French National Diagnostic and Care Protocols (NDCPs)

Generic Polyhandicap

PIMD (Profound intellectual and multiple disabilities)

Reference Centers

Neuropediatrics, Developmental Pathology, Trousseau APHP Hospital - Sorbonne University

Paediatric Polyhandicap of La Roche Guyon

Reference Centre for Polyhandicap and Intellectual Disabilities of Rare Causes

Reference Centre for Congenital Cerebral Palsy and Congenital Diseases of the Cerebellum

DéfiScience

National Health Network for rare neurodevelopmental disorders

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Text

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European Reference Network for Congenital Malformations and Rare Intellectual Disability

Summary

Polyhandicap

We have retained the linguistic terminology of the French word "**Polyhandicap"**.
This term has not become established in the international literature. Its equivalent, with some differences, can be found in the Anglo-Saxon literature under the term "Profound Intellectual and Multiple Disabilities" (PIMD) (Nakken 2007).This term defines the clinical features of the most highly disabled children, who require very specific human and material assistance. [Bourg, V. 2008*]*

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"Pioneer in the field of polyhandicap."
We would like to dedicate this National Protocol for the Diagnosis and Treatment of Generic Polyhandicap to Dr. Elisabeth Zucman, who passed away on Sept. 14th, 2019.

List of abbreviations

The acronyms are French and do not correspond exactly to English terminology, a literal translation has been made.

AAH	Allocation Adult Handicapped person
AEEH	Allocation for the Education of Disabled Children
AESH	Accompanying Pupils with Disabilities, former Auxiliary for School Life (AVS)
AGEFIPH	Association for the Management of the Fund for the Professional Insertion of Disabled People
ALD	Affection de Long Term
AMM/MA	Marketing Authorization
AMP	Medico-Psychological help
ANSM	National Agency for the Safety of Medicines and Health Products
AP-HP	Public Assistance - Paris Hospitals (Assistance publique – Hôpitaux de Paris)
AVS	School Life Auxiliary
CAFS	Specialised Family Reception Centre
CAJ	Day Activity Centres
CAMSP	Centre for Early Medical and Social Action
CAT	Help through Work Centre
CDAPHC	Commission on the Rights and Autonomy of Persons with Disabilities
CEFC	Training centre for health and social work
CEMC	Motricity Training Centre
CLIS	Classes of School Insertion
CMD	Multidisciplinary Consultation
CMPP	Medical-Psycho-Pedagogical Centre
CNCPH	National Consultative Council for People with Disabilities
CNSA	National Solidarity Fund for Autonomy (Caisse Nationale de Solidarité pour l'Autonomie)
DESS	Pain Child San Salvadour
DI	Intellectual Deficiency
EEAP	School for children and adolescents with multiple disabilities
EEP	Establishment for polyhandicapped children
EME	Medical-Educational Externat
ESMS	Social and Medico-Social Establishment
ESAT	Establishment and Help through Work Service (ex WCB)
FAM	Medical Nursing Home
FDV	Home life
FIPHFP	Fund for the Integration of Disabled People in the Public Service
GPE	Endoscopic percutaneous Gastrostomy
HAD	Hospitalisation at home
HAS	High Health Authority (Haute Autorité de Santé)
HDJ	Patient Day Hospital
IEM	Motricity training Institute
IMC	Cerebral Palsy, Brain Motor Infirmity (BMI)
IME	Medical-Educational Institute
IMOC	Motor Disorders of Cerebral Origin
IMP	Institut Médico-Pédagogique
IRM	Magnetic Resonance Imaging
ITEP	Therapeutic, Educational and Pedagogical Institute
LAP	List of Acts and Benefits
LPPR	List of Reimbursable Products and Services
MAS	Specialised Welcome House
MAT	Temporary shelter
MPR/PRM	Médecin physique et de réadaptation - Physical and Rehabilitation Medicine
MDPH	Departmental House for the Disabled (Maison Départementale des Personnes Handicapées)
NFS/CBC	numération formule sanguine - blood count blood count
PAIP	Individualised Welcome
PC	Project Cerebral
PCH	Disability Compensation Benefits
PLH	Polyhandicap
PIMD	**Profound intellectual and multiple disabilities (international definition)** (Cf Annex p116- Gérard Ponsot)
PNDS/NDCPs	**French National Diagnostic and Care Protocols**
PPH	Person with multiple disabilities (polyhandicap)
PPS	Personal Schooling Project
QI	Intellectual Quotient
RQTH	Recognition of the Quality of Disabled Workers
RTU	Temporary Use Recommendation
SAAD	Service of Help and Assistance at Home
SAMSAH	Medical and Social Service Service for Disabled Adults
SAVS	Social Life Support Service
SESSAD	Special Education and Home Care Services
SESSD	Education and Special Home Care Service
SPASAD	Multi-purpose home help and care service
SSAD	Service de Soins A Domicile
SSIAD	Service of Home Nursing Care
TED	Invasive Developmental Disorder
TSA	Autism Spectrum Disorders
ULIS	Localized Units for School Inclusion

Synthesis for the attending physician
Generic NDCPs Polyhandicap

1. Definition

The French decree of May 9, 2017 specifies those who are polyhandicapped : "Individuals presenting with early cerebral dysfunction or cerebral dysfunction that occurs during development, resulting in serious disturbances, with multiple and evolving manifestations, of motor, perceptive, and cognitive capacities and the construction of relationships with physical and human environment and a situation of extreme physical, psychological, and social vulnerability during which certain individuals may present, in a transitory or permanent manner, signs of autism".

This official recognition of Polyhandicap: a developmental disturbance that leads to a dramatic health condition with severe and complex disabilities, inability to walk, no meaningful oral language, severe and profound intellectual disability, constitutes the diagnosis of polyhandicap.

These disorders of the developing brain have evolving consequences throughout life: neurological, orthopaedic, digestive, respiratory, sensory, and other disorders. Behavioural disorders are common, as is pain. The perceptive, sensitive, and affective capacities of these people, their skills, and their appetite for communication must be considered to optimize their care and quality of life (*see appendix 4 in connection with the definition Chapter, Table 1: Polyhandicap, medical aspects*).

The causes are known in 70% of cases. They are mostly prenatal (70 to 80% of cases) then essentially genetic, perinatal (10 to 15% of cases), and postnatal (10 to 15% of cases). Etiological research is essential, favouring early care (prenatal or from birth) and sometimes genetic counselling.

2. Clinical signs and treatment

Rather than summarizing a too huge body of work, we have chosen to focus on the most important points to pay attention to in the daily life of the polyhandicapped child or adult with multiple disabilities, the support of whom it is essential to consistently obtain by talking to them.

2.1. Pain

The first point of particular concern is pain. Polyhandicapped children, as well as adults, very frequently suffer from pain of various origins. It can be nociceptive, neuropathic, or psychological, with polymorphic aspects. It is therefore always necessary to anticipate it. Each person must have a record of their basic pain, and each change in behaviour must lead to a new evaluation, ideally with the help of an evaluation grid.

It is essential to find the cause of the pain to propose the most suitable treatment possible. Otherwise a trial treatment is appropriate. The possibility of psychological suffering should not be overlooked.

2.2. Nutritional status

A second point of concern is the monitoring of nutritional status. Pain, like many potential dietary or digestive difficulties, can lead to food refusal by those who are already often in a precarious nutritional state.

Faced with the risk of undernutrition, a nutrition assistance strategy must be proposed. If needed, a gastrostomy can compensate for inadequate oral intake by allowing enteral feeding.

Undernutrition aggravates all other pathologies, promotes infections (respiratory or other), muscle wasting, osteoporosis, and trophic disorders (bedsores), and can be life-threatening.

Food or saliva inhalation linked to swallowing disorders are a cause of undernutrition, as well as recurrent respiratory infections. They are almost constant in the polyhandicapped but can go unnoticed because of the absence of a cough reflex.

2.3. Epilepsy

Epilepsy is one of the most common comorbidities of the polyhandicapped (50 to 65%, depending on the study). It can manifest itself as any clinical type of seizure, motor seizures being the most noticeable. They consist of severe lesional epilepsies, often pharmaco-resistant (25 to 31% of cases), with frequent states of malaise. If the epilepsy worsens, one must first search for the source of irritation: pain, regardless of its origin (often digestive, due to constipation, orthopaedic), pulmonary or urinary infection, change of life situation, lack of sleep, etc.

Substantive treatment should be given by a neurologist. Treatment of the seizure may involve protocols such as rectal or oral benzodiazepines or other specific protocols prescribed by the neurologist who is following the person (*see Appendix 4, in relation to Chapter 2.4, Epilepsy*).

2.4. Motor impairment and tone disorders

The polyhandicapped present disorders of motor control with disturbances of muscular tone (hyper or hypotonia) and possible autonomic movements. Of course, the orthopaedic and postural consequences are significant during growth but also throughout life. This aspect must be followed closely from very early childhood. Rehabilitation aims to improve motor development, stimulating and developing voluntary motricity and making it possible to settle comfortably into a lying or sitting position. In adults, the maintenance of good posture is essential, relying on well-adapted equipment and the continuation of rehabilitation, while encouraging daily periods of free movement.

2.5. Neuro-Orthopaedic disorders

These disorders may be related to the neurological pathology itself, associated with tonic-motor imbalances and growth. They require physical and rehabilitative medicine (PRM), rehabilitation, and orthopaedic intervention, particularly surgical. The main disorders are those of the spine (severe cypho-scoliosis), which can compromise respiratory function, the hips, which may affect sitting and lying down, and the feet.

Children must be treated as early as possible. This involves rehabilitation, orthopaedic surgery, if necessary, and orthopaedic devices. The objectives are to direct growth to allow pain-free sitting and lying down and to educate the caregivers.

In adults, the objectives are to improve overall comfort, facilitate transfers and nursing care, aid personal hygiene, optimize the conditions for social interaction, and minimize respiratory, digestive, and skin problems.

Bone fragility must be monitored on a daily basis, as it can cause spontaneous fractures and pain. Preventive and corrective measures must be undertaken, and treatment provided, if necessary, from childhood onwards.

2.6. Respiratory problems

Respiratory problems are the leading cause of death among the polyhandicapped. They are often the consequence of inhaling food/liquids. They may also be partially due to position, but susceptibility to viral or bacterial infections due to immune deficiency can also be responsible. The bronchopulmonary state of the individual is also conditioned by neurological disorders (respiratory control, inability to cough and spit) and deformations of the thoracic cage linked to spinal deformities, which are sometimes very significant. Any infection must therefore be treated quickly and effectively (adapted antibiotics, physiotherapy and bronchial drainage, and bronchodilators, if necessary). Non-invasive ventilation methods at night or during the day may also be considered.

2.7. Digestive disorders

Gastroesophageal reflux is very common and often serious because it is frequently diagnosed at the complication stage. It causes pain and aggravates oral disorders and undernutrition but is often amenable to improvement through long-term medical or surgical treatment.

Constipation is a major problem and must be systematically and rigorously prevented, foremost by sufficient water intake, a diet sufficiently rich in fiber, regular mobilisation and verticalization, abdominal massage, prevention of any drug-induced iatrogeny, and, if possible, the encouragement of defecation in a sitting position, particularly postprandial.

2.8. Numerous and frequently associated disorders

The prevention and treatment of dental appliance abnormalities are essential for lifelong quality nutrition. Consideration of vesico-sphincter disorders, basic vegetative disorders, and sleep disorders are essential for the daily life of the polyhandicapped individual. Hearing and visual disorders must be systematically sought; communication with them is essential as he or she has so much to "say"!

Any new clinical situation should be diagnosed and followed up in both children and adults. All components are intertwined throughout the course of their lives. The clinical condition of polyhandicapped individuals is fragile and progressive and can quickly become an emergency, requiring anticipation, networking, and systematic periodic specialised follow-up throughout life.

Behavioural and personality disorders may be part of the causal pathology and/or appear at various occasions during the person's life (adolescence, bereavement, pain, psychological suffering).

Any chronic disease can also affect the polyhandicapped and must therefore be subject to prevention and/or screening in the same way as for anyone else.

3. Care and monitoring

3.1. Daily life of the polyhandicapped

A polyhandicapped person is highly dependent on his or her family and friends, who are generally familiar with the polyhandicap and the particular characteristics of their loved one. They must be involved in all actions concerning him or her.

All activities of daily life of the polyhandicapped (washing, dressing, learning, emotional and sexual life...) must be carried out with attention for his/her health. Each polyhandicapped person must have personal information sheets (in any form: tables, lists, etc.) indicating their needs, habits, tastes, and personal vulnerabilities.

All staff accompanying the polyhandicapped individual is responsible for systematically reporting any changes in his or her clinical condition to his or her nurse or referring doctor.

3.2. Medical follow-up of the polyhandicapped

Various examinations are required at regular intervals. They involve all of the body's systems and aim to maintain the health of each polyhandicapped individual (*see table in Chapter 3.2.2, Medical monitoring model: Polyhandicap care pathway*).

Multidisciplinary consultations are recommended, using modern technologies, such as telemedicine, when necessary. Hospital check-ups should favour scheduled appointments. In emergencies, specific reception of the patient should be organised, with a "remarkable patient" file.

Life expectancy for polyhandicapped people has increased in recent years but is still below average. Many causes can lead to a serious and unstable clinical situation, requiring palliative care for both children and adults under the Claeys Leonetti law of 2016.

4. Role of the General Practitioner

4.1. Follow-up of medico-administrative measures

- Having a polyhandicap implies a disability rate of 80% from the time of diagnosis and consequently the granting of a disability card and an allowance (with possible supplements for children), to be requested from the Maison Départementale des Personnes Handicapées (MDPH), as well as a disability compensation benefit, especially for adults, as soon as the diagnosis of polyhandicap is made, to make life easier for the person's family and friends. Whenever necessary, the GP ensures that medical certificates are provided to accompany applications for the MDPH.
- The exemption of the co-payment for health expenses is requested from the Social Security Fund on which the person depends.

4.2. Medical follow-up

- Refer the child to a paediatric neurology service to confirm the diagnosis and set up the necessary ongoing follow-up.
- Support and accompany the family, especially when the person lives at home.
- Ensure that patient follow-up is carried out by a multidisciplinary team with knowledge of the specificities of polyhandicap, according to the recommendations of the NDCPs.
- Ensure the monitoring of progressive disorders, in coordination with the referring teams.
- Ensure the follow-up generally recommended for all patients (children or adults): vaccinations, specialized clinical and biological monitoring according to the NDCPs (monitoring table), systematic prevention and screening of all adult patients (osteopenia, cancer, etc.).

5. Further information

(confers NDCPs Text)

Contact details (see Annex 2)

- Accredited Reference and Competence Centres (see Appendix 2)
- DéfiScience National health network for rare neurodevelopmental disorders http://www.defiscience.fr
- General information - Internet source - http://www.orpha.net
- Patient associations and polyhandicap health networks (see Appendix 2)

Reference Texts

(confers NDCPs Argumentation)

There are countless references covering all subjects related to polyhandicap and a few reference texts that cover the entire subject:

- Decree No. 2017-982 of May 9, 2017 on the nomenclature of social and medico-social establishments and services for the disabled and chronically ill
- Camberlein P. and Ponsot G., The Polyhandicapped person, know him, accompany him, take care of him. Collection guide sante sociale DUNOD November 2017 (new edition to be published in 2020)
- Juzeau D., Vivre et grandir polyhandicapé, Dunod, 2010
- HAS recommendation, to be published in 2020, on the support of polyhandicapped people, which was collaboratively conceived and written, in particular the health section.

NDCPs Text

French National Diagnostic and Care Protocols (NDCPs)

Definition of Polyhandicap

Validated in June 2018 by the French Society of Paediatric Neurology

The official French definition of polyhandicap has been updated by decree N°2017-982 of May 9, 2017[(1)] on the nomenclature of social and medico-social establishments and services accompanying disabled or chronically ill individuals.

This text was incorporated as soon as it was published in the Code of Social Action and Families and describes the population concerned:

"Individuals with an early or developmental brain dysfunction resulting in severe, multiple, and progressive disturbances in motor, perceptual, and cognitive capacities and the construction of relationships with the physical and human environment, and a situation of extreme physical, mental, and social vulnerability in which some individuals may present, either temporarily or permanently, signs of autism".

This official recognition of severe and multiple disabilities (lack of independent walking, no meaningful oral language, profound intellectual disability) should make it possible to provide more appropriate responses to the needs of the people thus identified.

It highlights the situation of vulnerability caused by severe and early brain injury, usually before the age of two years. Since 1989, the date of the first "official" definition, other definitions have circulated, which are intended to be more complete or more positive or to include etiological elements.

The precise aetiology of a polyhandicap is only known in 70 to 80% of cases: when it is known, the cause is prenatal (65 to 80% of cases), essentially genetic, including progressive neurological diseases, perinatal (10 to 15% of cases), mostly linked to extreme prematurity, or post-natal (10 to 15% of cases).

These brain disorders always have multiple consequences, which remain progressive throughout life: multiple neurological (somatic and intellectual), orthopaedic, digestive, respiratory, sensory, bone, and other disorders *(See Appendix 4 in connection with the definition Chapter, Table 1: Polyhandicap, medical aspects)*. Behavioural disorders are frequent (30 to 40%). Pain is also a part of the daily life of these individuals. The perceptive, sensitive, and affective capacities of these people, their skills, and their appetite for communication must be considered to optimize the care they receive and their quality of life.

Why a generic PNDS Polyhandicap (NDCPs)?

The prevalence of polyhandicap is 0.50 per 1,000, so it is not uncommon. However, the etiologies of polyhandicap are by and large rare diseases, diagnosed or not. Multiple brain lesions and their consequences on growth and secondary repercussions on various organs are sufficiently characteristic for this situation of severe disability, from birth or early childhood, to be declared as a true pathological entity in a national summary document.

In accordance with the missions entrusted to it, the DéfiScience sector has undertaken to support and coordinate, through methodological support, the drafting of the National Diagnosis and Care Protocols for its Reference Centres. The implementation of the NDCPs dedicated to polyhandicap has been entrusted to the Rare Cause ID Reference Centres within the framework of the **DéfiScience** French National Network -

1 Introduction

The preparation of this NDCPs was particular because for the "Polyhandicap theme" it was essential to go beyond the framework of rare syndromes that contribute to a polyhandicap, but rather to make it the basis from which to describe the reference management of a patient with a polyhandicap (of rare or non-rare cause).

All stages of life are covered in this NDCPs, as well as all aspects of the daily environment, from hospital to home, medical to social, and short- to long-term stays in specialised facilities.

This NDCPs is intended to be a guide for health professionals, who in their practice may sometimes lack knowledge or advice on good practices for supporting polyhandicapped patients.

1.1 The objective of this National Diagnostic and Care Protocol (NDCPs)

The aim of this NDCPs is to explain the current optimal diagnostic and therapeutic management and healthcare path of a polyhandicapped patient to the professionals concerned so as to optimize and harmonize the support and monitoring of polyhandicapped people throughout the country.

However, the NSDPs cannot consider all specific cases, co-morbidities or complications, therapeutic particularities, hospital care protocols, etc. Nor can it replace the individual responsibility of the physician towards his or her patient.

The present NDCPs has been prepared according to the "Method for the preparation of a national protocol of diagnosis and care for rare diseases" published by the High Authority of Health in 2012 (methodological guide available on the HAS website: (www.has-sante.fr). A PNDS (NDCPs) is based "on the search for scientific evidence and the consultation of all concerned actors", taking into consideration the French or international recommendations or protocols already published on a pathological entity.

1.1.1 Identifying the polyhandicap

It is fundamental to first recognize the polyhandicap, so as to provide care and accompaniment of the person according to the proposed guides, taking into account his or her character, aspirations, and needs, like any other human being, regardless of the origin and particular characteristics of the polyhandicap and its severity.

The French term "polyhandicap" has not established itself in the international literature. Its equivalent, with some differences, can be found in the Anglo-Saxon literature under the term "Profound Intellectual and Multiple Disabilities" (PIMD) (Nakken 2007). The place of the various designations is detailed in Appendix 4 *(Appendix 4, in connection with the chapter on definition, Table 2, place of polyhandicap/PIMD in disabilities with motor impairment).*

Several studies on the prevalence of polyhandicap have been carried out over the last 20 years and provide figures between 0.50 and 0.73/1,000.

The prevalence of polyhandicap was estimated to be 0.50 per 1,000 for the generations from 1980 to 2009 (strict or extended polyhandicap in children over 7 years of age) from the RHEOP database (Registre des Handicaps de l'Enfant et Observatoire Péri natal). It was estimated to be 0.73/1,000 children by Rumeau Rouquette in 1998 and 0.70/1,000 children in 1999 by Juzeau. The DREES survey, ES Handicap de 2014, estimated there to be 9,400 polyhandicapped children and 23,200 polyhandicapped adults in France, i.e. 0.50/1,000 all ages combined. The frequency of deaths in children may explain the drop in prevalence in the general population. However, if children survive infancy, they increasingly reach adulthood, with an increasing life expectancy, explaining the growing demand for places in adult institutions.

1.1.2 Provide appropriate care

Polyhancap is the consequence of a fixed or early progressive multifocal brain injury, i.e. during the period of brain formation and development (anterior, peri, and postnatal periods up to the age of 2 years, by analogy

with cerebral palsy), even if brain maturation continues for many more years. Any lesion occurring during this period implies the disorganization of various processes, as well as the possibility to compensate the initial lesion by the activation of other not-yet specialized neuronal circuits; this is the purpose of early rehabilitation.

The resulting impairments do not simply add up, but become intertwined and potentiated, leading to multiple cascading problems. The appropriate care of these people must therefore be global, interdisciplinary, coordinated, coherent, and carried out in close collaboration with the family. Given their complete vulnerability, ethical implications must always be taken into consideration.

Physiological ageing, which can occur early, can alter acquired capacities; care must also be taken to detect and treat additional diseases (cancer, diabetes, kidney failure, etc.), as for anyone else.

1.1.3 Accompany the person and their loved ones

The aim of accompaniment is the optimal achievement of comfort, education, quality of life, well-being, and pleasure of life, starting with the absence of physical or psychological pain (importance of the evaluation of such pain on a daily basis).

The links between the health sector and the medico-social sector are described in the relevant chapters; they are essential for the implementation of a true-life project for each individual. The NDCPs provides guidelines on the need for care and the opportunities for accompaniment, to be adapted according to local realities. The objective of this NDCPs is to be a guide of quality practices to enable "agreement" between parents and professionals.

1.1.4 Issues not addressed

The following situations are not covered:

- Mild to moderate intellectual disabilities
- All brain injuries acquired after two years of age
- Dementia, except that beginning in early childhood.

1.2 Professionals involved

General practitioners, paediatricians, and other health professionals in the field are alerted by various signs of psychomotor retardation. Their role is to make the family aware of the need to consult specialists (Billette de Villemeur, 2012).

The first specialists to be consulted are, first and foremost, neuro-paediatricians, a "pivot specialty" in the case of diagnosis and follow-up of a child's polyhandicap, who will then refer him or her to the various complementary specialties required in each case, depending on the symptoms, and the prevention to be carried out. All specialties may be involved, depending on the clinical aspects previously described (Annals of Physical and Rehabilitation Medicine, 2013).

In the follow-up of polyhandicap in adults, the general practitioner coordinates the various types of care and follow-up that need to be regularly carried out. He follows the patient at home or in an institution. *(Cf: Appendix 3, Figure 1 - Participation of diagnostic professionals in the life project, according to profession and sector)*

The reference centres to be involved depend on the diagnosed pathologies. It is essential that each family be informed of the possible existence of these centres so that the follow-up of the child/adult benefits from the best possible quality and a pejorative evolution is avoided or at least delayed, if unavoidable.

Health and medico-social institutions are involved at all levels of the care path and the links between the health and medico-social sectors must remain strong to provide each individual with the care and support that suit him or her (Heron-Longé B. , Ponsot G., 2017).

Networks and associations are used as needed so that each family can find answers to their questions and raise their child within a social network that breaks the isolation that often follows the arrival of a polyhandicapped child in a family (Camberlein, 2017).

1.2.1 The role of polyhandicap associations and collectives

The experience of the families should also be recognized when discussing the care path, because they know the most about all the hazards and vicissitudes. Indeed, families are afforded an important role in the hearings of the High Health Authority (HAS). The experiences of the families are strewn with small victories and big disappointments or announcements, and they often know instinctively and by experience where and by whom their children will be best cared for and in whom they can place their trust (HAS, 2008).

This is why the relevant parent and/or professional associations were associated with the drafting of this NDCPs and the professionals of several collectives working on polyhandicap were invited to participate in its preparation, both in the writing and proofreading of the text.

This was already the belief of Stanislas Tomkiewicz, who participated with Elisabeth Zucman, with an unfailingly militant attitude to advocate for a true policy in favour of people with a polyhandicap:
"When you get to know them well, when you accept to open your brain and your heart, you discover human beings with such a fine affectivity that goes hand in hand with an often surprising intuition, and their potential for evolution, even if it is very limited, often exceeds the prognosis, which is often still too pessimistic."».

1.2.2 A team of professionals and support staff

This team consists of:

- Educational professionals, at all levels, who accompany children and adults in their daily and social life
- Professionals in chronic and acute care, for both short-lived acute pathologies and chronic monitoring of polyhandicap. All types of medical personnel may be concerned
- Professionals from the pedagogical domain, who accompany the polyhandiapped individual in their learning and permanent development of their cognitive capacities.

Polyhandicap must be treated as a public health problem that affects society as a whole, as it involves many professional actors, as well as various families and caregivers, and the whole of society, which participates in their inclusion in its activities.

1.3 Aetiologies and the diagnosis of polyhandicap

The few surveys published on the subject in the 1970s revealed a percentage of unknown causes on the order of 40% of cases. When an aetiology was found, it was specified as antenatal in 50% of cases, perinatal in 34%, and postnatal in 16%. However, it should be noted that these figures most often concerned only periods of brain damage rather than a precise diagnosis.

1.3.1 Why make the effort to establish a precise diagnosis?

For the parents and the child, knowledge of the etiological diagnosis serves several purposes. For the child, it can allow him or her to adapt to their medical, rehabilitative, and socio-educational care and follow-up. In addition, certain pathologies, particularly metabolic, can benefit from treatment. In other cases, prevention of over-disability can be effective. For the parents and the rest of the family, if necessary (X-linked disorders for example), the diagnosis opens the way to genetic counselling for subsequent pregnancies, with the possibility of prenatal or pre-implantation diagnosis. Moreover, after an often long and agonising path of etiological research - rightly described as veritable "diagnostic wandering" - many parents are relieved to be able to put a name to the pathology their child suffers from, meet other similar families through patient

associations or social networks, and put an end to a series of potentially invasive tests. Often, their testimony emphasizes the guilt-reducing nature of the genetic diagnosis.

For the care and education teams, a precise diagnosis makes it possible to understand and accept the care and support the approach, which differs according to the fixed, acquired, or progressive nature of the disease (Ponsot 2017).

Finally, for doctors, understanding the causes of the polyhandicap allows them to progress in their knowledge of them and therefore in their prevention and to be aware of and prevent possible progressive complications. It also makes it possible to improve our epidemiological knowledge (Rodriguez, 2017).

1.3.2 Results of etiological studies

Several aetiological studies have been detailed and compared and can be seen in Appendix 4 (*cf. Appendix 4, ch 1.3, Table 1: Comparative study of surveys carried out from 1992 to 2018*).

In particular, we cite a survey carried out on 318 documented cases (Monocentric study, PERNES, 2018) using a strict definition of polyhandicap and a precise diagnosis (the etiological diagnosis was considered to be a "precise" diagnosis when the physiopathology was clearly identified, the terms ante, peri, or post-natal encephalopathies or even epileptic encephalopathies being considered as a nosographic classification). It showed the following figures (birth years from 1974 to 2015): precise diagnoses were found for 73.8% of cases, antenatal causes, representing 67.5% of known aetiologies, dominated by genetic causes (31.6%), followed by cerebral malformations (18.8%) and acquired causes (14.1%); perinatal causes, representing 15%, dominated by anoxo-ischaemia (7 of 10 cases in premature babies), far ahead of infectious (3%) or metabolic causes (0.8%); and postnatal causes, remaining fairly high at 17.5%, mainly represented in this study by severe head trauma, mostly related to maltreatment *(cf. Appendix 4, ch 1.3, Table 2: Study of aetiologies over 3 successive periods)*.

In the Eval-PLH study (Rousseau 2019), carried out using questionnaires completed by investigators and covering 463 files of children and young adults (3 to 25 years old), the aetiologies remained unknown in 63 cases, i.e. 13.6%. Among the 400 cases with a known diagnosis (86.4%), the following breakdown is given (figures compared to 400 cases):

- Prenatal causes: 237 (59.2%), dominated by cerebral malformations (45.7%), of which neuro-metabolic encephalopathies (6.25%), neurogenetic encephalopathies (4.75%), and epileptic encephalopathies (2.5%) represented a total of 13.5%
- Perinatal causes: remaining high (129 or 32.25%), dominated by anoxia (15.25%), prematurity (8.5%), and infectious causes (7%)
- Postnatal causes: 34 cases or 8.5%, represented mainly by traumatic causes (5%).

In the Eval-PLH study, identification of the perinatal causes was imprecise, and they were probably over-represented due to the type of data collection, the aetiology not being the primary objective of this survey.

There is still progress to be made in the search for the causes of polyhandicap. However, significant progress has been made over the years: the percentage of specified aetiologies is increasing: from 59.2% for children born before 1992 (Boutin) to rates varying, according to subsequent studies, from 73.8 to 90.7%. Such an increase in the rate of diagnosis is particularly due to improved genetic techniques, with the CGH-array, followed by next-generation sequencing (targeted panels or whole genome sequencing), which should be proposed when encountering an antenatal cause with no identified aetiology.

The frequency of perinatal aetiologies has fallen sharply: from 34.4% in 1992 (Boutin) to 15% (Pernes 2018). The increase in antenatal causes (from 47.3 to 72.8%) is mainly linked to the growing identification of genetic causes. Postnatal aetiologies (up to 2 years) are in sharp decline, but remain dominated by head injuries, mostly in the context of abuse (shaken babies, in particular).

Focusing specifically on the years of birth from 2002 to 2015 (Pernes 2018), antenatal causes amounted to 81.7% versus 11.7% for the perinatal and 6.7% for the postnatal periods *(cf Annex 4, 1.3, Table 2: Study of aetiologies over 3 successive periods)*.

1.3.3 Diagnostic strategy

The etiological exploration in a patient with a polyhandicap is most often carried out in several stages. These steps are summarised in Appendix 3 *(see Appendix 3, Figure 2: Proposed diagnostic strategy for a patient with a polyhandicap).*

- **Anamnesis**

The first step consists of taking the history, which includes a family investigation and a history of the child from conception, followed by clinical assessment. This stage is crucial, making it possible on the one hand to potentially direct complementary examinations and, on the other, to explain the issues involved in genetic examinations to the parents, in particular high-throughput sequencing. Complementary morphological examinations (cerebral MRI, cardiac and renal ultrasounds, ophthalmological examination) are also useful, and can sometimes guide the search for an aetiology.

A three-generation family tree is constructed, and the family history recorded, with an emphasis on the notion of individuals with an intellectual deficiency, disability, epilepsy, or malformation(s). Possible parental consanguinity must also be sought. The data collected by the family tree may point to a genetic origin and a mode of transmission: a history in the maternal branch of the family may suggest an X-linked disorder; a history in the siblings may suggest a recessive pathology; repeated miscarriages and cases of infertility may suggest a family translocation being transmitted in a balanced or unbalanced state.

The history should include the perinatal history: progress of the pregnancy, the results of ultrasounds and any screening tests, the intake of toxic substances or medication, gestational diabetes, or maternal infection, suggesting a non-genetic cause or confounding factor, birth term and measurements, and Apgar score. The neonatal history is also taken - possible notion of feeding difficulties, and anomalies on neurological examination (hypotonia, bad eye contact, convulsions), or hospitalisation. Complementary examinations are carried out: imaging check-up to search fora malformation and auditory and ophthalmological examinations. Finally, the stages of psychomotor development and the paediatric history are of course detailed.

- **Somatic examination**

The somatic examination for genetic diagnosis must be complete. The measurements and growth curves (weight, height, and head circumference) of the child are noted, as well as the measurements of the parents in case of abnormalities (especially concerning the head circumference). The morphological examination focuses on the search for even minor peculiarities likely to point to an aetiology: facial morphology (including mouth, palate, uvula, and teeth), orthopaedic examination (spine, limbs, pectus anomaly, laxity), skin examination (pigmentation anomalies, vascular malformations), extremities, and external genitalia.

Far from making clinical examination obsolete and useless, high-throughput sequencing tests, which have revolutionized medical genetics in recent years, have, on the contrary, led to an awareness of the importance of fine patient phenotyping, more essential than ever for the relevant analysis of molecular results. Ideally, the initial clinical examination should make it possible to define a diagnostic framework that will guide the biologist in interpreting the results. In its absence, the variants identified may or may not be retained based on complementary clinical data obtained secondarily when the patient is reviewed with results of uncertain significance ("reverse-phenotyping"). Clinical-biological collaboration and a multidisciplinary approach to the patient are therefore a cornerstone of tomorrow's genetics.

- **Prescription of genetic tests**

Prescriptions for a genetic test are usually provided by the paediatrician, pediatric neurologist, or clinical geneticist.

If the clinician recognizes a genetic syndrome with a good level of conviction, a targeted examination may be requested at the outset. This may be FISH, if there is a suspicion of a microdeletion syndrome (deletion 1p36,

etc.), sequencing of a targeted gene (TCF4 gene for Pitt Hopkins syndrome, MECP2 for Rett syndrome, etc.), or sequencing of a limited number of genes by means of a panel, such as the "epileptic encephalopathies" panel.

If the test does not suggest an aetiology, or if the evoked syndrome is genetically heterogeneous, the proposed first-line test is chromosomal microarray analysis (CMA). CMA shows good diagnostic performance, up to 20% in intellectual disability, with or without a polyhandicap, depending on the series, and its genome-wide nature makes it a good first-line test. CMA is also performed if a targeted strategy is negative.

As a second step, genome-wide molecular tests are recommended. Within the framework of the France Genomic Medicine 2025 plan, the deployment of two very high-throughput sequencing platforms in France, planned for 2020, should make it possible to carry out whole-genome sequencing for polyhandicapped patients, with modalities that are still under discussion. In the meantime, exome sequencing or a panel of genes involved in intellectual disability will continue to be carried out.

- **Modalities of prescription**

CMA can be prescribed by a geneticist physician or a non-geneticist physician if he or she is competent to provide information to the patient on its principle and issues, as summarized above. In addition, before being carried out, informed consent of the patient must be obtained or that of his or her legal representatives.

Within the framework of the etiological exploration of a polyhandicap, it is preferable to refer the patient to a genetics service if these tests come back normal or as a first intention according to local practices. If this is not possible, the patient can be referred to a professional working in close collaboration with a rare disease reference centre (CRMR) specialised in intellectual disability and polyhandicap. Second-line tests (gene panels, exome, or genome sequencing) can be discussed and carried out if indicated.

1.4 Genetic counselling and prenatal diagnosis

The question of genetic counselling may arise when a relative of the polyhandicapped child (parents, a parent, aunt or uncle, an adult sibling) questions the risks of recurrence of the polyhandicap for his or her descendants. As polyhandicap does not have a single cause, but a wide range of aetiologies, genetic information aims to verify whether the precise aetiology of the polyhandicap has been demonstrated to inform (future) parents whether or not there is a risk of recurrence. Indeed, a certain etiological diagnosis is the *sine qua non* for reliable genetic counselling, hence the importance of implementing the most appropriate techniques in cooperation with clinical geneticists when an acquired cause has been eliminated: infection (infectious fetopathy such as CMV, congenital rubella, congenital toxoplasmosis, etc.), trauma, or asphyxiation (shaken child or drowning).

When the diagnosis of the disease responsible for the polyhandicap is demonstrated by the discovery of a definitive genetic abnormality, genetic counselling makes it possible to inform relatives of the risk of transmission of the abnormality. This risk depends on the mode of transmission (chromosomal aberration, dominant or X-linked mutation, autosomal recessive mutation, or mitochondrial disease), as well as on the position of the polyhandicapped child in the family tree in relation to the consulting relative (parents, uncle or aunt, cousin, brother, or sister). The risk of recurrence for the pregnancy of a relative is assessed individually. Genetic counselling is carried out by a geneticist (or genetic counsellor) in conjunction with the referring doctor.

Genetic counselling is therefore intended to inform parents and relatives about the risk of recurrence and the possibilities of prenatal diagnosis or preimplantation diagnosis (via *in vitro* fertilisation). When a genetic diagnosis is not determined (diseases for which the mode of transmission is not known, or those for which the genetic mechanism is not known and cannot be ruled out in the light of current knowledge), genetic counselling cannot provide a precise answer on the risk of recurrence.

1.5 Early detection

Early detection of a polyhandicap is necessary to enable the rapid management of disorders to limit the deleterious effects of the interaction between impaired neurological function and physiological development in childhood: brain maturation and skeletal growth. The detection of serious abnormalities at birth or during pregnancy makes it possible to establish specialised follow-up and to adapt care to the child's progress from birth.

Indeed, when the possibility of polyhandicap is detected during pregnancy, parents who wish to continue the pregnancy say that it allows them to better welcome their child.

Sometimes, screening during pregnancy can also lead parents and doctors to consider terminating the pregnancy. Parents are entitled to request medical termination of the pregnancy after the discovery of an abnormality of such severity that it is deemed to be incurable and likely to progress to a polyhandicap. Such a request occurs, in particular, when the parents already have a first polyhandicapped child: during a subsequent pregnancy, these parents may request medical termination of the pregnancy if brain lesions identical to those of their polyhandicapped child are found.

If the diagnosis of brain injury is made in the context of neonatal resuscitation, continued resuscitation may be considered unreasonable if the neurological prognosis is very severe. After a meeting of the medical team, the cessation of treatment that only results in keeping the child alive, with no hope of significant progress, may be decided after discussion with the parents.

In these situations, screening has thus two opposing but not contradictory purposes: to enable parents to prepare themselves in the event that they will have to care for their polyhandicapped child (by choice in prenatal or post-natal care) or to enable parents to request medical termination of the pregnancy or doctors to interrupt resuscitation if keeping the child alive seems unreasonable. In all cases, the decision is painful for the parents, as well as the doctors and caregivers. A consensus after discussion between parents and professionals should always be sought.

2 Clinical signs and treatment

A polyhandicap represents the consequences of an original brain injury on multiple organs and functions and not, as its name might suggest, the sum of several associated disabilities.

The evolution of the observed disorders is an element of particular importance. Although it is due, in its own right, to the evolution of progressive encephalopathies, it is also an element of fixed encephalopathies and their orthopaedic, respiratory, digestive, etc. consequences, belonging to what is known as a secondary disability *(See appendix 4 in connection with the definition Chapter, Table 1: polyhandicap, medical aspects)*.

The medical status of the polyhandicapped individual is therefore never fixed once and for all, and must be constantly reassessed and reconsidered. The prognosis is always uncertain, which is a continuous hardship for the patient and his or her entourage.

The increasing and necessary medicalisation of the therapeutic management of pathologies of polyhandicapped individuals has significantly improved and extended their lives. This has increasingly given rise to the question of the limits of such medicalisation. Caregivers find themselves confronted with new medical situations that are sometimes difficult to manage, both in terms of the strict therapeutic possibilities and the resulting ethical problems.

2.1 Assessment of cognitive impairment and skills

The need to provide polyhandicapped people with support and education from which they can fully benefit requires an assessment of their cognitive skills. This raises questions, particularly because of the complex interplay between significant motor, cognitive, and sensory impairments (Morris, Kurinczuk, Fitzpatrick & Rosenbaum, 2006). These impairments often render tests used in the practices of psychologists and professionals irrelevant. In particular, such tests require sensory and/or motor skills that these individuals do not have.

Evaluation is essential because family and professionals need to have agreed upon benchmarks to guide their interventions, make choices concerning education, activities, and pastimes, and define priorities for the interventions to be proposed (Nakken and Vlaskamp, 2007).

2.1.1 The ECP scale ("Evaluation - Cognition – Polyhandicap")

The ECP is an easy-to-use scale for professionals and families. Through the combined assessment of the evaluators, it provides insights into the cognitive skills and mental suffering of polyhandicapped individuals of all ages

The ECP was psychometrically validated between 2015 and 2017.

The specific features of the disorders require the psychologist to analyse the individual's behaviour in relation to certain tasks with great finesse. These characteristics must be considered in the assessment process.

Moreover, the evaluation shows a particular time-sensitive nature in these situations:

- Skills are sometimes revealed "by surprise" at an occasion that provides the polyhandicapped person the opportunity to show that they understand or remember something
- The reaction time of the polyhandicapped person is often very long: the more worried they are, the more they concentrate, and the longer they require to respond.

This leads us to propose consideration of the temporal aspect from two angles: the timing of the evaluation itself and its periodicity.

It is not simply a question of identifying skills and using them to initiate learning, but also of ensuring that such learning is meaningful and functional for the individual and those close to him or her (Scelles, 2013).

The cognitive assessment of these individuals requires consideration of their body, what it reveals, its physical and sensorial capacities, and how it is organized.

For a variety of reasons, polyhandicapped individuals live or have lived for long periods of time with a body that makes them suffer. Such pain, which is not necessarily noticeable or easily alleviated, has an impact on their cognitive life. Unrecognized and misunderstood, such pain can lead to withdrawal from the world or the refusal of affective relationships.

Goffman (1975) speaks of a "body dialect" that originates during early bonding to evoke the way people without verbal language express themselves. Indeed, people with a polyhandicap develop a type of body language with their loved ones over time. This is manifested both through voice (echolalia, vocalizations, onomatopoeia) and non-verbal signs (mimics, mimes, avoidance behaviours) (Lormet 2000).

In light of all of these considerations, creating a cognition assessment tool that is adapted to this population requires consideration of the non-verbal communication, extreme heterogeneity, and even evolutionary nature of these individuals, as well as their human and physical environment.

Rigorous, attentive, methodical observation is an essential tool, so that from smiles, eye movements, body movements, limb tension, etc. it is possible to grasp what the person understands and through what type of stimulation he or she is best able to learn and enjoy doing things.

Moreover, certain gestures and mimicry may not be voluntary but appear in the context of tension in the individual who wants to respond to a solicitation, or dystonia or dyskinesia.

Establishing a profile of the capacities of individuals within this population and seeing how the profile evolves over time helps in building their educational program. The differences in profiles constructed by parents, psychologists, and speech therapists, for example, are valuable sources of learning in this assessment process. An illustration of the profile constructed with the ECP is presented in the rationale (Chapter 2.1).

2.1.2 Use of the ECP

The ECP can be offered to children, adolescents, and adults as part of their longitudinal follow-up at all stages of their care. The obtention and analysis of the results imply two types of subjectivity: that inherent to direct and/or deferred observation and that related to the interpretation, which is made from what was observed (memories, as well as interpretation of facial expressions, body language, etc.).
The ECP can be used in two ways:

- For the regular assessment of polyhandicapped individuals (for example, to monitor their progress or identify the effects of a treatment or intervention); in this case the profile is constructed and appears with the report written by the psychologist in the file
- To discuss the skills of the polyhandicapped individual and his affective state with relatives, between relatives and professionals, or between professionals; it is thus a type of common observation grid that allows the exchange of information between people concerned about the psychic health and cognitive development of the polyhandicapped individual.

2.2 Motor deficiencies and tone disorders

The polyhandicapped individual does not have proper motor control of his or her trunk and limbs because of motor-control disorders on the one hand and disturbances in muscle tone (whether excessive or diminished) on the other.

Such disturbances lead to postural disorders (head posture, sitting posture) and strict motor disorders: paralysis, disturbances of voluntary movement, involuntary movements.

2.2.1 Clinical aspects

- **Examination at rest**

Examination at rest allows the assessment of basal contractions and abnormal postures. This examination is part of the overall observation of the polyhandicapped individual (Camberlein P., Ponsot G., 2017). The postural function of the trunk is described and its deficit is qualified, for example, by the SATCo (Segmental assessment of trunk control, Butler P., 2010), which is a scale that can be used for a child with significant disability and cognitive impairment.

- **Useful motor skills and tonicity**

It is most important to identify useful motor skills and tone, whether voluntary, involuntary, or automatic. Such useful motricity may involve the trunk and help the individual, for example, to hold his head up or actively bend over. In the upper limbs, motor control, tonicity, or involuntary movements may allow for grasping, upper limb positioning, or actions through the shoulders, arms, forearms, or hands. For the lower limbs and trunk, any motor control or tonicity that allows the individual to support his or her weight for transfers, propel themselves along the ground, or turn over in bed must be analysed and preserved or optimised.

- **Active motor skills**

Active motor skills are analysed during spontaneous, directed, or provoked activity. Motor control must be analysed in various situations and positions and at various times of the day. Such motor control can be analysed throughout motor development (Le Métayer, 1999).

- **Muscular strength**

Muscular strength is difficult to analyse because the muscle test is disturbed by difficulties in understanding the examination instructions, musculo-tendinous retraction, fatigability, tonicity disorders, and dyspraxia. However, an evaluation of muscular strength is necessary, as well as an evaluation of gesture selectivity. There are several existing scales. The MRC scale, rated from 0 to 5 *(see Appendix 4 and the link to Chapter 2.2, MRC scale),* for muscle strength is the most widely used, even if it is questionable in central injuries. Two scales are more commonly used for selective motor control, the Selective Control Assessment of the Lower Extremity (SCALE) and the modified Trost SMC (Smith, 2010).

- **Functional Evaluation**

There are many scales for the functional assessment of birth disabilities. These include two global assessment scales. The GMFCS (Gross Motor Function Classification of Cerebral Palsy) rates functional capacity for movement from 1 to 5 *(Annex 4, c.f. link Chapter 2.2, GMFCS scale, Imms C., 2010)*. The score is often 4 or 5 for polyhandicapped people. The Global Functional Motor Assessment (GMFA) (Alotaibi M1, 2014) is a quantitative scale of the child's global motor function and has been validated for cerebral palsy and other pathologies. For upper limb functionality, the MACS (Manual Ability Classification System) can be used *(Appendix 4, see link chapter 2.2, MACS scale)*.

- **Joint mobility**

The study of joint mobility is carried out after relaxation. The mobility of each joint must be measured, as well as the muscle length. Joint mobility depends both on joint stiffness and musculo-tendinous retraction.

- **Spasticity**

Spasticity results in involuntary resistance to an imposed movement. Such resistance increases with the speed of the movement. Spasticity can affect all four limbs and the trunk. It can be rated using the Modified Ashworth Scale *(Annex 4, see link Chapter 2.2, Modified Ashworth Scale) (Mutlu A, 2008)* or the Tardieu Scale (Gracies J-M, 2010).

- **Dystonia and dyskinesia**

Dystonia and dyskinesia are abnormal modes of posture (particular fixed segmental positioning) and/or movement, with involuntary, uncontrolled, generally abrupt, localised, recurrent, and sometimes

stereotypical movements. In the dystonic form, the basic tone is accentuated (hypertonia), whereas in the choreo-athetotic form, the basic tone is diminished (hypotonia). Rating scales useful for studies are little used in clinical practice (Stewart K, 2017).

- **Possible cerebellar disorders**

Cerebellar disorders cause global hypotonia, balance disorders, and voluntary movement control disorders (ataxia).

- **Stimulation of motor skills**

Motor skills and voluntary control are stimulated from childhood by including families and carers through motor education and multidisciplinary care (physiotherapy, balneotherapy, occupational therapy, psychomotricity). All activities that encourage taking initiative and using motor skills are to be encouraged, taking care to maintain the body in a favourable orthopaedic situation. Sensoriality should also be considered in all activities. Check the ability to walk (with or without assistance).

2.2.2 Therapeutic aspects

When muscle activity is excessive and uncomfortable, attempts are made to reduce it through medical or surgical treatment (M. R. Delgado, 2010).

- **Drug Treatment**

- Drug treatments for spasticity show only partial and inconsistent efficacy (Baclofen, Tizanidine, Diazepam, Dantrolene) and variable tolerance.
- Drug treatments for dystonia are only partially effective. They include anticholinergic antiparkinsonian drugs (e.g. Trihexyphenidyl/Artane®) or certain neuroleptic derivatives (Tetrabenazine/Xenazine®). However, the literature does not provide proof of the effectiveness of these treatments in cerebral palsy (Fehlings, D 2018).
- Treatment with intramuscular botulinum toxin injection is effective in decreasing spasticity or dystonia of the injected muscle. Botulinum toxin treatment also has an analgesic effect. Tolerance is generally good. There is a risk of generalized muscle weakness at high doses, called pseudo-botulism. Treatment should therefore be carried out with careful dosing (Strobl W. 2015). This treatment has multiple indications. It can reduce pain, facilitate nursing care, reduce the risk of retraction or stiffening, promote voluntary movement in the upper or lower limbs, and improve posture (cervical dystonia).

- **Surgical Treatment**

Surgical techniques (baclofen pump, radicotomy, deep brain stimulation) require a multidisciplinary evaluation (PRM, paediatric neurologist, neurosurgeon) to establish the indications and optimally manage the perioperative period.

- Intrathecal Baclofen treatment: this is a regional treatment for lower-limb spasticity that is resistant to usual treatments and disabling (difficulty in sitting, intractable pain, difficulties in allowing nursing care). This treatment is carried out by a specialised team. If clear improvement is demonstrated during an intrathecal Baclofen test by direct intrathecal injection, a subcutaneous intrathecal Baclofen (Lioresal®) pump can be implanted to reduce spasticity. This pump is compatible with a spinal arthrodesis or gastrostomy. It implies surgical constraints and regular filling by the transcutaneous route. This treatment may cause side effects, especially in case of withdrawal
- A rare treatment, called selective dorsal radicotomy, can be discussed (Ingale H, 2016) when hypertonia in the lower limbs is due to spasticity and prevents sitting or leads to joint deformities and pain
- Decreased innervation of the muscle by phenolization or neurotomy can be discussed in the treatment of locoregional spasticity. Obturator neurotomy or phenolization of the obturator nerves is often proposed in association with tendon release procedures to reduce spastic hip adduction
- Deep cerebral stimulation, most often bi-pallidal, has very rare indications for polyhandicapped individuals with disabling involuntary movements because the response to treatment is inconsistent and only very partial for such an invasive surgical procedure.

2.2.3 Motor education, rehabilitation

Educational/rehabilitative care and the fitting of devices are aimed at avoiding harmful prolonged positions and promoting motor skills, however minimal, in a variety of ways. Such care must be carried out without inducing pain.

- Orthopaedic prevention starts very early: in neonatology. Simple manipulations are necessary in the dorsal position in slight abduction, with the child keeping the knees in front (kneecaps at the zenith), or in the ventral position, taking care to lift the buttocks from the plane of the bed. Prevention of upper limb shortening, especially shortening of the trapezius in hyper-extended children, is ensured by bending the head and bringing the upper limbs back along the thorax.
- In all cases, active motor skills are encouraged, and the baby is taught to use his sensory, relational and, above all, motor skills. The principles of orthopaedic prevention are thus laid down from the very first days.
- The motor education initiated by physiotherapists and psychomotricians then aims to develop motor skills according to specific sequences, with the aim of promoting motor evolution (M. Le Metayer 1993-1999) and providing various sensory information to refine perceptions. Even if the capacities are limited, the sequences and turning are continued as a preventive measure. The physiotherapist and psychomotrician guide and solicit the muscles to maximally bring them into play. They also rely on sensory stimulation to improve muscle tone.
- Maintenance of joint mobility and muscle length: mastery of the relaxation manoeuvres by the family and professionals contributes to the daily maintenance of muscle length and comfort by virtue of constant mobilisation.
- Care should be taken to avoid harmful positions, such as sitting on the ground with the buttocks between the heels or forced internal rotation of the hips ("W" shaped), so frequently adopted by children, which is harmful to the hips, knees, and feet, as it promotes antitorsion, distension of the cruciate and patellar ligaments, and tibial rotation.
- Adults must be able to have a period of daily free movement on a floor mat.

2.2.4 Equipment

Devices (corset-seats, orthoses, etc.) are key elements of prevention, as is the alternation of positions, which is fundamental. The person must not remain in the same position for hours and his or her motor skills must be continuously stimulated. Changes in position must be carried out in the presence of carers in conjunction with occupational therapists. New technologies facilitate the taking of casts with the use of video scanning.

- **Corsets and other seats**

The seat corset is suitable for both:

- The functional state, to facilitate the activities of daily life: meals, technical aids for communication, leisure activities, social life, etc. Of course, the goal is a horizontal gaze
- The neuromotor status, especially insufficiency against gravity. In severe cases, tilting the seat backwards is necessary to reduce the harmful effects of gravity, which aggravates abnormal axial postures, especially those that are cervicocephalic. At the same time, great attention must be given to the position of the head and cervical spine to avoid a hyper-extended position, which leads to feeding difficulties and can be the cause of misalignment. The head is tilted at an angle of approximately 30° with a custom-made head or neck support. To maintain the relaxed state, the upper limbs can be held by wrap-around armrests. An increase in hip flexion is sometimes useful to control powerful hyper-extended contractions, especially in dystonic patients.

Orthopaedic prevention: for the pelvis, the aim is to re-centre the hips and obtain a balanced pelvis in all three planes, sometimes imposing an asymmetry of abduction or flexion in complex situations; control of retroversion (keeping the pelvis in a slightly more physiological anteversion) is important, as it contributes to spine alignment and straightening, if possible.

The situation of the activity also guides the choice of position. Thus a short-seat corset promotes active straightening. This proposal must respect the fatigability of the individual and can be used in alternation with more passive seat corsets.

A seat corset with carbon stays allows mobility of the trunk in rotation, flexion, and inclination, if the child/adult is able to do so.

- **Foam saddles and fittings**

- Foam saddles are very helpful, inducing activity and re-centring of the hips; installed on a mobile support, they promote movement and independence, with front support for "straight-leg" installation.
- Made of firm foam: the seat in the "sit-fit" position (which allows the child to straighten and control the pelvis) and the rabbit walker (positioned on the rabbit walker, the child can play and move around without sitting with his buttocks between his heels but on a higher plane).
- Expanded foam installations of various densities are used for individuals with complex neuro-orthopaedic conditions. The objective is to maintain comfort. The integration of visco-elastic foam in risk zones (gibbosity, greater trochanter, etc.) prevents skin complications caused by excessive support.

- **Verticalization orthoses**

Verticalization orthoses are most often posterior (sometimes anterior to stimulate the posterior plane). They should promote recentring of the hips (through their abduction and slight flexion) and maintain the length of the legs. The position of the feet is controlled by orthopaedic shoes on moulds or with a rigid shell to avoid common aggravation of the valgus. Radiographic control is necessary to verify centring of the hips and pelvic balance. Verticalization improves vital functions, such as digestion (transit), urination, and respiration, promotes growth, and prevents osteoporosis. The benefits, in terms of stimulation, are obvious. The regard of others changes when you are standing. Its integration into activities facilitate its acceptance. The stability of the device is imperative, as well as its ease of use (a harness that allows the child to be placed in the horizontal position of the orthosis with the use of a lift system). Verticalization is performed on a daily basis, if possible, for at least one hour or more, depending on the tolerance of the individual.

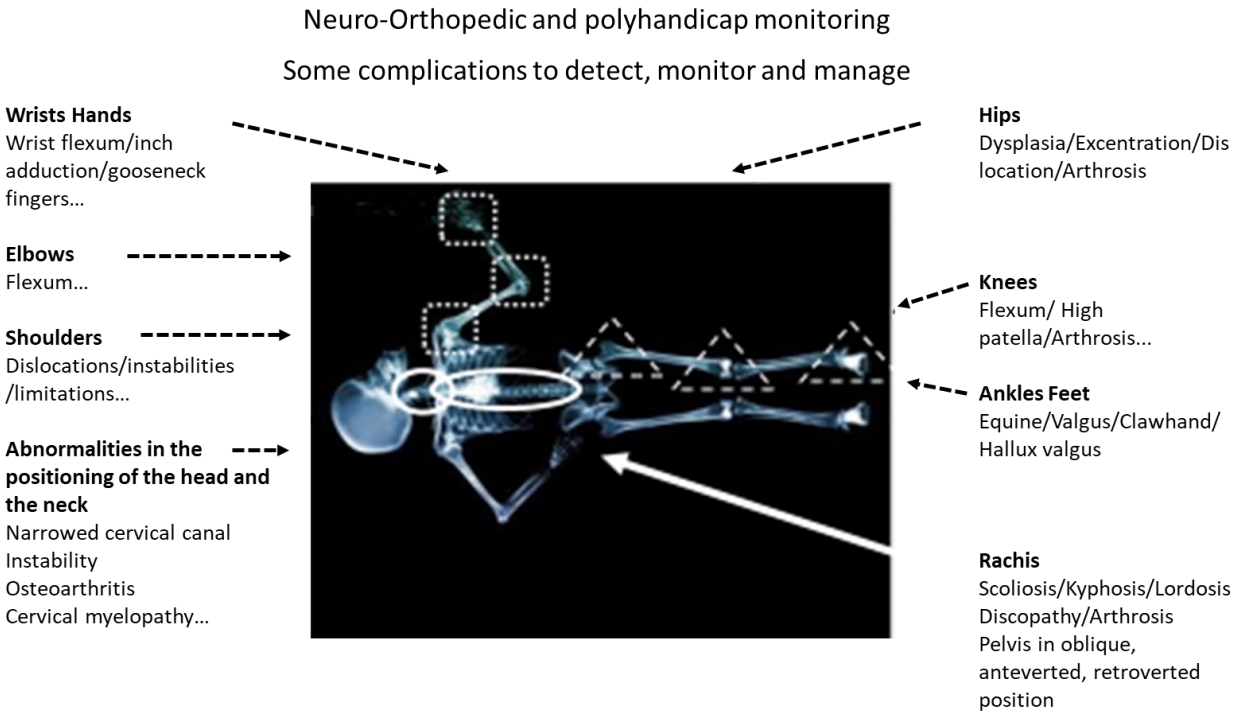
Verticalization must not to be abandoned in adults, even if verticalization orthoses for adults are more complex to make and use.

- **The decubitus orthosis**

The objectives are the same as for other devices: orthopaedic prevention, indolence, and comfort, with the search for the position of maximum relaxation and sufficient inclination of the trunk from the horizontal to avoid gastroesophageal reflux.

- The corset slows the progression of scoliosis. The "Garchois" corset is the most suitable for respiratory problems. A corset derived from the CTM (Cheneau Toulouse Munster) is often proposed.
- Various posture orthoses are used: leg braces, simple foot braces; anti-valgus braces, varus braces, with articulated lateral post; therapeutic shoes, with or without a rigid shell; orthopaedic insoles; orthopaedic shoes on moulds necessary for verticalization; functional orthoses (anti-shin with carbon blades, articulated, anti-talus) for those who walk (guided walking).
- Successive lengthening plasters: made after injection of toxin, successive lengthening plasters are used for subjects who stand upright or walk (guided walk).

2.3 Neuro-Orthopaedic and Bone Disorders



(Source Dr Célia Rech)

Neuro-orthopaedic disorders are very common in people with a polyhandicap. They develop quickly, sometimes as early as during the neonatal period.

Inadequate **bone mineralization is** also frequently observed and must be detected.

Neuro-orthopaedic disorders are a source of pain, discomfort, and loss of function, and have visceral consequences (respiratory, digestive). They require the rapid implementation of physical rehabilitation and physical care or surgical programs.

The analysis of neuro-orthopaedic disorders (Viehweger 2007 - Berard 2010) must attempt to differentiate between primary anomalies linked to neurological pathology (spasticity, lack of control, co-contractions, etc.), secondary anomalies linked to the growth of the child (musculo-tendinous retraction, hyperlaxity, structural deformation of the bones in the different planes, etc.), and tertiary anomalies, which are compensatory anomalies of neuro-orthopaedic disorders.

These disorders affect the median axis (oro-facial sphere, spine) and the limbs (upper and lower limbs).

In children, the emphasis is on early care, sometimes as early as during the neonatal period. The re-educational/motor education (Michel Le Metayer 1993 - 2006) and rehabilitative approach must be educational and the child must benefit from close, multi-year follow-up in physical and rehabilitation medicine (PRM), which is fundamental during this period of development. The PRM assessment makes it possible to determine the child's needed rehabilitation (physiotherapy - psychomotricity - occupational therapy - speech therapy) and technical equipment and aids (aid in sitting and lying down - verticalization mode - movement mode - limb orthoses - corsets - orthopaedic footwear, etc.). Studies are still needed to clarify the effectiveness of these various approaches (J. Robertson Journal of Applied Research in Intellectual Disabilities 31, 11-28, 2018). Physical medicine follow-up makes it possible to define the neuro-orthopaedic surgery needs (tendon lengthening, correction of bone disorders of the limbs, osteotomies, neurotomies, arthrodesis, etc.) and manage tonus disorders (spasticity - dystonia, etc.) by various methods (rehabilitative approach, drug treatment, Baclofen pump, botulinum toxin injections, etc.).

Screening for osteoporosis may be considered (see section on osteoporosis) and calcium and vitamin D intake are monitored and supplemented, if necessary. Mobility is encouraged.

The main objectives are to predict and guarantee the possibility of harmonious musculoskeletal growth, preserve orthopaedic capacity and bone mass, maximise the possibility of functional development (locomotion, manipulation, interactions, communication, cervicocephalic mobility, etc.), maintain the capacity to sit and lie down, guarantee comfort and prevent and control pain, and prevent the risk of fractures, as well as establish the education of caregivers (prevention of lumbago, etc.).

In adults, the objectives are mainly to prevent and control pain, control spasticity, dystonia and disabling dyskinesia, prevent and control the aggravation of musculoskeletal deformities of the spine and limbs (Hodgkinson 2002) and the risk of skin problems or osteoporosis with spontaneous fractures, ensure comfort in the various postures necessary for all activities of daily life, facilitate transfers and positioning, as well as nursing and hygiene care, optimize relational conditions, and minimize the respiratory and digestive impact.

Minimum annual monitoring by a PRM is recommended. Prevention and screening for osteoporosis is required. Physiotherapy, work on posture, verticalization, orthoses, corset-seats, orthopaedic shoes, moulded mattresses and large orthotic-foam devices for resting, adapted physical activities, psychomotricity, motor freedom on mats, anti-spastic treatments (including toxin), and orthopaedic surgery are part of the therapeutic arsenal.

Caregiver education also remains one of the challenges of monitoring.

2.3.1 Lower limbs

- **Hip:**

The hip is strategically positioned between the trunk and lower limbs. Its functioning has an impact both upstream on the spine and downstream on the lower limbs, potentially compromising lying down, sitting, standing, and movement. Limited abduction and eccentricity of the hip are frequently observed, with a reciprocal head-acetabular development defect. Disorders of the hip are all the more severe, as standing and walking are difficult or impossible. There is a close relationship between pelvic obliquity, the direction of scoliosis, windswept hip deformity, and the side of hip offset, as shown in the study by David Porter in Oxford of 747 child/adult patients with non-walking cerebral palsy (David Porter, 2008).

The risk of excentration increases with growth. The polyhandicapped non-walking quadriplegic child is the most at risk of hip excentration (Lespargot, 1991) and should be monitored in the early years of life by annual radiography (Shrader MW J Am acad Orthop Surg 2019). The risk is greater when the child presents with a windswept deformity of the lower limbs with one limb most often in flexion, adduction, internal hip rotation with the contralateral limb in flexion, abduction, or external hip rotation. The pelvis is often oblique and may be associated with lumbar scoliosis. The risk of progression to dislocation should be assessed and controlled, especially in non-walking children.

PRM treatment consists of adapting the rehabilitation to the deformity and reducing the activity of the luxating muscles (adductors, hamstrings, etc.). Sitting and lying positions aim to avoid the luxating position (hip abduction). Surgical treatment for musculo-tendinous lengthening can help to reduce or stop eccentricity. Hip-conserving bone surgery is sometimes proposed (femoral varus osteotomy, pelvic osteotomy). When the hip is dislocated, an evaluation of the impact of such a dislocation on the quality of life of the adult or child is necessary. Depending on the pain and possible difficulties in sitting, various treatments may be proposed: treatment of spasticity of the dislocated muscles, infiltrations of the neo-articulation for pain relief, wide periarticular tendon releases for pain relief, total hip replacement, and sometimes surgery to resect the femoral head and neck (D'Anjou 2016/ Khouri 2016).

Conversely, "froggy" attitudes and adduction defects are mainly observed in patients without lower limb support with overall hypotonia. In the supine position, the lower limbs are in bilateral external flexion-abduction-rotation. This promotes anterior dislocation of the femoral head. In this context, the seated and

supine positions aim to avoid excessive external abduction-rotation of the hip, and rehabilitative care aims to preserve the possibility of passive adduction of the hips as much as possible.

The appearance of hip flexum is promoted by a permanent sitting position. Severe hip flexum can compromise the ability to stay in a ventral decubitus or dorsal decubitus position; it is responsible for instability of the lower limbs. Compensatory lumbar or dorso-lumbar lordosis gradually develops and can become fixed. Physiotherapy is important to prevent such a flexum. Resting in a state of hip extension for an extended period of time is desirable, in a lying position or in verticalization to not be permanently in a position that favours short hip flexors. Treatment with botulinum toxin or hip flexor lengthening surgery can be offered.

Stiffened hips in extension are rarer. The lower limbs are thus in hip extension - external rotation favouring anterior hip dislocation. The sitting position may be compromised. Spasticity and/or dystonia of hip extensors (gluteus maximus, hamstrings) are assessed in the PRM. Local (botulinum toxin) or regional (baclofen pump) treatment may be offered. A high-quality device to aid the sitting position is necessary to preserve the sitting position for as long as possible by controlling the pelvis as well as possible. Neuro-orthopaedic surgery may be necessary if the sitting position is compromised by changes in the patient's condition (e.g. high hamstring release).

- **Knee:**

Knee flexum is often associated with a high patella with an active knee extension defect. Such abnormalities particularly develop in patients who do not stand and spend long periods sitting that are not compensated by extension or verticalization movements/postures. Severe knee flexum compromises comfortable sitting because it leads to pelvic retroversion with lumbar kyphosis. Devices that aid sitting should avoid leaving the knee free in hyper-flexum. Reclining (positioning cushions, moulded half-mattress, knee-ankle-foot orthoses) or verticalization aim to maintain knee extension for as long as possible. A treatment for hamstring spasticity or musculo-tendinous lengthening can be discussed, depending on the evolution or impact.

Recurvatum is less frequent. It is important to control it if it worsens and to avoid hamstring stretching in physiotherapy.

The torsion of the leg can be either in the direction of an external hypertorsion with flat foot valgus abductus or a rarer tibial hypotorsion.

- **Ankle:**

The most frequent deformity is equinus with limited dorsal flexion, a direct consequence of hypertonia of the sural triceps associated or not with weakness of the foot lifters. The natural evolution is towards fixed equinus.

Planovalgus abductus foot is very often associated with more or less marked equinus, which can progressively lead to a medio-tarsal dislocation in dorsal flexion and abduction associated with a collapse within the talus and a flattening of the plantar arch with secondary appearance of a valgus of the hind foot.

The talus foot, with limited plantar flexion, can be a true talus with excessive dorsal flexion of the hindfoot. It can also be a pseudo-talus, a talus of the forefoot associated with an equinus of the hindfoot masked by the medio-tarsal break.

The varus adductus foot consists of a varus of the hind foot associated or not with an adduction of the forefoot with retraction of the hallux adductor; it progressively evolves towards the varus hollow foot (varus, adduction, and supination).

Physiotherapy adapted to the deformities is desirable to maintain muscle length and joint flexibility. The use of night and/or day orthotics, such as "leg boots", aims to contain the evolution of the deformities. Successive lengthening casts, made after injection of botulinum toxin into the triceps, are sometimes used to control equinus in patients with the possibility to practice verticalization.

Adapted footwear is proposed, often consisting of moulded slippers or orthopaedic shoes. During growth, foot orthotics help to contain deformities. Botulinum toxin treatment of the prevalent muscles causing the deformities can be offered, as well as neuro-orthopaedic surgical treatment. Foot care is recommended if there are calluses or skin problems.

- **Deviated toes:**

Hallux valgus can regularly worsens, with pressure sores, for example, on the medial edge of the hallux (Van de Velde. Dev med child Neurol 2018). It can be associated with a dislocation of the sesamoid belt and metatarsophalangeal osteoarthritis.

Spasticity of the flexor muscles of the toes, whether or not it is associated with spasticity of the extensors, leads to claw toe. The position of the metatarsophalangeal joint can vary, and the interphalangeal joints are in flexion. This type of foot is frequently found in varus hollow feet and dystonic children. These deformities are responsible for problems with shoes and can cause pain and limit the possibility of verticalization or transfer.

Special attention is paid to maintaining passive mobility of the toes, the detection and treatment of skin lesions, pedicure care, proper fitting of foot devices (toe orthoses, orthopaedic shoes, moulded shoes, therapeutic shoes), and treatment of spasticity by botulinum toxin (e.g. in claw toe). Neuro-orthopaedic surgery to correct deformities is considered in cases of discomfort, pain, or unfavourable evolution.

2.3.2 Upper limbs

Retraction of the upper limbs occurs earlier and is more frequent with more severe motor impairment. Such retraction primarily concerns patients who cannot pick up objects and are severely limited, even for simple motor actions (Manual Ability Classification System -MACS- level V) (Hedberg-Graff J. 2019).

- **Shoulder:**

Anterior dislocation is a risk frequently observed in connection with the candlestick attitude of the upper limbs. Sitting and lying positions seek, in particular, to contain excessive external rotation of the shoulder. Rehabilitative management, possibly combined with botulinum toxin treatment, aims to maintain the shoulder's ability to maintain adduction-internal rotation.

- **Elbow:**

The contraction patterns associated with neurological hypertonia most often lead to a sometimes very severe flexum of the elbow, as well as only limited supination. Flexum and limited supination greatly reduce the perimeter of the grip, interfere with dressing, and can lead to maceration or skin lesions. The management strategy consists of preserving elbow extension as much as possible (physiotherapy, elbow extension orthoses, chair installation).

- **Wrist:**

Most often, flexum is observed in the wrist. Severe forms eventually lead to carpal deformity and pain. Postural support of the wrists (orthoses), associated or not with toxin injections, should be applied early.

- **Hand:**

Thumb adductus and swanneck finger deformities are very common. These deformities limit the gripping capacity and can lead to maceration or skin problems between the fingers.

2.3.3 Trunk

- **Scoliosis**

Scoliosis is a serious and frequent deformity of the spine that occurs in all three planes. Its severity corresponds with that of the motor impairment (Saito N. 1998). It is often progressive in adults when the curvature is greater than 40° at the age of 15 years. It is linked to asymmetry of muscle tone and retraction. The curvature is most often thoraco-lumbar, involving the pelvis, with a very large rotation of the vertebrae. It is possible to observe lumbar scoliosis with hyperlordosis, thoracic scoliosis with hyperkyphosis, scoliosis in inversion of the curvature, with a flat thoracic back, and scoliosis with thoracolumbar kyphosis. Severe

scoliosis affects respiratory capacity and digestive functions. **Rehabilitative management and positioning** aim to maintain spinal and thoracic flexibility, maintain respiratory capacity, and provide comfort. The use of a corset slows the evolution of scoliosis (De Lattre C. 2005). **Several types of corsets** can be proposed, the Garchois corset is one of the best adapted for maintaining respiratory function. **Surgical treatment** can be proposed if the scoliosis is severe, progressive, and painful.

The purpose of the intervention is to stop the evolution of scoliosis, reduce the existing curvature, and make it possible to eliminate the wearing of a restrictive corset.

The surgeon seeks to maintain the best possible balance between the pelvis and shoulders in the frontal and sagittal planes. He tries to balance the pelvis in the frontal plane to avoid asymmetrical buttock support, which can cause pressure sores and make sitting uncomfortable, and corrects the position of the pelvis in the sagittal plane (excess anteversion/lordosis or retroversion/kyphosis) by trying to achieve a harmonious lumbar curvature.

At the level of the thorax, it is desirable to balance the shoulders as well as possible in the fontal plane to favour verticality of the head and neck. In the sagittal plane, the correction of excess kyphosis aims to maintain the best possible thoracic capacity for the future and a horizontal gaze. Given the complexity of the procedure, the benefit/risk ratio of spinal fusion should be discussed by PRM and surgical teams with experience in this field.

The reference surgery is posterior spinal arthrodesis (more rarely posterior and anterior), which consists of fixing the spine with bone grafts held in place by a metal mount. This surgery is performed when the rib cage is sufficiently developed, and the deformity is still partly reducible. This major surgery, with a high prevalence of general and infectious complications (Vialle 2014), cannot be proposed if the young person's general condition is too precarious.

The young person must arrive at the operation in the best possible condition: the skin condition must be good, and skin lesions, acne, and maceration at the operating sites must be avoided. It is sometimes necessary to treat osteoporosis beforehand to obtain a better fit. Respiratory management must be optimised (see chapter on respiratory disorders). From a nutritional point of view, weight gain of polyhandicapped patients who are lean before the operation is desirable, if possible, to anticipate post-operative weight loss. Correction of any existing undernutrition is essential because undernutrition increases the risk of complications. Sometimes it is necessary to ask for a gastrostomy before the operation. It is also advisable to retain as much spinal flexibility as possible to facilitate the per-operative correction of curvature. Sometimes, the medical-surgical team proposes preoperative traction by cranial halo to optimize the per-operative reduction of curvature. Arthrodesis is a long operation under general anaesthesia, with high blood loss. Post-operative management can only be carried out by trained teams. A post-operative stay in intensive care is often necessary because it allows the management of ventilatory weaning, compensation for blood loss (transfusions...), and monitoring for the risk of infection and ensures nutritional intake and nursing care without risking the spinal assembly.

After resuscitation, a specialized PRM team is required. Spinal immobilization by post-operative corset is sometimes indicated, its duration depending on the type of scoliosis, the type of assembly, and the evolution of the grafts.

Instrumental surgery without definitive or progressive grafting (Miladi L. 2016) can also be proposed. This is a surgical technique that consists of a posterior operation with solid bipolar fixation (proximal thoracic upper and distal pelvic). A telescopic frame allows re-tensioning of the rods (every 18 to 24 months) to progressively reduce deformities and follow growth. The approach is less extensive (lumbosacral and upper thoracic cutaneous approach) and the post-operative consequences are less severe but the complications (especially infectious) are still important. This type of operation can be proposed earlier in childhood and to patients who would not tolerate conventional arthrodesis.

- **Kyphosis:**

Kyphosis is a deformation in the sagittal plane, isolated or related to scoliosis. The kyphosis can be lumbar, lumbo-sacral, or dorso-lombo-sacral with a retroverted pelvis. It decreases respiratory capacity. Positioning in the dorsal decubitus position at night is preferable and rehabilitative care is important to maintain respiratory capacity. The sitting position must include a high backrest ± a posterior rocker to prevent the trunk from projecting forward. Spinal arthrodesis surgery is sometimes necessary.

- **Lordosis:**

Hyperlordosis is mostly observed in the lumbar region. It most often manifests as lordo-scoliosis with the disappearance of dorsal kyphosis, a large flat back and lumbar hyperlordosis, resulting in a large anteversion of the pelvis, or a kyphoscoliosis, with thoracic hyperkyphosis and compensating lumbar hyperlordosis.

- **Cervical spine:**

Deviations of the head and neck are variable. Anterior hypotonic head fall, dystonia in all three planes, musculo-tendinous or spinal limitations, and deformities in inclination, rotation, and extension all affect participation in the activities of daily living, particularly meals and swallowing. Tonic or dystonic deviation is to be particularly monitored (dynamic radiographs/MRIs) and managed because of the long-term risk of upper spinal cord compression due to cervical myelopathy. When indicated and necessary, the reference technique of spinal cord decompression for dystonic patients is laminoplasty (Zhou 2016/ Jameson 2012).

2.3.4 Bone fragility

The prevention of bone fragility and fractures is essential from a very early age. Bone density and quality are disturbed by the absence of bearing weight, immobilisation, undernutrition, calcium and vitamin D deficiencies (particularly in adolescence), and corticosteroid and anticonvulsant treatments (phenobarbital, carbamazepine, phenytoin), which weaken the bone to the point of causing pathological fractures, i.e. spontaneous fractures or fractures following low-kinetic trauma. The clinical warning signs, apart from spontaneous fractures, are pain upon mobilization or unexplained pain at rest, as well as sleep disorders, weight loss, and alteration of the general condition with motor regression.

The phospho-calcium test is used to detect vitamin D deficiency and assess bone damage. The calcinuria/creatinuria ratio is an indicator of bone turnover, especially in children. It can be performed on urine using a urinocervix, a urine collection bag, urine collected on clean compresses placed in the diaper, or as a last resort, catheterization.

An assessment of nutritional status should be conducted, taking into consideration the correlation between bone fragility, undernutrition, and hormone secretion.

Bone densitometry is recommended if risk factors for bone fragility are present. This should be carried out on the spine in the absence of arthrodesis or on the long bones in paediatric hospitals equipped with specific software. Only the Z score is relevant.

Therapeutic management should consist of the prevention and active treatment of bone fragility. A program for the prevention of brittle bone fragility should be established for all patients.

- **Prevention must be systematic and include:**

- Verticalization and motor activity
- Adequate protein intake
- Age-appropriate calcium intake
- Vitamin D supplementation to reach a level of > 20 ng/ml 25 0H vitamin D, usually given orally by 80,000- to 100,000-IU ampoule every three months
- Hormonal treatment, in case of delayed puberty or early puberty (estimated in relation to chronological age).

- **Treatment**

In cases of pathological fracture, bone pain, or an indication for major surgery, treatment with bisphosphonate IV is indicated if the patient has proven bone fragility by densitometry.

Treatment with bisphosphonates, by pamidronate or zoledronate infusion, is carried out in a hospital setting.

The frequency of infusions (Wiedeman 2018) should be discussed and depends on the choice of medication (most often every 3 to 6 months for children and sometimes up to every year for adults) and for a period that is still a subject of debate. Most care teams propose two years, as in the study by Dr Anne-Sophie Lambert (A.S. Lambert 2017). Bisphosphonates block bone resorption and remain fixed to the bone matrix for a long time.

Adverse reactions include an influenza-like syndrome type immune response for two to three days after the first infusion and hypocalcaemia. Following treatment, a rapid decrease in pain within a few days, with functional improvement for some patients (Rett Syndrome), has been observed, as well as an improvement in bone strength, with little recurrence of fractures.

In children, "per os" treatment (alendronate) is not indicated, as it is likely to cause esophagitis; it is increasingly proposed only for the adult population. New treatments are being considered, such as parathormone or denosumab.

For adults, screening should be performed if it was done when they were children. There are sometimes difficulties in finding sites for bone densitometry studies that have not been altered by surgery (hips and spine). The indications for preventive and curative treatment are the same as for children.

Bone density improves in all patients. An ECG and dental examination are recommended prior to treatment with bisphosphonate IV. No dental procedures are contraindicated during treatment.

2.4 Epilepsy

Epilepsy is one of the most common co-morbidities of polyhandicapped individuals. The Eval-PLH study found a prevalence of 55 to 60% in children and among them, 25 to 31% have drug-resistant epilepsy and 25 to 33% have a history of epileptic disorder (Rousseau 2019, to be published).

2.4.1 Epileptic Seizures - Epilepsy

A seizure is the transient occurrence of signs due to excessive or abnormal synchronous neuronal activity in the brain. (Fischer 2014) It can manifest as:

- A sudden change in the state of consciousness (loss of consciousness)
- An alteration in the perception of the environment ("contact break")
- Motor, sensitivity, sensory, psychic, or vegetative phenomena.

The clinical signs depend on the topographical origin and the spread of neuronal discharges *(see Annex 4 Cf 2.4, Figure 1: classification of seizures)*:

- Widespread crises quickly evolve to involve bilateral neural networks; if they exist, motor manifestations are bilateral and roughly symmetrical from the outset
- Focal seizures involve neural networks limited to a single hemisphere; they are characterized by one or more symptoms, which may be subjective ("aura"), motor, vegetative, and/or dyscognitive, with altered or preserved consciousness/response to stimulation. Focal seizures may become secondarily generalized, hence the importance of the very first clinical sign of the seizure, which is of great value in localizing the focus.

Epilepsy is a brain disorder characterized by a long-lasting predisposition to generate epileptic seizures and by the neurobiological, cognitive, psychological, and social consequences of this condition (Fischer 2014).

Epilepsy (International League Against Epilepsy-ILAE 2014) is referred to as epilepsy in one of the following situations:

- At least two unprovoked seizures occurring more than 24 hours apart
- A single spontaneous crisis and the likelihood of subsequent crises occurring within the next 10 years, similar to the general risk of recurrence (at least 60%) observed after two unprovoked crises
- An epileptic syndrome has been identified.

2.4.2 Clinical aspects

In the case of the polyhandicapped, epilepsy is secondary to the brain injury or genetic disease causing the polyhandicap. It is often severe and within the framework of epilepsy with structural or metabolic causes. It represents one of the most frequent comorbidities among polyhandicapped people, but there are few studies on this subject in the literature.

- **Two situations can arise:**

- Sometimes, seizures of any type are part of the neurological syndrome, along with motor, sensory, and other disorders, and provide only an additional element of concern
- On the contrary, epilepsy may appear to be at the centre of medical problems in the context of epileptic encephalopathies.

- **Clinical study**

Regardless of the type of seizure, clinical observation of seizures is essential and several elements should be noted: for caregivers, it is important to describe them in simple words rather than trying to type the seizure *(Appendix ch 2.4, Figure 2, guide to help describe seizures)*.

The epileptic nature of a seizure is sometimes difficult to diagnose**.** There are therefore two major risks, with serious consequences: misunderstanding the epileptic nature of certain paroxysmal manifestations in a polyhandicapped child (or missing brief, poorly symptomatic seizures, particularly at night) or wrongly diagnosing epilepsy in the face of acute manifestations of other origins.

2.4.3 Treatment of seizures

Be vigilant about the duration of the seizure, as it is a factor that is known to aggravate the child's previous deficit if it is prolonged:

- For a short seizure: no special treatment is required. Respect the usual behaviour in the event of a seizure
- If the attack lasts more than 5 minutes = intra-rectal administration of Valium: 0.5 mg/kilo/administration (maximum 10 mg or 1 ampoule). No risk of respiratory depression by the intra-rectal route.

Valium can be renewed after 10 to 15 minutes. Beyond half an hour, the situation is serious, and hospitalisation is necessary to resort to injectable medication under monitoring.

Alternative: Buccolam (active ingredient Midazolam): MA (Marketing authorization) from 3 months to 18 years (from 3 to 6 months only in hospitals); convenience of administration due to the intra-jugal route.

2.4.4 Background treatment of epilepsy

The existence of a polyhandicap does not affect the choice of medication sequence, and the type of seizures, age, epilepsy syndrome if present, and aetiology are considered (Dulac, 2005).

- **Goals of treatment.**

The goal is not necessarily the disappearance of all seizures. The overriding principle is to find a compromise between the risks associated with the seizures and their possible consequences on the person's neuropsychic state and the fear of altering the patient's cognitive abilities and alertness as a result of the side effects of treatment. This balance is always difficult to achieve and needs to be explained to families and healthcare teams.

An increase in seizures should always elicit the search for a decompensating factor: pain of any origin (digestive, osteo-articular, etc.), pulmonary or urinary infection, or constipation. Otherwise it is necessary to evoke "intrinsic" aggravation of the epilepsy.

- **Type of treatment (See Appendix 4 - 2.4 Table 1 and Figure 3)**

It is of fundamental importance that observations and information be shared between the medical specialist, the treating physician, families, and teams to avoid any misunderstandings concerning treatment.

Treatment that lowers the epileptogenic threshold should be avoided if possible: neuroleptics, sedative antihistamines, opioids (Tramadol), and some antidepressants or antispasmodic drugs (Baclofen).

Adjunctive treatments should be avoided, as they may reduce the effectiveness of certain anti-epileptic drugs through drug interactions and promote seizures (e.g. certain antibiotics).

The following rules must be observed:

- Begin with a MONOTHERAPY, with a major antiepileptic, known as a first-line antiepileptic
- In case of failure (at least 1 month of treatment), change the medication
- If the second drug fails, changing to a third drug may be effective. Otherwise combine a second drug in BITHERAPY (using drugs with different targets of action), knowing that only 20% of patients will be improved by dual therapy
- Avoid POLYTHERAPY (Egunsola 2016) as much as possible, because no synergy of several drugs has been demonstrated. No study has shown with certainty the effectiveness of three drugs over two, and the accumulation of side effects of drugs, particularly on alertness, has been proven. On the contrary, it is necessary to know how to reduce the treatment of a child on polytherapy, which can lead to an improvement in more than 50% of cases (M Wu 1993).

- **Drug-resistant epilepsy**

The International Epilepsy League (ILAE) recommends that the epilepsy should be considered to be drug-resistant following failure of two antiepileptic drugs (adequate, well tolerated, and appropriately chosen), used in mono or dual therapy (Kwan 2010) (prevalence of 61.9% at the Centre de Vendin le Vieil for 87 patients). However, it should be noted that the error rate of diagnosis is from 10 to 20%, hence the need for video coupled with EEG.

- **Other therapeutic possibilities**

- A ketogenic diet is among the therapeutic resources for drug-resistant epilepsy in children (Dulac 2005).
- Vagus nerve stimulation provides indirect cerebral stimulation through the relays of the nerve centres.
- Surgical treatment consists of the removal or sometimes disconnection of the brain regions generating the seizures; it is possible for certain focal epilepsies in polyhandicapped children.
- Deep brain stimulation is currently being studied.

In addition, the entourage and team must remain vigilant in the event of disturbances of the person with a polyhandicap (intercurrent illness, fever, lack of sleep) by promoting rest, controlling fever, and hydration to avoid a resurgence of seizures.

Finally, any abrupt changes or discontinuation of antiepileptic treatment should be avoided, e.g. in case of vomiting. These treatments should be retained, sometimes even when fasting before surgery.

2.4.5 Surveillance is essentially clinical

- **Side effects of drug treatment:**

Side effects are particularly common, especially for the cognitive or psychological effects of sometimes difficult diagnoses. It is impossible to mention them all, but some can be serious (Mathieu 2017): hepatic, endocrinological, haematological, cutaneous, or ocular toxicity, the appearance of urinary lithiasis, etc. The most powerful molecules are also those with the most pronounced side effects: Topiramate can induce

encephalopathy (alteration of consciousness and extreme anorexia) that is sometimes so serious that parents prefer to return to the previous state of pharmaco-resistance.

If new drugs are introduced, always check the compatibility with the existing anti-epileptic treatment to avoid potential side effects.

- **Interest of blood assays in bi or polytherapy:**

➢ If seizures persist despite correct dosage
➢ To recognize adherence to or good absorption of medication
➢ If cognitive impairment, signs of toxicity, or unexplained side effects occur.

It is not necessary to repeat the assays for a child with balanced epilepsy.

- **Interest of EEG :**

The primary interest is to establish a syndromic diagnosis. An EEG is desirable in children, but the frequency of EEG monitoring in adults depends on the clinic. An EEG is desirable in case of worsening epilepsy, change in seizure type, or worsening or onset of cognitive impairment (ideally video EEG with wakefulness and sleep patterns). It may be necessary to offer pre-medication with Hydroxyzine or Melatonin, while avoiding Benzodiazepines.

- **Interest of brain imaging.**

Brain imaging must be carried out systematically to make an etiological diagnosis, particularly that of a focal lesion, as well as in the monitoring of certain pathologies, such as Bourneville's tuberous sclerosis (1x/year) or if the diagnosis is uncertain.

2.5 Pain

Those with a polyhandicap have multiple organ damage, often causing pain that they have little opportunity to communicate accurately to those around them or to those who examine them. Pain management is a prerequisite for any treatment, as it can block an already limited relational process. Moreover, it is a legal, but above all, ethical obligation.

Various surveys show that the pain of non-communicating people is still underestimated, undervalued, and under-treated. However, it has been proven that toddlers and polyhandicapped people suffer, on the contrary, from a deficit in the modulation of the pain message due to immature or damaged regulatory systems and a lack of cerebral integration of the meaning of their pain: their pain is therefore more global and accompanied by a major component of anxiety.

Prevalence: Prevalence surveys have been conducted in only a small number of studies in the context of profound intellectual and multiple disabilities (PIMD). The study of Stallard (2001) of a small number of children (34, mean age 9.4 years) revealed that 73.5% of children had at least one painful episode over a two-week period. The Eval-PLH study showed much lower figures: pain affects 4 to 11% of children and clearly increases after the age of 35 to reach 60% after the age of 50 (Rousseau 2019) *(see Annex 4 ch 2.5 Table 1, summary of 3 surveys).*

2.5.1 Physiopathology

- **Pain has four components:**

➢ A sensitivity component: neurophysiological mechanism for detecting the painful stimulus and analysing its intensity, location, and quality = what the person feels
➢ An affective and emotional component: unpleasant connotation linked to the perception of pain, which can lead to anxiety or depression = what the person experiences
➢ A cognitive component: mental processes likely to modulate the experience of pain = the interpretation of the situation, with references to past experiences

- A behavioural component: all the observable, physiological (vegetative phenomena, for example: redness, tachycardia, etc.), verbal (complaints, moaning), and motor (postures, analgesic attitude, mobility or agitation, limitation of activities, etc.) manifestations.

- **Acute and chronic pain *(see Annex 4 ch 2.5, Table 2)***

If it is persistent, an initially simple pain symptom (pain-alarm) can change and become a syndrome in its own right (pain-disease): the intensity of the emotional and direct signs of pain diminishes and the person stops complaining and withdraws into himself. After a few days, a pseudo-depressive semiology sets in, consisting of psychomotor atony: a frozen, inexpressive, even hostile face.

The different types of pain found in someone with a polyhandicap are the same as those found in an "ordinary" person.

2.5.2 The necessity and difficulties of evaluation

For these people, with no meaningful verbal language and limited opportunities for communication, interviewing parents and others is of paramount importance. The focus should therefore be on them:

- Assessment of the anxiety (AC) or emotional component of pain (crying, mimicry, moaning, etc.)
- Searching for direct signs of pain (DSP): analgesic attitude, protection of painful areas, defensive reactions, etc.
- The analysis of psychomotor alterations (PMA) resulting in psychic regression and an exacerbation of psychotic manifestations through interrogation of the entourage.

- **Analysis of unusual behaviour: signs of orientation**

The existence of pain is to be sought through the interrogation of relatives and careful observation of someone who does not communicate verbally to look for unusual changes in his or her behaviour. Some signs taken separately (e.g. crying) are not necessarily specific to pain, but their association can be significant. This is the interest of scales that group these signs together with the possibility of establishing pain scores.

- **The clinical examination: 4 main points**

- Watch: observe the person at rest, when active, during play, while undressing, during emotional interactions, etc. and note the position he or she chooses, in particular whether he or she obstinately maintains the same position, feverishly seeks an analgesic position, etc.
- Listen: interviewing parents and caregivers. "He (she) is not as usual"
- Dialogue: talk to the person to be able to examine him or her calmly
- Search: Clinical examination, if possible, in pairs. One maintains playful and empathetic contact with the person while the other examines him gently:
    - Explore sensitivity by light touch: touch with a familiar object to identify hyperalgesic areas, followed by local superficial palpation, then gentle pressure of muscle masses then bones; passive or even active joint examination if possible
    - Check for changes in pulse or respiration
    - Carry out a motor exploration.

- **Evaluation by a hetero-evaluation scale**

The assessment of pain without a specific tool varies from person to person, especially for emotional reasons. A validated and standardized scale:

- Allows an objective and reproducible evaluation of a subjective phenomenon
- Allows the evaluation of the initial intensity of the pain and visualization of its decrease after treatment
- Constitutes a common tool for all personnel using the same language
- Forces one to consider pain and reassess it from time to time.

- **When should a hetero-evaluation scale be used (always at least in pairs)?**

- When confronted with any pain, even if it seems obvious (initial intensity)
- When faced with abnormal behaviour (behavioural disorders, self-mutilation, aggressiveness, etc.), or an increase in epileptic symptoms

- For the regular reassessment of drug and non-drug therapy
- During any presumed painful treatment: allows the effectiveness of the premedication to be checked
- Post-operatively
- Before a consultation with the orthopaedic surgeon, pain being one of the elements of any decision, or before any consultation, especially physical medicine.

- **The different evaluation scales: *(see Annex 4 ch 2.5, Table 3)***

The presented scales are all downloadable from the internet, along with their mode of use (see bibliography). Give priority to the DESS (Pain San Salvadour Child), EDAAP (Expression of Pain in Children or Adults with a polyhandicap), or PPP (Profile Pain Pain) scales, which are specific for polyhandicap. The DESS and PPP require a basic file for each child or adult.

It is necessary that all staff be trained in their use if they are to be used routinely and that a basic "pain" record be created by the team with the aid of the parents. Under these conditions, less than 5 minutes are sufficient to complete a DESS or PPP once the basic file has been compiled.

- **Evaluation by physiological parameters**

Heart rate variability (HRV) correlates with autonomic nervous system tone and has been used to evaluate nociceptive stimuli. The PhysioDoloris™ (MDoloris Medical Systems SAS, Lille, France) is a monitor that has been developed to convert HRV into a numerical scale (Analgesia Nociception Index or ANI) using an algorithm. This tool has been studied using various surgical stimuli under general anaesthesia in adults. In patients who are awake, there is a negative linear relationship between the ANI and pain scores. Its applicability to children with cerebral palsy, especially non-communicative children, is the subject of a pilot study (Avez Couturier 2015).

Other neurophysiological assessment methods are also still under research.

2.5.3 Aetiologies of pain

The etiological diagnosis is often difficult, especially for chronic pain, because of the intricate nature of the pathologies presented by these children or adults, leading to a vicious circle. Nonetheless, it is necessary to focus on recognising the initial causal pathology to break it.

Pain related to the polyhandicap itself: nociceptive pain as a consequence of the polyhandicap

The most frequent types of pain are musculoskeletal pain and pain of digestive origin (80.7% of pain for Anne HUNT 2004):

- Neuro-orthopaedic disorders: pain from hip dislocation, pain related to retraction, immobility - cramps, paresthesia, pressure points -, and osteoporosis, which can lead to fractures and sometimes a late diagnosis
- Pain of digestive origin: constipation, gastroesophageal reflux, gastric dilatation, etc.
- Neurological pain: spasticity, major dystonia, intracranial hypertension
- Special attention should be paid to the discomfort and pain associated with swallowing problems
- Pain secondary to self-mutilation
- Bone pain secondary to spontaneous fractures or demineralization.

The possibility of pain of stomatological or dental, ENT, ophthalmological, cutaneous, or urinary origin (acute urine retention, lithiasis, sometimes of iatrogenic origin), or in connection with chronic underhydration must be considered.

- **Neuropathic pain**

Neuropathic pain must always be consdiered. It is often sought in progressive encephalopathies because it is part of their own symptomatology, but it is far from being absent from the signs of so-called fixed encephalopathies. The characterization of such pain is very important because its treatment is specific.

- **Psychological pain and suffering**

Psychological pain must always be considered. Psychological support, working with families and caregivers, is of utmost importance.

- **Pain related to care**

Pain related to care is essentially that related to daily life: washing, dressing, changing clothes, transfers, mobilization, positioning, installation of equipment, feeding, and oral care. Its treatment is above all preventive, forcing us to continually rethink our habits.

- Pain related to nursing procedures (gastrostomy care, aspiration, changing dressings, post-operative care, specimen collection) must also be evaluated and prevented (EMLA, ENTONOX/MEOPA).
- Rehabilitative care, including physiotherapy, should use the least aggressive methods possible and may involve the early prescription of analgesics.

2.5.4 Therapeutic aspects

If the cause is not found, it is necessary to prevent or alleviate the pain by means of medication (symptomatic treatment), always trying to find a balance between relieving the pain and maintaining the cognitive and social capacities of the person being treated.

Such analgesic treatment must be used in sufficient doses using medication that is adapted to the intensity and type of pain. The rules of prescription - compliance with the three levels of the WHO and the recommendations of the AFSSAPS (for children, 2009) - apply to the polyhandicapped population.

- **The management of the vast majority of pain is based first and foremost on prevention.**

Treatment must, above all, aim to avoid the emergence of potentially painful pathologies and aim to constantly improve the physical and psychological comfort of the child or adult and his or her well-being and quality of life.

The practices of care providers, educators, and physiotherapists have an essential preventive role: feeding techniques, transfers, positioning and posture, and personal hygiene (importance of protocols). It is important to stress the major role of speech: singing a song, broadcasting music, talking, telling a story, playing with a puppet, and manipulating objects, as well as relaxation.

When pain is present, it is essential to focus on recognizing the initial causal pathology to best adapt treatment.

- **Particularities to be considered for drug treatment:**

- Beware of drug interactions in a polyhandicapped patient
- Opioids: respiratory congestion is not a formal contraindication, provided that the prescription rules are strictly observed
- Digestive side effects: constipation, leading to reinforcement of hygienic, dietary, physical, and medication measures
- Routes of administration: the oral route is always preferred to the rectal or subcutaneous route; the IM route should be avoided
- The side effects of long-term treatment are not always well known, so treatment should be re-evaluated regularly
- Treatment of nociceptive pain.

- **Pain related to care:**

Reliance is placed on the analgesic treatments described in the three WHO tiers *(see Annex 4, ch 2.5, Table 4: Main analgesic drugs classified according to the three WHO tiers)*:

- Tier I: Non-opioid analgesics, known as peripheral analgesics, intended for mild pain: paracetamol and non-steroidal anti-inflammatory drugs (NSAIDs). NSAIDs should be used with caution for children with a polyhandicap because of the frequency of gastroesophageal reflux; their prescription should be combined with that of PPIs. Aspirin should no longer be used by children because of potentially serious side effects and its limited use in certain childhood viruses

- Tier II: Weak central opioid analgesics, reserved for moderate to severe pain: mainly codeine (children over 12 years old - HAS 2016) and Tramadol
- Tier III: Strong central opioid analgesics for severe pain: morphine and fentanyl, still underused in *a fortiori* polyhandicapped children: following the development of dependence, at the end of life, etc.

- **Treatment of neuropathic pain:**

- Gabapentin in the first line (10 to 30 mg/kg/day) or an amitriptyline-type antidepressant (0.3 to 1 mg/kg/day in one evening dose (1 drop/kg/day for the drinkable forms)
- The use of opioids is only indicated in cases of mixed pain.

- **Treatment of pain of psychological origin**

Pain of psychological origin is often a diagnosis of elimination that requires a psychiatric opinion.

- **Other drug treatments**

- Antispasmodics (phloroglucinol), muscle relaxants (benzodiazepines, baclofen)
- Etiological treatments: dystonia, gastroesophageal reflux disease (GERD), osteoporosis, spasticity (see specific chapters)

- **Drug treatments used for prevention :**

- Local anaesthetics: EMLA cream or patch for care, VERSATIS patch 5% in case of neuropathic pain.
- MEOPA: It is still little used, although there are few contraindications or side effects. It is rapidly anxiolytic (1 min) and provides rapid (4 min) surface analgesia.

Pain is a major problem for polyhandicapped individuals. Observation is crucial, and the use of a hetero-evaluation scale is essential. It is essential to find the cause of the pain, but if not possible, a test treatment is perfectly legal.

Treatment is mainly preventive and symptomatic treatment is initially non-medication based. Drug treatment must be adapted to the type of pain and prescribed in effective doses.

"Any change in the clinical, emotional, or social behaviour of a polyhandicapped child, provided that it can be observed in several stages and by several people, is highly suspicious of indicating pain, until proven otherwise". MR. MENIER

2.6 Dental disorders

The risks involved in poor oral health are many:

- Pain: the oro-facial sphere is the most highly innervated area of the body
- Poor hygiene => cavities, periodontitis, loss of teeth, bad breath
- Infection, with possible complications (cardiac, ENT, pulmonary, etc.)
- Difficulty chewing and swallowing.

It is essential to act almost from birth to avoid the later use of devices, which is always difficult for a polyhandicapped person.

2.6.1 Consequences of a polyhandicap on oro-facial functions

- **The jaw**

Good functioning of the jaws allows proper dental occlusion and efficient chewing (controlled extractions may sometimes be necessary).
Oral parafunctions (bruxism, lack of lip closure, repeated movements, tongue position) cause pain in the temporo-maxillary joint that is difficult to diagnose and that can be prominent because it is triggered by the slightest movement. Such pain increases the difficulty of chewing.

- **Food or salivary stasis**

If chewing is not sufficient, food remains stuck in the vestibules, greatly increasing the risk of caries. For the same reasons, tartar is highly present on the teeth of polyhandicapped individuals.

The presence of acid in the mouth during gastroesophageal reflux disease (GERD) weakens the enamel and supporting bone tissue, causing cavities and gingivitis.

Saliva is a major element of protection. It prevents the formation of caries and remineralises the enamel. Saliva can be missing in cases of hyposalivation (often linked to medication), whereas the child/adult drools because he does not swallow his saliva correctly in cases of hypersalivation, in which case it does not play its optimal protective role.

- **General consequences of dental diseases**

Many pathologies are aggravated or induced by an altered oral condition. The most often cited are endocarditis linked to a dental infection and ENT pathologies linked to local or loco-regional contamination, as well as rheumatic pathologies linked to poor dental occlusion. Diabetes is an additional risk factor that potentiates all the pathologies described below.

2.6.2 Clinical Aspects and Treatment

Refusal to eat must result in a careful examination of the mouth!

- Cavities and infections of supporting tissues

The bacteria involved are numerous and the infections are transmissible (mother to child). They must be treated, just like periodontal infections.

- Gingival pathologies

They are very frequent and often related to treatment (antiepileptics). The volume of the gums is such that it can affect chewing and be detrimental to tooth eruption and especially hygiene. The accumulation of plaque increases the risk of caries and aggravates the long-term consequences.

- Pathologies of the mouth
- Candidiasis, a superficial mycosis, can be the result of antibiotic treatment or nutritional deficiencies.

Any intraoral lesion must be treated, as it causes pain. The causes are local infection, difficult tooth extraction, bites during epileptic seizures, or simply fragility of the oral mucosa.

If recovery is slow, early (adult) cancer may need to be investigated.

2.6.3 Preventive recommendations

- **Prevention of caries and infections**

From the first teeth, place a compress on the gums to remove food deposits.
From the age of two years, brush once and then twice a day with a small brush and a small amount of toothpaste dosed according to age.

Of course, the diet must avoid sugar or acidic foods (ketchup, vinegar, fruit juice) or combine them with compensators (cheese, proteins, lipids). Always finish the meal with water and, at the very least, apply a compress at the end of the meal if it is not possible to brush.

In collaboration with the dental surgeon, adjust topical fluoride dosages according to the identified risk of cavities.

- **Role of the speech therapist**

Speech therapists work with the polyhandicapped person to learn proper swallowing, mouth closure to avoid drooling, and oro-facial praxis to promote chewing. The management of oral parafunctions (bruxism) also falls within the sphere of speech therapists.

- **Role of the dentist and stomatologist**

An annual visit to a dental surgeon should be scheduled (treatment under MEOPA if possible). Controlled extractions allow the permanent teeth to be better positioned.

The presence of a pacemaker must be reported, as it limits the use of certain devices.

The pain caused by parafunctions may warrant a retainer.

2.7 Eating disorders

Eating disorders in people with a polyhandicap can have serious, sometimes vital, consequences related to misdirection, gastroesophageal reflux, risk of undernutrition, and dehydration. These disorders also have consequences on phonation, non-verbal communication, and especially breathing. They limit the pleasure and conviviality of the meal and are not without consequences on socialization.

2.7.1 Swallowing disorders

From birth, most new-borns have a sucking reflex that allows them to suck. Then, as development progresses and through multiple oral explorations, the child builds new motor skills that allow him or her to eat with a spoon and then a fork, chew, and drink from a glass.

- **These functions call for multiple skills:**

- Skills of sensation to perceive and then analyse what is in the mouth
- Motor skills of the tongue, cheeks, lips, jaws, palate, pharynx, and oesophagus
- More global motor skills to maintain a well-adapted posture and sometimes use certain functional capacities.

- **Eating is comprised of three stages:**

- Chewing, voluntary and automated, if it is well mastered, which requires the presence of molars and lateral mobility of the tongue, essential for solid food. It allows the transformation of food into a homogeneous and lubricated food bolus
- Propulsion, also voluntary and automated, which leads the bolus to the pharynx through the elevation and retraction of the tongue
- Swallowing, which is a simple reflex triggered by contact of the sensitive zones of our aerodigestive intersection. It allows the transport of the food bolus into the oesophagus.

Successful swallowing therefore requires the adequate synchronization of these three stages, each conditioning the next (Lespargot, 1989; Rofidal, 2017).

- **Respiratory aspiration**

Swallowing disorders lead to various misalignments, either nasal due to poor contraction of the soft palate during propulsion, or tracheal due to a deficiency of the reflex.

- Direct tracheal respiratory aspiration during eating:
  - Fatal when a piece of food becomes blocked in the trachea and breathing is not possible and resuscitation with a Heimlich manoeuvre is not undertaken immediately
  - Severe when a piece of food that gets stuck in a bronchus causes inhalation pneumonitis, which can be life-threatening
  - Minimal and repetitive, related to the inhalation of liquids, semi-liquids, or mixed food, which may go unnoticed if coughing is minimal or absent; these accidents are the cause of permanent bronchial congestion responsible for severe pain, frequent superinfections, atelectasis, and dilation of the bronchi, which may be complicated by pneumopathies and evolve into a restrictive respiratory syndrome (Gautheron 2015).

- Indirect tracheal respiratory aspiration due to the absence of secondary swallowing of the alimentary bolus or a deficit of salivary swallowing, which can cause inhalation upon the resumption of breathing, or due to inhalation of regurgitated acid (gastroesophageal reflux, vomiting) and therefore more dangerous for the respiratory system.

2.7.2 Prevention of swallowing disorders

- **Meal conditions**

There are three essential conditions for the prevention of swallowing disorders:

- Meals should take place in a quiet environment to allow the person to concentrate on eating
- The positioning device must allow good flexion of the neck (the axis of the head must always be in front of the axis of the trunk, regardless of the position of the body) so as not to oppose the rise of the larynx triggered by the swallowing reflex and guaranteeing closure of the airways; it is most often the inclination of the sitting position, often up to 45 to 60° (interest of the "kit-cool") that allows the person, whether hypotonic or hypertonic, to avoid having to fight against axial gravity and to concentrate his energy on his oral and facial motor skills (Le Métayer, 2003). The hand of the carer helping the person with a polyhandicap to eat should be placed behind the head and never exert pressure on the forehead or under the chin to avoid tilting the head backwards (see Appendix 4, in connection with Chapter 2.7, Indications for Installation and Mouth Closure Aid Practices and Techniques)
- Modification of the texture of the food must be adapted to the oral-facial motor skills.

- **Texture of the food**

For food:

- A blended texture is intended for people whose tongue mobility is only antero-posterior and/or for those who are completely toothless; it must be smooth, it must separate the different components of the dishes, and its presentation must be neat (decoration, colours...)
- A ground texture is intended for people whose tongue is capable of lateral movement but whose chewing vigour is weak and/or the number of chews is insufficient or who do not have a sufficient chewing coefficient (molars in opposition); all foods crushed in the plate are served as they are and are well cooked, whereas the others are processed in a mixer until a texture is obtained that retains a certain subsistence but is well cooked (Rofidal, 2011) (Benigni, 2011, 2014)
- The use of a masticating mixer is very useful, allowing you to easily adapt the texture of a dish according to your needs (in institutions, with your family, in restaurants, etc.).

For beverages:

- The sensory information of the drink can be improved by the taste (limiting sweetened drinks to preserve the teeth), temperature, fizziness (carbonated water), and possibly spiciness (ginger).
- When it is necessary to slow the time of the sip in the mouth, modification of the textures calls for the use of a thickener (starch, for which carbohydrate intake must be considered) and/or a gelling agent (gelatine or agar).

- **Oral motricity and sensorial regulation**

Mealtimes are facilitated when the position of the subject, the sensory quality of the food, and the environment have been optimized to limit excessive tonic recruitment (Bullinger, 2006). Thus, to help a child nurse, one can help him/her to hold the teat with the lips, ensuring that the teat is well placed on the tongue, so that if forms a trough to allow effective nursing.

For the older child or adult, spoon feeding requires good mouth closure (jaws and lips), lingual motor skills at both the tip and base of the tongue, and sensory abilities that allow the person to perceive that they have food in their mouth and accept it.

When drinking from a cup, postural precautions should be reinforced to limit the risk of misfeeding, bringing the liquid into contact with the child's upper lip to help the child to aspirate. Under no circumstances should the liquid be poured into the child's mouth.
(Annex 4, see link to section 2.7, Indications of installations and practices).

2.7.3 Oral sensory disturbances

Some people with a polyhandicap show hyposensitivity or hypersensitivity in the oral sphere, which concerns tactile, olfactory, gustatory, and thermal sensations.

Hyporeactive people show little or no discrimination of taste nor pleasure or displeasure at mealtimes. Sensorial stimulation can be attempted by offering them larger spoons and tastier and more temperature-diverse foods (cold or hot), without forgetting to offer spices or mustard (spicy, hot), mint (refreshing), or sparkling water (sparkling) to stimulate "trigeminal sensitivity".

Hyperreactive people show food hyperselectivity and frequent food refusal, sometimes accompanied by "headaches" and displeasure at mealtimes. The prevalence of such sensory hyperreactivity is approximately 30% in this population and is correlated with undernutrition (Benigni, 2011). Those concerned must be treated by a specialised speech therapist to broaden their food repertoire. Their diet must be adapted to their tastes and enriched while awaiting the benefits of rehabilitation.

2.7.4 Swallowing assessment and objectives to be achieved

A precise multidisciplinary evaluation is carried out each time the caregiver doubts the comfort of the position or the match between the texture of the food proposed and oral-facial motricity. An assessment outside of mealtime under calm conditions with test foods (e.g. yoghurt, biscuits, fruit juice) is used to guide the observations during the meal.

- **Additional examinations may be carried out**

Additional examinations may be carried out, especially in cases of suspected respiratory aspiration or to support a decision to reduce oral intake in favour of gastrostomy in a patient with swallowing pneumopathies.

- Swallowing fibroscopy is a flexible nasal fibroscope examination under local anaesthesia that allows observation during swallowing of, for example, a spoonful of yoghurt in a seated position. This examination is quick but is not always accepted or tolerated. The presence of the fibroscope may disturb swallowing. It allows the examination of swallowing and paralytic or dystonic components by direct observation, whether the larynx is closed or not.
- Swallowing radio-cinema is the making of a film of the swallowing of a radio-opaque "food bolus". This complementary examination allows identification of primary or secondary respiratory aspiration by viewing the tracheal passage of the contrast product. Radio-cinema allows an analysis of oesophageal motricity by visualizing peristalsis. The texture of the radio-opaque bolus can be liquid or semi-liquid. Its taste is not always appreciated, which is one of the limitations of the examination.

- **It is essentially the clinic aspects that guide the organization of the meal**

- The aim is to achieve three essential objectives:
  - Ensure the safety of the person when feeding
  - Ensure the nutritional and hydration value of meals
  - Make meals moments of pleasure, comfort, and relationship.

Whenever possible, the functional abilities of the polyhandicapped person are used and promoted by technical aids: spoons made of an adapted material with a thickened or bent handle, non-slip mats, plate rims, cups with a nasal notch that keeps the head in flexion, possibly a *feeder to* aid upper limb motricity. But the main level of autonomy that is sought is, above all, that of swallowing to avoid respiratory aspiration.

An evaluation under usual conditions (meal given by the usual caregiver) allows for the observation of practices and recommendations for adjustments: quantity of food in the spoon, rhythm of bites, positioning of the caregiver and the person being fed, etc.

2.7.5 Other mealtime essentials

- **Caregiver comfort**

The person must sit on a height-adjustable stool fitted with castors, a right-handed person should always sit to the right of the polyhandicapped person (and vice versa for a left-handed person) so as to always keep the head of the person being fed bent in relation to the trunk, in particular by the position of the spoon, and to facilitate interactions and encourage the person to direct his or her gaze towards the spoon.

- **Facilitation of food and drink intake and oral motor skills**

Access to the mouth is made easier by tilting the entire sitting position backwards, by helping to close the mouth if necessary, and above all by the precise, non-hesitant insertion of the spoon into the mouth in the axis and removal without touching the teeth, but if necessary moving it towards the upper lip if the grip is poor, without straightening the person's head *(see Annex 4, section 2.7, Techniques for helping to close the mouth).*

- **Quality of all aspects of the meal**

The caregivers' know-how must ensure pleasure, comfort, and safety during the meal. This often means abandoning one's own representations of conviviality: sitting in an upright sitting position, offering the food in pieces, the meal shared by all at the same time around a table. Cohesion of the team is essential to allow pleasure, comfort, and safety and sometimes to achieve a certain level of progress for the polyhandicapped child or adult.

- **Card summarizing the main rules of mealtimes for each person**

Inclination of the seating arrangement, head control, food texture, drink texture, rituals at the beginning and end of the meal, technical aids, position in the room, rhythm of the meal, etc. This sheet will be prepared in the most practical form and regularly updated. A placemat or personalised communication tool made up of pictograms can be proposed to encourage communication during meals (I don't want it anymore, I'm thirsty, it's cold, etc.).

2.7.6 Reeducative and rehabilitative care

This care is multidisciplinary:

- Occupational therapists and physiotherapists intervene, in particular, by working on motor skills to promote the state of relaxation necessary to improve the person's autonomy and oral-facial possibilities; optimisation of the position is an essential prerequisite for the oral-facial approach (Le Métayer M. 1985)
- The speech therapist intervenes to improve oral motor skills (lip occlusion, the reduction of tongue protrusion, learning to suck up liquids), promote nasal breathing, limit oral sensory disturbances, train salivary swallowing, and practice chewing.

Each technique is explained to the caregiver to be used progressively during the meal and thus perpetuated. Each of the caregivers at mealtime must perceive the risks incurred by the person they are accompanying and use the appropriate procedures.

2.7.7 Salivary incontinence

Salivary incontinence (drooling) is only rarely associated with hypersalivation, although some medications may contribute to it, as may poor oral hygiene, gastroesophageal reflux, or poor oral trophic status. It is mainly due to swallowing problems, which are themselves linked to sensory and motor disturbances in the area of the mouth, sometimes aggravated by dental, and orthodontic problems.

Saliva secretion is constant and increases during contact (food or non-food) with the mucous membrane of the mouth and during irritation of the lower oesophagus (gastroesophageal reflux). The consequences of

salivary incontinence are many: dry mouth, digestive and transit problems, skin problems, wet clothing, or social rejection. A sponge bandana or pretty scarves are preferable to wearing infant bibs; sponge bracelets can also help people who have the motor skills to wipe their mouth regularly.

Facilitating salivary swallowing involves the same steps as those proposed to facilitate feeding: ensuring the proper positioning of the head in relation to the trunk, helping to close the mouth, encouraging nasal breathing, and proposing speech therapy (Bobath oral treatment). Sometimes, bending the neck accentuates drooling, but it limits the risk of saliva being inhaled and the pneumopathies that can result.

Medical treatment is mainly indicated for the inhalation of saliva. Scopolamine patches are intended to reduce saliva production. Placed behind the ear, they have an effect of 6 to 12 hours after application and for three days (72 hours). However, they are not without side effects, the main ones being drowsiness, thickening of bronchial secretions, constipation, and urinary retention.

Bilateral injections of botulinum toxin into the parotid and submaxillary glands can be carried out under ultrasound control and with analgesic measures. Their duration of action is 3 to 6 months.

Surgical treatment, consisting of the derivation of the main salivary glands, is a major operation.

2.8 Gastroesophageal reflux, esophagitis, gastritis, delayed gastric evacuation

2.8.1 Gastroesophageal reflux disease (GERD)

In polyhandicapped people, GERD is almost always pathological (complicated or responsible for embarrassing manifestations). It is extremely frequent, from 14 to 75% of cases (Vernon, 2013), particularly in cases of mitochondrial pathology (Kim, 2017). It has also been reported in 50% of a population of institutionalised polyhandicapped patients (Böhmer, 2000). It is often severe and causes pain and discomfort but can very often be improved by medical or surgical treatment.

- **Mechanisms of reflux in the polyhandicapped person**

- Inappropriate relaxation of the lower oesophageal sphincter (LES)
- Decreased oesophageal clearance
- Oesophageal dysmotricity and decreased oesophageal peristalsis
- Delayed gastric evacuation with chronic gastric dilatation
- Intra-abdominal hyperpressure: constipation, muscle spasms, scoliosis, coughing, abdominal compression with clothing or other means
- Decubitus with a decreased effect of gravity for oesophageal clearance
- Role of certain drugs (benzodiazepines, etc.).

- **Clinical signs**

The diagnosis of GERD is essentially clinical:

- Nausea, regurgitation; frequent voluntary post-prandial or nocturnal vomiting
- Eating difficulties, prolonged anorexia, refusal to eat, worsening swallowing problems
- Painful manifestations: nocturnal screams, development or aggravation of behavioural disorders, increased spasticity, irritability, dystonic movements (including Sandifer's Syndrome, a paroxysmal dystonia-like movement disorder occurring in association with GERD and, in some cases, hiatal hernia), aggravation of epilepsy
- Repeated ENT infections: pharyngitis, laryngitis, ear infections, etc.
- Hypersalivation, hands in the mouth.

- **Complications**

- Peptic esophagitis: mainly ulcerations of the lower oesophagus, resulting in bloody vomiting, hematemesis, or iron deficiency anaemia, painful dysphagia, and anorexia. It can lead to the formation of oesophageal stenosis or endobrachyoesophagus.

- Respiratory: repeated indirect inhalation pneumopathies, which can be very serious (Mendelson's syndrome by massive inhalation); laryngospasm, cough, apnea.
- Nutritional: weight loss, undernutrition (Benigni et al, 2011), and growth retardation as a consequence of dietary difficulties.

- **Various complementary examinations**

- **pH-metry**: the only useful baseline test for diagnosing persistent GERD, assessing the effectiveness of antisecretory therapy, or correlating symptoms, such as coughing with episodes of reflux. The severity of acid reflux does not correlate with the severity of symptoms or complications.
- **Impedancemetry**: coupled with pH-metry, it has the particularity of detecting acid and non-acid reflux (because potentially buffered by effective PPI treatment), liquid, solid, and gaseous reflux and correlates clinical signs with episodes of acid or non-acid reflux.
- **Oeso-gastro-duodenal** f**ibroscopy** (nowadays OGD "endoscopy", because it is no longer fibre optic): it confirms the diagnosis of oesophagitis, allows the study of the cardia, and identifies hiatal hernias. Biopsies allow the diagnosis of peptic, mycotic, or eosinophilic esophagitis or *Helicobacter pylori* gastritis.
- **Oeso-gastro-duodenal transit (OGDT):** carried out by the upper route or by gastrostomy; it is of little use for the diagnosis of reflux because it is neither very sensitive (46%) nor very specific (41% Saleh, 2015) and is responsible for false positives (possibility of triggering reflux depending on the patient's position). It provides information on morphological abnormalities: hiatal hernia, oesophageal stenosis. It is required by the surgeon before an operation.
- **Manometry**: useless in the diagnosis of GERD. It seeks to assess the dysmotricity of the oesophagus, the tone of the lower sphincter of the oesophagus, and the quality of peristalsis. It is useful in pre-operative care, as major disorders of oesophageal motility can make Nissen's results more uncertain.
- **Scintigraphy**: not recommended in the diagnosis of GERD. It can be very useful in the diagnosis of delayed gastric evacuation or bronchial inhalation.
- **Oesophageal ultrasound**: Non-contributing to the diagnosis of GERD.
- ***Helicobacter Pylori* serology*:*** in case of repeated unexplained hematemesis.

- **Indication of additional tests:**

Because of the frequency and severity of GERD in people with a polyhandicap, consider PPI therapy before further testing:

- <u>In adults</u>, it may be permissible to initiate PPI therapy (usual dosage) without prior investigation if GERD is clinically very likely. If the symptoms of reflux are improved, this treatment can be continued in the long term (AFSSAPS Recommendation - June 2008) and no further investigation is required. If there is no improvement, or if there are immediate signs of complications, fibroscopy should be performed to confirm the existence of esophagitis and adapt the dosage of the PPIs. pH measurement may be useful to assess the effectiveness of antisecretory therapy. Manometry and OGDT should be reserved for pre-operative use
- <u>In children</u>, Espghan (2017) recommends the use of objective tests for the diagnosis of GERD (pH-metry coupled with impedancemetry and/or upper GI endoscopy) in children with neurological impairment. However, given the high prevalence of GERD, a trial of PPI therapy with careful clinical follow-up is acceptable for these clinically fragile children.

- **Hygieno-dietary rules and prevention**

- Sitting for at least one hour after eating and avoiding tight clothing or straps.
- Prolonged bed rest at 30° (Rofidal, 2011) (bed rest should be as short as possible).
- Avoid acidic foods while waiting for the effect of PPIs and adapt the texture of the food to the chewing and swallowing capacity.
- If possible, recommend small, split meals.
- Fight constipation and spasticity, prevent spinal deformities, manage undernutrition. Practice respiratory physiotherapy before meals to avoid coughing after meals.
- Consider reflux medications: benzodiazepines, N acetyl cysteine, etc.

- **Drug treatment**

- Antacids and mucosal protectors are increasingly being considered in terms of their effectiveness and usefulness.
- Anti-H2: rapid action but important side effects and possible exhaustion of the effects with time. They have given way to PPIs.
- PPIs have no effect on reflux itself. They are superior to anti-H2 in healing esophagitis. They can have side effects in 14% of cases, dominated by nausea, diarrhoea, or constipation. Their long-term deleterious effects are not known. As already mentioned, a trial treatment with PPIs is perfectly legal. A study in polyhandicapped children with fibroscopically-proven esophagitis showed that 90% were asymptomatic after three months of treatment (Böhmer 1998).

- **Surgical treatment:**

The most frequently used intervention is fundoplicature (Nissen's intervention). This intervention is proposed in cases of failure of medical treatment, long-term dependence, or very severe and disabling GERD, especially with recurrent respiratory complications. It is performed only for documented and proven reflux. Resolution of vomiting can completely change the management of these children and their contact with their families and caregivers. The rate of reflux recurrence (reascension of the valve in the thorax) following the procedure is high: 12.3% versus 1.8% in children without neurological disorders, as reported by Podevin in 2006, with 44% re-interventions, and 12 to 30%, as reported by Vernon-Roberts (2013), with re-interventions in 12.6% of cases, including three fourths by Bianchi's intervention (Lauriti 2018). Recurrences after this type of intervention are much less frequent: 1.4% ± 1.1% (Lauriti 2018). Per and postoperative mortality is difficult to assess: from 3% to 9% according to surveys.

2.8.2 Delayed gastric evacuation

- **Physiopathology**

Gastric emptying is normally complete in adults in four hours, but takes more than 6 to 10 hours for those with a polyhandicap (objectively measured by measuring gastric residue or gastric scintigraphy): feeding occurs on a stomach still full from the previous meal.

Delayed gastric emptying leads to chronic gastric dilatation. It is associated with and aggravates GERD (28-50% of cases) and can lead to treatment failure.

- **The diagnosis is essentially clinical:**

- Abdominal bulge ++
- Tension of the abdominal wall
- Percussion tympany
- Discomfort, nausea.

X-rays show an increase in the shadow of the stomach extending from one hypochondrium to the other, horizontalization of the entire body of the stomach, and the absence of gas in the intestines.

- **Treatment of delayed gastric emptying**

In everyday life, gastric emptying can be improved by alternate positioning (Swendsen 2007) if spinal deformities allow it:

- Empty the stomach before eating by placing the person on his or her left side for a few minutes
- Give the meal in a corset-seat in a sitting position
- At least one hour later, put the subject on his right side to promote evacuation of the food content.

Prokinetic drugs may help in adults, but the risk/benefit ratio should always be assessed. If the person has a gastrostomy button: open the button regularly.

Pyloroplasty remains reserved for severe cases.

2.9 Transit disorders: constipation in the polyhandicapped person

Constipation is a decrease in the volume and frequency of stools (calendar) and a change in their consistency (Bristol scale). Rare bowel movements are not synonymous with constipation, but one must know one's usual rhythm. Any hard or loose stool (stasis diarrhoea) may indicate constipation.

The pathophysiological approach theoretically distinguishes between transit constipation and terminal constipation; in both cases there is a risk of faecal impaction, but the two phenomena are frequently associated in people with a polyhandicap.

Monitoring of stool output should be ongoing and recorded and the appearance of the stools should be noted.

2.9.1 Clinical aspects

- **Factors favouring normal transit**

For everyone, bowel movement and defecation are promoted by:

- Natural intestinal peristalsis that is effective in moving stools forward
- Stools that are sufficiently large and flexible to move forward under the effect of intestinal peristalsis
- Standing up to allow intestinal kneading by the movements of the diaphragm with each breath
- Walking, because the dissociation of the thoracic girdles (synchronous reverse rotation of the pelvic and shoulder girdle) mechanically stimulates the intestines
- Attempting to pass the stool as soon as the need is felt, without waiting and allowing the sensation of need to fade away
- The correct position for defecation, the best position being squatting or positions that approach it.

In the vast majority of cases, these factors cannot be met by polyhandicapped individuals.

- **Risk factors for constipation in the polyhandicapped individual**

There are many risk factors for constipation for people with a polyhandicap:

- Neurovegetative disorders with poor or slowed natural intestinal peristalsis
- Failure to stand, walk, or move, instead remaining in a predominantly fixed sitting position
- Diet insufficiently rich in fibre
- Insufficient hydration and lack of autonomy in fluid intake, which further increases dehydration of the faecal bolus already caused by to its slow progression through the colon
- Insufficient chewing and food texture sometimes unsuitable for chewing capacity
- Inability to express the need or difficulty in being able to satisfy it when it is felt, blunting of the feeling of need
- Difficult installation and stability on the toilet, postural conditions that do not facilitate defecation
- Intimacy insufficiently respected during defecation attempts, intermittent stays in a collective setting that can lead to temporary voluntary retention
- Orthopaedic or axial dystonic disorders or those of the hip, telescoping of the trunk associated with a scoliotic or kyphotic spine, with pneumo-digestive impaction
- Axial hypotonia and decreased thrust force
- Dyskinesia
- Sphincter spasticity, recto-anal dyssynergia
- Medications (anticholinergics, neuroleptics, antiepileptics, antispastics)
- Endocrine disorders (hypothyroidism), possible metabolic disorders (hypokalaemia).

As a result, constipation is very common among people with a polyhandicap. Moreover, X-rays very often show diffuse "stercoral stasis". It can be a major source of permanent discomfort (abdominal material and/or gas in the abdomen and distension), breathing difficulties, or pain, as well as an irritation that can increase

spasticity and even cause seizures in people with epilepsy. It may also promote gastroesophageal reflux by intra-abdominal hyperpressure. The emission of a stool does not mean that there is not stasis of material throughout the colon. Neither does diarrhoea, because the intestine can react to stasis by secreting water, causing terminal constipation to be expressed by such diarrhoea.

Terminal constipation can also be complicated by an occlusive syndrome. If the faeces are dehydrated, their stasis in the terminal part of the intestine exposes them to the risk of faecaloma, a hard aggregate that can no longer leave the sigmoid or pass through the anal sphincter.

One should try to distinguish between transit constipation and terminal constipation. However, it is not uncommon for the two phenomena to be associated. Only a digital rectal exam can make the differential diagnosis.

2.9.2 Therapeutic Resources

- **In all cases:**

Whether it is transit constipation or terminal constipation or a combination of both:

- Limitation of medicines that promote constipation *(Annex 4, see Chapter 3.5, "Iatrogenicity")*
- A daily diet rich in vegetable fibre (fruit, vegetables, legumes, wholegrain cereals, dried fruit)
- Adequate hydration: for children, aim for 100 ml/kg/day up to 1.5 litres, and for adults, 1.5 litres of water intake per day, all intake combined (including part of Hépar*); therefore hydrate systematically between meals as well. Note on the label of the bottle the name of the person and the date to check the day's consumption. Any drink can be gelled or thickened in case of difficulty swallowing liquids
- Verticalization (++) and daily free movement or physical activity, regardless of the type (on the ground on a floor mat or with a walking aid, such as a walking frame), according to the psychomotor level and motor capacities of the person; at least, practice regular changes of position
- Passive abdominal mobilization (mobilization of the hips); if the state of the hips allows it, circumduction of the lower limbs in the dorsal decubitus position, hips and knees bent to the maximum, provided by a caregiver, in a clockwise direction, outside the post-prandial periods
- Gentle abdominal massage to aid intestinal transit, given daily, performed by trained family caregivers or professional caregivers, clockwise, with the palm of the hand or with the shower, for example during morning personal hygiene
- Mild enemas; hypertonic enemas (Normacol) in case of failure
- Long-term transanal (Peristeen) irrigation, on gastroenterological advice.

- **If it is transit constipation:**

- Long-term laxative treatment: osmotic laxatives (if the person is not dehydrated), non-lactulose-containing laxatives (Macrogol-Polyethylene glycol -PEG-), or sweet laxatives (lactulose, which may cause abdominal pain).
- Ballast laxatives and lubricant laxatives are formally contraindicated for those with swallowing disorders, but care should also be taken with PEGs (severe pneumopathies described in children).

- **If it is terminal constipation:**

- Attempt to pass the stool as soon as the need is expressed, if it can be expressed; otherwise, systematic (persevering over a long period) presentation within 30 minutes postprandial to benefit from the gastrocolic reflex
- Ergonomic, psychological, and functional consideration of the defecation posture: stable, comfortable sitting position, if possible - depending on the neuro-orthopaedic status of the person – a posture approaching a squatting position (flexion of the hips greater than 100°, if necessary by using a step); visual, auditory, and olfactory privacy
- Possible rectal laxatives: Eductyl suppository as a trigger aid, or Microlax, occasionally.
- PEGs can also be useful in this case.

In conclusion, constipation in a person with a polyhandicap is a major scourge that requires rigorous management.

2.10 Nutritional disorders

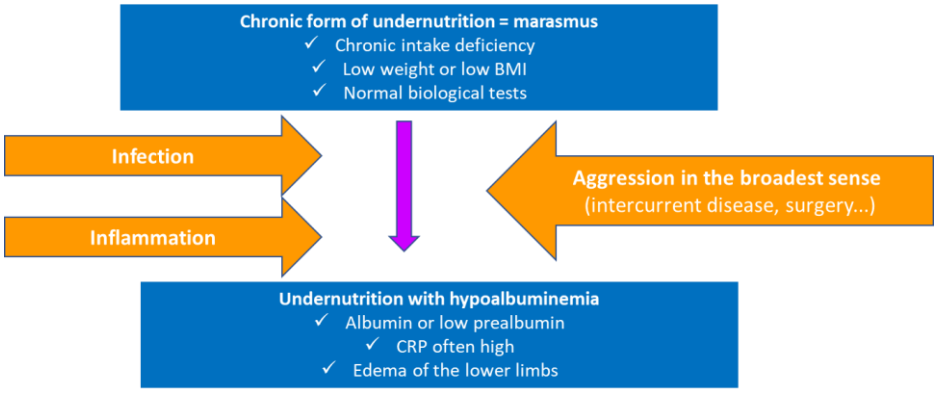
A study published in the Annals of Rehabilitation and Physical Medicine showed that nutritional status was the variable that most influenced general health (De Lattre, Hodgkingson, Bérard, 2007).

The ESPGHAN (European Paediatric Society of Gastroenterology Hepatology and Nutrition, 2018) working group suggests that nutritional assessment and monitoring should be carried out by a multidisciplinary team ideally including a physician, dietician, nurse, speech therapist, physiotherapist, psychologist, and occupational therapist.

2.10.1 Origins of undernutrition in people with a polyhandicap

- **Frequency and origins**

Undernutrition is common among people with a polyhandicap, with a 66% prevalence in children (Campanozzi, 2007), and a 60% prevalence in adults (Benigni, 2011). It should be systematically screened. Having a polyhandicap is inherently a risk factor for undernutrition: dependency on being fed, difficulties in expressing food cravings and freedom of choice, possible poor oral conditions, chewing and swallowing disorders, sensory disturbances in oral function, food fatigue, gastroesophageal reflux with possible esophagitis, slowed gastric emptying (gastric paresis), slowed transit and expulsion disorders, and high energy expenditure (muscle tone disorders, abnormal movements, vegetative disorders). The risk of undernutrition is increased by special circumstances: pain, hyperthermia, infection, bedsores, intercurrent disease, and surgery. The most frequent form of malnutrition in this population is marasmus, which can be complicated by hypoalbuminemic undernutrition.



- **Assessment of nutritional status**

This assessment is based on anthropometric measurements:

- Monthly weight measurement
- Height measurement: if height cannot be measured with a tape measure, segment the body from bony landmarks with a tape measure, then add these measurements together; take two measurements and average them.
- Assessment of waist size if waist size cannot be measured
- Brachial circumference measurement (at the midpoint of the non-dominant arm)
- Measurement of the tricipital skin fold

For children, the ESPGHAN recommends the use of an extrapolation equation based on the tibial length (between the outer joint space and the distal end of the lateral malleolus).

Boy: Height = 40.54 + (2.22 x tibial length)
Girl: Size = 43.21 + (2.15 x tibial length)

For adults, the Chumlea extrapolation formula can be used from the heel/knee distance if growth has been normal.

Male height cm = (2.02 x TG) - (0.04 x age) + 64.10
Female size cm = (1.83 x TG) - (0.24 x age) + 84.88

The measurements of weight and height allow calculation of the BMI (BMI = weight in kg / height² in m); this calculation is only valid for adults because the BMI is not a reliable criterion for polyhandicapped children. For children, it is better to follow the evolution of their height.

The ESPGHAN also recommends an annual bioassay of micronutrients (vitamin D, iron, calcium, phosphate, etc.) in the nutritional assessment of children with neurological disabilities.

Albumin, prealbumin, and CRP measurements should be reserved for unstable or decompensated patients. They remain normal and falsely reassuring in the event of marasmus, which is the most common type of undernutrition of people with a polyhandicap.

2.10.2 Criteria for undernutrition

- **In children (ESPGHAN criteria)**

- Stagnation in weight and/or slowing of statural growth
- Weight for age Z-score < -2 SD
- Thickness of the tricipital skin fold < 10th percentile for age and sex
- Brachial circumference < 10th percentile.

- **In adults**

The presence of only one of these criteria suggests a risk of undernutrition (practical information sheet from the Association Ressources Polyhandicap Hauts-de-France, available on the association's website):

- BMI < 16 Kg/m^2 in adults
- Loss of 3 kg if BMI < 16 or 5 kg if BMI > 16
- Slimming of 5% relative to the reference weight
- Brachial circumference < 22 cm if BMI < 16, or < 25 cm if BMI > 16 (1 cm = 2 kg)
- Reduction of ingesta by half relative to the usual ration
- Weight < 40 kg (Benigni et al, 2011)
- Albumin < 35 g/l
- Pre-albumin < 0.18 g/L.

However, a normal or high BMI or a normal biological assessment does not exclude undernutrition.

2.10.3 Consequences and therapeutic aspects

Undernutrition exposes everyone to a vicious circle: the main complications are infections, muscle wasting, osteoporosis, and trophic disorders (bedsores), which cause pain; all these disorders increase energy expenditure and worsen undernutrition.

A two-pronged nutritional assistance strategy is needed:

- **Enrich feeding by concentrating enrichment in a small volume**

(Benigni, 2019, practical sheet of the Association Ressources Polyhandicap Hauts-de-France, available on the website)

- Add protein: unsweetened condensed milk, powdered milk, formula, cheese, béchamel sauce, eggs, ham, tuna flakes, protein powder, etc.
- Add lipids: butter, oil (varying the oils), fresh cream, chocolate spread, peanut butter, almond purée, etc.
- Add carbohydrates: starchy foods, biscuits, cereals, porridge, pastries, breadcrumbs, salted crumble, almond powder, hazelnut powder, maltose dextrin, etc.

- **Favour calorie and protein intake over a balanced diet:**

- Respect the person's preferences
- Arrange the duration and rhythm of food intake to avoid prolonged fasting (possibly split the diet by adding snacks)
- Provide meal assistance/be flexible, do not conflict at mealtime
- Give drinks of nutritional interest (milk, fruit juice), preferably outside of meals
- Make sure you are providing the correct amount of food.

- **Use oral nutritional supplements (ONS)**

Nutritional supplements are of nutritional and practical interest. It is permissible to use home supplementation whenever possible to avoid fatigue and for economic reasons. As the name implies, ONSs are "supplements" to meals and should not replace them. They should be offered preferably at snack time and in the evening so as not to reduce the day's intake.

2.10.4 Enteral feeding

Enteral feeding is a nutrition technique that includes all the digestive feeding processes but bypasses the oral route. It allows the temporary or definitive intake of nutrition, liquids, medication by means of a probe or a button when *per os* feeding is not or no longer possible: swallowing problems, refusal of food, regardless of the origin, or because of persistent undernutrition despite the nutritional aid strategy implemented.

- **Routes**

- Naso-gastric tube: this route is only considered in the event of an acute situation (eating disorders, post-operative care, or intercurrent disease, especially pulmonary) and can only be a temporary solution before returning to eating by mouth or waiting for a gastrostomy (if the frequent complications, which can be serious, dissuade you from continuing with the oral route).
- Gastrostomy:
  - Percutaneous endoscopic (GPE) or the radiological route for some teams: the reference method when possible
  - Surgical procedure, if GPE is not possible or if anti-reflux is associated.

- **Indications for enteral nutrition**

- Swallowing disorders: repeated pulmonary aspiration, repeated pneumonia, discomfort ++.
- Eating disorders: refusal to eat, regardless of the cause.
- Undernutrition: often the consequence of the first two (knowing that one should not wait for its occurrence to resort to enteral nutrition).
- Gastric decompression (severe disorders of gastric emptying with gastric distension).

The ESPGHAN recommends the use of enteral feeding when the time required for oral feeding exceeds three hours per day (Romano 2017) or if the person only takes ONSs (Rofidal 2018).

It is necessary to always weigh the indications carefully with the family and the teams: explain the reasons and inform them about the benefits and risks. Stopping oral feeding, even partially, is a very difficult step to accept by the family and teams because of a strong feeling of personal failure. Informed consent of the family must be obtained.

It should always be remembered that GPE can be a temporary solution and does not prevent pleasure-feeding through the mouth if it is possible. GPE should never be a medically convenient solution but should also not be applied too late (lung damage, suffering of the child, etc.).

- **Percutaneous endoscopic gastrostomy (PEG):**

This is currently the most widely used route.

- Contraindications (Heusckel 2015): absolute: coagulation disorders, organ interposition (absence of transillumination), peritonitis; relative: subcostal stomach position, portal hypertension, ascites, renal failure in dialysis.
- Non-contraindications: anterior abdominal surgery, ventriculoperitoneal shunt.

- Techniques: Operating theatre for surgery under GA - short duration without opening the abdominal wall; average length of hospitalisation from 3 to 5 days.

Two-stage GPE is decreasingly performed: first, a gastrostomy tube is inserted and replaced 2 to 3 months later after the stomach is attached to the abdominal wall by a button during a new endoscopy under GA.

One-step GPE: a button is put in place in the first intention. The stomach wall is attached to the abdominal wall via three anchor sutures (gastropexy) that spontaneously fall out after three weeks (Druelle 2010 and Jacob-Beerens 2012). It avoids general anaesthesia and is less expensive. Nutrition can be started quickly, within 4 to 12 hours after application.

- Early complications: skin infection, pneumoperitoneum, usually minimal and spontaneously resolving, exceptionally digestive haemorrhage, colon or small intestine injury, or gastrocolic fistula.
- Late complications: site infection, burying of the collar and internal migration of the collar or balloon through the pylorus for the catheters, budding (gastric metaplasia), ulceration of the opposite stomach wall.

The overall mortality rate is 0.1 to 0.3%. (Faymendy 2015, Heuschkel 2015).

- **Onset or aggravation of GERD after gastrostomy**

Neurological impairment and the existence of pre-existing GERD appear to be significantly associated with the performance of anti-reflux surgery. The mortality rate associated with the procedure is 0.6%.

The ESPGHAN (Romano 2017) recommends that routine Nissen should not be used at the same time as GPEs, due to increased morbidity. The decision to perform a Nissen at the same time is restricted to children who at the time of the indication of their GPE have GERD resistant to medical treatment (c.f. ESPGHAN consensus).

- A retrospective study conducted on 684 children (mean age 2.9 years) showed there to be secondary Nissen's in 62 patients (9.1% of cases), on average, 20.7 months after gastrostomy, with a significant correlation between the occurrence of the Nissen's and severe neurological impairment (Ponsky, 2013).
- A prospective study published in 2018 on 326 children (follow-up from 2 to 13.5 years, average 3.5 years), 56% of whom had neurological damage, showed that reflux appeared in 11% of patients after GPE and pre-existing GERD was aggravated in 25% of cases (Aumar 2018). Only 53 patients (16%) required a Nissen, of whom 22 were treated within a year of the GPE.

- **Surveillance and monitoring (Brisse 2013 and Le Sidaner 2016)**

The presence of a catheter or button does not prevent bathing (bathtub, swimming pool or whirlpool, sea). and it does not prevent the intake of food by mouth (food for pleasure or an enteral nutritional supplement – ENS - to supplement food at night or insufficient hydration during the day).

Any accidental tearing of the catheter or button requires its emergency replacement: the unfitted orifice can close completely within a few hours (always have an advance button or, failing that, a Foley catheter if replacement is necessary and to preserve the ostomy orifice).

Inject drugs in liquid form or crush any capsules and dissolve and give them separately.

Rinse after each passage of medication or nutrients.

Turn the catheter or button a half turn every day. Do not cover with a bandage, especially an occlusive one. A gastrostomy opening rarely becomes infected: in this case, local rather than general care is appropriate.

- ***NB:*** **Jejunostomy**

The probe is placed beyond the pylorus into the jejunum. For some, this is an alternative to GPE + Nissen in cases of serious GERD with severe vomiting. Disadvantages: difficult to place, displacement and frequent obstruction (very thin catheter).

- **Conclusion on enteral nutrition**

Gastrostomy is currently a component of the usual management of polyhandicapped people. The indications must be carefully discussed with the family. Its indication requires clinical (weight, height, BMI) and nutritional (clinical and biological) assessments, the completion of food behaviour charts, the search for a cause for refusal to eat or eating difficulties (first and foremost, swallowing problems, followed by local or general pain, gastro-oesophageal reflux, poor positioning, etc.), and the completion of a speech and language assessment and then a swallowing assessment by an ENT specialist who is aware of the problem. The decision must be multidisciplinary (paediatric gastroenterologist, referring doctors, team), in agreement with the parents.

The GPE method is simple, reliable, and well tolerated. The introduction of enteral nutrition leads to a clear improvement in quality of life: reduced risk of bronchial inhalation, improved nutritional status, reduced number of hospitalisations (Di Leo 2019), and reduced parental and team stress. However, enteral nutrition via gastrostomy does not solve everything: persistence of respiratory aspiration, direct via saliva, indirect via GERD. It is advisable to continue working on swallowing and orality (speech therapist, team, parents). It is important to think of a gastrostomy as "one more mouth" that allows you to eat comfortably and safely. The original mouth is still there; even if it no longer allows eating, it retains its sensory capacities, which are sources of pleasure. For each polyhandicapped individual who is subject to enteral feeding, an ongoing team discussion should search for what it is possible to "dare" to propose, not to eat, but to taste (Rofidal 2018).

2.11 Respiratory disorders

People with a polyhandicap are highly vulnerable in terms of breathing for a combination of many reasons: lack of development of the pulmonary parenchyma, neurological disorders of respiratory control, narrowness and rigidity of the rib cage, scoliosis, inefficient coughing, etc. Added digestive problems hamper the efficiency of ventilation, such as respiratory aspiration of food or salivary, gastroesophageal reflux, bronchopulmonary superinfections, transit disorders, etc.

Respiratory damage is very common in the polyhandicapped person and is the leading cause of death (50 to 80% of deaths). Respiratory morbidity is proportional to the degree of neuromotor impairment. Restrictive syndrome and obstructive syndrome often combine, leading to increased respiratory effort and muscular and general fatigue.

Prevention and early detection are essential. Desaturation is rapid and fluctuates. Management must be comprehensive.

2.11.1 Preventive measures

- **Optimization of basic respiratory function**

Optimization of basic respiratory function involves the following measures:

- Adapt the position to allow a good head and trunk alignment; improve the often-hypotonic axial posture to optimize diaphragm activity
- Reduce the supine or semi-vertical position (moulded, thick foam total body positioning orthoses) and lateral and ventral free abdominal supine postures
- Treat scoliosis to limit the aggravation of the restrictive syndrome and dysfunction of the diaphragm
- Avoid chronic stercoral stasis for the same reasons
- Ensure proper hydration
- Evaluate and reduce the influence of iatrogenic drugs (respiratory depressants, such as morphine)
- Manage sleep respiratory disturbances, aggravated by pharyngeal hypotonia and frequent tracheal malaise (central pauses or neurological abnormalities of the breathing rhythm of Cheyne-Stockes type dyspnoea, obstructive pauses aggravated by pharyngeal hypotonia and glossoptosis, sometimes on macroglossia).

- **Prevention**

It also involves reducing the risk of bronchial inhalation and respiratory congestion *(see also Chapter 2.7 Eating disorders)*:

- Limit the risk of salivary inhalation and treat hypersalivation if necessary (good oral hygiene, scopolamine patches, atropine in infra-lingual drops in adults, injection of botulinum toxin into the salivary glands, surgery, even use of a mucosal aspirator)
- Adapt feeding to the chewing and swallowing ability to minimize any risk of misfeeding
- Include speech and language therapy
- Carefully ensure that the head and neck are positioned in a way that promotes swallowing, i.e. with the neck flexed (although this does not prevent a backward tilted sitting position), and follow measures to prevent swallowing problems related to the caregiver's position and feeding style *(see Chapter 2.7, Oral Timing and Swallowing Problems).*
- Avoid nasogastric tubes if possible
- Limit gastroesophageal reflux disease (GERD), which is so common due to the anatomical features of the oesophageal-gastric junction and gastroparesis, by straightening the sitting and lying postures (at least 60° of trunk straightening during any prolonged feeding phase - oral or enteral - and postprandial), avoiding overfeeding or excessive enteral flow at each meal, administering PPIs against the consequences of reflux, and using anti-reflux surgery if necessary (Nissen's procedure) *(see Chapter 2).8, GERD).*

- **The prevention of respiratory infections involves :**

- Routine vaccination (Influenza and Pneumococcus)
- Hygiene protocols: use of hydro-alcoholic solutions for workers, isolation measures in case of antibiotic-resistant bacteria.

2.11.2 Treatments

Whether it is a question of looking to reduce bronchial congestion or improve ventilation, the essential prerequisite for any respiratory care is the relaxation of the person. Some rely on basal stimulation.

- **Treatment of congestion and bronchopulmonary infections**

People with a polyhandicap are most often unable to induce an effective cough (hyposensitivity of the pharynx, poor muscle coordination).

The presence of sibilants on auscultation may justify the use of aerosol bronchodilators before the session. After gentle mobilization of the mandible and stretching of the oro-facial elements, manual and/or mechanical techniques of decongestion, drainage, and expectoration are used:

- In cases of atelectasis or peripheral obstruction, intra-pulmonary percussion with a mouth-nasal mask (percussion) is effective in moving secretions up the trachea without requiring the patient's participation. One can also resort to slow prolonged exhalations in the lateral decubitus position, postural drainage, or vibration.
- To clear the trachea and allow expectoration, either flow acceleration and/or pharyngeal suctioning (suctioning must not go beyond the intersection of the vocal cords), manual techniques dedicated to the proximal passages (rhinopharyngeal clearance by vallecular pumping, external tracheal pumping, induced cough), or coughing with mechanical assistance using a "Cough-assist" device should be practised in a manner adapted to the patient.

Respiratory distress requires appropriate antibiotic therapy and decongestion techniques. Manual drainage techniques (rhino-pharyngeal hygiene, vallecular pumping, induced cough) are coupled with mechanical techniques (aspiration, hyperinsufflation, intrapulmonary percussion ventilation, and non-invasive ventilation).

Decongestion is carried out in different postures (dorsal, lateral, and ventral decubitus) to improve lung recruitment and combat pathological contractures that considerably hinder ventilation. The implementation of these techniques seeks to limit the use of invasive ventilation (often ethically questionable) as much as possible.

- **Treatment of ventilatory disorders**

Attention should also be given to treating nocturnal and/or diurnal ventilatory disorders. They are also related to sleep disturbances, daytime sleepiness, with slowed ideation due to hypercapnia, epileptic flare-ups, or fatigue.

Attention to thoraco-abdominal positioning and trunk joint maintenance exercises is permanently required, as well as changes in position that affect ventilation. The lateral decubitus position facilitates ventilation of the upper side. The ventral decubitus position optimizes ventilation of the dorsal pulmonary segments.

Periods of motor freedom on mats, *a fortiori* with active turning exercises, assisted or not, even if the participation of the person is weak, aid cardio-respiratory functioning.

Breathing exercises using a pressure-relaxing device (Intermittent Positive pressure Breathing IPPB: Alpha 200 or Alpha 300 type) to perform hyperinsufflation promote lung expansion, mobilize the rib cage, and limit the risk of bronchial congestion. Theoretical contraindications, such as swallowing disorders, gastro-oesophageal reflux, and hypersalivation, require that this type of device only be used by experienced physiotherapists.

Non-invasive ventilation (with an adapted interface) aims to keep the airways open at all stages of the respiratory cycle. The benefit/risk ratio should be discussed for the polyhandicapped person without voluntary motor skills. It can be proposed as a first-line treatment for children or adults with daytime hypercapnia or sleep breathing disorders. In the presence of pharyngeal salivary congestion, care should be taken to avoid inhalation of the saliva and a nasal mask should be used rather than a mouth-nasal mask.

If there are mainly obstructive pauses, ventilation with a constant positive pressure of 8 to 10 cms of water should be used during the inspiratory and expiratory phases with a CPAP (*continuous positive airway pressure*) device.

If there is a restrictive hypercapnic syndrome and/or central breathing pauses, ventilation should be attempted at two different pressures for inspiration and expiration using a *bilevel positive airway pressure (*BIPAP) device by setting a sufficient inspiratory pressure level and a safe frequency. It may take some time to become accustomed to the device.

In cases of intense and frequent desaturation, oxygen therapy by oxygen extractor or portable tanks is a valid palliative treatment if there are no effective therapeutic means (installed pulmonary fibrosis).

There has been little investigation of nocturnal ventilation in polyhandicapped patients, which prevents the detection of pathologies, such as sleep apnea syndrome and/or global alveolar hypoventilation. Both of these impairments have important consequences on the quality of life. If evocative clinical signs are detected, and if the patient's life plan is consistent with the possibility of ventilatory treatment, additional examinations may be programmed: nocturnal pulse oxymetry, polygraphy, and capnography. Only after all other possible solutions have been investigated will the ventilatory treatment be implemented by a team of experts in this field. In practice, this is rarely feasible. Screening for possible sleep apnea is more justified for warning the family of respiratory precariousness and the risk of rapid lethal complications than for an appliance.

With increasing age, many phenomena lead to states of predominantly restrictive respiratory insufficiency (severe scoliosis, swallowing disorders having caused many episodes of pneumopathies, etc.). The seriousness of the situation, particularly with repeated episodes of pulmonary infection, leads to a discussion with the entourage and the multidisciplinary team on the course of action to be taken in case of respiratory decompensation, in particular possible limitations of care (avoiding a tracheotomy, intubation, invasive assisted ventilation).

In rarer cases, the decision may be in favour of performing a tracheotomy after discussion with the institution and on condition that day and night care staff with the authority to perform tracheal aspirations (nurse,

nurse's aide, or other appropriately trained personnel) are available. This can facilitate aspiration, limit bronchial aspiration of saliva when the tracheotomy is a balloon tracheostomy, and promote bronchopulmonary comfort by reducing congestion. Assisted ventilation by tracheotomy is very rarely proposed; it is more often the result of a failure of deventilation during an episode of pulmonary infection with invasive resuscitative management.

In general, treatment should be provided after assessing the benefit/burden balance and based on informed consent following discussion with the person's family and principal care team, as well as continuing support.

"The care project must be in the service of the life project of the person with a polyhandicap, and should be continuously reevaluated ..." (Elisabeth Zucman)

2.12 Urinary and vesico-sphincter disorders

It is important to point out that having a polyhandicap does not necessarily mean incontinence, in children or adults (information sheet on the website of the Association Ressources Polyhandicap Hauts-de-France - A. Blanchard). Careful screening for signs that express a need, the implementation of satisfactory urination conditions (with adapted facilities, particularly in terms of postural maintenance), or even scheduling urination at regular hours may enable some people with a polyhandicap to not to be forced to wear permanent protection. For older boys or men with urinary incontinence, a penis pouch device can be offered to avoid remaining in the urine, while monitoring the correct adaptation and tolerance of the device.

However, urinary behaviour is often disturbed by a variety of causes.

2.12.1 Urinary disorders of neurological origin

People with a polyhandicap almost always have abnormal vesico-sphincter function. Two of three have an unstable bladder due to the lack of inhibition usually exerted by the cortical centres on the urinary centre of the brain stem and spinal cord. Clinically, frequent urination during the day and night and urinary urgency, with or without leakage, are observed. Such bladder hyperactivity is often associated with vesico-sphincter dyssynergia (a lack of relaxation of the urinary sphincter during urination), causing dysuria with poor quality bladder emptying (post-void residual urine greater than 100 ml in adults, which tends to increase naturally with age) and potentially recurrent urinary tract infections. In the case of vesico-sphincter dyssynergia, the detrusor may tend to become muscular (bladder control), with a reduced possibility of compliance (distensibility), contributing to frequent urination or later even to vesico-ureteral reflux due to high-pressure urination.

Incontinence may also be related to a person's failure to recognize the need to urinate or to bladder sensory disturbances that delay the feeling of need and cause overflow urination.

Routine bladder-renal ultrasound screening, especially in very spastic individuals, searches therefore for the existence of a possible resistant bladder (thickened and diverticular bladder wall) and if this is the case, possible upper urinary tract abnormalities (vesico-ureteral reflux, pyelocalic dilatation, lithiasis).

Faced with this type of neurological urinary dysfunction, drug treatment to limit vesico-sphincter dyssynergia and retention is preferred, namely alpha-blocking therapy (e.g. alfuzosin, tamsulosin, etc.). Urinary leakage due to urgency may possibly justify the prescription of an anticholinergic treatment (e.g. solifenacin, trospium chloride, etc.) while closely monitoring the absence of side effects, such as constipation, urinary retention, or dry mouth. Other remedies, i.e. hetero-catheterisation, relief of male incontinence (by endo-urethral prosthesis or definitive sphincterotomy), or non-continent Bricker-type urinary diversion, should be discussed on a case-by-case basis with a team specialized in neuro-urology. The use of an indwelling catheter should be avoided as much as possible, as it may cause multiple medium- or long-term complications (infectious, lithiasis, neoplasia, etc.).

2.12.2 Other causes of urinary disorders

- **Possible causes:**

- Constipation
- The existence of some type of irritation (physical discomfort or suffering)
- Certain medications (especially those with an anticholinergic effect)
- Masked urinary tract infection (watch for changes in the frequency of urination, the colour or odour of the urine, or any general disturbance: change in behaviour, change in basal temperature)
- Behavioural disorders (urination behaviour in relation to a frontal syndrome unrelated to vesico-sphincter dysfunction).

- **Possible reviews (see Chapter 3.2.2, HPP monitoring table)**

Follow-up should be based on clinical elements, especially infectious, which most often show chronic retention, and on regular monitoring of post-void residual urine by *bladder scan* or bladder-renal ultrasound to check for upper urinary tract anomalies, only occasionally in the absence of alarming urinary signs or annually if there is a problem. The same applies to the renal biological check-up.

2.13 Sleep disorders

Sleep disturbances of the polyhandicapped are poorly described and insufficiently taken into account, despite their frequency and often significant impact on the individual and his or her family.

The ICSD3 (International Classification of Sleep Disorders) classifies sleep disorders into six subdivisions (ICSD 2014). In those with a polyhandicap, it is possible to schematically distinguish between three groups.

2.13.1 Sleep disorders of organic origin

- **To be suspected if there is/are:**

- Signs of pain (objectified by hetero-evaluation scale)
- Long awakenings (> 15 min), especially in the first part of the night, decreased sleep time
- Restless sleep between awakenings
- Abnormal daytime hyperactivity and/or behavioural disorders or, on the contrary, daytime fatigue and/or unexpected naps
- Deviation from the weight-height curve,
- Abnormal snoring, excessive sweating (sleep apnea syndrome, see below).

- **The causes:**

- Pain, regardless of its origin: orthopaedic, digestive, neurological (spasticity, dystonia, neuropathic pain, restless legs syndrome, etc.), stomatological, ENT, urinary, etc.
- Social and family anxiety and stress, parental fatigue
- Anxiety disorders during sleep, daytime behavioural disorders
- Epileptic seizures: disturbance of sleep physiology
- Anti-epileptic drugs and other drugs that cause daytime drowsiness (hypnotics, anti-allergy drugs, analgesics, etc.)
- Psychostimulants during the day
- Too frequent or long naps and rising too late
- Discomfort (wet or soiled diaper)
- Parasomnia (nightmares, night terrors)
- External disturbances (light, noise)
- Sleep apnea syndrome (SAS),
- Associated chronic diseases: certain chronic diseases (respiratory and cardiac) decompensate more readily at night and may lead to a pattern of excessive daytime drowsiness.

2.13.2 Problems with installation of the day/night rhythm

- **Clinical signs:**

- Difficulty falling asleep: when falling asleep, the brain inhibits existing excitatory stimuli; this process can be deficient in the polyhandicapped child
- Fragmentation of sleep, with frequent awakenings: short sleep cycles, as in small children = free running ultradian rhythm (hyper-ncthemeral syndrome), for example in blind children (an ultradian rhythm is a biological rhythm that occurs at a frequency higher than the circadian rhythm, with periods of a few minutes to a few hours, i.e. a multi-daily frequency)
- Loss of sleep-wake cycles or in the most extreme case, day-night inversion
- Delays, or more rarely phase advances, corresponding to a malfunction of the internal clock.

- **Causes: lack of time indicators**

These can also be considered as the absence of external synchronizers (Challamel 2004, Teulade 2017):

- Lack of day-night alternation (blind)
- Lack of sufficient daytime physical activity
- Meals not given at a fixed time (fragmented feeding or continuous enteral nutrition)
- Poor distribution of sleep over the 24 hours (late rising, too frequent or long naps, unfavourable role of drugs acting on the central nervous system)
- Poor appreciation of social time indicator related to cognitive impairment.

Specific abnormalities of the waking-sleeping rhythm are observed in Rett syndrome (progressive loss of day-night organization), Angelman syndrome (irregular waking-sleeping rhythm), and Smith Magenis syndrome: phase advance (inversion of the rhythm of melatonin secretion), irregular waking-sleeping rhythms (Challamel 2004).

2.13.3 Sleep disorders of respiratory origin

- **Two stages:**

- Upper Airway Resistance Syndrome (UARS): progressive increase in respiratory effort but without hypoxemia, without variations in oro-nasal airflow, followed by micro-awakening, which allows normalization of breathing but fragments sleep.
- Obstructive Sleep Apnea Syndrome (OSAS): obstructive apnea/hypopnea (≠central apnea): interruption or 50% decrease of ventilation for a minimum of 10 seconds in adults or 5 seconds in chidren, with hypoxemia and or awakening at least 10 times per hour of sleep in adult and 5 times in chidren.

- **Clinical signs:**

- Loud nocturnal snoring + daytime drowsiness: snoring, followed by interruption of snoring by apnea, followed by the resumption of sound breathing with agitation
- Mouth breathing, wet pillow, night sweats
- Morning headaches, cognitive problems during the day (attention, concentration, etc.), abnormal drowsiness, behavioural problems, unexpected naps
- Restless sleep - parasomnia.

- **Causes:**

- Hypotonia of the upper airways, atony of the soft palate, pharyngotracheo-malacia, tonsil hypertrophy, choanal atresia, craniofacial anomalies, facial hypoplasia
- Oro-pharyngeal incoordination, role of gastroesophageal reflux
- Neuromuscular diseases, with or without brain-stem involvement
- Diseases of overload, including muco-polysaccharidosis, hypothyroidism, etc.
- Genetic diseases.

- **Mainly clinical diagnosis**

Diagnosis is primarily based on the medical history, interviews of the caregivers, and a sleep record (the booklet "Sleep disorders and disability", which can be downloaded from the Réseau-Lucioles website: reseau-lucioles.org, can help).

Mechanical respiratory disorders of the sleep apnea type may justify a nocturnal SpO_2 recording (Stagnara 2011), which can be confirmed by polygraphy. Polysomnography, which is difficult to perform in the context of a polyhandicap, may be performed if there is any doubt. A 24-hour EEG may be useful if there is of suspicion of nocturnal seizures.

2.13.4 Prevention and treatment of sleep disorders

- **Prevention**

➢ Time indicators: creating a conducive environment by providing the person with time indicators (chronotherapy): teaching the person to fall asleep alone, reorganizing the distribution between sleeping and napping over 24 hours, going to bed and waking up at set times, bedtime ritual, transitional object, temperature, noise, light, meal times, physical and play activities.
➢ Prevention of organic causes: comfortable night-time position, prevention of pain, treatment of behavioural disorders.
➢ Luminotherapy: for sighted individuals with a polyhandicap, if there are sleep/wake rhythm disorders.

- **Drug Treatment**

Hypnotics should only be used temporarily, preferably with a short duration of action. Benzodiazepines can have a paradoxical effect.

Melatonin provides good results for people with a polyhandicap, especially for disorders of circadian rhythm, which it restores. The formulation can make administration difficult. Basic melatonin is preferable but it lacks the delayed effect of the slow-release formulation against midnight awakenings. It should be administered from 30 minutes to one hour before bedtime.

- **Treatment of sleep apnea**

➢ Discontinuation of medications that may cause pharyngeal hypotonia
➢ Adenoidectomy, ± uvula surgery ± maxillofacial surgery
➢ Non-invasive ventilation (NIV): in this case, a benefit-risk balance should be assessed, with treatment providing sufficient improvement in daytime quality of life to balance the strain, discomfort, and risks for a person without voluntary motor control of ventilation every night (Teulade J, 2017)

2.14 Visual disturbances

"It is estimated that 80% of our perception of the outside world is visual" (Meyniel P.Y. Robert 2017).

Vision is a complex neurosensory system that plays an important role in the motor, cognitive, relational, and social abilities of the person with a polyhandicap (Jacquier 2017):

➢ Vision is involved in controlling posture and movement
➢ Analysis of visual stimuli allows the development of fine motor skills
➢ Mental representations and visual exploration complement these functions and provide the possibility to master the environment.

2.14.1 Prevalence and clinical aspects

The EVAL PLH study reported the prevalence of visual disorders to be 25% (MC Rousseau 2019), without any details on the disorders presented.

Two types of disturbance are possible (Jacquier 2017): peripheral (the eye and its movements) and central (peripheral nerves and various nerve centres).

- **Peripheral disturbances**

The eye itself can harbour morphological abnormalities linked to embryonic pathologies, as well as cataracts, retinal detachment, eyelid eczema, etc. These pathologies can be congenital or the consequences of trauma or infections linked to various causes, including some that are stereotypical.

Eye movements enable the gaze, which can be used as a tool for inter-human communication or to even control a computer and develop alternative communication in the absence of verbal language. For this to happen, eye movements must be able to be directed voluntarily and the functioning of the annexes of the eye, in particular the oculomotor muscles, must not be disturbed, which may be the case during epileptic seizures or paralysis due to intracranial hypertension (HTIC) or any other pathology, such as nystagmus or cerebellar syndromes.

- **Central disturbances**

Neurological pathways can be affected by a pathology that causes a polyhandicap, and attempts should be made to explore optical pathways (visual field amputations), occipital areas (visual recognition abnormalities, perceptual-cognitive visual function disorders, total lack of conscious vision), the occipito-parietal pathway (analysis and action pathway), of which the impairment alters visuo-spatial perception, and the occipito-temporal pathway (visual recognition and focal vision pathway), of which impairment leads to agnosia, as well as imprecise gestures and fixation disorders.

2.14.2 Screening and observation

It is very difficult to accurately assess acuity and visual abilities because of the considerable communication difficulties of those with a polyhandicap, which make it difficult to understand instructions and answer questions. It is therefore necessary to focus on visual behaviour, taking into account the movements of the person with a polyhandicap, and to interview the person's family and friends who know him or her well. Acknowledging the lay knowledge of family members and caregivers also means valuing the role and expertise of parents and generally supporting the commitment of caregivers for today and tomorrow.

These exams must be performed at the right time, in the right environment, using the right equipment. Sometimes, the examination can be performed at the patient's bedside with portable equipment. Ophthalmologists and orthoptists trained in these techniques and experienced with polyhandicapped people can help to make a proper assessment.

- Refraction must be assessed systematically to detect myopia or hyperopia, astigmatism, and important refractive asymmetries between the two eyes; this should be done as early as possible in life so that maturation of the visual system occurs on the basis of the most normal information possible, then regularly and in case of behavioural changes.
- The visual field is assessed using double confrontation methods.
- Direct examination of the eyelids, conjunctiva, cornea, crystalline lens (cataract), fundus (detachment and other retinal pathologies) is essential but often difficult: it must be performed as rarely as possible under general anaesthesia.
- Complementary examinations are sometimes necessary in the exploration of vision: these include visual evoked potentials and/or an electroretinogram after MRI and EEG have raised a suspicion of visual impairment, which can only be confirmed by a clinical examination, as there is not always a direct link between MRI or EEG abnormalities and an individualised pathology.

Beyond the visual impairments themselves, the use of visual information can be problematic due to cognitive impairment, especially difficulties with attention and memory, which hinder all visual recognition and learning activities for vision control. Possible compensation must be sought and a lengthy coordinated effort by all stakeholders make it possible to put in place the compensations that will enable the person with a polyhandicap to acquire both visual skills and use them cognitively.

2.14.3 Support

After the optimal resolution of ophthalmological problems, optical corrections, surgery, and a complete evaluation of the individual’s general possibilities, professionals from several disciplines (orthoptists, occupational therapists, psychomotor therapists, educators, nurses, MPAs, caregivers, etc.) must work together to understand all the disorders and set motor, visual, and daily life improvement objectives.

The compensation of visual disabilities, based on the person's abilities, is sought through multidisciplinary work on the following themes:

- Motivation to visually examine the surroundings: quality of the visual environment, games, and comfort of the position, including a posture favouring the gaze
- The tracking of movements in the person's immediate environment and the communication possibilities this allows
- Peripheral visual stimulation to improve axial tone (e.g. Bullinger structured panels)
- The individual visual preferences of each person with a polyhandicap to adapt the individual project
- Regular repeated identical training for communication or locomotion activities involving vision, according to the person's possibilities
- The use of computerised or non-computerized material for compensation.

An important resource is available for all people with a polyhandicap and their families, as well as the professionals who work with them: the National Resource Centre for Rare Disabilities (CNRHR) in Loos (59). The CNRHR aims to aid people with visual impairment (blindness, low vision, neuro-visual disorders) associated with other disabilities; the association of such disabilities is very complex, always singular, and often intertwined with relationship and communication disorders. Each situation in a particular environment is unique and involves the cooperation of several partners (Contact details: CNRHR LA PEPINIERE 8 allée GLATIGNY 59120 LOOS, tel 03 20 97 17 31).

2.15 Hearing problems

2.15.1 Prevalence and clinical aspects

A single recent epidemiological survey (Rousseau 2019) carried out on 875 polyhandicapped people showed a prevalence of hearing problems of 6% versus 28% for visual disorders. A personal study at the Centre Antoine de St Exupéry in Vendin le Vieil (62) on 317 files estimated the prevalence to be 7.4%.

- Definition of deafness: elevation of the threshold of sound perception, regardless of the degree
- Degrees of deafness: *(Annex 4, see link ch 2.15, Table 1)*
- Conductive and sensorineural deafness: *(Appendix 4, see in connection with Chapter 2.15, Table 2, Location of deafness)*

Permanent deafness in the polyhandicapped person is of the perceptual type and its causes include certain pathologies that generate both the polyhandicap and deafness, often profound:

- Antenatal conditions: viral infections (rubella, CMV), degenerative diseases (mitochondrial cytopathies), CHARGE syndrome
- Perinatal: perinatal distress: anoxia, prematurity, cerebro-meningeal haemorrhage, kernicterus jaundice
- Post-natal: meningitis, meningoencephalitis, head trauma.

Nevertheless, it is important not to forget about conductive deafness, most often transitory, either due to obstruction (earwax plugs, foreign bodies) or seromucous otitis, acute infectious otitis, or even chronic otitis: any ear discharge requires an examination of the eardrum, especially if it is foul smelling or recurrent.

2.15.2 Screening for deafness

- **During the neonatal period**

The decree of November 3, 2014 relating to the national specifications of the screening programme for permanent neonatal deafness (PND) makes it compulsory to screen new-borns for deafness in maternity or neonatology departments using objective or non-objective methods.

Method: otoacoustic emissions (OAEs) or automated auditory evoked potentials (AEPs): see their indications and results *(Appendix 4, cf. in connection with Chapter 2.15, Table 3)*.

- Protocol: A first test is carried out (after 48 hours of life for OAEs and from D 1 for AEPs). If positive, a retest is performed before leaving the maternity ward and in the event of a new positive result, an additional electro-physiological assessment is performed in a specialized environment to determine the extent and type of deafness and assess possible central involvement (auditory evoked potentials

of the brain stem, etc.), in addition to a complete ENT examination and subjective audiometry (study of behavioural reactions to sound stimulation).

- Ideally, confirmation of the diagnosis is possible at 3 to 4 months of age, fitting at 6 months, and cochlear implantation at 12 to 18 months.

- **Screening for deafness after the neonatal period: deafness is unknown**

- When investigating the aetiology of a polyhandicap, a neuro-sensory assessment must be systematically performed.
- Deafness in these cases is often diagnosed late. It is a disability that often takes second place to motor, visual, and epileptic problems, all the more so as the deaf child may use other strategies to enter into communication, giving the impression that he or she can hear (Dorche 2017), or, on the contrary, present behavioural disorders or autistic withdrawal through sensory deprivation, masking their true origin.
- Prolonged observation of the reactions of the child or adult by family or professional carers is therefore essential (Leman 2014, Association Ressources Polyhandicap Hauts-de-France website):
  - Is the alert function (sound source outside the field of view) present?
  - Does the person respond to calling their first name in a normal voice or understand the meaning of the words?
  - Does he or she perceive a whispered voice out of sight?
  - Does he or she hear high-pitched sounds (bell)? [If not, the person "seems to hear" but cannot understand due to a lack of high-pitched perception.]
  - Does he or she understand better if he or she looks at the person speaking or if the words are accompanied with gestures?

In case of doubt, a referral can be given by the general practitioner (referral to calibrated sound toys, tuning fork) but a consultation with an ENT specialist accustomed to these types of patients will be necessary with subjective audiometry (study of behaviour during sound stimulation) followed by an electrophysiological check-up.

- **Late onset deafness. Presbycusis**

Presbycusis can become problematic from approximately 65 years of age. Hearing aids help to avoid increasing isolation.

2.15.3 Management of deafness

Sensorineural deafness should be diagnosed as early as possible, even if it is late, whether the hearing loss is bilateral or unilateral.

However, devices are sometimes poorly supported and removed, particularly when deafness is diagnosed late, the device is poorly adapted (under-correction or, conversely, discomfort), or there are autism spectrum disorders. Rehabilitation is also difficult and the duration of sessions must be kept short because of the individual's attentional lability. During the day, the device should ideally be worn all the times, or, if it is not well supported, at certain times when the person can be cared for exclusively. Fitting with hearing aids or even cochlear implants must provide an alert function, a perception of the temporo-spatial environment, and emotional communication (melody of the voice), with the hope of more-or-less coded language.

Communication aids (see this chapter) are also implemented: pictures, pictograms, Makaton.

Anyone fitted with a hearing aid must receive, in the initial stages of the fitting, auditory education by a speech therapist and audiophonological and audioprosthetic follow-up by an ENT specialist at least every year to ensure the effectiveness and good tolerance of the fitting.

2.16 Communication disorders

Communication is fundamental to every human being. A person with a polyhandicap is all the more vulnerable, as communication is difficult and disrupted by the motor, perceptive, and cognitive disorders that characterise this complex disability (decree N°2017-982 of 9 May 2017[2]).

As the person has little access to oral language, communication often remains non-verbal. It thus passes through posture, facial expressions, glances, intonations, pointing, and behaviour, which are also sometimes disturbed by motor and sensory disorders or by undesirable events (environment, pain, emotions, etc.) (D. Crunelle, 2018). Sometimes the interlocutor is distraught, does not perceive what the person expresses, or interprets ambiguously (Cataix-Negre, 2011).

However, polyhandicapped individuals communicates and must be recognized as communicative beings and acknowledged in the exchanges. Exchanges with brothers and sisters should also be supported so that, under the best possible conditions, they do not give up communicating.

"The polyhandicapped person probably has a thought process that escapes us, on the order of sensitive intelligence" (Zucman 2011).

2.16.1 Basic principles of communication with the polyhandicapped person

To communicate, the polyhandicapped depend on the person they are communicating with. He or she can only be heard if the caregiver agrees to hear him or her by reserving the necessary time for the exchange, taking care to give him or her the necessary tools, avoiding rapid interpretations, which often lead to misinterpretation, and favouring the expression of emotions and feelings. "If we know how to listen to them, people with a polyhandicap bring us incomparably more than we can offer them" (Tezenas du Montcel, 2017).

The most important thing is to be convinced that the person has things to tell us. It is, in particular, through everyday acts, which must be "facilitated, thought, and spoken" (Crunelle, 2018, Chavaroche, 2017), that the foundations of communication are built and that the person, over time, can share his or her experiences.

Certain very simple-to-use tools, such as a life book, logbook (Crunelle, 2018), or communication passport (Cataix Negre, 2017), provide content for communication with the person within his or her personal history, and allow communication to have a narrative function (Chavaroche, 2017).

2.16.2 Possible means of communication

All communication "aids" are grouped under the concept of enhanced alternative communication (EAC). EAC is implemented on the basis of a precise evaluation, first of motor, sensory, and cognitive skills (Beukelman et al, 2017) and then communication skills, both receptive and expressive (Crunelle, 2018), carried out in close consultation between natural and professional caregivers. "Over time, the people closest to those with a polyhandicap manage to differentiate between what is intentional communication, what manifests as an emotion, and what is merely reflex... It is the confrontation between the different views of the subject that can be the source of a better understanding of what he or she expresses" (Scelles, 2017).

Alternative and improved communication tools are built from the person's experience, desires, needs, and emotions, and used by all caregivers to support the oralization process. In a multimodal approach, all types of means can be used (Cataix 2017), i.e. communication through sensoriality (Fröhlich), body language, sign or French sign language (LSF), or signed codes. Each positive strategy that the person uses in a communication process must be recognized, encouraged, and taken up by all interlocutors, sometimes adapted to the interlocutor. "A person may be able to communicate with family members very effectively... and may need other approaches with unknown interlocutors" (Beukelman et al, 2017). Technical aids, which are methods

[2] Decree No. 2017-982 of 9 May 2017 on the nomenclature of social and medico-social establishments and services for disabled or chronically ill persons.

that involve equipment, may also be used. They may or may not be technology based. They range from a communication notebook/table to highly sophisticated tools.

To help the person develop his or her communication, each speaker must also be informed of the new words or ideas targeted, and of the chosen strategy; each person regularly uses the gesture, object, image or pictogram chosen, while at the same time oralizing what he or she evokes. It is such modelling that allows, little by little, the understanding of the new concept, and, perhaps, its expression. Thus, based on shared observations and with the support of all family and professional caregivers, an individual communication plan can be developed and become part of the person's project. It can be implemented by all those who surround him or her on a daily basis and take the time to wait for signs of engagement. The person's personalized tool becomes his or her passport to greater autonomy.

2.17 Thymic, behavioural, and psychiatric disorders

These consist of behavioural disorders and disruptive, challenging, and problem behaviours in people with a polyhandicap.

The term "behavioural challenge" has gradually replaced the term "behavioural disorder", showing that people with such disorders pose "true challenges to the organization of reception structures and community integration, particularly in the nature of the services that meet the needs of the person with a disability" (Lambert 2003).

Two recent studies summarize the difficulties of diagnosing and evaluating such disruptive behaviours, the complexity of their support, and the seriousness of their consequences for individuals and their families: "Disruptive behaviours in people with brain injuries acquired before the age of 2 years: prevention and care" (Haute Autorité de Santé, HAS 2014) and, more recently in 2016, the INSERM Expertise, Intellectual Disabilities, in several chapters.

2.17.1 High prevalence of challenging behaviours

There are few studies with sufficient series in this topic and prevalence data most often focus on one type of behavioural disorder rather than their entirety. The prevalence of challenging behaviours is high among people with a polyhandicap, approaching 30%. Although this figure is only an approximation, it is high. Additional data are in accordance with this estimate: such disorders are more frequent when there is a very severe intellectual disability and they are observed in nearly three-quarters of cases in disabled males.

2.17.2 A difficult diagnosis

- **A preliminary observation**

The situation of having a polyhandicap favours the appearance of behavioural disorders in these people because of their difficulties of adaptability due not only to their disability (especially mental) but also their insufficient acceptance from their social environment and society. Behavioural disorders should not be considered solely as a disorder inherent to the disabled person but as the result of interactions between that person and his or her environment.

- **Identification of behavioural disorders**

The identification of behavioural disorder requires:

- Prolonged shared observation by all the professionals accompanying these people, along with the parents and families, with the obligatory help of the psycho-psychiatric teams
- Establishing communication with these people, which is not simple.

There are three main categories of challenging behaviours: self-injurious behaviours, which are common among those with a polyhandicap, aggressive behaviours, and stereotyped behaviours, not to mention withdrawal, which too often goes unnoticed.

These behavioural disorders are very often intertwined with other psycho-pathological disorders. However, any abrupt change in behaviour must have a somatic cause (pain, digestive disorder, etc.); the two most frequent abrupt changes in behaviour are depressive states and pervasive developmental disorders (PDDs).

Identifying them is all the more difficult because of the presence of a mental disability and requires the help of psycho-psychiatric teams specializing in these problem behaviours who know how to use the scales and tools to evaluate their impact on the adaptability of these individuals, understand the origin of their occurrence, and improve them with adapted support methods.

The meaning of such behavioural disorders in these people, who have difficulties to adapt, must be systematically sought: their overall state and modalities to make them understood; it is essential to take this into account to care for these people.

In addition, shared observations make it possible to recognize:

- Whether these challenging behaviours are recent, as they are often due to a physical problem, such as pain, psychological suffering, or a modification of the environment; any change in the usual behaviour of these people with communication difficulties reported by someone who knows them ("I don't recognise him, he is not as usual") must be assessed for a cause
- Whether, on the contrary, they have existed for a long time, forming part of the psycho-pathological profile of the person or his environment, or even his quality of life.

2.17.3 Serious consequences

The consequences of such challenging behaviours are very serious for those with a polyhandicap or cerebral palsy and those around him or her.

- For the individual, these disorders are responsible for a decrease in adaptability and participation, and severely disrupt the relationships with those around them. They significantly complicate care, education, learning, and, ultimately, the person's quality of life. They run the risk of social rejection, refusal of admission, and disruption of their life course.
- For professionals, parents, and families, they are a source of discouragement, exhaustion, and stress, which can sometimes lead to intolerance towards these people, with the risk of abuse.

2.17.4 Difficult management of care

The modalities of care must be established by the psychiatric teams and shared with the professionals and families; we will highlight only two aspects. (Piveteau D., Rapport Zéro Sans Solution, 2014)

- Improve the acceptability and tolerance of the challenging behaviours of these people by all those around them (professionals, family members, and friends) through training, support, guidance, and the sharing of tasks among these stakeholders with the help of specialized teams, so that none of them find themselves in the unacceptable situation of "No Solution".
- Favour non-medicinal management first, with the exception of acute disruptive behaviours that require urgent therapy. Non-medicinal management, guidance, and treatment of a cause must always precede the taking of medication; it should be remembered that in 70% of cases, psychotropic drugs are used in these people without a precise diagnosis.

2.18 Special case of degenerative diseases

The polyhandicap of individuals is likely to worsen due to the interlocking nature of the disabilities and respiratory, osteoarticular, digestive, infectious, or other complications that can occur, despite active care and appropriate prevention. However, the term progressive or degenerative disease is reserved when the cause of the polyhandicap is itself progressive, leading to the progressive aggravation of the cerebral dysfunction that causes the polyhandicap.

- **Clinical aspects**

Progressive conditions are most often genetic or metabolic but can also sometimes be infectious diseases or certain forms of epilepsy. In these pathologies, the child may show perfectly normal development for a few

weeks, months, or even years before the neurological manifestations become noticeable and gradually more pronounced until they cause the polyhandicap. Throughout the period of normal clinical development, the disease is already present and causes cerebral dysfunction and lesions that become quietly established, but already affect the brain. One of the consequences of such degenerative diseases is that the signs of neurological disease occur gradually and the perception of the disease and disability is experienced differently by the parents than when they occur very early. Sometimes the child perceives the deterioration of his capacities, which may initially be more marked for motor skills than intellectual and communicative abilities. This difference in the perception of a polyhandicap by parents is also present when it occurs suddenly, as a consequence of an acute illness or a domestic accident for a child whose development was previously perfectly normal.

- **Etiological diagnosis**

The etiological diagnosis can sometimes be urgent, especially at the beginning of a new pregnancy. Indeed, a genetic diagnosis can then be made for the current pregnancy and make it possible to know the status of the foetus. These situations are extremely difficult. It is necessary to accompany them closely, and the family must be referred to specialised multidisciplinary structures with complete data on the disease.

- **Specificity of support**

Although, once installed, the time of care and treatment for degenerative forms of disability is similar to that of other forms of polyhandicap, the diagnosis and installation phase of the disorders requires special care, aiming to maximally preserve the possibilities of communication, perception, and interaction with the entourage. The end-of-life situation may thus occur earlier and this perspective must be an integral part of the management of care, without the lifespan ever being predictable. It is therefore necessary to refrain from any pronouncements of life expectancy, which would be detrimental to the child and his/her family, as well as to the dynamics of care.

At the same time, it is necessary, as with any situation of polyhandicap, to actively prevent over-disabilities, of which it is sometimes difficult to distinguish between those due to the degenerative disease itself and those that can be slowed down.

Some of these pathologies require specific management according to their aetiology, in addition to that of the polyhandicap, and may benefit from their own treatment, which can sometimes be preventive; they may also sometimes require special precautions.

2.19 Cancers and polyhandicap

The incidence of cancer in people with a polyhandicap is still poorly known, with a varying distribution among organs due to specific risk factors and lifestyle. Screening and surveillance thus need to be adapted.

2.19.1 Frequency and distribution of cancers

Indirect information can be derived from the distribution of cancers in people with severe and profound intellectual disability (SID) [Satgé 2016]. The distribution of cancers is different from that observed in the general population: there is less prostate, lung, and cervical cancer and an incidence close to that in the general population for breast, uterine, and ovarian cancer. On the contrary, there is a higher prevalence of digestive cancers and cancers of the testis (x10), gallbladder (x10), thyroid (x3), and brain (x3.5) [Patja et al 2001, Sullivan et al 2004].

2.19.2 Risk Factors

The risk factors for people with a polyhandicap differ from those in the general population due to the presence of fewer environmental agents, which account for 41% of current cancers in the general population: mainly tobacco, alcohol, occupational toxicants, ultraviolet radiation, being overweight, obesity. However, the physiopathological causes are more numerous. Within the group of environmental agents, the risks linked to a lack of physical activity and those attributable to infectious agents (which together are estimated to account for only 5% of the risk of cancer in the general population) remain. Chronic gastric infection with

Helicobacter pylori promotes gastric cancer and a history of hepatitis B or C increases the likelihood of liver cancer. A second group of risks concerns pathophysiological disorders, such as GERD and neurological bladder, which favour cancers of the lower oesophagus and bladder, respectively. A third group consists of cancers related to the genetic pathology responsible for the polyhandicap. Certain conditions increase the risk of cancer, whereas others reduce the risk for all cancers or some tumours. For example, trisomy 21 strongly favours leukaemia in childhood and testicular tumours in young men but protects against breast cancer and nerve tumours [Satgé et al 1998]. Bourneville's tuberous sclerosis increases the risk of brain and kidney tumours. These increases in frequency can be observed from childhood for many genetic disorders, for example, for the two conditions mentioned above.

2.19.3 The various cancers and their screening

- **Common cancers in the general population**

- Breast cancer: it is advisable to carry out organised screening, as performed in France, from 50 to 74 years of age with a mammogram every two years, as the risk is currently estimated to be equivalent for DIS women (who are also unprotected by pregnancy, breastfeeding, and regular physical activity). If screening by mammography is difficult because of marked intellectual impairment and physical disability, ultrasound is recommended, despite the higher number of false positives and this method being operator dependent [Poulos et al 2016, Iezzoni et al 2011].
- Colorectal cancer: because the risk is theoretically higher [Willis et al 2018] than in the general population, it is recommended that, pending more epidemiological data and as a precautionary measure, organized screening should be performed in the 50 to 74 age group, as in the general population.
- Cervical cancer: as this cancer is primarily related to sexual relations and has been reported only rarely in women with intellectual disabilities [Satgé 2016], the decision to screen should be evaluated by the physician following the person, based on individual criteria.

- **Other cancers - related to polyhandicap**

- For cancer of the lower oesophagus, which is very often linked to GERD, monitoring can be carried out by pH measurement, as the risk increases the lower the IQ is below 35 in cases of cerebral palsy and antiepileptic treatment [de Vire et al 2008] and symptoms can be difficult to detect [de Veer et al 2008].
- Chronic gastric infection with *Helicobacter pylori* is increasingly likely the longer one stays in a medical-social institution. It should be sought to prevent stomach cancer.
- Bladder cancers are favoured by the chronic inflammation associated with urinary stasis and repeated infections in cases of marked neurological urinary disorders. Monitoring by urinary cytology is desirable, especially in cases of spastic cerebral palsy and significant symptomatic urological dysfunction.
- Testicular cancer is 10 times higher in DIS men and those with cerebral palsy associated with significant comorbidities, as well as primarily bilateral cryptorchidism, than in the general population. Screening is performed by annual palpation of the testicles between the ages of 15 and 40 by a same-sex professional. It may be supplemented by ultrasound if there is any doubt [Peate and Maloret].

It is important to keep in mind that any type of cancer, even if uncommon, can occur in a person with a polyhandicap. This possibility should be sought in cases of unusual disorders and symptoms, including unexplained behavioural changes. Symptoms may be very different from those generally encountered in the general population.

2.19.4 Treatment and prognosis

Observations reported in the literature show that the treatment of cancer in people with a polyhandicap is often difficult due to biological vulnerabilities to chemotherapy, radiotherapy, and anaesthesia on the one hand, and psychological limitations that reduce patient compliance on the other.

However, it is still possible and generally requires the accompaniment of a caregiver. There is very little data on treatments and their results, probably due to the reduced life expectancy of these people in the past.

In conclusion, it is very important that people with a polyhandicap benefit from the same screening as that performed for the general population for breast and colon cancer, and be subject to targeted surveillance measures for the oesophagus, stomach, bladder, and testicles because of their specific risk. Experience with cancer treatment and treatment outcomes are currently very limited.

3 Follow-up of a person with a polyhandicap

3.1 Everyday life

In everyday life, the most highly qualified people for the supervision and care of a person with a polyhandicap are those who take care of them every day. The doctor can only make the right decisions if he or she listens to those who carry out the acts of daily life, whether family, friends, or professionals. Observation of the person's behaviour is of great importance to trigger active investigations as soon as the clinical condition changes.

3.1.1 Everyday activities

- **On the social front**

Daily life is an opportunity for the person with a polyhandicap to meet with peers or caregivers. It is also a way to experience the necessary separation from parents. The institution is also a way to experience the necessary separation from parents. Signs of affinity between two people are to be taken into consideration by the carers, in order to help them experience social interactions. Everyday life also provides each person multiple opportunities to express life choices: personal environment, relational preferences, etc.

Social life is sometimes a source of psychological suffering, such as during parting, and this can be expressed classically, with sadness, or through behavioural disorders (crying, tears, and stereotypes that can go as far as self-mutilation), as well as somatically: this should be considered if there are epileptic or digestive manifestations or frequent and unexplained intercurrent infections.

- **Bathing**

Bathing is a source of well-being and must be conducted by those who are experienced. The position of the polyhandicapped person must allow for gentle and respectful handling, with a spoken explanation of each step that reassures him or her. The person must be well positioned according to their orthopaedic constraints to avoid painful postures and the conditions must be ergonomic for the caregiver (height of the washstand, necessary objects within reach).

Every part of the body must be examined and washed and dried carefully, especially all places that are difficult to access (closed hands, sharply bent arms or knees, etc.) to avoid fungal infections or the development of bedsores. This period may be used to perform care around the gastrostomy or tracheostomy cannula. It can also be a privileged moment for the daily abdominal massage to help intestinal transit. In cases of neuropathic pain, any touch is a source of pain, and the gestures should be performed with a stronger touch. When bathing is a source of significant pain, despite the good quality of care, it is possible to offer premedication or, in institutions, to perform it under MEOPA (DVD "Time for a toilet" - Philippe Pernes).

- **Dressing**

Dressing is also a time when the person can participate and learn the gestures of daily life. Preferably the clothes are chosen with the person, favouring stretchy textiles and fine seams. They must be smooth at the end of putting them on, leaving no uncomfortable or irritating folds or creases.

- **Positioning of the person**

Positioning is the responsibility of the care givers and must be carried out with the greatest care, under conditions validated in orthopaedic, PRM, and/or occupational therapy consultation. The equipment must be optimized for the person: verticalization, chair with corset, moulded mattress, etc. Changes in position during the day are necessary. Free movement time on the floor must be organised for physical and psychological relaxation and to encourage spontaneous motor skills and discovery of the material environment.

Postural changes before or after meals are used to regulate gastric emptying and promote regular transit. Before a meal, the person remains on the left side for five minutes to evacuate air from the stomach. After the meal, the sitting position should be maintained for one to three hours (as gastric paresis is often

present), after which a nap on the right side helps gastric emptying. These people very often have gastroesophageal reflux and *a priori* all are suspected of having it. Thus, they should never be left in a flat decubitus position, but rather always maintain a certain degree of global or trunk uprightness (at least 30 to 40°).

- **Learning**

Learning must be organised through activities of discovery of the world, which aim to develop the skills of the person with a polyhandicap and increase his or her emerging capacities: understanding and recognizing places and time (Bataille 2011) and practicing the use of aids for moving about and autonomy for the essential acts of daily life. The same activities must be repeated sufficiently to lead to true cognitive acquisition.

All body awareness activities promote new learning through various means: Snoezelen (controlled multisensory environment), Fröhlich's Basal Stimulation (Rofidal 2017), music, swings, and flotation, as well as manual activities, such as pottery and painting, and finally, all adapted physical or sports activities: trekking chair racing, horseback riding therapy, swimming, etc.

These activities are also a vector for social participation and inclusion for people with a polyhandicap, in particular, outings or activities shared with other structures, regardless of what they may be (Desenfant 2010). These can include day trips (shops, museums, concerts, restaurants, etc.) or more prolonged stays in unfamiliar surroundings (winter sports, boating, etc.).

3.1.2 Emotional, intimate, and sexual life

It is common for parents and caregivers to be unsettled by or question manifestations of sexual function, for example, erections when changing diapers or using the toilet.

Between denying such manifestations of the body in the name of a taboo (in the past) or by interpreting any manifestation as a sexual need (today), the sexual rights proclaimed by the IPPF (International Planned Parenthood Federation[3]) are those of everyone, whether or not they live with a polyhandicap.

Between minimization and dramatization, we should value:

- **Gender identity... to replace the disabled identity alone**

We should avoid infantilizing the pubescent and then adult body as much as possible and instead make them feel their boy/girl and then young man/young woman, man/woman identites. Be creative in the choice of clothes. Dare to go as far as beauty care. Call them by their real first name instead of the eternal nicknames. Name the "private" parts of their body, even if their meaning escapes them; words humanize the relationship.

- **Modesty instead of exposing the intimate parts of the body**

In the bathroom, from gestures adapted to the existing modesty to unintentional immodesty due to a highly dependent body, the dignity of the person is at stake, despite his/her possible indifference in this respect. We should devote ourselves entirely and exclusively to the person during their intimate care in the face of his or her nudity. Do not allow third parties to enter, speak, or look.

- **The sensual body instead of solely the imprisoned body.**

Any manifestation of the intimate body should not be interpreted, from afar, as a sign of "sexual" need! Nevertheless, some people will need to manipulate their genitals in spite of the indispensable urinary protection. Do not tighten it too much, so the hand can be inserted, keeping in mind that the hand will later have to be removed and the protection retightened. Sometimes it may even be necessary to guide the hand into the opening, because certain rigidities or spastic movements do not allow a spontaneous autonomous gesture, but without ever providing direct help for masturbation! On the other hand, provide them time for such intimate contact with themselves outside of the collective or family time, taking care, however not to forget them... they won't call us!

[3] Sexual Rights Website : planning-familial.org

- **Emotional touching between peers instead of just touching the armrests of the chair**

Why not sometimes bring them closer together so that they can briefly experience the warmth and texture of the skin of others in their restricted lives as well? Through such modest body-to-body contact (without nudity or sexual contact) the relationship can be expressed between peers, as they lack the ability to speak, while assisting them closely so that their often clumsy, even unexpected, gestures do not cause physical discomfort or harm.

- **Tenderness in institutions instead of the taught "right distance"**

Their strong need for attachment is vital (and far removed from romantic attachment) and is nourished by the "right closeness", without infantilizing, imprisoning, or exciting. Unconditional love dictates that "Every desire to love is also a desire to be loved"[4]. This is an emotional impulse that must be exchanged between them and us.

- **In short, between denial of the sexualized body and hypersexualization...**

We should allow our beliefs to be challenged and accept the uncertainty of confrontation[5]. The work *they* and *we are* asked to perform often requires imagination, creativity, and certainly a great deal of attention for the other. This latter aspect must be the subject of open discussion within any care/educational/therapeutic team, in light of the sexuality of the polyhandicapped person in the hands of so many accompanying third parties. It is essential to consult with each other and critically analyse our various personal beliefs, without however increasing the propensity to doubt or falling into triviality or, conversely, a disproportionate zeal.

Finally, a high level of dependency leads to a particularly high level of vulnerability to *sexual abuse*; the sexuality of all the protagonists comes into play in the interactions of daily care.

Training courses must not neglect this aspect: this delicate subject must be addressed and repeated for professionals and should be discussed with families. Indeed, by definition, the person with a polyhandicap will not be able to express himself/herself in this respect. It is best if professionals and parents try to determine how to respond in the most appropriate way and not by solitude and silence.

3.2 Modalities of access to care

People with disabilities are dependent on the ordinary healthcare system for all their care. However, they encounter difficulties at several levels of the care path: difficulties of physical access to certain places, communication difficulties, insufficient attention given to the patient's entourage and their own expertise, and problems of coordination between professionals in different sectors (health, medical, and social). There are many areas for improvement, particularly in the field of coordinating care, to avoid situations of loss to follow-up, with particular vulnerability among adults with a polyhandicap (HAS 2017).

The person with a polyhandicap rarely expresses a desire for care. The role of the family or professional entourage is to help him/her achieve this to the best of his/her understanding and expressive abilities. To do so, they must be heard, as stipulated by the Act of March 4, 2002, whether as legal representatives for the parents of a minor or as guardians or relatives for the entourage of an adult. Professional carers often have a role to play if they know the person on a daily basis and can help hospital staff to decipher the requests of the polyhandicapped patient.

[4] Quote from Jacques LACAN, from a psychology course in Curative Pedagogy, Switzerland.

[5] From the Heart to the Body Programme, Swiss Prize for Curative and Specialised Pedagogy, intended for educators, carers, teachers, therapists, parents, principals and their assistants. Interactive training, based on life situations encountered in daily institutional and family life, including three levels of in-depth study. Contact: https://www.catherineagthe.ch

3.2.1 Long and multidisciplinary consultations

- **Interest of dedicated consultation systems**

Such systems are gradually being established on a nationwide basis under the initiative of the DGOS (Direction Générale de l'Offre de Soins) (Instruction 2015) by the ARS (Agence Regional de Santé). They aim to facilitate and organise the care path for people whose disability makes it too difficult for them to seek care under the usual conditions and makes it possible to meet the needs not covered for routine somatic care not related to their disability. Their operation requires coordination with the upstream and downstream aspects of both health and medico-social care and coordinated multi-professional organisation within an adapted framework. The system can involve mobile teams, organise consultations in the medico-social sector, and use telemedicine technology.

Dedicated consultation facilities receive financial aid: adaptation of certain types of consultations and financing of the required time for medical and paramedical coordination. They are subject to evaluation as a part of quality assurance/quality control. The aim is to provide a summary of the state of health of the polyhandicapped individual, direct them to the correct professionals, and coordinate follow-up.

The visibility of these measures for families and professionals still needs improvement and is provided by appropriate guides, some of which are published by local authorities (Seine Saint Denis 2015).

- **Points of vigilance**

Particular vigilance is expected for the following organizational points:

- Preparation of the consultation beforehand by involving the carers, which may include pre-visits (visit of the premises, meeting with professionals, use of equipment)
- Reduction of the waiting time (anticipation of administrative formalities, directly visiting without going through the waiting room, etc.)
- Involvement of the family or professional caregiver in the consultation (a common approach in paediatrics, still insufficient in the adult sector)
- Use of adapted communication: pictograms, "health comic book", etc.
- Integration of long consultations
- Organization of multidisciplinary consultations
- Organisation of care and check-ups during day hospitalisation. Easy access to technical platforms
- Consultations and care accompanied by the nurse coordinating the system: avoids waiting, ensures continuity
- Use of suitable premises and equipment: a more serene atmosphere
- Use of MEOPA for certain care, including dental care
- Training of hospital staff in the reception of people with a polyhandicap.

3.2.2 Screening and prevention of chronic diseases, minimum annual check-up

The person with a polyhandicap must benefit from screening and the prevention of complications specific to his or her condition, as well as from the usual chronic pathologies that are screened and treated for the general population.

- **Vaccination**

Vaccination, according to the vaccination schedule, is recommended to prevent diseases that could be very serious in these children and then fragile adults. Epilepsy is not a contraindication to vaccination (including pertussis vaccination). Particular attention should be paid to the management of post-vaccination fever. In cases of severe uncontrolled epilepsy, the recommendations of the neurologist or paediatric neurologist should be followed. Influenza and pneumococcal vaccinations are strongly recommended.

- Sensory screening (visual and auditory) should be conducted early by practitioners who can manage this type of patient. Such screening must be renewed if there are any changes in behaviour. The aim is to better understand their communication capacity and thus adapt the treatment. It is also important to avoid over-disability whenever possible.

- A hearing assessment should be performed during childhood and repeated as necessary in adulthood to account for any potential evolution of the impairment or to detect hearing impairment that starts later.

- In terms of vision, monitoring and screening for neuro-visual and ophthalmologic disorders should be performed early and renewed based on the child's disorders. It must be repeated in adults to detect complications (worsening of vision, glaucoma, etc.).

- **Basic clinical condition**

In view of the often particular homeostasis of people with a polyhandicap, good knowledge of the patient's basic clinical condition through repeated measurements in children of temperature, heart rate, blood pressure, and oxygen saturation is necessary. It is on the basis of these values that it is possible to better detect intercurrent pathologies, particularly infections. Screening for hypertension should be offered as in the general population, by adapting the circumference of the cuff.

- **Digestive tract**

- Oral follow-up is closer than in the general population: every six months for children, preferably every year for adults. Screening and management of orthodontic and dental disorders (tartar / caries) are important in terms of diet, comfort, and prevention of infectious complications.
- Screening and management of digestive disorders, particularly constipation and GERD, are recommended during childhood and should continue in adulthood. Specific follow-up is necessary for patients with a gastrostomy, twice a year for children and annually for adults. Screening for digestive cancers should be discussed for adults according to age, with particular attention to cancer of the oesophagus, which is favoured by chronic reflux.
- Screening and management of feeding and swallowing difficulties are multidisciplinary (speech therapist/ENT/etc.) and may be necessary from childhood. Regular reassessment in adults is necessary, as difficulties increase with age.
- Nutritional status monitoring is systematic and should be reassessed in the event of weight loss or excessive weight gain but should take into consideration the individual's individual state of equilibrium.

- **Endocrine monitoring**

- In terms of endocrine function, the types of investigation and frequency of the examinations are guided by the clinical signs. In childhood, pubertal disorders, hypogonadism, and ovarian failure should be monitored. Thyroid disorders should be screened at the first sign of any doubt, as clinical signs are difficult to individualize in the polyhandicapped individual.
- The search for diabetes in children or adults must be performed if there is the slightest doubt by a blood test, because the clinic signs are misleading, and must be systematic if there is a family history or unexplained worsening of their state of health. The search for dyslipidaemia can be performed as early as 20 years of age, as in the general population.

- **Gynaecological and urological follow-up**

- Gynaecological follow-up is oriented by hormonal problems (pubertal anomalies, menstrual disorders) during adolescence. The gynaecological consultation must be easy to request and performed by an experienced gynaecologist who should carry out a gynaecological examination adapted to teenage girls. In adulthood, screening for female cancers should be performed. For breast cancer, the examinations are adapted according to the difficulties of their realization (palpation / ultrasound / mammography, if it is possible).

- Vesico-sphincter examinations should be directed by the clinical signs. An up-to-date voiding record, with a search for post-void residue, for children and adults allows us to check for correct emptying of the bladder (bladder scan). A urine test strip or urinalysis should be performed at the slightest doubt and when there is a change in urinary or general behaviour. An ultrasound scan in children can confirm the normal state of the urinary tract and in adults it can detect asymptomatic vesico-ureteral reflux or lithiasis.

- **Neurological and psychiatric follow-up**

- Psychological and psychiatric follow-up is recommended from childhood onwards, particularly when there are mood or behavioural disorders.
- Neurological follow-up begins in childhood and continues to adulthood, depending on clinical signs (epilepsy, abnormal movements, etc.). An electroencephalogram should be requested if there is a suspicion of epilepsy and renewed according to the evolution of the epilepsy and the treatments.
- Brain imaging is routine in children for diagnosis and is age appropriate. In adults, reference brain imaging is desirable (sometimes simple CT scans), especially if there is no access to child imaging or if it was performed very early in life. Such imaging should be discussed according to the constraint/benefit ratio. In cases of a brain shunt, X-ray imaging of the valve and brain CT imaging should be adapted to the age and clinical signs.

- **Biological monitoring**

- An annual blood test is recommended for children and adults to detect complications of drug treatments, anaemia, vitamin deficiencies, endocrine and ionic disorders, etc.
- Screening for osteoporosis can be performed from early childhood, depending on the clinical signs and orthopaedic constraints, with follow-up adapted for age and possible treatments. Bone densitometry is of great interest in this context. A search for fractures should be carried out by a targeted radiological check-up at the slightest doubt, regardless of the age, particularly for vertebral fractures.

- **Other follow-up**

- A respiratory check-up is systematic from childhood and is adapted to the clinic signs, with an annual reassessment.
- It is advisable to screen children and adults for classic ENT complications (ear infections, sinusitis, etc.).
- An orthopaedic check-up, with X-ray imaging of the skeleton, should be carried out from childhood, particularly of the spine and pelvis. Ideally, the PRM follow-up is at least annual for children and its frequency for adults depends on their clinic state (rehabilitation / equipment / facilities / surgical discussions, etc.).
- Screening for sleep disorders and nocturnal ventilatory disorders depends on the clinic signs, including in children.
- Follow-up for the skin depends on the clinic signs. Any alteration of the skin or scalp must be diagnosed (bedsores, macerations, mycoses, lice, etc.).

The medical follow-up model, to be adapted to the polyhandicapped patient, is specified in the table below.

- Follow-up model: Polyhandicap care pathway (Source: Dr. Celia Rech)

Polyhandicap care pathway - Follow-up proposal to be adapted to the patient						
Items	**Clinical function**	**3 months**	**6 months**	**1 year**	**3/ 5 years**	**Details**
General						
Weight-Height		**X**		**X**		**Weight-height -> end of growth curve**
BMI		**X**				**Interest in longitudinal monitoring and nutritional assessment**
Cranial perimeter child				**X**		**More at the beginning of life**
Base temperature / HR / BP / SpO2	**X**	**X**				**Knowledge of the basic condition of the child or adult**
Ophthalmological follow-up						
Visual check-up					**X**	
Hearing monitoring						
Hearing check-up for children					**X**	**Clinical examination, auditory evoked potentials**
Adult hearing assessment	**x**					**Subjective audiometry**
Oral Follow-up						
Child			**x**			**Clinical assessment / care in an adapted environment sometimes under anaesthesia +/- imaging**
Adult				**x**		
Gastroenterology follow-up						
Gastroesophageal reflux disease				**X**		**Possibly pHmetry and/or fibroscopy if signs of gastroesophageal reflux. Screening for Helicobacter Pylori if upper gastrointestinal symptoms are resistant to treatment (PPI).**
Constipation				**X**		**Clinical follow-up / abdomen X-ray as needed / other**
Gastrostomy follow-up			**X**			**Adaptation of the nutrition / Change of button / biology: albumin, prealbumin**
Gynaecological Follow-up						
Gynaecological check-up	**X**				**x**	**At puberty and screening for women's cancers**
Endocrine Monitoring						
Screening for endocrine disorders	**X**					**Thyroid disorders / ante-pituitary insufficiency / hypogonadism / ovarian insufficiency / pubertal disorders**
Vesico-sphincter follow-up						
Vesico-sphincter assessment				**X**		**Urination record with bladder scan, urinalysis, +/- test strip**
					X	**Bladder-renal ultrasound (neurological bladder resonance) every 3-5 years**
	X					**Urodynamic assessment**
Aetiology and genetics						
Child	**X**					**Initial balance sheet**
If no aetiology					**X**	**Rethink the question of aetiology and if the parents / siblings want to get pregnant.**

Items	Clinical function	3 months	6 months	1 year	3/ 5 years	Details
Swallowing disorders						
Screening	**X**					**Clinic / ENT advice / Speech and language therapist / Specific exams**
Neurological follow-up						
Diagnosis, evaluation, global management	**X**					**Neuro-paediatric and then neurological follow-up at least once a year for children and then depending on the clinical signs.**
Epilepsy follow-up				**X**		**EEG / Sleep EEG: first seizure and seizure changes**
Child brain Imaging	**X**					**Diagnosis or scalability**
Adult brain Imaging	**X**					**At least one and in case of neurological changes**
Brain shunt imaging	**X**					**Valve and brain reference imaging and shunt tracking**
Psychological and Psychiatric Follow-up						
Psychologist/psychiatrist consultation	**X**					**To be adapted to the needs and include close relatives, siblings, caregivers, etc.**
Follow-up in Physical Medicine and Rehabilitation (PRM)						
Paediatric PRM assessment			**X**	**X**		
Adult PRM assessment	**X**			**X**		**Rhythm of follow-up to be adapted / adult**
Skeletal X-ray	**X**					**Depends on clinical signs, age, growth - According to PRM / orthopaedist opinion**
Large children's equipment				**X**		
Large adult equipment					**X**	
Wheelchair				**X**	**X**	**Depending on age and clinical state**
Orthopaedic shoes				**X**		
Follow-up orthopaedic surgery						
Paediatric and adult orthopaedic follow-up	**X**					**In collaboration with PRM - spine and limb monitoring**
Osteoporosis follow-up						
Osteodensitometry - specific biological assessment	**X**				**X**	**Clinic sign- and treatment-based screening and monitoring**
Bone imaging X-ray	**X**					**At the slightest sign of fracture**
Biological follow-up						
Biological assessment				**X**		**CBC, ferritin, albumin, prealbumin, blood ions, creatine urea, 25 OH vitamin D, calcium, phosphorus, alkaline phosphatase, magnesium. In case of macrocytic anaemia or severe undernutrition: vitamin B12, folates**
Specific biological assessment	**X**					**Follow-up of the side effects of neuroleptic, antiepileptic, and antispastic treatments (hepatic, pancreatic, lipid, glycaemic, and prolactin tests). Asthenia assessment - measurement of antiepileptics**
Respiratory Monitoring						
Adaptation of respiratory management				**X**		**Respiratory check-up - imaging - blood test - capnia - oxymetry**
Cardiological Follow-up						
ECG, cardiac ultrasound				**X**		**Depending on the clinic and aetiology**
Sleep disorders						
Screening for Sleep and nocturnal ventilatory disorders	**X**					**Sleep clinic/agenda/actimetry/polysomnography/oximetry/transcutaneous nocturnal capnometry**
Communication disorders Cognitive functions					**X X**	**As required**

3.2.3 Conditions for carrying out paraclinical examinations

The conditions for carrying out paraclinical investigations must be systematically evaluated, as well as the benefit/risk balance, depending on the nature and justification of the examinations envisaged. The family or close relatives may be involved whenever possible to facilitate the performance of the examinations.

Examinations that may lead to a change in management or symptomatic treatment should be prioritised: for example, the search for a cause of pain for which the mode of disclosure may be a challenging behaviour.

The development of "dedicated consultations" allows simplified access to technical imaging platforms: dedicated time slots, extended times, adapted premedication, presence of a family caregiver, equipment for transfers, etc. Certain examinations must be performed under general anaesthesia. The coordinated organisation of several procedures during the same anaesthesia is beginning to emerge. This makes it possible, for example, to combine a digestive fibroscopy and a dental care consultation.

3.2.4 Telemedicine and new technologies

The objective of telemedicine for people with a polyhandicap is to facilitate regular follow-up to prevent, as far as possible, the occurrence of complications that endanger their health, which often lead to repeated and/or prolonged hospitalisation. This is one of the main thrusts of the five-year strategy for the development of the medico-social offer for people with a polyhandicap.

An evaluation carried out in 2014 by Dr. Hully, Pr Billette de Villemeur, and the CESAP of nine different centres showed that 18% of these patients had no neuro-paediatric follow-up, and among the children with a referring neuro-paediatrician, 40% had had no consultation with their referring physician during the previous year, a paradoxical situation for a so-called fragile and at-risk population.

The French Eval-PLH cohort study coordinated by Pr Billette de VIllemeur recruited 875 patients divided by age (children and adults up to 25 years of age, N = 545, and aging adults, N = 474) in a neuro-paediatric hospital service (Hôpital Trousseau), a CESAP entity representing the ambulatory medico-social/medico-educational institutes and five services dedicated to polyhandicapped individuals, including four AP-HP dependents. Of the 875 patients, 400 were ambulatory or medico-educational institute patients (Rousseau 2018). This confirms the significant medical needs of these patients and the burden of their care, which increases with age.

Telemedicine is a new medical activity with an ecological dimension of care, in the sense that the person is seen in his or her usual environment and the opinion of local health professionals is shared with parents and expert doctors. This limits the use of expensive and exhausting medical transport for the person, and contributes to the continuous training of the local healthcare team. These teleconsultations do not replace a traditional consultation but constitute a new medical activity for the benefit of the person and the local services that follow him or her.

A trial of neuro-paediatric teleconsultation for children with a polyhandicap (polyhandicap MTCT) was conducted between 2015 and 2018 (co-piloted by the ESCAP and AP-HP). During such teleconsultation, the partners communicate directly by videoconference on a secure computer network, allowing them to meet and collectively propose a personalized and adapted care plan for each child (Billette de Villemeur 2017).

Among 118 teleconsultations carried out at the two pilot sites, the reasons for consultation were systematic follow-up (35.5%), management of epilepsy (48.3%), behavioural disorders (11.8%), a problem of nutrition, sleep, or pain, or preparation for surgery or scheduled hospitalisation. Thus, 70% of the teleconsultations resulted in a therapeutic adjustment by prescription.

Support sites for the implementation of these new technologies are beginning to emerge and help to develop specific responses to polyhandicap.

3.2.5 Hospitalisations: precautions to be taken

People with a polyhandicap are all likely to be hospitalized, so it is important to be prepared. Each person with polyhandicap (through family or professional caregivers) must have his or her own computerized medical record (shared medical record, health passport, or specific health record).

Professional secrecy is shared within the care team, in accordance with the Health Law of January 26, 2016, and therefore includes all professionals working with the person on a daily basis, at home or in a medical-social establishment.

Orientation tools, such as Via Trajectoire software, must be known to caregivers and medical and social professionals to quickly find the establishment that can accommodate the person in question in an adapted manner.

- **Encourage scheduled hospitalisations**

This improves the quality of care:

- If possible, organize direct entries for already known/fragile patients
- Establish the "adapted equipment list" with the caregiver before hospitalisation
- Check with the caregiver whether 24-hour assistance is required for a single room
- Prepare for the accompaniment of the entire family (ex: other children at home)
- Use day hospitalisation or, if possible, delay entry for 24 to 48 hours to admit the patient under the best possible conditions
- Promote home hospitalisation (HH) whenever possible to avoid uncomfortable or dangerous transport for the person
- Use mobile care teams whenever possible as an alternative.

- **Conditions to be respected**

For all health establishments, the reception of disabled individuals and, in particular those with a polyhandicap, requires compliance with the HAS recommendations published in 2017 to:

- Ensure continuity of care between the upstream and downstream stages of hospitalisation
- Ensure accessibility in the broadest sense
- Promote and formalize the role of the caregiver
- Change the view of such patients by hospital staff and develop skills internally
- Unite the teams around a project of change

There are two tools of particular interest for healthcare institutions that aim to improve the quality of care for people with a polyhandicap within the institution: the *checklist of* tools for the management team and the patient-tracer grid.

Healthcare institutions must also enable all staff in health or medico-social institutions to receive training in the assisted living relationship, so that they are ready to welcome the patients, detect risks, and react collectively to stop situations of abuse if they are detected at any point in the healthcare journey of the polyhandicapped person (Piveteau 2019 guidance note).

- **Emergency hospitalisation**

In the event of an emergency, healthcare institutions must be prepared to respond appropriately:

- Contact referents or "Disability" teams within the hospital
- Have a *checklist* available in the emergency department of what needs to be evaluated or rapidly implemented when you receive a person with a polyhandicap (see experience Handisanté 13) to:
  - Immediately place the patient on a bed with an anti-decubitus mattress
  - Welcome the caregiver and allow him or her to stay with the polyhandicapped person on a continuous basis
  - Assess pain and relieve it
  - Establish a Braden score (used to assess the risk of pressure ulcers)
  - Consider adding albuminemia, pre-albuminemia, PCR/CRP (C-reactive protein) in the entry blood workup if there is any doubt about nutritional status.

3.3 Specific recommendations

3.3.1 Respect the state of equilibrium

The homeostasis of people with a polyhandicap is sometimes outside the usual norms. The most frequent unusual situations are vital constants in the basal state (at rest or during sleep) that are outside the norms for age. These may include a temperature below 36°C, low blood pressure, and a very slow pulse. It is also common for blood oxygen saturation to be below 90 or even 85%. If these vital constants are regularly found in the same polyhandicapped person with the same values outside of the norm, apart from decompensation or intercurrent illness, they must be accepted as the person's state of equilibrium.

It is also common for height and weight to be much less than normal for age or for weight to be much less than expected for height and age. It is always necessary to ensure that such unusual values are not related to severe undernutrition, especially for weight, and to check for biologicalweight markers of undernutrition. For each person, it is important to define their « healthy weight », which should become the person's reference weight.

It is very important to not try to correct such equilibrium values for a given child by seeking treatments to bring them back to the accepted norm. Indeed, at best these attempts are ineffective and the child returns to his or her state of equilibrium as soon as treatment is stopped, or their values do not change in spite of treatment. However, attempts to correct such abnormal values in relation to the reference values, sometimes through aggressive treatment, may lead to severe deterioration of the child or even put him/her at risk of death.

It should therefore be remembered that disease is sometimes a new homeostatic equilibrium and that it is dangerous to try to correct it in an untimely manner, which Canguilhem defined well in his medical thesis in 1943 and which is still perfectly valid today (Canguilhem Georges, 2013).

3.3.2 The transition from childhood to adulthood

The transition from child to adult is an important passage for physical, psychological, and organizational reasons. Puberty upsets the balances patiently built up during childhood. The necessary changes within the entourage and in living arrangements can be brutal (place in a MAS (Specialised Welcome House) to be taken in the coming month or week, etc.) and often irreversible. Medical coordination is sometimes difficult to organize in the "Adult" sector.

The SOFMER recommendation published in 2012[6] and approved by the HAS provides some food for thought (see schematic presentation at the beginning of the recommendation):

- If possible, make medical transitions during periods of stability
- Organize consultations in paediatrician - adult doctor pairs
- Find the attending "Adult" physician before discharge from the "Children's" facility and determine his or her role in coordinating care
- Develop tools to aide support within adolescent structures
- Define a "transition programme" that is communicated to and understood by the young person, his or her parents, and the health or medico-social institutions concerned
- Develop health education for a better knowledge and understanding of the body by the adolescent and then the young adult
- Prepare for administrative changes and inform parents and the youth, especially about guardianship, before the age of 18 years
- Provide parents of teenagers with discussion groups that focus on this transition.

[6] SOFMER (French Society of Physical medicine and Rehabilitation), Motor handicaps and related: the transition from child to adult. Medical and medico-social stakes in the 15-25-year period: "Recommendations for Clinical Practice with public debate", July 2012.

3.4 Therapeutic education and the promotion of health

Therapeutic education for patients with a polyhandicap is aimed at parents and caregivers who live with the polyhandicapped person.
The main objectives of therapeutic education programs are:

- Train parents and caregivers in specialized care techniques that are necessary in daily life (e.g., nasopharyngeal suctioning techniques, tracheostomy care, pressure ulcer prevention, oral and gastrostomy feeding management, prevention of miscarriages, and analysis and management of behaviour indicating pain).
- Train caregivers in the identification and management of emergency situations (e.g., respiratory aspiration, states of acute agitation) and thus improve the safety of patients in a situation of polyhandicap.

Various therapeutic education programs have been developed in hospitals (short or medium stay). They are personalised and adapted to each patient, through an educational contract established with the parents/caregivers.

Therapeutic education sessions are organized in groups to promote the exchange of experiences among caregivers, or individually, particularly when preparing with families for a return home after a prolonged hospital stay (e.g., when changing feeding tubes or tracheotomies, emergency treatment of a prolonged epileptic seizure, or adaptation of oxygen therapy).

The therapeutic education is evaluated during physical consultations and then increasingly during telemedicine consultations, allowing close interactions with the staff of the medical-social centres where the patients with a polyhandicap live (Gaudon P., GPF, 2015).

3.5 Iatrogenies

A disorder or disease is iatrogenic if it is caused not only by drug therapy but also by any medical act, even in the absence of medical error.

3.5.1 Drug Iatrogeny

- **Polymedication**

Because of his or her multiple pathologies, the polyhandicapped person is most often poly-medicated. During a study carried out at the Antoine de St Exupéry Centre in Vendin le Vieil (62) on 87 cases of people with a polyhandicap, both children and adults, we found that 12 therapeutic classes used directly for the treatment of the pathologies encountered in the context of a polyhandicap were very frequently used in basic treatment (excluding antiepileptic drugs, which were studied elsewhere and not classified in order of the frequency of prescriptions).

Table 1 summarizes the MA (marketing authorization), precautions for use, and main side effects observed for each class *(Annex 4, cf. in connection with Chapter 3.5, Table 1)*.

- **Use of medicines outside marketing authorization**

Certain medicines are used without marketing authorization in children, either without a therapeutic indication (clonidine) or without an age indication (therapy not recommended in this age group or galenic form not appropriate before the age of 6 years. For example, tetrabenazine, used in dystonia, is not recommended before the age of 18, although its benefit is undeniable. Similarly, bisphosphonates do not have marketing authorization for children, but their use is subject to a proper prescription.

Certain antiepileptic drugs only have market authorization for adults (pregabalin) or children after 16 (lacosamide) or 12 years of age (perampanel, gabitril, gabapentin as first-line mono therapy).

- **Doses used are sometimes high**

The doses used for certain antidystonics, antiepileptics, and PPIs can be high.

- **Possible medication errors**

Errors in administration are possible due to polymedication and galenic forms that are not adapted to children. Adverse reactions and precautions for the use of these medicines are summarised in Table 2 *(Annex 4, see in connection with Chapter 3.5, Table 2).* It should be kept in mind that each drug has its own side effects and that their combined use will increase such deleterious effects, particularly in terms of the level of alertness.

Any prescription will have to account for the person's overall medical context, the risk/benefit balance, possible side effects, and compatibility with existing therapies. The treatment should be re-evaluated on a regular basis to avoid continuing unnecessary medication or medication with excessive side effects. One should also try to simplify the treatment (e.g. antiepileptics).

Certain side effects that are particularly disturbing for the comfort of the person's daily life should be systematically monitored: constipation, urinary retention, dry mouth, hypersalivation, bronchial congestion, hypocalcaemia, and flu syndrome *(Appendix 4, cf. in connection with Chapter 3.5, Table 2)*. Attention should also be paid to the timing of administration, especially at night.

3.5.2 Iatrogeny related to medical procedures

Any medical act must take into account the person as a whole, the goal sought, the possible harmful effects, its repercussions on the person's daily life, comfort, and quality of life, and the informed opinion of parents or legal guardians.

This applies in particular to certain daily procedures (correct intermittent urinary catheterisation, for example) and indications for surgery, which must be performed by a multidisciplinary team: a correctly centred hip after surgery can remain or become painful.

4 Life path and support

4.1 Role of the person and his or her family

4.1.1 Role of parents

For a child with a polyhandicap, as for any child, parents have parental authority until the age of adulthood and must guarantee their safety, accommodation, health, and education. The needs of a child with a polyhandicap are such that parents will face many demands over the years to ensure that care is provided. All situations can be encountered, from those in which parents assume almost all the care of their child, to delegating it to a health or medico-social institution. Every effort must be made to ensure that their quality of life is compromised as little as possible (Rousseau 2019).

- **Information and support for families**

Informing parents and siblings throughout the child's life journey remains a legal obligation for professionals, as for any child. The explanations to be given about a child with a polyhandicap are more complex and require frequent contact, and each step must be thought out for the child's greater well-being. Of course, maintaining a relationship between parents and a child with a polyhandicap who has become an adult is beneficial as long as it is possible, even if most of their life takes place outside the parents' home.

The process of informing and accompanying parents cannot be the subject of a generic protocol, as there are many situations: during pregnancy, during an acute episode, after a difficult delivery, etc. Nothing can be systematized. News that can generate anxiety, even anguish, is frequent and diverse in the life of a child with a polyhandicap and each parent has his or her own way of "hearing" it and then understanding it and taking responsibility for the choices that determine the future of their child. Parents and siblings must be supported, as recommended in DHOS/DGS/DGAS circular No. 2002-239 of April 18, 2002.

On the other hand, the doctor's constant concern must be to accompany the family, first by the local staff, who must be informed if painful news has been delivered, and by the psychologists available in the hospital department or medico-social establishment. Such accompaniment must continue if the child is at home and the modalities must be devised with the parents as and when necessary.

Parents should remain the ones who make life-changing decisions in these processes. Each communicates by his questions a part of the truth that he understands at the moment and the accompaniment takes on its full meaning when the professional rephrases what has just been said, using his professional language, which allows the parents to understand better, and thus with each back and forth, the explanations are adapted and become more complete (Juzeau, Husse, 2010).

Parents are often waiting for a prognosis. However, even with experience, no doctor can venture to define what will happen to the polyhandicapped child (Tezenas du Montcel, 2017). Finally, what is important for parents is to have someone to call or talk to in case of a hard blow or a confrontation with the "violence of appearances" (Zucman, 1998).

The most important thing is to give the child the life of a child and to insist on this with the parents so that they can give their child the love and happiness that they are capable of giving. Give the family perspectives in a systematic way, in the way things are said about leisure, 'schooling', family life, etc. (Juzeau, Husse, 2010).

- **Siblings**

There are two scenarios: either the polyhandicapped child is the eldest, in which case the situation is already known before the birth of the other children, or he arrives after other children, and the earthquake that the parents always talk about is then felt all the siblings.

The parents try as often as possible to lead "as normal a life as possible" for the sake of the other children, knowing full well that each child will have to adapt more than in other families, and that guilt over the siblings is always on the parents' minds. "There is no single choice or one that is absolutely better than another. "(Tezenas du Montcel, 2017).

The information given to other children must also be as clear, appropriate, and guilt-reducing as possible, especially for the youngest. It should be reviewed whenever necessary during the asking of questions, which is always the best source for providing the right answers. The right answer varies according to personality, those of the parents and that of each child, and brothers and sisters tend to hide their own difficulties in trying to protect their parents.

A word on friendships, of which many are lost, but many are also created through hardship, as well as within the extended family: parents and professionals also know well how strong friendships are born from the child's particular situation, through hospitalisations, association meetings, etc. (Tezenas du Montcel, 2017).

- **Aging parents**

As the lifespan of people with a polyhandicap increases, parents may find it increasingly difficult to cope with the maintenance of the parental bond or even die before their child. This situation poses new problems for both the polyhandicapped individual and his or her parents (Desenfant, 2010).

When the person is cared for in an establishment, the routines of visits and reception at the parents' home (weekends, holidays) change or are sometimes broken. This emotional imbalance is detrimental to the complex and fragile balance that has been built up over time for the person on the one hand and the parents on the other. The parents, already feeling guilty about having a polyhandicapped child, suffer even more as time passes by, as they are less able to care for their child and fear that they will not outlive him or her.

When parents provide legal protection, their own aging makes them decreasingly operational. Passing the baton to another family member is a logical development but it may be impossible or complex because family ties are not as strong as between mother/father and child. In such cases, it is sometimes another proxy, without any initial emotional ties, who will have to take over their protection.

4.1.2 The person with a polyhandicap himself or herself

- **The polyhandicapped child**

From early childhood, children learn much from their parents, both through imitation and through constant exchange and communication. Little by little, they must learn to manage frustration, like any child, which is probably more intense than for other children, and varies according to the quality of life of the parents, who send them positive or negative non-verbal and verbal messages.

Inclusion in a community, either day-care or school, is a source of enrichment for the child, as encounters with peers lead to new experiences and learning about life and knowledge. Such inclusion provides an opportunity to develop the sensory-motor awakening but also the cognitive capacities of the child, enabling him/her to learn how to make adults discover them by all the means at their disposal. For many years, the inclusion of young children with disabilities in early childhood facilities has been the subject of studies that have shown all the positive aspects for the child and his or her parents of such type of care (C. Zaouche Gaudron 2016).

It should be remembered that the right to education is also enshrined both in the Convention on the Rights of the Child and in the law governing medico-social establishments (Act No. 2002-2 of January 2, 2002 on social and medico-social action). Many associations and institutions are working to ensure that such a right to education and learning continues to evolve, as can be seen in their writings and in the approaches under way for inclusive education in many regions of France (A. Bataille 2011). Children and adolescents with a polyhandicap can be cared for in a system with adapted schooling. The advantage of this type of care is the access to interested and trained pedagogical teams, with a smaller number of students than in ordinary

schooling. This system also makes it possible to better adapt to the child's rhythm and care, with adapted schooling.

- **The polyhandicapped adult**

Of course, the person with a polyhandicap, given all the communication difficulties described, may be far from understanding his or her situation, and involving him or her in the development of his or her life project may be a difficult undertaking. However, legislation (law 2002-2 of January, 2 2002 on social and medico-social action, decree of May 9, 2017) and our understanding of the person as a human being mandate that ways to do so be established and implemented: adaptation of communication, explanations through various channels, and appropriation by the person of his or her choices, from the most intimate to the most concrete, from the choice of room neighbours to his or her food or activity preferences, taking into account his or her emotional preferences, primary needs, and possibilities for expression.

Many institutions take note of these legal obligations and bring them to life on a daily basis, based on the progress made in the observation, knowledge, and education of polyhandicapped people (Rofidal T., 2017).

4.2 Protective measures and decision-making

4.2.1 During childhood

It is unusual for a child to be entitled to a legal measure of protection, since his or her parents are naturally his or her legal representatives. However, this is possible if both parents are deceased or subject to the withdrawal of parental authority.

Consequently, after referral to the guardianship judge, the judge sets up and chairs a family council of at least four members, chosen on the basis of the interests of the child. This council appoints from among its members a tutor and a supervising tutor responsible for supervising the management of the tutor. The family council is responsible for regulating the general conditions for the maintenance and education of the minor and deliberates by majority vote (the tutor does not vote). If no one is able to provide guardianship, it is entrusted to the services of the Department. Guardianship is then exercised without a family council or a supervised guardian.

In the event of separation of the parents, parental authority may be attributed by the judge to only one of the parents, with the other parent being deprived of parental authority, in cases in which a risk of abuse has been identified by the judge.

4.2.2 Adult guardianship

Under the Civil Code, the legal protection of adults is the duty of families and the public community. From the age of 18, the need for guardianship is therefore in principle necessary, but quite often parents continue to manage their son/daughter's affairs for several years without having taken the necessary steps for guardianship, because the procedures are long and involve distancing from the child, and sometimes, if the family situation is complex, they may be afraid of rekindling conflict.

The guardian must both respect the autonomy of the protected person and his or her will, while making decisions for him or her without risking infantilizing him or her more than necessary. This is a hybrid mission between social work and justice, which is not without ethical questions (laws of January 3, 1968 and March 5, 2007)[7].

[7] Act No. 68-5 of 3 January 1968 reforming the law on incapable adults - Consolidated version as of May 6, 2019
Act No. 2007-293 of March 5, 2007 reforming the protection of children

Moreover, even in the case of legal protection, there is often an interaction between the family (harmonious or not) and the actors of the medico-social establishments that very often take care of the person with a polyhandicap.

In practice, protection must be requested from the guardianship judge who will choose the type (empowerment of the family and often guardianship for people with a polyhandicap), its scope (economic/social), and the representative (family, proxy service, individual representative, settlement worker). Judges often entrust guardianship to the parents, who may sometimes exercise it jointly. In certain cases, guardianship is entrusted to a specific association and is exercised by professional mandataries if the judge deems it necessary.

The proxy is accountable to the guardianship judge. When the mandate is given to a family member, he or she can be assisted by the Regional Family Guardian Information and Support Service (SRISTF).

A national website is currently under construction by the Ministry of Solidarity and Health for families who are considering setting up a guardianship for their loved one. It will be operational in 2020.

4.2.3 Health decision making (operations)

Guardianship is primarily concerned with asset management and making difficult medical decisions is sometimes problematic. The guardian may also be appointed by the judge to represent the individual himself or herself if they are unable to provide informed consent.

However, the person's adherence to any medical decision must always be sought and the appropriate information must always be provided.

4.3 Polyhandicap allowances

In the case of a proven polyhandicap, the person's disability rate is over 80%, regardless of age, and therefore benefits, such as the AEEH for the child or the AAH for the adult, are automatically acquired. The obligatory threshold for the transition to the adult system is the 20th birthday: up to the age of 20, child allowances apply, whereas the adult system takes over from this birthday onwards.

4.3.1 During childhood, from 0 to 20 years old

- **Allowance for the Education of a Disabled Child (AEEH)**

In addition to the AEEH, supplemental support may be granted:

- Supplements for expenses are assessed according to the level of expenditure, with a scale derived from the implementing decrees of the 2005 law[8].
- Supplements for the reduction of parents' working hours are allocated according to the reduction linked to the disability, which may change according to the orientations, the places available in the establishment, and the family's choices, or for the hiring of a third person paid during the parents' working hours.
- The sixth supplement can only be awarded if the active presence of a third person is required night and day, the child is not in an establishment more than four half days a week, and there is a reduction in working hours or the hiring of a third person to care for the child.

[8] Legislative files - Timeline - LAW No. 2005-102 of February 11, 2005 on equal rights and opportunities, participation and citizenship of persons with disabilities and links to the implementing decrees
https://www.legifrance.gouv.fr/affichLoiPubliee.do?idDocument=JORFDOLE000017759074ype=echeancieregislature=

- **The Disability Compensation Benefit (PCH) for the child**

This benefit can only be awarded if the child is entitled to the AEEH supplement and if certain expenses are linked to the disability and significant dependency, and/or to childcare expenses at home. This is often the case for a child who is growing up.

Parents always have the choice between the option of supplements or PCH, both of which must be quantified by the CDM when applying for PCH [9].

Additional costs for the fitting out of the dwelling or vehicle constitutes Component 3 of the PCH, which can be allocated in addition to the supplement for expenses or reduction in working time.

4.3.2 Adults over 20 years of age

- **Disabled Adult Allowance (AAH)**

The 80% disability rate attributed to a person with a polyhandicap entitles them to receive an AAH if they live outside a residential facility. During a full-time stay in a specialised care home, only part of the AAH is paid into the person's bank account (and therefore managed by the guardian if the measure is pronounced). For accommodation in a residential home, whether or not it is medicalized, the AAH is paid to the person, who must pay a part of it to the establishment.

- **The Disability Compensation Benefit (PCH) for adults**

The conditions of attribution are systematically fulfilled in for polyhandicapped adults. The expenses must be quantified precisely and only the estimates are accepted. Already paid invoices are not eligible for refunds. It has 5 parts:

- Part 1: human assistance; the scales vary depending on the assistance provided by the family, a provider, or an agent
- Part 2: technical aid; only the remainder after all possible social aid is considered
- Part 3: accommodation, vehicle, and additional transport costs; only additional costs linked to disability are considered
- Part 4: specific or exceptional aid: adapted sanitary protection for incontinence, holiday stays, training in adapted alternative communication, etc.
- Part 5: animal aid; little used for the polyhandicapped person who cannot truly grasp the concept of a pet or give orders to an animal.

4.4 Organisation of the care pathway

(Annex 4 cf. in connection with Chapter 4)

4.4.1 Early childhood (0 to 6 years)

This is an extremely difficult and crucial period for parents, who take the news of a polyhandicap very seriously (Tezenas du Montcel 2015). The promise of a normal and happy parenthood collapses and the transition to the realities of a polyhandicap is painful and long while it is important for the child to build a life project adapted to his or her capacities. At best, parents must acquire the conviction that their child has abilities that need to be discovered to make the most of them and then included in in a project that is suited for them.

During this period, hospitalisations can be long and frequent, and contact and support from doctors and caregivers is important. In hospital, the paediatric resuscitation and then paediatric departments, most often in university hospitals, are on the front line.

[9] The Disability Compensation Benefit (PCH) for adults - https://www.service-public.fr/particuliers/vosdroits/F14202

Certain services of the Follow-up Care and Rehabilitation Services (SSR) have developed specific skills in the field of polyhandicap and have specialised in the long-term care of polyhandicapped individuals.

The first medico-social resource services to come into play are the Centres d'Action Médico-Sociale Précoce (CAMSP), which provide outpatient screening, diagnosis, early rehabilitation, and family guidance. In certain exceptional cases, the Centres Médico Psycho-Pédagogiques (CMPP), of which the mission is focused on mental health, in accordance with the regulations, can offer support to children and parents from the age of three years.

The Special Education and Home Care Services (SESSAD) provide multidisciplinary aide for the child at home, in early childhood structures, or at school, after notification by the MDPH.

There are only a few medico-social accommodation facilities before the age of three years, apart from hospitals and several medical nurseries, whereas after the age of three, the child can benefit from the Etablissements pour Enfants ou Adolescents Polyhandicapés (EEAP) and the Instituts Médico Educatif (IME), which can take care of these children, with or without accommodation, after notification of the MDPH.

All of these measures are free for families.

4.4.2 Childhood and adolescence (7 to 12 years old)

Medico-social services are provided by the SESSAD, and the use of aid, with or without accommodation, EEAPs, and IMEs, is fairly common. Activities are adapted to the child's condition and capacities (C. Husse 2010).

These establishments are highly committed to learning, both practical and academic, and each child is solicited according to the potential he or she develops. Some EEAPs or IMEs have adapted teaching units with specialized teachers, for generally short periods of time and in very small groups (maximum 3 to 5 children).

Inclusion in schools is under way in many schools, and experiments in adapted schooling are being attempted. A study by the INSHEA, called the Polyscol study, partially funded by the CNSA, was made public in November 2018 on the subject (Toubert-Duffort 2018). Working groups were established in 2019 at the initiative of CNSA and La Direction Générale de l'Enseignement Scolaire (DGESCO).

4.4.3 The transition from childhood to adulthood (13 to 20 years old)

There are several transitions that take place during this period, all of which constitute risks of rupture and regression from an already fragile and delicate balance. They concern medical care (see above, Chapter 3.3.2), the practical difficulty of daily care, the type of accommodation, and the awakening of sexuality.

Medical care shifts from the paediatrician, who has been involved from infancy, to the adult doctor. Daily care, washing, dressing, and transfers, which are special moments for the polyhandicapped child are often carried out by the "natural" caregivers, who are the parents. When the child becomes an adult, this care may be entrusted to third parties because of the physical fatigue they cause and the distance created between the adolescent and his parents in terms of physical and psychological closeness.

Structures for children must normally stop providing care at the age of 20, except in the Creton scheme, which allows for continued care in children's institutions (at least part-time) if no place is available for the young adult.

Structures for the reception and accommodation of adults with a polyhandicap must cope with the permanent care needs of such individuals, and therefore have a nurse permanently available, as do the

Maisons d'Accueil Spécialisées (MAS), which, over and above the difficulties of finding a place, means a new care team and a new community.

4.4.4 Adulthood

The place of residence of the adult with a polyhandicap may be the parental home, the MAS in day care or as a resident, or in temporary care. Stays may last several years or even a lifetime, which goes hand in hand with a humane environment and relatively stable activities.

Everyday life can remain a source of individual projects, exchanges, and lifelong learning for these individuals, who learn at their own pace (Chavaroche 2006).

Regardless of the techniques used, the key lies in the project that underpins them and allows the person to grow up independently at any age (Rofidal 2017).

In these institutions, support by professionals must be adapted to the needs of people with a polyhandicap, always with a personalised project that supports their actions, accompanies the development of new potential, and avoids situations of burnout for carers (Rousseau 2017).

4.4.5 The advancing years

It is a new fact and a true advance that polyhandicapped individuals reach adulthood and continue to thrive.

The desires of the polyhandicapped person, even if he or she does not speak, must be heard, and are translated into a project. Parents still have a role to play with professionals, because their cooperation sustains the well-being of the child who has become an adult (A. Gambrelle 2015).

Medical needs, beyond the initial needs, increase with age: osteo-articular problems (spine, various joints), osteoporosis, hearing, vision, and dental problems, cancer, etc. Everything is difficult: functional symptoms are poorly expressed and are very intricated, examinations are more difficult to carry out, and surgical and anaesthetic procedures are feared, as well as their aftermath. Moreover, individual decisions based on full information are not possible and are thus the responsibility of the doctor, informed by the person's entourage.

Finally, parents, who are often unfailing caregivers even though it lowers their quality of life (Rousseau 2019), also age to varying degrees. They may no longer be able to provide full or transitional accommodation, visits to the MAS, or formal or informal guardianship. It is a source of deep anxiety for parents to risk disappearing before their child. Practical, psychological, and emotional adjustments must be made in a coordinated effort with families (parents, siblings, and others) and professional representatives.

4.5 The end of life

In 2013, the National Observatory on the End of Life (ONFV) reported "an invisible, forgotten, neglected, even hidden, end of life" for disabled people living in a MAS or FAM. However, there is no justification for the end of life being treated differently from that of the general population.

The ONFV report highlighted that procedures were too often not or poorly applied, especially as there were no healthcare personnel trained in end-of-life issues in such an environment, even though mobile palliative care units exist throughout the country.

In paediatrics, a Regional Resource Team for Paediatric Palliative Care (ERRSPP) has been created in each region to which parents and institutions can turn for short or medium-term support. The mobile palliative care teams can of course also intervene for children if the regional team is too distant or unavailable.

Specificities of the end of life of the polyhandicapped

Short-term life-threatening situations are common in the polyhandicapped, but do not involve the end of life; no matter how serious they appear, they can often be resolved.

The end-of-life situation is defined when no reversibility can be envisaged. The Leonetti law of April 22, 2005 and the Claeys-Leonetti law of February 2, 2016 are[10] fully applicable. However, certain provisions cannot simply be applied, as the law only considers very few situations of disability and does not specify anything about polyhandicaps.

Concerning situations that occur very early in life, the 2016 law favours a "culture of service" that progresses with time and continues to respect the equality of opinion of everyone, parents and professionals, taking into account the frequent uncertainty of the future of a child who is only a few hours old. Early diagnosis is the key to rapid decision-making in situations in which the situation is life-threatening.

For adults or older adolescents, the appointment of a trustworthy person and the drafting of advance directives should not be completely ruled out, although it is known that comprehension and expression problems often make them very difficult to establish. However, the comprehension of such individuals is often better than the absence or difficulty of oral expression would suggest and it is therefore obligatory to provide each person with a polyhandicap information about his or her condition, adapted to his or her comprehension, as specified in the Claeys-Leonetti Act.

As the polyhandicapped person is not able to clearly express his or her will, it is up to the doctor to make the decision to limit or stop active therapeutic access (LATA) after a meeting with colleagues, as defined by the law and recorded in the person's file. The family, legal representatives, and usual caregivers must be consulted and informed of the medical decision. In the case of LATA, any physical or psychological suffering must be avoided, including sufficient sedation, even if it may accelerate death.

4.5.1 Role of the legal guardian at the end of life

- **For the child:**

Children’s legal representatives are usually the parents. However, if parental authority has been withdrawn, the legal representative is appointed by the Judge for Family Affairs (JAF).

In cases of certain difficult decisions (tracheotomy, transfusion, etc.) and disagreement by one or both parents, the JAF may be asked to withdraw parental authority from the reluctant parent in the event of a life-threatening risk or a medical technique intended to increase the child's comfort or allow effective treatment of a serious pathology. Such a withdrawal of parental authority can then be lifted by the JAF as soon as the necessary act has been carried out.

Concerning the cessation of care, the Claeys-Leonetti law applies: the doctor must inform the parents as well as possible and in the case of a collegial procedure, he is obliged to inform the legal representative of the decisions to obtain his informed consent whenever possible.

If the decisions taken go against the parents' opinion, legal recourse is possible within a few days to oppose the contested decision. However parents and doctors often work in a dialogue that allows for shared decision-making.

- **For the adult:**

For adults with a polyhandicap under guardianship, medical decisions are generally subject to information being provided by the doctor and agreement by the guardian. The professional representatives usually agree with the doctor's opinion. Relatives must nevertheless be systematically informed under the terms of the

[10] LAW No. 2005-370 of April 22, 2005 on the rights of the sick and at the end of life Law No. 2005-370 of April 22, 2005 on the rights of the sick and at the end of life - Consolidated version as of May 20, 2019

Claeys-Leonetti law of February 2, 2016, and dialogue with them must be maintained under all circumstances if they are present during hospitalisation.

If one parent is a guardian, he or she will have to provide consent for any medical decision, even if both parents share guardianship, or if one parent and another family member share guardianship, which happens quite frequently.

If a non-guardian parent objects to the decision, he or she may appeal to the Judge. He or she is then involved as a close associate in the dialogue with the doctor, but decisions are not subject to his or her agreement. In the event of a collective decision, in particular on the cessation of care, appeals are possible during the few days following the date on which the relatives are informed of the decision to stop care.

4.5.2 Arrangements for accompanying a person in an institution

If the adult lives in a medico-social structure, since the decree of September 6, 2012, the establishment may be assisted by [11]a hospitalisation at home (HAD) service, insofar as the establishment does not feel able to manage end-of-life care on its own. This project must be established in an agreement between the establishment and the HAD service. The service must help the establishment, mainly in carrying out technical procedures for which it has trained staff (palliative care, pain management, etc.) and the necessary equipment. This can only be agreed upon for a short period of time to avoid making the hospitalisation difficult by the condition of the polyhandicapped person, for example.

Agreements can also be signed with mobile palliative care teams, whose role is principally to help the institution in its thinking and relations with the person's loved ones rather than the technical gestures that the service will not carry out itself. The decision to call on such teams is part of the partnership that the establishment has set up with other common-law services under the law of January 2, 2002[12].

4.6 The role of associations and other actors

4.6.1 Information transmission and networking

Since 2012 (Billette 2012), the "landscape" of the healthcare pathway has, in the end, changed little. After a few articles calling for coordination between the various health and medico-social institutions and city practitioners, possibly within networks or other coordinated mechanisms, little legislation has been passed in this area.

The changes in the pathway suggested in the Jacob Report in 2013 (Jacob 2013) could not be sustainably implemented; the financing of health networks has been tightening year after year, because it is also not sustainable. Nevertheless, coordination is highly desired by many players and has very recently been the subject of a model applicable to any patient in need of care. Indeed, the HAS proposed a model for a personalised health coordination pathway in 2019, with instructions for use and examples (HAS 2019). This work is not specific to polyhandicap, but is rather intended to meet the needs of everyone and should make it possible to adopt good practices and set clear objectives shared by all health stakeholders under the guidance of a path representative.

Law 2016-41 of January 26, 2016 on the modernisation of the healthcare system specified that the multidisciplinary healthcare team authorises the transmission of information between all actors involved in the care of a single person, regardless of the sector (hospital, medico-social, or home). The decrees implementing this law have authorised the transmission of necessary information, including medical

[11] Decree no. 2012-1030 of September 6, 2012 relating to the intervention of home hospitalisation establishments in social and medico-social establishments with accommodation - Consolidated version as at May 20, 2019
[12] Law No. 2002-2 of January 2, 2002 renewing social and medico-social action - Consolidated version as of May 20, 2019

information, to non-healthcare professionals working with a sick person. Multiple transmission tools have emerged since 2012 and continue to improve the daily lives of people with a polyhandicap and their family caregivers.

Finally, the care project must be integrated into the life project. This has been reaffirmed both in expert opinions and in legislative or regulatory recommendations (D. Héron 2017). Under no circumstances can somatic problems prevent the implementation of a coherent life project for the person with a polyhandicap (ANESM 2017).

4.6.2 Health democracy and parent or patient associations

Associations of disabled people or their families are essential partners of reference centres, centres of excellence, and health and medico-social establishments. They play an essential role in supporting families through the information, assistance, and support they provide. The contact details of the associations are systematically given to the families, but the decision to contact an association remains the choice of the family or patient. They are a significant source of information: patients and their families feel less alone by exchanging with others who are in the same situation and the associations provide practical advice to help them in their daily lives (Lucioles 2014).

Such associations generally concern parents or individuals for whom an etiological diagnosis is proven, but there is still no known aetiology for 30% of polyhandicapped people and, thus, no specific association to support them. They can, however, join more generalist associations concerning polyhandicaps. The associations are often a meeting place for professionals and parents. Professionals can use them to carry out studies that are the subject of various publication, on health topics or themes relating to the daily practice of parents at home or in coordination with health or medico-social establishments.
In the field of polyhandicaps, the work of professionals with parents has long taken on a very practical dimension within numerous collectives and associations, both national (UNAPEI white paper 2016, GPF) and local or regional. These associations organise numerous colloquia and training courses and publish tools resulting from research (ESCAP 2017) or the proceedings of their meetings.

4.6.3 Health networks, reference centres, and other organizations

The associations help to strengthen and support patient care, in collaboration with reference centres and centres of excellence, with the support of the DéfiScience network. They participate in research projects and may, if necessary, finance projects of major interest to patients *(see Appendix 2: List of Reference Centres and Associations)*.

Resource centres manage document collections on polyhandicaps and organise training courses which may be internal to an establishment or involve staff from several establishments, as well as actions/research and various studies: sleep, night work, IT, temporary care, Snoezelen, etc... Their long-term funding enables them to steadily develop**.**Various health networks also provide families with significant support in implementing a coherent care and life path for their polyhandicapped child. They have created tools available on the internet for the general public and professionals in all regions (R4P network's Compilio Compendium), have become recognised training organisations, guide families towards local professionals suitable for their child, and are financed in various ways, often not permanent, but recognised by the supervisory authorities.

A research action, named Polyscol (D. Toubert-Duffort 2018), is an example of an active partnership between various organizations: the subject of schooling for children with a polyhandicap has been explored by several establishments receiving such children, with the assistance of the Polyhandicap Resource Centre, the CREAI Rhône Alpes Auvergne, the Caisse Nationale de la Solidarité et de l'Autonomie (CNSA), the association APF département Handas, the CESAP Formation, and the INSHEA of Suresnes *(see Appendix 2 List of Reference Centres and Associations)*.

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- Pr Isabelle Laffont, MPR - President of SOFMER

Expressions of Interest

All coordinators involved in the development of the NSDP have completed an Expression of Interest (EOI).

Appendix 2. Addresses and Contact Information

- **Centres of reference and competence**
- **Patient associations and networks**
- **Resource Centre and Networks**
- **Further information**

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The addresses of the centres are available on line on the website http://www.defiscience.fr/filiere/.

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CR constitutive	CHU Dijon	Prof. Christel Thauvin
CR constitutiveCHU	Rennes	Dr. Laurent Pasquier
Constituent CRCHU	Strasbourg	Dr. Salima El Chehadeh
Constituent RCHCL	Lyon	Prof. Vincent Des Portes

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Competence CenterCHU	Besancon	Prof. Lionel Van Maldergem
Competence CentreCHU	Bordeaux	Prof. Cyril Goizet
Centre CompétencesCHU	Lille	Dr Audrey Riquet
Competence CentreCHU	Montpellier	Dr Pierre Meyer
Competence CentreCHU	Nancy	Dr Bruno Leheup
Competence CentreCHU	Nantes	Dr Bertrand Isidor
Competence CentreCHU	Nice	Dr. Fabienne Giuliano
Competence CenterCHU	Pointe à Pitre	Dr. Marilyn Lackmy Port Lis
Competence CentreCHU	Reims	Prof. Nathalie Bednarek
Competence CentreCHU	Toulouse	Dr. Caroline Karsenty
Competence CenterCHU	Tours	Prof. Annick Toutain

Centres of Reference for Congenital Cerebral Palsy and Congenital Diseases of the Cerebellum

Constituent Reference Centre	CHU Lille	Dr Audrey Riquet

Competence Centres for Cerebral Palsy and Congenital Cerebral Malformations and Diseases

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Competence Centre	Hospice Civils de Lyon	Dr Christelle Rougeot

Patient associations and networks

- Groupe Polyhandicap France GPF http://gpf.asso.fr/
- Réseau-Lucioles, Lyon: https://www.reseau-lucioles.org
- French Rett Syndrome Association - https://afsr.fr
- Association francophone du syndrome d'Angelman: AFSA: https://www.angelman-afsa.org
- Rare and poly handi : https://www.handyrareetpoly.fr
- Polycap - https://polycap.fr
- Association for the Defence of the Polyhandicapped ADEPO: https://www.adepo.fr/fr
- INJENO supports the parents of children with a polyhandicap and neuro-linguistic disabilities. https://www.facebook.com/injenoHDF/

Resource Centre and Networks

- CRMH - Multihandicap Resource Center of Ile de France - www.crmh.fr
- CESAP Training - Committee for Study, Education and Care of the Politically Handicapped https://www.cesap.asso.fr/cesap-formation-documentation-ressources/actuformation
- Association Ressources Plolyhandicap Hauts de France : http://ressourcespolyhandicap.org
- CRPGE - Centre Ressources Polyhandicap Grand Est (experimental) https://crpge.org
- HandiConnect - Multi-disability Tip Sheets https://handiconnect.fr/fiches-conseilsNDPC

Further information

ORPHANET - Source Internet : - http://www.orpha.net
Article for the general public
What to do in an emergency
Diagnostic Criteria
Focus Handicap
Clinical Genetics Review

Rare Disease Alliance - Federation of Rare Disease Organizations
Internet source: http://www.alliance-maladies-rares.org

Rare Diseases Info Service - Rare **Disease** Information and Call Platform
Source Internet: http://www.maladiesraresinfo.org/ Telephone: 01 56 53 81 36

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Appendix 3. Decision tree for etiological diagnosis

Figure 1: Participation of diagnostic professionals in the life project, according to professions and sectors (health, medical-social, educational)

(Source : Pr Vincent des Portes)

The "village" of the polyhandicapped child or adult has a large number of indispensable partners, and it grows over time in view of an ever richer and more adapted life project.

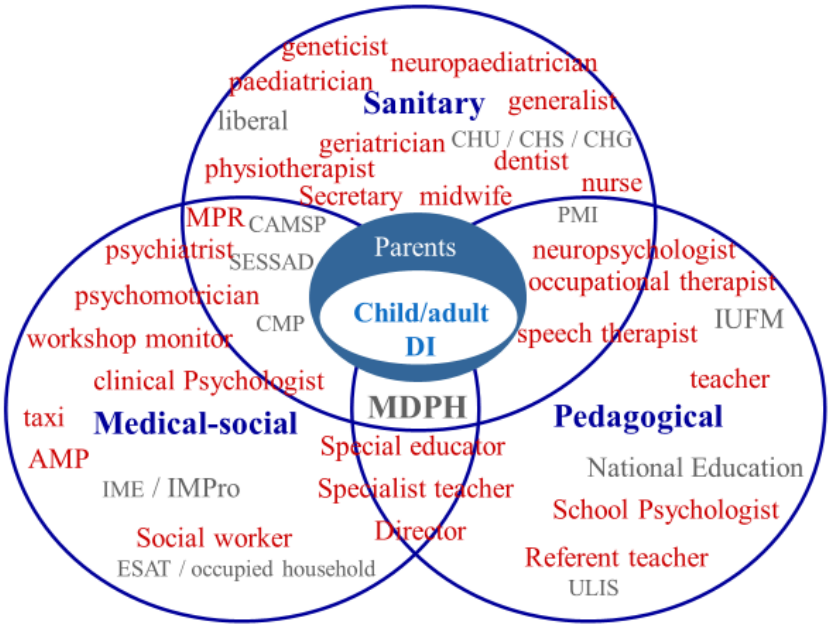
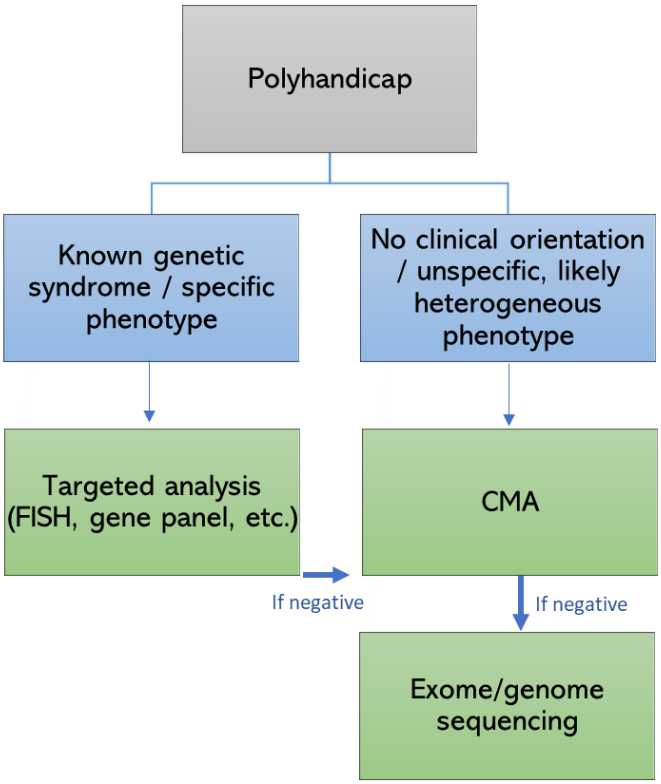


Figure 2: Proposed etiologic diagnostic strategy for a polyhandicap

(Source: Dr. Delphine Heron)



Appendix 4. Tables, figures and supplements

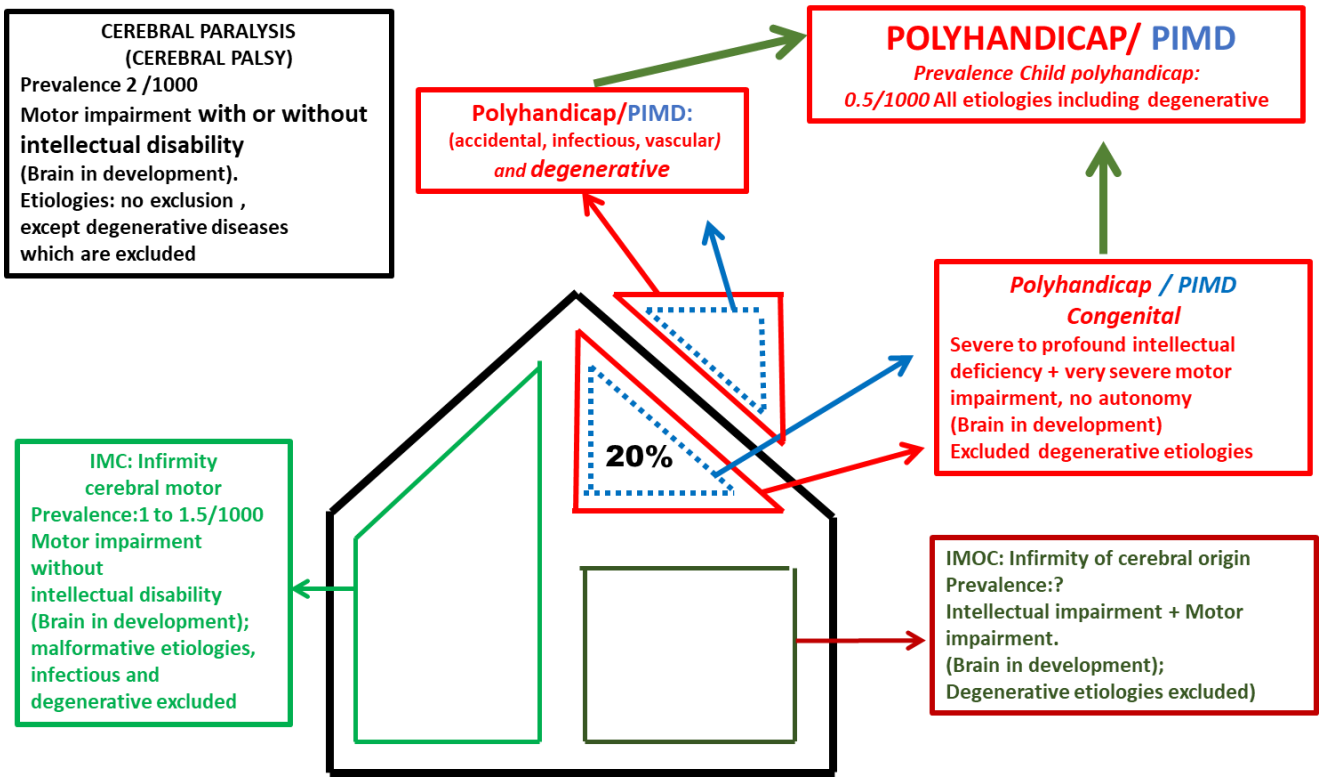
Cf. in relation to the chapter Definition of Polyhandicap

Table 1: Polyhandicap, medical aspects

<table>
<tr><td colspan="2">Brain injury = primary impairment</td></tr>
<tr><td>Multiple alterations of functions</td><td>Characteristics of the infringements</td></tr>
<tr><td>- Cognitive
- Motor (quadriplegia, abnormal mvts, tr. tonus, swallowing, no cough reflex...)
- Sensorial
- Sensitives
- Epilepsy
- Psychiatric disorders
- Neurovegetative disorders
- Sleep and alertness disorders</td><td>- Present from the discovery of a polyhandicap
- Usually fixed (except for degenerative diseases)
- Not accessible for curative treatment
- Need to exploit remaining opportunities (basis for rehabilitation)</td></tr>
<tr><td colspan="2">⇨ Committed vital prognosis</td></tr>
<tr><td colspan="2">Secondary alterations = direct consequences of primary alterations</td></tr>
<tr><td>Secondary Disorders</td><td>Characteristics of the infringements</td></tr>
<tr><td>- Osteo articular
- Growth
- Respiratory
- Digestive
- Urinary tract
- Speech and language
- Special problems: hydration, nutrition, bedsores, pain, microcirculation, iatrogenic risks.</td><td>- Very often absent initially
- Scalable in an intricate way
- Accessible prevention and treatment
- Sources of suffering
- Often the cause of morbidity and mortality (mainly respiratory disorders)</td></tr>
<tr><td colspan="2">Functional Disabilities</td></tr>
<tr><td colspan="2">- Communication disorders
- Behavioural and Relationship Disorders
- Posturo-motor and gesture disorders</td></tr>
</table>

(Source: Dr Philippe PERNES)

Table 2: The place of polyhandicap among neurological disabilities with motor impairment



(Source : Pr Gérard Ponsot – Graph)

Cf. in connection with the chapter: 1.3 Etiologies of polyhandicap

- **Table 1: Comparative study of surveys conducted from 1992 to 2018**

Authors	**L.George Janet 1992**			**AM. Boutin 1992**		**P.Pernes 2004**	**V Des Portes 2005**	**S.Mathieu, C Brisse 2006**	**J Furioli 2009**	**MC Rousseau 2013**	**P Pernes 2018 (unpublished)**
Population described	Comparative study over 3 periods of 3 groups of children monitored at ESCAP Poly, but also Pluri and Over-handicapped people			3 PA-HP Hospitals 470 files Children born between 1951 and 1987 (majority 1967-79) "Profoundly mentally retarded" + associated disabilities		Centre Antoine de St Exupéry Vendin le Vieil 204 cases children and adults born between 1974 and 2001 (majority 1979-90) Polyhandicapped persons (restrictive definition)	Bush Hospital Civil Hospitals of Lyon 81 gastrostomized patients CNS impairment in 95% of cases 2/3 fixed encephalopathies 1/3 progressive encephalopathies	La Roche Guyon Hospital 73 severely disabled children and adolescents Average age 17 years	Study of 40 polyhandicapped children hospitalized in general pediatrics (Mantes le Jolie Hospital)	Population of people with a polyhandicap in specialized SRG SSRs Studies of 133 deaths between 2006 and 2012 Heavily medically disabled persons GMFCS 4 or 5 70 children/63 adults	Centre Antoine de St Exupéry Vendin le Vieil 317 cases of children and adults born between 1974 and 2015 Polyhandicapped persons (restrictive definition) All GMFCS 5
	312 children followed between 1979 and 1982	389 children followed between 1983 and 1986	390 children followed between 1987 and 1990								
Precise diagnosis or time of onset	PERIOD OF BRAIN DAMAGE			REACH PERIOD	ACCURATE DIAGNOSIS	ACCURATE DIAGNOSIS	ACCURATE DIAGNOSIS	PERIOD OF BRAIN DAMAGE	ACCURATE DIAGNOSIS	ACCURATE DIAGNOSIS	ACCURATE DIAGNOSIS
Unknown	37.5%	31.5%	33.8%	27.9%	40.8%	26%	6.25%	22%	20%	14.3%	26.2%
Known	62.5%	68.5% Accurate diagnosis 55%	66.2% (54.6%)	72.1%	59.2%	74%	Certain: 57.5%. Suspected: 36.25%.	78%	80%	85.7%	73.8%
Birthdate	66.7%	61.3%	70.6%	56.7% (including 25 children with Down's syndrome 21)	47.3%	61.6%	82.6% (mostly genetic in origin)	61.4%	62.5	72.8% of which more than half are progressive encephalopathies	67.5%
native Peri	26.6%	28.7%	22.8%	28%	34.4%	16.6%	10.9%	22.8%	25	22.8%	15%
Post natal	7.7%	10%	6.6%	15.3%	18 ?3%	21.8%	6 ?5%	15.8%	12.5	4.4%	17 ?5%

(Source: Dr Philippe PERNES)

TABLE 2: Study of etiologies over 3 successive periods (Source: Dr Philippe PERNES)

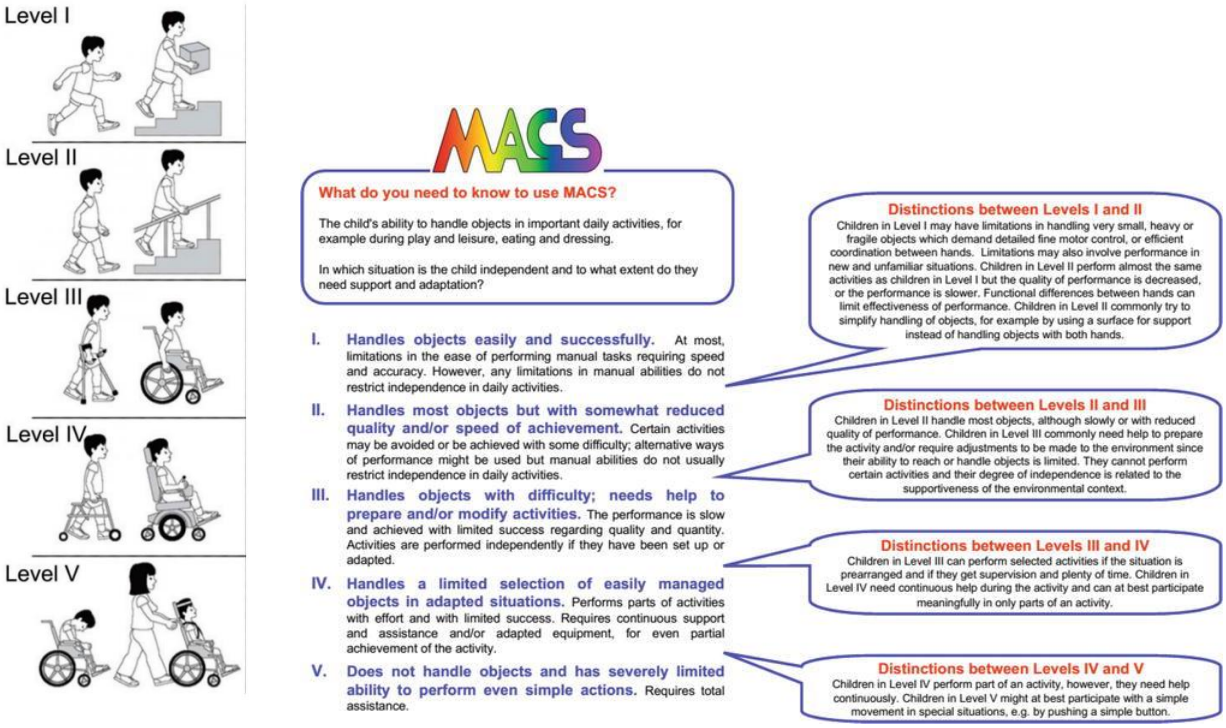
CENTRE ANTOINE DE St EXUPERY de VENDIN LE VIEIL. Dr PERNES **ETIOLOGICAL DIAGNOSIS 2017** **Updated: 17 02 2018**		**Years of Birth**	1974 -1985		1986-2001		2002-2015		TOTAL 1974-2015	
		Number of files	107+2 =109 files		97 +34 = 131 files		77 cases		317 files	
		Precise diagnosis	**Precise diagnosis 77/109 = 70.6%**		**Precise diagnosis 97/131 = 74 %**		**Precise diagnosis**		**Precise diagnosis 234= 73.8%**	
			Number of cases out of 77	%	Number of cases out of 97	%	Number of cases out of 60	%	Number of cases out of 234	%
NATAL ANTE PERIOD			**44/77**	**57.2%**	**65/97**	67%	49	81.6%	158	67.5%
A-GENETICS			15/77	19.5 %	33/97	34%	25	41.7%	74	31.6%
	1-Chromosomal disorders		5	6.5%	8	8.2%	16	26.7%	30	12.8%
	2-S.clinical defined with or without bio-moleculars	Rett, Angelman, Cornelia de Lange... Aicardi, Tuberous Sclerosis of B...	7	9.1%	7	7.2%	4	6.7%	19	8.2%
	3-Metabolic diseases		2	2.6%	15	15.5%	5	8.3%	21	8.9%
	4-Family cases with no genetic abnormality found		1	1.3%	3	3.1%	/	/	4	1.7%
B- ACQUISITIONS			14/77	18.2%	14/97	14.4%	6	10%	33	14.1%
	1-Infections		7	9.1%	2	2.1%	/	/	9	3.9%
	2-Toxic	endo or exotic	1	1.3%	5	5.1%	3	5%	8	3.4%
	3 -Vascular	Ischemia hemorrhage	6	7.8%	7	7.2%	3	5%	16	6.8%
C-CEREBRAL DEFORMITIES			12/77	15.6 %	16/97	16.5%	18	30%	44	18.8%
D- POLY-MALFORMATIVE SYNDROME			1	1.3%	1	1.%	/	/	3	1.3%
E-MISCELLANEOUS		CO poisoning, diaphragmatic hernia...	2	2.6%	1	1%	/	/	4	1.7%
NATAL PERIOD			**16/77**	**20.8%**	**12/97**	**12.4%**	**7**	**11.7%**	**35**	**15%**
	1- Anoxo - ischemia	Term / Prema++ / Post ma	10	13%	9	9.3%	7	11%	26	11.1%
	2- CNS Infection	Meningitis-encephalitis - septicaemia	4	5.2%	3	3.1%	/	/	7	3%
	3- Metabolic and miscellaneous		2	2.6%	0	/	/	/	2	0.8%
POSTNATAL PERIOD			**17/77**	**22.1%**	**20/97**	**20.6%**	**4**	**6.7%**	**41**	**17.5%**
	1- Head injuries	(Abuse ++)	4	5.2%	9	9.3%	2	3.3%	15	6.4%
	2- Infection		7	9.1%	3	3.1%	/	/	10	4.3%
	3- EDMal epileptic		4	5.2%	4	4.1%	/	/	9	3.9%
	4-Post natal anoxia	Staph. pleuro-pulmonary,. drowning...	2	2.6%	3	3.1%	/	/	5	2.1%
	5 HCM		/	/	1.	1%	1	1.7%	1	0.4%
	6 Malignant hyperthermia		/	/	/	/	1	1.7%	1	0.4%

Cf. in connection with chapter: 2.2 Motor deficiencies and tonicity disorder

- **MRC Mucle Power Scale**

0 =	**No contraction**
1 =	**Fliker or trace of contraction**
2 =	**Active movement, with gravity eliminated**
3 =	**Active movement againt gravity**
4 =	**Active movement againt gravity and resistance**
5 =	**Normal power**

- **GMFCS Scale and MACS Scale**

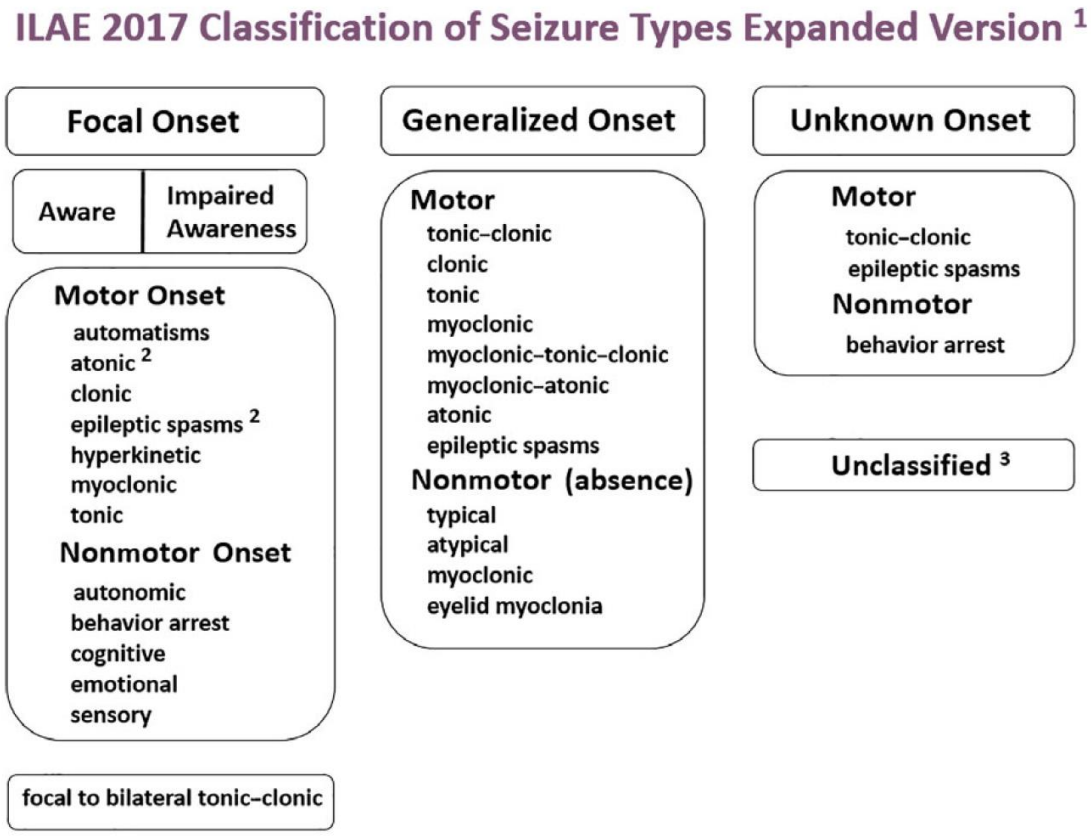


- **Modified Ashworth Scale**

Grade	Description
0	No increase in muscle tone
1	Slight increase in muscle tone, manifested by a catch or by minimal resistance at the end of the range of motion (ROM) when the affected part(s) is moved in flexion or extension
1+	Slight increase in muscle tone, manifested by a catch, followed by minimal resistance throughout the remainder (less than half) of the ROM
2	More marked increase in muscle tone through most of the ROM, but affected part(s) easily moved
3	Considerable increase in muscle tone, passive movement difficult
4	Affected part(s) rigid in flexion or extension
9	Unable to test

Cf. in connection with chapter: 2.4 Epilepsy

Figure 1 Classification of Seizures
Framework for classification of epilepsies. *Denoes onset of seizue. Epilepsia©ILAE
Detailed version of the ELAI Classification of Seizure Types



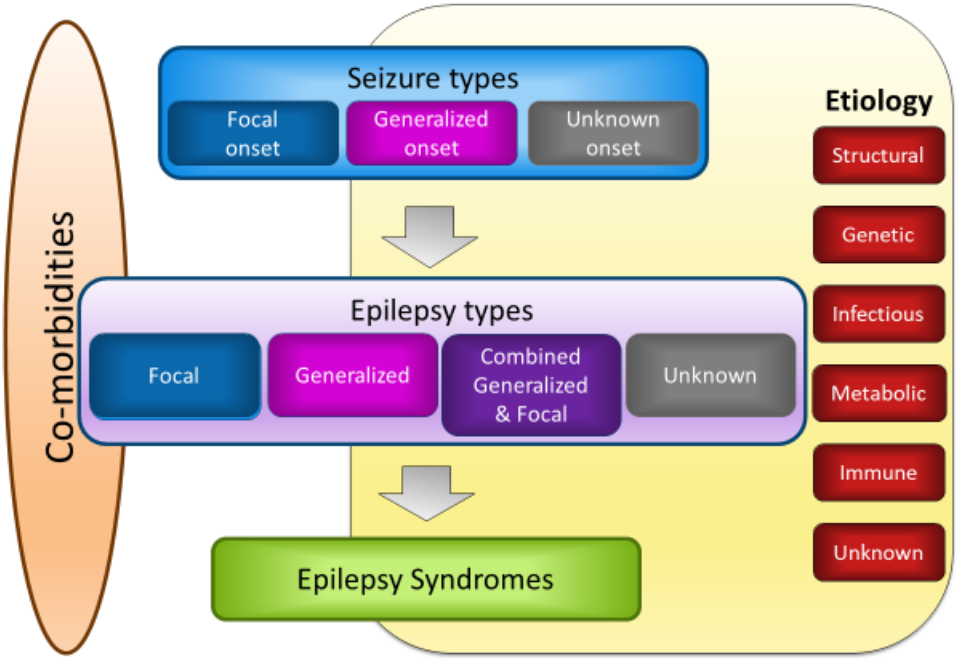


Figure 2 Seizure Descriptions Help Guide

- **Signs of onset**: gradual, abrupt onset; clinical **signs**
- **Triggering or facilitating factors**: noise, tactile stimulation, sleep or wakefulness, pain, hyperthermia, constipation, wakefulness or sleep.
- **Partial or generalised crisis**
  - Partial crisis
  - Secondary widespread partial crisis
  - Widespread crisis from the outset
- **Disorders of consciousness**
  - Total loss of consciousness
  - Contact failure
  - Absence of impairment of consciousness or alertness
- **Type of crisis**
  - Clonic
  - Tonic
  - Tonico-clonic
  - Myoclonic
  - Insulated contact break
  - Spasm(s)
  - Atonic
  - Seizure salvo (repeated seizures)
- **Schedule / Duration**
- **Accompanying signs**
  - Vegetative disorders: hypersalivation, swallowing, chewing, nausea, sweating, palpitations, redness, pallor.
  - Breathing disorders = noisy, snoring, due to the inertia of the palate veil, cyanosis
  - Digestive signs: sometimes vomiting at the end of a seizure
  - Gestural automatisms = stereotyped hand movements (rubbing), various verbal automatisms, onomatopoeia etc...
  - Small motor signs = fluttering eyelids, a labial commissure
- **End of the crisis**: gradual, brutal
- **Post critical disorders**: obsnubilation, paralysis, ...
- **Administration of emergency treatment**
- **Change in background treatment** (noted by the doctor) Videos made by family or professionals are often a great help; these items should be noted in a personal crisis diary.

(Source: Dr Philippe PERNES)

- **Table 1 The different generations of antiepileptics**

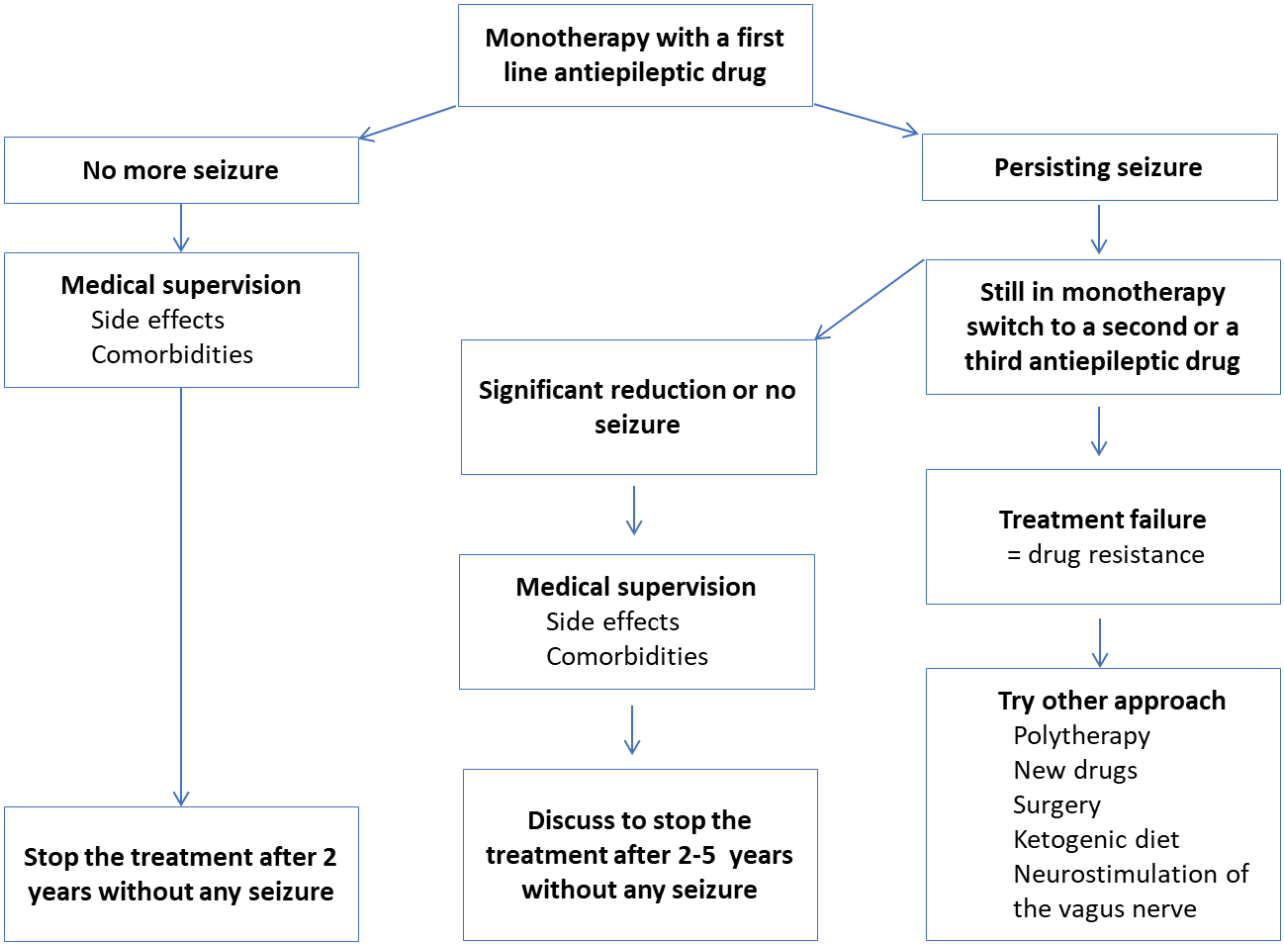
The different generations of antiepileptics (Vallée L., 2016)

First generation		Second generation		Third generation	
Phenobarbital	1912	Vigabatrin	1990	Stiripentol	2001
Phenitoin	1938	Gabapentin	1994	Levetiracetam	2093
Primidone °	1953	Felbamate	1995	Pregabaline	2004
Carbamazepine	1960	Lamotrigine	1995	Zonisamide	2005
Ethosuximide	1962	Clonazepam	1995	Rufinamide	2007
Diazepam	1963	Tiagabide	1997	Lacosamide	2008
Valproate	1967	Topiramate	1998	Eslicarbazepine	2009
° deleted from the pharmacopoeia		Oxcarbazepine	2000	Pérampanel	2012

(Source: Pr Louis Vallée, 2016)

Note: Cannabidiol has a European MA for Dravet's syndrome and Lennox-Gastaut syndrome.

Figure 3: Drug Management of Epilepsy



(Source: Dr Philippe PERNES)

Cf. in connection with chapter 2.5 Pain

Table 1. Three surveys on the prevalence of pain

Table 1.	P.Stallard 2001	LM Breau 2003	P Gallien 2009
Study population	34 children - Average age 9.4 years	94 children and teenagers from 3 to 18 years old Average age 10.1 years	258 adults Average age 37 years
Impairments	Severe intellectual disability without verbal language/ - 18 CNS reach set - 7 progressive encephalopathies - 9 chromosomal alterations	Moderate to profound intellectual disability Cerebral palsy: 44 out of 94	Cerebral Palsy Gross Motor Function Classification System (GMFCS): 43% level 4 or 5 Non-Marketers 35%.
Study period	Prospective study over 2 weeks	Retrospective study over 4 weeks in 1 year	Retrospective study over several years
Results	73.5 % > to 1 painful episode of which 84% > or = to 5 separate days Duration74 episodes > à 30 mn	78% > 1 painful episode Average duration > to 9 hours/week	82% > 1 painful episode - 40% daily - 28% weekly - 22% monthly 73% lasted > 1 year
Cause of pain	No Specified	30% Accidental 22% of digestive origin 20% of infectious origin 19% of musculoskeletal origin	Essentially musculoskeletal
Means used	Caregiver Questionnaire Classification as mild, moderate or severe pain	Caregivers; Use of the NCCPC Scale	Caregiver Questionnaire
Treatment	No children were being treated	No Specified	No Specified

(Source: Dr Philippe PERNES)

Table 2. Acute and chronic pain

Table 2	Acute pain (symptom)	Chronic pain (disease)
Biological purpose	Useful, Protective, Alarm signal	It's no use, Destructive, Full-blown disease
Generating mechanism	Single factor (e.g. trauma)	Plurifactorial
Neurovegetative reactions	Reactions	Habituation or maintenance
Emotional component	Anxiety	Depression (adult) or psychomotor atony (child)
Model of understanding	Classical medical	Multi-dimensional somato-psycho-social.

(Source: Dr Philippe PERNES)

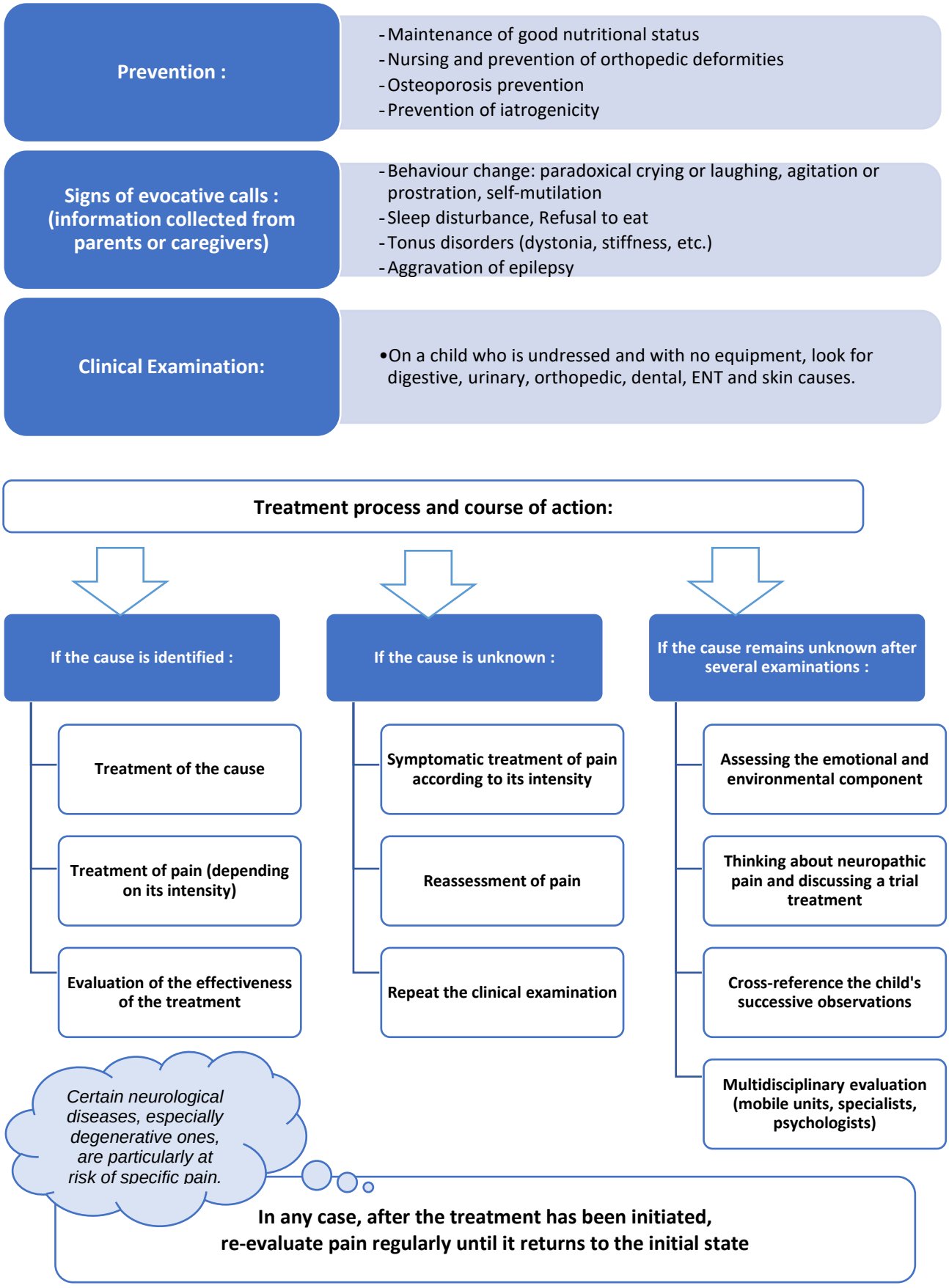
Table 3. Comparison of Pain scales for use in the polyhandicapped individual
(Source: Dr Philippe PERNES)

Scale	Target population	Age	Number of items	Threshold of Pain	Filling time	Basic backrest	Remarks
DESS Pain Child San Salvadour	Polyhandicap	Child but applicable to both the adolescent and the adult	10 items, rated from 0 to 4	Pain if = or > 6/40	5 min for a trained team	Yes	The only one really designed for polyhandicaps. Need to have a basic pain record More like chronic pain
EDAAP Expression of Pain for Adults and Adolescents with a polyhandicap	Polyhandicap	Adult and teenager	11items, Rated from 0 to 3, 4 or 5	Pain if >7 /41	5 to 10 mn	Yes	DESS/Doloplus Compromise Need to have a basic pain record
NCCPC Non Communicating Children's Pain Checklist = **GED-DI** Pain - Intellectual Disability Assessment Grid	Patient unable to express him/herself verbally in relation to a cognitive disability. Autistic children included in its validation	From 3 years to adulthood	30, rated from 0 to 3 Score from 0 to 90 -27, post-op. (score from 0 to 81 because 3 items less)	6 to 10: mild pain >11: moderate to severe pain	5 to 10 mn	No	No need to have a basic pain record Disadvantages: long More like acute pain
DOLOPLUS 2	Non-communicating person	Elderly person	10 items in 3 groups, Rated from 0 to 3	pain if > 5/30	A few minutes.	Yes	Need to have a basic pain record
FLACC Modified (Face Legs Activity Cry Consolability) French version modified for children with disabilities	Provided for post-operative and care-related pain	Validated from 2 months to 7 years, Can be used from birth to 18 years of age	5 items, rated from 0 to 2, Score d 0 to 10	1-3 = slight discomfort 4-6 = dl moderate 7-10 = dl intense or major discomfort	1 to 2 mn	No	Simple, quick but not very specific to polyhandicaps. English version validated in post-op French version modified for handicapped and autistic children Low sensitivity
PPP (Pediatric Pain Profile) = Pediatric Pain Profile	Children with disabilities neurological severe non-communication	1 to 18 years old	20 items, rated from 0 to 3, Score from 0 to 60	If >14 = moderate to severe pain	5 to 10 mn	Yes	Need to have a basic pain record

Table 4. Main analgesic drugs classified according to the 3 WHO levels
(Source: Dr Philippe PERNES)

Class	Generic	Specialty	Dosage	Side Effects	Remarks
Stage 1 Peripheral analgesics	Paracetamol	- Dafalgan powder at 80, 150 and 250mg -Dafalgan 500mg or 1g	E: 60 mg/kg/day in 4 intakes A : up to 4g/d	Allergic skin reactions, hepatotoxicity at supratherapeutic doses	From birth
	Ibuprofen	- Nurofen pro sol. drinkable at 20mg/ml - Nurofen cp 200 and 400 mg	E: 20 to 30mg/kg/d in 3 sockets A: max 3 cp at 400mg/d	Digestive (++ precautions if GERD), allergic, neurological	Child > 3 months
Stage 2 Weak central morphinics	Codeine	- Codenfan syrup - Dafalgan Codeine (500mg paracetamol and 30mg codeine)	E: 3 to 6 mg/kg/day A: 1 to 2 cp X 3 to 4/d	Opiates, but less so, especially nausea, constipation, drowsiness, dizziness, etc.	E > 12 years. In short cures After Tier 1 ineffectiveness. Never after tonsillectomy or adenoidectomy.
	Tramadol	-Topalgic floor. drinkable 1gtt = 2.5mg 50 and 100 mg gel	E: 1-2 mg/kg/intake x 3 to 4 times a day A: Max 400 mg/d in 4 to 6 intakes	Opiates, but less so, especially nausea, vomiting, feeling sick.	WMA from 15 years of age Discussed recently before 15 years old, as possible dependency
Stage 3 Powerful Central Analgesics	Immediate release morphine	- Oramorph Oral Solute2.5 mg/drop and single doses of 10, 30, 100mg - Actiskenan capsules 5, 10 and 20 mg - Sevredol cp at 10 and 20mg	1 dose every 4 hours Maintenance Dose : 1 mg/kg/day	Constipation to be prevented by the addition of PEGs Side effects: neurological, pruritus, bradycardia, urinary retention...	A and E > 6 months Out of MA (Marketing Authorization) at birth
	Sustained-release morphine	- Skenan capsules 10, 30, 60, 100 and 200 mg - Moscontin cp at 10, 30, 60, 100 mg	2 intakes per day		
	Fentanyl	- Durogesic patch at 12, 25, 50, 75, 100µg/hr. 1 patch / 72 hours	12 microg/hour corresponds to 30 mg/24 hours of Morphine.	Those of the morphinics	A and E > 2 years

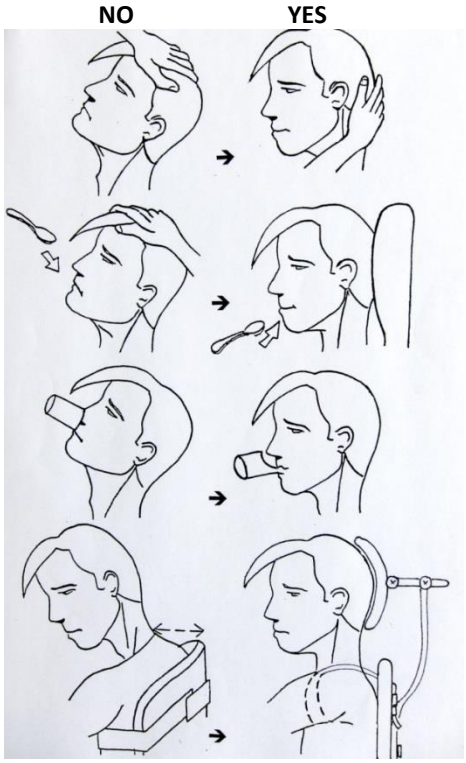
Figure 1: Prevention, diagnostic approach and proposal for pain management in children with neurological disabilities
(Source: Dr. Justine Avez)



Cf. in connection with Chapter 2.7: Eating disorders

(Source: Dr Philippe PERNES)

Indications of facilities and practices



Mouth closure aid techniques

Cf. in connection with Chapter 2.15: Hearing disorders

Table 1: Degrees of Severity of Hearing Impairment

Classification Bureau International d'Audio Phonologie (BIAP)

Degree of deafness	Hearing loss in the better ear		Perceived speech	Perceived Noise
Normal hearing	0 - 20dB			
Deafness light	21-40 dB		Speech perceived in a normal voice, not easily perceived in a low or distant voice	Mostly perceived familiar sounds
Moderate deafness	41 -70 dB	1st degree: 41-55 dB	Perceived speech if you raise your voice	Some familiar perceived noises
		2nd degree: 56-70 dB		
Deafness severe	71 -90 dB	1st degree: 71-80 dB	Loud speech near the ear	Perceived loud noises
		2nd degree: 81-90 dB		
Profound deafness	91-120 dB	1st degree: 91-100 dB	No perception of speech	Only very loud noises are perceived
		2nd degree: 101-110 dB		
		3rd degree: 111-120 dB		
Cophose	>120 dB		No perception of speech	Some very powerful noises can be heard (Formula 1 car, plane taking off).

Table 2: Anatomical location of deafness (Source: Dr Philippe PERNES)

→ **Conductive Deafness:** affected
- of the outer ear (atrial atresia, obstruction.)
- of the middle ear (eardrum, eardrum body, ossicles): serous otitis most often, acute or chronic otitis.

The nerve message to the central auditory pathways is consistent but attenuated. The inner ear is intact.
The treatment is either medical or medico-surgical.

→ **Sensorineural Deafness**

There are stabilized or evolving functional or lesional disturbances in the encoding, transfer, and/or decoding of acoustic information in retrocochlear and central auditory pathways. The message is of insufficient intensity and may be blurred or distorted.
They may be original:
- Cochlear: inner ear involvement: most common
- Retrocochlear: damage to the auditory nerve
- Central: damage to the auditory pathways of the brainstem, subcortical and cortical

Treatment: only cochlear injuries can benefit from hearing aids or cochlear implants. Rehabilitation is always necessary.
Endocochlear sensorineural deafness may be accentuated by transmissive damage - usually reversible - and increased by associated retrocochlear or central damage, which will penalise the developmental, particularly language, development.

Table 3: Screening for Permanent Deafness in Newborns (Source: Dr Philippe PERNES)

Oto Acoustic Emissions (OEA)

Sounds produced by the inner ear (contraction of the outer hair cells) in response to acoustic stimulation, recorded by a miniature microphone placed in the inner ear canal and analysed by computer
- Quick (5mn) and painless method
- To be practiced after 48 hours of life (previously persistence of amniotic fluid in the tympanic cavity).
- Allows the detection of deafnesses with more than 40 dB of loss
- Explore the inner ear (cochlea) but not the auditory nerve: therefore, they are not recommended for newborns in intensive care unit where retrocochlear hearing loss is more common.
- Information on high frequencies
- False positives (normo-hearing newborns screened for deafness) ranging from 8.8% in the first test to 0.7% in the test.

Automated Auditory Evoked Potentials
- Reflections of the passage of nerve impulses at the nerve and auditory pathways of the brain stem
- Method a little longer (15 minutes) and expensive
- Can be practiced from D1
- Evaluate a response at a fixed intensity (usually 30 dB)
- Explore central retrocochlear disease and are therefore recommended for newborns in the intensive care unit w this disease is more common.
- Information on high frequencies
- False positives ranging from 6.1% in the first test to 1.3% in the re-test.

The absence of a PEAO or AEPP may indicate a treble loss greater than 30 dB, to be confirmed by AMATCP.

Cf. in connection with chapter 3.5: Iatrogenies

Table 1: Drugs most commonly used in people with a polyhandicap (Source: Dr. Philippe PERNES)

CLASS	DCI	WMA	UNDESIRABLE EFFECTS	
NEUROLEPTICS	RISPERIDONE	Adult - Child > 5 YEARS	Drowsiness, asthenia, constipation, insomnia, orthostatic hypoTA, weight gain, extrapyramidal syndrome QT prolongation, torsades de pointes, rhythm disorders → ECG before treatment Rare malignant syndrome	
ANTI DYSTONICS	CLONIDINE	Adult (HTA) Not recommended for children	Drowsiness, constipation, dry mouth, digestive disorders, asthenia, orthostatic hypoTA, rash	
	TETRABENAZINE	Adult. Not recommended for children	Drowsiness, asthenia, parkinsonian syndrome, depressive syndrome; do not combine with L Dopa or dopaminergic agonists	
	TRIHEXIPHENIDILE	A/E	Drowsiness, dry mouth, urinary retention, constipation	
ANTISPASTIQUES	BACLOFENE	Adult - Child > 6 years due to the tablet dosage form	Drowsiness, gastrointestinal disturbances, rash, aggravation of axial hypotonia, lowering of epileptogenic threshold	
ANTIDEPRESSANTS	AMYTRIPTYLINE	Adult - Child 6 to 15 years old (indication: enuresis)	Drowsiness, sedation, dry mouth, constipation, accommodation disorders, orthostatic hypoTA, weight gain, conduction and rhythm disorders at high doses.	
HYPNOTICS	MELATONINE	Adult-Child > 6 years due to the tablet dosage form → use melatonin base	Rare abdominal pain, nausea, headache, constipation, dry mouth. Overdose: drowsiness	
ANTIEPILEPTICS	2nd and 3rd generation AEs	Adult - Child : according to specialities	Virtually all: drowsiness, cognitive problems, dizziness	
			Specific side effects	
			Felbamate: Hematologic and Hepatic Toxicity Lamotrigine: Rash, Stevens Johnson, Lyell Oxcarbazepine: Hyponatremia Topiramate: Anorexia, weight loss, urinary lithiasis, toxic encephalopathy	Stiripentol: Anorexia, ataxia Zonisamide: Anorexia, Urinary Lithiasis, Hematologic Disorders Perampanel: Anorexia Rufinamide: Ataxia Pregabalin: Neutropenia, QT prolongation
ANTALGICS	PARACETAMOL	Adult - Child from birth	Rare: allergy, thrombocytopenia. Hepatotoxicity at supratherapeutic doses	
	NSAIDS	Adult - Child > 3 months for Ibuprofen	Allergy, gastric ulcer, GI bleeding, increased transaminases, abnormal NF. Precautions in case of GERD. Strongly discouraged in case of chickenpox in children.	
	TRAMADOL and Morphinics	Adult - Child > 3 years old	For all morphinics but less: Drowsiness, nausea, vomiting, constipation, urinary retention.	
	GABAPENTINE	Adult - Child > 3 years (> 6 years in association)	Drowsiness, dizziness, asthenia, headache, weight gain, digestive disorders, skin rash, neutro and thrombocytopenia. In children: hyperkinesia	
IPP	ESOMEPRAZOLE	Adult - Child > 12 months	Digestive disorders: nausea, vomiting, abdominal pain, diarrhea. Very rare: skin reactions, agranulocytosis, hemolytic anemia, hyponatremia, decreased bone density, increased risk of Clostridium difficile infections.	
ANTICHOLINERGICS	Trans dermal SCOPOLAMINE	Adult - Child > 15 years old	Dry mouth, hyperviscosity of bronchial secretions, accommodation disorders, constipation, urinary retention, disorientation, confusion, agitation	
Alpha 1 Blockers	ALFUZOSINE	Adult - effectiveness not demonstrated in children 2 to 16 years of age.	Constipation, dry mouth, vomiting, asthenia, headache, skin rash, dizziness.	
BISPHOSPHONATES	PAMIDRONATE	Principle of the right prescription adapted for the polyhandicapped child	Influenza-like illness, fever, nausea, vomiting, cramps, abdominal pain, anemia, leukopenia, thrombocytopenia, impaired renal function, hypocalcemia, hypophosphatemia.	
BENZODIAZEPINES	CLONAZEPAM	Adult - Child without age limit Indication: epilepsy alone.	Drowsiness, hypotonia, bronchial congestion First prescription by neurologist or paediatrician on secure prescription	

Table 2: The side effects of drugs that interfere with comfort and daily life (Source: Dr Philippe PERNES)

Medications that promote constipation	**Medicines to promote urinary retention**
- Morphinic analgesics - Anti-asthmatics: Terbutaline - Tricyclic antidepressants: Amitryptillin - SRI: Fluoxetine, Paroxetine - Antiepileptics: Phenitoin, Clonazepam, Topiramate - Antihistamines - Atropinics: Atropine, Scopolamine - Muscle relaxants: Baclofen - Neuroleptics: Risperdone, Tetrabenazine... - Anti anaemia: Iron in all its forms - AntiParkinsonian: LDopa - Hypotensive: Clonidine - Alpha blockers: Alfusozine	- Morphinic analgesics - Anti-asthmatics: Ipatropium bromide - Antiemetics: Metopimazine - Tricyclic antidepressants: Amitryptillin - SRI: Fluoxetine, Paroxetine - Antiepileptics: Phenitoin, Clonazepam Pregabalin, Carbamazepine, - Antihistamines - Antipsychotics: Olanzapine, Chlorpromazine - Urinary antispasmodics: Oxybutinin - Atropinics: Atropine, Scopolamine - Muscle relaxants: Baclofen - Neuroleptics: Risperdone, Tetrabenazine... - AntiParkinsonian: LDopa
Medication and appetite	
Increased appetite - Antiepileptics: Sodium Valproate, Pregabalin - Neuroleptics - Antihistamines - Beta blockers: Avlocardyl - Corticotherapy	**Decreased appetite** - Antiepileptics: Topiramate, Stiripentol, Zonisamide, Perampanel - Methylphenidate
Drugs and saliva	
Hypersalivation - Benzodiazepines: Clonazepam	**Dry mouth** - Atropinics: Atropine, Scopolamine - Neuroleptics - Antiepileptics: Gabapentin - Antihistamines - Antidepressants: Amitryptilline - Anxiolytics - Alpha blockers: Alfusozine - Hypotensive: Clonidine

Cf. in connection with Chapter 4: Life journey and accompaniment

AAH (Disabled Adult Allowance)

The AAH is a financial aid that ensures a minimum of resources. This assistance is granted subject to compliance with criteria of disability, age and residence. It is applied for under the MDPH, granted by decision of the Commission on the Rights and Autonomy of Disabled Persons (CDAPH) systematically if the rate is higher than 80%. It is paid monthly by the CAF. Its amount supplements any other resources of the person with a disability, with rules of accumulation.

AEEH (Allowance for the Education of Disabled Children)

The AEEH is a lump-sum benefit designed to compensate for the costs of education and care of a child with a disability. This assistance is paid to the person who is responsible for the child's care. This allowance may be supplemented by a supplement which varies according to the costs related to the disability and the reduction in the parents' working time due to the child's disability. An increase for a single parent may be added.

It is means-tested and is paid to the family of a disabled child under the age of 20. It is applied for under the MDPH, granted by decision of the CDAPH, and systematically if the disability rate is over 80%. This family benefit is paid by the family allowance funds (CAF) or the agricultural social mutual insurance funds (MSA) for persons covered by the agricultural scheme.

PCH (Disability Compensation Benefit)

The PCH is a financial assistance provided by the Department. It covers certain disability-related expenses (e.g., home or vehicle modifications, use of a third party).

It is a personalised aid, adjustable according to needs, which can cover 5 fields: Human aid, Technical aid, Aid for housing development, Vehicle fitting out and additional costs linked to transport, Specific or exceptional charges, Animal aid. It is requested from the MDPH, and granted by decision of the CDAPH.

CAMSP (Centre for Early Medico-social Action)

The CAMSPs are designed to screen, provide outpatient treatment and rehabilitation, in collaboration with parents, for children under the age of 6 with sensory, motor or mental deficits, with a view to their social and educational adaptation in their natural environment. They carry out preventive actions. They are versatile or specialized, intervening only through consultations in centres or at home. They are freely solicited by parents or any other structures or interveners. There are about 300 of them.

CMPP (Centre médico-psycho-pédagogique)

The CMPPs are consultation centres that provide diagnosis and treatment for school-age children and adolescents with neuropsychological disorders (psychomotor, speech and language difficulties, learning disabilities) or behavioural disorders that are susceptible to medical therapy, medical-psychological re-education or psychotherapeutic or psycho-pedagogical re-education under medical authority. Approximately 500 exist on the territory.

CAFS (Specialized family reception centre)

Its aim is to provide children and adolescents with a psychological, educational and emotional environment that they cannot find in their own environment.

It is attached to an EMI, a CMPP or a CAMSP and only takes in children who are followed up.

Foster families are approved by the President of the Departmental Council for the reception of children up to the age of 21.

There are about 80 CAFS, with an average of 15 places.

IME (Institute for Medical Education)

EMIs are reception facilities providing adapted education and medical-social support to children and adolescents (3 to 20 years old) suffering from predominantly intellectual deficiencies linked to neuropsychic disorders (profound, moderate, mild intellectual deficiencies, with or without associated disorders). Admission is applied for at the MDPH, granted by decision of the CDAPH. They can function in a boarding school, semi-boarding school, on a prolonged basis or in temporary accommodation. A little more than 1,200 EMIs are listed in France for about 70,000 places, with a great heterogeneity according to regions (from 1.6 to 6 per 1,000 young people under 20 years old).

EEAP (Establishment for children or adolescents with a polyhandicap)

Their aim is to provide children and adolescents with a polyhandicap (from 3 to 20 years of age) with specialized follow-up aimed at their overall development and the reduction of their dependency. Admission is applied for under the MDPH, granted by decision of the CDAPH. They can operate in a boarding school, semi-boarding school, on an extended basis

or in temporary accommodation. A little more than 200 EEAPs are listed in France for around 6,000 places, with great heterogeneity according to region (from 0 to 0.72 per 1,000 young people under 20 years of age).

SSAD (Service de soins et aides à domicile)
The SSAD's action is oriented towards:

- On the one hand, early care for children from birth to 6 years of age, including advice and support for families and the child's family circle, the deepening of the diagnosis and the initial psychomotor development of the child as well as the development of communication;
- On the other hand, for all children and adolescents, support for the acquisition of autonomy, including all appropriate medical, paramedical and psychosocial means.

Admission is applied for under the CDM, granted by decision of the CDAPH.

FAM (Foyer d'accueil médicalisé) and MAS (Maison d'accueil spécialisée)
AFM and SAM are very close. They take in adults who are severely handicapped, either intellectually or physically, whose dependency makes them unfit for any professional activity and requires the assistance of a third person for most of the essential acts of life. They fulfil essential missions: to develop learning of daily life, to accompany in the essential acts of life, to provide a place to live in conditions that allow people to express themselves and to favour relational and cultural life, to ensure constant medical follow-up and care. Admission is requested to the MDPH, granted by decision of the CDAPH. They can operate in a boarding school, semi-boarding school, on a prolonged basis or in temporary accommodation. Approximately 850 FAM and 650 MAS are listed in France for about 25,000 places each, with great heterogeneity according to region (from 0 to 1 (FAM) and from 0.3 to 2 (MAS) per 1,000 adults under 60 years of age).

SAAD (Service d'aide et d'accompagnement à domicile)
SAADs provide assistance with daily living, assistance with essential tasks and support for social life in or from the home of adults. Access to the service is most often determined by the CDAPH. There are several professions that can intervene in the home, in particular: the home agent and the home employee; the social life assistant (AVS); the social and family intervention technician (TISF); the medico-psychological aid (AMP).

SAVS (Service d'accompagnement à la vie sociale),
The purpose of the SAVS is to provide adapted support by encouraging the maintenance or restoration of social ties in the family, university or professional environment and by facilitating access for disabled adults to all the services offered by the community. Their missions consist of assistance and accompaniment in essential acts of life as well as social accompaniment in an open environment.

SAMSAH (Medical-social support service for disabled adults)
The SAMSAHs' vocation is to carry out the social and professional integration missions also assigned to the SAVS within the framework of adapted medical-social support including care services. These services are aimed at more severely handicapped people in order to provide them with a multidimensional response integrating a therapeutic dimension. Admissions are requested from the MDPH, granted by decision of the CDAPH.

SPASAD (Service polyvalent d'aide et de soins à domicile pour personnes âgées et/ou handicapés adultes)
The Services polyvalents d'aide et de soins à domicile (SPASAD) provide both the missions of a Service de soins infirmiers à domicile (SSIAD) and the missions of a Service d'aide et d'accompagnement à domicile (SAAD). Their justification was aimed at strengthening the coordination of interventions with individuals and pooling interventions aimed at developing the individual project of assistance, accompaniment and care.

SSIAD (Home nursing care service for elderly and/or disabled adults)
The Home Nursing Care Services (HNCS) provides nursing care services in the form of technical care or basic and relational care for adults under 60 years of age with a disability. Under the responsibility of nurses, nursing assistants (as not indicated by the name SSIAD) provide basic and relational care and assist in the performance of essential life tasks.

Possible places to live and society's responses (Source: Pr Dominique Robert)

	Evaluation / outpatient resource services	**Financial assistance**	**Living at home with help (social worker, social and family intervention technician, care assistant)**	**Reception without accommodation**	**Reception with accommodation**
0 to 6 years	**CAMSP** (centre for early medical-social action) **CMPP** > 3 years (medico-psycho-educational centre) **SSAD** (home care and home help service)	**AEEH** (education allowance for disabled adult children)		**EEAP** > 3 years (facility for children or adolescents with disabilities) **EMI** > 3 years (medico-educational institute)	**EEAP** (institution for children or adolescents with disabilities) **EMI** > 3 years (medico-educational institute) **CAFS** (specialized family reception centre)
7 to 12 years old	**CMPP** (medico-psycho-educational centre) **SSAD** (home care and home help service)	**AEEH** (education allowance for disabled adult children)		**EEAP** (institution for children or adolescents with disabilities) **IME** (medical-educational institute)	**EEAP** (institution for children or adolescents with disabilities) IME (medical-educational institute) **CAFS** (specialized family reception centre)
13 to 18/20 years old	**CMPP** (medico-psycho-educational centre) **SSAD** (home care and home help service)	**AEEH** (education allowance for disabled adult children)		**EEAP** (institution for children or adolescents with disabilities) **IME** (medical-educational institute)	**EEAP** (institution for children or adolescents with disabilities) **IME** (medical-educational institute) **CAFS** (specialized family reception centre)
Adult		**AAH** (disabled adult allowance > 20 years old) **PCH** (disability compensation benefit)	**SAVS** (social support service) **SAMSAH** (medical-social support service for disabled adults) **SSIAD** (home nursing care service) **SAAD** (home help and accompaniment service) **SPASAD** (multi-purpose home help and care service)	**FAM** (nursing home) **MAS** (specialized care home)	**FAM** (nursing home) **MAS** (specialized care home)

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