PROGRAMME

Monday 19th May

Satellite Meetings

8.30	Registration UNESCO, 7 Place de Fontenoy			
9.30 – 11.00	**LSDB Curation Course** Salle III		**Breast Cancer Interest Group** Salle IV (by invitation only)	**Oesophageal Cancer Meeting** Salle V (by invitation only)
11.00 – 11.30	Coffee Break			
11.30 – 13.00	**LSDB Curation Course** Salle III	**HVP Country Node Establishment Workshop: new directions** Salle VI	**Breast Cancer Interest Group** Salle IV (by invitation only)	**Oesophageal Cancer Meeting** Salle V (by invitation only)
13.00 – 14.30	Lunch break at leisure			
14.30 – 16.00	**LSDB Curation Course** Salle III	**HVP Country Node Establishment Workshop: new directions** Salle VI	**Breast Cancer Interest Group** Salle IV (by invitation only)	
16.00 – 16.30	Coffee Break			
16.15 – 18.00	**Joint meeting of Gene and Disease Specific Database Advisory Council with International Confederation of Countries Advisory Council** Salle IV			

dmd [MM Paris Booklet]2014.indd 5 1/05/14 12:29 AM

Tuesday 20th May

8.30 – 9.30	Registration UNESCO, 7 Place de Fontenoy
9.00	**HVP Breast Cancer interest Group** **Salle III** *(by invitation only)*

Human Variome Project – 5th biennial meeting
Salle IV

SESSION I — WELCOME & INFORMATION
CHAIR: JOHN BURN

WELCOME ADDRESS

David Abraham
Chair, Human Variome Project International Board

Richard Cotton
Scientific Director, Human Variome Project International

9.35 – 9.45	**WELCOME ADDRESS** **Wendy Watson-Wright** Assistant Director-General for Natural Sciences *ad interim*, UNESCO
9.45 – 10.00	**Report from the International Scientific Advisory Council** Garry Cutting, Chair
10.00 – 10.15	**Report from Gene/Disease Specific Database Advisory Council** Raymond Dalgleish, Chair
10.15 – 10.30	**Report from International Confederation of Countries Advisory Council** Martina Wisch-Baumgartner, Chair
10.30 – 11.15	**Global Alliance** Peter Goodhand
11.15 – 11.25	**Assemble for group photograph** Foyer outside Salle IV
11.15 – 11.45	Coffee Break

SESSION II — COLLABORATION BETWEEN DEVELOPED AND DEVELOPING COUNTRIES
CHAIR: MARTINA WISCH-BAUMGARTNER

11.45 – 12.05	**Datasharing in the Belgium/Congo** Thomy de Ravel
12.05 – 12.25	**The Human Variome Project Australian Node in 2014** Graham Taylor
12.25 – 12.45	**A Human Variome Project Country Node: Taking the HVPA-BioGrid Experience as a Data Sharing Model** Nik Nor Liza Nik Hassan

dmd [MM Paris Booklet]2014.indd 6 1/05/14 12:29 AM

12.45 – 13.00	**Report from Minimum Content of a Country Specific Variant Database** Martina Wisch-Baumgartner
13.00 – 14.20	Lunch break at leisure

SESSION III — CURRENT DEVELOPMENTS IN AFRICA
CHAIR: RAJ RAMESAR

14.20 – 14.30	**Introduction speech** Lalla Aïcha Ben Barka Assistant Director-General for Africa, UNESCO
14.30 – 14.35	**Introduction to H3Africa** Raj Ramesar
14.35 – 14.50	**H3Africa Kidney Disease Research Network** (GHANA) Dwomoa Adu
14.50 – 15.05	**Clinical and Genetic Studies of Hereditary Neurological Disorders in Mali** (MALI) Guida Landoure
15.05 – 15.20	**Host and Microbial Genetic Determinants of febrile illness in West Africa** (CAMEROON, NIGERIA) Christian Happi
15.20 – 15.35	**Collaborative African Genomics Network (CAfGEN)** (BOTSWANA, UGANDA) Gabriel Anabwani
15.35 – 15.50	**TrypanoGEN: An integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis** (UGANDA) Enock Matovu
15.50 – 16.05	**Development of H3 Africa Biorepositories to facilitate studies on Biodiversity, Disease & Pharmacogenomics of African Populations** (SOUTHERN AFRICA) Akin Abayomi
16.05 – 16.20	**Visual Analytical Screening System for Disease Linked Gene Variants** (NIGERIA) Oyekanmi Nash
16.20 – 17.00	Poster Session 1 – Odd numbers (1, 3, 5 ...) o Poster presenters for ODD NUMBERS to stand by their poster

SESSION IV — INTEGRATION OF NGS DATA
CHAIR: HEIDI REHM

17.00 – 17.20	**Informatics infrastructure for secure access, visualization and analysis of NGS data** Ted Kalbfleisch
17.20 – 17.40	**Next-generation variant sharing between Dutch genome diagnostic laboratories** Moris Swertz
17.40 – 18.00	**Dealing with Next Generation sequencing data not generated in the TSC context** Rosemary Ekong

dmd [MM Paris Booklet]2014.indd 7 1/05/14 12:29 AM

18.00 – 19.30	**Welcome Reception** 7th Floor Bar

Wednesday 21st May

Salle IV

SESSION V — DATABASES AND DATA CURATION 1
CHAIR: JOHAN T. DEN DUNNEN

9.30 – 9.45	**Cracking rare disease: sharing exome/genome variant and phenotype data in LOVD3** Peter Taschner
9.45 – 10.00	**Lightweight integration tool for data collection** Andrei Loukianov
10.00 – 10.15	**Microattributions – increasing incentives for data submissions to Locus-Specific Databases** John-Paul Plazzer
10.15 – 10.30	**Whole Exome and Genome Sequencing Analysis (PhenoDB) and Gene Matching Tool for Identification of Individuals with Mutations in the Same Gene (GeneMatcher)** Ada Hamosh
10.30 – 10.45	**Cafe Variome: connecting diagnostic networks, disease consortia and diverse third parties** Raymond Dalgleish
10.45 – 11.00	**MUTALYZER 2: Improved Sequence Variant Descriptions from next generation sequencing data and gene variant databases** Peter Taschner
11.00 – 11.10	**Report from WG03: Minimal content for gene variant databases (LSDBs)** Peter Taschner
11.10 – 11.30	Coffee Break

SESSION VI — CONCURRENT WORKSHOPS

Workshop A 11.30 – 13.00 Salle IV	**InSiGHT Interpretation Committee** Chair: Finlay Macrae Observers welcome
Workshop B 11.30 – 13.00 Salle III	**Institutional Support for Data Sharing** Chair: Richard Cotton
11.30 – 11.45	**Report of World Health Organisation Project** Helen Robinson
11.45 – 12.00	**Ethical issues** Dafna Fienholz, UNESCO

dmd [MM Paris Booklet]2014.indd 8 1/05/14 12:29 AM

12.00 – 12.30	**Attracting EC Funding** Diana Salmen, European Commission
12.30 – 13.00	**OPEN DISCUSSION**
13.00 – 14.30	Lunch break at leisure
13.00 – 14.30	**WG04: Sequence variant description committee meeting** Salle III (by Invitation only)

SESSION VII — DATABASES AND DATA CURATION 2
CHAIR: MAUNO VIHINEN

14.30 – 14.45	**ClinGen: The Clinical Genome Resource** Heidi Rehm
14.45 – 15.00	**ClinVar: A Central Repository for Clinically Relevant Variants** Melissa Landrum
15.00 – 15.15	**MseqDR consortium: a grass-roots effort to establish a global resource aimed to empower genomic studies of mitochondrion diseases** Marcella Attimonelli
15.15 – 15.30	**MSV3d 2014: Database of human MisSense Variants mapped to 3D protein structure** Hoan Nguyen
15.30 – 15.40	**Report from WG05: Variant Database Quality Assessment** Mauno Vihinen
15.40 – 15.50	**Report from WG01: Disclaimer Statements on G/DSDBs Websites** John Paul Plazzer
15.50 – 16.00	**Report from WG04: Sequence variant description committee** Johan T. den Dunnen
16.00 – 16.30	Coffee Break

SESSION VIII — HUGO SESSION
CHAIR: STYLIANOS ANTONARAKIS

16.30 – 17.00	**The Renaissance of Personalized Medicine** Stylianos Antonarakis
17.00 – 17.30	**Functional annotation of personal genomes: transcriptome analysis in the GTEx project** Tuuli Lappalainen
17.30 – 18.00	**Clinical bioinformatics; a paradigm change in medicine** Jacques Beckmann

dmd [MM Paris Booklet]2014.indd 9 1/05/14 12:29 AM

Thursday 22nd May

Salle IV

SESSION IX	ASSESSMENT OF PATHOGENICITY OF VARIANTS 1 CHAIR: MARC GREENBLATT
9.30 – 9.50	**Development of rules for the interpretation of mismatch repair gene variants based on the 5-tiered IARC classification system** Maurizio Genuardi
9.50 - 10.10	**Using The InSiGHT Consensus Criteria for Classification of Mismatch Repair Gene Variants to Validate New Lines of Evidence for Use in the Classification Process** Marc Greenblatt
10.10 – 10.30	**Progress of the InSiGHT Mismatch Repair Gene Database and Variant Interpretation Committee – Application of Classification Rules and Dissemination of Results** Finlay Macrae
10.30 – 10.50	**Exonic splicing mutations in Mendelian disorders: more prevalent than currently estimated** Alexandra Martins
10.50 – 11.00	**Report from WG02: Assigning Pathogenicity to a Genetic Variant** Marc Greenblatt
11.00 – 11.40	Poster Session 2 – Even numbers (2, 4, 6 ...) • Poster presenters for EVEN NUMBERS to stand by their poster

SESSION X	ASSESSMENT OF PATHOGENICITY OF VARIANTS 2 CHAIR: MIKE WATSON
11.40 – 12.00	**Variant Server in a box** Joeri van der Velde
12.00 – 12.20	**A powerful literature review: impact index, a gene-based bibliomic pathogenicity score** Christopher Cassa
12.20 – 12.40	**A collaborative database for prospective studies in Lynch syndrome: First results and invitation to join** Pal Moller
12.40 – 13.00	**Is the assessment of pathogenicity of a variant in isolation appropriate for clinical application in the context of multigenic analyses of complex genetic pathology?** Desiree du Sart
13.00 – 14.30	Lunch break at leisure

dmd [MM Paris Booklet]2014.indd 10 1/05/14 12:29 AM

SESSION XI	PHENOTYPE AND TRANSLATION TO THE CLINIC CHAIR: ADA HAMOSH
14.30 – 14.50	**Clinical Significance of Transcript Alignment Discrepancies** Reece Hart
14.50 – 15.10	**The Curation of Molecular Pathology Cancer Samples** Kenneth Doig
15.10 – 15.30	**VarElect: phenotype-based variation prioritizer in GeneCards** Gil Stelzer
15.30 – 15.50	**Linking Genomic and Clinical Data: The HVPA-BioGrid Experience** Maureen Turner
15.50 – 16.10	**Phenotype terminologies in use for genotype-phenotype databases: need for a standardization** Ana Rath
16.10 – 16.20	**Report from WG06: Disease & Phenotype Descriptions in Gene/Disease Specific Databases** Peter Robinson
16.20 – 16.40	Coffee Break
16.40 – 17.30	**Meeting Summary and thoughts** Garry Cutting
	END HVP 5th biennial meeting

dmd [MM Paris Booklet]2014.indd 11 1/05/14 12:29 AM

Friday 23rd May

Advisory Council and International Scientific Advisory Committee Meetings

9.30 – 11.00	**Combined HVP Council Meetings** **• Gene/Disease Specific Database Advisory Council Meeting** **• International Confederation of Countries Advisory Council Meeting** **• International Scientific Advisory Council** Salle IV (all welcome)
11.00 – 11.15	Coffee Break
11.15 – 13.00	**Gene/Disease Specific Database Advisory Council Meeting** Salle IV (all welcome) **International Confederation of Countries Advisory Council Meeting** Salle III (all welcome) **MEETING END**

dmd [MM Paris Booklet]2014.indd 12 1/05/14 12:29 AM