

Chapter 14

Personalized Medicine in the Era of Genomics

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ABSTRACT

Personalized medicine is a model that aims at customizing healthcare and tailoring medicine according to an individual's genetic makeup. It classifies individuals that differ in their susceptibility to a particular disease or response to a particular treatment into subpopulations based on individual's unique genetic and clinical information along with environmental factors. The completion of Human Genome Project and the advent of high-throughput genome analysis tools has helped in building and strengthening this model. There lies a huge potential in the implementation of personalized medicine to significantly improve the clinical outcomes; however, its implementation into clinical practice remains slow and is a matter of concern. This chapter aims at acquainting readers with the underlying concepts and components of personalized medicine supplemented with some disease-based case studies, discussing challenges and recent advancements in the implementation of the model of personalized medicine.

INTRODUCTION

Personalized medicine is one of the most exciting topics revolutionizing healthcare industry today. It is a concept that has the potential to transform medical interventions by providing effective, tailored therapies based on the genetic profile of an individual by utilizing vast information contained in our genetic code. It simply means studying the person in the framework of disease rather than just studying the disease itself. The kind of genetic makeup a person has, the kind of protein produced in them and bio-signatures or biomarkers present in their genome are considered while tailoring treatment for an

DOI: 10.4018/978-1-5225-0427-6.ch014

individual. Offering right drug to the right disease at the appropriate time with the optimal dosage is what is required to improve clinical outcomes and this is what personalized medicine stands for (The Case for Personalized Medicine, 2014)

Traditionally healthcare industry followed a reactive approach to treatment, in which the treatment only begins after signs and symptoms appear. The same prescription is given to different people suffering from the same disease. The physicians only considered family history, social-economic and environmental factors. As a result, every year large number of people die due to severe effects of the medicine and even larger gets hospitalized from adverse effects of medication (ADR-Adverse Drug Reaction). According to a report, patient's response rate to medicines can be as low as 20% depending upon the drug (range= 20-75% response rate). The pharmaceutical industry is still developing medicine based on the observation and the mechanism of the disease. Thus, just treating the disease and not the person.

The modern medical treatment focusses on patient's disease to investigate exactly the state and the pathophysiology of disease, in order to "select" the most appropriate medication. "One drug size fits all" paradigm has lost its validity and it is also clear now that the drug response varies from patient to patient. Genetics play an important role in individual's response to a drug and in deciding optimal drug dose. So we have a model that takes into consideration all the aspects affecting a person's state of health from genetics to lifestyle, termed precision or personalized medicine. The following quote can easily summarize the concept behind personalized medicine *It's far more important to know what person, the disease has than what disease the person has* (Hippocrates of Cos (c. 460 BC – c. 370 BC)).

The development of personalized medicine as an approach to medicine, and as a new dimension in medicine, took off when the human genome was sequenced in 2001. A tectonic shift has started since then which is though subtle and perhaps imperceptible for ordinary individuals, but it is bound to influence the entire backdrop of how we look at our physical and emotional well-being. Modern techniques and the completion of Human Genome project have made it possible to understand the genetic part of the diseases and helped in making significant advancements in the drug development process. Personalized medicine is the new paradigm shift in the field of drugs or medicine which formulates treatment or medicine following genetic testing, proteome profiling, and metabolomic analysis to identify the impact of genetic variations on drug response. However, personalized medicine also requires doctors to collect the patient's personal information including the genotype, so as to understand the nature of the particular patient. Based on which the doctor will decide the most suitable medical treatment for that particular patient.

In particular, each person has different ability of drug metabolism owing to the activity of their metabolic enzymes. For instance, if a person has very weak enzymatic activity, the drug will not get completely metabolized and its concentration in the blood increases. Contrary to this, if a person has very strong enzymatic activity towards a particular drug, the drug gets metabolized very quickly and its concentration in the blood would remain low. In simpler words, when several patients are prescribed the same dosage of a drug, the actual effect of the drug would vary depending on its enzymatic activity and thus its genotype. Based on this observation, a new process of drug development requires studying the effectiveness of the drug against the genotype, including the adverse effects.

The whole genome is sequenced using next generation sequencing methods and the obtained genomic reads are then filtered, aligned and variants identified. Variants and Mutations are then annotated using publically available bioinformatics resources. Personalized treatment could then be suggested based on the biomarkers present and other molecular aspects.

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