THE HUMAN VARIOME PROJECT
sharing data • reducing disease

United Nations Educational, Scientific and Cultural Organization

6th Biennial Meeting
UNESCO, Paris
30th May – 3rd June 2016

Sunday 29th May	
16.00 – 18.00	The Human Variome Project International Board Meeting Novotel Tour Eiffel

Monday 30th May	
9.00 – 9.30	Registration Global Globin 2020 Meeting ONLY UNESCO, 7 Place de Fontenoy
9.30 – 17.00	Global Globin 2020 Meeting Salle IV

Tuesday 31st May

Satellite Meetings			
9.00 – 11.00	Registration - Satellites & HVP6 UNESCO, 7 Place de Fontenoy		
9.00 – 10.45	**Global Globin 2020 Initiative** (meeting continued) Salle IV		
10.45 – 11.00	Coffee Mixer		
11.00 – 13.00	**LSDB Curation Course** Salle VII	**UNESCO Ethics & Standards Committee Meeting** Salle V	**Global Globin 2020 Initiative** (meeting continued) Salle IV
13.00 – 14.30	Lunch break at leisure		
14.00 – 15.30	**LSDB Curation Course** (continued) Salle VII		**Global Globin 2020 Initiative** (meeting continued) Salle IV
15.30 – 16.00	**Variant Nomenclature Workshop** Salle VII		**Global Globin 2020 Initiative** (meeting continued) Salle IV
16.00 – 16.20	Coffee Break		
16.20 – 17.30	**Variant Nomenclature** (continued) Salle VII		**Global Globin 2020 Initiative** (meeting continued) Salle IV

Wednesday 1st June

Salle IV	
9.00 - 9.30	Registration UNESCO, 7 Place de Fontenoy
9.30 – 15.30	**BRCA Workshop** **Salle TBA**
SESSION I	**HVP6 Begins** **Opening Addresses** **Chair: John Burn**
9.30 – 9.35	**Welcome Address** **Chris Arnold** Chair, Human Variome Project International Board
9.35 – 9.45	**Welcome** Ms Flavia Schlegel Assistant-Director of Natural Sciences Sector, UNESCO
9.45 – 10.05	**Report from the International Scientific Advisory Council** John Burn
10.05 – 10.35	**HVP Roadmap 2016 – 2020** Chris Arnold
10.35 – 10.50	Assemble for Group Photograph
10.50 – 11.15	Coffee Break
11.15 – 11.45	**Richard Cotton Memorial Lectures** **Sponsored by Interactive Biosoftware** Mike Watson & Finlay Macrae
SESSION II	**Ethical, Legal and Technical Frameworks for Variant Data Sharing** **Chair: Chris Arnold**
11.45 – 11.55	**The UNESCO/HVP Ethics and Standards Project** Chris Arnold
11.55 – 12.15	**Underlying Principles for National Variant Data Sharing Policies** **Discussion**
12.105– 12.30	**Legal and regulatory challenges to data sharing for clinical genetics and genomics services in the UK** Alison Hall
12.30 – 12.45	**Title TBA** Dafna Feinholz

12.45 – 14.00	Official Opening of Rick Guidotti's Positive Exposure: The Spirit of Difference Exhibition with lunch Introduction by Thomy de Ravel *La Salle des Pas Perdus*
SESSION III	**Global Globin 2020** **Chairs:** 1. **Prof. R Ramesar, *Division of Human Genetics, Dept of Clinical Laboratory Services, University of Cape Town, South Africa*** 2. **Prof. Z bin Alwi, *School of Medical Sciences, Universiti Sains Malaysia, Kelantan, Malaysia***
14.00 – 14.40	An overview of the GG2020 – what it is, aims, how it is designed and details on various countries participating in GG2020 and how they currently deliver health services: • Examples from participating countries • Example of countries working together in regional groups: Central and South America, S E Asia
14.40 – 15.00	Specific Challenges for LMICs in genetics/genomics and public health, clinical practice arising from this HVP project-wide initiative
15.00 – 15.15	Report on what has been achieved so far including the outcomes of 30-31 May meeting
15.15 – 15.30	Questions & Discussion
15.30 – 16.00	**Poster Session: Poster *presenters to stand by their poster***
SESSION IV	**BRCA Challenge** **Chairs:** Prof.Diana Eccles, *Univ of Southampton UK* & Prof. Sir John Burn, *Newcastle Univ., UK*
16.00 – 16.10	The BRCA Challenge John Burn
16.10 – 16.30	**Developing and sustaining national databases: Experience with BRCA Share** Dr Claude Houdayer INSERM Paris
16.30 – 16.50	BRCAexchange: the response to the BRCA Challenge Dr Gunnar Ratsch USA

16.50 – 17.10	Collecting and Sharing genomic data safely Dr Jem Rashbass (Public Health England)
17.10 – 17.30	Panel Discussion
17.30 – 19.30	**Welcome Reception** 7th Floor Restaurant, UNESCO

Thursday 2nd June	
7.00 – 8.30	**International Scientific Advisory Committe Meeting (***by invitation only***)** *Novotel Tour Eiffel Hotel*
SESSION V	**Country Nodes** **Chair: Martina Witcsh-Baumgartner** **Salle IV**
9.00 – 9.20	**Report from International Confederation of Country Nodes Council** Martina Witcsh-Baumgartner, Chair
9.20 – 9.25	**Update on the Netherlands** Marielle van Gijn
9.25 – 9.30	**Update on the Malaysian Node** Zilfalil bin Alwi
9.30 – 9.35	**Progress at the Canadian Node** Matthew Lebo
9.35 – 9.40	**What's happening in Belgium and The Congo** Thomy de Ravel de l´Argentière
9.40 – 9.45	**Progress & Challenges at the Nigerian Node** Dr Oyekanmi Nash
9.45 – 9.50	**Status of Nepalese HVP country node: progress, specific issues and challenges in Nepal** Prof. Tilak Shrestha
9.50 – 9.55	**Kuwait and Lebanon, status report** Fahd Al-Mulla
9.55 – 10.30	**Discussion**
10.30 – 11.00	**Coffee Break**
SESSION VI	**Gene & Disease Specific Databases** **Chair: Peter Taschner** **Salle IV**
11.00 – 11.35	**Report from the Gene and Disease Specific Database Advisory Council** Peter Taschner, Chair
11.35 – 11.50	**Ratio of harmful amino acid substitutions: Case studies of BTK and MMR system** Mauno Vihinen
11.50 – 12.05	**Pathogenicity Decision Pathway in TSC**

	Rosemary Ekong	
12.05 – 12.30	**Discussion**	
12.30 – 14.00	Lunch break at leisure	
SESSION VII	**Sharing & Challenges** **Chair: Qasim Ayub** **Salle IV**	
14.00 – 14.20	**High-security DNA bank accounts to protect and share your genetic identity** Johan T. den Dunnen	
14.20 – 14.40	**Establishing validity, reproducibility, and utility of highly scalable genetic tests: Challenges and Solutions** Stephen Lincoln	
CONCURRENT SESSIONS 1	**Databases: Creation, Content, Curation** Chair: Marc Greenblatt Salle IV	**Knowledge** Chair: Johan T. den Dunnen Salle VII
14.45 – 15.00	**Human Variome Project quality assessment criteria for variation databases** Mauno Vihinen	**Checking the experts: compliance with author instructions regarding HGVS nomenclature and variant submission to databases in genetics and genomics journals** Jim Osmani
15.00 – 15.15	**Trust in digital data: the reliability, authenticity and accuracy of genomic variant data** Timothy Smith	**HIPBI-RD: Harmonising phenomics information for a better interoperability in the rare disease field** Ana Rath
15.15 – 15.30	**Towards formal specification of HGVS nomenclature enabling computational tool development** Jonathan Vis	**Diagnostic application of a tool to detect single exon copy number variations in targeted next-generation sequencing data** Annemieke van de Hout
15.30 – 16.00	Coffee Break	
SESSION VIII	**Database Updates** **Chair: Mauno Vihinen**	

	Salle IV
16.00 – 16.15	**Sharing dog exome/genome variant and phenotype data in Varda and LOVD3** Peter Taschner
16.15 – 16.30	**ClinVar: Aggregating Data to Improve Variant Interpretation** Melissa Landrum
16.30 – 16.45	**Data sharing in Canada through the Canadian Open Genetics Repository (COGR): a unified clinical genome database as a community resource for standardizing and sharing genetic interpretations** Matthew Lebo
16.45 – 17.00	**The PhenX Toolkit: Standard Measures for Collaborative Research** Wayne Huggins
17.00 – 17.15	**European Hereditary Tumour Group (former Mallorca Group) hosts The Prospective Lynch Syndrome Database and the European MMR cDNA Working Group and is open for all to join** Pal Moller
17.15 – 17.30	**kConFab: A national familial breast cancer consortium facilitating research** Heather Thorne
17.30 – 19.00	Free Time
19.00 -	**R.G.H Cotton Memorial Dinner Cruise (*Separate ticket required*)**

Friday 3rd June		
CONCURRENT SESSIONS 2	**Variation Interpretation & Pathogenicity Assessment** Chair: Finlay Macrae Salle IV	**HUGO SESSION** Title TBA Chair: TBA Salle VII
9.00 – 9.15	**Phenotype-based Matching Using PhenoDB Terms in BHCMG PhenoDB to Maximize Whole Exome/Genome Sequencing Data Interpretation** Nara Sobreira	**Rare and common variants, coding and non-coding, contribute to the complex inheritance of Hirschsprung disease** Stanislas Lyonnet Hôpital Necker-Enfants Malades, France
9.15 – 9.30	**Variation Ontology (VariO) for systematic annotation of variation effects and mechanisms** Mauno Vihinen	**TBA** **Not available at time of printing**
9.30 – 9.45	**The ClinGen Sequence Variant Interpretation Working Group- Refining Criteria for Interpreting the Pathogenicity of Genetic Variants** Marc Greenblatt	**TBA** **Not available at time of printing**
9.45 – 10.00	**Clinical Classification of Mismatch Repair Gene Variants by Multiple Lines of Evidence: the Contribution of Individual Components** Maurizio Genuardi	**TBA** **Not available at time of printing**
10.00 – 10.30	**Variations in LSDBs; when can they be considered pathogenic?** **Open Discussion**	
10.30 – 10.50	**Meeting summary and thoughts – John Burn**	
10.50	**MEETING END**	
10.50 – 11.10	**Coffee**	
11.10 – 13.00	**COUNCIL MEETINGS** **Salle IV**	

11.10 – 13.00 **ICCAC & GDSDBAC Joint Meeting (ISAC members welcome)**