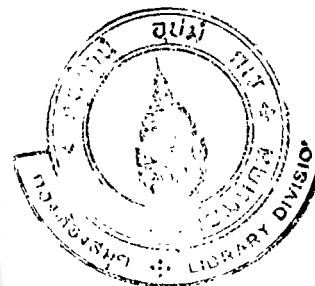


DOUBLE HETEROZYGOSITY FOR HEMOGLOBIN E AND A LEPORE-TYPE HEMOGLOBIN
FOUND IN A THAI WOMAN

BY

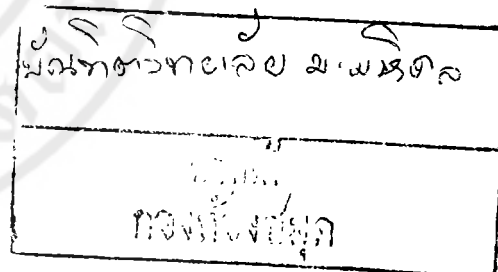
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ABSTRACT

This thesis describes the characterization of two abnormal hemoglobins from a Thai woman having mild anemia, associated with slight hepatosplenomegaly. The patient's hematological profile was as follows: Hb 11.4 g/dl, RBC: $4.91 \times 10^6 / \mu\text{l}$; Hct 34.7%; reticulocyte 2.4%; MCV: 70 fl; MCH: 23.3 pg; MCHC: 30.0 g/dl. Red cell morphology showed moderate hypochromia, slight aniso-poikilocytosis. Hemoglobin typing showed three bands : Hb F, Hb E and an unidentified band (HbX) between Hb F and Hb A₂. Hemoglobin X (12.7%), Hb E (53.0%) and Hb F (34.3%) were separated by DEAE-cellulose chromatography. The abnormal beta chains β^X and β^E , prepared by CM-cellulose chromatography, were subjected to peptide mapping in comparison to β^A . The β^E map showed the loss of peptide βTpIII and the appearance of two new peptides βTpIIIa and βTpIIIb , which suggested the change $\beta 26 \text{ Glu} \rightarrow \text{Lys}$. The β^X map showed the disappearance of the β -chain peptides βTpII , βTpIII , βTpV and βTpX and the appearance of new δ -chain peptides δTpII , δTpIII , δTpV , δTpX . Since β^X also contained β -chain peptides βTpXIIb and βTpXIII , the β^X chain is most likely a δ - β Lepore hybrid chain, where crossing over between the δ and β genes probably occurred somewhere between amino acid positions 87 and 116. DNA analysis by restriction endonuclease digestion showed an additional 3.8 kb XbaI fragment and 2.6 kb PstI fragment which confirmed the existence of δ - β hybrid chain. It could be concluded that this Thai woman was a double heterozygote for Hb E [$\alpha_2(\beta 26\text{Glu} \rightarrow \text{Lys})_2$] and Hb Lepore-Boston [$\alpha_2(\delta\text{-}\beta)_2$].

This is the first case of Hb Lepore to be reported in Thailand and the first report of Hb Lepore in association with Hb E.



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