



Points To Remember About Osteopetrosis

- Osteopetrosis is a rare disorder that causes bones to grow abnormally and become too dense. When this happens, bones can break easily.
- The disorder may be mild to severe, and symptoms may show up early after birth or later in adulthood.
- Living with osteopetrosis is different for each person. Some people have few or no symptoms, while others have many symptoms that affect how they live their lives.
- Doctors treat osteopetrosis with medications; physical and occupational therapy; bone care such as casts, splints, or surgery; and bone marrow transplant in certain cases.
- Other things you can do to help you live with osteopetrosis include seeing your health care providers regularly; following the doctor's recommendations about exercise; eating a healthy well-balanced diet; and asking loved ones for help when you need it.

What is osteopetrosis?

Osteopetrosis is a rare disorder that causes bones to grow abnormally and become too dense. When this happens, bones can break easily. In addition, bones may be misshapen and large, causing other problems in the body, such as problems with:

- Seeing and hearing.
- Fighting infection.
- Controlling bleeding.

Osteopetrosis is a genetic disease that a child inherits from their parents. The disorder may be mild to severe, and symptoms may show up early after birth or later in adulthood.

Who gets osteopetrosis?

People with a family history of the disease are at greater risk of getting it through an abnormal gene that is passed on from one or both parents.

What are the types of osteopetrosis?

The types of osteopetrosis are based on:

- How the disease is inherited.
- Age symptoms develop.
- How bad the disease is.

Some types show up after birth and other types develop in adulthood.

What are the symptoms of osteopetrosis?

Symptoms can vary depending on the type of osteopetrosis. They include:

- Broken bones from weak, dense bones.
- Nasal congestion from narrowing sinus cavities.
- Vision and hearing changes from enlarged bones pressing on nerves.
- Dental problems due to bone changes.
- Low blood cell levels.
- Infections in the bone.
- Chronic bone pain.

What causes osteopetrosis?

Osteopetrosis is a genetic disease that, in most cases, a child inherits from one or both parents. People with osteopetrosis have a gene that causes the body to make too few or abnormal cells that break down old bone. This leads to dense and weak bones.

Is there a test for osteopetrosis?

Doctors diagnose osteopetrosis by:

- Taking a family and medical history.
- Performing an exam.
- Ordering laboratory tests and imaging studies, such as:
 - X-rays.
 - Blood tests.
 - Genetic testing.
 - Other imaging tests.

How is osteopetrosis treated?

Doctors treat osteopetrosis based on the type, symptoms, and other problems you are experiencing from the disease. Treatments can include:

- Medications to delay the progress of the disease.
- Medications to help improve some blood cell levels.
- Physical and occupational therapy to help children develop motor and other skills.
- Bone care such as casts, splints, or surgery to correct broken and misshapen bones.
- Bone marrow transplant in certain cases.

Regular visits with the doctor are important to:

- Check symptoms.
- Learn about test results.
- Discuss treatment.

Who treats osteopetrosis?

Several different types of doctors diagnose and treat osteopetrosis. Often, people work with a team of doctors to provide the best treatment. These doctors may include:

- Endocrinologists, who treat hormonal and metabolic disorders.
- Dentists, who can help prevent dental problems and treat issues, including orthodontists and oral-maxillofacial surgeons.
- Geneticists, who study genes, especially how they are inherited and mutate.
- Hematologists, who specialize in blood disorders.
- Neurologists, who treat disorders and diseases of the spine, brain, and nerves.
- Neurosurgeons, who perform surgery for disorders and diseases of spine, brain, and nerves.
- Ophthalmologists, who treat disorders and diseases of the eye.
- Orthopaedists, who treat and perform surgery for bone and joint diseases.
- Otolaryngologists, who treat ear, nose, and throat disorders.

Living with osteopetrosis

Living with osteopetrosis is different for each person. Some people have few or no symptoms, while others have many symptoms that affect how they live their lives. The following tips may help.

- See your health care providers on a regular basis.
- Talk to your doctor or physical therapist about which types of exercises are best for you.
- Ask your doctor about eating a well-balanced diet.

- Ask family and friends for help when you need it.
- Reach out to online and community support groups.

For more info

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