

30 March, 2012

Secretariat
Strategic Review of Health & Medical Research
PO Box 4226
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Submission to Strategic Review of Health & Medical Research in Australia

Background to Submission

The Human Variome Project is an international consortium of scientists and health-care professionals who are working towards a significant reduction in the burden of genetic disease on the world's populations. The aim of the Human Variome Project is to ensure that all information on genetic variation can be collected, curated, interpreted and shared freely and openly. This will lead to speedier, better and cheaper diagnosis and treatment of genetic disorders, and better insight into the causes, severity and effect of common disease. The Human Variome Project achieves its aims by establishing and maintaining the necessary standards, systems and infrastructure, by providing education and training to scientists, clinicians, genetic counsellors, other healthcare professionals and the general public and by helping developing nations build their capacity in medical genetics and genomics. The Human Variome Project acts as an umbrella organisation and works to encourage communication and collaboration around its central vision.

The Human Variome Project doesn't establish databases itself; however it actively encourages the establishment of both Country and Gene/Disease Specific Databases for the sharing of data globally. As a part of this system of sharing Australia is establishing an HVP Country Node this Node will share de-identified molecular and clinical data with the international Gene/Disease databases, this data will assist in the interpretation of the pathogenicity of many disease states.

The importance of the Human Variome Project was recognised in 2011 by the United Nations Educational, Scientific and Cultural Organisation in the Project's admittance to Consultative Partner status.

Human Variome Project International Limited, a not-for-profit Australian public company, has been set up to act as the International Coordinating Office for the Human Variome Project. The International Coordinating Office is presently located in Melbourne.

Its role includes:

- providing strategic direction and oversight for the Project, including governance and business affairs
- supporting the operation of Project-wide committees and representative bodies
- monitoring and reporting on the progress of Project initiatives and activities

As such, the Human Variome Project International Limited has an interest in the strategic direction of health and medical research in Australia and we thank the panel for the opportunity to present this submission.

1) Why is it in Australia's interest to have a viable internationally competitive health and medical research sector?

- To minimise suffering and death to a level which equates to the top 5% of countries in the world.
- To have a seat at the table of important decision making bodies such as WHO and international societies etc.
- To enable rapid transfer of life saving and suffering-reducing procedures developed throughout the world to our patients and research projects.
- To enable rapid disaster response which are relevant to minimise mortality and morbidity.
- To generate patentable processes and medicines to allow corporate exploitation and creation of jobs

2) How might health and medical research be best managed and funded in Australia?

Managed:

- To avoid projects 'falling' between the stools' of research and healthcare, a set percentage of funds should be assigned to joint healthcare/ research activities.
- Major projects should be judged by relevant people at the coal face rather than high profile senior scientists who have migrated to administrative duties only.
- International reviews so personal preferences and favouritism may be eliminated.

Funded:

- Research should be treated the same way as Healthcare, each should have a fixed percentage of GDP allocated to them.
- Ideally 'Successful' companies arising out of government supported research should contribute to a 'research fund' to pay back their debt to government funded research.
- Funding of 'grant preparation' to selected bodies/ institutes specifically and especially to focus on international funds to bring money in. Top researchers do not have the time to write the multiple applications demanded at present
- Risk project funding should be awarded to proven innovators in Health and Medical research.

3) What are the health and medical research strategic directions and priorities and how might we meet them?

- **Data Collection and Sharing:** Healthcare especially is shackled by the fact that clinicians and lab heads are chronically underfunded due to budgeting constraints (E.g. clinicians spent 15 minutes per patient all day and a lab head still has to account for each 15 minutes of his day), therefore they do not have the time to think, let alone

do something to help their efficiency! In my field, as in many others, this refers to access to complete databases rather than perhaps having to have long searches on the web of information that is not even up to date or complete. Thus the direction and priority would be to support an experienced but 'outside' person who might be called a 'Priority Coordinator or Development Officer', even part-time to make sure that the data is collected and shared. If Data sharing was a mandated requirement then time and resources would be allocated to the task or automation developed.

- Lack of access to complete hospital records, world experience in the field and access to relevant experts, inhibits the best medical healthcare in many areas. This is especially so in genetic healthcare and I am sure, is not just confined to the field of genetics. Therefore, funding of the construction of databases, would save hundreds of hours for professionals and provide the clinician with the most up to date information when treating their patients. This would provide very cost effective and suffering-saving investment. Naturally confidentiality and privacy of the individual must be protected.
- Databases just mentioned also act as a substrate for clinical trials at least by providing cohorts for trials as well as determining the load of disease which might then inform investment by government.

4) How can we optimise translation of health and medical research into better health and wellbeing?

- Translation can be optimised by bringing the experience of the world via the sharing of genetic variation (or key data in any disease) to bear on an individual patient or disease at the click of a computer key.
- PhD/ Medicos need encouragement. Currently many talented medicos cannot afford to do research. High profile fellowships could be offered to 35 to 40 individuals to ensure that they bridge the research/ healthcare boundary to enhance translation.

Yours Sincerely,



Richard G.H. Cotton AM PhD DSc FRCPA(Hon.)

Scientific Director