Protocole National de Diagnostic et de Soins (PNDS)

Délétion 1p36

CHU de Reims
Centre de référence Anomalies du Développement et Syndromes Malformatifs de l'Interrégion Est,
Filière AnDDI-Rares

ARGUMENTAIRE DU PNDS
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Cet argumentaire a été élaboré par le centre de référence du CHU de Reims. Il a servi de base à l'élaboration du PNDS Délétion 1p36.
Le PNDS est téléchargeable sur le site du centre de la filière AnDDI-Rares (http://www.anddi-rares.org/).

Sommaire

Liste des abréviations

ACPA	Analyse Chromosomique sur Puce à ADN
ACTH	Adrénocorticotrophine
ADN	Acide Désoxyribonucléique
AJPP	Allocation Journalière de Présence Parentale
ALD	Affection de Longue Durée
AMM	Autorisation de Mise sur le Marché
CAMSP	Centre d'Action Médico-Sociale Précoce
CIA	Communication InterAtriale
CIV	Communication InterVentriculaire
CMPP	Centre Médico Psycho Pédagogique
CPDPN	Centre Pluridisciplinaire de Diagnostic Prénatal
CRMR (-AD)	Centre de référence Maladies rares (- Anomalies du Développement)
DI	Déficience Intellectuelle
EEG	Electroencéphalogramme
ERHR	Equipes Relais Handicap Rares
ETP	Education thérapeutique
FISH	Fluorescence In Situ Hybridization : L'hybridation in situ en fluorescence
FISH	Hybridation in situ en sonde fluorescente
HAS	Haute Autorité de Santé
IMC	Indice de Masse Corporelle
IME	Institut Médico-Educatif
IMG	Interruption Médicale de Grossesse
Inv dup	Inverted duplication
IRM	Imagerie par Résonance Magnétique
MDPH	Maison départementale des personnes handicapées
MLPA	Multiplex Ligation dependent Probe Amplification
PAG	Petit Poids pour l'âge gestationnel
PC	Périmètre Cranien
PCR / qPCR	Polimérase Chain Reaction / Quantitative Polimérase Chain Reaction
PEA	Potentiels Evoqués Auditifs
PNDS	Protocole National de Diagnostic et de Soins
RCIU	Retard de Croissance Intra-Utérin
RGO	Reflux Gastroœsophagien
SESSAD	Service d'Éducation Spéciale et de Soins à Domicile

Préambule

Le PNDS sur la délétion 1p36 a été élaboré selon la « Méthode d'élaboration d'un protocole national de diagnostic et de soins pour les maladies rares » publiée par la Haute Autorité de Santé en 2012 (guide méthodologique disponible sur le site de la HAS : www.has-sante.fr). Le présent argumentaire comporte l'ensemble des données bibliographiques analysées pour la rédaction du PNDS.

Il s'est appuyé sur la bibliographie internationale, sur les données clinique d'une cohorte de 96 patients élaborée en France et sur l'avis d'un groupe multidisciplinaire de professionnels concernés par le thème de ce PNDS.

Méthode de travail

La méthode utilisée pour l'élaboration de ce protocole national de diagnostic et de soins (PNDS) est celle des « Recommandations pour la pratique clinique » [1]. Elle repose, d'une part, sur l'analyse et la synthèse critiques de la littérature médicale disponible, et, d'autre part, sur l'avis d'un groupe multidisciplinaire de professionnels concernés par le thème du PNDS. La thèse du DR Clémence Jacquin basée sur ce travail été soutenue en septembre 2020.

Rédaction du PNDS

Un groupe de rédaction a été constitué par le CRMR du CHU de Reims. Après analyse et synthèse de la littérature médicale et scientifique pertinente, le groupe de rédaction rédige une première version du PNDS qui a été soumise à un groupe de lecture multidisciplinaire et multiprofessionnel. Il est composé de professionnels de santé, ayant un mode d'exercice public ou privé, d'origine géographique ou d'écoles de pensée diverses, d'autres professionnels concernés et de représentants d'associations de patients et d'usagers. Il a été consulté par mail, visioconférence et téléphone durant la période de COVID et a donné un avis sur le fond et la forme du document, en particulier sur la lisibilité et l'applicabilité du PNDS. Les commentaires du groupe de lecture ont été analysés et discutés par le groupe de rédaction qui a rédigé la version finale du PNDS.

[1] Cf. Les recommandations pour la pratique clinique - Base méthodologique pour leur réalisation en France. Anaes, 1999.

Argumentaire

1 Recherche documentaire

1.1 Bases de données bibliographiques

La recherche documentaire via Pubmed, a été réalisée en en utilisant successivement les **mots clefs** suivants :

- 1p36
- Deletion
- Syndrome
- Monosomy
- 1p36 deletion syndrome
- Monosomy 1p36

Analyse de la bibliographie et recrutement de la cohorte 2018-2021.

1.2 Stratégie de recherche

Tableau : Stratégie de recherche documentaire

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles retenus
Deletion	Sans limite-10/08/2021	291,400		
Deletion 1p36	Sans limite- 2021	857		
Deletion 1p36 syndrome	Sans limite- 2021	234	156	49
Monosomy 1p36	Sans limite- 2021	90	83	50
TOTAL			156 (doublons)	51

Non retenus :

- Les articles concernant uniquement des délétions en lien avec des translocations ou des remaniements complexes du chromosome 1 et autres chromosomes
- Les articles sans cartographie des délétions
- Les articles concernant des délétions acquises
- Les articles dans une langue différente du français ou de l'anglais
- Les articles concernant la méthode de détection des CNVs
- Les articles sans abstract
- Les articles traitant du dépistage prénatal
- Les articles traitant uniquement de mutation de gènes de la région 1p36
- Les articles traitant de techniques obsolètes
- Les articles taritant de délétions intragéniques

1.3 Critères de sélection des articles

* date de début et fin de la recherche, bases de données, mots clés renseignés

Tableau 1. Recommandations de bonne pratique

Auteur, année, référence, pays	Objectif	Stratégie de recherche bibliographique renseignée (oui/non)*	Recueil de l'avis des professionnels (non, oui, lesquels)	Recueil de l'avis des patients (non, oui)	Populations et techniques (ou produits) étudiées	Résultats (avec grade des recommandations si disponible)
Battaglia A et al., GeneReviews®, 2008 Feb 1 [updated 2013 Jun 6] PMID: 20301370 USA **1p36 Deletion Syndrome** - RETIRED CHAPTER, FOR HISTORICAL REFERENCE ONLY.	Guide	non	Oui	Non		Bilan de parcours
Sangu N et al., Congenit Anom, 2014 May;54(2):82-6. doi:10.1111/cga.12029. Japon Growth patterns of patients with **1p36 deletion syndrome**.	Description	Oui Rétrospectif	Oui	Non	44 patients	Bilan Retard de croissance ou obèsité
Carter LB et al., Am J Med Genet A, 2019 Aug;179(8):1543-1546. doi:10.1002/ajmg.a.61266. USA Perinatal distress in **1p36 deletion syndrome** can mimic hypoxic ischemic encephalopathy.	Evaluer la période périnatale	Etude retrospective	Oui	Non	72 patients 2 groupes avant et après terme	Observation de défaillance périnatale dans la majorité
Verrotti A et al., Acta Neurol Scand, 2018 Dec;138(6):523-530. doi: 10.1111/ane.13006 Italie **Electroclinical features of epilepsy** monosomy 1p36 syndrome **and their implications.**	Prevention	Etude retrospective	Oui	Non	22 patients avec epilepsie	Fréquence des spasmes infantiles Pharmaco-résistance
Stagi S et al., BMC Med Genet, 2014 Jan 30;15:16.doi: 10.1186/1471-2350-15-16 Italie Type II diabetes and impaired glucose tolerance due to severe hyperinsulinism in patients with **1p36 deletion syndrome** and a Prader-Willi-like phenotype.	Description		Oui	Non	2 enfants	Obèsité Hyperphagie Diabète
Vieira GH et al., Am J Med Genet A, 2011 May;155A(5):988-92. doi: 10.1002/ajmg.a.33960 USA Differential diagnosis of Smith-Magenis **syndrome**: **1p36 deletion syndrome**.	Description				1 cas avec del 1p36.32p36.33	Non retenu comme diagnostic différentiel
Bursztejn AC et al., Am J Med Genet A, 2009 Nov;149A(11):2493-500. France Molecular characterization of a **monosomy 1p36** presenting as	Description				1 cas avec remaniement complexe	Non retenu comme diagnostic différentiel

an Aicardi **syndrome** phenocopy.						
Robinson DM et al., Am J Med Genet A, 2008 Jun 15;146A(12):1571-4. doi:10.1002/ajmg.a.32096 USA Young-Simpson **syndrome** (YSS), a variant of del(1)(p36) **syndrome**?	Description				1 cas	Non retenu comme diagnostic différentiel
Battaglia A et al., GeneReviews®, 2008 Feb 1 [updated 2013 Jun 6]. PMID: 20301370 USA 1p36 Deletion Syndrome - RETIRED CHAPTER, FOR HISTORICAL REFERENCE ONLY.	Guide	Non	Oui	Non		Bilan de parcours

Tableau 2. Revues systématiques de la littérature

Auteur, année, référence, pays	**Objectif**	**Stratégie de recherche renseignée (oui/non)***	**Critères de sélection des études**	**Populations et techniques (ou produits) étudiées**	**Critères d'évaluation**	**Résultats et signification**
Jordan VK et al., Appl Clin Genet, 2015 Aug 27;8:189-200.doi: 10.2147/TACG.S65698 USA **1p36 deletion syndrome**: an update.	Review	Oui	Cartographie 1p36 Par FISH ou ACPA	25 articles 1987 - 2015	Carte Hg19	Relation génotype phénotype des gènes de la régions et modèles animaux
Gajecka M et al., Am J Med Genet C Semin Med Genet, 2007 Nov 15;145C(4):346-56.doi: 10.1002/ajmg.c.30154 USA **Monosomy 1p36 deletion syndrome**.	Review	Non	Deletion 1p36	44 articles 1981-2007 Environ 140 patients	Rétrospectifs	Description clinique, génétique, génique
Battaglia A. Brain Dev, 2005 Aug;27(5):358-61. doi:10.1016/j.braindev.2004.03.011 Italie Del 1p36 syndrome: a newly emerging clinical entity	Review		Monosomy 1p36		Nouveaux cas	Description clinique 0,5% a 1,2% des retards mentaux idiopathiques
Brazil A et al., Am J Med Genet A, 2014 Oct;164A(10):2496-503. doi: 10.1002/ajmg.a.36657 USA Delineating the phenotype of **1p36 deletion** in adolescents and adults.	Review	Oui	Patients >12 ans	40 patients Adultes et ado	Questionnaire rempli	Description clinique
Rocha et al., Genet Mol Res, 2016 Feb 22;15(1). doi:10.4238/gmr.15017942. Mini-Review: **Monosomy 1p36 syndrome**: reviewing the correlation between **deletion** sizes and phenotypes.	Review	Oui	Clinique et mesure de la taille de la délétion Deletion 1p36 pure	17 articles Entre 1999 et 2014 29 patients 20 femmes	rétrospectif	Relation génotype phénotype 1p36.11-p36. 33
Slavotineck A et al., J Med Genet 1999 ;36 :657-663	Review	Oui	Terminal deletion 1p	18 articles Entre 1987 et 1999	rétrospectif	Contiguous deletion syndrome clinique

Tableau 2. Revues systématiques de la littérature

Auteur, année, référence, pays	Objectif	Stratégie de recherche renseignée (oui/non)*	Critères de sélection des études	Populations et techniques (ou produits) étudiées	Critères d'évaluation	Résultats et signification
UK/USA Monosomy 1p36				39 patients		
Shimada S et al., Brain Dev, 2015 May;37(5):515-26.doi: 10.1016/j.braindev.2014.08.002 Microarray analysis of 50 patients reveals the critical chromosomal regions responsible for 1p36 deletion syndrome-related complications.	Description	Cartographie par ACPA		50 nouveaux cas sur 86 patients		Relation génotype phénotype Dont 3 cas de mosaique
Wu YQ et al., Hum Mol Genet, 1999 Feb;8(2):313-21.doi: 10.1093/hmg/8.2.313 USA Molecular refinement of the **1p36 deletion syndrome** reveals size diversity and a preponderance of maternally derived deletions.	Description	Etude prospective	FISH et microsatellites	30 cas		Relation génotype phénotype par cartographie
Kang SH et al., Clin Genet, 2007 Oct;72(4):329-38. doi: 10.1111/j.1399-0004 Identification of proximal **1p36** deletions using array-CGH: a possible new **syndrome**.	Description	oui	Interstitielle 1p36.23-p36.11	5 patients	prospectif	Relation génotype phénotype New syndrome
Giannikou K et al., Gene, 2012 Sep 15;506(2):360-8. doi: 10.1016/j.gene.2012.06.060 Grece Further delineation of novel **1p36** rearrangements by array-CGH analysis: narrowing the breakpoints and clarifying the "extended" phenotype.	Description	ACPA		7 cas		Relation génotype phénotype 7 cas dont 1 cas hérité
Buck A et al., Am J Med Genet A, 2011 Dec;155A(12):3164-9. doi: 10.1002/ajmg.a.34333 Allemagne Minimal genotype--phenotype correlation for small deletions within distal **1p36**.	Description			1 cas	Rejeté association del et dup	Relation génotype phénotype
Aagaard Nolting L et al., Clin Genet, 2020 Jun;97(6):927-932 doi:10.1111/cge.13739 Danemark A new **1p36**.13-**1p36**.12 microdeletion **syndrome** characterized by learning disability, behavioral abnormalities, and ptosis.	Description	Oui Cas chevauchants	Decipher Del <5 Mb	7 cas	Rejeté car interstitiel	Relation génotype phénotype Nouveau syndrome del 1p36.23p36.22

Tableau 3. Etudes cliniques

Auteur, année, référence, pays	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et signification
Yokoyama E et al., Mol Cytogenet, 2020 Sep 7;13:42. doi: 10.1186/s13039-020-00510-5 Mexique Non-classical **1p36 deletion** in a patient with Duane retraction **syndrome**: case report and literature review.	Review	Cartograhie Région 1p36.31p36.21 Gène GES3	1 cas			Duane syndrome
Murakoshi M et al., Am J Med Genet A, 2017 Feb;173(2):495-500. doi: 10.1002/ajmg.a.38020 Japon Abdominal paraganglioma in a young woman with **1p36 deletion syndrome**.	Description		1 cas			paragangliome
Battaglia A et al., Pediatrics, 2008 Feb;121(2):404-10. doi:10.1542/peds.2007-0929 Italie Further delineation of deletion 1p36 syndrome in 60 patients: a recognizable phenotype and common cause of developmental delay and mental retardation	Description	Prospectif	60 nouveaux cas 41 female, 19 male			Description clinique
Tandy PA. Intern Med J, 2012 Sep;42(9):1037-9. doi: 10.1111/j.1445-5994 Australie Dying at 23 with **1p36 deletion syndrome**: Laura's family story.	Description		1 cas de 23 ans			Risque vital Arret cardiaque
Okamoto N et al., J Hum Genet, 2002;47(10):556-9. doi: 10.1007/s100380200085 Japon A girl with **1p36 deletion syndrome** and congenital fiber type disproportion myopathy.	Description	FISH Deletion de SKI	1 cas de 4 ans			myopathy
Shimada S et al., Am J Med Genet A, 2014 Feb;164A(2):415-20. doi: 10.1002/ajmg.a.36304 Mild developmental delay and obesity in two patients with mosaic **1p36 deletion syndrome.**	Description	ACPA FISH	2 cas en mosaique			Obesity mosaicisme
Zagalo A et al., BMJ Case Rep, 2012 Mar 20;2012:bcr0120125503. doi:10.1136/bcr.01.2012.5503 Portugal Morbid obesity in a child with monosomy 1p36 syndrome.	Description	FISH Der(1)t(1 ;2)	1		Rejeté	obesity
Tsuyusaki Y et al., Pediatr Int, 2010 Aug;52(4):547-50. doi: 10.1111/j.1442-200X.2010.03090.x Japon **1p36 deletion syndrome** associated with Prader-Willi-like phenotype.	Description	FISH	2			obesity
Minami K et al., Eur J Pediatr, 2005	Description	FISH	1			intestinal malrotation and

Tableau 3. Etudes cliniques

Auteur, année, référence, pays	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et signification
Mar;164(3):193-4.doi: 10.1007/s00431-004-1581-z Japon **1p36 deletion syndrome** with intestinal malrotation and annular pancreas.						annular pancreas.
Kawashima H et al., Pancreas, 2011 Jan;40(1):171-3. doi:10.1097/MPA.0b013e3181df1ca9 Japon A case of **1p36 deletion syndrome** accompanied with anomalous arrangement of the pancreaticobiliary duct.	Description		1			pancreas
Heilstedt A et al., Am J Hum Genet, 2003 May;72(5):1200-12. doi: 10.1086/375179. USA Physical map of 1p36, placement of breakpoints in monosomy 1p36, and clinical characterization of the syndrome	Cartographier	Cartographie par marqueurs microsatellites et FISH	61 patients		observation	Relation génotype phénotype
Subin Jang et al., Front Pediatr, 2021 Jun 7;9:653633. doi: 10.3389/fped.2021.653633 USA 1p36 Deletion Syndrome and Left Ventricular Non-compaction Cardiomyopathy-Two Cases Report	Description		2 patients			LVNC transplantation
Thienpont B et al., Eur J Med Genet, May-Jun 2007;50(3):233-6 doi:10.1016/j.ejmg.2007.01.002 Belgique Left-ventricular non-compaction in a patient with **monosomy 1p36**.	Description	ACPA	1 patient			Heart 1er cas de LVNC
Lee J et al., Methodist Debakey Cardiovasc J, Oct-Dec 2014;10(4):258-9. doi: 10.14797/mdcj-10-4-258 USA Left ventricular noncompaction cardiomyopathy: adult association with **1p36 deletion syndrome**.	Evaluation LVNC	Case report	1 patient		NA	Heart 1st case
Halpern AV et al., J Am Acad Dermatol, 2006 Nov;55(5 Suppl):S98-9. doi: 10.1016/j.jaad.2005.09.035 Pemphigus vulgaris in a patient with **1p36 deletion syndrome**.	Description		1 patient			dermato
Zhang Z et al., J Dermatol, 2018 Jul;45(7):871-873. doi: 10.1111/1346-8138.14311 Cutis laxa in a patient with 1p36 deletion syndrome.	Description	CGH et exome 1p36.33p36.32	1 patient			Dermato Nouveau symptome
Kanabar G et al., Seizure, 2012 Jun;21(5):402-6. doi:10.1016/j.seizure.2012.02.004 UK	Description	retrospective	4 patient			Apnee et Epilepsie

Tableau 3. Etudes cliniques

Auteur, année, référence, pays	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et signification
Multiple causes of apnea in **1p36 deletion syndrome** include seizures.						
Saito Y et al., Brain Dev, 2011 May;33(5):437-41. doi: 10.1016/j.braindev.2010.07.004. Japon Polymicrogyria and infantile spasms in a patient with **1p36 deletion syndrome**.	Description	Der(1)t(1 ;4)	1 cas		Rejeté, remaniement complexe	Epilepsie Spasme infantile et polymicrgyrie
Bahi-Buisson N et al., Epilepsia, 2008 Mar;49(3):509-15. doi: 10.1111/j.1528-1167.2007.01424.x France Spectrum of epilepsie in terminal 1p36 deletion syndrome	Review 6 article et cas	Etude rétrospective 6 article	91 patients dont 64 feminin et 80 deletions pures			Epilepsie précoce
Kurosawa K et al., Brain Dev, 2005 Aug;27(5):378-82. doi: 10.1016/j.braindev.2005.02.004 Japon Epilepsy and neurological findings in 11 individuals with 1p36 deletion syndrome	Description		11 patients			Epilepsie et traitement
Çöllü M et al., Am J Med Genet A, 2016 Jul;170(7):1889-94 doi: 10.1002/ajmg.a.37666 Turquie Is **1p36 deletion** associated with anterior body wall defects?	Description	Carte : gènes NOCCL2, DVL1, MMP23B	1 cas			2eme cas d'extrophie vésicale avec del 1p36
Nistico' D et al., BMC Pediatr, 2020 May 9;20(1):201 doi: 10.1186/s12887-020-02049-1 Italie Dental anomalies as a possible clue of **1p36 deletion syndrome** due to germline mosaicism: a case report.	Description		2 sisters		Interstitial mosaicism	dents

Tableau 4. Etudes des gènes

Auteur, année, référence, pays	Objectif	Méthodologie, niveau de preuve	Population		Critères de jugement	Résultats et signification
Baranek C and Atanasoski S Epigenetics, 2012 Jul;7(7):676-9. doi: 10.4161/epi.20590 Suisse Modulating epigenetic mechanisms: the diverse functions of Ski during cortical development.	Thèse sur SKI				Rejeté trop général	SKI
Kim BJ et al., PLoS One, 2013;8(2):e57460. doi: 10.1371/journal.pone.0057460 USA An allelic series of mice reveals a role for RERE in the development of multiple organs affected in **chromosome 1p36** deletions.	RERE gene	Modèle animal	Modèle animal	Del 1p36 prox		RERE Association aux fentes oro-faciales

El Waly B et al., Eur J Hum Genet, 2020 Dec;28(12):1703-1713. doi: 10.1038/s41431-020-0659-z France Molecular characterization of a **1p36** chromosomal duplication and in utero interference define ENO1 as a candidate gene for polymicrogyria.	ENO1		1 cas		rejeté	Délétion centrée sur ENO1
Colmenares C et al., Nat Genet, 2002 Jan;30(1):106-9. doi: 10.1038/ng770 USA Loss of the SKI proto-oncogene in individuals affected with **1p36 deletion syndrome** is predicted by strain-dependent defects in Ski-/- mice.	Recherche gène SKI	Modèle animal	Modèle animal			SKI et fente faciale
Heilstedt HA et al., Epilepsia, 2001 Sep;42(9):1103-11. doi: 10.1046/j.1528-1157.2001.08801.x USA Loss of the potassium channel beta-subunit gene, KCNAB2, is associated with epilepsy in patients with **1p36 deletion syndrome**.	Description	FISH	24 patients dont 9 avec la del 1p36			KCNAB2 : suspicion de lien avec l'épilepsie
Perkowski JJ and Murphy GG. J Neurosci, 2011 Jan 5;31(1):46-54. doi:10.1523/JNEUROSCI.2634-10.2011 USA **Deletion** of the mouse homolog of KCNAB2, a gene linked to **monosomy 1p36**, results in associative memory impairments and amygdala hyperexcitability.	Recherche	Modèle murin de délétion de KCNAB2	Modèle murin			KCNAB2 impliqué dans la mémorisation et l'aprentissage
García-López J et al., Cell Rep, 2020 Jan 14;30(2):454-464.e5 doi:10.1016/j.celrep.2019.12.048 USA Large **1p36** Deletions Affecting Arid1a Locus Facilitate Mycn-Driven Oncogenesis in Neuroblastoma.	Description				Rejeté pathologie acquise	ARID1A
Arndt AKet al., Am J Hum Genet, 2013 Jul 11;93(1):67-77 doi: 10.1016/j.ajhg.2013.05.015 Allemagne Fine mapping of the **1p36 deletion syndrome** identifies mutation of PRDM16 as a cause of cardiomyopathy.	Cartographie des patients avec cadiomyopathie	Cartographie et etude fonctionnelle	75 patients prospective and retrospective			PRDM16
Frejeau B et al., Am J Hum Genet, 2016 May 5;98(5):963-970 doi: 10.1016/j.ajhg.2016.03.002 USA De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions	Recherche	Modèle animal Del 1p36 prox	Modèle animal			Fentes orofaciales

Tableau 5. Etudes anténatale						
Auteur, année, référence, pays	**Objectif**	**Méthodologie, niveau de preuve**	**Population**		**Critères de jugement**	**Résultats et signification**
Chen CP et al., Taiwan J Obstet Gynecol, 2010 Dec;49(4):473-80..doi: 10.1016/S1028-4559(10)60100-3 Taiwan **Chromosome 1p36 deletion syndrome**: prenatal diagnosis, molecular cytogenetic characterization and fetal ultrasound findings.	Description	Derivé t(1 ;10)	1 cas		**Rejeté remaniement complexe**	Signes echographique
Guterman S et al., Prenat Diagn, 2019 Sep;39(10):871-882. doi: 10.1002/pd.5498 Prenatal findings in 1p36 deletion syndrome: New cases and a literature review	Review	Description et cartographie	10 cas de 2012 a 2017			Signes d'appel échographique
Zhang X et al., J Matern Fetal Neonatal Med, 2021 Jul;34(13):2180-2184. doi: 10.1080/14767058.2019.1660764 Epub 2019 Sep 4. Chine Prenatal detection of **1p36 deletion syndrome**: ultrasound findings and microarray testing results.		Etude retrospective cartographie	10 cas			Signes d'appel échographique
Toshimitsu M et al., Case Reports in Obstetrics and Gynecology, Volume 2019, Article ID 6753184 Japon DOI: 10.1155/2019/6753184 Exome-First Approach in Fetal Akinesia Reveals Chromosome 1p36 Deletion Syndrome	Description	Approche par exome	1 cas			Carte Del 1p36.33p36.22
Chawla V et al., J Investig Med High Impact Case Rep, 2018 Jul 24;6:2324709618790613. doi.org/10.1177/2324709618790613 USA A Novel Case of Biliary Atresia in a Premature Neonate With 1p36 Deletion Syndrome	Description		1 cas			Atrésie biliaire

2 Argumentaire sur le planning de la rédaction

Le syndrome de la délétion ou monosomie 1p36 (OMIM:607862 et ORPHA:1606) est l'un des syndromes de délétion terminale le plus fréquent 1/5000 a 1/10000 nouveau né vivant [Shimada 2014]. Cela représente 0,5 to 1,2% des cas de DI syndromique [Guterman 2019, Battaglia genereview 2008 updated 2021, Heilstedt 2003].

Ce syndrome est du à une délétion hétérozygote du bras court (p) du chromosome 1. On observe Les délétions pures (52-67%), les délétions interstitielles (9.7-29%), les délétions dérivées de remaniements de chromosomes (7-16.4%), et de réarrangements plus complexes (12%) [Battaglia generewiew 2008 updated 2021, Rocha 2016]. Le syndrome de la deletion 1p36 du Chromosome 1 peut être diagnostiquée par les techniques de la cytogénétique, notamment la technique d'hybridation *in situ* en Fluorescence fluorescence (FISH) et la CGH-array ou ACPA. La technique de l'ACPA permet l'identification de

remaniements complexes, [Battaglia generewiew 2008 updated 2021, Rocha 2016]. La technique d'étude du génome permet aujourd'hui également de faire ce diagnostic.

Afin de rédiger ce PNDS initié en 2018 nous avons repris les données de la littérature et les données des travaux réalisés au sein de l'association des Cytogénéticiens de Langue Française. Ainsi nous avons pu recruter une grande cohorte de patients français en prénatal et en postnatal. La cohorte des observations de prénatal a été publiée en 2019: "Prenatal findings in 1p36 deletion syndrome: New cases and a literature review. » *Guterman S, Beneteau C, Redon S, Dupont C, Missirian C, Jaeger P, Herve B, Jacquin C, Douet-Guilbert N, Till M, Tabet AC, Moradkhani K, Malan V, Doco-Fenzy M, Vialard F*. Prenat Diagn. 2019 Sep;39(10):871-882. doi: 10.1002/pd.5498. Epub 2019 Jul 5. PMID: 31172545 » ; La deuxième cohorte des patients en postnatal formée de 86 patients à fait l'objet d'un manuscript décrivant les signes cliniques de cette grande cohorte. Le Dr Clémence Jacquin a soutenu sa thèse sur ce sujet à la faculté de medecine de Reims le 25 septembre 2020. L'article collaboratif est soumis, en revision dans la revue Am J Med Genet "1p36 deletion syndrome: review and mapping with further characterization of the phenotype, a new cohort of 86 patient » *C. Jacquin, E. Landais, C. Poirsier, M. Descharmes, M. Spodenkiewick, L. Herissant, D. Sanlaville, P. Jaeger, D. Bosquet, AC. Tabet, J. Levy, A. Verloes, A. Bonnard, JM. Dupont, A. Heddar, B. Isidor, C. Lecaignec, D. Martin Coignard, N. Gruchy, C. Missirian, G. Jouret, G. Vieville, P. Callier, L. Burglen, A. Afenjar, S. Chantot, C. Mignot, N. Lopez, D. Heron, B. Keren, J. Puechbertty, C. Coubes, V. Gatinois, L. Pinson, B. Delobel, Z. Manssens, P. Kuentz, S. Redon, C. Pebrel-Richard, C. Yardin, S. Guterman, F. Vialard, M. Doco-Fenzy.*

Ce travail a permis de montrer que la délétion 1p36 est une maladie rare avec une fréquence 10 fois inférieure a celle de la délétion 22q11. Nous avions publié une cohorte de 700 patients avec une délétion 22q11 en 2016 (*Poirsier C, Besseau-Ayasse J, Schluth-Bolard C, Toutain J, Missirian C, Le Caignec C, Bazin A, de Blois MC, Kuentz P, Catty M, Choiset A, Plessis G, Basinko A, Letard P, Flori E, Jimenez M, Valduga M, Landais E, Lallaoui H, Cartault F, Lespinasse J, Martin-Coignard D, Callier P, Pebrel-Richard C, Portnoi MF, Busa T, Receveur A, Amblard F, Yardin C, Harbuz R, Prieur F, Le Meur N, Pipiras E, Kleinfinger P, Vialard F, Doco-Fenzy M. A French multicenter study of over 700 patients with 22q11 deletions diagnosed using FISH or aCGH. Eur J Hum Genet. 2016 Jun;24(6):844-51).* Ce travail préparatoire a permis de mettre en évidence des points de vigilence concernant les patients porteurs de ces délétions 1p36. En particulier la fréquence des décès précoces en lien avec des cardiomyopathies ou avec des malformations cardiaques.

Les signes cliniques peuvent etre également différents en fonction de la position de la déétion (distale ou proximale) avec des malformations cardiaques, des troubles du comportement et une épilepsie plus fréquent dans les délétions distales et la microcéphalie plus fréquente dans les délétions proximales.

Le délais pour la rédaction de ce PNDS s’explique par tout ce travail réalisé en amont par tous les partenaires qui sont à la base du diagnostic et du suivi des patients.

Annexe 1. Liste des participants

Ce travail a été coordonné par le Pr Martine DOCO-FENZY, Centre de référence du CHU de REIMS - Centre de référence Anomalies du Développement et Syndromes Malformatifs de l'Interrégion Est.

Ont participé à l'élaboration du PNDS :

Rédacteurs & Groupe de travail multidisciplinaire

- Pr Martine DOCO-FENZY, génétique, REIMS ;
- Dr Céline Poirsier, génétique, REIMS
- Dr Clémence Jacquin, Génétique, REIMS ;
- Pr Nathalie Bednarek, neuropédiatrie, REIMS ;
- Dr Ahmad Akhavi, cardio pédiatre, REIMS ;
- Mme Isabelle MARCHETTI-WATERNAUX, présidente de l'association Valentin-APAC ;
- Dr Pierre François Souchon, endocrino pédiatre, REIMS ;
- Pr Nadia Bahi-Buisson neurologue, Necker, PARIS ;
- Dr Anne Sophie David, médecin traitant, NANTES.

Groupe de relecture

- Dr Elise SCHAEFFER, Généticienne, STRASBOURG ;
- Pr Florence PETIT, Généticienne, LILLE ;
- Pr Anne-catherine ROLLAND, pédopsychiatre, REIMS ;
- Pr Annick TOUTAIN, Généticienne, TOURS ;
- Pr Laurence FAIVRE, Généticienne, DIJON ;
- Dr Claire BENETEAU, Pédiatre Généticienne, NANTES ;
- Dr Guylène LE MEUR, MCU-PH en Ophtalmologie, NANTES.

Déclarations d'intérêt

Tous les participants à l'élaboration du PNDS ont rempli une déclaration d'intérêt. Les déclarations d'intérêt sont en ligne et consultables sur le site internet de la filière AnDDI-Rares.

Modalités de concertation du groupe de travail multidisciplinaire

Réunions visioconférence ou e-meeting

- Nombreux échanges par mail ou téléphone car petit nombre de patients environ 600 à 700 publiés en postnatal

- Dates des réunions :

  - 2018- 17 septembre
  - 2020- 10 aout
  - 2020- 25 septembre
  - 2020- 24 novembre
  - 2021- 28 juin
  - 2022 - 06 février
  - 2022 – 14 juin
  - 2022 – 21 juin

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