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Abstract Topic:- Evolutionary and population genetics

Abstract Title:- Genetic diversity in the populations of India - Impact of founder events.

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Aims:-Disease prevalence varies across population groups, determined by their ethnicity, geographical isolation, and cultural practices. Identification of genetic etiology of such diseases provides a key understanding of their biology, diagnosis, treatment, counseling and prevention. India is a country with remarkable diversity, inhabited by approximately more than 5,000 anthropologically well-defined groups. Most of these are endogamous communities with high IBD (Identity-by-decent) scores and significant barriers to gene flow due to social, linguistic and cultural factors. A previous work of genome-wide SNP analysis on 263 distinct South Asian groups identified 81 populations with IBD scores significantly higher than the widely studied archetype population, Finns and the Ashkenazi Jews. Such strong founder events often result in a population with a distinct genetic composition. Some of these population specific alleles might be deleterious, possibly leading to high occurrence of certain genetic diseases in these population groups.

We aimed to identify the population structure and genetic diversity in 4 selected groups with high IBD scores (Yadavs, Kallar, Reddy and Kalinga) from South India.

Methods:- We generated whole exome sequence data and SNP genotype array data for 339 and 96 individuals from these groups, respectively.

Results:- We report the presence of IBD regions, with ~3 times higher IBD score in these groups when compared to the Finns, although the founder age was estimated to be comparable. From around 80,000 total variants identified, we discerned the known and novel (not reported as of date) variants for each group. We identified several known pathogenic variants in significantly higher frequencies than seen in 1000 Genomes and GenomeAsia100k data. Using in-silico prediction tools, we delineated potentially highly deleterious novel variants in these groups. We also report around ~50% of these populations to be inbred. Further, we also annotated and cataloged the presence of several actionable pharmacogenomic alleles following the CPIC-PharmGKB recommendations.

Conclusions:- We believe that our work constitutes a comprehensive investigation into the implications and the clinical relevance of already known strong founder events in India.

Keywords:- Founder event, IBD score, Inbreeding coefficient, Deleterious variants, Pharmacogenomics