

New screening halves the number of children born with Down syndrome

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A new national screening strategy in Denmark has halved the number of infants born with Down's syndrome and increased the number of infants diagnosed before birth by 30%, according to a study published on bmj.com today.

Many countries, including England, Australia and New Zealand, are trying to introduce national screening strategies for Down's syndrome, but are facing a variety of problems because of a lack of consensus about the screening policy and logistical challenges.

In 2004, the Danish National Board of Health issued new guidelines for prenatal screening and diagnosis. These included the offer of a combined test for Down's syndrome (based on combination of maternal age, plus serum and nuchal screening) in the first trimester. This test gave women a risk assessment for Down's syndrome at an early stage in the pregnancy. Women whose risk was higher than a defined cut off were referred for invasive diagnostic tests (chorionic villus sampling or amniocentesis).

In the previous guidelines screening for Down's syndrome was based on maternal age and a diagnostic test was mainly offered to women above 35 years.

Professor Ann Tabor and colleagues from Denmark, evaluated the impact of the new national screening strategy on the number of infants born with Down's syndrome and the number of referrals for invasive

procedures. They analysed data from the 19 Danish departments of gynaecology and obstetrics and the national cytogenetic registry, for an average of 65,000 births each year, between 2000 and 2007.

Uptake was good, by June 2006 all 15 Danish counties followed the guidelines from 2004 and offered the new screening strategy. In 2006 approximately 84% of pregnant women had a risk assessment for Down's syndrome.

The researchers found that the new strategy was associated with improved earlier detection of Down's syndrome, low false positive rates, and more than a 50% decrease in the number of invasive tests carried out each year.

They report that the number of infants born with Down's syndrome decreased from 55 per year during 2000, to 31 in 2005 and 32 in 2006. The total number of invasive tests fell sharply from 7524 in 2000 to 3510 in 2006.

The detection rate in the screened population was 86% in 2005 and 93% in 2006. With 3.9% (17) of women receiving a false positive result in 2005 and 3.3% (7) in 2006.

The authors point out that the value of this new screening strategy is that all women can be assessed early in pregnancy (in the first trimester). The national guidelines emphasise that risk assessment should only be done if women choose the test on the basis of informed choice, therefore despite the programme being available to all pregnant women in Denmark, some will still choose not to be screened.

The authors conclude by emphasising Denmark's success at building a strong national organisation for fetal medicine and a national quality database that allows follow-up of all screened women at a national level

and quality control of the new national screening programme.

Source: British Medical Journal

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