

Workshop on Genetics of Neurological Disorders

30.01.2020 | *Thursday*

8h30 Secretarial Opening and Registration

08h50 Welcome Address (Natália Oliva-Teles, Paula Jorge)

09h00 Neurodevelopmental Disorders (NDDs): Multidisciplinary Approach

Joana Damásio | Ataxias and movement disorders: the neurologist's view

Susana Silva Santos | Pedopsychiatry contribution to NDDs: CNV syndromes clinical approach

Carla Carmona | Neurological disorders: A psychologist's experience

Chair: Natália Oliva-Teles

10h30 Questions and Comments

Coffee Break

10h50 Autism Spectrum Disorders and Copy Number Variants (CNV)

Ana Beleza | Challenges in the diagnosis and counselling of low penetrance CNVs in the work-up of children with neurodevelopmental disorders

Catarina Prior | 16p11.2 and 22q11.2 deletions and duplications

Chair: M. João Sá

11h30 Trouble-shooting Session: Bring your clinical cases, discuss with the experts!

Case presentations

12h45 Questions and Comments

13h00 Lunch

14h00 Rare Copy Number Variants in Neurological Disorders

Yolanda | Rare, but more frequent CNVs in neurological disorders

Arjan de Brouwer | Diagnosing NDDs by genome sequencing

Chair: Paula Jorge

15h00 Questions and Comments

15h10 Neurogenomics

Yavuz Oktay | Severe NDDs: new approaches

Alex Garanto | RNA-based therapies for rare inherited diseases

Chair: François Foulquier

16h00 Paula Jorge | COST Action 16210 Funding Opportunities: STSM, ITC

16h15 Joris Vermeesch | MINDDS-connect (M-con) Project – an introduction

Chairs: (Natália Oliva-Teles, Paula Jorge)

17h15 End of Workshop

18h00 Porto de Honra / Port of Honour

Venue: Auditorium CHUP/CGM

Praça Pedro Nunes, 88 – 4099-028 , Porto, Portugal



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