## Increased Awareness Of Rare Diseases Helps Patients With Gaucher Disease Map The Right Blueprint

(NAPSA)—In the United States there are nearly 7,000 rare diseases. Because of the small patient populations and a variety of symptoms that often mimic other disorders, many physicians have difficulty diagnosing rare disorders such as Gaucher disease.

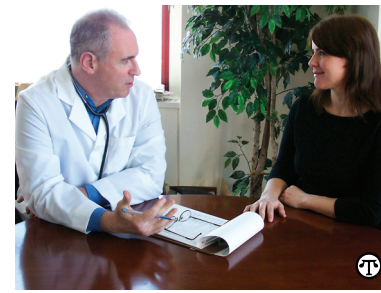
Patients can feel isolated or scared because they do not have clarity around a diagnosis or treatment path. Those who do have a proper diagnosis have often suffered through years of misdiagnosis and exposure to unnecessary treatments that may not address the underlying disease or its symptoms.

Gaucher disease (pronounced: go-shay) is a chronic and progressive disorder with a varying age of onset and with many different symptoms that can range from patients having no symptoms to severe disease. Gaucher disease is an inherited genetic disease that often strikes individuals within the same extended family and affects many of the body's organs and tissues including the liver, spleen and bones.

It is estimated that between 1 in 50,000 to 1 in 100,000 people have Gaucher disease. However, it is found most frequently in the Eastern European (Ashkenazi) Jewish population, where it is estimated that 1 in 15 people are carriers of the defective gene.

Many patients do not experience symptoms of Gaucher disease until the second decade of life, but Michelle Neumann was one of a few patients to be diagnosed with Gaucher disease at birth.

"At the time I was diagnosed, it was tabula rasa—no one knew how to treat, you just treated the symptoms for Gaucher disease. Having been diagnosed at birth, I didn't understand the gravity of my disease until recently, when I was out with friends from summer camp and I discovered that they didn't expect me to live past college. That was an eye-opener," said Michelle.



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Advances in early screening and treatment for rare conditions such as Gaucher disease have enabled many patients to confirm their diagnosis earlier and begin appropriate treatment sooner. Today, Gaucher disease can be diagnosed from a single blood test.

Early and accurate diagnosis also allows patients and their families to seek out support networks that can help them cope with the disease and share with others treatment strategies that have worked for them. This was the case for patient Wayne Rosenfield.

"I can remember going to the library after receiving my diagnosis [Gaucher disease] and scouring the research material with my father. Everything we read was ominous and offered no hope," said Wayne. "Then a few years later, I saw an article about Gaucher disease in the newspaper. It was a feeling I'll never forget; I was no longer alone. I still have that newspaper."

Today, patients can benefit from available therapy options. Patients have a lot to be optimistic about, in fact, doctors believe we are entering a new era of treatment for rare diseases. Thanks to ongoing efforts, future generations of patients may have the proper blueprint to manage their Gaucher disease.

To find out more information about Gaucher disease, please visit www.gaucherdisease.org.