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Vexas Syndrome Decoding: Somatic UBA1 Mutations, AI-Driven 3D Genomic Fingerprinting, New Frontier in Diagnosing Global and In India, Therapeutic Pathways, Curative Stem Cell Transplantation

Yash Srivastav[1*], Shivani Singh[1], Vaishali Bhagwani[2], Amita Singh[1],Brijesh Kumar Pal[1], Kamini Prajapati[1], Stuti Verma[2]

[1]D.K.R.R Pharmacy College, Amberpur, Sitapur (Uttar Pradesh), India. 261303
[2]K.P. Singh Memorial Institute of Pharmacy, Sitapur, U.P, India
[3]Aryakul College of Pharmacy and Research, Sitapur, Uttar Pradesh, India. 261303

***Corresponding Email: yashsrv.108@gmail.com**

Abstract:

VEXAS syndrome (Vacuoles, E1 enzyme, X-linked, Autoinflammatory, Somatic syndrome) is a more recently described, acquired somatic mutation in the UBA1 gene, and is an autoinflammatory disorder of adults. Its clinical features include systemic inflammation, clonal hematopoiesis, abnormalities of the bone marrow and various hematological, rheumatological and dermatologic changes. This review discusses the molecular pathogenesis of VEXAS syndrome with emphasis on the UBA1 mutations, immune dysfunction and disease pathogenesis. In addition, it emphasizes the importance of artificial intelligence, next generation sequencing (NGS), integration of multi-omics and three-dimensional (3D) genomic fingerprinting in the improvement of diagnosis and molecular characterization. The current therapeutic options such as the use of corticosteroids, JAK inhibitors, azacitidine and biologic agents, as well as novel stem cell-based therapies are reviewed. Special focus is on the most promising potentially curative therapy, namely, allogeneic hematopoietic stem cell transplantation (HSCT). Although genomic medicine and precision diagnostics has come a long way, there are still issues to be addressed, particularly with respect to diagnosis at an early stage and longer-term management. The synergy of genomics, artificial intelligence (AI), and regenerative medicine could have a profound impact on the care of VEXAS syndrome patients in the future.

Keywords: VEXAS Syndrome; UBA1 Mutation; Clonal Hematopoiesis; Artificial Intelligence; Genomic Medicine; 3D Genomic Fingerprinting; Precision Medicine; Next-Generation Sequencing; Stem Cell Transplantation; Autoinflammatory Disorders.

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1. INTRODUCTION

Genomic medicine has made tremendous progress, and complex inflammatory and hematological diseases are much better understood. One of the most significant findings in recent years is VEXAS syndrome which is a new adult onset auto inflammatory condition caused by somatic mutations of the UBA1 gene[1]. The syndrome has been the subject of much attention since its discovery in 2020, as it is associated with systemic inflammation, bone marrow dysfunction and clonal hematopoiesis. VEXAS syndrome has shed new light on the connection between somatic mutations and immune-mediated diseases.

At the same time, breakthroughs in both AI and next NGS and multi-omics technologies have revolutionized the rare disease diagnosis and precision medicine. The identification of early signs and molecular characterization of VEXAS syndrome is possible with the use of AI-assisted genomic analysis and emerging 3D genomic fingerprinting approaches[2]. Moreover, an increasing body of evidence indicates that allogeneic HSCT could be a therapeutic option of potential cure; thus, coupling genomics, computational medicine, and advanced therapeutics in disease management is important.

1.1 Background and Context

VEXAS syndrome was first identified by Beck et al. in 2020 after somatic mutations were found within the UBA1 gene among patients exhibiting unexplained features of inflammation and blood disorders. The UBA1 gene is known to code for an enzyme that is necessary for protein ubiquitination and cell stability. Mutation within this particular gene results in dysregulation of the body's immunity, causing inflammation, and bone marrow diseases. Some symptoms of this disease include repeated high fever, neutrophilic skin conditions, vasculitis, lung disease, macrocytic anemia, and myelodysplasia. With increased usage of genomic sequencing techniques, more cases have been reported around the world[3].

1.2 Objectives of the Review

The objectives of the present review are:

- To study the molecular mechanism and pathogenesis of VEXAS syndrome.
- To analyze the impact of AI-enabled genomics and 3D genomic fingerprinting on VEXAS syndrome diagnosis.
- To discuss global as well as the Indian perspective on VEXAS syndrome.
- To explore therapeutic interventions and the curability of VEXAS syndrome by stem cell transplantation.

1.3 Importance of the Topic

VEXAS syndrome is one of the important discoveries in genomics-based medicine as it reveals the possibility for the development of acquired mutations leading to systemic inflammation. It is difficult to diagnose VEXAS syndrome since it overlaps with other diseases such as autoimmune conditions in the field of rheumatology, dermatology, and hematology[4]. Hence, early detection of the disease is vital as late detection will contribute to worsening disease prognosis and morbidity. Another reason to emphasize the significance of the topic lies in the ability to utilize

AI-based genome technologies and precision medicine techniques. Finally, the introduction of stem cell transplantation in treating the disease gives it additional value.

2. MOLECULAR PATHOGENESIS, UBA1 MUTATIONS AND CLINICAL SPECTRUM OF VEXAS SYNDROME

Knowledge of the molecular mechanism of VEXAS syndrome is critical to comprehending the syndrome's unique presentation in terms of inflammation and hematologic involvement. It was shown, following the discovery of the syndrome in 2020, that somatic mutations in the UBA1 gene play an important role in the onset of the syndrome[5]. The mutations impair key cellular mechanisms involved in protein metabolism and the functioning of the immune system, thereby causing chronic systemic inflammation as well as hematological disorders such as clonality. Recent progress in sequencing has shed more light on the interaction between these various components. For these reasons, study into VEXAS syndrome's genetics and molecular mechanisms is highly relevant.

2.1 Genomic Basis and Somatic UBA1 Mutations

Acquired somatic mutations that target the UBA1 gene, which is found in the chromosome Xp11.23, are the cause of VEXAS syndrome. UBA1 codes for ubiquitin-like modifier activating enzyme 1, the main enzyme in charge of initiating the ubiquitin-proteasome pathway through the E1 pathway[6]. The ubiquitination system contributes to protein breakdown, intracellular communication, cell cycle regulation, and immune balance. Pathogenic mutations typically affect the methionine-41 (p.Met41) location; however, other pathogenic mutations have been reported recently.

The mutations affect the expression of the UBA1b isoform in the cytoplasm. Consequently, ubiquitination is affected and there is a problem with the activation of the inflammatory pathways. The effects that the mutation causes at the molecular level provide the basis for disease pathogenesis and cause the clinical manifestations of the disease.

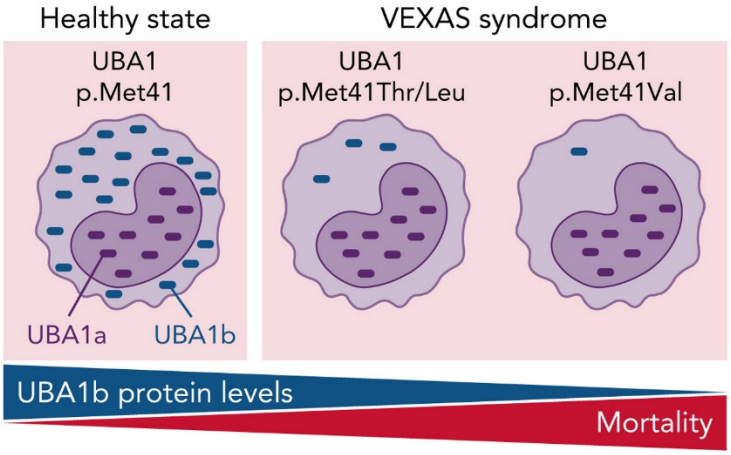

Figure 1: Effect of UBA1 p.Met41 Variants on UBA1b Expression and Disease Severity in VEXAS Syndrome

Whereas genetically inherited conditions result from germline mutations present since conception, the VEXAS syndrome is caused by somatic mutations that occur in hematopoietic

stem and progenitor cells later on in life[7]. These abnormal cells proliferate and give rise to clonal hematopoiesis, which is highly correlated with both inflammatory symptoms and hematological abnormalities. Recent genomic research has shown the presence of various UBA1 mutations in relation to different clinical presentations, pointing to the possibility that the nature and amount of mutations can determine the severity of VEXAS syndrome.

2.2 Molecular Pathogenesis and Disease Development

The causative factor behind VEXAS Syndrome is mainly the ubiquitination dysfunction due to faulty UBA1 function. Ubiquitination is an essential cellular function that plays a key role in protein degradation, signal transduction, and immune system homeostasis. Gene mutations associated with the UBA1 gene can result in the disruption of these processes, causing improper functioning of the cells and ultimately producing abnormal proteins[8]. This leads to chronic inflammation due to disrupted immune signaling.

Studies have shown that mutant myeloid cells produce elevated levels of pro-inflammatory cytokines, including interleukin-6 (IL-6), tumor necrosis factor-alpha (TNF-α), and interferon-gamma (IFN-γ)[9]. These inflammatory mediators promote tissue damage, vascular inflammation, and immune dysregulation across multiple organ systems. The resulting cytokine-driven inflammatory environment is considered a major factor underlying the diverse clinical manifestations observed in affected patients.

Vacuolation in myeloid and erythroid precursors in the bone marrow is another typical hallmark of VEXAS syndrome. Further accumulation of clones of mutant hematopoietic cells contributes to an increased inflammatory response and the risk of development of hematologic disorders, especially MDS. The combination of clonal hematopoiesis, cytokine dysregulation, and immune dysfunction forms a multifactorial pathogenetic background for the disease.

2.3 Clinical Manifestations and Global–Indian Disease Landscape

VEXAS syndrome has been noted to present itself in a clinically heterogeneous way affecting multiple body systems. VEXAS is a disease characterized by both inflammation and hematological abnormalities which make it difficult to diagnose since these symptoms may mimic other rheumatologic, dermatologic, and hematologic conditions. Patients experience different signs depending on various factors like mutation type, clonal load, and disease severity[10].

Clinical symptoms associated with VEXAS Syndrome include:

- **Hematological features:** Macrocytic anemia, thrombocytopenia, leukopenia, vacuoles in the bone marrow, and myelodysplastic syndrome.
- **Rheumatological manifestations:** Relapsing polychondritis, vasculitis, inflammatory arthritis, and symptoms similar to connective tissue disease.
- **Dermatological involvement:** Neutrophilic dermatosis, Sweet's syndrome, erythematous plaques, vasculitic lesions, and recurring skin inflammation.
- **Systemic manifestations:** Recurrence of fever, fatigue, weight loss, lung inflammation, eye problems, and thromboembolic problems.

Since the discovery of VEXAS syndrome, more instances have been reported in North America, Europe, Japan, and elsewhere, implying that there might be under-diagnosis of the disease globally[11]. The majority of the patients diagnosed with VEXAS syndrome are usually older male patients since the UBA1 gene is X-linked; yet, there are a few reports of female cases with distinct genetic predisposition. Knowledge about the existence of VEXAS syndrome is still scarce in India, where only a handful of cases have been identified so far[12]. Yet, the growing availability of next-generation sequencing technology and awareness regarding rare auto-inflammatory diseases should lead to better detection rates in the future. The creation of national patient registries, raising awareness among physicians, and utilizing genome-based diagnostics in the routine clinical practice could greatly help in revealing the full extent of the problem.

3. AI-DRIVEN GENOMICS, 3D GENOMIC FINGERPRINTING AND DIAGNOSTIC INNOVATIONS

Recent advances in genomics-based medicine, AI, and NGS technology have greatly contributed to a better understanding and identification of the VEXAS syndrome, a relatively rare condition that is usually hard to diagnose because it presents similar symptoms with other autoimmune-inflammatory and hematological disorders[13]. The use of advanced techniques such as AI-assisted analysis, multi-omics approach, and novel 3D Genomic Fingerprinting has helped detect pathogenic mutations in the UBA1 gene, among other aspects.

3.1 Current Diagnostic Approaches and Genomic Technologies

Diagnosis of the VEXAS syndrome is complex due to the high level of clinical heterogeneity associated with the disease along with overlapping clinical features with other rheumatological, cutaneous, and hematological conditions[14]. These include repeated fevers, relapsing polychondritis, vasculitis, lung inflammation, neutrophilic cutaneous conditions, and cytopenias without clear reason. Thus, patients have to go through various clinical assessments until the genetic basis of the condition is recognized. Routine blood tests like elevated levels of inflammatory markers and macrocytic anemia can be indicative of the disease but cannot confirm diagnosis[15].

Bone marrow examination holds a great value in the diagnostic process for the cases in question. One of the features observed is that of myeloid and erythroid cell precursors containing cytoplasmic vacuoles, a finding after which the disease was named VEXAS syndrome[16]. The above-mentioned symptoms are not specific to the condition in question, but rather may suggest conducting molecular studies. With the advent of NGS, diagnosing VEXAS syndrome became easier due to direct detection of UBA1 somatic mutations.

Modern genomics techniques allow identifying mutations, quantifying the clonal size, and analyzing the heterogeneity of the disease. As a result, molecular genetic testing has become a gold standard in confirming the diagnosis[17].

3.2 Artificial Intelligence and Multi-Omics Integration

AI has become an important constituent of genomics medicine owing to its capacity to deal with big amounts of biological information. Using AI techniques is helpful in detecting the existence of pathological alterations and molecular signatures within a genomic sample in relation to the VEXAS syndrome, where machine learning algorithms allow one to analyze sequences in order

to make conclusions regarding their impact. Another approach to genomics research involves deep learning algorithms, which enable the integration of multiple molecular parameters into diagnostics[18].

Deep learning is especially effective at facilitating early diagnosis and reducing the number of patients whose diseases are undiagnosed because of various reasons. Moreover, such algorithms help classify diseases based on various genomic signatures. Given the continuous development of genomic datasets, the importance of AI will continue to grow.

Alongside the use of AI in studying VEXAS syndrome, another effective method which has become popular is that of multi-omics integration[19]. This technique involves the merging of genomics, transcriptomics, proteomics, metabolomics, and epigenomics in order to get an overall picture of the disease and understand the complicated biological mechanisms behind the process. In addition, multi-omics approach might help in identifying potential biomarkers for early detection and disease monitoring.

3.3 3D Genomic Fingerprinting and Precision Medicine Applications

With advancements made in recent times in the field of genome biology, it has been found that, apart from genetic mutations, the spatial organization of the genome in the cell nucleus plays a major role in disease pathogenesis[20]. With this discovery, there is the emergence of a new science known as 3D genomics, which entails studying genome architecture and its spatial interaction. While the traditional sequencing technique mainly emphasizes changes in the genetic sequence of the DNA, 3D genomics technology provides information on the influence of structural genome arrangements on gene expression and cell functions[21]. In diseases involving inflammation and hematological processes, such as VEXAS syndrome, chromatin structure may play a role in immune signaling and other cellular responses.

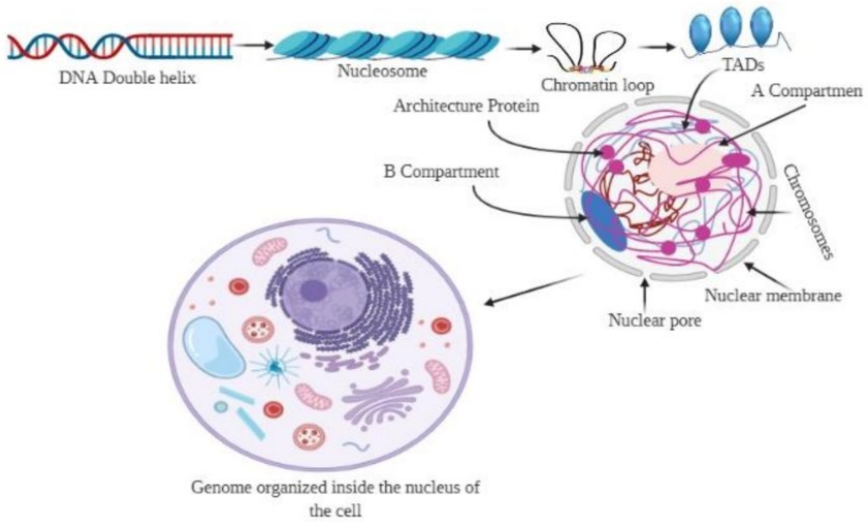

Figure 2: Hierarchical Organization of the Genome and Chromatin Architecture in 3D Genomics

Combining 3D genomics fingerprinting with the use of artificial intelligence and precision medicine offers many possibilities regarding disease profiling and personalized care. Using both

the spatial data from the genomic structure and the clinical or molecular data, scientists will be able to develop genomic signatures for diseases. Such fingerprints would provide better diagnostic results, predict prognoses, and aid in developing treatment plans based on personal molecular data[22].

The major application of 3D genomics fingerprinting in VEXAS syndrome is:

- Chromatin structure analysis and studies of gene regulatory networks.
- Studies related to spatial genomic interactions that relate to inflammatory pathways.
- Identification of molecular signatures characteristic of specific diseases.
- Integrating genomic data with patient information for personalized diagnosis[23].
- Development of precision medicines and personalized treatment plans.

Further development in 3D genomics technology will be important in improving knowledge about the biology behind diseases and in the future of precision medicine[24]. While still largely experimental in nature, combining these technologies with AI-driven analytics and multi-omics platforms could potentially change how we diagnose and treat VEXAS syndrome and other rare genetic conditions.

4. THERAPEUTIC PATHWAYS AND CURATIVE STEM CELL TRANSPLANTATION

Management of VEXAS syndrome continues to pose challenges, owing to the complexity of its pathogenesis, multiple organ/system involvement, and treatment-refractoriness to many conventional treatments[25]. Given the fact that VEXAS syndrome results from somatic mutations in UBA1 and clonal hematopoiesis, the mainstay of treatment is mainly geared towards managing the inflammatory processes and minimizing the associated complications rather than treating the underlying mutation itself. Over the past few years, remarkable advances have been witnessed in terms of elucidation of the current status and future prospects of therapeutics in relation to VEXAS syndrome. Of all the modalities discussed in the preceding section, the most promising one appears to be HSCT, which can even be considered curative[26].

4.1 Current Therapeutic Strategies

The management of VEXAS syndrome largely revolves around addressing systemic inflammation, mitigating the symptoms, and dealing with hematological disturbances. The underlying cause of this condition is somatic UBA1 mutation and clonal hematopoiesis. Therefore, existing treatment options only manage the symptoms and do not address the primary pathology. Steroids have been used extensively for the treatment of VEXAS syndrome and have proven to be effective for alleviating inflammatory symptoms in the early stages of the disease. Unfortunately, steroids are often used for an extended duration in most cases, leading to side effects[27].

The common treatments for VEXAS syndrome are:

- **Corticosteroids:** First-line agents that can be used for suppressing systemic inflammation and disease flares[28].
- **JAK inhibitors:** Target cytokine-induced inflammatory pathway and have been successful in controlling disease activities.

- **Azacitidine:** Specifically useful in individuals who have co-existing myelodysplastic syndromes by improving hematological and inflammatory factors.
- **Biologic agents:** Medications that inhibit inflammatory mediators such as IL-6 and TNF-α, despite varying effectiveness in individual patients.

Even though they are beneficial in a clinical setting, these medications have not been successful in eradicating the underlying mutant hematopoietic clone[29].

4.2 Stem Cell-Based Therapeutic Approaches

The use of stem cells as a treatment modality has gained momentum due to its capability of providing solutions for correcting cellular anomalies responsible for VEXAS syndrome. The uniqueness of stem cells lies in their capability of undergoing self-renewal and differentiation to become different types of cells. This makes stem cells suitable for the treatment of hematologic disorders and immune-related diseases, as pathogenic mutations in UBA1 arise from hematopoietic stem and progenitor cells, which may render the use of anti-inflammatory drugs insufficient.

In addition to being involved in cellular replacement, stem cells display remarkable immunomodulatory functions that can regulate inflammation. The production of numerous cytokines, growth factors, and exosomes by stem cells plays an essential role in the regulation of the immune system and maintenance of cellular balance[30]. Thus, much attention has been devoted to exploring the potential of stem cells in regulating chronic inflammation.

Despite the fact that the use of stem cell-based treatments for VEXAS syndrome remains mostly experimental, there are still some advances in the field of regenerative medicine that can help in this matter. Today's studies are aimed at understanding how stem cells affect the immune system and aid in repairing tissues. Such advances may help in developing a new method of treating patients diagnosed with VEXAS syndrome[31].

4.3 Allogeneic Hematopoietic Stem Cell Transplantation: Curative Potential and Challenges

At present, allogeneic HSCT is the most likely potentially curative option for VEXAS syndrome. While traditional treatments involve suppressing inflammation caused by VEXAS syndrome, transplant therapy acts on the basic principle behind the pathogenesis of the condition by removing the mutant clones in patients and replacing them with healthy donor cells[32].

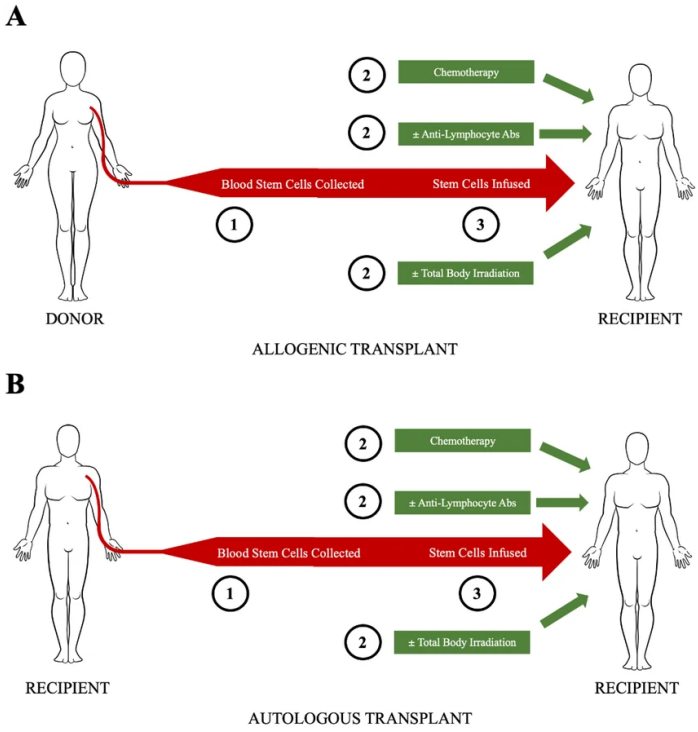

Figure 3: Schematic Representation of Allogeneic Hematopoietic Stem Cell Transplantation (HSCT) as a Potentially Curative Therapy for VEXAS Syndrome

Clinical improvement has been reported in recent case series, cohorts, and multi-center trials after transplantation[33]. There have been positive clinical effects in terms of inflammatory symptoms, hematological conditions, and improved quality of life among most individuals. This indicates the possibility of the use of HSCT to achieve long-term disease control since it addresses the underlying genetic and cellular basis of the disease.

However, the use of HSCT comes with many clinical difficulties. Age, co-morbidity, and severity of the condition among others can contribute to various complications associated with transplantation including graft-versus-host disease, infections, organ toxicity, and graft rejection[34]. Therefore, the choice of suitable candidates and donor, as well as proper conditioning regimens, should be prioritized to ensure positive outcomes from therapy.

5. DISCUSSION

With an increasing number of research findings on VEXAS syndrome, our knowledge about molecular mechanisms, signs and symptoms, diagnoses, and treatment strategies of the condition has greatly improved. For example, the discovery of somatic mutations in the UBA1 gene has shown the connection between clonal hematopoiesis and systemic inflammation, and new developments in the field of genomics have led to better diagnosis and characterization of VEXAS syndrome. Additionally, innovative technologies such as artificial intelligence and stem cell therapy open up promising possibilities for precise disease diagnosis and management.

Table 1: Summary of Selected Studies on VEXAS Syndrome, AI Diagnostics, and Stem Cell Transplantation

Author(s) & Year	Study Focus	Key Findings	Relevance to Present Review
Sofi et al. (2024)[35]	Clinical presentation of VEXAS syndrome	Reported diffuse alveolar haemorrhage as a rare but severe manifestation of VEXAS syndrome.	Highlights the diverse and multisystem clinical spectrum of the disease.
Stiburkova et al. (2023)[36]	Novel UBA1 mutation identification	Identified a previously unreported somatic UBA1 variant associated with VEXAS syndrome.	Supports the discussion on genetic heterogeneity and molecular diagnosis.
van Leeuwen-Kerkhoff et al. (2022)[37]	Allogeneic stem cell transplantation	Demonstrated successful upfront allogeneic HSCT in a patient with VEXAS syndrome.	Provides evidence for the curative potential of stem cell transplantation.
Zhang et al. (2015)[38]	Mesenchymal stem cell therapy	Discussed opportunities and challenges of allogeneic mesenchymal stem cells in regenerative medicine.	Supports the broader role of stem cell-based therapeutic strategies.
Zhang & Wang (2024)[39]	Artificial intelligence in genomics	Demonstrated the application of machine learning in genomic data analysis and biological research.	Reinforces the importance of AI-driven diagnostics and precision medicine.

5.1 Interpretation and Analysis of Findings

These studies show that VEXAS syndrome is an autoinflammatory disease with somatic mutations in the UBA1 gene and clonal hematopoiesis. There is ample evidence of the close link between genetic abnormalities, immune dysregulation, and organ involvement. Thanks to progress in genome sequencing techniques, disease diagnosis has been facilitated. Moreover, thanks to new analytical tools based on artificial intelligence, the complexity of genetic information is easier to decipher. On top of that, recent publications related to stem cell transplantation point to the possibility of eliminating mutant clone hematopoiesis and treating VEXAS syndrome.

5.2 Implications and Significance

VEXAS syndrome has emerged as a clinically significant entity, and this condition is likely to have an impact on both medical practice and genomics. First of all, early detection of UBA1 gene mutations will aid in precise diagnosis and timely initiation of therapy[40]. The incorporation of artificial intelligence and genomic innovations could enhance the accuracy of diagnostics and

patients' stratification. Moreover, successful allogeneic stem cell transplantation opens new perspectives for personalized medicine.

5.3 Research Gaps and Limitations

Despite the progress made, there are still many challenges in the current knowledge of VEXAS syndrome. Most of the information is based on case series and reports involving few patients; thus, conclusions may not apply to all patients. Moreover, epidemiological information is lacking, especially in third-world countries where people lack access to genetic testing. Finally, the impact of the various UBA1 gene mutations on the disease and patients' response to treatment remains unclear. More research needs to be done in order to fill these gaps.

5.4 Future Research Directions

Research into the future needs to concentrate more on increasing the size of international patient registries and conducting genotype-phenotype correlation studies. Validation of AI diagnostics is also an area that requires more study and investigation. The potential use of 3D genomic fingerprinting can be looked at for clinical purposes as well. Research is required in stem cell transplantation results evaluation to refine treatment approaches and determine patient eligibility. Gene editing and molecular therapy may also have possibilities for development.

6. CONCLUSION

VEXAS is an emerging autoinflammatory disorder associated with somatic mutations in the UBA1 gene, which connect clonal hematopoiesis with systemic inflammation and multisystem involvement. Innovations like genomic sequencing, artificial intelligence, multilayer omics analysis, and 3D genomic signature mapping have enhanced our comprehension of the disorder's etiopathogenesis and diagnostic applications. Although various treatments including corticosteroids, Jak inhibitors, azacitidine, and biologic therapy manage the condition, none of these eradicate the mutational clone. On the other hand, allogenic bone marrow transplantation holds immense potential as a promising curative modality. There are various issues surrounding early detection, limited epidemiological information, and treatment efficacy that need addressing.

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