Newsbites

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The Life Saving Cord Blood Bank

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TAY-SACHS DISEASE- AN AUTOSOMAL RECESSIVE GENETIC DISORDER

Tay-Sachs Disease (TSD) is an inherited disorder that progressively destroys nerve cells (neurons) in the brain and spinal cord.

Symptoms:

There are different forms of Tay-Sachs Disease and the form is determined by the age of the individual when symptoms first appear.

1. Classic Infantile Tay-Sachs: Symptoms appear around 6 months of age. Parents may notice a reduction in vision and tracking and the baby does not outgrow normal startle response.

2. Juvenile Tay-Sachs: Symptoms typically appear between ages 2 and 5, but can occur anytime during childhood. Early symptoms of Juvenile Tay-Sachs include lack of coordination or clumsiness and muscle weakness such as struggling with stairs. A child may also exhibit slurred speech, swallowing difficulties and muscle cramps.

3. Late Onset Tay-Sachs: Symptoms typically appear in adolescence or early adulthood, but can appear later. This includes clumsiness and muscle weakness in the legs. Once diagnosed, adults often reflect back to their childhood and may notice experiencing symptoms much earlier such as not being athletic and/or speech difficulties or a stutter as a child or teenager.

Incidence:

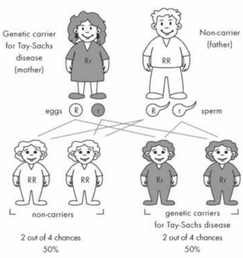
Only one form of Tay-Sachs occurs in a family. If a child has Infantile, older siblings are not at risk to develop Juvenile or Late Onset Tay-Sachs later in life.

·High prevalence has been reported in children with neurological disorders from the southern region of India, consanguinity is more common [1].

·Higher incidence of Tay-Sachs Disease was observed in the SC community of Gujarat [2].

Causes:

Mutations in the Hex-A gene cause Tay-Sachs Disease. The Hex-A gene provides instructions for making part of an enzyme called beta-hexosaminidase A, which plays a critical role in the brain and spinal cord. This enzyme is located in lysosomes, which are structures in cells that break down toxic substances and act as recycling centers. Within lysosomes, beta-hexosaminidase A helps break down a fatty substance called GM2 ganglioside.



Mutations in the Hex-A gene disrupt the activity of beta-hexosaminidase A, which prevents the enzyme from breaking down GM2 ganglioside. As a result, this substance accumulates to toxic levels, particularly in neurons in the brain and spinal cord.

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Treatment:

There is no effective treatment for the condition as of now. All the treatments available focuses on management of the symptoms.

Stem cell therapy:

Usage of umbilical cord stem cells are under research. Martin, Carter, Kernan, Sahdev, Wall, Pietryga et al. (2006) investigated the use of stem cells in cord blood to treat lysosomal and peroxisomal storage diseases (LSDs) in 69 children (mean age = 1.8 years), 3 of whom had Tay-Sachs Disease. Survival improved with higher matches of antigens of the cord blood cells to the patient. Results suggest that stem cell transplantation is a viable method of prolonging life in Lysosomal storage diseases patients[3].

Treatments	Reasons for Treatments
Medication	To manage pain or seizures in TSD patients
Respiratory Care	To solve the difficulties with breathing. e.g.: Chest physiotherapy is used to reduce mucus.
Feeding Tubes	To solve the swallowing problem. e.g.: Nasogastric (NG) tube helps to deliver nutrients through the nose and esophagus into the stomach. Percutaneous esphago-gastromy(PEG) tubes inserted surgically.
Physical Therapy	To prevent deterioration of neurological and motor functions in TSD patients.

Sources:

1. Nalini A, Christopher R. Cerebral glycolipidoses: clinical characteristics of 41 pediatric patients. J Child Neurol. 2004;19(6):447–452.
2. Mistri M, Tamhankar P, Sheth F, et al. Identification of novel mutations in HEXA gene in children affected with Tay Sachs disease from India. PLoS One. 2012;7(6):e39122. doi: 10.1371/journal.pone.0039122.
3. Martin, L.P., Carter, S.L., Kernan, N.A., Sahdev, I., Wall, D., Pietryga, D., et al. (2006). Results of the Cord Blood Transplantation Study (COBLT): Outcomes of Unrelated Donor Umbilical Cord Blood Transplantation in Pediatric Patients with Lysosomal and Peroxisomal Storage. Diseases. Biology of Blood and Marrow Transplantation 12, 184- 194.

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