

Facts about Paroxysmal Nocturnal Hemoglobinuria (PNH)

Paroxysmal Nocturnal Hemoglobinuria (PNH) is a rare blood disorder that affects both males and females equally. In this condition, red blood cells break down prematurely due to the absence of specific proteins that normally shield them from the body's immune system. This process, known as *hemolysis*, can lead to many of the symptoms that are associated with PNH, like potentially life-threatening blood clots.

- PNH is a very rare disease, affecting 0.5 to 1.5 cases per million in the general population
- While PNH can affect people of any age, the typical age of diagnosis is around 30

Being diagnosed with a complex disease like PNH can affect you and your loved ones. Access to specialized care and effective treatments can significantly improve your health and support from family, friends, and dedicated groups can help you navigate the ups and downs.

Some Signs & Symptoms of PNH

- Fatigue
- Blood clots
- Stroke or heart attack
- Shortness of breath
- Abdominal pain
- Rapid heartbeat
- Dark urine
- Difficulty swallowing
- Erectile dysfunction
- Pale skin
- Cognitive difficulties

What Causes PNH?

- PNH is caused by a mutation in the PIG-A gene in our bodies which is crucial for producing protective proteins for blood cells
- Mutated stem cells in the body replicate, resulting in more bone marrow stem cells with the PIG-A mutation
- These mutated stem cells, develop into blood cells referred to as PNH blood cells or PNH clones

Why does PNH develop?

- It's not entirely clear why some people develop PNH while others do not
- PNH is not inherited or contagious
- PNH can occur in isolation or be associated with other blood issues such as aplastic anemia or myelodysplastic syndrome

How is PNH treated?

Your medical team will work with you to establish a treatment plan. Treatment for PNH can include:

Supportive Therapy

- **Anticoagulation therapy** to prevent or treat blood clots by thinning the blood
- **Blood transfusions** to boost blood counts
- **Iron chelation therapies** to reduce excess iron from blood transfusions
- **Folic acid (folate)** to increase folate levels

Drug Therapy

- **Complement inhibitors** to decrease hemolysis, prevent blood clots, and improve anemia and survival by preventing the immune system from destroying red blood cells prematurely

Targeted Treatment

- **Bone marrow transplantation** to replace the patient's bone marrow stem cells with healthy stem cells from a donor, which is considered a potential cure for certain patients with PNH

Living with PNH

Learning about and actively managing your disease, seeking support, and making necessary care adjustments can help you maintain a fulfilling life with PNH.

Knowledge:

- Engage with healthcare professionals and PNH patient organizations to **understand the disease**, and **educate your social circle** to create a supportive environment

Self-Care:

- Prioritize a **balanced diet** rich in iron and vitamins, **stay hydrated** to reduce blood clots, and engage in **mindfulness practices** like yoga to enhance your overall wellness

Social Support:

- **Seek support** from counselling, family and friends, or support groups to cope with the emotional and psychological impacts of PNH

Healthy Lifestyle:

- **Modify your daily routines** to manage PNH symptoms, incorporate **appropriate exercise** after consulting your physician, and consult your healthcare provider about **vaccinations and preventive care**

Regular Monitoring and Care:

- **Adhere to prescribed treatments**, **manage side effects** in consultation with your healthcare team, **monitor for signs** of hemolysis and blood clots, and keep a **detailed record** of treatments and symptoms for medical visits

For more detailed **information about PNH**, check out the [AAMAC Educational Series: Paroxysmal Nocturnal Hemoglobinuria](#) resource.

To learn about support groups and peer-to-peer support programs in Canada for people living with PNH, please **contact** the **Aplastic Anemia & Myelodysplasia Association of Canada (AAMAC)** at info@aamac.ca or 1-888-840-0039, or visit www.aamac.ca.

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