ISSN (Online): 3139-7689

Available online: www.ijsrmes.com

International Journal for Scientific Research in Modern Engineering and Science

Published in Conference Special Issue: Vol. 1, Special Issue 1, pp. 541–546, 2026

PERSONALIZED MEDICINE: THE FUTURE OF HEALTHCARE

Shipra Jha [1*]

[1]*Department of MBBS, Shri Shankaracharya Institute of Medical Sciences, Bhilai.*

Abstract

Abstract: Personalized medicine refers to the treatment of the patient by considering the genomic sequences of that patient and providing essential therapy accordingly. Advancement in this field has led healthcare professionals to treat diseases based on the genomic sequence of an individual. This review aims to explain the advantages of precision medicine and healthcare along with giving implications on how with the advancement of the modern medicine, it has been beneficial in conquering diseases. Diseases such as different types of cancer, cardiac problems are nowadays being treated by first sequencing the DNA of a person following with drug administration. Adverse drug reactions are a major challenge being faced by many patients but by the application of precision medicine, it can be lessened or even eliminated. This paper has aimed to explain how certain genes break down drug and how it can influence the adverse events faced by the patients. Most of the clinical trials in the past were done on the European men, due to which drug dosage that are standard, are in accordance to their bodies and metabolic activities. Asian patients are devoid of certain genes that metabolize drugs and thus often do not respond well to the drugs prescribed to them. This review presents the examples and the milestones that are achieved by personalized drug therapy and aims to make people aware of it so that what is now available as 'one size fits all' becomes more tailored for the people, with lesser side effects and more advantages.

Keywords: *Personalized medicine, precision medicine, genomics, cancer.*

* *Corresponding author*

1. Introduction

Genomic sequence of a patient is the way the DNA bases are present in that patient's genome, where genome refers to the complete set of genetic material. Before genes were discovered, scientists knew that there was a factor present in the body of a human that makes the children resemble their parents. When resemblance is talked about in today's era, it does not only refer to the outer appearance. Genes have a huge role in determining how an individual responds to the environmental changes. It is widely known that certain diseases tend to run in families due to inherited genetic variations. Risks of a patient to develop diseases like cancer and diabetes are often linked to the patient's family history. Some people are more likely to develop certain diseases, and every individual responds differently to the same drug provided. A patient may develop adverse reactions to a fixed dose, simultaneously another patient may not at all respond to the same drug given in the same dose.

Genes are the inherited units that are passed down from the parents to their offsprings leading to shared biological traits. Genes are present on the chromosomes of a person. Chromosomes are made of a double helical structure

called DNA (deoxyribonucleic acid). A child gets 50% of the genes from the mother and the other 50% from the father. The mother/father in return had their genes inherited from their own parents. This is why healthcare workers are likely to ask for the family history of diseases to a patient to conclude the susceptibility of that patient for certain diseases. A person has trillions of cells, and each cell consists of 23 pairs of chromosomes. Modern studies have revealed that there are around 20,000 to 21,000 genes in the human body, out of which approximately 1-2% of the whole genome codes for protein. Everyone has different genes which brings about the diversity among humans, in addition to that different individuals show different response to the same drugs administered because of these variations.

Pharmacogenomics is a crucial component of personalized medicine and focuses on the study of genetic variations that can impact the drug metabolism, efficacy and toxicity. It combines pharmacology and genetics to understand the pharmacokinetics and pharmacodynamics of drugs. Clinicians, with the help of pharmacogenomics, can identify genetic variations which will allow them to make better drug and dosage selection. Genetic differences among individuals determine whether a person will show poor effect, good effect or adverse reaction to a drug.

Cytochrome P450 enzyme family consists of drug-metabolizing enzymes responsible for metabolism of most of the drugs used clincally[2]. Variations in this family of genes determines how rapidly or slowly a person will metabolize the drug. For instance, if a person is a poor metabolizer and a drug is given in the standard dosage, the patient is likely to suffer from overdose and adverse side-effects, whereas if a patient is a rapid metabolizer, the standard dosage won't be sufficient to bring about the desired results.

Similarly, BRCA 1 and BRCA 2 genes, when mutated increases the risk of cancer. In the study done in 1994[3], BRCA1 gene was established as the susceptible gene for breast cancer. If mutation is found in a woman, her chances of breast and ovarian cancer increase. This could be decreased through genomic sequencing by determining the approximate chances of breast cancer in an individual as was in the case of a well-known example of Angelina Jolie[4]. She found out that her chances of breast cancer were approximately 87% through genetic testing, so she went through bilateral mastectomy, which reduced her risk of cancer to approximately 5%. Therefore, with genetic testing, risks of diseases with high mortality rate can be managed. A patient with a specific mutation inherited can learn to manage the risks of diseases by getting genetic testing and precision treatment.

Modern gene sequencing technologies have made personalized treatment increasingly accessible for patients at higher risk of genetic diseases. Adverse reactions will be minimized as any drug before administration will be largely tested for compatibility with the genome of a patient.

Conventional treatments focus more on the general disease characteristics rather than biological differences among the individuals. Clinical trials are held where drugs are tested on large populations, and the drug is then given to a patient for that disease. A trial-and-error approach is considered where when a patient shows no improvement or adverse events, the drug given, is switched to a different drug. Although the age, height, weight, and other general information of a patient is considered, conventional medicine mostly fails to acknowledge the genetics behind it.

Therefore, by integrating genomic information into conventional clinical practice, modern healthcare will be transformed for the better. Identifying genetic variations that influence drug metabolism will help the clinicians to move towards a better healthcare practice, that is, personalized medicine. Therapies will be tailored to an individual, thus, more effective with fewer reports of adverse reactions. Thus, the future of medicine will significantly be improved by the clinical application of personalized medicine.

2. Methodology/Approach

This paper presents a review of the concepts of Personalized Medicine. Academic databases such as PubMed and Google Scholar were used. Keywords used for searching the information were "personalized medicine," "precision medicine," "pharmacogenomics," "clinical applications of personalized medicine." Articles explaining the concepts of pharmacogenomics and the clinical applications of genomic medicine were selected and reviewed. The selected literature was analyzed and summarized to provide a clear overview of the topic.

The objective of this review paper was to present a broad understanding of how conventional medicine often fails to consider the genetic variation of an individual and relies solely on the "one size fits all" approach. Through the understanding and future implementation of personalized medicine, patients are likely to be well treated. Thus, this review aims to highlight how personalized medicine can help improve drug therapy and reduce adverse reactions.

3. Results and Discussion

Modern advancements have demonstrated that variations in the genetics of an individual significantly influence drug response, efficacy, and toxicity. Traditional medicine is often prescribed primarily considering the characteristics of the disease rather than individual genetic difference. Genetic differences dramatically alter the therapeutic outcomes. Several clinical examples have been documented that supports the usage of personalized medicine.

Pharmacogenomic variation influences the effectiveness of hormonal therapy in breast cancer. Tamoxifen, a widely used drug in the treatment of estrogen receptor-positive breast cancer, requires the activation by the enzyme that is encoded by the gene CYP2D6 to convert into its active metabolite, endoxifen[6]. If an individual has reduced CYP2D6 activity, the individual may produce insufficient levels of the active metabolite resulting in decreased therapeutic effect and higher risk of cancer recurrence. Therefore, testing of CYP2D6 variant may help optimize drug therapy and help the patients benefit more from it.

One of the most widely known examples is of Codeine. Codeine is converted to its active form, morphine, by the action of the enzyme encoded by the CYP2D6 gene. In a case reported in 2006, a breastfeeding mother was an ultrarapid metabolizer of CYP2D6 gene which resulted in unusually high levels of morphine after being prescribed codeine for her postpartum pain. The morphine was transferred to her infant through her breast milk, resulting in toxicity in her infant. Subsequently, the infant died of morphine toxicity at the age of 13 days old[5]. This case

highlights the critical importance of incorporation of pharmacogenomic information into clinical practice before administering drugs.

Abacavir is an antiretroviral drug used in the treatment of HIV infection. Hypersensitivity reactions to abacavir can occur in patients carrying the HLA-B*57:01 allele[7]. These reactions could be life-threatening. The patient may present with fever, rash, gastrointestinal disturbances, and respiratory complications. Genetic screening for the gene HLA-B*57:01 allele prior to initiating the abacavir therapy has been shown to significantly reduce the adverse reactions and it is in fact now recommended as a routine clinical practice[7].

An example of application of precision medicine was observed in cancer therapy of a 55-year-old woman with metastatic breast cancer. She underwent genomic sequencing of her tumor where it was revealed that she had an amplification of the ERBB2 gene, also known as HER2[8]. Due to this alteration, she became an eligible target for Trastuzumab drug therapy. Based on her genomic sequence information, the treatment was significantly improved for her, which demonstrated how precision medicine is essential for oncology treatments. Tailored treatments according to the molecular characteristic of the tumor will result in lesser side-effects of the drugs used in cancer treatment.

Another anticancer drug, Irinotecan is metabolized through glucuronidation by the enzyme encoded by UGT1A1 gene. Individuals carrying the UGT1A128 polymorphism have reduced enzyme activity leading to impaired drug metabolism and accumulation of the active metabolite SN-38[9]. This results in these patients being at a higher risk of severe adverse reactions. Therefore, pharmacogenetic testing of the UGT1A1 variant can help guide the dosage of the drug administered.

A very important example highlighting the clinical relevance of precision medicine in the administration of the antiepileptic drug Carbamazepine. Certain individuals carry the HLA-B*15:02 allele and thus have an increased risk of developing severe cutaneous adverse reactions such as Steven-Johnson syndrome to the drug carbamazepine[10]. This allele is particularly frequent among the Asian population. Traditional medicine has historically been based primarily on populations of European ancestry. The data used for the clinical samples largely come from individuals from the European descent, while Asians and Africans and others alike remain underrepresented[11]. This imbalance creates significant problems as the underrepresented population is likely to suffer through more and severe adverse reactions. Additionally, women have historically been underrepresented in clinical trials, resulting in drug dosage being optimized primarily for male physiology thus making them more susceptible to drug overdosage because the same drug concentration given from the male-dominated clinical trials produce higher blood drug concentration in women[12]. This is why personalized medicine is essential as it considers the individual differences and is not biased towards any region or gender as it is tailored for every type of individuals separately.

4. Conclusion

Personalized medicine represents a shift from the traditional therapeutic approach by incorporating genetic, and individual differences. Evidence from modern studies have shown that genetic variations are necessary to be studied before therapy as it can substantially influence drug metabolism and adverse drug reactions. Studies have

highlighted how most clinical trials are done on European men due to which the underrepresented populations are likely to get the wrong-dosage or suffer from more hypersensitivity reactions. With the help of precision medicine, more diversity in the drug industry could be acknowledged, leading to safer and more effective treatment strategies facilitating all individuals without any bias.

With increasing advancements in the field of modern technology and the integration of pharmacogenomics testing into clinical practice, personalized medicine has the potential to improve drug safety, emerging as the future of healthcare.

References

[1] Pertea M, Shumate A, Pertea G, Varabyou A, Breitwieser FP, Chang YC, et al. CHESS: a new human gene catalog curated from thousands of large-scale RNA sequencing experiments reveals extensive transcriptional noise. Genome Biol. 2018;19(1):208. doi:10.1186/s13059-018-1590-2

[2] Evans WE, Relling MV. Pharmacogenomics: translating functional genomics into rational therapeutics. Science. 1999;28(5439):487-491. doi:10.112/science.286.5439.487.

[3] Miki Y, Swenson J, Shattuck-Eidens D, Futreal PA, Harshman K, Tavtigian S, et al. Science. 1994;266:66-71.

[4] Evans DG, Barwell J, Eccles DM, Collins A, Izatt L, Jacobs C, et al. The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services. Breast Cancer Res. 2014;16(5):442.

[5] Koren G, Cairn J, Chitayat D, Gaedigk A, Leeder JS. Pharmacogenomics of morphine poisoning in a breast-fed neonate of a codeine-prescribed mother. Lancet. 2006;368:704.

[6] Goetz MP, Rae JM, Suman VJ, et al. Pharmacogenetics of tamoxifen biotransformation is associated with clinical outcomes of efficacy and hot flashes. J Clin Oncol. 2005;23:9312-9318.

[7] Mallal S, Phillips E, Carosi G, et al. HLA-B5701 screening for hypersensitivity to abacavir. N Engl J Med. 2008;358:568-579.

[8] Slamon DJ, Leyland-Jones B, Shak S, et al. Use of chemotherapy plus a monoclonal antibody against HER2 for metastatic breast cancer that overexpresses HER2. N Engl J Med. 2001;344:783-792.

[9] Iyer L, Das S, Janisch L, et al. UGT1A128 polymorphism as a determinant of irinotecan disposition and toxicity. J Clin Oncol. 2002;20:3473-3478.

[10] Chung WH, Hung SI, Hong HS, et al. Medical genetics: a marker for Stevens-Johnson syndrome. Nature. 2004;428:486.

[11] Popejoy AB, Fullerton SM. Genomics is failing on diversity. Nature. 2016;538(7624):161-164. doi:10.1038/538161a.

[12] Zucker I, Prendergast BJ. Sex differences in pharmacokinetics predict adverse drug reactions in women. Biol Sex Differ. 2020;11(1):32.