Jain et al. (2017) Can J Biotech, Volume 1, Special Issue, Page 80
https://doi.org/10.24870/cjb.2017-a67

Canadian Journal of Biotechnology

ISSN 2560-8304

Poster Presentation OPEN ACCESS

Category: Clinical Genomics

Incidental and clinically actionable genetic variants in 1005 whole exomes and genomes from Qatar

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Abstract

Next generation sequencing (NGS) technologies such as whole genome and whole exome sequencing has enabled accurate diagnosis of genetic diseases through identification of variations at the genome wide level. While many large populations have been adequately covered in global sequencing efforts little is known on the genomic architecture of populations from Middle East, and South Asia and Africa. Incidental findings and their prevalence in populations have been extensively studied in populations of Caucasian descent. The recent emphasis on genomics and availability of genome-scale datasets in public domain for ethnic population in the Middle East prompted us to estimate the prevalence of incidental findings for this population. In this study, we used whole genome and exome data for a total 1005 non-related healthy individuals from Qatar population dataset which contained 20,930,177 variants. Systematic analysis of the variants in 59 genes recommended by the American College of Medical Genetics and Genomics for reporting of incidental findings revealed a total of 2 pathogenic and 2 likely pathogenic variants. Our analysis suggests the prevalence of incidental variants in population-scale datasets is approx. 0.6%, much lower than those reported for global populations. Our study underlines the essentiality to study population-scale genomes from ethnic groups to understand systematic differences in genetic variants associated with disease predisposition.

Citation: Jain, A., Gandhi, S., Koshy, R. and Scaria, V. Incidental and clinically actionable genetic variants in 1005 whole exomes and genomes from Qatar [Abstract]. In: Abstracts of the NGBT conference; Oct 02-04, 2017; Bhubaneswar, Odisha, India: Can J biotech, Volume 1, Special Issue, Page 80. https://doi.org/10.24870/cjb.2017-a67