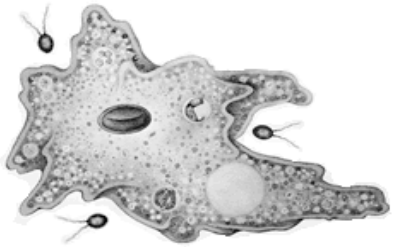
MCDB 2222 coreBIO: Midterm 1 - Spring 2018 NAME: _____



Directions: The exam is worth 100 points.

Carefully read each question to determine what the question wants you to do. **DO QUESTIONS 1-16 first - and then 17/18 if you have time**

Q1 (6 points): Draw a schematic that contains the common elements present in most eukaryotic genes.

enhancer, promotor (coding region - exons / introns)

Q2 (10 points): For each of these elements, describe how an amorphic mutation could "work"; if you think that such an amorphic mutation is impossible, explain why.

mutation inactivates enhancer - no transcription - no gene product made
mutation inactivates promotor - no transcription - no gene product made
mutation inactivates start code / introduces stop codon - no functional gene product made
mutation inactivates exon/intron junction - no functional gene product

Q3 (5 points): Explain how an amorphic mutation could produce a dominant allele.

The amount of gene product produced by a single allele is not sufficient to generate the wild type phenotype - the amorphic allele is (dominant).

Q4 (6 points) In a eukaryote, a mutation on a chromosome leads to the inability to form a centromere. Draw a picture of how such a mutant chromosome would behave during mitosis and explain why.

The chromosome would not be able to connect to the mitotic spindle (chromosome segregation) machinery.

Q5 (4 points) Draw the effects of such a mutation on the two progeny cells produced by cell division.

Without connecting to the mitotic spindle (chromosome segregation machine), the duplicated chromosome would go to one or the other of the daughter cells; one would get two copies, the other, none.

Q6 (6 points) Provide a short description of a condition under which you might expect sexual dimorphism to arise. Explain how the two sexes will be impacted.

If the two sexes make unequal contributions to the development or nurturing of the offspring, they will be subject to different selective pressures, leading to different adaptations.

The nature of the impact will depend upon the difference in investment, as well as the value of various traits - for example, the value of cooperation in raising offspring successfully.

Q7 (4 points) Consider a situation in which the female parent provide substantial resources to insure the success of its offspring. Such as situation can favor a form of sexual selection, a form in which females choose which male(s) to mate with. What features in a male would likely be selected for, explain your answer.

The female may choose the most robust male, based on visible markings of health and such. This can lead to selection based on size or other markers, the male peacock's tail is an example.

Alternatively, the female may look for signs of male cooperation, the willingness to form a loyal bond and help to nurture the offspring (e.g. bring food and such).

Q8 (5 points) Consider two homologous chromosomes during meiosis. The maternal chromosome has a large duplication (not present in the paternal chromosome). Assume (for the sake of simplicity) that the duplicated region DOES NOT include the centromere. Generate a drawing that displays the ways that the two chromosomes could align in meiosis.

There will be looping out of the duplicated region.

Q9 (5 points) Now, consider the effects of various crossing over events between these two chromosomes. Do any such crossing over events have negative effects?

Crossing over events will not have any negative effects; one chromosome will emerge with the duplicated region, the other will not. Of course the original duplication might have effects.

Q10 (6 points). There are a number of reasons that one, but not the other gene in a diploid organism might be expressed in a particular cell. Assuming that both alleles have exactly the same sequence, describe the various ways (at least two) that this is possible.

If the gene is on the X, then in a female, X-inactivation would lead to the gene on one chromosome being expressed in one cell lineage, while the gene on the other chromosome could be expressed in the other cell lineage.

Another alternative would be imprinting, such that only the maternal or paternal allele is expressed. Another alternative is that the effect could be stochastic, depending on which regulatory factors were present and how the effect the ability of the gene's promoter to be activated.

Q11 (5 points): Assume that the two alleles are not identical, what might be the effect on the cells or the organism.

If the alleles have different effects on cellular behavior, such as survival, rate of cell division, or the behavior of cells, then the cell's expressing different alleles will be different.

If the effect is on cell division, and the choice of allele is permanent (as in X-inactivation or imprinting) then there could be somatic evolution, with the adult made up of different percentages of cells expressing one or the other allele. The choice of allele and percentage of cells expressing it could also impact tissue function.

Q12 (6 points): Consider a female human being. Excluding the effects of newly arising mutations (that appear during the development of the organism), explain why not all of the cells of her body will be identical.

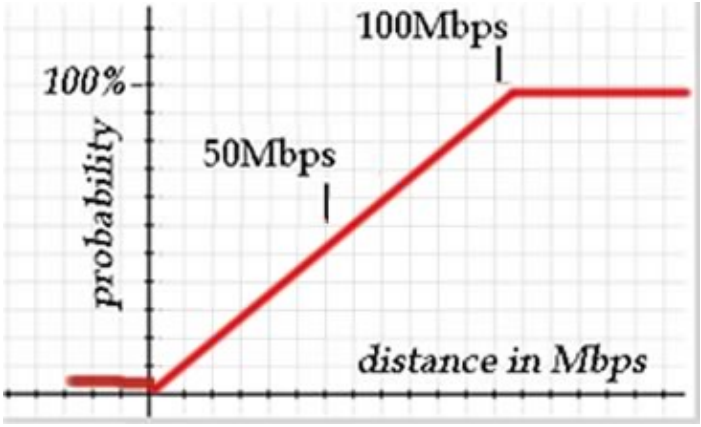
In a human female, only one of the two X chromosomes is expressed. There can be effects on cell behavior (see above) depending upon which chromosome is expressed.

Q13 (5 points): Explain why it is that males are more susceptible to some genetic diseases than females.

Human (and mammalian) males have only one X chromosome. If alleles of the genes on the X contribute to a disease's phenotype (or appearance), it is more likely that males are effectively homozygous (only one type of allele is present and expressed) compared to females, where there is a chance that the other X chromosome has a wild type (or non-disease) associated allele. Of course, it will depend (in the female) on which cells express which X chromosome.

Q14 (5 points) In a sexually reproducing eukaryotic organism, there is a 200 megabase long chromosome, its centromere is located in the middle of the chromosome. Consider three genes located on this chromosome. Two of these genes are located within 2 megabases on either side of the centromere, while the third gene is located 100 Mbps away from one of these two genes. Generate a schematic of the chromosome.

Q15. (6 points) Assuming that crossing over is suppressed in the region of the centromere. Generate a graph in which the centromere is located at X=0. On the Y axis, display the probability that one or more crossing over events occur between these genes (we can assume that each gene occurs in two different allelic forms in this individual so that crossing over events can be detected. Label the axes of your graph.



Q16 (6 points) An individual has two different alleles of a gene. Both lead to changes in a single amino acid in the coding region. How could a new allele with both of these amino acid changes in the same polypeptide be generated? Illustrate (with a drawing) and explain your proposed mechanism.

A crossing over event between the sites of the encoded amino acid changes would lead to 1 chromosome with neither change and one chromosome with both changes. (alternative: a rare mutation that generates the second amino acid change in a gene that already has the other change).

Q17 (6 points) Assume that the polypeptide with both animo acid changes provides a strong selective advantage (more offspring). Starting with organisms with neither amino acid change, which type of population would be expected to more rapidly (in terms of generation numbers) produce such a double changed polypeptide, a sexual or an asexual organism and explain why.

The sexual reproducing organism, because independent events in two different lineages can be combined (through fertilization). In an asexual organism, these two independent events would need to occur within the same lineage.

Q18 (4 points) Assume that organisms that are heterozygous for the double mutant polypeptide can survive infection by a virus, while homozygous individuals die before reproductive age. Predict whether the allele encoding the double mutant polypeptide will occur within a population, and what factors will influence its prevalence (the percentage of organism that carry it).

If the organism lives in a region where the virus is present (endemic) so that each individual can be expected to be infected, and infection has a high probability of leading to death or decreased reproductive potential, heterozygous individuals will be strongly selected for. If the disease is mild

or rare, they will not be expected to be selected for, but there will be strong negative selection against the homozygous individuals.