Gaucher Disease

Last updated: 09/9/2024
Years published: 1984, 1985, 1986, 1987, 1988, 1989, 1990, 1992, 1993, 1994, 1995, 1996, 1997, 1998, 1999, 2001, 2002, 2003, 2004, 2007, 2008, 2011, 2014, 2018, 2024

Acknowledgment

NORD gratefully acknowledges Ozlem Goker-Alpan, MD, Lysosomal Research and Treatment Center (LDRTC), Fairfax, VA, and the Gaucher Community Alliance for assistance in the preparation of this report.

Disease Overview

Gaucher Disease (GD) is a lysosomal disorder characterized by an enlarged liver and/or spleen (hepatosplenomegaly), low levels of circulating red blood cells (anemia), low levels of platelets (thrombocytopenia) and skeletal abnormalities. In some forms of GD, the nervous system is primarily affected. The disease is caused by changes (variants) in the *GBA1* gene, leading to a deficiency of the enzyme glucocerebrosidase, resulting in the accumulation of glucosylceramide (Gb1) and its derivative glucosylsphingosine (Lyso-Gb1) in lysosomes, affecting various organs and systems.

GD is categorized into three types: non-neuronopathic (GD1), acute neuronopathic (GD2) and chronic neuronopathic (GD3). Symptoms vary depending on the type, with GD1 often presenting from early childhood to adulthood, GD2 manifesting before birth (prenatally) or within the first weeks of life and GD3 symptoms typically appearing during early childhood.

GD3 (chronic neuronopathic) typically manifests in early childhood, with neurological symptoms developing gradually, such as cognitive decline, lack of coordination (ataxia) and myoclonic seizures. The progression and cognitive involvement in GD3 can vary significantly, with some individuals presenting with eye movement abnormality as the only neurological abnormality.

Subtypes of GD3:

- GD3a is associated with myoclonic epilepsy.
- GD3b is characterized by severe visceral involvement, including enlarged liver and spleen (hepatosplenomegaly), growth delay and skeletal anomalies. Neurological issues in this subtype are usually milder and progress more slowly.
- GD3c, also known as the cardiac type, involves specific genetic variants that contribute to its unique clinical features, including heart valve abnormalities and other cardiovascular complications.

Next section >

Programs & Resources

RareCare® Assistance Programs

Gaucher's Disease Premium Copay Assistance

Closed

Phone: 203-296-3486 Email: gaucherdiseases@rarediseases.org Fax: 203-349-3189

Accepting Applications

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Additional Assistance Programs

Rare Disease Educational Support Program

Ensuring that patients and caregivers are armed with the tools they need to live their best lives while managing their rare condition is a vital part of NORD's mission.
https://rarediseases.org/patient-assistance-programs/rare-disease-educational-support/

Rare Caregiver Respite Program

This first-of-its-kind assistance program is designed for caregivers of a child or adult diagnosed with a rare disorder.

https://rarediseases.org/patient-assistance-programs/caregiver-respite/

Patient Organizations

Gaucher Community Alliance (GCA)

NORD Member

Email: info@gauchercommunity.org

https://rarediseases.org/organizations/gaucher-community-alliance-gca/

Vaincre Les Maladies Lysosomales

Email: accueil@vml-asso.org

https://rarediseases.org/organizations/vaincre-les-maladies-lysosomales/

National Gaucher Foundation

Email: ngf@gaucherdisease.org

https://rarediseases.org/organizations/national-gaucher-foundation/

NIH/National Institute of Neurological Disorders and Stroke

Phone: 301-496-5751 Fax: 301-402-2186

https://rarediseases.org/organizations/nih-national-institute-of-neurological-disorders-and-stroke/

Gauchers Association (UK)

Email: ga@gaucher.org.uk

https://rarediseases.org/organizations/gauchers-association-uk/

Proyecto Pide un Deseo MÃ©xico, i.a.p.

Email: prayecto.pdeundese.mexico@gmail.com

https://rarediseases.org/organizations/proyecto-pide-un-deseo-maxico-i-a-p/

More Information

The information provided on this page is for informational purposes only. The National Organization for Rare Disorders (NORD) does not endorse the information presented. The content has been gathered in partnership with the MONDO Disease Ontology. Please consult with a healthcare professional for medical advice and treatment.

GARD Disease Summary

The Genetic and Rare Diseases Information Center (GARD) has information and resources for patients, caregivers, and families that may be helpful before and after diagnosis of this condition. GARD is a program of the National Center for Advancing Translational Sciences

(NCATS), part of the National Institutes of Health (NIH).

View report

Orphanet

Orphanet has a summary about this condition that may include information on the diagnosis, care, and treatment as well as other resources. Some of the information and resources are available in languages other than English. The summary may include medical terms, so we encourage you to share and discuss this information with your doctor. Orphanet is the French National Institute for Health and Medical Research and the Health Programme of the European Union.

View report