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Clinical and Genetic Factors of Thrombogenic Risk in Patients with Retinal Vein Occlusion

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Annotation: Retinal vein occlusion (RVO) is a significant cause of visual impairment worldwide. Its pathogenesis is multifactorial, involving both clinical and genetic risk factors. This study aimed to evaluate the association of thrombogenic genetic markers and clinical characteristics with the risk of RVO. A total of 120 patients with RVO and 100 age- and sex-matched controls were examined. Genetic testing focused on Factor V Leiden, prothrombin G20210A mutation, and MTHFR polymorphisms. Clinical evaluation included systemic conditions, laboratory parameters, and ophthalmologic findings. Results indicated that patients with RVO exhibited higher prevalence of hypertension, diabetes mellitus, and hyperlipidemia. Factor V Leiden mutation was significantly more frequent in the RVO group, while prothrombin and MTHFR polymorphisms also contributed to increased thrombogenic risk. These findings highlight the importance of combined clinical and genetic assessment in identifying patients at high risk of RVO and guiding preventive strategies. Retinal vein obstruction is a common retinal vascular

disorder that can lead to severe visual impairment. Its occurrence is influenced by multiple determinants, including systemic health conditions and inherited prothrombotic variants. This investigation aimed to evaluate the relationship between hereditary coagulation anomalies and systemic features with the incidence of venous blockage in the retina. A total of 120 affected individuals and 100 matched healthy volunteers were analyzed. Laboratory procedures focused on detecting Factor V Leiden, prothrombin G20210A, and MTHFR gene variants. Clinical evaluation encompassed metabolic, cardiovascular, and ocular assessments. Findings revealed a substantial association of hypertension, impaired glucose tolerance, and dyslipidemia with retinal venous obstruction. Genetic analysis demonstrated an increased frequency of Factor V Leiden among patients, with prothrombin and MTHFR polymorphisms also contributing to heightened thrombotic susceptibility. These outcomes underline the significance of comprehensive clinical-genetic profiling for early recognition of at-risk subjects and the implementation of preventive measures.

Keywords: Retinal vein occlusion, thrombogenic risk, Factor V Leiden, prothrombin mutation, MTHFR, clinical risk factors, genetic predisposition.

Introduction:

Retinal vein occlusion is the second most common retinal vascular disorder after diabetic retinopathy and is a leading cause of vision loss, particularly in elderly patients. RVO can be classified as central retinal vein occlusion (CRVO) or branch retinal vein occlusion (BRVO), depending on the site of venous obstruction. The pathophysiology of RVO involves a combination of local vascular factors and systemic thrombogenic conditions. Classical risk factors include

hypertension, diabetes mellitus, dyslipidemia, and glaucoma. Additionally, genetic predisposition plays a crucial role in determining individual susceptibility to venous thrombosis. Polymorphisms in the Factor V, prothrombin, and MTHFR genes have been associated with hypercoagulability and may contribute to RVO development. Understanding the interplay between clinical and genetic risk factors is essential for early diagnosis, risk stratification, and prevention of complications associated with RVO. Retinal venous occlusion represents a substantial cause of sudden or gradual vision loss and is frequently observed in aging populations. Blockages can occur in central or branch retinal veins, resulting in retinal ischemia, hemorrhage, and macular edema. Multiple physiological and pathological mechanisms are implicated, including vascular endothelial dysfunction, impaired blood flow, and systemic prothrombotic conditions. Chronic illnesses such as elevated blood pressure, diabetes, lipid disorders, and ocular hypertension are recognized contributors. Additionally, inherited coagulation abnormalities predispose certain individuals to venous obstruction. Mutations in genes coding for coagulation proteins, such as Factor V, prothrombin, and methylenetetrahydrofolate reductase, can enhance thrombotic tendencies. Identifying the interplay between systemic disturbances and genetic predisposition is essential to develop strategies for timely intervention and the prevention of vision-threatening complications.

Materials and Methods:

This observational case-control study included 120 patients diagnosed with RVO at the Department of Ophthalmology, Samarkand State Medical University, from January 2023 to June 2025. The control group comprised 100 healthy individuals matched by age and sex. All participants underwent a detailed ophthalmologic examination, including fundus photography, optical coherence tomography (OCT), and fluorescein angiography. Clinical evaluation recorded medical history, blood pressure, fasting glucose, lipid profile, and body mass index. Genetic analysis involved polymerase chain reaction (PCR) testing for Factor V Leiden (G1691A), prothrombin (G20210A), and MTHFR (C677T and A1298C) mutations. Statistical analysis included chi-square tests for categorical variables and logistic regression to assess independent risk factors for RVO. The study included 120 participants diagnosed with retinal vein blockage and 100 healthy controls matched by age and sex. Detailed ophthalmologic examinations were performed, including fundus photography, optical coherence tomography, and fluorescein angiography. Clinical parameters evaluated included arterial pressure, glycemic status, serum lipid levels, and body mass index. Genetic screening utilized polymerase chain reaction techniques to detect Factor V Leiden (G1691A), prothrombin (G20210A), and MTHFR (C677T, A1298C) mutations. Statistical analyses employed chi-square assessments and multivariable logistic regression to determine independent associations between clinical and genetic variables and retinal venous obstruction risk.

Results:

Among RVO patients, 65% had hypertension, 40% had diabetes mellitus, and 35% had hyperlipidemia. Factor V Leiden mutation was detected in 18% of patients compared to 4% of controls ($p<0.01$). Prothrombin G20210A mutation was present in 10% of RVO patients and 3% of controls ($p<0.05$). MTHFR C677T polymorphism was found in 22% of patients and 12% of controls, while A1298C was observed in 15% versus 7% in controls. Multivariate logistic regression identified hypertension (OR 2.5), diabetes (OR 1.8), hyperlipidemia (OR 1.6), and Factor V Leiden mutation (OR 3.2) as independent predictors of RVO. Patients with combined clinical and genetic risk factors demonstrated the highest probability of developing RVO. Hypertension was present in 65% of affected individuals, impaired glucose regulation in 40%, and abnormal lipid profile in 35%. Factor V Leiden mutation was identified in 18% of patients versus 4% of controls, while prothrombin G20210A appeared in 10% versus 3%, and MTHFR variants C677T and A1298C were found in 22% and 15% of patients compared to 12% and 7% in the control group. Regression analysis demonstrated that elevated blood pressure, dysglycemia, lipid irregularities, and Factor V Leiden carriage were independent predictors of retinal vein

obstruction. Patients carrying multiple risk factors exhibited the highest susceptibility to venous blockage events.

Discussion:

Our findings indicate that both clinical and genetic factors significantly contribute to the development of RVO. Hypertension, diabetes mellitus, and hyperlipidemia remain dominant clinical risk factors, consistent with previous studies. The presence of thrombophilic mutations, particularly Factor V Leiden, substantially increases the risk of venous occlusion, supporting the role of inherited hypercoagulability in RVO pathogenesis. MTHFR and prothrombin mutations also exhibit contributory effects, albeit to a lesser extent. These results emphasize the need for comprehensive risk assessment incorporating both systemic health and genetic predisposition. Early identification of high-risk individuals allows for targeted preventive strategies, including anticoagulant therapy, lifestyle modification, and careful management of comorbidities. Further longitudinal studies are necessary to evaluate the predictive value of combined clinical and genetic risk assessment in RVO recurrence and progression. The investigation confirms the combined influence of systemic and inherited determinants in retinal venous occlusion. Cardiovascular and metabolic disorders significantly elevate thrombotic potential within retinal veins. Inherited coagulation anomalies, particularly Factor V Leiden, markedly enhance susceptibility. Prothrombin and MTHFR polymorphisms further modulate risk, though to a lesser extent. These findings emphasize the necessity for dual assessment encompassing clinical and genetic evaluation to identify high-risk individuals. Preventive interventions, such as anticoagulation management, lifestyle modification, and stringent control of metabolic disorders, can mitigate the likelihood of occlusive events. Future longitudinal studies are warranted to determine the prognostic value of integrated clinical-genetic risk profiling for recurrence and progression of retinal vein obstruction.

Conclusion:

Retinal vein occlusion is a multifactorial disease influenced by systemic conditions and genetic thrombophilia. The study demonstrates that hypertension, diabetes, hyperlipidemia, and Factor V Leiden mutation are the most significant predictors of RVO. Integrating clinical and genetic evaluation can improve early detection, prevention, and individualized patient management, ultimately reducing the risk of vision loss. Retinal venous blockage is a multifactorial condition influenced by systemic comorbidities and hereditary thrombophilia. Hypertension, impaired glucose metabolism, lipid abnormalities, and Factor V Leiden mutation are principal risk determinants. Comprehensive assessment combining clinical evaluation and genetic testing enhances early detection, risk stratification, and personalized preventive strategies, potentially preserving visual function and reducing complications.

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