

# Talk:Leukodystrophy

**Leukodystrophy** represents a group of genetically determined diseases that are manifested by morphological changes in the white matter of central nervous system. <sup>[1]</sup>

The cause of leukodystrophies turns out to be defects in the genes that code for various enzymes, the deficiency of which results in a metabolic disorder affecting myelin (e.g. impaired peroxisome function or a defect in the metabolism of sphingolipids). The basic characteristic of this group of diseases is myelin dysfunction. Myelin can be damaged either in the sense of **demyelination**, i.e. loss of normally formed myelin, or **dysmyelination**, i.e. loss of damaged myelin. Simple demyelination is typical of Sudanophilic leukodystrophies (X-ALD, Pelizaeus-Merzbacher disease). In dysmyelination, abnormal lipids are deposited in the myelin sheath, which is defective, and is manifested by metachromasia (metachromatic leukodystrophy) when stained).<sup>[1]</sup>

Leukodystrophies are the focus of several organizations, including the *United Leukodystrophy Foundation* (<https://ulf.org/>), *The Myelin Project* (<https://www.myelin.org/>) and last but not least, the *Stennis Foundation for Leukodystrophies* (<http://www.stennisfoundation.org/VjUXZ/ZgOcZ/OegPd/web/>).

## Overview of leukodystrophies

- **Pelizaeus-Merzbacher disease**
  - **Cockayne syndrome**
  - **Alexander's disease**
  - **Canavan disease**
  - **[[Krabbe's disease]**
  - **Metachromatic leukodystrophy**
  - **Peroxisomal leukodystrophy - - adrenoleukodystrophy** (ALD) – is manifested by progressive damage not only to brain, but also to the adrenal glands.
1. **X-linked ALD** (X-ALD)
  2. **Neonatal ALD** – (autosomal recessive)
  3. **Refsum's Disease**
  4. **Zellweger Syndrome**<sup>[1]</sup>

## Links

### Related Articles

- Peroxisomal Disease
- Peroxisomal Leukodystrophy
- X-linked adrenoleukodystrophy

### External Links

- <https://en.wikipedia.org/wiki/Leukodystrophies>
- <http://www.stennisfoundation.org/VjUXZ/ZgOcZ/OegPd/web/>
- <https://www.myelin.org/>
- <https://ulf.org/>

### Reference

- 1.

Kategorie:Patologie Kategorie:Neurologie Kategorie:Pediatrie Kategorie:Genetika