GOMED
Genomics and other omics tools for Enabling Medical Decision
CSIR INSTITUTE OF GENOMICS & INTEGRATIVE BIOLOGY

Genomics and other *omics* technologies for Enabling Medical Decision

Genetic diseases, though are individually rare, cumulatively affect a large number of individuals. It has been estimated that genetic diseases affect **over 70 million Indians** (ORDI 2015) and a total of **over 300 million worldwide**. In majority of the cases, an appropriate diagnosis is not arrived at, due to lack of general awareness on genetic diseases, lack of access and high-cost of appropriate genetic diagnostic services. It is estimated that an average patient with rare disease has to go through multiple healthcare visits and diagnostic tests and approximately 7 years to arrive at a precise diagnosis. Nevertheless, an equitable access to genetic diagnostic services could potentially provide a better quality of life to the individual, family and society at large. For example, early genetic diagnosis and treatment of a large and increasing number of genetic diseases like achondroplasia, Turner syndrome and Gaucher's disease, could significantly improve the quality of life and outcome of patients. Diagnosis of a genetic disease in adulthood is also equally important as this could potentially prevent the transmission of genetic diseases to the future generations through counselling and/or companion prenatal genetic diagnostics. This has been majorly due to the lack of general awareness on genetic diseases, and the lack of an easily available and affordable system for offer genetic testing to aid clinicians. We hope the availability of an accessible, affordable system and equitable access to the genetic testing services would go a big way to significantly reduce the disease, social and economic burden due to these debilitating disorders.

CSIR Institute of Genomics and Integrative Biology (CSIR-IGIB) with its rich expertise in the area of genomics and a wide network of clinical partners across the country has embarked on a unique outreach programme, to **enable equitable access to genetic testing for frequent genetic diseases**. This programme called GOMED (Genomics and other omics technologies for *Enabling Medical Decision*) aims to provide a platform for clinicians to tap into the rich and varied expertise of CSIR-IGIB in disease genomics to solve clinical problems. The Institute brings to table its pioneering expertise in genomics in the country, demonstrated over almost a decade through the Indian Genome variation project, the sequencing of first indian personal genome and ongoing clinical genomics efforts in rare and common diseases with a large number of public and private healthcare institutions in the country. Through this effort, we make available this in-house expertise to a larger community of clinicians to enable precise diagnosis and prognostication. It is proposed that a dedicated in-house project could potentially meet the gap in making available these assays to clinicians.

The present portfolio includes over 80 genes and sequencing of mitochondrial loci for mitochondrial disorders and would significantly improve over time. In the initial phase, we hope to be able to reach needy patients through our clinical network and make a difference in their lives through providing appropriate genetic testing services. **In this endeavour, we look forward to partner with like-minded individuals, organisations and philanthropists to improve the access, affordability, reach and portfolio of genetic and genomic tests**.

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