How is haemophilia diagnosed?

Haemophilia is a medical condition where a person has low levels of clotting factor VIII or factor IX. These low factor levels or deficiencies are caused by gene variants or mutations in the clotting factor VIII or IX gene.

In women and girls haemophilia is usually diagnosed through:

- The physical signs that you have unusual bleeding problems

And • Checking the family history for bleeding problems

And • Laboratory tests on a blood sample for your clotting factor levels

And • Genetic testing that shows you have the gene alteration for haemophilia.

Diagnostic categories for females

In the past women and girls with bleeding symptoms were generally called ‘symptomatic carriers’. In Australia, diagnostic categories have now changed.[1]

These categories can be helpful for many women and girls when their results are clear but if a woman or girl has borderline factor levels, singling out which category is appropriate can be challenging.

Factor level	Diagnostic category
Females with • an F8 or F9 gene alteration • and clotting factor levels in the range for haemophilia (0-40% of normal clotting factor levels)	**Haemophilia**
• Some females who carry the gene alteration and have factor levels at the lower end of normal (40-50%) may also experience abnormal bleeding. • If further investigation indicates this bleeding is related to a factor VIII or factor IX deficiency, they will be treated as having haemophilia.	**Symptomatic haemophilia carrier**
• Most females who carry the gene alteration will have normal factor VIII or factor IX levels and will not have bleeding problems.	**Asymptomatic haemophilia carrier**

The journey to diagnosis

Haemophilia is rare and some women have not been diagnosed until later in life, although they have bleeding symptoms. There is ongoing work worldwide to increase awareness about haemophilia in females, so that women and girls can be diagnosed earlier and the pathway to diagnosis can be a smoother experience.

> *“When I was diagnosed in my 40s, I was the first person I had ever known with haemophilia. Little did I know I had had it all my life. I was unsure how I was never tested before with all the bleeding and bruising I experienced throughout my life.”*

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NB: All photos in this booklet are stock images.

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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.

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