Gaucher Disease (GBA)

Gaucher Disease is caused by mutations in the GBA gene. This gene encodes for the enzyme beta-glucocerebrosidase, which breaks down the substance glucocerebroside. The buildup of glucocerebroside causes damage to various organs. There are various types of Gaucher disease. Type 1 is the most common, and involves anemia, lung disease, enlargement of the spleen and liver, easy bruising of the skin, and skeletal disorders such as arthritis and high risk of fractures. The nervous system is not affected in type 1 Gaucher disease. Types 2 and 3 involve serious damage to the nervous system, with type 2 being the more aggressive, leading normally to early mortality. A perinatal form of the disease is also known, leading to prompt death after birth. Finally, a cardiovascular form of the disease mainly involves damage to the heart valves.

In the general population, Gaucher disease is found in 1 in 60,000 to 1 in 80,000 new births. It is much more prevalent in various ethnic groups. Among those of Ashkenazi Jewish descent, the disease is found in 1 in 855 people (nearly all Type 1), with around 1 in 18 people being carriers. The disease is autosomal recessive, typically requiring both parents to be asymptomatic carriers of the faulty gene copy.

Sources

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