Introduction

This leaflet aims to provide you with general information about the signs and symptoms your child may have when they have Severe Haemophilia B. 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 haemophilia B?

Haemophilia B is a disorder of the blood clotting (coagulation) system where there is a reduction or absence of clotting factor IX (9). Men and boys affected by Haemophilia B bleed for longer, but not faster than other people.

Where did my son’s haemophilia come from?

Haemophilia B is an inherited disorder that affects 1:50,000 males. Often there is a family history of the disorder, but in a third of cases this can be “spontaneous” and your son may be the first person in the family known to be affected.

How is the diagnosis made?

The diagnosis is made by taking a “bleeding history”. You will be asked if your son 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.

As well as asking you questions we will take some blood to check how the blood is clotting and if the factor 9 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 9 gene can help us to discover where the factor 9 deficiency has come from, give you accurate information and advice as well as to manage your child’s disorder.

Inheritance of haemophilia B

Haemophilia B is an inherited X linked recessive disorder; this means that women can be carriers of the disorder but the symptoms are seen in males. Sometimes women who are carriers of haemophilia have lower Factor 9 levels. These women are classed as low level haemophilia carriers and can have symptoms similar to men and boys who have mild haemophilia.

In each pregnancy a carrier for Haemophilia has a 1:2 (50%) risk of passing the affected gene on to her child. A man with haemophilia B cannot have a son affected by haemophilia B but his daughters will all be obligate carriers for Haemophilia B. We can arrange for the necessary genetic tests to be carried out at Alder Hey or an appointment can be arranged to be seen by a geneticist.
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 does severe Haemophilia mean for day to day life?**

Although it is tempting you do not need to “wrap your son up in cotton wool”. You can get on with day to day life and join in almost all childhood activities. We do advise against some contact sports such as rugby, judo and karate.

**What is Severe haemophilia B?**

Haemophilia B has three classifications

**Severe: Factor 9 less than 3%**

Moderate: Factor 9 3.1-7.9%

Mild: Factor 9 8-40%

The severity of haemophilia runs true within a family so that if other family members have severe haemophilia then your son will also have severe haemophilia. In some forms of severe haemophilia B the factor 9 level increases gradually so that by young adulthood the levels are at the lower end of normal. We will let you know if your son is in this group of boys.
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 severe haemophilia B?

- Bruising more than other children often without any known injury, 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
- Muscle bleeds
- Joint bleeds
How should you ask for advice?

If you are worried about your son please 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 haemophilia.

Situations when you should ask for advice

If your son:

- 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, 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,

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

Can my child join in with sports?

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. Other sports including football, swimming, dancing, basketball and cycling are encouraged and your child should be allowed to join in.

How can Haemophilia B be treated?

If a muscle or joint 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 son 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 you 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 a knee, ankle, elbow or wrist bleed. 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.

There is no cure for haemophilia 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 9 can be given as an injection into the vein. It is used to stop bleeding into muscles and joints, during surgery or dental work and sometimes for nose bleeds that do not stop. It works by replacing the factor 9 in the blood stream bringing the factor levels up to normal so that the bleeding will stop.

Boys with severe haemophilia have problems with bleeding into muscles and joints, If these are not managed well they can be painful and eventually lead to long term damage.

Traumatic Bleeds: are those which happen after a known injury.

Spontaneous Bleeds: develop when there has been no known injury. Bleeding episodes should always be treated with replacement clotting factor 9, so that the bleeding can be switched off. With severe haemophilia we would aim for primary prophylaxis in most instances.

What is Prophylaxis?

Most boys with Severe Haemophilia begin a prophylaxis treatment schedule at approximately 12 months of age. Prophylaxis is preventative treatment which is given to reduce the risk of bleeding into joints and muscles. Most prophylaxis programmes are for treatment 1-2 times a week. The aim of the programme is for the Factor 9 levels to be kept above 3% reducing the risk of bleeding episodes

Usually prophylaxis is gradually introduced starting with one treatment per week increasing to twice if required

There are different types of prophylaxis

- **Primary prophylaxis:** Factor 9 replacement given regularly once a week. It is used as long term management of haemophilia and is usually started after the first joint or muscle bleed or by the age of two years.
- **Secondary Prophylaxis:** Factor 9 replacement given regularly once a week. It is used as long term management of haemophilia and is started after two or more joint bleeds have occurred in order to prevent joint damage
- **Prophylaxis for activity:** Treatments prior to activities for example every Wednesday before football

Prophylaxis aims to keep the factor 9 level above 3%. When the factor 9 levels fall below this there is an increased risk of a breakthrough bleed.

Why give prophylaxis?

We need to keep your son’s joints and muscles healthy, to do this we need to be able to keep bleeds to a minimum. Recurrent bleeding into a joint causes damage to the soft spongy tissue, the cartilage, and the thin layer of tissue lining the inside of the joint (synovium). Joint damage is more commonly noticed in adults but without a good treatment and prophylaxis plan minor damage can begin at a much earlier age.
What is a breakthrough bleed?

Prophylaxis aims to keep Factor 9 levels between 50% (just after the injection) and 3% (just before the next dose of treatment is due). Breakthrough bleeds can happen “spontaneously” despite prophylaxis; they are most likely to occur as the factor 9 level falls to 3% or below.

What is a target joint?

A target joint is one in which 3 spontaneous bleeds have occurred in a six month period. In this situation we would urgently want to evaluate and update prophylaxis and treatment plans.

Treating your son at home

We want you have the opportunity to become independent, but not isolated from Alder Hey. As you become more familiar with haemophilia, the signs and symptoms of a bleed and bleed management you may also begin to feel that giving treatment at home is a possibility

Home Treatment can be helpful for your son, you and the rest of the family as:

- Bleeds can be treated more quickly
- You rely less on hospital based treatment giving you more time to be at home, school or work.
- It increases your confidence in managing the haemophilia
- You can be more independent

How is treatment given?

Treatment is given one of two ways either directly into the vein with a butterfly needle or into a surgically implanted device called a port a cath; more detailed information is in the leaflet “My child needs a port a cath2”. Ask the nurse specialists if you have any questions, concerns or would like more information on these treatment methods.

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.

We might advise you to talk to family members who may need to ask a doctor to check if they could be affected by haemophilia.

You might find it helpful to tell friends about haemophilia so that they can support you and so that if your son 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 son at diagnosis and then in clinic for primary immunisations. We give all immunisations just under the skin, to reduce risk of bleeding into the muscle. 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 mild haemophilia. After this we will see you in clinic first at 3 and then 6 month intervals. You might feel happier coming more often and 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 son paracetamol if he needs a pain killer or something to reduce a temperature.

Links

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

Further booklets and information sheets are available from the Haematology Treatment Room waiting area, with further information from the Haemophilia Society.

Contact Information

Haematology Nurses: 8.30am-5.00pm 0151 252 5070 with answer machine for non urgent queries which 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 son with the appropriate member of staff. Do not rely on this leaflet alone for information about your son’s treatment. Specific information about your son’s treatment will be included in the clinic letter you are sent and is also available on the hospital’s computer system.

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