What is the risk in pregnancy?

Pregnancy itself carries a small increase in the risk of thrombosis. If you have Antithrombin deficiency we recommend treatment with low dose heparin during and for a few weeks after pregnancy.

Royal United Hospital Bath

Department of Haematology

Antithrombin Deficiency
Information for Patients

June 2006
What is Antithrombin?

Antithrombin is a natural anticoagulant. It was the first recognised cause of thrombophilia in 1965. This is part of the body’s safety mechanism to stop clots forming where they are not needed. When there is a deficiency of Antithrombin the natural balance between clotting and protective anti clotting is changed. This increases the risk of thrombosis. Inheritance of Antithrombin Deficiency is rare but important to recognise as it is one of the thrombophilias with the greatest risk of thrombosis.

How is Antithrombin Deficiency inherited?

We have two Antithrombin genes – one from our mother and one from our father. Therefore there are three possibilities:

- We inherit normal Antithrombin genes from each parent (no increased risk of thrombosis)
- We inherit one normal gene from one parent and one abnormal from the other. This is known as being heterozygous and the Antithrombin levels in our body will be approximately half normal.
- If we inherit the abnormal gene from both our parents we are homozygous and our Antithrombin levels will be very low.

Therefore if a parent is heterozygous they will have one abnormal and one normal gene. This means that their children will have a 50% chance of inheriting the abnormal gene.

If both Antithrombin genes are abnormal the patient is homozygous and will produce very little Antithrombin. This is associated with a severe risk of thrombosis and presents at birth or soon after.

Why is Antithrombin Deficiency important?

Antithrombin deficiency is associated with an increase in the risk of venous thrombosis (clots in the veins), especially in the legs. There is also a lesser association with arterial clots at a young age. Treatment with the anti clotting drug Heparin can also give low results on testing and so we need to know about your medical history and medications when looking at the results of Antithrombin levels.

Treatment

Your doctor can advise you about ways to reduce the risk of thrombosis such as during long-haul flights. Should you require a surgical procedure or long stay in hospital you may need low dose heparin injections to prevent clots. If you develop a thrombosis with Antithrombin deficiency we are likely to recommend lifelong Warfarin.

Should my Family be tested?

Since Antithrombin deficiency has a tendency to run in families, we usually recommend that if one member of a family is found to have Antithrombin deficiency, other members of the immediate family should be tested. Although specific treatment is not required in affected family members, simple precautions in situations of increased risk are advised.

Is the oral contraceptive or hormone replacement safe?

The use of the oral contraceptive pill (OCP) or hormone replacement therapy (HRT) is associated with a small increase in the risk of thrombosis in everybody. This risk is increased if you have Antithrombin deficiency. We don’t advise the use of the pill or HRT if you have Antithrombin deficiency.