

## 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.
- Aliviameita, A. and Puspitasari, P., 2024. Pemeriksaan Hematologi Rutin. *Umsida Press*, pp.1-96.
- Amalia, R.A., 2020. *Perbedaan Hasil Hitung Jumlah Eritrosit Menggunakan Larutan Hayem dan Larutan Gower (Doctoral dissertation, Universitas Muhammadiyah Semarang)*.
- Anna, A. & G. Monika. 2018. Splicing mutations in human genetic disorders: examples, detection, and confirmation. *Journal of Applied Genetics*. 59(3): 253–268.
- Apriliansi, Indah Nur. 2014. *Hitung Jumlah Eritrosit Pada Pekerja Penambang Batu Kapur di Tegal* (Thesis, Fakultas Ilmu Keperawatan Dan Kesehatan Universitas Muhammadiyah, Semarang).
- 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.
- Arumdapta, K.T. 2026. *Deteksi Mutasi Cd26 Gen HBB pada Keluarga Pasien  $\beta$ -Thalassemia di RSUD Tidar Kota Magelang* (Skripsi, UGM).
- Bain, B. J. 2020. *Haemoglobinopathy Diagnosis*. 3rd Edition. John Wiley & Sons. UK. pp.1–6; 9.
- Basu, D. & R. Kulkarni, 2014. Overview of blood components and their preparation. *Indian Journal of Anaesthesia*. 58(5): 529–537.
- 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.
- Chaudhary, H. K., M. Shrestha, P. Chaudhary, & B. H. Poudel. 2017. Amplification refractory mutation system – polymerase chain reaction for rapid detection of rpoB gene mutation in *Mycobacterium tuberculosis*. *International Journal of Applied Science and Biotechnology*. 5(1): 81–85.
- Chojnowska, S., Baran, T., Wilińska, I., Sienicka, P., Cabaj-Wiater, I. and Knaś, M., 2018. Human saliva as a diagnostic material. *Advances in medical sciences*, 63(1), pp.185-191.
- Farashi, S. & C. L. Harteveld. 2018. Molecular basis of  $\alpha$ -thalassemia. *Blood Cell, Molecules, and Diseases*. 70:43–53.
- Firdayanti, Umar, A., Susanti, Ismawatie, E., Sari, A.I., Supriyanta, B., Dewi, Y. R., Yashir, M., Chairani, Anggraini, F. T., Rahayu, M., Gunawan, L. S., Tuntun, M., Wibowo, S., Thaslifia., & Wenty, D. 2024. *Dasar-dasar Hematologi*. Purbalingga: Eureka Media Aksara.
- Galanello, R. & R. Origa. 2010. Beta-thalassemia. *Orphanet Journal of Rare Disease*. 5(11): 1–15.
- Guyton, A. C. & J. E. Hall. 2011. *Medical Physiology*. 12th Edition. Saunders Elseiver. 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. Yussof, S. Salleh, & B. A. Zilfalil. 2014. Multiplex amplification refractory mutation system (MARMS) for the detection of  $\beta$ -globin gene mutations among the transfusion-dependent  $\beta$  thalassemia Malay patients in Kelantan, Northeast of Peninsular Malaysia. *American Journal of Blood Research*. 4(1): 33–40.
- Handayani, N. S. N., N. Husna, G. Rahmil, R. A. Ghifari, L. Widyawati, & I. Lesmana. 2020. Splice-site and frameshift mutation of  $\beta$ -globin gene found in thalassemia carrier screening in Yogyakarta special region, Indonesia. *Indones Biomed J*. 13(1): 55–60.
- Harahap, A.S., 2017. UJI KUALITAS DAN KUANTITAS DNA BEBERAPA POPULASI POHON KAPUR SUMATERA: Ariani Syahfitri Harahap. *Jasa Padi*, 2(2): 1-6.
- Haris, N., Hajrial, A, Nurita. T.M, dan Agus. P. 2003. Kemiripan genetik klon karet (*Hevea brasiliensis* Muell Arg.) berdasarkan metode amplified fragment length polymorphisms (AFLP). *Menara Perkebunan*, 71(1): 1-15.
- Hassan, S., R. Ahmad, Z. Zakaria, Z. Zulkafli, & W. Z. Abdullah. 2012. Detection of  $\beta$ -globin gene mutations among  $\beta$ -thalassemia carriers and patient in Malaysia: application of multiplex amplification refractory mutation system-polymerase chain reaction. *Malaysian Journal Medicine Science*. 20(1):13–20.
- Hidayati, N. I., N. Wijayanti, & N. S. N. Handayani. 2020. Detection of HBB:c.92+5G>C and HBB:c.108delC mutations in  $\beta$ -thalassemia carriers using high-resolution melting analysis. *Molecular Biology Reports*. 47:5665–5671.
- Hoffbrand, A. V. & P. A. H. Moss. 2011. *Essential Haemology*. 6th Edition. John Wiley & Sons, Inc. UK.
- Indrawati, V.N. 2022. *Deteksi Mutasi Cd 26 dan Cd 35 Gen Pengkode  $\beta$ -Globin pada Pasien  $\beta$ -thalassemia di RSUD Tidar Kota Magelang* (Skripsi, UGM). <https://etd.repository.ugm.ac.id/penelitian/detail/216063>
- Jannah, M. 2014. *Profil Hematologis dan Deteksi Molekular Pembawa Sifat Hemoglobin E di Yogyakarta*. Tesis. Tidak Diterbitkan. UGM. Yogyakarta.
- Lie-Injo, L.E., Cai. S.P, I. Wahidijat, S. Moeslichan, M.L. Lim, L. Evangelista, M. Doherty & Y.W. Kan. 1989.  $\beta$ -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.
- Marengo-Rowe, A. J. M. D. 2007. The thalassemias and related disorders. *Baylor University Medical Center Proceedings*. 20(1):27–31.
- Nienhuis, A. W. & D. G. Nathan. 2012. Pathophysiology and clinical manifestations of the  $\beta$ -thalassemias. *Cold Spring Harb Perspect Med*. 2:a011726.
- Nur, A. and Yamamoto, Z., 2022. Saliva sebagai sumber DNA genom manusia. *Jurnal Kedokteran Syiah Kuala*, 22(2): 127-131.
- 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.
- Old, J., C. L. Hartevelde, J. Traeger-Synodinos, M. Petrou, M. Angastiniotis, & R. Galanello. 2012. *Prevention Of Thalassaemias And Other Haemoglobin Disorders Volume 2: Laboratory Protocols*. 2nd Edition. Thalassaemia International Federation Publication. Cyprus. pp. 89–91.

- Pardosi, Z.R.H., 2025. *PEMERIKSAAN KADAR HEMOGLOBIN PADA PASIEN GAGAL GINJAL KRONIS SEBELUM DAN SESUDAH MELAKUKAN HEMODIALISA DI RSUD H. ABDUL MANAN SIMATUPANG KISARAN* (Doctoral dissertation, Poltekkes Medan).
- Roca, X., R. Sachidanandam, & A. R. Krainer. 2003. Intrinsic differences between authentic and cryptic 5' splice sites. *Nucleic Acids Research*. 31(21):6321–6333.
- Rongers, K. 2011. *The Human Body Blood Physiology and Circulation*. Britannica Educational Publishing. New York. pp. 17–19.
- Rujito, L., M. Basalamah, S. Mulatsih, & A. Salam. 2015. Molecular scanning of  $\beta$ -thalassemia in the Southern Region of Central Java, Indonesia; a step towards a local prevention program. *Journal Hemoglobin*. 39(5): 330–333.
- Rujito, L. 2019. *Talasemia: Genetik Dasar dan Pengelolaan Terkini*. Universitas Jendral Soedirman Press. Purwokerto.
- Saha, D., M. Patgaonkar, A. Shroff, K. Ayyar, T. Bashir, & K. V. R. Reddy. 2014. Hemoglobin expression in nonerythroid cells novel or ubiquitous. *International Journal of Inflammation*. 2014:1–8.
- Schaller, J., S. Gerber, U. Kämpfer, S. Lejon, & C. Trachsel. 2008. *Human Blood Plasma Proteins Structure and Function*. John Wiley & Sons Ltd. West Sussex. pp. 7–8.
- Tamam, M., S. Hadisaputro, Sutaryo, I. Setianingsih, R. Astuti, & A. Soemantri. 2010. Hubungan antara tipe mutasi gen globin dan manifestasi klinis penderita talassemia. *Jurnal Kedokteran Brawijaya*. 26:48–52.
- Thein, S. L., P. Winichagoon, C. Hesketh, S. Best, S. Fucharoen, P. Wasi, & D. J. Weatherall. 1990. The molecular basis of  $\beta$ -thalassemia in Thailand: application to prenatal diagnosis. *Am J Hum Genet*. 47:369–375.
- Thein, S. L. 2013. The molecular basis of  $\beta$ -thalassemia. *Cold Spring Harb Perspect Med*. 3:a011700.
- Treisman, R., S. H. Orkin, & T. Maniatis. 1983. Specific transcription and rna splicing defects in five cloned  $\beta$ -thalassaemia genes. *Nature*. 302: 591–596.
- Ulya, N.M. 2022. *Deteksi Mutasi IVSI-5 (G>C) dan IVSI-1 (G>T) Gen Pengkode  $\beta$ -globin pada Pasien  $\beta$ -thalassemia di RSUD Tidar Kota Magelang* (Skripsi, UGM). <https://etd.repository.ugm.ac.id/penelitian/detail/216122>
- Ulya, N.M., Indrawati, V.N., Wulansari, W.T., Lesmana, I. and Handayani, N.S.N. 2023. Mutation Spectrum of  $\beta$ -Globin Gene in Patients with  $\beta$ -Thalassemia at Tidar Hospital, Magelang, Central Java, Indonesia. *Hemoglobin*, 47(4), pp.152-156.
- Wahed, A. & A. Dasgupta. 2015. *Hematology and Coagulation*. Elsevier. USA. pp. 55–61.
- Wattendorf, D.J. and Hadley, D.W., 2005. Family history: the three-generation pedigree. *American family physician*, 72(3), pp.441-448.
- Yang, L., I. Ijaz, J. Cheng, C. Wei, X. Tan, M. A. Khan, X. Fu, & J. Fu. 2018. Evaluation of amplification refractory mutation system (ARMS) technique for quick and accurate prenatal gene diagnosis of CHM variant in choroideremia. *The Application of Clinical Genetics*. 11:1–8.
- Yayasan Thalassemia Indonesia. 2021. *Laporan kasus thalassemia Indonesia*. Jakarta.