CONDITION

Fibrodysplasia ossificans progressiva

Fibrodysplasia ossificans progressiva is a rare genetic condition that gradually causes bone to form in muscles and other soft tissues.

WHAT IT IS
Progressive bone formation within muscles and connective tissues

BODY SYSTEM
Muscles, connective tissues, joints, and skeleton

MAIN SYMPTOMS
Malformed big toes, painful swellings, stiffness, reduced movement

WHAT CAUSES IT
Genetic ACVR1 change, usually not inherited

HOW IT IS DIAGNOSED
Clinical findings and ACVR1 genetic testing

MAIN TREATMENT
Specialist flare care, symptom relief, and targeted medicine

CAN IT BE CURED?
No - existing abnormal bone cannot be removed safely

IS IT AN EMERGENCY?
Sometimes - breathing trouble needs emergency care

LONG-TERM OUTLOOK
Progressive movement loss, with an unpredictable pace

Overview

Fibrodysplasia ossificans progressiva, usually called FOP, is a very rare genetic condition. It causes bone to form in muscles, tendons, ligaments, and other connective tissues.

This is called heterotopic ossification, meaning bone formation outside the normal skeleton. Over time, extra bone can bridge joints, restrict movement, and become permanent.

FOP often causes painful swelling episodes called flare-ups. Some lead to new bone, although bone can also form without an obvious flare-up. Triggers may include minor injuries, falls, injections into muscle, dental procedures, or surgery.

Most people with FOP are born with short, bent, or inward-turning big toes. Recognising this feature early can prevent biopsies, unnecessary surgery, and other tissue injuries that may

worsen the condition.

The tongue, diaphragm, heart muscle, smooth muscles of internal organs, and muscles controlling the eyes are generally spared. However, bone around the chest can make the rib cage less flexible and restrict breathing.

Symptoms

The earliest visible sign is usually an abnormal shape of both big toes. They may be short, turned inward, or have an abnormal joint. Thumb differences may also occur.

Flare-ups often begin during childhood and may cause:

Swellings around the scalp, neck, or back may appear suddenly and can be mistaken for tumours or infections. Some settle, while others become hard as bone develops.

Bone formation commonly begins around the neck, shoulders, and upper back before affecting the arms, hips, knees, or ankles. The order and pace vary.

Other effects can include:

The abnormal bone is not cancer and does not spread like a cancerous tumour.

- Painful or tender swelling
- Warmth or redness
- A firm lump beneath the skin
- Stiffness or reduced movement
- Tiredness or mild fever
- Difficulty moving the neck or raising the arms
- Trouble walking or balancing
- Jaw stiffness affecting eating and dental care
- Hearing loss
- Pressure sores from limited movement
- Shortness of breath as the chest becomes less flexible

Causes and risk factors

FOP is caused by a harmful change in the ACVR1 gene, also called ALK2 . This gene helps

control signals involved in bone growth and repair. In FOP, altered signalling can cause soft tissue to form bone after inflammation or injury, or without a clear trigger.

Most affected people are the first in their family to have FOP. The gene change usually occurred by chance in the egg, sperm, or early embryo. It was not caused by anything a parent did during pregnancy.

Less often, FOP is inherited from an affected parent in an autosomal dominant pattern. A person with FOP has a 1 in 2 chance of passing the altered gene to each child.

Events that may trigger flare-ups include:

Not every injury causes a flare-up, and some flare-ups have no identifiable trigger.

- Falls, bumps, muscle strain, or other injury
- Injections into a muscle
- Surgery or biopsy
- Dental procedures that stretch or injure the jaw
- Viral illnesses
- Prolonged pressure on soft tissues

Diagnosis

FOP is usually suspected when malformed big toes occur with soft-tissue swellings or abnormal bone formation. Assessment may include a medical history, physical examination, and imaging.

A blood or saliva test can identify a disease-causing ACVR1 change and confirm the diagnosis. Genetic testing is particularly useful when the signs are not typical.

X-rays or CT scans may show mature extra bone. MRI or ultrasound can show early soft-tissue changes, but cannot confirm FOP by themselves.

Biopsy is usually avoided when FOP is suspected because cutting into a swelling may provoke rapid bone formation. Removing abnormal bone for diagnosis is unnecessary when clinical findings and genetic testing can establish the condition.

Because FOP is rare, swellings may initially be mistaken for cancer or another disorder. Assessment by a specialist familiar with FOP can reduce the risk of harmful procedures.

Treatment and management

There is no cure for FOP, and existing abnormal bone cannot generally be removed safely. Care aims to reduce new bone formation when possible, manage symptoms, preserve independence, and prevent avoidable injuries.

Palovarotene is a targeted medicine approved in some countries for certain adults and children with FOP. It may reduce new heterotopic bone but does not remove existing bone. Eligibility depends on age, growth, pregnancy potential, and local approval.

Side effects include dry skin and lips. In growing children, palovarotene may cause early closure of growth plates and affect growth. It can severely harm an unborn baby, so specialist monitoring and strict pregnancy precautions are required where relevant.

For some major flare-ups, a specialist may recommend a short course of corticosteroids like prednisone or hydrocortisone , particularly after significant injury or when a major joint is affected. Anti-inflammatory medicines or other pain relief may also be used. These treatments do not reliably prevent every flare-up from forming bone.

Supportive care may include:

Forceful movement, manipulation, or stretching can injure tissue and trigger bone formation. Activity should remain within a comfortable range.

Surgery to remove extra bone is generally avoided because it often grows back, sometimes more extensively. If surgery or anaesthesia is unavoidable, it requires detailed specialist planning. Restricted jaw and neck movement can make airway management difficult.

- Physiotherapy focused on safe function rather than forceful stretching
- Occupational therapy, mobility equipment, and home adaptations
- Pressure-relieving seating, bedding, and skin care
- Nutrition support if jaw movement is limited
- Hearing assessment and hearing aids
- Respiratory monitoring and prompt treatment of chest infections
- Carefully planned dental care and anaesthesia

Complications

Progressive bone formation can lock joints in fixed positions, affecting washing, dressing, eating, writing, walking, and transfers. Some people eventually need wheelchairs or substantial daily support.

Jaw restriction can make eating and dental care difficult, leading to poor nutrition or

unintended weight loss. Limited movement can also contribute to pressure injuries, falls, fractures, chronic pain, nerve compression, reduced bone strength, anxiety, or social isolation.

Bone around the chest wall may prevent normal expansion. This can cause thoracic insufficiency syndrome, in which the chest cannot support breathing effectively. Respiratory infections may then become more serious.

Anaesthesia carries added risks because of limited mouth opening, a fixed neck, and restricted breathing. Planned and emergency procedures must account for these issues.

Prevention

FOP cannot currently be prevented. It results from a genetic change rather than diet, activity, or parenting.

Some triggers can be reduced by preventing falls, avoiding injections into muscle when a safe alternative exists, and avoiding unnecessary biopsies or surgery. These precautions cannot prevent every flare-up.

Routine vaccination remains important, particularly because respiratory infections can be serious. However, vaccine route and timing may need specialist planning because injections into muscle can trigger local bone formation.

Preventive dental care can reduce the need for complex treatment. Procedures involving jaw stretching, local injections, sedation, or anaesthesia should be planned in advance.

People considering pregnancy may choose genetic counselling to discuss inheritance, testing, and reproductive options.

Living with it and what to expect

FOP is lifelong and usually progresses over years. Rapid changes may alternate with long periods when little new bone forms. The timing and location of future flare-ups cannot be predicted.

Movement lost because a joint is bridged by mature bone is usually permanent. Treatment and careful planning focus on preserving remaining movement and independence.

Helpful support may include powered mobility, adapted clothing, voice-controlled technology, personal assistance, and pressure-relieving seating. Children with FOP usually have typical intelligence and can take part in education. A school safety plan can reduce accidental injuries while supporting inclusion.

Regular specialist follow-up may assess breathing, hearing, nutrition, dental health, mobility, skin care, and emotional wellbeing. It also provides an opportunity to plan procedures before they become urgent.

Life expectancy can be shortened, particularly when severe chest restriction causes breathing or heart-related complications. The course varies between individuals, and advances in targeted treatment and supportive care may improve future outcomes.

When to get medical help

Contact your FOP or medical team promptly after a new flare-up, significant pain, reduced movement, fall, or injury. Early advice may affect how a major flare-up is managed.

Arrange routine follow-up for increasing stiffness, equipment problems, hearing changes, pressure areas, dental concerns, or difficulty maintaining weight.

Seek urgent medical assessment after a serious fall, head injury, fracture, or major wound. Tell the medical team that you have FOP before injections, surgery, airway procedures, or attempts to move a stiff joint.

Call emergency services for:

If possible, carry an emergency information card describing FOP, airway concerns, and potentially harmful procedures. Emergency treatment should not be delayed when breathing or life is at risk.

- Severe or rapidly worsening difficulty breathing
- Blue or grey lips or skin
- Inability to swallow saliva or signs of choking
- New confusion, fainting, or marked drowsiness
- A severe allergic reaction after medicine

Common questions

Is FOP a type of cancer?

No. The swellings and extra bone are not cancer. However, early flare-ups can resemble tumours, so genetic testing is important when FOP is suspected.

Can extra bone from FOP be removed?

Surgery is generally avoided because injury can trigger new bone and removed bone may grow

back. It may occasionally be considered for a serious complication after specialist planning.

Can exercise stop my joints from becoming stiff?

No. Gentle activity within a comfortable range may support function and general health, but forceful stretching, manipulation, or exercising through pain can cause injury and should be avoided.

Does everyone with FOP need a wheelchair?

No. The timing and degree of mobility loss vary. Many people eventually use wheelchairs or other equipment to improve independence, conserve energy, and reduce falls.

Can someone with FOP have children?

Yes, but pregnancy and childbirth may carry additional risks related to breathing, restricted movement, or airway management. FOP can also be inherited. Early planning with FOP, obstetric, anaesthetic, and genetics specialists is essential.

Can vaccinations be given to someone with FOP?

Yes. Vaccination remains important, but injections into muscle may trigger flare-ups. Vaccine type, route, and timing should be planned with clinicians familiar with FOP.

Sources

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