

DAFTAR PUSTAKA

- Bajwa H and Basit H. Thalassemia. (2023) [Updated 2023 Aug 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan-. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK545151/>
- Ban, Q., Lee, J., Shi, Z., Lu, D., Qiao, L., Yang, P., Li, X., Cheng, H., Zhang, M., Hou, J., Yao, J.H., Wang, J., Huang, P.-Y., Tseng, H., Zhu, Y., Chen, L.-C., Hui, W. and Liu, D. (2022). Intraosseous injection of SMNP vectors enables CRISPR/Cas9-mediated knock-in of HBB gene into hematopoietic stem and progenitor cells. *Nano Today*, 47, pp.101659–101659. doi:<https://doi.org/10.1016/j.nantod.2022.101659>.
- Chu, S.N., Soupene, E., Sharma, D., Sinha, R., McCreary, T., Hernandez, B., Shen, H., Wienert, B., Bowman, C., Yin, H., Lesch, B.J., Jia, K., Romero, K.A., Kostamo, Z., Zhang, Y., Tran, T., Cordero, M., Homma, S., Hampton, J.P. and Gardner, J.M. (2025). Dual α -globin-truncated erythropoietin receptor knockin restores hemoglobin production in α -thalassemia-derived erythroid cells. *Cell Reports*, 44(1), pp.115141–115141. doi:<https://doi.org/10.1016/j.celrep.2024.115141>.
- Darudhita, A.B. (2025). Deteksi mutasi IVSI-5 (G>C) Gen B-Globin pada Keluarga Pasien B-Thalassemia di RSUD Tidar, Kota Magelang [undergraduate's thesis]. Yogyakarta (YK): Universitas Gadjah Mada.
- Ekwattanakit, S., Monteerarat, Y., Riolueang, S., Tachavanich, K. and Viprakasit, V. (2012). Association of *XmnI* Polymorphism and Hemoglobin E Haplotypes on Postnatal Gamma Globin Gene Expression in Homozygous Hemoglobin E. *Advances in Hematology*, 2012, pp.1–5. doi:<https://doi.org/10.1155/2012/528075>.
- Harewood J and Azevedo AM. Alpha Thalassemia. (2025). [Updated 2023 Sep 4]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK441826/>
- Hernaningsih, Y., Syafitri, Y., Indrasari, Y.N., Rahmawan, P.A., Andarsini,

- M.R., Lesmana, I., Moses, E.J., Abdul Rahim, N.A. and Yusoff, N.M. (2022). Analysis of Common Beta-Thalassemia (β -Thalassemia) Mutations in East Java, Indonesia. *Frontiers in Pediatrics*, 10. doi:<https://doi.org/10.3389/fped.2022.925599>.
- Huang, N., Wang, Y., Huang, H., Chen, Z. and Zhang, Z. (2024). Genotype phenotype correlation analysis of patients with thalassemia in Quanzhou city, Southeast of China. *Heliyon*, 10(22), pp.e40144–e40144. doi:<https://doi.org/10.1016/j.heliyon.2024.e40144>.
- Jannah M. (2014). Profil hematologis dan deteksi molekular pembawa sifat hemoglobin E di Yogyakarta [master's thesis]. Yogyakarta (YK): Universitas Gadjah Mada.
- Jarujareet, U., Wiratchawa, K., Petiwathayakorn, T., Koonyosying, P., Hantrakool, S., Srichairatanakool, S. and Intharah, T. (2024). Classification of beta-thalassemia major and HbE/beta-thalassemia via deep learning of image structure function image. *Biomedical Signal Processing and Control*, 102, pp.107265–107265. doi:<https://doi.org/10.1016/j.bspc.2024.107265>.
- Kountouris, P., Michailidou, K., Christou, S., Hadjigavriel, M., Sitarou, M., Kolnagou, A., Kleanthous, M. and Telfer, P. (2020). Effect of HBB genotype on survival in a cohort of transfusion-dependent thalassemia patients in Cyprus. *Haematologica*, 106(9), pp.2458–2468. doi:<https://doi.org/10.3324/haematol.2020.260224>.
- Kriukiene, E., Lubiene, J., Lagunavicius, A. and Lubys, A. (2005). *MnII*—The member of H-N-H subtype of Type IIS restriction endonucleases. *Biochimica et Biophysica Acta (BBA) - Proteins and Proteomics*, 1751(2), pp.194–204. doi:<https://doi.org/10.1016/j.bbapap.2005.06.006>.
- Kuchay, M.S., Mithal, A. and Yedla, N. (2015). Hemoglobin E disease and glycosylated hemoglobin. *Indian Journal of Endocrinology and Metabolism*, 19(5), p.683. doi:<https://doi.org/10.4103/2230-8210.163211>.
- Lama, R., Yusof, W., Shrestha, T.R., Hanafi, S., Bhattarai, M., Hassan, R. and Zilfalil, B.A. (2021). Prevalence and distribution of major β -thalassemia mutations and HbE/ β -thalassemia variant in Nepalese ethnic groups.

Hematology/Oncology and Stem Cell Therapy.

doi:<https://doi.org/10.1016/j.hemonc.2021.01.004>.

Langer AL. Beta-Thalassemia. (2000) [Updated 2024 Feb 8]. In: Adam MP, Feldman J, Mirzaa GM, *et al.*, editors. *GeneReviews® [Internet]*. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1426/>

Liebhaber, S.A., Cash, F.E. and Ballas, S.K. (1986). Human alpha-globin gene expression. The dominant role of the alpha 2-locus in mRNA and protein synthesis. *Journal of Biological Chemistry*, 261(32), pp.15327–15333. doi:[https://doi.org/10.1016/s0021-9258\(18\)66871-1](https://doi.org/10.1016/s0021-9258(18)66871-1).

Matsuno, Y., Yamashiro, Y., Yamamoto, K., Hattori, Y., Yamamoto, K., Ohba, Y., and Miyaji, T. (1992). A possible example of gene conversion with a common beta-thalassemia mutation and chi sequence present in the beta-globin gene. *Hum. Genet.* 88: 357-358.[PubMed: [1733840](#)] [[Full Text](#)]

Mitra, N., Chowdhury, P. and Basu, A. (2025). Exploring the functional and immune landscape of E- β thalassemia patients through RNA sequencing of peripheral blood mononuclear cells. *Heliyon*, 11(1), p.e41255. doi:<https://doi.org/10.1016/j.heliyon.2024.e41255>.

Moiz, B., M.R. Hashmi, A. Nasir, A. Rashid and T. Moatter. (2012). Hemoglobin E Syndrome in Pakistani Population. *BMC Blood Disorder* 12 (3): 6.

Needs T, Gonzalez-Mosquera LF, Lynch DT. Beta Thalassemia. (2023) [Updated 2023 May 1]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK531481/>

Old, J., Harteveld, C.L., Traeger-Synodinos, J., Petrou, M., Angastiniotis, M. and Galanello, R. (2014). *Prevention of thalassaemias and other haemoglobin disorders: volume 2: laboratory protocols.*

Origa, R. (2017). β -Thalassemia. *Genetics in Medicine*, 19(6), pp.609–619. doi:<https://doi.org/10.1038/gim.2016.173>.

Rahaman, M., Bhattacharya, S., Vaddi, R., Mukherjee, M., Mani, D., Jain, M.,

- Dolai, T.K., Chakrabarti, P., Purwar, S., Dhingra, B., Shukla, P.C., Mukherjee, G., Mukherjee, B. and Chakravorty, N. (2025). Two missense mutations (p.H63D and p.C282Y) in HFE gene elevate the risk of iron-overload in HbE/ β -thalassemia disease. *Gene Reports*, [online] 39, p.102165. doi:<https://doi.org/10.1016/j.genrep.2025.102165>.
- Ropero, P., Fernández, F.A.G., Nieto, J.M., Recasens, V., Montañés, Á., Murúzabal, M.J., Sarasa, M., Fernández, C., Villegas, A. and Benavente, C.C. (2022). Does size matter? Two new deletions in the HBB gene cause β^0 -thalassemia. *Annals of hematology*, [online] 101(7), pp.1465–1471. doi:<https://doi.org/10.1007/s00277-022-04837-4>.
- Roth, I.L., Lachover, B., Koren, G., Levin, C., Zalman, L. and Koren, A. (2018). Detection of B Thalassemia Carriers by Red Cell Parameters Obtained from Automatic Counters Using Mathematical Formulas. *Mediterranean Journal of Hematology and Infectious Diseases*, [online] 10(1), p.2018008. doi:<https://doi.org/10.4084/mjhid.2018.008>.
- Sahiratmadja, E., Maskoen, A.M., Reniarti, L. and Prihatni, D. (2022). Erythrocyte Indices MCV and/or MCH as First Round Screening Followed by Hb-analysis for β -thalassemia Carrier State. *The Indonesian Biomedical Journal*, 14(3), pp.282–8. doi:<https://doi.org/10.18585/inabj.v14i3.1960>.
- Shafique, F., Ali, S., Almansouri, T.S., van Eeden, F., Shafi, N., Khalid, M., Khawaja, S., Andleeb, S. and Ul Hassan, M. (2023). Thalassemia, a human blood disorder. *Brazilian Journal of Biology*, 83. doi:<https://doi.org/10.1590/1519-6984.246062>.
- Soremekun, O.S., Ezenwa, C., Isewon, I., Soliman, M., Idowu, O., Nashiru, O. and Fatumo, S. (2020). Computational and drug target analysis of functional single nucleotide polymorphisms associated with Haemoglobin Subunit Beta (HBB) gene. *Computers in Biology and Medicine*, 125, p.104018. doi:<https://doi.org/10.1016/j.combiomed.2020.104018>.
- Tamary H and Dgany O. Alpha-Thalassemia. (2005) [Updated 2024 May 23]. In: Adam MP, Feldman J, Mirzaa GM, *et al.*, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1435/>

- Tari, K., Ardalan, P.V., Dibavar, M.A., Atashi, A., Jalili, A. and Gheidishahran, M. (2018). Thalassemia an update: molecular basis, clinical features and treatment. *International Journal of Biomedicine and Public Health*, 1(1), pp.48–58. doi:<https://doi.org/10.22631/ijbmph.2018.56102>.
- 'Ulya, N. M., Indrawati, V. N., Wulansari, W. T., Lesmana, I., and Handayani, N. S. N. (2023). Mutation Spectrum of β -Globin Gene in Patients with β -Thalassemia at Tidar Hospital, Magelang, Central Java, Indonesia. *Hemoglobin*, 47(4), 152–156. <https://doi.org/10.1080/03630269.2023.2244429>
- Viljoen, C.D., Booysen, C. and Tantuan, S.S. (2022). The suitability of using spectrophotometry to determine the concentration and purity of DNA extracted from processed food matrices. *Journal of Food Composition and Analysis*, [online] 112, p.104689. doi:<https://doi.org/10.1016/j.jfca.2022.104689>.
- 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. doi:<https://doi.org/10.1016/j.tjog.2023.09.024>.
- Wahidiyat, P.A., Sari, T.T., Rahmartani, L.D., Iskandar, S.D., Pratanata, A.M., Yapiy, I., Setianingsih, I., Atmakusuma, T.D. and Lubis, A.M. (2022). Thalassemia in Indonesia. *Hemoglobin*, 46(1), pp.39–44. doi:<https://doi.org/10.1080/03630269.2021.2023565>.
- Wang, W.-H., Lin, C.-Y., Jain, S.-H., Lu, P.-L. and Chen, Y.-H. (2024). Development of the novel Gene Chip and Restriction Fragment Length Polymorphism (RFLP) methods for rapid detection of Mycobacterium tuberculosis complex in broth culture. *Journal of Microbiology Immunology and Infection*, 58(1). doi:<https://doi.org/10.1016/j.jmii.2024.09.003>.
- Xu, M.R., Yang, B.C., Chang, H.C., Kuo, C.L., Lin, C.H., Chen, H.J., Cheng, J.H. and Lee, M.S. (2022). Molecular authentication of the medicinal crop *Portulaca oleracea* and discrimination from its adulterants in herbal markets

using PCR-restriction fragment length polymorphism (PCR-RFLP) analysis. *Industrial Crops and Products*, 183, pp.114934–114934. doi:<https://doi.org/10.1016/j.indcrop.2022.114934>.

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*. doi:<https://doi.org/10.1016/j.clinthera.2025.01.008>.

Zhou, J., Liu, C., Hao, N., Feng, J., Quan, Z., Chen, L. and Liu, J. (2025). Thalassemia genetic screening of pregnant women with anemia in Northern China through comprehensive analysis of thalassemia alleles (CATSA). *Clinica Chimica Acta*, 569, p.120151. doi:<https://doi.org/10.1016/j.cca.2025.120151>