From Diagnosis to Advocacy: How a Patient Turned His Experience Into a Positive Force

Derick, an outgoing 22-year-old diagnosed with a mild platelet function defect, a mild bleeding disorder and possible Ehlers-Danlos (a connective tissue disorder that may increase the risk of bleeding – see page 2), will be spending the next year at the University of Ulster, Belfast, Ireland. There is an interesting connection between his diagnosis and his educational path. He reports that when he was diagnosed at an early age, his parents took the necessary precautions to keep him safe, but did not let his disorder stop him from leading a normal life. As a family they attended Advocacy Conferences through Great Lakes Hemophilia Foundation. There he learned about the public health policy issues that would eventually lead to the pursuit of a career in health care.

In both middle and high school, Derick was restricted from playing football and other contact sports, a common frustration experienced by many young people living with bleeding disorders. However, as he grew older, he made the conscious decision to take ownership of his disorder, becoming a passionate advocate for the bleeding disorder community. He lobbied his congress people and developed a real interest in health care on a more advanced level. This in turn led him to his current career path – the intersection of public policy and health care. Living with a chronic condition and seeing others dealing with similar situations has made him sensitive to the concerns of people with bleeding disorders.

Derick was recently awarded the Mitchell Fellowship for a year of graduate studies in Ireland, where he will be working toward a Masters in Health Communications. He received the fellowship by authoring Congressional legislation to increase screening for and awareness of bleeding disorders. After completion of his Masters he hopes to work on a second Masters in Business Administration. He plans to eventually work in the health care industry. Derick’s advice to other young people with the same or similar disorder is to learn as much as you can about your disorder and to be your own best advocate.
Ehlers–Danlos Syndrome (EDS) refers to a group of heritable connective tissue disorders, characterized by joint hypermobility, skin extensibility, and tissue fragility. There are six distinct types of EDS that vary greatly in symptoms and severity. A diagnosis of EDS is made upon clinical findings and, when applicable, family history.

The most common form of EDS is hypermobility type, which is generally considered the least severe type of EDS. The major manifestation of EDS, hypermobility type is joint hyperextension and dislocation, which can occur spontaneously or with minimal trauma and can be acutely painful, with pain lasting for hours or days after an event. Chronic pain, fatigue, and degenerative joint disease are common and can occur at a younger age than in the general population. The skin is often soft or velvety and may be mildly hyperextensible.

Some individuals with EDS, hypermobility type also have mild bleeding symptoms, including easy bruising, frequently without obvious cause; mildly prolonged bleeding with cuts or epistaxis (nosebleeds); and heavy, prolonged, or breakthrough menstrual bleeding. This can mimic von Willebrand disease or a platelet function disorder, but von Willebrand factor, platelet number and function, and coagulation factor studies are almost always normal in a patient with EDS.

Other common features of EDS, hypermobility type include:
- Headaches, especially migraine
- Functional bowel disorders, reflux (causing heartburn) and stomach ulcers
- Atypical chest pain, heart palpitations at rest or on exertion, and/or dizziness and fainting
- Dilatation of the aorta, usually of a mild degree, occurs in one-quarter to one-third of individuals with EDS

Fragility of soft tissues with spontaneous ruptures or tears of internal organs is, by definition, not a feature of hypermobility type of EDS. Such manifestations should be brought to the attention of a physician for prompt consideration of other hereditary connective tissue disorders.

In most individuals with EDS, hypermobility type, the specific genetic cause is unknown. EDS, hypermobility type is inherited in an autosomal dominant manner. Most individuals diagnosed with the syndrome have an affected parent. Each child of an individual with EDS, hypermobility type has a 50 percent chance of inheriting the disorder.
Blood is carried throughout the body within a network of blood vessels. When tissues are injured, damage to a blood vessel may result in leakage of blood through holes in the blood vessel wall. Normally, bleeding stops through two processes that work together to form a blood clot: 1) the formation of a platelet plug, and 2) stabilization of that platelet plug by the formation of a fibrin clot.

What are Platelets?
Platelets are small disc shaped cells that circulate in the blood. Platelets play an important role in the clotting of blood and the repair injured blood vessels. Platelets stick to, and spread on, areas of damaged blood vessel walls (platelet adhesion). These spreading platelets release substances that activate other nearby platelets which clump at the site of injury to form a platelet plug (platelet aggregation). The surface of these activated platelets then provides a site for blood clotting to occur. Clotting proteins that circulate in the blood are activated on the surface of the platelets to form a mesh-like fibrin clot.

Description of the Disorder
There are several different types of platelet function defects, or PFD, as well as varying degrees of severity. The number of platelets is usually normal; however, the function of the platelets is abnormal.
Most PFDs diagnosed are of the mild hereditary type. These PFDs do not usually “go away.” A patient diagnosed with the hereditary type of PFD will most likely have it for the rest of their life. There are also acquired PFDs. These include PFDs caused by certain medical conditions or from the use of medications, which inhibit platelet function such as aspirin, non-steroidal anti-inflammatory drugs (such as ibuprofen and naproxen), blood thinners, and some antibiotics, antidepressants, anesthetics, and heart drugs. Platelet function returns to normal when these medications are stopped. Bernard-Soulier Syndrome is a rare hereditary PFD that results in an inability of platelets to stick and spread at sites of blood vessel injury. Glanzmann thrombasthenia is also a rare hereditary PFD that results in an inability of platelets to aggregate.

Signs and Symptoms of a Platelet Function Defect
• Easy bruising: bruising with little or no known injury; bruising that occurs in more than one part of the body; bruises which are two inches or larger; firm or tender bumps in center of bruises (hematomas)
• Heavy or prolonged menstrual periods: periods that last longer than one week; need to change tampon or pad every one to two hours during the heavy days of the menstrual cycle; use more than two dozen (24) tampons or pads in one month; frequent need to miss work/school because of heavy menstrual flow
• Frequent prolonged nosebleeds: nosebleeds that occur more than once a year and are difficult to control or take more than 10 minutes to stop even with pressure
• Prolonged or unusual bleeding after injury, surgery, childbirth or dental work: bleeding that has caused your doctor to tell you that you bleed more than expected or that required blood transfusions

Severity of symptoms can vary even among family members who have the same genetic platelet function defect. The majority of people will have mild symptoms and many individuals have no symptoms unless they have a serious injury or an operation. In a few individuals, platelet function disorders can cause severe bleeding.

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Diagnosing the Disorder
When a bleeding disorder is suspected, diagnosis is made by obtaining a thorough personal history, family history, physical examination, and laboratory testing (blood tests). These blood tests include:

- Complete blood count: to make sure the platelet count (number of platelets in the blood) is adequate to provide normal clotting.
- Bleeding time: measured by inflating a blood pressure cuff on the upper arm and using a device to make a very small cut on the forearm. The time it takes this cut to stop bleeding is measured using a stopwatch. The bleeding time might be longer than normal in patients with abnormal platelet function. However, quite often the bleeding time is normal in patients with mild abnormal platelet function. The bleeding time is being replaced by a test called the Platelet Function Screen (PFA-100). The test measures the time it takes for whole blood to clot and stop the flow of blood in a cone-like device coated with various substances that help platelets stick to each other. Like the bleeding time, it might be longer than normal in patients with abnormal platelet function, but may be normal.
- Platelet aggregation study: Platelets are collected from a blood sample and stirred in tubes where they can be monitored for clumping (aggregation). Activating substances are added to stimulate aggregation and the effects are recorded on a chart. Failure of platelets to aggregate under these conditions suggest that there are abnormalities in platelet release or aggregation.

Treating the Disorder
The treatment of PFDs will depend on the particular type of PFD, as well as the location and severity of bleeding. Most patients require NO treatment on a regular basis, but DO need treatment at the time of surgical procedures (including dental procedures), or following accidental injury.

- IV DESMOPRESSIN (DDAVP) - This is a medication that is injected intravenously (into a vein) before surgery or after an accident, to prevent or decrease excessive bleeding. The response to DDAVP varies among individuals. Patients who do not respond to DDAVP may require platelet transfusion in the event of bleeding. Side effects of DDAVP may include headache and facial flushing. DDAVP causes fluid retention and careful monitoring of fluid intake after use of DDAVP is very important to avoid the possibility of fluid overload with resultant seizures. (IV DDAVP is usually not effective in treating severe bleeding in patients with Glanzmann thrombasthenia or Bernard-Soulier syndrome).
- STIMATE - This is the nasal spray form of DDAVP. This medication is often used by patients with PFD at home to control nosebleeds, heavy menstrual periods, and other bleeding symptoms. Some patients with Glanzmann or Bernard-Soulier may respond to Stimate for minor bleeding. (Note: There is an intranasal spray form of desmopressin by the brand name “DDAVP”. This should NOT be used to treat bleeding because this particular brand is too dilute. The brand “STIMATE” must be used.)
- PLATELET TRANSFFUSION - Some of the more severe types of PFD, including Bernard-Soulier syndrome or Glanzmann thrombasthenia, may not respond to DDAVP and may require treatment with platelets for bleeding or before surgery.
- AMICAR - This medication is used to treat mouth bleeding (and sometimes to treat nosebleeds) along with DDAVP or Stimate. Amicar works to stabilize the blood clot in the mouth or nose, to allow the area to heal and to prevent rebleeding. It is used four times per day for several days. Amicar or tranexamic acid (Lysteda) can also be given for heavy menstrual bleeding.
- OINTMENT - For frequent nosebleeds, Vaseline applied to the nasal passages may help prevent
We’d Like to See You When You Turn 18

Turning 18 marks an exciting time in your life. You may be graduating from high school and looking forward to college, technical school or a new job. You may be moving into your own apartment, ready to begin your life as an independent adult.

It is also time to take ownership of your health and learn to manage responsibilities that were previously handled by your parents. One of those responsibilities is managing your bleeding or clotting disorder.

Now that you are an adult, we would like to see you in clinic to make sure you have all the information you need to take good care of yourself through the coming years. For patients who have hemophilia or type 2 or 3 von Willebrand Disease, we will automatically schedule a clinic visit for you. For patients with other bleeding or clotting diagnoses who are age 18 or older and have not yet scheduled a clinic visit, please call us at (414) 257-2424 so that we can set up a visit at a time that is convenient for you.

drying of the nasal tissues. Saline nasal spray or saline gel (Ayr gel) may also help to keep the nose moist.

• **BIRTH CONTROL PILLS** - Some women with PFD may have heavy menstrual bleeding. Birth control pills may help control the heavy bleeding.

• **RECOMBINANT FACTOR VIIa** - For patients with more severe PFDs (Glanzmann or Bernard-Soulier) who do not respond to treatment with platelets, recombinant factor VIIa is usually helpful. It is a genetically engineered activated clotting factor that may bypass the need for platelets to help form a stable clot.

**Things to Remember**

• Avoid aspirin and ibuprofen products! Learn to read medication labels. Ask your pharmacist.

• Preventive dental care can decrease the need for dental surgery.

• Carry identification to alert medical personnel to your specific bleeding disorder.

• Always wear a helmet when biking, skating, or participating in activities with a risk of head injury.

**When to Call Your Hematologist**

• Before any surgical procedures, major dental procedure or extraction, you may need pre- and post-procedure treatment to prevent or control bleeding.

• In the event of serious injury.

• If experiencing nosebleeds that are difficult to control or occur very frequently.

• If experiencing other bleeding that seems prolonged (i.e., mouth bleeding, cuts, etc.)

• If experiencing heavy menstrual bleeding, i.e., longer than seven days, or requiring a change of protection more often than every two to three hours.

• With any questions or concerns.
Meet Our New CCBD Staff

Hello, my name is Margaret Thew and I am happy to be joining the Comprehensive Center for Bleeding Disorders as a Nurse Practitioner. I look forward to getting to know all the patients, their families, and the staff involved with CCBD. I will be working directly with patients to evaluate and coordinate surgeries and develop educational materials. I will work with and support the entire CCBD team to deliver high quality, exceptional patient care.

I received my Master’s degree as a certified Family Nurse Practitioner from the University of Wisconsin-Milwaukee in 1997. I have worked in various roles as a Family Nurse Practitioner at Children’s Hospital, the Medical College of Wisconsin, and ProHealth Care. When I am not working, I enjoy coaching volleyball and spending time with my family. I look forward to working with the patients, families, and staff at CCBD.

Hello, my name is Michelle Zoerb and I am excited to have joined CCBD in September as a Social Work Intern. I am working on my Master’s degree at the University of Wisconsin-Milwaukee and will be with CCBD one day a week until May 2012. I am thrilled to have the opportunity to learn from Jane Volkmann (Supervisor-Social Worker) and Sheri Robbins (Social Worker/Financial Counselor), as well as the many other skilled staff at CCBD. During my time here, I will be participating in comprehensive clinic appointments with patients with bleeding disorders, as well analyzing ethical issues that may impact the care of patients and families affected by hemophilia. I am also currently interning with another BloodCenter program at the Wisconsin Donor Network. Additionally, I have worked as a Quality Assurance Care Coordinator in the Wraparound program at My Home, Your Home for five years, and continue to work there part-time. I look forward to meeting and talking with many of you over the upcoming months.

Holiday Product Ordering & Delivery Schedule – CCBD Holiday Hours

Since the holidays are fast approaching it seems like a good time to address product delivery from BloodCenter of Wisconsin. Please be aware of your inventories (product and supplies) at all times, and double check them on a weekly basis to assure proper inventories. You should never have less than two to three doses on hand at any time. At this time of year, place product orders when you have four to five infusions remaining at home, as delivery times may not be running as usual. And if you are going on vacation, please order enough for the ENTIRE vacation, PLUS another three infusions.

Place your order at least two days prior to the desired delivery date, taking into consideration any upcoming holiday or weekend. There is a lot of background information needed to process your order and waiting until the last minute may delay your delivery. (Although BloodCenter of Wisconsin drivers can make next day or same day deliveries in emergency/life threatening occurrences, this service should never be used as a convenience)!

Keep in mind there are no deliveries on the holidays or on weekends. Additionally, CCBD is closed on the following days with no staff available to take any orders:

• Thanksgiving and the Friday after (November 24 & 25)
• Monday, December 26, 2011
• Monday, January 2, 2012

Also, please let us know of any changes in your insurance as soon as they occur. If you have any questions please call your patient services coordinator at 414-257-2424.
HAVE YOU MOVED?
Please complete the form below and return to us at the Comprehensive Center for Bleeding Disorders, PO Box 2178, Milwaukee, WI 53201-2178. It is important that we keep our mailing lists current so that you can be sure to receive current medical information along with announcements regarding our special medical programs.

Patient Name: _____________________________

DOB: _____________________________

New Address: __________________________________________

City, State, Zip Code: __________________________________________

New Phone Number: _____________________________

New Dentist or Primary Doctor: _____________________________

Office Phone Number: _____________________________

Is this the address of patient’s:
MOTHER     FATHER  BOTH

HAVE YOU RECENTLY TURNED 18 YEARS OLD?
Check the appropriate boxes telling us how we may contact you and who we may speak with regarding your medical care and return it to us at the Comprehensive Center for Bleeding Disorders, PO Box 2178, Milwaukee, WI 53201-2178. Because you are legally an adult, CCBD cannot speak to anyone but you regarding your medical care without your authorization.

I authorize CCBD staff to:

☐ Contact me at my work phone number:

(Detailed messages will not be left)

☐ Leave a detailed message on my home phone/voicemail:

☐ I authorize CCBD staff to speak or leave information with person(s) in my home as follows:

Name / Relationship to Patient

Name / Relationship to Patient

Patient Name: _____________________________

DOB: _____________________________

Patient Signature: _____________________________

Date: _____________________________

WE WOULD LIKE YOUR INPUT

Remember, this is your newsletter. We welcome any requests you might have for future articles. Just give us a call and let us know of your ideas, suggestions or call and let us know if you feel we are providing you with an informative newsletter.
BloodCenter of Wisconsin advances patient care by providing life-saving solutions grounded in unparalleled medical and scientific expertise.