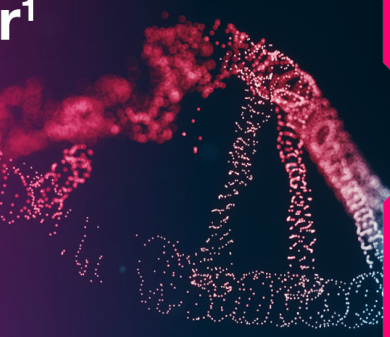
Bringing science to life

Spotlight on... Gaucher disease (GD)

GD is a rare genetic disorder[1]

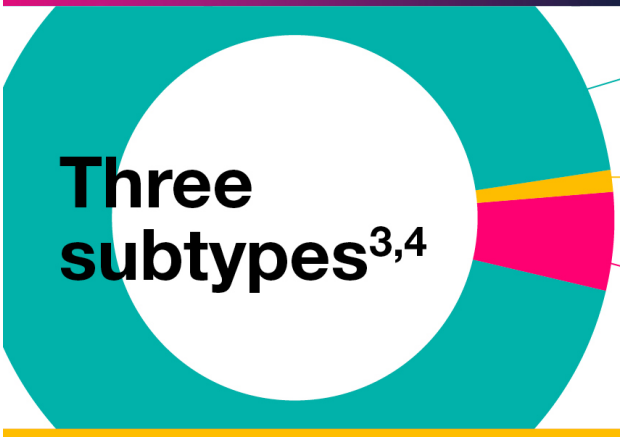
Affects **0.70–1.75** in **100,000** globally, but **1 in ~850** among people of **Ashkenazi Jewish** ethnicity[1]



GD is caused by a deficiency of an enzyme called **glucocerebrosidase**[2]

It leads to a build up of a **fatty substance called glucosylceramide** in cells, which damages tissues and organs[2]

Three subtypes[3,4]



Type 1: 94% of cases
Doesn't affect central nervous system (CNS)

Type 2: 1% of cases
Affects CNS; typically fatal by age 2–3 years

Type 3: 5% of cases
Affects CNS and progresses slowly

Prevalence data from Gaucher Registry[5]

Symptoms of GD worsen over time[1]

Anaemia & thrombocytopenia	Types 1–3[6]
Lung disease	
Increased bleeding & bruising	
Enlarged liver & spleen	
Bone pain & fractures	
Neurological impairment	Types 2&3 only[3,6]

Diagnosis

Blood test for active glucocerebrosidase[5]

Genetic testing to identify carriers[5]

Treatment

GD can't be cured yet, but **symptoms can be treated** with enzyme replacement therapy[1,3]

Early diagnosis and initiation of treatment may improve outcomes for patients with GD[7]

1. Nalysnyk L, et al. *Hematology.* 2017;22(2):65–73. **2.** Rosenbloom BE, Weinreb NJ. *Crit Rev Oncog.* 2013;18(3):163–175. **3.** Linari S, Castaman G. *Clin Cases Miner Bone Metab.* 2015;12(2):157–164. **4.** National Gaucher Foundation. https://www.gaucherdisease.org/about-gaucher-disease/what-is/type-2-3/. Accessed 01 Oct 2021. **5.** GeneReviews®. https://www.ncbi.nlm.nih.gov/books/NBK1269/. Accessed 30 Sep 2021. **6.** Nagral A. *J Clin Exp Hepatol.* 2014;4(1):37–50. **7.** Peters H, et al. *Int Med J.* 2020;50(S4):5–27.

cognite
Science brought to life