

Down syndrome screening in Australia: More luck than choice

As recently summarised in this Journal,¹ the last 20 years has witnessed major improvements in Down syndrome screening such that detection rates of approximately 80%, for screen-positive rates of under 5%, are now readily achievable by either first or second trimester screening.^{2–4} However, such detection rates are only afforded by optimised screening: either combined (nuchal translucency and biochemistry) first trimester screening or second trimester triple or quadruple biochemical screening (free β -hCG, inhibin A, α -fetoprotein (AFP) \pm unconjugated oestriol). Other approaches to screening, such as nuchal translucency alone or dual analyte second trimester biochemistry (hCG and AFP), while considered acceptable only five or so years ago, are now simply not sufficiently adequate to be recommended for routine screening.

In this issue of this Journal, Peter O'Leary and colleagues report a survey of Down syndrome screening utilisation and practices across the states and territories of Australia.⁵ The survey clearly shows that there are significant disparities in the provision and availability of Down syndrome screening services across Australia. Some states have well-established screening programs encompassing both first and second trimester options while others would appear to have only rudimentary services. In the Northern Territory, for example, O'Leary *et al.* report that there are no accredited providers of nuchal translucency, suggesting that the provision of quality first trimester screening is compromised, whereas in Queensland, the Northern Territory and Tasmania, Down syndrome screening is taken up by less than half of all women. The survey not only identifies such apparent inequities of access to screening across Australia, but also highlights important differences in the operation of the various screening programs, including the indications for screening and the reporting of either risks at birth or risks at testing. Taken together, the picture that O'Leary and colleagues paint is one that we should not be proud of. It is one of haphazard screening where access to screening is more a matter of luck than choice. To make matters worse this is not the first time we have been shown this picture. Similar discrepancies in the provision of Down syndrome screening has been reported previously by Lawrence and Crowther⁶ through a survey of Australian hospitals, and Halliday *et al.* have described more local differences in access to prenatal diagnosis in Victoria.⁷ We are also not alone. New Zealand would appear to have developed a similarly inadequate patchwork of screening services.⁸ Further afield, in 2002 the UK National Screening Committee (NSC) published a survey of Down syndrome screening practices in England,⁹ revealing widespread differences in practices and standards very similar to those identified today by O'Leary *et al.*

However, the UK NSC has used the findings of their survey to guide the development of a coordinated national screening program that is 'equitable and of uniformly good quality', as directed and underpinned by their national screening policy.¹⁰ It is time for a similar national approach here. Hence, O'Leary and his colleagues⁵ are right to call for a uniform national screening policy in Australia, as is TC Chang in New Zealand.⁸ It is only in this way that we can ensure real and equitable choice for women in our respective countries, irrespective of their age, ethnic background or place of residence. Moreover, as we push for the development of those national policies and screening programs, preferably by our governments, we need to ensure that all aspects of those programs receive due attention. Effective screening for Down syndrome is more than an ultrasound scan and/or a blood test. Best practice requires the provision of high quality information to women and health professionals alike – and by all accounts, including a report in this issue of the Journal,¹¹ such information is badly needed. Rowe and colleagues report that, at least in the women they studied, the majority of decisions regarding uptake of screening were not well informed and that, in particular, women choosing to have screening were not sufficiently aware of the potential need for subsequent diagnostic testing and the risks inherent in that.¹¹ Others have previously reported similar findings for both women and health professionals,^{12–14} emphasising that, to be successful, a national screening program will need to audit the quality and provision of information given to women as part of their screening and their understanding of that information. This will be no small task. It will also be critical that providers of screening are required to formally report the performance of their screening program through linkage of screening undertaken to birth outcomes, as has been recently done in Victoria.³ Such reporting will be fundamental to ensuring adequate access to quality screening and the provision of real choice, not luck.

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