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# Frequency of FV 1299 His-Arg (A4070G) in Turkish Cypriots

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## ABSTRACT

FV 1299 His-Arg polymorphism was recently determined to be common in Turkish population (8.5%). This polymorphism contribute to the activated protein C resistance phenotype. We aimed to examine the frequency of this polymorphism in Turkish Cypriots.

Key Words: FV 1299, Polymorphism, Turkish Cypriots.

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The A4070G polymorphism in exon 13 of the factor V (FV) gene which replaces His by Arg at position 1299 of the B domain, was shown to influence circulating FV levels and to contribute to the activated protein C (APC) resistance phenotype. Double heterozygosity for FV 1691A and FV (A4070G) conferred a 3-to 4-fold increase in the relative risk of venous thromboembolism compared with FV 1691A alone<sup>[1-4]</sup>. We previously reported the high frequencies of thrombophilic alterations FV 1691A (12.2%) and Prothrombin 20210A (8.1%) in Turkish Cypriot population<sup>[5-6]</sup>.

FV 1299 His-Arg was recently determined as a common polymorphism in Turkish population (8.5%)<sup>[7]</sup>. So we aimed to examine the frequency of this polymorphism in Turkish Cypriots.

DNA was extracted by conventional methods and polymerase chain reaction of exon 13 of the FV gene

was performed according to previously described method<sup>[9]</sup>. The distribution of FV 4070 mutation is shown in Table 1. Of the 118 Turkish Cypriots. Five (4.2%) carried 4070G allele in the heterozygous condition (frequency of the G allele was (0.021). None of the 4070G carries had FV1691A.

As both factor V1691A and 4070G mutations are very frequent (i.e. for every 150-200 healthy individual, one would carry both mutations), thus it can be said that the prevalence of these mutations is very high in Turkish Cypriots, almost similar to thalassemia syndromes which are the commonest genetic disease in Cyprus where they cause a very serious health problem<sup>[8]</sup>. So the effect of their coinheritance on the risk of venous thromboembolism might represent a clinically relevant issue in Turkish Cypriots, and screening for FV4070 A-G mutation in carriers of factor V 1691A mutation should be considered.

Table 1. Distribution of FV 4070 A-G in Turkish Cypriots

	n	FV4070G	(%)	Chromosomes	FV4070G frequency
Turkish Cypriots	118	5	4.2	236	0.021

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