Fibrous Dysplasia Overview

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What Is Fibrous Dysplasia?

Fibrous dysplasia (FD) is a skeletal disorder in which bone-forming cells fail to mature and produce too much fibrous, or connective, tissue. Areas of healthy bone are replaced with this fibrous tissue. The replacement of normal bone in fibrous dysplasia can lead to pain, misshapen bones, and fracture, especially when it occurs in the long bones (arms and legs). When it occurs in the skull, there can also be a replacement of the normal bone with fibrous tissue, resulting in changes in the shape of the face or skull, pain, and, in rare circumstances, hearing or vision loss.

Some people with fibrous dysplasia have only one bone involved (monostotic), whereas other people have more than one bone involved (polyostotic). The disease may occur alone, or as part of a condition known as the McCune-Albright syndrome. McCune-Albright syndrome is characterized by fibrous dysplasia and other symptoms such as patches of pigmented skin (light brown or "café-au-lait" spots) and endocrine problems such as early puberty (precocious puberty), hyperthyroidism (excess thyroid hormone), excess growth hormone (gigantism or acromegaly), excess cortisol (Cushing's syndrome), and other rare conditions.

Fibrous dysplasia can affect any bone in the body. The most common sites are the bones in the skull and face, femur (thighbone), tibia (shinbone), humerus (upper arm), pelvis, and ribs. Although many bones can be affected at once—and affected bones are often found on one side of the body—the disease does not "spread" from one bone to another; that is, the pattern of which bones are involved is established very early in life and does not change with age.

Who Is Affected?

Fibrous dysplasia is an uncommon disorder. It is usually diagnosed in children and young adults, and is present throughout life. The likelihood of getting the disease does not appear to be influenced by gender, race, ethnic background, geographic location, or by any environmental exposures.

What Is the Cause?

Fibrous dysplasia is caused by a defective gene in the cells that form bone and other affected tissues. The defect occurs at some point after conception, most

likely early in fetal development. This means that the disorder is not inherited from an affected person's parents, nor can an affected person with the condition pass it on to his or her children.

What Are the Symptoms?

The most common symptoms of the disorder are painful, misshapen, and/or broken bones (fractures). Fractures are more common between the ages of 6 and 10, but often persist into adulthood. The problems a person experiences depend on which bones are affected. For example, the legs can be of different lengths, leading to a limp and the need for a shoe lift. The bones of the sinuses can be affected, leading to chronic sinus congestion. Only very rarely do serious problems such as vision loss or cancer occur.

How Is the Disorder Diagnosed?

The bones in people with fibrous dysplasia have a characteristic appearance on x rays, which is usually sufficient to make the diagnosis. Other imaging tests, such as magnetic resonance imaging (MRI) or computed tomography (CT) may also be indicated. In some cases, a doctor may need to obtain a small bone specimen (a biopsy) to confirm the diagnosis. The usefulness of gene testing is not clear. Since the mutated gene is only present in fibrous dysplasia tissues, it is best to test only the DNA from affected tissue, but even then, doctors do not know with certainty how useful such a test is.

How Is Fibrous Dysplasia Treated?

There is no cure for fibrous dysplasia. Like most medical conditions, one treats the symptoms or problems as they arise. Fractures often require surgery, but can sometimes be treated with just a cast. Surgeries are recommended if a fracture is likely to occur, or in an effort to correct the shape of the bone. Surgery may also be indicated to relieve bone pain. Medications known as bisphosphonates—approved by the U.S. Food and Drug Administration for the treatment of other bone diseases—have been shown to reduce pain associated with the disease. Bone-healthy strategies such as physical activity (with physician approval), and adequate calcium, phosphorus, and vitamin D intake are also important.

What Research Is Being Done?

Scientists at the National Institutes of Health (NIH) are studying the natural history of fibrous dysplasia to better understand the course of the disease. Their research has provided insights into managing the disease and its consequences. NIH scientists have also developed a tool that measures the impact of fibrous dysplasia on the quality of life of people with the disorder. Additionally, researchers are evaluating the impact of bisphosphonates and other medications in patients with the disease. Studies continue to explore the genetic and molecular basis of the disease with the hope of one day developing better treatments.

Resources

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

Genetic and Rare Disease Information Center
Website: www.rarediseases.info.nih.gov/GARD

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

Fibrous Dysplasia Foundation
Website: www.fibrousdysplasia.org

The MAGIC Foundation
Website: www.magicfoundation.org

Rare Bone Disease Patient Network
Website: www.rarebonedisease.org

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For Your Information

This publication contains information about medications used to treat the health condition discussed here. When this publication sheet was developed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact the Food and Drug Administration toll free at 888–INFO–FDA (463–6332) or visit its website at www.fda.gov. For additional information on specific medications, visit Drugs@FDA at www.accessdata.fda.gov/scripts/cder/drugsatfda. Drugs@FDA is a searchable catalog of FDA-approved drug products.

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