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# Genetic counseling in congenital heart diseases

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Congenital heart diseases (CHD) are the most common birth defect with worldwide incidence of 1.3million children with CHD born each year and estimated prevalence 21 million in adults worldwide. In India, among the 27 million births, 10% of them have CHD and one third of them have identifiable genetic etiology. Approximately, 25-40% of cases with CHD are associated with other anomalies and 30% children with a chromosomal abnormality are found to have CHD. Newer diagnostic tests help in identifying genetic causes in many of these children with CHD. Congenital heart diseases include structural heart defects (ventricular septal defects, arch defects), valvular diseases (aortic stenosis, pulmonary stenosis), diseases of myocardium (cardiomyopathy), rhythm disturbances (prolonged QT syndrome, Wolff–Parkinson–White syndrome (WPW) syndrome) or vascular diseases (aortic aneurysms). These diseases can be inherited in an autosomal dominant, autosomal recessive, X linked, mitochondrial or multifactorial (teratogens) manner.

The genetic cause for CHD is suspected in the following situations: If there is a family history of CHD and if there are other associated abnormalities such as developmental issues (delayed development, intellectual disability, speech or motor abnormalities), growth issues (Intrauterine growth restriction, decreased height, globally small, microcephaly), dysmorphic face and/or other parts of body- hands, feet, skin, hair, voice and exclude exposure to teratogens, intrauterine infections and maternal metabolic conditions.

With the advances in the technology, many genetic conditions are now amenable to confirmatory diagnostic testing. The choice of genetic test depends on the underlying molecular defects, availability of the test and the cost. The genetic tests that may be useful are karyotyping, florescent in-situ hybridization (FISH), array comparative genome hybridization, multiplex ligation probe amplification (MLPA), DNA tests for single gene disorders (polymerase chain reaction (PCR), genetic sequencing), DNA sequencing for multiple genes in single test (next generation sequencing, exome sequencing), tests for inherited metabolic conditions and non-invasive prenatal testing (NIPT).

Genetic counseling in the context of CHDs of genetic origin is complex and requires detailed clinical and laboratory evaluation. The important step in the genetic counseling is establishment of confirmed molecular diagnosis and carrier status determination of parents. This helps in predicting course of disease and co-morbidities and decision on cardiac surgery / intervention. The molecular diagnosis also helps in estimation of recurrence risk is subsequent pregnancy, prenatal diagnosis and offspring risks for affected individual. The recurrence risk depends on the mode of inheritance and carrier status of the parents. It also provides an opportunity for the development of targeted gene therapies.