DEPOSITION OF HOMOGENTISIC ACID IN LONG BONES WITH DIAPHYSEAL LYTIC CHANGES IN A PATIENT WITH OCHRONOSIS

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SUMMARY - Presentation is made of a 58-year-old female patient with alkaptonuria and ochronosis. Calcification and ossification of intervertebral disks in the lumbar, thoracic and cervical spine were developed. In addition, diaphyseal lytic changes of the femur, tibia and fibula were observed. The lesions were diffuse and unequal. Diaphysis of the long bones had a ‘moth-eaten’ bone appearance. No data on such changes were found in the available literature.

Key words: Ochronosis; Ochronosis - pathology; Ochronosis - complications; Homogentisic acid

Introduction

Ochronosis is caused by inherent flaw due to the absence of the enzyme homogentisic acid oxidase. This enzyme normally aids in the oxidation of tyrosine/phenylalanine in the complete breakdown of amino acids to water and carbon dioxide. Excessive homogentisic acid in the blood and urine results in the deposition of black pigment in soft tissues and cartilage (meniscus, disk, joints, ear, nose). To our knowledge, there are no literature reports on the pigment deposition in bone marrow and bone tissue leading to lytic lesions. That is why the case presented appears to be quite interesting and intriguing.

Case Report

Our patient was a 58-year-old woman, a housewife, suffering from alkaptonuria from birth. She had two adult children. She had no had any serious illness. In 1998, she fell and sustained fracture of the left femur neck. After the operation and implantation of total hip prosthesis, she underwent rehabilitation and walked on a crutch for a long time. Radiologic examination was performed several hours after the fracture.

On presentation, blue auricles, tip of the nose, both sclerae and fingertips were observed. Thoracic kyphosis and reduced cervical and lumbar lordosis were present. Spine movements (inclination and reclinatio) were reduced. The patient had been becoming shorter, and she could reach ever lower spot on her leg with her fingers. The coastal arch ‘sat’ on the iliac crest.

Laboratory findings: ESR 30, erythrocytes 4.02, hemoglobin 111, leukocytes 9.1. Other hematology and biochemistry findings were normal. The homogentisic acid test was positive in the blood and urine. Other hematological and biochemical markers were normal. The homogentisic acid test was positive in the blood and urine. Other hematological and biochemical markers of bone turnover, and bone or synovium tissue PHD were not determined.

Radiologic finding: Beside ochronotic changes including calcification and ossification of the intervertebral disks in the lumbar, thoracic and cervical spine (Fig. 1) and ochronotic peripheral arthropathy, diaphyseal lytic changes of the femur, tibia and fibula were also recorded (Figs. 2 and 3). Osteolytic changes diffusely affected bone tissue, especially in the sponge area, wherefrom the lesions extended into the cortex causing erosions. Radiologic images showed the bone tissue appearing like a ‘moth-eaten’ bone. Os-
Discussion

There are no radiologic changes early in the course of the disease. Changes occur in the second and third decade, and most distinctive findings are calcification and ossification of multiple intervertebral disks, which result in reduced inclination and reclinination of the spine. Changes of peripheral joints usually develop secondary to the changes of the spine. Subchondral cysts are not pronounced in ochronosis, and lytic changes have not been mentioned in the available literature.

Homogentisic acid is being deposited in the joint cartilage causing its destruction. Similarly, it is deposited in the bone tissue causing lytic lesions, their severity depending on the homogentisic acid amount. Lytic changes are likely to more easily develop in an osteoporotic area.

In conclusion, we consider that the lytic changes of the diaphyseal bone were due to the effect of homogentisic acid, which is secreted in excess in patients with ochronosis.

References

Sažetak

ODLAGANJE HOMOGENTIZINSKE KISELINE U DUGE KOSTI S LITIČKIM PROMJENAMA DIJAFAZA U BOLESNICE S OHRONOZOM

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Ključne riječi: Ochronozoa; Ochronozoa, patologija; Ochronozoa, komplikacije; Homogentizinska kiselina