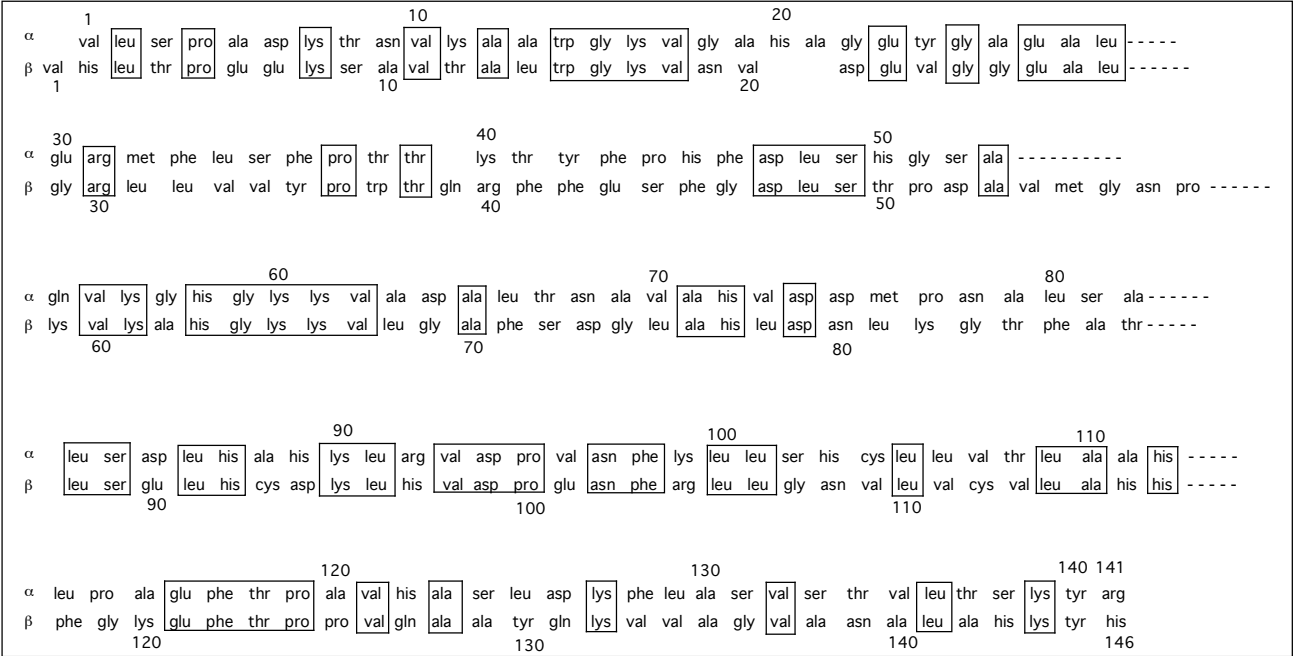


1



2

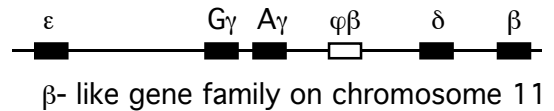
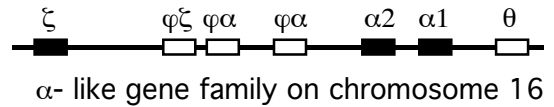
|                  | $\alpha$ - chain |         |         |         |         |         |
|------------------|------------------|---------|---------|---------|---------|---------|
| normal sequence: | 1                | 5       | 15      | 30      | 54      | 116     |
|                  | val--            | --ala-- | --gly-- | --glu-- | --gln-- | --glu   |
| HbJ Toronto      |                  | --asp-- |         |         |         |         |
| HbJ Oxford       |                  |         | --asp-- |         |         |         |
| HbG Chinesee     |                  |         |         | --gln-- |         |         |
| Hb Mexico        |                  |         |         |         | --glu-- |         |
| Hb Shimonoseki   |                  |         |         |         | --arg-- |         |
| HbO Indonesia    |                  |         |         |         |         | --lys-- |

|                  | $\beta$ - chain |         |         |         |         |
|------------------|-----------------|---------|---------|---------|---------|
| normal sequence: | 1               | 2       | 6       | 7       | 26      |
|                  | val--           | --his-- | --glu-- | --glu-- | --glu-- |
| Hb Tokuchi       |                 | --try-- |         |         |         |
| HbC              |                 |         | --lys-- |         |         |
| HbS              |                 |         | --val-- |         |         |
| Hb San Jose      |                 |         |         | --gly-- |         |
| Hb Siriraj       |                 |         |         | --lys-- |         |
| HbE              |                 |         |         |         | --lys-- |

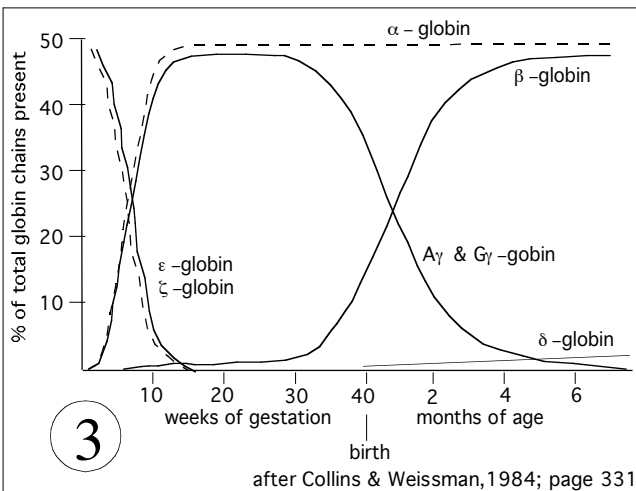
3a

embryonic (<8 weeks)  $\zeta_2 \epsilon_2 \zeta_2 \gamma_2 \alpha_2 \epsilon_2$   
 fetal (3-9 months)  $\alpha_2 \gamma_2$   
 adult (from birth)  $\alpha_2 \delta_2 \alpha_2 \beta_2$  after Genes VII



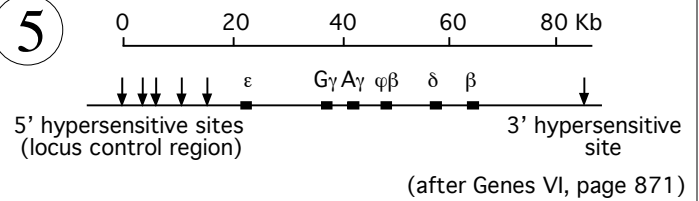
4

(after Genes VI, page 692)



3

5



|               |     |               |     |
|---------------|-----|---------------|-----|
| Alanine       | ala | Leucine       | leu |
| Arginine      | arg | Lysine        | lys |
| Asparagine    | asn | Methionine    | me  |
| Aspartic Acid | asp | Phenylalanine | phe |
| Cysteine      | cys | Proline       | pro |
| Glutamine     | gln | Serine        | ser |
| Glutamic Acid | glu | Threonine     | thr |
| Glycine       | gly | Tryptophan    | trp |
| Histidine     | his | Tyrosine      | tyr |
| Isoleucine    | ile | Valine        | val |

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1          ttagacctcaccctgtggagccaca
2  ccctagggcagttgg ccaat ctactccaggagcaggaggaggcaggagcgggtgcc ata aaagtcagggca
3  ggaccatctattgcttACATTTGCTTCTACGACACAACCTGTGTTCTAGCAACCTCAAACAGACACC
4  met val his leu thr pro glu glu lys ser ala val thr ala leu trp gly lys val asn
   ATG GTG CAC CTG ACT CCT GAG GAG AAG TCT GCC GTT ACT GCC CTG TGG GGC AAG GTG AAC
5  val asp glu val gly gly glu ala leu gly ar
   GTG GAT GAA GTT GGT GGT GAG GCC CTG GGC AGGTTG GTATCAAGGTTACAAGAC AGGTTT
6  AAGGAGACCAATAGAAACTGGGCATGTGGAGACAGAGAAGACTCTTGGGTTTCTGATAGGCACTGACTCTCTC
          g leu leu val val tyr pro trp thr gln arg
7  TGCCTATTGGTCTATTTT CCCACCCTTAGG CTG CTG GTG GTC TAC CCT TGG ACC CAG AGG
          phe phe glu ser phe gly asp leu ser thr pro asp ala val met gly asn pro lys val
8  TTC TTT GAG TCC TTT GGG GAT CTG TCC ACT CCT GAT GCT GTT ATG GGC AAC CCT AAG GTG
          lys ala his gly lys lys val leu gly ala phe ser asp gly leu ala his leu asp asn
9  AAG GCT CAT GGC AAG AAA GTG CTC GGT GCC TTT AGT GAT GGC CTG GCT CAC CTG GAC AAC
          leu lys gly thr phe ala thr leu ser glu leu his cys asp lys leu his val asp pro
10 CTC AAG GGC ACC TTT GCC ACA CTG AGT GAG CTG CAC TGT GAC AAG CTG CAC GTG GAT CCT
          glu asn phe arg
11 GAG AAC TTC AGG GTG AGTCTATGGGACCCTTGATGTTTTCTTCCCTTCTTTTCTATGGTTAAGTTCAT
22 GTCATAGGAAGGGGAGAAGTAAACAGGGTACAGTTTAGAATGGGAAACAGACGAATGATTGCATCAGTGTGGAAG
23 TCTCAGGATCGTTTTAGTTCTTTTTATTTGCTGTTTCATAACAATGTTTTCTTTTGTTAATCTTCTGTTTCTT
24 TTTTTTCTTCTCCGCAATTTTACTATTATACTTAATGCCTTAACATTGTGTATAACAAAAGGAAATATCTCT
25 GAGATACATTAAGTAACTTAAAAAAAACCTTACACAGTCTGCCTAGTACATTACTATTTGGAAATATATGTGTG
26 CTTATTTGCATATTCATAATCTCCCTACTTTATTTCTTTTATTTTTAAATGATACATAATCATTATACATATT
27 TATGGGTTAAAGTGAATGTTTTAATATGTGTACACATATTGACCAAATCAGGGTAATTTTGCAATTTGTAATTT
28 TAAAAAATGCTTCTTCTTTAATAATACTTTTTGTTTATCTTATTTCTAATACTTCCCTAACTCTTTCTTT
29 CAGGGAATAATGATACAATGTATCATGCCTCTTGCACCATCTAAAGAATAACAGTGATAATTTCTGGGTTA
30 AGGCAATAGCAATATTTCTGCATATAAATTTCTGCATATAAATTTGTAAGTATGTAAGAGGTTTCATATTGC
31 TAATAGCAGCTACAATCCAGCTACCATTCTGCTTTTATTTATGTTGGGATAAGGCTGGATTATTCTGAGTCC
          leu leu gly asn val leu
22 AAGCTAGGCCCTTTTGCTAATCATGTTTCATACCTCTTATCTTCTCCACAGCTCCTG GGC AAC GTG CTG
          val cys val leu ala his his phe gly lys glu phe thr pro pro val gln ala ala tyr
23 GTCTGTGTGCTGGCCCAT CAC TTT GGC AAAGAATTC ACCCCACAGTG CAGGCT GCC TAT
          gln lys val val ala gly val ala asn ala leu ala his lys thr his
24 CAGAAA GTG GTG GCT GGT GTG GCT AAT GCC CTG GCC CAC AAG TAT CAC TAAGCTCGCTT
25 CTTGCTGTCCAATTTCTATTAAGGTTCTTTGTTCCCTAAGTCCAACACTAAACTGGGGGATATTATGAAG
26 GGCCTTGAGCATCTGGATTCTGCCTAATAAAAAACATTTATTTTCATTGC AATGA TGATTTAAATTTATTTCT
27 GAATATTTTACTAAAAAGGGAATgtgggaggtcagtgcat taaacataaaagaaatgaagagctagttcaa
28 acctgggaaatacactatcttaactccatgaaagaaggtgaggtgcaaacagctaatgcacattggcaac
29 agccctgatgcctatgcctattcatcctcagatgctaaaggattcaagtagaggttgattggaggttaagttt
30 atgctgtatttacattactattgttttagctgctcctcatgaatgcttttcacaccattgcttatcctgcactctc
31 tcagccttgactccactcagtt

```

Nucleotide sequence of beta-globin gene

non-transcribed strand shown

5' ..... 3'

lower case (a) -- not transcribed  
upper case (A) -- transcribed

5' leader to ATG;  
translated bases with amino acid designations;  
introns between translated bases;  
intron splice sites: at 5' end first base of intron underlined, at 3' end first base of next exon underlined

First base transcribed = +1  
Base to the left = -1

ccaat = cat box;  
ata = tata box  
double underline = other promoters

underline at 3' end = sequences required for poly-A site and ? termination of transcription

dot = attachment site poly A

References

sequence and promoters from Collins and Weissman; Prog. Nucleic Acid Res. and Mol. Biol. v.31; 1984

3' details from Singer and Berg, 1991

For sequence details see :  
[http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?cmd=Retrieve&db=nucleotide&list\\_uids=455025&dopt=GenBank&term=HUMHBB&qty=1](http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?cmd=Retrieve&db=nucleotide&list_uids=455025&dopt=GenBank&term=HUMHBB&qty=1)

Diagram of some important regions of the beta-globin gene (not to scale)

