

Senate Hearing Gene patents

Summary of evidence given by Jennifer
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Foundation

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Australian Marfan Foundation

- I am a Director and Secretary of the AMF
- The AMF is a recently established not-for-profit organisation
- The aims of the Foundation are
 - To provide support for subjects with Marfan syndrome
 - To stimulate research into MFS
 - Increase community awareness of MFS and its complications by the development of educational tools for patients, schools and medical practitioners

Marfan Syndrome

- An inherited disorder affecting 1 in 3-5000 in the community (Oxford Desk Reference Clinical Genetics 2005)
- Caused by a mutation in the DNA sequence of the fibrillin gene, a gene involved in ensuring the integrity or strength of connective tissue
- Main risk of MFS is development of aortic aneurysm with potential for rupture or dissection
- Other physical problems include eye, skeletal, skin and respiratory abnormalities

My background

- I am a registered nurse
- I work coordinated a diagnostic and follow-up MFS clinic at Prince Charles Hospital in Brisbane for the last 15 years. PCH is the main hospital in Qld that deals with MFS and cardiac surgery
- I have been a member of the Qld Marfan Association for 18 years and act as Almoner
- I have a significant background in dealing with MFS subjects and their multiple clinical problems
- Together with my husband and other colleagues around Australia we established the AMF in 2009

Diagnosis of MFS

- Up to the present the diagnosis of MFS has been based on satisfying the clinical criteria for MFS (determined at an international conference in gent, Belgium in 1996). However it has been found that not all subjects with MFS fully satisfy the criteria
- DNA testing for mutation sites in the fibrillin gene is now the gold standard. Unfortunately since each family has a different mutation mutation testing is not simple. Multiple sites in the gene need to be assessed. DNA testing is thus complex and not readily available.
- There is no patented test for MFS. DNA testing is carried out by both not for profit and commercial organisations. There is minimal government supported MFS testing
- It is possible that a surrogate DNA test (using DNA haplotypes) will be developed but there is no such test available at present

Issues for Consideration

The 5 points we have identified as relevant to this

Hearing are:

1. DNA gene sequences are not inventions and should be available to anyone
2. Diagnostic or treatment innovation is **NOT** more likely to occur if a gene sequence is patented
3. Proposed new laws regarding patents for genes should involve ethicists, scientists and the public as well as legal practitioners
4. The law regarding patentability of genes needs to be reviewed (ALRC report disagrees with this)
 - Gene testing should be regulated, accessible and benefits and limitations publicised

Issues for Consideration 1.

- A gene sequence is a naturally occurring set of chemical data within the human genome. Therefore it is not an invention. It should not be able to be patented. The data should be freely available. Likewise the discovery of a mutation within the gene sequence is also a natural information and not an inventio. An invention is the way in which natural information is used in a unique fashion or innovative way.

Issues for Consideration 2.

- The development of diagnostic tests, novel treatments or further understanding of a biological phenomenon does not depend on whether the gene sequence is patented.
- Patenting limits access to new information. Greater access to information would seem to be more likely to stimulate innovation

Issues for Consideration 3.

- Any review of the law or recommendations by this Committee relating to gene patenting should involve a wide cross section of the community including ethicists, scientists and the general public. Any such review should not be confined to legal practitioners and patent attorneys

Issues for Consideration 4.

- The AMF believes that the current law as outlined in the Australian Law Reform Commission report *Genes and Ingenuity (2004)* should be reviewed and changed.
- The Australian Law Reform Commission report *Genes and Ingenuity (2004)* recommended the *status quo* to apply *ie* no change in current laws and the ability to patent gene sequences to continue. **The AMF disagrees with this recommendation.**

Issues for Consideration 5.

- Genetic testing should be regulated by Government and controlled by registered professional providers
- Genetic testing should be accessible to the Australian public via medical practitioners
- The community should be made aware of the benefits, limitations and consequences of genetic testing and subjects undergoing such testing should likewise be given similar information

Benefits and Costs of DNA Gene Testing for Marfan Syndrome

Benefits

- Ability to make a correct and watertight diagnosis
- Ability to determine whether other family members have the genetic abnormality
- Ability to commence early treatments
- Removal of need for expensive lifelong monitoring for aortic aneurysm in family members where DNA testing is negative

Benefits and Costs of DNA Gene Testing for Marfan Syndrome

Costs

- Impact on ability to obtain life insurance
- Impact on social life
- Family planning issues