




The aim of this workshop is to develop an international consortium for the curation of all the human genetic variants associated to spastic paraparesis and motor neuron diseases, within the scope and vision of the Human Variome Project. Experts in the clinical, genetic, and cellular and informatics aspects from different countries will meet in order to discuss the challenges and delineate actions towards the construction of a coordinated mutation collection and database. These databases are of crucial importance for the research and diagnosis of genetic disorders. Clinicians and scientists worldwide interested in spastic paraparesis and motor neuron disorders are welcome to join this initiative.

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WORKING MEETING SPASTIC PARAPARESIS MUTATION DATABASE

12 -14 September 2010

Reference Center for Rare Diseases
(CREER).

Burgos. Spain.

Participants

Jorge Amigo, Santiago de Compostela, Spain

Juan Diego Arroyo, Valencia, Spain

Peter Bauer, Tübingen, Germany

Juan Bautista, Sevilla, Spain

Sara Benedetti, Milano, Italy

Corey Braastad, Worcester, MA, USA

Fabio Cancarè, Milano, Italy

Paola Carrera, Milano, Italy

Christina Divincenzo, Worcester, MA, USA

Christopher Elzinga, Worcester, MA, USA

John Fink, Ann Arbor, MI, USA

Andrés Ordóñez, Santiago de Compostela, Spain

Ignacio Pascual, Madrid, Spain

Maria-Jesús Sobrido, Santiago de Compostela, Spain

Stephan Züchner, Miami, FL

Programm

Sunday 12th

Arrival of participants

Dinner at the CREER

Monday 13th

8:00-9:00h: Breakfast at the CREER

9:00-14:00h: Morning sessions

Session I: Overview of current representation of motor neuron diseases in LSDBs, aims and scope of the proposed database.

Session II: Database fields: genetic data, reference sequences.

11:00-11:30: COFFEE BREAK

Session III: Database fields: clinical data.

Session IV: Assessment of pathogenicity.

13:30-15:00h: Lunch at the CREER

15:00-20h: Afternoon sessions

Session V: Ethical and legal aspects: declaration of intent, informed consent, ethics committee.

Session VI: Work flow and organization, data collection, entry and access of data.

17-17:30: COFFEE BREAK

Session VII: Long term sustaining, funding, invitation of others who are relevant to join, dissemination.

Session VIII: Planning specific goals, time frame, next steps.

20:00h-21:00h: Dinner at the CREER

Optional after dinner walk in Burgos

Tuesday 14th

8:00-9:00h: Breakfast at the CREER

9:00-14:00h: Morning sessions

Session IX: Structure of the database, informatics tools, hosting, flow, management and security of data.

Session X: Review of key issues, summary, conclusions.

14-15:30h: Lunch at the CREER

Departure