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.

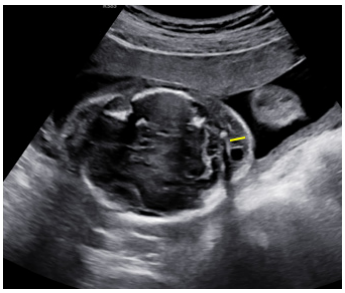

Fig 1. 8.7 mm nuchal fold with septation and cystic appearance

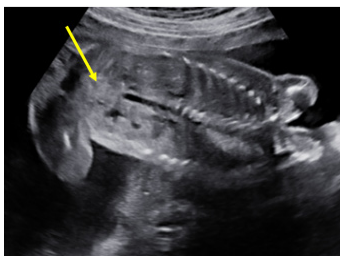

Fig 2. Lower poles of the fetal kidneys merges at the level of the aortic bifurcation

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.