MLD can progress rapidly[1] – don't miss the symptoms

MLD – an unrelenting neurodegenerative disease

Metachromatic leukodystrophy (MLD) is a rare, life-threatening, inherited neurometabolic disease that results in intralysosomal sulfatide accumulation, which causes demyelination and progressive neurodegeneration.[2,3] MLD is caused by mutations in the *ARSA* gene resulting in functional deficiency of arylsulfatase A (ARSA), a lysosomal enzyme.[2]

Know the signs and symptoms of MLD

MLD has a heterogeneous clinical presentation.[1,3]

Symptoms can include[1,4]

- Developmental delay
- Hypotonia
- Clumsiness
- Ataxia
- Psychomotor regression
- Nystagmus
- Esotropia
- Impaired speech
- Spasticity
- Seizures

Patient with MLD, diagnosed at age 6

Presymptomatic

Progressive disease

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MLD results in progression to a decerebrated state and eventually to death, typically as a result of an infection such as pneumonia.[5]

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