ABSTRACT

Background: Chromosome 22q11 deletion is one of the important cause of cardiovascular malformations mainly conotruncal defects.

Aims: 1) We aimed to determine frequency of chromosomal aberrations especially 22q11 deletion in children with conotruncal heart defects. 2) To identify phenotypic features that potentially predicts chromosome 22q11 deletion. 3) Determine mean platelet volume (MPV) and platelet count in children with and without deletion. 4) Screening for TBX1 gene in children with or without 22q11 deletion and conotruncal defects.

Material and Methods: 254 consecutive children with conotruncal heart defect underwent genetic analysis (Karyotyping and FISH). The association of 22q11 deletion with abnormal aortic arch and the frequency of chromosomal aberrations in parents of children with deletion were also investigated. The phenotypes were evaluated in 226 children. In addition, retrospective analysis of the MPV and platelet count was done. Children with or without 22q11 deletion were tested for TBX1 gene.

Results and Discussion: This is first major study performed in India showing chromosome 22q11 deletion in children ≤ 2 years of age with conotruncal defects. Chromosomal abnormalities were identified in 52 (21%) children, of whom 49 (94%) showed 22q11 deletion and 3(6%) had other chromosomal abnormalities. The chromosome 22q11 deletion was observed 19.3% of total children. None of the children with tetralogy of Fallot with absent pulmonary valve showed deletion. The association of deletion with right aortic arch varied with the type of conotruncal defects. Maternally derived deletion was observed in two cases. The eight extracardiac features in combination showed 93.5% agreement with presence of deletion. The best cut-off value of MPV for predicting deletion was noticed to be 8.32 fL. Multiple genes including TBX1 were found to be responsible for conotruncal defects.

Conclusions: The study suggests that specific extracardiac features along with specific type of conotruncal defect and associated cardiovascular anomaly should alert the clinician for 22q11 deletion testing. The MPV above 8.3 fL can be an indicator of 22q11 deletion. The study indicates a need for further research in order to understand the basis for variation in frequency of chromosome 22q11 deletion in subtypes of conotruncal defects.