Other bleeding disorders

Rare clotting factor deficiencies

Rare clotting factor deficiencies are another group of inherited bleeding disorders, caused when the body does not produce enough of a certain clotting factor, or when the factor does not work properly. Less is known about these disorders because they are very uncommon. In fact, many have only been discovered in the last 40 years.

Both females and males can be affected by rare clotting factor deficiencies.

Rare clotting factor deficiencies include:

- Factor I (1) deficiency – often called *fibrinogen deficiency*
- Factor II (2) deficiency
- Factor V (5) deficiency
- Combined factor V (5) and VIII (8) deficiency
- Factor VII (7) deficiency
- Factor X (10) deficiency
- Factor XI (11) deficiency
- Factor XIII (13) deficiency.

Photo by Bess Hamiti from Pexels

Symptoms

The particular pattern of bleeding symptoms in rare clotting factor deficiencies depends on the type of deficiency. It can also depend on the type of genetic alteration a person has inherited (called a *phenotype*). In some factor deficiencies, the symptoms can also be mild, moderate or severe depending on the amount of factor in the person's blood.

Finding out more about your disorder

If you have a rare clotting factor deficiency or inherited platelet disorder, ask your HTC for an information brochure that is specifically about your type of bleeding disorder.

There is also more information about RARE CLOTTING FACTOR DEFICIENCIES and INHERITED PLATELET DISORDERS on these websites if you look under BLEEDING DISORDERS:

- World Federation of Hemophilia – www.wfh.org
- Canadian Hemophilia Society - www.hemophilia.ca

Inheritance

Most of these conditions are inherited in what is known as an **autosomal recessive** pattern (like the inheritance pattern for VWD on page 13). This means that the person with the rare factor deficiency has nearly always inherited an altered gene from both parents. Some are found more often in areas of the world or communities where marriage between close relatives is common. In other families, parents may not be aware that they have the gene because they have few or no symptoms and it can be very unexpected when their child is diagnosed.

Exceptions are factor XI (11) deficiency and some types of factor I (1) deficiency, which can be inherited in either an **autosomal recessive** or **autosomal dominant** pattern (like the VWD inheritance patterns on pages 13-14).

Inherited platelet function disorders

Examples of inherited platelet function disorders are:

- Glanzmann's Thrombasthenia
  Affects the fibrinogen receptor
- Bernard-Soulier syndrome
  Affects the von Willebrand factor (VWF) receptor

In platelet function disorders, the platelet plug does not form properly, leading to a tendency to bleed for longer than normal or bruise easily. Since platelets have many roles in blood clotting, platelet function disorders can cause bleeding that ranges from mild to severe.

Acquired haemophilia and VWD

Nearly all people with haemophilia and von Willebrand disease (VWD) are born with these health conditions. These are called inherited bleeding disorders. However, a person can develop a different kind of haemophilia or VWD throughout life, usually as an adult, although this is very rare. This is known as an acquired bleeding disorder.

Acquired haemophilia can occur when a person's immune system produces antibodies that mistakenly target their own factor VIII. It is very rare but can sometimes occur in older people and young women who are in the later stages of pregnancy or have recently given birth. The bleeding pattern is different to inherited haemophilia and it is usually curable with treatment.

Acquired von Willebrand disease can also occur but is extremely rare.

Acquired bleeding disorders are not inherited or passed on to children. Men and women are equally likely to be affected by an acquired bleeding disorder.

If you have an acquired bleeding disorder, ask your HTC for more information about your disorder.

Factor deficiency	How rare? (estimates)	Inheritance	Severity of bleeding
Factor I (1): Afibrinogenemia	5 in 10 million people	Autosomal recessive	Usually mild, except in afibrinogenemia.
Hypofibrinogenemia	Not available	Autosomal recessive or dominant	Some can experience thrombosis (blood clots in veins or arteries) instead of bleeding
Dysfibrinogenemoa	1 in 1 million people	Autosomal recessive or dominant	
Factor II (2)	1 in 2 million people	Autosomal recessive	Usually mild. Moderate to severe when factor levels are low
Factor V (5)	1 in 1 million people 1 in 100,000 in some populations, including Israel, Iran and Italy	Autosomal recessive	Usually mild. Moderate to severe when factor levels are low
Combined Factor V (5) and Factor VIII (8)	1 in 1 million people	Autosomal recessive (very rarely, factor VIII deficiency can be inherited separately from only one parent)	Usually mild
Factor VII (7)	1 in 500,000 people	Autosomal recessive	Severe when factor levels are low
Factor X (10)	1 in 1 million people	Autosomal recessive	Moderate to severe when factor levels are low
Factor XI (11)	1 in 100,000 people	Autosomal recessive or dominant	Variable
Factor XIII (13)	1 in 3 million people	Autosomal recessive	Moderate to severe when factor levels are low
Bernard-Soulier Syndrome	1 in 1 million people	Autosomal recessive	Variable (usually moderate to severe) [1]
Glanzmann thrombasthenia	1 in 1 million people	Autosomal recessive	Variable (usually moderate to severe) [2]

Source: Adapted from National Blood Authority. Australian Bleeding Disorders Registry Annual Report 2015-16. NBA: Canberra, 2017; World Federation of Hemophilia. Characteristics of rare clotting factor deficiencies. WFH; Montreal, 2014.

For more information visit factoredin.org.au

More Information

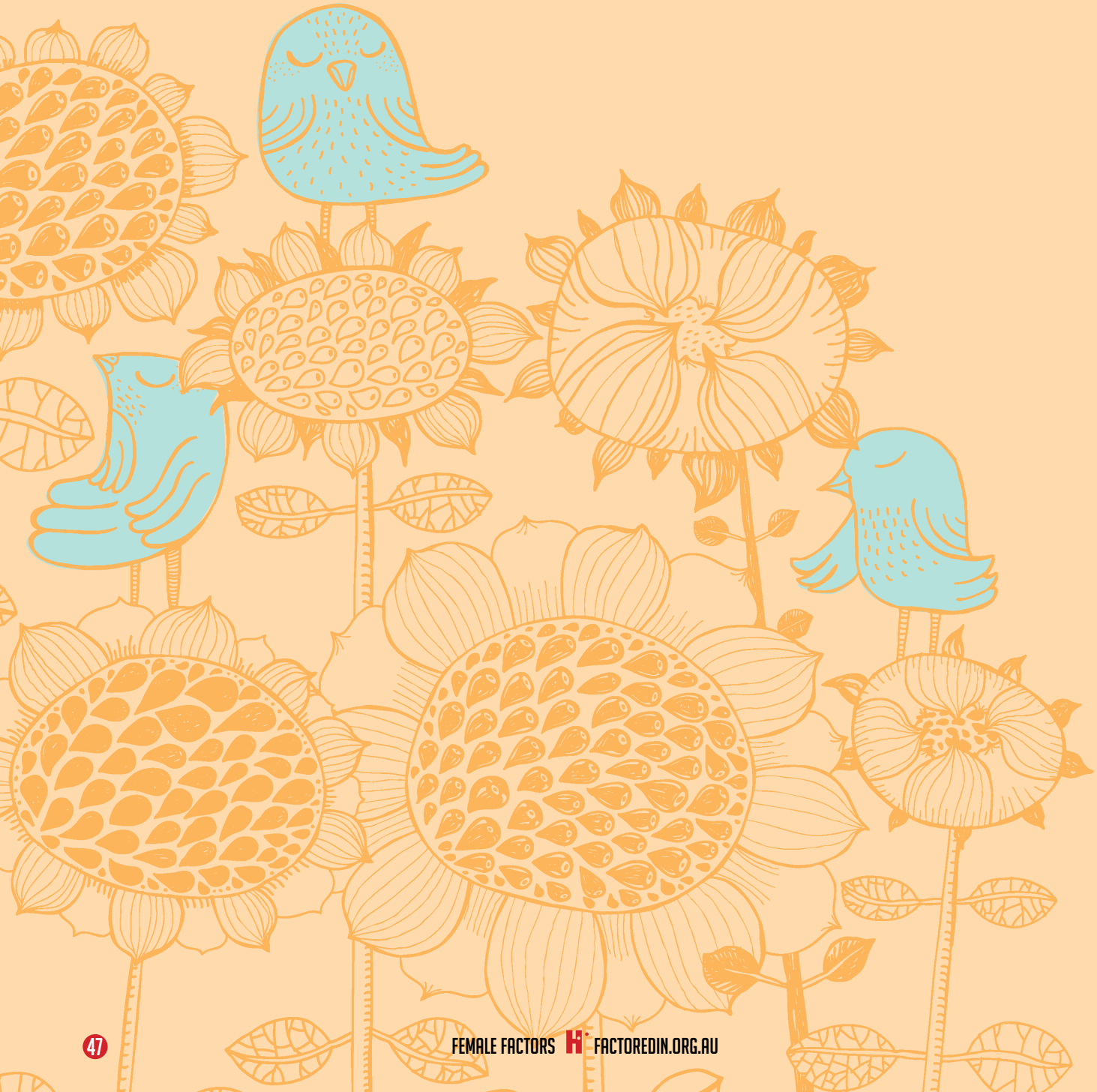
For more information about bleeding disorders, or to find out how to get in touch with your local Haemophilia Foundation or a specialist Haemophilia Treatment Centre, contact:

Haemophilia Foundation Australia

P: **1800 807 173**

E: **hfaust@haemophilia.org.au**

W: **www.haemophilia.org.au**



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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.