Metabolic osteopathy

This article has been translated from WikiSkripta; ready for the **editor's review**.

[1]Metabolic osteopathies are diseases caused by disturbances in the balance between bone formation and resorption and disturbances in bone mineralization → osteopenia and decreased bone mass (osteoporosis, osteomalacia) or bone sclerotization and increased bone mass (Paget's disease , osteopetrosis). These osteopathies are caused by bone cell dysfunction, genetic abnormalities (defects in the synthesis of collagen I etc.), increased expression of bone morphogenetic proteins, kidney function disorders, endocrinopathies, tumor secretion of substances affecting bone metabolism[2]

Osteoporosis

Osteoporosis

Rickets

Rachitis

Osteomalacia

Ostoemalacia

Definition of Disease

Osteomalacia/ Rickets is a metabolic bone disease characterized by inadequate bone mineralization. The remodeling cycle proceeds normally through osteoid formation, but mineral calcification fails to occur. Rickets is the term used to describe the condition prior to the closing of the physis. Osteomalacia is used after physis closure.

Pathophysiology

Crystallization of minerals in osteoid requires adequate concentrations of ionized calcium and phosphate. Vitamin D influences these levels after its dihydroxylation into calcitriol (hepatic position 25 and renal position 1). When concentrations are too low crystallization does not proceed normally. Vitamin D disrupts mineralization because it normally regulates and enhances the absorbtion of calcium in the intestine. A lack of vitamin D causes plasma calcium concentrations to fall. Low plasma calcium levels stimulate parathyroid hormone (PTH). PTH raises calcium concentration but also increases renal clearance of phosphate. When phosphate decreases below a critical level, mineralization cannot proceed normally.

Genetics

Hereditary forms of osteomalacia include:

• Hypophosphatemic vitamin D resistant rickets, an X-linked, autosomal dominant disorder,

• Hypophosphatemic rickets with hypercalciuria autosomal recessive

• Vitamin D dependant rickets caused by a mutation in the renal tubular 25-hydroxyvitamin D hydrolase

Hereditary forms of vitamin D deficiency and resistance, identified in childhood, are associated with osteomalacia in adults, but these disorders are rare.

Epidemiology

Osteomalcia is rare in the United States and Western Europe. There is a growing prevalence of vitamin D deficiency in many countries, which when severe and prolonged results in hypocalcemia, secondary hyperparathyroidism, secondary hypophosphatemia, and osteomalacia. Populations at risk include:

• The homebound elderly who have little sun exposure and insufficient dietary calcium and vitamin D

• Patients with malabsorption related to gastrointestinal bypass surgery, crohns, or celiac disease

• Vegetarian diets without vitamin D supplementation

• Women who wear traditional veils or dresses that prevent sun exposure

• Patient on long term anticonvulsant therapy i.e. phenytoin and phenobarbital

• Rifampin and glucocorticoics

• Tumor induced osteomalacia i.e. paraneoplastic syndrome of renal phosphate wasting by tumor secretion of phoshatonin

Disease described

The principle abnormality associated with Osteomalacia/ Rickets is a defect in the mineralization of the osteoid matrix. It can be definitively diagnosed by bone biopsy. The clinical syndrome associated with osteomalacia consists of pain, myopathy, and fracture.

Sign and Symptoms

• Pain, sometimes severe, in bones, particularly in the pelvis, lower back and legs. Tenderness may sometimes be felt in the shins and in other bones.

• The patient usually walks with feet rather widely separated and may appear to waddle. Deformities of the pelvis and long bones may be obvious.

• Tetany is manifested by involuntary twitching of the muscles of the face or by carpopedal spasm.

• Spontaneous fractures may be a feature. Before the deformities are clinically detectable, diagnosis may be made by X-ray examination, which will show rarefaction or decalcification of bones all over the body.

Diagnosis

• Blood and urine tests. In cases of osteomalacia caused by vitamin D deficiency or by phosphorus loss, abnormal levels of vitamin D and the minerals calcium and phosphorus are often detected.

• X-ray. Slight cracks in bones that are visible on X-rays, referred to as Looser transformation zones, are a characteristic feature of osteomalacia.

• Bone biopsy. High specificity in detecting osteomalacia, it's not often needed to make the diagnosis.

• ALP and PTH may be done to rule out renal disorders causing the problem.

Treatment

Osteomalacia is managed by treating the underlying cause. If vitamin-D deficiency is diagnosed, repletion can be accomplished with:

• Oral vitamin D, 1000 IU per day.

• Neutral phosphate salts, 500 mg four times daily.

• Long-term anticonvulsant therapy may be supplemented with 400 to 800 IU of vitamin D daily.

• Hepatobiliary disease or chronic renal failure is managed with supplemental 25(OH)D (calcifediol) and 1,25(OH)2D (calcitriol) (Lexicomp, 2014).

Links

http://www.mayoclinic.org/diseases-conditions/osteomalacia/basics/definition/con-20029393

http://my.clevelandclinic.org/orthopaedics-rheumatology/diseases-conditions/hic-osteomalacia.aspx

Related current articles

http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1819593/

http://onlinelibrary.wiley.com/store/10.1111/j.1753-4887.2008.00100.x/asset/j.1753-4887.2008.00100.x.pdf?v=1&t=hta9wzlo&s=bd3005296b46d52d150162df20d17a80f485c5da

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Lexicomp (2014). Osteomalacia and rickets (2.0.1) [Mobile application software]

McCance, K., & Heuther, S. (2014). Pathophysiology: The biological basis for disease in adults and children (7th ed.). St. Louis, MO.: Elsevier/Mosby.

Osteodystrophia fibrosa cystica generalisata

Osteodystrophia fibrosa cystica generalisata (*morbus Recklinghausen, primary hyperparathyroidism*) is a disease belonging to the group of acquired systemic diseases of the bone system. **It must be distinguished from morbus von Recklinghausen, which is synonymous with neurofibromatosis - type 1.**

Etiopathogenesis

It mainly affects women (mainly in the 2nd decade of life). **The cause is an adenoma of the parathyroid glands** , which leads to hyperproduction of PTH , PTH releases phosphates and calcium salts from the skeleton, thereby increasing phosphaturia and increasing calciuria (it increases calcium resorption in the ascending limb of the loop of Henle, but due to high calcium values, hypercalciuria still occurs) . This results in hypophosphatemia and hypercalcemia .

At the same time, there is an increased formation of osteoid (fibrous remodeling of cancellous bone). Cystic destruction of the skeleton and generalized osteoporosis occur . Fractures/infractions with intraosseous hemorrhages occur at the site of significant weakening of the load-bearing parts of the skeleton .

Clinical picture

Fatigue with reduced physical performance, occasional **pain** in the spine and limbs. In the later stage of the disease, minor **limb deformities** , or spontaneous fractures. **Kidney damage** : nephrolithiasis to nephrocalcinosis .

Laboratory finding

- hypercalcemia
- hypercalciuria
- hypophosphatemia
- hyperphosphaturia

X-ray image

X-ray examination performed only after the occurrence of a spontaneous fracture (cystic deposits, thinning of the compacts, enlargement of the medullary cavity). A decrease in the thickness of the vertebral bodies, their expansion, and the occurrence of multiple compression fractures are common . Subperiosteal bone reduction, most commonly seen on the middle phalanges of the fingers. Structural changes in the calf are common.

In the advanced stages of the disease, there are multiple angulations and severe deformities of the load-bearing parts of the skeleton. On CT sometimes parathyroid adenoma.

Therapy

Causal treatment only **surgical** (removal of parathyroid adenoma). Hypercalcemic crises are treated with hydration and adjustment of the mineral economy.

Orthopedic therapy consists in corrective osteotomy of the resulting deformities, possibly in combination with prolongation performances .

Differential diagnosis

Fibrous dysplasia (Jaffe-Lichtenstein), cortical fibrous defect, juvenile solitary pseudocyst , myeloma . In all of these diseases (with the exception of plasmacytoma) unilocular/monomelic occurrence, but in fibrous dysplasia the bones of almost the entire skeleton are affected .

Links

Related Articles

- Metabolic osteopathy
- Osteoporosis
- Osteomalacia
- Rickets

References

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Albers-Schönberg disease

Morbus Albers-Schönberg

Links

Related Articles

- Metabolic bone disease of immaturity

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2. GALLO, George, et al. *Orthopedics for students of medical and health faculties.* 1. edition. Olomouc : Palacký University in Olomouc, 2011. ISBN 978-80-244-2486-6.