

Hb D-Punjab + Beta-thal.

(Hb D-Punjab is also called Hb D-Los Angeles, Hb D-Chicago, Hb D-North Carolina, Hb D-Portugal or Hb Oak Ridge)

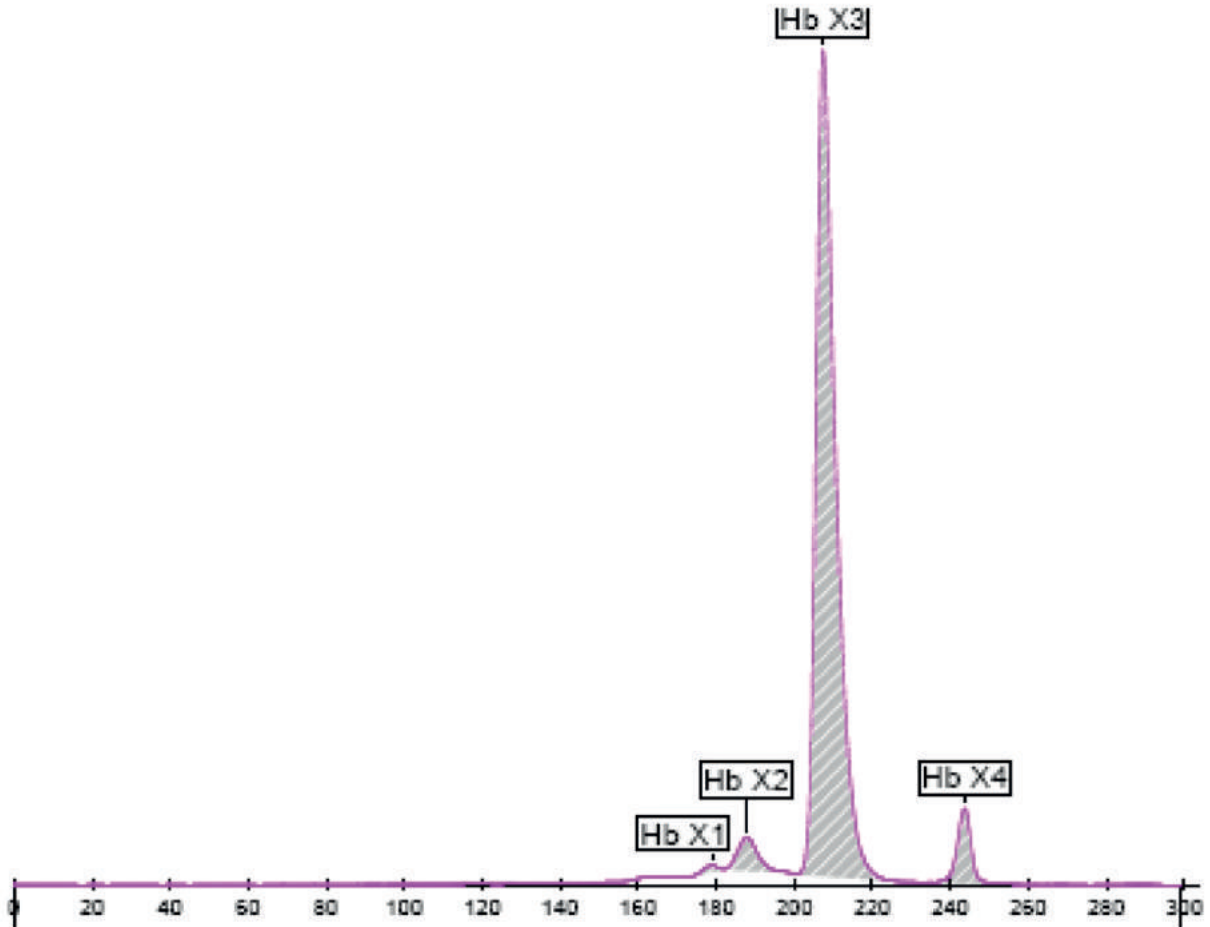
Globin chain(s) involved: **Beta**

Status: **Compound heterozygous**

Migration in zone(s): -

Migration in position(s): -

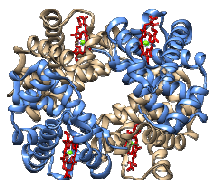
Peak position may vary +/- 1



Capillary Electrophoresis

Fractions	Value %
Denat. Hb (Hb X1)	0,5
Denat Hb (Hb X2)	3,7
Hb D (Hb X3)	90,2
Hb A2 (Hb X4)	5,6

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.



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Mutation data

Status:	Compound heterozygous
Hb D-Punjab	
Mutation	Beta 121(GH4) Glu>Gln
Nomenclature	HBB:c.364G>C
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	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. Elevated Hb A2

Other information

Clinical context:	
Clinical presentation	Normal
Genetic risk	Combinations Hb S / Hb D and Beta-thalassemia / Beta-thalassemia are severe
Advice	Partner and family analysis
About this variant:	
Stability	Normal
Oxygen affinity	Normal
Found in	While homozygosis for less common hemoglobin variants is rare in non consanguineous couples, compound heterozygosis can be more frequent. Here, a Hb D / Beta-thalassemia combination is common in Asians, Indians and in populations along the Silk Road

Comments:

References: Fucharoen S. et al., Hemoglobin. 2002 Aug;26(3):261-9.