

LIVING WITH PAROXYSMAL NOCTURNAL HEMOGLOBINURIA

About 6 per 1 million people are diagnosed with PNH each year.

Paroxysmal nocturnal hemoglobinuria (PNH) is a rare blood disorder that damages the red blood cells and platelets. This disease usually presents in early adulthood and continues throughout the patient's life. People with PNH have sudden, recurring episodes of symptoms. During these episodes, the affected individual may pass dark colored urine due to the premature breakdown of red blood cells. If left untreated, PNH can lead to hemolytic anemia, chronic kidney disease, or thrombosis.

01 | Causes

PNH is caused by a mutation to the PIGA gene in a blood-forming stem cell. This mutation triggers a series of events that result in abnormal red blood cells and platelets. These abnormal red blood cells break apart prematurely, and hemoglobin is released, which causes many of the symptoms associated with PNH.

02 | Symptoms

The symptoms of PNH can vary greatly from one person to another. Some possible symptoms include:

- Hemoglobinuria
- Anemia
- Fatigue
- Rapid heartbeat
- Headaches
- Blood clots
- Shortness of breath or difficulty breathing with exercise

03 | Treatment

New treatments for PNH are currently being discovered. Historically, treatment was mostly supportive, focused on managing symptoms. Individuals with this disorder would receive regular blood transfusions, iron therapy, and steroids. At that time, the average lifespan was 10 to 22 years after diagnosis. In recent years, researchers have advanced drug development for this disorder, focusing on treating the root cause and preventing red blood cell damage.

For more information on paroxysmal nocturnal hemoglobinuria, please visit clevelandclinic.org.

Did You Know?

Experts estimate between 400 and 500 cases of PNH are diagnosed in the U.S. each year.

References

<https://my.clevelandclinic.org/health/diseases/22871-paroxysmal-nocturnal-hemoglobinuria>

<https://www.ncbi.nlm.nih.gov/books/NBK562292>

<https://emedicine.medscape.com/article/207468-overview#a6>

<https://www.aamds.org/pnh>

<https://medlineplus.gov/genetics/condition/paroxysmal-nocturnal-hemoglobinuria/>

[https://www.mayoclinicproceedings.org/article/S0025-6196\(25\)00419-7/fulltext](https://www.mayoclinicproceedings.org/article/S0025-6196(25)00419-7/fulltext)

<https://www.chop.edu/conditions-diseases/paroxysmal-nocturnal-hemoglobinuria#treatment>