



THE 5TH INTERNATIONAL CONFERENCE ON
UNSTABLE MICROSATELLITES & HUMAN DISEASE



PROGRAM SCHEDULE

(All Speaker and Workshop Presentations will be held in the Parque de las Ciencias)

Saturday, November 11

12:00 p.m.	Check-in	Vincci Hoteles
2:00-5:00 p.m.	Registration	Vincci Hoteles
5:30 p.m.	Buffet Dinner	Parque de las Ciencias
6:50 p.m.	Opening Comments Christopher E. Pearson, Laura Ranum, Al La Spada, Xavier Estivill	Parque de las Ciencias
	Keynote Addresses - Mechanisms of Mutation & Pathogenesis Chair: Christopher E. Pearson	Parque de las Ciencias
7:00-8:00 p.m.	Harry Orr <i>Neurodegenerative Disease, Mutations, Pathogenesis, Steps Toward Treatments</i>	
8:00-9:00 p.m.	Jan Hoeijmakers <i>DNA Repair Defects and Neurodegeneration</i>	
9:00-11:00 p.m.	Reception	

Sunday, November 12

6:45-7:30 a.m.	Breakfast Buffet	Vincci Hoteles
8:00-4:00 p.m.	Poster Set-Up (all posters will be up through to Tuesday afternoon)	Parque de las Ciencias
8:00 a.m.	Repeat Instability, DNA Replication, Repair & Epigenetics Chair and Discussion Leaders: Geneviève Gourdon & Christopher E. Pearson	Parque de las Ciencias
8:00-8:25 a.m.	Geneviève Gourdon <i>Big Jumps: Mice Can Also Do It With No Doping Drugs!</i>	
8:25-8:50 a.m.	Darren Monckton <i>Instability in Myotonic Dystrophy and Huntington's Disease</i>	
8:50-9:15 a.m.	Christopher E. Pearson <i>DNA Replication, Repair, Epigenetics and Repeat Instability.</i>	
9:15-9:45 a.m.	Coffee Break	Parque de las Ciencias
9:45-10:10 a.m.	Bé Wieringa <i>DMI, if it isn't the DNA it's the RNA, if it isn't the RNA it is the Protein</i>	
10:10-10:35 a.m.	Sherif El-Khamisy <i>DNA Single-Strand Break Repair and Human Ataxia</i>	
10:35-11:00 a.m.	Cynthia T. McMurray <i>CAG expansions arises in the process of Repairing oxidative DNA damage</i>	

11:00-11:25 a.m.	John H. Wilson <i>Modeling Repeat Instability in Human Cells and Mice</i>	
11:25-11:55 a.m.	Workshop I presentations	
11:55-12:30 p.m.	Discussion led by Geneviève Gourdon & Christopher E. Pearson	
12:30-4:00 p.m.	Free time	
4:00-4:30 p.m.	Refreshments (open bar)	Parque de las Ciencias
4:30-6:15 p.m.	Poster Session I (Even Numbers)	Parque de las Ciencias
6:15-7:30 p.m.	Buffet Dinner	Parque de las Ciencias
8:00 p.m.	Repeat Instability & Fragile Sites Chair and Discussion Leader: Tom Glover	Parque de las Ciencias
8:00-8:25 p.m.	Sergei Mirkin <i>Microsatellites and Beyond: from Replication to Instability</i>	
8:25-8:50 p.m.	Tom Glover <i>Fragile Site Instability: Lessons from Common Fragile Sites</i>	
8:50-9:15 p.m.	Catherine Freudenreich <i>Determining the Molecular Basis for Chromosome Fragility at Human Common Fragile Site FRA16D</i>	
9:15-9:40 p.m.	Kirill Lobachev <i>Gene Amplification Triggered by Secondary Structure Forming Repeats</i>	
9:40-10:20 p.m.	Workshop II presentations	
10:20-10:50 pm	Discussion led by Tom Glover	

Monday, November 13

6:45-7:30 a.m.	Breakfast Buffet	Vincci Hoteles
8:00 a.m.	Fragile X / Fragile X-associated Tremor Ataxia Syndrome Chair and Discussion Leader: Steve Warren	Parque de las Ciencias
8:00-8:25 a.m.	Steve Warren <i>Fragile X Syndrome: Molecular mechanisms and therapeutic implications</i>	
8:25-8:50 a.m.	Ben Oostra <i>CGG Instability in the Mouse FMR1 gene</i>	
8:50-9:10 a.m.	Workshop III presentations	
9:10-9:30 a.m.	Discussion led by Steve Warren	
9:30-10:00 a.m.	Coffee Break	Parque de las Ciencias
10:00 a.m.	Facioscapulohumeral Muscular Dystrophy Chair and Discussion Leader: Charles Thornton	
10:00-10:25 a.m.	Sylvère van der Maarel <i>D4Z4 Macrosatellite Repeat Instability in FSHD: Causes and Consequences</i>	

- 10:25-10:50 a.m. **Rossella Tupler**
Facioscapulohumeral Muscular Dystrophy: How Deletion of Repetitive Elements Leads to Muscular Dystrophy
- 10:50-11:00 a.m. Discussion led by Charles Thornton
- 11:00 a.m. **Polyglutamine / Polyalanine / Protein Gain-of-Function**
Chair and Discussion Leader: Al La Spada
- 11:00-11:25 a.m. **Albert La Spada**
Molecular and Cellular Basis of Spinocerebellar Type 7
- 11:25-11:50 a.m. **Jozef Gécz**
Spectrum and Molecular Pathology of ARX Gene Mutations
- 11:50-12:30 p.m. **Workshop IV** presentations
- 12:30-12:45 p.m. Discussion led by Al La Spada
- 12:45-5:00 p.m. Free Time
- 5:00-5:30 p.m. Refreshments (open bar), mingle, meet and greet Parque de las Ciencias
- 5:30-6:15 p.m. Discussion and mingle
- 6:15 -7:30 p.m. Tapas Dinner Parque de las Ciencias
- 8:00 p.m. **RNA Pathogenesis Part I: DM1 / DM2** Parque de las Ciencias
Chair and Discussion Leader: Maurice Swanson
- 8:00-8:25 p.m. **Maurice Swanson**
Impact of Dynamic Mutations on Post-Transcription Gene Regulation
- 8:25-8:50 p.m. **Thomas A. Cooper**
CELF Proteins in Myotonic Dystrophy Pathogenesis
- 8:50-9:15 p.m. **Charles Thornton**
Myotonia as a Window on the Pathogenesis of Myotonic Dystrophy
- 9:15-9:40 p.m. **Mani S. Mahadevan**
Reversible Model of RNA Toxicity and Cardiac Conduction Defects in Myotonic Dystrophy
- 9:40-10:10 p.m. **Workshop V** presentations
- 10:10-10:30 p.m. Discussion led by Maurice Swanson

Tuesday, November 14

- 6:30-7:30 a.m. Breakfast Buffet Vincci Hoteles
- 8:00 a.m. **RNA Pathogenesis Part II: DM1 / DM2 / SCA8 / SCA10 / HDL2** Parque de las Ciencias
Chair and Discussion Leader: Laura Ranum
- 8:00-8:25 a.m. **Gala Filippova**
Studies of the DM1 Locus Suggest that CTG/CAG Repeats are Associated with an RNA Mediated Chromatin Modification and Insulator Function.

8:25-8:50 a.m.	Laura Ranum <i>SCA8: Bidirectional Expression of CUG and CAG Expansion Transcripts Evidence for RNA and Protein Gain of Function Effects.</i>	
8:50-9:15 a.m.	Russell Margolis <i>Huntington's Disease-like 2: A Potential Role for RNA Toxicity in Neurodegeneration</i>	
9:15-9:45 a.m.	Coffee Break	Parque de las Ciencias
9:45-10:10 a.m.	Tetsuo Ashizawa <i>RNA-Mediated Gain of Function in SCA10</i>	
10:10-10:50 a.m.	Workshop VI presentations	
10:50-11:10 a.m.	Discussion led by Laura Ranum	
11:10-4:00 p.m.	Free Time	
4:00-4:30 p.m.	Refreshments (open bar)	Parque de las Ciencias
4:15-6:15 p.m.	Poster Session II (Odd Numbers)	Parque de las Ciencias
6:15-7:30 p.m.	Buffet Dinner	Parque de las Ciencias
8:00 p.m.	Genome Polymorphism / Friedreich's Ataxia Chair and Discussion Leader: Xavier Estivill	Parque de las Ciencias
8:00-8:25 p.m.	Xavier Estivill <i>Structural Variants of the Human Genome and Disease Susceptibility: Twins, Families and Populations</i>	
8:25-8:50 p.m.	Hélène Puccio <i>Molecular Genetics of Friedreich's Ataxia: Insights Through Mouse Models</i>	
8:50-9:15 p.m.	Sanjay Bidichandani <i>Somatic Instability as a Phenotype Determinant in Friedreich Ataxia</i>	
9:15-9:25 p.m.	Workshop VII presentations	
9:25-9:35 p.m.	Discussion led by Xavier Estivill	

Wednesday, November 15

6:45-7:30 a.m.	Breakfast Buffet	Vincci Hoteles
8:00 a.m.	Treatment Strategies Chair and Discussion Leader: David L. Nelson and Christopher E. Pearson	Parque de las Ciencias
8:00-8:25 a.m.	Joel M. Gottesfeld <i>Histone Deacetylase Inhibitors Reverse Gene Silencing Caused by Expanded GAA-TTC Repeats in Friedreich's Ataxia Histone Deacetylase In</i>	
8:25-8:50 a.m.	Gen Sobue <i>Molecular Targeted Therapy for Spinal and Bulbar Muscular Atrophy</i>	
8:50-9:15 a.m.	Wlodzimierz Krzyzosiak <i>Towards Allele-Specific Silencing of Mutant Genes Implicated Triplet Repeat Expansion Diseases.</i>	

9:15-9:35 a.m.	Discussion led by David L. Nelson and Christopher E. Pearson	
9:35-10:05 a.m.	Coffee Break	Parque de las Ciencias
10:05 a.m.	Autophagy Neurodevelopment and Neurodegeneration Chair and Discussion Leader: Al La Spada	
10:05-10:30 a.m.	Ana Maria Cuervo <i>Autophagy and Misbehaving Proteins: Who is Attacking Whom?</i>	
10:30-10:55 a.m.	David C. Rubinsztein <i>Intracytosolic Aggregate-Prone Proteins are Autophagy Substrates</i>	
10:55-11:15 a.m.	Discussion led by Al La Spada	
11:15-11:45 p.m.	Wrap-Up	
12:00	Bus to Vincci Hoteles	from Parque de las Ciencias
1:30 p.m.	Bus to Alhambra and boxed lunch	from Vincci Hoteles
3:00-5:00 p.m.	Tour of Alhambra	Alhambra
5:30 p.m.	Bus to Vincci Hoteles	from Alhambra
6:45 p.m.	Bus to Bodegas Señorío de Nevada	from Vincci Hoteles
7:00 pm	Cocktails	Bodegas Señorío de Nevada
7:30-11:00 p.m.	Closing Banquet and Flamenco Dance	Bodegas Señorío de Nevada

Thursday, November 16

6:45-9:00 a.m.	Breakfast Buffet	Vincci Hoteles
	Departure	

