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ORIGINAL ARTICLE

# Comparative Analysis of the Clinical Course of Retinal Vein Occlusion in Patients With and Without Thrombophilic Mutations

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## Abstract

Retinal vein occlusion is one of the most common vascular diseases of the retina and a frequent cause of sudden visual impairment. Although traditional systemic factors such as hypertension, diabetes mellitus, and dyslipidemia play an important role in its pathogenesis, increasing attention has been directed toward the contribution of genetic predisposition to thrombosis. Mutations associated with thrombophilia may influence the severity of retinal vascular obstruction and affect the response to treatment. The present study provides a comparative analysis of the clinical course of retinal vein occlusion in patients with and without thrombophilic mutations. A total of 86 patients diagnosed with retinal vein occlusion were evaluated using clinical, ophthalmological, and genetic methods. Particular attention was given to polymorphisms of the JAK2 and MTHFR genes, which are associated with abnormalities in coagulation regulation and homocysteine metabolism. The results demonstrated that the presence of these mutations was significantly associated with a more severe clinical course of the disease, slower recovery of visual acuity, and a higher frequency of complications including persistent macular edema and recurrent vascular events. The findings emphasize the importance of genetic screening in patients with retinal vein occlusion in order to identify individuals at higher risk of unfavorable outcomes and to optimize therapeutic strategies. Retinal vein occlusion is one of the most common retinal vascular disorders and represents a major cause of sudden vision impairment in ophthalmic practice. Although traditional systemic risk factors such as hypertension, metabolic disorders, and vascular degeneration play a substantial role in its development, recent investigations indicate that genetic predisposition to thrombosis may significantly influence disease progression and clinical outcomes. Mutations associated with thrombophilia, particularly in genes regulating coagulation and homocysteine metabolism, may contribute to increased susceptibility to vascular occlusion and determine the severity of retinal damage. The present analysis focuses on a comparative evaluation of the clinical course of retinal vein occlusion in individuals with and without thrombophilic genetic mutations. Special attention is given to variants of the JAK2 and MTHFR genes, which are associated with alterations in hematologic regulation and vascular endothelial function. The results indicate that patients carrying these genetic variants tend to experience a more aggressive disease course, slower improvement of visual function, and a higher incidence of complications. Recognition of these molecular determinants allows improved prognostic assessment and supports the implementation of personalized diagnostic and therapeutic strategies for patients affected by retinal vascular pathology.

**Keywords:** *retinal vein occlusion; thrombophilia; JAK2 mutation; MTHFR polymorphism; vascular thrombosis; macular edema; ophthalmology; retinal vascular disease; visual acuity recovery; genetic risk factors*

## 1. Introduction

Retinal vascular disorders remain a major cause of visual impairment worldwide, and retinal vein occlusion is among the most frequently encountered conditions in ophthalmic practice. The disease develops when the normal outflow of blood from the retinal circulation becomes obstructed, leading to increased venous pressure, vascular leakage, hemorrhage, and ischemic damage to retinal tissues. These changes may result in macular edema and progressive loss of visual function. Traditionally, the development of retinal vein occlusion has been associated with systemic cardiovascular and metabolic conditions such as arterial hypertension, diabetes mellitus, atherosclerosis, and disorders of lipid metabolism. These factors contribute to endothelial dysfunction and increased blood viscosity, thereby creating conditions that favor thrombus formation within retinal vessels. However, the clinical course of the disease varies significantly among patients, suggesting that additional biological factors may influence disease severity and recovery. Recent research has highlighted the role of genetic predisposition in vascular disorders, particularly mutations associated with thrombophilia. Thrombophilia refers to a condition characterized by an increased tendency for blood clot formation due to inherited or acquired abnormalities in coagulation pathways. Among the genetic variants most frequently studied in relation to vascular diseases are mutations affecting the JAK2 and MTHFR genes. The JAK2 gene is involved in intracellular signaling pathways that regulate hematopoiesis and inflammatory responses, while the MTHFR gene plays an important role in homocysteine metabolism and vascular health. Polymorphisms affecting these genes may alter the balance of coagulation and increase susceptibility to thrombotic events. Understanding how these genetic factors influence the clinical course of retinal vein occlusion may help improve diagnostic evaluation and allow more individualized management of patients suffering from this condition. Vascular diseases of the retina are among the leading causes of visual disability worldwide, and occlusion of the retinal venous system represents a significant portion of these conditions. This disorder occurs when the normal outflow of blood through retinal veins becomes obstructed, resulting in venous congestion, increased hydrostatic pressure, and leakage of blood components into surrounding retinal tissues. As the pathological process progresses, hemorrhages, edema, and ischemic damage may develop, ultimately leading to impairment of central vision. Clinical manifestations vary widely, ranging from mild visual disturbances to severe and irreversible vision loss. The development of retinal venous obstruction has long been associated with systemic cardiovascular conditions including arterial hypertension, metabolic abnormalities related to diabetes, and degenerative vascular changes. These factors contribute to endothelial dysfunction and structural changes in vascular walls, creating conditions favorable for thrombus formation. However, the variability in clinical presentation and response to treatment among patients suggests the involvement of additional biological mechanisms. In recent years, advances in molecular medicine have highlighted the importance of inherited predisposition to thrombosis as a contributing factor in retinal vascular disorders. Genetic mutations affecting coagulation pathways, inflammatory signaling, and homocysteine metabolism may significantly increase the probability of vascular occlusive events. Variants of the JAK2 gene are known to influence hematopoietic activity and cellular signaling processes related to inflammation and thrombosis. At the same time, polymorphisms of the MTHFR gene affect the metabolism of homocysteine, an amino acid that can damage vascular endothelium when present at elevated levels. These molecular abnormalities may alter the course of retinal vascular disease by enhancing thrombotic activity and prolonging tissue ischemia. Understanding the influence of such genetic factors is essential for improving the prediction of disease outcomes and optimizing management strategies for patients with retinal vein occlusion.

## 2. Materials and Methods

The present study included 86 patients diagnosed with retinal vein occlusion who underwent detailed ophthalmological and laboratory examination. Patients were divided into two groups according to the presence or absence of thrombophilic genetic mutations. Comprehensive ophthalmic evaluation was performed for all participants, including measurement of best corrected visual acuity, slit-lamp biomicroscopy, intraocular pressure assessment, and dilated fundus examination. Optical coherence tomography was used to evaluate retinal structure and determine the presence and severity of macular edema. In selected cases fluorescein angiography was performed to assess retinal perfusion and identify ischemic areas. Blood samples were collected from each participant for molecular genetic analysis. DNA testing was conducted to detect polymorphisms associated with thrombophilia, with particular focus on the JAK2 and MTHFR genes. Clinical follow-up included monitoring of visual acuity, retinal morphology, and the occurrence of complications during the observation period. Statistical analysis was performed to compare clinical outcomes between patients carrying genetic mutations and those without such alterations.

## 3. Results

Analysis of the collected data revealed significant differences in the clinical course of retinal vein occlusion between the two study groups. Patients without detected thrombophilic mutations generally exhibited a more favorable clinical

progression. Reduction of macular edema and improvement of retinal morphology were observed during follow-up examinations, and visual acuity gradually improved following treatment. In contrast, individuals carrying JAK2 and MTHFR mutations demonstrated a more severe manifestation of the disease. These patients frequently presented with extensive retinal hemorrhages, pronounced macular edema, and signs of retinal ischemia during initial examination. Recovery of visual acuity occurred more slowly and remained incomplete in many cases. The frequency of complications, including persistent edema and recurrent vascular occlusion, was also higher in this group. Statistical evaluation confirmed a significant association between the presence of thrombophilic genetic variants and unfavorable clinical outcomes. The comparative evaluation of clinical data revealed notable differences between patients carrying thrombophilic genetic variants and those without such mutations. Individuals lacking detectable polymorphisms generally demonstrated a more favorable clinical course. Following appropriate treatment, gradual reduction of retinal edema and partial restoration of visual acuity were observed during follow-up examinations. Structural imaging of the retina showed progressive improvement of macular morphology and stabilization of retinal circulation. In contrast, patients with JAK2 or MTHFR gene mutations frequently presented with more severe initial manifestations, including extensive retinal hemorrhages and pronounced macular edema. These individuals often experienced delayed improvement in visual function and persistent morphological alterations of the retina. Recovery of visual acuity occurred more slowly and remained incomplete in several cases. Furthermore, the occurrence of secondary complications such as recurrent vascular obstruction, prolonged edema, and ischemic changes was significantly higher in this group. These observations indicate that genetic predisposition to thrombosis may contribute to a more aggressive course of retinal vascular disease and may influence the effectiveness of therapeutic interventions.

## 4. Discussion

The results of this study support the hypothesis that genetic predisposition plays an important role in determining the severity and progression of retinal vein occlusion. While traditional systemic risk factors contribute to vascular damage and thrombus formation, inherited abnormalities in coagulation pathways may further increase susceptibility to retinal vascular obstruction. Mutations affecting the JAK2 gene may influence hematologic regulation and inflammatory signaling pathways, potentially enhancing thrombotic activity within retinal vessels. Similarly, polymorphisms in the MTHFR gene may lead to elevated homocysteine levels, which are known to promote endothelial dysfunction and vascular injury. These mechanisms may explain the more severe clinical course observed in patients carrying these genetic variants. The identification of such molecular risk factors has important clinical implications. Genetic screening in patients presenting with retinal vein occlusion may help identify individuals with underlying thrombophilic predisposition who require closer monitoring and more comprehensive systemic management. Such an approach may improve therapeutic outcomes and reduce the risk of recurrent vascular complications. The findings of this investigation emphasize the importance of considering genetic factors when evaluating patients with retinal vein occlusion. While systemic cardiovascular conditions remain major contributors to the development of retinal vascular obstruction, inherited abnormalities affecting coagulation pathways may significantly modify disease progression and prognosis. Mutations associated with thrombophilia may enhance the tendency toward thrombus formation within the retinal circulation, thereby intensifying vascular congestion and prolonging ischemic injury to retinal tissues. In the case of JAK2 gene variants, alterations in intracellular signaling mechanisms may influence hematologic activity and inflammatory processes that contribute to vascular dysfunction. Similarly, disturbances in homocysteine metabolism related to MTHFR polymorphisms may lead to endothelial damage and increased susceptibility to vascular occlusion. The combined effect of these mechanisms may explain the more severe clinical manifestations observed in patients with thrombophilic mutations. Recognition of these molecular influences has important implications for clinical practice. Genetic evaluation may assist clinicians in identifying patients who are at greater risk of unfavorable outcomes and who may benefit from more intensive monitoring or systemic therapeutic interventions. Such an approach reflects the principles of personalized medicine, in which diagnostic and treatment strategies are adapted according to the biological characteristics of each patient.

## 5. Conclusion

The comparative analysis demonstrated that the presence of thrombophilic mutations significantly influences the clinical course of retinal vein occlusion. Patients carrying genetic variants of the JAK2 and MTHFR genes tend to experience more severe disease manifestations, slower recovery of visual acuity, and a higher incidence of complications. These findings highlight the importance of incorporating genetic evaluation into the diagnostic assessment of patients with retinal vascular disorders. Early identification of thrombophilic mutations may support personalized treatment strategies and contribute to improved prognosis by addressing both ocular manifestations and systemic risk factors associated with vascular thrombosis. Comparative assessment of the clinical course of retinal vein

occlusion demonstrates that the presence of thrombophilic genetic mutations is associated with a more severe disease pattern, delayed recovery of visual acuity, and an increased frequency of complications. These findings highlight the significance of incorporating molecular genetic analysis into the diagnostic evaluation of patients with retinal vascular disorders. Early identification of thrombophilic predisposition allows clinicians to develop more individualized management strategies that address both ocular manifestations and systemic thrombotic risk. Integration of genetic diagnostics with conventional ophthalmic examination may improve prognostic accuracy and contribute to more effective prevention of visual impairment associated with retinal vein occlusion.

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