XIIIth
MEETING OF THE INTERNATIONAL SOCIETY OF HAEMATOLOGY
(EUROPEAN & AFRICAN DIVISION)

ABSTRACT BOOK

3-8 September 1995
İstanbul, TÜRKİYE

765

ABNORMAL HEMOGLOBINS IN THE CZECH AND SLOVAK POPULATION

K.INDRÁK[1],V.DIVOKÝ[1],V.BRABEC[2],H.FOŘTOVÁ[2],M.DIVOKÁ[1],L.CHROBÁK[3],
D.POSPÍŠILOVÁ[1],A.SAKALOVÁ[4],T.H.J.HUISMAN[5]

Dept.Haematol. in Olomouc(1), Praha(2), Hradec Králové(3), CR
Dept.Haematol. in Bratislava(4), SR
MCG, Augusta, GA, USA(5)

A review of clinical and laboratory findings of nine structural hemoglobin (Hb) variants is presented.

Unstable Hb variants - Hb-Köln, Hb-St.Louis, Hb-Nottingham, Hb-Santa Ana and two new unstable variants were found, namely: Hb-Hradec Králové or $\alpha_2\beta_2$ 115(G17) Ala-Asp - severely unstable Hb resulting in a dominant β-thal trait; Hb-Haňá or $\alpha_2\beta_2$ 63(E7) His-Asn higly unstable variant with a distal histidine substitution resulting in a moderate hemolytic anemia and mild methemoglobinemia in the heterozygotes.

A new variant is also Hb-Olomouc or $\alpha_2\beta_2$ 86(F2) Ala-Asp with a high oxygen affinity and erythrocytosis in father and son.

A combination of Hb-E with β-thal mutation (IVS-I-1/G-A/) and α-globin gene triplication lead to a severe hemolytic anemia and posttransfusion hemosiderosis. The haplotype of two Hb-E families suggests a Czech origin of the mutation.

Hb-M-Milwaukee or $\alpha_2\beta_2$ 67(E11) is manifested rather by hemolysis with Heinz bodies formation than by classical cyanosis.

The family studies show a spontaneous ("de novo") origin of most of the mutations for abnormal β-globin chains synthesis.

766

SELECTIVE AMINOACIDURIA AND HYPERZINCURIA IN THALASSEMIA

Y.Yıldırmak, S.Kemahlı, N.Dinçer, G.Biberoğlu, A.Hasanoğlu, A.Arcasoy
Departments of Paediatrics, Faculty of Medicine, Ankara University and Gazi University, Ankara, Turkey

Objective: Growth retardation and delayed or absent puberty have been reported to be a common problem in transfusion-dependent thalassemics. In our previous studies on thalassemia we have documented zinc deficiency in the presence of hyperzincuria. The cause of increased urinary zinc excretion in thalassemia is not clear. Zinc is known to bind avidly to certain amino acids, especially cysteine and histidine.In children with untreated histidinemia, urinary zinc excretion was positively correlated with urinary histidine excretion. We have investigated the urinary excretion of 13 amino acids and the possible role of these amino acids in hyperzincuria in thalassemic patients.

Design and Patients: Urinary amino acids were assayed by high performance liquid chromatography (HPLC) in 14 thalassemics and 10 healthy children. Urinary zinc excretion was measured by atomic absorption spectrophotometry. Urinary amino acid excretion of the patient and control groups were compared.

Results: Of the 13 urinary amino acids assayed histidine, glycine, lysine and tyrosine were found to be significantly increased in the patients compared with the control group. Although there was marked hyperzincuria in thalassemic patients no correlation was found between increased urinary zinc levels and any of the urinary amino acids.

	Histidine	Cysteine	Lysine	Tyrosine
Patients	872.00±369.3	100.23±65.18	101.52±78.23	120.61±102.07
Controls	325.18±45.2	39.00±18.38	22.18±9.12	41.86±21.37
p	<0.05	<0.01	<0.01	<0.05