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**Cytogenetic study of subtypes of Down syndrome and its relation with pattern of congenital cardiac defects**

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**Aim:** Although a number of studies had dealt with this topic but in case of Pakistani population data is not only insufficient but also not very well acclaimed. The study determined the relationship between the subtypes of Down syndrome and pattern of congenital cardiac defects and reports the frequency of different sub-types of Down syndrome and severity and factors influencing the cardiac defects in them.

**Method:** Using a non-probability purposive sampling technique, a cross sectional study was conducted in the Department of Genetics, Children Hospital Lahore. 160 patients were interviewed, examined and subjected to karyotypic analysis and echocardiography.

**Result:** On karyotypic analysis of 160 cases of Down syndrome, the frequency of Trisomy 21 was 96.2%, translocation 3.1% and mosaicism 0.6%. In these cases, the frequency of the congenital cardiac defects was found to be 39.4%. Of all the cardiac defects patent ductus arteriosus was the most prevalent followed by ventricular septal defects, ASD, CAVSD and TOF. 9.5% had other defects. The isolated cardiac defects existed 76% in Down syndrome cases and multiple cardiac defects occur in 23.8%. ASD existed as the double defect in 56.2% of cases. Although the congenital cardiac defects were higher in female cases 47%. The increased prevalence of PDA and VSD is found to be associated with increasing parental age.

**Conclusion:** A pattern was established between sub-types of Down syndrome and congenital cardiac defects and different predisposing factors were identified. We highly recommend further genetic studies to be carried out in this regard to evaluate the complex etiology of the disease.

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