

DAFTAR PUSTAKA

- Arika, W.M., Nyamai, D.W., Musila, M.N., Ngugi, M.P., & Njagi, E.N.M. 2016. Hematological Markers of *In Vivo* Toxicity. *Journal of Hematology and Thromboembolic Diseases*, 4 (2) : 1-7.
- Bachir, D. & Galacteros, F. 2004. *Hemoglobin E Disease*. Orphanet Encyclopedia. Creteil, pp. 4.
- Beck, N. 2009. *Diagnostic Hematology*. Springer Science. London, pp. 213.
- Birchard, G.F. 1997. Optimal Hematocrit : Theory, Regulation, and Implications. *American Zoology*, 37 : 65-72.
- Cao, A. & Galanello, R. 2010. Beta-thalassemia. *Genetics in Medicine*, 12 (2) : 61-76.
- Cao, A. & Moi, P. 2000. Genetic Modifying Factors in B-Thalassemia. *Clinical Chemistry and Laboratory Medicine*, 38 (2) : 123-132.
- Chakrabarti, I., Sinha, S.W., Ghosh, N., & Goswami, B.K. 2012. Beta-Thalassemia Carrier Detection by NESTROFT : An Answer in Rural Scenario. *Iranian Journal of Pathology*, 7 (1) : 19-26.
- Chang, Y.C., Chang, Y., Chang, C.C., Liu, T.C., Ko, Y.C., Lee, C.C., Chang, S.J., & Chang, J.G. 2016. Development of a High-Resolution Melting Method for the Screening of *TNFAIP3* Gene Mutations. *Oncology Report*, 35 : 2936-2942.
- Chassanidis, C., Boutou, E., Voskaridou, E., & Balassopoulou, A. 2016. Development of a High-Resolution Melting Approach for Scanning Beta Globin Gene Point Mutations in the Greek and Other Mediterranean Populations. *PLOS ONE*, 11 (6) : 1-22.
- Chui, D.H. 2005. Alpha-Thalassemia : HbH Disease and Hb Barts Hydrops Fetalis. *Annals of the New York Academy of Sciences*, 10 (54) : 25-32.
- Clarke, G.M. & Higgins, T.N. 2000. Laboratory Investigation of Hemoglobinopathies and Thalassemias : Review and Update. *Clinical Chemistry*, 46 : 8.
- Daar, S. & Gravell, D. 2006. Diagnosis of Beta-Thalassaemia Carriers in the Sultanate of Oman. *Sultan Qaboos University Medical Journal*, 6 (1) : 1-5.
- Deo, M.G. & Pawar, P.V. 2014. Alpha Thalassaemia in Tribal Communities of Coastal Maharashtra India. *Indian Journal of Medical Research*, 140 (2) : 231-237.
- Desjardins, P. & Conklin, D. 2010. NanoDrop Microvolume Quantitation of Nucleic Acids. *Journal of Visualized Experiments*, 45 : 2565.
- Devereux, R.D. & Wilkinson, S.S. 2004. *Amplification of Ribosomal RNA Sequences*, pp. 509-522. In Akkermans, A.D.L. (Eds.). *Molecular Microbial Ecology Manual*. Dordrecht : Kluwer Academic Publishers.
- Eleftheriou, A. 2007. *About Thalassemia*. Thalassemia International Federation. Nicosia, pp. 17.
- Fahim, F. & Ahmed, M.G.S. 2010. Genetic Haemoglobin Disorders. *Haematology Updates*, pp. 16 & 21-22.
- Fitarelli-Kiehl, M., Macedo, G. S., Schlatter, R. P., Koehler-santos, P., Matte, S., Ashton-prolla, P. & Giacomazzi, J. 2016. Comparison of Multiple Genotyping Methods for the Identification of the Cancer Predisposing Founder Mutation p.R337H in TP53. *Genetic and Molecular Biology*. 39 (2) : 203-209.
- Forget, B.G & Hardison, R.C. 2010. *The Normal Structure and Regulation of Human Globin Gene Clusters*. Cambridge University Press. Cambridge, pp. 46-48.
- Fucharoen, S. & Weatherall, D.J. 2010. *Hemoglobin E Disorders*. Cambridge University Press. Cambridge, pp. 417-420.



- Fucharoen, S. & Winichagoon, P. 2002. Thalassemia and Abnormal Hemoglobin. *International Journal of Hematology*, 76 (2) : 83-89.
- Fucharoen, S. 2012. The Hemoglobin E Thalassemias. *Cold Spring Harbor Perspectives in Medicine*, 2 : 1-15.
- Galanello, R. & Origa, R. 2010. Beta-Thalassemia. *Orphanet Journal of Rare Diseases*, 5 (11) : 1-15.
- Guyton, A.C. & Hall, J.E. 2006. *Textbook of Medical Physiology*. 11th Edition. Elsevier Saunders. Philadelphia, pp. 419 & 424.
- Hanafi, S.B., Abdullah, W.Z., Adnan, R.A., Bahar, R., Johan, M.F., Azmam, N.F., Rashid, N.D., Ahmad, S.A.A., Hassan, R., & Zilfalil, B.A. 2016. Genotype-phenotype Association of HbE/ β -thalassemia Disease and The Role of Genetic Modifier. *Malaysian Journal of Pediatrics and Child Health*, 22 : 1-16.
- Harahap, A., Megawati, D., Nainggolan, I.M., Swastika, M., & Setianingsih, I. 2015. The Role of Complete Blood Count in the Diagnosis of Hemoglobin E in Pseudo High Level of Hemoglobin A₂. *Open Journal of Hematology*, 1-6.
- Hidayati, N.I. 2017. Deteksi Mutasi IVSI-5 (G→A) dan Cd 35 (Del C) Gen Pengkode β -Globin pada Carrier β -Thalassemia dengan *High-Resolution Melting Analysis*. Tesis. Program Pascasarjana Fakultas Biologi Universitas Gadjah Mada Yogyakarta, hal. 45.
- Higgs, D.R. & Weatherall, D.J. 2009. The Alpha Thalassemias. *Cellular and Molecular Life Sciences*, 66 (7) : 1154-1162.
- Hoffbrand, A.V. & Moss, P.A.H. 2011. *Essential Haematology*. 6th Edition. Wiley Blackwell Publishing. London, pp. 17-23 & 87-91.
- Jones, K.W. 2009. *Evaluation of Cell Morphology and Introduction to Platelet and White Blood Cell Morphology*, pp. 93-112. In Harmening, D.M. (Eds.). *Clinical Hematology and Fundamentals of Homeostasis*. 5th Edition. Philadelphia: F. A. Davis Company.
- Khosravinia, H., Murthy, H.N.N., Parasad, D.T., & Pirany, N. 2007. Optimizing Factors Influencing DNA Extraction from Fresh Whole Avian Blood. *African Journal of Biotechnology*, 6 (4) : 481-486.
- Lin, M., Jia, J.W., Zhan, X.H., Zhan, X.F., Pan, M.C., Wang, J.L., Wang, C.F., Zhong, T.Y., Zhang, Q., Yu, X., Wu, J.R., Yang, H.T., Lin, F., Tong, X., & Yang, L.Y. 2014. High Resolution Melting Analysis: A Rapid Screening and Typing Tool for Common β -Thalassemia Mutation in Chinese Population. *PLOS ONE*, 9 (8) : 1-7.
- Luokopoulos, D.L. 2002. *Haemoglobinopathies Encyclopedia of Life Science*. Macmillan Publisher. New York, pp. 1-8.
- Marashi, S. J., Eshkoo, S. A., Mirinargesi, M., Sarookhani, M. R., Rahmat, A., & Ismail, P. 2012. Detection of Eight Common β -globin Gene Mutation in Thalassemia Major Patients using Real Time Polymerase Chain Reaction (PCR) - High Resolution Melting and EvaGreen™ Dye. *African Journal of Biotechnology*, 11 (2) : 448-459.
- Marsella, M., Salvagno, G., Dolcini, B., Ferlini, A., Ravani, A., & Hartevel, C.L. 2014. Characterization of Hb Calvino : A New Silent β -globin Gene Variant Found in Coexistence with A-Thalassemia in a Family of African Origin. *Hemoglobin*, 38 (5) : 369-372.
- Meijer, M. & Meskhat, M. 2010. The Effectiveness of Lentiviral Correction of Thalassemia Major and The Possibility of Adverse Effects. *Erasmus Journal of Medicine*, 1 (1) : 46-50.
- Moiz, B., Hashmi, M.R., Nasir, A., Rashid, A., & Moatter, T. 2012. Hemoglobin E Syndromes in Pakistani Population. *BMC Blood Disorders*, 12 (3): 1-6.



- Muncie, H.L.Jr. & Campbell, J. 2009. Alpha and Beta Thalassemia. *American Family Physician Journal*, 80 (4) : 339-44.
- Passarge, E. 2001. *Color Atlas of Genetics*. 2nd Edition. Thieme Stuttgart Publishing. New York, pp. 336-338.
- Perng, C-L., Chen, H-Y., Chiueh, T-S., Wang, W-Y., Huang, C-T. & Sun, J-R. 2012. Identification of Non-Tuberculous Mycobacteria by Real-Time PCR Coupled with a High-Resolution Melting System. *Journal of Medical Microbiology*, 61 : 944-951.
- Philipsen, S. & Wood, W.G. 2010. *The Molecular, Cellular, and Genetic Basis of Hemoglobin Disorders : Erythropoiesis*. Cambridge University Press. Cambridge, pp. 26-27.
- Piel, F.B. & Weatherall, D.J. 2014. The α -Thalassemias. *The New England Journal of Medicine*, 371 (20) : 1908-1916.
- Putri, C. 2018. Deteksi Mutasi Cd 26 (G→A) Gen Pengkode β -Globin pada Pembawa HbE dengan Metode *Tm-Shift-Real Time* PCR. *Skripsi (unpublished)*. Fakultas Biologi, Universitas Gadjah Mada, Yogyakarta.
- Rahimi, Z., Rezaei, M., Nagel, R.L., & Muniz, A. 2008. Molecular and Hematologic Analysis of Hemoglobin Q-Iran and Hemoglobin Aetif in Iranian Families. *Archives of Iranian Medicine*, 11 (4) : 382-386.
- Ramezanzadeh, M., Salehi, M., & Salehi, R. 2016. Assessment of High Resolution Melt Analysis Feasibility for Evaluation of Beta-Globin Gene Mutations as a Reproducible, Cost-Efficient and Fast Alternative to the Present Conventional Method. *Advanced Biomedical Research*, 5 : 71.
- Reed, G.H. & Wittwer, C.T. 2004. Sensitivity and Specificity of Single Nucleotide Polymorphism Scanning by High Resolution Melting Analysis. *Clinical Chemistry*, 50 (10) : 1748-1754.
- Reed, G.H., Kent, J.O., & Wittwer, C.T. 2007. High Resolution DNA Melting Analysis for Simple and Efficient Molecular Diagnostics. *Pharmacogenomics*, 8 (6) : 597-608.
- Rivella, S. 2009. Ineffective Erythropoiesis and Thalassemias. *Current Opinion in Hematology*, 16 (3) : 187-194.
- Rogers, K. 2011. *The Human Body : Blood Physiology and Circulation*. 1st Edition. Britannica Educational Publishing. New York, pp. 181-183.
- Rund, D. & Rachmilewitz, E. 2005. β -Thalassemia : Medical Progress. *The New England Journal of Medicine*, 353 (11) : 1135-1146.
- Ruskova, L. & Raclavsky, V. 2011. The Potential of High Resolution Melting Analysis (HRMA) to Streamline, Facilitate, and Enrich Routine Diagnostics in Medical Microbiology. *Biomedical Papers of the Medical Faculty of the University Palacky, Olomouc, Czechoslovakia*, 155 : 1-14.
- Sachdeva, A., Lokeshwar, M.R., Shah, N., Agarwal, B.R., Khanna, V.K., Yadav, S.P., & Jain, V. 2006. *Hemoglobinopathies*. Jaypee Brothers Medical Publisher. New Delhi, pp. 78.
- Sheerwood, L. 2010. *Human Physiology : From Cells to Systems*. 7th Edition. Brooks Cole Cengage Learning. Belmont, pp. 394-395.
- Solomon, E.P., Berg, L.R., & Martin, D.W. 2010. *Biology*. 9th Edition. Brooks Cole Cengage Learning. Belmont, pp. 975-976.
- Steinberg, M.H. & Nagel, R.L. 2010. *Hemoglobins of the Embryo, Fetus, and Adult*. Cambridge University Press. Cambridge, pp. 119 & 124.



- Steinberg, M.H. & Rodgers, G.P. 2015. HbA₂ : Biology, Clinical Relevance and a Possible Target for Ameliorating Sickle Cell Disease. *British Journal of Haematology*, 170: 781-787.
- Stephens, A. 2005. Haemoglobinopathies : A Way Forward in Reporting Results. *The Biomedical Scientist*, pp. 448.
- Surapon, T. 2011. *Thalassemia Syndrome, Advances in the Study of Genetic Disorders*. InTech China. Shanghai, pp. 101.
- Synodinos, J.T. & Hartevelde, C.L. 2014. Advances in Technologies for Screening and Diagnosis of Hemoglobinopathy. *Biomarkers in Medicine*, 8 (1) : 119-131.
- Taylor, C.F. 2009. Mutation Scanning Using High-Resolution Melting. *Biochemical Society Transactions*, 37 (0) : 433-437.
- Uddin, M.K., Aziz, M.A., Sardar, M.H., Hossain, M.Z., Bhuya, M.F., Uddin, M.M., Kobir, M.A., & Rahman, M.J. 2010. Electrophoretic Pattern of Hereditary Haemoglobin Disorders in Bangladesh. *Journal of Dhaka Medical College*, 19 (1) : 39-42.
- Virprakasit, V., Lee, C.L., Chong, Q.T., Lin, K.H., & Khuhapinant, A. 2009. Iron Chelation Therapy in the Management of Thalassemia : The Asian Perspective. *International Journal of Hematology*, 90 : 435-445.
- Vossen, R.H.A.M., Aten, E., Roos, A., & Dunnen, J.T. 2009. High-Resolution Melting Analysis (HRMA) - More Than Just Sequence Variant Screening. *Human Mutation*, 30 (0) : 1-7.
- Weatherall, D.J. & Clegg, J.B. 2001. Inherited Haemoglobin Disorders : An Increasing Global Health Problem. *Bulletin of the World Health Organization*, 79 (8) : 704-712.
- Wimbley, T.D.J. & Graham, D.Y. 2011. Diagnosis and Management of Iron Deficiency Anemia in the 21st Century. *Therapeutic Advances in Gastroenterology*, 4 (3) : 177-184.
- Wittwer, C.T. 2009. High Resolution DNA Melting Analysis : Advancements and Limitations. *Human Mutation*, 30 (6) : 857-859.
- Xiong, F., Huang, Q., Chen, X., Zhou, Y., Zhang, X., Cai, R., Chen, Y., Xie, J., Q., Zhang, T., Luo, S., Yang, X., Hao, Y., Qu, Y., Li, Q., & Xu, X. 2011. A Melting Curve Analysis-Based PCR Assay for One-Step Genotyping of β -Thalassemia Mutations : A Multicenter Validation. *The Journal of Molecular Diagnostics*, 13 (4): 427-436.
- Yu, J., Xie, J., Luo, L., & Li, Z. 2014. An Alu Element-Mediated 28.5kb α -Thalassemia Deletion Found in a Chinese Family. *Hemoglobin*, pp. 4.