# The LSD Collaborative:

## General Election Manifesto 2019

AGSD UK Association for Glycogen Storage Disease · BDFA · CATS Cure & Action for Tay-Sachs Foundation · Gauchers Association · Krabbe uk · MPS Society Transforming lives through support, research and awareness · npuk


## Our Key Messages:

**The LSD Collaborative calls on the next government to commit to the following pledges:**

1) No child or adult with a Lysosomal Storage Disorder in the UK should die due to lack of funding for the available treatment that could save their life.
2) Medicine manufacturers should strive to provide safe, effective and affordable treatments for LSDs
3) That full consideration be given to the imbalance between the very few disorders screened for at birth in the UK, compared to the number of disorders screened for in other European countries and the rest of the World.

If the next government fulfils these pledges then each year the lives of over 100 newly diagnosed children and many adults living with LSD conditions will lead happier and healthier lives.

## Who is the LSD Collaborative?

The UK LSD Patient Collaborative Group is comprised of patient organisations representing those affected by Lysosomal Storage Disorders (LSDs). We have joined together to form an action group to advocate and work on behalf of LSD patients and their families in the UK. The group is made up of representatives from 7 charities in the UK who provide support and information to patients and families affected by different LSDs. Together we represent over 70 rare and ultra rare diseases and almost 2500 patients and their families.



## Our Mission Statement:

To undertake joint promotion and a shared understanding of LSDs, to advance standards of care and to enhance the health and 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.

AGSD UK Association for Glycogen Storage Disease · BDFA · CATS Cure & Action for Tay-Sachs Foundation · Gauchers Association · Krabbe uk · MPS Society Transforming lives through support, research and awareness · npuk

## Introduction:

**Why we have a manifesto:** Lysosomal Storage Disorders (LSDs) are a subset of rare diseases which are often overlooked.

**What we want our manifesto to achieve:** We ask those who support us and those we serve to make sure their prospective parliamentary candidates are aware of this manifesto and to commit to ensuring that their party commits to the actions, requests and recommendations.

## What is a Lysosomal Storage Disorder (LSD)?

Lysosomal storage diseases are inherited metabolic diseases that are characterized by an abnormal build-up of various toxic materials in the body's cells because of enzyme deficiencies. There are more than 70 of these disorders altogether, and they may affect different parts of the body, including the skeleton, brain, skin, heart, and central nervous system. New lysosomal storage disorders continue to be identified. While clinical trials are in progress on possible treatments for some of these diseases, there is currently no approved treatment for many of the lysosomal storage diseases.

## Pledge 1) No child or adult with a Lysosomal Storage Disorder in the UK should die due to lack of funding for the available treatment that could save their life:

LSDs are a group of complex diseases, many of which have no known cure and few treatments. Access to new treatments depends on where in the UK a person lives and is subject to rigorous evaluation. While appraisal processes are necessary for safety, the current system has tilted towards an over-emphasis on the cost of treatment that discriminates against rare and ultra rare diseases such as the LSDs. This needs to be rebalanced. The processes can be protracted and, in some cases, this has led to people needlessly dying. These are largely new treatments for diseases where no medicines or therapies were previously available. A rigorous and transparent evaluation process is always welcomed.

The knowledge that there are other countries in which treatment is available – in some instances even in neighbouring countries in the UK - places families under a huge amount of emotional and mental strain. Families often resort to drastic measures including moving abroad in order to access treatment.

We believe that there are straight forward measures that the new government can take to achieve this pledge.

**To achieve this, there needs to be:**

• A dedicated Rare Disease Drug Fund, similar to the Cancer Drug Fund, that is dedicated to funding new treatments for LSDs and other rare disease as this is proven to increase access to innovative treatments

• Parliamentary scrutiny of the current NICE review of their drug appraisal process to ensure it does not discriminate against ultra rare diseases such as LSDs in England.

• Understanding of the impact of not funding available treatments for LSDs including early death, irreversible damage and disability, such factors as families leaving England for treatment elsewhere, the total burden on the public purse, and long-term mental health issues within affected families.

• Equality of access to treatment across all Nations of the United Kingdom.



## Pledge 2) Medicine manufacturers should strive to provide safe, effective and affordable treatments for LSDs:

New treatments and medicines are the result of research and development mostly undertaken by private pharmaceutical companies. There has been a number of new treatments that have not been approved or that have taken a protracted time to be agreed, allegedly due to issues with pricing set by the manufacturer. It requires government intervention in order to make sure that all patients in the UK can receive safe and effective treatments at prices that are affordable to the health system.

**To achieve this, there needs to be:**

• Encouragement by the UK Government for medicine manufacturers to be innovative in their approach and to strive to provide only the most effective and affordable treatments for LSDs.

• Parliamentary oversight of commercial arrangements should be made as part of the overall health system in each of the devolved Nations.

• Creation of an environment that encourages all parties to be transparent in their processes to benefit patients and those who care for them.

### What is Newborn Screening?

Early diagnosis of disease at birth in the UK is done by taking a blood test from the infant, generally on the 5th day of life. This is known as the 'heel prick' test. The heel prick bloodspot test is a tried and tested method and does no harm to the infant. The bloodspot sample is then screened in newborn screening laboratories to identify severe disorders and life limiting diseases which are difficult to detect clinically at this stage in an infant's life. Early detection of a disorder at birth allows clinicians to give immediate treatment to the child, preferably before any symptoms appear. Treatment at this earliest opportunity can prevent physical disability, neurological disability and death. Improved treatments in recent years for some metabolic disorders, including LSDs, have highlighted the real benefits of early recognition of these disorders. Simultaneously, the analytical potential to screen for such conditions has offered an opportunity to respond to this need.

## Pledge 3) That full consideration be given to the imbalance between the very few disorders screened for at birth in the UK, compared to the higher number of disorders screened for in other European countries and the rest of the World:

In comparison to many other countries the UK has a very disappointing record in preventing disability and death in children by screening at birth. The first disease screened for nationally in England was PKU in 1969. Since 1969 only 8 additional disorders have been added to the UK national screening programme. Countries across Europe and other countries of the World screen for many more conditions. (See Table of figures at Annex 1)

All Countries follow the World Health Organisation criteria for determining which disorders are appropriate for screening at birth. It remains unclear why the UK has such a disappointing record when screening for more diseases could save the lives of children and prevent disability, irreversible damage and death. Legislation has been introduced in Italy that every child born should have the right to be screened for almost 40 conditions for which there is a viable treatment. (Bill no.167/August 2016). The law is written in such a way to ensure that when new treatments become available, the list of disorders included in the screening programme will be continuously updated.
Indeed a meeting was held in the EU Parliament on 30th January 2019 to welcome the introduction of the Italian model and to discuss how this model might be adopted by all EU member states. We believe that that children born in the UK should similarly have the right to be screened.

**To achieve this, there needs to be:**

• An All Party Parliamentary Group to consider why there is such an imbalance in the number of diseases screened for at birth in the United Kingdom in comparison to Europe and the rest of the World and to make recommendations to Parliament on how the situation across the UK can be improved. The UK LSD Collaborative Group are willing to take a supportive role to an All Party Parliamentary Group

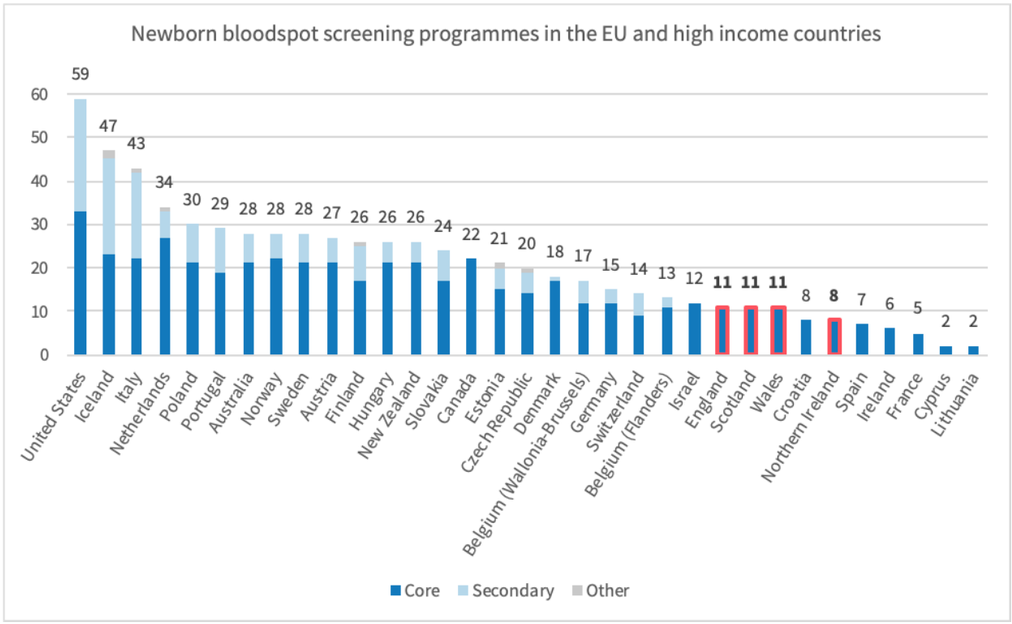
• Greater support from the UK Government for scientists, clinicians and patient organisations who are making submissions to the UK National Screening Committee proposing an additional disorder to the UK newborn screening programme.



• Consideration of the structure of the current decision making committees on newborn screening to ensure participation of members with expertise in childhood metabolic disorders.

• A real commitment by Government to include the need for improvements in a future Rare Disease Framework.

**Appendix A:**




Note: To allow for comparison the nomenclature used in the United States Recommended Uniform Screening Panel has been applied. Sickle Cell disease is listed as 3 separate conditions which accounts for the UK panel of 9 being increased by 2.

Secondary Conditions are those which are detected when testing for the Core or Primary Conditions. These are included where the testing results are reported to the family.

Other conditions are those which are only screened for by one country, often because of local priorities. These are difficult to categorise as core or secondary.

*Document Source: Genetic Alliance UK. Fixing the Present - building for the Future. July 2019.*


AGSD UK
Association for Glycogen Storage Disease

BDFA

CATS
CURE & ACTION FOR TAY-SACHS FOUNDATION

Gauchers ASSOCIATION

Krabbe uk

MPS

MPS Society
Transforming lives through support, research and awareness

npuk

**For further information please visit: lsdcollaborative.org.uk**