Oxford NANOPORE Technologies

ASHG
American Society of Human Genetics

OXFORD NANOPORE AT ASHG 2024

Booth 203

Presentations featuring nanopore sequencing

Wednesday, November 6th

Session 11: All the single cells (8:00–9:30)

Room: 501

8:45–9:00 Single-cell long-read sequencing analysis in endemic pemphigus foliaceus

Ticiana Farias, The University of North Carolina at Charlotte, US

Thursday, November 7th

Session 47: From variant to function: predicting and understanding variants function (10:15–11:45)

Room: Mile High Ballroom 2&3

11:15–11:30 *De novo* precise splice site predictor using deep learning and integration with minimap2 for enhanced long-read sequence alignment

Siying Yang, Harvard Medical School, US

Friday, November 8th

Session 74: The non-coding genome: from nucleotide to protein (10:15–11:45)

Room: Four Seasons Ballroom 1

10:30–10:45 Nanopore sequencing of chromatin accessibility

Phillip James, Oxford Nanopore Technologies, UK

Session 82: Read all about it: transcriptomic insights from new sequencing technologies (13:15–14:15)

Room: 501

13:30–13:45 Single-Cell Omics for Transcriptome CHaracterization (SCOTCH): isoform-level characterization of gene expression through long-read single-cell RNA sequencing

Zhuoran Xu, University of Pennsylvania, US

Session 83: Splice splice baby: isoform expression in health and disease (13:15–14:15)

Room: 505

13:15–13:30 An atlas of expressed transcripts in the prenatal and postnatal human cortex

Rosemary Bamford, University of Exeter, UK

Saturday, November 9th

Session 89: Long-read sequencing offering new insights into neurological disease (8:00–9:00)

Room: 505

8:00–8:15 Long-read sequence and assembly of autism reference genomes

Yang Sui, University of Washington, US

8:15–8:30 Long-read sequencing to diagnose autosomal recessive Parkinson's disease in diverse populations

Kensuke Daida, NIH, US

Posters featuring nanopore sequencing

Wednesday, November 6th

14:30–16:30

Omics technologies

1106W: Assessing the readiness of Oxford Nanopore sequencing for clinical genomics applications

Judith Arres, M42, UAE, *Presenter 007*

1111W: Building large-scale population databases of structural variant calls from low- and high-coverage long-read sequencing data

Danny E Miller, University of Washington, US, *Presenter 012*

1152W: Long-read RNA sequencing reveals isoform diversity in human immune cells and implications for diseases

Jun Inamo, Tokyo Medical and Dental University, JP, *Presenter 053*

1181W: Telo-seq: native, multiplexed telomere sequencing of individual chromosomes using 3' end adapter ligation

Scott Hickey, Oxford Nanopore Technologies, US, *Presenter 082*

Epigenetics

1009W: Comprehensive profiling of genetic and epigenetic features for FSHD diagnosis using targeted nanopore sequencing

Florentine Scharf, MGZ Medical Genetics Center, DE, *Presenter 009*

1013W: DNA methylation patterns and differences in two long-read sequencing platforms in a clinical pediatric cohort

Sergey Batalov, Rady Children's Hospital, US, *Presenter 013*

Mendelian phenotypes

2049W: Long-read RNA sequencing reveals Parkinson's disease-associated transcription in oligodendrocytes

James Evans, The Francis Crick Institute, UK, *Presenter 047*

Molecular and cytogenetic diagnostics

3060W: Long-read genomic sequencing reveals expanded GAA-GGA chimeric alleles in Friedreich ataxia

Sanjay Bidichandani, University of Oklahoma Health Sciences Center, US, *Presenter 020*

3067W: Resolving complex genotypes in residual clinical samples with long range-PCR and nanopore sequencing assay

Cody Edwards, Asuragen, US, *Presenter 026*

Complex traits and polygenic disorders

5131W: Nanopore long-read sequencing of Lewy body dementia cases from the DementiaSeq1 cohort for structural variation and integrative genetics study

Elizabeth Rice, The American Genome Center, US, *Presenter 128*

5041W: Detecting gene and isoform expression changes in the IBDs using long-read sequencing

Meaghan Kennedy Ng, University of North Carolina at Chapel Hill, US, *Presenter 040*

Prenatal, perinatal, and developmental genetics

7040W: Long-read whole genome sequencing for comprehensive genomic profiling of men with infertility

Thomas Garcia, Baylor College of Medicine, US, *Presenter 016*

Thursday, November 7th

14:30–16:30

Complex traits and polygenic disorders

5186T: Whole-genome identification of short tandem repeats underlying idiopathic peripheral neuropathy using short-read and long-read sequencing

Zitian Tang, Washington University School of Medicine, US, *Presenter 183*

Genetic, genomic, and epigenomic resources and databases

1055T: Characterization of AZF deletions on the Y chromosome using nanopore long-read sequencing: advances and diagnostic applications

AbdelAli Zrhidri, Inovie — GenBio, FR, *Presenter 014*

1061T: Construction of a Korean pan-genome based on long-read sequences

Jae-Yoon Kim, Korea Research Institute of Bioscience and Biotechnology, KR, *Presenter 020*

1080T: Long-read sequencing of hundreds of ancestrally diverse brains provides insight into the impact of structural variation on gene expression and methylation

Kimberley Billingsley, NIH, US, *Presenter 039*

Mendelian phenotypes

2032T: Enhanced detection and genotyping of disease-associated tandem repeats using HMMSTR and targeted long-read sequencing

Kinsey Van Deynze, University of Michigan, US, *Presenter 030*

2068T: Pathological short tandem repeats analysis and imprinting disorders by long-read sequencing in affected individuals

Hagar Mor-Shaked, Hadassah Medical Organization, IL, *Presenter 065*

Molecular and cytogenetic diagnostics

3052T: Expanding the diagnostic toolkit using long-read RNA-sequencing

Kylie Montgomery, UCL GOS ICH, UK, *Presenter 012*

3060T: Molecular genetic testing of congenital adrenal hyperplasia with massive parallel long-read sequencing

Qizong Lao, NIH Clinical Center, US, *Presenter 020*

Omics technologies

1110T: Automated and scalable single technology telomere-to-telomere assembly of human genomes

Sean McKenzie, Oxford Nanopore Technologies, US, *Presenter 011*

1113T: Benchmarking of Straglr for the detection of short-tandem repeats using high-coverage Cas9-enriched nanopore sequencing data

Veronika Scholz, MGZ Medical Genetics Center, DE, *Presenter 014*

1133T: Enhanced library preparation methods for targeted Oxford Nanopore Technologies (ONT) long-read sequencing

Yu-Chi Ho, National Taiwan University, TW, *Presenter 034*

1139T: From validation to diagnosis: metadata analysis of long-read sequencing for clinical insights

Suzanne Drury, Geneyx Genomics, UK, *Presenter 040*

1156T: needLR: A structural variant filtering and prioritization tool for long-read sequencing data

Jonas Gustafson, University of Washington, US, *Presenter 057*

1162T: Piloting the 48-hour sequencing workflow for BadgerSeq: a decentralized model for ultra-rapid long-read diagnostic genome sequencing

Stephen Meyn, University of Wisconsin School of Medicine, US, *Presenter 063*

Pharmacogenomics

7022T: Resolution of *CYP2D6* pharmacogene complex haplotypes with end-to-end Oxford Nanopore sequencing workflows

Carly Tyer, Oxford Nanopore Technologies, US, *Presenter 022*

Friday, November 8th

14:30–16:30

Epigenetics

1037F: Using long-read sequencing for genomic and epigenomic analysis in patient-derived samples with somatic *IDH1* mosaicism

Carolina Maria Montano, Johns Hopkins University, US, *Presenter 037*

Evolutionary and population genetics

3024F: Long-read sequencing on diverse populations identifies an association of a haplotype with two CAA interruptions in the CAG trinucleotide repeats of the *ATXN2* gene

Beoung Hun Lee, University of Pennsylvania, US, *Presenter 02*

Genetic, genomic, and epigenomic resources and databases

1044F: AI cloud-based end-to-end technology for accurate, fast and affordable newborn screening of genetic disorders/rare diseases

Chris Kyriakidis, gMendel, DK, *Presenter 003*

1080F: Long-read based detection of large copy number variants with potential functional significance using the ContextSV structural variant caller

Jonathan Perdomo, CHOP, US, *Presenter 039*

Mendelian phenotypes

2024F: Complete nanopore repeat sequencing of *SCA27B* (GAA-FGF14 ataxia) in Japanese

Naomichi Matsumoto, Yokohama City University Graduate School of Medicine, JP, *Presenter 022*

Molecular and cytogenetic diagnostics

3045F: Beyond short-read WGS: long-read WGS as a stand-alone tool for genetic diagnosis of neurological diseases

Wei Ma, The Hong Kong Genome Institute, CN, *Presenter 005*

Molecular effects of genetic variation

6050F: Leveraging long-read Oxford Nanopore Technologies and a national genome program to better understand structural variation inheritance and medical impact

Daniel Matias Sanchez, M42, UAE, *Presenter 048*

Omics technologies

1173F: Streamlined, multimodal, epigenetic measurements using CUT&RUN combined with nanopore sequencing

Paul Hook, Johns Hopkins University, US, *Presenter 074*

1143F: Long-read sequencing to diagnose Mendelian diseases

Yulia Mostovoy, Massachusetts General Hospital, US, *Presenter 044*

1155F: Phasing accuracy of long-read whole genome sequencing is coverage and reference genome dependent

Jiayu Fu, NHGRI, NIH, US, *Presenter 056*

Statistical genetics and genetic epidemiology

4051F: Evaluating long-read phasing methods in clinically-relevant genes using data from the 1000 Genomes ONT Sequencing Consortium

Nikhita Damaraju, University of Washington, US, *Presenter 04*

Oxford NANOPORE Technologies

nanoporetech.com

Oxford Nanopore Technologies activities at ASHG

On-booth activities (Booth 203)

Wednesday, November 6th

Single cell demo: Individual cells matter

10:30

Carly Tyer, *Oxford Nanopore Technologies*

ElysION: from sample to analysis, end-to-end nanopore sequencing on a single device

13:00

Steph Wilbraham, *Oxford Nanopore Technologies*

Large-scale comprehensive Nanopore sequencing identifies regulatory loci driven by structural variants in diverse Human Brains

15:30

Melissa Meredith, *Graduate Student Researcher, Computational Genomics Lab, UC Santa Cruz*

Thursday, November 7th

Data for Breakfast - Many samples to answers: scaling up long-read WGS projects with nanopore sequencing and EPI2ME wf-human-variation

9:45

Bryan Leland, *Oxford Nanopore Technologies*

EPI2ME demo: Comprehensive germ line variant calling and 5mC/5hmC detection with wf-human-variation

15:30

Bryan Leland, *Oxford Nanopore Technologies*

Friday, November 8th

Data for Breakfast - Nanopore sequencing tackles challenging genes

10:30

Phill James, *Oxford Nanopore Technologies*

Single cell demo: Individual cells matter

12:30

Carly Tyer, *Oxford Nanopore Technologies*

EPI2ME Demo: Analyzing full-length transcripts with wf-single-cell

15:30

Bryan Leland, *Oxford Nanopore Technologies*

Industry education session

Thursday, November 7th

Bridging the gap: Discover what you’re missing with comprehensive and accessible genomic research

12:00–13:00

Room: 109

CoLab sessions

Wednesday, November 6th

Scalable long-read enrichment and phamacogenomic analysis with Oxford Nanopore Technologies

14:30–15:00

Room: Theater 2, Exhibit & Poster Hall/Upper Level

From arrays to sequencing: Enhancing methylation analysis for biomarker discovery and clinical applications (Presented by Wasatch Biolabs)

12:00–12:30

Room: Theater 1, Exhibit & Poster Hall/Upper Level

Thursday, November 7th

Telomere-to-telomere genome assembly using Oxford Nanopore’s long and ultra-long reads

16:00–16:30

Room: Theater 1, Exhibit & Poster Hall/Upper Level

Oxford Nanopore evening reception

Thursday, November 7th

20:00pm–12:00am

Venue: 1134 Broadway

Location: 1134 N Broadway, Denver, CO 80203

Please register to join members of Oxford Nanopore Technologies and users of nanopore technology for an evening of substantial canapés, drinks, and networking. The Oxford Nanopore Technologies team and speakers from our conference events will be on hand all evening at 1134 Broadway, just a five minute drive from the conference venue!

Find out more and register for Oxford Nanopore's activities at ASHG:

Oxford NANOPORE Technologies

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