Hb E + Beta-thal.

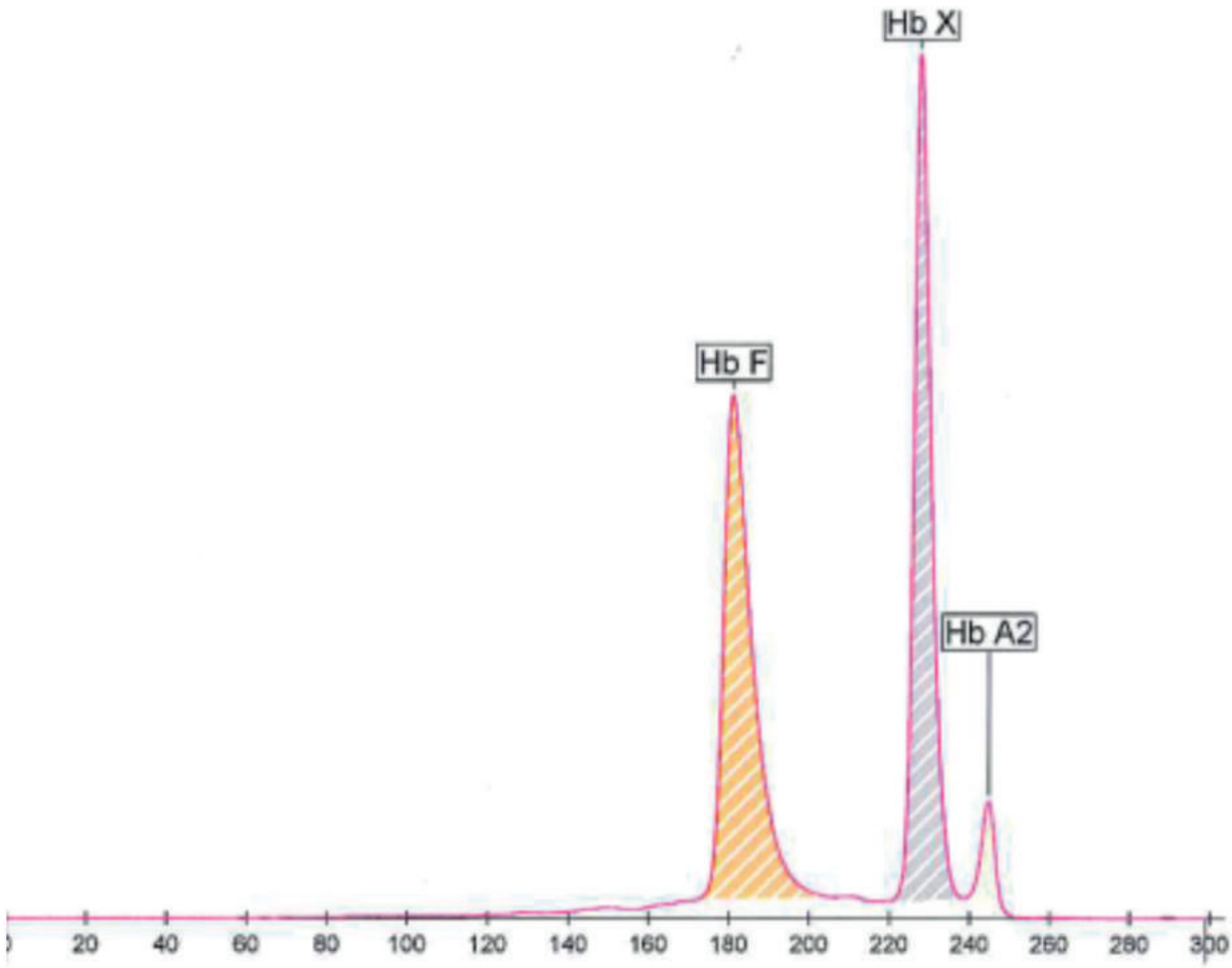
Globin chain(s) involved: **Beta**

Status: **Compound heterozygous**

Migration in zone(s): **-**

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

Peak position may vary +/- 1



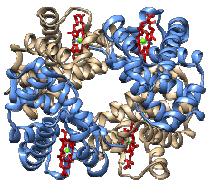
Capillary Electrophoresis

Fractions	Value %
Hb F	47,3
Hb E (Hb X)	47,9
Hb A2	4,8

Comments on this profile: Hb A2 elevated as expected for Beta-thalassemia.
In case of absence of Hb A and/or Hb A2, analyse the sample mixed with Normal Hb A2 Control to display the zones (see Package Insert).
Values of each fractions must be taken on the native sample.

Confirmed by amino acid or DNA sequencing

Profile generated with Phoresis Software release < 6.50



Hb E + β-thalassemia

Mutation data

Status:	Compound heterozygous
Hb E	
Mutation	Beta 26(B8) Glu>Lys
Nomenclature	HBB:c.79G>A
In combination with: **β-thalassemia**	
Mutation	One of the many described Beta gene defects reported on http://globin.cse.psu.edu/hbvar/menu.html
Nomenclature	

Comments:

Hematology

Hematological parameters	Results
RBC	
Hemoglobin	Low
Hematocrit	Low
MCV	Low

Hematological parameters	Results
MCH	Low
Blood smear	Severe thalassemic smear
Serum iron and ferritin	

Comments on hematology:	Severely microcytic hypochromic. Hemolysis

Other information

Clinical context:	
Clinical presentation	Severe hemolytic anemia symptoms
Genetic risk	Severe risk in combination with Hb S, Hb E, Beta-thalassemia, Hb Lepore and other Beta-thalassemic variants
Advice	Partner and family analysis
About this variant:	
Stability	Midly unstable, sensitive to oxidative stress
Oxygen affinity	Normal
Found in	Observed mainly in populations from South East Asia, along the Silk Road and the Middle East

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.