

Cheme355

name \_\_\_\_\_

Autumn 02

Module 2 Take home exam (75 pts)

DUE: November 25, at class

In the tissues module, we've learned how the genetic makeup of an organism influences the phenotype. We've used the disease Osteogenesis Imperfecta as a model of how the structure of a protein affects its function, how that structure is affected by a genetic mutation, and how this mutation can be passed on to further generations. Apply what you've learned to understand one of the disorders below.

**Resources:**

Work on this on your own, do not discuss it with other members of the class. You can use any web or printed resources to help, and you can ask any of the course instructors or the TA for clarification help.

General Biology Textbooks

Web Searches

Library

OMIM:

<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>

PubMed:

<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=PubMed>

Biology on-line textbook: [http://highered.mcgraw-](http://highered.mcgraw-hill.com/sites/0073031208/student_view0/essential_study_partner.html)

[hill.com/sites/0073031208/student\\_view0/essential\\_study\\_partner.html](http://highered.mcgraw-hill.com/sites/0073031208/student_view0/essential_study_partner.html)

Genetics on-line:

<http://www.botany.uwc.ac.za/mirrors/MIT-bio/bio/mg/mgdir.html>

Meiosis: (real player)

[http://www.mhhe.com/biosci/esp/2001\\_gbio/folder\\_structure/ge/m1/s5/assets/real/ge\\_m1s5\\_8.rm](http://www.mhhe.com/biosci/esp/2001_gbio/folder_structure/ge/m1/s5/assets/real/ge_m1s5_8.rm)

recombination: (shockwave)

[http://www.mhhe.com/biosci/esp/2001\\_gbio/folder\\_structure/ge/m1/s6/gem1s6\\_2.htm](http://www.mhhe.com/biosci/esp/2001_gbio/folder_structure/ge/m1/s6/gem1s6_2.htm)

crossover: (Shockwave)

[http://www.mhhe.com/biosci/esp/2001\\_gbio/folder\\_structure/ge/m1/s6/gem1s6\\_4.htm](http://www.mhhe.com/biosci/esp/2001_gbio/folder_structure/ge/m1/s6/gem1s6_4.htm)

**Topic/Questions:**

Choose one of the following inherited diseases:

- Sickle Cell Anemia
- Wilson Disease
- Alcohol Intolerance, acute

**Research the disease you've chosen on OMIM and the other resources, and answer the following questions:**

1. What is the gene that is mutated in people with this disease? (Genotype)
2. What is the role of this protein in cells? Provide information about location, function, importance, etc...
3. How is the disease inherited, is it sex linked, autosomal? Is it dominant, recessive, or does the phenotype mirror the genotype? (one mutated allele and one normal allele causes milder disease than two mutated alleles)
4. What are the symptoms and the affected tissues, organs, and/or structures? Discuss known or postulated reasons that this mutation could cause these effects. (Phenotype)
5. What is the current treatment or therapy for this disease? How/why does this compensate for the mutation or relieve symptoms of the disease?

6. Module 2 Wrap-up: What are the main concepts we have covered in the Tissues (OI) Module (list 2-3 main concepts per class session for the sessions starting Oct. 28)