



A testing time...

Could you cope or afford a handicapped child? Would you consider a termination?

Gillian Handley looks at the ethical dilemmas posed by pregnancy testing and how she coped.

Prenatal testing can tell what's in store, but it can't tell you what to do. Everyone hopes for a perfect baby, but there are no guarantees: one in 20 babies born in Australia has some type of congenital abnormality and three per cent of these are serious.

The range of tests to check if anything is wrong range from screening to more invasive procedures. But the choice to test is more than a decision about the range of tests available, it also raises tough questions about what we do if we don't get the result we want.

An innocent approach

Recently, I had an amniocentesis. This was more by accident than design, and while I recognise the medical logic behind it, I wonder if I would do it again.

I am 38, so before I'd even met my obstetrician, I was referred to a genetic counsellor, just as a matter of course.

Resentful that I was part of a number-crunching exercise, I made an appointment to see the counsellor. What I should have done was spend more time examining my husband's and my own feelings about the whole concept of testing.

Ultimately, I chose the nuchal translucency test (see *Pregnancy testing guide* on opposite page) to, hopefully, eliminate the need for further tests.

The nuchal translucency test requires an ultrasound. Although I could see the foetus quite clearly on screen, I tried to pretend it wasn't mine. What on earth do I do if something is seriously wrong?

While the result was within the "high range of normal", it revealed an increased risk of foetal abnormality. I was left with the option of doing nothing and living with the uncertainty of a high-risk assessment, or choosing one of the invasive procedures for a definite result.

The figures reckoned my risk of foetal abnormality was higher than my risk of miscarriage. At this point, I really began to consider the implications of a damaged foetus. Could I cope with or afford a handicapped child? Termination: would I consider it? I found the best way through this minefield of decisions was to take them one step at a time.

Considering my increased risk, I reluctantly chose to take further tests. Since I was going through all the moral, ethical and emotional stress of anticipating the decision, I thought I should have the test to see if all this angst was necessary.

“Testing raises very complex issues. I found the best way to deal with them was to move through each issue and option one by one”

As if the decisions themselves were not difficult enough, I had to keep an eye on the clock. The nuchal translucency is done between 11 and 13 weeks. I was too late to have chorionic villus sampling (CVS), so at 14 weeks I would have to have an amniocentesis. This was far easier than I expected. It took 15 minutes and was practically painless.

I took the doctor's advice and rested for the day to minimise the risk of a miscarriage. The results took two weeks, which was an agonising wait, but at least I knew that if there was something wrong with the baby, then the decision about what to do would be much worse.

Worst case scenario

What I feared most was that the results would show something was wrong, but not how badly – I would still be making a choice partly in the dark.

The baby may have Down's syndrome, but there would be no way of knowing how mild or how severe the condition would be. What would I do if the baby was seriously damaged? I found it confusing, emotionally draining and overwhelming to think about all my possible options.

Ultimately, my test results were normal. This was a blinding relief, but it left me feeling a bit off balance. I still feel like I dodged some serious moral and ethical questions that I should have been able to resolve somehow.

From my experience I would offer this advice: find out about prenatal testing before you agree to it. Decide beforehand, if you can, why you want the information and what you will do with it. Spend some time with a genetic counsellor and talk frankly about any of your concerns.

Right or wrong?

Testing raises very complex issues. I found the best way to deal with them was to move through each issue and option one by one. Testing may raise more questions than answers, but there is no right or wrong action, either about prenatal testing or what to do if the results show an abnormality. It's your baby and your choice.

Today's testing standards

Traditionally, women aged over 35 were advised to attend genetic counselling. Today, however, the more accurate screening tests mean women of all ages can have less invasive tests.

Seventy per cent of all women having babies are aged under 35. If age alone is used as a screen, then only 30 per cent of Down's syndrome babies in the population would be detected.

WHAT IS A CHROMOSOME DISORDER?

A chromosome contains genetic material called DNA. This carries all the information needed to make a new human being. If there are too many or too few chromosomes, or if they are damaged, then this information will be incorrect and a congenital abnormality may occur.

In the future, more specialised and specific screening may phase out invasive testing. Active research into a blood test that can pick up foetal blood cells in the maternal blood stream is ongoing.

AGE OLD RISKS

Maternal age	Risk of Down's syndrome	Risk of all chromosome abnormalities
20	1:1923	1:526
25	1:1205	1:476
30	1:885	1:384
35	1:365	1:178
40	1:109	1:63
45	1:32	1:18

Pregnancy testing guide

There are two types of tests: screening tests to work out a risk factor, which are usually less invasive; and diagnostic tests that confirm or exclude chromosomal abnormality, but are more invasive.

Screening tests

TRIPLE TEST

This is a blood test from the mother, usually between 15 and 18 weeks. The test works out the risk specifically for each woman and can detect 60 per cent of Down's syndrome. It also screens for Neural tube defects like spina bifida. If the result is abnormal for either defect, you'll be offered another test.

NUCHAL TRANSLUCENCY ULTRASOUND

This non-invasive procedure is done between 11 and 14 weeks. It involves either an abdominal or vaginal ultrasound, depending on the site of the placenta or shape of the uterus, and can detect between 70 and 80 per cent of Down's syndrome cases. The ultrasound measures the fluid under the skin at the back of the neck of the foetus. This fluid tends to be greater when a foetus has a chromosome disorder. If the risk is high, diagnostic testing, such as chorionic villus sampling (CVS), may be offered.

Diagnostic procedures

AMNIOCENTESIS

This is an invasive test done at 14 to 16 weeks. A sample of amniotic fluid is taken by passing a needle through the abdomen. The procedure is done under ultrasound guidance to ensure the baby isn't hurt. Results take about two weeks and the chance of miscarriage following is less than one per cent, or one in 200. Amniocentesis provides a definitive answer about Down's syndrome, but doesn't pinpoint single gene defects, such as cystic fibrosis.

CHORIONIC VILLUS SAMPLING (CVS)

This test is similar to an amniocentesis, whereby the amniotic fluid is aspirated through the cervix, however it carries a slightly higher risk of miscarriage. Since CVS is performed between 10 and 12 weeks, if the decision is taken to terminate the pregnancy, the woman can have a suction dilatation and curettage (D&C), rather than go through labour.

FISH (FLUORESCENT IN SITU HYBRIDISATION)

This test gives a fast result, 24 hours following an Amnio or CVS, but it only screens for certain chromosomes.

by Julie Hamilton