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TITLE

SOMATIC MED12 GENE MUTATIONS IN WOMEN WITH BURDENED ANAMNESIS

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ABSTRACT

According to the Ministry of Health and Social development of the Russian federation the frequency of uterine fibroid is 30% among women of reproductive age. Timely detection of uterine fibroids and proper assessment of rates and causes of its growth help not only to clarify the diagnosis, but also to make an individual prognosis and to determine adequate tactics.

In recent years attention has focused on genetic mechanisms of development of uterine fibroids. In this regard, mutations in the gene of MED 12 are interesting for investigations.

Objective: to evaluate and compare the rate and the character of MED 12 exon 2 somatic mutations in 2 groups: the 1st group presented by women with aggravated myoma history, the 2nd group without aggravated history.

Patients: we examined tissues of 75 myomas from 32 patients, as well as aliquots of blood from every patient. The 1st group -17 patients, collected 44 sample myomas. The 2nd group -15 patients, collected 31 sample myomas.

Methods: in our research we carried out DNA purification and MED 12 exon 2 amplification, PCR reaction with primers in the segment of MED 12 exon 2 with determination of a sequence of fragments by Sanger sequencing followed by analysis of somatic mutations.

Results: the proportion of patients with mutations from the 1st group amounted to 71%, the proportion of myomatous nodules with mutations totaled 66%. In the 2nd group the proportion of patients with mutations amounted to 60%. While the proportion of myomatous nodules was of 48%. The proportion of deletions in women with aggravated history amounted to 47% compared to 20% in the 2nd group.

Conclusion: the obtained data confirmed the direct relation of mutation of the gene under study to the state with aggravated history. According to the obtained data the hereditary component determines the selection abundance in MED 12 mutations.

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