Long QT Syndrome

How is it passed down in families?

Information about the genetics of Long QT syndrome and genetic testing

Long QT syndrome is genetic

Long QT syndrome is a condition of the heart that can lead to abnormal heart beat patterns. Long QT can sometimes be due to other medical problems. It can also be passed on from generation to generation as a hereditary condition caused by a non-working gene. In most Gitxsan, Long QT is a problem caused by a gene.

What is a gene?

A gene is like a blueprint for our body. Genes give instructions that determine traits, such as blood type and height, but genes are also important in making sure the body stays healthy. We have thousands of genes. Some of these genes control heart beat patterns.

We have 2 copies of each of our genes

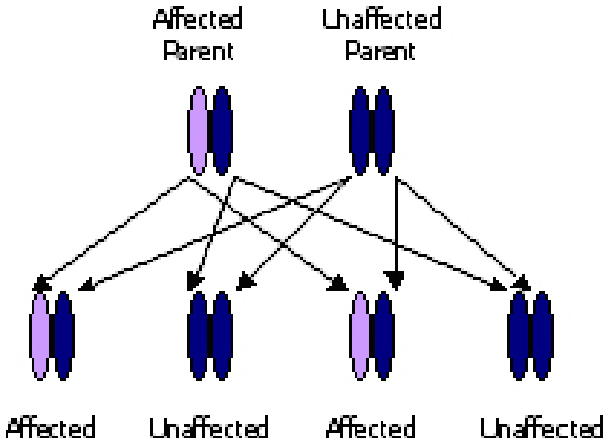
Our genes come in pairs. This is because we get one copy of a gene pair from our mother and the other copy from our father. Our eggs and sperm only have one copy of each gene. When we have children, we pass on only one copy of each gene, through our egg or sperm.

Long QT is caused by a gene change

Sometimes, one of the genes in a pair has a change which stops the gene from working properly. In a person with Long QT syndrome, one copy of the gene works normally, but the other has a change in it, making it non-working. This gene change causes the problem with the pattern of heart beats. This can lead to the problems of Long QT syndrome (fainting, dizziness and abnormal heart beats).

We pass genes to our children

When we have children, we pass on only one copy of each of the genes we carry. For a person with Long QT syndrome, this means that each child has a 50% (or 1 in 2) chance of inheriting the non-working gene causing Long QT syndrome. Each child also has a 50% chance of inheriting the working gene. The gene for Long QT syndrome affects men and women, boys and girls, just the same. If a person does not have the gene for Long QT, they cannot pass it to their children.



The same gene can have different effects

Even in the same family, where the same gene change is passed down from generation to generation, each individual may be affected differently. Some individuals may be healthy until old age, some may have fainting episodes, some may need to take medicine everyday, and some may need to have an implanted defibrillator (a device to keep the heart beating regularly). Because of this we can't predict how a child will be affected by looking at how the parent is affected. This is called *variable expressivity*.

Is there a gene test?

Gene changes can happen in families without causing the disease. So, we need to know first whether a gene change causes disease. If the gene change is known in a family and we can show that the change causes the gene to be non-working, other family members can have a blood test to see if they have the gene change. The blood test will not be able to tell how severely or mildly the person will be affected.

But: There are many different genes for Long QT

There are at least 12 different genes (and probably more which we do not know about yet) which can cause Long QT. Once the gene change causing Long QT syndrome in a family is found, it is easy to

test other family members for that gene change. But, if the gene change is not known in a family, it is difficult and expensive to find it. For some people, even with extensive testing, the gene change causing the condition will never be known.

When should the gene test be done?

A diagnosis of Long QT syndrome can often be made by a heart specialist. Sometimes the diagnosis is not easy to make and a gene test (if the gene change is known in the family) may be more accurate. But, some people are worried about gene testing. These are some of the things they might think about: Do I want to know if I have the gene change? Will it make a difference to my choice to have children? If I already have children, will it make me feel guilty? If I know that I do not have Long QT, will I feel even worse for family members who have it? There are a lot of things to think about before the test is done and also after the results are received. For this reason, it is important to talk to someone about the testing before deciding to have it (a genetic counsellor, your doctor, a health nurse or a community counsellor).

For more information, please contact:

- Dr. Laura Arbour, a researcher at UBC looking at genetic causes of Long QT in the Gitxsan, or Sarah McIntosh (genetic counsellor/research assistant) on our toll-free line: 1-888-853-8924
- Community research assistants, Gwen Weget-Simms at (250) 842-6295 or Julia Sundell at (250) 842-5234