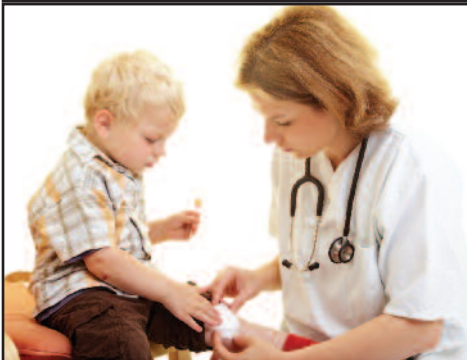


Irish Haemophilia Society



Introduction to Haemophilia



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Introduction

Haemophilia is an inherited bleeding disorder where one of the bloods clotting proteins is absent or present in a reduced amount. A diagnosis of haemophilia in you or your child can be a traumatic experience as your knowledge of haemophilia may be very limited or rooted in a past reality where adequate and safe treatment was not available. The good news is that such fears are unfounded. Current state of the art treatment for haemophilia is available in Ireland. The life expectancy of a child born with haemophilia now in Ireland is essentially normal and their quality of life will be excellent. Haemophilia is a lifelong condition and it is important that you and your child learn about the condition in order to live in a positive manner with the condition. Information and education are vital. We hope this booklet starts you on that road, and be assured that the Irish Haemophilia Society will be there to help you in every step of your journey to a healthy and normal life.

Brian O'Mahony
CEO,
Irish Haemophilia Society

WHAT IS HAEMOPHILIA?

Haemophilia is a genetic blood disorder, which is usually inherited. The gene is passed down from a parent to a child. A person who is born with haemophilia will have it for life. Some of the signs of haemophilia are large bruises, bleeding into muscles and joints, spontaneous bleeding and bleeding for a long time after a cut or surgery.

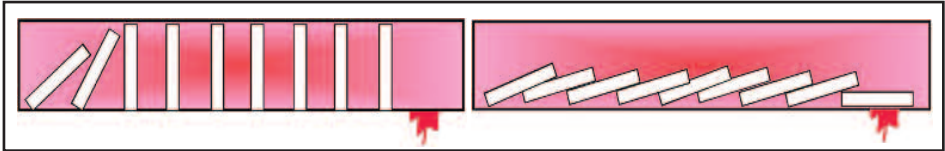


Figure 1: Normal clotting process

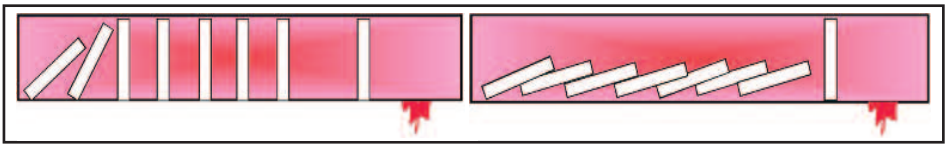


Figure 2: Clotting process in a person with haemophilia where a clotting factor is missing.

The general term haemophilia describes a group of inherited blood disorders in which there is a life long defect in the clotting mechanism of the blood. Blood contains many proteins called clotting factors, and these work to stop bleeding. The lack of clotting factor causes people with haemophilia to bleed for longer periods of time than people whose blood factor levels are normal. However, people with haemophilia do not bleed faster than other people. Most bleeding in haemophilia occurs internally into the joints or muscles. The joints that are most often affected are the knee, ankle and elbow. Repeated bleeding without prompt treatment can damage a joint. The incidence of haemophilia in the general population is 1 in 10,000 (therefore about 1 in 5,000 of the male population has haemophilia).

There are two types of haemophilia. Haemophilia A which is a deficiency in Factor VIII and haemophilia B (or Christmas Disease) is a deficiency in Factor IX. Both types of haemophilia share the same symptoms and inheritance pattern, only blood tests can differentiate which factor is affected. The severity of the condition is related to the degree of deficiency of the relevant clotting factor in the blood.

SEVERITY

The severity of haemophilia is related to the degree of deficiency of the relevant clotting factor in the blood. There are three levels: severe, moderate and mild. A person with less than 1% of normal clotting activity is described as having severe haemophilia. A person with between 1% and 5% of normal clotting activity is described as having moderate haemophilia and a person with over 5% but less than 50% of normal activity is described as having mild haemophilia.

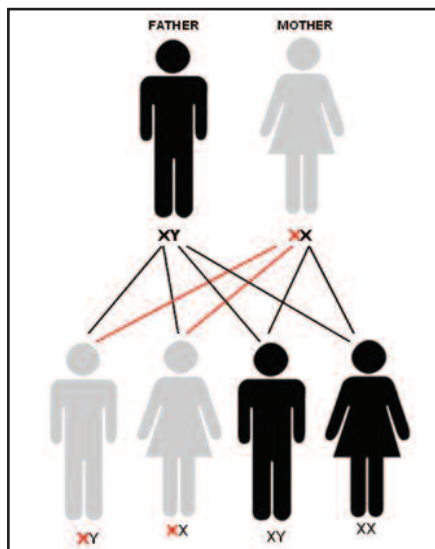


Figure 3: Inheritance pattern when the mother is a carrier of Haemophilia.

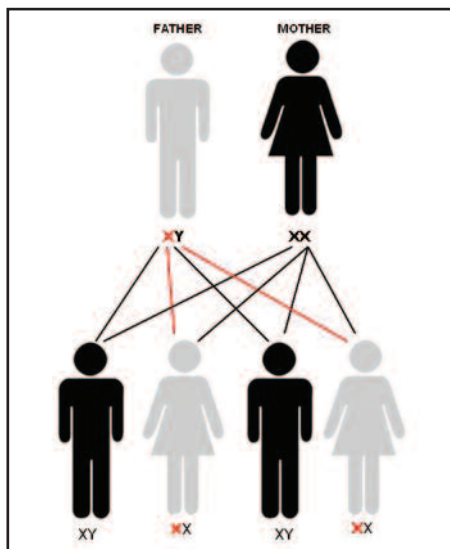


Figure 4: Inheritance pattern when the father has Haemophilia.

INHERITANCE PATTERN

Haemophilia is an inherited condition. However, it is possible for the condition to appear in any family – it is thought that at least 30% of people with haemophilia have no family history of haemophilia. It is difficult to be exact about this because of the way in which haemophilia is inherited. Technically, it is a sex-linked recessive inheritance condition. This means that while only males are affected by the condition, it is passed through the female member of the family.

The sex of a newly conceived baby is determined by the type of chromosomes it receives – one from each parent. A boy inherits his mother's X chromosome and his father's Y chromosome, and a girl has two X chromosomes, one from each parent.

The defect that causes haemophilia resides in the X chromosome. Therefore, all daughters of a man with haemophilia will inherit his X chromosome and will be carriers of the haemophilia gene (obligate carrier). Since the sons of a man with haemophilia must inherit his Y chromosome they will not be affected by haemophilia nor will they be carriers. A carrier female has one normal and one defective X chromosome. If she has a son, the son has a 50:50 chance of receiving his mothers defective X chromosome and therefore has a 50:50 chance of having haemophilia.

The daughter of a carrier also has a 50:50 chance of being a carrier herself.

DIAGNOSIS

A blood sample from the newborn baby can be used to make a diagnosis. This applies to mild, moderate and severe cases. In at least 30% of cases of haemophilia there is no known family history, the occurrence of haemophilia is presumed to be the result of a spontaneous genetic mutation.



The first symptom of haemophilia is usually in the form of extensive bruising as the child learns to crawl or walk. Unfortunately, this is sometimes suspected to be a result of non accidental injury, but increasingly in such cases it is automatic that coagulation tests are used to investigate the possibility of a child having haemophilia or a bleeding disorder.

Moderate and mild haemophilia may not be diagnosed until later in childhood or in some cases even in adulthood. The process of diagnosis involves many complex laboratory tests on blood samples and takes several days to complete. The time around diagnosis can be a difficult period for parents particularly where there is no family history of the condition. It is important that parents become informed about haemophilia and the impact it is likely to have on their child



and other members of the family. The Irish Haemophilia Society is available to offer support to all family members and provides a range of education and support programmes for all age groups.

SIGNS AND SYMPTOMS

CUTS AND SCRATCHES

In most cases minor cuts and scratches do not pose any problems for people with haemophilia. A little pressure is usually enough to stop the bleeding. A person with a bleeding disorder does not cut more easily, bleed more profusely or bleed faster than normal. They simply bleed for longer.



JOINTS AND MUSCLES

For those severely affected, a major problem can be internal bleeding into joints, muscles and soft tissues. All of us damage our muscles in small ways in the activities of everyday life. Most people repair that damage automatically. For the person with a severe bleeding disorder however, the tiny breaks in the blood vessels in joints and muscles may continue to bleed as a result of normal everyday activity. These bleeds are sometimes described as “spontaneous” because it is impossible to identify a cause.

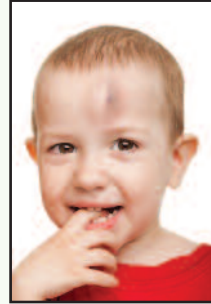
An ache or irritation in an affected area is usually an indication that a person with haemophilia is getting a bleed. If left untreated pain may become excruciating. In the case of joint bleeding, the blood which has escaped into the joint has a very damaging effect on the surface of that joint. Once a joint becomes damaged then bleeding will occur more frequently resulting in a “target joint”. The majority of bleeds into joints and muscles occur in the lower limbs, with ankles and knees being the worst affected in most people.

SOFT TISSUE BRUISES

Soft tissue bruises will always occur in people with bleeding disorders. Although these may look serious they usually do not require any treatment. Sometimes if the bruise is increasing in size and causing pain, then treatment with factor may be recommended.

HEAD, FACE AND NECK INJURIES

After a blow to the head, face or neck, sufficient factor must be given immediately to bring the factor level to 100%. These injuries must be assessed at a treatment centre. If the injury is significant enough, a person with a bleeding disorder may need to be admitted to hospital and may require a CT scan to ascertain the extent of bleeding, if any.

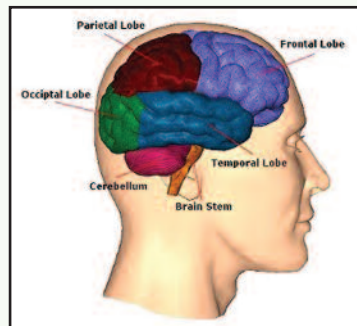


MINOR HEAD INJURIES

These are injuries that can lead to bruising or even small cuts on the head. These injuries should always be treated either at home or at the treatment centre.

SERIOUS HEAD INJURIES

These result from a severe bump on the head. A head injury is always serious if the person is knocked unconscious. These injuries should be treated as quickly as possible and the patient must be taken to the nearest haemophilia centre.



OTHER SERIOUS BLEEDING EPISODES

- Vomiting blood
- Coughing up blood
- Passing blood in bowel motions (stools will be black in colour)



TREATMENT

Haemophilia is treated by replacing the missing clotting factor in the blood through an intravenous infusion of clotting factor concentrate. Each bleeding episode must be promptly treated. Once the bleeding stops, pain rapidly diminishes and use of the limb returns. The clotting factor concentrate is manufactured as a white powder and is reconstituted with the sterile water provided with the factor concentrate. The Factor VIII and Factor IX used for the treatment of haemophilia is called “recombinant factor”.

What cannot be emphasised enough is that a person with haemophilia must have treatment as soon as a bleed starts. It prevents further bleeding, pain and most importantly, reduces the likelihood of permanent damage to joints (target joints).



SURGERY

All surgical procedures, including dental extractions and fillings, will require treatment beforehand and should be organised through your Haemophilia Treatment Centre.

HOME TREATMENT

Both preventive and on demand treatment can be administered at home. Home treatment is the ideal method of treatment from a medical viewpoint as a minimum amount of time is lost between the recognition of a bleed and treatment. This has many advantages, it reduces the disruption caused by a bleeding episode to the person with haemophilia and his family and the patient feels more able to control his condition. The benefits of self infusion at home not only include increased independence and the bonus of not having to travel to the hospital for treatment, but school and work attendance is more regular. If bleeds are treated promptly, the period of incapacity caused by each episode can be reduced. In adults and teenagers home treatment is usually carried out by the affected person. From a young age children will be taught how to self infuse. Alternatively, a device called a Port-a-Cath (Freddie) can be used to facilitate venous access until self infusion using the veins is practical.

PROPHYLAXIS

It is recommended that all children with severe factor deficiencies should be commenced on a programme of factor concentrate prophylaxis. Prophylaxis involves small regular (2-3 per week) infusions of factor concentrate to prevent spontaneous bleeding and to minimise traumatic bleeding. This treatment regime, although it can be time consuming and at times difficult to learn, will prevent joint damage and lead to an improved quality of life.

PORT-A-CATHS

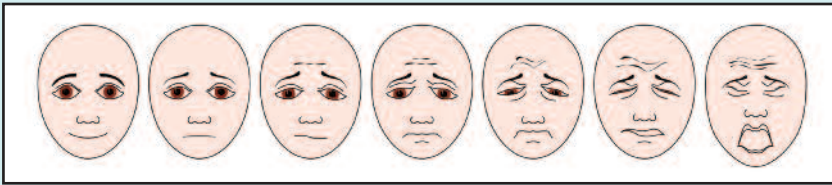
In some young children where venous access is difficult and injections are stressful for both the child and the parent it is recommended that a Port-a-Cath be fitted. Port-a-Caths are used to provide access to veins and are inserted under general anaesthetic. The Port-a-Cath is inserted under the skin usually in the chest area. When the Port has been inserted there is no longer a need for injections into veins so treatment is painless and therefore less stressful.



INHIBITORS TO FACTOR CONCENTRATE

Some people with severe haemophilia develop an antibody (inhibitor) to factor concentrates. The effect of this is that when factor is administered there is no clinical response because the activity is immediately neutralised. Often the first sign that a person has inhibitors is that the treatment does not appear to be working.

People who have inhibitors to Factor VIII and Factor IX are generally treated with a recombinant factor called Factor VIIa (Novoseven). This is an activated factor, which bypasses the need for Factor VIII and Factor IX in the clotting cascade. It is given by intravenous injection but only gives a very short-term response, therefore needs to be given every 2-3 hours. Eradicating Factor VIII or Factor IX inhibitors involves highly specialised and intensive treatment regimes, which may not always be successful. It is good practice to screen small children with haemophilia regularly for inhibitors.



PAIN MANAGEMENT

It is important to treat bleeding episodes without delay as this will minimise the pain caused by internal bleeding into joints or muscles. Before prompt and adequate treatment became available in the 1970's pain was a regular feature of life for people with severe haemophilia. Untreated bleeds resulted in excruciating pain and arthritic deterioration. Long term arthritic deterioration can lead to constant pain in a damaged joint. Many people with haemophilia still have constant pain as a result of arthritic damage to their joints. You should speak to your haematologist or GP in relation to what medications should be taken.

Remember: People with bleeding disorders should not take Aspirin in any form as this can aggravate bleeding.



SAFETY ISSUES

In the 1970's, factor concentrates transformed the lives of people with haemophilia. It meant that people could be active, treat themselves at home and not suffer acute pain in their joints. However the factor concentrates used at the time were made from pooled plasma, and many people who were treated with factor concentrates during the 1970's and 1980's were exposed to Hepatitis C and HIV.

Hepatitis C and HIV gave rise to many new fears and problems just at a time when life for people with haemophilia appeared to be better than it had been in the days before effective treatment was available. It was essential for people who were infected, and their families, to attend counselling. The Irish Haemophilia Society responded by introducing a range of support programmes to meet the needs of these members.

Currently, recombinant (synthetic) factor replacement therapy is used to treat Factor VIII and Factor IX deficiency. All other factor deficiencies are treated with products which are made from human donated plasma. Von Willebrands Disease is treated with a synthetic substance called DDAVP or with a human plasma derived Factor Concentrate.

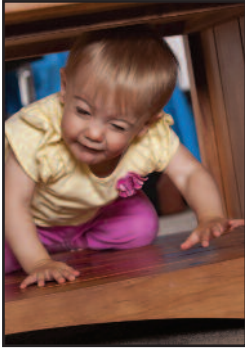
Nowadays, blood products are safer than ever, as they are extensively screened for viruses and treated to inactivate any viruses which may be present. Patients are also offered a vaccination programme against Hepatitis A and Hepatitis B. However, vigilance must be maintained. The Irish Haemophilia Society now has a formal role in the selection and tender process for all treatment products purchased in Ireland for haemophilia, von Willebrands Disease and other bleeding disorders. Two representatives of the Irish Haemophilia Society are members of the Haemophilia Product Selection Monitoring & Advisory Board (HPSMAB). The HPSMAB formally selects the products used for the treatment of Haemophilia, von Willebrand Disease and other bleeding disorders from all the products available. The group also monitors developments in relation to these products on an ongoing basis. The Society has an independent expert advisor who attends meetings of the HPMSAB as necessary and who advises the Society in relation to their work on this committee.



YOUR CHILD WITH HAEMOPHILIA

When a child is diagnosed with haemophilia or a related bleeding disorder, the parents may have feelings of worry, disappointment and perhaps guilt. These emotions are temporary. This may be an especially hard time for those who have no family history of the condition. Some people talk about things while others suppress their worries. Parents and other members of the extended family need to have the reality of haemophilia carefully, patiently and repeatedly explained to them by members of the comprehensive haemophilia care team. After a while, parents may even find that it is not nearly as bad as they first imagined. The unknown is often more frightening than the reality.

The impact of factor concentrate replacement therapy on the lives of people with haemophilia needs to be explained. Likewise when a Port-a-Cath is recommended because of difficulty with venous access, the surgical procedure needs to be fully explained to the parents. It is important to remember that with good treatment the child with haemophilia has every chance of growing up as an active, fit child who can participate fully in family, school and later working life. Because haemophilia is rare, parents may feel isolated and alone and it is very helpful to be put in touch with others in a similar position.



THE EARLY DAYS

A big worry for parents of a baby with haemophilia is how they can recognise when their child has a bleed. An infant or young child with a bleed may appear to be irritable for no apparent reason. He may avoid using the affected limb. The area may feel warm or swollen. His limbs may look uneven. If in doubt you should call your haemophilia treatment centre for advice. With experience and a little vigilance parents will soon learn to recognise the signs and severity of bleeding episodes and learn to respond effectively.



SOCIAL SKILLS

It is important for the child and for the other members of the family that haemophilia does not dominate the life of the family. A child with haemophilia has to learn to become independent in the same way as any other child. While it is natural for parents to want to offer as much protection as possible, over protection is not in anyone's interest. Obviously at the toddler stage, when all children are liable to cut, bump and bruise themselves, careful observation of the child is important. However, as children get older they can become more involved in self care and the recognition of bleeds. Young children with bleeding disorders learn very quickly to recognise when a bleeding episode is starting in a joint or muscle. They should be encouraged to tell a parent as soon as possible so that treatment can be given and disruption to normal life reduced to a minimum.

EDUCATION

Academically there is no difference between a child with haemophilia or a related bleeding disorder and an unaffected child. Most severely affected children receive preventative treatment 2-3 times weekly at home which prevents most bleeding episodes. This is known as prophylaxis and requires intravenous (i.v.) injections given by the parents or the child themselves. It is important for them to attend school as much as possible. However, sometimes they may miss school when they are recovering from a bleed. Teachers should do their best to make them comfortable when they come back, and to help them catch up on work they have missed.



PLAYGROUP AND PRIMARY SCHOOL

At playgroup, normal play activities present little in the way of problems and the child should be allowed to play alongside the other children. Primary school level sport is usually less competitive and children with haemophilia should be allowed to take part in all activities unless they have specific problems.

SECONDARY SCHOOL

In secondary school sport often becomes more competitive and injuries are more common. Sports with a high level of physical contact such as rugby or boxing are not recommended. Sports booklets are also available for more details.

(For further information on this please see our publication “Information for Teachers and Playgroup Leaders”)

SPORTS

Physical activity is an important part of a healthy life. Participation in sports, games and exercise is a fun way to improve the quality of our lives – physically, mentally and emotionally. This does not change because you have haemophilia. In fact, physical activity may be even more important to someone with haemophilia, since it helps to develop strong bones and muscles that protect the joints from the traumas of daily living.

It is vital that anyone with haemophilia discuss plans for physical activity with their haematologist or physiotherapist, who is familiar with their individual situation. Only they can properly advise and recommend appropriate physical activities.



COLLEGE / EMPLOYMENT

On a programme of prophylaxis, a child with haemophilia should not miss out on schooling or education as a result of bleeds or joint damage. The choice of college education and employment, therefore, for someone with a bleeding disorder should not differ from any of his peers.

In each case, the choice of occupation is related firstly to the interests, abilities and skills of the individual and secondly to the severity of haemophilia.

It must be emphasised that the majority of people with haemophilia find and keep satisfactory regular employment.

Home treatment has enabled the person with haemophilia to also self infuse factor concentrates at his place of employment, if required, which ensures normal work attendance and promotes self confidence with independence.

CARRIERS

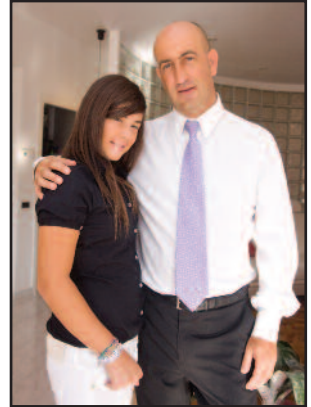
CARRIER DETECTION

The inheritance pattern of haemophilia is a difficult one to understand at first. It is very important to understand it properly so that families who know there is a history of haemophilia can get accurate advice about their own situation.

OBLIGATE CARRIERS

(The following are definitely carriers!)

- A daughter of a man with haemophilia
- A mother with more than one son with haemophilia
- A mother with one son with haemophilia and a blood relative with haemophilia



POSSIBLE CARRIERS

When a man has haemophilia the following family members should be tested for their carrier status:

- Mother
- Sisters
- Granddaughters
- Cousins
- Nieces
- Aunts



It is important for carriers to be aware of their factor levels, as treatment may be necessary at times of severe injury or surgery. When a haemophilia carrier or potential carrier is pregnant, she should inform her obstetrician of her carrier status. It should not be assumed that a young girl who has grown up with a father or brother with haemophilia is aware of her possible carrier status. A young girl needs to be given information relevant to her age and using language that she can understand. This information needs to be given on an ongoing basis as she matures to help her understand it at her level. When the time comes for a young girl to be tested for her carrier status (usually at the age of 16) she will need to be given extra support.



GENETIC COUNSELLING

For carriers or possible carriers it is advisable to undergo genetic counselling preferably before becoming pregnant. Genetic Counselling will provide information to enable a woman to make an informed decision in relation to family planning. Genetic counselling and carrier testing is available at the National Centre for Hereditary Coagulation Disorders in St. James's Hospital, Dublin. Tel: 01 416 2141.



WHAT THE IRISH HAEMOPHILIA SOCIETY OFFERS

The Irish Haemophilia Society was founded in 1968 to provide support and advice for members and to improve the quality of life for people with haemophilia.

The Irish Haemophilia Society offers education and support to all members living with haemophilia, von Willebrands Disease and related bleeding disorders. From the time of diagnosis there are many issues facing people with haemophilia. Support is also offered to their parents, families and friends on issues such as how to recognise a bleed, treatment and support services. Coming to terms with a diagnosis can

be very difficult especially when there is no history of haemophilia in the family. Through our educational programmes, AGM & Conference, Parents' Conference, Members Conference, Information Meetings and Regional Visits, we give our members as much information as possible. We also provide publications and up to date information on our website. We constantly endeavour to help members with any queries or questions they may have.

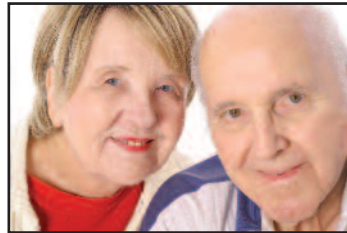


The following are a list of educational meetings and conferences that the IHS hold:

- AGM & Conference
- Parents' Conference
- Members' Conference
- Relatives Days
- Regional Meetings
- Treatment Updates
- Conference for Young Men
- Women and Bleeding Disorders Information Days
- Peer Group Meetings
- Conference for members with HIV and/or Hepatitis C

We also provide support and services to specific categories of members:

- Adults with haemophilia
- Ageing adults with haemophilia
- Children with haemophilia and their families
- Families with a new diagnosis of haemophilia
- Persons with haemophilia who have been affected with HIV and / or Hepatitis C
- The families of those who have died of either or both viruses
- Carriers of haemophilia
- Persons with von Willebrands Disease
- Women with Bleeding Disorders
- Individuals with rare bleeding disorders such as Factor VII, Factor X and Factor XIII deficiency
- Persons with haemophilia and their families who have moved to Ireland from abroad



The Society employs a Nurse/Counsellor who meets with members on a one to one basis to help them deal with all the issues associated with haemophilia. The Counsellor also meets with partners and other family members and provides Psychosocial and Practical Support especially in times of a crisis and when a member is ill. The Counsellor also facilitates access for members to specialised external counselling, as required. The Counsellor meets members to assist and advise on all aspects of living with a bleeding disorder including assistance for family matters and relationship issues.



The Irish Haemophilia Society also provides:

- Assistance for members who are ill at home including liaison and advocacy with the health services on their behalf.
- Assistance for parents in dealing with a new diagnosis of haemophilia through education, information, meetings and home visits where required.
- Assisting members in dealing with medical personnel and attending meetings with members if requested.
- Information talks to the staff at schools or crèches at the request of the parents of the child with haemophilia who is attending the school or crèche.
- Provision of advice and support to young persons with a bleeding disorder on matters such as inheritance of haemophilia, treatment and education. Specific programmes for young people are an integral part of our annual meeting and family conferences.
- Individual financial assistance when necessary including facilitating access for members to external financial advice.
- Provision of advice and information on benefits.

The ongoing level of personal contact between staff and members is very high. Each member of staff is trained and educated sufficiently about haemophilia and the ramifications of living with a bleeding disorder to enable them to proactively engage with and assist members.

ADVOCACY

The Society advocates for optimum provision of healthcare for people with haemophilia, von Willebrands Disease and related bleeding disorders. We provide information, education materials and advocacy to any person with a bleeding disorder regardless of whether they are a member of the Irish Haemophilia Society, or not. Our goal is to have an educated and informed population of parents and people with haemophilia who are well equipped to deal with this life long condition.

We make recommendations to the Department of Health & Children, the Health Service Executive and the Health Care System through the formal statutory bodies, the National Haemophilia Council and the Consultative Council on Hepatitis C. We are involved in the selection of factor replacement therapy for haemophilia through our role on the Haemophilia Product Selection and Monitoring Advisory Board (HPSMAB). We are involved in the development of policy for haemophilia, on a European and Global level, through our activities on the European Haemophilia Consortium and the World Federation of Hemophilia. This external work also ensures that we are up to date on key developments in relation to haemophilia care worldwide.

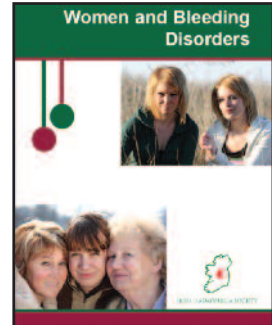
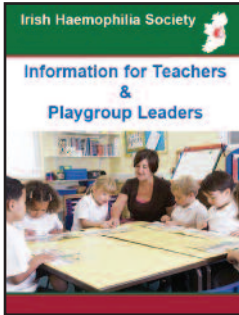
We are involved in the provision of services for persons with disability through our membership with the Disability Federation of Ireland.

We work in close co-operation with the haemophilia care teams at the Comprehensive Haemophilia Treatment Centres in St. James's Hospital, Our Lady's Children's Hospital Crumlin, Dublin and Cork University Hospital. We meet regularly with the haemophilia care teams in the treatment centres to discuss issues of mutual concern and to ensure that we can work in a co-operative and mutually beneficial way in providing the best treatment, services and support for persons with haemophilia and related bleeding disorders.



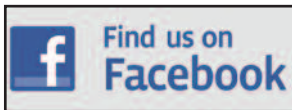
EDUCATIONAL GRANTS AND ASSISTANCE

The Irish Haemophilia Society provides a number of educational grants to assist persons with haemophilia or related bleeding disorders, to avail of third level education. Grants are also available to relatives of a person with haemophilia or related bleeding disorder who is attending college. Grants are awarded following an application, review and selection process including a rigorous scoring system.



PUBLICATIONS

Four magazines are published each year with reports on what is happening locally and in the wider world of haemophilia. The Society produces an annual report outlining the activities and achievements for the year. Information booklets, leaflets, and fact-sheets on specific topics are produced on a regular basis when required.



WEBSITE

The Irish Haemophilia Society website, www.haemophilia.ie is updated regularly with information on haemophilia and related bleeding disorders. In addition, relevant media articles are uploaded onto the site on a daily basis. The website content includes:

- Sections for Parents, Young Adults and Children
- Online registrations, donations and membership renewal
- Educational material from the hospitals/treatment centres
- Fundraising
- Entitlements & Benefits
- Reports from events
- Treatment Centre Audits
- A Discussion Forum & Facebook page



MEMBERSHIP

ORDINARY MEMBERS

Ordinary members pay an annual fee of €30 and have voting rights. Ordinary members must be 18 years or over.

ASSOCIATE MEMBERS

Associate members are included on the mailing list but do not have a vote. This category can include other national haemophilia organisations, healthcare workers and service providers.

LIFE MEMBERS

These are voting members. They pay a one off fee of €650.

HONORARY LIFE MEMBERS

Members who have 30 years continuous membership and who have attained the age of 65 may be given Honorary Life Membership at the discretion of the Board.

Irish Haemophilia Society
First Floor, Cathedral Court,
New Street,
Dublin 8, Ireland.
Tel: 00 353 1 6579900
Fax: 00 353 1 6579901
Email: info@haemophilia.ie
Website: www.haemophilia.ie