**REVIEW QUESTIONS FOR MIDTERM 1 FALL 2013**

**The only places you can get feedback on these questions are in the office hour sessions we will hold Sept 30 – Oct 4th or on the discussion board with you colleagues. These questions will not be covered in tutorial as there is not enough time and you need to cover the material in the Week 5 tutorial.**

**1.** Loppins (*Loppinicus loopy*) are fictitious, but very useful diploid invertebrates with a total of 6 chromosomes in their somatic cells. Of those 6 chromosomes, 4 are autosomes and 2 are sex chromosomes. Like humans, male loppins are XY while females are XX.

The gene that determines loppins' blood type is called *bt* and is on chromosome 1, the gene that determines the presence or absence of eyelashes is called *eye* and is on chromosome 2, and the gene that determines ability to digest cellulose is called *cel* and is on the X chromosome.

$bt^A$, $bt^B$; $eye^W$, $eye^D$, *celWT* and *celM* are alleles of these three genes.

a) A female loppin is a triple heterozygous with the genotype $bt^A/bt^B$; $eye^{WT}/eye^D$; $X^{celWT}/X^{celM}$.

Her mother was homozygous for $bt^A$, for $eye^D$ and for *celWT*.

Draw a somatic cell of our triple heterozygous female loppin in G1 stage of the cell cycle (that is, before DNA replication). Make sure that the chromosomes are properly drawn and clearly label all the relevant genes and alleles. **(4 marks)**

**b)** What alleles did the triple heterozygous female loppin inherit from her mother, and what alleles did she inherit from her father? **(2 marks)**

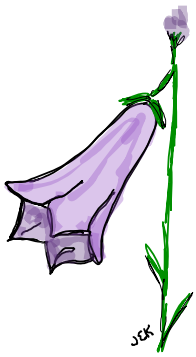
c) For research purposes you remove three meiocytes from the triple heterozygous female, you let them undergo the cell cycle and meiosis, and you analyze the genotype of the gametes that are produced. The first meiocyte produces two gametes of genotype $bt^{A}$; $eye^{WT}$; $X^{celWT}$ and two gametes of genotype $bt^{B}$; $eye^{D}$; $X^{celM}$.

i) Draw this meiocyte at metaphase of meiosis I (this is when the homologous chromosomes are paired and lined up in the centre of the cell). Make sure to clearly label all the relevant genes and alleles. **(3 marks)**

ii) The second meiocyte produces two gametes of genotype $bt^{B}$; $eye^{WT}$; $X^{celWT}$ and two gametes of genotype $bt^{A}$; $eye^{D}$; $X^{celM}$. Explain what must have happened differently in this meiocyte compared to the one in part i) to produce this result. You may refer to the diagram that you drew above to illustrate your rationale. **(1 mark)**

iii) The third meiocyte produces one gamete of genotype $bt^{A}$; $eye^{WT}$; $X^{celWT}$, one gamete of genotype $bt^{B}$; $eye^{WT}$; $X^{celWT}$, one gamete of genotype $bt^{B}$; $eye^{D}$; $X^{celM}$ and one gamete of genotype $bt^{A}$; $eye^{D}$; $X^{celM}$. Briefly explain what must have happened to produce this result. You may refer to previous drawings if you need to. **(1 mark)**

iv) If we looked at the gametes produced by 100 different meiocytes from this triple heterozygous female loppin, what are all the genotypes that we would expect to find, and in what proportions? *(Note: 100 different meiocytes will produce a total of 400 gametes)*

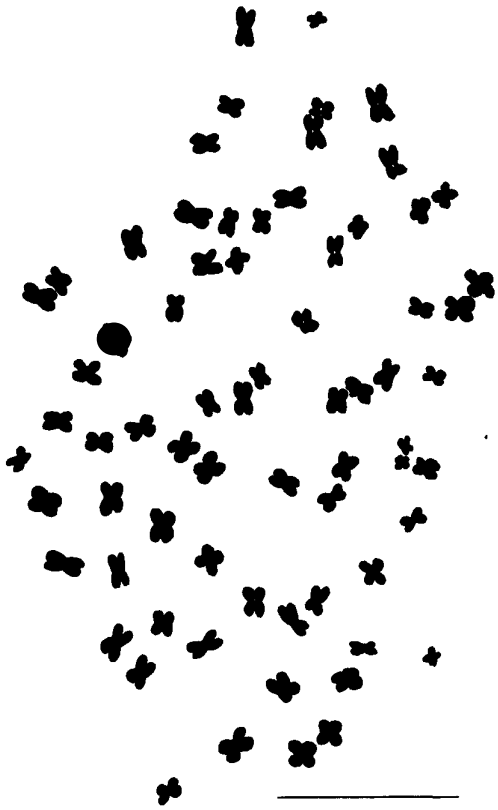


2. Bluebells (*Campanula rotundifolia*) are a common wildflower that have purple bell-shaped flowers. You find a bluebell plant growing on the edge of Pacific Spirit Park, but it has white flowers rather than purple.

*a) Suggest one possible environmental cause that made the flowers white. You don't need to know much botany. (2 marks)

b) Suggest one genetic cause that made the flowers white. It's ok if you don't know much biochemistry. (2 marks)

c) Design an experiment to distinguish between an environmental versus a genetic cause of the white flower phenotype, and explain how you will interpret the data (i.e.: **explain how the results of your experiment will allow you to distinguish between environmental and genetic causes**.) Your answer should be point form and clear, it shouldn't take up a lot of space. (4 marks)



3. The image on the right shows all 68 chromosomes from one root tip cell of *Campanula rotundifolia* after DNA replication.

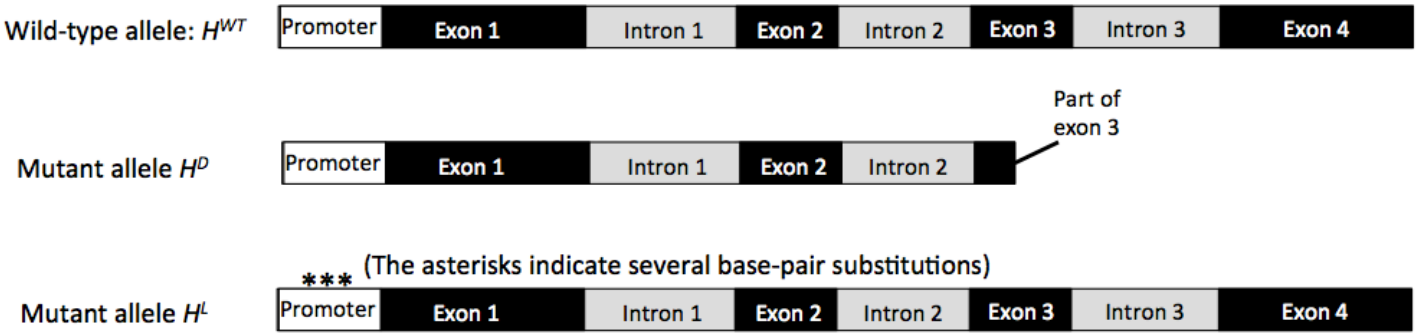
a) How many chromatids does one of these chromosomes have? (1 mark)

b) How many DNA double helices is each one of chromosomes made of? (1 mark)

c) It is very difficult to recognize homologous chromosomes on the picture, but knowing that *Campanula rotundifolia* is diploid, how many pairs of homologous chromosomes are present in one of its root tip cells? (1 mark)

d) Draw a clear diagram of two homologous chromosomes from *Campanula rotundifolia* <u>before</u> DNA replication. (2 marks)

4. One of the genes involved in determining blue flower colour in *Campanula rotundifolia* codes for an enzyme called hydroxylase. The diagram below shows the simplified structure of the wild-type allele of the *hydroxylase* gene (hydroxylase converts a chemical compound into a purple pigment) and the changes in DNA sequence that were detected in two mutant alleles of this gene.

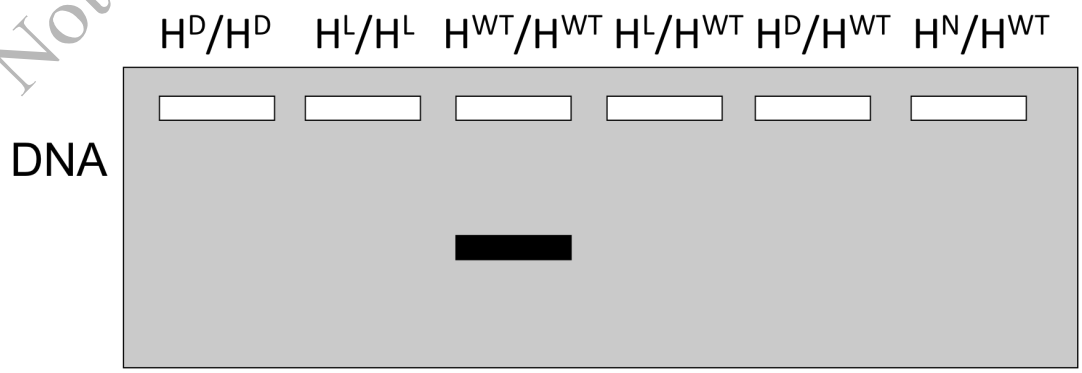


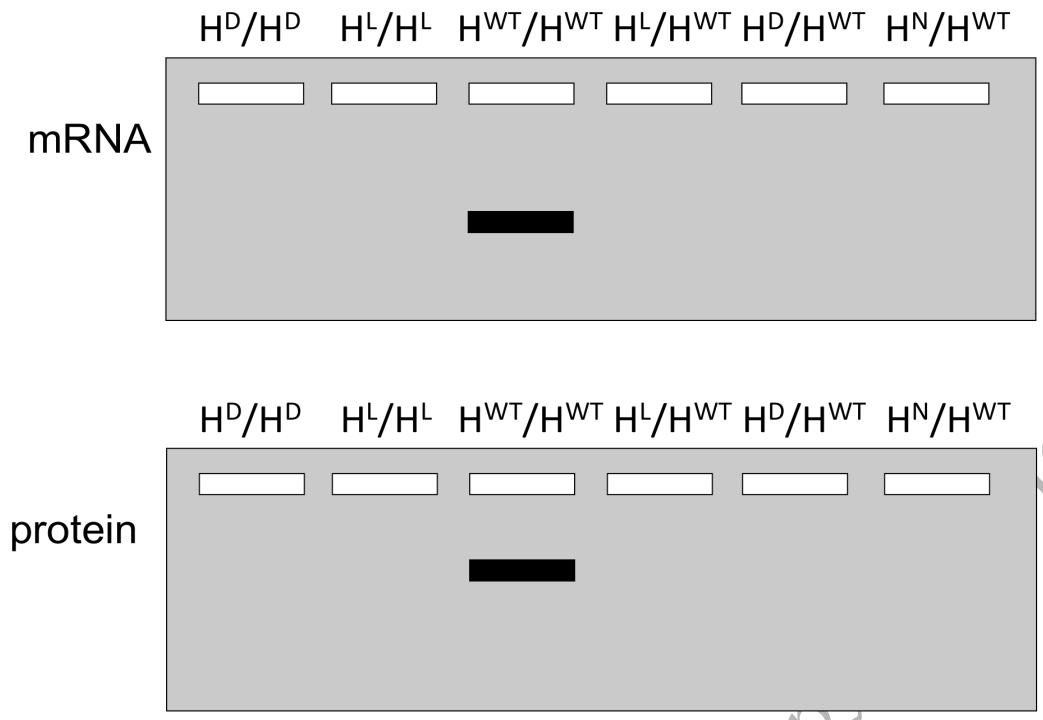

b) Predict the relationship between the $H^D$ and the wild-type ($H^{WT}$) alleles of the *hydroxylase* gene if both alleles are present in one organism and the phenotype of interest is "Flower colour". Briefly justify your answer. (2 marks)

c) In the lab, you have generated a novel allele of the *hydroxylase* gene (that you call $H^N$) that has a single base-pair substitution in exon 3 compared to the wild-type. This results in a single amino acid difference between the wild-type Hydroxylase enzyme and the enzyme encoded by your $H^N$ allele.

Surprisingly, *Campanulae* that are homozygous $H^N/H^N$ make flowers that are a much darker purple than the wild-type colour. Explain how this may be possible. (2 marks)

**d)** Draw a DNA, RNA, and protein gel with bands of DNA, RNA, and protein from the following genotypes: $H^D/H^D$, $H^L/H^L$ , $H^L/H^{WT}$, $H^D/H^{WT}$, and $H^N/H^{WT}$. $H^{WT}/H^{WT}$ is done for you. **(See next page also) (**7.5 marks….*0.5 marks per correct banding pattern*)






**5**. Imagine a spontaneous mutation occurs in a cell that is n = 3. This mutation is a 20 bp deletion within a centromere sequence of one chromosome.

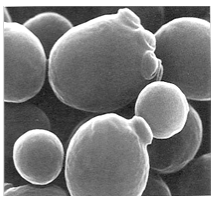
a) What are the phenotypes (in terms of chromosome number) of possible daughter cells from mitosis if the mutation prevents chromatids of this one chromosome from being held together after DNA replication. Draw the different possible daughter cells with the chromosomes they would contain.

b) The n=3 cell with the spontaneous centromere mutation is a gamete, and this gamete fused with another, normal n=3 gamete to produce a diploid cell (2n=6).

If this 2n=6 cell is to then undergo mitosis to grow and develop will it be viable? Explain and use drawings to aid in your explanation.

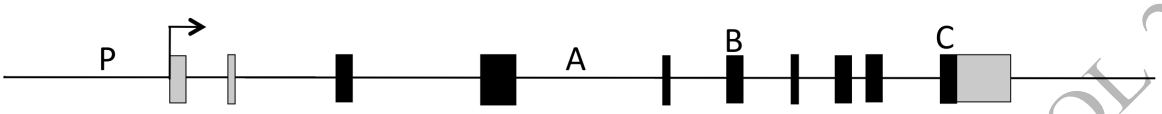
**6.** Arbutus trees are native to the Pacific Northwest coast. You find an Arbutus tree, that unlike normal trees has many dark spots on the leaves. Briefly outline your strategy to determine whether these spots have a genetic or environmental basis.

**7.** Below is shown the gene structure of the gene *isocitrate dehydrogenase* (*ic*) from yeast *Saccharomyces cerevisiae* . Based on sequence analysis there are no known enhancers in this gene. The protein coded for by this gene is an enzyme within the citric acid cycle operating in the mitochondria that converts isocitrate to alpha ketoglutarate. The ultimate purpose of this cycle is produce reduced NADH which is then converted to energy through other major metabolic reactions. Yeast cells that cannot produce NADH through this cycle then have to perform anerobic fermentation to obtain energy.



*Saccharomyces cerevisiae*

The locations of 3 different mutations are shown.




Mutation A is an insertion of 2 nucleotides in the middle of the 4th intron.
Mutation B is the deletion of 4 nucleotides in the 6th exon, which also happens to be part of the active site of the enzyme.
Mutation C is the substitution of 1 nucleotide which causes a missense mutation in the last exon and causes the enzyme to be found in the cytoplasm rather than the mitochondria where it is normally found.

a) In the table below explain what the phenotype of each of these mutations will likely be (when homozygous) at the different levels:

| genotype | mRNA (sequence, size amount) | Protein (sequence, size, amount, function) | Whole organism (single yeast cell) |
|---|---|---|---|
| $ic^A/ic^A$ | | | |
| $ic^B/ic^B$ | | | |
| $ic^C/ic^C$ | | | |

b) If the normal job of isocitrate dehydrogenase is to convert isocitrate into alpha ketoglutarate explain whether or not each mutation could be rescued by adding alpha ketoglutarate to the cell (and we are assuming the alpha ketoglutarate makes it into the mitochondria)

c) Rank and explain the relative severity of the three heterozygous allele combinations. State any assumptions you are making.

$ic^A/ic^B$;

$ic^A/ic^C$;

$ic^B/ic^C$

**8.** In hogs, a dominant allele *B* results in a white belt around the body. At a separate locus the dominant allele *S* causes fusion of the two parts of the normally cloven hoof resulting in a condition known as syndactyly. A belted syndactylous sow was crossed to an unbelted cloven-hoofed boar, and in the litter there were:

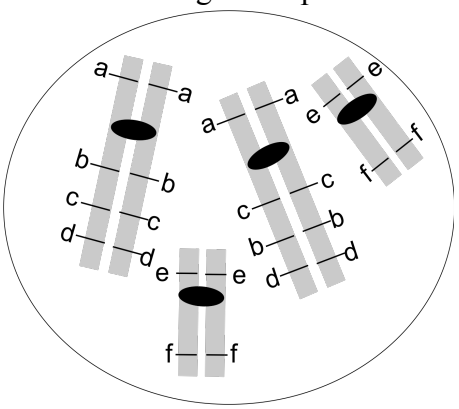
18 belted syndactylous
21 belted cloven
19 unbelted syndactylous
20 unbelted cloven

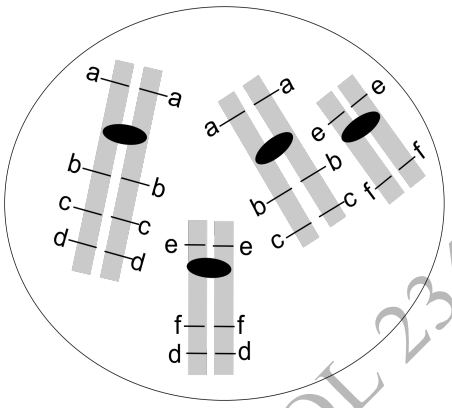
a. Write out the possible genotypes for the litter phenotypes listed above.

b. What are the parental genotypes? Show all your work and indicate what part of your work verifies that your hypotheses are correct.

c. If two belted syndactylous animals were mated, what would you expect in the F2?

**9.** The following two diploid cells have their genes labelled onto the chromosomes as letters a - f.






a) i) What type of a mutation has occurred to create one of the large chromosomes in the left cell?

ii) What type of mutation has occurred in the cell on the right?

b) Draw all the different possible gametes that could be created by each cell underneath the cell. Assume there is no crossing over between homologous chromosomes.

c) Do each of these gametes above that you drew above contain a complete genome's worth of DNA? Label each gamete as "complete", "incomplete" or "more than 1 complete genome" as necessary.

**10. Chapter 6 #23**
In a maternity ward, four babies become accidentally mixed up. The ABO phenotypes of the four babies are known to be: baby #1 O, baby #2 A, baby #3 B, and baby #4 AB. The ABO phenotypes of the four sets of parents are:
Parents:
(a) AB × O

(b) A × O

(c) A × AB

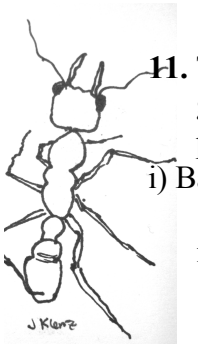
(d) O × O

a. Analyze this information to predict which parental set (a-d) could be the parents of each baby. Show your work, and then complete the table below. Use the following symbols for alleles: O – $i^O$, A – $i^A$, B – $i^B$,

| Phenotype | Genotype or possible genotypes Possible parents |
|---|---|
| Baby #1 - O | |
| #2 – A | |
| #3 – B | |
| #4 - AB | |

b. For baby #4 (B blood type phenotype), explain and/or show the work you have done to confirm that your hypothesis about the parental and baby #4 genotypes are correct.

c. Draw a cell from an AB parent in **prophase** and **anaphase of meiosis I** (separation of homologous chromosomes). Label chromatids with alleles. What are the resulting gamete genotypes, and in what proporations, from this meiotic division?

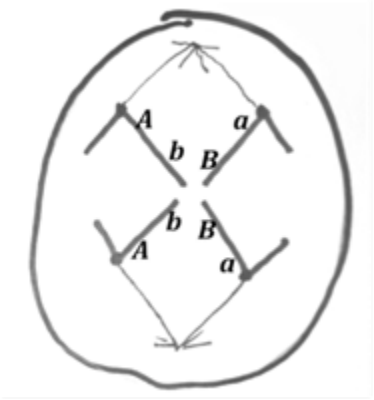



**11.** The jack jumper ant (*Myrmecia pilosula)* has the chromosome number 2n = 2. An ant has the genotype *a B/Ab* (the A and B loci are on the same chromosome). It was produced from two pure-breeding parents with the genotypes *a B/a B* and *A b/A b* respectively.

i) Based on the structures separating, what stage of meiosis this cell is. (1 mark)

ii) Given your answer to part i, what is incorrect about this cell, and what, if anything is correct. (3 marks)

b) For the stage shown in a) draw what the meiotic cell SHOULD correctly look like for the ant described above. (3 marks)




**12. Modified from Chapter 2 #44**

The ability to taste the chemical phenylthiocarbamide is an autosomal dominant phenotype, and the inability to taste it is recessive. A taster woman with a nontaster father marries a taster man who in a previous marriage had a nontaster daughter. The taster man and woman are going to have a child. What are the possible genotypes and phenotypes, including probabilities, of their child?

**13. Chapter 3 #29**

In tomatoes, two alleles of one gene determine the character difference of purple (P) versus green (G) stems, and two alleles of a separate, independent gene determine the character difference of "cut" (C) versus "potato" (Po) leaves. The results for five matings of tomato-plant phenotypes are as follows:

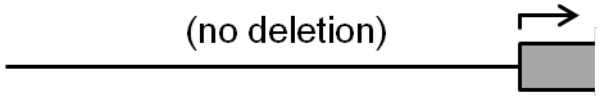
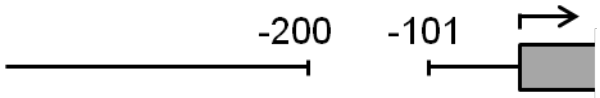
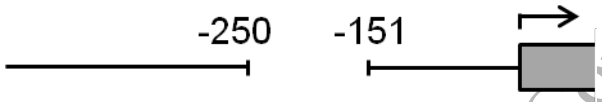
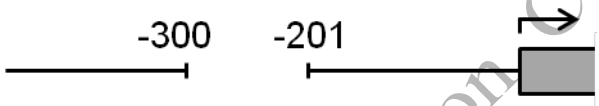
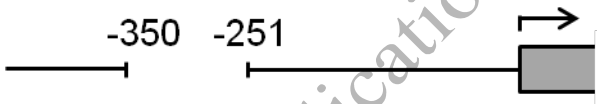
| | | *Number of progeny* | | | |
|---|---|---|---|---|---|
| Mating | Parental phenotypes | P, C | P, Po | G, C | G, Po |
| 1 | P, C × G, C | 321 | 101 | 310 | 107 |
| 2 | P, C × P, Po | 219 | 207 | 64 | 71 |
| 3 | P, C × G, C | 722 | 231 | 0 | 0 |
| 4 | P, C × G, Po | 404 | 0 | 87 | 0 |
| 5 | P, Po × G, C | 70 | 91 | 86 | 77 |

****It's recommended that you come up with new gene and allele symbols that you find easy to keep track of - the P/G and C/Po is notoriously annoying.*

a. Determine which alleles are dominant. What are the most probable genotypes for the parents in each cross?

a. For at least one cross s*how how you determined the dominance relationships, and how you checked/proved that your hypothesis is correct.*

**14.** To test for the presence and the location of suspected enhancer elements upstream of the *atm* gene, we generated 5 deletion alleles (*a* to *e*). These deletions take out a part of or all of the sequence from the 101st to the 350th nucleotide upstream of *atm*'s transcriptional start site. The amount of *atm* mRNA is measured for the wild-type (*WT*) and each of the five deletion alleles. The results are summarized below.

| **Allele** | **Change to the *atm* upstream region** | **Deletion size (bp)** | **Relative mRNA amount** |
|---|---|---|---|
| *WT* |  | 0 | ++++ |
| *a* |  | 250 | *atm* mRNA not detectable |
| *b* |  | 100 | ++ |
| *c* |  | 100 | ++ |
| *d* |  | 100 | ++++ |
| *e* |  | 100 | ++ |

Is there evidence of enhancer elements upstream of *atm*? If so, where are the enhancer elements located?