Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Antenatal screening tests

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

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

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Easy read, audio and British Sign Language versions are available on www.antenatalscreening.wales.nhs.uk

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Clarity approved by Plain English Campaign

www.antenatalscreening.wales.nhs.uk
December 2020
IFW 1st Edition

This booklet includes the following sections.

Section 1 – Introduction to antenatal screening tests

Your choices in pregnancy

During your pregnancy, you will be offered a number of different screening tests. This booklet contains important information about antenatal screening and testing. The information in this booklet 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.

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

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 may affect either you or your baby. They do not show for certain whether you or your baby are affected. They show what chance there is that you or your baby are affected.

The tests offered include blood tests and ultrasound scans.

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

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.

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.

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 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:

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

You can have a test to find out your chance of having a baby with Down's syndrome and your chance of having a baby with Edwards' syndrome or Patau's syndrome. The test can often be done as part of the early pregnancy dating scan offered to you when you are between 11 and 14 weeks pregnant. You would also need to have a blood test on the same day.

If you go to the scan appointment and you are more than 14 weeks pregnant, you can have a blood test to find out your chance of having a baby with Down's syndrome. This is normally carried out between 15 and 18 weeks of pregnancy.

At 18 to 20 weeks

You can have a fetal anomaly scan to see if there are any problems with the way your baby is developing.

Deciding if you want to have the tests

It can be difficult to decide what tests to have. Some women do not want to know if they have any infections that could affect their baby or if there are problems with the way their baby is developing. Others want to know so they can protect their baby against infections or, if there are major problems with their baby, they can think about whether 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.

The sections in this booklet have information about each of the tests.

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.

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

Section 2 – Your Blood group and pregnancy

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

- find out your blood group
- find out your Rhesus D group, and
- look for antibodies.

The people looking after you during your pregnancy need to know your blood group in case you ever need a blood transfusion. It is also important to know your Rhesus D group.

What is your blood group?

Your blood group will be one of the following four main groups.

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

You also have another blood group called the D (Rhesus) group. You can be either Rhesus D positive or Rhesus D negative. For example, your overall blood group might be written as 'O Rh D positive'.

What are red cell antibodies?

Red cells carry oxygen in your blood. Antibodies are your body's natural defence against anything that your body thinks is foreign. You may form antibodies if blood cells with a different blood group from your own enter your bloodstream.

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. This can damage your baby's blood. Sometimes it causes a rare condition called haemolytic disease of the fetus and newborn (HDFN). Symptoms of HDFN include jaundice and anaemia (lack of red blood cells). Affected babies usually need to be admitted to hospital, where treatment includes phototherapy (treatment with light) and sometimes blood transfusions.

The test

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

If you are Rhesus D negative

If you are Rhesus D negative and your baby is Rhesus D positive, and some of your baby's blood cells enter your bloodstream, your body might produce antibodies to destroy these 'foreign' Rhesus D positive blood cells. This sometimes happens during pregnancy, or more likely when your baby is delivered. If you are Rhesus D positive, this problem does not usually happen.

Can haemolytic disease of the fetus and newborn (HDFN) be prevented?

To help prevent HDFN, if you are Rhesus D negative, you will be offered an injection or injections of anti-D immunoglobulin during pregnancy and after delivery if your baby is Rhesus D positive. This can help stop your body making Rhesus D antibodies and reduces the risk of problems in future pregnancies.

What is anti-D immunoglobulin?

Anti-D immunoglobulin (injection) is a blood product made from blood collected from donors.

Is anti-D immunoglobulin safe?

Occasionally anti-D immunoglobulin causes allergic reactions. How it is produced is very strictly controlled, so the risk of a known virus being passed on to you from the 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 Rhesus D positive or Rhesus D negative. It is less common to be Rhesus D negative.

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 might carry the risk of affecting your baby as explained above. If this happens you and your baby can be given specialist care.

About 15% (15 in 100) of the population are Rhesus D negative.

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.

Diagnostic tests for blood group and red cell antibodies

The screening test for blood group and red cell antibodies is very accurate. Sometimes you will need extra blood tests if a problem is suspected or found.

If you know that you are Rhesus D negative

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

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

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

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 (invasive tests)
- you have a miscarriage or after your baby is delivered, or
- you have an ectopic pregnancy.

Section 3 – Infections in pregnancy

Protecting your baby

This section explains about some of the infections that can cause problems 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, your baby could catch the infection from you during the 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 problems 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 your baby catching the infection.

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.

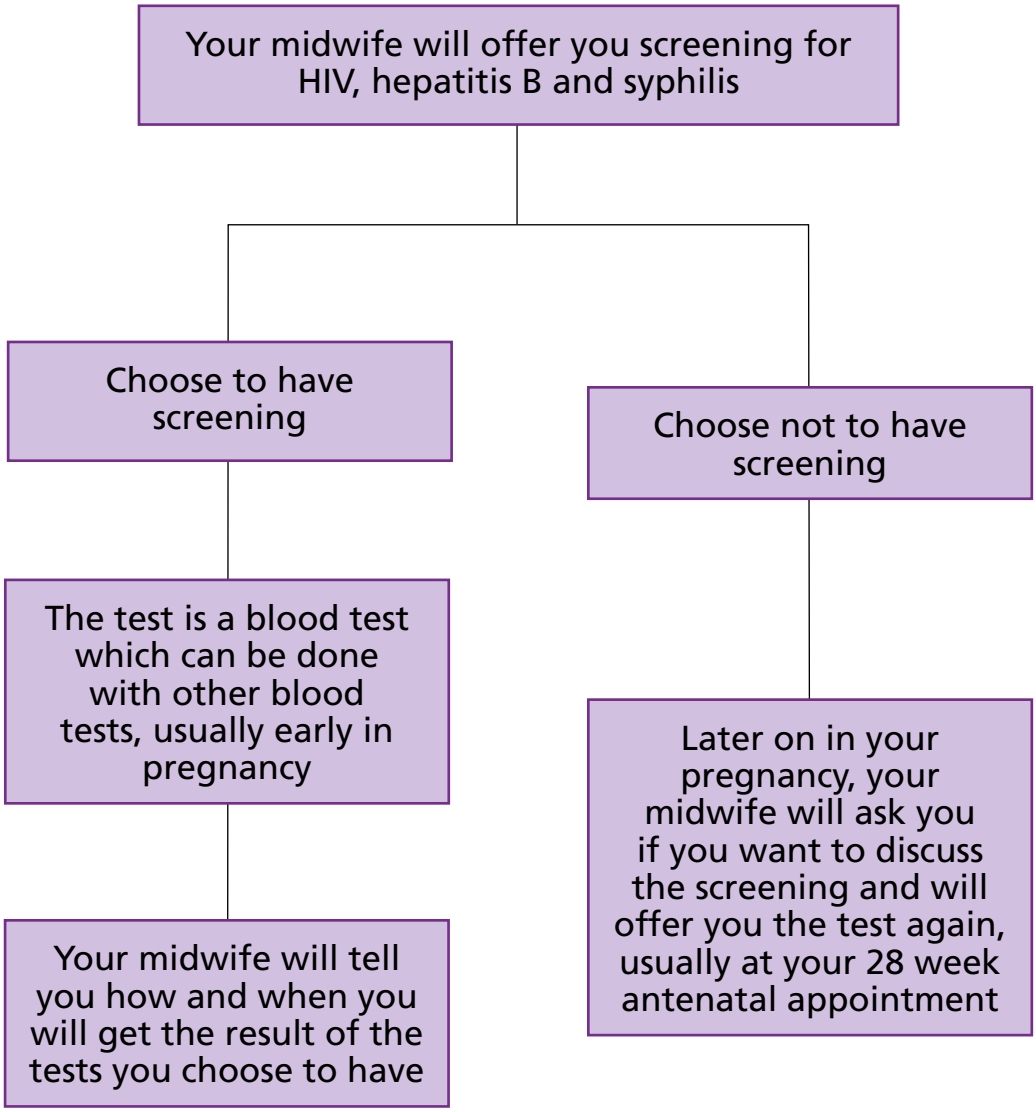
Why should I have the test?

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 unborn child.

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

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, 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.



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 infected with HIV can look and feel well for many years. They may not know they are infected unless they have a blood test. However, this virus 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 is infected passing it to her baby during pregnancy, the delivery or breastfeeding
- **any** unprotected sexual activity (without a condom) with someone who is infected
- a blood transfusion or blood products containing the virus (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.

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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 (3 in 1000 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 baby catching the virus from you.

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 catching the virus.

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 catch 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 may be needed to confirm you do not have an 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 virus.

Treatment will include drug therapy. This treatment will not cure you but it will improve your health. You may also be advised to have a Caesarean delivery and to feed your baby formula milk.

Does having the HIV screening test affect insurance policies?

Insurance companies should not ask if someone applying for insurance has had a test for HIV. They can only ask whether someone has had a positive test result. If you already have a life insurance policy it will not be affected by taking a HIV test, even if the result is positive, as long as you did not withhold any important facts when you took the policy out.

Hepatitis B

What is hepatitis B?

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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 the virus. People who are carriers may develop serious liver disease.

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

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 catching the virus. 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 become infected.

If you are found to have hepatitis B infection your baby can be vaccinated, and the chance of your baby being infected 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 from catching the virus 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 can catch 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 may be needed to confirm you do not have an 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.

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 serious problems later in life, including brain damage and heart problems.

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 an infected person.

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 catching 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 your baby from developing major problems.

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 your baby from developing major problems.

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 catch syphilis when you are pregnant. If you change your sexual partner during the pregnancy, you should use a condom.

What are the diagnostic tests for syphilis?

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Sometimes the syphilis blood test can give an unclear (reactive) result and further tests may 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 diseases. 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 problem.

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.

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 or GUM clinic.

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.

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.

Catching 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 problems) or even heart problems 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 catch measles in pregnancy your symptoms can be more severe.

Parvovirus

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

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 father of your baby will also be offered testing. There are a number of different types of carrier.

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 father of your baby are both carriers of 'important types' 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 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 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 affected?

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.

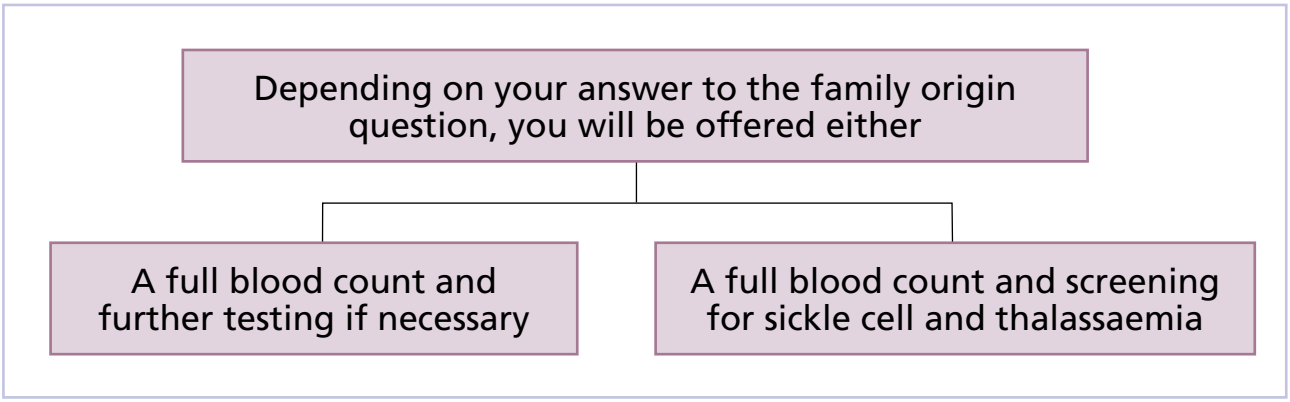
The full blood count can also find some types of thalassaemia.

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 is affected. This could be done by either chorionic villus sampling (CVS) or amniocentesis. If your baby is affected, 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 is affected. Because the invasive tests can cause 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 father of your baby will be offered testing too.

If the 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 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 is affected. 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 is affected, 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 abnormalities. 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.

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 for possible problems with your pregnancy. It is a screening test. This means it may show up problems that would then need to be checked by other tests. A scan cannot pick up all problems.

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 hips. 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 find problems before your baby is born. Finding out about a problem your baby may have before the birth can help you and your partner prepare yourselves. Sometimes it can help plan treatment for after your baby is born. Sometimes when women find out that there is a major problem, 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 a problem is found. If you prefer not to know about any problems that your baby has before the birth, 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 problems 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 problems are found on the scan, the sonographer will tell you about them, and this is not a suitable situation for children.

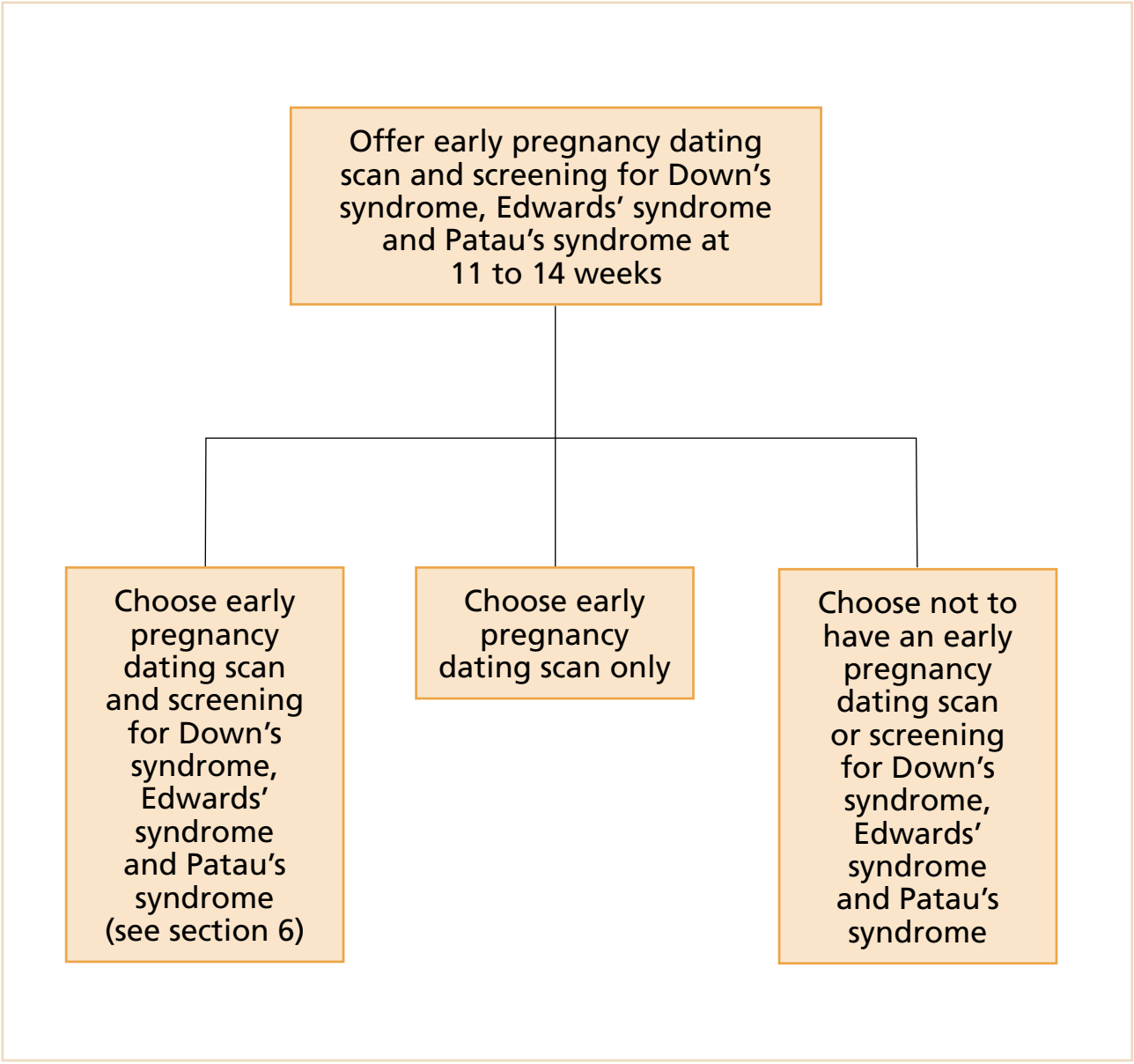
Can I have a picture of my baby?

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 the use of mobile phones within the ultrasound room and during the scan is not allowed.

The early pregnancy dating scan

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



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How long will my scan take?

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

The scan is done to:

- check 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 problems can be detected).

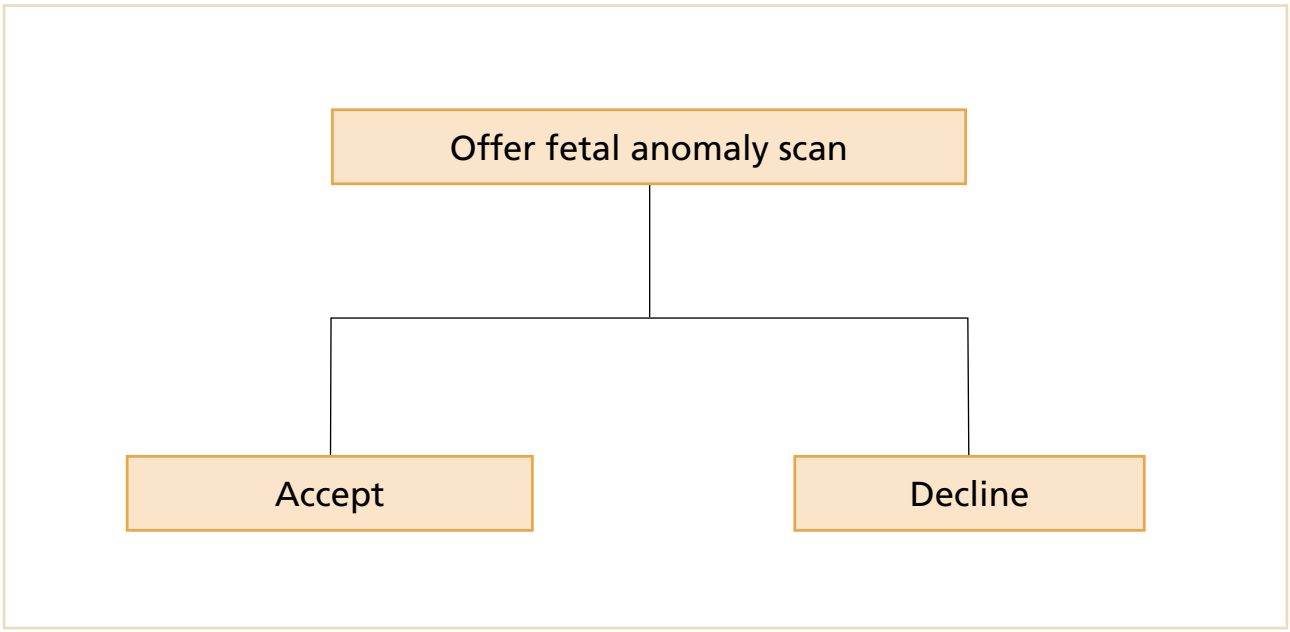
Will I need another early pregnancy dating scan?

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 vaginal 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 will be offered 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 problems (also called anomalies) such as spina bifida 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 problems can the scan find?

Some problems with your baby 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 problems even though no problem was seen during the scan.

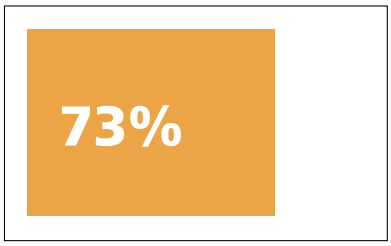
A scan can show some problems with your baby's development but not all.

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

Some issues 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.

Other very rare conditions can be detected on the fetal anomaly scan, such as major kidney and limb problems. Some conditions cannot be detected on this scan, such as small problems with the way your baby is developing, problems that develop after the scan, and conditions like cerebral palsy and autism.

Table 1

The problem	The chance of the problem being seen on an ultrasound 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.	
Major heart condition, for example, tetralogy of fallot Tetralogy of fallot is a serious heart condition where the heart has not developed in the same way as a normal heart in the womb. This condition will need surgery, usually in the first year of birth.	
Autistic spectrum disorders (autism) Autism cannot be picked up on a scan as there is no structural abnormality.	

Data from: Welsh Congenital Anomaly Register and Information Service (CARIS)
www.caris.wales.nhs.uk/antenatal-detection (Accessed 30/09/20)

What will happen if a problem is found, or suspected, during the scan?

If the sonographer finds a problem, 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 bad news can be distressing. We recommend that your partner or one adult only comes with you to the scan appointment.

Some small problems may only need more scans at a later stage of your pregnancy.

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

Invasive tests following a detailed 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 about a problem your baby may have before the birth can help you and your partner prepare yourselves. Information about the type of problem 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 problems can be treated before your baby is born.

If the problem 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 problem 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, Edwards' syndrome or Patau's syndrome. 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). It is recommended that you watch a film clip before you see your midwife for the first time.

The film clip, which gives you more information about screening for these conditions, is available from: www.antenatalscreening.wales.nhs.uk/public/home

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?

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 and leukaemia. 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 problems, 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 get more information about Down's syndrome from the Down's Syndrome Association at www.downs-syndrome.org.uk and you can view some family stories about living with Down's syndrome on the Antenatal Screening Wales website: www.antenatalscreening.wales.nhs.uk/public/home

What are Edwards' syndrome and Patau's syndrome?

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 problems with 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 syndromes 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 get more information about Edwards' syndrome and Patau's syndrome at www.soft.org.uk. You can view family stories about living with Edwards' syndrome on the Antenatal Screening Wales website: www.antenatalscreening.wales.nhs.uk/public/home

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 it is difficult to measure the nuchal translucency. 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.

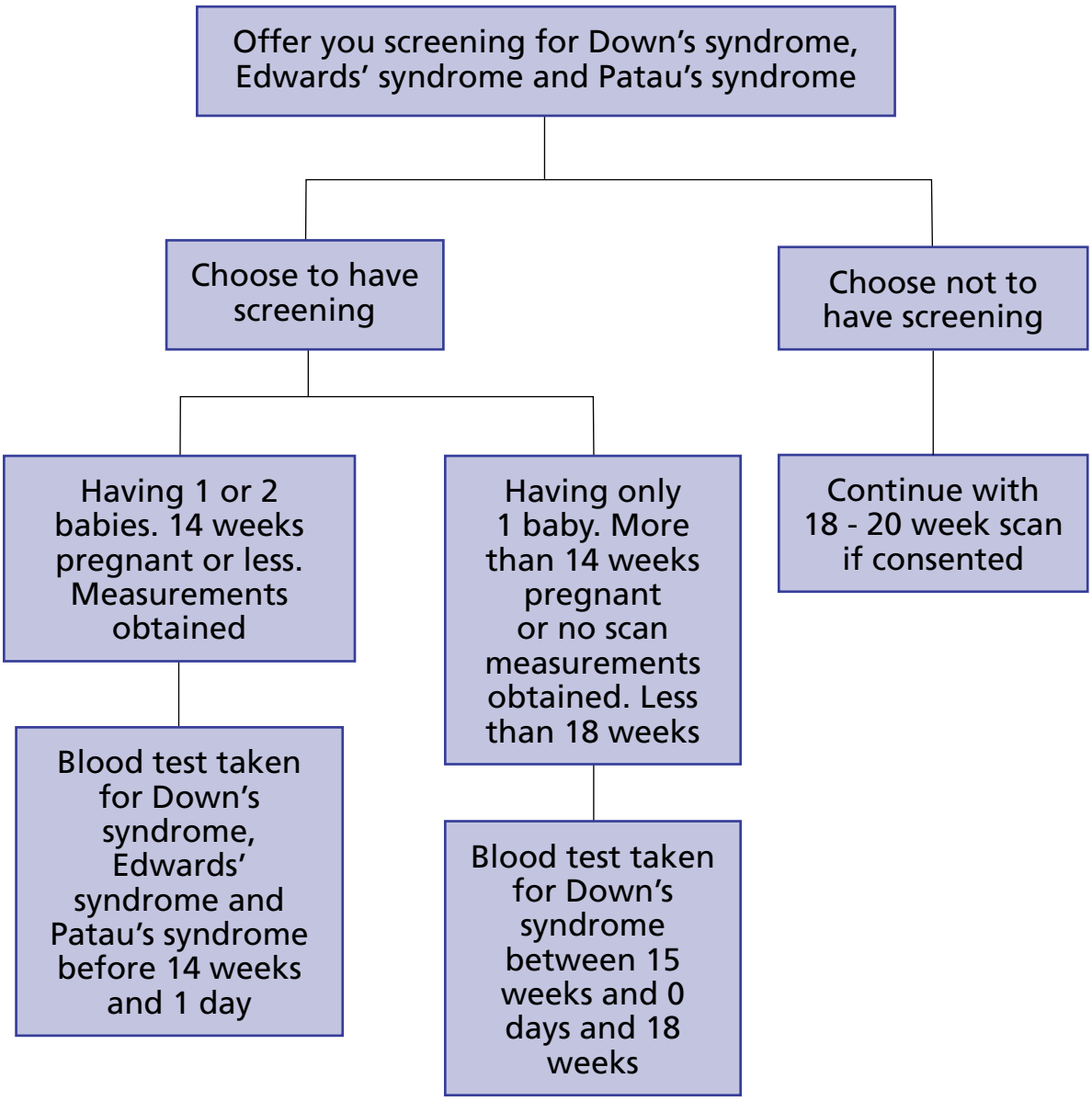
The quadruple screening test is not offered in Wales in twin pregnancies as it is not as accurate as in single pregnancies.

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

You will be offered a quadruple test if the sonographer cannot measure the nuchal translucency 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.

When screening tests are carried out



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 before you are 14 weeks 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 and 18 weeks) 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, for every 10 babies with these conditions only around seven or eight will be identified by screening. This means two or three in every 10 babies with these conditions will not be identified by the screening test.

What are the advantages of having screening for these syndromes?

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 syndromes?

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 syndromes.

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, Edwards' syndrome and Patau's syndrome?

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.

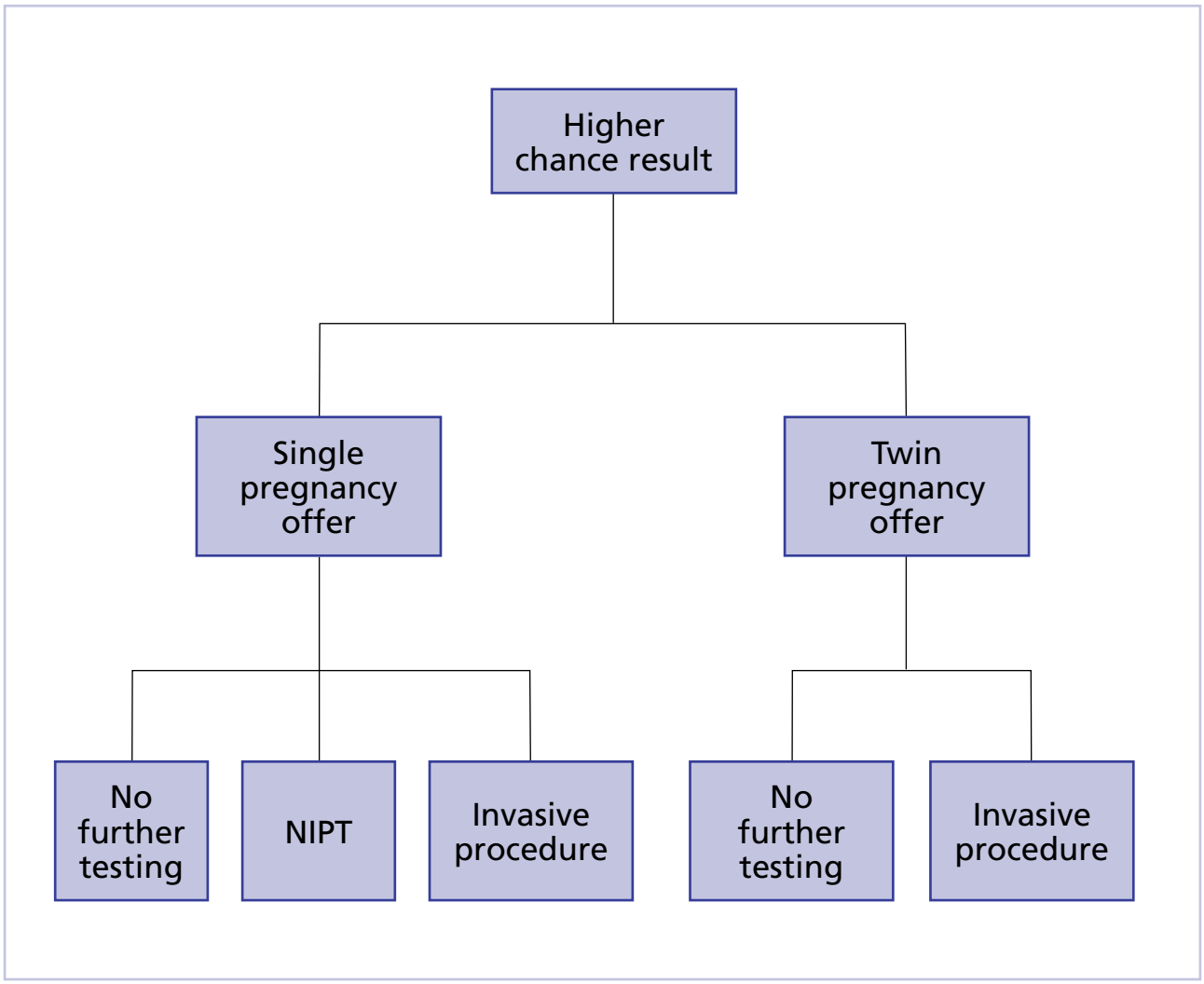
What happens if I have a higher chance result?

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 or 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.

NIPT tests are not available in Wales in twin pregnancies. Between 2% and 4% of women (that is, between two and four in 100) 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 syndromes.

Options for a higher chance result



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 (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.

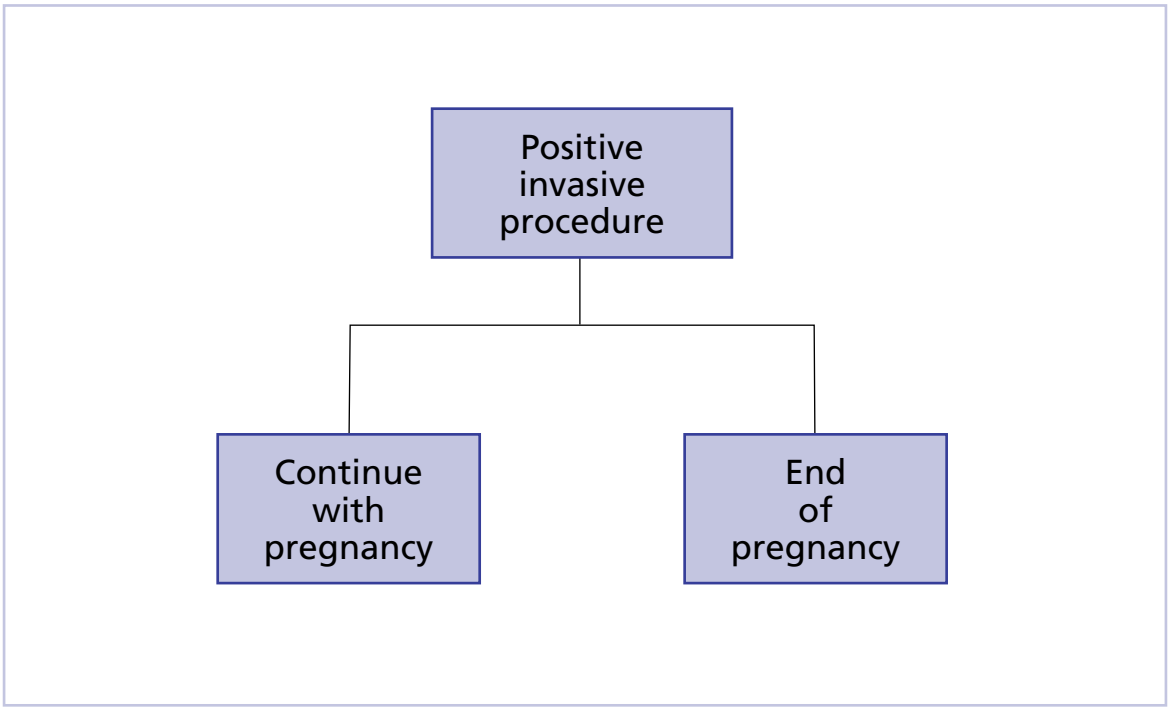
If you choose this test it usually takes about two weeks to get the result. In about 2% (2 in 100 pregnancies) of cases the NIPT test will not be able to give a result and you will be offered an invasive test or you may decide to have no further tests.

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?

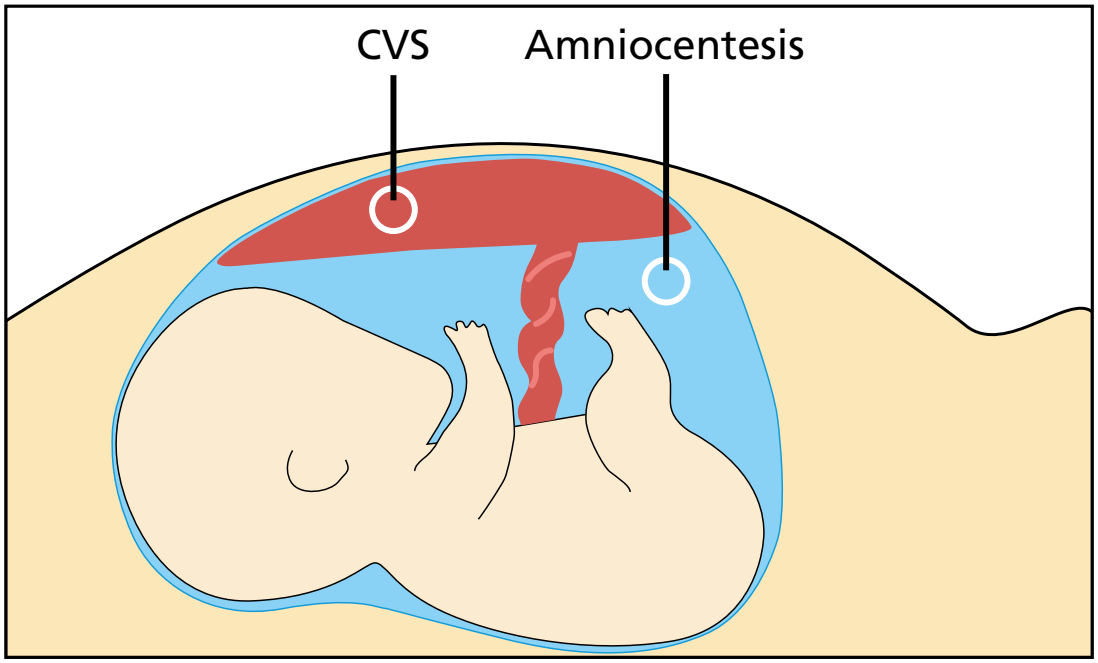
The result would tell you if your baby or babies have Down's syndrome, Edwards' syndrome or Patau's syndrome. If your baby or babies have one of these conditions, you can decide whether to prepare for the birth of your baby or to end your pregnancy.

Film clip

There is a film clip available to give you more information about the screening for Down's syndrome, Edwards' syndrome and Patau's syndrome.

At the end of the film clip there are some questions that you may want to consider before you have your first appointment with your midwife. We have listed some of these questions below.

- Do I know enough about Down's syndrome, Edwards' syndrome or Patau's syndrome in order to make decisions about whether I would want testing for these conditions?
- Do I know how to find out more information if I want it?
- How important it is for me to know whether my baby has Down's syndrome, Edwards' syndrome or Patau's syndrome before it is born, and
    - would I be prepared to have an invasive test that has a 1% to 2% risk of miscarriage to find this out?
- Would I continue with the pregnancy if my baby has Down's syndrome, Edwards' syndrome or Patau's syndrome?
    - Would I need more information about the support available in this instance?
- Would I consider ending the pregnancy for any of these conditions?
- If I take the combined or quadruple test alone, would only knowing my chance of having a baby with one of these conditions be enough for me?
- If I had a higher chance result from the first test, would I want to have a NIPT blood test knowing that it could come back high chance?
- Would I be happy to take the NIPT test in the hope it would come back low chance?
- How important would finding out sooner be (using the invasive test that risks a miscarriage) rather than delaying the result for a NIPT test that may rule out the condition?



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. If you are having twins, the risk of miscarriage will be more than 2% and less than 3% (between 2 and 3 in 100 pregnancies).

Having a CVS carries a risk of miscarriage in more than 1% but less than 2% of pregnancies (between 1 in 50 and 1 in 100 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 a risk of miscarriage in around 1% of pregnancies (1 in 100 pregnancies).

If you are having twins, the risk of miscarriage will be around 2% (2 in 100 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: www.antenatalscreening.wales.nhs.uk

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.

Antenatal Results and Choices (ARC)

Helpline: 0845 077 2290 or 0207 713 7486 from a mobile
E-mail: info@arc-uk.org
Website: www.arc-uk.org

Down's Syndrome Association (DSA)

Phone: 0333 1212 300
E-mail: wales@downs-syndrome.org.uk
Website: www.downs-syndrome.org.uk

Support Organisation for Trisomy 13/18 (Edwards' syndrome and Patau's syndrome) (SOFT UK)

E-mail: enquiries@soft.org.uk
Website: www.soft.org.uk

Sickle Cell Society

Website: www.sicklecellsociety.org
E-mail: info@sicklecellsociety.org

UK Thalassaemia Society

Website: www.ukts.org
E-mail: info@ukts.org

CARIS is the Congenital Anomaly Register & Information Service and childhood rare disease register for Wales.

CARIS collects information about conditions that affect the way babies develop in pregnancy. These conditions are called congenital anomalies, malformations or birth defects. They include Down's syndrome, heart defects or cleft lip.

It is very important to know more about these sorts of conditions and their causes.

CARIS uses the information to find out how common these conditions are. The information CARIS collects also helps to show how good the health service is at picking up these conditions and is used for planning future services.

If it is suspected from your screening tests that there are these types of conditions with your pregnancy, the midwife or obstetrician will pass information about this to CARIS.

The information held on the CARIS register is strictly confidential. We will never pass your name to anyone else or publish it.

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