URL of this page: https://medlineplus.gov/leukodystrophies.html

Leukodystrophies

What are leukodystrophies?

Leukodystrophies are a group of rare genetic disorders [https://medlineplus.gov/geneticbraindisorders.html] that affect the central nervous system (CNS). The CNS is made up of your brain and spinal cord. Leukodystrophies damage the white matter of your CNS. The white matter includes:

- Nerve fibers, also called axons, which connect your nerve cells
- Myelin, a layer of proteins and fatty materials that covers and protects the nerve fibers. It also helps speed up signals between the nerve cells.

When the white matter is damaged, it can slow down or block the signals between nerve cells. This can cause many different symptoms, including trouble with movement [https://medlineplus.gov/movementdisorders.html] , vision [https://medlineplus.gov/visionimpairmentandblindness.html] , hearing [https://medlineplus.gov/hearingdisordersanddeafness.html] , and thinking.

There are over 50 types of leukodystrophies. Some types are present at birth, while others may not cause symptoms until a child becomes a toddler. A few types mainly affect adults. Most types get worse over time.

What causes leukodystrophies?

Leukodystrophies are caused by genetic changes. These changes are usually inherited, meaning that they are passed from parent to child.

What are the symptoms of leukodystrophies?

The symptoms of leukodystrophies depend on the type; they can include a gradual loss of:

- Muscle tone
- Balance [https://medlineplus.gov/balanceproblems.html] and mobility
- Walking
- Speech
- Ability to eat
- Vision
- Hearing
- Behavior

There can also be other symptoms, such as:

- Learning disabilities [https://medlineplus.gov/learningdisabilities.html]
- Bladder issues [https://medlineplus.gov/bladderdiseases.html]
- Breathing problems [https://medlineplus.gov/breathingproblems.html]
- Developmental disabilities [https://medlineplus.gov/developmentaldisabilities.html]
- Muscle control disorders

- Seizures [https://medlineplus.gov/seizures.html]

How are leukodystrophies diagnosed?

Leukodystrophies can be hard to diagnose because there are so many different types which can have different symptoms. Your health care provider may use many tools to make a diagnosis:

- Physical and neurological [https://medlineplus.gov/lab-tests/neurological-exam/] exams
- A medical history, including asking about family history
- Imaging tests [https://medlineplus.gov/diagnosticimaging.html] , such as an MRI [https://medlineplus.gov/mriscans.html] or CT scan [https://medlineplus.gov/ctscans.html]
- Genetic testing [https://medlineplus.gov/genetictesting.html] to look for genetic changes that could cause leukodystrophies
- Lab tests

What are the treatments for leukodystrophies?

There is no cure for leukodystrophies. Treatment focuses on relieving symptoms and providing support. It may include:

- Medicines to manage muscle tone, seizures, and spasticity (muscle stiffness)
- Physical, occupational, and speech therapies to improve mobility, function, and cognitive problems
- Nutritional therapy for eating and swallowing problems
- Educational and recreational programs

Stem cell [https://medlineplus.gov/stemcells.html] or bone marrow [https://medlineplus.gov/bonemarrowtransplantation.html] transplantation can be helpful for a few types of leukodystrophy.

One type of leukodystrophy, CTX, is treatable if it is diagnosed early. It is treated with chenodeoxycholic acid (CDCA) replacement therapy.

NIH: National Institute of Neurological Disorders and Stroke

Start Here

- Leukodystrophy [https://www.ninds.nih.gov/health-information/disorders/leukodystrophy]
  NIH (National Institute of Neurological Disorders and Stroke)
  Also in Spanish [https://www.ninds.nih.gov/es/health-information/disorders/leucodistrofia]
- What Is Leukodystrophy? [https://ulf.org/leukodystrophies/what-is-leukodystrophy/] (United Leukodystrophy Foundation)

Treatments and Therapies

- Krabbe Disease (GLD) [https://www.nmdp.org/patients/understanding-transplant/diseases-treated-by-transplant/krabbe-disease] (National Marrow Donor Program)

Specifics

- Aicardi-Goutieres Syndrome [https://www.ninds.nih.gov/health-information/disorders/aicardi-goutieres-syndrome]
  NIH (National Institute of Neurological Disorders and Stroke)
- CADASIL [https://www.ninds.nih.gov/health-information/disorders/cadasil]
  NIH (National Institute of Neurological Disorders and Stroke)
  Also in Spanish [https://www.ninds.nih.gov/es/health-information/disorders/cadasil]
- Leukodystrophy [https://www.ninds.nih.gov/health-information/disorders/leukodystrophy#refsum-disease-adult]
  NIH (National Institute of Neurological Disorders and Stroke)
- Lipid Storage Diseases [https://www.ninds.nih.gov/health-information/disorders/lipid-storage-diseases]
  NIH (National Institute of Neurological Disorders and Stroke)

Genetics

- Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/adult-onset-leukoencephalopathy-with-axonal-spheroids-and-pigmented-glia] (National Library of Medicine)
- Aicardi-Goutières syndrome: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/aicardi-goutieres-syndrome] (National Library of Medicine)
- Alexander disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/alexander-disease] (National Library of Medicine)
- Autosomal dominant leukodystrophy with autonomic disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/autosomal-dominant-leukodystrophy-with-autonomic-disease] (National Library of Medicine)
- Canavan disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/canavan-disease] (National Library of Medicine)
- D-bifunctional protein deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/d-bifunctional-protein-deficiency] (National Library of Medicine)
- Hypomyelination and congenital cataract: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hypomyelination-and-congenital-cataract] (National Library of Medicine)
- Hypomyelination with brainstem and spinal cord involvement and leg spasticity: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/hypomyelination-with-brainstem-and-spinal-cord-involvement-and-leg-spasticity] (National Library of Medicine)
- Krabbe disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/krabbe-disease] (National Library of Medicine)
- Leukoencephalopathy with thalamus and brainstem involvement and high lactate: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/leukoencephalopathy-with-thalamus-and-brainstem-involvement-and-high-lactate] (National Library of Medicine)
- Leukoencephalopathy with vanishing white matter: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/leukoencephalopathy-with-vanishing-white-matter] (National Library of Medicine)
- Megalencephalic leukoencephalopathy with subcortical cysts: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/megalencephalic-leukoencephalopathy-with-subcortical-cysts] (National Library of Medicine)
- Metachromatic leukodystrophy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/metachromatic-leukodystrophy] (National Library of Medicine)
- Pelizaeus-Merzbacher disease: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pelizaeus-merzbacher-disease] (National Library of Medicine)
- Pelizaeus-Merzbacher-like disease type 1: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pelizaeus-merzbacher-like-disease-type-1] (National Library of Medicine)
- Peroxisomal acyl-CoA oxidase deficiency: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/peroxisomal-acyl-coa-oxidase-deficiency] (National Library of Medicine)
- Pol III-related leukodystrophy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/pol-iii-related-leukodystrophy] (National Library of Medicine)
- RNAse T2-deficient leukoencephalopathy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/rnase-t2-deficient-leukoencephalopathy] (National Library of Medicine)
- TUBB4A-related leukodystrophy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/tubb4a-related-leukodystrophy] (National Library of Medicine)
- X-linked adrenoleukodystrophy: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/x-linked-adrenoleukodystrophy] (National Library of Medicine)

- Zellweger spectrum disorder: MedlinePlus Genetics [https://medlineplus.gov/genetics/condition/zellweger-spectrum-disorder] NIH (National Library of Medicine)

Clinical Trials

- ClinicalTrials.gov: Adrenoleukodystrophy [https://clinicaltrials.gov/search?cond=%22Adrenoleukodystrophy%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)
- ClinicalTrials.gov: Canavan Disease [https://clinicaltrials.gov/search?cond=%22Canavan+Disease%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)
- ClinicalTrials.gov: Leukodystrophy, Globoid Cell [https://clinicaltrials.gov/search?cond=%22Leukodystrophy,+Globoid+Cell%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)
- ClinicalTrials.gov: Leukodystrophy, Metachromatic [https://clinicaltrials.gov/search?cond=%22Leukodystrophy,+Metachromatic%22&aggFilters=status:not%20rec] NIH (National Institutes of Health)

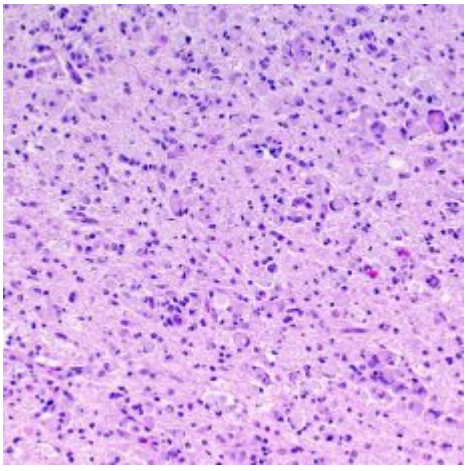
Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Amelioration of Inflammation and Metabolic Blockage in GALC Deficient Mice After... [https://www.ncbi.nlm.nih.gov/pubmed/40692539]
- Article: Self-Assembly of Accumulated Sphingolipids into Cytotoxic Fibrils in Globoid Cell Leukodystrophy... [https://www.ncbi.nlm.nih.gov/pubmed/40603002]
- Article: Metachromatic Leukodystrophy: New Therapy Advancements and Emerging Research Directions. [https://www.ncbi.nlm.nih.gov/pubmed/40577679]
- Leukodystrophies -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Leukodystrophy%2C+Globoid+Cell%22%5Bmajr%3Anoexp%5D+OR+%22Leukodystrophy%2C+Metachromatic%22%5Bmajr%3Anoexp%5D+OR+%22Canavan+Disease%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+]

Find an Expert

- Genetic and Rare Diseases Information Center [https://rarediseases.info.nih.gov/] NIH
- National Institute of Neurological Disorders and Stroke [https://www.ninds.nih.gov/] NIH



MEDICAL ENCYCLOPEDIA

Adrenoleukodystrophy [https://medlineplus.gov/ency/article/001182.htm]

Canavan disease [https://medlineplus.gov/ency/article/001586.htm]

Krabbe disease [https://medlineplus.gov/ency/article/001198.htm]

Metachromatic leukodystrophy [https://medlineplus.gov/ency/article/001205.htm]

Related Health Topics

Degenerative Nerve Diseases [https://medlineplus.gov/degenerativenervediseases.html]

National Institutes of Health

The primary NIH organization for research on *Leukodystrophies* is the National Institute of Neurological Disorders and Stroke [http://www.ninds.nih.gov/]