Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

This information is for you if you have been offered further tests for suspected chromosomal or genetic conditions

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

April 2024

Version 8

You have been given this information because your health professional suspects that your baby may have a higher chance of having a chromosomal or genetic condition.

This booklet includes the following sections:

Not all of the sections in this booklet will be relevant to you. Your health professional will have discussed the sections that apply to you and this will depend on:

- why they suspect there may be a higher chance of your baby having a chromosomal or genetic condition, and
- how many weeks pregnant you are.

- [ ] **If you, or the father of your baby, have a family history or carry a chromosomal or genetic change, or if either of you have had a previous pregnancy that had a chromosomal or genetic condition,** you should read sections 1, 5, 8, 9, 10, 11, 12, 13 and 16 and your health professional will tell you if any other sections are relevant.

- [ ] **If your baby has a higher chance of a chromosomal or genetic change because of something identified on the early pregnancy dating scan,** you should read sections 1, 2, 3, 4, 6, 9, 10, 11, 14 and 16 and your obstetrician will tell you if any other sections are relevant.

- [ ] **If you have a higher chance result from the combined or quadruple screening test,** you should read sections 1, 2, 3, 4, 7, 8, 9, 10, 11 and 16 and your midwife will tell you if any other sections are relevant.

- [ ] **If you have a high chance result from the non-invasive prenatal test (NIPT),** you should read sections 1, 2, 3, 4, 10, 11 and 16 and your midwife will tell you if any other sections are relevant.

- [ ] **If your baby has a higher chance of a chromosomal change because of something identified on the anomaly scan,** you should read sections 1, 10, 11, 14, 15 and 16 and your obstetrician will tell you if any other sections are relevant.

Section 1 – What are chromosomes and what causes chromosomal changes in babies?

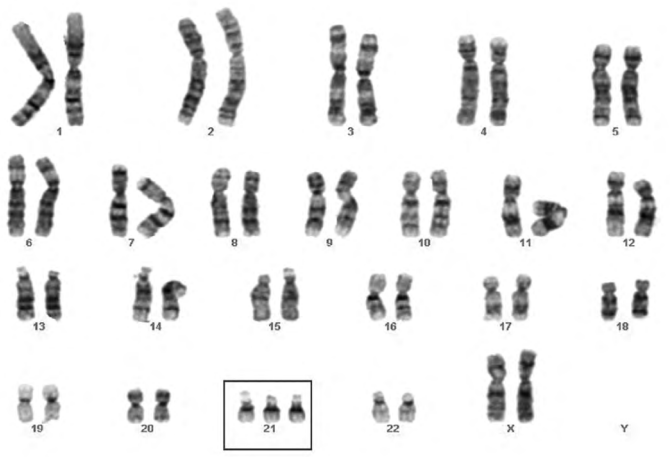
Each cell in the body is usually made up of 23 pairs of chromosomes. Parents contribute one chromosome from the sperm and the egg to each pair of chromosomes in the baby.

- Sometimes changes can happen by chance in the sperm or the egg which lead to an extra chromosome in all or some of the baby's cells. Most chromosomal conditions in babies are caused this way. Nothing you or the baby's father has done or not done can make any difference to this type of condition.
- Sometimes changes can happen by one chromosome breaking off and attaching to another chromosome. These types of changes are often referred to as translocations, and in Down's syndrome, for example, they happen in around 4% (1 in 25) of cases.
- Occasionally, in around 1% (1 in 100) of cases some of the cells in the body are affected by the changes and some are not. This tends to make the effect of the chromosomal changes less serious. This is known as a mosaic chromosomal change.

The most common chromosomal change is called Down's syndrome. This is caused by an extra copy of chromosome 21 in some or all of the cells. An extra copy of chromosome 18 causes Edwards' syndrome. An extra copy of chromosome 13 causes Patau's syndrome. The picture below shows the chromosomes from the cells of a baby as seen under a microscope. There is a square around the three copies of chromosome 21 that cause Down's syndrome − you will see that all the others are pairs.

There is a wide range of potential changes to the chromosomes that may affect babies in different ways.

If the midwife or obstetrician suspects any other chromosomal changes in your baby, they will talk to you about those conditions.



Trisomy 21: 47,XX,+21

Section 2 – What is Down's syndrome and living with Down's syndrome?

What is Down's syndrome?

Down's syndrome is a genetic condition caused by an extra chromosome 21 in all or some cells. Down's syndrome is also known as trisomy 21, or T21. A person with Down's syndrome has 47 chromosomes instead of the usual 46. The extra chromosome cannot be removed from cells even if Down's syndrome is diagnosed before the baby is born.

All women have a chance of having a baby with Down's syndrome. Nothing you or the baby's father have done or not done can make any difference to this chance.

All people with Down's syndrome will have a learning disability. This means they may be delayed in their development and take longer to learn new things. There is a greater understanding these days of how children with Down's syndrome learn, and help is provided in education settings. Around 80% (8 in 10) of children will go to mainstream primary schools, although individuals vary greatly in how they develop and will have different health and support needs. The antenatal tests cannot tell you what the health and support needs of your baby may be.

You can watch some family stories on living with Down's syndrome at: phw.nhs.wales/antenatal-screening

There are support organisations available for pregnant women and for families who have a child with Down's syndrome. These include the Down's Syndrome Association (DSA). Helpline 0333 12 12 300.
Website: www.downs-syndrome.org.uk

Children and adults with Down's syndrome

In Wales 90% (9 in 10) of children with Down's syndrome live past their 5th birthday. For babies without serious health conditions survival is similar to that of other children, and most people with Down's syndrome will live into their 60s.

Most children and adults who have Down's syndrome lead healthy and fulfilled lives and are included in their community. Most say they enjoy their lives and relationships. Many adults are capable of work and live in their own accommodation, with support.

People born with Down's syndrome are more likely than most to have some medical conditions.

- In Wales about 60% (6 in 10) of children with Down's syndrome will have a heart condition, and around 30% (3 in 10) will need an operation.
- Most children with Down's syndrome will have some sight difficulty that will need monitoring or treatment. More serious sight difficulty is less common, for example, 0.4% (1 in 250) of children with Down's syndrome are born with cataracts.
- Around 60% (6 in 10) of children with Down's syndrome will have some hearing loss which may cause challenges with speech and language.
- Infections of the ears, nose and throat are more likely.
- Leukaemia is more common in children with Down's syndrome and for most this will not cause any health conditions. 0.5% (1 in 200) of children with Down's syndrome will need treatment for leukaemia. Following treatment, most children with leukaemia will recover with no related health issues.

Babies and children with Down's syndrome will be under the care of a specialist medical team who will be aware of the increased chances of these medical conditions and will do tests to monitor their wellbeing. Many of these conditions can be treated effectively.

Section 3 – What is Edwards' syndrome and living with Edwards' syndrome?

What is Edwards' syndrome?

Edwards' syndrome is a genetic condition caused by an extra chromosome 18 in all or some cells. Edwards' syndrome is also known as trisomy 18, or T18. A person with Edwards' syndrome has 47 chromosomes instead of the usual 46. The extra chromosome cannot be removed from cells even if Edwards' syndrome is diagnosed before the baby is born.

All women have a chance of having a baby with Edwards' syndrome. Nothing you or the baby's father have done or not done can make any difference to this chance. Antenatal tests will not be able to tell you what specific challenges your baby might have.

Most babies with Edwards' syndrome will die before they are born or shortly after birth. Of the babies who survive, about 13% (1 in 7) will live for more than a year. Some babies with the less severe types of Edwards' syndrome (such as those with a mosaic chromosomal change) can survive into early childhood but rarely survive to adulthood.

You can watch some family stories on living with Edwards' syndrome at: phw.nhs.wales/antenatal-screening

All people with Edwards' syndrome will have severe lifelong learning disabilities.

There are support organisations available for families who have a child with Edwards' syndrome. These include the Support Organisation for Trisomy 13/18 (Patau's syndrome and Edwards' syndrome) (SOFT).
Website: www.soft.org.uk

Babies born with Edwards' syndrome will have a narrow but often serious range of conditions.

- A significant delay in their development (all babies).
- Heart conditions (many babies).
- Feeding difficulties (many babies).
- Medically fragile, especially with respiratory conditions (most babies, especially when small).
- Cleft lip and palate (some babies).
- Prone to conditions such as urinary tract infections (UTIs).

3

At the other end of the scale, research shows, for example, that:

- babies and children make progress, however slowly
- older babies and children show some level of communication
- some will stand and walk with help, and
- parents consistently report a high quality of life for their babies and children, because they are involved in family activities.

Babies and children with Edwards' syndrome will be under the care of a specialist medical team who will be aware of the increased chances of the medical conditions and will do tests to monitor their wellbeing. Some of these conditions can be treated effectively.

Children born with Edwards' syndrome usually develop and learn more slowly than other children. They have a range of specific challenges. They will need to attend special school.

Section 4 – What is Patau's syndrome and living with Patau's syndrome?

What is Patau's syndrome?

Patau's syndrome is a genetic condition caused by an extra chromosome 13 in all or some cells. Patau's syndrome is also known as trismoy 13, or T13. A person with Patau's syndrome has 47 chromosomes instead of the usual 46. The extra chromosome cannot be removed from cells even if Patau's syndrome is diagnosed before the baby is born.

All women have a chance of having a baby with Patau's syndrome. Nothing you or the baby's father have done or not done can make any difference to this chance.

Antenatal tests will not be able to tell you what specific challenges your baby might have.

Around 90% (9 in 10) of babies with Patau's syndrome will die before they are born or shortly after birth. Of the babies who survive, about 12% (1 in 8) live past the age of one. Some babies with the less severe types of Patau's syndrome, such as those with a mosaic chromosomal change, do survive beyond a year and can survive to adulthood, but this is rare.

All people with Patau's syndrome will have severe lifelong learning disabilities.

There are support organisations available for families who have a child with Patau's syndrome. These include the Support Organisation for Trisomy 13/18 (Patau's syndrome and Edwards' syndrome) (SOFT).
Website: www.soft.org.uk

Babies born with Patau's syndrome will have a narrow but often serious range of conditions.

- A significant delay in their development (all babies).
- Heart conditions (many babies).
- Feeding difficulties (many babies).
- Medically fragile, especially with respiratory conditions (most babies, especially when small).
- Cleft lip and palate (some babies).
- Prone to conditions such as urinary tract infection (UTIs).

At the other end of the scale, research shows, for example, that:

- babies and children make progress, however slowly.
- older babies and children show some level of communication.
- some will stand and walk with help, and
- parents consistently report a high quality of life for their babies and children, because they are involved in family activities.

Babies and children with Patau's syndrome will be under the care of a specialist medical team as babies and children with this condition will have complex needs.

Children born with Patau's syndrome usually develop and learn more slowly than other children. They have a range of complex challenges. They will need to attend special school.

Section 5 – What happens if I or the father of my baby has a family history, or if either of us carries a chromosomal or genetic change, or if either of us has had a previous pregnancy with a chromosomal or genetic condition?

This will depend on the type of chromosomal or genetic change that you or the father of the baby carries. We may:

- offer you or the father of the baby a referral to medical genetics to discuss your options
- offer the partner of the person who carries a chromosomal change a blood test to see if there is a chance that your baby may have the chromosomal change, or
- offer you chorionic villus sampling (CVS) or amniocentesis to see whether your baby has the chromosomal change.

If we offer you CVS or amniocentesis you should be aware that both of these tests can cause a miscarriage.

5

Section 6 – What happens if my baby has a higher chance of a chromosomal change because of something identified on the early pregnancy dating scan?

Nuchal translucency (NT) 3.5mm and above

The NT is the small collection of fluid at the back of the baby's neck that the sonographer will measure if they identify that it looks big or if you have asked for screening for Down's syndrome (T21), Edwards' syndrome (T18) and Patau's syndrome (T13).

Most babies where the NT measures 3.5mm and above will have no other problems. However, it is more likely that these babies could have certain issues, which the health professional will have discussed with you. One of these is that the baby is slightly more likely than most to have a chromosomal change.

You may have chosen to have screening for Down's syndrome, Edwards' syndrome and Patau's syndrome and the obstetrician may have discussed the result of this test with you.

We may offer you chorionic villus sampling (CVS) or amniocentesis to see whether your baby has a chromosomal change. If we offer you CVS or amniocentesis you should be aware that both of these tests have a small risk of causing a miscarriage.

Other findings on the early pregnancy dating scan – the health professional will discuss the findings of the scan with you and tell you which tests they will offer. They will tell you which sections of this booklet are relevant to you.

6

Section 7 – What does a higher chance result from combined or quadruple screening mean?

If your test result showed you had a higher chance of having a baby with Down's syndrome (T21) or a higher chance of having a baby with Edwards' (T18) or Patau's (T13) syndrome (that is, a chance of 1 in 2 to 1 in 150), your midwife or obstetrician will have explained your test result to you in detail, including your individual chance. Remember that the lower the number, the higher the chance. So, for example, 1 in 80 is a higher chance of having a baby with Down's syndrome than 1 in 140.

Between 2% and 3% (two and three in 100) of women who have the screening test have a result which shows they have a higher chance of having a baby with one of these conditions.

If you are having twins you will have had an in-depth discussion with an obstetrician or midwife before having your combined test. This is because the decisions are more complicated as there are many issues to consider. The discussion will have included information on the type of twins you are having and what that could mean in relation to the chances of either one or both babies having one of the conditions screened for.

As you have a higher chance result, your choices are:

- no further testing.
- NIPT, or
- invasive testing.

No further testing

Some women will want to prepare themselves for the birth knowing that their baby or babies have a chromosomal condition, while others may decide not to continue with the pregnancy.

You can discuss whether or not you want to have a non-invasive prenatal test (NIPT) or an invasive procedure.

You may face some difficult decisions after an invasive procedure that you need to be aware of beforehand. If, for example, you are pregnant with twins, and the invasive test (chorionic villus sampling (CVS) or amniocentesis) shows that only one baby has the condition, deciding whether to continue with the pregnancy can be very difficult. Ending the pregnancy for the baby with the condition could also cause a miscarriage of the other twin. Your obstetrician will discuss this with you in more detail.

7

Section 8 – What is the non-invasive prenatal test (NIPT)?

You will be offered the NIPT screening test if you have a higher chance result from combined or quadruple screening and you are having only one baby or twins.

NIPT is a further screening test that is more accurate than the combined or quadruple test. It will not give a definite result. It is a blood test taken from you in the usual way – your blood will have your DNA (genetic material) in it and some of your baby's DNA in it which is released from the placenta (afterbirth). The blood is sent to the laboratory and screened for Down's syndrome (T21), Edwards' syndrome (T18) and Patau's syndrome (T13). If there is more DNA than expected for chromosomes 21,18 or 13 in your blood, it could mean that your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome. The result will tell you whether there is a low or high chance that your baby has one of the conditions. This test will not give a result for any other conditions, or the sex of your baby. If you choose this test it takes about two weeks to get the result.

The reason why this test is not 100% accurate is because it tests the DNA from the placenta and, in rare cases, this is not identical to the baby's DNA. This is called confined placental mosaicism.

This test can be performed up to 20 weeks of pregnancy.

You will be unable to have the NIPT if:

- this pregnancy was initially a twin pregnancy and one baby died during pregnancy, or
- you have had certain medical conditions, including if you have had a tumour, transplant or recent blood transfusion.

Your midwife will be able to talk to you about this.

What do the results mean?

As NIPT is a screening test, it will not give you a definite result. Instead, it will give you a:

- low chance result, or
- high chance result.

NIPT will give an accurate result for most women who choose to have the test, but there can be false positive and false negative results.

NIPT may also be less accurate in a twin pregnancy.

A false positive result means getting a high chance result when the baby does not have the condition.

A false negative result means getting a low chance result when the baby does have the condition.

Low chance results

Most women who have a NIPT will receive a low chance result for Down's syndrome, Edwards' syndrome or Patau's syndrome. If you receive a low chance result, you will not be offered an invasive test (chorionic villus sampling (CVS) or amniocentesis). For most women the result is accurate.

You should discuss your individual result of your combined or quad test with your health professional before deciding on further testing.

High chance results

If you have a high chance NIPT result for Down's syndrome, Edwards' syndrome or Patau's syndrome, you will be offered an invasive test (chorionic villus sampling (CVS) or amniocentesis) or you may choose to have no further testing.

- At least 90% (9 in 10) of women who have received a high chance NIPT result for Down's syndrome will be carrying a baby who has the condition.

NIPT is less accurate in finding babies with Edwards' syndrome or Patau's syndrome than it is for Down's syndrome.

- Around 80% (8 in 10) of women who have received a high chance NIPT for Edwards' syndrome or Patau's syndrome will be carrying a baby with one of the conditions.

No result from NIPT

Around 0.3% (1 in 300) of tests will not give a result, often because there are not enough of the baby's cells in your blood. In this case you will be offered an invasive test to give you a result. Your midwife will be able to talk to you about this.

Section 9 – What is chorionic villus sampling (CVS)?

CVS is a procedure during which an obstetrician removes a small amount of tissue from your placenta (afterbirth) during pregnancy. The cells in this tissue are tested in the laboratory to look at your baby's chromosomes. You can usually have CVS between 11 and 14 weeks of pregnancy.

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.

A miscarriage is most likely to happen up to three weeks after CVS. No one knows why this happens or who it will happen to. It can happen whether or not your baby has a chromosomal change. CVS is done early in pregnancy, which is when miscarriages are slightly more common in all pregnant women.

Because CVS is a specialised procedure, you may not be able to have it done locally. Instead, you may be offered an appointment at a different maternity unit.

Your midwife or obstetrician can explain the test to you. **It is your choice if you want this procedure or not.**

If you decide to go ahead with CVS, we will ask you to sign a consent form agreeing to the procedure before it is carried out.

Preparing for CVS

- You can have breakfast or a light lunch before your appointment.
- You will need a full bladder for the scan you will have before CVS.
- You may be asked to empty your bladder for the CVS procedure itself.
- You may be more comfortable if you wear loose clothing.
- You can bring your partner or a friend with you for support during and after the procedure, but please don't bring any children with you.
- If possible, you should arrange for someone to drive you home.

Having CVS

The procedure takes about 20 minutes and you will have it done as an outpatient, usually in the antenatal clinic. You will be awake for the procedure, and lying down.

You will have an ultrasound scan before CVS. This is to check the baby's position and to find the best place to take the sample from your placenta.

The obstetrician will give you an injection of local anaesthetic to numb the skin of your abdomen. Your abdomen is then cleaned with an antiseptic solution to reduce the risk of infection. The obstetrician inserts a needle through your skin and the wall of your womb, and then takes a small sample from the placenta. The obstetrician will be watching the ultrasound scan to guide the needle and so avoid getting close to your baby. You may find the test a little uncomfortable.

You will also be asked to give a blood sample so that the laboratory can be sure that the results they get from the CVS sample are for your baby rather than for you. Occasionally the procedure cannot be done due to the position of the baby. If this happens, the obstetrician may suggest that the procedure is done on another day.

What happens after the procedure?

After the procedure you should rest in the clinic for up to 30 minutes. You may have tummy cramps afterwards, rather like period pains.

If your blood group is Rhesus negative, you will be offered an injection of anti-D after the procedure. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is Rhesus positive.

Some obstetricians may advise you to take things easy for a couple of days after the procedure, and to avoid having sex, or doing any heavy lifting or strenuous exercise. You won't need complete bed rest.

If you have any pain or discomfort, you can take a normal dose of paracetamol.

Most women are back to normal after two days.

What you should look out for

- Severe pain, which you can't control by taking mild painkillers (like paracetamol).
- Any bleeding or unpleasant discharge from your vagina.
- Any fluid leaking from your vagina.
- If you suddenly feel unwell, with a high temperature or flu-like symptoms.

These symptoms do not always mean there is a problem, but you may need further care and attention. For advice, please contact:

- the clinic where you had CVS, or
- your midwife.

If your blood group is D negative, you will be offered a test called cell free fetal DNA which will predict your baby's blood group.

An injection of anti-D after the procedure will be recommended if you have not had the cell free fetal DNA test, or if your baby is predicted to be D positive or you have had an inconclusive result. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is D positive.

Section 10 – What is amniocentesis?

Amniocentesis is a procedure during which an obstetrician removes a small amount (about 15 to 20 millilitres) of amniotic fluid from around your baby in your womb. The cells from your baby that are floating in this fluid can be tested in the laboratory to look at the chromosomes. Amniocentesis is usually done after 15 weeks of pregnancy.

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.

A miscarriage is most likely to happen up to three weeks after the amniocentesis. No one knows why this happens or who it will happen to. It can happen whether or not your baby has a chromosomal change.

Because amniocentesis is a specialised procedure, you may not be able to have it done locally. Instead, you may be offered an appointment at a different maternity unit.

If you have an infection such as HIV, hepatitis B or hepatitis C, you may need extra information and advice from a doctor who specialises in infectious diseases before you decide whether to have amniocentesis.

Your midwife or obstetrician can explain the test to you. **It is your choice if you want this procedure or not.**

If you decide to go ahead with an amniocentesis, we will ask you to sign a consent form agreeing to the procedure before it is carried out.

Preparing for an amniocentesis

- You can have breakfast or a light lunch before your appointment.
- You will need a full bladder for the scan you will have before the amniocentesis.
- You may be asked to empty your bladder for the amniocentesis procedure itself.
- You may be more comfortable if you wear loose clothing.
- You can bring your partner or a friend with you for support during and after the procedure, but please do not bring children with you.
- If possible, you should arrange for someone to drive you home.

Having an amniocentesis procedure

The procedure takes about 10 minutes and you will have it done as an outpatient, usually in the antenatal clinic. You will be awake for the procedure, and lying down.

You will have an ultrasound scan before the amniocentesis. This is to check the position of your baby and to look for the best place to take the sample of fluid from inside your womb. Your abdomen is cleaned with an antiseptic solution to reduce the risk of infection. The obstetrician inserts a needle through your skin and the wall of your womb, and then takes a small amount of fluid from around your baby. The obstetrician will be watching the ultrasound scan to guide the needle and so avoid getting close to your baby. You may find the test a little uncomfortable.

Occasionally the procedure cannot be done due to the position of the baby. If this happens, the obstetrician may suggest that the procedure is done on another day.

There is a small chance that the obstetrician will not be able to get any amniotic fluid from around your baby. This means the test cannot be done. The obstetrician may suggest that the procedure is done again on another day.

You will also be asked to give a blood sample so that the laboratory can be sure that the results they get from the amniocentesis are for your baby rather than for you.

What happens after the procedure?

After the procedure you should rest in the clinic for up to 30 minutes. You may have tummy cramps afterwards, rather like period pains.

10

If your blood group is Rhesus negative, you will be offered an injection of anti-D after the procedure. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is Rhesus positive.

Some obstetricians may advise you to take things easy for a couple of days after the procedure and to avoid having sex, or doing any heavy lifting or strenuous exercise. You won't need complete bed rest.

If you have any pain or discomfort, you can take a normal dose of paracetamol.

Most women are back to normal after two days.

What to look out for

- Severe pain, which you can't control by taking mild painkillers (like paracetamol).
- Any bleeding or unpleasant discharge from your vagina.
- Any fluid leaking from your vagina.
- If you suddenly feel unwell and have a high temperature or flu-like symptoms.

These symptoms do not always mean there is a problem, but you may need further care and attention. For advice, please contact:

- the clinic where you had the amniocentesis, or
- your midwife.

If your blood group is D negative, you will be offered a test called cell free fetal DNA which will predict your baby's blood group.

An injection of anti-D after the procedure will be recommended if you have not had the cell free fetal DNA test, or if your baby is predicted to be D positive or you have had an inconclusive result. This is to reduce the chance of antibodies developing in your blood, which could happen if your baby's blood group is D positive.

10

Section 11 – What is a quantitative fluorescence-polymerase chain reaction (QF-PCR) test?

QF-PCR test on a chorionic villlus sampling (CVS) or amniocentesis sample following a higher chance result from the combined or quadruple screening tests or a high chance NIPT test

The QF-PCR test will look for the following chromosome conditions in the baby.

- Down's syndrome (T21)
- Edwards' syndrome (T18)
- Patau's syndrome (T13)

It will not tell you the sex of your baby.

QF-PCR test for any other reason than a higher/ high chance result

If your QF-PCR test is being done for any other reason, your obstetrician or genetic counsellor will discuss the specific conditions the test will give you a result for.

QF-PCR result

If you have had an amniocentesis procedure, your QF-PCR result is usually available within three calendar days.

About 4% (1 in 25) of amniocenteses will not be able to give a result within this time. In these cases the cells from your baby that are floating in the amniotic fluid are grown (cultured) in the laboratory. This result will be available in 10 to 14 calendar days. This does not mean it is more likely that there is a problem with your baby.

If you have had CVS the result is usually available within 10 to 14 calendar days.

The person taking the sample will have asked you how you would like to receive your results.

In about 4% or 5% (four or five in 100) of QF-PCR tests we may need blood samples from you and the baby's father to help us get a clearer result. Again this does not mean it is more likely that there is a problem with your baby.

If the test shows your baby has Down's syndrome, Edwards' syndrome or Patau's syndrome, the laboratory will do a further test called a karyotype test to see if any other babies you may have in the future could have the condition.

11

Section 12 – What is a karyotype?

This laboratory test takes longer than the QF-PCR test but will detect more chromosomal changes. It is not done for higher chance screening results from the combined or quadruple test or NIPT, unless these show your baby has Down's syndrome (T21), Edwards' syndrome (T18) or Patau's syndrome (T13). The laboratory will carry out a karyotype test to see if any other babies you may have in the future could have the condition.

To do this test, the cells from your baby that are floating in the amniotic fluid or from the chorionic villus sampling (CVS) are grown (cultured) in the laboratory (this takes about 10 to 14 calendar days). The laboratory staff will examine the cells under a microscope to look for changes in the number and appearance of your baby's chromosomes.

The karyotype test will not detect:

- changes in single genes, such as cystic fibrosis (each chromosome contains thousands of genes)
- microdeletions (loss of small segments of a chromosome), or
- other small changes in chromosomes.

Karyotype test results

These results are usually ready within about two weeks.

Sometimes it takes a bit longer to complete these tests, and you will have to wait a few extra days. (This does not mean there is more chance of a problem with your baby).

In some cases, a karyotype may give a result that is not clear. Your obstetrician will discuss this with you if this happens.

The karyotype test will show the sex of your baby. If you do not want to know the sex of your baby, please tell your midwife or obstetrician.

12

Section 13 – What are single-gene tests?

Single-gene disorders

Single-gene disorders are caused by changes in a single gene. If you have a family history of an inherited disorder that is caused by an alteration in a single gene (such as cystic fibrosis), you may be offered a specific test, called a single-gene test, just for this. You may already have spoken to someone at the genetics service, or your midwife or obstetrician can refer you to the genetics service if you tell them you have a family history of an inherited disorder. The genetics team will discuss the options with you and explain the risks. Options may include invasive tests, such as chorionic villus sampling (CVS) or amniocentesis, or non-invasive methods such as a detailed ultrasound scan or a blood test. If a single-gene test is done on an amniotic fluid or CVS sample, you will usually be offered a QF-PCR test as well.

Single-gene test results

Results of tests for single-gene disorders take different lengths of time. The genetics team will discuss with you how you will get the results of single-gene tests.

13

Section 14 – What is a single nucleotide polymorphism array (SNP) test?

This test is offered **only** when there are certain structural changes seen on an ultrasound scan.

A single nucleotide polymorphism array (SNP) is a test that looks at the chromosomes in the cells in the chorionic villus sampling (CVS) or amniotic fluid sample. The test looks for small changes in genetic material. A change in this genetic material may explain the cause of the abnormalities seen on the ultrasound scan.

The SNP array test:

- will not detect changes in single genes, such as cystic fibrosis (each chromosome contains thousands of genes).
- may identify a chromosomal change that may not be related to the findings identified on the ultrasound scan (these changes may have implications for the future health of your baby or other members of your family), and
- may show some chromosomal changes that have an unknown effect on your baby.

This test looks for chromosomal changes in more detail than the QF-PCR or karyotype test.

SNP array test results

These results are usually ready within about 14 calendar days.

Sometimes it takes a bit longer to complete these tests (this does not mean there is more chance of a problem with your baby).

Occasionally, in about 0.4% (1 in 250) of tests you will not get a result. If this happens, you may be offered another amniocentesis.

In some cases a SNP array test may give a result that is not clear.
Your obstetrician will discuss this with you if this happens.

The SNP array test will show the sex of your baby. If you do not want to know the sex of your baby, please tell your midwife or obstetrician.

14

Section 15 – What happens if my baby has a higher chance of a chromosomal change or a genetic condition because of something identified on the fetal anomaly scan?

You will have been told that there is something identified on your fetal anomaly scan which means that your baby may be at higher chance of having a chromosomal change or a genetic condition. The health professional will discuss the findings of your scan with you and tell you which tests they will offer. This may include SNP array or other genetic tests.

We may offer you an amniocentesis to see whether the baby has a chromosomal change or a genetic condition. If we offer you an amniocentesis you should be aware that this test can cause a miscarriage.

If you have had chorionic villus sampling (CVS) or amniocentesis earlier in pregnancy for a higher chance result from the screening test for Down's syndrome, Edwards' syndrome or Patau's syndrome, the laboratory may be able to carry out extra tests on the sample to look for other changes in chromosome or genes.

15

Section 16 – What are my options if my baby has Down's syndrome, Edwards' syndrome or Patau's syndrome or another chromosomal or genetic change found on an invasive test?

There will be some pregnancies where a chromosomal or genetic change is found. You will be given information and support to help you make a decision about your pregnancy. You will be able to discuss the result with an obstetrician and with a midwife who specialises in antenatal screening. You will also be able to discuss the result with someone who specialises in genetics if that is needed.

Finding out that your baby has a chromosomal or genetic change can cause a mixture of emotions, and deciding what to do is hard. There are experts who can give you information to help you decide what to do. These include fetal medicine consultants, geneticists and paediatricians. 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. If you want, your partner or a friend can come to hospital appointments with you.

There are support organisations available for families who have a child with Down's syndrome (T21), Edwards' syndrome (T18) or Patau's syndrome (T13).

You can watch some family stories on living with either Down's syndrome or Edwards' syndrome at:
phw.nhs.wales/antenatal-screening

Some women will want to prepare themselves for the birth knowing that their baby has a chromosomal or genetic condition, while others may decide not to continue with the pregnancy. Whichever decision you make, the person looking after you will support you and agree a plan of care.

These are difficult decisions and you will be given time and information to help you make a decision that is right for you.

Whatever decision you make, the people caring for you will respect your wishes.

More information

You can get more information from the following.

Antenatal Results and Choices (ARC)
Helpline: 0845 077 2290 or 0207 713 7486 from a mobile
Email: info@arc-uk.org
Website: www.arc-uk.org

Down's Syndrome Association (DSA)
Phone: 0333 1212 300
Email: info@downs-syndrome.org.uk
Website: www.downs-syndrome.org.uk

Support Organisation for Trisomy 13/18 (Patau's syndrome and Edwards' syndrome (SOFT UK)
Email: enquiries@soft.org.uk
Website: www.soft.org.uk