

Personal genomes to precision medicine

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Received: November 10, 2017 | **Published:** November 14, 2017

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One of the major advances that happened in the last decade has been the availability of fast, efficient and cost-effective DNA sequencing which has seen tremendous applications in healthcare and research. This has been largely possible with the advent of newer sequencing approaches which offer a higher throughput, lower cost and faster turnaround times, and appropriate computational tools to enable the handling, processing and mining of information from them. On one end, this technology enables to elucidate the genomes of individuals and how genomic variations could influence the life of the individual, while on the other side, it enables one to peek into the pathophysiology of diseases and have a mechanistic view of the disease processes. It is widely believed that these advances would be immensely useful in healthcare through Predictive, Preventive, Precise, Personalized and

Participatory Medicine.

In the present talk, we would describe our experiences with personal genome sequencing and use of genomics approaches and model systems towards this end. We have employed a number of novel approaches towards harnessing the BigData challenge including involvement of students towards analysis and model-building. Systematic annotation of datasets for personalized genomics for the OpenPGx consortium and analytical tasks through the 2C4C programmed for Cheminformatics could be crowd sourced through the platform. The talk would also discuss the way forward, detailing the ongoing initiatives including the GUARDIAN Consortium involving a large clinical network towards realizing the dream.