Protocole National de Diagnostic et de Soins (PNDS)

Arthrogryposes Multiples Congénitales

Argumentaire

Centres de Référence :
Anomalies du développement et Syndromes Malformatifs et Maladies neuromusculaires rares

Filières maladies rares AnDDI-Rares et FILNEMUS

Septembre 2021

Cet argumentaire a été élaboré par le centre de référence Anomalies du développement et Syndrome Malformatifs. Il a servi de base à l'élaboration du PNDS « Arthrogryposes multiples congénitales ».
Le PNDS est téléchargeable sur le site de la Haute Autorité de Santé
www.has-sante.fr

Sommaire

Liste des abréviations

ALD	Affection de Longue Durée
AMM	Autorisation de Mise sur le Marché
HAS	Haute Autorité de Santé
PNDS	Protocole National de Diagnostic et de Soins

Préambule

Le PNDS sur les Arthrogryposes Multiples Congénitales 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.

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.

Rédaction du PNDS

Un groupe de rédaction est constitué par le Centre de Référence Anomalies du Développement et Syndrome Malformatifs. Après analyse et synthèse de la littérature médicale et scientifique pertinente, le groupe de rédaction rédige une première version du PNDS. Cette version est soumise à un groupe de relecteurs multidisciplinaires. 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 est consulté lors d'une réunion physique ou virtuelle et donne 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 relecture sont ensuite analysés et discutés par le groupe de rédaction qui rédige la version finale du PNDS. Une réunion physique est organisée en cas de besoin.

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

Argumentaire

Recherche documentaire

Sources consultées	Bases de données : PUBMED, Orphanet, OMIM
Période de recherche	Non limitée, sauf précisé autrement
Langues retenues	Anglais/français/allemand
Mots clés utilisés	Cf. stratégie de recherche
Nombre d'études recensées	2444
Nombre d'études retenues	430

Stratégie de recherche

Tableau : Stratégie de recherche documentaire

AMC + diagnosis

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles ajoutés manuellement	Nombre d'articles retenus
Arthogryposis multiplex congenita AND Chromosomal microarray	2003 – 2019	144	44	0	44
Arthogryposis multiplex congenita AND whole exome sequencing	2009 – 2019	33	33	0	33
Arthogryposis multiplex congenita AND SMN1	1996 – 2019	88	5	1	6
Arthogryposis multiplex congenita AND DMPK	1993 – 2019	20	5	0	5
Arthogryposis multiplex congenita AND Prader-Willi syndrome	1983 – 2019	7	2	0	2
Arthogryposis multiplex congenita AND electromyography	1999 – 2019	80	14	0	14
Arthogryposis multiplex congenita AND creatine kinase	1999 – 2019	40	13	1	14
Arthogryposis multiplex congenita AND muscle	2009 – 2019	101	33	0	33

biopsy					
Distal arthrogryposis AND muscle biopsy	1982 – 2019	62	14	2	16
Amyoplasia AND muscle biopsy	1933 – 2019	17	11	1	2
Arthogryposis multiplex congenita AND spinal MRI	1999 – 2019	38	1	0	1
Arthogryposis multiplex congenita AND muscle MRI	1999 – 2019	41	24	0	5
Arthogryposis multiplex congenita AND myasthenia	1964 – 2019	76	24	6	30
Arthogryposis multiplex congenita AND aminoacidopathies	1950 – 2019	13	1	2	3
Arthogryposis multiplex congenita AND oxidation metabolic disorders	1967 – 2019	6	0	0	0
Arthogryposis multiplex congenita AND citric acid cycle	1970 – 2019	7	0	2	2
Arthogryposis multiplex congenita AND mitochondrial diseases	1970 – 2019	60	12	4	16
Arthogryposis multiplex congenita AND peroxisomal disorders	1990 – 2019	11	1	0	1
Arthogryposis multiplex congenita AND lysosomal disorders	1985 – 2019	14	5	0	5
Arthogryposis multiplex congenita AND metabolic	1964 – 2019	97	3	6	9
Arthogryposis multiplex congenita AND heart AND congenital anomaly	1955 – 2019	95	6	0	6
Arthogryposis multiplex congenita AND ophthalmology AND abnormalities	1989 – 2019	11	7	0	7
Arthogryposis multiplex congenita AND tethered spinal cord	1989 – 2019	3	1	0	1
Arthogryposis multiplex congenita AND neural tube defects	1980 – 2019	74	9	0	9
Total		**1138**	**268**	**25**	**264**

AMC + Medecine Physique et Réadaptation

Thème / mot(s) clé(s)	Période	Nombre total de références	Nombre d'articles	Nombre d'articles	Nombre d'articles

		obtenues	analysés	ajoutés manuellement	retenus
Arthrogryposis multiplex congenita AND braces	all	21	10	0	8
Arthrogryposis multiplex congenita AND casting	all	30	6	0	5
Arthrogryposis multiplex congenita AND clinical care	all	68	10	0	6
Arthrogryposis multiplex congenita AND gait analysis	all	13	5	0	5
Arthrogryposis multiplex congenita AND neurostimulation	all	1	0	0	0
Arthrogryposis multiplex congenita AND orthoses	all	49	6	0	4
Arthrogryposis multiplex congenital AND medical NOT orthopaedic	all	64	3	7	2
Arthrogryposis multiplex congenita AND physiotherapy	all	82	5	0	2
Arthrogryposis multiplex congenita AND scoliosis AND medical	all	50	1	0	1
Arthrogryposis multiplex congenita AND treatment AND medical NOT orthopaedic	all	121	4	1	3
Arthrogryposis multiplex congenita AND treatment AND recommendations	all	5	3	2	4
arthrogryposis multiplex congenita AND joint mobilization	all	20	10	0	0
Arthrogryposis multiplex congenita AND wheelchair	All	13	8	0	1
Arthrogryposis multiplex congenita AND participation restriction	All	1	1	0	0
Arthrogryposis multiplex congenital AND driver licence	All	0	0	0	0
Arthrogryposis multiplex congenital AND technical aids	All	3	3	0	0
TOTAL		**541**	**75**	**10**	**41**

<u>Amyoplasie + Medecine Physique et Réadaptation</u>

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles ajoutés manuellement	Nombre d'articles retenus
Amyoplasia AND braces	all	3	2	0	2
Amyoplasia AND clinical care	all	6	3	0	1
Amyoplasia AND gait analysis	all	2	0	0	0
Amyoplasia AND neurostimulation	all	0	0	0	0
Amyoplasia AND orthoses	all	8	0	0	0
Amyoplasia AND medical NOT orthopaedic	all	11	1	1	2
Amyoplasia AND scoliosis AND medical	all	2	0	0	0
Amyplasia AND treatment AND medical NOT orthopaedic	all	10	1	0	1
Amyoplasia AND treatment AND recommendations	all	0	0	0	0
TOTAL		**42**	**7**	**1**	**6**

AMC + chirurgie

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles ajoutés manuellement	Nombre d'articles retenus
Arthogryposis multiplex congenita AND surgery AND scoliosis	all	30	5	0	3
Arthrogryposis multiplex congenita AND surgery AND achilles tendon release	all	8	7	1	3
Arthrogryposis multiplex congenita AND surgery AND adducted thumb	all	2	2	0	2
Arthrogryposis multiplex congenita AND surgery AND anaesthesia	all	55	11	0	10
Arthrogryposis multiplex congenita AND surgery AND club feet	all	96	18	0	9
Arthrogryposis multiplex congenital AND surgery AND elbow	All	68	34	0	24

Arthrogryposis multiplex congenita AND surgery AND hip	all	73	17	4	11
Arthrogryposis multiplex congenita AND surgery AND hip dislocation	All	49	9	0	4
Arthrogryposis multiplex congenita AND surgery AND knee	all	77	30	1	23
Arthrogryposis multiplex congenita AND surgery AND muscle transfer	all	27	18	0	2
Arthrogryposis multiplex congenita AND surgery AND pain	all	42	11	0	0
Arthrogryposis multiplex congenita AND surgery AND pes convex	all	7	6	0	2
Arthrogryposis multiplex congenita AND surgery AND ponseti	all	9	2	2	1
Arthrogryposis multiplex congenita AND surgery AND pterygium	all	7	0	0	0
Arthrogryposis multiplex congenita AND surgery AND skin web	all	2	1	0	1
Arthrogryposis multiplex congenita AND surgery AND spinal arthrodesis	all	27	5	0	1
Arthrogryposis multiplex congenital AND durgery AND talectomy	All	13	10	0	8
Expert review or recos		-	-	15	2
TOTAL		**579**	**142**	**23**	**103**

AMC + douleur

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles ajoutés manuellement	Nombre d'articles retenus
Arthogryposis multiplex congenita AND pain	all	73	10	0	7

AMC + anesthesie

Thème / mot(s) clé(s)	Période	Nombre total de références obtenues	Nombre d'articles analysés	Nombre d'articles ajoutés	Nombre d'articles retenus

				manuellement	
Arthogryposis multiplex congenita AND anaesthesia	all	71	20	0	7

Sélection des articles

Dans un premier temps, le comité des rédacteurs a établi les mots clés de recherche biobliographique, puis sélectionné les articles sur abstract. Seuls les recommandations de bonnes pratiques, les articles de revue, les séries d'au moins 5 cas ou cas unique si le patient était génotypé ou avec diagnostic clinique certain et apportant des informations importantes pour établir des recommandations ont été retenus.
Dans un second temps, les articles sans rapport spécifique à l'arthrogrypose ou ne correspondant pas aux critères de sélection en format complet n'ont pas été retenus.
Certains articles jugés utiles pour la rédaction du PNDS, mais identifiés par la recherche bibliographique dans des thématiques sans rapport avec les mots clés spécifiques, ont été reclassé (« ajouté manuellement »).

Recommandations de bonnes pratiques

Tableau 1. Recommandations de bonne pratique

Référence	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)	Population et techniques (ou produits) étudiées	Résultats (avec grade des recommandations si disponible)
van Bosse HJP, Pontén E, Wada A, Agranovich OE, Kowalczyk B, Lebel E, Şenaran H, Derevianko DV, Vavilov MA, Petrova EV, Barsukov DB, Batkin SF, Eylon S, Kenis VM, Stepanova YV, Buklaev DS, Yilmaz G, Köse O, Trofimova SI, Durgut F. Treatment of the Lower Extremity Contracture/Deformities. J Pediatr Orthop. 2017 Jul/Aug;37 Suppl 1:S16-S23. doi: 10.1097/BPO.0000000000001005. PMID: 28594688.		non	oui	non	Traitement des luxations de hanche, flessum de genoux et malposition des pieds (PBVE, pied creux, pieds convexes)	Accord d'experts
Komolkin I, Ulrich EV, Agranovich OE, van Bosse HJP. Treatment of Scoliosis Associated With Arthrogryposis Multiplex Congenita. J Pediatr Orthop. 2017 Jul/Aug;37 Suppl 1:S24-S26. doi: 10.1097/BPO.0000000000000993. PMID: 28594689.		non	oui	non	corset jusqu'à 18 mois/corsets plâtrés/ chirurgies non fusion > 50° puis arthrodèse	Accord d'experts

Revues systématiques de la litérature

Tableau 2. Revues systématiques de la litérature

Référence	Objectif	Stratégie de recherche renseignée (oui/ non)	Critères de sélection des études	Populations et techniques (ou produits) étudiées	Critères d'évaluation	Résultats et signification
Néant						

Etudes cliniques

AMC + diagnosis

Analyse chromosomique sur puce à ADN						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Okubo Y, et al, Brain Dev. 2018 Apr;40(4):334-338.	Génotype	Case report	DD, AMC	CMA	NA	Heterozygous eletion Xq11.2 (ZC4H2)
Mohmaed am, et al, Am J Med Genet Part A 167A:128–136	Génotype	Family report	DD, AMC	CMA	NA	Duplication 16p13.3, deletion 1p36.3
Thevenon et al, Am J Med Genet A. 2014 Dec;164A(12):3027-34.	Génotype	4 unrelated patients	DD, AMC	CMA	NA	Heterozygous deletion 3p14.1p13
Inbar-Feigenberg et al, Ultrasound Obstet Gynecol 2014; 44: 486 – 490	Génotype	2 cases, 1 with deletion	AMC	CMA/FBN2 gene analysis	NA	FBN2 deletion
Carrascosa-Romero et al, Am J Med Genet A. 2013 Sep;161A(9):2281-90	Génotype	Case report	MCA, DD, AMC	CMA	NA	21q22.11
Zanzottera C et al, Am J Med Genet A. 2017 May;173(5):1358-1363.	Génotype	Case report	DD, AMC	CMA	NA	heterozygous deletion Xq11.2, 429kb, encompassing only ZC4H2
Balta A et al, Am J Med Genet A. 2017, Oct;173(10):2798-2802.	Génotype	Case report	MCA, DD, AMC	CMA	NA	concurrent 5p14.1-p15.2 and 5q14.3-q23.2 deletions
Au et al, Am J Med Genet A. 2016 Nov;170(11):2984-2987	Génotype	Case report	MCA, DD, AMC	CMA	NA	recurrent 2.65Mb deletion of 8q13.2q13.3
Jonsson et al, Eur J Med Genet. 2012 Jun;55(6-7):437-40.	Génotype	Case report	Dysmorphic Features, DD, AMC	CMA	NA	1.13Mb deletion of 12q13.13
Tabet et al, Am J Med Genet A. 2010 Jul;152A(7):1781-8.	Génotype	case report	Dysmorphic features, DD, AMC	CMA	NA	6q24.2q25.3 duplication interrupting UTRN
Lukusa et al, Genet Couns. 2010;21(1):25-34.	Génotype	Case report	Dysorphic features, AMC	CMA	NA	17q25.3 duplication
Busche et al, Eur J Med Genet. 2008 Nov-Dec;51(6):615-21.	Génotype	Case report	Dysmorphic features, ID, AMC	CMA	NA	Unbalanced translocation t(2;9)(p25.2;q34.3)
Li Z et al. Gene. 2013 Dec 1;531(2):502-5.	Génotype	Case report	ID, dysmorphic features	CMA	NA	16p13.3 interstitial duplication

Hancorova M et al. Eur J Med Genet. 2013 Mar;56(3):171-3.	Génoty page	Case report	MCA, AMC, Dysmorphic features	CMA	NA	12q13.13 deletion
Chen CP et al. Gene. 2013 Mar 1;516(1):132-7.	Génoty page	Case report	MCA, AMC	CMA	NA	11.31-Mb duplication at 3q27.3–q29 and a 19.56-Mb deletion at 14q31.3–q32.33.
Chen CP et al. Taiwan J Obstet Gynecol. 2011 Dec;50(4):479-84	Génoty page	Case report	AMC, choroid plexus cysts	Chromosome analysis	NA	48,XYY+18
Rameiri V et al.J Craniofac Surg. 2011 Nov;22(6):2124-8.	Génoty page	Case report	Dysmorphic Features	Chromosome annalysis	NA	chromosome 3 interstitial deletion q22.1-q25.2
Hagen A et al. Am J Med Genet A. 2011 Dec;155A(12):3075-81.	Génoty page	Case report	Dysmorphic features, DD, AMC	Array CGH	NA	Deletion of 3.46 Mb at 10p15.3p15.2 and gain of 32.21 Mb at 11q22.2q25. T
Tabet AC Am J Med Genet A. 2010 Jul;152A(7):1781-8	Génoty page	Case report	Dysmorphic features, DD, AMC	Chromosome analysis, Array CGH	NA	6q24.2q25.3 duplication
Liewlick T et al.J Child Neurol. 2011 Aug;26(8):1005-8.	Génoty page	Case report	Dysmorphic features, AMC	Chromosome analysis, microarray CGH	NA	614-Kb duplication of chromosome 22q11.2 (16950312-17564416)
Hiraki Y et al. Am J Med Genet A. 2011 Feb;155A(2):409-14	Génoty page	Case report	Dysmorphic features, DD, AMC	Chromosome analysis, CMA	NA	6.5-Mb deletion at 20q11.21–q12
Kim YJ et al. Korean J Lab Med. 2011 Jan; 31(1): 49–53	Génoty page	Case report	Dysmorphic features, DD, AMC	Chromosome analysis, CGH	NA	arr17p13.3p13.2 (28,969-4,653,156)×1 pa
Thienpont B et al.J Med Genet. 2010 Mar;47(3):155-61	Génoty page	9 patients with 16p13.3 deletion	Dysmorphic features, DD, AMC	CGH	NA	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrom
Nakamura-Pereira M et al Clin Ultrasound. 2009 Oakamura-Pereira Mct;37(8):471-4.	Génoty page	Case Report	MCA	Chromosome analysis	NA	Tetrasomy 9 p
Ansari M et al.European Journal of Medical Genetics 57 (2014) 587e595	Génoty page	6 Cases	Dysmorpic features, MCA, ID, AMC	Array CGH, FISH	NA	5q23 cases
Otake K et al. et al. Pediatr Surg Int (2009) 25: 827	Génoty page	1 case	Congenital diaphragmatic hernia, dysmorphic features, AMC	Chromosome analysis	NA	1q12q23 deletion
Chen CP et al. Taiwan J Obstet Gynecol. 2008 Mar;47(1):93-4	Génoty page	1 case	MCA, AMC	Chromosome analysis	NA	Trisomy 18
Kosaki R et al. Congenit Anom (Kyoto). 2006 Jun;46(2):115-7	Génoty page	1 case	Dysmorphic features	Chromosome analysis	NA	Trisomy 9 mosaicism

Peng HH et al. Prenat Diagn 2005; 25: 470–474	Génotypage	1 case	MCA	Chromosome analysis , Array CGH and chromosome painting	NA	46, XY,der(4), t(4;12) (q35.1; q21.2)
Grati FR et al. Am J Med Genet A. 2005 Jul 30;136(3):254-8	Génotypage	3 cases	Dysmorphic features, AMC, akinesia	Chomosome analysis, STR mapping	NA	Case 1 and 2: two deleted regions (6q22 and 6q25.1-q25.2) and a duplication of 6q23-q25.1. Case 3:deletion of 6q14-q16
Destree et al. Prenat Diagn 2005; 25: 354–357	Génotypage	1 case	Dysmorphic features, AMC	Chromosome analysis, FISH	NA	Trisomy 6 mosaicism
Chen CP et al.	Génotypage	Niveau 4/ case series - 11 cases	MCA, AMC	Chromosome analysis	NA	Preuve C; Trisomy 18
Castro Gago M et al. J Child Neurol. 2005 Jan;20(1):76-8	Génotypage	1 case	AMC, undescended testes, gracile bones, velopharyngeal incompetance, GE reflux	Chromosome analysis, FISH	NA	22q11.2 deletion
Denizot S et. al.J Perinatol. 2004 Nov;24(11):733-4	Génotypage	1 case	Hypotonia, feeding difficulties, AMC	Chromosomes; DNA methylation	NA	Uniparental maternal disomy for Prader Willi syndrome
Devriendt K et al. Eur J Pediatr. 2004 Jun;163(6):329-30	Génotypage	1 case	Hypotonia, congenital heart defect, distal arthrogryposis	Chromosomes, FISH	NA	22q11.2 deletion
Punal E et al. Rev Neurol. 2003 Oct 1-15;37(7):601-	Génotypage	Niveau 4/ 16 cases	Brain malformations, DD, AMC	FISH, DNA amplification with PCR	NA	Preuve C; 22q11.2 deletion
Wu YC et al.Am J Med Genet A. 2003 Mar 15;117A(3):278-81	Génotypage	Case report	Distal arthrogryposis	Chromosomes, FISH	NA	Mosaic tetrasomy 10p
Enya T et al. Am J Med Genet A. 2018 Mar;176(3):707-711. doi: 10.1002/ajmg.a.38606. Epub 2018 Jan 23.	Génotypage	Case reports; 2 sibs and 1 unrelated patient	Hypotonia, MCA, AMC	Chromosomes, Array GCH, WES	NA	MAGEL2 mutation
Hague J et al. Am J Med Genet A. 2016 Jun;170(6):1608-12. doi:	Génotypage	Single case report	Dysmorphic features, AMC	Next gen seq panel	NA	MYH3 mutation

10.1002/ajmg.a.37631. Epub 2016 Mar 2						
Mejlachowicz D, et al; Am J Hum Genet. 2015 Oct 1;97(4):616-20. doi: 10.1016/j.ajhg.2015.08.010. Epub 2015 Sep 10	Génotypage	Two families	Dysmorphic features, AMC, polyhydramnios, club foot	SNP array, WES	NA	MAGEL2 mutation
Alamillo CL et al; Prenat Diagn. 2015 Nov;35(11):1073-8. doi: 10.1002/pd.4648. Epub 2015 Aug 3	Génotypage	Niveau 4/ 7 cases	MCA, AMC	CMA, karyotype; WES	NA	Preuve C; COL1A2, GBE1, OFD1, RAPSN1
Karaman A et al. Genet Couns. 2015;26(1):77-9.	Génotypage	Case report	Dysmorphic, MCA	Karyotype	NA	Trisomy 18
Chen CP Prenat Diagn. 2003 Jan;23(1):85-7	Génotypage	Case report	Decreased fetal movement; polyhydramnios, single UA, AMC	Karyotype	NA	Mosaic isochromosome 20q

Séquençage très haut débit (exome, génome)						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Zho H et al.; EMBO Mol Med. 2019. Feb 18.pii:e9709.doi: 10. 15252/emmm.201809709	Génotypage	Case report of large extended family		WES	NA	MET
Am J Med Genet A. 2018 ;176(2):359-367. doi: 10.1002/ajmg.a.38577. Epub 2017 Dec 23	Génotypage	4 affected proband with 2 sibs from one family and 1 sib each from a second and third unrelated family		WGS	NA	NEB; ACTA1 and BICD2
Chervinsky E et al. Am J Med Genet A.2018;176(4):1001-1005. doi: 10.1002/ajmg.a.38639.	Génotypage	WES of multiple affected family members in consanguineous family		WES	NA	TTN
Montiero FP et al. Eur J Med Genet. 2019. pii: S1769-7212	Génotypage	Two unrelated families with two sibs from one family and a single affected patient from second		WES	NA	ATP1A2
Jobling R et al.J Med Genet.2018 May;55(5):316-321.doi:10:1136/jmedgenet-2017-105222	Génotypage	Three unrelated families with 6 affected patients		WES/WGS	NA	MAGEL2
Abiusi E, et al.Hum Mol Genet. 2017 Oct 15;26(20): 3989-3994, doi:10.1093/hmg/ddx288	Génotypage	Single family and single affected patient		Homozyosity mapping/WES in a consan	NA	UNC50

				guineous family with lethal arthrogryposis		
Tan QK et al.Cold Spring Harb Mol Case Stud. 2017 Nov 21;3(6). pii: a002063. doi: 10.1101/mcs.a002063	Génotypage	Single affected individual and parents		Trio WES	NA	GLE1
Berkenstadt JM et al. J Ultrasound Med. 2018 Jul;37(7):1827-1833. doi: 10.1002/jum.14520. Epub 2018 Jan 13	Génotypage	Two affected fetuses in a single nonconsanguineous family		WES	NA	LMOD3
Casey J et al. Prenat Diagn. 2016;36(11):1020-1026. doi: 10.1002/pd.4925. Epub 2016 Oct 2.	Génotypage	Affected fetus and mother in single family		WES	NA	RYR1
Bayram Y et al. J Clin Invest. 2016 Feb;126(2):762-78. doi: 10.1172/JCI84457. Epub 2016 Jan 11	Génotypage	52 patients with arthrogryposis from 42 families		WES	NA	CENPJ, GBE1, IDS FBN3, MYO9A, PSD3; A second homozygous or compound heterozygous variant identified a novel gene or in another known gene, in addition to the homozygous variant in a known gene :(vacuolar protein sorting 8 [VPS8 (novel)] occurs with POLR3A, MYBPC2 [novel] occurs with GPR126, CHRNG occurs with ERCC2; COL6A3 occurs with BICD2;LIFR occurs with PI4KA; LIFR occurs with MYH14; MYO18B occurs with MYH7B; and RIPK4 occurs with LMNA); 58.8% of families had molecular dx.
Bauche S et al. Am J Hum Genet. 2016;99(3):753-761. doi: 10.1016/j.ajhg.2016.06.033. Epub 2016 Aug 25	Génotypage	Six unrelated families		WES and Sanger	NA	SLC5A7

Enya T et al. Am J Med Genet A. 2018 Mar;176(3):707-711. doi: 10.1002/ajmg.a.38606. Epub 2018 Jan 23.	Génotypage	Two sibs from a single family and one proband from a second family		WES/Target sequence analysis	NA	MAGEL2
Reinstein E et al. Clin Genet. 2018 Jan;93(1):160-163. doi: 10.1111/cge.13018. Epub 2017 Jul 26	Génotypage	Large kindred		WES	NA	ERG1C1
Mroczek M et al. J Appl Genet. 2017; 58(2): 199–203	Génotypage	Single affected patient		WES	NA	TPM2
Knierim E et al. Am J Hum Genet. 2016 Mar 3;98(3):473-489. doi: 10.1016/j.ajhg.2016.01.006. Epub 2016	Génotypage	4 families with multiple affected individuals; 1 of these families is consanguineous		Autozygosity mapping and WES	NA	TRIP4 and ASCC1
Jo HY et al.Clin Genet. 2016 Aug;90(2):177-81. doi: 10.1111/cge.12714. Epub 2016 Mar 4	Génotypage	2 patients with CMT1A and 1 patient with HNPP		Exome CNV, CONIFER, and CEQer, with comparison to aCGH	NA	Deletion and duplication events associated with CMT1A/HNPP
Wambach JA et al. Am J Hum Genet. 2016 Nov 3;99(5):1206-1216. doi: 10.1016/j.ajhg.2016.09.019. Epub 2016 Oct 27	Génotypage	10 individuals from 4 independent consanguineous families		WES, SNPchip-based linkage analysis, DNA microarray, and Sanger sequencing	NA	PIEZO2
Ekhilevitch N et al. Clin Genet. 2016 Jul;90(1):84-9. doi: 10.1111/cge.12707. Epub 2016 Jan 20	Génotypage	Multiple affected members of consanguineous family		WES	NA	MYBPC1
Wambach JA et al. Hum Mutat. 2017;38(11):1477-1484. doi: 10.1002/humu.23297. Epub 2017 Aug 17	Génotypage	6 infants children from 4 unrelated families		WES	NA	GLDN
Todd EJ et al. Orphanet J Rare Dis. 2015 Nov 17;10:148. doi: 10.1186/s13023-015-0364-0	Génotypage	45 patients with fetal akinesia/hypokinesia, arthrogryposis or severe congenital myopathies belonging to 38 unrelated families		WES	NA	18/38 families had conclusive diagnosis; mutations identified in 8 known genes: CHRND, CHNRG, ECEL1, GBE1, MTM1, MYH3, NEB and RYR1; 4

						novel genes: GPR126, KLHL40, KLHL41 and SPEG
Wang B et al. Am J Med Genet A. 2016 ;170A(1):135-41. doi: 10.1002/ajmg.a.37391. Epub 2015 Sep 16	Génotypage	3 affected members of a single family		WES	NA	TNN12
Mejlachowicz D et al. Am J Hum Genet. 2015;97(4):616-20. doi: 10.1016/j.ajhg.2015.08.010. Epub 2015 Sep 1	Génotypage	2 families; 3 affected/deceased siblings of first family; 1 affected proband of second family		Linkage analysis and WES	NA	MAGEL2
Ravenscroft G et al. Am J Hum Genet. 2015;96(6):955-61. doi:10.1016/j.ajhg.2015.04.014. Epub 2015 May 21	Génotypage	3 consanguineous families affected by lethal AMC		WES, targeted exome sequence	NA	GPR126
Okubo M et al.Am J Med Genet A. 2015;167A(5):1100-6. doi: 10.1002/ajmg.a.36881. Epub 2015 Feb 25	Génotypage	Multiple affected individuals in a single family		WES	NA	PIEZO2
Dohrn N et al. Am J Med Genet A. 2015;167A(4):731-43. doi: 10.1002/ajmg.a.37018. Epub 2015 Feb 23	Génotypage	2 affected fetuses in a consanguineous family		WES	NA	ECEL1
Wilbe M et al. J Med Genet. 2015 Mar;52(3):195-202. doi: 10.1136/jmedgenet-2014-102730. Epub 2015 Jan 2	Génotypage	Affected fetus in one family with 5 affected fetuses		WES	NA	MuSK
Seo J et al. J Hum Genet. 2015;60(4):213-5. doi: 10.1038/jhg.2015.2. Epub 2015 Jan 22	Génotypage	Three affected patients from two families		WES	NA	CHRNG
Patel N et al. Hum Mol Genet. 2014;23(24):6584-93. doi: 10.1093/hmg/ddu384. Epub 2014 Jul 23el	Génotypage	Stillborn infant in consanguineous family with male and female affected stillbirths		Autozygome and WES	NA	ZBTB42
Shaaban S et al. Clin Genet. 2014;85(6):562-7. doi: 10.1111/cge.12224. Epub 2013 Jul 19	Génotypage	Consanguineous pedigree with two affected sibs		Homozygosity mapping and WES	NA	ECEL1
Davidson AE et al. Brain. 2013;136(Pt 2):508-21. doi: 10.1093/brain/aws344	Génotypage	WES and targeted sequence analysis of affected individuals in 4 families		WES	NA	TPM2
Ravenscroft G et al.Neuromuscul Disord. 2013;23(2):165-9. doi: 10.1016/j.nmd.2012.11.005. Epub 2012 Dec 3	Génotypage	Two siblings in non consanguineous family		WES	NA	GBE1

Markus B et al. Hum Mutat. 2012 Oct;33(10):1435-8. doi: 10.1002/humu.22122. Epub 2012 Jun 7	Génotypage	WES in two individuals of different consanguineous tribes		WES	NA	LCCS4
McMillin MJ et al.Am J Hum Genet. 2013 Jan 10;92(1):150-6. doi: 10.1016/j.ajhg.2012.11.014. Epub 2012 Dec 20	Génotypage	WGS analysis of 15 affected individuals in 5 families (1 reported to be consanguineous)		WGS	NA	ECEL1

Maladie de Steinert/ DMPK						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Kahn JA, Cataltepe S. A 35-week neonate with respiratory failure, hypotonia, and joint contractures. Curr Opin Pediatr. 1996 Dec;8(6):583-8.	Phénotypage	case report	NA	NA	NA	Bilateral flexion contractures on elbows and hips
Martinello F, Piazza A, Pastorello E, Angelini C, Trevisan CP. Clinical and neuroimaging study of central nervous system in congenital myotonic dystrophy. J Neurol. 1999 Mar;246(3):186-92.	Phénotypage	série de cas	NA	NA	NA	Bilateral flexion contractures of elbows and of the left knee (n=1)
González de Dios J, Martínez Frías ML, Egües Jimeno J, Gairi Tahull JM, Gómez Sabrido F, Morales Fernández MC, Paisán Grisolía L, Pardo Romero M, Medina Rams M. [Epidemiological study of Steinert's congenital myotonic dystrophy: dysmorphological characteristics]. An Esp Pediatr. 1999 Oct;51(4):389-96.	Phénotypage	série de cas	NA	NA	NA	arthrogryposis (n=1)
Akiyama M, Yuza Y, Yokokawa Y, Yokoi K, Ariga M, Eto Y. Differences in CTG triplet repeat expansion in leukemic cells and normal lymphocytes from a 14-year-old female with congenital myotonic dystrophy. Pediatr Blood Cancer. 2008 Oct;51(4):563-5.	Phénotypage	case report	NA	NA	NA	extensive contractures of multiple joints of the lower extremities
Schilling L, Forst R, Forst J, Fujak A. Orthopaedic Disorders in Myotonic Dystrophy Type 1: descriptive clinical study of 21 patients. BMC Musculoskelet Disord. 2013 Dec 1;14:338. doi: 10.1186/1471-2474-14-338.	Phénotypage	Niveau 4/ série de cas	NA	NA	NA	5 patients suffered from contractures at at least one other joint level than feet

Lowry RB, Sibbald B, Bedard T, Hall JG. Birth Defects Res A Clin Mol Teratol. 2010 Dec;88(12):1057-61. doi: 10.1002/bdra.20738.	Phénotypage	103 patient cohort of MCC patients	NA	NA	NA	1 patient of group III (lethal or central forms) had a diagnosis of MD

Prader-Willi syndrome						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Denizot S, Boscher C, Le Vaillant C, Rozé JC, Gras Le Guen C. J Perinatol. 2004 Nov;24(11):733-4.	Génotypage et phénotypage	Case report		Maternal UPD 15q11	NA	Peripheral and axial hypotonia with bilateral articular stiffness of hands and feet
Bigi N, Faure JM, Coubes C, Puechberty J, Lefort G, Sarda P, Blanchet P. Prenat Diagn. 2008 Sep;28(9):796-9.	Génotypage et phénotypage	2 case reports		Paternal 15q11-q13 deletion; maternal UPD15	NA	Oddly positioned hands and feet with extended legs and feet and flexed toes prenatally, adducted thumbs over the index and middle fingers and flexed fingers, feet in varus position with flexed toes disappearing after a few days (1); Extended feet and flexed toes, permanently clenched hands prenatally (2)

Amyotrophie spinale/ SMN1						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Bingham PM, Shen N, Rennert H, Rorke LB, Black AW, Marin-Padilla MM, Nordgren RE. Arthrogryposis due to infantile neuronal degeneration associated with deletion of the SMNT gene. Neurology. 1997 Sep;49(3):848-51.	Phénotypage	4 case reports	NA	NA	NA	Case 1: moderate flexion contractures of fingers, elbows and knees at birth; case 2: elbows, wrists, hips, knees, ankles, vertebral column; case 3: severe flexion contractures at all joints in both arms and legs; case 4: multiples contractures at birth
Bürglen L, Amiel J, Viollet L, Lefebvre S, Burlet P, Clermont O, Raclin V, Landrieu P, Verloes A, Munnich A, Melki J. Survival motor neuron gene deletion in the arthrogryposis multiplex congenita-spinal muscular atrophy association. J Clin Invest. 1996 Sep 1;98(5):1130-2.	Phénotypage	Niveau 4/ cohort	NA	NA	NA	12 cases with multiples congenitales contractures in upper and lower limbs and hips

Falsaperla R, Romeo G, Di Giorgio A, Pavone P, Parano E, Connolly AM. Long-term survival in a child with arthrogryposis multiplex congenita and spinal muscular atrophy. J Child Neurol. 2001 Dec;16(12):934-6.	Phénotypage	case report	NA	NA	NA	Contractures of fingers, wrists, knees, toes, ankles; scoliosis
Nadeau A, D'Anjou G, Debray G, Robitaille Y, Simard LR, Vanasse M. A newborn with spinal muscular atrophy type 0 presenting with a clinicopathological picture suggestive of myotubular myopathy. J Child Neurol. 2007 Nov;22(11):1301-4.	Phénotypage	case report	NA	NA	NA	arthrogryposis noted on elbows, fingers, hips, knees
Grotto S, Cuisset JM, Marret S, Drunat S, Faure P, Audebert-Bellanger S, Desguerre I, Flurin V, Grebille AG, Guerrot AM, Journel H, Morin G, Plessis G, Renolleau S, Roume J, Simon-Bouy B, Touraine R, Willems M, Frébourg T, Verspyck E, Saugier-Veber P. Type 0 Spinal Muscular Atrophy: Further Delineation of Prenatal and Postnatal Features in 16 Patients. J Neuromuscul Dis. 2016 Nov 29;3(4):487-495.	Phénotypage	Niveau 4; 16 patient cohort	NA	NA	NA	Preuve C; 15 of 16 patients with type 0 SMA had joint contractures
MacLeod MJ, Taylor JE, Lunt PW, Mathew CG, Robb SA. Prenatal onset spinal muscular atrophy. Eur J Paediatr Neurol. 1999;3(2):65-72.	Phénotypage	5 patient report	NA	NA	NA	Fixed flexion contractures of fingers, elbows and knees (case 3), restricted movements at elbows, hips, shoulders

Electroneuromyographie						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, Pantaleoni C. ZC4H2 deletions can cause severe phenotype in female carriers. Am J Med Genet A. 2017 May;173(5):1358-1363.	Phénotypage	Case report	ZC4H2	NA	NA	normal at 6 years; neurogenic in deltoid and moderate myopathic anomalies at distal muscles of lower limbs
Zafeiriou DI, Pitt M, de Sousa C. Clinical and neurophysiological characteristics of congenital myasthenic syndromes presenting in early infancy. Brain Dev. 2004 Jan;26(1):47-52.	Phénotypage	11 patients cohort study	CMS with episodic apnoe	NA	NA	decrement at low frequency repetitive stimulation, increment high frequency RS

Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Novel BICD2 mutation in a Japanese family with autosomal dominant lower extremity-predominant spinal muscular atrophy-2. Brain Dev. 2018 Apr;40(4):343-347.	Phénotypage	family report SMALED2	BICD2	NA	NA	Chronic neurogenic change in one index patient
Yıldırım Y, Orhan EK, Iseri SA, Serdaroglu-Oflazer P, Kara B, Solakoğlu S, Tolun A. A frameshift mutation of ERLIN2 in recessive intellectual disability, motor dysfunction and multiple joint contractures. Hum Mol Genet. 2011 May 15;20(10):1886-92.	Phénotypage	family report	ERLIN2	NA	NA	normal EMG and NCV in the 4 patients investigated
Verma S, Goyal P, Guglani L, Peinhardt C, Pelzek D, Barkhaus PE. COL6A and LAMA2 Mutation Congenital Muscular Dystrophy: A Clinical and Electrophysiological Study. J Clin Neuromuscul Dis. 2018 Mar;19(3):108-116.	Phénotypage	8 patients COL6A, 6 patients LAMA2 cohort EMG study	COL6A, LAMA2	NA	NA	proximal myopathic and distal neurogenic features in all patients
Kang PB, Lidov HG, David WS, Torres A, Anthony DC, Jones HR, Darras BT. Diagnostic value of electromyography and muscle biopsy in arthrogryposis multiplex congenita. Ann Neurol. 2003 Dec;54(6):790-5.	Phénotypage	38 patient cohort; diagnostic comparison between NCV/ EMG and muscle biopsy	various	NA	NA	low sensitivities and PPV;
Reddel S, Ouvrier RA, Nicholson G, Dierick I, Irobi J, Timmerman V, Ryan MM. Autosomal dominant congenital spinal muscular atrophy--a possible developmental deficiency of motor neurones? Neuromuscul Disord. 2008 Jul;18(7):530-5.	Phénotypage	family report	SMALED	NA	NA	Increased motor unit action potential amplitude
Rabie M, Jossiphov J, Nevo Y. Electromyography (EMG) accuracy compared to muscle biopsy in childhood. J Child Neurol. 2007 Jul;22(7):803-8.	Phénotypage	Cohort study on diagnostic accuracy of EMG	Neurogenic	NA	NA	100% detection rate in neurogenic forms of AMC
Ko JM, Choi IH, Baek GH, Kim KW. First Korean family with a mutation in TPM2 associated with Sheldon-Hall syndrome. J Korean Med Sci. 2013 May;28(5):780-3.	Phénotypage	family report	TPM2/ Sheldon-Hall	NA	NA	Myopathic EMG pattern

Kiselev A, Vaz R, Knyazeva A, Khudiakov A, Tarnovskaya S, Liu J, Sergushichev A, Kazakov S, Frishman D, Smolina N, Pervunina T, Jorholt J, Sjoberg G, Vershinina T, Rudenko D, Arner A, Sejersen T, Lindstrand A, Kostareva A. De novo mutations in FLNC leading to early-onset restrictive cardiomyopathy and congenital myopathy. Hum Mutat. 2018 Sep;39(9):1161-1172.	Phénotypage	2 family report	FLNC	NA	NA	Diffuse myopathic pattern
Gaitanis JN, McMillan HJ, Wu A, Darras BT. Electrophysiologic evidence for anterior horn cell disease in amyoplasia. Pediatr Neurol. 2010 Aug;43(2):142-7.	Phénotypage	5 patients cohort	Amyoplasia	NA	NA	4 patients have a neurogenic pattern with apparent predilection for the upper cervical myotomes (C5-C6)
Fusco C, Frattini D, Salerno GG, Canali E, Bernasconi P, Maggi L. New phenotype and neonatal onset of sodium channel myotonia in a child with a novel mutation of SCN4A gene. Brain Dev. 2015 Oct;37(9):891-3.	Phénotypage	Case report	SCN4A myotonia	NA	NA	Myotonic discharges and myopathic changes
Fleming J, Quan D. A case of congenital spinal muscular atrophy with pain due to a mutation in TRPV4. Neuromuscul Disord. 2016 Dec;26(12):841-843.	Phénotypage	Family Report	TRPV4	NA	NA	Normal NCV; long duration motor unit potentials and reduced recruitment on EMG
Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316-9.	Phénotypage	cohort report	various	NA	NA	definition of cohort according to EMG findings when neurogenic

Créatine Phosphokinase CPK						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Kölbel H, Abicht A, Schwartz O, Katona I, Paulus W, Neuen-Jacob E, Weis J, Schara U. Characteristic clinical and ultrastructural findings in nesprinopathies. Eur J Paediatr Neurol. 2019 Mar;23(2):254-261.	Phénotypage	3 family report	nesprinopathy	2 of 5 patients with myopathic AMC; CK normal	NA	N

Astrea G, Romano A, Angelini C, Antozzi CG, Barresi R, Battini R, Battisti C, Bertini E, Bruno C, Cassandrini D, Fanin M, Fattori F, Fiorillo C, Guerrini R, Maggi L, Mercuri E, Morani F, Mora M, Moro F, Pezzini I, Picillo E, Pinelli M,Politano L, Rubegni A, Sanseverino W, Savarese M, Striano P, Torella A, Trevisan CP, Trovato R, Zaraieva I, Muntoni F, Nigro V, D'Amico A, Santorelli FM; Italian CMD Network. Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet J Rare Dis. 2018 Sep 26;13(1):170.	Phénotypage	13 patient report	GMPPB	1 patient of 13 with AMC, PMG, micrcephaly, cataracts, sudden heart block; high CK level	NA	high
Conant A, Curiel J, Pizzino A, Sabetrasekh P, Murphy J, Bloom M, Evans SH, Helman G, Taft RJ, Simons C, Whitehead MT, Moore SA, Vanderver A. Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogryposis. J Child Neurol. 2018 Sep;33(10):642-650.	Phénotypage	2 patient report	CNTNAP1	2 patients with hypomyelinating neuropathy and central hypomyelination with early respiratory failure	NA	N
Sivaraman I, Friedman NR, Prayson RA. Muscle biopsy findings in a child with NALCN gene mutation. J Clin Neurosci. 2016 Dec;34:222-223.	Phénotypage	Case report	NALCN	1 patient with global developmental delay, generelized hypotonia, ulnar deviation of hands and bilateral clawing of toes; CK normal	NA	N
Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Novel BICD2 mutation in a Japanese family with autosomal dominant lower extremity-predominant spinal muscular atrophy-2. Brain Dev. 2018 Apr;40(4):343-347.	Phénotypage	Family report	BICD2	2 patients with SMALED2, father and son; son with resolving AMC; CK at 30y was 1478UI/l (N<100), but normal at 20y, and not measured in his son.	NA	high in one patient on one occasion
Janecke AR, Li B, Boehm M, Krabichler B, Rohrbach M, Müller T, Fuchs I, Golas G, Katagiri Y, Ziegler SG, Gahl WA, Wilnai Y, Zoppi N, Geller HM, Giunta C, Slavotinek A, Steinmann B. The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to CHST14 mutations. Am J Med Genet A. 2016 Jan;170A(1):103-15.	Phénotypage	Cohort report	CHST14	7 patients with musculo-skeletal EDS of whom 3/5 had elvated CK levels varying from 300 to 3000IU/L	NA	high in 3/5 patients on several occasions

Hunter JM, Ahearn ME, Balak CD, Liang WS, Kurdoglu A, Corneveaux JJ, Russell M, Huentelman MJ, Craig DW, Carpten J, Coons SW, DeMello DE, Hall JG, Bernes SM, Baumbach-Reardon L. Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Mol Genet Genomic Med. 2015 Jul;3(4):283-301.	Phénotypage	Cohort report	COL6A6	1 patient with congenital club feet, COL6A3 AR mutations, CK 600-1000IU/L; 1 patient with AMC and COL6A6 mutations but no CK measure	NA	no CK level in AMC patient
Echaniz-Laguna A, Dubourg O, Carlier P, Carlier RY, Sabouraud P, Péréon Y, Chapon F, Thauvin-Robinet C, Laforêt P, Eymard B, Latour P, Stojkovic T. Phenotypic spectrum and incidence of TRPV4 mutations in patients with inherited axonal neuropathy. Neurology. 2014 May 27;82(21):1919-26.	Phénotypage	Cohort report	TRPV4	2 patients out of 17 have had congenital SMA with arthrogryposis	NA	high in both, 420IU and 300IU respectively (n<200)
Dlamini N, Josifova DJ, Paine SM, Wraige E, Pitt M, Murphy AJ, King A, Buk S, Smith F, Abbs S, Sewry C, Jacques TS, Jungbluth H. Clinical and neuropathological features of X-linked spinal muscular atrophy (SMAX2) associated with a novel mutation in the UBA1 gene. Neuromuscul Disord. 2013 May;23(5):391-8.	Phénotypage		UBA1	SMAX2 patient report with AMC	NA	High CK levels at 3 days of age 4348kIU/l (n<5,8kIU/l), returning to normal over 4 weeks)
Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316-9.	Phénotypage	Cohort report	diverse	AMC cohort review	NA	Only slighltly raised in 5/12 neurogenic forms of AMC (max. 347IU/l) and normal in all other 11 patients.
Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. A mutation in the fast skeletal muscle troponin I gene causes myopathy and distal arthrogryposis. Neurology. 2006 Aug 22;67(4):597-601.	Phénotypage	Family report	TNNI2	Family report on 5 affected patients over 3 generations with DA2B	NA	Mildly elevated CK levels in 4 adults
Chauveau C, Bonnemann CG, Julien C, Kho AL, Marks H, Talim B, Maury P, Arne-Bes MC, Uro-Coste E, Alexandrovich A, Vihola A, Schafer S, Kaufmann B, Medne L, Hübner N, Foley AR, Santi M, Udd B, Topaloglu H, Moore SA, Gotthardt M, Samuels ME, Gautel M, Ferreiro A. Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. Hum Mol Genet. 2014 Feb 15;23(4):980-91.	Phénotypage	Cohort report	TTN	5 patients with distal arthrogryposis, core myopathy and heart disease	NA	Mildly elevated CK levels in 4 patients (<5N)
Philpot J, Counsell S, Bydder G, Sewry CA, Dubowitz V, Muntoni F. Neonatal arthrogryposis and absent limb muscles: a muscle developmental gene defect? Neuromuscul Disord. 2001 Jul;11(5):489-93.	Phénotypage	Case report	Neurogenic Amyopalsia-like AMC	Case report	NA	N

Vasli N, Harris E, Karamchandani J, Bareke E, Majewski J, Romero NB, Stojkovic T, Barresi R, Tasfaout H, Charlton R, Malfatti E, Bohm J, Marini-Bettolo C, Choquet K, Dicaire MJ, Shao YH, Topf A, O'Ferrall E, Eymard B, Straub V, Blanco G, Lochmüller H, Brais B, Laporte J, Tétreault M. Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain. 2017 Jan;140(1):37-48.	Phénotypage	5 families report	ZAK	in 1 family : 2 siblings with hypotonia and contractures at birth, slowly progrssiive motor weakness, motor delay, hyperlaxity, cervical spine hypermotility and contractures (ankle and fingers), scoliosis, no CNS involvement. Contractures not observed in the 4 other families.	NA	N

Biopsie musculaire - Arthrogryposes distales						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Sandaradura SA, Bournazos A, Mallawaarachchi A, Cummings BB, Waddell LB, Jones KJ, Troedson C, Sudarsanam A, Nash BM, Peters GB, Algar EM, MacArthur DG, North KN, Brammah S, Charlton A, Laing NG, Wilson MJ, Davis MR, Cooper ST. Nemaline myopathy and distal arthrogryposis associated with an autosomal recessive TNNT3 splice variant. Hum Mutat. 2018 Mar;39(3):383-388.	Phénotypage	case report	TNNT3 homozygous splice mutation	NA	NA	Bilateral vertical talus, knee and finger contractures, bilateral hip dislocation; selective and marked atrophy type 2 fibres, nemalin rods in type 2 fibres
Haliloglu G, Becker K, Temucin C, Talim B, Küçükşahin N, Pergande M, Motameny S, Nürnberg P, Aydingoz U, Topaloglu H, Cirak S. Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects. J Hum Genet. 2017 Apr;62(4):497-501.	Phénotypage	case report	homozygous nonsense PIEZO2 mutation	NA	NA	variation in fibre size, mild increase in endomyseal tissue and focal fatty infiltration, rounded myofibres, internal nuclei; predominant type 1 fibres
Sivaraman I, Friedman NR, Prayson RA. Muscle biopsy findings in a child with NALCN gene mutation. J Clin Neurosci. 2016 Dec;34:222-223.	Phénotypage	case report	NALCN	NA	NA	Ulnar deviation of hands and bilateral clawing of toes; small focus of chronic endomysial inflammation and a mild variation in muscle fiber type, type 2 fiber type atrophy

Thevenon J, Monnier N, Callier P, Dieterich K, Francoise M, Montgomery T, Kjaergaard S, Neas K, Dixon J, Dahm TL, Huet F, Ragon C, Mosca-Boidron AL, Marle N, Duplomb L, Aubriot-Lorton MH, Mugneret F, Vokes SA, Tucker HW, Lunardi J, Faivre L, Jouk PS, Thauvin-Robinet C. Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. Am J Med Genet A. 2014 Dec;164A(12):3027-34.	Phénotypage	4 patients	deletion 3p14.1 p13	NA	NA	Patient 1: normal muscle biopsy
Davidson AE, Siddiqui FM, Lopez MA, Lunt P, Carlson HA, Moore BE, Love S, Born DE, Roper H, Majumdar A, Jayadev S, Underhill HR, Smith CO, von der Hagen M, Hubner A, Jardine P, Merrison A, Curtis E, Cullup T, Jungbluth H, Cox MO, Winder TL, Abdel Salam H, Li JZ, Moore SA, Dowling JJ. Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. Brain. 2013 Feb;136(Pt 2):508-21.	Phénotypage	3 family report	TPM2 delK7	NA	NA	Distal and proximal AMC in 2 families, with limited jaw opening; wide variation of fiber diameter, rod-like inclusions, myofibrillar disarray, minicore-like areas
Dieterich K, Quijano-Roy S, Monnier N, Zhou J, Fauré J, Smirnow DA, Carlier R, Laroche C, Marcorelles P, Mercier S, Mégarbané A, Odent S, Romero N, Sternberg D, Marty I, Estournet B, Jouk PS, Melki J, Lunardi J. The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Hum Mol Genet. 2013 Apr 15;22(8):1483-92.	Phénotypage	6 family report	ECEL1	NA	NA	Type 1 fiber predominance, mild fiber size variability, no major structural abnormalities of the sarcolemma nor signs of denervation
Tajsharghi H, Kimber E, Holmgren D, Tulinius M, Oldfors A. Distal arthrogryposis and muscle weakness associated with a beta-tropomyosin mutation. Neurology. 2007 Mar 6;68(10):772-5.	Phénotypage	2 case report	TPM2 R133W	NA	NA	type 1 fiber predominance, normal ultrastructure
Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. Distal arthrogryposis: clinical and genetic findings. Acta Paediatr. 2012 Aug;101(8):877-87.	Phénotypage	DA cohort	MYH3 DA2B, MYH3 DA2A, TNNI2 DA1 ou DA2B	NA	NA	type 1 fiber predominance, increased variability in fiber size; type 2 fiber changes
Vydyanath A, Gurnett CA, Marston S, Luther PK. Axial distribution of myosin binding protein-C is unaffected by mutations in human cardiac and skeletal muscle. J Muscle Res Cell Motil. 2012 May;33(1):61-74.	Phénotypage	2 cases	MYBPC1	NA	NA	DA1; normal histology and ultrastructure
Oldfors A, Lamont PJ. Thick filament diseases. Adv Exp Med Biol. 2008;642:78-91.	Phénotypage	1 family report	MYH2	NA	NA	DA10; dystrophic pattern, rimmed vacuoles, tubulofilamentous inclusions

Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. A mutation in the fast skeletal muscle troponin I gene causes myopathy and distal arthrogryposis. Neurology. 2006 Aug 22;67(4):597-601.	Phénotypage	1 family report	TNNI2 p.K175del	NA	NA	type 1 fiber predominance, internalized nuclei, scattered regenerating fibers, mild increase in endomysial connective tissue
Tajsharghi H, Kimber E, Kroksmark AK, Jerre R, Tulinius M, Oldfors A. Embryonic myosin heavy-chain mutations cause distal arthrogryposis and developmental myosin myopathy that persists postnatally. Arch Neurol. 2008 Aug;65(8):1083-90.	Phénotypage	3 family report	MYH3	NA	NA	Increased variability of fiber size due to scattered, small type 1 fibers, mild type 1 fiber predominance
Hageman G, Jennekens FG, Vette JK, Willemse J. The heterogeneity of distal arthrogryposis. Brain Dev. 1984;6(3):273-83.	Phénotypage	1 case report	no gene identified	NA	NA	DA1; increase in internal nuclei (10%), occasionnal small clusters of one fiber type, small atrophic fibers predominantly type 1
Monnier N, Lunardi J, Marty I, Mezin P, Labarre-Vila A, Dieterich K, Jouk PS. Absence of beta-tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. Neuromuscul Disord. 2009 Feb;19(2):118-23. doi: 10.1016/j.nmd.2008.11.009. Epub 2009 Jan 19. PMID: 19155175.	Phénotypage	1 family report	TPM2 nonsense mutation			Escobar syndrome; slight type 1 fiber predominance, subsarcolemmal and internal aggregates of rods, fibro-adipose tissue replacement
Sung SS, Brassington AM, Krakowiak PA, Carey JC, Jorde LB, Bamshad M. Mutations in TNNT3 cause multiple congenital contractures: a second locus for distal arthrogryposis type 2B. Am J Hum Genet. 2003 Jul;73(1):212-4. doi: 10.1086/376418. PMID: 12865991; PMCID: PMC1180583.		1 family report	TPM2 R91G			no rods

Biopsie musculaire - Amyoplasie						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Gaitanis JN, McMillan HJ, Wu A, Darras BT. Pediatr Neurol. 2010 Aug;43(2):142-7.	Phénotypage	cohort report	NA	NA	NA	5 patients with Amyoplasia; patient 1 A: left quadriceps revealed nonspecific changes: mild variability in fiber size, no atrophy, hypertrophy, fiber splitting, fiber-type atrophy, or grouping. Electron microscopy normal.
Price DS. Arch Dis Child. 1933 Oct;8(47):343-54	Phénotypage	case report	NA	NA	NA	Striated muscle fibres, areas of fibrous tissue, and fat replacement. There were also some infrequent zones of small atrophied fibres and chains of nuclei with degenerated sheaths.

Biopsie musculaire - arthrogryposes multiples congénitales						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Carrera-García L, Natera-de Benito D, Dieterich K, de la Banda MGG, Felter A, Inarejos E, Codina A, Jou C, Roldan M, Palau F, Hoenicka J, Pijuan J, Ortez C, Expósito-Escudero J, Durand C, Nugues F, Jimenez-Mallebrera C, Colomer J, Carlier RY, Lochmüller H, Quijano-Roy S, Nascimento A. Am J Med Genet A. 2019 Jun;179(6):915-926.	Phénotypage	cohort report; only 1 with muscle biopsy results	CHRNG	NA	NA	Non lethal multiple pterygium syndrom; histological report on muscle pathology in case 3: non specific mild myopathic changes, occasional neonatal myosin heavy chain staining, small sized neuromuscular junctions and abnormal shape
Alkhunaizi E, Shuster S, Shannon P, Siu VM, Darilek S, Mohila CA, Boissel S, Ellezam B, Fallet-Bianco C, Laberge AM, Zandberg J, Injeyan M, Hazrati LN, Hamdan F, Chitayat D. Am J Med Genet A. 2019 Mar;179(3):386-396.	Phénotypage	cohort report	RYR1	NA	NA	FADS, lethal MPS, hydrops; pahology reports in 4 families of 5: small myocytes and fasciles, reduced number or focal disarray of Z-bands, excess of connective tissue and lipid inclusions; muscle histology did NOT assist the diagnosis

Bonnin E, Cabochette P, Filosa A, Jühlen R, Komatsuzaki S, Hezwani M, Dickmanns A, Martinelli V, Vermeersch M, Supply L, Martins N, Pirenne L, Ravenscroft G, Lombard M, Port S, Spillner C, Janssens S, Roets E, Van Dorpe J, Lammens M, Kehlenbach RH, Ficner R, Laing NG, Hoffmann K, Vanhollebeke B, Fahrenkrog B. PLoS Genet. 2018 Dec 13;14(12):e1007845.	Phénotypage	2 family report	NUB88	NA	NA	FADS, 2 family reports; no detailed histological analysis; weaker staining of Rapsyn antibody compared to controls
Berkenstadt M, Pode-Shakked B, Barel O, Barash H, Achiron R, Gilboa Y, Kidron D, Raas-Rothschild A. J Ultrasound Med. 2018 Jul;37(7):1827-1833.	Phénotypage	Family report	LMOD3	NA	NA	FADS; family report; muscle atrophy, hip, knees, ankles contracted; muscle fiber loss and fibrosis, variability of fiber size, nemalin bodies
Ahmed AA, Skaria P, Safina NP, Thiffault I, Kats A, Taboada E, Habeebu S, Saunders C. Am J Med Genet A. 2018 Feb;176(2):359-367.	Phénotypage	4 case reports	NEB, ACTA1, BICD2	NA	NA	Lethal multiple pterygia syndrome and FADS; amyoplasia only mentionned as histological finding in NEB mutations as absence of muscle tissue; nemalin bodies + variation in fiber size in ACTA mutations; BICD2 : scattered atrophic and hypoplastic muscle fibers separated by lobules of fibrofatty tissue
Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Brain Dev. 2018 Apr;40(4):343-347.	Phénotypage	Case report	BICD2	NA	NA	features of denervation: occasional scattered angular fibres
Sandaradura SA, Bournazos A, Mallawaarachchi A, Cummings BB, Waddell LB, Jones KJ, Troedson C, Sudarsanam A, Nash BM, Peters GB, Algar EM, MacArthur DG, North KN, Brammah S, Charlton A, Laing NG, Wilson MJ, Davis MR, Cooper ST. Hum Mutat. 2018 Mar;39(3):383-388.	Phénotypage	Case report	TNNT3	NA	NA	Severe weakness, hypotonia, contractures, congenital scoliosis; muscle biopsy: marked atrophy and degeneration of fast fibers with striking nemalin rods, and hypertrophy of slow fibers that were histologically normal

Tan QK, McConkie-Rosell A, Juusola J, Gustafson KE, Pizoli CE, Buckley AF, Jiang YH. Cold Spring Harb Mol Case Stud. 2017 Nov 21;3(6). pii: a002063. doi: 10.1101/mcs.a002063.	Phénotypage	Case report	GLE1	NA	NA	Muscle weakness, hypotonia, distal congenital contractures; Muscle biopsy showed signs of a chronic active neuromuscular insult consistant with anterior horn cell disease: moderate variation in fiber diameter, clusters of atrophic fibers and frequent fiber hypertrophy, uneven staining of COX, mild non specific mitochondrial changes (balling, vacuolation, thickening of cristae with increased matrix material)
Quélin C, Loget P, Rozel C, D'Hervé D, Fradin M, Demurger F, Odent S, Pasquier L, Cavé H, Marcorelles P. Eur J Med Genet. 2017 Jul;60(7):395-398.	Phénotypage	Case report	HRAS p.Gly12Ser	NA	NA	Clenched hands with overlapping fingers, camptodactyly of third fingers, bilateral talips equinovarus; Muscle biopsy: excess of connective tissue, unusual lobulated pattern of the muscle, excess of muscle spindles
Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, Pantaleoni C. Am J Med Genet A. 2017 May;173(5):1358-1363.	Phénotypage	Case report	heterozygous ZC4H2 deletion	NA	NA	non specific hypotrophy of type II fibers
Chimelli L, Melo ASO, Avvad-Portari E, Wiley CA, Camacho AHS, Lopes VS, Machado HN, Andrade CV, Dock DCA, Moreira ME, Tovar-Moll F, Oliveira-Szejnfeld PS, Carvalho ACG, Ugarte ON, Batista AGM, Amorim MMR, Melo FO, Ferreira TA, Marinho JRL, Azevedo GS, Leal JIBF, da Costa RFM, Rehen S, Arruda MB, Brindeiro RM, Delvechio R, Aguiar RS, Tanuri A. Acta Neuropathol. 2017 Jun;133(6):983-999.	Phénotypage	Cohort report	ZIKV	NA	NA	neurogenic muscle atrophy
Fernández-Marmiesse A, Carrascosa-Romero MC, Alfaro Ponce B, Nascimento A, Ortez C, Romero N, Palacios L, Jimenez-Mallebrera C, Jou C, Gouveia S, Couce ML. Neuromuscul Disord. 2017 Feb;27(2):188-192.	Phénotypage	Case report	TTN	NA	NA	Weak suction, severe axial hypotonia, distal contractures, hip dislocation; cytoplasmic bodies, connective tissue, variable fiber size, internal myonuclei
Haliloglu G, Becker K, Temucin C, Talim B, Küçükşahin N, Pergande M, Motameny S, Nürnberg P, Aydingoz U, Topaloglu H, Cirak S. J Hum Genet. 2017 Apr;62(4):497-501.	Phénotypage	Case report	PIEZO2 LoF	NA	NA	Variable fiber size, round myofibers, increased connective tissue, type 1 fiber predominance

Baumann M, Steichen-Gersdorf E, Krabichler B, Petersen BS, Weber U, Schmidt WM, Zschocke J, Müller T, Bittner RE, Janecke AR. Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotype-phenotype correlation. Eur J Hum Genet. 2017 Feb;25(2):262-266.	Phénotypage	Case report	SYNE1 nonsense Ho	NA	NA	Variation in muscle fiber size, no internal myonuclei, honeycombing of myonuclei
Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. Neuromuscul Disord. 2016 Oct;26(10):681-687.	Phénotypage	Case report	GBE1	NA	NA	Rimmed and on rimmed vacuoles, reduction in glycogen content on PAS, polyglycosan bodies on some muscle fibers
Sivaraman I, Friedman NR, Prayson RA. J Clin Neurosci. 2016 Dec;34:222-223.	Phénotypage	Case report	NALCN	NA	NA	Type 2 muscle fiber atrophy, increased endomysial fibrosis, mild variation of fiber size, ragged red fibers
Rudnik-Schöneborn S, Deden F, Eggermann K, Eggermann T, Wieczorek D, Sellhaus B, Yamoah A, Goswami A, Claeys KG, Weis J, Zerres K. Muscle Nerve. 2016 Sep;54(3):496-500.	Phénotypage	Family report	BICD2	NA	NA	neurogenic muscle atrophy, type 2 fiber predominance
Lazier J, Mah JK, Nikolic A, Wei XC, Samedi V, Fajardo C, Brindle M, Perrier R, Thomas MA. Neuromuscul Disord. 2016 Jan;26(1):56-9.	Phénotypage	Case report	RYR1	NA	NA	Variable fiber size, round myofibers, increased connective tissue, central cores
Evangelista T, Bansagi B, Pyle A, Griffin H, Douroudis K, Polvikoski T, Antoniadi T, Bushby K, Straub V, Chinnery PF, Lochmüller H, Horvath R. Neuromuscul Disord. 2015 Jun;25(6):516-21.	Phénotypage	2 case report	TRPV4	NA	NA	increased variation in fibre size with both scattered and small groups of atrophic fibres. ATPase stain showed type I fibre predominance, areas of fibre type grouping, indicative of a chronic neuropathy.
Dohrn N, Le VQ, Petersen A, Skovbo P, Pedersen IS, Ernst A, Krarup H, Petersen MB. Am J Med Genet A. 2015 Apr;167A(4):731-43.	Phénotypage	Family report	ECEL1	NA	NA	Variation in fiber diameter, central nuclei (13WG), glycogen accumulation
Wilbe M, Ekvall S, Eurenius K, Ericson K, Casar-Borota O, Klar J, Dahl N, Ameur A, Annerén G, Bondeson ML. J Med Genet. 2015 Mar;52(3):195-202.	Phénotypage	Case report	MUSK	NA	NA	large variation in fibre size with significant atrophy, fibres co-expressed neonatal and developmental myosin, Increased amounts of loose connective tissue

Scoto M, Rossor AM, Harms MB, Cirak S, Calissano M, Robb S, Manzur AY, Martínez Arroyo A, Rodriguez Sanz A, Mansour S, Fallon P, Hadjikoumi I, Klein A, Yang M, De Visser M, Overweg-Plandsoen WC, Baas F, Taylor JP, Benatar M, Connolly AM, Al-Lozi MT, Nixon J, de Goede CG, Foley AR, Mcwilliam C, Pitt M, Sewry C, Phadke R, Hafezparast M, Chong WK, Mercuri E, Baloh RH, Reilly MM, Muntoni F. Neurology. 2015 Feb 17;84(7):668-79.,	Phénotypage	cohort report	DYNC1H1	NA	NA	Chronic denervation, some with core-like areas
Tan-Sindhunata MB, Mathijssen IB, Smit M, Baas F, de Vries JI, van der Voorn JP, Kluijt I, Hagen MA, Blom EW, Sistermans E, Meijers-Heijboer H, Waisfisz Q, Weiss MM, Groffen AJ. Eur J Hum Genet. 2015 Sep;23(9):1151-7.	Phénotypage	cohort report	MUSK	NA	NA	small and rounded atrophic fibers and an increased number of intracellular nuclei relative loss of type I fibers
Rossor AM, Oates EC, Salter HK, Liu Y, Murphy SM, Schule R, Gonzalez MA, Scoto M, Phadke R, Sewry CA, Houlden H, Jordanova A, Tournev I, Chamova T, Litvinenko I, Zuchner S, Herrmann DN, Blake J, Sowden JE, Acsadi G, Rodriguez ML, Menezes MP, Clarke NF, Auer Grumbach M, Bullock SL, Muntoni F, Reilly MM, North KN. Brain. 2015 Feb;138(Pt 2):293-310.	Phénotypage	cohort report	BICD2	NA	NA	marked variation in fibre size (5–150 mm) with increased internal nuclei increased connective tissue, whorled and split fibres and frequent mini-core like lesions
Chauveau C, Bonnemann CG, Julien C, Kho AL, Marks H, Talim B, Maury P, Arne-Bes MC, Uro-Coste E, Alexandrovich A, Vihola A, Schafer S, Kaufmann B, Medne L, Hübner N, Foley AR, Santi M, Udd B, Topaloglu H, Moore SA, Gotthardt M, Samuels ME, Gautel M, Ferreiro A. Hum Mol Genet. 2014 Feb 15;23(4):980-91.	Phénotypage	cohort report	TTN	NA	NA	Minicores, central nuclei, type 1 predominance
Bharucha-Goebel DX, Santi M, Medne L, Zukosky K, Dastgir J, Shieh PB, Winder T, Tennekoon G, Finkel RS, Dowling JJ, Monnier N, Bönnemann CG. Neurology. 2013 Apr 23;80(17):1584-9.	Phénotypage	cohort report	RYR1	NA	NA	Cores or multi-mini cores, central nuclei, type 1 fiber predominance, mild fibrosis
Dlamini N, Josifova DJ, Paine SM, Wraige E, Pitt M, Murphy AJ, King A, Buk S, Smith F, Abbs S, Sewry C, Jacques TS, Jungbluth H. . Neuromuscul Disord. 2013 May;23(5):391-8.	Phénotypage	Case report	UBA1	NA	NA	variation in fiber size, atrophic fibers, marked perivascular inflammation linked to acute denervation, type 1 fiber predominance, neurogenic changes
Davidson AE, Siddiqui FM, Lopez MA, Lunt P, Carlson HA, Moore BE, Love S, Born DE, Roper H, Majumdar A, Jayadev S, Underhill HR, Smith CO, von der Hagen M, Hubner A, Jardine P, Merrison A, Curtis E, Cullup T, Jungbluth H, Cox MO, Winder TL, Abdel Salam H, Li JZ, Moore SA, Dowling JJ. Brain. 2013 Feb;136(Pt 2):508-21.,	Phénotypage	Case report	TPM2 delK7	NA	NA	increased variation in fiber size, internalized nuclei, type 1 fiber predominance, large nemaline rods on Gomori trichrome staining and EM, numerous cores

Ravenscroft G, Thompson EM, Todd EJ, Yau KS, Kresoje N, Sivadorai P, Friend K, Riley K, Manton ND, Blumbergs P, Fietz M, Duff RM, Davis MR, Allcock RJ, Laing NG. Neuromuscul Disord. 2013 Feb;23(2):165-9.	Phénotypage	1 family report	GBE1	NA	NA	severely dystrophic pattern, marked fibrosis, fatty replacement, PAS positive inclusion
Yonath H, Reznik-Wolf H, Berkenstadt M, Eisenberg-Barzilai S, Lehtokari VL, Wallgren-Pettersson C, Mehta L, Achiron R, Gilboa Y, Polak-Charcon S, Winder T, Frydman M, Pras E. Prenat Diagn. 2012 Jan;32(1):70-4.	Phénotypage	2 case report	NEB	NA	NA	nemaline bodies
Yıldırım Y, Orhan EK, Iseri SA, Serdaroglu-Oflazer P, Kara B, Solakoğlu S, Tolun A. Hum Mol Genet. 2011 May 15;20(10):1886-92.	Phénotypage	1 family report	ERLIN2	NA	NA	normal muscle biopsy
Hernandez-Lain A, Husson I, Monnier N, Farnoux C, Brochier G, Lacène E, Beuvin M, Viou M, Manéré L, Claeys KG, Fardeau M, Lunardi J, Voit T, Romero NB. Eur J Med Genet. 2011 Jan-Feb;54(1):29-33.	Phénotypage	1 family report	RYR1	NA	NA	variable fiber size, core and rod/ nemaline bodies
Gurnett CA, Desruisseau DM, McCall K, Choi R, Meyer ZI, Talerico M, Miller SE, Ju JS, Pestronk A, Connolly AM, Druley TE, Weihl CC, Dobbs MB. Hum Mol Genet. 2010 Apr 1;19(7):1165-73.	Phénotypage	3 case report	MYBPC1	NA	NA	type 1 fiber atrophy in 1/3 patients

IRM médullaire						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Aragao MFVV, Holanda AC, Brainer-Lima AM, Petribu NCL, Castillo M, van der Linden V, Serpa SC, Tenório AG, Travassos PTC, Cordeiro MT, Sarteschi C, Valenca MM, Costello A. Nonmicrocephalic Infants with Congenital Zika Syndrome Suspected Only after Neuroimaging Evaluation Compared with Those with Microcephaly at Birth and Postnatally: How Large Is the Zika Virus "Iceberg"? AJNR Am J Neuroradiol. 2017 Jul;38(7):1427-1434. doi: 10.3174/ajnr.A5216. Epub 2017 May 18. PMID: 28522665; PMCID: PMC7959892.	Phénotypage	retrospective quantitative and qualitative MRI analysis in 12 congenital Zika syndrome patients.	Zika	NA	NA	Comparative analysis of spinal cord, spinal root and brain MRI in congenital Zika syndrome: 4 patients with AMC, 8 patients without AMC; 4/4 AMC+ patients had reduced thoracic spinal cord thickness, vs. 6/8 AMC- patients; in AMC+ patients, in entire spinal cord, in reduced and thinner conus medullaris anterior roots, and brain stem hypoplasia; no polymicrogyria.

Muscle MRI						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Carrera-García L, Natera-de Benito D, Dieterich K, de la Banda MGG, Felter A, Inarejos E, Codina A, Jou C, Roldan M, Palau F, Hoenicka J, Pijuan J, Ortez C, Expósito-Escudero J, Durand C, Nugues F, Jimenez-Mallebrera C, Colomer J, Carlier RY, Lochmüller H, Quijano-Roy S, Nascimento A. CHRNG-related nonlethal multiple pterygium syndrome: Muscle imaging pattern and clinical, histopathological, and molecular genetic findings. Am J Med Genet A. 2019 Jun;179(6):915-926. doi: 10.1002/ajmg.a.61122. Epub 2019 Mar 14. PMID: 30868735.	Phénotypage	Case series				marked muscle bulk reduction is the predominant finding, mostly affecting the spinal erector muscles and gluteus maximus. Fatty infiltration was only observed in deep paravertebral muscles and distal lower limbs
Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. A novel neuromuscular form of glycogen storage disease type IV with arthrogryposis, spinal stiffness and rare polyglucosan bodies in muscle. Neuromuscul Disord. 2016 Oct;26(10):681-687. doi: 10.1016/j.nmd.2016.07.005. Epub 2016 Jul 25. PMID: 27546458.	Phénotypage	Case report				fibroadipose muscle replacement but sparing ofthe sartorius, gracilis, adductor longus and vastus intermedialis muscles.
Jarraya M, Quijano-Roy S, Monnier N, Béhin A, Avila-Smirnov D, Romero NB, Allamand V, Richard P, Barois A, May A, Estournet B, Mercuri E, Carlier PG, Carlier RY. Whole-Body muscle MRI in a series of patients with congenital myopathy related to TPM2 gene mutations. Neuromuscul Disord. 2012 Oct 1;22 Suppl 2:S137-47. doi: 10.1016/j.nmd.2012.06.347. PMID: 22980765.	Phénotypage	Case series				predominant involvement of masticatory and distal leg muscles with the other regions rel-atively spared
Mercuri E, Manzur A, Main M, Alsopp J, Muntoni F. Is there post-natal muscle growth in amyoplasia? A sequential MRI study. Neuromuscul Disord. 2009 Jun;19(6):444-5. doi: 10.1016/j.nmd.2009.03.006. Epub 2009 May 27. PMID: 19477646.	Phénotypage	Case report				diffuse involvement of both upper and lower limbs with relativepreservation of the trunk muscles; elative sparing of thebiceps femoris, sartorius, gracilis and, on the right thigh, partly alsoof the vasti; overall increased muscle bulk as a result not onlyof hypertrophy of the muscles which were relatively spared in theprevious scans, but also due to the appearance of muscle ofapparently normal signal intensity in previously severely affectedmuscles

Philpot J, Counsell S, Bydder G, Sewry CA, Dubowitz V, Muntoni F. Neonatal arthrogryposis and absent limb muscles: a muscle developmental gene defect? Neuromuscul Disord. 2001 Jul;11(5):489-93. doi: 10.1016/s0960-8966(00)00221-2. PMID: 11404123.	Phénotypage	case report				virtual absence of muscles in the limbs with sparing of the axial muscle

Myasthénie						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Barnes PR, Kanabar DJ, Brueton L, Newsom-Davis J, Huson SM, Mann NP, Hilton-Jones D. Neuromuscul Disord. 1995 Jan;5(1):59-65.	Phénotypage	series of cases				4 AMC cases born from a mother retrospectively diagnosed with MG : 3 were born alive but died in the neonatal period, 1 was alive with isolated DA, and another died IU at 17WG.
Brueton LA, Huson SM, Cox PM, Shirley I, Thompson EM, Barnes PR, Price J, Newsom-Davis J, Vincent A. Am J Med Genet. 2000 May 1;92(1):1-6.	Phénotypage	series of cases				6 patients with FADS, whose mother later had a diagnosis of asymptomatic MG with the identification of AChR ab.
Cantagrel S, Maury L, Yamamoto AM, Maheut J, Toutain A, Castelnau P. Am J Perinatol. 2002 Aug;19(6):297-301.	Phénotypage	case report				1 case of AMC : major akinesia with hydramnios, then arthrogryposis and craniosysnostosis at birth. Asymptomatic mother with AChR ab vs gamma subunit, elevated gamma/epsilon ratio. Outcome : progressive improvement of contracture, persistence of facial diplegia.
Carr SR, Gilchrist JM, Abuelo DN, Clark D. Obstet Gynecol. 1991 Sep;78(3 Pt 2):485-9.	Phénotypage	case report				2 AMC cases with prenatal onset born from a mother with MG, who died in the neonatal period. She previously had a child with TNMG.
Chieza JT, Fleming I, Parry N, Skelton VA. Int J Obstet Anesth. 2011 Jan;20(1):79-82.	Phénotypage	case report				Diagnosis of MG in a previously asymptomatic mother at 20 WG, thanks to identification of maternal symptoms associated to a Pena Shokeir fetal sequence. Elevated AChR antibodies concentrations. Child born at 34 WG with AMC, died soon after birth.

Dalton P, Clover L, Wallerstein R, Stewart H, Genzel-Boroviczeny O, Dean A, Vincent A. Neuromuscul Disord. 2006 Aug;16(8):481-91.	Phénotypage	experimental : cohort of AMC patients vs controls				detection of AChR ab in asymptomatic mothers with AMC children : n=179 vs 20 controls. 3/179 were positive (1.5%). For seronegative ones : reactivity with undefined muscle or neuronal antigens in 33% of cases, especially when babies had CNS involvement, suggesting the existence of other antibodies? No MUSK.
Dinger J, Prager B. Neuromuscul Disord. 1993 Jul;3(4):335-9. Review.	Phénotypage	case report				1 AMC case with prenatal onset in a mother with MG : born alive, favorable outcome of contractures despit persistant hypotonia.
Dulitzky F, Sirota L, Landman J, Homburg R. Helv Paediatr Acta. 1987 Oct;42(2-3):173-6.	Phénotypage	case report				1 AMC case with prenatal onset born from a asymptomatic mother, died at 37 do.
Eymard B, Morel E, Dulac O, Moutard-Codou ML, Jeannot E, Harpey JP, Rondot P, Bach JF. Rev Neurol (Paris). 1989;145(10):696-701. French.	Phénotypage	cohort of children born from mother with MG				42 pregnancies in 39 mothers with MG : 3 AMC cases. At least 1 was born alive with favorable outcome.
Gilhus NE, Hong Y. Eur J Neurol. 2018 Dec;25(12):1402-1409.	Phénotypage	litterature review				in a cohort of 127 children born from MG mothers, 3,9% (5/127) had "severe skeletal anomalies" including 2 siblings from the same mother, whose brother has severe myopathy.
Hacohen Y, Jacobson LW, Byrne S, Norwood F, Lall A, Robb S, Dilena R, Fumagalli M, Born AP, Clarke D, Lim M, Vincent A, Jungbluth H. Neurol Neuroimmunol Neuroinflamm. 2014 Dec 23;2(1):e57.	Phénotypage	series of cases				8 pts/4 families born from MG mothers including one asymptomatic. 4 have limb contractures. The 2 children from asymptomatic mother with antibodies directed against the foetal form have multiple congenital contracutres of good outcome despite mild learning difficulties and unilateral hear loss for one/ normal development for the other). 1 child was diagnosed with AMC, born from a woman who became symptomatic from 20WG with high levels of uncharacterized AChR antibodies, who further had another child with NMG that died in the neonatal period. The last child had hips and fingers contractures, and was born from a mother that was symptomatic before pregnancy with AChR antibodies, mostly directed against the fetal form; he died at 5 mo. Ratio gamma/epsilon may be correlate

						with severity. High titers of antibodies blocking the fœtal function of AChR are rare in women with MG. These children usually have normal electrophysiological studies and low response to traditionnal drugs.
Hellmund A, Berg C, Geipel A, Müller A, Gembruch U. Arch Gynecol Obstet. 2016 Oct;294(4):697-707.	Phénotypage	cohort of FADS				cohort of 79 FADS cases (severe cases, TOP 86%). 20/79 mothers were tested for AChR antibodies, only 1 was positive in a asymptomatic woman. This mother had fetal FADS in two subsequent pregnancies, 1 with TOP and 1 with live birth but AMC and requirement for respiratory support, ongoing physiotherapy, then minor residual troubles such as attention issues, abnormal motor function, need for hearing aid devices and recurrent lung infections.
Hoff JM, Daltveit AK, Gilhus NE. Acta Neurol Scand Suppl.2006;183:26-7.	Phénotypage	cohort of children born from mother with MG				study of 176 births from 79 mothers with previously diagnosed MG. 4 newborns with severe AMC or FADS including twins, all died in the neonatal period. Previous or further siblings with NMG --> siblings of an affected child either with NMG or AMC have an increased risk to develop either NMG or AMC, independantly of MG mother's clinical state.
Hoff JM, Daltveit AK, Gilhus NE. Eur J Neurol. 2007 Jan;14(1):38-43.	Phénotypage	cohort of mothers with MG				135 birth from 73 mothers with MG were identified. 0 case of arthrogryposis.
Hoff JM, Loane M, Gilhus NE, Rasmussen S, Daltveit AK. Eur J Obstet Gynecol Reprod Biol. 2011 Dec;159(2):347-50.	Phénotypage	cohort of AMC patients				757 AMC cases. None was related to maternal MG --> MG does not appear as a risk factor for arthrogryposis
Holmes LB, Driscoll SG, Bradley WG. J Pediatr. 1980 Jun;96(6):1067-9.	Phénotypage	case report				1 AMC case with prenatal onset (both hands) who died in the neonatal period, born from a mother with known MG, décès NN. She had 2 other children : 1 with TNMG, and 1 normal child.
Mikou F, Kaouti N, Ghazli M, El Kerroumi M, Sefrioui O, Morsad F, Matar N, Elmoutawakil B, Mouden M, Gam I, Slassi I. J Gynecol Obstet Biol Reprod (Paris). 2003 Nov;32(7):660-2.	Phénotypage	case report and review				Patient with known MG, who had a child with AMC associated to pulmonary hypoplasia, who died at 19 do. Review of 27 cases from 12 publications of fetuses presenting with arthrogryposis.
Morel E, Bach JF, Briard ML, Aubry JP. J Neuroimmunol. 1984 Aug;6(5):313-7.	Phénotypage	case report				1 AMC case born from a MG mother. AChR ab were found in amniotic fluid.

Moutard-Codou ML, Delleur MM, Dulac O, Morel E, Voyer M, De Gamara E. Presse Med. 1987 Apr 11;16(13):615-8. French.	Phénotypage	cases reports				2 AMC cases with prenatal onset : bilateral hand contractures, pulmonary hypoplasia, hydramnios, who died in the neonatal period. Born from a previously MG diagnosed mother. + 1 AMC case born from a previously undiagnosed mother with favourable outcome but persistance of facial diplegia.
Pasternak JF, Hageman J, Adams MA, Philip AG, Gardner TH. J Pediatr. 1981 Oct;99(4):644-6.	Phénotypage	case report				1 AMC case with prenatal onset born from a mother with MG (contractures of fingers, toes), favorable outcomes.
Polizzi A, Huson SM, Vincent A. Teratology. 2000 Nov;62(5):332-41. Review.	Phénotypage	review				reviewed the role of AChR antibodies in mothers with MG and affected babies : found transient signs of MG in 10-15% of babies born to mother with MG and only few cases of AMC (identification in the litterature of 32 AMC cases from 13 mothers with MG). The severity of AMC in children born to MG mothers is variable and has not been found to correlate with the severity of the mother's MG, neither at onset time nor during pregnancy. In anti AChR ab associated AMC, fetal or NN death is common, and is sometimes associated to CNS anomalies. HIgh recurrent risk for further pregnancies.
Reimann J, Jacobson L, Vincent A, Kornblum C. Neurology. 2009 Nov 24;73(21):1806-8.	Phénotypage	case report				1 AMC case with prenatal onset (reduced fetal movements, FADS). AChR ab directed against the gamma subunit were found at high level in the mother (not in the child at 4 months old), which was asymptomatic but had decrement on electrophysiology, she developped symptoms only 2 years after delivery. She also reported a TOP at 26WG for FADS. MB in children showed complement deposition colocalizating with gamma subunits.
Riemersma S, Vincent A, Beeson D, Newland C, Hawke S, Vernet-der Garabedian B, Eymard B, Newsom-Davis J. J Clin Invest. 1996 Nov 15;98(10):2358-63.	Phénotypage	experimental				Serum analysis of 5 patients previously described (Morel, Vernet der, Vincent and Barnes) : high levels of AChR directed against the foetal form. Also tested 20 sera from women that had a child with AMC : none had AChR ab but 3 had a functional inhibition of the receptor.

Shepard MK. Birth Defects Orig Artic Ser. 1971 Feb;7(2):127.	Phénotypage	case report				2 AMC cases with prenatal onset in a patient with MG, IU death
Smit LM, Barth PG. Dev Med Child Neurol. 1980 Jun;22(3):371-4.	Phénotypage	case report				1 AMC case with prenatal onset (lower limbs only) born from a mother with MG. Results of ultrastructural studies of motor endplates described in another article in 1984 by Smit PMID: 6094728
Stoll C, Ehret-Mentre MC, Treisser A, Tranchant C. Prenat Diagn. 1991 Jan;11(1):17-22. Review.	Phénotypage	case report				2 AMC cases with prenatal onset born from mother with previously diagnosed MG : 1 born alive with neonatal death, 1 for which TOP was chosen.
Tranchant C, Ehret C, Labouret P, Gasser B, Warter JM. Rev Neurol (Paris). 1991;147(1):62-4. Review. French.	Phénotypage	case report				2 AMC cases with prenatal onset born from a woman with MG : 1 stillbirth, 1 TOP.
Vincent A, McConville J, Farrugia ME, Bowen J, Plested P, Tang T, Evoli A, Matthews I, Sims G, Dalton P, Jacobson L, Polizzi A, Blaes F, Lang B, Beeson D, Willcox N, Newsom-Davis J, Hoch W. Ann N Y Acad Sci. 2003 Sep;998:324-35. Review.	Phénotypage	cohort of mothers who had AMC children				Look for antibodies in 200 mothers who previously had a child with a diagnosis of AMC : only a small proportion of the women had histories of MG. In the remainning mothers, AchR were found in only two individuals, for the seronegative ones, antibodies binding to different neonatal mouse tissues were found, suggesting that other antibodies might be involved in causing AMC.
Vincent A, Newland C, Brueton L, Beeson D, Riemersma S, Huson SM, Newsom-Davis J. Lancet. 1995 Jul 1;346(8966):24-5.	Phénotypage	case report				case of a mother with asymptomatic MG due to AChR ab at high concentration : 1 normal pregnancy followed by 6 pathologic pregnancies, 1 stillborn at 35WG, then 5 TOP at 17-18WG for AMC after interruption of active fetal movements from 15W. Antibodies were also found in fetal sera.
Vincent A, Waters P, Leite MI, Jacobson L, Koneczny I, Cossins J, Beeson D. Ann N Y Acad Sci. 2012 Dec;1274:92-8.	Phénotypage	experimental				3 exemples of mother with AMC children. 1 asymptomatic mother with 3 children with AMC for which anti MUSK antibodies were evidenced. 1 mother with symptomatic MG and AChR antibodies mostly against the fetal form, 4 children with AMC. 1 asymptomatic mother with 4 AMC children and AChR antibodies mostly against the fetal form. No detail about clinical evolution.

Aminoacidopathies						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Kurolap A, Armbruster A, Hershkovitz T, Hauf K, Mory A, Paperna T, HannappelE, Tal G, Nijem Y, Sella E, Mahajnah M, Ilivitzki A, Hershkovitz D, Ekhilevitch, N, Mandel H, Eulenburg V, Baris HN. Am J Hum Genet. 2016 Nov 3;99(5):1172-1180.	Phénotypage	series of 4 cases				5 patients from 2 families who presented at birth with facial dysmorphism, encephalopathy, arthrogryposis (multiple levels in 4/5), hypotonia and respiratory failure. Only one survived with severe sequellae on neurodevelopment. High glycine in CSF but not in blood in 2. Identificaion of truncating mutations in SLC6A9 encoding GLYT1.
Chiang MC, Huang SF, Hsueh C, Lai MW, Hou JW. Turk J Pediatr. 2008 Sep-Oct;50(5):492-4.	Phénotypage	case report				restrictive dermopathy associated with dysmorphic features, generalized arthrogyrposis, pulmonary hypoplasia. 1 case of restrictive dermopathy with urine and blood generalized organic aciduria and low free carnithine levels.
El-Hattab AW, Shaheen R, Hertecant J, Galadari HI, Albaqawi BS, Nabil A, Alkuraya FS. J Inherit Metab Dis. 2016 May;39(3):373-381.	Phénotypage	case report				Mutation in PGDH, PSAT, PSP result in Neu-Laxova syndrome (lethal, with contractures and pterygia) to less severe disorders. 3 case reports with serine biosynthesis defects, one with contractures and hypertonia with PSAT1 mutation. Plasma levels of serine decreased.

Citric acid cycle						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
van Dijk T, Ferdinandusse S, Ruiter JPN, Alders M, Mathijssen IB, Parboosingh JS, Innes AM, Meijers-Heijboer H, Poll-The BT, Bernier FP, Wanders RJA, Lamont RE, Baas F. Eur J Hum Genet. 2018 Dec;26(12):1752-1758.	Phénotypage	series of cases				Mutation in COASY encoding coenzyme A synthase for CoA synthesis. CoA involved in numerous enzyme reactions (PDH, 2 ketoglutarate dehydrogenase, acyl CoA synthase..) affecting metabolic processes (citric acid cycle, fatty acid oxidation, amino-acid degradation) + other roles. 4 individuals with PCH, prenatal microcephaly, arthrogryposis. Lethal (1 TOP at 22WG, the other died at 1 month)

Winters L, Van Hoof E, De Catte L, Van Den Bogaert K, de Ravel T, De Waele L, Corveleyn A, Breckpot J. Eur J Paediatr Neurol. 2017 Sep;21(5):745-753.	Phénotypage	2 case reports				in-house panel in 2 cases of FADS. PDHA1 in a fœtus with TOP. First case ever reported. Had brain anomalies.

Mitochondrial disorders						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Piard J, Umanah GKE, Harms FL, Abalde-Atristain L, Amram D, Chang M, Chen R, Alawi M, Salpietro V, Rees MI, Chung SK, Houlden H, Verloes A, Dawson TM, Dawson VL, Van Maldergem L, Kutsche K. Brain. 2018 Mar 1;141(3):651-661.	Phénotypage	case report				Mut ATAD1 encoding Thorase that plays a role in the function and integrity of mitochondria and peroxisomes. 3 siblings presenting with severe, lethal encephalopathy, stiffness and arthrogryposis. For one only adducted thumbs at birth and AMC at 2 months, his brother adducted thumbs and clenched toes, for the other one AMC from birth with adducted thumbs, ulnar deviation of wrists and club feet.
Sivaraman I, Friedman NR, Prayson RA. J Clin Neurosci. 2016 Dec;34:222-223.	Phénotypage	case report				Mut NALCN encoding a sodium leak channel lead to neurodevelopmental impairment, hypotonia and congenital contracutes, episodic ataxia, neuroaxonal dystrophy. AMC especially associated with de novo heterozygous mutations. 1 case here, metabolic tests was normal except for mildly elevated multiple metabolites in urine suggestive of mild dysfunction of multiple mitochondrial enzymes. Muscle biopsies reveales ragged red fibe changes and an increased number of mitochondria, so possible mitochondrial bioenergetic dysfunction.
Fukumura S, Ohba C, Watanabe T, Minagawa K, Shimura M, Murayama K, Ohtake A, Saitsu H, Matsumoto N, Tsutsumi H. J Hum Genet. 2015 Sep;60(9):509-13.	Phénotypage	case report				Mut GMF2, a nuclear gene responsible for Leigh syndrome. 2 females with AMC, OA, severe MR. CSF lactate/pyruvate not increased but lactate peak on MRI, CSF and serum lactate/pyruvate beta-hydroxybutyrate/acetoacetate ratios high, levels of owidative phosphorylation in skin fibroblasts reduced. Contractures in all extremities

Ajit Bolar N, Vanlander AV, Wilbrecht C, Van der Aa N, Smet J, De Paepe B, Vandeweyer G, Kooy F, Eyskens F, De Latter E, Delanghe G, Govaert P, Leroy JG, Loeys B, Lill R, Van Laer L, Van Coster R. Hum Mol Genet. 2013 Jul 1;22(13):2590-602.	Phénotypage	case report				Mut IBA57, a gene involved in biosynthesis of mitochondrial proteins. 2 siblings died perinatally, hypotonia, respiratory insufficiency, arthrogryposis, microcephaly, congenital brain malformations, hyperglycinemia. Catalytic activities of the mitochondrial respiratory complexes I and II were deficient suggesting an inborn error in mitochondrial biogenesis
Wilnai Y, Seaver LH, Enns GM. Am J Med Genet A. 2012 Sep;158A(9):2353-7.	Phénotypage	case report				Mut SURF1, causing mitochondrial respiratory chain complex IV deficiency and Leigh syndrome. Case of a boy with amyoplasia congenita in the neonatal period, then at 10.5 months hypotonia and choreathetosis on a viral onset. CSF lactate levels were elevated. " a variety of mitochondrial respiratory chain complex deficiencies have been associated with contractures of varying severity, therefore mitochondrial disorders should be considered in the differential diagnosis of neonatal arthogryposis, especially if other characteristic findings such as lactic acidemia or basal ganglia abnormalities are present.
Tulinius M, Oldfors A. Semin Fetal Neonatal Med. 2011 Aug;16(4):229-35.	Phénotypage	review				Review of muscular manifestations of neonates and infants with mitochondrial diseases. Table : arthrogryposis associated with complex V deficiencies with mutation in ATPAF2 or TMEM70 (nuclear assembly factor genes of the RC enzyme complex subunits) and with combined deficiencies with mutations in EFG1 and MRPS16 (nuclear translation factor genes of the RCEC)
Spiegel R, Khayat M, Shalev SA, Horovitz Y, Mandel H, Hershkovitz E, Barghuti F, Shaag A, Saada A, Korman SH, Elpeleg O, Yatsiv I. J Med Genet. 2011 Mar;48(3):177-82.	Phénotypage	case report				Mut TMEM70 causing ATP synthase deficiency (complex V deficiency), profound lactic acidosis and 3-methylgluthaconic aciduria. Joint contractures attributed to fetal hypertonia while usually fetal hypotonia is reported. Contractures improved with time. Usually also hypertrophic cardiomyopathy, life threatening lactic acidosis.
McPherson E, Zabel C. Mitochondrial mutation in a child with distal arthrogryposis. Am J Med Genet A. 2006 Jan 15;140(2):184-5. doi: 10.1002/ajmg.a.31041. PMID: 16353243.10.1002/ajmg.a.31041. PMID: 16353243.	Phénotypage	respiratory chain studies				Distal arthrogryposis with normal milestones. Talipes equinovarus (requiring surgery), camptodactyly, adducted thumbs. MELAS T3271C.

von Kleist-Retzow JC, Cormier-Daire V, Viot G, Goldenberg A, Mardach B, Amiel J, Saada P, Dumez Y, Brunelle F, Saudubray JM, Chrétien D, Rötig A, Rustin P, Munnich A, De Lonlay P. Antenatal manifestations of mitochondrial respiratory chain deficiency. J Pediatr. 2003 Aug;143(2):208-12. doi: 10.1067/S0022-3476(03)00130-6. PMID: 12970634.	Phénotypage	cohort study				300 patients with proven respiratory chain enzyme deficiency, retrospectively reviewed for fetal development. 2 cases : case 1 talipes equinovarus and Complex III deficiency. Case 2 : arthrogryposis and complex V deficiency, Conclusion : a number of metabolic diseases undergo a symptom-free period, but respiratory chain disorders may have an early antenatal expression, because of gene expression in the embryofetal period.
Gire C, Girard N, Nicaise C, Einaudi MA, Montfort MF, Dejode JM. Clinical features and neuroradiological findings of mitochondrial pathology in six neonates. Childs Nerv Syst. 2002 Nov;18(11):621-8. doi: 10.1007/s00381-002-0621-0. Epub 2002 Sep 12. PMID: 12420122.	Phénotypage	cohort study				MRI in 6 neonate cases with diagnosis of mitochondrial cytopathy. Antenatal onset in 5 cases. Arthrogryposis, polyhydramnios. Complex I deficiency. Conclusion : fetal expression of the disease.
Vielhaber S, Feistner H, Schneider W, Weis J, Kunz WS. Mitochondrial complex I deficiency in a female with multiplex arthrogryposis congenita. Pediatr Neurol. 2000 Jan;22(1):53-6. doi: 10.1016/s0887-8994(99)00097-1. PMID: 10669207.	Phénotypage	case report				girl with AMC (all distal limbs, limited mouth opening and closing, flexion contractures of fingers II-V in the proximal IP joints, camptodactyly, ulnar deviation of the hands, limited rotation and extension of the elbows) and mild myopathy. Complex I deficiency
Laubscher B, Janzer RC, Krähenbühl S, Hirt L, Deonna T. Ragged-red fibers and complex I deficiency in a neonate with arthrogryposis congenita. Pediatr Neurol. 1997 Oct;17(3):249-51. doi: 10.1016/s0887-8994(97)00082-9. PMID: 9390702.	Phénotypage					Contractures of wrists, elbows, shoulders, hips and knees, severe talipes equinovarus. Death from respiratory failure at 14 days. Autopsy : numerous ragged-red fibres and central nervous system abnormalities consistent with a mitochondrial disease. Complex I deficiency. Conclusion : mitochondrial cytopathies can be associated with AMC and should be sought in children presenting with severe arthrogyrposis.
Lee JS, Hwang JS, Ryu KH, Lee EH, Kim SH. Mitochondrial respiratory complex I deficiency simulating spinal muscular atrophy. Pediatr Neurol. 2007 Jan;36(1):45-7. doi: 10.1016/j.pediatrneurol.2006.07.007 . PMID: 17162196..	Phénotypage	case report				1/2 patients with contractures of wrists and great toe since birth. Complex I deficiency

Enns GM, Hoppel CL, DeArmond SJ, Schelley S, Bass N, Weisiger K, Horoupian D, Packman S. Relationship of primary mitochondrial respiratory chain dysfunction to fiber type abnormalities in skeletal muscle. Clin Genet. 2005 Oct;68(4):337-48. doi: 10.1111/j.1399-0004.2005.00499.x. PMID: 16143021.	Phénotypage					Children with variation in the size and proportion of type 1 and 2 muscle fibers on MB. Analysis of muscle mitochondrial respiratory chain in 10 children. 1 child with distal extremity contractures, hypotonia, torticollis. Complex I deficiency. Conclusion : mitochondrial electron transport chain disorders are a cause of fiber type abnormalities and should be investigated even in the absence of histological features of mitochondrial disorders.
De Meirleir L, Seneca S, Lissens W, De Clercq I, Eyskens F, Gerlo E, Smet J, Van Coster R. Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. J Med Genet. 2004 Feb;41(2):120-4. doi: 10.1136/jmg.2003.012047. PMID: 14757859; PMCID: PMC1735674.	Phénotypage	case report				Mutations in the complex V assembly gene ATPAF2, rocker bottom feet and flexion contractures of the limbs associated with camptodactylia. Hypertonic at birth.
de Koning TJ, de Vries LS, Groenendaal F, Ruitenbeek W, Jansen GH, Poll-The BT, Barth PG. Pontocerebellar hypoplasia associated with respiratory-chain defects. Neuropediatrics. 1999 Apr;30(2):93-5. doi: 10.1055/s-2007-973467. PMID: 10401692.	Phénotypage	case report				Decreased fetal movements, multiple contractures (np). Pontocerebellar hypoplasia and cortical and diffuse periventricular white matter abnormalities. Biochemical analysis of skin fibroblast demonstrated multiple deficiencies of respirtory chain enzymes. Complex I, III, IV deficiency. conclusion : the diagnosis workup of pontocerebellar hypoplasia should include search for respiratory chain impairement.

Peroxisomal disorders						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Nakano K, Zhang Z, Shimozawa N, Kondo N, Ishii N, Funatsuka M, Shirakawa S, Itoh M, Takashima S, Une M, Kana-aki RR, Mukai K, Osawa M, Suzuki Y. J Pediatr. 2001 Dec;139(6):865-7.	Phénotypage	case report				D-bifunctional protein deficiency (gene identified later : HSD17B4) responsible for abnormal peroxisomal beta-oxidation. Polyhydramnios, chylous fetal onset ascites, generalized hypotonia at birth with dysmorphism, claw hands and hammer toes probably because of decreased IU movement (first description) cystic lesion of the anterior horns, peripheral pulmonary artery stenosis.

Lysosomal disorders

Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
van der Beek J, Jonker C, van der Welle R, Liv N, Klumperman J. disease. J Cell Sci. 2019 May 15;132(10). pii: jcs189134.	Phénotypage	review				CHEVI = class C Homologs in Endosome Vesicle interaction = Multi Subunits Tethering Complex involved in endo lysosomal transport steps. Mutations in CHEVI subunits lead to ARC syndrome (Arthrogryposis, renal dysfunction and Cholestasis), mutation in VPS33B and VIPAS39, caused by imparied alpha granule formation because the transport of cargo proteins from the golgi to alpha granule progenitor organelles is disrupted
BenHamida E, Ayadi I, Ouertani I, Chammem M, Bezzine A, BenTmime R, Attia L, Mrad R, Marrakchi Z. Pan Afr Med J. 2015 Jun 10;21:110.	Phénotypage	case report				Gaucher disease : deficiency in glucocerebrosidase, the most common type of lysosomal storage disorders. Type 2 manifests either prenatally or in the first months of life. Sometimes lethal in the perinatal period but rarely : presents with non immune hydrops fetalis. Here : case report with hydrops fetalis and hypokinesia in utero. At birth arthrogryposis and hypertonia. Low glucerebrosidase enzymatic activity on leucocytes. Died in the first day of life. Diagnosis confirmed by sequencing of GBA1. Conclusion : hydrops fetalis are associated with several lysosomal storage disorders, when assiciated to hepatosplenomegaly, arthrogryposis and ichtyosis, GD should be sought.
Cullinane AR, Straatman-Iwanowska A, Zaucker A, Wakabayashi Y, Bruce CK, Luo G, Rahman F, Gürakan F, Utine E, Ozkan TB, Denecke J, Vukovic J, Di Rocco M, Mandel H, Cangul H, Matthews RP, Thomas SG, Rappoport JZ, Arias IM, Wolburg H, Knisely AS, Kelly DA, Müller F, Maher ER, Gissen P. Nat Genet. 2010 Apr;42(4):303-12. Erratum in: Nat Genet. 2011 Mar;43(3):277.	Phénotypage	series of cases				First description of ARC syndrome in link with VIPAR/VIPAS39 mutations. Effects of VIPAR et VPS33B mutations : modify apical basolateral polaritiy in the liver and kidney, leads to protein destruction by mis-sorting, i.e. sorting toward late endosomes and lysosomes where protein degradation ocurs.
Gissen P, Johnson CA, Morgan NV, Stapelbroek JM, Forshew T, Cooper WN, McKiernan PJ, Klomp LW, Morris AA, Wraith JE, McClean P, Lynch SA, Thompson RJ, Lo B, Quarrell OW, Di Rocco M, Trembath RC, Mandel H, Wali S, Karet FE, Knisely AS, Houwen RH, Kelly DA, Maher ER. Nat	Phénotypage	series of cases				First description of ARC syndrome in link with VPS33B mutations

Genet. 2004 Apr;36(4):400-4.						
Mignot C, Gelot A, Bessières B, Daffos F, Voyer M, Menez F, Fallet Bianco C, Odent S, Le Duff D, Loget P, Fargier P, Costil J, Josset P, Roume J, Vanier MT, Maire I, Billette de Villemeur T. Am J Med Genet A. 2003 Jul 30;120A(3):338-44.	Phénotypage	series of perinatal Gaucher disease cases				Gaucher Disease = purely visceral, sometimes neurological signs. Most often after a symptom-free period but in rare cases with fetal onset. 8 original cases of GD with fetal onset : non immune hydrops fetalis, HSMG, ichthyosis, arthrogryposis, facial dysmorphy. Often IU fetal death or in the first three months. Review of natural history of 41 cases of perinatal-lethal gaucher disease. In 3/8 cases, arthrogryposis was diagnosed prenatally. Arthrogryposis ranged from severe generalized contractures to distal joint contractures (camptodactyly, fixed overlapping fingers, club foot). Hypokinesia 43% and may be associated with contractures (30%).

Metabolic disorders						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Schorling DC, Rost S, Lefeber DJ, Brady L, Müller CR, Korinthenberg R, Tarnopolsky M, Bönnemann CG, Rodenburg RJ, Bugiani M, Beytia M, Krüger M, van der Knaap M, Kirschner J. Neurology. 2017 Aug 15;89(7):657-664.	Phénotypage	case report				5 patients from 3 families. 3 had congenital contractures. Died < 1 yo. Mutations in ALG14 causing a glycosylation defect (N-glycosylation). Delineation of a new genetic syndrome combining myasthenic features and severe neurodegeneration with therapy refractory epilepsy. Gene previously identified in a childhood onset isolated myasthenic syndrome. Here no clear glycosylation abnormalities could be observed of transferrin in the only family were it has been done.
Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. Neuromuscul Disord. 2016 Oct;26(10):681-687.	Phénotypage	case report				Glycogen storage disease type IV. 3 forms based on the time of onset. 1st type antenatal onset with FADS and AMC. Neuromuscular forms with FADS, lethal myopathy or mild hypotonia and weakness. Case : 3 yo boy with arthrogryposis (neck and upper limbs), rigid spine. Polyglucosan in muscle. GBE1 analyssis. Recognizable mMRI pattern. Ref 8 à 14 : cases of antenatal AMC GSD IV. cf Raju 2008.

Hunter JM, Kiefer J, Balak CD, Jooma S, Ahearn ME, Hall JG, Baumbach-Reardon L. Am J Med Genet A. 2015 May;167A(5):931-73.	Phé noty page	review				syndromes avec contractures LX : SLC9A6 (ganglioside GM2), EIF2S3 (mitochondrial), MTM1 (myopathy myotubular), OFD1, AP1S2 (iron deposits), RBM10, Tiemann (elevated glycogene in muscle).
Wu PL, Yang YN, Tey SL, Yang CH, Yang SN, Lin CS. Pediatr Int. 2015 Aug;57(4):746-9.	Phé noty page	case report and review of the littérature				case = female infant with PFK deficiency (= GSD type VII), floppy infant syndrome, congenital joint contracture, cleft palate, duplication of the pelvicalyceal system, died at corrected age of 6 mo due to respiratory failure. Review of 13 other cases in the litterature : arthrogryposis occurs in 64.3%, also hypotonia, encephalopathy, cardiomyopathy, low survival rate.
Ganetzky R, Izumi K, Edmondson A, Muraresku CC, Zackai E, Deardorff M, Ganesh J. Am J Med Genet A. 2015 Oct;167A(10):2411-7.	Phé noty page	case report				case = newborn female with AMC and CDG DPAGT1, fœtal onset. To date 17 reported cases of DPAGT1 CDG including two similar cases : CDG has to be considered as a differential diagnosis for arthrogryposis.
Clayton PT, Grunewald S. J Inherit Metab Dis. 2009 Dec;32 Suppl 1:S137-9.	Phé noty page	case report				first detailed description of the clinical phenotype CDG In due to RFT1 mutations. It was a severe disorder affecting intrauterine development and movement, and leading to intrauterine growth retardation. The child was born with several musculoskeletal abnormalities including arthrogryposis. Postnatally, severe reflux and irregular bowl movements contributed to failure to thrive. The patient showed very little development and no vision and suffered from drug-resistant epilepsy. Abnormal coagulation resulted in thrombosis and the patient died at the age of 4 years from a pulmonary embolus.
Ravenscroft G, Thompson EM, Todd EJ, Yau KS, Kresoje N, Sivadorai P, Friend K, Riley K, Manton ND, Blumbergs P, Fietz M, Duff RM, Davis MR, Allcock RJ, Laing NG. Neuromuscul Disord. 2013 Feb;23(2):165-9.	Phé noty page	family case report				WES in 2 Siblings of fœtal akinesia with lethal MPS, TOP. Splice and missense mutations in GBE1, which has a severe fœtal akinesia subphenotype. Confirmation on muscle pathology and biochemically.
Tiemann C, Bührer C, Burwinkel B, Wirtenberger M, Hoehn T, Hübner C, van Landeghem FK, Stoltenburg G, Obladen M. Am J Med Genet A. 2005 Aug 30;137(2):125-9.	Phé noty page	series of cases				3 male siblings from inbred family : arthrogryposis multiplex congenita, deafness, inguiinal hernia, hiccup like diaphragmatic contractions, inability to suck, died from respiratory failure during the first 3 months. Muscle : elevated glycogen content, deposits in liver as well. AR/LX? No mutation in GBE1 or SMN1 del.

Shin YS, Plöchl E, Podskarbi T, Muss W, Pilz P, Puttinger R. J Inherit Metab Dis. 1994;17(1):153-5.	Phénotypage	case report				case report of a child with generalized arthrogryposis and phosphorylase b kinase deficiency (GSD type IXb)

Congenital heart anomalies						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Kane MS, Diamonstein CJ, Hauser N, Deeken JF, Niederhuber JE, Vilboux T. Genes Dis. 2019 Jan 7;6(1):56-67.		2 case reports				Mutations in KIAA1 109 gene : disorder with neurological malformations, seizures, arthrogryposis, craniofacial or cardiac malformations. 2 patients lacking joint contractures and cardiac anomalies.
Gueneau L, Fish RJ, Shamseldin HE, Voisin N, Tran Mau-Them F, Preiksaitiene E, Monroe GR, Lai A, Putoux A, Allias F, Ambusaidi Q, Ambrozaityte L, Cimbalistienė L, Delafontaine J, Guex N, Hashem M, Kurdi W, Jamuar SS, Ying LJ, Bonnard C, Pippucci T, Pradervand S, Roechert B, van Hasselt PM, Wiederkehr M, Wright CF; DDD Study, Xenarios I, van Haaften G, Shaw-Smith C, Schindewolf EM, Neerman-ArbezM, Sanlaville D, Lesca G, Guibaud L, Reversade B, Chelly J, Kučinskas V, Alkuraya FS, Reymond A. Am J Hum Genet. 2018 Jan 4;102(1):116-132.		13 patients cohort				LOF mutations or missense in KIAA1109, Alkuraya Kucinskas syndrome : brain atrophy, clubfoot, arthrogryposis 10/13 and/or talipes valgus/talipes equinovarus/club foot 12/13, cardiac 6/13 (all types) and ophtalmologic anomalies. Overlaps with Aase-Smith syndrome 1 (arthrogryposis and cardiac malformations as well). some letal cases.
Stevenson RE, Vincent V, Spellicy CJ, Friez MJ, Chaubey A. Am J Med Genet A. 2018 Sep;176(9):1968-1971.		cohort of fetuses				Heterozygous SOX10 variants = Waardenburg syndrome. Here homozygous mutations with severe phenotype of fetal akinesia with arthrogryposis, abnormla pigmentation, canthi dystopia. One patient had a cleft, 1 had cardiac malformation (Fallot tetralogy).
Quélin C, Loget P, Rozel C, D'Hervé D, Fradin M, Demurger F, Odent S, Pasquier L, Cavé H, Marcorelles P. Eur J Med Genet. 2017 Jul;60(7):395-398.		case report				Severe form of Costello syndrome with arthrogryposis without cardiac anormalies. In the litterature 2 patients with CMH or septal hypertrophy and PFO asscoaited to AMC AMC. Burkitt-Wright 2012
Verloes A, Di Donato N, Masliah-Planchon J, Jongmans M, Abdul-Raman OA, Albrecht B, Allanson J, Brunner H, Bertola D, Chassaing N, David A, Devriendt K, Eftekhari P, Drouin-Garraud V, Faravelli F, Faivre L, Giuliano F, Guion Almeida L, Juncos J, Kempers M, Eker HK, Lacombe D, Lin A, Mancini G, Melis D, Lourenço CM, Siu VM, Morin G,		cohort				BWS : possible congenital arthrorgyposis and heart defects

Nezarati M, Nowaczyk MJ, Ramer JC, Osimani S, Philip N, Pierpont ME, Procaccio V, Roseli ZS, Rossi M, Rusu C, Sznajer Y, Templin L, Uliana V, Klaus M, Van Bon B, Van Ravenswaaij C, Wainer B, Fry AE, Rump A, Hoischen A, Drunat S, Rivière JB, Dobyns WB, Pilz DT. Eur J Hum Genet. 2015 Mar;23(3):292-301.						
Burkitt-Wright EM, Bradley L, Shorto J, McConnell VP, Gannon C, Firth HV, Park SM, D'Amore A, Munyard PF, Turnpenny PD, Charlton A, Wilson M, Kerr B. Am J Med Genet A. 2012 May;158A(5):1102-10.		cohort				2 patients with hypertrophic cardiomyopathy or septal hypertrophy and PFO, associated to AMC.

Ophtalmological anomalies						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Gueneau L, Fish RJ, Shamseldin HE, Voisin N, Tran Mau-Them F, Preiksaitiene E, Monroe GR, Lai A, Putoux A, Allias F, Ambusaidi Q, Ambrozaityte L, Cimbalistienė L, Delafontaine J, Guex N, Hashem M, Kurdi W, Jamuar SS, Ying LJ, Bonnard C, Pippucci T, Pradervand S, Roechert B, van Hasselt PM, Wiederkehr M, Wright CF; DDD Study, Xenarios I, van Haaften G, Shaw-Smith C, Schindewolf EM, Neerman-ArbezM, Sanlaville D, Lesca G, Guibaud L, Reversade B, Chelly J, Kučinskas V, Alkuraya FS, Reymond A. Am J Hum Genet. 2018 Jan 4;102(1):116-132.		small cohort				mut LOF, missense in KIAA1109, Alkuraya Kucinskas syndrome : brain atrophy, clubfoot, arthrogryposis 10/13 and/or talipes valgus/talipes equinovarus/club foot 12/13, cardiac 6/13 (all types) and ophtalmologic anomalies 8/10 (congenital cataracts, microphtalmia, blepharophimosis, oculomotor apraxia, hypermetropia, strabismus, astigmatism). some letal cases.
Zin AA, Tsui I, Rossetto J, Vasconcelos Z, Adachi K, Valderramos S, Halai UA, Pone MVDS, Pone SM, Silveira Filho JCB, Aibe MS, da Costa ACC, Zin OA, Belfort R Jr, Brasil P, Nielsen-Saines K, Moreira MEL. JAMA Pediatr. 2017 Sep 1;171(9):847-854.		cohort				cohort of infants whose mothers had PCR-confirmed Zika virus infection during pregnancy. N=112, 21.4% (n=24) eye abnormalities (optic nerve and retinal abnormalities), association with arthrogryposis (OR, 29.0), 6.3% (/112) and 29,9% (/24) had arthrogryposis. Type of arthrogryposis not specified.
Meneses JDA, Ishigami AC, de Mello LM, de Albuquerque LL, de Brito CAA, Cordeiro MT, Pena LJ. Clin Infect Dis. 2017 May 15;64(10):1302-1308.		cohort				87 infants with laboratory-confirmed congenital Zika syndrome. 20,7% (n=18) had arthrogryposis. The presence of ophtalmologic abnormalities were significantly associated with a smaller head circumference.

Khan AO, Shaheen R, Alkuraya FS. J AAPOS. 2014 Aug;18(4):362-7. Erratum in: J AAPOS. 2014 Oct;18(5):517.		family report				3/4 siblings with ECEL1-related DA : bilateral ptosis with bilateral congenital fibrosis of the extraocular muscles, right ptosis with ipsilateral Y exotropia and right ptosis with ipsilateral Duane retraction syndrome. 26 patients reported in the litterature, all have arthrogryposis, 19 had ptosis, 4 complex strabisumus, 1 both. Form of Congenital cranial dysinnervation disorder (CCDD)
Verloes A, Di Donato N, Masliah-Planchon J, Jongmans M, Abdul-Raman OA, Albrecht B, Allanson J, Brunner H, Bertola D, Chassaing N, David A, Devriendt K, Eftekhari P, Drouin-Garraud V, Faravelli F, Faivre L, Giuliano F, Guion Almeida L, Juncos J, Kempers M, Eker HK, Lacombe D, Lin A, Mancini G, Melis D, Lourenço CM, Siu VM, Morin G, Nezarati M, Nowaczyk MJ, Ramer JC, Osimani S, Philip N, Pierpont ME, Procaccio V, Roseli ZS, Rossi M, Rusu C, Sznajer Y, Templin L, Uliana V, Klaus M, Van Bon B, Van Ravenswaaij C, Wainer B, Fry AE, Rump A, Hoischen A, Drunat S, Rivière JB, Dobyns WB, Pilz DT. Eur J Hum Genet. 2015 Mar;23(3):292-301.		cohort				Baraitser-Winter syndrome : 2 patients had congenital arthrogryposis. Often iris or retenial coloboma. Heart defects are seen in some cases.
Rajab A, Hoffmann K, Ganesh A, Sethu AU, Mundlos S. Am J Med Genet A. 2005 Apr 15;134A(2):151-7		small cohort				Escobar + ophtalmologic anomalies but phenotype of ECEL1??
Brooks JG Jr, Coster DJ. Aust N Z J Ophthalmol. 1994 May;22(2):127-32		cases report				All ophtalmologic anomalies that can be seen in AMC

Neural tube defects						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Naja AS, El Khatib H, Baydoun A, Nasser Eddine M. Am J Case Rep. 2019 May 20;20:719-722.		case report				one case of 28do boy with lower limb arthrogryposis (fully extended at the knee joint, flexed 45 degrees at the hip joint with extensive contraction and limited passive range of motion due to joint contractures and hyper spasticity, bilateral club feet) , with myelomeningocele and Chiari II malformation all diagnosed prenatally

Weidman EK, Morgenstern PF, Phillips CD, Greenfield JP, Schwartz TH, Heier LA. Int J Pediatr Otorhinolaryngol. 2019 Feb;117:26-29.		case report				Case of Beals syndrome with temporal encephalocele on imaging, middle and inner ear dysplasia unilateral with hearing loss. Encephalocele never reported before. Additional and independant finding??
Kowalczyk B, Feluś J. Arch Med Sci. 2016 Feb 1;12(1):10-24.		review				Abnormal neural tube development, e.g. in meningomyelocele or in sacral agenesia, may result in secondary limitation of active fetal movements and congenital multiple joint contractures; their severity depend on the level of injury of the neural tube.
van Bosse HJ. Foot Ankle Clin. 2015 Dec;20(4):619-44.		review				Myelomeningocele : especially foot deformities of various types according to the level of lesion and can be either congenital or developmental
Chen CP, Chang TY, Lin HH, Chern SR, Wang W. Taiwan J Obstet Gynecol. 2008 Mar;47(1):93-4.		case report				1 fetal case of trisomy 18 with neural tube defect (anencephaly) and arthrogryposis.
Pungavkar SA, Sainani NI, Karnik AS, Mohanty PH, Lawande MA, Patkar DP, Sinha S. Korean J Radiol. 2007 Jul-Aug;8(4):351-5.		case report				A cause with iniencephaly and arthrogryposis in the fetal period, early death after preterm delivery (gemellar pregnancy).
Luedemann WO, Tatagiba MS, Hussein S, Samii M. J Neurosurg. 2000 Jul;93(1 Suppl):130-2. Review		case report				Case report of a woman with congenital arthrogryposis, atlantoaxial subluxation and dysraphic abnormalities. Pas accès.
Podder S, Shepherd RC, Shillito P, Tolmie JL. Clin Dysmorphol. 1995 Jan;4(1):70-4.		case report				no access
Westcott MA, Dynes MC, Remer EM, Donaldson JS, Dias LS. Radiographics. 1992 Nov;12(6):1155-73. Review.		review				myelomeningocele : contractures of lower limbs that can be congenital.

Tethered spinal cord						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
McGirt MJ, Mehta V, Garces-Ambrossi G, Gottfried O, Solakoglu C, Gokaslan ZL, Samdani A, Jallo GI. J Neurosurg Pediatr. 2009 Sep;4(3):270-4.		observational study				Patients with TCS and scoliosis that underwent untethering. Try to determine factor of progression after surgery. 1 case of TCS was due to arthrogryposis but no detail : congenital??

Muscle MRI

Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Carrera-García L, Natera-de Benito D, Dieterich K, de la Banda MGG, Felter A, Inarejos E, Codina A, Jou C, Roldan M, Palau F, Hoenicka J, Pijuan J, Ortez C, Expósito-Escudero J, Durand C, Nugues F, Jimenez-Mallebrera C, Colomer J, Carlier RY, Lochmüller H, Quijano-Roy S, Nascimento A. CHRNG-related nonlethal multiple pterygium syndrome: Muscle imaging pattern and clinical, histopathological, and molecular genetic findings. Am J Med Genet A. 2019 Jun;179(6):915-926. doi: 10.1002/ajmg.a.61122. Epub 2019 Mar 14. PMID: 30868735.	Phénotypage	Case series				marked muscle bulk reduction is the predominant finding, mostly affecting the spinal erector muscles and gluteus maximus. Fatty infiltration was only observed in deep paravertebral muscles and distal lower limbs
Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. A novel neuromuscular form of glycogen storage disease type IV with arthrogryposis, spinal stiffness and rare polyglucosan bodies in muscle. Neuromuscul Disord. 2016 Oct;26(10):681-687. doi: 10.1016/j.nmd.2016.07.005. Epub 2016 Jul 25. PMID: 27546458.	Phénotypage	Case report				fibroadipose muscle replacement but sparing ofthe sartorius, gracilis, adductor longus and vastus intermedialis muscles.
Jarraya M, Quijano-Roy S, Monnier N, Béhin A, Avila-Smirnov D, Romero NB, Allamand V, Richard P, Barois A, May A, Estournet B, Mercuri E, Carlier PG, Carlier RY. Whole-Body muscle MRI in a series of patients with congenital myopathy related to TPM2 gene mutations. Neuromuscul Disord. 2012 Oct 1;22 Suppl 2:S137-47. doi: 10.1016/j.nmd.2012.06.347. PMID: 22980765.	Phénotypage	Case series				predominant involvement of masticatory and distal leg muscles with the other regions rel-atively spared
Mercuri E, Manzur A, Main M, Alsopp J, Muntoni F. Is there post-natal muscle growth in amyoplasia? A sequential MRI study. Neuromuscul Disord. 2009 Jun;19(6):444-5. doi: 10.1016/j.nmd.2009.03.006. Epub 2009 May 27. PMID: 19477646.	Phénotypage					diffuse involvement of both upper and lower limbs with relativepreservation of the trunk muscles; elative sparing of thebiceps femoris, sartorius, gracilis and, on the right thigh, partly alsoof the vasti; overall increased muscle bulk as a result not onlyof hypertrophy of the muscles which were relatively spared in theprevious scans, but also due to the appearance of muscle ofapparently normal signal intensity in previously severely affectedmuscles

Philpot J, Counsell S, Bydder G, Sewry CA, Dubowitz V, Muntoni F. Neonatal arthrogryposis and absent limb muscles: a muscle developmental gene defect? Neuromuscul Disord. 2001 Jul;11(5):489-93. doi: 10.1016/s0960-8966(00)00221-2. PMID: 11404123.	Phénotypage	case report				virtual absence of muscles in the limbs with sparing of the axial muscle

AMC et Médecine Physique et réadaptation

Braces						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Yingsakmongkol W, Kumar SJ. Scoliosis in arthrogryposis multiplex congenita: results after nonsurgical and surgical treatment. J Pediatr Orthop. 2000 Oct;20(5):656–61.	Rééducation	Case series				Scoliosis: bracing in patients who are ambulators and have a curve of <30 degrees
Herron LD, Westin GW, Dawson EG. Scoliosis in arthrogryposis multiplex congenita. J Bone Joint Surg Am. 1978 Apr;60(3):293–9.	Rééducation	Case series				Scoliosis: treatment by corrective casts or a Milwaukee brace was ineffective
Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 1985 Sep;10(7):609–13.	Rééducation	Case series	birth - 15y			Spinal problems can occur in arthrogryposis patients, are poorly controlled nonoperatively
Thomas B, Schopler S, Wood W, Oppenheim WL. The knee in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):87–92.	Rééducation	Case series				Knee: Physiotherapy alone rarely resulted in clinically significant improvement unless accompanied by prolonged casting and bracing
Södergård J, Ryöppy S. The knee in arthrogryposis multiplex congenita. J Pediatr Orthop. 1990 Apr;10(2):177–82.	Rééducation	Case series	1 - 36y			Knee : primary treatment nonoperative in most cases. In spite of the initial promising results, significant number of patients underwent operation for residual symptoms
Komolkin I, Ulrich EV, Agranovich OE, van Bosse HJP. Treatment of Scoliosis Associated With Arthrogryposis Multiplex Congenita. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S24–6.	Rééducation	recommandation de bonne pratqiue ; avis d'expert				cf. recommandations de bonnes pratiques
van Bosse HJP, Marangoz S, Lehman WB, Sala DA. Correction of arthrogrypotic clubfoot with a modified Ponseti technique. Clin Orthop Relat Res. 2009 May;467(5):1283–93.	Rééducation	Case series				Arthrogrypotic club foot: ankle foot orthoses maintained correction

Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.	Rééducation	Case series				Hip: bracing may contribute to some functional ambulation but high need for surgery

Casting						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Thomas B, Schopler S, Wood W, Oppenheim WL. The knee in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):87–92.	Rééducation	Case series				Knee: Physiotherapy alone rarely resulted in clinically significant improvement unless accompanied by prolonged casting and bracing
Bernstein RM. Arthrogryposis and amyoplasia. J Am Acad Orthop Surg. 2002 Dec;10(6):417–24.	Rééducation	Case series				Casting as first tier treatment. High recurrence risk and surgical interventions often necessary.
Smith DW, Drennan JC. Arthrogryposis wrist deformities: results of infantile serial casting. J Pediatr Orthop. 2002 Feb;22(1):44–7.	Rééducation	Case series				Authors recommend early casting of infant wrist deformities for both forms of arthrogryposis, Amyoplasia and DA. If the wrist deformity recurs, repeat serial casting is unlikely to improve wrist extension.
Pontén E. Management of the knees in arthrogryposis. J Child Orthop. 2015 Dec;9(6):465–72.	Rééducation	Case series				Knee: just after birth, traction and mobilization followed by serial casting could often greatly improve the range of motion.
van Bosse HJP. Orthopaedic care of the child with arthrogryposis: a 2020 overview. Curr Opin Pediatr. 2020 Feb;32(1):76–85.	Rééducation	Case series				Avis d'expert

Clinical care						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Binkiewicz-Glińska A, Wierzba J, Szurowska E, Ruckeman-Dziurdzińska K, Bakuła S, Sokołów M, et al. Arthrogryposis multiplex congenital - multidisciplinary care - including own experience. Dev Period Med. 2016;20(3):191–6.	Rééducation	Avis d'expert				Early rehabilitation treatment paramount in a multidisciplinary setting at reference hospitals. when orthopedic treatment is required, it should always be preceded and followed by rehabilitation.
Dahan-Oliel N, Bedard T, Darsaklis VB, Hall JG, van Bosse HJP, Hamdy RC. Development of a research platform for children with arthrogryposis multiplex congenita: study protocol for a pilot registry. BMJ Open. 2018 30;8(6):e021377.	Rééducation	Case series				North American research plateform for children with AMC to establish large scale studies in patients with AMC
Dieterich K, Kimber E, Hall JG. Central nervous system involvement in arthrogryposis multiplex congenita: Overview of causes, diagnosis, and care. Am J Med Genet C Semin Med Genet. 2019;181(3):345–53.	Rééducation	Avis d'expert				Importance of the diagnosis in planning adequate treatment, in genetic counselling. A multidiciplinary team is needed both in investigation and treatment.
Valdés-Flores M, Casas-Avila L, Hernández-Zamora E, Kofman S, Hidalgo-Bravo A. Characterization of a group unrelated patients with arthrogryposis multiplex congenita. J Pediatr (Rio J). 2016 Feb;92(1):58–64.	Rééducation	Case series				important to establish patient-specific physical therapy and rehabilitation programs. A multidisciplinary approach is necessary, with medical, surgical, rehabilitation, social and psychological care, including genetic counseling.
Villard L, Nordmark-Andersson E, Crowley B, Straub V, Bertoli M. Multidisciplinary Clinics. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S29–30.	Rééducation	Avis d'expert				Multidisciplinary clinics are the ideal setting to provide coordinated and comprehensive care to patients with special needs
Williams MS, Elliott CG, Bamshad MJ. Pulmonary disease is a component of distal arthrogryposis type 5. Am J Med Genet A. 2007 Apr 1;143A(7):752–6.	Rééducation	Family report				individuals diagnosed with DA5 (PIEZO2) should be evaluated for chest disease, alveolar hypoventilation

Gait analysis						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Bartonek Å. The use of orthoses and gait analysis in children with AMC. J Child Orthop. 2015 Dec;9(6):437–47.	Rééducation	Case series				Gait analysis as adjunct to evaluate the efficacy of KAFOs

Bjornson KF, Song K, Zhou C, Coleman K, Myaing M, Robinson SL. Walking stride rate patterns in children and youth. Pediatr Phys Ther. 2011;23(4):354–63.	Rééducation	Case series	2 - 16y			Feasability of gait analysis in a cohort of children with AMC
Eriksson M, Bartonek Å, Pontén E, Gutierrez-Farewik EM. Gait dynamics in the wide spectrum of children with arthrogryposis: a descriptive study. BMC Musculoskelet Disord. 2015 Dec 9;16:384.	Rééducation	Case series				Gait analysis as adjunct to evaluate the efficacy of orthoses.
Eriksson M, Villard L, Bartonek A. Walking, orthoses and physical effort in a Swedish population with arthrogryposis. J Child Orthop. 2014 Aug;8(4):305–12.	Rééducation	Case series				Gait analysis as adjunct to evaluate the efficacy of orthoses.
Rodriguez LM, Bickley C, Russo S, Barnes D, Gagnon M, Hamdy R, et al. Perspectives on gait and motion analysis in the management of youth with arthrogryposis multiplex congenita. Am J Med Genet C Semin Med Genet. 2019;181(3):404–9.	Rééducation	Avis d'expert				Power of quantitative motion analysis for most effective treatment interventions

Orthoses						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Bartonek A, Lidbeck CM, Pettersson R, Weidenhielm EB, Eriksson M, Gutierrez-Farewik E. Influence of heel lifts during standing in children with motor disorders. Gait Posture. 2011 Jul;34(3):426–31.	Rééducation	Case series	AMC, CP			heel wedges had a positive influence on pelvis, hip and knee positions in patients with AMC
Eriksson M, Gutierrez-Farewik EM, Broström E, Bartonek A. Gait in children with arthrogryposis multiplex congenita. J Child Orthop. 2010 Feb;4(1):21–31.	Rééducation	Case series				Description of gait patterns with orthoses
Eriksson M, Jylli L, Villard L, Kroksmark A-K, Bartonek Å. Health-related quality of life and orthosis use in a Swedish population with arthrogryposis. Prosthet Orthot Int. 2018 Aug;42(4):402–9.	Rééducation	Case series				Children with AMC were quite satisfied with their orthoses
Fucs PMMB, Svartman C, de Assumpção RMC, Lima Verde SR. Quadricepsplasty in arthrogryposis (amyoplasia): long-term follow-up. J Pediatr Orthop B. 2005 May;14(3):219–24.	Rééducation	Case series				Allongement Z quadriceps + capsulotomie antérieure + platre 3 semaines à 90° puis attelles alternées

Outcome + medical - orthopaedic

Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316–9.	Rééducation	Case series	AMC			Most children improved with physiotherapy and well-fitted orthoses
Carlson WO, Speck GJ, Vicari V, Wenger DR. Arthrogryposis multiplex congenita. A long-term follow-up study. Clin Orthop Relat Res. 1985 Apr;(194):115–23.	Rééducation	Case series				In addition to appropriate surgical correction, good family support, a proper educational environment, and promotion of independence at an early age are required to achieve maximal adult function
Donohoe M, Pruszcynski B, Rogers K, Bowen JR. Predicting Ambulatory Function Based on Infantile Lower Extremity Posture Types in Amyoplasia Arthrogryposis. J Pediatr Orthop. 2019 Aug;39(7):e531–5.	Rééducation	Case series				Amyoplasia can be sorted by infantile position of lower extremities and muscle strength into 5 types to predict ambulatory function
Dubousset J, Guillaumat M. Long-term outcome for patients with arthrogryposis multiplex congenita. J Child Orthop. 2015 Dec;9(6):449–58.	Rééducation	Case series	Amyoplasia, other causes of AMC			priority goes to the upper limbs function, because majority of these patients have a high level of intelligence
Ho CA, Karol LA. The utility of knee releases in arthrogryposis. J Pediatr Orthop. 2008 May;28(3):307–13.	Rééducation	Case series				Decline in ambulatory ability with age
Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.	Rééducation	Case series				definit les ambulatoires comme flessum de hanche <30° et de genou<20°. Importance de la notion de marchant a évaluer à 10 ans. Chirurgies après 10 ans chez le marchant = dos et pieds (triple arthrodèse par exemple) pbve plus gênant que pied convexe pour la marche car moins plantigrade. Difficultés d'extrapoler la position de pied à la possibilité de marche
Nouraei H, Sawatzky B, MacGillivray M, Hall J. Long-term functional and mobility outcomes for individuals with arthrogryposis multiplex congenita. Am J Med Genet A. 2017 May;173(5):1270–8.	Rééducation	Case series				Similar or higher scores for SF36 compared to US general population; Less physically active. At least half experienced pain.
Sawatzky B, Jones T, Miller R, Noureai H. The relationship between joint surgery and quality of life in adults with arthrogryposis: An international study. Am J Med Genet C Semin Med Genet. 2019;181(3):469–73.	Rééducation	Case series				knee and/or shoulder surgeries were more likely to have a negative correlation with Physical Capacity Score; elbow surgery, however, showed a positive correlation, as elbow function may impact independent function

Södergård J, Hakamies-Blomqvist L, Sainio K, Ryöppy S, Vuorinen R. Arthrogryposis multiplex congenita: perinatal and electromyographic findings, disability, and psychosocial outcome. J Pediatr Orthop B. 1997 Jul;6(3):167–71.	Rééducation	Case series				Patients cope well socially.
Wall LB, Vuillerman C, Miller PE, Bae DS, Goldfarb CA, CoULD Study Group. Patient-reported Outcomes in Arthrogryposis. J Pediatr Orthop. 2020 Feb 7;	Rééducation	Case series				Amyoplasia patients were functionally worse than distal arthrogryposis patients

Physiotherapy						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Hamdy RC, van Bosse H, Altiok H, Abu-Dalu K, Kotlarsky P, Fafara A, et al. Treatment and outcomes of arthrogryposis in the lower extremity. Am J Med Genet C Semin Med Genet. 2019;181(3):372–84.	Rééducation	Avis d'expert				
Murray C, Fixsen JA. Management of knee deformity in classical arthrogryposis multiplex congenita (amyoplasia congenita). J Pediatr Orthop B. 1997 Jul;6(3):186–91.	Rééducation	Case series				Knee deformaties: physiotherapy and splintage successful in all except 1 patient in the extended-knee group, but only 7 to 26 knees responded to physiotherapy and splintage alone in the flexed-knee group
Palmer PM, MacEwen GD, Bowen JR, Mathews PA. Passive motion therapy for infants with arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):54–9.	Rééducation	Case series				Daily intensive passive stretching of joints and serial splinting have substantially increased patient function in this population
Samekova H, Bialik V, M Bialik G. Arthrogryposis multiplex congenita - local experience. Ortop Traumatol Rehabil. 2006 Feb 28;8(1):69–73.	Rééducation	Case series				physiotherapy and occupational therapy supported by orthotic equipment as first tier treatment option in patients with AMC

<u>Amyoplasie et Médecine Physique et réadaptation</u>

Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Braces						
Lampasi M, Antonioli D, Donzelli O. Management of knee deformities in children with arthrogryposis. Musculoskelet Surg. 2012 Dec;96(3):161–9.	Rééducation	case series	AMC + Amyoplasia			Reprise de toutes les techniques détaillées et des cas de flexion et hyper extension. Conclusion : prise en charge MPR précoce. Chirurgie pour

						les rétractions non vaincues par traitement orthopédique la 1ère année. PEC globale. En cas de récidive, parfois attendre fin de croissance
Yang SS, Dahan-Oliel N, Montpetit K, Hamdy RC. Ambulation gains after knee surgery in children with arthrogryposis. J Pediatr Orthop. 2010 Dec;30(8):863–9.	Rééducation	case series	Amyoplasia			Knee flexion contracture. Femoral distal extension osteotomy effective. Loss of ROM or recurrence of flexion contracture did not limit ambulatory gains
Clinical care						
Ayadi K, Trigui M, Abid A, Cheniour A, Zribi M, Keskes H. [Arthrogryposis: clinical manifestations and management]. Arch Pediatr. 2015 Aug;22(8):830–9.		case series	AMC + Amyoplasia			Results of surgical treatment of congenital convex pes valgus (10 non-idiopathic feet)
Outcome + medical – orthopaedic						
Sells JM, Jaffe KM, Hall JG. Amyoplasia, the most common type of arthrogryposis: the potential for good outcome. Pediatrics. 1996 Feb;97(2):225–31.		case series	Amyoplasia			Excellent physical, social, and intellectual outcome under rehabilitative and surgical interventions
Physiotherapy						
Lake AL, Oishi SN. Hand therapy following elbow release for passive elbow flexion and long head of the triceps transfer for active elbow flexion in children with amyoplasia. J Hand Ther. 2015 Jun;28(2):222–6; quiz 227.		case series	Amyoplasia			expert opinion on rehabilitation including physiotherapy after elbow relezse and long head triceps transfer
Treatment + medical – orthopaedic						
Kroksmark A-K, Kimber E, Jerre R, Beckung E, Tulinius M. Muscle involvement and motor function in Amyoplasia. Am J Med Genet A. 2006 Aug 15;140(16):1757–67		case series	Amyoplasia			Good correlation between muscle strength and motor function; early stimulation of active movement

AMC et chirurgie

Achilles tendon release						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, Keskes H. Results of surgical treatment of congenital convex pes valgus (ten nonidiopathic feet). Rev Chir Orthop Reparatrice Appar Mot. 2008 Dec;94(8):e28-34. doi: 10.1016/j.rco.2007.12.020. Epub 2008 Jun 24. PMID: 19070711.	Chirurgical	Case series				Results of surgical treatment of congenital convex pes valgus (10 non-idiopathic feet)
Ramanoudjame M, Loriaut P, Seringe R, Glorion C, Wicart P. The surgical treatment of children with congenital convex foot (vertical talus): evaluation of midtarsal surgical release and open reduction. Bone Joint J. 2014 Jun;96-B(6):837-44. doi: 10.1302/0301-	Chirurgical	Case series				concerne le pied convexe. 31 pieds.15% de reprise

620X.96B6.32313. PMID: 24891587.						
Zimbler S, Craig CL. The arthrogrypotic foot plan of management and results of treatment. Foot Ankle. 1983 Jan-Feb;3(4):211-9. doi: 10.1177/107110078300300406. PMID: 6832664.	Chirurgical	Case series				insiste sur difficultés pied et arthrogryposes. Préconise plâtres dès que possible/LPI et talectomie comme salvage procédure

Adducted thumb						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794-798. doi: 10.1177/1753193417712863. Epub 2017 Jun 12. PMID: 28602132.	Chirurgical	Case series				69 pouces 39 patients age moyen 30 mois. Cure de pouce flexus add. Lambeau cutané dorsal d'approfondissement de commissure
Rodríguez RN, Capdevila-Leonori R, Nualart-Hernández L. [Opening of the first web space in patients with multiple congenital arthrogryposis and adducted thumb with a dorsoradial index finger flap]. Acta Ortop Mex. 2014 Jan-Feb;28(1):23-7. PMID: 26031134.	Chirurgical	Case series				technique lambeau dorsal 1ere commissure

Anesthesia						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Oberoi GS, Kaul HL, Gill IS, Batra RK. Anaesthesia in arthrogryposis multiplex congenita: case report. Can J Anaesth. 1987 May;34(3 (Pt 1)):288–	Chirurgical					utilisation curare et kétamines

90.						
Hopkins PM, Ellis FR, Halsall PJ. Hypermetabolism in arthrogryposis multiplex congenita. Anaesthesia. 1991 May;46(5):374–5.	Chirurgical					risk malignant hyperthermia
Szmuk P, Ezri T, Warters DR, Katz J. Anesthetic management of a patient with arthrogryposis multiplex congenita and limited mouth opening. J Clin Anesth. 2001 Feb;13(1):59–60.	Chirurgical					aperture buccale et intubation
Nguyen NH, Morvant EM, Mayhew JF. Anesthetic management for patients with arthrogryposis multiplex congenita and severe micrognathia: case reports. J Clin Anesth. 2000 May;12(3):227–30.	Chirurgical					aperture buccale et intubation
Ion T, Cook-Sather SD, Finkel RS, Cucchiaro G. Fascia iliaca block for an infant with arthrogryposis multiplex congenita undergoing muscle biopsy. Anesth Analg. 2005 Jan;100(1):82–4.	Chirurgical					réalisation de blocs périphériques
Ponde V, Desai AP, Shah D. Comparison of success rate of ultrasound-guided sciatic and femoral nerve block and neurostimulation in children with arthrogryposis multiplex congenita: a randomized clinical trial. Paediatr Anaesth. 2013 Jan;23(1):74–8.	Chirurgical					réalisation de blocs périphériques
Gleich SJ, Tien M, Schroeder DR, Hanson AC, Flick R, Nemergut ME. Anesthetic Outcomes of Children With Arthrogryposis Syndromes: No Evidence of Hyperthermia. Anesth Analg. 2017;124(3):908–14.	Chirurgical					Absence de risque d'hyperthermie maligne
Savenkov AN, Pajardi GE, Agranovich OE, Zabolskiy D, van Bosse HJP. Anaesthesiology for Children With Arthrogryposis. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S27–8.	Chirurgical					condition générales: ouverture bouche + extension cou +micrognathie = intubation/ entretien anesthesique/bloc
Isaacson G, Drum ET. Difficult airway management in children and young adults with arthrogryposis. World J Otorhinolaryngol Head Neck Surg. 2018 Jun;4(2):122–5.	Chirurgical					aperture buccale et intubation

Club feet						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Eidelman M, Katzman A. Treatment of arthrogrypotic foot deformities with the Taylor Spatial Frame. J Pediatr Orthop. 2011 Jun;31(4):429–34.	Chirurgical	Case series				PBVE et arthrogryposes. 7 patients de 4 à 16ans; tsf permet une amélioration de correction, n'est pas le ttt de 1ere intention, ne corrige pas la raideur, ne prévient pas des récidives. 16 semaines de temps de port de fixateur minimum
Iskandar HN, Bishay SNG, Sharaf-El-Deen HA-R, El-Sayed MM. Tarsal decancellation in the residual resistant arthrogrypotic clubfoot. Ann R Coll Surg Engl. 2011 Mar;93(2):139–45.	Chirurgical	Case series				curetage intraspongieux
Spires TD, Gross RH, Low W, Barringer W. Management of the resistant myelodysplastic or arthrogrypotic clubfoot with the Verebelyi-Ogston procedure. J Pediatr Orthop. 1984 Nov;4(6):705–10.	Chirurgical	Case series				description de verebelyi-ogston procedure = bone decancellation for talus et cuboid. Reserve sur l'évolution post chirurgicale
Choi IH, Yang MS, Chung CY, Cho TJ, Sohn YJ. The treatment of recurrent arthrogrypotic club foot in children by the Ilizarov method. A preliminary report. J Bone Joint Surg Br. 2001 Jul;83(5):731–7.	Chirurgical	Case series				
Widmann RF, Do TT, Burke SW. Radical soft-tissue release of the arthrogrypotic clubfoot. J Pediatr Orthop B. 2005 Mar;14(2):111–5.	Chirurgical	Case series				6 patients. Chirurgie avant un an.8 % de récidive des LPI avant un an. indication d'attelles jusqu'à maturité osseuse. Indication d'un release complet et circonférentiel et de ténectomies.
Machida J, Inaba Y, Nakamura N. Management of foot deformity in children. J Orthop Sci. 2017 Mar;22(2):175–83.	Chirurgical	Case series				techniques chirurgicales
van Bosse HJP. Challenging clubfeet: the arthrogrypotic clubfoot and the complex clubfoot. J Child Orthop. 2019 Jun 1;13(3):271–81.	Chirurgical	Case series				ponseti modifie Tenotomies iteratives, platres, ilizarov dernier recours

Hip						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.	Chirurgical					definit les ambulatoires comme flessum de hanche <30° et de genou<20°. Importance de la notion de marchant a évaluer à 10 ans. Chirurgies après 10 ans chez le marchant = dos et pieds (triple arthrodèse par exemple) pbve plus gênant que pied convexe pour la marche car moins plantigrade. Difficultés d'extrapoler la position de pied à la possibilité de marche
St Clair HS, Zimbler S. A plan of management and treatment results in the arthrogrypotic hip. Clin Orthop Relat Res. 1985 Apr;(194):74–80.	Chirurgical					groupe 1a: bilat luxées ms atteints : ne rien faire. 1b : bilat luxées ms ok : OR 1ere année, raccourcissement femoral par approche anterolaterale ; groupe 2 unilat luxée : chir si echec traitement closed reduction ; groupe 3 subluxation : kine et posture, chirurgie selon evolution. groupe 4 contracture : kiné posture.
Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop. 1987 Dec;7(6):681–5.	Chirurgical					préconisation open reduction medial apporach entre 3 et 6 mois avec chirurgie combinée pied cheville genou et plâtre 5 à 6 semaines; en opposition avec Aydin et al, 2016
Yau PWP, Chow W, Li YH, Leong JCY. Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. J Pediatr Orthop. 2002 Jun;22(3):359–63.	Chirurgical					échec des traitements closed, échec des releases antérieurs pour flessum : préférer ostéotomies d'extension. Abord anterolateral, or + raccourcissement femoral + geste osseux bassin + plâtre. Bénéfice à traiter les hanches luxées bilat.
Rocha LEM da, Nishimori FK, Figueiredo DC de, Grimm DH, Cunha LAM da. OPEN REDUCTION OF HIP DISLOCATION IN PATIENTS WITH ARTHROGRYPOSIS MULTIPLEX CONGENITA - AN ANTEROMEDIAL APPROACH. Rev Bras Ortop. 2010 Oct;45(5):403–8.	Chirurgical					préconise approche antérolatérale avec racc fémoral dans les unilatérales. Reprend les ccls et préconisations des autres auteurs.
Wada A, Yamaguchi T, Nakamura T, Yanagida H, Takamura K, Oketani Y, et al. Surgical treatment of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop B. 2012 Sep;21(5):381–5.	Chirurgical					Intérêt de traiter les lch bilatérales. Calendrier : pieds et genoux entre 3 et 10 mois, hanches vers 10/18 mois Smith Petersen, geste fémoral + bassin + open. Echec des ttts closed type pavlik

Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.	Chirurgical					préconisation 1/ pieds 3-4 mois 2/ genoux 3/hanches vers 1 an. Importance de l'évaluation globale
Bradish C. The hip in arthrogryposis. J Child Orthop. 2015 Dec;9(6):459–63.	Chirurgical					reprend les differents types d'atteintes, contractures, luxation uni ou bi et flexion ++. En faveur d'une MAOR uni ou bi type staheli, release pour les flexions > 45° antérieur et kiné pour raideur fixé en abduction
Aydin BK, Yilmaz G, Senaran H, Durgut F. Short-term results of early (before 6 months) open reduction of dislocated hips in arthrogryposis multiplex congenita. J Pediatr Orthop B. 2016 Nov;25(6):509–13.	Chirurgical					pas d'indication à réduction sanglante avant 6 mois AMC 7 patts, faire chirurgie plus tardive. Pas de données sur marche et déambulation des patients opérés
Dalton DM, Magill P, Mulhall KJ. Bilateral total hip replacement in arthrogryposis multiplex congenita. BMJ Case Rep. 2015 Nov 25;2015.	Chirurgical					étude d'un cas d'AMC, LCH bilat, ostéotomie de Chiari vers 27 et 29 ans, chirurgies des membres inf. arthroplastie des 2 hanches à 56 ans, succès sur douleurs et déambulation autonome.
van Bosse HJP, Saldana RE. Reorientational Proximal Femoral Osteotomies for Arthrogrypotic Hip Contractures. J Bone Joint Surg Am. 2017 Jan 4;99(1):55–64.	Chirurgical					seul article sur les ostéotomies de réorientation dans les contractures de hanches excellent

Hip dislocation						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Wada A, Yamaguchi T, Nakamura T, Yanagida H, Takamura K, Oketani Y, et al. Surgical treatment of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop B. 2012 Sep;21(5):381–5.	Chirurgical					Intérêt de traiter les lch bilatérales. Calendrier : pieds et genoux entre 3 et 10 mois, hanches vers 10/18 mois Smith Petersen, geste fémoral + bassin + open. Echec des ttts closed type pavlik
Aydin BK, Yilmaz G, Senaran H, Durgut F. Short-term results of early (before 6 months) open reduction of dislocated hips in arthrogryposis multiplex congenita. J Pediatr Orthop B. 2016 Nov;25(6):509–13.	Chirurgical					pas d'indication à réduction sanglante avant 6 mois AMC 7 patts, faire chirurgie plus tardive. Pas de données sur marche et déambulation des patients opérés

Yau PWP, Chow W, Li YH, Leong JCY. Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. J Pediatr Orthop. 2002 Jun;22(3):359–63.	Chirurgical					échec des traitements closed, échec des releases antérieurs pour flessum : préférer ostéotomies d'extension. Abord anterolateral, or + raccourcissement femoral + geste osseux bassin + plâtre. Bénéfice à traiter les hanches luxées bilat.
Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop. 1987 Dec;7(6):681–5.	Chirurgical					préconisation open reduction medial apporach entre 3 et 6 mois avec chirurgie combinée pied cheville genou et plâtre 5 à 6 semaines; en opposition avec Aydin et al, 2016

Knee						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.	Chirurgical	Case series				facteurs péjoratifs pour capacité de marche : flessum hanche > 30 °, flessum genou >30°
Saleh M, Gibson MF, Sharrard WJ. Femoral shortening in correction of congenital knee flexion deformity with popliteal webbing. J Pediatr Orthop. 1989 Oct;9(5):609–11.	Chirurgical	Case series				Raccourcissement fémoral + arthrolyse posterieure. Correction persistance à 3 ans et 2 mois. Recul de 2 ans. Marchante.
Södergård J, Ryöppy S. The knee in arthrogryposis multiplex congenita. J Pediatr Orthop. 1990 Apr;10(2):177–82.	Chirurgical	Case series				Généralités sur une cohorte. Descriptions des techniques.
Murray C, Fixsen JA. Management of knee deformity in classical arthrogryposis multiplex congenita (amyoplasia congenita). J Pediatr Orthop B. 1997 Jul;6(3):186–91.	Chirurgical	Case series				Extension moins grave et moins de conséquences que flessum, moins difficile à traiter. Ostéotomie < 3 ans = risque de récidive.
Brunner R, Hefti F, Tgetgel JD. Arthrogrypotic joint contracture at the knee and the foot: correction with a circular frame. J Pediatr Orthop B. 1997 Jul;6(3):192–7.	Chirurgical	Case series				Age moyen 11,9 ans. 13 genoux preop flessum 38,9°, post op 6,5 plus grand recul 17,9 ans. Plaide pour fixateur ext, insuffisance des traitements orthopédiques
Mooney JF, Koman LA. Knee flexion contractures: soft tissue correction with monolateral external fixation. J South Orthop Assoc. 2001;10(1):32–6.	Chirurgical	Case series				Fixateur externe latéral monotube. Efficacité. Récidive. Série de 7 patients pas que arthrogryposes
Fucs PMMB, Svartman C, de Assumpção RMC, Lima Verde SR. Quadricepsplasty in arthrogryposis (amyoplasia): long-term follow-up. J Pediatr Orthop B. 2005 May;14(3):219–24.	Chirurgical	Case series				Allongement Z quadriceps + capsulotomie antérieure + platre 3 semaines à 90° puis attelles alternées

van Bosse HJP, Feldman DS, Anavian J, Sala DA. Treatment of knee flexion contractures in patients with arthrogryposis. J Pediatr Orthop. 2007 Dec;27(8):930–7.	Chirurgical	Case series				Ilizarov + ; 10 genoux de 72,5 ° à 20,5° au dernier suivi. Age moyen 7,3 ans.
Ho CA, Karol LA. The utility of knee releases in arthrogryposis. J Pediatr Orthop. 2008 May;28(3):307–13.	Chirurgical	Case series				Recidive inevitable, moindres déconvenues dans les ttt des flessums. Conclusion : bien avertir les parents des évolutions prévisibles, s'occuper des membres supérieurs pour être opérationnels avec un bon fauteuil
Klatt J, Stevens PM. Guided growth for fixed knee flexion deformity. J Pediatr Orthop. 2008 Sep;28(6):626–31.	Chirurgical	Case series				Cohorte multiples étiologies. Intérêt du 8 plate.
Borowski A, Grissom L, Littleton AG, Donohoe M, King M, Kumar SJ. Diagnostic imaging of the knee in children with arthrogryposis and knee extension or hyperextension contracture. J Pediatr Orthop. 2008 Jun;28(4):466–70.	Chirurgical	Case series				Localisation supéro laterale de rotule en echo et irm quand pas de palpation clinique possible. Bénéfice d'une plastie quadriceps et repositionnement de rotule
Palocaren T, Thabet AM, Rogers K, Holmes L, Donohoe M, King MM, et al. Anterior distal femoral stapling for correcting knee flexion contracture in children with arthrogryposis--preliminary results. J Pediatr Orthop. 2010 Mar;30(2):169–73.	Chirurgical	Case series				8 plates 10 patients age moyen 7 ans diagnostic arthrogrypose
Sud A, Kumar N, Mehtani A. Femoral shortening in the congenital dislocation of the knee joint: results of mid-term follow-up. J Pediatr Orthop B. 2013 Sep;22(5):440–4.	Chirurgical	Case series				Femoral shortening. Mauvais résultats chez patients AMC
Gavriliu S, Georgescu I, Ulici A, Ghita R, Japie EM, Pandea N, et al. Herbert capsuloplasty and Burnei tenomyoplasty for the correction of genu flexum in cerebral palsy, arthrogryposis and posttraumatic. Chirurgia (Bucur). 2013 Dec;108(6):866–73.	Chirurgical	Case series				Patient origines multiples : AMC, PC, post trauma. Description des techniques : capsulotomie postérieure, allongement des IJ, ostéotomie de recurvatum
Lampasi M, Antonioli D, Donzelli O. Management of knee deformities in children with arthrogryposis. Musculoskelet Surg. 2012 Dec;96(3):161–9.	Chirurgical	Case series				Reprise de toutes les techniques détaillées et des cas de flexion et hyper extension. Conclusion : prise en charge MPR précoce. Chirurgie pour les rétractions non vaincues par traitement orthopédique la 1ère année. PEC globale. En cas de récidive, parfois attendre fin de croissance
Eamsobhana P, Kaewpornsawan K, Vanitcharoenkul E. Walking ability in patients with arthrogryposis multiplex congenita. Indian J Orthop. 2014 Jul;48(4):421–5.	Chirurgical	Case series				facteurs péjoratifs pour capacité de marche : flessum hanche > 30 °, flessum genou >30°

Oleas-Santillán G, Bowen JR. Tension band plate-guided growth of knee-flexion deformity in arthrogryposis multiplex congenita in which metaphyseal funnelization induced screw encroachment upon the neurovascular bundle. J Pediatr Orthop B. 2020 Jan;29(1):62–4.	Chirurgical	Case report				Description d'une complication 8 plate et bris de vis avec menace axe VN poplité

Muscle transfer						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Lake AL, Oishi SN. Hand therapy following elbow release for passive elbow flexion and long head of the triceps transfer for active elbow flexion in children with amyoplasia. J Hand Ther. 2015 Jun;28(2):222–6; quiz 227.	Chirurgical	Case series				physiotherapie post release triceps et transfert triceps. Rappel des indications et timing article non chirurgical
Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794-798. doi: 10.1177/1753193417712863. Epub 2017 Jun 12. PMID: 28602132.	Chirurgical	Case series				69 pouces 39 patients age moyen 30 mois. Cure de pouce flexus add. Lambeau cutané dorsal d'approfondissement de commissure
Hagemann C, Stücker R, Breyer S, Kunkel POS. Nerve transfer from the median to musculocutaneous nerve to induce active elbow flexion in selected cases of arthrogryposis multiplex congenita. Microsurgery. 2019 Nov;39(8):710–4.	Chirurgical	Case series				Transfert nerveux pour réanimer la flexion du coude. Pas d'emg ni irm musculaire preopératoire. Simple etude echographique confirmant la présence d'un biceps brachial. Transfert nerveux du médian ou ulnaire sur musculo cutané. 5 patients opérés entre 2 et 5 ans. Kiné et rééducation post op.

Pes convex						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, et al. Results of surgical treatment of congenital convex pes valgus (ten nonidiopathic feet). Rev Chir Orthop Reparatrice Appar Mot. 2008 Dec;94(8):e28-34.	Chirurgical	Case series				Results of surgical treatment of congenital convex pes valgus (10 non-idiopathic feet) techniques chirurgicales bien decrites. Ttt difficile du pied convexe arthrogryposes.preferer une approche dorsale. Liberation releveurs, allongement fibulaires, release calcacub, brochage talo m1 + calcacub +/- talo calca, liberation anterieure ++ age 15 mois
Guidera KJ, Drennan JC. Foot and ankle deformities in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):93–8.	Chirurgical					pbve doivent être traités par talectomie ; pied convexe= mauvais résultats

Ponseti						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Funk JF, Lebek S, Seidl T, Placzek R. [Comparison of treatment results of idiopathic and non-idiopathic congenital clubfoot : prospective evaluation of the Ponseti therapy]. Orthopade. 2012 Dec;41(12):977–83.	Chirurgical	Case series				interet du ponseti sur pbve non idiop. 17% de reprise chez non idiop à 3 ans contre 9% idiop. Pas de lpi, transfert +, achille itératif, fascia et liberation dorsomediale
van Bosse HJP. Challenging clubfeet: the arthrogrypotic clubfoot and the complex clubfoot. J Child Orthop. 2019 Jun 1;13(3):271–81.	Chirurgical	Avis d'expert				high recurrence rate suggests the difficulty in maintaining the deformity after correction. Emphasis on minimally invasive methods. Expert opinion.

Scoliosis						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Herron LD, Westin GW, Dawson EG. Scoliosis in arthrogryposis multiplex congenita. J Bone Joint Surg Am. 1978 Apr;60(3):293–9.	Chirurgical	Case series				18patients scolioses amc. Importance du retablissement de l'obliquité pelvienne avant traitement de la colonne = chir hanches. Milwaukee insuffisant. perte 2dr de correction frequente après arthrodese. Courbures rigides et fixes a age precoce. Proposition arthrodese post precoce pour courbure souple. Approche ant et post pour raides.prise sacrée +/- halo preop ? Malformations associées vertébrales
Yingsakmongkol W, Kumar SJ. Scoliosis in arthrogryposis multiplex congenita: results after nonsurgical and surgical treatment. J Pediatr Orthop. 2000 Oct;20(5):656–61.	Chirurgical	Case series				46 patients. Groupe non chir < 30°/ groupe chir moyenne cobb 78.5°corset = patients ambulatoires <30°. Resultats satisfaisants de chir si abord ant et post combines. Pas de diminution de deambulation post op
Fletcher ND, Rathjen KE, Bush P, Ezaki M. Asymmetrical arthrogryposis of the upper extremity associated with congenital spine anomalies. J Pediatr Orthop. 2010 Dec;30(8):936–41.	Chirurgical	Case series				analyse groupe scoliose et asymétire MS chez amc.6 patients. Atteintes ms multiples +chirurgies. Ccl : = interet d'évalution de scoliose cervicale/ rachis cervical et imagerie

Skin web						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794-798. doi: 10.1177/1753193417712863. Epub 2017 Jun 12. PMID: 28602132.	Chirurgical	Case series				69 pouces 39 patients age moyen 30 mois. Cure de pouce flexus add. Lambeau cutané dorsal d'approfondissement de commissure. Release 1ere comm (1er interosseux add du pouce). Chondrodese de la mp. Allongement lfp. +/- greffe de peau totale

Spinal arthrodesis						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations

Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 1985 Sep;10(7):609–13.	Chirurgical	Case series				A Review of 16 Patients attention à la lordose thoracique et desequilibre sagittal necessitant un ttt precoce. Raideur ++. Difficultes du ttt orthopedique article toutefois ancien

AMC et douleur

Douleur						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Altiok H, Flanagan A, Krzak JJ, Hassani S. Quality of life, satisfaction with life, and functional mobility of young adults with arthrogryposis after leaving pediatric care. Am J Med Genet C Semin Med Genet. 2019;181(3):461–8.	algologie	Case series				Average pain intensity was mild at 2.6 out of 10, with pain frequently reported in the legs and feet.
Cirillo A, Collins J, Sawatzky B, Hamdy R, Dahan-Oliel N. Pain among children and adults living with arthrogryposis multiplex congenita: A scoping review. Am J Med Genet C Semin Med Genet. 2019;181(3):436–53.	algologie	Scoping review				Pain appears to be more commonly experienced in adults with AMC compared with children with AMC, with individuals having undergone multiple corrective procedures self-reporting pain more often. In adult populations, musculoskeletal chronic pain is a significant problem, resulting in restrictions in activities of daily living, mobility, and participation. Pain in adults with AMC should be acknowledged to offer proper client-centered interventions throughout the lifespan.
Dai S, Dieterich K, Jaeger M, Wuyam B, Jouk P-S, Pérennou D. Disability in adults with arthrogryposis is severe, partly invisible, and varies by genotype. Neurology. 2018 01;90(18):e1596–604.	algologie	Case series				90% of patients complained about pain, especially in lower back and extremities.
Jones T, Miller R, Street JT, Sawatzky B. Validation of the Oswestry Disability Index for pain and disability in arthrogryposis multiplex congenita. Ann Phys Rehabil Med. 2019 Mar;62(2):92–7.	algologie	Case series				Oswestry Disability Index: valid outcome tool for low-back and lower-extremity pain-related disability for patients with AMC
Kimber E, Tajsharghi H, Kroksmark A-K, Oldfors A, Tulinius M. Distal arthrogryposis: clinical and genetic findings. Acta Paediatr. 2012 Aug;101(8):877–87.	algologie	Case series				Fourteen patients reported pain related to muscle and joint affection.

Nicomedez FPI, Li YH, Leong JCY. Tibiocalcaneal fusion after talectomy in arthrogrypotic patients. J Pediatr Orthop. 2003 Oct;23(5):654–7.	algologie	Case series				Tibiocalcaneal fusion may improve the function and the pain symptoms of arthrogrypotic patients, but it can lead to early degenerative arthritis of the adjacent joints.
Wall LB, Vuillerman C, Miller PE, Bae DS, Goldfarb CA, CoULD Study Group. Patient-reported Outcomes in Arthrogryposis. J Pediatr Orthop. 2020 Feb 7;	algologie	Case series				PROMIS pain was in the normal range for both amyopasia and distal arthrogryposis. Median PODCI pain 85 to 88 for all patients with no statistical difference between groups.

AMC et anesthésie

Anesthésie						
Référence	Objectif	Méthodologie, niveau de preuve	Population	Intervention	Critères de jugement	Résultats et significations
Baines DB, Douglas ID, Overton JH. Anaesthesia for patients with arthrogryposis multiplex congenita: what is the risk of malignant hyperthermia? Anaesth Intensive Care. 1986 Nov;14(4):370–2.	Anesthésie	Case series				over a 32-year period revealed no episode of malignant hyperthermia occurring in patients with arthrogryposis multiplex congenita
Hopkins PM, Ellis FR, Halsall PJ. Hypermetabolism in arthrogryposis multiplex congenita. Anaesthesia. 1991 May;46(5):374–5.	Anesthésie	case reports				Two patients with AMC developed hypermetabolic reactions during anaesthesia. Reaction distinct from malignant hyperthermia and independent of the anaesthetic agents used.
Ma L, Yu X. Arthrogryposis multiplex congenita: classification, diagnosis, perioperative care, and anesthesia. Front Med. 2017 Mar;11(1):48–52.	Anesthésie	scoping review				discussion on treatment and perioperative management of patients with AMC undergoing surgery
Jung JW, Heo BY, Oh EJ, Chung YH. Anesthesia in patients with arthrogryposis multiplex congenita: a report of 10 patients. Korean J Anesthesiol. 2014 Dec;67(Suppl):S89-90.	Anesthésie					
Gleich SJ, Tien M, Schroeder DR, Hanson AC, Flick R, Nemergut ME. Anesthetic Outcomes of Children With Arthrogryposis Syndromes: No Evidence of Hyperthermia. Anesth Analg. 2017;124(3):908–14.	Anesthésie	Cas - témoins				challenging peripheral IV placement, difficult airway management and intraoperative hemodynamic instability. Authors did not find evidence of increased odds of intraoperative hyperthermia or hypermetabolic responses.

Savenkov AN, Pajardi GE, Agranovich OE, Zabolskiy D, van Bosse HJP. Anaesthesiology for Children With Arthrogryposis. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S27–8.	Anesthésie	Avis d'expert				Intubation challenging in patients with AMC. Peripheral blocks as adjunct to post-operative pain management
Isaacson G, Drum ET. Difficult airway management in children and young adults with arthrogryposis. World J Otorhinolaryngol Head Neck Surg. 2018 Jun;4(2):122–5.	Anesthésie	review				75% of patients with AMC without difficult airway access. Careful advances planning of surgeries.

Annexe

Annexe 1. Liste des participants

Ce travail a été coordonné par le Dr DIETERICH Klaus, Centre de référence Anomalies du développement et syndromes malformatifs Sud-Est, site constitutif Grenoble, et Centre de compétence Maladies neuromusculaires rares PACARARE, site Grenoble (CHU Grenoble Alpes).

Ont participé à l'élaboration du PNDS :

Groupe de rédacteurs

- Dr Klaus Dieterich, médecin généticien, Grenoble
- Dr Véronique Bourg, médecin MPR, Grenoble
- Dr Emeline Bourgeois, chirurgien orthopédique, Grenoble
- Pr Jacques Griffet, chirurgien orthopédique, Grenoble
- Mme Gipsy Billy, conseillère en génétique, Grenoble
- Mme Marjolaine Gauthier, infirmière de recherche clinique, Grenoble
- Mme Claire Huzar, kinésithérapeute, Grenoble
- Mme Charlotte Marion, psychologue, Grenoble
- Mme Véronique Thellier, ergothérapeute, Grenoble

Groupe de travail multidisciplinaire (ordre alphabétique)

- Dr Hassan Al Khoury, orthopédiste, Saint Etienne
- Dr Beatrice Bayle, médecin de MPR, Saint Etienne, filière FILNEMUS
- Mme Soizic Benezech, ergothérapeute, Leguevin
- Pr Sylvain Brochard, médecin de MPR, Brest, filière FILNEMUS
- M Charles Campana, kinésithérapeute libéral, Le Havre
- Dr Claude Cances, neuropédiatre, Toulouse, filière FILNEMUS
- Dr Marieke Chamberon, médecin de MPR, Saint Maurice, filière AnDDI-Rares
- M Sylvain Darrous, ergothérapeute, Grenoble
- Pr Jean Dubousset, orthopédiste, Paris
- Dr Alice Fassier, orthopédiste, Lyon
- Mme Marion Fiat, kinésithérapeute, Saint Maurice, filière AnDDI-Rares
- Pr Franck Fitoussi, orthopédiste, Paris, filière AnDDI-Rares
- Dr Véronique Forin, médecin de MPR, Paris
- Dr Cyril Gitiaux, neuropédiatre, Paris, filière FILNEMUS
- Pr Pierre Simon Jouk, généticien, Grenoble, filière AnDDI-Rares
- Mme Violaine Maes Pascal, kinésithérapeute libérale, Grenoble
- Dr Véronique Manel, neuropédiatre, Lyon, filière FILNEMUS
- Dr Xenia Martin, généticienne, Grenoble, filière FILNEMUS/ AnDDI-Rares
- Dr Judith Melki, généticienne, Paris, filière AnDDI-Rares
- Dr Marie Doriane Merard, médecin de MPR, Lyon, filière FILNEMUS
- Dr Christian Morin, orthopédiste, Lyon
- Dr Mélanie Portes, médecin de MPR, Nimes
- Dr Susana Quijano-Roy, neuropédiatre, Paris, filière FILNEMUS
- Dr Nathalie Quintero, médecin de MPR, Saint Maurice, filière AnDDI-Rares
- Dr Marc Roquebert, médecin généraliste, Le Havre
- Dr Jean Solin, médecin du sport, Paris
- Dr Philippe Thoumié, médecin de MPR, Paris
- Dr Sandra Whalen, généticienne, Paris, filières FILNEMUS/ AnDDI-Rares

- Association Alliance Arthrogrypose

Gestion des intérêts déclarés
Tous les participants à l'élaboration du PNDS sur l'Arthrogrypose ont rempli une déclaration d'intérêt disponible sur le le site internet de l'HAS.

Les déclarations d'intérêt ont été analysées et prises en compte, en vue d'éviter les conflits d'intérêts, conformément au guide HAS « Guide des déclarations d'intérêts et de gestion des conflits d'intérêts » (HAS, 2010).

Modalités de concertation du groupe de travail multidisciplinaire

Date	Réunion	Emargement	Contenu
2019.09.09	Présentielle	Groupe des rédacteurs	Choix des mots clés pour la bibliographie Elaboration du plan du PNDS
2020.02.24	Présentielle	Groupe des rédacteurs	Lecture des parties rédigées par chaque rédacteur, corrections et envoi aux relecteurs.
2020.03.13	Présentielle/visio	Groupe des rédacteurs Groupe des relecteurs	Synthèse de la version 0.1
2020.05.04	Présentielle	Groupe des rédacteurs	Prise en compte des modifications du 13.03 et organisation de la poursuite de la rédaction.
2020.09.07	Présentielle	Groupe des rédacteurs	Lecture des parties rédigées par chaque rédacteur, corrections et envoi aux relecteurs.
2020.09.18	visio	Groupe des rédacteurs Groupe des relecteurs	Synthèse de la version 0.2
2021.03.08	Présentielle	Groupe des rédacteurs	Lecture des modifications apportées lors de la précédente synthèse et organisation de la poursuite de la rédaction.
2021.06.14	Présentielle	Groupe des rédacteurs	Lecture de la version finale et dernières corrections apportées avant envoi au groupe de relecteurs

Références bibliographiques

AMC + Diagnosis

Chromosomal microarray

Okubo Y, et al, A severe female case of arthrogryposis multiplex congenita with brain atrophy, spastic quadriplegia and intellectual disability caused by ZC4H2 mutation. Brain Dev. 2018 Apr;40(4):334-338.

Mohmaed am, et al, Intellectual disability secondary to a 16p13 duplication in a 1;16 translocation. Extended phenotype in a four-generation family. Am J Med Genet Part A 167A:128–136

Thevenon et al, Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. Am J Med Genet A. 2014 Dec;164A(12):3027-34.

Inbar-Feigenberg et al, Beals syndrome (congenital contractural arachnodactyly): prenatal ultrasound findings and molecular analysis.Ultrasound Obstet Gynecol 2014; 44: 486 – 490

Carrascosa-Romero et al, A 2.84 Mb Deletion at 21q22.11 in a Patient Clinically Diagnosed With Marden – Walker Syndrome. Am J Med Genet A. 2013 Sep;161A(9):2281-90

Zanzottera C et al, ZC4H2 deletions can cause severe phenotype in female carriers. Am J Med Genet A. 2017 May;173(5):1358-1363.

Balta A et al, Interstitial deletion 5p14.1-p15.2 and 5q14.3-q23.2 in a patient with clubfoot, blepharophimosis, arthrogryposis, and multiple congenital abnormalities. Am J Med Genet A. 2017, Oct;173(10):2798-2802.

Au et al, Review of the recurrent 8q13.2q13.3 branchio-oto-renal related microdeletion, and report of an additional case with associated distal arthrogryposis. Am J Med Genet A. 2016 Nov;170(11):2984-2987

Jonsson et al, A de novo 1.13 Mb microdeletion in 12q13.13 associated with congenital distal arthrogryposis, intellectual disability and mild dysmorphism. Eur J Med Genet. 2012 Jun;55(6-7):437-40.

Tabet et al, Molecular characterization of a de novo 6q24.2q25.3 duplication interrupting UTRN in a patient with arthrogryposis. Am J Med Genet A. 2010 Jul;152A(7):1781-8.

Lukusa et al, Pure de novo 17q25.3 micro duplication characterized by micro array CGH in a dysmorphic infant with growth retardation, developmental delay and distal arthrogryposis. Genet Couns. 2010;21(1):25-34.

Busche et al, A cryptic unbalanced translocation t(2;9)(p25.2;q34.3) causes the phenotype of 9q subtelomeric deletion syndrome and additional exophthalmos and joint contractures Eur J Med Genet. 2008 Nov-Dec;51(6):615-21.

Li Z et al. Phenotypic expansion of the interstitial 16p13.3 duplication: a case report and review of the literature. Gene. 2013 Dec 1;531(2):502-5.

Hancorova M et al. Chromosome 12q13.13 deletions involving the HOXC gene cluster: phenotype and candidate genes. Eur J Med Genet. 2013 Mar;56(3):171-3.

Chen CP et al. Prenatal diagnosis of partial trisomy 3q (3q27.3→qter) and partial monosomy 14q
(14q31.3→qter) of paternal origin associated with fetal hypotonia, arthrogryposis,scoliosis and hyperextensible joints. Gene. 2013 Mar 1;516(1):132-7.

Chen CP et al. Double aneuploidy with Edwards-Klinefelter syndromes (48,XXY,+18) of maternal origin: prenatal diagnosis and molecular cytogenetic characterization in a fetus with arthrogryposis of the left wrist and aplasia of the left thumb. Taiwan J Obstet Gynecol. 2011 Dec;50(4):479-84

Rameiri V et al. Microdeletion 3q syndrome J Craniofac Surg. 2011 Nov;22(6):2124-8.

Hagen A et al. Combined partial trisomy 11q and partial monosomy 10p in a 19-year-old female patient: phenotypic and genotypic findings. Am J Med Genet A. 2011 Dec;155A(12):3075-81.

Tabet AC 7 Mb de novo deletion within 8q21 in a patient with distal arthrogryposis type 2B (DA2B). Am J Med Genet A. 2010 Jul;152A(7):1781-8

Liewlick T et al. A Novel Sporadic 614-Kb Duplication of the 22q11.2 Chromosome in a Child With Amyoplasia J Child Neurol. 2011 Aug;26(8):1005-8.

Hiraki Y et al. A de novo deletion of 20q11.2-q12 in a boy presenting with abnormal hands and feet, retinal dysplasia, and intractable feeding difficulty. Am J Med Genet A. 2011 Feb;155A(2):409-14

Kim YJ et al. Miller-Dieker Syndrome with der(17)t(12;17)(q24.33;p13.3)pat Presenting with a Potential Risk of Mis-identification as a de novo Submicroscopic Deletion of 17p13.3. Korean J Lab Med. 2011 Jan; 31(1): 49–53

Thienpont B et al. Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. J Med Genet. 2010 Mar;47(3):155-61

Nakamura-Pereira M et al Sonographic findings in a case of tetrasomy 9p associated with increased nuchal translucency and Dandy-Walker malformation. Clin Ultrasound. 2009 Oakamura-Pereira Mct;37(8):471-4.
Ansari M et al. A syndromic form of Pierre Robin sequence is caused by 5q23 deletions encompassing FBN2 and PHAX European Journal of Medical Genetics 57 (2014) 587e595
Otake K et al. Congenital diaphragmatic hernia with a pure duplication of chromosome 1q: report of the first surviving case Pediatr Surg Int (2009) 25: 827
Chen CP et al. Concomitant omphalocele and anencephaly associated with trisomy 18 and arthrogryposis diagnosed in early pregnancy. Taiwan J Obstet Gynecol. 2008 Mar;47(1):93-4
Kosaki R et al. Discrepancies in cytogenetic results between amniocytes and postnatally obtained blood: trisomy 9 mosaicism. Congenit Anom (Kyoto). 2006 Jun;46(2):115-7
Peng HH et al. Prenatal diagnosis of partial trisomy 12q: clinical presentations and outcome. Prenat Diagn 2005; 25: 470–474
Grati FR et al. Three cases with de novo 6q imbalance and variable prenatal phenotype. Am J Med Genet A. 2005 Jul 30;136(3):254-8
Destree et al. Prenatal diagnosis of trisomy 6 mosaicism. Prenat Diagn 2005; 25: 354–357
Chen CP. Arthrogryposis of the wrist and ankle associated with fetal trisomy 18. Prenat Diagn. 2005
May;25(5):423-5. doi: 10.1002/pd.1141. PMID: 15906415.
Castro Gago M et al. Neurogenic Arthrogryposis Multiplex Congenita and Velopharyngeal Incompetence Associated With Chromosome 22q11.2 Deletion. J Child Neurol. 2005 Jan;20(1):76-8
Denizot S et. al. Distal Arthrogryposis and Neonatal Hypotonia: an Unusual Presentation of Prader–Willi Syndrome (PWS). J Perinatol. 2004 Nov;24(11):733-4
Devriendt K et al. Velocardiofacial syndrome presenting as distal arthrogryposis. Eur J Pediatr. 2004 Jun;163(6):329-30
Punal E et al. Phenotypic variability of deletion 22q11.2. An analysis of 16 observations with special emphasis on the neurological manifestations. Rev Neurol. 2003 Oct 1-15;37(7):601-
Wu YC et al. Prenatal diagnosis of mosaic tetrasomy 10p associated with megacisterna magna, echogenic focus of left ventricle, umbilical cord cysts and distal arthrogryposis. Am J Med Genet A. 2003 Mar 15;117A(3):278-81
Enya T et al. Three patients with Schaaf-Yang syndrome exhibiting arthrogryposis and endocrinological abnormalities. Am J Med Genet A. 2018 Mar;176(3):707-711. doi: 10.1002/ajmg.a.38606. Epub 2018 Jan 23.
Hague J et al. Molecularly proven mosaicism in phenotypically normal parent of a girl with Freeman-Sheldon Syndrome caused by a pathogenic MYH3 mutation. Am J Med Genet A. 2016 Jun;170(6):1608-12. doi: 10.1002/ajmg.a.37631. Epub 2016 Mar 2
Mejlachowicz D, et al; Truncating Mutations of MAGEL2, a Gene within the Prader-Willi Locus, Are Responsible for Severe Arthrogryposis. Am J Hum Genet. 2015 Oct 1;97(4):616-20. doi: 10.1016/j.ajhg.2015.08.010. Epub 2015 Sep 10
Alamillo CL et al; Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. Prenat Diagn. 2015 Nov;35(11):1073-8. doi: 10.1002/pd.4648. Epub 2015 Aug 3
Karaman A et al. Concomitant omphalocele, anencephaly and arthrogryposis associated with trisomy 18. Genet Couns. 2015;26(1):77-9.
Chen CP Detection of mosaic isochromosome 20q in amniotic fluid in a pregnancy with fetal arthrogryposis multiplex congenita and normal karyotype in fetal blood and postnatal samples of placenta, skin, and liver. Prenat Diagn. 2003 Jan;23(1):85-7

Whole exome/ genome sequencing

Zho H et al.; EMBO Mol Med. 2019. Feb 18.pii:e9709.doi: 10. 15252/emmm.201809709
Chervinsky E et al. Am J Med Genet A.2018;176(4):1001-1005. doi: 10.1002/ajmg.a.38639.
Montiero FP et al. Eur J Med Genet. 2019. pii: S1769-7212
Jobling R et al.J Med Genet.2018 May;55(5):316-321.doi:10:1136/jmedgenet-2017-105222
Abiusi E, et al.Hum Mol Genet. 2017 Oct 15;26(20): 3989-3994, doi:10.1093/hmg/ddx288
Tan QK et al.Cold Spring Harb Mol Case Stud. 2017 Nov 21;3(6). pii: a002063. doi: 10.1101/mcs.a002063
Berkenstadt JM et al. J Ultrasound Med. 2018 Jul;37(7):1827-1833. doi: 10.1002/jum.14520. Epub 2018 Jan 13
Casey J et al. Prenat Diagn. 2016;36(11):1020-1026. doi: 10.1002/pd.4925. Epub 2016 Oct 2.
Bayram Y et al. J Clin Invest. 2016 Feb;126(2):762-78. doi: 10.1172/JCI84457. Epub 2016 Jan 11
Bauche S et al. Am J Hum Genet. 2016;99(3):753-761. doi: 10.1016/j.ajhg.2016.06.033. Epub 2016 Aug 25
Enya T et al. Am J Med Genet A. 2018 Mar;176(3):707-711. doi: 10.1002/ajmg.a.38606. Epub 2018 Jan 23.
Reinstein E et al. Clin Genet. 2018 Jan;93(1):160-163. doi: 10.1111/cge.13018. Epub 2017 Jul 26
Mroczek M et al. J Appl Genet. 2017; 58(2): 199–203
Knierim E et al. Am J Hum Genet. 2016 Mar 3;98(3):473-489. doi: 10.1016/j.ajhg.2016.01.006. Epub 2016

Jo HY et al.Clin Genet. 2016 Aug;90(2):177-81. doi: 10.1111/cge.12714. Epub 2016 Mar 4
Wambach JA et al. Am J Hum Genet. 2016 Nov 3;99(5):1206-1216. doi: 10.1016/j.ajhg.2016.09.019. Epub 2016 Oct 27
Ekhilevitch N et al. Clin Genet. 2016 Jul;90(1):84-9. doi: 10.1111/cge.12707. Epub 2016 Jan 20
Wambach JA et al. Hum Mutat. 2017;38(11):1477-1484. doi: 10.1002/humu.23297. Epub 2017 Aug 17
Todd EJ et al. Orphanet J Rare Dis. 2015 Nov 17;10:148. doi: 10.1186/s13023-015-0364-0
Wang B et al. Am J Med Genet A. 2016 ;170A(1):135-41. doi: 10.1002/ajmg.a.37391. Epub 2015 Sep 16
Mejlachowicz D et al. Am J Hum Genet. 2015;97(4):616-20. doi: 10.1016/j.ajhg.2015.08.010. Epub 2015 Sep 1
Ravenscroft G et al. Am J Hum Genet. 2015;96(6):955-61. doi:10.1016/j.ajhg.2015.04.014. Epub 2015 May 21
Okubo M et al.Am J Med Genet A. 2015;167A(5):1100-6. doi: 10.1002/ajmg.a.36881. Epub 2015 Feb 25
Dohrn N et al. Am J Med Genet A. 2015;167A(4):731-43. doi: 10.1002/ajmg.a.37018. Epub 2015 Feb 23
Wilbe M et al. J Med Genet. 2015 Mar;52(3):195-202. doi: 10.1136/jmedgenet-2014-102730. Epub 2015 Jan 2
Seo J et al. J Hum Genet. 2015;60(4):213-5. doi: 10.1038/jhg.2015.2. Epub 2015 Jan 22
Patel N et al. Hum Mol Genet. 2014;23(24):6584-93. doi: 10.1093/hmg/ddu384. Epub 2014 Jul 23el
Shaaban S et al. Clin Genet. 2014;85(6):562-7. doi: 10.1111/cge.12224. Epub 2013 Jul 19
Davidson AE et al. Brain. 2013;136(Pt 2):508-21. doi: 10.1093/brain/aws344
Ravenscroft G et al.Neuromuscul Disord. 2013;23(2):165-9. doi: 10.1016/j.nmd.2012.11.005. Epub 2012 Dec 3
Markus B et al. Hum Mutat. 2012 Oct;33(10):1435-8. doi: 10.1002/humu.22122. Epub 2012 Jun 7
McMillin MJ et al.Am J Hum Genet. 2013 Jan 10;92(1):150-6. doi: 10.1016/j.ajhg.2012.11.014. Epub 2012 Dec 20

Maladie de Steinert/ DMPK

Kahn JA, Cataltepe S. A 35-week neonate with respiratory failure, hypotonia, and joint contractures. Curr Opin Pediatr. 1996 Dec;8(6):583-8.
Martinello F, Piazza A, Pastorello E, Angelini C, Trevisan CP. Clinical and neuroimaging study of central nervous system in congenital myotonic dystrophy. J Neurol. 1999 Mar;246(3):186-92.
González de Dios J, Martínez Frías ML, Egües Jimeno J, Gairi Tahull JM, Gómez Sabrido F, Morales Fernández MC, Paisán Grisolía L, Pardo Romero M, Medina Rams M. [Epidemiological study of Steinert's congenital myotonic dystrophy: dysmorphological characteristics]. An Esp Pediatr. 1999 Oct;51(4):389-96.
Akiyama M, Yuza Y, Yokokawa Y, Yokoi K, Ariga M, Eto Y. Differences in CTG triplet repeat expansion in leukemic cells and normal lymphocytes from a 14-year-old female with congenital myotonic dystrophy. Pediatr Blood Cancer. 2008 Oct;51(4):563-5.
Schilling L, Forst R, Forst J, Fujak A. Orthopaedic Disorders in Myotonic Dystrophy Type 1: descriptive clinical study of 21 patients. BMC Musculoskelet Disord. 2013 Dec 1;14:338. doi: 10.1186/1471-2474-14-338.
Lowry RB, Sibbald B, Bedard T, Hall JG. Birth Defects Res A Clin Mol Teratol. 2010 Dec;88(12):1057-61. doi: 10.1002/bdra.20738.

Prader Willi syndrome

Denizot S, Boscher C, Le Vaillant C, Rozé JC, Gras Le Guen C. J Perinatol. 2004 Nov;24(11):733-4.
Bigi N, Faure JM, Coubes C, Puechberty J, Lefort G, Sarda P, Blanchet P. Prenat Diagn. 2008 Sep;28(9):796-9.

Amyotrophie spinale/ SMN1

Bingham PM, Shen N, Rennert H, Rorke LB, Black AW, Marin-Padilla MM, Nordgren RE. Arthrogryposis due to infantile neuronal degeneration associated with deletion of the SMNT gene. Neurology. 1997 Sep;49(3):848-51.
Bürglen L, Amiel J, Viollet L, Lefebvre S, Burlet P, Clermont O, Raclin V, Landrieu P, Verloes A, Munnich A, Melki J. Survival motor neuron gene deletion in the arthrogryposis multiplex congenita-spinal muscular atrophy association. J Clin Invest. 1996 Sep 1;98(5):1130-2.
Falsaperla R, Romeo G, Di Giorgio A, Pavone P, Parano E, Connolly AM. Long-term survival in a child with arthrogryposis multiplex congenita and spinal muscular atrophy. J Child Neurol. 2001 Dec;16(12):934-6.
Nadeau A, D'Anjou G, Debray G, Robitaille Y, Simard LR, Vanasse M. A newborn with spinal muscular atrophy type 0 presenting with a clinicopathological picture suggestive of myotubular myopathy. J Child Neurol. 2007 Nov;22(11):1301-4.

Grotto S, Cuisset JM, Marret S, Drunat S, Faure P, Audebert-Bellanger S, Desguerre I, Flurin V, Grebille AG, Guerrot AM, Journel H, Morin G, Plessis G, Renolleau S, Roume J, Simon-Bouy B, Touraine R, Willems M, Frébourg T, Verspyck E, Saugier-Veber P. Type 0 Spinal Muscular Atrophy: Further Delineation of Prenatal and Postnatal Features in 16 Patients. J Neuromuscul Dis. 2016 Nov 29;3(4):487-495.
MacLeod MJ, Taylor JE, Lunt PW, Mathew CG, Robb SA. Prenatal onset spinal muscular atrophy. Eur J Paediatr Neurol. 1999;3(2):65-72.

Electromyographie

Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, Pantaleoni C. ZC4H2 deletions can cause severe phenotype in female carriers. Am J Med Genet A. 2017 May;173(5):1358-1363.
Zafeiriou DI, Pitt M, de Sousa C. Clinical and neurophysiological characteristics of congenital myasthenic syndromes presenting in early infancy. Brain Dev. 2004 Jan;26(1):47-52.
Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Novel BICD2 mutation in a Japanese family with autosomal dominant lower extremity-predominant spinal muscular atrophy-2. Brain Dev. 2018 Apr;40(4):343-347.
Yıldırım Y, Orhan EK, Iseri SA, Serdaroglu-Oflazer P, Kara B, Solakoğlu S, Tolun A. A frameshift mutation of ERLIN2 in recessive intellectual disability, motor dysfunction and multiple joint contractures. Hum Mol Genet. 2011 May 15;20(10):1886-92.
Verma S, Goyal P, Guglani L, Peinhardt C, Pelzek D, Barkhaus PE. COL6A and LAMA2 Mutation Congenital Muscular Dystrophy: A Clinical and Electrophysiological Study. J Clin Neuromuscul Dis. 2018 Mar;19(3):108-116.
Kang PB, Lidov HG, David WS, Torres A, Anthony DC, Jones HR, Darras BT. Diagnostic value of electromyography and muscle biopsy in arthrogryposis multiplex congenita. Ann Neurol. 2003 Dec;54(6):790-5.
Reddel S, Ouvrier RA, Nicholson G, Dierick I, Irobi J, Timmerman V, Ryan MM. Autosomal dominant congenital spinal muscular atrophy--a possible developmental deficiency of motor neurones? Neuromuscul Disord. 2008 Jul;18(7):530-5.
Rabie M, Jossiphov J, Nevo Y. Electromyography (EMG) accuracy compared to muscle biopsy in childhood. J Child Neurol. 2007 Jul;22(7):803-8.
Ko JM, Choi IH, Baek GH, Kim KW. First Korean family with a mutation in TPM2 associated with Sheldon-Hall syndrome. J Korean Med Sci. 2013 May;28(5):780-3.
Kiselev A, Vaz R, Knyazeva A, Khudiakov A, Tarnovskaya S, Liu J, Sergushichev A, Kazakov S, Frishman D, Smolina N, Pervunina T, Jorholt J, Sjoberg G, Vershinina T, Rudenko D, Arner A, Sejersen T, Lindstrand A, Kostareva A. De novo mutations in FLNC leading to early-onset restrictive cardiomyopathy and congenital myopathy. Hum Mutat. 2018 Sep;39(9):1161-1172.
Gaitanis JN, McMillan HJ, Wu A, Darras BT. Electrophysiologic evidence for anterior horn cell disease in amyoplasia. Pediatr Neurol. 2010 Aug;43(2):142-7.
Fusco C, Frattini D, Salerno GG, Canali E, Bernasconi P, Maggi L. New phenotype and neonatal onset of sodium channel myotonia in a child with a novel mutation of SCN4A gene. Brain Dev. 2015 Oct;37(9):891-3.
Fleming J, Quan D. A case of congenital spinal muscular atrophy with pain due to a mutation in TRPV4. Neuromuscul Disord. 2016 Dec;26(12):841-843.
Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316-9.

Créatine Phosphokinase

Kölbel H, Abicht A, Schwartz O, Katona I, Paulus W, Neuen-Jacob E, Weis J, Schara U. Characteristic clinical and ultrastructural findings in nesprinopathies. Eur J Paediatr Neurol. 2019 Mar;23(2):254-261.
Astrea G, Romano A, Angelini C, Antozzi CG, Barresi R, Battini R, Battisti C, Bertini E, Bruno C, Cassandrini D, Fanin M, Fattori F, Fiorillo C, Guerrini R, Maggi L, Mercuri E, Morani F, Mora M, Moro F, Pezzini I, Picillo E, Pinelli M,Politano L, Rubegni A, Sanseverino W, Savarese M, Striano P, Torella A, Trevisan CP, Trovato R, Zaraieva I, Muntoni F, Nigro V, D'Amico A, Santorelli FM; Italian CMD Network. Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet J Rare Dis. 2018 Sep 26;13(1):170.
Conant A, Curiel J, Pizzino A, Sabetrasekh P, Murphy J, Bloom M, Evans SH, Helman G, Taft RJ, Simons C, Whitehead MT, Moore SA, Vanderver A. Absence of Axoglial Paranodal Junctions in a Child With CNTNAP1 Mutations, Hypomyelination, and Arthrogryposis. J Child Neurol. 2018 Sep;33(10):642-650.
Sivaraman I, Friedman NR, Prayson RA. Muscle biopsy findings in a child with NALCN gene mutation. J Clin Neurosci. 2016 Dec;34:222-223.

Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Novel BICD2 mutation in a Japanese family with autosomal dominant lower extremity-predominant spinal muscular atrophy-2. Brain Dev. 2018 Apr;40(4):343-347.
Janecke AR, Li B, Boehm M, Krabichler B, Rohrbach M, Müller T, Fuchs I, Golas G, Katagiri Y, Ziegler SG, Gahl WA, Wilnai Y, Zoppi N, Geller HM, Giunta C, Slavotinek A, Steinmann B. The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to CHST14 mutations. Am J Med Genet A. 2016 Jan;170A(1):103-15.
Hunter JM, Ahearn ME, Balak CD, Liang WS, Kurdoglu A, Corneveaux JJ, Russell M, Huentelman MJ, Craig DW, Carpten J, Coons SW, DeMello DE, Hall JG, Bernes SM, Baumbach-Reardon L. Novel pathogenic variants and genes for myopathies identified by whole exome sequencing. Mol Genet Genomic Med. 2015 Jul;3(4):283-301.
Echaniz-Laguna A, Dubourg O, Carlier P, Carlier RY, Sabouraud P, Péréon Y, Chapon F, Thauvin-Robinet C, Laforêt P, Eymard B, Latour P, Stojkovic T. Phenotypic spectrum and incidence of TRPV4 mutations in patients with inherited axonal neuropathy. Neurology. 2014 May 27;82(21):1919-26.
Dlamini N, Josifova DJ, Paine SM, Wraige E, Pitt M, Murphy AJ, King A, Buk S, Smith F, Abbs S, Sewry C, Jacques TS, Jungbluth H. Clinical and neuropathological features of X-linked spinal muscular atrophy (SMAX2) associated with a novel mutation in the UBA1 gene. Neuromuscul Disord. 2013 May;23(5):391-8.
Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316-9.
Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. A mutation in the fast skeletal muscle troponin I gene causes myopathy and distal arthrogryposis. Neurology. 2006 Aug 22;67(4):597-601.
Chauveau C, Bonnemann CG, Julien C, Kho AL, Marks H, Talim B, Maury P, Arne-Bes MC, Uro-Coste E, Alexandrovich A, Vihola A, Schafer S, Kaufmann B, Medne L, Hübner N, Foley AR, Santi M, Udd B, Topaloglu H, Moore SA, Gotthardt M, Samuels ME, Gautel M, Ferreiro A. Recessive TTN truncating mutations define novel forms of core myopathy with heart disease. Hum Mol Genet. 2014 Feb 15;23(4):980-91.
Philpot J, Counsell S, Bydder G, Sewry CA, Dubowitz V, Muntoni F. Neonatal arthrogryposis and absent limb muscles: a muscle developmental gene defect? Neuromuscul Disord. 2001 Jul;11(5):489-93.
Vasli N, Harris E, Karamchandani J, Bareke E, Majewski J, Romero NB, Stojkovic T, Barresi R, Tasfaout H, Charlton R, Malfatti E, Bohm J, Marini-Bettolo C, Choquet K, Dicaire MJ, Shao YH, Topf A, O'Ferrall E, Eymard B, Straub V, Blanco G, Lochmüller H, Brais B, Laporte J, Tétreault M. Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. Brain. 2017 Jan;140(1):37-48.

Biopsie musculaire – arthrogryposes distales

Sandaradura SA, Bournazos A, Mallawaarachchi A, Cummings BB, Waddell LB, Jones KJ, Troedson C, Sudarsanam A, Nash BM, Peters GB, Algar EM, MacArthur DG, North KN, Brammah S, Charlton A, Laing NG, Wilson MJ, Davis MR, Cooper ST. Nemaline myopathy and distal arthrogryposis associated with an autosomal recessive TNNT3 splice variant. Hum Mutat. 2018 Mar;39(3):383-388.
Haliloglu G, Becker K, Temucin C, Talim B, Küçükşahin N, Pergande M, Motameny S, Nürnberg P, Aydingoz U, Topaloglu H, Cirak S. Recessive PIEZO2 stop mutation causes distal arthrogryposis with distal muscle weakness, scoliosis and proprioception defects. J Hum Genet. 2017 Apr;62(4):497-501.
Sivaraman I, Friedman NR, Prayson RA. Muscle biopsy findings in a child with NALCN gene mutation. J Clin Neurosci. 2016 Dec;34:222-223.
Thevenon J, Monnier N, Callier P, Dieterich K, Francoise M, Montgomery T, Kjaergaard S, Neas K, Dixon J, Dahm TL, Huet F, Ragon C, Mosca-Boidron AL, Marle N, Duplomb L, Aubriot-Lorton MH, Mugneret F, Vokes SA, Tucker HW, Lunardi J, Faivre L, Jouk PS, Thauvin-Robinet C. Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. Am J Med Genet A. 2014 Dec;164A(12):3027-34.
Davidson AE, Siddiqui FM, Lopez MA, Lunt P, Carlson HA, Moore BE, Love S, Born DE, Roper H, Majumdar A, Jayadev S, Underhill HR, Smith CO, von der Hagen M, Hubner A, Jardine P, Merrison A, Curtis E, Cullup T, Jungbluth H, Cox MO, Winder TL, Abdel Salam H, Li JZ, Moore SA, Dowling JJ. Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. Brain. 2013 Feb;136(Pt 2):508-21.
Dieterich K, Quijano-Roy S, Monnier N, Zhou J, Fauré J, Smirnow DA, Carlier R, Laroche C, Marcorelles P, Mercier S, Mégarbané A, Odent S, Romero N, Sternberg D, Marty I, Estournet B, Jouk PS, Melki J, Lunardi J. The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthrogryposis. Hum Mol Genet. 2013 Apr 15;22(8):1483-92.
Tajsharghi H, Kimber E, Holmgren D, Tulinius M, Oldfors A. Distal arthrogryposis and muscle weakness associated with a beta-tropomyosin mutation. Neurology. 2007 Mar 6;68(10):772-5.
Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. Distal arthrogryposis: clinical and genetic findings. Acta Paediatr. 2012 Aug;101(8):877-87.

Vydyanath A, Gurnett CA, Marston S, Luther PK. Axial distribution of myosin binding protein-C is unaffected by mutations in human cardiac and skeletal muscle. J Muscle Res Cell Motil. 2012 May;33(1):61-74.
Oldfors A, Lamont PJ. Thick filament diseases. Adv Exp Med Biol. 2008;642:78-91.
Kimber E, Tajsharghi H, Kroksmark AK, Oldfors A, Tulinius M. A mutation in the fast skeletal muscle troponin I gene causes myopathy and distal arthrogryposis. Neurology. 2006 Aug 22;67(4):597-601.
Tajsharghi H, Kimber E, Kroksmark AK, Jerre R, Tulinius M, Oldfors A. Embryonic myosin heavy-chain mutations cause distal arthrogryposis and developmental myosin myopathy that persists postnatally. Arch Neurol. 2008 Aug;65(8):1083-90.
Hageman G, Jennekens FG, Vette JK, Willemse J. The heterogeneity of distal arthrogryposis. Brain Dev. 1984;6(3):273-83.
Monnier N, Lunardi J, Marty I, Mezin P, Labarre-Vila A, Dieterich K, Jouk PS. Absence of beta-tropomyosin is a new cause of Escobar syndrome associated with nemaline myopathy. Neuromuscul Disord. 2009 Feb;19(2):118-23. doi: 10.1016/j.nmd.2008.11.009. Epub 2009 Jan 19. PMID: 19155175.
Sung SS, Brassington AM, Krakowiak PA, Carey JC, Jorde LB, Bamshad M. Mutations in TNNT3 cause multiple congenital contractures: a second locus for distal arthrogryposis type 2B. Am J Hum Genet. 2003 Jul;73(1):212-4. doi: 10.1086/376418. PMID: 12865991; PMCID: PMC1180583.

Biopsie musculaire – Amyoplasie

Gaitanis JN, McMillan HJ, Wu A, Darras BT. Pediatr Neurol. 2010 Aug;43(2):142-7.
Price DS. Arch Dis Child. 1933 Oct;8(47):343-54

Biospie musculaire – Arthrogrypose multiple congénitale

Carrera-García L, Natera-de Benito D, Dieterich K, de la Banda MGG, Felter A, Inarejos E, Codina A, Jou C, Roldan M, Palau F, Hoenicka J, Pijuan J, Ortez C, Expósito-Escudero J, Durand C, Nugues F, Jimenez-Mallebrera C, Colomer J, Carlier RY, Lochmüller H, Quijano-Roy S, Nascimento A. Am J Med Genet A. 2019 Jun;179(6):915-926.
Alkhunaizi E, Shuster S, Shannon P, Siu VM, Darilek S, Mohila CA, Boissel S, Ellezam B, Fallet-Bianco C, Laberge AM, Zandberg J, Injeyan M, Hazrati LN, Hamdan F, Chitayat D. Am J Med Genet A. 2019 Mar;179(3):386-396.
Bonnin E, Cabochette P, Filosa A, Jühlen R, Komatsuzaki S, Hezwani M, Dickmanns A, Martinelli V, Vermeersch M, Supply L, Martins N, Pirenne L, Ravenscroft G, Lombard M, Port S, Spillner C, Janssens S, Roets E, Van Dorpe J, Lammens M, Kehlenbach RH, Ficner R, Laing NG, Hoffmann K, Vanhollebeke B, Fahrenkrog B. PLoS Genet. 2018 Dec 13;14(12):e1007845.
Berkenstadt M, Pode-Shakked B, Barel O, Barash H, Achiron R, Gilboa Y, Kidron D, Raas-Rothschild A. J Ultrasound Med. 2018 Jul;37(7):1827-1833.
Ahmed AA, Skaria P, Safina NP, Thiffault I, Kats A, Taboada E, Habeebu S, Saunders C. Am J Med Genet A. 2018 Feb;176(2):359-367.
Yoshioka M, Morisada N, Toyoshima D, Yoshimura H, Nishio H, Iijima K, Takeshima Y, Uehara T, Kosaki K. Brain Dev. 2018 Apr;40(4):343-347.
Sandaradura SA, Bournazos A, Mallawaarachchi A, Cummings BB, Waddell LB, Jones KJ, Troedson C, Sudarsanam A, Nash BM, Peters GB, Algar EM, MacArthur DG, North KN, Brammah S, Charlton A, Laing NG, Wilson MJ, Davis MR, Cooper ST. Hum Mutat. 2018 Mar;39(3):383-388.
Tan QK, McConkie-Rosell A, Juusola J, Gustafson KE, Pizoli CE, Buckley AF, Jiang YH. Cold Spring Harb Mol Case Stud. 2017 Nov 21;3(6). pii: a002063. doi: 10.1101/mcs.a002063.
Quélin C, Loget P, Rozel C, D'Hervé D, Fradin M, Demurger F, Odent S, Pasquier L, Cavé H, Marcorelles P. Eur J Med Genet. 2017 Jul;60(7):395-398.
Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, Pantaleoni C. Am J Med Genet A. 2017 May;173(5):1358-1363.
Chimelli L, Melo ASO, Avvad-Portari E, Wiley CA, Camacho AHS, Lopes VS, Machado HN, Andrade CV, Dock DCA, Moreira ME, Tovar-Moll F, Oliveira-Szejnfeld PS, Carvalho ACG, Ugarte ON, Batista AGM, Amorim MMR, Melo FO, Ferreira TA, Marinho JRL, Azevedo GS, Leal JIBF, da Costa RFM, Rehen S, Arruda MB, Brindeiro RM, Delvechio R, Aguiar RS, Tanuri A. Acta Neuropathol. 2017 Jun;133(6):983-999.
Fernández-Marmiesse A, Carrascosa-Romero MC, Alfaro Ponce B, Nascimento A, Ortez C, Romero N, Palacios L, Jimenez-Mallebrera C, Jou C, Gouveia S, Couce ML. Neuromuscul Disord. 2017 Feb;27(2):188-192.
Haliloglu G, Becker K, Temucin C, Talim B, Küçükşahin N, Pergande M, Motameny S, Nürnberg P, Aydingoz U, Topaloglu H, Cirak S. J Hum Genet. 2017 Apr;62(4):497-501.
Baumann M, Steichen-Gersdorf E, Krabichler B, Petersen BS, Weber U, Schmidt WM, Zschocke J, Müller T, Bittner RE, Janecke AR. Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotype-phenotype correlation. Eur J Hum Genet. 2017 Feb;25(2):262-266.

Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. Neuromuscul Disord. 2016 Oct;26(10):681-687.
Sivaraman I, Friedman NR, Prayson RA. J Clin Neurosci. 2016 Dec;34:222-223.
Rudnik-Schöneborn S, Deden F, Eggermann K, Eggermann T, Wieczorek D, Sellhaus B, Yamoah A, Goswami A, Claeys KG, Weis J, Zerres K. Muscle Nerve. 2016 Sep;54(3):496-500.
Lazier J, Mah JK, Nikolic A, Wei XC, Samedi V, Fajardo C, Brindle M, Perrier R, Thomas MA. Neuromuscul Disord. 2016 Jan;26(1):56-9.
Evangelista T, Bansagi B, Pyle A, Griffin H, Douroudis K, Polvikoski T, Antoniadi T, Bushby K, Straub V, Chinnery PF, Lochmüller H, Horvath R. Neuromuscul Disord. 2015 Jun;25(6):516-21.
Dohrn N, Le VQ, Petersen A, Skovbo P, Pedersen IS, Ernst A, Krarup H, Petersen MB. Am J Med Genet A. 2015 Apr;167A(4):731-43.
Wilbe M, Ekvall S, Eurenius K, Ericson K, Casar-Borota O, Klar J, Dahl N, Ameur A, Annerén G, Bondeson ML. J Med Genet. 2015 Mar;52(3):195-202.
Scoto M, Rossor AM, Harms MB, Cirak S, Calissano M, Robb S, Manzur AY, Martínez Arroyo A, Rodriguez Sanz A, Mansour S, Fallon P, Hadjikoumi I, Klein A, Yang M, De Visser M, Overweg-Plandsoen WC, Baas F, Taylor JP, Benatar M, Connolly AM, Al-Lozi MT, Nixon J, de Goede CG, Foley AR, Mcwilliam C, Pitt M, Sewry C, Phadke R, Hafezparast M, Chong WK, Mercuri E, Baloh RH, Reilly MM, Muntoni F. Neurology. 2015 Feb 17;84(7):668-79.,
Tan-Sindhunata MB, Mathijssen IB, Smit M, Baas F, de Vries JI, van der Voorn JP, Kluijt I, Hagen MA, Blom EW, Sistermans E, Meijers-Heijboer H, Waisfisz Q, Weiss MM, Groffen AJ. Eur J Hum Genet. 2015 Sep;23(9):1151-7.
Rossor AM, Oates EC, Salter HK, Liu Y, Murphy SM, Schule R, Gonzalez MA, Scoto M, Phadke R, Sewry CA, Houlden H, Jordanova A, Tournev I, Chamova T, Litvinenko I, Zuchner S, Herrmann DN, Blake J, Sowden JE, Acsadi G, Rodriguez ML, Menezes MP, Clarke NF, Auer Grumbach M, Bullock SL, Muntoni F, Reilly MM, North KN. Brain. 2015 Feb;138(Pt 2):293-310.
Chauveau C, Bonnemann CG, Julien C, Kho AL, Marks H, Talim B, Maury P, Arne-Bes MC, Uro-Coste E, Alexandrovich A, Vihola A, Schafer S, Kaufmann B, Medne L, Hübner N, Foley AR, Santi M, Udd B, Topaloglu H, Moore SA, Gotthardt M, Samuels ME, Gautel M, Ferreiro A. Hum Mol Genet. 2014 Feb 15;23(4):980-91.
Bharucha-Goebel DX, Santi M, Medne L, Zukosky K, Dastgir J, Shieh PB, Winder T, Tennekoon G, Finkel RS, Dowling JJ, Monnier N, Bönnemann CG. Neurology. 2013 Apr 23;80(17):1584-9.
Dlamini N, Josifova DJ, Paine SM, Wraige E, Pitt M, Murphy AJ, King A, Buk S, Smith F, Abbs S, Sewry C, Jacques TS, Jungbluth H. . Neuromuscul Disord. 2013 May;23(5):391-8.
Davidson AE, Siddiqui FM, Lopez MA, Lunt P, Carlson HA, Moore BE, Love S, Born DE, Roper H, Majumdar A, Jayadev S, Underhill HR, Smith CO, von der Hagen M, Hubner A, Jardine P, Merrison A, Curtis E, Cullup T, Jungbluth H, Cox MO, Winder TL, Abdel Salam H, Li JZ, Moore SA, Dowling JJ. Brain. 2013 Feb;136(Pt 2):508-21.,
Ravenscroft G, Thompson EM, Todd EJ, Yau KS, Kresoje N, Sivadorai P, Friend K, Riley K, Manton ND, Blumbergs P, Fietz M, Duff RM, Davis MR, Allcock RJ, Laing NG. Neuromuscul Disord. 2013 Feb;23(2):165-9.
Yonath H, Reznik-Wolf H, Berkenstadt M, Eisenberg-Barzilai S, Lehtokari VL, Wallgren-Pettersson C, Mehta L, Achiron R, Gilboa Y, Polak-Charcon S, Winder T, Frydman M, Pras E. Prenat Diagn. 2012 Jan;32(1):70-4.
Yıldırım Y, Orhan EK, Iseri SA, Serdaroglu-Oflazer P, Kara B, Solakoğlu S, Tolun A. Hum Mol Genet. 2011 May 15;20(10):1886-92.
Hernandez-Lain A, Husson I, Monnier N, Farnoux C, Brochier G, Lacène E, Beuvin M, Viou M, Manéré L, Claeys KG, Fardeau M, Lunardi J, Voit T, Romero NB. Eur J Med Genet. 2011 Jan-Feb;54(1):29-33.
Gurnett CA, Desruisseau DM, McCall K, Choi R, Meyer ZI, Talerico M, Miller SE, Ju JS, Pestronk A, Connolly AM, Druley TE, Weihl CC, Dobbs MB. Hum Mol Genet. 2010 Apr 1;19(7):1165-73.

IRM médullaire

Aragao MFVV, Holanda AC, Brainer-Lima AM, Petribu NCL, Castillo M, van der Linden V, Serpa SC, Tenório AG, Travassos PTC, Cordeiro MT, Sarteschi C, Valenca MM, Costello A. Nonmicrocephalic Infants with Congenital Zika Syndrome Suspected Only after Neuroimaging Evaluation Compared with Those with Microcephaly at Birth and Postnatally: How Large Is the Zika Virus "Iceberg"? AJNR Am J Neuroradiol. 2017 Jul;38(7):1427-1434. doi: 10.3174/ajnr.A5216. Epub 2017 May 18. PMID: 28522665; PMCID: PMC7959892.

IRM musculaire

Carrera-García L, Natera-de Benito D, Dieterich K, de la Banda MGG, Felter A, Inarejos E, Codina A, Jou C, Roldan M, Palau F, Hoenicka J, Pijuan J, Ortez C, Expósito-Escudero J, Durand C, Nugues F, Jimenez-

Mallebrera C, Colomer J, Carlier RY, Lochmüller H, Quijano-Roy S, Nascimento A. CHRNG-related nonlethal multiple pterygium syndrome: Muscle imaging pattern and clinical, histopathological, and molecular genetic findings. Am J Med Genet A. 2019 Jun;179(6):915-926. doi: 10.1002/ajmg.a.61122. Epub 2019 Mar 14. PMID: 30868735.

Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. A novel neuromuscular form of glycogen storage disease type IV with arthrogryposis, spinal stiffness and rare polyglucosan bodies in muscle. Neuromuscul Disord. 2016 Oct;26(10):681-687. doi: 10.1016/j.nmd.2016.07.005. Epub 2016 Jul 25. PMID: 27546458.

Jarraya M, Quijano-Roy S, Monnier N, Béhin A, Avila-Smirnov D, Romero NB, Allamand V, Richard P, Barois A, May A, Estournet B, Mercuri E, Carlier PG, Carlier RY. Whole-Body muscle MRI in a series of patients with congenital myopathy related to TPM2 gene mutations. Neuromuscul Disord. 2012 Oct 1;22 Suppl 2:S137-47. doi: 10.1016/j.nmd.2012.06.347. PMID: 22980765.

Mercuri E, Manzur A, Main M, Alsopp J, Muntoni F. Is there post-natal muscle growth in amyoplasia? A sequential MRI study. Neuromuscul Disord. 2009 Jun;19(6):444-5. doi: 10.1016/j.nmd.2009.03.006. Epub 2009 May 27. PMID: 19477646.

Philpot J, Counsell S, Bydder G, Sewry CA, Dubowitz V, Muntoni F. Neonatal arthrogryposis and absent limb muscles: a muscle developmental gene defect? Neuromuscul Disord. 2001 Jul;11(5):489-93. doi: 10.1016/s0960-8966(00)00221-2. PMID: 11404123.

Myasthenia

Barnes PR, Kanabar DJ, Brueton L, Newsom-Davis J, Huson SM, Mann NP, Hilton-Jones D. Neuromuscul Disord. 1995 Jan;5(1):59-65.

Brueton LA, Huson SM, Cox PM, Shirley I, Thompson EM, Barnes PR, Price J, Newsom-Davis J, Vincent A. Am J Med Genet. 2000 May 1;92(1):1-6. "

Cantagrel S, Maury L, Yamamoto AM, Maheut J, Toutain A, Castelnau P. Am J Perinatol. 2002 Aug;19(6):297-301.

Carr SR, Gilchrist JM, Abuelo DN, Clark D. Obstet Gynecol. 1991 Sep;78(3 Pt 2):485-9.

Chieza JT, Fleming I, Parry N, Skelton VA. Int J Obstet Anesth. 2011 Jan;20(1):79-82.

Dalton P, Clover L, Wallerstein R, Stewart H, Genzel-Boroviczeny O, Dean A, Vincent A. Neuromuscul Disord. 2006 Aug;16(8):481-91.

Dinger J, Prager B. Neuromuscul Disord. 1993 Jul;3(4):335-9. Review.

Dulitzky F, Sirota L, Landman J, Homburg R. Helv Paediatr Acta. 1987 Oct;42(2-3):173-6.

Eymard B, Morel E, Dulac O, Moutard-Codou ML, Jeannot E, Harpey JP, Rondot P, Bach JF. Rev Neurol (Paris). 1989;145(10):696-701. French.

Gilhus NE, Hong Y. Eur J Neurol. 2018 Dec;25(12):1402-1409.

Hacohen Y, Jacobson LW, Byrne S, Norwood F, Lall A, Robb S, Dilena R, Fumagalli M, Born AP, Clarke D, Lim M, Vincent A, Jungbluth H. Neurol Neuroimmunol Neuroinflamm. 2014 Dec 23;2(1):e57.

Hellmund A, Berg C, Geipel A, Müller A, Gembruch U. Arch Gynecol Obstet. 2016 Oct;294(4):697-707.

Hoff JM, Daltveit AK, Gilhus NE. Acta Neurol Scand Suppl.2006;183:26-7.

Hoff JM, Daltveit AK, Gilhus NE. Eur J Neurol. 2007 Jan;14(1):38-43.

Hoff JM, Loane M, Gilhus NE, Rasmussen S, Daltveit AK. Eur J Obstet Gynecol Reprod Biol. 2011 Dec;159(2):347-50.

Holmes LB, Driscoll SG, Bradley WG. J Pediatr. 1980 Jun;96(6):1067-9.

Mikou F, Kaouti N, Ghazli M, El Kerroumi M, Sefrioui O, Morsad F, Matar N, Elmoutawakil B, Mouden M, Gam I, Slassi I. J Gynecol Obstet Biol Reprod (Paris). 2003 Nov;32(7):660-2.

Morel E, Bach JF, Briard ML, Aubry JP. J Neuroimmunol. 1984 Aug;6(5):313-7.

Moutard-Codou ML, Delleur MM, Dulac O, Morel E, Voyer M, De Gamara E. Presse Med. 1987 Apr 11;16(13):615-8. French.

Pasternak JF, Hageman J, Adams MA, Philip AG, Gardner TH. J Pediatr. 1981 Oct;99(4):644-6.

Polizzi A, Huson SM, Vincent A. Teratology. 2000 Nov;62(5):332-41. Review.

Reimann J, Jacobson L, Vincent A, Kornblum C. Neurology. 2009 Nov 24;73(21):1806-8.

Riemersma S, Vincent A, Beeson D, Newland C, Hawke S, Vernet-der Garabedian B, Eymard B, Newsom-Davis J. J Clin Invest. 1996 Nov 15;98(10):2358-63.

Shepard MK. Birth Defects Orig Artic Ser. 1971 Feb;7(2):127.

Smit LM, Barth PG. Dev Med Child Neurol. 1980 Jun;22(3):371-4.

Stoll C, Ehret-Mentre MC, Treisser A, Tranchant C. Prenat Diagn. 1991 Jan;11(1):17-22. Review.

Tranchant C, Ehret C, Labouret P, Gasser B, Warter JM. Rev Neurol (Paris). 1991;147(1):62-4. Review. French.

Vincent A, McConville J, Farrugia ME, Bowen J, Plested P, Tang T, Evoli A, Matthews I, Sims G, Dalton P, Jacobson L, Polizzi A, Blaes F, Lang B, Beeson D, Willcox N, Newsom-Davis J, Hoch W. Ann N Y Acad Sci. 2003 Sep;998:324-35. Review.

Vincent A, Newland C, Brueton L, Beeson D, Riemersma S, Huson SM, Newsom-Davis J. Lancet. 1995 Jul 1;346(8966):24-5.
Vincent A, Waters P, Leite MI, Jacobson L, Koneczny I, Cossins J, Beeson D. Ann N Y Acad Sci. 2012 Dec;1274:92-8.

Aminoacidopathies

Kurolap A, Armbruster A, Hershkovitz T, Hauf K, Mory A, Paperna T, HannappelE, Tal G, Nijem Y, Sella E, Mahajnah M, Ilivitzki A, Hershkovitz D, Ekhilevitch, N, Mandel H, Eulenburg V, Baris HN. Am J Hum Genet. 2016 Nov 3;99(5):1172-1180.
Chiang MC, Huang SF, Hsueh C, Lai MW, Hou JW. Turk J Pediatr. 2008 Sep-Oct;50(5):492-4.
El-Hattab AW, Shaheen R, Hertecant J, Galadari HI, Albaqawi BS, Nabil A, Alkuraya FS. J Inherit Metab Dis. 2016 May;39(3):373-381.

Citric acid cycle

van Dijk T, Ferdinandusse S, Ruiter JPN, Alders M, Mathijssen IB, Parboosingh JS, Innes AM, Meijers-Heijboer H, Poll-The BT, Bernier FP, Wanders RJA, Lamont RE, Baas F. Eur J Hum Genet. 2018 Dec;26(12):1752-1758.
Winters L, Van Hoof E, De Catte L, Van Den Bogaert K, de Ravel T, De Waele L, Corveleyn A, Breckpot J. Eur J Paediatr Neurol. 2017 Sep;21(5):745-753.

Mitochondrial disorders

Piard J, Umanah GKE, Harms FL, Abalde-Atristain L, Amram D, Chang M, Chen R, Alawi M, Salpietro V, Rees MI, Chung SK, Houlden H, Verloes A, Dawson TM, Dawson VL, Van Maldergem L, Kutsche K. A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain. 2018 Mar 1;141(3):651-661. doi: 10.1093/brain/awx377. PMID: 29390050; PMCID: PMC5837721.Verloes A, Dawson TM, Dawson VL, Van Maldergem L, Kutsche K. Brain. 2018 Mar 1;141(3):651-661.
Sivaraman I, Friedman NR, Prayson RA. Muscle biopsy findings in a child with NALCN gene mutation. J Clin Neurosci. 2016 Dec;34:222-223. doi: 10.1016/j.jocn.2016.06.018. Epub 2016 Jul 26. PMID: 27473021.
Fukumura S, Ohba C, Watanabe T, Minagawa K, Shimura M, Murayama K, Ohtake A, Saitsu H, Matsumoto N, Tsutsumi H. Compound heterozygous GFM2 mutations with Leigh syndrome complicated by arthrogryposis multiplex congenita. J Hum Genet. 2015 Sep;60(9):509-13. doi: 10.1038/jhg.2015.57. Epub 2015 May 28. PMID: 26016410.
Ajit Bolar N, Vanlander AV, Wilbrecht C, Van der Aa N, Smet J, De Paepe B, Vandeweyer G, Kooy F, Eyskens F, De Latter E, Delanghe G, Govaert P, Leroy JG, Loeys B, Lill R, Van Laer L, Van Coster R. Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Hum Mol Genet. 2013 Jul 1;22(13):2590-602. doi: 10.1093/hmg/ddt107. Epub 2013 Mar 5. PMID: 23462291.Govaert P, Leroy JG, Loeys B, Lill R, Van Laer L, Van Coster R. Hum Mol Genet. 2013 Jul 1;22(13):2590-602.
Wilnai Y, Seaver LH, Enns GM. Atypical amyoplasia congenita in an infant with Leigh syndrome: a mitochondrial cause of severe contractures? Am J Med Genet A. 2012 Sep;158A(9):2353-7. doi: 10.1002/ajmg.a.35533. Epub 2012 Aug 7. PMID: 22887355.
Tulinius M, Oldfors A. Neonatal muscular manifestations in mitochondrial disorders. Semin Fetal Neonatal Med. 2011 Aug;16(4):229-35. doi: 10.1016/j.siny.2011.04.001. Epub 2011 May 18. PMID: 21596636.
Spiegel R, Khayat M, Shalev SA, Horovitz Y, Mandel H, Hershkovitz E, Barghuti F, Shaag A, Saada A, Korman SH, Elpeleg O, Yatsiv I. J Med Genet. 2011 Mar;48(3):177-82.
McPherson E, Zabel C. Mitochondrial mutation in a child with distal arthrogryposis. Am J Med Genet A. 2006 Jan 15;140(2):184-5. doi: 10.1002/ajmg.a.31041. PMID: 16353243.10.1002/ajmg.a.31041. PMID: 16353243.
von Kleist-Retzow JC, Cormier-Daire V, Viot G, Goldenberg A, Mardach B, Amiel J, Saada P, Dumez Y, Brunelle F, Saudubray JM, Chrétien D, Rötig A, Rustin P, Munnich A, De Lonlay P. Antenatal manifestations of mitochondrial respiratory chain deficiency. J Pediatr. 2003 Aug;143(2):208-12. doi: 10.1067/S0022-3476(03)00130-6. PMID: 12970634.
Gire C, Girard N, Nicaise C, Einaudi MA, Montfort MF, Dejode JM. Clinical features and neuroradiological findings of mitochondrial pathology in six neonates. Childs Nerv Syst. 2002 Nov;18(11):621-8. doi: 10.1007/s00381-002-0621-0. Epub 2002 Sep 12. PMID: 12420122.
Vielhaber S, Feistner H, Schneider W, Weis J, Kunz WS. Mitochondrial complex I deficiency in a female with multiplex arthrogryposis congenita. Pediatr Neurol. 2000 Jan;22(1):53-6. doi: 10.1016/s0887-8994(99)00097-1. PMID: 10669207.

Laubscher B, Janzer RC, Krähenbühl S, Hirt L, Deonna T. Ragged-red fibers and complex I deficiency in a neonate with arthrogryposis congenita. Pediatr Neurol. 1997 Oct;17(3):249-51. doi: 10.1016/s0887-8994(97)00082-9. PMID: 9390702.
Lee JS, Hwang JS, Ryu KH, Lee EH, Kim SH. Mitochondrial respiratory complex I deficiency simulating spinal muscular atrophy. Pediatr Neurol. 2007 Jan;36(1):45-7. doi: 10.1016/j.pediatrneurol.2006.07.007. PMID: 17162196.
Enns GM, Hoppel CL, DeArmond SJ, Schelley S, Bass N, Weisiger K, Horoupian D, Packman S. Relationship of primary mitochondrial respiratory chain dysfunction to fiber type abnormalities in skeletal muscle. Clin Genet. 2005 Oct;68(4):337-48. doi: 10.1111/j.1399-0004.2005.00499.x. PMID: 16143021.
De Meirleir L, Seneca S, Lissens W, De Clercq I, Eyskens F, Gerlo E, Smet J, Van Coster R. Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. J Med Genet. 2004 Feb;41(2):120-4. doi: 10.1136/jmg.2003.012047. PMID: 14757859; PMCID: PMC1735674.
de Koning TJ, de Vries LS, Groenendaal F, Ruitenbeek W, Jansen GH, Poll-The BT, Barth PG. Pontocerebellar hypoplasia associated with respiratory-chain defects. Neuropediatrics. 1999 Apr;30(2):93-5. doi: 10.1055/s-2007-973467. PMID: 10401692.

Peroxisomal disorders

Nakano K, Zhang Z, Shimozawa N, Kondo N, Ishii N, Funatsuka M, Shirakawa S, Itoh M, Takashima S, Une M, Kana-aki RR, Mukai K, Osawa M, Suzuki Y. J Pediatr. 2001 Dec;139(6):865-7.

Lysomsomal disorders

van der Beek J, Jonker C, van der Welle R, Liv N, Klumperman J. disease. J Cell Sci. 2019 May 15;132(10). pii: jcs189134.
BenHamida E, Ayadi I, Ouertani I, Chammem M, Bezzine A, BenTmime R, Attia L, Mrad R, Marrakchi Z. Pan Afr Med J. 2015 Jun 10;21:110.
Cullinane AR, Straatman-Iwanowska A, Zaucker A, Wakabayashi Y, Bruce CK, Luo G, Rahman F, Gürakan F, Utine E, Ozkan TB, Denecke J, Vukovic J, Di Rocco M, Mandel H, Cangul H, Matthews RP, Thomas SG, Rappoport JZ, Arias IM, Wolburg H, Knisely AS, Kelly DA, Müller F, Maher ER, Gissen P. Nat Genet. 2010 Apr;42(4):303-12. Erratum in: Nat Genet. 2011 Mar;43(3):277.
Gissen P, Johnson CA, Morgan NV, Stapelbroek JM, Forshew T, Cooper WN, McKiernan PJ, Klomp LW, Morris AA, Wraith JE, McClean P, Lynch SA, Thompson RJ, Lo B, Quarrell OW, Di Rocco M, Trembath RC, Mandel H, Wali S, Karet FE, Knisely AS, Houwen RH, Kelly DA, Maher ER. Nat Genet. 2004 Apr;36(4):400-4.
Mignot C, Gelot A, Bessières B, Daffos F, Voyer M, Menez F, Fallet Bianco C, Odent S, Le Duff D, Loget P, Fargier P, Costil J, Josset P, Roume J, Vanier MT, Maire I, Billette de Villemeur T. Am J Med Genet A. 2003 Jul 30;120A(3):338-44.

Metabolic diseases

Schorling DC, Rost S, Lefeber DJ, Brady L, Müller CR, Korinthenberg R, Tarnopolsky M, Bönnemann CG, Rodenburg RJ, Bugiani M, Beytia M, Krüger M, van der Knaap M, Kirschner J. Neurology. 2017 Aug 15;89(7):657-664.
Malfatti E, Barnerias C, Hedberg-Oldfors C, Gitiaux C, Benezit A, Oldfors A, Carlier RY, Quijano-Roy S, Romero NB. Neuromuscul Disord. 2016 Oct;26(10):681-687.
Hunter JM, Kiefer J, Balak CD, Jooma S, Ahearn ME, Hall JG, Baumbach-Reardon L. Am J Med Genet A. 2015 May;167A(5):931-73.
Wu PL, Yang YN, Tey SL, Yang CH, Yang SN, Lin CS. Pediatr Int. 2015 Aug;57(4):746-9.
Ganetzky R, Izumi K, Edmondson A, Muraresku CC, Zackai E, Deardorff M, Ganesh J. Am J Med Genet A. 2015 Oct;167A(10):2411-7.
Clayton PT, Grunewald S. J Inherit Metab Dis. 2009 Dec;32 Suppl 1:S137-9.
Ravenscroft G, Thompson EM, Todd EJ, Yau KS, Kresoje N, Sivadorai P, Friend K, Riley K, Manton ND, Blumbergs P, Fietz M, Duff RM, Davis MR, Allcock RJ, Laing NG. Neuromuscul Disord. 2013 Feb;23(2):165-9.
Tiemann C, Bührer C, Burwinkel B, Wirtenberger M, Hoehn T, Hübner C, van Landeghem FK, Stoltenburg G, Obladen M. Am J Med Genet A. 2005 Aug 30;137(2):125-9.
Shin YS, Plöchl E, Podskarbi T, Muss W, Pilz P, Puttinger R. J Inherit Metab Dis. 1994;17(1):153-5.

Congenital heart anomalies

Kane MS, Diamonstein CJ, Hauser N, Deeken JF, Niederhuber JE, Vilboux T. Genes Dis. 2019 Jan 7;6(1):56-67.

Gueneau L, Fish RJ, Shamseldin HE, Voisin N, Tran Mau-Them F, Preiksaitiene E, Monroe GR, Lai A, Putoux A, Allias F, Ambusaidi Q, Ambrozaityte L, Cimbalistienė L, Delafontaine J, Guex N, Hashem M, Kurdi W, Jamuar SS, Ying LJ, Bonnard C, Pippucci T, Pradervand S, Roechert B, van Hasselt PM, Wiederkehr M, Wright CF; DDD Study, Xenarios I, van Haaften G, Shaw-Smith C, Schindewolf EM, Neerman-ArbezM, Sanlaville D, Lesca G, Guibaud L, Reversade B, Chelly J, Kučinskas V, Alkuraya FS, Reymond A. Am J Hum Genet. 2018 Jan 4;102(1):116-132.
Stevenson RE, Vincent V, Spellicy CJ, Friez MJ, Chaubey A. Am J Med Genet A. 2018 Sep;176(9):1968-1971.
Quélin C, Loget P, Rozel C, D'Hervé D, Fradin M, Demurger F, Odent S, Pasquier L, Cavé H, Marcorelles P. Eur J Med Genet. 2017 Jul;60(7):395-398.
Verloes A, Di Donato N, Masliah-Planchon J, Jongmans M, Abdul-Raman OA, Albrecht B, Allanson J, Brunner H, Bertola D, Chassaing N, David A, Devriendt K, Eftekhari P, Drouin-Garraud V, Faravelli F, Faivre L, Giuliano F, Guion Almeida L, Juncos J, Kempers M, Eker HK, Lacombe D, Lin A, Mancini G, Melis D, Lourenço CM, Siu VM, Morin G, Nezarati M, Nowaczyk MJ, Ramer JC, Osimani S, Philip N, Pierpont ME, Procaccio V, Roseli ZS, Rossi M, Rusu C, Sznajer Y, Templin L, Uliana V, Klaus M, Van Bon B, Van Ravenswaaij C, Wainer B, Fry AE, Rump A, Hoischen A, Drunat S, Rivière JB, Dobyns WB, Pilz DT. Eur J Hum Genet. 2015 Mar;23(3):292-301.
Burkitt-Wright EM, Bradley L, Shorto J, McConnell VP, Gannon C, Firth HV, Park SM, D'Amore A, Munyard PF, Turnpenny PD, Charlton A, Wilson M, Kerr B. Am J Med Genet A. 2012 May;158A(5):1102-10.

Ophtalmological anomalies

Gueneau L, Fish RJ, Shamseldin HE, Voisin N, Tran Mau-Them F, Preiksaitiene E, Monroe GR, Lai A, Putoux A, Allias F, Ambusaidi Q, Ambrozaityte L, Cimbalistienė L, Delafontaine J, Guex N, Hashem M, Kurdi W, Jamuar SS, Ying LJ, Bonnard C, Pippucci T, Pradervand S, Roechert B, van Hasselt PM, Wiederkehr M, Wright CF; DDD Study, Xenarios I, van Haaften G, Shaw-Smith C, Schindewolf EM, Neerman-ArbezM, Sanlaville D, Lesca G, Guibaud L, Reversade B, Chelly J, Kučinskas V, Alkuraya FS, Reymond A. Am J Hum Genet. 2018 Jan 4;102(1):116-132.
Zin AA, Tsui I, Rossetto J, Vasconcelos Z, Adachi K, Valderramos S, Halai UA, Pone MVDS, Pone SM, Silveira Filho JCB, Aibe MS, da Costa ACC, Zin OA, Belfort R Jr, Brasil P, Nielsen-Saines K, Moreira MEL. JAMA Pediatr. 2017 Sep 1;171(9):847-854.
Meneses JDA, Ishigami AC, de Mello LM, de Albuquerque LL, de Brito CAA, Cordeiro MT, Pena LJ. Clin Infect Dis. 2017 May 15;64(10):1302-1308.
Khan AO, Shaheen R, Alkuraya FS. J AAPOS. 2014 Aug;18(4):362-7. Erratum in: J AAPOS. 2014 Oct;18(5):517.
Verloes A, Di Donato N, Masliah-Planchon J, Jongmans M, Abdul-Raman OA, Albrecht B, Allanson J, Brunner H, Bertola D, Chassaing N, David A, Devriendt K, Eftekhari P, Drouin-Garraud V, Faravelli F, Faivre L, Giuliano F, Guion Almeida L, Juncos J, Kempers M, Eker HK, Lacombe D, Lin A, Mancini G, Melis D, Lourenço CM, Siu VM, Morin G, Nezarati M, Nowaczyk MJ, Ramer JC, Osimani S, Philip N, Pierpont ME, Procaccio V, Roseli ZS, Rossi M, Rusu C, Sznajer Y, Templin L, Uliana V, Klaus M, Van Bon B, Van Ravenswaaij C, Wainer B, Fry AE, Rump A, Hoischen A, Drunat S, Rivière JB, Dobyns WB, Pilz DT. Eur J Hum Genet. 2015 Mar;23(3):292-301.
Rajab A, Hoffmann K, Ganesh A, Sethu AU, Mundlos S. Am J Med Genet A. 2005 Apr 15;134A(2):151-7
Brooks JG Jr, Coster DJ. Aust N Z J Ophthalmol. 1994 May;22(2):127-32

Tethered spinal cord

McGirt MJ, Mehta V, Garces-Ambrossi G, Gottfried O, Solakoglu C, Gokaslan ZL, Samdani A, Jallo GI. J Neurosurg Pediatr. 2009 Sep;4(3):270-4.

Neural tube defects

Naja AS, El Khatib H, Baydoun A, Nasser Eddine M. Am J Case Rep. 2019 May 20;20:719-722.
Weidman EK, Morgenstern PF, Phillips CD, Greenfield JP, Schwartz TH, Heier LA. Int J Pediatr Otorhinolaryngol. 2019 Feb;117:26-29.
Kowalczyk B, Feluś J. Arch Med Sci. 2016 Feb 1;12(1):10-24.
van Bosse HJ. Foot Ankle Clin. 2015 Dec;20(4):619-44.
Chen CP, Chang TY, Lin HH, Chern SR, Wang W. Taiwan J Obstet Gynecol. 2008 Mar;47(1):93-4.
Pungavkar SA, Sainani NI, Karnik AS, Mohanty PH, Lawande MA, Patkar DP, Sinha S. Korean J Radiol. 2007 Jul-Aug;8(4):351-5.
Luedemann WO, Tatagiba MS, Hussein S, Samii M. J Neurosurg. 2000 Jul;93(1 Suppl):130-2. Review
Podder S, Shepherd RC, Shillito P, Tolmie JL. Clin Dysmorphol. 1995 Jan;4(1):70-4.

Westcott MA, Dynes MC, Remer EM, Donaldson JS, Dias LS. Radiographics. 1992 Nov;12(6):1155-73. Review.

AMC + Anesthesia

Baines DB, Douglas ID, Overton JH. Anaesthesia for patients with arthrogryposis multiplex congenita: what is the risk of malignant hyperthermia? Anaesth Intensive Care. 1986 Nov;14(4):370–2.
Cruickshanks GF, Brown S, Chitayat D. Anesthesia for Freeman-Sheldon syndrome using a laryngeal mask airway. Can J Anaesth. 1999 Aug;46(8):783–7.
Gleich SJ, Tien M, Schroeder DR, Hanson AC, Flick R, Nemergut ME. Anesthetic Outcomes of Children With Arthrogryposis Syndromes: No Evidence of Hyperthermia. Anesth Analg. 2017;124(3):908–14.
Hopkins PM, Ellis FR, Halsall PJ. Hypermetabolism in arthrogryposis multiplex congenita. Anaesthesia. 1991 May;46(5):374–5.
Isaacson G, Drum ET. Difficult airway management in children and young adults with arthrogryposis. World J Otorhinolaryngol Head Neck Surg. 2018 Jun;4(2):122–5.
Jung JW, Heo BY, Oh EJ, Chung YH. Anesthesia in patients with arthrogryposis multiplex congenita: a report of 10 patients. Korean J Anesthesiol. 2014 Dec;67(Suppl):S89-90.
Ma L, Yu X. Arthrogryposis multiplex congenita: classification, diagnosis, perioperative care, and anesthesia. Front Med. 2017 Mar;11(1):48–52.
Oberoi GS, Kaul HL, Gill IS, Batra RK. Anaesthesia in arthrogryposis multiplex congenita: case report. Can J Anaesth. 1987 May;34(3 (Pt 1)):288–90.
Savenkov AN, Pajardi GE, Agranovich OE, Zabolskiy D, van Bosse HJP. Anaesthesiology for Children With Arthrogryposis. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S27–8.

AMC + Pain

Altiok H, Flanagan A, Krzak JJ, Hassani S. Quality of life, satisfaction with life, and functional mobility of young adults with arthrogryposis after leaving pediatric care. Am J Med Genet C Semin Med Genet. 2019;181(3):461–8.
Aroojis AJ, King MM, Donohoe M, Riddle EC, Kumar SJ. Congenital vertical talus in arthrogryposis and other contractural syndromes. Clin Orthop Relat Res. 2005 May;(434):26–32.
Cirillo A, Collins J, Sawatzky B, Hamdy R, Dahan-Oliel N. Pain among children and adults living with arthrogryposis multiplex congenita: A scoping review. Am J Med Genet C Semin Med Genet. 2019;181(3):436–53.
Dai S, Dieterich K, Jaeger M, Wuyam B, Jouk P-S, Pérennou D. Disability in adults with arthrogryposis is severe, partly invisible, and varies by genotype. Neurology. 2018 01;90(18):e1596–604.
Jones T, Miller R, Street JT, Sawatzky B. Validation of the Oswestry Disability Index for pain and disability in arthrogryposis multiplex congenita. Ann Phys Rehabil Med. 2019 Mar;62(2):92–7.
Kimber E, Tajsharghi H, Kroksmark A-K, Oldfors A, Tulinius M. Distal arthrogryposis: clinical and genetic findings. Acta Paediatr. 2012 Aug;101(8):877–87.
Nicomedez FPI, Li YH, Leong JCY. Tibiocalcaneal fusion after talectomy in arthrogrypotic patients. J Pediatr Orthop. 2003 Oct;23(5):654–7.
Spencer HT, Bowen RE, Caputo K, Green TA, Lawrence JF. Bone mineral density and functional measures in patients with arthrogryposis. J Pediatr Orthop. 2010 Aug;30(5):514–8.
van Bosse HJP. Orthopaedic care of the child with arthrogryposis: a 2020 overview. Curr Opin Pediatr. 2020 Feb;32(1):76–85.
Wall LB, Vuillerman C, Miller PE, Bae DS, Goldfarb CA, CoULD Study Group. Patient-reported Outcomes in Arthrogryposis. J Pediatr Orthop. 2020 Feb 7;

AMC + MPR

Braces

Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 1985 Sep;10(7):609–13.
Florence J. The orthotic management of arthrogryphosis. Prosthet Orthot Int. 1977 Aug;1(2):111–3.
Herron LD, Westin GW, Dawson EG. Scoliosis in arthrogryposis multiplex congenita. J Bone Joint Surg Am. 1978 Apr;60(3):293–9.
Komolkin I, Ulrich EV, Agranovich OE, van Bosse HJP. Treatment of Scoliosis Associated With Arthrogryposis Multiplex Congenita. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S24–6.

Siebold RM, Winter RB, Moe JH. The treatment of scoliosis in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1974;(103):191–8.
Södergård J, Ryöppy S. The knee in arthrogryposis multiplex congenita. J Pediatr Orthop. 1990 Apr;10(2):177–82.
Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.
Thomas B, Schopler S, Wood W, Oppenheim WL. The knee in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):87–92.
van Bosse HJP, Marangoz S, Lehman WB, Sala DA. Correction of arthrogrypotic clubfoot with a modified Ponseti technique. Clin Orthop Relat Res. 2009 May;467(5):1283–93.
Yingsakmongkol W, Kumar SJ. Scoliosis in arthrogryposis multiplex congenita: results after nonsurgical and surgical treatment. J Pediatr Orthop. 2000 Oct;20(5):656–61.

Casting

Bernstein RM. Arthrogryposis and amyoplasia. J Am Acad Orthop Surg. 2002 Dec;10(6):417–24.
Hahn G. Arthrogryposis. Pediatric review and habilitative aspects. Clin Orthop Relat Res. 1985 Apr;(194):104–14.
Pontén E. Management of the knees in arthrogryposis. J Child Orthop. 2015 Dec;9(6):465–72.
Smith DW, Drennan JC. Arthrogryposis wrist deformities: results of infantile serial casting. J Pediatr Orthop. 2002 Feb;22(1):44–7.
Thomas B, Schopler S, Wood W, Oppenheim WL. The knee in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):87–92.
van Bosse HJP. Orthopaedic care of the child with arthrogryposis: a 2020 overview. Curr Opin Pediatr. 2020 Feb;32(1):76–85.

Clinical care

Binkiewicz-Glińska A, Wierzba J, Szurowska E, Ruckeman-Dziurdzińska K, Bakuła S, Sokołów M, et al. Arthrogryposis multiplex congenital - multidisciplinary care - including own experience. Dev Period Med. 2016;20(3):191–6.
Bissinger RL, Koch FR. Nonlethal multiple pterygium syndrome: Escobar syndrome. Adv Neonatal Care. 2014 Feb;14(1):24–9.
Dahan-Oliel N, Bedard T, Darsaklis VB, Hall JG, van Bosse HJP, Hamdy RC. Development of a research platform for children with arthrogryposis multiplex congenita: study protocol for a pilot registry. BMJ Open. 2018 30;8(6):e021377.
Dieterich K, Kimber E, Hall JG. Central nervous system involvement in arthrogryposis multiplex congenita: Overview of causes, diagnosis, and care. Am J Med Genet C Semin Med Genet. 2019;181(3):345–53.
Valdés-Flores M, Casas-Avila L, Hernández-Zamora E, Kofman S, Hidalgo-Bravo A. Characterization of a group unrelated patients with arthrogryposis multiplex congenita. J Pediatr (Rio J). 2016 Feb;92(1):58–64.
Villard L, Nordmark-Andersson E, Crowley B, Straub V, Bertoli M. Multidisciplinary Clinics. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S29–30.
Williams MS, Elliott CG, Bamshad MJ. Pulmonary disease is a component of distal arthrogryposis type 5. Am J Med Genet A. 2007 Apr 1;143A(7):752–6.

Gait analysis

Bartonek Å. The use of orthoses and gait analysis in children with AMC. J Child Orthop. 2015 Dec;9(6):437–47.
Bjornson KF, Song K, Zhou C, Coleman K, Myaing M, Robinson SL. Walking stride rate patterns in children and youth. Pediatr Phys Ther. 2011;23(4):354–63.
Eriksson M, Bartonek Å, Pontén E, Gutierrez-Farewik EM. Gait dynamics in the wide spectrum of children with arthrogryposis: a descriptive study. BMC Musculoskelet Disord. 2015 Dec 9;16:384.
Eriksson M, Villard L, Bartonek A. Walking, orthoses and physical effort in a Swedish population with arthrogryposis. J Child Orthop. 2014 Aug;8(4):305–12.
Rodriguez LM, Bickley C, Russo S, Barnes D, Gagnon M, Hamdy R, et al. Perspectives on gait and motion analysis in the management of youth with arthrogryposis multiplex congenita. Am J Med Genet C Semin Med Genet. 2019;181(3):404–9.

Orthoses

Bartonek A, Lidbeck CM, Pettersson R, Weidenhielm EB, Eriksson M, Gutierrez-Farewik E. Influence of heel lifts during standing in children with motor disorders. Gait Posture. 2011 Jul;34(3):426–31.
Eriksson M, Gutierrez-Farewik EM, Broström E, Bartonek A. Gait in children with arthrogryposis multiplex congenita. J Child Orthop. 2010 Feb;4(1):21–31.
Eriksson M, Jylli L, Villard L, Kroksmark A-K, Bartonek Å. Health-related quality of life and orthosis use in a Swedish population with arthrogryposis. Prosthet Orthot Int. 2018 Aug;42(4):402–9.
Fucs PMMB, Svartman C, de Assumpção RMC, Lima Verde SR. Quadricepsplasty in arthrogryposis (amyoplasia): long-term follow-up. J Pediatr Orthop B. 2005 May;14(3):219–24.

Outcome + medical – orthopaedic

Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of arthrogryposis multiplex congenita with reference to the neurogenic variant. Eur J Paediatr Neurol. 2011 Jul;15(4):316–9.
Carlson WO, Speck GJ, Vicari V, Wenger DR. Arthrogryposis multiplex congenita. A long-term follow-up study. Clin Orthop Relat Res. 1985 Apr;(194):115–23.
Donohoe M, Pruszcynski B, Rogers K, Bowen JR. Predicting Ambulatory Function Based on Infantile Lower Extremity Posture Types in Amyoplasia Arthrogryposis. J Pediatr Orthop. 2019 Aug;39(7):e531–5.
Dubousset J, Guillaumat M. Long-term outcome for patients with arthrogryposis multiplex congenita. J Child Orthop. 2015 Dec;9(6):449–58.
Ho CA, Karol LA. The utility of knee releases in arthrogryposis. J Pediatr Orthop. 2008 May;28(3):307–13.
Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.
Nouraei H, Sawatzky B, MacGillivray M, Hall J. Long-term functional and mobility outcomes for individuals with arthrogryposis multiplex congenita. Am J Med Genet A. 2017 May;173(5):1270–8.
Sawatzky B, Jones T, Miller R, Noureai H. The relationship between joint surgery and quality of life in adults with arthrogryposis: An international study. Am J Med Genet C Semin Med Genet. 2019;181(3):469–73.
Södergård J, Hakamies-Blomqvist L, Sainio K, Ryöppy S, Vuorinen R. Arthrogryposis multiplex congenita: perinatal and electromyographic findings, disability, and psychosocial outcome. J Pediatr Orthop B. 1997 Jul;6(3):167–71.
Wall LB, Vuillerman C, Miller PE, Bae DS, Goldfarb CA, CoULD Study Group. Patient-reported Outcomes in Arthrogryposis. J Pediatr Orthop. 2020 Feb 7;

Physiotherapy

Hamdy RC, van Bosse H, Altiok H, Abu-Dalu K, Kotlarsky P, Fafara A, et al. Treatment and outcomes of arthrogryposis in the lower extremity. Am J Med Genet C Semin Med Genet. 2019;181(3):372–84.
Murray C, Fixsen JA. Management of knee deformity in classical arthrogryposis multiplex congenita (amyoplasia congenita). J Pediatr Orthop B. 1997 Jul;6(3):186–91.
Palmer PM, MacEwen GD, Bowen JR, Mathews PA. Passive motion therapy for infants with arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):54–9.
Samekova H, Bialik V, M Bialik G. Arthrogryposis multiplex congenita - local experience. Ortop Traumatol Rehabil. 2006 Feb 28;8(1):69–73.
Thompson GH, Bilenker RM. Comprehensive management of arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):6–14.

Scoliosis + medical

Li Y, Sheng F, Xia C, Xu L, Qiu Y, Zhu Z. Risk Factors of Impaired Pulmonary Function in Arthrogryposis Multiplex Congenital Patients With Concomitant Scoliosis: A Comparison With Adolescent Idiopathic Scoliosis. Spine. 2018 15;43(8):E456–60.

Treatment + medical – orthopaedic

Bernstein RM. Arthrogryposis and amyoplasia. J Am Acad Orthop Surg. 2002 Dec;10(6):417–24.
Dieterich K, Kimber E, Hall JG. Central nervous system involvement in arthrogryposis multiplex congenita: Overview of causes, diagnosis, and care. Am J Med Genet C Semin Med Genet. 2019;181(3):345–53.
Dillon ER, Bjornson KF, Jaffe KM, Hall JG, Song K. Ambulatory activity in youth with arthrogryposis: a cohort study. J Pediatr Orthop. 2009 Mar;29(2):214–7.
Valdés-Flores M, Casas-Avila L, Hernández-Zamora E, Kofman S, Hidalgo-Bravo A. Characterization of a group unrelated patients with arthrogryposis multiplex congenita. J Pediatr (Rio J). 2016 Feb;92(1):58–64.

Wagner LV, Cherry JS, Sawatzky BJ, Fąfara A, Elfassy C, Eriksson M, et al. Rehabilitation across the lifespan for individuals with arthrogryposis. Am J Med Genet C Semin Med Genet. 2019;181(3):385–92.

Treatment + recommendation

Elfassy C, Darsaklis VB, Snider L, Gagnon C, Hamdy R, Dahan-Oliel N. Rehabilitation needs of youth with arthrogryposis multiplex congenita: Perspectives from key stakeholders. Disabil Rehabil. 2019 Feb 11;1–7.
Funk JF, Lebek S, Seidl T, Placzek R. [Comparison of treatment results of idiopathic and non-idiopathic congenital clubfoot : prospective evaluation of the Ponseti therapy]. Orthopade. 2012 Dec;41(12):977–83.
Guerra-Jasso JJ, Valcarce-León JA, Quíntela-Núñez-Del Prado HM. [Evidence level and grade of recommendation of the Ponseti method for arthrogryposis-related syndromic talipes equinovarus and Moebius syndrome: a systematic review]. Acta Ortop Mex. 2017 Aug;31(4):182–8.
Kroksmark A-K, Kimber E, Jerre R, Beckung E, Tulinius M. Muscle involvement and motor function in amyoplasia. Am J Med Genet A. 2006 Aug 15;140(16):1757–67.
Poling MI, Morales Corado JA, Chamberlain RL. Findings, phenotypes, and outcomes in Freeman-Sheldon and Sheldon-Hall syndromes and distal arthrogryposis types 1 and 3: protocol for systematic review and patient-level data meta-analysis. Syst Rev. 2017 06;6(1):46.

Weelchair

De Souza LH, Frank AO. Rare diseases: matching wheelchair users with rare metabolic, neuromuscular or neurological disorders to electric powered indoor/outdoor wheelchairs (EPIOCs). Disabil Rehabil. 2016 Aug;38(16):1547-56. doi: 10.3109/09638288.2015.1106599. Epub 2015 Dec 30. PMID: 26714619; PMCID: PMC4926775.

AMC + Surgery

Scoliosis

Astur N, Flynn JM, Flynn JM, Ramirez N, Glotzbecker M, van Bosse HJ, et al. The efficacy of rib-based distraction with VEPTR in the treatment of early-onset scoliosis in patients with arthrogryposis. J Pediatr Orthop. 2014 Jan;34(1):8–13.
Fletcher ND, Rathjen KE, Bush P, Ezaki M. Asymmetrical arthrogryposis of the upper extremity associated with congenital spine anomalies. J Pediatr Orthop. 2010 Dec;30(8):936–41.
Herron LD, Westin GW, Dawson EG. Scoliosis in arthrogryposis multiplex congenita. J Bone Joint Surg Am. 1978 Apr;60(3):293–9.
Paul JC, Lonner BS, Vira S, Feldman D, Errico TJ. Does Reoperation Risk Vary for Different Types of Pediatric Scoliosis? J Pediatr Orthop. 2018 Oct;38(9):459–64.
Yingsakmongkol W, Kumar SJ. Scoliosis in arthrogryposis multiplex congenita: results after nonsurgical and surgical treatment. J Pediatr Orthop. 2000 Oct;20(5):656–61.

Achilles tendon release

Adelaar RS, Williams RM, Gould JS. Congenital convex pes valgus: results of an early comprehensive release and a review of congenital vertical talus at Richmond Crippled Children's Hospital and the University of Alabama in Birmingham. Foot Ankle. 1980 Sep;1(2):62–73.
Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, et al. [Results of surgical treatment of congenital convex pes valgus (10 non-idiopathic feet)]. Rev Chir Orthop Reparatrice Appar Mot. 2008a Apr;94(2):128–34.
Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, et al. Results of surgical treatment of congenital convex pes valgus (ten nonidiopathic feet). Rev Chir Orthop Reparatrice Appar Mot. 2008b Dec;94(8):e28-34.
Chang CH, Huang SC. Surgical treatment of clubfoot deformity in arthrogryposis multiplex congenita. J Formos Med Assoc. 1997 Jan;96(1):30–5.
Guidera KJ, Drennan JC. Foot and ankle deformities in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):93–8.
Ramanoudjame M, Loriaut P, Seringe R, Glorion C, Wicart P. The surgical treatment of children with congenital convex foot (vertical talus): evaluation of midtarsal surgical release and open reduction. Bone Joint J. 2014 Jun;96-B(6):837–44.

Schwering L. Surgical correction of the true vertical talus deformity. Oper Orthop Traumatol. 2005 Jun;17(2):211–31.
Zimbler S, Craig CL. The arthrogrypotic foot plan of management and results of treatment. Foot Ankle. 1983 Feb;3(4):211–9.

Adducted thumb

Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794–8.
Rodríguez RN, Capdevila-Leonori R, Nualart-Hernández L. [Opening of the first web space in patients with multiple congenital arthrogryposis and adducted thumb with a dorsoradial index finger flap]. Acta Ortop Mex. 2014 Feb;28(1):23–7.

Anaesthesia

Gleich SJ, Tien M, Schroeder DR, Hanson AC, Flick R, Nemergut ME. Anesthetic Outcomes of Children With Arthrogryposis Syndromes: No Evidence of Hyperthermia. Anesth Analg. 2017;124(3):908–14.
Hopkins PM, Ellis FR, Halsall PJ. Hypermetabolism in arthrogryposis multiplex congenita. Anaesthesia. 1991 May;46(5):374–5.
Ion T, Cook-Sather SD, Finkel RS, Cucchiaro G. Fascia iliaca block for an infant with arthrogryposis multiplex congenita undergoing muscle biopsy. Anesth Analg. 2005 Jan;100(1):82–4.
Isaacson G, Drum ET. Difficult airway management in children and young adults with arthrogryposis. World J Otorhinolaryngol Head Neck Surg. 2018 Jun;4(2):122–5.
Nguyen NH, Morvant EM, Mayhew JF. Anesthetic management for patients with arthrogryposis multiplex congenita and severe micrognathia: case reports. J Clin Anesth. 2000 May;12(3):227–30.
Oberoi GS, Kaul HL, Gill IS, Batra RK. Anaesthesia in arthrogryposis multiplex congenita: case report. Can J Anaesth. 1987 May;34(3 (Pt 1)):288–90.
Ponde V, Desai AP, Shah D. Comparison of success rate of ultrasound-guided sciatic and femoral nerve block and neurostimulation in children with arthrogryposis multiplex congenita: a randomized clinical trial. Paediatr Anaesth. 2013 Jan;23(1):74–8.
Rozkowski A, Smyczek D, Birnbach DJ. Continuous spinal anesthesia for cesarean delivery in a patient with arthrogryposis multiplex congenita. A clinical report. Reg Anesth. 1996 Oct;21(5):477–9.
Savenkov AN, Pajardi GE, Agranovich OE, Zabolskiy D, van Bosse HJP. Anaesthesiology for Children With Arthrogryposis. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S27–8.
Sivaci R, Balci C, Maralcan G, Kuru I. Management of difficult airway in a child with arthrogryposis multiplex congenita during general anesthesia. Saudi Med J. 2005 Oct;26(10):1657–9.
Szmuk P, Ezri T, Warters DR, Katz J. Anesthetic management of a patient with arthrogryposis multiplex congenita and limited mouth opening. J Clin Anesth. 2001 Feb;13(1):59–60.

Club feet

Al Kaissi A, Kenis V, Melchenko E, Chehida FB, Ganger R, Klaushofer K, et al. Corrections of lower limb deformities in patients with diastrophic dysplasia. Orthop Surg. 2014 Nov;6(4):274–9.
Chang CH, Huang SC. Surgical treatment of clubfoot deformity in arthrogryposis multiplex congenita. J Formos Med Assoc. 1997 Jan;96(1):30–5.
Choi IH, Yang MS, Chung CY, Cho TJ, Sohn YJ. The treatment of recurrent arthrogrypotic club foot in children by the Ilizarov method. A preliminary report. J Bone Joint Surg Br. 2001 Jul;83(5):731–7.
Eidelman M, Katzman A. Treatment of arthrogrypotic foot deformities with the Taylor Spatial Frame. J Pediatr Orthop. 2011 Jun;31(4):429–34.
Eulert J. [Clinical aspects and treatment of arthrogryposis multiplex congenita. Lower extremity]. Z Orthop Ihre Grenzgeb. 1984 Oct;122(5):661–9.
Gibson DA, Urs ND. Arthrogryposis multiplex congenita. J Bone Joint Surg Br. 1970 Aug;52(3):483–93.
Iskandar HN, Bishay SNG, Sharaf-El-Deen HA-R, El-Sayed MM. Tarsal decancellation in the residual resistant arthrogrypotic clubfoot. Ann R Coll Surg Engl. 2011 Mar;93(2):139–45.
Lichtblau S. Choices in surgical treatment of rigid neurogenic and arthrogrypotic clubfeet. Mt Sinai J Med. 1989 Jan;56(1):17–22.
Machida J, Inaba Y, Nakamura N. Management of foot deformity in children. J Orthop Sci. 2017 Mar;22(2):175–83.
Niki H, Staheli LT, Mosca VS. Management of clubfoot deformity in amyoplasia. J Pediatr Orthop. 1997 Dec;17(6):803–7.

Popihn H. [Contribution to the surgical treatment of congenital multiple arthrogryposis of the lowe extremity]. Beitr Orthop Traumatol. 1980 Oct;27(10):580–4.
Samekova H, Bialik V, M Bialik G. Arthrogryposis multiplex congenita - local experience. Ortop Traumatol Rehabil. 2006 Feb 28;8(1):69–73.
Segal LS, Mann DC, Feiwell E, Hoffer MM. Equinovarus deformity in arthrogryposis and myelomeningocele: evaluation of primary talectomy. Foot Ankle. 1989 Aug;10(1):12–6.
Södergård J, Ryöppy S. Foot deformities in arthrogryposis multiplex congenita. J Pediatr Orthop. 1994 Dec;14(6):768–72.
Somppi E, Sulamaa M. Early operative treatment of congenital club foot. Acta Orthop Scand. 1971;42(6):513–20.
Spires TD, Gross RH, Low W, Barringer W. Management of the resistant myelodysplastic or arthrogrypotic clubfoot with the Verebelyi-Ogston procedure. J Pediatr Orthop. 1984 Nov;4(6):705–10.
Stauffer RN, Nelson GE, Bianco AJ. Calcaneal osteotomy in the treatment of the cavovarus foot. Mayo Clin Proc. 1970 Sep;45(9):624–35.
van Bosse HJP. Challenging clubfeet: the arthrogrypotic clubfoot and the complex clubfoot. J Child Orthop. 2019 Jun 1;13(3):271–81.
Widmann RF, Do TT, Burke SW. Radical soft-tissue release of the arthrogrypotic clubfoot. J Pediatr Orthop B. 2005 Mar;14(2):111–5.

Elbow

Andrisano A, Manfrini M, Zucchi M, Mignani G. Arthromyolysis of the elbow in arthrogryposis. Ital J Orthop Traumatol. 1988 Jun;14(2):239–42.
Atkins RM, Bell MJ, Sharrard WJ. Pectoralis major transfer for paralysis of elbow flexion in children. J Bone Joint Surg Br. 1985 Aug;67(4):640–4.
Axt MW, Niethard FU, Döderlein L, Weber M. Principles of treatment of the upper extremity in arthrogryposis multiplex congenita type I. J Pediatr Orthop B. 1997 Jul;6(3):179–85.
Bayne LG. Hand assessment and management of arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):68–73.
Bennett JB, Hansen PE, Granberry WM, Cain TE. Surgical management of arthrogryposis in the upper extremity. J Pediatr Orthop. 1985 Jun;5(3):281–6.
Cao J, Stutz C, Beckwith T, Browning A, Mills J, Oishi SN. Elbow Release and Tricepsplasty in Arthrogrypotic Patients: A Long-Term Follow-Up Study. J Hand Surg Am. 2020 Jun;45(6):549.e1-549.e7.
Carroll RE, Hill NA. Triceps transfer to restore elbow flexion. A study of fifteen patients with paralytic lesions and arthrogryposis. J Bone Joint Surg Am. 1970 Mar;52(2):239–44.
Chomiak J, Dungl P, Včelák J. Reconstruction of elbow flexion in arthrogryposis multiplex congenita type I: results of transfer of pectoralis major muscle with follow-up at skeletal maturity. J Pediatr Orthop. 2014 Dec;34(8):799–807.
Doyle JR, James PM, Larsen LJ, Ashley RK. Restoration of elbow flexion in arthrogryposis multiplex congenita. J Hand Surg Am. 1980 Mar;5(2):149–52.
Ezaki M. Treatment of the upper limb in the child with arthrogryposis. Hand Clin. 2000 Nov;16(4):703–11.
Frizzell K, Kozin SH, Zlotolow DA. Bipolar Latissimus Dorsi Transfer for Arthrogryposis Multiplex Congenita: Minimum 10-Month Follow-Up. J Hand Surg Am. 2020 Nov;45(11):1084.e1-1084.e7.
Gagnon E, Fogelson N, Seyfer AE. Use of the latissimus dorsi muscle to restore elbow flexion in arthrogryposis. Plast Reconstr Surg. 2000 Dec;106(7):1582–5.
Gogola GR, Ezaki M, Oishi SN, Gharbaoui I, Bennett JB. Long head of the triceps muscle transfer for active elbow flexion in arthrogryposis. Tech Hand Up Extrem Surg. 2010 Jun;14(2):121–4.
Goldfarb CA, Burke MS, Strecker WB, Manske PR. The Steindler flexorplasty for the arthrogrypotic elbow. J Hand Surg Am. 2004 May;29(3):462–9.
James MA, Millar KL, Manske MC, Van Heest AE. Posterior Elbow Capsulotomy and Triceps Lengthening for Elbow Extension Contracture in Children with Arthrogryposis Multiplex Congenita. JBJS Essent Surg Tech. 2020 Mar;10(1).
Lahoti O, Bell MJ. Transfer of pectoralis major in arthrogryposis to restore elbow flexion: deteriorating results in the long term. J Bone Joint Surg Br. 2005 Jun;87(6):858–60.
Mailänder P, Machens HG, Rieck B, Wittig K, Berger A. [Bilateral biceps replacement-plasty by transposition of the latissimus dorsi muscle in arthrogryposis multiplex congenita. A case report]. Handchir Mikrochir Plast Chir. 1996 Mar;28(2):59–63.
Martin G, Perrot P, Duteille F. [Treatment of congenital deficit of elbow flexion in a case of arthrogryposis]. Chir Main. 2009 Apr;28(2):116–9.
Martini AK, Banniza von Bazan U. [Steindler-plasty in paralysis of the elbow flexor muscles]. Handchir Mikrochir Plast Chir. 1985 Jan;17(1):23–6.

Mennen U. Early corrective surgery of the wrist and elbow in arthrogryposis multiplex congenita. J Hand Surg Br. 1993 Jun;18(3):304–7.
Meyn M, Ruby L. Arthrogryposis of the upper extremity. Orthop Clin North Am. 1976 Apr;7(2):501–9.
Oishi SN, Agranovich O, Pajardi GE, Novelli C, Baindurashvili AG, Trofimova SI, et al. Treatment of the Upper Extremity Contracture/Deformities. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S9–15.
Parsch K, Pietrzak S. [Congenital multiple arthrogryposis]. Orthopade. 2007 Mar;36(3):281–90; quiz 291.
Puchała J, Sułko J, Radło W. [Surgical treatment of upper extremities of cases arthrogryposis multiplex congenita]. Chir Narzadow Ruchu Ortop Pol. 2001;66(2):143–9.
Ramirez RN, Richards CJ, Kozin SH, Zlotolow DA. Combined Elbow Release and Humeral Rotational Osteotomy in Arthrogryposis. J Hand Surg Am. 2017 Nov;42(11):926.e1-926.e9.
Richards C, Ramirez R, Kozin S, Zlotolow D. The Effects of Age on the Outcomes of Elbow Release in Arthrogryposis. J Hand Surg Am. 2019 Oct;44(10):898.e1-898.e6.
Sawatzky B, Jones T, Miller R, Noureai H. The relationship between joint surgery and quality of life in adults with arthrogryposis: An international study. Am J Med Genet C Semin Med Genet. 2019;181(3):469–73.
Takagi T, Seki A, Kobayashi Y, Mochida J, Takayama S. Isolated Muscle Transfer to Restore Elbow Flexion in Children with Arthrogryposis. J Hand Surg Asian Pac Vol. 2016;21(1):44–8.
Van Heest A, James MA, Lewica A, Anderson KA. Posterior elbow capsulotomy with triceps lengthening for treatment of elbow extension contracture in children with arthrogryposis. J Bone Joint Surg Am. 2008 Jul;90(7):1517–23.
Van Heest A, Waters PM, Simmons BP. Surgical treatment of arthrogryposis of the elbow. J Hand Surg Am. 1998 Nov;23(6):1063–70.
Van Heest A, James MA, Lewica A, Anderson KA. Posterior elbow capsulotomy with triceps lengthening for treatment of elbow extension contracture in children with arthrogryposis. J Bone Joint Surg Am. 2008 Jul;90(7):1517–23.
Williams PF. The elbow in arthrogryposis. J Bone Joint Surg Br. 1973 Nov;55(4):834–40.
Williams PF. Management of upper limb problems in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):60–7.
Zargarbashi R, Nabian MH, Werthel J-D, Valenti P. Is bipolar latissimus dorsi transfer a reliable option to restore elbow flexion in children with arthrogryposis? A review of 13 tendon transfers. J Shoulder Elbow Surg. 2017 Nov;26(11):2004–9.
Zlotolow DA, Kozin SH. Posterior elbow release and humeral osteotomy for patients with arthrogryposis. J Hand Surg Am. 2012 May;37(5):1078–82.

Expert review recommendations

Campbell RM. Spine deformities in rare congenital syndromes: clinical issues. Spine. 2009 Aug 1;34(17):1815–27.
Eulert J. [Clinical aspects and treatment of arthrogryposis multiplex congenita. Lower extremity]. Z Orthop Ihre Grenzgeb. 1984 Oct;122(5):661–9.
Ezaki M. Treatment of the upper limb in the child with arthrogryposis. Hand Clin. 2000 Nov;16(4):703–11.
Hamdy RC, van Bosse H, Altiok H, Abu-Dalu K, Kotlarsky P, Fafara A, et al. Treatment and outcomes of arthrogryposis in the lower extremity. Am J Med Genet C Semin Med Genet. 2019;181(3):372–84.
James MA, Millar KL, Manske MC, Van Heest AE. Posterior Elbow Capsulotomy and Triceps Lengthening for Elbow Extension Contracture in Children with Arthrogryposis Multiplex Congenita. JBJS Essent Surg Tech. 2020 Mar;10(1).
Komolkin I, Ulrich EV, Agranovich OE, van Bosse HJP. Treatment of Scoliosis Associated With Arthrogryposis Multiplex Congenita. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S24–6.
Meyn M, Ruby L. Arthrogryposis of the upper extremity. Orthop Clin North Am. 1976 Apr;7(2):501–9.
Oishi SN, Agranovich O, Pajardi GE, Novelli C, Baindurashvili AG, Trofimova SI, et al. Treatment of the Upper Extremity Contracture/Deformities. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S9–15.
Parsch K, Pietrzak S. [Congenital multiple arthrogryposis]. Orthopade. 2007a Mar;36(3):281–90; quiz 291.
Parsch K, Pietrzak S. [Congenital multiple arthrogryposis]. Orthopade. 2007b Mar;36(3):281–90; quiz 291.
Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.
van Bosse HJP. Orthopaedic care of the child with arthrogryposis: a 2020 overview. Curr Opin Pediatr. 2020;32(1):76–85.
van Bosse HJP, Pontén E, Wada A, Agranovich OE, Kowalczyk B, Lebel E, et al. Treatment of the Lower Extremity Contracture/Deformities. J Pediatr Orthop. 2017a Aug;37 Suppl 1:S16–23.
Williams P. The management of arthrogryposis. Orthop Clin North Am. 1978 Jan;9(1):67–88.

Hip

Akazawa H, Oda K, Mitani S, Yoshitaka T, Asaumi K, Inoue H. Surgical management of hip dislocation in children with arthrogryposis multiplex congenita. J Bone Joint Surg Br. 1998 Jul;80(4):636–40.
Aydin BK, Yilmaz G, Senaran H, Durgut F. Short-term results of early (before 6 months) open reduction of dislocated hips in arthrogryposis multiplex congenita. J Pediatr Orthop B. 2016 Nov;25(6):509–13.
Bradish C. The hip in arthrogryposis. J Child Orthop. 2015 Dec;9(6):459–63.
Catterall A. Congenital dislocation of the hip: the indications and technique of open reduction. Acta Orthop Belg. 1990;56(1 Pt B):229–31.
Dalton DM, Magill P, Mulhall KJ. Bilateral total hip replacement in arthrogryposis multiplex congenita. BMJ Case Rep. 2015 Nov 25;2015.
Eulert J. [Clinical aspects and treatment of arthrogryposis multiplex congenita. Lower extremity]. Z Orthop Ihre Grenzgeb. 1984 Oct;122(5):661–9.
Grill F. [The hip joint in arthrogryposis]. Z Orthop Ihre Grenzgeb. 1990 Aug;128(4):384–90.
Huurman WW, Jacobsen ST. The hip in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):81–6.
Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.
Huurman WW, Jacobsen ST. The hip in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):81–6.
Radło W, Miklaszewski K, Feluś J, Sułko J. [Reconstructions of the teratogenic hip dislocation in childrens with AMC]. Chir Narzadow Ruchu Ortop Pol. 2007 Feb;72(1):9–13.
Rocha LEM da, Nishimori FK, Figueiredo DC de, Grimm DH, Cunha LAM da. OPEN REDUCTION OF HIP DISLOCATION IN PATIENTS WITH ARTHROGRYPOSIS MULTIPLEX CONGENITA - AN ANTEROMEDIAL APPROACH. Rev Bras Ortop. 2010 Oct;45(5):403–8.
Samekova H, Bialik V, M Bialik G. Arthrogryposis multiplex congenita - local experience. Ortop Traumatol Rehabil. 2006 Feb 28;8(1):69–73.
Södergard J. [Hip in arthrogryposis multiplex congenita]. Rev Chir Orthop Reparatrice Appar Mot. 1996;82(5):403–9.
St Clair HS, Zimbler S. A plan of management and treatment results in the arthrogrypotic hip. Clin Orthop Relat Res. 1985 Apr;(194):74–80.
Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop. 1987 Dec;7(6):681–5.
Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.
Szöke G, Staheli LT, Jaffe K, Hall JG. Medial-approach open reduction of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop. 1996 Feb;16(1):127–30.
van Bosse HJP, Saldana RE. Reorientational Proximal Femoral Osteotomies for Arthrogrypotic Hip Contractures. J Bone Joint Surg Am. 2017 Jan 4;99(1):55–64.
Wada A, Yamaguchi T, Nakamura T, Yanagida H, Takamura K, Oketani Y, et al. Surgical treatment of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop B. 2012 Sep;21(5):381–5.
Yau PWP, Chow W, Li YH, Leong JCY. Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. J Pediatr Orthop. 2002 Jun;22(3):359–63.

Hip dislocation

Akazawa H, Oda K, Mitani S, Yoshitaka T, Asaumi K, Inoue H. Surgical management of hip dislocation in children with arthrogryposis multiplex congenita. J Bone Joint Surg Br. 1998 Jul;80(4):636–40.
Aydin BK, Yilmaz G, Senaran H, Durgut F. Short-term results of early (before 6 months) open reduction of dislocated hips in arthrogryposis multiplex congenita. J Pediatr Orthop B. 2016 Nov;25(6):509–13.
Miao M, Cai H, Wang Z, Hu L, Bian J, Cai H. Early open reduction of dislocated hips using a modified Smith-Petersen approach in arthrogyposis multiplex congenita. BMC Musculoskelet Disord. 2020 Mar 4;21(1):144.
Rocha LEM da, Nishimori FK, Figueiredo DC de, Grimm DH, Cunha LAM da. OPEN REDUCTION OF HIP DISLOCATION IN PATIENTS WITH ARTHROGRYPOSIS MULTIPLEX CONGENITA - AN ANTEROMEDIAL APPROACH. Rev Bras Ortop. 2010 Oct;45(5):403–8.
Rombouts JJ, Rossillon R. Teratologic dislocation of the hip: review of a series of 17 cases. Acta Orthop Belg. 1990;56(1 Pt A):181–9.
Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop. 1987 Dec;7(6):681–5.
Szöke G, Staheli LT, Jaffe K, Hall JG. Medial-approach open reduction of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop. 1996 Feb;16(1):127–30.
Wada A, Yamaguchi T, Nakamura T, Yanagida H, Takamura K, Oketani Y, et al. Surgical treatment of hip dislocation in amyoplasia-type arthrogryposis. J Pediatr Orthop B. 2012 Sep;21(5):381–5.

Yau PWP, Chow W, Li YH, Leong JCY. Twenty-year follow-up of hip problems in arthrogryposis multiplex congenita. J Pediatr Orthop. 2002 Jun;22(3):359–63.

Knee

Auñón-Martín I, Jiménez-Díaz V, Zorrilla-Sánchez De Neira J, Capel-Agúndez A. [Femur fracture in a patient with dog-leg deformity secondary to arthrogryposis. Description of a unique osteosynthesis method]. Acta Ortop Mex. 2017 Jun;31(3):148–51.
Borowski A, Grissom L, Littleton AG, Donohoe M, King M, Kumar SJ. Diagnostic imaging of the knee in children with arthrogryposis and knee extension or hyperextension contracture. J Pediatr Orthop. 2008 Jun;28(4):466–70.
van Bosse HJP, Feldman DS, Anavian J, Sala DA. Treatment of knee flexion contractures in patients with arthrogryposis. J Pediatr Orthop. 2007 Dec;27(8):930–7.
van Bosse HJP, Pontén E, Wada A, Agranovich OE, Kowalczyk B, Lebel E, et al. Treatment of the Lower Extremity Contracture/Deformities. J Pediatr Orthop. 2017 Aug;37 Suppl 1:S16–23.
Brunner R, Hefti F, Tgetgel JD. Arthrogrypotic joint contracture at the knee and the foot: correction with a circular frame. J Pediatr Orthop B. 1997 Jul;6(3):192–7.
DelBello DA, Watts HG. Distal femoral extension osteotomy for knee flexion contracture in patients with arthrogryposis. J Pediatr Orthop. 1996 Feb;16(1):122–6.
Eamsobhana P, Kaewpornsawan K, Vanitcharoenkul E. Walking ability in patients with arthrogryposis multiplex congenita. Indian J Orthop. 2014 Jul;48(4):421–5.
Eulert J. [Clinical aspects and treatment of arthrogryposis multiplex congenita. Lower extremity]. Z Orthop Ihre Grenzgeb. 1984 Oct;122(5):661–9.
Fisher KA, Fisher DA. Total hip and knee replacement in a patient with arthrogryposis multiplex congenita. Am J Orthop. 2014 Apr;43(4):E79-82.
Fucs PMMB, Svartman C, de Assumpção RMC, Lima Verde SR. Quadricepsplasty in arthrogryposis (amyoplasia): long-term follow-up. J Pediatr Orthop B. 2005 May;14(3):219–24.
Gavriliu S, Georgescu I, Ulici A, Ghita R, Japie EM, Pandea N, et al. Herbert capsuloplasty and Burnei tenomyoplasty for the correction of genu flexum in cerebral palsy, arthrogryposis and posttraumatic. Chirurgia (Bucur). 2013 Dec;108(6):866–73.
Ho CA, Karol LA. The utility of knee releases in arthrogryposis. J Pediatr Orthop. 2008 May;28(3):307–13.
Hoffer MM, Swank S, Eastman F, Clark D, Teitge R. Ambulation in severe arthrogryposis. J Pediatr Orthop. 1983 Jul;3(3):293–6.
Klatt J, Stevens PM. Guided growth for fixed knee flexion deformity. J Pediatr Orthop. 2008 Sep;28(6):626–31.
Ko JY, Shih CH, Wenger DR. Congenital dislocation of the knee. J Pediatr Orthop. 1999 Apr;19(2):252–9.
Lampasi M, Antonioli D, Donzelli O. Management of knee deformities in children with arthrogryposis. Musculoskelet Surg. 2012 Dec;96(3):161–9.
Lénárt G, Mészáros T. [Knee results of the treatment of traction contractures using the quadriceps plasty by Bennett (author's transl)]. Arch Orthop Unfallchir. 1975 Jun 27;82(3):225–30.
Mirazimov BM, Dzhuraev AM. [Our experience in the treatment of deformations of the knee joint in arthrogryposis]. Ortop Travmatol Protez. 1989 Aug;(8):12–5.
Mooney JF, Koman LA. Knee flexion contractures: soft tissue correction with monolateral external fixation. J South Orthop Assoc. 2001;10(1):32–6.
Murray C, Fixsen JA. Management of knee deformity in classical arthrogryposis multiplex congenita (amyoplasia congenita). J Pediatr Orthop B. 1997 Jul;6(3):186–91.
Oleas-Santillán G, Bowen JR. Tension band plate-guided growth of knee-flexion deformity in arthrogryposis multiplex congenita in which metaphyseal funnelization induced screw encroachment upon the neurovascular bundle. J Pediatr Orthop B. 2020 Jan;29(1):62–4.
Palocaren T, Thabet AM, Rogers K, Holmes L, Donohoe M, King MM, et al. Anterior distal femoral stapling for correcting knee flexion contracture in children with arthrogryposis--preliminary results. J Pediatr Orthop. 2010 Mar;30(2):169–73.
Parsch K, Pietrzak S. [Congenital multiple arthrogryposis]. Orthopade. 2007 Mar;36(3):281–90; quiz 291.
Pontén E. Management of the knees in arthrogryposis. J Child Orthop. 2015 Dec;9(6):465–72.
Qin S, Chen J, Zheng X, Jiao S. [Ilizarov technique for correcting flexion deformity of the knee of arthrogryposis multiplex congenita]. Zhonghua Wai Ke Za Zhi. 2004 Aug 22;42(16):993–6.
Saleh M, Gibson MF, Sharrard WJ. Femoral shortening in correction of congenital knee flexion deformity with popliteal webbing. J Pediatr Orthop. 1989 Oct;9(5):609–11.
Södergård J, Ryöppy S. The knee in arthrogryposis multiplex congenita. J Pediatr Orthop. 1990 Apr;10(2):177–82.
Stilli S, Antonioli D, Lampasi M, Donzelli O. Management of hip contractures and dislocations in arthrogryposis. Musculoskelet Surg. 2012 Jun;96(1):17–21.

Sud A, Kumar N, Mehtani A. Femoral shortening in the congenital dislocation of the knee joint: results of mid-term follow-up. J Pediatr Orthop B. 2013 Sep;22(5):440–4.
Thomas B, Schopler S, Wood W, Oppenheim WL. The knee in arthrogryposis. Clin Orthop Relat Res. 1985 Apr;(194):87–92.
Yang SS, Dahan-Oliel N, Montpetit K, Hamdy RC. Ambulation gains after knee surgery in children with arthrogryposis. J Pediatr Orthop. 2010 Dec;30(8):863–9.

Muscle transfer

Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794–8.
Bayne LG. Hand assessment and management of arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):68–73.
Chomiak J, Dungl P. [Restoration of elbow joint flexion using pectoral muscle transfer in patients with arthrogryposis multiplex congenita. Part I: surgical method, rehabilitation and clinical results]. Acta Chir Orthop Traumatol Cech. 2002;69(6):333–43.
Chomiak J, Dungl P, Grim M. [Restoration of elbow joint flexion by transfer of the pectoralis major muscle in patients with arthrogryposis multiplex congenita. Part II. Results of electromyographic and histologic examinations]. Acta Chir Orthop Traumatol Cech. 2003;70(1):25–30.
Chomiak J, Dungl P, Včelák J. Reconstruction of elbow flexion in arthrogryposis multiplex congenita type I: results of transfer of pectoralis major muscle with follow-up at skeletal maturity. J Pediatr Orthop. 2014 Dec;34(8):799–807.
Doyle JR, James PM, Larsen LJ, Ashley RK. Restoration of elbow flexion in arthrogryposis multiplex congenita. J Hand Surg Am. 1980 Mar;5(2):149–52.
Gogola GR, Ezaki M, Oishi SN, Gharbaoui I, Bennett JB. Long head of the triceps muscle transfer for active elbow flexion in arthrogryposis. Tech Hand Up Extrem Surg. 2010 Jun;14(2):121–4.
Goldfarb CA, Burke MS, Strecker WB, Manske PR. The Steindler flexorplasty for the arthrogrypotic elbow. J Hand Surg Am. 2004 May;29(3):462–9.
Hagemann C, Stücker R, Breyer S, Kunkel POS. Nerve transfer from the median to musculocutaneous nerve to induce active elbow flexion in selected cases of arthrogryposis multiplex congenita. Microsurgery. 2019 Nov;39(8):710–4.
Lahoti O, Bell MJ. Transfer of pectoralis major in arthrogryposis to restore elbow flexion: deteriorating results in the long term. J Bone Joint Surg Br. 2005 Jun;87(6):858–60.
Lake AL, Oishi SN. Hand therapy following elbow release for passive elbow flexion and long head of the triceps transfer for active elbow flexion in children with amyoplasia. J Hand Ther. 2015 Jun;28(2):222–6; quiz 227.
Meyn M, Ruby L. Arthrogryposis of the upper extremity. Orthop Clin North Am. 1976 Apr;7(2):501–9.
Schwering L. Surgical correction of the true vertical talus deformity. Oper Orthop Traumatol. 2005 Jun;17(2):211–31.
Stevanovic M, Sharpe F. Functional free muscle transfer for upper extremity reconstruction. Plast Reconstr Surg. 2014 Aug;134(2):257e–74e.
Takagi T, Seki A, Kobayashi Y, Mochida J, Takayama S. Isolated Muscle Transfer to Restore Elbow Flexion in Children with Arthrogryposis. J Hand Surg Asian Pac Vol. 2016;21(1):44–8.
Thione A, Cavadas PC, Espadero AP, Rubi CG. Interbrachial Pinch by Trapezius Transfer in Amyoplasia Congenita: A Case Report. Plast Reconstr Surg Glob Open. 2017 May;5(5):e1292.
Van Heest A, Waters PM, Simmons BP. Surgical treatment of arthrogryposis of the elbow. J Hand Surg Am. 1998 Nov;23(6):1063–70.
Zargarbashi R, Nabian MH, Werthel J-D, Valenti P. Is bipolar latissimus dorsi transfer a reliable option to restore elbow flexion in children with arthrogryposis? A review of 13 tendon transfers. J Shoulder Elbow Surg. 2017 Nov;26(11):2004–9.

Pes convex

Adelaar RS, Williams RM, Gould JS. Congenital convex pes valgus: results of an early comprehensive release and a review of congenital vertical talus at Richmond Crippled Children's Hospital and the University of Alabama in Birmingham. Foot Ankle. 1980 Sep;1(2):62–73.
Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, et al. [Results of surgical treatment of congenital convex pes valgus (10 non-idiopathic feet)]. Rev Chir Orthop Reparatrice Appar Mot. 2008a Apr;94(2):128–34.
Ayadi K, Trigui M, Gdoura F, Zribi W, Zribi M, Elleuch MH, et al. Results of surgical treatment of congenital convex pes valgus (ten nonidiopathic feet). Rev Chir Orthop Reparatrice Appar Mot. 2008b Dec;94(8):e28-34.

Guidera KJ, Drennan JC. Foot and ankle deformities in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985 Apr;(194):93–8.
Ramanoudjame M, Loriaut P, Seringe R, Glorion C, Wicart P. The surgical treatment of children with congenital convex foot (vertical talus): evaluation of midtarsal surgical release and open reduction. Bone Joint J. 2014 Jun;96-B(6):837–44.
Yen CC, Huang SC. Surgical treatment of congenital convex pes valgus. J Formos Med Assoc. 1997 Jun;96(6):424–8.

Ponseti

Boehm S, Limpaphayom N, Alaee F, Sinclair MF, Dobbs MB. Early results of the Ponseti method for the treatment of clubfoot in distal arthrogryposis. J Bone Joint Surg Am. 2008 Jul;90(7):1501–7.
Church C, McGowan A, Henley J, Donohoe M, Niiler T, Shrader MW, et al. The 5-Year Outcome of the Ponseti Method in Children With Idiopathic Clubfoot and Arthrogryposis. J Pediatr Orthop. 2020 Feb 6;
Funk JF, Lebek S, Seidl T, Placzek R. [Comparison of treatment results of idiopathic and non-idiopathic congenital clubfoot : prospective evaluation of the Ponseti therapy]. Orthopade. 2012 Dec;41(12):977–83.
Guerra-Jasso JJ, Valcarce-León JA, Quíntela-Núñez-Del Prado HM. [Evidence level and grade of recommendation of the Ponseti method for arthrogryposis-related syndromic talipes equinovarus and Moebius syndrome: a systematic review]. Acta Ortop Mex. 2017 Aug;31(4):182–8.
Matar HE, Beirne P, Garg N. The effectiveness of the Ponseti method for treating clubfoot associated with arthrogryposis: up to 8 years follow-up. J Child Orthop. 2016 Feb;10(1):15–8.
Moroney PJ, Noël J, Fogarty EE, Kelly PM. A single-center prospective evaluation of the Ponseti method in nonidiopathic congenital talipes equinovarus. J Pediatr Orthop. 2012 Sep;32(6):636–40.
van Bosse HJP. Challenging clubfeet: the arthrogrypotic clubfoot and the complex clubfoot. J Child Orthop. 2019 Jun 1;13(3):271–81.

Skin web

Abdel-Ghani H, Mahmoud M, Shaheen A, Abdel-Wahed M. Treatment of congenital clasped thumb in arthrogryposis. J Hand Surg Eur Vol. 2017 Oct;42(8):794–8.

Spinal arthrodesis

Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 1985 Sep;10(7):609–13.
Greggi T, Martikos K, Pipitone E, Lolli F, Vommaro F, Maredi E, et al. Surgical treatment of scoliosis in a rare disease: arthrogryposis. Scoliosis. 2010 Nov 9;5:24.
Ho AL, Mohole J, Sussman ES, Pendharkar AV, Singh H. Cervical Stenosis in Adult Arthrogryposis: A Case Report and Review of the Literature. J Neurol Surg Rep. 2018 Jan;79(1):e19–22.
Siebold RM, Winter RB, Moe JH. The treatment of scoliosis in arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1974;(103):191–8.
Xu L, Chen Z, Qiu Y, Chen X, Li S, Du C, et al. Case-matched comparative analysis of spinal deformity correction in arthrogryposis multiplex congenita versus adolescent idiopathic scoliosis. J Neurosurg Pediatr. 2018 12;23(1):22–9.

Amyoplasia + MPR

Braces

Lampasi M, Antonioli D, Donzelli O. Management of knee deformities in children with arthrogryposis. Musculoskelet Surg. 2012 Dec;96(3):161–9.
Yang SS, Dahan-Oliel N, Montpetit K, Hamdy RC. Ambulation gains after knee surgery in children with arthrogryposis. J Pediatr Orthop. 2010 Dec;30(8):863–9.

Clinical care

Ayadi K, Trigui M, Abid A, Cheniour A, Zribi M, Keskes H. [Arthrogryposis: clinical manifestations and management]. Arch Pediatr. 2015 Aug;22(8):830–9.

Outcome + medical – orthopaedic

Sells JM, Jaffe KM, Hall JG. Amyoplasia, the most common type of arthrogryposis: the potential for good outcome. Pediatrics. 1996 Feb;97(2):225–31.

Physiotherapy

Lake AL, Oishi SN. Hand therapy following elbow release for passive elbow flexion and long head of the triceps transfer for active elbow flexion in children with amyoplasia. J Hand Ther. 2015 Jun;28(2):222–6; quiz 227.

Treatment + medical – orthopaedic

Kroksmark A-K, Kimber E, Jerre R, Beckung E, Tulinius M. Muscle involvement and motor function in Amyoplasia. Am J Med Genet A. 2006 Aug 15;140(16):1757–67