**Q1: How could we use the DNA alignments (or lack thereof) for each chromosome to verify that each individual is the sex we expect them to be?**

**Q2: For each of the following variants, answer the following questions:**

a) what type of polymorphism is this: a SNP (single-nucleotide polymorphism) or an INDEL (Insertion-deletion polymorphism)?

b) using a diagram such as the one below (Fig. 1), draw the inheritance pattern of the variant from the parents to the child. For each individual, mark their genotype (that is, if one chromosome is clearly a G and the other is clearly an A, mark the genotype as A/G). Is there anything unexpected about the inheritance pattern? That is, could the alleles present in the child be explained given the alleles found in the parents? If not, what might have happened?

c) What is the evidence that this is a functionally important mutation?

   a. does the variant change an amino acid? (i.e., is it synonymous or non-synonymous?
      i. If it is non-synonymous, what is the amino acid change:
         1. E.g., from I to T
   b. does it delete or insert an amino acid?
   c. Does it cause a shift in the coding frame (a "frameshift")

**Note: if the variant is an INDEL, do not worry about providing the genotypes for each individual. Just tell us if it whether or not it disrupts the gene's reading frame and deletes amino acids.**

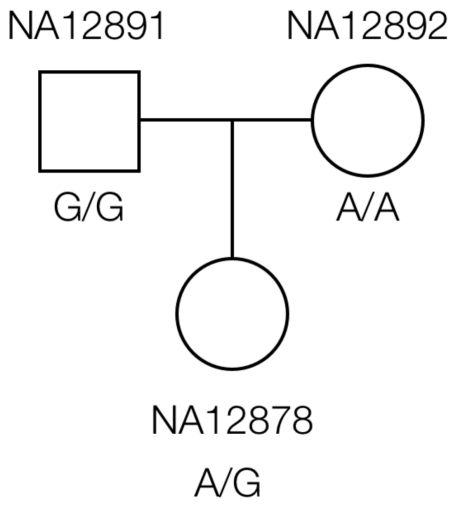


**Figure 1. Example diagram for the drawing the inheritance patterns of the questions below.**

**Variant #1: chr1:247978318-247978319  (hint: just paste in these coordinates to IGV)**

**Variant #2: chr1:248004847-248004848**

**Variant #3: chr1:216595305-216595306**

**Variant #4: chr1:236729956-236729956**

**Variant #5: chr1:241755348-241755348**

**Variant #6: chr1:248059702-248059703**
**What else is interesting about this variant and the nearby C/T variant at 258059712?**

**Variant #7: chr1:216172379-216172380**

**Variant #8: chr1:216172379-216172380**

**Variant #9: chr1:47080678-47080679**

**Variant #10: chr1:67242086-67242087**

**Variant #11: chr1:248,024,002-248,024,144**

**Variant #12: chr1:100316588-100316589**

**Variant #13: chr14:56763333-56763373**

**Variant #14: chr16:30,768,388-30,768,596**

**Variant #15: chr19:54,663,828-54,663,963  HINT: hover over the purple "I"s for more information.**

**Variant #16: chr19:52,887,077-52,887,212**

**Variant #17: chr17:76,798,538-76,798,558**