PLNT3140 INTRODUCTORY CYTOGENETICS
FINAL EXAMINATION
December 19, 2020

Answer any combination of questions totaling to exactly 100 points. If you answer questions totaling more than 100 points, answers will be discarded at random until the total points equal 100. There are 13 questions to choose from, totaling 120 points. This exam is worth 35% of the final grade.

Ways to write a readable and concise answer:
i. Just answer the question. Save time by specifically addressing what is asked. Don't give irrelevant background if it doesn't contribute to the question that was asked.
ii. Avoid stream of consciousness. Plan your answer by organizing your key points, and then write a concise, coherent answer. Make your point once, clearly, rather than repeating the same thing several times with no new information.
iii. Point form, diagrams, tables, bar graphs, figures are welcome. Often they get the point across more clearly than a long paragraph.
iv. Your writing must be legible. If I can't read it, I can't give you any credit.

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1. (10 points) Suppose you wish to map the gene for cystic fibrosis. If the Human genome has been mapped to a resolution of 1cM (ie. at least one marker every cM), which formula best describes the number of markers you must screen to be sure of finding at least one marker linked within 5cM of the gene for cystic fibrosis? (Assume you already have saturated the genome with markers, and have access to all mapped markers.)

a)

$$N = \frac{\ln(1 - 0.99)}{\ln\left(1 - \frac{5\,cM}{2708\,cM}\right)} = 2492\,markers$$

b)

$$N = \frac{\ln(1 - 0.99)}{\ln\left(1 - \frac{2 \cdot 5\,cM}{2708\,cM}\right)} = 1245\,markers$$

c)

$$N = \frac{2708\,cM}{2 \cdot 5\,cM} = 271\,markers$$

d)

$$N = \frac{2708\,cM}{5\,cM} = 542\,markers$$

2. (10 points)

a) (5 points) It is commonly observed that the size of a genome can change drastically due to gain or loss of middle repetitive sequences such as transposable elements. For example an increase in transposition might double the size of the middle repetitive fraction of the genome, due to proliferation of a family of transposons. In other words, there is twice as much middle repetitive DNA, after the transposition events.

To keep the math simple, consider an imaginary species with a diploid DNA content of 10 pg per nucleus. The following describes the components of that genome:

Population A - Original population

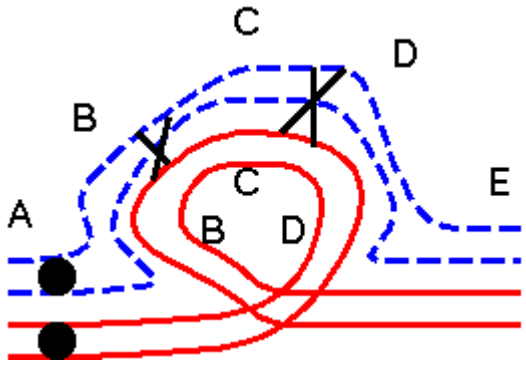
	percent of genome	pg DNA per nucleus
highly repetitive	15%	1.5
middle repetitive	75%	7.5
single copy	10%	1
total	100%	10.0

Suppose that in one population of this species (Population B), a drastic amplification of transposons took place, such that the middle repetitive fraction doubled in size. Create a new table, similar to the one above, for the genome of Population B.

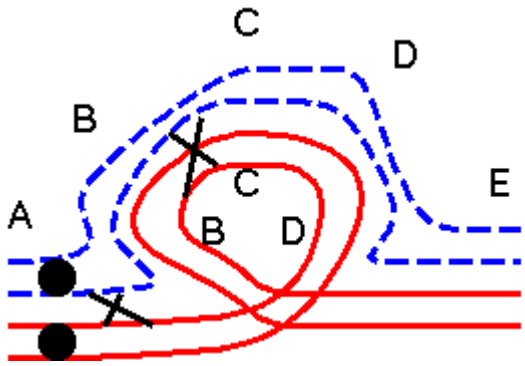
b) (5 points) With Population A as a starting point, what would happen if a third population of this species (Population C) had undergone a tetraploidization event. Create a table to represent Population C.

3. (10 points) For each of the following diagrams, indicate the outcome of double crossovers in meiosis by listing the number of normal, inverted, dicentric, or acentric chromosomes.

a



b

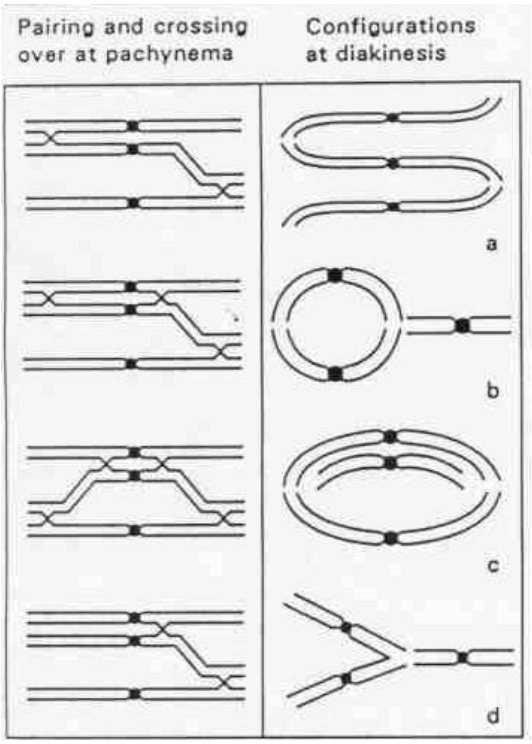


4. (5 points) Define the term “homeologous chromosomes”.

5. (10 points)

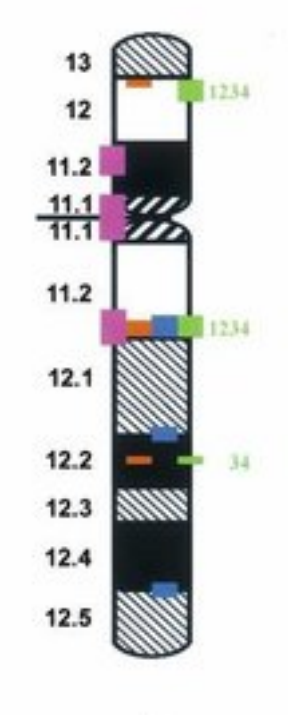
a) What is being illustrated in the figure at right?

b) Why do the chromosomes in diakinesis form chains?

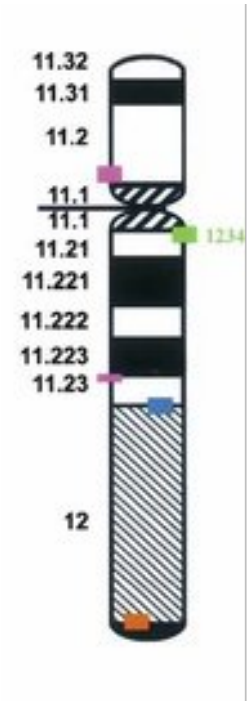


6. (10 points) Using correct terminology, describe and compare the following two ideograms to each other, making note of arm ratios, NORs, and other features. Which features are conserved? Which features have changed?
Note: diagonally hatched sections represent repetitive sequences with estimated length.

Gorilla Y chromosome



Human Y chromosome



7. (10 points) PCR-based markers were mapped in a segregating population. Data for 20 individual progeny are shown, with scores for five loci, A - E. Plus (+) indicates presence of a band in PCR, and minus (-) indicates absence of the band.

	20 segregating progeny																			
A	+	-	+	+	+	+	+	-	+	+	+	+	-	+	+	+	-	+	+	-
B	+	+	+	-	+	+	-	+	+	+	-	+	+	-	+	+	-	+	+	+
C	+	+	-	+	+	-	+	+	+	+	+	-	+	+	-	+	+	-	+	+
D	+	+	-	+	-	+	+	+	-	+	+	+	-	+	+	-	+	+	+	+
E	+	+	-	+	+	-	+	+	+	+	+	-	+	+	-	+	+	-	+	+

a) At any given locus, you will observe that more individuals give a + score than a - score. Explain the reason for that observation.

b) Of the 5 loci, which two are the most tightly linked? Explain your reasoning.

8. (10 points) You are beginning a new project studying ribosomal RNA genes, which are typically present in > 200 copies per haploid genome. The project will involve a lot of Southern blots, which normally require a 20 hour hybridization time to detect a single copy gene. You realize that it should be possible to do your Southern blots in a shorter time, because of the fact that rRNA genes are present in high copy numbers. If you want to get bands with the same intensity as you would get for a single copy gene, how long should you hybridize? Hint: Consider the definition of C_0t. Show your work.

9. (10 points) We have spent a lot of time describing how chromosomal abnormalities such as translocations, inversions, deletions and duplications can help drive speciation, through their effects on pairing at meiosis.

Describe a mechanism by which amplification or deletion of middle-repetitive sequence families might also create reproductive barriers between populations within a species?

10. (10 points)

a) Given the following sequence in an mRNA,

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5'CUA AAA CUG CCA AUC NNN NNN ...3'
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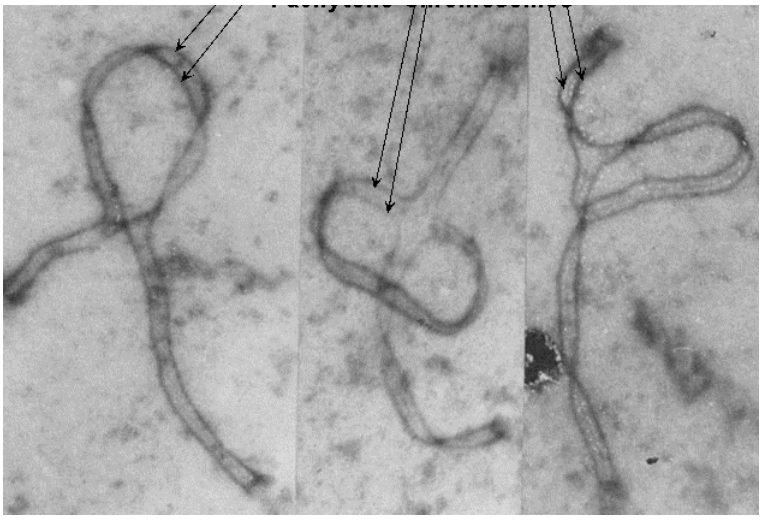
What is the corresponding amino acid sequence?

b) If an A in the 2nd codon was deleted, what would the new amino acid sequence be?

Hints:
1. Show as many amino acids as possible
2. Use a fixed font such as Courier to show RNA or amino acid sequences

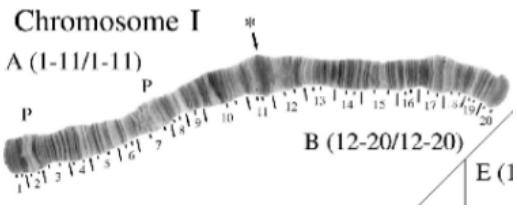
11. (10 points) The chromosomes below are from cells at pachytene.
a) Why do the chromosomes appear as two threadlike-structures, rather than four?
b) What is the most likely explanation for the loops seen in each?



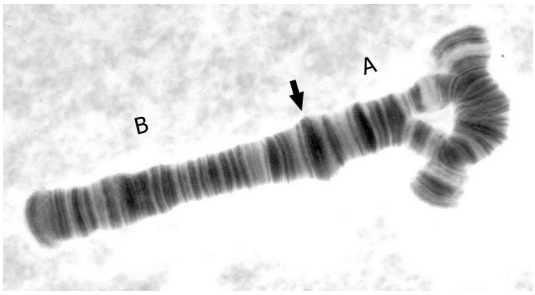
12. (5 points) Multi-slect.

Chromomsome I from the salivary glands of two individuals of a midge species are shown. Several hypotheses are stated below. Check all that are consistent with the data shown.

A



B



a) A wild type chromosome.

b) B is an inversion heterozygote

c) A is an inversion homozygote

d) B is an inversion homozygote

e) B is a wild type chromosome

13. (10 points) The table below shows the number of transcribed loci reported on each human chromosome. In light of this data, explain why the only human trisomies that survive to birth are for chromosomes X, Y, 13, 18 and 21.

chromosome	1	2	3	4	5	6	7	8	9	10	11	12
# of loci	2468	1692	1372	926	1152	1311	1201	886	1015	992	1534	1279
chromosme	13	14	15	16	17	18	19	20	21	22	X	Y
# of loci	427	730	746	1056	1433	372	1590	748	290	613	1081	119