Sturge-Weber syndrome

Sturge-Weber syndrome is a condition that affects the development of certain blood vessels, causing abnormalities in the brain, skin, and eyes from birth. Sturge-Weber syndrome has three major features: a red or pink birthmark called a port-wine birthmark, a brain abnormality called a leptomeningeal angioma, and increased pressure in the eye (glaucoma). These features can vary in severity and not all individuals with Sturge-Weber syndrome have all three features.

Most people with Sturge-Weber syndrome are born with a port-wine birthmark. This type of birthmark is caused by enlargement (dilatation) of small blood vessels (capillaries) near the surface of the skin. Port-wine birthmarks are typically initially flat and can vary in color from pale pink to deep purple. In people with Sturge-Weber syndrome, the port-wine birthmark is most often on the face, typically on the forehead, temple, or eyelid. The port-wine birthmark is usually only on one side of the face but can be on both sides. Over time, the skin within the port-wine birthmark can darken and thicken.

In Sturge-Weber syndrome, there is usually abnormal formation and growth of blood vessels within the two thin layers of tissue that cover the brain and spinal cord. This abnormality, which is called leptomeningeal angioma, can affect one or both sides of the brain and impair blood flow in the brain and lead to loss of brain tissue (atrophy) and deposits of calcium (calcification) in the brain below the angioma. The decrease in blood flow caused by leptomeningeal angiomas can cause stroke-like episodes in people with Sturge-Weber syndrome. These episodes often involve temporary muscle weakness on one side of the body (hemiparesis), vision abnormalities, seizures, and migraine headaches. In affected individuals, these episodes usually begin by age 2. The seizures usually involve only one side of the brain (focal seizures), during which the port-wine birthmark may darken and individuals may lose consciousness. People with Sturge-Weber syndrome have varying levels of cognitive function, from normal intelligence to intellectual disability. Some individuals have learning disabilities with problems focusing similar to attention-deficit/hyperactivity disorder (ADHD).

In individuals with Sturge-Weber syndrome, glaucoma typically develops either in infancy or early adulthood and can cause vision impairment. In some affected infants, the pressure can become so great that the eyeballs appear enlarged and bulging (buphthalmos). Individuals with Sturge-Weber syndrome can have tangles of abnormal blood vessels (hemangiomas) in various parts of the eye. When these abnormal blood vessels develop in the network of blood vessels at the back of the eye (choroid), it is called a diffuse choroidal hemangioma and occurs in about one-third of individuals with Sturge-Weber syndrome. A diffuse choroidal hemangioma can cause vision loss. When
present, the eye abnormalities typically occur on the same side of the head as the port-
weight birthmark.

Frequency
Sturge-Weber syndrome is estimated to affect 1 in 20,000 to 50,000 individuals.

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

The \textit{GNAQ} gene mutation that causes Sturge-Weber syndrome results in the
production of a protein with impaired function. As a result, the altered \textit{G}\alpha \textit{q} 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.

Inheritance Pattern
Sturge-Weber syndrome is not inherited. The mutation that causes this disorder is
somatic, which means it occurs after conception. In Sturge-Weber syndrome, the
mutation is thought to occur in a cell during early development before birth. As that
cell continues to grow and divide, the cells derived from it, specifically certain cells in
the brain, eyes, and skin that are involved in blood vessel formation, also have the
mutation, while the body’s other cells do not. This situation is called mosaicism. The
mosaic nature of the mutations helps to explain why the abnormal blood vessel growth
occurs in some parts of the body but not in others.

Other Names for This Condition
- angiomatosis aculoorbital-thalamic syndrome
- encephalofacial hemangiomatosis
- encephalofacial hemangiomatosis syndrome
- meningo-oculo-facial angiomatosis
- meningofacial angiomatosis-cerebral calcification syndrome
- neuroretinoangiomatosis
- phakomatosis, Sturge-Weber
- Sturge-Weber-Dimitri syndrome
• Sturge-Weber-Krabbe syndrome
• SWS

Diagnosis & Management

Genetic Testing Information
• What is genetic testing?
  https://primer/testing/genetictesting
• Genetic Testing Registry: Sturge-Weber syndrome

Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov

Other Diagnosis and Management Resources
• Boston Children's Hospital: Sturge-Weber Syndrome Clinic
  http://www.childrenshospital.org/Centers-and-Services/Programs/O_-_Z/sturge-weber-syndrome-clinic-program
• Children's Hospital of Philadelphia: Capillary Vascular Malformations: Port Wine Stains
  https://www.chop.edu/conditions-diseases/capillary-vascular-malformations-port-wine-stains
• Children's Hospital of Philadelphia: Neurocutaneous Syndromes in Children
  https://www.chop.edu/conditions-diseases/neurocutaneous-syndromes-children
• Great Ormond Street Hospital for Children (UK): Sturge-Weber Clinic
• Great Ormond Street Hospital for Children (UK): Sturge-Weber Clinic
• Kennedy Krieger Institute: Hunter Nelson Sturge-Weber Syndrome Center
• MedlinePlus Encyclopedia: Sturge-Weber Syndrome
  https://medlineplus.gov/ency/article/001426.htm
• The Sturge-Weber Foundation: Centers of Excellence
  https://sturge-weber.org/for-patients/
Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Port-wine Stain
  https://medlineplus.gov/ency/article/001475.htm

- Encyclopedia: Sturge-Weber Syndrome
  https://medlineplus.gov/ency/article/001426.htm

- Health Topic: Vascular Diseases
  https://medlineplus.gov/vasculardiseases.html

Genetic and Rare Diseases Information Center

- Sturge-Weber syndrome

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Sturge-Weber Syndrome Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Sturge-Weber-Syndrome-Information-Page

Educational Resources

- Cincinnati Children’s Hospital
  https://www.cincinnatichildrens.org/health/s/sturge-weber-syndrome

- Johns Hopkins Health Library
  https://www.hopkinsmedicine.org/health/conditions-and-diseases/sturgeweber-syndrome

- KidsHealth from Nemours: Neurocutaneous Syndromes

- MalaCards: sturge-weber syndrome
  https://www.malacards.org/card/sturge_weber_syndrome

- Merck Manual Consumer Version

- Orphanet: Sturge-Weber syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3205

- Seattle Children's Hospital
  https://www.seattlechildrens.org/conditions/common-childhood-conditions/sturge-weber-syndrome-sws
Patient Support and Advocacy Resources

- American Foundation for the Blind
  https://www.afb.org/

- Contact a Family (UK)
  https://contact.org.uk/advice-and-support/medical-information/conditions/s/sturge-weber-syndrome/

- Epilepsy Foundation

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/sturge-weber-syndrome/

- Rare Diseases Clinical Research Network: Brain Vascular Malformation Consortium
  https://www.rarediseasesnetwork.org/cms/BVMC/Learn-More/Disorder-Definitions#SWS

- Sturge-Weber Syndrome Community
  https://swscommunity.org/

- The Sturge-Weber Foundation
  https://sturge-weber.org/

- Vascular Birthmarks Foundation
  https://birthmark.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Sturge-Weber+Syndrome%5BMAJR%5D%29+AND+%28Sturge-Weber+syndrome%5BTI%5D%29+AND+english%5Bl%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- STURGE-WEBER SYNDROME
  http://omim.org/entry/185300

Medical Genetics Database from MedGen

- Sturge-Weber syndrome
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27268758

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26564078

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27238857

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23656586
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3749068/


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