

Whole exome sequencing for identification of rare variants in cardiac disorders: Experiences from India

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Received: November 10, 2017 | **Published:** November 14, 2017

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The availability of the human genome sequence and advances in next generation sequencing technologies has opened opportunities for a new era of genomic medicine implicating on diagnosis, treatment and preventive care relating to genetic diseases. One of the major applications of such genomic technologies in the clinical settings is in the identification and annotation of variants associated with rare genetic diseases. It has been estimated from various projections that India is home to over 70 million people suffering from a genetic disease.

In order to provide benefit of the emerging field of genomic medicine to clinicians and patients suffering from rare genetic diseases, we establish the Genomics for Understanding Rare genetic Diseases India Alliance Network (GUARDIAN). We applied the power of genomics for systematic characterization and diagnosis of rare genetic diseases within the GUARDIAN consortium. We share our proof-of-concept experience in the utility of whole exome sequencing for identification of rare variants in cardiac genetic diseases from India.