THE HUMAN VARIOME PROJECT
an NGO official partner of UNESCO
sharing data • reducing disease

14th International Symposium on Variants in the Genome: detection, sequencing & interpretation

5 - 7 June 2017

NH Collection Santiago
Santiago de Compostela
Spain

Scientific Programme Committee

Prof. Johan T. den Dunnen (Leiden, Nederland) CHAIR

Prof. Sir John Burn (Newcastle, UK)

Prof. Angel Carracedo (Santiago de Compostela, Spain)

Dr Reece Hart (San Francisco, CA, USA)

Dr Andreas Laner (Munich, Germany)

Dr Maria-Jesus Sobrido (Santiago de Compostela, Spain)

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5 - 7 June 2017, Santiago de Compostela, Spain

FINAL PROGRAM

Monday 5th June

8.30 - 14.00	Exhibitor Bump In	
8.00 - 10.30	**REGISTRATION**	
9.00 - 10.40	**PRE-MEETING WORKSHOP 1** **Variant Nomenclature** **Room: Allariz + Noia** *(Included in registration)* Johan T. den Dunnen *Leiden Univ. Medical Center, Leiden, Netherlands* *Please read the Workshop Summary before attending.*	**PRE-MEETING SPONSORED WORKSHOP 2** **Clinical Genomics - Interpretation & Reporting with QIAGEN Bioinformatics** **Room: Obradoiro** *(Included in registration)* *Please read the Workshop Summary before attending regarding requirements e.g. laptop required.* *QIAGEN is a sponsor of the 14th International Symposium on Variants in the Genome*
10.40 - 11.00	**Coffee Break** **Room: Obradoiro Terrace**	
11.00 - 11.10	**INTRODUCTION & WELCOME** **Introduction** Johan T. den Dunnen *Leiden Univ. Medical Center, Leiden, Netherlands* **Welcome** Maria-Jesus Sobrido & Angel Carracedo *Instituto de Investigaciones Sanitarias de Santiago / Fundación Pública Galega de Medicina Xenómica*	
11.10 - 13.00	**PLENARY SESSION 1 - Variant Calling And Annotation For Diagnostic Applications** **Room: Obradoiro** **Chair: Johan T. den Dunnen**	
11.10 - 11.50	**Phenotype Driven Genomic Diagnostics** Peter Robinson *Computational Biology Group at the Jackson Lab. for Genomic Medicine, Farmington CN, USA*	

11.50 - 12.20	**CNV detection from targeted next-generation panel sequencing data in routine diagnostics** Anna Benet-Pagès *Medizinisch Genetisches Zentrum, Munich, Germany*
	PRESENTATIONS FROM ABSTRACTS
12.20 - 12.40	**GeneHancer and VarElect: disease interpretation of whole genome sequence variants** Doron Lancet *Weizmann Institute of Science, Rehovot, Israel*
12.40 - 13.00	**Chromium™: Full spectrum genome analysis with Linked-Reads** Steve Giavas *10x Genomics Inc.* *10x Genomics is a sponsor of the 14th International Symposium on Variants in the Genome*
13.00 - 14.00	**Lunch** **Room: Azabache Restaurant**
14.00 - 15.30	**PLENARY SESSION 2 - The BRCA Challenge** **Room: Obradoiro** **Chair: Stephen Chanock (TBC)**
14.00 - 15.00	**The BRCA Challenge** Prof. Sir John Burn *Institute of Genetic Medicine, International Centre for Life, Newcastle upon Tyne, UK* Gunnar Ratsch *ETH Zurich, Memorial Sloan-Kettering Cancer Center, Weill Cornell Medical College*
15.00 - 15.30	**BRCA Exchange Mobile: Enabling Patient Access, Notification, and Case-Level Data Ingress** Faisal Alquaddoomi *ETH Zurich, Switzerland*
15.30 - 16.10	**Poster Session 1** **Room: Quintana & Lobby** ***Odd numbers to present: POS001, POS003 etc.***
16.10 - 16.20	**Assemble for Group Photograph** **Room: Obradoiro Terrace**
16.20 - 18.00	**PLENARY SESSION 3 - The BRCA Challenge (continued)** **Room: Obradoiro** **Chair: Prof. Sir John Burn**
16.20 - 16.50	**The Genomics England 100,000 Genomes Project: Establishing a centralised national resource of genomic data** Claire Turnbull *Genomics England, Queen Mary University of London and Institute of Cancer Research, London, UK*

16.50 - 17.20 **Novel genes involved in Fanconi anemia, DNA repair and cancer predisposition: the clinical relevance of functional studies of genetic variants**

Jordi Surrallés Calonge
Universitat Autònoma de Barcelona, Catalunya, Spain

PRESENTATIONS FROM ABSTRACTS

17.20 - 17.40 **A unified framework for prioritization of variants of uncertain significance In hereditary breast and ovarian cancer (HBOC)**

Peter Rogan
University of Western Ontario and CytoGnomix Inc., London, Canada

17.40 - 18.00 **Pilot multi-gene testing in Hereditary Breast-Ovarian Cancer**

Paola Carrera
IRCCS San Raffaele Scientific Institute, Milano, Italy

19.00 - 20.30 **WELCOME RECEPTION**
Room: Obradoiro Terrace

Tuesday 6th June

8.30 - 10.30 **PLENARY SESSION 4 - Variant Interpretation In The Clinic**
Room: Obradoiro
Chair: Reece Hart

8.30 – 9.00 **A systematic framework for the clinical interpretation of chromosomal copy number variants**

Swaroop Aradhya
On behalf of the ACMG/ClinGen Structural Variant Working Group

9.00 - 9.30 **Challenges in Variant Interpretation - How to minimize inter and intra-laboratory inconsistencies**

Andreas Laner
Medizinisch Genetisches Zentrum, Munich, Germany

9.30 - 10.00 **From a list of variants to a diagnostic report: extracting clinically relevant information**

María-Jesús Sobrido
Instituto de Investigaciones Sanitarias de Santiago / Fundación Pública Galega de Medicina Xenómica

10.00 - 10.30 **Accessing the full size-spectrum of human genetic variation using PacBio long-read SMRT sequencing on the Sequel System**

Luke Hickey
Senior Director of Human Biomedical Sciences, PacBio
PacBio is a Sponsor of the 14th International Symposium on Variants in the Genome

10.30 - 11.00 **Coffee Break**
Room: Quintana & Lobby

11.00 - 13.10	**PLENARY SESSION 5 - Population Genetics & Forensic Applications** **Room: Obradoiro** **Chair: Maria-Jesus Sobrido**
11.00- 11.30	**Challenges with the compilation and naming of new variation revealed by massively parallel sequencing of forensic markers** Christopher Phillips *University of Santiago de Compostela, Galicia, Spain*
11.30 - 12.00	**Naming Genetic Variation in Forensic Science: Alignment and Nomenclature of Next Generation Sequence Alleles** Walther Parson *President International Society of Forensic Genetics, Institute of Legal Medicine, Innsbruck, Austria*
	PRESENTATIONS FROM ABSTRACTS
12.00 - 12.20	**BaseSpace Variant Interpreter: A new platform to improve the speed of genomic interpretation and facilitate collaborative knowledge sharing** Jennifer Harrow *Programme Manager Population Sequencing, Illumina Inc., UK*
12.20 - 12.40	**Safe Variant Annotation Sharing Across Laboratories** Beat Wolf *HES - SO, Informatics, Fribourg, Switzerland*
12.40 - 13.10	**LGC SPONSORED PRESENTATION** **Variant detection and the challenges beyond: what is needed to implement genetic information for clinical use?** Prof. Dr. Daniela Steinberger *Human Geneticist, Medical Director, bio.logis Center for Humangenetics, Frankfurt am Main, Germany*
13.10 - 14.10	**Lunch Break** **Room: Azabache Restaurant**
14.10 - 15.40	**SPONSORED WORKSHOP - Alamut** **Room: Obradoiro** Andre Blavier Interactive Biosoftware *Please read the Workshop Summary before attending regarding requirements e.g. laptop required.* *Interactive Biosoftware is a sponsor of the 14th International Symposium on Variants in the Genome*
15.40 - 16.20	**Poster Session 2** **Room: Quintana & Lobby** ***Even numbers to present: POS002, POS004 etc.***

16.20 - 18.30 **WORKSHOP - Genetic Variant Interpretation**
Room: Obradoiro

Organizers:

- Andreas Laner
- Reece Hart
- Maria—Jesus Sobrido

18.30 **DAY END - Evening at leisure**

Wednesday 7th June

8.30 - 10.30 **PLENARY SESSION 6 - Bioinformatics And Big Data**
Room: Obradoiro
Chair: Angel Carracedo

8.30 - 9.00 **Challenges in bioinformatics for genetic diagnosis**

Joaquin Dopazo
Fundacion Progreso y Salud, Clinical Bioinformatics Research Area, Sevilla, Spain

9.00 - 9.30 **A community-developed data model for representing sequence variation**

Reece Hart
Invitae, San Francisco, USA

9.30 - 10.00 **KEYNOTE SPEAKER**

Meiosis, recombination, and the origin of a species

Peter Donnelly
Wellcome Trust Centre for Human Genetics, Oxford, UK

10.00 - 10.30 **Raising the bar in NGS diagnostics : Challenges in Variants Identification**

Zelie Dubreucq
Subject Matter Expert, at Sophia Genetics
Sophia Genetics is a Sponsor of the 14th International Symposium on Variants in the Genome

10.30 - 11.00 **Coffee Break**
Room: Quintana & Lobby

11.00 - 14.00 **PLENARY SESSION 7 - New Genomic Technologies**
Room: Obradoiro
Chair: Andreas Laner

11.00 - 11.30 **Strategies for assembling high quality genome sequences**

Ivo Gut
Centro Nacional de Análisis Genómico, CNAG-CRG, Barcelona, Spain

11.30 - 12.00 **Latest applications of innovative technologies in our research and diagnostics**

Johan T. den Dunnen
Leiden Univ. Medical Center, Leiden, Netherland

PRESENTATIONS FROM ABSTRACTS

12.00 - 12.20 **Accelerating diagnosis of hereditary diseases analytically with a knowledge network and the ACMG guidelines**

Ruth Burton
Qiagen, Redwood City, USA

12.20 - 12.50 **Rapid Genome Wide Mapping at the Single Molecule Level Using Nanochannel Arrays for Structural Variation Analysis and de novo Assembly**

Sven Bocklandt
Senior Application Specialist, BioNano Genomics
BioNano Genomics is a Sponsor of the 14th International Symposium on Variants in the Genome

12.50 - 13.50 **Lunch Break**
Room: Azabache Restaurant

13.50 - 15.20 **PLENARY SESSION 8 - Personalised Medicine & Pharmacogenomics**
Room: Obradoiro
Chair: Peter Donnelly

13.50 - 14.20 **Advancing Drug Discovery - Translating Small & Big Data Into Insight**

Guna Rajagopal
Global Head of Computational Sciences within Discovery Sciences Janssen Pharmaceuticals R & D, Spring House, PA, USA

14.20 - 14.50 **Innopharma: the Pharmacogenomics platform of the University of Santiago de Compostela**

Mabel Loza/ Ángel Carracedo
University of Santiago de Compostela, Galicia, Spain

14.50 - 15.20 **Advances in Personalised Medicine - Liquid Biopsy**

Alexander Sartori
Agena BioScience
Agena BioScience is a Sponsor of the 14th International Symposium on Variants in the Genome

15.20 - 15.50 **Coffee Break**
Room: Quintana & Lobby

15.50 - 17.00 **PLENARY SESSION 9 - Presentations From Selected Abstracts**
Room: Obradoiro
Chair: Ivo Gut

15.50 - 16.10 **PacBio long read sequencing for improved resolution of complex genomic variation**

Henk Buermans
Leiden Genome Technology Center, Leiden, Netherlands

16.10 - 16.30 **Tracing mitochondrial mutations in 3D in primary tumor, lymph node and liver metastasis**

Per Ekstrom
The Norwegian Radium Hospital, Oslo, Norway

16.30 - 16.50 **The TP53 mutation database: a paradigm for the analysis of cancer genes**

Thierry Soussi
Karolinska Institutet, Stockholm, Sweden

16.50- 17.00 **SUMMARY - CLOSING**

Johan T. den Dunnen, Maria Jesus Sobrido, Angel Carracedo

17.00 MEETING END