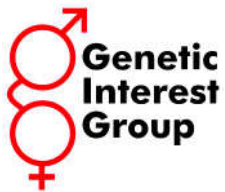


Acknowledgements.

We would like to thank all those who took part in the focus groups, interviews, and on-line survey and all who helped in the development of this guide.

The *Family Route Maps* project is funded by the charity Jeans for Genes and an unrestricted educational grant from Genzyme Therapeutics Ltd.



Genetic Interest Group (GIG)

Unit 4D, Leroy House

436 Essex Road, London, N1 3QP

Tel: 0207 7043141

www.gig.org.uk

Registered Charity No 1114195

Company Registered by Guarantee 05772999

This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor Barth Syndrome Trust can be held responsible for the accuracy of the information it contains. Links to other organisations are included for information purposes only and are not recommendations from GIG or Barth Syndrome Trust .

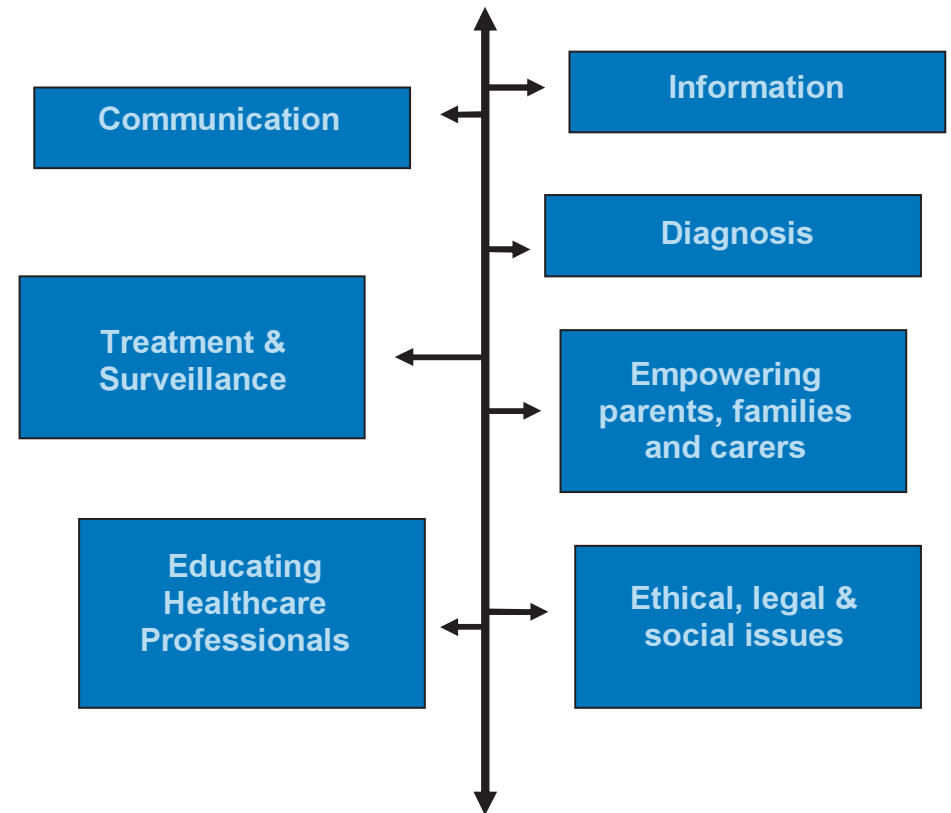
Date published: February 2008

Date for review: February 2009

Barth Syndrome

Family Route Map

This Family Route Map is a guide to current services and information.



Registered Charity Number 1100835

Introduction

This leaflet provides signposts to sources of current information and appropriate services for patients, their families, carers and healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) identified seven themes (see front cover) which have been used as the basis for developing this resource. This guide will help you find the information you need about Barth syndrome quickly and easily.

Barth syndrome is a genetic condition affecting only boys, which includes symptoms of heart problems, failure to thrive, being prone to infections, muscle weakness and tiredness. People with Barth syndrome do not always have all of the above symptoms at the same time.

More information about Barth syndrome can be found at www.barthsyndrome.org.uk or by contacting:

Barth Syndrome Trust

1 The Vikings

Romsey

SO51 5RG

Tel: 01794 518785

E-mail: info@barthsyndrome.org.uk

Information and **Signposts** have been divided into the following categories and some issues that may be relevant for you have also been explored:

- Diagnostic Testing
- Newly Diagnosed Boys
- Infants up to 2 years old
- 2 year olds to Pre-school children
- Boys aged 5 – 11 years old
- Boys aged 11 -16 years old
- Post 16 year old
- Information for healthcare professionals
- Other Information
- Useful Links

Other Information

Please contact the Barth Syndrome Trust for information about:

- Travel Insurance - list of sympathetic insurers
- MedicAlert - www.medicalert.org.uk
- Psychological help - finding counselling
- Personal Passport - keeping medical details
- Blue Badge - a disabled parking badge
- Wheelchair/quad bike - ways of getting around
- Physiotherapy/Occupational Therapy - specially tailored exercises

Links:

<u>Organisation</u>	<u>Website</u>	<u>Telephone</u>
British Heart Foundation	www.bhf.org.uk	08450 70 80 70
Cardiomyopathy Association	www.cardiomyopathy.org	0800 018 1024
Contact-A-Family provide a wide range of advice, particularly about claiming benefits	www.cafamily.org.uk	020 7608 8700
Children's Heart Federation	www.childrens-heart-fed.org.uk	0808 808 5000
Directgov provide a website with help on benefits and entitlements	www.direct.gov.uk	
Disability Benefits Office		08457 123456
Disablement Income Group Scotland	www.digscotland.org.uk	0131 555 2811
Early Support	www.earlysupport.org.uk	0845 602 2260
GIG (Genetic Interest Group) have a range of patient leaflets	www.gig.org.uk/eurogentest_patientleaflets.htm	020 7704 3141
IPSEA provide free and independent advice about special educational needs	www.ipsea.org.uk	0800 0184016
The Family Fund	www.familyfund.org.uk	0845 1304542
The UK Genetic Testing Network has produced a patient leaflet www.ukgtn.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html		
Whizz Kidz can provide help to fund equipment. They have a Family Advice Worker	www.whizz-kidz.org.uk	020 7798 6114

Information for healthcare professionals

Barth syndrome is caused by a defect of the *G4.5* gene, resulting in an inborn error of metabolism. The following information is available:

Short Medical Brochure

See: www.barthsyndrome.org.uk This includes information about:

- major clinical problems
- incidence statistics
- cardiac decompensation, transplantation, arrhythmias
- accelerated growth in late teenage years
- ways to manage neutropaenia and hypoglycaemia
- useful contacts in the UK

Detailed Medical Brochure

See: www.barthsyndrome.org.uk (Click on Physician references, then Overview). Treatment Guidelines are currently being developed. In the interim, there is much information about treatment options in this brochure. It also includes highlights of clinical knowledge and a bibliography of published journal articles.

International Medical Database and Biobank - launched in 2006, UK patients enrolled in 2007. For more details, please see www.peds.ufl.edu/barthsyndromeregistry/contact.htm

International Barth Syndrome Scientific and Medical Conference

Please contact the Barth Syndrome Trust if you are interested in attending or receiving a DVD of the past presentations.

Doctors' Listserv

Do you have a Barth patient and need to discuss the case with another doctor who has experience with the disorder? If so, then please sign up to the specialist e-mail forum which facilitates open dialogue between healthcare professionals.

See: www.barthsyndrome.org.uk for further details.

The NHS National Genetics Education and Development Centre provide information and resources for healthcare professionals.

See: www.geneticseducation.nhs.uk

Diagnostic Testing

To diagnose this syndrome in boys the following may be used:

- Clinical diagnosis based on symptoms
- DNA testing – a genetic test (from a blood sample which can take up to four months to complete)
- Cardiolipin analysis which involves a preliminary blood spot test. Results are rapid and reliable. This test can be arranged through the Barth Syndrome Trust.

Because this is an hereditary condition, other members of the family may be affected – please see '**Inheritance**' on the Barth Syndrome website www.barthsyndrome.org.uk or request a copy of their Factsheet '**Genetics of Barth Syndrome**'.

You can ask for a second opinion or referral within the NHS to see a specialist who has experience of diagnosing and treating boys with Barth syndrome.

For information on the referral process please see: www.gig.org.uk/docs/referrals.pdf

Newly Diagnosed Boys

There is an annual multidisciplinary **Barth Syndrome Clinic** that sees children from infants to adulthood, held at Bristol Royal Hospital for Children. Here patients and their families, together with expert doctors and clinical staff, can meet to discuss latest treatments and share knowledge in a supportive environment.

Your son may be referred to a number of different specialities because Barth syndrome affects so many different body systems. Specialist doctors and/or departments involved include:

- Cardiology
- Haematology
- Immunology
- Neurology
- Nutrition/dietetics
- Occupational therapy
- Paediatrics
- Endocrinology
- Clinical genetics
- Physiotherapy
- Gastroenterology
- Metabolic units

Infants up to 2 years old

It's important to note that your child may not have all of the symptoms below but information about the following is available should you need it.

Please see the following fact sheets from the Barth Syndrome Foundation available at the website: www.barthsyndrome.org.uk

- **Heart failure**
- **Neutropaenia**
- **Medications**
- **Neurology**
- **Metabolism**
- **Nutrition and feeding problems**
- **Care Plan**
- **Practical tips for parents**

Other common problems during this period include:

- Muscle weakness and delayed milestones
- Hypoglycaemia (low blood sugar)
- Diarrhoea and/or vomiting

TIP

A typed summary sheet at the front of your child's hospital notes will assist clinical staff to quickly find details of medication and other treatment required.

2 year olds to Pre-school children

Any symptoms may now become apparent *or* in some cases previous symptoms may improve. However, considerations now additionally include:

- The need for hand washing and good hygiene when mixing with other young children because of the possibility of catching infections.
- Discussing needs with nursery staff, for example, special diets or food fads, and managing tiredness.

Affected boys are often bright and social but need extra help for physical symptoms and fatigue. A statutory assessment of any special educational needs from your local authority would be helpful at this time to help your son achieve his full potential. See the fact sheet, 'Education and Learning' available at www.barthsyndrome.org.uk

Boys aged 5 – 11 years old

The Barth Syndrome Foundation has produced information on education strategies, please visit www.barthsyndrome.org.uk

Symptoms may lessen a little at this stage and boys may be in a period when they are stable and appear fairly well.

- A statement of special educational needs might be needed.
- Adaptations at home and school may be useful. (e.g. extra set of books left at school, a memory stick to avoid carrying heavy loads, using a laptop or a scribe if writing is tiring, extra supervision during playtimes etc). This is often a good time to take holidays and enjoy social activities and hobbies.
- Careful health monitoring must, however, be continued.
- Planning for secondary school should also be commenced.

Boys aged 11 – 16 years old

Heart function may deteriorate during this period and/or abnormal rhythms might start to occur. Close monitoring is advised. Please discuss any concerns you may have with your child's cardiologist or electrophysiologist.

- Transition into adult care for health services will start at 16.
- Forward planning for further education and/or career can be assisted by **Connexions** at : www.connexions.gov.uk or phone: 080 800 13 2 19

Post 16 years old

- Growth spurts can happen now and may need managing.
- Arrhythmias may occur and our latest registry data show that the majority of young people fitted with Internal Cardioverter Defibrillators (ICD's) were 16+.
- Physical aspects may require boys to work around their excessive growth and manage the possible associated fatigue and muscle weakness at this time.

TIP

Ask for a copy of your clinic notes when you see a doctor so that you always have the latest information at hand.