Discussion: Numerous studies have investigated over 400 cases involving fetuses with VACTERL syndrome and partial caudal regression syndrome. The reported incidence rates of various abnormalities were as follows: spine abnormalities ranged from 60% to 80%, anal atresia from 55% to 90%, tracheoesophageal abnormalities from 50% to 80%, cardiac malformations from 40% to 80%, limb malformations from 40% to 50%, and kidney malformations from 50% to 80%. In conclusion, prenatal ultrasonic diagnosis plays an indispensable role in identifying VACTERL syndrome and partial caudal regression syndrome, offering valuable guidance for obstetric treatment. Its clinical implementation is highly warranted.[1]

Conclusion: VACTERL syndrome is typically defined by the presence of at least three of the following congenital malformations: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and limb abnormalities. In addition to these main component features, patients may have other congenital anomalies. It is possible to suspect the diagnosis by antenatal ultrasonography. It is important to detect, suspect and investigate further when the first signs appear on a routine ultrasound scan.

Keywords: Cardiac malformation, limb anomaly, vertebral defect, VACTERL

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PP-19 Prenatal diagnosis of isolated bilateral congenital cataract

Büsra Berfin Polat[1], Rauf Melekoglu[1]
[1]Inonu University Faculty of Medicine, Department of Obstetrics and Gynecology, Malatya, Türkiye

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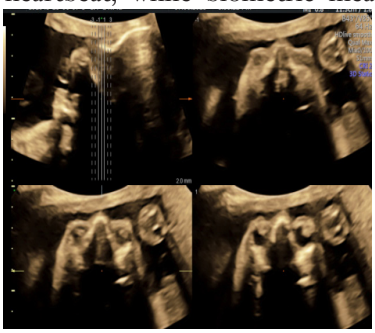
Objective: Congenital cataract stands as an orbital anomaly characterized by lens opacity, manifesting unilaterally or bilaterally, with an incidence of 1 in 10,000 births. Genetic syndromes are identifiable in roughly 10% of cases; nonetheless, congenital infections have been discerned in approximately 30% of instances. In the presence of unilateral or bilateral congenital cataracts, a comprehensive ultrasound evaluation, encompassing neurosonography, invasive prenatal diagnostic testing for karyotyping, and maternal TORCH panel analysis for fetal infections, becomes imperative. Furthermore, maternal utilization of pharmaceuticals (including steroids), radiation exposure, or exposure to potential teratogens, along with any underlying metabolic conditions, should be meticulously investigated for their potential etiological implications. In the context of this report, our objective is to present a case involving the prenatal diagnosis of an isolated instance of bilateral congenital cataract..

Case: A 36-year-old patient, gravida 6, parity 4, at 24 weeks and 4 days of gestation, was admitted to the prenatal diagnosis and treatment unit at our clinic for fetal anomaly screening. The patient had a history of pregestational diabetes mellitus and was meticulously monitored. Ultrasonographic examination confirmed a positive fetal heartbeat, while biometric measurements aligned with the gestational age, and amniotic fluid volume remained within normal ranges. Notable findings in fetal facial assessment included increased opacity observed in bilateral lenses within the coronal plane and orbital assessment (figure 1). Fetal neurosonography and abdominal evaluation yielded no evidence of periventricular calcification, hepatic/splenic calcification, or hyperechogenic bowel. Given the patient's history of congenital cataract in a previous child, an isolated diagnosis of congenital cataract was considered. Upon fetal cardiac evaluation, borderline myocardial hypertrophy and perimembranous ventricular septal defect (VSD) were identified. At this juncture, the option of invasive prenatal diagnostic testing or cell-free fetal DNA analysis in maternal blood was presented. Maternal blood TORCH panel analysis yielded no abnormal findings, and in light of the patient's preferences, she chose not to undergo invasive prenatal diagnostic testing. Throughout this process, the family was provided with comprehensive information regarding potential neonatal complications.



Discussion: Genetic testing and prenatal ultrasound have become primary methods of diagnosing congenital cataracts. The analysis by Yue Qin et al found a total of 41 cases of congenital cataracts diagnosed prenatally among 788,751 women who underwent the mid-tirmester fetal anatomical scan. Based on sonographic features, 16/41 (39.0%) had an intense echogenic pattern, 15/41 (36.6%) had a hyperechogenic spot, and 10/41 (24.4%) had a "double ring" sign. 17/41 (41.5%) were isolated and 24/41 (58.5%) had associated intraocular and extraocular findings. Microphthalmia, cardiac abnormalities and central nervous system abnormalities were the most common associated abnormalities. Potential etiology regarding the disease, 6 cases had known family history of congenital cataracts, 4 cases had confirmed congenital rubella infection, and 2 cases had aneuploidy.[3]

Conclusion: Congenital cataract comprises 7.4-15.5% of all cases of childhood blindness. Detecting congenital cataract during the prenatal phase and discerning any potential associations with other pathologies are critical measures for timely intervention and treatment of conditions that could lead to morbidity, mortality, or vision impairment. Emphasizing the significance of precise prenatal diagnosis of congenital cataract, this process greatly aids patients and their families by enabling personalized genetic counseling. Therefore, meticulous sonographic evaluations, taking into consideration the patient's specific risk factors, are paramount to ensuring comprehensive care.

Keywords: Congenital cataract, lens, opacity, ultrasound

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PP-20 Prenatal sonographic diagnosis of Ebstein anomaly

Büsra Berfin Polat[1], Rauf Melekoglu[1]
[1]Inonu University Faculty of Medicine, Department of Obstetrics and Gynecology, Malatya, Türkiye
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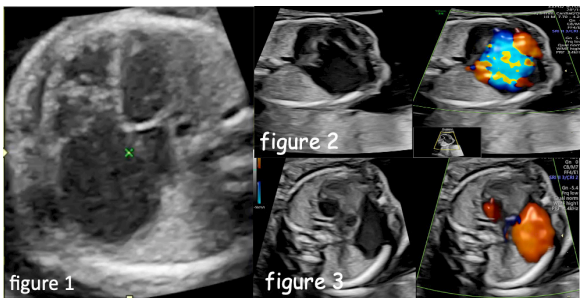
Objective: Ebstein's anomaly, which accounts for 1% of congenital cardiac anomalies, is a rare congenital cardiac anomaly with a prevalence of 0.3 -0.5% and an incidence of 1 in 20000 live births. Pathologically, it is characterized by abnormal positioning of the septal and/or posterior leaflet of the tricuspid valve towards the right ventricular apex. The right ventricular area is reduced and the infundibulum is obstructed by the anterior leaflet secondary to atrialization of the portion of the right ventricle between the level of the true annulus and the level of the false annulus. Antenatal diagnosis is usually made by fetal echocardiography. The main findings of this pathology seen on fetal echocardiography are apical displacement of the tricuspid valve and consequent atrialization of the right ventricle, right ventricular failure, cardiomegaly, tricuspid valve insufficiency, ventricular septal defect and atrial septal defect. Antenatal diagnosis is very important because it is a rare congenital cardiac anomaly and mortality is significantly reduced with appropriate neonatal management. In this report, we aimed to present the prenatal diagnosis of a very rare case of Ebstein anomaly.

Case: A 35-year-old patient with gravida 3, parity 2, 22 weeks and 1 day gestation according to the last menstrual period was referred to the prenatal diagnosis and treatment unit of our clinic due to suspicion of cardiac anomaly. In the ultrasonographic examination of the patient, fetal heartbeat was positive, amniotic fluid volume was high, and biometric measurements were compatible with the gestational week. Fetal cardiac examination revealed severely dilated right atrium (figure 1), severe tricuspid regurgitation (tricuspid regurgitation) (figure 2) and severe pulmonary hypoplasia (figure 3). The family was informed in detail about the possible poor fetal/neonatal prognosis of the fetus with Ebstein's anomaly and invasive prenatal diagnosis was offered as an option. Fetal echocardiography was planned for the patient who did not want to undergo invasive prenatal diagnostic testing. Pregnancy follow-up and delivery were recommended to be performed in a tertiary care center with pediatric cardiovascular surgery facilities.



Discussion: In the multicenter fetal cohort with Ebstein's anomaly and tricuspid valve dysplasia reported by Freud et al., perinatal mortality was found about 45%. This was considerably higher than other types of congenital heart disease in the current era. However, a greater proportion of fetuses survived to birth compared with previous series of single-center Ebstein's anomaly and tricuspid valve dysplasia in the last few decades. This can be attributed to a combination of factors, including a lower rate of termination of pregnancy and an increased likelihood that progress in prenatal diagnosis will identify less severely affected fetuses.[3]

Conclusion: The symptoms of Ebstein's anomaly, a very rare congenital cardiac anomaly, vary depending on the degree of tricuspid regurgitation, whether ventricular function is impaired, whether the infundibulum is obstructed, and whether fetal arrhythmia is present. In this case, which we suspected on the basis of dilatation of the right atrium and the accompanying cardiac findings, the prognosis depends on the severity of the malformation. Severe cases may result in intrauterine death. Accordingly, in cases where surgical treatment is necessary and intrauterine death does not occur, Ebstein anomaly should be considered in cases with abnormally located tricuspid valve, right atrial dilatation, pulmonary stenosis or functional atresia. Since the mortality rate in these cases is significantly reduced with appropriate prenatal and neonatal management, it is important to detect, suspect and investigate further when the first signs appear on a routine ultrasound scan.[4]