VII. References

Aru,B.,Dessi,C., Galanello,R.,Paglietti,E., Giau,L.,Barella,S., Argiolu,F., Cocco, S., and Cao, A., (1989). Abstract, 3rd International Conference on Thalassemia and Hemoglobinopathies, Sardinia.

Bunn,H.F., Forgot,B.G.,and Ranney,H.M., (1977). Hemoglobinopathies, p.28. In W.B.Saunders (Ed.), U.S.A.

Bunn, H.F., and Forgot, B.G., (1986). Hemoglobin : Molecular, Genetic and Clinical Aspects, p.257-261. In W.B.Saunders, Philadelphia.

Bowden,D.K.,Hill, A.V., Higgs, D.R., Oppenheimer,S.J.,Weatherall,D.J., and Clegg, J.B., (1989). Different hematologic phenotypes are associated with the leftward (-alpha4.2) and rightward (-alpha3.7) alpha+- thalassemia deletions. J.Clin.Invest., 79:39-43.

Clegg, J.B., and Weatherall,D.J.,(1967).Haemoglobin synthesis in alpha-thalassemia (haemoglobin H disease). Nature, 215:1241-1243.

Clegg,J.B.,Weatherall,D.J.,and Milner,P.F.,(1971).Hemoglobin Constant Spring - a chain termination mutant?. Nature, 234:337-340.

Chan, V., Chan, T.K., Liang,S.T., Ghosh, A., Kan, Y.W., and Todd, D., (1985). Hydrops fetalis due to an unusual form of Hb H disease. Blood, 66:224-228.

Chee, B.M., Hsia,Y.E., Hunt, J.A., and Yuen, J., (1991). Gap PCR for rapid detection of single alpha-globin gene deletions. Am.J.Hum. Genet.,49:184.

Deisseroth,A.,Nienhuis,A.,Turner,P.,Velez,R.,Anderson,W.F.,Ruddle,F., Lawrence,J., Creagan, R.,and Kucherlapati,R.,(1977). Localization of the human α-globin structuregene to chromosome 16 in somatic cell hybrids by molecular hybridization assay. Cell,12:205-218.

Deisseroth,A., Nienhuis,A., Lawrence,J., Giles,R.,Turner,P.,Ruddle,F.H., (1978). Chromosomal localization of human β-globin gene on human chromosome 11 in somatic cell hybrids. Proc.Natl.Acad. Sci.U.S.A. 75:1456-1460

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-thalassemia. Nature, 280:605-607.

Embury,S.H.,Miller,J.A., Dozy,A.M.,Kan,Y.W., Chan, V., and Todd,D., (1980). Two different molecular organization account for the single α globin gene of the α-thalassemia-2 genotype. J. Clin. Invest., 66:1319-1325.

Felber, B.K., Orkin, S.H., and Hamer, D.H., (1982). Abnormal RNA splicing causes one form of alpha thalassemia.Cell, 29:895-902.

Flint,J., Hill,A.V., Bowden,D.K., Oppenheimer,S.J.,Sill,P.R.,Serjeantson, S.W., Bana - Koiri, J., Bhatia, K., Alpers, M.P., and Boyce, A.J., (1986). High freguencies of alpha-thalassemia are the result of natural selection by malaria. Nature, 321:744-750.

Fischel-Ghodsian,N., Higgs,D.R., and Beyer,E.R.,(1987). Function of a new globin gene [letter]. Nature, 329:397.

Fodde,R., Losekoot,M.,Van Den Broek,M.H.,Oldenberg,M., Rashida,N., Schreuder,A.,Wijnen,J.T.,Giordano,P.C., Nayudu,N.V.,and Khan, P.M., (1988). Prevalence and molecular heterogeneity of alfa + thalassemia in two tribal populations from Andhra Pradesh India. Hum.Genet.80:157-160.

Fischel-Ghodsian,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., Schwart,Z.E.,and Surrey,S.,(1988). A large deletion encompassing the entire alpha-like globin gene cluster in a family of Northern European extraction. Nucleic.Acids.Res., 16:11223-11225.

Fucharoen, S.,Winichagoon, P.,Pootrakul, P.,Piankijagum, A., and Wasi, P.,(1988). Thalassemia : Pathophysiology and Management ,Part A,p.309 In S. Fucharoen (Ed), Alan R.Liss,Inc.,New york.

Goodbourn, S.E.Y.,Higgs,D.R.,Clegg, J.B., and Weatherall, D.J., (1982). Molecular basis of length polymorphism in the human zeta-globin gene complex. Proc.Natl.Acad.Sci.U.S.A., 80:5022-5026.

Galanello, R., Pirastu, M., Melis,M.A., Paglietti, E., Moi,P., and Cao, A., (1983). Phenotype-genotype correlation in haemoglobin H disease in childhood. J.Med.Genet.,20:425-429.

Galanello, R., Maccioni, L., Ruggeri, R., Perseu, L., and Cao, A., (1984). Alpha thalassemia in Sardinian newborns. Br. J. Haematol., 58 : 361-368.

Heywood, J.D., Karon, M., and Weissman, S., (1964). Amino acids incorporation into alpha- and beta - chains of hemoglobin by normal and thalassemic reticulocytes. Science, 146:530-531.

Higgs, D.R., Pressley,L.,Aldridge,B.,Clegg,J.B.,Weatherall, D.J.,Cao, A., Hadjiminas, M.G., Kattamis, C., Metaxotou - Mavromati, A., Rachmilewitz, E.A.,Sophocleous,T.,(1981). Genetic and molecular diversity in non-deletion Hb H disease. Proc.Natl.Acad.Sci.U.S.A., 78:5833-5837.

Higgs,D.R., Goodbourn, S.E.Y., Lam,J.,Clegg, J.B.,and Weatherall,D.J., (1983). Alpha - thalassemia caused by a polyadenylation signal mutation. Nature, 306:398-400.

Higgs, D.R., and Weatherall, D.J.,(1983). Alpha Thalassemia, p37. In S. Piomelli (Ed), Current Topics in Hematology.AR Liss,New York.

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.

Hill,A.V.S.,(1986). The distribution and molecular basis of thalassemia in Oceania. Ph.D. Thesis, Oxford University.

Hardison, R.C., Sawada, I., Cheng, J.F., Shen, C.K., and Schmid, C.W., (1986). A previously undetected pseudogene in the human alpha globin gene cluster.Nucleic.Acids.Res.,14:1903-1911.

Hsu,S.L., Marks,J.,Shaw,J.P.,Tam,M., Higgs,D.R., Shen,C.C., and Shan, C.K.,(1988). Structure and expression of the human theta 1 globin gene. Nature,331:94-96.

Hsia, Y.E., Ford, C.A., Shapiro, L.J., Hunt, J.A., and Ching, N.S.P., (1989). Molecular screening for Haemoglobin Constant Spring. Lancet, 1: 988 - 991.

Higgs,D.R., Vickers, M.A., Wilkie, 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.

Ireland, J.H., Hsia, Y.E., Chu, B.M., and Chui, D.H.K., (1991). Anti-zeta will reliably detect (--SEA) double alpha deletions. Am.J.Hum. Genet., 49:329.

Jarman,A.P., Nicholls,R.D.,Weatherall, D.J., Clegg,J.B.,and Higgs,D.R., (1986). Molecular characterization of a hypervariable region downstream of the human alpha-globin gene cluster. EMBO.J., 5:1857-1863.

Jarman, A.P., and Higgs, D.R., (1988). A new hypervariable marker for the human alpha-globin gene cluster. Am.J.Hum.Genet.,43:249-256.

Kan,Y.W.,Schwartz,E.,and Nathan,D.G.,(1969). Globin chain synthesis in the alpha thalassemia syndromes. J.Clin.Invest.,47:2512-2522.

Kan, Y.W., Dozy, A.M., Stamatoyannopoulos, G., Hadjiminas, M.G., Zachariades, Z.,Furbetta, M.,and Cao,A., (1979). Molecular basis of hemoglobin - H disease in the Mediterranean population. Blood, 54 : 1434-1438.

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.

Kattamis, C., Tzotzos, S., Kanavakis, E., Synodinos, J., and Metaxou-Mavromati, A., (1988). Correlation of clinical phenotype to genotype in haemoglobin H disease. Lancet,1:442-444.

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.

Langer, P.R., Waldrop, A.A., and Ward, D.C., (1981). Enzymatic synthesis of biotin-labeled polynucleotides : nucleic acid affinity probes. Proc. Natl. Acad.Sci.U.S.A.,78:6333-6337.

Leary, J.J., Brigati, D.J., and Ward, D.C.,(1983). Rapid and sensitive colorimetric method for visualizing biotin- labeled- DNA probes hybridized to DNA or RNA immobilized on nitrocellulose : Bio-blots. Proc.Natl.Acad.Sci. U.S.A.,80:4045-4049.

Liebhaber,S.A.,Cash,F.E.,and Main,D.M.,(1985). Compensatory increase in alpha 1-globin gene expression in individuals heterozygous for the alpha-thalassemia-2 deletion. J.Clin.Invest.76:1057-1064.

Liebhaber,S.A.,(1989). α-Thalassemia. Hemoglobin,13:685-731.

Laig,M.,Pape,M.,Hundriesser,J.,G.,Sanguansermsri,T.,Das,D.M.,Deka,R. , Yongvanit,P.,and Mularlee,N.,(1990). The distribution of the Hb Constant Spring gene in Southeast Asian populations.Hum.Genet. ,84:188-190.

Lebo, R.V., Saiki, R.K., Swanson, K., Montano, M.A., Erlish, H.A., and Golbus, M.S., (1990). Prenatal diagnosis of α - thalassemia by polymerase chain reaction and dual restriction enzyme analysis. Hum.Genet.,85:293-299.

Morle, F., Lopez, B., Henni, T., and Godet, J., (1985). α -Thalassemia associated with the deletion of two nucleotides at position-2 and -3 preceeding the AUG codon. EMBO.J., 5:1245-1250.

Maniatis,T., Fritsch,E.E., Lauer,J., Lawn, R.M., Proudfoot, N.J.,Shancer, H.M.,and Chen, C.K.J., (1981). The structure and chromosomal arrangement of human globin genes, p. 15-31. In Stamatoyannopoulos (Ed), Organization and Expression of Globin Genes. Alan R Liss, New York.

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.

Muglia, M., Annesi, G., Gabriele, A.L., Covello, M., and Brancati, C., (1993). α-Thalassemia in a Southern Italian population (detection by a non- radioactive procedure).Hemoglobin,17:285-287.

Nicholls, R.D., Fischel - Ghodsian, N., and Higgs, D.R., (1987). Recombination at the human alpha-globin gene cluster: sequence features and topological constrains. Cell,49:369-378.

Orkin, S.H., Goff, S.C., and Hechman, R.L., (1981). Mutation in an intervening sequence splice junction in man. Proc.Natl.Acad.Sci. U.S.A.,78:5041-5045.

Oppenheimer, S.J., Higgs, D.R., Weatherall, D.J., Barker, J., and Spark, R.A., (1984). Alpha thalassemia in Papua New Guinea. Lancet,1: 424-426.

Olivieri, N.F., Chang, L.S., Poon, A.O., Michelson, A.M., and Orkin, S.H., (1987). An alpha-globin gene initiation codon mutation in a black family with Hb H disease. Blood,70:729-732.

Pressley,L.,Higgs,D.R., Clegg, J.B., and Weatherall, D.J., (1980). Gene

Pressley,L.,Higgs,D.R., Clegg, J.B., and Weatherall, D.J., (1980). Gene deletions in an α thalassemia prove that 5' locus is functional. Proc.Natl.Acad.Sci.U.S.A.,77:3586-3589.

Proudfoot,N.J.,and Maniatis,T.,(1980). The structure of a human alpha-globin pseudogene and its relationship to alpha - globin gene duplication. Cell, 21:537-544.

Pressley, L., Higgs, D.R., Clegg, J.B., Perrine, R.P., Pembrey, M.E., and Weatherall, D.J., (1981). A new genetic basis for hemoglobin H disease. N.Engl.Med.,303:1383-1388.

Proudfoot, N.J., Gil,A., and Maniatis, T., (1982). The structure of the human zeta-globin gene and a closely linked , nearly identical pseudogene. Cell, 31:553-563.

Pirastu, M., Saglio, G., Chang, J.C., Cao, A., and Kan, Y.W., (1984). Initiation codon mutation as a cause of α-thalassemia. J.Biol. Chem.,259:12315- 12317.

Peschle,C.,Mavilio,F.,Care,A., Migliaccio,G., Migliaccio,A.R., Salvo,G., Samoggia, P., Petti, S., Guerriero, R., and Marinucci, M., (1985). Hemoglobin switching in human embryos : asynchrony of zeta--alpha and epsilon - gamma - globin switches in primitive and definitive erythropoietic lineage. Nature, 313:235-238.

Paglietti, E., Galanello, R., Moi, P., Pirastu, M., and Cao, A., (1986). Molecular pathology of haemoglobin H disease in Sardinians. Br.J Haematol.,63:485-496.

Southern, E.M., (1975). Detection of specific sequences among DNA fragments seperated by gel electrophoresis.J.Mol.Biol.,98:503-517

Stephen,A., Liebhaber, S.A., Faith, E., Cash, F.E., and Samir, K., (1986). Human alpha globin gene expression : The dominant role of the alpha 2 locus in mRNA and protien synthesis.J.Biol.Chem.,261: 15327-15333.

Stamatoyannopoulos, G., Nienhuis, A.W., Leder, P., and Majerus, P.W., (1987).The alpha - thalassemias, p. 113-115. In S.H. Orkin (Ed.), The Molecular Basis of Blood Disease. W.B. Saunders, Philadelphia.

Trent, R.J.,Wilkinson,T.,Yakas, J.,Carter, J.,Lammi, A.,and Kronenberg, H., (1986). Molecular defects in 2 samples of severe Hb H disease. Scand. J.Hematol.,36:272-279.

Wasi,,P., Pootrakul, S., Pootrakul, P., Pravatmuang, P., Winichagoon,P., and Fucharoen,S.,(1980).Thalassemia in Thailand. Ann.NY.Acad. Sci.,344:352- 363.

Weatherall, D.J., Clegg, J.B., (1981). The Thalassemia Syndromes. (3rd) Oxford, Blackwell.

Wasi,P.,(1983). Hemoglobinopathies in Southeast Asia Distribution and Evolution of Hemoglobin and globin loci, p. 179-208. In J.E. Bowman (Ed.), Elsevier Science Publishing Co.,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.

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 α2 globin gene. EMBO.J.,5: 2915- 2922.

Winichagoon, P., Chalalaksananukul, W., Panyim, S., Fucharoen,S., and Wasi,P.,(1987). Alpha mRNA level in the two types of Hb H disease. Birth Defects, 23:55-60.

Winichagoon,P.,Thonglairoam,V., Fucharoen,S., Tanphaichitr,V.S., and Wasi, P.,(1988). α-Thalassemia in Thailand. Hemoglobin, 12:485-498.

Yenchitsomanus,P.T.,Summers,K.M., Bhatia,K.K., Cattani,J.,and Board, P.G., (1985). Alpha-zero- and beta-zero- thalassemia in a Thai family: unusually mild homozygous beta-zero-thalassemia without alpha-globin gene deletion. Am.J.Hum.Genet.,69:375-377.