HEMOPHILIA B:

HER VOICE, HER LIFE

B2B

Your Voice. Your Life.

FOREWORD

The B2B (hemophilia B patient to hemophilia B patient) program began in 2005. It was created so that people who have hemophilia B could speak to others with hemophilia B with the goal of helping each other learn how to live with this bleeding disorder.

The program's original mission remains the same today:

- ✔ **Help people with hemophilia B address their challenges**
- ✔ **Strengthen the support system and educational network in the hemophilia B community**

These books were created to provide information and tips about living with hemophilia B from people who are doing it every day. The books contain personal stories from people in varying life stages from infants to mature adults, parents of children with hemophilia, and other members of the family. The books also address topics like personal empowerment and include insights from health care professionals who treat children and adults with hemophilia B.

This 10th book in the B2B series, titled *Hemophilia B: Her Voice, Her Life,* was created to build awareness, understanding, and acceptance of women in the hemophilia community. It addresses women diagnosed with hemophilia B, their struggles with validation and treatment, and what they envision for the future standard of care.

Hemophilia B: Her Voice, Her Life also explores some issues women face when they are carriers of the hemophilia B gene, what that means for their health and family planning, and the emotional impact of carrier status. It discusses men with hemophilia, focusing on the need for them to become educated about women's issues and on the importance of uniting the hemophilia B community around these topics.

Too often, hemophilia is thought of as only a male condition. Over the past several years, the topic of women with hemophilia has been receiving more attention in the medical community. This book aims to educate and encourage women to seek a diagnosis if symptoms and family history are present in their lives.

Hemophilia B: Her Voice, Her Life provides a platform for members of the hemophilia B community to share their stories and aims to shine light on a segment of the community that historically has been underserved. Members of the hemophilia B community and our own B2B Advisory Board offer thoughts and speak about their experiences as women in the hemophilia B community.

To those who shared their experience and knowledge about life as a woman or as an advocate of women with hemophilia B, we are greatly appreciative: Kirstin D., Nina D., Shelby S., Elizabeth V., Becky V., Pam W., Rocky W., and Dr. Robert Sidonio.

I think women need to feel like the health concerns they have are valid and that they aren't alone. Women need to know how to go about getting tested, what questions they need to ask, and what kinds of information they need to be collecting to help their doctor make an accurate diagnosis and determine appropriate treatment.

-BECKY

Daughter has hemophilia B

The views and opinions expressed in this book are those of patients, parents, and hemophilia care specialists who are members of an advisory board within the hemophilia B community and not of Pfizer Inc.

The information in this book should in no way replace the advice of your health care professional (HCP). Be sure to talk with your HCP, nurse, or hemophilia treatment center (HTC) staff regarding any form of medical advice or treatment.

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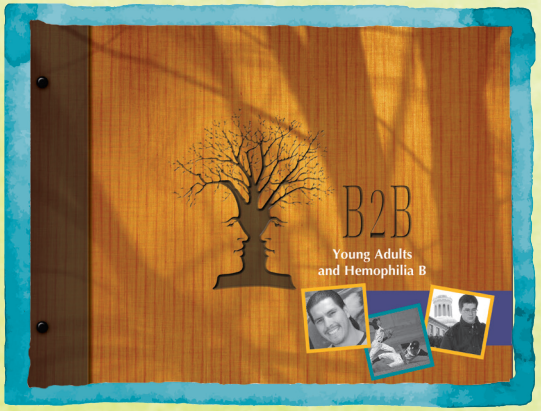
Throughout the book, professional input is provided by Dr. Sidonio, a faculty member at Emory University Medical Center. Dr. Sidonio's research focuses on managing bleeding disorders in women.

Hemophilia of Georgia Clinical Scientist Award
HTRS MRA Scholar
Assistant Professor of Pediatrics
Emory University/Children's Healthcare of Atlanta

Medical suggestions and recommendations offered by Dr. Sidonio throughout this book are for awareness only; consult your doctor for treatment recommendations that are specific to you.

As part of the B2B program, 9 books have been developed:

- *Young Adults and Hemophilia B*
- *Learn From Experience: A Guide for Mature Adults*
- *Navigating the Preteen Years*
- *Hemophilia B in Early Childhood*
- *Hemophilia B: Your Point of View*
- *Many Faces of Hemophilia B: Challenges and Opportunities*
- *Hemophilia B: A Family Perspective*
- *Hemophilia B: Paths to Empowerment*
- *Know Your Numbers: Knowledge Is Power*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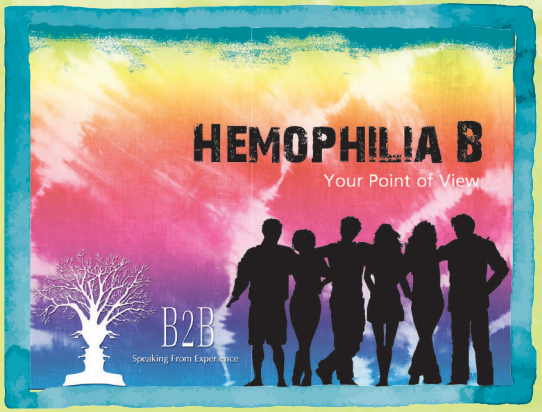
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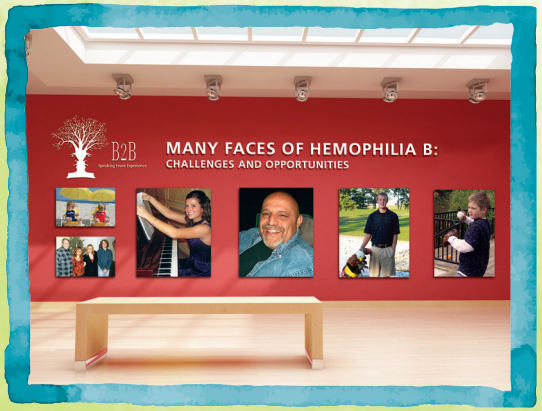
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BRINGING WOMEN TO THE FOREFRONT OF HEMOPHILIA B CARE

INTRODUCTION

As awareness regarding manifestations of hemophilia in women is increasing, it is becoming clear that education on this diagnosis is needed in the hemophilia community.

Historically, hemophilia was believed to affect only males. Because more women are now being diagnosed with hemophilia, there is an opportunity to hear their voices as they share their stories.

The good news is that women with hemophilia B and their families can become active participants in the care they receive. This can help reduce the feeling of being out of control that may be common with a chronic condition.

Living with hemophilia B can be challenging. Bleeds, pain, and, in some patients, musculoskeletal challenges, which can interfere with the most basic activities of life: school, work, recreation, play, or sports, not to mention the challenges girls and women face in puberty and childbirth.

Q&A with Dr. Sidonio

Q: As we create new patient education programs to support women with hemophilia, we want to rally around a mission statement. In your opinion, what should be our mission or objective?

A: The mission should be to support women at risk or already diagnosed to advocate for themselves through education and good communication with their providers and local hemophilia treatment center (HTC), and for appropriate treatment and resources.

In the past, many women may have felt that they are on their own with respect to their bleeding symptoms. The truth is that some women with hemophilia can experience the same kinds of symptoms that men with hemophilia do. However, because it has long been thought that women could only be hemophilia carriers, some women have had the extra burden of convincing health care professionals (HCPs) that their hemophilia diagnosis is legitimate. In fact, there have even been times when women have been discounted with respect to their symptoms as well as their feelings.[1]

This burden has been the experience of some of the women B2B Advisory Board members as they describe living with hemophilia. This book provides information on what to do when diagnosed with hemophilia B; struggles with menstruation, pregnancy and childbirth, and what it means to be a carrier of hemophilia; and how men with hemophilia can help the women in their lives.

Understanding hemophilia and how it affects the lives of everyone can help

- Bring a sense of control and balance
- Improve coping skills
- Assist in better management of hemophilia B because of improved communication with and assistance from HCPs
- Define desired goals and what needs to be done to achieve them
- Raise awareness and self-esteem, and instill a feeling of success and accomplishment

A breakthrough would be a woman not receiving any questioning about her diagnosis with a bleeding disorder.

-ELIZABETH
Has hemophilia B

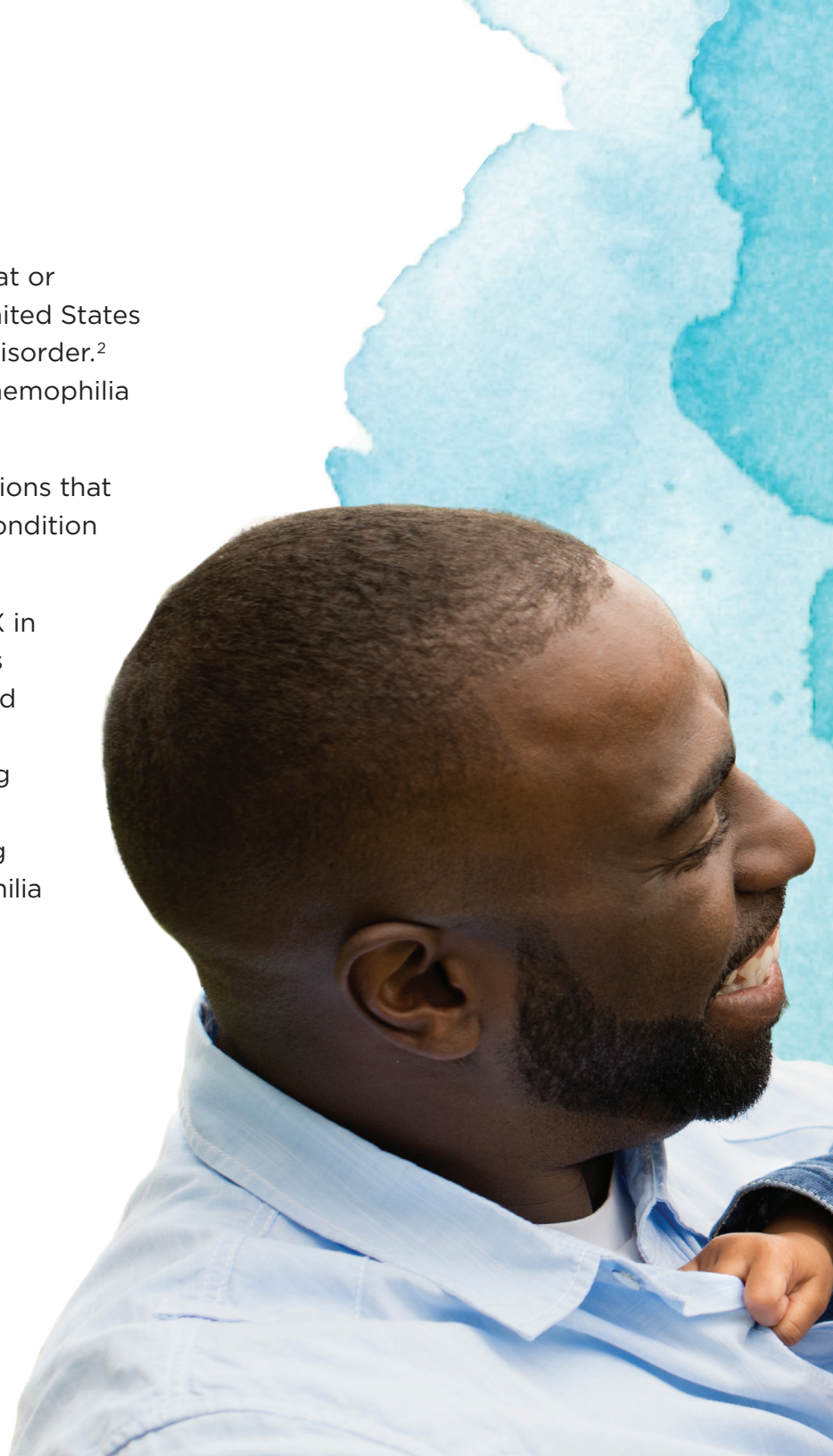
HEMOPHILIA B

A Quick Review

Hemophilia is a congenital bleeding disorder, which means it exists at or before birth and is usually inherited. About 20,000 people in the United States have hemophilia. Each year, another 400 babies are born with the disorder.[2] Most cases of hemophilia are hemophilia A; about 20% of cases of hemophilia are hemophilia B.

The term *bleeding disorder* refers to a wide range of medical conditions that lead to poor blood clotting and continuous bleeding. This type of condition may also be referred to as a coagulopathy or clotting disorder.

People with hemophilia do not have enough clotting factor VIII or IX in their blood; sometimes, they do not have any. The missing protein is called a clotting factor, which works together with other proteins and platelets to help blood clot. Platelets are small blood cells that are formed in the bone marrow. When blood vessels are injured, clotting factors help the platelets stick together to plug breaks in the blood vessel at the site of the injury to stop the bleeding. Without clotting factors, blood clotting cannot take place.[3] When people with hemophilia are injured and bleeding occurs, the bleeding takes longer to stop.[2]



There are two main types of hemophilia.

Hemophilia A

Hemophilia A is the most common type of hemophilia.[2] The blood has little or no functional clotting factor VIII. About 8 out of 10 people with hemophilia have hemophilia A.[3]

Hemophilia B

Hemophilia B is the second most common type of hemophilia. It is also known as factor IX deficiency or Christmas disease. The blood has little or no functional clotting factor IX.[4]

Being a female with hemophilia requires that I am fully educated about my condition, so I can help to educate others well.

–KIRSTIN
Has hemophilia B

How Hemophilia Is Diagnosed

Hemophilia is a blood disorder that can range from mild to severe.[3] The HCP performs a blood test to measure the level of circulating factor IX activity in the blood. Table 1 shows how the severity of hemophilia B is categorized based on the results of clotting factor tests (also called assays).[4]

Table 1. Levels of Factor IX in the Blood of Normal People and People With Hemophilia of Different Severities

SEVERITY[4]	LEVEL OF FACTOR IX IN THE BLOOD[4,5]
Normal (person who does not have hemophilia)	40% to 150%
Mild hemophilia	5% to less than 40%
Moderate hemophilia	1% to less than 5%
Severe hemophilia	Less than 1%

Mild Hemophilia B

People with mild hemophilia B (5% to 40% factor level[5]), about 30% of the hemophilia B population, usually have problems with bleeding only after serious injury, trauma, or surgery. In some cases, if there is no family history of hemophilia, mild hemophilia B is not discovered until an injury, surgery, or tooth extraction results in unusual bleeding. The first episode may not occur until adulthood.[4]

Moderate Hemophilia B

People with moderate hemophilia B (1% to 5% factor level), about 40% of the hemophilia B population, tend to have bleeding episodes after injuries. They may also experience occasional bleeding episodes without obvious cause. These are called spontaneous bleeding episodes.[4]

Severe Hemophilia B

People with severe hemophilia B (less than 1% factor level), about 30% of the hemophilia B population, can have bleeding following an injury and may have frequent spontaneous bleeding episodes, especially into joints.[4] Left untreated, severe hemophilia B causes severe bleeding throughout life.[5]

Important Considerations When Caring for a Person With Hemophilia B

Prevention of bleeding episodes should be a primary goal. A secondary goal involves treating bleeding episodes early and aggressively.[6]

Standard treatment is infusion of factor IX concentrates to replace the defective clotting factor. The amount of factor IX infused depends on the severity of bleeding, the site of the bleeding, and the weight and height of the patient.[7]

Following an infusion, the HCP may perform a blood test to measure the level of circulating factor IX activity in a patient's blood, also called recovery.[8]

I think the one thing that makes me feel validated is that I do have a specific diagnosis of severe hemophilia B. If I was classified as a symptomatic carrier as many women are, I would want to be diagnosed with mild, moderate, or severe, if that was necessary. That way I could receive the care that I need through factor replacement.

-ELIZABETH

Has hemophilia B

WOMEN WITH A DIAGNOSIS OF HEMOPHILIA B

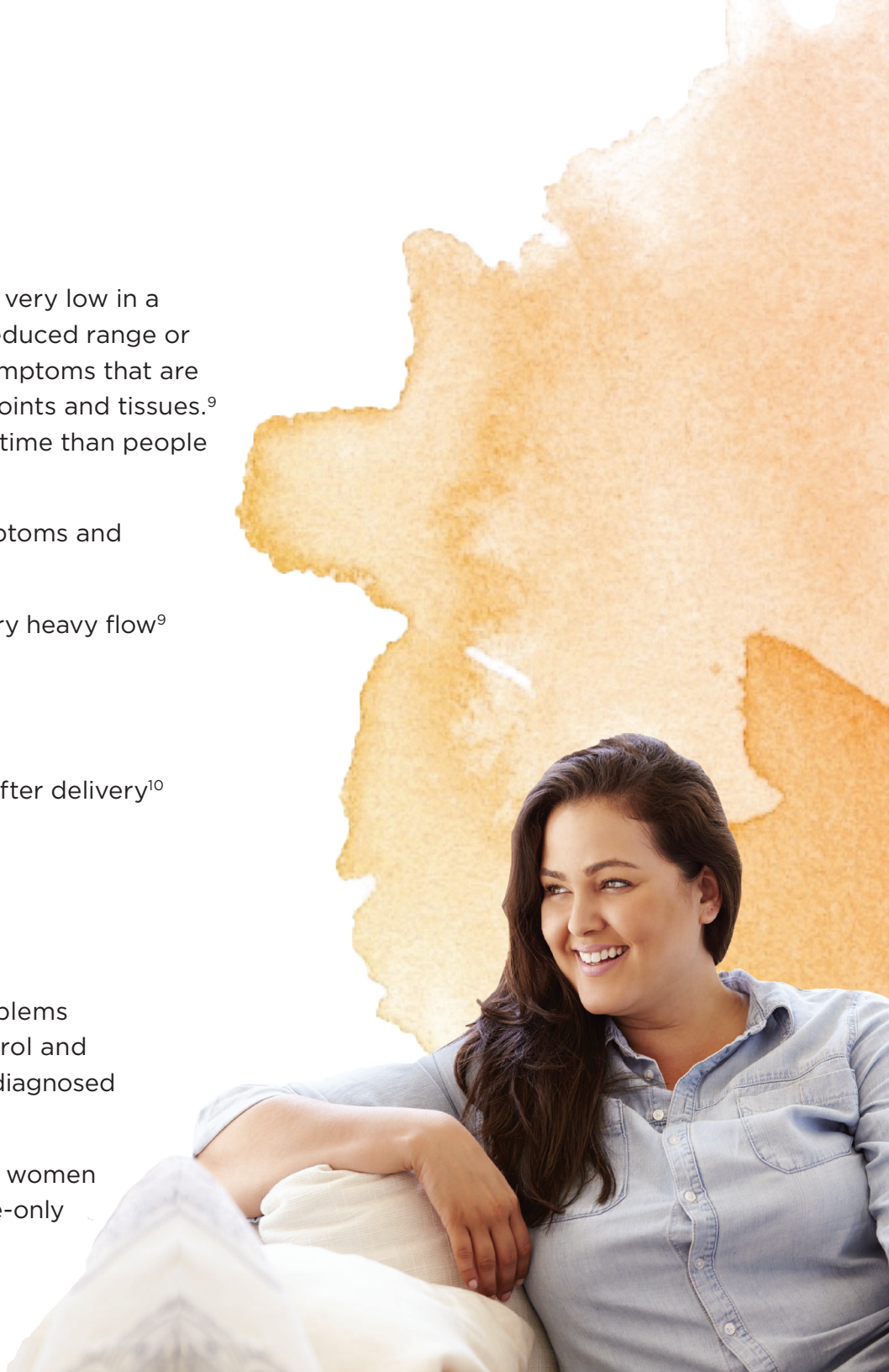
Depending on the factor IX level, which occasionally can be very low in a woman with hemophilia B (but more often is in the mildly reduced range or is normal), some women with hemophilia B contend with symptoms that are identical to those experienced by men, such as bleeds into joints and tissues.[9] Both men and women with hemophilia B bleed for a longer time than people who do not have the condition.[2]

Women with hemophilia also may have a unique set of symptoms and may encounter management issues, such as:

- Menstrual periods that last longer than 7 days with a very heavy flow[9]
- Passing menstrual blood clots larger than a quarter[9]
- Changing a tampon or pad at least every 2 hours[9]
- Postpartum bleeding that lasts longer than expected after delivery[10]
- Prenatal testing for hemophilia[11]
- Ensuring that forceps are not used in childbirth[10]
- Avoiding an epidural and unnecessary episiotomy[10]

Heavy menstrual bleeding is one of the most common problems women report to their doctors. A Centers for Disease Control and Prevention (CDC) study found that bleeding disorders were diagnosed in nearly 11% of women with heavy menstrual bleeding.[12]

It is important to understand the impact of hemophilia B on women and to challenge the misconception that hemophilia is a male-only disorder. These issues affect the level of medical care and emotional support that women receive.[1]



For people diagnosed with hemophilia B, the deficient or missing blood protein is called factor IX. The gene for factor IX is found on the X chromosome, which is a sex chromosome; this is why hemophilia B is called a sex-linked disorder.[4] Men are more likely to be affected by hemophilia because they carry a single X chromosome as opposed to women who carry two chromosomes. This is significant because having two X chromosomes means that women have two copies of the genes that are found on that chromosome whereas men have only one copy. In women, therefore, if one X chromosome carries an abnormal factor IX gene, it is likely that the factor IX gene on the other chromosome is unaffected.[4]

Bringing Women to the Forefront of Hemophilia B Care

Until recently, most women in the hemophilia B community have been labeled simply as carriers. However, many carriers have bleeding symptoms. There is a movement by the National Hemophilia Foundation, called the V4W Health Initiative, to upgrade the diagnosis of carrier to mild hemophilia when qualifying symptoms are shown and laboratory values are present. Depending on the level of factor IX, moderate or severe hemophilia diagnoses are also possible.[9]

Q&A with Dr. Sidonio

Q: What is something about women with hemophilia that you wish the hemophilia community knew?

A: I wish there was more clarity from our governing bodies regarding the correct terminology to use for hemophilia carriers. To me, a hemophilia carrier with normal factor levels (>50%) should be called a symptomatic hemophilia carrier, and a female with a level in the hemophilia range should be called a hemophilia patient. For example, a hemophilia carrier with a factor IX level of 35% should be called a mild hemophilia B patient; she is also a hemophilia B carrier.

Our first major breakthrough was actually getting the diagnosis of hemophilia B and factor levels. Our second major breakthrough was working with our hematologist to identify my daughter's bleeds and treating them.

-NINA

Daughter has hemophilia B

Advocacy Through Better Communication With HCPs

Because males have been considered as the only people who can have hemophilia, many women have experienced barriers in communication and treatment with their HCPs.

Many HCPs do not have protocols or guidelines for treating women diagnosed with hemophilia. As a result, women may need to have a stronger voice when communicating with their HCPs.

It is important for every woman with hemophilia to enlist a team of doctors that includes a primary care physician (PCP), an obstetrician/gynecologist (OB/GYN), an anesthesiologist experienced in bleeding disorders, and a hematologist who can coordinate care and needs.[10]

One way women can manage their symptoms is by working in tandem with their HCPs, perhaps by using some or all of the following tools[13]:

- Care plans designed for patients to help them facilitate care coordination with HCPs
- Online diaries that help track progress and treatment plans
- Self-monitoring assistance for better symptom accuracy
- Apps for smartphones that allow women to track their hemophilia and hemophilia care

In an emergency situation, how do you convince someone that you are a woman with hemophilia if they haven't realized that women with hemophilia even exist?

–BECKY

Daughter has hemophilia B

I want other women with hemophilia B bleeding symptoms to have access to factor replacement therapies. I have been lucky to see how much they can help, and I want other women to feel the benefits of factor as well.

–ELIZABETH
Has hemophilia B

Know Your Numbers

After diagnosis of hemophilia, women can feel isolated in what seems to be a man's world. Having a care plan to follow can help make life with hemophilia B more manageable. Part of this plan includes a thorough understanding of what the numbers mean.

Numbers such as peak and trough factor levels, baseline factor level, and cholesterol level play a significant part in treatment options. One of the first ways to start this education is to read and understand screening lab tests, then ask questions.

Women can take a part in managing their health care. They can let the members of their health care team know that they desire to be involved. Understanding the numbers will help women make better and more informed decisions in the management of hemophilia B.

Bonding and Hemophilia B

It is important for women to come together as a community and to involve loved ones in their care. Talking about hemophilia and how it affects women's lives may help negate any disbelief about the diagnosis and its impact.

Check out the National Hemophilia Foundation at www.hemophilia.org. HTCs provide information on support groups in your local area and are a resource for volunteering in your community.

For more information on important numbers in hemophilia B treatment, check out the B2B *Know Your Numbers* book, available for download at www.B2Byourvoice.com.

Puberty and Helping Daughters Manage Hemophilia

Puberty on its own can be a difficult experience, but with a hemophilia diagnosis, it can become even more complicated. A girl may start to experience heavier than normal periods (known as menorrhagia), which may be a sign that she should be tested for a bleeding disorder.[14]

Some symptoms of menorrhagia include[14]:

- A menstrual period lasting longer than 1 week
- Changing pads or tampons at least every 2 hours on the heaviest day
- Passing blood clots that are larger than a quarter
- Daily activities affected because of excessive bleeding

Another symptom to be aware of is anemia due to blood loss. If a girl who has recently begun to have heavy periods complains of feeling weak, tired, or has a paleness to her skin, she may be experiencing anemia. She should see her primary care physician and hematologist for further evaluation.[14]

Q&A with Dr. Sidonio

Q: What do you wish women and girls knew about being a carrier?

A: I wish that hemophilia carriers would attend hemophilia clinics with their brothers to learn more about the disorder for support and how it is relevant to them. I think this might motivate more females to get tested and prepare for procedures and pregnancy. I see too many carriers suffer from bleeding because their family was timid about advocating for them.

Tracking With Apps

There are apps for iPhone® and Android™ that allow girls and women to track menstrual flow volume and period duration. The data can help a gynecologist or other HCP determine a baseline and suggest ways to control bleeding issues.

Tips to Help Teens Control Their Menstrual Periods[14]

- Prepare the teen for what she will experience before her first period. Talk with her and give her information on how to manage it successfully
- Make sure she has a personal supply of monthly products as well as a way to carry them with her
- Give her confidence with honest, accurate information about menstruation and the added impact hemophilia may have on it

Camp

There are hemophilia camps for girls that can be a great way to connect with others who have a bleeding disorder. The National Hemophilia Foundation website has a page dedicated to finding a camp that is a good match: www.hemophilia.org/Community-Resources/Locate-a-Camp-Near-You.

Because my daughter had access to treatment, she did not have to endure years of heavy menstrual bleeding or joint bleeds.

-BECKY

Daughter has hemophilia B

Pregnancy and Childbirth: Issues Unique to Women

Pregnancy, childbirth, and passing on the hemophilia B gene to their children are issues women face. This can potentially cause serious problems if planning is not done ahead of time.

It is important to begin addressing potential issues with an OB/GYN before problems arise. This includes family planning and any concerns about children inheriting hemophilia.

During pregnancy, prenatal testing can be performed to determine whether the gene mutation has been passed on to the unborn child. Frank discussions with HCPs are helpful in deciding whether to conduct tests, as they can be invasive and carry some risk.[11]

After my second son was born, I bled for 6 to 8 weeks. I just thought it was normal.

–PAM

Hemophilia B carrier

Treatment Team

Your pediatrician, primary care physician, and obstetrician should be in communication with your hematologist to develop a plan for pregnancy and delivery. Special consideration should be made for receiving enough factor prior to and after childbirth, if it is required. Having team members who have experience with bleeding disorders is crucial.[10,15]

Knowing about a diagnosis of hemophilia ahead of time can help avoid serious problems during delivery. Avoiding the use of forceps or a vacuum extractor during delivery is an important safety measure, especially in the case of a male baby who may have inherited the abnormal factor IX gene and may therefore have hemophilia.[15]

When doctors work as a team, they can form a comprehensive treatment plan for making important health care decisions.

Often, because I'm female, two hematologists in the same practice may have a difference of opinion on how to proceed when issues arise, as some hematologists still have trouble accepting that women bleed and aren't just carriers. I have learned that I need to advocate for myself and align with a care team that supports me.

-KIRSTIN
Has hemophilia B

When considering pregnancy with a diagnosis of hemophilia B, ensure good communication between your obstetrician and your hemophilia specialist. This is the time to discuss issues such as postnatal bleeding and whether to plan a C-section or vaginal birth.

Following birth, the baby can be tested for hemophilia using a sample from the umbilical cord blood. Talk with your HCPs if any of the following applies[15]:

- There is a family history of hemophilia
- The mother is diagnosed with hemophilia or is a carrier
- The child has bleeding issues during delivery

Q&A with Dr. Sidonio

Q: Is it possible for a hemophilia carrier to be at risk for surgical bleeding?

A: Many patients and providers often ask this. My response is that it is not clear how many hemophilia carriers are at risk for bleeding, for example, after a procedure. What we do know is that it is recommended to check the factor level (factor VIII or factor IX) before a procedure, obtain a bleeding inventory (possibly through a bleeding score), and verify hemophilia carrier status genetically. From my work and others, there is definitely an increase in bleeding tendency following procedures, and that risk extends to some carriers with normal levels (>50%).

CHALLENGES FOR CARRIERS OF HEMOPHILIA B

What It Means to Be a Carrier

For years women were known only as hemophilia carriers until women with bleeding symptoms stepped forward.

A carrier of hemophilia (a female who has inherited one abnormal X chromosome) is a woman with the ability to pass the abnormal X chromosome on to her children. Being called a carrier is a genetic designation. There are two types of hemophilia carriers: obligate carriers and possible carriers.[16]

Obligate carriers are[16]:

- Daughters of a father with hemophilia
- Mothers of a son with hemophilia and who have at least one other family member with hemophilia
- Mothers of a son with hemophilia and who have at least one other family member who is a known carrier of the hemophilia gene
- Mothers of 2 or more sons with hemophilia

Possible carriers are[16]:

- Daughters of a carrier
- Mothers of a son with hemophilia but who do not have other family members with hemophilia
- Sisters, mothers, maternal grandmothers, aunts, nieces, and female cousins of carriers

Both types of carriers can be symptomatic. This means they can have the gene mutation as well as hemophilia symptoms.

Carriers face issues that range from physical symptoms, such as bleeds and joint problems, to the psychological impact from hemophilia.[1,9] They should be informed about necessary testing and the importance of building good relationships with HCPs in order to get the best treatment.

Q&A with Dr. Sidonio

Q: What should carriers and women with hemophilia know about their joints?

A: Right now we have only preliminary data about the possibility of hemarthrosis in hemophilia carriers based on range of motion from those enrolled in the Universal Data Collection Project and on a pilot study of carriers with normal levels who underwent an MRI at Vanderbilt University. For those carriers with factor levels in the hemophilia range, the same guidelines as hemophilia patients should be followed and the HTC contacted for concerns about a joint bleed. When the factor levels are higher, it may take visual observation by an HTC professional and potentially ultrasound in a situation in which it is not clear. Documentation and physician physical exam are important to establish whether a joint bleed is occurring. I like to use a simple ultrasound when it is not clear. For long-term repeat joint bleeding in a young adult, an MRI may be helpful but can be difficult to get insurance approval.

I envision us coming together and being recognized as more than symptomatic carriers. We are women with a bleeding disorder—it should be recognized as that. My hope is for education, advocacy, and acceptance.

-KIRSTIN
Has hemophilia B

A Carrier Perspective: Pam's Story

What goes through a woman's mind when she's presented with the diagnosis of hemophilia carrier? Is she surprised, bewildered, or feels affirmed? All of those feelings are justified and possible, and they may be related to whether or not she knew hemophilia existed in her family medical tree. Even then, it can be overwhelming as thoughts and questions arise, such as, "Do I want to get pregnant and have children, potentially passing on the hemophilia gene?" "How do I tell my potential spouse that our children may have hemophilia?" "How do I even explain hemophilia to those who may only remember the Ryan White story and thus see hemophilia as a risk for AIDS and death?" And finally, "How do I explain hemophilia to my potential in-laws and what it means for their grandchildren?" While these questions may seem harsh, they are very real...and reality begins to hit home when you have to face the big picture of hemophilia for your future.

Pam, a hemophilia B carrier

As a child, my diagnosis (and my father's diagnosis) should have come many years before it did. I often bruised very easily and when I reached puberty, my menstrual periods were erratic and extremely heavy. I can remember having to keep extra clothes in my gym locker, just in case something happened. As I entered my 20s, my periods didn't improve, and I often passed large clots and bled for seven or more days. After the birth of my first son, there was a great deal of blood and my son bled longer than usual following his circumcision. Still no medical personnel made the connection or even seemed to worry about it. After the birth of my second son, I bled heavily for 6 weeks postpartum. My midwife didn't seem too concerned: "It's probably just because you went back to work one week after childbirth and your body hasn't had time to relax. You just need to slow down." So I lived with the heavy periods, the ruined clothing and bed linens, and moved on with my life.

I was 37 years old when I received my diagnosis. My father was 62 when he received his hemophilia B diagnosis, following the diagnosis of my sons aged 11 and 6. Our older son had ear surgery, and when the surgeon attempted to remove the postoperative dressing, the ear gushed blood and wouldn't stop. Luckily for us, he worked closely with the hematology department and knew which blood tests to order to confirm our son's diagnosis. The next day, we were admitted to the hospital and given the diagnosis by the hematologist. We were visited by nurses, social workers, and therapists…overwhelming, to say the least. We had no idea that hemophilia existed in our family. Looking back, we should have figured it out. All the signs were there, but we lived in rural Vermont and our family physician was a general practitioner who saw everything from colds to heart problems.

I have to wonder, if I had known in my early years that I was a hemophilia carrier and had understood what that diagnosis truly meant, would I have made the same choices in my life?

Would other women in my situation face the same questions? I asked the question to various mothers in the hemophilia circle and the responses were balanced. Each person, each couple, needs to make the best decision for themselves; raising a child with chronic medical needs can be demanding. If you know in advance, then you can make a more concrete decision.

My advice to a girl who may have a family history of hemophilia is that as she enters puberty and begins menstruation, she should always be aware of how heavy her periods become and talk with another female authority figure. That person could be her mother, her aunt, or even the school nurse. I would tell her to not accept the notion that "All the women in our family have heavy periods; it's nothing to worry about." I would encourage her to seek more definitive answers, to ask questions, to explore her symptoms, and to find someone who will really listen to her concerns.

The Importance of Testing

As can be seen in *Know Your Numbers: Knowledge Is Power*, the best way to address the issue of whether a woman is a carrier or a carrier with symptoms is to have factor level and other relevant tests. This helps set a course of action for treatment and further education.

For example, a woman with a male family member with hemophilia B should consider being tested especially if she is pregnant.

One advantage of knowing a diagnosis of hemophilia B (especially if it is moderate or severe) earlier in life rather than later is to accelerate management through a comprehensive HTC. At the HTC, important decisions, such as whether factor replacement and assessment of joint function are needed, can be made.

I was diagnosed at 10 years of age after a study to find out where hemophilia came from in our family. They believed they were only carrier testing me. They sent me back for tests several times to confirm my hemophilia B diagnosis.

-KIRSTIN

Has hemophilia B

Q&A with Dr. Sidonio

Q: What sort of testing should carriers have?

A: All female relatives at risk for being a carrier should consider genetic testing and definitely obtain a baseline factor VIII or factor IX level (depending on the hemophilia type) prior to undergoing a procedure or starting their first period.

Types of Testing

To accurately assess whether a female is a carrier or a carrier with symptoms, start with a factor IX activity level. In the event that the results do not produce a definite diagnosis, genetic testing may be the best next step.[17]

The next set of tests is molecular testing when the factor IX genotype may be unknown.[18] The test that is performed is called polymerase chain reaction (PCR)/fluorescent DNA sequencing with a hemophilia carrier testing algorithm.

If there is a diagnosis of hemophilia B in a family member, the carrier testing algorithm can be performed. The next step is to learn whether a factor IX gene mutation has been identified before. If not, a factor IX gene mutation screening can be performed.[19]

If prior identification has occurred, a different test can be performed. It is called a factor IX gene known mutation screening. If the family history of a gene mutation is not known, this screening cannot be performed.[19]

When to Test

The Medical and Scientific Advisory Council (MASAC) of the National Hemophilia Foundation recommends that testing be done on females at risk of being a carrier prior to pregnancy. During pregnancy, clotting factor levels could rise, so any testing done while pregnant may be unreliable.[17] Seeking the advice of a genetic counselor with hemophilia experience can be valuable.

Some families with at-risk daughters (for instance, if their fathers have hemophilia B) have them tested at puberty, but others prefer to wait. At a minimum, it is important to have clotting factor levels tested if there is any concern, and particularly if any surgery is planned.[17]

Q&A with Dr. Sidonio

Q: What advice do you have for carriers?

A: All obligate hemophilia carriers should discuss their carrier status with a surgeon, dentist, or primary care provider before undergoing a procedure or prior to pregnancy. Unfortunately, it is still common for obligate carriers to go through pregnancy without consulting their local HTC for advice.

Being a Carrier: Working With an OB/GYN

Pregnancy and childbirth present special circumstances in which a woman needs the support of her health care team. The OB/GYN needs to know a woman's carrier status in order to take the necessary steps for delivery.[15]

If a woman is working with specialists at an HTC, close coordination is critical between those medical providers and the delivery team. The OB/GYN should take safety measures during delivery, such as avoiding the use of instruments like forceps or vacuum extractors, and follow any additional guidance that the hemophilia team provides.[15]

Standards of Care

Women carriers and those with hemophilia B have spent years working within a medical system that does not have a standardized regimen of care for HCPs and HTCs to follow. The principal reason for this book is education, so that the conversation can begin to help women feel a sense of normalcy.

Because not everyone with hemophilia experiences the same types of bleeding events, it is important to increase communication and education so that individualized care plans can be developed. These treatment guidelines for women should include managing their periods, pregnancy, childbirth, breastfeeding, and any related gynecological issues.

Standardized treatment guidelines for proper routine testing and care for dealing with menses, pregnancy, breastfeeding, and gynecological issues would aid in validation of a woman with a hemophilia diagnosis.

-NINA

Daughter has hemophilia B

Q&A with Dr. Sidonio

Q: What can carriers and women with hemophilia do to involve their OB/GYN in their comprehensive care?

A: I think gynecological providers should become integrated into the HTC model if possible, and if not, they should maintain close collaboration with the HTC. Many women's clinics are being established throughout the country in response to the needs of carriers and those with other bleeding disorders like von Willebrand disease. I think the gynecologist can assist in management of heavy menstrual bleeding. We currently have a joint clinic for adolescent girls with bleeding disorders on a different day than our traditional comprehensive care clinic, and the response has been great. I feel we can better address the needs of females with bleeding disorders with a gynecologist available for consultation. The Foundation for Women and Girls with Blood Disorders has completed an inventory of the current services for women with bleeding disorders, and it is searchable.

Puberty and Young Female Carriers

Puberty may be the time when hemophilia symptoms become apparent. Heavy periods and anemia due to blood loss may hinder young female carriers' lifestyles in school and their participation in certain sports. Typically, a period lasting more than 7 days with the passing of blood clots or the need to change feminine products every couple of hours should signal the need for further evaluation.[14]

Puberty is a good time to develop a health care plan and assemble a team to help manage symptoms. Following a young girl's diagnosis of hemophilia, it is recommended that she wear a medical identifier at all times. This is especially important if she participates in sports that could cause a bleeding event, and she is unable to tell emergency responders about her condition.

For cases of very heavy periods, oral contraceptives sometimes can be helpful, and this option should be discussed with a health care team to weigh the pros and cons before trying it. There are also apps for tracking menstrual period flow that can be helpful tools for management and care.

Hemophilia camps also offer a great source of community with other carriers, which may help young girls learn how to be their own best advocate.

Why didn't the aunt or the mom realize that something was wrong when a girl has a very heavy period? Often it's the same kind of bleeding pattern they had. So the only people you ever talked to about your menstrual cycle think it's normal—and it's not.

-MELISSA

Son has hemophilia B

Q&A with Dr. Sidonio

Q: How would you recommend women initiate and continue conversations with medical personnel whom they feel need to be convinced that they have hemophilia?

A: This can be challenging for sure. It is always best to approach in a nonconfrontational way with a respectful tone and by stating the bleeding they have had over their lifetime. Then state recently published data to support this. It may take a few visits and discussions with the nurse and physician. Bring in documentation of bleeding events with pictures and menstrual diaries to make your case stronger. Also it may be helpful to involve the local hemophilia chapter for tips on how to approach the specific physician. In the end, if the provider is not convinced, then consider a colleague who sees more females with bleeding disorders or has some experience with carrier bleeding. This may be the only solution.

My best response is to always educate HCPs on the fact that women can and do have hemophilia—whether that's through explaining my own genetic mutation or not.

-ELIZABETH
Has hemophilia B

Questions to Ask HCPs

After a diagnosis of hemophilia, it can be helpful to prepare a list of questions to ask when visiting a hematologist. The answers to these questions may allow a new patient to understand her condition and what she needs to know to protect herself.

Some questions may include[20]:

- What are the treatment options?
- Are there restrictions on activities?
- Are there any long-term complications?
- Is there a list of genetic counselors to talk with?

This list is not complete, of course, and each patient, including affected children, may have more questions. Compile a comprehensive list before the initial doctor visit to get as much information as possible.

A lot of men with hemophilia don't realize the experiences that many women go through. They don't understand that yes, even though a person is labeled a carrier, she has been through a lot of the same experiences that they've had in their own lives. That education is so important.

–ROCKY
Has hemophilia B

Handling the Psychological Impact of Being a Carrier

Besides physical symptoms, some women may have emotional issues to overcome as they deal with a new diagnosis. An HTC can help with guidance and treatment options as well as education.

Because carriers tend to have heavy periods, young girls may feel the need to isolate themselves from family and friends because they are afraid of a staining accident. Fear of embarrassment can be the driving force behind reduced self-image or confidence.

Young girls may find it particularly difficult because they may not know that heavy periods are not a normal menstrual process. This may keep them from reaching out to an HCP for advice and counsel.

Heavy periods may also affect a woman with a successful professional career. A heavy monthly cycle could cause her to miss work and career opportunities because of the need to take time off.

A support network and educated HCPs may be able to assist women in making informed choices as a carrier when it comes to marriage and family life.

Q&A with Dr. Sidonio

Q: What can you tell us about possible communication gaps between carriers and HCPs?

A: There are some providers who have little experience with women and bleeding disorders and even less with hemophilia carriers, but they are learning that it is possible for a hemophilia carrier to have excessive bleeding. This is a fairly new concept with data published only recently as support. It may take some time for those providers, but I see more and more of them believing the data and reporting back to me as well. Some HTCs are not well funded and are only able to focus on those with severe bleeding disorders. This is not an excuse but a reality. You may need to find a well-organized, well-funded center to help support you and manage your potential bleeding.

When my daughter was diagnosed, we felt very alone. Over the years, more and more women are sharing their stories. Elizabeth is lucky that she was treated for hemophilia from birth. Her life story is so different because she hasn't suffered from severe menstrual bleeding or untreated joint bleeds. Everyone deserves timely diagnosis and appropriate treatment.

-BECKY

Daughter has hemophilia B

COMING TOGETHER AS A COMMUNITY

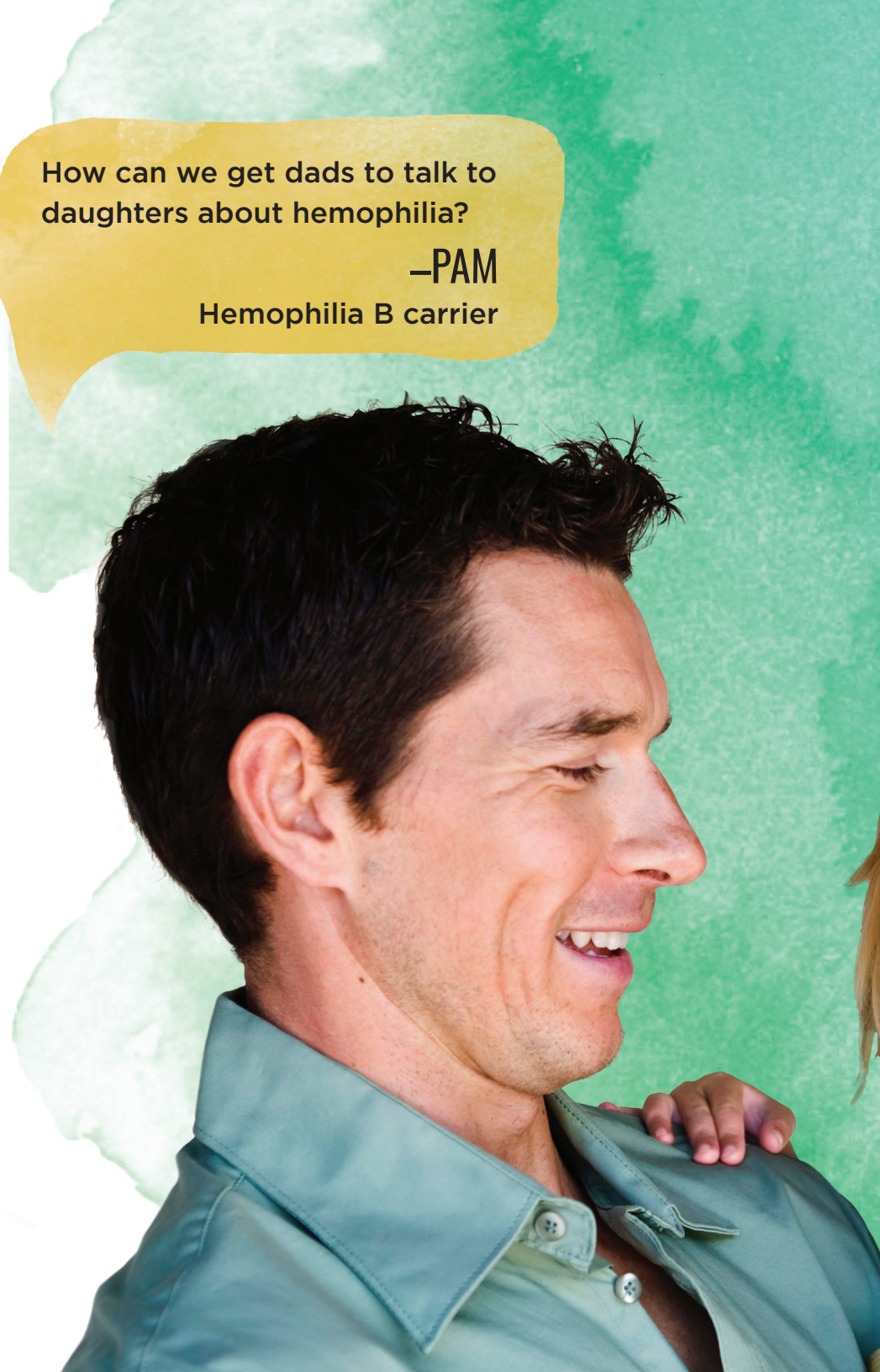
For many years, hemophilia has been a disorder for men, but there is a growing understanding that women are also affected and can struggle with the same symptoms. The hemophilia community has advocated for many things in its history, and now it is time that education about women with hemophilia becomes a priority.

Men with hemophilia should think about all of the women in their lives and have an open dialogue to encourage them to be tested. Better education about women and bleeding disorders can lead to more constructive conversations.

How can we get dads to talk to daughters about hemophilia?

–PAM

Hemophilia B carrier



Men with hemophilia should be educated, and we should be women's champions.

–SHELBY

Has hemophilia B

Community Ties

Although hemophilia is a sex-linked disorder and may be passed down to children via the mother, it affects everyone in a family. And even though more males than females are diagnosed, girls and women share the same concerns and should have a voice in their treatment options.

It is normal for women to feel depressed, stressed, or sad after receiving a hemophilia diagnosis. It is also important to seek help when they feel overwhelmed and to share these emotions with family. This sharing, this community, is what unites everyone touched by hemophilia and helps women's voices be raised in unison.

When women reach out and share their experiences, knowledge, and even struggles, an empowerment occurs that sparks an entire community. That community can be as small as a family unit or as large as a town, region, or state.

Open communication and education will help women with hemophilia flourish and be heard. Sometimes, all it takes is one conversation to open up a world of knowledge and understanding.

It's important to teach carriers that this is real, and in one way or another hemophilia can impact your life.

-ROCKY
Has hemophilia B

In the Women's Corner: A Male Advocate

Several local chapters, including my own Hemophilia Association of the Capital Area, have developed programming to address issues that women with bleeding disorders face. I would advise women to get involved because these local organizations will be their greatest resource for the day-to-day issues of the disorder. The HTCs and physicians are great at setting up robust treatment plans, but other resources may be necessary to address the daily issues and the accommodations that must be made for the bleeding disorder. That's where the community comes in.

Shelby, a male with hemophilia and community advocate

I'm still learning and getting up to speed on the specifics of carriers and women with hemophilia. Largely, I think the community has overlooked the problems or has been dismissive of the seriousness of the disorder. Now, like the rest of the community, I'm learning how bleeding disorders impact women's health and how I can support women. Much of the programming and available literature fails to speak to women, so this should be a starting place for community advocacy. Just as men need emotional, financial, and medical support, women should be able to access these resources that, for me, have been invaluable.

I think our role as men with hemophilia is to embrace these women as we have been embraced.

Helping to secure education and to advocate for drug therapy for women seems a logical and fruitful approach. We must be there to support the women through what I imagine for many may be a difficult acceptance, especially if they get the diagnosis later in life. For most men, the diagnosis came early, and we have never known life outside of hemophilia. For women, the diagnosis is just now coming as science is catching up with the times. I have to imagine that it is difficult to transition from one experience of living without hemophilia to one with hemophilia.

Become a Hemophilia Advocate

Women, men, and everyone else can become advocates to drive education forward and empower those who have been diagnosed. The goal is that no one should have to go through the experience of living with hemophilia alone.

Here are 4 national advocacy groups that support women with hemophilia.

- **The Coalition for Hemophilia B**
  www.hemob.org
- **National Hemophilia Foundation**
  www.hemophilia.org
- **Hemophilia Federation of America**
  www.hemophiliafed.org
- **World Federation of Hemophilia**
  www.wfh.org

Knowledge truly is power when it comes to education about women diagnosed with hemophilia B, and it starts with action and discussion.

Q&A with Dr. Sidonio

Q: What do you envision for the future for women with hemophilia?

A: I think the future is bright for women who are hemophilia carriers. There is more knowledge with the advent of better genetic testing (current testing is >97% accurate) that is widely available through an initiative called My Life Our Future for participating ATHN (American Thrombosis and Hemostasis Network) hemophilia treatment centers and a better understanding of the unique bleeding symptoms. Hemophilia carriers should be better able to advocate for themselves for proper resources. I am also hopeful that fewer hemophilia carriers will undergo procedures without proper planning, which puts them at risk for severe or life-threatening bleeding. It is great that the hemophilia community has been supportive in bringing the issues of our hemophilia carriers to the forefront.

CONCLUSION

The topic of women with hemophilia is a significant one, and it is time for the voices of those affected to be heard. It is important to learn some of the history of women with hemophilia and to discuss the future of this important part of the community. Being tested is the first and most critical step, but there are two other crucial pieces of this puzzle: education and preparation to become the best advocates possible.

HCPs include specialists to help guide women through diagnosis, pregnancy, childbirth, and growing older. Having a preselected team on hand can help women prepare for bleeding events and, in some cases in which the factor IX level is very low, may help prevent joint damage.

Support groups provide community and education to females with hemophilia B and help them to form an important sense of connection, because they do not need to be alone.

This book has brought together parts of the conversation and can be used as a resource for women in the hemophilia B community. It is important to keep the focus on improving the lives of women who need support and education and to promote open communication so that the voices of these women can continue to be heard.

We have often been told that we were mistaken and that our daughter must have von Willebrand disease because "girls don't get hemophilia." When this happens, we just calmly and politely explain that she does indeed have hemophilia, and we explain the chromosomal defect that caused the hemophilia.

-BECKY

Daughter has hemophilia B

Better communication and coordination of care is needed among the specialists (eg, hematologist, OB/GYN, orthopedist, and physical therapist) to attain better health outcomes and consistency in treatment. This is not always an easy task, as it can take days sometimes for the care to be coordinated. Because there are no standard treatment guidelines for females, this can exacerbate the matter. There are not enough specialists who understand treating females with hemophilia. This is probably more complicated when there are no OB/GYNs at adult HTCs who understand their issues.

-NINA

Daughter has hemophilia B

I am a true testament that factor replacement therapies can help when it comes to being a woman with hemophilia. Not only do we have to deal with the joint bleeds that men do, we also have specific issues as women. Factor can help when it comes to having a period and the length and heaviness of it.

-ELIZABETH

Has hemophilia B

RESOURCES

Pfizer Hemophilia Connect

We're committed to helping the hemophilia community.

For the past two decades, Pfizer has been a part of the hemophilia community. We recognize it can be difficult to understand what support is available to help families with bleeding disorders, and to help address this challenge, we created Pfizer Hemophilia Connect.

Pfizer Hemophilia Connect is a one-stop destination to access all of our resources for eligible patients.

Soozie Courter Hemophilia Scholarship Program

Pfizer provides scholarships to students with hemophilia A or hemophilia B who are high school seniors, have a graduate equivalency diploma (GED), or are currently enrolled in an accredited junior college, college (undergraduate or graduate), or vocational school. Awards are based on academics, recommendations, and a personal statement from the student.

Visit www.HemophiliaVillage.com to download an application.

HemophiliaVillage.com

The Pfizer-sponsored website, www.HemophiliaVillage.com, provides information for the hemophilia community. Consumers and professionals alike can find product information and learn about programs and services.

B2Byourvoice.com

The Pfizer B2B Consumer Advisory Board was developed to directly connect Pfizer to hemophilia B patients and caregivers in order to gain firsthand feedback from the hemophilia B community. As a result, the B2B program has created tools and resources to support the community and continues to evolve to address needs as they change.

Visit www.b2byourvoice.com to check out a series of books, videos, patient stories, and other resources for people living with hemophilia B.

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THE COALITION FOR HEMOPHILIA B, INC.

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