

Gaucher Disease and Bones:

Everything You Need to Know



Gaucher is an autosomal recessive disease caused by a defect in the **GBA** gene.¹

Gaucher disease type 1 is one of the most common lysosomal disorders and affects approximately **one in 44,000** people worldwide.²

1.Futerman AH, Platt FM. The metabolism of glucocerebrosides - From 1965 to the present. Mol Genet Metab. 2017;120:22-26.
2.Pastores GM, Hughes, D. Gaucher Disease. GeneReviews (online). <https://www.ncbi.nlm.nih.gov/books/NBK1269/> . Accessed April 15, 2021.

Gaucher disease causes significant bone complications, including:

- **reduced bone density and strength (osteopenia/osteoporosis)**
- **fractures**
- **bone death (avascular necrosis)**
- **severe bone pain/bone crisis**³



3.Kaplan P, Andersson H, Kacena K, et al. The clinical and demographic characteristics of nonneuropathic Gaucher disease in 887 children at diagnosis. Arch Pediatr Adolesc Med. 2008;160:603-608.

90%

Of Gaucher patients are affected by bone disease.⁴

4.Marcucci, G., Zimran, A., Bembi, B. et al. Gaucher disease and bone manifestations. Calcif Tissue Int. 2014;95: 477.

81%

Of children show evidence of radiologic bone disease.⁵

5.Charrow, J., Dulisse, B., Grabowski, G., et al. The effect of enzyme replacement therapy on bone crisis and bone pain in patients with type 1 Gaucher disease. Clinical Genetics. 2007;71: 205-211.

25%

Of patients continue to have physical limitations due to bone disease after 2 years of ERT.⁶

6.Giraldo P, Solano V, Pérez-Calvo J, et al. Quality of life related to type 1 Gaucher disease: Spanish experience. Qual Life Res. 2005;4(2):453-62.

VS

Bone pain

Bone crisis

- Sharp, "take your breath away" pains
- Occurs with activity
- Goes away quickly
- "Dull and nagging"
- Happens daily
- "Spikes" of pain

- "Trapped in a closing vice"
- "Excruciating"
- "Unrelenting"
- "Paralyzing"
- "Shocking"
- "Hell"

Patients differentiated between chronic, daily bone pain and a bone crisis: although bone crises are not everyday occurrences, they are exceptionally debilitating and can take weeks to months to resolve.⁷

7.Copeland FD, Correlia IA, White ML. Uncovering the burden of Gaucher disease type 1: patient perspectives on unaddressed symptoms, impact of disease, and the future of treatment. Poster presented at : 17th Annual WORLDSymposium 2021; February 8, 2021.

Bone disease impairs quality of life and permanently alters health status due to chronic pain, disability, and potential need for surgical interventions.⁴

4.Marcucci, G., Zimran, A., Bembi, B. et al. Gaucher disease and bone manifestations. Calcif Tissue Int. 2014;95: 477.

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To learn more about Gaucher disease and a clinical trial evaluating an investigational gene therapy for Gaucher disease, visit:
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