Living with MLD

(Metachromatic Leukodystrophy)

Information for Families affected by MLD

mld
SUPPORT
ASSOCIATION UK

This leaflet has been produced to provide information on MLD (Metachromatic Leukodystrophy) for families affected by this disease.

What is MLD?

MLD is an acronym for Metachromatic Leukodystrophy. MLD is an autosomal recessive genetic disorder which at the moment has no cure. MLD is directly caused by a deficiency of the enzyme Arylsulfatase-A. Without this enzyme sulfatides build up, eventually destroying the myelin sheath of the nervous system. The myelin sheath is a fatty covering that protects nerve fibres. Without it, the nerves in the brain and the peripheral nerves cease to function properly.

What is the Incidence of MLD?

There are many forms of Leukodystrophy, but Metachromatic Leukodystrophy (MLD) is one of the most common forms. The incidence of MLD is now estimated to occur in 1 case in 40,000 live births. However, with modern diagnostic tools such as MRI Scans and genetic sequencing, it means that there are fewer incorrect diagnoses and it is possible that the incidence may prove to be higher.

What are the Symptoms and Types of MLD?

There are several forms of MLD, which are generally classified as **late-infantile, Early-Juvenile** and **Juvenile,** and **Adult-Onset.**

Late Infantile MLD

In late-infantile MLD, after a period of normal growth and development, affected children experience difficulty when they are crawling or walking, usually at 15-24 months. Further deterioration then occurs, including muscle wasting and weakness, muscle rigidity, developmental delays, progressive loss of vision leading to blindness, convulsions, impaired swallowing, paralysis, and dementia. Most children with this form of MLD die by age 5, often much sooner.

Once symptoms have appeared it is too late for any treatment. Currently, a trial is on-going in Milan for Gene Therapy for children who are pre-symptomatic.

Early Juvenile and Juvenile MLD

Children with a juvenile form of MLD (onset between 3 and 16 years of age) often show impairments in fine motor skills with increasing difficulties with balance, movement, co-ordination and walking. They can also develop behavioural problems, particularly at school. They then develop symptoms similar to the late-infantile form, but with slower progression. Age of death is variable, but normally within 10 to 15 years of symptom onset.

Gene Therapy is currently undergoing trials in Milan for children who are diagnosed with Early Juvenile or Juvenile MLD before symptoms appear.

Adult-Onset MLD

The adult form of MLD commonly begins after age 16 and is often misdiagnosed as a psychiatric disorder because of personality changes. Initially, the symptoms are cognitive rather than physical, leading to progressive dementia and, ultimately, physical disability similar to the earlier onset forms. Adult-Onset MLD progresses slowly with a protracted course of a decade or more.

Sufferers of Adult-Onset MLD can benefit most from a Bone Marrow or Stem Cell Transplant. In the UK and in the USA there are now sufferers who have had transplants who are showing little or no degeneration after 25 years.

The Dangers of Anaesthesia?

Over the last 25 years, it has been shown that general anaesthesia can cause degeneration in sufferers of MLD. It is advised to avoid the use of gas anaesthesia as most deterioration has been seen following this. Ensure that you speak to the Anaesthetist personally and see if a General Anaesthetic can be avoided. If it cannot, then ask them to use the smallest amount of anaesthesia for the least amount of time. It is preferable to use local anaesthesia. Ask them to treat your child or affected adult as they would someone with a head injury, Multiple Sclerosis or Cerebal Palsy. These are all conditions anaesthetists are familiar with. Also, it will probably take your child or affected adult a little longer to recover from an anaesthetic.

MLD Support Association UK was set up by two families who have a child with MLD. We understand how life-shattering the diagnosis can be, and how alone you are feeling. We are here to help.

About us

MLD Support Association UK was set up to bring hope to families in the fight to eradicate Metachromatic Leukodystrophy (MLD). We aim to provide support to families, personally, through our Website and Facebook group, and at Family Conferences and Fun Days. This enables families to share their experiences of living with MLD.

MLD is a rare disease and health professionals know little about the symptoms and outcomes. MLD Support Association UK is committed to providing information to help with a correct diagnosis and also to provide details of on-going care or treatment options.

We are currently working on a Registry of all known cases of MLD in the UK. Patient information in this registry will be used to create a natural history of MLD types for research and experimental clinical trials.

MLD Support Association also organises a bi-annual Scientific Conference where scientists and clinicians who specialise in Leukodystrophies can meet up to learn about current treatment and research.

Information for Families affected by MLD

At present MLD has no cure. In pre-symptomatic or mildly symptomatic adults a Bone Marrow or Stem Cell Transplant may be an option. Gene Therapy is currently being trialled in Milan and they are recruiting children who are pre-symptomatic but with a confirmed diagnosis of Late Infantile MLD or Juvenile MLD.

At MLD Support Association UK we can help all families living with MLD by providing information and access to a specially selected team of health professionals.

Contact us

MLD Support Association UK
Floor 5, Amphenol Business Complex
Thanet Way, Whitstable, CT5 3SB
Tel: 07414 529392
Email: admin@mldsupportuk.org.uk

www.facebook.com/groups/MLDsupportUK

www.mldsupportuk.org.uk

mld SUPPORT ASSOCIATION UK

Registered Charity Number 1150542