

WHO IS AFFECTED BY GAUCHER DISEASE?

While there are three types of Gaucher disease, type 1 Gaucher is the most common form of Gaucher disease, representing about 95% of all cases.

~1-9 | N 100,000 in the overall population has type 1 Gaucher disease

~1 | N 600 within the Ashkenazi Jewish population has type 1 Gaucher disease ~1 IN 17 within the Ashkenazi Jewish population is a Carrier of a Gaucher cell mutation

Because Gaucher (-shay) disease is rare, the average person has probably never heard of it. At Takeda, we're working to change that by arming people with the information they need to better understand this lifelong condition, to find out if they (and their families) may be at risk, and to learn what to do if they have concerns.

THE THREE SIMPLE STEPS IN THIS BROCHURE WILL HELP YOU GET STARTED.







WHAT IS GAUCHER DISEASE?

Gaucher (foll-shay) disease is a rare disease caused by the deficiency of an enzyme your body needs. Over time, this can result in the build-up of a fatty substance in cells, causing damage to tissues and organs.

Gaucher disease can impact people of all ages and genders. Additionally, many symptoms can overlap with other disorders making Gaucher disease difficult to diagnose. It may take years to arrive at a conclusive diagnosis.

Gaucher disease is hereditary, which means that people can be Carriers of the Gaucher cell mutation without having the condition themselves. It also means that if two Carriers have children together, they can pass the Gaucher cell mutation on to their children and there would be a chance their children could develop Gaucher disease.

If both parents are Carriers







Then there is a 50% chance of the child also being a Carrier

A 25% chance that the child will have Gaucher disease

And a 25% chance the child will be unaffected

GO! EVALUATE

WHAT SYMPTOMS SHOULD I BE LOOKING OUT FOR?

One of the reasons Gaucher disease can be so difficult to diagnose is that it often resembles other conditions.





HOW CAN I FIND OUT IF I HAVE GAUCHER DISEASE?

If you're concerned about Gaucher disease, talk to your doctor. There are tests that can be performed to learn more.



TO LEARN IE YOU HAVE GAUCHER DISEASE:

A simple test, called the beta-glucosidase leukocyte (BGL) test, measures the levels of the enzyme glucocerebrosidase. Low levels of this enzyme will confirm a diagnosis of Gaucher disease.



ADDITIONAL TESTING:

Genetic testing can identify the specific genetic mutations that result in Gaucher disease. Genetic testing may also be used to identify Carriers. While Carriers do not experience symptoms of Gaucher disease, it is important to identify Carriers because it is a hereditary condition.

It you have additional questions about the tests, condition, treatment options, or anything else dealing with Gaucher disease