

# Store Your Precious Bio samples and the derived Data Today with Transcell Biolife – Your Trusted Bio bank in India Providing Unique Bio bank Applications

Dec 2019

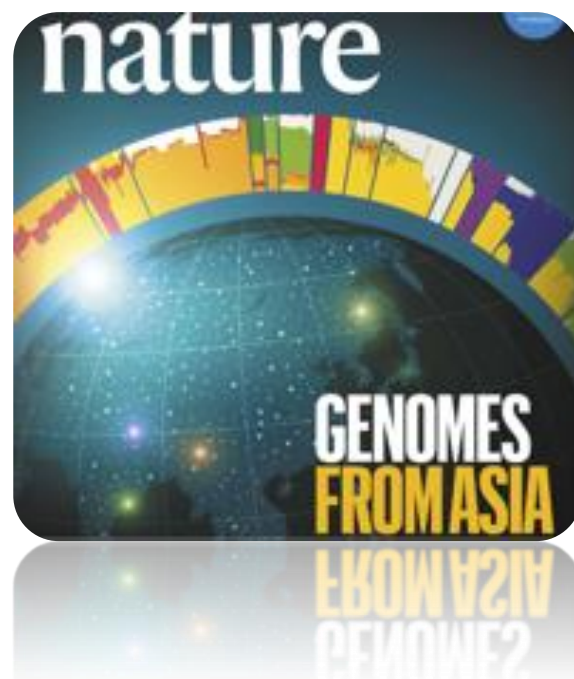


The Transcom Newsletter

Version 1

## Understanding the genetic basis

of human disease in the context of diagnosis to prevent and find cure is possible from an increase in the number and scale of disease-association studies that are carried out in all populations. To the reader's knowledge, there are 51 contemporary ethnic (category of population with presumed common genealogy) groups in the world today. Population groups with differences determined by ethnicity also show differences in terms of illness behaviour which is a result of genome related variation. Medical conditions such as sickle cell disease and Creutzfeldt-Jakob disease; cardiovascular disease and type II diabetes are classic examples with altered prevalence and patterns in different ethnic groups.



It has come to a point now that Indian population specific genome data is an important asset to create, analyze datasets that may allow mapping out strategies focused on population specific understanding of how genetic variants affect disease susceptibility and drug responses. The Indo Aryans are the diverse group of people in India: Assamese, Bengali, Gujarati, Hindi, Kashmiri, Konkani, Marathi and Punjabi. Outside of South India where most Indians are of Dravidian origin.

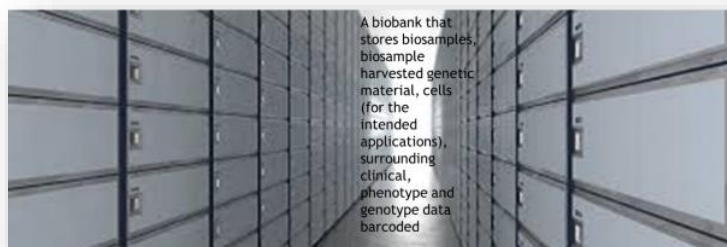
Stem cells obtained from the biological discards like Umbilical cord and Tooth (one time opportunity in an individual's life) are the best materials for genome sequencing with multiple benefits to the donor in applications to do with biobanked samples and data.

*Transcell Biolife is the only biobank in India for the intended application*

### Extracts of the recent publication:

Wall, J.D., Stawiski, E.W., Ratan, A. et al. The GenomeAsia 100K Project enables genetic discoveries across Asia. Nature 576, 106-111 (2019).

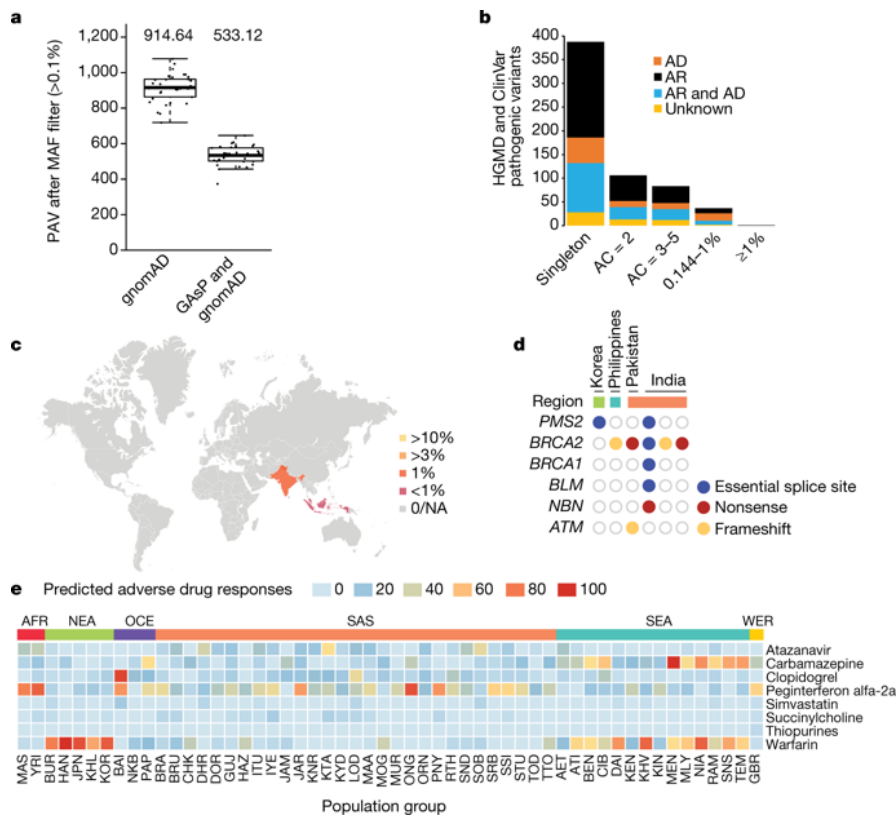
This study reports sequence from 1,267 individuals that includes 598 individuals representing 55 ethnic groups that span the major language groups across India. Importantly, this study found many large population groups from India in which individuals were more related to each other by descent.



These groups are similar to the Finnish population group where many disease gene discoveries were made. The Finnish-equivalent Indian groups are going to be a great resource for disease gene discovery and they will aid in target identification, drug development and disease management.

This study has identified many genetic variants that are specific to Indian population groups that were previously not known. Some of these are common variants in the Indian groups, but when first identified by previous studies from India involving smaller sample size, they were thought to be disease causing (for example in diabetes) as they were not represented in the Eurocentric variant database.

### Genetic Variants Predispose Individuals to Adverse Drug Effects Observed Adverse Effects For Certain Drugs Linked To Genetic Variation



Several variants that predispose individuals to higher cancer risk were identified in this study. Once this part of the work is expanded, the data from this can be used to screen individuals to understand the disease risk and provide appropriate monitoring and proactive treatment. Similarly, variants linked to increase in adverse effect in individuals for certain drugs were found. Understanding this will allow doctors to provide alternate safer drugs to such patients.

Genome sequencing has come a long way, but majority of genetic mutations or variations are still not validated in Indian population. Biobanking is the only way to have complete understanding of South-Asian Genome and in specific the Indian genome. Majority of metabolic or genetic disease diagnostics and therapy monitoring are dependent on genomic data obtained from non-Indian genomes, thus clinical outcomes are poor. For example, Clopidogrel which is a first line therapy given in heart diseases like coronary artery disease, does not work for several Indian patients due to the fact that they carry SNPs in CYP19C gene which is needed for metabolizing this pro-drug to drug in liver. It is important to identify such genotypes and validate in Indian population. Such developments of molecular markers for diagnostic or therapeutic value are possible if we promote biobanking technologies done at Transcell Biolife and Institute for Applied Research and Innovation.

