

Prevalence of factor V Leiden and prothrombin G20210A gene mutation

To the Editor

I have read the article "Prevalence of factor V Leiden and prothrombin G20210A gene mutation" by Irdem et al¹ with great interest and appreciate the authors' efforts. However, I was concerned that the study was carried out on children and adolescents rather than adults. It is recommended that predictive genetic testing should only be performed on children if it is in their best interest. This view is shared by most leading authorities in Medical Genetics including the American Society of Human Genetics, the American College of Medical Genetics, and the Canadian College of Medical Genetics, in addition to guidelines set forth by the American Medical Association, the American Academy of Pediatrics and the Canadian Pediatric Society. Genetic testing of asymptomatic children for adult onset disorders is discouraged for various reasons including: removing the child's autonomy, removing the child's right to confidentiality, changing the family's perception of the child, potential damage of self-esteem, anxiety generation, enabling social stigmatization, production of parental obligation to determine the right time to inform the child, enabling discrimination (education, employment, insurance) and removing the opportunity for gradual adaptation to potential disease. It is therefore endorsed that the main justification for genetic testing in children should be timely medical benefit to the child involved.²⁻⁴

Even in adults, DNA testing for asymptomatic individuals has many drawbacks and requires careful pre-testing genetic counseling by an experienced professional to ensure the patient understands the pros and cons of testing, including the potential harms involved before proceeding further.⁵ I did not see anything in the methods

section regarding genetic counseling of the subjects or their caregivers prior to testing. This may have been carried out but not written. I considered the possibility that perhaps the patients were never informed of the results, therefore, removing the necessity of counseling and the potential harm of testing. If that is the case, I would still recommend that this be mentioned especially in a Medical Journal with such a wide range of readers, most of whom are not in the genetic field, as this may send the inappropriate message that genetic testing of asymptomatic children or adults is with no restrictions.

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Reply from the Author

No reply received from the Author.

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