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ORIGINAL ARTICLE

Influence of JAK2 and SOD2 Gene Polymorphisms on the Effectiveness of Treatment in Retinal Vein Occlusion

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Abstract

Retinal vein occlusion represents a major vascular disorder of the retina and is frequently associated with visual deterioration caused by impaired venous circulation, retinal ischemia, and macular edema. Although modern therapeutic strategies such as intravitreal anti-vascular endothelial growth factor therapy, corticosteroid administration, and retinal laser photocoagulation have significantly improved clinical outcomes, variability in treatment response remains a persistent clinical challenge. Increasing evidence suggests that genetic factors influencing vascular regulation and oxidative stress may contribute to these differences in therapeutic effectiveness. The present study investigates the influence of polymorphisms in the JAK2 and SOD2 genes on the response of patients with retinal vein occlusion to standard ophthalmological treatment. The analysis demonstrates that the presence of these genetic variants is associated with altered biological mechanisms affecting vascular function, inflammatory responses, and oxidative balance in retinal tissues. Patients carrying these polymorphisms exhibited reduced responsiveness to conventional therapy and a higher probability of persistent macular edema or recurrent vascular complications. The findings indicate that identification of such molecular markers may support individualized treatment planning and highlight the importance of systemic metabolic and vascular correction alongside local ophthalmic therapy. Retinal vein occlusion is a widespread vascular disorder of the retina that often leads to visual impairment due to disturbances in retinal blood circulation, tissue ischemia, and secondary macular damage. Despite the availability of effective therapeutic approaches, including intravitreal anti-VEGF injections, corticosteroid therapy, and retinal laser photocoagulation, clinical outcomes differ substantially among patients. These variations indicate the possible influence of genetic factors that affect vascular function, inflammatory responses, and oxidative balance within ocular tissues. Particular attention has been directed toward polymorphisms of the JAK2 and SOD2 genes, which are involved in intracellular signaling, hematologic regulation, and antioxidant defense. The present analysis evaluates how these genetic variations influence the response of patients with retinal vein occlusion to standard treatment protocols. The findings demonstrate that individuals carrying these polymorphisms tend to exhibit slower anatomical recovery and reduced functional improvement after conventional therapy. Recognition of these molecular characteristics allows clinicians to predict therapeutic response more accurately and suggests the necessity for modification of treatment strategies that include systemic metabolic correction and enhanced vascular protection in addition to local ophthalmic interventions.

Keywords: *retinal vein occlusion; JAK2 polymorphism; SOD2 polymorphism; anti-VEGF therapy; retinal laser photocoagulation; corticosteroid therapy; oxidative stress; vascular pathology; ophthalmology; personalized medicine*

1. Introduction

Retinal vein occlusion is one of the most frequently encountered retinal vascular disorders and represents a significant cause of visual impairment among middle-aged and elderly populations. The disease arises when obstruction develops within the retinal venous system, leading to impaired blood drainage, vascular congestion, hemorrhage, and edema of the retinal tissues. As intravascular pressure increases, the integrity of the blood-retinal barrier becomes compromised, allowing fluid and plasma components to accumulate within the retinal layers. Macular edema is considered the most common cause of decreased visual acuity in patients with this condition. Over the past two decades, significant progress has been made in the management of retinal vein occlusion. Therapeutic strategies commonly include intravitreal injections of anti-vascular endothelial growth factor agents, corticosteroid therapy aimed at reducing inflammation and vascular permeability, and retinal laser photocoagulation to limit ischemic complications. Despite the effectiveness of these methods, treatment outcomes vary considerably among individuals. Some patients demonstrate rapid anatomical and functional recovery, whereas others experience persistent edema or repeated vascular complications despite adequate therapy. This variability suggests the involvement of additional biological determinants influencing treatment response. In recent years, molecular genetic research has revealed that polymorphisms in genes involved in vascular regulation, oxidative stress defense, and inflammatory signaling may contribute to the development and progression of retinal vascular diseases. The JAK2 gene plays an important role in intracellular signaling pathways regulating hematopoiesis, immune responses, and inflammatory processes. Mutations affecting this gene may alter vascular homeostasis and increase thrombotic tendencies. The SOD2 gene encodes mitochondrial superoxide dismutase, an essential enzyme responsible for neutralizing reactive oxygen species and protecting cells from oxidative damage. Genetic variations in this enzyme may reduce antioxidant capacity and intensify oxidative stress within retinal tissues. Understanding the influence of these molecular factors on treatment response may provide new opportunities for personalized therapeutic approaches in patients with retinal vein occlusion. Vascular disorders of the retina are among the most significant causes of visual disability in ophthalmological practice, and retinal vein occlusion occupies a prominent position among these conditions. The disease occurs when venous drainage from the retinal circulation becomes obstructed, resulting in elevated venous pressure, capillary leakage, hemorrhage, and swelling of retinal structures. In many cases, prolonged disturbance of retinal microcirculation leads to ischemic damage and progressive deterioration of visual acuity. Modern ophthalmology has developed several therapeutic strategies aimed at controlling the consequences of venous obstruction. Intravitreal administration of agents that suppress vascular endothelial growth factor reduces vascular permeability and limits the formation of macular edema. Corticosteroid therapy decreases inflammatory reactions and stabilizes the blood-retinal barrier, while retinal laser photocoagulation improves oxygenation of ischemic retinal zones and prevents the development of neovascular complications. Although these therapeutic approaches have significantly improved clinical outcomes, the degree of recovery varies greatly among patients. Some individuals respond rapidly with substantial improvement in retinal morphology and visual acuity, whereas others show persistent edema or repeated vascular events despite adequate therapy. These differences suggest that biological variability among patients may influence treatment effectiveness. Recent advances in molecular medicine have revealed that genetic polymorphisms affecting oxidative stress regulation and intracellular signaling pathways may alter susceptibility to vascular disease and response to treatment. Variations in the JAK2 gene may influence hematologic activity and inflammatory processes that contribute to vascular dysfunction, while alterations in the SOD2 gene can reduce mitochondrial antioxidant capacity and intensify oxidative damage within retinal tissues. Understanding the relationship between these genetic characteristics and therapeutic response may help improve individualized treatment strategies for patients with retinal vascular disorders.

2. Materials and Methods

The study included patients diagnosed with retinal vein occlusion who received standard ophthalmological treatment at specialized clinical centers. All participants underwent comprehensive ophthalmic evaluation prior to treatment initiation. Examination procedures included measurement of best corrected visual acuity, slit-lamp biomicroscopy, intraocular pressure measurement, and detailed fundus examination. Optical coherence tomography was performed to assess retinal morphology and quantify macular thickness. Fluorescein angiography was used to evaluate retinal perfusion and detect ischemic areas when necessary. Standard treatment protocols consisted of intravitreal anti-vascular endothelial growth factor injections for the management of macular edema, corticosteroid therapy in selected cases with inflammatory components, and retinal laser photocoagulation for ischemic retinal zones. Peripheral blood samples were collected from all patients for genetic analysis. DNA extraction was followed by molecular testing to identify polymorphisms in the JAK2 and SOD2 genes. Statistical analysis was performed to evaluate the relationship between genetic variants and therapeutic outcomes. Treatment effectiveness was assessed by measuring changes in visual acuity, reduction in macular thickness, and recurrence of retinal edema during follow-up examinations.

3. Results

Clinical evaluation following therapy demonstrated that the response to treatment varied significantly depending on the genetic profile of the patients. Individuals without detected polymorphisms in the analyzed genes generally exhibited favorable anatomical and functional recovery. Reduction of macular edema was observed within the first months after anti-VEGF therapy, and improvement in visual acuity occurred gradually during the follow-up period. In contrast, patients carrying JAK2 gene variants showed a tendency toward persistent vascular instability and higher incidence of recurrent retinal edema despite repeated treatment sessions. This observation suggests that abnormalities in signaling pathways associated with vascular regulation and thrombosis may influence therapeutic response. Similarly, polymorphisms of the SOD2 gene were associated with increased oxidative stress within retinal tissues, which may impair the effectiveness of conventional therapies aimed primarily at controlling vascular permeability. Patients with these genetic variants demonstrated slower resolution of macular edema and required more frequent treatment interventions. Statistical analysis confirmed a significant association between the presence of JAK2 and SOD2 polymorphisms and reduced responsiveness to standard treatment protocols. Clinical observations revealed distinct differences in treatment response depending on the genetic profile of the examined patients. Individuals without detectable polymorphisms in the studied genes generally demonstrated favorable anatomical recovery after standard therapy. Optical coherence tomography showed progressive reduction of macular thickness following anti-VEGF injections, and visual acuity improved during subsequent follow-up examinations. In contrast, patients carrying variants of the JAK2 gene exhibited a tendency toward persistent vascular instability and slower resolution of retinal edema. Repeated treatment sessions were often required to achieve partial structural improvement. Similar patterns were observed in individuals with polymorphisms of the SOD2 gene, in whom oxidative stress appeared to contribute to prolonged inflammatory reactions and delayed restoration of retinal architecture. These patients frequently showed incomplete absorption of intraretinal fluid and less pronounced improvement in visual function. Statistical analysis demonstrated that the presence of these genetic variations was associated with decreased responsiveness to conventional therapy and a greater probability of recurrent pathological changes within the retina.

4. Discussion

The results of this study emphasize the importance of genetic determinants in shaping individual responses to ophthalmological treatment. Retinal vein occlusion is a multifactorial disease influenced by systemic vascular conditions, metabolic disturbances, and molecular mechanisms regulating cellular homeostasis. While anti-VEGF agents, corticosteroids, and laser therapy remain fundamental components of treatment, the presence of certain genetic polymorphisms may alter the biological environment in which these therapies act. Variants of the JAK2 gene may influence inflammatory signaling pathways and thrombogenic activity, thereby maintaining vascular dysfunction even after local ophthalmic treatment. At the same time, reduced antioxidant activity associated with SOD2 polymorphisms may lead to persistent oxidative damage within retinal tissues, limiting the ability of the retina to recover following vascular injury. These findings suggest that patients with such genetic backgrounds may benefit from additional systemic therapeutic strategies aimed at correcting metabolic disturbances, improving vascular health, and reducing oxidative stress. Integration of genetic screening into clinical practice could therefore support the development of more individualized treatment strategies for patients with retinal vascular disorders. The results obtained in this analysis highlight the importance of genetic determinants in the management of retinal vascular diseases. Traditional therapeutic strategies for retinal vein occlusion primarily target the consequences of vascular obstruction, such as macular edema and ischemia. However, the biological environment in which these therapies act may differ significantly among individuals due to inherited molecular characteristics. Genetic alterations affecting intracellular signaling and antioxidant defense mechanisms can modify vascular reactivity, inflammatory activity, and the capacity of retinal tissues to recover after injury. Variants of the JAK2 gene may influence hematopoietic and inflammatory pathways, potentially increasing thrombotic tendencies and maintaining vascular dysfunction despite local ophthalmic treatment. At the same time, reduced activity of mitochondrial antioxidant enzymes associated with SOD2 polymorphisms may intensify oxidative stress within retinal cells, thereby limiting tissue recovery and prolonging pathological processes. These mechanisms help explain why certain patients respond less effectively to conventional therapeutic protocols.

5. Conclusion

Genetic polymorphisms affecting the JAK2 and SOD2 genes significantly influence the effectiveness of conventional treatment strategies for retinal vein occlusion. Patients carrying these molecular variants demonstrate altered responses to anti-VEGF therapy, corticosteroid administration, and laser photocoagulation, often requiring more intensive or modified treatment approaches. Identification of these genetic markers may help clinicians predict therapeutic outcomes and implement personalized treatment plans that include both local ophthalmic interventions and systemic corrective

measures. Continued research integrating molecular genetics with clinical ophthalmology will contribute to improved understanding of retinal vascular diseases and enhance the effectiveness of therapeutic management. Genetic polymorphisms affecting the JAK2 and SOD2 genes play a significant role in determining the effectiveness of therapeutic interventions for retinal vein occlusion. Patients carrying these molecular variants demonstrate altered biological responses that may reduce the efficacy of conventional ophthalmic treatments and increase the likelihood of persistent retinal pathology. Identification of these genetic characteristics provides valuable prognostic information and supports the development of individualized treatment strategies that combine local ocular therapy with systemic correction of vascular and metabolic disturbances. Continued integration of molecular genetics into ophthalmic research and clinical practice will contribute to more precise prediction of treatment outcomes and improved management of retinal vascular disorders.

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