

Chapter 1

What Is von Willebrand Disease?

Von Willebrand disease (VWD) is an inherited bleeding disorder. Although you may have only recently heard of it, VWD is the most common inherited bleeding disorder on earth, affecting about 1% to 2% of the world's population.^{1,2} Between 60 and 120 million people worldwide may have VWD.³

It is upsetting to learn that you or your loved one has a chronic, lifelong disorder. Although you are definitely not alone, if you have VWD you may *feel* alone. Even though at least 60 million people worldwide may have VWD, many, if not most, don't even know they have it. Because VWD is not a well-publicized disorder, the general public and many healthcare providers have not heard of it. Some people with VWD may live their entire life without being properly diagnosed. Have you heard people say, "I bruise easily," "I take *forever* to stop bleeding after I cut myself," or "My periods last for weeks"? It is quite possible they have VWD and don't even know it.

If you are reading this book, it is likely that you or a family member has VWD or you know someone who does. Educating yourself is an important step. Because VWD is still classified as a rare disorder, many healthcare providers have little knowledge of how to diagnose and treat it. Like other rare, chronic disorders, patients or their loved ones often know more about the disorder than the healthcare provider who is treating them. If you have VWD, you will have to take an active part to ensure that you receive the best health care.

The Discovery of von Willebrand Disease

I can remember looking out at our next door neighbor's willow trees and using the word willow to try to remember Willebrand. —J.K., New York

You may wonder how this disorder got its strange name. Dr. Erik Adolf von Willebrand, a Finnish physician, first described VWD in 1926. Born in Vaasa, a seaport in west central Finland on the Gulf of Bothnia, he received his PhD from the University of Helsinki, Finland, in 1899. The following year, he received his medical degree from the same university. He began to study an unusual bleeding disorder in the local communities called "Alandic Hemorrhagic Disease."

¹Rodeghiero F, Castaman G, Dini E. Epidemiological investigation of the prevalence of von Willebrand's disease. *Blood*. 1987;69:454-459.

²Werner EJ, Broxson EH, Tucker EL, et al. Prevalence of von Willebrand disease in children: a multiethnic study. *The Journal of Pediatrics*. 1993;123:893-898.

³World POPclock Projection. US Census Bureau. Available at: www.census.gov. Accessed March 14, 2006.

A Guide to Living With von Willebrand Disease

In 1925, while in Helsinki, Dr. von Willebrand was asked to evaluate a five-year-old girl named Hjördis S. from the remote island of Föglö belonging to Åland, a group of islands in the Baltic Sea between Sweden and Finland. She was suffering from excessive bleeding from the lip following an injury. She had a history of nosebleeds and bleeding following tooth extraction. She was the ninth of 12 children, 10 of whom showed bleeding symptoms. Sadly, four of her siblings died between the ages of two and four of uncontrollable bleeding. Tragically, this young girl later bled to death during her fourth menstrual period. When Dr. von Willebrand studied the extended family, he discovered that 23 of 66 family members (16 females and seven males) also had bleeding problems. The history of the original Finnish family has now been traced back to 1750 and reveals over 1,000 persons with 125 known affected members, 12 of whom died from bleeding. Fortunately, most people are not as severely affected by VWD as these family members were. Nonetheless, this case history shows that VWD can be severe in some families.

The type of bleeding affecting this family was different from any other bleeding disorders known at the time. Dr. von Willebrand concluded this was a previously unknown type of hemophilia, characterized by prolonged **bleeding time**⁴ (how long it takes for bleeding to stop) and **mucocutaneous** (mucous membranes and skin) bleeding. This disorder affected both males and females. He termed this new disorder “pseudohemophilia.” He later renamed it “constitutional thrombopathy” because he believed platelets were involved. In 1928, four American doctors described a similar disorder, but it wasn’t until decades later that the actual **protein** involved was identified. The protein—**von Willebrand factor (VWF)**—and the disorder were named after the doctor who first described the unusual symptoms.

Who Has von Willebrand Disease?

Our families were under the impression that “only boys have bleeding disorders.” —M.T., Connecticut

Our doctor didn’t believe that a girl could have a bleeding disorder!
—M.T., Ohio

⁴Terms that appear in bold type are defined in the glossary at the end of the book.