Screening for Down, Edwards and Patau Syndromes in Pregnancy

What is the test?

You will be offered a screening test called the Combined test. In England this is offered to all pregnant women between 11+2 and 14+1 weeks of pregnancy. A screening test will give you a result which will show how likely it is that your baby has one of these conditions. It will give you a 'high' or 'low' chance result but cannot tell for certain whether the baby will be affected or not. The test measures substances in the mother's blood which are transferred from the placenta, together with a measurement taken by ultrasound scan of the thickness of the fluid at the back of the baby's neck (nuchal translucency). A computer program uses these results and together with your age, weight and how many weeks pregnant you are will give a chance for each individual pregnancy. It is important to know your ethnicity, whether you smoke or not, whether you are on medication such as insulin and also if this is an IVF pregnancy - as all these factors can affect the result.

What are the syndromes that are being tested for?

Usually people are born with 46 chromosomes - these are packets of genetic material inherited from both the mother and father. Occasionally when the sperm and egg meet to form a baby there can be an extra copy of one of these chromosomes. An extra copy of chromosome 21 will give Down syndrome, an extra copy of chromosome 18 will give Edwards syndrome and an extra copy of chromosome 13 will give Patau syndrome. People with Down syndrome, like everyone, vary a great deal in appearance, personality and ability. There will always be a certain degree of learning difficulties but these can vary with each individual and cannot be predicted at birth. There are a number of health problems associated with Down syndrome but these too will vary from person to person.Edwards syndrome and Patau syndrome are rarer than Down syndrome, but much more serious. Many babies with Edwards and Patau syndromes are often miscarried or stillborn and sadly, if these babies are born alive, most will not survive the first few months of life. More information can be found in your "Screening tests for you and your baby" booklet.

What information will the test tell me?

The result will tell you the chance of the baby being affected by these syndromes and will be recorded as a chance of '1 in …' For example if your result was 1 in 500 it means for 500 women like you, there would be 499 babies born without these syndromes, and one baby would be affected. If you choose to be screened for all 3 syndromes you will be given 2 results, one for the chance of your baby having Down syndrome and one for the chance of your baby having either Edwards or Patau syndrome. It is not possible to give separate results for Edwards and Patau syndrome.

How will I get my results?

If your result shows the chance of the baby being affected is less than 1 in 150 (for example 1 in 500) then this would be classed as a lower chance result and you will receive a letter in the post with this information. If you have not received a letter 10-14 days after having the test please contact your Community Midwife or the Screening Coordinator (contact number at the end of this leaflet). If the result shows the chance of the baby being affected is greater than 1 in 150 (for example 1 in 5) then this is an increased or higher chance result and you will be contacted directly to discuss the result further.
About 1 in 30 women will have a higher chance result but even with a higher chance result, it is still more likely that your baby will not be affected.

One of the substances measured in your blood is called PAPP-A (Pregnancy Associated Plasma Protein – A). Occasionally the blood test will show a low level of PAPP-A. This by itself does not put the baby at risk of having one of these syndromes, but we know that sometimes the baby can grow more slowly in later pregnancy. If this is the case in your pregnancy we will contact you directly, arrange further appointments to check how your baby is growing and send you further information.

Do I have to have the test?

No. The test is optional so you can decide whether you wish to have the test or not. Some women will wish to know about some of the syndromes but not all of them. You can choose to have screening for all 3 syndromes, just Down syndrome or just Edwards and Patau syndrome. Your choices will be documented on the screening request form.

You should think about the possible results before you have the test and whether you feel it is important for you to know whether your baby has one of these conditions or not. If you have a higher chance of the baby being affected would this change anything about the way you feel about your pregnancy?

What happens if I get a higher chance result?

If you get a result which shows that your baby is at an increased chance of these syndromes you will be offered a diagnostic test - a test that will tell you a definite yes or no about whether your baby has one of these syndromes or not. A diagnostic test involves putting a fine needle through the mother's abdomen and taking a sample of placental tissue or amniotic fluid. Unfortunately there is a small chance of miscarriage associated with the diagnostic test of around 1 in 100 or 1%. If you have opted for one particular syndrome to be screened for and declined the others it is important to know that a diagnostic test will give you an answer for all 3. It is not possible to test for individual syndromes at that point.

Are there other tests available?

The UK National Screening Committee (UKNSC) is the expert group who advise which tests should be offered to pregnant women. They advise that screening for Down, Edwards and Patau syndromes should be offered on the Combined test. Some women however, can be unsure of their dates and it can be too late for the combined test or occasionally it is not possible to measure the nuchal translucency at the dating scan due to position of the baby or poor views. In this case the UKNSC advises we offer the Quadruple test, but only screening for Down syndrome. The Quadruple test measures four substances in the mother's blood and is available from 14+2 and 20+0 weeks. Screening for Edwards and Patau syndromes is not possible on the Quadruple test but is a recommended part of the Fetal Anomaly Scan at 18+0 to 20+6 weeks.

One of the substances measured in your blood if you have a quadruple test is called Alpha-fetoprotein (AFP). Occasionally the blood test will show a higher level of AFP. Most babies with a higher level of AFP do not have any problems but very occasionally it can be associated with Spina Bifida or an abdominal wall defect (a hole in the tummy wall). If this is the case in your pregnancy we will contact you directly, and arrange an early scan to check your baby's spine and tummy wall. The recommended pathway for screening for spina bifida or abdominal wall defects is the fetal anomaly scan at 18+0 – 20+6 weeks of pregnancy so if you have a combined test, you will still be screened for these conditions.

Unfortunately it is not possible to offer a test for any of these syndromes after 20 weeks as the combined scan and blood test information will not give an accurate result.

Other tests, using free Fetal DNA, are available but at present only privately. These analyze genetic information (DNA) from the baby's cells which are present in the mother's blood in small amounts. These tests can give a risk result for all 3 syndromes but is a blood test from the mother so does not carry a chance of miscarriage. These still only act as a screening test and you would be offered a diagnostic test if you were given a higher chance result.

Where can I find more information about these syndromes and the tests?

Antenatal Screening Coordinator 01603 286802 (Monday-Friday)

National Screening Committee website https://www.gov.uk/topic/population-screening-programmes/fetal-anomaly

Antenatal Results and Choices www.arc-uk.org and helpline 0207 6310285