Understanding haemophilia

What is haemophilia?

Haemophilia is a genetic bleeding disorder where blood doesn't clot properly. It is caused when blood does not have enough clotting factor. A clotting factor is a protein in blood that controls bleeding.

There are two types of haemophilia. Both have the same symptoms.

Haemophilia A is the most common form and is caused by having low levels of **clotting factor VIII (8)**.

Haemophilia B is caused by having low levels of **clotting factor IX (9)**.

Haemophilia is not contagious. It is a genetic condition and a person with haemophilia is born with it.

What causes haemophilia?

Everyone has the genes responsible for making factor VIII (the F8 gene) and factor IX (the F9 gene). These factors are necessary for blood to clot.

Haemophilia is caused by a mutation or alteration in the F8 or F9 gene. This alteration interferes with the way the gene functions, so that it doesn't work as well as it should. As a result, the body does not produce enough factor VIII or IX for blood to clot properly.

- Men and boys with the gene alteration **always** have haemophilia
- Most women and girls with the gene alteration do not have bleeding problems
- Some women and girls with the gene alteration have bleeding symptoms and may have haemophilia.

How common is haemophilia?

Haemophilia is rare. It occurs in all races and socio-economic groups.

Haemophilia in males

- Approximately 1 in 6,000 males has haemophilia A
- Approximately 1 in 25,000-30,000 males has haemophilia B

Severity in males

In Australia around **53%** of men and boys with haemophilia have **mild haemophilia**.

Around **47%** of men and boys with haemophilia have **moderate or severe haemophilia**.[2]

Haemophilia in females

- Research is being conducted worldwide to understand how haemophilia affects females and how common it is.
- One study of Haemophilia Treatment Centre patients found 1.6 female carriers to 1 male with haemophilia – but noted there would also be other females who did not know they were carriers.[3]

Severity in females

In Australia **most** women or girls who carry the gene alteration for haemophilia have normal factor VIII or factor IX levels and **do not have bleeding symptoms**.

Around **20-30%** of girls or women with the gene alteration causing haemophilia have reduced factor levels and may have **a bleeding tendency**.

If a girl or woman's factor levels are in the range for mild haemophilia (5-40% of normal clotting factor), she is now recognised as having **mild haemophilia**.

Some females with factor levels at the lower end of normal (40-50%) have abnormal bleeding. If investigation shows this is related to haemophilia, they will be treated as having mild haemophilia.

Very rarely, girls or women have particularly low factor levels (below 5% of normal clotting factor) causing them to have **moderate or severe haemophilia**.[1, 4]

What happens when you have haemophilia?

A key characteristic of haemophilia is to have a bleeding tendency.

Haemophilia is a lifelong condition. It can't yet be cured, but with current treatments it can be managed effectively.

There are different levels of severity in haemophilia related to the amount of clotting factor in the blood: mild, moderate and severe.

A person with haemophilia does not bleed any faster than anyone else, but bleeding can continue for longer if it is not treated, causing poor healing. Minor bruising or scratches on the skin are not usually a problem.

For people with haemophilia, situations become more serious when there is internal or prolonged bleeding. If normal first aid does not stop the bleeding, without other treatment the bleeding can continue for days. Specialised treatment will be needed so blood can clot normally.

A problem for many people with haemophilia is internal bleeding episodes or 'bleeds' into muscles, or organs or joints, especially knees, ankles or elbows. They cause painful swelling and bruising. They can often happen as a result of injury. Some bleeds don't seem to have an obvious cause (sometimes called 'spontaneous bleeds') - this is more common in people with severe haemophilia.

Treatment

Current haemophilia treatment helps the blood to clot normally.

Treatment may be used to prevent bleeding episodes or control a bleeding episode once it starts.

For more information on treatment, see the **Treatment Plan** section on page 29.

Sources

References

1. van Galen KPM, d'Oiron R, James P, et al. A new hemophilia carrier nomenclature to define hemophilia in women and girls: Communication from the SSC of the ISTH. Journal of Thrombosis and Haemostasis. 2021;19(8):1883-1887. Accessed 24 March 2022. Available from https://doi.org/10.1111/jth.15397.

2. Australian Bleeding Disorders Registry data provided to HFA by the National Blood Authority in 2020, following an HFA request approved by the ABDR Steering Committee in 2020 and 2022.

3. Kasper CK, Lin JC. How many carriers are there? Haemophilia 2010;16:842. Accessed 24 March 2022. Available from https://doi.org/10.1111/j.1365-2516.2010.02210.x.

4. Hermans C, Kulkarni R. Women with bleeding disorders. Haemophilia. 2018;24(Suppl. 6):29-36. Accessed 25 March 2022. Available from https://doi.org/10.1111/hae.13502.

5. Srivastava A, Santagostino E, Dougall A, et al. WFH guidelines for the management of hemophilia, 3rd edition. Haemophilia. 2020: 26(Suppl 6): 1-158. Accessed 24 March 2022. Available from https://doi.org/10.1111/hae.14046.

Other sources

Alabek M, Mohan R, Raia MA. Genetic counselling for hemophilia. Rev. edn. Treatment of hemophilia No 25. Montreal: World Federation of Hemophilia, 2015. Accessed 24 March 2022. Available from http://www1.wfh.org/publications/files/pdf-1160.pdf.

Australian Haemophilia Centre Directors' Organisation. Guidelines for the management of haemophilia in Australia. Melbourne; Canberra: AHCDO; National Blood Authority, 2016. Accessed 24 March 2022. Available from https://www.blood.gov.au/haemophilia-guidelines.

McLintock, C. Women with bleeding disorders: Clinical and psychological issues. Haemophilia 2018;24(Suppl. 6):22–28. Accessed 24 March 2022. Available from https://doi.org/10.1111/hae.13501.

World Federation of Hemophilia. Carriers and women with hemophilia. Montreal: WFH, 2012. Accessed 24 March 2022. Available from https://www1.wfh.org/publication/files/pdf-1471.pdf.

NB: All photos in this booklet are stock images.

Acknowledgements

Written by Suzanne O'Callaghan, HFA Policy Research and Education Manager, and Marg Sutherland, health educator.

Quotes and personal stories in this resource were contributed by Australian women with haemophilia or who carry the gene alteration. We thank them for their generosity in sharing their experiences.

Our thanks also to the HFA Women's Consumer and Health Professional Review Groups for their suggestions on topics and content to include.

Reviewers

Australia and New Zealand Haemophilia Psychosocial Group: Nicoletta Crollini, Dr Moana Harlen, Jane Portnoy.

Australian Haemophilia Centre Directors' Organisation: A/Prof Chris Barnes, Dr Stephanie P'ng.

Australian Haemophilia Nurses' Group: Jaime Chase, Susan Dalkie, Janine Furmedge, Penny McCarthy, Joanna McCosker, Megan Walsh.

Genetics and genetic counselling: Clinical A/Prof Kristi Jones, Senior Staff Specialist in Clinical Genetics, and Lucy Kevin, genetic counsellor, The Children's Hospital at Westmead, Sydney.

Haemophilia Foundation Australia: Sharon Caris.

HFA Women's Consumer Review Group – individuals not named for privacy reasons.

Maurice Blackburn Lawyers: Katherine Bedford, Senior Associate.

More information

To find more information about haemophilia and carrying the gene alteration, or to find out how to get in touch with your local Haemophilia Foundation or a specialist Haemophilia Treatment Centre, contact:

Haemophilia Foundation Australia

7 Dene Ave Malvern East Victoria 3145

T: 03 9885 7800 Toll free: 1800 807 173

E: hfaust@haemophilia.org.au

Or visit the HFA website: www.haemophilia.org.au

Important note

This booklet was developed by Haemophilia Foundation Australia for education and information purposes only and does not replace advice from a treating health professional. Always see your health care provider for assessment and advice about your individual health before taking action or relying on published information.

© Haemophilia Foundation Australia, May 2022.

This factsheet may be printed or photocopied for education purposes.

HAEMOPHILIA FOUNDATION AUSTRALIA