13th International Symposium on Mutation in the Genome: detection, genome sequencing & interpretation

26th – 30th April 2015
Holiday Inn, Leiden, The Netherlands

PROGRAMME

Sunday 26th April

09.00 – 09.30	**Registration Mutation Detection Meeting & LSDB Curation Course:** *Outside Gouda*
09.30 – 17.00	**LSDB Curation Course –** *Gouda Room*

Monday 27th April

09.00 – 10.45	**Registration** – *Patio* *Coffee Mixer - Refreshments from 9.45– catchup with old friends, meet new colleagues*
Session 1	**Chair: Johan T. den Dunnen**
10.45 – 10.55	**Welcome** Johan T. den Dunnen
10.55 – 11.40	**KEYNOTE SPEAKER** **New technologies applied in the diagnostic laboratory** Joris Vermeesch *Lab. for Cytogenetics & Genome Research, KU Leuven, Leuven, Belgium*
11.40 – 12.10	**KEYNOTE SPEAKER** **Clinical Exome Sequencing at a Large Academic Medical Center: Diagnostic Yield, Variant Spectrum, and Lessons Learned** Wayne Grody *UCLA School of Medicine, CA, USA*
12.10 – 12.30	**PacBio sequencing: Improving mutation detection in complex genomic regions** Seyed Yahya Anvar *Leiden University Medical Center, The Netherlands*
12.30 – 13.40	**Lunch –** *Garden Restaurant*
Session 2	**Chair: Andreas Laner**
13.40 – 14.00	**Multiplex targeted long amplicon sequencing method for Cyp2d6 genotyping using the PacBio RSII** Henk Buemans *Leiden University Medical Centre, Leiden, The Netherlands*
14.00 – 14.20	**Unravelling the origin of variants identified which may or may not be part of a patient's genotype** Desiree Du Sart *Victorian Clinical Genetics Services, Melbourne, Australia*
14.20 - 14.40	**Validating Bioinformatic Pipelines for the Clinic** Kenneth Doig *Peter MacCallum Cancer Institute, Melbourne, Australia*
14.40 – 15.00	**Combination of MLPA and Illumina MiSeq Amplicon Sequencing provides maximum**

mutation detection coverage for TSC1/TSC2 gene analyses in patients with Tuberous Sclerosis Complex
Teguh Haryo Sasongko
School of Medical Sciences, Universiti Sains Malaysia, Kubang Kerian, Malaysia

15.00 – 15.20

COMPANY LECTURE

MLPA 2.0 - Multiplex Ligation-dependent Probe Amplification on Illumina NGS platforms using a 600+ probe assay
Jan Schouten
MRC - Holland

15.20 – 15.35 **RAPID FIRE POSTER PRESENTATIONS**

15.35 – 16.10 **Poster Session 1**

Session 3 **Chair: Peter Taschner**

16.10 – 16.55 **3Gb-TEST SPONSORED SPEAKER**
Is exome sequencing of single patients with intellectual disability an effective diagnostic strategy? And what about whole genome sequencing?
Claudia Ruivenkamp
Leiden University Medical Center, The Netherlands

16.55 – 17.15 **A novel POLE variant, identified by exome sequencing, in a family with high burden of colorectal- and extra-colonic cancers**
Maren F. Hansen
Norwegian University of Science and Technology, Trondheim, Norway

17.15 – 17.35 **Target Locus Amplification (TLA): A comprehensive new DNA test for detection of fusion genes in leukemia**
Birgit Sikkema-Raddatz
University of Groningen, University Medical Center, Groningen, The Netherlands

17.35 – 17.55 **Pathway analyses of whole genome sequence data identifies novel candidate Intellectual Disability genes**
Farah Zahir
Canada's Michael Smith Genome Sciences Center, Vancouver, BC, Canada

17.55 **End of Day – Evening at Leisure**

Tuesday 28th April

Session 4	Chair: Maria Jesus Sobrido
09.00 – 09.40	**KEYNOTE SPEAKER** **RNA, The Neglected Molecule** Johan T. den Dunnen *Leiden University Medical Centre, Leiden, The Netherlands*
09.40 – 10.00	**Traditional vs. Next-Generation Panel Testing of Hereditary Breast and Ovarian Cancer Genes in a Large Clinical Population** Stephen Lincoln *Invitae, San Francisco, CA, USA*
10.00 – 10.20	**Next Generation Sequencing of Short Tandem Repeats: additional variation takes Forensic mixture analysis to the next level** Kristiaan van der Gaag *Leiden University Medical Centre, Leiden, The Netherlands*
10.20 – 10.40	**COMPANY LECTURE** BIONANO GENOMICS **The Irys System; Rapid Genome Wide Mapping at the Single Molecule Level Using Nanochannel Arrays for Structural Variation Analysis and de novo Assembly** Jack Peart *BioNano Genomics, UK*
10.40 – 11.10	**Coffee Break -** *Patio*
Session 5	**Chair: Stefan White**
11.10 – 11.55	**KEYNOTE SPEAKER** **De novo Genome Assembly using Diverse Data Types** Ivo Gut *Fundacio Privada Parc Cientific de Barcelona, Barcelona, Spain*
11.55 – 12.15	**Rapid screening for monogenic diseases in severely ill newborns using whole genome sequencing** Cleo van Diemen *University Medical Center Groningen, The Netherlands*
12.15 – 12.35	**The impact of genetic variations in miRNA binding sites on the miRNA-mediated regulation of genes associated with cardiometabolic traits** Mohsen Ghanbari *Erasmus University Medical Center, Rotterdam, The Netherlands*
12.35 – 13.50	**Lunch –** *Garden Restaurant*

Session 6	**Chair: Ivo Gut**
13.50 – 14.10	**HGVS SPONSORED SPEAKER** **New Approaches to Pathogenicity Interpretation in the Age of Precision Medicine** Marc Greenblatt *University of Vermont College of Medicine, Burlington, VT, USA*
14.10 – 14.30	**Evaluating inheritance pattern for genetic variant interpretation: are we generally oversymplistic?** Maris Jesus Sobrido *Fundacion Publica Galega de Medicina Xenomica-Instituto de Investigacion Sanitaria de Santiago, Santiago de Compostela, Spain*
14.30 – 14.50	**Using Selective Constraint at the Domain Level to Assess Variant Pathogenicity** Christopher Cassa *Harvard Medical School, Cambridge, MA, USA*
14.50 – 15.10	**PON-P2 and PON-Diso - reliable prediction of variation pathogenicity** Mauno Vihinen *Lund University, Lund, Sweden*
15.10 – 15.50	**Poster Session 2**
Session 7	**Chair: Mauno Vihinen**
15.50 – 16.10	**The European Variation Archive: A New Genetic Variation Resource at EMBL-EBI** Garry Saunders *European Bioinformatics Institute (EMBL-EBI) Wellcome Trust Genome Campus, Cambridge, Great Britain*
16.10 – 16.30	**Computer analysis of context dependencies of SNP sites in human genome** Nataly Safranova *Institute of Citology and Genetics SB RAS, Novosibirsk, Russia*
16.30 – 16.50	**An Efficient Algorithm for the Extraction of HGVS Descriptions** Jonathan Vis *Dept. of Molecular Epidemiology, Leiden University Medical Center and Leiden Institute of Advanced Computer Science, Leiden University, Leiden, The Netherlands*
17.10 Departure	**Welcome Reception –** *Leiden City Hall* • *Remainder of evening at leisure (no return to hotel)*

Wednesday 29th April	
Session 8	**Chair: Mats Nilsson**
09.00 – 09.45	**KEYNOTE SPEAKER** **Developments in single cell sequencing** Thierry Voet *Department of Human Genetics, KU Leuven, Leuven, Belgium*
9.45 – 10.05	**Reverse Diagnosis of Missense Variants In DNA Mismatch Repair Genes in Lynch syndrome** Mark Drost *Leiden University Medical Cee, Leiden, The Netherlands*
10.05 – 10.25	**Calibrating an in vitro functional assay for use in the diagnosis of Lynch syndrome due to missense variants in the mismatch repair genes** Marc Greenblatt *University of Vermont College of Medicine, Burlington, VT, USA*
10.25 – 11.00	**Coffee Break -** *Patio*
Session 9	**Chair: Joris Vermeesch**
11.00 – 11.45	**KEYNOTE SPEAKER** **Spatially resolved mutation detection and sequencing *in situ*** Mats Nilsson *Stockholm University, Sweden*
11.45 – 12.05	**Full-length mRNA sequencing uncovers a widespread coupling between transcription and mRNA processing** Peter A.C. 't Hoen *Leiden University Medical Center, Leiden, The Netherlands*
12.05 – 12.25	**How to choose the right predictor for variation interpretation** Mauno Vihinen *Lund University, Lund, Sweden*
12.25 – 13.30	**Lunch –** *Garden Restaurant*
Session 10	**RD-Connect/Global Alliance for Genomics and Health: data sharing and integration for rare diseases: "Knowledge from sharing"** **Chair: Peter A.C. 't Hoen**
13.30 – 13.50	**What data sharing means for patients with rare diseases: knowledge from sharing** Milan Macek Jr. *Charles University, Prague, Czech Republic*
13.50 – 14.10	**A charter and code of practice enabling data sharing: ethics, privacy and legal issues** Mats Hansson *Centre for Research Ethics & Bioethics, Uppsala University, Uppsala, Sweden*

14.10 – 14.30	**Data integration for rare diseases facilitated by the RD-Connect platform** Ivo Gut *Fundacio Privada Parc Cientific de Barcelona, Barcelona, Spain*
14.30 – 14.50	**Genetic variation interoperability standards: Global Alliance for Genomics and Health data sharing beacon project** Peter Goodhand *The Global Alliance for Genomics and Health, Toronto, Canada*
14.50 – 15.10	**Genetic variation interoperability standards: Semantic interoperability standards; data FAIRports** Barend Mons *Center for Human and Clinical Genetics, LUMC, Leiden, The Netherlands*
15.10 – 15.30	**General Discussion**
15.30 – 16.00	**Coffee Break -** *Patio*
16.00 – 17.15	Free Time
17.15 Departure	**Conference Dinner Surprise** • *Assemble in hotel lobby for prompt departure* • *Location will be revealed at the meeting for those staying in other hotels* • *Please wear warm outer clothing*

Thursday 30th April

Session 11	**Chair: Marc Greenblatt**
09.00 – 9.45	**KEYNOTE SPEAKER** **Rare, non-coding DNA variation and disease** Stefan White *Leiden University Medical Center, Leiden, The Netherlands*
09.45 – 10.05	**GeneMatcher: A Matching Tool for Identification of Individuals with Mutations in the Same Gene** Nara Sobreira *Johns Hopkins University, Baltimore, MD, USA*
10.05 – 10.25	**Regression and overdispersion correction improves sensitivity of non-invasive prenatal testing** Lennart Johanasson *University Medical Center Groningen, Groningen, The Netherlands*
10.25 – 11.00	**Coffee Break**
Session 12	**Chair: Johan T. den Dunnen**
11.00 – 11.45	**KEYNOTE SPEAKER**

	Integrated genome and transcriptome sequencing of the same cell Siddharth Subhas Dey *Hubrecht Institute, Utrecht, The Netherlands*
11.45 – 12.05	**Genome in a Bottle: You may have sequenced, but how well did you do?** Stephen Lincoln *Invitae, San Francisco, CA, USA*
12.05 – 12.35	**KEYNOTE SPEAKER** **The Global Variome** Sir Prof. John Burn *Institute of Genetic Medicine Newcastle University Centre for Life Central Parkway, Newcastle upon Tyne, United Kingdom*
12.35 – 12.45	**Windup & Summary** **MEETING END**
12.45	**Lunch –** *Garden Restaurant*