CONDITION

Encephalofacial angiomatosis

A condition present from birth that causes abnormal blood vessels in the brain, face, or eyes.

WHAT IT IS
Congenital blood-vessel malformation affecting the brain, face, or eyes

BODY SYSTEM
Nervous system, skin, and eyes

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

WHAT CAUSES IT
Random, non-inherited mosaic GNAQ gene change

HOW IT IS DIAGNOSED
Clinical assessment, contrast brain MRI, complete eye examination

MAIN TREATMENT
Seizure control, eye treatment, laser therapy, rehabilitation

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

IS IT AN EMERGENCY?
Sometimes - prolonged seizures, sudden weakness, or acute vision loss

LONG-TERM OUTLOOK
Often manageable, though neurological and eye problems can persist

Overview

Encephalofacial angiomatosis is a condition involving abnormal blood vessels in the brain, skin, or eyes. It is present from birth, although some effects may not become clear until later. It is often called Sturge-Weber syndrome.

A common feature is a flat red or purple birthmark called a port-wine birthmark, often affecting one side of the forehead, scalp, or eyelid. However, not everyone with the condition has a facial birthmark, and most people with a port-wine birthmark do not have brain involvement.

Abnormal vessels may also form in the thin tissues covering the brain, called the meninges. They can interfere with blood flow and oxygen delivery, leading to seizures, weakness, headaches, learning differences, or stroke-like episodes.

The eyes can be affected by glaucoma. Increased pressure inside the eye can damage the optic nerve and reduce vision. Glaucoma may begin in infancy or later, so ongoing eye examinations are important even when vision seems normal.

Effects vary widely. Some people have only a birthmark or mild symptoms, while others need long-term treatment for seizures, movement difficulties, learning needs, or glaucoma.

Symptoms

Symptoms depend on which blood vessels are affected. They may be noticeable at birth, begin during infancy, or appear later.

Possible features include:

The birthmark may be pale at first but can become darker, thicker, or more textured. Soft tissue or bone beneath it may also grow more than expected.

Seizures often begin in childhood, but their timing and severity vary. Fever, illness, lack of sleep, or missed seizure medicine may make seizures more likely in someone with epilepsy.

Glaucoma may have no early symptoms. Possible signs include light sensitivity, tearing, cloudiness, pain, redness, or vision changes. Regular testing is more reliable than waiting for symptoms.

- A red, pink, or purple port-wine birthmark, often on the forehead or upper eyelid
- Seizures affecting one side or the whole body
- Weakness or reduced movement on one side
- Developmental delay, learning difficulties, or attention problems
- Headaches, including migraine-like headaches
- Brief stroke-like episodes involving weakness, speech, or vision
- Glaucoma, sometimes causing an enlarged eye in an infant
- Reduced vision or loss of part of the visual field
- Differences in balance, coordination, speech, or muscle stiffness

Causes and risk factors

Encephalofacial angiomatosis usually results from a change in a gene called GNAQ . The change happens by chance in one cell early in development after conception.

Some groups of cells carry the change while others do not, a pattern called mosaicism . This explains why only certain areas of the skin, brain, or eye may be affected.

The gene change is not usually present in the egg or sperm, so the condition is not considered inherited. Parents do not usually pass it to their children, and it is not caused by anything someone did during pregnancy.

There are no established lifestyle risk factors. A port-wine birthmark involving the forehead or upper eyelid is associated with a greater chance of brain or eye involvement, but its location does not prove that the brain is affected.

Diagnosis

Diagnosis begins with a medical history and examination. A clinician will assess the birthmark, neurological development, movement, vision, and possible seizures.

A brain MRI may be used to look for abnormal vessels in the tissues covering the brain. Contrast material given into a vein can make them easier to see. MRI can also show changes in nearby brain tissue.

An early MRI can occasionally appear normal even when symptoms develop later. Decisions about when to scan or repeat imaging depend on symptoms, age, birthmark location, and specialist assessment. MRI in a young child may require sedation or a general anaesthetic.

An EEG, or electroencephalogram, records the brain's electrical activity. It can help investigate seizures and identify where they may begin, but it cannot by itself confirm or exclude the condition.

A complete eye examination checks eye pressure, the optic nerve, the front of the eye, and the retina. Examinations may need to continue throughout life because glaucoma can develop later.

Additional assessments may cover hearing, speech, movement, learning, or visual fields. Genetic testing is not always useful because the gene change may occur only in affected tissue and may not appear in blood.

Treatment and management

There is no treatment that removes the underlying blood-vessel changes. Management aims to control symptoms, protect vision and brain function, and support development.

Anti-seizure medicines are used when epilepsy is present. The choice depends on seizure type,

age, other health needs, and side effects. A written seizure plan may explain when to give prescribed rescue medicine and seek emergency help.

If seizures continue despite appropriate medicines, a specialist epilepsy team may consider surgery. This is reserved for carefully selected people, particularly when seizures arise from a severely affected area on one side of the brain.

Some specialist teams use low-dose aspirin in selected people to reduce clotting within abnormal vessels and possibly limit stroke-like episodes. Evidence is limited, and aspirin can cause bleeding. It should only be used under a specialist familiar with the condition.

Glaucoma may be treated with eye drops, surgery, or both. Treatment lowers eye pressure to protect the optic nerve, and more than one procedure may be needed.

Pulsed-dye laser treatment can lighten a port-wine birthmark by targeting abnormal surface vessels. Several sessions are often required, and the birthmark may not disappear completely. Laser treatment does not affect abnormal vessels in the brain or eye.

Physical, occupational, and speech and language therapies can support movement, daily activities, communication, and swallowing. Educational assessment can identify classroom support. Headaches and mental health concerns can also be treated.

Complications

Repeated or prolonged seizures can affect safety, learning, and quality of life. A seizure lasting several minutes or repeated seizures without recovery may become status epilepticus, which requires emergency treatment.

Reduced blood flow around abnormal brain vessels can cause temporary stroke-like episodes or lasting weakness. Over time, affected brain tissue may shrink or develop calcium deposits.

Glaucoma can permanently damage the optic nerve if pressure remains too high. Because early glaucoma may be painless, missing routine eye follow-up can allow unnoticed vision loss.

Other complications include reduced visual fields, muscle tightness, walking or speech difficulties, and developmental or learning disabilities. A visible facial birthmark may also affect confidence and social experiences. Psychological support and school planning may help.

Prevention

Encephalofacial angiomatosis cannot currently be prevented. The gene change occurs randomly during early development and is not linked to known actions before or during

pregnancy.

Early recognition and regular care may limit complications. This includes monitoring seizures, attending eye-pressure checks, following treatment plans, and arranging developmental support when needed.

Laser treatment may reduce darkening or thickening of a port-wine birthmark, but it does not prevent brain or eye involvement.

Living with it and what to expect

The abnormal blood vessels are present from birth. They do not spread from person to person or turn into cancer, but their effects can change over time.

Seizures may improve, remain stable, or become harder to control. Some children develop steadily, while others progress more slowly or lose skills after severe seizures or stroke-like episodes. Early therapy and educational support can help, but cannot guarantee a particular outcome.

Weakness on one side may remain stable or become more noticeable as physical demands increase. Braces, stretching, therapy, mobility aids, or treatment for muscle tightness may support independence.

A port-wine birthmark usually persists and may darken or thicken. Laser treatment can improve its appearance, but decisions are personal and may reflect skin changes, discomfort, cost, and access to care.

Glaucoma remains a lifelong possibility. Someone with normal eye pressure during childhood may still need checks as an adult.

Many people attend school, work, build relationships, and take part in everyday activities. Those with severe epilepsy, visual loss, or major movement and learning difficulties may need substantial long-term assistance. Regular neurology and eye care should continue during the transition from children's to adult services.

When to get medical help

Call emergency services for:

Sudden neurological symptoms may resemble previous stroke-like episodes, but still need urgent assessment because stroke or another serious problem cannot be excluded at home.

Arrange prompt medical review for new seizures, a clear increase in seizure frequency, new

weakness, loss of developmental skills, persistent headaches, or vision changes. An enlarging, cloudy, or persistently watery eye in a baby also needs prompt eye assessment.

Routine follow-up includes scheduled eye-pressure checks, seizure reviews, developmental or educational assessments, and monitoring of the birthmark. These visits remain important even when symptoms are stable.

- A first seizure, especially without an established seizure plan
- A seizure lasting longer than five minutes, or longer than the person's emergency plan allows
- Repeated seizures without waking or returning to normal between them
- Breathing difficulty, serious injury, or persistent unresponsiveness after a seizure
- Sudden weakness, facial drooping, speech trouble, severe confusion, or loss of vision
- Sudden severe eye pain, redness, vomiting, halos around lights, or rapid vision loss

Common questions

Is encephalofacial angiomatosis inherited?

Usually not. The gene change occurs by chance after conception and is present in only some cells. The chance of a parent passing the condition to another child is thought to be very low.

Does a port-wine birthmark mean the brain is affected?

No. Most port-wine birthmarks occur without brain involvement. A birthmark on the forehead or upper eyelid raises concern, but examination and appropriate imaging are needed to assess the brain and eyes.

Can encephalofacial angiomatosis be cured?

There is no cure for the underlying blood-vessel malformations. Treatment can control seizures, lower eye pressure, lighten the birthmark, improve function, and reduce complications.

Will the seizures get worse with age?

Not necessarily. Some people achieve good control, while others have persistent or changing seizures. The course depends partly on the extent of brain involvement and response to treatment.

Can laser treatment remove the birthmark completely?

It may lighten the birthmark substantially, but complete removal is not guaranteed. Several treatments may be needed, and the colour can return or deepen over time.

Can someone with this condition live a full life?

Yes. Many people take part in school, work, relationships, and community life. Support needs range from occasional monitoring to lifelong help with epilepsy, vision, movement, or learning.

Sources

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