

# The Chronic Granulomatous Disorder Research Trust

## Research Strategy

### **Aims and objectives**

Chronic Granulomatous Disorder (CGD) is a rare inherited genetic condition that affects approximately 1 in 200,000 people. CGD is a complex life threatening condition. The Trust is the only UK charity funding research into all aspects of this genetic condition. The CGD Research Trust has five strategic aims:

1. The support of medical and scientific research projects aimed towards understanding the cause, inheritance, and management of this disorder. The CGD Research Trust is committed to funding pioneering research that will result in the translation of laboratory based research into clinical benefit.
2. Providing specialist support for affected people and their families and ensuring they get the best possible care and assistance in all aspects of their lives.
3. Raising awareness of CGD with the general public and among medical professionals.
4. Dissemination of clinical and scientific knowledge.
5. Building partnerships with other charities who fund research and care for individuals affected by other single gene disorders so that additive value can be gained in tackling problems common to other genetic conditions and primary immune deficiencies.

### ***Priority research and scientific areas – what we support***

The CGD Research Trust supports scientific and applied research so as to make the maximum impact in establishing the cause and treatment of CGD and the quality of life of people affected by the condition. CGD is a complex disorder requiring considerable specialist treatment. The research addresses the serious symptoms and complications of the disease, which include infection, tissue damage caused by inflammation, bowel, digestive and urinary tract complications. It is vital that CGD patients remain as healthy as possible so that they will be able to benefit from the curative treatment options currently available and/or under development. The Trust supports projects in the UK and internationally and has funded research in over five different countries.

### **Priority research and support areas:**

#### ***Gene Therapy***

Since the identification of one of the genes that causes CGD in 1986, research is increasingly focused on correcting the genetic defect by gene therapy. This may provide a treatment option for individuals affected by CGD when bone marrow transplantation is not a viable option. A large part of our research income is therefore given to projects that are developing gene therapy procedures to replace the defective gene in the bone marrow of CGD patients with a correctly functioning gene. Over £1.5 million has been committed to this area over the last eight years. It is anticipated that CGD patients will then have a correctly functioning immune system that can fight infections. CGD Research Trust funded projects have already found clinical evidence that gene therapy is effective in some CGD patients and the CGD Research Trust leads the field in this area. Results from this research will also help in the development of treatments for many other genetic diseases.

#### ***Controlling Infection and Inflammation***

It is essential to find more effective means of reducing and fighting infections and dampening down of tissue-damaging inflammation. Infections caused by the spores from the fungus *Aspergillus* can cause serious problems in CGD because such infections are invasive, increase rapidly and are often difficult to eradicate. The Trust supports work that will advance the development of better anti-fungal drugs. The Trust

is committed to funding research into why the faulty gene in CGD causes severe and inflammatory problems such as tissue and organ abscesses. Twelve projects have been supported in these areas since 1998.

### ***Understanding more about the genetic defect***

In CGD, parts of a specialised enzyme that is important for killing bacteria and fungi is either missing or defective. The CGD Research Trust supports work that will help understand how the genetic mutations in CGD affect the working of this key enzyme. This will add to the chain of progress by improving knowledge and understanding of the complex medical conditions that occur in CGD.

### ***Clinical support and quality of life issues***

The CGD Research Trust funds the only CGD clinical nurse specialist in the country. This support service was set up in 2000 to provide essential support to people with CGD and their families. The service provides practical advice and counselling, promotes the interests of CGD patients, provides liaison between all the professionals involved in the care of a patient and raises awareness of the condition at medical conferences and among healthcare professionals. This service begins at diagnosis and continues through adolescence and adulthood.

The Trust is committed to expanding these services and hopes to raise an additional £2.5 million over the next three years.

### **How we raise our funds and our grant award schemes**

The CGD Research Trust raises a large proportion of income through the Jeans for Genes annual campaign. Funds are shared between four partner charities and a number of guest charities. The CGD Research Trust is the founder member of the Jeans for Genes Campaign and owns the J4G trademark. Since 1996, J4G has raised over £24 million spreading the support to other genetic conditions. The CGD Research Trust works with its partner charities to ensure optimum use is made of the funds available, that there is no duplication of research effort and to canvass and lobby for better healthcare provision on behalf of patients with primary immunodeficiencies.

To further the aims of the CGD Research Trust has four types of grant schemes:

1. Post-doctoral project grants of up to three years duration are available for applied biomedical/clinical research.
2. Ph.D. studentships for exceptional candidates in laboratories that have a proven track record research in CGD. The studentships are designed to encourage the best young science graduates to embark on a research career in CGD.
3. One-year 'start up' grants (maximum £40, 000) are offered for more basic research proposals that may lead to new avenues of investigation into the diagnosis and treatment of CGD.
4. Three-year research fellowships are available for medically qualified individuals wishing to specialise in CGD research in an appropriate laboratory or hospital unit.

There are no geographical restrictions concerning the allocation of funding. The criteria for funding are excellent science and relevance to and benefit for CGD. All clinical and research projects are subjected to full peer review and are scrutinised by a panel of medical and scientific experts. The CGD Research Trust currently funds projects both nationally and internationally at different institutions, including hospitals, universities and research centres of excellence and encourages and promotes dialogue between researchers, clinicians, patients and carers. The Trust is committed to spending an extra £1 million on gene therapy research over the next five years and we are anticipating raising an extra £5 million for other research areas.