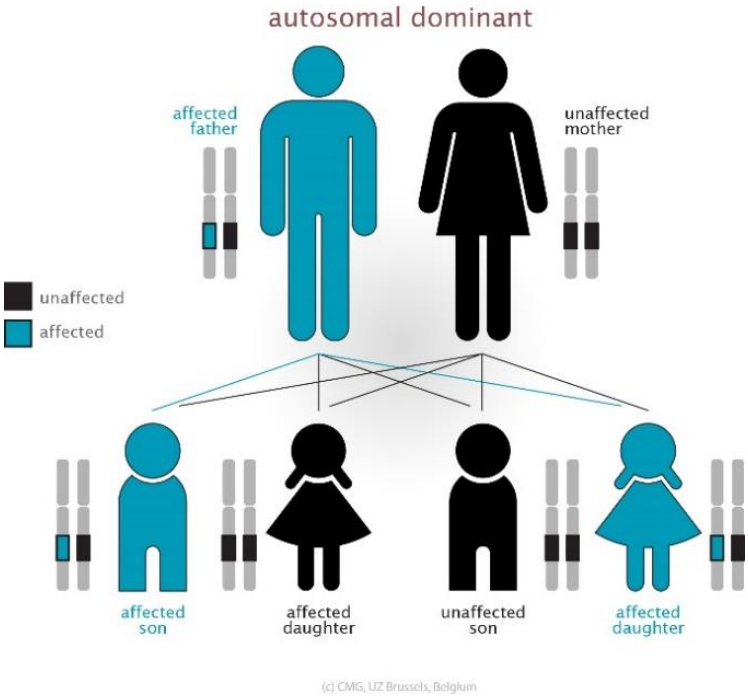
THE MATER HOSPITAL

Cardio Genetics Information

Inherited Cardiac Condition

You have been diagnosed with or are suspected of having an Inherited Cardiac Condition.
The condition you have may be passed through your genes from a parent to a son or daughter.
A gene is part of our DNA which contains a code for making a molecule (a protein).



Every person has two copies of each gene. Cardiac Genetic conditions are caused by a mutation in the genes that contain codes for molecules (proteins) in the heart.
A mutation in only **one** of the two copies of one of these genes (from the father or from the mother) is enough to develop some conditions.
This is called **Autosomal Dominant** and a parent who carries it has a 50% (1in 2) chance of passing the mutation to each child. The chance that a child will not inherit the mutated gene is also 50 percent.

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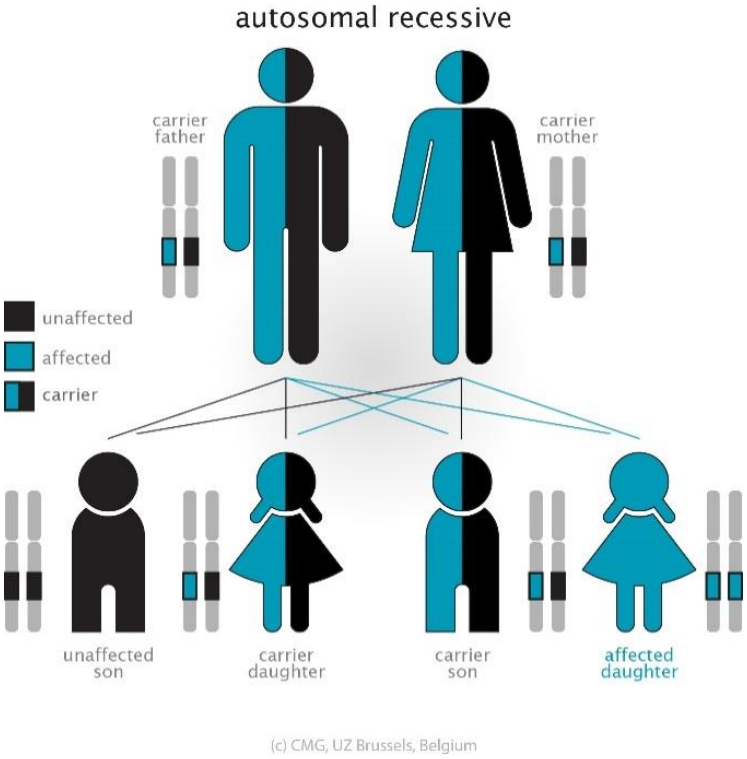

Sometimes, conditions can also be Autosomal Recessive. This means that you need mutations on both copies of a gene (from both father and mother) to develop the condition.

In recessive inheritance patterns, parents who are both carriers of a rare disorder, with no evidence of the condition, will have children with a 1 in 4 chance of not being a carrier, a 1 in 2 chance of being a carrier and a 1 in 4 chance of developing the condition which can be quite severe (autosomal recessive).

In some cases, a **New (de novo) Mutation** can occur in the egg or sperm cells or in an embryo. In these cases individuals do not inherit the genetic condition from a parent but rather it develops spontaneously in the individual and is absent in both parents.

Gene Variant Testing

Genes are made of a chemical called DNA.
DNA is a very long molecule, the components of which can line up in sequence creating the code responsible for production of proteins. We use specialised testing equipment called gene sequencers to read the code which we represent through a four-lettered labelling system (A, T, C and G).
An example of a gene sequence would be 'ATTCGAACGCTTACG'.
These genes are the building blocks of and are present in every cell in our body.

Changes in our gene sequence by insertion, deletion or duplication of one or more of the letters results in a 'variant'. Variants can cause inheritable conditions.

Gene variants are classified as:

Class	Type	Likelihood
Class V	Pathogenic	>99% likelihood it is responsible for the condition
Class IV	Likely Pathogenic	90-99% likelihood it is responsible for the condition
Class III	Variant of Unknown Significance (VUS)	10 – 90% likelihood it is responsible for the condition

Genetic testing has limitations and cannot tell us everything.

Not everybody however who inherits a gene variant develops the condition associated with that gene variant. This is called variable expression meaning that if two family members carry the same gene variant, it is possible that one may develop the associated condition and another may not most likely due to the influence of multiple other genes, lifestyle or other factors such sporting activity or high blood pressure which can make it more likely for people to develop some forms of cardiomyopathy or certain medications which make it more likely to develop long QT syndrome.

If genetic testing shows you have a DNA pathogenic variant (called a “positive result”), it does not always mean you have a genetic heart disease. A positive result might signal that you have an increased risk to develop a heart problem that you may never develop.
If genetic testing shows you do not have a DNA variant (called a “negative result”), that does not mean you will never develop a heart problem. Depending on your family history, you may still be at increased risk to develop a genetic heart problem.
If genetic testing shows you have a DNA variant of unknown significance (called a “VUS result”), that means your risk for an inherited heart disease is not yet known, but this could become clear over time as more is learned.

According to Irish law patients who are diagnosed with a genetic or inherited cardiac condition cannot be penalised on the basis of a genetic test result. Insurance companies, banks or mortgage providers are prevented by law from asking about or being influenced by genetic test results.

Women of child-bearing age with certain genetic conditions are advised to specifically meet with a Genetics Counsellor to discuss family planning options to explore options with in -vitro fertilisation or other techniques to reduce the risk of a genetic condition being passed onto a son or a daughter.

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