Attend Our Workshop

Medical Genetics:
Identification of Hidden Structural Variants with Long-Read Sequencing
Alexander Hoischen, Ph.D., Radboud University Medical Center
Thursday, February 15, 9:40 a.m. - 10:00 a.m.
Bonnet Creek Ballroom I-VI

Hear Lightning Talks in Our Hospitality Suite

Columbia Room
Tuesday, February 13, 8:30 a.m.
What’s New With the Sequel® System? More Data, Better Biology
Jonas Korlach, Ph.D., CSO

Tuesday, February 13, 10:45 a.m.
PacBio® Long-Read WGS for Structural Variant Discovery
Aaron Wenger, Ph.D., Staff Scientist

Wednesday, February 14, 10:30 a.m.
No-Amp Targeted Enrichment & SMRT® Sequencing of Repeat-Expansion Disease Causative Genomic Regions
Jonas Korlach, Ph.D., CSO

Visit Our SMRT Sequencing Posters
Authors will present odd-numbered posters on Tuesday and even-numbered posters on Wednesday.

307: Mitochondrial DNA Sequencing Using the PacBio Sequel System - Meredith Ashby, Ph.D.
413: Multiplexed Complete Microbial Genomes on the Sequel System with Accelerated, Single-Tube SMRTbell® Library Prep - Cheryl Heiner, Ph.D.
913: Full-length Transcript (Iso-Seq®) Profiling for Improved Genome Annotations - Michelle Vierra
1003: Population-Scale Discovery of Structural Variants with PacBio SMRT Sequencing - Aaron Wenger, Ph.D.
1106: Amplification-free Targeted Enrichment and SMRT Sequencing of Repeat-Expansion Disease Causative Genomic Regions - Janet Ziegle
1212: High-quality De Novo Genome Assembly and Intra-Individual Mitochondrial Instability in the Critically Endangered Kākāpō - Jonas Korlach, Ph.D.
TYPES OF GENOMIC VARIATION

SMALL VARIANTS
< 50 BASE PAIRS / VARIANT

INDELS
1-49 BASE PAIRS / VARIANT

STRUCTURAL VARIANTS
> 50 BASE PAIRS / VARIANT

DELETION
INSERTION
TANDEM DUPLICATION
INTERSPERSED DUPLICATION
INVERSION
TRANSLOCATION

SNV
1 BASE PAIR / VARIANT

KNOWN DISEASES CAUSED BY STRUCTURAL VARIANTS

SCHIZOPHRENIA
Deletion, Duplication

CARNEY COMPLEX
Deletion

POOR DRUG METABOLISM
Duplication

HEREDITARY BREAST & OVARIAN CANCER
Deletion, Insertion

NEUROFIBROMATOSIS
Insertion

CHRONIC MYELOID LEUKEMIA
Translocation

+ 1,000's MORE KNOWN...
...AND MANY TO BE DISCOVERED

> 20,000 STRUCTURAL VARIANTS IN A HUMAN GENOME

STRUCTURAL VARIANTS COMPRIS MORE THAN HALF OF GENOMIC VARIATION

60%