

INHERITED THROMBOPHILIA

INTRODUCTION

Inherited thrombophilia is a blood coagulation disorder that increases the risk for venous thromboembolism (VTE). During the last decades, the practice of testing has evolved from testing selected populations, leading to high perceived risks, to broad testing for various conditions that included VTE, arterial thrombosis, and pregnancy complications. Inherited thrombophilia is widely known to be associated with reproduction, both in men and in women. Current areas of research are whether women with inherited thrombophilia and pregnancy complications benefit from anticoagulant therapy with regard to improving the chance of a successful pregnancy. Potential effects of inherited thrombophilia, most notably factor V Leiden, on improved embryo implantation in women and sperm counts in men are intriguing, but are currently poorly understood.

INHERITED THROMBOPHILIA AND PREGNANCY COMPLICATIONS

A large number of studies have shown an association between inherited thrombophilia and recurrent miscarriage. It has been hypothesized that anticoagulant therapy might reduce the number of miscarriages and stillbirth in these women. In the ALIFE study, the subgroup of women with inherited thrombophilia (n = 47) showed a trend toward a benefit of LMWH and aspirin (relative risk for live birth, 1.31 for the LMWH and aspirin vs placebo; relative risk for live birth, 1.22 for aspirin, with corresponding absolute difference in live birth rates of 16.3% and 11.8%, respectively. LMWH enoxaparin 40 mg subcutaneously once daily started prior to 7 weeks gestational age.

Reference: Middeldorp S. Inherited thrombophilia: a double-edged sword. *Hematology Am Soc Hematol Educ Program*. 2016;2016:1-9

Reasons to Test for Inherited Thrombophilia

- To decide on preventive measures in case of a positive test result; eg, Not to take oral contraceptives
- To use anticoagulant prophylaxis in high-risk situations; eg, pregnancy, after minor surgical procedures (arthroscopy)
- To have an explanation for the disease
- Patient's request (eg, the patient feels thoroughly investigated well taken care)

THROMBOSIS PROPHYLAXIS AROUND PREGNANCY

- Postpartum prophylaxis should be considered in asymptomatic women with all types of inherited thrombophilia and a positive family history of VTE, based on a risk for pregnancy-related VTE of 2% to 4%, and in all women who are homozygous for factor V Leiden or prothrombin 20210A.
- Although the risk for pregnancy-related VTE in women who do not have the inherited thrombophilic defect but come from families with VTE is higher than in the general population, the estimated 0.5% risk does not justify 8 months of antenatal prophylaxis.
- In women who are homozygous for factor V Leiden and who have a positive family history of VTE, the risk for pregnancy-related VTE of 16% is so high that antepartum prophylaxis also is recommended.
- For the deficiencies, most notably antithrombin deficiency, the indication for antepartum prophylaxis remains a matter of debate, and an individual decision based on the physicians' and patients' preference should be made.