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CASE REPORT

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A newborn with multiple fractures

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Summary

Background: Sometimes newborns with multiple fractures are diagnosed as osteogenesis imperfecta in spite of absence of radiographic findings supporting this diagnosis.

Case Report: A newborn with multiple fractures was diagnosed as osteogenesis imperfecta. Analysis of the structure of the long bones, pattern of fractures and poorly developed muscles suggested the diagnosis of fetal akinesia deformation syndrome. This was confirmed by pregnancy history and clinical findings.

Conclusions: Multiple fractures in a newborn may present with diagnostic radiographic features as in osteogenesis imperfecta, or as in lethal gracile bone dysplasias or achondrogenesis type IA. If those features are absent, other diseases should be considered. Radiographs should be compared with pregnancy history and clinical findings in the newborn.

Key words: newborn • fractures • osteogenesis imperfecta • gracile bones dysplasia • polyhydramnios • oligohydramnios

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Background

Multiple fractures are characteristic of a number of inherited bone dysplasias and metabolic disorders. Usually they appear well after the newborn period, during childhood or later in life. Each of these conditions should be evaluated together with the clinical history and other often diagnostic radiographic features.

Case Report

The proband was the first child of healthy, young non-consanguineous Caucasian parents both aged 27 years. No relevant past history was found. She was delivered at term by Caesarean section because of breech position and suspected fetal asphyxia. Ultrasounds reported at the age of 19 and 39 weeks were normal

Birth weight was 2900 g. The mother noticed that during the pregnancy the baby was inactive.

Soon after birth the infant was hypotonic and in respiratory distress requiring artificial respiration. Low set ears, small nose with anteverted nostrils, quadrate skull with wide forehead were identified, along with gothic palate. The eyes were normal,

The hands were small with conic fingers and without flexion creases apart from those at the metacarpo-phalangeal joints, The nails were hypoplastic.

The karyotype was 46XX, The biochemical tests showed hypoproteinemia.

Radiographic examination documented symmetrical fractures of the femora and left humerus.

The girl died at 21 days because of multi-organ failure from hypoxia (generalized edema with renal failure, hypoxy-ischemic encephalopathy, bleeding into the respiratory and gastrointestinal tracts).

Table 1.

Disorder	Phenotype	Radiographic findings	Remarks
Osteogenesis Imperfecta Types IIA, IIC and III/IIB	Variably dysmorphic phenotype	Osteoporosis, thin or thick twisted tubular bones, undermineralised skull	Blue sclerae Hyperlaxity Hypotonia
Gracile Bone Dysplasias	Dysmorphic phenotype	Fish bone diaphyses Metaphyseal flaring Undermineralised calvarium	Often low birth weight
Oligohydramions Deformation Sequence	Dysmorphic phenotype	Gracile long bones Asymmetrical fractures	Asymmetrical contractures
Fetal Akinesia Deformatio0n Sequence	Dysmorphic phenotype	Gracile long bones Reduction of muscle bulk Symmetrical fractures	Polyhydramnios Inactive fetus Symmetrical contractures
Achondrogenesis Type IA	Dysmorphic phenotype	Multiple rib fractures	Short trunk and very short extremities
Traumatic birth	Local deformities	Single usually clavicle; asymmetrical if multiple fractures	Complicated birth

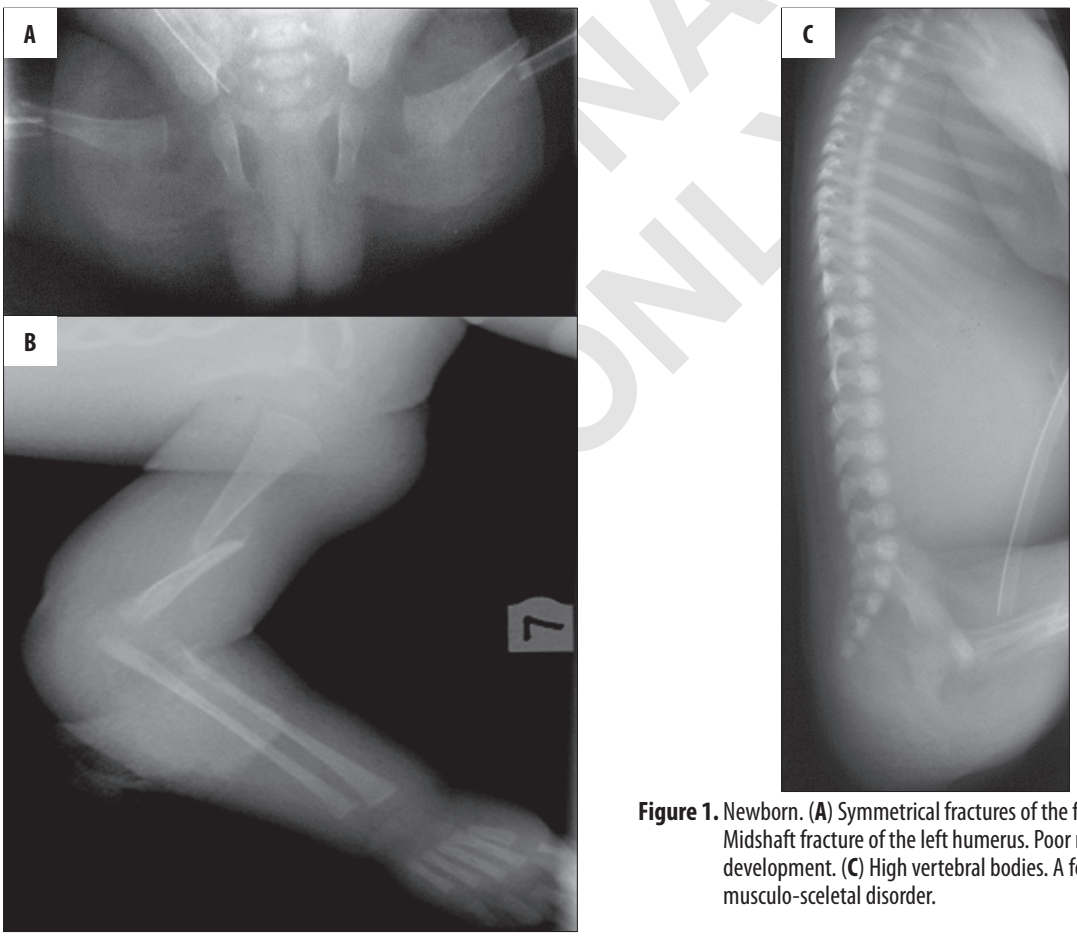

Figure 1. Newborn. (**A**) Symmetrical fractures of the femora. (**B**) Midshaft fracture of the left humerus. Poor muscular development. (**C**) High vertebral bodies. A feature of a musculo-sceletal disorder.

Histological examination of the muscle tissue showed severe muscle atrophy with hemorrhage and reparatory connective tissue process. Femoral nerve biopsy documented atrophy, demyelinisation, mononuclear infiltration around some lemniscuses and fibrosis of some connective tissue. In the brain there was spongiform edema and diffuse activation of microglia, There was bronchopulmonary dysplasia with patches of atelectasis. In the kidneys necrosis of proximal tubules was present.

Discussion

In newborns, the differential diagnosis of multiple fractures comprises only few entities (Table 1). The most common disorder is **osteogenesis imperfecta** (OI). Generalised osteoporosis with underossified skull and spine, anisospondyly, multiple long bone and rib fractures make the diagnosis of OI usually easy.

The heterogeneous group of **lethal gracile bone dysplasias** (LGBD) is characterized by distinctive facial dysmorphology, moderately short limbs, small hands and feet. Absence/aplasia of the spleen is a distinctive feature. The diagnostic radiographic findings include extremely slender tubular bones (fish bone diaphyses), flared tubular bones and undermineralised skull simulating cloverleaf skull.

Multiple fractures with slender bones are a feature of **oligohydramnios deformation syndrome** (ODS) and **fetal akinesia deformation sequence** (FADS). Both are a result of akinesia – primary in ODS and secondary in FADS. Both are characterized by a short umbilical cord, limb positional deformities and pulmonary hypoplasia. ODS is caused by oligohyramnios, FADS is the result of neuromuscular disorders. Multiple rib fractures are characteristic of **Achondrogenesis type IA**. The most common single fracture of the clavicle may occur during **traumatic delivery.**

Neonatal fractures should be differentiated from **pseudofractures** such as occur in the clavicles in cleido-cranial dysplasia or as isolated **congental pseudoarthrosis** most common in the clavicle and tibia.

Absence of fetal movements is an important forewarning sign of a most likely musculo-skeletal abnormality. In our case symmetry of fractures in a hypotonic newborn were consistent with the diagnosis of a fetal akinesia deformation sequence. The differential diagnosis of FADS disorders comprises *Spinal Muscular Atrophy Type O (Werdnig-Hoffmann disease) (WHD), Arthrogryposis multiplex congenita syndromes (AMCS)* and *Congenital Myotonic Dystrophy (CMD).* WHD type O denotes most severe form of the disease such as in our patient. It is characterized by intrauterine onset with neonatal asphyxia, severe diffuse weakness and early death secondary to aspiration and respiratory complications. Types 1–3 denote forms with later onset and less dramatic course. AMCS are characterized by fixed position of multiple joints with limitation of movement. CMD shares pregnancy characteristics – polyhydramnios and absence of fetal movemets - with WHD. Neonatal hypotonia, facial diplegia and arthrogryposis of the lower extremities are distinctive clinical features of CMD. Impaired swallowing, aspiration and respiratory complications result in early fatal outcome.

Conclusions

The implication of this case to radiologists, neonatologists and orthopaedic surgeons is that although OI is the most common disease in a newborn with fractures other possibilities should not be overlooked. In case of a newborn with multiple fractures without diagnostic radiographic features of OI, LGBD or Achondrogenesis type IA, radiograms should be evaluated with the history of the mother's pregnancy and the clinical findings in the child.

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