

## CARRYING THE HAEMOPHILIA GENE: SPECIAL ISSUES FOR GIRLS AND WOMEN

### CLOTTING FACTOR LEVELS AND SYMPTOMS

Many girls or women who carry the gene causing haemophilia do not have signs or symptoms of a bleeding disorder. If at least one of their X chromosomes has a factor VIII or IX gene that works, their body can usually produce normal or near normal levels of factor and they do not have bleeding problems.

However, some girls or women who carry the haemophilia gene may have a bleeding tendency. Females are often described as “symptomatic carriers”. If their factor levels fall in the range for mild haemophilia (5 – 40% of normal clotting factor), they may sometimes also be referred to as having “mild haemophilia”.

Examples of having a bleeding tendency may include:

- Bruising easily
- Having heavy menstrual bleeding
- Having excessive bleeding after dental surgery or extractions, other surgery or accidents
- Have prolonged bleeding after childbirth.

All females who carry the gene should have testing for their clotting factor levels periodically, as their factor levels may change with age, pregnancy and hormonal medications. If their factor level is low, they will need a treatment plan to manage situations if they occur or prevent them.

*“Heavy periods aren’t normal, and can make life pretty hard. It’s OK (and even good!) to talk about it and share information with doctors and others, to find out more, to understand the options that you have, and to support others by sharing your story about being a carrier.”*

## MANAGING SYMPTOMS

Heavy bleeding with menstrual periods (menorrhagia) may be a symptom of carrying the haemophilia gene and can involve:

- Heavy menstrual periods (e.g., soaking through a tampon and pad around two hourly, or needing to change during the night)
- Menstrual bleeding for longer than normal (e.g., longer than 8 days)
- Bleeding with clots bigger than a 50 cent piece in size.

Heavy menstrual bleeding can lead to anaemia (low red blood cell count/low blood iron levels), with symptoms of fatigue, paleness, lack of energy and shortness of breath.

Although these can be symptoms related to carrying the gene, they can also be symptoms of a gynaecological disorder, so it is important to consult a gynaecologist.

With diagnosis and appropriate treatment, these bleeding problems can usually be reduced or managed.

If you are a woman or girl who carries the haemophilia gene, a holistic or comprehensive care approach to your health care can help you to achieve better health and quality of life. Specialist gynaecological care over your lifetime is important to manage any gynaecological issues that occur. These may not be related to haemophilia, but in some cases the bleeding disorder may make the bleeding problems worse.

At times in their life, some symptomatic women may need to have gynaecological surgery or procedures. If this happens, it is important that this is managed in a team, with discussion between the woman, the Haemophilia Centre and the gynaecologist and/or surgeon.

Ideally your medical care team should work together on your health care and should include:

- A gynaecologist
- A haematologist specialising in bleeding disorders
- A GP or paediatrician or obstetrician, if relevant at the time.

*For more information about treatment and managing other bleeding problems, see the TREATMENT AND CARE section, page 15.*

## **GENETIC TESTING**

A normal factor VIII or factor IX level test does not mean that a girl or woman does not carry the haemophilia gene.

Finding out whether a girl or woman carries the haemophilia gene is a process which may take some time. This can involve:

- Meeting with a genetic counsellor
- Looking at the family tree to identify other family members who may carry the haemophilia gene
- Blood tests for other affected family members, if known, to identify the particular gene alteration causing haemophilia in her family
- Laboratory tests on a blood sample from the girl or woman to see if she has the same family gene alteration
- A blood test to check clotting factor levels if they are not known.

Genetic counselling is available to girls or women and their parents or partners and many find it helpful. The Haemophilia Centre can help with information and advice about genetic testing and provide a referral to a genetic counsellor.

## **PREGNANCY AND CHILDBIRTH**

With good management, women who carry the haemophilia gene have no more problems with delivering a healthy baby than other mothers.

How to prepare:

- Ideally, if you are planning a pregnancy, contact your Haemophilia Centre for a referral to a genetic counsellor
- When you become pregnant, contact your Haemophilia Centre for advice on local obstetric services they already work with
- Ask your haemophilia and obstetrics teams to consult with each other to plan for a smooth and safe pregnancy and delivery and care for the newborn

- Check with your Haemophilia Centre before having any invasive procedures, such as amniocentesis
- Discuss suitable choices for anaesthesia, especially an epidural, with your Haemophilia Centre and obstetrics teams.

A normal vaginal delivery is usually recommended unless there are obstetric complications.

