BI 441/541 HUMAN DISEASE GENETICS REPORT

BACKGROUND

Many human diseases have genetic underpinnings. While we will mention several of these diseases in class, we will not get to them all. To allow you to explore one disease in-depth and practice scientific writing skills, your assignment is to author a 2-3 page paper on a human disease caused by abnormal genetics. Specifics on finding a genetically-caused disease are below. Please choose a disease with no more than three (3) genes involved.

DIRECTIONS

1. To browse through disease choices, go to the Online Mendelian Inheritance in Man website listing human genetic diseases, http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd&ref=toc.

From here, you may search for diseases by affected body system, using links in the left-hand column. I worked with the campus library to set up a link specifically for this assignment. You may link from Moodle, or directly from the library website, http://www.wou.edu/provost/library/ (then class stuff).

2. Read through several descriptions before choosing a disease. Sign up on the sheet posted by my office door – you will need the name of the disease and the chromosome on which the involved gene is located. Every student is responsible for choosing one disease upon which to write a paper. No more than two students may choose the same disease, so you may want to have a back-up choice or two. First student (or students) to sign up has "dibs" on that disease.

3. You will need to find additional resources to support your research. At least three references are required, in addition to the OMIM website (thus four total), and at least one reference must be a book (besides our textbook) or journal article. You must cite your references in your paper.

A note on reading scientific literature: As you read through the information, there may be many unknown terms, phrases, and ideas.

Read through the information once, circling terms that are unfamiliar, and noting sections that do not make sense.

Look up unknown terms, reread these assignment directions, and read through your literature again, building an understanding of the overall point of the article.

You may need to read through the information, or sections of info, several times to gain a full understanding; this is okay and expected.

4. Write a two- or three-page, typed, description of your disease (not including title page if you have one). You should include the disease name, phenotype/manifestations, responsible gene(s), and a description of what type of mutation is the cause and *how* the mutation leads to the disease traits. You will also want to include other information as you see fit (for example: predominantly affected ethnic groups, available treatments, any preventative measures, etc.). You may follow MLA, APA, or CBE/CSE writing guidelines (a brief description below). For more specifics see online sites.

MLA: *American Psychological Association* formatting style. Double-spaced, paper (8.5" x 11") with 1" margins, 10-12 pt Times New Roman or a similar font. Title Page, Abstract, Main Body, References.

APA: *Modern Language Association* formatting style. Double-spaced, paper (8.5" x11") with 1" margins, 12 pt font. No title page - information included at top of first page, Main Body, References.

CBE/CSE: *Council of Biology/Science Editors* formatting style. Double-spaced, paper (8.5" x11") with 1" margins, 12 pt font. Title page, Abstract Page, Main Body, References.

GRADING

Your paper is due at the beginning of class on **Thursday, February 26th 2009.** It will be graded out of 50 points maximum. Five (5) points will be deducted for each day it is late.
You have some leeway in compiling your paper and expressing your written "voice". However, grades will be assigned according to the criteria below. Criteria to receive full points are listed in the right-hand column, with columns to the left indicating categories of partial points.

Grading Rubric

Element	Misses	Approaches	Meets
Introduction **5 points**	▪ Poor sentence structure, little background information ▪ Does not mention genes involved or chromosome	▪ Complete sentences, some background information ▪ Does not mention genes involved	▪ Introduction well written with complete sentences and background information ▪ Mention disease name, gene(s) involved, and chromosome
Phenotype Description **10 points**	▪ Fails to describe disease symptoms or characteristics ▪ Does not describe the disease inheritance pattern	▪ Describes some diseases symptoms, is too brief or unclear ▪ Haphazard description	▪ Clear description of disease symptoms/characteristics ▪ Indicates how the disease is inherited in families
Normal Gene Function **10 points**	▪ Does not describe how the normal gene works or what protein is made	▪ Describes what gene is involved but does not mention how it normally functions	▪ Describes clearly how the normal gene works including what protein is made and the role of that protein
Abnormal Gene Effects **10 points**	▪ Fails to mention the type of mutation ▪ Does not explain what the problem is with the mutant form of the gene ▪ Does not describe how the mutant gene leads to the disease phenotype	▪ Mentions mutation name but not how DNA is changed ▪ Discusses the gene product but not how the mutant form is different from normal ▪ Talks about the abnormal gene but no clear link to the disease symptoms	▪ Clear description of what type of mutation is involved ▪ Explanation of how the gene and/or gene product fails to function in its abnormal form ▪ Describes how abnormal gene function leads to the disease phenotypes
Content **5 points**	▪ Lacks any additional information to what is asked for above	▪ Includes some information in addition to above, although description not complete ▪ Information is disorganized in the way it is included	▪ In addition to the information above provides a full picture of what this disease involves and how people who are diagnosed with it are affected. ▪ Tied in smoothly with topics
Quality of Writing **10 points**	▪ No specific formatting ▪ Many grammar, punctuation, spelling errors ▪ Lacks organization ▪ Sections blend together ▪ Uses 1st person often ▪ No citations or references	▪ Formatted sporadically ▪ Few grammar, punctuation and spelling errors ▪ Not organized w/in paragraphs ▪ Sections not well labeled ▪ Uses 1st person occasionally ▪ Citations and references included, but incorrect	▪ Appropriate formatting ▪ Grammar, punctuation and spelling are correct ▪ Paper well-organized w/in and between paragraphs ▪ Sections clearly labeled ▪ 3rd person used throughout ▪ Correct citations and complete references listed

Example Grade Sheet **Human Heredity Genetic Disease Report**

Student Name:_____

Element	Your category is circled			Your Score
Introduction	Missed: 1-2 points	Approached: 3-4 points	Met: 5 points	
Phenotype Description	Missed: 2-5 points	Approached: 6-9 points	Met: 10 points	
Normal Gene Function	Missed: 2-5 points	Approached: 6-9 points	Met: 10 points	
Abnormal Gene Effects	Missed: 2-5 points	Approached: 6-9 points	Met: 10 points	
Content	Missed: 1-2 points	Approached: 3-4 points	Met: 5 points	
Quality of Writing	Missed: 2-5 points	Approached: 6-9 points	Met: 10 points	

Total: / 50

Comments: