HGVS
HUMAN GENOME
VARIATION SOCIETY

Clinical Interpretation of Variants from Next - Generation Sequencing

Hotel Vincci Maritimo
Barcelona, Spain
20th May 2016

8.00 – 8.55	Registration Speakers arrive and hand in presentations
9.00 – 9.10	**Introduction** William Oetting
Session 1	**Chair: Peter Taschner**
9.10 – 9.40	**The 100,000 Genomes Project** Tim Hubbard *Head of Genome Analysis, Genomics England, & Head of Department of Medical & Molecular Genetics Director of Bioinformatics King's Health Partners/King's College London, Faculty of Life Sciences & Medicine, King's College London*
9.40 – 10.00	**Comparing WGS to WES in a clinical setting** Martin Elferink
10.00 – 10.20	**Genetic testing for hereditary cancer: is exome sequencing ready or there is still room for *ad hoc* designed panels?** Lidia Feliubadalo
10.20 – 10.30	Discussion
10.30 – 11.00	**Coffee Break & Poster Session**

Session 2	**Chair: Anthony Brookes**
11.00 – 11.30	**Are we done with variant calling?** Ivo Gut *Director, CNAG, Barcelona, Spain*
11.30 – 11.50	**Towards formal specification of HGVS nomenclature enabling computational tool development** Jonathan Vis
11.50 – 12.10	**An efficient and accurate end-to-end solution leveraging network analytics to infer patient syndrome and identify causal mutations in rare disease cases** Sohela Shah
12.10 – 12.30	**What do public databases of clinical variants really tell us about classification concordance?** Stephen Lincoln
12.30 – 12.40	**Discussion**
12.40 – 14.00	**Lunch**
Session 3	**Chair: Christophe Beroud**
14.00 – 14.30	**Variants annotation and filtration in NGS context** David Salgado *NGS Team Manager at UMR_S910 - Aix-Marseille Université, France*
14.30 – 14.50	**Improving the annotation of clinically important genes to aid identification of missing causal variants** Adam Frankish
14.50 – 15.10	**Sitting on the fence: variant interpretation in RASopathies** Vijaya Ramachandran
15.10 – 15.20	**Discussion**
15.20 – 15.40	**Coffee Break**

Session 4	**Chair: William Oetting**
15.40 – 16.10	**Ethical issues of NGS and diagnosis of human genetics diseases** Heidi Howard
16.10 – 16.20	**Congenital haemolytic anaemia study with a targeted NGS panel** Celeste Bento
16.20 – 16.30	**Molecular Analysis of Single Nucleotide Polymorphism (SNPs) for β- Globin Gene in Tribes of Madhya-Pradesh, India** Ruchira Chaudhary
16.30 – 16.40	**SNPs based Genetic variability in tribes of Madhya-Pradesh (India) in reference to G-6-PD gene** Nitin Pathak
16.40 – 16.50	**Characterizing the intrinsic component of disease severity** Oscar Marin
16.50 – 17.00	**Meeting Summary & End**