



## **TAY-SACHS DISEASE**

### **WHAT IS TAY-SACHS DISEASE?**

Tay-Sachs Disease is an inherited, neurodegenerative disorder found in eastern European Jewish families and some French-Canadian families. It is characterized by progressive mental and physical retardation and early death.

### **WHAT ARE THE SYMPTOMS OF TAY-SACHS DISEASE?**

Symptoms first appear around 6 months of age. At that time the infant learns no new skills and gradually loses skills acquired to that point. After 1 year of age, convulsions and seizures begin, as well as atrophy of the optic nerve. Blindness, spasticity, dementia, and paralysis follow. A specific characteristic of Tay Sachs disease is the appearance of a cherry-red spot on the retina. Death occurs between 2 and 4 years of age.

### **WHAT CAUSES TAY-SACHS DISEASE?**

Ashkenazi Jews sometimes carry an autosomal recessive trait that is transmitted to their offspring. The trait causes a deficiency in hexosaminidase A, an enzyme that is important in the metabolism of fatty acids. When the fatty acids don't metabolize properly, they accumulate in the brain and cause neurological deterioration.

### **HOW IS TAY-SACHS DIAGNOSED?**

Tay Sachs disease can be identified by chorionic villus sampling or amniocentesis. Genetic screening of parents can be done before conception. Tay-Sachs occurs in 1 of 3600 infants with Ashkenazi Jewish or French-Canadian parents.

### **HOW IS TAY-SACHS DISEASE TREATED?**

The only treatment is to attempt to relieve the symptoms associated with the disease.

**THOMAS G. STOVALL, M.D.**

Dr. Stovall is a Clinical Professor of Obstetrics and Gynecology at the University of Tennessee Health Science Center in Memphis, Tennessee and Partner of Women's Health Specialists, Inc.

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**Health Information Provided by Women's Health Specialists**

7800 Wolf Trail Cove, Germantown, TN 38138, (901) 682-9222, [www.whsobgyn.com](http://www.whsobgyn.com)

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