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ORIGINAL ARTICLE

Clinical and Genetic Model for Predicting Outcomes in Patients with Retinal Vein Occlusion

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Abstract

Retinal vein occlusion is one of the most common vascular disorders of the retina and represents a significant cause of visual impairment and irreversible vision loss among middle-aged and elderly populations. The disease develops due to obstruction of venous blood flow in the retinal circulation, resulting in retinal ischemia, hemorrhage, macular edema, and progressive deterioration of visual function. Despite advances in ophthalmic diagnostics and treatment strategies, the clinical course of retinal vein occlusion remains highly variable, making prediction of visual outcomes and recurrence risk challenging. The present study focuses on the development of a clinical and genetic predictive model designed to evaluate the prognosis of patients diagnosed with retinal vein occlusion. The model integrates traditional systemic risk factors such as arterial hypertension, diabetes mellitus, and dyslipidemia with molecular genetic markers including JAK2, SOD2, and MTHFR gene polymorphisms. The analysis demonstrates that combining clinical parameters with genetic indicators significantly improves the accuracy of predicting visual outcomes, the likelihood of disease progression, and the risk of recurrence. The proposed predictive model may contribute to more individualized management strategies, early identification of high-risk patients, and improved prevention of severe visual complications associated with retinal vascular occlusive disorders. Retinal vein occlusion represents a significant retinal vascular disorder that frequently leads to visual impairment due to compromised retinal circulation, ischemic damage, and secondary macular complications. The severity of visual dysfunction and the risk of recurrence vary considerably among patients, which indicates the involvement of multiple systemic and genetic determinants in disease progression. This study explores a comprehensive prognostic approach that integrates traditional clinical risk factors with molecular genetic indicators in order to improve prediction of disease outcomes. Particular emphasis is placed on systemic conditions such as arterial hypertension, diabetes mellitus, and dyslipidemia, as well as genetic polymorphisms associated with vascular dysfunction and oxidative stress, including JAK2, SOD2, and MTHFR gene variants. The combined evaluation of these parameters allows the identification of individuals with increased susceptibility to severe retinal damage and recurrent vascular events. The results demonstrate that the integration of clinical and genetic information significantly enhances prognostic accuracy and provides valuable insights for personalized patient management. Implementation of such predictive strategies may contribute to improved monitoring, earlier therapeutic intervention, and prevention of progressive visual deterioration in patients affected by retinal venous occlusive disorders.

Keywords: *retinal vein occlusion; predictive model; genetic markers; JAK2 gene; SOD2 gene; MTHFR polymorphism; vascular eye disease; ophthalmology; visual prognosis; personalized medicine*

1. Introduction

Retinal vein occlusion is considered the second most common retinal vascular disorder after diabetic retinopathy and represents a major cause of vision impairment worldwide. The condition occurs when venous outflow from the retina becomes obstructed, leading to increased intravascular pressure, vascular leakage, retinal hemorrhage, and ischemic damage to retinal tissues. Depending on the location of the obstruction, the disease may manifest as central retinal vein occlusion or branch retinal vein occlusion, both of which can lead to serious structural and functional alterations of the retina. A number of systemic and local risk factors contribute to the development of retinal venous obstruction. Among the most significant systemic conditions are arterial hypertension, diabetes mellitus, dyslipidemia, and disorders of blood coagulation. These factors promote vascular endothelial dysfunction, increased blood viscosity, and thrombotic events within the retinal circulation. However, recent research has demonstrated that genetic predisposition also plays an important role in determining susceptibility to vascular occlusive diseases. Molecular genetic markers associated with oxidative stress, thrombosis, and vascular inflammation have been identified as potential contributors to the pathogenesis and progression of retinal vein occlusion. Polymorphisms in genes such as JAK2, SOD2, and MTHFR have been linked to altered cellular responses to oxidative stress, impaired vascular regulation, and increased thrombotic risk. Despite the growing body of knowledge regarding these mechanisms, predicting the clinical course and long-term outcomes of retinal vein occlusion remains complex. Development of a comprehensive prognostic model that integrates both clinical and genetic determinants may significantly improve the ability of clinicians to evaluate disease progression and select appropriate treatment strategies for affected patients. Retinal venous occlusion is considered one of the most prevalent vascular diseases of the retina and is recognized as a major cause of visual disability in ophthalmic practice. The condition occurs when obstruction develops within the retinal venous system, resulting in impaired blood drainage, increased intravascular pressure, and subsequent retinal hemorrhage and edema. In many cases, prolonged circulatory disturbance leads to retinal ischemia and structural damage affecting the neurosensory layers of the retina. The clinical course of the disease may vary widely, ranging from mild visual disturbances to severe and irreversible loss of vision. Numerous systemic conditions have been identified as important contributors to the development of this pathology. Chronic arterial hypertension, metabolic disturbances related to diabetes mellitus, and abnormalities in lipid metabolism significantly increase the probability of vascular occlusive events. These disorders promote endothelial dysfunction, vascular wall thickening, and increased blood viscosity, which collectively contribute to thrombotic processes in retinal vessels. In addition to systemic risk factors, modern biomedical research has highlighted the importance of genetic predisposition in vascular diseases of the eye. Variations in genes involved in oxidative stress defense, homocysteine metabolism, and hematologic regulation may influence the vulnerability of retinal vessels to occlusion and subsequent complications. Understanding the combined influence of clinical and molecular determinants is essential for improving prognostic evaluation and developing more individualized therapeutic strategies for patients suffering from retinal vein occlusion.

2. Materials and Methods

The present study involved patients diagnosed with retinal vein occlusion who underwent comprehensive ophthalmological and systemic evaluation. Clinical data were collected to assess the presence of major systemic risk factors including arterial hypertension, diabetes mellitus, and lipid metabolism disorders. Ophthalmic examination included measurement of best corrected visual acuity, slit-lamp biomicroscopy, intraocular pressure assessment, fundus examination, and optical coherence tomography to evaluate structural changes within the retina. Fluorescein angiography was performed when necessary to assess retinal perfusion and identify areas of ischemia. In addition to clinical assessment, molecular genetic analysis was conducted in order to identify polymorphisms associated with vascular dysfunction and oxidative stress. Peripheral blood samples were obtained from all participants for DNA extraction and genotyping of selected markers. Particular attention was given to genetic variants of the JAK2 gene associated with abnormal hematologic activity, the SOD2 gene involved in antioxidant defense mechanisms, and the MTHFR gene which influences homocysteine metabolism and vascular health. Statistical analysis was performed to evaluate the association between clinical risk factors, genetic markers, and disease outcomes. A predictive model was constructed using multivariate analytical methods to determine the probability of visual deterioration and recurrence of retinal vein occlusion.

3. Results

The analysis revealed a strong association between systemic vascular risk factors and the severity of retinal vein occlusion. Patients with arterial hypertension and diabetes mellitus demonstrated more pronounced retinal ischemia and higher incidence of macular edema compared with individuals without these conditions. Dyslipidemia was also found to contribute to vascular endothelial dysfunction and increased risk of retinal vascular obstruction. Genetic analysis

demonstrated significant correlations between certain molecular markers and disease progression. Variants of the MTHFR gene were associated with elevated homocysteine levels and increased thrombotic tendency within the retinal circulation. Polymorphisms of the SOD2 gene were linked to reduced antioxidant activity, which may enhance oxidative stress and promote vascular damage in retinal tissues. Mutations in the JAK2 gene showed potential association with altered hematological parameters and increased risk of recurrent vascular occlusion. Integration of these genetic indicators with clinical risk factors allowed the development of a comprehensive predictive model. The model demonstrated high accuracy in identifying patients at increased risk of severe visual loss and recurrent retinal vein occlusion. Patients categorized as high-risk based on the combined clinical and genetic profile showed significantly poorer visual prognosis during follow-up observation. Comprehensive evaluation of the examined patient population revealed clear associations between systemic vascular disorders and the severity of retinal damage. Individuals with long-standing arterial hypertension showed more pronounced retinal hemorrhages, greater macular involvement, and higher incidence of ischemic complications. Patients diagnosed with diabetes mellitus exhibited increased retinal vascular permeability and greater frequency of macular edema, which significantly affected visual function. Dyslipidemia was also identified as an important factor contributing to endothelial dysfunction and impaired retinal microcirculation. Molecular analysis demonstrated that specific genetic polymorphisms were associated with increased susceptibility to disease progression and recurrence. Variants of the MTHFR gene were correlated with elevated homocysteine levels, which may promote endothelial injury and thrombotic activity within retinal vessels. Alterations in the SOD2 gene were linked to reduced antioxidant capacity, thereby intensifying oxidative stress within retinal tissues and contributing to vascular damage. Genetic variations in the JAK2 gene showed potential influence on hematological parameters and thrombogenic activity. When these molecular markers were analyzed in combination with clinical risk factors, a predictive framework was established that demonstrated strong ability to identify patients at increased risk of severe visual impairment and recurrent occlusive events. The integrated model provided higher prognostic precision compared with traditional risk factor evaluation alone.

4. Discussion

The findings of this study support the concept that retinal vein occlusion is a multifactorial disorder influenced by both systemic clinical conditions and genetic predisposition. Traditional vascular risk factors remain fundamental contributors to the development of retinal venous obstruction. However, the addition of molecular genetic markers provides a deeper understanding of the biological mechanisms that influence disease progression and clinical outcomes. Genetic polymorphisms affecting oxidative stress regulation, homocysteine metabolism, and hematologic activity appear to modify individual susceptibility to retinal vascular damage and recurrence of occlusive events. The proposed clinical-genetic model demonstrates the potential value of integrating personalized genetic information into routine clinical assessment. Such an approach may allow early identification of patients with increased vulnerability to severe retinal complications and may guide the selection of preventive or therapeutic strategies. Furthermore, the use of predictive modeling can support ophthalmologists in determining prognosis and optimizing follow-up schedules for patients with retinal vascular disorders. The observations obtained from this investigation confirm that retinal vein occlusion is a multifactorial vascular disorder influenced by both systemic pathological conditions and inherited biological characteristics. While traditional cardiovascular and metabolic risk factors play a central role in initiating vascular obstruction, genetic predisposition may significantly affect the severity of disease progression and the likelihood of recurrence. Oxidative stress mechanisms appear to represent an important pathogenic pathway, particularly in individuals carrying variants of genes responsible for antioxidant defense. Similarly, disturbances in homocysteine metabolism caused by polymorphisms in the MTHFR gene may enhance endothelial injury and increase the risk of thrombotic events within retinal circulation. The involvement of the JAK2 gene suggests that alterations in hematologic regulation and cellular signaling pathways may further contribute to vascular instability. Integrating these molecular indicators with conventional clinical assessment provides a broader understanding of the pathophysiological processes underlying retinal venous occlusion. Such an approach supports the development of personalized diagnostic strategies and may guide the selection of preventive or therapeutic interventions aimed at reducing the likelihood of severe retinal complications.

5. Conclusion

The development of a clinical and genetic predictive model significantly enhances the ability to evaluate prognosis in patients with retinal vein occlusion. Incorporation of systemic risk factors together with molecular genetic markers such as JAK2, SOD2, and MTHFR improves the accuracy of predicting visual outcomes and the probability of disease recurrence. Application of this integrated approach may facilitate early identification of high-risk individuals, support personalized treatment strategies, and contribute to improved prevention of severe visual impairment associated with

retinal vascular occlusive disease. Continued research combining clinical ophthalmology and molecular genetics is expected to further refine prognostic tools and improve management of patients affected by retinal vein occlusion. The integration of clinical parameters with molecular genetic markers significantly improves the ability to evaluate the prognosis of patients with retinal vein occlusion. A combined analytical approach that includes systemic vascular risk factors together with genetic polymorphisms associated with oxidative stress and thrombosis allows more accurate identification of individuals at increased risk of visual deterioration and disease recurrence. The implementation of predictive models based on both clinical and genetic information may support personalized patient monitoring, facilitate earlier therapeutic intervention, and ultimately reduce the incidence of severe vision loss associated with retinal vascular disorders. Continued research in the field of ophthalmic genetics and vascular biology is expected to further refine prognostic tools and enhance clinical management strategies for patients affected by retinal vein occlusion.

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