

Living with MLD

(Metachromatic Leukodystrophy)

**Information for Families
affected by MLD**





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 and 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. At the moment the only treatment approved by the NHS is Gene Therapy, but this is not available for all forms of MLD.

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 **Late-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.

Unfortunately, once symptoms have appeared it is too late for any treatment. Gene Therapy is now available at specialist centres on the NHS. This treatment has proved effective, but only for children who are pre-symptomatic.

Early-Juvenile and Late-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, coordination 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.

Currently, Gene Therapy is available at specialist centres on the NHS, but only for children with Early-Juvenile MLD who are pre-symptomatic or early-symptomatic. A clinical trial is currently being carried out in Milan for children who are pre-symptomatic or early-symptomatic with Late-Juvenile MLD.

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 30 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 Cerebral 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 Database of all known cases of MLD in the UK. Patient information in this Database will

be used to help us keep in touch with all affected families.

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 the moment the only treatment approved by the NHS is Gene Therapy, but this is not available for all forms of 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

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