Metachromatic Leukodystrophy

Metachromatic leukodystrophy is a lysosomal storage disease caused by a defect in the enzyme arylsulfatase A. Deficiency of this enzyme can cause buildup of cerebroside sulfate in tissues and affect the growth and development of myelin. Due to demyelination, patients typically exhibit symptoms of ataxia and dementia.

PLAY PICMONIC

Pathophysiology

Autosomal Recessive

Recessive-chocolate

Metachromatic leukodystrophy is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be present in order for the disease to develop.

Arylsulfatase A Deficiency

Arrow-sulfur-match A Broken

This enzyme breaks down cerebroside sulfate into cerebroside and sulfate. This enzyme is deficient in metachromatic leukodystrophy.

Increased Cerebroside Sulfate

Up-arrow Cerebro-guy-on-side with Sulfur-matches

Deficiency of arylsulfatase A causes buildup of cerebroside sulfate in tissues.

Signs and Symptoms

Demyelination

Unraveling-myelin on neuron

Buildup of cerebroside sulfate affects the growth and development of myelin leading to demyelination.

Ataxia

A-taxi

Ataxia refers to lack of coordination of muscle movements caused by damage to the nervous system.

Dementia

Demented-D-man

Dementia is loss of cognitive ability due to damage to the CNS and is commonly seen in metachromatic leukodystrophy.