# How Technological Advancements In Genomics Are Helping Prevention And Cure Of Diseases!

Guest February 15, 2016

From electronic health records, wearable technology, telemedicine, self-service kiosks and genome sequencing, the last decade saw the most innovation in the healthcare domain. With its speed, precision and accessibility, rapid advances in technology have helped take healthcare services out of the confines of hospital walls and integrate them with high end, user friendly devices which have made patient care easy and accessible. The field is moving rapidly from a reactive to preventative approach where predictive analysis and precision medicine are at the forefront, all thanks to the growing popularity of genomic tools.

A study of your genome—chromosomes, genes, and their functions; genomics focuses on the aspects of locating variations in your genetic material which can be helpful in spotting, preventing and controlling diseases. More than a decade after the successful mapping of the whole human genome, clinical genomics has permeated into the healthcare scenario at large and can be considered as one of the major technological innovations in the field.

Applying genomic data in clinical practice has opened new avenues for improved diagnosis, targeted treatment and precision personalized care. The true value of genomic medicine rests in understanding, analysing and incorporating genomic information in an individual's health and well-being.

## Clinical Genomics: Impact On Healthcare

Genomics as a field is beginning to show its potential in reforming the healthcare sector and in turn improve patient care. The debate is no longer focused on 'if genomic medicine will impact healthcare' as much as how rapidly this impact will begin to show. The evolution of next generation sequencing techniques, availability of more than a thousand different genetic tests, and the use of pharmacogenomics and targeted therapies in treatment plans suggest that we are already moving towards a major healthcare reform.

Genomic medicine will allow us to take significant pre-emptive measures years before the onset of a disease, even before the symptoms start appearing. It will also allow clinicians to define which drugs and interventions hold the most likelihood for success, thereby changing care from interventional to preventative. As genomic technologies continue to evolve, genetic information will become an integral part of every patients medical record. Significant benefits are already starting to surface, although in more specialised settings.

## Improving Diagnosis through Screening

Every individual has a unique genetic makeup which gives him a completely distinctive profile for developing any particular disease. The use of clinical genomic tools in improving diagnosis mainly involves:

1. Prenatal testing: Involves screening for chromosomal aberrations (Down's syndrome) in newborns
2. Risk analysis: Includes testing for specific mutations in genes (BRCA1 and BRCA2 for breast cancer), rare genetic disorders which can develop in adulthood, lifestyle diseases like diabetes, heart diseases etc. which are broadly multi factorial.
3. Pharmacogenomics testing: Helps determine the most effective treatment based on your genetic profile
4. Molecular imaging: Makes use of biomarker probes for improved diagnosis

## Improving Treatment through Targeted Therapies

A fundamentally different approach for therapeutic use, targeted therapy is changing the traditional pharmaceutical model of 'one size fits all'. A given drug can produce a spectrum of responses in different individuals. Using genetic testing it is now possible to identify if a particular drug will have significant effect in treating a patient. It also helps to determine if a drug will produce any adverse events or have no effect at all.

In parallel, genomic data is helping researchers suggest new therapeutic applications for existing treatments, popularly known as repositioning. This approach known as 'drug-target isolation' offers a range of advantages as compared to conventional treatments.

**Improving Disease Prevention & Management**

Genome wide scans have made it possible to obtain a more accurate assessment of an individual's risk for developing any particular disease or diseases. This information can be utilised by hospitals to tailor their prevention and management programs and focus on a more personalized, molecular approach, especially for preventing chronic diseases.

**Role Of Big Data & Artificial Intelligence In Genomic Medicine**

Unlike conventional diagnostic tests, genomic tests produce a huge amount of data, as large as 300GB. However, looking at a massive number of data points is pivotal, to find out all the areas of interests and their clinical implications, if any. This is where computationally intensive tools like artificial intelligence, big data and machine learning come in the picture.

Big data and artificial intelligence platforms help us to use the efficiency and high output capacity of a computer to find genetic variations, discover patterns, predict the best available treatment, and most importantly transform medical records from mere e-filings to full-fledged comprehensible reports that can deliver clinically relevant and high quality data in real time. Data analytics also provides an excellent opportunity for researchers and manufacturers to understand and replicate the data obtained through clinical trials.

**Genomic Literacy: A Key Factor For Genomics**

In order to fully harness the potential of genomic tools and to successfully incorporate them in healthcare, genomic literacy is important. Genetic information is complex and consumers can have difficulty in understanding and interpreting this data and how it will impact their lives. Similarly, the understanding of genomics and the corresponding data is not widespread in the medical community, either.

With a growing demand for genomic services, we need highly qualified and trained professionals who can understand the importance of genetic data and convey this sensitive information to the consumer in an appropriate manner. Investing in genomic literacy will not only increase awareness amongst patients but also professionals who can then offer the necessary counselling to their patients.

**The Way Forward**

With increased awareness and consistent developments, it is becoming easier for the general population to gain access to genomic tests through direct-to-consumer testing services. Integrating genomic and clinical data requires a strong investment in computational capacity. Given the large amounts of data flowing in, it is imperative to adopt tools like big data and artificial intelligence to built a knowledge repository to help clinicians and health care providers interrogate a person's genomic map against a clinical decision.

Genomics represents a cutting edge and innovative field with strong transformative capacity. By directing appropriate investment in the infrastructure to acquire and share data obtained through genomic profiling we sure are on our way to a 'genomic revolution'.

(Image Credits: discoveryeye.org)

**About The Author:**

Samarth Jain is the founder & CEO of Positive Bioscience. Samarth has Majored in Economics at Urbana-Champaign, University of Illinois. In USA, while working on an assignment on Wall Street, Samarth came across upcoming Genomics technologies and how it was revolutionizing the practice of medicine there. He was impressed by the way genomics was improving outcomes in cancer. Along with Dr Meetha Medhora (expert in genomics from University of Wisconsin) and Bapsy Jain (CFO) as co-founders Samarth started Positive Bioscience in October 2012.