



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 (Studi di RSUD Tidar Kota Magelang). *Jurnal Kesehatan Masyarakat (Undip)* 8(1): 23-29.
- Arif, A.A., A.D. An-Nizamiya, C. Putri, M. Nashrurrokhman, N. Husna, Mulyati, S. Hadisusanto & N.S.N. Handayani. 2020. Comparison between three molecular diagnostics for the identification of heterozygous hemoglobin E. *Pakistan Journal of Biological Sciences* 23(1): 17-26.
- Billett, H.H. 1990. Hemoglobin and Hematocrit. In: Walker HK, Hall WD, Hurst JW (Ed). Clinical Methods: The History, Physical, and Laboratory Examinations. 3rd edition. Boston: Butterworths.
- Brancaleoni, V., I. Di Pierro, I. Motta & M.D. Cappellini. 2016. Laboratory diagnosis of thalassemia. *Int J Lab Hematol* 1:32-40.
- Brinkley, P., D.S. Bautista & F.L. Graham. 1991. The Cleavage Site of Restriction Endonuklease *MnII*. *Gene* 100:267-268.
- Cao, A. & R. Galanello. 2010. β -thalassemia. *Genet Med.* 12(2):61-76
- Coviello, A.D., B. Kaplan, K.M. Lakshman, T. Chen, A.B. Singh & S. Bhasin. 2008. Effects of graded doses of testosterone on erythropoiesis in healthy young and older men. *J Clin Endocrinol Metab.* 93(3):914-919.
- Datkhile, K.D., M.N. Patil, R.D. Vavhal & T.S. Khamkar. 2015. The spectrum of β -globin gene mutations in thalassemia patients of south-western maharashtra: A cross sectional study. *Journal of Krishna Institute of Medical Sciences* 4(3): 49-58.
- Dzierzak, E. & S. Philipsen. 2013. Erythropoiesis: development and differentiation. *Cold Spring Harb. Perspect. Med.* 3:a011601.
- El-Gawhary, S., S. El-Shafie, M. Niazi, M. Aziz & A. El-Beshlawy. 2007. Study of β -thalassemia mutations using the polymerase chain reaction-amplification refractory mutation system and direct DNA sequencing techniques in a group of Egyptian thalassemia patients. *Hemoglobin* 31(1): 3-9.
- Farashi, S. & C.L. Harteveld. (2018). Molecular basis of α -thalassemia. *Blood Cells, Molecules, and Diseases* 70: 43–53.
- Ford, J. 2013. Red blood cell morphology. *Int. Soc. Lab. Hematol.* 35: 351–357
- Forget, B.G. & R.C. Hardison. 2008. The normal structure and regulation of human globin gen clusters. *NIH grants* 3: 3-5.
- Fucharoen, S. & D.J. Weatherall. 2012. The haemoglobin e thalassemia. *Cold Spring Harb Perspect Med.* 2(8): 1-15.
- Fucharoen, S. & P. Winichagoon. 2011. Haemoglobinopathies in Southeast Asia. *The Indian journal of medical research* 134(4): 498–506.
- Gallagher, S. 2001. Quantitation of nucleic acids with absorption spectroscopy. *Curr Protoc Protein Sci:* 18429085.
- Galanello, R. & A. Cao. 2011. Gene test review Alpha-thalassemia. *Genet Med*13(2): 83-87.
- Galanello R. & R. Origa. 2010. Beta-thalassemia. *Orphanet Journal of Rare Diseases* 5 (11):1-15.
- Galanello, R. & A. Cao. 2011. Alpha-thalassemia. *Genet Med* 13: 83– 88.



- Grosso, M., R. Sessa, S. Puzone S, M. Rosasria & P. Izzo. 2015. Molecular Basis of Thalassemia in Anemia, Dr. Donald Silverberg (Ed.) (pp.342-348). Intech. Rijeka
- Handayani, N.S.N. & R. Purwanto. 2015. CD35 (DEL C) frameshift mutation in ekson 2 of β -globin gene on β -thalassemia carriers. *Biomedical Engineering*. 1(1): 19-23.
- Handayani, N.S.N., N. Husna, G. Rahmil, R.A. Ghifari, L. Widyawati & I. Lesmana. 2021. Splice-site and frameshift mutations of β -globin gene found in thalassemia carrier screening in Yogyakarta Special Region, Indonesia. *The Indonesian Biomedical Journal*. 13(1): 55-60.
- Hernanda, P.Y., L. Tursilowati, S.G. Arkesteijn, I.D. Ugrasena, M.C. Larasati, S.M. Soeatmadji, P.C. Giordano & C.L. Harteveld. 2012. Towards a prevention program for β -thalassemia. The molecular spectrum in East Java, Indonesia. *Hemoglobin* 36 (1):1-6.
- Higgins, J. 2015. Red blood cell population dynamics. *Clin. Lab. Med.* 35: 43-57.
- Husna, N., I. Sanka, A.A. Arif, C. Putri, E. Leonard & N.S.N. Handayani. 2017. Prevalence and distribution of thalassemia trait screening. *J Med Sci* 49 (3): 106-113.
- Jannah, M. 2014. Profil Hematologis dan Deteksi Molekular Pembawa Sifat Hemoglobin E di Yogyakarta. Tesis. Tidak Diterbitkan. UGM. Yogyakarta.
- 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.
- Kong, Y., S. Zhou, A.J. Kihm, A.M. Katein, X. Yu, D.A. Gell, J.P. Mackay, K. Adachi, L. Foster-Brown, C.S. Louden, A.J. Gow & M.J. Weiss. 2004. Loss of alpha-hemoglobin-stabilizing protein impairs erythropoiesis and exacerbates beta-thalassemia. *The Journal of clinical investigation* 114(10): 1457–1466.
- Kumar, A. & N. Garg. 2005. *Genetic Engineering*. Nova Science Publisher Inc, New York. pp.54-58.
- Lie-Injo, L.E., Cai. S.P, I. Wahidijat, S. Moeslichan, M.L. Lim, L. Evangelista, M. Doherty & Y.W. Kan. 1989. β -thalassemia mutations in Indonesia and their linkage to b haplotypes. *Am J Hum Genet*. 45(6): 971–975.
- Little S. 2001. Amplification-refractory mutation system (ARMS) analysis of point mutations. *Curr Protoc Hum Gene*: 1-2.
- Lorenz, T. C. 2012. Polymerase chain reaction: basic protocol plus troubleshooting and optimization strategies. *Journal of visualized experiments* 63: 1-15
- McPherson, M.J. & S.G. Moller. 2006. *PCR: The Basics*, 2nd Edition. Taylor and Francis Group, New York. pp. 9-29.
- Moiz, B., M.R. Hashmi, A. Nasir, A. Rashid & T. Moatter. 2012. Hemoglobin E Syndrome in Pakistani Population. *BMC Blood Disorder* 12 (3): 6.
- Nienhuis, A. W. & D.G. Nathan. 2012. Pathophysiology and Clinical Manifestations of the β -Thalassemias. *Cold Spring Harbor perspectives in medicine* 2 (12): 1-9.
- Olivieri, N.F. 1999. The β -thalassemias. *N Engl J Med* 341(2):99–109.
- Origa, R. 2015. β -Thalassemia. NCBI Bookshelf. A service of the National Library of Medicine, National Institutes of Health. pp.1-30.



- Perera, S., A. Allen, I. Silva, M. Hapugoda, M.N. Wickramarathne, I. Wijesiriwarden & A. Premawardhena. 2019. Genotype-phenotype association analysis identifies the role of α globin genes in modulating disease severity of β -thalassaemia intermedia in Sri Lanka. *Scientific Reports* 9 (1): 1-9.
- Rogers, K. 2011. *The Human Body Blood Physiology and Circulation*. Britannica Educational Publishing, New York. pp. 11-12, 19, 35.
- Rujito, L. 2019. *Buku Referensi Thalassemia: Genetik Dasar dan Pengelolaan Terkini*. UNSOED Press, Purwokerto. pp. 8-37.
- 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): 330-333.
- Sherwood, L. 2010. *Human Physiology: From Cells to Systems*, 7th edition. Books/Cole Cengage Learning, Belmont. pp. 394-395.
- Susanto, Z., W. Siswandari. & L. Rujito. 2020. Cd60 (GTG > GAG)/Hb Cagliari mutation was found in scanning of β -thalassemia alleles from patients of East Kalimantan, Indonesia. *Molecular Genetics and Metabolism Reports* 22: 100550.
- Susanto, Z.A., W. Siswandari & L. Rujito. 2020. Korelasi Gneotipe-Fenotipe Pasien Talasemia Beta di Kota Samarinda Kalimantan Timur Tahun 2019. *Buletin Penelitian Kesehatan* 48 (2): 91-98.
- Siswandari, W., L. Rujito, V. Indriani & W. Djatmiko. 2019. Mentzer Index Diagnostic Value in Predicting Thalassemia Diagnosis. *IOP Conf. Series: Earth and Environmental Science* 255: 1-6.
- Tamam M., S. Hadisaputro, Sutaryo, I. Setianingsih, R. Astuti & A. Soemantri. 2010. Hubungan antara tipe mutasi gen globin dan manifestasi klinis penderita thalassemia. *Jurnal kedokteran brawijaya* 26: 48-52.
- Thein, S.L. 2013. The molecular basis of β -thalassemia. *Cold Spring Harb Perspect Med* 13 (5): 1-24.
- 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.
- Thom, C.S., C.F. Dickson, D.A. Gell & M.J. Weiss. 2013. Hemoglobin variants: biochemical properties and clinical correlates. *Cold Spring Harbor perspectives in medicine* 3 (3): a011858.
- Traivaree, C., C. Monsereenusorn, P. Rujkijyanont, W. Prasertsin & B. Boonyawat. 2018. Genotype-phenotype correlation among beta-thalassemia and beta-thalassemia / HbE disease in Thai children: predictable clinical spectrum using genotypic analysis. *J. Blood. Med*: 35- 41
- Vichinsky, E. 2010. Complexity of α thalassemia: growing health problem with new approaches to screening, diagnosis, and therapy. *Ann N Y Acad Sci* 12 (2) :180-187.
- Virprakasit, V., C. L. Lee, Q. T. Chong, K. H. Lin & A. Khuhapinant. 2009. Iron Chelation Therapy in the Management of Thalassemia: The Asian Perspective. *International Journal of Hematology* 90: 435-4
- Yayasan Thalassemia Indonesia. 2018. Laporan Kasus Thalassemia Indonesia. Jakarta: