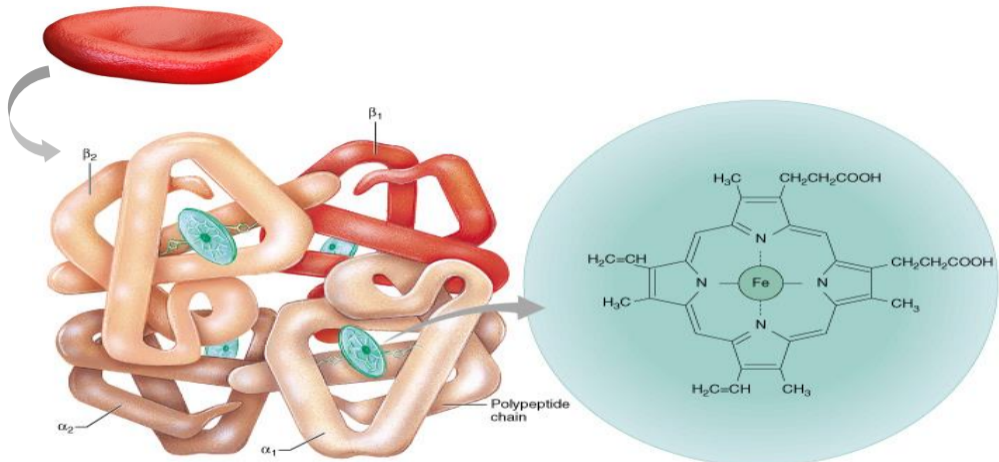


First Observation of Hemoglobin Hamilton in Turkey

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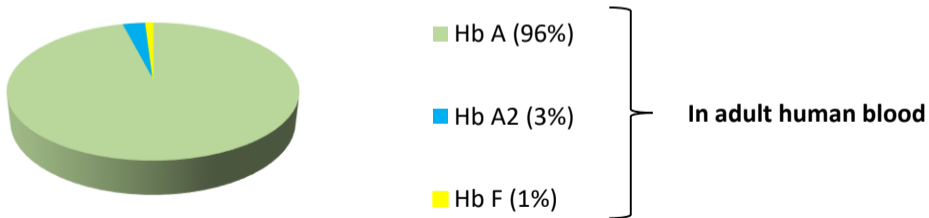


(a) Hemoglobin

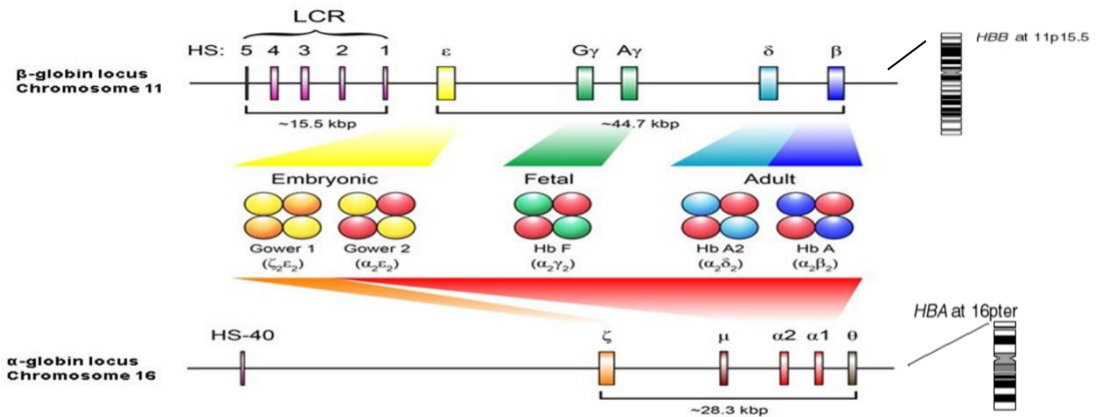
(b) Iron-containing heme group

Structure of Hemoglobin Molecule

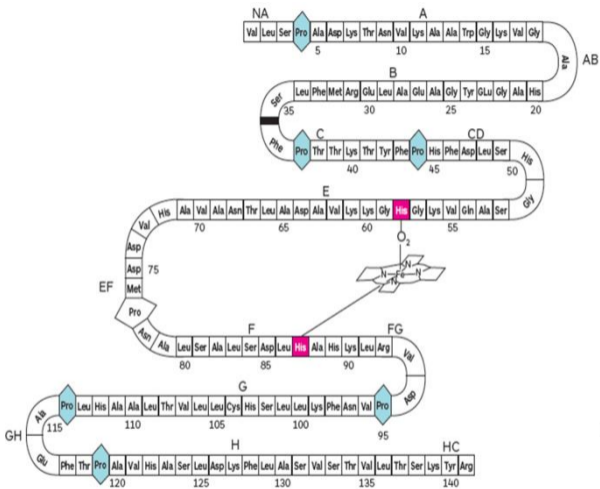
Embryonic Hemoglobin	Fetal Hemoglobin	Adult Hemoglobin
Hb Gower 1 ($\zeta_2\varepsilon_2$)	Hb F ($\alpha_2\gamma_2$)	Hb A ($\alpha_2\beta_2$)
Hb Gower 2 ($\alpha_2\varepsilon_2$)		Hb A2 ($\alpha_2\delta_2$)
Hb Portland ($\zeta_2\gamma_2$)		



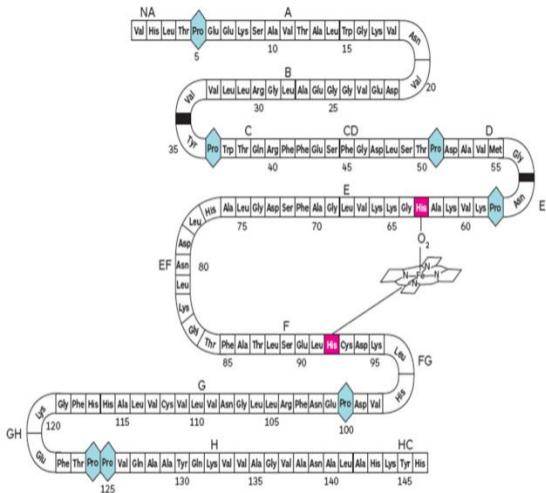
Normal Human Hemoglobin



Alpha and Beta Like Globin Gene Clusters



Alpha Globin Chain



Beta Globin Chain

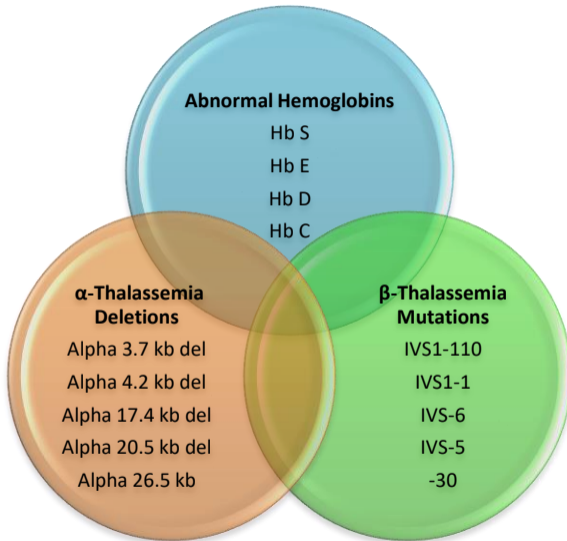
HEMOGLOBINOPATHIES

Hemoglobin variants

- α variants
- β variants
- γ variants
- δ variants

Thalassemia

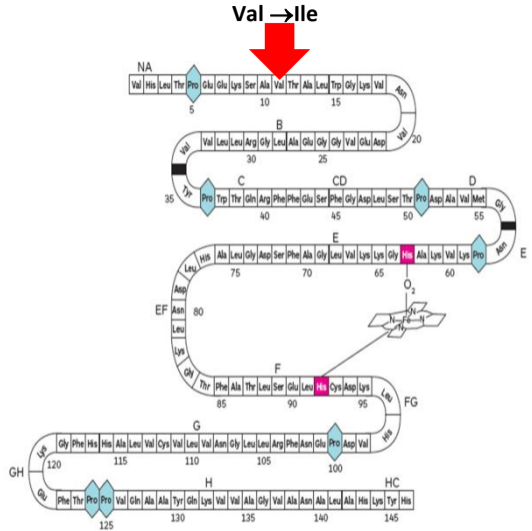
- α-Thalassemia
- β-Thalassemia



No	Name	Chain/Mutation	No	Name	Chain/Mutation
1	Adana*	α59 Gly>Asp	31	J-Antakya*	β65 Lys>Met
2	Ankara*	β10 Ala>Asp	32	J-Iran	β 77 His>Asp
3	A2 Yialousa	δ 82 Ala>Ser	33	J-Meerut	α 120 Ala>Glu
4	Antalya*	Deletion/insertion	34	Knosos	β 27 Val>Ser
5	Başkent*	γ 128 Ala>Thr	35	Köln	β 98 Val>Met
6	Beograd	β 121 Glu>Val	36	Lepore-Boston	Hybrid
7	Brocton	β 138 Ala>Pro	37	M-Iwate	α 87 His>Tyr
8	Bronova	α 103 His>Leu	38	Moabit	α 86 Leu>Arg
9	C	β 6 Glu>Lys	39	Monthgomery	α 48 Leu>Arg
10	City of Hope	β 69 Gly>Ser	40	M-Saskatoon	β 63 His>Tyr
11	Costant Spring	Prolonged α chain	41	N-Baltimore	β 95 Lys>Glu
12	Crete	β 129 Ala>Pro	42	Noah	δ 143 His>Tyr
13	Çapa*	α94 Asp>Gly	43	O-Arab	β 121 Glu>Lys
14	D-Iran	β 22 Glu>Gln	44	O-Podova	α 30 Glu>Lys
15	D-Punjab	β 121 Glu>Gln	45	P-Nilotic	Hybrid
16	D-Ouled Rabah	β 19 Asn>Lys	46	Pyrgos	β 83 Gly>Asp
17	E	β 26 Glu>Lys	47	Q-Iran	α 75 Asp>His
18	Ernz	β 123 Thr>Asn	48	S	β 6 Glu>Val
19	E-Saskatoon	β 22 Glu>Lys	49	Sarrebourg	β 131 Gln>Arg
20	G-Copenhagen	β 47 Asp>Asn	50	Setif	α 94 Asp>His
21	G-Cousatta	β 22 Glu>Ala	51	Siirt*	β27 Ala>Gly
22	G-Georgia	α 95 Pro>Leu	52	South Florida	β 1 Val>Met
23	G-Szuhu	β 80 Asn>Lys	53	Strumica	α 112 His>Arg
24	Hakkari*	β31 Leu>Arg	54	Summer Hill	β 52 Asp>His
25	Hamadan	β 56 Gly>Arg	55	Tunis	β 124 Pro>Ser
26	Hamilton	β 11 Val>Ile	56	Tyne	β 5 Pro>Ser
27	Hasharon	α 47 Asp>His	57	Ube-2	α 68 Asn>Asp
28	Istanbul*	β92 His>Gln	58	Volga	β 27 Ala>Asp
29	Izmir*	β86 Ala>Val	59	Yauzi	β 79 Asp>Asn
30	J-Anatolia	α 61 Lys>Thr	60	Westeinde	α 125 Leu>Gln

*Discovered in Turkish Patients

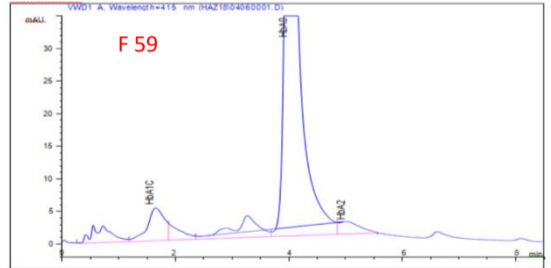
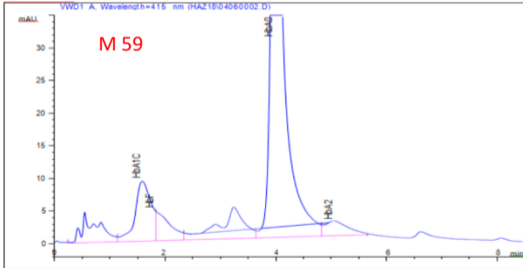
- Hb Hamilton, beta 11(A8)Val→Ile, is a silent mutation and the substitution of isoleucine for valine in the 11th position of the beta chain.
- This mutation does not change the function of the hemoglobin molecule.
- The variant not detectable by cellulose acetate, starch or agar gel electrophoresis and HPLC.



Hb Hamilton

Sex/Age	RBC 10 ¹² /L	Hb g/dL	MCV fL	MCH pg	MCHC g/dL	HbA ₂ %
M59	6.51 ↑	12.7 ↓	63.0 ↓	19.5 ↓	31.0	3.5 ↑
F59	4.36	13.0	97.7	29.8	30.5	2.4

Hematological data



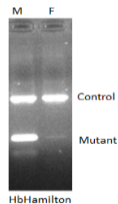
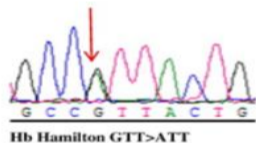
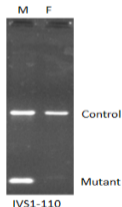
Hemoglobin variants on HPLC

661 cagtgccaga agagccaagg acaggtacgg ctgtcatcac ttagacctca cctgtggag
 721 ccacacccta gggttggcca atctactccc aggagcaggg agggcaggag ccagggctgg
 781 gcataaaagt cagggcagag ccattctattg cttacatttg cttctgacac aactgtgttc

841 actagcaacc tcaaacagac acc**ATGGTGC** **ACCTGACTCC** **TGAGGAGAAG** **TCTGCCGTTA**
901 CTGCCCTGTG **GGGCAAGGTG** **AACGTGGATG** **AAGTTGGTGG** **TGAGGCCCTG** **GGCAG**gttgg
 961 **tatcaaggtt** acaagacagg tttaaggaga ccaatagaaa ctgggcatgt ggagacagag

IVS1-110 (g>a)

1021 aagactcttg ggtttctgat aggcactgac tctctctgcc tattggtcta ttttcccacc
 1081 cttag**GCTGC** **TGGTGGTCTA** **CCCTTGGACC** **CAGAGGTTCT** **TTGAGTCCTT** **TGGGGATCTG**
1141 TCCACTCCTG **ATGCTGTTAT** **GGGCAACCCT** **AAGGTGAAGG** **CTCATGGCAA** **GAAAGTGCTC**
1201 GGTGCCTTTA **GTGATGGCCT** **GGCTCACCTG** **GACAACCTCA** **AGGGCACCTT** **TGCCACACTG**
1261 AGTGAGCTGC **ACTGTGACAA** **GCTGCACGTG** **GATCCTGAGA** **ACTTCAGG**gt gagtctatgg
 1321 gacccttgat gttttctttc cccttctttt ctatgggtaa gttcatgtca taggaagggg
 1381 agaagtaaca gggtacagtt tagaatggga aacagacgaa tgattgcatc agtgtggaag
 1441 tctcaggatc gtttttagttt cttttatttg ctgttcataa caattgtttt cttttgttta



Hb Hamilton Mutation Point

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Thank you for attention and participation...