A case of 3D diagnosis of Apert syndrome
Karahanoglu E, Akpinar F, Esin S, Yalvac ES, Kandemir NO
Etlik Zubeyde Hanim Womens Health and Education Hospital, Ankara, Turkey

Objective
To evaluated the 3D ultrasound for diagnosis of Apert Syndrome.

Methods
A 32 year old, gravida 2 para 1, at 21 weeks gestation was referred to the outpatient clinic for routine prenatal care. A 2-dimensional ultrasonography reveals frontal bossing and suspected closure of coronal suture with exophthalmos. In neurosonography no specific finding was detected. In 3D ultrasound examination exophthalmos, midface hypoplasia low set ear, widening of sagittal and metopic suture were detected (Figure 1A-B). While evaluating the hand although pollex was separated from index finger, there is complete syndactyly between 2th, 3th and 4th digit and partial syndactyly between 4th and 5th digit (Figure 1C). No other anomaly was detected. At 24 week preterm birth ensued and a 700 gr fetus was delivered by vaginally. The fetus died at 4 hour after delivery due to extreme prematurity. Post mortem finding confirmed prenatal diagnosis. Frontal bossing, midface hypoplasia, complete syndactyly of 2th, 3th, 4th digit (Figure 2).

Results
Second trimester Apert syndrome is challenging diagnosis. However evolving of ultrasound technologies and 3D ultrasonography, it becomes easier (5). Most striking feature of Apert syndrome is frontal bossing. However there are to many conditions that may cause frontal bossing. First step after detection of frontal bossing should be the evaluation of intra cranial anatomy and morphology of cranial sutures (coronal, sagittal and metopic). Objective and subjective assessment of width of sagittal, metopic, and coronal sutures has been defined (6, 7). Appropriate 3D images of the suture can be obtained by adjusting the ultrasound probe parallel to plane that passes the through suture that is going to be evaluated (7). During the examination if any of the sutures closed with reciprocal widening of other sutures, craniosynostosis syndromes should be suspected. Apert syndrome is the one that is coronal sutures closed with reciprocal widening of metopic and sagittal suture. Coronal suture can be evaluated easily with 2D sonography (Figure 3). However evaluation of sagittal and metopic suture are more challenging with 2D ultrasound. 3D dimensional evaluation of these stures are more easy. Midfacial hypoplasia and low set ear can easily be detected by 3D ultrasound. Hand and finger are important aspect of craniosynostosis syndromes for differential diagnosis (Phiffer, Apert, Carpenter). Although preaxial polydactyl is pathognomonic for Phiffer syndrome also syndactyly can bee seen. However in Phiffer syndrome diagnostic skull shape is clover leaf. Although polydactyl can be detected 2D ultrasound, assessment of syndactyly sometimes difficult. A 3D ultrasound helpful for evaluation syndactyly and webs between fingers. Pathognomonic hand finding of Apert syndrome (Mitten hand, spade hand deformity) can easily be detected by 3D ultrasound. Apert syndrome sometimes associated with other system abnormalities like diaphragma hernia. Careful anatomic survey should be done to detect other abnormalities.

Conclusion
Although it hasn’t been shown superiority of 3D ultrasound on 2D ultrasound for diagnosis of congenital anomalies and syndromes, in some condition diagnosis can be done easily with the aid of 3D technique.