Case Report

Neurosurgical Implications of Osteogenesis Imperfecta in a Child after Fall: Case Illustration

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Osteogenesis imperfecta (OI) is a group of hereditary genetic pathologies of connective tissue, which is characterized by bone fragility and fractures. It is classified into types I, II, III, IV, V, and VI. The disorder is caused by an autosomal-dominant mutation in one of the two genes that encode the alpha chains of type I collagen, COL1A1 and COL1A2. Several central nervous system abnormalities have been described in children with OI, however, it has been through various case reports. The neurological abnormalities that have been described are macrocephaly, ventriculomegaly, myelopathy, cranial neuropathy, basilar invagination, obstructive hydrocephalus, cranial fractures, and intracranial hemorrhage. In this report, we describe the clinical case of a child with parietal fracture; the main objective of this work being to show one of the several neurological implications that children with OI can present, and their implications for the pediatric neurosurgeons as neurosurgical complications are very frequent.

Keywords: Fracture, neurosurgical implications, osteogenesis imperfecta, pediatric

INTRODUCTION

Osteogenesis imperfecta (OI) represents a rare heterogeneous group of inherited disorders characterized by low bone mass and an increase in bone fragility.[¹-³] Different clinical manifestations have been described in these patients, is the most frequent: blue sclerotic, opalescent teeth, hearing loss, long bone deformity and spine, joint hyperextensibility motor development delay and short stature.

Neurological manifestations can also occur.[⁴-¹¹] In children, very few cases of neurological abnormalities have been reported, the most frequent being macrocephaly, acute intracranial hematoma, and obstructive hydrocephalus among others. The neurosurgical complications that these patients may present are serious and frequent.

CASE REPORT

A male patient of 1 year and 6 months of age presented with no family history, antenatal diagnosis of OI, and fracture of both legs and forearms at 5 months of age. He was born by scheduled cesarean delivery at 37 weeks of gestation with a weight of 2.930 g and size of 44 cm. He was clinically diagnosed of OI. He was started on a treatment of calcium + gluconate, vitamin D, and pamidronate. On June 4, 2014, he fell from a bed, 40-cm high, hitting the right parietal region. Enter in glasgow coma scale 14, without focal or long way deficit [Figures 1 and 2].

DISCUSSION

Imperfect osteogenesis is a disorder of the connective tissues that results in skeletal fragility, ligamentous laxity, and scleral discoloration.[⁴,⁸] This disorder is caused by a mutation in the gene encoding for type I collagen, the main structural protein in bone and many other connective tissues.

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OI is usually diagnosed early in life when a child presents with multiple bone fractures following minor trauma and is often misdiagnosed as a consequence of nonaccidental injury.[5]

Neurological aberrations observed in OI have not historically been classified with any particular subtype. However, in the reports that have been made, it has been observed that the most associated phenotype is type III.[11]

**Macrocephaly in patients with OI**

Macrocephaly has been observed previously in patients with OI.[10-12] A large retrospective analysis performed by the National Institutes of Health in 76 patients with OI found macrocephaly in 13.1% patients.[10,11] It was observed most frequently in patients with OI type III (6 [18%] of 33 patients).[12] Skull deformity, in general, occurs frequently in OI and may be manifested by flattening of the anteroposterior diameter and prominence of the frontal and occipital bones with a helmet-type appearance.[4]

**Bleeding diathesis**

Bleeding abnormalities have been reported in the literature.[9,13] The underlying origins have included abnormalities with vessel fragility and platelet dysfunction. Platelet function tests were performed in 15 patients with OI and their parents. The results showed that all patients exhibited impaired platelet factor 3 release. A study of 58 patients with OI found an increased rate of capillary fragility as well as reduced platelet retention.[4]

**Acute intracranial hematoma**

Two of our patients underwent emergency evacuation of an epidural hematoma, one without preceding trauma and the other following a minor fall. Two additional cases in the literature have been reported describing severe intracerebral hemorrhage after two young men with OI fell from their wheelchairs.[2,11] A patient with OI type I and concomitant end-stage renal disease developed an acute subdural hematoma (SDH) while undergoing dialysis without antecedent trauma, prompting neurosurgical evacuation with operative findings significant in the absence of an associated fracture. An analysis of death certificates in a cohort of patients with OI showed an increased incidence of intracranial hemorrhage secondary to trauma. The risk for the development of acute hemorrhage necessitating surgical intervention is increased in this patient population. Patients presenting with neurological complaints with or without antecedent trauma should be examined accordingly for possible hematoma formation.[4]

**Chronic SDH**

The incidence of chronic SDH in OI without inciting trauma is low as reported in the literature. Sasaki-Adams et al.[6] revealed three cases in which children were shown to have significant bilateral subdural fluid collections that were consistent with chronic SDHs. Two of these patients underwent operative intervention with placement of a subdural peritoneal shunt that was later converted to a ventriculoperitoneal shunt.

**Hydrocephalus in patients with OI**

Hydrocephalus has been described in OI in both communicating and noncommunicating forms. Basilar
invagination often underlies the obstructive subtype.\textsuperscript{6,12} The incidence of hydrocephalus in patients with OI is not known. Sulcal prominence with ventriculomegaly attributed to hydrocephalus was observed in only 3 of the 76 patients in the study conducted by the National Institutes of Health. In comparison, 17 of the 76 cases of patients with ventriculomegaly and sulcal prominence were attributed to cerebral atrophy.

**Basilar invagination**

Basilar invagination has been observed in multiple series of patients with OI as well as with other osteodysplastic syndromes.\textsuperscript{4,7} The experience of Sawin and Menezes\textsuperscript{12} at the University of Iowa provided the most in-depth discussion regarding the natural history of this entity in this population and strategies in its management. Over the 10-year period, 25 patients with OI or related osteochondrodysplastic with basilar invagination were evaluated. Asymptomatic patients were treated with the external orthosis. Symptomatic patients underwent a trial of cervical traction in an attempt at reduction. In those patients in whom reduction occurred, a posterior fossa decompression was undertaken followed by posterior fusion by using a wire construct. In those patients with ventral compression, a ventral decompression was first performed followed by posterior fusion. All patients showed adequate fusion of mass postoperatively. However, 80\% of patients showed the progression of basilar invagination.\textsuperscript{4}

**Spine fractures and deformity**

Spinal fractures and deformity are observed with a relatively high frequency in patients with OI. Scoliotic-type deformities are seen commonly in patients with OI. They may be found in between 40\% and 90\% of cases with the more severe subtypes. The treatment of choice appears to be operative fusion with instrumentation and possible administration of methyl methacrylate as an adjunctive measure.\textsuperscript{4}

In conclusion, although OI is a rare genetic disorder, the neurosurgical implications in children can be associated with serious and fatal neurological abnormalities.

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**Conflicts of interest**

There are no conflicts of interest.

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