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Review Article

Review Article: Application of Genomic Data in Precision Medicine

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Abstract

Introduction*: Precision medicine used in individual genetic characteristic to provide tailored medical treatments (person specific treatment), improving therapeutic efficacy and reduce undesirable side effects. One-size-fits-all approach to more precise, person-specific treatments that can produce better therapeutic outcomes with fewer side effects. development in genomic innovation have strongly enhanced the ability to investigation, treat, and cure diseases based on genetic data.* ***Objective****: With an importance on its utility in disease diagnosis, target-specific therapy, adverse effect prevention, and its potential to improve healthcare outcomes, this article aims to examine the role of genetic data in precision medicine.* ***Methodology****: A thorough summary of the most recent uses of genetic data in precision medicine is given in this review paper. It points out the application of CRISPR-Cas9 gene editing innovation, pharmacogenomics, (WGS) whole-genome sequencing and (NGS) next-generation sequencing. Drug dosing, pharmacogenomics, unusual genetic illnesses, cancer identification, and disease prophylactic are all providing in broad overview the review.* ***Results****: The detect and target treatment therapy of a number of illnesses, including tumor and uncommon genetic mutation deformity, have been entirely convert by genomic data. The discovery of genomic mutation is made possible by methods such as WGS and NGS, which improve treatment efficacy and direct targeted therapy. Pharmacogenomics help in alter drug dosages to minimize side effects and guarantee therapeutic efficacy. There is also a lot of potential in the use of genomic data for disease prevention, such as predicting malignancy risk linked via genetic testing. But problem with unethical and illegal genetic discrimination, data secrecy, and equal access to genomic technology continue to be major concern.* ***Conclusion****: By facilitating more precision, focused, and efficient treatment plans, genomic data contributes significantly to the advancement of precision medicine. For all that combining genetic data into clinical therapy has many benefits, it is imperative to address ethical, legal, and societal concerns such data secrecy and accessibility. Genomic Enovation have the capability to highly increase patient outcomes and change healthcare globally as they develop further.*

Keywords*:* ***Precision Medicine, Genomic Data, Pharmacogenomics, Gene Editing, Next-Generation Sequencing, Personalized Medicine, Disease Prevention, Side effects***

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Introduction

The rapidly growing domain of precision medicine is important, it is a transformative shift in health maintenance and it enables more specialized and actual medical interventions which are based on individual genetic characteristics. In the past, medical treatments were based on a uniform approach, where medications and therapy were developed to treat thegeneral population. But, the latest development in genomic innovation have discover the significant of the Genetic mutations in the function of different genes and their function in determining the susceptibility of an individual to develop a certain disease, as well as response to treatment. [1]

The use of genetic information data in clinical practice treatment has make it possible to transfer from the idea that there is a one-size-fits-all approach to more precise, person-specific treatments that can produce better therapeutic outcomes with less side effects. One of the main pillars of precision medicine is genomic identification, the study of an organism's entire genome, or (Set of DNA). This field aids in detect genetic alteration and individual differences in therapy response and identified target treatment and specific dose and less adverse effect.[2] The purpose of this study is to highlight the importance of genomic data in precision medicine and its potential applications in illness research, treatment plan management, medication development, and personalized healthcare planning.[3]

1.Cancer Diagnosis and Genomics:

By allow the genomic study of a patient's cancer and detect specific type of gene mutations, gene amplifications and duplication, or chromosomal rearrangements (A mutation that occurs when a chromosome breaks and the pieces are arrange) that may contribute to the development of tumor, next-generation sequencing (NGS) innovation have entirely changed the way that tumor is diagnosed. For instance: Non-small-cell lung cancer (NSCLC) with EGFR mutations: EGFR inhibitors, like Erlotinib or Gefitinib, can help patients whose cancer cells contain EGFR[4].

EGFR(epidermal growth factor receptor) is transmembrane protein involve in cell growth and differentiations after mutations that cause uncontrolled growth of cells. EGFR mutated this receptor activate without ligand bindings These drugs target and bind the mutant EGFR receptor that cause cancer cells to grow out of control. By detecting these mutation through genomic testing, targeted medicines are administered to only the right patients.EGFR is mutated its remain activate without ligand binding activate cancer promoting pathway.AKT and mTor pathway cell survival and resistance to apoptosis. JAK-STAT pathway – increase tumor growth.

RAS/RAF/MEK/ERK Pathway-Increase tumor proliferation.[5]

Rare Genetic Disorders:

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Whole-genome sequencing (WGS) and whole-exome sequencing (WES) have become essential tools for diagnosing uncommon genetic disorders. By analyzing a patient's genetic material, healthcare providers can identify mutations that might elude conventional diagnostic techniques. This methodology has resulted in enhanced early identification, tailored treatment, and genetic guidance for families dealing with hereditary diseases such as cystic fibrosis, Huntington's disease, and sickle cell anemia.[6]

2. Pharmacogenomics: Tailoring Drug Therapy Based on Genetic Profiles

Pharmacogenomics studies how an individual's genetics influence their reactions to medications. By recognizing these genetic elements, healthcare professionals can customize drug treatments for each patient, resulting in more efficient therapies and reduced side effects[7]

Adverse Drug Reactions (ADR):

Particularly genetic alteration differences can increase the probability of adverse reactions to drugs, Genetic variations between populations can affect how drugs are metabolized which can impact the drug's effectiveness and safety.[8]

Warfarin and CYP450 Enzymes: Differences in genetic polymorphisms such as CYP2C9 or CYP1A2 and CYP3A4 can affect how medications like warfarin are metabolized.[9]

which is used as an anticoagulant. Some people breakdown warfarin very slowly, increase their risk of bleeding, while others breakdown of drug is increase, which decrease its effectiveness. Genomic testing can help in identifying these genetic variation, identified to personalized dosing of warfarin to ensure that patients receive the most suitable effective and safe dose, thus minimizing the chances of both bleeding and lack of effectiveness.Particularly CYP2C9 or CYP450 Enzymes, may experience altered metabolism of frequently prescribed medications such as warfarin (an anticoagulant) or codeine (a pain reliever).[10]

Personalized Drug Dosing:

Pharmacogenomic testing can guide drug dosing, making sure that patients receive the appropriate dosage of medication according to their genetic profile. [11]

HER2(Human epidermal growth factor receptor-2)-Positive Breast Cancer and Trastuzumab: Trastuzumab (Herceptin) is administered to breast cancer patients whose tumors show positive results for HER2 gene mutation in particular patient. Testing for the HER2 mutation guarantees that only patients who are HER2-positive get this targeted therapy, enhancing its effective result while preventing

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unnecessary treatment in those who are HER2-negative and associate its side effect and minimize disease progression duration and cost effective treatment.[12]

Thiopurine Methyltransferase (TPMT) and Azathioprine: Azathioprine is immunosuppressant drugs frequently used for autoimmune disorders such as RA (Rheumatoid arthritis). TPMT enzyme plays a critical role in drug degradation. Individuals with low TPMT activity may face negative side effects such as bone marrow suppression. Genomic testing for TPMT variations helps determine appropriate azathioprine dosage for patients, improving efficacy and decreasing toxicity.[13]

Cancer Pharmacogenomics:

By examining the genetic traits of the patient and the tumour, genomic data can be used in tumour therapy to identify the most effective treatments. The gene that codes for the B-Raf protein is called BRAF (B-Rapidly Accelerated Fibrosarcoma). A serine/threonine kinase that controls cell division and proliferation through the RAS-RAF-MEK-ERK (MAPK) signalling cascade. Increase kinase activity leading to continuous MAPK pathway activation. BRAF inhibitors like vemurafenib are utilized for treating melanoma patients who have certain mutations in the BRAF gene. Conducting tests for these mutations guarantees that only patients with BRAF mutations are prescribed the drug, resulting in improved outcomes when compared to unusual chemotherapy.[14]

3. Genomic Data in Disease Prevention and Risk Stratification

Assessing disease risk, enabling prompt interventions, customised preventative strategies, and improving healthcare outcomes all depend on genomic data. Risk classification based on genetic components could change how diseases are managed.[15]

Cardiovascular Disorder

hereditary variations in the PCSK9 and LDL receptor genes on chromosome 19 can result in hypercholesterolemia, a hereditary condition characterised by abnormally elevated blood cholesterol levels. Testing for these mutations can help guide the use of PCSK9 inhibitors, like alirocumab, to help control cholesterol levels and lower the risk of cardiovascular disease.[16]

(PRS) Polygenic Risk Scores:

Using polygenic risk scores (PRS), a healthcare professional can assess a patient's genetic risk factor for developing several factor diseases such as diabetes, obesity, autoimmune disorders, and cancer. PRS are numerical numbers that indicate a person's genetic susceptibility to a trait or illness. They are based

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on information from several genetic markers linked to a particular illness. PRS can be used to determine a person's risk of contracting an illness. These results can direct target preventative strategies, lifestyle changes, and the early initiation of screening programs.[17]

Pharmacogenomic Screening for Disease Prevention:

Genomic data is used to analyse information from millions of locations in the genome in order to anticipate treatment reactions, investigate traits, and evaluate susceptibility to certain diseases. For example, genomic diagnostics can identify those who have a higher risk of breast cancer because of changes in the BRCA1 (breast cancer gene 1) and BRCA2 (breast cancer gene 2) genes. This means that by using genetic mutation diagnose to detect people who are at increased risk for breast tumor, early screening method to detect such as mammograms can be used. Additionally, if a genetic predisposition mutation is confirmed, but it means they have higher risk compared to someone without the genetic background it may be possible to take preventive measures like taking drug tamoxifen, a medication used for chemoprevention, or having prophylactic surgery, which involves surgically removing breast tissue before cancer develops. [18]

4. Advancements in Genomic Technologies for Precision Medicine

Incorporating a patient's genetic information like DNA sequencing data, directly into electronic healthcare record.Which have improved the accessibility andaverage cost of genomic testing[12].

(NGS)Next-Generation Sequencing

The development of(NGS) next-generation sequencing technologies has enabled the facilitate high capacity and cost effective sequencing of an individual's entire genome or specific regions, prominent to the discovery of rare version of gene and extensive genomic research, which has increase diagnostic accuracy and facilitated the development of new targeted therapy.[7]

CRISPR-Cas9 and Gene Editing:

The expansion and development ofediting gene technology CRISPR-Cas9 offers extremely promise for personalized medicine by permit genetic mutations to be corrected directly in the DNA, probably leading to the development of permanent cures for genetic diseases like spinal muscular dystrophy(SMA), and cystic fibrosis sickle cell anemia.[8]

Liquid Biopsy:

Liquid biopsy refers to examining RNA orDNA, biomarkerobtained from a body fluid such a blood, urine, Cerebrospinal fluid (CSF) enabling non-penetrating and non- surgical testing for cancer mutations. This technique has positive the way for new opportunities in early cancer detection,

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assessment therapy response, and monitoring minimal residual disease in cancers such as lung cancer, breastcancer, and colon cancer.[1]

5. Ethical, Legal, and Social Considerations in Genomic Medicine

Many legal, ethical, and societal concerns must be addressed as genetic data becomes safer for hacker and misuse and changing gene rise question about ethics and safety prevalent in therapeutic decision-making.[5]

Data information Security and Privacy:

Data security and patient privacy are concerns raised by the store and collect of genetic data.It can also expose family history and sensitivity to certain diseases. Safety this data involves implementing strong coding method methods translate this code only expert in healthcare professions only, secure data storage, and clear data access policies. This guarantees that only authorized individuals and authorized organizations will use the information. Assurance to legal structure such as the Health Insurance Portability and Accountability Act (HIPAA) in the United States and the General Data Protection Regulation (GDPR) in Europe is required compulsory to secure personal health information and genomic data. Continue trust in genomic healthcare depends on securing the appropriate authorization and safeguarding the privacy of genetic data.[19]

Discrimination:

People may be subjected to differentiation by employers or insurance companies because of their genetic susceptibilities to certain illnesses. (GINA) Genetic Information Nondiscrimination Act and other laws in the United State are intended to protect people from this kind of discrimination.[4]

Equity and Access:

The cost of genetic diagnosis and treatment might be high. This makes accessing these treatments less possible for persons in poor locations or without enough health insurance. The necessary tests and treatments notbe available to residents of rural locations or communities with limited healthcare resources. Equal access to these crucial technologies is a concern. A crucial element of precision medicine's future is ensuring that these advancements are available to lower class populations.[10]

Conclusion

The utilization of genetic data in designing targeted therapies for genetic subpopulation medicine and has completely transformed healthcare by offering more individualized medicines that improve

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therapeutic efficacy, efficiency, and treatment choices. By improving disease illness and alternative choices, predicting disease susceptibility, and optimizing drug dosages, genomic information influences the evolution of medical practices.

Despite the substantial advantages, the ethical, legal, and societal ramifications of genetic data in the healthcare sector must be considered. As technology develops and more genetic data becomes available, precision medicine is positioned to offer highly customized treatments that potentially enhance patient outcomes and quality of life worldwide.[20]

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