



Antenatal Screening

Antenatal Screening for Down's Syndrome
and Edwards Syndrome

The Quadruple Test

Questions and Answers for women considering
the test

The Wolfson Institute of Preventive Medicine
Barts and The London School of Medicine and Dentistry

Antenatal Screening

This leaflet answers some of the common questions women ask about their screening test – we hope you find it helpful. You are welcome to discuss the test with your midwife or consultant before you decide whether you would like to be screened. If you have any further questions screening staff at the Wolfson Institute are available to talk to you on 020 7882 6293.

What is Down's syndrome?

Down's syndrome (trisomy 21) is defined by the presence of an extra chromosome number 21 in the cells of the fetus or affected individual. In an unscreened population about 1 in every 500 babies is born with Down's syndrome. Usually it is not inherited and so a baby can be affected even if there is no history of Down's syndrome in the family.

Down's syndrome is the most common cause of severe learning disability and is often associated with physical problems such as heart defects and difficulties with sight and hearing. It is not possible to assess the degree of disability before the baby is born. About 9 out of 10 babies with Down's syndrome will survive their first year and nearly half of these will reach 60 years of age.

What is Edwards syndrome?

Edwards syndrome (trisomy 18) is defined by the presence of an extra chromosome number 18 in the cells of the fetus or affected individual. Edwards syndrome affects multiple organs with a high risk of fetal death.

At 16 weeks of pregnancy Edwards syndrome has a prevalence of about 1 in 1,500 in an unscreened population.

Babies born with Edwards syndrome, on average, live for around two weeks with only 1 in 12 surviving for one year or more. Babies born with Edwards syndrome typically have severe structural defects (including defects of the heart, kidney, brain, skeleton and face) as well as severe learning disability.



What is the screening test procedure?

If you have the test one blood sample will be taken from the 14th to 22nd week of pregnancy.

The Quadruple test involves:

- i) taking a sample of your blood to measure the concentration of alphafetoprotein (**AFP**), unconjugated oestriol (**uE₃**), total human chorionic gonadotrophin (**total hCG**) and **inhibin-A**.

In pregnancies with Down's syndrome, the AFP and uE₃ levels *tend* to be low and the total hCG and inhibin levels *tend* to be raised.

In pregnancies with Edwards syndrome the AFP, uE₃ and total hCG levels *tend* to be low.

What is a 'risk'?

A risk is the chance of an event occurring. For example, a risk of Down's syndrome of 1 in 100 means that if 100 women have this risk, we expect that 1 of these women will have a baby with Down's syndrome and that 99 will not. This is the same as a 1% chance that the baby has Down's syndrome and a 99% chance that the baby does not.

When will the screening result be available?

For most women the result of the test is usually ready within 3 days of the sample being taken. Results are sent to your antenatal clinic and a letter is sent to you to let you know that the result is ready.

Your screening result is either screen-positive or screen-negative.

What does a screen-positive result mean?

A screen-positive result means that you are in a high risk group (equal to or greater than 1 in 150) for having a baby with Down's or Edwards syndrome. If you are in this group, you will be offered a diagnostic amniocentesis or possibly a chorionic villus sampling (CVS) (see page 5). Most women with screen-positive results will not have a pregnancy with Down's or Edwards syndrome.

What does a screen-negative result mean?

If the risk of Down's or Edwards syndrome is lower than 1 in 150 then the result is called screen-negative.

Although a screen-negative result indicates that your risk of having a baby with Down's or Edwards syndrome is low it cannot rule out the possibility of a pregnancy with these disorders (false-negative result).

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Does the Quadruple test detect all pregnancies with Down's or Edwards syndrome?

About 7 to 8 out of 10 cases of Down's syndrome are detected (screen-positive). This means that 2 to 3 out of 10 pregnancies with Down's syndrome are missed (screen-negative).

About 6 to 7 out of 10 cases of Edwards syndrome are detected. About 3 to 4 out of 10 cases are missed.

Is my age taken into account?

Yes, any woman may have a baby with Down's or Edwards syndrome but the chance of this happening increases as a woman gets older and so we use age as one of the factors when working out your risk of having a pregnancy with any of these disorders.

What happens if the ultrasound scan shows that I am too late for the Quadruple test?

Unfortunately if you are too late for the Quadruple test there is no other screening test available. It is possible an ultrasound scan could be performed to identify whether any major abnormalities associated with Down's or Edwards syndrome are present



What are the diagnostic tests?

If your Quadruple test result is screen-positive, you will be offered a diagnostic test, usually an **amniocentesis** or possibly an **chorionic villus sampling (CVS)**.

The diagnostic test will determine whether or not the pregnancy is actually affected with Down's syndrome, Edwards syndrome.

Amniocentesis

An amniocentesis is a widely used procedure. It is performed at about 16 weeks of pregnancy and involves collecting a small sample of amniotic fluid from around the baby by inserting a needle through the abdominal wall. This fluid contains cells from the baby. Down's and Edwards syndrome are diagnosed using a technique called quantitative fluorescence polymerase chain reaction (QF-PCR) which provides a rapid diagnosis for Down's or Edwards (and Patau) syndrome usually within 48 hours of the amniocentesis being performed.

Chorionic Villus Sampling (CVS)

CVS involves taking a sample of placental tissue, by inserting a needle through the abdominal wall or a fine instrument through the cervix. Down's, Edwards syndrome are diagnosed using QF-PCR which also provides a rapid diagnosis usually within 48 hours. There is a small chance (about 1 in 100) that the CVS will not provide a conclusive diagnosis. If this happens you will be offered an amniocentesis in order to provide a definitive diagnosis.

Will these diagnostic tests guarantee that my baby is free of all birth defects?

No test can guarantee that your baby will be free of all birth defects but if the result of the amniocentesis or CVS is negative it will almost certainly rule out Down's, Edwards syndrome.

Is it safe to have a CVS or an amniocentesis?

Amniocentesis and CVS have been offered for many years but the procedure is not without risk. It is estimated that about 1 in 100 women will have a miscarriage as a result of the procedure.

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If the diagnostic test is negative and I do not have an affected pregnancy how could I have had a screen-positive result?

Most women with Quadruple test screen-positive results will not have an affected pregnancy. Although the screening test offered here discriminates well between women with affected and unaffected pregnancies it cannot do so perfectly.

Your screening result is based on your age, the AFP, uE₃, total hCG and Inhibin values. The four markers naturally vary between women and therefore there is usually no apparent reason for women having either high or low levels and so most women with screen-positive results will not have an affected pregnancy. A screen-positive result only indicates who is in a higher risk group so that we know who should be offered a diagnostic test.

What happens if my baby does have Down's or Edwards syndrome?

If your baby does have one of these abnormalities you will be offered counselling to discuss the implications and your options. If you decide to continue with the pregnancy you could discuss the support that is available with your consultant or midwife. If you decide to have a termination of pregnancy your consultant or midwife can make arrangements for this.



OTHER USEFUL ORGANISATIONS

Down's Syndrome Association

www.downs-syndrome.org.uk 0333 1212 300

info@downs-syndrome.org.uk

Antenatal Results and Choices (ARC)

www.arc-uk.org 0845 077 2290

info@arc-uk.org

Support Organisation For Trisomy 18/13 (SOFT) UK

www.soft.org.uk 0121 351 3122

enquiries@soft.org.uk

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or find us at:

www.wolfson.qmul.ac.uk/service-1/antenatal-screening

The Wolfson Institute of Preventive Medicine has played a leading role in the discovery, development and implementation of antenatal screening methods. It is committed to improving the efficacy and safety of screening. We use information collected as part of our screening programme, including measurements on stored blood samples, to audit our screening programme and ensure that it is meeting our expected quality standards. Such information may also be used to help discover and validate new tests that improve the quality of screening services.