Section1

Q1: Nucleotide to nucleotide.

Q2:

Homo sapiens hemoglobin subunit beta (HBB), mRNA 854 854 100% 0.0 99% NM\_000518.4

Select seq XM\_508242.4 PREDICTED: Pan troglodytes hemoglobin subunit beta (HBB), mRNA 848 848 100% 0.0 99% XM\_508242.4

Select seq XM\_003819029.2 PREDICTED: Pan paniscus hemoglobin subunit beta (LOC100976465), mRNA 848 848 100% 0.0 99% XM\_003819029.2

Select seq AY136510.1 Homo sapiens hemoglobin beta chain variant Hb S-Wake (HBB) mRNA, complete cds 848 848 100% 0.0 99% AY136510.1

Q3: 99%

Q4:

Top hit: 466/468

Lowest hit: 409/464

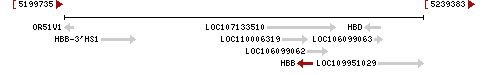
Q5:

Official Symbol: HBB

Official Full Name: hemoglobin subunit beta

Q6: Chromosome 11.

Q7:



Q8: 3 exons, 2 introns.

Q9:

GeneRIFs: Gene References Into FunctionsWhat's a GeneRIF?

Upon oxygenation, changes in the conformational flexibility across different regions of alpha and beta globin chains in the tetrameric HbS molecule were investigated. It was observed that oxygenation led to perturbation in the conformation of several residues around the hydrophobic patch, groove of a tetramer and axial, lateral contacts across the double strands that are involved in HbS polymerization.

Vaginal cell-derived Hb-alpha and Hb-beta may have antioxidative properties and protect cells from oxidative stress-induced inflammation possibly through Nrf2 downstream signaling.

a novel, positive role for PKR activation and eIF2alpha phosphorylation in human globin mRNA splicing, is reported.

human bone marrow stromal cells-derived induced pluripotent stem sacs allow for more efficient erythroid cell generation with higher beta-globin production, likely due to heightened emergence of immature progenitors.

In this paper, the experimental results of resonance Raman, terahertz time-domain, and 1H nuclear magnetic resonance spectroscopy for the Hb M are described in detail. The heterogeneity of alpha subunits, which has been observed previously, is reconfirmed, and its implications for the dynamical structure of the alpha2beta2 tetramer and the regulation of O2 affinity are discussed.

study reports 3 novel HBB mutations - one in the promoter (-90 C>G), a 20 bp deletion in exon 2 (FS Cd 78/85) and a mutation in the donor site of intron 2 (IVS2:2 T>G); considering these novel mutations and the first observation of IVS1:6T>C, the molecular spectrum of beta-thalasemia in Mexicans comprises 21 different mutations, confirming the high allelic heterogeneity in Mexicans

Embryonic stem cell sacs serve as hemangioblast-like progenitors capable to generate definitive erythroid cells that express beta-globin.

Data suggest that, in crystal form of Hb containing three protein molecules--Hbalpha(1)beta(2), allosteric effectors (phosphate, bezafibrate) induce conformational changes in direction from relaxed-state to tense-state; binding of each effector energetically stabilizes lowest affinity conformer more strongly than intermediate affinity conformer; minor Hb conformers have disproportionate effects on overall O(2) affinity.

In the presence of free alpha subunits and H2O2, both HbA and HbE showed bCys93 oxidation which increased with higher H2O2 concentrations. In the presence of Alpha-hemoglobin stabilizing protein (AHSP)Cys93 oxidation was substantially reduced in both proteins.in the presence of excess free alpha-subunit and under the same oxidative conditions, these events are substantially increased for HbE compared to HbA

This study presents comparison of two neutron structures of hemoglobins, human deoxyhemoglobin (T state) and equine cyanomethemoglobin (R state) and offers a direct observation of histidine residues that are likely to contribute to the Bohr effect.

Q10:

Associated conditions

| Description | Tests |
| --- | --- |
| [alpha Thalassemia](https://www.ncbi.nlm.nih.gov/gtr/conditions/C0002312)  MedGen: [C0002312](https://www.ncbi.nlm.nih.gov/medgen/C0002312) OMIM: [604131](https://www.ncbi.nlm.nih.gov/omim/604131) GeneReviews: [Alpha-Thalassemia](https://www.ncbi.nlm.nih.gov/books/NBK1435) | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C0002312%5bDISCUI%5d&condition=C0002312&compare_labs=1) |
| [beta Thalassemia](https://www.ncbi.nlm.nih.gov/gtr/conditions/C0005283)  MedGen: [C0005283](https://www.ncbi.nlm.nih.gov/medgen/C0005283) OMIM: [613985](https://www.ncbi.nlm.nih.gov/omim/613985) GeneReviews: [Beta-Thalassemia](https://www.ncbi.nlm.nih.gov/books/NBK1426) | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C0005283%5bDISCUI%5d&condition=C0005283&compare_labs=1) |
| [Beta-thalassemia, dominant inclusion body type](https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858990)  MedGen: [C1858990](https://www.ncbi.nlm.nih.gov/medgen/C1858990) OMIM: [603902](https://www.ncbi.nlm.nih.gov/omim/603902) GeneReviews: Not available | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C1858990%5bDISCUI%5d&condition=C1858990&compare_labs=1) |
| [Fetal hemoglobin quantitative trait locus 1](https://www.ncbi.nlm.nih.gov/gtr/conditions/C1841621)  MedGen: [C1841621](https://www.ncbi.nlm.nih.gov/medgen/C1841621) OMIM: [141749](https://www.ncbi.nlm.nih.gov/omim/141749) GeneReviews: Not available | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C1841621%5bDISCUI%5d&condition=C1841621&compare_labs=1) |
| [Hb SS disease](https://www.ncbi.nlm.nih.gov/gtr/conditions/C0002895)  MedGen: [C0002895](https://www.ncbi.nlm.nih.gov/medgen/C0002895) OMIM: [603903](https://www.ncbi.nlm.nih.gov/omim/603903) GeneReviews: [Sickle Cell Disease](https://www.ncbi.nlm.nih.gov/books/NBK1377) | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C0002895%5bDISCUI%5d&condition=C0002895&compare_labs=1) |
| [Heinz body anemia](https://www.ncbi.nlm.nih.gov/gtr/conditions/C0700299)  MedGen: [C0700299](https://www.ncbi.nlm.nih.gov/medgen/C0700299) OMIM: [140700](https://www.ncbi.nlm.nih.gov/omim/140700) GeneReviews: Not available | [Compare labs](https://www.ncbi.nlm.nih.gov/gtr/tests/?term=C0700299%5bDISCUI%5d&condition=C0700299&compare_labs=1) |
| [Susceptibility to malaria](https://www.ncbi.nlm.nih.gov/gtr/conditions/C1970028)  MedGen: [C1970028](https://www.ncbi.nlm.nih.gov/medgen/C1970028) OMIM: [611162](https://www.ncbi.nlm.nih.gov/omim/611162) GeneReviews: Not available |  |

Section2:

Q11: Accession: NM\_000518.4

Q12: exon1: 1-142

Q13:51-494

Section3:

Q14: 50 (not coding)

Q15:2

Q16:70/20 and 212/162

GAG vs GUG: yes. Glutamic acid to valine

GCA vs GCT: no. silent mutation

Q17: yes. 1HBS

Q18: Very close

Section 5

Gene 1:

Homo sapiens peroxisome proliferator activated receptor gamma (PPARG)

Gene2:

Homo sapiens potassium voltage-gated channel subfamily H member 2 (KCNH2), transcript variant 3, mRNA