



THE  
**HUMAN VARIOME**  
PROJECT

sharing data • reducing disease

A Non Governmental Organisation in Operational Relations with UNESCO

*Working Meeting Beijing*

**Co-organised by**

Human Variome Project Coordinating Office, Melbourne, Australia  
Human Variome Project China Office, Beijing, China  
National Association of Health Industry and Enterprise Management,  
Science & Technology Management Branch  
Beijing YuanTang Institute of Gene Science  
Beijing Science Technology Management College

**8th – 12th December 2011**  
**Radegast Lake View Hotel**  
**Beijing, China**

# Thursday 8th December

- 9.30 – 12.00 Human Variome Project International Limited  
**Meeting of Interim Board**
- 12.00 – 14.00 Human Variome Project International Limited  
**Members Luncheon**  
Attendees: Company Members, Interim Directors and Interim Scientific Advisory Committee Members
- 14.00 – 18.00 **Meeting of Interim Human Variome Project Scientific Advisory Committee**
- 14.30 – 16.30 Registration for HVP Beijing Meeting Open
- 18.30 Dinner – Radegast Lake View Hotel



# Friday 9th December

- 8.30 – 9.00 **Registration**

## **SESSION I Summit Opening & Setting the Guidelines - Frame of Reference for the Summit**

**Co - Chairs: David Abraham & Xitao Li**

- 9.00 – 9.05 **Musical Interlude**
- 9.05 – 9.10 **Opening Address**  
*David Abraham*  
Chairman, Human Variome Project International Limited
- 9.10 – 9.15 **Opening Address**  
Central Government Officer
- 9.15 – 9.20 **Opening Address**  
*Ramasamy Jayakumar*  
Programme Specialist for Science, Technology and Environment  
United Nations Educational, Scientific and Cultural Organisation
- 9.20 – 9.25 **Opening Address**  
*Richard Cotton*  
Scientific Director, Human Variome Project International Ltd.
- 9.25 – 9.30 **Opening Address**  
*Gang Wan*  
Minister, Ministry of Science and Technology

9.30 – 9.35	<b>Opening Address</b> <i>Zhu Chen</i> Minister, Ministry of Health
9.35 – 10.00	Ceremony to formally accept China as a Human Variome Project Partner
10.00 – 10.20	<b>Expected Outcomes and Outline of Topics</b> <i>David Rimoin</i> Chair, Scientific Advisory Committee
10.20 – 10.50	Group Photograph
10.50 – 11.20	Media Interviews
10.50 -11.20	<b>Coffee Break</b>

## SESSION 2 Plenary

**Chair: Richard Cotton**

11.20 – 11.30	<b>Video Address</b> <i>Peter C. Doherty</i> The Nobel Prize in Physiology or Medicine 1996
11.30 – 12.20	<b>Plenary Lecture</b> Human genetic variation and disease: the importance of global collaboration <i>Lynn Jorde</i> President, American Society of Human Genetics
12.20 – 13.30	<b>Lunch</b>

## SESSION 3 The Human Variome Project Chinese Node

**Chair: Stephen Lam**

13.30 -14.00	<b>What is an HVP Country Node?</b> <i>Richard Cotton</i> Scientific Director – Human Variome Project
14.00 – 14.15	<b>The Human Variome Project's Significance to Human Health; the role in clinics, treatment and prevention</b> <i>Denian Ba</i> Clinical Expert
14.15 – 14.30	<b>International Cooperation Development Strategy for the Future Genetics Applications</b> <i>Huanming Yang</i> Genetics Expert
14.30 – 14.45	<b>Future Development Strategy of Gene Industry Application</b> <i>Zhongjing Zhao</i> Representative of Global Gene Industrialisation Foundation and Global Industrialisation Association



14.45 – 14.50 **Human Variome Project China Node: Roadmap for Action**  
*Xitao Li*  
Board Director, Human Variome Project International  
Director, National Gene Centre of Sino Food and Drug Administration

14.50 – 15.15 **The Chinese Node 10 year Plan**  
*Ming Qi*  
Chief Coordinator of the Chinese Node

15.15 – 15.30 **Cooperation of Human Variome Project Chinese Node – Implementation plan**  
*Yufu Ren*  
Representative of the Executive Council of the Chinese Node

15.30 – 16.00 Discussion

16.00 – 16.30 **Coffee Break**



## **SESSION 4 Experiences from the Global Perspective**

**Chair: Aida Falcon De Vargas**

16.30 – 16.50 **The UK Experience**  
*Andrew Devereau*  
MutDB

16.50 – 17.10 **Collection from Large Laboratories – the example of MutaDATABASE, a universal database of DNA variants leading to human genetic disease**  
*Patrick Willems*  
MutaDATABASE

17.10 – 17.30 **The Australian Experience**  
*Timothy Smith*  
HVP Australian Node

17.30 – 18.00 Discussion

19.00 **Dinner**  
Radegast Lake View Hotel

# Saturday 10th December

## SESSION 5

### Key Issues in Developing a Country Node

**Chair: Yoichi Matsubara**

9.00 – 9.20

**Collection Within a Country: the French DMD and CFTR Databases**

*Mireille Claustres*

Medical Diagnostic Laboratory Expert

9.20 – 9.40

**Ethical Issues Arising in the Sharing of Human Genetic Data**

*Sue Povey*

Clinical Geneticist & Database Curator

9.40 – 10.00

**Ensuring the Integrity of the HVP Ethics Guidelines in the Face of Next Gen Technologies, Exponential Amounts of Data & Increasingly Global R & D and Marketing**

*Carol Isaacson Barash*

Co-Chair HVP Ethics Committee

10.00 – 10.20

**The Issues Faced in Low/Middle Income Countries**

*Aida Falcon de Vargas*

Clinical Genetics Expert

10.20 – 10.50

**Coffee Break**

## SESSION 6

### Key issues in developing a Country Node (cont.)

**Chair: Helen Robinson**

10.50 – 11.10

**Faced Issues in Middle and Low Income Countries of the Middle East**

*Mona El Ruby*

Clinical Genetics Expert

11.10 – 11.30

**Whole Genome Sequencing**

*Juan Wang*

Beijing Genomics Institute

11.30 – 11.40

**Whole Genome Sequencing from the Clinical Perspective**

*Rolf Sijmons*

Clinical Geneticist in the field of Oncogenetics

11.40 – 12.15

Discussion: Potential Areas for Collaboration Recommendations

12.15 – 13.30

**Lunch**





## Session 7

### Establishment of Gene/Disease Specific Databases (LSDBs)

**Chair: Finlay Macrae**

13.30 – 14.00

**Web-based Collection of Gene Variants Using LSDB-in-a-Box Software**

*Peter Taschner*

Database Software Expert

14.00 – 14.30

**Experience with a Large Mutations Database: Cystic Fibrosis**

*Garry Cutting*

Database Curator & Clinical Geneticist

14.30 – 15.00

**Current Initiation Stage and Future Requirements of the Chinese Node**

*Ming Qi*

Chief Coordinator Chinese Node

15.00 – 15.30

**Student Volunteer Program to assist the Establishment of Gene/Disease Specific Databases**

*Peikuan Cong*

PhD Candidate

15.30 -16.00

**Coffee Break**

## SESSION 8

### Establishment of Gene/Disease Specific Databases (LSDBs) (cont.)

**Chair: Arleen D. Auerbach**

16.00 – 16.30

**China, InSiGHT and the Human Variome Project**

*Finlay Macrae*

Honorary Secretary, International Society for Gastrointestinal Hereditary Tumours

16.30 – 17.30

Discussion

18.00

Departure for Gala Dinner: Grand Mansion Xicui Theatre Restaurant

Delegates to meet in hotel foyer for prompt departure by coach

# Sunday 1 1th December

## SESSION 9

### Key Issues in Establishing a Gene/Disease Specific Database (LSDB)

**Chair: Wayne Grody**

9.00 – 9.20

#### **Pathogenicity – Determination of Sequence Changes**

*Thomas Weber*

Gastroenterologic surgeon

9.20 – 9.40

#### **Curation of a Gene/Disease Specific Database (LSDB)**

*Arleen D. Auerbach*

Database Curator

9.40 – 10.00

#### **Phenotype Representation in Gene/Disease Specific Databases (LSDBs)**

*Peter Robinson*

Phenotype Ontology expert & clinician

10.00 – 10.30

Discussion

10.30 – 11.00

**Coffee Break**

## SESSION 10

### Key Issues in Establishing a Gene/Disease Specific Database (LSDB) (cont.)

**Chair: John Burn**

11.00 – 11.20

#### **Technical Standards and Data Models for System Interoperability**

*Anthony Brookes*

GEN2PHEN project

11.20 – 11.40

#### **Database Accreditation & the Gene/Disease Specific Database Advisory Council**

*Mauno Vihinen*

Database Curator and Standards Expert

11.40 – 12.10

Discussion

12.10 – 13.30

**Lunch**



## SESSION 11

### Global Collaborations

**Chair: Garry Cutting**

13.30 – 14.10

#### **Integration from the Global Perspective**

*John Burn*

UK Genetics lead, National Institute of Health Research and Chair of the British Society for Human Genetics

14.10 – 14.30

#### **The Role of IBSP/UNESCO and other international organisations**

*Angelo Azzi*

IBSP/UNESCO

14.30 – 14.50

#### **China's Role in Assisting Gene/Disease Specific Databases**

*Ming Qi*

Chief Coordinator Chinese Node

14.50 – 15.00

#### **Mechanisms for Assistance/Creation of Databases**

*David Rimoin*

Chair, Human Variome Project International Scientific Advisory Committee

15.00 – 15.30

Discussion

15.30 – 16.00

Recommendations

16.00 – 16.30

**Coffee Break**

## SESSION 12

### Meeting Summary

**Chair: Chris Arnold**

16.30 – 17.00

#### **Confirmation of Meeting Outcomes and Next Steps**

*David Rimoin and Ming Qi*

17.00 – 17.30

#### **Closing Addresses**

*Richard Cotton – HVP Scientific Director*

*Xitao Li – HVP China*

*Ramasamy Jayakumar - UNESCO*





THE  
**HUMAN VARIOME**  
PROJECT

**International Confederation of  
Countries Advisory Council  
Inaugural Meeting**

**12th December, 2011  
Radegast Beijing Lake View Hotel  
Beijing, China**



**Discussants:**

**HVP Country Node Representatives**

Australian Node	Heather Howard
Austrian Node	Martina Witsch-Baumgartner
Belgian Node	Thomy de Ravel de l'Argentiere
Chinese Node	Ming Qi
Cypriot Node	Andreas Hadjisavvas
Egyptian Node	Sherifa Ahmed Hamed
Hellenic Node	George Patrinos
Kuwaiti Node	Fahd Al Mulla
Malaysian Node	Zilafalil bin Alwi
Nepalese Node	Tilak Shreshta
Spanish Node	Maria Jesus Sobrido
Vietnamese Node	Chí Dũng Vŭ

**Human Genetics Society Representatives & others**

Najib Al Khaja	President Centre for Arab Genomic Studies
Kevin Carpenter	President Human Genetics Society of Australasia
Aida Falcon de Vargas	Co- President Latin American Network of Human Genetics Societies
Wayne Grody	President American College of Medical Genetics
Lynn Jorde	President American Society of Human Genetics
Jin-Sung Lee	President East Asian Union of Human Genetics Societies
Milan Macek	Past –President & Representative European Society of Human Genetics
Yoichi Matsubara,	President International Federation of Human Genetics Societies
Carmencita D. Padilla	Ex-President Asia Pacific Society of Human Genetics (currently Phillipine Genome Centre)
Feliciano Ramos	President Spanish Society of Human Genetics
Thomy de Ravel de l'Argentiere	Secretary Belgian Society of Human Genetics
August Rojas-Martinez	President Latin American Network of Human Genetics
Raj Ramesar	Liaison & Representative African Society of Human Genetics
Graham Taylor	President Human Genome Variation Society

**Interim Scientific Advisory Committee Members Attending**

Arleen D. Auerbach	John Burn
Mireille Claustres	Richard G.H. Cotton
Mona El-Ruby	Aída Falcón de Vargas
Yoichi Matsubara	Mauno Vihinen
David Rimoïn	

**Liason**

Julia Hasler	UNESCO Liason
Helen Robinson	WHO Liason

**Genetics Journals**

Charlotte Liu	Managing Director, Greater China, Macmillan Group
Mara Hvistendahl	Science Magazine, Asia Correspondent
David Swinbanks	Nature, Managing Director Asia Pacific



9.00 – 9.30

### **The Role of the Council**

*Richard Cotton*

Scientific Director, Human Variome Project International Ltd.

9.30 – 10.30

### **HVP Country Node Representatives: 5 minute introductions & updates**

*Chair: Richard Cotton*

Australian Node – Timothy Smith

Austrian Node - Martina Witsch-Baumgartner

Belgian Node – Thomy de Ravel de l'Argentiere

Chinese Node – Ming Qi

Cypriot Node – Andreas Hadjisavvas

Egyptian Node – Sherifa Ahmed Hamed

Hellenic Node – George Patrinos

Kuwaiti Node – Fahd Al Mulla

Malaysian Node – Zilafalil bin Alwi

Nepalese Node – Tilak Shreshta

Spanish Node – Maria Jesus Sobrido

Vietnamese Node - Chí Dũng Vũ

10.30 – 11.00

### **Coffee Break**

### **Open Discussion**

*Discussion Leader: George Patrinos*

11.00 – 12.30

### **Discussion Topics**

- Which countries are models
- Areas requiring standards and guidelines for countries
- Software and systems between countries and LSDBs and strategies for sharing
- Patient registries and other possible synergies
- Listing of needs

12.30 - 13.30

Lunch

### **Open Discussion**

*Discussion Leader: John Burn*

13.30 – 15.30

### **Discussion Topics**

- Strategies for dealing with different Provinces/Cities
- Relationship of China and LMIC Nodes
- Strategies for engaging with government and funders
- The role of the Coordinating Office staff and the frequency of future meetings

15.30 – 16.00

### **Coffee Break**

### **Discussion**

*Discussion Leader: Richard Cotton*

16.00 – 16.50

### **Recommendations, Plan of Action and next steps**

16.50 – 17.00

### **Closing Address**

*Richard Cotton & Ming Qi*

### **End of Meeting**