

Aktuelles zur Diagnostik und Therapie der paroxysmalen nächtlichen Hämoglobinurie

- 1) Parker C, Omine M, Richards S et al. Diagnosis and management of paroxysmal nocturnal hemoglobinuria. *Blood* 2005;106:3699-709.
- 2) Hillmen P, Young NS, Schubert J et al. The complement inhibitor eculizumab in paroxysmal nocturnal hemoglobinuria. *N Engl J Med* 2006;355:1233-43.
- 3) Hillmen P, Lewis SM, Bessler M et al. Natural history of paroxysmal nocturnal hemoglobinuria. *N Engl J Med* 1995;333:1253-8.
- 4) Socie G, Mary JY, De Gramont A et al. Paroxysmal nocturnal haemoglobinuria: long-term follow-up and prognostic factors. French Society of Haematology [see comments]. *Lancet* 1996;348:573-7.
- 5) Dacie JV, Lewis SM. Paroxysmal nocturnal haemoglobinuria: clinical manifestations, haematology, and nature of the disease. *Ser Haematol* 1972;5:3-23.
- 6) Luzzatto L. Paroxysmal nocturnal hemoglobinuria: an acquired X-linked genetic disease with somatic-cell mosaicism. *Curr Opin Genet Dev* 2006;16:317-22.
- 7) Bessler M, Mason PJ, Hillmen P et al. Mutations in the PIG-A gene causing partial deficiency of GPI-linked surface proteins (PNH II) in patients with paroxysmal nocturnal haemoglobinuria. *Br J Haematol* 1994;87:863-6.
- 8) Bessler M, Mason PJ, Hillmen P et al. Paroxysmal nocturnal haemoglobinuria (PNH) is caused by somatic mutations in the PIG-A gene. *EMBO J* 1994;13:110-7.
- 9) Ware RE, Rosse WF, Howard TA. Mutations within the Piga gene in patients with paroxysmal nocturnal hemoglobinuria. *Blood* 1994;83:2418-22.
- 10) Mortazavi Y, Merk B, McIntosh J et al. The spectrum of PIG-A gene mutations in aplastic anemia/paroxysmal nocturnal hemoglobinuria (AA/PNH): a high incidence of multiple mutations and evidence of a mutational hotspot. *Blood* 2002.
- 11) Hugel B, Socie G, Vu T et al. Elevated levels of circulating procoagulant microparticles in patients with paroxysmal nocturnal hemoglobinuria and aplastic anemia. *Blood* 1999;93:3451-6.
- 12) Vu T, Griscelli Bennaceur A, Gluckman E et al. Aplastic anaemia and paroxysmal nocturnal haemoglobinuria: a study of the GPI-anchored proteins on human platelets. *Br J Haematol* 1996;93:586-9.
- 13) Rother RP, Bell L, Hillmen P et al. The clinical sequelae of intravascular hemolysis and extracellular plasma hemoglobin: a novel mechanism of human disease. *JAMA* 2005;293:1653-62.
- 14) Schrezenmeier H, Hertenstein B, Wagner B et al. A pathogenetic link between aplastic anemia and paroxysmal nocturnal hemoglobinuria is suggested by a high frequency of aplastic anemia patients with a deficiency of phosphatidylinositol glycan anchored proteins [published erratum appears in *Exp Hematol* 1995 Feb;23(2):181]. *Exp Hematol* 1995;23:81-7.
- 15) Schrezenmeier H, Hildebrand A, Rojewski M et al. Paroxysmal nocturnal haemoglobinuria: a replacement of hematopoietic tissue? *Acta Haematol* 2000;103:41-8.
- 16) Araten DJ, Nafa K, Pakdeesuwan K et al. Clonal populations of hematopoietic cells with paroxysmal nocturnal hemoglobinuria genotype and phenotype are present in normal individuals. *Proc Natl Acad Sci U S A* 1999;96:5209-14.



- 17) Nebe T, Schubert, J. et al. Flow cytometric analysis of GPI-deficient cells for the diagnosis of paroxysmal nocturnal hemoglobinuria (PNH). *J Lab Med* 2003;27:257-65.
- 18) Spath-Schwalbe E, Schrezenmeier H, Heimpel SH. [Paroxysmal nocturnal hemoglobinuria. Clinical experiences with 40 patients at one center over 25 years] Paroxysmale nachtliche Hamoglobinurie. Klinische Erfahrungen bei 40 Patienten in einem Zentrum über 25 Jahre. *Dtsch Med Wochenschr* 1995;120:1027-33.
- 19) Audebert HJ, Planck J, Eisenburg M et al. Cerebral ischemic infarction in paroxysmal nocturnal hemoglobinuria report of 2 cases and updated review of 7 previously published patients. *J Neurol* 2005;252:1379-86.
- 20) Frickhofen N, Heimpel H, Kaltwasser JP et al. Antithymocyte globulin with or without cyclosporin A: 11-year follow-up of a randomized trial comparing treatments of aplastic anemia. *Blood* 2003;101:1236-42.
- 21) Saso R, Marsh J, Cevreska L et al. Bone marrow transplants for paroxysmal nocturnal haemoglobinuria. *Br J Haematol* 1999;104:392-6.
- 22) Brecher ME, Taswell HF. Paroxysmal nocturnal hemoglobinuria and the transfusion of washed red cells. A myth revisited. *Transfusion* 1989;29:681-5.
- 23) Knobloch K, Lichtenberg A, Schubert J et al. Paroxysmal nocturnal hemoglobinuria--pre- and perioperative rationale during cardiac surgery using extracorporeal circulation. *Eur J Cardiothorac Surg* 2004;26:1058-9.
- 24) Knobloch K, Zardo P, Gohrbandt B et al. Cardiac surgery in a patient with paroxysmal nocturnal hemoglobinuria. *Haematologica* 2002;87:ECR29.
- 25) Sloand EM, Maciejewski JP, Dunn D et al. Correction of the PNH defect by GPI-anchored protein transfer. *Blood* 1998;92:4439-45.
- 26) Sloand EM, Mainwaring L, Keyvanfar K et al. Transfer of glycosylphosphatidylinositol-anchored proteins to deficient cells after erythrocyte transfusion in paroxysmal nocturnal hemoglobinuria. *Blood* 2004;104:3782-8.
- 27) Hall C, Richards S, Hillmen P. Primary prophylaxis with warfarin prevents thrombosis in paroxysmal nocturnal hemoglobinuria (PNH). *Blood* 2003;102:3587-91.
- 28) Moyo VM, Mukhina GL, Garrett ES et al. Natural history of paroxysmal nocturnal haemoglobinuria using modern diagnostic assays. *Br J Haematol* 2004;126:133-8.
- 29) Hill A, Ridley SH, Esser D et al. Protection of erythrocytes from human complement-mediated lysis by membrane-targeted recombinant soluble CD59: a new approach to PNH therapy. *Blood* 2006;107:2131-7.
- 30) Hillmen P, Hall C, Marsh JC et al. Effect of eculizumab on hemolysis and transfusion requirements in patients with paroxysmal nocturnal hemoglobinuria. *N Engl J Med* 2004;350:552-9.
- 31) Hill A, Hillmen P, Richards SJ et al. Sustained response and long-term safety of eculizumab in paroxysmal nocturnal hemoglobinuria. *Blood* 2005;106:2559-65.
- 32) Hill A, Rother RP, Hillmen P. Improvement in the symptoms of smooth muscle dystonia during eculizumab therapy in paroxysmal nocturnal hemoglobinuria. *Haematologica* 2005;90:ECR40.
- 33) Hillmen, P., Muus, P., Dührsen, U., Risitano, A., Schubert, J., Young, N. S., Schrezenmeier, H., Szer, J., Brodsky, R. A., Hill, A., Socie, G., Rollins, S. A., Rother, R. P., Bell, L., and Luzzatto, L. The terminal complement inhibitor eculizumab reduces thrombosis in patients with paroxysmal nocturnal hemoglobinuria. *Blood* 108, 41a. 2006.