How do you know if you have the gene alteration that causes haemophilia?

There are a number of steps to identify if you have an alteration in your F8 or F9 gene that causes haemophilia. The steps are outlined here and then explained in greater detail in the rest of this education resource.

Obligate carriers

In some cases a family history of haemophilia will identify whether you have the gene alteration. In genetics this is described as being an ***obligate carrier***, because the pattern of inheritance means you **must** have the altered gene.

Obligate carriers include:

- **ALL** daughters of a man with haemophilia
- Mothers of one child with haemophilia, and who have at least one other family member with haemophilia
- Mothers of one child with haemophilia, and who have a family member who is a known haemophilia carrier
- Mothers of two or more children with haemophilia.

Gene changes

Everyone has many gene changes throughout their DNA. These gene changes are called **variants or mutations** or **alterations**. Some are common and some are rare. Some gene changes have no effect on your health, while others can cause genetic conditions, like the alterations to the F8 and F9 gene that can result in haemophilia.

What is a *haemophilia carrier*?

In genetics females who have the gene alteration for haemophilia are described as 'carriers'. Most carry the gene alteration without symptoms. Some with the gene alteration have a bleeding tendency and have the medical condition haemophilia. A female with the gene alteration can pass it onto her children, whether she has bleeding symptoms or not.

Clotting factor level tests

- Clotting factor level tests do not take the place of genetic testing. However, if factor VIII (8) or factor IX (9) tests show that you have lower than normal factor levels, this suggests that you are likely to have the gene alteration and be a carrier.
- Normal or borderline clotting factor level tests will not tell you whether you carry the gene alteration. Many women and girls have normal factor levels and no symptoms but still have the gene alteration that causes haemophilia.

Genetic carrier testing

You may need to have genetic testing to confirm if you have the altered copy of the F8 or F9 gene.

There are a number of gene alterations causing haemophilia and families will share a specific alteration.

Identifying the gene alteration in your family can be part of the process in genetic testing. If your family gene alteration is not already known, this will involve some steps before you have genetic testing:

- Usually genetic testing of a male with haemophilia in your family - or less commonly, an affected female
- Once the genetic alteration in your family is known, you can have genetic testing for the same known or familial alteration to see if you are a carrier.

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