

REFERENCES

- Adair, G.S. (1925) The osmotic pressure of hemoglobin in the absence of salts. Proc. R. Soc. Lond. (Biol.), 1109A : 292.
- Antonarakis, S.E., Boehm, C.D., Giardina, P.V.J., and Kazazian, H.H. (1982) Non random association of polymorphic restriction sites in the β - globin gene complex. Proc Natl Acad Sci, USA, 79: 137.
- Baralle, F.E., Shoulders, C.C., and Proudfoot, N.J. (1980) The primary structure of the human ϵ - globin gene. Cell, 21: 621.
- Beggs, A., Koenig, M., Boyce, F., and Kunkel, L. (1990) Detection of 98% of DMD/BMD deletions by PCR. Human Genetics, 86: 45-48.
- Bernini F.L. (1998) Alpha - thalassemia. Clinical Hematology, 11:53-90.
- Bowden, D.K., Hill, A.V.S., Higgs, D.R., Oppenheimer, S.J., Weatherall, D.J., and Clegg, J.B. (1987) Different hematologic phenotypes are associated with the leftward (- alpha 4.2) and rightward (- alpha 3.7) alpha⁺ thalassemia deletions. J Clin Invest., 79:39.
- Bowden, D.K., Vickers, M.A., and Higgs, D.R. (1992) A PCR based strategy to detect the common severe determinants of α - thalassemia. British Journal of Hematology, 81:104-108.
- Brown, B.A. (1988) Hematology : Principles and procedures (5 ed.) Lea & Febbiger, Philadelphia.
- Bunn, H.F. , and Forget, B.G. (1986) Hemoglobin: Molecular genetic and Clinical Aspects. Philadelphia, Saunders, 257-261.

- Chamberlain, J.S., Gibbs, R.A., Ranier, J.E., Nguen, P.N., and Caskey, C.T. (1988)
Deletion screening of the Duchene muscular dystrophy locus via multiplex
DNA amplification. *Nucleic Acids Research*, 16: 1141-1156.
- Chan, V., Chan, V.W., Tang, M., Lau, K., Todd, D., and Chan, T.K. (1997)
Molecular defects in HbH hydrops fetalis. *British Journal of Haematology*,
96 : 224-228.
- Dacie J.V., and Lewis, S.M. (1964) *Practical Haematology* (ed.4.). Grune & Stratton,
New York,
- Dozy, A.M., Kan, Y.W., Embury, S.H., Mentzer, W.C., Wang, W.C., Lubin B.,
Davis, J.R., and Konig, H.M. (1979) Alpha – globin gene organization in
blacks precludes the severe form of alpha-halassemia. *Nature*, 280:605-607.
- Erich, H.A. (1992) *PCR Technology : Principles and applications for DNA
amplification*. W.H. Freeman and Company, New York.
- Fei, Y.J., Oner, R. and Bozkurt, G., Gu, W.H., Altay, C., Gurgey, A., Fattoum, S.,
Basal, E and Huisman, T. H. (1992) Hb H disease caused by a
homozygosity for the AATAAA→AATAAG mutation in the polyadenylation
site of the alpha 2 - globin gene: Hematological observations. *Acta
Haematologica*, 88: 82-85.
- Fischel-Ghodian N., Vickers, M.A., Seip, M., Winichagoon, P., and Higgs, D.R.
(1988) Characterization of two deletion that remove the entire human
zeta - alpha globin gene complex (--THAI and --FIL). *Br.J. Haematol.*,
70: 233-238.
- Fortina, P., Delgrosso K., Rappaport E., Ponez, M., Ballas S.K., Schwat, Z.E., and
Surreys (1988) A large deletion encompassing the entire alpha-like globin

gene cluster in a family of Northern European extraction. *Br.J.Haematol.*,70: 233-238.

Fritsh, E.F., Lawn, R.M., Maniatis, T.(1980) Molecular cloning and Characterization of the human β -like globin gene cluster. *Cell*, 19 : 959.

Galanello, R., Aru, B., Dessi, C., Addis, M., Pagliatti, E., Melis, M.A., Massa, P., Giagu, N., and Barella, S. (1992) Hb H disease in Sardinia : Molecular, hematological and Clinical aspects. *Acta Haematologica*, 88: 1-6.

Galanello, R. Sallaino, C., Paglietti, E., Barella, S., Perra, C., Doneddu, I., Pirroni, M.A., Maccioni, L., and Cao, A. (1998) α -Thalassemia carrier identification by DNA analysis in the screening for thalassemia *Am J Hematol*, 59: 273- 278.

Guyer, R.L. and Koshland, Jr. D.F. (1990) The molecule of the year. *Science*, 246 : 1543-1544.

Higgs, D.R., Hill, A., Bowden, D.K., Weatherall, D.J., and Clegg, J.B. (1984) Independent recombination events between the duplicated human alpha-globin genes : Implications for their concerted evolution. *Nucleic Acids. Res.*, 12: 6965-6977.

Higgs, D.R., Wainscoat, J.S., Flint, J., Aill, A.V., Thein, S.L., Nicholls, R.D., Teal, H., Ayyub, H., Peto, T.E. and Falusi, A.G. (1986) Analysis of the human α - globin gene cluster reveals a highly informative genetic locus. *Proc.Natl Acad Sci , USA*, 83: 5156.

Higgs, D.R., Vickers, M.A., Wilielkie, A.O.M., Pretorius, I.M., Jarman, A.P., and Weatherall, D.J. (1989). A review of the molecular genetics of the human α - globin gene cluster. *Blood*, 73:1081-1104.

- Honig, G.R., Shamsuddin, M., Zaizov, R., Steinherz, M., Solar, I., and Kirschman, C. (1981) A new unstable variant associated with α - thalassemia -like expression. *Blood*, 57:705.
- Honig, G.R., Shamsuddin, M., Vida, L.N. Mompoin, M., Valcouat, E., Bowie, L.J., Jones, E.C., Powers P.A., Spritz, R.A. and Guis, M. (1984) Hemoglobin Evanston (A 14 Trp > Arg) An unstable α - chain variant expressed as α - thalssemia, *J Clin Invest*, 73:1740.
- Honig, G.R., and Adams III, J.G. (1986) *Human hemoglobin Genetics*. Springer-Verlag, Wein, NY.
- Hundrieser, J., Sanguanserm, T. Papp, T., and Flatz, G. (1988) Alpha- thalassemia in northern Thailand. Frequency of deletion types characterized at the DNA level. *Hum Hered*, 38: 211-5.
- Innis, A.M., Gelfand, H.D., Sninsky, J.J., and White, T.J. (1990) *PCR protocol : A guide to methods and applications*. Academic Press, California.
- Jeffrey, A.J. (1979) DNA sequences in the α γ - , α γ - , δ - and β - globin genes of man. *Cell*, 18:1.
- Jones, J.A., Broszeit, H.K., LeCrone, C.N., and Dellter, J.C. (1981) An improved method for detection of red cell hemoglobin H inclusions, *Am J Med Tech.*, 47: 94.
- Kanavakis, E., Tzotzos, S., Liapaki, K., Metaxou-Mavromati, A., and Kattamis, C. (1988) Molecular basis and prevalence of alpha thalassemia in Greece. *Birth Defects*, 23:377-380.
- Kanavakis, E., Traeger-Synodinos, J., Papasotiriou, I., Vrettou, C., Metaxotou-Mavromati, A., Stamoulakatou, A., Lagona, E., and Kattamis, C. (1996)

The interaction of alpha zero thalassemia with Hb Icaria : Three unusual cases of hemoglobinopathy, H. British Journal of Hematology, 92: 332-335.

Kerem, B.S., Rommens, J.M., Buchanan, J.A., Markiewicz, D., Cox, T.K., (1988)
Characterization of the cystic fibrosis gene : genetic analysis. Science,
245: 1073-1080.

Lauer, J., Shen, C.K.J., and Maniatis, T. (1980) The chromosomal arrangement of
human α - like globin gene. Sequence homology and α -globin gene deletions.
Cell, 20:119-130.

Lemmens-Zygulska, M., Eigel, A. Helbig, B. and Sanguansermsri, T., and Flatz, G
(1996) Prevalence of alpha-thalassemias in northern Thailand. HumGenet,
98: 3345-7.

Liebhaber, S.A., Goossens, N., and Kan, Y.W. (1980) Cloning and complete
nucleotide sequence of human 5'- α - globin gene. Proc Natl Acad Sci. USA,
77:7054.

Liebhaber, S.A., Goossens, N., and Kan, Y.W. (1981) Homology and concerted
evolution at the $\alpha 1$ and $\alpha 2$ loci of human α - globin. Nature, 290:26.

Liebhaber, S.A., and Kan, Y.W. (1983) α - thalassemia caused by an unstable α -
globin mutant. J. Clin Invest, 71: 461.

Liebhaber, S.A. (1989) Alpha - thalassemia. Hemoglobin, 13:685-731

Lui, T.C., Chiou, S.S., Lin, S.F. Chen, T.P., Tseng, W.P., Chen, P.H., and Chang,
J.G. (1994) Molecular basis and hematological characterization of HbH
disease in southeast Asia. American Journal of Hematology, 45: 293-297.

- Makonkawkeyoon, L., Sanguansermisri, T., Asato, T., Nakashima, Y., and Takei, H. (1993a) Rapid detection of chain termination mutations in the $\alpha 2$ globin gene. *Blood*, 82: 3503-3504.
- Makonkawkeyoon, L., Masaru Nagamine M., Sanguansermisri, T., and Takei, H. (1993b) Molecular Characterization and the severity of Hemoglobin H disease in Northern Thailand. *Ryukyu Med. J.*, 13 : 159 - 166
- Miller, M.A., Dykes, D.D., and Polesky, H.F. (1988) A simple salting out procedure for extracting DNA from human nucleated cells. *Nucleic Acids Res*, 16:1215.
- Nicholls, R.D., Fischel-Ghodsian, N., and Higgs, D.R. (1987) Recombination at the human alpha - globin gene cluster : Sequence features and topological constraints. *Cell*, 49: 369-378.
- Nienhuis, A.W., and Maniatis, T. (1987) Structure and expression of globin genes in erythroid cells, in the molecular Basis of blood disease, edited by G Stamatoyannopoulos, A.W., Nienhuis, P., Leder, P.W. Majerus, Saunder, Philadelphia, 28
- Oppenheimer, S.T., Higgs D.R., Weatherall D.J., and Spark, R.A. (1984) Alpha thalassemia in Papua New Guinea. *Lancet*, 1:424-426.
- Oppenheimer, S.T., Higgs D.R., Weatherall D.J., and Spark, R.A. (1988)
- Orkin, S.H. (1978) The duplicated human α - globin genes lie close together in cellular DNA. *Proc Natl Acad Sci USA.*, 75 : 5950
- Perutz, M.F. (1986) X-ray analysis of hemoglobin. *Science*, 140:863.
- Proudfoot, N.J., and Maniatis, T. (1980) The structure of a human α - globin psuedogene and its relationship to α - globin duplication. *Cell*, 21 : 537.

- Raven, J.L., and Tooze, J.A. (1973) α - thalassemia in Britain, Br Med J., 4:486
- Saiki, R.k., Bugawan, T.L., Horn, G.T., Mullis, K.B., and Erlich, H.A. (1986)
Analysis of enzymatically amplified β - globin and HLA-Dq α DNA with
allele specific oligonucleotide probes. Nature, 324: 163-166.
- Sanguansermisri, T., Matrogon, S., Changlosh, L., and Fletz, G. (1979) Hemoglobin
Suan - Dok (α 2^{109 (G16) LEU.ARG} β 2) : An unstable variant associated
with α -thalassemia. Hemoglobin, 3:161.
- Scharf, S.J., Horn, G.T., and Erlich, H.A. (1986) Direct cloning and sequence
analysis of enzymatically amplified genomic sequences, Science, 253:
1076-1078.
- Schmidt, R. M., and Brosius, E.M. (1974) Basic Laboratory Methods of
Hemoglobinopathy Detection, HEW Pub. USA. No. (CDC) 74-8266.
- Shepard, M.K., Weatherall, D.J., and Conley, C.L. (1962) Semiquantitative
estimation of the distribution of fetal hemoglobin in red cell populations,
Bull. Johns Hopkins Hosp., 110: 293.
- Slightom, J.L., Blechl, A.E., Smithies, O. (1980) Human γ^G and γ^A - globin genes :
Complete nucleotide sequences suggest that DNA can be exchanged
between these duplicated genes. Cell, 21:627.
- Smetina, N.S., and Huisman, T.H. (1996) Detection of α -thalassemia2 (-3.7 kb) and
its Corresponding Triplication $\alpha\alpha\alpha$ (Anti -3.7 kb) by PCR : An improved
Technical Change. Am. J. Hematol., 53:3 202-3
- Spritz, R.A., Deriel, J.K., Forget, B.G., and Weissman, S.M. (1980) Complete
nucleotide sequence of the human δ - globin gene. Cell, 21:639.

- Svedberg, T., and Fahraeus, R. (1926) A new method for the determination of the molecular weight of the proteins. *J. Am. Chem. Soc.*, 48:430.
- Thein, S.L., Wallace, R.B., Pressley, L., Clegg, J.B., Weatherall, D.J., and Higgs, D.R. (1988). The polyadenylation site mutation in the α -globin gene cluster. *Blood*, 71:313.
- Wainscoat, J.S., Hill, A.V.V., Boya, A., Flint, J., Hernandez, M., Thein, S.L., Old, J.M., Lynch, J.R., Falusi, A.G., and Weatherall, D.J. (1982) Evolutionary relationships of human populations from an analysis of nuclear DNA polymorphisms. *Nature*, 319 : 491.
- Wasi, P. (1983) Hemoglobinopathies in Southeast Asia Distribution and Evolution of Hemoglobin and globin loci. In J.E. Bowman (Ed.), Elsevier Science Publishing Co., Inc.
- Whitelaw, E. and Proudfoot, N., (1986) α -thalassemia caused by a poly (A) site mutation reveals that transcriptional termination is linked to 3' end processing in the human $\alpha 2$ globin gene. *EMBO. J.*, 5: 2915-2922.
- Weatherall, D.J., and Clegg, J.B. (1981) The thalassemia Syndromes (ed. 3). Liverpool. Blackwell.
- Weatherall, D.J. (1995) The thalassemia. *Hematology* (ed.5). McGraw – Hill, Inc.
- Winichagoon, P., Higgs, D.R., Goodbourn, S.E.Y., Clegg, J.B., Weatherall, D.J. and Wasi, P., (1984) The molecular basis of alpha thalassemia in Thailand. *EMBO J.*, 3: 1813-1818.

Winichagoon, P., Fucharoen, S., and Wasi, P. (1992) The molecular basis of α - thalassemia in Thailand. *Southeast Asian J. Trop. Med. Pulic Health.* , 23: 7-13.

Yenchitsomanus, P.T., Summers, K.M., Bhatia, K.K., Cattani, J., and Board, P.G. (1985) Alpha - zero - and beta - thalassemia in Thai family : Unusually mild homozygous beta - zero - thalassemia without alpha - globin gene deletion. *Am.J.Hum.Genet.*, 37:778.