

Abstract Title: Traversing complex Genetic landscape of India: A journey of three decades

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Abstract: With a population of over 1.4 billion in India and 23 million babies are born in the country every year, there is no dearth of rare genetic disorders in the country. It is estimated that the burden of rare diseases out passes the burden of diabetes and cardiovascular diseases with an estimated 70 million people afflicted with rare diseases. Our journey started in 1980 with the first report of prenatal diagnosis of omphalocele by measuring AFP serum marker using Rocket electrophoresis which opened our journey to the understanding of neural tube defects and its prevention. This further opened up the new area of rare diseases study in India like identification of the gene locus for Clouston syndrome and lysosomal storage disorders. Our first report on the burden of LSDs identifying Gaucher disease as the most common storage disorders with L444P mutation was followed by a task force for LSDs in India. The study has further identified many rare diseases with founder mutation in the Indian population like P77R in a Gujarati community for Morquio-A disease, E462V and D322Y in a Parmar community for Tay Sachs disease, recurrent variant c.298TA in CCN6 gene in a Patni community from Gujarat for Progressive Pseudorheumatoid Dysplasia. Our recent study has identified 337 rare disease burden in India with LSDs as the most common, followed by Neuromuscular and haematological disorders that will help to make a National policy for the prevention of the most common rare diseases in the country. Our recent study to understand the variable phenotype with a common genotype of L444P in GD has identified this mutation as a heterozygous in all normal population in the GBAP1 gene likely to provide the answer to understand the phenotype variability with same genotype in GD.

Area of expertise: Lysosomal storage disorders, Metabolic disorders, Mandelina disorders, Genetic counseling, molecular genetics