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NICE
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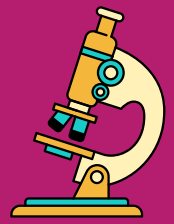
Our organisation was established in **1981** by Peter and Lesley Greene following the diagnosis of their daughter, with cystinosis, a rare IMD after finding little support or information for people living with IMDs.

Today we support the 1.43 million people living with one of over 2000 inherited metabolic disorders worldwide.Our community makes up almost 20% of the all rare diseases.

Our values are:
Positive Disruption, Bridging Connections, Enduring Support

Individual Support



Insight & Advocacy

Community Building

Comms & Campaigns

METABOLIC SUPPORT

Living well with a rare disease

What are inherited metabolic disorders (IMDs)?

METABOLIC SUPPORT
Living well with a rare disease

IMDs are a large group of rare genetic conditions caused by defects in enzymes or proteins involved in metabolic pathways. These defects disrupt biochemical processes, leading to the accumulation of toxic substances or deficiencies of essential compounds required for normal body function.

IMDs can result in serious health complications, including developmental delay, neurological impairment, and organ damage. Management often requires lifelong treatment, such as specialised diets, medications, enzyme replacement therapies, or organ transplantation. Early diagnosis, particularly through newborn screening, is essential to improve outcomes (7/10 of UK newborn screening panel are IMDs).

We are heavily involved with varied medicines consultation including:

FCS: Health Inequity & Treatment Choice - treatments that work for diverse communities
LC-FAODs: Currently under Early Access Scheme
Early Access Scheme Different Outcomes: Hypophosphatasia & Batten Disease
Nephropathic Cystinosis: Disparity across devolved nations

Examples of Inherited Metabolic Disorders

TMAU

Trimethylaminuria

- Impaired breakdown of trimethylamine causing odour symptoms, often diagnosed late
- Not life-threatening but life-altering with significant psychosocial impact.
- Managed mainly through diet and lifestyle, with limited specialist support.

OTCD

Ornithine Transcarbamylase Deficiency

- Urea cycle disorder affecting how ammonia is processed in the body.
- Acute onset, often life-threatening in infancy.
- Requires emergency management, dietary restriction, and specialised hospital care.
- Gene Editing & Gene Therapy Trials

Driving access to transformative treatments and supporting a thriving life sciences industry…what does this mean to our patient community?