CSCI 5465
Lab#2
Quincy Gu

Part I (2) Questions

a. Name the gene you chose and briefly describe its function and its association with the disease.

   The gene I chose is 'BRCA1'.

   Function Description:

   BRCA1 gene forms many distinct complexes together with variety of adaptor proteins, and normally serve as a negative regulatory of mammary epithelial cell growth (Thompson et al., 1995). It has a strong effect on DNA repair, cell cycle checkpoint control, and maintaining genomic stability (Wang et al., 2009).

   BRCA1's association with the disease:

   1. BRCA1 carriers has higher risk in breast and ovarian cancer based on the family history research (Gayther et al., 1995);
   2. BRCA1 associated hereditary ovarian cancer patients' survival conditions usually better than non-hereditary patients (Boyd at al.,2000);
   3. The deficiency of PTEN protein expression was significantly associated with the highly proliferative, poorly differentiated and prognosis basal-like cancer subtype in BRCA1-deficient breast cancer (Saal et al., 2008).

b. How many allelic variants are associated with your gene? List a few examples, and describe the mutations associated with each variant (e.g. substitution vs. insertion/deletion).

   There are 44 allelic variants associated with the 'BRCA1' gene I chose.

   Examples & Variant associated mutations descriptions:

   1. Index .003 – Mutation type is deletion associated with BRCA1, 2-BP, 185AG;
   2. Index .005 – Mutation type is insertion associated with BRCA1, 1-BP;
   3. Index .0031 – Mutation type is duplication associated with BRCA1, 6-KB, EX13.

   Reference Link for these above examples is the following:

   https://omim.org/allelicVariants/113705

c. On which chromosome is your gene located in the human genome?

The chromosome 17 is where BRCA1 gene located in the human genome.

d. Does your gene have an ortholog in any other species? If so, name 2-3 other species and report the sequence similarity to the human gene.

The BRCA1 gene does have orthologs in some other species except human.

Examples with species names and its sequence similarity to the human gene:

1. Mouse – 74.24 (n) is the sequence similarity to the human gene;
2. Chicken – 54.18 (n) is the sequence similarity to the human gene;
3. Lizard – 37 (a) is the sequence similarity to the human gene;
4. African clawed frog – no sequence similarity to the human gene info listed in the Gene Cards webpage.

Reference Link for the above examples in the following:

https://www.genecards.org/cgi-bin/carddisp.pl?gene=BRCA1&keywords=BRCA1#orthologs