

## ADULT OSTEOPETROSIS – CASE REPORT

COSMINA DIACONU<sup>1</sup>, CRISTINEL GABRIEL DIACONU<sup>2</sup>, FLORINA LIGIA POPA<sup>3</sup>

<sup>1,2,3</sup>“Lucian Blaga” University of Sibiu

**Keywords:**

osteopetrosis,  
radiologic features,  
therapeutic approach

**Abstract:** Osteopetrosis is a sclerosing bone dysplasia. Several clinical forms, different by clinical expressions, genetic and biochemical findings, have been reported. There are two major clinical forms of the disease: the autosomal dominant, adult, benign type (incidence has been reported at 1:20.000 births) and the autosomal recessive, infantile malignant type (incidence has been reported at 1:250.000 births). We intend to present a rare case of adult osteopetrosis, we discovered accidentally having radiographs taken for poliarticular pathology.

### INTRODUCTION

Osteopetrosis, also known as marble bone disease or Albers-Schönberg disease is a disorder caused by a defective bone remodeling that leads to a generalized growth of bone mass.

In disease pathogenesis, there is incriminated an abnormality in the functioning of osteoclasts, expressed through a failure of osteoclast-mediated resorption of the skeleton. The exact mechanism underlying the osteoclast dysfunction is not known.

There have been described several clinical forms of the disease:

- *Type I benign (autosomal dominant), adult osteopetrosis* - with adult onset, more patients present few or no symptoms, but the radiological abnormalities become apparent during childhood. It affects long bones which are very brittle and prone to fracture. In addition, this type of osteopetrosis may be associated with osteoarthritis, carpal tunnel syndrome, facial palsy, deafness, impaired vision or hearing, osteomyelitis of the mandible and psychomotor developmental delay.
- *Type II malignant (autosomal recessive), infantile osteopetrosis* – diagnosed immediately or shortly after birth. If it is untreated is fatal during infancy or early childhood. In the spinal canal, due to excessive bone development occurs marrow insufficiency, that leads to hepatosplenomegaly and poliadenopathy.
- *Type III intermediate (autosomal recessive)* - less common, occurs during childhood, when there are some signs of malignant osteopetrosis, but the impact on survival is not well defined.
- *Type IV syndrome called osteopetrosis with renal tubular acidosis and cerebral calcifications (autosomal recessive)* occurs because carbonic anhydrase II deficiency due to inborn error of metabolism.

### CASE REPORT

Female patient, 53 years old, from the urban environment, who was admitted in the Clinical Rehabilitation Department I of Sibiu Emergency County Clinical Hospital

presenting:

- pain in cervicodorsolumbar spine that spread in the both thighs (radicular bilateral L3 syndrome),
- mechanical, polyarticular pain (knees, hips, in the small joints of the hands).

From the family history and from the personal history, it can be noted:

- her father and a son, incidentally diagnosed in adulthood, respectively in late childhood with marble bone disease.
- early menopause at 42 years old, cholecystectomy for cholelithiasis at 31 years old when, during colecistography, she is diagnosed incidentally with osteopetrosis.

The patient underwent repeated rehabilitation treatment in our ward for mechanical pain in the joints and in cervicolumbar spine.

The patient did not present any fracture until now.

Objectively, we established: moderate rachidian lumbar syndrome: lumbar flattening, decreased spinal mobility – on flexion (Schober test = 10/12 cm) and on lateral inflexion; Lasegue (straight test – raising sign) and Lasegue reversed test negative, patellar tendon reflexes decreased and Achilles tendon reflexes normal, no motor deficit signs, knees with crackles on mobilization, left knee painful, which tend to deviate in varum; right hip with diminished mobility in abduction and internal rotation, painless Heberden’s nodes.

Among the *laboratory findings*, we mention: serum calcium= 5.0 mEq/L(4.2-5.1), urinary calcium= 9.1 mEq/1-13.6 mEq/24h(5-15 mEq/l) ,serum phosphate= 4.3mg/dl (2.7-4.5), urinar phosphate=1.47g/24h (VN=0.4-1.3g/24h), serum alkaline phosphatase =110U/L (40-150 U/L), vitamin D=22.2 µg/I (optimum level>30 µg/I), blood count was normal, as blood glucose level and urine exam.

*Radiologic features:* Abdominal ultrasound does not reveal hepatosplenomegaly.

*Treatment:* According to the medical literature, there is no specific drug treatment for the adult type.

A low-calcium/high phosphate diet can be recommended to our patient.

<sup>1</sup>Corresponding author: Cosmina Diaconu, B-dul Mihai Viteazul, Bl. P 15, Nr. 19, Sibiu, România, E-mail: cosmina\_75dia@yahoo.com. Phone: +40727 887829

Article received on 08.01.2014 and accepted for publication on 07.02.2015  
ACTA MEDICA TRANSILVANICA March 2015;20(1):58-59

## CLINICAL ASPECTS

If the patient has symptoms of hypocalcemia, she may need calcium intake.

**Figure no. 1. Lumbar spine radiograph (lateral left view): Sclerotic bands on the upper and lower margins of each vertebra T8-L5; vertebrae have a „bone in bone” (endobone), „rugger jersey” or a „sandwich” configuration**



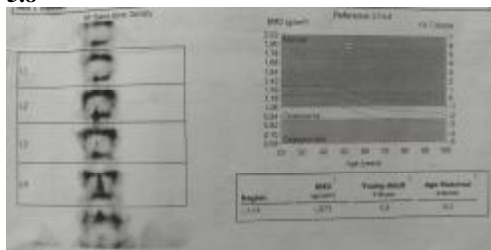
**Figure no. 2. Pelvic radiograph: Alternating sclerotic and lucent bands in the iliac wings and at the femoral heads**



**Figure no. 3. Both knee radiograph (anteroposterior view): knee osteoarthritis, Kellgren-Lawrence II at the right knee and III at the left knee; osteosclerotic areas in distal part of the both femoral bones and in the proximal epiphyses and metaphyses of both tibias, more evident in the left leg**



**Figure no. 4. Central skeletal DXA (dual-energy x-ray absorptiometry) – Bone mineral density (BMD) exam of the spine in an anterior-posterior (AP) view of L1-L4: T-score=5.8**



In infantile osteoporosis, large doses of vitamin D (calcitriol) appear to help by stimulating dormant osteoclasts to dissolve and absorb bone. The positive effects on bone remodelling are not sustained when the therapy is discontinued. Even low doses of vitamin D stimulate bone mineralization (stimulate intestinal absorption of calcium and phosphate), that is why this treatment is not useful in our case despite the fact that our patient presents a low level of vitamin D.

In adult osteopetrosis, surgical treatment may be needed, in the first place, for functional reasons (e.g., loss of function, multiple and repeated fractures - especially at femoral shaft, inferior neck of femur or posterior tibia fractures, deformity of the bones) and, in the second place for aesthetic reasons (notable facial or joint deformity). Fracture healing on osteopetrotic bones seems to occur at a normal rate, calus formation is variable. Severe degenerative joint disease (knee or hip osteoarthritis) may warrant surgical intervention as well. Life expectancy for the autosomal dominant adult subtype is normal.

Our patient presents moderate knee osteoarthritis and does not present any facial deformity. For this reasons, we consider that the best option is conservative treatment, with medication (pain-killer, chondroprotector) and periodic rehabilitation treatment.

The objectives of the rehabilitation therapy are:

1. Controlling pain and muscular contracture;
2. Preventing limitation and restoring ROM – Range of Motion in affected joints by the tonification of the abdominal and gluteal muscle, stretching the paravertebral and iliopsoas muscle;
3. Maintaining and improving muscle strength, especially at stabilizer muscle of the knees and hips, to improve normal motor function at the level of the bearing joints;
4. Reducing the risk of bone fractures, especially the limb fractures.

### CONCLUSIONS

1. The patient was diagnosed incidentally with autosomal dominant osteopetrosis due to the presence of typical radiographic findings, age onset and inheritance pattern, in spite of the fact she was asymptomatic.
2. Early menopause represents a risk factor for osteoporosis, but our patient presents, as it can see on Dual-energy X-ray absorptiometry (DEXA), an increased bone mass, due to impaired bone turnover, with abnormalities in osteoclast function.
3. The patient must be counselled on adequate lifestyle modification to prevent fractures.

### REFERENCES

1. Whyte MP. Sclerosing Bone Dysplasias. In: Favus MJ, ed. Primer on the Metabolic Bone Diseases and Disorders of Mineral Metabolism, Third Edition, Philadelphia, Lippincott-Raven Publishers. 1996;68:363-366.
2. Blank P. Chief Editor: George T Griffing. Osteopetrosis. 2014; Dec 17 [Medscape Reference ].
3. Baron R. Anatomy and Ultrastructure of Bone. In: Favus MJ, ed. Primer on the Metabolic Bone Diseases and disorders of Mineral Metabolism. 4<sup>th</sup> ed. Philadelphia, Lippincott, Williams and Wilkins. 1999:3-10.
4. Stark Z, Savarirayan R. Osteopetrosis. Orphanet J Rare Dis. Feb 20 2009;4:5 [Medline].