Sturge-Weber Syndrome

WHAT IS STURGE-WEBER SYNDROME?

Sturge-Weber syndrome is characterized by a reddish discoloration of the skin on one side of the face (port-wine birthmark) as well as malformations of the blood vessels in the brain [See figure 1].

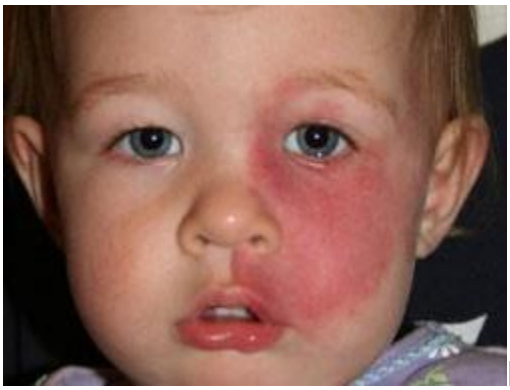

Fig. 1: Sturge-Weber Syndrome is characterized by a reddish discoloration of the skin on one side of the face.

DOES EVERY CHILD WITH A PORT-WINE BIRTHMARK OF THE SKIN HAVE STURGE-WEBER SYNDROME?

No. The reddish discoloration of the face may be the only finding. Sturge-Weber syndrome is diagnosed when the brain is involved as well.

IN WHAT WAY IS THE BRAIN INVOLVED?

Abnormal blood vessels in the brain may cause a variety of symptoms (leptomeningeal malformations). Seizures, neurological deficits, developmental delay, and weakness are some of the more common findings. Many children, however, have normal intelligence and development.

WHAT TESTS ARE DONE TO LOOK FOR BRAIN INVOLVEMENT?

Neuro-imaging studies (MRI / CT) may be utilized to evaluate for abnormal blood vessels of the brain.

WHAT CAUSES STURGE-WEBER SYNDROME?

Sturge-Weber syndrome is not a genetically inherited disease. The abnormalities associated with Sturge-Weber syndrome develop very early in pregnancy - probably between the second and third month of pregnancy.

WHEN IS STURGE-WEBER DIAGNOSED?

The port-wine birthmark is usually noted shortly after birth. Further testing may reveal involvement of the brain (Sturge-Weber syndrome).

WHY IS THE SKIN OF THE FACE SO RED?

Redness (port-wine birthmark) of the skin is caused by numerous enlarged small blood vessels. The forehead and upper eyelid are often involved while the lower eyelid and cheek are occasionally involved.

IS THERE ANY TREATMENT FOR THE SKIN?

The skin appearance can be improved in some situations by laser treatment.

WHAT EYE PROBLEMS CAN OCCUR WITH STURGE-WEBER SYNDROME?

Any child with a port-wine birthmark should be evaluated by an ophthalmologist. Children diagnosed with Sturge-Weber Syndrome should be examined by an ophthalmologist promptly and should be monitored regularly by an ophthalmologist for eye problems.

Some children with Sturge-Weber have eyes that are different colors. The surface of the eye occasionally has enlarged blood vessels which cause a pink or red eye appearance. These blood vessel abnormalities can occur inside the eye as well, leading to an elevated pressure within the eye. If this elevated pressure begins to cause damage to the optic nerve, this condition is referred to as “glaucoma.”

Glaucoma is the most serious eye problem associated with Sturge-Weber syndrome. About 50% of children with Sturge-Weber develop glaucoma between infancy and late childhood. Signs of glaucoma may include an enlarged eye, a cloudy appearing eye, light sensitivity, or excessive tearing.

If the optic nerve or visual areas of the brain are involved, a decrease in peripheral vision may result.

HOW IS GLAUCOMA TREATED?

Eye drops may be used to reduce the pressure inside of the eye. Sometimes, eye surgery may be needed.

WHERE CAN I LEARN MORE ABOUT STURGE-WEBER SYNDROME?

- Sturge-Weber Foundation

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