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Heart ailment, diabetes have genetic link: Study

Will Aid Early Detection In South Asians

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Bengaluru: Scientists and doctors from several cities in India and the US say they have provided the first evidence for a shared genetic link between cardiomyopathy (condition of heart muscle leading to failure) and metabolic syndrome (high blood sugar, excess body fat around the waist and abnormal cholesterol levels) among South Asians. This information, the team says, can aid early identification and development of preventive strategies. While recent epidemiological and clinical studies suggest that around 10% of cardiomyopathy patients coexist with diabetes, the genetic link underlying it is less understood. Findings published in the latest issue of “Science Advances”, an international peer-reviewed journal, by the team led by Dhandapany Perundururai, investigator at the Centre for Cardiovascular Biology and Disease (CCBD) at Bengaluru’s Institute for Stem Cell Science and Regenerative Medicine (DBT-inStem), provide evidence for this.

What they found

Perundururai’s lab screened 2,552 cardiomyopathy patients with and without diabetes, and identified novel and ultra-rare variants in a metabolism-related gene called “Adiponectin receptor protein 1(ADIPO1R)”.

“Importantly, these gene variants were absent in South Asian-specific 1,800 healthy individuals. ADIPO1R is a receptor for the hormone adiponectin, which regulates glucose and lipid metabolism in human cells,” the team said. To understand these variants’ role in causing heart disease, researchers used cardiomyocytes — cells involved in heart function — and showed significant dysregulations in glucose metabolism and increased cell size with enhanced expression genetic markers associated with cardiac hypertrophy.

“As each of these variants is found to be affecting a unique signalling molecule and its biological process, therapeutic strategies should be tailor-made based on the nature and type of the variant associated with this disease,” Perundururai explained. “A simple genetic test would help spot individuals highly susceptible to cardiac muscle disorder. Early diagnosis of an ‘at-risk’ population will help prescribe appropriate preventive health plans to subdue harmful effects associated with this disease,” Perundururai added.