HGVS
HUMAN GENOME VARIATION SOCIETY

PROGRAM

Copy Number Variation: data collection & analysis

Room J2
Gothia Towers
Gotteborg, Sweden

14th June 2019

(a satellite of ESHG)

8:00 - 9:00	**Registration**
9:00 - 9:05	**Welcome** Christophe Béroud
Session 1	**Chair: Christophe Béroud**
9:05 - 9:50	**KEYNOTE SPEAKER** **ELIXIR, data federation, and the Human Copy Number Variation Community** Gary Saunders *ELIXIR, Wellcome Genome Campus, Hinxton, Cambridgeshire, UK*

9:50 - 10:15 **KEYNOTE SPEAKER**

Annotation and display of structural variation data in Ensembl

Sarah Hunt
Ensembl, EMBL-EBI, Hinxton, Cambridgeshire, UK

10:15 - 10:25 **Presentation from selected Abstract**

Towards an optimised workflow for the identification of CNVs from heterogeneous WES data, within the scope of the SolveRD project

Steven Laurie
Centro Nacional de Análisis Genómico (CNAG-CRG), Center for Genomic Regulation, Barcelona Institute of Science and Technology (BIST), Barcelona, Spain

10:25 - 10:35 **Presentation from selected Abstract**

Assessing performance of copy number variation detection tools using simulated data

Iria Roca Otero
Genomes and Disease Group, Center for Research in Molecular Medicine and Chronic Diseases (CiMUS), University of Santiago de Compostela, Spain

10:35 - 10:40 **Rapid Fire Virtual Poster Presentations (*non - CNV*)**

- **AnnotSV 2.0: Annotation and Ranking of Human Structural Variations** - *Veronique Geoffroy*

- **Human genetics associated with dengue severity in Thailand** - *Unchana Arayasongsak*

10:40 - 11:15 Coffee Break

Session 2	Chair: Michael Baudis
11:15 - 12:00	**KEYNOTE SPEAKER** **hCNV in the context of the French Genomic Medicine 2025 Plan and the national BANCCO database** Damien Sanlaville *Claude Bernard University, Lyon, France*
12:00 - 12:10	**Presentation from selected Abstract** **Detection of copy number variations using whole-exome sequencing improves diagnostic yield of patients with rare Mendelian diseases** Paulo Silva *CGPP-IBMC – Centro de Genética Preditiva e Preventiva, Instituto de Biologia Molecular e Celular and i3S – Instituto de Investigação e Inovação em Saúde, Universidade do Porto, Portugal*
12:10 - 12:20	**Presentation from selected Abstract** **New CNV quality score enables discovering novel phenotype associations from genome-wide CNV analysis** Maarja Lepamets *Estonian Genome Center, Institute of Genomics, and Institute of Molecular and Cell Biology, University of Tartu, Tartu, Estonia*
12:20 - 12:30	**Presentation from selected Abstract** **Identification of mobile retroCNVs during genetic testing: consequences for routine diagnostics** Nicolas Chatron *Service de génétique, CHU Lyon and Equipe GENDEV, CRNL, INSERM U1028, CNRS UMR5292, UCBL1, Lyon, France*
12:30 - 13:45	Lunch

Session 3	Chair: Gary Saunders
13:45 - 14:30	**KEYNOTE SPEAKER** **CNV detection from exon capture data** Anna Benet-Pages *MGZ- Medical Genetics Centre, Munich, Germany*
14:30 - 14:40	**Presentation from selected Abstract** **Detection of copy number variations from NGS data using read depth information: a diagnostic performance evaluation** Olivier Quenez *Dept. of Genetics and CNR-MAJ, Normandie Univ, UNIROUEN, Inserm U1245 and Rouen University Hospital, Normandy Centre for Genomic and Personalized Medicine, Rouen, France*
14:40 - 14:50	**Presentation from selected Abstract** **Most rare and high-risk CNV carriers do not have major health, cognitive or socioeconomic consequences** Elmo Saarentaus *Institute for Molecular Medicine Finland FIMM, University of Helsinki, Finland*
14:50 - 15:00	**Presentation from selected Abstract** **Copy number variation detection tool for targeted sequencing data** Ashish Singh *Dept. of Medical Genetics, St. Olavs Hospital and Norwegian University of Science and Technology, Trondheim, Norway*
15:00 - 15:20	Coffee Break

Session 4	Chair: Anna Benet Pages
15:20 - 16:05	**KEYNOTE SPEAKER** **Association analysis of SVs/CNVs using NGS data** Victor Guryev *European Research Institute for the Biology of Ageing (ERI-BA), University Medical Center, Groningen, The Netherlands*
16:05 - 16:50	**KEYNOTE SPEAKER** **Implementation Driven Development of Standards for Genomic Data Exchange from Cancer Genome Data Collections** Michael Baudis *Institute of Molecular Life Sciences and Swiss Institute of Bioinformatics, University of Zurich, Switzerland*
16:50 - 17:00	Closing Remarks
17.00	**MEETING END**