Coexistence of Two Prothrombotic Mutations, Factor V 1691 G-A and Prothrombin 20210 G-A, and the Risk of Thrombosis in Turkish Population

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ABSTRACT

This report summarizes the coexistence of two mutations; Factor V 1691 G-A and prothrombin 20210 G-A in Turkish population and emphasises on the point that, this coexistence increases the risk of thrombosis in such patients. In thrombophilia screening programs, these two variants should be included, particularly in Turkish population.

Key Words: Factor V gene, Prothrombin gene, Thrombosis.

ÖZET

İki Protrombotik Mutasyonun (Faktör V 1691 G-A ve Protrombin 20210 G-A) Birlikteliği ve Türk Toplumunda Tromboz Riski

Bu yazıda Türk toplumunda Faktör V 1691 G-A ve protrombin 20210 G-A mutasyonlarının birarada bulunma sıklığı özetlenmekte ve bu birlikteliğin tromboz riskini arttırdığı vurgulanmaktadır. Türk toplumu trombofili tarama programları için bu iki varyant eklenmelidir.

Anahtar Kelimeler: Faktör V geni, Protrombin geni, Tromboz.

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Point mutations in the factor V gene (FV 1691 G-A) and the prothrombin gene (PT 20210 GA) are the most common causes of inherited thrombophilia. There exist few reports on the risk of thrombosis among carriers of both mutations[1-4]. We previously reported the frequencies of FV 1691A and PT 20210A mutati-

ons to be 9.8% and 2.7% respectively in Turkish population, expecting that for every 400-500 healthy individual would carry both mutations[5,6]. Our present study compiles our data concerning FV and PT mutations in Turkish thromboembolic (TE) patients.

Both mutations were screened according to previously reported techniques. Two- hundred-sixtynine thromboembolic patients with age range of 2 months to 72 years and one-hundred-ninety healthy individuals without any familial history of thrombosis and stroke were included to the study.

Genotype distributions of FV 1691G-A and PT 20210 G-A are given in Table 1. Only one healthy individual was found to carry both mutations (0.5%). Her mother, although carried both mutations (case 11) didnot experience thrombosis. On the other hand, ten individuals were found to carry both mutations in TE group (3.7%). The difference between two groups was statistically different (p= 0.06). Clinical data of the patients carrying both mutations in given in Table 2.

Our data indicated that FV 1691A and PT 20210A mutations are important risk factors for the occurence of thrombosis. The risk of the patient increases when these two mutations coexist in a patient with an OR: 7.3 (1-57.5; CI: 95%). So a careful search for these two variants should be included in thrombophilia screening programs, particularly in Turkish population.

Table 1. Distribution of FV 1691 G-A and PT 20210 G-A in healthy controls and thrombosis patients

	N	FV 1691 G-A	%	OR	CI	PT 20210 G-A	%	OR	CI	Both	%	OR	CI
Normal controls	190	20	10.5	1		4	2.1	1		1	0.5	1	
Patients with thrombosis	269	58	21.5	2.3	1.35-4.0	25	9.2	4.76	1.63-14.0	10	3.7	7.3	1.0-57.5

Table 2. Clinical data of the individuals carrying both mutations

Age/sex	Diagnosis/site of thrombosis	Recurrence
25 F	Deep vein thrombosis	nr
25 M	Pulmonary thromboembolism	r
49 M	Inferior mesenteric artery	nr
72 F	Anterior tibial artery	r
66 F	Femoropopliteal artery	r
28 M	Peripheral artery	nr
15 F	Familial mediterranean fever/vasculitis	nr
18 month F	Cerebral infarct	nr
7 M	Budd-chiari syndrome	nr
2 M	Cerebral infarct	nr
49 F	Normal (detected during family screening)	-
30 F	Normal (detected during population screening)	-

r: Recurrence, nr: Norecurrence.

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