Hb S + Beta-thal.

Patient under transfusion therapy. Profile was done just before exchange transfusion

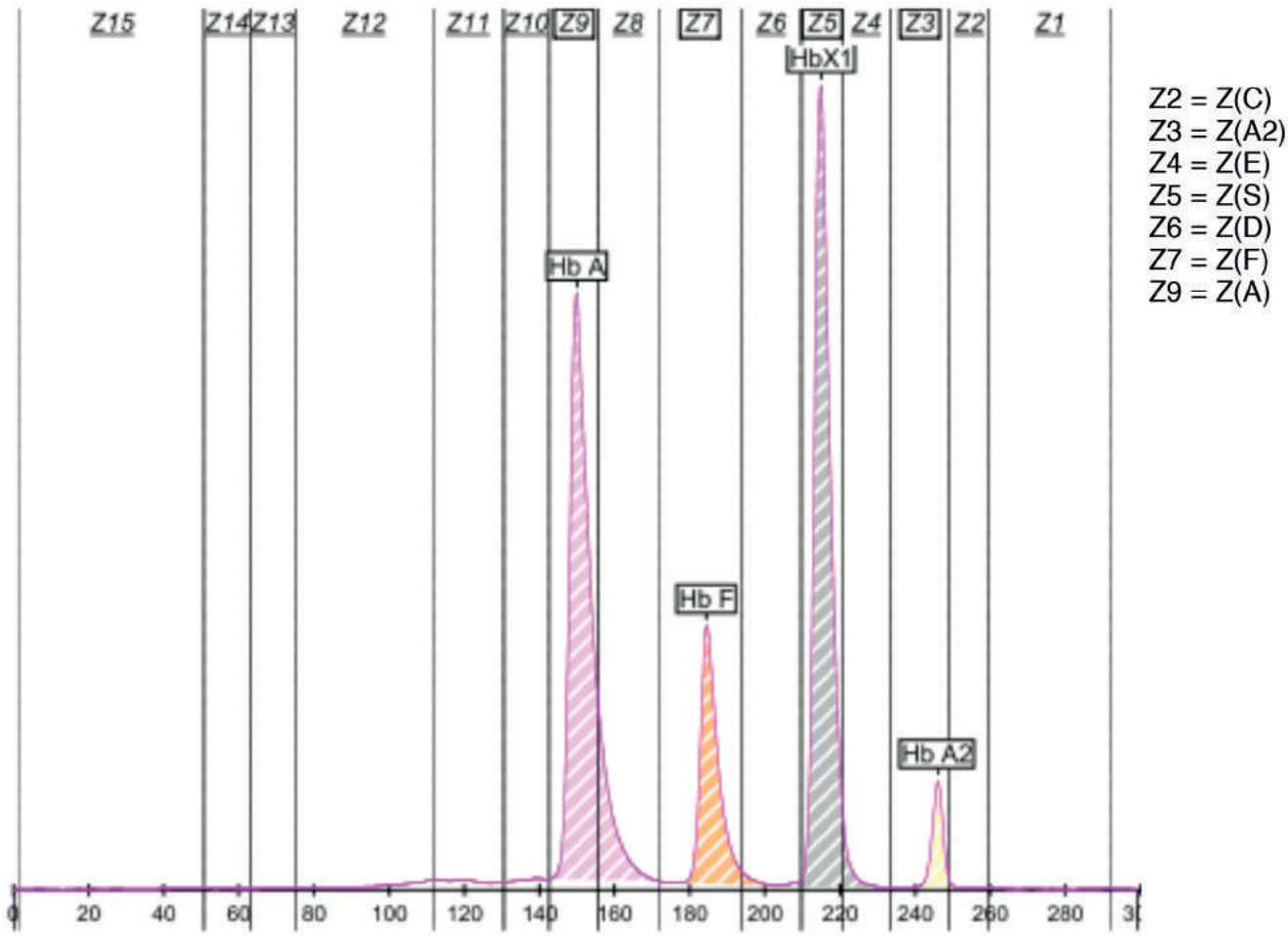
Globin chain(s) involved: **Beta**

Status: **Compound heterozygous**

Migration in zone(s): **Z(S) (=Z5)**

Migration in position(s): **214**

Peak position may vary +/- 1



Capillary Electrophoresis

Fractions	Value %
Hb A	40,4
Hb F	14,6
Hb S (Hb X1)	41,3
Hb A2	3,7

Comments on this profile: Hb A comes from the last transfusion. Hb F is elevated. Hb A2 measurement has no clinical significance due to transfusion.

Hb S + Beta-thal.

Patient under transfusion therapy. Profile was done shortly after exchange transfusion

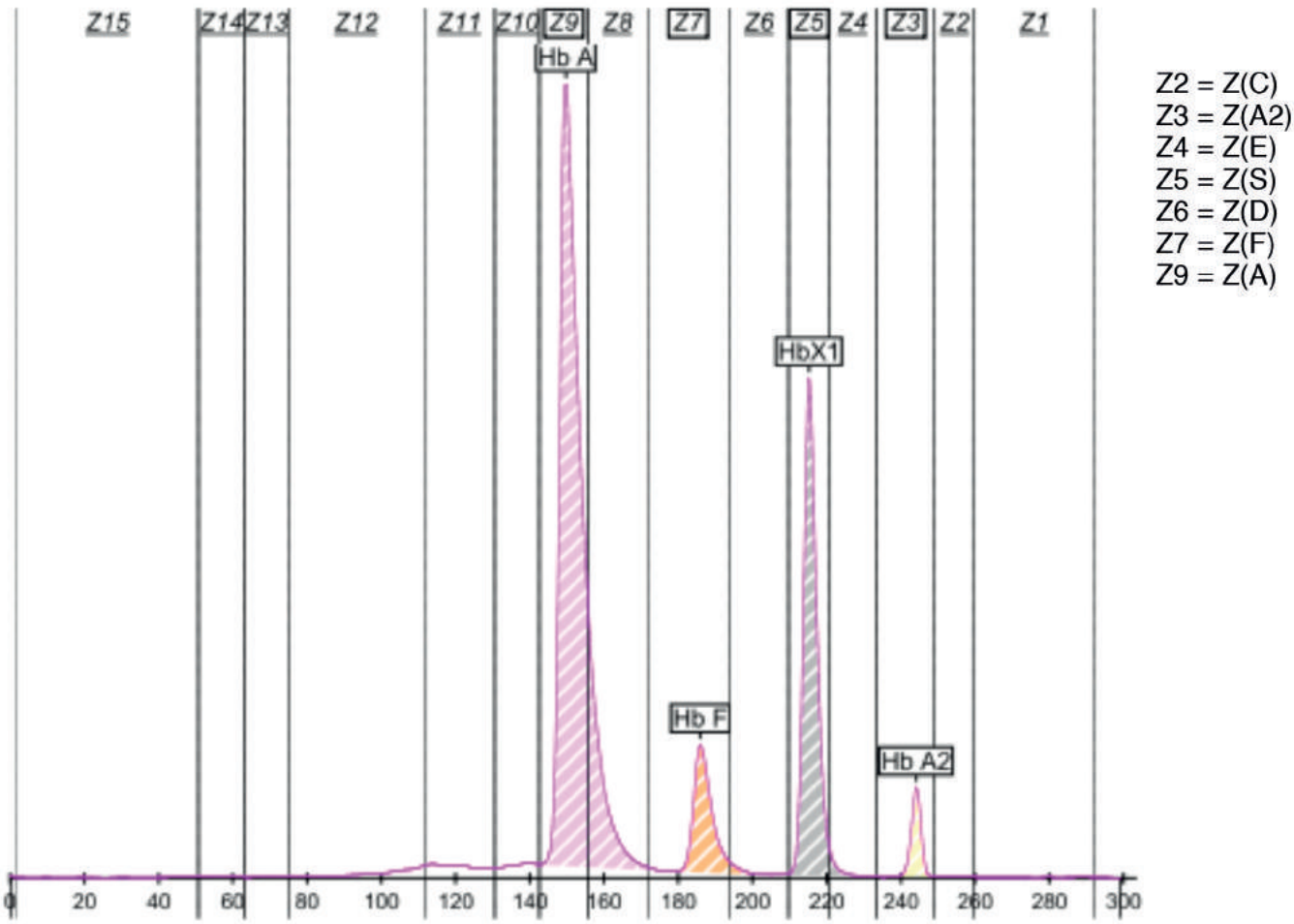
Globin chain(s) involved: **Beta**

Status: **Compound heterozygous**

Migration in zone(s): **Z(S) (=Z5)**

Migration in position(s): **214**

Peak position may vary +/- 1



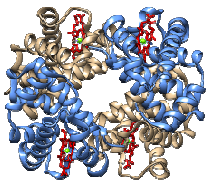
Capillary Electrophoresis

Fractions	Value %
Hb A	63,8
Hb F	8,6
Hb S (Hb X1)	24,4
Hb A2	3,2

Comments on this profile: Hb F and Hb A2 are reduced due to transfusion.

Confirmed by amino acid or DNA sequencing

Profile generated with Phoresis Software release < 6.50



Hb S + β-thalassemia

Patient under transfusion therapy

Mutation data

Status:	Compound heterozygous
Hb S	
Mutation	Beta 6 (A3) Glu>Val
Nomenclature	HBB:c.20A>T
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	
Hematocrit	
MCV	

Hematological parameters	Results
MCH	
Blood smear	Sickle cells
Serum iron and ferritin	

Comments on hematology:	Sickle cell test positive. Due to transfusion, hematological parameters should not be taken into account.

Other information

Clinical context:	
Clinical presentation	No data
Genetic risk	Severe risk in combination with Hb S, Hb C, Hb E, Hb D, Beta-thalassemia, Hb Lepore and other less common hemoglobin variants
Advice	Partner and family analysis
About this variant:	
Stability	No data
Oxygen affinity	No data
Found in	Black, Asian, Mediterranean populations

Comments:

References: -