

Hb H

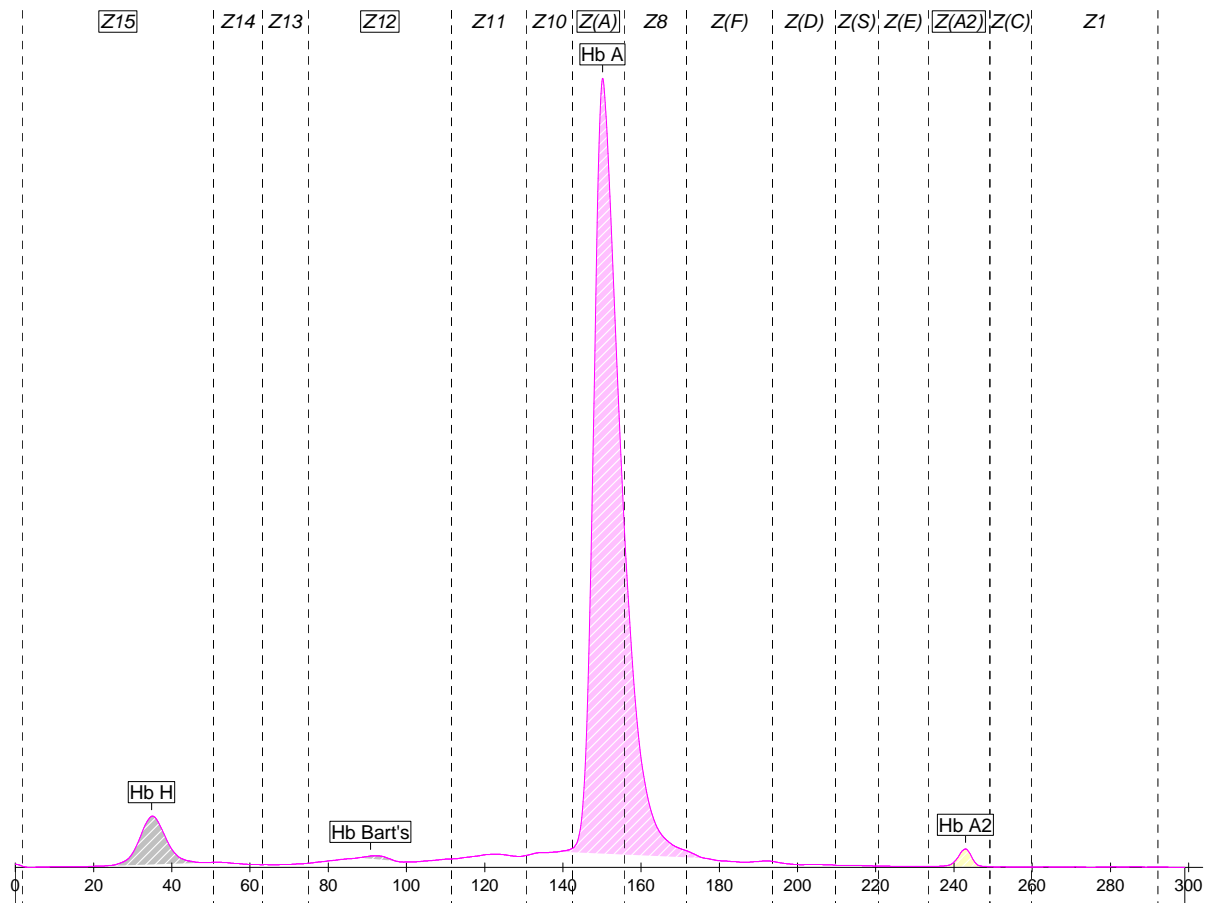
Globin chain(s) involved: **3 Alpha genes deleted**

Status: **Compound heterozygous**

Migration in zone(s): **Z15**

Migration in position(s): **7 to 46**

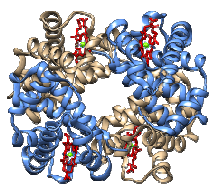
Peak position may vary +/- 1



Capillary Electrophoresis

Fractions	Value %
Hb H	5,6
Hb Bart's	0,3
Hb A	93,1
Hb A2	1,0

Comments on this profile: Hb H is highly unstable and can disappear in few days.



Hb H

Mutation data

Status:	Compound heterozygous
Hb H	
Mutation	loss of expression of 3 of the 4 Alpha genes due to deletions or point mutations
Nomenclature	
In combination with:	
Mutation	
Nomenclature	

Comments:

Hematology

Hematological parameters	Results
RBC	
Hemoglobin	
Hematocrit	
MCV	

Hematological parameters	Results
MCH	
Blood smear	Hb H inclusion bodies
Serum iron and ferritin	

Comments on hematology:

Microcytic, hemolytic parameters. Strongly positive Inclusion Bodies staining

Other information

Clinical context:	
Clinical presentation	No data
Genetic risk	Hb H or hydrops fetalis with partners carriers of Alpha + or Alpha zero thalassemia defects
Advice	Partner and family analysis. Further investigation at the DNA level for Alpha-thalassemia deletions and point mutations
About this variant:	
Stability	Hb H is very unstable
Oxygen affinity	Hb H binds no oxygen
Found in	Observed mainly in populations from South East Asia, the Mediterranean basin and the Middle East

Comments:

References: -