**Curriculum Vitae**

Name and surname: Jordi Diaz Manera

Date: November the 1st, 2019

**1. Personal data**

| Name and surname | National ID Number |
|---|---|
| Jordi Diaz Manera | 43090431T |

| Nationality | Sex |
|---|---|
| Spanish<br>Born in: August 27th 1976 | ☒ Man ☐ Woman |

| Direction |
|---|
| C/ Napols 215; 6e 4ª, 08013 BARCELONA |

**2. Professional situation at the moment**

| Insitution | Place of work |
|---|---|
| Hospital de la Santa Creu i Sant Pau | Hospital Santa Creu i Sant Pau |

| Department | Direction |
|---|---|
| Neurology | Carrer Sant Antoni Mª Claret 167 |

| Postal code | City | Province |
|---|---|---|
| 08025 | Barcelona | Barcelona |

| Professional Category | Starting date |
|---|---|
| Senior Associate Member of the department | 2006 |

| Administrative situation |
|---|
| X Full time permanent ☐ Part Time permanent ☐ Grant. |

**3. Scientific identification**

| | | |
|---|---|---|
| x ORCID ID: 0000-0003-2941-7988 | ☐ Scopus ID: | ☐ |
| Researcher ID: | | |

**4. Academic degree**

| Studies | University | Date |
|---|---|---|
| 1. Degree in Medicine and Surgery | 1. Universitat Rovira i Virgili | 1. 2000 |
| **Postdegree studies** | **University** | **Date** |
| 1. PhD in Medicine | 1. Universitat Autònoma Barcelona | 1. 2013 |

**5. Other Scientific or professional activities**

| Activity | Institution | Period |
|---|---|---|
| 1. Directive Board Member | 1. Catalan Society Neurology | 1. 2017-2018 |
| 2. Director of the Neuromuscular Disorders Study Group | 2. Spanish Society Neurology | 2. 2012-2014 |
| 3. Young Fellowship Grant | 3. European Federation of Neurological Societies | 3. 2008-2009 |
| 4. Neuromuscular disorders Grant | 4. Spanish Society of Neurology | 4. 2005-2006 |

**6. Languages (B: basic, A= advance, P = proficiency)**

| Language | Speaking | Wirting | Reading |
|---|---|---|---|
| 1. Catalan | 1. P | 1. P | 1. P |
| 2. Spanish | 2. P | 2. P | 2. P |
| 3. English | 3. A | 3. A | 3. A |
| 4.Italian | 4.A | 4.A | 4.A |

**7. Doctoral Thesis**

**Name:** New diagnostic and terapeuthic aspects of dysferlinopathy

**Director:** Isabel Illa Sendra

**University:** Universitat Autónoma de Barcelona

**Department:** Medicine

**Qualification:** Cum Laude, Selected as Best Thesis of the Yea

**European Doctoral Thesis:** ☒ **Sí** ☐ **No**

**Date of the defense (dd/mm/aaaa):** May the 10th 2013

**Date of issue of the title (dd/mm/aa):** July the 29th 2013

**Summary of my scientific trajectory**

I studied medicine at the Rovira i Virgili University in Tarragona from 1994 to 2000. Being a small university, I soon came into contact with professors who had a research activity. In my case, I collaborated with the Histology Unit, a unit that had a series of ongoing studies on the development of neuromuscular union in murine models.
I obtained a National Grant for students that wished to collaorate with University Departments. I developed this grant in the Histology Unit. This stay awakened my curiosity for neurology, neuromuscular diseases and research.
In 2001, I started my neurology residency at the Hospital de la Santa Creu i Sant Pau in Barcelona (HSCSP). From the first months of my residency, it was clear to me that I wanted to share my time between basic clinical research and care.
In fact, in the third year of residency I published my first scientific article reviewing the clinical characteristics of a group of patients with myasthenia Musk. This work was used to achieve research sufficiency and have access to Postgraduate studies (needed to perform a PhD in Medicine).
After finishing my neurology residency in 2005, I had the opportunity to start a training scholarship at the Neuromuscular Unit of the HSCSP under the direction of Prof. Illa. In 2006 I was hired by the Deparment of Neurology of the HSCSP as an associate, dedicated to neuromuscular disorders.
In the first formative years I shared my healthcare tasks with research, both clinical and basic. During these years I was very interested in Myasthenia Gravis as a model of autoimmune disease, and in fact my publications of that time are related to this disease.
But in 2008 I had the opportunity to carry out a pre-doctoral stay in Italy, at the Stem Cell Research Institute of the Ospedalle San Rafaelle in Milan in the laboratory of Dr. Cossu. This stay was funded by a grant from the Spanish Ministry of Health and by a Young Fellowship Grant from the European Federation of Neurological Societies. In Italy, I developed a cell therapy project in a murine model of muscular dystrophy due to dysferline deficit.
This disease has been the focus of my doctoral thesis research, which I defended in 2013 and which obtained the qualification of cum laude an was selectes as one of the Best Thesis of the Year by the University. My stay in Italy opened my eyes to a world of basic research, I learned to pose and solve problems with the tools of the laboratory. In 2010 I came back from Italy to Barcelona, and since that moment I was in charge of the diagnosis and follow-up of patients with hereditary and acquired myopathies at our Unit.
My clinical research has been focused on the application of magnetic resonance in the diagnosis and follow-up of patients with muscle disorders. This has allowed me to publish numerous papers in international journals and collaborate with scientific groups around the world. As a result of this effort, I have been part of the Action-COST called MYO-MRI aimed to disseminate the use of MRI among clinicians dedicated to neuromuscular diseases and coordinating the last two years of the COST some of the group meetings. This participation has opened the door to new studies in which our Unit is participating and I have been able to take our centre to the forefront of research in this aspect.
Basic research is the other pillar of my research routine. For years we have been interested in the physiopathology and the development of new treatments for muscular dystrophies. The work carried out in this field has allowed us to find a new indication for an antifibrotic drug, with a European patent shared with Boehringer-Ingelheim, which is the pharmaceutical company that has developed the drug and began a series of research projects grantes with both private and public fundings
Finally, the third aspect of my research is Pompe's disease, a metabolic myopathy of which we have done a series of basic research and radiological work that have allowed us to publish articles in first quartile international journals.
I have been coordinator of the neuromuscular diseases study group of the Spanish Society of Neurology from 2012 to 2014 and member of the board of the Catalan Society of Neurology from 2017 to 2019. I am an associate professor at the Autonomous University of Barcelona. I have directed 3 doctoral Thesis that have been defended already, and I am currently directing 5 other thesis focused on muscled disorders

**Scientific projects (public funding, competitive projects)**

As a principal investigator:

1. Name of the project: “Effect of nintedanib in muscle fibrosis in a mouse model of sarcoglycanopathy and in the function of human and mouse FAP cells”
   Funding Insitution: Association Francaise contre les Myopathies
   Grant number: AFM Trampolin Grant 22525
   Funding obtained: 49.201,00 €
   Duration of the grant: 2020
   PI: Jordi Diaz Manera

2. Name of the project: “Advances in imaging techniques and therapy in muscular dystrophies: evaluation and treatment of fibrosis”
   Funding Insitution: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health.
   Grant number: FIS 18/1525
   Funding obtained: 129.470,00 €
   Duration of the grant: 2019-2021
   PI: Jordi Diaz Manera

3. Name of the project: “PDGF as a new biomarker and terapeuthic target in patients with muscular dystrophies”
   Funding Insitution: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health.
   Grant number: FIS 15/1822
   Funding obtained: 92.565,00 €
   Duration of the grant: 2016-2018
   PI: Jordi Diaz Manera

4. Name of the project: “Nintedanib as a new antifibrotic drug for Duchenne muscular dystrophy”
   Funding Insitution: Duchenne Parent Project Spain
   Grant number: -
   Funding obtained: 50.000,00 €
   Duration of the grant: 2014-2016
   IP: Jordi Diaz Manera

As a coinvestigator:

5. Name of the project: “Functional study of dysferlinopathy: therapeutic implications”.
   Funding entity: Fondos de Investigación Sanitaria del Instituto de Salud Carlos III, Spanish Ministry of Health.
   Grant number: FIS 06/0455
   Funding otained: 131.164,00 €
   Funding period: 2007-09
   IP: Eduard Gallardo Vigo

6. Name of the project: Research on neuromuscular diseases. Consolidated Research Group, Generalitat de Catalunya
   Funding entity: Departament de Salut, Generalitat de Catalunya
   Grant number: SGR1004
   Funding otained: 43.680,00 €
   Funding period: 2010-2014
   IP: Eduard Gallardo Vigo

7. Name of the project: Research on neuromuscular diseases. Consolidated Research Group, Generalitat de Catalunya
   Funding entity: Departament de Salut, Generalitat de Catalunya
   Grant number: SGR1004
   Funding obtained: 30.000,00 €
   Funding period: 2014-2018
   IP: Eduard Gallardo Vigo

8. Name of the project: “Study of the pathophysiology of muscular dystrophies with mutations in the gene DYSF by means of the analysis of the interactome and the signaling pathways in wich dysferlin is involved”
   Funding entity: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health.
   Grant number: FIS 09/1944
   Funding obtained: 140.960,00 €
   Funding period: 2010-2012
   IP: Eduard Gallardo Vigo

9. Name of the project:“Development of new diagnostic assays for dysferlinopathy using peripheral blood monocytes”.
   Entitat Finançadora: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health
   Grant number: ETES 08/ 90622
   Funding obtained: 111.920,00 €
   Funding period: 2008-2010
   IP: Isabel Illa Sendra

10. Name of the project:“Treatment of symptomatic carriers of dysferlin myopathy with vitamin D”.
    Funding entity: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health.
    Grant number: FIS EC07/ 90601
    Funding obtained: 38.700,00 €
    Funding duration: 2008-2010
    IP: Isabel Illa Sendra

11. Name of the Projecte: “Search for plasma biomarkers in muscular dystrophies”
    Funding entity: Fondos de Investigación Sanitaria del Insituto de Salud Carlos III, Spanish Ministry of Health.
    Grant number: FIS 12/02291
    Funding obtained: 109.500,00 €
    Funding duration: 2013-15
    IP: Eduard Gallardo Vigo

**Scientific projects (private funding, non competitive projects)**

As a principal Investigator:

1. Project Name: “New MRI tools for the identification of glycogen in patients with Pompe disease”
   Funding entity: Sanofi-Genzyme
   Funding otained: 119.000,00 €
   Funding period: 2020-2022
   IP: Jordi Diaz Manera – Glenn Walter (University of Florida)

2. Project Name: “New therapeutic tools for Muscular Dystrophies”
   Funding entity: Fundacion Isabel Gemio
   Funding obtained: 150.000,00 €
   Funding period: 2020-2022
   IP: Jordi Diaz Manera – Eduard Gallardo

3. Project Name:“Spanish Registry of Pompe disease”.
   Funding entity: Sanofi-Genzyme Spain
   Funding obtained: 36.000,00 €
   Funding period: 2020
   IP: Jordi Diaz Manera

4. Project Name:“Nintedanib as a new treatment for a sarcoglycanopathy and dysferlinopathy murine model”.
   Funding entity: Boehringer Ingelheim
   Funding obtained: 60.000,00 €
   Funding period: 2019-2020
   IP: Jordi Diaz Manera

5. Project name:“Nintedanib in Duchenne Muscle Dystrophy”.
   Funding entity: BOEHRINGER INGELHEIM
   Funding obtained: 32.000,00 €
   Funding period: 2015-2016
   IP: Jordi Diaz Manera

6. Project name: “Use of Muscle MRI as a Biomarker tool in Pompe disease”.
   Funding entity: Sanofi-Genzyme
   Funding obtained: 200.000 Euros
   Duration: 2013-2018
   IP: Jordi Diaz Manera

7. Project name:“Clinical Outcome Study for Dysferlinopathy”.
   Funding entity: Jain Foundation (Seattle, USA)
   Funding obtained: 60.000 euros
   Duration: 2011-2016
   IP: Jordi Diaz Manera

As a coinvestigator:

8. Name of the Project:“Combined cell transplantation as a therapeutic approach for dysferlinopathies and transversal study to evaluate the frequency of carriers of dysferlinopathy in Caucasian population using a monocyte’s test to detect dysferlin expression”.
   Funding entity: Jain Foundation (Seattle, USA)
   Funding obtained: 84.000,00
   Duration: 2012-2014
   IP: Eduard Gallardo Vigo

9. Name of the project: “Development of new therapies for muscular dystrophies: mesoangiolasts and bone marrow transplantion for a murine model of muscular dystrophy”.
   Funding entity: Fundación Isabel Gemio
   Funding obtained: 400.000,00
   Duration: 2012-2014
   IP: Eduard Gallardo Vigo – Jordi Diaz Manera – Isabel Illa

10. Títol del Projecte: “Serum biomarkers study of Pompe disease”
    Fuding entity: Sanofi-Genzyme
    Funding obtained: 134.928,00
    Duration: 2016-2019
    IP: Eduard Gallardo Vigo

**Clinical trials (private funding, non competitive projects)**

1. Name of the trial: An International Phase III multicentric, double blind clinical trial analysing the effectiveness of Tadalafil in Duchenne Muscular dystrophy.
   Sponsor: Lilly
   Duration: 2015-2016
   Principal Investigator: Isabel Illa
   Role: Subinvestigator.

2. Name of the trial: An international Phase III multicentric clinical trial assessing the effectiveness of Eculizumab in Myasthenia Gravis.
   Sponsor: Alexion
   Duration: 2015-17
   Principal Investigator: Isabel Illa
   Role: Sub-investigator

3. Name of the trial: A prospective, noninterventional, observational study of late-onset pompe disease (BMN 701-901 Study)
   Sponsor: Biomarin
   Durada: 2015
   Princial Investigator: Jordi Díaz Manera

4. Name of the trial: A phase Ib, open label study to evaluate the safety, tolerability, pharmacokinetics and pharmacodynamics of multiple ascending oral doses of rimeporide in patients with duchenne muscular dystrophy (ESPERARE_RIM_001)
   Sponsor: ESPERARE Fundation
   Durada: 2016
   Princial Investigator: Jordi Díaz Manera

5. Name of the trial: A randomized, multicenter, multinational, double-blinded study of neogaa and alglucosidade alfa in approximately 96 treatment na√Øve late-onset pompe disease patients aged 3 years and older.
   (EFC14028 COMET)
   Sponsor: Sanofi-Genzyme
   Duration: 2016-
   Principal investigator: Jordi Díaz Manera

6. Name of the trial: A double-blind, placebo-controlled, multicenter study with an open-label extension to evaluate the efficacy and safety of srp-4045 and arp-4053 in patients with duchenne muscular dystrophy (4045-301 ESSENCE)
   Sponsor: Sarepta
   Duration: 2017-2019
   Principal investigator: Jordi Diaz Manera

7. Name of the trial: A double-blind, placebo-controlled, multicenter study with an open-label extension to evaluate the efficacy and safety of srp-4045 and arp-4053 in patients with duchenne muscular dystrophy (4045-302 ESSENCE)
   Sponsor: Sarepta
   Duration: 2019-
   Principal investigator: Jordi Diaz Manera

8. Name of the trial: Aphase 2 randomized, double blind, placebo controlled study of ace-083 in patients with facioscapulohemeral muscular dystrophy (ACE8302 ACCELERON)
   Sponsor: Acceleron
   Duration: 2018-
   Principal investigator: Jordi Diaz Manera

9. Name of the trial: A randomized, double blind, placebo controlled, multicentre study to evaluate the efficacy and safety of givinostat in ambulant patients with duchenne muscular dystrophy (DSC/14/2357/48).

Sponsor: ITALFARMACO
Duration: 2018-
Principal investigator: Jordi Diaz Manera

10. Name of the trial: A phase 3 double-blind ranzomized study to assess the efficacy and safety of intravenous atb200 co-administered with oral at2221 in adult subjects with late-onset pompe disease compared with alglucosidase alfa (ATB200-03 AMICUS)
    Sponsor: Amicus
    Duration: 2019-
    Principal investigator: Jordi Diaz Manera

11. Name of the trial: A multicenter, randomized, parallel group, double blind, multiple dose, placebo controlled study to assess the efficacy and safety of mnk-1411 in male subjects 4 to 8 years of age with duchenne muscular dystrophy (MNK14112096 MALLINCROFT).
    Sponsor: MALLINCROFT
    Duration: 2019-
    Principal investigator: Jordi Diaz Manera

12. Name of the trial: An open-label extension study to evaluate the long-term effects of ace-083 in patients with facioscapulohumeral muscular dystrophy (fshd) previously enrolled in study a083-02 and patients with charcot-marie tooth (cmt) disease types 1 and x previously enrolled in study a083-03
    Sponsor: ACCELERON
    Duration: 2019-
    Principal investigator: Jordi Diaz Manera

13. Name of the trial: A phase 2, randomized, double-blind, placebo-controlled, 24-week, parallel-group study of the efficacy and safety of losmapimod in treating subjects with facioscapulohumeral muscular dystrophy (FSHD)(FIS-002-2019)
    Sponsor: FULCRUM
    Duration: 2019-
    Principal investigator: Jordi Diaz Manera

14. Name of the trial: A randomized, double-blind, placebo-controlled, efficacy and safety study of WVE-210201 in abulatory patients with duchenne muscular dystrophy (WVE-DMDX51-003)
    Sponsor: WAVE THERAPEUTICS
    Duration: TO BE STARTED IN 2020
    Principal investigator: Jordi Diaz Manera

15. Name of the trial: A randomized, double-blind, dose comparison study evaluating the safety and efficacy of two dose levels of eteplirsen in patients with duchenne muscular dystrophy amenable to exon 51 skipping (Sarepta 4658-402)
    Sponsor: SAREPTA
    Duration: TO BE STARTED IN 2020
    Principal investigator: Jordi Diaz Manera

16. Name of the trial: A phase 2, two-part, multiple-ascending-dose study of SRP-5051 for dose determination, then dose expansion, in patients with duchenne muscular dystrophy amenable to exon 51- Skipping treatment (Sarepta 5051-201)
    Sponsor: SAREPTA
    Duration: TO BE STARTED IN 2020
    Principal investigator: Jordi Diaz Manera

## B. SCIENTIFIC PUBLICATION

**As a summary the publications that Dr. Jordi Diaz Manera has co-authored are**

| *Ítem* | *Nombre* |
|---|---|
| B.1 Scientific papers in indexed journals | 102 |
| B.2 Scientific papers in non indexed journals | 1 |
| B.3 Book chapters | 4 |
| B.4 Other pulications | 2 |
| B.5 Congress abstracts published | 63 |
| | |

## SCIENTIFIC PAPERS PUBLISHED IN INDEXED JOURNALS

**1.** 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. Study of the accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology (in press)

**2.** Vissing J, Johnson K, Töpf A, Nafissi S, Díaz-Manera J, French VM, Schindler RF, Sarathchandra P, Løkken N, Rinné S, Freund M, Decher N, Müller T, Duno M, Krag T, Brand T, Straub V. POPDC3 gene variants associate with a new form of limb girdle muscular dystrophy. Ann Neurol. 2019 Oct 14. doi: 10.1002/ana.25620

**3.** Barp A, Laforet P, Bello L, Tasca G, Vissing J, Monforte M, Ricci E, Choumert A, Stojkovic T, Malfatti E, Pegoraro E, Semplicini C, Stramare R, Scheidegger O, Haberlova J, Straub V, Marini-Bettolo C, Løkken N, Diaz-Manera J, Urtizberea JA, Mercuri E, Kynčl M, Walter MC, Carlier RY. European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). J Neurol. 2019 Sep 25. doi: 10.1007/s00415-019-09539-y

**4.** Wood L, Bassez G, Bleyenheuft C, Campbell C, Cossette L, Jimenez-Moreno AC, Dai Y, Dawkins H, Díaz-Manera J, Dogan C, El Sherif R, Fossati B, Graham C, Hilbert J, Kastreva K, Kimura E, Korngut L, Kostera-Pruszczyk A, Lindberg C, Lindvall B, Luebbe E, Lusakowska A, Mazanec R, Meola G, Orlando L, Takahashi MP, Peric S, Puymirat J, Rakocevic-Stojanovic V, Rodrigues M, Roxburgh R, Schoser B, Segovia S, Shatillo A, Thiele S, Tournev I, van Engelen B, Vohanka S, Lochmüller H.
"Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease."
Orphanet J Rare Dis. 2019 Aug 15;14(1):199. doi: 10.1186/s13023-019-1157-7.

**5.** 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 Munáin 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 Jul 23. pii: S1096-7192(18)30784-4

**6.** 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*. *:Co-corresponding authors
"Identification of serum microRNAs as potential biomarkers in Pompe disease"
Ann Clin Transl Neurol. 2019 Jul;6(7):1214-1224

**7.** Carreño-Gago L, Blázquez-Bermejo C, Díaz-Manera J, Cámara Y, Gallardo E, Martí R, Torres-Torronteras J, García-Arumí E.
“Identification and Characterization of New RNASEH1 Mutations Associated With PEO Syndrome and Multiple Mitochondrial DNA Deletions.”
Front Genet. 2019 Jun 14;10:576

**8.** 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). 2019 Jun 25

**9.** Hogarth MW, Defour A, Lazarski C, Gallardo E, Diaz Manera J, Partridge TA, Nagaraju K, Jaiswal JK.
“Fibroadipogenic progenitors are responsible for muscle loss in limb girdle muscular dystrophy 2B”.
Nat Commun. 2019 Jun 3;10(1):2430

**10.** 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

**11.** Domínguez-González C, Hernández-Laín A, Rivas E, Hernández-Voth A, Sayas Catalán J, Fernández-Torrón R, Fuiza-Luces C, García García J, Morís G, Olivé M, Miralles F, Díaz-Manera J, Caballero C, Méndez-Ferrer B, Martí R, García Arumi E, Badosa MC, Esteban J, Jimenez-Mallebrera C, Encinar AB, Arenas J, Hirano M, Martin MÁ, Paradas C.
Late-onset thymidine kinase 2 deficiency: a review of 18 cases
Orphanet J Rare Dis. 2019 May 6;14(1):100

**12.** Gutiérrez Gutiérrez G, Díaz-Manera J, Almendrote M, Azriel S, Eulalio Bárcena J, Cabezudo García P, Camacho Salas A, Casanova Rodríguez C, Cobo AM, Díaz Guardiola P, Fernández-Torrón R, Gallano Petit MP, García Pavía P, Gómez Gallego M, Gutiérrez Martínez AJ, Jericó I, Kapetanovic García S, López de Munaín Arregui A, Martorell L, Morís de la Tassa G, Moreno Zabaleta R, Muñoz-Blanco JL, Olivar Roldán J, Pascual Pascual SI, Peinado Peinado R, Pérez H, Poza Aldea JJ, Rabasa M, Ramos A, Rosado Bartolomé A, Rubio Pérez MÁ, Urtizberea JA, Zapata-Wainberg G, Gutiérrez-Rivas E.
Clinical guide for the diagnosis and follow-up of myotonic dystrophy type 1, MD1 or Steinert's disease.

Neurologia. 2019 Apr 16. pii: S0213-4853(19)30019-2

**13.** 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

**14.** 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

**15.** 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

**16.** Duchateau L, Martín-Aguilar L, Lleixà C, Cortese A, Dols-Icardo O, Cervera-Carles L, Pascual-Goñi E, Diaz-Manera J, Calegari 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

**17.** 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

**18.** 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

**19.** Gutiérrez Gutiérrez G, Díaz-Manera J, Almendrote M, Azriel S, Eulalio Bárcena J, Cabezudo García P, Camacho Salas A, Casanova Rodríguez C, Cobo AM, Díaz Guardiola P, Fernández-Torrón R, Gallano Petit MP, García Pavía P, Gómez Gallego M, Gutiérrez Martínez AJ, Jericó I, Kapetanovic García S, López de Munaín Arregui A, Martorell L, Morís de la Tassa G, Moreno Zabaleta R, Muñoz-Blanco JL, Olivar Roldán J, Pascual Pascual SI, Peinado Peinado R, Pérez H, Poza Aldea JJ, Rabasa M, Ramos A, Rosado Bartolomé A, Rubio Pérez MÁ, Urtizberea JA, Zapata-Wainberg G, Gutiérrez-Rivas E.
Clinical guide for the diagnosis and follow-up of myotonic dystrophy type 1, MD1 or Steinert's disease.
Med Clin (Barc). 2019 Jul 19;153(2):82.e1-82.e17

**20.** Moore U, Jacobs M, James MK, Mayhew AG, Fernandez-Torron R, Feng J, Cnaan A, Eagle M, Bettinson K, Rufibach LE, Lofra RM, Blamire AM, Carlier PG, Mittal P, Lowes LP, Alfano L, Rose K, Duong T, Berry KM, Montiel-Morillo E, Pedrosa-Hernández I, Holsten S, Sanjak M, Ashida A, Sakamoto C, Tateishi T, Yajima H, Canal A, Ollivier G, Decostre V, Mendez JB, Sánchez-Aguilera Praxedes N, Thiele S, Siener C, Shierbecker J, Florence JM, Vandevelde B, DeWolf B, Hutchence M, Gee R, Prügel J, Maron E, Hilsden H, Lochmüller 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, Díaz-Manera J, Bello L, Semplicini C, Pegoraro E, Mendell JR, Bushby K, Straub V; Jain COS Consortium.
Assessment of disease progression in dysferlinopathy: A 1-year cohort study.
Neurology. 2019 Jan 9

**21.** 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

**22.** 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

**23.** GóMez-Andrés D, Díaz-Manera J, Alejaldre A, Pulido-Valdeolivas I, GonzáLez-Mera L, Olivé M, Vilchez JJ, De Munain AL, Paradas C, Muelas N, SáNchez-MontáÑez Á, Alonso-Jimenez A, De la Banda MGG, Dabaj I, Bonne G, Munell F, Carlier RY, Quijano-Roy S.
Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for follow-up.
Muscle Nerve. 2018 Dec;58(6):812-817

**24.** Díaz-Manera J, Luna S, Roig C.
Ocular ptosis: differential diagnosis and treatment.
Curr Opin Neurol. 2018 Oct;31(5):618-627

**25.** 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

**26.** 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

**27.** 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

**28.** Cabrera-Serrano M, Mavillard F, Biancalana V, Rivas E, Morar B, Hernández-Laín A, Olive M, Muelas N, Khan E, Carvajal A, Quiroga P, Diaz-Manera J, Davis M, Ávila R, Domínguez C, Romero NB, Vílchez JJ, Comas D, Laing NG, Laporte J, Kalaydjieva L, Paradas C.
A Roma founder BIN1 mutation causes a novel phenotype of centronuclear myopathy with rigid spine.
Neurology. 2018 Jul 24;91(4):e339-e348

**29.** Cortés-Vicente E, Rojas-Garcia 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

**30.** Siles AM, Martínez-Hernández E, Araque J, Diaz-Manera J, Rojas-Garcia 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

**31.** Diaz-Manera J, Fernandez-Torron 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, Coppenrath 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

**32.** Lostal W, Urtizberea JA, Richard I; calpain 3 study group.
233rd ENMC International Workshop:: Clinical Trial Readiness for Calpainopathies, Naarden, The Netherlands, 15-17 September 2017.
Neuromuscul Disord. 2018 Jun;28(6):540-549

**33.** Erdocia-Goni A, Alonso-Jimenez A, Ramon-Carbajo C, Garcia-Arumi E, Casquero P, Gallardo E, Diaz-Manera J.
Isolated girdle weakness: expansion of the phenotypic spectrum of the MERRF 8344A>G mutation of mitochondrial DNA.Rev Neurol. 2018 Apr 16;66(8):268-270

**34.** 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

**35.** Moore UR, Jacobs M, Fernandez-Torron R, Jang J, James MK, Mayhew A, Rufibach L, Mittal P, Eagle M, Cnaan A, Carlier PG, Blamire A, Hilsden H, Lochmüller 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, Diaz-Manera J, Bello L, Semplicini C, Pegoraro E, Mendell JR, Bushby K, Straub V.
Teenage exercise is associated with earlier symptom onset in dysferlinopathy: a retrospective cohort study.
J Neurol Neurosurg Psychiatry. 2018 Nov;89(11):1224-1226

**36.** Alonso-Jiménez A, Ramón C, Dols-Icardo O, Roig C, Gallardo E, Clarimón J, Núñez-Peralta C, Díaz-Manera J.
Corpus callosum agenesis, myopathy and pinpoint pupils: consider Stormorken syndrome.
Eur J Neurol. 2018 Feb;25(2):e25-e26

**37.** Martinez-Martinez L, Lleixà MC, Boera-Carnicero G, Cortese A, Devaux J, Siles A, Rajabally Y, Martinez-Piñeiro A, Carvajal A, Pardo J, Delmont E, Attarian S, Diaz-Manera J, Callegari I, Marchioni E, Franciotta D, Benedetti L, Lauria G, de la Calle Martin 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

**38.** Koeks Z, Bladen CL, Salgado D, van Zwet E, Pogoryelova O, McMacken G, Monges S, Foncuberta ME, Kekou K, Kosma K, Dawkins H, Lamont L, Bellgard MI, Roy AJ, Chamova T, Guergueltcheva V, Chan S, Korngut L, Campbell C, Dai Y, Wang J, Barišić N, Brabec P, Lähdetie J, Walter MC, Schreiber-Katz O, Karcagi V, Garami M, Herczegfalvi A, Viswanathan V, Bayat F, Buccella F, Ferlini A, Kimura E, van den Bergen JC, Rodrigues M, Roxburgh R, Lusakowska A, Kostera-Pruszczyk A, Santos R, Neagu E, Artemieva S, Rasic VM, Vojinovic D, Posada M, Bloetzer C, Klein A, Díaz-Manera J, Gallardo E, Karaduman AA, Oznur T, Topaloğlu H, El Sherif R, Stringer A, Shatillo AV, Martin AS, Peay HL, Kirschner J, Flanigan KM, Straub V, Bushby K, Béroud C, Verschuuren JJ, Lochmüller H.
Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database
J Neuromuscul Dis. 2017;4(4):293-306

**39.** Querol L, Siles AM, Alba-Rovira R, Jáuregui A, Devaux J, Faivre-Sarrailh C, Araque J, Rojas-Garcia R, Diaz-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

**40.** 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 Oct 20. pii: S1474-4422(17)30369-1.

**41.** Natera-de Benito D, Töpf A, Vilchez JJ, González-Quereda L, Domínguez-Carral J, **Díaz-Manera J**, Ortez C, Bestué M, Gallano P, Dusl M, Abicht A, Müller JS, Senderek J, García-Ribes A, Muelas N, Evangelista T, Azuma Y, McMacken G, Paipa Merchan A, Rodríguez Cruz PM, Camacho A, Jiménez E, Miranda-Herrero MC, Santana-Artiles A, García-Campos O, Dominguez-Rubio R, Olivé M, Colomer J, Beeson D, Lochmüller H, Nascimento A.
"Molecular characterization of congenital myasthenic syndromes in Spain".
Neuromuscul Disord. 2017 Aug 18. pii: S0960-8966(17)30475-3. doi: 10.1016/j.nmd.2017.08.003. [Epub ahead of print]

**42.** Tasca G, Monforte M, **Díaz-Manera J**, Brisca G, Semplicini C, D'Amico A, Fattori F, Pichiecchio A, Berardinelli A, Maggi L, Maccagnano E, Løkken N, Marini-Bettolo C, Munell F, Sanchez A, Alshaikh N, Voermans NC, Dastgir J, Vlodavets D, Haberlová J, Magnano G, Walter MC, Quijano-Roy S, Carlier RY, van Engelen BGM, Vissing J, Straub V, Bönnemann CG, Mercuri E, Muntoni F, Pegoraro E, Bertini E, Udd B, Ricci E, Bruno C.
"MRI in sarcoglycanopathies: a large international cohort study".
J Neurol Neurosurg Psychiatry. 2017 Sep 9.

**43.** Rodríguez MA, Del Rio Barquero LM, Ortez CI, Jou C, Vigo M, Medina J, Febrer A, Ramon-Krauel M, **Diaz-Manera J**, Olive M, González-Mera L, Nascimento A, Jimenez-Mallebrera C.
"Differences in Adipose Tissue and Lean Mass Distribution in Patients with Collagen VI Related Myopathies Are Associated with Disease Severity and Physical Ability".
Front Aging Neurosci. 2017 Aug 8;9:268.

**44.** De Luna N, Suárez-Calvet X, Lleixà C, **Diaz-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.

**45.** 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

**46.** Van der Ploeg AT, Kruijshaar ME, Toscano A, Laforêt P, Angelini C, Lachmann RH, Pascual Pascual SI, Roberts M, Rösler K, Stulnig T, van Doorn PA, Van den Bergh PYK, Vissing J, Schoser B; **European Pompe Consortium** (forma part d'aquest grup).
"European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience".
Eur J Neurol. 2017 Jun;24(6):768-e31

**47.** 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.

**48.** Garibaldi M, Tasca G, **Diaz-Manera J**, Ottaviani P, Laschena F, Pantoli D, Gerevini S, Fiorillo C, Maggi L, Tasca E, D'Amico A, Musumeci O, Toscano A, Bruno C, Massa R, Angelini C, Bertini E, Antonini G, Pennisi EM. "Muscle MRI in neutral lipid storage disease (NLSD)".
J Neurol. 2017 Jul;264(7):1334-1342. Erratum in: J Neurol. 2017 Jun 12.

**49.** Cortés-Vicente E, Pradas J, Marín-Lahoz J, De Luna N, Clarimón J, Turon-Sans J, Gelpí E, **Díaz-Manera J**, Illa I, Rojas-Garcia 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.

**50.** Marsolier J, Laforet P, Pegoraro E, Vissing J, Richard I, **Sarcoglycanopathies Working Group** (Barnerias C, Carlier RY, **Díaz-Manera J**, Fayssoil A, Galy A, Gazzerro E, Górecki D, Guglieri M, Hogrel JY, Israeli D, Leturcq F, Moussu H, Prigent H, Sandona D, Schoser B, Semplicini C, Talim B, Tasca G, Urtizberea A, Udd B.).
"1st International Workshop on Clinical trial readiness for sarcoglycanopathies 15-16 November, 2016, Evry, France".
Neuromuscul Disord. 2017 Jul;27(7):683-692.

**51.** 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.

**52.** 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 Cortactin".
JAMA Neurol. 2016 Sep 1;73(9):1099-104.

**53.** Harris E, Bladen CL, Mayhew A, James M, Bettinson K, Moore U, Smith FE, Rufibach L, Cnaan A, Bharucha-Goebel DX, Blamire AM, Bravver E, Carlier PG, Day JW, **Díaz-Manera J**, Eagle M, Grieben U, Harms M, Jones KJ, Lochmüller H, Mendell JR, Mori-Yoshimura M, Paradas C, Pegoraro E, Pestronk A, Salort-Campana E, Schreiber-Katz O, Semplicini C, Spuler S, Stojkovic T, Straub V, Takeda S, Rocha CT, Walter MC, Bushby K; Jain COS Consortium.
"The Clinical Outcome Study for dysferlinopathy: An international multicenter study".
Neurol Genet. 2016 Aug 4;2(4):e89.

**54.** Garibaldi M, **Diaz-Manera J**, Gallardo E, Antonini G.
"Teaching Video NeuroImages: The Beevor sign in late-onset Pompe disease".
Neurology. 2016 Jun 14;86(24):e250-1.

**55.** Cruz S, Figueroa-Bonaparte S, Gallardo E, de Becdelièvre A, Gartioux C, Allamand V, Piñol P, Garcia 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.

**56.** 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.

**57.** **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.

**58.** Paco S, Casserras T, Angels Rodriguez M, Jou C, Puigdelloses M, Ortez C I, **Diaz-Manera J**, Gallardo E, Colomer J, Nascimento A, Kalko S G, Jimenez-Mallebrera C.
"Transcriptome analysis of ullrich congenital muscular dystrophy fibroblasts reveals a disease extracellular matrix signature and key molecular regulators".
PLoS One. 2015 Dec 15;10(12):e0145107.

**59.** **Diaz-Manera J**, Llauger J, Gallardo E, Illa I.
"Muscle MRI in muscular dystrophies".
Acta Myol. 2015 Dec;34(2-3):95-108.

**60.** Teresa Osuna M, Querol L, Olivas-Chacon C I, Lejarreta-Andres S, Robert J, Ailouti-Caballero N, Ramos-Duran L, **Diaz-Manera J**, Belvis R.
"Toxic demyelinating neuropathy and leukoencephalopathy in patients who take the slimming products Thermatrim (R) and Pura Alegria (R)".
Rev Neurol. 2015 Dec 1;61(11):527-8. Spanish.

**61.** Querol L, Rojas-Garcia R, **Diaz-Manera J**, Barcena J, Pardo J, Ortega-Moreno A, Sedano, M J, Sero-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.

**62.** Gutierrez-Rivas E, Bautista J, Vilchez JJ, Muelas N, **Diaz-Manera J**, Illa I, Martinez-Arroyo A, Olive M, Sanz I, Arpa J, Fernandez-Torron R, Lopez de Munain A, Jimenez L, Solera J, Lukacs Z.
"Targeted screening for the detection of Pompe disease in patients with unclassified limb-girdle muscular dystrophy or asymptomatic hyperCKemia using dried blood: A Spanish cohort".
Neuromuscul Disord. 2015 Jul;25(7):548-53.

**63.** Ramos-Fransi A, Rojas-Garcia R, Segovia S, Marquez-Infante C, Pardo J, Coll-Canti J, Jerico 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 Jul;22(7):1056-61.

**64.** Camara Y, Carreno-Gago L, Martin M A, Melia M J, Blazquez A, Delmiro A, Aitor, Garrabou G, Moren C, **Diaz-Manera J**, Gallardo E, Bornstein B, Lopez-Gallardo E, Hernandez-Lain A, San Millan B, Cancho E, Samuel Rodriguez-Vico J, Marti R, Garcia-Arumi E.
"Severe TK2 enzyme activity deficiency in patients with mild forms of myopathy".
Neurology. 2015 Jun 2;84(22):2286-8.

**65.** Bladen C L, Salgado D, Monges S, Foncuberta M E, Kekou K, Kosma K, Dawkins H, Lamont L, Roy A J, Chamova T, Guergueltcheva V, Chan S, Korngut L, Campbell C, Dai Y, Wang J, Barisic N, Brabec P, Lahdetie J, Walter M C, Schreiber-Katz O, Karcagi V, Garami M, Viswanathan V, Bayat F, Buccella F, Kimura E, Koeks Z, van den Bergen J C, Rodrigues M, Roxburgh R, Lusakowska A, Kostera-Pruszczyk A, Zimowski J, Santos R, Neagu E, Artemieva S, Rasic V M, Vojinovic D, Posada M, Bloetzer C, Jeannet P-Y, Joncourt F, **Diaz-Manera J**, Gallardo E, Karaduman A A, Topaloglu H, El Sherif R, Stringer A, Shatillo A V, Martin A S, Peay H L, Bellgard M I, Kirschner J, Flanigan K M, Straub V, Bushby K, Verschuuren J, Aartsma-Rus A, Beroud C, Lochmueller H.
"The TREAT-NMD DMD global database: analysis of more than 7,000 duchenne muscular dystrophy mutations".
Hum Mutat. 2015 Apr;36(4):395-402. doi: 10.1002/humu.22758. Epub 2015 Mar

**66.** Gomez-Anson B, Roman E, Fernandez de Bobadilla R, Pires-Encuentra P,
**Diaz-Manera J**, Nunez Fidel, Martinez-Horta S, Vives-Gilabert Y, Pagonabarraga J, Kulisevsky J, Cordoba J, Guarner C, Soriano G.
"Alterations in cerebral white matter and neuropsychology in patients with cirrhosis and falls".
PLoS One. 2015 Mar 20;10(3):e0118930.

**67.** Gutierrez-Rivas E, Bautista J, Vilchez J J, Muelas N, **Diaz-Manera J**, Illa I, Martinez-Arroyo A, Olive M, Sanz I, Arpa J, Fernandez-Torron R, Lopez de Munain A, Jimenez L, Solera J, Lukacs Z.
"Dried blood spot for screening for late-onset pompe disease: a spanish cohort".
J Neuromuscul Dis. 2015;2(s1):S42.

**68.** Grau-Rivera O, Gelpi E, Nos C, Gaig C, Ferrer I, Saiz A, Llado A, Molinuevo J L, Graus F, Sanchez-Valle R. **Neurological Tissue Bank (autoria conjunta)**.
"Clinicopathological correlations and concomitant pathologies in rapidly progressive dementia: a brain bank series".
Neurodegener Dis. 2015;15(6):350-60.

**69.** Gallardo E, Martinez-Hernandez E, Titulaer M J, Huijbers M G, Angeles Martinez M, Ramos A, Querol L, **Diaz-Manera J**, Rojas-Garcia R, Hayworth C R, Verschuuren J J, Balice-Gordon R, Dalmau J, Illa I.
"Cortactin autoantibodies in myasthenia gravis".
Autoimmun Rev. 2014 Oct;13(10):1003-7.

**70.** **Diaz-Manera J**, Querol L, Alejaldre A, Rojas-Garcia 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. Erratum in J Hum Genet. 2014 Aug;59(8):477.

**71.** Gallardo E, Ankala A, Nunez-Alvarez Y, Hegde M, **Diaz-Manera J**, De Luna N, Pastoret A, Suelves M, Illa I.
"Genetic and epigenetic determinants of low dysferlin expression in monocytes".
Hum Mutat. 2014 Aug;35(8):990-7.

**72.** Suarez-Calvet X, Gallardo E, Nogales-Gadea G, Querol L, Navas M, **Diaz-Manera J**, Rojas-Garcia R, Illa I.
"Altered RIG-I/DDX58-mediated innate immunity in dermatomyositis".
J Pathol. 2014 Jul;233(3):258-68.

**73.** **Diaz-Manera J**, Alejaldre A, Llauger J, Mirabet S, Rojas-Garcia 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.

**74.** Nogales-Gadea G, Ramos-Fransi A, Suarez-Calvet X, Navas M, Rojas-Garcia R, Luis Mosquera J, **Diaz-Manera J**, Querol L, Gallardo E, Illa I.
"Analysis of serum miRNA profiles of myasthenia gravis patients".
PLOS ONE. 9 - 3, PUBLIC LIBRARY SCIENCE, 17/03/2014.
PLoS One. 2014 Mar 17;9(3):e91927.

**75.** Querol L, Nogales-Gadea G, Rojas-Garcia R, **Diaz-Manera J**, Pardo J, Ortega-Moreno A, Jose Sedano M, 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.

**76.** Flix B, Suarez-Calvet X, **Diaz-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.

**77.** Querol L, Nogales-Gadea G, Rojas-Garcia R, Martinez-Hernandez E, **Diaz-Manera J**, Suarez-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.

**78.** **Diaz-Manera J**, Gallardo E, De Luna N, Navas M, Soria L, Garibaldi M, Rojas-Garcia 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 Dec;228(4):544-53.

**79.** Juan-Mateu J, Rodriguez M J, Nascimento A, Jimenez-Mallebrera C, Gonzalez-Quereda L, Rivas E, Paradas C, Madruga M, Sanchez-Ayaso P, Jou C, Gonzalez-Mera L, Munell F, Roig-Quilis M, Rabasa M, Hernandez-Lain A, **Diaz-Manera J**, Gallardo E, Pascual J, Verdura E, Colomer J, Baiget M, Olive M, Gallano P.
"Prognostic value of X-chromosome inactivation in symptomatic female carriers of dystrophinopathy".
Orphanet J Rare Dis. 2012 Oct 23;7:82.

**80.** Alejaldre A, **Diaz-Manera J**, Ravaglia S, Tibaldi E C, D'Amore F, Moris G, Muelas N, Vilchez J J, Garcia-Medina A, Uson M, Martinez Garcia F, Francisco A, 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.

**81. Diaz-Manera J**, De Luna N, Paradas C, Iturriaga C, Rojas-Garcia R, Araque J, Genebriera M, Gich I, Illa I, Gallardo E.
"1 alpha,25(OH)(2)-vitamin D3 increases dysferlin expression in vitro and in a human clinical trial".
Mol Ther. 2012 Oct;20(10):1988-97.

**82. Diaz-Manera J**, Rojas Garcia R, Illa I.
"Treatment strategies for myasthenia gravis: an update".
Expert Opin Pharmacother. 2012 Sep;13(13):1873-83. Review.

**83. Diaz-Manera J**, Martinez-Hernandez E, Querol L, Klooster R, Rojas-Garcia R, Suarez-Calvet X, Munoz-Blanco J L, Mazia C, Straasheijm K R, Gallardo E, Juarez C, Verschuuren J J, Illa I.
"Long-lasting treatment effect of rituximab in MuSK myasthenia".
NEUROLOGY. 78 - 3, pp. 189 - 193. LIPPINCOTT WILLIAMS & WILKINS, 01/01/2012.
Neurology. 2012 Jan 17;78(3):189-93.

**84.** Gallardo E, de Luna N, **Diaz-Manera J**, Rojas-Garcia R, Gonzalez-Quereda L, Flix B, de Morree 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.

**85. Diaz-Manera J**, Querol L, Clarimon J, Yaguee S, Illa I.
"Unique post-exercise electrophysiological test results in a new Andersen-Tawil syndrome mutation".
Clin Neurophysiol. 2011 Dec;122(12):2537-9.

**86.** Pinos T, Marotta M, Gallardo E, Illa I, **Diaz-Manera J**, Gonzalez-Vioque E, Garcia-Arumi E, Andreu A L, Marti 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.

**87. Diaz-Manera J**, Touvier T, Dellavalle A, Tonlorenzi R, Tedesco F S, 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.

**88.** Paradas C, Llauger J, **Diaz-Manera J**, Rojas-Garcia R, De Luna N, Iturriaga C, Marquez C, Uson 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.

**89.** Fiaschi t, Tedesco F S, Giannoni E, **Diaz-Manera J**, Parri M, Cossu G, Chiarugi P.
"Globular Adiponectin as a complete mesoangioblast regulator: role in proliferation, survival, motility, and skeletal muscle differentiation".
Mol Biol Cell. 2010 Mar 15;21(6):848-59.

**90.** Tedesco F S, Dellavalle A, **Diaz-Manera J**, Messina G, Cossu G.
"Repairing skeletal muscle: regenerative potential of skeletal muscle stem cells".
J Clin Invest. 2010 Jan;120(1):11-9.

**91. Diaz-Manera J**, Rojas-Garcia R, Illa I.
"Treatment strategies for myasthenia gravis".
Expert Opin Pharmacother. 2009 Jun;10(8):1329-42.

**92. Diaz-Manera J**, Rojas-Garcia R, Gallardo E, Illa I.
"Response to methotrexate in a chronic inflammatory demyelinating polyradiculoneuropathy patient".
Muscle Nerve. 2009 Mar;39(3):386-8.

**93.** Marti-Fabregas J, Martinez-Ramirez S, Martinz-Corral M, **Diaz-Manera J**, Querol L, Suarez-Calvet M, de Juan M, Santalo M, Marin R, Marti-Vilalta, J-L.

"Blood pressure is not associated with haematoma enlargement in acute intracerebral haemorrhage".
Eur J Neurol. 2008 Oct;15(10):1085-90.

**94.** 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.

**95.** Rojas-Garcia R, Gallardo E, Povedano M, de Luna N, Bruna J, Juarez C, **Diaz-Manera J**, Martinez-Matos J A, Illa I.
"Antibodies against disialosyl and terminal NeuNAc(alpha 2-3)Gal ganglioside epitopes in acute relapsing sensory ataxic neuropathy".
J Neurol. 2008 May;255(5):764-6.

**96.** Aguirregomozcorta M, Pagonabarraga J, **Diaz-Manera J**, Pascual-Sedano B, Gironell A, Kulisevsky J.
"Efficacy of botulinum toxin in severe Tourette syndrome with dystonic tics involving the neck".
Parkinsonism Relat Disord. 2008;14(5):443-5.

**97. Diaz-Manera J**, Rojas-Garcia R, Gallardo E, Juarez C, Martinez-Domeno A, Martinez-Ramirez S, Dalmau J, Blesa R, Illa I.
"Antibodies to AChR, MuSK and VGKC in a patient with myasthenia gravis and Morvan's syndrome".
Nat Clin Pract Neurol. 2007 Jul;3(7):405-10.

**98.** Marti-Fabregas J, Bravo Y, Cocho D, Marti-Vilalta J-L, **Diaz-Manera J**, San Roman L, Puig M, Blanco M, Castellanos M, Millan M, Roquer J, Obach V, Maestre J.
"Frequency and predictors of symptomatic intracerebral hemorrhage in patients with ischemic stroke treated with recombinant tissue plasminogen activator outside clinical trials".
Cerebrovasc Dis. 2007;23(2-3):85-90.

**99.** Rojas-Garcia R, Martinez-Lage M, Gallardo E, de Luna N, Marti-Vilalta J-L, **Diaz-Manera J**, Juarez C, Blesa R, Illa I.
"A novel antiganglioside pecificity against terminal NeuNAc(alfa 2-3)Gal in acute bulbar palsy".
J Neuroimmunol. 2006 Jul;176(1-2):219-22.

**100.** Cocho D, Borrell M, Marti-Fabregas J, Montaner J, Castellanos M, Bravo Y, Molina-Porcel L, Belvis R, **Diaz-Manera J**, Martinez-Domeno A, Martinez-Lage M, Millan M, Fontcuberta J, Marti-Vilalta JL.
"Pretreatment hemostatic markers of symptomatic intracerebral hemorrhage in patients treated with tissue plasminogen activator".
Stroke. 2006 Apr;37(4):996-9.

**101.** Cocho D, Belvis R, Marti-Fabregas J, Bravo Y, Aleu A, Pagonabarraga J, Molina-Porcel L, **Diaz-Manera J**, San Roman L, Martinez-Lage M, Martinez, A, Moreno M, Marti-Vilalta JL.
"Does thrombolysis benefit patients with lacunar syndrome?".
Eur Neurol. 2006;55(2):70-3.

**102.** Marti-Fabregas J, Borrell M, Cocho D, Belvis R, Castellanos M, Montaner J, Pagonabarraga J, Aleu A, Molina-Porcel L, **Diaz-Manera J**, Bravo Y, Alvarez-Sabin J, Davalos A, Fontcuberta J, Marti-Vilalta JL.
"Hemostatic markers of recanalization in patients with ischemic stroke treated with rt-PA".
NEUROLOGY. 65 - 3, pp. 366 - 370. LIPPINCOTT WILLIAMS & WILKINS, 09/08/2005.
Neurology. 2005 Aug 9;65(3):366-70.

**103.** Illa I, **Diaz-Manera J**, Juarez C, Rojas-Garcia R, Molina-Porcel L, Aleu A, Pradas J, Gallardo E.
"Seronegative" myasthenia gravis and antiMuSK positive antibodies: Description of Spanish series".
MEDICINA CLINICA. 125 - 3, pp. 100 - 102. 18/06/2005.
Med Clin (Barc). 2005 Jun 18;125(3):100-2. Spanish.

**104.** Cocho D, Belvis R, Marti-Fabregas J, Molina-Porcel L, **Diaz-Manera J**, Aleu A, Pagonabarraga J, Garcia-Bargo D, Mauri A, Marti-Vilalta JL.
"Reasons for exclusion from thrombolytic therapy following acute ischemic stroke".
Neurology. 2005 Feb 22;64(4):719-20.

**105.** Marti-Fabregas J, Gomis M, Arboix A, Aleu A, Pagonabarraga J, Belvis R, Cocho D, Roquer J, Rodriguez A, Garcia M D, Molina-Porcel L, **Diaz-Manera J**, Mart-Vilalta J L.
"Favorable outcome of ischemic stroke in patients pretreated with statins".
Stroke. 2004 May;35(5):1117-21.

## SCIENTIFIC PAPERS PUBLISHED IN NON INDEXED JOURNALS

1. **Diaz-Manera J**, Illa I. "Aproximació diagnòstica a les miopaties".
   Neurologia Catalana (Butlletí de la Societat Catalana de Neurologia), Número 28, pàgines 16-23.

## BOOK CHAPTERS

**1**. Rojas-García R, **Díaz-Manera J**, Illa I.
"Enfermedades de músculo, unión neuromuscular y nervio periférico".
IN: Manual de diagnóstico y terapéutica neurológicas (2a edició).

Pages: 485-535
Editors: J. A. Molina, M. E. Luquin i F.J.Jiménez-Jiménez
Editorial: Viguera, Barcelona
ISBN-13: 978-84-85424-65-8
Year: 2007

**2**. Rojas-García R, **Díaz-Manera J**, Illa I.
"Análisis de la actividad muscular espontánea".
In: Manual de electromiografía clínica.

Editors: Eduardo Gutiérrez-Rivas, Dolores Jiménez, Julio Pardo i Manuel Romero
Editorial: Ergon, Majadahonda (Madrid)
ISBN-13: 978-84-8473-600-4
Year: 2008

**3**. **Díaz-Manera J**, Rojas García R, Illa I.
"Trastornos de la union neuromuscular".
In: Tratado de Neurología.

Pages: 1117-1149
Editors: Julio Pascual
Editorial: Luzan 5. Madrid
ISBN: 978-84-7989-698-0
Year: 2012

**4**. **Díaz-Manera J**, Gallardo E.
"Distrofinopatías"
En: Manual de Enfermedades Neuromusculares

Pages 375-386
Editor: Eduardo Gutierrez Rivas
Editorial: Ergon
ISBN: 978-84-16732-35-7
Year: 2017

**OTHER PUBLICATIONS**

**1.** Autors: Neuromuscular Disorders Study Group-Spanish Society of Neurology.
Títol: "Diagnostics Algorithms for Neuromuscular Disorders"
Year: 2013
Pages: Published as Phone Applications
http://www.sen.es/aplicaciones-sen/2162-algoritmos-enfermedades-neuromusculares
https://play.google.com/store/apps/details?id=com.mindtheapp.algoritmos&hl=es
https://itunes.apple.com/es/app/algoritmos-enfermedades-neuromusculares/id1053800020?mt=8
Editorial: Ediciones SEN – Editoral de la Sociedad Española de Neurología

**2.** Autors: Díaz-Manera J, Illa I.
Name: Therapeutoc guidelines of Myasthenia Gravis
Year: 2011
Pages: 381-401
Editorial: Catalan Society of Neurology
ISBN: 978-84-614-7767-8
Dipòsit legal: LL-116-2011

## SCIENTIFIC PUBLICATIONS AS A RESULT OF CONGRESS PRESENTATIONS

1. Luciana Martel, MD, Alicia Alicia, MD, Helena Basuñana, MD, Jordi Diaz-Manera, PhD, Jaume Llauger, MD, Claudia Nuñez-Peralta, MD, Betina Biagetti, MD, Paula Montesinos, PhD, Susan Webb, MD,PHD, Elena Valassi, MD,PHD
   "MON-LB074 Ultrasonography May Reliably Assess Muscle Architecture in Patients with Cushing's Syndrome in Remission: Comparison with Gold-Standard Muscle MRI"Journal of the Endocrine Society, Volume 3, Issue Supplement_1, April-May 2019, MON-LB074

2. Luciana Martel, MD, Helena Bascuñana, MD, Alicia Alonso, MD, Jordi Diaz-Manera, PHD, Jaume Llauger, MD, Claudia Nuñez-Peralta, MD, Betina Biagetti, MD, Paula Montesinos, PHD, Susan Webb, MD,PHD, Elena Valassi, MD, PHD
   "MON-LB073 The Serum Creatinine to Serum Cystatin C Ratio Is a Reliable Surrogate Marker of Sarcopenia in Patients with Cushing's Syndrome in Remission" Journal of the Endocrine Society, Volume 3, Issue Supplement_1, April-May 2019, MON-LB073

3. Luciana Martel1, Alicia Alonso2, Helena Bascuñana3, Jordi Diaz Manera2, Jaume Llauger4, Claudia Nuñez-Peralta4, Betina Biagetti5, Paula Montesinos6, Susan Webb1 & Elena Valassi1
   Skeletal muscle fatty infiltration in the thigh, as assessed by MRI T2-weighted and 3-point Dixon sequences, is associated with poor performance on muscle function testing in patients with Cushing's syndrome in remission
   Edocrine Abstracts (2019) 63 GP159 | DOI: 10.1530/endoabs.63.GP159

4. Linda Lowes, Meredith James, Anna Mayhew, Lindsay Alfano, Marni Jacobs, Simone Spuler, Kristi J. Jones, John Day, Diana Bharucha-Goebel, Emmanuelle Campana-Salort, Alan Pestronk, Maggie Walter, Carmen Paradas, Tanya Stojkovic, Madoka Mori-Yoshimura, Elena Bravver, Elena Pegoraro, Jordi Diaz Manera, Tina Duong, Kristy Rose, Jerry Mendell, Kate Bushby, Volker Straub
   Examining longitudinal functional changes in Dysferlinopathy: The JAIN Clinical Outcome Study (P5.429)
   April 10, 2018; 90 (15 Supplement) APRIL 26, 2018

5. D. Natera-de Benito1, A.R. Foley2, J. Diaz-Manera3, M. Fink2, A. Mebrahtu2, J. Minal2, S. Donkervoort2, Y. Hu2, D. Cuadras2, L. Carrera-Garcia1, M. Alarcón1, J. Milisenda4, R. Dominguez5, M. Olive5, C. Jou1, J. Colomer1, C. Jimenez-Mallebrera1, C. Bönnemann2, A. Nascimento1
   P.330Long-term motor function in collagen VI-related myopathies is associated with the maximal motor ability achieved: a classification proposal
   Neuromuscular disorders 2018 28(S2)p.130

6. S. Figueroa-Bonaparte, S. Segovia, I. Belmonte, I. Pedrosa, E. Montiel, J. Llauger, A. Alonso-Jimenez, E. Gallardo, I. Illa, J. Diaz-Manera
   Antibodies anti-ERT do not influence muscle fatty infiltration in a long cohort of patients with late onset Pompe disease
   Neuromuscular disorders 2018 28(S2)p.162

7. P. Marti1, N. Muelas1, J. Diaz-Manera2, J. Vilchez1
   Usefulness of MRI in cases of hyperCKemia
   Neuromuscular disorders 2018 28(S2)p.162

8. L. Gonzalez-Quereda1, M. Rodriguez2, A. Nascimento3, C. Ortez3, C. Jou3, J. Milisenda4, J. Diaz-Manera2, I. Jerico5, I. Tejada6, P. Gallano1

Two different NGS approaches to address molecular diagnosis of congenital neuromuscular diseases
Neuromuscular disorders 2018 28(S2)p.197

9. N. De Luna1, X. Suárez-Calvet1, M. Garicano1, E. Fernández-Simón1, R. Rojas-Garcia2, J. Diaz-Manera2, L. Querol2, I. Illa2, E. Gallardo1 Effect of MAPK Inhibition on the differentiation of rhabdomyosarcoma cell line TE671 combined with CRISPR/Cas9 technology: an in vitro model for the study of human muscle diseases
Neuromuscular disorders 2018 28(S2)p.33

10. M. James1, A. Mayhew1, M. Jacobs2, S. Spuler3, J. Day4, K. Jones5, D. Bharucha-Goebel2, E. Salort-Campana6, A. Pestronk7, M. Walter8, C. Paradas9, T. Stojkovic10, M. Mori-Yoshimura11, E. Bravver12, J. Diaz Manera13, E. Pegoraro14, J. Mendell15, K. Bushby1, V. Straub
Rasch analysis of the individualised neuromuscular Quality of Life Questionnaire administered to patients with dysferlinopathy
Neuromuscular disorders 2018 28(S2)p.33

11.

12. Alonso-Jimenez, A.; Alejaldre-Monforte, A.; Dominguez-Gonzalez, C.; Cortes-Vicente, E.; Rojas-Garcia, R.; Tasca, G.; Carlier, R.; Monforte, M.; Laforet, P.; Gutierrez-Gutierrez, G.; Lopez de Munain, A.; Fernandez-Torron, R.; Ilia, I.; Diaz-Manera, J.
"Magnetic resonance image in oculopharyngeal muscular dystrophy"
"NEUROMUSCULAR DISORDERS". 27, pp. S122 - S123.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

13. Figueroa-Bonaparte, S.; Segovia, S.; Belmonte, I.; Pedrosa, I.; Montiel, E.; Llauger, J.; Alonso-Jimenez, A.; Gallardo, E.; Ilia, I.; Diaz-Manera, J.
"Antibodies anti-ERT do not influence muscle fatty infiltration in a long cohort of patients with late onset Pompe disease"
"NEUROMUSCULAR DISORDERS". 27, pp. S162 - S162. 01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

14. Gomez Andres, D, Diaz Manera, J, Garcia de la Banda, M. Gomez, Sanchez-Carpintero, A Alonso-Jimenez, A, Alejaldre-Monforte, A, Munell, F, Carlier, R, Quijano-Roy, S.
"Random forest approach to assess relationships of subjective muscle fatty infiltration with age at clinical onset and time of disease evolution in LMNA-related muscle disorders"
NEUROMUSCULAR DISORDERS, 27: S138-S138
DOI: 10.1016/j.nmd.2017.06.166 Fecha de publicación: OCT 2017
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

15. Gonzalez-Quereda, L.; Rodriguez, M.; Nascimento, A.; Ortez, C.; Jou, C.; Milisenda, J.; Diaz-Manera, J.; Jerico, I.; Tejada, I.; Gallano, P."
"Two different NGS approaches to address molecular diagnosis of congenital neuromuscular diseases"

"NEUROMUSCULAR DISORDERS". 27, pp. S197 - S197.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

16. James, M.; Jacobs, M.; Mayhew, A.; Feng, J.; Spuler, S.; Day, J.; Jones, K.; Bharucha-Goebel, D.; Salort-Campana, E.; Pestronk, A.; Walter, M.; Paradas, C.; Stojkovic, T.; Mori-Yoshimura, M.; Bravver, E.; Diaz-Manera, J.; Pegoraro, E.; Mendell, J.; Bushby, K.; Straub, V.
"Clinical outcome study of dysferlinopathy: what are the best outcome measures for dysferlinopathy patients?"
"NEUROMUSCULAR DISORDERS". 27, pp. S227 - S227.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

17. Marti, P.; Muelas, N.; Diaz-Manera, J.; Vilchez, J.
"Usefulness of MRI in cases of hyperCKemia"
"NEUROMUSCULAR DISORDERS". 27, pp. S123 - S123.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

18. Pinol, P.; Fernandez-Simon, E.; Suarez, X.; de Luna, N.; Molins, A.; de Oliva, N.; Martinez, A.; Escudero, L.; Sanchez, D.; Navarro, X.; Illa, I.; Gallardo, E.; Diaz-Manera, J.
"Nintedanib as a new therapeutic agent for Duchenne muscular dystrophy: preclinical in vitro and in vivo studies"
"NEUROMUSCULAR DISORDERS". 27, pp. S191 - S191.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

19. Torron, R. Fernandez; Harris, E.; Bourke, J.; Bettinson, K.; Hilsden, H.; Spuler, S.; Day, J.; Jones, K.; Bharucha-Goebel, D.; Salort-Campana, E.; Pestronk, A.; Walter, M.; Paradas, C.; Stojkovic, T.; Mori-Yoshimura, M.; Bravver, E.; Diaz-Manera, J.; Pegoraro, E.; Mendell, J.; Bushby, K.; Straub, V.
"Is cardiac dysfunction a feature of dysferlinopathy? Data from the clinical outcome study of dysferlinopathy"
"NEUROMUSCULAR DISORDERS". 27, pp. S145 - S145.
01/10/2017. ISSN 0960-8966, ISSN 1873-2364
22nd International Annual Congress of the World-Muscle-Society (WMS)
Saint Malo, FRANCE Fecha: OCT 03-07, 2017

20. Alonso-Jimenez A, Belvis-Nieto R, Diaz-Manera J, et ál.
"Acute demyelinating polyneuropathy resembling guillain-barre syndrome in a patient taking the slimming product pura alegria (R)".
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM".
Volum: 22 (3): 233-234. SEP 2017
Peripheral-Nerve-Society Meeting. Sitges. JUL 08-12, 2017.

21. Siles A M, Assylbekova D, Diaz-Manera J, et ál.
"Regulatory B cell frequencies increase after ivig therapy in inflammatory neuropathies".
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 22 (3): 233-234. SEP 2017
Peripheral-Nerve-Society Meeting. Sitges. JUL 08-12, 2017.

22. Siles A M, Martinez-Hernandez E, Diaz-Manera J, et ál.
"Antibodies against cell adhesion molecules and neural structures in paraneoplastic neuropathies".
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM".
Volum: 22 (3): 233-234. SEP 2017
Peripheral-Nerve-Society Meeting. Sitges. JUL 08-12, 2017.

23. Fernandez-Torron R, James M, Diaz Manera J, et ál.

"Functional outcome measures and muscle MRI pattern recognition in dysferlinopathy: The JAIN COS Study".
"EUROPEAN JOURNAL OF NEUROLOGY". 24 (S1)1:96-97 JUL 2017
3rd Congress of the European-Academy-of-Neurology Ubicación. Amsterdam. JUN 2017.

24. Perez-Perez J, Diaz-Manera J, Pagonabarraga J, et ál.
"Hung up knee jerk reflex in Huntington's disease: A clinical and neurophysiological study".
"MOVEMENT DISORDERS". 32 (2): 480 2017
21st International Congress of Parkinson's Disease and Movement Disorders. Vancouver. JUN 04-08, 2017.

25. Ghaoui R, Meredith J, Fernandez-Torron R, Roberto; Autoría conjunta: Jain COS Consortium.
"The international clinical outcome study in dysferlinopathy".
"JOURNAL OF NEUROLOGY NEUROSURGERY AND PSYCHIATRY". Volum: 88 Número: 5 Abstract de reunió: 46. MAY 2017
Annual Scientific Meeting of the Australian-and-New-Zealand-Association-of-Neurologists (ANZAN). Gold Coast, Australia. May 09-12, 2017.

26. Cruz S, Figueroa-Bonaparte S, Gallardo E, de Becdelievre A, Gartioux C, Allamand V, Pinol P, Rodriguez-Garcia M, Jimenez-Mallebrera C, Llauger J, Gonzalez-Rodriguez L, Cortes-Vicente E, Illa I, Diaz-Manera J.
"Bethlem myopathy phenotypes and follow up: Description of 8 patients in the mildest end of the spectrum".
"NEUROMUSCULAR DISORDERS". 26, pp. S187 - S188. 01/10/2016.
21st International Congress of the World-Muscle-Society. Granada. 2016.
ISSN 0960-8966,ISSN 1873-2364.

27. Figueroa-Bonaparte S, Llauger J, Segovia S, Belmonte I, Alejaldre A, Pedrosa I, Gallardo E, Illa I, Diaz-Manera J.
"Quantitative magnetic resonance imaging in late-onset Pompe disease: A prospective observational study of 19 patients".
"NEUROMUSCULAR DISORDERS". 26, pp. S100 - S100. 01/10/2016.
21st International Congress of the World-Muscle-Society. Granada. 2016.
ISSN 0960-8966, ISSN 1873-2364.

28. Garibaldi M, Tasca G, Diaz-Manera J, Antonini G, Pennisi E, Italian Network NLSD.
"Muscle MRI in neutral lipid storage disease (NLSD)".
"NEUROMUSCULAR DISORDERS". 26, pp. S100 - S101. 01/10/2016.
21st International Congress of the World-Muscle-Society. Granada. 2016.
ISSN 0960-8966, ISSN 1873-2364.

29. Gonzalez-Quereda L, Gallano, P, Gallardo E, Rodriguez M, Lleixa C, Straub V, Topf A, Diaz-Manera J.
"LGMD2D intrafamilial clinical heterogeneity caused by alternative splicing of SGCA gene".
"NEUROMUSCULAR DISORDERS". 26, pp. S91 - S91. 01/10/2016.
21st International Congress of the World-Muscle-Society. Granada. 2016.
ISSN 0960-8966, ISSN 1873-2364.

30. Querol L, Alba-Rovira R, Jauregui A, Devaux J, Faivre-Sarrailh C., Araque J, Rojas-Garcia R, Diaz-Manera J, Cortes-Vicente E, Nogales-Gadea G, Navas-Madronal M, Gallardo E, Illa I.
"Antibodies against peripheral nerve antigens in chronic inflammatory demyelinating polyradiculoneuropathy".
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 21 - 3, pp. 202 - 203. 01/09/2016.
Inflammatory Neuropathy Consortium and GBS 100 Centenary Symposium and Ceilidh. Glasgow. 2016.
ISSN 1085-9489, ISSN 1529-8027.

31. Diaz-Manera J, Figueroa S, Llauger J, Belmonte I, Segovia S, Suarez-Cuartin G, Mayos M, Gallardo E, Illa I, S Spanish Pompe Study Grp.

“Whole body muscle mri correlates with muscle function in patients with adult onset pompe disease”.
"JOURNAL OF THE NEUROLOGICAL SCIENCES". 357,pp. E333 - E333. 15/10/2015.
22nd World Congress of Neurology (WCN). Santiago. 2015.
ISSN 0022-510X, ISSN 1878-5883

32. Diaz-Manera J, Segovia S, Llauger J, Belmonte L, Suarez-Cuartin G, Mayos M, Alejaldre A, Figueroa-Bonaparte S, Pedrosa I, Querol L, Rojas-Garcia R, Gallardo E, Illa I, Spanish Pompe Study Grp.
“Whole body muscle MRI correlates with muscle function in patients with adult onset Pompe disease”.
"NEUROMUSCULAR DISORDERS". 25, pp. S188 - S188. 01/10/2015.
20th International Congress of the World-Muscle-Society. Brighton. 2015.
ISSN 0960-8966, ISSN 1873-2364.

33. Gallardo E, Suarez-Calvet X, De Luna N, Rojas-Garcia R, Diaz-Manera J, Illa I.
“Role of hypoxia in innate immunity activation in dermatomyositis”.
"NEUROMUSCULAR DISORDERS". 25, pp. S246 - S247. 01/10/2015.
20th International Congress of the World-Muscle-Society. Brighton. 2015.
ISSN 0960-8966, ISSN 1873-236.

34. Querol Gutierrez L A, Rojas-Garcia R, Diaz-Manera J, Barcena J, Pardo Fernandez J, Ortega-Moreno A, Sedano M J, Sero-Ballesteros L, Carvajal A, Ortiz N, Gallardo E, Illa I.
“Rituximab is effective in treatment-resistant CIDP with IgG4 antibodies against paranodal proteins”.
"EUROPEAN JOURNAL OF NEUROLOGY". 22, pp. 39 - 39. 01/06/2015.
1st Congress of the European-Academy-of-Neurology. Berlin, 2015.
ISSN 1351-5101, ISSN 1468-1331.

35. Tasca G, Monforte M, Brisca G, D'Amico A, Diaz-Manera J, Maggi L, Al Shaik N, Pichiecchio A, Berardinelli A, Lokken N, Munell F, Sanchez A, Morandi L, Voermans N, Dastgir J, Walter M C, Boennemann C, Straub V, Quijano-Roy S, Carlier R, Vissing J, Mercuri E, Muntoni F, Ricci E,
Bertini E, Udd B, Bruno C.
“Muscle MRI in sarcoglycanopathies”.
"EUROPEAN JOURNAL OF NEUROLOGY". 22, pp. 206 - 206. 01/06/2015.
1st Congress of the European-Academy-of-Neurology. Berlin, 2015.
ISSN 1351-5101, ISSN 1468-133.

36. Querol Gutierrez L A, Rojas-Garcia R, Diaz-Manera J, Barcena J, Pardo Fernandez J, Ortega-Moreno A, Sedano M J, Sero-Ballesteros L, Carvajal A, Ortiz N, Gallardo E, Illa I.
“Rituximab is effective in treatment-resistant cidp with igg4 antibodies against paranodal proteins”.
Biennial Meeting of the Peripheral-Nerve-Society. Quebec. 2015.
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 20 - 2, pp. 214 - 215.
01/06/2015.
ISSN 1085-9489, ISSN 1529-8027.

37. Diaz-Manera J, Alejaldre A, Gonzalez L, Rojas-Garcia R, Olive M, Llauger J, Gallardo E, Gonzalez-Quereda L, Carbonell P, Marquez C, Muelas N, Vilchez J J, Fernandez-Torron R, Lopez de Munain A, Illa I.
“Muscle MRI and CT help to differentiate between mutations in emerin and lamin A/C gene in patients with Emery Dreifuss clinical phenotypes”.
NEUROMUSCULAR DISORDERS". 24 - 9-10, pp. 844 - 844. 01/10/2014.
19th International Congress of the World-Muscle-Society. Berlin. 2014
ISSN 0960-8966, ISSN 1873-2364.

38. Gallardo E, Ankala A, Nunez-Alvarez Y, Hedge M, Diaz-Manera J, De Luna N, Pastoret A, Suelves M, Illa I.
“Genetic and epigenetic determinants of low dysferlin expression in monocytes”.
"NEUROMUSCULAR DISORDERS". 24 - 9-10, pp. 903 - 903. 01/10/2014.
19th International Congress of the World-Muscle-Society. Berlin. 2014.
ISSN 0960-8966, ISSN 1873-2364.

39. Diaz-Manera J. “MRI studies on limb girdle muscle dystrophies”.
"JOURNAL OF NEUROLOGY". 261, pp. S462 - S463. 01/05/2014.
Joint Congress of European Neurology. Istanbul. 2014.
ISSN 0340-5354, ISSN 1432-1459.

40. Diaz-Manera J. “MRI studies on limb girdle muscle dystrophies”.
"EUROPEAN JOURNAL OF NEUROLOGY". 21, pp. 715 - 716. 01/05/2014.
Joint Congress of European Neurology. Istanbul. 2014.
ISSN 1351-5101, ISSN 1468-1331.

41. Querol L, Nogales-Gadea G, Rogas-Garcia R, Diaz-Manera J, Pardo J, Ortega-Moreno A, Sedano M J, Gallardo E, Berciano J, Blesa R, Dalmau J, Illa I. “Antibodies against neurofascin-155 (NF155) in CIDP associate with disabling tremor, distal weakness and poor response to IVIg”.
"JOURNAL OF NEUROLOGY". 261, pp. S61-S61. 01/05/2014.
Joint Congress of European Neurology. Istanbul. 2014.
ISSN 0340-5354, ISSN 1432-1459.

42. Querol L, Nogales-Gadea G, Rogas-Garcia R, Diaz-Manera J, Pardo J, Ortega-Moreno A, Sedano M J, Gallardo E, Berciano J, Blesa R, Dalmau J, Illa I. “Antibodies against neurofascin-155 (NF155) in CIDP associated with disabling tremor,distal weakness and poor response to IVIg”.
"EUROPEAN JOURNAL OF NEUROLOGY". 21, pp. 83 - 83. 01/05/2014.
Joint Congress of European Neurology. Istanbul. 2014.
ISSN 1351-5101, ISSN 1468-1331.

43. Suarez-Calvet X, Gallardo E, Nogales-Gadea G, Navas M, Querol L, Diaz-Manera J, Rojas-Garcia R, Illa I.
“Dysregulation of innate immunity-related genes in Dermatomyositis”.
"NEUROMUSCULAR DISORDERS". 23 - 9-10, pp. 813 - 813. 01/10/2013.
18th International Congress of the World-Muscle-Society (WMS).
Asilomar, (CA) USA. 2013.
ISSN 0960-8966.

44. Diaz-Manera J, Gallardo E, De Luna N, Pastoret A, Navas M, Illa I.
“PDGF-BB is expressed by muscle fibers in myopathies and regulates growth and migration of satellite cells and pericytes”.
"NEUROMUSCULAR DISORDERS". 23 - 9-10, pp. 837 - 837. 01/10/2013.
18th International Congress of the World-Muscle-Society (WMS).
Asilomar, (CA) USA. 2013.
ISSN 0960-8966.

45. Querol L, Nogales-Gadea G, Rojas-Garcia R, Suarez-Calvet X, Diaz-Manera J, Gallardo E, Illa I.
“Antibodies to contactin-1 and neurofascin-155 in chronic inflammatory demyelinating polyneuropathy”.
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 18, pp. 91 - 91. 01/06/2013.
Meeting of the Peripheral-Nerve-Society. Saint Malo. 2013.
ISSN 1085-9489.

46. Suarez-Calvet X, Gallardo E, Nogales-Gadea G, Querol L, Rojas-Garcia R, Diaz-Manera J, Vidal S, Illa I.
“Persistent abnormal TLR response and the effect of IVIG on TLR7 in guillain-barre syndrome patients”.
"JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 18, pp. 111 - 111. 01/06/2013.
Meeting of the Peripheral-Nerve-Society. Saint Malo. 2013.
ISSN 1085-9489.

47. Nogales-Gadea G, Querol L, Suarez-Calvet X, Araque J, Navas M, Ortiz E, Diaz-Manera J, Gallardo E, Rojas-Garcia R, Illa I.
“Autoantibody heterogeneity in chronic inflammatory demyelinating polyneuropathy”.
"JOURNAL OF NEUROLOGY". 260, pp. S55 - S56. 01/06/2013.

ISSN 0340-5354, ISSN 1432-1459.

48. Juan-Mateu J, Verdura E, Rodriguez M J, Gonzalez-Quereda L, Colomer J, Diaz-Manera J, Gallardo E, Gonzalez-Mera L, Macaya A, Munell F, Nascimento A, Navarro C, Olive M, Pascual J, Pou A, Rivas E, Roig M, Baiget M, Gallano P, Madruga M, Jimenez-Mallebrera C.
"Dystrophinopathy in manifesting female carriers: Clinical and genetic characterization in a cohort of 20 patients".
"NEUROMUSCULAR DISORDERS". 21 - 9-10, pp. 646 - 647. 01/10/2011.
16th International Congress of the World-Muscle-Society. Algarve. 2011
ISSN 0960-8966.

49. Hankiewicz K, Rojas-Garcia R, Diaz-Manera J, Gallardo E, Llauger J, Gallano P, Illa I.
"Caveolinopathy: Further clinical heterogeneity".
"NEUROMUSCULAR DISORDERS". 21 - 9-10, pp. 669 - 669. 01/10/2011.
16th International Congress of the World-Muscle-Society. Algarve. 2011.
ISSN 0960-8966.

50. Diaz-Manera J, Touvier T, Dellavalle A, Tonlorenzi R, Gallardo E, 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".
"EUROPEAN JOURNAL OF NEUROLOGY". 17, pp. 656 - 656. 01/09/2010.
14th Congress of European-Federation-of-Neurological-Societies. Geneve. 2010.
ISSN 1351-5101.

51. Diaz-Manera Ji, Touvier T, Tonlorenzi R, Perani L, Dellavalle A, Messina G, Pessina P, Gallardo E, Illa I, Torrente Y, Cossu G.
"Adult Murine Derived Mesoangioblasts Successfully Recovered Dysferlin Expression in a Murine Model of Dysferlinopathy".
"NEUROLOGY". 74 - 9, pp. A284 - A284. 02/03/2010.
62nd Annual Meeting of the American-Academy-of-Neurology. Toronto. 2010.
ISSN 0028-3878.

52. Illa I, Llauger J, Rojas-Garcia R, Diaz-Manera J, Martinez-Hernandez E, De Luna N, Gallardo E, Marquez C, Fernandez A, Paradas C.
"Shaping the Phenotypes of Dysferlin Myopathies According to a Clinical and Muscle Imaging Follow-Up".
"NEUROLOGY". 72 - 11, pp. A305 - A305. 17/03/2009.
61st Annual Meeting of American-Academy-of-Neurology. Seattle. 2009.
ISSN 0028-3878

53. De Luna N, Gallardo E, Rojas-Garcia R, Dominguez-Perles R, Diaz-Manera J, De La Torre C, Gallano P, Illa I.
"Quantification of dysferlin in monocytes: A useful tool for the detection of patients and carriers of dysferlinopathy".
"NEUROMUSCULAR DISORDERS". 18 - 9-10, pp. 790 - 791. 01/10/2008.
13th International Congress of the World-Muscle-Society. Newcastle upon Tyne. 2008.
ISSN 0960-8966.

54. Dominguez-Perles R, De Luna N, De la Torre C, Cooper S T, Robles-Cedeno R, Rojas-Garcia R, Diaz-Manera J A, Illa I, Gallardo E.
"Increased lysosomes trafficking and poly(ADP-ribose)polymerase-1 expression in dysferlin myopathy: Implications in muscle fiber necrosis".
"NEUROMUSCULAR DISORDERS". 18 - 9-10, pp. 792 - 792. 01/10/2008.
13th International Congress of the World-Muscle-Society. Newcastle upon Tyne. 2008.
ISSN 0960-8966.

55. Diaz Manera J, Rojas-Garcia R, Pradas J, Juarez C, Gallardo E, Illa I. "Seronegative generalized myasthenia gravis: clinical analysis and response to treatment in a series of 15 patients".
"EUROPEAN JOURNAL OF NEUROLOGY". 15, pp. 152 - 152. 01/08/2008.
12th Congress of the European-Federation-of-Neurological-Societies. Madrid. 2008.

ISSN 1351-5101.

56. Rojas-Garcia R, De Luna Salva N, Gallardo E, Povedano M, Bruna J, Fages E, Valldeoriola F, Carvajal A, Espinal J, Diaz-Manera J, Juarez C, Illa I. “Bulbar involvement in patients with antiganglioside antibodies against anti-neunac (alpha 2-3)Gal”. "JOURNAL OF THE PERIPHERAL NERVOUS SYSTEM". 13 - 2, pp. 180 - 181. 01/06/2008.
Inflammatory Neuropathy Consortium Meeting. Paris. 2008.
ISSN 1085-9489.

57. Illa I, Diaz-Manera J, Rojas-Garcia R, Pradas J, Rey A, Blesa R, Juarez C, Gallardo E. “Rituximab in refractory myasthenia gravis: A follow-up study of patients with antil-MuSK or anti-MuSK antibodies”.
"NEUROLOGY". 70 - 11, pp. A301 - A301. 11/03/2008.
60th Annual Meeting of the American-Academy-of-Neurology. Chicago. 2008.
ISSN 0028-3878, ISSN 1526-632X.

58. Diaz-Manera J, Rojas-Garcia R, Juarez C, Pradas J, Gallardo E, Illa I. “Are immunosupresors as effective in MuSK plus MG as in AChR plus MG patients?: Evaluation of 150 myasthenic patients treated with the same protocol”.
"NEUROLOGY". 68 - 12, pp. A300 - A300. 20/03/2007.
59th Annual Meeting of the American-Academy-of-Neurology. Boston. 2007.
ISSN 0028-3878.

59. Diaz-Manera J, Juarez C, Martinez-Ramirez S, Rojas-Garcia R, Gallardo E, Dalmau J, Illa I.
“Severe muscle atrophy and respiratory failure requiring intubation in a 45-year-old woman”.
"EUROPEAN JOURNAL OF NEUROLOGY". 13, pp. 304 - 304. 01/09/2006.
ISSN 1351-5101.

60. Diaz-Manera J, Juarez C, Martinez-Ramirez S, Rojas-Garcia R, Gallardo E, Dalmau J, Illa I.
“High titers of AChR, MuSK and VGKC-Abs in one patient: Clinical and immunological evaluation and response to rituximab therapy”.
"NEUROLOGY". 66 - 5, pp. A58 - A58. 14/03/2006.
58th Annual Meeting of the American-Academy-of-Neurology. San Diego. 2006.
ISSN 0028-3878.

61. Marti-Fabregas J, Cocho D, Belvis R, Borrell M, Montaner J, Castellanos M, Pagonabarraga J, Aleu A, Diaz-Manera J, Molina-Porcel L, Martinez-Lage M, Martinez-Domeno A, Davalos A, Jose AS, Fontcuberta J, Marti-Vilalta JL. “Baseline profile of coagulation and fibrinolysis predict the probability of recanalization in patients treated with intravenous rtPA”.
"STROKE". 36 - 2, pp. 445 - 445. 01/02/2005.
30th International Stroke Conference. New Orleans. 2005.
ISSN 0039-2499.

62. Pagonabarraga J, Diaz-Manera J, Gironell A, Pascual-Sedano B, Kulisevsky J. “Isolated tongue tremor: Three patients, three etiologies”.
"MOVEMENT DISORDERS". 20, pp. S167 - S167. 01/01/2005.
9th International Congress of Parkinsons Disease and Movement Disorders. New Orleans. 2005.
ISSN 0885-3185.

63. Belvi R, Marti-Fabregas J, Cocho D, Santamaria A, Aleu A, Molina-Porcel L, Diaz-Manera J, Martinez-Lage M, Borrell M, Fontcuberta J, Marti-Vilalta JL.
“Diagnostic yield and therapeutic modifications following prothrombotic state studies in cryptogenic stroke”.
"STROKE". 35 - 1, pp. 302 - 302. 01/01/2004.
29th International Stroke Conference. San Diego. 2004.
ISSN 0039-2499.

## PATENTS

1. Inventors: **Jordi Diaz Manera** and Boehringer Ingelheim
   Name of the patent: Nintedanib for Use in Methods for the Treatment of Muscular Dystrophy
   European Patent: EP 17163355.5
   Country: Germany
   Date: 2017
   Institution: Boehringer Ingelheim
   Countries where it applies: Europe
   Company with the commercial rights: Boehringer Ingelheim

**E. Congressos**
**(Consigneu únicament els 10 més rellevants i especifiqueu clarament les conferències invitades)**

**Autors/ores (per ordre de signatura):**

**Títol:**

**Tipus de contribució (pòster, comunicació oral, etc.):**

**Congrés:**

**Publicació:**

**Lloc:** **Any:**

**Organisme/institució que l'organitza:**

***Nota: cal indicar correctament totes les dades demanades. No deixar cap apartat en blanc.***

1. Autors: **Díaz-Manera J**, Touvier T, Messina G, Dellavalle G, Tedesco S, Tonlorenzi R, Illa I and Cossu G.
   Títol: "Mesoangioblast transplantation improves a murine model of dysferlinopathy".
   Tipus de contribució: Comunicació oral
   Congrés: Anual Meeting of the American Academy of Neurology
   Lloc: Toronto (CAN) Any: 2010
   Organisme que organitza: American Academy of Neurology

2. Autors: **Díaz-Manera J**, Touvier T, Messina G, Dellavalle G, Tonlorenzi R, Illa I and Cossu G.
   Títol: "Mesoangioblast transplantation is useful in a murine model of dysferlinopathy".
   Tipus de contribució: Comunicació oral
   Congrés: Anual Meeting of the European Federation of Neurological Societies
   Lloc: Basilea (SWI) Any: 2010
   Organisme que organitza: European Federation of Neurological Societies

3. Autors: **Díaz-Manera J**, De Luna N, Paradas C, Iturriaga C, Gallardo E, Illa I
   Títol: "Vitamin D treatment increases Dysferlin expression in carriers of one mutation in the DYSF gene". (Conferencia invitada)
   Tipus de contribució: Comunicació oral
   Congrés: Jain Foundation Dysferlin Meeting
   Lloc: Seatle (USA) Any: 2011
   Organisme que organitza: Jain Foundation

4. Autors: **Díaz-Manera J**, Fernandez-Torron R, Llauger J and the dysferlin COS study consortium (Conferencia invitada)
   Títol: "Pattern of Muscle MRI involvement in a large cohort of patients with dysferlinopathy"
   Tipus de contribució: Comunicació oral
   Congrés: Jain Foundation Dysferlin Meeting
   Lloc: Toronto Any: 2016
   Organisme que organitza: Jain Foundation

5. Autors: **Díaz-Manera J**, Fernandez-Torron R, Llauger J and the dysferlin COS study consortium (Conferencia invitada)
   Títol: "Outcomes measures in a large International study of patients with dysferlinopathy"
   Tipus de contribució: Comunicació oral
   Congrés: Jain Foundation Dysferlin Meeting
   Lloc: Orlando Any: 2016
   Organisme que organitza: Jain Foundation

6. Autors: **Díaz-Manera J** representeing the dysferlin COS study consortium (Conferencia invitada)
   Títol: “MRI and Physiotherapy Outcome Measures in a global multi-center dysferlinopathy study”
   Tipus de contribució: Comunicació oral
   Congrés: New Directions on Biology and Disease of Skeletal muscle
   Lloc: Orlando  Any: 2016
   Organisme que organitza: University of Florida

7. Autors: **Díaz-Manera J** representing the dysferlin COS study consortium  (Conferencia invitada)
   Títol: “Clinical Outcome Study for Dysferlinopathy: Moving towards Trial Readiness”
   Tipus de contribució: Comunicació oral
   Congrés: Muscle Study Group Meeting
   Lloc: Salt Lake City  Any: 2017
   Organisme que organitza: Muscle Study Group

8. Autors: **Díaz-Manera J** (Conferencia invitada)
   Títol: “Muscle MRI in muscle dystrophies produced by laminopathies”
   Tipus de contribució: Comunicació oral
   Congrés: MYO-MRI Meeting
   Lloc: Malaga (Spain)  Any: 2015
   Organisme que organitza: MYO-MRI Consortium

9. Autors: **Díaz-Manera J** (Conferencia invitada)
   Títol: “Clinical evaluations in patients with Pompe disease”
   Tipus de contribució: Comunicació oral
   Congrés: First Pompe academy
   Lloc: Barcelona (Spain)  Any: 2015
   Organisme que organitza: Sanofi-Genzyme

10. Autors: **Díaz-Manera J**, Figueroa-Bonaparte S, Segovia S, Llauger J, Belmonte I, Pedrosa I, Gallardo E, Illa I
    Títol: “Muscle MRI is useful in the follow-up of patients with late-onset Pompe disease”
    Tipus de contribució: Poster
    Congrés: Next steps in Pompe disease
    Lloc: Amsterdam (HOL)  Any: 2016
    Organisme que organitza: Sanofi-Genzyme

## SCIENTIFIC STAYS AT OTHER INSTITUTIONS

1. Institution: University of Florida, Barry Byrne Lab
   City: Gainesville, Florida
   Country: USA
   Year: 2017
   Period: 1 month
   Related to: Utility of muscle MRI in the study of Pompe disease

2. Insitution: Ospedalle San Rafaelle
   City: Milano
   Country: Italy
   Year: 2008/2009
   Duration: 1 any - 4 mesos
   Related to: Treatment of a murine model of dysferlinopathy with mesoangioblasts, a type of stem cell.

## COORDINATION OF THESIS

### THESIS ALREADY PRESENTED

1. Directors: Isabel Illa Sendra, Eduard Gallardo, **Jordi Díaz-Manera**
   Name: Utility of the muscle imaging techniques in the diagnosis of axial myopathies.
   Date of defense: November the 8th 2016
   Name of the student: Aida Alejaldre Monforte
   Institution: Universitat Autònoma de Barcelona

2. Directors: **Jordi Díaz-Manera**
   Name: A prospective study using muscle MRI in late onset Pompe disease
   Date of defense: October 26th 2018
   Name of the student: Aida Alejaldre Monforte
   Institution: Universitat Autònoma de Barcelona

3. Directors: **Jordi Díaz-Manera,** Eduard Gallardo
   Name: Role of PDGF-BB in muscle regeneration and nintedanib as a treatment for a murine model of Duchenne muscular dystrophy.
   Date of defense: November the 7th 2019
   Name of the student: Aida Alejaldre Monforte
   Institution: Universitat Autònoma de Barcelona

### THESIS IN PROCESS:

4. Directors: **Jordi Díaz-Manera,** Noemi de Luna
   Name: PDGF as a biomarker of muscle disorders and new treatment for muscle fibrosis.
   Name of the student: Esther Fernández-Simón
   Institució: Universitat Autònoma de Barcelona

5. Directors: **Jordi Díaz-Manera,** Eduard Gallardo
   Name: Biomarkers for Pompe disease
   Name of the student: Ana Carrasco Rozas
   Institució: Universitat Autònoma de Barcelona

6. Directors: **Jordi Díaz-Manera,** Elena Valassi
   Name: Utility of muscle MRI in the diagnostic and follow-up of patients with muscle dystrophies
   Name of the student: Alicia Alonso
   Institució: Universitat Autònoma de Barcelona

7. Directors: **Jordi Díaz-Manera**
   Name: Muscle MRI in the follow-up of Pompe disease
   Name of the student: Claudia Nuñez-Peralta
   Institució: Universitat Autònoma de Barcelona

## POST-DEGREE TEACHING COURSES

1. Activity category: Master
   Name: Treatment of patients with critical illness
   Years: 2007-2008
   Number of hours: 1,5 hours
   Institution: Universitat de Barcelona – Hospital Clinic

2. Activity category: Master
   Name: Muscle regeneration in muscle dyatrophies
   Years: 2011-2018
   Number of hours: 2 hours
   Institution: Universidad de Asturias

3. Activity category: European Teaching course
   Name: Summer school on Muscle MRI
   Years: 2015-2017
   Number of hours: 3 hours
   Institution: MYO-MRI Action Cost – Salpetriere Hospital Paris (FRA)

4. Activity category: Summer School Universitat de Barcelona
   Name: Rares diseases: from the diagnostic to the cure
   Years: 2016 and 2019
   Number of hours: 1 hour
   Institution: Universitat de Barcelona

5. Activity category: Teaching course
   Name: Teaching course on neuromuscular disorders
   Years: 2012-2019
   Number of hours: 5 hours
   Institution: Hospital de la Santa Creu I Sant Pau

6. Activity category: Teaching course
   Name: Pompe disease: 2 days course
   Years: 2017-2018
   Number of hours: 12 hours
   Institution: Hospital de la Santa Creu I Sant Pau – Sanofi-Genzyme

PERSONAL GRANTS FOR SCIENTIFIC PURPOSES

1. Aim: Formation stay at Ospedalle San Rafaelle de Milan
   Funding Entity: BAE grant from the Spanisg MInistry of Health 2008-2009
   Duration: 2008-2009
   Institution: Ospedalle San Rafaelle de Milan

2. Aim: Formation stay at Ospedalle San Rafaelle de Milan
   Funding Entity: Young fellowship grant. European Federation of Neurological Societies (EFNS)
   Duration: 2008
   Institution: Ospedalle San Rafaelle de Milan

3. Aim: Grant for university student to collaborate with a department
   Funding Entity: Spanish moinistry of education
   Duration: 1999-2000
   Institution: Universitat Rovira I Virgili

**Other contribution of scientific interest**

**Advisory or scientific committees:**

1. Entitity: Catalan Society of Neurology
   Role: Member of the board
   Institution category: Public research institution
   City: Barcelona, Spain
   Period: 2017-2019

2. Entitity: Spanish Society of Neurology
   Role: Coordination of the Neuromuscular disorders study group
   Institution category: Public research institution
   City: Barcelona, Spain
   Period: 2012-2014

3. Entitity: ASEM (Spanish Association of patients with neuromuscular disorders)
   Role: Scientific committee
   Institution category: Patients association
   City: Barcelona, Spain
   Period: 2010-

4. Entitity: Spanish Association of patients with glycogenosis
   Role: Scientific committee
   Institution category: Patients association
   City: Barcelona, Spain
   Period: 2013-

5. Entitity: Duchenne parent project Spain
   Role: Scientific committee
   Institution category: Patients association
   City: Barcelona, Spain
   Period: 2014-

6. Entitity: Fundacion Isabel Gemio
   Role: Scientific committee
   Institution category: Patients association
   City: Barcelona, Spain
   Period: 2014-

7. Entitity: Sanofi-Genzyme Spain
   Role: External advisory board
   Institution category: Pharma company
   City: Barcelona, Spain
   Period: 2015-

8. Entitity: Sarepta
   Role: External advisory board
   Institution category: Pharma company
   City: Barcelona, Spain
   Period: 2019-

**Organization of teaching courses:**

1. Name: First Pompe day - Barcelona
   Type of activity: Scientific meeting for patients with pompe disease and neurologists
   Insitution: Unitat de Malalties Neuromusculars, Hospital Sant Pau
   City: Barcelona

Dates: April 2019

2. Name: Pompe disease: 2 days of inmmersion in a specialized center
   Type of activity: Teaching course for Spanish neurologists
   Insitution: Unitat de Malalties Neuromusculars, Hospital Sant Pau
   City: Barcelona
   Dates: December 2018

3. Name: Spring meeting of the Neuromuscular disorders study group
   Type of activity: Scientific congress
   Insitution: Spanish society of neurology
   City: Barcelona
   Dates: February 2018

4. Name: Pompe disease: 2 days of inmmersion in a specialized center
   Type of activity: Teaching course for Spanish neurologists
   Insitution: Unitat de Malalties Neuromusculars, Hospital Sant Pau
   City: Barcelona
   Dates: October 2017

5. Name: What do we know about hereditary myopathies
   Type of activity: Scientific meeting for patients
   Insitution: Unitat de Malalties Neuromusculars, Hospital Sant Pau
   City: Barcelona
   Dates: May 2016

6. Name: Spring meeting of the Neuromuscular disorders study group
   Type of activity: Scientific congress
   Insitution: Spanish society of neurology
   City: Oviedo
   Dates: May 2014

7. Name: Spring meeting of the Neuromuscular disorders study group
   Type of activity: Scientific congress
   Insitution: Spanish society of neurology
   City: Valencia
   Dates: April 2013

**Scientific Prizes**

1. Best oral communication in neuromuscular disorders. Annual meeting of the Spanish society of Neurology 2016

2. Scietific prize for research in neuromuscular disorders. Spanish society of neurology 2015

3. Best oral communication in neuromuscular disorders. Annual meeting of the Spanish society of Neurology 2011

4. Best oral communication in neuromuscular disorders. Annual meeting of the Spanish society of Neurology 2010

**Invited conferences**

1. “Muscle MRI as an outcome measure”. World Muscle Society meeting, 2019
2. “Muscle MRI as an outcome measure”. Annual meeting of the AFM, 2019
3. “Muscle glycogenosis”. Annual meeting of the Spanish society of Neurology 2017
4. “Recent advances in Pompe disease” I Meeting of Hereditary myopathies Hospital 12 de Octubre. Madrid, 2017
5. “New treatments for inflammatory myopathies”. Annual meeting of the Spanish society of Neurology 2013
6. “Utility of stem cell therapies for muscular dystrophies” Annual meeting of the Spanish society of Neurology 2012
7. “Muscle MRI in muscle dystrophies”. Annual Meeting of the European Federation of Neurological Societies. Estambul, 2014
8. “Muscle MRI in Pompe disease”. Jornadas Iberoamericamas de la Enfermedad de Pompe. Buenos Aires, 2016
9. “Differential diagnosis of Pompe disease”, Jornada Iberia-LATAM. Madrid, 2015