



**University
Hospitals Sussex**
NHS Foundation Trust

Combined screening in pregnancy

Patient information

This leaflet has been written to give you more information about having the combined screening test. This leaflet should be read in conjunction with 'Screening tests for you and your baby' which you can access via your digital notes portal or at Screening tests for you and your baby (STFYAYB) - GOV.UK (www.gov.uk).

What is combined screening?

Combined screening is a test offered to all pregnant women and people between 11 weeks and two days and 14 weeks and one day to help identify pregnancies that have a higher chance of being affected with one of three chromosomal conditions: Down's syndrome (trisomy 21), Edwards' syndrome (trisomy 18) and Patau's syndrome (trisomy 13).

It is a screening test, which means it will not tell you definitely whether your baby has any of these conditions. This test can only tell you the chance of your baby being affected. You can then use this information to help decide whether to go on and have further tests. This test is optional-it is your choice whether to have this test or not.

Your midwife will ask if you would like screening for:

- All three conditions (one chance for Down's syndrome and one chance for Edwards' and Patau's syndromes combined).
- Down's syndrome only
- Edwards' and Patau's syndromes (one chance for both conditions)
- None of the conditions.

How does combined screening identify pregnancies at higher chance of Down's, Edwards' or Patau's syndrome?

It is called combined screening because it combines information from an ultrasound scan and blood test to work out the chance of your baby having a chromosomal condition. The ultrasound scan checks your dates and measures the amount of fluid behind the baby's neck, called the nuchal translucency. An increased amount of fluid is more likely in babies with chromosomal conditions. The blood test is taken from you after the scan. It measures the levels of two substances found in your blood during pregnancy which may be altered if the baby has a chromosomal condition.

Other factors which can affect the result are also taken into account: these include your family origins, age, weight, smoking history, pregnancy history and medical history.

How reliable is combined screening?

Combined screening identifies about 85% of babies with Down's syndrome and 80% of babies with Edwards' or Patau's syndrome.

When will I get the results?

This depends on whether your result is assessed as a higher or lower chance for the conditions you chose to be screened for.

A lower chance result (also called a low chance result):

Any result below 1 in 150 is considered to be a 'lower chance'. (This is less than 0.7%, such as 1 in 200, 1 in 850, 1 in 4400, etc.). The bigger the number, the lower the chance. This does not rule out the condition in your baby but means it is less likely. We will write to you within two weeks of having the test if you have a low chance result. If you have not received your result by this time, please contact the screening midwives (contact phone number available in the 'For More Information' section).

A higher chance result (also called a high chance result):

Any result of 1 in 150 or higher (such as 1 in 120, 1 in 50, 1 in 8, etc.) is considered to be 'higher chance'. The smaller the number, the higher the chance. In these cases, a screening midwife will call you, usually within three working days. We will offer you an appointment to discuss the result in more detail and discuss whether you want to have further tests. Remember, a higher chance result does not mean there is definitely a problem with your baby and most women and people given this result will go on to have a baby that does not have Down's, Edwards' or Patau's syndrome.

Further tests

If you have a lower-chance result you will not be offered a further test. If you have a higher-chance result, you may decide:

- not to have any further testing
- to have a second screening test (called NIPT) to obtain a more accurate screening result before choosing whether or not to have a diagnostic test
- to have a diagnostic test straight away.

Whatever results you get from any of the screening or diagnostic tests, you will get support and care to help you decide what to do next.

Non-invasive prenatal testing (NIPT)

The second screening test is another blood test called NIPT (non-invasive prenatal testing). NIPT is more accurate than the combined or quadruple test. It works by measuring DNA (genetic material) in your blood. Some of this DNA will be from the baby's placenta. If there is more DNA than expected from chromosomes 21, 18 or 13 it may mean that your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome. Like every screening test, though, it does not give a definite answer. NIPT cannot harm your baby. Most women and people will get their result within 2 weeks.

Most people will get a lower-chance result, meaning your chance of having a baby with the condition is low. You will not be offered a diagnostic test.

If your NIPT result shows a higher-chance result then the chance of your baby having the condition is high. You will then be offered a diagnostic test, though it is up to you whether or not to have this.

Diagnostic tests (CVS or amniocentesis)

Diagnostic tests give a definite answer. They test cells from the placenta or fluid surrounding your baby.

1 out of 200 women and people (0.5%) who have a diagnostic test will miscarry as a result of the test. There are 2 types of diagnostic test: chorionic villus sampling (CVS) and amniocentesis. More information about diagnostic tests is available in the leaflet 'CVS and Amniocentesis' or you can speak to your doctor or midwife.

What happens if I am over 14 weeks and one day pregnant at scan?

If you are over 14 weeks and one day at the scan it is too late to have combined screening. However we can offer you another screening test, called the 'quadruple test'. This is a blood test that measures the amount of four substances in your blood to work out the chance of the baby having Down's syndrome. Unfortunately it is not possible to offer women and people who book after 20 weeks of pregnancy any screening tests for chromosomal conditions.

Can combined screening be used if I am having twins?

Yes, combined screening can be performed if you are expecting twins.

Can any other anomalies be identified by combined screening?

If the amount of fluid on the baby's neck is increased at 3.5mm or more, this may indicate a rarer chromosomal, genetic or structural problem in the baby. In such cases we would discuss this with you and offer a detailed heart scan as well as invasive testing. Finding an increased amount of fluid does not mean there is definitely a problem.

It is important to remember that no test can guarantee that your baby will be free of all anomalies.

For more information

If you have further questions about combined screening, you can speak to your midwife or doctor. You might also want to contact the following:

Antenatal Screening Midwives:

A team of local midwives specialising in antenatal screening and testing. Please contact them on:

Mobile: 07876 357 423

Royal Sussex Country Hospital: 01273 696955 Ext. 67477/62755

Princess Royal Hospital: 01444 441881 Ext. 65404

ARC – Antenatal Results and Choices:

National organisation providing support and information about screening and diagnosis of fetal anomaly.

www.arc-uk.org Helpline 0207 713 7486

Down's Syndrome Association:

Provides information and support for people with Down's syndrome, their families and carers.

www.downs-syndrome.org.uk Helpline 0333 1212300

S.O.F.T. UK:

Provides information support for families affected by Patau's syndrome

(trisomy 13) and Edward's syndrome (trisomy 18).

www.soft.org.uk Helpline 0300 102 7638

If you have vision, mobility or access issues please contact the specialist screening midwife team on **07876 357 423** for further information.

This leaflet is intended for patients receiving care in Brighton & Hove or Haywards Heath

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