



An Aborted Fetus in the Second Trimester of Pregnancy due Osteogenesis Imperfecta (Diagnosed After Miscarriage): A Case Report

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Abstract

Background: Osteogenesis imperfecta (OI) is a group of disorders characterized by osteopenia, fractures, short stature, bone deformity, blue sclera and hyper laxity of ligaments and skin, with a prevalence 6-7 per 100 000 individuals.

There are four main kinds of OI with type II being the most severe; it is inherited in an autosomal recessive pattern with prenatally lethally deforming OI with multiple congenital fractures, micromelia and severe lung disease. OI could be caused by mutations in COL1A1 (Collagen Type 1, Alpha 1) or COL1A2 (Collagen Type 1, Alpha 2) with pathogenesis related to quantitative or qualitative abnormalities of type I collagen. Prenatal diagnosis for at risk pregnancies by fetal ultrasonography in the early 2nd Trimester is possible and it enables care. Case presentation: a twenty-year old Syrian woman reported that she could no longer feel her fetal movement during the second trimester of her first pregnancy, no fetal pulse was registered on fetal ECG (electrocardiogram). The absence of the fetus heart activity led to abortion and stillbirth due to respiratory failure. The aborted fetus had multiple bone fractures and severe limb malformations, blue sclera as well as clubfoot. All of the above led to clinically diagnosing Osteogenesis imperfecta type II. The patient did not have a family history of similar cases.

Conclusion: Osteogenesis imperfecta (OI) is a rare inherited bone disease, with many different types ranging from perinatal lethality to individuals with severe skeletal deformities and multiple fractures to nearly asymptomatic individuals with mild predisposition to fractures and normal life span. There are many difficulties facing healthcare professionals in diagnosing OI over other skeletal dysplasia. OI type II is a severe form with early fatal skeletal defects, thus early prenatal sonographic diagnosis is possible. It is im-

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Keywords: Osteogenesis imperfecta type II; aborted fetus; Diagnosis after miscarriage; Case report.



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portant to recognize that it can be difficult to distinguish between severe OI and other lethal skeletal dysplasias such as campomelic dysplasia or thanatophoric dysplasia. The primary tool for prenatal screening and diagnosis of skeletal dysplasias is ultrasonography, but a precise diagnosis cannot be made in many times. Infants with OI type IIA have very short curved limbs, long bones fractures, small chest and blue or gray sclera.

Background

Osteogenesis imperfecta (OI) is a group of disorders characterized by osteopenia, fractures, short statures, bone deformity, blue sclera and hyper laxity of ligaments and skin [1,2]. Its prevalence is about 6-7 per 100 000 individuals [3]. Severity of OI varies from type to type, ranging from perinatal lethality to individuals with severe skeletal deformities and multiple fractures to nearly asymptomatic individuals with mild predisposition to fractures and normal life span [2]. Various researchers tried to describe and classify this disorder into various types: The most acceptable classification was by Silience Et al., [4] in 1979, in which they grouped the disorder into four types: Type I; which is dominantly inherited with blue sclera, type II; is inherited in an autosomal recessive pattern with prenatally lethally deforming OI with multiple congenital fractures, micromelia and severe lung disease, Type III; is a progressively deforming OI with normal sclera and Type IV; is a dominantly inherited OI with normal sclera. All the different types of OI could be caused by mutations in COL1A1 or COL1A2 in an autosomal dominant pattern with pathogenesis related to quantitative or qualitative abnormalities of type I collagen. It consists of a heterogeneous group of skeletal dysplasia characterized by clinical variability and genetic polymorphism [5]. OI type II (OMIM (Online Mendelian Inheritance in Man) 166210) can be detected using ultrasound after 14 weeks of gestation with findings such as broad, crumpled and shortened limbs, thin ribs, angulation or fractures of long bones, abnormal fractures, and clinical findings of micromelia of long bones, beaded ribs and severe bone deformity at birth [6]. Prenatal diagnosis for at risk pregnancies by fetal ultrasonography in the early 2ndTrimester is possible and it enables care [7,8].

Case presentation

We report a case of a twenty-year old Syrian woman who reported that she could no longer feel her fetal movement during the second trimester of her first pregnancy. Fetal ECG showed no signs of the fetus heart activity, and no fetal pulse was registered. The absence of the fetus heart activity led to abortion and stillbirth due to respiratory failure. The patient used to have ultrasound tests to monitor her pregnancy, and they did not show any abnormalities, she did not have any other medical tests or medications during pregnancy. The aborted fetus had multiple bone fractures and severe limb malformations, blue sclera as well as clubfoot **figures (1-3)**. The fetus had a normal spinal cord as well as normal chest wall, heart, lungs, kidneys, liver, spleen and placenta. The aborted fetus was sent to the pathology lab, and the examination showed it was a male embryo, with a crown-rump length of 12cm and a head circumference of 2cm. There were no signs of a TORCH infection nor a hydatid form Mole or choriocarcinoma, all of the above led to clinically diagnosing Osteogenesis imperfecta type II. The patient did not have a family history of similar cases.



Figure 1: Pictures of the aborted fetus.

Discussion

Osteogenesis imperfecta (OI) is a rare inherited bone disease caused by defects in type I collagen synthesis secondary to mutations in type I collagen genes. Its prevalence is about 6-7 per 100 000 individuals [3]. There are many difficulties facing healthcare professionals in diagnosing OI over other skeletal dysplasia.

Severity of OI varies from type to type, ranging from perinatal lethality to individuals with severe skeletal deformities and multiple fractures to nearly asymptomatic individuals with mild predisposition to fractures and normal life span [2]. OI type II is a severe form with early fatal skeletal defects, thus early prenatal sonographic diagnosis is possible. In fact, early prenatal diagnosis of OI type II is possible. Authors reported early prenatal diagnosis of type (II) OI during the first trimester using transvaginal sonography by detecting shortened curved long bones, multiple fractures and hypo echogenicity of the skeleton [9].

It is important to recognize that it can be difficult to distinguish between severe OI and other lethal skeletal dysplasias such as campomelic dysplasia or thanatophoric dysplasia. The primary tool for prenatal screening and diagnosis of skeletal dysplasias is ultrasonography, but a precise diagnosis cannot be made in many times. Infants with OI type IIA have very short curved limbs, long bones fractures, small chest and blue or gray sclera. Respiratory and swallowing problems are common. Macrocephaly may be present, microcephaly is rarely present. There are many characteristic radiologic features for OI which includes absent or limited mineralization; flat vertebral bodies; broad femurs and broad short ribs [3]. The outcome of type IIA is poor with death in the perinatal period. The lethal form of OI is frequently associated with severe respiratory insufficiency secondary to pulmonary hypoplasia (10). The diagnosis of OI type II in our patient was based on the Clinical features of multiple fractures at birth, severe limb malformations, blue sclera as well as clubfoot. There was no positive family history of OI or other skeletal malformations, it was noted that sporadic cases were most likely in type II [4]. Akiode et al., in Sagamu [11] and Akinola et al., in Lagos [12], in Western Nigeria, both reported a single and two cases respectively of female babies based on clinical findings of multiple fractures, blue sclera at birth and

radiological evidence of defective Ossification of the skull. Prenatal diagnosis by fetal ultrasound is possible [7], but no Abnormality was detected in our case prenatally nor in the first case earlier reported in Nigeria, while the other report did not state the ultrasonography findings. The fractures might have occurred during delivery in both cases since prenatal diagnosis is mainly based on the presence of fractures. Fractures can occur even on minimal stress, but there is no empiric data to prove that caesarean section will improve the outcome over vaginal birth even when diagnosed prenatally [13].

This is the first recorded case of type II Osteogenesis imperfecta in Syria.

Conclusion

Osteogenesis imperfecta type II is a rare inherited bone disease, this study proves its existence in Syrian ethnicity, and it might be discovered during the second trimester of pregnancy without having any previous family history nor having any ultrasound finding during pregnancy.

Declarations

Ethics Approval and consent to participate

Not applicable

Consent to publication

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images.

Availability of data and materials

Not applicable.

Competing interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

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Author's contribution

HIJ was the doctor responsible of diagnosing and managing the case. NA, NAA, HH were responsible of writing the first draft of the paper and doing part of the literature review. MSM was responsible of editing, proofreading and writing the final form of the paper. All authors read and approved the final manuscript.

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