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Features Of The Mdr1 (C3435t) Polymorphism In Non-Syndromic Congenital Orofacial Clefts

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Abstract

The present study investigated the MDR1 (C3435T) polymorphism in patients with non-syndromic congenital orofacial clefts in Uzbekistan. Comparative analysis of allele and genotype frequencies demonstrated no statistically significant differences between affected patients and healthy controls. Based on the obtained findings, the MDR1 (C3435T) polymorphism cannot be considered an independent genetic marker associated with an increased risk of non-syndromic congenital orofacial clefts in the Uzbek population.

Keywords: orofacial clefts, MDR1, C3435T polymorphism, cleft palate, cleft lip, genetics, allele frequency, genotype distribution

Introduction

Non-syndromic congenital orofacial clefts (NSCOCs) represent a complex group of multifactorial developmental disorders that include isolated cleft palate (Q35), isolated cleft lip (Q36), and cleft lip with palate (Q37). The MDR1 gene encodes P-glycoprotein, a transmembrane transporter involved in xenobiotic detoxification. The C3435T polymorphism may affect transporter activity and has been proposed as a candidate factor in congenital anomalies.

Materials and Methods

The study included 105 children with NSCOCs and 103 healthy controls. Patients were divided into Q35 (n=35), Q36 (n=33), and Q37 (n=37) groups.

DNA was isolated from peripheral blood leukocytes and MDR1 C3435T genotyping was performed using Rotor-Gene Q (Qiagen, Germany). Statistical analysis was performed using OpenEpi 9.2.

Results

Genotype distributions corresponded to Hardy–Weinberg equilibrium. Frequencies of C and T alleles in patients were 73.8% and 26.2%, respectively, compared with 72.3% and 27.7% in controls. No statistically significant differences were found in allele or genotype frequencies between patients and controls or between clinical subgroups. Although the T/T genotype tended to occur more frequently in some patient groups, none of the associations reached statistical significance.

Discussion

The findings indicate that MDR1 C3435T is unlikely to be a major genetic determinant of susceptibility to non-syndromic orofacial clefts in Uzbekistan. Minor variations observed among subgroups were not supported by statistical significance.

Conclusion

No statistically significant association was identified between the MDR1 (C3435T) polymorphism and non-syndromic congenital orofacial clefts. Therefore, this polymorphism cannot be considered an independent genetic predictor of disease susceptibility in the studied population.

References

15 references from the original manuscript retained and translated into international citation style.