Hb E + Alpha-thal.

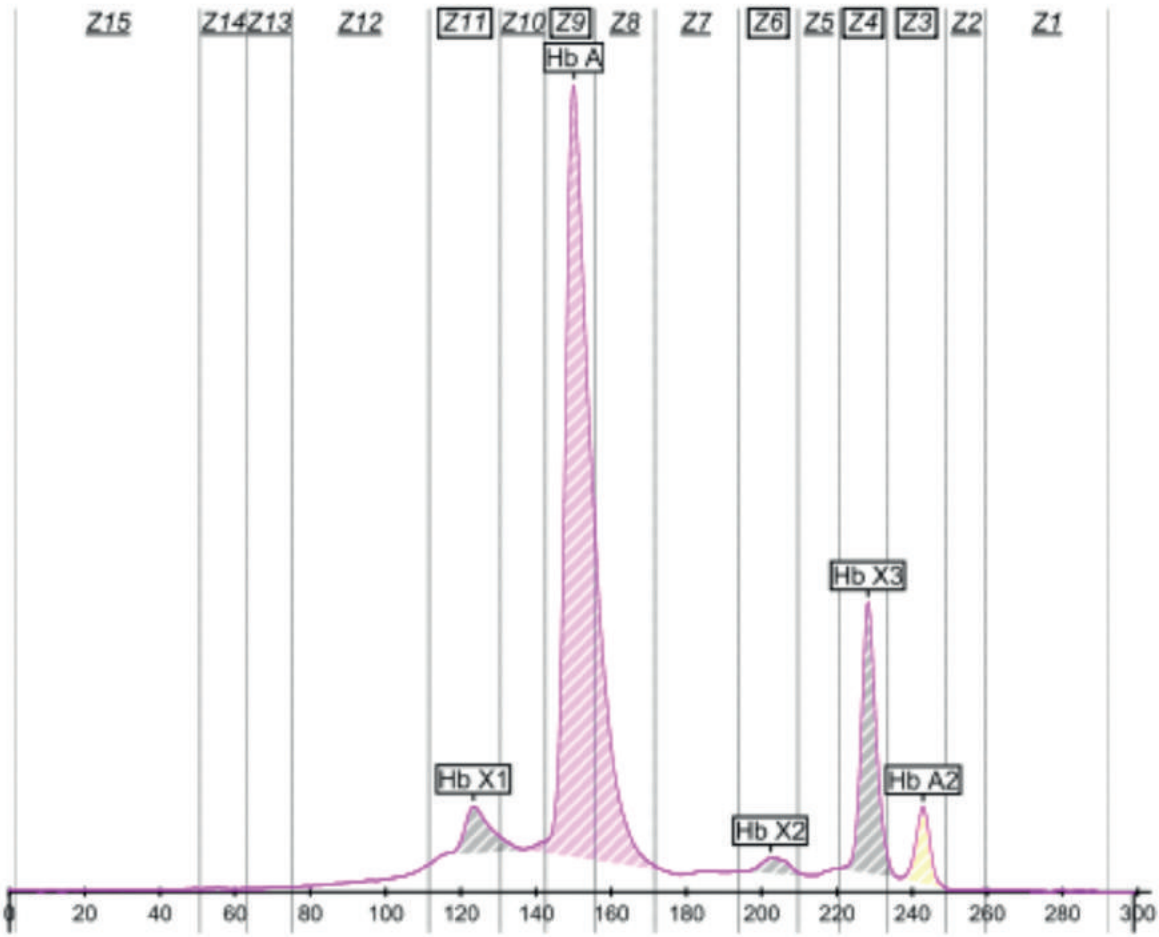
Globin chain(s) involved: **Beta, Alpha**

Status: **Heterozygous Hb E + heterozygous Alpha zero thalassemia**

Migration in zone(s): **Z(E) (=Z4)**

Migration in position(s): **228**

Peak position may vary +/- 1



Z2 = Z(C)
Z3 = Z(A2)
Z4 = Z(E)
Z5 = Z(S)
Z6 = Z(D)
Z7 = Z(F)
Z9 = Z(A)

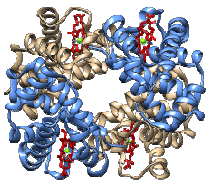
Capillary Electrophoresis

Fractions	Value %
Denat. Hb (Hb X1)	4,4
Hb A	75,6
Denat. Hb (Hb X2)	1,4
Hb E (Hb X3)	14,8
Hb A2	3,8

Comments on this profile: -

Confirmed by amino acid or DNA sequencing

Profile generated with Phoresis Software release < 6.50



Hb E + α-thalassemia

Mutation data

Status:	Heterozygous Hb E + heterozygous Alpha zero thalassemia
Hb E	
Mutation	Beta 26(B8) Glu>Lys
Nomenclature	HBB:c.79G>A
In combination with: **α-thalassemia**	
Mutation	SEA Alpha zero thalassemia deletion Heterozygous (--/αα)
Nomenclature	

Comments:

Hematology

Hematological parameters	Results
RBC	Normal or elevated
Hemoglobin	Low
Hematocrit	Low
MCV	Low

Hematological parameters	Results
MCH	Low
Blood smear	Thalassemic smear
Serum iron and ferritin	

Comments on hematology:	Microcytic hypochromic

Other information

Clinical context:	
Clinical presentation	Mild chronic anemia symptoms
Genetic risk	Mild risk in Homozygous Hb E but intermediate to severe risk in combination with Hb S, all Beta-thalassemia defects, Hb Lepore and other less common hemoglobin variants. Risk for Hb H disease or hydrops fetalis with Alpha + or Alpha zero thalassemic partner
Advice	Partner and family analysis
About this variant:	
Stability	Midly unstable, sensitive to oxidative stress
Oxygen affinity	Normal
Found in	Common combination in South East Asians

Comments:

References:

Kazazian HH Jr. et al., Am J Hum Genet. 1984 Jan;36(1):212-7.
Nakatsuji T. et al., Am J Hum Genet. 1986 Jun;38(6):981-3.
Indrak K., Ann Hematol. 1991 Jul;63(1):42-4.