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Precision medicine: Present and future

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Precision medicine is a medical model that proposes treatment of an individual based on his or her genetic content. Thus various and appropriate diagnostic tests are needed in a patient to find his/her genetic contents. Various tools employed in precision medicine may include molecular diagnostics, imaging, and analytics.

There is no universal definition for the term "precision medicine," although probably, the most widely accepted is the one provided by the US National Institutes of Health (NIH) that defines it as ''an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person'' [1]. This at times is used synonymously with personalized or individualized medicine.

Precision medicine, as defined by the National Institutes of Health's Precision Medicine Initiative Working Group, is "an approach to disease treatment and prevention that seeks to maximize therapeutic effectiveness by taking into account the individual genetic, molecular, environmental, and lifestyle differences."

It is estimated that only around 40% of the drugs, we prescribe, are effective or appropriate for the concerned patients. Today we practice trial and error medicine which does not consider in detail, person to person variability which is the main reason for the differences observed in effectiveness of the drugs on different individuals.

Precision medicine looks at the root cause of an illness, rather than addressing the symptoms alone. It takes into account genotypic and phenotypic factors, that is individual variability in genes, environment, and lifestyle for each person [2]. Thus, it is involved in identifying potential biomarkers and utilizes targeted therapy.

Each individual is an island and now since the molecular complexity of disease has been deciphered, the role of genetics, environment, and epigenetics is clearly telling us that all patients need to receive treatment designed for their respective physiology and body type. There are several targeted therapies that work only on patients who have a specific genotype or genomic variation, hence, particular patients need to be adequately tested before the therapy is prescribed.

In various clinical and nonclinical situations genetic testing is undertaken to find out changes in chromosomes or genes [2], for example, diagnostic testing, predictive and pre-symptomatic testing, carrier testing, pre-implantation genetic diagnosis, prenatal testing, new born screening, pharmacogenomic testing, etc.

With the advent of precision medicine there is a paradigm shift in medicine. By its use appropriate strategies can be adopted in individual patients depending on their genetic constitution. Thus suitable and appropriate drugs can be used in patients. It avoids unnecessary use of drugs which are not only going to be in-effective in a particular patient but may in fact be doing harm to him/her.

Pharmacogenetics provides information that can help predict how an individual will respond to a medication. Changes in certain genes affect pharmacodynamics and pharmacokinetics [3]. Identifying these changes makes it possible to identify patients who are at increased risk for adverse effects from drugs or who are likely to be non-responders. Pharmacogenomic testing allows healthcare providers to tailor therapies by adjusting the dose or drug for an individual patient [4].

As on date there are only 30 approved pharmacogenomic drugs in use in oncology, which are benefiting only a small number of patients suffering from malignancy. The effectiveness of these drugs is also limited due to intratumor heterogeneity [5, 6] and resistance mechanisms which are poorly understood [5, 7]. The time elapsed between obtaining patients biopsy up to starting personalized treatment for cancer is much more than that is acceptable to clinicians. The delay in starting personalized treatment by not more than 10–14 days would be acceptable to patients and physicians [8].

The situation is evolving rapidly, and the penetration of Next Generation Sequencing (NGS) [9] technologies in the clinical realm has prompted the development of

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new laboratory guidelines and standards for NGS data production, analysis, and sharing. These efforts have been made by a variety of groups and institutions around the world, which resulted in the publication of numerous partially overlapping guidelines, some extremely general and others focusing on specific diseases, or specific steps in the process, such as the return of results to patients and clinicians or the development of specific bioinformatic pipelines for NGS data analysis.

Precision medicine will offer a possible answer by reducing the cost of hospital stay, reducing the overall financial, physical, and psychological costs of the try and test approach of medicine. It is akin to the direct benefit transfer (DBT) scheme of Govt of India for transferring the subsidies amount direct to the linked bank accounts of intended individuals through unique genetic identification by way of Aadhaar biometrics thereby achieving precise and targeted delivery of benefits without any side effects of wastage of public money. Likewise in precision medicine the intended therapeutic agent is delivered directly on to the targeted cells or tumor without causation of collateral damage to normal cells/tissues thus reducing morbidity and mortality significantly.

Advancement in precision medicine is likely to improve the health of general population by application of individualized treatment and public health approach. The technology used for precision medicine will not be limited to genes and disease. The same technologies and big data that are propelling precision medicine forward are leading to a new era of precision public health [10]. In case of public health, application of methods and technology to study various diseases, their pathogens and susceptibility at community level will help in developing precision to prevent the diseases.

In the years to come the precision medicine is going to become the main approach to treatment as the cost of genetic sequencing and data is coming down gradually. The use of artificial intelligence (AI) to perform algorithm analysis and in decision making for optimal drug selection, for more effective drugs, for large number patients in lesser time, is further going to revolutionize it thus making precision medicine truly a personalized medicine. In the meantime countries have to bring out enabling legislations for DNA usage policy in all encompassing and holistic manner. The United States has already rolled out Precision Medicine Initiative (PMI) [1] since 2015 and is now renamed as “All of Us” to take the scope forward in a big way by federal funding and participation of number of reputed institutions. In the United Kingdom, there has been a similar commitment to creating more precise treatments through the Medical Research Council’s Stratified Medicine Initiative and through key partnerships between industry and the UK’s National Health Service. These investments highlight the promise of precision medicine to revolutionize many other fields of medicine in the decades to come.

In a country like India the initiative of precision medicine may be considered as an overambitious program since lack of proper nutrition and poverty are major killers. But being a diverse country with nearly 4000 population groups and a significant percentage of consanguineous marriages has a heavy burden of inherited diseases driven by unique genetic characteristics in the subpopulations. With 1.3 billion people, the absolute number of patients suffering from diseases in which genetics plays a role is significantly large. To an outsider India may appear confusing, for example, so many people in abject poverty on one end and so many billionaires/multimillionaires on the other side, underdeveloped infrastructure on one side and one of the top four countries in space and nuclear science field on the other end, producer of largest number of doctors and paramedics in the world, state of the art health care facilities and yet affordable quality health care is out of reach to people in vast rural hinterland, finest software manpower provider to the world yet lagging at home front, etc. Add to this young demography and rising and aspiring middle class. However, unlike in the past, the present political leadership in India is leapfrogging by taking quantum strides rather than incremental steps in all spheres of development. Precision Medicine Initiative has also caught the attention of Indian Prime Minister who has announced to put all the effort toward the comprehensive DNA bill of India. India will achieve excellence in this field as well. Just imagine a patient taking a blood or saliva test at a laboratory, and in the next meeting his/her doctor is well prepared with the medical horoscope including the information on chemical compounds or combination of drugs useful or harmful to him/her, based on the tried and tested genetic test report. In India Precision Medicine, though in nascent stage, is being practiced in several specialties like oncology, cardiology, psychiatry, and diabetology. However, oncology seems to be the specialty in which precision medicine is most advanced.

Lot of investment is required in the field of precision medicine like infrastructure for producing, sharing, and storing the data which needs high throughput computing systems. Investment also is required for educating general public, insurance companies, and treating clinicians. Unless proper education is given to various stakeholders and interdisciplinary research is encouraged or done, precision medicine cannot be enabled.

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Author Contributions

Piaray Lal Kariholu – Conception of the work, Design of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Sanjeev Kumar – Conception of the work, Design of the work, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

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Conflict of Interest

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Data Availability

All relevant data are within the paper and its Supporting Information files.