Paroxysmal nocturnal hemoglobinuria (PNH) is a rare and acquired hematological disorder characterized by the premature destruction of red blood cells (hemolysis), increased blood clotting (thrombosis), and bone marrow failure. PNH is primarily caused by a genetic mutation in blood-forming stem cells, leading to the absence of certain proteins that normally protect red blood cells from the immune system's attack.

Here are some key points about PNH:

Genetic Mutation: PNH is associated with a mutation in the PIG-A gene, which is responsible for the synthesis of glycosylphosphatidylinositol (GPI) anchors. These anchors attach various proteins, including those that protect red blood cells from the immune system. The genetic mutation leads to a deficiency of GPI-anchored proteins on the surface of blood cells.

Hemolysis: Due to the deficiency of GPI-anchored proteins, red blood cells become susceptible to the immune system's attack. This leads to hemolysis, where red blood cells are destroyed prematurely, resulting in anemia.

Thrombosis: PNH also increases the risk of blood clots (thrombosis) in various parts of the body, including veins, arteries, and small blood vessels. This can lead to serious complications, such as strokes and pulmonary embolism.

Symptoms: Common symptoms of PNH include fatigue, weakness, pale skin, dark urine (due to hemoglobin breakdown), abdominal pain, and the formation of blood clots. Symptoms can vary in severity among individuals.

Diagnosis: Diagnosis typically involves blood tests, such as flow cytometry, which can identify the deficiency of GPI-anchored proteins on blood cells. A bone marrow biopsy may also be performed to confirm the diagnosis.

Treatment: Treatment options for PNH aim to manage the condition's symptoms and reduce complications. These may include blood transfusions, immunosuppressive therapy, and medications to reduce clotting risk. Eculizumab (Soliris) is a targeted therapy that inhibits the immune system's attack on red blood cells and is commonly used in PNH treatment.

Bone Marrow Transplant: In some cases, allogeneic hematopoietic stem cell transplantation (bone marrow transplant) can be considered as a potential cure for PNH. However, this is a complex and high-risk procedure.

PNH is a chronic condition that requires ongoing medical management. It is important for individuals with PNH to work closely with healthcare professionals, including hematologists, to monitor the condition, manage symptoms, and reduce the risk of complications. Research and advancements in targeted therapies have improved the outlook for individuals with PNH, but the disease can still be associated with significant health challenges.