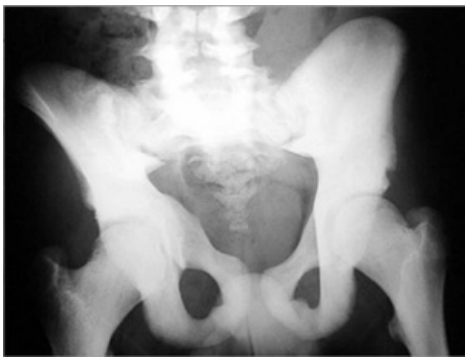
Albers-Schönberg Disease

Albers-Schönberg disease (*marbling of the bones, osteosclerosis, osteopetrosis*) is a rare hereditary disease with a malfunction of osteoclasts (disorder of bone resorption). Disturbed **balance of activity of osteoblasts and osteoclasts** → bone becomes extremely compact (**bone sclerosis)**. New bone formation normal → brittle and brittle bone, strengthening of metaphyses and diaphysis. The primary medullary cavity is filled with homogeneous irregular bone substance → **extramedullary hematopoiesis**.


Osteoporosis of pelvis (X-Ray)

- 3 forms:
  - ***Classic congenital form*** (m. Albers-Schönberg) – malignant osteopetrosis (AR hereditary).
  - ***Osteopetrosis tarda*** – benign (AD hereditary).
  - ***Osteopetrosis due to renal tubular acidosis*** (Hereditary AR).

Clinical picture

- different course and prognosis – mild forms to severe or fatal forms (e.g. early infantile form ending fatally before the age of 10),
- disorders of hematopoiesis → anemia, immunodeficiency,
- macrocephaly, tooth defects, osteosclerosis, exophthalmos (by compression of nerves in bone canals),
- pathological fractures.

X-ray image

- the basic image is an increased density of bone tissue (corticalis and spongiosis cannot be distinguished, filling of the marrow cavity of the bones),
- striated osteosclerotic zones of vertebral bodies and covering plates,
- thickening of the cortex of the long bones and the peripheral parts of the pelvis,
- radial thickening of the bony beam in the arm and leg skeleton,
- periosteal apposition to spicules, often in traction zones of the skeleton (club-shaped metaphysis).

Laboratory finding

- increased acid phosphatase (possibly also alkaline phosphatase) in the serum,
- calcium and phosphate levels normal,
- in the renal form, manifestations of acidosis.

Prenatal diagnosis

- sonographically increased bone density,
- radiological certificate from the 25th week of pregnancy.

Therapy

- causal is not,
- **non-orthopedic treatment** – treatment of anemia or pancytopenia (bone marrow transplantation, corticoids, IFN-α),
- **orthopedic treatment** – pathological fractures (mostly transverse) , bone healing is prolonged, a longer period of immobilization of fractures is necessary.

Differential diagnosis

- other sclerosing bone diseases (pycnodysostosis, progressive diaphyseal dysplasia, metaphyseal dysplasia, metal poisoning, syphilis, myelofibrosis) – they do not have severe anemia.

Links

Related articles

- Neurofibromatosis (m. von Recklinghausen)
- Osteogenesis imperfecta (osteopsatyrhosis, fragilitas ossium)
- Osteopoikilosis (osteopoicilia)

References

- SOSNA, A. – VAVŘÍK, P. – KRBEC, M.. *Základy ortopedie.* 1. edition. Triton, 2001. ISBN 80-7254-202-8.
- DUNGL, P.. *Ortopedie.* 1. edition. Grada Publishing, 2005. ISBN 80-247-0550-8.

Reference

1. SOSNA, A., P. VAVŘÍK a M. KRBEC, et al. *Základy ortopedie.* 1. vydání. Praha : Triton, 2001. ISBN 80-7254-202-8.
2. ↑ Skočit nahoru k:a b c d e f g h DUNGL, P., et al. *Ortopedie.* 1. vydání. Praha : Grada Publishing, 2005. ISBN 80-247-0550-8.