
Fibrous dysplasia

Summary

- Fibrous dysplasia is a condition that causes abnormal growth or swelling of bone.
 - The cause seems to be a genetic mutation that alters the normal growth of the bone's connective tissue.
 - Treatment includes surgery to remove diseased section of bone.
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Fibrous dysplasia is a condition that causes abnormal growth or swelling of bone. The affected bone becomes enlarged, brittle and warped. Fibrous dysplasia can occur in any part of the skeleton but the bones of the skull and face, thigh, shin, ribs, upper arm and pelvis are most commonly affected.

This rare disorder is usually diagnosed in childhood or early adulthood and can affect one or several bones. Males and females of any race are equally affected.

Fibrous dysplasia is not a form of cancer and does not increase a person's susceptibility to cancer. In some cases, hormone problems and changes in skin colour also occur. Fibrous dysplasia is incurable but can be managed with medical treatment.

Symptoms of fibrous dysplasia

In some cases, fibrous dysplasia has no symptoms and is only diagnosed by accident during investigations for an unrelated medical problem. When symptoms occur, they can include:

- unusual gait – for example, 'waddling' or rocking from side to side when walking
- pain – caused by the expansion of bone or the pressure of the expanding bone against a nerve
- irregular bone growth
- bone deformity
- increased susceptibility to bone fractures.

Complications of fibrous dysplasia

Fibrous dysplasia can cause a range of complications:

- Rickets may develop in some cases.
- Affected pelvis and leg bones increase the risk of arthritis developing in the hip and knee joints.
- Eyesight problems or other sensory problems may develop if the bones of the skull are affected.

Types of fibrous dysplasia

The main types of fibrous dysplasia include:

- **Monostotic fibrous dysplasia** – only one bone is affected. This accounts for about seven cases out of every 10. The most commonly affected site in monostotic fibrous dysplasia is the skull.
- **Polyostotic fibrous dysplasia** – two or more bones are affected.
- **McCune-Albright syndrome** – fibrous dysplasia can be associated with hormone disturbances and skin pigment changes. About one case of fibrous dysplasia in every 10 is diagnosed as McCune-Albright syndrome.

McCune-Albright syndrome

Some people with fibrous dysplasia have hormone problems and changes in skin colour. This is known as McCune-Albright syndrome. Generally, people who have fibrous dysplasia in more than one bone are at increased risk of developing McCune-Albright syndrome.

Symptoms of McCune-Albright syndrome may include:

- onset of puberty before 10 years of age
- overactive pituitary gland, which could lead to abnormal height
- overactive thyroid gland (hyperthyroidism)
- darkened spots on the skin (café-au-lait spots).

Causes of fibrous dysplasia

Fibrous dysplasia is caused by a mutation in the gene called GNAS1. The mutation causes the GNAS1 gene to become more active than normal. The gene mutation is not inherited from the parents, nor is it present at the time of conception. It actually arises in an early cell division in the developing embryo.

As a result, the mutation is present in only some cells of the person's body, and the effects may be confined to just one location. The same underlying mechanism is responsible for McCune-Albright syndrome, but in McCune-Albright syndrome, the effects are more widespread.

Diagnosis of fibrous dysplasia

Sometimes, the symptoms of fibrous dysplasia mimic the symptoms of other conditions. For example, fibrous dysplasia of the vertebrae (backbones) may be misdiagnosed as idiopathic scoliosis.

Fibrous dysplasia is diagnosed using a number of tests, including:

- medical history
- physical examination
- blood tests
- x-ray examinations
- CT scan
- bone biopsy (a small sample of bone is collected for examination in a laboratory).

Treatment of fibrous dysplasia

Treatment may include:

- treatment to reduce the risk of complications such as rickets or fractures
- medication to strengthen bones (such as drugs commonly used in the treatment of osteoporosis)
- medication to treat pain, hormone imbalances and other problems
- pain management therapy
- physiotherapy to improve joint mobility
- surgery to correct bone deformities.

Surgery options for fibrous dysplasia

Surgery is recommended in the case of painful or fractured bones, or when joints can no longer move freely. Fibrous dysplasia that causes complications such as pressure against the brain, spinal cord or nerves is also treated with surgery.

Usually, the section of diseased bone is removed. The remaining bone is strengthened with grafts of healthy bone tissue taken from other unaffected areas of the skeleton. In some cases, pins, rods and other permanent devices may be inserted to further bolster the bone.

Surgery may successfully treat the condition if only one bone is affected. If several bones are affected, it may not be possible to remove every section of diseased tissue.

Treatment team

Treatment depends on the person's age, general health and severity of the condition. Specialist doctors may include an orthopaedic surgeon to treat bone problems, a plastic surgeon to treat facial deformities, a

neurosurgeon to treat complications of the central nervous system (brain and spinal cord) and an endocrinologist to treat hormone imbalances in the case of McCune-Albright syndrome.

Exercise is important

Regular weight-bearing exercise helps to strengthen bone, increase joint mobility and maintain a healthy weight. It is important that the exercise program is performed under the supervision of a doctor, because a person with fibrous dysplasia is at increased risk of bone fractures.

Where to get help

- Your **GP (doctor)**
- **Orthopaedic surgeon**
- **Plastic surgeon**
- Neurosurgeon
- **Endocrinologist**

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