

Prothrombin Gene Mutation Information for patients and families

What is prothrombin gene mutation?

Prothrombin gene mutation is a blood clotting disorder, not a disease. It causes the body to make too much prothrombin, which makes the blood more likely to clot. Blood clots in your lungs or the deep veins of your legs can be dangerous.

Prothrombin Gene Mutation is also called:

- Factor II mutation
- Prothrombin 20210 mutation
- Prothrombin 20210A mutation

| What the medical words mean | |
|-----------------------------|--|
| Prothrombin | <ul style="list-style-type: none"> • Prothrombin is a protein in the blood that is needed for the blood to clot properly. It is also called factor II (two). |
| Gene | <ul style="list-style-type: none"> • Genes are found in every cell of the body. They carry instructions for making proteins. This “genetic code” determines the characteristics of a living thing. • You have two copies of each gene, one inherited from each parent. Together they determine your unique physical features. • One gene carries the instructions for making the prothrombin protein. |
| Mutation | <ul style="list-style-type: none"> • A change in a gene. • If the gene for making prothrombin has a mutation, the body will make too much of the prothrombin protein. This makes the blood more likely to clot. • One or both parents can pass on the prothrombin gene mutation to their child. |

How does the prothrombin gene mutation affect clotting?

When you get injured, your body stops bleeding by forming a plug (clot) to prevent more blood from leaking through damaged blood vessels.

Blood clots are made of platelets and the blood clotting protein 'fibrin'. Prothrombin is a blood clotting protein that is needed to form fibrin.

The prothrombin gene mutation, causes your body to make too much prothrombin. This makes your blood more likely to form clots, which can be dangerous.

How is the diagnosis made?

The diagnosis of a prothrombin gene mutation is made by a blood test. The blood is then sent to a laboratory, where the genes are examined for the prothrombin gene mutation.

How did I get the prothrombin gene mutation?

Everyone inherits two copies of the prothrombin gene. You inherit one copy of the gene from your mother and one from your father.

There are two types of prothrombin gene mutation:

1. Heterozygous:

- You have inherited one copy of the prothrombin gene mutation, from one parent. The other copy of the prothrombin gene, inherited from your other parent, is normal.

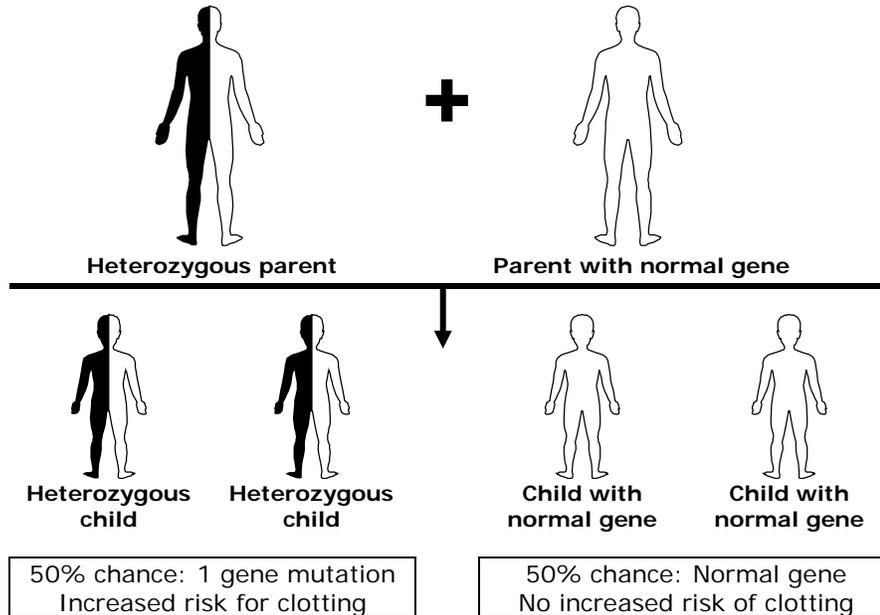
2. Homozygous:

- You have inherited two copies of the prothrombin gene mutation, one from each parent. This is more rare than the heterozygous type.

Your brothers and sisters may also have inherited a prothrombin gene mutation, just as you did.

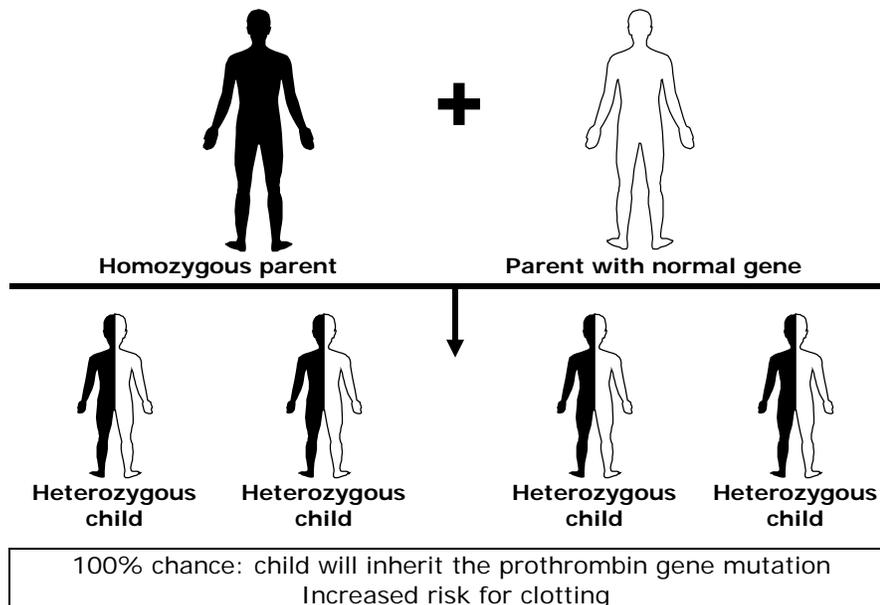
Heterozygous prothrombin gene mutation

You inherited 1 prothrombin gene with a mutation from one parent and 1 normal prothrombin gene from your other parent.



Homozygous prothrombin gene mutation

You inherited one prothrombin gene with a mutation from one parent, and another from your father.



Can I pass on the prothrombin gene mutation?

Yes, you can pass on the mutation to your children.

If you have the heterozygous type, your child has a 50% of inheriting the prothrombin gene mutation from you. There is also a 50% chance that your child will inherit your normal prothrombin gene.

If you have the homozygous type, your child will inherit the prothrombin gene mutation. Your child will have either the heterozygous or homozygous type, depending on whether the prothrombin gene inherited from his or her other parent is normal (no mutation) or abnormal (with the mutation).

What does this mean for me?

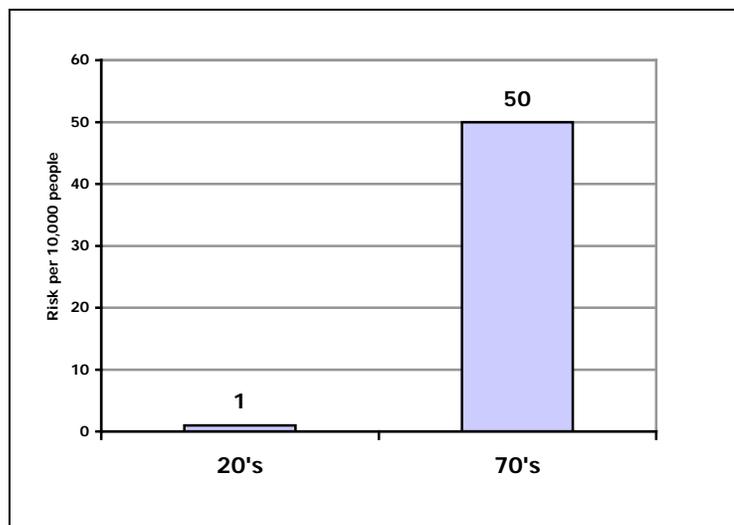
Risk for blood clots

Having a prothrombin gene mutation means that you have a higher risk of developing a blood clot in your legs (called Deep Vein Thrombosis or DVT) and/or lungs (called Pulmonary Embolus or PE).

About 1 in every 1000 people will develop a DVT or PE each year.

The risk of developing a DVT or PE increases with age.

The risk for people in their seventies is much greater than for people in their twenties.



Many people with the prothrombin mutation may never develop a blood clot in their lifetime. People who have the prothrombin mutation and develop a blood clot often have other risk factors that increase their chances of getting a blood clot.

Other risks

Most research studies show that prothrombin gene mutations do not increase risk for heart attack and stroke in middle-aged and elderly people. However, some studies suggest that having the prothrombin gene mutation may increase the risk of heart attacks in young women, in particular those who smoke.

Some research suggests that prothrombin gene mutations increase the risk for certain pregnancy complications such as pregnancy loss (miscarriage) and high blood pressure during pregnancy (preeclampsia). However it is not clear how strong the association may be. Most women with the prothrombin gene mutation have normal pregnancies.

Can the prothrombin gene mutation be treated?

No treatment can prevent the prothrombin gene mutation or make it go away. The goal of treatment is to prevent blood clots (see next question and page 7).

If you have had a blood clot, you may be treated with medications called anticoagulants. Anticoagulants are used to help to prevent blood clots. They are also known “blood thinners.” If you have never had a blood clot, you do not need to take anticoagulants.

You may need to take anticoagulants during times when there is a higher chance of getting blood clots (such as surgery, a leg cast, a serious illness, or a pregnancy). You can stop taking them when the risk has passed.

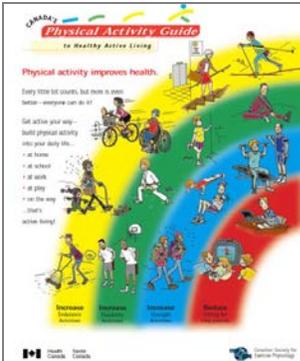
What can I do to avoid blood clots?

Keep your weight in a healthy range.



- Eat well. You can eat a variety of foods from the 4 food groups.
- Follow Canada's Food Guide. www.hc-sc.gc.ca/fn-an/food-guide-aliment/index_e.html
- If you are overweight or obese, consider the benefits of losing weight. You can talk with your health care provider for advice and help on how to lose weight.

Build physical activity into your daily life.



- Be active by doing activities you enjoy. Exercise regularly, such as walking, playing sports, swimming or riding a bike 3 times a week.
- Follow Canada's Physical Activity Guide. www.phac-aspc.gc.ca/pau-uap/paguide/
- If you have an inactive (sedentary) lifestyle, consider how you could gradually increase your level of physical activity at home, school, work or play. You can talk with your health care provider for advice on how to do this safely.

Take care when travelling long distances.



- Long periods of immobility, including times of travel, can increase chances of developing a blood clot.
- During long car rides, stop every few hours and take a short walk. This helps keep the blood flowing in your legs and prevent a blood clot.
- On a long flight, take regular walks down the aisle of the plane. You can also do exercises (including calf raises and frequent toe squeezes). Drink plenty of fluids (without alcohol). Your doctor can tell you how much fluid is right for your age, health and activity level.

Do not smoke.



- Smoking increases the risk for developing blood clots. If you smoke, consider the benefits of quitting.
- You can talk to your health care provider for advice on how to quit. There are many resources such as programs and websites that can help you quit.

What about risks I can't avoid?

- Serious illness, casts, and surgery can increase your risk of developing blood clots. When you need health care services or go to the hospital, tell the health care team that you have the prothrombin gene mutation. This will help them plan the care you need. The health care team may treat you with blood thinners until the risk has passed.
- During pregnancy, women have a higher risk of developing blood clots. Talk with your doctor before becoming pregnant about how your risk of blood clots can be managed during pregnancy, childbirth, and for 6 weeks after delivery (postpartum). During this time, you may be treated with blood thinners.
- The risk of blood clots is increased in women who take hormones and birth control pills. Talk with your doctor about the risks and benefits of taking these medications.

How will I know if I get a blood clot?

Learn these signs of a blood clot so that you can get help as soon as possible.

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| If you have any of these signs, call your doctor right away. | |
| Signs of DVT: A clot in the deep veins of your legs | <ul style="list-style-type: none"> • Pain or tenderness in the back of your calf or inside your thigh • Swelling or warmth in your arm or leg • Redness on your arm or leg |
| Signs of PE: A clot in your lungs | <ul style="list-style-type: none"> • Shortness of breath • Chest pain • Passing out • Coughing up blood |
| If you cannot reach your doctor go to the nearest hospital emergency room. | |

Healthy Living

Preventing blood clots

- Drink plenty of fluids
- Follow Canada's Food Guide
- Work with your health team to manage your health
- Balanced lifestyle
- If you smoke, consider the benefits of quitting
- Take care when traveling long distances
- Talk to your health care professional if you have questions
- Follow Canada's Physical Activity Guide

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