



Pattern of aneuploidies in fetuses with pleural effusion detected on antenatal ultrasound examination

Vaishali M, Reeth S, Radhakrishnan P
Bangalore Fetal Medicine centre, Bangalore, India

Objective

To determine the prevalence of chromosomal abnormalities in fetuses with prenatally diagnosed pleural effusions and to identify factors associated with increased risk of aneuploidy.

Methods

A retrospective analysis of singleton pregnancies found to have pleural effusion on antenatal scan at Bangalore Fetal Medicine Centre from January 2008 to July 2017 was performed. All the scans were performed according to standard Fetal Medicine Foundation (FMF) protocols by FMF certified operators for the study of fetal anatomy.

Results

There were 104 pregnancies in whom fetal pleural effusion was identified. These were divided into 3 groups, based on gestational age at diagnosis ie, first trimester [11-14 wks], second [15-27 wks] and third trimester [28-40 wks]. In the first trimester, 19/36 [52%] opted for fetal karyotyping. The prevalence of chromosomal abnormalities was 12/19 [63.2%]; 7/19 [36.8%] were normal. Out of the 12 aneuploidies, 6/12 [50%] had Turner Syndrome, 3/12 [25%] had Trisomy 21, 2/12 [16.6%] had Trisomy 18 and 1/12 [8.3%] had Trisomy 13. In second trimester, 28/48 [58%] opted for fetal karyotyping. The prevalence of chromosomal anomalies was 2/28 [8.2%] of which 1 was Trisomy 21 and 1 diagnosed to have mutation for Noonan syndrome, postnatally. In third trimester, 11/20 [55%] opted for fetal karyotyping and all were normal.

Conclusion

Chromosomal analysis is warranted after prenatal detection of fetal pleural effusion. The risk of aneuploidy is significantly greater with first trimester detection of pleural effusion and Turner Syndrome is the most frequent aneuploidy.