



Simposio Internacional: Enfermedades neuromusculares: es el tiempo para el tratamiento

International Symposium: Neuromuscular diseases: It's time for treatment

Madrid, 15 y 16 de noviembre de 2012

Madrid, November 15-16, 2012

Isabel Illa

A. Positions and Honors (Chronological order)

Positions and Employment

Post-Graduate Training:

- 1976-1981 **Resident.** Department of Neurology
Hospital de la Sta. Creu i Sant Pau Universitat Autònoma de Medicina,
Barcelona, Spain
- 1981-1983 **Fellow.** Neuromuscular Diseases
Hospital de la Sta. Creu i Sant Pau and Hospital Vall d'Hebron
Barcelona, Spain

Clinical Appointments:

- 1983-present **Attending Physician**
Department of Neurology. Section Neuromuscular Diseases
Hospital de la Sta. Creu i Sant Pau . Barcelona, Spain
- 1993-present **Chief Neuromuscular Diseases Section** Neurology Department
Hospital Santa Creu i Sant Pau. Barcelona, Spain
- 8/93; 3/94; 7/95 **Invited Scientist**
National Institutes of Health, NDS, NINDS
Bethesda, Maryland
- 8/97 Invited Scientist. Mayo Clinic Rochester, Minnesota

Academic Appointments:

- 1989-1992 **Visiting Associate.** National Institutes of Health
Neuromuscular Diseases Unit NINDS Bethesda, Maryland
- 1992-1996 **Assistant Professor.** Autonomous University of Barcelona
Barcelona, Spain
- 1996- 2005 **Professor of Neurology** Autonomous University of Barcelona
Barcelona, Spain
- 2005- **Chair of Neurology** Autonomous University of Barcelona
Barcelona, Spain

Honors and Awards

- 1991- 1993 Councillor of the Spanish Society of Neurology
- 1993- 1995 Vice- President, Spanish Society of Neurology
- 2000- 2002 President, Spanish Society of Neurology
- 2003-2009 Management Committee, European Federation Neurological
Societies.
- 2011-2014 Management Committee, European Federation Neurological
Societies
- 2009- present Vicepresident CIBERNED



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2011- present President Committee for grants on Neurology and Psiquiatry
Instituto Salud Carlos III

B. Selected peer-reviewed publications (Chronological order - last 3 years)

0. Luis Querol*, Gisela Nogales-Gadea PhD*, Ricard Rojas-García , Eugenia Martínez-Hernández, Jordi Díaz-Manera Xavier Suárez-Calvet, Miquel Navas, Josefa Araque, Eduard Gallardo, Isabel Illa. Antibodies to contactin-1 in chronic inflammatory demyelinating polyneuropathy. *Ann. Neurol* 2012 (Accepted September 2012)
1. Luna ND, Díaz-Manera J, Paradas C, Iturriaga C, Rojas-García R, Araque J, Genebriera M, Gich I, Illa I, Gallardo E. $1\alpha,25(\text{OH})_2$ -Vitamin D3 Increases Dysferlin Expression in vitro and in a Human Clinical Trial. *Mol Ther.* 2012 Aug 21. doi: 10.1038/mt.2012.156. [Epub ahead of print]
2. Díaz-Manera J, Gallardo E, de Luna N, Navas M, Soria L, Garibaldi M, Rojas-García R, Tonlorenzi R, Cossu G, Illa I. The increase of pericyte population in human neuromuscular disorders supports their role in muscle regeneration in vivo. *J Pathol.* 2012 Jul 30. doi: 10.1002/path.4083. [Epub ahead of print]
3. Díaz-Manera J, Rojas García R, Illa I. Treatment strategies for myasthenia gravis: an update. *Expert Opin Pharmacother.* 2012 Sep;13(13):1873-83. Epub 2012 Jul 9.
4. Rojas-García R, Querol L, Gallardo E, De Luna N, Juarez C, Garces M, Fages E, Casanovas C, Illa I. Clinical and serological features of acute sensory ataxic neuropathy with antiganglioside antibodies. *J Peripher Nerv Syst.* 2012 Jun;17(2):158-68.
5. Vanhoutte EK, Faber CG, Merkies IS; PeriNomS Study Group. MRC sum-score in the ICU: good reliability does not necessarily reflect "true reliability". *Muscle Nerve.* 2012 May;45(5):767-8
6. Barba-Romero MA, Barrot E, Bautista-Lorite J, Gutierrez-Rivas E, Illa I, Jimenez LM, Ley-Martos M, Lopez de Munain A, Pardo J, Pascual-Pascual SI, Perez-Lopez J, Solera J, Vilchez-Padilla JJ. Clinical guidelines for late-onset Pompe disease. *Rev Neurol.* 2012 Apr 16;54(8):497-507.
7. Díaz-Manera J, Martínez-Hernández E, Querol L, Klooster R, Rojas-García R, Suárez-Calvet X, Muñoz-Blanco JL, Mazia C, Straasheijm KR, Gallardo E, Juárez C, Verschuuren JJ, Illa I. Long-lasting treatment effect of rituximab in MuSK myasthenia. *Neurology.* 2012 Jan 17;78(3):189-93.
8. Gallardo E, de Luna N, Diaz-Manera J, Rojas-García R, Gonzalez-Quereda L, Flix B, de Morrée A, van der Maarel S, Illa I. Comparison of dysferlin expression in human skeletal muscle with that in monocytes for the diagnosis of dysferlin myopathy. *PLoS One.* 2011;6(12):e29061.
9. Vanhoutte EK, Faber CG, van Nes SI, Jacobs BC, van Doorn PA, van Koningsveld R, Cornblath DR, van der Kooi AJ, Cats EA, van den Berg LH, Notermans NC, van der Pol WL, Hermans MC, van der Beek NA, Gorson KC, Eurelings M, Engelsman J, Boot H, Meijer RJ, Lauria G, Tennant A, Merkies IS; PeriNomS Study Group. Modifying the Medical Research Council grading system through Rasch analyses. *Brain.* 2012 May;135(Pt 5):1639-49.
10. Resmini E, Morte B, Sorianello E, Gallardo E, de Luna N, Illa I, Zorzano A, Bernal J, Webb SM.



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Identification of novel GH-regulated genes in C2C12 cells. *Horm Metab Res.* 2011 Dec;43(13):919-30.

11. Berciano J, Sevilla T, Casasnovas C, Sivera R, Vélchez JJ, Infante J, Ramón C, Pelayo-Negro AL, Illa I; Guidelines for molecular diagnosis of Charcot-Marie-Tooth disease. Programa 3 (Enfermedades Neuromusculares) del Centro de Investigación Biomédica en Red de Enfermedades Neurodegenerativas (CIBERNED) del Instituto de Salud Carlos III. *Neurologia.* 2012 Apr;27(3):169-78.

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

13. Krahn M, Illa I, Lévy N, Bushby K. 172nd ENMC International Workshop: dysferlinopathies 29-31 January 2010, Naarden, The Netherlands. *Neuromuscul Disord.* 2011 Jul;21(7):503-12.

14. Peters MJ, van Nes SI, Vanhoutte EK, Bakkers M, van Doorn PA, Merkies IS, Faber CG; PeriNomS Study group. Revised normative values for grip strength with the Jamar dynamometer. *J Peripher Nerv Syst.* 2011 Mar;16(1):47-50.

15. Gallardo E, Saenz A, Illa I. Limb-girdle muscular dystrophy 2A. *Handb Clin Neurol.* 2011;101:97-110.

16. Lancaster E, Huijbers MG, Bar V, Boronat A, Wong A, Martinez-Hernandez E, Wilson C, Jacobs D, Lai M, Walker RW, Graus F, Bataller L, Illa I, Markx S, Strauss KA, Peles E, Scherer SS, Dalmau J.

Investigations of caspr2, an autoantigen of encephalitis and neuromyotonia. *Ann Neurol.* 2011 Feb;69(2):303-11.

17. Díaz-Manera J, Touvier T, Dellavalle A, Tonlorenzi R, Tedesco FS, Messina G, Meregalli M, Navarro C, Perani L, Bonfanti C, Illa I, Torrente Y, Cossu G. Partial dysferlin reconstitution by adult murine mesoangioblasts is sufficient for full functional recovery in a murine model of dysferlinopathy. *Cell Death Dis.* 2010 Aug 5;1:e61.

18. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society guideline on management of multifocal motor neuropathy. Report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society--first revision. *J Peripher Nerv Syst.* 2010 Dec;15(4):295-301.

19. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society Guideline on management of paraproteinemic demyelinating neuropathies. Report of a Joint Task Force of the European Federation of Neurological Societies and the Peripheral Nerve Society--first revision. *J Peripher Nerv Syst.* 2010 Sep;15(3):185-95.

20. Pinós T, Marotta M, Gallardo E, Illa I, Díaz-Manera J, Gonzalez-Vioque E, García-Arumí E, Andreu AL, Martí R. A novel mutation in the mitochondrial tRNA(Ala) gene (m.5636T>C) in a patient with progressive external ophthalmoplegia. *Mitochondrion.* 2011 Jan;11(1):228-33.

21. Hernández S, Texidó L, Calderó J, Ciutat D, Piedrafita L, Casanovas A, Blasi J, Solsona C, Povedano M, Rojas R, Illa I, Caress J, Prevette D, Oppenheim RW, Milligan C, Esquerda JE.



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Increased intramuscular nerve branching and inhibition of programmed cell death of chick embryo motoneurons by immunoglobulins from patients with motoneuron disease. *J Neuroimmunol.* 2010 Dec 15;229(1-2):157-68.

22. Paradas C, Llauger J, Diaz-Manera J, Rojas-García R, De Luna N, Iturriaga C, Márquez C, Usón M, Hankiewicz K, Gallardo E, Illa I. Redefining dysferlinopathy phenotypes based on clinical findings and muscle imaging studies. *Neurology.* 2010 Jul 27;75(4):316-23.

23. De Luna N, Gallardo E, Sonnet C, Chazaud B, Dominguez-Perles R, Suarez-Calvet X, Gherardi RK, Illa I.

Role of thrombospondin 1 in macrophage inflammation in dysferlin myopathy. *J Neuropathol Exp Neurol.* 2010 Jun;69(6):643-53.

24. Van den Bergh PY, Hadden RD, Bouche P, Cornblath DR, Hahn A, Illa I, Koski CL, Léger JM, Nobile-Orazio E, Pollard J, Sommer C, van Doorn PA, van Schaik IN; European Federation of Neurological Societies; Peripheral Nerve Society. European Federation of Neurological Societies/Peripheral Nerve Society guideline on management of chronic inflammatory demyelinating polyradiculoneuropathy: report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society - first revision. *Eur J Neurol.* 2010 Mar;17(3):356-63.

25. Halter J, Schüpbach WM, Casali C, Elhasid R, Fay K, Hammans S, Illa I, Kappeler L, Krähenbühl S, Lehmann T, Mandel H, Marti R, Mattle H, Orchard K, Savage D, Sue CM, Valcarcel D, Gratwohl A, Hirano M.

Allogeneic hematopoietic SCT as treatment option for patients with mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): a consensus conference proposal for a standardized approach. *Bone Marrow Transplant.* 2011 Mar;46(3):330-7.

26. Joint Task Force of the EFNS and the PNS. European Federation of Neurological Societies/Peripheral Nerve Society Guideline on management of chronic inflammatory demyelinating polyradiculoneuropathy: report of a joint task force of the European Federation of Neurological Societies and the Peripheral Nerve Society--First Revision. *J Peripher Nerv Syst.* 2010 Mar;15(1):1-9. Erratum in: *J Peripher Nerv Syst.* 2010 Dec;15(4):373.

27. Skeie GO, Apostolski S, Evoli A, Gilhus NE, Illa I, Harms L, Hilton-Jones D, Melms A, Verschuuren J, Horge HW; European Federation of Neurological Societies. Guidelines for treatment of autoimmune neuromuscular transmission disorders. *Eur J Neurol.* 2010 Jul;17(7):893-902.

28. Rojas-Garcia R, Gallardo E, De Luna N, Juarez C, Martinez-Hernandez E, Carvajal A, Casasnovas C, Fages E, Davila-González P, Illa I. Bulbar involvement in patients with antiganglioside antibodies against NeuNAc(alpha2-3)Gal. *J Neurol Neurosurg Psychiatry.* 2010 Jun;81(6):623-8.

C. Editorial activities

Reviewer for:
Annals of Neurology
Neurology
Lancet Neurology



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European Journal of Neurology

PLoS ONE

Archives of Neurology

J Neurol Neurosurg Psychiatry

D. Professional activities

Member of the evaluating committee of the 2011 E-RARES call.

President of the evaluating committee of the 2006, 2007, 2008, 2009 FIS, ISCIII Ministry of Health projects on Neurology call.

President of the evaluating committee of the 2013 FIS, ISCIII Ministry of Health projects on Neurology call.

On-line reviewer for several AFM calls

Member of the scientific committee for the research laboratories located at the Henri Mondor hospital in Créteil, France.

Member of the scientific committee of the Instituto de Investigación IBIS, Sevilla, Spain.

E. Patents

RH Brown, MF Ho, I Illa, E Gallardo

Blood-Based Assay for Dysferlinopathies

REGISTRY N° : 7,172,858

6 de Febrero de 2007

Massachusetts General Hospital, Boston/ Hospital Sta. Creu i Sant Pau, Barcelona

COUNTRIES: : USA, Canadá and Japon

F. Citation Report.

Results : 224

Sum of the times cited : 5008

Citing articles :3998

Citing articles without self-citation: 3911

H-index: 22