Screening for Down’s syndrome

Professor Nicholas J Wald and others (8 October, p 883) report a new screening test for Down’s syndrome with a higher detection rate and a lower false positive rate than currently available tests, which is likely to be welcomed by epidemiologists, obstetricians, and prospective parents. But as Drs Ian Donnai and Tony Andrews (8 October, p 876) argue, the problems in introducing any new screening test should not be underestimated.

One such problem is the effect upon women of receiving a false positive result. Although distress in women who have been told that they have raised maternal serum α-fetoprotein concentrations (indicating an increased risk of an open neural tube defect) has been documented,1 there has been no study of the effects of screening for Down’s syndrome. In a prospective study currently underway we are studying the impact of various prenatal test results on women consecutively booked for antenatal care at this hospital. Forty two women had abnormally low maternal serum α-fetoprotein concentrations that were subsequently shown to be false positive results. We report some initial findings that illustrate the psychological impact of receiving false positive results.

We report separately on women aged 38 and over (who are routinely treated as being at extra risk from the beginning of pregnancy and are offered amniocentesis for chromosomal analysis in this region) and women aged under 38 (a group who are not encouraged to consider themselves at increased risk of having a baby with Down’s syndrome), for whom an abnormal maternal serum concentration of α-fetoprotein is a new challenge to the pregnancy. In the older group those with an abnormal result show similar changes in anxiety at the time of the result to those with a normal result. Three weeks later when the results of any subsequent tests have shown the result to be false they were no more anxious than those with a normal result. The results are quite different for younger women receiving a false positive result: they show much more anxiety both at the time of receiving the test result and three weeks later when compared with women with normal results (figure). The mean score for women aged less than 38 receiving an abnormal result was 53·8 (SD 2·8), a score well above normal (mean 35·1 (9·2)), and within the range for patients with a diagnosis of general anxiety disorder (mean 49·0 (11·6)).2

Inadequate understanding of a test and poor preparation for potentially bad news are likely to be two factors contributing to this increased distress in younger women. Women do not have a good understanding of such tests; in a previous study 39% of the women could not even identify whether they had had blood taken to test for spina bifida.3 With adequate preparation the impact of a positive test result is likely to be reduced, as is evident in the pre-counselling of those using HIV antibody screening services.4 An association between maternal stress and obstetric outcome has been documented.5 Before introducing any new screening tests into routine obstetric care we need to ensure adequate counselling to minimise the distress now known to arise for women undergoing such tests.

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