

DAFTAR PUSTAKA

- Adiratna, W., A. Udiyono, & L. D. Saraswati (2020). Hubungan pengetahuan dan dukungan sosial terhadap skor kepatuhan minum obat kelasi besi pada pasien thalassemia. *Jurnal Kesehatan Masyarakat*. 8(1):23–30.
- Aldakeel, S.A., Ghanem, N.Z., Al-Amodi, A.M., Osman, A.K., Al Asoom, L.I., Ahmed, N.R., Almandil, N.B., Akhtar, M.S., Azeez, S.A. dan Borgio, J.F. (2020) 'Identification of seven novel variants in the β -globin gene in transfusion-dependent and normal patients', *Archives of Medical Science*, 16(2), pp. 453–459.
- Alberts, B., Johnson, A., Lewis, J., Morgan, D., Raff, M., Roberts, K. and Walter, P. (2014) *Molecular Biology of the Cell*. 6th edn. New York: Garland Science.
- Arumdapta, K.T. (2025) *Deteksi Mutasi Cd26 Gen HBB pada Keluarga Pasien β -Thalassemia di RSUD Tidar Kota Magelang* (Skripsi, UGM).
- Cao, A. & R. Galanello. (2010). β -thalassemia. *Genet Med*. 12(2):61-76
- Clancy, S. (2008) 'RNA Splicing: Introns, Exons and Spliceosomes', *Nature Education*, 1(1), p. 31.
- Bennett, R. L., French, K. S., Resta, R. G., & Doyle, D. L. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, 17(5), 419–433.
- Burns, M., L. Foster, & M. Walker. (2020). *DNA techniques to verify food authenticity applications in food fraud*. CPI Group (UK) Ltd. UK. pp. 39–44.
- Brancaleoni, V., E. D. Pierro, I. Motta, & M. D. Cappellini. (2016). Laboratory diagnosis of thalassemia. *International Journal of Laboratory Hematology*. 38(1):32–40.
- Flint, J., et al. (1993). *The population genetics of the haemoglobinopathies*. Bailliere's Clinical Haematology, 6(1), 215-262
- Fucharoen, S. & D.J. Weatherall. (2012). *The haemoglobin e thalassemia*. Cold Spring Harb Perspect Med. 2(8): 1-15.
- Farashi, S. & C. L. Harteveld. (2018). *Molecular basis of α -thalassemia*. Blood Cell, Molecules, and Diseases. 70:43–53.
- Firani, N. K. (2018). *Mengenali Sel Darah Merah: Mengenal Anemia dan Penyakit Hemoglobin*. UB Press.
- Ford, J., (2013). Red blood cell morphology *Mengenali Sel-Sel Darah dan Kelainan Darah*. *International Journal of Laboratory Hematology*, 35 (3), pp.351- 357.
- Galanello, R., & Origa, R. (2010). Beta-thalassemia. *Orphanet Journal of Rare Diseases*, 5(11). 1-15
- Grosso, M., R. Sessa, S. Puzone S, M. Rosasria & P. Izzo. (2015). *Molecular Basis 45 of Thalassemia in Anemia* .pp.342- 348
- Ghifari, R., Harahap, A. and Setianingsih, I. (2018) Genetic Diversity and Spectrum of β -Thalassemia Mutations in the Indonesian Archipelago, *Indonesian Journal of Biotechnology*, 23(1), pp. 45–52.

- Guyton, A. C., & Hall, J. E. (2020). *Textbook of Medical Physiology* (14th ed.). Philadelphia, PA: Elsevier.
- Guyton, A. C. & J. E. Hall. (2011). *Medical Physiology*. 12th Edition. Saunders Elsevier. Philadelphia. pp 415.
- Hanafi, S., R. Hassan, R. Bahar, W. A. Abdullah, M. F. Johan, N. D. Rashid, N. F. Azman, A. Nasir, S. Hassan, R. Ahmad, A. Othman, M. I. Ibrahim, S. Sukeri, S. Sulong, S. Yusoff, N. S. Mohamad, A. Hussein, R. Hassan, N. Yusoff, B. H. Yahya, E. Ismail, N. K. N. Yusoff, S. Salleh, & B. A. Zilfalil (2014). Multiplex amplification refractory mutation system (MARMS) for the detection of β -globin gene mutations among the transfusion-dependent β thalassemia Malay patients in Kelantan, Northeast of Peninsular Malaysia. *American Journal of Blood Research*. 4(1): 33–40.
- Hassan, S., R. Ahmad, Z. Zakaria, Z. Zulkafli, & W. Z. Abdullah (2012). Detection of β -globin gene mutations among β -thalassemia carriers and patient in Malaysia: application of multiplex amplification refractory mutation system-polymerase chain reaction. *Malaysian Journal Medicine Science*. 20(1):13–20.
- Haldane, J. B. S. (1949). *The rate of mutation of human genes*. Proceedings of the Eighth International Congress of Genetics. Hereditas. pp. 267– 273.
- Hoffbrand, A. V. & P. A. H. Moss (2011). *Essential Haemology*. 6th Edition. John Wiley & Sons, Inc. UK.
- Hoffbrand, A. V., & Steensma, D. P. (2020). *Hoffbrand's Essential Haematology*. Wiley-Blackwell.
- Hoffbrand, A. V., Moss, P. A. H., & Pettit, J. E. (2016). *Hoffbrand's Essential Haematology* (7th ed.). Wiley-Blackwell.
- Kaushansky, K., M.A. Lichtman, T.P. Josef, M.L. Marcel, W.P. Oliver, J.B. Linda & A.C. Michael. (2016). *Williams Hematology*, 9th Edition. McGraw-Hill Education, New York. pp. 729-733.
- Little S. (2001). *Amplification-refractory mutation system (ARMS) analysis of point mutations*. *Curr Protoc Hum Gene*: 1-2.
- Muncie, H.L. and Campbell, J.S. (2009) 'Alpha and Beta Thalassemia', *American Family Physician*, 80(4), pp. 339–344.
- Moore, K. L., Dalley, A. F., & Agur, A. M. R. (2018). *Clinically Oriented Anatomy* (8th ed.). Lippincott Williams & Wilkins.
- Mattick, J.S. (2004). RNA regulation: a new genetics?. *Nature Reviews Genetics*, 5(4), pp. 316–323.
- Nayak, R., S. Rai, & A. Gupta. (2012). *Essentials in Hematology and Clinical Pathology*. 1st Edition. Jaypee Brothers Medical Publishers. New Delhi.

pp. 10–12; 45.

- Pedoman Nasional Pelayanan Kedokteran Tata laksana Talasemia, Pub. L. No. HK.01.07/MENKES/I/2018 (2018).
- Rongers, K. (2011). *The Human Body Blood Physiology and Circulation*. Britannica Educational Publishing. New York. pp. 17–19.
- Rujito, L. (2019). *Talasemia: Genetik Dasar dan Pengelolaan Terkini*. Universitas Jendral Soedirman Press. Purwokerto.
- Rujito, L., B. Muhammad, M. Sri & S.S.M. Abdul. (2015). Molecular Scanning of β -Thalassemia in the Southern Region of Central Java, Indonesia; a Step Towards a Local Prevention Program. *Hemoglobin*.39(5). pp 330-333.
- Ratih, N., Watkins, R. J., Syafira, N. and Setianingsih, I. (2017) 'Molecular characterization of beta-thalassemia in North Sumatra, Indonesia', *Journal of Human Genetics*, 62(10), pp. 891-896.
- Setianingsih, I., Williamson, R., Marzuki, S., Harahap, A. and Forrest, S.M. (1998) Molecular basis of β -thalassaemia in Indonesia: whole- genome analysis of the β -globin gene cluster, *Journal of Medical Genetics*, 35(11), pp. 929–936.
- Setianingsih, I., Williamson, R., Marzuki, S., Harahap, A. and Tamam, M. (2003) Molecular basis of beta-thalassemia in Indonesia: Application to prenatal diagnosis. *Molecular Biology Reports*, 30(2), pp. 71-81
- Sherwood, L. (2010). *Human Physiology: From Cells to Systems*, 7th edition. Books/Cole Cengage Learning, Belmont. pp. 394-395.
- Standring, S. (2020). *Gray's Anatomy: The Anatomical Basis of Clinical Practice* (42nd ed.). Elsevier
- Sari, D. P., Wahidiyat, P. A., Setianingsih, I. (2022). Hematological Parameters in Individuals with β -Thalassemia Trait in South Sumatra, Indonesia. *Journal of Blood Medicine*.
- Salim, Y., N. Sukartini, & A. Setiawati. (2016). Erythrocyte indices to differentiate iron deficiency anemia from β trait thalassemia. *Clinical Pathology and Medical Laboratory*. 23(1):50–55.
- Taher, A., Isma'eel, H. and Cappellini, M. D. (2006) Thalassemia intermedia: Revisited, *Blood Cells, Molecules, and Diseases*, 37(1), pp. 12-20.
- Origa, R. (2015). *β -Thalassemia*. NCBI Bookshelf. A service of the National Library of Medicine, National Institutes of Health. pp.1-30.
- Origa, R. (2017). β -Thalassemia. *Genetics in Medicine*, 19(6), pp.609–619.
- Old, J. M. (1991). *Detection of mutations by the Amplification Refractory Mutation System (ARMS)*. In: Mathew C.G. (eds) *Protocols in Human Molecular Genetics*. Methods in Molecular Biology. Vol 9. Springer. Totowa, NJ.

- Thein, S.L. (2011). *Abnormalities of the structure and synthesis of hemoglobin. In Blood and Bone Marrow Pathology* (pp. 132-135). StatPearls Publishing. Treasure Island.
- Treisman, R., S. H. Orkin, & T. Maniatis. 1983. Specific transcription and rna splicing defects in five cloned β -thalassaemia genes. *Nature*. 302: 591–596.
- ‘Ulya, N.M., V.N. Indrawati, W.T. Wulansari, I.Lesmana & N.S.N. Handayani. (2023). Mutation spectrum of β -globin gene in patients with β -thalassemia at Tidar Hospital, Magelang, Central Java, Indonesia. *Hemoglobin*. 47 (4): 152-156.
- ‘Ulya, N.M. (2022). *Deteksi Mutasi IVSI-5 (G>C) dan IVSI-1 (G>T) Gen Pengkode β -globin pada Pasien β -thalassemia di RSUD Tidar Kota Magelang*(Skripsi,UGM).<https://etd.repository.ugm.ac.id/penelitian/detail/216122>
- Wahed, A. & A. Dasgupta. (2015). *Hematology and Coagulation*. Elsevier. USA. pp. 55–61.
- Weatherall, D.J. and Clegg, J.B. (2001) *The Thalassemia Syndromes*. 4th edn. Oxford: Blackwell Science.
- Whipple, G. H., & Bradford, W. L. (1932). Racial or Mediterranean anemia: Thalassemia. *American Journal of Diseases of Children*, 44(2), 336-365.
- Yang, H., Han, S., Xu, J., He, S., Lu, Q., Luo, T., Chen, S., Dang, L., Wang, G., Li, J., Huang, M., Liao, Y., He, Y., Cai, N., Huang, L., Zhou, M., Mo, Y., Zhu, W., Wu, Z. and Zhou, G. (2025). Effects of Thalidomide on Metabolism and Lifespan of Red Blood Cell in Patients With β -Thalassemia Major: A Post Hoc Analysis of a Randomized Controlled Trial. *Clinical Therapeutics*.
- Vuong, H., Nguyen, P.-D., Thi, N.N., Thi, P.L., Minh, T., Nguyen, M.H., Tran, H.A., Dang-Tran, N.-M., Bui, T.-H., Tran, T.H., Ta, T.V. and Tran, V.K. (2024). Application of short tandem repeats (STRs) in the preimplantation genetic diagnosis (PGD) of α -thalassemia. *Taiwanese journal of obstetrics and gynecology*, 63(3), pp.375–380.
- Vichinsky, E. P. (2005). Changing patterns of thalassemia worldwide. *Annals of the New York Academy of Sciences*, 1054(1), 18-24.
- Zheng, J.-T., Lin, C.-X., Fang, Z.-Y. dan Li, H.-D. (2020) Intron Retention as a Mode for RNA-Seq Data Analysis. *Frontiers in Genetics*. 11. p.586.