Review Article

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Personalized medicine and genomics in the contemporary world

Abstract

Personalized medicine (PM) is an evolving branch of life sciences which employs tailoring and development of targeted therapies against specific diseases and conditions. With the advent of molecular diagnostics, it is now possible to obtain the genetic make-up of an individual and zero in on the possible mutation or aberration which might be the cause behind a particular condition. Targeted therapies formulated like this have prospects for both the healthcare providers as well as the pharmaceutical industries in terms of improved medication and increase in market shares and profits. This is a revolutionary approach which could be used to eradicate every medical condition in the contemporary medical dictionary.

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Abbreviations: PM, personalized medicine; CF, cystic fibrosis; PKU, phenylketonuria; CFTR, cystic fibrosis transmembrane conductance regulator

Introduction

Personalized medicine refers to tailoring a particular therapy in accordance with a patient's genomic make-up. As the US Food and Drug Administration puts it "providing the right patient with the right drug at the right dose at the right time". The main aim of personalized medicine (PM), a developing branch of life sciences as a whole, over the years, has been to curtail error rates in prescribing patients with therapies and zero-in on appropriate solutions to problems (diseases) and treat them from the very roots, so as to minimise or totally eradicate chances of recurrence or remission.[1,2]

A normal approach taken by medical professionals in treating a patient involves obtaining phenotypic and demographic information from him/her along with environmental factors which could have played a role in the development of the disease in the first case. Apart from this, owing to recent advances in molecular techniques and especially, with the completion of the Human Genome Project, it is now possible to study the genetic make-up of an individual, sequence his/her genome and get an idea of what possibly could affect a particular gene which would lead to the disease. In short, the root cause of the disease often lies in a person's genetic make-up, prior knowledge of which helps a physician, not only prescribe the correct medication, but also helps pharmaceutical companies come up with more appropriate therapies to the rarest of rare conditions, and also, prevent them from adverse drug reactions.

PM has a little something for both diagnostic laboratories and pharmaceutical companies, which in the long run, would help in the economic stability and ensure a strong grip on market shares for investors. While diagnostic laboratories could use modern sequencing techniques to zero-in on any genetic mutation or defect which could possibly differ from the normal genome map of an individual, provide diagnostic bio-markers which indicate disease development, the pharmaceutical companies could improvise on these and come up with formulations which could treat and cure the condition. This would boost sales and improve profit margins, providing better resources for further research and development. Broadly speaking, PM holds the potential to shift the basis of medical treatment to a more pro-active form of 'prevention' from its reactive counterpart 'reaction'. The 'trial-and-error' method of treatment is slowly getting replaced by a more accurate form of medical remedy, along with revealing alternate forms of therapies for known diseases or drug candidates (which could prove wonders in the growing world of antibiotic-resistance in infectious diseases.[3–5]

Some common applications of PM that have already made milestones in the medical world include cancer therapies and categorizations depending on genetic mutations, which collectively comprise a branch of genomics called 'oncogenomics' a concept of 'pharmacogenomics' has evolved which uses the human's genomic map as a reference to provide a tailored prescription for a drug pertaining to a particular disease; and not to forget, developing drugs for rare conditions (often denoted as Orphan Diseases) with a host of possible genetic mutations to account for. Conditions like these include identifying genes responsible for inherited rare diseases like cystic fibrosis (CF), Tay-Sachs disease, phenylketonuria (PKU) and so on. There exist as many as 15,000 tests for more than 2800 genes sequenced so far for identifying a host of genetic disorders.[1,3–5]

Applications and future prospects

A good example of how PM has helped in tailoring therapeutics in rare diseases would be the FDA approval of the drug Kalydeco (ivacaftor), developed and marketed by Vertex Pharmaceuticals, targeted at a very specific, G551D mutation, of the CFTR (Cystic Fibrosis Transmembrane Conductance Regulator) gene, which is one among 1500 possible mutations leading to CF. The CFTR gene is mainly concerned with production of sweat, digestive fluids and mucus. Those with a mutated gene have impaired muco-ciliary clearance in the lungs which paves way for mucoid secretion and *Pseudomonas aeruginosa* infections (complicated with antibiotic resistance in most cases), impaired secretion of digestive juices as well as salt-water imbalance in the body which mostly proved to be fatal. The G551D mutation is a cause of CF in only 4% of the cases and it deals with regulation of sodium channels to maintain salt and water transport across membranes. Normally, a sweat test would be the first approach to such a case, but owing to molecular genomics, it is now

easy to sequence the genome and predict the 'most-likely' mutation out of them, which leads to the production of a faulty protein and its defective functioning. This gave Vertex the opportunity to develop a drug that could restore the function of the mutated protein and ensure sodium-water equilibrium in the body. This further prevents the build up of mucus in the lungs and avoids risk of bacterial infections as a whole. Kalydeco (approved by the FDA on January 31st, 2012) was the first drug of its kind to have focussed on the underlying genetic defect, rather than concentrating on reducing symptoms and clinical worsening. Patients who have resorted to treatment with this drug have so far given a positive feedback, with the drug successfully improving their quality of life as a whole.[4]

Adequate research goes behind the development of PM as well. It needs intensive background study as well as knowledge of a person's genetic history to work on such cases. But one can surely hope, in due course of time, PM might muster the potential to eradicate every possible disease existing in the contemporary medical dictionary!

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Conflict of interest

The author declares no conflict of interest.

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