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ANALYSIS OF IL6 GENE POLYMORPHISM (rs1800795) IN HEMORRHAGIC VASCULITIS

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Abstract

Objective. To investigate allelic polymorphism of the IL6 gene (rs1800795) in patients with immune microthrombotic vasculitis (hemorrhagic vasculitis).

Materials and Methods. The study included 75 patients with hemorrhagic vasculitis (age range: 16–80 years) and 73 conditionally healthy controls without hemostatic system pathology, matched by age. Detection of the IL6 polymorphism (rs1800795) was performed using SNP-PCR.

Results and Conclusions. Carriage of the IL6 gene polymorphism (rs1800795) was not associated with increased risk for hemorrhagic vasculitis.

Keywords: IL6 gene polymorphism (rs1800795), hemorrhagic vasculitis, allele, genotype, association, risk.

INTRODUCTION

Among human diseases, disorders of hemostasis (bleeding diatheses) increasingly attract researchers' attention due to their high prevalence, diverse clinical manifestations, and frequent development of severe hemorrhagic and thrombohemorrhagic complications that may lead to death [3,4]. Within the overall spectrum of hemostatic disorders across populations and age groups, acquired primary hemostasis pathology—specifically immune microthrombotic

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vasculitis (hemorrhagic vasculitis, HV)—occupies an important place with regard to polymorphism and clinical severity.

Various endogenous and exogenous factors negatively affect vessel wall integrity, which subsequently leads to disturbances in the hemostatic system. The nature and clinical manifestations of these disturbances in HV are characterized by complex formation mechanisms that remain insufficiently understood and are the subject of ongoing research [1,2].

A review of the literature shows considerable progress in studying different aspects of HV formation. However, despite more than two centuries of investigation, many causal factors and pathogenic mechanisms remain incompletely elucidated [10]. This incomplete understanding and contradictory findings in the literature contribute to the high rates of complications and recurrences seen in HV [5].

Although the molecular bases underlying HV development are not fully known, evidence suggests that genetic factors play a decisive role in the disease pathogenesis [8]. Cytokine genes are important regulators of immune and inflammatory responses; polymorphic variants in these genes can alter inflammatory and immune regulation [6]. Among cytokines, interleukin-6 (IL-6) is a major proinflammatory mediator with a wide spectrum of biological effects. Single nucleotide substitutions (SNPs) in regulatory regions of the IL6 gene often change its biological function, leading to increased or decreased IL-6 production depending on the substitution site [7].

Studies examining the association between IL-6 and HV have yielded mixed results [8,9]. Given the absence of a consensus, investigating IL6 gene polymorphism (rs1800795) in HV is a relevant and interesting research question.

Study Objective

To study features of allelic polymorphism of the IL6 gene (rs1800795) in adult patients with immune microthrombotic vasculitis in Uzbekistan.

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MATERIALS AND METHODS

Seventy-five patients with HV (aged 16 to 80 years) who were followed at the outpatient clinic of RSNPMCG of the Ministry of Health of the Republic of Uzbekistan between 2017 and 2018 were included in the study, along with 73 conditionally healthy controls without hemostatic pathology matched by age. All participants were divided into two groups: the main group (n = 75) consisting of patients with HV, and the control group (n = 73) of healthy individuals. The main group was further divided by disease stage into two subgroups: subgroup A — patients with active HV (n = 41), and subgroup B — patients in remission (n = 34). The diagnosis of HV was verified according to the contemporary classification criteria EULAR/PRINTO/PReS (2010).

Detection of IL6 polymorphism (rs1800795) was performed using SNP-PCR on an Applied Biosystems 2720 programmable thermal cycler (USA) with test systems from Litekh (Russia), following the manufacturers' instructions. Statistical analysis was carried out using the OpenEpi statistical package, Version 9.3.

RESULTS AND DISCUSSION

We analyzed the distribution of allele and genotype frequencies of the IL6 polymorphism (rs1800795) in the control group and in patients with HV.

Comparative analysis of allele and genotype frequencies between the main and control groups showed no statistically significant differences: allele C frequency was 85.3% in patients and 80.1% in controls, allele G frequency 14.7% and 19.9%, respectively. Genotype frequencies C/C were 70.7% in patients and 61.6% in controls; C/G 29.3% and 37.0%, respectively ($\chi^2 = 1.401$; $p = 0.2372$; OR = 0.6934; 95% CI: 0.3775–1.274). The homozygous mutant genotype G/G, which is considered the most unfavorable with respect to increased IL-6 production, was observed in 1.4% (n = 1) of individuals only in the control group ($\chi^2 = 2.137$; df = 2, $p = 0.3436$) (Table 1).

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Table 1. Differences in allele and genotype frequencies of IL6 (rs1800795) polymorphism in controls and HV patients

Polymorphism		Alleles/Genotypes	Control group (n=73)		Patient group, (n=75)		Statistical significance
			n	%	n	%	
C174G (IL6)	Alleles	C	117	80,1	128	85,3	χ^2= 1.401; p= 0.2372; OR= 0.6934; 95% CI: 0.3775-1.274
		G	29	19,9	22	14,7	
	Genotype	C/C	45	61,6	53	70,7	χ^2= 2.137; df=2, p= 0.3436
		C/G	27	37	22	29,3	
		G/G	1	1,4	0	0	

No significant difference in carriage rates of alleles C and G compared with the control group was found when analyzed by disease stage. Among patients with active HV (subgroup A) the allele frequencies were C = 85.4% and G = 14.6% (χ^2 = 0.9734; p = 0.3238; OR = 0.6916; 95% CI: 0.3316–1.442). In patients in remission (subgroup B) the frequencies were C = 85.3% and G = 14.7% (χ^2 = 0.8279; p = 0.3629; OR = 0.6956; 95% CI: 0.3174–1.524). Consequently, comparative analysis of genotype frequencies C/C, C/G and G/G between both patient subgroups and the control group revealed no statistically significant association between IL6 (rs1800795) polymorphism and HV risk. In subgroup A the frequencies of genotypes C/C and C/G were 70.7% and 29.3% (χ^2 = 1.353; df = 2; p = 0.5084), and in subgroup B — 70.6% and 29.4% (χ^2 = 1.138; df = 2; p = 0.5660).

Observed genotype frequencies of IL6 (rs1800795) in both the main and control groups corresponded to Hardy–Weinberg equilibrium. Specifically, observed C/C genotype frequencies were 70.7% and 61.6% (expected 72.82% and 64.22%); observed C/G frequencies were 29.3% and 32.0% (expected 25.03% and 31.84%). The differences between observed and expected genotype frequencies were not statistically significant (χ^2 = 2.22; p = 0.1366 and χ^2 = 1.91; p = 0.4, respectively).

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Thus, our study found no significant differences in allele and genotype distributions of IL6 (rs1800795), which supports the absence of an association between this genetic polymorphism and the risk of developing hemorrhagic vasculitis. These results are consistent with findings from international authors R. López-Mejías, B. Sevilla Pérez, F. Genre et al. (2013), who also reported no significant differences in IL6 polymorphism frequencies (rs2069827, rs1800795 and rs2069840) between HV patients and healthy controls [7].

CONCLUSION

No association was found between the IL-6 (rs1800795) gene polymorphism and the development or severity of immune microthrombotic vasculitis.

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