



## Osteopetrosis Overview

### National Institutes of Health Osteoporosis and Related Bone Diseases National Resource Center

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### What Is Osteopetrosis?

Osteopetrosis is a disorder in which an imbalance in the formation and breakdown of bone causes bones to be overly dense, yet weak and prone to break easily. It is a congenital condition, which means people are born with it.

Excessive bone density can interfere with vital tissues and structures, causing serious problems throughout the body. Compression of nerves in the skull, for example, can lead to problems such as vision loss, hearing loss, or paralysis of facial muscles. It can also cause crowding of the bone marrow, which is the tissue inside the bones that produces blood cells and platelets. This can lead to low levels of cells needed to fight infection, carry oxygen to the body's cells, or control bleeding. In the most severe cases, these problems can be life-threatening. The specific complications and their severity depend largely on the type of osteopetrosis.

### What Are the Different Forms?

There are two major types of osteopetrosis: malignant infantile and adult. The malignant infantile form is evident at or shortly after birth and can greatly reduce life expectancy. Despite its name, it is not related to cancer. The formal name for this is autosomal recessive osteopetrosis.

The adult form is milder and may not be diagnosed until adolescence or adulthood. The formal name for this is autosomal dominant osteopetrosis.

Because some cases of osteopetrosis do not fit clearly into these two categories, some scientists recognize a third type called intermediate osteopetrosis. Found in children younger than age 10, this form is more severe than the adult form, but less severe than the malignant infantile form. The vast majority of the patients falling into this category have a severe form of autosomal dominant osteopetrosis (the "adult" disease).

### What Is the Cause?

Osteopetrosis is caused by defects in one or more genes involved in the formation, development, and function of cells called osteoclasts. These cells break down bone tissue during bone remodeling. Remodeling is a normal process in which old bone is removed and new bone is created to replace it.

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The malignant infantile form of osteopetrosis occurs when a child inherits a copy of the defective gene from both parents.

Only one copy of the defective gene is necessary for the adult form. This can be passed from one parent. However, in many cases, the abnormal gene does not come from either parent, but may be the result of a new gene mutation that happens at conception.

## What Are the Symptoms?

The severity of symptoms differs for the three forms of osteopetrosis. The most common symptoms are bone fractures, low blood cell levels, impaired vision and hearing, and dental problems related to infection.

## How Is Osteopetrosis Diagnosed?

The bones in people with osteopetrosis appear unusually dense and chalky white on x rays. A bone biopsy is generally not recommended.

Doctors may use other tests to diagnose and gain additional information about specific problems related to osteopetrosis. These may include hearing and vision tests, blood tests, computerized axial tomography (CAT) scans, and magnetic resonance imaging (MRI).

## How Is Osteopetrosis Treated?

Treatment for osteopetrosis depends, in part, on the form. For children who are severely affected, a bone marrow transplant may be performed. In this procedure, abnormal osteoclasts are replaced with normal ones. This can completely stop bone and bone marrow abnormalities. Unfortunately, it cannot reverse damage that has already occurred. Furthermore, finding an appropriate bone marrow donor can be difficult, and the procedure itself is very risky.

Other treatments for children or adults include:

- Interferon gamma-1b, which is an injected drug designed to delay disease progression and the only therapy specifically approved by the U.S. Food and Drug Administration (FDA) for osteopetrosis.
- Calcitriol, which is the active form of vitamin D. It stimulates osteoclasts and can help reduce bone density.

- Prednisone, a hormone similar to cortisone produced by the body that may be taken in the short term to improve blood cell and platelet counts.
- Physical and occupational therapy to help children develop motor and other skills.
- A balanced diet to support normal growth and development.
- Orthopaedic care for fractures.
- Monitoring of the eye, ear, nose, and throat
- Good dental care to decrease the chance of dental infections.

## What Types of Doctors Diagnose and Treat Osteopetrosis?

Several different types of doctors may be involved in the diagnosis and treatment of osteopetrosis. Often patients see a team of doctors who work together to provide the best treatment. These doctors may include:

- **Hematologists**, who are blood disease specialists.
- **Endocrinologists**, who specialize in hormonal and metabolic disorders.
- **Orthopaedists**, who are doctors who specialize in the treatment of bone fractures and musculoskeletal disorders.
- **Ophthalmologists**, who specialize in eye care.
- **Otolaryngologists**, who are ear, nose, and throat (ENT) specialists.

## What Research Is Being Done?

Some researchers supported by the National Institutes of Health (NIH) are examining the genetic and molecular mechanisms of abnormal bone formation and breakdown. Their goal is to find new targets for therapy. Others are studying the surgical management of complications. Elsewhere, scientists are looking at modifications to bone marrow transplantation that would make the procedure more readily accessible to children shortly after diagnosis, with the hope of stopping the disease and preventing its complications.

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## Resources

For more information on osteopetrosis, contact the:

**NIH Osteoporosis and Related Bone Diseases  
National Resource Center**

Website: [www.bones.nih.gov](http://www.bones.nih.gov)

**National Institute of Arthritis and Musculoskeletal  
and Skin Diseases (NIAMS)**

Website: [www.niams.nih.gov](http://www.niams.nih.gov)

**Genetic and Rare Diseases Information Center**

Website: [www.rarediseases.info.nih.gov/GARD](http://www.rarediseases.info.nih.gov/GARD)

**Genetics Home Reference**

Website: [www.ghr.nlm.nih.gov/  
condition/osteopetrosis](http://www.ghr.nlm.nih.gov/condition/osteopetrosis)

**American Academy of Orthopaedic Surgeons**

Website: [www.aaos.org](http://www.aaos.org)

**Orphanet: The portal for rare diseases and  
orphan drugs**

Website: [www.orpha.net](http://www.orpha.net)

**St. Jude Children's Research Hospital**

Website: [www.stjude.org](http://www.stjude.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](http://www.fda.gov). For additional information on specific medications, visit Drugs@FDA at [www.accessdata.fda.gov/scripts/cder/drugsatfda](http://www.accessdata.fda.gov/scripts/cder/drugsatfda). Drugs@FDA is a searchable catalog of FDA-approved drug products.

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