



## Human genome research in the context of P4 medicine- How rosy and not so rosy is the picture?

by

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Genetics in health and illness has engaged both observational and experimental scientists for centuries. Of late, human genome research has undoubtedly witnessed one of the most exciting phases, with discovery genomics dramatically changing the fundamental understanding of human/disease biology. Newer technologies have contributed notably to the incredible progress in this field which in turn has paved the way for new paradigms. Outcome of the elegant discovery genomics is now propelling translation of the findings for fulfilling the goals of P4 (predictive, preventive, personalized and participatory) medicine.

In the first part of my talk, the exciting journey of decoding the human genome which has revolutionized biomedical research will be presented, with examples from our studies. In the second, translational potential of some of the genomic findings, particularly for familial disorders will be highlighted. To this end, a novel initiative of newborn screening for inborn errors of metabolism, an example of effective technology for the masses, that we have undertaken will be showcased. Next, the not so rosy part, of the otherwise engaging genome research, which primarily includes the major limitation for prediction and prevention of common complex disorders such as rheumatoid arthritis, type 2 diabetes, cardiovascular disease etc will be shared. Finally, how we believe, that Ayurgenomics, a combination of the principles of Ayurveda, our ancient holistic system of medicine and contemporary genomics, may be able to tackle this daunting limitation to achieve predictive health will be discussed.



Professor Thelma B.K. is a faculty member and Team leader of the Centre of excellence in Genome sciences and predictive medicine in the Department of Genetics at the University of Delhi south campus, New Delhi.

Unraveling the genetic basis of both Mendelian disorders and common complex traits is the major focus of her research work. Identification of novel disease causing/risk conferring gene variants for X-linked intellectual disability, Parkinson's disease, Schizophrenia, Rheumatoid arthritis and Ulcerative colitis are some of the highlights of her recent work. Population specific common and rare variant data generated by genome-wide association studies and whole exome sequencing on Indian cohorts are a rich resource generated in her laboratory. As a natural sequel, understanding disease biology by functional characterization of promising genetic leads and computational biology are being pursued in the laboratory. Translation of genetic findings to DNA based diagnostics, pharmacogenetics and lead molecule development is the ultimate goal of her group. To overcome limitations faced in complex trait genetics, an alternate paradigm of Ayurgenomics- combining Ayurveda doctrines with contemporary genome analysis tools is being explored.

Prof. Thelma has several national and international research projects and has published her research in international journals of repute. Prof. Thelma is a fellow of all the three science academies in India. She serves as an expert member in several task force committees of funding agencies and has also served as a member on the Science and Engineering Research Board, India and on the Scientific Advisory Council to the Prime Minister of India.

**THURSDAY, November 15, 2018 | 04:00 p.m. | Faculty Hall, Main Building, IISc.,  
Tea will be served following the talk**

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