The Max Foundation

Accelerating health equity,
one patient at a time

Paroxysmal Nocturnal Hemoglobinuria (PNH)

This brochure will help you:

- Understand PNH and how it affects the body, including the causes and symptoms
- Learn about diagnosis and treatment for PNH
- Learn about supportive care and wellbeing

What is Paroxysmal Nocturnal Hemoglobinuria (PNH)?

PNH is a rare, blood disorder characterized by the destruction of red blood cells. It occurs due to a mutation in the PIGA gene, which is found in bone marrow. This mutation leads to a lack of protective proteins on red blood cells. Without this protection, the red blood cells become vulnerable to attack by the body's own immune system. The destruction of red blood cells may increase over time.

How does PNH affect the body?

As more red blood cells are affected, people with PNH generally start to notice more symptoms.

Some of the most common symptoms from PNH include:

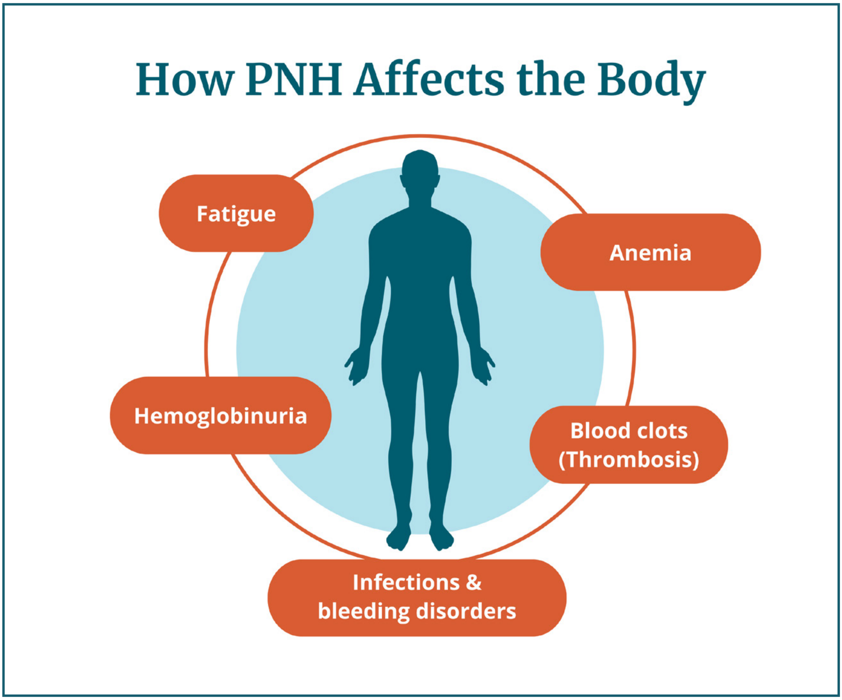
- **Fatigue** is one of the primary symptoms of PNH. Fatigue from PNH exceeds the usual levels one might feel after physical or mental exertion and is not improved by rest or sleep.

- **Anemia** means lower than normal levels of hemoglobin, the protein in your red blood cells that carries oxygen throughout your body. Anemia caused by PNH can result in symptoms like fatigue, dizziness, weakness, and pale skin.

- **Hemoglobinuria** is a condition where more hemoglobin is released through the kidneys, causing darker colored urine. Urine can appear reddish or reddish-brown and can be more noticeable in the morning.

- **Blood clots (Thrombosis)** may occur in veins or arteries, potentially leading to serious conditions like heart attacks or liver failure.

- **Infections and bleeding disorders** can occur as reduced bone marrow function causes lower white blood cell and platelet counts.



How PNH Affects the Body

Diagnosis

PNH is diagnosed through physical examination and specialized blood tests that detect PNH cells and assess the severity of the disease. Blood tests that your physician will order include:

- **Flow Cytometry** - This test detects the absence of protective proteins on red and white blood cells. It helps determine the percentage of PNH cells in the blood.

- **Complete Blood Count (CBC)** - Checks for low hemoglobin levels (anemia) and lower than normal numbers of white blood cells and platelets.

- **Lactate Dehydrogenase (LDH) Test** - Measures the levels of the LDH enzyme, which are higher than normal in PNH due to red blood cell destruction.

Treatment

Iptacopan (Fabhalta®) is a type of drug called a Factor B inhibitor. This drug treats PNH by reducing red blood cell destruction and the risk of blood clots. It is a gel capsule taken by mouth twice daily.

Depending on the severity of the disease and the symptoms a patient is experiencing, supportive treatment such as folic acid & iron supplements, blood transfusions, and antibiotics might be given.

Guidelines for Taking Iptacopan (Fabhalta®)

- Take iptacopan exactly as your healthcare provider tells you. Do not change the dose or stop taking iptacopan unless your healthcare provider tells you.

- Take one iptacopan capsule twice each day, at the time of day indicated by your physician, with or without food.

- Swallow the capsules whole. Do not open, break, or chew capsules.

- If you miss a dose or doses of iptacopan, take it as soon as you remember, even if it is almost time to take your next scheduled dose, and then take your next dose of iptacopan at your regularly scheduled time.

The importance of vaccinations:

Iptacopan affects part of your immune system and may increase your chance of getting serious infections such as pneumonia, meningitis, and influenza type b.

To reduce the risk of infections, patients must be vaccinated against these bacteria before starting treatment and receive a booster vaccination every five years. If you have been vaccinated against these bacteria in the past, you might need additional vaccinations before starting iptacopan. Your health care provider will decide if you need additional vaccinations.

The importance of taking iptacopan every day:

Do not stop taking iptacopan without talking with your health care provider, even if your symptoms have improved. Stopping treatment can lead to elevated destruction of red blood cells, worsening symptoms of anemia and hemoglobinuria, and life-threatening blood clots.

Supportive Care and Wellbeing

Living with PNH can impact overall wellbeing, but it is possible to lead a fulfilling life with proper management and support.

• Regular, light exercise can be beneficial for sleep and overall quality of life in people diagnosed with PNH. However, intense exercise or physical activity can make the condition worse. It’s helpful to keep a diary to document how you’re feeling, including your symptoms and daily activities to discuss with your healthcare team.

• A chronic disease such as PNH can become a psychological burden, causing anxiety and depression. The uncertainty of living with the disease and managing its symptoms understandably contribute to stress. Talk to your doctor if you are experiencing stress so that you can discuss options for support.

• It can also be helpful to communicate with others through participating in support groups and patient organizations. The PNH Global Alliance is a non-profit umbrella organisation of global formal and informal PNH patient groups. Our vision is that all PNH patients globally have access to optimal care and treatment and our mission to achieve this is by collectively advocating on behalf of all PNH patients.

The contact details are:
website : www.pnhglobalalliance.org
email: contact@pnhga.org

Frequently Asked Questions

1. Is PNH a genetic disorder?

PNH is not a genetic disorder. It is an acquired disorder caused by a mutation in the PIGA gene, which occurs in the cells in the bone marrow. This means family members do not inherit PNH, it cannot be passed on to future generations, and it is not contagious.

2. I am pregnant, what are the risks for me and my baby?

If you suspect you are pregnant or you want to have children, it is important that you discuss this with your physician for more information.

3. Is PNH a Chronic Disease?

Yes, it is a chronic disease, so for most patients, it is something that will need to be managed for the rest of their lives. While symptoms and severity vary, most patients require regular monitoring and ongoing treatment to manage the disease and prevent complications. However, with treatment and support, people with PNH can still live full and productive lives.

4. Is there a cure for PNH?

The only potential cure for PNH is a bone marrow transplant but this is rarely performed because it carries significant risks.

5. At what age does PNH typically occur, and does it affect men and women differently?

PNH can occur at any age, but it is most commonly diagnosed in young adults between 25 and 40 years old. It affects both men and women equally.

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