

# Haemophilia testing in women and girls

If you are a woman or girl who may be affected by haemophilia, there are tests to help with your diagnosis. These tests can tell whether you have haemophilia or carry a genetic alteration that causes haemophilia.

This information has been developed for women, girls and their parents to explain what is involved in the tests and answer some common questions.



# Why test for haemophilia?

## There are important reasons for testing for haemophilia in women and girls:

- To know whether a woman or girl is affected by haemophilia
- If a woman or girl has a bleeding tendency and/or haemophilia, she will need a treatment plan to prevent bleeding complications
- Information for pregnancy or family planning
- Preparing a safe childbirth for mother and baby
- To help with diagnosing haemophilia in other family members.

**Haemophilia** is an inherited genetic bleeding disorder. It is caused by a **mutation** or alteration in the DNA code for **the factor VIII/F8 or factor IX/F9 gene**. As a shorthand, this F8 or F9 gene alteration is often referred to as 'the gene'. Some people in the community call it the 'haemophilia gene'.

- Females can carry the gene alteration for haemophilia without symptoms and pass it on to their children.
- Some women and girls with the gene alteration may also have a bleeding tendency and have haemophilia themselves.

## If you are female, why might your doctor suggest you be tested for haemophilia?

- There is a history of haemophilia in your family
- Or a close relative like your brother, sister or mother has recently been diagnosed
- Or your child has haemophilia or is a carrier
- Or you have bleeding symptoms that suggest you may have haemophilia.

Types of haemophilia testing:

- **Genetic testing:** to identify if a woman or girl has an alteration in her F8 or F9 gene associated with haemophilia
- **Factor level testing:** to see if a woman or girl has low levels of factor VIII or IX and may have a bleeding tendency or haemophilia.

You may need to have both genetic testing and factor level testing. For example, a woman or girl can have normal factor levels and still carry the gene alteration that causes haemophilia.

# 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: AHCCO; National Blood Authority, 2016. Accessed 24 March 2022. Available from <https://www.blood.gov.au/haemophilia-guidelines>.
- McIntock, 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 Furnedgedge, 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](mailto:hfaust@haemophilia.org.au)

Or visit the HFA website: [www.haemophilia.org.au](http://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.

