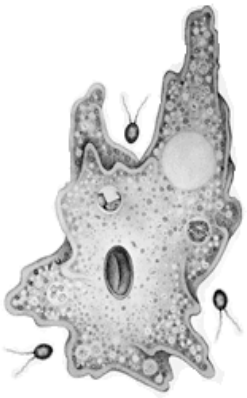


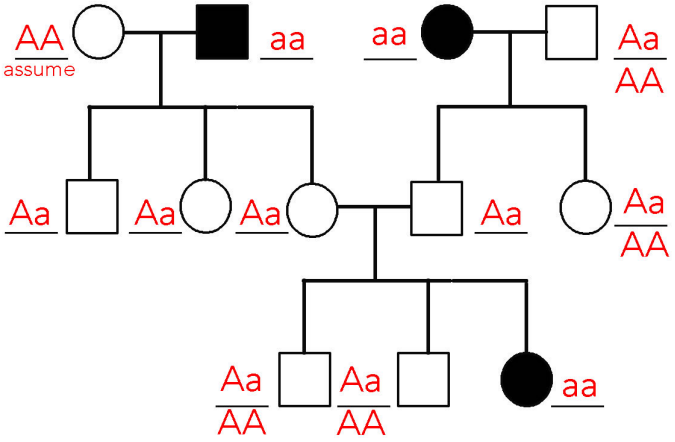
MCDB 2222 coreBIO II: FINAL - Spring 2018 NAME: _____

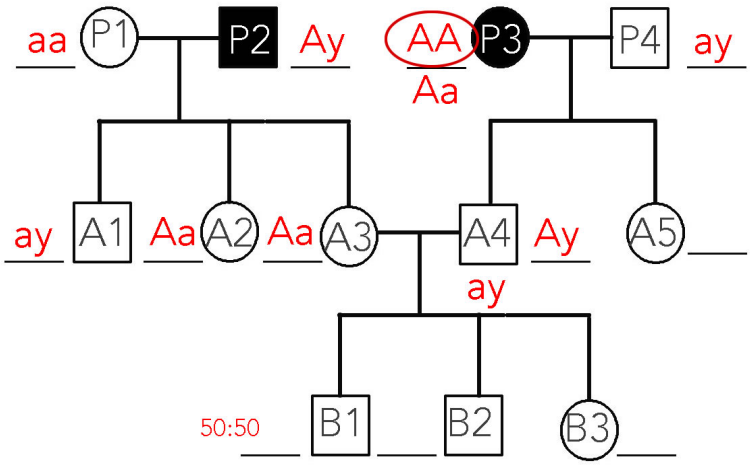
Directions: The exam is worth 150 points. Carefully read each question to determine what the question wants you to do.

I am taking ☐ IKIN 1 (midterm 1) ☐ IKIN 2 (midterm 2)
Each is worth up to 20 points added to your midterm score.

Q1 (10 points): Here is a pedigree, in which the individuals (■ ●) display the trait - the trait behaves in a simple Mendelian manner and is autosomal. What are the likely genotypes of the eleven individuals shown?

Is the allele determining the trait dominant or recessive?





Q2 (10 points): Consider how the pedigree might change if the allele for the trait were dominant and the gene for the trait was located on the X-chromosome. Remember that ■=male & ●= female.

1) predict whether A1, A2, and A3 display the trait.

2) to be 100% certain that A4 will display the trait, what **has** to be true for P3?

3) Assume that A4 **does not** display the trait, what is the probability that a B individual displays the trait?

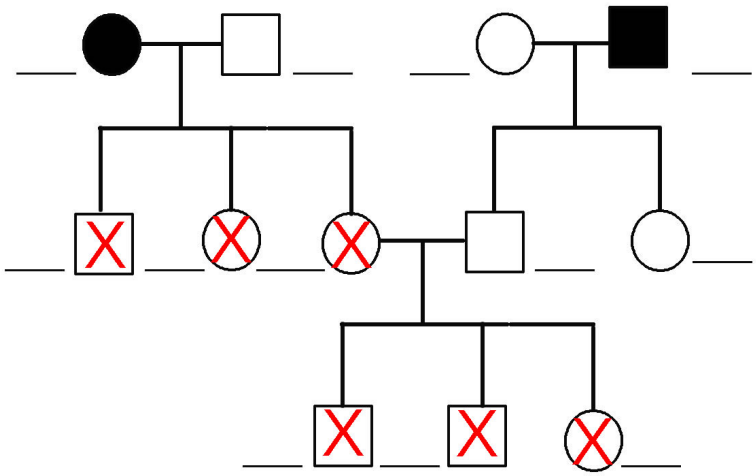
Writing out possible genotypes can help.

Q3: (10 points) What would happen to maternal inheritance if mitochondria did not have their own genetic material (DNA), and why exactly is it thought that they contain genetic material in the first place?

There would be no maternal inheritance associated with mitochondria - mitochondrial behavior would be determined by nuclear genes.

Mitochondria are assumed to be derived by previously free-living bacteria, which had their own genomes.

Q4: (10 points): You are considering a family tree in which a defect is inherited maternally. Two of the grandparents of this family display the trait. Fill in the symbols of the offspring who display the trait.



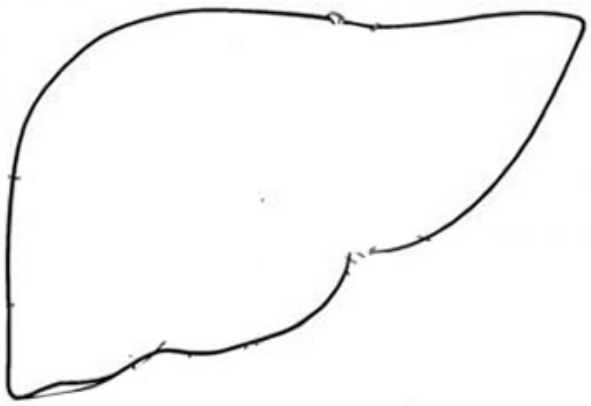
Q5 (10 points) Consider a gene on the X chromosome. You have identified an allele that fails to produce any RNA - what type of morph is that allele likely to be?

☐ neomorph ☐ hypermorph ☐ hypomorph ☐ antimorph X amorph ☐ impossible to predict.

What factor(s) will determine whether it behaves as a dominant or recessive allele?

It will be dominant in males (no wild type allele to rescue) and may be recessive in female, depending on whether the cells (50% of all cells) that express the wild type allele are sufficient to produce the wild type phenotype (assuming that the female is heterozygous).

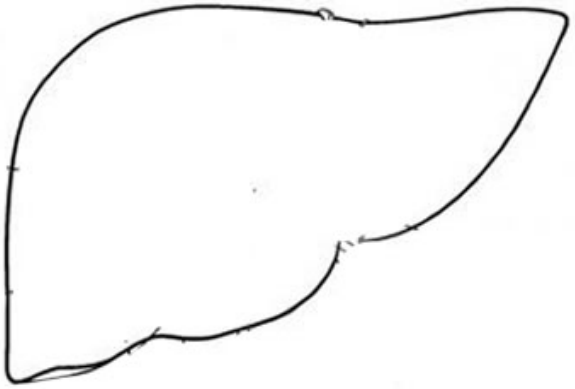
Q6 (10 points) The X-linked gene described in question 5 is expressed in all liver cells. In a post-mortem analysis of a woman heterozygous for this gene, you prepare sections of the liver and analyze gene expression using in situ hybridization with an anti-sense RNA probe. Describe a plausible pattern of gene expression that might be observed and explain your reasoning.



splotchy dependent on which X chromosome is act in which cells

↑ X-linked
↓ Autosomal

How (and why) would this pattern of gene expression be likely to change if the gene were located on an autosome (non-sex chromosome) and the individual were heterozygous for the allele?



It would like uniform, the wild type allele would lead to mRNA synthesis (it would be lower than that seen in a homozygous wild type however).

Q7 (10 points) A gene encodes a gene product that assembles into a functional (dimeric) protein. The polypeptide has no function as a monomer. You identify a mutant allele (MUT) that produces the same amount of gene product as the wild type allele (WT); but when the MUT polypeptide combines with itself (MUT/MUT) or the WT (MUT/WT) polypeptide, the resulting dimer is non-functional. In a heterozygous (MUT/WT) individual, the mutant allele acts as a
☐neomorph, ☐ hypermorph ☐ hypomorph ☐ antimorph ☐ amorph ☐ impossible to predict.

In a homozygous (MUT/MUT) individual the mutant allele acts as a
☐neomorph, ☐ hypermorph ☐ hypomorph ☐ antimorph ☐ amorph ☐ impossible to predict.

Explain the logic behind your answers. In the heterozygous case, the MUT gene produce would can to inhibit (though dimerization) some of the WT gene product - reducing the level of active dimers (an antimorphic effect).

In the homozygous MUT/MUT case, no functional dimer could form, an amorphic outcome.

Q8 (10 points): A mutation arises in the soma of an organism; this mutation has two effects - it has a dominant phenotype that results in cells dividing more rapidly than wild type cells, and a recessive phenotype that leads to an increase in mutation rate. Describe how you might expect cells carrying this mutation will behave over time, particularly if resistance to the drugs used to control cell division arise through mutations in other genes?

Initially the cell would divide more rapidly can surrounding normal cells, with each division the chance of other mutations would increase - if a second mutation occurred that would lead to an increase in overall mutation rate (homozygous for recessive phenotype). This could increase odds that cell's carrying the original mutation could development drug resistance.

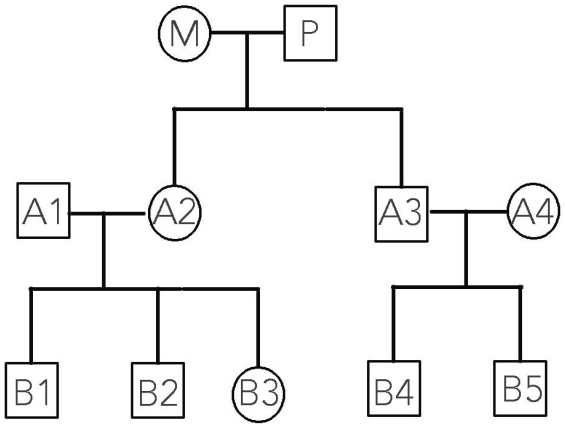
Q9 (10 points): You are interested in determining whether a person has inherited a mutated allele of a particular gene. You decide to use reverse transcription and PCR. The wild type gene is ~8 kilobases long, whereas the mutant gene has a internal deletion that removes the middle 4 kilobases of the gene. Draw a schematic of the wild type and mutant genes.

You design PCR primers that enable you to amplify the gene and you have a method to determine the size of the amplified region. On your schematic of the gene, indicate where in the gene your PCR primers would bind, and give an estimate of the size of the amplified PCR product (DNA) that you would expect to be generated from cells homozygous or heterozygous for the mutant allele.

one product would be long the other would be short (4 kb shorter than the wild type) assuming your PCR primers flanked the region of the deletion.

Q10 (10 points): Consider an autosomal gene that is maternally imprinted, so that the allele inherited from mom (the maternal allele) is not expressed. Assume that mom (M) was homozygous for a dominant allele of a gene that leads to blindness at birth; there is no history of congenital blindness in P's family.

1) is Mom (M) blind? yes
2) Will either of her children (A2 or A3) be blind? explain your reasoning.



no, because the dominant allele she passes down is imprinted and so not expressed.

Q11 (5 points): Which grandchildren (B1-5) have a possibility to inherit an active, dominant blindness allele? explain your reasoning. yes, both B4 and B5 because they could inherit the allele from Dad - where it is not imprinted (and so is expressed)

Q12 (10 points): You discover a SNP that maps near a gene's coding region. The presence of the SNP correlates with a trait (cold insensitivity + frequent migraines). The coding regions of the nearby genes appear identical. How might the polymorphism effect the gene?

The SNP could influence expression of these genes

Q13 (10 points): When someone says that an allele displays incomplete penetrance, what does that mean – consider this in the case of a dominant allele. Does the penetrance of an allele influence whether it is passed on to the next generation? Explain your thinking.

It would mean that because you have a dominant allele you do not necessarily express the trait, presumably due to factors elsewhere in your genome.

It does not influence whether you pass the allele on to the next generation.

Q14 (10 points): You have generated two pure breeding mouse lines that are either large and black or small and white. When you cross these animals you discover the F1 generation is uniformly medium sized and grey. Now you cross the F1 animals - what phenotypes and genotypes, and in what ratios, would you expect to find. In order to be able to make such predictions, what assumptions do you need to make?

You would have to assume that they are unlinked in order to make a prediction.

In a typical cross you would get 9:3:3:1, but this is for fully penetrant dominant and negative alleles

for this case of co-dominance in both traits, the result would look like (16) 1: large:black
2: large:grey
2: small: grey
2: med:black
2: med: grey
2: med: white
2: small: grey
1: large: white
1: small: black
1: small: white

L - large, l - small, B - black, b- white

F1 Ll Bb

	LB	Lb	lB	lb
LB	LLBB large, black	LLBb large, grey	LlBB med, black	LlBb med, grey
Lb	LLBb large, grey	LLbb large, white	LlBb med, grey	Llbb med, white
lB	LlBB med, black	LlBb med, grey	llBB small, black	llBb small, grey
lb	LlBb med, grey	Llbb med, white	llBb small, grey	llbb small, while

Q15 (15 points): A. What factors impact the ability of a person to modify their own somatic cells, for example their brain, using CRISPR or some future technology? The ability of get the modifying reagents to the correct cells, and to influence a high enough percentage of the cells

B. Should people be free to genetically modify their own germ lines, or should such modifications be restricted to their somatic tissues. Briefly explain your thinking.

C. Should people be free to genetically modify the DNA of their children or their pets? Briefly explain your thinking.

D. Which aspects of genetic engineering be regulated by the legal system? Briefly explain your thinking. (answers can be continued on the next page).

Question 15: More space to write if you need it.

IKIN 1: (10 points) In a eukaryote, a deletion mutation on a chromosome leads to the inability to form a centromere and the loss of a set of genes essential for cell survival. Draw a picture of how such a mutant chromosome would behave during mitosis and the possible effects on the cells' offspring.

IKIN 2: (10 points) Provide a short description of a condition under which you would **not** expect sexual dimorphism to arise. How does sexual dimorphism influence a process such as pregnancy (for example, in humans)?

IKIN 2:1 (5 points): How might an neomorphic allele behave in a dominant manner.

IKIN 2:2 (**5 points**): How might an antimorphic allele behave in a dominant manner.

IKIN 2:3 (10 points): You are carrying out a cross with simple mendelian (dominant and recessive) alleles of genes that control two distinct traits, skull size (large or small) and hair (bald or luxuriant). You generate F1 individuals that are all large + bald. When you cross F1 individuals you get

large head + bald	2805
large head + luxuriant hair	420
small head + bald	385
small head + luxuriant hair	2850

What were the phenotypes of the parental (original stocks) and are the genes responsible for these traits linked or not? explain your reasoning.