Paroxysmal Nocturnal Hemoglobinuria (PNH)

Thrombo-Guide
vom Labor zur Diagnose

Description:

PNH is a rare, acquired clonal hematologic disorder. An acquired somatic mutation in the phosphatidylinositol glycan class A (PIGA) gene within hematopoietic stem cells leads to a deficiency of glycosylphosphatidylinositol (GPI)-anchored proteins on the cell surface, disrupting normal cellular function. Clinical hallmarks comprise intravascular hemolysis, a severe thrombophilic state with a propensity for both typical and atypical thromboses, and variable cytopenias that can progress to profound pancytopenia.

Diagnostics:

- Cytopenia, typically pancytopenia; laboratory markers of hemolysis.
- Definitive diagnosis is established via flow cytometry demonstrating a distinct cell population lacking GPI-anchored surface proteins.

Treatment:

- Complement cascade inhibition using eculizumab, ravulizumab, crovalimab, pegcetacoplan, danicopan, or iptacopan.
- Clinical management should be coordinated at a specialized referral center.
- For more comprehensive information, consult the current Onkopedia PNH guidelines: https://www.onkopedia.com/de/onkopedia/guidelines/paroxysmale-naechtliche-haemoglobinurie-pnh/@@guideline/html/index.html

References:

Thomas L, Labor und Diagnose, 2023, Release 5: https://www.labor-und-diagnose.de/index.html
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