SALFORD AREA HEALTH AUTHORITY (TEACHING)

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Introduction to Haemophilia

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INTRODUCTION TO HAEMOPHILIA

In a normal human being at least 12 factors are needed for blood to clot. In haemophilia one of these factors is missing. The blood clots slowly, the clot is very unstable and jelly-like. It will break down causing the wound or blood vessels to bleed.

Haemophilia is divided into two main categories:

Haemophilia 'A' in which Factor VIII is missing or reduced.

Haemophilia 'B' or Christmas Disease in which Factor IX is missing or reduced.

Haemophilia is a sex linked disease.

INHERITANCE

There are 46 chromosomes to the body cell. On each chromosome are fixed the genes.

44 chromosomes and their genes are the same for both sexes, but the sex chromosomes are different. Females have 2 X chromosomes, males have an X and a Y. The Y is smaller and lacks some genetic material, which includes factors VIII and IX.

Females are therefore XX and Males are XY.

Genes carry all the different messages necessary to make a human being. They contain the D.N.A. (Desoxyribo-Nucleic Acid) which holds all the relevant information and instruction on the formation of protein from which the body and blood is made.

Sometimes these messages get confused and mixed up which is hardly surprising as there are millions of genes with millions of messages. When the information passed on by a gene becomes confused, the protein laid down by this confused message must be faulty and will not be able to function properly in the body. In fact, the instructions may be so mixed up that no protein at all is produced.

A genetic change in which a piece of this D.N.A. is altered or lost is called a mutation. Once a mutation has occurred it is permanent — the misprinted gene is fixed to its chromosome and copied every time the cell divides. The misprint may appear in the sperm or egg to be passed to the next generation.

About 70% of people with haemophilia have a family history, 30% are recent mutations.

In haemophilia 'A' the mutation affects the gene governing the production of the blood clotting F VIII and in haemophilia 'B' the mutation affects the gene governing the production of the blood clotting F IX and either:

(a) No F.VIII or F.IX is produced.

(b) An abnormal F.VIII or F.IX is produced, which will not function correctly.

(c) Only a small amount of F.VIII or F.IX is produced.

As a result, the severity of the disease varies from case to case.

The abnormal gene is on the X chromosome. Females who are the carriers of this disease have one normal X and one abnormal X. They may show very little or no sign of abnormality in their blood clotting mechanism because their sound X chromosome carries a normal gene.

The males are affected because they have only one X chromosome, which is an abnormal one. Their other sex chromosome is a Y which is too small to carry the F.VIII gene and cannot make up for the deficiency as the female's other X does.

SCHOOL AND HAEMOPHILIA

Public opinion is still based on presumptions and poor information, and so it is not unusual to find haemophiliacs viewed with suspicion as individuals who, when cut, will die in a pool of blood.

This obviously is far from the truth and it is not necessary for the haemophilic child to be singled out in school for special attention.

However, it is perhaps wise not to allow him to participate in any competitive sport which carries a risk of physical contact and injury, such as football or rugby. Swimming, however, is beneficial and should be encouraged.

School life contains an element of stress — starting school, changing school and last years at school. A period of difficult behaviour or emotional unsettlement may be shown at home or school.

One of the main problems is loss of school work due to frequent visits to hospital, in-patient treatment or a period or rest at home following severe bleeding episodes. Perhaps the school teacher could help and give support by arranging current work to be sent to the child at home or even visiting the home.

We like to keep our haemophilic children at normal schools as they have a wider curriculum and higher standards of attainment than at a special school. Also socially, normal schools offer the child a better prospect of learning how to adjust to life in the community.

TREATMENT

Treatment consists of one or more injections of the deficient clotting factor which is given intravenously to raise the blood level so that the blood will clot normally. This arrests the bleeding. The injection may sometimes need to be repeated. It is also necessary to provide general support, e.g. bandages or splints for muscles and joints, bed rest, physiotherapy, etc. while the injured part recovers.

BLEEDING EPISODES

Surface cuts: Cuts serious enough to need stitches will have to go to hospital any way. In haemophilia they will also need injections to encourage proper healing. Small cuts and abrasions, however, will heal normally.

Mouth bleeds: Injury to the mouth, e.g. on lollipop sticks, sharp pencils, bitten tongues, loose teeth, etc. These can ooze for a long time and if not stopped can cause quite a loss of blood. These need treatment.

Nose bleeds: Note which nostril and tell the doctor. Need treatment if cannot be stopped by usual pressure.

Eye bleeds: of any sort must be reported always.

Internal bleeding: Coughing blood is rare. So is bleeding into the bowel — sticky tarry black stools, abdominal pain. Blood in the water or haematuria can occur without any reason in the haemophiliac — bear in mind a tiny quantity of blood stains a lot of urine red and so haematuria looks far more frightening than it really is. Nevertheless, it must be reported and treated.

Closed bleeds: These are bleeds into soft tissue, joints and bones, muscles and into the skull.

Joint bleeds: Joints mainly affected are elbows, knees, ankles, and shoulders, but other joints in the body can also be affected. Joint bleeds are variable, they may grow very gradually without pain — the joint becomes full but not tense, or they may start suddenly, the joint filling rapidly with blood and becoming tense, hot and painful.

These bleeds need treatment which is the replacing of the missing factor.

Before going for treatment a crepe bandage should be firmly wrapped round the affected joint, this will help the pressure of the tissues to contain the bleed.

Bleeds must be reported during early stages. If not, the more blood that is allowed to enter a joint space, the more damage to the surrounding tissues. The period of recovery is prolonged and more treatment than normal is needed.

Children must be encouraged to report bleeds as soon as one is recognised.

Muscle bleeds: Bleeds in the arm and leg muscles need treatment as soon as possible. The affected muscle becomes hot, swollen and tense. The child holds the limb in odd positions so as to rest the affected muscle.

Prompt treatment is important because muscle fibres can be killed and, if a bleed is allowed to progress over a long period, joints worked by the muscle will be affected, leading to contractures and deformities.

Head Injuries: e.g. Bad knocks on the head, a heavy fall, or injuries to the face and skull.

These knocks may cause internal haemorrhage in the skull which will continue to spread and as the bleed grows it will begin to compress the brain. There are several warning signs which include:

(a) intense headache.

(b) irritability.

(c) dislike of bright lights.

(d) nausea and vomiting.

(e) drowsiness.

If a cerebral haemorrhage is suspected, urgent hospital admission and factor replacement are necessary.

AFTER TREATMENT

If a joint needs a period of rest, which most do after a bleed, children may need to come to school in a wheelchair or on crutches, with an arm in a sling or with a plaster of Paris back slab, or just a bandage. If this is the case keep the child indoors away from the playground until the bleed has settled and the joint is normal again. But do give him encouragement and do not be frightened of these appliances. To the child and his parents they become second nature. Some children may need long-term orthopaedic supports such as leg irons or polythene splints.

HOME TREATMENT

Some patients are on home treatment. This means that the injection is given at home by the parent or the child himself. We hope in time to have all our severe cases on home treatment. Perhaps some older boys who have learnt to give their own injections may be allowed to give their treatment at school?

Prophylaxis: Some children may be able to have an injection to provide cover for a day or so for an important examination, or to tide them over a period when bleeds are particularly troublesome.

DENTAL CARE

Care of the teeth is important because if the child has dental decay, an abscess or toothache may develop and he will have to be admitted into hospital for extraction. The child should be discouraged from eating sugar-containing foods and drinks between meals, as this causes dental decay; he should also be discouraged from "wobbling" loose milk teeth because bleeding may occur. Regular visits to the dentist are essential in order to maintain dental health.

OTHER RARE BLEEDING DISEASES

In the foregoing we have described what happens in classical haemophilia and Christmas Disease. There are other, even more rare, congenital bleeding diseases. The commonest of these is von Willebrand's disease. It has a different pattern of inheritance, as it affects either sex and is passed on directly from an affected parent. Patients have a low factor VIII concentration, as in haemophilia, but the mechanism is different. However, treatment is similar and patients are given cryoprecipitate or factor VIII concentrate. In these rare disorders, the severity is variable but the general management of illness is the same as for haemophilia.

GENETIC PATTERNS

NORMAL	CARRIER	HAEMOPHILIAC
Mother 1, 2 X X — Father 3, 4 X Y	Mother 1, 2 *X X — Father 3, 4 X Y	Mother 1, 2 X X — Father 3, 4 *X Y
Possible Combinations	**Possible Combinations**	**Possible Combinations**
1, 3 — Female Child X X	1, 3 — Female Carrier *X X	1, 3 — Female Carrier X X*
1, 4 — Male Child X X	1, 4 — Male Affected *X Y	1, 4 — Male Normal X Y
2, 3 — Female Child X X	2, 3 — Female Normal X X	2, 3 — Female Carrier X X*
2, 4 — Male Child X Y	2, 4 — Male Normal X Y	2, 4 — Male Normal X Y
	1 in 4 chance of affected boy 1 in 4 chance of carrier girl 50/50 Chance of normal child	All boys will be normal All girls will be carriers

*— Indicates a haemophilic chromosome