

Centerpoints

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Director's Corner



**Stacy E. Croteau,
MD, MMS
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Happy spring everyone and welcome to an exciting edition of our Centerpoints newsletter.

Inheriting a bleeding disorder

Sometimes an individual is the first in their family to be diagnosed with a bleeding disorder and other times there are many family members with the same diagnosis. Understanding how our bleeding disorder is inherited is important, so you and your medical team can understand which other family members may also be at risk of a bleeding disorder or having a bleeding disorder gene. Hemophilia A and B are unique among bleeding disorders, because the genes for these bleeding disorders are on the X chromosome. Since females have two X chromosomes and males only have one, females generally impacted less severely (and in many cases not at all) by a given hemophilia gene, compared to males in the same family that have that hemophilia gene. Recognizing which females in the family may have a hemophilia gene — even if they have normal factor levels and no bleeding symptoms — is necessary, because their future children could be more severely affected. Wondering if you or a loved one may have a hemophilia gene? Speak to your doctor or one

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Connecting the dots:

Lessons learned from a family's hemophilia history

by Emily Williams

At 5 a.m. the morning of May 18, just hours after returning home with their newborn son, the Durkins were doing what most parents ordinarily do — changing a diaper. “I opened his diaper up, and there was all this blood,” remembers Sara Durkin. Panicked, Sara and her husband, Derek, returned to the hospital, where doctors tried to stop the bleeding from baby Ryan's recent circumcision — but it wouldn't stop.

Sara thumbed through Ryan's lab records on her cell phone. It couldn't be, she thought. She knew the blood clotting disorder hemophilia A was part of her family history, but Ryan couldn't possibly have it. It had been ruled out during prenatal or newborn testing...or so she thought. “I was scrolling through the lab work, and I saw the hemophilia test was for factor IX (9),” Sara says. “And I'm thinking, Where's factor VIII (8)?”

It appeared the lab had tested Sara for hemophilia B, associated with low levels of the clotting factor IX protein, rather than hemophilia A, associated with insufficient factor VIII.

With this new information, the Durkins rushed to Boston Children's Hospital. “When we got there, we went to take Ryan out of his car seat and the blood had completely soaked through his diaper and his clothes. It was one of those situations where we had eight members of the emergency room staff suddenly surrounding him. It was very traumatic.”

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Connecting the dots (continued)

Within hours, a team from Urology and Hematology was able to stop the bleeding and put Ryan safely back in his parents' arms. Behind the scenes, Dr. Stacy Croteau, medical director of the Boston Hemophilia Center at Dana-Farber/Boston Children's Cancer and Blood Disorders Center, focused in on learning more about Sara's family history of hemophilia.

"Just from Ryan's short history everything aligned with hemophilia," says Dr. Croteau. "But what I also discovered a strong family history of inhibitors."

Inhibitors and hemophilia

Inhibitors are antibodies developed by the immune system that can target factor VIII. These antibodies "inhibit" clotting factor before it has a chance to work by quickly targeting and destroying it. Typically, inhibitor development is not a major concern in children, like Ryan, who have mild hemophilia. However, Dr. Croteau points out, that for some individuals the specific factor VIII variant puts them at significantly higher risk of developing an inhibitor so "We have to be really careful exposing them to factor VIII concentrates, because we risk turning a patient with mild hemophilia into a patient with severe hemophilia."

With Ryan, it was fortunate Dr. Croteau had his family history of inhibitor development early on, so she could avoid exposing him to factor VIII and work with Urology to treat the bleed using local measures and other supportive medications to control the bleeding. Remarkably, Sara was not overwhelmed when Ryan was diagnosed with mild hemophilia. She's grown up with it, observed how five cousins have managed it, and knows her son can live a normal life. But she wasn't familiar with the risk and impact inhibitors can have for hemophilia patients.

"The fact Dr. Croteau understood my family history and was able to actually inform me about inhibitors, really helped us make a good plan for Ryan moving forward," Sara says. That plan will include a specialized treatment approach, potentially including newly approved therapies or those under clinical investigation. It's part of the benefit of going to a hemophilia treatment center, where the team is experienced in treating bleeding disorders and has access to clinical trials.



Did you know?

Most women who are carrying a hemophilia gene experience no bleeding symptoms but can pass on the gene to their sons and daughters? If hemophilia runs in your family, spread the word. Encourage those who are thinking of starting a family to speak with their doctors about testing for hemophilia. Contact your local Hemophilia Treatment Center with questions.

"We work closely with many local pediatricians, and to some extent other hematologists, especially for folks that live further from the Center," says Dr. Croteau. "We don't want distance to be a barrier to expert care or providers to feel like they're in isolation. Instead, we want to empower and educate the local provider team, so patients can have the best care possible."

Looking back and moving ahead

The Durkins are fortunate to live within close proximity to Boston and to have Ryan under the watchful eye of Dr. Croteau and her team.

In hindsight, Sara wishes she had thought to have her prenatal testing done at Boston Hemophilia Center. "Now I know," she says. "I feel like hemophilia is one of those conditions people know about, but don't have enough information to test properly. Moving forward, I'll make sure my sister gets tested through a Hemophilia Treatment Center and doesn't trust anyone else."

Ryan, who Sara describes as an easy, mellow baby, has not had any complications since that scary day in May. He's "a chill dude, who likes to be held, and he especially loves his big sister, Claire."

Although French Canadian-born Dad, Derek, is was initially disappointed hemophilia will keep Ryan off the ice, he and Sara are okay with that. "Maybe instead of playing hockey, he'll be like me and want to dance, sing or be an actor. Who knows?"

As far as Dr. Croteau is concerned, despite the potential risks, it's important for kids to live their life and embrace adventure. "We have to be cautious but can't keep them in bubbles. We have to let them play, learn and grow."

Learn more about the Boston Hemophilia Center. ♦

Making Healthy Happen



By Kristen Benya, PT, DPT, Physical Therapist, Boston Hemophilia Center

Speaking with patients this year in the HTC has brought to light the challenges of maintaining a healthy lifestyle routine during the pandemic. How do we balance daily activities that improve our physical wellbeing while attending to the demands of work and home life as well as the unique stressors of the pandemic? Resetting our leisure time to include self-care for our physical health can also dramatically impact other aspects of our life, including our mental health and clarity. I'd like to offer up some simple yet effective tools to begin this process and suggestions of different modes of exercise to fit any lifestyle.

One of the most important steps in the process of integrating healthful activity into your life is setting a S.M.A.R.T goal for yourself. S.M.A.R.T is a mnemonic that creates a goal that is Specific, Measurable, Achievable, Realistic, and anchored within a Time Frame. Understanding your reason for setting your specific goal is also a very important piece to this puzzle and an excellent practice in self-reflection. I encourage my patients to verbalize it or write it down to begin their efforts towards it. The more specific and objective the goal, the easier it will be to see those positive changes and appreciate your progress, which can be a huge motivator! Rather than "I want to lose weight", a more achievable and measurable goal could be "I want to lose 5 pounds by the end of the month". This opens the door to a modest goal for which you can begin brainstorming strategies that will get you there like diet and exercise.

Now that you've set a goal, it's time to implement the plan to achieve it. Simple as that, right? Wrong! We've come to the most challenging part in the process and this is often where many people lose their motivation. A simple yet effective way to combat this slump is to find an exercise/activity partner. According to the CDC, when you exercise with a friend you're

more likely to stay motivated, remain consistent, and are even more likely to try new activities than if you were solo. It may take time to find the right partner with similar interests, ability levels, and availability, but once you find the right fit you'll be well on your way to reaching your goal. Remember, our furry friends count too. A daily walk in the neighborhood or local park with your dog is a great way to stay active with an exercise partner who relies on you.

If those obstacles were not enough to overcome, we now have even more pandemic-specific constraints to wellness activities. Many of my patients in the bleeding disorders community still feel uncomfortable attending exercise classes in their community or working out at their local YMCA. Now that the days are getting shorter and the temperatures are dropping, many outdoor activities are also becoming a less attractive option. One positive that has come out of these past two years is the availability of virtual exercise opportunities. This could be a live stream of your favorite local yoga instructor, a free class you found on YouTube, or even a weekly Zoom exercise date with a friend. Using these virtual tools and platforms available to us now is a great way to stay active in the comfort of your own home.

Joint and muscle health are critical pieces of the health and wellbeing of those living with a coagulation disorder. The benefits of light to moderate physical activity to the musculoskeletal and cardiovascular systems have been well documented, with even a little going a long way to establish a healthy lifestyle routine that will have a great impact for many years to come. If you have questions about what type of activity is most appropriate for you, your healthcare team at the HTC is a great place to start the process towards a healthier you. ♦

Director's Corner

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the hemophilia treatment center (HTC) providers to learn more.

Your voice matters

We strive to continue to improve our HTC for all members of the bleeding disorder community. Our goal is to provide exceptional patient care, education, and ease of access to our facilities and services. Your comments and feedback are a critical part of how we can continue to bring you useful and engaging clinical services and educational content. We have a few exciting things in development to help us improve how you can easily share your

ideas with our team. Please be on the look out for information on feedback surveys, formation of a patient/family advisory council, and an electronic suggestion box.

Centers for Disease Control (CDC) Community Counts!

Community Counts is an important national monitoring program, funded by CDC's Division of Blood Disorders, which gathers and shares information about key health outcomes that affect people with bleeding disorders cared for in U.S. HTCs. Boston Hemophilia Center is the regional lead for New England, and all of our New England HTCs participate. The CDC has worked

to increase the ease by which individuals can access these data with the data visualization tool. Our data managers have shared a snapshot from our center later in the newsletter.

We regularly update our website with new research and publications information, communications and educational content. Help us continue to build our Community Corner section by sharing your photos and stories with us; we also want to continue to promote resources and businesses within our bleeding disorder community. Please be sure to bookmark our website for easy access to these materials. Also like us on Facebook (@BostonHemophilia) as we post and link new content there as well. ♦

New Staff



Welcome Jessica!

Jessica Gonzalez joined the team as the Bleeding Disorder team Program Coordinator in January 2022. She graduated from Florida International University with her B.A in Psychology in August 2018 after completing the Pre-Med track. Outside her studies, she volunteered at Baptist Hospital of Miami and Caring for Miami. She moved to Boston after graduating and worked at BWH expanding her healthcare experience as a Medical Assistant.

Jessica was born and raised in Cuba and migrated to South Florida at the age of 10. She is very excited to work with the Bleeding disorder patients and families and research at BHC and hopes to pursue a career in medicine one day. In her spare time, she enjoys traveling around the world and taking photographs.



Welcome Natalie!

The Boston Hemophilia Center welcomes **Natalie Asselin** to our Bleeding Disorders Team in February 2022. She was previously employed as a pediatric primary care nurse practitioner for 5 years at UMass Memorial Children's Medical Center in Worcester, MA. She graduated from the pediatric nurse practitioner program at the University of Pennsylvania in 2015. She completed her nursing degree at UMass Amherst in 2010. She also has experience as a pediatric triage nurse for several years at Pioneer Valley Pediatrics in Longmeadow, MA.

Natalie is originally from Chicopee, MA. She enjoys spending time with family, long distance running, hiking and traveling. She is thrilled to be starting the new bleeding disorders nurse practitioner position at Boston Children's.

Community Corner

The Covid-19 pandemic has been so hard on many small businesses. Our local restaurants have felt the devastating effects of declining numbers of customers and resulting financial losses. The Boston Hemophilia Center cares about our community and economy. In this new section, Community Corner, we highlight two of our Center's family businesses. Please check out these restaurants and their websites. Support our local businesses!

About Distraction Brewing Co.:

Distraction Brewing Co. is a small, family-run craft brewery in Boston, MA. Its charming Roslindale Square storefront, once operating as a bank, now serves up a rotating selection of handcrafted draft and canned beers, its proprietary maltini cocktails, and light fare featuring snacks and bites from some of Boston's best chefs. Since opening in 2019, the 75-seat taproom and outdoor beer garden run by husband and wife duo, Mike and Lora Estey, hosts several rotating pop-up events, entertainment and music. Distraction Brewing is the perfect place for after-work drinks with friends, family and coworkers, and is also available for private events. Distraction Brewing is located at 2 Belgrade Avenue, Boston, MA 02131. For hours of operation or for more information, please call (617) 477-3637 or visit distractionbrewingco.com.