eugene

PRE-PREGNANCY CARRIER SCREENING

What is it, how does it work and should future parents consider it?

For more info, visit www.eugenelabs.com/carrier

What is genetic carrier screening?

Pre-pregnancy carrier screening is like a checkup for your genes — it involves testing healthy adults to see if either parent carries a genetic variation that could cause a serious inherited disorder in their child. Some of the more common disorders screened for include Cystic Fibrosis, Spinal Muscular Atrophy, Thalassemia, and Tay-Sachs disease, but there are more than 100 others that can be tested for.

Most conditions are “recessive,” which means:

1. A person can be a healthy carrier — and show no signs of the disease, but still have the ability to pass it on to their children.
2. Most people carry at least one recessive genetic condition.
3. Both parents must be carriers to have a child affected by the disease.
4. Often there are no clues in the family to suggest a risk.
5. Examples include Cystic Fibrosis & Tay-Sachs disease.

Some conditions are “sex-linked,” which means:

1. Only one parent needs to be a carrier to have a child affected by the disease.
2. Here too a person can be a healthy carrier and have the ability to pass it on to their children.
3. The child’s gender influences the health risk.
4. Examples include FragileX syndrome & Duchenne Muscular Dystrophy.

This test clarifies a couple’s risk of passing on an inherited genetic disease to their child and can better inform their planned pregnancy choices.

Many genetic conditions are rare, but a recent study found that 1 in 4 people tested were carriers of at least one variant. The average risk of having a child with one of these diseases is higher than that of having a child with Down syndrome . What's more, these conditions will not be detected by routine prenatal tests. Also, CVS and amniocentesis tests can only identify risk when you have had a carrier screening first.

One in 25 Australians carry the Cystic Fibrosis gene.
That’s one kid in every class.

1. Plantinga, M, etal (2016) Population Based Preconception Carrier Screening, European Journal of Human Genetics

How does it work?

Every human has about 23,000 genes. These genes are filed on structures called chromosomes. Most of us have 23 pairs of chromosomes. For each pair, we inherit one copy from our biological mother and one copy from our biological father.

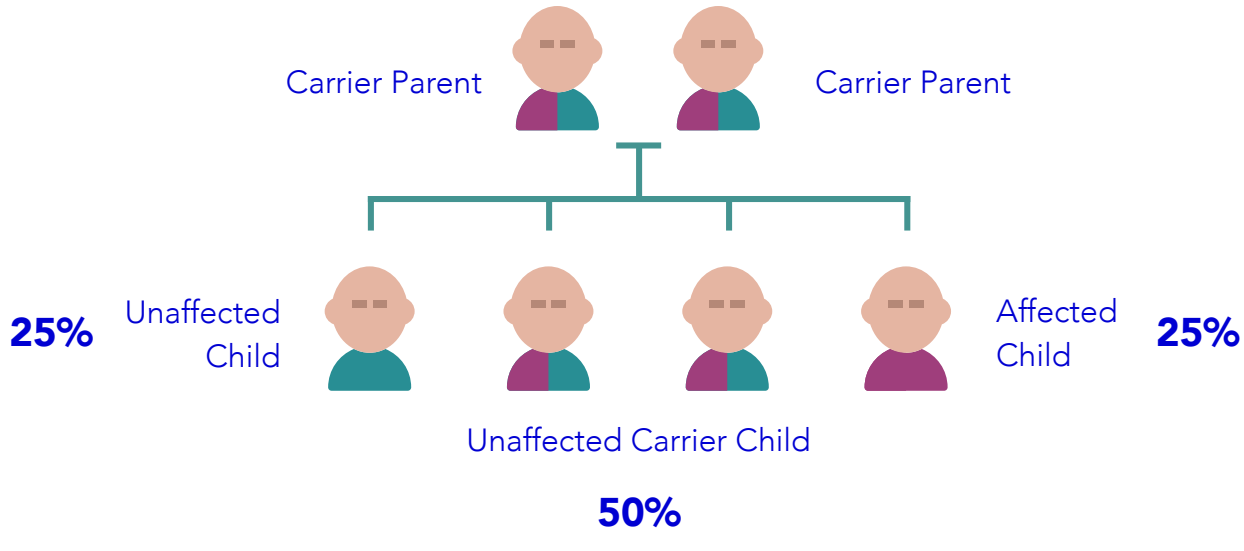
This is how parents pass on genes that can influence their child's physical characteristics like their hair color or the shape of their nose. In the same way, parents also pass on genes that influence their child's health and development. Sometimes these genes cause inherited diseases, even if the parents themselves don't have any symptoms or the disease.

It's important to stress that all of this is completely normal. We're all different and we all carry these gene variations, we just don't know about them. Recent advances in genetic technologies means that it is becoming easier to find out more about these risks and the possibility of passing on inherited conditions to our children. When we know about these risks, it also enables us to take action and make informed choices that help reduce the risk itself, and if not reduce it, be prepared for it.

Recessive Conditions

For recessive conditions, if a person has one gene (or half of a pair), they are a *carrier*, meaning — they are healthy because they also have a working copy of the gene. But, they can still pass their non-working copy to their child.

If the other parent also happens to be a carrier of the same gene, there is a 25% (1 in 4) chance that they *both* pass this gene to their child — and as such, have a child affected by the disease.



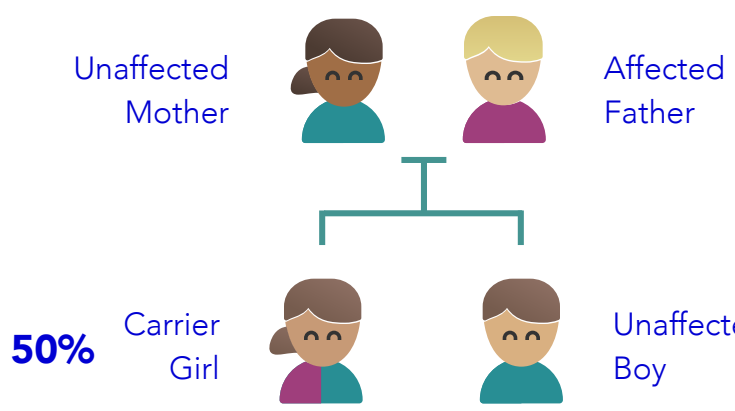
That's why it's important to test both parents for these genes — because people can be carriers and remain unaffected. 90% of children that have a recessive genetic disease had no previous family history of it, which feels completely out of the blue for the parents.

If both parents are carriers, there's a one in four chance that their children could develop symptoms.

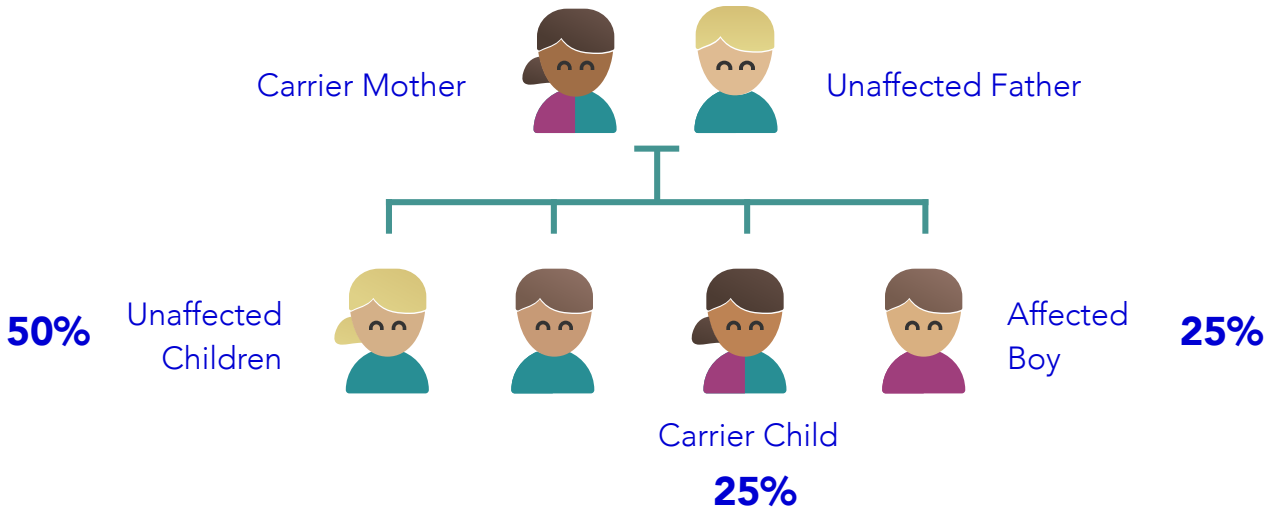
X-Linked Conditions (Also known as sex-linked conditions)

For x-linked conditions the sex of the parent who passes on the gene variation influences the risk for the children.

If the father is affected, all his daughters will be healthy carriers. Sons can only inherit these x-linked conditions from their mother and therefore will not be affected by the disease.



If the mother is a healthy carrier and they have a son, there is a 50% chance that they could be affected by the disease. If they have a daughter, there is a a 50% chance that they will be a healthy carrier (like her mother).



Another way of looking at this is that there is a 25% chance of having an affected son and a 75% chance that the child will not be affected personally by the disease.

Why do it?

The biggest benefit of carrier screening is that it can help future parents understand their risk so they can be ready and make informed decisions.

If neither partner are carriers, it provides reassurance and peace of mind that the risk of having a child with a genetic disease is low.

If a couple is found to be at risk of having a child with a genetic disease, we know that it can be complicated, confusing, and even scary. That's why we have trained genetic counsellors to help you consider your options and make empowered, informed choices that feel right for you. These choices can potentially reduce the risk of having a child affected by the disease, but also help parents be emotionally, mentally and practically prepared for it in a much more meaningful way.

Eugene's guided genetic screening can be done from the comfort of your own home — it's just a saliva test. Each member is also paired with a trained genetic counsellor who is there to support and guide you every step of the way and the entire experience is facilitated through the Eugene app.

Knowing the risk in advance can help families treat or even prevent the disease in the first place.

"Genetic carrier screening should be recommended to all prospective parents."

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists

Excellence in Women's Health

HUMAN GENETICS SOCIETY OF AUSTRALASIA

ACOG

THE AMERICAN CONGRESS OF OBSTETRICIANS AND GYNECOLOGISTS

The American College of Obstetricians and Gynecologists

WOMEN'S HEALTH CARE PHYSICIANS

Why Eugene?

We started Eugene to create the healthcare experience we'd want for ourselves and our loved ones. Our at-home guided genetic tests are simple, ethical, actionable and all facilitated through the Eugene app.

Every member is also paired with a genetic counsellor for mindful support and guidance every step of the way. We help you understand your results, and if necessary, help you consider their options and make choices that feel right for you. We can even provide a referral if appropriate.

Your results, reports, plans and appointments all sit in the app, and always in simple language. You can learn more at your pace and even request another call from your counsellor who can assist with your personalized care at every step.

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www.eugenelabs.com/carrier

Email your referral to counsellor@eugenelabs.com
or fax a referral to 03 9923 6320