

# Child abuse and osteogenesis imperfecta: how can they be still misdiagnosed? A case report

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## Summary

**Osteogenesis imperfecta (OI) is a rare hereditary disease caused by mutations in genes coding for type I collagen, resulting in bone fragility. In literature are described forms lethal in perinatal period, forms which are moderate and slight forms where the only sign of disease is osteopenia. Child abuse is an important social and medical problem. Fractures are the second most common presentation after skin lesions and may present specific patterns. The differential diagnosis between slight-moderate forms of OI and child abuse could be very challenging especially when other signs typical of abuse are absent, since both could present with multiple fractures without reasonable explanations. We report a 20 months-old female with a history of 4 fractures occurred between the age of three and eighteen months, brought to authorities' attention as a suspected child abuse. However when she came to our department physical exa-**

mination, biochemical tests, total body X-ray and a molecular analysis of DNA led the diagnosis of OI.

Thus, a treatment with bisphosphonate and a physical rehabilitation process, according to Vojta method, were started with improvement in bony mineralization, gross motor skills and absence of new fracture.

In conclusion our case demonstrates how in any child presenting fractures efforts should be made to consider, besides child abuse, all the other hypothesis even the rarest as OI.

*KEY WORDS:* osteogenesis imperfecta; child abuse; physical rehabilitation; neridronate.

## Introduction

Osteogenesis imperfecta (OI) is a rare disease, genetically determined, due, in 90% of cases, to mutations in genes coding for type I collagen's chains. The disease state encompasses a phenotypically and genotypically heterogeneous group of inherited disorders that may result in alterations at level of all those tissues where collagen type I is present, especially bone, sclera, dentin and ligaments (1). The main feature of OI is bone fragility, with fractures occurring even for ordinary trauma.

OI is classified according to the classification of Sillence et al. (2, 3) in four types. This classification was later enlarged by Glorieux et al. (4, 5) with five new type of OI divided on the basis of clinical manifestation such as blue sclera, low stature, deformity and dentinogenesis.

Treatment of OI is essentially the orthopedic surgery but a medical treatment with bisphosphonate is also indicate to increase bone mineral density. Moreover physical rehabilitation is important, since it could ameliorate the clinical management of OI. The prolonged immobilization, the fear of the pain, the hyper-protection of the family, are the main factors that limit the patient's physical activity and may therefore worsen the muscular hypotonia and the osteoporosis, that are typical of this condition.

Child abuse is an important social and medical problem which represents a major cause of morbidity and mortality among children. Fractures are the second most common presentation after skin lesions and they present specific patterns. The differential diagnosis includes other conditions leading to fractures like slight-moderate forms of OI. Characteristics of OI, which may be useful in differentiation from child abuse, include blue sclera, tooth involvement, osteopenia, family history, Wormian bones on skull X-ray, and deformity (6). However, in patients with milder forms of OI, where those signs are absent, the diagnosis can be particularly vexing.

We report a case of OI type I misdiagnosed as child abuse in which medical and physiotherapy treatment was successful despite a tardive diagnosis.

## Case report

A female baby of 20-months was referred to our Department of Pediatrics for suspicion of skeleton's disease, after a history of four fractures and a signaling for child abuse.

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She was born full-term vaginally. Her birth weight was 3650 g and the neonatal period was normal. She is second of two children, brother and parents in good health, grandmother suffering for osteoporosis. Her first fracture occurred at the age of 4 months when the little baby sustained an accidental fall from an outpatient couch. The CT scanning performed at the emergency department showed the presence of a fracture of the right parietal bone with epidural hemorrhagic shedding.

At the age of 5 months as a consequence of a simple domestic accident she suffered a displaced distal supracondylar humerus fracture that required a prompt closed reduction. When patient was 16 months old she reported a displaced fracture of left radius and ulnar midshafts treated with immobilization in plaster cast (Figure 1). The parents told the event as result of an extremely soft blow. The fourth access at the emergency department was 2 months later when she fractured her right tibial midshaft (Figure 2). Once more the parent reported simply an ordinary accident occurred at home but, in this occasion, the case was reported to the Authority as a suspected case of child abuse and a series of investigation on the family began. From the Department of Orthopedics and Traumatology where she had been hospitalized after her last fracture, the child was referred to our observation for a suspected skeleton's disease.

On admission, physical examination showed blue sclera, muscular hypotrophy of lower limbs, ligamentous laxity of hands and a mild hypotonia of trunk and lower limbs.

Biochemical tests highlighted an alteration of all the main markers of bony metabolism: alkaline phosphatase 293U/L (v.n. 39-111 U/L) bone alkaline phosphatase 134 U/L (v.n. <20 U/L)  $\alpha$ -crosslaps 3291  $\mu$ g/mM (v.n. 45-1035 g/mM) osteocalcin 43 mg/mL (v.n. 2,7-12,2 mg/mL).

A total body X-ray was performed, showing a generalized reduction of bones calcium content and in particular, at the level of cranium, diploe's sponginess and lambdoid perisutural accessory bones. Presence of multiple fractures of different ages appeared on the limbs associated with a valgus condition of femoral necks and a mild bending of distal part of femoral shafts, especially for the left. Moreover a varus condition of tibias was evident.

Thus, the clinical findings, biochemical markers and radiographic images addressed our diagnosis to OI, later confirmed by a molecular analysis of DNA, which revealed the heterozygote mutation of the exon 19 of gene Col1A1.

We started, every 3 months, a therapy with intravenous Neridronate, a bisphosphonate licensed in Italy for OI treatment. It was prolonged for two years with an improvement in bony mineralization, biological markers and absence of new fractures.

The patient, admitted to the Rehabilitation Center, presented poor motor initiative in the spontaneous behaviour and she appeared to be afraid to get toys. There were no observable post-traumatic and post-immobilization stiff joints, but the muscles of trunk and lower limbs were hypotonic. The Gross Motor Function Measure GMF-88 was performed and resulted 76/264 (7). The first rehabilitation approach consisted in Vojta method exercises to restore the muscular strength according to the motor development milestones, integrated with exercises in game forms, in order to facilitate the spontaneous motility (8). The parents were trained to the Vojta method exercises in order to continue the rehabilitation process after discharge. After 3 months of treatment the patient was able to walk without assistance. The GMFM score was 189/264.

## Discussion

In our case child abuse could have been excluded for the absence of clinical and radiographic signs characteristic of violence and the presence, instead, of a pattern suggestive for OI.



Figure 1 - Radiograph of the left arm shows displaced fracture of radial and ulnar midshafts.



Figure 2 - Lateral radiograph of right lower limb showing a spiral fracture of tibial midshaft.

A high suspicion was present since the first access to the emergency department, represented by the blue sclerae and the Wormian bones.

Blue sclerae are an anomaly due to a reduction in thickness of scleral fibrous tissue that makes appear the blue colour of the

uvea below. Blue sclerae could be normal in baby up to 4 months of age, but after this time they are a sign indicative for connective tissue alterations.

Wormian bones are perisutural accessory cranium bones. They could be simply an anatomic variant without any pathological impact or could be a marker of a specific syndrome, such as OI. Anyway, the contemporary presence of blue sclerae and wormian bones, that considered singularly are already rare in normal population, should have certainly lead the suspect of OI. This hypothesis could have been reinforced, in occasion of the subsequent access, analysing accurately fracture's type and location.

In fact, voluntary inferred injuries have so characteristic location and radiographic pattern that lesions can be classified as of high, moderate or low specificity for child abuse (9). According to this classification the lesions with the highest specificity for abuse are rib fractures and classical metaphyseal lesion.

Rib fracture has a predictive value of 95% for child abuse (10), especially if posteromedial and when occurring in children with less than three years of age. The characteristic mechanism that they need to be produced is a compression around the chest accompanied by the act of squeezing anteriorly and posteriorly the thorax (11). This type of fracture is extremely rare in patient with OI and it occurred generally only in the most severe forms but on lateral side.

Regarding the classical metaphyseal lesions, they are highly specific in children in their first year of life. The accepted mechanism for this type of fracture is violent shaking of a young child, causing a whiplash-type injury at the level of the zone of provisional calcification (12, 13). They have never been described in patient with OI.

In our patient none of the lesions occurred showed the character of high specificity for abuse. In fact, except for the first fracture involving cranium, all the other were at the level of long bone's midshaft as typical for OI. In fact, the most brittle bone's part in those patients, is represented by the midshaft and not by the physis, where type I collagen is absent (14). In addition all the other sign of abuse, such as bruises or burning were absent. Therefore in our patient the investigations should have been made looking for OI besides a condition of abuse.

However our case, although the diagnosis has been delayed, demonstrates as a rehabilitation process, through a specific and comprehensive physiotherapeutic approach, could anyway contribute to the optimal management of the disease, as shown by the improvement of GMFM scores. The Vojta method we adopted, due to its peculiarity of reproducing the physiological simulation of body structures, and being in keeping with the motor development milestones, represents a secure way to promote the spontaneous motility of the child and give the possibility to continue the therapy at home. The Vojta

method a few decades ago was described as a constraining and hard method. The case shows as this method can be used in secure way in case of bone fragility since parents were trained to its application at home, establishing a deep and mild contact with their daughter without any fear of new traumas.

In conclusion our case demonstrates how in any child presenting multiple fractures, efforts should be made to consider all the causes even the rarest as OI. It is true that child abuse is pervasive while OI is a rare disease, but this last hypothesis has to be considered, since the consequences of a wrong diagnosis should be dramatic for the family accused (5).

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