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Precision medicine: A primer

Matthew NO Sadiku¹, Yonghui Wang², Suxia Cui³, Sarhan M Musa⁴

¹⁻⁴ Roy G. Perry College of Engineering, Prairie View A&M University, Prairie View, TX 77446, USA

Abstract

Traditionally, most medical treatments are designed for the "average patient," as a one-size-fits-all-approach. In contrast, precision medicine (PM) is an emerging field in medicine which refers to the tailoring of medical treatment to the individual patient. Because it deals with individuals, PM is highly personalized. Widespread adoption of PM may have a serious impact on American economy and competitiveness. This paper provides a brief introduction to PM.

Keywords: precision medicine, personalized medicine, predictive medicine

Introduction

Today, most medical treatments are designed for average or "typical" patients. Most doctors today still prescribe therapies based on population averages. But experience has shown that treatments that work well for some patients may not work for others. Physicians need tools that will help them provide the best care by incorporating many factors that affect each patient. This is where precision medicine comes in.

Precision medicine (PM) is a medical model that proposes the customization of healthcare with medical treatments, tailored to the individual patient. The word "precision" in PM means both "accurate" and "precise." Precision medicine is also known as personalized medicine, individualized medicine, or predictive medicine although there are differences between the terms ^[1]. Some regard PM as a new name for molecular medicine. Precision medicine matches each patient with the treatment that will work best for them. It takes individual variation in our genes, environment, lifestyle, and other individual factors into account. This is in contrast to a one-size-fits-all approach, in which disease treatment is developed for the average person, ignoring the differences between individuals.

The announcement of Precision Medicine Initiative (PMI) by US President Barack Obama in January 2015 during his State of the Union address has brought PM to the forefront for health care providers, researchers, regulators, and stakeholders. Since the launching of PMI, PM has created immense possibilities for breakthroughs in medicine.

The medical community is now at the transition from the era of evidence-based medicine to the era of precision medicine [2]

Basic Features

PM is about prediction and diagnosis of health with a great precision at the molecular level taking into account genetic variations at a personal level. It takes into account individual variability in genetic and environmental factors and leads neither to undertreatment nor to overtreatment. Precision medicine has the following attributes [3, 4]:

- 1. An understanding of the etiology and pathogenesis of disease
- 2. The ability to detect specific causal factors

- 3. The ability to specifically treat the root causes(s) effectively
- 4. It is being driven by advances in science, genomics and other "omics."
- 5. PM is based on widespread generic testing and integration of the genomic data with clinical information.
- 6. The delivery of targeted treatment to the patient at an appropriate time.
- 7. PM offers more targeted and cost-effective care.

Properly combining observational data with experimental data can facilitate PM. A model of precision medicine approaches is shown in Figure 1 [5].

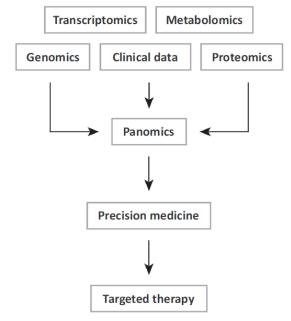


Fig 1: A model of precision medicine approaches ^[5].

Enabling Technologies

Tools that are commonly employed in PM include molecular diagnostics, imaging, and analytic. AI techniques are used in precision cardiovascular medicine to understand genotypes and phenotypes in existing diseases. Machine learning algorithms are used for genomic sequence. Big data

is also required to manage the huge medical data.

Applications

These are some of the ways precision medicine is making a significant different ^[6]:

- Precision oncology is the branch of precision medicine that addresses cancer in precision medicine. Cancer treatment takes many forms: surgery, radiation therapy, chemotherapy, target-based therapy, and immunotherapy. In several areas of oncology, PM is the current standard of care. PM approaches cancer treatment and prevention by taking patients' individual variability in genes, environment, and behavior into account. Cancer is caused by a wide range of cell mutations. Identification of these mutations has led to the development of effective drugs. An overview of PM in oncology is shown in Figure 2 [7].
- Tracking Infectious Diseases: Attempts to tailor disease treatment to individuals based on their DNA have met with equivocal success. Looking at the DNA of germs can help track disease outbreaks when they occur. Knowing exactly which germ is making their patient sick can help doctors determine the treatment that will work best.
- Cigarette Smoking: This is a major global health problem. Smoking tobacco is the leading cause of preventable death worldwide. By stopping smoking, the risk of mortality is greatly reduced. Identification of what contributes to smoking cessation is important for understanding nicotine dependence. To implement PM for smoking cessation treatments, we need meta-analyses of diverse populations [8].

Other applications include diabetes, leukemia, cystic fibrosis, hearing loss, obesity, cardiovascular disease, and Alzheimer's disease.

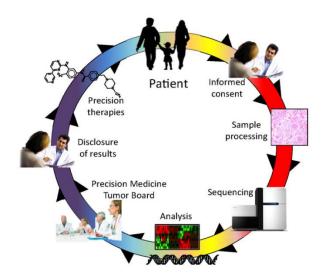


Fig 2: An overview of precision medicine oncology [7].

Benefits

Potential benefits from PM range across all aspects of medicine: research, prevention, diagnosis, treatment, and cure. Precision medicine promises to remake the healthcare industry and change the paradigm for drug development. It will transform the entire pharmaceutical value chain. It will improve how physicians detect and treat diseases. It can help physicians tailor medicines of the specific needs of

individual patients [9].

Challenge

In order for precision medicine to realize its potential, the patient's history, environment, and lifestyle must also be taken into account. PM raises significant concerns about patient privacy. Since privacy is a major social value, the potential consequences of privacy breaches can be significant. It is difficult to manage such sensitive medical data while ensuring patient privacy.

A big bottleneck for PM is computational. PM faces a daunting computational challenge in supporting the genomic, image processing, and analytics needed for millions of patients. Computational methods play a crucial role for assisting physicians in their decision making. Research findings take too long to be implemented into clinical practice.

Another challenge is the misalignment of interests of the patient, physician, health system, and health insurance. Managing these interests in a cost-effective manner may be difficult. Regulatory issues may be a barrier to the progress of PM ^[10]. Taking care of these challenges is critical to the success of PM and will result in limitless advances in PM.

Conclusion

Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual characteristics. It is targeted to the needs of a patient based on his/her own genetic or psychosocial characteristics. It involves understanding how factors from the environment interact with genetic variations to influence an individual's health. Some see PM as the golden highway from disease identification to cure.

Some institutions of higher learning across US have established courses on PM. PM offers a tool to revolutionize how medicine is taught and practiced. As the society looks to the future, PM aims to meet the medical needs of all people, regardless of their gender, race, age, religion, or disability [11]. PM is poised to change the practice of medicine. More information on precision medicine can be found in [12, 13].

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