

Lipidoses and Glycogenosis

Overview

Lipidoses are genetic diseases caused by enzyme deficiencies, which result in the cellular accumulation of lipids.

Glycogenosis is a broad term for a group of genetic disorders that cause the abnormal use and storage of glycogen in the body's tissues.

Lipidoses

Lipids are fat-like substances (such as oils, fatty acids, waxes, steroids, cholesterol, and estrogen) that are important parts of the membranes found within and between cells and in the myelin sheath that coats and protects the nerves.

Gaucher Disease is caused by a deficiency of the enzyme glucocerebrosidase. Fatty material can collect in the brain, spleen, liver, kidneys, lungs, and bone marrow.

Niemann-Pick Disease is a group of autosomal recessive disorders caused by an accumulation of fat and cholesterol in cells of the liver, spleen, bone marrow, lungs, and, in some instances, brain.

- **Type A** – the most severe form. Infants appear normal at birth but develop profound brain damage by 6 months of age, an enlarged liver and spleen, swollen lymph nodes, and nodes under the skin (xanthomas).
- **Type B** – does not generally affect the brain but most children develop ataxia, damage to nerves exiting from the spinal cord (peripheral neuropathy), and pulmonary difficulties that progress with age.
- **Type C** – is not caused by a deficiency of sphingomyelinase but by a lack of the NPC1 or NPC2 proteins. As a result, various lipids and particularly cholesterol accumulate inside nerve cells and cause them to malfunction.
- **Type D** – a progressive neurodegenerative disorder characterized by the accumulation of tissue cholesterol and sphingomyelin.

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Fabry Disease causes a buildup of fatty material in the autonomic nervous system eyes, kidneys, and cardiovascular system. Fabry disease is the only X-linked lipid storage disease.

Sphingolipidoses are a class of lipid storage disorders or degenerative storage disorders caused by deficiency of an enzyme that is required for the catabolism of lipids that contain ceramide, also relating to sphingolipid metabolism.

Signs and symptoms may present as:

- Cherry-red halo around the center of the retina.
- Brain damage.
- Burning pain in the arms and legs.

Complications such as:

- Progressive neurological decline.
- Heart disease, renal failure, stroke.
- Elevated susceptibility to infection.

Treatment options of:

- Bone marrow transplant.
- Joint replacement surgery.
- Medication intervention.

Glycogenosis

Glycogen is a form of sugar (glucose) that is stored in the liver and muscles.

Von Gierke Disease is a condition in which the body cannot break down glycogen. This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

Pompe Disease results from the deficiency of an enzyme called acid alfa glucosidase (GAA), which breaks down complex sugars in the body.

Cori Disease is an inherited disorder caused by the buildup of a complex sugar. The accumulated glycogen is structurally abnormal and impairs the function of certain organs and tissues, especially the liver and muscles.

McArdle Disease results from changes (mutations) in the gene for the enzyme muscle phosphorylase.

Signs and symptoms may present as:

- Mild intellectual disability.
- Worsening muscle weakness.
- Muscle pain and cramping during exercise.

Complications such as:

- GI concerns such as IBS.
- Tumors in the liver.
- Cirrhosis of the liver.

Treatment options may consist of:

- Involve multiple specialists.
- PT, OT, or SLP.
- Diet management.

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Documentation Best Practices

Provider documentation should include elements such as:

- ✓ Detailed and thorough physical exam.
 - Organ enlargement that is palpable.
- ✓ Lab and imaging tests ordered along with their results.
- ✓ Diagnostic test results should be incorporated into the progress note during a face-to-face encounter.
- ✓ Detailed family history.

ICD-10-CM Information

E74 Other disorders of carbohydrate metabolism

E74.00	Glycogen storage disease, unspecified
E74.01	von Gierke disease
E74.02	Pompe disease
E74.03	Cori disease
E74.04	McArdle disease
E74.05	Lysosome-associated membrane protein 2 [LAMP2] deficiency
E74.09	Other glycogen storage disease

E75 Disorders of sphingolipid metabolism and other lipid storage disorders

E75.21	Fabry (-Anderson) disease
E75.22	Gaucher disease
E75.23	Krabbe disease
E75.240	Niemann-Pick disease type A
E75.241	Niemann-Pick disease type B
E75.242	Niemann-Pick disease type C
E75.243	Niemann-Pick disease type D
E75.244	Niemann-Pick disease type A/B
E75.248	Other Niemann-Pick disease
E75.249	Niemann-Pick disease, unspecified
E75.3	Sphingolipidosis, unspecified

References

“ICD-10”. Centers for Medicare & Medicaid Services, CMS.gov.
<http://www.cms.gov/medicare/coding/icd10>

“Pompe disease”. MedlinePlus, National Library of Medicine, 1 February 2016
<https://medlineplus.gov/genetics/condition/pompe-disease/>

“Glycogen Storage Disease”. Johns Hopkins Medicine, The Johns Hopkins University, Hospital, and Health System, 2022,
<https://www.hopkinsmedicine.org/health/conditions-and-diseases/glycogen-storage-disease>