

Fall 2003 Biology 111 Exam #2.5 - Molecular Genetics Half Exam KEY

There is no time limit on this test, though I have tried to design one that you should be able to complete within 1.5 hours, except for typing. You are not allowed to use your notes, old tests, any electronic sources, any books, nor are you allowed to discuss the test with anyone until all exams are turned in by class on Monday November 3. **EXAMS ARE DUE AT CLASS TIME ON MONDAY NOVEMBER 3.** You may use a calculator and/or ruler. The **answers to the questions must be typed on a separate sheet of paper** unless the question specifically says to write the answer in the space provided. If you do not write your answers on the appropriate pages, I may not find them unless you have indicated where the answers are. There are 3 pages to this exam, including this cover sheet.

- 3 pts if you do not follow this direction.

Please do not write or type your name on any page other than this cover page. Staple all your pages (INCLUDING THE TEST PAGES) together when finished with the exam.

Name (please print):

Write out the full pledge and sign:

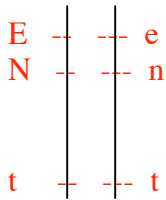
How long did this exam take you to complete (excluding typing)?

10 pts.

1) Three recessive traits exist in a species that has orange round heads. The recessive traits are triangle eyes, pyramid nose and one tooth. Determine which alleles were on each parent's chromosomes, the order of the loci and the distance between each locus. Use the space provided here for your answer:

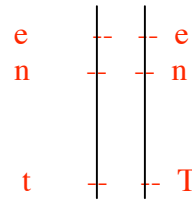
round eyes, single tooth, wedge nose

Draw Mother's Alleles below



triangle nose, many teeth, pyramid nose

Draw Father's Alleles below



e = eyes triangle eyes

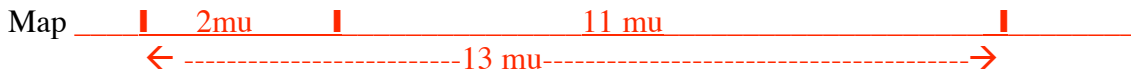
t = single tooth

n = pyramid nose

E = round eyes

T = many teeth

N = wedge nose



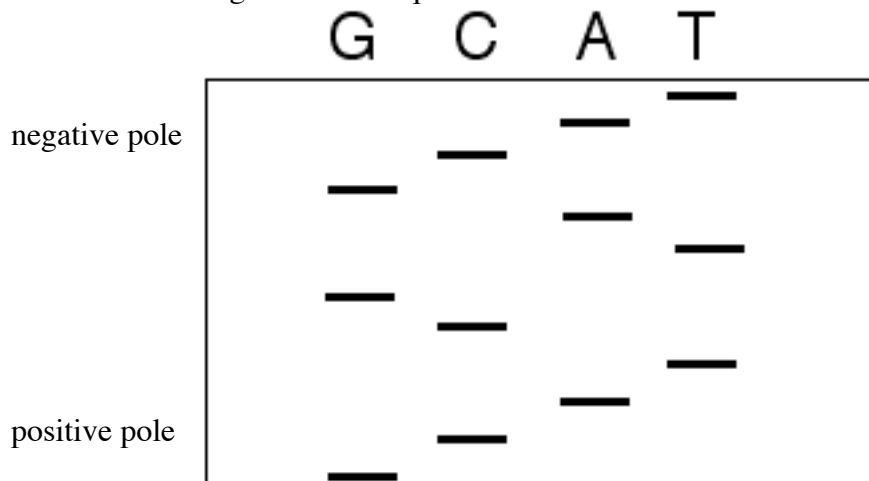
Phenotype

Number of Individuals

| | |
|---|------------|
| round eyes, single tooth, wedge nose | 370 |
| round eyes, many teeth, pyramid nose | 9 |
| round eyes, many teeth, wedge nose | 47 |
| round eyes, single tooth, pyramid nose | 1 |
| triangle eyes, many teeth, pyramid nose | 369 |
| triangle eyes, single tooth, wedge nose | 8 |
| triangle eyes, single tooth, pyramid nose | 45 |
| triangle eyes, many teeth, wedge nose | <u>1</u> |
| | 850 |

6 pts.

2) In the space provided, draw a picture of a sequencing gel if each dideoxynucleotide reaction was loaded onto a different lane of a gel and the sequence was GCATCGTAGCAT.



6 pts.

3) Go to this <<http://www.ncbi.nlm.nih.gov:80/BLAST/>> and determine the gene, accession number, and species from which this sequence was extracted.

MVHLTPEEKSAVTALWGKVNVDVEVGGEALGRLLVVYPWTQRFFE

Homo sapiens beta globin gene (variant) # AAN84548

6 pts.

4) Draw a picture of CFTR in two ways. One way should show the major features of the protein as determined from the sequence analysis online, including the most common mutation. The second way should show what CFTR looks like in its normal 3D shape while performing its normal function.

Your first picture should have looked like the one we drew in class and your second one should have looked like the one we viewed online with two channels side by side.

4 pts.

5) Why is the mucus thick in people with two $\Delta F508$ alleles?

- the protein is stuck in the ER
- this leads to a lack of Cl^- ions in the mucus and therefore not as much water leaves the cells and enters the mucus
- the mucus is thicker than normal due to the reduced water
- two alleles are needed to have the disease since this is a loss of function (recessive allele)

6 pts.

6) a. Describe the type of mutation that causes Huntington's disease.

Trinucleotide repeat that results in too many glutamine amino acids in a row in the mutant protein.

This makes the protein too sticky and thus dominant (over active).

b. What happens over time to a family's mutant allele that causes the allele to disappear from the human gene pool?

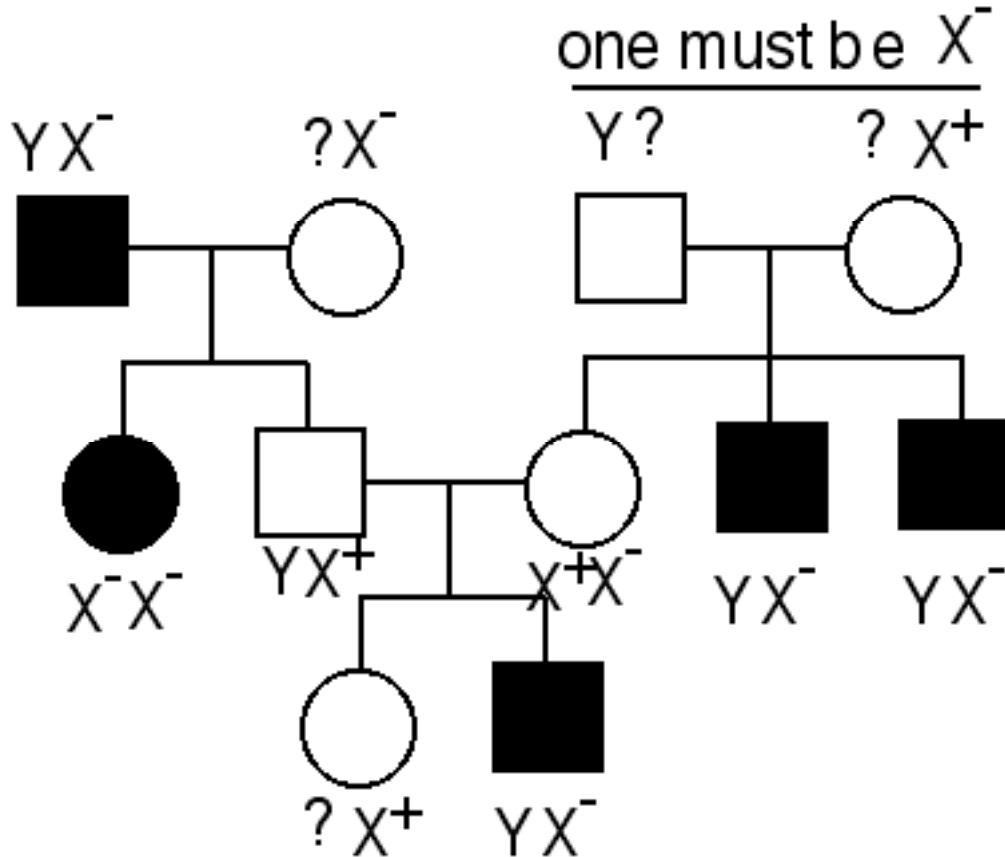
As the repeat expands with each generation, the age of onset is younger. This leads to death before reproduction and the allele disappears from the family because all affected people die before passing on the allele.

12 pts.

7) a. Draw a pedigree that describes a family with Duchene's muscular dystrophy and meets the conditions below. Be sure to include as many grandparents as you can.

- 1) The patient is a boy. His parents have no symptoms. He has a twin sister who does not have any symptoms.
- 2) Both of the boy's maternal uncles have muscular dystrophy.
- 3) The boy's paternal aunt has muscular dystrophy.

b. Write in the genotypes of all individuals in the pedigree. If you are uncertain about a particular person, put in a ? for any uncertain alleles.



c. Is it possible that this disease is sex-linked or not? Explain your answer.

Yes, the data are consistent with a sex-linked disease. See genotypes for explanation.