Introduction

This leaflet aims to provide you with general information about the signs and symptoms your child may have when they have Factor 7 deficiency. If you are ever worried about your child please contact your IBD Team or take your child to the Accident and Emergency Department.

What is Factor 7 deficiency?

Factor 7 deficiency is a disorder of the blood clotting (coagulation) system where there is a reduction or absence of clotting factor 7. Girls and boys who are affected by factor 7 deficiency bleed for longer, but not faster than other people.

This leaflet is about severe forms of Factor 7 deficiency, the symptoms are often noticed early in life. If your child has a mild form of the disorder we can give you a leaflet on mild bleeding disorders.

Where did my child’s Factor 7 deficiency come from?

Factor 7 deficiency is a very rare inherited disorder affecting 1:300,000 - 1:500,000 people. Sometimes there can be a family history of the disorder; however your child may be the first in the family affected by severe Factor 7 deficiency.

How is the diagnosis made?

The diagnosis is made by taking a “bleeding history”. You will be asked if your child bruises or bleeds easily and about problems with any surgery or dental procedures. If your child is very young you may not be able to answer some of these questions and we understand this. You will also be asked if other family members are affected by any bleeding or bruising.

As well as asking you questions we will take some blood to check how the blood is clotting and to see if the Factor 7 is low. Sometimes these bloods will need to be repeated as the levels can fluctuate. Further blood tests which may include looking at the Factor 7 gene can help us to discover where the factor 7 deficiency has come from, give you accurate information and advice as well as to manage your child’s disorder. Genetic tests can only be undertaken with consent.

Inheritance of Factor 7 deficiency

Factor 7 deficiency is an autosomal recessive disorder. This means that a child has to inherit the faulty gene from both parents to develop a severe form of Factor 7 deficiency. Each pregnancy carries a 25 per cent risk of having an unaffected child, 25 per cent risk of having an affected child, and 50 per cent risk of having a child who is a carrier of the affected gene.
Once there is genetic confirmation of your child’s disorder we will be able to advise you if you or other family members need to attend for investigations.

**What is Factor 7 deficiency?**

Some people have a mild form of the disorder which means that the symptoms may not be noticed until later in life.

There are two types of Factor 7 deficiency:

- Type 1 is where the Factor 7 is absent or in low levels in the blood.
- Type 2 is where Factor 7 is present but it does not work properly.

The severity of symptoms ranges from mild to severe depending on the amount of Factor 7 present in the blood and its activity.
How does blood clot?

Clotting factors are proteins in the blood that control bleeding.

When a blood vessel is injured, the walls of the blood vessel contract to limit the flow of blood to the damaged area. Then, small blood cells called platelets stick to the site of injury and spread along the surface of the blood vessel to stop the bleeding.

At the same time, chemical signals are released from small sacs inside the platelets that attract other cells to the area and make them clump together to form what is called a platelet plug.

On the surface of these activated platelets, many different clotting factors work together in a series of complex chemical reactions (known as the coagulation cascade) to form a fibrin clot. The clot acts like a mesh to stop the bleeding.

Coagulation factors circulate in the blood in an inactive form. When a blood vessel is injured, the coagulation cascade is initiated and each coagulation factor is activated in a specific order to lead to the formation of the blood clot. Where one or more of the clotting factors is reduced or not working properly the coagulation cascade is affected slowing the time for the fibrin clot to form.

What are the signs and symptoms of factor 7 deficiency?

- Bleeding around the umbilicus after birth
- Bruising (more than other children); these bruises often have hard lumps in the middle which can last long after the bruise has gone
- Bleeding gums
- Prolonged bleeding from cuts
- Nosebleeds
- Bruising or bleeding after vaccinations
- In those with severe factor 7 deficiency (less than 3% factor 7), bleeds into muscles and joints
- Heavy periods
When should we ask you for advice?

If you are worried about your child please try to contact the IBD Team on the numbers provided. However if it is out of hours, and you have been unsuccessful, please do not wait at home for a response but attend the Accident and Emergency Department. Make sure you tell them on arrival that your child has factor 7 deficiency.

If your child has:

- Has a significant head injury with any signs of the following signs:
  - Babies: any signs of high pitched crying increased sleepiness decreased alertness, reduced feeding or vomiting.
  - Older children: any signs of visual disturbances, loss of consciousness, severe or ongoing headache, loss of coordination, tiredness and decreased alertness, difficulty in thinking or severe or persistent vomiting
- is showing signs of a joint bleed: Restricted movement in the affected area, swelling, tenderness and heat when touched and pain on movement. Reluctant to use that arm or leg.
- is showing signs of a muscle bleed: Firm, tender and swollen to touch with restricted movement.
- has a prolonged nosebleed which does not seem to be slowing after 10-15 minutes
- has a loose tooth that is bleeding
- has an uncontrollable period

Please let us know if your child needs surgery or dental extractions. It is important for us to know so that we can make a simple plan to make sure the procedure runs smoothly.

Can my child join in with sports with Factor 7 deficiency?

Yes, most sports are encouraged as they can help to make your child’s muscles and joints stronger. Contact sports such as rugby, mixed martial arts and boxing should be avoided, tag rugby can be participated in. All other sports football, swimming, dancing, basketball and cycling are encouraged and you child should be allowed to join in.

How can Factor 7 deficiency be treated?

Severe forms of factor 7 deficiency your child may need treatment for injuries, surgery and dental extractions, and sometimes to prevent bleeding episodes.

If a bleed is suspected use normal first aid measures first; this includes Protect Rest Ice Compression Elevation otherwise known as PRICE

Protect and (pain killers). Make sure your child is in a safe place and that the injured or painful area is not at risk of being knocked, paracetamol may be needed to help the pain to settle

Rest - Rest the affected joint or muscle. If your son has an elbow or shoulder bleed he should rest his arm and not use it to lift or carry things. If it is a joint or muscle bleed in his leg he should not walk but rest as much as possible.

Ice - Apply ice, wrapped in a towel, or a cold pack to the affected joint either by using an ice pack, crushed ice in plastic bag or frozen vegetables, apply for 10-15 minutes. Ice helps to reduce pain and muscle spasms as well as reducing swelling and redness. Ice alternating with heat can also help to reduce pain, particularly when the bleed is into a muscle.
Compression - Applying pressure to the injured area will help slow blood flow; the pressure can also provide comfort. A Tubigrip bandage is a good example of compression and can be used over the knee, ankle, elbow or wrist. Compression is often too painful in the early stages of a bleed.

Elevation - Raise the injured arm or leg above the heart although this may seem difficult you can prop an arm and leg up with cushions or pillows. This helps to reduce the blood flow to the area and lower blood congestion to slow the bleeding.

Currently there is no cure for Factor 7 deficiency, but there are medicines and treatments available that help to keep symptoms under control.

**Tranexamic acid** is helpful for gum bleeds, nose bleeds and dental extractions (mucosal bleeding). It works by helping to stabilise blood clots while the blood vessel repairs itself. It is usually given orally but may be given as an infusion intravenously.

**Factor 7** is given as an injection into the vein. It can be used to prevent bleeding episodes as well as to stop bleeding into muscles and joints, during surgery or dental work and sometimes for nose bleeds that do not stop. It works by increasing the factor 7 in the bloods stream bringing the factor levels up to normal so that the bleeding will stop.

**Who do I tell?**

It is helpful for other healthcare professionals to know including your GP and Health Visitor.

Child minder, play group, nursery and schools need to know as do the leaders of any club or group he joins. We routinely visit schools to talk to teachers and lunchtime staff and provide them with written information.

You might find it helpful to tell friends about the factor 7 deficiency so that they can support you and so that if your child develops bruises while playing with their children they can understand why.

**How often will we see the IBD Team?**

We will see you or speak to you as often as you would like. We know that lots of questions or worries do not wait for a clinic appointments.

Routinely we will see you with your child at diagnosis and then in clinic for primary immunisations, we give all immunisations just under the skin. This is a good time for us to get to know you, and for you to get to know the team. It is a good time for you to ask any questions and for us to talk a little about factor 7 deficiency. After this we will see you in clinic, first at 3 and then 6 month intervals. You might feel happier coming more often this can be easily arranged.

**Medicines that should be avoided:**

Please avoid using pain killers known as NSAIDs including ibuprofen, naproxen and diclofenac as these interfere with the way the blood clots by affecting the function of platelets, NSAIDs can also irritate the lining of the stomach, causing bleeding. You can give your child paracetamol if he needs a painkiller or something to reduce a temperature.
Further booklets and information sheets are available from the Haematology Treatment Room waiting area, with further information from the Haemophilia Society.

Links:
The Haemophilia Society website: [https://haemophilia.org.uk/](https://haemophilia.org.uk/)
Alder Hey Children’s NHS Foundation Trust: [https://alderhey.nhs.uk/](https://alderhey.nhs.uk/)

Contact Information:
Haematology Nurses: 8.30am-5.00pm  0151 252 5070 with answer machine non urgent queries will be reviewed by the end of the next working day
Nicki Mackett:  07584 234 526  nicki.mackett@nhs.net
Catherine Benfield:  07876 132 163  catherine.benfield@nhs.net
For urgent clinical needs please go directly to Accident and Emergency Department

This leaflet only gives general information. You must always discuss the individual treatment of your child with the appropriate member of staff. Do not rely on this leaflet alone for information about your child’s treatment.

This information can be made available in other languages and formats if requested.

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