PROVIDING THE MOST COMPLETE VIEW OF HUMAN GENETIC VARIATION

PACBIO®

ESHG 2019

PRESENTATIONS

E01.2 - Resolving human genetic variation with long-read single-molecule sequencing

M. J. P. Chaisson, University of Southern California

Saturday, June 15, 8:30 - 9:00
Session: New technologies (Sponsored by Illumina) - Room: Hall C

C03.1 - Intronic expansions of an ATTTC pentamer in the STARD7 gene underlie Familial Adult Myoclonic Epilepsy linked to chromosome 2 (FAME2)

J. Gecz, The University of Adelaide

Saturday, June 15, 18:30 - 18:45
Session: Neurogenetic and psychiatric disorders - Room: F1+F2+F3

E05.1 - How long do we need? The relative value of emerging sequencing technologies in genomic medicine

M. Talkowski, Cambridge

Sunday, June 16, 8:30 - 9:15
Session: The longer the better? Third generation sequencing technologies - Room: K2+K3

C19.1 - Phasing of complex genomic rearrangements reveal involvement of both homologous chromosomes in pre- and post-zigotic events

C. M. Carvalho, Baylor College of Medicine

Monday, June 17, 13:00 - 13:15
Session: From genome architecture to RNA biology - Room: F4+F5

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POSTERS

Day	Time	Pres #	Title and Author	Session
Sunday, June 16	10:15 - 11:15	P12.100A	**Variant analyses of PMS2 by single-molecule long-read sequencing** K. Neveling, Radboud UMC, Nijmegen, Netherlands	Cancer genetics
		P16.45A	**Long read sequencing of patient-parent trios with intellectual disability** C. Gilissen, Radboud UMC, Nijmegen, Netherlands	Omics - Bioinformatics
		P16.69A	**PopDel calls medium-size deletions jointly in tens of thousands of genomes** S. Roskosch, Berlin Institute of Health / Charité, Berlin, Germany	Omics - Bioinformatics
		P16.85A	**Comprehensive variant detection in a human genome with highly accurate long reads** W. J. Rowell, Pacific Biosciences, Menlo Park, CA, United States	Omics - Bioinformatics
		P18.81A	**Increasing the value of the Swedish 1000 whole-genome data resource** A. Ameur, Uppsala University, Uppsala, Sweden	Genetic epidemiology - Population genetics - Statistical methodology - Evolutionary genetics
	16:45 - 17:45	P10.27B	**Distinguishing Highly Similar SMN1/2 Genes and Identifying Novel SMN Transcripts by Targeted Capture of PacBio Single-molecule Long-read Sequencing** M. Dai, Department of Genetic Counseling, Xinhua Hospital, School of Medicine, Shanghai Jiao Tong University, Shanghai, China.	Neuromuscular disorders
		P14.003B	**Sequencing the previously unsequencable using amplification-free targeted enrichment powered by CRISPR-Cas9** J. Ekholm, Pacific Biosciences, Menlo Park, CA, United States	New diagnostic approaches - Technical aspects - Quality control
		P14.051B	**CRISPR/Cas9-targeted enrichment and long-read sequencing of a non-coding corneal dystrophy-associated TCF4 triplet repeat** N. J. Hafford-Tear, UCL Institute of Ophthalmology, London, United Kingdom	New diagnostic approaches - Technical aspects - Quality control
Monday, June 17	10:15 - 11:15	P14.012C	**Utilizing long-read amplicon sequencing for comprehensive pan-ethnic spinal muscular atrophy (SMA) carrier screening** D. A. Zeevi, Dor Yeshorim Committee for Prevention of Jewish Genetic Diseases, Jerusalem, Israel	New diagnostic approaches - Technical aspects - Quality control
		P14.068C	**The power of 16kb Long-Read Circular Consensus Sequencing for the homologous OPN1 genes** K. Neveling, Department of Human Genetics, Nijmegen, Netherlands	"New diagnostic approaches - Technical aspects - Quality control
		P16.71C	**Using multiple sequencing platforms to identify and characterise disease-causing genome alterations.** G. Gallone, Max Planck Institute for Molecular Genetics, Berlin, Germany	Omics - Bioinformatics
	16:45 - 17:45	P14.029D	**Evaluation of preliminary benchmark deletions for the reference sample NA12878** R. Rajagopalan, Division of Genomic Diagnostics, Children's Hospital of Philadelphia, Philadelphia, PA, United States	New diagnostic approaches - Technical aspects - Quality control
		P16.12D	**Deciphering complex genomic rearrangements by traveling through 3rd generation sequencing in rare diseases: two chromothripsis cases** S. Verdez, UF Innovation en diagnostic génomique des maladies rares, Dijon, France	Omics - Bioinformatics

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