Haemophilia

You have just been diagnosed with haemophilia.
Haemophilia is a hereditary genetic disorder affecting blood clotting.
People suffering from haemophilia do not produce enough

- factor VIII, a blood factor necessary to normal clotting, in the case of haemophilia A
- clotting factor IX, in the case of haemophilia B (or Christmas disease after the name of the first patient studied with the disease)

In both Haemophilia A or B, the clot is more fragile and has difficulty to form, inducing a risk of bleeding (haemorrhage).

Medecin's notes :

In haemophiliacs the genes responsible for the coagulation factors have mutated onto the FVIII gene for haemophilia A or on the FIX gene for haemophilia B. The haemophilia genes are on the X chromosome.
Females have two X chromosomes (=XX), whereas males have one X chromosome and one Y chromosome (=XY).
Transmission of the mutated gene may run in a family, but the mutation may also occur in people with healthy parents (spontaneous mutation).

- If a woman carries the gene for haemophilia, she is said to be a 'healthy carrier' (i.e. she does not have the disease even though she carries the mutated gene), because her second X chromosome 'makes up' for the mutation on the first one. Some of these carriers, however, may have lower rates of FVIII or FIX.
- If a man carries the gene for haemophilia, he is haemophiliac because his Y chromosome cannot 'make up' for the mutation on his single X chromosome.

Both sick people and healthy carriers may pass on the mutated gene to their children.
Haemophilia effects almost exclusively males with frequency at birth of 1/5,000 boys for haemophilia A and 1/30,000 for haemophilia B. There are about 5,000 haemophiliacs in France.

Symptoms of the disease

Severity differs from one person to another depending on how deficient they are in clotting factors. There are three forms of haemophilia:

- a severe form, with a coagulation factor rate lower than 1%;
- a moderate form, with a coagulation factor rate between 1% and 5%;
- a weak or attenuated form, with a coagulation factor rate between 6% and 30%.

The risk of bleeding depends on the severity of the disease. If your levels of the deficient clotting factor are very low, you may experience spontaneous bleeding, but if your levels of the deficient clotting factor are slightly to moderately low, you may bleed only after surgery or trauma. Bleeding may affect any organ: skin injuries, bleeding under the skin or into a muscle (haematoma), into a joint (haemarthrosis), sometimes into an internal organ or into the brain.
Haematomas and haemarthrosis are the most commonly reported events. If untreated they may spread and cause compression. They result in :

- pains;
- stiff movements if the haematoma affects a limb;
- vessel or nerve compression;
- a drop (sometimes sharp) of the red blood cell count (anaemia).

Bleeding may become really serious if it affects sensitive areas of the body, such as the brain.

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Haemophilia (suite)

The follow-up network

The 'France-Coag' network registers all haemophiliacs living in France and allows to follow their treatments and health problems. The data are anonymous and meet the CNIL criteria for confidentiality and freedom. The network is ruled over by Institut de Veille Sanitaire (InVS), a NICE equivalent, and works with the different CRTH doctors and patient representatives.

Your CRTH doctor will ask you to register with this network. You can refuse to register, but can also ask to register if you think you have been left out.

School-age children

Any haemophiliac child going to school must benefit from an 'individualised project' tailored to their case and designed by the school doctor advised by the family doctor.

Your treatment may induce adverse effects and carry risks. Your doctor will keep you informed and tell you what symptoms to watch out for before you start on the suggested treatment.

Useful contacts:

- **Secretarial / appointment:**
- **Nursing consultation:**
- **Consulting psychologist:**
- **Social worker:**
- **In an emergency:**

Diagnosis

Diagnosis rests on blood clotting tests made from a simple blood sample.
Antenatal diagnosis is also possible after screening the foetus for the mutated genes.

Management

Each region of France has a local haemophilia management centre (Centre Régional de Traitement de l'Hémophilie, CRTH) with health professionals specialised in haemophilia (doctors, surgeons, physiotherapists, nurses, etc). They ensure patient management and education and organise local care.
Bleeding management, whether curative or preventive, consists in infusing the deficient clotting factor intravenously. The dosage depends on the location and severity of the bleeding.
In some cases, because of the presence of a coagulation-inhibiting antibody, special drugs are used instead of the haemophilia factor concentrates and specialist advice is required before any therapeutic treatment.
In case of established haemarthrosis, pain management is essential and the joint must be immobilised.
For many patients haemarthrosis prevention consists in long-term prophylaxis (with regular systematic injections of the deficient clotting factor) from infancy, according to medical prescriptions. Physical exercise and physiotherapy allow to preserve both muscle tone and joint movement in case of damage due to repeated bleeding. Strenuous or dangerous efforts, at work or for recreation (especially sports), are better avoided.

Complication prevention

- Regular medical supervision, coordinated by the Centre Régional de Traitement de l'Hémophilie, in relation with the family doctor, is recommended.
- Haemophiliac patients must never take any painkiller with aspirin or non-steroid anti-inflammatory drugs (NSAIDs), as these medicines increase the risk of bleeding by interfering with coagulation. Neither should they get any intramuscular injection.
- Patients must always carry their haemophilia card, their latest prescription and their follow-up document and systematically show them on any medical appointment or in case of an accident.
- In case of surgery or any invasive event (suture, tooth extraction…) coagulation must be corrected with some initial treatment. Therefore it is important to get the CRTH doctor's advice before any such procedure. Both the anaesthetist and the surgeon must be informed of the bleeding risk, even in case of a slight deficiency in FVIII or FIX, and even if bleeding is scarce.

Patients association → Association Française des Hémophiles (AFH)
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