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Prothrombin Gene Mutation

What is the Prothrombin Gene Mutation?

The Prothrombin Gene Mutation is a change in the gene that controls a blood clotting protein called prothrombin, which circulates in the blood. This gene change leads to an increase in the level of this protein in the blood.

How did I get the Prothrombin Gene Mutation?

It's an inherited condition. We inherit one copy of each gene from each parent — one (or both) of your parents will have passed the mutation on to you, and you may pass it on to your children.

How common is the Prothrombin Gene Mutation?

It's relatively common — occurring in about 1 in every 50 people in Australia, more common than many other gene changes. Because it's so common, it has been suggested there may be some “benefit” to having it.

What are the main problems with having the Prothrombin Gene Mutation?

The main problem is an increased risk of developing blood clots, which can sometimes travel to different parts of the body, including the lungs. If you have one copy of the mutation (heterozygote), you are at around 3 times more risk of developing a blood clot compared to someone without it.

Am I going to get a blood clot because I have this mutation?

Most people with one copy of the Prothrombin Gene Mutation do not develop blood clots. Clots usually only develop when other “risk factors” are present — for example, after long flights, after surgery, or in people with other serious medical conditions such as cancer or heart failure.

What about the use of oral contraceptive pills?

The pill can increase the risk of developing a blood clot. Women on the pill have around a 3–4 fold increased risk compared to those not on it. If a woman also has one copy of the Prothrombin Gene Mutation, this risk increases to around 16 times. These figures sound high, but clots remain relatively uncommon in the community.

Are there any benefits of having the Prothrombin Gene Mutation?

Because blood clots more easily in people with this mutation, it has been suggested this may be beneficial during bleeding episodes (e.g. menstruation or childbirth). This is only a theory and has not been confirmed by studies.

Is there any treatment for the Prothrombin Gene Mutation?

No treatment to change genes is currently available. Most people do not need any treatment, but should be careful at times when clot risk may be increased (e.g. after surgery, during long flights — see information sheet on Clots and Flights). Some people may need blood thinning medication depending on personal or family history of clots.

Is there anything else I should do to protect myself against getting blood clots?

Maintaining a healthy weight, stopping smoking, staying active, and keeping other medical conditions under control should also help. Tell your doctor or surgeon that you have the Prothrombin Gene Mutation before any operations or prolonged periods of bed rest.

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Should my family be tested for the Prothrombin Gene Mutation?

Testing is easily done with a simple blood test, but you and your family should think carefully before testing, as there may be implications for health and life insurance policies. Clots in children are very uncommon, so it's generally best to wait until children are older (usually in their late teens) and can decide for themselves whether to be tested.

References

1. Investigation and management of heritable thrombophilia. British Journal of Haematology 2001;114(3):512-528.

Further Questions?

The information presented in this fact sheet is intended as a general guide only.

Patients should seek further advice and information about **Prothrombin Gene Mutation** and their individual condition from their treating haematologist or doctor.

For additional information about blood disorders and their treatment, or to contact one of our specialist haematologists, visit the Melbourne Haematology website:

www.melbournehaematology.com.au