

What is

# MLD?

Metachromatic leukodystrophy

A rare,  
life-threatening,  
inherited  
neurometabolic  
disease

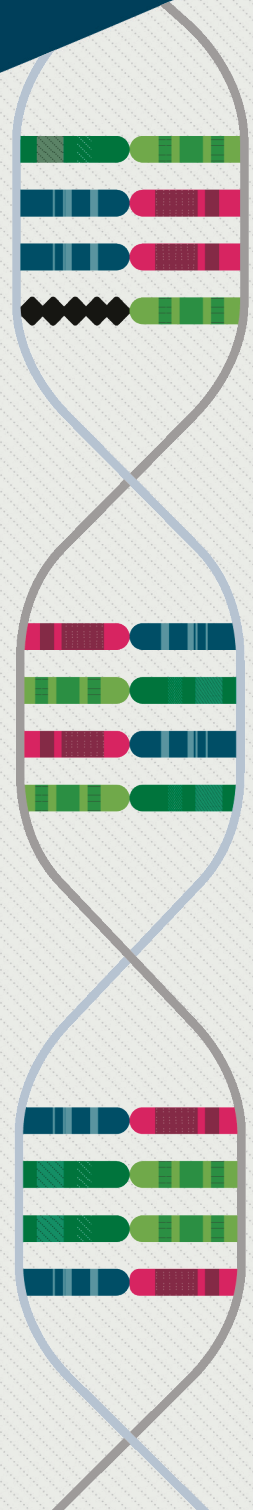
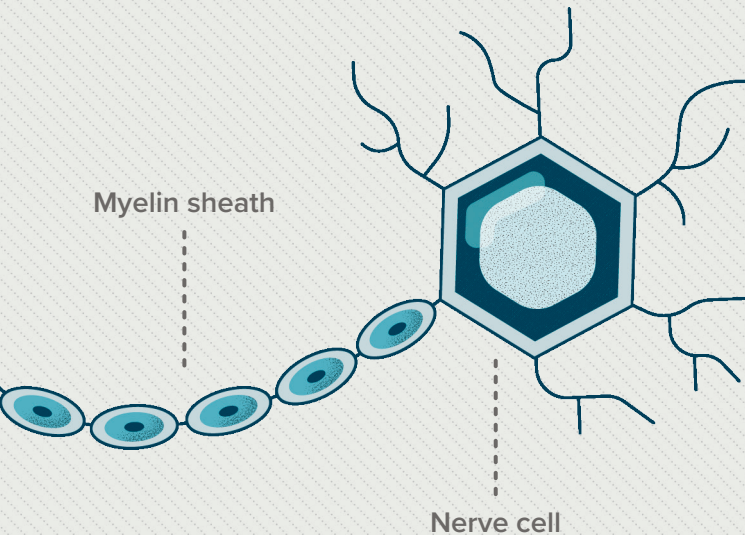


# What are the causes of MLD?

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.



Affected

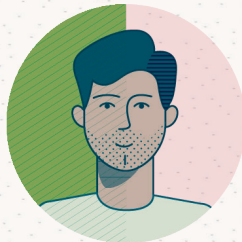


Unaffected



Carrier

Carrier father



Carrier mother



Affected child



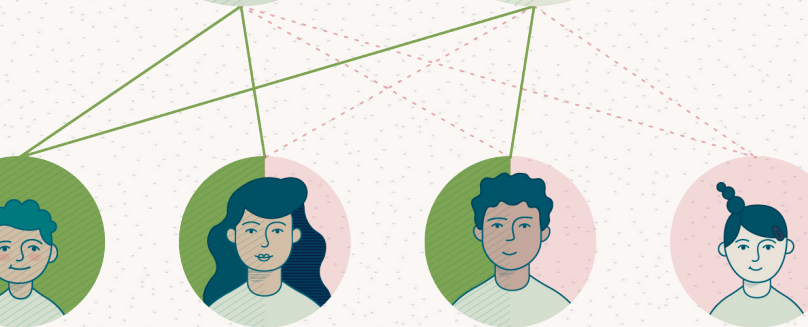
Carrier child



Carrier child



Unaffected child



# How does MLD affect the body?

Symptoms may include:



People with MLD can experience mobility, behavioural 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

Behaviour 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.

## Gene

Small sections of DNA that contain the instructions for individual characteristics, like eye and hair colour, 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.

## 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).

## Seizure

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

## 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.

## 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.

## Neurometabolic diseases

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

## Muscle spasticity

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

---

## References

- Kehrner C et al. Dev Med Child Neurol 2011; 53(9):850-855.
- Genetics Home Reference [Internet]. Bethesda (MD): The Library. Published: April 19, 2021. <https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns>
- National Organization for Rare Disorders [Internet]. "Metachromatic Leukodystrophy." Last modified: 2022. <https://rarediseases.org/rarediseases/metachromatic-leukodystrophy>

