

## **Gene study of head and neck carcinoma**

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**Abstract:** the research was performed on 117 patients of Uzbek ethnicity with malignant tumors of maxillo-facial area. Results of the research showed that developing of malignant tumors of maxillo-facial area involving face skin and oral mucosa was associated with the carrier of a heterozygous Arg/Pro genotype.

**Keywords:** maxillo-facial area, neoplasm, genetic aberration, genotype, polymorphic marker, carcinoma, Uzbek ethnicity, amplification, heterozygous.

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Neoplasms of maxillo-facial area involving face skin and oral mucosa according to numerous studies have a growing trend. Despite the visual accessibility of the localization over 70% of patients with malignant neoplasms are diagnosed in last clinical stages.

As well known, the gene TP53 plays an important role in regulation of cell cycle and maintenance of genome stability, preventing mutations. The decrease of the gene TP53 expression or its mutation is probably one of the most prevailing genetic aberrations leading to cancer. The most frequent polymorphism of the gene TP53 in the codon 72 of the exon 4 (dbSNP ID: rs1042522)

designated as Arg72Pro may influence on the development risk and/or prognosis of many cancer localizations, as it is reported 1, 2, 3, 4.

**The purpose of the study** is to investigate the distribution features of genotypes of polymorphic marker Arg72Pro of the gene TP53 in patients with carcinoma of Uzbek ethnicity.

**Materials and methods of research.** The main group (n=117) consisted of patients of Uzbek ethnicity with carcinoma, among them there were 90 male and 27 female patients. The average age is 54.3+8.6 years old. The control group (n=70) consisted of volunteers of Uzbek ethnicity with average age 51.0+6.2 years

old with no oncologic diseases reported.

Carcinomas were diagnosed on the base of cytomorphological researches, inconformity between the clinical symptoms and the morphological results were not observed.

The isolation of genomic DNA was performed from whole peripheral blood with the device Diatom DNA Prep 200 ("Isogen Laboratory", Russia) according to the producer protocol. The polymerase chain reaction was performed on amplifier GeneAmp® PCR System 9700 (Applied Biosystems, USA) using set for genotyping on polymorphic marker Arg72Pro of the gene TP53 (GenTest, Russia).

**The conditions of amplification:**

The first denaturation	10 cycles	30 cycles
95°C – 5 min	94°C – 20 sec	94°C – 20 sec
	TD* – 50 sec	64°C – 50 sec
	72°C – 20 sec	72°C – 20 sec

**\* - TD - Touchdown PCR**

Detection of the amplification results was carried out by horizontal electrophoresis in a 1% agarose gel at field strength of ~ 15V / cm for 30 minutes. The gel was stained with a solution of ethidium bromide followed by viewing in the ultraviolet spectrum (wave length 254 nm) on a transilluminator. The obtained results were documented by photographing.

To determine the genotype, subsequent restriction of the amplification products was performed using the restriction enzyme HspAI at 50 ° C for 2 hours.

Thus, by gel electrophoresis, a 252 bp fragment was found that matched the amplified DNA fragments containing the polymorphic sequences of Arg72Pro of the TP53 gene.

To determine the genotype, restriction of amplification products was performed

using the restriction enzyme TP53-Arg72Pro. The DNA fragment containing the Arg/Arg genotype was 252 bp long. The fragment containing the Pro/Pro genotype was separated by a restriction enzyme into fragments of size 228 and 24 bp. Fragments of 252, 228, and 24 bp in size corresponded to the heterozygous genotype Arg/Pro.

The distribution of genotypes in both groups corresponded to the Hardy-Weinberg law. For comparison of genotypes, the standard Pearson  $\chi^2$  test was used. The relative risk (odds ratio, OR) of the disease in the carrier of a particular genotype was calculated according to the multiplicative inheritance model.

**Results of the study.** The frequency of distribution of the Arg72Pro genotypes polymorphism of the TP53 gene in patients with carcinoma of Uzbek ethnicity of Arg/Arg was 0,11, Arg/Pro was 0,68, Pro/Pro was 0,21;  $\chi^2= 99,308$ ,  $p = 0,000$ . In healthy volunteers, the Arg/Arg:Arg/Pro:Pro/Pro ratio was 0,27:0,52:0,21,  $\chi^2= 21,57$ ,  $p = 0,000$ .

The heterozygous genotype dominated in patients with carcinoma, as well as in the control group. Nevertheless, the number of persons with a heterozygous genotype was significantly higher in the main group - 0,68, in the control group - 0,52 ( $p = 0,03$ ). The homozygous Arg/Arg genotype was rare in patients with carcinoma - 0,11 in the main group and 0,27 in the control group ( $p = 0,005$ ). The increased risk of developing of carcinoma was associated with the accumulation of the Arg/Pro genotype [OR: 1,93 (95% CI:1,05-3,55), while the carrier of Arg/Arg genotype was associated with a decrease in this risk [OR:0,34 (95% CI:0,15-0,73)].

**Conclusions.** Thus, the results of the study showed that an increased risk of developing of carcinoma was associated with the carrier of a heterozygous Arg/Pro genotype, which, nevertheless, was occurred quite often in the healthy group. Carrier of Arg/Arg genotype was associated with decrease in the risk of the disease.

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