Research Output Journal of Public Health and Medicine 3(2)5-9, 2024

ROJPHM

ISSN ONLINE: 1115-9715
ISSN PRINT: 1115-6147

https://rojournals.org/roj-public-health-and-medicine/

https://doi.org/10.59298/ROJPHM/2024/325902

The Role of Precision Medicine in Autoimmune Diseases

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ABSTRACT

Autoimmune disorders are a broad category of complex, chronic ailments caused by the immune system wrongly targeting the body's tissues. The variety of over 80 diverse autoimmune illnesses makes effective therapy difficult. Precision medicine, a personalised approach based on individual genetic, environmental, and lifestyle factors, has emerged as a viable treatment method for autoimmune illnesses. Genomic technology and biomarkers are critical in identifying and tailoring therapy options for patients. This study investigates the concept of precision medicine, its applicability in autoimmune illnesses, and the obstacles that come with its implementation. Real-world case studies demonstrate its ability to transform treatment outcomes. The future of autoimmune disease management through precision medicine is dependent on addressing ethical problems, progressing genomic research, and increasing access to these cutting-edge medications.

Keywords: Precision medicine, autoimmune diseases, genomics, biomarkers, personalized treatment

INTRODUCTION

Autoimmune diseases represent a diverse collection of more than 80 different conditions that primarily affect women and are among the top ten leading causes of death for women. These diseases occur when the immune system mistakenly targets and attacks the tissues of the body, generating an immune response. The onset of autoimmune diseases is associated with different factors, including but not limited to genetic predisposition, environmental, and infectious triggers. Symptoms associated with autoimmune conditions vary considerably depending on the autoimmune disease and the tissues primarily affected by the immune response; as an example, multiple sclerosis primarily attacks the central nervous system, while rheumatoid arthritis primarily affects joints. Some conditions are systemic and involve widespread tissues, as is the case with systemic lupus erythematosus [1, 2]. The primary role of the immune system is to recognize, attack, and eliminate foreign pathogens such as viruses and bacteria to prevent infection and disease. While the immune system is designed to be beneficial, it can also lead to autoimmune disease and other inflammatory conditions through inappropriate immune responses against 'self' tissues in a genetically and environmentally predisposed host. Although much is known about the immune responses mounted during autoimmunity, the complex etiology of these disorders remains unclear. The heterogeneity, diversity, and multifactorial nature of immune-mediated diseases make diagnosing and treating autoimmune diseases extremely challenging, especially since patients are typically older and likely present comorbidities that need to be carefully managed. Management of these diseases includes lifelong therapies with immunosuppressive drugs to manage symptoms and prevent tissue damage. Given the stark heterogeneity of these diseases, many customized treatments based on the patient's clinical course are often essential. It is therefore critical to develop tools to diagnose and treat autoimmune disease patients at the earliest possible stage for a better and more effective outcome [3, 4].

DEFINITION AND TYPES OF AUTOIMMUNE DISEASES

Autoimmune diseases are characterized by self-imperative and antibody-driven immunities that disrupt the body's regular functions. They can be classified as organ-specific or systemic. Organ-specific diseases, such as Type 1 Diabetes Mellitus and Hashimoto's Thyroiditis, target specific organs, resulting in distinct symptoms. Systemic diseases, like Systemic Lupus Erythematosus and Rheumatoid Arthritis, affect multiple organs and present a diagnostic challenge. Diagnosis relies on comprehensive evaluation and paraclinical investigations. These diseases are prevalent and more common in females [4, 5].

PRECISION MEDICINE: CONCEPT AND PRINCIPLES

Precision medicine is a transformative approach for improving complex disease prediction, prevention, and treatment. It focuses on the individual, determining the best therapeutic strategy and lifestyle choices. This approach considers genetic information, environmental conditions, and behavior. Precision medicine collects personalized data using advanced technology and interdisciplinary collaborations. It enables the design of personalized protocols for autoimmune diseases, which are ideal candidates for this approach due to their complexity [6, 7].

GENOMICS AND PERSONALIZED MEDICINE

Personalized medicine has been placed within the precision medicine paradigm, which aims to use an individual's genetics, environment, and lifestyle to provide disease prevention and treatment strategies. Genomic technologies, particularly next-generation sequencing, have allowed the identification of genetic variations such as single nucleotide polymorphisms, insertions, deletions, duplications, and rearrangements related to the onset and progression of autoimmune diseases. There is also evidence that environmental factors may contribute to genetic regulation related to autoimmune diseases [8]. Biomarkers are essential for the implementation of personalized medicine, particularly for precision approaches in disease therapeutics. In general, a biomarker is a molecule found in blood, other body fluids, or tissues that can indicate the presence or progression of a disease. In the case of autoimmune diseases, blood biomarkers are appealing as they can reflect gene expression in immune cells and have specific cell functions in the immune response and inflammation. There are specific molecular connections between genotype and phenotype related to autoimmune diseases. The transcriptional signatures obtained from gene expression arrays revealed intense genetic imprinting and distinct phenotypic subclasses in the suboptimal response of patients with autoimmune diseases. This sensitivity of the immune response also provides mechanistic biomarkers with potential applications for monitoring early events of inflammation or immunosuppression in clinical trials or adding efficacy data to clinical and diagnostic research [9]. The two hallmarks of a gene signature in autoimmune diseases are related to the amount of upregulated or downregulated genes and the immune relatedness of the genes. For instance, the responsiveness of a molecular gene signature cannot be reached without a sufficient number of genes regulated at the mRNA level. Ideally, these genes should be among the most regulated genes in the tissue of interest. The consortium has identified many differentially expressed genes that were related to more than one-third of each person's transcriptome. The genes shared common downstream network connectivity through many key pathways such as inflammation, neutrophils, leukocyte extravasation, primary immunodeficiency, cytokine signaling, and Type I diabetes [10]. Genetic mutations can modify the disease severity before the start of any medication when the patients are analyzed in the early stages of autoimmune diseases. These mutations could be beneficial for early treatment planning. Often, people with autoimmune diseases may seek the cause of their condition for years before they are diagnosed. A basic gene test may increase the probability of accelerating to a correct diagnosis and ensure appropriate treatment. There have been rapid advances in technology and knowledge on gene mutations in the last 10 years. Perceptive evolution in the study of gene mutations causing specific disorders is focused on providing individualized treatment for patients using different gene tests, as well as accessing a significant change. Genetic testing can provide essential information to help in the early detection and care of these conditions, based on the increasing evidence of the genetic background of autoimmune diseases [11].

APPLICATION OF PRECISION MEDICINE IN AUTOIMMUNE DISEASES

Precision medicine is increasingly used to manage autoimmune diseases, integrating genetic and genomic data into personalized treatment plans. This reduces decision-making time and adjusts treatment after failed therapies. Precision medicine in autoimmune or rheumatic disorders tailors treatment based on the patient's risk, progression, and symptoms. It offers various options, empowering patients to actively participate in therapeutic decisions and adhere to the intervention plan. Real-world applications show the efficacy of targeted approaches in treating severe autoimmune diseases. Personalized medicine options guide current and future treatment regimens. For example, the molecular basis of myasthenia gravis informs evolving treatments, avoiding generic and non-molecular therapies [12, 13].

CASE STUDIES AND SUCCESS STORIES

It is often illustrative to provide relevant case studies that clarify precise patient details, especially when discussing precision medicine. Unfortunately, there are relatively few relevant case studies in this context on which we can draw. However, there are case reports where personalized treatments for patients have been applied with some success. One descriptive study randomized patients to biologic treatments based on genomic testing or biologics based on the likelihood of response to treatment and found that 58.9% of patients randomized to genomically driven therapy were in remission within 48 weeks, compared with 42.4% of patients randomized to the other treatment strategy, which was not based on genomics [14, 15]. The study above is particularly interesting, as it comprises a relatively straightforward real-world application of the principles involved in precision medicine, as indicated by a patient whose story we have already discussed. As it is currently one of the few studies offering an example of precision treatment for an autoimmune condition, it is worth discussing in more detail. The patient not only has active rheumatoid arthritis, but also long-term cholesterol levels that are above optimal targets, and the normal treatment agents such as steroids have caused significant side effects. In 2019, the patient was admitted to undergo treatment for high cholesterol levels. Although he signed off to only having a specific procedure, jointly between the medical and rheumatology teams, the decision was made to infuse healthy umbilical cord delivered mesenchymal stem cells to help his rheumatoid arthritis in an attempt to put the disease into remission. It is now almost 20 months following his single treatment of umbilical cord and therapy that the patient experienced benefits [4, 16].

CHALLENGES AND FUTURE DIRECTIONS

The lack of access to genotypic and phenotypic information hinders precision medicine implementation, resulting in patients not receiving comprehensive diagnoses. Translating molecular data into disease mechanisms remains unclear, and clinical trials offer conflicting evidence on their practicability. Barriers to precision medicine must be addressed to effectively translate its therapeutic potential. Autoimmune diseases are complex and currently defined by patients' symptoms, not genetic similarities. Advanced therapies are limited by costs and potential toxicity. Inequities in healthcare delivery and access to therapies persist. Training could help alleviate these issues. Collaborative research and harmonization of findings are needed for increased patient impact. Specific guidance is necessary for combining different compounds. Genomic medicine faces challenges, and registries should include critical information. Academic institutions should be involved in long-term intelligence and early-phase clinical trials. Regulatory procedures are being reviewed, and progress is needed. Rare immune-mediated diseases associated with systemic autoimmunity present opportunities for improved therapeutic management. The future of precision medicine relies on delivering on these expectations [17, 18].

ETHICAL AND REGULATORY CONSIDERATIONS

The implementation of precision medicine in the clinical management of patients may face several ethical limitations that differ from those of broader applications. It is generally accepted that shared genetic, epigenetic, and environmental factors cause a high interplay between autoimmune diseases and cancer. [19, 20]. Personalized medicine raises ethical questions about data ownership and the impact of genetic and health information on the patient's social life. Negative results and issues related to employer insurance, social stigma, and anxiety must be addressed. Patient involvement in treatment selection based on multiomic research requires robust informed consent. Clinical trials face new challenges, such as drug-vaccination interactions and the use of electronic health records. Collaboration among stakeholders is necessary to address ethical considerations and establish clear guidelines. The ethical and legal principles guiding research and practical application of personalized medicine need further refinement. Precision medicine has great potential but requires the joint commitment of all stakeholders to address ethical concerns [21, 22].

CONCLUSION

By combining genomes, biomarkers, and personalised treatment options, precision medicine provides a game-changing approach to the diagnosis and treatment of autoimmune illnesses. Precision medicine, which tailors medications based on individual genetic and environmental profiles, has the potential to enhance patient outcomes, reduce therapy failure, and empower individuals to manage their ailments. Despite the potential, problems like as cost, accessibility, and ethical concerns must be solved before precision medicine can be effectively integrated into clinical practice. Continued study, collaboration, and technical breakthroughs will be required to overcome these obstacles and realise the full potential of precision medicine for autoimmune illnesses.

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CITATION: Mugo Moses H. (2024). The Role of Precision Medicine in Autoimmune Diseases. Research Output Journal of Public Health and Medicine 3(2):5-9. https://doi.org/10.59298/ROJPHM/2024/325902