

Glossary

ABDR: The Australian Bleeding Disorders Registry (ABDR) is the system used by Haemophilia Treatment Centres around Australia for the clinical care of their patients.

Antibody: A protein which is produced by the body's immune system in response to and to defend against substances that the body recognises as foreign. Antibodies to clotting factor concentrates that occur in people with haemophilia are called inhibitors.

Arthritis: Inflammation of a joint causing pain and stiffness. Problems related to arthritis include inflammation of the synovial lining (the membrane that lines joints such as elbows and knees), and damage to joint cartilage (the tissue that covers the ends of bones, enabling them to move against each another). In haemophilia, arthritis is caused by repeated bleeding into the joint cavity.

Blood clot: The jelly-like mass that results when blood platelets and fibrin mesh to seal a leaking blood vessel.

Carrier: A person who has a certain genetic alteration in his or her genetic make-up but has no symptoms; the genetic alteration can be passed to offspring. In haemophilia carriers are females who have an X chromosome where there is a mutation in the factor VIII (8) or factor IX (9) gene. Their sons and daughters may inherit this altered gene from them.

Central venous line (CVL): Also known as a central venous catheter. It is a long, thin, soft tube that is inserted into a large vein leading to the heart. In haemophilia it is used to give regular doses of factor replacement therapy over a long period of time, usually a few weeks or more, to protect the small veins in arms and legs. It makes it easier, quicker and less painful to give small children their replacement factor therapy.

Christmas Disease: Another name for haemophilia B or factor IX deficiency. It was named after Stephen Christmas, the first patient with haemophilia B described in medical journals.

Chromosome: The part of a cell in the body that contains the genes. Genes determine some individual characteristics of a human, such as the colour of a person's eyes.

Chronic health condition: a health condition that persists for a long time. Haemophilia is often described as a chronic health condition because a person has haemophilia for their lifetime.

Classical haemophilia: another term for haemophilia A or factor VIII (8) deficiency

Clotting: The process to stop bleeding when a blood vessel is injured and begins to leak blood. This process is also known as coagulation. One part of the process is a chain reaction with clotting factor proteins, where they link to form a chain called fibrin at the site of a break in the blood vessel wall. The fibrin chain becomes a plug to seal the blood vessel. In haemophilia the body does not produce enough clotting factor VIII (8) or IX (9) for the fibrin plug to form properly and blood continues to ooze, delaying healing.

Clotting factor concentrate: A condensed, powdered preparation of clotting proteins, which is dissolved in sterile water and then infused (injected) to correct a bleeding disorder such as haemophilia. These concentrates can be manufactured by recombinant technology to make a synthetic product or from human plasma. There are concentrates to correct deficiencies in factors I, II, VII, VIII, IX, X, XI, XIII and von Willebrand factor. Factor VIII or factor IX concentrates are used to treat haemophilia. See *factor replacement therapy*.

Coagulation: The process of blood clotting. See *clotting*.

Comprehensive care: Care that covers all aspects of one's wellbeing and health, including mental, emotional and physical care. Comprehensive care for haemophilia involves a specialist team with experience in bleeding disorders, with key elements of diagnostic services, factor replacement therapy, rehabilitation and physiotherapy, and psychosocial support.

Counsellor: a health professional who is trained to help their clients talk about their problems, understand them, and make positive changes in their lives. They do not tell their clients what to do, but help them to make their own decisions. They provide confidential support.

Factor: A blood plasma protein that is an agent in the clotting process.

Factor VIII (8): a protein in the blood that is essential for clotting. Factor VIII levels are low in a person with haemophilia A.

Factor IX (9): a protein in the blood that is essential for clotting. Factor IX levels are low in a person with haemophilia B.

Factor replacement therapy: a treatment for haemophilia to stop or prevent bleeding. The person with haemophilia is given an injection containing (or infused with) the blood clotting factor product required. See *clotting factor concentrate*.

Gene: Tiny structures of DNA that give the directions for forming the body, including individual characteristics such as the colour of a person's hair or eyes. Genes are the basic unit in heredity and are passed down from parent to child through the generations. Genes are located on chromosomes. Haemophilia is caused by an alteration in the factor VIII or factor IX gene.

Gene alteration: A change (mutation) that alters the directions carried by a gene, and sometimes produces a new characteristic or trait not found in either parent. This change in the hereditary unit is permanent. In haemophilia there is an alteration in the factor VIII or factor IX gene resulting in low levels of factor VIII or factor IX in the body. This can cause bleeding to continue for longer than normal.

Gene therapy: A treatment where a copy of a normally functioning gene is inserted into a patient's cells so that the body will reproduce the normally functioning gene. In haemophilia this is a normal copy of either the factor VIII gene or the factor IX gene, whichever gene is currently altered in the patient.

Genetic testing: Laboratory tests that in most cases can identify the specific factor VIII or IX gene mutation in an individual with haemophilia or a carrier. This type of test can also be used for prenatal diagnosis to determine whether or not a fetus has the genetic mutation.

Haemophilia: A chronic, genetic blood disorder characterised by a bleeding tendency where bleeding continues for longer than normal. Most bleeding episodes are internal, into the joints or muscles. It is caused by a mutation in a protein (factor VIII or factor IX) needed for blood clotting. This causes the body to produce little or no factor VIII or factor IX.

Haemophilia A: A type of haemophilia caused by low levels of factor VIII (8). This is also called classical haemophilia and factor VIII deficiency.

Haemophilia B: A type of haemophilia caused by low levels of factor IX (9). This is also called Christmas disease and factor IX deficiency.

Haemophilia nurse: a nurse who is in a dedicated role to provide nursing care to people with bleeding disorders as part of the team at the Haemophilia Treatment Centre.

Haemophilia Treatment Centre (HTC): A designated medical centre staffed by a team of health professionals who specialise in bleeding disorders. The team will typically include specialist doctors (haematologists), nurses, physiotherapists and psychosocial workers and access to specialist laboratory services. The Centre co-ordinates and often provides specialist diagnosis, treatment, support and information to people with haemophilia and their families.

Haematologist: A doctor with specialist qualifications to manage blood diseases. These doctors provide explanations and guidance and will recommend the treatment that is appropriate for your child. They will review your child regularly, monitor their progress and adjust your child's treatment accordingly

Hereditary: Passed in the genes from parent to offspring; the basic unit of heredity is the gene.

Infusion: In haemophilia this is usually referring to the injection of a clotting factor treatment product into the bloodstream via a vein to help stop or prevent bleeding.

Inherited platelet disorders: Platelet function disorders are conditions where platelets don't work the way they should, resulting in a tendency to bleed or bruise. Inherited platelet disorders include Bernard-Soulier syndrome, Glanzmann thrombasthenia and storage pool deficiencies.

Inhibitors: Antibodies produced by the body's immune system in reaction to factor replacement therapy that is being used to treat or prevent bleeds. These antibodies are produced when the body's immune system recognises the treatment product as 'foreign'. Inhibitors reduce the effectiveness of the treatment.

Joint: The place where two or more bones come together.

MedicAlert or SOS talisman: A medical identity bracelet or necklace which is engraved with important information about a person's health condition or allergies for emergency situations. The intention is that this information will be read by emergency medical staff, particularly in situations where the person cannot speak for themselves.

MyABDR: A secure smartphone app and website for people with bleeding disorders. It is used to record bleeds and treatments at home. MyABDR links directly to the ABDR, which is the system used by Haemophilia Treatment Centres for the clinical care of their patients.

Physiotherapist: A health professional that assesses, diagnoses, treats, and works to prevent disease and disability through physical means. In haemophilia treatment and care, the physiotherapist plays an important role in the prevention of long-term joint and muscle damage.

Plasma: The pale yellow fluid part of blood that contains proteins such as clotting factors.

Plasma-derived factor concentrate: Plasma-derived factor concentrate is a factor replacement therapy manufactured from the plasma in human blood.

Platelets: Very small cells circulating in the blood, which stick to and spread on the walls of damaged blood vessels to promote blood clotting.

Port: A device surgically implanted under the skin in the chest wall to give easier access to a vein and to protect veins from continued needle sticks. It is a temporary device that makes infusing factor replacement therapy easier and quicker in children. A port may also be referred to as an implanted central venous access device (CVAD) or an infusaport.

PRICE: The Protection, Rest, Ice, Compression and Elevation (PRICE) treatment method is a form of immediate treatment used to prevent a bleed from developing, prevent further bleeding, reduce swelling and speed up recovery.

Prophylaxis: Infusing factor replacement therapy on a regular basis. The intention is to keep factor levels at a sufficient level so as to prevent most, if not all, bleeds.

Psychosocial worker: A broad term used to describe a psychologist, social worker or counsellor. In haemophilia psychosocial workers have an important role in providing or co-ordinating counselling and support for people with haemophilia, their partner and family.

Psychologist: a health professional who specialises in the science of mental processes and how people think, feel, and behave. They can evaluate, diagnose and provide help with issues related to the mental health and social emotional wellbeing of people.

Rare clotting factor deficiencies: These are bleeding disorders other than haemophilia and von Willebrand disease caused when a particular factor or protein in the blood that helps blood to clot is lower than normal, missing or doesn't work properly. They include factor I, II, V, VII, X, XI, XIII deficiencies and combined factor V and factor VIII deficiency.

Recombinant factor: Synthetic factor concentrate made in a laboratory by genetic engineering. It contains little or no material from human blood or animals.

Social worker: a health professional who helps people in times of crisis. Social workers can provide their clients with practical support, counselling, information and emotional support.

Spontaneous bleed: Bleeds that appear to happen for no obvious reason. This means that there is no specific identified injury that caused this bleed to occur. Sometimes, on reflection, parents or children can identify an activity that may have contributed to the bleed. Spontaneous bleeds are more common in people with severe haemophilia and occur less often in people with moderate haemophilia. They are uncommon in mild haemophilia.

Synovial fluid: The thick, sticky fluid produced in the spaces between certain joints to reduce friction and help joints to move smoothly.

Target joints: The joints where bleeds occur most often. A target joint is defined as a joint that has had three or more bleeds without an apparent cause within a six-month period.

Tolerisation: A treatment method for inhibitors where the person receives high doses of factor product. Also known as immune tolerance induction (ITI).

Vein: Blood vessel which carries blood from any part of the body back to the heart.

Von Willebrand Disease (VWD): Another type of inherited bleeding disorder. People with VWD have a problem with a protein in their blood called von Willebrand factor (VWF) that helps control bleeding. They do not have enough VWF or it doesn't work properly and it takes longer for blood to clot and bleeding to stop. VWD is the most common bleeding disorder worldwide. Also known as von Willebrand disorder.