

IV CRG ANNUAL SYMPOSIUM

New challenges in the mechanistics of human disorders

CONNECTING THE GENOME WITH DISEASE

Friday, 2 December and Saturday, 3 December, 2005

Center for Genomic Regulation

Barcelona, Catalonia, Spain

Monogenic diseases research has provided knowledge about the molecular alterations that cause many severe human hereditary disorders. Despite this enormous progress, the genetic bases of common disorders that affect a large number of subjects are largely unsolved. In this symposium, we will evaluate some of the molecular mechanisms potentially involved in human disorders with special emphasis in neurological-related disorders. The symposium will bring together the best experts in the fields of small RNAs, epigenetics, genomic variation, and gene expression variation. We should evaluate to which extend these phenomena participate in the chain of events that lead to complex disorders.

Agenda

Friday 2, December

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| 14:00 – 14:30 | Registration |
| 14:30 – 15:45 | Welcome
Miguel Beato , Center of Genomic Regulation, Barcelona, Spain |
| 14:45 – 15:00 | Opening Comments
Xavier Estivill , Center of Genomic Regulation, Barcelona, Spain |
| SESSION 1: | Expanding trinucleotide expansions
Moderator: Jerome Cavaille |
| 15:00 – 15:30 | Jean Louis Mandel , Institut de Génétique et de Biologie Moleculaire et Cellulaire, Strasbourg, France |
| 15:30 – 16:00 | Albert R. La Spada , University of Washington, Seattle, USA |
| 16:00 – 16:30 | Gillian P. Bates , Guy's Hospital, London, UK |
| 16:30 – 17:00 | Beverly L. Davidson , University of Iowa, Iowa City, USA |
| 17:00 – 17:30 | Coffee Break |

- SESSION 2:** **Genomic variants and disease**
Moderator: Jean Louis Mandel
- 17:30 – 18:00** **James R. Lupski**, Baylor College of Medicine, Houston, USA
- 18:00 – 18:30** **Stephen W. Scherer**, Hospital for Sick Children, Toronto, Canada
- 18:30 – 19:00** **Luis Perez-Jurado**, Universitat Pompeu Fabra, Barcelona, Spain
- 19:00 – 19:30** **Elizabeth A. Lindsay**, University of Naples, Naples, Italy

Saturday, 3 December

- SESSION 3:** **The rising of the small RNAs' world**
Moderator: Bernhard Horsthemke
- 9:00 – 9:30** **Witold Filipowicz**, Friedrich Miescher Institute, Basel, Switzerland
- 9:30 – 10:00** **Oliver Hovert**, Columbia University, New York, USA
- 10:00 – 10:30** **Michel Georges**, University of Liège, Liège, Belgium
- 10:30 – 11:00** **Jerome Cavaille**, Université P. Sabatier, Toulouse, France

11:00 – 11:30 **Coffee Break**

- SESSION 4:** **Epigenetic mechanisms of genetic disease**
Moderator: Gillian P. Bates
- 11:30 – 12:00** **Bernhard Horsthemke**, Institut für Humangenetik, Universitätsklinikum Essen, Essen, Germany
- 12:00 – 12:30** **Nicholas Katsanis**, Johns Hopkins University School of Medicine, Baltimore, USA
- 12:30 – 13:00** **Manel Esteller**, Centro Nacional de Investigaciones Oncológicas, Madrid, Spain
- 13:00 – 13:30** **Peter Seeburg**, Max-Planck-Institut für medizinische Forschung, Heidelberg, Germany

13:30 – 15:00 **Lunch Break**

- SESSION 5:** **Variability in gene expression and disease**
Moderator: James R. Lupski
- 15:00 – 15:30** **Vivian G. Cheung**, Children's Hospital of Philadelphia, Philadelphia, USA
- 15:30 – 16:00** **Emmanouil Dermitzakis**, Wellcome Trust Sanger Institute, Cambridge, UK
- 16:00 – 16:30** **Takeshi Yagi**, Osaka University, Osaka, Japan
- 16:30 – 17:00** **Andrew Chess**, Massachusetts General Hospital, Harvard Medical School, Boston, USA
- 17:00 – 17:30** **Closing Remarks**, Informal Discussion, and Departure