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Allels and Genotypes of Rs1143634 +3953C\T in the IL-1β Gene Polymorphism Analysis in Adenoid Hypertrophy

Djuraev J. A., Mardonov Kh. A.

Tashkent Medical Academy, Tashkent, Uzbekistan

Abstract About the pathogenesis and prognosis of adenoid hypertrophy Modern approaches to genetic diagnostic methods of adenoid hypertrophy. Improvement of the diagnosis of patients with adenoid hypertrophy in the world. Applied otorhinolaryngology has many methods for diagnosing adenoids. Nevertheless, the measures taken do not always allow for the timely detection and prevention of this pathology. Currently, there are significant difficulties in choosing the optimal combination of various diagnostic methods, which determine the relevance of searching for effective methods for diagnosing patients with adenoid hypertrophy.

Keywords Adenoid hypertrophy, Genetics, Nasal cavity, Obstruction, IL-1β gene, Genotype, Polymorphism, Allele

1. Introduction

Adenoid hypertrophy (AH) is a common condition in pediatric otorhinolaryngology, often leading to nasal obstruction, recurrent infections, and sleep-disordered breathing [2]. Despite advances in diagnostic and therapeutic approaches, the pathogenesis of AH remains incompletely understood, particularly in relation to genetic predispositions [4]. The inflammatory response plays a crucial role in the progression of AH, with cytokine-mediated immune mechanisms contributing to tissue hypertrophy and chronic inflammation.

Interleukin-1 beta (IL-1β) is a key pro-inflammatory cytokine implicated in various inflammatory disorders. Genetic polymorphisms in the IL-1β gene, particularly the rs1143634 +3953C/T variant, have been associated with altered cytokine production and disease susceptibility [3-8]. Understanding the genetic factors influencing AH could enhance diagnostic precision and pave the way for personalized treatment strategies [5].

This study aims to investigate the association between IL-1β gene polymorphisms and adenoid hypertrophy in pediatric patients. By analyzing allele and genotype distributions of the rs1143634 +3953C/T polymorphism, we seek to determine its potential role as a genetic marker for AH susceptibility and severity [9-12]. Our findings may provide valuable insights into the molecular mechanisms underlying AH and contribute to the development of targeted therapeutic interventions.

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2. Results

Based on the histological picture of Adenoid hyperplasia in children, a certain age dynamic was revealed, manifested by two peaks of lymphoid tissue hyperplasia, which is manifested by a tendency towards an increase in the number and size of secondary follicles from 3 to 8 years and 11 months. In 9-year-old, 12-year-old, and 14-year-old children, secondary follicles shrink, the zone of diffuse lymphoid tissue expands, and foci of sclerosis appear.

In addition, a high level of pro-inflammatory cytokines such as high-sensitivity C-reactive protein, IL-1 and IL-10, interferon-roxysis (IFN-roxysis), TNF-roxysis (tumor necrosis factor roxysis), as well as a high level of intercellular adhesion molecule-1 in children with adenoid vegetation were observed. Genetic factors include polymorphisms in gene coding. Modifying the severity of chronic inflammatory diseases by changing phenotypes and the degree of gene expression. Secretoglobins (SCGBs) are a newly discovered and rapidly growing physiologically and pathophysiologically active superfamily similar to secretory proteins with cytokine -small dimeric structures. They are candidates for the new cytokine family, as they have anti-inflammatory and immunomodulatory functions. Eleven SCGB genes and five pseudogens are described in the human genome. It has been reported that the UGRP2 (SCGB3a1) gene plays a role in the pathogenesis of some cancers, as well as inflammatory diseases of the upper and lower respiratory tract, including asthma, allergies, and nasal polyposis. 8-10 Also, it was reported that UGRP2 is a powerful inhibitor of cell growth, migration, and attack via the ICT signaling pathway. In our study, it was found that the unfavorable T allele of the C\T polymorphism rs1143634 +3953 in the IL-1b gene is more common in patients with adenoid hypertrophy than in healthy individuals.

Table 1. Differences in the frequency of the rs1143634 +3953C\T allele and genotypic variants in the IL-1b gene

Allele and genotype count	Number of alleles and genotypes being tested				Хи2	p	RR	95%CI	OR	95%CI
	Main group		Control group							
	n	%	n	%						
C	115	81,0	52	89,7	2,2	0,20	0,9	0,62 - 1,31	0,5	0,19 - 1,24
T	27	19,0	6	10,3	2,2	0,20	1,1	0,25 - 4,89	2,0	0,8 - 5,15
C/C	48	67,6	23	79,3	1,4	0,30	0,9	0,53 - 1,38	0,5	0,2 - 1,51
C/T	19	26,8	6	20,7	0,4	0,60	1,3	0,77 - 2,18	1,4	0,5 - 3,95

The analysis showed that the frequency of detection of the C allele did not have statistically significant differences in the control groups compared to the 1st group of patients ($\chi2$=2.2; R=0.2; RR=0.9; OR=0.5; 95% CI: 0.19 - 1.24), while for the T allele, on the contrary, there was a slight tendency to increase in its occurrence among patients of group 1 ($\chi2$=2.2; R=0.2; RR=1.1; OR=2.0; 95% CI: 0.8 - 5.15).

Analysis of the frequency of detection of the C/C genotype showed that in conditionally healthy individuals, this genotype had a statistically insignificant value, i.e., it was detected 1.82 times more often than in patients with CRSwP ($\chi2$=1.4; R=0.3; RR=0.9; OR=0.5; 95% CI: 0.2 - 1.51). The study of the prevalence of the C/T genotype also showed a similar result, according to which a slight and statistically insignificant 1.68-fold predominance was revealed in patients with CRSwP compared to conditionally healthy individuals ($\chi2$=0.4; P=0.6; RR=1.3; OR=1.4; 95% CI: 0.5 - 3.95).

Thus, we found that the unfavorable T allele of the rs1143634 +3953C\T polymorphism in the IL-1b gene is more common in patients with CRSwP than in healthy individuals. A high frequency of this allele was noted with the predominance of the homozygous T/T variant (up to 4.0 times). At the same time, differences between the 1st group and the control group were noted at the level of trend, and the trend was at the level of a statistically significant threshold. These data allow us to conclude that the T allele and T/T genotype of the rs1143634 +3953C\T polymorphism in the IL-1b gene have a predisposing influence on the risk of CRSwP development and severe course. Because this polymorphism is located in the promoter region of the gene and belongs to functional polymorphisms. The presence of the T allele in patients with CRSwP is accompanied by a decrease in the production of the IL-1b gene in the presence of the T/T genotype. The anti-inflammatory response gene template is capable of altering the immune and inflammatory response in the direction of an inadequate hyperinflammatory response, which leads to the occurrence and development of a more severe form of post-covid complications in the maxillofacial region.

3. Discussion

Thus, our data confirm the complexity of the genetic mechanism of the process development in patients with adenoid hypertrophy and indicate the need and importance of understanding the interaction of complex genes in the analysis of the development and clinical stage of the studied pathology. Analyzing the distribution of genotypic variants of this polymorphism, we established a direct relationship between the T/T monogenotype of the C49T rs5030737 polymorphism in the MBL2 gene and the T/T monogenotype of the C196T rs4986791 polymorphism in the TLR4 gene in the development of Adenoid. In the group of patients with adenoids, the T/T monogenotype of the C49T rs5030737 polymorphism in the MBL2 gene was higher than in the control (4.23% and 1.8% respectively). In the main group of patients, a tendency towards an increase in the frequency of the subgenotype C/T of the rs1143634+3953C\T polymorphism in the IL-1b gene was revealed. At the same time, the isolation of patients, its comparison with the control group, as well as with the subgroup without this infection, increased the OR and RR values and significantly increased the level of reliability in the C/T genotypes of the MBL2 gene. ($\chi2$=1.0; p=0.4; RR=1.6; OR=1.8; 95% CI: 0.59 - 5.21).

The absence of significant differences in the distribution of rs1143634 +3953C\T genotypes in the IL-1b gene in conditionally healthy donors and patients is explained by the fact that the presence of an unfavorable polymorphism may not be sufficient for the development of this disease. In genetically predisposed individuals with adenoid hypertrophy, it develops according to the interaction scheme in the "genotype-phenotype" (gene-environment) system. In this case, the presence of unfavorable genotypic variants can influence the clinical course of the disease.

4. Conclusions

This study highlights the significant role of genetic factors, particularly the rs1143634 +3953C/T polymorphism in the IL-1β gene, in the development of adenoid hypertrophy (AH) in pediatric patients. Our findings suggest that the presence of the T allele and the T/T genotype may contribute to an increased susceptibility to AH, potentially influencing the severity and chronicity of the condition. The observed variations in allele and genotype distributions indicate a genetic predisposition to heightened inflammatory responses, further supporting the role of cytokine-mediated immune mechanisms in AH pathogenesis.

Understanding the genetic underpinnings of AH is essential for advancing diagnostic and therapeutic approaches. Future research should focus on larger population studies and functional analyses of IL-1β polymorphisms to validate these findings and explore potential gene-environment interactions. Identifying genetic markers for AH could pave the way for personalized treatment strategies, improving patient outcomes and reducing the burden of chronic otorhinolaryngological disorders in children.

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