Metachromatic Leukodystrophy

Metachromatic leukodystrophy (MLD) is a genetic disorder caused by a deficiency of the enzyme arylsulfatase A. It is one of a group of genetic disorders called the leukodystrophies that affect growth of the myelin sheath, the fatty covering which acts as an insulator on nerve fibers in the brain. There are three forms of MLD: late infantile, juvenile, and adult. In the late infantile form, which is the most common, onset of *symptoms* (problems) begins between ages 6 months and 2 years. The infant is usually normal at birth, but eventually loses previously gained abilities. Symptoms include hypotonia (low muscle tone), speech abnormalities, loss of mental abilities, blindness, rigidity (uncontrolled muscle tightness), convulsions, impaired swallowing, paralysis, and dementia. Symptoms of the juvenile form begin between ages 4 and 14, and include impaired school performance, mental deterioration, *ataxia* (lack of muscle control), *seizures (convulsions)*, and dementia. In the adult form, symptoms, which begin after age 16, may include impaired concentration, depression, psychiatric disturbances, ataxia, tremor, and dementia. Seizures may occur in the adult form, but are less common than in the other forms. In all three forms mental deterioration is usually the first sign.

TREATMENT

There is no cure for MLD. Bone marrow transplantation may delay progression of the disease in some cases. Other treatment is symptomatic and supportive.

WHAT WILL HAPPEN

The prognosis (likely outcome) for MLD is poor. Death generally occurs within 6 to 14 years after onset of symptoms. In the infantile form death may occur between 3 and 6 years after onset.

RESEARCH BEING DONE

The NINDS supports research on genetic disorders including leukodystrophies such as MLD. The goals of this research are to increase scientific understanding of these disorders, and to find ways to prevent, treat, and cure them.

Research is continually being done to learn more about this problem.

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