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Case Report

Unusual presentation of chronic headaches revealing osteopetrosis: A case report ☆

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ABSTRACT

Osteopetrosis, also known as the disease of marbled bones, refers to a group of constitutional bone diseases resulting from a defect in bone metabolism. This condition is characterized by its manifestation, most often at a young age, and is typically revealed by its complications, primarily fractures. Diagnosis is currently confirmed through genetics but also relies on imaging such as standard radiography and computed tomography. We report the case of a child, aged 13 years, presenting with osteopetrosis revealed by atypical symptoms, confirmed by computed tomography imaging mainly in our country in Morocco where access to care is sometimes difficult for some patients.

Introduction

Osteopetrosis comprise a group of rare hereditary diseases affecting the skeletal system, characterized by increased bone density throughout the skeleton due to a defect in bone resorption by osteoclasts [1]. We report the case of a child presenting with osteoporosis revealed by atypical symptoms, confirmed through computed tomography.

Case presentation

A 13-year-old female patient was admitted to the pediatric emergency department of CHU HASSAN II due to acute onset intracranial hypertension syndrome. The patient is the product of a first-degree consanguineous marriage and has a history of chronic headaches since the age of 5 years, for which she had consulted several doctors without definitive etiology.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Her most recent consultation, 6 months prior to admission, was with an ophthalmologist who requested a brain MRI and conducted a normal examination, as well as normal brain MRI. A week before her emergency department admission, she experienced severe worsening headaches associated by projectile vomiting and photophobia.

Upon general examination, her vital signs were normal, with blood pressure at 100/70 mmHg. Signs of intracranial hypertension were noted, along with normal neurological, ENT, and ophthalmological findings. Lumbar puncture revealed an intracranial pressure of 21 mmHg, with cerebrospinal fluid showing white blood cell count of less than 3 cells/mm^3, red blood cells at 1100 cells/mm^3 (negative culture), protein level of 0.2 g/L, and glucose level of 0.69 g/L, yielding a cerebrospinal fluid glucose/blood glucose ratio of 0.6.

Standard laboratory tests (Table 1) showed hemoglobin level of 11.3 g/dL, white blood cell count of 6390 cells/mm^3 (with neutrophils at 5010 cells/mm^3 and lymphopenia at 1000 cells/mm^3), and platelets at 156,000 cells/mm^3. Infectious workup was negative, with CRP at 3.2 mg/L and sedimentation rate 05/20. Phosphocalcic balance was normal, but acidosis with alkaline reserves at 13 mmol/L was observed.

Ophthalmological examination revealed normal visual acuity in both eyes, normal anterior chamber, and normal fundus.

Brain imaging via computed tomography (CT) scan identified a lytic lesion at the spheno-occipital suture surrounded by osteocondensation. Osteocondensation of the axis tooth was also noted. A subsequent CT scan of the spine revealed diffuse bone marrow densification affecting various bones in both the axial and peripheral skeleton, consistent with osteopetrosis (Figs. 1–5). The diagnosis of osteopetrosis was confirmed through imaging. Symptomatic treatment with anti-edema medication (acetazolamide) resulted in regression of the intracranial hypertension syndrome. The patient was closely monitored by our team for potential complications.

Table 1 – Summary of the biological assessment of the patient who presents with osteopetrosis.

Biological assessment	Value	Normal range
CSF tap		
WC	3	<3
RC	1100	<3
Culture	Negative	Negative
CSF protein	0.2	0.15-0.45 g/L
CSF glucose	0.69	0.6-0.8 g/L
Intracranial pressure ICP	21	10-15 mmHg
WBC	6390	4000-10,000/mm^3
Hb	11.3	12-16 g/dL
Platelet count	156,000	150000-400000/μL
CRP	3.2	0-5 mg/L
Creatinine	7	7-11 mg/L
Urea	0.2	0.15-0.45 g/L
Albumin	36	35-50 g/L
Serum calcium	88	84-102 mg/L
Phosphorus	23	23-47 mg/L
Sedimentation rate	05/20	< 10 mm

Discussion

Osteopetrosis comprises a group of rare hereditary bone diseases characterized by abnormal bone resorption by osteoclasts [1]. They are classified into 3 forms based on genetic transmission. These include autosomal dominant osteopetrosis (ADO), also known as Albers-Schönberg disease, with an incidence of 5/100,000 births, predominantly seen in adults. Autosomal recessive osteopetrosis (ARO), also known as malignant infantile form, is rarer with an incidence of 1/250,000 births. The third form, X-linked osteopetrosis, is lethal and extremely rare [2,3].

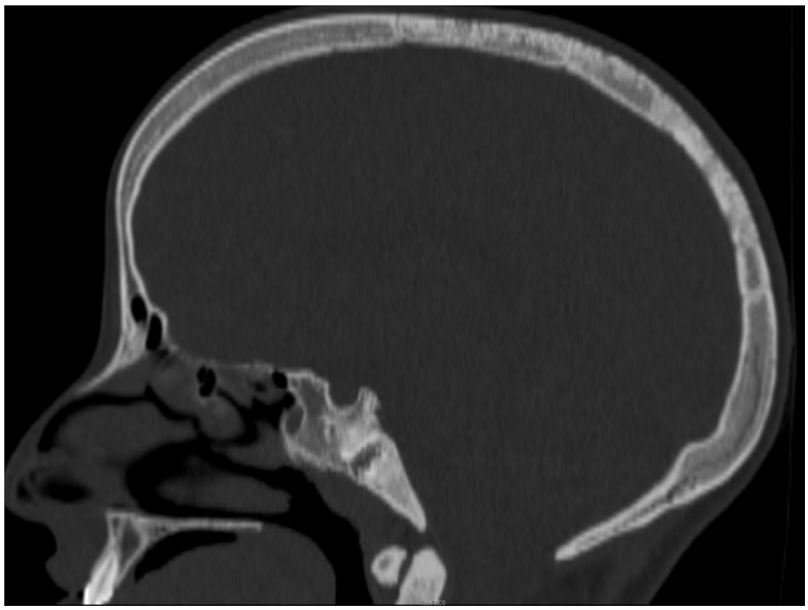

Fig. 1 – Sagittal CT scan section showing osteocondensation of the cranial vault and base of the skull.

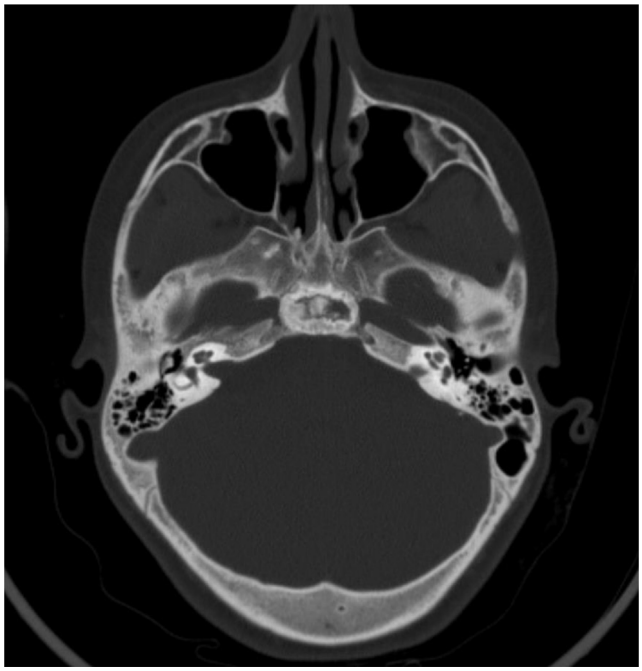

Fig. 2 – Axial CT scan section demonstrating osteocondensation of the cranial vault and base of the skull.

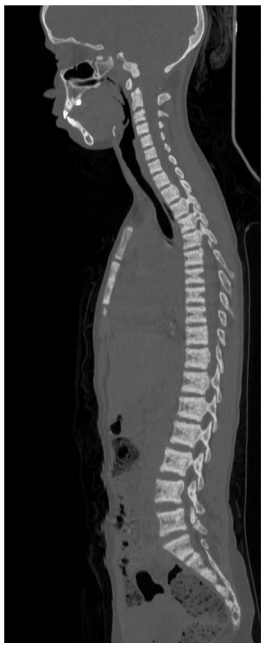

Fig. 3 – Sagittal CT image of the entire spine showing vertebrae with a "sandwich vertebrae" appearance.

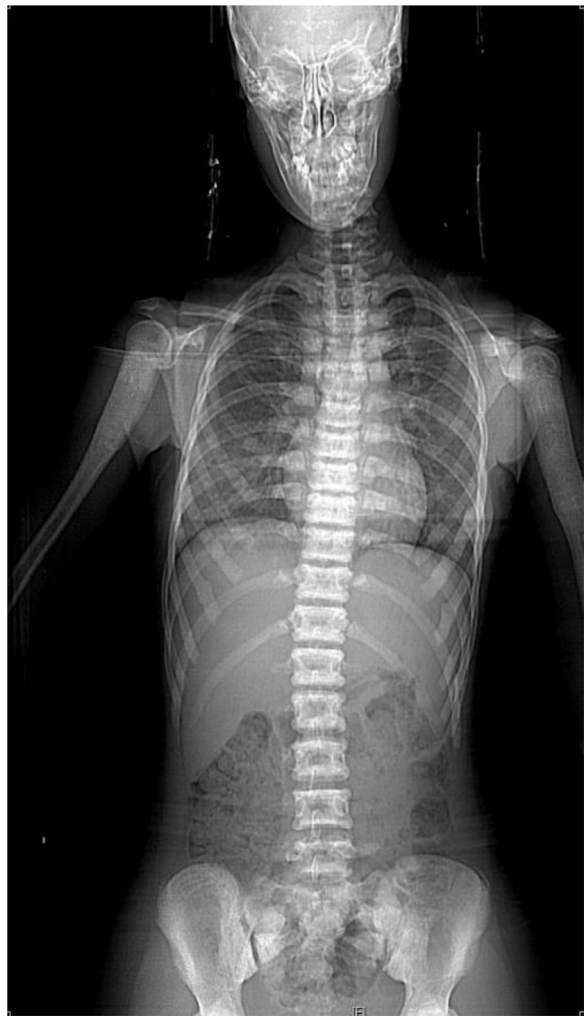

Fig. 4 – CT image showing metaphyseal widening with cortical thinning, resulting in an "Erlenmeyer flask" appearance in both humeri.

The clinical presentation of this group of pathologies varies widely, ranging from asymptomatic patients to lethal forms in infancy, depending on the mode of genetic transmission [2]. The dominant form (ADO) is further divided into 2 types: type 1, resulting from LRP5 anomaly, and type 2, known as Albers-Schönberg disease, resulting from CLCN7 mutation [1,4]. Albers-Schönberg disease is considered "benign," manifesting with radiological anomalies, fractures, osteomyelitis, or even pancytopenia [1]. The recessive form (ARO) is severe and can lead to death, manifesting early in life with small stature, fractures, frontal bossing, macrocephaly, typical facies, dental anomalies, and caries [5]. Neurologically, there is a significant risk of blindness, deafness, facial paralysis, and seizures. Biochemically, hypocalcemia with pancytopenia is observed [1,2]. The X-linked form generally associates with immune deficiency [2,6]. Diagnosis is primarily clinical and confirmed by radiographs, showing increased bone density, metaphyseal widening with cortical thinning, giving rise to an "Erlenmeyer flask" appearance in long bones and a "sandwich vertebra" appearance in the spine [1,2,7]. Cranially, there may be basal skull densification, as observed in our patient, confirming the diagnosis. Management of osteopetrosis is multidisciplinary, involving various specialties throughout the patient's life from childhood to adulthood, ranging from symptomatic treatment to transfusion needs to stem cell transplantation, depending on the clinical presentation [8,9]. Prognosis depends on the clinical presentation and the presence of complications [2].

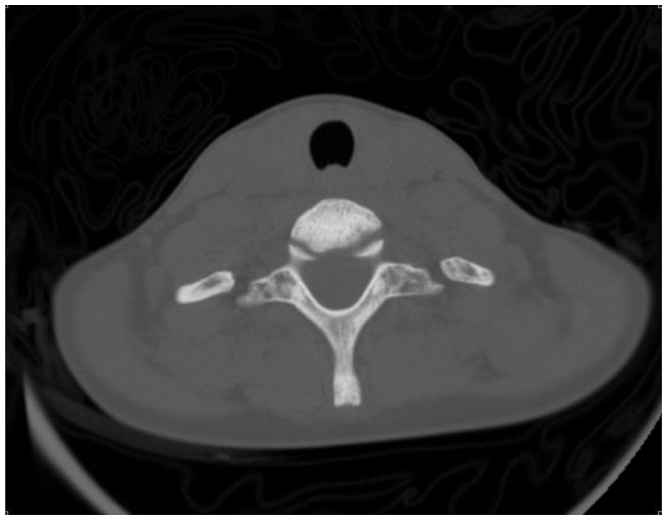

Fig. 5 – Axial CT scan image demonstrating osteocondensation of the vertebral body.

Conclusion

Osteopetrosis represents a group of diseases with highly variable clinical expression, which should be familiar to pediatricians to consider even in the presence of atypical symptoms. The key to management remains multidisciplinary, aimed at preserving the patient's prognosis.

Patient consent

I declare, I, the author of the article, have obtained the consent of the patient's legal guardian in order to publish of the patient's parents in order to publish the article for scientific purposes.

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