Overview of Fibrous Dysplasia

Fibrous dysplasia happens when abnormal fibrous (scar-like) tissue replaces healthy bone. The fibrous tissue weakens the bone over time, which can lead to fractures (breaks) and misshapen bones.

Some people with fibrous dysplasia have no symptoms, or only a few symptoms, usually in one bone (monostotic). Other people may have multiple affected bones (polyostotic) and experience more symptoms. The disease may occur alone or as part of a condition known as McCune-Albright syndrome, which affects the bone as well as the skin and endocrine (hormone-producing) tissues.

Unfortunately, there is no cure for fibrous dysplasia; however, treatments may help to relieve pain, and supportive measures such as physical therapy may help strengthen muscle and improve range of motion.

What Happens in Fibrous Dysplasia?

Fibrous dysplasia happens when a gene mutates (changes) while the baby is developing in the womb. The changes in the gene cause bone-forming cells to fail to mature. Instead, they produce abnormal fibrous tissue in certain bones. Because the gene change happens while the baby is developing, only specific bones will have the disease. This means fibrous dysplasia does not spread from one bone to another.

The disease can affect any bone in the body; however, for some people, it occurs in the bones on one side of the body. The most common bones affected by fibrous dysplasia include:

- Skull and facial bones.
- Femur (upper leg) and tibia and fibula (lower leg).
- Humerus (upper arm bone).
- Pelvis.
- Ribs.

Who Gets Fibrous Dysplasia?

Fibrous dysplasia is not common, but anyone can develop the disease. It is usually diagnosed in children and young adults; however, it was probably present but not diagnosed at birth. Race, sex, environmental exposures, and geographic location do not influence who develops the disease.

Symptoms of Fibrous Dysplasia

People with a milder form of the disease may not have any symptoms and do not learn they have fibrous dysplasia until they have an x-ray for another reason. Other people may have a more severe form of the disease and develop symptoms in early childhood. The most common symptoms of fibrous dysplasia include:

- Bone pain, which may happen because of fractures or fibrous tissue changes in the bones.
- Misshapen bones or bowing of bones. This is most common in the femur (thigh bone) and is called a coxa vara (shepherd's crook).
- Fractures due to weak bone structure.

Other symptoms may develop depending on which bones are affected. Changes in the:

- Leg bones may cause the bones to shorten, bow, or change in length, leading to a limp or changes in mobility.
- Facial bones and sinuses can cause long-term sinus congestion.
- Spine can lead to scoliosis.
- Skull and facial bones around the eyes and ears may rarely lead to vision and hearing loss.

In very rare cases, some people may develop a malignant form of bone cancer.

Cause of Fibrous Dysplasia

Fibrous dysplasia happens when a gene mutates (changes) after conception, early in the pregnancy. There is nothing that the mother can do to prevent this from happening. Unfortunately, researchers do not know what causes the gene to mutate; however, they have identified the gene and continue to study why fibrous dysplasia develops. Because the disease develops from a mutated gene, children do not inherit the gene from their parents and will not pass the disease to their children.

Diagnosis of Fibrous Dysplasia

Depending on the location and severity of symptoms, your doctor may order one of the following

tests:

- X-rays. This is the most common test that doctors use to diagnose fibrous dysplasia. An x-ray can evaluate the bone structure for the disease and diagnose fractures and misshapen bones.
- Magnetic resonance imaging (MRI) or computed tomography (CT). These tests provide detailed images that are analyzed by a computer and are helpful in evaluating the skull and facial bones for the disease.
- Bone scan. This test evaluates the entire skeleton, helping doctors understand the amount of bone in the body affected by the disease.
- Bone biopsy. During this test, a doctor takes a small amount of bone tissue from an area affected by the disease to examine under a microscope.

Some children may need additional testing to determine if fibrous dysplasia is part of another syndrome or disorder. Genetic testing is usually performed on a case-by-case basis.

Treatment of Fibrous Dysplasia

There is no cure for fibrous dysplasia. The goals for treatment may include:

- Treating and preventing fractures.
- Correcting misshapen bones when the bowing is severe.
- Managing pain.

If you or your child do not have any symptoms and are not at risk for a facture, your doctor may recommend monitoring the condition. If symptoms exist, treatments may include:

- Physical therapy to help strengthen muscle and improve range of motion.
- Cast, splint, or brace to immobilize fractures or improve mobility.
- Surgery to prevent and repair fractures, treat scoliosis, and repair misshapen bone.

Although there are no medicines approved by the U.S. Food and Drug Administration to treat fibrous dysplasia, your doctor may recommend a therapy approved for a related condition. Your doctor may prescribe:

- Pain medicines to treat pain caused by broken bones and chronic bone pain.
- Medicines to treat the hormone problems some patients with fibrous dysplasia may have.

Who Treats Fibrous Dysplasia?

You or your child may see different health care professionals, depending on the location of the disease and the severity of the symptoms. Most people work with a team of doctors and medical

professionals, which may include:

- Orthopaedists, who treat and perform surgery for bone and joint diseases.
- Dental providers such as dentists and oral-maxillofacial surgeons, who provide dental care and treat problems of the mouth and jaw.
- Endocrinologists, who treat bone problems and problems related to the glands and hormones.
- Mental health providers, who provide counseling and treat mental health disorders.
- Occupational therapists, who teach how to safely perform activities of daily living.
- Ophthalmologists, who specialize in treating disorders and diseases of the eye.
- Otolaryngologists, who treat ear, nose, and throat disorders.
- Physiatrists, who specialize in physical and rehabilitation medicine.
- Physical therapists, who teach ways to build muscle strength, recover from broken bones, and prevent broken bones
- Primary care physicians, who diagnose and treat adults and children.

When possible, try to work with health care professionals familiar with treating fibrous dysplasia.

Living With Fibrous Dysplasia

Living with fibrous dysplasia is different for each person. Some people have few or no symptoms, while others have many symptoms that affect their ability to perform daily activities. The following tips may help.

- See your health care providers on a regular basis to help keep bones as healthy as possible.
- Talk to your doctor or physical therapist about which types of exercises are best.
- Ask your doctor about taking calcium, vitamin D, and phosphorus supplements to support general bone health.
- Ask family and friends for help when you need it.
- Reach out to online and community support groups.

Research Progress Related to Fibrous Dysplasia

Researchers are studying fibrous dysplasia to better understand how to manage the disease and its symptoms. Research activities include:

- Insights into the genetics and biology of the disease to help build better treatments.
- A tool that measures how fibrous dysplasia affects quality of life and how health care providers can tailor treatment plans to improve quality of life.
- Evaluation of medications to treat the disease.

Related Resources

U.S. Food and Drug Administration
Toll free: 888-INFO-FDA (888-463-6332)
Website: https://www.fda.gov

Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics
Website: https://www.cdc.gov/nchs

Genetic and Rare Diseases Information Center
Website: https://rarediseases.info.nih.gov/diseases/6444/fibrous-dysplasia

National Institute of Dental and Craniofacial Research
Website: https://www.nidcr.nih.gov

American Academy of Orthopaedic Surgeons
Website: https://www.aaos.org

Fibrous Dysplasia Foundation
Website: https://www.fibrousdysplasia.org

The MAGIC Foundation
Website: https://www.magicfoundation.org

Rare Bone Disease Alliance
Website: http://rbdalliance.org

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- Asian Language Health Information
- Spanish Language Health Information