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Genomic Data Is Used In Person-Centered Medicine to Enhance Diagnosis By Focusing On The Immune System

Hossein Amin-Anaraki[1*], Hamidreza Ashrafi[2]

[1] Biotechnology Research Center, Islamic Azad University, East-Tehran Branch, Tehran, Iran.
[2] Department of Biology, Faculty of Basic Science, Islamic Azad University, East-Tehran Branch, Tehran, Iran.

Corresponding Author's E-mail: Hosseinanaraki2012@gmail.com

Abstract:

Genomic data is crucial for precise treatment, known as customized medicine. Its use in treating autoimmune diseases will lead to a significant advancement in medicine. Autoimmune illnesses occur when the body's immune response identifies and reacts to self-antigens. Various methods may incorporate precision medical data into the clinical care of autoimmune disorders, such as identification, prognosis, classification, and prediction of therapy reactions. Various indicators are available to assist in clinical decision-making, with other indicators currently being discovered and suggested. This article focuses on information and databases in personalized medicine for autoimmune medical conditions and exchanging information. The discussion focused on personalized medicine for specific autoimmune disorders, examining different indicators for identification, prognosis, classification, and treatment response tracking.

Keywords: Genomic data, Immunological systems, Personalized Healthcare

Introduction

The immunological system's role is to defend the body against illness by eliminating infectious organisms that assault the body or are transmitted via vaccination (1). The body's defense system operates via tightly controlled biological mechanisms that help identify and distinguish between bodily and foreign cells (2). The body's immune system cells often cohabit alongside other cells that possess a self-marker protein (3). Immune responses are initiated when a substance, such as a bacterium, part of an organism, or a chemical, is detected on the outermost layer of a cell and recognized by the body's defensive mechanisms (4).

The human defense mechanism consists of two divisions: innate and developed immunity. Innate immunity acts as the first protection against infectious diseases upon recognition by the body, whereas acquired immunity eliminates infections in the later stages of illness. When the body's immune system is activated, it identifies and eliminates alien entities (5). However, under some aberrant circumstances, the immune system may exhibit insensitivity to antigens, hypersensitivity to substances or mistake cells with self-markers as alien cells (6).

Various medical problems may impact the immune

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system's function, resulting in various symptoms collectively known as immunological disorders. Immune-mediated illnesses contain congenital and acquired immune deficiency immune-proliferative diseases such as cancers (multiple myeloma, lymphoma, leukemia), autoimmune conditions like rheumatoid arthritis, and immunological hypersensitivities like allergies (7, 8). Primary immune deficiency, frequently referred to as inherited immune deficiency, encompasses many immunological illnesses that affect the growth and operation of the immune system and its functions (9). Primary immunodeficiency refers to disorders caused by monogenic germline mutations that lead to loss of function, gain of function, or loss of expression. Externally and environmental variables may negatively impact the immune system, leading to acquired immunodeficiency. This is a regular occurrence in clinical settings and can result from several illnesses (10).

Personalized healthcare refers to the shift in medical procedures, particularly in medical care and diagnosis, from a generalized approach to a more individualized and genetic-based classification of patients to gather more specific information regarding the illness and the individual receiving treatment (1, 10, 11). The intricate nature of the immune system in the body and the cells' capacity to transition between several activation stages in both standard and abnormal settings contribute to the variety of treatment strategies (12). Immune disorders may be different, leading to variances in reaction to treatment. The variation in the illness progression underscores the need for identifying individualized markers for diagnosing immunological diseases. Genetic evaluation is crucial for selecting the most effective therapy method among several possibilities with varied mechanisms, dangers, and effectiveness (13, 14).

This article covers medical precision kinds of data, genomic information in precision medicine, genome-wide and customized medicine database servers, and data sharing, access, and usage. This article also discusses the application of genomic techniques and information in disease comprehension, evaluation employing specific indicators, forecasting monitoring employing prognosis indicators, personalized therapy for immune-related conditions, and monitoring response to therapy employing reaction biomarkers.

Personalized medicine for distinct autoimmune conditions

What's referred to as the immune system?

Immunization is the body's capacity to avoid illness through fighting against the growth of dangerous microorganisms known as pathogenic organisms (15). Immunity may be roughly divided into two types:

1. Innate or Natural Immunity 2. Acquired Protection

Innate or natural immunity

Humans encounter several possible infections every day via touch, ingestion, and inhalation. The immune system's adaptive function plays a crucial role in our capacity to prevent infection by recognizing and eliminating specific pathogens upon subsequent contact (16). Upon first exposure to a novel pathogen, adaptive immune reactions are delayed due to the activation and expansion of particular clones of B and T cells, resulting in a successful response taking about a week to develop (17). On the other hand, a single microorganism with a doubling period of one hour may generate about 20 million offspring within one day, causing a complete infection (18). Thus, in the first crucial hours and days of encountering a novel pathogen, our natural immune system is essential for shielding us against infection (19).

Unlike adaptive immune system reactions, innate immune system reactions lack specificity towards a particular disease (20). They rely on a set of molecules and phagocytic cells that identify common characteristics of infections and rapidly become active to assist in eliminating intruders. The adaptive immune system, or immune system, emerged in evolution fewer than 500 million years ago and is exclusive to vertebrates (21). In contrast, innate immune reactions are present in both invertebrates and vertebrates, as well as in plants, with conserved regulatory mechanisms (20, 21). Natural immunity, controlled by phagocytes, is the first line of defense versus disease-causing substances (22). The human body's innate defense detects intruding germs via germline-encoded pattern-recognition receptors (PRRs). Pattern Recognition Receptors (PRRs), such as Toll-like and cytoplasmic receptors, identify specific microbial components of invading pathogens and stimulate immune cells (20-22).

The way in which innate or natural immunity functions

Upon detecting non-self-agents, Pattern Recognition Receptors (PRRs) located on the cell's outer membrane, intracellular components, or in body fluids carry out opsonization, stimulate complement and coagulation processes, facilitate phagocytosis, initiate pro-inflammatory signaling processes, trigger apoptosis (23). The intracellular signaling pathways produce overlapping and distinct genes that play a role in inflammatory immune reactions and are crucial in personalized medicine. Innate immune reactions include phagocytes (neutrophils, monocytes, and macrophages), inflammatory mediator-releasing cells (basophils, mast cells, and eosinophils), and natural killer (NK) cells (24).

Acquired immunity

Adaptive immunity is not established at birth. It has been acquired (25). The learning process begins when an individual's immune system comes into contact with external intruders and identifies non-self-substances known as antigens (26). The different elements of acquired immunity develop to effectively target each antigen and create a memory specific to that antigen. Acquired immune systems, also known as particular immunity, target a specific antigen met earlier. Its distinguishing features are its capacity to acquire knowledge, adjust, and retain information (25, 26).

Adaptive immunity requires a period to establish after initial exposure to a novel antigen. Following the first exposure, the immune system retains memory of the antigen, resulting in faster and more efficient responses upon repeated encounters (27).

The white blood cells contributing to acquired immunity include Lymphocytes consisting of T cells and B cells. Other components of acquired immunity include Dendritic cells and cytokines (28). The immune system's complement system boosts the efficacy of antibodies. Acquired immunity is the body's defense system that develops after exposure to a pathogen, either through previous infection or through the transfer of chemicals that protect from mother to child. Adaptive immunity is mediated by T and B lymphocytes that are clonally dispersed and exhibit specificity and memory. Stimulation of the innate body's immune system often stimulates acquired immunity. Helper T cell subset formation and cytokine production impact adaptive immunity (25-28).

Mechanism of Acquired Immunity

Naïve T-helper cells develop into two subsets that are TH1 and TH2 when triggered by Antigen-presenting Cells (APCs). Interferon-γ (IFN-γ) is secreted by TH1 cells and enhances explicitly cellular immunity. TH2 cells generate interleukin 4, 5, 10, and 13 (IL-4, IL-5, IL-10, and IL-13). IL-12 drives TH1 differentiation, whereas IL-4 promotes TH2 differentiation. TH2 has a significant role in enhancing humoral immunity (27-29).

Development of immunological disorders

Immunological illness arises from the dysfunction of many components of the mammalian immune system's function. The immune system reaction identifies and removes antigens while also tolerating its tissues (30). The classification of immune-mediated illnesses depends on the significant immune pathology lesion. Immune-mediated illnesses may be categorized into acute hypersensitivity, autoimmune diseases, immune-complex illness, and delayed-type hypersensitivity. Autoimmunity may be categorized into adaptive immunity-mediated and innate immunity-mediated. Most illnesses are characterized by positive pathogenic feedback among adaptive and innate immune systems. Figure 1 below illustrates the development of immunological disorders (31).

Personalized Healthcare

Personalized healthcare involves customizing diagnostic methods, therapy, and preventative measures based on specific patient features to achieve the best possible result for every individual, focusing on availability and cost-effectiveness (32). Personalized healthcare utilizes an individual's unique genetic composition to inform clinical treatment decisions (33). Researchers constantly seek prognostic, diagnostic, and predictive biomarkers to assist in clinical decision-making and guarantee that the most appropriate medicine is provided to the correct patient at the optimal moment (34). Figure 2

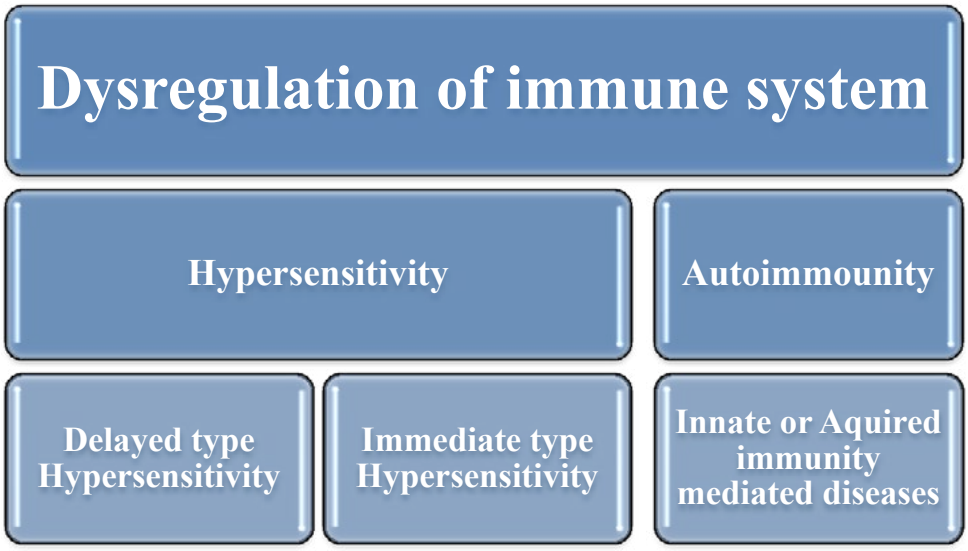

Fig 1. Diagram illustrating the development of immunological disorders.

illustrates the subdivision of customized medicine. The method of application of precision medicine is given in Figure 3.

Individualized medicine and genetic information

Personalized healthcare often consists of a large amount of genetic data. Advancements in power systems have enhanced the application of massive amounts of information in customized or precise healthcare. The advancement of genomics data provides several opportunities for developing clinical procedures, diagnostics, and preventative strategies and predicting the most effective therapies for various illnesses associated with distinct locations and lineages (35).

Types of data used in precision healthcare

Information about patients is being systematically collected and becoming more complicated, especially in neuroimaging, where over 10 petabytes of data are generated annually. Precision healthcare research utilizes various types of data including imaging information (CT, PET, ultrasound, and MRI), bio-sample information (serum, plasma, and urine amount), molecular information, genomics information (nucleotide sequences), proteomic identifying information (mass spectrometry), digital pathology information, biomedical instrument information (blood pressure, heart rate, and insulin level), and clinical information (death/survival data, demographics, and medical-based questionnaire) among others (35, 36).

Advancements in personalized medicine have resulted in the development of tailored brain models for patients with intractable epilepsy and breakthroughs in understanding the epigenetic mechanisms of hematopoiesis. A comprehensive grasp of several informatics domains, such as data science, data management, data curation, and bioinformatics, is essential for combining and

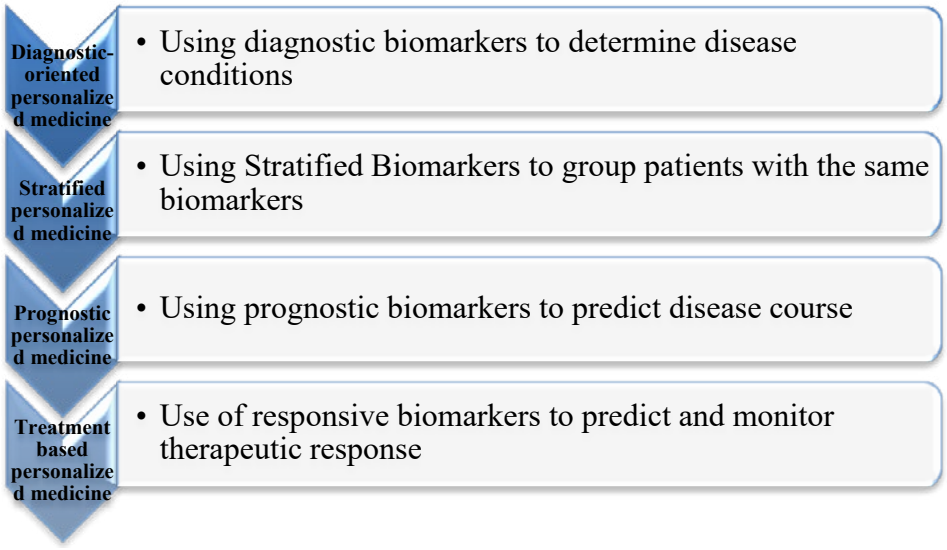

Fig 2. A diagram illustrating the many divisions of precision healthcare.

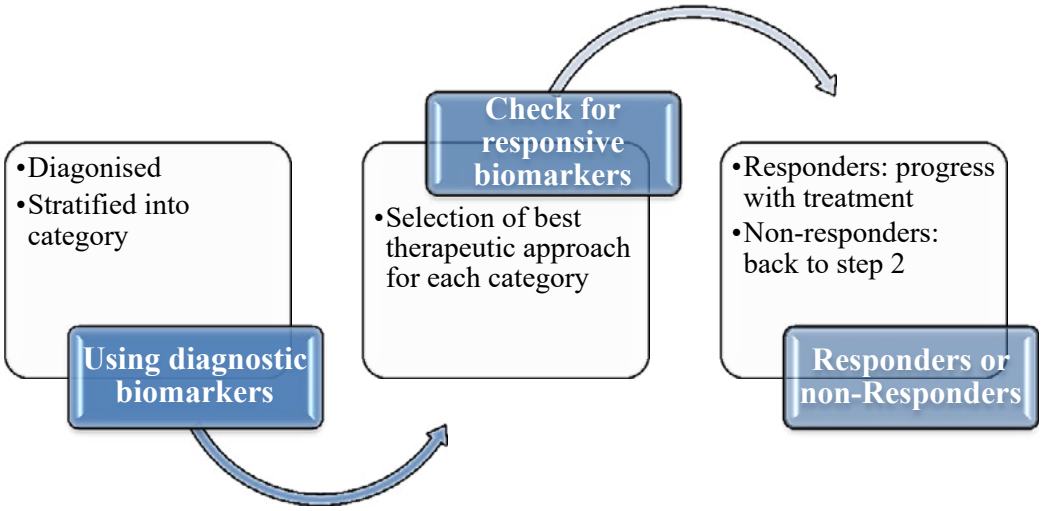

Fig3. The implementation of the precision healthcare approach is shown in Figure 3.

integrating numerous data kinds (37).

Platform for genomic and customized healthcare

A database of information is an organized collection of information managed by the database management system (DBMS) on a computing device. A relational database management system consisting of data, DBMS, and related applications is often called a database. Each database includes certain forms of data. Here, we shall introduce databases related to customized or precision healthcare (38).

Immune Epitope Database (IEDB)

The IEDB is an open-access database valuable for vaccine and pharmaceutical research. It contains experimental information on immunoglobulins Major Histocompatibility Complex (MHC), attaching information gathered from various antigenic resources, as well as Helper T Lymphocyte (HTL) and Cytotoxic T Lymphocyte (CTL) epitopes for human and other animal species. The collected information also assists in forecasting and analyzing different types of epitopes (39). The database is accessible at https://www.iedb.org/.

Prostate cancer-related lifestyle database (PCaLiStDB)

Lifestyle therapy focuses on the relationship between lifestyle choices and chronic or immune-related illnesses. PCaLiStDB is a lifestyle collection focused on accuracy in preventing prostate cancer and other lifestyle-related disorders. The database contains lifestyle-related genes, biomarkers linked with lifestyle types, and individualized predictors of lifestyle-related diseases (40). The database URL is http://www.sysbio.org.cn/pcalistdb/.

Clinical Genome Resources (ClinGen)

The ClinGen database structure, supported by the National Institute of Health (NIH), collects clinically significant data for personalized research and medicine. This data collection extracts clinically significant genes and variations for accurate diagnosis and therapy (41, 42). The database may be accessible via the website https://clinicalgenome.org/.

Personal Genome Project (PGP)

The personal genomics research collection is considered a significant advancement in healthcare technology. This information collection is accessible to anyone focused on creating a tool for individualized treatment and furthering research. The database contains diverse data sets for several locations, such as PGP-UK, PGP-AUSTRIA, PGP-CHINA, PGP-CANADA, and PGP-UNITED STATES. The information system provides Genome, Methylome, transcriptome, and phenotypic data for the application of precision medicine (43). The genome database may be accessed at https://www.personalgenomes.org/.

Online mendelian inheritance in man (OMIM)

The information system was established at the beginning of the 1960s, with an online version developed in 1985. OMIM is an open-access library for specialists specializing in genetic diseases, genetics researchers, and advanced medical students. Information about human genes, genetic diseases, clinical characteristics, phenotypes, and genes (44). The URL for this database is https://www.omim.org/.

Human gene mutation database (HGMD)

This collection of files compiles known gene mutations responsible for human hereditary disorders. Data collection contains precise healthcare information, including gene symbols, genomics coordinates, splicing, various diseases, phenotypes, and alterations in the human chromosome (45). The database may be accessed at http://www.hgmd.cf.ac.uk/ac/index.php.

Clinical Genome Database (CGD)

The Clinical Genomic Library is a crucial resource at the intersection of clinical and genomic healthcare, providing medically relevant genetic information and possible therapies. The CGD provides information regarding allelic circumstances, gene symbols, clinical categories (manifestation and therapies), affected populations, mode of inheritance, and pathogenic mutations for all captured illnesses in the database (46). The database is accessible at the following link: https://research.nhgri.nih.gov/CGD/.

Other database related to precision/personalized medicine

Other collection initiatives are also under progress to enhance the current ones, such as The Human Variome Project (47). Additionally, several websites and information related to precision healthcare are beyond the scope of this article to cover in detail. Table 1 below contains more database information on precision healthcare in general and their corresponding connections (40-47).

Application of Genomic and Personalized Healthcare Information

The exchange of information involves the transmission of a single data source across several applications or users, allowing for the exchange, access, and reuse of copies of information. Data may be categorized as open access (publicly accessible) or controlled (limited). Distributing data includes both original information (such as nucleotide sequences) and additional information (previously used or processed information) (63). Access to precision healthcare data, such as clinical information, may be open or limited. Authorization from an authorized individual is required to utilize the data for therapeutic, diagnostic, and research purposes (64).

Table 1. The database is associated with precision healthcare in general and its connections**.**

Database	Link	Ref
NetPath (signal transduction)	http://www.netpath.org/	48
Entrez – (encompasses sub-Databases)	http://www.ncbi.nlm.nih.gov/sites/gquery	49
GeneCards	http://www.genecards.org/	50
Human Genome Resources	http://www.ncbi.nlm.nih.gov/projects/genome/guide/human/	51
Ensembl Human Genome Browser	http://www.ensembl.org/IIomo_sapiens/Info/	52
Online Mendelian Inheritance in Man (OMIM)	http://www.ncbi.nlm.nih.gov/omim/	53
Gene Expression Omnibus	http://www.ncbi.nlm.nih.gov/geo/	54
ENCODE Project: ENCyclopedia of DNA Elements, NHGRI	http://www.genome.gov/ENCODE/	55
PubChem	http://pubchem.ncbi.nlm.nih.gov/	56
PhenX Toolkit	https://www.plienxtoolkit.org/	57
Human Genome Project, NHGRI	http://www.genome.gov/10001772	58
NCBI BioSystems	http://www.ncbi.nlm.nih.gov/biosystems/	59
National Human Genome Research Institute (NHGRI)	http://genome.gov	60
ExPASy Proteomics Server	http://expasy.org/	61
HUPO: Human Proteome Organization	http://www.hupo.org/	62

Conclusions

Genomics information is crucial for precision healthcare since it helps explain individual variability and development. However, the practical use of chromosomal information in clinical settings must be enhanced to address issues identified by researchers, such as the disparity between the molecular and medical data forms poses a challenge due to the vast amount of genomic information, making it difficult to handle clinical data in practice without further manipulation. Genomic and observational information utilized in clinical contexts varies due to the vast amount of data in genomic operations, making it distinct from data in clinical systems. Challenges arise when aligning genomic and clinical information for medical interpretation, particularly in specific sequencing, where information is often processed before medical analysis. There needs to be more global validation for the biomarkers being used, highlighting the need for international cooperation to evaluate the existing biomarkers. Conquering these obstacles will provide further possibilities for using genetic data in therapeutic settings.

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Authors' Contribution

Hossein Amin-Anaraki and Hamidreza Ashrafi were involved in the conceptualization, design, and support of the study. All authors read and confirmed the final manuscript.

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All data are obtainable after an appeal from the corresponding author.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

Competing interests

The authors declare they have no conflicts of interest regarding the publication of this article.

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