

Introduction

The Niemann-Pick Disease Group (UK) is a small charity representing just over 100 families afflicted by the Niemann-Pick diseases. It became an independent charity in 1996 and was registered as a company limited by guarantee in September 2011. The Charity is governed by Articles of Association and conforms to the requirements of both Companies House and the Charity Commission.

The objects of the Charity are to relieve sickness among families afflicted by Niemann-Pick disease and any distress that may arise there from and to advance the education of such families, interested professionals and the general public in all matters concerning the disease as the committee may determine

The NPDG (UK) and its Associate Members raise funds to sustain the services offered in support of families in line with its objects. These are described as Care and Support, Information and Research. Restricted donations are set aside for the purpose intended.

Policy

- The NPDG (UK) will support organisations, universities and pharmaceutical companies who work together to better understand the nature of Niemann-Pick diseases and who have the potential to develop therapies that may halt disease progression or contribute to the effective treatment of Niemann-Pick diseases.
- The Board of Trustees will consider applications for funding and will determine the nature of research to be undertaken in consultation with its Research Co-ordinator and Medical Adviser. Where appropriate, guidance will be sought from the Scientific Advisory Board that currently provides advice to the National Niemann-Pick Disease Foundation in the U.S.A. - Where the size of the grant is deemed to be significant, a peer review process will be employed.
- The Board of Trustees have the authority to set a research budget and to agree funding for research projects and other research related activities in furtherance of the objects.
- The charity will periodically review its research policy and strategy in line with its objects and priorities.
- The charity will not contribute to the Full Economic Costs of funding research but will instead pay for the actual costs that are identifiable as part of the project. These may include labour, consumables, agreed travel / subsistence and publication costs
- The charity will not act as employers with respect to people or organisations benefiting from funding.
- Researchers are required to meet the various legal and regulatory requirements placed upon them and will have obtained all the necessary licenses and approvals as appropriate.
- Researchers are required to maintain a separate cost account for work in association with funding and keep records to support the costs incurred. Payment of invoices will be made against an agreed schedule.
- Regular progress reports are to be submitted to the Charity and the findings from research activities, funded by the charity, should be made available to all interested parties and are to be published in a scientific journal where appropriate.

Strategy

The Charity will share knowledge, expertise and where possible work in partnership and collaboration with other charities, medical authorities and scientific institutions across the world. It will support scientific establishments in the UK and elsewhere and, where possible, will fund appropriate research in order to improve medical and scientific knowledge and understanding of Niemann-Pick diseases.

Strategic objectives

Medical and scientific studies of Niemann-Pick diseases have been progressed over many decades and a substantial body of knowledge has been accumulated. A great deal of work remains to be carried out in order to gain a full understanding of the diseases, their diagnosis, pathology and treatment. It is recognised that improvements and significant effort is required in areas such as:-

- Diagnosis – improvement is needed to the reliability and speed of diagnosis.
- Genetics – improvement in identification of disease causing mutations.
- Cellular biology – identification of protein functions and resulting cellular abnormalities
- Cellular pathology – identification of the pathways leading to cell malfunction and death
- Therapeutic intervention – identification of therapeutic targets and approaches to phenotype correction or modification especially in relation to the brain
- Clinical trials – translation of therapeutic strategies into clinical trials
- Disease prevention – improved education and access to cascade genetic testing and partner compatibility testing in the general population. Improvements in PGD/IVF

Research studies in pursuance of the above objectives will normally be considered under the following general headings:-

Diagnostic studies

These will normally be pilot studies or larger research projects that investigate the cause and progression of Niemann-Pick Disease. Such studies are aimed at providing an understanding of the basic science associated with the disease and are essential in identifying how different tissues, cells and organs react to the genetic lesion. This knowledge in turn, helps to inform therapeutic development strategies. Studies may include genetics, cellular function abnormalities, pathology, clinical progression, effects on tissues and organs, natural history, biomarkers, genetic and biochemical methods of diagnosis.

Studies may include a range of animal, cell, and computer models and improvements to these models to more closely represent the human defect is an important area for research.

This is not an exhaustive list of topics.

Therapeutic studies

This area of work is concerned with the identification of therapeutic targets at all levels from genetic through to whole person. Whilst the ultimate goal is to find a cure, interim measures aimed at ameliorating disease symptoms are likely to be more practicable in the short and medium term.

Studies may include therapeutic approaches such as gene or enzyme transfer, substrate reduction, epigenetic approaches, chaperone molecules, inhibitors of inflammation and other disease modifying concepts including combinatorial approaches.

Any potential therapy must be capable of delivery to the brain (central nervous system – CNS). Approaches to this are therefore of great importance and should be considered concurrently with any therapeutic study.

Whenever a pre-clinical study into a potential therapy indicates significant efficacy, the charity will progress discussions aimed at facilitating clinical trials in the UK or in providing access to clinical trials planned elsewhere. In this respect the charity will seek to collaborate with medical authorities, pharmaceutical companies and other stakeholders in bringing such trial to fruition.

Strategy management

In general the charity will encourage researchers to submit requests for funding and will maintain a dialogue to assist in forecasting work that is planned in the short and medium terms.

Research into medical science is expensive and requires a collaborative approach with other groups such as healthcare authorities, the medical science industry and other patient organisations. The charity will work with other groups as appropriate in order to progress the Strategic Objectives. Grant giving organisations that fund research into medical science will also continue to be approached for assistance.

Collaboration and information exchange with other funders of Niemann-Pick disease research will assist in the avoidance of duplication of effort or repeating studies already carried out. The collaborative approach also provides a major benefit in being able to access scientific advisory boards served by scientists and clinicians at the forefront of disease research.

In order to encourage research into the diseases, the charity will utilise the following approaches to funding:-

- Research grants (directed) – Advertise for specific research in designated areas.
- Research grants (undirected) – Advertise for applications for general NPD research.
- Facilitative funding – for seed grants, research fellowships, partnerships, ad hoc stop gap funding.
- Employment of research officer/nurse – to co-ordinate information and keep the membership informed about developments.
- The Peter Carlton Jones Memorial Award – An award made annually to young scientists working in the field of Niemann-Pick disease.

The research strategy will be subject to review from time to time in response to changing research environment, the introduction of new technology, availability of funding and other factors which may influence current knowledge and understanding.

Prevention

Prevention is the most certain way of reducing the incidence of Niemann-Pick disease. Science and technology will increasingly be able to provide genetic tests to determine if a sibling or a potential partner is carrying a defective Niemann-Pick gene.

The Niemann-Pick Disease Group (UK) will talk to any family who wish to make informed judgements about prevention programmes as a way of reducing the risk of future generations inheriting the Niemann-Pick gene. Options that may be available include carrier testing, prenatal testing, IVF and preimplantation genetic diagnosis.

However, taking the first step towards prevention is a personal choice and may be prove very difficult. In all cases, families should seek the advice of their GP who will arrange a meeting with a genetic counsellor who may help in deciding the best outcome.