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 Prothrombin G20210A. We don't advise the use of the pill or HRT if you have Prothrombin G20210A and have had a thrombosis. If you have Prothrombin G20210A but have never had a thrombosis, the decision is more difficult. We would need to make decision cased on an assessment of all your risks of thrombosis.

What is the risk in pregnancy?

Pregnancy itself carries a small increase in the risk of thrombosis. If you have had a previous thrombosis and have Prothrombin G20210A we recommend treatment with low dose heparin during and for a few weeks after pregnancy. If you have Prothrombin G20210A but have never had a thrombosis, then in most cases, heparin will not be prescribed during pregnancy

Royal United Hospital Bath

Department of Haematology

Prothrombin Gene change G20210A Information for Patients

June 2006

What is Prothrombin G20210A?

This is an inherited Thrombophilia which is associated with a small increase in the risk of thrombosis (clot formation), particularly in the legs. The abnormal gene causes higher than normal levels of the clotting factor called prothrombin. About 2% of western Europeans have this abnormality which increases the risk of thrombosis about three-fold. This seems like a large increase, but because the overall risk of a thrombosis in the general population is so low, despite this increase, most people with prothrombin G20210A never have a thrombosis.

How is Prothrombin G20210A inherited?

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

- We inherit normal Prothrombin 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 half the Prothrombin we produce will be abnormal.
- If we inherit the abnormal gene from both our parents we are *homozygous* and all the prothrombin the body produces will be abnormal.
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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 a parent is *homozygous* both their Prothrombin genes will be abnormal and one copy will be passed on to all their children.

Why is Prothrombin G20210A important?

Prothrombin G20210A is associated with a small increase in the risk of venous thrombosis (clots in the veins), especially in the legs. This risk is highest in patients who have two genes affected (homozygous). Some doctors believe that it may contribute occasionally to an increased risk of heart disease. For this reason we encourage people with Prothrombin G20210A to avoid other risk factors for heart disease such as high blood pressure, smoking and high cholesterol.

Treatment

Most people with Prothrombin G20210A require no specific 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 have two abnormal Prothrombin genes, another thrombophilia in addition to Prothrombin G202101A or repeated episodes of thrombosis, we may recommend lifelong Warfarin treatment.

Should my Family be tested?

Since Prothrombin G202110A has a tendency to run in families, we usually recommend that if one member of a family is found to have Prothrombin G20210A, 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. Because the risk of thrombosis in children with Prothrombin G20210A is so small, we do not recommend testing in the under sixteens.