

Prenatal Genetic Screening Tests

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Screening tests do not give a yes/no definitive answer. Screening can tell you how likely is that a baby has a genetic condition; in other words, whether there is a low chance or a higher-than-average chance of a baby having a genetic condition. If a screening test finds your baby has a higher chance of having a genetic condition, you are likely to be offered a procedure called chorionic villus sampling (CVS) or amniocentesis (amnio) so that genetic testing can be done to give a yes/no answer.

1. Screening tests offered by the NHS

All women in England, Scotland and Wales are offered screening tests for Down's syndrome, Edwards syndrome and Patau's syndrome and sickle cell disease and thalassaemia. The conditions are also known as trisomy 21 (T21), trisomy 18 (T18) and trisomy 13 (T13), because in each case a baby has three copies of the chromosome rather than the typical two.

These tests are optional; it is your individual choice to have these screening tests or not. It can help you decide if you know about the conditions being screened for. Find out more here:

- Down's syndrome: <https://www.downs-syndrome.org.uk/>
- Edwards' syndrome and Patau's syndrome: <https://www.soft.org.uk/>

- Sickle cell disease: <https://www.sicklecellsociety.org/>
- Thalassaemia: <https://ukts.org/>

2. Non-invasive prenatal testing (NIPT)

NIPT is also known as non-invasive prenatal screening (NIPS) or cell-free DNA screening and is carried out using a maternal blood test (blood is taken from the mother's arm). You may be familiar with some of the brand names such as 'Harmony', 'Panorama' or the 'SAFE test'. All of these are examples of NIPT. In the laboratory, DNA fragments that have come out of cells from the mother and fragments that have 'leaked' from the placenta are extracted from the blood sample. The analysis checks for extra DNA material associated with chromosomes 21 (suggesting Down's syndrome), chromosome 18 (suggesting Edwards' syndrome) and chromosome 13 (suggesting Patau's syndrome) and gives an assessment of the baby's chance of having these conditions.

We know NIPT is a very reliable screening test for Down's syndrome, and what the test result predicts is usually correct. NIPT for Edwards syndrome and Patau's syndrome is not as reliable. The results from NIPT are more accurate than conventional screening (e.g. nuchal translucency scan + blood test) but it is important to remember that it does not provide a yes/no answer. Occasionally, NIPT will not give a result. This can be for a variety of reasons. A second test will always be offered. If there is no result a second time it is important to seek expert clinical advice.

In England, Scotland and Wales, NIPT is offered to women whose screening result for Down's syndrome, Edwards' syndrome or Patau's syndrome shows there is a greater than 1 in 150 chance (so 1 in 2 to 1 in 150).

Some private clinics offer NIPT for a wider range of conditions. There is limited evidence on how accurate NIPT is for other conditions, so it may be helpful to discuss having NIPT for other conditions with an independent genetic counsellor before proceeding.

Non-invasive prenatal diagnosis (NIPD) refers to testing using the same technique that can give a definite answer for some single gene disorders.

3. Ultrasound scans

Ultrasound scans can show images of a baby's body, inside and outside. Although a scan cannot show genetic changes, it can show physical features in the baby that might suggest a genetic condition. Often it will be unexpected scan findings that lead to the offer of invasive procedures - usually chorionic villus sampling (CVS) or amniocentesis (amnio) – so that genetic diagnostic testing can be done. In the NHS, the two main scans will be offered at 12 weeks and around 20 weeks of pregnancy.

ARC - Call our helpline

Want to talk? We're here as often as you need us. Our helpline is answered by trained staff.

020 7713 7486

Monday to Friday: 10.00am – 5.30pm

Or text: **07908 683004**

Tuesday and Thursday: 8 – 10pm – email
info@arc-uk.org to arrange an evening call.

Disclaimer

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