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<pre class="genbank">LOCUS          HUMHBB          73308 bp    DNA    linear    PRI 16-OCT-2008
DEFINITION Human beta globin region on chromosome 11.
ACCESSION  U01317 J00093 J00094 J00096 J00158-J00175 J00177 J00178 J00179
            K01239 K01890 K02544 M18047 M19067 M24868 M24886
VERSION    U01317.1
KEYWORDS   Alu repeat; HPFH; KpnI repetitive sequence; RNA polymerase III;
            allelic variation; alternate cap site; beta-1 pseudogene;
            beta-globin; delta-globin; epsilon-globin; gamma-globin; gene
            duplication; globin; polymorphism; promoter mutation; pseudogene;
            repetitive sequence; thalassemia.
SOURCE     Homo sapiens (human)
  ORGANISM <a href="/Taxonomy/Browser/wwwtax.cgi?id=9606">Homo sapiens</a>
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE  1 (bases 62409 to 62631; 63482 to 63610)
  AUTHORS  Marotta,C.A., Forget,B.G., Weissman,S.M., Verma,I.M.,
            McCaffrey,R.P. and Baltimore,D.
  TITLE    Nucleotide sequences of human globin messenger RNA
  JOURNAL  Proc. Natl. Acad. Sci. U.S.A. 71 (6), 2300-2304 (1974)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/4135409">4135409</a>
REFERENCE  2 (bases 63602 to 63646)
  AUTHORS  Forget,B.G., Marotta,C.A., Weissman,S.M. and Cohen-Solal,M.
  TITLE    Nucleotide sequences of the 3'-terminal untranslated region of
            messenger RNA for human beta globin chain
  JOURNAL  Proc. Natl. Acad. Sci. U.S.A. 72 (9), 3614-3618 (1975)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/1059150">1059150</a>
REFERENCE  3 (bases 63593 to 63626)
  AUTHORS  Proudfoot,N.J. and Brownlee,G.G.
  TITLE    Nucleotide sequences of globin messenger RNA
  JOURNAL  Br. Med. Bull. 32 (3), 251-256 (1976)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/788834">788834</a>
REFERENCE  4 (bases 63673 to 63743)
  AUTHORS  Proudfoot,N.J. and Longley,J.I.
  TITLE    The 3' terminal sequences of human alpha and beta globin messenger
            RNAs: comparison with rabbit globin messenger RNA
  JOURNAL  Cell 9 (4 PT 2), 733-746 (1976)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/1035137">1035137</a>
REFERENCE  5 (sites)
  AUTHORS  Proudfoot,N.J. and Brownlee,G.G.
  TITLE    3' non-coding region sequences in eukaryotic messenger RNA
  JOURNAL  Nature 263 (5574), 211-214 (1976)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/822353">822353</a>
REFERENCE  6 (bases 63596 to 63743)
  AUTHORS  Proudfoot,N.J.
  TITLE    Complete 3' noncoding region sequences of rabbit and human
            beta-globin messenger RNAs
  JOURNAL  Cell 10 (4), 559-570 (1977)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/67897">67897</a>
REFERENCE  7 (bases 62137 to 62193)
  AUTHORS  Baralle,F.
  TITLE    Complete nucleotide sequence of the 5' noncoding region of human
            alpha-and beta-globin mRNA
  JOURNAL  Cell 12 (4), 1085-1095 (1977)
  PUBMED  <a href="https://www.ncbi.nlm.nih.gov/pubmed/597858">597858</a>

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Weissman,S.M.
TITLE Human beta-globin messenger RNA. I. Nucleotide sequences derived from complementary RNA
JOURNAL J. Biol. Chem. 252 (14), 5019-5031 (1977)
PUBMED [873928](https://www.ncbi.nlm.nih.gov/pubmed/873928)
REFERENCE 9 (bases 62187 to 63610)
AUTHORS Cohen-Solal,M., Forget,B.G., Prensky,W., Marotta,C.A. and Weissman,S.M.

TITLE Human beta-globin messenger RNA. II. nucleotide sequences derived from 125I-labeled globin messenger RNA
JOURNAL J. Biol. Chem. 252 (14), 5032-5039 (1977)
PUBMED [318159](https://www.ncbi.nlm.nih.gov/pubmed/318159)
REFERENCE 10 (bases 54790 to 54881; 62409 to 62631; 63482 to 63715)
AUTHORS Marotta,C.A., Wilson,J.T., Forget,B.G. and Weissman,S.M.

TITLE Human beta-globin messenger RNA. III. Nucleotide sequences derived from complementary DNA
JOURNAL J. Biol. Chem. 252 (14), 5040-5053 (1977)
PUBMED [68958](https://www.ncbi.nlm.nih.gov/pubmed/68958)
REFERENCE 11 (bases 62137 to 62189)
AUTHORS Chang,J.C., Temple,G.F., Poon,R., Neumann,K.H. and Kan,Y.W.
TITLE The nucleotide sequences of the untranslated 5' regions of human alpha- and beta-globin mRNAs
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 74 (11), 5145-5149 (1977)
PUBMED [270752](https://www.ncbi.nlm.nih.gov/pubmed/270752)
REFERENCE 12 (bases 56103 to 56184)
AUTHORS Lawn,R.M., Fritsch,E.F., Parker,R.C., Blake,G. and Maniatis,T.
TITLE The isolation and characterization of linked delta- and beta-globin genes from a cloned library of human DNA
JOURNAL Cell 15 (4), 1157-1174 (1978)
PUBMED [728996](https://www.ncbi.nlm.nih.gov/pubmed/728996)
REFERENCE 13 (bases 35854 to 35946; 63482 to 63531)
AUTHORS Little,P., Curtis,P., Coutelle,C., Van den Berg,J., Dalgleish,R., Malcolm,S., Courtney,M., Westaway,D. and Williamson,R.
TITLE Isolation and partial sequence of recombinant plasmids containing human alpha-, beta- and gamma-globin cDNA fragments
JOURNAL Nature 273 (5664), 640-643 (1978)
PUBMED [318161](https://www.ncbi.nlm.nih.gov/pubmed/318161)
REFERENCE 14 (bases 62187 to 63742)
AUTHORS Wilson,J.T., Wilson,L.B., deRiel,J.K., Villa-komaroff,L., Efstratiadis,A., Forget,B.G. and Weissman,S.M.
TITLE Insertion of synthetic copies of human globin genes into bacterial plasmids
JOURNAL Nucleic Acids Res. 5 (2), 563-581 (1978)
PUBMED [345245](https://www.ncbi.nlm.nih.gov/pubmed/345245)
REFERENCE 15 (bases 34478 to 34563; 39414 to 39499)
AUTHORS Chang,J.C., Poon,R., Neumann,K.H. and Kan,Y.W.
TITLE The nucleotide sequence of the 5' untranslated region of human gamma-globin mRNA
JOURNAL Nucleic Acids Res. 5 (10), 3515-3522 (1978)
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REFERENCE 16 (bases 40839 to 40985)
AUTHORS Poon,R., Kan,Y.W. and Boyer,H.W.
TITLE Sequence of the 3'-noncoding and adjacent coding regions of human gamma-globin mRNA
JOURNAL Nucleic Acids Res. 5 (12), 4625-4630 (1978)
PUBMED [318163](https://www.ncbi.nlm.nih.gov/pubmed/318163)
REFERENCE 17 (bases 35826 to 35907; 40742 to 40823)
AUTHORS Smithies,O., Blechl,A.E., Denniston-Thompson,K., Newell,N., Richards,J.E., Slightom,J.L., Tucker,P.W. and Blattner,F.R.
TITLE Cloning human fetal gamma globin and mouse alpha-type globin DNA: characterization and partial sequencing
JOURNAL Science 202 (4374), 1284-1289 (1978)
PUBMED [725604](https://www.ncbi.nlm.nih.gov/pubmed/725604)

TITLE Nucleotide sequences of the untranslated 5' and 3' regions of human alpha-, beta-, and gamma-globin mRNAs

JOURNAL (in) Stamatoyannopoulos, G. and Newhouse, A.W. (Eds.); CELLULAR AND MOLECULAR REGULATION OF HEMOGLOBIN SWITCHING: 0-0; Grune and Stratton, New York (1979) In press

REFERENCE 19 (sites)

AUTHORS Chang, J.C. and Kan, Y.W.

TITLE beta 0 thalassemia, a nonsense mutation in man

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 76 (6), 2886-2889 (1979)

PUBMED [88735](https://www.ncbi.nlm.nih.gov/pubmed/88735)

REFERENCE 20 (bases 19846 to 19965)

AUTHORS Proudfoot, N.J. and Baralle, F.E.

TITLE Molecular cloning of human epsilon-globin gene

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 76 (11), 5435-5439 (1979)

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REFERENCE 21 (sites)

AUTHORS Chang, J.C., Kan, Y.W., Trecartin, R.F. and Temple, G.F.

TITLE Nonsense mutation as a cause of beta 0 thalassemia

JOURNAL Ann. N. Y. Acad. Sci. 344, 113-119 (1980)

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REFERENCE 22 (sites)

AUTHORS Fritsch, E.F., Lawn, R.M. and Maniatis, T.

TITLE Molecular cloning and characterization of the human beta-like globin gene cluster

JOURNAL Cell 19 (4), 959-972 (1980)

PUBMED [6155216](https://www.ncbi.nlm.nih.gov/pubmed/6155216)

REFERENCE 23 (bases 19462 to 21741)

AUTHORS Baralle, F.E., Shoulders, C.C. and Proudfoot, N.J.

TITLE The primary structure of the human epsilon-globin gene

JOURNAL Cell 21 (3), 621-626 (1980)

PUBMED [6254663](https://www.ncbi.nlm.nih.gov/pubmed/6254663)

REFERENCE 24 (bases 34422 to 36069; 39358 to 40986)

AUTHORS Slightom, J.L., Blechl, A.E. and Smithies, O.

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JOURNAL Cell 21 (3), 627-638 (1980)

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REFERENCE 25 (bases 54618 to 56602)

AUTHORS Spritz, R.A., DeRiel, J.K., Forget, B.G. and Weissman, S.M.

TITLE Complete nucleotide sequence of the human delta-globin gene

JOURNAL Cell 21 (3), 639-646 (1980)

PUBMED [7438204](https://www.ncbi.nlm.nih.gov/pubmed/7438204)

REFERENCE 26 (bases 62034 to 64083)

AUTHORS Lawn, R.M., Efstratiadis, A., O'Connell, C. and Maniatis, T.

TITLE The nucleotide sequence of the human beta-globin gene

JOURNAL Cell 21 (3), 647-651 (1980)

PUBMED [6254664](https://www.ncbi.nlm.nih.gov/pubmed/6254664)

REFERENCE 27 (sites)

AUTHORS Efstratiadis, A., Posakony, J.W., Maniatis, T., Lawn, R.M., O'Connell, C., Spritz, R.A., DeRiel, J.K., Forget, B.G., Weissman, S.M., Slightom, J.L., Blechl, A.E., Smithies, O., Baralle, F.E., Shoulders, C.C. and Proudfoot, N.J.

TITLE The structure and evolution of the human beta-globin gene family

JOURNAL Cell 21 (3), 653-668 (1980)

PUBMED [6985477](https://www.ncbi.nlm.nih.gov/pubmed/6985477)

REFERENCE 28 (bases 19864 to 19983)

AUTHORS Baralle, F.E., Shoulders, C.C., Goodbourn, S., Jeffreys, A. and Proudfoot, N.J.

TITLE The 5' flanking region of human epsilon-globin gene

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REFERENCE 29 (bases 32353 to 43728)

JOURNAL Cell 26 (2 PT 2), 191-203 (1981)
PUBMED 7332928
REFERENCE 30 (sites)
AUTHORS Busslinger,M., Moschonas,N. and Flavell,R.A.
TITLE Beta + thalassemia: aberrant splicing results from a single point mutation in an intron

JOURNAL Cell 27 (2 PT 1), 289-298 (1981)
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REFERENCE 31 (bases 32353 to 33218; 51978 to 52472)
AUTHORS Duncan,C.H., Jagadeeswaran,P., Wang,R.R. and Weissman,S.M.
TITLE Structural analysis of templates and RNA polymerase III transcripts of Alu family sequences interspersed among the human beta-like globin genes

JOURNAL Gene 13 (2), 185-196 (1981)
PUBMED 6165649
REFERENCE 32 (sites)
AUTHORS Orkin,S.H. and Goff,S.C.
TITLE Nonsense and frameshift mutations in beta 0-thalassemia detected in cloned beta-globin genes

JOURNAL J. Biol. Chem. 256 (19), 9782-9784 (1981)
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REFERENCE 33 (bases 62454 to 62613)
AUTHORS Orkin,S.H., Goff,S.C. and Nathan,D.G.
TITLE Heterogeneity of DNA deletion in gamma delta beta-thalassemia

JOURNAL J. Clin. Invest. 67 (3), 878-884 (1981)
PUBMED 6162860
REFERENCE 34 (sites)
AUTHORS Trecartin,R.F., Liebhaber,S.A., Chang,J.C., Lee,K.Y., Kan,Y.W., Furbetta,M., Angius,A. and Cao,A.
TITLE beta zero thalassemia in Sardinia is caused by a nonsense mutation

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REFERENCE 35 (sites)
AUTHORS Westaway,D. and Williamson,R.
TITLE An intron nucleotide sequence variant in a cloned beta +-thalassaemia globin gene

JOURNAL Nucleic Acids Res. 9 (8), 1777-1788 (1981)
PUBMED 6264391
REFERENCE 36 (sites)
AUTHORS Moschonas,N., de Boer,E., Grosveld,F.G., Dahl,H.H., Wright,S., Shewmaker,C.K. and Flavell,R.A.
TITLE Structure and expression of a cloned beta o thalassaemic globin gene

JOURNAL Nucleic Acids Res. 9 (17), 4391-4401 (1981)
PUBMED 6272205
REFERENCE 37 (bases 60489 to 60948)
AUTHORS Spritz,R.A.
TITLE Duplication/deletion polymorphism 5' - to the human beta globin gene

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PUBMED 7312624
REFERENCE 38 (bases 59345 to 59593)
AUTHORS Miesfeld,R., Krystal,M. and Arnheim,N.
TITLE A member of a new repeated sequence family which is conserved throughout eucaryotic evolution is found between the human delta and beta globin genes

JOURNAL Nucleic Acids Res. 9 (22), 5931-5947 (1981)
PUBMED 6273813
REFERENCE 39 (bases 16595 to 17840)
AUTHORS Di Segni,G., Carrara,G., Tocchini-Valentini,G.R., Shoulders,C.C. and Baralle,F.E.
TITLE Selective in vitro transcription of one of the two Alu family repeats present in the 5' flanking region of the human

PUBMED 7335492
REFERENCE 40 (sites)
AUTHORS Adams,J.G. 3rd, Steinberg,M.H., Newman,M.V., Morrison,W.T., Benz,E.J. Jr. and Iyer,R.
TITLE beta-Thalassemia present in cis to a new beta-chain structural variant, Hb Vicksburg [beta 75 (E19)Leu leads to 0]
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 78 (1), 469-473 (1981)
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REFERENCE 41 (bases 61921 to 63824)
AUTHORS Spritz,R.A., Jagadeeswaran,P., Choudary,P.V., Biro,P.A., Elder,J.T., deRiel,J.K., Manley,J.L., Gefter,M.L., Forget,B.G. and Weissman,S.M.
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REFERENCE 42 (sites)
AUTHORS Baird,M., Driscoll,C., Schreiner,H., Sciarratta,G.V., Sansone,G., Niazi,G., Ramirez,F. and Bank,A.
TITLE A nucleotide change at a splice junction in the human beta-globin gene is associated with beta 0-thalassemia
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 78 (7), 4218-4221 (1981)
PUBMED 6270663
REFERENCE 43 (sites)
AUTHORS Grosveld,F., Busslinger,M., Grosveld,G., Groffen,J., DeKleine,A. and Flavell,R.A.
TITLE The structure and expression of the haemoglobin genes
JOURNAL Adv. Exp. Med. Biol. 158, 65-80 (1982)
PUBMED 6297279
REFERENCE 44 (sites)
AUTHORS Treisman,R., Proudfoot,N.J., Shander,M. and Maniatis,T.
TITLE A single-base change at a splice site in a beta 0-thalassemic gene causes abnormal RNA splicing
JOURNAL Cell 29 (3), 903-911 (1982)
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REFERENCE 45 (bases 63823 to 64083)
AUTHORS Poncz,M., Ballantine,M., Solowiejczyk,D., Barak,I., Schwartz,E. and Surrey,S.
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JOURNAL J. Biol. Chem. 257 (11), 5994-5996 (1982)
PUBMED 7076659
REFERENCE 46 (bases 61873 to 62198; 62375 to 62448)
AUTHORS Gorski,J., Fiori,M. and Mach,B.
TITLE A new nonsense mutation as the molecular basis for beta thalassaemia
JOURNAL J. Mol. Biol. 154 (3), 537-540 (1982)
PUBMED 6896219
REFERENCE 47 (bases 50716 to 51215)
AUTHORS Jagadeeswaran,P., Tuan,D., Forget,B.G. and Weissman,S.M.
TITLE A gene deletion ending at the midpoint of a repetitive DNA sequence in one form of hereditary persistence of fetal haemoglobin
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PUBMED 6174873
REFERENCE 48 (sites)
AUTHORS Orkin,S.H., Kazazian,H.H. Jr., Antonarakis,S.E., Goff,S.C., Boehm,C.D., Sexton,J.P., Waber,P.G. and Giardina,P.J.
TITLE Linkage of beta-thalassaemia mutations and beta-globin gene polymorphisms with DNA polymorphisms in human beta-globin gene cluster
JOURNAL Nature 296 (5858), 627-631 (1982)
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REFERENCE 49 (sites)

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