

20 September, 2007

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(via email humangenetics@nhmrc.gov.au)

Dear Dr Ash,

**Disclosure of Genetic Information to a patient's genetic relatives
Targeted consultation**

Thank you for the opportunity to comment on this excellent document. The case scenarios are very effective in exemplifying and unbundling the issues that arise when dealing with the need to contact genetic relatives when a serious inherited disorder has been identified.

I note that this document is relevant to the private sector and as such will be relevant to our organisation, the Victorian Clinical Genetic Services. However these issues arise from time to time in all clinical genetic services and attention will need to be given at either Federal or State level on how this can be legally applied in those settings.

My comments on the specifics of the documents follow:

1. The term "**health practitioner**" needs to be defined. Since there is a need for special professional judgment in the consideration of disclosure and in providing such disclosure, I suggest that this should be confined to medical practitioners, genetic counsellors and social workers who would be expected to have those skills.
2. The summary (p5) clearly states that "**the disclosure must not reveal the identity of the patient**". This is clarified on p30 "that a heritable tendency has been found in a family member" and invites them to attend for testing. There is also excellent information on p34. Such clarification should be made on p5 since how such disclosure can be done would not be obvious. However I suggest that the wording be changed to "a heritable factor that could potentially affect your health has been identified in your family. If you would like to learn more about it and consider to be tested to see whether you have inherited this tendency please contact us".
3. **Predictive genetic testing** (p15), comment also needs to be made about differences in penetrance and expression of genes.
4. **Right not to know** - is problematic. How can a decision be informed without information?

5. Getting the **patient to assist in disclosure to relatives** (p22) details very well what information needs to be passed on and that it needs to be provided in writing. The point could be made on p7 in relation to the flow chart that when the patient is asked to contact relatives, it is a process that needs to be assisted with appropriate written information and it is a process that needs to be supported so that the relatives don't "shoot the messenger" or if they do the health practitioner is there to provide support.
6. **Testing opportunistically** (p27) for glycosuria can be part of the care of any patient but it should be a matter of caution to point out that gene testing can only be done with specific informed consent.
7. **"Psychological threat associated with making a reproductive health choice"** as serious potential harm is certainly worth considering. Most families choose to make deliberate decisions about the children they have. Whether or not they choose testing, they want to be in the knowing situation to make that choice. 70% of pregnant women in Victoria choose MSS and 85% choose prenatal diagnosis when they know of increased risk. The choices may be different for adult onset conditions but people still want the information.
8. **Scenario 6** (p30) has an unusual ending that may be worth reconsidering. It would be most unusual for a person who has had predictive counselling to be so shocked with a positive result that they would refuse any further contact with the counsellors. They may however decide adamantly not to share their information and refuse consent for disclosure.
9. **Telephone answering machines** (p32). This note of caution is well founded. It may be worth clarifying that the caller has no knowledge of who might access the voicemail eg. the patient's personal assistant!
10. **Scenario 8** (p33) neonatal myotonic dystrophy is often lethal. It would be worth stating this to underline the potential seriousness of non timely disclosure.

With best wishes,



Professor Agnes Bankier
Director
Genetic Health Services and VCGS Pathology