

What is MLD?

Metachromatic leukodystrophy

A rare,
life-threatening,
inherited
neurometabolic
disease

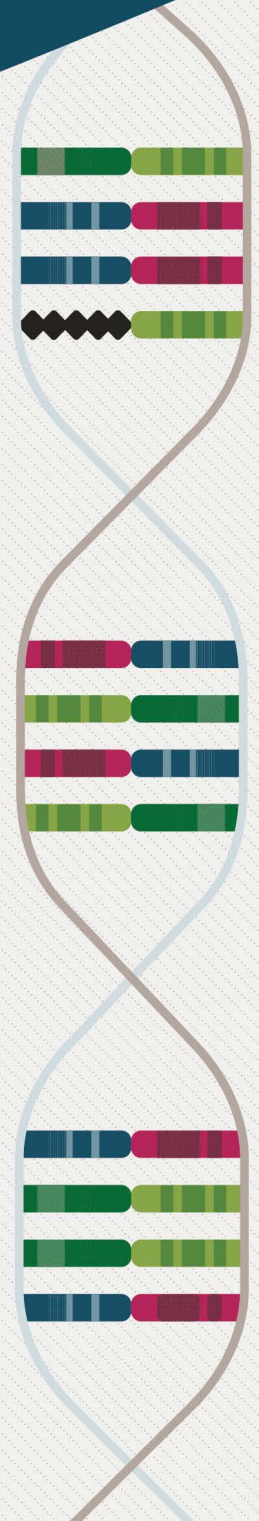
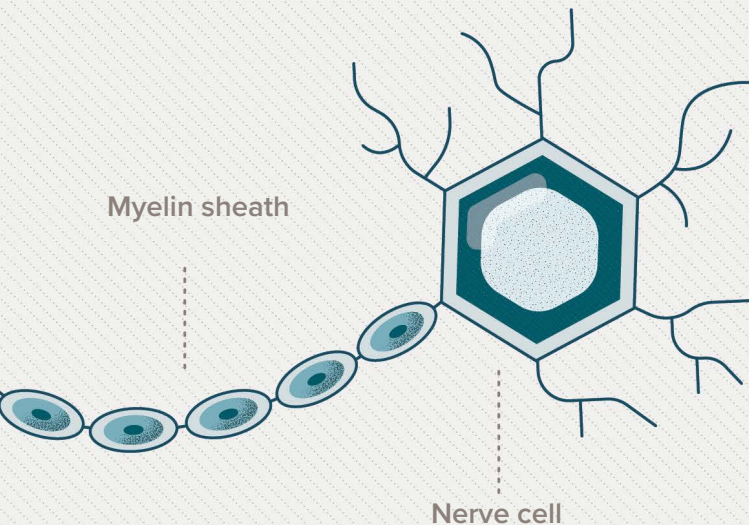


What are the causes of MLD?

Faulty gene
in the DNA

MLD is caused by a change, or mutation, in the arylsulfatase A (ARSA) gene that results in the accumulation of materials called sulfatides in the brain, nerves and other areas of the body, including the liver, gallbladder and kidneys.

This causes damage to the protective layer surrounding the nerves (myelin sheath), damaging the nervous system and leading to severe neurological problems.

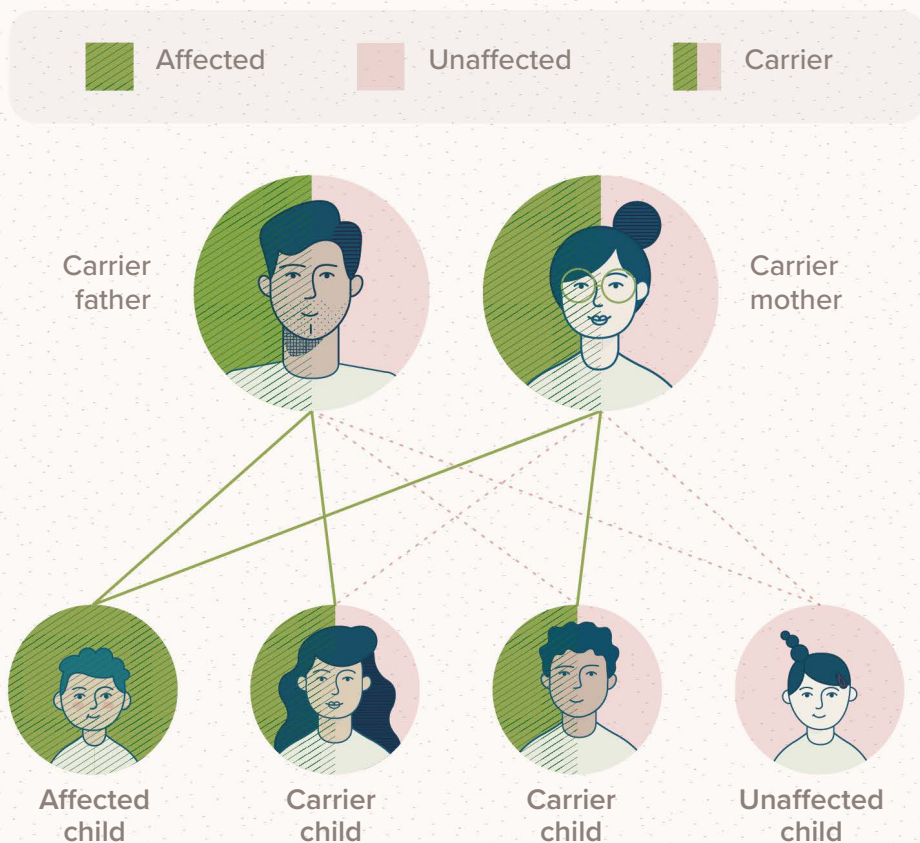


How is MLD inherited?

Approximately
**1 in every
100,000**
babies are born
with MLD

MLD is an autosomal recessive condition. This means that a child must inherit the faulty gene from both parents to have the condition.

MLD affects both boys and girls.



How does MLD affect the body?

**Symptoms
may include:**

People with MLD can experience mobility, behavioral and learning problems, and find it increasingly difficult to move, talk, swallow, eat and see.

Symptoms, age at symptom onset and disease progression, may vary depending on the form of MLD and the parts of the brain that are affected. Over half of people affected by MLD show symptoms before their 3rd birthday.

For more information about MLD and its management, please contact your healthcare provider.



Developmental
delays in children

Difficulty walking

Progressive
loss of skills

Behavior changes

Intellectual
disability

Muscle
spasticity

Pain

Seizures

Useful terms

ARSA gene

The gene responsible for making the enzyme arylsulfatase A (ARSA), that works to break down substances called sulfatides. If these aren't broken down, they can build up in the brain, nerves and other areas of the body causing problems.

Enzyme

A type of protein produced by the body's cells that increases the rate of chemical reactions, enabling the body to build up, or break down substances that are necessary for life and normal functioning.

Gene

Small sections of DNA that contain the instructions for individual characteristics, like eye and hair color, and how to make proteins, the functional building blocks of the cell. Proteins are responsible for making sure that the cells in the body function properly.

Mutation

A change in the structure of a gene or group of genes. Such changes can be passed on from parent to child. Many mutations cause no harm, but others can cause genetic disorders, such as MLD.

Myelin sheath

A protective fatty layer that surrounds the nerves in the brain (central nervous system) and in the nerves in your body (peripheral nervous system).

Neurometabolic diseases

A group of conditions causing problems with both metabolism (how cells chemically break down nutrients to get energy) and brain function.

Seizure

A sudden electrical disturbance in the brain causing physical and behavioral changes.

Muscle spasticity

A condition causing abnormal increase in muscle stiffness, which could interfere with movement.

References

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