

BIO 2133 (Dr. Montpetit)
Take Home Test #1 (35 Points)
Due – February 3rd 830am - midnight.

INSTRUCTIONS:

1. Please download a copy of the exam. You have the option to use the **WORD or PDF version.**
2. This exam is open book. Answer **ALL** questions as best you can with the information on hand.
3. This test must be completed individually. Should two or more students submit identical answers or answers that constitute plagiarism in any form, all students with those answers will face allegations of academic fraud. We are not using **Respondus** or any other lockdown software.
4. The test is designed to be written in a **90-minute block of time**; however, you have up until **midnight** to submit your answers to the test.
5. You can answer questions in any way you choose/are able. This includes but is not limited to:
 - a. Printing the exam, filling it in by hand, then scanning/photographing it.
 - b. Answering each question by hand on a blank paper, then scanning/photographing it.
**You do not need to recopy the questions to your sheet of paper but be sure to clearly indicate which question you are answering and if you must copy sequences/images/etc... to your sheet, do so very carefully to avoid making errors.*
 - c. Handwriting your answers digitally. (e.g. with an Apple pencil or stylus capable of writing in e-ink)
 - d. Typing your responses directly into the file.
6. You may ask questions via email. They will be answered only during regular business hours (9am to 5pm). Only questions of a technical nature (i.e. – submission issues) will be answered in the final 30 minutes of the exam. Please do not expect quick answers given the size of the class.
7. Please name and save the digital file (**as a PDF file- ONLY**) containing your completed exam **student#_BIO2133_surname.filetype** (e.x - I would name my completed exam – 12345678_BIO2133_Montpetit.pdf).
8. You must submit your completed exam by **midnight on Feb. 3rd**. **This is a hard deadline.**
 - a. Upload **a single file** (.pdf) of the entire completed exam by **midnight on Feb. 3rd**.
 - b. You will receive an email confirmation of the receipt of your submission.
 - c. If you experience technical difficulties resulting in you missing the submission deadline, please contact me before the deadline to let me know (colin.montpetit@uottawa.ca).
 - d. Late submissions will not be assessed and considered an absence.

Helpful Hints and Tips:

- If completing the exam digitally, it is recommended that before beginning to write, that you name and save the document. I also recommended that you activate any automatic save function that your software may have.
- If scanning/photographing your exam to digitize a handwritten file, you may find it helpful to use a free app/program such as clear scanner (for [android](#) or [iPhone](#)) or [pdf merge](#) (web based) to compile

multiple pages into a single file. Please try to create a PDF file prior to the exam, in order to test out your ability to successfully create a PDF file and to avoid any issues when trying to submit your exam.

Please use 12pt Times New Rom font if you type your answers.

Write your answers in the space provided.

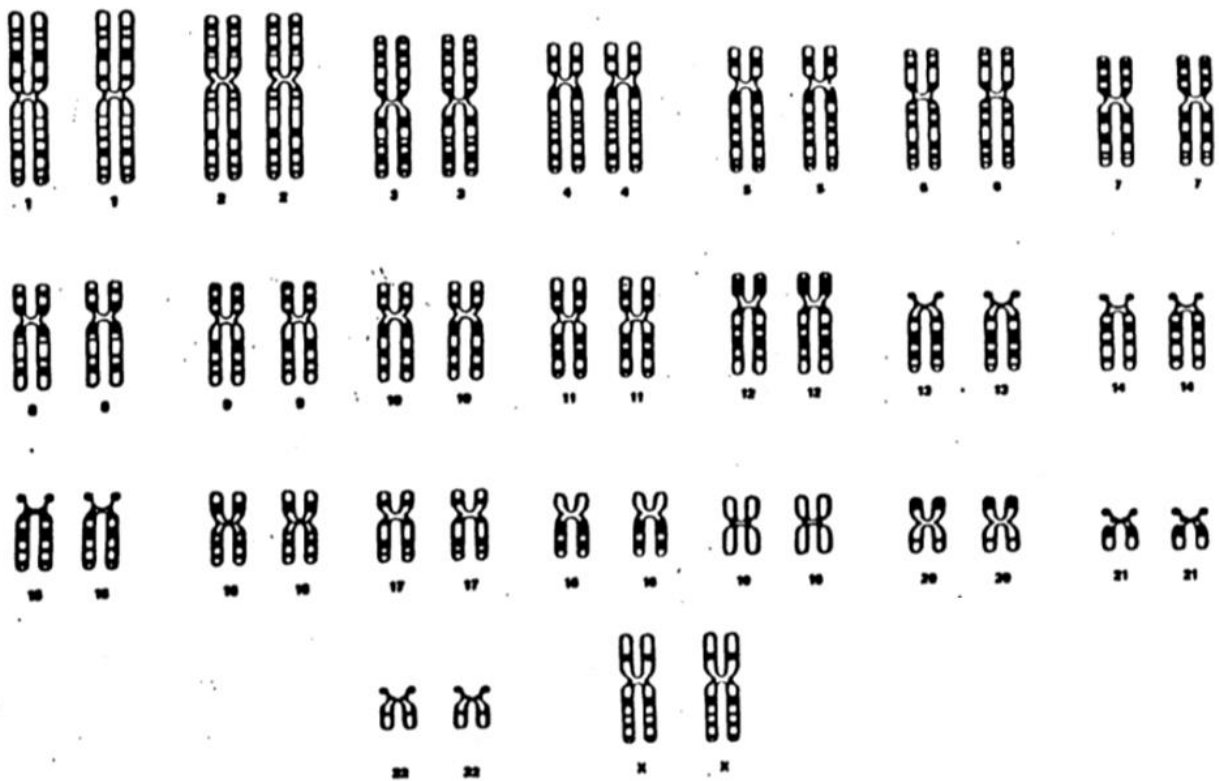
- The figure below shown below is from a human individual. This karyotype was generated by collecting the chromosomes following duplication and condensation, but prior to entering mitosis.

This particular individual is:

- Heterozygote for the leptin gene: O/o
- Homozygote dominant for the tyrosinase gene: C/c
- Afflicted with an autosomal recessive form of the Ehlers-Danlos condition (Gene E)

Suppose that the leptin and tyrosinase genes are linked; The gene for Ehlers -Danlos is not linked to the leptin and tyrosinase genes.

Using labels directly on the karyotype, provide a representation of the location of the leptin, tyrosinase and Ehlers-Danlos genes and alleles on the chromosomes and sister chromatids in such a way **that is consistent** with the scenario described above. Choose the chromosomes of your liking to show an appropriate representation (3 marks)



2. Priyanka, an undergraduate research assistant, recently identified a new allele linked to cystic fibrosis. Genetic tests show that this form of CF is inherited in an autosomal dominant fashion. DNA sequencing revealed that this new allele has a point mutation in the 3' untranslated region and causes the mRNA to degrade at a very high rate. This is unlike the $\Delta 504$ mutation which causes an autosomal recessive form of cystic fibrosis. Priyanka's study also demonstrated that 0.01% of the population is tested positive for this new allele.

Based on this information, answer the following questions:

- a. How many individuals in the population has the Cystic Fibrosis (CF) gene and why are some individuals afflicted with CF while others are not? Explain your reasoning. **(2 marks)**
- b. Every individual who tested positive for this new allele for the CF gene are heterozygote (they have the new allele and a copy of the wild-type allele). Provide a genetic mechanism that could explain why this new allele causes CF in an autosomal dominant fashion. **(2 marks)**

3. You are studying a gene called COL5A1. You discover a deletion of 1 nucleotide in a variant of the COL5A1 gene. You are about to conduct experiments to compare the lengths of the wild-type COL5A1 protein and the mutant COL5A1 protein gene when your supervisor asks for your predictions.

List 3 predictions and for each provide your reasoning (**6 points**)

4. Joanne is about to have a baby. Ultrasounds and a karyotype have revealed that their child is trisomic for chromosome 21. Joanne asks the genetic counsellor if the chromosome nondisjunction occurred in her gamete or the biological father's and in which stage of meiosis did it occur.

To answer the question, a DNA test determined the presence or absence of the **STR** (short tandem repeat) **marker D21S11** in Joanne and the baby. D21S11 is located on chromosome #21 and 4 variants of the marker (A1, A2, A3 and A4 variants) can be found in the population.

The DNA test revealed the following:

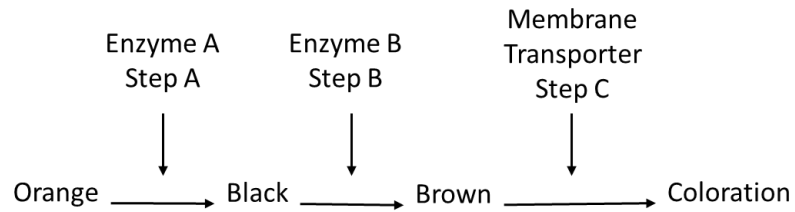
- Joanne – A₂A₄
- baby – A₁A₂A₃

Based on the test results, what is your conclusion and explain your reasoning. **(4 marks)**

5. During translation, the tRNA molecule carrying the correct amino acid corresponding to its anticodon sequence must base-pair bind with the codon of the mRNA. What would happen in the case where a tRNA carrying its correct amino acid incorrectly binds with the codon of the mRNA? Would this be considered a mutation? Explain your reasoning. **(3 marks)**

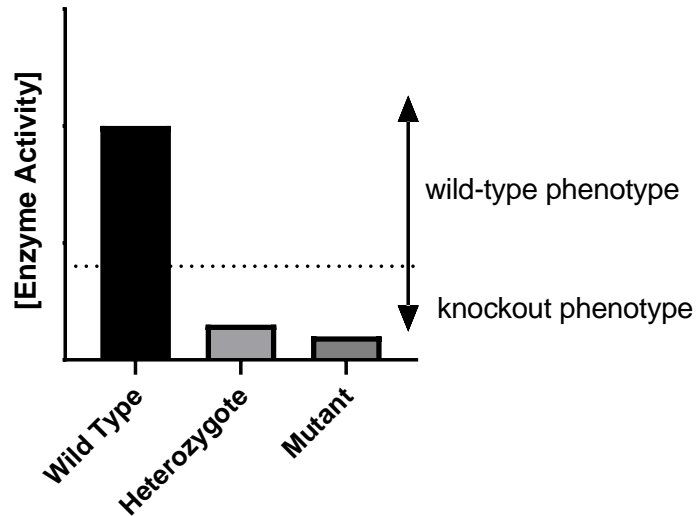
6. Wild-type corn snakes are brown, however, autosomal recessive mutations in the steps of colour determination can result in either orange, black or brown coloration or no colouration (white). Assume that the dominant alleles are haplosufficient.

The biochemical pathway for colour determination is the following:



- a. Assuming no confounding factors, which genotype will result in a phenotypically black corn snake? (use a clearly defined annotation system to indicate a genotype) (2 mark)
- b. Assuming that the transcription of enzyme A mRNA is completely inhibited by a transcription factor, what will be the snake's phenotype? (1 mark)
- c. Assuming a dysfunctional transporter, what will be the colour? (1 mark)

7. Knockout mice are mice that have gone through genetic modification to inactivate certain genes that codes for an enzyme. One method of achieving knockout is through homologous recombination (you do not need to know how this is done for the purposes of this question). After breeding a homozygous wild-type mouse with a homozygous knockout mouse, you find the following results:



- a. Based on the results, what conclusions can you draw about this gene? (1 marks)
- b. How would you annotate the alleles to represent the genotypes of the wild-type, heterozygotes, and homozygote mutants? (2 marks)
- c. Assume there is a third allele of this gene. A heterozygote mouse with a copy of the wild-type allele and this third allele displays an enzyme activity that is lower than the homozygous wild-types but is above threshold for normal activity. What would be the order of dominance of the 3 alleles? (in other words which allele is the most dominant and so on) – rank the alleles with a 1 (most dominant), 2 or 3 (most recessive): (2 marks)

Wild type: Knockout allele: 3rd allele:

3' TAC CTA CTG AGT ATT AAA TAC TGA GTA ACT 5'
5' ATG GAT GAC TCA TAA TTT ATG ACT CAT TGA 3'

8. Consider the DNA molecule above, assuming no introns: **(total 6 marks)**

a. How many reading frames can you identify? _____ **(1 mark)**

b. How many open reading frames can you identify? _____ **(1 mark)**

c. Predict the amino acid sequence of each open reading frames you have identified:
(4 marks – be aware that the number of marks does not necessarily reflect the number you need to identify).