Stanford HEALTH CARE

Ashkenazi Jewish Genetic Screening

What is an Ashkenazi Jewish Disease?
Ashkenazi is the term used to describe Jewish individuals who have ancestors from Eastern Europe. Roughly 90% of the six million Jewish individuals in the United States are of Ashkenazi descent. Similar to most ethnic populations, the Ashkenazi Jewish population has a higher prevalence of certain genetic disorders. Individuals of Jewish descent should be screened for Tay-Sachs disease, Canavan disease and Gaucher's disease.

What Is Tay-Sachs Disease?
Tay-Sachs disease is a fatal genetic disorder that occurs more frequently in the Ashkenazi (Eastern European) Jewish population. Approximately 1 in 27 Ashkenazi Jewish individuals are carriers of this disease. A baby with Tay-Sachs disease appears normal at birth, but after six months of age, the child progressively develops mental retardation followed by paralysis, blindness, and seizures. Death usually occurs by the age of five. Tay-Sachs disease is caused by a deficiency of an enzyme called Hex-A. As a result of this deficiency, there is an accumulation of certain substances, which damage the nervous system.

What is Canavan Disease?
Canavan disease is a progressive disorder in which the brain and nervous system degenerate. Symptoms of Canavan disease include brain damage, mental retardation, feeding difficulties, blindness, and a large head. There is no treatment, and death usually occurs in the first decade of life.

What is Gaucher's Disease?
Gaucher's Disease is an inborn error of metabolism that results from a specific malfunction in on of the body's individual chemical processes. Although there are at least 34 mutations known to cause Gaucher Disease, there are 4 genetic mutations which account for 95% of the Gaucher's Disease in the Ashkenazi Jewish population. The carrier rate is 1 in 14 Jewish people of Eastern European ancestry and 1 in 100 of the general population.

How Are These Diseases Inherited?
All three diseases are inherited in an autosomal recessive pattern. For an individual to be affected, he/she must inherit one copy of the abnormal (mutated) gene from each parent. Individuals having one copy of the particular disease-causing gene and one copy of the normal gene are known as carriers. Carriers usually do not have any symptoms of the disorder. If both parents carry the same mutated gene, their child has a 25% chance of having the disease. If only one parent carries the disease gene, their child is not at risk for having that disease but has a 50% chance of being a carrier. If both parents are carriers, the couple should undergo prenatal genetic counseling.