Genetic diagnosis of congenital heart disease : Where do we stand today?

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Abstract:

Congenital Heart Disease (CHD) is still the most common congenital disorder affecting infants worldwide. In India alone it has been estimated about 200,000 children per year are born with CHD. Abnormal development during heat development causes defects which could range from minor defect/s without any symptoms to major defect/s which could be severe and, in some cases, life-threatening. As heart does not have the capacity to repair or regenerate, patients sometimes need surgical intervention to correct the defects.

With the advancement of technology there has been a remarkable improvement in the survival rate of children in the recent times. That means, children who were born with CHD grow to be adults and have children of their own. But, in such cases there is about 5% chance that the child born to them would also have CHD. Although, we still don't know the exact reason/s that causes CHD, several studies have supported the fact that chromosomal variations; interaction of several genes and environmental factors contribute to different types of CHD.

Based on this, nowadays we are able to provide genetic investigations and testing for both syndromic and non-syndromic types of CHD. Genetic investigation, in the form of pedigree analysis is crucial for early screening to determine possible inherited defects. Today, genetic tests are available that analyse chromosomal variations and genes associated with both syndromic and non-syndromic CHD. A proper diagnosis genetically could lead clinicians to determine if there is a need for additional evaluation or screening to assist in management of CHD and its recurrence.

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