What is Gaucher Disease?

Gaucher disease refers to a collection of rare inherited metabolic disorders characterised by the buildup of harmful levels of lipid in various body cells and tissues.[1] Gaucher disease results from mutations to the GBA gene.[2,3]

There are several types of the condition, which are classed according to their specific characteristics and severity:[1,2,3]

Gaucher Disease

Type 1	Type 2	Type 3	Type 3c	Perinatal lethal
Non-neuronopathic	Acute neuronopathic	Chronic neuronopathic	Causes hardening of the heart valves	Most severe form

Signs and Symptoms[4]

The symptoms typically vary between individuals.[4] However, the most common symptoms include:[4]

- Enlarged spleen and liver
- Anaemia
- Haemophilia
- Easy bruising
- Fatigue
- Bone pain
- Respiratory problems

Onset of Symptoms[1]

- **Prenatal** — Before birth
- **Newborn** — Birth–4 weeks
- **Infant** — 1–23 months
- **Child** — 2–11 years
- **Adolescent** — 12–18 years
- **Adult** — 19–65 years
- **Older adult** — 65+ years

The typical ages for symptoms of the disease to appear

Potential Complications[1]

- Ataxia
- Recurring bone fractures
- Developmental regression

Stay Aware of The Rare. Get Screened For Gaucher Disease Now!

References

1. Genetic and Rare Diseases Information Center. Gaucher disease. Available at https://rarediseases.info.nih.gov/diseases/8233/gaucher-disease. Accessed on 12 September 2022. **2.** National Library of Medicine. Gaucher disease. Available at https://medlineplus.gov/genetics/condition/gaucher-disease/. Accessed on 12 September 2022. **3.** National Organization for Rare Disorders. Gaucher disease. Available at https://rarediseases.org/rare-diseases/gaucher-disease/#causes. Accessed on 12 September 2022. **4.** National Gaucher Foundation. Gaucher Disease Symptoms. Available at https://www.gaucherdisease.org/about-gaucher-disease/symptoms/. Accessed on 12 September 2022.

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