17th World Congress in Fetal Medicine

Effect of the presence of structural defects on fetuses with markers on aneuploidies at the 11-13 weeks' scan

Vaghasia D, Robert K, Acharya V, Radhakrishnan P
Bangalore Fetal Medicine Centre, Bangalore, India

Objective

To determine the effect of presence of structural defects on aneuploidies in fetuses with marker(s) at the 11+0 – 13+6 weeks' scan.

Methods

681/ 12085 (5. 6%) fetuses with at least one marker were analysed at the 11+0 – 13+6 weeks' scan for the presence of additional defects. All had completed outcomes with chromosomal and structural defects. All the scans were performed according to the unit protocol by Fetal Medicine Foundation (FMF) certified operators for the study of fetal anatomy, Nuchal Translucency, Nasal bone and Tricuspid Doppler. 3 markers, increased nuchal translucency (NT), absent nasal bone (ANB) and tricuspid regurgitation (TR), were studied in all the fetuses according to the guidelines as per the FMF.

Results

467 (68. 6%), 242 (35. 5%) and 88 (12. 9%) had increased NT, ANB and TR respectively. Of these 125 (26. 8%), 56 (23. 1%) and 33 (37. 5%) fetuses with increased NT, ANB and TR respectively had additional structural defects. 22/125 (17. 6%), 11/56 (19. 6%) and 8/33 (24. 2%) fetuses with fetal defects and increased NT, ANB and TR respectively had major aneuploidies. However, 38/ 342 (11. 1%), 26/186 (13. 9%) and 9/55 (16. 3%) fetuses without defects and increased NT, ANB and TR respectively had major aneuploidies. 595 (87. 4%), 73 (10. 7%) and 13 (1. 9%) had 1, 2 and 3 markers respectively. Of these, 117 (19. 7%), 37 (50. 7%) and 6 (46. 2%)%) fetuses with 1, 2 and 3 markers respectively had additional structural defects. 17/117 (14. 5%), 8/37 (21. 6%) and 2/13 (15. 4%) fetuses with fetal defects and 1, 2 and 3 markers respectively had major aneuploidies. However, 34/478 (7. 1%), 14/36 (38. 9%) and 2/7 (28. 6%) fetuses without defects and 1, 2 and 3 markers respectively had major aneuploidies. The overall LR for fetuses with increased NT, ANB and TR with associated structural defects is 1. 6, 1. 4 and 1. 5 respectively. In fetuses with 1 marker the presence of an associated fetal defect will increase the risk of aneuploidy by a LR of 2. However, if there is more than 1 marker, even in the absence of fetal defect, the risk for aneuploidy remains significantly high.

Conclusion

Increased NT is the commonest marker followed by absent nasal bone and tricuspid regurgitation. The presence of fetal structural defects along with any of these markers increases the risk for aneuploidies, irrespective of which marker it is. When there is more than 1 marker, irrespective of the presence of fetal defect, the risk of aneuploidy remains high. Hence, when performing the 11+0 – 13+6 weeks' scan, in the presence of any marker, it is vital to perform a good anatomical survey to assess for fetal defects as the presence of a defect will reaffirm the need for fetal karyotyping.