CONDITION

Sturge-weber syndrome

Sturge-Weber syndrome is a condition affecting blood vessels in the skin, brain, and sometimes the eyes.

WHAT IT IS
Congenital blood-vessel condition affecting the skin, brain, and eyes

BODY SYSTEM
Skin, nervous system, and eyes

MAIN SYMPTOMS
Port-wine birthmark, seizures, glaucoma, one-sided weakness

WHAT CAUSES IT
Random genetic change after conception, not usually inherited

HOW IT IS DIAGNOSED
Contrast-enhanced brain MRI, skin assessment, and eye examination

MAIN TREATMENT
Seizure control, eye care, laser therapy, and rehabilitation

CAN IT BE CURED?
No - underlying blood-vessel changes remain

IS IT AN EMERGENCY?
Sometimes - prolonged seizure or sudden stroke-like symptoms

LONG-TERM OUTLOOK
Many live fulfilling lives; neurological effects differ widely

Overview

Sturge-Weber syndrome is a condition involving unusual blood vessels in the skin, the covering of the brain, and sometimes the eyes. It is present from birth, although some effects may not appear until later.

A common visible feature is a port-wine birthmark. This is a flat red, pink, or purple area caused by widened tiny blood vessels in the skin. In Sturge-Weber syndrome, it is usually on one side of the face, often over the forehead or upper eyelid.

Abnormal blood vessels may also develop in the thin tissues covering the brain. This is called a leptomeningeal vascular malformation. It can affect blood flow and lead to seizures, headaches, weakness, or difficulties with development and learning.

Some people develop glaucoma, meaning increased pressure inside the eye that can damage the optic nerve. Glaucoma may be present in infancy or develop later.

The effects of Sturge-Weber syndrome cover a wide range. Some people have a facial birthmark with few other problems. Others have frequent seizures, vision loss, or significant physical and learning disabilities. A port-wine birthmark by itself does not mean that someone has Sturge-Weber syndrome.

Symptoms

The port-wine birthmark is usually visible at birth. It may become darker or thicker over time. Small raised areas can develop in adulthood. A birthmark affecting the forehead or upper eyelid is more closely associated with brain or eye involvement than one limited to other parts of the face.

Seizures are a common neurological symptom and often begin in infancy or early childhood. A seizure may cause stiffening, rhythmic jerking, staring, reduced awareness, or unusual movements. Symptoms may affect one side of the body.

Other possible symptoms include:

Glaucoma may cause an enlarged or cloudy eye in a baby. Older children and adults may have blurred vision, eye pain, redness, or halos around lights. However, glaucoma can develop without obvious symptoms, which is why regular eye checks matter.

Not everyone has every feature. Brain and eye involvement may occur on the same side as the facial birthmark, but the pattern is not identical in every person. Rarely, brain involvement occurs without a noticeable facial mark.

- Weakness or reduced movement on one side
- Migraine-like headaches, sometimes with an aura
- Short-lived episodes of weakness, speech difficulty, or vision change
- Delayed development or learning difficulties
- Problems with attention, memory, behaviour, or mood
- Loss of part of the field of vision
- Poor coordination or difficulty walking

Causes and risk factors

Sturge-Weber syndrome is caused by a genetic change that usually happens by chance shortly after conception. Most cases involve a gene called GNAQ , which helps control how cells grow and communicate.

The change affects only some cells. This is called somatic mosaicism . Which cells carry the change helps determine whether the skin, brain coverings, eyes, or a combination of these areas are involved.

Sturge-Weber syndrome is not usually inherited from a parent and is not caused by anything someone did during pregnancy. People with the condition generally do not pass it to their children through the usual pattern of genetic inheritance.

Having a facial port-wine birthmark does not automatically mean that a child has Sturge-Weber syndrome. The chance of associated brain or eye changes is higher when the birthmark includes the forehead, scalp, or upper eyelid. A specialist assessment considers the birthmark's location and any neurological or eye symptoms.

Diagnosis

Diagnosis is based on the pattern of skin, brain, and eye findings. A clinician will examine the birthmark and ask about seizures, headaches, development, movement, vision, and family history.

A brain MRI is the main imaging test for suspected brain involvement. Contrast material may be given into a vein to make abnormal blood vessels easier to see. MRI can show changes in the brain coverings, underlying brain tissue, and veins.

An early MRI in a baby does not always show the changes clearly. If the first scan is inconclusive, the specialist team may recommend observation or later imaging based on the child's symptoms and risk pattern. Repeated scans are not automatically needed for everyone.

An electroencephalogram, or EEG, records the brain's electrical activity. It can help assess seizures and guide treatment, but it does not confirm Sturge-Weber syndrome by itself.

A detailed eye examination checks vision, the optic nerve, eye size, and pressure inside the eye. These checks may need to continue throughout life because glaucoma can appear later.

A routine blood or saliva genetic test often cannot confirm the condition. Because the genetic change is present in only some cells, it may not be found outside affected tissue.

Treatment and management

There is no treatment that removes all the underlying blood-vessel changes. Care focuses on controlling symptoms, protecting sight, supporting development, and helping the person take part in daily life.

Anti-seizure medicines are used when seizures occur. The choice depends on the type of seizure, age, other health needs, and possible side effects. Keeping a record of seizure timing, duration, and recovery can help the neurology team judge how well treatment is working.

If seizures continue despite medicines, specialist assessment may include epilepsy surgery. Surgery is considered only after detailed testing shows where seizures begin and how treatment could affect movement, language, and vision.

Some specialists use low-dose aspirin for selected people with repeated stroke-like episodes or particular neurological concerns. Evidence is limited, and aspirin can increase bleeding or bruising. It should only be used when a specialist has assessed the possible benefits and risks.

Glaucoma may be treated with eye drops, surgery, or both. Treatment aims to lower eye pressure and protect the optic nerve. More than one procedure may be needed, and long-term monitoring remains important even when pressure improves.

Pulsed-dye laser therapy can lighten a port-wine birthmark by targeting blood vessels in the skin. Several sessions may be needed, and the mark may not disappear completely. Treatment can also be considered later if the skin becomes thicker or develops raised areas.

Physiotherapy, occupational therapy, speech and language therapy, and educational support may help with weakness, movement, communication, and learning. Headaches and emotional or behavioural difficulties can also be assessed and treated.

Complications

Repeated seizures can affect safety, learning, and quality of life. A prolonged seizure is called status epilepticus and requires urgent treatment. Some people develop epilepsy that is difficult to control with medicines.

Changes in brain blood flow can lead to stroke-like episodes. These may cause temporary or lasting weakness, speech problems, visual changes, or reduced awareness. They are not always caused by the same process as a typical blood-clot stroke, but sudden symptoms still need urgent assessment.

Long-term neurological effects can include one-sided weakness, tight muscles, reduced coordination, visual-field loss, and learning disability. The degree of disability is different for each person.

Untreated glaucoma can cause permanent loss of vision. Because damage may begin before symptoms are noticed, scheduled eye examinations are important.

A facial birthmark can also affect confidence and emotional wellbeing. Children and adults may experience unwanted attention, questions, or bullying. Psychological support and contact with others who have visible differences may help.

Prevention

Sturge-Weber syndrome cannot currently be prevented. The genetic change happens randomly during early development and is not usually inherited.

Nothing done before or during pregnancy is known to cause the condition. Parents should not blame themselves.

Complications cannot always be prevented, but planned neurological and eye follow-up can identify changes early. Taking seizure medicine as directed, attending glaucoma checks, and following an individual seizure safety plan can reduce some risks.

Living with it and what to expect

Sturge-Weber syndrome is lifelong, but its course is not the same for everyone. The extent of brain involvement, age when seizures begin, seizure control, glaucoma, and access to rehabilitation can all influence long-term needs.

The port-wine birthmark usually grows in proportion with the face. It may darken, thicken, or become more raised over the years. Laser treatment can lighten it, but later treatment or ongoing skin care may still be needed.

Neurological symptoms often become clear during childhood. Seizures may become well controlled, continue at intervals, or remain difficult to manage. Weakness and learning needs can change as a child grows and faces more complex physical or school tasks.

Glaucoma requires long-term attention because it can develop or return at different ages. Regular eye examinations may be recommended even when vision seems normal and previous pressure readings were reassuring.

Many people attend school, work, form relationships, and live independently. Others need continuing help with mobility, communication, personal care, or decision-making. Support should reflect the person's abilities and goals rather than the appearance of the birthmark alone.

Routine follow-up may involve neurology, ophthalmology, dermatology, rehabilitation services, and educational or psychological support. Adults may need help moving from children's services to adult care so that seizure management and eye monitoring continue without interruption.

When to get medical help

Call emergency services now for:

Follow an existing emergency seizure plan if one has been provided.

Contact the usual healthcare team promptly if seizures become more frequent, change in pattern, or take longer to recover from. New headaches, changes in walking, loss of previously gained skills, or new problems at school also deserve assessment.

Arrange an eye review for persistent redness, light sensitivity, blurred vision, changes in eye size, or concerns about sight. Keep routine neurology and eye appointments even when symptoms appear stable.

- A first seizure
- A seizure lasting five minutes or longer
- Repeated seizures without normal recovery between them
- New facial drooping, one-sided weakness, speech difficulty, confusion, or sudden vision loss
- Severe breathing difficulty, serious injury, or failure to recover after a seizure
- Sudden severe eye pain with redness, vomiting, or reduced vision

Common questions

Is Sturge-Weber syndrome inherited?

Usually not. The genetic change generally occurs by chance after conception and affects only some cells. The condition rarely follows an inherited family pattern.

Does every port-wine birthmark mean Sturge-Weber syndrome?

No. Most port-wine birthmarks do not mean that someone has Sturge-Weber syndrome. Marks involving the forehead or upper eyelid are more likely to need assessment for brain or eye involvement.

Can Sturge-Weber syndrome get worse with age?

Some features can change. A birthmark may darken or thicken, glaucoma can appear later, and seizure or headache patterns may change. This does not mean that everyone will develop increasing disability.

Can the port-wine birthmark be removed?

Pulsed-dye laser treatment can often lighten it, but complete removal is not guaranteed. Several sessions may be needed, and some colour or thickening can return over time.

Can someone with Sturge-Weber syndrome live a normal life?

Many people live active and independent lives. Others need long-term support because of epilepsy, vision loss, weakness, or learning disability. Appearance alone does not predict neurological ability.

Is there a cure for Sturge-Weber syndrome?

There is no cure for the underlying blood-vessel changes. Treatment can control many seizures, lower eye pressure, lighten the birthmark, and support movement, learning, and independence.

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