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Hemophilia society warns MDs: bleeding disorders not a male domain

Barbara Sibbald

En bref

Bien des femmes affligées de troubles héréditaires de saignement sont frustrées par les diagnostics erronés et le scepticisme des médecins. La Société canadienne d'hémophilie tente de faire connaître les deux principaux problèmes, la maladie de von Willebrand et les troubles de fonction plaquelettaire, qui touchent des milliers de Canadiennes.

Ania Szabo bled for 4 weeks after a miscarriage. She kept returning to a Toronto emergency room, where staff kept upping the ante. "When you soak a pad in 2 hours, 1 hour, half an hour, 20 minutes — then we'll do something," she was told. She told them repeatedly that she had von Willebrand's disease and needed to see a hematologist. Finally, she was referred.

The hematologist told her the referring physician had said Szabo didn't really need to see a specialist — she just needed reassurance. "He was very angry because he knew I needed drugs," said Szabo. He determined that Szabo had type 3 von Willebrand's disease — the most severe form. It was a diagnosis — and experience — that launched Szabo's involvement in the Canadian Hemophilia Society (CHS).

She is the newest member of a CHS task force that is attempting to educate and raise awareness about women and inherited bleeding disorders. "Of all the pain and physical trouble I've had, especially as a kid, the thing about not being believed hurt the most," says Szabo.

Many people associate bleeding disorders with hemophilia, a predominately male disease, and think women do not experience these problems. "We have to get past the mind-set that this only happens to men," says Dr. Sara Israels, a pediatric hematologist and oncologist in Winnipeg.

This assumption, coupled with the fact that many of the signs relating to bleeding disorders can be mistaken for gynecologic problems, leads to frequent misdiagnosis. "One of the major presentations is anemia associated with heavy periods," says Israels. "Physicians immediately think this is a gynecological issue."

Undiagnosed bleeding disorders can have horrific repercussions: periods that last 37 days, multiple transfusions, serious problems during surgery and hemorrhaging after birth. It can be life-threatening for young women at menarche. Nineteen-year-old Connie Bielinski of Toronto once needed 2 blood transfusions after her period. But the real horror stories come from older women who had scant hope of diagnosis and few treatment options. Some underwent hysterectomies to stop heavy bleeding. Szabo says at least 4 of the 12 women attending her first society workshop had undergone hysterectomies because of heavy bleeding. "I was shocked by the stories," she says. "Women with towels between their legs, passing out at school, all sorts of horror stories."

Tom Feiler photo

Ania Szabo has type 3 von Willebrand's disease. Her children Luke, 2, and Ela, 4, have the milder type 1 form of the disease.

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Women can experience 2 broad types of bleeding disorders: von Willebrand's disease, and platelet-function disorders such as deficiencies of fibrinogen or of factors V, VII or XI; as well, half of all carriers of hemophilia A or B have levels of factors VIII or IX that are below the normal range, and about 20% of these patients have levels low enough to cause symptoms of a bleeding disorder.

Von Willebrand's disease, named after the Finnish physician who first described the problem in 1925, is the most common inherited bleeding disorder and affects up to 1% of the total population (male and female), or about 150 000 Canadian females; hemophilia affects roughly 1500 Canadian males. Von Willebrand (vW) Factor is a protein critical to the initial stages of blood clotting; it helps platelets adhere to the site of injury and also carries and stabilizes Factor VIII.

People with von Willebrand's disease have insufficient or abnormal vW Factor. Type 1 disease, which accounts for about 80% of confirmed cases, is usually considered mild — these patients produce 25% to 35% of normal levels of vW Factor. Type 2 disease involves an abnormality in the vW Factor itself, while type 3 is the most severe type; these patients produce levels of vW Factor that range from less than 5% of normal to undetectable. Symptoms can be similar to hemophilia, and include joint bleeding.

Despite the prevalence of von Willebrand's disease, only 800 Canadians are registered in the von Willebrand registry run by Dr. David Lillicrap of Queen's University in Kingston, Ont. Hematologists speculate that many type 1 or type 2 patients may be unaware they have the disorder.

Platelet-function disorders may be as common if not more common than von Willebrand's disease, says Israels. Unfortunately, testing for the former is less certain and somewhat more complicated than for the latter, and some parts of Canada don't have the laboratory facilities to diagnose platelet disorders. A registry for these patients is being established by Dr. Margaret Rand at Toronto's Hospital for Sick Children.

Resources available for physicians, patients

A pamphlet on platelet function disorders is available through local hemophilia clinics or from the Canadian Hemophilia Society, 625, ave Président Kennedy, bur1210, Montréal, Que H3A 1K2; 800 668-2686.

Related Web sites have been developed by the World Federation of Hemophilia, **www.wfh.org/von_w.html**, and the National Hemophilia Foundation, **www.infonhf.org/bleeding_info/willebrand/**.

Diagnosing a bleeding disorder

A detailed history can reveal signs that a patient may have a bleeding disorder. These signs can include a family history of bleeding problems, easy bruising, swollen, painful joints, prolonged bleeding after a dental extraction or minor surgery, frequent nose bleeds, unusual bleeding from the gums, and an unusually heavy period when there is no apparent cause, such as an intrauterine device. ("Unusually heavy" means a period that lasts more than 7 days or soaks more than 1 or 2 pads or tampons every 2 to 3 hours.)

Physicians in major centres should refer these patients for specialized hemostatic testing, said Dr. Sara Israels, a pediatric hematologist in Winnipeg. In smaller centres or rural areas, screening tests including prothrombin time (PT), partial thromboplastin time (PTT) and bleeding time may be done, but they are not refined enough to detect all bleeding disorders. In other words, they may result in false negatives. "If these patients have a significant history, they should get a blood work-up," Israels says.

Half of all carriers of hemophilia A or B have levels of factors VIII or IX that are below the normal range, and about 20% of them have a level low enough to cause bleeding-disorder symptoms. "Compared to hemophilia, we're talking about a huge population," says Israels.

But the biggest problem remains initial diagnosis: deciding to do the test. "You can't diagnose something you don't think about," Israels points out. "Being aware of the possibility is the key."

Because of advances made in the last decade, testing techniques and treatment at Canada's 23 bleeding-disorder clinics and other centres has improved. Treatment for acute bleeding may include the use of desmopressin, a synthetic hormone that works by causing vW Factor to be released from storage sites in the lining of blood vessels. For persons who do not respond to it, clotting-factor concentrates are the preferred treatment; responding to concern about potential HIV or hepatitis infection, the hemophilia society says these products "are screened for viruses such as hepatitis and HIV and treated with heat or chemicals to further purify them." As well, oral contraceptives or other hormonal therapies can cut blood loss during menses.

"Bleeding disorders are so common, especially in their mild forms, and the repercussions of not knowing are horrible," says Szabo. "Physicians need to be more aware." ⁇