The present findings are: (a) prevalence of CHD in Mysore was 11.08 per 1000 livebirths from 2000 to 2005; (b) males were more affected with CHD than females; (c) the most frequent type of CHD recorded was VSD followed by ASD, TOF and PDA; (d) pedigree analysis revealed that sporadic CHD were more frequent than the familial cases; (e) first-cousin and uncle-niece marriages were the major risk factors for the etiology of CHD and the most common types of CHD associated with parental consanguinity were ASD and PDA; (f) about 16.18% of the CHD patients showed chromosomal aberrations, they are, trisomy 21, isochromosome 18p, a novel subtelomeric reciprocal translocation between chromosomes 9q and 13q and trisomy 21, another novel complex variant translocations between chromosomes 5q, 9p and 13q and pericentric inversion in chromosome 9 [inv(9)(p11-q13)]; (g) microdeletion 22q11.2 was not found in any of the isolated CHD patients and (h) transition mutation in GATA4 (exon 3) was also not found in the CHD patients which could be population specific.

Thus, these findings are the maiden report from Mysore, which has contributed richly towards the understanding the genetics of CHD. Since, chromosomes 5, 9 and 13 are involved in causing CHD; these could be called as killer chromosomes of CHD. More studies on CHD is needed in our country to prevent, manage, treat and cure the disease to save millions of children. These novel findings and their implications are discussed in the thesis.