

# Centerpoints

Issue 21/Spring  
2023

## Director's Corner



**Stacy E. Croteau,  
MD, MMS  
Medical Director**

Happy spring everyone and welcome to an exciting edition of our Centerpoints newsletter.

### Gene therapy for bleeding disorders

We are steadily seeing new prophylaxis options for patients with bleeding disorders, specifically hemophilia A & B. The latest addition to our bleed prevention toolbox is gene therapy. The first hemophilia gene therapy product was licensed by the FDA (Food & Drug Administration) in November 2022. This product is intended for individuals with clinically severe hemophilia B (factor IX deficiency). This past summer, a hemophilia gene therapy product for people with severe hemophilia A (factor VIII deficiency) received conditional marketing authorization throughout the EU and is now under review by the FDA.

These gene therapy products work by providing liver cells (hepatocytes) the instructions (DNA—F8 or F9 gene) needed to make the desired clotting factor proteins (factor VIII or factor IX). The F8 or F9 gene are provided through a one-time intravenous (IV) infusion typically performed as an outpatient procedure.

*(continued on page 2)*

## Meet Our New Assistant Medical Director!

Nathan T. Connell, MD, MPH, FACP is Chief of Medicine at Brigham and Women's Faulkner Hospital, Vice Chair of Medicine for BWFH Clinical Services and Education at Brigham and Women's Hospital, Associate Director of the Boston Hemophilia Center, and an Associate Professor of Medicine at Harvard Medical School.



A graduate of Cornell University, Dr. Connell received his medical degree from the University of Miami. He subsequently completed an internal medicine residency at Brown University and Rhode Island Hospital, where he also served as chief medical resident. He remained at Brown to complete his fellowship in hematology and medical oncology while also earning a Master of Public Health degree in Clinical Effectiveness at the Harvard School of Public Health.

In partnership with the American Society of Hematology (ASH), Dr. Connell has helped define the field of systems-based hematology. His research includes the application of decision-analytic models to evaluate the most cost-effective strategies for the diagnosis and treatment of hematologic conditions. He has shown that rapid turnaround of clinical laboratory information significantly reduces unnecessary blood plasma utilization at a health-systems level. He is past chair of the ASH Working Group on Systems-Based Hematology and prior faculty for the ASH Medical Educators Institute. Dr. Connell served as the co-chair of the ASH ISTH NHF WFH 2021 Guidelines on the Diagnosis and Management of von Willebrand disease and has both published and spoken extensively on opportunities to improve the care of those living with inherited bleeding disorders.

Dr. Connell is looking forward to this new role within the Boston Hemophilia Center, saying: "I've been caring for patients with inherited bleeding disorders for many years and have had the opportunity to connect with many in the local community through events with the New England Hemophilia Association. In taking on the Associate Director role, I hope to work with our staff to optimize transition programs, standardize clinical operations, and expand our services to support those living with bleeding disorders."

## Director's Corner

(continued from page 1)

Gene therapy itself is very interesting, yet complex, process. To protect the instructions for the clotting factor proteins while they travel to the liver cells, the gene is "packaged" in what is essentially the shell of a virus. At this time, the most common "packaging" used for hemophilia gene therapy is adeno-associated virus (AAV). While AAV does not make humans sick, it is very effective at interacting with our liver cells and can safely hand off the F8 or F9 gene information. Once the liver cells have the new instructions on how to make factor VIII or IX clotting factor, the expectation is that within a few weeks of the one-time gene therapy infusion, liver cells will begin to produce and secrete factor VIII or factor IX into the bloodstream, which will lead to detectable factor levels and protection from bleeding.

So far, testing of gene therapy products

has focused on adult (>18-years-old) male participants with severe disease (factor levels generally  $\leq 2\%$ ). There are potential restrictions and extra precautions people with liver disease (including history of HIV or hepatitis C or liver fibrosis) must take, making it very important to discuss this topic with your hemophilia provider before considering gene therapy.

To date, gene therapy infusions have been well-tolerated, but some people have experienced reactions to the infusion including: headache, fever, chest tightness, lightheadedness and rash during or shortly after receiving gene therapy. From a safety perspective the most common event that has occurred is a change in ALT (alanine transaminase) enzyme levels in the liver. ALT is a common indicator used in blood monitoring to determine liver health. The increase in ALT is not associated with symptoms but has been noted to occur after gene therapy infusion. In some cases, the increase in ALT requires that a patient

start on a multi-week course of steroids or potentially another similar therapy.

There is a wide range of factor levels that patients have achieved in the weeks and months just after the infusion, as well as the years following gene therapy. Data reported from the most recent hemophilia A and B clinical trials demonstrate that most participants achieve factor levels at least in the mild hemophilia range (5-40%), but some have had normalization of their levels. Others, though initially showing some response, do lose their factor levels over time. The long-term success of gene therapy, as marked by the factor levels patients can maintain over 5 years after the infusion, is still being studied.

You can learn more about gene therapy from your HTC team members. There are also educational resources through WFH, <https://elearning.wfh.org/resource/what-is-gene-therapy/> and others.

## Presentations & Conferences

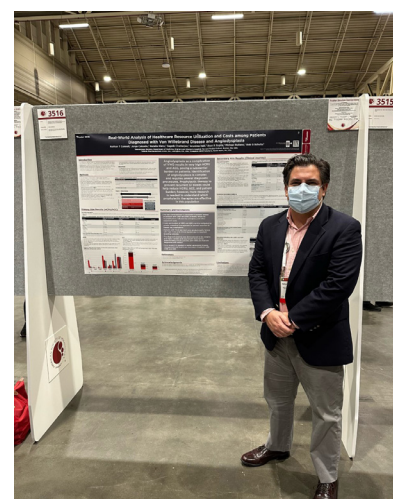
### National Hemophilia Foundation Bleeding Disorders Conference

In August, Amanda Stahl (one of our clinical social workers) and Drs. Nathan Connell and Aric Parnes presented at the National Hemophilia Foundation Bleeding Disorders Conference (NHF) in Houston Texas.

Nathan presented the findings of his paper Real-World Analysis of Healthcare Resource Utilization and Costs Among Patients Diagnosed with Von Willebrand Disease and Angiodysplasia. Nathan and his fellow authors examined the economic and clinical impact of frequent hospitalizations related to patients diagnosed with both von Willebrand Disease and Angiodysplasia. Von Willebrand's Disease is an inherited bleeding disorder characterized by a deficiency (or dysfunction) of von Willebrand Factor; and angiodysplasia is a relatively common source of gastrointestinal (GI) bleeding. The paper's findings point to there being a notable financial cost placed on patient and clinical teams stemming from these paired issues. A cost that could be reduced with steps to better understand these diagnosis and how to better manage patient symptoms in an outpatient setting.

Amanda and Aric presented their paper Characteristics and Prevalence of Posttraumatic Stress Disorder and Posttraumatic Stress Symptoms Among Adults with Hemophilia A and B. Working with the teams at the Boston Hemophilia Center (MA), Mount Sinai Hemophilia Center (NY) and M Health Fairview Center for Bleeding and Clotting Disorders (MN), the paper's authors committed to interviewing patients over the age of 20 about their experiences with PTSD and Posttraumatic Stress Symptoms (PTSS). They found that it was more common for people with hemophilia to experience PTSD or PTSS when compared to the population at large. Leading the authors to suggest additional testing and consultation to help inform patient care.

These are just two of the dozens of papers and presentations produced by the team working with the Boston Hemophilia Center over the last year. And shows our commitment to not only providing the best care to our patients, but by sharing our knowledge with our colleagues across our field.





## Foundation for Women & Girls with Blood Disorders Educational Conference

In September of 2022, Natalie Asselin (Pediatric Nurse Practitioner) attended the Foundation for Women and Girls with Blood Disorders (FWGBD) educational conference held in Arlington, Virginia. The FWGBD works to ensure that all women and girls with blood disorders are given optimal care and are properly diagnosed. They seek to raise awareness among healthcare professionals, foster collaboration between the various disciplines involved in treating women and girls with blood disorders, and share exciting updates and research being done for this patient population.

Reflecting on her experience, Natalie said, "The FWGBD conference was a great weekend of networking and learning from fellow colleagues who specialize in women's care of various blood disorders."

## Community Events

In October 2022, the Boston Hemophilia Center was very excited to have our first in-person community event since the beginning of 2020. We held a private screening of the documentary *Bombardier Blood* and a Q&A session with the movie's subject, bleeding disorder advocate and mountaineer, Chris Bombardier.

The documentary follows Chris, who is a hemophiliac, as he prepares and summits Mount Everest in his then ongoing mission to climb the Seven Summits, which are the tallest peaks on each of the seven continents. Aside from detailing his mountaineering exploits, the film also delves into Chris's experiences with his bleeding disorder, the ways his family has supported him throughout his journey, and his exposure to the precarious circumstances some bleeding disorder patients are born into around the world – which would eventually lead to his advocacy work.

Chris, now the executive director of the international nonprofit Save One Life, provides direct financial aid to patients with hemophilia in developing countries. To learn more about their mission, and how you might help, please visit their website at: <https://saveonelife.net/>

We would like to extend our thanks to Chris and his team for helping to make this community event possible! And would like to encourage those who are able to consider participating in the factor donation program Save One Life operates: <https://saveonelife.net/how-you-can-help/project-share/donate-factor>



## Upcoming Events

This spring the Family Medical Coping Initiative (FMCI) and members of the Blood Disorders Team at the Boston Children's Hospital will be holding an online Fear of Needles (Trypanophobia) information session. The session will introduce participants to the FMCI; strategies on how to discuss a newly diagnosed bleeding disorder; and several possible techniques to reduce fears, increase confidence, and improve the experience surrounding procedures involving needles.

Please stay tuned to our social media (Facebook: @bostonhemophilia) for further information about this and other future educational opportunities!

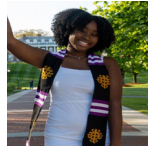


# New Staff



## Welcome Eva!

In June of 2022, **Eva Resto-Estrada** joined our team as a Program Coordinator with the Boston Hemophilia Center at Boston Children's Hospital. After working as a Paraprofessional in the Boston Public School system, she has returned to her roots in healthcare by working with us at the center. She was born and raised in Boston, Massachusetts and received her degree from Southern New Hampshire University. She adores the fall, and in her free time she loves to explore with her dog and husband.



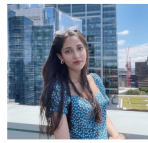
## Welcome Jess!

Also in June 2022, **Jessica Bowman** began working with the BWH Division of Hematology as a Research Assistant, where she plays a critical role in many of our on-going studies. Jessica grew up in Boston, Massachusetts and graduated from Stonehill College with a Bachelor of Arts in Psychology and Anthropology. When not at work she enjoys reading, exploring the town, and playing volleyball.



## Welcome Greg!

**Greg Schiurring** joined the Hematology Division as a Project Manager for the Boston Hemophilia Center in July of 2022. Previously a Research Lab Manager at the Spaulding Rehabilitation Hospital, Greg is excited to join our team and to assist wherever he might. Originally from Austin, Texas, Greg graduated from Texas State University – San Marcos with a BSc in Biology. He is happily learning about- and acclimating to life in New England. Especially our winters.



## Welcome Sana!

In September 2022, **Sana Adil** came onboard as the Boston Hemophilia Center's new Financial Analyst. She came to us from Tufts Medical Center, where she had worked in Hospital Administration for the last 2 years. Sana has a BSc in Healthcare Administration and enjoys kickboxing and reading in her free time. She is very much looking forward to bringing her expertise to help and support the growth of the Boston Hemophilia Center at BWH.



## Welcome Megan!

**Megan Candito** joined the Boston Hemophilia Center team in November 2022 as a Communications Specialist. Previously, she was a Marketing & Administrative Coordinator at Massachusetts General Hospital. She received her MA from Emerson College in Marketing & Data Analytics, and her BA from the University of Massachusetts, Amherst in Communication. Megan is from Worcester, MA. In her free time, she enjoys hiking, photography, and reading. She is excited to be a part of such a unique team and is looking forward to contributing to the BHC's growth.

## Community Resources

Below are general links to resources which may be available to you. We highly recommend you speak with the care team at your local hemophilia treatment center for information more specific to your location and circumstances.

### Mental Health

<https://www.hemophilia.org/educational-programs/education/mental-health>

<https://hemaware.org/mind-body/putting-spotlight-mental-health-and-bleeding-disorders>

### Food Assistance

<https://www.boston.gov/departments/food-justice/find-your-food-pantry>

### Heating Assistance

<https://www.mass.gov/how-to/apply-for-home-heating-and-energy-assistance>

### Rental Assistance

<https://www.mass.gov/rental-assistance-programs>

### Public Housing

<https://www.mass.gov/public-housing-assistance-programs>

### Emergency Housing

<https://www.mass.gov/emergency-housing-assistance-programs>