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www.reachmd.com
info@reachmd.com
(866) 423-7849

Alleviating the Burden of Bleeding Disorders on Patients & Families

Dr. Caudle:
Welcome to *Clinician's Roundtable* on ReachMD. I'm your host, Dr. Jennifer Caudle. And joining me to discuss the work that's being done to help alleviate the impact of bleeding disorders on our patients and their families is Dr. Leonard Valentino, who's the president and chief executive officer at the National Hemophilia Foundation. Dr. Valentino, welcome to the program.

Dr. Valentino:
Thank you for having me. It's a pleasure to be here today.

Dr. Caudle:
Well, we're happy that you're here. So, to start us off, Dr. Valentino, can you tell us a little bit about inheritable blood and bleeding disorders?

Dr. Valentino:
Sure. So inheritable blood disorders are a group of non-cancerous conditions. Inheritable means that it's passed in a family from one generation to the next. And those blood disorders can be variable. Things like sickle cell disease, which affects about 100,000 people in the United States, to bleeding disorders, like hemophilia or von Willebrand disease, or some rare and ultra-rare bleeding disorders. Hemophilia affects about 1 in 5,000 live male births, but von Willebrand disease is far more common. About 1 in 1,000 people have von Willebrand disease, and about 95% of those people don't know that they have an inheritable bleeding disorder.

Dr. Caudle:
So then once a patient is diagnosed with a bleeding disorder, what are some of the chronic health challenges they may face?

Dr. Valentino:
Well, those health challenges depend, of course, on what kind of blood disorder they have. For people living with sickle cell disease, chronic pain is one of the challenges that they have because the sickled cells occlude blood vessels, and this can result in acute and chronic pain. But more importantly, the sickled cells can also obstruct the long blood vessels, resulting in pneumonia or infarction or death of lung tissue. More importantly, it can result in strokes and cognitive deficiencies or even death.

Where bleeding disorders, the constellation of symptoms are a little bit different. And as the name implies, people bleed. And this can be bleeding into joints--the knees, the ankles, and the elbows--or it could be bleeding in more significant places, such as in the brain. And again, that can result in cognitive deficiencies as well as death. We also have rare bleeding disorders that frequently result in bleeding into vital organs and structures, including the brain where von Willebrand disease is a more mild disorder. People present with easy bruising, nose bleeding, or gum bleeding that's prolonged and protracted and frequent. But they also can present with, for example, in young girls and women, heavy menstrual bleeding that oftentimes is not easily controlled and can result in unnecessary procedures, like a hysterectomy in a very young woman because of uncontrolled bleeding. So, the bleeding disorders a result in a significant morbidity if they're not addressed by diagnosis and appropriate treatment.

Dr. Caudle:
And how about the patient's family members and loved ones? How does this diagnosis impact them?

Dr. Valentino:
Well, both of these diseases that we've spoken about, sickle cell disease as well as hemophilia and other bleeding disorders, are in families. This is a family affair. And at the National Hemophilia Foundation, we have programs that address not only education for people who live with the bleeding disorder or blood disorder, but also for their family members. Because as a carer or a family member

of an individual, oftentimes these are people also who have the same symptoms or disease as the loved one that they're caring for. So, these are conditions that are passed in families and can really result in an altered family dynamic. So, we try to address the entire family in all of our work, whether that's education, advocacy work, or in research.

Dr. Caudle:
Thank you very much for that. And for those of you who are just tuning in, you're listening to *Clinician's Roundtable* on ReachMD. I'm your host Dr. Jennifer Caudle, and today I'm speaking with Dr. Leonard Valentino about the impact of bleeding disorders on our patients.

So, Dr. Valentino, now that we have a better understanding of some of the challenges our patients and their families face, I'd like to turn our attention now to the research that's being done to help alleviate this burden. Can you share some of the treatments and developments that have been uncovered so far?

Dr. Valentino:
That's a great question because this is an unprecedented time for innovation and discovery, especially in the area of hemophilia and in sickle cell disease.

The first challenge, of course, is identifying individuals. And that comes with awareness. So, there's a lot of research that's being done to help understand how people come to recognition of their healthcare problems. And what can be done to continue to diagnose more people. As I said, von Willebrand disease affects probably more than 3.5 million people in the United States, but many of those are undiagnosed.

There have been some really exciting innovations in terms of new products to treat bleeding disorders, as well as a revolution in sickle cell disease, with a number of new mechanisms of action that have been developed in the last decade to address the serious complications of sickle cell disease.

And now we're on the cusp of having potentially curative treatments for both sickle cell disease and for hemophilia in the form of gene therapies. These one-time administered treatments could really revolutionize the treatment of these otherwise chronic diseases that people live with for their entire lifetime. So, we're now facing the opportunity to be able to treat people with a one-time therapy that really can be life changing. So, it's been really exciting to watch these developments occur over the last decade and more. It's been an exciting time to be a hematologist in this field.

Dr. Caudle:
I can understand. And if we look into the future, where do you think those research efforts are headed?

Dr. Valentino:
Well, it's another great question because we think that research really needs to be focused on the needs of the patient, the needs of the person who lives with these disorders. So, there's a whole group of research that's being focused on what are the appropriate outcomes to be tracking and looking at. There's a lot of mental health issues in all of these inheritable blood disorders. And we need to have research into the best way to address and deal with mental health issues.

But we also need to be thinking about the medical issues as well. And its research into new curative treatments. So, for example, we're again beginning on a new frontier of gene editing, and the opportunity to really remove these diseases from the individual through these genetic treatments. And I also think that there's a lot of research that's going on in terms of identifying new diagnostic procedures and tests that can be used with those awareness programs that I spoke to previously. Coupling awareness and new research in terms of genetic diagnosis is going to be ever more powerful moving into the future. And will help more and more families that have inheritable blood disorders.

Dr. Caudle:
And lastly, Dr. Valentino, if any of our listeners would like to learn more, what resources are available to them and their patients?

Dr. Valentino:
Well, of course, our website is available. The National Hemophilia Foundation has a website www.hemophilia.org. And of course we're on social media. And we have a lot of information that's distributed through these different channels. One of the most important ways that we deliver resources in terms of education is through an annual conference, our Bleeding Disorders Conference, that we have each year. Those are important routes to deliver education, provide advocacy.

Dr. Caudle:
Well, those all sound like great resources for those of us caring for patients with bleeding disorders. And as that brings us to the end of today's program, I'd like to thank you, Dr. Valentino, not only for joining me to share your insights, but for all the work you're doing to

spread awareness on such an important topic. It was great having you on the program.

Dr. Valentino:
Thank you for having me.

Dr. Caudle:
I'm Dr. Jennifer Caudle, and to access this episode and others in our series, please visit ReachMD.com/CliniciansRoundtable where you can Be Part of the Knowledge. Thanks for listening.