The frequency of 22q11 Micro Deletion Syndrome in Fetuses with Conotruncal Heart Disease

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Objectives

We sought to investigate the frequency of 22q11 microdeletion syndrome (22q11DS) in fetuses diagnosed with conotruncal heart disease (CTD) and to correlate 22q11DS with phenotypic CTD for prenatal parental counseling.

Methods:

• We retrospectively reviewed 30009 fetal echocardiograms from October 2010 to February 2018 in Beijing Anzhen Hospital. 744 fetuses were diagnosed as CTD in which 208 cases diagnosed were reviewed. Fetal umbilical cord tissue and parental peripheral blood specimens were performed to ascertain 22q11DS using low coverage whole genome sequencing (WGS) and whole exome sequencing (WES).

Results:

In this cohort, 32 (6.25%) fetuses with CTD were identified with 22q11DS. Peripheral blood samples were collected from 27 parents, 2 mothers and 3 fathers (18.51%) were detected with the same 22q11DS with their fetuses, and the others were de novo. The most common phenotypes of 22q11DS were interruption of aortic arch of Type B (IAA-B) (100%,5/5), posterior alignment ventricular septal defect with coarctation of aorta (paVSD/CoA) (20%,1/5), persistent truncus arteriosus (PTA) (33.33%,5/15), tetralogy of Fallot (TOF) (17.39%,12/69), pulmonary atresia with ventricular septal defect (PA/VSD) (16.67%, 6/36), and double outlet right ventricle (DORV) (8.3%, 3/36). The other kinds of CTD did not merge with 22q11DS. What’s more, the other minor phenotypes, right aortic arch (34.38%,11/32), arterial duct abnormality (31.25%,10/32) and mandibular enlargement (34.38%,11/32) were common in the fetuses with 22q11DS.

Conclusion

The frequency of 22q11DS is high in fetuses with CTD. Fetuses with some CTD (e.g IAA-B, paVSD/CoA, etc.) had higher frequency of association with 22q11DS, while the other kinds of CTD did not usually find 22q11DS. The data may aid prenatal diagnosis, prognosis and parental counseling of 22q11DS in CHD.