CONDITION

Francois dyscephaly syndrome

Francois dyscephaly syndrome is a rare condition affecting facial growth, eyes, teeth, hair, skin, and sometimes breathing

WHAT IT IS
Rare condition affecting facial and skull development

BODY SYSTEM
Face and skull, eyes, teeth, skin, and airways

MAIN SYMPTOMS
Distinctive facial growth, sparse hair, dental problems, vision problems

WHAT CAUSES IT
Unknown, usually occurring without a family history

HOW IT IS DIAGNOSED
Clinical examination, eye assessment, dental assessment, and imaging

MAIN TREATMENT
Coordinated care for vision, breathing, feeding, and teeth

CAN IT BE CURED?
No - structural differences are lifelong

IS IT AN EMERGENCY?
Sometimes - severe breathing trouble or blue lips

LONG-TERM OUTLOOK
Many reach adulthood; breathing and eye complications influence health

Overview

Francois dyscephaly syndrome is a very rare condition affecting development before birth. It mainly changes skull and facial growth but can also affect the eyes, teeth, hair, skin, height, and upper airway.

The condition is commonly called Hallermann-Streiff syndrome in medical literature. Older descriptions may use terms such as "bird-like face," referring to a pattern that includes a small lower jaw and a thin, prominent nose. It is not a judgment about someone's appearance.

The condition is present from birth, although features such as missing permanent teeth may become clearer during childhood. Differences in the jaw and airway can affect breathing and feeding, while eye changes may reduce vision. Dental development can affect chewing, speech, and facial growth. Severity varies widely.

Intelligence is often unaffected. However, reduced vision, hearing problems, illness, feeding difficulties, or missed schooling can influence learning and development. Early support can help children participate at home, at school, and in the community.

Symptoms

The most recognizable features involve the head and face. A baby may have a small lower jaw, a narrow nose, and a small or unusually shaped head. The eyes may appear small or deep-set, and the soft spots between skull bones may remain open longer than expected.

Common features include:

A congenital cataract is clouding of the eye's natural lens that is present at birth. It may affect one or both eyes and interfere with visual development. Some people also develop glaucoma, which is raised pressure inside the eye, or other structural eye problems.

A small lower jaw can leave less room for the tongue and upper airway. This may cause noisy breathing, snoring, pauses in breathing during sleep, or difficulty breathing while lying flat. Infants may tire during feeds or struggle to coordinate sucking, swallowing, and breathing.

Dental findings can include delayed tooth eruption, absent teeth, weak enamel, or unusually small or irregular teeth. These changes may contribute to decay, gum problems, difficulty chewing, or unclear speech.

Not everyone has every feature, and different parts of the condition may vary in severity.

- A small lower jaw and distinctive facial shape
- Small eyes, congenital cataracts, or other vision problems
- Sparse scalp hair, eyebrows, or eyelashes
- Thin skin, especially over the nose
- Missing, small, poorly formed, or crowded teeth
- Short stature with generally proportional growth
- Feeding, swallowing, breathing, or speech difficulties

Causes and risk factors

The cause of Francois dyscephaly syndrome is unknown. It appears to involve an early difference in the development of facial bones and other tissues, but researchers have not identified one responsible gene or environmental trigger.

Most reported cases have occurred without a family history, and no consistent inheritance pattern has been confirmed. Parents should not assume that something they did before or during pregnancy caused the condition.

There are no established lifestyle, pregnancy, or parental risk factors. Because the condition is rare and its cause remains uncertain, estimating whether it will happen again in a family can be difficult.

A genetics professional can review the person's features and family history, look for other genetic conditions, and explain what is known about recurrence. A precise estimate may not be possible.

Diagnosis

Diagnosis is based mainly on the pattern of physical findings. No single blood test or genetic test confirms Francois dyscephaly syndrome.

A clinical genetics or craniofacial team may assess facial growth, skull shape, hair, skin, height, and development. Classic descriptions include distinctive facial structure, abnormal teeth, sparse hair, thin skin, small eyes, cataracts, and short stature, although a person may not have every feature.

Assessment may include:

Genetic testing may be offered when the diagnosis is uncertain, mainly to look for other conditions with similar facial, growth, eye, or skin findings. A negative result does not rule out Francois dyscephaly syndrome.

Diagnosis may take time because the condition is rare and some features develop with age. Review by clinicians familiar with craniofacial conditions can help.

- A complete eye examination, including cataract and eye-pressure checks
- Dental examination and dental X-rays when appropriate
- Hearing assessment
- Evaluation of feeding, swallowing, speech, and growth
- Sleep or airway testing for snoring or interrupted breathing
- Skull, jaw, or airway imaging for treatment planning

Treatment and management

There is no treatment that reverses the underlying developmental differences. Care focuses on protecting breathing and vision, supporting nutrition and communication, and managing dental and facial problems.

Treatment usually involves several specialists, such as eye doctors, dentists, orthodontists, airway specialists, craniofacial surgeons, genetics professionals, dietitians, and speech or feeding therapists.

Eye care should begin early because clear vision is important for visual development. Cataracts and other eye problems may require surgery, glasses, contact lenses, or treatment for reduced vision. Surgery can be technically complex when the eyes are unusually small, so planning is individualized.

Breathing problems may be managed with sleep assessment, positioning advice, treatment of airway blockage, breathing support, or surgery in selected cases. Surgical and dental teams must know about the small jaw and possible narrow airway before sedation or general anesthesia because placing a breathing tube may be difficult.

Feeding support may include a swallowing assessment, changes in food texture, nutrition support, or therapy. More intensive support may be needed if a child cannot take enough food safely by mouth.

Dental care may include decay prevention, replacement of missing teeth, orthodontics, or prosthetic teeth. Treatment is often staged as the jaws and remaining teeth develop.

Speech therapy can support speech clarity, language, and communication. Children with reduced vision may benefit from large print, suitable classroom seating, mobility support, and other educational accommodations.

Complications

A narrow upper airway may cause obstructive sleep apnea, in which breathing repeatedly slows or stops during sleep. Untreated sleep apnea can affect behavior, learning, growth, sleep quality, and heart or lung health.

Young children may be especially vulnerable to breathing problems during respiratory infections. A difficult airway also makes anesthesia more complex, making careful advance planning essential.

Eye complications include reduced vision from cataracts, glaucoma, or abnormal eye development. Vision may not fully recover if the brain did not receive clear images during early childhood. Regular follow-up remains important after cataract treatment.

Feeding and swallowing problems can cause poor growth or aspiration, when food or liquid enters the airway. Aspiration may lead to coughing, choking, or repeated chest infections.

Dental abnormalities can cause pain, decay, gum disease, chewing difficulties, and speech changes. Differences in appearance or communication may also lead to bullying or social isolation, so psychological and school support can be important.

Prevention

Francois dyscephaly syndrome cannot currently be prevented. Its cause is unknown, and no action during pregnancy has been shown to cause or prevent it.

There is no established screening test for people without symptoms or for routine use during pregnancy. Prenatal imaging may identify some facial or growth differences, but these are not specific enough to confirm the condition.

Genetic counseling may help affected people or parents considering another pregnancy. It can review whether another diagnosis should be considered and explain the limits of current testing.

Secondary problems may be reduced through regular eye examinations, dental care, growth monitoring, airway assessment, and early feeding or communication support.

Living with it and what to expect

Francois dyscephaly syndrome is lifelong. Facial, eye, hair, and skin differences generally remain, although their appearance may change as the skull and jaw grow. Short stature may continue into adulthood.

Health priorities often change over time. Infancy may involve breathing, feeding, cataracts, and growth. Dental development, speech, education, and social wellbeing may become more important during childhood. Adults may continue to need eye, dental, airway, and hearing care.

Some complications can be treated successfully, but several procedures or long-term follow-up may be needed. Vision may remain reduced after cataract treatment, dental rehabilitation may require revision, and sleep-related breathing problems can appear or become more noticeable with age.

Many people reach adulthood and participate in education, work, relationships, and community life. Support needs depend more on breathing, vision, feeding, and developmental needs than on the diagnosis alone.

Routine follow-up may include eye-pressure checks, dental reviews, hearing tests, growth and nutrition monitoring, and reassessment of sleep or breathing symptoms. Emotional support may help with medical stress, appearance-related concerns, or social experiences.

When to get medical help

Arrange a routine medical review for new or worsening snoring, daytime sleepiness, feeding difficulty, poor weight gain, dental pain, vision changes, or concerns about hearing, speech, or development. These concerns may not be emergencies, but they should not be ignored.

Seek urgent medical assessment if a baby or child repeatedly chokes during feeds, cannot take enough fluid, has signs of dehydration, or develops persistent coughing or breathing problems after eating.

Get emergency help for:

A respiratory infection also needs prompt assessment if breathing becomes much harder than usual, especially in an infant or someone with a known narrow airway.

- Severe difficulty breathing or struggling for each breath
- Blue, grey, or unusually pale lips or skin
- A prolonged pause in breathing or inability to wake normally
- Sudden severe eye pain, vomiting, or rapid loss of vision
- Choking with inability to breathe, cry, or cough

Common questions

Is Francois dyscephaly syndrome the same as Hallermann-Streiff syndrome?

Medical publications commonly use Hallermann-Streiff syndrome for the same pattern of facial, eye, dental, hair, skin, and growth differences. Older medical records may use different wording.

Is it inherited from a parent?

Most reported cases occur without a family history, and no consistent inheritance pattern has been established. Genetics assessment can check for similar conditions with known genetic causes.

Will my child have typical intelligence?

Many affected people have typical intelligence. Vision loss, hearing difficulties, illness, or

missed learning opportunities can still affect development. Individual assessment is more useful than assumptions based on the diagnosis.

Can the facial differences be corrected with surgery?

Some jaw, dental, airway, or facial concerns may be treated surgically, but no operation removes the condition. Decisions consider function, growth, potential benefits, and anesthesia risks.

Can congenital cataracts be treated?

Sometimes. Cataract surgery or other treatment may improve the opportunity for sight, but results depend on eye size, other abnormalities, and how early clear vision is established. Long-term eye follow-up remains important.

Does the condition get worse with age?

It is not usually considered steadily degenerative. The developmental differences remain, while needs change with growth. Dental problems, glaucoma, sleep apnea, and social or communication needs may require more attention later.

Sources

- Cohen MM Jr. "Hallermann-Streiff syndrome: a review." American Journal of Medical Genetics . 1991;41(4):488-499.
- National Organization for Rare Disorders. "Hallermann-Streiff Syndrome." Rare Disease Database .
- Orphanet. "Hallermann-Streiff syndrome." ORPHA:2108.
- National Institutes of Health, Genetic and Rare Diseases Information Center. "Hallermann-Streiff syndrome."
- Online Mendelian Inheritance in Man. "Hallermann-Streiff syndrome." OMIM 234100.