Metachromatic Leukodystrophy

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Metachromatic Leukodystrophy Information Page.

Metachromatic leukodystrophy (MLD) is one of a group of genetic disorders characterized by the toxic buildup of lipids (fatty materials such as oils and waxes) and other storage materials in cells in the white matter of the central nervous system and peripheral nerves. The buildup of storage materials impairs the growth or development of the myelin sheath, the fatty covering that acts as an insulator around nerve fibers. (Myelin, which lends its color to the white matter of the brain, is a complex substance made up of a mixture of fats and proteins.) MLD is one of several lipid storage diseases, which result in the harmful buildup of lipids in brain cells and other cells and tissues in the body. People with lipid storage diseases either do not produce enough of one of the enzymes needed to break down (metabolize) lipids or they produce enzymes that do not work properly. MLD, which affects males and females, is cause by a deficiency of the enzyme *arylsulfatase A*.

MLD has three characteristic forms:

- *Late infantile* MLD typically begins between 12 and 20 months following birth. Infants appear normal at first but develop difficulty walking after the first year of life and eventually lose the ability to walk. Other symptoms include muscle wasting and weakness,developmental delays, progressive loss of vision leading to blindness, impaired swallowing, and dementia before age 2. Most children with this form of MLD die by age 5.
- *Juvenile formof* MLD (which begins between 3-10 years of age) includes impaired school performance, mental deterioration, an inability to control movements, seizures, and dementia. Symptoms continue to get worse, and death eventually occurs 10 to 20 years following disease onset.
- *Adult* MLD commonly begins after age 16, with symptoms that include psychiatric disturbances, seizures, tremor, impaired concentration, depression, and dementia. Death generally occurs within 6 to 14 years after onset of symptoms.