Inherited Disorders of Fibrinogen

What is Fibrinogen?

Fibrinogen is a protein that is made in the liver and is present in blood and is essential to the blood clotting process. Fibrinogen is converted into fibrin by thrombin in the presence of calcium ions. Fibrin is a stringy protein involved in giving a blood clot its character and thrombin is an enzyme made in plasma during the clotting process.

Types of abnormalities of Fibrinogen

1. **Afibrinogenaemia** occurs when there is no Fibrinogen able to be measured in the blood.

2. **Hypofibrinogenaemia** is a low level of normal fibrinogen

3. **Dysfibrinogenaemia** occurs when there is fibrinogen present but it is not normal and does not function as normal fibrinogen does.

Your child has______________________________

What are the signs and symptoms?

**Afibrinogenaemia and Hypofibrinogenaemia**

Bleeding; this varies and there can be long periods where there are no problems with bleeding. The most common areas for bleeding are the mucus membranes, which line many parts of the body such as the mouth and the gut. Bleeding into muscles and joints can occur but are not common. Indications of this include pain, heat and swelling of the joint/muscle.

Hypofibrinogenaemia the bleeding pattern is similar but appears to be milder and bleeding may follow invasive procedures (ie surgery).
Both hypofibrinogenemia and afibrinogenemia have been associated with recurrent miscarriage and bleeding during pregnancy and after delivery of a baby.

Rare symptoms can include clots and impaired wound healing after surgery.

**Dysfibrinogenaemia**

Studies have shown that the way dysfibrinogenaemia can present varies with many people having no symptoms whilst some have problems with bleeding and others with clots.

For those with bleeding symptoms, problems may occur after the birth of a baby, after surgery and dental extraction. In some cases where there are low levels, bleeding has occurred into soft tissues and the central nervous system. Some people also experience slow wound healing after surgery.

Thrombosis (or clots) has been found to occur in some people with dysfibrinogenaemia. The symptoms are not common but can result in deep vein thrombosis and thrombophlebitis (inflammation of a vein often seen with the formation of a clot).

**Diagnosis**

This is done by a blood test and in some cases testing of other family members may be required.

**Treatment**

How this is treated is will depend on your child’s personal and family history of bleeding and thrombosis. In many cases no treatment is required, however if treatment is required Fibrinogen concentrate will be considered.

Cryoprecipitate (a good source of FI, not often used as not virally inactivated). Its use may be considered in an emergency situation if no suitable alternative is available.

Fibrin glue may be used to treat minor wounds or after dental extractions.

Tranexamic acid can be useful to treat mucosal bleeding or prevent bleeding for procedures such as dental extraction, avoiding the need for blood products. For those that have problems with clots this should be used with caution.
Your doctor will advise you on the appropriate treatment for your child and when you should see your local GP or Haematologist.

It is important to remember this is not a contagious disorder and children can lead normal lives. If bleeding occurs it is advised to contact your local GP or Haematologist.

Apply RICE (rest, ice, compression, elevate) if bleeding occurs in a joint/muscle. If bleeding occurs from the mouth seek advice.

If planning to fly it is advisable to first check with your Haematologist.

**You should see your local GP for treatment when?**
(to be completed by medical staff)

**You should contact your child’s haematologist when?**
(to be completed by medical staff)

Further information is available from your Haematologist

**Reference**
The rare coagulation disorders-review with guidelines for management from the UKHCDO. *Haemophilia (2004)*, 10, 593-628