Objectives

To investigate the echocardiographic diagnosis for fetal ventricular non-compaction cardiomyopathy (VNCC) and to analyze the genetic associations with VNCC.

Methods:

• A total of 13 fetuses were diagnosed as VNCC from 30009 fetal echocardiographic studies in Beijing Anzhen Hospital from September 2010 to February 2018. Fetal umbilical cord tissue samples and the parental pewere collected after the families opted for termination of pregnancy, in addition to the peripheral blood collected from the parents. Whole exome sequencing (WES) and Sanger sequencing were performed using the biological specimens.

Results:

• In this cohort, 6 fetuses were diagnosed with biventricular, 5 with left ventricle, and 2 with right ventricle and gestational ages of the pregnant patients range from 24 to 37 years (mean, 28.60 ± 3.75 years). Among the 13 fetuses with VNCC, 11 cases were terminated after a detailed prognosis consultation during pregnancy. 1 fetus was born, 1 case lost follow-up. In addition, 6 cases were associated with congenital heart defects (CHD), and 4 cases with fetal arrhythmia, and the average cardiovascular profile score of fetuses was 7.08. There were VNCC mostly involved the apical and lateral wall of the LV. The non-compacted layer to compaction layer ratio of RV is more than LV. For genomic study, 1 case had LDB3 gene mutation (c. T964C / p. S322P), and 1 had PRKAG2 gene mutation (c.1592G>A / p. Arg531Gln), both of the two mutations were de novo. In addition, 3 cases came from one families were associated with uncertain significant mutation which inherited from their mothers.

Conclusion

Fetal VNCC can involve both the left and the right ventricles. Prenatal echocardiography combined with WGS could play an important role in the timely diagnosis, prognosis and parental counseling of patients with VNCC.