

REFERENCES

- Al-Soud**, W.A. & Radstrom, P. (2000) Effects of amplification facilitators on diagnostic PCR in the presence of blood, feces, and meat. *J Clin Microbiol*, 38, 4463-70.
- Al-Soud**, W.A. & Radstrom, P. (2001) Purification and characterization of PCR-inhibitory components in blood cells. *J Clin Microbiol*, 39, 485-93.
- Bianco**, I., Cappabianca, M.P., Foglietta, E., Lerone, M., Deidda, G., Morlupi, L., Grisanti, P., Ponzini, D., Rinaldi, S. & Graziani, B. (1997) Silent thalassemias: genotypes and phenotypes. *Haematologica*, 82, 269-80.
- Bu**, Y., Huang, H. & Zhou, G. (2008) Direct polymerase chain reaction (PCR) from human whole blood and filter-paper-dried blood by using a PCR buffer with a higher pH. *Anal Biochem*, 375, 370-2.
- Burckhardt**, J. (1994) Amplification of DNA from whole blood. *PCR Methods Appl*, 3, 239-43.
- Cao**, A. & Galanello, R. (2010) Beta-thalassemia. *Genet Med*, 12, 61-76.

Castley, A., Higgins, M., Ivey, J., Mamotte, C., Sayer, D.C. & Christiansen, F.T. (2005)
Clinical applications of whole-blood PCR with real-time instrumentation. *Clin Chem*, 51,
2025-30.

Chan, A.Y., So, C.K. & Chan, L.C. (1996) Comparison of the HbH inclusion test and a
PCR test in routine screening for alpha thalassaemia in Hong Kong. *J Clin Pathol*, 49,
411-3.

Chang, J.G., Lee, L.S., Lin, C.P., Chen, P.H. & Chen, C.P. (1991) Rapid diagnosis of
 α -thalassemia-1 of southeast Asia type and hydrops fetalis by polymerase chain
reaction. *blood*, 78, 853-4.

Chapple, L., Harris, A., Phelan, L. & Bain, B.J. (2006) Reassessment of a simple
chemical method using DCIP for screening for haemoglobin E. *J Clin Pathol*, 59, 74-6.

Chong, S.S., Boehm, C.D., Higgs, D.R. & Cutting, G.R. (2000) Single-tube multiplex-
PCR screen for common deletional determinants of alpha-thalassemia. *Blood*, 95, 360-
2.

Clark, B.E. & Thein, S.L. (2004) Molecular diagnosis of haemoglobin disorders. *Clin
Lab Haematol*, 26, 159-76.

Clarke, G.M. & Higgins, T.N. (2000) Laboratory investigation of hemoglobinopathies and thalassemias: review and update. *Clin Chem*, 46, 1284-90.

Clegg, J.B. & Gagnon, J. (1981) Structure of the ζ chain of human embryonic hemoglobin. *Proc Natl Acad Sci U S A*, 78, 6076-80.

Cotton, F., Lin, C., Fontaine, B., Gulbis, B., Janssens, J. & Vertongen, F. (1999) Evaluation of a capillary electrophoresis method for routine determination of hemoglobins A₂ and F. *Clin Chem*, 45, 237-43.

Dieffenbach, C.W. & Dveksler, G.S. (1995) PCR primer : a laboratory manual. Cold Spring Harbor Laboratory Press, United States of America.

Edwards, M.C. & Gibbs, R.A. (1994) Multiplex PCR: advantages, development, and applications. *PCR Methods Appl*, 3, 65-75.

Elnifro, E.M., Ashshi, A.M., Cooper, R.J. & Klapper, P.E. (2000) Multiplex PCR: optimization and application in diagnostic virology. *Clin Microbiol Rev*, 13, 559-70.

Eng, B., Patterson, M., Borys, S., Chui, D.H. & Waye, J.S. (2000) PCR-based diagnosis of the Filipino ((FIL)) and Thai ((THAI)) alpha-thalassemia-1 deletions. *Am J Hematol*, 63, 54-6.

Flint, J., Harding, R.M., Boyce, A.J. & Clegg, J.B. (1998) The population genetics of the haemoglobinopathies. *Baillieres Clin Haematol*, 11, 1-51.

Foglietta, E., Deidda, G., Graziani, B., Modiano, G. & Bianco, I. (1996) Detection of alpha-globin gene disorders by a simple PCR methodology. *Haematologica*, 81, 387-96.

Frackman, S., Kobs, G., Simpson, D. & Storts, D. (1998) Betaine and DMSO: Enhancing Agents for PCR. Promega Notes, 65, 27.

Fucharoen, G., Sanchaisuriya, K., Sae-ung, N., Dangwibul, S. & Fucharoen, S. (2004) A simplified screening strategy for thalassaemia and haemoglobin E in rural communities in south-east Asia. *Bull World Health Organ*, 82, 364-72.

Fucharoen, S., Fucharoen, G., Sanchaisuriya, K. & Pengjam, Y. (2002) Molecular analysis of a thai β -thalassaemia heterozygote with normal haemoglobin A₂ level: implication for population screening. *Ann Clin Biochem*, 39, 44-9.

Fucharoen, S., Sanchaisuriya, K., Fucharoen, G., Panyasai, S., Devenish, R. & Luy, L. (2003) Interaction of hemoglobin E and several forms of α -thalassemia in Cambodian families. *Haematologica*, 88, 1092-8.

Fucharoen, S., Singsanan, S., Hama, A., Fucharoen, G. & Sanchaisuriya, K. (2007)

Rapid molecular characterization of Hb Queens and Hb Siam: two variants easily misidentified as sickle Hb. *Clin Biochem*, 40, 137-40.

Fucharoen, S. & Winichagoon, P. (1997) Hemoglobinopathies in Southeast Asia: molecular biology and clinical medicine. *Hemoglobin*, 21, 299-319.

Fucharoen, S. & Winichagoon, P. (2011) Haemoglobinopathies in southeast Asia. *Indian J Med Res*, 134, 498-506.

Galanello, R. & Origa, R. (2010) Beta-thalassemia. *Orphanet J Rare Dis*, 5, 11.

Higgs, D.R., Vickers, M.A., Wilkie, A.O., Pretorius, I.M., Jarman, A.P. & Weatherall, D.J. (1989) A review of the molecular genetics of the human α -globin gene cluster. *Blood*, 73, 1081-104.

Ho, P.J., Hall, G.W., Luo, L.Y., Weatherall, D.J. & Thein, S.L. (1998) Beta-thalassaemia intermedia: is it possible consistently to predict phenotype from genotype. *Br J Haematol*, 100, 70-8.

Ho, S.S., Chong, S.S., Koay, E.S., Chan, Y.H., Sukumar, P., Chiu, L.L., Wang, W., Roy, A., Rauff, M., Su, L.L., Biswas, A. & Choolani, M. (2007) Microsatellite markers within --SEA breakpoints for prenatal diagnosis of HbBarts hydrops fetalis. *Clin Chem*, 53, 173-9.

<http://globin.cse.psu.edu/>. [July 10, 2010]

<http://www.ncbi.nlm.nih.gov/pubmed/>. [July 10, 2010]

Jadaon, M.M., Dashti, A.A., Lewis, H.L. & Habeeb, F.M. (2009) Whole-blood polymerase chain reaction and restriction fragment length polymorphism: a simplified method by microwave irradiation. *Med Princ Pract*, 18, 280-3.

Joseph, S. & Russell, D.W. (2001) Molecular Cloning; A Laboratory Manual. Cold Spring Harbor Laboratory, New York.

Kim, J.E., Kim, B.R., Woo, K.S., Kim, J.M., Park, J.I. & Han, J.Y. (2011) Comparison of capillary electrophoresis with cellulose acetate electrophoresis for the screening of hemoglobinopathies. *Korean J Lab Med*, 31, 238-43.

Kreader, C.A. (1996) Relief of amplification inhibition in PCR with bovine serum albumin or T4 gene 32 protein. *Appl Environ Microbiol*, 62, 1102-6.

Kutlar, F. (2007) Diagnostic approach to hemoglobinopathies. *Hemoglobin*, 31, 243-50.

Lebnak, T., Fucharoen, P., Fucharoen, S., Sirithanawattanakul, N. & Tunpichit, W.

(2005) Thalassemia: From molecular biology to clinical medical (in Thai). Nontaburi, Chumnum Sahakorn Thai Printing.

Lin, L.I., Lin, K.S., Lin, K.H. & Chang, H.C. (1991) The spectrum of β -thalassemia mutations in Taiwan: identification of a novel frameshift mutation. *Am J Hum Genet*, 48, 809-12.

Liu, J.Z., Xiao, B., Wang, Q.T. & Wang, L.R. (2009) Rapid diagnosis of the α -thalassemia-1 Southeast Asian type deletion using a single tube real-time SYBR-polymerase chain reaction combined with dissociation curve analysis. *Hemoglobin*, 33, 546-9.

Liu, Y.T., Old, J.M., Miles, K., Fisher, C.A., Weatherall, D.J. & Clegg, J.B. (2000) Rapid detection of α -thalassaemia deletions and α -globin gene triplication by multiplex polymerase chain reactions. *Br J Haematol*, 108, 295-9.

Luawsombut, W. (1998) Thalassemia (in Thai). Bangkok, O.S. Printing House.

McCusker, J., Dawson, M.T., Noone, D., Gannon, F. & Smith, T. (1992) Improved method for direct PCR amplification from whole blood. *Nucleic Acids Res*, 20, 6747.

Mercier, B., Gaucher, C., Feugeas, O. & Mazurier, C. (1990) Direct PCR from whole blood, without DNA extraction. *Nucleic Acids Res*, 18, 5908.

Munkongdee, T., Vattanaviboon, P., Thummarati, P., Sewamart, P., Winichagoon, P., Fucharoen, S. & Svasti, S. (2010) Rapid diagnosis of α -thalassemia by melting curve analysis. *J Mol Diagn*, 12, 354-8.

Musso, M., Bocciardi, R., Parodi, S., Ravazzolo, R. & Ceccherini, I. (2006) Betaine, dimethyl sulfoxide, and 7-deaza-dGTP, a powerful mixture for amplification of GC-rich DNA sequences. *J Mol Diagn*, 8, 544-50.

Nopparatana, C., Panich, V., Saechan, V., Sriroongrueng, V., Rungjeadpha, J., Pornpatkul, M., Laosombat, V. & Fukumaki, Y. (1995) The spectrum of β -thalassemia mutations in southern Thailand. *Southeast Asian J Trop Med Public Health*, 26 Suppl 1, 229-34.

Ou-Yang, H., Hua, L., Mo, Q.H. & Xu, X.M. (2004) Rapid, accurate genotyping of the common - α (4.2) thalassaemia deletion based on the use of denaturing HPLC. *J Clin Pathol*, 57, 159-63.

Panyasai, S., Fucharoen, S., Surapot, S., Fucharoen, G. & Sanchaisuriya, K. (2004)

Molecular basis and hematologic characterization of $\delta\beta$ -thalassemia and hereditary persistence of fetal hemoglobin in Thailand. *Haematologica*, 89, 777-81.

Polski, J.M., Kimzey, S., Percival, R.W. & Gross, L.E. (1998) Rapid and effective processing of blood specimens for diagnostic PCR using filter paper and Chelex-100.

Mol Pathol, 51, 215-7.

Pornphannukool, S., Fucharoen, S., Fucharoen, G., Sae-ung, N. & Sanchaisuriya, K. (2008) Effective routine screening for α - thalassemia 1 carrier with the SEA deletion. *J Med Tech Phy Ther*, 20, 201-9.

Pornprasert, S., Wiengkum, T., Srithep, S., Chainoi, I., Singbootra, P. & Wongwiwatthanakit, S. (2011) Detection of α -thalassemia-1 Southeast Asian and Thai type deletions and β -thalassemia 3.5-kb deletion by single-tube multiplex real-time PCR with SYBR Green1 and high-resolution melting analysis. *Korean J Lab Med*, 31, 138-42.

Quaife, R., al-Gazali, L., Abbes, S., Fitzgerald, P., Fitches, A., Valler, D. & Old, J.M. (1994) The spectrum of β thalassaemia mutations in the UAE national population. *J Med Genet*, 31, 59-61.

Radstrom, P., Knutsson, R., Wolffs, P., Lovenklev, M. & Lofstrom, C. (2004) Pre-PCR processing: strategies to generate PCR-compatible samples. *Mol Biotechnol*, 26, 133-46.

Rees, W.A., Yager, T.D., Korte, J. & von Hippel, P.H. (1993) Betaine can eliminate the base pair composition dependence of DNA melting. *Biochemistry*, 32, 137-44.

Sanguansermsri, T., Lebnak, T. & Fucharoen, P. (1998) Thalassemias: Laboratory guideline (in Thai). Thalassemia Foundation of Thailand, Bangkok.

Santoro, M.M., Liu, Y., Khan, S.M., Hou, L.X. & Bolen, D.W. (1992) Increased thermal stability of proteins in the presence of naturally occurring osmolytes. *Biochemistry*, 31, 5278-83.

Shuber, A.P., Grondin, V.J. & Klinger, K.W. (1995) A simplified procedure for developing multiplex PCRs. *Genome Res*, 5, 488-93.

Singsanan, S., Fucharoen, G., Savongsy, O., Sanchaisuriya, K. & Fucharoen, S. (2007) Molecular characterization and origins of Hb Constant Spring and Hb Pakse in Southeast Asian populations. *Ann Hematol*, 86, 665-9.

Sirichotiyakul, S., Saetung, R. & Sanguansermsri, T. (2003) Analysis of β -thalassemia mutations in northern Thailand using an automated fluorescence DNA sequencing technique. *Hemoglobin*, 27, 89-95.

Sirichotiyakul, S., Saetung, R. & Sanguansermsri, T. (2009a) Prenatal diagnosis of β -thalassemia/Hb E by hemoglobin typing compared to DNA analysis. *Hemoglobin*, 33, 17-23.

Sirichotiyakul, S., Tantipalakorn, C., Sanguansermsri, T., Wanapirak, C. & Tongsong, T. (2004) Erythrocyte osmotic fragility test for screening of α -thalassemia-1 and β -thalassemia trait in pregnancy. *Int J Gynaecol Obstet*, 86, 347-50.

Sirichotiyakul, S., Wanapirak, C., Srisupundit, K., Luewan, S. & Tongsong, T. (2009b) A comparison of the accuracy of the corpuscular fragility and mean corpuscular volume tests for the α -thalassemia 1 and β -thalassemia traits. *Int J Gynaecol Obstet*, 107, 26-9.

Sirinawin, J., Limwong, C., Ruengwutlaed, P., Sukpanichanun, S., Wanacheewanawin, W. & Tunpichit, W. (2004) Thalassemia: Prevention and control (in Thai). Faculty of Medicine, Siriraj Hospital, Mahidol University, Bangkok.

Siriratmanawong, N., Fucharoen, G., Sanchaisuriya, K., Ratanasiri, T. & Fucharoen, S. (2001) Simultaneous PCR detection of β - thalassemia and α - thalassemia 1 (SEA type) in prenatal diagnosis of complex thalassemia syndrome. *Clin Biochem*, 34, 377-80.

Sritong, W., Tatu, T., Seatung, R. & Sanguansermsri, T. (2004) Rapid identification of β -thalassemia mutations by mutagenically separated polymerase chain reaction. Chiang Mai Med Bull, 43, 133-41.

Steinberg, M.H., Forget, B.G., Higgs, D.R. & Nagel, R.L. (2001) Disorders of Hemoglobin : Genetics, Pathophysiology, and Clinical Management (First edition). New York, Cambridge University Press.

Suwannasin, S. (2009) Study of molecular background of hematologically silent α -thalassemia and development of polymerase chain reaction to detect double heterozygous α/β -thalassemia commonly found in Thais (MS Thesis). Master of Science (Medical Technology). Chiang Mai University, Chiang Mai.

Tan, A.S., Quah, T.C., Low, P.S. & Chong, S.S. (2001) A rapid and reliable 7-deletion multiplex polymerase chain reaction assay for α -thalassemia. Blood, 98, 250-1.

Tangvarasittichai, O., Jeenapongsa, R., Sitthiworanan, C. & Sanguansermsri, T. (2004) Diagnostic value of combined parameters for α -thalassemia-1 screening in pregnant women. Naresuan University Journal, 12, 19-24.

Tatu, T. & Kasinrerk, W. (2011) A novel test tube method of screening for hemoglobin E. Int J Lab Hematol, 34, 59-64.

Tatu, T., Kiewkarnkha, T., Khuntarak, S., Khamrin, S., Suwannasin, S. & Kasinrerk, W.

(2012) Screening for co-existence of α -thalassemia in β -thalassemia and in HbE heterozygotes via an enzyme-linked immunosorbent assay for Hb Bart's and embryonic ζ -globin chain. *Int J Hematol*, 95, 386-93.

Tujinda, S., Tunpichit, W., Fucharoen, S., Fucharoen, G., Limwong, C., Fucharoen, P., *et al.* (2010) The laboratory guideline for thalassemia and hemoglobinopathy diagnosis (in Thai). Bangkok, Mat-Ded Co. Ltd.

Vrettou, C., Traeger-Synodinos, J., Tzetis, M., Malamis, G. & Kanavakis, E. (2003)

Rapid screening of multiple β -globin gene mutations by real-time PCR on the LightCycler: application to carrier screening and prenatal diagnosis of thalassemia syndromes. *Clin Chem*, 49, 769-76.

Walsh, P.S., Metzger, D.A. & Higuchi, R. (1991) Chelex 100 as a medium for simple extraction of DNA for PCR-based typing from forensic material. *Biotechniques*, 10, 506-13.

Wang, W., Ma, E.S., Chan, A.Y., Prior, J., Erber, W.N., Chan, L.C., Chui, D.H. & Chong, S.S. (2003) Single-tube multiplex-PCR screen for anti-3.7 and anti-4.2 α -globin gene triplications. *Clin Chem*, 49, 1679-82.

Wasi, P. (1983) Population Screening. In Weatherall, D.J. (ed) : The Thalassemias ; Methods in Hematology. New York, Churchill Livingstone, pp 134-44.

Waye, J.S. & Chui, D.H. (2001) The α -globin gene cluster: genetics and disorders. Clin Invest Med, 24, 103-9.

Weatherall, D.J. (1983) The Thalassemia ; Methods in Hematology. New York, Churchill Livingstone.

Weatherall, D.J. & Clegg, J.B. (2001) The Thalassemia Syndromes (Fourth edition). Oxford, Blackwell Science.

Yang, Y.G., Kim, J.Y., Song, Y.H. & Kim, D.S. (2007) A novel buffer system, AnyDirect, can improve polymerase chain reaction from whole blood without DNA isolation. Clin Chim Acta, 380, 112-7.

Zheng, T., Brittain, T., Watmough, N.J. & Weber, R.E. (1999) The role of amino acid α 38 in the control of oxygen binding to human adult and embryonic haemoglobin Portland. Biochem J, 343 , 681-5.