PATHOLOGY TESTS EXPLAINED

Information about pathology tests to help everyone take control of their health and make the right decisions about their care.

WHAT YOU SHOULD KNOW ABOUT **REPRODUCTIVE CARRIER SCREENING**

If you're thinking of starting a family or are in the early stages of pregnancy, reproductive carrier screening is a way to find out if you and your partner could be carrying a genetic alteration that puts you at risk of having a child with a genetic condition like cystic fibrosis, spina bifida, and more.

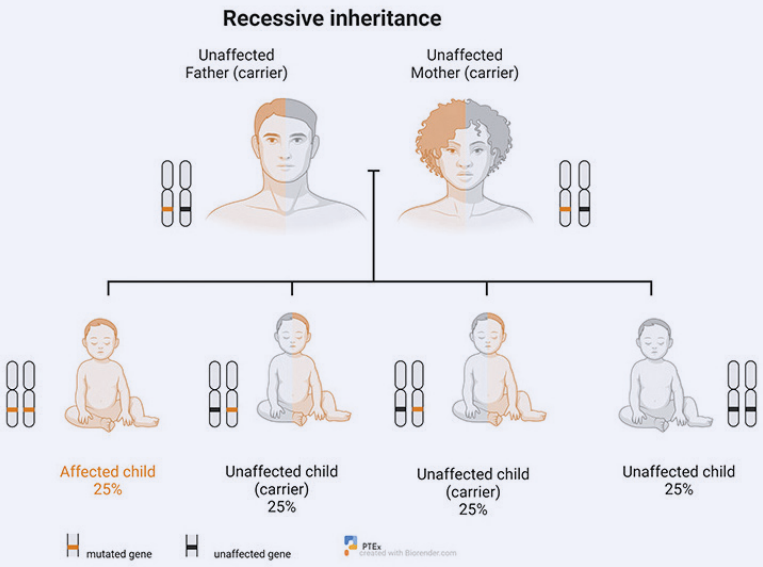
Chromosomes

How we inherit our genes

Everyone has two sets of genes. One inherited from their mother and one from their father. These genes in the form of DNA are packaged up in chromosomes which are found in most of the cells in our bodies. Humans have 23 pairs of chromosomes – one set comes from the father and one from the mother.

What is a carrier?

Because we have two sets of genes, one from each parent, we often don't know if we have a faulty gene in one of the pairs because the other healthy gene can do the job. If you're a carrier, it means you have a genetic mutation on one of the pairs that is known to be associated with a particular disorder, even though you do not have the condition yourself.



How we inherit our genes (continued)

What is a recessive condition?

Many genetic disorders need both parents to be carriers of mutations in the same gene for their child to be at risk of developing a condition. This is called a recessive condition. If two people carry changes in the same gene, they have a one in four chance with each pregnancy of having a child affected by that condition.

What is an X-linked condition?

Things are a little bit different for X-linked conditions. They will usually only appear in boys.

Our 23 sets of genes are made up of:

- 22 pairs called autosomes.
- 1 pair of sex chromosomes.

Typically, males have one X and one Y sex chromosome (XY) while females have two X sex chromosomes (XX). This means:

- A man can contribute an X or Y sex chromosome to his children.
- A woman can only pass on an X chromosome.
- Looking at it the other way, the Y chromosome can only be passed down from father to son.

Females have two X chromosomes so any faulty gene may be carried without her knowing. If she is carrying a mutation in one gene, with a normal gene on the other X chromosome, she is usually unaffected. However, any boys she has must inherit one of her X chromosomes and the Y chromosome from dad. If the boy inherits the X chromosome with the genetic change, he will inherit the genetic condition. This means the mother has a one in four chance of having an affected boy.

Types of testing

Three-gene screening (CF, SMA, Fragile-X)

Although many genetic conditions can be screened for, most testing looks at three:

- Cystic fibrosis (CF)
- Spinal Muscular Atrophy (SMA)
- Fragile X syndrome (FXS)

Carrier screening gene panels

Carrier screening panels test for a wider range of conditions. Because many more genes are screened there is a very high chance of one partner being a carrier for one or more of these conditions. For this reason, this type of testing is usually performed on both partners.

Specific screening tests

People from some specific ethnic groups are more likely to carry certain genetic mutations than others. In this case, a doctor may recommend several other screenings.

Getting your results

If your results show you have genetic variants that put your baby at risk, you will be referred to a genetic counsellor. About 1 in 20 couples will both be carriers of the same condition.

A negative test does not mean there is no chance of having a baby with a rare disorder. However, having a negative result can show that the likelihood is greatly reduced.

Questions to ask your doctor

Why should I do this test?
Which conditions are detected, and which are not?
What will the results tell me?
Who will help me understand the results?
What will happen if my partner and I are carriers?
What does it mean if the test is negative?

For more detailed information on these and many other tests go to **pathologytestsexplained.org.au**

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www.pathologytestsexplained.org.au

Please use this QR code to access more information

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My Health Record

You'll find a direct link to the Pathology Tests Explained website embedded in the pathology results pages of your My Health Record.

Click on the link to find information about what your tests are investigating or measuring and what your results can tell your doctor.