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

References

1. Rahi JS, Dezateux C and British Congenital Cataract Interest Group. Measuring and interpreting the incidence of congenital ocular anomalies: lessons from a national study of congenital cataract in the UK. Invest Ophthalmol Vis Sci 2001; 42: 1444–1448.
2. Yi J, Yun J, Li ZK, et al. Epidemiology and molecular genetics of congenital cataracts. Int J Ophthalmol2011; 4: 422–432.
3. Qin Y, Zhong X, Wen H, Zeng Q, Liao Y, Luo D, Liang M, Tang Y, Guo J, Cao H, Yang S, Tian X, Luo G, Li S. Prenatal Diagnosis of Congenital Cataract: Sonographic Features and Perinatal Outcome in 41 Cases. Ultraschall Med. 2022 Dec;43(6):e125-e134. English.

PP-20 Prenatal sonographic diagnosis of Ebstein anomaly

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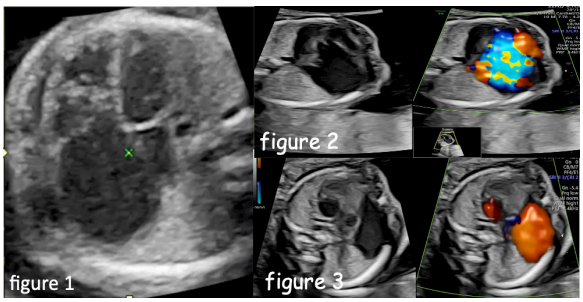
Objective: Ebstein's anomaly, which accounts for 1% of congenital cardiac anomalies, is a rare congenital cardiac anomaly with a prevalence of 0.3 -0.5% and an incidence of 1 in 20000 live births. Pathologically, it is characterized by abnormal positioning of the septal and/or posterior leaflet of the 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]

Keywords: Ebstein,fetal echo, tricuspid valve

References

1. McIntosh N, Chitayat D, Bardanis M, Fouron JC. Ebstein anomaly: report of a familial occurrence and prenatal diagnosis. Am J Med Genet. 1992 Feb 1;42(3):307-9.
2. Downing KF, Riehle-Colarusso T, Gilboa SM, Lin AE, Oster ME, Tinker SC, Farr SL; National Birth Defects Prevention Study. Potential risk factors for Ebstein anomaly, National Birth Defects Prevention Study, 1997-2011. Cardiol Young. 2019 Jun;29(6):819-827.
3. Freud LR, Escobar-Diaz MC, Kalish BT, Komarlu R, Puchalski MD, Jaeggi ET, Szwast AL, Freire G, Levasseur SM, Kavanaugh-McHugh A, Michelfelder EC, Moon-Grady AJ, Donofrio MT, Howley LW, Tierney ES, Cuneo BF, Morris SA et al. Outcomes and Predictors of Perinatal Mortality in Fetuses With Ebstein Anomaly or Tricuspid Valve Dysplasia in the Current Era: A Multicenter Study. Circulation. 2015 Aug 11;132(6):481-9.
4. Yuan SM. Ebstein's Anomaly: Genetics, Clinical Manifestations, and Management. Pediatr Neonatol. 2017 Jun;58(3):211-215.

PP-21 Noonan syndrome presented with cystic hygroma in the first trimester

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Objective: It is aimed to point out the importance of advanced genetic testing in fetuses with cystic hygroma.

Case: A pregnant woman aged 33 years, G1P0, 20 weeks and 5 days referred to us from an external center was evaluated. She had chorionic villus sampling because of a cystic hygroma seen in the first trimester sonographic screening. Fetal karyotype and microarray analysis results had been reported as normal so a genetic mutation panel had been studied for Noonan syndrome. Pathogenic missence variant was found in PTPN11 gene and pathogenic splice acceptor variant in MYBPC3 gene. The ultrasonographic findings were thought to be related to the heterozygous pathogenic variant detected in the PTPN11 gene. In the ultrasonographic evaluation of the fetus, it was observed that the nuchal fold was 8.7 mm, with septation and cystic appearance, and the lower poles of the fetal kidneys merged at the level of the aortic bifurcation. Fetal gender was female. Fetal echocardiography revealed subvalvular aortic stenosis. By reason of clinical findings and genetic examination results were thought to be compatible with the autosomal inherited Noonan syndrome, termination of pregnancy was offered to the family by the perinatology council. The family accepted the option of termination of pregnancy.

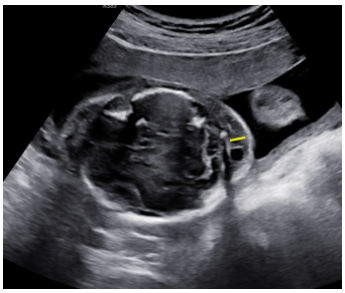

Fig 1. 8.7 mm nuchal fold with septation and cystic appearance

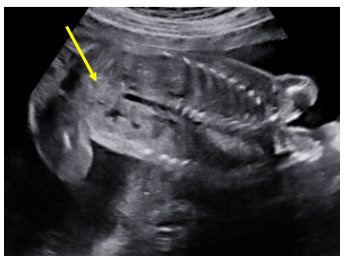

Fig 2. Lower poles of the fetal kidneys merges at the level of the aortic bifurcation

Discussion: Noonan syndrome is a common autosomal-dominant condition clinically and genetically heterogeneous and two-thirds of patients are the first affected person in their family due to a de novo pathogenic variant. Although so many gene mutations are described for this syndrome, approximately %50 of patients have a pathogenic variant in protein tyrosine phosphatase, nonreceptor type 11 (PTPN11).[1] Fetuses with Noonan syndrome may present with manifestations of disordered lymphatic development, which include most commonly increased nuchal translucency. Renal and valvular cardiac anomalies may represent, too.[2] Clinical features such as difficulties with feeding in early life; vision, hearing, and growth problems; specific learning difficulties may develop in the postnatal period.

Conclusion: Even if fetal karyotyping is normal in cystic hygroma cases detected in the first trimester screening, it should be kept in mind that genetic mutations that may accompany related syndromes may also be present.

Keywords: Noonan syndrome, cystic hygroma, PTPN11 gene mutation, prenatal genetic test.

References

1. Tartaglia M, Cordeddu V, Chang H, et al. Paternal germline origin and sex-ratio distortion in transmission of PTPN11 mutations in Noonan syndrome. Am J Hum Genet. 2004;75(3):492-497.
2. Croonen EA, Nillesen WM, Stuurman KE, et al. Prenatal diagnostic testing of the Noonan syndrome genes in fetuses with abnormal ultrasound findings. Eur J Hum Genet. 2013;21(9):936-942.