

Prenatal Diagnosis - making difficult decisions

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An expanded and updated version of this article is available in Sarah's book: *Gentle Birth, Gentle Mothering: The wisdom and science of gentle choices in pregnancy, birth, and parenting*. Details are available at www.sarahjbuckley.com.

Prenatal diagnosis - that is, testing for abnormalities in unborn babies - has become an almost universal part of pregnancy care in Australia in 1999. Close to 100 per cent of pregnant women choose to have an ultrasound to check for major physical abnormalities at 18 to 20 weeks, which is the most common and most obvious form of prenatal diagnosis.

Maternal Serum screening

Rising numbers of women are also accepting maternal serum screening (MSS), which involves a blood test taken between 15 and 18 weeks, though exact dates are needed for accurate results. MSS can detect babies at higher risk of Down's syndrome and the neural tube defects - NTDs - anencephaly, where the brain is very underdeveloped, and spina bifida, where the spinal cord coverings are incomplete and the lower body may be paralysed. Local experience has shown that a range of other uncommon conditions may sometimes be picked up by MSS; however, because MSS is a screening test, a definite diagnosis is never possible from MSS alone.

Uptake of MSS currently varies from state to state, with South Australia having the most organised and popular programme. In SA 75 per cent of women choose to have this test, while numbers in other states are much lower. It is likely that in the future, MSS will be offered to all pregnant women, as it is in the UK, and an earlier MSS is currently being developed.

Nuchal translucency

This is a new test which is increasingly used. It takes the form of a specialised ultrasound, performed at 11 to 13 weeks, and uses specific equipment and analysis to measure the thickness of the pocket of fluid that all babies of this age have at the back of the neck. Babies with extra fluid have an higher risk of Down's syndrome, and the result of this test, which also takes into account the mother's age and number of weeks pregnant, indicates whether the risk is high enough to recommend amniocentesis or CVS for chromosomal analysis. Nuchal Translucency can sometimes detect babies with other chromosomal abnormalities.

Other tests on the increase

As well as these tests, increasing numbers of older women, or women who have had a "positive" result from MSS or Nuchal Translucency, are choosing to have amniocentesis or Chorionic Villus Sampling (CVS). These are two different methods of obtaining cells for chromosomal analysis, which is the definitive test for Down's syndrome and other chromosomal abnormalities. Chromosomal abnormalities are caused by defects in one of the 23 pairs of chromosomes that, deep inside our cells, carry all our genetic characteristics.

Amniocentesis involves taking a 15 ml sample of the amniotic fluid, which surrounds the baby, at 15-17 weeks. Fluid removed by amniocentesis can also be checked for spina bifida. In CVS, a small fragment of placenta is removed with a syringe, either through the abdomen or via the vagina, at 10 to 12 weeks..

Diagnostic vs Screening tests

Tests for prenatal diagnosis are either diagnostic or screening tests. Diagnostic tests, such as amniocentesis or CVS, can tell you whether or not your baby has the abnormality that is being tested for. In contrast, screening tests such as MSS and Nuchal Translucency, are not accurate enough to give a definite yes or no. Instead, you are given an estimation of risk, which is usually "positive", or "high-risk", when the chance of abnormality is around one in 250 or greater. This estimate of risk can be used in deciding whether to proceed with a diagnostic test, which is usually amniocentesis, CVS or, in the case of spina bifida, ultrasound.

Levels of accuracy

The inaccuracy of screening tests means that there is an unavoidable down side. Not all affected babies will be identified, and many mothers carrying unaffected babies will have the unnecessary worry of a “positive” result. With nuchal translucency, for example, 19 out of 20 women who get a “positive” result will not be carrying an affected baby, but will go through counselling and amniocentesis (with a 1 in 200 risk of causing miscarriage), then wait days to weeks before reassuring results are back. Some women who have been through this experience report that they felt anxious about their baby even after this reassurance, and others have felt that they lost some of the enjoyment of their pregnancy.

New developments

In the area of prenatal diagnosis, many new developments are designed because of problems with existing tests. For example, CVS was developed as an early test, so that women can have the results of their chromosomal analysis when a early termination, which is a much simpler procedure, is still an option. In contrast, after amniocentesis at 15 to 17 weeks, termination is usually performed by an induced labour and birth, which can be a traumatic and painful procedure.

For the same reasons, MSS will soon be available earlier in pregnancy - probably from 10 weeks - making CVS, at 10 to 12 weeks, an option for women with “positive” results. A major problem with this approach, however, is that some women will have a termination for an affected baby who would naturally have miscarried soon afterwards.

MSS and nuchal translucency were developed as “no-risk” tests that could ensure that only those babies who are most likely to have an abnormality would be subjected to amniocentesis and CVS, both of which slightly increase the chance of miscarriage afterwards. Because of their safety, MSS and nuchal translucency encourage younger women to accept screening. This further increases the detection of babies with Down’s syndrome, the majority of which are born to women in their younger years.

Ideally, early and safe tests would be also be available for chromosomal analysis. Researchers are currently refining methods to detect the small numbers of fetal cells that circulate in the mother’s blood, even in the early weeks. At present, this is too expensive, and requires too much blood, to be practical, but if it were possible, it would remove the need for CVS or amniocentesis.

Problems with chromosomal analysis have generated new ways of testing cells from amniocentesis and CVS. Some services are now using the Fluorescent In-Situ Hybridisation (FISH) test, which detects selected chromosomal abnormalities with a 24 hr processing time. This is in contrast to standard cell-culture which takes around two weeks to give results. Standard testing, however, is more comprehensive.

Abnormalities looked for

It is estimated that around two per cent of babies are born with a major congenital abnormality. We still do not know the causes of most of these abnormalities, so prevention has a limited place at present. The only exceptions are the Neural Tube Defects (spina bifida and anencephaly). Starting folic acid supplements before conception can drastically reduce the chance of NTD.

Spina bifida can usually be detected on routine ultrasound at 18–20 weeks. Ultrasound can usually give an estimate of severity. Anencephaly, which is always fatal, is obvious even on an early ultrasound, and is also picked up on MSS. Overall, MSS can detect up to 90 per cent of babies with severe NTD.

Down’s syndrome is the most common chromosomal abnormality in our population. It is generally not inherited, but women giving birth in their later years are more likely to have a baby with Down’s syndrome. Down’s syndrome is associated with intellectual disability, but it is not possible whether a baby will be mildly or severely affected. Babies born with Down’s syndrome can also have heart and gut abnormalities, most of which are operable.

Babies with Down’s syndrome have a high rate of miscarriage. Even after 16 weeks, 1 in 3 affected babies will miscarry. Parents faced with the prenatal diagnosis of Down’s syndrome can have a very difficult decision.

Single gene defects such as cystic fibrosis and thalassemia can be identified by analysing fetal cells using genetic probes. Most single gene defects are hereditary. Families who may be at risk of inherited conditions need to see a genetic counsellor to discuss the use of these tests, which are extremely expensive outside the public system, and which are carried out after amniocentesis or CVS.

Shades of Grey

While Down's syndrome and spina bifida are the conditions that are being targeted, there are many more abnormalities that can be detected by current methods of prenatal diagnosis. However for many of these conditions, the outcome for the baby can be unclear. For example, disorders of the sex chromosomes, which account for about one third of reported chromosomal abnormalities, can give subtle or uncertain levels of abnormality. Parents who are given an uncertain or ambiguous diagnosis are particularly in need of skilled counselling and support.

High-quality counselling is also needed before testing to ensure that women make an informed decision about accepting the tests. Reports from research, and from women's experience indicates that this is not always provided. Doctors may be too busy to take the time to explain, and providing trained counsellors for every woman who opts for prenatal diagnosis would be very difficult and expensive.

While prenatal diagnosis is said to be economically cost-effective, especially for conditions where a child would require constant and ongoing support and supervision, the "cost" to women and their families of this new technology is not often explored. Some women are definitely grateful for the opportunity to terminate an affected pregnancy and "start again", but for other women the pain of choosing to terminate a wanted pregnancy can take years to resolve.

Prenatal diagnosis is based on the assumption that discovering and terminating an affected pregnancy is advantageous, even when the condition detected would be lethal to the baby at a later time. The reported experiences of a few couples who did not choose termination in this circumstance, and whose babies miscarried or died naturally soon after birth, would contradict this preconception. For some women, not being able to see or hold their baby can prolong the grieving.

Most women choose to have prenatal diagnosis, because they want the reassurance that their baby is normal. However our current tests cannot give this guarantee. Perhaps we are expecting too much of this technology, and in our striving for the perfect baby, we are producing a system that has its own share of heartache.

The future

If we follow overseas trends, all pregnant women will soon be faced with these decisions about risk and worry, and a few will be waiting anxiously for results which will determine whether they will terminate wanted pregnancies. Women who receive bad news will be faced with choosing "the tragedy of her choice" as Barbara Katz Rothman, author of *The Tentative Pregnancy*, describes this difficult decision.

If you are considering tests for prenatal diagnosis, you will need to think through the possible consequences. *Which Tests for my Unborn Baby* and *Prenatal Testing: Making Choices in Pregnancy* which are both co-authored by Lachlan de Crespigny, an expert in this area, are highly recommended, along with *The Tentative Pregnancy*. Good luck!