Why can girls be carriers of Barth Syndrome? - As girls have two X chromosomes they will have two copies of the TAZ gene. So it is possible for a girl to have a spelling mistake in one copy of her TAZ gene which stops that copy of the gene working properly, but provided that the other copy of TAZ is normal this will act as a back-up and she won't develop the symptoms of Barth Syndrome. She is called a "carrier" of Barth Syndrome. She may have inherited the TAZ gene with the spelling mistake from either her Mum or her Dad or it may have occurred for the first time in her when she was made.

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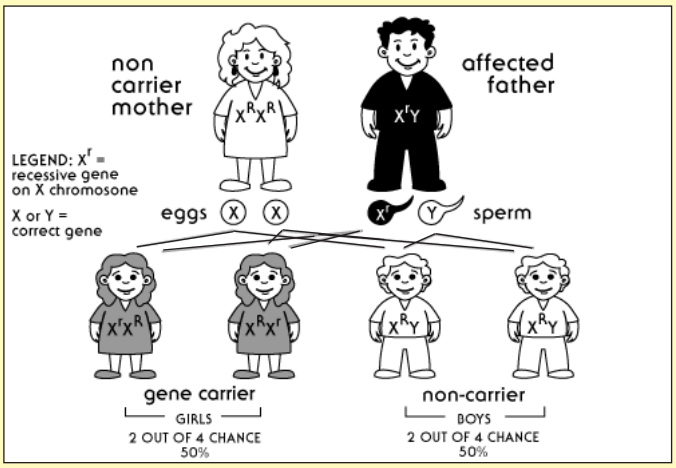
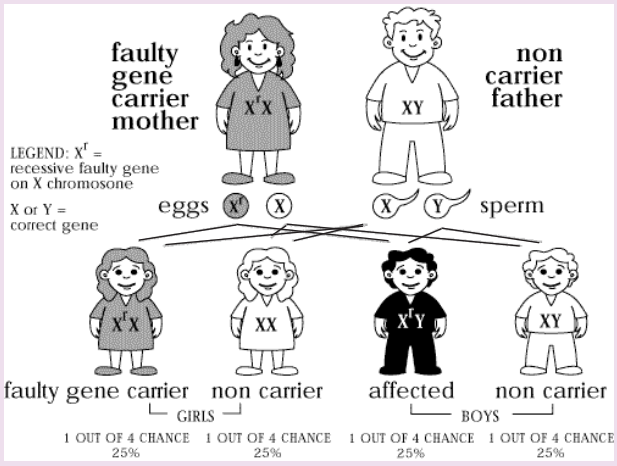
If I have a baby, will it have Barth Syndrome?

If a man who has Barth Syndrome has a child with a woman who is not a carrier of Barth Syndrome then there would not be an increased risk of any sons they have being affected by Barth Syndrome. However all of his daughters will be carriers of Barth Syndrome.

If a carrier female has a child there would be a 1 in 4 (25%) chance in each pregnancy that the baby will be a boy affected by Barth Syndrome. There will also be a 1 in 4 (25%) chance in each pregnancy that the baby will be a girl who is a carrier of Barth Syndrome.





Options when starting a family - If a couple want to find out more about their chances of having a boy with Barth Syndrome then we would recommend that they ask their GP for a referral to their local Clinical Genetics Service or they can ask to see the Genetic Counsellors at the National Barth Syndrome Service clinic in Bristol.

There are a number of options available to couples where there is a chance of having a child with Barth Syndrome. These would include having extra scans in the pregnancy, having a test during the pregnancy or having screening of embryos before they are put into the mother's womb. All the options are very personal choices and it can be helpful to have the chance to discuss these with a Genetic Counsellor.

Key facts on Genetics for Males with Barth Syndrome

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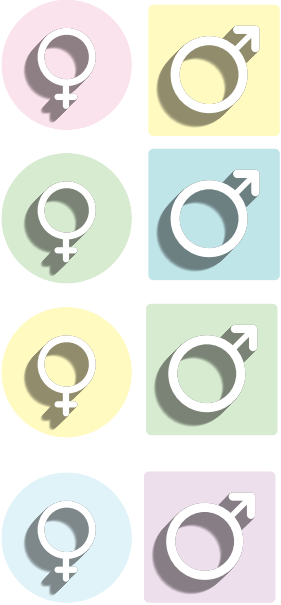
Chromosomes - Our bodies are made up of millions of cells. In the middle of each cell is a nucleus where our genetic material is stored. If you looked down a microscope at the nucleus you would see the chromosomes. Humans have 46 chromosomes which come in 23 pairs; we inherit one of each pair from our Mum and the other from our Dad. Chromosome pairs 1-22 are the same in boys and girls. The 23rd pair are the sex chromosomes - they determine whether a person is a boy or a girl. Girls have two X chromosomes (XX) and boys have an X and a Y chromosome (XY).



Genes - Each chromosome is made up of a substance called DNA. The DNA forms the genetic code and within the genetic code are individual pieces called genes. We have about 23,000 genes in total. A gene is like a recipe that our bodies can read to make proteins. Many different types of proteins are needed to help us grow and function properly. If there is an change (a bit like a spelling mistake) in the genetic code within a gene, this can affect how the protein is made and this can cause a genetic condition.

Why do some Boys get Barth Syndrome? - Barth Syndrome is caused by a spelling mistake in the TAZ gene. This gene is found on the X chromosome. All Boys only have one copy of the TAZ gene (because they only have one X chromosome), so if they have a spelling mistake in this gene that affects how the gene works, then they will have Barth Syndrome. They may have inherited the copy of the TAZ gene with the spelling mistake from their Mum or the spelling mistake may have occurred for the first time in the egg that made them.