Mediterranean fever gene mutation analysis in infertile Turkish males

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ABSTRACT. Male infertility is a common barrier that prevents successful conception. There have been reports of azoospermia in men with familial Mediterranean fever, some of whom had not been treated with colchicine. Variation in this disorder could be a risk factor for amyloidosis associated with azoospermia. We determined the frequency of 6 of the most common Mediterranean fever gene mutations, M680I, M694V, M694I, V726A, P369S, and A744S, in 74 infertile men, 155 men diagnosed with familial Mediterranean fever and 55 healthy fertile men in eastern Turkey. All three groups were screened for the 6 mutations using an amplification refractory mutation system and restriction fragment length polymorphism methods. Allelic frequencies were 2.7% for M694V and 1.35% for V726A in the infertile patient group and 1.8% for M694V and 1.8% for V726A in healthy subjects. Other mutations were not detected in patients or controls. The mutation frequency was not found to be significantly higher in infertile patients when compared with healthy fertile male controls. To our knowledge, this is the first study to determine the frequency of Mediterranean fever gene mutations in infertile male and the infertility rate of male patients with familial Mediterranean fever.

Key words: Familial Mediterranean fever; Male infertility; Mediterranean fever gene