THE ROYAL COLLEGE OF
PATHOLOGISTS OF AUSTRALASIA

Position Statement

Subject: **Patenting of Human Genes**
Approval Date: 25 July 2001
Review Date: July 2006
Number: 3/2001

The Royal College of Pathologists of Australasia considers that genes and their mutations are naturally occurring substances that should not be patented. As a consequence any intellectual property rights should not reside in the actual gene sequence itself but only in any process by which its nature was ascertained. Patenting of an actual gene sequence inhibits access to the basic knowledge of genes. As a consequence, such gene patents limit development, improvement and eventual use of tests based on that knowledge.

General consequences of such monopolisation are likely to include:

- reduced patient access to testing,
- increased costs of testing,
- increased division between those who can afford tests and those who cannot,
- dictation of standards of care for testing by patent holders,
- reduction in the vital element of peer review in test performance,
- creation of unacceptable conflicts of interest,
- restriction of further research activity.

In addition, the specific consequences of tests used in medically complex tests, including testing for cancer susceptibility genes such as BRCA1 and 2, are likely to include:

- Disruption or loss of the critical clinician-laboratory interface, which is necessary for skilled interpretation of these complex tests.

- Difficulties in establishing an open and effective regime of external quality assurance.

- Loss of opportunities for training of laboratory scientists, geneticists, pathologists and physicians.

- Entrepreneurs adopting testing strategies that are determined by profit rather than a medical necessity. This is likely to result in a limited repertoire of high volume tests being completed rather than offering a wide range of testing strategies to look for different types of mutations or rare mutations. For example, in persons with a strong family history of breast cancer, but negative initial mutation screening for BRCA1 and 2, it is sometimes necessary to undertake additional testing (such as genetic linkage, promoter mutations, methylation status, large deletions) in a specialised reference laboratory. Use of any of these methods might infringe the patent and the reference laboratory would be compelled to cease testing. Although the licensed laboratory cannot offer the full range of these tests, it will not be possible to fully investigate the more difficult cases. This would impair the standard of care.

Durham Hall 207 Albion Street Surry Hills NSW 2010 Australia
Tel: 61 2 8356 5858 Fax: 61 2 8356 5828
Email: rcpa@rcpa.edu.au

- Identification of mutations and their clinical significance usually requires testing of a large patient database and utilising linkage disequilibrium studies. This is normally done either by, or in conjunction with, public institutions such as major teaching hospitals or university medical departments. Thus, much of the work can already be considered to be in the public domain.

- There may have already been a universal uptake of testing for a disease or gene mutation before the patent has been issued, or before the owner of the patent decides to enforce. This is highly likely for disease genes with relatively few single nucleotide polymorphisms (SNPs) and where the prevalence of the disease is quite high and the disease has a high profile such as in Haemochromatosis (HFE gene). In Australia, HFE testing is now so universally accepted that it has been incorporated into the Medicare Benefits Schedule (MBS). Tests are normally placed on the MBS when it is considered that they should be universally available within Australia.

It may be argued however, that patents are essential if genetic venture companies are to continue to put money into research. However, where the downstream development costs of diagnostic applications are minimal (as in many SNP tests), then it may also be argued that any royalty or license payments should also be minimal. This is especially so where large numbers of tests may be performed in the interests of good medicine.

Given the large number of genes with the potential to impact across almost every known human disease, this is a development with far-reaching implications. For these reasons it is vital that the issues raised receive urgent but thorough consideration for development of appropriate public policy before restrictive genetic testing through patent enforcement becomes widespread.