

Hb D-Punjab + Hb "A2-Yialousa"

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

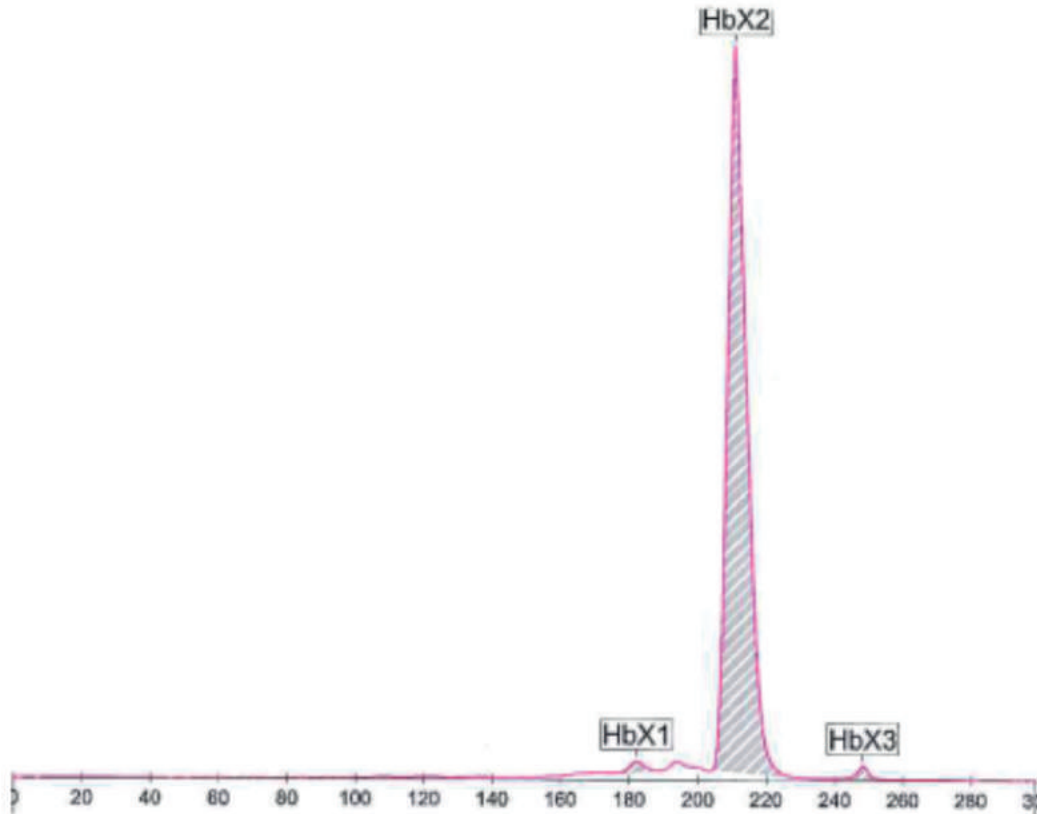
Globin chain(s) involved: **Beta, Delta**

Status: **Double homozygous**

Migration in zone(s): -

Migration in position(s): -

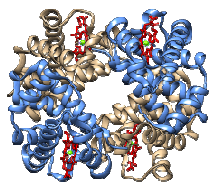
Peak position may vary +/- 1



Capillary Electrophoresis

Fractions	Value %
Denatured Hb (Hb X1)	1,1
Hb D-Punjab (Hb X2)	97,8
Hb "A2-Yialousa" (Hb X3)	1,1

Comments on this profile: Lower Hb A2 level is due to a co-inherited homozygosity for Hb A2-Yialousa, a fairly common Delta thalassemic Hb A2 variant.



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Mutation data

Status:	Double homozygous
Hb D-Punjab	
Mutation	Beta 121(GH4) Glu>Gln
Nomenclature	HBB:c.364G>C
In combination with: Hb A2-Yialousa	
Mutation	Delta 27(B9) Ala>Ser
Nomenclature	HBD:c.82G>T

Comments:

Hematology

Hematological parameters	Results
RBC	
Hemoglobin	
Hematocrit	
MCV	

Hematological parameters	Results
MCH	
Blood smear	
Serum iron and ferritin	

Comments on hematology: Normocytic normochromic

Other information

Clinical context:	
Clinical presentation	Normal
Genetic risk	Combinations of Hb D-Punjab with Hb S are severe
Advice	Partner and family analysis
About this variant:	
Stability	Normal
Oxygen affinity	No data
Found in	Although homozygosis for less common hemoglobin variants is rare, Hb D-Punjab homozygosis may occur in Asians, Indians and populations along the Silk Road due to consanguinity.

Comments:

References: Fucharoen S. et al., Hemoglobin. 2002 Aug;26(3):261-9.
 Moi F. et al., Blood. 1988 Aug;72(2):530-3.
 Loudianos G. et al., Blood. 1990 Jan 15;75(2):526-8.