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NIBMG Participates in 'The GenomeAsia 100K Project'

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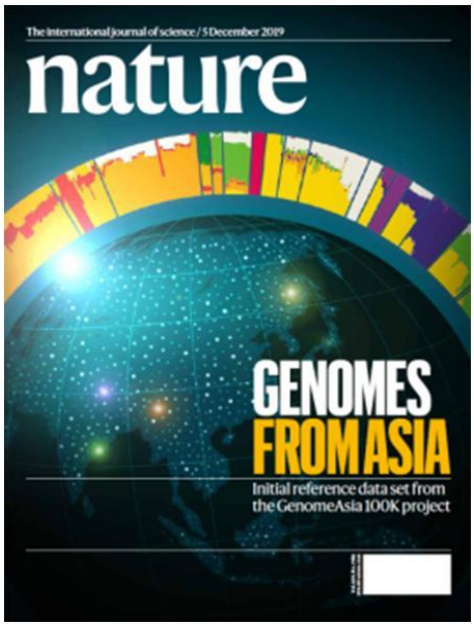

Researchers are making huge expenditures to assay genotypes of Asians by using the DNA microarrays constructed largely on the basis of data of non-Asians. A deep baseline genomic variation data are required from Asian populations to optimally design DNA microarrays for use in Asian populations. Such data is expected to stimulate the biotechnology industry in Asia.

Therefore, recognizing the scarcity of data on Asian populations, the GenomeAsia 100K Consortium was established in 2016 with the mission *"Let's Map the Gap in Genomic Data."*

The consortium comprises of Scientists from academic institutions such as Nanyang Technological University, Singapore; National Institute of Biomedical Genomics, Kalyani, India; University of California at San Fransisco, USA; etc. and the biotechnology industry such as Genentech, USA; Macrogen, South Korea; MedGenome, India; etc.

Baseline data on genetic variation among humans is warehoused in various global databases, such as 1000G, ExAc, etc. Asia comprises about half of the world's population, however, less than 10% of genomic data in global databases is from Asian populations. This under-representation has severely prevented the efficient conduct of research for identifying genomic bases of diseases which are of importance to Asians.

Further, design of DNA microarrays that are widely used for inexpensively discovering genes involved in diseases and for understanding human movements, admixtures and evolution has

been severely hampered. A large fraction (appox. 30-40%) of the data generated by the currently available DNA microarrays does not provide useful information on Indian populations.

This study conducted at the National Institute of Biomedical Genomics (NIBMG), Kalyani is the largest genomic study conducted among population groups of Asia. It comprised generation and analyses of whole-genome sequences of 1739 individuals from 219 populations spread across most countries of Asia, out of which 598 are from Indian tribal and non-tribal populations.

Study discovered about 200,000 previously unreported novel DNA variants among Asians. Alterations of proteins are usually associated with disease, but, 23% of protein-altering variants found in Asia are unreported in existing databases. Therefore, the novel data are critical for discovering genes related to diseases that are of high prevalence in Asia, and to design of better DNA microarrays for use in Asia.

Study revealed that the structures of Asia populations are complex. They have admixed with multiple (about 14) ancestral populations. Besides, different regions have also admixed with different and smaller subsets of ancestors. The genome sequence data obtained has summarized the earlier findings based on DNA microarray data. Earlier five predominant ancestral admixture events seem to have taken place to give rise to the present-day Indian populations, including the populations of Andaman & Nicobar Islands.

Furthermore, it was discovered that even some large urban populations of India (e.g., the population of Chennai) exhibit genetic characteristics that are usually found in isolated populations. Team have discovered that two different admixture events with archaic humans (Denisovans) in Southeast Asia. Understanding population structure makes disease gene discovery studies efficient and cost-effective.

Using these data, team has discovered novel DNA variants even in many well-studied genes implicated in various diseases, such as diabetes, thalassemia and breast cancer. Many drugs have adverse effects on individuals with specific DNA variants. Researchers found that Carbamezepine, a drug used for treatment of some mental disorders, may have adverse effects on about 400 million speakers of Austronesian language residing in South-East Asia.

GenomeAsia 100K consortium will continue to gather and analyze DNA sequence data from Asians with a view to disease gene discovery, precision medicine and drug development.

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