Hands-On Eight
Beta Thalassemia

A) We are going to use a gene predictor to see the exons and introns of the beta globin gene.

- Obtain the sequence "beta_globin_sequence".
- Open a web browser and go to the Genscan server at: http://genes.mit.edu/GENSCAN.html
- Paste the sequence into the DNA field.
- Keep the default options and press "Run Genscan".
- Read the following explanation and answer the questions related to the table that GenScan generated.

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Gn.Ex : gene number, exon number (for reference)
Type  : Init = Initial exon (ATG to 5' splice site)
        Intr = Internal exon (3' splice site to 5' splice site)
        Term = Terminal exon (3' splice site to stop codon)
        Sngl = Single-exon gene (ATG to stop)
        Prom = Promoter (TATA box / initation site)
        PlyA = poly-A signal (consensus: AATAAA)
S     : DNA strand (+ = input strand; - = opposite strand)
Begin : beginning of exon or signal (numbered on input strand)
End   : end point of exon or signal (numbered on input strand)
Len   : length of exon or signal (bp)
Fr    : reading frame (a forward strand codon ending at x has frame x mod 3)
Ph    : net phase of exon (exon length modulo 3)
I/Ac  : initiation signal or 3' splice site score (tenth bit units)
Do/T  : 5' splice site or termination signal score (tenth bit units)
CodRg : coding region score (tenth bit units)
P     : probability of exon (sum over all parses containing exon)
Tscr  : exon score (depends on length, I/Ac, Do/T and CodRg scores)
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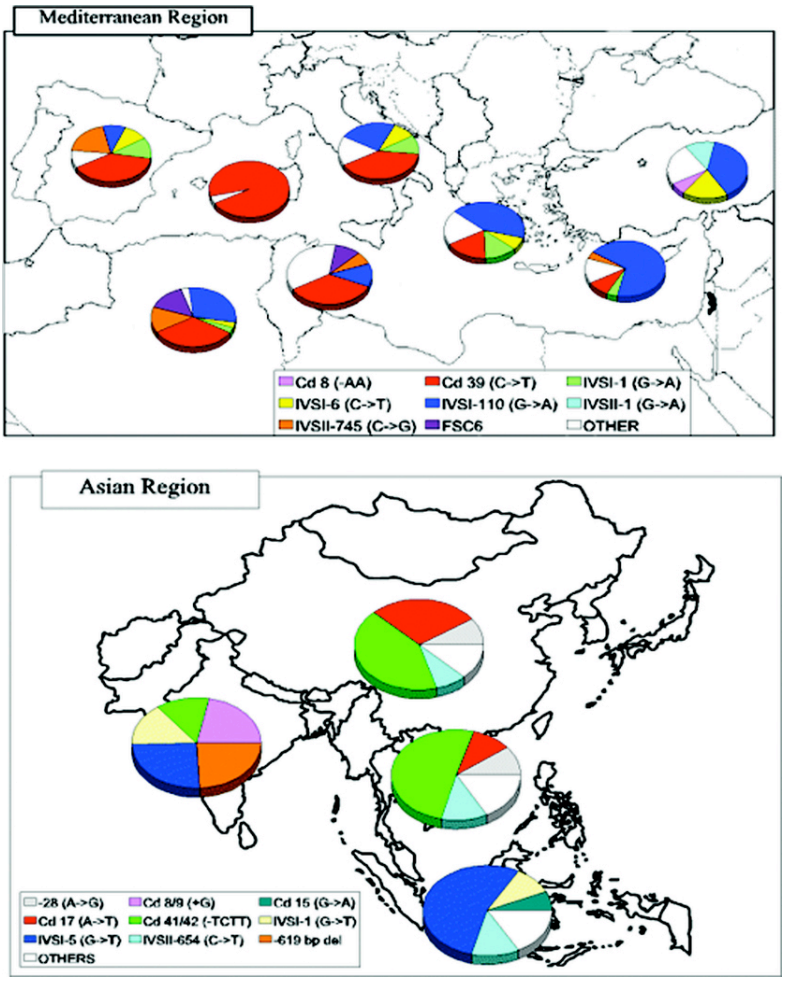
1. How many exons are predicted? _____ .
2. On which strand (+ or -) is the gene located? _____ .
3. Fill in the table:

Exon or Signal Number	Is it an Exon?	Exon or Signal Start Position	Exon or Signal End Position	Probability
1				
2				
3				
4				

B) "**Beta thalassemia**" by Cao et al. appeared in Genetics IN Medicine • Volume 12, Number 2, February 2010 (pages 61 to 76).

The introduction of the article claims that: "Beta-thalassemia is one of most common autosomal recessive disorders worldwide. High prevalence is present in populations in the Mediterranean, Middle-East, Transcaucasus, Central Asia, Indian subcontinent, and Far East."

Figure 3 of the article gives the most common beta-thalassemia mutations in different countries.



For each of the following three mutations that appear in Figure 3:

a) locate the mutation on the sequence, and

b) explain what consequences on the protein it might have.

1) Cd 39 (C->T)
2) Cd 41/42 (-CTTT)
3) IVSI-110 (G -> A)

Recall the anatomy of an intron: