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CASE REPORT

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Osteogenesis imperfecta type V, spot diagnosis

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Summary

Background: The first case of Osteogenesis Imperfecta Type V in the Polish literature is reported.

Case Report: Skeletal survey of an 8 year old girl with a history of multiple fractures and bilateral dislocation of radial heads was received for consultation.

Conclusions: Generalised osteoporosis with multiple fractures, periosteal thickening and bilateral dislocation of the radial heads are characteristic signs of osteogenesis imperfecta Type V. The Nosology and Classification of Genetic Skeletal Disorders 2006 Revision classified Osteogenesis Imperfecta into 8 major types. Type V is recognizable on the basis of skeletal survey alone.

Key words: osteoporosis • fractures • radial head dislocation • periosteal thickening • ectopic ossification • osteogenesis imperfecta

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Background

Osteogenesis imperfecta (OI) is a group of different disorders characterized by abnormal phenotype of variable expressivity, increased bone fragility, hyperextensibility of the joints, blue sclerae and dentinogenesis imperfecta. The characteristic radiographic findings include osteopenia, multiple fractures, and usually Wormian bones [1,3]

Sillence [4] classified OI in four types I-IV on the basis of clinical and radiographic findings. Later Type II characterized by multiple fractures and severe deformities was subdivided in Types A, B, and C. Types V and VI were included by Glorieux et al. [5] due to similarity of microscopic appearances to types I–IV. Type VII was added in a Seminar on OI published by Lancet [6] and Type VIII by Cabral et al. [7] because of recessive type of inheritance and distinctive gene abnormalities (Table 1).

Discussion

Skeletal survey of our patient showed all the features - osteoporosis, anisospondyly, bilateral dislocation of the radial heads and localised hyperplastic callus formation – of "Unclassified demineralizing bone disease – new entity", as reported by Kozlowski and Bittner in 1981 [2] and renamed OI Type V by Glorieux et al. in 2000 [5]. Ectopic interosseous membrane calcification appears in older individuals. It is possible that the two siblings reported by Coenegracht et al. [8] as "Diaphyseal aclasia combined with fragilitas ossium" represent advanced cases of OI Type V.

This author believes that incorporation of OI Type V into OI group is erroneous because of normal phenotype and different radiographic findings – long bones fractures are close to metaphyses, there is bilateral radial head dislocation, distinctive asymmetrical localized hyperplastic periosteal thickening and absence of Wormian bones. Additionally abnormal bone-modeling markers [2,9] are not observed in Types OI Types I-IV, VI, VII and VIII. Diagnosis of OI Type V is easy if the disease is known to the observer [2,5].

Other types of OI which can be diagnosed on radiographic examination alone are OI Type IIA (lethal, thick bone variety) and OI Type IIC (lethal thin bone variety) [3]. The severe Type IIB presents with osteoporosis and variable pattern of fractures and deformities. Diagnosis of the remaining types of OI requires other clinical information. Clinical history (inheritance) and phenotypic evaluation are most important, but bone histomorphology and DNA investigations may be necessary in some patients.

The differential diagnosis of OI Type V varies with the age of the patient. The early stages of OI V are unknown as all the reported cases are children in school age. In every child with grossly normal phenotype and limitation of elbow movements and/or bilateral radial head dislocations OI

Table 1. Classification of osteogenesis imperfecta 2009 [1].

Type	Severity	Mutation	Inheritance	Biochemical markers	Radiology
OI I	+/++	COL1A1	AD	–	+/++
OI II					
A	+++	COL1A1/2	AD	–	+++ Diagnostic
B	+++	COL1A1/2 CRTAP LEPRE1	AD AR	–	+++
C	+++	?	AD	–	+++ Diagnostic
OI III	++/+++	COL1A1/2	AD	–	++/+++
OI IV	+/++	COL1A1/2	AD	–	+/+ +
OI V	**+**	**?**	**AD**	**+**	**+ Diagnostic**
OI VI	++/+++	?	AR	+/–	++/+++
OI VII	+/++	CRTAP	AR	–	+/++
O I VIII	+/++	LEPRE1	AR	–	+/++

+ mild to moderate; ++ moderate to severe; +++ severe, usually lethal; +/– alk phosphatase may be raised; AD – autosomal dominant; AR – autosomal recessive.

Table 2. Differential diagnosis of OI Type V.

Disorder	Phenotype	Inheritance	Clinical diagnostic characteristics	Radiographic diagnostic characteristics	Genetic defect
OI V	-	AD	Radial head dislocation	Osteopenia, fractures, periosteal callus formation. Radial head dislocation	?
OI other types	+/-	AD AR	Deformities, blue sclerae, dentinogenesis imperfecta, thin skin, hypotonia	Osteopenia, fractures, slender bones, deformities, large head, wormian bones	COL1A1 COL1A2 CRTAP LEPRE1
Idiopathic osteoporosis	–	?	–	Osteopenia, Fractures	?
Child abuse syndrome	–	-	Soft tissues injuries	Fractures – different stages	-
Bruck syndrome	+	AR	Congental joint contractures, pterygia	Osteopenia, fractures, slender bones, deformities, wormian bones	Bone specific telopeptide lysil hydroxylase
Osteoporosis psudoglioma syndrome	+	AR	Ocular abnormalities, blindness	Osteopenia, fractures	LRP5
Cole-Carpenter syndrome	+	?	Orbital hypoplasia, ocular proptosis	Osteopenia, fractures, cranio-synostosis	?

Type V should be considered. During childhood the differential diagnosis is with all diseases presenting some of the features of OIV such as osteoporosis, anisospondyly, multiple fractures, periosteal thickenimg and radial head dislocation. These *includes predominantly other types of OI*. None of them shows asymmetrical hyperplastic callus, bilateral symmetrical radial head dislocation or calcified interosseous membrane. *Idiopathic osteoporosis* shows some features of OI such as osteoporosis, anisospondyly and fractures, but localized asymmetrical hyperplastic callus, interosseous calcification and bilateral radial head dislocations are not features of IO. *Child abuse injury* (*Battered baby syndrome)* presenting with osteoporosis and multiple fractures may be misdiagnosed as OI, especially when adults in charge of the child, deny child abuse. However periosteal reaction in battered baby syndrome shows different stages of evolution, extraosseous soft tissues lesions are usually present and Wormian bones are absent.

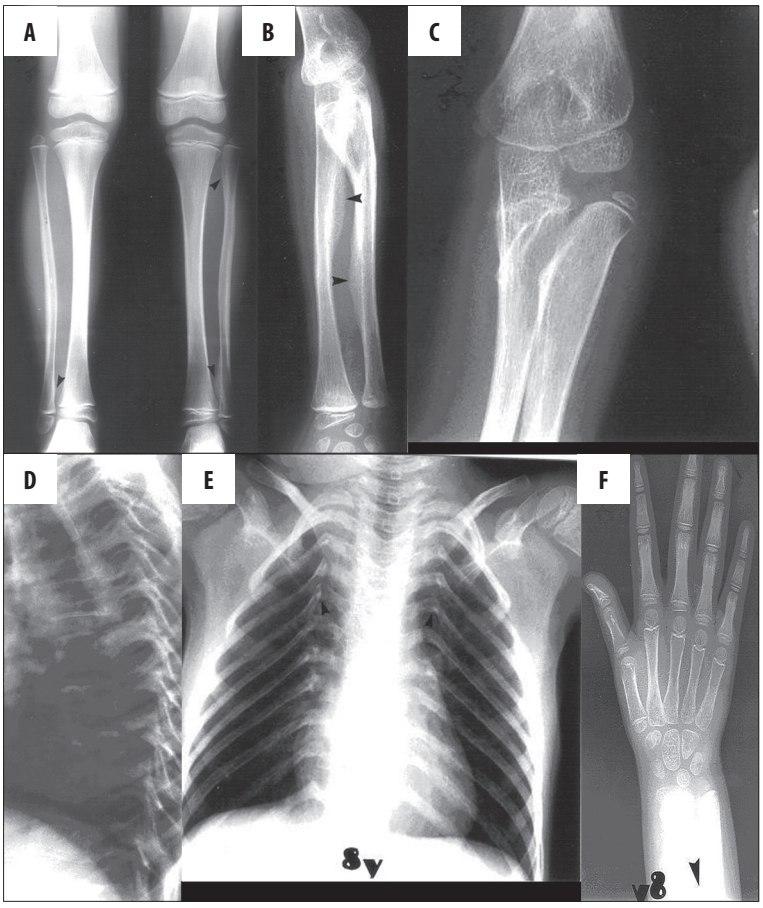

Figure 1. Girl 8 years old. Generalised advanced osteoporosis. (**A,B**) Localised periosteal thickening (arrowed). Note absence of rickets like changes! (**C**) Bilateral radial head dislocation. (**D**) Thoracic spine – anisospondyly. (**E**) Thin, obliquely positioned ribs. (**F**) Normal bone age. Localised periosteal thickening – arrowed.

Other congenital syndromes associated with osteoporosis and bone fragility such as *Bruck syndrome* [10], *osteoporosis-psudoglioma syndome* [11] and *Cole-Carpenter syndrome* [12] are unlikely to cause confusion if the radiographic findings are correlated with phenotype and clinical history of the patient (Table 2),

Disorders causing periosteal reaction with or without osteoporosis and fractures such as *Melnick- Needles syndrome, periosteal tumours, bleeding disorders* – and *juxtacortical myositis ossificans* are unlikely to be misdiagnosed. They show different patterns of periosteal reaction and usually have a definite positive etiological clinical history.

In instances when only one x-ray is presented for diagnosis, confusion with cortical or parosteal osteosarcoma, periostitis, myositis ossificans, subperiosteal hematoma secondary to trauma, bleeding disorders, or mesodermal dysplasia can occur. MR, CT and/or nuclear scan may be helpful in the differential diagnosis [13].

In adults the diagnosis of OI Type V may be more difficult because of superimposed traumatic and osteoarthritic changes.

Conclusions

An 8 year-old girl with OI Type V is reported. Radiographic examination documented osteoporosis, anisospondyly, fractures, bilateral radial head dislocation and hyperplastic callus. Presence of this type of malformation pattern allows a spot diagnosis of OI Type V and makes other diagnostic tests unnecessary.

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