Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Antenatal screening tests

Contains information on cell free fetal DNA (cffDNA) screening for women who are D negative.

GIG CYMRU NHS WALES
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Public Health Wales

This information is available in Welsh, English, large print and Braille. Please ask your midwife.

Mae'r llyfryn hwn ar gael yn Gymraeg, Saesneg, print bras a Braille. Gofynnwch i'ch bydwraig.

Easy read, audio and British Sign Language versions are available on phw.nhs.wales/antenatal-screening

Crystal Mark 23463
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Section 1 – Introduction to antenatal screening tests

Your choices in pregnancy

During your pregnancy, you will be offered a number of different screening tests. Here you will find information about the screening tests, which will help you decide whether to have, or not have, some or all of the tests. You will have the chance to talk about the tests with your midwife.

Your NHS number is unique to you and this number identifies you on NHS computer systems. If you have a copy of your NHS number, please have the number available when you first see the midwife or go to the hospital for your maternity care.

What are screening tests?

Screening tests can help to detect some of the conditions that you or your baby may have. They show what chance there is that you or your baby has the condition.

They do not show for certain whether you or your baby has the condition screened for.

The tests offered include blood tests and ultrasound scans.

Sometimes the blood test or ultrasound scan can give an unclear result and you may be offered further tests.

If the result of a screening test shows you, or your baby, have a higher chance of having a certain condition, you may be offered further screening or an invasive test, for example an amniocentesis. Invasive tests give more certain results, but there is a small chance of them causing a miscarriage. For this reason, these tests are only offered where there is a higher chance of having a condition. You can find information on invasive tests in section 7 of this booklet.

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Screening tests can miss the condition they are screening for. The midwife can tell you how often this can happen with the tests.

What tests are available?

You will be offered the following screening tests.

In early pregnancy, usually before 14 weeks

You will be offered blood tests to detect:

- infections that could harm you and your baby (these are HIV, hepatitis B and syphilis)
- your blood group and Rhesus (Rh) D group, and whether there are any antibodies in your blood, and
- certain blood disorders which are inherited, such as sickle cell and thalassaemia.

All of these tests can be taken at the same time. You can choose which tests are carried out. Your midwife will tell you where the tests can be done.

You will also be offered:

- screening for Down's syndrome, Edwards' syndrome and Patau's syndrome.

Usually at 11 to 14 weeks

An ultrasound scan to see:

- how many weeks pregnant you are
- if your baby's heart is beating, and
- if you are having more than one baby.

At 18 to 20 weeks

You can have a fetal anomaly scan to see the way your baby is developing.

Deciding if you want to have the tests

Screening is your choice. It can be difficult to decide what tests to have.

Some women do not want to know if they have any health conditions that could affect them or their baby. Others want to know if they have any health conditions that could affect them or their baby so that they can make decisions on treatment or, if there are unexpected findings, to prepare for the birth or consider ending the pregnancy.

Take time to think before you decide. You can talk about the tests with your midwife and ask any questions you may have. When you have chosen which tests you want to have, the midwife will make arrangements for you.

Where will the tests be done?

Your midwife will tell you where you can have the tests done.

Results

How will I get the result of my screening tests?

Your midwife will tell you how and when you will get the result of your tests.

Will my results be confidential?

The NHS keeps the results of all tests confidential. Hospital policies vary on how many health-care professionals have access to your test results. Your midwife will be able to explain the local arrangements to you.

Please remember

- You can choose which tests you want to have.
- No test will be done unless you agree.
- If you do not come for a test you may not be sent a reminder.

If you change your mind

You can change your mind about your choices. If you decide to have a test and then change your mind before you have the test, please remember to tell your midwife so they can make a note in your maternity records.

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Section 2 – Your blood group and pregnancy

This section explains the tests that can be done during pregnancy to:

- find out your ABO blood group
- find out your D group (sometimes known as RhD group/Rhesus D) group, and
- look for antibodies.

The people looking after you during pregnancy need to know your blood group (type of blood) in case you ever need a blood transfusion. The screening test for blood group and red cell antibodies is very accurate. Sometimes you will need extra blood tests if antibodies are found.

The test

This test can be done with other blood tests, usually early in pregnancy. It is offered to you again around the 28th week of pregnancy.

What your blood group test result will tell you Your blood group will be one of the following four main groups.

- Group O
- Group A
- Group B
- Group AB

Within your blood group, you will either be D positive or D negative. For example, your overall blood group might be written as 'O RhD positive'.

About your blood

Your blood is made up of:

- red blood cells
- white blood cells, and
- platelets

These are found in a liquid called plasma. Your blood group is identified by antigens and antibodies contained in your blood.

- Antigens are protein molecules found on the surface of the red blood cells.
- Antibodies are proteins found in the liquid part of the blood called plasma. Antibodies are part of your body's natural defence.

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Red blood cells sometimes have another antigen known as D antigen.

- If this is present, your blood group is known as D positive.
- If it is not present in your blood, your blood group is D negative.
- Most people, 85% (85 out of 100) will be D positive.

What are red cell antibodies?

Red cell antibodies are part of your body's natural defence and fight against anything the body thinks is foreign. You may form antibodies if blood cells with a different blood group to yours, enter your blood stream. This can happen because of a blood transfusion or from your baby during pregnancy.

Red cell antibodies can pass from your bloodstream into your baby's blood, where anti-D antibodies may develop. Sometimes this can cause a rare condition called haemolytic disease of the fetus and newborn (HDFN), making your baby unwell. Babies with this rare condition usually need to be admitted to hospital.

What if you are D negative?

- If your blood group is D negative and your baby's blood group is positive you may develop anti-D antibodies (If you are D positive, this problem does not usually happen).
- If you are D negative, you will be offered a blood test at around the 16th week of pregnancy. This test is called cell free fetal DNA (cffDNA) and will tell you if your baby is D negative or D positive.

What is the cell free fetal DNA (cffDNA) test?

If you are D negative, the cell free fetal DNA test can predict if:

- your baby is D negative (the same blood group as you), or
- D positive.

This test will look at small amounts of your baby's DNA in your blood. DNA is the genetic information inside the body's cells. If you are D negative and your baby is D positive you will be offered anti-D immunoglobulin injections. This is referred to as anti-D in this information. This will greatly reduce HDFN developing.

What are anti-D injections?

Anti-D will help to stop antibodies being made if any of your baby's blood may have entered your blood stream, and so will reduce the chance of haemolytic disease of the newborn. Anti-D injections are made from plasma, which is the fluid part of blood that carries oxygen and blood cells around your body. The plasma used to make anti-D is collected from blood donors. You may need to be offered anti-D injections in future pregnancies.

Is anti-D safe?

Yes. Anti-D injections can cause some mild pain when they are injected into the muscle. Occasionally anti-D injections can cause allergic reactions. How it is produced is strictly controlled, so the risk of a known virus being passed on to you from a donor is very low.

What are the advantages of having screening for my blood group and antibodies?

If you have this test, you will know your blood group and whether you are D positive or D negative.

It is less common to be D negative. If you are D negative, you will be offered a cell free fetal DNA test. If your baby is D negative, you would not be offered anti-D injections.

The test will also look for antibodies. It is important to know about these so that if you ever need a blood transfusion this could be given safely. Rarely, antibodies present in your blood carry a risk to your baby. If this happens you and your baby can be given specialist care.

What are the disadvantages of having screening for my blood group and antibodies?

Screening is a simple blood test. The only risk would be the same as having any blood test.

If you know that you are D negative

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Contact your midwife or your hospital doctor (obstetrician) as soon as possible to remind them that you are D negative if you:

- have vaginal bleeding after 12 weeks of pregnancy
- have a miscarriage after 12 weeks of pregnancy, or
- suffer an injury to your abdomen (for example, an injury from a seat belt in a car accident or by falling over).

If any of these things happen, your body may start to produce antibodies and you may need an anti-D injection.

Other reasons to be offered an anti-D injection

You should be offered an anti-D injection to reduce the risk of you producing antibodies. This includes if:

- you have chorionic villus sampling (CVS) or an amniocentesis (see section 7 for information on invasive tests)
- you have a miscarriage
- you have an ectopic pregnancy
- your baby's blood group is not known, or
- after your baby is delivered they are found to be D positive.

Section 3 – Infections in pregnancy

Protecting your baby

This section explains about some of the infections that can cause complications for you or your baby, but which can be treated.

These are:

- human immunodeficiency virus (HIV)
- hepatitis B, and
- syphilis.

There are screening tests for all these infections and your midwife will offer you these.

Why are the tests recommended?

If you have one of these infections and it is not treated, you could pass the infection to your baby during your pregnancy, the birth or after the birth.

Screening for HIV, hepatitis B and syphilis is offered and recommended in every pregnancy.

All of these infections can be serious and if not treated they may cause complications for you or your baby. Most people with these infections will not feel ill and will not know they have these infections. If you have one of these infections, treatment will significantly reduce the chance of you passing it to your baby.

The test

The test is a blood test which can be done with other blood tests, usually early in pregnancy.

How are the tests done?

The screening tests for HIV, hepatitis B and syphilis can all be carried out on one blood sample. You can choose which tests are carried out. Only a small amount of blood is needed.

Why should I have the test?

By accepting screening for HIV, hepatitis B and syphilis you are deciding to find out if you have the infection so that everything possible can be done to protect your baby.

What if I decide not to be screened for HIV, hepatitis B or syphilis?

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If you decide not to be screened for HIV, hepatitis B or syphilis your midwife will ask you the reasons why to make sure you have understood the reasons for the test. Later on in your pregnancy, usually before 21 weeks, your midwife will ask you if you want to discuss the screening and will offer you the test again.

> You can ask to be screened for HIV, hepatitis B or syphilis at any time in your pregnancy.

If, while you are pregnant, you are worried that you might have caught HIV, hepatitis B, syphilis or other diseases which can be passed between people, you can ask your midwife to do another test at any time during your pregnancy. You can also get confidential testing from your nearest sexual-health clinic.

If you need further information you can speak to your nearest NHS sexual-health clinic – phone your local hospital and ask for the sexual health clinic.

Human immunodeficiency virus (HIV)

What is HIV?

HIV is a virus that attacks the immune system. It is the virus that can lead to acquired immune deficiency syndrome (AIDS). A person living with HIV can look and feel well for many years. They may not know they have the infection unless they have a blood test. However, HIV can be passed on to their baby during pregnancy, childbirth or by breastfeeding.

How can HIV be caught?

HIV can be caught through:

- a mother who has the infection passing it to her baby during pregnancy, the delivery or breastfeeding
- having unprotected sexual activity (without a condom) with someone who has HIV
- a blood transfusion or blood products containing HIV (these are tested in the UK but not in all other countries)
- sharing infected needles and injecting equipment, and
- contact with unclean needles used in body piercing and tattooing.

What are the advantages of having screening for HIV in pregnancy?

25% (1 in 4) of babies born to mothers who have an untreated HIV infection, or who do not know that they have an infection, will become infected with HIV.

If you are found to have HIV, a number of things can be done to lower the chance of you passing it to your baby. You will be offered specialist treatment and care. The treatment may also help to keep you in better health.

If you are found to have HIV and accept the treatment offered, the risk to your baby of having HIV will be reduced to 0.3% or less (1 in 300 or less).

What are the disadvantages of having screening for HIV in pregnancy?

There is no good time to find out that you are HIV positive. However, if you find out when you are pregnant, you can have treatment to help stop your passing the infection to your baby.

Should I have the screening test for HIV?

Only you can decide whether to have the test or not. All hospitals in Wales recommend the screening test for HIV because if you are HIV positive it is possible to reduce the chance of your baby getting the infection.

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What will the screening test result tell me?

A negative screening result tells you that you are very unlikely to have HIV infection.

If you have caught HIV in the few weeks before the blood sample is taken, your body may not have started producing antibodies and the test will not be able to detect the infection.

It is important to remember that you can get HIV when you are pregnant. If you change your sexual partner during the pregnancy, you should use a condom.

What is the diagnostic test for HIV?

Sometimes the HIV blood test can give an unclear (reactive) result and further tests will be needed to confirm you do not have the infection.

If the screening test shows that you are HIV positive, you will have another blood test to confirm the infection and guide treatment options.

What if I have HIV?

If the test shows you have HIV, you will be able to plan with your midwife or hospital doctor what happens next. You will be offered specialist medical care and treatment to help with the infection. This will help reduce the risk of your baby getting the infection.

Treatment will include drug therapy.

You can get more information about HIV from:
111.wales.nhs.uk/Encyclopaedia/h/article/hivandaids

Hepatitis B

What is hepatitis B?

Hepatitis B is a virus that infects the liver. Many people who have hepatitis B do not know they have it. Most adults with hepatitis B make a full recovery, but a small number become 'carriers' of hepatitis B. People who are carriers may develop serious liver disease.

If a pregnant woman has hepatitis B, her baby can be exposed to the infection during the delivery. A baby who gets the infection may have it for life and may be at risk of liver disease.

How can hepatitis B be caught?

Hepatitis B can be caught through:

- a mother who has hepatitis B passing it to her baby during delivery
- having unprotected sexual activity (without a condom) with someone who has the infection
- having contact with the body fluids of someone who has the infection
- having contact with unclean needles used in body piercing and tattooing
- sharing infected needles and injecting equipment
- a blood transfusion or blood products containing hepatitis B (these are tested in the UK but not in all other countries), and
- living for a long time in close contact with someone who has the infection.

What are the advantages of having screening for hepatitis B in pregnancy?

Testing for hepatitis B is important because if doctors know about the infection before a baby is born, a course of vaccinations started soon after the birth can help stop your baby getting the infection. The vaccinations protect most babies from developing hepatitis B. If you have hepatitis B, there is up to a 90% (9 in 10) chance that your baby will get it.

If you are found to have hepatitis B infection your baby can be vaccinated, and the chance of them having the infection will be less than 5% (five in 100).

What are the disadvantages of having screening for hepatitis B in pregnancy?

There is no good time to find out that you have hepatitis B. However, if you find out when you are pregnant, your baby can be vaccinated to help prevent them getting the infection from you.

Should I have the screening test for hepatitis B?

Only you can decide to have the test or not. All hospitals in Wales recommend the screening test for hepatitis B because if you have hepatitis B your baby can be vaccinated to help stop them catching the virus from you.

What will the screening test result tell me?

A negative screening result tells you that you are very unlikely to have hepatitis B infection.

If you have caught hepatitis B in the few months before the blood sample is taken, the test will not be able to detect the infection.

It is important to remember that you get hepatitis B when you are pregnant. If you change your sexual partner during the pregnancy, you should use a condom.

What is the diagnostic test for hepatitis B?

Sometimes the hepatitis B blood test can give an unclear (reactive) result and further tests will be needed to confirm you do not have the infection.

If the screening test shows that you have hepatitis B, you will need another blood test to confirm the infection.

What if I have hepatitis B?

If you have hepatitis B, your midwife or doctor will talk to you about how it will affect you and to plan the vaccinations your baby will need.

Vaccinations should take place:

- within 24 hours of birth (some babies will also need an injection of antibodies – hepatitis B immunoglobulin) at their first vaccination
- at four weeks of age
- at eight, 12 and 16 weeks of age (as part of normal baby vaccinations), and
- at one year of age.

Your baby will also need a blood test at 12 to 13 months to make sure they have not got the infection.

You may also be worried that other people in your family have the infection. They can also be tested and vaccinated if necessary.

You can get more information about hepatitis B from:
111.wales.nhs.uk/Encyclopaedia/h/article/hepatitisb

Syphilis

What is syphilis?

Syphilis is a serious bacterial infection. Most people who have syphilis are unwell for only a short time at first and they may not be aware they have it. But if syphilis is not treated, it can cause brain damage and heart conditions later in life.

How can syphilis be caught?

Syphilis can be caught through:

- a woman who has syphilis passing the infection to her unborn baby during pregnancy, or
- having unprotected sexual activity (without a condom) with someone who has the infection.

What are the advantages of having screening for syphilis in pregnancy?

A syphilis infection will be treated with antibiotics in early pregnancy and this will usually prevent your baby from getting syphilis. Occasionally, babies may also need antibiotics when they are born.

What are the disadvantages of having screening for syphilis in pregnancy?

There is no good time to find out that you have syphilis. However, if you find out when you are pregnant, you can have treatment to help prevent passing the infection to your baby.

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If you are found to have a syphilis infection and it is not treated, there is a risk that the infection can lead to a miscarriage or harm your baby.

Should I have the screening test for syphilis?

Only you can decide to have the test or not. All hospitals in Wales recommend the screening test for syphilis because, if you have syphilis, treatment with antibiotics can help prevent you passing it to your baby.

What will the screening test result tell me?

A negative screening result tells you that you are very unlikely to have syphilis infection.

If you have caught syphilis in the few weeks before the blood sample is taken, your body may not have started producing antibodies and the test will not be able to detect an infection.

It is important to remember that you can get syphilis when you are pregnant. If you change your sexual partner during the pregnancy, you should use a condom.

What is the diagnostic test for syphilis?

Sometimes the syphilis blood test can give an unclear (reactive) result and further tests will be needed to confirm you do not have an infection.

If the screening test is positive, you will be given an appointment with a doctor specialising in these types of infections. This doctor will ask you questions, including questions about previous infections, to make a diagnosis and decide on the best treatment.

The results of the screening test for syphilis are not always easy to understand. Sometimes the result of the screening test will come back positive because you have had syphilis in the past and have been treated, or you have a different and less serious infection.

What if I have syphilis?

If you have syphilis, your midwife or hospital doctor will talk to you about how it will affect you. You will probably be given antibiotics and need more blood tests.

You can get more information about syphilis from:
111.wales.nhs.uk/Encyclopaedia/s/article/syphilis

Other infections

If you get a rash or come into contact with someone who has a rash when you are pregnant, you need to tell your midwife or doctor. You may need to have other blood tests to find out what has caused the rash.

There are a few infections that cause a rash that are important to know about in pregnancy. These are German measles (rubella), chickenpox, measles and parvovirus infection (commonly known as slapped cheek).

Rubella

You will be protected from rubella if you have ever had two doses of a vaccine containing rubella. You will need two doses of the vaccine if you haven't had or cannot remember having had the vaccine.

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You will need these after your baby is born. You will be given the first vaccine usually at your doctor's surgery, and the second dose a month later.

If you are not sure what vaccinations you have had, you should ask your GP surgery to check your immunisation history, which may be recorded in your GP records.

Having rubella during pregnancy is extremely rare in Wales but it can be very serious for your baby. It can cause a condition called congenital rubella syndrome (CRS). This can lead to deafness, blindness, cataracts (eye conditions) or even heart conditions in your baby. It can also, very rarely, result in the death of your baby.

Chickenpox

Chickenpox is a very common infection and most women will have had chickenpox as a child and will be immune. If you come into contact with someone with chickenpox or have chickenpox while you are pregnant, you may be offered treatment. This will help to prevent or reduce the symptoms of the infection.

Measles

You will be protected from measles if you have previously had the infection or if you have ever had two doses of a vaccine containing measles (for example, measles-rubella or measles-mumps-rubella in school, as a child, or at your GP's surgery). If you come into contact with someone with measles while you are pregnant or if you have measles in pregnancy, you may be offered treatment. This will help to prevent or reduce the symptoms of the infection.

Measles is rare in the UK. If you have measles in pregnancy your symptoms can be more severe.

Parvovirus

Parvovirus is usually a very mild infection in women but can occasionally cause complications in unborn babies. There is no vaccine to prevent this infection. In rare cases when women get parvovirus in early pregnancy their unborn baby may have complications. If you have parvovirus in early pregnancy you will be offered some extra scans to look for signs of these complications in your baby.

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Section 4 – Screening for sickle cell and thalassaemia in pregnancy

This section explains the tests you can have during pregnancy to find out if you are a carrier of sickle cell or thalassaemia. If you are a carrier, the biological father of your baby will also be offered testing.

What are sickle cell disorders and beta thalassaemia major?

Sickle cell disorders and beta thalassaemia major are serious inherited blood conditions. They affect the haemoglobin in the red blood cells. Haemoglobin is important because it carries oxygen around the body. People who have these conditions will need specialist care throughout their lives. There are also other, less common haemoglobin disorders. Many of these are not as serious.

If you and the biological father of your baby are both carriers of sickle cell or thalassaemia, your baby could inherit a sickle cell disorder or thalassaemia major.

Sickle cell disorders

People with a sickle cell disorder can:

- have tissue and organ damage and varying degrees of symptoms
- have attacks of severe pain where they need to stay in hospital, and
- be more prone to serious infections.

Beta thalassaemia major

People with beta thalassaemia major have:

- severe anaemia and need blood transfusions every four to six weeks as well as other treatments.

How are the disorders inherited?

Sickle cell and thalassaemia are genetic disorders. They are passed on in families. If only one biological parent (either the mother or father) has the sickle cell or thalassaemia gene, it is very unlikely that their baby will have a sickle cell disorder or thalassaemia major. But their baby may be a carrier. This means that, like the mother or father, the baby will have the sickle cell or thalassaemia gene, but the gene does not usually cause problems.

If both biological parents carry a sickle cell or thalassaemia gene, the baby may have a 25% (one in four) chance of having a sickle cell disorder or beta thalassaemia major.

Who can be a carrier of sickle cell or thalassaemia?

Anyone can be a carrier of sickle cell or thalassaemia. The chances of being a carrier of sickle cell or thalassaemia are higher for certain groups of people.

You are more likely to be a carrier if your family, no matter how many generations back, come from the Mediterranean, Africa, the Caribbean, the Middle East, South Asia, South America or South East Asia.

The test

The test is a blood test which can be done with other blood tests, usually early in pregnancy. The test is only offered to women with a higher chance of carrying sickle cell or thalassaemia. See 'Who is offered the test?' to see if you are in this group. As part of your antenatal care, you will be offered a routine blood test (a full blood count) to check your haemoglobin level to see if you are anaemic.

The full blood count can also find some types of thalassaemia.

The midwife will ask you if you would like to have thalassaemia screening as part of the full blood count test. If your full blood count test suggests that you might carry thalassaemia, the laboratory may also screen your blood for sickle cell disorders and thalassaemia.

Who is offered the test?

Your midwife will ask you about your family origins. You should be offered the test if:

- you or the biological father of your baby has a family history of sickle cell or thalassaemia
- you, the biological father of your baby, anyone in the biological father's family or your family, no matter how many generations back, came from anywhere in the world apart from the UK or Ireland, or
- you or the biological father of your baby does not know your family history – for example, you or the biological father of your baby were adopted.

What are the advantages of having screening for sickle cell and thalassaemia in pregnancy?

If you are a carrier of sickle cell or thalassaemia, it is important to know so you can have the right kind of care during your pregnancy.

Women who know their baby has a high chance of inheriting a sickle cell disorder or thalassaemia major can have an invasive test to find out if their baby has the condition. This could be done by either chorionic villus sampling (CVS) or amniocentesis. If your baby has the condition, you can decide whether to prepare for the birth of your baby with one of these conditions or to end your pregnancy.

What are the disadvantages of having screening for sickle cell and thalassaemia in pregnancy?

Having the test may make you anxious if you find out you carry sickle cell or thalassaemia. Some women would be offered an invasive test to see if their baby has the condition. Because the invasive tests have a small chance of causing a miscarriage, many women find this a difficult decision. Some women may wish they had not had the screening test because making this decision is difficult.

Should I have the blood test for sickle cell and thalassaemia?

Only you can decide to have the test or not. Some women want to find out if their baby has sickle cell or thalassaemia, and some do not. Having the test may cause anxiety as the result may mean that you are offered further tests.

What will the results tell me?

If the result shows you are not a carrier, it is very unlikely your baby could have a sickle cell disorder or thalassaemia major. Although the test is very accurate, a small number of results may be unclear. If this happens, you will be offered another test.

If the test shows you are a carrier or a possible carrier, you will be able to talk to a specialist midwife or genetic specialist and they will give you more information. They may suggest you ask the biological father of your baby to have a blood test to find out if he is a carrier. If his test result shows he is not a carrier, it is very unlikely your baby will have a sickle cell disorder or thalassaemia major.

If you are a carrier or have a sickle cell or thalassaemia disorder the biological father of your baby will be offered testing too.

If the biological father of your baby is a carrier or has sickle cell disorder or declines testing you will be offered specialist advice on the condition and invasive testing (see section 7).

What if the biological father is also a carrier?

If the test shows the biological father of your baby is a carrier, there may be a 25% (one in four) chance your baby could have a sickle cell disorder or thalassaemia major. You can then decide whether to have more tests to find out if your baby has the condition. These tests are called invasive tests (see section 7). If you choose not to have more tests, your baby can be tested at birth for sickle cell disorders or for thalassaemia major. This means that if your baby has the condition, treatment can start early.

What are the possible results from invasive tests?

If you have CVS or amniocentesis, the result may show:

- that your baby does not have this condition, or
- your baby has a sickle cell disorder or thalassaemia major. You can then decide whether to prepare for the birth of a baby with sickle cell or thalassaemia major or to end your pregnancy.

CVS and amniocentesis can detect other chromosome changes. The specialist midwife or genetic counsellor will give you more information.

You will be offered newborn bloodspot screening after your baby is born. This test will also look for sickle cell disorder.

You can get more information about sickle cell and thalassaemia from:

111.wales.nhs.uk/Encyclopaedia/s/article/sicklecelldisease

111.wales.nhs.uk/Encyclopaedia/t/article/thalassaemia

Section 5 – Ultrasound scans in pregnancy

This section explains the two ultrasound screening tests you will be offered during pregnancy. You can choose whether or not to have these tests.

It explains:

- what the tests are
- why they are carried out, and
- when they are done.

The scan is a way of checking that your baby appears to be developing as expected. This means it may show up unexpected findings that would then need to be checked by other tests. A scan cannot find all conditions screened for.

What is an ultrasound scan?

An ultrasound machine uses sound waves to create an image on a computer screen.

The person who does the scan is called a sonographer. The sonographer will explain to you what they are doing and what they are looking for.

How is the scan arranged?

If you decide you would like to have a scan, your midwife will tell you where it can be done and will arrange the appointment for you.

If you have been given an appointment and then decide you do not want a scan, please tell your midwife and cancel the appointment.

How is the scan done?

You will be asked to lie on your back to have the scan. You do not need to wear any specific clothing for the scans. However, you will be asked to raise your upper clothes to your chest and lower your skirt or trousers to your bikini line. Gel is spread on your lower abdomen so that a device called a transducer can be passed backwards and forwards over your abdomen. The sonographer may need to press on your abdomen with the transducer to see your baby properly.

Ultrasound waves do not pass through air, so the gel makes sure there is good contact between your skin and the transducer. Your clothing will be protected from the gel with tissue paper.

The transducer passes sound waves through your abdomen into the womb. The sound waves bounce back off your baby and are translated into an image on a screen.

The sonographer will get a clearer image of your baby if your bladder is not completely empty when you have your scan. Please try not to pass urine for about an hour before your appointment.

For the sonographer to see your baby clearly on the screen, the scan is carried out in a dimly lit room. Scanning involves a lot of concentration, so the room is also kept quiet. The sonographer will explain to you what they are doing and what they are looking for.

Are scans safe?

As far as we know, the early pregnancy dating scan and the fetal anomaly scan we offer are safe for mother and baby.

What are early pregnancy dating and fetal anomaly scans?

Both of these scans can detect unexpected findings before your baby is born. Finding this out before the birth can help you prepare. Sometimes it can help plan treatment for after your baby is born. Sometimes when women find out that there are unexpected findings, they may want to consider ending the pregnancy.

What are the disadvantages of having these scans?

Having the scans may make you anxious, especially if there is an unexpected finding. If you prefer not to know, you need to think carefully whether you should have the scans. You should discuss your concerns with your midwife.

Results

How do I get the results of these scans?

The sonographer will tell you the results of your scan at the end of the examination.

Can I bring family or friends with me when I have the scan?

Most babies are healthy, but because the scans can show unexpected findings you may want to ask your partner or one adult who can support you to come with you to your scan.

It is best not to bring children to the appointment. They can distract you and the sonographer during the scan. If unexpected findings are found on the scan, the sonographer will tell you about them, and this is not a suitable situation for children.

Can I have a picture of my baby?

It is sometimes possible to buy pictures of your baby taken during the scan. Please tell the sonographer if you would like to do this **before** the scan starts. The sonographer will obtain the best picture of your baby within the appointment time.

Video recording or using mobile phones in the ultrasound scan room is not recommended. You will need to check your local hospital's policy on this.

The early pregnancy dating scan

This scan is offered to all women, usually at 11 to 14 weeks of pregnancy.

How long will my scan take?

The early pregnancy dating scan takes about 10 to 20 minutes.

The scan is done to:

- look for your baby's heartbeat
- find out if you are carrying more than one baby (if this is the case you will need extra antenatal care, and it is important to know if your babies are sharing the same placenta)
- measure your baby to check how pregnant you are and the date your baby is due (this is especially important if you are thinking of having more screening tests)
- measure the nuchal translucency (the small collection of fluid at the back of your baby's neck), if you have asked for Down's syndrome, Edwards' syndrome and Patau's syndrome screening, and
- check your baby's development (your baby's development is not very clear at this early stage, but sometimes serious conditions can be detected).

Will I need another early pregnancy dating scan?

Sometimes your baby cannot be seen clearly using an abdominal transducer, so the sonographer may suggest you have an internal scan. This is called a transvaginal scan, and it can give a more detailed picture. You will be asked to empty your bladder before this scan. A small transducer is inserted into your vagina, similar to having a tampon inserted.

A transvaginal ultrasound scan is not usually painful. The sonographer will explain about the scan and ask for your agreement to do it. If you do not want to have an internal scan, please tell the sonographer. You may need an appointment for another abdominal scan.

Fetal anomaly scan

This scan is offered at 18 to 20 weeks of pregnancy.

How long will my scan take?

The fetal anomaly scan usually takes about 15 to 30 minutes.

The scan is done to:

- check your baby's physical development
- help detect structural changes (also called anomalies) such as spina bifida, kidney conditions, or heart conditions
- check the amount of fluid around your baby in the womb, and
- look at the position of the placenta.

Looking for the sex of your baby is not part of the scan and is not 100% accurate.

If you want to know the sex of the baby and the sonographer can see it, they will tell you at the time of the scan. They will not write it down.

Will I need another fetal anomaly scan?

The sonographer will use an all-Wales agreed checklist to look for certain conditions (such as spina bifida) and at structures (such as the heart). The sonographer has to concentrate very hard during the scan, so please make sure you and the person supporting you do not distract them. Sometimes it is not possible to see everything on the list during your scan. This can be because:

- your baby was lying in a position which made the examination difficult, or
- you are above average weight for your height and this made looking at your baby difficult as the images were not clear.

If this happens, you will be given another appointment to come back for one more scan to see if the sonographer can complete the checklist. It is not always possible for the sonographer to complete the list, even on the second appointment.

What is the scan able to find?

A scan can show some unexpected findings with your baby's development but not all. Some may develop after 20 weeks and some may not show up on the scan. This is why, in a small number of cases, babies are born with undiagnosed conditions.

Table 1 has a list of some examples of conditions. The right-hand column shows how likely it is that a fetal anomaly scan could identify each condition. **This list does not include all conditions that may be seen.**

Some conditions may be caused by your baby having a chromosome change which affects the way your baby develops. If a chromosome change is suspected, you may be offered an amniocentesis. You can find information on this test in section 7.

Table 1

The condition	The chance of the condition being seen on a fetal anomaly scan at 18 to 20 weeks
Spina bifida (skin or bone not covering the spinal cord) Spina bifida is a fault in the development of the spine and spinal cord which leaves a gap in the spine. The spinal cord connects all parts of the body to the brain.	**98% (around 9 in 10)**
Major heart condition, for example, coarctation of the aorta Coarctation of the aorta is a serious heart condition where the main artery (the aorta) has not developed in the normal way in the womb. This condition often requires surgery in the first weeks of life (or in some cases may only need monitoring in clinic by a specialist heart doctor).	**58% (around 5 in 10)**

Data from: Welsh Congenital Anomaly Register and Information Service (CARIS)
phw.nhs.wales/services-and-teams/caris (Accessed 23/02/24)

What will happen if an unexpected finding is suspected during the scan?

If the sonographer detects an unexpected finding, they will tell you about it and you will be able to talk to the midwives or hospital doctor (obstetrician) in your antenatal clinic.

Receiving unexpected news can be distressing. We recommend that your partner or one adult only comes with you to the scan appointment.

Sometimes it is not possible, at the first fetal anomaly scan, for the sonographer to tell definitely what the unexpected finding is. You might be offered another scan in a different department or with a specialist dealing with the type of condition your baby is suspected of having.

Invasive tests following a fetal anomaly scan

You might also be offered another test, such as amniocentesis (see section 7). You will be given more information on any other tests by your midwife or your hospital doctor (obstetrician).

What will happen if a definite anomaly is found?

Finding out that your baby may have a condition before the birth can help you prepare. Information about the type of condition can be used to prepare for how, when and where your baby is delivered. Your baby may need to be born in a different hospital that can provide the specialised staff and care that your baby may need.

A very small number of conditions can be treated before your baby is born.

If the condition is serious, you may decide to continue with your pregnancy or consider ending your pregnancy. These are difficult decisions and you will be given time, information and support to help you make a decision that is right for you.

Finding out that your unborn baby has a condition is distressing and deciding what to do is hard. Most women want and need some support. This might come from your partner, family or friends or from the health professionals who are caring for you. You can choose to bring your partner or one adult only to hospital appointments with you.

Section 6 – Screening for Down's syndrome, Edwards' syndrome and Patau's syndrome in pregnancy

This section explains the tests that can be done during pregnancy to find out if your baby has Down's syndrome (trisomy 21 or T21), Edwards' syndrome (trisomy 18 or T18) or Patau's syndrome (trisomy 13 or T13. Women who find out that their baby has a higher chance of these conditions will be offered a further screening test or an invasive procedure, such as chorionic villus sampling (CVS) or amniocentesis (see section 7).

You can choose whether or not to have these tests. Some women want to find out if their baby has one of the conditions and some do not.

What is Down's syndrome (T21)?

Down's syndrome is caused by an extra copy of chromosome 21 in all or some cells of the body.

People with Down's syndrome can have a good quality of life and most say they enjoy their lives. With support, many people with Down's syndrome are able to get jobs, have relationships and live semi-independently in adulthood.

A person with Down's syndrome will have some level of learning disability. This means they will find it harder than most people to understand and to learn new things. They may have communication challenges and difficulty managing some everyday tasks. Most children with Down's syndrome attend mainstream schools but need additional support.

People with Down's syndrome are more likely to have some medical conditions, including heart conditions. Many of these conditions can be treated, with good outcomes.

In Wales, 90% (9 in 10) children with Down's syndrome live past their fifth birthday. For babies without serious health conditions, survival rates are similar to that of other children. With good health care, most people will live into their 60s.

All women have a chance of having a baby with Down's syndrome. The chance increases with age but babies are also born with Down's syndrome to younger women. This is why women of all ages are offered the screening test.

Down's syndrome happens in 1 in 415 pregnancies in Wales.

You can view some family stories about living with Down's syndrome on the Antenatal Screening Wales website: phw.nhs.wales/antenatal-screening

You can get more information about Down's syndrome from: 111.wales.nhs.uk/encyclopaedia/d/article/downssyndrome

What are Edwards' syndrome (T18) and Patau's syndrome (T13)?

Babies with Edwards' syndrome have an extra copy of chromosome 18 in all or some cells. Babies with Patau's syndrome have an extra copy of chromosome 13 in all or some cells.

Sadly, the survival rates are low and, of those babies born alive in Wales, only around 13% (1 in 7) live past their first birthday.

Some babies may survive to adulthood but this is rare.

All babies born with Edwards' syndrome and Patau's syndrome will have a learning disability and a wide range of physical challenges, which can be extremely serious. They may have conditions affecting their heart, limbs, kidneys and digestive system.

Around half of babies with Patau's syndrome will have a cleft lip and palate. Babies with Edwards' syndrome and Patau's syndrome will have a low birthweight.

Despite their difficulties, children can slowly make progress in their development. Older children with either condition would need to attend a specialist school.

All women have a chance of having a baby with Edwards' syndrome or Patau's syndrome. The chance increases with age but babies are also born with these conditions to younger women. This is why women of all ages are offered the screening test.

Edwards' syndrome happens in about 1 in 1656 pregnancies in Wales and Patau's syndrome in 1 in 4201.

You can view family stories about living with Edwards' syndrome on the Antenatal Screening Wales website: phw.nhs.wales/antenatal-screening

You can get more information about Edward's syndrome and Patau's syndrome from:

111.wales.nhs.uk/Encyclopaedia/e/article/edwardssyndrome(trisomy18)

111.wales.nhs.uk/Encyclopaedia/p/article/pataussyndrome

What screening test will I be offered?

Combined screening – this test is taken between 11 and 14 weeks of pregnancy

You will be offered an ultrasound scan (if possible, the sonographer will measure the small collection of fluid (nuchal translucency) at the back of the baby's neck). You will then be offered a blood test. The measurements taken at the scan, the results of the blood test and your age are used to work out the chance of your baby having Down's syndrome and the chance of your baby having Edwards' syndrome or Patau's syndrome.

Sometimes the measurements can be difficult to get. For example, the baby may be lying in the wrong position or you may be above average weight for your height and this makes looking at the baby difficult because the images are not clear. If the person performing the scan (the sonographer) cannot get a measurement, they will tell you.

If you are having twins, you will be offered the combined screening test.

The quadruple screening test is not offered in Wales in twin pregnancies as it is not as accurate as in single pregnancies.

Quadruple screening – this test is taken between 15 and 18 weeks of pregnancy

You will be offered a quadruple test if the measurements that are needed cannot be taken or if you go for the scan appointment later than 14 weeks pregnant. This involves having a blood test to find out your chance of having a baby with Down's syndrome but not Edwards' syndrome or Patau's syndrome.

You can choose whether or not to have an early pregnancy dating scan. You can also choose to have an early pregnancy dating scan but not to have the screening test for Down's syndrome, Edwards' syndrome and Patau's syndrome.

What will the screening test result tell me?

The screening test can tell you what chance you have of your baby having one of the conditions.

The combined test

- This is taken on or before you are 14 weeks and 1 day pregnant.
- If you have the combined test you will get two different results:
    - One will tell you your chance of your baby having Down's syndrome.
    - The other will tell you your chance of your baby having Edwards' syndrome or Patau's syndrome.

The quadruple test

- This is taken at around 16 weeks of pregnancy (but can be taken between 15 weeks and 0 days and 18 weeks and 0 days pregnant) and will only tell you your chance of having a baby with Down's syndrome.

The screening test does not identify all babies with these conditions. For example, on average, 70% to 80% (around seven or eight in 10) of babies with these conditions will be identified by screening. This means 20% to 30% (around two or three in 10) of babies with these conditions will not be identified by the screening test.

What are the advantages of having screening for these conditions?

If your baby has one of these conditions, you will be able to make choices about your pregnancy. For example, you can decide whether to prepare for the birth of a baby with one of these conditions or to end your pregnancy.

What are the disadvantages of having screening for these conditions?

Having the test may make you anxious, especially if you have a result which shows you have a higher chance of having a baby with one of these conditions. 'Higher chance' is how we describe your result if it is between 1 in 2 and 1 in 150. If the result is between 1 in 2 and 1 in 150, you will be offered either:

- no further testing
- another screening test called non-invasive prenatal testing (NIPT), which is more accurate than the combined or quadruple test, or

- an invasive test to see if your baby definitely has one of these conditions.

Because the invasive test, called amniocentesis or chorionic villus sampling (CVS), has a small chance of causing a miscarriage, many women find this a difficult decision.

Should I have the screening test for Down's syndrome (T21), Edwards' syndrome (T18) and Patau's syndrome (T13)?

Only you can decide whether to have the screening test or not. Some women want to find out if their baby has these conditions, and some don't. All hospitals in Wales offer women a screening test but the decision whether to have the test or not is yours. You can discuss with your midwife what you want to do. Your decision will be respected and health-care professionals will support you whatever you decide.

The scans you have in pregnancy can occasionally see things that may identify that your baby could have one of these conditions and you may be offered an invasive test at this stage.

Results

The results are given as either 'higher chance' or 'lower chance'.

What happens if I have a lower chance result?

If your test result shows you have a lower chance of having a baby with Down's syndrome, Edwards' syndrome or Patau's syndrome, no more tests are offered. Please remember that having a lower chance does not mean that you have no chance of having a baby with one of these conditions.

What happens if I have a higher chance result?

If your test result shows you have a higher chance of having a baby or babies with one of the conditions screened for (that is, a chance of 1 in 2 to 1 in 150), you will be offered an appointment with a midwife or hospital doctor (obstetrician). They will explain your test result to you in detail, including your individual chance, and you can discuss whether or not you want to have a more accurate screening test called a non-invasive prenatal test (NIPT), or an invasive procedure (CVS or amniocentesis), which will give you a definite result but has a small risk of miscarriage. You may face some difficult decisions after an invasive procedure that you need to be aware of beforehand.

Remember that the lower the number, the higher the chance. So, for example, 1 in 80 is a higher chance of your baby having Down's syndrome than 1 in 140.

Between 2% and 4% (between two and four in 100) of women who have the combined or quadruple screening tests have a result which shows they have a higher chance of having a baby with one of these conditions.

Non-invasive prenatal test (NIPT)

NIPT is a further screening test and will not give a definite result. However, it is more accurate than the combined or quadruple test. It is a blood test taken from you in the usual way – your blood will have some of your baby's DNA (genetic material from the placenta) in it. The laboratory screens this blood sample to tell you if there is a 'high' or 'low' chance that your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome.

If you choose this test it usually takes about two weeks to get the result. In about 0.3% (1 in 300) of pregnancies the NIPT test will not be able to give a result and you will be offered an invasive test or you may decide to have no further tests.

If the NIPT result is high chance you will be offered an invasive test to tell you for definite if your baby has one of the three conditions. If you choose not to have an invasive test you will be supported in your decision, and we can give you as much information as you want about the syndrome that the NIPT is high chance for.

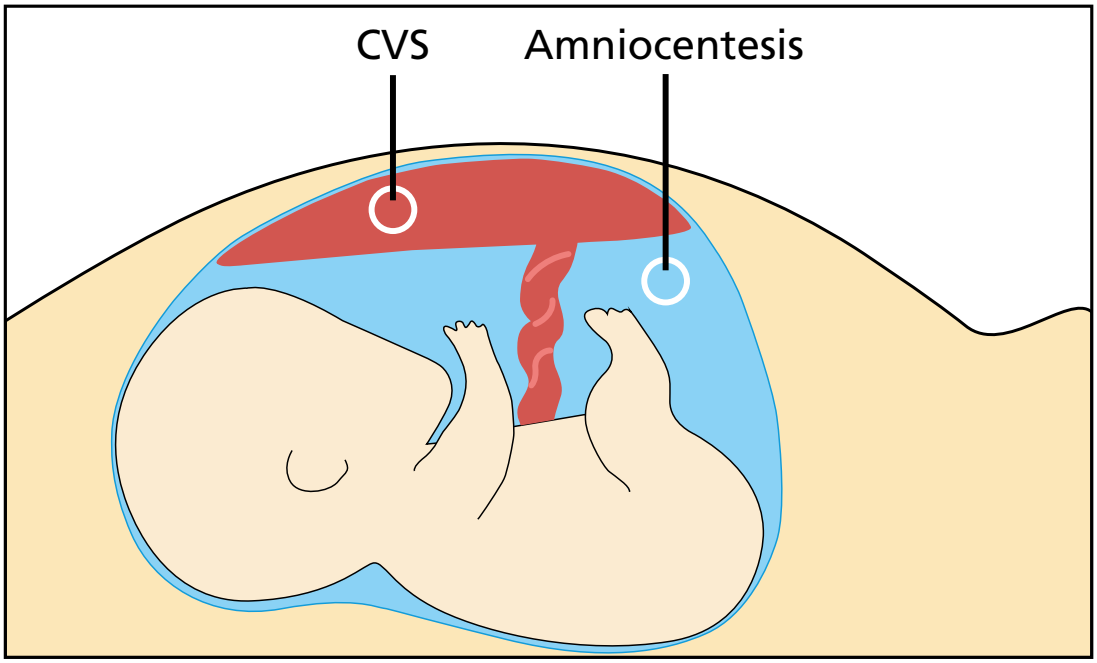
If the NIPT is low chance, it is unlikely that your baby will have Down's syndrome, Edwards' syndrome or Patau's syndrome and you will not be offered any further testing.

What invasive procedures will I be offered if I have a higher or high chance screening result?

Depending on how many weeks pregnant you are, you will be offered either a chorionic villus sampling (CVS) or amniocentesis procedure (see section 7).

What would an invasive test result tell me?

The result would tell you if your baby or babies have Down's syndrome, Edwards' syndrome or Patau's syndrome. If your baby or babies have one of these conditions, you can decide whether to prepare for the birth of your baby or to end your pregnancy.



What is chorionic villus sampling (CVS)?

CVS is a procedure during which a hospital doctor (obstetrician) removes a small amount of tissue from your placenta (afterbirth) during your pregnancy. The cells in this tissue are tested in the laboratory to look for the condition that you are higher chance for. You can usually have CVS after you are 11 weeks and before 14 weeks pregnant.

Having a CVS carries an additional risk of miscarriage which is likely to be below 0.5% (around 1 in 200) of pregnancies.

The additional risk of miscarriage following CVS in a twin pregnancy is around 1% (1 in 100) of pregnancies.

What is amniocentesis?

An amniocentesis is a procedure to remove about 15 to 20 millilitres (that is, three to four teaspoons) of amniotic fluid from around your baby in the womb. The cells from your baby that are floating in this fluid can be tested in the laboratory to look for the condition that you are higher chance for. It can be done after you are 15 weeks pregnant.

Having an amniocentesis carries an additional risk of miscarriage which is likely to be below 0.5% (around 1 in 200) of pregnancies.

The additional risk of miscarriage following amniocentesis in a twin pregnancy is around 1% (1 in 100) of pregnancies.

Section 8 – More information

You can also get information about screening tests from your midwife or your hospital doctor (your obstetrician) and from the Antenatal Screening Wales website at: phw.nhs.wales/antenatal-screening

If you move home

If you move home during your pregnancy, please tell your midwife so that they can update your medical records.

Private tests

The quality of the screening offered by the NHS in Wales is monitored. Some women pay privately to have screening tests. Screening done by private clinics is not monitored by the NHS. This means that your midwife will have no information about the quality and accuracy of any screening tests carried out by private clinics.

CARIS is the Congenital Anomaly Register & Information Service and childhood rare disease register for Wales. CARIS collects information about congenital anomalies and rare diseases.
The information held on the CARIS register is strictly confidential. We will never pass your name to anyone else or publish it.

We hope everyone will want to be included to help us plan and improve services for future parents and children. If you would like us to remove your details from the register, please contact the CARIS team.

Website:phw.nhs.wales/services-and-teams/caris/about-caris/

Email:caris@wales.nhs.uk

Phone:01792 285241

If you want more information on the Public Health Wales privacy notice, please visit:-

phw.nhs.wales/use-of-site/privacy-notice

Or write to:

The Data Protection Officer
Public Health Wales NHS Trust
2 Capital Quarter, Tyndall Street
Cardiff, CF10 4BZ