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MedGenome shows novel variations in common genetic disorders in India

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In a recent research study, Bengaluru based MedGenome Labs, the leaders in genetic testing in India, in partnership with Sir Ganga Ram Hospital, New Delhi, conducted a pilot study to determine the carrier frequency and to look for any novel mutations seen in the Indian population for common genetic disorders.

The study was conducted over a period of 22 months with a sample size of 200 unrelated individuals, in the North Indian population. After pre-test genetic counseling, the 200 individuals were screened for pathogenic variants in shortlisted 88 genes using Next Generation Sequencing (NGS) technology. These variants were classified as per the guidelines of American College of Medical Genetics. The study was facilitated by MedGenome Laboratories Bangalore, who carried out the molecular analysis, using NGS and the data was re-analyzed at Sir Ganga Ram Hospital, New Delhi.

Out of the 200 participants, 52 (26%) were found to be carriers of one or more rare genetic disorders, 12 individuals (6%) were identified to be carriers for congenital deafness and the 9 individuals (4.5%) were observed to be carriers for cystic fibrosis. Three individuals were detected to be carriers for Pompe disease. This study showed a higher carrier frequency for these disorders which was contrary to the generally held view about their low prevalence in Asian Indians.

Another interesting finding was that the disease-causing variants observed for disorders such as deafness, cystic fibrosis, Pompe disease, Canavan disease, primary hyperoxaluria, junctional epidermolysis bullosa, galactosemia, medium chain acyl CoA deficiency etc. were different from what is seen in the Western population. Thus, this pilot study highlights the importance of having a Genetic Variant Database for the Indian population.