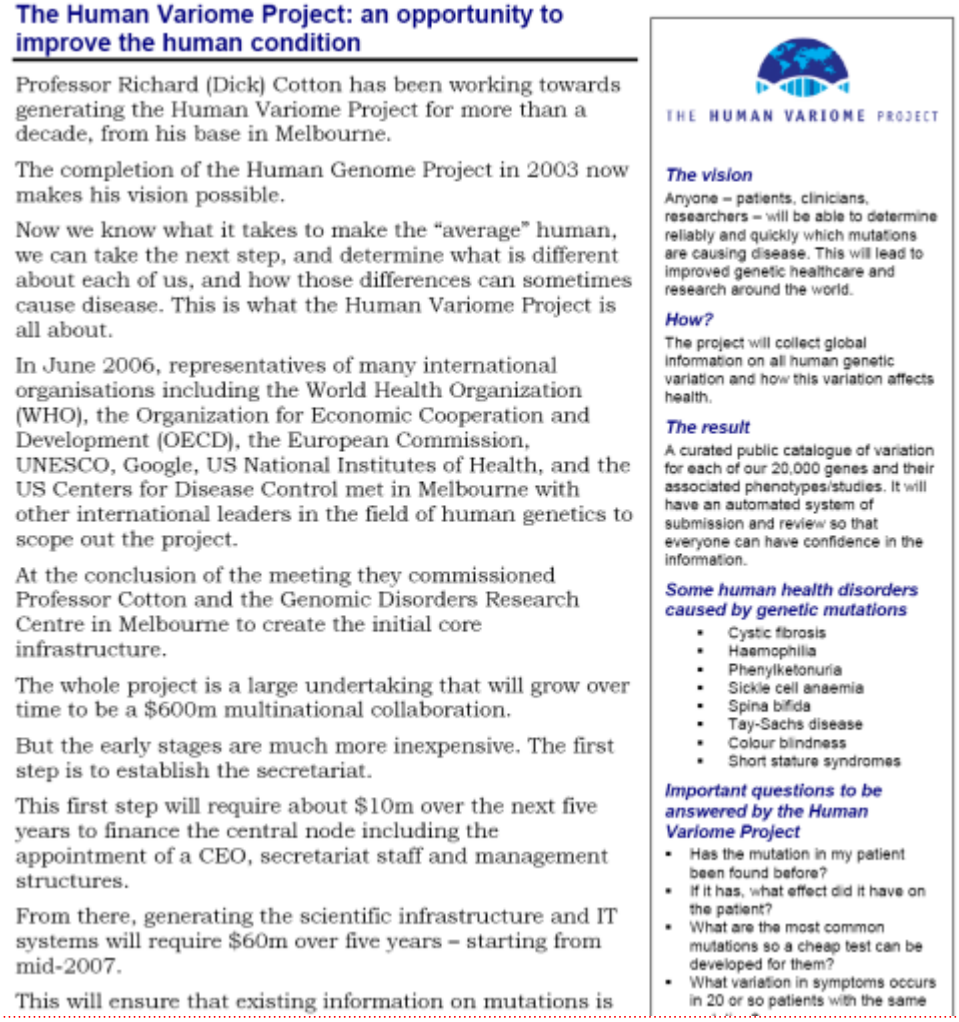
GENETICS, SOFTWARE

HUMAN GENOME VARIATION

5.02.2007 3 COMMENTS

Being a former curator for a genetic disease database, I received a PM that explained why the foundation of the Human Genome Variation Society did not include most of the HUGO Mutation Database Owners — most did not join as they found it difficult to pay for membership. This reflects the overall frustration in obtaining funds for databases projects that are between research and service. Now, a new initiative for the Human Variome Project (HVP) is started to create a focus pulling the whole vision together and to assist in fundraising. Meeting details are at www.humanvariomeproject.org. I strongly support this initiative. All genetic variation databases are sharing a high interest in the community but zero interest at funding bodies (more). I have a dream...

the flyer...



The Human Variome Project: an opportunity to improve the human condition

Professor Richard (Dick) Cotton has been working towards generating the Human Variome Project for more than a decade, from his base in Melbourne.

The completion of the Human Genome Project in 2003 now makes his vision possible.

Now we know what it takes to make the "average" human, we can take the next step, and determine what is different about each of us, and how those differences can sometimes cause disease. This is what the Human Variome Project is all about.

In June 2006, representatives of many international organisations including the World Health Organization (WHO), the Organization for Economic Cooperation and Development (OECD), the European Commission, UNESCO, Google, US National Institutes of Health, and the US Centers for Disease Control met in Melbourne with other international leaders in the field of human genetics to scope out the project.

At the conclusion of the meeting they commissioned Professor Cotton and the Genomic Disorders Research Centre in Melbourne to create the initial core infrastructure.

The whole project is a large undertaking that will grow over time to be a $600m multinational collaboration.

But the early stages are much more inexpensive. The first step is to establish the secretariat.

This first step will require about $10m over the next five years to finance the central node including the appointment of a CEO, secretariat staff and management structures.

From there, generating the scientific infrastructure and IT systems will require $60m over five years – starting from mid-2007.

This will ensure that existing information on mutations is

THE HUMAN VARIOME PROJECT

The vision

Anyone – patients, clinicians, researchers – will be able to determine reliably and quickly which mutations are causing disease. This will lead to improved genetic healthcare and research around the world.

How?

The project will collect global information on all human genetic variation and how this variation affects health.

The result

A curated public catalogue of variation for each of our 20,000 genes and their associated phenotypes/studies. It will have an automated system of submission and review so that everyone can have confidence in the information.

Some human health disorders caused by genetic mutations

- Cystic fibrosis
- Haemophilia
- Phenylketonuria
- Sickle cell anaemia
- Spina bifida
- Tay-Sachs disease
- Colour blindness
- Short stature syndromes

Important questions to be answered by the Human Variome Project

- Has the mutation in my patient been found before?
- If it has, what effect did it have on the patient?
- What are the most common mutations so a cheap test can be developed for them?
- What variation in symptoms occurs in 20 or so patients with the same

3 THOUGHTS ON “HUMAN GENOME VARIATION”

Pingback: Gene Expression

Pingback: Just Science

Richard Cotton

15.06.2007 AT 06:45

The situation where some HUGO Mutation Database owners (curators) are excluded because they can’t afford the fees is regrettable and has been a chronic problem. We have to address this at the upcoming meeting in San Diego on October 23rd 2007 (www.hgvs.org/meetings/sandiego.html). Any ideas on how to address this problem are welcome.

COMMENTS ARE CLOSED.