



THE HUMAN VARIOME PROJECT

COLLECTION OF HUMAN GENE VARIATION

**HUMAN VARIOME PROJECT
IMPLEMENTATION AND INTEGRATION MEETING**

**UNESCO Headquarters
Paris, France**

10th– 14th May, 2010

Hosted by UNESCO

In association with:

Genomic Disorders Research Centre (GDRC) – HVP Coordinating Office

Co-Chairs:

**Richard G.H. Cotton
Michael S. Watson**

Satellite Meetings May 10th:

EMBL-EBI Training Workshop

The HVP Neurogenetics Consortium

International Society for Gastrointestinal Hereditary Tumours (InSiGHT)

NUGO: The Micronutrient Genomics Project (MGP)

Scientific Committee

Richard G.H. Cotton (Chair), Australia
Mike Watson, USA
Arleen D. Auerbach, USA
Segolène Ayme, France
Jean-Jacques Cassiman, Belgium
Mireille Claustres, France
Johan T. den Dunnen, Netherlands
Michael Katz, USA
Finlay Macrae , Australia
Yoichi Matsubara, Japan
Gabriela Möslein, Germany
Rajkumar Ramesar, South Africa
Sue Richards, USA
David Rimoin, USA (TBC)
Graham Taylor, UK
Thomas Weber, USA

Meeting Secretariat

Julia Hasler, UNESCO, France
Rania Horaitis, Australia
Heather Howard, Australia
Lauren Martin, Australia

Meeting Overview

A meeting co-hosted with the World Health Organization and supported by the American College of Medical Genetics was held in Melbourne, Australia in 2006 to initiate the Human Variome Project (*Pharmacogenomics*. Oct 2006;7 (7):969-972). This meeting brought together the most important people and organisations of the field and included representatives from the European Commission, UNESCO, OECD, NCBI and EBI among many other senior geneticists. In 2008 a planning meeting was held to formulate actions indicated from the 96 recommendations arising from the initiating meeting (*Nat Genet*. Apr 2007; 39(4):433-436). The planning meeting report including these actions were published in *Human Mutation* (*Hum Mutat*. Jan 20 2009; 30(4):496-510). The third Human Variome Project meeting will address the implementation and integration issues pertaining to this unique project, which was formed to address the urgent need in the areas of single gene disorders: translation of research to genetic healthcare, and in the near future, personalized medicine. This need is being able to efficiently access complete, accurate and up to date lists of human gene variation causing disease. This will allow those responsible to deliver timely and accurate genetic healthcare and inform research and therapy in the area including personalized medicine when everyone can cheaply get their genome sequenced. Systematic and complete collection of this data and its expert curation is not readily available and/or efficiently searchable at this time, despite numerous and some large scale, well meaning and expertly curated and designed databases being available.

This third meeting will formally discuss the next steps to be taken and planned for the project particularly surrounding “**Implementation and Integration**”. The aim is to determine the major ways in which the recommendations and actions from the first meetings and elsewhere can be best implemented in a global collaborative context to prepare the systems necessary to routinely and systematically gather the deluge of faults in genes now being discovered. There will be at least, 100 to 1000 faults in each of the 20,000 human genes affecting human health. This meeting will bring together key individuals who are currently working, and will work, on the necessary components and the development of the systems relevant to the task, including those working on pilot systems. Individuals include bioinformaticians, clinical geneticists, researchers, counsellors, database curators, diagnostic laboratory heads, common disease variation experts, genome sequencers, support groups and will address and include topics impacting on:- clinical data collection, genetic data collection, distributed databasing (country and gene specific), country specific collection, key pilot/current projects, central databasing, transfer to central databases, incentives for databasing, ethics of databasing and funding.

Abstracts are invited and presentations will be chosen from those who have or are planning systems containing actions which are relevant to the collection and display of “all mutations in all genes from all countries”. All abstracts will be forwarded to Chairs, discussed and taken into account even if authors are not speaking or attending in the interest of inclusiveness.

Space is limited to 200 so abstract content and key words of our applicant’s interest may be needed if the meeting is oversubscribed.

Objectives

1. To ensure progression of the objectives of the Human Variome Project to “collect variation causing disease (mutations) in all genes world wide”.
2. To gather experts and leaders in the strategies and systems required, together with those responsible for collecting data and its users.
3. To gather those who are already collecting mutations within their countries and others who wish to establish a system, especially those from developing countries.
4. To present projects currently underway or in the planning stage that contribute to the achievement of the objectives of the Human Variome Project objective.
5. To examine all areas of the implementation and integration of the overall and individual parts of the project.
6. To identify specific new projects and develop formal collaborative groups to undertake the work and to develop standards.
7. To gather those who are experienced in funding to consider optimal ways for the Human Variome Project to move forward and to identify potential funding sources for the projects.

The delegates will hear of programs now underway and planned in the listed areas, with a particular focus on the implementation of integrated systems into the healthcare strategies and systems globally and develop specific plans and collaborations. As we are meeting on behalf of thousands of potential collaborators, their potential to be involved should be remembered and ways to involve them developed.

PROGRAMME (Draft as of 12TH February 2010)

Day 1 – Monday May 10th

9.00 – 9.30	Registration of satellite meetings EBI Workshop - Room HVP Neurogenetics Consortium - Room InSiGHT - Room NUGO - Room
9.30 – 16.30	Satellite meetings
14.00 – 17.00	Registration for Human Variome Project Meeting - Room
16.30 – 17.00	Chairs discussion – Room
17.00 – 19.00	Human Variome Project Welcome Reception – UNESCO Headquarters

Day 2 – Tuesday May 11th

8.30 – 9.00	Registration
9.00 – 9.10	Welcoming addresses Walter Erdelen (UNESCO)/Ala Alwan (WHO)
9.10 – 9.30	Report on forums, session objectives & standards Richard Cotton/Mike Watson (Outline overall meeting plan, how to achieve meeting outcomes)
9.30 – 9.50	The Human Variome Project Roadmap Richard Cotton
9.50 – 10.50	Plenary Lecture Sir Prof. David Weatherall Chair: Julia Hasler
10.50 – 11.20	Coffee Break SESSION 1: Ethics Chairs: Sue Povey/Carol Isaacson Barash
11.20 – 11.40	Progress and potential directions – TBA
11.40 – 12.00	Abstract TBA
12.00 – 12.20	Abstract TBA
12.20 – 13.00	Discussion of future directions, projects and standards
13.00 – 14.00	Lunch SESSION 2: Overall Data Integration & Access – Central Systems Chairs: Donna Maglott/Fiona Cunningham
14.00 – 14.20	Progress and potential directions –TBA
14.20 – 14.35	Abstract TBA

14.35 – 14.50	Abstract TBA
14.50 – 15.05	Abstract TBA
15.05 – 15.20	Abstract TBA
15.20 – 16.00	Discussion of future directions, projects and standards
16.00 – 16.20	Coffee Break
	SESSION 3: Nomenclature and Standards Chairs: Johan den Dunnen/Richard Cotton
16.20 – 16.40	Progress and potential directions – TBA
16.40 – 17.30	Discussion
17.30	End of Day 2

Day 3 – Wednesday May 12th

9.00 – 10.30 **CONCURRENT SESSIONS 4, 5, 6 & 7**

CONCURRENT SESSION 4	CONCURRENT SESSION 5
Data Collection from Clinics Chairs: Gabriela Möslein/Garry Cutting 9.00 – 9.10 Progress and potential directions -TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract TBA	Data Collection from Laboratories Chairs: Graham Taylor/Sue Richards 9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract TBA
CONCURRENT SESSION 6	CONCURRENT SESSION 7
Publication, Credit & Incentives Chairs: Myles Axton/John Burn 9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract TBA	Assessment of Pathogenicity Chairs: Marc Greenblatt/Sean Tavgian 9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract TBA

10.30 – 11.00 **Coffee break**

11.00 – 12.30

CONTINUING CONCURRENT SESSIONS 4, 5, 6, & 7

CONCURRENT SESSION 4 CONTINUING	CONCURRENT SESSION 5 CONTINUING
Data Collection from Clinics Chairs: Gabriela Möslein/Garry Cutting 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract	Data Collection from Laboratories Chairs: Graham Taylor/Sue Richards 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract
CONCURRENT SESSION 6 CONTINUING	CONCURRENT SESSION 7 CONTINUING
Publication, Credit & Incentives Chairs: Myles Axton/John Burn 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract	Assessment of Pathogenicity Chairs: Marc Greenblatt/Sean Tavtigian 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract

12.30 – 13.30

Lunch

13.30 – 15.00

CONTINUING CONCURRENT SESSIONS 4, 5, 6 & 7

Discussion of future directions, projects and standards

15.00 – 15.30

Coffee Break

15.30 – 17.00

CONTINUING CONCURRENT SESSIONS 4, 5, 6 & 7

Consensus & Compilation of Reports

17.00 – 19.00

Free Time

19.00 –

Gala Dinner

Meet at Port de Javel Haut (Station Javel-Ligne C)

Day 4 – Thursday May 13th

9.00 – 10.30 **CONCURRENT SESSIONS 8, 9, 10 & 11**

CONCURRENT SESSION 8	CONCURRENT SESSION 9
<p>Overall Data Integration & Access – Peripheral Systems/Software</p> <p>Chairs: Christophe Beroud /Inge Bernstein</p> <p>9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract 10.10 – 10.30 Abstract</p>	<p>Translation to Healthcare and Personalised Medicine</p> <p>Chairs: Annika Lindblom/Ingrid Winship</p> <p>9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract</p>
CONCURRENT SESSION 10	CONCURRENT SESSION 11
<p>Country Specific Collection</p> <p>Chairs: Raj Ramesar/Jumana Al-Aama</p> <p>9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract TBA 10.10 – 10.30 Abstract</p>	<p>Data Transfer, Databasing and Curation</p> <p>Chairs: Arleen Auerbach/Johan den Dunnen</p> <p>9.00 – 9.10 Progress and potential directions - TBA 9.10 – 9.30 Abstract TBA 9.30 – 9.50 Abstract TBA 9.50 – 10.10 Abstract 10.10 – 10.30 Abstract</p>

10.30 – 11.00 Coffee Break

11.00 – 12.30 **CONTINUING CONCURRENT SESSIONS 8, 9, 10 & 11**

CONCURRENT SESSION 8 CONTINUING	CONCURRENT SESSION 9 CONTINUING
<p>Overall Data Integration & Access – Peripheral Systems/Software</p> <p>Chairs: Christophe Beroud /Inge Bernstein</p> <p>11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract TBA 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract</p>	<p>Translation to Healthcare and Personalised Medicine</p> <p>Chairs: Annika Lindblom/Ingrid Winship</p> <p>11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract TBA 11.30 – 11.45 Abstract TBA 11.45 – 12.00 Abstract TBA 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract</p>

CONCURRENT SESSION 10 CONTINUING	CONCURRENT SESSION 11 CONTINUING
Country Specific Collection Chairs: Raj Ramesar/Jumana Al-Aama 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract TBA 11.30 – 11.45 Abstract TBA 11.45 – 12.00 Abstract TBA 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract	Data Transfer, Databasing and Curation Chairs: Arleen Auerbach/Johan den Dunnen 11.00 – 11.15 Abstract TBA 11.15 – 11.30 Abstract 11.30 – 11.45 Abstract 11.45 – 12.00 Abstract 12.00 – 12.15 Abstract 12.15 – 12.30 Abstract

12.30 – 13.30 Lunch

13.30 – 15.00 **CONTINUING CONCURRENT SESSIONS 8, 9, 10 & 11**
Discussion of future directions, projects and standards

15.00 – 15.30 Coffee Break

15.30 – 17.00 **CONTINUING CONCURRENT SESSIONS 8, 9, 10 & 11**
Consensus & Compilation of Report

17.00 End of Day 4

Day 5 – Friday May 14th

9.00 – 10.30 SESSION 12: Funding Mechanisms and Sustainability

Chairs: Michael Katz/Daniela Seminara

9.00 – 9.15 Progress and potential directions – Michael Katz

9.15 – 9.30 Abstract TBA

9.30 – 9.45 Abstract TBA

9.40 – 10.20 Open Discussion

10.20 – 10.40 Coffee Break

10.40 – 13.00 SESSION 13: REPORT BACK of Plans and Project definition and standards

Chair: David Rimoin

10.40 – 11.00 Ethics
Sue Povey/ Carol Isaacson Barash

11.00 – 11.20 Overall Data Integration & Access – Central Systems
Donna Maglott/Fiona Cunningham

11.20 – 11.40 Nomenclature and Standards
Johan den Dunnen/Richard Cotton

- 11.40 – 12.00 Data Collection from Clinics
Gabriela Möslein/Garry Cutting
- 12.00 – 12.20 Data Collection from Laboratories
Graham Taylor/Sue Richards
- 12.20 – 12.40 Publication, Credit & Incentives
Myles Axton/John Burn
- 12.40 – 13.00 Assessment of Pathogenicity
Marc Greenblatt/Sean Tavtigian
- 13.00 – 14.00 Lunch**
- 14.00 – 16.00 SESSION 14: CONTINUING REPORT BACK of Plans and Projects
definition and standards**
- Chair: Richard Cotton**
- 14.00 – 14.20 Overall Data Integration & Access-Peripheral systems/software
Christophe Beroud/Inge Bernstein
- 14.20 – 14.40 Translation to Healthcare/Personalised Medicine
Annika Lindblom/Ingrid Winship
- 14.40 – 15.00 Country Specific Collection
Graham Taylor/Jumana Al-Aama
- 15.00 – 15.20 Data Transfer, Databasing and Curation
Arleen Auerbach/Johan den Dunnen
- 15.20 – 15.40 Funding Mechanisms and sustainability
Michael Katz/Daniela Seminara
- 15.40 – 16.00 Coffee Break**
- 16.00 – 17.00 SUMMARY & CLOSE**
Richard Cotton