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Case Report

A case of infantile osteopetrosis: The radioclinical features with literature update☆

Tamer Ahmed EL-Sobky [a,*], Ezzat Elsobky [b], Ismaiel Sadek [c], Solaf M. Elsayed [b], Mohamed Fawzy Khattab [d]

[a] Department of Orthopaedic Surgery, Division of Paediatric Orthopaedics, Faculty of Medicine, Ain-Shams University, 38 Abbasia, Cairo, Egypt
[b] Division of Medical Genetics, Department of Paediatrics, Faculty of Medicine, Ain-Shams University, 38 Abbasia, Cairo, Egypt
[c] Department of Paediatrics, Faculty of Medicine, Ain-Shams University, 38 Abbasia, Cairo, Egypt
[d] Department of Orthopaedic Surgery, Faculty of Medicine, Ain-Shams University, 38 Abbasia, Cairo, Egypt

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ABSTRACT

Background: Osteopetrosis is a rare hereditary metabolic bone disorder characterized by generalized skeletal sclerosis caused by a defect in bone resorption and remodelling. Infantile autosomal recessive osteopetrosis is one of three subtypes of osteopetrosis and the most severe form. The correct and early diagnosis of infantile osteopetrosis is important for management of complications and for future genetic counselling. Diagnosis is largely based on clinical and radiographic evaluation, confirmed by gene testing where applicable.
Methods: Therefore, in this case study the classical clinical and radiological signs of a boy with infantile osteopetrosis will be presented with a comprehensive literature update. The differentiating signs from other causes of hereditary osteosclerosing dysplasias are discussed.
Results: This case study and review of available literature show that there tends to be a highly unique clinical and skeletal radiographic pattern of affection in infantile osteopetrosis.
Conclusion: Although tremendous advances have been made in the elucidation of the genetic defect of osteopetrosis over the past years, the role of accurate clinical and radiological assessment remains an important contributor to the diagnosis of infantile osteopetrosis.

1. Introduction

Osteopetrosis is a rare hereditary metabolic bone disorder characterized by generalized skeletal sclerosis caused by a defect in bone resorption and remodelling. The defect in bone turnover characteristically results in skeletal fragility despite increased bone mass, and it may also cause haematopoietic insufficiency. The actual incidence is unknown but it is estimated to be 1 case per 100,000–500,000 population (Stark and Savarirayan, 2009). Three distinct clinical forms of the disease—infantile, intermediate, and adult onset—are identified based on age and clinical features. Infantile autosomal recessive osteopetrosis is the more severe form that tends in the first few months of life. Hence, it is referred to as "infantile" and "malignant" compared to the autosomal dominant adult osteopetrosis (Stark and Savarirayan, 2009). The correct and early diagnosis of infantile osteopetrosis (IO) is important for management of complications and for future genetic counselling. Diagnosis is largely based on clinical and radiographic evaluation, confirmed by gene testing where applicable (Sit et al., 2015). It is therefore important to be familiar with the radiological features of IO. Therefore, in this case study the classical clinical and radiological signs of a boy with infantile osteopetrosis will be presented and a comprehensive literature update. The differentiating signs from other causes of hereditary osteosclerosing dysplasias are discussed.

2. Case report

A one and a half year old boy was brought to our outpatient clinic with complains of delayed milestones. He was first in order to a first cousin parents. Birth and family history were unremarkable.

Clinical examination revealed macrocephaly with opened anterior fontanel, frontal bossing, nystagmus of left eye and retromicrognathia, near normal stature and proportions and abnormal dentition. He had small chest cavity with ptosed liver and mildly enlarged spleen. The boy was only able to ambulate with parents' assistance.

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* Corresponding author.
E-mail addresses: tamersh@hotmail.com, tamer.ahmed@med.asu.edu.eg (T.A. EL-Sobky).

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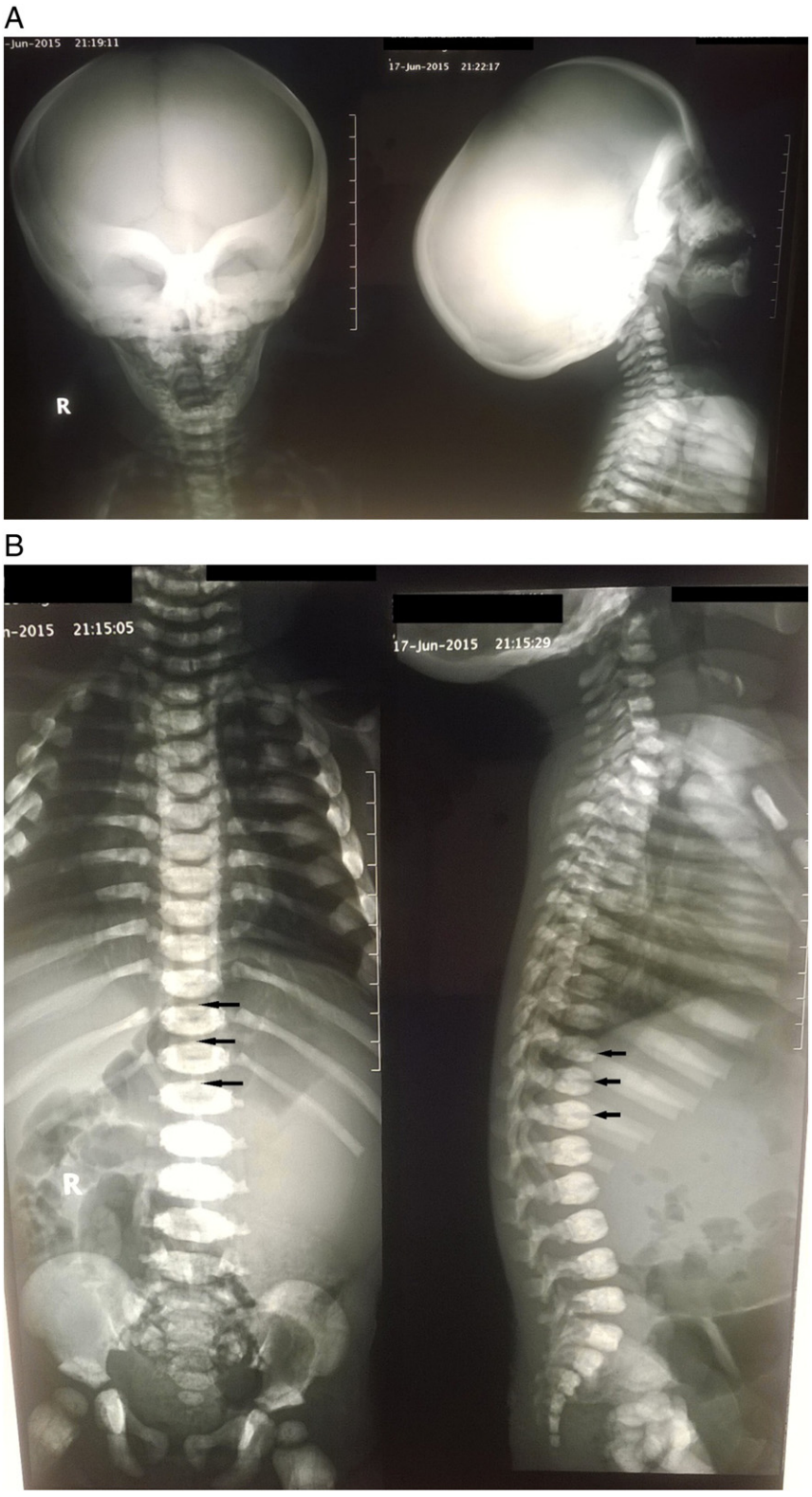

Fig. 1. Anteroposterior and lateral radiographs of the skull (A) and the spine (B) demonstrating an overall increased density of the bones with fundamental involvement of the medullary portion. Note the classic endobone or "bone-within-bone" appearance in the spine, the "sandwich vertebra" appearance, characterized by dense endplate sclerosis with sharp margins (black arrows).

Radiographic examination of the axial and appendicular skeleton revealed generalized osteosclerosis within the medullary portion of the bone with relative sparing of the cortices. Detailed radiographic abnormalities are depicted in Figs. 1A, B and 2A, B, C, D. The pelvi-abdominal sonography revealed splenomegaly with no focal lesions. The blood picture revealed moderate anaemia. Our patient's parents were informed that data concerning the case would be submitted for publication. The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper. No financing was received