



THE HUMAN VARIOME PROJECT
COLLECTION OF HUMAN GENE VARIATION

PLANNING MEETING

In Association with

Genomic Disorders Research Centre (GDRC) – HVP Coordinating Office
United Nations Educational, Scientific and Cultural Organization (UNESCO)
American College of Medical Genetics (ACMG)
Genotype 2 Phenotype (GEN2PHEN), A consortium of the EC
Human Genome Variation Society (HGVS)
International Society for Gastrointestinal Hereditary Tumours (InSiGHT)

25 – 29 May 2008
Hotel Eden Roc
San Feliu de Guixols, Spain

Co-Chairs: Richard G.H. Cotton and Michael S. Watson



Genomic Disorders Research Centre



Day 1 – Sunday May 25th

- 15.00 – 18.00 Registration
- 19.00 – 21.30 Welcome Reception & Dinner

Day 2 – Monday May 26th

Session Chair
Richard Cotton

- 8.30 – 8.40 Welcoming address & session objectives**
- Speaker:** Richard Cotton, Convenor, Human Variome Project, Melbourne, Australia.
- 8.40 – 9.10 Keynote Lecture**
Myles Axton, Editor *Nature Genetics*
Microattribution for community annotation of the human genome
- 9.10 – 9.30 **Overview: Pilot Projects**
Finlay Macrae / Samir Brahmachari
- 9.30 – 9.40 Questions/Discussion
- 9.40 – 10.00 **Overview: Data Collection from Clinics**
XXXX / Gabriela Möslein
- 10.00 – 10.10 Questions/Discussion
- 10.10- 10.30 **Overview: Data Collection from Laboratories**
Graham R. Taylor / C. Sue Richards
- 10.30 – 10.40 Questions/Discussion
- 10.40 – 11.00 Coffee Break**

- 11.00 – 11.20 **Overview: Data Transfer & Databasing (Gene Specific/LSDB)**
Johan T. den Dunnen / Arleen D. Auerbach
- 11.20 – 11.30 Questions/Discussion
- 11.30 – 11.50 **Overview: Overall Data Integration & Access**
Donna R. Maglott / Anthony J. Brookes
- 11.50 – 12.00 Questions/Discussion
- 12.00 – 12.20 **Overview: Assessment of Pathogenicity**
Marc Greenblatt / Sean Tavtigian
- 12.20 – 12.30 Questions/Discussion
- 12.30 – 12.50 **Overview: Publication, Credit & Incentives**
Myles Axton / John Burn
- 12.50 – 13.00 Questions/Discussion
- 13.00 – 14.00 Lunch**
- Session Chair**
Michael Watson
- 14.00 – 14.30 Keynote Lecture**
Garry Cutting, Johns Hopkins University School of Medicine
Complementing Mutation-Driven Databases with Phenotype-Rich Repositories: The Clinical and Functional Translation of CFTR (CFTR2) Project
- 14.30 – 14.50 **Overview: Developing/Emerging Countries & Worldwide**

Collection

Joji Utsunomiya / Raj Ramesar

14.50 – 15.00 Questions/Discussion

15.00 – 15.20 **Overview: Funding Mechanisms & Governance**
Michael Katz / Richard Cotton / Daniela Seminara

15.20 – 15.30 Questions/Discussion

15.30 – 16.00 Keynote Lecture
James O'Leary, COO, Genetic Alliance
Openness and Collaboration in the Variome

16.00 – 16.30 Coffee break

16.30 – 16.50 **Overview: Ethics**
Sue Povey / Jan Helge Solbakk

16.50 – 17.00 Questions/Discussion

SESSIONS COMMENCE

17.00 – 19.30 ETHICS
Chairs: Sue Povey / Jan Helge Solbakk

17.00 – 17.30 **Sue Povey**
Ethical issues arising in the curation of locus-specific variation databases

17.30 – 18.00 **Jan Helge Solbakk**
Why research biobanks are still an ethical challenge

18.00 – 18.30 **Anne Cambon-Thomsen**
Sharing samples and data: ethics embedded in the tools, the tricks and the incentives

18.30 – 19.30 **Open discussion**

19.30 – 20.00 Free Time

20.00 – 22.00 Dinner

Day 3 – Tuesday May 27th

8.30 – 10.30 PILOT PROJECTS
Chairs: Finlay Macrae / Samir Brahmachari

8.30 – 8.40 **Finlay Macrae**
The International Society for Gastrointestinal Hereditary Tumours' (InSiGHT) collaboration with the Human Variome Project (HVP)

8.40 – 8.50 **Samir Brahmachari**
TBA

8.50 – 9.00 **Inge Bernstein**
System development, implementation and experiences from a pilot study dealing with electronic exchange of data on phenotype – genotype in families with Hereditary Colon Cancer

9.00 – 9.10 **Steven Brenner**
The Genome Commons and The Genome Commons Navigator

9.10 – 9.20 **Fahd Al-Mulla**
Kuwait genome/variome project: linking clinicians and researchers to the world

9.20 – 9.30 **Dagmar Scheilbe**
Longterm follow-up of patients with rare metabolic disorders by the RAMEDIS database

9.30 – 9.40 **James Kaput**
Analyzing human genetic variation and food cultures: developing Nutrigenomics research for personalized nutrition and medicine

9.40 – 9.50 **Joji Utsunomiya**
An attempt to construct cancer genetic medicine infrastructure in Japan

9.50 – 10.00 **Judith Savige**
Cataloging genetic variants in X-linked Alport syndrome is enhanced where their effect on structural sequences and ligand-binding motifs is evident in an ‘interactome’

10.00 – 10.10 **Peter J. Oefner**
Validation of microarray- based resequencing of 93 worldwide mitochondrial genomes

10.10 – 10.30 **Open discussion**

Other abstracts to be discussed

David Hansen

Towards an architecture to support collaborative sharing and accessing of human mutation data

Peter Propping

The German HNPCC Consortium: Structure, Aims and Accomplishments

10.30 – 11.00 Coffee Break

11.00 – 13.00 CONTINUING SESSION: PILOT PROJECTS

Open discussion

13.00 – 14.30 Lunch

14.30 – 16.30 CONCURRENT SESSIONS 1, 2 & 3

CONCURRENT SESSION 1	CONCURRENT SESSION 2	CONCURRENT SESSION 3
Data Transfer & Databasing (Gene Specific; LSDB)	Assessment of Pathogenicity	Developing/Emerging Countries & Worldwide Collection
Chairs:	Chairs:	Chairs:

Johan T. den Dunnen / Arleen D. Auerbach	Marc Greenblatt / Sean Tavtigian	Joji Utsunomiya / Raj Ramesar
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16.30 – 17.00 Coffee break

17.00 – 19.00 CONTINUING CONCURRENT SESSIONS 1, 2 & 3

Data Transfer & Databasing (Gene Specific; LSDB)	Assessment of Pathogenicity	Developing/Emerging Countries & Worldwide Collection
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19.00 – 20.00 Free time

20.00 – Gala Dinner

Day 4 - Wednesday May 28th

8.30 – 10.30 CONCURRENT SESSIONS 4, 5 & 6

CONCURRENT SESSION 4	CONCURRENT SESSION 5	CONCURRENT SESSION 6
Funding Mechanisms & Governance	Data Collection from Laboratories	Overall Data Integration and Access
Chairs: Michael Katz / Richard Cotton / Daniela Seminara	Chairs: Graham R. Taylor / C. Sue Richards	Chairs: Donna R. Maglott / Anthony J. Brookes

10.30 – 11.00 Coffee Break

11.00 – 13.00

**NEW CONCURRENT SESSIONS 7 & 8 & CONTINUING
CONCURRENT SESSION 6**

CONCURRENT SESSION 7	CONCURRENT SESSION 8	CONTINUING CONCURRENT SESSION 6
Publication, Credit & Incentives	Data Collection from Clinics	Overall Data Integration and Access
Chairs: Myles Axton / John Burn	Chairs: XXX / Gabriela Möslein	

13.00 – 14.30

Lunch

Session Chairs

Richard Cotton / Michael Watson

14.30 – 16.35

**REPORT BACKS BY DISCUSSION LEADERS
(Questions and open discussion)**

14.30 – 14.55

Ethics

14.55 – 15.20

Pilot Projects

15.20 – 15.45

Data Transfer & Databasing

15.45 – 16.10

Assessment of Pathogenicity

16.10 – 16.35

Developing/Emerging Countries

16.35 – 17.05

Coffee Break

17.05 – 19.10

REPORT BACKS BY DISCUSSION LEADERS CONTINUED

17.05 – 17.30	Funding Mechanisms & Government
17.30 – 17.55	Publication, Credit & Incentives
17.55 – 18.20	Data Collection from Laboratories
18.20 – 18.45	Data Collection from Clinics
18.45 – 19.10	Overall Data Integration and Access
19.10 – 19.30	Free time
19.30 –	Dinner

Day 5 – Thursday May 29th

Session Chairs

Richard Cotton / Michael Watson

9.00 – 11.50	Summation Circulation of headings for three draft pages by discussion leaders and questions
9.00 – 9.15	Circulation of papers
9.15 – 9.30	Ethics
9.30 – 9.45	Pilot Projects
9.45 – 10.00	Data Transfer & Databasing
10.00 – 10.15	Assessment of Pathogenicity
10.15 – 10.30	Developing/Emerging Countries
10.30 – 10.45	Funding Mechanisms & Government
10.45 – 11.00	Publication, Credit & Incentives
11.00 – 11.15	Data Collection from Laboratories
11.15 – 11.30	Data Collection from Clinics
11.30 – 11.45	Overall Data Integration and Access
11.45 – 12.00	Conclusion

12.00 – Lunch

After Lunch

Optional Excursion to the Dali Museum in Figueras & return to hotel before departure

Program for Concurrent Sessions

Day 3 – Tuesday May 27th

- 14.30 – 16.30 **CONCURRENT SESSION 1**
Data Transfer & Databasing (Gene Specific; LSDB)
- Chairs:** Johan T. den Dunnen / Arleen D. Auerbach
- 14:30 – 14.45 **Mireille Claustres**
UMD-LSDBs: From genomic annotation to clinical trials
- 14:45 – 15.00 **Johan T. den Dunnen**
Web-based collection of gene-specific sequence variants and their phenotypic consequence
- 15:00 – 15.15 **Lawrence Cavedon**
Enabling more efficient LSDB curation with more effective automatic search and text mining tools
- 15:15 – 15.30 **Tim Smith**
VariVis: A visualisation toolkit for variation databases.
- 15:30 – 15.45 **Jon Teague**
COSMIC, data standards and handling next generation sequencing data
- 15.45 – 16.00 **Arleen D. Auerbach**
Conversion of an LSDB to a genotype-phenotype repository: the Fanconi anemia mutation database experience
- 16.00 – 16.15 **Ming Qi**
Bio-bank and genetic database of major diseases in China
- 16.15 – 16.30 **Steven Dobrowolski**
Traditional single gene mutation databases, their strengths,

shortcomings, and means by which these may be scrutinized to increase their utility

16.30 – 17.00

Coffee break

17.00 – 19.00

CONTINUING CONCURRENT SESSION 1
Data Transfer & Databasing (Gene Specific; LSDB)

Open Discussion

Other abstracts to be discussed

Carsten Bermann

LSDB for PKHD1 in autosomal recessive polycystic kidney disease (ARPKD)

David Cooper

The Human Gene Mutation Database (HGMD®)

1. LSDB software/tools.
2. Lessons from LSDB disease-specific curation.
3. National Data Collections.
4. General Discussion and Summation.

14.30 – 16.30

CONCURRENT SESSION 2
Assessment of Pathogenicity

Chairs: Marc Greenblatt / Sean Tavtigian

14:30 – 15.00

Mauno Vihinen

Pathogenic or not? Analysis of the effects of missense mutations by bioinformatics methods

15:00 – 15.30

Rolf Sijmons

Database to support the clinical interpretation of human mismatch repair gene variants

15:30 – 16.00

Maurizio Genuardi

Assessment of variants of undetermined significance in mismatch repair genes: role of the InSiGHT/Human Variome Project

- 16:00 – 16.30 **Sean Tavtigian and Marc Greenblatt**
Analysis of unclassified variants in BRCA1 and BRCA2: the breast cancer information consortium (BIC) approach and planned extension to other high-risk cancer susceptibility genes
- 16.30 – 17.00 **Coffee break**
- 17.00 – 19.00 **CONTINUING CONCURRENT SESSION 2**
Assessment of Pathogenicity
- Open discussion**
- Other abstracts to be discussed**
- Peter Rogan**
Translation of research mutation databases and software resources for clinical diagnosis: US regulatory considerations

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- 14.30 – 16.30 **CONCURRENT SESSION 3**
Developing/Emerging Countries & Worldwide Collection
- Chairs:** Joji Utsunomiya / Raj Ramesar
- 14:30 – 14.50 **Joji Utsunomiya**
Developing/Emerging countries and world collection
- 14:50 – 15.10 **Ana Maria Oller de Ramirez**
The mission of an Argentinean genetic center in the Human Variome Project (HVP).
- 15:10 – 15.30 **Makia Marakie**
Kuwait genome/variome project: our vision and future prospect
- 15:30 – 15.50 **Jumana Al-Aama**
Obstacles to Genetic Data Collection in Emerging Countries: Experience in Saudi Arabia
- 15:50 – 16.10 **Raj Ramesar**
Genomic variation on the African continent
- 16.10 – 16.20 **Maria Golubenko**

Monogenic and common diseases in populations of north Asia
– phenotype and genetic data collection

16.20 – 16.30 **Open discussion**

16.30 – 17.00 **Coffee break**

17.00 – 19.00 **CONTINUING CONCURRENT SESSION 3**
Developing/Emerging Countries & Worldwide Collection

Open discussion

Day 4 - Wednesday May 28th

8.30 – 10.30 **CONCURRENT SESSION 4**
Funding Mechanisms & Governance

Chairs: Michael Katz / Richard Cotton / Daniela Seminara

8.30 – 8.40 **Richard G.H. Cotton**
Uniting those interested in mutation collection worldwide

8.40 – 8.50 **David Quin**
Obtaining global agreement on communication in genetics:
the Human Variome Project role as an interface between
researchers and the international standards organization

8.50 – 9.00 **Richard G.H. Cotton**
How to link new and existing projects with the Human
Variome Project

9.00 – 9.10 **Richard G.H. Cotton**
LSDB sponsorship as a mechanism to fund curators “Adopt A
Gene”

9.10 – 9.20 **Jong Bhak**
\$0 genomics project towards variomics slogan: “Open-free
Personal Genome”

9.20 – 9.30 **Daniela Seminara**
TBA

9.30 – 10.30 **Open discussion**

10.30 – 11.00 Coffee Break

**8.30 – 10.30 CONCURRENT SESSION 5
Data Collection from Laboratories**

Chairs: Graham R. Taylor / C. Sue Richards

8.30 – 8.45 **C. Sue Richards**
The Clinical Laboratory: 2008 Progress Report for U.S.
Laboratories

8.45 – 9.00 **Graham R. Taylor**
DMuDB: A Variome Database for Genetics Service
Laboratories in the UK

9.00 – 9.15 **Simon B. Flanagan**
Inter-laboratory variation in the molecular diagnosis of triplet
repeat disorders

9.15 – 9.30 **Maija Kohonen-Corish**
Mapping of epigenetic sites in cancer

9.30 – 10.30 **Open discussion**

10.30 – 11.00 Coffee Break

**8.30 – 10.30 CONCURRENT SESSION 6
Overall Data Integration and Access**

Chairs: Donna R. Maglott / Anthony J. Brookes

8.30 – 8.40 **Jong Bhak**
Human variome resource and infrastructure

8.40 – 8.50 **Paul Flicek**

- The role of European bioinformatics institute in the Human Variome Project
- 8.50 – 9.00 **Anthony J. Brookes**
HGVBase-G2P: A central genetic association study database
- 9.00 – 9.10 **Marta Gwinn**
Knowledge Base on Genetic Variation and Human Health: HuGE Navigator
- 9.10 – 9.20 **Ross Hardison**
Two paths to central repositories: PhenCode and dbSNP
- 9.20 – 9.30 **Terence M. Harrison**
The Evidence Based Medicine and Human Genetics search interface: where to next?
- 9.30 – 9.40 **Jongsun Jung**
New storing and databasing technology for variome database
- 9.40 – 9.50 **Donna R. Maglott**
Managing variome data at the National Center for Biotechnology Information (NCBI)
- 9.50 – 10.00 **William S. Oetting**
The Data Interoperability Steering Committee (DISC)
- 10.00 – 10.10 **Mollie Ullman-Cullere**
Structuring Clinical Genetic Data in the Electronic Health Record
- 10.10 – 10.20 **Yoon Shin Cho**
Korea Association Resource Database (KAReDB), a user friendly interactive browser for Korea genome-wide association study
- 10.20 – 10.30 **John Hancock**
Man and mouse – proposed collaboration with the CASIMIR project on syntactic and semantic interoperability of human and mouse databases
- 10.30 – 11.00 **Coffee Break**

11.00 – 13.00 **CONTINUING CONCURRENT SESSION 6**
Overall Data Integration and Access

Open discussion

Other abstracts to be discussed

George Patrinos

GEN2PHEN: An international effort towards the universal databasing of gene-disease relationships

11.00 – 13.00 **CONCURRENT SESSIONS 7**
Publication, Credit & Incentives

Chairs: Myles Axton / John Burn

11.00 – 11.15 **Richard G.H. Cotton**
Inducements, aids, guidelines and rules to ensure collection of mutation/variation useful in research, diagnostics and prognostics

11.15 – 11.30 **George Patrinos**
Database-journals: The example of *FINDbase* for connecting a database with a scientific journal

11.30 – 13.00 **Open discussion**

13.00 – 14.30 **Lunch**

11.00 – 13.00 **CONCURRENT SESSION 8**
Data Collection from Clinics

Chairs: **xxxx** / Gabriela Möslein

11.00 – 11.15 **Gabriela Möslein**
TBA

11.15 – 11.30 **TBA**

TBA

11.30 – 11.45 **Paola Carrera**
Clinical-molecular data-base and definition of diagnostic flowcharts: the example of Charcot-Marie-Tooth neuropathies

11.45 – 12.00 **Rosemary Ekong**
Our experience of collecting data for *tsc1* and *tsc2* databases from diagnostic laboratories

12.00 – 12.15 **David Quin**
The Australian BioGrid as a model infrastructure architecture for the Human Variome Project

12.15 – 13.00 **Open discussion**

Other abstracts to be discussed

Judith G. Hall
A Clinician's Perspective

John C. MacMillan
Addressing the weak link in annotating the human genome and its phenotypes, the busy clinician

Raoul CM Hennekam
Defining Morphology

13.00 – 14.30 **Lunch**

