Haemophilia is a genetic bleeding disorder that affects approximately 1 in 10 000 boys. In haemophilia, the blood takes a long time to clot or does not clot at all. As a result, haemophilia is characterized by persistent or excessive bleeding following a minor injury, dental extraction or surgical procedure; or in more severe cases, by spontaneous bleeding into the skin or joints. Haemophilia A (Classic haemophilia) is the most common form of the disorder, affecting about 1 in 5000 boys. It is caused by deficient activity of factor VIII (anti-haemophilic globulin), a clotting factor in the blood. Haemophilia B, also known as Christmas disease, is due to a deficiency of factor IV. Both haemophilia A and B have x-linked recessive inheritance. This means that haemophilia is controlled by a sex-linked gene, occurs almost exclusively in males and is transmitted to them, by women who carry the disease without being affected themselves. Haemophilia C, caused by a factor XI deficiency, is an autosomol-recessive disease, occurring with high frequency in the Jewish population. Haemophilia A and B have similar clinical problems, with the severity of the conditions corresponding to the levels of Factor XIII and IV in the blood. Haemophilia C is a milder disease (Concise Medical Dictionary, 1994; Coovadia, H.M. & Wittenberg, D.F., 1998; Houston, J.C., Joiner, C.L., & Trounce, J.R., 1966; Dr. Stoppard, M., 2005).

What is clotting?

An injury to a blood vessel, such as a cut, triggers a complex chain of events that results in a blood clot. This clotting process is also referred to as blood coagulation. Clotting is the body's response to bleeding and prevents us from losing too much blood. Excessive blood loss is potentially life threatening and may be damaging to the internal organs (www.medic8.com).

What is a clotting factor?

Clotting factors are certain proteins in the blood, that work together with platelets, to help the blood clot. If blood vessels are damaged, clotting factors help the platelets stick together, in order to plug cuts and breaks at the site of the injury. Babies born with haemophilia are missing or have low levels of these proteins, that are required for normal blood coagulation. If blood clotting is particularly slow or ineffective; heavy blood loss can occur, bodily organs and tissues may sustain damage, and in severe cases, this damage may be permanent or result in death (www.medic8.com).

Causes:

Both haemophilia A and B are due to a deficiency of protein (Factor XIII and IX respectively) involved in blood clotting. The faulty gene involved is different in the two disorders but in both cases the abnormal gene is located on the X chromosome. As a result, nearly all haemophiliacs are males, while most females in haemophiliac families are female. Women do not tend to develop the disease because they have two X chromosomes and the normal gene on the other X chromosome, compensates for the abnormal gene. Males who inherit the abnormal gene, do develop the disease because they only have one X chromosome (and one Y chromosome) and therefore do not have a normal copy of the gene to compensate for the defective one. Females only tend to develop the disease if their father is a haemophilia sufferer and their mother is a carrier. Female carriers may pass on the faulty gene to both their male and female children. Each child has a 50% chance of inheriting the faulty gene. In one third of all cases, haemophilia and Christmas disease are a result of a spontaneous gene abnormality and there is no previous family history of the disorders (Collins, J., 2003; Dr. Stoppard, M., 2005).

Symptoms:

The severity of haemophilic symptoms varies, according to the degree of deficiency of anti-haemophilic globulin (i.e.. how much Factor VIII or IX is actually produced) The symptoms generally develop during infancy, when the child begins to crawl and walk. However, children with very mild haemophilia may not have any noticeable symptoms for years. The disorder often only becomes apparent after they experience heavy bleeding, following a dental procedure, accident or injury. Children with mild to moderate haemophilia may not display any signs or symptoms at birth. Males with severe haemophilia may bleed excessively after circumcision.

In children the most common signs are:

- Easy bruising, even after a minor injury. In children that are learning to walk and tend to fall frequently, bruises and bumps generally develop easily.
- Persistent bleeding after cuts and abrasions, or a minor surgical procedure, such as the extraction of teeth. In young children, haemophilia may become evident when they experience heavy bleeding from their gums as they cut their baby teeth.
- Sudden, painful swelling of the soft tissue, muscles and joints, particularly the knee joint, due to internal bleeding.
- Female carriers are generally asymptomatic because they have sufficient clotting factors from their normal gene, that helps prevent any serious bleeding problems.

- In older children and adults, the most common symptoms of haemophilia are:
- Bleeding into the joints (haemarthrosis). This most frequently occurs in the knees, elbows and ankles, but can occur in any joint. Symptoms of haemarthrosis include; tightness and pain in the joint before there are visible signs of bleeding, pain on bending or extending the joint, swelling, the joint may become hot to the touch, and over time all movement to the joint may be lost and pain becomes severe. The bleeding tends to slow after several days when the joint is full of blood. Without treatment, prolonged bleeding into the joints may cause long term damage, and may eventually lead to deformity of the joints.
- Bleeding and bruising in the soft tissue and muscles.
- Bleeding into the mouth due to an injury such as a cut or bite, or the loss of a tooth.
- Unexplained nosebleeds.
- Blood in the urine, due to bleeding in the kidneys or bladder (hematuria).
- Blood in the stool, from bleeding in the intestines or stomach
- One of the most serious complications of haemophilia, is bleeding on the brain. This requires emergency medical intervention and may present with the following symptoms; long-lasting painful headaches, severe vomiting, behavioral changes or sleepiness, a sudden weakness or clumsiness of the leg or arm, stiffness or pain in the neck, double vision, difficulty walking, and convulsions or seizures (Houston, J.C., Joiner, C.L. & Trounce, J.R., 1966; Dr. Stoppard, M., 2005; ww.medic8.com).

Diagnosis:

In making a diagnosis of haemophilia, your doctor will take a personal and family history, perform a physical examination, and arrange for blood tests. Blood tests may include; a complete blood count (CBC), prothrombin time (PT), activated partial thromboplastin time (PTT), factor VIII level, and factor IX. Blood tests are used to establish; how long your child's blood takes to clot and to measure the level of Factor VIII and IX. The tests will show if your child has haemophilia, the type of haemophilia they have, and how severe the condition is. The severity of haemophilia A and B are inversely proportional to the factor levels (XIII or IV) in the blood. When the levels are between 5-30%, mild bleeding occurs. In moderate haemophilia, the factor levels are between 1-5%. Severe haemophilia, with serious symptoms and spontaneous bleeds, occurs when the levels are less than 1% (Coovadia, H.M. & Wittenberg, D.F., 1998; Dr. Stoppard, M., 2005; The Nemours Foundation, 1995-2011).

Treatment:

The aim of treatment for haemophilia is to maintain the clotting factors at high enough levels to prevent bleeding. The frequency and severity of bleeding differs from one boy to another. Some

haemophiliacs only suffer from the occasional episode of minor bleeding, while others, with more severe forms of the condition, experience recurrent internal bleeding. The parents and child must be fully informed about the disease. The child should also wear an identity disc displaying the diagnosis of the disease.

General and conservative measures for haemophilia include; avoiding trauma and IM injections, ensuring pressure bandages and cold packs are on-hand in case of injury, physiotherapy and rehabilitation if necessary, and the application of thrombin locally on affected areas.

Doctors may recommend splinting an affected joint, for a short period of time and to apply ice to the area. This helps to decrease inflammation, promotes clotting and relieves pain. All pain killers containing aspirin or NSAIDS (non-steroidal anti-inflammatory drugs), should be avoided, because they can affect blood platelets and lead to increased bleeding.

In severe cases, bleeding in haemophilia probably needs to be treated with transfusions of plasma or fresh blood, containing the missing factor. Transfusions must be given daily until the bleeding stops and usually have to be repeated for several days.

In milder forms of either condition, haemophiliacs may only require intravenous injections of Factor VIII or IX, to boost the levels of these factors in the blood, following an injury or before surgery.

Factor replacement may be given through an intravenous line, either at the hematology clinic or at home by a visiting nurse or by parents who have undergone special training. Training teaches parents how to prepare the concentrated clotting factor and when and how to inject it into their child's vein.

If your child has severe haemophilia and clotting factors need to be infused 2-3 times per week, a central venous catheter can be surgically placed in their vein, to allow the concentrates of clotting factors to be given frequently without pain.

Joint pain and swelling caused by bleeding into a joint, either as a result of injury or

spontaneously due to the disease itself, can be treated with immobilization, cold compresses, and the correction of the disorder itself. Removal of blood from the joint may help to ease the pain.

In severe cases, prolonged bleeding into the joint may require surgery to remove damaged joint tissue. This procedure is known as synovectomy.

Haemophiliacs must always be admitted to hospital for dental extractions and must be transfused before and after the operation. The socket can be effectively plugged, with a dental splint, that is made before the operation.

Desmopressin, which contains pituitary hormone, may be prescribed to boost levels of Factor VIII. Unfortunately some people develop antibodies to Factor VIII supplements, making treatment particularly difficult. In these cases, immunosuppressant drugs may be prescribed to destroy theses antibodies (Dr. Stoppard, M., 2005; Collins, J., 2003; Coovadia, H.M. & Wittenberg, D.F., 1998; Houston, J.C., Joiner, C.L., & Trounce, J.R., 1966; Hubpages, Inc., 2011; Martin, E.A. (ed.), 1994; The Nemours Foundation, 1995-2011).

How to help your child:

Ensure that your child has a safe environment, whether it be at home, day care, school, or at the babysitter's.

Provide them with a healthy diet, so that they are able to attain and maintain a healthy weight for their size. Excess weight can cause strain on certain areas of the body and increase the risks of bleeding.

Encourage healthy behaviours, to prevent problems associated with haemophilia. For example, exercise strengthens the muscles and helps decrease bleeding from injuries. Swimming is an ideal form of exercise because it exercises all the major muscle groups without putting stress on the joints.

Provide your child with education about their condition, in a way that they are able to understand and reassure them, that their condition is not their fault.

Children with haemophilia generally sense when a bleed has occurred. They often describe a bubbling or tingling sensation in the joint. Encourage your child to tell you immediately if they feel they have had a bleed. Early infusions are essential to preventing long-term damage.

On a practical level, children need protection from things in the home and elsewhere, that may cause injury or bleeding;

Use proper car seats or seat belts when travelling in a car. Protect your toddler with knee pads, elbow pads and protective helmets. All children should wear safety helmets when riding a tricycle or bicycle. Ensure that your child is strapped into a high chair or stroller to prevent falls.

Remove furniture with sharp edges or pad them, especially when your child is a toddler.

If your house is tiled or has hard wood floors, consider installing carpets or purchasing rugs to soften the floor surface.

Remove all small and sharp, or other items that may cause bleeding or hurt your child.

Use electrical outlet covers.

Use baby security gates to prevent your child from going into areas (e.g. stairs) where they may injure themselves.

Check all play equipment and outdoor areas for potential hazards.

Keep cold packs in the freezer, to use as directed in the case of an injury.

Keep a bag packed with essential items, in case your child needs to go to the emergency room.

Ensure that anyone responsible for your child, such as teachers, babysitters, coaches etc, knows that they have haemophilia.

Learn how to examine your child and recognize the signs of bleeding (The Nemours Foundation, 1995-2011www.medic8.com).

Call your doctor immediately if your child has a bleed or injury involving the following:

You suspect any trauma to their central nervous system- head, neck or back

They have sustained an injury to their face, including their eyes or ears.

Their throat or another portion of the airway, has been affected.

Gastrointestinal tract, evinced by bright red or black blood in the stool.

The bladder and urinary tract, which might produce blood in the urine.

The iliopsoas muscle in the trunk. This might produce signs that mimic a hip or abdominal bleed, including pain in the abdomen, groin or upper thigh area, and an inability to raise the leg on the affected side.

The genital area.

Bleeds in the hips or shoulders, can be complicated because they involve rotator joints.

Large muscle compartments, such as the thighs (The Nemours Foundation, 1995-2011).

Prognosis:

Individuals with haemophilia A or Christmas disease are able to lead an active lifestyle but need to avoid sustaining any injuries. Physical activities such as running, walking, golf and swimming are beneficial; but contact sports such as wrestling or rugby should be avoided. Regular dental care is required to avoid bleeding from inflamed gums. Provided that an affected child receives prompt treatment with Factor VIII or IX when bleeding occurs, or has regular infusions, the muscles and joints are less likely to sustain any damage, and their life expectancy should be normal (Collins, J., 2003; Dr. Stoppard, M., 2005).

Prevention:

Women with a family history of haemophilia can be tested to find out if they have the haemophilia gene. If they do, they should obtain medical advice when planning a pregnancy. Genetic counselling is also available to help them assess the risk of having an affected child (Collins, J, 2003).