

# Paroxysmal Nocturnal Hemoglobinuria

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The distinct and rather peculiar characteristics of paroxysmal nocturnal hemoglobinuria (PNH) have puzzled hematologists for more than a century. PNH is characterized by a decreased number of red blood cells (anemia), and the presence of blood in the urine (hemoglobinuria) and plasma (hemoglobinemia), which is evident after sleeping. PNH is associated with a high risk of major thrombotic events, most commonly thrombosis of large intra-abdominal veins. Most patients who die of their disease die of thrombosis. PNH blood cells are deficient in an enzyme known as PIG-A, which is required for the biosynthesis of cellular anchors. Proteins that are partly on the outside of cells are often attached to the cell membrane by a glycosylphosphatidylinositol (GPI) anchor, and PIG-A is required for the synthesis of a key anchor component. If PIG-A is defective, surface proteins that protect the cell from destructive components in the blood (complement) are not anchored and therefore absent, so the blood cells are broken down. The PIG-A gene is found on the X chromosome. Although not an inherited disease, PNH is a genetic disorder, known as an acquired genetic disorder. The affected blood cell clone passes the altered PIG-A to all its descendants-red cells, leukocytes (including lymphocytes), and platelets. The proportion of abnormal red blood cells in the blood determines the severity of the disease. For Diagnosis and treatment **click here** Important Links Gene sequence [[www.ncbi.nlm.nih.gov/LocusLink/list.cgi?Q=paroxysmal+nocturnal+hemoglobinuria](http://www.ncbi.nlm.nih.gov/LocusLink/list.cgi?Q=paroxysmal+nocturnal+hemoglobinuria) ORG=Hs V=0] collection of generelated information BLink [[www.ncbi.nlm.nih.gov/sutils/blink.cgi?pid=11863130](http://www.ncbi.nlm.nih.gov/sutils/blink.cgi?pid=11863130) org=1] related sequences in different organisms The literature Research articles online full text Books online books section OMIM [[www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=311770](http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=311770)]