



**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

## Susana Quijano-Roy



Position: Full-time Pediatric neurologist at the Pediatric Garches Neuromuscular Center, France.

1999 -Inter-University Diploma of Myologie : UPMC (Paris VI) (Pr Fardeau)  
1999 ECFMG – FMGEMS, Certification for Foreign Medical Graduates at the United States

2001- EMG and Neuromuscular Fellowship Program, HARVARD and TUFTS University, Boston, MA (USA)

2004- Ph-Degree (European doctoral Science Thesis)

Pediatric Neurologist expert in Diagnosis and treatment of Neuromuscular Disorders, in particular early onset diseases with orthopaedic and respiratory severe complications (SMA, congenital muscular dystrophies (CMD) congenital myopathies and myasthenic syndromes); specialized consultation, daily hospital at R. Poincaré Hospital. Head of the pediatric EMG laboratory at Necker Enfants Hospital, Paris.

Teaching on Congenital muscular dystrophies at the *Inter-University Diploma of Myology* (Pr B Eymard, UPMC Jussieu Paris- Marseille), every year since 2003. *Summer School of Myology* (Paris), every year since 2003.

Teaching on diagnosis and respiratory, orthopaedic and global management of early onset neuromuscular disorders, and on muscle MRI at the *Summer School EVELAM* (Latin-American Myology Group), every year since 2008. Variable teaching in neuromuscular aspects in various University Diploma: Pediatric Neurology (DIU), Genetics (DEA), Physical therapists (DU) and Myology (DIU).

Clinical research: diagnosis, identification of new genes in CMDs, congenital myopathies, Ehler-danlos Sd, arthrogryposis; treatment and multidisciplinary management of early onset neuromuscular disorders. Coordinator of the French Clinical Research Group on CMD from 2002. Participant in national and international Neuromuscular Expert groups : CMD network (ENMC workshops), Pediatric EMG, Congenital Myasthenias, Laminopathies, arthrogryposis, whole-body muscle MRI, Garches brace (AFM) Attached to INSERM U974 (T. Voit), Salpêtrière, Paris. Soon to join Dr L Garcia's team (UVSQ)

### INTERNATIONAL PUBLICATIONS : (H-Index: 17)

46 publications in various international journals with an "impact factor" above 2.

Five examples :

\* Quijano-Roy S, et al. LAMA2-Related Muscular Dystrophy . In: Pagon RA, Bird TD, Dolan CR, Stephens K, Adam MP, editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. 2012 Jun 07. Available at <http://www.genetests.org>.

\* Quijano-Roy S, et al. Muscle imaging in congenital myopathies. *Semin Pediatr Neurol* 2011;18:221-9.

\* Quijano-Roy S, et al. De novo LMNA mutations cause a new form of Congenital Muscular Dystrophy (L-CMD). *Ann Neurol* 2008;64:177–186

\* Quijano-Roy S, et al. EMG and nerve conduction studies in children with congenital muscular dystrophy. *Muscle Nerve* 2004 Feb;29(2):292-9



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\*Ferreiro A, Quijano-Roy S, et al. Mutations of the selenoprotein N gene, which is implicated in rigid spine muscular dystrophy, cause the classical phenotype of multiminicore disease: reassessing the nosology of early-onset myopathies. *Am J Hum Genet* 2002 Oct;71(4):739.

REVIEWING : 2-5 reviews per year for neurology and neuromuscular journals, the most frequent: (in order of frequency) Neuromuscular Disorders; Muscle and Nerve.