

BIBLIOGRAPHIE DU CENTRE DE REFERENCE GNMH (mise à jour fin 2016)

GNMH-Garches-Enfants

1. Gerin I, Ury B, Breloy I, Bouchet-Seraphin C, Bolsée J, Halbout M, Graff J, Vertommen D, Muccioli GG, Seta N, Cuisset JM, Dabaj I, Quijano-Roy S, Grahn A, Van Schaftingen E, Bommer GT. ISPD produces CDP-ribitol used by FKTN and FKRP to transfer ribitol phosphate onto α -dystroglycan. *Nature Commun.* 2016 May 19;7:11534.
2. Schartner V, Romero NB, Donkervoort S, Treves S, Munot P, Pierson TM, Dabaj I, Malfatti E, Zaharieva IT, Zorzato F, Abath Neto O, Brochier G, Lornage X, Eymard B, Taratuto AL, Böhm J, Gonorazky H, Ramos-Platt L, Feng L, Phadke R, Bharucha-Goebel DX, Sumner CJ, Bui MT, Lacene E, Beuvin M, Labasse C, Dondaine N, Schneider R, Thompson J, Boland A, Deleuze JF, Matthews E, Pakleza AN, Sewry CA, Biancalana V, Quijano-Roy S, Muntoni F, Fardeau M, Bönnemann CG, Laporte J. Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy. *Acta Neuropathol.* 2016 Dec 23. doi: 10.1007/s00401-016-1656-8. [Epub ahead of print]
3. Quijano-Roy S, Khirani S, Colella M, Ramirez A, Aloui S, Wehbi S, de Becdelievre A, Carlier RY, Allamand V, Richard P, Azzi V, Estournet B, Fauroux B. Diaphragmatic dysfunction in Collagen VI myopathies. *Neuromuscul Disord.* 2014 Feb;24(2):125-33. 24314752.
4. Heller F, Dabaj I, Mah JK, Bergounioux J, Essid A, Bönnemann CG, Rutkowski A, Bonne G, Quijano-Roy S, Wahbi K. Cardiac manifestations of congenital LMNA-related muscular dystrophy in children: three case reports and recommendations for care. *Cardiol Young.* 2016 Dec 12:1-7.
5. Gitiaux C, Chemaly N, Quijano-Roy S, Barnerias C, Desguerre I, Hully M, Chiron C, Dulac O, Nabbout R. Motor neuropathy contributes to crouching in patients with Dravet syndrome. *Neurology.* 2016 Jul 19;87(3):277-81.
6. Gómez-Andrés D, Dabaj I, Mompoin D, Hankiewicz K, Azzi V, Ioos C, Romero NB, Ben Yaou R, Bergounioux J, Bonne G, Richard P, Estournet B, Yves-Carlier R, Quijano-Roy S. Pediatric laminopathies: Whole-body magnetic resonance imaging fingerprint and comparison with Sepn1 myopathy. *Muscle Nerve.* 2016 Aug;54(2):192-202.
7. Mazuet C, Yoon EJ, Boyer S, Pignier S, Blanc T, Doehring I, Meziane-Cherif D, Dumant-Forest C, Sautereau J, Legeay C, Bouvet P, Bouchier C, Quijano-Roy S, Pestel-Caron M, Courvalin P, Popoff MR. A penicillin- and metronidazole-resistant *Clostridium botulinum* strain responsible for an infant botulism case. *Clin Microbiol Infect.* 2016 Apr 21. pii: S1198-743X(16)30068-4.
8. Cavassa E, Tordjman M, Ferreiro A, Carlier R, Quijano-Roy S. [Diagnostic orientation of « Rigid spine » familial case with whole body muscle MRI]. *Med Sci (Paris).* 2016 Nov;32 Hors série n°2:14-16. French.
9. Sauvagnac-Quera R, Vabre C, Azzi V, Tirolien S, Leiba N, Poisson F, Miladi L, Carlier R, Glorion C, Leclair D, Estournet B, Quijano-Roy S. Prevention and treatment of scoliosis by Garches Brace in children with type Ib SMA. *Ann Phys Rehabil Med.* 2016 Sep;59S:e92.
10. Béchir N, Pecchi É, Relizani K, Vilmen C, Le Fur Y, Bernard M, Amthor H, Bendahan D, Giannesini B. Mitochondrial impairment induced by postnatal ActRIIB blockade does not alter function and energy status in exercising mouse glycolytic muscle in vivo. *Am J Physiol Endocrinol Metab.* 2016 Apr 1;310(7):E539-49.

11. Béchir N, Pecchi E, Vilmen C, Le Fur Y, Amthor H, Bernard M, Bendahan D, Giannesini B. ActRIIB blockade increases force-generating capacity and preserves energy supply in exercising mdx mouse muscle in vivo. *FASEB J*. 2016 Oct;30(10):3551-3562.
12. Stantzou A, Ueberschlag-Pitiot V, Thomasson R, Furling D, Bonnieu A, Amthor H, Ferry A. Effect of constitutive inactivation of the myostatin gene on the gain in muscle strength during postnatal growth in two murine models. *Muscle Nerve*. 2016 Jun 16. doi: 10.1002/mus.25220. [Epub ahead of print]
13. 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 arthrogyriposis, spinal stiffness and rare polyglucosan bodies in muscle. *Neuromuscul Disord* 2016 (In Press)
14. Petkova MV, Morales-Gonzales S, Relizani K, Gill E, Seifert F, Radke J, Stenzel W, Garcia L, Amthor H, Schuelke M. Characterization of a Dmd (EGFP) reporter mouse as a tool to investigate dystrophin expression. *Skelet Muscle*. 2016 Jul 5;6:25.
15. Richard I, Laurent JP, Cirak S, Vissing J; ENMC FKRP Study Group. 216th ENMC international workshop: Clinical readiness in FKRP related myopathies January 15-17, 2016 Naarden, The Netherlands. *Neuromuscul Disord*. 2016 Oct;26(10):717-724.
16. Khirani S, Ramirez A, Olmo-Arroyo J, Amaddeo A, Quijano-Roy S, Desguerre I, Fauroux B. [Are respiratory muscle testing helpful to prompt sleep studies in children with neuromuscular disease?]. *Med Sci (Paris)* 2015 Nov;31 Spec No 3:14-7. French
17. Hankiewicz K, Carlier RY, Lazaro L, Linzoain J, Barnerias C, Gómez-Andrés D, Avila-Smirnow D, Ferreira A, Estournet B, Guicheney P, Germain DP, Richard P, Bulacio S, Mompont D, Quijano-Roy S. Whole-body muscle magnetic resonance imaging in SEP11-related myopathy shows a homogeneous and recognizable pattern. *Muscle Nerve*. 2015 Nov;52(5):728-35.
18. Goyenvallé A, Griffith G, Babbs A, El Andaloussi S, Ezzat K, Avril A, Dugovic B, Chausseot R, Ferry A, Voit T, Amthor H, Bühr C, Schürch S, Wood MJ, Davies KE, Vaillend C, Leumann C, Garcia L. Functional correction in mouse models of muscular dystrophy using exon-skipping tricyclo-DNA oligomers. *Nat Med*. 2015 Mar;21(3):270-5.
19. Fauroux B, Quijano-Roy S, Desguerre I, Khirani S. The value of respiratory muscle testing in children with neuromuscular disease. *Chest*. 2015 Feb;147(2):552-9.
20. Carrasco-Marina ML, Quijano-Roy S, Iglesias-Escalera G, Jorge-Blanco A, Carro-Martinez A, Gutierrez-Cruz N. [Ullrich congenital muscular dystrophy. The usefulness of muscular magnetic resonance imaging in its diagnosis]. *Rev Neurol*. 2015 Jul 1;61(1):44-6. Spanish.
21. Seferian AM, Moraux A, Annoussamy M, Canal A, Decostre V, Diebete O, Le Moing AG, Gidaro T, Deconinck N, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Maincent K, Desguerre I, Thémar-Noël C, Cuisset JM, Tiffreau V, Denis S, Jousten V, Quijano-Roy S, Voit T, Hogrel JY, Servais L. Upper limb strength and function changes during a one-year follow-up in non-ambulant patients with Duchenne Muscular Dystrophy: an observational multicenter trial. *PLoS One*. 2015 Feb 2;10(2):e0113999
22. Catteruccia M, Vuillerot C, Vaugier I, Leclair D, Azzi V, Viollet L, Estournet B, Bertini E, Quijano-Roy S. Orthopedic Management of Scoliosis by Garches Brace and Spinal Fusion in SMA Type 2 Children. *J Neuromuscul Dis*. 2015 Nov 21;2(4):453-462.
23. Gitiaux C, Blin-Rochemaure N, Hully M, Echaniz-Laguna A, Calmels N, Bahi-Buisson N, Desguerre I, Dabaj I, Wehbi S, Quijano-Roy S, Laugel V. Progressive demyelinating neuropathy correlates with clinical severity in Cockayne syndrome. *Clin Neurophysiol*. 2015 Jul;126(7):1435-9.
24. Goyenvallé A, Griffith G, Avril A, Amthor H, Garcia L. [Functional correction and cognitive improvement in dystrophic mice using splice-switching tricyclo-DNA oligomers]. *Med Sci (Paris)*. 2015 Mar;31(3):253-6.
25. Seferian AM, Moraux A, Canal A, Decostre V, Diebete O, Le Moing AG, Gidaro T, Deconinck N, Van Parys F, Vereecke W, Wittevrongel S, Annoussamy M, Mayer M, Maincent K, Cuisset JM,

- Tiffreau V, Denis S, Jousten V, Quijano-Roy S, Voit T, Hogrel JY, Servais L. Upper limb evaluation and one-year follow up of non-ambulant patients with spinal muscular atrophy: an observational multicenter trial. *PLoS One*. 2015 Apr 10;10(4):e0121799.
26. Amthor H. [The basic concept of therapeutic approaches for DMD]. *Arch Pediatr*. 2015 Dec;22(12 Suppl 1):12S63-8.
 27. Khirani S, Dabaj I, Amaddeo A, Ramirez A, Quijano-Roy S, Fauroux B. The value of respiratory muscle testing in a child with congenital muscular dystrophy. *Respirol Case Rep*. 2014 Sep;2(3):95-8.
 28. Bolocan A, Quijano-Roy S, Seferian AM, Baumann C, Allamand V, Richard P, Estournet B, Carlier R, Cavé H, Gartioux C, Blin N, Le Moing AG, Gidaro T, Germain DP, Fardeau M, Voit T, Servais L, Romero NB. Congenital muscular dystrophy phenotype with neuromuscular spindles excess in a 5-year-old girl caused by HRAS mutation. *Neuromuscul Disord*. 2014 Nov;24(11):993-8.
 29. Nicole S, Chaouch A, Torbergesen T, Bauché S, de Bruyckere E, Fontenille MJ, Horn MA, van Ghelue M, Løseth S, Issop Y, Cox D, Müller JS, Evangelista T, Stålborg E, loos C, Barois A, Brochier G, Sternberg D, Fournier E, Hantai D, Abicht A, Dusl M, Laval SH, Griffin H, Eymard B, Lochmüller H. Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. *Brain*. 2014 Sep;137(Pt 9):2429-43.
 30. Mouisel E, Relizani K, Mille-Hamard L, Denis R, Hourdé C, Agbulut O, Patel K, Arandel L, Morales-Gonzalez S, Vignaud A, Garcia L, Ferry A, Luquet S, Billat V, Ventura-Clapier R, Schuelke M, Amthor H. Myostatin is a key mediator between energy metabolism and endurance capacity of skeletal muscle. *Am J Physiol Regul Integr Comp Physiol*. 2014 Aug 15;307(4):R444-54.
 31. Mosler S, Relizani K, Mouisel E, Amthor H, Diel P. Combinatory effects of siRNA-induced myostatin inhibition and exercise on skeletal muscle homeostasis and body composition. *Physiol Rep*. 2014 Mar 20;2(3):e00262.
 32. Laquérière A, Maluenda J, Camus A, Fontenas L, Dieterich K, Nolent F, Zhou J, Monnier N, Latour P, Gentil D, Héron D, Desguettes I, Landrieu P, Beneteau C, Delaporte B, Bellesme C, Baumann C, Capri Y, Goldenberg A, Lyonnet S, Bonneau D, Estournet B, Quijano-Roy S, Francannet C, Odent S, Saint-Frison MH, Sigaudy S, Figarella-Branger D, Gelot A, Mussini JM, Lacroix C, Drouin-Garraud V, Malinge MC, Attié-Bitach T, Bessieres B, Bonniere M, Encha-Razavi F, Beaufrère AM, Khung-Savatovsky S, Perez MJ, Vasiljevic A, Mercier S, Roume J, Trestard L, Saugier-veber P, Cordier MP, Layet V, Legendre M, Vigouroux-Castera A, Lunardi J, Bayes M, Jouk PS, Rigonnot L, Granier M, Sternberg D, Warszawski J, Gut I, Gonzales M, Tawk M, Melki J. Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axonal defects. *Hum Mol Genet*. 2014 May 1;23(9):2279-89.
 33. Relizani K, Mouisel E, Giannesini B, Hourdé C, Patel K, Morales Gonzalez S, Jülich K, Vignaud A, Piétri-Rouxel F, Fortin D, Garcia L, Blot S, Ritvos O, Bendahan D, Ferry A, Ventura-Clapier R, Schuelke M, Amthor H. Blockade of ActRIIB signaling triggers muscle fatigability and metabolic myopathy. *Mol Ther*. 2014 Aug;22(8):1423-33.
 34. Vuillerot C, Rippert P, Kinet V, Renders A, Jain M, Waite M, Glanzman AM, Girardot F, Hamroun D, Iwaz J, Ecochard R, Quijano-Roy S, Bérard C, Poirot I, Bönnemann CG; CDM MFM Study Group. Rasch analysis of the motor function measure in patients with congenital muscle dystrophy and congenital myopathy. *Arch Phys Med Rehabil*. 2014 Nov;95(11):2086-95.
 35. Bertrand AT, Ziaei S, Ehret C, Duchemin H, Mamchaoui K, Bigot A, Mayer M, Quijano-Roy S, Desguettes I, Lainé J, Ben Yaou R, Bonne G, Coirault C. Cellular microenvironments reveal defective mechanosensing responses and elevated YAP signaling in LMNA-mutated muscle precursors. *J Cell Sci*. 2014 Jul 1;127(Pt 13):2873-84.
 36. Malfatti E, Lehtokari VL, Böhm J, De Winter JM, Schäffer U, Estournet B, Quijano-Roy S, Monges S, Lubieniecki F, Bellance R, Viou MT, Madelaine A, Wu B, Taratuto AL, Eymard B, Pelin K, Fardeau M, Ottenheijm CA, Wallgren-Pettersson C, Laporte J, Romero NB. Muscle

- histopathology in nebulin-related nemaline myopathy: ultrastructural findings correlated to disease severity and genotype. *Acta Neuropathol Commun*. 2014 Apr 12;2:44.
37. North KN, Wang CH, Clarke N, Jungbluth H, Vainzof M, Dowling JJ, Amburgey K, Quijano-Roy S, Beggs AH, Sewry C, Laing NG, Bönnemann CG; International Standard of Care Committee for Congenital Myopathies. Approach to the diagnosis of congenital myopathies. *Neuromuscul Disord*. 2014 Feb;24(2):97-116.
 38. Bönnemann CG, Wang CH, Quijano-Roy S, Deconinck N, Bertini E, Ferreira A, Muntoni F, Sewry C, Bérout C, Mathews KD, Moore SA, Bellini J, Rutkowski A, North KN; Members of International Standard of Care Committee for Congenital Muscular Dystrophies. Diagnostic approach to the congenital muscular dystrophies. *Neuromuscul Disord*. 2014 Apr;24(4):289-311.
 39. Quijano-Roy S, Carlier RY. Muscle magnetic resonance imaging: a new diagnostic tool with promising avenues in therapeutic trials. *Neuropediatrics*. 2014 Oct;45(5):273-4.
 40. Barnérias C, Quijano S, Mayer M, Estournet B, Cuisset JM, Sukno S, Peudenier S, Laroche C, Chabrier S, Sabouraud P, Vuillerot C, Chabrol B, Halbert C, Cancès C, Beze-Beyrie P, Ledivenah A, Viillard ML, Desguerre I. [Multicentric study of medical care and practices in spinal muscular atrophy type 1 over two 10-year periods]. *Arch Pediatr*. 2014 Apr;21(4):347-54.
 41. Vuillerot C, Rippert P, Roche S, Bérard C, Margirier F, de Lattre C, Poirot I, Berruyer A, Tiffreau V, Fournier-Mehouas M, Bouhour F, Urtizberea JA, Renders A, Ecochard R; Le groupe d'étude NM-Score. Development and validation of a motor function classification in patients with neuromuscular disease: the NM-score. *Ann Phys Rehabil Med*. 2013 Dec;56(9-10):673-86.
 42. Estournet B. Respiratory care in neuromuscular disorders. *Handb Clin Neurol*. 2013;113:1485-90.
 43. 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 arthrogyposis. *Hum Mol Genet*. 2013 Apr 15;22(8):1483-92.
 44. Eymard B, Hantaï D, Estournet B. Congenital myasthenic syndromes. *Handb Clin Neurol*. 2013;113:1469-80.
 45. Eymard B, Stojkovic T, Sternberg D, Richard P, Nicole S, Fournier E, Béhin A, Laforêt P, Servais L, Romero N, Fardeau M, Hantaï D; Membres du réseau national Syndromes Myasthéniques Congénitaux. [Congenital myasthenic syndromes: difficulties in the diagnosis, course and prognosis, and therapy--The French National Congenital Myasthenic Syndrome Network experience]. *Rev Neurol (Paris)*. 2013 Feb;169 Suppl 1:S45-55.
 46. Sartori R, Schirwis E, Blaauw B, Bortolanza S, Zhao J, Enzo E, Stantzou A, Mouisel E, Toniolo L, Ferry A, Stricker S, Goldberg AL, Dupont S, Piccolo S, Amthor H, Sandri M. BMP signaling controls muscle mass. *Nat Genet*. 2013 Nov;45(11):1309-18.
 47. Foley AR, Quijano-Roy S, Collins J, Straub V, McCallum M, Deconinck N, Mercuri E, Pane M, D'Amico A, Bertini E, North K, Ryan MM, Richard P, Allamand V, Hicks D, Lamandé S, Hu Y, Gualandi F, Auh S, Muntoni F, Bönnemann CG. Natural history of pulmonary function in collagen VI-related myopathies. *Brain*. 2013 Dec;136(Pt12):3625-33.
 48. Giannesini B, Vilmen C, Amthor H, Bernard M, Bendahan D. Lack of myostatin impairs mechanical performance and ATP cost of contraction in exercising mouse gastrocnemius muscle in vivo. *Am J Physiol Endocrinol Metab*. 2013 Jul 1;305(1):E33-40.
 49. Servais L, Deconinck N, Moraux A, Benali M, Canal A, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Desguerre I, Maincent K, Themar-Noel C, Quijano-Roy S, Serari N, Voit T, Hogrel JY. Innovative methods to assess upper limb strength and function in non-ambulant Duchenne patients. *Neuromuscul Disord*. 2013 Feb;23(2):139-48.
 50. North KN, Wang CH, Clarke N, Jungbluth H, Vainzof M, Dowling JJ, Amburgey K, Quijano-Roy S, Beggs AH, Sewry C, Laing NG, Bönnemann CG; International Standard of Care Committee for Congenital Myopathies. Approach to the diagnosis of congenital myopathies. *Neuromuscul Disord*. 2014 Feb;24(2):97-116.

51. Laquérière A, Maluenda J, Camus A, Fontenas L, Dieterich K, Nolent F, Zhou J, Monnier N, Latour P, Gentil D, Héron D, Desguerres I, Landrieu P, Beneteau C, Delaporte B, Bellesme C, Baumann C, Capri Y, Goldenberg A, Lyonnet S, Bonneau D, Estournet B, Quijano-Roy S, Francannet C, Odent S, Saint-Frison MH, Sigaudy S, Figarella-Branger D, Gelot A, Mussini JM, Lacroix C, Drouin-Garraud V, Malinge MC, Attié-Bitach T, Bessieres B, Bonniere M, Encha-Razavi F, Beaufrère AM, Khung-Savatovsky S, Perez MJ, Vasiljevic A, Mercier S, Roume J, Trestard L, Saugier-veber P, Cordier MP, Layet V, Legendre M, Vigouroux-Castera A, Lunardi J, Bayes M, Jouk PS, Rigonnot L, Granier M, Sternberg D, Warszawski J, Gut I, Gonzales M, Tawk M, Melki J. Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. *Hum Mol Genet.* 2014 May 1;23(9):2279-89.
52. van de Kamp JM, Betsalel OT, Mercimek-Mahmutoglu S, Abulhoul L, Grünewald S, Anselm I, Azzouz H, Bratkovic D, de Brouwer A, Hamel B, Kleefstra T, Yntema H, Campistol J, Vilaseca MA, Cheillan D, D'Hooghe M, Diogo L, Garcia P, Valongo C, Fonseca M, Frints S, Wilcken B, von der Haar S, Meijers-Heijboer HE, Hofstede F, Johnson D, Kant SG, Lion-Francois L, Pitelet G, Longo N, Maat-Kievit JA, Monteiro JP, Munnich A, Muntau AC, Nassogne MC, Osaka H, Ounap K, Pinard JM, Quijano-Roy S, Poggenburg I, Poplawski N, Abdul-Rahman O, Ribes A, Arias A, Yaplito-Lee J, Schulze A, Schwartz CE, Schwenger S, Soares G, Sznajder Y, Valayannopoulos V, Van Esch H, Waltz S, Wamelink MM, Pouwels PJ, Errami A, van der Knaap MS, Jakobs C, Mancini GM, Salomons GS. Phenotype and genotype in 101 males with X-linked creatine transporter deficiency. *J Med Genet.* 2013 Jul;50(7):463-72.
53. Rul B, Quijano-Roy S, Golse A, Beynier D, Estournet B, Desguerres I, Barnerias C, Herve C. [What a tracheostomy changes in a child with a neuromuscular disease]. *Rech Soins Infirm.* 2013 Sep;(114):46-57. French.
54. Bonne G, Quijano-Roy S. Emery-Dreifuss muscular dystrophy, laminopathies, and other nuclear envelopathies. *Handb Clin Neurol.* 2013;113:1367-76.
55. de Lattre C, Payan C, Vuillerot C, Rippert P, de Castro D, Bérard C, Poirot I; MFM-20 Study Group. Motor function measure: validation of a short form for young children with neuromuscular diseases. *Arch Phys Med Rehabil.* 2013 Nov;94(11):2218-26.
56. Letellier G, Mok E, Alberti C, De Luca A, Gottrand F, Cuisset JM, Denjean A, Darmaun D, Hankard R. Effect of glutamine on glucose metabolism in children with Duchenne muscular dystrophy. *Clin Nutr.* 2013 Jun;32(3):386-90.
57. Schirwis E, Agbulut O, Vadrot N, Mouisel E, Hourdé C, Bonnieu A, Butler-Browne G, Amthor H, Ferry A. The beneficial effect of myostatin deficiency on maximal muscle force and power is attenuated with age. *Exp Gerontol.* 2013 Feb;48(2):183-90.
58. Servais L, Deconinck N, Moraux A, Benali M, Canal A, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Desguerres I, Maincent K, Themar-Noel C, Quijano-Roy S, Serari N, Voit T, Hogrel JY. Innovative methods to assess upper limb strength and function in non-ambulant Duchenne patients. *Neuromuscul Disord.* 2013 Feb;23(2):139-48. 23219352.
59. Gitiaux C, Bergounioux J, Magen M, Quijano-Roy S, Blanc T, Bonnefont JP, Desguerres I. Diaphragmatic weakness with progressive sensory and motor polyneuropathy: case report of a neonatal IGHMBP2-related neuropathy. *J Child Neurol.* 2013 Jun;28(6):787-90.
60. Quijano-Roy S, Sparks S, Rutkowski A. LAMA2-Related Muscular Dystrophy. 2012 Jun 7. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.
61. Baujat G, Huber C, El Hokayem J, Caumes R, Do Ngoc Thanh C, David A, Delezoide AL, Dieux-Coeslier A, Estournet B, Francannet C, Kayirangwa H, Lacaille F, Le Bourgeois M, Martinovic J, Salomon R, Sigaudy S, Malan V, Munnich A, Le Merrer M, Le Quan Sang KH, Cormier-Daire V. Asphyxiating thoracic dysplasia: clinical and molecular review of 39 families. *J Med Genet.* 2013 Feb;50(2):91-8.
62. Ravenscroft G, Miyatake S, Lehtokari VL, Todd EJ, Vornanen P, Yau KS, Hayashi YK, Miyake N, Tsurusaki Y, Doi H, Saitsu H, Osaka H, Yamashita S, Ohya T, Sakamoto Y, Koshimizu E,

- Imamura S, Yamashita M, Ogata K, Shiina M, Bryson-Richardson RJ, Vaz R, Ceyhan O, Brownstein CA, Swanson LC, Monnot S, Romero NB, Amthor H, Kresoje N, Sivadorai P, Kiraly-Borri C, Haliloglu G, Talim B, Orhan D, Kale G, Charles AK, Fabian VA, Davis MR, Lammens M, Sewry CA, Manzur A, Muntoni F, Clarke NF, North KN, Bertini E, Nevo Y, Willichowski E, Silberg IE, Topaloglu H, Beggs AH, Allcock RJ, Nishino I, Wallgren-Pettersson C, Matsumoto N, Laing NG. Mutations in KLHL40 are a frequent cause of severe autosomal-recessive nemaline myopathy. *Am J Hum Genet.* 2013 Jul 11;93(1):6-18.
63. Wang CH, Dowling JJ, North K, Schroth MK, Sejersen T, Shapiro F, Bellini J, Weiss H, Guillet M, Amburgey K, Apkon S, Bertini E, Bonnemann C, Clarke N, Connolly AM, Estournet-Mathiaud B, Fitzgerald D, Florence JM, Gee R, Gurgel-Giannetti J, Glanzman AM, Hofmeister B, Jungbluth H, Koumbourlis AC, Laing NG, Main M, Morrison LA, Munns C, Rose K, Schuler PM, Sewry C, Storhaug K, Vainzof M, Yuan N. Consensus statement on standard of care for congenital myopathies. *J Child Neurol.* 2012 Mar;27(3):363-82.
64. Didier N, Hourdé C, Amthor H, Marazzi G, Sassoon D. Loss of a single allele for Ku80 leads to progenitor dysfunction and accelerated aging in skeletal muscle. *EMBO Mol Med.* 2012 Sep;4(9):910-23.
65. Baumann M, Giunta C, Krabichler B, Rüschemdorf F, Zoppi N, Colombi M, Bittner RE, Quijano-Roy S, Muntoni F, Cirak S, Schreiber G, Zou Y, Hu Y, Romero NB, Carlier RY, Amberger A, Deutschmann A, Straub V, Rohrbach M, Steinmann B, Rostásy K, Karall D, Bönnemann CG, Zschocke J, Fauth C. Mutations in FKBP14 cause a variant of Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss. *Am J Hum Genet.* 2012 Feb 10;90(2):201-16.
66. Cuisset JM, Estournet B; French Ministry of Health. Recommendations for the diagnosis and management of typical childhood spinal muscular atrophy. *Rev Neurol (Paris).* 2012 Dec;168(12):902-9.
67. Quijano-Roy S, Avila-Smirnow D, Carlier RY; WB-MRI muscle study group. Whole body muscle MRI protocol: pattern recognition in early onset NM disorders. *Neuromuscul Disord.* 2012 Oct 1;22 Suppl 2:S68-84.
68. Ochala J, Gokhin DS, Péniisson-Besnier I, Quijano-Roy S, Monnier N, Lunardi J, Romero NB, Fowler VM. Congenital myopathy-causing tropomyosin mutations induce thin filament dysfunction via distinct physiological mechanisms. *Hum Mol Genet.* 2012 Oct 15;21(20):4473-85.
69. Amthor H, Hoogaars WM. Interference with myostatin/ActRIIB signaling as a therapeutic strategy for Duchenne muscular dystrophy. *Curr Gene Ther.* 2012 Jun;12(3):245-59.
70. Pinard JM, Azabou E, Essid N, Quijano-Roy S, Haddad S, Cheliout-Hérou F. Sleep-disordered breathing in children with congenital muscular dystrophies. *Eur J Paediatr Neurol.* 2012 Nov;16(6):619-24
71. 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.
72. Jungbluth H, Dowling JJ, Ferreira A, Muntoni F. 182nd ENMC International Workshop: RYR1-related myopathies, 15-17th April 2011, Naarden, The Netherlands. *Neuromuscul Disord.* 2012 May;22(5):453-62.
73. Rul B, Carnevale F, Estournet B, Rudler M, Hervé C. Tracheotomy and children with spinal muscular atrophy type 1: ethical considerations in the French context. *Nurs Ethics.* 2012 May;19(3):408-18.
74. Ohana M, Quijano-Roy S, Colas F, Lebreton C, Vallée C, Carlier RY. Axonotmesis of the sciatic nerve. *Diagn Interv Imaging.* 2012 May;93(5):398-400.
75. Blin-Rochemaure N, Quinet B. [Should a lumbar puncture be performed in any child with acute peripheral facial palsy and clinical suspicion of Lyme borreliosis?]. *Arch Pediatr.* 2012 Dec;19(12):1354-61.

76. Betsalel OT, Pop A, Rosenberg EH, Fernandez-Ojeda M; Creatine Transporter Research, Group, Jakobs C, Salomons GS. Detection of variants in SLC6A8 and functional analysis of unclassified missense variants. *Mol Genet Metab.* 2012 Apr;105(4):596-601.
77. Sparks S, Quijano-Roy S, Harper A, Rutkowski A, Gordon E, Hoffman EP, Pegoraro E. Congenital Muscular Dystrophy Overview. 2001 Jan 22 [updated 2012 Aug 23]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.
78. Matsakas A, Macharia R, Otto A, Elashry MI, Mouisel E, Romanello V, Sartori R, Amthor H, Sandri M, Narkar V, Patel K. Exercise training attenuates the hypermuscular phenotype and restores skeletal muscle function in the myostatin null mouse. *Exp Physiol.* 2012 Jan;97(1):125-40.

GNMH-Garches Adultes

1. Birnbaum S, Ghout I, Demeret S, Bolgert F, Eymard B, Sharshar T, et al. Translation, cross-cultural adaptation, and validation of the french version of the 15-item Myasthenia Gravis Quality Of life scale. *Muscle & nerve.* 2016 Aug 17. PubMed PMID: 27533497.
2. Boentert M, Prigent H, Vardi K, Jones HN, Mellies U, Simonds AK, et al. Practical Recommendations for Diagnosis and Management of Respiratory Muscle Weakness in Late-Onset Pompe Disease. *International journal of molecular sciences.* 2016 Oct 17;17(10). PubMed PMID: 27763517. Pubmed Central PMCID: 5085764.
3. Boussaid G, Devaux C, Lofaso F. [Respiratory care in patient with neuromuscular disease: the existing and the desirable]. *Medecine sciences : M/S.* 2016 Nov;32 Hors serie n degrees 2:27-9. PubMed PMID: 27869074. La prise en charge respiratoire chez le patient neuromusculaire - L'existant et le souhaitable.
4. Boussaid G, Lofaso F, Santos DB, Vaugier I, Pottier S, Prigent H, et al. Impact of invasive ventilation on survival when non-invasive ventilation is ineffective in patients with Duchenne muscular dystrophy: A prospective cohort. *Respiratory medicine.* 2016 Jun;115:26-32. PubMed PMID: 27215500.
5. Boussaid G, Lofaso F, Santos DB, Vaugier I, Pottier S, Prigent H, et al. Factors influencing compliance with non-invasive ventilation at long-term in patients with myotonic dystrophy type 1: A prospective cohort. *Neuromuscular disorders : NMD.* 2016 Oct;26(10):666-74. PubMed PMID: 27542439.
6. Brasil Santos D, Vaugier I, Boussaid G, Orlikowski D, Prigent H, Lofaso F. Impact of Noninvasive Ventilation on Lung Volumes and Maximum Respiratory Pressures in Duchenne Muscular Dystrophy. *Respiratory care.* 2016 Nov;61(11):1530-5. PubMed PMID: 27794082.
7. Fayssoil A, Lazarus A, Wahbi K, Oagna A, Nardi O, Lofaso F, et al. Cardiac implantable electronic devices in tracheotomized muscular dystrophy patients: Safety and risks. *International journal of cardiology.* 2016 Nov 01;222:975-7. PubMed PMID: 27526372.

8. Khirani S, Louis B, Leroux K, Ramirez A, Lofaso F, Fauroux B. Improvement of the trigger of a ventilator for non-invasive ventilation in children: bench and clinical study. *The clinical respiratory journal*. 2016 Sep;10(5):559-66. PubMed PMID: 25515939.
9. Meric H, Falaize L, Pradon D, Lacombe M, Petitjean M, Orlikowski D, et al. Short-term effect of volume recruitment-derecruitment manoeuvre on chest-wall motion in Duchenne muscular dystrophy. *Chronic respiratory disease*. 2016 Dec 06. PubMed PMID: 27923984.
10. Oгна A, Lofaso F. Mouthpiece ventilation: Individualized patient care is the key to success. *Chronic respiratory disease*. 2016 Aug 09. PubMed PMID: 27507834.
11. Oгна A, Nardi J, Prigent H, Quera Salva MA, Chaffaut C, Lamothe L, et al. Prognostic Value of Initial Assessment of Residual Hypoventilation Using Nocturnal Capnography in Mechanically Ventilated Neuromuscular Patients: A 5-Year Follow-up Study. *Frontiers in medicine*. 2016;3:40. PubMed PMID: 27679799. Pubmed Central PMCID: 5020068.
12. Oгна A, Prigent H, Falaize L, Leroux K, Santos D, Vaugier I, et al. Bench evaluation of commercially available and newly developed interfaces for mouthpiece ventilation. *The clinical respiratory journal*. 2016 Dec 27. PubMed PMID: 28026119.
13. Orlikowski D, Prigent H, Ambrosi X, Vaugier I, Pottier S, Annane D, et al. Comparison of ventilator-integrated end-tidal CO₂ and transcutaneous CO₂ monitoring in home-ventilated neuromuscular patients. *Respiratory medicine*. 2016 Aug;117:7-13. PubMed PMID: 27492508.
14. Salluh JI, Sharshar T, Kress JP. Does this patient have delirium? *Intensive care medicine*. 2016 Sep 12. PubMed PMID: 27620296.
15. Oгна A, Prigent H, Falaize L, Leroux K, Santos D, Vaugier I, Orlikowski D, Lofaso F. Accuracy of tidal volume delivered by home mechanical ventilation during mouthpiece ventilation: A bench evaluation. *Chron Respir Dis*. 2016 May 3.
16. Fayssoil A, Oгна A, Chaffaut C, Chevret S, Guimarães-Costa R, Leturcq F, Wahbi K, Prigent H, Lofaso F, Nardi O, Clair B, Behin A, Stojkovic T, Laforet P, Orlikowski D, Annane D. Natural History of Cardiac and Respiratory Involvement, Prognosis and Predictive Factors for Long-Term Survival in Adult Patients with Limb Girdle Muscular Dystrophies Type 2C and 2D. *PLoS One*. 2016 Apr 27;11(4):e0153095.
17. Garguilo M, Lejaille M, Vaugier I, Orlikowski D, Terzi N, Lofaso F, Prigent H. Noninvasive Mechanical Ventilation Improves Breathing-Swallowing Interaction of Ventilator Dependent Neuromuscular Patients: A Prospective Crossover Study. *PLoS One*. 2016 Mar 3;11(3):e0148673
18. Ambrosi X, Lamothe L, Heming N, Orlikowski D. [Respiratory and intensive care aspects of muscular dystrophies]. *Arch Pediatr*. 2015 Dec;22(12 Suppl 1):12S51-7.
19. Meric H, Falaize L, Pradon D, Orlikowski D, Prigent H, Lofaso F. 3D analysis of the chest wall motion for monitoring late-onset Pompe disease patients. *Neuromuscul Disord*. 2016 Feb;26(2):146-52.

20. Nardi J, Leroux K, Orlikowski D, Prigent H, Lofaso F. Home monitoring of daytime mouthpiece ventilation effectiveness in patients with neuromuscular disease. *Chron Respir Dis*. 2016 Feb;13(1):67-74.
21. Ognà A, Quera Salva MA, Prigent H, Mroue G, Vaugier I, Annane D, Lofaso F, Orlikowski D. Nocturnal hypoventilation in neuromuscular disease: prevalence according to different definitions issued from the literature. *Sleep Breath*. 2016 May;20(2):575-81.
22. Santos DB, Boussaid G, Stojkovic T, Orlikowski D, Letilly N, Behin A, Butel S, Lofaso F, Prigent H. Respiratory muscle dysfunction in facioscapulohumeral muscular dystrophy. *Neuromuscul Disord*. 2015 Aug;25(8):632-9.
23. Carlier PG, Azzabou N, de Sousa PL, Hicks A, Boisserie JM, Amadon A, Carlier RY, Wary C, Orlikowski D, Laforêt P. Skeletal muscle quantitative nuclear magnetic resonance imaging follow-up of adult Pompe patients. *J Inherit Metab Dis*. 2015 May;38(3):565-72.
24. Dervaux B, Szwarcensztein K, Josseran A; participants of round table N°4 of Giens XXX; Barna A, Carbonneil C, Chevre K, Debroucker F, Grumblat A, Grumel O, Massol J, Maugeudre P, Méchin H, Orlikowski D, Roussel C, Rumeau-Pichon C, Sales JP, Vicaut E. Assessment and non-clinical impact of medical devices. *Thérapie*. 2015 Jan-Feb;70(1):57-68.
25. Sansone VA, Gagnon C; participants of the 207th ENMC Workshop. 207th ENMC Workshop on chronic respiratory insufficiency in myotonic dystrophies: management and implications for research, 27-29 June 2014, Naarden, The Netherlands. *Neuromuscul Disord*. 2015 May;25(5):432-42.
26. Annane D, Orlikowski D, Chevret S. Nocturnal mechanical ventilation for chronic hypoventilation in patients with neuromuscular and chest wall disorders. *Cochrane Database Syst Rev*. 2014 Dec 13;12:CD001941.
27. Fayssoil A, Ritzenthaler T, Luis D, Hullin T, Clair B, Annane D, Orlikowski D. Be careful about abdominal discomfort in adult patients with muscular dystrophy. *Rev Neurol (Paris)*. 2014 Aug-Sep;170(8-9):548-50.
28. Lacombe M, Del Amo Castrillo L, Boré A, Chapeau D, Horvat E, Vaugier I, Lejaille M, Orlikowski D, Prigent H, Lofaso F. Comparison of three cough-augmentation techniques in neuromuscular patients: mechanical insufflation combined with manually assisted cough, insufflation-exsufflation alone and insufflation-exsufflation combined with manually assisted cough. *Respiration*. 2014;88(3):215-22.
29. Khirani S, Ramirez A, Delord V, Leroux K, Lofaso F, Hautot S, Toussaint M, Orlikowski D, Louis B, Fauroux B. Evaluation of ventilators for mouthpiece ventilation in neuromuscular disease. *Respir Care*. 2014 Sep;59(9):1329-37.
30. Rajabally YA, Durand MC, Mitchell J, Orlikowski D, Nicolas G. Electrophysiological diagnosis of Guillain-Barré syndrome subtype: could a single study suffice? *J Neurol Neurosurg Psychiatry*. 2015 Jan;86(1):115-9.
31. de Boysson H, Zuber M, Naggara O, Neau JP, Gray F, Bousser MG, Crassard I, Touzé E, Couraud PO, Kerschen P, Oppenheim C, Detante O, Faivre A, Gaillard N, Arquizan C, Bienvenu B, Néel A, Guillevin L, Pagnoux C; French Vasculitis Study Group and the French NeuroVascular Society. Primary angiitis of the central nervous system: description of the first

fifty-two adults enrolled in the French cohort of patients with primary vasculitis of the central nervous system. *Arthritis Rheumatol.* 2014 May;66(5):1315-26.

32. Fayssoil A, Nardi O, Annane D, Orlikowski D. Diastolic Function in Steinert's Disease. *Neurol Int.* 2014 Mar 31;6(1):5140.
33. Fayssoil A, Abdallah C, Orlikowski D. Left ventricular aneurysm in a patient with Duchenne muscular dystrophy. *Presse Med.* 2014 Jun;43(6 Pt 1):731-2.
34. Fayssoil A, Nardi O, Annane D, Orlikowski D. Left ventricular function in alpha-sarcoglycanopathy and gamma-sarcoglycanopathy. *Acta Neurol Belg.* 2014 Dec;114(4):257-9.
35. Fayssoil A, Nardi O, Annane D, Orlikowski D. Right ventricular function in late-onset Pompe disease. *J Clin Monit Comput.* 2014 Aug;28(4):419-21.
36. Fayssoil A, Nardi O, Annane D, Orlikowski D. Successful cardiac resynchronisation therapy in Duchenne muscular dystrophy: a 5-year follow-up. *Presse Med.* 2014 Mar;43(3):330-1.
37. Falaize L, Leroux K, Prigent H, Louis B, Khirani S, Orlikowski D, Fauroux B, Lofaso F. Battery life of portable home ventilators: effects of ventilator settings. *Respir Care.* 2014 Jul;59(7):1048-52.
38. Sivadon-Tardy V, Porcher R, Orlikowski D, Ronco E, Gault E, Roussi J, Durand MC, Sharshar T, Annane D, Raphael JC, Megraud F, Gaillard JL. Increased incidence of *Campylobacter jejuni*-associated Guillain-Barré syndromes in the Greater Paris area. *Epidemiol Infect.* 2014 Aug;142(8):1609-13.
39. Mayaud L, Congedo M, Van Laghenhove A, Orlikowski D, Figère M, Azabou E, Cheliout-Heraut F. A comparison of recording modalities of P300 event-related potentials (ERP) for brain-computer interface (BCI) paradigm. *Neurophysiol Clin.* 2013 Oct;43(4):217-27.
40. Lofaso F, Prigent H, Tiffreau V, Menoury N, Toussaint M, Monnier AF, Stremler N, Devaux C, Leroux K, Orlikowski D, Mauri C, Pin I, Sacconi S, Pereira C, Pépin JL, Fauroux B; Association Française Contre les Myopathies research group. Long-term mechanical ventilation equipment for neuromuscular patients: meeting the expectations of patients and prescribers. *Respir Care.* 2014 Jan;59(1):97-106.
41. Nicolas G, Annane D, Orlikowski D. [Acute inflammatory polyradiculoneuritis. Guillain Barré syndrome]. *Rev Prat.* 2013 Apr;63(4):573-9. French.
42. Fayssoil A, Amara W, Annane D, Orlikowski D. Wolff-Parkinson-White syndrome in Duchenne muscular dystrophy. *Int J Cardiol.* 2013 Aug 10;167(3):e53-4.
43. Fayssoil A, Nardi O, Orlikowski D, Annane D. Cardiac asynchrony in Duchenne muscular dystrophy. *J Clin Monit Comput.* 2013 Oct;27(5):587-9.
44. Quijano-Roy S, Avila-Smirnow D, Carlier RY; WB-MRI muscle study group. Whole body muscle MRI protocol: pattern recognition in early onset NM disorders. *Neuromuscul Disord.* 2012 Oct 1;22 Suppl 2:S68-84.
45. Fayssoil A, Orlikowski D, Nardi O, Pellegrini N, Annane D. Right ventricular function in Steinert's disease. *Int J Cardiol.* 2013 Jul 15;167(1):291.

46. Fayssoil A, Drouet T, Luis D, Orlikowski D, Alamowitch S, Annane D. Acute ischemic stroke in gamma-sarcoglycanopathy. *Presse Med.* 2013 Apr;42(4 Pt1):484-6.
47. Sharshar T, Polito A, Porcher R, Merhbene T, Blanc M, Antona M, Durand MC, Friedman D, Orlikowski D, Annane D, Marcadet MH. Relevance of anxiety in clinical practice of Guillain-Barre syndrome: a cohort study. *BMJ Open.* 2012 Aug 24;2(4). pii: e000893.
48. Prigent H, Orlikowski D, Laforêt P, Letilly N, Falaize L, Pellegrini N, Annane D, Raphael JC, Lofaso F. Supine volume drop and diaphragmatic function in adults with Pompe disease. *Eur Respir J.* 2012 Jun;39(6):1545-6.
49. Nardi J, Prigent H, Garnier B, Lebagry F, Quera-Salva MA, Orlikowski D, Lofaso F. Efficiency of invasive mechanical ventilation during sleep in Duchenne muscular dystrophy. *Sleep Med.* 2012 Sep;13(8):1056-65.
50. Fayssoil A, Nardi O, Orlikowski D, Annane D. [Heart involvement in sarcoglycanopathies]. *Rev Neurol (Paris).* 2012 Nov;168(11):779-82.
51. Nardi J, Prigent H, Adala A, Bohic M, Lebagry F, Quera-Salva MA, Orlikowski D, Lofaso F. Nocturnal oximetry and transcutaneous carbon dioxide in home-ventilated neuromuscular patients. *Respir Care.* 2012 Sep;57(9):1425-30.
52. Prigent H, Orlikowski D, Letilly N, Falaize L, Annane D, Sharshar T, Lofaso F. Vital capacity versus maximal inspiratory pressure in patients with Guillain-Barré syndrome and myasthenia gravis. *Neurocrit Care.* 2012 Oct;17(2):236-9.
53. Prigent H, Lejaille M, Terzi N, Annane D, Figere M, Orlikowski D, Lofaso F. Effect of a tracheostomy speaking valve on breathing-swallowing interaction. *Intensive Care Med.* 2012 Jan;38(1):85-90.

GNMH-Necker-Enfants Malades

1. Grotto S, Cuisset JM, Marret S, Drunat S, Faure P, Audebert-Bellanger S, Desguerre I, Flurin V, Grebille AG, Guerrot AM, Journal 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
2. Marey I, Ben Yaou R, Deburgrave N, Vasson A, Nectoux J, Leturcq F, Eymard B, Laforet P, Behin A, Stojkovic T, Mayer M, Tiffreau V, Desguerre I, Boyer FC, Nadaj-Pakleza A, Ferrer X, Wahbi K, Becane HM, Claustres M, Chelly J, Cossee M. Non Random Distribution of DMD Deletion Breakpoints and Implication of Double Strand Breaks Repair and Replication Error Repair Mechanisms. *J Neuromuscul Dis.* 2016 May 27;3(2):227-245

3. Bauché S, O'Regan S, Azuma Y, Laffargue F, McMacken G, Sternberg D, Brochier G, Buon C, Bouzidi N, Topf A, Lacène E, Remerand G, Beaufrere AM, Pebrel-Richard C, Thevenon J, El Chehadeh-Djebbar S, Faivre L, Duffourd Y, Ricci F, Mongini T, Fiorillo C, Astrea G, Burloiu CM, Butoianu N, Sandu C, Servais L, Bonne G, Nelson I, Desguerre I, Nougues MC, Bœuf B, Romero N, Laporte J, Boland A, Lechner D, Deleuze JF, Fontaine B, Stochlic L, Lochmuller H, Eymard B, Mayer M, Nicole S. Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. *Am J Hum Genet.* 2016 Sep 1;99(3):753-61
4. 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 arthrogyriposis, spinal stiffness and rare polyglucosan bodies in muscle. *Neuromuscul Disord.* 2016 Oct;26(10):681-687.
5. Gitiaux C, Chemaly N, Quijano-Roy S, Barnerias C, Desguerre I, Hully M, Chiron C, Dulac O, Nabbout R. Motor neuropathy contributes to crouching in patients with Dravet syndrome. *Neurology.* 2016 Jul 19;87(3):277-81.
6. Gitiaux C, De Antonio M, Aouizerate J, Gherardi RK, Guilbert T, Barnerias C, Bodemer C, Brochard-Payet K, Quartier P, Musset L, Chazaud B, Desguerre I, Bader-Meunier B. Vasculopathy-related clinical and pathological features are associated with severe juvenile dermatomyositis. *Rheumatology (Oxford).* 2016 Mar;55(3):470-9.
7. Aloui S, Khirani S, Ramirez A, Colella M, Louis B, Amaddeo A, Fauroux B. Whistle and cough pressures in children with neuromuscular disorders. *Respir Med.* 2016 Apr;113:28-36.
8. Desguerre I, Laugel [Diagnosis and natural history of Duchenne muscular dystrophy]. *V. Arch Pediatr.* 2015 Dec;22(12 Suppl 1):12S24-30
9. Chabrol B, Desguerre I. [Muscular dystrophies: From Duchenne to Becker]. *Arch Pediatr.* 2015 Dec;22(12 Suppl 1):12S1-2
10. Pinto-Mariz F, Rodrigues Carvalho L, Prufer De Queiroz Campos Araujo A, De Mello W, Gonçalves Ribeiro M, Cunha Mdo C, Cabello PH, Riederer I, Negroni E, Desguerre I, Veras M, Yada E, Allenbach Y, Benveniste O, Voit T, Mouly V, Silva-Barbosa SD, Butler-Browne G, Savino W. CD49d is a disease progression biomarker and a potential target for immunotherapy in Duchenne muscular dystrophy. *Skelet Muscle.* 2015 Dec 10;5:45.
11. Barnérias C, Bassez G, Schischmanoff O. [Chanarin-Dorfman syndrome in a 7-year-old child: when myopathy and skin involvement are all but one]. *Med Sci (Paris).* 2015 Nov;31
12. Latroche C, Gitiaux C, Chrétien F, Desguerre I, Mounier R, Chazaud B. *Physiology (Bethesda). Skeletal Muscle Microvasculature: A Highly Dynamic Lifeline.* 2015 Nov;30(6):417-27.
13. Hankiewicz K, Carlier RY, Lazaro L, Linzoain J, Barnerias C, Gómez-Andrés D, Avila-Smirnow D, Ferreira A, Estournet B, Guicheney P, Germain DP, Richard P, Bulacio S, Mompoin D, Quijano-Roy S. Whole-body muscle magnetic resonance imaging in SEPN1-related myopathy shows a homogeneous and recognizable pattern. *Muscle Nerve.* 2015 Nov;52(5):728-35
14. Frémond ML, Gitiaux C, Bonnet D, Guiddir T, Crow YJ, de Pontual L, Bader-Meunier B. Mosaic Tetrasomy 9p: A Mendelian Condition Associated With Pediatric-Onset Overlap Myositis. *Pediatrics.* 2015 Aug;136(2)

15. Gitiaux C, Blin-Rochemaure N, Hully M, Echaniz-Laguna A, Calmels N, Bahi-Buisson N, Desguerre I, Dabaj I, Wehbi S, Quijano-Roy S, Laugel V. Progressive demyelinating neuropathy correlates with clinical severity in Cockayne syndrome. *Clin Neurophysiol*. 2015 Jul;126(7)
16. Bourgeois F, Messéant J, Kordeli E, Petit JM, Delers P, Bahi-Buisson N, Bernard V, Sigoillot SM, Gitiaux C, Stouffer M, Francis F, Legay C. A critical and previously unsuspected role for doublecortin at the neuromuscular junction in mouse and human. *Neuromuscul Disord*. 2015 Jun;25(6):461-73.
17. Rutkowski A, Chatwin M, Koumbourlis A, Fauroux B, Simonds A; CMD Respiratory Physiology Consortium. 203rd ENMC international workshop: respiratory pathophysiology in congenital muscle disorders: implications for pro-active care and clinical research 13-15 December, 2013, Naarden, The Netherlands. *Neuromuscul Disord*. 2015 Apr;25(4):353-8.
18. Fauroux B, Quijano-Roy S, Desguerre I, Khirani S The value of respiratory muscle testing in children with neuromuscular disease.. *Chest*. 2015 Feb;147(2):552-9
19. Seferian AM, Moraux A, Annoussamy M, Canal A, Decostre V, Diebate O, Le Moing AG, Gidaro T, Deconinck N, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Maincent K, Desguerre I, Thémar-Noël C, Cuisset JM, Tiffreau V, Denis S, Jousten V, Quijano-Roy S, Voit T, Hogrel JY, Servais L. Upper limb strength and function changes during a one-year follow-up in non-ambulant patients with Duchenne Muscular Dystrophy: an observational multicenter trial. *PLoS One*. 2015 Feb 2;10(2)
20. Seferian AM, Moraux A, Annoussamy M, Canal A, Decostre V, Diebate O, Le Moing AG, Gidaro T, Deconinck N, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Maincent K, Desguerre I, Thémar-Noël C, Cuisset JM, Tiffreau V, Denis S, Jousten V, Quijano-Roy S, Voit T, Hogrel JY, Servais L. Upper limb strength and function changes during a one-year follow-up in non-ambulant patients with Duchenne Muscular Dystrophy: an observational multicenter trial. *PLoS One*. 2015 Feb 2;10(2)
21. Khirani S, Ramirez A, Olmo-Arroyo J, Amaddeo A, Quijano-Roy S, Desguerre I, Fauroux B. [Are respiratory muscle testing helpful to prompt sleep studies in children with neuromuscular disease?]. *Med Sci (Paris)*. 2015 Nov;31 Spec No 3:14-7.
22. Khirani S, Dabaj I, Amaddeo A, Ramirez A, Quijano-Roy S, Fauroux B. The value of respiratory muscle testing in a child with congenital muscular dystrophy. *Respirol Case Rep*. 2014 Sep;2(3):95-8.
23. Marttila M, Lehtokari VL, Marston S, Nyman TA, Barnerias C, Beggs AH, Bertini E, Ceyhan-Birsoy O, Cintas P, Gerard M, Gilbert-Dussardier B, Hogue JS, Longman C, Eymard B, Frydman M, Kang PB, Klinge L, Kolski H, Lochmüller H, Magy L, Manel V, Mayer M, Mercuri E, North KN, Peudener-Robert S, Pihko H, Probst FJ, Reisin R, Stewart W, Taratuto AL, de Visser M, Wilichowski E, Winer J, Nowak K, Laing NG, Winder TL, Monnier N, Clarke NF, Pelin K, Grönholm M, Wallgren-Pettersson C. Mutation update and genotype-phenotype correlations of novel and previously described mutations in TPM2 and TPM3 causing congenital myopathies. *Hum Mutat*. 2014 Jul;35(7):779-90.

24. Khirani S, Ramirez A, Delord V, Leroux K, Lofaso F, Hautot S, Toussaint M, Orlikowski D, Louis B, Fauroux B. Evaluation of ventilators for mouthpiece ventilation in neuromuscular disease. *Respir Care*. 2014 Sep;59(9):1329-37
25. Lofaso F, Prigent H, Tiffreau V, Menoury N, Toussaint M, Monnier AF, Stremier N, Devaux C, Leroux K, Orlikowski D, Mauri C, Pin I, Sacconi S, Pereira C, Pépin JL, Fauroux B; Association Française Contre les Myopathies research group. Long-term mechanical ventilation equipment for neuromuscular patients: meeting the expectations of patients and prescribers. *Respir Care*. 2014 Jan;59(1):97-106
26. Quijano-Roy S, Khirani S, Colella M, Ramirez A, Aloui S, Wehbi S, de Becdelievre A, Carlier RY, Allamand V, Richard P, Azzi V, Estournet B, Fauroux B. Diaphragmatic dysfunction in Collagen VI myopathies. *Neuromuscul Disord*. 2014 Feb;24(2):125-33
27. Fauroux B, Khirani S. Neuromuscular disease and respiratory physiology in children: putting lung function into perspective. *Respirology*. 2014 Aug;19(6):782-91.
28. Khirani S, Bersanini C, Aubertin G, Bachy M, Vialle R, Fauroux B. Non-invasive positive pressure ventilation to facilitate the post-operative respiratory outcome of spine surgery in neuromuscular children. *Eur Spine J*. 2014 Jul;23 Suppl 4:S406-11.
29. Khirani S, Ramirez A, Aubertin G, Boulé M, Chemouny C, Forin V, Fauroux B. Respiratory muscle decline in Duchenne muscular dystrophy. *Pediatr Pulmonol*. 2014 May;49(5):473-81
30. Barnérias C, Quijano S, Mayer M, Estournet B, Cuisset JM, Sukno S, Peudenier S, Laroche C, Chabrier S, Sabouraud P, Vuillerot C, Chabrol B, Halbert C, Cancès C, Beze-Beyrie P, Ledivenah A, Viallard ML, Desguerre I. [Multicentric study of medical care and practices in spinal muscular atrophy type 1 over two 10-year periods]. *Arch Pediatr*. 2014 Apr;21(4):347-54.
31. Laquérière, A ; Maluenda, J ; Camus, A ; Fontenas, L ; Dieterich, K ; Nolent, F & al. Mutations in CNTNAP1 and ADCY6 are responsible for severe arthrogryposis multiplex congenita with axoglial defects. *Hum. Mol. Genet*. 2014.
32. Bönnemann CG, Wang CH, Quijano-Roy S, Deconinck N, Bertini E, Ferreira A, Muntoni F, Sewry C, Bérout C, Mathews KD, Moore SA, Bellini J, Rutkowski A, North KN Diagnostic approach to the congenital muscular dystrophies.; Members of International Standard of Care Committee for Congenital Muscular Dystrophies. *Neuromuscul Disord*. 2014 Apr;24(4):289-311.
33. Bertrand AT, Ziaei S, Ehret C, Duchemin H, Mamchaoui K, Bigot A, Mayer M, Quijano-Roy S, Desguerre I, Lainé J, Ben Yaou R, Bonne G, Coirault C Cellular microenvironments reveal defective mechanosensing responses and elevated YAP signaling in LMNA-mutated muscle precursors.. *J Cell Sci*. 2014 Jul 1;127(Pt 13):2873-84
34. Rul B, Quijano-Roy S, Golse A, Beynier D, Estournet B, Desguerre I, Barnerias C, Herve C. [What a tracheostomy changes in a child with a neuromuscular disease]. *Rech Soins Infirm*. 2013 Sep;(114):46-57. French.
35. Gitiaux C, Kostallari E, Lafuste P, Authier FJ, Christov C, Gherardi RK. Whole microvascular unit deletions in dermatomyositis. *Ann Rheum Dis*. 2013 Mar;72(3):445-52

36. Khirani S, Colella M, Caldarelli V, Aubertin G, Boulé M, Forin V, Ramirez A, Fauroux B. Longitudinal course of lung function and respiratory muscle strength in spinal muscular atrophy type 2 and 3. *Eur J Paediatr Neurol*. 2013 Nov;17(6):552-60.
37. Gitiaux C, Bergounioux J, Magen M, Quijano-Roy S, Blanc T, Bonnefont JP, Desguerre I. Diaphragmatic weakness with progressive sensory and motor polyneuropathy: case report of a neonatal IGHMBP2-related neuropathy. *J Child Neurol*. 2013 Jun;28(6):787-90
38. Mercier S, Toutain A, Toussaint A, Raynaud M, de Barace C, Marcorelles P, Pasquier L, Blayau M, Espil C, Parent P, Journal H, Lazaro L, Andoni Urtizberea J, Moerman A, Faivre L, Eymard B, Maincent K, Gherardi R, Chaigne D, Ben Yaou R, Leturcq F, Chelly J, Desguerre I. Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. *Eur J Hum Genet*. 2013 Aug;21(8):855-63.
39. Chelly J, Desguerre I. Progressive muscular dystrophies. *Handb Clin Neurol*. 2013;113:1343-66.
40. Servais L, Deconinck N, Moraux A, Benali M, Canal A, Van Parys F, Vereecke W, Wittevrongel S, Mayer M, Desguerre I, Maincent K, Themar-Noel C, Quijano-Roy S, Serari N, Voit T, Hogrel JY. Innovative methods to assess upper limb strength and function in non-ambulant Duchenne patients. *Neuromuscul Disord*. 2013 Feb;23(2):139-48.
41. Fardeau M, Desguerre I. Diagnostic workup for neuromuscular diseases. *Handb Clin Neurol*. 2013;113:1291-7
42. Desguerre I, Arnold L, Vignaud A, Cuvellier S, Yacoub-Youssef H, Gherardi RK, Chelly J, Chretien F, Mounier R, Ferry A, Chazaud B. A new model of experimental fibrosis in hindlimb skeletal muscle of adult mdx mouse mimicking muscular dystrophy. *Muscle Nerve*. 2012 Jun;45(6):803-14.
43. Renaldo F, Amati-Bonneau P, Slama A, Romana C, Forin V, Doummar D, Barnerias C, Bursztyn J, Mayer M, Khouri N, Billette de Villemeur T, Burglen L, Reynier P, Bernabe Gelot A, Rodriguez D. MFN2, a new gene responsible for mitochondrial DNA depletion. *Brain*. 2012 Aug;135(Pt 8):e223
44. Bader-Meunier B, Monnet D, Barnerias C, Halphen I, Lambot-Juhan K, Chalumeau M, Costedoat-Chalumeau N, Ribeil JA, Bodemer C, Gherardi R. Thrombotic microangiopathy and Purtscher-like retinopathy as a rare presentation of juvenile dermatomyositis. *Pediatrics*. 2012 Mar;129(3):e821-4
45. Bader-Meunier B, Decaluwe H, Barnerias C, Gherardi R, Quartier P, Faye A, Guignonis V, Pagnier A, Brochard K, Sibilia J, Gottenberg JE, Bodemer C. Safety and efficacy of rituximab in severe juvenile dermatomyositis: results from 9 patients from the French Autoimmunity and Rituximab registry; Club Rhumatismes et Inflammation. *J Rheumatol*. 2011 Jul;38(7):1436-40

1. Gherardi RK, Aouizerate J, Cadusseau J, Yara S, Authier FJ. Aluminum adjuvants of vaccines injected into the muscle: Normal fate, pathology and associated disease. *Morphologie*. 2016 Mar 2. pii: S1286-0115(16)00025-4.
2. Aouizerate J, Valleyrie-Allanore L, Limal N, Ayache SS, Gherardi RK, Audard V, Authier FJ. Ischemic myopathy revealing systemic calciphylaxis. *Muscle Nerve*. 2016 Dec 9. doi: 10.1002/mus.25505.
3. Van Der Gucht A, Aoun-Sebaiti M, Kouv P, Guedj E, Aouizerate J, Verger A, Gherardi RK, Bachoud-Levi AC, Authier FJ, Itti E. FDG-PET/CT Brain Findings in a Patient With Macrophagic Myofasciitis. *Nucl Med Mol Imaging*. 2016 Mar;50(1):80-4
4. Dogan C, De Antonio M, Hamroun D, Varet H, Fabbro M, Rougier F, Amarof K, Arne Bes MC, Bedat-Millet AL, Behin A, Bellance R, Bouhour F, Boutte C, Boyer F, Campana-Salort E, Chapon F, Cintas P, Desnuelle C, Deschamps R, Drouin-Garraud V, Ferrer X, Gervais-Bernard H, Ghorab K, Laforet P, Magot A, Magy L, Menard D, Minot MC, Nadaj-Pakleza A, Pellieux S, Peroon Y, Preudhomme M, Pouget J, Sacconi S, Sole G, Stojkovich T, Tiffreau V, Urtizbera A, Vial C, Zagnoli F, Caranhac G, Bourlier C, Riviere G, Geille A, Gherardi RK, Eymard B, Puymirat J, Katsahian S, Bassez G. Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1 Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional Observational Study. *PLoS One*. 2016 Feb 5;11(2):e0148264.
5. Vuong V, Duong TA, Aouizerate J, Authier FJ, Ingen-Housz-Oro S, Valeyrie-Allanore L, Ortonne N, Wolkenstein P, Gherardi RK, Chosidow O, Cosnes A, Sbidian E. Dermatomyositis: factors predicting relapse. *J Eur Acad Dermatol Venereol*. 2016 May;30(5):813-8.
6. Griger Z, Nagy-Vincze M, Bodoki L, Gherardi RK, Dankó K, Hortobágyi T. Late onset dysferlinopathy mimicking treatment resistant polymyositis. *Joint Bone Spine*. 2016 May;83(3):355-6. doi: 10.1016/j.jbspin.2015.03.017.
7. Gitiaux C, De Antonio M, Aouizerate J, Gherardi RK, Guilbert T, Barnerias C, Bodemer C, Brochard-Payet K, Quartier P, Musset L, Chazaud B, Desguerre I, Bader-Meunier B. Vasculopathy-related clinical and pathological features are associated with severe juvenile dermatomyositis. *Rheumatology (Oxford)*. 2016 Mar;55(3):470-9.
8. Béhin A, Acquaviva-Bourdain C, Souvannanorath S, Streichenberger N, Attarian S, Bassez G, Brivet M, Fouilhoux A, Labarre-Villa A, Laquerrière A, Pérard L, Kaminsky P, Pouget J, Rigal O, Vanhulle C, Eymard B, Vianey-Saban C, Laforêt P. Multiple acyl-CoA dehydrogenase deficiency (MADD) as a cause of late-onset treatable metabolic disease. *Rev Neurol (Paris)*. 2016 Mar;172(3):231-41.
9. Wahbi K, Sebag FA, Lellouche N, Lazarus A, Bécane HM, Bassez G, Stojkovic T, Fayssoil A, Laforêt P, Béhin A, Meune C, Eymard B, Duboc D. Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. *Neuromuscul Disord*. 2016 Mar;26(3):227-33.
10. Van Der Gucht A, Aoun-Sebaiti M, Kouv P, Guedj E, Aouizerate J, Verger A, Gherardi RK, Bachoud-Levi AC, Authier FJ, Itti E. FDG-PET/CT Brain Findings in a Patient With Macrophagic Myofasciitis. *Nucl Med Mol Imaging*. 2016 Mar;50(1):80-4.
11. Dogan C, De Antonio M, Hamroun D, Varet H, Fabbro M, Rougier F, Amarof K, Arne Bes MC, Bedat-Millet AL, Behin A, Bellance R, Bouhour F, Boutte C, Boyer F, Campana-Salort E,

Chapon F, Cintas P, Desnuelle C, Deschamps R, Drouin-Garraud V, Ferrer X, Gervais-Bernard H, Ghorab K, Laforet P, Magot A, Magy L, Menard D, Minot MC, Nadaj-Pakleza A, Pellieux S, Pereon Y, Preudhomme M, Pouget J, Sacconi S, Sole G, Stojkovich T, Tiffreau V, Urtizbera A, Vial C, Zagnoli F, Caranhac G, Bourlier C, Riviere G, Geille A, Gherardi RK, Eymard B, Puymirat J, Katsahian S, Bassez G. Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1 Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional Observational Study. *PLoS One*. 2016 Feb 5;11(2):e0148264.

12. Vuong V, Duong TA, Aouizerate J, Authier FJ, Ingen-Housz-Oro S, Valeyrie-Allanore L, Ortonne N, Wolkenstein P, Gherardi RK, Chosidow O, Cosnes A, Sbidian E. Dermatomyositis: factors predicting relapse. *J Eur Acad Dermatol Venereol*. 2016 May;30(5):813-8.
13. Griger Z, Nagy-Vincze M, Bodoki L, Gherardi RK, Dankó K, Hortobágyi T. Late onset dysferlinopathy mimicking treatment resistant polymyositis. *Joint Bone Spine*. 2016 May;83(3):355-6. doi: 10.1016/j.jbspin.2015.03.017.
14. Béhin A, Acquaviva-Bourdain C, Souvannanorath S, Streichenberger N, Attarian S, Bassez G, Brivet M, Fouilhoux A, Labarre-Villa A, Laquerrière A, Pérard L, Kaminsky P, Pouget J, Rigal O, Vanhulle C, Eymard B, Vianey-Saban C, Laforêt P. Multiple acyl-CoA dehydrogenase deficiency (MADD) as a cause of late-onset treatable metabolic disease. *Rev Neurol (Paris)*. 2016 Mar;172(3):231-41.
15. Wahbi K, Sebag FA, Lellouche N, Lazarus A, Bécane HM, Bassez G, Stojkovic T, Fayssoil A, Laforêt P, Béhin A, Meune C, Eymard B, Duboc D. Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. *Neuromuscul Disord*. 2016 Mar;26(3):227-33.
16. Dogan C, Puymirat J, Bassez G. [DM-SCOPE, an intermediary appraisal report and benefits of databases in neuromuscular disorders]. *Med Sci (Paris)*. 2015 Nov;31 Spec No 3:18-9.
17. Barnérias C, Bassez G, Schischmanoff O. [Chanarin-Dorfman syndrome in a 7-year-old child: when myopathy and skin involvement are all but one]. *Med Sci (Paris)*. 2015 Nov;31 Spec No 3:11-3.
18. Laustriat D, Gide J, Barrault L, Chautard E, Benoit C, Auboeuf D, Boland A, Battail C, Artiguenave F, Deleuze JF, Bénit P, Rustin P, Franc S, Charpentier G, Furling D, Bassez G, Nissan X, Martinat C, Peschanski M, Baghdoyan S. In Vitro and In Vivo Modulation of Alternative Splicing by the Biguanide Metformin. *Mol Ther Nucleic Acids*. 2015 Nov 3;4:e262.
19. Dany A, Barbe C, Rapin A, Réveillère C, Hardouin JB, Morrone I, Wolak-Thierry A, Dramé M, Calmus A, Sacconi S, Bassez G, Tiffreau V, Richard I, Gallais B, Prigent H, Taiar R, Jolly D, Novella JL, Boyer FC. Construction of a Quality of Life Questionnaire for slowly progressive neuromuscular disease. *Qual Life Res*. 2015 Nov;24(11):2615-23.
20. van Engelen B; OPTIMISTIC Consortium. Cognitive behaviour therapy plus aerobic exercise training to increase activity in patients with myotonic dystrophy type 1 (DM1) compared to usual care (OPTIMISTIC): study protocol for randomised controlled trial. *Trials*. 2015 May 23;16:224.
21. Bouchard JP, Cossette L, Bassez G, Puymirat J. Natural history of skeletal muscle involvement in myotonic dystrophy type 1: a retrospective study in 204 cases. *J Neurol*. 2015 Feb;262(2):285-93.

22. Mercier S, Küry S, Salort-Campana E, Magot A, Agbim U, Besnard T, Bodak N, Bou-Hanna C, Bréhéret F, Brunelle P, Caillon F, Chabrol B, Cormier-Daire V, David A, Eymard B, Faivre L, Figarella-Branger D, Fleurence E, Ganapathi M, Gherardi R, Goldenberg A, Hamel A, Igual J, Irvine AD, Israël-Biet D, Kannengiesser C, Laboisse C, Le Caignec C, Mahé JY, Mallet S, MacGowan S, McAleer MA, McLean I, Méni C, Munnich A, Mussini JM, Nagy PL, Odel J, O'Regan GM, Péréon Y, Perrier J, Piard J, Puzenat E, Sampson JB, Smith F, Soufir N, Tanji K, Thauvin C, Ulane C, Watson RM, Khumalo NP, Mayosi BM, Barbarot S, Bézieau S. Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. *Orphanet J Rare Dis*. 2015 Oct 15;10:135.
23. Gitiaux C, De Antonio M, Aouizerate J, Gherardi RK, Guilbert T, Barnerias C, Bodemer C, Brochard-Payet K, Quartier P, Musset L, Chazaud B, Desguerre I, Bader-Meunier B. Vasculopathy-related clinical and pathological features are associated with severe juvenile dermatomyositis. *Rheumatology (Oxford)*. 2016 Mar;55(3):470-9.
24. Crépeaux G, Eidi H, David MO, Tzavara E, Giros B, Exley C, Curmi PA, Shaw CA, Gherardi RK, Cadusseau J. Highly delayed systemic translocation of aluminum-based adjuvant in CD1 mice following intramuscular injections. *J Inorg Biochem*. 2015 Nov;152:199-205.
25. Mescam-Mancini L, Allenbach Y, Hervier B, Devilliers H, Mariampillay K, Dubourg O, Maisonobe T, Gherardi R, Mezin P, Preusse C, Stenzel W, Benveniste O. Anti-Jo-1 antibody-positive patients show a characteristic necrotizing perifascicular myositis. *Brain*. 2015 Sep;138(Pt 9):2485-92.
26. Eidi H, David MO, Crépeaux G, Henry L, Joshi V, Berger MH, Sennour M, Cadusseau J, Gherardi RK, Curmi PA. Fluorescent nanodiamonds as a relevant tag for the assessment of alum adjuvant particle biodisposition. *BMC Med*. 2015 Jun 17;13:144. doi: 10.1186/s12916-015-0388-2.
27. Van Der Gucht A, Aoun Sebaiti M, Itti E, Aouizerate J, Evangelista E, Chalaye J, Gherardi RK, Ragunathan-Thangarajah N, Bachoud-Levi AC, Authier FJ. Neuropsychological Correlates of Brain Perfusion SPECT in Patients with Macrophagic Myofasciitis. *PLoS One*. 2015 Jun 1;10(6):e0128353.
28. Kostallari E, Baba-Amer Y, Alonso-Martin S, Ngoh P, Relaix F, Lafuste P, Gherardi RK. Pericytes in the myovascular niche promote post-natal myofiber growth and satellite cell quiescence. *Development*. 2015 Apr 1;142(7):1242-53.
29. Preusse C, Goebel HH, Pehl D, Rinnenthal JL, Kley RA, Allenbach Y, Heppner FL, Vorgerd M, Authier FJ, Gherardi R, Stenzel W. Th2-M2 immunity in lesions of muscular sarcoidosis and macrophagic myofasciitis. *Neuropathol Appl Neurobiol*. 2015 Dec;41(7):952-63.
30. Gherardi RK, Eidi H, Crépeaux G, Authier FJ, Cadusseau J. Biopersistence and brain translocation of aluminum adjuvants of vaccines. *Front Neurol*. 2015 Feb 5;6:4.
31. De Bleecker JL, De Paepe B, Aronica E, de Visser M; ENMC Myositis Muscle.
32. Biopsy Study Group, Amato A, Aronica E, Benveniste O, De Bleecker J, de Boer O, De Paepe B, de Visser M, Dimachkie M, Gherardi R, Goebel HH, Hilton-Jones D, Holton J, Lundberg IE, Mammen A, Mastaglia F, Nishino I, Rushing E, Schroder HD, Selcen D, Stenzel W. 205th ENMC International Workshop: Pathology diagnosis of idiopathic inflammatory myopathies

part II 28-30 March 2014, Naarden, The Netherlands. *Neuromuscul Disord.* 2015 Mar;25(3):268-72.

33. Rigolet M, Aouizerate J, Couette M, Ragunathan-Thangarajah N, Aoun-Sebaiti M, Gherardi RK, Cadusseau J, Authier FJ. Clinical features in patients with long-lasting macrophagic myofasciitis. *Front Neurol.* 2014 Nov 28;5:230.
34. Aouizerate J, De Antonio M, Bassez G, Gherardi RK, Berenbaum F, Guillevin L, Berezne A, Valeyre D, Maisonobe T, Dubourg O, Cosnes A, Benveniste O, Authier FJ. Myofiber HLA-DR expression is a distinctive biomarker for antisynthetase-associated myopathy. *Acta Neuropathol Commun.* 2014 Oct 23;2:154.
35. Drouot L, Allenbach Y, Jouen F, Charuel JL, Martinet J, Meyer A, Hirschberger O, Bader-Meunier B, Kone-Paut I, Campana-Salort E, Eymard B, Tournadre A, Musset L, Sibilia J, Marie I, Benveniste O, Boyer O; French Myositis Network [CN]. Exploring necrotizing autoimmune myopathies with a novel immunoassay for anti-3-hydroxy-3-methyl-glutaryl-CoA reductase autoantibodies. *Arthritis Res Ther.* 2014 Feb 3;16(1):R39.
36. Drouot L, Allenbach Y, Jouen F, Charuel JL, Martinet J, Meyer A, Hirschberger O, Bader-Meunier B, Kone-Paut I, Campana-Salort E, Eymard B, Tournadre A, Musset L, Sibilia J, Marie I, Benveniste O, Boyer O; French Myositis Network [CN]. Exploring necrotizing autoimmune myopathies with a novel immunoassay for anti-3-hydroxy-3-methyl-glutaryl-CoA reductase autoantibodies. *Arthritis Res Ther.* 2014 Feb 3;16(1):R39.
37. Quartier P, Gherardi RK. Juvenile dermatomyositis. *Handb Clin Neurol.* 2013;113:1457-63. doi: 10.1016/B978-0-444-59565-2.00014-9. Review.
38. Cadusseau J, Ragunathan-Thangarajah N, Surenaud M, Hue S, Authier FJ, Gherardi RK. Selective elevation of circulating CCL2/MCP1 levels in patients with longstanding post-vaccinal macrophagic myofasciitis and ASIA. *Curr Med Chem.* 2014;21(4):511-7.
39. Gherardi RK, Cadusseau J, Authier FJ. [Biopersistence and systemic distribution of intramuscularly injected particles: what impact on long-term tolerability of alum adjuvants?]. *Bull Acad Natl Med.* 2014 Jan;198(1):37-48; discussion 49-53.
40. Mercier S, Küry S, Shaboodien G, Houniet DT, Khumalo NP, Bou-Hanna C, Bodak N, Cormier-Daire V, David A, Faivre L, Figarella-Branger D, Gherardi RK, Glen E, Hamel A, Laboisie C, Le Caignec C, Lindenbaum P, Magot A, Munnich A, Mussini JM, Pillay K, Rahman T, Redon R, Salort-Campana E, Santibanez-Koref M, Thauvin C, Barbarot S, Keavney B, Bézieau S, Mayosi BM. Mutations in FAM111B cause hereditary fibrosing poikiloderma with tendon contracture, myopathy, and pulmonary fibrosis. *Am J Hum Genet.* 2013 Dec 5;93(6):1100-7.
41. De Bleeker JL, Lundberg IE, de Visser M; ENMC Myositis Muscle Biopsy Study Group. 193rd ENMC International workshop Pathology diagnosis of idiopathic inflammatory myopathies 30 November - 2 December 2012, Naarden, The Netherlands. *Neuromuscul Disord.* 2013 Nov;23(11):945-51.
42. Ragunathan-Thangarajah N, Le Beller C, Boutouyrie P, Bassez G, Gherardi RK, Laurent S, Authier FJ. Distinctive clinical features in arthro-myalgic patients with and without aluminum hydroxyde-induced macrophagic myofasciitis: an exploratory study. *J Inorg Biochem.* 2013 Nov;128:262-6.

43. Virgone-Carlotta A, Uhlrich J, Akram MN, Ressenkoff D, Chrétien F, Domenget C, Gherardi R, Despars G, Jurdic P, Honnorat J, Nataf S, Touret M. Mapping and kinetics of microglia/neuron cell-to-cell contacts in the 6-OHDA murine model of Parkinson's disease. *Glia*. 2013 Oct;61(10):1645-58.
44. Biondi O, Villemeur M, Marchand A, Chretien F, Bourg N, Gherardi RK, Richard I, Authier FJ. Dual effects of exercise in dysferlinopathy. *Am J Pathol*. 2013 Jun;182(6):2298-309.
45. Khan Z, Combadière C, Authier FJ, Itier V, Lux F, Exley C, Mahrouf-Yorgov M, Decrouy X, Moretto P, Tillement O, Gherardi RK, Cadusseau J. Slow CCL2-dependent translocation of biopersistent particles from muscle to brain. *BMC Med*. 2013 Apr 4;11:99.
46. Mercier S, Toutain A, Toussaint A, Raynaud M, de Barace C, Marcorelles P, Pasquier L, Blayau M, Espil C, Parent P, Journal H, Lazaro L, Andoni Urtizberea J, Moerman A, Faivre L, Eymard B, Maincent K, Gherardi R, Chaigne D, Ben Yaou R, Leturcq F, Chelly J, Desguerre I. Genetic and clinical specificity of 26 symptomatic carriers for dystrophinopathies at pediatric age. *Eur J Hum Genet*. 2013 Aug;21(8):855-63.
47. Gitiaux C, Kostallari E, Lafuste P, Authier FJ, Christov C, Gherardi RK. Whole microvascular unit deletions in dermatomyositis. *Ann Rheum Dis*. 2013 Mar;72(3):445-52.
48. Sène D, Cacoub P, Authier FJ, Haroche J, Créange A, Saadoun D, Amoura Z, Guillausseau PJ, Lefaucheur JP. Sjögren Syndrome-Associated Small Fiber Neuropathy: Characterization From a Prospective Series of 40 Cases. *Medicine (Baltimore)*. 2013 Aug 26.
49. Fréret M, Drouot L, Obry A, Ahmed-Lacheheb S, Dauly C, Adriouch S, Cosette P, Authier FJ, Boyer O. Overexpression of MHC class I in muscle of lymphocyte-deficient mice causes a severe myopathy with induction of the unfolded protein response. *Am J Pathol*. 2013 Sep;183(3):893-904.
50. Yiou R, Hogrel JY, Loche CM, Authier FJ, Lecorvoisier P, Jouany P, Roudot-Thoraval F, Lefaucheur JP. Periurethral skeletal myofibre implantation in patients with urinary incontinence and intrinsic sphincter deficiency: a phase I clinical trial. *BJU Int*. 2013 Jun;111(7):1105-16.
51. Bachinski LL, Baggerly KA, Neubauer VL, Nixon TJ, Raheem O, Sirito M, Unruh AK, Zhang J, Nagarajan L, Timchenko LT, Bassez G, Eymard B, Gamez J, Ashizawa T, Mendell JR, Udd B, Krahe R. Most expression and splicing changes in myotonic dystrophy type 1 and type 2 skeletal muscle are shared with other muscular dystrophies. *Neuromuscul Disord*. 2014 Mar;24(3):227-40.
52. Vuillerot C, Rippert P, Roche S, Bérard C, Margirier F, de Lattre C, Poirot I, Berruyer A, Tiffreau V, Fournier-Mehouas M, Bouhour F, Urtizberea JA, Renders A, Ecochard R; Le groupe d'étude NM-Score. Development and validation of a motor function classification in patients with neuromuscular disease: the NM-score. *Ann Phys Rehabil Med*. 2013 Dec;56(9-10):673-86.
53. Echenne B, Bassez G. Congenital and infantile myotonic dystrophy. *Handb Clin Neurol*. 2013;113:1387-93.
54. Hernández-Hernández O, Guiraud-Dogan C, Sicot G, Huguet A, Luillier S, Steidl E, Saenger S, Marciniak E, Obriot H, Chevarin C, Nicole A, Revillod L, Charizanis K, Lee KY, Suzuki Y, Kimura T, Matsuura T, Cisneros B, Swanson MS, Trovero F, Buisson B, Bizot JC, Hamon M, Humez S,

Bassez G, Metzger F, Buée L, Munnich A, Sergeant N, Gourdon G, Gomes-Pereira M. Myotonic dystrophy CTG expansion affects synaptic vesicle proteins, neurotransmission and mouse behaviour. *Brain*. 2013 Mar;136(Pt 3):957-70.

55. Laforêt P, Stojkovic T, Bassez G, Carlier PG, Clément K, Wahbi K, Petit FM, Eymard B, Carlier RY. Neutral lipid storage disease with myopathy: a whole-body nuclear MRI and metabolic study. *Mol Genet Metab*. 2013 Feb;108(2):125-31.
56. Huguet A, Medja F, Nicole A, Vignaud A, Guiraud-Dogan C, Ferry A, Decostre V, Hogrel JY, Metzger F, Hoeflich A, Baraibar M, Gomes-Pereira M, Puymirat J, Bassez G, Furling D, Munnich A, Gourdon G. Molecular, physiological, and motor performance defects in DMSXL mice carrying >1,000 CTG repeats from the human DM1 locus. *PLoS Genet*. 2012;8(11):e1003043.
57. Boërio D, Lefaucheur JP, Bassez G, Hogrel JY. Central and peripheral components of exercise-related fatigability in myotonic dystrophy type 1. *Acta Neurol Scand*. 2012 Jan;125(1):38-46.
58. Liao H, Franck E, Fréret M, Adriouch S, Baba-Amer Y, Authier FJ, Boyer O, Gherardi RK. Myoinjury transiently activates muscle antigen-specific CD8+ T cells in lymph nodes in a mouse model. *Arthritis Rheum*. 2012 Oct;64(10):3441-51.
59. Desguerre I, Arnold L, Vignaud A, Cuvellier S, Yacoub-Youssef H, Gherardi RK, Chelly J, Chretien F, Mounier R, Ferry A, Chazaud B. A new model of experimental fibrosis in hindlimb skeletal muscle of adult mdx mouse mimicking muscular dystrophy. *Muscle Nerve*. 2012 Jun;45(6):803-14.
60. Bader-Meunier B, Monnet D, Barnerias C, Halphen I, Lambot-Juhan K, Chalumeau M, Costedoat-Chalumeau N, Ribeil JA, Bodemer C, Gherardi R. Thrombotic microangiopathy and Purtscher-like retinopathy as a rare presentation of juvenile dermatomyositis. *Pediatrics*. 2012 Mar;129(3):e821-4.
61. Gherardi RK, Authier FJ. Macrophagic myofasciitis: characterization and pathophysiology. *Lupus*. 2012 Feb;21(2):184-9.
62. Liao H, Franck E, Fréret M, Adriouch S, Baba-Amer Y, Authier FJ, Boyer O, Gherardi RK. Myoinjury transiently activates muscle antigen-specific CD8+ T cells in lymph nodes in a mouse model. *Arthritis Rheum*. 2012 Oct;64(10):3441-51.
63. Lesault PF, Theret M, Magnan M, Cuvellier S, Niu Y, Gherardi RK, Tremblay JP, Hittinger L, Chazaud B. Macrophages improve survival, proliferation and migration of engrafted myogenic precursor cells into MDX skeletal muscle. *PLoS One*. 2012;7(10):e46698.
64. Cohen C, Mekinian A, Saidenberg-Kermanac'h N, Stirnemann J, Fenaux P, Gherardi R, Fain O. Efficacy of tocilizumab in rituximab-refractory cryoglobulinemia vasculitis. *Ann Rheum Dis*. 2012 Apr;71(4):628-9.

sous-section bibliographie amylose

1. Gillmore JD, Maurer MS, Falk RH, Merlini G, Damy T, Dispenzieri A, Wechalekar AD, Berk JL, Quarta CC, Grogan M, Lachmann HJ, Bokhari S, Castano A, Dorbala S, Johnson GB,

- Glaudemans AW, Rezk T, Fontana M, Palladini G, Milani P, Guidalotti PL, Flatman K, Lane T, Vonberg FW, Whelan CJ, Moon JC, Ruberg FL, Miller EJ, Hutt DF, Hazenberg BP, Rapezzi C, Hawkins PN. Non-Biopsy Diagnosis of Cardiac Transthyretin Amyloidosis. *Circulation*. 2016 Apr 22.
2. Bodez D, Guellich A, Kharoubi M, Covali-Noroc A, Tissot CM, Guendouz S, Hittinger L, Dubois-Randé JL, Lefaucheur JP, Planté-Bordeneuve V, Adnot S, Boyer L, Damy T. Prevalence, Severity, and Prognostic value of Sleep Apnea Syndromes in Cardiac Amyloidosis. *Sleep*. 2016 Apr 12.
 3. Galat A, Guellich A, Bodez D, Slama M, Dijos M, Zeitoun DM, Milleron O, Attias D, Dubois-Randé JL, Mohty D, Audureau E, Teiger E, Rosso J, Monin JL, Damy T. Aortic stenosis and transthyretin cardiac amyloidosis: the chicken or the egg? *Eur Heart J*. 2016 Feb 22.
 4. Damy T, Maurer MS, Rapezzi C, Planté-Bordeneuve V, Karayal ON, Mundayat R, Suhr OB, Kristen AV.
 5. Clinical, ECG and echocardiographic clues to the diagnosis of TTR-related cardiomyopathy. *Open Heart*. 2016 Feb 8;3(1):
 6. Ternacle J, Bodez D, Guellich A, Audureau E, Rappeneau S, Lim P, Radu C, Guendouz S, Couetil JP, Benhaiem N, Hittinger L, Dubois-Randé JL, Plante-Bordeneuve V, Mohty D, Deux JF, Damy T. Causes and Consequences of Longitudinal LV Dysfunction Assessed by 2D Strain Echocardiography in Cardiac Amyloidosis. *JACC Cardiovasc Imaging*. 2016 Feb;9(2):126-38.
 7. Damy T, Costes B, Hagège AA, Donal E, Eicher JC, Slama M, Guellich A, Rappeneau S, Gueffet JP, Logeart D, Planté-Bordeneuve V, Bouvaist H, Huttin O, Mulak G, Dubois-Randé JL, Goossens M, Canoui-Poitrine F, Buxbaum JN. Prevalence and clinical phenotype of hereditary transthyretin amyloid cardiomyopathy in patients with increased left ventricular wall thickness. *Eur Heart J*. 2015 Nov 3.
 8. Damy T, Bodez D, Habibi A, Guellich A, Rappeneau S, Inamo J, Guendouz S, Gellen-Dautremer J, Pissard S, Loric S, Wagner-Ballon O, Godeau B, Adnot S, Dubois-Randé JL, Hittinger L, Galactéros F, Bartolucci P. Haematological determinants of cardiac involvement in adults with sickle cell disease. *Eur Heart J*. 2016 Apr 7;37(14):1158-67.
 9. Galat A, Rosso J, Guellich A, Van Der Gucht A, Rappeneau S, Bodez D, Guendouz S, Tissot CM, Hittinger L, Dubois-Randé JL, Plante-Bordeneuve V, Itti E, Meignan M, Damy T. Usefulness of (99m)Tc-HMDP scintigraphy for the etiologic diagnosis and prognosis of cardiac amyloidosis. *Amyloid*. 2015;22(4):210-20.
 10. Van Der Gucht A, Galat A, Rosso J, Guellich A, Garot J, Bodez D, Plante-Bordeneuve V, Hittinger L, Dubois-Randé JL, Evangelista E, Sasanelli M, Chalaye J, Meignan M, Itti E, Damy T. [18F]-NaF PET/CT imaging in cardiac amyloidosis. *J Nucl Cardiol*. 2015 Sep 24.
 11. Ng Wing Tin S, Planté-Bordeneuve V, Salhi H, Goujon C, Damy T, Lefaucheur JP. Characterization of Pain in Familial Amyloid Polyneuropathy. *J Pain*. 2015 Nov;16(11):1106-14.
 12. Galat A, Van Der Gucht A, Colombat M, Attias D, Itti E, Meignan M, Lebras F, Molinier-Frenkel V, Benhaiem N, Guellich A, Rosso J, Damy T. (99m)Tc-HMDP scintigraphy rectifies wrong diagnosis of AL amyloidosis. *J Nucl Cardiol*. 2015 Aug;22(4):853-7.

13. Damy T, Judge DP, Kristen AV, Berthet K, Li H, Aarts J. Cardiac findings and events observed in an open-label clinical trial of tafamidis in patients with non-Val30Met and non-Val122Ile hereditary transthyretin amyloidosis. *J Cardiovasc Transl Res*. 2015 Mar;8(2):117-27.
14. Deux JF, Mihalache CI, Legou F, Damy T, Mayer J, Rappeneau S, Planté-Bordeneuve V, Luciani A, Kobeiter H, Rahmouni A. Noninvasive detection of cardiac amyloidosis using delayed enhanced MDCT: a pilot study. *Eur Radiol*. 2015 Aug;25(8):2291-7.
15. Deux JF, Damy T, Rahmouni A, Mayer J, Planté-Bordeneuve V. Noninvasive detection of cardiac involvement in patients with hereditary transthyretin associated amyloidosis using cardiac magnetic resonance imaging: a prospective study. *Amyloid*. 2014 Dec;21(4):246-55.
16. Rosso J, Gallat A, Guellich A, Damy T; Pour le réseau amylose Mondor. [Answer to the letter from Mrs Aurélie Dumas about our article "senile systemic amyloidosis: definition, diagnosis, why thinking about?"]. *Presse Med*. 2014 Sep;43(9):1027-8.
17. Mohty D, Damy T, Cosnay P, Echahidi N, Casset-Senon D, Virost P, Jaccard A. Cardiac amyloidosis: updates in diagnosis and management. *Arch Cardiovasc Dis*. 2013 Oct;106(10):528-40.
18. Damy T, Plante-Bordeneuve V, Dogan A. Characterization of untyped cardiac amyloidosis by mass spectrometry in a patient with Gly6Ser transthyretin polymorphism in fatal cardiogenic shock. *Arch Cardiovasc Dis*. 2014 Dec;107(12):706-8.
19. Damy T, Deux JF, Moutereau S, Guendouz S, Mohty D, Rappeneau S, Guellich A, Hittinger L, Loric S, Lefaucheur JP, Plante-Bordeneuve V. Role of natriuretic peptide to predict cardiac abnormalities in patients with hereditary transthyretin amyloidosis. *Amyloid*. 2013 Dec;20(4):212-20.
20. Damy T, Mohty D, Deux JF, Rosso J, Benhaïem N, Lellouche N, Sabbah L, Guendouz S, Tissot CM, Rappeneau S, Pongas D, Bodez D, Krypciak S, Guellich A, Dubois-Randé JL, Hittinger L, Lefaucheur JP, Jaccard A, Planté-Bordeneuve V. [Senile systemic amyloidosis: definition, diagnosis, why thinking about?]. *Presse Med*. 2013 Jun;42(6 Pt 1):1003-14.
21. Lefaucheur JP, Ng Wing Tin S, Kerschen P, Damy T, Planté-Bordeneuve V. Neurophysiological markers of small fibre neuropathy in TTR-FAP mutation carriers. *J Neurol*. 2013 Jun;260(6):1497-503.
22. Damy T, Plante-Bordeneuve V, Valleix S. Diagnosis of cardiac amyloidosis by magnetic resonance imaging due to a new mutation in the transthyretin gene. *Arch Cardiovasc Dis*. 2012 Nov;105(11):614-5.

1. Richard P, Trollet C, Stojkovic T, de Becdelievre A, Perie S, Pouget J, Eymard B; Neurologists of French Neuromuscular Reference Centers CORNEMUS and FILNEMUS. Correlation between PABPN1 genotype and disease severity in oculopharyngeal muscular dystrophy. *Neurology*. 2016 Dec 23. [Epub ahead of print]
2. Richard I, Hogrel JY, Stockholm D, Payan CA, Fougerousse F; Calpainopathy Study Group, Eymard B, Mignard C, Lopez de Munain A, Fardeau M, Urtizbera JA. Natural history of LGMD2A for delineating outcome measures in clinical trials. *Ann Clin Transl Neurol*, 2016 Mar 4;3(4):248-65.
3. Udd B, Brignol TN, Andoni Urtizbera J. [Finland: an ideally valued genetic heritage]. *Med Sci (Paris)*. 2016 Nov;32 Hors série n°2:52-54. French. PubMed. PMID: 27869077.
4. Modrego PJ, Gazulla J, Cobo AM, Andoni Urtizbera J. Une cause inhabituelle d'hyperCKémie. *Med Sci (Paris)*. 2016 Nov;32 Hors série n°2:12-13. French. PubMed PMID: 27869070.
5. Dogan C, De Antonio M, Hamroun D, Varet H, Fabbro M, Rougier F, Amarof K, Arne Bes MC, Bedat-Millet AL, Behin A, Bellance R, Bouhour F, Boutte C, Boyer F, Campana-Salort E, Chapon F, Cintas P, Desnuelle C, Deschamps R, Drouin-Garraud V, Ferrer X, Gervais-Bernard H, Ghorab K, Laforet P, Magot A, Magy L, Menard D, Minot MC, Nadaj-Pakleza A, Pellieux S, Pereon Y, Preudhomme M, Pouget J, Sacconi S, Sole G, Stojkovich T, Tiffreau V, Urtizbera A, Vial C, Zagnoli F, Caranhac G, Bourlier C, Riviere G, Geille A, Gherardi RK, Eymard B, Puymirat J, Katsahian S, Bassez G. Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1. Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional. Observational Study. *PLoS One*. 2016 Feb 5;11(2):e0148264.
6. Woudt L, Di Capua GA, Krahn M, Castiglioni C, Hughes R, Campero M, Trangulao A, González-Hormazábal P, Godoy-Herrera R, Lévy N, Urtizbera JA, Jara L, Bevilacqua JA. Toward an objective measure of functional disability in dysferlinopathy. *Muscle Nerve*. 2016 Jan;53(1):49-57.
7. Fatehi F, Nafissi S, Urtizbera JA, Blanck-Labelle V, Lévy N, Krahn M, Dbouk MB, Attarian S. Dysferlinopathy in Iran: Clinical and genetic report. *J Neurol Sci*. 2015 Dec 15;359(1-2):256-9.
8. Brignol TN, Urtizbera JA. [Ptosis in rare muscle and neuromuscular junction disorders: A literature review and diagnostic flowchart]. *J Fr Ophtalmol*. 2015. Dec;38(10):e253-4.
9. Urtizbera JA, Lochmuller H, Tournev I. [Myology and ethnic minorities: all roads lead to the Roma]. *Med Sci (Paris)*. 2015 Nov;31 Spec No 3:34-8.
10. Urtizbera JA, Béhin A. [GNE myopathy]. *Med Sci (Paris)*. 2015 Nov;31 Spec No 3:20-7.
11. Nectoux J, de Cid R, Baulande S, Leturcq F, Urtizbera JA, Penisson-Besnier I, Nadaj-Pakleza A, Roudaut C, Criqui A, Orhant L, Peyroulan D, Ben Yaou R, Nelson I, Cobo AM, Arné-Bes MC, Uro-Coste E, Nitschke P, Claustres M, Bonne G, Lévy N, Chelly J, Richard I, Cossée M. Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. *Eur J Hum Genet*;2014 Oct 29.
12. Foley AR, Menezes MP, Pandraud A, Gonzalez MA, Al-Odaib A, Abrams AJ, Sugano K, Yonezawa A, Manzur AY, Burns J, Hughes I, McCullagh BG, Jungbluth H, Lim MJ, Lin JP, Megarbane A, Urtizbera JA, Shah AH, Antony J, Webster R, Broomfield A, Ng J, Mathew AA, O'Byrne JJ, Forman E, Scoto M, Prasad M, O'Brien K, Olpin S, Oppenheim M, Hargreaves I,

Land JM, Wang MX, Carpenter K, Horvath R, Straub V, Lek M, Gold W, Farrell MO, Brandner S, Phadke R, Matsubara K, McGarvey ML, Scherer SS, Baxter PS, King MD, Clayton P, Rahman S, Reilly MM, Ouvrier RA, Christodoulou J, Züchner S, Muntoni F, Houlden H. Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. *Brain*. 2014 Jan;137(Pt 1):44-56.

13. Xi J, Blandin G, Lu J, Luo S, Zhu W, Bérout C, Pécheux C, Labelle V, Lévy N, Urtizbera JA, Zhao C, Krahn M. Clinical heterogeneity and a high proportion of novel mutations in a Chinese cohort of patients with dysferlinopathy. *Neurol India*, 2014; 62(6):635-9.
14. Vuillerot C, Rippert P, Roche S, Bérard C, Margirier F, de Lattre C, Poirot I, Berruyer A, Tiffreau V, Fournier-Mehouas M, Bouhour F, Urtizbera JA, Renders A, Ecochard R; Le groupe d'étude NM-Score. Development and validation of a motor function classification in patients with neuromuscular disease: the NM-score. *Ann Phys Rehabil Med*; 2013 Dec;56(9-10):673-86.
15. Khademian H, Mehravar E, Urtizbera J, Sagoo S, Sandoval L, Carbajo R, Darvish B, Valles-Ayoub Y, Darvish D. Prevalence of GNE p.M712T and hereditary inclusion body myopathy (HIBM) in Sangesar population of Northern Iran. *Clin Genet*, 2013;Dec;84(6):589-92.
16. Nalini A, Gayathri N, Richard P, Cobo AM, Urtizbera JA. New mutation of the desmin gene identified in an extended Indian pedigree presenting with distal myopathy and cardiac disease. *Neurol India*. 2013 Nov-Dec;61(6):622-6.
17. Funalot B, Topilko P, Arroyo MA, Sefiani A, Hedley-Whyte ET, Yoldi ME, Richard L, Touraille E, Laurichesse M, Khalifa E, Chauzeix J, Ouedraogo A, Cros D, Magdelaine C, Sturtz FG, Urtizbera JA, Charnay P, Bragado FG, Vallat JM. Homozygous deletion of an EGR2 enhancer in congenital amyelinating neuropathy. *Ann Neurol*, 2012 May;71(5):719-23.
18. Böhm J, Biancalana V, Dechene ET, Bitoun M, Pierson CR, Schaefer E, Karasoy H, Dempsey MA, Klein F, Dondaine N, Kretz C, Haumesser N, Poirson C, Toussaint A, Greenleaf RS, Barger MA, Mahoney LJ, Kang PB, Zanoteli E, Vissing J, Witting N, Echaniz-Laguna A, Wallgren-Pettersson C, Dowling J, Merlini L, Oldfors A, Bomme Ousager L, Melki J, Krause A, Jern C, Oliveira AS, Petit F, Jacqueline A, Chaussonnet A, Mowat D, Leheup B, Cristofano M, Poza Aldea JJ, Michel F, Furby A, Llona JE, Van Coster R, Bertini E, Urtizbera JA, Drouin-Garraud V, Bérout C, Prudhon B, Bedford M, Mathews K, Erby LA, Smith SA, Roggenbuck J, Crowe CA, Brennan Spitale A, Johal SC, Amato AA, Demmer LA, Jonas J, Darras BT, Bird TD, Laurino M, Welt SI, Trotter C, Guicheney P, Das S, Mandel JL, Beggs AH, Laporte J. Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. *Hum Mutat*, 2012. Jun;33(6):949-59.
19. Blandin G, Beroud C, Labelle V, Nguyen K, Wein N, Hamroun D, Williams B, Monnier N, Rufibach LE, Urtizbera JA, Cau P, Bartoli M, Lévy N, Krahn M. UMD-DYSF, a novel locus specific database for the compilation and interactive analysis of mutations in the dysferlin gene. *Hum Mutat*, 2012 Mar;33(3):E2317-31.
20. Sacconi S, Féasson L, Antoine JC, Pécheux C, Bernard R, Cobo AM, Casarin A, Salviati L, Desnuelle C, Urtizbera A. A novel CRYAB mutation resulting in multisystemic disease. *Neuromuscul Disord*, 2012;22:66-72.
21. Johnson JO, Gibbs JR, Megarbane A, Urtizbera JA, Hernandez DG, Foley AR, Arepalli S, Pandraud A, Simón-Sánchez J, Clayton P, Reilly MM, Muntoni F, Abramzon Y, Houlden H,

Singleton AB. Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. *Brain*, 2012;135:2875-82.

BIBLIOGRAPHIE DU COORDONNATEUR Dr. Pascal LAFORET

1. Wahbi K, Babuty D, Probst V, Wissocque L, Labombarda F, Porcher R, Bécane HM, Lazarus A, Béhin A, Laforêt P, Stojkovic T, Clementy N, Dussauge AP, Gourraud JB, Pereon Y, Lacour A, Chapon F, Milliez P, Klug D, Eymard B, Duboc D. Incidence and predictors of sudden death, major conduction defects and sustained ventricular tachyarrhythmias in 1388 patients with myotonic dystrophy type 1. *Eur Heart J*. 2016 Dec 9.
2. El Mendili MM, Lenglet T, Stojkovic T, Behin A, Guimarães-Costa R, Salachas F, Meininger V, Bruneteau G, Le Forestier N, Laforêt P, Lehericy S, Benali H, Pradat PF. Cervical Spinal Cord Atrophy Profile in Adult SMN1-Linked SMA. *PLoS One*. 2016 Apr 18;11
3. Scalco RS, Snoeck M, Quinlivan R, Treves S, Laforêt P, Jungbluth H, Voermans NC. Exertional rhabdomyolysis: physiological response or manifestation of an underlying myopathy? *BMJ Open Sport Exerc Med*. 2016 Sep 7;2(1)
4. Marey I, Ben Yaou R, Deburgrave N, Vasson A, Nectoux J, Leturcq F, Eymard B, Laforet P, Behin A, Stojkovic T, Mayer M, Tiffreau V, Desguerre I, Boyer FC, Nadaj-Pakleza A, Ferrer X, Wahbi K, Becane HM, Claustres M, Chelly J, Cossee M. Non Random Distribution of DMD Deletion Breakpoints and Implication of Double Strand Breaks Repair and Replication Error Repair Mechanisms. *J Neuromuscul Dis*. 2016 May 27;3(2):227-245.
5. Masat E, Laforêt P, De Antonio M, Corre G, Perniconi B, Taouagh N, Mariampillai K, Amelin D, Mauhin W, Hogrel JY, Caillaud C, Ronzitti G, Puzzo F, Kuranda K, Colella P, Mallone R, Benveniste O, Mingozzi F; French Pompe Registry Study Group. Long-term exposure to Myozyme results in a decrease of anti-drug antibodies in late-onset Pompe disease patients. *Sci Rep*. 2016 Nov 4;6:36182.
6. Laforêt P. What have we learned about glycogenosis in recent years? *Rev Neurol(Paris)*. 2016 Oct;172(10):541-545. doi: 10.1016/j.neurol.2016.08.001. Review.
7. Decostre V, Laforêt P, Nadaj-Pakleza A, De Antonio M, Leveugle S, Ollivier G, Canal A, Kachel K, Petit F, Eymard B, Behin A, Wahbi K, Labrune P, Hogrel JY. Cross-sectional retrospective study of muscle function in patients with glycogen storage disease type III. *Neuromuscul Disord*. 2016 Sep;26(9):584-92.
8. Perniconi B, Vauthier-Brouzes D, Morélot-Panzini C, Dommergues M, Nizard J, Taouagh N, Hogrel JY, Canal A, Servais L, Laforêt P. Multidisciplinary care allowing uneventful vaginal delivery in a woman with Pompe disease. *Neuromuscul Disord*. 2016 Sep;26(9):610-3.
9. Schoser B, Laforêt P, Kruijshaar ME, Toscano A, van Doorn PA, van der Ploeg AT; European POMpe Consortium (EPOC).. Minutes of the European POMpe Consortium (EPOC) Meeting March 27 to 28, 2015, Munich, Germany. *Acta Myol*. 2015 Dec;34(2-3):141-3.

10. Thevenon J, Laurent G, Ader F, Laforêt P, Klug D, Duva Pentiah A, Gouya L, Maurage CA, Kacet S, Eicher JC, Albuissou J, Desnos M, Bieth E, Duboc D, Martin L, Réant P, Picard F, Bonithon-Kopp C, Gautier E, Biquet C, Thauvin-Robinet C, Faivre L, Bouvagnet P, Charron P, Richard P. High prevalence of arrhythmic and myocardial complications in patients with cardiac glycogenosis due to PRKAG2 mutations. *Europace*. 2016 May 17.
11. Sentner CP, Hoogeveen IJ, Weinstein DA, Santer R, Murphy E, McKiernan PJ, Steuerwald U, Beauchamp NJ, Taybert J, Laforêt P, Petit FM, Hubert A, Labrune P, Smit GP, Derks TG. Glycogen storage disease type III: diagnosis, genotype, management, clinical course and outcome. *J Inher Metab Dis*. 2016 Sep;39(5):697-704.
12. Allenbach Y, Keraen J, Bouvier AM, Jooste V, Champtiaux N, Hervier B, Schoindre Y, Rigolet A, Gilardin L, Musset L, Charuel JL, Boyer O, Jouen F, Drouot L, Martinet J, Stojkovic T, Eymard B, Laforêt P, Behin A, Salort-Campana E, Fain O, Meyer A, Schleinitz N, Mariampillai K, Grados A, Benveniste O. High risk of cancer in autoimmune necrotizing myopathies: usefulness of myositis specific antibody. *Brain*. 2016 Aug;139(Pt 8):2131-5.
13. Béhin A, Acquaviva-Bourdain C, Souvannanorath S, Streichenberger N, Attarian S, Bassez G, Brivet M, Fouilhoux A, Labarre-Villa A, Laquerrière A, Pérard L, Kaminsky P, Pouget J, Rigal O, Vanhulle C, Eymard B, Vianey-Saban C, Laforêt P. Multiple acyl-CoA dehydrogenase deficiency (MADD) as a cause of late-onset treatable metabolic disease. *Rev Neurol (Paris)*. 2016 Mar;172(3):231-41.
14. Dogan C, De Antonio M, Hamroun D, Varet H, Fabbro M, Rougier F, Amarof K, Arne Bes MC, Bedat-Millet AL, Behin A, Bellance R, Bouhour F, Boutte C, Boyer F, Campana-Salort E, Chapon F, Cintas P, Desnuelle C, Deschamps R, Drouin-Garraud V, Ferrer X, Gervais-Bernard H, Ghorab K, Laforet P, Magot A, Magy L, Menard D, Minot MC, Nadaj-Pakleza A, Pellieux S, Pereon Y, Preudhomme M, Pouget J, Sacconi S, Sole G, Stojkovich T, Tiffreau V, Urtizbera A, Vial C, Zagnoli F, Caranhac G, Bourlier C, Riviere G, Geille A, Gherardi RK, Eymard B, Puymirat J, Katsahian S, Bassez G. Gender as a Modifying Factor Influencing Myotonic Dystrophy Type 1 Phenotype Severity and Mortality: A Nationwide Multiple Databases Cross-Sectional Observational Study. *PLoS One*. 2016 Feb 5;11(2)
15. Allenbach Y, Leroux G, Suárez-Calvet X, Preusse C, Gallardo E, Hervier B, Rigolet A, Hie M, Pehl D, Limal N, Hufnagl P, Zerbe N, Meyer A, Aouizerate J, Uzunhan Y, Maisonobe T, Goebel HH, Benveniste O, Stenzel W; French Myositis Network.. Dermatomyositis With or Without Anti-Melanoma Differentiation-Associated Gene 5 Antibodies: Common Interferon Signature but Distinct NOS2 Expression. *Am J Pathol*. 2016 Mar;186(3):691-700.
16. Herlin B, Laforêt P, Labrune P, Fournier E, Stojkovic T. Peripheral neuropathy in glycogen storage disease type III: Fact or myth? *Muscle Nerve*. 2016 Feb;53(2):310-2.
17. Béhin A, Salort-Campana E, Wahbi K, Richard P, Carlier RY, Carlier P, Laforêt P, Stojkovic T, Maisonobe T, Verschuere A, Franques J, Attarian S, Maues de Paula A, Figarella-Branger D, Bécane HM, Nelson I, Duboc D, Bonne G, Vicart P, Udd B, Romero N, Pouget J, Eymard B. Myofibrillar myopathies: State of the art, present and future challenges. *Rev Neurol (Paris)*. 2015 Oct;171(10):715-29.
18. Wahbi K, Bougouin W, Béhin A, Stojkovic T, Bécane HM, Jardel C, Berber N, Mochel F, Lombès A, Eymard B, Duboc D, Laforêt P. Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. *Eur Heart J*. 2015 Nov 7;36(42):2886-93.

19. Schoser B, Laforêt P, Kruijshaar ME, Toscano A, van Doorn PA, van der Ploeg AT; European Pompe Consortium (EPOC). 208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26-28 September 2014. *Neuromuscul Disord*. 2015 Aug;25(8):674-8.
20. Heslop E, Csimma C, Straub V, McCall J, Nagaraju K, Wagner KR, Caizergues D, Korinthenberg R, Flanigan KM, Kaufmann P, McNeil E, Mendell J, Hesterlee S, Wells DJ, Bushby K; TACT.. The TREAT-NMD advisory committee for therapeutics (TACT): an innovative de-risking model to foster orphan drug development. *Orphanet J Rare Dis*. 2015 Apr 23;10:49.
21. Semplicini C, Vissing J, Dahlqvist JR, Stojkovic T, Bello L, Witting N, Duno M, Leturcq F, Bertolin C, D'Ambrosio P, Eymard B, Angelini C, Politano L, Laforêt P, Pegoraro E. Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*. 2015 Apr 28;84(17):1772-81.
22. Preisler N, Laforêt P, Madsen KL, Prahm KP, Hedermann G, Vissing CR, Galbo H, Vissing J. Skeletal muscle metabolism is impaired during exercise in glycogen storage disease type III. *Neurology*. 2015 Apr 28;84(17):1767-71.
23. Hogrel JY, van den Bogaart F, Ledoux I, Ollivier G, Petit F, Koujah N, Béhin A, Stojkovic T, Eymard B, Voermans N, Laforêt P. Diagnostic power of the non-ischaemic forearm exercise test in detecting glycogenosis type V. *Eur J Neurol*. 2015 Jun;22(6):933-40.
24. Michon CC, Gargiulo M, Hahn-Barma V, Petit F, Nadaj-Pakleza A, Herson A, Eymard B, Labrune P, Laforet P. Cognitive profile of patients with glycogen storage disease type III: a clinical description of seven cases. *J Inherit Metab Dis*. 2015 May;38(3):573-80.
25. Ørngreen MC, Vissing J, Laforêt P. No effect of bezafibrate in patients with CPTII and VLCAD deficiencies. *J Inherit Metab Dis*. 2015 Mar;38(2):373-4.
26. Böhm J, Biancalana V, Malfatti E, Dondaine N, Koch C, Vasli N, Kress W, Strittmatter M, Taratuto AL, Gonorazky H, Laforêt P, Maisonobe T, Olivé M, Gonzalez-Mera L, Fardeau M, Carrière N, Clavelou P, Eymard B, Bitoun M, Rendu J, Fauré J, Weis J, Mandel JL, Romero NB, Laporte J. Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. *Brain*. 2014 Dec;137(Pt 12):3160-70.
27. Tchikviladzé M, Gilleron M, Maisonobe T, Galanaud D, Laforêt P, Durr A, Eymard B, Mochel F, Ogier H, Béhin A, Stojkovic T, Degos B, Gourfinkel-An I, Sedel F, Anheim M, Elbaz A, Viala K, Vidailhet M, Brice A, Jardel C, Lombès A. A diagnostic flow chart for POLG-related diseases based on signs sensitivity and specificity. *J Neurol Neurosurg Psychiatry*. 2015 Jun;86(6):646-54.
28. Sacconi S, Wahbi K, Theodore G, Garcia J, Salviati L, Bouhour F, Vial C, Duboc D, Laforêt P, Desnuelle C. Atrio-ventricular block requiring pacemaker inpatients with late onset Pompe disease. *Neuromuscul Disord*. 2014 Jul;24(7):648-50.
29. Allenbach Y, Drouot L, Rigolet A, Charuel JL, Jouen F, Romero NB, Maisonobe T, Dubourg O, Behin A, Laforet P, Stojkovic T, Eymard B, Costedoat-Chalumeau N, Campana-Salort E, Tournadre A, Musset L, Bader-Meunier B, Kone-Paut I, Sibia J, Servais L, Fain O, Larroche C, Diot E, Terrier B, De Paz R, Dossier A, Menard D, Morati C, Roux M, Ferrer X, Martinet J, Besnard S, Bellance R, Cacoub P, Arnaud L, Grosbois B, Herson S, Boyer O, Benveniste O;

French Myositis Network. Anti-HMGCR autoantibodies in European patients with autoimmune necrotizing myopathies: inconstant exposure to statin. *Medicine (Baltimore)*. 2014 May;93(3):150-7.

30. Tegtmeyer LC, Rust S, van Scherpenzeel M, Ng BG, Losfeld ME, Timal S, Raymond K, He P, Ichikawa M, Veltman J, Huijben K, Shin YS, Sharma V, Adamowicz M, Lammens M, Reunert J, Witten A, Schrapers E, Matthijs G, Jaeken J, Rymen D, Stojkovic T, Laforêt P, Petit F, Aumaître O, Czarnowska E, Piraud M, Podskarbi T, Stanley CA, Matalon R, Burda P, Seyyedi S, Debus V, Socha P, Sykut-Cegielska J, van Spronsen F, de Meirleir L, Vajro P, DeClue T, Ficicioglu C, Wada Y, Wevers RA, Vanderschaeghe D, Callewaert N, Fingerhut R, van Schaftingen E, Freeze HH, Morava E, Lefeber DJ, Marquardt T. Multiple phenotypes in phosphoglucomutase 1 deficiency. *N Engl J Med*. 2014 Feb 6;370(6):533-42.
31. Drouot L, Allenbach Y, Jouen F, Charuel JL, Martinet J, Meyer A, Hirschberger O, Bader-Meunier B, Kone-Paut I, Campana-Salort E, Eymard B, Tournadre A, Musset L, Sibilia J, Marie I, Benveniste O, Boyer O; French Myositis Network [CN]. Exploring necrotizing autoimmune myopathies with a novel immunoassay for anti-3-hydroxy-3-methyl-glutaryl-CoA reductase autoantibodies. *Arthritis Res Ther*. 2014 Feb 3;16(1):R39
32. Ørngreen MC, Madsen KL, Preisler N, Andersen G, Vissing J, Laforêt P. Bezafibrate in skeletal muscle fatty acid oxidation disorders: a randomized clinical trial. *Neurology*. 2014 Feb 18;82(7):607-13
33. Auré K, Dubourg O, Jardel C, Clarysse L, Sternberg D, Fournier E, Laforêt P, Streichenberger N, Petiot P, Gervais-Bernard H, Vial C, Bedat-Millet AL, Drouin-Garraud V, Bouillaud F, Vandier C, Fontaine B, Lombès A. Episodic weakness due to mitochondrial DNA MT-ATP6/8 mutations. *Neurology*. 2013 Nov 19;81(21):1810-8.
34. Wahbi K, Algalarrondo V, Bécane HM, Fressart V, Beldjord C, Azibi K, Lazarus A, Berber N, Radvanyi-Hoffman H, Stojkovic T, Béhin A, Laforêt P, Eymard B, Hatem S, Duboc D. Brugada syndrome and abnormal splicing of SCN5A in myotonic dystrophy type 1. *Arch Cardiovasc Dis*. 2013 Dec;106(12):635-43.
35. Perrin L, Féasson L, Furby A, Laforêt P, Petit FM, Gautheron V, Chabrier S. PNPLA2 mutation: a paediatric case with early onset but indolent course. *Neuromuscul Disord*. 2013 Dec;23(12):986-91.
36. Laforêt P, Laloui K, Granger B, Hamroun D, Taouagh N, Hogrel JY, Orlikowski D, Bouhour F, Lacour A, Salort-Campana E, Penisson-Besnier I, Sacconi S, Zagnoli F, Chapon F, Eymard B, Desnuelle C, Pouget J; French Pompe Registry Study Group. The French Pompe registry. Baseline characteristics of a cohort of 126 patients with adult Pompe disease. *Rev Neurol (Paris)*. 2013 Aug-Sep;169(8-9):595-602.
37. Gargiulo M, Herson A, Michon CC, Hogrel JY, Doppler V, Laloui K, Herson S, Payan C, Eymard B, Laforêt P. Attitudes and expectations of patients with neuromuscular diseases about their participation in a clinical trial. *Rev Neurol (Paris)*. 2013 Aug-Sep;169(8-9):670-6.
38. Malfatti E, Olivé M, Taratuto AL, Richard P, Brochier G, Bitoun M, Gueneau L, Laforêt P, Stojkovic T, Maisonobe T, Monges S, Lubieniecki F, Vasquez G, Streichenberger N, Lacène E, Saccoliti M, Prudhon B, Alexianu M, Figarella-Branger D, Schessl J, Bonnemann C, Eymard B, Fardeau M, Bonne G, Romero NB. Skeletal muscle biopsy analysis in reducing body myopathy and other FHL1-related disorders. *J Neuropathol Exp Neurol*. 2013 Sep;72(9):833-45.

39. Donadille B, D'Anella P, Auclair M, Uhrhammer N, Sorel M, Grigorescu R, Ouzounian S, Cambonie G, Boulot P, Laforêt P, Carbonne B, Christin-Maitre S, Bignon YJ, Vigouroux C. Partial lipodystrophy with severe insulin resistance and adult progeria Werner syndrome. *Orphanet J Rare Dis*. 2013 Jul 12;8:106.
40. Périé S, Trollet C, Mouly V, Vanneaux V, Mamchaoui K, Bouazza B, Marolleau JP, Laforêt P, Chapon F, Eymard B, Butler-Browne G, Larghero J, St Gully JL. Autologous myoblast transplantation for oculopharyngeal muscular dystrophy: a phase I/IIa clinical study. *Mol Ther*. 2014 Jan;22(1):219-25.
41. Degos B, Laforêt P, Jardel C, Sedel F, Jossay-Winter M, Romero NB, Lyon-Caen O, Tourbah A. POLG mutations associated with remitting/relapsing neurological events. *J Clin Neurosci*. 2014 Jan;21(1):186-8.
42. Nilsson J, Schoser B, Laforet P, Kalev O, Lindberg C, Romero NB, Dávila López M, Akman HO, Wahbi K, Iglseider S, Eggers C, Engel AG, Dimauro S, Oldfors A. P olyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. *Ann Neurol*. 2013 Dec;74(6):914-9.
43. Preisler N, Laforêt P, Echaniz-Laguna A, Ørngreen MC, Lonsdorfer-Wolf E, Doutreleau S, Geny B, Stojkovic T, Piraud M, Petit FM, Vissing J. Fat and carbohydrate metabolism during exercise in phosphoglucomutase type 1 deficiency. *J Clin Endocrinol Metab*. 2013 Jul;98(7):E1235-40
44. Palmio J, Evilä A, Chapon F, Tasca G, Xiang F, Brådvik B, Eymard B, Echaniz-Laguna A, Laporte J, Kärppä M, Mahjneh I, Quinlivan R, Laforêt P, Damian M, Berardo A, Taratuto AL, Bueri JA, Tommiska J, Raivio T, Tuerk M, Göllitz P, Chevessier F, Sewry C, Norwood F, Hedberg C, Schröder R, Edström L, Oldfors A, Hackman P, Udd B. Hereditary myopathy with early respiratory failure: occurrence in various populations. *J Neurol Neurosurg Psychiatry*. 2014 Mar;85(3):345-53.
45. Preisler N, Pradel A, Husu E, Madsen KL, Becquemin MH, Mollet A, Labrune P, Petit F, Hogrel JY, Jardel C, Maillot F, Vissing J, Laforêt P. Exercise intolerance in Glycogen Storage Disease Type III: weakness or energy deficiency? *Mol Genet Metab*. 2013 May;109(1):14-20.
46. Eymard B, Stojkovic T, Sternberg D, Richard P, Nicole S, Fournier E, Béhin A, Laforêt P, Servais L, Romero N, Fardeau M, Hantaï D; Membres du réseau national Syndromes Myasthéniques Congénitaux.. [Congenital myasthenic syndromes: difficulties in the diagnosis, course and prognosis, and therapy--The French National Congenital Myasthenic Syndrome Network experience]. *Rev Neurol (Paris)*. 2013 Feb;169 Suppl 1:S45-55.
47. Voermans NC, Jungbluth H, Brusse E, van Engelen BG, Lafôret P. Exertional hyperckemia might be the first manifestation of a genetic disorder. *Muscle Nerve*. 2013 Sep;48(3):461-2.
48. Böhm J, Chevessier F, Maues De Paula A, Koch C, Attarian S, Feger C, Hantaï D, Laforêt P, Ghorab K, Vallat JM, Fardeau M, Figarella-Branger D, Pouget J, Romero NB, Koch M, Ebel C, Levy N, Krahn M, Eymard B, Bartoli M, Laporte J. Constitutive activation of the calcium sensor STIM1 causes tubular-aggregate myopathy. *Am J Hum Genet*. 2013 Feb 7;92(2):271-8
49. Malfatti E, Laforêt P, Jardel C, Stojkovic T, Behin A, Eymard B, Lombès A, Benmalek A, Bécane HM, Berber N, Meune C, Duboc D, Wahbi K. High risk of severe cardiac adverse events in patients with mitochondrial m.3243A>G mutation. *Neurology*. 2013 Jan 1;80(1):100-5.

50. Billot S, Hervé D, Akman HO, Froissart R, Baussan C, Claeys KG, Piraud M, Sedel F, Mochel F, Laforêt P. Acute but transient neurological deterioration revealing adult polyglucosan body disease. *J Neurol Sci.* 2013 Jan 15;324(1-2):179-82.
51. Wahbi K, Béhin A, Bécane HM, Leturcq F, Cossée M, Laforêt P, Stojkovic T, Carlier P, Toussaint M, Gaxotte V, Cluzel P, Eymard B, Duboc D. Dilated cardiomyopathy in patients with mutations in anoctamin 5. *Int J Cardiol.* 2013 Sep 20;168(1):76-9.
52. Mochel F, Schiffmann R, Steenweg ME, Akman HO, Wallace M, Sedel F, Laforêt P, Levy R, Powers JM, Demeret S, Maisonobe T, Froissart R, Da Nobrega BB, Fogel BL, Natowicz MR, Lubetzki C, Durr A, Brice A, Rosenmann H, Barash V, Kakhlon O, Gomori JM, van der Knaap MS, Lossos A. Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. *Ann Neurol.* 2012 Sep;72(3):433-41.
53. van der Ploeg AT, Barohn R, Carlson L, Charrow J, Clemens PR, Hopkin RJ, Kishnani PS, Laforêt P, Morgan C, Nations S, Pestronk A, Plotkin H, Rosenbloom BE, Sims KB, Tsao E. Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. *Mol Genet Metab.* 2012 Nov;107(3):456-61.
54. Preisler N, Laforet P, Madsen KL, Hansen RS, Lukacs Z, Ørngreen MC, Lacour A, Vissing J. Fat and carbohydrate metabolism during exercise in late-onset Pompe disease. *Mol Genet Metab.* 2012 Nov;107(3):462-8.
55. Prigent H, Orlikowski D, Laforêt P, Letilly N, Falaize L, Pellegrini N, Annane D, Raphael JC, Lofaso F. Supine volume drop and diaphragmatic function in adults with Pompe disease. *Eur Respir J.* 2012 Jun;39(6):1545-6.
56. Gentil C, Leturcq F, Ben Yaou R, Kaplan JC, Laforet P, Péniisson-Besnier I, Espil-Taris C, Voit T, Garcia L, Piétri-Rouxel F. Variable phenotype of del45-55 Becker patients correlated with nNOS μ mislocalization and RYR1 hypernitrosylation. *Hum Mol Genet.* 2012 Aug 1;21(15):3449-60.
57. Laforêt P, Ørngreen M, Preisler N, Andersen G, Vissing J. Blocked muscle fat oxidation during exercise in neutral lipid storage disease. *Arch Neurol.* 2012 Apr;69(4):530-3.
58. Michot C, Hubert L, Romero NB, Gouda A, Mamoune A, Mathew S, Kirk E, Viollet L, Rahman S, Bekri S, Peters H, McGill J, Glamuzina E, Farrar M, von der Hagen M, Alexander IE, Kirmse B, Barth M, Laforet P, Benlian P, Munnich A, JeanPierre M, Elpeleg O, Pines O, Delahodde A, de Keyser Y, de Lonlay P. Study of LPIN1, LPIN2 and LPIN3 in rhabdomyolysis and exercise-induced myalgia. *J Inherit Metab Dis.* 2012 Nov;35(6):1119-28.
59. Wahbi K, Meune C, Porcher R, Bécane HM, Lazarus A, Laforêt P, Stojkovic T, Béhin A, Radvanyi-Hoffmann H, Eymard B, Duboc D. Electrophysiological study with prophylactic pacing and survival in adults with myotonic dystrophy and conduction system disease. *JAMA.* 2012 Mar 28;307(12):1292-301.
60. Malfatti E, Birouk N, Romero NB, Piraud M, Petit FM, Hogrel JY, Laforêt P. Juvenile-onset permanent weakness in muscle phosphofructokinase deficiency. *J Neurol Sci.* 2012 May 15;316(1-2):173-7.

61. Béhin A, Jardel C, Claeys KG, Fagart J, Louha M, Romero NB, Laforêt P, Eymard B, Lombès A. Adult cases of mitochondrial DNA depletion due to TK2 defect: an expanding spectrum. *Neurology*. 2012 Feb 28;78(9):644-8.
62. Young SP, Piraud M, Goldstein JL, Zhang H, Rehder C, Laforet P, Kishnani PS, Millington DS, Bashir MR, Bali DS. Assessing disease severity in Pompe disease: the roles of a urinary glucose tetrasaccharide biomarker and imaging techniques. *Am J Med Genet C Semin Med Genet*. 2012 Feb 15;160C(1):50-8.
63. Herson S, Hentati F, Rigolet A, Behin A, Romero NB, Leturcq F, Laforêt P, Maisonobe T, Amouri R, Haddad H, Audit M, Montus M, Masurier C, Gjata B, Georger C, Cheraï M, Carlier P, Hogrel JY, Herson A, Allenbach Y, Lemoine FM, Klatzmann D, Sweeney HL, Mulligan RC, Eymard B, Caizergues D, Voït T, Benveniste O. A phase I trial of adeno-associated virus serotype 1-γ-sarcoglycan gene therapy for limb girdle muscular dystrophy type 2C. *Brain*. 2012 Feb;135(Pt 2):483-92.
64. Preisler N, Orngreen MC, Echaniz-Laguna A, Laforet P, Lonsdorfer-Wolf E, Doutreleau S, Geny B, Akman HO, Dimauro S, Vissing J. Muscle phosphorylase kinase deficiency: a neutral metabolic variant or a disease? *Neurology*. 2012 Jan 24;78(4):265-8.
65. Wahbi K, Béhin A, Charron P, Dunand M, Richard P, Meune C, Vicart P, Laforêt P, Stojkovic T, Bécane HM, Kuntzer T, Duboc D. High cardiovascular morbidity and mortality in myofibrillar myopathies due to DES gene mutations: a 10-year longitudinal study. *Neuromuscul Disord*. 2012 Mar;22(3):211-8.

SITE COCHIN (Coordinateur Dr. Karim WAHBI)

1. Wahbi K, Babuty D, Probst V, Wissocque L, Labombarda F, Porcher R, Bécane HM, Lazarus A, Béhin A, Laforêt P, Stojkovic T, Clementy N, Dussauge AP, Gourraud JB, Pereon Y, Lacour A, Chapon F, Milliez P, Klug D, Eymard B, Duboc D. Incidence and predictors of sudden death, major conduction defects and sustained ventricular tachyarrhythmias in 1388 patients with myotonic dystrophy type 1. *Eur Heart J*. 2016 Dec 9.
2. Heller F, Dabaj I, Mah JK, Bergounioux J, Essid A, Bönnemann CG, Rutkowski A, Bonne G, Quijano-Roy S, Wahbi K. Cardiac manifestations of congenital LMNA-related muscular dystrophy in children: three case reports and recommendations for care. *Cardiol Young*. 2016 Dec 12:1-7.
3. Fayssoil A, Laforêt P, Bougouin W, Jardel C, Lombès A, Bécane HM, Berber N, Stojkovic T, Béhin A, Eymard B, Duboc D, Wahbi K. Prediction of long-term prognosis by heteroplasmy levels of the m.3243A>G mutation in patients with the mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes syndrome. *Eur J Neurol*. 2016 Nov 21.
4. Marey I, Ben Yaou R, Deburgrave N, Vasson A, Nectoux J, Leturcq F, Eymard B, Laforet P, Behin A, Stojkovic T, Mayer M, Tiffreau V, Desguerre I, Boyer FC, Nadaj-Pakleza A, Ferrer X, Wahbi K, Bécane HM, Claustres M, Chelly J, Cossee M. Non Random Distribution of DMD

Deletion Breakpoints and Implication of Double Strand Breaks Repair and Replication Error Repair Mechanisms. *J Neuromuscul Dis.* 2016 May 27;3(2):227-245.

5. Avila-Smirnow D, Gueneau L, Batonnet-Pichon S, Delort F, Bécane HM, Claeys K, Beuvin M, Goudeau B, Jais JP, Nelson I, Richard P, Ben Yaou R, Romero NB, Wahbi K, Mathis S, Voit T, Furst D, van der Ven P, Gil R, Vicart P, Fardeau M, Bonne G, Behin A. Cardiac arrhythmia and late-onset muscle weakness caused by a myofibrillar myopathy with unusual histopathological features due to a novel missense mutation in FLNC. *Rev Neurol (Paris).* 2016 Oct;172(10):594-606.
6. Fayssoil A, Lazarus A, Wahbi K, Ognà A, Nardi O, Lofaso F, Clair B, Orlikowski D, Annane D. Cardiac implantable electronic devices in tracheotomized muscular dystrophy patients: Safety and risks. *Int J Cardiol.* 2016 Nov 1;222:975-7.
7. Decostre V, Laforêt P, Nadaj-Pakleza A, De Antonio M, Leveugle S, Ollivier G, Canal A, Kachetel K, Petit F, Eymard B, Behin A, Wahbi K, Labrune P, Hogrel JY. Cross-sectional retrospective study of muscle function in patients with glycogen storage disease type III. *Neuromuscul Disord.* 2016 Sep;26(9):584-92.
8. Cattin ME, Ferry A, Vignaud A, Mougnot N, Jacquet A, Wahbi K, Bertrand AT, Bonne G. Mutation in lamin A/C sensitizes the myocardium to exercise-induced mechanical stress but has no effect on skeletal muscles in mouse. *Neuromuscul Disord.* 2016 Aug;26(8):490-9.
9. Fayssoil A, Ognà A, Chaffaut C, Chevret S, Guimarães-Costa R, Leturcq F, Wahbi K, Prigent H, Lofaso F, Nardi O, Clair B, Behin A, Stojkovic T, Laforet P, Orlikowski D, Annane D. Natural History of Cardiac and Respiratory Involvement, Prognosis and Predictive Factors for Long-Term Survival in Adult Patients with Limb Girdle Muscular Dystrophies Type 2C and 2D. *PLoS One.* 2016 Apr 27;11(4):e0153095.
10. Freyermuth F, Rau F, Kokunai Y, Linke T, Sellier C, Nakamori M, Kino Y, Arandel L, Jollet A, Thibault C, Philipps M, Vicaire S, Jost B, Udd B, Day JW, Duboc D, Wahbi K, Matsumura T, Fujimura H, Mochizuki H, Deryckere F, Kimura T, Nukina N, Ishiura S, Lacroix V, Campan-Fournier A, Navratil V, Chautard E, Auboeuf D, Horie M, Imoto K, Lee KY, Swanson MS, Lopez de Munain A, Inada S, Itoh H, Nakazawa K, Ashihara T, Wang E, Zimmer T, Furling D, Takahashi MP, Charlet-Berguerand N. Splicing misregulation of SCN5A contributes to cardiac-conduction delay and heart arrhythmia in myotonic dystrophy. *Nat Commun.* 2016 Apr 11;7:11067.
11. Wahbi K, Sebag FA, Lellouche N, Lazarus A, Bécane HM, Bassez G, Stojkovic T, Fayssoil A, Laforêt P, Béhin A, Meune C, Eymard B, Duboc D. Atrial flutter in myotonic dystrophy type 1: Patient characteristics and clinical outcome. *Neuromuscul Disord.* 2016 Mar;26(3):227-33.
12. Béhin A, Salort-Campana E, Wahbi K, Richard P, Carlier RY, Carlier P, Laforêt P, Stojkovic T, Maisonobe T, Verschueren A, Franques J, Attarian S, Maues de Paula A, Figarella-Branger D, Bécane HM, Nelson I, Duboc D, Bonne G, Vicart P, Udd B, Romero N, Pouget J, Eymard B. Myofibrillar myopathies: State of the art, present and future challenges. *Rev Neurol (Paris).* 2015 Oct;171(10):715-29.
13. Wahbi K, Bougouin W, Béhin A, Stojkovic T, Bécane HM, Jardel C, Berber N, Mochel F, Lombès A, Eymard B, Duboc D, Laforêt P. Long-term cardiac prognosis and risk stratification in 260 adults presenting with mitochondrial diseases. *Eur Heart J.* 2015;36:2886-2893.

14. Algalarrondo V, Wahbi K, Sebag F, Gourdon G, Beldjord C, Azibi K, Balse E, Coulombe A, Fischmeister R, Eymard B, Duboc D, Hatem SN. Abnormal sodium current properties contribute to cardiac electrical and contractile dysfunction in a mouse model of myotonic dystrophy type 1. *Neuromuscul Disord*. 2015;25:308-320.
15. Deconinck N, Richard P, Allamand V, Behin A, Lafôret P, Ferreira A, de Becdelievre A, Ledeuil C, Gartioux C, Nelson I, Carlier RY, Carlier P, Wahbi K, Romero N, Zobot MT, Bouhour F, Tiffreau V, Lacour A, Eymard B, Stojkovic T. Bethlem myopathy: long-term follow-up identifies COL6 mutations predicting severe clinical evolution. *J Neurol Neurosurg Psychiatry*. 2015;86:1337-1346.
16. Sacconi S, Wahbi K, Theodore G, Garcia J, Salviati L, Bouhour F, Vial C, Duboc D, Laforêt P, Desnuelle C. Atrio-ventricular block requiring pacemaker in patients with late onset Pompe disease. *Neuromuscul Disord*. 2014;24:648-650.
17. Laforêt P, Stojkovic T, Bassez G, Carlier PG, Clément K, Wahbi K, Petit FM, Eymard B, Carlier RY. Neutral lipid storage disease with myopathy: A whole-body nuclear MRI and metabolic study. *Mol Genet Metab*. 2013;108:125-131.
18. Malfatti E, Laforêt P, Jardel C, Stojkovic T, Behin A, Eymard B, Lombès A, Bécane HM, Berber N, Meune C, Duboc D, Wahbi K. High risk of severe cardiac adverse events in patients with mitochondrial m.3243A>G mutation. *Neurology*. 2013;80:100-105.
19. Wahbi K, Algalarrondo V, Bécane HM, Fressart V, Beldjord C, Azibi K, Lazarus A, Berber N, Radvanyi-Hoffman H, Stojkovic T, Béhin A, Laforêt P, Eymard B, Hatem S, Duboc D. Brugada syndrome and abnormal splicing of SCN5A in myotonic dystrophy type 1. *Arch Cardiovasc Dis*. 2013;106:635-643.
20. Nilsson J, Schoser B, Laforet P, Kalev O, Lindberg C, Romero NB, Dávila López M, Akman HO, Wahbi K, Iglseder S, Eggers C, Engel AG, Dimauro S, Oldfors A. Polyglucosan body myopathy caused by defective ubiquitin ligase RBCK1. *Ann Neurol*. 2013;74:914-919.
21. Laforêt P, Stojkovic T, Bassez G, Carlier PG, Clément K, Wahbi K, Petit FM, Eymard B, Carlier RY. Neutral lipid storage disease with myopathy: A whole-body nuclear MRI and metabolic study. *Mol Genet Metab*. 2013;108:125-131.
22. Wahbi K, Béhin A, Bécane HM, Leturcq F, Cossée M, Laforêt P, Stojkovic T, Carlier P, Toussaint M, Gaxotte V, Cluzel P, Eymard B, Duboc D. Dilated cardiomyopathy in patients with mutations in anoctamin 5. *Int J Cardiol*. 2013 Sep 20;168(1):76-9
23. Wahbi K, Meune C, Porcher R, Bécane HM, Lazarus A, Laforêt P, Stojkovic T, Béhin A, Radvanyi-Hoffmann H, Eymard B, Duboc D. Electrophysiological study with prophylactic pacing and survival in adults with myotonic dystrophy and conduction system disease. *JAMA*. 2012;307:1292-1301.
24. Wahbi K, Béhin A, Charron P, Dunand M, Richard P, Meune C, Vicart P, Laforêt P, Stojkovic T, Bécane HM, Kuntzer T, Duboc D. High cardiovascular morbidity and mortality in myofibrillar myopathies due to DES gene mutations: a 10-year longitudinal study. *Neuromuscul Disord*. 2012;22:211-218.

25. Meune C, Khouzami L, Wahbi K, Caramelle P, Decostre V, Bonne G, Pecker F. Blood glutathione decrease in subjects carrying lamin A/C gene mutations is an early marker of cardiac involvement. *Neuromuscul Disord.* 2012;22:252-257.