# UK Gauchers Association

**Project Title: Patient Advocacy** with an emphasis on newly diagnosed and Neuronopathic Gaucher disease (nGD); a feasibility study.

## Background

**a) Gaucher Disease -** Gaucher Disease is a genetically inherited, enzyme deficiency disorder. Symptoms range from mild to severe and can appear at any time, from infancy to old age. They may include anaemia, fatigue, easy bruising and a tendency to bleed. An enlarged spleen and liver with a protruding abdomen may also occur, as well as bone pain, demineralisation and fractures.

People with Gaucher disease lack sufficient levels of an enzyme called *glucocerebrosidase*. When worn-out cells break down, a fatty substance called *glucocerebroside* is released. This is normally broken down by glucocerebrosidase. It therefore accumulates in patients with Gaucher disease, particularly in the spleen, liver, bone marrow, and sometimes in the central nervous system.

The most common form of Gaucher Disease (Type 1) affects 1 in 100,000 of the general population but 1 in 850 of Ashkenazi descent, although not all those who inherit the mutated genes for this disorder will show symptoms.

In the rare Neuronopathic (Types 2 and 3) Gaucher Disease, neurological symptoms occur; they include an eye movement disorder (oculomotor apraxia), unsteadiness (ataxia), fits, loss of skills and a central auditory processing disorder. Children with Type 2 Gaucher disease die within one or two years of birth.

Detailed information is available on the Gauchers Association website at www.gaucher.org.uk

Additional publications on Neuronopathic Gaucher disease and a separate leaflet on Type 2 Gaucher disease are available from the Gauchers Association, and can also be downloaded from the Gauchers Association website.

**b) The UK Gauchers Association -** Since 1991, The Gauchers Association has been active in promoting awareness and research, providing general and specific information - including keeping our members up-to-date on the latest research developments, and perhaps most importantly, establishing a support network for those affected by Gaucher disease.

Most of our members are in the UK and Ireland. We are a registered charity that relies solely on fundraising and voluntary donations. The Association aims to:

- Provide information about Gaucher disease and keep families and medical advisers up to date with the latest developments.
- Encourage the availability of treatment including enzyme replacement therapy.
- Keep families in touch for support.

- Actively promote medical research into Gaucher disease.

**c) Need for this study –** With the development of treatment for Gaucher disease, patients are living longer. However with the increase in survival, new manifestations of the disease are being seen such as mental health issues, challenges with employment and independence. This need to be understood and the scope of support required needs to be identified and documented for the Gauchers Association to be able to provide the necessary support that members and families need.

**d) Patient advocacy for similar conditions –** Gaucher disease is one of a group of diseases called 'Lysosomal Storage Disorders (LSDs)', two other charities that support patients and their families with an LSD are the MPS Society and the Niemann-Pick Disease Group (UK). Both these charities offer advocacy services; a description of these is given below for information.

**The MPS Society** provides, through a team of skilled staff, an individual advocacy support service to its members. The service is flexible and a wide range of support is offered, led by members' requests. Individuals affected by and families caring for a child or adult with an MPS or related disease experience specific problems and difficulties arising from the rarity of this group of diseases. This rarity means that the vast majority of social care and health professionals know very little if anything about the diseases and without such knowledge are unable to make accurate assessments to include both short and long term needs. Specific needs of MPS Society members may also be neglected by policies, which do not uphold these. The rights of sufferers and their families may also be neglected or undermined through policies and practices which do not address the multi-systemic nature of these diseases, sufferers of whom are increasingly living into adulthood and for whom standard provision is often unsuitable. The requirement for social services provision to meet clinical needs such as gastrostomy feeding and transition from family-centred children's palliative care services to those provided for adults are two examples, which demonstrate these challenges.

**The Niemann-Pick Disease Group (UK)** employs a full time Clinical Nurse Specialist for Niemann-Pick Disease that offers a dedicated service to families. As the disease progresses, those affected can develop complex needs that can change rapidly. The service provides support to the whole family, ranging from friendly advice over the phone to home visits, or liaising with other care professionals to ensure the families receive the help they need.

Other LSD charities including the Association for Glycogen Storage Diseases and Batten Disease family Association also offer advocacy and support services to their members and families.

**e) Current status of patient advocacy in the UK for Gaucher disease –** current provision for patient advocacy is carried out by the Association's Chief Executive as and when requests are made by members for support. The Chief Executive is the mother of a Type III patient and has developed the role using her personal experience. However, this is limited and mainly focuses on emotional support.

**Aims of the Project:**

To explore with patients, families and medical professionals their thoughts about the remit and benefits of a permanent advocacy post, looking in particular at what is – or is not - happening currently and what they would, ideally, like to see available, with an emphasis on newly diagnosed and Neuronopathic Gaucher disease (nGD).

**This will be achieved by**

Collating and analysing information on past experiences of families/patients, to highlight common issues and identify areas of weakness/unmet need to be addressed for future patients and their families. Such factual information will also strengthen our ability to develop productive and complementary relationships with statutory service providers and other relevant agencies. Seeking families' views on how these issues can be addressed; in doing so helping them understand what their choices are, particularly in relation to issues around social care and health services and education.

**How the information will be collected**

***Home visits with families/patients***

- Conducting semi-structured interviews to gain qualitative information
- Exploring experiences, needs and anxieties about the present and future for their child/themselves and the wider family.
- Extending/enhancing information gathered through other face-to-face meetings telephone and/or e-mail follow-up
- Other structured questionnaires, utilising the Association's interactive website facility
- Focus groups, run primarily at other regional, national and international conferences/meetings re GD, optimising on engagement with all GA families and medical/healthcare professionals

***Visits to the Seven LSD Centres and other social care and health and education providers***

- Meeting each of the professionals at the Centres of Excellence involved in the clinical management of patients to identify/discuss issues faced by patients/families from the professionals' perspective.
- Focus groups, run primarily at other regional, national and international conferences/meetings re GD, optimising on engagement with all GA families and medical/healthcare professionals
- Conducting interviews with Social services, palliative care, schools, mental health teams who have had experience of working with Gaucher patients and their families in the community to identify/discuss issues faced by patients/families from the professionals' perspective

**What will be done with the information?**

The information collected during the project from the interviews will be written up into a formal report and also for publication in an appropriate journal. The report will make a series of recommendations which will be presented to the Board of Trustees of the Gauchers Association for consideration to enable the appropriate service to be developed to support members and their families. The information will also be shared with the eight designated LSD centres in England and those physicians and clinical staff and service providers who support Gaucher patients in the rest of the UK to ensure a coherent service is provided.