

## DAFTAR PUSTAKA

- Adewoyin, A., dan Nwogoh, B. 2014. Peripheral blood Film- A REVIEW. *Annals of Ibadan Postgraduate Medicine*. 12(2):71–79.
- Alanazi, M., Abduljaleel, Z., Khan, W., Warsy, A. S., Elrobh, M., Khan, Z., Bazzi, M. D. (2011). *In Silico* Analysis of Single Nucleotide Polymorphism (SNPs) in Human  $\beta$ -Globin Gene. *PLoS ONE*. 6(10): e25876.
- Ansari, M, I., dan Patel, N, G. 2015. Characterization of  $\beta$ -Thalassemia Mutations from North Maharashtra Region. *IOSR Journal of Pharmacy and Biological Sciences*. 10(3): 13-16.
- Ariani, Y., Soeharso, P., dan Sjarif, D, R. 2017. Genetics and genomic medicine in Indonesia. *Molecular Genetics & Genomic Medicine*. 103-109. Wiley Periodicals, Inc.
- Aycicek, A., Koç, A., Özdemir, Z, C., Bilinç, H., Koçyiğit, A., dan Dilmeç, F. 2011.  $\beta$ -globin gene mutations in children with  $\beta$ -thalassemia major from Şanlıurfa province, Turkey. *Turkish Journal of Hematology*. 28: 264-268.
- Buckingham, L dan Flaws, M. 2007. *Molecular Diagnostic: Fundamentals, Methods, & Clinical Applications*. Philadelphia: F.A Davis Company.
- Batt, K., dan Reske, T. 2010. Hemoglobinopathies. *Hospital Physician Hematology Board Review Manual*. 5(3): 1-12.
- Baig, S, M., Rabbi, F., Hameed, U., Qureshi, J, A., Mahmood, Z., Bokhari, S, H., Kiani, A., Hassan, A., Baig, J, M., Azhar, A., dan Zaman, T. 2005. Molecular characterization of mutations causing  $\beta$ -thalassemia in Faisalabad Pakistan using the amplification Refractory Mutation System (ARMS-PCR). *Indian Journal of Human Genetics*. 11 (2): 80-83.
- Calvino, N. 2003. Connective tissue:Vascular and Hematological (blood) Support. *Journal of Chiropractic Medicine*. 2(1):25-36
- Cao, A., dan Galanello, R. 2010.  $\beta$ -Thalassemia. *Genetics in Medicine*. 12(2): 61-76.
- Cao, A., dan Kan, Y, W. 2013. The prevention of Thalassemia. *Cold Spring Harbor Perspectives in Medicine*. 1-16.
- Carlice-dos-Reis, T., Viana, J., Moreira, F, C., Cardoso, G, L., Guerreiro, J., Santos, S., dan Ribeiro-dos-Santos, A. 2017. Investigation of mutations in the HBB gene using the 1,000 genomes database.. *PLoS ONE* 12(4): 1-9.
- Chen, P., Lian, Q., Huang, T., Liu, T., dan Li, M. 2016. A Simple, Rapid, and Highly Sensitive Electrochemical DNA Sensor for the Detection of  $\alpha$ - dan  $\beta$ - Thalassemia in China. *Journal of Clinical Laboratory Analysis*. 00: 1-8.

- Chin, J. Y., Kuan, J. Y., Lonkar, P. S., Krause, D. S., Seidman, M. M., Peterson, K. R., Nielsen, P. E., Kole, R., dan Glazer, P. M. 2008. Correction of a splice-site mutation in the  $\beta$ -globin gene stimulated by triplex-forming peptide nucleic acids. *Proceedings of the National Academy of Sciences*. 105(36): 13514–13519.
- Colah, R., Nadkarni, A., Gorakshakar, A., Phanasgaonkar, S., Surve, R., Subramaniam, P. G., Bondge, N., Pujari, K., Ghosh, K., dan Mohanty, D. 2004. Impact of  $\beta$ -globin gene mutations on the clinical phenotype of  $\beta$ -thalassemia in India. *Blood Cells Molecules and Diseases*. 33(2):153-157
- Das, S. K., dan Talukder, G. 2002.  $\beta$ -Globin Gene and Related Diseases: A review. *International Journal of Human Genetics*. 2 (3): 139-152.
- Das, R., dan Sharma, P. 2016. *Molecular Genetics of Thalassemia Syndromes*. Morgan & Claypool Life Science.
- Danckwardt, S., Neu-Yilik, G., Thermann, R., Frede, U., Hentze, M. W. Dan Kulozik, A. E. 2002. Abnormally spliced  $\beta$ -globin mRNAs: a single point mutation henerate transcripts sensitive and insensitive to nonsense-mediated mRNA decay. *Blood*. 99 (5): 1811-1816.
- Dell'edera, D., Pacella, E., Epifania, A. A., Benedetto, M., Tinelli, A., Mazzone, E., Laterza, F., dan Malvasi, A. 2011. Importance of molecular biology in the characterization of  $\beta$ -thalassemia carriers. *European Review for Medical and Pharmacological Sciences*. 15: 79-86.
- Diez-Silva, M., Dao, M., Han, J., Lim, C., dan Suresh, A. 2010. Shape and Biomechanical Characteristics of Human Red Blood Cells in Health and Disease. *MRS Bulletin*. 35(5): 382–388.
- Ford, J. 2013. Review: Red Blood Cell Morphology. *International Journal of Laboratory Hematology*. 35, 351–357
- Forget, B. G. dan Bunn, F. 2013. Classification of the Disorders of Hemoglobin. *Cold Spring Harbour Perspective in Medicine*. 3: a011684.
- Fox, S. I. 2011. *Human Physiology*. 8th edition. Boston: McGraw-Hill Companies. Hal: 403-412.
- Fucharoen, S. dan Winichagoon P. 1992. Thalassemia in SouthEast Asia: problems and strategy for prevention and control. *Southeast Asian Journal of Tropical Medicine and Public Health*. 23(4):647-55.
- Fucharoen, S. dan Winichagoon, P. 2011. Haemoglobinopathies in Southeast Asia. *Indian Journal of Medical Research*. 134(4): 498–506.

- Galanello, R. dan Origa, R. 2010.  $\beta$ -Thalassemia. *Orphanet Journal of Rare Diseases*. 5:11
- Galanello, R., dan Cao, A. 2011. Alpha-thalassemia. *Genetics in Medicine*. 13: 83–88.
- Ghifari, A, R. 2017. Deteksi mutasi IVS I-2 T>C Gen Penyandi  $\beta$ -globin pada  $\beta$ -Thalassemia Carrier. Skripsi. Yogyakarta. Universitas Gadjah Mada.
- Gen, M., dan Ahmed, S. 2014. Amplification Refractory Mutation System (ARMS). Diakses di <http://grcpk.com/wp-content/uploads/2014/10/7.-ARMS.pdf>. pada tanggal 26 Oktober, 2017
- George-Gay, B., dan Parker, K. 2003. Understanding the Complete Blood Count With Differential. *Journal of PeriAnesthesiaNursing*. 18(2): 96-117.
- Gupta, A., Hattori, Y., Gupta, U, R., Nigham, N., Singhal, P., dan Agarwal, S. 2003. Molecular Genetic Testing of  $\beta$ -Thalassemia Patients of Indian Origin and a Novel 8-bp Deletion Mutation at Codons 36/37/38/39. *Genetic Testing*, 7(2): 163-168.
- Greer, J, P., Foerster, J., Rodgers, G, M., Paraskevas, F., Glader, B., Arber, D, A., dan Means, R, T. 2009. *Wintrobe's Clinical Hematology* 12<sup>th</sup> Edition. Philadelphia: Lippincott Williams & Wilkins.
- Hajihoseini, S., Motovali-Bashi, M., Honardoost, M, M., dan Alerasool, N. 2015. Tetra-Primer ARMS PCR Optimization for Detection of IVS-II-I (G>A) and FSC 8/9 InsG Mutation in  $\beta$ -Thalassemia Major Patients inj Isfahan Population. *Iranian Journal of Public Health*. 44(3): 380-387.
- Hanafi, S., Hassan, R., Bahar, R., Abdullah, W,Z., Noor, M, F, J., Rashid, D., Azman, N, F., Nasir, A., Ahmad, R., Othman, A., Ibrahim, M, I., Sukeri, S., Sulong, S., Yusoff, S., Mohammad, N, S., Hussein, S., Hassan, R., Yusoff, N., Yahaya, B, H., Ismail, E., Yussof, N, K, N., Salleh, S dan Zifail, B, A. 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., Husna, N. dan Sanka, I. 2017.  $\alpha$ -globin alteration in  $\alpha$ -thalassemia disorder: prediction and interaction defect. *Pakistan Journal of Biological Sciences*. 20 (7): 343-349.
- Handayani, N.S.N. dan Purwanto, R. 2015. Cd35 (Del C) Frameshift Mutation in Exon 2 of  $\beta$ -Globin Gene on  $\beta$ -Thalassemia Carriers. *Biomedical Engineering*. 1(1): 19-23.

- Harteveld, C., Voskamp, A., Phylipsen, M., Akkermans, N., den Dunnen, J. T., White, S., dan Giordano, P. (2005). Nine unknown rearrangements in 16p13.3 and 11p15.4 causing  $\alpha$ - and  $\beta$ -thalassaemia characterised by high resolution multiplex ligation-dependent probe amplification. *Journal of Medical Genetics*. 42(12): 922–931.
- Hidayati, N, I. 2017. Deteksi Mutasi IVSI-5 (G>C) Dan Cd 35 (Del C) Gen Pengkode  $\beta$ -Globin Pada Carrier  $\beta$ -Thalassemia dengan High-Resolution Melting Analysis. Tesis. Yogyakarta: Universitas Gadjah Mada.
- Hernanda, P.Y.,Tursilowati, L., Arkesteijn, S.G.J., Ugrasena, I.D.G., Larasati, M.C.S., Soeatmadji, S.M., Giordano,P.C., dan Harteveld, C.L. 2012. Towards a Prevention Program for  $\beta$ -Thalassemia: The Molecular Spectrum in East Java, Indonesia. *Hemoglobin*. 36(1):1–6.
- Hoffbrand, A. V., Moss, P. A. H. & Pettit, J. E. 2006. *Essential Haematology*. 5th edition. Oxford: Blackwell Publishing Ltd.
- Huang, S., Eng, F., Ren, Z., Lu, Z., Rodgers, G, P., Schechter, A, N., dan Zeng, Y. 1994. RNA transcripts of the  $\beta$ -thalassaemia allele IVS-2-654 C >T: a small amount of normally processed P-globin mRNA is still produced from the mutant gene. *British Jourrial of Haematology*. 88: 541-546
- Hug, N., Longman, D., dan Caceres, J, F. 2016. Survey and Summary: Mechanism and Regulation of the non-sense mediated decay pathway. *Nucleic Acids Research*. 44(4): 1483-1495.
- Jagannathan-Bogdan, M dan Zon, L, I. 2013. Hematopoiesis. *Development*. 140: 2463-2467.
- John, G, S, M., dan Takeuchi, S. 2011. Understanding Tools and Techniques in Protein Structure Prediction. *Systems and Computational Biology - Bioinformatics and Computational Modeling*. INTECH Open Access Publisher. 185-212.
- Kakavas V, K., , Plageras, P., Vlachos, T, A., Papaioannou, A., dan Noulas. V. A., 2008. PCR-SSCP: a method for the molecular analysis of genetic diseases. *Molecular Biotechnology*. 38(2):155-63.
- Kawahara, R. 2007. Hematopoiesis. In xPharm: The Comprehensive Pharmacology Reference. *Biomedical Science*. Elsevier. 1-5.
- Kazazian, H, H, Jr. dan Boehm, C, D.1988. Molecular basis and prenatal diagnosis of  $\beta$ -thalassemia. *Blood*. 72 (4): 1107-16.
- Khare, P., Raj, V., Chandra, S., dan Agarwal, S. 2014. Quantitative and qualitative assessment of DNA extracted from saliva for its use in forensic identification. *Journal of Forensic Dental Sciences*. 6(2): 81–85.

- Kotila, T, R. 2010. Guidelines for the diagnosis of Haemoglobins in Nigeria. *Annals of Ibadan Postgraduate Medicine*. 8 (1): 25-29.
- Kotila, T, R. 2012. Thalassaemia is a Tropical Disease. *Annals of Ibadan Postgraduate Medicine*. 10 (2): 11-15.
- Kohne, E. 2011. Hemoglobinopathies: clinical manifestations, diagnosis, and treatment. *Deutsches Ärzteblatt International*. 108(31–32): 532–40.
- Kulshreshtha, S., Chaudhary, V., Goswami, G.K., dan Mathur, N. 2016. Computational approaches for predicting mutant protein stability. *Journal of Computer-Aided Molecular Design*. 30: 401.
- Kumar, A. dan Garg, N. 2005. *Genetic Engineering*. New York: Nova Biomedical Books.
- Lahiry, P., Al-Attar, S, A., dan Hegele, R, A. 2008. Understanding  $\beta$ -Thalassaemia with Focus on the Indian Subcontinent and the Middle East. *The Open Hematology Journal*, 2: 5-13
- Lee, Y., Park, S, S., Kim, J., dan Cho, H. 2002. RFLP Haplotypes of  $\beta$ -Globin Gene Complex of  $\beta$ -Thalassaemic Chromosomes in Koreans. *Journal of Korean Medical Science*. 17: 475-478.
- Lie-Injo, L. E., Cai, S. P., Wahidijat, I., Moeslichan, S., Lim, M. L., Evangelista, L., Doherty, M. & Kan, Y. W. 1989.  $\beta$ -Thalassaemia Mutations in Indonesia and Their Linkage to  $\beta$ -Haplotypes. *American Journal of Human Genetics*. 45: 971-975.
- Little S. 2001. Amplification-refractory mutation system (ARMS) analysis of point mutations. *Curr Protoc Hum Genet*. Chapter 9:Unit 9.8.
- Mishra, K., dan Tiwari, A. 2014. Screening and Molecular Characterization of  $\beta$ -Thalassaemia Mutations in Parents and Siblings of  $\beta$ -Thalassaemia Major Patients. *Indian Journal of Basic and Applied Medical Research*. 3(2): 481-486.
- Modell, B., dan Darlison, M. 2007. Epidemiological Estimates for Haemoglobin Disorders: WHO South East Asian Region by Country. World Health Organization.
- Murray, D., Doran, P., MacMathuna, P., dan Moss, A, C. 2007. In silico gene expression analysis – an overview. *Molecular Cancer*. 6(50): 1-10.
- Musallam, K, M., Taher, A, T., dan Rachmilewitz, E, A. 2012.  $\beta$ -Thalassaemia Intermedia: A Clinical Perspective. *Cold Spring Harbor Perspectives in Medicine*. 2(7): 1-15.

- Najmabadi, H., Teimourian, S., Khatibi, T., Neishabury, M., Pourfarzad, F., Jalil-Nejad, S., Azad, M., Oberkanins, C., dan Krugluger, W. 2001. Amplification Refractory Mutation System (Arms) And Reverse Hybridization In The Detection Of B-Thalassemia Mutations. *Archives of Iranian Medicine*. 4 (4): 165-170.
- Newton, C, R., Graham, A., dan Heptinstall, L, E. 1989 . Analysis of any point mutation in DNA. The amplification refractory mutation system (ARMS). *Nucleic Acids Research*. 17:2503-2516.
- Nussbaum, R, L., McInnes, R, R., dan Willard, H, F. 2007. *Thompson & Thompson: Genetic in Medicine*. Philadelphia: Saunders Elsevier.
- 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., Hartevelde, C., Traeger-Synodinos, J., Petrou, M., Angastiniotis, M., Galanello, R. 2012. *Prevention Of Thalassaemias And Other Haemoglobin Disorders : Volume 1: Laboratory Protocols*. 2<sup>nd</sup> edition. Thalassaemia International Federation Publication. Hal 78-89.
- Old, J., Angastiniotis, M., Eleftheriou, A., Galanello, R., Hartevelde, C, L., Petrou, M., dan Traeger-Synodinos, J. 2013. *Prevention Of Thalassaemias And Other Haemoglobin Disorders : Volume 1: Principles*. 2<sup>nd</sup> edition. Thalassaemia International Federation Publication. Hal: 1.
- Olson, N, D dan Morrow, J, B. 2012. DNA extract characterization process for microbial detection methods development and validation. *BMC Research Notes*. 5:668.
- Origa, R dan Moi, P. 2016. Alpha-Thalassemia. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Diakses di <https://www.ncbi.nlm.nih.gov/books/NBK1435> pada Tanggal 8 November 2017.
- Peixeiro, I., Silva, A. L. dan Romao, L. 2011. Control of Human  $\beta$ -Globin mRNA Stability and Its Impact on Beta-Thalassemia Phenotype. *Haematologica*. 96(6): 905-913.
- Provan, D., Singer, C, R, J., Baglin, T., dan Lilleyman, J. 2004. *Oxford Handbook of Clinical Hematology*. 2<sup>nd</sup> edition. Oxford University Press.
- Rachmilewitz, E, A., dan Giardina, P, J. 2011. How I treat thalassemia. *Blood*. 118(13) : 3479-3488.

- Reece, J. B., Urry, L. A., Cain, M. L., Wasserman, S. A., Minorsky, P. V., dan Jackson, R. B. 2011. *Campbell Biology*. 9<sup>th</sup> edition. San Fransisco: Pearson Benjamin Cummings. Hal:70
- Rhoades, R. A., dan Bell, D. R. 2013. *Medical Physiology Principles for Clinical Medicine Fourth Edition*. China: Lippincott Williams & Wilkins, a Wolters Kluwer business.
- Rogers, K. 2011. *The Human Body: Blood Physiology and Circulation*. Newyork: Britanica Educational Publishing.
- Rujito, L., Basalamah, M., Mulatsih, S. & Sofro, A. S. M. 2015. Molecular Scanning of  $\beta$ -Thalassemia in the Southern Region of Central Java, Indonesia; a Step Towards a Local Prevention Program. *Hemoglobin*. 39(5):330-33
- Santella, R. M. 2006. Approaches to DNA/RNA Extraxtion and Whole Genome Amplification. *Cancer Epidemiology, Biomarkers & Prevention*. 15(9): 1585 – 1587.
- Saste, S. R., Ghalsasi, P. M., Kataria, R. S., Josh, B. K., Mishra, B. P., dan Nimbkar, C. 2012. ARMS-PCR as an alternative, cost effective method for detection of *FecB* in Sheep. *Indian Journal of Biotechnology*. 11: 274-279.
- Schechter, A. N. 2008. Hemoglobin research and the origins of molecular medicine. *Blood*. 112(10): 3927-3938.
- Setianingsih, I., Alida, H., dan Nainggolan, I. M. 2003. Alpha Thalassaemia in Indonesia: Phenotypes and Molecular Defects. *Advances in Experimental Medicine and Biology*. 531: 47-56.
- Setianingsih, I., Williamson, R., Marzuk, S., Harahap, A., Tamam, M., dan Forrest, S. 1998. Molecular Basis of  $\beta$ -Thalassemia in Indonesia: Application to Prenatal Diagnosis. *Molecular Diagnosis*. 3(1): 11-20.
- Shakeel, M., Arif, M., Rehman, S. U., dan Yaseen, T. 2016. Investigation of molecular heterogeneity of  $\beta$ -thalassemia disorder in District Charsadda of Pakistan. *Pakistan Journal of Medical Sciences*. 32(2): 491-494.
- Sirichotiyakul, S., Wanapirak, C., Saetung, R., dan Sanguansermisri, T. 2010. High resolution DNA melting analysis: an application for prenatal control of  $\alpha$ -thalassemia. *Prenatal Diagnosis*. 30: 348–351.
- Sonati, M. F., dan Costa, F. F. 2008. The genetics of blood disorders: hereditary hemoglobinopathies. *Jornal de Pediatria*. 84(4): 40-51.

- Stamatoyannopoulos, G. 2005. Control of globin gene expression during development and erythroid differentiation. *Experimental Hematology*. 33(3): 259.
- Surapon, T. 2011. Thalassemia Syndrome, Advances in the Study of Genetic Disorders, Dr. Kenji Ikehara (Ed.), *InTech*, DOI: 10.5772/18051.
- Suraya, N. A. 2015. Analisis Sekuen IVS II Gen B-Globin pada Carrier  $\beta$ -Thalassemia. Tesis. Yogyakarta: Universitas Gadjah Mada.
- Tamam, M., Hadisaputro, S., Sutaryo, Setianingsih, I., Astuti, R. & Soemantri, A. 2010. Hubungan antara Tipe Mutasi Gen Globin dan Manifestasi Klinis Penderita Talasemia. *Jurnal Kedokteran Brawijaya*. 26: 48-52.
- Thein, S. L. 2005. Patophysiology of  $\beta$ -Thalassemia – A Guide to Molecular Therapies. *Hematology American Society of Hematology Educational Program*. 1: 31-37.
- Thein, S. L. 2013. The Molecular Basis of  $\beta$ -Thalassemia. *Cold Spring Harbor Perspectives in Medicine*. 3:a011700.
- Theml, H., Diem, H., dan Haferlach, T. 2004. Color Atlas of Hematology: Practical Microscopic and Clinical Diagnosis. Germany: Georg Thieme Verlag. Hal: 12-13.
- Thom, C, S., Dickson, C, F., Gell, D, A., dan Weiss, M, J. 2013. Hemoglobin Variants: Biochemical Properties and Clinical Correlates. *Cold Spring Harbor Perspectives in Medicine*. 3:a011858.
- Treisman, R., Orkin, S. H., dan Maniatis, T. 1983. Specific Transcription and RNA Splicing Defects in Five Cloned  $\beta$ -Thalassaemia Genes. *Nature*. 302: 591596.
- Tsiftoglou, A, S., Vizirianakis, I, S, dan Strouboulis, J. 2009. Critical Review Erythropoiesis: Model Systems, Molecular Regulators, and Developmental Programs. *Life*. 61(8): 800–830.
- Vander, A, J., Sherman, J., dan Luciano, D, S. 2001. *Human Physiology: The Mechanism of Body Function*. 8<sup>th</sup> edition. The McGraw-Hill Companies.
- Weatherall, D, J., dan Clegg, J, B. 2001. Inherited Haemoglobin Disorders: an Increasing Global Health Problem. *Bulletin of the World Health Organization*. 79(8): 704-712.
- Wen, W. Molecular Diagnosis and Mutation Characterization in Thalassemia. Thesis. Singapura. National University of Singapore.
- Wick, M., Pinggera., W., dan Lehmann, P. 2013. *Iron Metabolism: Diagnosis and Therapy of Anemias*. Springer Science & Business Media.

- Widmaier, E, P., Raff, H., dan Strang, K, T. 2006. *Human Physiology: The Mechanism of Body Function*. 11<sup>th</sup> edition. The McGraw-Hill Companies
- Widyawati, L. 2017. Deteksi mutasi IVS I-5 G>C Gen Penyandi  $\beta$ -globin pada  $\beta$ -Thalassemia Carrier. Skripsi. Yogyakarta. Universitas Gadjah Mada.
- Yang, L., Ijaz, I., Cheng, J., Wei, C., Tan, X., Khan, M. A., Fu, X., dan Fu, J. 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.
- Yokoyama, A., Nakamaki, T., Yamada, K., Koike, M., Tomoyasu, S., Hirayama, N., Tsuruoka, N., dan Harano, T. 1993.  $\beta^0$ -Thalassemia Trait (IVS-I-1 G>T) in Japanese Family. *Internal Medicine*. 32: 865-868.