Case study

Health & Medicine, Nursing

Tay - Sachs disease affiliation Tay - Sachs disease-it is a rare autosomal-recessive, continuous and finally calamitous neurodegenerative disorder. The symptoms surface before the baby attains six months of age. Symptoms include; loss of motor skills, startle reaction, muscle paralysis, seizures, slow growth, decreased eye contact and delayed mental and social skills (Ohno, Saito, Sugawara, & Sakuraba, 2008).

2. The inheritance pattern; Tay-Sachs is a genetic disease that is only transferred to a child if both parents carry a Tay-Sachs gene. A child who inherits two defective genes from both parents has nonfunctional Hex-A enzyme which causes Tay-Sachs disease.

3. Testing for carrier or affected individuals with Tay-Sachs disease; blood screening for hexosaminidase-A (Hex-A) mutation in suspected individual that either have family history or are of Ashkenazi Jewish descent is used to identify carriers. Blood test is performed antenatal using chronic villus sampling and amniocentesis or after birth.

4. Organelle dysfunction underlying this disease; lysosomes are the main organelles associated with Tay-Sachs disease

5. Function of lysosomes; they produce enzyme Hexosaminidase-A (Hex-A) that prevents buildup of fatty materials called GM2 ganglioside in the cells brain and nerves. This further averts malfunctioning of nerves and brain cells (Americo, Filho, & Shapiro, 2010).

6. Role of Hexosaminidase-A enzyme; this enzyme is responsible for abasement of GM2 ganglioside and many other biological molecules that have terminal N-acetyl hexosamins in both the brain cells and nervous cells. Deficiency of enzyme hexosaminidase A causes Tay-Sachs disease due to

increased buildup of toxic GM2 ganglioside a fatty substance that destroys brain cells and nerves. This results in motor difficulties and other signs and symptoms.

7. Diagnosis of Tay-Sachs disease; blood test is analyzed for enzyme assay or biochemical examination that reveals the levels of hexosaminidase- A in an individual. Affected individuals have less -hexosaminidase A blood and other cells than non-carriers.

8. Management; Tay-Sachs disease has no cure; only management is to relieve symptoms. Prevent airway and lung problems; relieve dysphagia and medication for fits, muscle stiffness and eye problems (Chamoles, Blanco, Gaggioli, & Casentini, 2002).

Reference

Americo, J., Filho, F., & Shapiro, B. E. (2010). Tay-Sachs disease. Arch Neurol, 61(11), 1466–1468.

Chamoles, N. A., Blanco, M., Gaggioli, D., & Casentini, C. (2002). Tay-Sachs and Sandhoff diseases: Enzymatic diagnosis in dried blood spots on filter paper: Retrospective diagnoses in newborn-screening cards. Clinica Chimica Acta, 318(1-2), 133–137. doi: 10. 1016/S0009-8981(02)00002-5

Ohno, K., Saito, S., Sugawara, K., & Sakuraba, H. (2008). Structural consequences of amino acid substitutions causing Tay-Sachs disease. Molecular Genetics and Metabolism, 94(4), 462–468. doi: 10. 1016/j. ymgme. 2008. 04. 006

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