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

Myelophthisic Anemia
Molotov Anemone
As bone mineralization continues uninhibited, it replaces bone marrow. Whenever bone marrow is displaced by another tissue, it is called a myelophthisic 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 ALPine-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.