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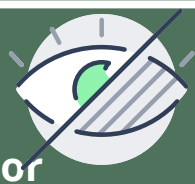
STURGE WEBER DISEASE

Sturge-Weber syndrome is a condition that affects the development of certain blood vessels, causing abnormalities in the brain, skin, and eyes from birth.

Characteristics



The illness has three main characteristics which include red or pink birthmark, leptomeningeal angioma, and glaucoma. Not all the individuals with the disease have all the three features. It can vary from patient to patient. The red marks are caused by the dilation of the blood vessels around the skin. The color of the mark can be from pink to purple and the mark is flat on the skin. The leptomeningeal angioma can cause less blood flow to the brain, which could lead to atrophy and calcification inside the brain tissue. Some people have learning disability issues like ADHD as part of the disease. Vision impairment is common among individuals with glaucoma. The glaucoma can start in infancy or in the early 20s time period. Furthermore, in babies with severe sturge weber disease, the eyeballs could become large because of immense pressure build up.

Cause

Sturge-Weber syndrome is caused by a mutation in the GNAQ gene. This gene provides instructions for making a protein called guanine nucleotide-binding protein G(q) subunit alpha (Gαq). The Gαq protein is part of a group of proteins (complex) that regulates signaling pathways to help control the development and function of blood vessels.

The GNAQ gene mutation that causes Sturge-Weber syndrome results in the production of a protein with impaired function. As a result, the altered Gαq protein cannot play its part in regulating signaling pathways, resulting in abnormally increased signaling. The enhanced signaling likely disrupts the regulation of blood vessel development, causing abnormal and excessive formation of vessels before birth in people with Sturge-Weber syndrome.

Treatment

The disease is not curable. However, there are treatments to reduce the symptoms of the disease. Laser treatment can be used to remove the birthmark. Also, anticonvulsant medications can be used to reduce seizures. People who cannot be treated with anticonvulsants can go through surgical removal of the damaged epileptic brain parts. Also, they can use eye drops or go through surgery to control the eye problems.

Reference

https://medlineplus.gov/genetics/condition/sturge-weber-syndrome/ https://www.ninds.nih.gov/health-information/disorders/sturge-weber-syndrome