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CAMURATI -ENGELMAN DISEASE- A RARE CAUSE ON BONE SCINTIGRAPHY: A CASE REPORT AND REVIEW OF THE LITERATURE

Sulejmanović Maja[1], Sabina Salkić[2], Fatima Mujarić -Bousbia[1]

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Sulejmanović Maja
Sabina Salkić
Fatima Mujarić -Bousbia

ABSTRACT

Camurati-Engelmann disease (CED) or progressive diaphyseal dysplasia is a rare autosomal dominant hereditary disorder, characterized by simmetrical progressive sclerosis of long bones cortical diaphysis. This disease is caused by mutation in the TGFβ1 gene [1,2]. Considering the rarity of this disorder (to date around 300 affected individuals have been reported) [3], we would like to share Bosnian and Herzegovina patient diagnosis with CED. To the best of our knowledge, no case of CED has been reported in Bosnia and Herzegovina till date. We evaluated a 10-year-old girl had history pain in the left leg and difficult movement since age of four. During the time, neuromuscular disease was suspected, but not confirmed. Symmetrical enhanced uptake seen on technetium hydroxymethylene diphosphonate (99m Tc-HDP) bone scintigraphy at diaphyses of longitudinal bones and cortical thickening of long bones seen at radiograms. Those finding pointed to Camurati-Engelmann diagnosed misdiagnosed for the long time.
Keywords: Camurati-Engelman disease, diaphyseal dysplasia,bone scintigraphy,

INTRODUCTION

Camurati- Engelmann disease (CED) or progressive diaphyseal dyspasia is a rare autosomal dominant hereditary disorder, characterized by simmetrical progressive sclerosis of long bones cortical diaphysis. In 1920 Cockayne descrobred a case of unusual bone thickening in 9-years old child [4]. Two years leter, camurati published a report of father and son having identical changes in lower limbs. He name this disease 'symmetrical hereditary osteitis', and determined later the indentical condition in four generation of the same family [5]. In 1929 Engelmann describred similar condition in 8-years old boy and named it » osteopathica hyperostotica (sclerotisans) multiplex infantilis« [6]. This sindrome is called today Camurati-Engelmann disease (CED) or progressive diaphyseal dysplasia. It is an autosomal dominant disease, a part of the group variability of simptoms. Most of the patients experience pain in the extremities, waddling gait, easy fatigability and muscle weakness [7]. Symptoms mostly occur at younger age, although not necessarily. Some of the patients have systemic manifestation like anaemia, leucopaenia and hepatosplenomegaly. Main pathology is cortical thickening-hyperostosis, appearing on the diaphyses of the long bones. The changes are present in bilateral and symmetrical pattern; long bones of lower extremities are affected first, than long bones and pelvis have sclerotic changes. Radiologically it is recognized by the symmetrical enlargement with sclerosis of the cortex of long bones. Bone scan shows abdormal uptake of technetium hydrosy diphosphonate-(99m Tc HDP) in mentioned locations [8,9]. In most of the cases Camurati-Engelmann disease is caused by a mutation of coding region TGFβ-1 (transforming growth factor beta 1) located on chromosome 9q13 [10]. TGFβ-1 is a stimulator of osteoblastic bone formation. The result of osteoblast enhanced action is failure of bone resorption creating bilateral symmetrical cortical thickening of the diaphyses of the long bones [11]. We report a case of a 10-year-old girl with bone pain and difficult movement since the age of 3 years.

CASE REPORT

A 10-year old girl was hospitalized to the pediatric clinic for an evaluation. The curent disease started a year ago due to a swelling in the lower right leg. On physical examination the girl was swelling in the

Affiliations:
[1] Clinic for Radiology and Nuclear medicine, University Clinical Center Tuzla, Trnovac bb, 75000 Tuzla, Bosnia and Herzegovina
[2] Family Medical Services, Public Healthcare Institution,Medical Centre Tuzla, Tuzla, Bosnia and Herzegovina

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Corresponding author:
Sulejmanović Maja
Email:majasulejmanovic@yahoo.com

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distal part of the lower leg, asthenic stature, hipotrophic musculature of the lower extremities and flat feet. A detailed history taking revealed that the presence of difficult movement, pain in the left leg and she can not get rising up from the squat since the age of third. During the past years diagnostic procedures were directed to determine nature of her neuromuscular symptoms, though no neurological or muscular demege was confirmed (electroneurography, neurological examination were normal) She had elevated iron and vitamin D in the serum. Thyroid funtion test were normal. ECG and echocardiographic findings were normal. Two mounts ago she had a blunt trauma in the left tibia. Bone scintigraphy was performed to evaluate the involved bones and provide effective signs for the diagnosis. The images were obtained using a 2-head gamma camera (GE) with high resolution and low energy collimator, 256x256 matrix, After 2 hours of the intravenous administration 99mTc-technetium-99m-hydroxy-methylene diphosphonate (HDP) with a dose of 740MBq, the planar whole body image was obtained (Figure 1). The images revealed diffuse, symmetric intense uptake in the long bones of upper and lower extremities. The tracer distribution in the other parts of the body was within normal limits. The x-ray of upper extremities showed thickening diaphyseal cortical, sclerotic and medullary canal stenosis (Figure 2). These findings were consistent with the increased uptake observed on bone scintigraphy images. On the basis of two findings, we believed that the diagnosis is Camurati-Engelmann disease.

DISCUSSION

The CED has been known by the names of progressive epiphyseal dysplasia, generalized hyperostosis, congenital multiple hyperostotic disease, sclerosing dysplasia and symmetrical osteosclerosis [12] but the eponym CED is the name most widely known and accepted. The first description was made by Cockayne in 1920 [13], 2 years later Camurati suggested its hereditary nature [5] and in 1920 Engelmann reported a case characteristic of the disease [14].

The child in our report presented with the typical clinical features of leg pains and difficult movement. The Whole body bone scintigraphy detected icreased osteoblastic activity in the diaphyseal portion of the long bones and lower extremities.As increased tracer uptake can be perceived even before sclerosis becomes radiologically visible, scintigraphy is a valuable technique for diagnosing CED in an early stage of disease. The bone scintigraphy could also be used to help assess treatment effects and early diagnosis with the advantage of more sensitivity than x-ray [15-17].The child is on corticosteroids therapy. We believe that this case highlights the need to consider skeletal dysplasias in the differential diagnosis of non-specific limb pains in addition to arthropathy, neuromuscular disorders, malignancy and non-organic causes.

CONCLUSION

Bone scintigraphy is a simple, noninvasive technique that enable the study of whole body, showing the areas with the greatest involvement in patients with generalized bone disease. The indicental finding of cortical thuckening on imaging studies should be examined by an extensive bone scan to improve staging and its classification.

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