Prenatal diagnosis of osteogenesis imperfecta type II
Kara O, Kirbas A, Daglar Hk, Timur H, Uygur D, Caglar T
Zekai Tahir Burak Women's Health Education and Research Hospital, Ankara, Turkey

Objective
To characterize sonographic features of osteogenesis imperfecta type ii diagnosed prenatally.

Methods
A 29 year-old, g2 p1 women has been referred to our perinatology clinic at 20th weeks with findings of hypomineralization of calvarium and thoracic hypoplasia for further evaluation. She did not have any medical history unremarkable and has a 3 healthy years old child. At ultrasonography, biparietal diameter, head circumference and abdominal circumference were consistent with 19th weeks pregnancy. Both femur length were below 5th percentile and amniotic fluid was normal. Skull hypo mineralization, small thorax, bowing, angulation, fractures of femur and humerus (figure1), club foot (figure 2) were observed suggesting a lethal form of oi type ii. Club hands (figure 3) were noticed due to fractures. Cranial bones and thorax were easily deformed when pressure was applied with transducer (figure 4 and 5). Fetal echocardiography was normal. The family was informed about the prognosis of the disease and they chose to terminate the pregnancy. Male fetus was delivered at 21 weeks with double-balloon catheter application.

Results
Postnatal skeletal x-ray confirmed the diagnosis of oi type ii ( figure 6).

Conclusion
Oi is seen rarely. The incidence is about 1/25, 000 to 30, 000 in obstetric patients. Oi has 4 major types. Type 2 which is a lethal form of oi gives intrauterine signs. It can be diagnosed while performing routine ultrasonography. The disorder is characterized by early prenatal onset of severe bone shortening and bowing due to multiple fractures affecting all long bones and ribs and poor mineralization of the skull. Death occurs either prenatally or shortly after birth because of respiratory failure. A case of lethal osteogenesis imperfecta detected prenatally has been reported.