BioSpectrum
the business of Bio & Health Sciences

Researchers identify rare form of hypertrophic cardiomyopathy

27 November 2020 | News

This study was the first of its kind from India that analysed the inheritance pattern of the mutation in a large number of multigenerational affected members from three unrelated families

Hypertrophic cardiomyopathy (HCM) affects the health of the heart muscle. Genetic cause of ~40-50 per cent of HCM are not known. The mutations responsible for HCM often localised to genes encoding sarcomeric proteins. In a study published in the journal Nature Scientific Reports, Scientists from MedGenome, Bengaluru, India and Amrita Institute of Medical Science and Research (Amrita Hospital), Cochin, India report a rare form of HCM where they have identified mutation in a non-sarcomeric protein PRKAG2.

This study was the first of its kind from India that analysed the inheritance pattern of the mutation in a large number of multigenerational affected members from three unrelated families.

The genetic data obtained helped refine the clinical diagnosis providing a template for personalised treatment applying genomics in the clinic. The families in the study were followed longitudinally for over seven years to understand the natural history and clinical outcomes of the affected individuals adding to our knowledge of the disease and interventions needed.

Dr Sameer Phalke, Senior Scientist, MedGenome and senior study co-author said, “Genetic testing for HCM diagnosis and screening for risk can now be used routinely for overall disease management.”

“As far as inherited cardiovascular disease is concerned, it is quite clear that we are now in the era of leveraging the potential of cardiovascular genetic testing for the prompt recognition of potentially life-threatening disease as well as choosing an appropriate management strategy to achieve optimum outcomes in our patients.” said Dr Hisham Ahmed, Clinical Associate Professor of Cardiology, Amrita Institute of Medical Sciences and Research, Kochi, the lead clinical scientist and senior study co-author.