



HUMAN GENETICS SOCIETY OF AUSTRALASIA

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October 11th, 2007

Dear Greg,

Re: Targeted consultation on draft genetics privacy guidelines

Thank you for your letter of September 12th. These draft guidelines were reviewed by three senior members of the Human Genetics Society of Australasia (Drs David Amor, Clara Gaff and myself), whose range of expertise covered clinical genetics, genetic counseling and molecular genetics diagnosis and research. We thought that this was a very impressive draft document, written in an extremely clear manner and which benefited greatly from the use of scenarios to engage the reader and illustrate realistic examples that may confront physicians. We regarded definitions such as "serious" and "genetic relative" as appropriate.

One area where we suggest the document could be improved is to be more specific about the issue of reproductive risk. We recognise that this is a sensitive area, but it is one of the most likely to cause confusion and uncertainty in the future. We suggest that either a specific scenario be added to address this issue directly, or one of the existing ones be modified. This could be helpful particularly to private practitioners who may be influenced by their own views of termination of pregnancy and may not be used to considering issues of reproductive risk.

At present there are two scenarios where we thought the issue of reproductive risk could be addressed more directly. In Scenario 3 (Fragile X syndrome), the implications for relatives of FXTAS and premature menopause are specified, but no mention is made of the fact that females with a premutation are at risk of having a child with Fragile X syndrome and might alter their reproductive choices if they discover they are carriers. Under 3.2.3 there is indirect reference to reproductive risk in so far as "serious threat to health" is stated to include a threat to psychological health "that might be associated with making a reproductive health choice". The implication of this statement seems to be that it is appropriate to inform at risk relatives of a reproductive risk, on the basis that having an affected child represents a risk to psychological health (similar to the situation with termination of pregnancy). If this interpretation is correct, why not include the issue specifically in one of the Scenarios?

Scenario 8 touches more closely on reproductive risk although the emphasis is on when disclosure may not be possible. The statement "Knowledge of her potential to carry the disorder may also have led the daughter to consider testing before a pregnancy" could perhaps be

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expanded by finishing the sentence with "in case this was relevant to her decision on having children or to considering reproductive options such as donor gamete IVF, pre-implantation genetic diagnosis or prenatal diagnosis."

The issue of reproductive risk in scenarios 3 and 8 is diluted by the fact that in both examples there is a risk of adult-onset disease in the women that distracts from the issue of reproductive risk. We suggest it may be clearer if, for example, scenario 8 was changed to an X-linked recessive condition without manifestations in a female carrier. This would allow the issue of reproductive risk to be distinguished from direct health risks to the individual.

On p.6 under Key Points for Best Practice on Discussing Disclosure, the wording of the second point seems weaker than when this issue is discussed later in the text. We suggest changing "If necessary, refer patients to a health practitioner with expertise ..." to "Consider referring patients to a health practitioner with expertise ...". Otherwise the layout, sentence structures and proof-reading of the document are excellent with just one typographical error being noted, on p.25 in the 4th line of text for scenario 3, "Fragile X syndrome gene was identified" should read "Fragile X syndrome was identified"

Thank you again for the opportunity to comment on this excellent draft document. We look forward to circulating the revised draft document more widely to our membership when it is available for public consultation.

Yours sincerely,



Assoc. Prof. David Thorburn
President, Human Genetics Society of Australasia

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