



Lysosomal storage disorders (LSDs) are a group of approximately 40 rare inherited metabolic disorders that result from defects in lysosomal function. Lysosomal storage disorders result when a specific organelle in the body's cells - the lysosome - malfunctions. All lysosomal storage disorders share a common pathogenesis: a genetic defect in a specific lysosomal enzyme, receptor target, activator protein, membrane protein, or transporter, leading to accumulation of substrates in cell lysosomes.

UK LSD Patient Organisation Collaboration

Patient Organisations representing those affected by Lysosomal Storage Disorders have joined together to form a new action group to work and lobby on behalf of LSD patients and their families in the UK.

The Group is made up of representatives from the Association for Glycogen Storage Disease, Batten Disease Family Association, The Gauchers Association, The Society for Mucopolysaccharide Diseases (the MPS Society), and The Niemann-Pick Disease Group (UK).

Our Mission Statement:

To undertake joint promotion and a shared understanding of LSDs, to advance standards of care and to enhance the wellbeing of those affected. To stimulate interest and work in partnership to establish a Forum in which members can discuss together matters of common interest and contribute to the development and dissemination of good practice amongst them.

LSD UK Patient Organisation Secretariat

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Objectives:

- To enable the development of a stronger voice with which to influence national policy, stimulate interest in and further the knowledge and understanding of, Lysosomal Storage Disorders.
- To encourage close working relationships between Member Associations, in order to facilitate the sharing of information and the effective use of resources.
- To oversee the provision of clinical care for those affected by LSDs; to promote clinical efficiency and best practices in the health service.
- To share information regarding potential therapies and treatments for LSDs.
- To consider the cost implications of potential treatments and therapies; to support and encourage all those affected by LSDs in accessing treatments and therapies.
- To seek to provide a positive influence, leading to increased engagement in the delivery of government policy on LSDs and other rare diseases.
- To consider ways of encouraging new initiatives; including the commissioning of further research into the causes and possible treatments of LSDs.

To be achieved by:

- The dissemination of ideas, examples of good practice and other relevant information to researchers/health care professionals in the UK.
- The provision of considered and co-ordinated advice and information regarding LSDs to all interested parties.
- The establishment of strong links with industry, researchers, clinicians and individuals with an interest in LSDs.
- The implementation of a well-organised campaign to highlight the need for effective, accessible and safe therapies for LSDs and better need for health and social care.
- Lobbying the government and other relevant agencies for improvement in clinical standards of care for those affected by LSDs, including; the early and accurate diagnosis of, and screening programs for, LSDs.
- Through combined activity, the provision of an effective contribution to the updating and ongoing improvement of the highest possible standard of clinical care.
- Working in partnership to raise awareness of LSDs amongst health and social care professionals and the general public.

In 2009/2010 our work programme will include the following:

To assist in the development of a patient group for Metachromatic Leukodystrophy (MLD)

To organise a family day to bring families together and then support the development of a Metachromatic Leukodystrophy/Leukodystrophy Patient Organisation

The development of a Clinical Study Group for Inherited Metabolic Disorders in partnership with the Medicines for Children Research Network (MCRN)

To develop an action plan for the continued designation of LSDs as a specialised service post 2012

To organise a half day workshop on Transition for LSD patients and their families

To organise an LSD patient meeting in Scotland and Northern Ireland

Who are the Members of the UK LSD Collaborative?

Association for Glycogen Storage Disease: *Allan Muir*

The Batten Disease Family Association: *Jan Sablitzky*

The Gauchers Association: *Tanya Collin-Histed*

The Society for Mucopolysaccharide Diseases (MPS Society): *Christine Lavery*

The Niemann-Pick Disease Group (UK): *Toni Mathieson*

Join Us:

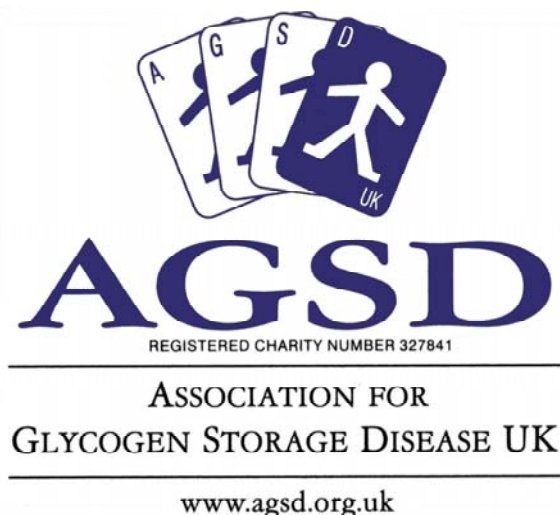
We are keen to open membership of the Group to other LSD conditions.

If you would like more information contact:

Tanya Collin-Histed, Executive Director, The Gauchers Association,

by telephone: 00 44 1453 549231, or e-mail at Tanya@gaucher.org.uk

The Association for Glycogen Storage Disease (UK)



The Association for Glycogen Storage Disease (UK) provides support and help for families affected by any one of the GSDs by providing information, issuing Newsletters and holding Conferences and Workshops.

Glycogen Storage Disease (GSD) occurs when there is an absence or deficiency of an enzyme needed to produce or break down glycogen in the body. GSD primarily affects the liver and/or muscles.

Pompe disease, also known as GSD Type II, or Acid Maltase Deficiency, is a rare neuromuscular genetic disorder that occurs in babies, children, and adults who inherit a defective gene from each of their parents. Progressive muscle weakness is the most common symptom of both the infantile-onset and the late-onset forms of Pompe disease. The muscles most often affected are those used for breathing and mobility. In infants, the heart muscle is often severely affected and without treatment can lead to death within the first two years of life.

Pompe disease is also a Lysosomal Storage Disease, which is why the Pompe Group of the AGSD-UK is proud to be a member of the UK LSD Patient Organisation Collaboration.

For further information about Pompe Disease and the AGSD (UK) please contact:

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Batten Disease Family Association

Batten Disease is one of the main paediatric neurodegenerative disorders in the UK affecting 150-200 children and young adults. Children who inherit this rare disease are born seemingly healthy but then develop epilepsy, lose their ability to see, talk, walk, eat and develop dementia before dying between the ages of 5 - 30 depending on what type of Batten Disease they have. At present there is no cure. Having Batten Disease profoundly changes the child's and their family's life forever.

Being a rare disease, the families of Batten Children are left feeling isolated and numb as most professionals have not heard of it and do not understand the course of the disease and the needs the child and families have. This has clearly been identified in the audited report of existing support and services: 'Batten Disease Family Support Project: Final Report' completed in 2008, commissioned by the BDFA and funded by Jeans for Genes.

The BDFA was founded ten years ago and is the only dedicated UK Batten Disease charity. Our vision is to be the central point of excellence in the UK for support, guidance and networking of families and professionals affected by Batten Disease as well as to increase vital awareness and national resources and facilitate research into Batten Disease.

We have a dedicated and growing team of volunteers and part-time paid support to manage and develop our activities to support families and professionals in communities across the UK. These now include: providing a helpline for carers and professionals; maintaining a website and forum; networking affected families and professionals; working with Batten's specialists as our professional advisors; providing educational conferences, training events and supportive information resources; providing updates on research developments; increasing awareness of Batten Disease



Bringing light to Batten's amongst the public, medical and research world; lobbying for better resources for Health, Social Care and Education; funding research into understanding the cause and progress of the disease and facilitating the development of potential therapies or new methods of treatment.

For further information about the BDFA, please contact:

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The Gauchers Association



Registered Charity No: 1095657

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.

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 stomach may also occur as well as bone pain, demineralisation and fractures.

People with Gaucher disease lack sufficient activity levels of an enzyme called glucocerebrosidase. The enzyme helps the body break down worn-out cells and as a result of the enzyme deficiency, a fatty substance called glucocerebroside accumulates 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 Jewish (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 which 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.

For further information about the Gauchers Association, please contact:

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The Society for Mucopolysaccharide Diseases

The Society for Mucopolysaccharide Diseases founded in 1982, is a voluntary support group which from throughout the United Kingdom represents the interests of over 1200 children and adults suffering from Mucopolysaccharide and related Lysosomal Storage diseases.

The MPS Society supports affected individuals and their families through a dedicated Advocacy Service providing support in areas of access to clinical management and treatment, home adaptations, special educational needs, respite care, palliative care, pre and post bereavement.

The MPS Society holds a National Weekend Conference for MPS Families and professionals every two years and regional conferences in Scotland in the intervening years. As funds allow, a programme of local events throughout the UK are organised.

Over the past 25 years the MPS Society has made grants for research exceeding £7,000,000 and currently support the

work of the MPS Stem Cell Group at the University of Manchester and the Blood Brain Barrier Group at Kings College, London.

MPS IH Hurler
MPS IHS Hurler Scheie
MPS IS Scheie
MPS II Hunter
MPS III Sanfilippo
MPS IVA Morquio
MPS VI Maroteaux Lamy
Geleo Physic Dysplasia
MPS VII Sly
MPS IX Natowicz
MLI Neuramidase Deficiency
ML II I-Cell
ML III Pseudo Hurler Polydystrophy
ML IV
Fucosidosis
Fabry Disease
Mannosidosis
Sialic Acid Storage Disease
Sialidosis
Multiple Sulphatase Deficiency
Aspartylglycosaminuria
Winchester Syndrome
GM1 Gangliosidosis (MPS IVB)



For further information about the MPS Society, please contact:

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Registered Charity No. 287034

The Niemann-Pick Disease Group (UK)



Registered Charity No. 1061881

The Niemann-Pick Disease Group (UK) is a charitable organisation offering care, support and information to families affected by Niemann-Pick diseases. The main aims and objectives of NPDG (UK) are; to make a positive difference to the lives of those affected by Niemann-Pick diseases (NPD), relieve sickness and any distress which may arise there from, and to advance the education and awareness of families, professionals and the general public in all matters concerning the disease.

Niemann-Pick is the name given to a group of rare, inherited, life-limiting diseases, of which there are two recognised forms; Niemann-Pick disease, Type A/B (or ASMD Niemann-Pick Disease), and Type C. Niemann-

Pick Type A and Type B are caused by an enzyme deficiency, causing a build up of toxic materials in the body's cells. In Type A this accumulation occurs very quickly, an affected child will usually die before reaching three years of age. Type B does not usually affect the brain, and, although growth may be slow, those affected will survive into adolescence or early adulthood, but not without experiencing health problems.

Niemann-Pick Type C is not caused by an enzyme deficiency, but the end result is the same; an accumulation of materials (cholesterol and other fatty acids) in the body's cells. The brain and other organs are affected, leading to progressive intellectual decline, loss of motor skills, seizures and dementia. Speech can become slurred and swallowing problems may develop, in the later stages the child will be bedridden and tube-fed. Affected children do not usually survive into adulthood.

The Group funds the salary of a full time Clinical Nurse Specialist, providing expert care and practical advice, plus home visits whenever necessary. Genetic counselling and advocacy services are also provided. To further support families, the Group holds

an Annual Family Conference, which aims to address the needs of all those affected by Niemann-Pick diseases, their families and the health and social care professionals involved in their care.

In 2006, the Group employed a Clinical Research Nurse to enable the collection of much needed data regarding clinical aspects of this group of diseases. This post has the potential to improve the understanding of Niemann-Pick diseases, stimulate research and assist in the development of therapies. The NPDG (UK) also offer an annual Trustee Memorial Award to young professionals who are able to provide an original contribution to the scientific or public understanding of the Niemann-Pick diseases and/or their treatment or cure.

For further information about the Niemann-Pick Disease Group (UK) please contact:

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