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Intracranial meningioma in a patient with osteogenesis imperfecta

Case Report

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Abstract: Osteogenesis imperfecta (OI) is a heritable disorder characterized mainly by connective tissue manifestations. In dinstinct cases, several neurological features have also been described. A 46-year-old male with a known family history of OI type I presented with progressive gait disturbances and diminished muscle strength. Brain MRI scans revealed an infiltrative intracranial mass occupying both frontoparietal lobes. The patient underwent surgical intervention. The histological diagnosis was an atypical (Grade II) meningioma. The bony parts demonstrated a mixture of osseous defects due to OI and infiltration by the tumor. At one-year follow up the patient's muscle power partially returned while repeat MRI scans were negative for tumor recurrence.

Keywords: *Osteogenesis Imperfecta • Meningioma • Tumor • Bone anomalies • Connective tissue*

1. Introduction

Osteogenesis imperfecta (OI) is a heterogenous genetic disease that affects the connective tissue. It is characterized by increased bone fragility and low bone mass [1,2]. Four main types can be distinguished [3]. The nervous system can be also affected and several neurologic features have been periodically reported [1,4-6].

The coexistence of an intracranial neoplasm with OI is considered to be extremely rare and only two cases have been reported [2].

A unique case of a patient with OI type IB associated with an atypical bilateral parasagittal meningioma is described.

2. Case Report

A 46-year-old male was admitted to the department of Neurology for investigation of progressive lower limb weakness and gait disturbances during the previous six months. He had a known family history of OI. In the past he had undergone several orthopedic operations for management of long bone fractures.

Upon physical examination, the patient was fully alert with normal mental functions. He had characteristic blue sclera, relatively short stature and a prominent hyperostotic cranial vault. Cranial nerve functions as well as upper limb motor activities were intact. Besides this he expressed diminished muscle strength in both lower limbs particularly during plantar flexion and extension and difficulty in walking. The Babinski sign was bilaterally positive. Sensitivity, higher functions and micturition were undisturbed.

The patient underwent cranial computed tomography (CT) and magnetic resonance imaging (MRI), which

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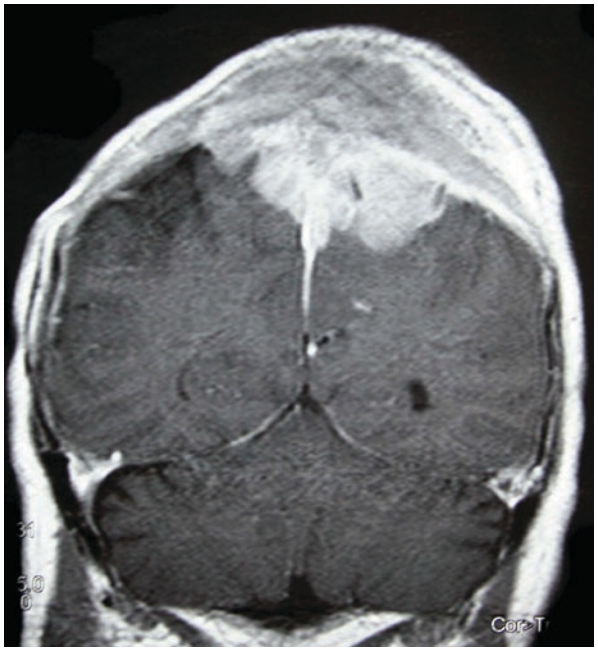
Figure 1. Preoperative coronal T1-weighted MRI study demonstrating a parasagittal infiltrative frontoparietal mass accompanied by bone hypeostosis.



revealed a large infiltrative intracranial mass compressing both frontal and parietal lobes anterior and posterior to the central sulcus in the cranial vault (Figure 1). He was then transferred to the Department of Neurosurgery for surgical treatment. In the course of the operation, some skull areas as well as a large part of the superior sagittal sinus seemed to be infiltrated by the tumor and the affected bone was extremely soft. A large craniectomy was performed. An extensive amount of dura was also inflitrated by the mass and it was excised. The tumor was gross totally removed. Meticulous duroplasty with a dural substitute was achieved. In the early post-operative period, the patient did not show any clinical improvement. Bony parts submitted for pathological examination demonstrated a mixture of osseous defects due to OI and tumor infiltration. The histological diagnosis was a Grade II meningioma (atypical) (Figure 2). The patient was discharged from the clinic and scheduled for cranioplasty for the bone defect. At one-year follow up, he manifested improved muscle power. Repeat MRI scans were negative for tumor recurrence (Figure 3).

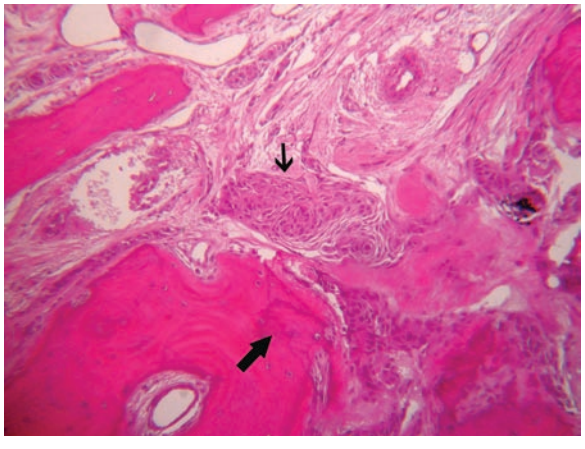
3. Discussion

Osteogenesis Imperfecta (OI) is a group of genetic diseases expressing, as a predominant feature, congenital bone fragility. Type I collagen is the defective protein of OI. Most patients with a clinically diagnosed OI express a mutation in one of the two genes that encode the α-chains of collagen Type I (COLIA1 and COLIA2) [1,7]. The result is a mixture of normal and abnormal collagen [8]. A positive collagen Type I mutation confirms the diagnosis. In patients with positive family history accompanied by certain clinical features the diagnosis can be established clinically [1].

The most widely adapted classification of OI was introduced by Silence [3]. It is based exclusively on clinical characteristics. Accordingly, four types are distinguished: Type I is referred to mild forms of the disease with minor deformities. Type II is typically lethal in the perinatal period. OI Type III OI is associated with major bone deformities secondary to bone fractures after minor trauma and with serious respiratory problems that may lead them to death [9]. OI Type IV is the second mildest form with affected patients usually demonstrating mild to moderate deformities [1,2]. It must be noted

Figure 2. a. Photomicrograph. Abnormal lamellar pattern indicative of osteogenesis imperfecta (large arrow). Note the infiltration of meningioma clusters between the bone trabecula (small arrow). H&E*200. b. Photomicrograph. Positive immunohistochemical vimentin stain for meningioma (arrow).

a



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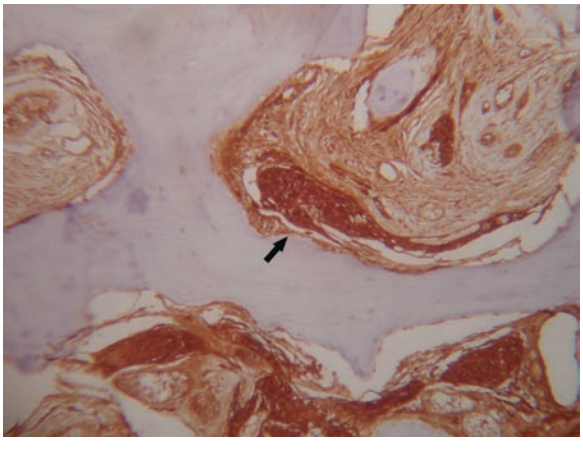
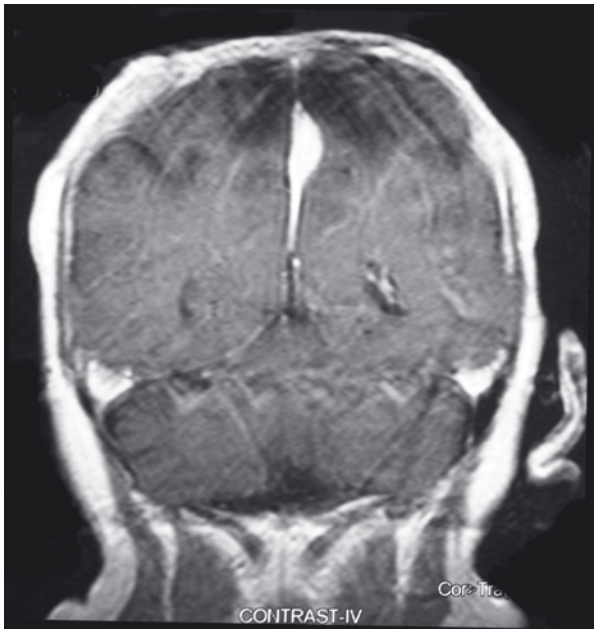


Figure 3. Postoperative coronal T1-weighted MRI study with contrast administration after 1 year. Craniectomy. No evidence of tumor recurrence. Small residual tumor in the falx.



however that the heterogeneity of Type IV has led to the identification of three new clinical entities based on the clinical and pathological characteristics [1].

Since Type I collagen fibers are found in several structures apart from the bones (organ capsules, fascia, cornea, sclera, tendons, meninges, and dermis), the extraskeletal manifestations of OI can be variable [10, 11]. Although frequent bone fractures prevail, associated manifestations of OI include mainly the presence of blue sclera, short stature, dentinogenesis imperfecta, hearing disturbances, hyperlaxity of ligaments and skin and evidence of wormian bodies on skull plain radiographs [1].

The spectrum of neurological features in patients with OI is wide, however, it has not been investigated thoroughly. The presence of basilar impression and/or invagination is an exception since numerous such cases have been repeatedly reported and analyzed [2,4,12,13]. Apart from basilar invagination which frequently leads to brain stem compression, other neurological complications identified in patients with OI include ventricular enlargement, sulcal prominence, macrocephaly, seizures, skull fractures, long tract signs, hydrocephalus, syringohydromyelia and intracranial hypertension [2,12].

According to the pertinent literature recorded intracranial neoplasms have been developed in two patients with OI. They included a grade II astrocytoma and a posterior fossa dermoid tumor [2]. The present case is the first reported presence of an intracranial meningioma in an individual with OI. One possible explanation of this coexistence could be the generalized disorganization of collagen type I in patients with this disease. This disorganization might have influenced and affected the meninges since collagen Type I is also abundant there, and led to the development of the tumor. A genetic correlation between a meningioma and OI has not been established so far. Several chromosomal anomalies have been implicated in the formation of meningiomas, but they seem to be unrelated to the pathogenesis of OI [14].

Currently, there is no cure for OI. Treatment with biphosphonate can reduce the risk of fractures during childhood, but there is not sufficient information concerning its use in adults [1,15]. Growth hormone has also been tested in small studies [1]. The use of bone marrow stem cells may offer beneficial results in the future [1].

In conclusion, although OI is not a very uncommon disease, most of its complex pathology and potential combinations with other entities remain poorly understood. Future advances, mainly in genetic medicine, are expected to shed further light on this entity and may pave the way for an efficient treatment.

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