



PUSTAKA ACUAN

- Alauddin, H., K. Kamarudin, T.Y. Loong, R.Z. Azma, A. Ithnin, N. Jalil, N.F. Razak, D. Koh-Xuan-Rong, E. Ismail, L. C-Khai, Z.A. Latiff, H. Alias, & A. Othman. 2018. A unique interaction of IVS-I-1 (G>A) (HBA2: c.95+1G>A) with Hb Adana (HBA2: c.179G>A) presenting as transfusion-dependent α -Thalassemia. *Hemoglobin*, 42(4), 247–251.
- Aldakeel, S.A., N.Z. Ghanem, A.M. Al-Amadi, A.K. Osman, L.I.A. Asoom, N.R. Ahmed, N.B. Almandil, M.S. Akhtar, S.A. Azeez, & J.F. Borgio. 2018. Identification of seven novel variants in the β -globin gene in transfusion-dependent and normal patients. *Archives of Medical Science*, 16(2): 453-459.
- Anna, A., & G. Monika. 2018. Splicing mutations in human genetic disorders: examples, detection, and confirmation. *Journal of Applied Genetics*, 59(3): 253-268.
- Arviananta, R., Syuhada, Aditya. 2020. Perbedaan jumlah eritrosit antara darah segar dan darah simpan di UTD RSAM Bandar Lampung. *Jurnal Ilmiah Kesehatan Sandi Husada*, 9(2): 686-694.
- Bachir, D., & F. Galacteros. 2004. Hemoglobin E. *Orphanet Encyclopedia*.
- Borah, M.S., P.K. Bhattacharya, & M.S. Pathak. Study of IVS 1-5 (G>C) mutation in beta-thalassemia patients of a tertiary care hospital of North East India. *International Journal of Science and Research*, 6(6): 246-249.
- Brancaeloni, V., E.D. Pierro, I. Motta, & M.D. Cappellini. 2016. Laboratory diagnosis of thalassemia. *International Journal of Laboratory Hematology*, 38(1): 32-40.
- Cappellini M.D, A. Cohen, A. Eleftheriou, A. Piga, J. Porter, & A. Taher. 2008. *Guidelines for the clinical management of thalassemia* [Internet]. 2nd Revised edition. Thalassaemia International Federation. Nicosia (CY). Chapter 2, Blood Transfusion Therapy in β -Thalassaemia Major.
- Cao, A., & P. Moi. 2002. Regulation of globin genes. *Pediatric Research*, 51(4): 415-421.



- Cao, A., & R. Galanello. 2010. β -thalassemia. *Genetics in Medicine*, 12(2): 61-76.
- Cao, A., & Y.E. Kan. 2013. The prevention of thalassemia. *Cold Spring Harbor Perspective in Medicine*, 3(2): a011775.
- Chaudhry H.S., & M.R. Kasarla. 2023. Microcytic Hypochromic Anemia. [Updated 2022 Aug 22]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK470252/>
- Clancy, S. 2008. RNA splicing: introns, exons, and splicesome. *Nature Education*, 1(1): 31.
- Das, S.W., & G. Talukder. 2002. Beta globin gene and related diseases: a review. *International Journal of Human Genetics*, 2(3): 139-152.
- Das, R., & P. Sharma. 2016. *Molecular genetics of thalassemia syndromes*. Morgan & Claypool Life Science.
- Fucharoen, S., & P. Winichagoon. 2011. Haemoglobinopathies in Southeast Asia. *Indian Journal of Medical Research*, 134(4): 498-506.
- Galanello, R., & A. Cao. 2011. Alpha-thalassemia. *Genetics in Medicine*, 13: 83-88.
- Galanello, R., & R. Oringa. 2010. Beta-thalassemia. *Orphanet Journal of Rare Disease*, 5(11): 1-15.
- Goh, L.P.W., E.T.J. Chong, & P-C. Lee. 2020. Prevalence of alpha (α)-thalassemia in Southeast Asia (2010-2020): a meta-analysis involving 83,674 subjects. *International Journal of Environmental Research and Public Health*, 17(7354): 1-11.
- Haddad, A., P. Tyan, A. Radwan, N. Mallat, & A. Taher. 2014. β -Thalassemia intermedia: a bird's-eye view. *Turkish journal of haematology: official journal of Turkish Society of Haematology*, 31(1), 5–16.
- Hassan, S., R. Ahmad, Z. Zakaria, Z. Zulkafli, & W.Z. Abdullah. 2013. Detection of β -globin gene mutations among β -thalassaemia carriers and patients in Malaysia: application of multiplex amplification refractory mutation system-polymerase chain reaction. *The Malaysian journal of medical sciences: MJMS*, 20(1): 13–20.



- Hanafi, S.B., W.Z. Abdullah, R.A. Adnan, R. Bahar, M.F. Johan, N.F. Azman, N.D. Rashid, S.A.A. Ahmad, R. Hassan, B.A. Zilfalil. 2016. Genotype-phenotype association of HbE/β thalassemia disease and the role of genetic modifiers. *Malaysian Journal of Paediatrics and Child Health*, 22: 1-16.
- Handayani, N.S.N., N. Husna, G. Rahmil, R.A. Ghifari, L. Widyawati, & I. Lesmana. 2021. Split-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.
- Handayani, N.S.N., & A.T. Onggo. 2014. Identifikasi mutasi gen β globin ekson 1 pada pembawa thalassemia. *GENESIS: Jurnal Ilmiah Biologi*, 2(1): 63-69.
- Handayani, N.S.N., & R. Purwanto. 2015. CD35 (DEL C) frameshift mutation in exon 2 of β-globin gene on β-thalassemia carriers. *Biomedical Engineering*, 1(1): 19-23.
- Hapsari, A.T., & L. Rujito. 2015. Diagnostic test of blood index and molecular identification of β-thalassemia carrier in blood donors of Banyumas area. *Jurnal Kedokteran Brawijaya*, 281(1): 233-237.
- Hernaningsih Y., Y. Syafitri, Y.N. Indrasari, P.A. Rahmawan, M.R. Andarsini, & I. Lesmana, E.J. Moses, N.A.A. Rahim, & N.M. Yusoff. 2022. Analysis of common beta-thalassemia (β -thalassemia) mutations in East Java, Indonesia. *Frontiers in Pediatrics*, 10: 925599.
- Hidayati, N.I., N. Wijayanti., & N.S.N Handayani. 2020. Detection of HBB:c.92+5G and HBB:c.108delC mutations in β-thalassemia carriers using high-resolution melting analysis. *Molecular Biology Reports*, 47: 5665-5671.
- Huang, T-L., T-Y. Zhang, C-Y. Song, Y-B. Lin, B—H. Sang, Q-L. Lei, Y. Lv, C-H. Yang, N. Li, X. Tian, Y-H. Yang, & X-W. Zhang. 2020. Gene mutation spectrum of thalassemia among children in Yunan province. *Frontiers in Pediatrics*, 8(159): 1-5.
- Husna, N., I. Sanka, A.A. Arif, C. Putri, E. Leonard, N.S.N. Handayani. 2017. Prevalence and distribution of thalassemia trait screening. *Journal of Medical Science*, 49(3): 106-113.



- İlgün, D, Y.Z. Aral, M. Akcan, Ö. Cartı, & G. Bozkurt. 2021. Evaluation of beta globin gene mutations in beta thalassemia carrier children in Aydin Province and its environment. *Trends in Pediatrics*, 2(1): 8-17.
- Jha, R., & S. Jha. 2014. Beta thalassemia – a review. *Journal of Pathology of Nepal*, 4: 663-671.
- Kattamis, A., G.L. Forni, Y. Aydinok, V. Viprakasit. 2020. Changing patterns in the epidemiology of β -thalassemia. *European Journal of Haematology*, 105(6): 692-703.
- Körber, C., A. Wölfler, M. Neubauer, & C. Robier. 2017. Red blood cell morphology in patients with β -thalassemia minor. *LaboratoriumsMedizin*, 41(1): 49-52.
- Lama, R., W. Yusof, T.R. Shrestha, S. Hanafi, M. Bhattarai, R. Hassan, & B.A. Zilfalil. 2021. Prevalence and distribution of major β -thalassemia mutations and HbE/ β -thalassemia variant in Nepalese ethnic groups. *Hematology/Oncology and Stem Cell Therapy*, 1-7.
- Lie-Injo, LE., S.P. Cai, I. Wahidijat, S. Moeslichan, M.L. Lim, L. Evangelista, M. Doherty, & Y.W. Kan. 1989. Beta-thalassemia mutations in Indonesia and their linkage to beta haplotypes. *American Journal of Human Genetics*, 45(6): 971-975.
- Lucena-Aguilar, G., A.M. Sánchez-López, C. Barberán-Aceituno, J.A. Carrillo-Ávila, J.A. López-Guerrero, & R. Aguilar-Quesada. 2016. DNA source selection for downstream applications based on DNA quality indicators analysis. *Biopreservation and biobanking*, 14(4): 264–270.
- Lutz, I., J. Miranda, P. Santana, T. Martins, C. Ferreira, I. Sampaio, M. Vallinoto, & G.E. Gomes. 2023. Quality analysis of genomic DNA and authentication of fisheries products based on distinct methods of DNA extraction. *PloS one*, 18(2): e0282369.
- Manning, L.R., J.E. Russell, J.C. Padovan, B.T. Chait, A. Popowicz, R.S. Manning, & J.M. Manning. 2007. Human embryonic, fetal, and adult hemoglobins have different subunit interface strengths. Correlation with lifespan in the red cell. *Protein Science*, 16(8): 1641-1658.



- Marengo-Rowe, A.J. 2006. Structure-function relations of human hemologis. *Proceedings*. Baylor University. Medical Center, 19(3): 239-245.
- Mehta, A.B., & Hoffbrand, A.V. 2014. *Haematology at a glance* 4th ed. Wiley Blackwell. UK.
- Mohammad, S. 2021. *Physiology of blood*. 10.13140/RG.2.2.25618.99529.
- Muncie, H.L., & J.S. Campbell. 2009. Alpha and beta thalassemia. *American Family Physician*, 80 (4): 339-344.
- Nienhuis, A.W., & D.G. Nathan. 2012. Pathophysiology and clinical manifestations of the β-thalassemias. *Cold Spring Harbor perspectives in medicine*, 2(12): a011726.
- 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, 9. Springer, Totowa, NJ.
- Old, J.M, C. Hateveld, J. Traeger-Synodinos, M. Petrou, M. Angastiniotis, R. Galanello. 2012. *Pervention of thalassemias and other haemoglobin disorders: volume 1: laboratory protocols*. 2nd edition. Thalassaemia International Federation Publication.
- O'Neill, M., J. McPartlin, K. Arthure, S. Riedel, & N.D. McMillan. 2011. Comparison of the TLDA with the nanodrop and the reference qubit system. *Journal of Physics: Conference Series*, 307, 012047.
- Oringa, R. 2017. β-Thalassemia. *Genetics in Medicine*, 19(6): 609-619.
- Orkin, S.H., & H.H. Kazazian. 1984. The mutation and polymorphism of the human β-globin gene and its surrounding DNA. *Annual Reviews Genetics*, 18: 131-171.
- Peate, I. 2017. Anatomy and physiology, 3. the blood. *British Journal of Healthcare Assistants*, 11(10): 474-478.
- Pratama, B., & I. Kurniati. 2019. Pendekatan diagnosis berbasis molekuler pada pasien thalassemia. *Medula*, 9(2): 339-345.
- Putri, A.R.S. 2017. Penapisan Hemoglobin E pada Siswi SMA Negeri Kecamatan Singosari di Kabupaten Malang, Jawa Timur. *Skripsi*. Fakultas Kedokteran dan Ilmu Kesehatan. Universitas Islam Negeri Syarif Hidayatullah. Jakarta.



- Polat, C., N. Mete, & M. Soker. 2021. Mutation analysis of beta-thalassemia major patients and their parents in Diyarbakir Province, Turkey. *Dicle Medical Journal*, 48(1): 47-54.
- Rivella S. (2009). Ineffective erythropoiesis and thalassemias. *Current opinion in hematology*, 16(3): 187–194.
- Rujito, L., M. Basalamah, S. Mulatsih, & A.S.M Sofro. 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.
- Rujito, L. 2019. *Talasemia: genetik dasar dan pengelolaan terkini*. UNSOED Press. Purwokerto.
- Saleh-Gohari, N., & M. Bazrafshani. 2010. Distribution of β -Globin Gene Mutations in Thalassemia Minor Population of Kerman Province, Iran. *Iranian journal of public health*, 39(2), 69–76.
- Silverthorn, D.U. 2019. *Human physiology* 8th ed. Pearson. USA.
- Sinha, S., M.L. Black, S. Agarwal, R. Colah, R. Das, K. Ryan, M. Bellgard, & A.H. Bittles. 2009. Profiling β -thalassaemia mutations in India at state and regional levels: implications for genetic education, screening and counselling programmes. *The HUGO journal*, 3(1-4), 51–62.
- Shawkat, AJ., & A.H. Jwalid. 2019. Clinical complications of beta-thalassemia major. *Iraqi Journal of Pharmaceutical Sciences*, 28(1): 1-8.
- Sofro, A.S.M., J.B. Clegg, F. Lanni, O, Sianipar, Himawan, & R.V. Liliani. 1996. Application of ARMS primers for the molecular characterization of β -thalassemia carrier in Palembang, South Sumatera. *Indonesia Journal of Biotechnology*, (December): 59-65.
- Susanto, ZA., W. Siswandari, & L. Rujito. 2020a. 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: 1-3.
- Susanto, ZA., W. Siswandari, & L. Rujito. 2020b. Korelasi genotip-fenotip pasien talasemia beta di Kota Samarinda Kalimantan Timur tahun 2019. *Buletin Penelitian Kesehatan*, 48(2): 91-98.



- Tabassum, S., M. Khakwan, A. Fayyaz, & N. Taj. 2022. Role of Mentzer index for differentiating iron deficiency anemia and beta thalassemia trait in pregnant women. *Pakistan Journal of Medical Sciences*, 38(4Part-II): 878–882.
- Taeger-Synodinos, J., C.L. Harteveld, J.M. Old, M. Petrou, R. Galanello, P. Giordano, M. Angastioniotis, B.D.I. Salle, S. Henderson, & A. May. 2015. EMQN best practice guidelines for molecular and haematology methods for carrier identification and prenatal diagnosis of the haemoglobinopathies. *European Journal of Human Genetics*, 23: 426-437.
- 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.
- Teh, L.K., G. Elizabeth, M.I. Lai, J.A.M.A. Tan, L. Wong & P. Ismail. 2013. Molecular basis of transfusion dependent beta-thalassemia major patients in Sabah. *Journal of Human Genetics*, 59: 119-123.
- Thein, S.L. 2013. The molecular basis of β -thalassemia. *Cold Spring Harbor perspectives in medicine*, 3(5): a011700.
- Thein S. L. 2018. Molecular basis of β thalassemia and potential therapeutic targets. *Blood Cells, Molecules & Diseases*, 70: 54–65
- Treisman, R., N.J. Proudfoot, M. Shander, & T. Maniatis. 1982. A single-base change at a splice site in a β 0-thalassemic gene causes abnormal RNA splicing. *Cell*, 29(3): 903–911.
- Ware, D. A. 2020. The complete blood count and white blood cell differential. In: W. Clarke and M. A. Marzinke (eds) *Contemporary Practice in Clinical Chemistry*. 4th Edition. Academic Press. UK. 429–433.
- Wienert, B., G.E. Martyn, A.P.W. Funnell, K.G.R. Quinlan, & M. Crossley. 2018. Wake-up sleepy gene: reactivating fetal globin for β -hemoglobinopathies. *Review Special Issue: Gene Expression in Time and Space*, 34(12): 927-940.
- Zivot, A., J.M. Lipton, A. Narla, & L. Blanc, 2018. Erythropoiesis: insights into pathophysiology and treatments in 2017. *Molecular Medicine*, 24(11): 1-15.