



Barth Syndrome Education Guide

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This booklet is produced by
the Barth Syndrome Trust
in conjunction with
the NHS Barth Syndrome Service.

Introduction

**For emergency care please refer to the Care Plan provided by parents.
See Appendix B for suggested template.**

This booklet is for parents and teachers. It provides information and guidance about the needs of boys with Barth syndrome in the school environment. We have included tips and practical advice from parents, teachers and clinicians to help you support your child/pupil so that he is able to participate in school as much as possible and achieve his full potential.

Boys with Barth syndrome have very different needs so it is essential to talk to your child about what he feels would help him. We recommend that parents keep in close contact with the school and regularly assess their child to ensure that his needs are met. We advise parents to complete an individualised Care Plan for each affected child as this will contain specific advice about a particular child's needs (See Appendix B).

Barth syndrome is a rare condition affecting mainly males, approximately 1 in 300,000 births. It affects the heart, immune system, muscles and growth.

You may find that a child with Barth syndrome has:

- ♥ Difficulties concentrating on school work and tasks in the classroom
- ♥ Difficulties in handwriting
- ♥ Muscle weakness which may lead to tiredness affecting his ability to join in classroom and physical education activities
- ♥ Neutropenia – a reduction in neutrophils, a type of white blood cell important for fighting bacterial infection. At these times young people may be more prone to infection.
- ♥ Heart failure, heart rhythm problems or a heart transplant
- ♥ Fatigue – can be variable and occasionally severe
- ♥ Growth delay - most young people of school age will be below average in height and weight.

The cruellest irony about Barth syndrome is how deceptively healthy those who have it may appear. A casual observer would never appreciate them to have such a devastating illness. ~

*Peter Barth, MD, PhD,
Paediatric Neurology (rtd),
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Academic Medical Centre,
Amsterdam*

What support can you have in playgroup or school?

An overview

Why do some children with Barth syndrome need statements around their physical and or learning needs?

The term 'special educational needs' or "SEN" has a legal definition. Children with special educational needs all have learning difficulties or disabilities that make it harder for them to learn than most of the children of the same age. These children may need extra or different help from that given to other children of the same age.

If you think that your child may have a special educational need that hasn't been identified, you should talk to the Special Educational Needs Coordinator (SENCO) at school, class teacher or Early Years teacher.

The Special Educational Needs Code of Practice gives guidance to help identify, assess and provide help for children with special educational needs. It sets out the process and procedures that all these organisations must follow to meet the needs of the child.

The child's teacher or SENCO may ask for advice from people outside the school to form an "Early Years Action Plus" or "School Action Plus", and careful records will be kept on the child's progress, with parents included in any discussions.

The law states that all state schools must do their best to see that special help is provided for all children with SEN. Most children's needs can be met by their mainstream school, sometimes with the help of a few outside specialists. In a few cases, the local authority (LA) will have to make an assessment of a child's educational needs, based on specialist advice. If the LA then decides that the child needs special help, they must write a statement of special educational needs - usually called 'a statement'. This describes all the child's needs and all the special help required. The child's ordinary school can usually provide this help with support from the LA.

The child's school or early years educational setting can ask the LA to carry out a statutory assessment, with communication with the parent,

Or

If a parent feels that their child is not making sufficient progress, they can ask the LA to carry out a statutory assessment. The process has a formal schedule and timings are dictated; the whole process should not take longer than six months.

Visit www.education.gov.uk where you can download “Special Educational Needs (SEN) – A guide for parents and carers” and

Contact a Family who have a wide range of excellent resources e.g.
<http://www.cafamily.org.uk/pdfs/educatio.pdf>

Physical Challenges

NB: See Care Plan (Appendix B)

Physical challenges faced by boys with Barth syndrome can include:

- Fine motor skills (the co-ordination between eyes, hands and fingers)
- Gross motor skills (the activities done by the arms, legs and large muscle groups)
- Exhaustion which is far more severe than the usual tiredness felt by healthy people
- Difficulty with handwriting
- Physical discomfort when sitting on the floor without support – this can often lead to fidgeting or adopting unusual positions to compensate
- Absences which can be as much as 50% due to illness, fatigue or hospital visits

Physical and medical safety

A child's physical safety is the most important factor needing attention when he is at school. Things to think about include:

Training

School staff should have regular training on:

- Resuscitation (and defibrillation if the child has an Automated External Defibrillator or AED)
- Symptoms and warning signs (see Appendix B for Care Plan)

Procedures

- Develop a clear procedure in the event of an emergency and ensure that it is known and accessible to all staff
- Advise parents when a contagious illness is spreading through the classroom or school

Other procedures

An assessment by a physiotherapist, occupational therapist and/or speech and language therapist may be required, with a programme which is implemented regularly at school.

Physical mobility and adaptations in the class room

An adapted curriculum might be required to provide time to rest.

A buggy or wheelchair might be needed for school trips

Simple things can make life at school easier, for example:

- a quiet place to work away from excessive distractions or stimuli
- extra time to complete a task
- a comfortable quiet place to rest when tired
- an angled writing table or board
- appropriate grip for pens and pencils

- easy grip or self-opening scissors
- hand putty exercises or something similar to strengthen hands early use of laptops and/or scribing assistance
- a flash drive so that work can be completed at home on the computer and then printed off at school
- an adapted toilet with hand rails or a foot box

- a specialist chair with arm rests like a Brookfield chair with a foot rest to ensure a stable base of support
- help with tying laces, buttons and zips
- help with eating (see separate section below)
- regular rests, stretches or breaks during longer sessions

Secondary School

In secondary schools think about:

- timetabling lessons in class rooms with close proximity
- being allowed to get up and move during long lessons
- using chairs, footstools and high science stools that have backs and/or armrests for to give support
- lockers at either end of the school
- a separate set of text books at home

- online resources such as Studywiz to post any catch-up work for easy access from home
- a lightweight notebook or laptop and memory stick
- an electric wheelchair for larger campuses or having someone to carry heavy bags and books
- creative timetabling with disapplication from certain non-core subjects if the work load becomes too heavy

- extra time for exams
- scribing
- being able to leave before the rush in the corridor
- finding out which time of day is best for the young person as some young people are more productive in the afternoons than in the mornings.

Physiotherapy and occupational therapy at school

Participation in PE with peers is important; this can be adapted for all, for example inclusion with the whole class for stretches and balance exercises during warm up which are often the physiotherapy exercises children with Barth syndrome have to do. Having somewhere to rest during this time should also be provided.

Fatigue

Genuine fatigue or a reluctance to do the work?

One of the recurring problems faced by young people, their families and their teachers is how to accurately gauge the true level of fatigue. There is no easy answer; however please bear the following in mind:

- The fatigue is totally different from the tiredness felt by a healthy person
- It can come and go at any time of the day
- It is often accompanied by pallor
- Some students feel at their best in the afternoon or evening and may struggle in the morning when teachers might expect them to be at their best
- Sleep studies have shown documented sleep apnoea which suggests that a child is not sleeping well enough to feel refreshed
- Over-exertion can result in fatigue which may last for days afterwards
- Some boys find it difficult to self-regulate themselves to preserve energy
- Sometimes they feel no-one believes them when they say they're tired
- They might not always wish to draw attention to themselves
- And sometimes, like many other children, they might not always be quite as tired as they say!

Possible strategies during periods of fatigue

- Try encouraging him to have a rest, snack and/or drink.
- Break down the task into smaller, more manageable sections and ask the child to do one section to the best of his ability and the rest at a later stage.

- Do not make him do any extraneous work and offer assistance with things like cutting and pasting work into books, colouring etc so that energy can be conserved for the most important part of the task.
- The boys often need a quiet, comfortable space away from everyone to rest
- Send home any missed work by email as soon as possible so that it can be done as soon as the child feels better.

How much should school staff be doing for the child?

This is another difficult question. Occasionally children with Barth syndrome may save their energy for the more enjoyable activities and might ask adults to help them do tasks that they are actually able to do themselves. The school environment is a wonderful arena in which to slowly build independence and self-confidence by encouraging children to do as much as they are able to do. Learning to become more organised is a skill that will serve them well throughout their school and adult lives. Finding creative and sensitive ways in which to deal with these issues will undoubtedly be beneficial and ensuring that parents and young people are actively involved in these discussions and provide guidance as to what they can and cannot do is essential.

Supporting a child with eating and drinking

Like all children, boys with Barth syndrome need a variety of foods from the five food groups, to promote growth and well-being. However boys with Barth syndrome often have a different body size, body composition, growth rate and different physical activity levels to those of the same age group, therefore their nutritional needs are affected.

Some boys with Barth syndrome have additional difficulties with eating and need extra help and understanding. School can provide a positive reinforcement of good eating habits and help develop the diet of Barth boys.

Boys with Barth syndrome need:

Smaller portion sizes and smaller sized food

As their bodies and tummies are smaller, expect the amounts eaten to be appropriate to their physical size not age

Lower physical activity and slower growth means that the amount of energy (calories) required is less.

They often tire easily when eating and are slow at eating. Make sure that they aren't last in the queue at lunchtime and help to make sure they can open their food and drink.

Some find chewing difficult. For younger children cut up food well and choose softer meats where possible.

A good supply of protein

Boys have lower muscle stores, and they are encouraged to have some protein at each meal and snack to help build and protect their muscle. Good sources of protein are foods such as milk, cheese, yoghurt, meats, nuts, fish, beans and eggs.

Regular Snacks

Although they need less calories overall, a small snack between meals helps to keep their energy up and provides a regular supply of protein. Low fat yoghurt or a yoghurt drink, a small handful of nuts, a cereal bar with seeds and nuts and fruit or raw vegetables are all healthy snacks.

Salty food and sauces

We don't yet know why boys with Barth syndrome are attracted to very salty food and sauces, but it is common in boys all across the world. Some boys even eat raw salt or lick salt from snacks. Eating a diet high in salt is linked to developing high blood pressure and therefore UK general healthy eating guidelines suggest that salt in the diet should be reduced. However, at present there is no evidence to show that their higher salt diet is detrimental for Barth boys; furthermore adding salt to food or dipping food in salty and spicy sauces can help to expand the diet of those boys who eat very little or very few foods. A more liberal approach to salt in the diet is accepted for Barth boys although as many salty foods are high in calories and fat, parents are encouraged to keep intake of these foods at a sensible level.

Restricted diets are helped by eating with friends

Many boys have a lack of variety in their diet and their diets contain higher amounts of milk, cheese and bread and are lower in meat, fruit and vegetables. This in part is due to limited exposure to different foods at critical developmental times because of repeated illnesses when young, weaker oromotor skills and possibly periods of tube feeding. Boys can also have a pronounced gag reflex and find it physically difficult to try new foods and tastes. Eating with their friends can help desensitise them to new foods and increase their motivation to try new foods. A number of the boys' diets have been really improved from eating with friends at school, and gentle persuasion to try new foods with no pressure can be beneficial.

Using a knife and fork

Sometimes standard cutlery can be difficult to use. There are lots of different types of adapted cutlery that can be tried e.g. using a knife, fork/or spoon with a larger handle. Simple techniques such as using a steak knife all the time for cutting up food can help.

Tube feeding

A number of boys with Barth syndrome have a reduced appetite and need long term tube feeding to provide additional calories. Without tube feeding they would not be able to eat enough to maintain their growth. They are given specially designed milk usually via a tube surgically placed directly into their stomach, called a gastrostomy. Provision should be made for children to receive their gastrostomy feeds at school if necessary, however it is important to include the child in usual mealtimes with friends even if very little is eaten orally.

Weight gain

Although many boys struggle to gain weight and need extra help using tube feeding, for others excess weight is easily gained. This can occur during early adolescence when their slower rate of growth in height and reduced physical activity contribute to weight gain particularly around the abdominal area. This can make the boys feel self-conscious.

Educational Challenges

In surveys and reports, parents of boys with Barth syndrome and boys themselves have suggested that that they may face many challenges in school. These may include:

- staying focused in lessons
- short and long term memory problems and
- difficulties with reading and comprehension, retention and mathematics
- gaps in knowledge caused by frequent absences.

Absences

Have regular meetings with the family to plan support for children through absences. Good home/school liaison is vital. Try to ensure that any missed work is sent home as quickly as possible. Suggest revision guides for children to use at home to ensure there are no gaps in knowledge.

Learning

Research (Mazzocco, Henry & Kelley, 2007) suggests that boys with Barth syndrome are at increased risk of academic difficulties. Assessments in the UK show that boys with Barth syndrome experience varying difficulties in attention and concentration. Their general academic ability is also generally lower than average.

As each young person's ability to learn may be affected in different ways and to different degrees, it is important for parents and teachers to assess his needs and progress at key developmental stages to ensure that appropriate support can be provided.

It may be important to consider how the child or young person learns best. They may need written instructions for tasks or may need extra verbal help to ensure they know what is asked of them. As some children get tired when writing they may need to negotiate answering some questions verbally instead of writing them down.

Please contact us for further details of testing available through the NHS Barth Syndrome Service. See page 15 for contact details.

Social and Emotional Challenges

Many boys with Barth syndrome also face many social and emotional challenges at school; they must contend with being significantly smaller and weaker than their class mates and because of their levels of exhaustion in many activities like sports it is difficult for them to keep up with their peers.

Absences may make it harder for them to keep up with school work. . As teaching becomes more didactic in secondary school any difficulties in levels with exhaustion, handwriting, attention and memory may mean that some boys with Barth syndrome find this a more difficult time to learn.

As boys get older, they may struggle with loneliness as their peers become more independent. With the demands of school life and the effects of fatigue, it can be difficult to find time for social activities outside school.

Increasing awareness of their limitations might result in feelings of anger or frustration and learning more about their condition might result in feelings of anxiety.

It is therefore important for parents and teachers to highlight any issues early so that the right support systems and strategies are in place to help young people enjoy greater autonomy, success and well-being throughout their time at school.

Joining in activities in the classroom and playground

It is important for children and young people with Barth syndrome to be an active part of their peer group and for other students to learn that children with health conditions are valuable peers and friends. It is through peer relationships and being part of activities that young people develop their identity and build their self-esteem. Young children with Barth syndrome may lack balance and strength and need someone to keep an eye on them during busy playground breaks.

Below are some tips from families who have found ways to help their sons join in at school:

- Have regular meetings with school and talk to your son to build a picture of the activities he enjoys.
- Set up a diary at school or at home where the child or young person can use smiley faces or a rating scale to rate which activities they liked or didn't like. This may allow you to talk about what they didn't like about an activity or what they did like and what they could do more of.
- Talk to the teacher about ways in which the child or young person can join in part of an activity if doing the whole activity is not possible.

- Boys with Barth syndrome are in the best position to know how much they can cope with in a classroom or playground situation. If young people do not feel able to say when they need a break, teachers and parents could consider a traffic light system or a way for the young person to signal that they need a break. Examples that have worked well include a mini flipchart on the desk with pages for “I feel fine 😊”, “I feel tired 😫” and “I feel exhausted 🤯” or just wearing a cap during sports and turning the peak to face backwards when needing a break.
- Be creative about activities outside of school that the child or young person could do. This enables them to meet different friends and gain self-esteem through different activities.
- Don’t worry if your child or young person plays with girls more than boys as girls often play less active games.
- For younger children, a tricycle with a handle at the rear so they can be pushed when they get tired can be an ideal way of keeping up with peers without getting too tired. Older children might be able to use an electric quad bike or electric bicycle to allow them to go out with friends without getting too tired.

Helping a child’s peers understand Barth syndrome

There are many benefits in a young person and their peers understanding about Barth syndrome. It may engender support amongst peers and reduce anxiety and stress for the young person with Barth syndrome. It is natural for other children to be curious about another child’s illness, particularly if they have physical aids or are unable to join in some activities. It is often helpful for teachers to deal with any questions about a child’s condition openly but without embarrassing the child. The following tips are ways to help children and young people talk about illness openly and appropriately.

- Take cues from the child with Barth syndrome. If another child asks him a blatant question, such as “Why aren’t you joining in PE?” don’t rush to intervene. The child or young person may feel comfortable to answer the question or have a preferred response.
- Do intervene if another child is using derogatory language or is disrespectful to the child.
- Do provide opportunities for the child to talk about his disability, but don’t force it if he chooses not to, and don’t talk about the child if he or she is not there.

- Sharing exercises that the whole class participates in, such as writing their autobiographies or talking about a problem they had and how they solved it, may naturally draw out information about living with Barth syndrome.
- Some children may want to share stories or diaries about being in hospital or visiting the Barth Syndrome Conference, while others may feel embarrassed or prefer for Barth syndrome to not be a topic of conversation.
- Consider having a Disability Awareness Day or do a general presentation during an assembly. Invite non-disabled students to use a wheelchair, walker, or crutches for a day to learn about what it might be like using mobility aids and use these as opportunities to educate fellow students as well as allay fears.
- For younger children, teachers can read books about children who have illnesses to show that children may have different health problems.
- Consider having a session on different illnesses and include Barth syndrome if appropriate.
- Don't forget to support other children if they are worried or upset about another child's illness.

What if my child needs to talk to someone?

Talking to someone else can help your child see things in a different light. Suggest speaking to parents, carers, teachers or friends. They may be able to help resolve the problem, or suggest a solution.

The Barth Syndrome Service also includes a Clinical Psychologist who regularly sees affected boys and their families at clinics and who may be able to help. See Resources.

The Barth Syndrome Trust can put you in touch with other affected families who may be going through, or have some experience of, issues being faced. See Resources.

Get confidential advice

For free and confidential help or advice, you can contact a careers adviser by phone, email, web chat or text message.

Telephone: 0800 100 900

Or Connexions on http://www.direct.gov.uk/en/DI1/Directories/DG_10011904

What if my child is being bullied?

No one deserves to be bullied. Bullying doesn't always mean being physically hurt - you can be bullied in a number of ways.

If you're worried about bullying at school, follow the link below to get help and advice on how to stop it.

<http://www.direct.gov.uk/en/YoungPeople/HealthAndRelationships/Bullying/index.htm>

Resources

Further information and contacts

This guide has been developed jointly by the NHS Barth Syndrome Service in Bristol and the Barth Syndrome Trust after a series of consultations with affected individuals, their families, specialised educational staff and medical staff. If you have any questions or require any further information, we would be happy to hear from you. Contact details can be found below.

Barth Syndrome Service

Service Lead: Dr Colin Steward
Telephone – 01173428044

Principal Clinical Psychologist: Dr Vanessa Garratt

Clinical Nurse Specialist: Debbie Riddiford

Specialist Dietician: Nicol Clayton

Physiotherapist: Hannah Harbidge

Occupational Therapist: Emma Davies

www.barthsyndromeservice.nhs.uk

info@barthsyndromeservice.nhs.uk

Barth Syndrome Trust

Chairperson: Michaela Damin
Telephone – 01794 518785
www.barthsyndrome.org.uk

info@barthsyndrome.org.uk

Other useful links

IPSEA (Independent Panel for Special Education Advice): www.ipsea.org.uk

Contact a Family: cafamily.org.uk

Appendix A

What Is Barth Syndrome – An Overview

Barth syndrome is a genetic disease which affects only boys. It has always been thought to be relatively rare and less than 200 affected boys have ever been diagnosed in the world. However cases are now being recognised more frequently as techniques for testing for the disease improve and already 30 families affected by the disease have been identified in the UK alone. Since April 2010, NHS Specialised Services has funded a national Barth Syndrome Service for boys and young men affected by the disease run in conjunction between the Barth Syndrome Trust and the University Hospitals Bristol NHS Foundation Trust.

The disease affects production of a phospholipid chemical (called “cardiolipin”) which is an important component of some of the membranes within cells. Although the mechanisms by which this causes problems are not well understood, the consequence is compromised muscle function and white blood cell production.

Effects on the heart and other muscles

Barth syndrome tends to affect the cells that make up our heart muscle and many boys are diagnosed after developing heart failure during the first few years of life. The function of the heart can often be improved using medicines and in some boys it will return completely to normal. Most, however, will stay on one or more drugs to help heart function on a long-term basis. However, in other boys heart function either does not improve with drugs or improves initially but then deteriorates again; as a result, approximately one quarter of affected boys require heart transplantation. There can also be rhythm problems with the heart, particularly during adolescence.

Most boys are reviewed by a cardiologist on at least a six monthly basis during early childhood and at least annually in later childhood and adulthood. The effectiveness of pumping of the heart is monitored by an ultrasound examination – this is called “echocardiography”.

Muscles throughout the body are also affected, especially the powerful muscles at the top of the legs. This often causes problems in running and playing games/sport, and boys with Barth syndrome often suffer very significantly from lethargy and fatigue. Fine muscles can also be affected so that handwriting for sustained periods may be difficult. These are dealt with in more detail in the sections which follow.

Effects on blood cells: Neutropenia

Production of the white blood cells which fight bacterial germs – called neutrophils – is also reduced although in an unpredictable fashion. Many boys with the disease therefore have intermittent or persistently low blood neutrophil counts, a condition which doctors refer to as "neutropenia" or "being neutropenic". Low neutrophil counts make them excessively prone to bacterial infections, such as cellulitis (infection of the skin), pneumonia or septicaemia (where bacterial germs are actively growing in the bloodstream). These infections may have sudden onset and can worsen rapidly. The most reliable common early sign of low neutrophil counts are sore gums with red margins or mouth ulcers, although some boys may just look run down or have minor flu-like symptoms whilst they are neutropenic.

Boys who get these problems or other bacterial infections are usually treated preventatively:

With a medicine called granulocyte colony stimulating factor (G-CSF). This is typically given by parents as an injection just under the skin (subcutaneous injection) three times per week, usually on Mondays, Wednesdays and Fridays. This stops the neutrophil count falling too low and greatly reduces the risk of bacterial infection. The effectiveness of the G-CSF is monitored at a frequency suggested by the Barth Syndrome Service often by parents or the boys themselves performing fingerprick blood counts which are then analysed by their GP or local hospital.

With penicillin (or other antibiotic in penicillin allergic boys) once or twice per day on a permanent basis.

Low neutrophil counts do not increase susceptibility to viral infections and most boys with this disease will recover from viral upper respiratory tract infections, gastroenteritis, etc. normally. Despite this it is always important to err on the side of safety and therefore any boy with Barth syndrome would usually be assessed in hospital if they develop a temperature of 38°C or above or become unwell with unexplained headache, vomiting or other medical problems.

Effects on growth and feeding

Boys with the disease also grow slowly, usually following one of the lower lines on their growth chart. They tend to go into puberty late and will often continue to grow until 21 years. Most will eventually reach a normal adult height and some may even become tall despite having been one of the smallest boys in their class for most of their childhood.

Muscle cells are also important in the gut and many boys are relatively picky eaters. They may even have food fads for savoury foods such as pickles, gherkins, cheese or crisps. This can cause excessive concern, especially when accompanied by slow growth, but in fact most boys will eventually grow satisfactorily despite these dietary concerns.

Appendix B – Care Plan

Download the Care Plan template from the BST Website [here](#) or print the PDF copy below:



Care plan.pdf