Preliminary program

- Kate Bushby. University of Newcastle and Newcastle upon Tyne Hospitals Trust. Newcastle, United Kingdom. Treatment and managing of Duchenne muscular dystrophy.
- Bjarne Udd. University of Tampere Medical School. Tampere, Finland. Genetics and pathogenesis of distal muscular dystrophies.
- **Francesc Palau.** Instituto de Biomedicina, CSIC, and Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER). Valencia, Spain.  
  **Role of mitochondria in the pathogenesis of Mendelian neuromuscular disorders.**

- **Salvatore DiMauro.** Columbia University College of Physicians & Surgeons. New York, USA.  
  **Pathogenesis and treatment of mitochondrial disorders.**

- **Michael E. Shy.** Wayne State University School of Medicine. Detroit, Michigan, USA.  
  **Biology of peripheral inherited neuropathies: Schwann cell and axon cross-talking.**

- **Michael Sereda.** Max Planck Institute of Experimental Medicine and University of Göttingen. Göttingen, Germany.  
  **Experimental therapies of demyelinating neuropathies.**

- **Vincent Timmerman.** Department of Molecular Genetics, University of Antwerp, Antwerp, Belgium.  
  **Genetic and cell heterogeneity of Charcot-Marie-Tooth disease.**

- **Juan J. Vilchez.** Hospital Universitari La Fe. Valencia, Spain.  
  **Rare forms of Charcot-Marie-Tooth disease: clinical and genetic aspects.**

- **José Berciano.** Hospital Universitario Marqués de Valdecilla. Santander, Spain.  
  **Phenotype and clinical evolution of Charcot-Marie-Tooth disease type 1A.**

- **Davide Pareyson.** IRCCS Foundation, C. Besta Neurological Institute. Milano, Italy.  
  **Natural history and treatment of peripheral inherited neuropathies.**

- **Gart A. Nicholson.** Concord Hospital. Sidney, Australia.  
  **Phenotype and genotype of peripheral inherited neuropathies.**

- **James R. Lupski.** Baylor College of Medicine and Texas Children's Hospital. Houston, Texas, USA.  
  **Molecular basis of curcumin treatment in Charcot-Marie-Tooth disease.**

- **Eduardo Tizzano.** Hospital de la Santa Creu i Sant Pau and Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER). Barcelona, Spain.  
  **Proximal spinal muscular atrophy: Genotype-phenotype correlation and therapeutic perspectives.**

- **Judith Melki.** Hadassah University Hospital. Jerusalem, Israel.  
  **Inherited upper and lower motor neuron disease: insights from genetics and animal models.**
- **Hélène Puccio.** Institut de Génétique et de Biologie Moléculaire et Cellulaire. Illkirch, France.
  Pathophysiology and therapeutics of Friedreich ataxia.

- **Alexis Brice.** Institut National de la Santé et de la Recherche Médicale (INSERM). Paris-Salpêtrière, France.
  Natural history, genetics and pathogenesis of spinocerebellar ataxias.

- **Alexandra Dürr.** Institut National de la Santé et de la Recherche Médicale (INSERM). Paris-Salpêtrière, France.
  Natural history, genetics and pathogenesis of spastic paraparesias.

- **Jesús Esteban.** Hospital Universitario 12 de Octubre. Madrid, Spain.
  Genetic forms of amyotrophic lateral sclerosis.

- **Peter Nigel Leigh.** Institute of Psychiatry, King’s College London. London, United Kingdom.
  Amyotrophic lateral sclerosis: pathogenic mechanisms and therapies.
About the Symposium

Ten years ago Alan Emery wrote in the preface of *Neuromuscular Disorders: Clinical and Molecular Genetics* the following comments: “It has been estimated than more that one person in every 3000 has a serious disabling inherited neuromuscular disorder. The suffering caused by these disorders is considerable, but, until the last decade or so, virtually nothing was known of their pathogenesis. Any rationale approach to treatment was therefore out of the question. However, matters are now changing rapidly. The genes for many of these disorders have been localised and characterised and their gene products identified and studied. The detection of preclinical disease, the identification of heterozygous carriers and prenatal diagnosis are all becoming possible, and, hopefully, effective treatments may no be too far distant”. Now, ten years later, more genes associated with neuromuscular disorders have been reported, confirming the wide genetic heterogeneity of most of diseases of the peripheral nervous system. Thinking genetically has become more important and more compelling. It allows the unequivocal diagnosis of neonatal, pediatric and adult diseases whose etiology has a genetic basis, thus providing a more accurate prediction of natural history and prognosis, and reproductive planning for the family, not only offering genetic counselling and prenatal diagnosis but also preimplantational genetic diagnosis. Moreover, for a number of them molecular and cell pathogenesis is suggesting new molecular targets and, more relevant, novel therapeutic approaches are currently developing to manage and treat these disorders, including new drugs and gene and cellular therapies.

The aim of the symposium is to discuss the state-of-the-art of neuromuscular diseases as a whole, including muscular dystrophies, mitochondrial disorders, peripheral neuropathies, spinal muscular atrophy, motoneurone disease and Friedreich ataxia. We will be able to confront and compare pathogenic mechanisms and molecular targets for the different diseases, as a forum for discussion of the rational basis of the new therapeutic approaches.

Meeting Venue

The International Symposium on Rare Diseases, *Inherited Neuromuscular Diseases: Translation from Pathomechanisms to Therapies* will be held in the Santiago Grisolía Auditorium in the Museo de las Ciencias Príncipe Felipe of Valencia (Spain). The museum, designed by Valencian architect Santiago Calatrava, and with an auditorium of 2.800 square metres, provides the perfect venue for this symposium.

These emblematic projects have turned Valencia, with its heritage of over two thousand years of history, into a most modern city. The city is easily accessible by road, rail, sea and air. Its culture, cuisine and academic tradition along with its location on the Mediterranean coast and its pleasant climate, make the city the ideal setting for this symposium.

Auditorium Santiago Grisolía  
Museo de las Ciencias Príncipe Felipe  
Autovía del Saler, 7  
46013 Valencia, Spain
Call for abstracts

Participants are invited to submit abstracts for oral or poster presentation, the majority being presented as poster. Acceptance will be based upon the quality and relevance of the submissions.

Abstracts, not exceeding one page in length (width 12 cm; height 20 cm, font Times New Roman size 12), with single-spaced text, should preferably be sent by e-mail to catedrasg@cac.es, before the deadline expires on September 30th, 2008.

Each abstract must include: title (in CAPITAL LETTERS) and the names of the authors (the name of presenting author should be underlined). After the name of the last author the address of the institution or organization should be stated.

Abstracts will be reproduced in the same condition in which they were received for inclusion in conference documentation. They will not be subject to editing.

Authors of accepted abstracts will be notified from October 13th onwards.

The poster exhibition will be situated in the hall of the auditorium. It will be open throughout the meeting.

Scientific & organizing committee

Francesc Palau
Instituto de Biomedicina, CSIC, and Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER). Valencia, Spain

Santiago Grisolía

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**Symposium secretariat**

Cátedra Santiago Grisolía  
Fundación Ciudad de las Artes y las Ciencias  
Prolongación Paseo de la Alameda, 42-B, 1er piso pta. 1  
46023 Valencia, Spain.

Tel: 0034 96 197 46 70  
Fax: 0034 96 197 45 98  
E-mail: catedrasg@cac.es  
www.fundacioncac.es/catedrasg

**Travel & accommodation**

The travel agency Viajes Iberia has booked rooms in several hotels within walking distance of the venue. Information can be obtained from:

Ms. Matilde García-Conde  
Tel: 0034 96 353 61 65  
Fax: 0034 96 394 06 06  
E-mail: valencia.hernancortes23@viajesiberia.com

**Registration fee**

Up to September 30\textsuperscript{th}: 100€  
After September 30\textsuperscript{th}: 200€  
Students: 50€

Registration fee for participants includes scientific sessions, coffee breaks and the symposium material.

A certificate of attendance will be given to all registered participants.

**Payment**

Bank transfer to Fundación Ciudad de las Artes y las Ciencias at the bank BANCAJA.

- **Swift code for international bank transfers:**  
  IBAN: ES48 2077 0737 74 3100345279  
  BIC: CVALESVVXXX

- **Account number for Spanish bank transfers:**  
  2077 0737 74 3100345279
Please send a copy of the bank transfer with your name either by e-mail to catedrasg@cac.es or by fax 0034 96 197 45 98.

**Language**

The lectures will be given in English.

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Please send the registration form to the symposium secretariat by mail, fax or e-mail. You can also register online on the website: http://www.fundacioncac.es

*Note: Your registration will be only considered upon receiving the full payment.*

In compliance with Act 34/2002, dated 11th July, on services for the Information Society and Electronic Trade and the Organic Act 15/1999, dated 13th December on Protection of Personal Data, we hereby inform you that your data will be included and processed in the I.T. File of the Fundación Ciudad de las Artes y las Ciencias with the purpose of being able to send information to you regarding our services, news and activities. At any time the user can exercise the rights of access, correction, cancellation and opposition to the use of personal data by sending an e-mail to catedraac@cac.es or a fax to: 0034 96 197 45 98.