A case of prenatal diagnosis of Apert syndrome with postmortem computed tomography findings
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Objective
Case Report: Prenatal 2D/3D Ultrasonographic findings and postnatal Computed Tomography findings of Apert Syndrome.

Methods
Apert syndrome (AS), also called Acrocephalosyndactyly type I, is an autosomal dominant disorder with the prevalence of 1 in 80,000 live births. Mutations in the fibroblast growth factor receptor 2 gene (FGFR2) are responsible in AS. Majority of the cases are sporadic. Coronal craniosynostosis, midfacial hypoplasia, and symmetric bony syndactyly of the hands and feet are most commonly detected findings of AS both in prenatal and postnatal diagnosis. A prenatally diagnosed Apert Syndrome is presented with comparison of Three-Dimensional Ultrasonography and Post-mortem Computed Tomography findings.

Results
A 34-year-old woman, gravida 2, parity 1 was referred at 23 weeks of gestation with the suspicious of mild ventriculomegaly. She and her family had both unremarkable medical histories. The anomaly scan showed irregular head shape, acrocephaly, prominent forehead, bilateral mild ventriculomegaly (right ventricle was 11 mm, left ventricle was 12 mm), hypertelorism, and midfacial hypoplasia, with a depressed nasal bridge (Figure 1a-b-c, grey scale ultrasound, 3-dimensional surface mode and postmortem photography, respectively). Syndactyly of the digits of both hands and feet were detected during the extremity examination on grey scale ultrasound and polyhydramnios was an additional finding of the case. A 3-dimensional (3D) ultrasound examination (Voluson E6, GE Healthcare, Milwaukee, Wis) with surface rendering and maximum mode was performed. Facial profile with depressed nasal bridge and prominent forehead was demonstrated with surface rendering mode on Figure 1b. Examination of the fetal head with maximum mode revealed synostosis of both coronal sutures causing frontal bossing. Because of early closure of coronal sutures (Figure 2a), wide metopic suture was also detected on 3-dimentional ultrasound examination. The diagnosis of Apert Syndrome was suspected based on the findings mentioned above. The patient was counselled with a genetic expert and consented for amniocentesis. This was performed and screened for most common mutations of Apert Syndrome. The diagnosis was confirmed by the identification of the heterozygote mutation P253R, which is the most common mutation of Apert Syndrome. The patient was counselled and the pregnancy was terminated at maternal request. Prenatal ultrasound findings were demonstrated on postmortem images on Figure 1c and 3a-b. Postmortem computed Tomography of the fetus also showed wide metopic suture and early closing of both coronal sutures on Figure 2b. Osseous syndactyly of hands and feet were demonstrated with Computed Tomography imaging in Figure 3 c-d.

Conclusion
Craniosynostosis is characterized by premature fusion of cranial sutures. Demonstration of premature closure of coronal suture should rise suspicion of Apert Syndrome. 2D Ultrasound have an important role both suspicion and diagnosis of Apert Syndrome. On the other hand complementary use of 3D ultrasound could be useful to demonstrate characteristic features of Apert Syndrome especially premature closure of sutures. Actual diagnosis should be based on demonstration of specific genetic mutation. Due to the premature closure of the sutures approximately one half of affected individuals are mentally retarded excluding functional handicap due to hands feet anomalies. Termination of pregnancy may be an option for families.