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Review Article

Precision medicine- Where do we stand now and what lies ahead of us?

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ABSTRACT

Precision medicine is an approach to disease prevention and treatment that aims at maximizing therapeutic effectiveness by analysing the genetic, environmental and lifestyle differences in an individual. Pharmacogenomics is an important part of precision medicine. Globally, precision medicine is tried for the prevention and treatment of various diseases but cancer appears to be the most invested entity. But most significant fact is that not just the non-communicable chronic diseases but precision medicine has been tried in the COVID-19 pandemic as well. The concept of precision medicine requires collaboration between the various stakeholders. This includes providers or the patients, health care workers, diagnostic partners, payers, regulatory bodies and IT partners. Currently, the concept of Precision medicine in India is in the early adoption stage.

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1. Introduction

When any patient is given an option to choose between a special treatment that is carefully selected just for them and a treatment that works for most of the people, no doubt most would choose something that is specially meant for them. Today there is a need to have faith on this human instinct and invest our efforts, knowledge, intelligence and funds on this idea. This is in fact the basis for the advent of precision medicine.

Precision medicine is an approach to disease prevention and treatment that aims at maximizing therapeutic effectiveness by analysing the genetic, environmental and lifestyle differences in an individual. The commonly practiced trial and error medicine is more convenient but it may not be successful in more than half of the cases. Precision medicine avoids the usage of drugs that are going to be ineffective in a particular patient and also avoids the possible harm that may result by using such drugs.

But this does not mean a new drug or device is designed for each patient. Rather precision medicine involves sub classifying individuals with similar or same symptoms or diseases depending on the variations in the genetical make up, lifestyle and environment. It is followed by selecting the best treatment or preventive modality out of the available options and modifying it accordingly for that particular individual.^{1,2}

The term 'precision medicine' is used interchangeably with 'personalised medicine' throughout the literature. There is significant amount of overlap between the two terms. In fact, there are sources suggesting that 'personalised medicine' is the older term for 'precision medicine'. But as the term 'personalised medicine' might be misinterpreted as developing treatments and preventions uniquely for each individual, the term 'precision medicine' is preferred. Also, misinterpreting 'precision medicine' with the 'p drug concept' should be avoided as they are two different concepts.²

Pharmacogenomics is an important part of precision medicine. Genetic variations in individuals is known to

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significantly influence the drug response. A knowledge about the genetic make-up of the individual thus helps in predicting the therapeutic outcome to a greater extent. But it is important to note that the variations in therapeutic response cannot be just attributed to the genetic makeup of the individual. Hence precision medicine takes lifestyle and environmental factors into account.²

Historically, the concept of precision medicine dates back to 1998 where it had its first mention in a monograph title. But the basic idea is credited to a Canadian physician, Sir William Osler (1849-1919) who recognised that no two individuals respond similarly under an abnormal condition like presence of a disease. In the United Kingdom, an investigation on the state of application of genomics in medicine was conducted in 2009. In 2012 it was followed up by an action plan on application of genomics in healthcare. Whereas, in the United States the president's council of advisors on science and technology submitted a report on the priorities of precision medicine. It was followed by the 'precision medicine acts' in 2008 and 2010 with the aim of being available to every citizen. These acts chalked out an action plan and defined programme management. They outlined the role of government agencies and financing. In January 2015, president Obama, in his address to the nation announced the precision medicine initiative (PMI) with a funding of 215 million dollars. It was followed by the publishing of a cohort structure programme comprising of more than a million patients. It was planned to be followed for 10 years or more.³ In India, the developments in the concept of precision medicine is in the very early stage. It is being tried in certain areas like oncology, cardiology, psychiatry and diabetology. Various institutions across the country are involved in the research works in this area. But most of the research is fragmented and driven by individual researchers and not very well translated to patient care. Funding for the research is again a concern.

Globally Precision medicine is tried for the prevention and treatment of various diseases but cancer appears to be the most invested entity. But most significant fact is that not just the non-communicable chronic diseases but precision medicine has been tried in the COVID-19 pandemic as well. Studies have discovered various inherited and genetic factors accounting for the varied susceptibility of an individual to the SARS COV-2 virus including variations across the Major histocompatibility complex (MHC), blood groups and genes including Tcam2, ACE2, TMPRSS2, CD147/BSG, DSTN, CFL1, CFL2, MUC5B, TERT, DPP4, locus 9q34.2 and locus 3p31.21. Environmental exposure and lifestyle may also explain the individual variation in the susceptibility to SARS COV-2. This explains the role of precision medicine in designing newer clinical trials as well as prevention and treatment of COVID 19.⁴

The concept of precision medicine requires collaboration between the various stakeholders. This includes providers

or the patients, health care workers, diagnostic partners, payers, regulatory bodies and IT partners. India being the home for a diverse population, a wide variation in the genomes of individuals favours the genomic research. Even though there are significant similarities between the Caucasian population, there are significant differences too. Hence genomic study of large number of individuals does not seem to be a vain. For this step of pooling in volunteers to be successful, a detailed scenario of precision medicine and its possible advantages should be made available to the patients in the languages and terms they can interpret, understand and decide.³ In this era where the patients refer the information available on the electronic media and involve themselves in self-diagnosis and self-medication practices, it is important to divert them into thinking differently and motivating them to discuss their symptoms, family back ground, lifestyle and environment with a specialist. It is important to make every patient aware that each one of us are different at the genetic level and the environmental and lifestyles variations make us even more different. This would help the patients in understanding the concept of precision medicine. Emotional and ethical issues related to the patients should be carefully considered and addressed.

The next important stakeholder is the health care worker. When we refer to the term precision medicine, it is not just about the high-tech institutes, super specialists and the expert researchers. The practice should start from the grass root level. The medical officers working at the Primary Health Care facilities should be given informative sessions to begin with. In turn the Auxiliary Nurse Midwives, ASHA workers and other field workers should be well informed. By this we can make sure the information reaches every individual, even a patient living in a remote village with no or little access to newer advances in healthcare and technology. This will ensure well informed and voluntary participation of the patients in the collaboration. When it comes to the healthcare worker, since they are already informed and the patient is well informed, it makes the process of referral to higher centres hassle free and systematic. Necessary arrangements for referral, tracking and follow up should be considered. Patients should be informed about the possible delay in the process due to the various analysis and expert procedures involved to avoid the patient from losing faith over the system.^{1,3}

Another deciding stakeholder in the success of precision medicine collaboration are the Diagnostic partners. It is important to first list, contact and communicate the diagnostic partners. Following this, suitable diagnostic partner should be chosen based on the accessibility, accuracy, reliability, cost and speed of early diagnosis. The ability to handle large number of testing should also be considered prior to the decision.^{1,3}

Currently, most of the funding in India for precision medicine is by individual organisation and research institutes. Whereas, in the United States apart from the initial budget for the PMI, the NIH has awarded 3.7 million dollars to the Washington university School of Medicine to fund an open source database that aims to boost precision medicine. Apart from these, US has many private firms contributing to the development of precision medicine. Hence, we should make attempts on receiving international funding and also make draft plans and allocate some funding from the national level as well. Attempts to involve private firms in funding should also be carried out.^{1,3}

The regulatory bodies play an important role in cementing all the stakeholders together in this collaboration. The regulatory bodies are required to be well informed about the plans and protocols and the need for the development and funding in this particular section. Only with the understanding and co-operation of the regulatory bodies we can expect a smooth initiation, translation, implementation and success of the precision medicine concept.^{1,3}

Last but not least we need assistance from the Information Technology experts. India with its diverse population is expected to produce huge amount of very diverse data over the next 10 years through genomic typing which is the important part of precision medicine. It is important to record, store, organise and access this data for future analysis. It is important to make the data available across the institutions in the country and also globalisation of this information. Decisions should be made on who can contribute and access the data base to avoid misuse of the data. We need to invest on Bio information technology platforms which can handle such huge and diverse data and are capable of formulating algorithms and deriving relations and conclusions from the available data. Reliability, standard, security and affordability should be prioritized.^{1,3}

Currently, as mentioned earlier, the concept of Precision medicine in India is in the early adoption stage. There are no 'acts' similar to that of PMI from the U S. The formal research works related to precision medicine are being carried out individually at various institutes across the country.⁵ Also, it is not surprising if majority of the patients and a lot of Health Care Workers are unaware or have a misconception of the terminology of precision medicine as such. But this can be easily overcome by a few informative sessions. More articles regarding the concept need to be encouraged, more studies and case scenarios should be published, researchers and research institutes involved in this area should be identified and assisted and collaboration with existing projects should be attempted.

More recently, inspired from the Human Genome Project, the Department of Biotechnology has initiated the Genome India project (GIP) on 3rd January 2020. It aims at collecting 10,000 genetic samples from citizens across

India to build a reference genome. This is a collaboration between 20 institutions across India. It is a project with 238 crore funding. The data is expected to be available to researchers across the world for analysis. This is expected to lay a strong foundation for the development of precision medicine in India.^{6–8}

2. Conclusion

This is bird view of where we started, where we stand, what lies ahead of us and what can be done to conquer the future in precision medicine. In other words, in order to see a more positive and practical picture of future of the precision medicine concept, we need to focus on the little steps and use the available resources very carefully with prior planning and intelligence. The concept of precision medicine is undoubtedly an expensive affair. But with certain amount of awareness and planning the existing resources can be utilised without having to invest much exclusively. One of the best ways would be collaborating with existing genomic studies and collaborating with the stakeholders and creating awareness. Precision medicine as a whole may appear too much hyped concept since we definitely have grass root level health and nutrition problems to address. But it is still worth the attempt to organise and plan to collaborate the existing resources for the development of precision medicine and watch it develop while we work on our grass root level problems. By this we will not need to question ourselves 'what next?' once the current basic healthcare goals are achieved. It is always better if we have a roof ready to get inside after a long struggle through the stormy sea rather than waiting at the shore wondering what next.

3. Conflict of Interest

The authors declare no relevant conflicts of interest.

4. Source of Funding

None.

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