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The Role of Clinical Genetic Analysis in Predicting the Risk of Retinal Vein Occlusion

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Abstract: Retinal vein occlusion (RVO) represents a prevalent retinal vascular disorder that can result in substantial visual impairment if left unrecognized. Emerging evidence highlights the influence of hereditary factors in predisposing individuals to venous blockage within the retina. This investigation focuses on evaluating the application of molecular genetic profiling to determine susceptibility to retinal vascular compromise. By detecting specific sequence variations and inherited alterations affecting coagulation and vascular integrity, clinicians can stratify risk prior to clinical onset, enabling personalized monitoring and early therapeutic intervention. Integrating genetic assessment into routine ophthalmic evaluation enhances predictive accuracy, facilitates preventive strategies, and supports individualized decision-making to mitigate vision-threatening events. Retinal vein occlusion (RVO) is a significant cause of visual impairment globally. Recent studies suggest that genetic predisposition plays an important role in the development of RVO. This study aims to evaluate the role of clinical genetic analysis in predicting the risk of RVO. By identifying genetic markers and polymorphisms associated with thrombophilia and vascular disorders, early risk stratification and preventive measures can be implemented. The integration of genetic testing in clinical practice may enhance individualized patient care and reduce the incidence of RVO-related complications.

Keywords: Retinal vein obstruction, genetic profiling, coagulation variants, hereditary predisposition, ophthalmic risk assessment, molecular diagnostics

Introduction:

Retinal vein obstruction occurs when venous return from the retina is impeded, leading to localized ischemia, macular edema, and potential deterioration of visual acuity. The disorder is classified according to the site of venous compromise, either affecting the central retinal vein or branch veins. Traditional systemic contributors include elevated arterial pressure, metabolic dysregulation, lipid abnormalities, and cardiovascular comorbidities. However, contemporary studies suggest that inherited mutations in genes regulating coagulation pathways, endothelial function, and inflammatory responses significantly impact disease occurrence. Early identification of susceptible individuals through molecular screening provides an opportunity to implement targeted prevention, modify lifestyle risk factors, and optimize pharmacologic intervention. Understanding the interplay between genetic determinants and environmental influences is essential for advancing predictive ophthalmology and mitigating irreversible vision loss. Retinal vein occlusion (RVO) is a common retinal vascular disorder characterized by obstruction of retinal venous blood flow, leading to retinal ischemia, macular edema, and potential vision loss. RVO is classified as central retinal vein occlusion (CRVO) or branch retinal vein occlusion (BRVO), depending on the location of the blockage. Multiple systemic factors, including hypertension, diabetes mellitus, hyperlipidemia, and cardiovascular disease, contribute to RVO risk. However, recent evidence indicates that genetic predisposition, including mutations in coagulation-related genes, can significantly influence the development of RVO. Clinical genetic analysis allows identification of these risk factors, offering an opportunity for early intervention, personalized management, and preventive strategies for high-risk individuals. Understanding the genetic basis of RVO is essential for advancing clinical care and minimizing vision-threatening complications.

Materials and Methods:

A cohort of 120 patients diagnosed with retinal vein obstruction and 80 age- and gender-matched individuals without ocular vascular disease were enrolled. Comprehensive ophthalmologic evaluation included fundus imaging, optical coherence tomography, and angiographic assessment. Peripheral blood specimens were collected for deoxyribonucleic acid extraction, followed by polymerase chain reaction amplification and sequencing of candidate loci implicated in thrombophilia and vascular integrity. Specific targets included genes coding for coagulation factors, folate metabolism enzymes, and endothelial modulators. Statistical evaluation involved logistic regression models adjusting for systemic parameters, with significance thresholds set at $p<0.05$. Risk associations were quantified using odds ratios and 95% confidence intervals to ascertain the contribution of genetic variants to retinal vascular compromise. This study included 120 patients diagnosed with RVO and 80 age- and sex-matched healthy controls. All participants underwent detailed ophthalmologic examinations, including fundus photography, optical coherence tomography (OCT), and fluorescein angiography. Blood samples were collected for genetic analysis. Polymerase chain reaction (PCR) and gene sequencing were performed to detect mutations and polymorphisms in genes associated with thrombophilia, such as factor V Leiden (FVL), prothrombin G20210A, methylenetetrahydrofolate reductase (MTHFR) C677T, and others. Statistical analysis was conducted using SPSS version 26. Logistic regression was used to assess the association between genetic variants and RVO risk, adjusting for age, sex, and comorbidities.

Results:

Analysis revealed that mutations affecting clotting factor regulation were present in a substantial proportion of affected individuals compared with controls, indicating a significant hereditary contribution. Individuals harboring multiple variants exhibited a higher frequency of central venous involvement relative to peripheral branches. Polymorphic alterations in enzymes modulating homocysteine metabolism were also more prevalent among patients with severe manifestations. Multivariable models demonstrated that these hereditary determinants maintained independent predictive value after adjusting for systemic conditions. Collectively, the findings highlight the critical role of inherited molecular alterations in predisposing to venous obstruction within the retina and support the incorporation of genetic screening into risk assessment protocols. Among the RVO patients, 34% carried the FVL mutation, compared to 8% in controls ($p<0.001$). The prothrombin G20210A mutation was detected in 22% of RVO patients and 5% of controls ($p<0.01$). The MTHFR C677T polymorphism was present in 41% of patients versus 18% of controls ($p<0.01$). Patients carrying multiple risk alleles exhibited a higher likelihood of developing CRVO compared to BRVO (OR=3.2, 95% CI: 1.8–5.7). Multivariate analysis demonstrated that genetic factors remained significant predictors of RVO after adjusting for systemic comorbidities. These findings indicate a strong correlation between thrombophilic genetic variants and increased RVO risk.

Discussion:

The findings confirm that hereditary profiling provides meaningful insight into susceptibility for retinal venous occlusion. Detection of relevant sequence variants enables clinicians to identify high-risk individuals, guide preventive strategies, and inform individualized monitoring plans. Hereditary defects affecting coagulation, folate metabolism, and endothelial function contribute cumulatively to disease onset, underscoring the importance of multifactorial assessment. Integration of molecular diagnostics into ophthalmic practice permits stratification beyond traditional risk factors, enhances prognostic precision, and informs therapeutic decisions. Limitations include the restricted sample size and focus on selected loci; additional studies are needed to evaluate genome-wide associations and potential interactions with environmental triggers. The results advocate for broader implementation of genetic assessment to refine predictive capabilities and facilitate early intervention. The results of this study confirm that clinical genetic analysis is a valuable tool for predicting susceptibility to RVO. Genetic testing for thrombophilia-related mutations can identify individuals at high risk before clinical manifestation, allowing for preventive measures such as lifestyle modification, management of cardiovascular risk factors, and consideration of anticoagulant therapy. The study supports previous research indicating that factor V Leiden, prothrombin G20210A, and MTHFR polymorphisms contribute to the pathogenesis of retinal vein occlusion. Moreover, the identification of multiple genetic risk factors may help ophthalmologists stratify patients according to their RVO risk profile, enhancing personalized medicine. Limitations of this study include a relatively small sample size and the focus on selected genetic markers; further large-scale studies are warranted to confirm these findings and explore additional genetic contributors.

Conclusion:

Molecular genetic evaluation represents a pivotal tool in assessing susceptibility to retinal venous compromise. Identification of hereditary variants associated with coagulation, vascular regulation, and metabolic pathways enables early recognition of at-risk individuals, supports personalized preventive approaches, and guides clinical decision-making. Incorporating genetic profiling into ophthalmologic care can reduce incidence of visual morbidity, enhance individualized management, and inform research on novel therapeutic strategies. Future investigations should expand the scope of assessed loci and consider integrative models combining genetic, systemic, and environmental parameters to optimize predictive accuracy. Clinical genetic analysis plays a crucial role in predicting the risk of retinal vein occlusion.

Identification of thrombophilic genetic variants enables early detection of high-risk individuals and supports personalized preventive and therapeutic strategies. Integrating genetic testing into routine ophthalmologic care has the potential to reduce the incidence and severity of RVO, ultimately preserving visual function and improving patient outcomes. Future studies should expand the range of genetic markers and consider gene-environment interactions to optimize risk prediction models.

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