

# Paroxysmal Nocturnal Hemoglobinuria (PNH)

## Knowing the signs and symptoms of Paroxysmal Nocturnal Hemoglobinuria (PNH)

It might be easy to dismiss fatigue, headaches, bruising, and shortness of breath as something not necessarily serious, especially if they come and go without warning. However, abdominal pain and dark or blood-colored urine may be harder to ignore, and a sign that something could be wrong.<sup>1-5</sup>

All of these symptoms, mild or severe, could be a sign of a rare, chronic, and serious blood disorder called paroxysmal nocturnal hemoglobinuria (PNH), one of a number of diseases that involve the complement system – the part of the body’s immune system that protects from infection and clearing damaged cells.<sup>1-6</sup> In PNH, red blood cells (RBCs) are destroyed prematurely by the complement system, leading to a range of potentially debilitating symptoms.

The name paroxysmal nocturnal hemoglobinuria indicates just one of the symptoms of the disease – discolored urine due to hemoglobin (hemoglobinuria) that occurs suddenly (paroxysmal) overnight (nocturnal) and is often evident in the morning. It is important to be aware of all the signs and symptoms associated with PNH and how it is caused, as a first step in identifying a blood disorder that can potentially have a significant impact on a patient’s health and overall quality of life.

## What causes PNH?

Typically, hematopoietic stem cells, which are located in the bone marrow, grow and develop into RBCs, white blood cells (WBCs), and platelets. In PNH, however, an acquired mutation can arise in the body’s hematopoietic stem cells, which makes the RBCs more susceptible to destruction by the complement immune system, in a process called hemolysis.<sup>1,6</sup>

Hemolysis can be intravascular, occurring within the blood vessels, or extravascular, occurring outside the blood vessels, within the spleen and liver.<sup>1</sup> PNH is characterized by hemolysis, thrombosis (formation of blood clots), and impaired bone marrow function.<sup>3</sup> If left untreated, PNH can be life-threatening.

## PNH and its impact

Because the main job of RBCs is to carry oxygen to organs and tissues throughout the body, when they are destroyed prematurely as in PNH, people living with the disease can experience a range of symptoms. These can include hemoglobinuria (episodes of hemoglobin, the protein RBCs use to carry oxygen, being excreted in the urine), as well as low hemoglobin levels (insufficient RBC levels), which itself can lead to anemia, fatigue and other debilitating symptoms.<sup>5</sup> As mentioned, severe PNH can be fatal, with blood clots that cause heart attacks or strokes, which are the leading cause of death for people with PNH.

Anemia can be one of the most challenging parts of living with PNH. As a result of anemia, people can experience a range of hard-hitting symptoms, including weakness, fatigue, shortness of breath, dizziness,

headaches, and chest pain.<sup>6</sup> These symptoms may impair patients' ability to perform everyday activities and diminish their quality of life.

While anemia may be diagnosed through blood tests measuring hemoglobin levels, additional tests are needed to diagnose PNH with certainty. These may include a range of specialized blood tests to identify PNH cells (RBCs that are missing a critical protein), as well as other telltale signs of the disease.<sup>1</sup>

## A rare disease with a life-long burden

Even after a diagnosis, the limited number of current treatments leaves a large proportion of people with PNH with persistent anemia, fatigue, and dependence on red blood cell transfusions.<sup>1,2,6,8,9</sup> The nature of current standards of care, such as the need for lifelong infusions, adds to the already existing burden that PNH patients face daily.<sup>5,6,8,9</sup>

## Novartis commitment

At Novartis, we believe that everyone has the right to live a life free from the impact of PNH and its symptoms. We are committed to developing innovative medicines with the potential to transform the lives of people living with CMDs, including PNH.

For more information on serious blood disorders, such as PNH, and our ongoing work in hematology, click [here](#). To hear more about the unmet needs of people living with PNH and the challenges they face, click [here](#)

### References

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