

Collaborative working to advance standards of care and the well being of lysosomal disorder patients and families in the UK



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WORKING TOGETHER SHAPING SERVICES FOR PATIENTS

Gauchers Association, Gloucestershire, UK ¹
 The Society for Mucopolysaccharide Diseases (MPS Society), Buckinghamshire, UK ²
 Niemann-Pick UK (NPUK), Tyne and Wear, UK ³
 Krabbe UK, Leeds, UK ⁴
 AGSD-UK, (Pompe), Southampton, UK ⁵
 Batten Disease Family Association, Farnborough, UK ⁶
 The CATS Foundation, (Tay Sachs), London, UK ⁷

In 2005 following the designation by the Department of Health of central funding for enzyme replacement therapy (ERT) for patients with lysosomal storage disorders (LSDs) in England, seven registered patient associations representing Batten, Gaucher, Fabry & MPS, Krabbe, Pompe, Niemann-Pick, and Tay-sachs agreed to work together to improve standards of care for those affected by LSDs.

The LSD Collaborative has a Facebook page and website. Members of the Collaborative are the Chief Executives of the associations and each took on a responsibility in representing the patient voice on national stakeholder groups for commissioning, homecare, drug framework development and the Expert Advisory Group (EAG) meetings with NHS England and the eight LSD Centres of Excellence responsible for the clinical management of all LSD patients in England.



Future Proposals

Description:
 The Collaborative have an annual business plan which is a live document and is updated each time it meets, that sets out our annual plans.

- Outcomes:**
- To develop a set of disease models for LSDs that will assist in health economics.
 - To promote the use of telephone clinics to support LSD families whose children have moved to palliative care.



Clinical Reference Group

Description:
 A multi-stakeholder group led by NHS England Commissioning to review and recommend new services, treatments, and guidelines

- Outcomes:**
- A member of the Collaborative has had a seat on the CRG for 4 years

National Centres of Excellence

Description:
 8 clinical centres in England where expert multi-disciplinary teams are based that clinically manage all patients with LSD's, prescribe treatment and run clinical trials

- Outcomes:**
- Annual meetings between the Collaborative and each centre once a year for updates and discuss challenges and opportunities for joint working i.e. patient satisfaction survey
 - New patient information card for newly diagnosed patients
 - Operate multi-disciplinary clinics for some diseases
 - Patient advocacy representatives attend clinic in the centres to support patients and family members/carers



Future Proposals

Clinical Reference Group

National Centres of Excellence

Mental Health Workshop

Description:
 Setting up and running a one-day workshop to raise the issue of mental health challenges in patients and parents with LSDs, followed by a survey of patients and parents to identify the challenges they face and their experience of access to services

- Outcomes:**
- All National Centres were represented at the workshop
 - Recognition of the need to include 'think mental health' in clinic with patients
 - The sharing of best practice across the centres

Expert Advisory Group

Description:
 A bi-annual meeting of doctors from the 8 national centres of excellence with NHS England and representatives of the patient organisations.

- Outcomes:**
- A maximum BMI of 27 set for dosing ERT
 - Standard Operating Procedures for all Diseases
 - A national drugs tender to achieve cost efficiencies
 - Collaborative research studies
 - Standardisation of Hospital to Home for Home Therapy i.e. Type 3 Gaucher infusions

Expert Advisory Group

Transition Pathway

Description:
 An NHS England funded piece of research to look at the transition processes in place when a young person with an LSD moves from paediatric to adult health services and to make recommendations to improve and develop this service in the best interests of the young people who are approaching this period of transition in their lives.

- Outcomes:**
- Improved clinical outcomes
 - Improved patient outcomes
 - Improved process time in the patient journey
 - Patient focused booklet explaining transitioning for those affected by LSDs

Transition Pathway

Medics4Rare Diseases

Description:
 Targeting medical professionals to increase awareness of rare diseases especially around diagnostics. M4RD will supersede students4rare diseases which was an initiative of medical students from Barts in London, UK, in 2011, to educate medical students about rare diseases.

S4RD was supported by the Collaborative to access funding to further the aims of this project and to transition into Medics 4 Rare Diseases which is now its own organisation

- Outcomes:**
- Interest from medical students at 33 medical schools
 - 6 medical schools holding rare disease events in England, Wales and Ireland
 - An iconic brand, a zebra with a stethoscope
 - Employment of an educational co-ordinator
 - Face, twitter and website sites
 - Annual symposium and evening seminars on rare diseases with parent/patient involvement
 - Interest in this model from medical students in Prague, Beirut and the University of Western Australia

Diagnostic Workshop

Description:
 A one-day workshop initiated by the LSD Collaborative and developed in conjunction with lead scientists to discuss the current challenges in diagnostics i.e. access to materials, succession planning for staff and to work jointly to resolve the issues to benefit both labs and patients.

- Outcomes:**
- Representation from the key UK laboratories
 - Agreement for an annual meeting
 - A paper to outline the challenges and some recommendations to present to NHS England for future service development



LSD Collaborative

Diagnostic Workshop

Homecare Framework

Description:
 A national framework for homecare services to all LSD patients across England, Scotland and Wales, overseen by NHS England and developed by representatives from the 8 National centres and a representative of the LSD Collaborative

- Outcomes:**
- Standardised services and care for patients
 - A drive to increase cost efficiency and care
 - An increase in patient independence through training to become self-infusers
 - Different levels of nursing care to support individual needs
 - The majority of LSD patients receive their treatment at home

LSD Patient Survey

Description:
 An independent on line and paper survey sent to all LSD patients and parents/carers to understand their access to information, services and the quality of their experience when using the national centres of excellence and homecare services

- Outcomes:**
- A total of 264 respondents across the diseases
 - Feedback broken down for each centre
 - Feedback broken down for each homecare company
 - Feedback broken down for each disease/patient organisations
 - This will now be an annual survey



Contact Information

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