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RESEARCH ARTICLE

OSTEOGENESIS IMPERFECTA IN THE NEWBORN: ABOUT A CASE

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Abstract

Osteogenesis imperfect (OI) is a rare hereditary constitutional disease showing varying severity characterized by bone fragility, secondary to a defect in the synthesis of collagen type I. Its management is multidisciplinary. We report a case of neonatal discovery of OI at Mohammed the VI University Hospital in order to identify diagnostic and therapeutic difficulties and improve the vital prognosis.

Introduction:

Osteogenesis imperfecta (OI) is a hereditary disease characterized by bone fragility, secondary to a defect in the synthesis of type I collagen (1). We report a case of neonatal discovery of OI at Mohammed the VI University Hospital ,with the aim of raising the diagnostic and therapeutic difficulties.

Methods:-

Through our observation we report the clinical, para-clinical, evolutionary aspect as well as the therapeutic management of neonatal osteogenesisi mperfecta, a disease quite rare in our context.

Observation:

New born male admitted on day 10 of life for etiological assessment of multiple fractures suspected of malformations of the four limbs; Having as a family history, a sister who died at 6 months of life because of osteogenesis imperfecta.

The mother benefited from obstetric ultrasounds revealing bone deformities. The serologies including toxoplasmosis, rubella, syphilis were all normal. No intercurrent pathology was noted during pregnancy. Delivery was by caesarean section at term (40 weeks + 4 days) without incident.

The examination on admission finds a pink new born, reactive, gesticulates spontaneously, dysmorphic facies, with deformation of the 4 limbs, afebrile at 36.4 normocardia at 140 bpm eupneic 40 cpm with a weight at 3kg500, a height at 50cm and head circumference at 36cm. good axial and peripheral tone with enlarged anterior and posterior fontanelle, four arched deformed limbs, no pain on palpation, slight perineal edema, the rest of the examination is unremarkable.
Frontal thoraco-abdominal and limb radiograph showing demineralization of the entire bone structure, particularly the long bones. There are several pathological fractures with malunions on the long bones of the limbs (figures 1-2).

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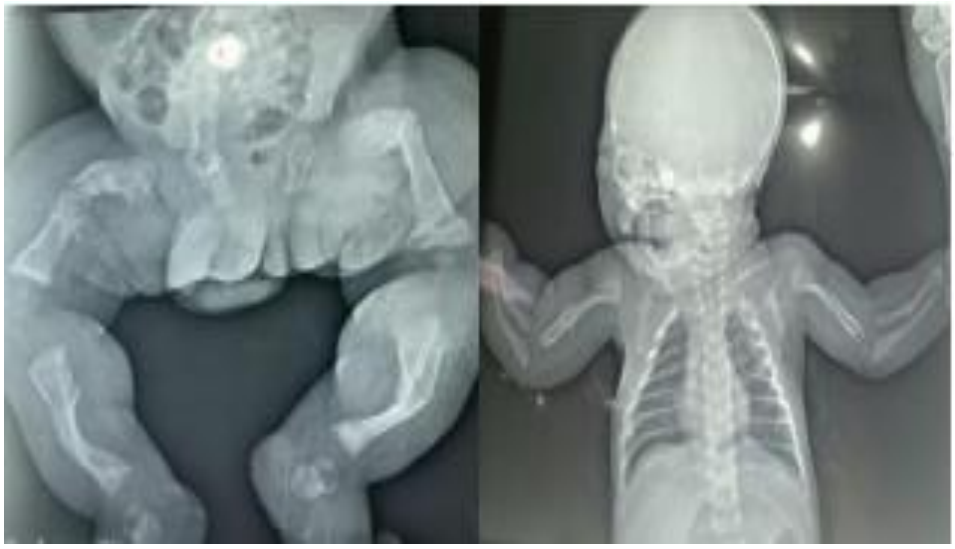

Figure 1-2:- Frontal thoraco-abdominal and limb radiograph.

In the phosphocalcic balance of the newborn, calcemia at 111 mg/l; phosphorus at 53 mg/l; hypovitaminosis D at 17.73 ng/ml; Parathormone at 46 pg/ml; Alkaline phosphatase at 117u/L. the heart, renal and transfontanelle ultrasound realized in this patient not revealing an anomaly The genetic study not made.

Patient was put on a bisphosphonate (zoledronate) at a dose of 0.025 mg/kg/day to be renewed every 6 months.The evolution was marked by the recovery of all the initial fractures, the appearance of two new fractures, and the persistence of the pain. The follow up is one year

Discussion:

Osteogenesis imperfect is a rare disease characterized by bone fragility and osteopenia. It affects one new born out off 25 to 50,000 births, with no gender or race predominance and no preferential geographical distribution (1).

OI is one of a group of genetic and inherited conditions characterized by abnormal collagen type 1 synthesis leading to reduced production and abnormal collagen. These two genes COL1 A1 and COL1 A2 code respectively for the synthesis of the a1 and a2 chains of type I collagen. These mutations are mostly dominant (2).

The clinical picture combines skeletal signs (fractures in the absence of trauma and bone deformities) and inconstant extra-skeletal signs (blue sclera, dentinogenesis imperfecta, ligament hyperlaxity, persistence of arterial duct)(3).

Our patient presented with type III (4) OI features. Nephrocalcinos is secondary to hypercalciuria, linked to bone hyper remodelling, is usual. Hydrocephalus exists in 20 to 33% of cases, often asymptomatic, it does not require any treatment (5).

From a therapeutic point of view, it is a multidisciplinary approach. Regarding medical treatment, the use of Bisphosphonates has revolutionized the management of this condition. They potently inhibit bone resorption mainly by preventing the action of osteoclasts (6).

The course of OI depends on the type and severity of the clinical expression. A long side the regressive types I and IV forms of SILLENCE, which are rare forms with a favorable evolution, there are severe forms, types II b and III, which are the most frequent, often fatal between 3 months and 3 years (7).

Conclusion:

Osteogenesis imperfect is a rare and potentially serious genetic disease. The phenotype is very heterogeneous. The need for ultrasound monitoring of pregnancies in search of antenatal forms is essential in sick parturients or with sick ascendants or collaterals. Medical treatment with bisphosphonates has become an integral part in the management of this disease.

Conflict Of Interest statement :
We declare that we have no conflict of interest .

References:

(1)Silence DO, Senn A and Danks DM. Genetic heterogeneity in osteogenesis imperfecta. J Med Genet. 1979; 16(2):101-16. PubMed | Google Scholar 3.
(2)Forin V. Osteogenesis imperfecta. Orphanet Encyclopedia. June 2007 www.orpha.net/data/ patho/Pro/fr/OsteogenesisImparfaite.Pdf. Accessed 2007-11-12. PubMed | Google Scholar.
(3)Cissé R, Tapsoba TL, Lankoandé J et al. Imperfectosteogenesis apropos of a case at the Yalgado Ouédraogo National Hospital Center in Ouagadougou (Burkina Faso). Med Afr Noire. 1998;45(5):335-9. PubMed | Google Scholar.
(4)Ipach I, Kluba T, Wolf P et al. The influence of ibandronate treatment on bonedensity and biochemicalbone markers in patients with osteogenesis imperfect. OrthopRev (Pavia). 2012; 4(3):29. PubMed | Google Scholar.
(5) Chinas A, Petersen DJ, Schranck FW, et al. Hypercalciuria in childrens everely affected with osteogenesis imperfecta. J Pediatr 1991;119:51-7.
(6)Younes M, Touzi M, Mahmoud I et al. Lobstein's disease. A propos of five cases. Journal of rheumatism. 2006; 73(10-11):1135. PubMed | Google Scholar.
(7)Kaux JF, Le Goff C, Debray FG et al. Osteogenesis imperfecta. Rev Med Liege. 2009; 64(1): 9-13. PubMed | Google Scholar.