

Second-trimester diagnosis of osteogenesis imperfecta associated with schizencephaly by sonography

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ABSTRACT

Osteogenesis imperfecta is a congenital connective tissue disorder characterized with multiple bone fractures, short limbs, membranous calvarium with wormian bones, and sometimes blue sclerae. Osteogenesis imperfecta is rarely accompanied by other major malformations. Although associations with microcephaly, congenital heart defects or anencephaly have been reported previously, association with schizencephaly was not found on literature review. We report a case of osteogenesis imperfecta associated with schizencephaly diagnosed at 21 weeks of gestation using 2-dimensional ultrasound. The present case shows that prenatal ultrasonographic examination is a very important tool to detect such intrauterine abnormalities in which, management of pregnancy would be changed significantly compared to normal pregnancies.

Saudi Med J 2007; Vol. 28 (8): 1289-1290

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Received 2nd July 2006. Accepted 11th November 2006.

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Osteogenesis imperfecta (OI) is a connective tissue disorder with a variable expression resulting in multiple fractures, short limbs, membranous calvarium with wormian bones, and sometimes blue sclerae.¹ The OI is an autosomal disorder related mainly, but not exclusively, to mutations in the collagen type 1 gene. The most severe form of the disease, referred to as OI congenita or type II, is lethal, and thus usually appears as the result of a sporadic new mutation. It is rarely accompanied by other major malformations, although associations with microcephaly, congenital heart defects

or anencephaly have been reported previously.¹⁻³ Our objective is to report for the first time, a case of OI associated with schizencephaly diagnosed at 21 weeks of gestation with the use of conventional 2-dimensional ultrasound (2-D ultrasound).

Case Report. A 29-year-old woman, gravida 3, para 2, was referred for routine obstetric ultrasonographic control at 21-weeks of the gestation. Ultrasonographic examination (Applio80, Toshiba, Japan) using a 3.5-MHz transabdominal probe, showed limb abnormalities associated with an abnormal shaped lateral ventricle, highly suggestive of osteogenesis imperfecta. The appearance of the fetus' head on ultrasonographic examination was relevant with a unilateral close loop schizencephaly. Bilaterally, upper, and lower extremities were abnormally placed horizontally across the anterior part of the abdomen. Deformities in the femur, humerus, radius, and ulna were suggestive of prenatal fractures (**Figure 1**). The pregnancy was terminated at 21 weeks of gestation, with the permission of both parents. Postmortem examination confirmed our prenatal diagnosis with the posterior defect of the skull, associated with short crumpled long bones, and beaded ribs. Histopathological bone examination showed a normal cartilage growth plate with unremarkable resting, proliferating, and hypertrophic chondrocyte zones. The main pathology in the bones was severe deficiency of the trabecular, cortical ossification with narrow delicate columns of calcified cartilage matrix, associated with numerous fractures, and callus formation.

Discussion. We report a case of OI associated with schizencephaly diagnosed at 21 weeks of gestation using 2-D ultrasound. Prenatal diagnosis of isolated OI by 2-D ultrasound in the first trimester has been reported before,^{4,5} however, its' association with neurological abnormalities has rarely been reported.^{1,2} Bronshtein et al,³ reported a case of OI diagnosed at 14 weeks of gestation using conventional ultrasonography. However, anencephaly in this case was diagnosed during postmortem pathological examination. The 2-D

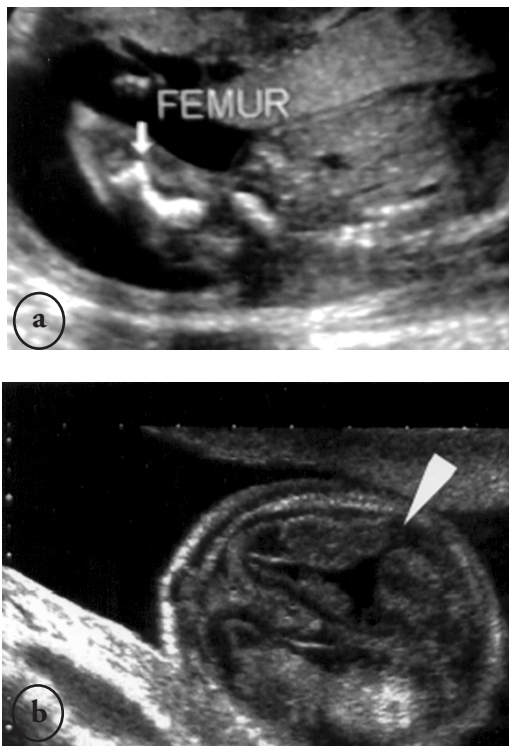


Figure 1 - Ultrasound imaging of the fetus showing a) curving of the femur (arrow), and b) the small schizencephalic cleft (arrowhead). Note the cleft extending from the ventricle to the pial surface.

ultrasound images are easier to interpret in cases with relevant family history. Otherwise, global view provided by 3-dimensional ultrasound (3-D) might be helpful for the diagnosis. The bone mode of 3-D ultrasound can show the bone fractures, and deformities, as well as postmortem x-ray examination. It is possible to rotate the volume image into a standard or oblique anatomic orientation, as well as to record, and manipulate data in the 3-D ultrasound examination.⁶⁻⁸ Recent studies have suggested the potential advantage of using 3-D ultrasound to improve the accuracy of prenatal diagnosis of skeletal dysplasias. However, the use of 3-D ultrasound was not helpful in the present case due to fact that the images of all the bony structures of the fetus were overlapped, and interpretation was difficult.

Prenatal 3-D helical computer tomography has been successfully used in the prenatal diagnosis of other fetal malformations, and additionally, ultrafast MRI is of great value in complementing conventional 2-D ultrasound for the diagnosis, and subclassification of OI in the fetus.^{9,10}

In conclusion, the preterm diagnosis of such abnormalities is very important during routine obstetric examination of a pregnant woman, since management of pregnancy changes significantly as in the present case, which was terminated.

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