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Predicting susceptibility to coronary artery disease using single nucleotide polymorphisms with a large-scale data extraction from pubmed and validation in an asian population subset

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Coronary artery disease (CAD) has a genetic component. Single Nucleotide Polymorphisms may help to identify a disease's underlying causes. This study aimed to identify SNPs associated with CAD. The research involved a deep literature mining process using bioinformatic techniques and manual efforts

The identified SNPs were then validated in Asian population. The aim of this study was twofold. Firstly, to identify and select SNPs associated with CAD using a combination of bioinformatics and manual approaches. Secondly, to validate the identified SNPs in an Asian population subset.Firstly, an automated pipeline was employed to extract structured information on SNPs, Population, and Diseases from approximately 28 million PubMed abstracts.

Natural Language Processing (NLP) techniques and Python scripts were utilized. The collected data was then curated, filtered, and categorized into 24 hierarchical groups using Named Entity Recognition (NER) algorithms and 466 unique PubMed Identifiers (PMIDs) and 694 SNPs related to coronary artery disease (CAD) were identified. Further, a thorough manual examination of all studies was carried out. SNPs demonstrating susceptibility to CAD and exhibited a positive Odds Ratio (OR) were selected and final pool of 324 SNPs was compiled. In second phase, identified SNPs were validated using DNA samples from 96 CAD patients and 37 healthy controls of an Asian Indian population subset through Global Screening Array. From initial pool of 324 SNPs, only 108 SNPs were successfully expressed in samples. Among these, four SNPs showed a significant difference in minor allele frequency between the CAD patients and healthy controls, namely rs187238 (IL- 18 gene), rs731236 (VDR gene), rs11556218 (IL16 gene), and rs5882, have been linked to various pathways related to endothelial damage, vitamin D receptor susceptibility, and HDL-cholesterol levels. Of these, only rs731236 had been previously studied in Indian population, specifically in diabetes and vitamin D deficiency..

This study reported, for the first time, the association of these SNPs with CAD in the Asian Indian population. The pool of 324 SNPs identified in this study serves as a unique resource for uncovering risk associations in CAD. Additionally, validating these SNPs in different populations can provide valuable insights and contribute to the development of a screening tool. This research enables the implementation of primary prevention strategies targeting vulnerable population.

Biography

Presently pursuing PhD in Pharmacology Department of All India Institute of Medical Sciences. On the professional front, to utilize my knowledge and skills in the best interest of my organization and the mankind. On the personal front, to keep myself updated in my field of work and to put in my best efforts in performing my duty. My research interests are preventive cardio vascular research, pharmacology, basic research, genomics, traditional medicine, Ayurveda Sciences, Medicinal plants.

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