Exercise – UCSC Genome Browser

This exercise is a basic introduction to the UCSC genome browser. Read the text and answer the question written in red.

- Open UCSC browser http://genome.ucsc.edu/ and find the human gene Artn (select isoform 1)
    - Via 'Genome Browser' & 'position or search term'

1. Understanding the graphical view
    a. Meaning of color coding ?
    b. What does the 3 line types (blocks) represent:
        i. The lines ?
        ii. The relative thin blocks ?
        iii. The fat blocks ?

- Add the track SNPs (131) – Color on SNPs
    - Zoom in on the SNPs – click on 'base position' bar in top of window
    - Genetic code - http://en.wikipedia.org/wiki/Genetic_code
    - Donor/acceptor pictures are shown at the end of exercise

2. What is the color of the Non-Synonymous SNPs ?
3. What is the implication of SNP rs2242637 ?
4. What is the implication of SNP rs12737332 ? (use figures 1 & 2)

- Orthologues

5. What is the % identity between human and mouse Artn ?
6. What are the up/down stream neighbours of Artn ?
7. Is the synteny conserved in mouse ?

- Chromosomal location

BRCA1 (BReast CAncer 1 gene) is a tumor suppressor gene where mutations can have a strong correlation to breast cancer. Mutations in BRCA1 are thought to be responsible for 45% of inherited breast cancer. Moreover, BRCA1 carriers have a 4-fold increased risk of colon cancer, whereas male carriers face a 3-fold increased risk of prostate cancer. Cells lacking BRCA1 show defects in DNA repair by homologous recombination.

1. At which locus is BRCA1 found ?
2. Is BRCA1 located at forward or reverse strand ?

Zoom into this region chr17:41,215,327-41,215,401
3. how many basepairs does the region span ?
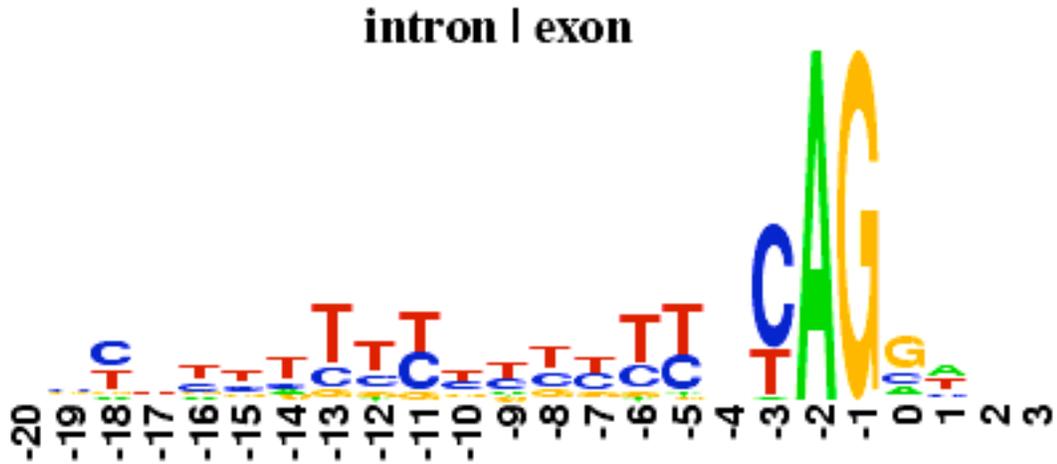
Donor/Acceptor sites
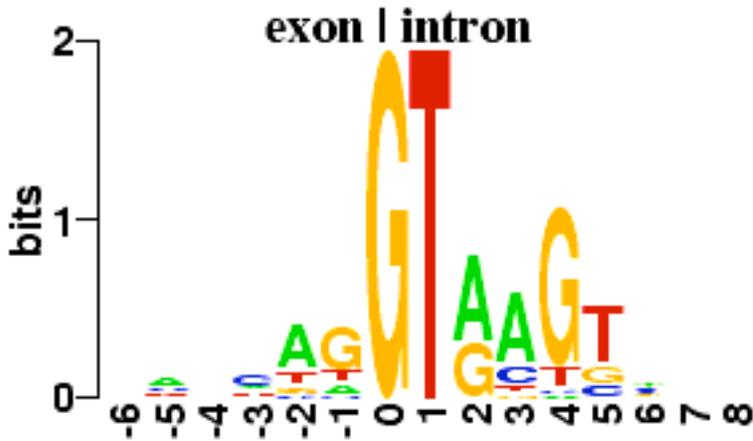

Figure 1 shows logo for a small set of human Acceptor sites

Figure 2 shows logo for a small set of human donor sites