**CHM/BIO 422000 Biochemistry: Metabolism**

**Directions for the Metabolic Disease Research Project**

**Due Dates:**

**March 21, 2013** Choose a topic and begin your research at the OMIM site. Start a Word file to collect information and references as you go. Read through the sections of the report that are needed and use the sites listed there to perform your research.

**March 28, 2013** Report outline and basic summary of mutation linked to disease (explain the genotype to phenotype correlation). – Please turn in only one page and print this out to turn in – no email submission. I’ll return it with comments in a week.

**April 18, 2013** Final Report is due.

For this project, you can choose your own topic from the OMIM list of the genes with known correlations to diseases. Alternatively, you can use the “Clinical Insights” sections from your text to choose a topic. After choosing a genetic disease to research, choose a particular mutation to study from the “Allelic Variants” section of the OMIM site. The OMIM entry will be your starting point for the research project, but you will need to gather additional information from the list of sources below:

**Formatting directions:**

Times or Arial 12 pt font and double-spaced

**Report section headings**:

(Use the protein name as the heading – example **Hemoglobin**) – In this section, describe the background of the normal function of the protein and the normal functioning of whatever pathway it is involved. For example, for hemoglobin you would describe the quaternary structure of hemoglobin and how it functions cooperatively to bind oxygen in the lungs and release it at tissues. You should use the OMIM website, UniProtKB, the KEGG database and your textbook for information in this section.

(Use the disease name for this section – example **Sickel Cell Anemia**) – In this section, describe the physiology of the disease including the symptoms and issues that patients experience. If molecular details are known (like protein aggregation, etc) describe that here also. You will probably get this from the OMIM website as a starting point, but you should obtain at least one additional journal article

(Use the amino acid mutation notation as your section title – example **Glu6Val**) Description of the mutation you have chosen to explore. This should include a discussion of the chemical differences of the amino acid side chains and how that change could be related to the change in protein function. For example, for hemoglobin, this section would describe that Glu is negative and when replaced with Val, this new hydrophobic side chain sticks out and fits into a hydrophobic pocket of adjacent hemoglobin tetramers. For this section, you should use the Firstglance in Jmol site to model the mutation and make a figure of the protein for your report.

**Conclusion** – This section should make the final connection summarizing how the mutation leads to a change in protein function and how, by understanding the pathways that change, the symptoms of the disease can possibly be explained. It’s okay to put your own interpretation here and explain what you think is happening even if it isn’t really all known yet.

**Figures –** Each figure needs to be numbered and referred to in the text. You need at least two figures, but you can have up to 5 if the figures really help you explain the story. Be careful not to insert a figure and then forget to explain it in the text. Each figure should also have a brief figure legend with one or two sentences explaining what the figure shows. The reference for the figure can be put at the end of the figure legend, but should be in your final reference list as well.

**References** – Use ACS (American Chemical Society Style Guide) for formatting references. Example formats are shown below.

Website citation format:

ACS Publications Division Home Page. http://pubs.acs.org

(accessed Nov 7, 2004)

Journal article example format:

Klingler, J. Influence of Pretreatment on Sodium Powder. *Chem.*

*Mater.* **2005**, *17*, 2755–2768.