Q&A: Addressing unmet needs for people living with PNH, a rare and chronic blood disorder

Hear from Director of Patient Services at AAMDSIF on how she supports patients with PNH, a blood disorder that significantly impacts quality of life.

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“Am I going to live?”

“What is my life going to be like, now?”

“What are my options?”

Leigh Clark explains that any one of these questions can be racing through a patient’s mind after they learn they have paroxysmal nocturnal hemoglobinuria (PNH)— a rare, chronic, and serious blood disorder.[1] In Leigh’s role as Director of Patient Services at the Aplastic Anemia and MDS International Foundation (AAMDSIF), it is precisely her responsibility to address concerns like these and provide ongoing support to people who live with this disease.

“It’s challenging for some patients to deal with the disease every day,” Clark says. “Patients may have to structure their lives around managing this new lifelong condition that they’ve likely never heard of. Our objective is to educate patients about their diagnosis and help them manage life after diagnosis.”

On top of being a continual task, managing PNH can be lonely for the patients who face this rare disease—Clark explains that some patients may never meet another person who lives with it. The rarity and lack of awareness of PNH and its symptoms can make it an extremely isolating experience for people living with the condition and their caregivers and families. This reality creates a significant burden on overall quality of life, health care systems, and society.

To understand the experience of those affected by PNH, it's helpful to know how the disease is caused and its impact.

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>
> **Leigh Clark**, Director of Patient Services at AAMDSIF

What is PNH and how is it caused?

It is estimated that 10-20 people per million worldwide live with PNH[1] and, while the disease can develop at any age, it is often diagnosed in people who are between 30-40 years old.[2,3]

PNH is caused by an acquired genetic mutation that produces red blood cells (RBCs) that are more likely to be destroyed by the immune system.[1] This process lowers the levels of circulating RBCs (anemia), causes blood clots to form (thrombosis), and causes fatigue and other hard-hitting symptoms that can impact quality of life.[1,4]

PNH is just one of several diseases that involve part of the body's immune system – the complement system – functioning incorrectly. These diseases are collectively known as complement-mediated diseases (CMDs).[5]

Though PNH is rare, there are organizations with a mission to raise awareness and help patients who face the condition along their journey.

The Aplastic Anemia and MDS International Foundation (AAMDSIF)

The Aplastic Anemia and MDS International Foundation (AAMDSIF) is a nonprofit health organization dedicated to supporting patients and families living with PNH and related diseases.[6] With the help of people like Leigh Clark, the foundation provides answers, support, and hope to thousands of patients and their networks around the world. Groups like AAMDSIF are an invaluable resource to both patients and their support systems as they navigate these physically and mentally challenging diseases.

In the following conversation, Clark shares her experience in supporting people with PNH, the challenges that patients are currently facing, and some of the work needed to provide them with better care.

How did PNH become a focus area for the Aplastic Anemia and MDS International Foundation (AAMDSIF)?

Originally, there was a PNH Foundation that was founded to help address the unmet needs of people living with PNH. Over time, the foundation became increasingly connected to AAMDSIF, as aplastic anemia is intrinsically linked to PNH. This means that people with one of these conditions are always at risk for developing the other condition. Due to this and because the needs were continuing to grow for patients living with PNH, it made sense to work together under one larger, umbrella organization.

What is your focus as Director of Patient Services?

A significant part of my role involves educating patients and helping them make sense of their disease, in a way that is accessible and digestible, at whatever stage they are in their journey. Although we support patients

at all stages of their journey, those who contact the foundation usually are newly diagnosed. Therefore, the foundation provides support in discussing their diagnosis and any complications associated with it. This involves providing as much disease information as possible — what it looks like, what patients may experience in the future, and their options. With all that is happening in the field of PNH, education about PNH should be lifelong and patients should stay up to date on the latest developments.

What challenges do people living with PNH face?

To me, the delay in diagnosis is one of the greatest challenges for people living with PNH. From my experience, a diagnosis can take months or even years. As a result of this delay, patients do not get the care they need, which can have significant consequences on their quality of life. I am hopeful that increased education and awareness of PNH will help lead to patients being diagnosed faster.

Additionally, the current treatments for patients living with PNH can come with challenges such as the burden of infusions. Some patients who need to travel to a health care setting to receive treatment may have difficulty due to physical or financial burdens, or they may feel restrained from activities others take for granted such as going on vacation or attending special occasions. While staying on treatment is critically important for success, we hope developments that can make the experience easier for patients can minimize the impact of being on lifelong treatment.

Currently, there is also a remaining unmet need. It involves symptom control, which can be both physically and mentally exhausting for people living with PNH. Even on treatment, some patients may continue to suffer and may find themselves having to compromise their quality of life to manage symptoms such as fatigue, shortness of breath, brain fog, and more.

There is another component in dealing with mental health. When you're diagnosed with something that's extremely rare, and that you've likely never heard of, you may never meet another patient dealing with it. This can be very isolating.

What are some of the ways that the Aplastic Anemia and MDS International Foundation (AAMDSIF) helps address these challenges for patients?

We feel very fortunate we're able to provide virtual support groups and offer hybrid conferences so patients can still participate even if they're not feeling great that day. This offers the opportunity to get helpful information, connect with others, and talk to leading PNH experts in the field. Additionally, we provide educational materials such as the PNH Patient Toolkit, to help patients better understand their diagnosis, and provide financial assistance to help patients with PNH visit an expert.

The Global PNH Patient Registry, is another tool we have for patients to report information about their condition and build more knowledge to help address the challenges of this rare disease.

Another part of what we do at the foundation is support patients in advocating for themselves when attending medical appointments. We inform them about who they should speak with, and make sure they are aware of their treatment options. It really does take a village.

What gives you hope?

Efforts to address the unmet needs for patients have come a long way, but there is always room for improvement. Patients with PNH need to know this is a lifelong journey – so we need to support them through every stage. I continue to be amazed by the resilience of our small but mighty patient community and how they manage the constant challenges they face.

The need for research is ongoing. Someday, I hope there will be no need to say PNH is a lifelong disease.

Novartis Commitment

Addressing the unmet needs for people living with diseases such as PNH is a critical focus for Novartis as we advance our commitment to transform and improve patients' lives. Together, with organizations like AAMDSIF, we believe we can do just that.

Understanding PNH

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