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Precision medicine: Modern era for diagnosis of disease.

Punam N. Mungase, Maya Y. Gaikwad*
Student SMBT Institute of D. Pharmacy, Dhamangaon, Nashik
Lecturer SMBT Institute of D. Pharmacy, Dhamangaon, Nashik

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ABSTRACT
Cancer is lethal disease that kills a great number of people each year. Standard treatment such as chemotherapy or radiation are only effective in a small percentage of individual due to illness variability .tumors can be caused by a variety of genetic factors and express a variety of protein depending on the individual. Cancer is devasting disease that takes the lives of hundreds of thousands of people every year due to disease heterogeneity, standard treatment such as chemotherapy or radiation are effective in only subsets of the patient population. Some PPM products are already available to link these difference to an effective drug .it is clear that PPM cancer treatment. However, broader change to the healthcare and insurance systems must be addressed, if PPM is to be addressed, if PPM is to become part of standard cancer care. Precision Medicine also includes the terms like Genomics, genetic sequencing, CRISPR, pharmacogenetics, next generation sequencing, biobanking and biomarkers.

KEYWORDS: Clonal evaluation, Genomic diagnosis, Cancer treatment, Personalized medicine, Precision medicine.

I. INTRODUCTION

Cancer through a broad term refers to the uncontrolled proliferation of cell that are prone to stochastic somatic mutation on oncogens and tumor suppresser genes. These can be inherited but exogenous factors can also introduced them into a person's life over the course of ones lifetime (e.g. carcinogenic chemical). According to report published by the American cancer society there will be 18,98,160 new cancer cases and cancer will claim the lives of 6,08,570 people in 2021. [1]

- **Examples Of Pricision Medicine Biomarker In Cancer-**

Her 2- Human Epidermal Growth Factor Receptor 2.
Bcr –Abl –Pridinitotinib, Imitinib ,Ponatinib
Braf V-Raf –Marine Sarcoma Viral Growth Factor
Alk-Anaplastic Lyphoma Kinase.[2]

- **WHAT IS PRICISION MEDICINE?**
- **Definition Of Pricision Medicine**

Precision Medicine (PM) is a novel medical model that was first investigated in 2011 by National Research Council. Precision medicine proposed area in healthcare system by applying some medical decisions, treatments, practices or products being tailored on specific individual patient.

Tools Applying on Precision Medicines:

- Molecular diagnostics
- Genetic imaging
- Analytics [3]

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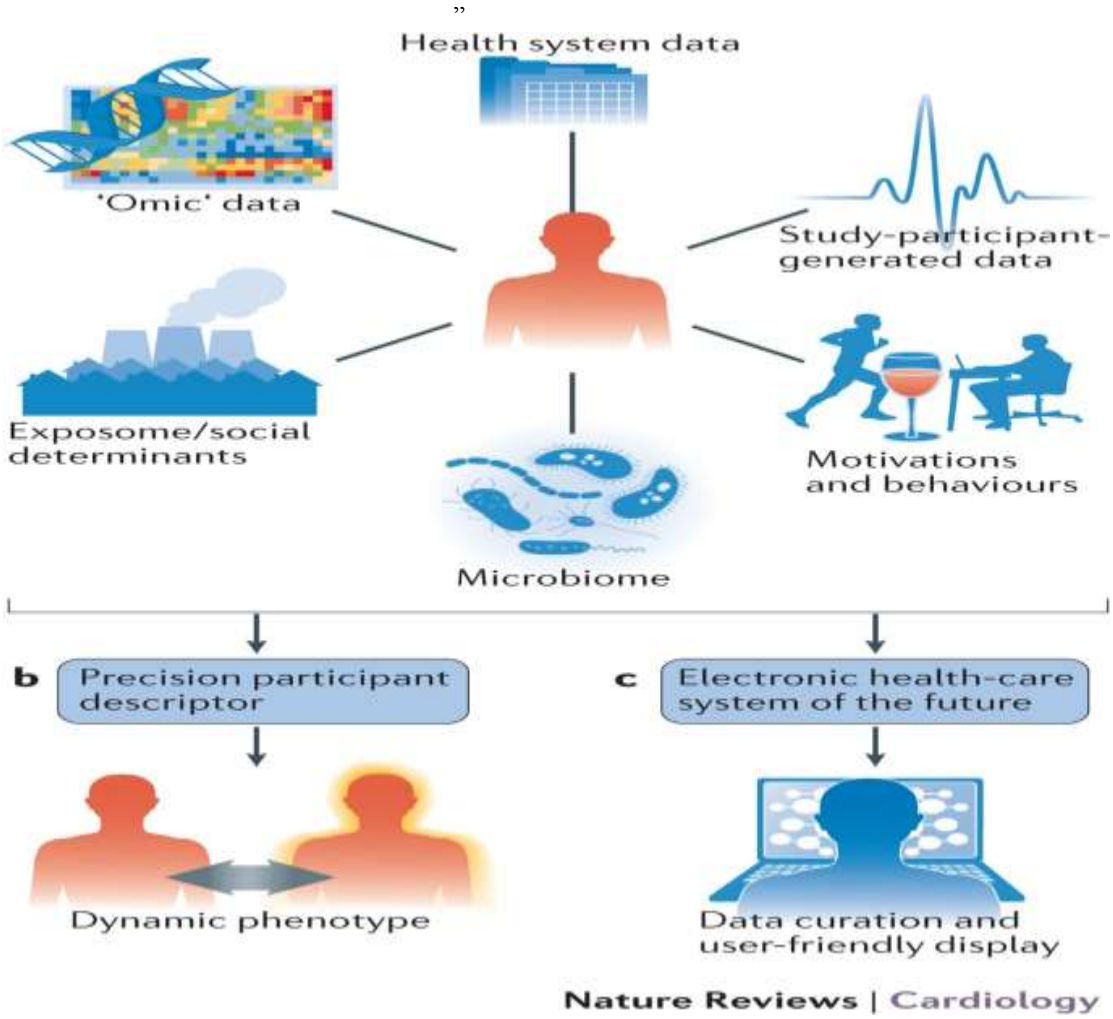

Fig. Precision Medicine in Cardiology

PRICISION HEALTH: IMPROVING HEALTH FOR EACH OF US ALL OF US:

Your gene, behaviors such as exercise and eating habits and environment are all factors that affect your health. The goal of precision health is to protect your health by measuring these environmental factors and acting on them.

UPDATES OF GENOMICS AND PRICISION HEALTH INFORMATION ON COVID-19

We conducted a seroepid etiologic survey from October 22 to December 9,2021,in Gauteng to determine the seroprevalance of SARS-CoV-2 IgG .We also evaluated covid-19 epidemiologic trends In Gauteng that include individual cases , mainly record in hospital deaths and excess death from the start of the pandemic through january12,2022. Epidemiologic data showed a decoupling of hospitalization death from infections while omicron was circulating.

On the 25th February 2016, the white house declared to execute several activities including a health research with one million participants to encourage and promote president Obama's proposal on precision medicine. It hoping to recruit one million volunteers by 2019, collecting such relevant data as medical records, genetic information, lifestyle etc. the aim in 2016 is to recruit 79,000 volunteers. When we come across back recent 10 years, we can say that we have marked a great progress on medical field's cancer, Alzheimer's disease and others. [4]

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ADVANTAGES:

- Increase the chances of doctor to use the patient genetic and molecular information
- Enhance the ability to understand underlying mechanism of disease.
- It helps in preventing diagnosis and treating a range of disease .
- Improved approaches to preventing diagnosis and treating a wide range of disease.

DISADVANTAGES:

- The patient must has to be aware of the family history.
- Time-consuming.
- Fear of genetic discrimination.
- Access and availability of genetic testing
- High cost associated with precision medicine [5]

Examples of Precision medicine application. [1]

Medical Field	Disease	Biomarker	Intervention
Cancer	Chronic myeloid leukemia	BCR- ABL	Imatinib
	Lung cancer	EML4-ALK	Crizotinib
Hematology	Thrombosis	Factor V Leiden	Avoid prothrmbotic drugs
Infectious disease	HIV/ AIDS	CD4+T cells, HIV viral load	Highly active antiviral therapy
Cardiovascular disease	Coronary artery disease	CYP2C19	Clopidogrel
Pulmonary disease	Cystic fibrosis	G551D	Ivacaftor
Renal disease	Transplant rejection	Urinary gene signature	Antirejection drugs
Liver disease	Hepatitis C	Hepatitis C viral load	Direct acting antiviral agents
Endocrine diseases	Multiple endocrine neoplasia type 2	RET	Prophylactic
Metabolic diseases	Hyperlipidemia	LDL cholesterol	Statins
Neurology	Autoimuune encephalitis	CXCL13	Immunotherapy
Psychiatry	Alcohol use disorder	GRIK1	Topiramate
Pharmacogenomics	Smoking cessation	CYP2A6	Varenicline
Ophthalmology	Leber's congenital amaurosis	RPE65	Gene therapy

TODAY USE IN PRICISION MEDICINE

One current use for this is a target therapy to treat a specific type of cancer like HER2 –POSITIVE breast cancer. [6]

USES:

Use information about a person's own genes or proteins to prevent diagnosis or treat disease. In cancer diagnosis precision medicine uses specific information about a person's tumor to help make a diagnosis, plan treatment, find out how well treatment is working.[6]

FUTURE:

Precision medicine offers great opportunity to shape the future of healthcare. While it is currently most sophisticated technology in oncology. Precision medication also has wider, exciting applications beyond oncology and late-stage disease, such as in rare and genetic diseases, it also holds to treating COVID-19.

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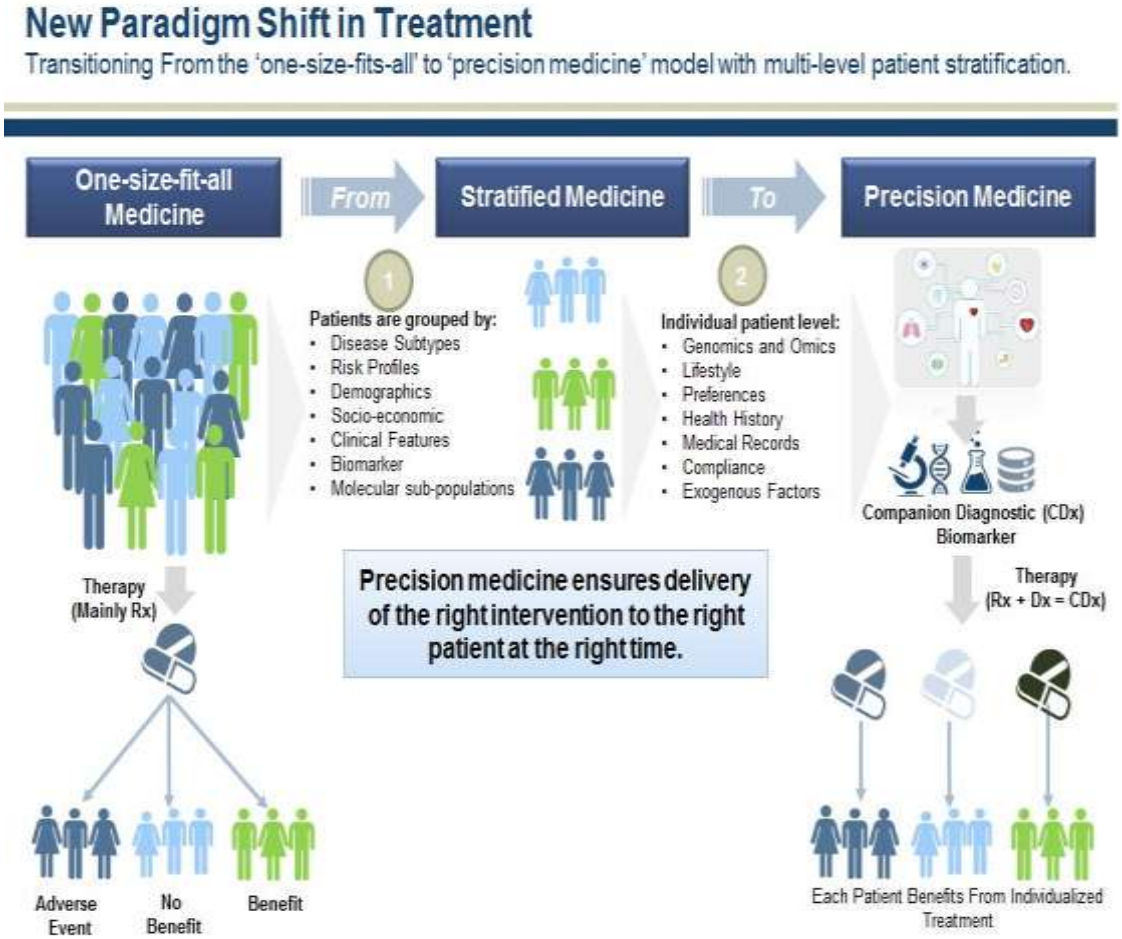

Fig. Modern Era of Precision Medicine

Precision medicine, also generally referred to as personalized medicine, is one of the most promising approaches to tackling diseases that have thus far eluded effective diagnosis, treatments or cures. Cancer is one of the neurodegenerative diseases, and rare genetic conditions take a huge toll on individuals, families and societies.

In 2017, approximately 1.7 million new cancer cases were diagnosed in the United States. Around 600,000 deaths were expected throughout that year, according to the American Cancer Society. The Agency for Healthcare Research and Quality adds that the direct economic impact of cancer is around $80 billion per year is due to loss of productivity, wages, and caregiver requires sap billions more from the economy.

Precision medicine refers to the diagnosing disease, medical treatment to the individual characteristics of each patient. It does not exactly mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease or in their response to a specific treatment. Even though the term "personalized medicine" is also used to express this meaning, that term is sometimes misinterpreted as implying that unique treatments can be designed for each individual. [4]

Key Terms Within Precision Medicine

Genomics

The genome is an organism's entire set of genetic material. DNA includes coding regions, or genes, that oversee the function of proteins, as well as non-coding sequences that perform regulatory functions.

Genetics

A gene is a subset of the genome that codes for a molecule that has a precise function, such as governing a person's eye color, blood type,

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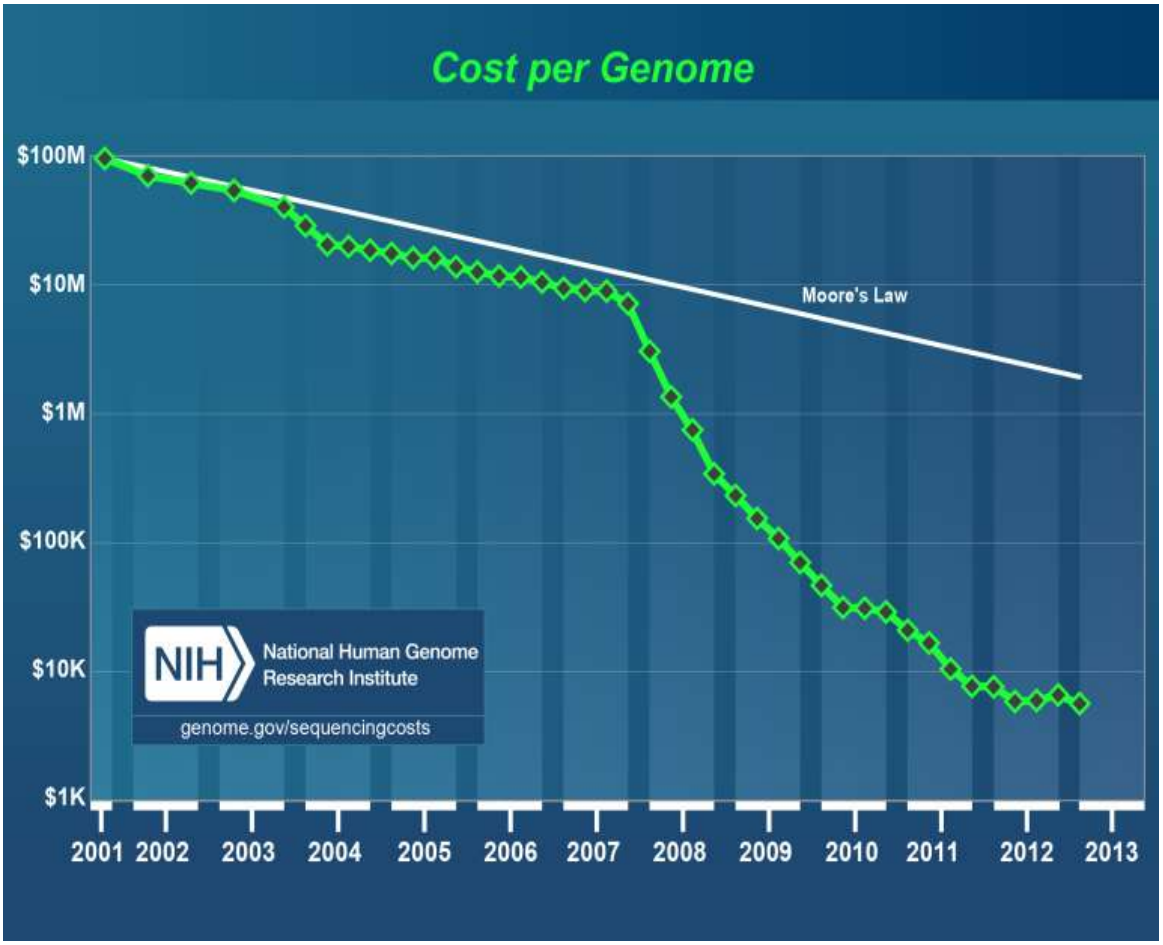
or predisposition for certain diseases. Genes can obtain mutations when passed along through families, resulting in inherited conditions. Variations in an individual's phenotype, or the sum of its observable physical or behavioral characteristics, are due part to how individual genes combine to produce those behavior.

Researchers are still discovering complex relationships between genes and diseases. The rapid advance of genetic research in the early years of the 21st century has been supported by initiatives like the Human Genome Project, which produced a multiple genetic sequencing that is freely available to the public. Scientists can evaluate cancer cells, for example, with this data to understand where and how specific mutations occur and what their impacts might be. [7]

Genetic sequencing

Genetic sequencing, or DNA sequencing, is the process of detecting the order of the four chemical building blocks of DNA (adenine, guanine, cytosine, and thymine) for an individual organism. The order of these chemicals in each strand of DNA dictates and identifies the type of genetic information included in a segment of DNA.

Researchers can recognize which sections of a DNA molecule contain genes and which include regulatory information, allowing them to place differences between individuals with certain traits and those without. The human genome contains around 3 billion pairs of the four chemical bases of DNA, which all collectively provide the "instruction manual" for a living organism.



Genetic sequencing start in the early 1970s, genetic sequencing has become so quick and inexpensive that healthcare providers can usually order genetic testing for patients suspected of having conditions with a genetic component.

The falling prices and rising speed of these tests has been a significant catalyst for

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precision medicine. Interest in genetic testing has become skyrocketed in recent years, with 90 percent of patients in one recent survey expressing some level of interest about receiving a genetic test.

Next-generation sequencing

Next-generation (next-gen) sequencing is a collection of techniques that have further improved the speed and detail of genetic sequencing.

Instead of sequencing an individual's entire genetic code scratch every time, next-gen techniques sequence fragments of an individual's DNA, called "reads," and then use algorithms to compare the results to a DNA library to fill in the gaps. Any differences or mutations can be recognized during the process.

Next-gen sequencing allows laboratories to complete the process more quickly, that helps to meet the growing demand for their services. The techniques are being developed and refined very rapidly as life science companies rush to provide researchers and healthcare organizations with one of the essential tools for precision care.

CRISPR

CRISPRs (Clustered Regularly Interspaced Short Palindromic Repeats), was first discovered by Francisco Mojica, scientist at the University of Alicante in Spain, are part of an organism's bacterial defense system,

They "consist of repeating sequences of genetic code, interrupted by 'spacer' sequences – leftovers of genetic code from past invaders. The system serves as a genetic memory that helps the cell detection and destroy invaders (called 'bacteriophage') when they return," says the Broad Institute.

This innovative capability may allow scientists to correct damaging mutations or susceptibilities to diseases in living organisms devoid of side effects from traditional pharmaceutical therapies – and even resolve genetic conditions for which no pharmaceutical option available.[7]

Pharmacogenetics

Pharmacogenetics is the study of how genetic differences affects the metabolic pathways of drugs, or how individuals respond to specific pharmaceutical interventions based on its unique genetic makeup. This science forms on the basis of many precision medicine efforts, such as selecting individualized drug combinations to attack a specific type of malignancy.

Biomarkers

Biomarkers are widely used in all forms of clinical practice to indicate that a disease, infection, toxicity, or other process is taking place inside an organism.

Biomarkers may include major levels in the blood, antibodies after an infection, thyroid hormone levels, or prostate specific antigen (PSA), as well as molecular signatures can indicate whether a patient is likely to respond to certain advanced therapies or not.

The exploration of biomarkers for identification, cancer treatment and similar conditions with genetic components is one of the main branches of precision medicine.

Biobanking

In many greetings, genomic research is at its most useful when conducted at extent range. In order to identify patterns in populations in a consistent, suitable and repeatable manner, researchers must have access to analyse large volumes of patient data.

Biobanking, offer the platform for collection and storing samples of DNA from groups of individuals (typically blood, saliva, and/or urine), that allows researchers to access larger pools of potential.

Large health systems such as Geisinger Health System, North well Health, and Kaiser Permanente have established their own biobanks to support their research communities, while the Department of Veterans Affairs currently oversees one of the chief biobanks in the country. [7]

daily challenges of pricision medicine

- Inadequate technologies, limited knowledge and gaps in research are major obstacles to adding precision medicine to routine clinical care.
- Developing comprehensive infrastructure and technologies.
- Provide education and training programme to health care professionals.
- Perpetuated care disparities and under representation
- Innumerable economic, regulatory, social and technical issue that need novel solution s.
- Convincing evidentiary support for precision medicine currently, more research is needed to

make the case for precision medicine adoption, which would lead to significantly improved outcomes. [8]

II. CONCLUSION

It's to be expected that healthcare difficulties and illness patterns vary by country to country and are influenced by multiple factors. Precision medicine is predicted to generate good opportunities in the health care sector as well as job opportunities.

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