

Selected peer-reviewed publications (Chronological order 2008-2022)

1. Caballero-Ávila M, Álvarez-Velasco R, Moga E, Rojas-García R, Turon-Sans J, Querol L, Olivé M, Reyes-Leiva D, Illa I, Gallardo E, Cortés-Vicente E. Rituximab in myasthenia gravis: Efficacy, associated infections and risk of induced hypogammaglobulinemia. **Neuromuscul Disord**. 2022 Jun 21:S0960-8966(22)00192-4. doi: 10.1016/j.nmd.2022.06.006. Online ahead of print.
2. Reyes-Leiva D, López-Contreras J, Moga E, Pla-Juncà F, Lynton-Pons E, Rojas-García R, Turon-Sans J, Querol L, Olive M, Álvarez-Velasco R, Caballero-Ávila M, Carbayo Á, Vesperinas-Castro A, Domingo P, Illa I, Gallardo E, Cortés-Vicente E. Immune Response and Safety of SARS-CoV-2 mRNA-1273 Vaccine in Patients With Myasthenia Gravis. **Neurol Neuroimmunol Neuroinflamm**. 2022 Jun 21;9(4):e200002. doi: 10.1212/NXI.000000000200002. Print 2022 Jul.
3. Carrasco-Rozas A, Fernández-Simón E, Suárez-Calvet X, Piñol-Jurado P, Alonso-Pérez J, de Luna N, Schoser B, Meinke P, Domínguez-González C, Hernández-Lafín A, Paradas C, Rivas E, Illa I, Olivé M, Gallardo E, Díaz-Manera J. BNIP3 Is Involved in Muscle Fiber Atrophy in Late-Onset Pompe Disease Patients. **Am J Pathol**. 2022 May 21:S0002-9440(22)00146-8. doi: 10.1016/j.ajpath.2022.05.003. Online ahead of print.
4. Cortés-Vicente E, Álvarez-Velasco R, Pla-Junca F, Rojas-García R, Paradas C, Sevilla T, Casanovas C, Gómez-Caravaca MT, Pardo J, Ramos-Fransi A, Pelayo-Negro AL, Gutiérrez-Gutiérrez G, Turon-Sans J, López de Munain A, Guerrero-Sola A, Jericó I, Martín MA, Mendoza MD, Morís G, Vélez-Gómez B, García-Sobrino T, Pascual-Goñi E, Reyes-Leiva D, Illa I, Gallardo E. Drug-refractory myasthenia gravis: Clinical characteristics, treatments, and outcome. **Ann Clin Transl Neurol**. 2022 Feb;9(2):122-131. doi: 10.1002/acn3.51492. Epub 2022 Jan 26.
5. Doets AY, Lingsma HF, Walgaard C, Islam B, Papri N, Davidson A, Yamagishi Y, Kusunoki S, Dimachkie MM, Waheed W, Kolb N, Islam Z, Mohammad QD, Harbo T, Sindrup SH, Chavada G, Willison HJ, Casanovas C, Bateman K, Miller JAL, van den Berg B, Verboon C, Roodbol J, Leonhard SE, Benedetti L, Kuwabara S, Van den Bergh P, Monges S, Marfia GA, Shahrizaila N, Galassi G, Péréon Y, Bürmann J, Kuitwaard K, Kleyweg RP, Marchesoni C, Sedano Tous MJ, Querol L, Illa I, Wang Y, Nobile-Orazio E, Rinaldi S, Schenone A, Pardo J, Vermeij FH, Lehmann HC, Granit V, Cavaletti G, Gutiérrez-Gutiérrez G, Barroso FA, Visser LH, Katzberg HD, Dardiotis E, Attarian S, van der Kooi AJ, Eftimov F, Wirtz PW, Samijn JPA, Gilhuis HJ, Hadden RDM, Holt JKL, Sheikh KA, Karafiath S, Vytopil M, Antonini G, Feasby TE, Faber CG, Gijssbers CJ, Busby M, Roberts RC, Silvestri NJ, Fazio R, van Dijk GW, Garssen MPJ, Straathof CSM, Gorson KC, Jacobs BC; IGOS Consortium. Predicting Outcome in Guillain-Barré Syndrome: International Validation of the Modified Erasmus GBS Outcome Score. **Neurology**. 2022 Feb 1;98(5):e518-e532. doi: 10.1212/WNL.0000000000013139. Epub 2021 Dec 22.
6. Luijten LWG, Leonhard SE, van der Eijk AA, Doets AY, Appeltshauer L, Arends S, Attarian S, Benedetti L, Briani C, Casanovas C, Castellani F, Dardiotis E, Echaniz-Laguna A, Garssen MPJ, Harbo T, Huizinga R, Humm AM, Jellema K, van der Kooi AJ, Kuitwaard K, Kuntzer T, Kusunoki S, Lascano AM, Martinez-Hernandez E, Rinaldi S, Samijn JPA, Scheidegger O, Tsouni P, Vicino A, Visser LH, Walgaard C, Wang Y, Wirtz PW, Ripellino P, Jacobs BC; IGOS consortium. Guillain-Barré syndrome after SARS-CoV-2 infection in an international prospective cohort study. **Brain**. 2021 Dec 16;144(11):3392-3404. doi: 10.1093/brain/awab279.
7. Siddiqi ZA, Nowak RJ, Mozaffar T, O'Brien F, Yountz M, Patti F; REGAIN Study Group. Eculizumab in refractory generalized myasthenia gravis previously treated with rituximab: subgroup analysis of REGAIN and its extension study. **Muscle Nerve**. 2021 Dec;64(6):662-669. doi: 10.1002/mus.27422. Epub 2021 Oct 14.
8. Martín-Aguilar L, Lleixà C, Pascual-Goñi E, Caballero-Ávila M, Martínez-Martínez L, Díaz-Manera J, Rojas-García R, Cortés-Vicente E, Turon-Sans J, de Luna N, Suárez-Calvet X, Gallardo E, Rajabally Y, Scotton S, Jacobs BC, Baars A, Cortese A, Vegezzi E, Höftberger R, Zimprich F, Roesler C, Nobile-Orazio E, Liberatore G, Hiew FL, Martínez-Piñeiro A, Carvajal A, Piñar-Morales R, Usón-Martín M, Albertí O, López-Pérez MÁ, Márquez F, Pardo-Fernández J, Muñoz-Delgado L, Cabrera-Serrano M, Ortiz N, Bartolomé M, Duman Ö, Bril V, Segura-Chávez D, Pitarokoili K, Steen C, Illa I, Querol L. Clinical and Laboratory Features in Anti-NF155

Autoimmune Nodopathy. **Neurol Neuroimmunol Neuroinflamm.** 2021 Nov 2;9(1):e1098. doi: 10.1212/NXI.0000000000001098. Print 2022 Jan.

9. Lleixà C, Martín-Aguilar L, Pascual-Goñi E, Franco T, Caballero M, de Luna N, Gallardo E, Suárez-Calvet X, Martínez-Martínez L, Díaz-Manera J, Rojas-García R, Cortés-Vicente E, Turón J, Casasnovas C, Homedes C, Gutiérrez-Gutiérrez G, Jimeno-Montero MC, Berciano J, Sedano-Tous MJ, García-Sobrino T, Pardo-Fernández J, Márquez-Infante C, Rojas-Marcos I, Jericó-Pascual I, Martínez-Hernández E, Morís de la Tassa G, Domínguez-González C, Juárez C, Illa I, Querol L. Autoantibody screening in Guillain-Barré syndrome. **J Neuroinflammation.** 2021 Nov 1;18(1):251. doi: 10.1186/s12974-021-02301-0.
10. Suárez-Calvet X, Fernández-Simón E, Piñol-Jurado P, Alonso-Pérez J, Carrasco-Rozas A, Lleixà C, López-Fernández S, Pons G, Soria L, Bigot A, Mouly V, Illa I, Gallardo E, Jaiswal JK, Díaz-Manera J. Isolation of human fibroadipogenic progenitors and satellite cells from frozen muscle biopsies. **FASEB J.** 2021 Sep;35(9):e21819. doi: 10.1096/fj.202100588R.
11. Cortés-Vicente E, Álvarez-Velasco R, Illa I, Gallardo E. Comment to "Autoantibodies to cortactin and agrin in sera of patients with myasthenia gravis". **J Neuroimmunol.** 2021 Sep 15;358:577659. doi: 10.1016/j.jneuroim.2021.577659. Epub 2021 Jul 13.
12. Alonso-Pérez J, Casasús A, Gimenez-Muñoz Á, Duff J, Rojas-García R, Illa I, Straub V, Töpf A, Díaz-Manera J. Late onset Sandhoff disease presenting with lower motor neuron disease and stuttering. **Neuromuscul Disord.** 2021 Aug;31(8):769-772. doi: 10.1016/j.nmd.2021.04.011. Epub 2021 May 27.
13. Álvarez-Velasco R, Gutiérrez-Gutiérrez G, Trujillo JC, Martínez E, Segovia S, Arribas-Velasco M, Fernández G, Paradas C, Vélez-Gómez B, Casasnovas C, Nedkova V, Guerrero-Sola A, Ramos-Fransi A, Martínez-Piñeiro A, Pardo J, Sevilla T, Gómez-Caravaca MT, López de Munain A, Jericó I, Pelayo-Negro AL, Martín MA, Morgado Y, Mendoza MD, Pérez-Pérez H, Rojas-García R, Turon-Sans J, Querol L, Gallardo E, Illa I, Cortés-Vicente E. Clinical characteristics and outcomes of thymoma-associated myasthenia gravis. **Eur J Neurol.** 2021 Jun;28(6):2083-2091. doi: 10.1111/ene.14820. Epub 2021 Mar 30.
14. Alonso-Jiménez A, Fernández-Simón E, Natera-de Benito D, Ortez C, García C, Montiel E, Belmonte I, Pedrosa I, Segovia S, Piñol-Jurado P, Carrasco-Rozas A, Suárez-Calvet X, Jimenez-Mallebrera C, Nascimento A, Llauger J, Nuñez-Peralta C, Montesinos P, Alonso-Pérez J, Gallardo E, Illa I, Díaz-Manera J. Platelet Derived Growth Factor-AA Correlates With Muscle Function Tests and Quantitative Muscle Magnetic Resonance in Dystrophinopathies. **Front Neurol.** 2021 Jun 11;12:659922. doi: 10.3389/fneur.2021.659922. eCollection 2021.
15. Pascual-Goñi E, Fehmi J, Lleixà C, Martín-Aguilar L, Devaux J, Höftberger R, Delmont E, Doppler K, Sommer C, Radunovic A, Carvajal A, Smyth S, Williams L, Mazanec R, Potočková V, Hinds N, Cassereau J, Viala K, Lefilliatre M, Nicolas G, Foley P, Leypoldt F, Keddie S, Lunn MP, Zimprich F, Nunkoo VS, Löscher WN, Martínez-Martínez L, Díaz-Manera J, Rojas-García R, Illa I, Rinaldi S, Querol L. Antibodies to the Caspr1/contactin-1 complex in chronic inflammatory demyelinating polyradiculoneuropathy. **Brain.** 2021 May 7;144(4):1183-1196. doi: 10.1093/brain/awab014.
16. Pascual-Goñi E, Martín-Aguilar L, Lleixà C, Martínez-Martínez L, Simón-Talero MJ, Díaz-Manera J, Cortés-Vicente E, Rojas-García R, Moga E, Juárez C, Illa I, Querol L. Author Correction: Clinical and laboratory features of anti-MAG neuropathy without monoclonal gammopathy. **Sci Rep.** 2021 Feb 10;11(1):3902. doi: 10.1038/s41598-021-81911-3.
17. Mantegazza R, Wolfe GI, Muppidi S, Wiendl H, Fujita KP, O'Brien FL, Booth HDE, Howard JF Jr; REGAIN Study Group. Post-intervention Status in Patients With Refractory Myasthenia Gravis Treated With Eculizumab During REGAIN and Its Open-Label Extension. **Neurology.** 2021 Jan 26;96(4):e610-e618. doi: 10.1212/WNL.0000000000011207. Epub 2020 Nov 23.
18. Narayanaswami P, Sanders DB, Wolfe G, Benatar M, Cea G, Evoli A, Gilhus NE, Illa I, Kuntz NL, Massey J, Melms A, Murai H, Nicolle M, Palace J, Richman D, Verschuuren J. International Consensus Guidance for Management of Myasthenia Gravis: 2020 Update. **Neurology.** 2021 Jan 19;96(3):114-122. doi: 10.1212/WNL.0000000000011124. Epub 2020 Nov 3.
19. Muppidi S, Guptill JT, Jacob S, Li Y, Farrugia ME, Guidon AC, Tavee JO, Kaminski H, Howard JF Jr, Cutter G, Wiendl H, Maas MB, Illa I, Mantegazza R, Murai H, Utsugisawa K, Nowak RJ;

- CARE-MG Study Group. COVID-19-associated risks and effects in myasthenia gravis (CARE-MG). **Lancet Neurol.** 2020 Dec;19(12):970-971. doi: 10.1016/S1474-4422(20)30413-0.
20. Savarese M, Vihola A, Oates EC, Barresi R, Fiorillo C, Tasca G, Jokela M, Sarkozy A, Luo S, Díaz-Manera J, Ehrstedt C, Rojas-García R, Sáenz A, Muelas N, Lonardo F, Fodstad H, Qureshi T, Johari M, Välipakka S, Luque H, Petiot P, de Munain AL, Pane M, Mercuri E, Torella A, Nigro V, Astrea G, Santorelli FM, Bruno C, Kuntzer T, Illa I, Vílchez JJ, Julien C, Ferreiro A, Malandrini A, Zhao CB, Casar-Borota O, Davis M, Muntoni F, Hackman P, Udd B. Genotype-phenotype correlations in recessive titinopathies. **Genet Med.** 2020 Dec;22(12):2029-2040. doi: 10.1038/s41436-020-0914-2. Epub 2020 Aug 11.
 21. Fernández-Simón E, Lleixà C, Suarez-Calvet X, Diaz-Manera J, Illa I, Gallardo E, de Luna N. Proteasome inhibitors reduce thrombospondin-1 release in human dysferlin-deficient myotubes. **BMC Musculoskelet Disord.** 2020 Nov 27;21(1):784. doi: 10.1186/s12891-020-03756-7.
 22. Martín-Aguilar L, Camps-Renom P, Lleixà C, Pascual-Goñi E, Díaz-Manera J, Rojas-García R, De Luna N, Gallardo E, Cortés-Vicente E, Muñoz L, Alcolea D, Lleó A, Casanovas C, Homedes C, Gutiérrez-Gutiérrez G, Jimeno-Montero MC, Berciano J, Sedano-Tous MJ, García-Sobrino T, Pardo-Fernández J, Márquez-Infante C, Rojas-Marcos I, Jericó-Pascual I, Martínez-Hernández E, Morís de la Tassa G, Domínguez-González C, Illa I, Querol L. Serum neurofilament light chain predicts long-term prognosis in Guillain-Barré syndrome patients. **J Neurol Neurosurg Psychiatry.** 2020 Nov 5:jnnp-2020-323899. doi: 10.1136/jnnp-2020-323899. Online ahead of print.
 23. Alonso-Pérez J, González-Quereda L, Bello L, Guglieri M, Straub V, Gallano P, Semplicini C, Pegoraro E, Zangaro V, Nascimento A, Ortez C, Comi GP, Dam LT, De Visser M, van der Kooi AJ, Garrido C, Santos M, Schara U, Gangfuß A, Løkken N, Storgaard JH, Vissing J, Schoser B, Dekomien G, Udd B, Palmio J, D'Amico A, Politano L, Nigro V, Bruno C, Panicucci C, Sarkozy A, Abdel-Mannan O, Alonso-Jimenez A, Claeys KG, Gomez-Andrés D, Munell F, Costa-Comellas L, Haberlová J, Rohlenová M, Elke V, De Bleecker JL, Dominguez-González C, Tasca G, Weiss C, Deconinck N, Fernández-Torrón R, López de Munain A, Camacho-Salas A, Melegh B, Hadzsiev K, Leonardis L, Koritnik B, Garibaldi M, de Leon-Hernández JC, Malfatti E, Fraga-Bau A, Richard I, Illa I, Díaz-Manera J. New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. **Brain.** 2020 Sep 1;143(9):2696-2708. doi: 10.1093/brain/awaa228.
 24. Lee I, Kuo HC, Aban IB, Cutter GR, McPherson T, Kaminski HJ, Sussman J, Ströbel P, Oger J, Cea G, Heckmann JM, Evoli A, Nix W, Ciafaloni E, Antonini G, Witoonpanich R, King JO, Beydoun SR, Chalk CH, Barboi AC, Amato AA, Shaibani AI, Katirji B, Lecky BRF, Buckley C, Vincent A, Dias-Tosta E, Yoshikawa H, Waddington-Cruz M, Pulley MT, Rivner MH, Kostera-Pruszczyk A, Pascuzzi RM, Jackson CE, Verschuuren JJG, Massey JM, Kissel JT, Werneck LC, Benatar M, Barohn RJ, Tandan R, Mozaffar T, Conwit R, Minisman G, Sonett JR, Wolfe GI; MGTX study group. Minimal manifestation status and prednisone withdrawal in the MGTX trial. **Neurology.** 2020 Aug 11;95(6):e755-e766. doi: 10.1212/WNL.0000000000010031. Epub 2020 Jul 1.
 25. Nuñez-Peralta C, Alonso-Pérez J, Llauger J, Segovia S, Montesinos P, Belmonte I, Pedrosa I, Montiel E, Alonso-Jiménez A, Sánchez-González J, Martínez-Noguera A, Illa I, Díaz-Manera J. Follow-up of late-onset Pompe disease patients with muscle magnetic resonance imaging reveals increase in fat replacement in skeletal muscles. **J Cachexia Sarcopenia Muscle.** 2020 Aug;11(4):1032-1046. doi: 10.1002/jcsm.12555. Epub 2020 Mar 4.
 26. Mantegazza R, O'Brien FL, Yountz M, Howard JF Jr; REGAIN study group. Consistent improvement with eculizumab across muscle groups in myasthenia gravis. **Ann Clin Transl Neurol.** 2020 Aug;7(8):1327-1339. doi: 10.1002/acn3.51121. Epub 2020 Jul 22.
 27. Vissing J, Jacob S, Fujita KP, O'Brien F, Howard JF; REGAIN study group. 'Minimal symptom expression' in patients with acetylcholine receptor antibody-positive refractory generalized myasthenia gravis treated with eculizumab. **J Neurol.** 2020 Jul;267(7):1991-2001. doi: 10.1007/s00415-020-09770-y. Epub 2020 Mar 18.
 28. International MG/COVID-19 Working Group, Jacob S, Muppidi S, Guidon A, Guptill J, Hehir M, Howard JF Jr, Illa I, Mantegazza R, Murai H, Utsugisawa K, Vissing J, Wiendl H, Nowak RJ. Guidance for the management of myasthenia gravis (MG) and Lambert-Eaton myasthenic

- syndrome (LEMS) during the COVID-19 pandemic. **J Neurol Sci.** 2020 May 15;412:116803. doi: 10.1016/j.jns.2020.116803. Epub 2020 Mar 25.
29. Suárez-Calvet X, Alonso-Pérez J, Castellví I, Carrasco-Rozas A, Fernández-Simón E, Zamora C, Martínez-Martínez L, Alonso-Jiménez A, Rojas-García R, Turón J, Querol L, de Luna N, Milena-Millan A, Corominas H, Castillo D, Cortés-Vicente E, Illa I, Gallardo E, Díaz-Manera J. Thrombospondin-1 mediates muscle damage in brachio-cervical inflammatory myopathy and systemic sclerosis. **Neurol Neuroimmunol Neuroinflamm.** 2020 Mar 6;7(3):e694. doi: 10.1212/NXI.0000000000000694. Print 2020 May.
 30. Cortés-Vicente E, Álvarez-Velasco R, Segovia S, Paradas C, Casasnovas C, Guerrero-Sola A, Pardo J, Ramos-Fransi A, Sevilla T, López de Munain A, Gómez MT, Jericó I, Gutiérrez-Gutiérrez G, Pelayo-Negro AL, Martín MA, Mendoza MD, Morís G, Rojas-García R, Díaz-Manera J, Querol L, Gallardo E, Vélez B, Albertí MA, Galán L, García-Sobrino T, Martínez-Piñeiro A, Lozano-Veintimilla A, Fernández-Torrón R, Cano-Abascal Á, Illa I. Clinical and therapeutic features of myasthenia gravis in adults based on age at onset. **Neurology.** 2020 Mar 17;94(11):e1171-e1180. doi: 10.1212/WNL.0000000000008903. Epub 2020 Feb 18.
 31. Verdú-Díaz J, Alonso-Pérez J, Nuñez-Peralta C, Tasca G, Vissing J, Straub V, Fernández-Torrón R, Llauger J, Illa I, Díaz-Manera J. Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. **Neurology.** 2020 Mar 10;94(10):e1094-e1102. doi: 10.1212/WNL.0000000000009068. Epub 2020 Feb 6.
 32. De Luna N, Turon-Sans J, Cortes-Vicente E, Carrasco-Rozas A, Illán-Gala I, Dols-Icardo O, Clarimón J, Lleó A, Gallardo E, Illa I, Rojas-García R. Downregulation of miR-335-5P in Amyotrophic Lateral Sclerosis Can Contribute to Neuronal Mitochondrial Dysfunction and Apoptosis. **Sci Rep.** 2020 Mar 9;10(1):4308. doi: 10.1038/s41598-020-61246-1.
 33. Alonso-Pérez J, Segovia S, Domínguez-González C, Olivé M, Mendoza Grimón MD, Fernández-Torrón R, López de Munain A, Muñoz-Blanco JL, Ramos-Fransi A, Almendrote M, Illa I, Díaz-Manera J. Spanish Pompe registry: Baseline characteristics of first 49 patients with adult onset of Pompe disease. **Med Clin (Barc).** 2020 Feb 14;154(3):80-85. doi: 10.1016/j.medcli.2019.03.036. Epub 2019 Jun 26.
 34. Murai H, Uzawa A, Suzuki Y, Imai T, Shiraishi H, Suzuki H, Okumura M, O'Brien F, Wang JJ, Fujita KP, Utsugisawa K; REGAIN Study Group. Long-term efficacy and safety of eculizumab in Japanese patients with generalized myasthenia gravis: A subgroup analysis of the REGAIN open-label extension study. **J Neurol Sci.** 2019 Dec 15;407:116419. doi: 10.1016/j.jns.2019.08.004. Epub 2019 Aug 3.
 35. Fernández-Simón E, Carrasco-Rozas A, Gallardo E, González-Quereda L, Alonso-Pérez J, Belmonte I, Pedrosa-Hernández I, Montiel E, Segovia S, Suárez-Calvet X, Llauger J, Mayos M, Illa I, Barba-Romero MA, Barcena J, Paradas C, Carzorla MR, Creus C, Coll-Cantí J, Díaz M, Domínguez C, Fernández-Torrón R, García-Antelo MJ, Grau JM, López de Munain A, Martínez-García FA, Morgado Y, Moreno A, Morís G, Muñoz-Blanco MA, Nascimento A, Parajuá-Pozo JL, Querol L, Rojas R, Robledo-Strauss A, Rojas-Marcos Í, Salazar JA, Usón M, Díaz-Manera J. Study of the effect of anti-rhGAA antibodies at low and intermediate titers in late onset Pompe patients treated with ERT. **Mol Genet Metab.** 2019 Sep-Oct;128(1-2):129-136. doi: 10.1016/j.ymgme.2019.07.013. Epub 2019 Jul 23.
 36. Andersen H, Mantegazza R, Wang JJ, O'Brien F, Patra K, Howard JF Jr; REGAIN Study Group. Correction to: Eculizumab improves fatigue in refractory generalized myasthenia gravis. **Qual Life Res.** 2019 Aug;28(8):2255. doi: 10.1007/s11136-019-02204-x.
 37. Andersen H, Mantegazza R, Wang JJ, O'Brien F, Patra K, Howard JF Jr; REGAIN Study Group. Eculizumab improves fatigue in refractory generalized myasthenia gravis. **Qual Life Res.** 2019 Aug;28(8):2247-2254. doi: 10.1007/s11136-019-02148-2. Epub 2019 Mar 23.
 38. Muppidi S, Utsugisawa K, Benatar M, Murai H, Barohn RJ, Illa I, Jacob S, Vissing J, Burns TM, Kissel JT, Nowak RJ, Andersen H, Casasnovas C, de Bleecker JL, Vu TH, Mantegazza R, O'Brien FL, Wang JJ, Fujita KP, Howard JF Jr; Regain Study Group. Long-term safety and efficacy of eculizumab in generalized myasthenia gravis. **Muscle Nerve.** 2019 Jul;60(1):14-24. doi: 10.1002/mus.26447. Epub 2019 Mar 8.
 39. Carrasco-Rozas A, Fernández-Simón E, Lleixà MC, Belmonte I, Pedrosa-Hernandez I, Montiel-Morillo E, Nuñez-Peralta C, Llauger Rossello J, Segovia S, De Luna N, Suarez-Calvet X, Illa I;

- Pompe Spanish Study group, Díaz-Manera J, Gallardo E. Identification of serum microRNAs as potential biomarkers in Pompe disease. *Ann Clin Transl Neurol*. 2019 Jul;6(7):1214-1224. doi: 10.1002/acn3.50800. Epub 2019 Jun 12.
40. Alonso-Jimenez A, Kroon RHMJM, Alejaldre-Monforte A, Nuñez-Peralta C, Horlings CGC, van Engelen BGM, Olivé M, González L, Verges-Gil E, Paradas C, Márquez C, Garibaldi M, Gallano P, Rodriguez MJ, Gonzalez-Quereda L, Dominguez Gonzalez C, Vissing J, Fornander F, Eisum AV, García-Sobrino T, Pardo J, García-Figueiras R, Muelas N, Vilchez JJ, Kapetanovic S, Tasca G, Monforte M, Ricci E, Gomez MT, Bevilacqua JA, Diaz-Jara J, Zamorano II, Carlier RY, Laforet P, Pelayo-Negro A, Ramos-Fransi A, Martínez A, Marini-Bettolo C, Straub V, Gutiérrez G, Stojkovic T, Martín MA, Morís G, Fernández-Torrón R, Lopez De Munaín A, Cortes-Vicente E, Querol L, Rojas-García R, Illa I, Diaz-Manera J. Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. *J Neurol Neurosurg Psychiatry*. 2019 May;90(5):576-585. doi: 10.1136/jnnp-2018-319578. Epub 2018 Dec 8.
 41. Duchateau L, Martín-Aguilar L, Lleixà C, Cortese A, Dols-Icardo O, Cervera-Carles L, Pascual-Goñi E, Diaz-Manera J, Callegari I, Franciotta D, Rojas-Garcia R, Illa I, Clarimon J, Querol L. Correction: Absence of pathogenic mutations in CD59 in chronic inflammatory demyelinating polyradiculoneuropathy. *PLoS One*. 2019 Apr 17;14(4):e0215784. doi: 10.1371/journal.pone.0215784. eCollection 2019.
 42. Pascual-Goñi E, Martín-Aguilar L, Lleixà C, Martínez-Martínez L, Simón-Talero MJ, Díaz-Manera J, Cortés-Vicente E, Rojas-García R, Moga E, Juárez C, Illa I, Querol L. Clinical and laboratory features of anti-MAG neuropathy without monoclonal gammopathy. *Sci Rep*. 2019 Apr 16;9(1):6155. doi: 10.1038/s41598-019-42545-8.
 43. Ruiz-García R, Martínez-Hernández E, García-Ormaechea M, Español-Rego M, Sabater L, Querol L, Illa I, Dalmau J, Graus F. Caveats and Pitfalls of SOX1 Autoantibody Testing With a Commercial Line Blot Assay in Paraneoplastic Neurological Investigations. *Front Immunol*. 2019 Apr 12;10:769. doi: 10.3389/fimmu.2019.00769. eCollection 2019.
 44. Wolfe GI, Kaminski HJ, Aban IB, Minisman G, Kuo HC, Marx A, Ströbel P, Mazia C, Oger J, Cea JG, Heckmann JM, Evoli A, Nix W, Ciafaloni E, Antonini G, Witoonpanich R, King JO, Beydoun SR, Chalk CH, Barboi AC, Amato AA, Shaibani AI, Katirji B, Lecky BRF, Buckley C, Vincent A, Dias-Tosta E, Yoshikawa H, Waddington-Cruz M, Pulley MT, Rivner MH, Kostera-Pruszczyk A, Pascuzzi RM, Jackson CE, Verschuuren JJGM, Massey JM, Kissel JT, Werneck LC, Benatar M, Barohn RJ, Tandan R, Mozaffar T, Silvestri NJ, Conwit R, Sonett JR, Jaretzki A 3rd, Newsom-Davis J, Cutter GR; MGTX Study Group. Long-term effect of thymectomy plus prednisone versus prednisone alone in patients with non-thymomatous myasthenia gravis: 2-year extension of the MGTX randomised trial. *Lancet Neurol*. 2019 Mar;18(3):259-268. doi: 10.1016/S1474-4422(18)30392-2. Epub 2019 Jan 25.
 45. Manso C, Querol L, Lleixà C, Poncelet M, Mekaouche M, Vallat JM, Illa I, Devaux JJ. Anti-Neurofascin-155 IgG4 antibodies prevent paranodal complex formation in vivo. *J Clin Invest*. 2019 Mar 14;129(6):2222-2236. doi: 10.1172/JCI124694.
 46. Duchateau L, Martín-Aguilar L, Lleixà C, Cortese A, Dols-Icardo O, Cervera-Carles L, Pascual-Goñi E, Diaz-Manera J, Callegari I, Franciotta D, Rojas-Garcia R, Illa I, Clarimon J, Querol L. Absence of pathogenic mutations in CD59 in chronic inflammatory demyelinating polyradiculoneuropathy. *PLoS One*. 2019 Feb 22;14(2):e0212647. doi: 10.1371/journal.pone.0212647. eCollection 2019.
 47. Fernández-Simón E, Carrasco-Rozas A, Gallardo E, Figueroa-Bonaparte S, Belmonte I, Pedrosa I, Montiel E, Suárez-Calvet X, Alonso-Pérez J, Segovia S, Nuñez-Peralta C, Llauger J, Mayos M, Illa I; Spanish Pompe Study Group, Díaz-Manera J. PDGF-BB serum levels are decreased in adult onset Pompe patients. *Sci Rep*. 2019 Feb 14;9(1):2139. doi: 10.1038/s41598-018-38025-0.
 48. Diaz-Manera J, Fernandez-Torrón R, Llauger J, James MK, Mayhew A, Smith FE, Moore UR, Blamire AM, Carlier PG, Rufibach L, Mittal P, Eagle M, Jacobs M, Hodgson T, Wallace D, Ward L, Smith M, Stramare R, Rampado A, Sato N, Tamaru T, Harwick B, Rico Gala S, Turk S, Copenrath EM, Foster G, Bendahan D, Le Fur Y, Fricke ST, Otero H, Foster SL, Peduto A, Sawyer AM, Hilsden H, Lochmuller H, Grieben U, Spuler S, Tesi Rocha C, Day JW, Jones KJ, Bharucha-Goebel DX, Salort-Campana E, Harms M, Pestronk A, Krause S, Schreiber-Katz O, Walter MC, Paradas C, Hogrel JY, Stojkovic T, Takeda S, Mori-Yoshimura M, Bravver E, Sparks S, Bello L, Semplicini C, Pegoraro E, Mendell JR, Bushby K, Straub V; Jain COS Consortium.

- Muscle MRI in patients with dysferlinopathy: pattern recognition and implications for clinical trials. **J Neurol Neurosurg Psychiatry**. 2018 Oct;89(10):1071-1081. doi: 10.1136/jnnp-2017-317488. Epub 2018 May 7.
49. Doets AY, Verboon C, van den Berg B, Harbo T, Cornblath DR, Willison HJ, Islam Z, Attarian S, Barroso FA, Bateman K, Benedetti L, van den Bergh P, Casasnovas C, Cavaletti G, Chavada G, Claeys KG, Dardiotis E, Davidson A, van Doorn PA, Feasby TE, Galassi G, Gorson KC, Hartung HP, Hsieh ST, Hughes RAC, Illa I, Islam B, Kusunoki S, Kuwabara S, Lehmann HC, Miller JAL, Mohammad QD, Monges S, Nobile Orazio E, Pardo J, Pereon Y, Rinaldi S, Querol L, Reddel SW, Reisin RC, Shahrizaila N, Sindrup SH, Waqar W, Jacobs BC; IGOS Consortium. Regional variation of Guillain-Barré syndrome. **Brain**. 2018 Oct 1;141(10):2866-2877. doi: 10.1093/brain/awy232.
 50. De Luna N, Suarez-Calvet X, Garicano M, Fernandez-Simon E, Rojas-García R, Diaz-Manera J, Querol L, Illa I, Gallardo E. Effect of MAPK Inhibition on the Differentiation of a Rhabdomyosarcoma Cell Line Combined With CRISPR/Cas9 Technology: An In Vitro Model of Human Muscle Diseases. **J Neuropathol Exp Neurol**. 2018 Oct 1;77(10):964-972. doi: 10.1093/jnen/nly078.
 51. Gonzalez-Quereda L, Gallardo E, Töpf A, Alonso-Jimenez A, Straub V, Rodriguez MJ, Lleixa C, Illa I, Gallano P, Diaz-Manera J. A new mutation of the SCGA gene is the cause of a late onset mild phenotype limb girdle muscular dystrophy type 2D with axial involvement. **Neuromuscul Disord**. 2018 Aug;28(8):633-638. doi: 10.1016/j.nmd.2018.06.002. Epub 2018 Jul 13.
 52. Hughes R, Dalakas MC, Merkies I, Latov N, Léger JM, Nobile-Orazio E, Sobue G, Genge A, Cornblath D, Merschhemke M, Ervin CM, Agoropoulou C, Hartung HP; FORCIDP Trial Investigators. Oral fingolimod for chronic inflammatory demyelinating polyradiculoneuropathy (FORCIDP Trial): a double-blind, multicentre, randomised controlled trial. **Lancet Neurol**. 2018 Aug;17(8):689-698. doi: 10.1016/S1474-4422(18)30202-3. Epub 2018 Jul 9.
 53. Figueroa-Bonaparte S, Llauger J, Segovia S, Belmonte I, Pedrosa I, Montiel E, Montesinos P, Sánchez-González J, Alonso-Jiménez A, Gallardo E, Illa I; Spanish Pompe group, Díaz-Manera J. Quantitative muscle MRI to follow up late onset Pompe patients: a prospective study. **Sci Rep**. 2018 Jul 18;8(1):10898. doi: 10.1038/s41598-018-29170-7.
 54. Piñol-Jurado P, Suárez-Calvet X, Fernández-Simón E, Gallardo E, de la Oliva N, Martínez-Muriana A, Gómez-Gálvez P, Escudero LM, Pérez-Peiró M, Wollin L, de Luna N, Navarro X, Illa I, Díaz-Manera J. Nintedanib decreases muscle fibrosis and improves muscle function in a murine model of dystrophinopathy. **Cell Death Dis**. 2018 Jul 10;9(7):776. doi: 10.1038/s41419-018-0792-6.
 55. Ladislau L, Suárez-Calvet X, Toquet S, Landon-Cardinal O, Amelin D, Depp M, Rodero MP, Hathazi D, Duffy D, Bondet V, Preusse C, Bienvenu B, Rozenberg F, Roos A, Benjamim CF, Gallardo E, Illa I, Mouly V, Stenzel W, Butler-Browne G, Benveniste O, Allenbach Y. JAK inhibitor improves type I interferon induced damage: proof of concept in dermatomyositis. **Brain**. 2018 Jun 1;141(6):1609-1621. doi: 10.1093/brain/awy105.
 56. Cortés-Vicente E, Turon-Sans J, Gelpi E, Clarimón J, Borrego-Écija S, Dols-Icardo O, Illán-Gala I, Lleó A, Illa I, Blesa R, Al-Chalabi A, Rojas-García R. Distinct Clinical Features and Outcomes in Motor Neuron Disease Associated with Behavioural Variant Frontotemporal Dementia. **Dement Geriatr Cogn Disord**. 2018;45(3-4):220-231. doi: 10.1159/000488528. Epub 2018 Jun 8.
 57. Cortés-Vicente E, Rojas-García R, Díaz-Manera J, Querol L, Casasnovas C, Guerrero-Sola A, Muñoz-Blanco JL, Bárcena-Llona JE, Márquez-Infante C, Pardo J, Martínez-Fernández EM, Usón M, Oliva-Nacarino P, Sevilla T, Illa I. The impact of rituximab infusion protocol on the long-term outcome in anti-MuSK myasthenia gravis. **Ann Clin Transl Neurol**. 2018 Apr 14;5(6):710-716. doi: 10.1002/acn3.564. eCollection 2018 Jun.
 58. Siles AM, Martínez-Hernández E, Araque J, Diaz-Manera J, Rojas-García R, Gallardo E, Illa I, Graus F, Querol L. Antibodies against cell adhesion molecules and neural structures in paraneoplastic neuropathies. **Ann Clin Transl Neurol**. 2018 Mar 26;5(5):559-569. doi: 10.1002/acn3.554. eCollection 2018 May.

59. Painous C, López-Pérez MÁ, Illa I, Querol L. Head and voice tremor improving with immunotherapy in an anti-NF155 positive CIDP patient. **Ann Clin Transl Neurol.** 2018 Mar 7;5(4):499-501. doi: 10.1002/acn3.539. eCollection 2018 Apr.
60. Díaz-Manera J, Sotoca-Fernández J, Alonso-Jiménez A, Marzo ME, Gallardo E, Segovia-Simón S, Siles AM, Illa I, Pagonabarraga J. McLeod syndrome is a new cause of axial muscle weakness. **Muscle Nerve.** 2018 Jan 30. doi: 10.1002/mus.26086. Online ahead of print.
61. Illa I, Cortés-Vicente E, Martínez MÁ, Gallardo E. Diagnostic utility of cortactin antibodies in myasthenia gravis. **Ann N Y Acad Sci.** 2018 Jan;1412(1):90-94. doi: 10.1111/nyas.13502. Epub 2017 Oct 25.
62. Illa I. ARTHUR ASBURY LECTURE: Chronic inflammatory demyelinating polyradiculoneuropathy: clinical aspects and new animal models of auto-immunity to nodal components. **J Peripher Nerv Syst.** 2017 Dec;22(4):418-424. doi: 10.1111/jns.12237. Epub 2017 Nov 15.
63. Howard JF Jr, Utsugisawa K, Benatar M, Murai H, Barohn RJ, Illa I, Jacob S, Vissing J, Burns TM, Kissel JT, Muppidi S, Nowak RJ, O'Brien F, Wang JJ, Mantegazza R; REGAIN Study Group. Safety and efficacy of eculizumab in anti-acetylcholine receptor antibody-positive refractory generalised myasthenia gravis (REGAIN): a phase 3, randomised, double-blind, placebo-controlled, multicentre study. **Lancet Neurol.** 2017 Dec;16(12):976-986. doi: 10.1016/S1474-4422(17)30369-1. Epub 2017 Oct 20.
64. Martínez-Martínez L, Lleixà MC, Boera-Carnicero G, Cortese A, Devaux J, Siles A, Rajabally Y, Martínez-Piñeiro A, Carvajal A, Pardo J, Delmont E, Attarian S, Díaz-Manera J, Callegari I, Marchioni E, Franciotta D, Benedetti L, Lauria G, de la Calle Martín O, Juárez C, Illa I, Querol L. Anti-NF155 chronic inflammatory demyelinating polyradiculoneuropathy strongly associates to HLA-DRB15. **J Neuroinflammation.** 2017 Nov 16;14(1):224. doi: 10.1186/s12974-017-0996-1.
65. Querol L, Siles AM, Alba-Rovira R, Jáuregui A, Devaux J, Faivre-Sarrailh C, Araque J, Rojas-García R, Díaz-Manera J, Cortés-Vicente E, Nogales-Gadea G, Navas-Madroñal M, Gallardo E, Illa I. Antibodies against peripheral nerve antigens in chronic inflammatory demyelinating polyradiculoneuropathy. **Sci Rep.** 2017 Oct 31;7(1):14411. doi: 10.1038/s41598-017-14853-4.
66. Querol L, Devaux J, Rojas-García R, Illa I. Autoantibodies in chronic inflammatory neuropathies: diagnostic and therapeutic implications. **Nat Rev Neurol.** 2017 Sep;13(9):533-547. doi: 10.1038/nrneurol.2017.84. Epub 2017 Jul 14.
67. Cortés-Vicente E, Pradas J, Marín-Lahoz J, De Luna N, Clarimón J, Turon-Sans J, Gelpí E, Díaz-Manera J, Illa I, Rojas-García R. Early diagnosis of amyotrophic lateral sclerosis mimic syndromes: pros and cons of current clinical diagnostic criteria. **Amyotroph Lateral Scler Frontotemporal Degener.** 2017 Aug;18(5-6):333-340. doi: 10.1080/21678421.2017.1316408. Epub 2017 Apr 25.
68. De Luna N, Suárez-Calvet X, Lleixà C, Díaz-Manera J, Olivé M, Illa I, Gallardo E. Hypoxia triggers IFN-I production in muscle: Implications in dermatomyositis. **Sci Rep.** 2017 Aug 17;7(1):8595. doi: 10.1038/s41598-017-09309-8.
69. Piñol-Jurado P, Gallardo E, de Luna N, Suárez-Calvet X, Sánchez-Riera C, Fernández-Simón E, Gomis C, Illa I, Díaz-Manera J. Platelet-Derived Growth Factor BB Influences Muscle Regeneration in Duchenne Muscle Dystrophy. **Am J Pathol.** 2017 Aug;187(8):1814-1827. doi: 10.1016/j.ajpath.2017.04.011. Epub 2017 Jun 12.
70. Sivera R, Frasquet M, Lupo V, García-Sobrino T, Blanco-Arias P, Pardo J, Fernández-Torrón R, de Munain AL, Márquez-Infante C, Villarreal L, Carbonell P, Rojas-García R, Segovia S, Illa I, Frongia AL, Nascimento A, Ortez C, García-Romero MDM, Pascual SI, Pelayo-Negro AL, Berciano J, Guerrero A, Casasnovas C, Camacho A, Esteban J, Chumillas MJ, Barreiro M, Díaz C, Palau F, Vélchez JJ, Espinós C, Sevilla T. Distribution and genotype-phenotype correlation of GDAP1 mutations in Spain. **Sci Rep.** 2017 Jul 27;7(1):6677. doi: 10.1038/s41598-017-06894-6.
71. Suárez-Calvet X, Gallardo E, Pinal-Fernandez I, De Luna N, Lleixà C, Díaz-Manera J, Rojas-García R, Castellví I, Martínez MA, Grau JM, Selva-O'Callaghan A, Illa I. RIG-I expression in perifascicular myofibers is a reliable biomarker of dermatomyositis. **Arthritis Res Ther.** 2017 Jul 24;19(1):174. doi: 10.1186/s13075-017-1383-0.

72. Delmont E, Manso C, Querol L, Cortese A, Berardinelli A, Lozza A, Belghazi M, Malissart P, Labauge P, Taieb G, Yuki N, Illa I, Attarian S, Devaux JJ. Autoantibodies to nodal isoforms of neurofascin in chronic inflammatory demyelinating polyneuropathy. **Brain**. 2017 Jul 1;140(7):1851-1858. doi: 10.1093/brain/awx124.
73. Aragonés JM, Altimiras J, Roura-Poch P, Homs E, Bajo L, Povedano M, Cortés-Vicente E, Illa I, Al-Chalabi A, Rojas-García R. Amyotrophic lateral sclerosis: A higher than expected incidence in people over 80 years of age. **Amyotroph Lateral Scler Frontotemporal Degener**. 2016 Oct-Nov;17(7-8):522-527. doi: 10.1080/21678421.2016.1187175. Epub 2016 May 25.
74. Figueroa-Bonaparte S, Segovia S, Llauger J, Belmonte I, Pedrosa I, Alejaldre A, Mayos M, Suárez-Cuartín G, Gallardo E, Illa I, Díaz-Manera J; Spanish Pompe Study Group. Muscle MRI Findings in Childhood/Adult Onset Pompe Disease Correlate with Muscle Function. **PLoS One**. 2016 Oct 6;11(10):e0163493. doi: 10.1371/journal.pone.0163493. eCollection 2016.
75. Cortés-Vicente E, Gallardo E, Martínez MÁ, Díaz-Manera J, Querol L, Rojas-García R, Illa I. Clinical Characteristics of Patients With Double-Seronegative Myasthenia Gravis and Antibodies to Contactin. **JAMA Neurol**. 2016 Sep 1;73(9):1099-104. doi: 10.1001/jamaneurol.2016.2032.
76. Wolfe GI, Kaminski HJ, Aban IB, Minisman G, Kuo HC, Marx A, Ströbel P, Mazia C, Oger J, Cea JG, Heckmann JM, Evoli A, Nix W, Ciafaloni E, Antonini G, Witoonpanich R, King JO, Beydoun SR, Chalk CH, Barboi AC, Amato AA, Shaibani AI, Katirji B, Lecky BR, Buckley C, Vincent A, Dias-Tosta E, Yoshikawa H, Waddington-Cruz M, Pulley MT, Rivner MH, Kostera-Pruszczyk A, Pascuzzi RM, Jackson CE, Garcia Ramos GS, Verschuuren JJ, Massey JM, Kissel JT, Werneck LC, Benatar M, Barohn RJ, Tandan R, Mozaffar T, Conwit R, Odenkirchen J, Sonett JR, Jaretzki A 3rd, Newsom-Davis J, Cutter GR; MGTX Study Group. Randomized Trial of Thymectomy in Myasthenia Gravis. **N Engl J Med**. 2016 Aug 11;375(6):511-22. doi: 10.1056/NEJMoa1602489.
77. Sanders DB, Wolfe GI, Benatar M, Evoli A, Gilhus NE, Illa I, Kuntz N, Massey JM, Melms A, Murai H, Nicolle M, Palace J, Richman DP, Verschuuren J, Narayanaswami P. International consensus guidance for management of myasthenia gravis: Executive summary. **Neurology**. 2016 Jul 26;87(4):419-25. doi: 10.1212/WNL.0000000000002790. Epub 2016 Jun 29.
78. Manso C, Querol L, Mekaouche M, Illa I, Devaux JJ. Contactin-1 IgG4 antibodies cause paranode dismantling and conduction defects. **Brain**. 2016 Jun;139(Pt 6):1700-12. doi: 10.1093/brain/aww062. Epub 2016 Mar 26.
79. Cruz S, Figueroa-Bonaparte S, Gallardo E, de Becdelièvre A, Gartioux C, Allamand V, Piñol P, García MA, Jiménez-Mallebriera C, Llauger J, González-Rodríguez L, Cortes-Vicente E, Illa I, Díaz-Manera J. Bethlem Myopathy Phenotypes and Follow Up: Description of 8 Patients at the Mildest End of the Spectrum. **J Neuromuscul Dis**. 2016 May 27;3(2):267-274. doi: 10.3233/JND-150135.
80. Suárez-Calvet X, Gallardo E, Illa I. Comment to "Role of Toll-like receptors and retinoic acid inducible gene I in endogenous production of type I interferon in dermatomyositis". **J Neuroimmunol**. 2016 Feb 15;291:125. doi: 10.1016/j.jneuroim.2015.12.008. Epub 2015 Dec 18.
81. Huijbers MG, Vink AF, Niks EH, Westhuis RH, van Zwet EW, de Meel RH, Rojas-García R, Díaz-Manera J, Kuks JB, Klooster R, Straasheijm K, Evoli A, Illa I, van der Maarel SM, Verschuuren JJ. Longitudinal epitope mapping in MuSK myasthenia gravis: implications for disease severity. **J Neuroimmunol**. 2016 Feb 15;291:82-8. doi: 10.1016/j.jneuroim.2015.12.016. Epub 2016 Jan 5.
82. Cruz S, Cortes-Vicente E, Illa I, Rojas-García R. Transthyretin-related hereditary amyloid polyneuropathy presenting with large fibre involvement and cardiomyopathy. **Amyloid**. 2016;23(1):64-5. doi: 10.3109/13506129.2015.1127223. Epub 2016 Jan 23.
83. Díaz-Manera J, Alejaldre A, González L, Olivé M, Gómez-Andrés D, Muelas N, Vílchez JJ, Llauger J, Carbonell P, Márquez-Infante C, Fernández-Torrón R, Poza JJ, López de Munáin A, González-Quereda L, Mirabet S, Clarimon J, Gallano P, Rojas-García R, Gallardo E, Illa I. Muscle imaging in muscle dystrophies produced by mutations in the EMD and LMNA genes. **Neuromuscul Disord**. 2016 Jan;26(1):33-40. doi: 10.1016/j.nmd.2015.10.001. Epub 2015 Oct 22.
84. Díaz-Manera J, Alejaldre A, González L, Olivé M, Gómez-Andrés D, Muelas N, Vílchez JJ, Llauger J, Carbonell P, Márquez-Infante C, Fernández-Torrón R, Poza JJ, López de Munáin A,

- González-Quereda L, Mirabet S, Clarimon J, Gallano P, Rojas-García R, Gallardo E, Illa I. Muscle imaging in muscle dystrophies produced by mutations in the EMD and LMNA genes. **Neuromuscul Disord.** 2015 Oct 22. pii: S0960-8966(15)00743-9. doi: 10.1016/j.nmd.2015.10.001. [Epub ahead of print]
85. Querol L, Rojas-García R, Diaz-Manera J, Barcena J, Pardo J, Ortega-Moreno A, Sedano MJ, Seró-Ballesteros L, Carvajal A, Ortiz N, Gallardo E, Illa I. Rituximab in treatment-resistant CIDP with antibodies against paranodal proteins. **Neurol Neuroimmunol Neuroinflamm.** 2015 Sep 3;2(5):e149. doi: 10.1212/NXI.0000000000000149. eCollection 2015 Oct.
 86. Vanhoutte EK, Faber CG, van Nes SI, Cats EA, Van der Pol WL, Gorson KC, van Doorn PA, Cornblath DR, van den Berg LH, Merkies IS; PeriNomS Study Group. Rasch-built Overall Disability Scale for Multifocal motor neuropathy (MMN-RODS(©)). **J Peripher Nerv Syst.** 2015 Sep;20(3):296-305. doi: 10.1111/jns.12141.
 87. Halter JP, Michael W, Schüpbach M, Mandel H, Casali C, Orchard K, Collin M, Valcarcel D, Rovelli A, Filosto M, Dotti MT, Marotta G, Pintos G, Barba P, Accarino A, Ferra C, Illa I, Beguin Y, Bakker JA, Boelens JJ, de Coo IF, Fay K, Sue CM, Nachbaur D, Zoller H, Sobreira C, Pinto Simoes B, Hammans SR, Savage D, Martí R, Chinnery PF, Elhasid R, Gratwohl A, Hirano M. Allogeneic haematopoietic stem cell transplantation for mitochondrial neurogastrointestinal encephalomyopathy. **Brain.** 2015 Oct;138(Pt 10):2847-58. doi: 10.1093/brain/awv226. Epub 2015 Aug 10.
 88. Querol L, Illa I. Paranodal and other autoantibodies in chronic inflammatory neuropathies. **Curr Opin Neurol.** 2015 Oct;28(5):474-9. doi: 10.1097/WCO.0000000000000233.
 89. Draak TH, Pruppers MH, van Nes SI, Vanhoutte EK, Bakkers M, Gorson KC, Van der Pol WL, Lewis RA, Notermans NC, Nobile-Orazio E, Léger JM, Van den Bergh PY, Lauria G, Bril V, Katzberg H, Lunn MP, Pouget J, van der Kooi AJ, van den Berg LH, van Doorn PA, Cornblath DR, Hahn AF, Faber CG, Merkies IS; PeriNomS study group. Grip strength comparison in immune-mediated neuropathies: Vigorimeter vs. Jamar. **J Peripher Nerv Syst.** 2015 Sep;20(3):269-76. doi: 10.1111/jns.12126.
 90. Pruppers MH, Draak TH, Vanhoutte EK, Van der Pol WL, Gorson KC, Léger JM, Nobile-Orazio E, Lewis RA, van den Berg LH, Faber CG, Merkies IS; PeriNomS study group. Outcome measures in MMN revisited: further improvement needed. **J Peripher Nerv Syst.** 2015 Sep;20(3):306-18. doi: 10.1111/jns.12124.
 91. Vanhoutte EK, Hermans MC, Faber CG, Gorson KC, Merkies IS, Thonnard JL; PeriNomS Study Group. Rasch-ionale for neurologists. **J Peripher Nerv Syst.** 2015 Sep;20(3):260-8. doi: 10.1111/jns.12122. Review.
 92. Vanhoutte EK, Draak TH, Gorson KC, van Nes SI, Hoeijmakers JG, Van der Pol WL, Notermans NC, Lewis RA, Nobile-Orazio E, Léger JM, Van den Bergh PY, Lauria G, Bril V, Katzberg H, Lunn MP, Pouget J, van der Kooi AJ, Hahn AF, van Doorn PA, Cornblath DR, van den Berg LH, Faber CG, Merkies IS; PeriNomS Study Group. Impairment measures versus inflammatory RODS in GBS and CIDP: a responsiveness comparison. **J Peripher Nerv Syst.** 2015 Sep;20(3):289-95. doi: 10.1111/jns.12118.
 93. Draak TH, Vanhoutte EK, van Nes SI, Gorson KC, Van der Pol WL, Notermans NC, Nobile-Orazio E, Lewis RA, Léger JM, Van den Bergh PY, Lauria G, Bril V, Katzberg H, Lunn MP, Pouget J, van der Kooi AJ, Hahn AF, van den Berg LH, van Doorn PA, Cornblath DR, Faber CG, Merkies IS; PeriNomS Study Group. Comparing the NIS vs. MRC and INCAT sensory scale through Rasch analyses. **J Peripher Nerv Syst.** 2015 Sep;20(3):277-88. doi: 10.1111/jns.12127.
 94. Huijbers MG, Querol LA, Niks EH, Plomp JJ, van der Maarel SM, Graus F, Dalmau J, Illa I, Verschuuren JJ. The expanding field of IgG4-mediated neurological autoimmune disorders. **Eur J Neurol.** 2015 Aug;22(8):1151-61. doi: 10.1111/ene.12758. Epub 2015 May 29.
 95. Ramos-Fransi A, Rojas-García R, Segovia S, Márquez-Infante C, Pardo J, Coll-Cantí J, Jericó I, Illa I; Myasthenia NMD-ES Study Group. Myasthenia gravis: descriptive analysis of life-threatening events in a recent nationwide registry. **Eur J Neurol.** 2015 Apr 6. doi: 10.1111/ene.12703. [Epub ahead of print]

96. Gutiérrez-Rivas E, Illa I, Pascual-Pascual SI, Pérez-López J, Vílchez-Padilla JJ, Bautista-Lorite J, Barrot E, López de Munain A.[Guidelines for monitoring late-onset Pompe disease]. **Rev Neurol.** 2015 Apr 1;60(7):321-8.
97. Aragonès JM, Altimiras J, Roura P, Alonso F, Bufill E, Munmany A, Alfonso S, Illa I. Prevalence of myasthenia gravis in the Catalan county of Osona. *Neurologia.* 2014 Nov 11. pii: S0213-4853(14)00203-5. doi: 10.1016/j.nrl.2014.09.007. [Epub ahead of print]
98. Gallardo E, Martínez-Hernández E, Titulaer MJ, Huijbers MG, Martínez MA, Ramos A, Querol L, Díaz-Manera J, Rojas-García R, Hayworth CR, Verschuuren JJ, Balice-Gordon R, Dalmau J, Illa I. Contactin autoantibodies in myasthenia gravis. **Autoimmun Rev.** 2014 Oct;13(10):1003-7. doi: 10.1016/j.autrev.2014.08.039. Epub 2014 Sep 3.
99. Aragonès JM, Roura-Poch P, Hernández-Ocampo EM, Alonso F, Pont-Lluelles M, Xandri I, Bolívar I, Illa I. Myasthenia gravis: a disease of the very old. **J Am Geriatr Soc.** 2014 Jan;62(1):196-7.
100. Díaz-Manera J, Querol L, Alejaldre A, Rojas-García R, Ramos-Fransi A, Gallardo E, Illa I. Severe exacerbation of Andersen-Tawil syndrome secondary to thyrotoxicosis. **J Hum Genet.** 2014 Aug;59(8):465-6. doi: 10.1038/jhg.2014.43. Epub 2014 May 22.
101. Gallardo E, Ankala A, Núñez-Álvarez Y, Hegde M, Diaz-Manera J, Luna ND, Pastoret A, Suelves M, Illa I. Genetic and epigenetic determinants of low dysferlin expression in monocytes. **Hum Mutat.** 2014 Aug;35(8):990-7. doi: 10.1002/humu.22591. Epub 2014 Jun 24.
102. Díaz-Manera J, Alejaldre A, Llauger J, Mirabet S, Rojas-García R, Ramos-Fransi A, Gallardo E, Illa I. Cranial, axial and proximal myopathy and hypertrophic cardiomyopathy caused by a mutation in the globular head region of the MYH7 gene. **Eur J Neurol.** 2014 Jun;21(6):e51-2. doi: 10.1111/ene.12416.
103. Nogales-Gadea G, Ramos-Fransi A, Suárez-Calvet X, Navas M, Rojas-García R, Mosquera JL, Díaz-Manera J, Querol L, Gallardo E, Illa I. Analysis of serum miRNA profiles of myasthenia gravis patients. **PLoS One.** 2014 Mar 17;9(3):e91927. doi: 10.1371/journal.pone.0091927. eCollection 2014.
104. Suárez-Calvet X, Gallardo E, Nogales-Gadea G, Querol L, Navas M, Díaz-Manera J, Rojas-García R, Illa I. Altered RIG-I/DDX58-mediated innate immunity in dermatomyositis. **J Pathol.** 2014 Jul;233(3):258-68. doi: 10.1002/path.4346. Epub 2014 Apr 29.
105. Querol L, Nogales-Gadea G, Rojas-García R, Diaz-Manera J, Pardo J, Ortega-Moreno A, Sedano MJ, Gallardo E, Berciano J, Blesa R, Dalmau J, Illa I. Neurofascin IgG4 antibodies in CIDP associate with disabling tremor and poor response to IVIg. **Neurology.** 2014 Mar 11;82(10):879-86. doi: 10.1212/WNL.0000000000000205. Epub 2014 Feb 12.
106. Labasque M, Hivert B, Nogales-Gadea G, Querol L, Illa I, Faivre-Sarrailh C. Specific contactin N-glycans are implicated in neurofascin binding and autoimmune targeting in peripheral neuropathies. **J Biol Chem.** 2014 Mar 14;289(11):7907-18. doi: 10.1074/jbc.M113.528489. Epub 2014 Feb 4.
107. Evilä A, Vihola A, Sarparanta J, Raheem O, Palmio J, Sandell S, Eymard B, Illa I, Rojas-García R, Hankiewicz K, Negrão L, Löppönen T, Nokelainen P, Kärppä M, Penttilä S, Screen M, Suominen T, Richard I, Hackman P, Udd B. Atypical phenotypes in titinopathies explained by second titin mutations. **Ann Neurol.** 2014 Feb;75(2):230-40. doi: 10.1002/ana.24102. Epub 2014 Feb 24.
108. Querol L, Illa I. Myasthenia gravis and the neuromuscular junction. **Curr Opin Neurol.** 2013 Oct;26(5):459-65. doi: 10.1097/WCO.0b013e328364c079. Review.
109. Flix B, Suárez-Calvet X, Díaz-Manera J, Santos-Nogueira E, Mancuso R, Barquinero J, Navas M, Navarro X, Illa I, Gallardo E. Bone marrow transplantation in dysferlin-deficient mice results in a mild functional improvement. **Stem Cells Dev.** 2013 Nov 1;22(21):2885-94. doi:10.1089/scd.2013.0049. Epub 2013 Jul 26.
110. Rojas-García R, Gallardo E, Illa I. Paraproteinemic neuropathies **Presse Med.** 2013 Jun;42(6 Pt 2):e225-34. doi: 10.1016/j.lpm.2013.02.329. Epub 2013 Apr 22. Review.
111. Sarkozy A, Hicks D, Hudson J, Laval SH, Barresi R, Hilton-Jones D, Deschauer M, Harris E, Rufibach L, Hwang E, Bashir R, Walter MC, Krause S, van den Bergh P, Illa I, Péniisson-

- Besnier I, De Waele L, Turnbull D, Guglieri M, Schrank B, Schooser B, Seeger J, Schreiber H, Gläser D, Eagle M, Bailey G, Walters R, Longman C, Norwood F, Winer J, Muntoni F, Hanna M, Roberts M, Bindoff LA, Brierley C, Cooper RG, Cottrell DA, Davies NP, Gibson A, Gorman GS, Hammans S, Jackson AP, Khan A, Lane R, McConville J, McEntagart M, Al-Memmar A, Nixon J, Panicker J, Parton M, Petty R, Price CJ, Rakowicz W, Ray P, Schapira AH, Swingle R, Turner C, Wagner KR, Maddison P, Shaw PJ, Straub V, Bushby K, Lochmüller H. ANO5 gene analysis in a large cohort of patients with anoctaminopathy: confirmation of male prevalence and high occurrence of the common exon 5 gene mutation. **Hum Mutat.** 2013 Aug;34(8):1111-8. doi: 10.1002/humu.22342. Epub 2013 Jun 12.
112. de Morrée A, Flix B, Bagaric I, Wang J, van den Boogaard M, Grand Moursel L, Frants RR, Illa I, Gallardo E, Toes R, van der Maarel SM. Dysferlin regulates cell adhesion in human monocytes **J Biol Chem.** 2013 May 17;288(20):14147-57. doi: 10.1074/jbc.M112.448589. Epub 2013 Apr 4.
113. Querol L, Rojas-García R, Casasnovas C, Sedano MJ, Muñoz-Blanco JL, Alberti MA, Paradas C, Sevilla T, Pardo J, Capablo JL, Sivera R, Guerrero A, Gutierrez-Rivas E, Illa I. Long-term outcome in chronic inflammatory demyelinating polyneuropathy patients treated with intravenous immunoglobulin: a retrospective study. **Muscle Nerve.** 2013 Dec;48(6):870-6. doi: 10.1002/mus.23843. Epub 2013 Sep 11.
114. Querol L, Nogales-Gadea G, Rojas-García R, Martínez-Hernández E, Díaz-Manera J, Suárez-Calvet X, Navas M, Araque J, Gallardo E, Illa I. Antibodies to contactin-1 in chronic inflammatory demyelinating polyneuropathy. **Ann Neurol.** 2013 Mar;73(3):370-80. doi: 10.1002/ana.23794. Epub 2012 Dec 31.
115. Alejaldre A, Díaz-Manera J, Ravaglia S, Tibaldi EC, D'Amore F, Morís G, Muelas N, Vilchez JJ, García-Medina A, Usón M, Martínez García FA, Illa I, Pichiecchio A. Trunk muscle involvement in late-onset Pompe disease: study of thirty patients. **Neuromuscul Disord.** 2012 Oct 1;22 Suppl 2:S148-54. doi: 10.1016/j.nmd.2012.05.011.
116. Luna ND, Díaz-Manera J, Paradas C, Iturriaga C, Rojas-García R, Araque J, Genebriera M, Gich I, Illa I, Gallardo E. 1 α ,25(OH)(2)-Vitamin D3 Increases Dysferlin Expression in vitro and in a Human Clinical Trial. **Mol Ther.** 2012 Aug 21. doi: 10.1038/mt.2012.156. [Epub ahead of print.]
117. Díaz-Manera J, Gallardo E, de Luna N, Navas M, Soria L, Garibaldi M, Rojas-García R, Tonlorenzi R, Cossu G, Illa I. The increase of pericyte population in human neuromuscular disorders supports their role in muscle regeneration in vivo. **J Pathol.** 2012 Jul 30. doi: 10.1002/path.4083. [Epub ahead of print].
118. Díaz-Manera J, Rojas García R, Illa I. Treatment strategies for myasthenia gravis: an update. **Expert Opin Pharmacother.** 2012 Sep;13(13):1873-83. Epub 2012 Jul 9.
119. Rojas-García R, Querol L, Gallardo E, De Luna Salva N, Juárez C, Garces M, Fages E, Casasnovas C, Illa I. Clinical and serological features of acute sensory ataxic neuropathy with antiganglioside antibodies. **J Peripher Nerv Syst.** 2012 Jun;17(2):158-68.
120. Vanhoutte EK, Faber CG, Merkies IS; PeriNomS Study Group. MRC sum-score in the ICU: good reliability does not necessarily reflect "true reliability". **Muscle Nerve.** 2012 May;45(5):767-8.
121. Barba-Romero MA, Barrot E, Bautista-Lorite J, Gutierrez-Rivas E, Illa I, Jimenez LM, Ley-Martos M, Lopez de Munain A, Pardo J, Pascual-Pascual SI, Perez-Lopez J, Solera J, Vilchez-Padilla JJ. Clinical guidelines for late-onset Pompe disease. **Rev Neurol.** 2012 Apr 16;54(8):497-507.
122. Díaz-Manera J, Martínez-Hernández E, Querol L, Klooster R, Rojas-García R, Suárez-Calvet X, Muñoz-Blanco JL, Mazia C, Straasheijm KR, Gallardo E, Juárez C, Verschuuren JJ, Illa I. Long-lasting treatment effect of rituximab in MuSK myasthenia. **Neurology.** 2012 Jan 17;78(3):189-93.
123. Vanhoutte EK, Faber CG, van Nes SI, Jacobs BC, van Doorn PA, van Koningsveld R, Cornblath DR, van der Kooij AJ, Cats EA, van den Berg LH, Notermans NC, van der Pol WL, Hermans MC, van der Beek NA, Gorson KC, Eurlings M, Engelsman J, Boot H, Meijer RJ, Lauria G, Tennant A, Merkies IS; PeriNomS Study Group. Modifying the Medical Research Council grading system through Rasch analyses. **Brain.** 2012 May;135(Pt 5):1639-49.

124. Berciano J, Sevilla T, Casanovas C, Sivera R, Vílchez JJ, Infante J, Ramón C, Pelayo-Negro AL, Illa I; Guidelines for molecular diagnosis of Charcot-Marie-Tooth disease. Programa 3 (Enfermedades Neuromusculares) del Centro de Investigación Biomédica en Red de Enfermedades Neurodegenerativas (CIBERNED) del Instituto de Salud Carlos III. **Neurologia**. 2012 Apr;27(3):169-78.
125. Gallardo E, de Luna N, Diaz-Manera J, Rojas-García R, Gonzalez-Quereda L, Flix B, de Morrée A, van der Maarel S, Illa I. Comparison of dysferlin expression in human skeletal muscle with that in monocytes for the diagnosis of dysferlin myopathy. **PLoS One**. 2011;6(12):e29061.
126. Resmini E, Morte B, Sorianello E, Gallardo E, de Luna N, Illa I, Zorzano A, Bernal J, Webb SM. Identification of novel GH-regulated genes in C2C12 cells. **Horm Metab Res**. 2011 Dec;43(13):919-30.
127. Díaz-Manera J, Querol L, Clarimón J, Yagüe S, Illa I. Unique post-exercise electrophysiological test results in a new Andersen-Tawil syndrome mutation. **Clin Neurophysiol**. 2011 Dec;122(12):2537-9.
128. Krahn M, Illa I, Lévy N, Bushby K. 172nd ENMC International Workshop: dysferlinopathies 29-31 January 2010, Naarden, The Netherlands. **Neuromuscul Disord**. 2011 Jul;21(7):503-12
129. Peters MJ, van Nes SI, Vanhoutte EK, Bakkens M, van Doorn PA, Merkies IS, Faber CG; PeriNomS Study group. Revised normative values for grip strength with the Jamar dynamometer. **J Peripher Nerv Syst**. 2011 Mar;16(1):47-50.
130. Gallardo E, Saenz A, Illa I. Limb-girdle muscular dystrophy 2A. **Handb Clin Neurol**. 2011;101:97-110.
131. Lancaster E, Huijbers MG, Bar V, Boronat A, Wong A, Martinez-Hernandez E, Wilson C, Jacobs D, Lai M, Walker RW, Graus F, Bataller L, Illa I, Markx S, Strauss KA, Peles E, Scherer SS, Dalmau J. Investigations of caspr2, an autoantigen of encephalitis and neuromyotonia. **Ann Neurol**. 2011 Feb;69(2):303-11.
132. Halter J, Schüpbach WM, Casali C, Elhasid R, Fay K, Hammans S, Illa I, Kappeler L, Krähenbühl S, Lehmann T, Mandel H, Marti R, Mattle H, Orchard K, Savage D, Sue CM, Valcarcel D, Gratwohl A, Hirano M. Allogeneic hematopoietic SCT as treatment option for patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): a consensus conference proposal for a standardized approach. **Bone Marrow Transplant**. 2011 Mar;46(3):330-7.
133. Pinós T, Marotta M, Gallardo E, Illa I, Díaz-Manera J, Gonzalez-Vioque E, García-Arumí E, Andreu AL, Martí R. A novel mutation in the mitochondrial tRNA(Ala) gene (m.5636T>C) in a patient with progressive external ophthalmoplegia. **Mitochondrion**. 2011 Jan;11(1):228-33.
134. Díaz-Manera J, Touvier T, Dellavalle A, Tonlorenzi R, Tedesco FS, Messina G, Meregalli M, Navarro C, Perani L, Bonfanti C, Illa I, Torrente Y, Cossu G. Partial dysferlin reconstitution by adult murine mesoangioblasts is sufficient for full functional recovery in a murine model of dysferlinopathy. **Cell Death Dis**. 2010 Aug 5;1:e61.
135. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society guideline on management of multifocal motor neuropathy. Report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society--first revision. **J Peripher Nerv Syst**. 2010 Dec;15(4):295-301.
136. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society Guideline on management of paraproteinemic demyelinating neuropathies. Report of a Joint Task Force of the European Federation of Neurological Societies and the Peripheral Nerve Society--first revision. **J Peripher Nerv Syst**. 2010 Sep;15(3):185-95.
137. Hernández S, Texidó L, Calderó J, Ciutat D, Piedrafita L, Casanovas A, Blasi J, Solsona C, Povedano M, Rojas R, Illa I, Caress J, Prevet D, Oppenheim RW, Milligan C, Esquerda JE. Increased intramuscular nerve branching and inhibition of programmed cell death of chick embryo motoneurons by immunoglobulins from patients with motoneuron disease. **J Neuroimmunol**. 2010 Dec 15;229(1-2):157-68.

138. Paradas C, Llauger J, Diaz-Manera J, Rojas-García R, De Luna N, Iturriaga C, Márquez C, Usón M, Hankiewicz K, Gallardo E, Illa I. Redefining dysferlinopathy phenotypes based on clinical findings and muscle imaging studies. **Neurology**. 2010 Jul 27;75(4):316-23.
139. De Luna N, Gallardo E, Sonnet C, Chazaud B, Dominguez-Perles R, Suarez-Calvet X, Gherardi RK, Illa I. Role of thrombospondin 1 in macrophage inflammation in dysferlin myopathy. **J Neuropathol Exp Neurol**. 2010 Jun;69(6):643-53.
140. Van den Bergh PY, Hadden RD, Bouche P, Cornblath DR, Hahn A, Illa I, Koski CL, Léger JM, Nobile-Orazio E, Pollard J, Sommer C, van Doorn PA, van Schaik IN; European Federation of Neurological Societies; Peripheral Nerve Society. European Federation of Neurological Societies/Peripheral Nerve Society guideline on management of chronic inflammatory demyelinating polyradiculoneuropathy: report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society - first revision. **Eur J Neurol**. 2010 Mar;17(3):356-63.
141. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society Guideline on management of chronic inflammatory demyelinating polyradiculoneuropathy: report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society--First Revision. **J Peripher Nerv Syst**. 2010 Mar;15(1):1-9. Erratum in: *J Peripher Nerv Syst*. 2010 Dec;15(4):373.
142. Skeie GO, Apostolski S, Evoli A, Gilhus NE, Illa I, Harms L, Hilton-Jones D, Melms A, Verschuuren J, Horge HW; European Federation of Neurological Societies. Guidelines for treatment of autoimmune neuromuscular transmission disorders. **Eur J Neurol**. 2010 Jul;17(7):893-902.
143. Rojas-García R, Gallardo E, De Luna N, Juárez C, Martínez-Hernández E, Carvajal A, Casasnovas C, Fages E, Davila-González P, Illa I. Bulbar involvement in patients with antianglioside antibodies against NeuNAc(alpha2-3)Gal. **J Neurol Neurosurg Psychiatry**. 2010 Jun;81(6):623-8.
144. Díaz-Manera J, Rojas-García R, Illa I. Treatment strategies for myasthenia gravis. **Expert Opin Pharmacother**. 2009 Jun;10(8):1329-42.
145. Berciano J, García A, Villagrà NT, González F, Ramón C, Illa I, Berciano MT, Lafarga M. Severe Guillain-Barré syndrome: sorting out the pathological hallmark in an electrophysiological axonal case. **J Peripher Nerv Syst**. 2009 Mar;14(1):54-63.
146. De la Torre C, Illa I, Faulkner G, Soria L, Robles-Cedeño R, Dominguez-Perles R, De Luna N, Gallardo E. Proteomics identification of differentially expressed proteins in the muscle of dysferlin myopathy patients. **Proteomics Clin Appl**. 2009 Apr;3(4):486-97.
147. Response to methotrexate in a chronic inflammatory demyelinating polyradiculoneuropathy patient. Díaz-Manera J, Rojas-García R, Gallardo E, Illa I **Muscle Nerve**. 2009 Mar;39(3):386-8.
148. Paradas C, González-Quereda L, De Luna N, Gallardo E, García-Consuegra I, Gómez H, Cabello A, Illa I, Gallano P. A new phenotype of dysferlinopathy with congenital onset. **Neuromuscul Disord**. 2009 Jan;19(1):21-5.
149. Sáenz A, Azpitarte M, Armañanzas R, Leturcq F, Alzualde A, Inza I, García-Bragado F, De la Herran G, Corcuera J, Cabello A, Navarro C, De la Torre C, Gallardo E, Illa I, López de Munain A. Gene expression profiling in limb-girdle muscular dystrophy 2A. **PLoS One**. 2008;3(11):e3750. doi: 10.1371/journal.pone.0003750. Epub 2008 Nov 18.
150. Hackman P, Marchand S, Sarparanta J, Vihola A, Péniisson-Besnier I, Eymard B, Pardal-Fernández JM, Hammouda el-H, Richard I, Illa I, Udd B. Truncating mutations in C-terminal titin may cause more severe tibial muscular dystrophy (TMD). **Neuromuscul Disord**. 2008 Dec;18(12):922-8.
151. Guallar JP, Rojas-García R, García-Arumi E, Domingo JC, Gallardo E, Andreu AL, Domingo P, Illa I, Giralt M, Villarroya F. Impaired expression of mitochondrial and adipogenic genes in

- adipose tissue from a patient with acquired partial lipodystrophy (Barraquer-Simons syndrome): a case report. **J Med Case Rep.** 2008 Aug 27;2:284.
152. Illa I, Diaz-Manera J, Rojas-Garcia R, Pradas J, Rey A, Blesa R, Juarez C, Gallardo E. Sustained response to Rituximab in anti-AChR and anti-MuSK positive Myasthenia Gravis patients. **J Neuroimmunol.** 2008 Sep 15;201-202:90-4.
153. Blázquez L, Azpitarte M, Sáenz A, Goicoechea M, Otaegui D, Ferrer X, Illa I, Gutierrez-Rivas E, Vilchez JJ, López de Munain A. Characterization of novel CAPN3 isoforms in white blood cells: an alternative approach for limb-girdle muscular dystrophy 2A diagnosis. **Neurogenetics.** 2008 Jul;9(3):173-82.
154. Rojas-Garcia R, Gallardo E, De La Torre C, Sanvito L, Illa I. Chronic sensorimotor polyradiculopathy with antibodies to P2: an electrophysiological and immunoproteomic analysis. **Muscle Nerve.** 2008 Jul;38(1):933-8.
155. Rojas-Garcia R, Gallardo E, Povedano M, de Luna N, Bruna J, Juarez C, Diaz-Manera J, Martinez-Matos JA, Illa I. Antibodies against disialosyl and terminal NeuNAc(alpha2-3)Gal ganglioside epitopes in acute relapsing sensory ataxic neuropathy. **J Neurol.** 2008 May;255(5):764-6
156. Berciano J, Gallardo E, Domínguez-Perles R, Gallardo E, García A, García-Barredo R, Combarros O, Infante J, Illa I. Autosomal-dominant distal myopathy with a myotilin S55F mutation: sorting out the phenotype. **J Neurol Neurosurg Psychiatry.** 2008 Feb;79(2):205-8.