Understanding Chromosome & Gene Disorders

Do you have 22q11 deletion syndrome?

Are you planning to have a baby? Do you want to find out if the pregnancy has 22q11?

rarechromo.org

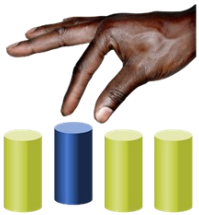
This Easy Read leaflet is for people with **22q11 deletion syndrome** who are thinking about having a baby.

22q11 deletion syndrome or **DiGeorge syndrome** is a condition you may have from birth.

22q11 deletion syndrome can cause a range of lifelong issues, like heart problems and learning difficulties.

22q11 deletion syndrome is also called **Velo-cardio-facial syndrome** or **VCFS**.

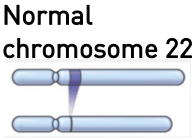
If you read this leaflet it will:



- help you to **choose** what to do

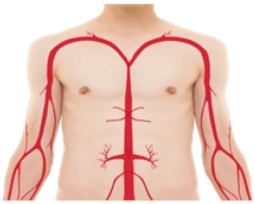
- give you information about testing for **22q11 deletion syndrome** in pregnancy

If you have **22q11 deletion syndrome** and are thinking about having a baby, **a test** could help you.

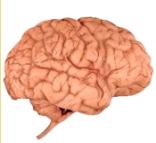


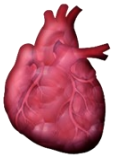
22q11 deletion syndrome happens when a piece of **chromosome number 22** is missing.

Everyone has **chromosomes** in their body.



Chromosomes tell our body how to work.



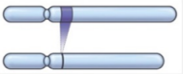


The missing piece of **chromosome 22** tells the brain and heart how to work.

The missing piece of **chromosome 22** explains some of the issues that people with **22q11 deletion syndrome** have.

So, if you have a child, there is a high chance that they have:

Normal chromosome 22



Chromosome 22 with q11.2 deleted

- **chromosome 22** that has not changed
- **chromosome 22** with a piece missing

The chances of having an affected child is **50%** (**1 in 2**) for **each** pregnancy.

Every person with **22q11 deletion syndrome** is different and has different issues.

If your child has **22q11 deletion syndrome**:

- they may have more serious medical problems than you do
- they may only have mild medical problems

Doctors **cannot** tell you what these problems might be.

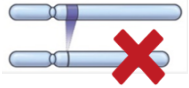
But it is your choice if you want to have a baby.

22q11 deletion syndrome when you are pregnant

Many people have **22q11 deletion syndrome** because their Mum or Dad has it too.

It may not worry you if your child has **22q11 deletion syndrome**.

Normal chromosome 22



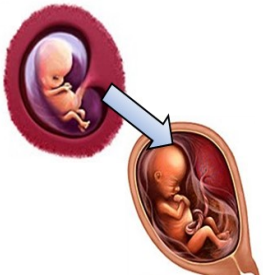
Chromosome 22 with q11.2 deleted

Or you might not want your child to have **22q11 deletion syndrome**.

If you want to find out how **testing** could help you, ask your GP to make a hospital appointment.

Testing and pregnancy

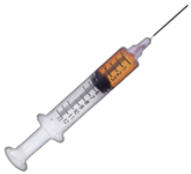
When a baby grows in the womb, it gets protected by water called **amniotic fluid**.



The **amniotic fluid** helps to protect the baby as it grows in the mother's womb.

Tests can be carried out early in the pregnancy when the baby is very small.

You can find out if the baby has **22q11 deletion syndrome** by testing some of the **amniotic fluid** around it.



A **needle** is put into the mother's stomach to take some **amniotic fluid** from around the baby.

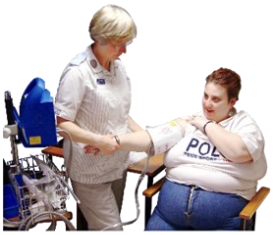
The mother takes **medicine** so that she does not feel the needle when it goes into her belly.

Then the amniotic fluid is tested to see if the baby has **22q11 deletion syndrome**.

There is a small risk that you could **lose your baby** when you have this test.

If the baby has **22q11 deletion syndrome,** you could choose to **end the pregnancy**.

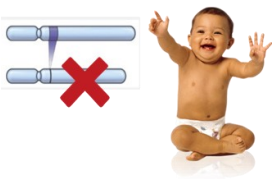
When you end a pregnancy, this is called **abortion** or **termination**.



You could also choose to **continue the pregnancy** and have more regular checks.

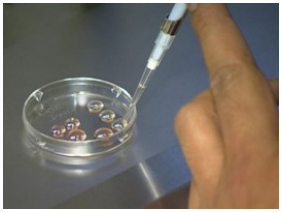
To have an abortion, the mother must take **medicine** or have an operation.

Tests before you have a baby

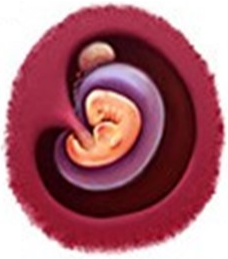


This part of the leaflet tells you what you can do to stop your baby from getting **22q11 deletion syndrome**.

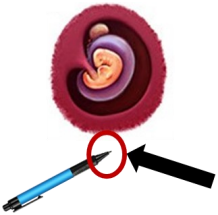
You start by taking an **egg** from the mother and a **sperm** from the father.



Then the **sperm** and **egg** are put together in a laboratory to make an **embryo.**

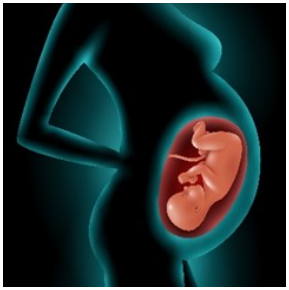


An **embryo** is the first **8** weeks of a baby. It does not look human at this stage.



A baby starts as an **embryo** and it is very small, about the same size as the tip of a **pen**.

We can test the **embryo** for **22q11 deletion syndrome**.



If the embryo does not have **22q11 deletion syndrome,** it is put in the mother's womb to grow.

This test is called **Preimplantation Blood Diagnosis** or **PGD**.

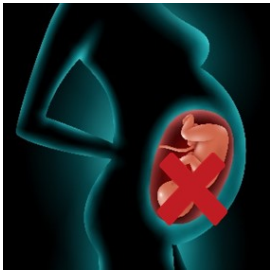
Some people with **22q11 deletion syndrome** have used **PGD** to have a child that does not have **22q11 deletion syndrome**.

It is important to know that the **PGD test** can make the mother feel ill.

Some **medicines** that help the mother to produce an egg may make them **feel ill**.

Your GP can tell you what is good and what is not so good about the **PGD test**.

If you have a **PGD test,** it might not work.



Not all embryos put into the womb will grow into a baby.

Also, you may need to have **more than 1 PGD test** before it works.

The hospital pays for the **PGD test**.

You **cannot** have **a PGD test** if you:

- already have a child that does not have **22q11 deletion syndrome**

- **smoke**

- are **overweight**

It can take a long time to get **PGD treatment.**

Do you want to find out more about PGD?

First, you must ask your GP to make an appointment to see a **specialist doctor** at a hospital about **PGD**.

The **specialist doctor** is called a **Clinical Geneticist**.

You should make the appointment about **1 year** before you want to have a baby.

If you talk to your GP about **PGD**, you **do not** need to have the **PGD test** when you have a baby.

It is **your** choice.

Inform Network Support

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