

UNDERSTANDING PNH

PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA

This infographic has been developed and funded by Novartis for UK healthcare professionals.

PNH is an ultra-rare, acquired, clonal disorder of haematopoietic stem cells that is characterised by **haemolysis, bone marrow failure, and thrombosis**

The estimated prevalence in Great Britain of PNH is

~10–20
per million¹

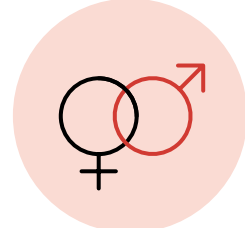
There are currently

~1,025
patients living with PNH in the UK²

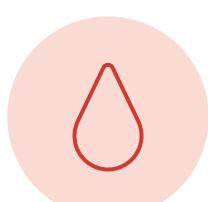
36
years

Median age at onset³

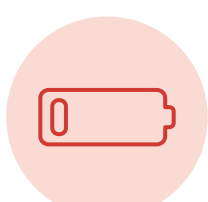
PNH affects males & females equally^{3,4}



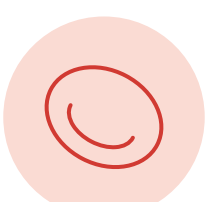
Key symptoms include:³



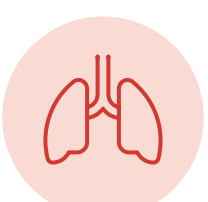
Anaemia



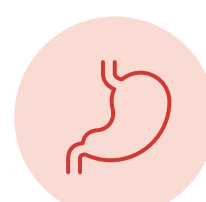
Fatigue



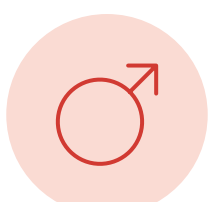
Haemoglobinuria



Dyspnoea



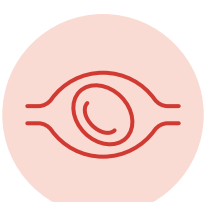
Abdominal pain



Erectile dysfunction



Dysphagia



Thrombosis

PNH varies in presentation and is often associated with other clinical conditions, such as **aplastic anaemia** and **myelodysplastic syndrome**⁴

Up to:
67%

of PNH deaths with a known cause are attributed to **thrombotic events**⁵

Diagnosis involves a thorough clinical evaluation, a detailed patient history and a variety of specialised tests, including:^{6,7}

Abdominal ultrasound

Full blood count

Bone marrow examination

Kidney function test

CT scan

Lactate dehydrogenase (LDH)

D-dimer

Reticulocyte count

Echocardiogram

Bilirubin levels



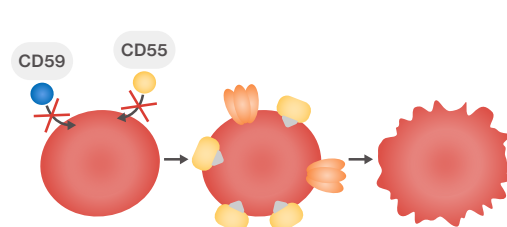
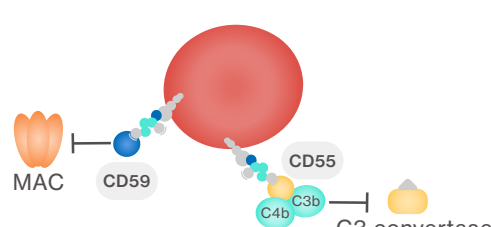
A diagnosis of PNH is confirmed by flow cytometry⁴

Mechanism of disease:

Acquired mutation in the **PIG-A** gene results in loss of **GPI** anchors on the cell surface^{1,8}

CD55 and **CD59** proteins, responsible for preventing complement-mediated cell destruction, cannot attach to cell due to loss of **GPI** anchors⁸

Lack of **CD55** and **CD59** proteins make red blood cells susceptible to complement-mediated haemolysis^{1,8}



There are a variety of different therapeutic options for PNH⁴
Approved therapies inhibit components of the complement system, halting the destruction of red blood cells and can reduce the risk of thrombosis⁴

The National PNH Service⁹

All patients who have PNH cells detected in their blood should be referred to the PNH Service to receive treatment at one of two dedicated centres.

Visit The National PNH Service for more information: <https://pnhserviceuk.co.uk/>

PNH Dedicated Centres

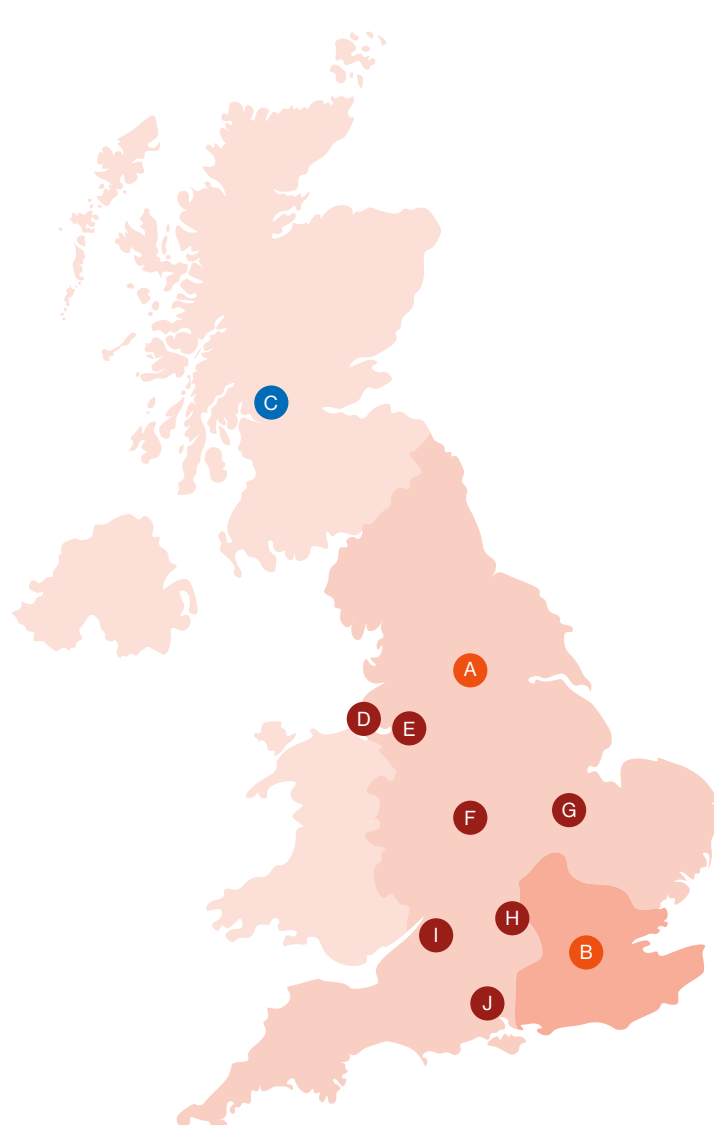
- A** Leeds Teaching Hospitals
- Leeds Teaching Hospitals catchment area
- B** King's College Hospital
- King's College Hospital catchment area

PNH Scotland Outreach

- C** Lanarkshire Monklands Hospital

PNH Outreach Clinics

- D** Royal Liverpool University Hospital
- E** Manchester Royal Infirmary
- F** Birmingham Queen Elizabeth Hospital
- G** Peterborough City Hospital
- H** Oxford Churchill Hospital
- I** Bristol Southmead Hospital
- J** Southampton Central Hospital



Abbreviations:

CT, computed tomography; GPI, glycosylphosphatidylinositol; LDH, lactate dehydrogenase; PNH, paroxysmal nocturnal haemoglobinuria.

References:

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2. The National PNH Service. Annual Report. Available at: <https://pnhserviceuk.co.uk/healthcare-professionals/annual-report/>. Accessed June 2024.
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5. Hill A, et al. *Blood*. 2013;121(25):4985-4997.
6. PNH Support (2024). Diagnosis. Available at: <https://pnhuk.org/what-is-pnh/diagnosis/>. Accessed June 2024.
7. EMBT (2015). Understanding PNH. Available at: https://www.ebmt.org/sites/default/files/migration_legacy_files/document/EBMT_Practical%20Guides%20for%20Patients_Paroxysmal%20Nocturnal%20Haemoglobinuria%20%28PNH%29_UK.pdf. Accessed June 2024.
8. Brodsky RA. *Blood*. 2014;124(18):2804-2811.
9. PNH National Service (2024). PNH Service Home. Available at: <https://pnhserviceuk.co.uk/>. Accessed June 2024.