Reply

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We would like to thank Castoldi et al. [1] for their valuable comments on our previously published article describing a novel mutation in the clotting factor V gene found in 2 Arab patients with venous thrombosis who were living in Kuwait [factor V Kuwait (FVK) mutation or FV H1254R polymorphism] [2]. As stated by Castoldi et al. [1], the FVK mutation is a very rare one. We have to state here that this polymorphism had not been registered in the human gene bank before we found it in our patients, giving us the impression that it had not been described before. We were the first to register it in the human gene bank with the accession number AY881018. In addition, the FVK mutation was not described as a part of the HR2 haplotype, the haplotype we were originally studying when we found the FVK mutation. In fact, both our patients who had the FVK did not have the HR2 haplotype.

As clarified by Castoldi et al. [1], this mutation appeared to be present mainly in populations in Africa or of African origin. Our article was the first to discover the presence of this mutation in cases of Arabic origin, noting the close geographic location between Africa and the Arabian region (the Middle East). We do not know yet what the origin of this mutation is and whether our patients and the African carriers of the mutation had a common ancestor. We agree with Castoldi et al. [1] that comparative genomic studies are needed to determine the origin of this mutation

(African or Arabic) or confirm a multiple occurrence of this mutation in different parts of the world; this mutation is present especially in an area of the factor V gene known to be a highly repetitive region.

Overall, we agree on the rarity of this mutation in the world, being confined to Africa and the Middle East. The mutation was discovered in only 12 cases worldwide. We added 2 cases from the Arab world. We also repeat our previous suggestion on the need for identifying a larger number of cases before confirming any possible risk of developing venous thrombosis in carriers. We suggest an international collaboration and a registry for cases with this mutation. Finally, we believe it is very important that researchers should register all discovered polymorphisms in the gene bank or other similar databases in order to be available to all other investigators worldwide.

References

- 1 Castoldi E, Lunghi B, Rosing J, Bernardi F: Factor V Kuwait alias factor V R3: a rare polymorphism of uncertain functional significance. *Med Princ Pract* 2007;16:244–246.
- 2 Jadaon MM, Dashti AA, Lewis HL: Factor V Kuwait: a novel mutation in the coagulation factor V gene discovered in Kuwait. *Med Princ Pract* 2006;15:102–105.