Find Me a Project!
A Helpful Walkthrough by Zach Gaudette

1. Log onto http://www.genome.gov/10001204 or utilize Google searches to learn about the causes, symptoms, and significance of multiple genetic diseases.

2. Discuss which diseases interest you and why. Select ~2 diseases that you would like to research further. For those diseases, determine which genes are affected. Record the genes’ common symbols/nomenclature for future use. For example, the disease Cystic Fibrosis is caused by mutations to the “Cystic Fibrosis Transmembrane Conductance Regulator” gene – more often abbreviated as the CFTR gene.

3. Log onto http://omim.org and enter a gene of interest into the search bar.

4. The first search result will often be the name of your gene, preceded by a red asterisk or other symbol. Click on it.

5. The resulting page contains a great wealth of information on your gene of interest and should be bookmarked immediately. You should explore this entire page yourself but, for now, scroll down to the bolded subheading “ALLELIC VARIANTS.”

6. Each of the entries in this section, often labeled with numbers like “.0001,” describes the clinical and scientific significance of a specific mutation of your gene. Unless you have a much cooler project in mind, your work this semester will likely be based around detection of specific mutations like these. Read about multiple mutations. Ensure that their presence leads to (or is a marker of) your disease of interest.

7. Select a few mutations and research them further. You can do so by clicking on links to past research papers. Example: Yang et al. (2003) Clicking on these links will take you down the page to the bibliography section, where the paper’s full information is listed. The bibliography entry is often followed by links to PubMed or other full-text versions of the paper.

8. Determine which mutation(s) you would like to base your semester’s work on. Return to the subheading (mentioned is step 6) of that/those mutation(s). There is often a blue link below the mutation’s heading with a format similar to this example: [dbSNP:rs5030858]. Don’t worry if such a link is not present. This often means that your mutation affects more than just a few nucleotides of DNA. Such large mutations often make for an especially interesting experimental design and project but, for now, we will assume that a link is available. This means that the mutation affects a small number of nucleotides, often just one. Click on the link.
9. This link will take you to a site containing more cataloged information about your mutation. Scroll down to the bolded subheading “Explore this variation.” It should contain multiple buttons that you’d be wise to explore on your own but, for now, click on the button titled “Flanking sequence.”

10. The resulting page should look similar, but notice that the “Explore this variation” subheading has been replaced with the title “Flanking Sequence.” Below this subheading will now be a list of letters. This page is presenting you will the DNA nucleotide sequence directly adjacent to your mutation of interest. This is great information to construct your PCR primers from – bookmark this page.

11. To avoid confusion, look to the left of this page and click on the blue button with a cogwheel symbol labeled “Configure this page.”

12. From the three selection bars present on the resulting pop-up screen, select:
   1) Length of reference flanking sequence to display: 1000bp
   2) Sequence selection: Upstream and downstream sequences
   3) Show variations in flanking sequence: No

**Once finished, click the checkmark in the upper-right corner to return to the sequence screen

13. Now the nucleotide sequence flanking your mutation of interest (the red underlined letter) has been cleared of all other genetic variations possible in the area, and spans for 1000 base pairs to each side. Highlight this string of letters and transfer them to a blank Microsoft Word document. Delete the resulting spaces, but not the letters! Save this document and you will have a workable piece of genomic sequence to develop PCR primers from later.

14. Rejoice!