

Predictive genetic testing in children

Fiona H Richards,* Roslyn J Tassicker,†
Jennifer G R Kromberg,‡ Barbara M
Singaram§

*Senior Social Worker, Predictive Testing Program, Department of Clinical Genetics, The Children's Hospital at Westmead, Locked Bag 4001, Westmead, NSW 2145; †Predictive Testing Counsellor, Huntington Disease Predictive Testing Services, Genetic Health Services Victoria, Melbourne; ‡Principal Genetic Counsellor, Queensland Clinical Genetics Service, Royal Children's Hospital, Brisbane; §Coordinator/Senior Social Worker, South Australian Huntington's Disease Service, Flinders Medical Centre, Bedford Park, Adelaide.

TO THE EDITOR: We are writing to express concern about the article by Savulescu in the 1 October 2001 issue of the Journal.¹ As social workers with extensive experience in the area of predictive testing for Huntington disease (HD), we wish to make the following points:

■ The author fails to mention that specialist multidisciplinary teams with training and experience in all aspects of predictive testing (and associated ethical issues) are based in genetics services in every Australian State. Experienced counsellors in these teams can explore the subtle psychological processes that may underlie a parent's request for predictive testing of their child. For example, in the hypothetical clinical encounter depicted by the author, "Mrs Smith" may feel guilty that her children are at risk, and might be motivated by a desire to know that her children have *not* inherited the HD mutation from her.

■ Through our work with adults who undertake predictive testing, we are acutely aware of the complex and subtle adverse psychological effects on the individual and on family relationships that often follow such testing. These effects are likely to be even more evident in families in which children's risk for an untreatable, adult-onset disease is known.

■ The child's right to autonomy in making his or her own decision about testing for such conditions, *when he or she reaches the age of 18*, is supported in a number of guidelines, including the international guidelines for predictive testing in Huntington disease.² These guidelines were formulated by experienced professionals and representatives of HD families worldwide — their wisdom in this matter should not be ignored.

■ In our experience, basing a decision about predictive testing for a minor on an assessment of the child's competence (not the same as intelligence) does not take into account other important factors, such as

the level of insight and maturity to make a life decision that has such potentially far-reaching implications.

■ The author misrepresents research on suicide and predictive testing (in the survey by Almqvist et al,³ the five people who suicided had not only tested positive but were also symptomatic), and refers to an article that was based on small numbers of people who underwent linkage testing.⁴ There is much more recent and reliable published research that highlights the complexities of reactions to predictive test results.⁵

1. Savulescu J. Predictive genetic testing in children. *Med J Aust* 2001; 175: 379-381.
2. International Huntington Association and World Federation of Neurology Research Group on Huntington's Disease. Guidelines for the molecular genetics predictive test in Huntington's disease. *J Med Genet* 1994; 31: 555-559.
3. Almqvist E, Bloch M, Brinkman R, et al. A worldwide assessment of the frequency of suicide, suicide attempts, or psychiatric hospitalization after predictive testing for Huntington disease. *Am J Hum Genet* 1999; 64: 1293-1304.
4. Wiggins S, Whyte P, Huggins M, et al. The psychological consequences of predictive testing for Huntington's disease. *N Engl J Med* 1992; 327: 1401-1405.
5. DudokdeWit A, Tibben A, Duivenvoorden H, et al. Predicting adaptation to presymptomatic DNA testing for late onset disorders: who will experience distress? *J Med Genet* 1998; 35: 745-754. □

Ethics and evidence-based medicine

Gil M Anaf

President, National Association of Practising Psychiatrists, PO Box 12, Arncliffe, NSW 2205
ganaf@ozemail.com.au

TO THE EDITOR: Comments made by Parker et al¹ in response to Leeder and Rychetnik's article on evidence-based medicine (EBM)² do not reflect the reality of the dilemmas clinicians face in practice — arguably, because of political misuse of the concept of EBM, which Leeder and Rychetnik warned against.

Parker et al take issue with the "worry that EBM might be misused in public policy...where evidence is difficult to obtain", and argue that this is not the case. However, the previous Health Minister, Dr Wooldridge, was a great admirer of the Cochrane Collaboration, and, based on a perceived lack of evidence, he cut Medicare rebates in 1996 (by 50%) for patients needing long-term intensive psychiatric outpatient treatment. Although, after much protest, this decision was amended somewhat, Item 319 of the Medical Benefits Schedule remains today as a stark reminder of how some patients cannot access fully the treatment they desperately need.

There is abundant evidence (international and local) as to the efficacy of this

form of intensive treatment.³ There is also abundant and clear evidence that all who seek this treatment are traumatised by previous failed shorter treatments, often have comorbid disorders, and have established DSM-IV diagnoses of long standing.⁴ All this evidence was made available to the Minister — but Item 319 remains, with its exclusionary and discriminatory criteria to ration access, in my opinion due in large part to political misuse of the concept of EBM. Contrary to the assertion of Parker et al, there is a great deal to worry about.

In addition, Parker and colleagues make the claim that mental health is attracting government attention and funding. Again, in reality, a great deal of money is being spent on promoting education and awareness — and certain kinds of treatment. There is no evidence that short-term treatments (which are heavily promoted) actually help the group excluded by Item 319 regulations. Yet public policy is being pushed along the lines of "one size fits all". It does not.

All this is evidence of misuse of the idea of EBM reflected in public policy, and patients are suffering as a result. To make matters worse, cuts in one area are mindlessly used to push agendas that in clinical reality will be unworkable in other areas — all of which devalues professional expertise and judgement.

1. Parker MH, Del Mar CB, Glasziou PP. Ethics and evidence-based medicine [letter]. *Med J Aust* 2001; 176: 138.
2. Leeder SR, Rychetnik L. Ethics and evidence-based medicine. *Med J Aust* 2001; 175: 161-164.
3. Doidge N. In: Cameron PM, Ennis J, Deadman JC, editors. Standards and guidelines for the psychotherapies. Toronto: University of Toronto Press, 1998.
4. Doidge N, Simon B, Gillies LA, Ruskin R. Characteristics of psychoanalytic patients under a nationalised health plan: DSM-III-R diagnoses, previous treatment and childhood trauma. *Am J Psych* 1994; 151: 586-590. □

Malcolm H Parker,* Chris B Del Mar,†
Paul P Glasziou‡

Senior Lecturer in Ethics and Professional Development, †Professor of Evidence-Based Practice, School of Medicine, ‡Director, and Professor of General Practice, Centre for General Practice, University of Queensland, Herston, QLD
m.parker@mailbox.uq.edu.au

IN REPLY: Anaf seems to have missed our point that the choice by Leeder and Rychetnik¹ of mental health as an area relatively devoid of good quality evidence was a poor one — quality research has revealed mental health as an area of considerable need, and mental illness as a significant component of the global burden of disease. Despite the fact that evidence is often more difficult to obtain within the mental health area, much evidence exists — for example, the Cochrane Collaboration