

**Part A:** Please check that you have correctly bubbled your student number and course code on the scantron. Choose one answer for each question. (1 mark each)

		Second base of codon				
		U	C	A	G	
First base of codon	U	UUU } Phe	UCU } Ser	UAU } Tyr	UGU } Cys	U
		UUC } Phe	UCC } Ser	UAC } Tyr	UGC } Cys	C
		UUA } Leu	UCA } Ser	UAA } Stop	UGA } Stop	A
		UUG } Leu	UCG } Ser	UAG } Stop	UGG } Trp	G
	C	CUU } Leu	CCU } Pro	CAU } His	CGU } Arg	U
		CUC } Leu	CCC } Pro	CAC } His	CGC } Arg	C
		CUA } Leu	CCA } Pro	CAA } Gln	CGA } Arg	A
		CUG } Leu	CCG } Pro	CAG } Gln	CGG } Arg	G
	A	AUU } Ile	ACU } Thr	AAU } Asn	AGU } Ser	U
		AUC } Ile	ACC } Thr	AAC } Asn	AGC } Ser	C
		AUA } Met	ACA } Thr	AAA } Lys	AGA } Arg	A
		AUG } Met	ACG } Thr	AAG } Lys	AGG } Arg	G
	G	GUU } Val	GCU } Ala	GAU } Asp	GGU } Gly	U
		GUC } Val	GCC } Ala	GAC } Asp	GGC } Gly	C
		GUA } Val	GCA } Ala	GAA } Glu	GGA } Gly	A
		GUG } Val	GCG } Ala	GAG } Glu	GGG } Gly	G

**KEY**

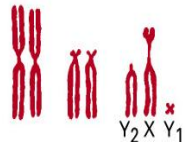
Ala = alanine
Arg = arginine
Asn = asparagine
Asp = aspartic acid
Cys = cysteine
Gln = glutamine
Glu = glutamic acid
Gly = glycine
His = histidine
Ile = isoleucine
Leu = leucine
Lys = lysine
Met = methionine
Phe = phenylalanine
Pro = proline
Ser = serine
Thr = threonine
Trp = tryptophan
Tyr = tyrosine
Val = valine

1. Which of the following probes would hybridize to the following target sequence

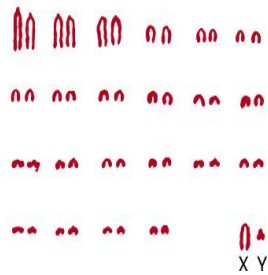
5'...ATTCGACATT...3'

- A. 5'...ATTCGACATT...3'
- B. 5'...TTACAGCTTA...3'
- C. 5'...AATGTCGAAT...3'
- D. 5'...TAAGCTGTAA...3'

2. A colleague shows you the following karyotypes for two new recently discovered organisms. Which of the following predictions could you make?



Organism A

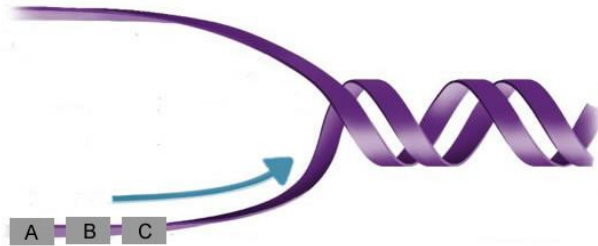


Organism B

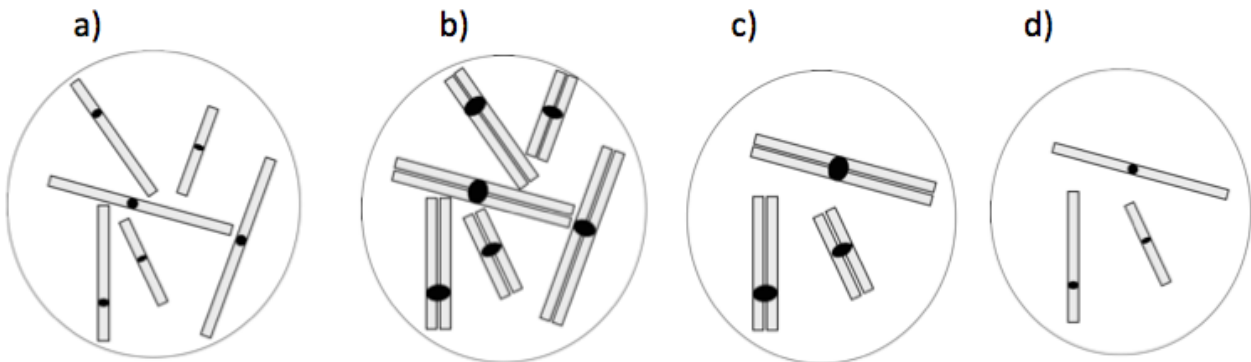
- A. Organism A's genome likely contains more nucleotides than that of organism B.
- B. Organism B's genome likely contains more nucleotides than that of organism A.
- C. Organism B is likely a male; Organism A is not male.
- D. You can't make any firm predictions on the nucleotide content nor gender.

3. On the right is a double strand DNA helix. Transcription is occurring and the mRNA transcript is represented by the arrow. Box B is the promoter segment. The arrow at the end of the mRNA indicates the direction of the transcription. The DNA sequence encoding the start codon for this gene is located in:

- A. Box A
- B. Box B
- C. Box C
- D. Note located anywhere



4. A certain cell is diploid and has a total of six chromosomes. If we pretend that its chromosomes remain condensed throughout the cell cycle, which of the diagrams below correctly represents the chromosomes of this cell before DNA replication. (A)



5. The object represented beside is composed of

- A. four single-stranded DNA molecules
- B. one double stranded DNA molecule
- C. two double-stranded DNA molecules
- D. two single-stranded DNA molecules



6. What are the total number of potential nucleotide sequences (combinations) that can be produced from a mRNA molecule containing 8 nucleotides.
- A. About 32 combinations
  - B. About 4,000 combinations
  - C. About 70,000 combinations
  - D. About 300,000 combinations
  - E. About 1 million combinations
7. The drug 5-bromouracil (5BU) is used to treat certain forms of cancer. This toxic compound is a base analog of thymine (T) and is incorporated into growing DNA chains. If 5BU is provided to a cancer cell entering the S (synthesis) phase, where will this drug be found in the chromosomes of newly formed daughter cells following mitosis?
- A. All of the chromosomes inherited by all daughter cells would contain 5BU.
  - B. Only half of the chromosomes inherited by any given daughter cell would contain 5BU.
  - C. Only half of the daughter cells would have 5BU in all of their chromosomes.
  - D. Only half of the daughter cells would have no 5BU in their chromosomes.
  - E. Both C and D will be seen.
8. Suppose the gene DKN1 is over 2000kb (kilobases) in length; however, the mRNA produced by this gene is only about 14 kb long. What is likely the cause of this discrepancy?
- A. The introns have been spliced out during mRNA processing and are not part of the mature mRNA.
  - B. The DNA represents a double-stranded structure, while the RNA is single stranded.
  - C. When the mRNA is produced, it is highly folded and therefore less long.
  - D. There are more amino acids coded for by the DNA than the mRNA.
  - E. The exons have been spliced out during mRNA processing and are not part of the mature mRNA.

9. A woman with no mutations in her X chromosomes has a daughter with a man whose X chromosome has a mutation in the Xist gene. The Xist gene produces a protein that when functional causes the inactivation of that particular chromosome (the chromosome does not get repressed as in the case of the Barr body example). This man also has the X-linked recessive disorder, haemophilia, which impairs the ability to stop bleeding through blood clotting. What are the effects on their daughter in terms of the inactivation of an X chromosome and the daughter's phenotype?
- A. There is no effect; the daughter's phenotype is normal (she does not have haemophilia).
  - B. The paternal X chromosome is inactivated in all of the daughter's cells; she has a normal phenotype.
  - C. Both the maternal & the paternal X chromosomes remain active in all the daughter's cells; she does not have haemophilia.
  - D. The maternal and paternal X chromosomes are randomly inactivated in the daughter's cells; she may have haemophilia if her maternal chromosome is inactivated in blood system cells.
  - E. The maternal X chromosome is inactivated in all of the daughter's cells; she therefore has haemophilia.

10. Assume that an mRNA molecule is synthesized using the following DNA template:

3'-CTTACATGGCATCC-5''

See the genetic code table on page 2. The second codon (counting the start codon as the first codon) directs the incorporation of which amino acid in the polypeptide?

- A. Asparagine
- B. Tyrosine
- C. Arginine
- D. Proline

11. Below are two DNA coding strand sequences. The 5' ATG encodes the "in-frame" start codon.

Hardeep's DNA sequence is: 5' ATG CGCTTA CCC TTA CTC CTA TAA 3'

Karen's DNA sequence is: 5' ATG CGCTAA CCC TTA CTC CTA TAA 3'

Karen's mutation causes the premature termination of:

- A. Replication
- B. Transcription
- C. Translation
- D. Both B and C

12. 13-deoxydanolidide is an antibiotic that binds to the E site of the ribosome. If 13-deoxydanolidide is added right before translation starts, which one of the statements is True?

- A. Translation would not happen.
- B. Translation would not be affected.
- C. The end product carries a 13-deoxydanolidide before the first amino acid Met.
- D. The end product has 2 amino-acids.

13. Which of the following macromolecules is primarily responsible for the differences between a neuron and a muscle cell?

- A. Carbohydrates
- B. DNA
- C. Lipids
- D. mRNA
- E. Proteins

14. A plant is homozygous for a mutation in gene *Bfr* (for this gene, the locus on both homologous chromosomes have the mutation). This plant produces a normal Bfr protein in normal amounts. More detailed analysis reveals that the *Bfr* mRNA produced by this plant is two nucleotides shorter than wild type plants (the locus for this gene on both homologous chromosomes of the wild-type plant do not possess the mutation). Where is it most likely that the two base pair deletion occurred?

- A. Downstream of the stop codon in the last exon of gene *Bfr*.
- B. In an intron of gene *Bfr* away from the splice sites
- C. In the open reading frame of gene *Bfr*.
- D. Within the promoter region of gene *Bfr*.

## PART B – SHORT ANSWERS

- ANSWER QUESTIONS 15 TO 23 ON THE SCANTRON.
- ANSWER QUESTIONS 24 AND 25 IN THE SPACE PROVIDED.
- Write/print clearly and neatly in the space provided.

### True or False (1 mark each)

An analysis of cheek cell DNA from members of a family that are afflicted by a “given” genetic condition reveals that there are 52 chromosomes in total. Close inspection of their karyotype indicate that there are 3 sets of chromosome #2, 3 sets of chromosome #5, 3 sets of chromosome #17, and 3 extra sets of the X chromosome. Determine if the following statements are true (**T**) or false (**F**).

15. The individuals afflicted by this genetic condition are considered triploids.
- True
  - False
16. The banding patterns on the Q arm of chromosomes #2 and #17 should be the same.
- True
  - False
17. Afflicted females in this family would exhibit 3 Barr bodies (3 inactivated chromosomes).
- True
  - False

Determine if the following statements about gene expression are True or False

18. Both strands of a DNA molecule could serve as coding strands for the same gene.
- True
  - False
19. Posttranslational modification can result in the production of different functional proteins
- True
  - False
20. Different open reading frames can create overlapping genes that when expressed into proteins can produce variants of the protein and even different proteins.
- True
  - False

Determine if the following statements about genes, genomes and chromosomes are True or False.

21. Sister chromatids do not share the same set of DNA markers (e.g. VNTRs).
- True
  - False

22. Multiple copies of the same gene can be found on different chromosomes.

- a. True
- b. False

23. Transcription regulatory elements of a given gene are only found upstream and close to the gene it regulates.

- a. True
- b. False

24. On February 15, 2012, Postmedia news published a report on a first study in nearly two decades showing that infertility is on the rise in Canada (measured by the proportion of Canadian couples who are having difficulty conceiving). The researchers didn't set out to discover why the numbers are increasing.

Based on the "excerpt" of the report described above, formulate two "genetic-related" research questions that could potentially form the basis of a research investigation to explain the rise of infertility. Write one research question/box (5 marks)

Students must write two questions (and only two questions). Questions must show the following indicators:

- (1) The questions must indicate a **dependant and independent** variable and (2) a **link** between these two variables.

For example: What are the effects of environmental pollutants (independent variable) on the transcription rate of gene X (dependant variable).

### **2.5 pts/question**

2.5 pts: questions addresses 1) and 2) and is directly linked to the excerpt above

1.5-2 pts: evidence for 1), more or less clear and coherent.

1 pt: evidence for 1), not very coherent;

0-0.5pts: vague, unclear, incoherent; does not address the excerpt.

25. Briefly explain what the following terms means. Where possible, include two solid pieces of information with a relevant example to which the term applies. **For this part, provide answers for 2 of the following 4 terms.** (Note: if you explain more than 3 terms, the first 3 will be evaluated). Each is worth **3 marks**.

The following is **a guide** that was provided to the TAs to assess answers. We have held many discussions with TAs to help them assess answers that were not necessarily the answers I communicate here.

**2.5-3 pts** – two solid facts and an example – answer is relevant, clear and concise

**2-2.5 pts** – two solid facts and an example – answer more or less relevant, clear and/or concise – or minor/major error

**1.5 - 2 pts** – two solid facts, no example

**1-1.5 pts** – two solid facts, no example – more or less relevant, clear and/or concise.

Minor/major errors

**0-1 pt:** - facts, not relevant or failure to understand relevance; incorrect

**I've given more information than is necessary for the students to obtain top marks – this is simply to provide you with much information to assess the different answers.**

**Mark in 0.5 pts.....**

**Beware: some answers may be correct which don't necessarily follow my answers. Please be flexible when this occurs and consider what the student has written. If definitions clearly provides a clear explanation within a given context appropriate for that term, please give credit where credit is due. Also, I have not necessarily added examples for every term as many examples may be given. Please consider the example and consider what the student has written.**

**In case of any doubt, please consult with me!**

Variable number of tandem repeats:

- (1) Is a location (locus) in a genome where a nucleotide sequence is organized as a tandem repeat.
- (2) These can be found on many chromosomes and often show variation in length between individuals.
- (3) Each variant acts as an inherited allele, allowing them to be used for personal or parental identification. (They may also describe how it is used in genetics).

Example: an drawing or representation of a tandem repeat ( ATCG ATCG ATCG).

Homologous chromosomes:

- (1) Is a set of one maternal chromosome and one paternal chromosome;
- (2) Each of the chromosome contain the same genes at specific loci but may carry different alleles;
- (3) In karotypes, these chromosomes would have the same length, banding pattern, and centromere position.

Heterochromatin:

- (1) **Heterochromatin** is a tightly packed form of DNA (histones);
- (2) May find repetitive DNA in these regions;
- (3) areas of repressed gene expression activity.

Transcription:

1. **Transcription** is the first step of gene expression, in which a particular segment of DNA is copied into RNA by the enzyme RNA polymerase;
- (2) activity controlled by transcription factors binding on the transcription regulatory elements;
- (3) the 3' → 5' strand is used as template for RNAP.