17th World Congress in Fetal Medicine

Retroesophageal anomalous left brachiocephalic vein in a fetus - not just a normal variant

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Objective

To evaluate the significance of ultrasound finding of a retroesophageal anomalous left brachiocephalic vein in a fetus.

Methods

1. Report five consecutive prenatal cases of retroesophageal anomalous left brachiocephalic vein 2. Describe the congenital abnormalities that were associated with these cases and the fetal outcomes 3. Discuss the significance of association between these abnormalities and retroesophageal anomalous left brachiocephalic vein.

Results

Retroesophageal anomalous left brachiocephalic vein (REALBCV) is very rare and is highly associated with congenital heart diseases in postnatal cases. After our first prenatal diagnosis of REALBCV in 2015, we have identified four more cases since then. All of them were detected because of initial finding of dilated azygos vein draining into superior vena cava in the three vessels view during mid trimester fetal anomaly scan. Three had increased nuchal translucency (NT) ≥3 mm in first trimester and either increased nuchal fold (NF), nuchal oedema, cystic hydroma, dilated jugular lymphatic sacs (JLS) or generalised hydrops in second trimester. Genetic study for Noonan and related syndromes (NRS) showed pathogenic mutations in all the three cases, two in RAF1 and one in BRAF gene. One of these fetuses also had a small ventricular septal defect (VSD). All of the patient opted for termination of pregnancy. NT was <3 mm in two cases, one >95th percentile for gestational age but with low risk combined Down syndrome screening and the other normal. The one with increased NT also had a small VSD and both karyotyping and chromosomal microarray (CMA) showed de novo inverted duplication and deletion of chromosome 8p. The pregnancy was terminated. A retrospective NRS genetic study was negative. The one with normal NT was one of the dichorionic twin fetuses and REALBCV was an isolated finding in this fetus. Parents declined amniocentesis and postnatal genetic study for NRS was negative. The baby was found to have oesophageal atresia (EA) and trachea-oesophageal fistula (TEF) on day one after birth and operation was done. No other congenital abnormalities were identified. The association of NRS with three of the five cases of REALBCV is not coincidental. Although all the three cases of NRS had prenatal ultrasound features, one only had borderline NT of 3 mm and second trimester NF of 6 mm. Unlike those with frank nuchal oedema, cystic hygroma or dilated JLS, NRS may not be tested in these milder cases as the diagnostic yield is low. The additional finding of REALBCV may increase the specificity of ultrasound diagnosis of NRS and therefore, increase the diagnostic yield of genetic testing. In all the four cases with genetic abnormalities, NT was increased. Three of the four cases also had associated abnormalities other than soft markers. In the only case without genetic abnormalities, it was associated with a structural abnormality. In none of the cases in this series REALBCV was found in a normal fetus. It is worth noting that the associated abnormalities in three of the five cases were subtle, including small VSD, borderline increased NF and EA with TEF, which could easily be missed in prenatal scan. Therefore, discussion for amniocentesis may be necessary even when REALBCV is apparently isolated.

Conclusion

REALBCV in a fetus is highly associated with genetic and structural abnormalities. We advised looking for any dilated azygos vein during routine examination of the three vessels view in fetal anomaly scan and if found, detailed examination for any REALBCV should follow. Finding of a REALBCV should indicate a careful and meticulous search for any associated abnormalities, which can be subtle. Amniocentesis for karyotyping and CMA should be discussed especially if NT is increased or other abnormalities are detected. Genetic testing for NRS is strongly indicated if karyotyping and CMA are normal and increased NT is associated with ultrasound signs of lymphatic obstruction even with borderline second

trimester increased NF.