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MedGenome to Unveil the South Asian Genome Variation Database (SAGVDB) Capturing Genetic Variations in South Asian Populations

PRNewswire MedGenome May 6, 2016 9:30 AM

FOSTER CITY, California, May 6, 2016 /PRNewswire/ --

MedGenome will present its abstract titled 'SAGVDB - An integrated resource for South Asian genome variation' at the **Big Data Bioinformatics Conference**, to be held on **May 26th-27th 2016** in **Boston, MA.**

(Logo: http://photos.prnewswire.com/prnh/20160112/784044)

A deep understanding of human genetic variation is essential for investigating the relationship between genotype and phenotype. The genetic sequence variations in individuals from different groups in South Asia is still not extensively explored. MedGenome's SAGVDB (South Asian Variation Database) comprises of 215 whole genomes and 121 whole exome from India, Pakistan, Sri Lanka and Bangladesh. The raw data compiled from various publications were processed using MedGenome's whole genome data analysis pipeline. The database contains ~ 34.2 million variants, of which ~ 31 million are SNPs and ~ 3.2 million are short InDels and includes variants from South Asian samples present in 1000-genome-phase3 and ExAC databases. The database also integrates variants from other studies including 1000-genome, ExAC, UK10K, GoNL, Iceland, EVS, 1000Japanese, Wellderly. In addition to showing allele frequencies, statistics for population/sub-population, a genome browser with adequate genome tracks, gene/transcript level annotation, etc. can enable researchers to dig deeper into the variants and their significance.

South Asian population, predominantly contributed by Indian sub-continent, represents a unique blend of genetically homogeneous, but socially and geographically stratified groups of individuals that can be leveraged to investigate complex diseases and disease phenotypes. MedGenome believes that the SAGVDB will be a great resource to enable further research into human diseases, identify new disease-causing variants, which will have a positive impact on global healthcare outcomes.

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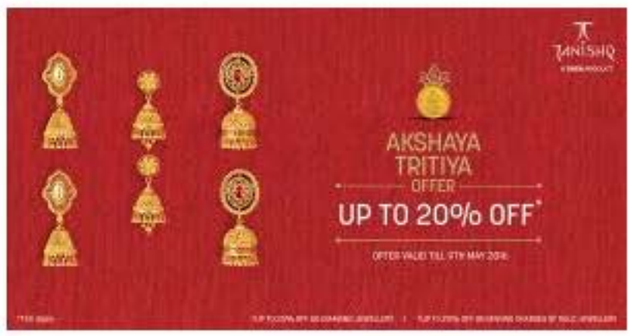

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