## Overview of MLD and Clinical Trails

**Prof Tim Cox MD, FRCP, FMedSci**
Emiritus Professor of Medicine, University of Cambridge. Director of Research and Honorary Consultant Physician at Addenbrooke's Hospital, Cambridge

All Lysosomal Storage Diseases have common factors

- It occurs when the body recycles cell contents and components
- When things go wrong the 'junk' accumulates and causes symptoms
- They are multi system diseases
- Nurodegenerative diseases have strong genetic determinants
- The need to overcome theraputic obstacles
- Progressive disease of the myelin leads to disease of the white matter in the brain
- In MLD there is a failure to metabolise Sulphatide

MLD is a rare disease, an orphan disease.

There is a massive current need and this is expressed by lobbying, fundraising, agitating for research and understanding treatment.

Haematopoietic stem cell transplantation is available for adult-onset MLD, but early intervention is essential to be beneficial.

Unrelated umbilical cord blood transplant can be used for pre-symptomatic juvenile MLD

A Clinical trial for Gene Therapy for MLD is currently taking place in Milan, Italy. They are recruiting pre-symptomatic children with a confirmed diagnosis of Late Infantile MLD or Juvenile MLD. It is often the case that an older sibling is diagnosed after displaying symptoms, and the younger sibling is tested prior to symptoms appearing.

Other possible therapies:

Stem cells

Enzyme Replacement Therapy

Substrate reduction therapy – that is, not increasing production of the missing enzyme, but taking away the waste product produced i.e. the sulfatides which cause de-myelination.