

The Human Variome Project in Europe

John Burn

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





THE
HUMAN VARIOME
PROJECT
sharing data · reducing disease



Michigan



Bangkok



Beijing



*August 2010 China National Highway 110
traffic jam, China, world's worst traffic jam
ever....*

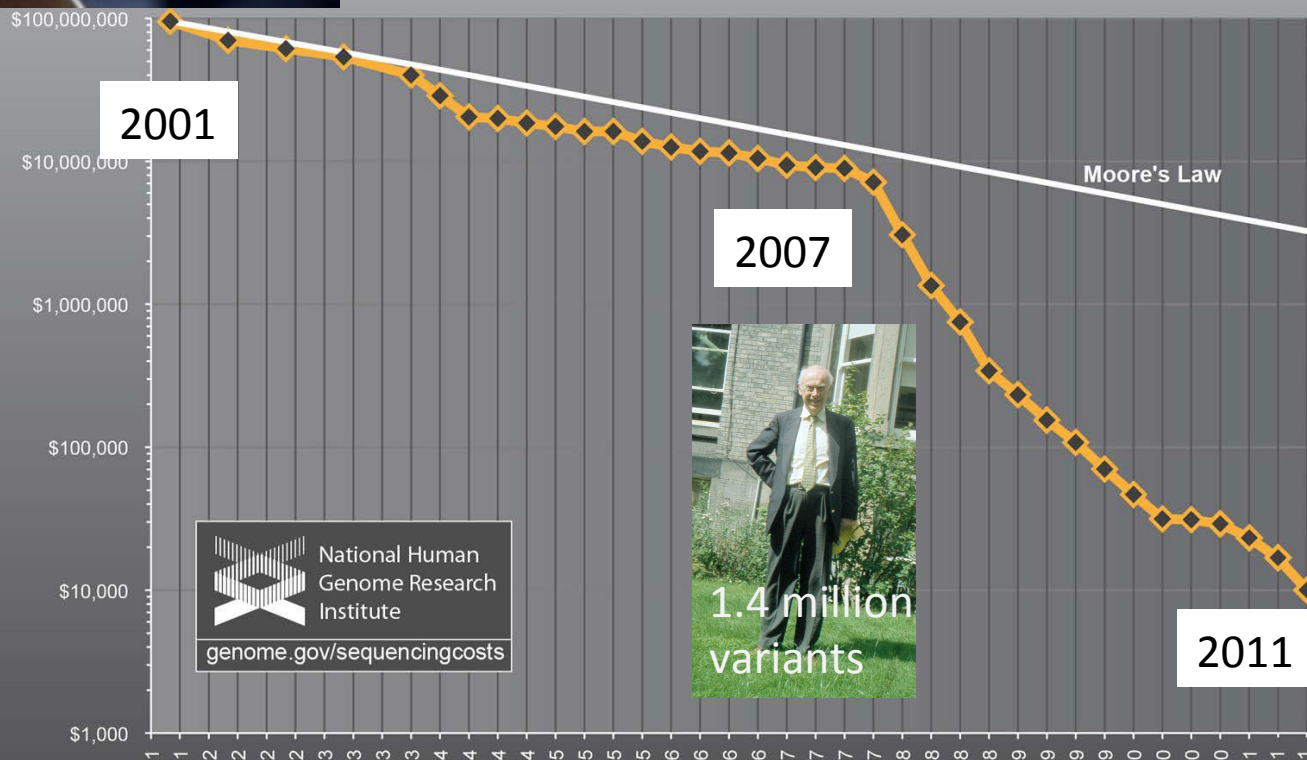
*more than 100 kilometres from August 14 - 26,
inc. at least 11 days of total gridlock.*





Sequencing costs have fallen more than ten thousand fold since 2000

Cost per Genome





Collaborative Curation of the Variome
sponsored by the UK Collaborative Group for Genetics in Healthcare
Hinxtion September 1st 2011
www.humanvariomeproject.org

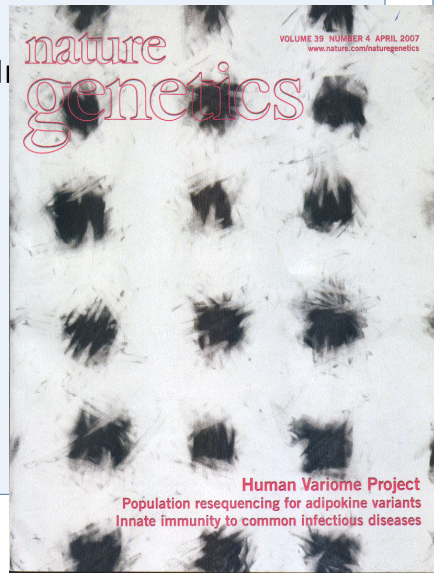


Human Variome project launched in 2006 under Richard Cotton, Melbourne

Members

- John Burn UK
- David Rimoin USA
- Jean Jacques Cassiman Belgium
- Eric Haan Australia
- David Abraham Australia
- Richard Cotton
- Dr. Xitao Li National Institute of Health, China

Nat Genet, 2007. **39**:433-6



HVP Scientific Advisory Committee elected May 2010, UNESCO, Paris

- Arleen Auerbach USA
- Marc Greenblatt USA
- Garry Cutting USA
- David Rimoin USA
- Mireille Claustres France
- Mona El Ruby Egypt
- Finlay Macrae Australia
- Yoichi Matsubara Japan
- Gert-Jan B. van Ommen The Netherlands
- Johan T. den Dunnen The Netherlands
- Mauno Vihinen Finland
- Christine v. Broeckhoven Belgium
- Aida Falcon Vargas Venezuela

Coopted:

- Richard Gibbs USA
- John Burn UK



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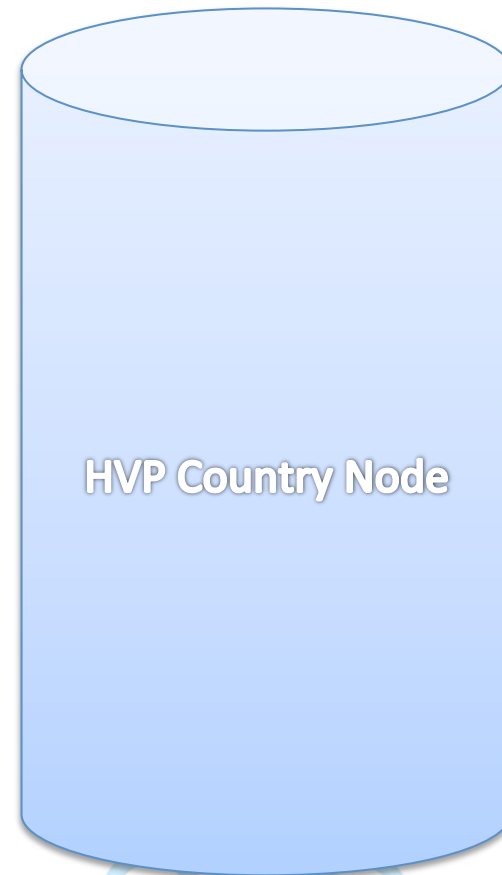
“For the Human Variome Project to have maximum benefit, we must all be able to share our knowledge freely and openly” Professor Xitau Li UNESCO June 2012

\$300m over 10 yrs. Inc. core support & \$1M for dev. Country nodes
(\$75,000 over 2 years for developed/developing country partnerships)

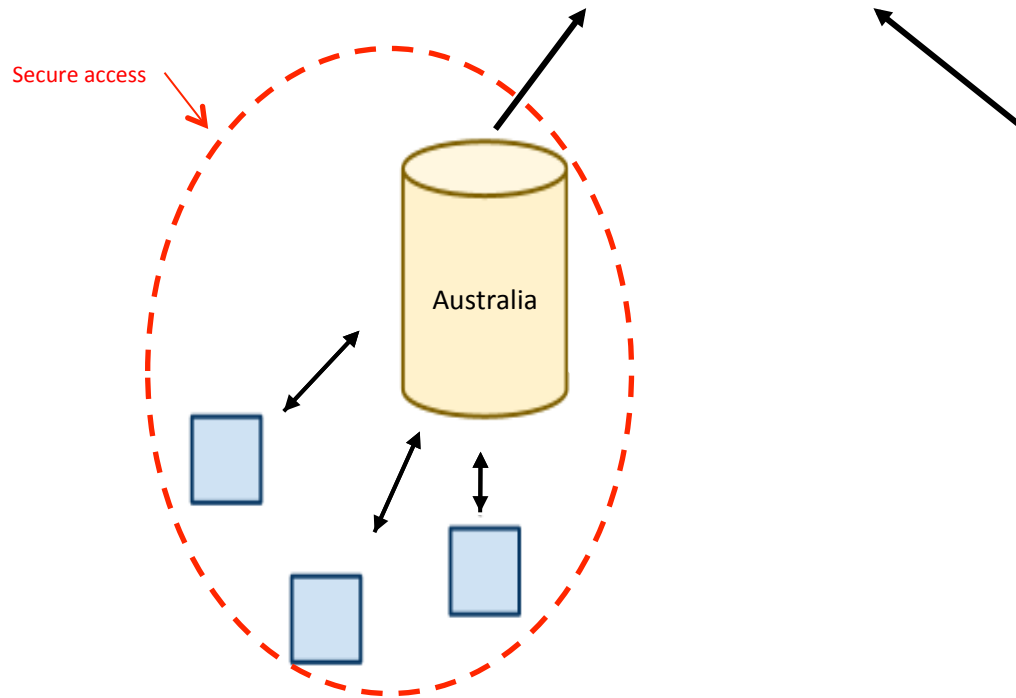


A Single Node

- Stores data from a single country
- Managed locally
- Local access
- Single legal and ethical framework
- Provides direct in-country benefits
- Relatively simple way for a country to contribute to the global effort



International Human Variome Project



How this idea works

- Each country forms a Node Consortium
 - Engage all stakeholders
 - Clinicians, Pathologists, Government, Patients
- Consortium decides what to collect and the methodology to do it
- Consortium constructs a repository to hold this data
- Consortium decides what information to provide to the international community

Everything is defined and controlled by the local consortium

HVP country nodes and support

- Country Nodes
- Support



1. Set normative functions
2. Behave ethically
3. Share knowledge
4. Build capacity

HVP needs input

Curators can be a little possessive



HVP is a recognised UNESCO NGO our aspiration

- An online free access reference database to all variants which have had functional significance validated
- A background “pool” of all variants detected
- Shared resource correlates phenotype with genotypes
- Rewards and regulation to ensure timely and complete collection of data
- A standard approach to assignment of probability



Proposal

- Seek formal endorsement of ESHG for HVP
- Use National Chairs committee to promote development of a european country node network
- Explore EU funding ? Under rare disease initiative
- Propose a “continental HVP board member” reporting to ESHG
- I volunteer to fill that position until 2014