J. Andoni URTIZBEREA

Born on January 24th, 1959 at St Jean de Luz, France. Married, three children.
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Degrees and Diplomas
1983 : Medical Degree, Paris University, France.
1984 : Institut d'Etudes Politiques, Paris, France.
1987 : University Degree, Tropical Medecine, Aix-Marseille, France.
1990 : certified in Paediatrics, Paris, France.
1991 : certified in Physical Medecine & Rehabilitation, Paris, France.
1993 : short genetic course, Bar Harbor, Maine, USA.
1999 : University Degree, Myology, Paris-6 university, France.

Research :
Master 2 in Sciences et Techniques adaptés au handicap, Dijon University, France.
Master 2 in Neurosciences, Paris, France.
Habilitation à diriger les recherches, Paris, France.

Research Director, European Neuromuscular Center, Baarn, The Netherlands (1999-2005)
General Secretary of the Institut de Myologie, Hopital de la Salpetriere, Paris (1997-2000)
Deputy-director, Center of Excellence for NMD (GNMH), Hendaye (2004-cont.)

Teaching activities
Ex-Professor in Physical Medecine & Rehabilitation, Versailles-St Quentin University, France
Currently : Head, Summer School of Myology, Institut de Myologie, UPMC, Paris, France.

Positions held

1993-1997 :	Medical Director, Muscular Dystrophy Association (AFM), France
2001-2003 :	Head, Department of Physical Medecine & Rehab., Garches Hospital
2004-2005 :	Head, Division of Myology, Muscular Dystrophy Association – Evry, France
2005- cont.:	Praticien Hospitalier, Hopital Marin, AP-HP, Hendaye, France
1997- cont.:	Head, Summer School of Myology, Institut de Myologie, Paris, France

Main Publications
105 publications in Pubmed.

1. Stum M, Davoine CS, Vicart S, Guillot-Noel L, Topaloglu H, Carod-Artal FJ, Kayserili H, Hentati F, Merlini L, **Urtizberea JA**, Hammouda EH, Quan PC, Fontaine B, Nicole S. Spectrum of HSPG2 (perlecan) mutations in patients with Schwartz-Jampel syndrome. *Hum Mutat,* 2006 ;27 :1082-1091.

2. Shore EM, Xu M, Feldman GJ, Fenstermacher DA, Cho TJ, Choi In, Connor JM, Delai P, Glaser DL, Le Merrer M, Morhart R, Rogers JG, Smith R, Triffit JT, **Urtizberea JA**, Zasloff, Brown MA, Kaplan FS. A recurrent mutation in the BMP type I receptor ACVR1 causes inherited and sporadic fibrodysplasia ossificans progressiva. *Nat Genet,* 2006;38:525-527.

3. **Urtizberea JA**, Bassez G, Leturcq F, Nguyen K, Krahn M, Levy N. Dysferlinopathies. *Neurol India*, 2008;56:289-97.

4. Senderek J, Garvey SM, Krieger M, Guergueltcheva V, **Urtizberea A**, Roos A, Elbracht M, Stendel C, Tournev I, Mihailova V, Feit H, Tramonte J, Hedera P, Crooks K, Bergmann C, Rudnik-Schöneborn S, Zerres K, Lochmüller H, Seboun E, Weis J, Beckmann JS, Hauser MA, Jackson CE. Autosomal-dominant distal myopathy associated with a recurrent missense mutation in the gene encoding the nuclear matrix protein, matrin 3. *Am J Hum Genet*, 2009;84:511-8.

5. Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, **Urtizberea A**, von der Hagen M, Huebner A, Palace J, Bushby K, Straub V, Beeson D, Abicht A, Lochmüller H. Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *Am J Hum Genet,* 2011 ;88:162-172.

6. Sacconi S, Féasson L, Antoine JC, Pécheux C, Bernard R, Cobo AM, Casarin A, Salviati L, Desnuelle C, **Urtizberea A**.
A novel CRYAB mutation resulting in multisystemic disease. Neuromuscul Disord, 2011 sept. (ahead of print)