

Osteopetrosis (Marble Bone Disease)

Osteopetrosis, also known as marble bone disease, is an inherited disorder of bone metabolism that is caused by functional osteoclast deficiency. Lack of bone resorption produces overly thick but fragile bones in this disease. Although there are several subtypes of osteopetrosis, this Picmonic summarizes the shared characteristics of each.



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Pathophysiology

Defective Osteoclast Resorption

Broken Ostrich-claws

These bone cells are responsible for breaking down bone. When they fail, bone formation by osteoblasts is unopposed, and thick, dense bones are formed.

Carbonic Anhydrase II Mutation

Carbon-fiber Hydra in (2) Tutu

This enzyme is present inside osteoclasts and if a patient has a mutation in the CA2 gene, they will be deficient of the enzyme and bone resorption will fail to occur. A mutation in the CLCN7 gene will cause a hydrogen-chloride channel dysfunction with a similar outcome.

Lack of Acidic Environment

No acid from Acidic-lemon

Osteoclasts normally utilize carbonic anhydrase to make a local acidic environment that is necessary for the breakdown of bone matrix within a resorption pit. Enzyme deficiency halts this process.

Symptoms

Fractures

Fracture

Although the unopposed osteoblasts produce thick and heavy bone, it is fragile and prone to breakage just like a piece of marble. Therefore, these patients have an increased incidence of fractures.

Nerve Palsies

Nerve Pause

As bones thicken in the skull, they compress any surrounding structures, including nerves. This may cause a multitude of cranial nerve palsies and manifest with symptoms of paralysis, paresthesias, and hearing and vision impairment.

Diagnosis

Myelophthistic Anemia

Molotov Anemone

As bone mineralization continues uninhibited, it replaces bone marrow. Whenever bone marrow is displaced by another tissue, it is called a myelophthistic process. Lack of functioning bone marrow results in anemia, thrombocytopenia, and leukopenia.

Erlenmeyer Flask Bones

Erlenmeyer Flask Bone

Radiographs often depict a distal flaring of long bones on either end, producing a plain film image that resembles an Erlenmeyer flask.

Normal ALP and PTH

[Normal ALP](#) [Pine-skier](#) and [Para-thigh-droid](#)

As a way to distinguish osteopetrosis from Paget's, alkaline phosphatase will be normal in this disease. Parathyroid hormone is not involved in pathology and remains unaffected.

Normal Calcium and Phosphorus

[Normal Calcium](#) [cow](#) and [Phosphorus-P](#)

On lab evaluation, osteopetrosis patients have normal calcium and phosphate levels. Only in late and severe presentations will a decreased calcium value be identified.

Bone-in-bone Appearance

[Bone Inside Bone](#)

Overly thick and dense bones produce a characteristic x-ray image in which it appears that a bone has another bone inside of it.

Treatment

Bone Marrow Transplant

[Bone Train-plant](#)

Since a patient lacks functional osteoclasts, the only available treatment is to perform bone marrow transplant and allow for monocyte evolution into new osteoclasts.