

GENETICS OF INTELLECTUAL DISABILITY - AN UPDATE

Life and Health Sciences Research Institute (ICVS)
School of Health Sciences | University of Minho
Braga | Portugal

June 25-27, 2015

Thursday, June 25th 2015

- 08h45 Registration ::: ICVS/ECS hall
09h00 Welcome address ::: room G2.02
09h15 The clinical and genetic diagnosis process in intellectual disability
[Raoul Hennekam] ::: room G2.02
10h30 Coffee-break ::: room G2.01
11h00 Genetic variation and disease - an introduction
[Patricia Maciel] ::: room G2.02
12h00 Novel technical possibilities for genetic diagnosis of intellectual disability - introduction to aCGH and NGS methodologies
[Mafalda Barbosa] ::: room G2.02
13h00 Lunch ::: ICVS/ECS bar
14h30 From clinic to gene and back - the example of Fragile X
[Jean Louis Mandel] ::: room G2.02
15h30 Coffee-break with poster presentations from participants ::: ICVS/ECS hall
17h00 Gene identification in intellectual disability using genomic approaches: what have we learned about pathogenesis?
[Michael Talkowski] ::: room G2.02
18h15 Students papers/activities ::: room G2.02
Dinner at the historic center of Braga

Friday, June 26th 2015

- 09h00 Epigenetics and intellectual disability
[Michael Talkowski] ::: room G2.02
10h00 Coffee-break with poster presentations from participants / case discussion ::: ICVS/ECS hall
10h45 Hands-on introduction to Genetic Databases
[Patricia Maciel, Fátima Lopes and Mafalda Barbosa] ::: room A2.08
12h00 Genetics of cortical malformation and intellectual disability
[Jamel Chelly] ::: room G2.02
13h00 Lunch ::: ICVS/ECS bar
14h30 Finding novel genes underlying Rett syndrome-like clinical presentation
[Fátima Lopes] ::: room G2.02
15h00 Copy number variations as risk factors for neurodevelopmental disorders
[Patricia Maciel] ::: room G2.02
15h30 Mutation or not mutation, that is the question
[Jean-Louis Mandel] ::: room G2.02
16h00 Coffee-break ::: room G2.01
16h30 Round table: Translation of novel genetic approaches to the clinical context - benefits and challenges
[Jean-Louis Mandel, Mafalda Barbosa, Raoul Hennekam] ::: room G2.02
17h00 New models for study of nervous system disease mechanisms: human inducible pluripotent stem cells
[Luis Pereira de Almeida] ::: room G2.02
18h00 Round Table: Gene discovery in ID and autism spectrum disorders - challenges and approaches
[Jean-Louis Mandel, Astrid Vicente] ::: room G2.02

Saturday, June 27th 2015

- 09h00 Neuroimaging in ID research
[Adriana Sampaio] ::: room G2.02
10h00 From animal models to clinical trials in intellectual disability: the case of Rett syndrome
[Laura Ricceri] ::: room G2.02
11h00 Coffee break ::: room G2.01
11h20 From animal models to clinical trials in intellectual disability: the case of Down syndrome
[Mara Dierssen] ::: room G2.02
12h20 Closure; evaluations ::: room G2.02
12h40 Farewell