References for Bone Marrow Failure

- 1. Camitta BM, Rappeport JM, Parkman R, Nathan DG. Selection of patients for bone marrow transplantation in severe aplastic anemia. *Blood.* 1975; 45(3): 355-63.
- 2. Gluckman E, Devergie A, Poros A, Degoulet P. Results of immunosuppression in 170 cases of severe aplastic anaemia. Report of the European Group of Bone Marrow Transplant (EGBMT). *BJH.* 1982; 51(4): 541-50.
- 3. Bacigalupo A, Hows J, Gluckman E, Nissen C, Marsh J, Van Lint MT, et al. Bone marrow transplantation (BMT) versus immunosuppression for the treatment of severe aplastic anaemia (SAA): a report of the EBMT SAA working party. *BJH.* 1988; 70(2): 177-82.
- 4. Frickhofen N, Heimpel H, Kaltwasser JP, Schrezenmeier H, German Aplastic Anemia Study G. Antithymocyte globulin with or without cyclosporin A: 11-year follow-up of a randomized trial comparing treatments of aplastic anemia. *Blood.* 2003; 101(4): 1236-42.
- 5. Scheinberg P, Nunez O, Weinstein B, Scheinberg P, Biancotto A, Wu CO, et al. Horse versus rabbit antithymocyte globulin in acquired aplastic anemia. *NEJM.* 2011; 365(5): 430-8.
- 6. Scheinberg P, Wu CO, Nunez O, Scheinberg P, Boss C, Sloand EM, et al. Treatment of severe aplastic anemia with a combination of horse antithymocyte globulin and cyclosporine, with or without sirolimus: a prospective randomized study. *Haematologica*. 2009; 94(3): 348-54.
- 7. Scheinberg P, Nunez O, Wu C, Young NS. Treatment of severe aplastic anaemia with combined immunosuppression: anti-thymocyte globulin, ciclosporin and mycophenolate mofetil. *BJH.* 2006; 133(6): 606-11.
- 8. Kim H, Min YJ, Baek JH, Shin SJ, Lee EH, Noh EK, et al. A pilot dose-escalating study of alemtuzumab plus cyclosporine for patients with bone marrow failure syndrome. *Leuk Res.* 2009; 33(2): 222-31.
- 9. Brodsky RA, Chen AR, Dorr D, Fuchs EJ, Huff CA, Luznik L, et al. High-dose cyclophosphamide for severe aplastic anemia: long-term follow-up. *Blood.* 2010; 115(11): 2136-41.
- 10. Marsh JC, Kulasekararaj AG. Management of the refractory aplastic anemia patient: what are the options? *Blood.* 2013; 122(22): 3561-7.
- 11. Olnes MJ, Scheinberg P, Calvo KR, Desmond R, Tang Y, Dumitriu B, et al. Eltrombopag and improved hematopoiesis in refractory aplastic anemia. *NEJM.* 2012; 367(1): 11-9.
- 12. Tiu RV, Sekeres MA. The role of AMG-531 in the treatment of thrombocytopenia in idiopathic thrombocytopenic purpura and myelodysplastic syndromes. *Expert Opin Biol Ther.* 2008; 8(7): 1021-30.
- 13. Desmond R, Townsley DM, Dumitriu B, Olnes MJ, Scheinberg P, Bevans M, et al. Eltrombopag restores trilineage hematopoiesis in refractory severe aplastic anemia that can be sustained on discontinuation of drug. *Blood.* 2014; 123(12): 1818-25.
- 14. Scheinberg P, Wu CO, Nunez O, Young NS. Predicting response to immunosuppressive therapy and survival in severe aplastic anaemia. *BJH.* 2009; 144(2): 206-16.

- 15. Young NS. Telomere biology and telomere diseases: implications for practice and research. *Hematology Am Soc Hematol Educ Program.* 2010; 2010: 30-5.
- 16. Calado RT, Cooper JN, Padilla-Nash HM, Sloand EM, Wu CO, Scheinberg P, et al. Short telomeres result in chromosomal instability in hematopoietic cells and precede malignant evolution in human aplastic anemia. *Leukemia* 2012; 26(4): 700-7.
- 17. Scheinberg P, Cooper JN, Sloand EM, Wu CO, Calado RT, Young NS. Association of telomere length of peripheral blood leukocytes with hematopoietic relapse, malignant transformation, and survival in severe aplastic anemia. *JAMA*. 2010; 304(12): 1358-64.
- 18. Doney K, Leisenring W, Storb R, Appelbaum FR. Primary treatment of acquired aplastic anemia: outcomes with bone marrow transplantation and immunosuppressive therapy. Seattle Bone Marrow Transplant Team. *Ann Intern Med.* 1997; 126(2): 107-15.
- 19. Schrezenmeier H, Passweg JR, Marsh JC, Bacigalupo A, Bredeson CN, Bullorsky E, et al. Worse outcome and more chronic GVHD with peripheral blood progenitor cells than bone marrow in HLA-matched sibling donor transplants for young patients with severe acquired aplastic anemia. *Blood.* 2007; 110(4): 1397-400.
- 20. Young NS, Bacigalupo A, Marsh JC. Aplastic anemia: pathophysiology and treatment. *Biol Blood Marrow Transplant*. 2010; 16(1 Suppl): S119-25.
- 21. Parker CJ. Paroxysmal nocturnal hemoglobinuria: an historical overview. *Hematology Am Soc Hematol Educ Program.* 2008: 93-103.
- 22. Krawitz PM, Hochsmann B, Murakami Y, Teubner B, Kruger U, Klopocki E, et al. A case of paroxysmal nocturnal hemoglobinuria caused by a germline mutation and a somatic mutation in PIGT. *Blood.* 2013; 122(7): 1312-5.
- 23. Luzzatto L. PNH from mutations of another PIG gene. *Blood* 2013; 122(7): 1099-100.
- 24. Józsi M. Anti-Complement Autoantibodies in Membranoproliferative Glomerulonephritis and Dense Deposit Disease Chapter 3 in An Update on Glomerulopathies Etiology and Pathogenesis. InTech. 2011.
- 25. Parker C, Omine M, Richards S, Nishimura J, Bessler M, Ware R, et al. Diagnosis and management of paroxysmal nocturnal hemoglobinuria. *Blood.* 2005; 106(12): 3699-709.
- 26. Risitano AM, Rotoli B. Paroxysmal nocturnal hemoglobinuria: pathophysiology, natural history and treatment options in the era of biological agents. *Biologics*. 2008; 2(2): 205-22.
- 27. Borowitz MJ, Craig FE, Digiuseppe JA, Illingworth AJ, Rosse W, Sutherland DR, et al. Guidelines for the diagnosis and monitoring of paroxysmal nocturnal hemoglobinuria and related disorders by flow cytometry. Cytometry B Clin Cytom. 2010; 78(4): 211-30.
- 28. Parker CJ. Management of paroxysmal nocturnal hemoglobinuria in the era of complement inhibitory therapy. *Hematology Am Soc Hematol Educ Program.* 2011: 21-9.
- 29. Hillmen P, Muus P, Roth A, Elebute MO, Risitano AM, Schrezenmeier H, et al. Long-term safety and efficacy of sustained eculizumab treatment in patients with paroxysmal nocturnal haemoglobinuria. *BJH.* 2013; 162(1): 62-73.

- 30. Nishimura J, Yamamoto M, Hayashi S, Ohyashiki K, Ando K, Brodsky AL, et al. Genetic variants in C5 and poor response to eculizumab. *N Engl J Med.* 2014; 370(7): 632-9.
- 31. Lamy T, Loughran TP, Jr. How I treat LGL leukemia. *Blood*. 117:2764-2774.
- 32. Koskela HL, Eldfors S, Ellonen P et al. Somatic Mutations in Large Granular Lymphocytic Leukemia. *N Engl J Med*. 2012; 366:1905-13
- 33. Yamamoto JF, Goodman MT. Patterns of leukemia incidence in the United States by subtype and demographic characteristics, 1997-2002. *Cancer Causes Control.* 2008;19:379-390.
- 34. Josefsen D, Myklebust JH, Lynch DH, Stokke T, Blomhoff HK, Smeland EB. Fas ligand promotes cell survival of immature human bone marrow CD34+CD38-hematopoietic progenitor cells by suppressing apoptosis. *Exp Hematol.* 1999;27:1451-1459.
- 35. Lundell R, Hartung L, Hill S, Perkins SL, Bahler DW. T-cell large granular lymphocyte leukemias have multiple phenotypic abnormalities involving pan-T-cell antigens and receptors for MHC molecules. *Am J Clin Pathol.* 2005;124:937-946.
- 36. Visconte V and Tiu RV. Rare Bone Marrow Failure Syndromes. Textbook of Uncommon Cancers. 4th Edition. D. Raghavan, C. Blanke, DH. Johnson, PL Moots, GH. Reaman, PG. Rose, MA. Sekeres (eds). Wiley-Blackwell UK. 2012.
- 37. Taniguchi T, D'Andrea AD. Molecular pathogenesis of Fanconi anemia: recent progress. Blood. 2006 Jun 1;107(11):4223-33.
- 38. Tischkowitz MD, Hodgson SV. Fanconi anaemia. J Med Genet. 2003 Jan;40(1):1-10.
- 39. Mozdarani H, Ashtiani KA, Mohseni-Meybodi A. Evaluation of concentration and storage effects of mitomycin C in the diagnosis of Fanconi anemia among idiopatic aplastic anemia patients. Indian J Hum Genet. 2011 Sep;17(3):145-51.
- 40. Smith AR, Wagner JE. Current clinical management of Fanconi anemia. Expert Rev Hematol. 2012 Oct;5(5):513-22.
- 41. Kee Y, D'Andrea AD. Molecular pathogenesis and clinical management of Fanconi anemia. J Clin Invest. 2012 Nov 1;122(11):3799-806.
- 42. Eiler E, Frohnmayer D, Frohnmayer L, Larsen K, Owen J (Eds). Fanconi Anemia. Guidelines for Diagnosis and management. 3rd Ed. 2008
- 43. Kirwan M, Dokal I. Dyskeratosis congenita, stem cells and telomeres. Biochim Biophys Acta. 2009 Apr;1792(4):371-9.
- 44. Ballew BJ, Savage SA. Updates on the biology and management of dyskeratosis congenita and related telomere biology disorders. Expert Rev Hematol. 2013 Jun;6(3):327-37.
- 45. Gadalla SM, Sales-Bonfim C, Carreras J, Alter BP, Antin JH, Ayas M, et al. Outcomes of allogeneic hematopoietic cell transplantation in patients with dyskeratosis congenita. *Biol Blood Marrow Transplant.* 2013; 19(8): 1238-43.
- 46. Ayas M, Nassar A, Hamidieh AA, Kharfan-Dabaja M, Othman TB, Elhaddad A, et al. Reduced intensity conditioning is effective for hematopoietic SCT in dyskeratosis congenita-related BM failure. *Bone Marrow Transplant.* 2013; 48(9): 1168-72.
- 47. Vlachos A, Muir E. How I treat Diamond-Blackfan anemia. *Blood.* 2010; 116(19): 3715-23.

- 48. Dutt S, Narla A, Lin K, Mullally A, Abayasekara N, Megerdichian C, Wilson FH, Currie T, Khanna-Gupta A, Berliner N, Kutok JL, Ebert BL. Haploinsufficiency for ribosomal protein genes causes selective activation of p53 in human erythroid progenitor cells. Blood. 2011 Mar 3;117(9):2567-76.
- 49. Chirnomas SD and Kupfer GM. The inherited Bone Marrow Failure Syndromes. Pediatr Clin N Am. 2013; 1291-1310
- 50. Khincha PP, Savage SA. Genomic characterization of the inherited bone marrow failure syndromes. *Semin Hematol.* 2013; 50(4): 333-47.
- 51. Buchanan GR, International Diamond-Blackfan Anemia Study G. Oral megadose methylprednisolone therapy for refractory Diamond-Blackfan anemia. International Diamond-Blackfan Anemia Study Group. *J Pediatr Hematol Oncol.* 2001; 23(6): 353-6.
- 52. Fagioli F, Quarello P, Zecca M, Lanino E, Corti P, Favre C, et al. Haematopoietic stem cell transplantation for Diamond Blackfan anaemia: a report from the Italian Association of Paediatric Haematology and Oncology Registry. *BJH.* 2014; 165(5): 673-81.