

BARTH SYNDROME



Teenage booklet

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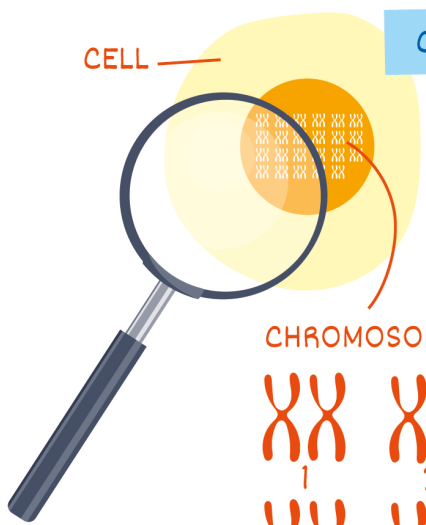


Hi! My name is Peter
and I have Barth syndrome.

This is a very rare, genetic condition that affects me in different ways. Don't worry, it's not at all contagious!

As you'll see from this booklet, there are quite a few things that I can't do because I have Barth syndrome but there are lots of things that I CAN do and quite a few things that I can join in, with a few adaptations.

Here is some specific information about how Barth syndrome affects me personally....



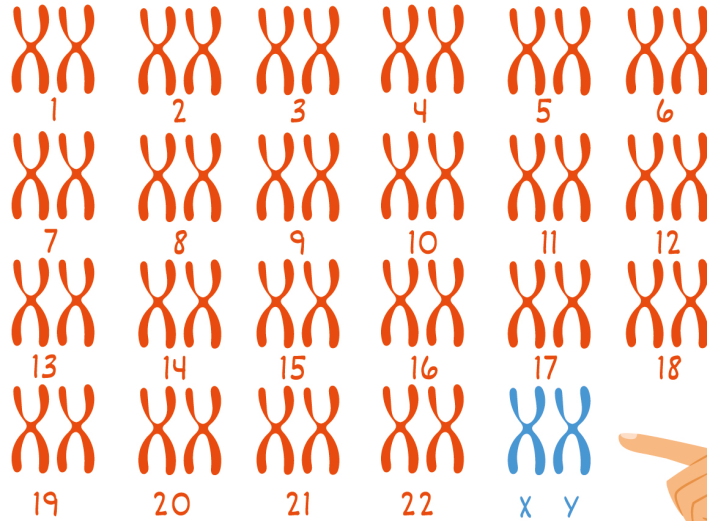
CHROMOSOMES AND GENES

The human body is made up of trillions of cells.

In each cell, there are 46 chromosomes:
men have 22 pairs of chromosomes, plus an X chromosome and a Y chromosome.

Women also have 22 pairs of chromosomes,
plus two X chromosomes.

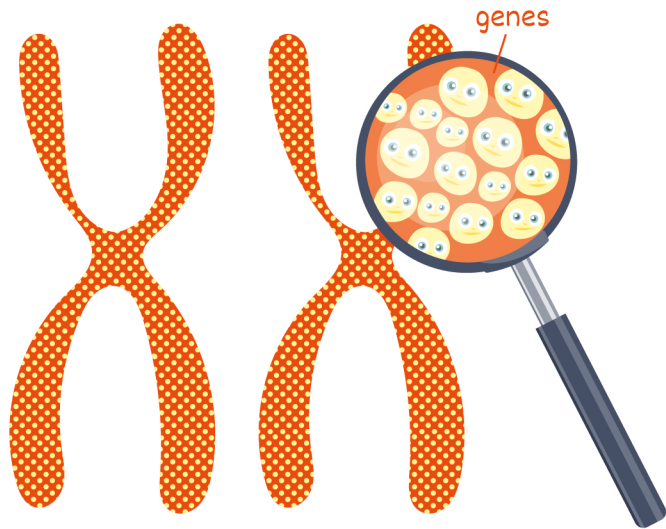
CHROMOSOMES



(I'm a boy)



BARTH SYNDROME IS A GENETIC CONDITION



This change affects all the cells in the body.

Our chromosomes are made up of DNA and this is where our genes are found. A gene is like a recipe to make our bodies work. Sometimes these genes have changes (called "variants") that can cause genetic conditions.



In Barth syndrome, the change is found on a gene (the TAZ gene) on the X chromosome.

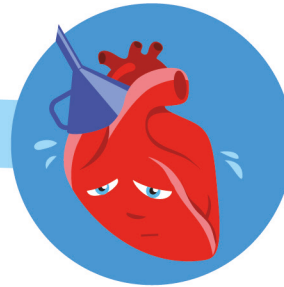
HOW DO YOU GET BARTH SYNDROME?

Barth syndrome can:

- be transmitted from a mother to her son (the mother is a carrier of the faulty gene, but does not have the condition) or
- occur for the first time in an egg or sperm that goes on to become a child (de novo mutation)



HOW BARTH SYNDROME AFFECTS MY HEART



Heart cells are particularly affected in Barth syndrome and this can be a very serious problem.

The heart is a pump that circulates blood through the body: when it doesn't work well, it can't pump efficiently. This can lead to cardiomyopathy (diseased heart muscle) and heart failure (where fluid starts to build up in the lungs, causing breathlessness).

Our hearts sometimes start to beat irregularly, too fast or too slowly. This is called arrhythmia. Many of us have a machine called a defibrillator. This can be used if we have an abnormal heart rhythm or cardiac arrest. The defibrillator gives the heart an electric shock to get it back into a normal rhythm again. Some of us even have an internal defibrillator implanted directly under the skin, close to the heart!

There is no cure for cardiomyopathy but treatment with medicines can help to control symptoms and stop it from getting worse. Sometimes the heart function gets so bad that the only thing that can be done is a heart transplant.

HOW BARTH SYNDROME AFFECTS MY BLOOD AND IMMUNE SYSTEM?



People with Barth syndrome often don't have enough neutrophils (white blood cells that protect the body from bacteria) so we are at danger from infection.

That's why we have to be careful and stay away from sick and contagious people. Also, getting sick can also put extra pressure on our hearts.

MUSCLE WEAKNESS AND FATIGUE

The TAZ gene also plays a role in the body's energy production. Our bodies produce less energy; that's why we can't be as active as we'd like. We get tired very quickly, even from everyday things like climbing stairs, running or even just eating and writing!

We are often very tired and forced to rest when we'd rather do the same things as our friends ...





DELAYED GROWTH

We're often significantly shorter on average during childhood and teenage years. Puberty usually occurs later than usual. The physical difference (small size, very young-looking face) can be difficult to live with when all we want is to look the same as our friends and do what they're doing.

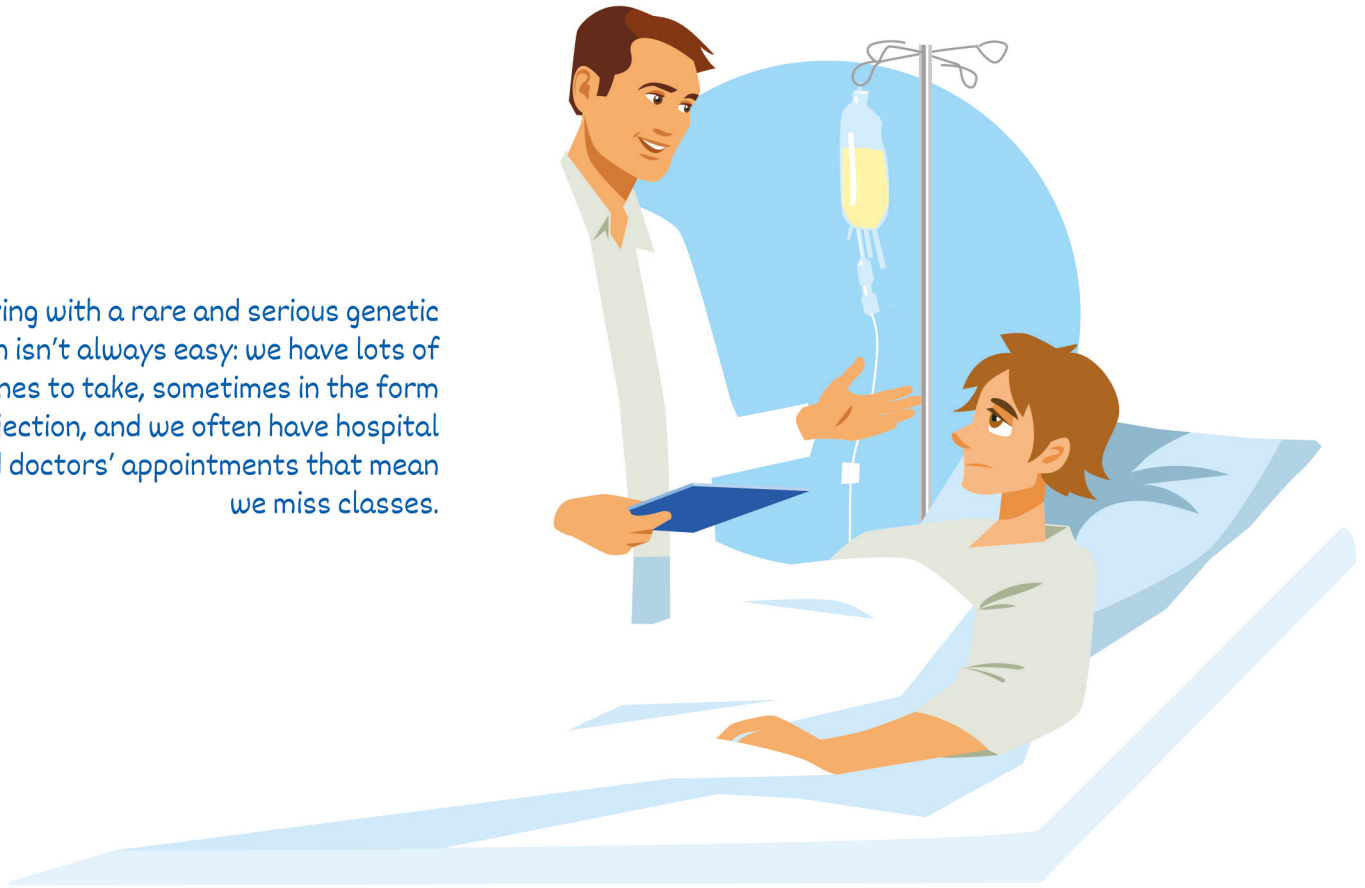
Luckily, many of us catch up on our growth in later teenage years and reach normal height in adulthood.

OTHER DAILY CHALLENGES

Other problems can also arise: pain, low blood sugars, eating problems ... Barth syndrome can mean different symptoms for different people, but it can also be different symptoms over one person's lifetime.



Living with a rare and serious genetic condition isn't always easy: we have lots of medicines to take, sometimes in the form of an injection, and we often have hospital and doctors' appointments that mean we miss classes.



FINDING BALANCE

We must constantly find a balance between our physical limitations and our social life, knowing that Barth syndrome is a serious illness, from which people still die.

Even if our health is stable, we have to be careful of a bacterial infection or a heart problem which can have immediate and dramatic consequences.





ONE IN A MILLION!

Barth syndrome is a very rare condition and we know that it's under diagnosed. Studies show that there should be approximately 4 000 cases in the world.

But we only know of fewer than 300 people. Doctors think it affects 1 in every million males. There are some rare cases where it affects girls too.



RESEARCH

Doctors and researchers are working hard to find medicines and solutions to help us live better and longer lives.

Medical research is coordinated and funded by the Barth Syndrome Foundation in the United States together with its affiliates in Canada, France, Italy and the United Kingdom.

Join us to help or to find more information:
www.barthsyndrome.org - USA and worldwide
www.barthsyndrome.ca - Canada
www.syndromedebarth.fr - France
www.barthitalia.org - Italy
www.barthsyndrome.org.uk - United Kingdom





I hope this booklet has helped you understand a bit more about my condition.
Thanks for taking the time to read it and I hope to see you soon!

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Illustrations : www.zoe-illustratrice.com