Abstract

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

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Editorial

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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Biography:

Dr. Smitha Gawde is a driven molecular geneticist with 15 plus years experience working in molecular diagnostics, human genetics research as well as teaching. She has published both in national and international journals. Her specialties lies in Cytogenetics, Molecular Biology, Molecular Diagnostics and Translation Research.

An energetic, effective and visionary organizational leader, who believes that "good enough" is never good enough. Known as an innovative thinker with strong life science and genetics acumen. Demonstrated success developing and executing plans in complex regulated organizational structures. Passionate in helping others achieve their potential through ensuring that well-being and integrity are supported by inspiration and curiosity.

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