Consequences of thrombophilia screening for life quality in women before prescription of oral contraceptives and family members of VTE patients

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Summary
A large number of hereditary and acquired alterations in the coagulation system that are associated with an increased risk of venous thrombosis have been described. Screening for these thrombophilic defects has become particularly popular in women before the prescription of oral contraceptives. The relevance of the results with regard to the management of the patients remains, however, to be questioned. In a recent review of six articles that were aimed to determine the nature and extent of psychological impact of thrombophilia screening, no valid conclusions could be drawn about the psychological impact due to heterogeneity of the data and lack of methodological accuracy. We performed a questionnaire-based study in 247 women with and in 132 women without factor V Leiden who were referred for factor V Leiden testing before oral contraceptive intake. A large proportion (76%) of the women reported being emotionally disturbed by genetic testing. 16% of women with wildtype factor V were discouraged from OC use, while 3% of women with factor V Leiden were encouraged to take OC. This indicates that recommendations after testing are not consistently driven by the test result, which compromises the quality of patient care. Given the large number of women who are taking oral contraceptives unequivocal guidelines for counseling prior to their prescription are urgently needed. These guidelines should integrate the risks and benefits of oral contraceptives but also the impact of screening on quality of life and aspects of counselling.

Thrombophilia and oral contraceptive use

The relationship between oral contraceptive use and an increased risk of thrombosis is known since the early 1960ies. The risk of thrombosis is about three- to six-fold higher among users of oral contraceptives as compared with non-users. Despite these well known thrombogenic effects, oral contraceptives became well accepted among women and gynecologists because of their safety and convenience with regard to contraception as well as other benefits that include reduction of menstrual irregularities and reduced risk of ovarian and endometrial cancer. It became common practice to inform women about the risk of thrombosis and to refrain from prescribing oral contraceptives in case of increased thrombotic risk as judged by a personal history of venous or arterial thrombosis, older age, a positive family history, smoking or obesity. With the identification of new and common thrombotic risk factors these prescription patterns came under new scrutiny. Factor V Leiden results in resistance to activated protein C (APC-resistance) and is the most frequent genetic risk factor of venous thrombosis. Factor V Leiden is found in about 5% of Caucasian populations, and heterozygous carriers have a seven-fold increased venous thrombotic risk as compared with non-carriers. Importantly, the combination of heterozygous factor V Leiden and oral contraceptive use cumulates in a 35-fold higher relative risk of venous thrombosis.

Psychological impact of thrombophilia screening

Overall, data on the quality of life and psychological impact of testing for (genetic) thrombotic risk factors are scarce. In a recent review only six studies which focused on the psychological impact of testing for thrombophilia and were of decent methodology could be identified from large databases. The studies were very heterogeneous with regard to the patient...
population and the thrombotic defects that had been investigated. None of the studies focused on women who were tested before the prescription of oral contraceptives. Overall, a large number of participants reported to be satisfied with the knowledge of being a carrier of thrombophilia. About one third of the participants, however, were more worried after being tested positive for a thrombophilic defect. In addition, some participants also reported that positive testing for factor V Leiden could have led to stigmatization and problems with health insurance. The majority of the participants had either a personal history of VTE or a relative with VTE. It is highly conceivable that being tested for a risk factor has less impact in a patient who is already affected by the disease or is well informed about the disease because of his relationship to a patient with VTE. The impact of testing for a risk factor – particularly if it is hereditary – on young women who are otherwise healthy may be quite different.

Emotional impact of screening for factor V Leiden before the prescription of oral contraceptives

We performed a descriptive study to investigate emotional reactions to being tested for the potential presence of a hereditary risk factor (factor V Leiden) as well as aspects of counseling in asymptomatic women before prescription of oral contraceptives (6). A questionnaire was sent to 247 women who had been tested heterozygous for the factor V Leiden mutation and to 132 women who had been tested negative for the mutation. The response rate was high (about 50%), which suggests that genetic testing is of particular concern to these women. A major finding of the study was that the emotional burden of genetic testing and of the knowledge of being a potential carrier of a hereditary thrombotic risk factor was considerable. 56% of women were uncomfortable with being tested for a genetic risk factor, and 21% reported severe insecurity or fear. Importantly, the degree of emotional disturbance did not differ between women who were finally diagnosed as carriers of factor V Leiden and women who had wildtype factor V. Another important finding was that recommendations given by the physicians after testing were not consistently driven by the test result. Four percent of women with factor V Leiden were encouraged by their gynaecologist to take oral contraceptives, and 14% of those without the mutation were discouraged from oral contraceptive use. Nine percent of factor V Leiden carriers got no recommendation from their gynecologists regarding use of oral contraceptives. In addition, a large proportion of women with factor V Leiden continued to use hormonal contraceptives despite of recommendations to the contrary. These findings implied that even in the absence of medical consequences women were exposed to psychological as well as social stress.

Our study showed that testing for factor V Leiden had strong emotional impact on young women who underwent “routine” screening before prescription of oral contraceptives and were otherwise healthy. Recommendations after testing were heterogeneous and were not consistently driven by the test results, which compromised the quality of patient care.

Conclusions

Only very few studies investigated the impact of thrombophilia screening on aspects of quality of life. Data are very heterogeneous with regard to participants, the thrombophilic defect(s) under investigation and methodology, and valid conclusions cannot be drawn. The emotional impact of genetic testing before prescription of oral contraceptives in young and otherwise healthy women seems to be considerable and the benefits of routine testing is uncertain. The number of women who need to be exposed to thrombophilia screening before prescription of oral contraceptives would be large and many authorities advise against routine screening (5, 14). More studies on the impact of thrombophilia screening on quality of life particularly before the prescription of oral contraceptives are needed. The results of these studies should be integrated in urgently needed unequivocal guidelines for counseling prior to the prescription of oral contraceptives.

References


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