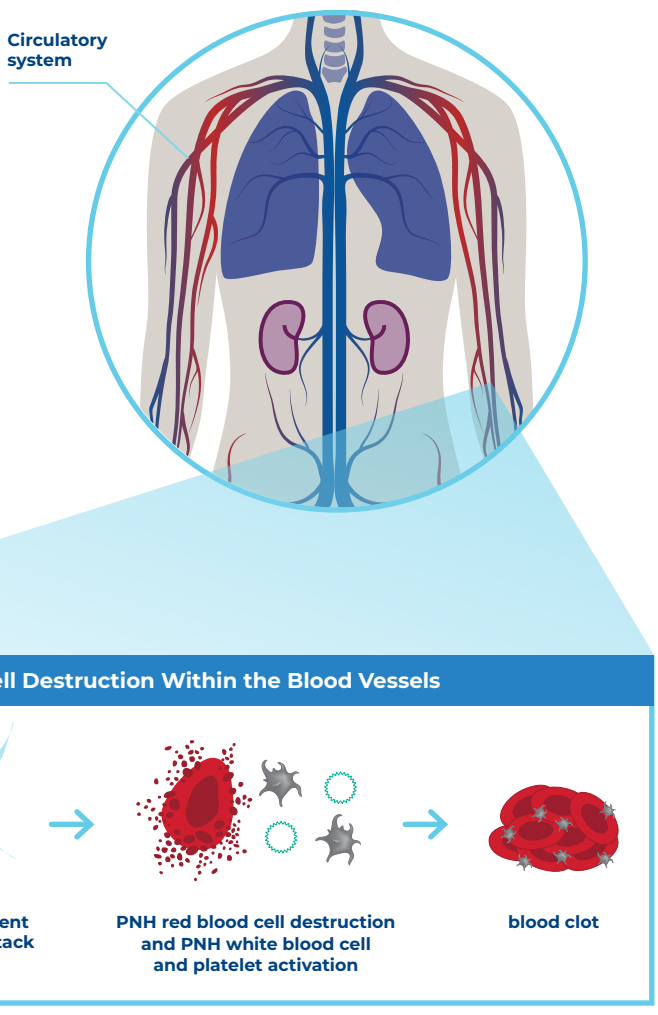
Paroxysmal Nocturnal Haemoglobinuria (PNH)

WHAT IS PNH?

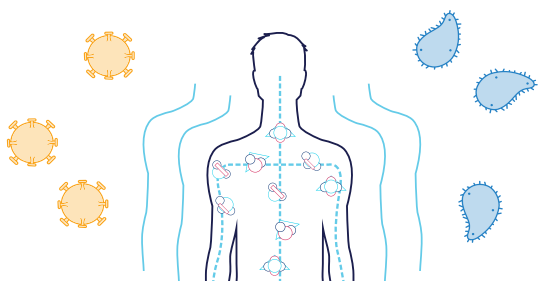
Paroxysmal nocturnal haemoglobinuria (PNH) is a **rare, chronic, progressive, and potentially life-threatening blood disorder.**

PNH is characterised by **red blood cell (RBC) destruction** within blood vessels (also known as **intravascular haemolysis**, or IVH) and **white blood cell and platelet activation**, which may lead to blood clots (thrombosis).

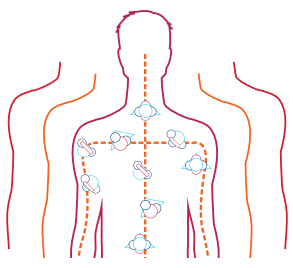
PNH is caused by an **acquired genetic mutation** (not inherited) that may happen any time after birth and results in the **production of abnormal blood cells** that are **missing important protective blood cell surface proteins**. These missing proteins enable the complement system to 'attack' and **destroy or activate these abnormal blood cells**.[1,2]



THE COMPLEMENT SYSTEM



The complement system is a part of the immune system and is **essential to the body's defence against infection**.[3]



When the system is **thrown out of balance**, or dysregulated, these proteins can **trigger a dangerous, uncontrolled cascade of reactions** that attack cells and tissues resulting in **harmful inflammation** and the **destruction of healthy cells**.[3]

Diagnosed prevalence in adults is

~14K[4] ~6K[4] ~6K[4] ~2K[4]

PNH can occur in **children and adults** at any age; the average age of diagnosis is **in the 30s**.[5]

PNH affects both **men and women** and people of **every racial and ethnic group**.[5]

Living with PNH can be debilitating, and signs and symptoms may include[5]

- **Blood clots (thrombosis)**
- **Abdominal pain**
- **Difficulty swallowing**
- **Erectile dysfunction**
- **Shortness of breath**
- **Excessive fatigue**
- **Anaemia**
- **Dark-coloured urine (haemoglobinuria)**

PNH can lead to **thrombosis**, which can occur in blood vessels throughout the body, and/or damage to other vital organs, such as **kidneys and lungs**. This can result in an overall **impaired quality of life** and **potentially premature death**.[1,5]

HOW IS PNH DIAGNOSED AND MONITORED?

Diverse symptoms and varied clinical presentation **can delay diagnosis**, in some cases by 10 or more years.[6]

PNH can be **diagnosed from a simple blood test** (high-sensitivity flow cytometry), which can detect and count PNH blood cells.[7,8]

Another type of blood test is used to **monitor ongoing PNH disease activity**. This test measures lactate dehydrogenase (LDH), an enzyme that is released from red blood cells during IVH.[7]

Monitoring LDH regularly is a very important part of managing PNH. If high levels of LDH are present, it means that many red blood cells have been destroyed within blood vessels. This has been shown to correlate with complications, such as thrombosis and early mortality.[2]

Content created by Alexion, AstraZeneca Rare Disease

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