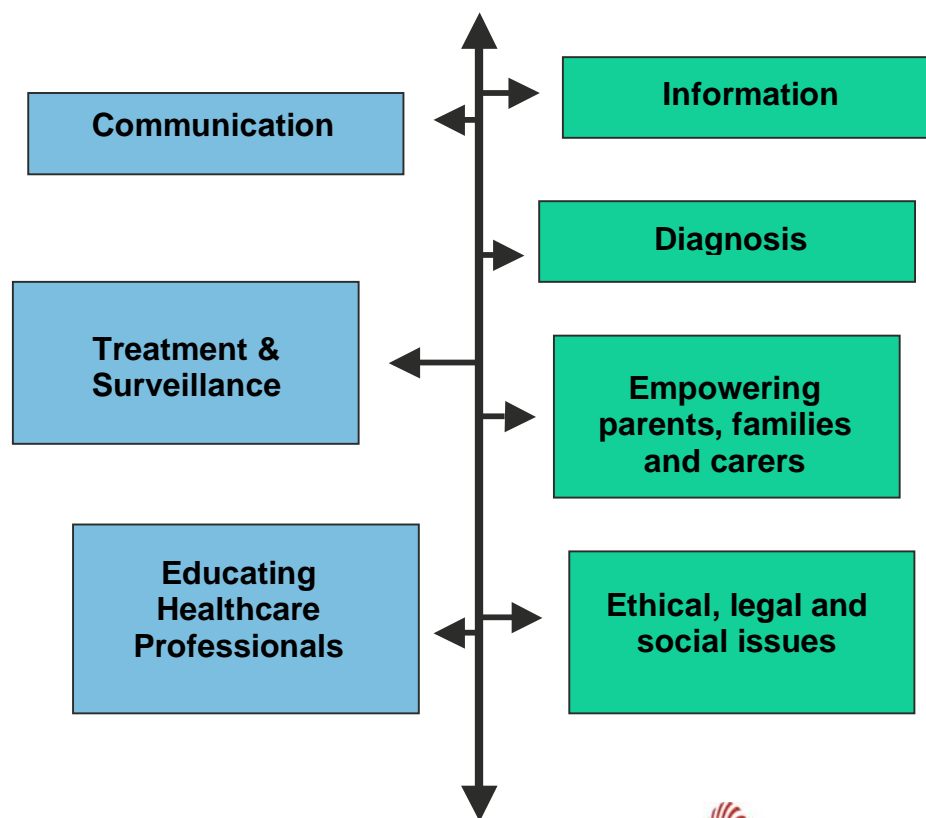


Chronic Granulomatous Disorder

Family Route Map

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



Challenging genetic disorders



CgD

The CGD Research Trust
Registered Charity Number 1003425

The Chronic Granulomatous Disorder Research Trust

Registered Charity Number 100342

INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families and carers, together with 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 Chronic Granulomatous Disorder (CGD) quickly and easily.

The Chronic Granulomatous Disorder Research Trust was founded in the UK in 1991. It is the only organisation in the UK and rest of Europe that specifically works for people and families affected by CGD. The CGD Research Trust is a patient support group and provides a point of contact for affected families as well as providing information for adults and parents and medical professionals on diagnosis, treatment and care. The CGD Research Trust funds a specialist CGD nursing service and a psychological service that can help with emotional support. CGD RT runs its own website, produces newsletters, organises medical conferences, get together weekends and fun days for families and funds state of the art research into the treatment, management and potential cure of CGD.

The Chronic Granulomatous Disorder Research Trust

- Provides a point of contact and support for individuals and families affected by CGD
- Provides specialist care for people affected by CGD through CGD specialist nurses
- Promotes mental well-being for those affected by CGD through psychological support
- Promotes and funds research into the cause, symptoms, treatment and cure for CGD and disseminate the results of such research
- Raises awareness of CGD throughout the medical profession and the general public

Contact details:

***Mrs Rosemarie Rymer
The CGD Research Trust
Manor Farm
Wimborne St Giles
Dorset
BH21 5NL***

**Tel No: +44 (0) 1725 517977
Email: cgd@cgdrt.co.uk
Web: www.cgd.org.uk**



The Chronic Granulomatous Disorder Research Trust

The CGD Research Trust is a unique and special charity striving for a cure and care for people affected by chronic granulomatous disorder.

INFORMATION

CGD is an inherited life-threatening disorder that prevents the immune system working properly to fight off certain bacterial and fungal infections. The exact incidence of the condition is unknown but is thought to affect approximately 8-10 people in a million. People with CGD carry a faulty bone marrow gene, which means that white blood cells, known as phagocytes, do not function correctly. These cells are responsible for killing fungi and bacteria and the defect makes people with CGD more susceptible to fungal and bacterial infections.

The hallmark of the disorder is early onset of severe recurrent bacterial and fungal infections. Three-quarters of people are diagnosed during the first five years of life but it is likely that the medical profession under-diagnoses CGD throughout the population. Pneumonia can be one of the most common symptoms in addition to infections of the skin, lungs, gastrointestinal tract, lymph nodes, liver, and spleen. Another hallmark of CGD is the development of granulomas (nodules of inflammatory tissue) in the skin, gastrointestinal tract, and urinary tract. At diagnosis, some people present with symptoms related to these granulomas. Bowel problems can also be characteristic. The wide variety of symptoms, and the fact symptoms can be common to other conditions, mean that misdiagnosis can occur. For example, symptoms can lead to misdiagnoses such as Crohn's, TB and leukaemia.

Medication in the form of antibiotics and anti-fungal therapy is necessary. These are taken on a daily basis to keep infection at bay but problems can still arise that can lead to serious illnesses and prolonged periods in hospital. The quality of life of those with the condition may also be severely affected. Bone marrow transplant is an effective treatment for the condition, although this is not without risk. It is becoming possible for a greater number of affected people using related and more recently, unrelated matched donors. There are some people for whom transplant will not be possible. Progress is being made in other forms of treatment such as gene replacement therapy.

DIAGNOSIS AND GENETIC TESTING

If CGD is suspected, patients should be referred with urgency to an immunology specialist. Genetic testing is not required to make a diagnosis of CGD and appropriate treatment can be given in the absence of knowing the precise genetic mutation involved. Diagnosis of CGD can be made using a functional blood test rather than a genetic test. This test, called the nitroblue tetrazolium dye reduction test is based on the ability of patient's phagocyte cells to produce an oxygen burst. It is this oxygen burst that is important for neutralising bacteria and fungi within cells. Increasingly this test is slowly being replaced by a more sophisticated method called dihydrorhodamine (DHR) test. Here phagocytic cells reduce DHR to the strongly fluorescent compound rhodamine, individual fluorescent cells are then counted, and the amount of fluorescence per cell is quantified with a specialised piece of equipment known as a flow cytometer. As CGD is an inheritable condition, a diagnosis of CGD must be followed up by appropriate genetic counseling and predictive gene testing for the whole family. Pinpointing the specific gene mutation is useful to establish the genetic inheritance pattern and to help with family counseling. The low incidence of CGD and the large number of unique mutations preclude standardized genetic testing. Therefore individual genetic analysis remains the domain of specialized research laboratories. Samples will be sent to these laboratories by the treating clinician.

CGD can result from mutations in one of four of the components responsible for the oxidative burst. These mutations cause either a complete absence of one of the components or affect

it so that it does not function properly. The most common form of CGD is called X-linked CGD. Here only boys are affected and it is caused by inheritance of the genetic fault from the mother. The other forms of CGD represent about 40% of cases and are inherited in a different manner and are due to both parents carrying a gene mutation.

There are a number of issues surrounding genetic testing particularly in relation to children and as such, many patients may wish to be seen and counselled by a consultant clinical geneticist as early on as possible. The Genetic Interest Group (GIG) website has a series of leaflets explaining more about inherited conditions and includes; a glossary of terms used in genetics, and useful questions to ask when going for an appointment.

Please see www.gig.org.uk/eurogentest_patientleaflets.htm

The UKGTN (UK Genetic Testing Network) has produced a patient leaflet to help understand testing. Available at their website: www.ukgt.nhs.uk/gtn/Home

For more information on the Referral Process please see the GIG website:

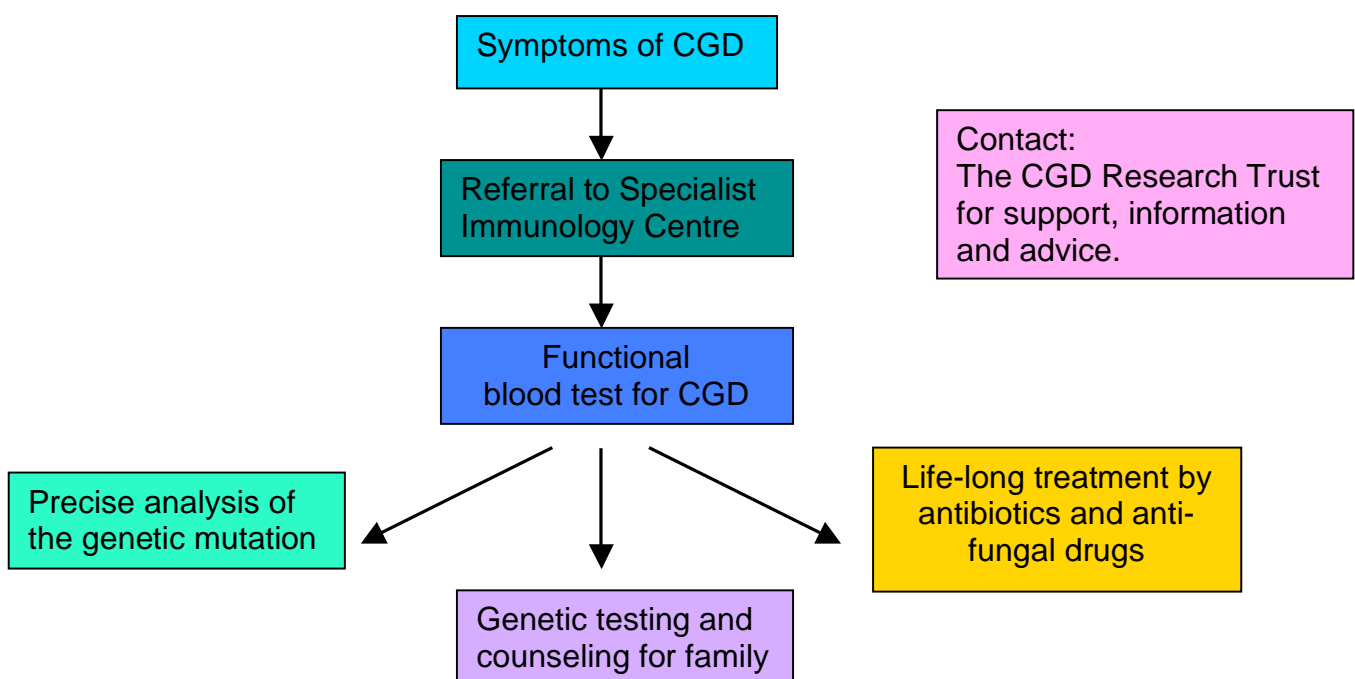
www.gig.org.uk/docs/referrals.pdf

Second Opinion at diagnosis

In the UK, patients have the right to ask for a second opinion via their first specialist or through their GP. They are not, however, entitled to receive that second opinion if the clinicians do not think it necessary. A patient may appeal against any decision via their Primary Care Trust (PCT).

TREATMENT AND SURVEILLANCE

Treatment is by daily use of antibiotics and antifungal drugs. Other medicines may be given to deal with problems associated with CGD such as inflammation. It is extremely important to contact a doctor if an infection is suspected so that prompt action can be given. CGD patients should look out for the signs of infection. Simple steps and changes in life style can be taken to avoid infection. Details can be obtained by contacting the CGD Research Trust. Bone marrow transplant is the best treatment option at the present time. This can be a difficult procedure, requiring a prolonged hospital stay. Research and refinement of gene replacement therapy procedures are currently being explored.



TIP

Talk to other families who have affected children through forums and contacting the CGD Research Trust. www.cgd.org.uk

INFORMATION FOR HEALTHCARE PROFESSIONALS

The following information is available through the CGD Research Trust website www.cgd.org.co.uk or by contacting the charity directly:

- **Short Guide to CGD for Medical Professionals** - This is written by medical experts on CGD for other medical professionals.
- Information from workshops and conferences at www.cgd.org.uk/cgd/medics

PATIENT-AIMED INFORMATION

The CGD Research Trust offers tailored information for parents, teenagers and young adults

- **Information booklet about CGD – A Guide for CGD Patients and their Families.** This contains easy to understand information on CGD and tries to answer the common questions that are often asked by parents and guardians of children with CGD, and by the children themselves.

Fact Sheets and Information Sheets

- **About your medicines** - Children and adults with CGD take medications every day to help prevent bacterial and fungal infection. This fact sheet gives information on an antibiotic drug and antifungal drug that are commonly given to people with CGD.
- **Dealing with a liver abscess** - People with CGD are prone to getting abscesses in lots of places and a liver abscess may be the feature that leads to a diagnosis of CGD. This fact sheet gives information on why they occur and how they are treated.
- **Looking after your skin** - People with CGD may find that they are prone to sensitive or dry skin conditions. These can be uncomfortable and may sometimes cause eczema-like rashes. Simple steps are highlighted that help protect skin and minimise the chances of skin irritation and infection in CGD.
- **Coping with bowel problems in CGD** - People with CGD often have problems with their bowels. It is very common for people with CGD to find that their bowel function is varied and that the severity of the problems can vary. This comprehensive fact sheet covers what problems may be encountered, practical solutions that may help and what medical treatments are on offer.
- **Immunisations** - This fact sheet aims to tell you a little more about what immunisation should be given or avoided in CGD.
- **Keeping myself well** - This general fact sheet gives tips on how to keep well with a chronic condition such as CGD. It gives wide-ranging advice on all aspects of personal health and well-being.
- **Looking after your lungs** - This gives information on why it is important to look after respiratory health in CGD.
- **Lupus in carrier mothers** – This gives information about the condition lupus that may affect some mothers who are carriers of CGD.
- **Food for Thought** - This information sheet provides a brief introduction to food and CGD including dietary advice, food facts, recipes and on feeding children.
- **Travel and CGD** - This provides useful information and advice on what to consider when going away from home on holiday or traveling.

OTHER INFORMATION FOR PATIENTS, FAMILIES AND CARERS

Shared Care

In the UK, a joint care system means that you can see your local consultant regularly *and* make an annual visit to a CGD specialist consultant, who has the advantage of seeing a number of affected patients and will know about the latest developments in treatment. He/she will liaise with your local consultant and any others involved in your care. To teak advantage of the shared care system, you need to ask your local consultant to refer you to a specialist centre, or your child to a paediatric CGD specialist. If you feel you need more advice about this, please ring the CGD Research Trust on 01725 517 977.

The CGD Research Trust

The CGD Research Trust provides the following services for individuals affected by CGD

- CGD Specialist Nursing Service - For parents of children with CGD and affected adults the CGD Research Trust can offer the help and support of two CGD Specialist Nurses who can help in all aspects of care.
- Psychology support service
- Pre-paid prescription charge certificates for individuals with CGD aged 16+
- Notes on the Management of CGD – available in plastic wallet or portable memory stick. These provide information on CGD, management of the condition and have space for you to keep your own notes. A valuable record to have when you see GP's and other healthcare professionals
- Careers help through an arrangement with Connexions Merseyside

Insurance

It is common to have difficulties finding adequate and affordable insurance policies once you have a pre-existing condition. The CGD Research Trust cannot make any recommendations but members have used insurance broker J.D. Consultants and found them very helpful. They will find you travel insurance and they do not charge you for their services or put any additional cost on to the insurance policy. Phone: 01689 859102

Using their services may be the best way of finding cover.

Other advice

You can get advice on finance and non-medical topics from our CGD CAB (Citizen's Advice Bureau) Helpline on 01202 857108. Alternatively, CAB (Citizens Advice Bureau) can help with advice locally about benefits and can be found through your phone directory.

TIPs

Don't be afraid:

- to say if you don't understand something
- to ask the doctor to repeat what he has said or to give you further explanation.
- to say how you feel: if you don't want medical students participating in your appointments; or clinical staff discussing your child in front of them, don't be afraid to say so.
- to phone back after an appointment if you get home and find you are not sure what was said

When you get into an appointment, it can be difficult to remember all the questions you meant to ask. It is useful to jot these down and take the piece of paper with you, as a reminder

UK Hospitals specialising in treating CGD

Great Ormond Street Hospital - www.gosh.org

Newcastle General Hospital - www.newcastle-hospitals.org.uk

Royal Free Hospital - www.royalfree.nhs.uk

Bone Marrow Registries

Bone marrow transplantation, BMT, (also known as haematopoietic stem cell transplantation) can be an effective treatment option for CGD. The CGD Research Trust offers specific information on this treatment for CGD. For more information, inserting Bone Marrow Transplant in the search box at www.cgd.org.uk.

The following organisations also offer useful information on BMT and donors:

Anthony Nolan Trust: www.anthonynolan.org.uk

National Blood Service for England and North Wales: www.blood.co.uk

Scottish Blood Transfusion Service: www.scotblood.co.uk

The Welsh Blood Service: www.welsh-blood.org.uk

Information on Gene Therapy

Gene Therapy for CGD www.cgd.org.uk/cgd/research/projects_we_support/gene_therapy

British Society for Gene Therapy - www.bsgt.org

Gene Therapy Advisory Committee - www.advisorybodies.doh.gov.uk/genetics/gtac/

Understanding fungal infections

Fungal infections can be a major problem in CGD. To find out more Information on Aspergillus and anti-fungal drugs visit www.aspergillus.org.uk (added .uk)

General organisations that can help

The National Organisation for Rare Diseases www.rarediseases.org

Orphanet (European database) www.orpha.net - Free-access website providing information on rare diseases. Search on 'Granulomatous disease, chronic'

Carers UK www.carersuk.org Tel: 0808 808 7777

Contact-A-Family: www.cafamily.org.uk Tel: 020 7608 8700

Department of Health: www.dh.gov.uk

Department of Health have produced a set of 'Questions to ask' to take with you to your appointment with a specialist. Available in several different languages; use the Search option

on their website.

NHS Telling stories – Understanding Real Life Genetics
www.geneticseducation.nhs.uk

GIG (Genetic Interest Group) have a range of patient leaflets on their website
www.gig.org.uk/eurogentest_patientleaflets.htm Tel: 020 7704 3141

The NHS National Genetics Education and Development Centre
www.geneticseducation.nhs.uk provide information and resources for healthcare professionals.

Genetics in Family Medicine: The Australian Handbook for General Practitioners
www.gpgenetics.edu.au

Claiming benefits

Disability Benefits Office Tel: 08457 123456

Directgov www.direct.gov.uk A website with help on benefits and entitlements

Disablement Income Group Scotland www.digscotland.org.uk Tel: 0131 555 2811

Information on Genetics

British Society for Human Genetics (BSHG): www.bshg.org.uk

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The CGD Research Trust
Manor Farm
Wimborne St Giles
Dorset, BH21 5NL
Tel No: +44 (0) 1725 517977
Email: cgd@cgdrt.co.uk; Web: www.cgd.org.uk

This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor the Chronic Granulomatous Disorder Research 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 the Chronic Granulomatous Disorder Research Trust. Date published: August 2008; Date for review: August 2009

