CONDITION

Syndactylic oxycephaly

A rare genetic condition in which skull bones fuse early and fingers and toes are joined

WHAT IT IS
Genetic condition affecting skull, face, hands, and feet

BODY SYSTEM
Bones, brain, breathing, hearing, vision, hands, and feet

MAIN SYMPTOMS
Unusual head shape, midface flattening, joined fingers, joined toes

WHAT CAUSES IT
FGFR2 gene change, usually new rather than inherited

HOW IT IS DIAGNOSED
Physical features, skull imaging, and FGFR2 genetic testing

MAIN TREATMENT
Staged surgery and lifelong specialist monitoring

CAN IT BE CURED?
No, the genetic change remains lifelong

IS IT AN EMERGENCY?
Sometimes, with breathing trouble or raised brain pressure

LONG-TERM OUTLOOK
Many reach adulthood; needs and abilities differ widely

Overview

Syndactylic oxycephaly is a rare genetic condition affecting how the skull, face, hands, and feet develop. In current medical literature, the term usually refers to Apert syndrome.

"Oxycephaly" describes a tall or pointed head shape. It happens when some of the joints between the skull bones fuse too early. These joints are called sutures, and their early fusion is called craniosynostosis.

"Syndactylic" refers to syndactyly, meaning fingers or toes are joined. In this condition, several fingers and toes may be connected by skin and deeper tissues, including bone.

The condition is present from birth. Its effects differ considerably between people. Some need several operations and ongoing support, while others have fewer medical or developmental difficulties.

Early assessment matters because restricted skull growth can sometimes increase pressure around the brain. Differences in the middle of the face can also affect breathing, sleep, hearing, eyesight, teeth, and feeding.

Symptoms

The most recognisable features involve the skull, face, hands, and feet.

Skull growth may produce:

Early fusion most often affects the sutures running over the head from ear to ear. The exact head shape depends on which sutures have fused and how the brain and remaining skull bones grow.

Facial features may include a flat or underdeveloped middle face, a small upper jaw, widely spaced or prominent eyes, and a beaked nose. The eyes may not line up together. Shallow eye sockets can make it difficult for the eyelids to cover the eyes fully.

The fingers are commonly joined in a symmetrical pattern on both hands. The thumbs may be partly separate, while the middle fingers may be joined by skin, soft tissue, or bone. The feet may have similar joining of the toes. Hand function depends on the bones and joints involved, not only on how the fingers look.

Other possible features include:

Not everyone has every feature. Intelligence and development range widely and cannot be predicted from head shape alone.

- A tall, short, or pointed head shape
- A broad forehead
- Flattening at the back of the head
- A soft spot on the top of the head that closes later than expected
- Noisy breathing, mouth breathing, or pauses in breathing during sleep
- Repeated ear infections or hearing loss
- Crowded teeth and difficulty bringing the upper and lower teeth together
- Feeding or speech difficulties
- Heavy sweating and intolerance of hot weather
- Acne that may become more noticeable during adolescence

- Differences in learning, development, or coordination

Causes and risk factors

Syndactylic oxycephaly is caused by a harmful change in the FGFR2 gene. This gene helps control signals involved in bone growth and development. The altered signal causes some skull bones to join too early and affects formation of the face, hands, and feet.

Most affected children are born to parents who do not have the condition. In these cases, the genetic change occurred newly in the egg, sperm, or very early embryo. This is called a de novo change.

Older paternal age is associated with a higher chance of a new FGFR2 change. However, most children born to older fathers do not have this condition, and nothing a parent did during pregnancy caused it.

The condition follows an autosomal dominant inheritance pattern. This means a person with the condition has a 1 in 2 chance of passing the altered gene to each child. The chance is the same for every pregnancy.

When neither parent has the condition, the chance of it happening again is usually low but is not always zero. A genetics professional can explain recurrence chances using the family's test results.

Diagnosis

The condition may be suspected before birth if an ultrasound shows an unusual skull shape or joined fingers and toes. These findings are not always clear on routine pregnancy scans. Prenatal genetic testing may be considered when a specific familial FGFR2 change is known or when imaging strongly suggests the condition.

After birth, diagnosis begins with an examination of the head, face, hands, and feet. A clinician will also assess breathing, feeding, eye protection, hearing, movement, and development.

Imaging helps identify which skull sutures have fused. A low-dose CT scan can show the skull bones and may be used when the diagnosis or surgical anatomy is unclear. MRI does not show bone as clearly, but it may be used to examine the brain, fluid spaces, or other structures without radiation.

A blood or saliva test can look for a disease-causing change in the FGFR2 gene. Finding a characteristic FGFR2 change confirms the genetic diagnosis and supports family testing and

genetic counselling.

Further assessments may include hearing tests, eye examinations, sleep studies, dental imaging, developmental assessment, and scans for fluid build-up around the brain. These tests look for associated problems rather than confirming the condition itself.

Treatment and management

There is no treatment that removes the FGFR2 gene change. Care focuses on protecting the brain, keeping the airway and eyes safe, improving hand function, and supporting hearing, speech, learning, and dental health.

Treatment is usually coordinated by a specialist craniofacial team. This may include skull and facial surgeons, neurosurgeons, hand surgeons, geneticists, dentists, ear specialists, eye specialists, respiratory teams, therapists, and psychologists.

Skull surgery is commonly performed during infancy or early childhood. It creates more room for the growing brain and reshapes the skull. The operation chosen and its timing depend on the fused sutures, head growth, signs of raised pressure, and the centre's surgical approach.

Some people later need surgery to move the middle of the face forward. This may improve breathing, protect prominent eyes, and help the upper and lower teeth meet more effectively. Jaw or dental treatment may continue into adolescence or adulthood.

Hand surgery can separate selected fingers and improve the ability to grip, pinch, and manage daily tasks. It is often done in stages because skin, nerves, blood vessels, tendons, joints, and bones may all be involved. Fully typical hand movement may not be possible, but meaningful functional improvement often is.

Other management can include:

Treatment plans are individual. Not every visible difference requires surgery, and the likely benefits must be weighed against the burdens and risks of each procedure.

- Treatment for sleep-related breathing obstruction
- Glasses, eye lubrication, or surgery for eye alignment or exposure
- Hearing aids or treatment of persistent middle-ear fluid
- Dental and orthodontic care
- Speech, occupational, and physical therapy
- Educational assessment and learning support

- Skin care for acne and heavy sweating
- Emotional and social support

Complications

Craniosynostosis can restrict skull growth and contribute to raised pressure around the brain. Signs may be subtle, especially in young children. Regular head measurements, eye examinations, developmental review, and specialist follow-up help detect problems.

Fluid can sometimes collect in or around the brain. This includes hydrocephalus , an increase in fluid within the brain's internal spaces. Some people also develop a Chiari malformation, where tissue at the lower back of the brain extends into the opening at the base of the skull.

The underdeveloped middle face can narrow the nose and throat. This may cause obstructive sleep apnoea, in which breathing repeatedly pauses during sleep. Untreated severe obstruction can affect sleep quality, behaviour, learning, growth, and heart strain.

Prominent eyes may become dry or injured if the eyelids cannot close fully. Hearing loss may result from repeated middle-ear problems or differences in the ear structures. Both hearing and vision difficulties can affect communication and learning if they are not recognised.

Dental crowding, an abnormal bite, and reduced hand movement may affect eating and self-care. Operations can also have complications, including bleeding, infection, scarring, and the need for further procedures as the skull and face grow.

Prevention

Syndactylic oxycephaly cannot usually be prevented. It results from a genetic change, and most cases arise unexpectedly rather than from anything done before or during pregnancy.

Genetic counselling can help someone with the condition, or parents of an affected child, understand inheritance and reproductive options. If the family's FGFR2 change is known, testing during pregnancy or testing embryos created through in vitro fertilisation may be possible. These are personal choices rather than ways to prevent the condition in an individual who already has it.

Living with it and what to expect

The condition is lifelong, but its needs change over time. Infancy and early childhood often

involve the most intensive monitoring because the brain, skull, airway, eyes, and hands are developing quickly.

Some children have staged skull and hand operations. School-age care may focus more on hearing, speech, learning, dental development, sleep, and social wellbeing. Facial or jaw procedures may be considered later as growth continues.

Developmental outcomes differ widely. Some people attend mainstream school without major learning support. Others have intellectual disability or need help with language, attention, coordination, or everyday skills. Early support can address specific needs, but it cannot predict or guarantee a particular outcome.

Adults may continue to need dental, hearing, eye, breathing, neurological, or psychological care. Some require further surgery, while others mainly have routine monitoring. Many adults study, work, form relationships, and live with differing levels of independence.

Visible facial and hand differences can affect confidence and expose a person to unwanted attention. Age-appropriate explanations, contact with peer-support organisations, and access to mental health support may help. Care should include the person's own goals, not only medical or cosmetic concerns.

When to get medical help

Arrange routine or prompt medical review for new or gradually worsening concerns such as:

Seek urgent medical assessment for a severe headache with repeated vomiting, unusual sleepiness, confusion, new weakness, a seizure, or a sudden change in vision. These symptoms can have several causes but may indicate increased pressure or another neurological problem.

Call emergency services for severe breathing difficulty, blue or grey lips, prolonged pauses in breathing, collapse, or difficulty waking the person. A painful, very red, or suddenly cloudy eye also needs urgent assessment, particularly if the eyelids do not protect the eye.

Routine specialist appointments remain important even when the person seems well. Raised pressure, hearing loss, and sleep-related breathing problems are not always obvious at home.

- Increasing headaches or a change in headache pattern
- Snoring, restless sleep, daytime tiredness, or witnessed breathing pauses
- New squinting, reduced vision, eye redness, or difficulty closing an eye
- Changes in hearing, speech, school progress, balance, or coordination
- Repeated ear infections or dental pain

- Loss of hand function or pain affecting daily activities

Common questions

Is syndactylic oxycephaly the same as Apert syndrome?

The term is an older descriptive name generally used for Apert syndrome. "Syndactylic" describes joined fingers or toes, while "oxycephaly" describes the altered skull shape. Current genetic and clinical sources more commonly use the name Apert syndrome.

Can someone with this condition have typical intelligence?

Yes. Intelligence and learning ability range widely. Some people have typical intellectual development, while others have mild to significant learning difficulties. Hearing, vision, sleep, raised skull pressure, and access to early support may also affect development and school progress.

Can joined fingers be separated?

Often, selected fingers can be separated surgically. The result depends on which bones, joints, nerves, tendons, and blood vessels are shared. Surgery aims to improve useful movement and independence rather than create a typical-looking or fully typical-functioning hand.

Does skull surgery cure the condition?

No. Surgery can make more room for the brain and change the skull shape, but it does not remove the genetic change. Skull and facial growth continue, so ongoing monitoring and sometimes further procedures are needed.

Can it be passed on to children?

Yes. A person with the condition has a 1 in 2 chance of passing the altered FGFR2 gene to each child. Most initial cases are caused by a new genetic change, so the parents are usually unaffected. Genetic testing can clarify the change present in a particular family.

Is life expectancy affected?

Many people live into adulthood. Life expectancy is influenced by the severity and treatment of complications, especially breathing obstruction, raised pressure around the brain, and neurological problems. An individual outlook cannot be predicted from appearance alone.

Sources

- GeneReviews: Apert Syndrome , NCBI Bookshelf

- MedlinePlus Genetics: Apert syndrome , U.S. National Library of Medicine
- Orphanet: Apert syndrome , INSERM
- Mathijssen IMJ and colleagues. Updated Guideline on Treatment and Management of Craniosynostosis , Journal of Craniofacial Surgery , 2020
- Dias MS and colleagues. Identifying the Misshapen Head: Craniosynostosis and Related Disorders , American Academy of Pediatrics, Pediatrics , 2020
- Apert syndrome , Great Ormond Street Hospital for Children NHS Foundation Trust