Understanding MLD (Metachromatic Leukodystrophy)

Information for Schools, Nurseries, Paediatric clinics and GP Surgeries.
Information for Health Professionals

This leaflet has been produced to provide information on MLD (Metachromatic Leukodystrophy) to school nurses, nursery school assistants, paediatric clinics, GP surgeries and others coming into regular contact with children and young adults.

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 the sheath, the nerves in the brain (Central Nervous System – CNS) and the peripheral nerves (Peripheral Nervous System – PNS) cease to function properly.

Symptoms and types of MLD

There are several forms of MLD, which are generally classified as late-infantile, juvenile, and adult-onset.

Late Infantile MLD

In the late-infantile form affected children have difficulty walking after the first year of life, usually at 15–24 months. Symptoms include 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.
Juvenile MLD

Children with the juvenile form of MLD (onset between 3 and 16 years of age) often show impairments in fine motor skills with increasing difficulties with 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.

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 as well. 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 Transplant. In the UK and the USA there are now sufferers who have had a Bone Marrow Transplant and are showing little or no degeneration after 20 years.

Incidence

There are many forms of Leukodystrophy, but Metachromatic Leukodystrophy is the most common. The incidence of Metachromatic Leukodystrophy is now estimated to occur in 1 case in 40,000 live births. However, with modern diagnostic tools such as MRI scans meaning fewer incorrect diagnoses it is quite possible that the incidence may be higher.

Anaesthesia and MLD

Over the last 18 years, it has been shown that general anaesthesia can cause degeneration in sufferers of MLD. It is advised to avoid the use of gas anesthesia 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 patient 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 the patient a little longer to recover from an anaeasthetic.
About us
MLD Support Association UK was set up to bring hope to families in the fight to eradicate Metachromatic Leukodystrophy (MLD). Our aim is to provide support by way of shared information from people in similar circumstances who have already experienced the effects of the condition and/or any treatments available. We also support research into therapeutic procedures for MLD through research grants to institutes specialising in MLD research.

Finally, as MLD is a rare disease, many health professionals know very little about the disease. MLD Support Association UK is committed to providing information to ensure a correct and early diagnosis and current details of on-going care or treatment options.

Information for Health Professionals
If you are a health professional dealing with a case of Metachromatic Leukodystrophy (MLD) then we can offer information on the disease, details of palliative and therapeutic care and specialists in the field. Please contact us on admin@mldsupportuk.org.uk with your questions and we will endeavour to answer your queries, or put you in touch with the relevant specialists.

Getting Involved
We are happy to provide support and information to anyone who wishes to organise fund raising events to support the charity. We are now registered with JustGiving. Go to www.justgiving.com and you can search for MLD Support Association UK. If you are holding an event you can set up a page on their website for your supporters to give funds to our charity. If you need any help or information please contact us on admin@mldsupportuk.org.uk and we will be happy to discuss your needs.

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