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Congenital fibrinogen deficiency

Congenital fibrinogen deficiency is a very rare, inherited blood disorder in which the blood does not clot normally. It affects a protein called fibrinogen. This protein is needed for the blood to clot.

Causes

This disease is due to a deficiency of the amount or function of fibrinogen. How fibrinogen is affected depends on how the genes that determine how it is formed are inherited:

- When the abnormal gene is passed down from both parents, a person will have a complete lack of fibrinogen (afibrinogenemia).
- When the abnormal gene is passed down from one parent, a person will have either a reduced level of fibrinogen (hypofibrinogenemia) or a problem with the function of fibrinogen (dysfibrinogenemia). Sometimes, these two fibrinogen problems can occur in the same person.

Symptoms

People with a complete lack of fibrinogen may have any of the following bleeding symptoms:

- Bruising easily
- Bleeding from the umbilical cord just after birth
- Bleeding in the mucous membranes
- Bleeding in the brain (very rare)
- Bleeding in the joints
- Heavy bleeding after injury or surgery
- Nosebleeds that do not stop easily

People with a reduced level of fibrinogen bleed less often and the bleeding is not as severe when compared to people with no fibrinogen at all. Those with a problem with the function of fibrinogen often don't have symptoms.

Exams and Tests

If your health care provider suspects this problem, you will have lab tests to confirm the type and severity of the disorder.

Tests include:

- Bleeding time
- Fibrinogen test and reptilase time to check fibrin level and quality
- Partial thromboplastin time (PTT)
- Prothrombin time (PT)
- Thrombin time

Treatment

The following treatments can be used for bleeding episodes or to prepare for surgery:

- Cryoprecipitate (a blood product containing concentrated fibrinogen and other clotting factors)
- Fibrinogen (RiaSTAP)
- Plasma (the liquid portion of the blood containing clotting factors)

People with this condition should get the hepatitis B vaccine. Having many transfusions raises your risk of getting hepatitis.

Outlook (Prognosis)

Excessive bleeding is common with this condition. These episodes may be severe, or even fatal. Bleeding in the brain is a leading cause of death in people with this disorder.

Possible Complications

Complications may include:

- Blood clots with treatment
- Development of antibodies (inhibitors) to fibrinogen with treatment
- Gastrointestinal bleeding
- Miscarriage
- Rupture of the spleen
- Slow healing of wounds

When to Contact a Medical Professional

Contact your provider or seek emergency care if you have excessive bleeding.

Tell your surgeon before you have surgery if you know or suspect you have a bleeding disorder.

Prevention

This is an inherited condition. There is no known prevention.

Alternative Names

Afibrinogenemia; Hypofibrinogenemia; Dysfibrinogenemia; Factor I deficiency

References

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