

Non-invasive prenatal testing (NIPT)

Information for women who have previously had a pregnancy or child with Down's syndrome, Edwards' syndrome or Patau's syndrome

It is your choice whether to have antenatal screening tests for chromosomal conditions such as Down's syndrome (also known as trisomy 21 or T21), Edwards' syndrome (also known as trisomy 18 or T18) and Patau's syndrome also known as (trisomy 13 or T13). Screening can give you information that may help you prepare for a baby with a particular condition and make sure that you get the right care during pregnancy, birth and immediately after the birth. As an initial screening test, NIPT is more accurate than the combined or quadruple tests (see Screening tests below). We understand that going through antenatal screening after a previous diagnosis may be a worrying time. There are support organisations available for pregnant women and families. You can find details at phw.nhs.wales/antenatalusefullinks

What tests are available if I have previously had a pregnancy or child with Down's syndrome, Edwards' syndrome or Patau's syndrome?

We understand that you might need different information for your current pregnancy when you have previously had a baby with Down's syndrome, Edwards' syndrome or Patau's syndrome. The information on this page outlines the available options for screening and invasive testing.

You may have been given information about these conditions during previous pregnancies. Or you may have lost a baby with, or have a child or family member living with, Down's syndrome, Edwards' syndrome or Patau's syndrome. It is important that you have accurate information about all three conditions when making choices about screening or invasive tests in pregnancy.

Your midwife or genetic counsellor will support you in whatever you decide.

Down's syndrome

You can find information on Down's syndrome and living with Down's syndrome at:

[Section 2 - What is Down's syndrome and living with Down's syndrome? - Public Health Wales](#)

You can watch some family stories on living with Down's syndrome at www.downs-syndrome.org.uk/about-downs-syndrome/pregnancy-and-baby/new-parent-stories

Edwards' syndrome and Patau's syndrome

You can find information on Edwards' syndrome and Patau's syndrome and living with the conditions at:

[Section 3 – What is Edwards' syndrome and living with Edwards' syndrome? - Public Health Wales](#)

[Section 4 – What is Patau's syndrome and living with Patau's syndrome? - Public Health Wales](#)

You can watch some family stories on living with Edwards' syndrome and Patau's syndrome at www.soft.org.uk/living-with-trisomy

Screening tests

All pregnant women who visit their midwife early in their pregnancy are offered the choice of screening tests to work out the chance of their baby having Down's syndrome, Edwards' syndrome or Patau's syndrome. You can find more information about the screening tests, Down's syndrome, Edwards' syndrome and Patau's syndrome, and other screening tests offered to you in pregnancy, at: [Antenatal Screening Tests Information about the screening tests you will be offered in pregnancy](#)

Your midwife can also give you this information.

As you have had a baby with Down's syndrome, Edwards' syndrome or Patau's syndrome, you will be offered different tests to the combined test or quadruple test. We explain the options available to you in more detail in the next section.

Deciding which tests to have during your pregnancy is a personal choice and your midwife will support you in whatever you choose. You may want to have someone with you for support during the tests and when you have your results.

Your options

1. No testing: If you decide not to have any tests, you will still be offered all other parts of your routine antenatal care. Your midwife will explain what this means for you.

2. Non-invasive prenatal testing (NIPT): NIPT is a screening test that is more accurate than the combined or quadruple test. It can be done from 10 weeks of pregnancy. Your midwife or genetic counsellor will discuss this with you in more detail. There is more information at:

[Section 8 – What is the non-invasive prenatal test \(NIPT\)? - Public Health Wales](#)

If you receive no result from a first NIPT, you will be offered a further NIPT or an invasive test (see below) to give you a result. Around 0.3% (1 in 300) of tests will not give a result, often because there are not enough of the baby's cells in your blood. Your midwife will be able to talk to you about this.

3. Invasive procedures: Invasive testing involves taking a sample from the placenta (known as chorionic villus sampling or CVS) or amniotic fluid (a procedure known as amniocentesis). The chance of miscarriage from either of these tests is around 0.5% (1 in 200). It usually takes two to three days to get a result that can tell you if your baby does or does not have Down's syndrome, Edwards' syndrome or Patau's syndrome.

Your midwife will discuss these procedures with you. There is more information at:

[Section 9 – What is chorionic villus sampling \(CVS\)? - Public Health Wales](#)

[Section 10 – What is amniocentesis? - Public Health Wales](#)

The test that is done on your CVS or amniocentesis sample is called a quantitative fluorescence-polymerase chain reaction (QF-PCR) test. You can find out more about this test at:

[Section 11 – What is a quantitative fluorescence-polymerase chain reaction \(QF-PCR\) test? - Public Health Wales](#)

This is general information, and the testing options may not be suitable for all parents with a previous pregnancy with Down's syndrome, Edwards' syndrome or Patau's syndrome. Always discuss options with your midwife or genetic counsellor.

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