



GROWING
UP WITH

BARTH ♥ SYNDROME

GROWING UP WITH BARTH SYNDROME

We have created this booklet to support you in day to day living with Barth Syndrome. This includes information about Barth Syndrome, spotting when you are feeling unwell, knowing where to get medicines and knowing who to contact when you have questions. There are also top tips from young people and their siblings about making sure that Barth Syndrome does not rule your life.

WHAT IS BARTH SYNDROME?

Barth Syndrome is a rare genetic condition that affects males. The main symptoms include:

- Heart muscle weakness (cardiomyopathy)
- Neutropenia (lack of white blood cells needed to fight bacterial infections)
- Fatigue and general muscle weakness
- Growth issues



NEUTROPENIA?

Nine out of every ten males with Barth Syndrome get neutropenia at some time. Some young people are neutropenic some or all of the time and others only get this occasionally.

What is it?

The body produces neutrophils; these are a type of white blood cell that are important in fighting bacteria. Most people have lots of these (more than 1,500,000,000 in every litre of blood!) which the doctors write as 1.5. If you have less than 1.5 neutrophils this is called being neutropenic. Having less than 0.5 neutrophils is called “severe neutropenia”. This means that it is more difficult for your body to fight bacteria properly so you might need to go to hospital for a blood test and get given antibiotics. Neutropenia can also make you feel tired, brushing your teeth might hurt and you might get frequent or very painful mouth ulcers.

WHITE BLOOD CELL

WHAT IS G-CSF?

G-CSF is a type of protein that is called a growth factor. It is made in the bone marrow and stimulates the bone marrow to make

neutrophils. The bone marrow is the material inside the bones where blood cells are made. G-CSF is given by injection just under the skin by your parents or you when you are at home. If you tend to have frequent low neutrophil counts and these are causing frequent mouth ulcers or making you get bacterial infections, these G-CSF injections will usually make you feel much better. Most people need injections three times per week (often on Mondays, Wednesdays and Fridays) but some people need them only twice a week and others on alternate days. You need to have your blood checked regularly to make sure you are on the right dose. We will usually ask you to check your blood count either just before you have a dose of G-CSF (we call this a trough count) or on the day after your injection (we call this a peak count). This is to try to make sure that your neutrophil count isn't going either too low or too high.

TOP TIPS

Finger prick blood tests can be done at home. Here are a few top tips:

ABOUT BLOOD TEST

- 1 Wash your hands in warm water to clean them and warm them up.
- 2 Give your hands a good shake to get the blood to the end of your finger tips.
- 3 Avoid pricking the finger's tip, this part of the finger is very sensitive and it can hurt more. Aim for the side of your finger instead.
- 4 Once you've pricked your finger, rub a tiny amount of Vaseline from a clean cotton bud over the area you pricked. This will help the blood to fall in individual droplets, right into your vial/tube. Wait a couple of seconds before squeezing your finger.
- 5 Hold your hand down below your waist with the tube positioned to catch the drops as they fall. If you need to, you can gently squeeze your finger at the base (near the hand) and move towards the tip.
- 6 Shake the tube gently; there is a chemical on the sides of the sample tube that stops the sample from clotting. The blood needs to mix with this, otherwise it will clot and the sample will be of no use.
- 7 Try not to use the same place for your injections or blood tests each time. Some boys choose to give their G-CSF around their tummy button like the numbers on a clock, so that they are using a different place each time.

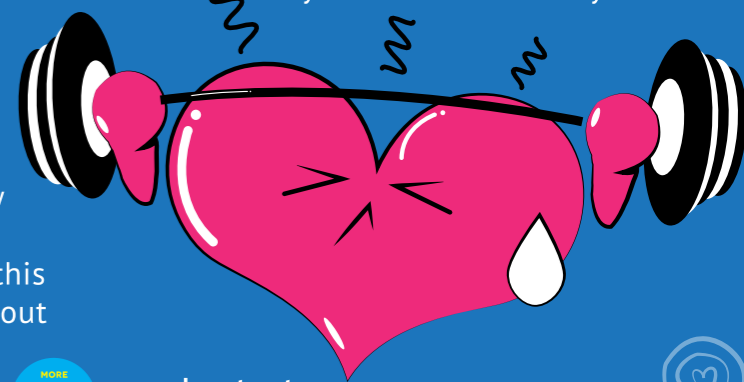


CARDIOMYOPATHY

The term cardiomyopathy means that the heart does not work as well as it should, this can be due to many reasons but usually is

because the heart becomes too big or the muscle is too thick. This can mean that the heart finds it harder to pump blood around the body.

You cannot catch cardiomyopathy from somebody else. In many cases we can help your heart pump better by giving you medications but sometimes this doesn't work well and we will then talk to you about other treatments.



MORE
INFORMATION
ABOUT HEART
CONDITIONS
CAN BE FOUND
AT:

www.yheart.net

www.cardiomyopathy.org

WHY

IT RUNS IN FAMILIES?

Everyone's body is made up of millions of cells. Inside these cells are tiny things called genes. The genes tell cells what to do. Genes are passed down from parents to children and some can cause Barth Syndrome. The active gene only gets passed to boys but girls can carry the gene without knowing as they won't have any of the signs of Barth Syndrome (they would be called carriers). This means that a carrier woman may have a boy with Barth Syndrome. A man who has Barth Syndrome himself and who has children of his own will always have either healthy boys without Barth Syndrome or carrier girls.

You can chat to a genetic counsellor if you want to talk about having children or find out more about how Barth Syndrome can be passed down in families.



GETTING ON WITH LIFE

If you have Barth Syndrome you can usually do most of the things that other young people may do, but you may not be able to do them as often or for as long. There may be times

when you feel fed up with having Barth Syndrome. Writing down in a diary what your worries are and letting others know what is getting you down can sometimes help. You can also talk to your parents, someone in the Barth Syndrome Trust or a psychologist, nurse or doctor. This may help you cope with your worries or answer your questions.

You may find that you have made some good friends with other young people with Barth Syndrome.



OTHER YOUNG PEOPLE

WITH BARTH SYNDROME HAVE SAID:

'Don't let it get in the way of your life'

'You will find you have amazing friends'

'Don't sit on the couch'

'Live your life, don't climb a mountain everyday but you can do it once if you want to'

'Its never really stopped me'



HEALTHY EATING ?

Healthy Eating means eating the right amount of those foods your body needs for energy, to grow and to repair. Including a wide variety of foods will help you to get the vitamins and minerals you need.

Carbohydrates—such as bread, potatoes, rice and pasta. Choose wholegrain when possible. This type of food will give you energy.

Protein—include one of these at each meal: meat, fish, eggs, beans, and pulses (e.g. lentils). Don't forget protein at breakfast too, try a low fat yoghurt smoothie or egg on toast. Make sure you drink the milk on your cereal as this also counts. These foods help build your muscles.

Dairy—such as milk, yoghurt and cheese. These foods make great snacks as they contain energy, protein and calcium to help build strong bones. Keep an eye on portion size.

Fruit and vegetables - You should try and eat five portions every day. Try to vary the fruit and veg that you eat – different types, colours, raw or cooked are all good. These are good low fat snacks too.



You should eat only a small amount of foods and drinks high in sugar or fat (like crisps, chocolate or fizzy drinks). Too many of these foods can make you quickly gain weight.



MY BROTHER HAS BARTH SYNDROME



You may feel lots of things about having a brother with Barth Syndrome. It may be annoying or scary and at other times fun. You could try talking to someone about what it is like for you.

OTHER YOUNG PEOPLE WHO HAVE A BROTHER WITH BARTH SYNDROME HAVE SAID:

'Try not to get angry with your brother but tell someone if you are'

'It's good to meet other young people - go to the Barth Syndrome Trust days'

'Stay away from your brother if you both feel upset and later when you're calm try telling someone about what things make you feel cross, upset or scared.'



FREQUENTLY ASKED QUESTIONS

Why am I shorter and have less energy than everyone else?

Barth Syndrome means that you grow more slowly than most other boys and you also start puberty later. You may do most of your growing after you have left school.

Doctors can sometimes look at your growth by looking at how many little bones have formed in your wrist by taking an X-ray. They call this a "bone age" test. It is one way of knowing how many extra years of growth you have. For instance, if you have an X-ray at 15 years but your X-ray looks like the wrist of someone of 12 years of age, then the doctors know that you should have three years of extra growth compared to other boys. If you and your doctors become worried about your growth they may do a growth hormone test to see whether your growth could be speeded up by giving growth hormone injections (but this doesn't happen very often).

Your muscles aren't as strong as many boys, particularly the muscles at the tops of your legs. This can make it more difficult to run or to kick a football hard and you might get tired much more easily, making it difficult to keep up with your classmates. Barth Syndrome makes it harder for your muscle cells to make energy, especially in large muscles like those at the tops of your legs and in your heart. It's important to tell your teachers and those around you about your condition so that they understand if you have to take things at your own pace.



Should I tell the people at work, school or college?

Yes, we would always advise you to tell the people that you work with so that they can help you if you need it. They are almost certain to be more understanding about things such as your

energy levels or tasks that you may find more difficult if they know about your condition. You may also need their help, for instance if you developed a problem with your heart rhythm. It's easy to feel that you are the odd one out and that everyone else is well, but in fact most people will have been to see their doctor in the past or be seeing their doctor regularly because of a medical condition you might not know about.

If I am on holiday and I feel ill what should I do?

Before you go on holiday you should always ask for a letter from your local

hospital which will contain medical information you need including your medicines. This letter will also contain details of who to contact if you become unwell and need to go to see a doctor. If you do feel ill you should always go straight to hospital and explain about your condition and the fact that it can cause a low neutrophil count and infections.



What will happen as I grow older?

If you have cardiomyopathy you may need to continue to take medications to help your heart cope. If your heart recovers you may not need to take these. It is always possible for your heart to develop a funny beat and this can make you feel dizzy or faint. It is really important that you tell your doctor about this so that they can check your heart and see if you need any extra help.

Neutropenia or the problems that can go with it such as mouth ulcers can get better, stay the same or even get worse as you get older. This means that sometimes people need to start G-CSF when they are older.

Having Barth Syndrome may mean that you find it harder to cope with bacteria and viruses and so might miss more school or work but it does not mean that you cannot do many things that other young people do – you can still go to university, work and be in a relationship with someone. You might want to get married or have children too. Having Barth Syndrome means that you might need to be a bit more careful and make sure that you look after your health but does not mean that you cannot have a full and fun life!



COME AND
TALK TO US
ABOUT WHAT
YOU NEED
TO KNOW:

Barth Syndrome Trust
www.barthsyndrome.org.uk

Barth Syndrome Service
www.uhbristol.nhs.uk/barthsyndromeservice



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