PLNT3140 INTRODUCTORY CYTOGENETICS
FINAL EXAMINATION
December 19, 2019

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 12 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 want make a BAC library for a genome. Given a genome with a total haploid size of 4 x 10^9 bp, how many BACs do we need to have a 99% likelilood of having a complete library ie. having at least one clone for every gene? Show your work, using the Clark and Carbon formula. Assume an average BAC insert size of 100 kb.

$$N = \frac{\ln(1 - P)}{\ln(1 - f)}$$

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

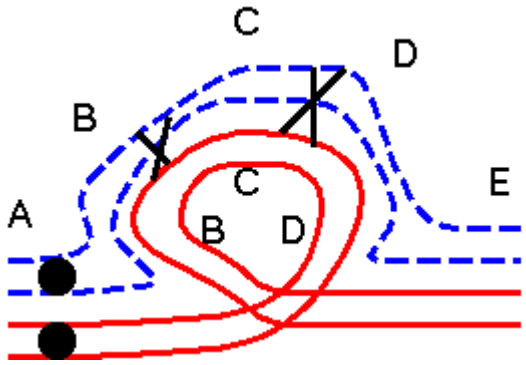
	percent of genome	pg DNA per nucleus
highly repetitive	15%	1.5
middle repetitive	75%	7.5
single copy	10%	1
total	100%	10.0

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.

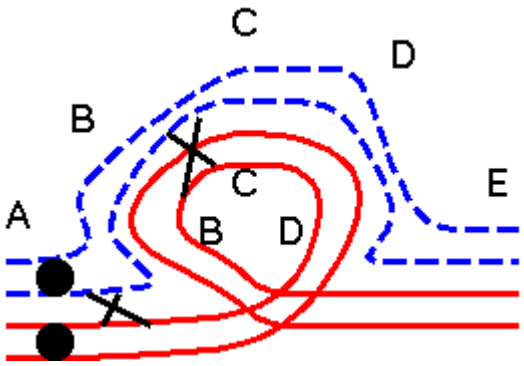
b) (5 points) With Population A as a starting point, what would happen if a third population of this species (Population C) had undergone a tetraploidization event. Create a table to represent Population C.

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



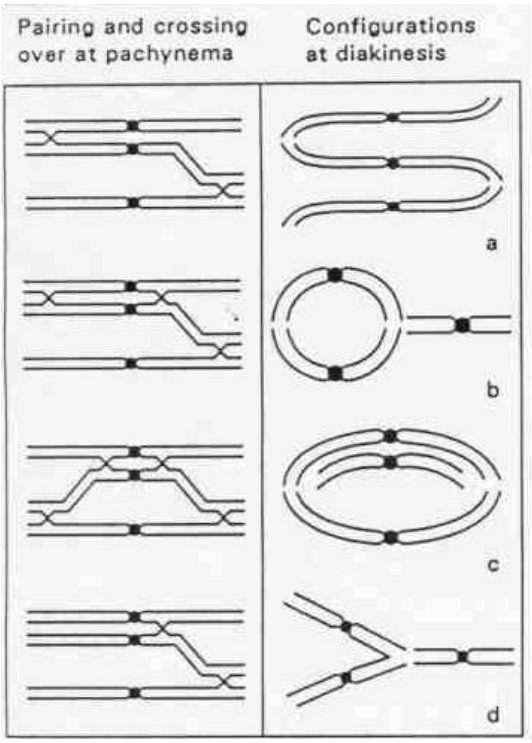
b



4. (10 points)

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

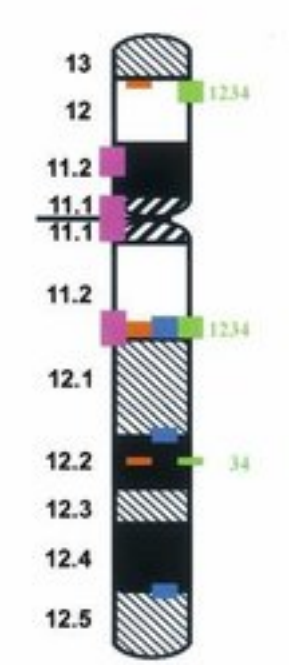
b) Why do the chromosomes in diakinesis form chains?



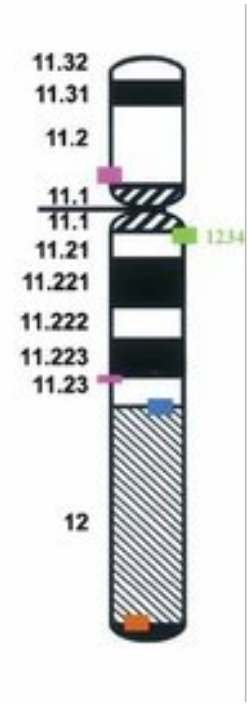
5. (5 points) Define the term “homeologous chromosomes”.

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 have changed? Which features remain roughly the same?
Note: diagonally hatched sections represent repetitive sequences with estimated length.

Gorilla Y chromosome

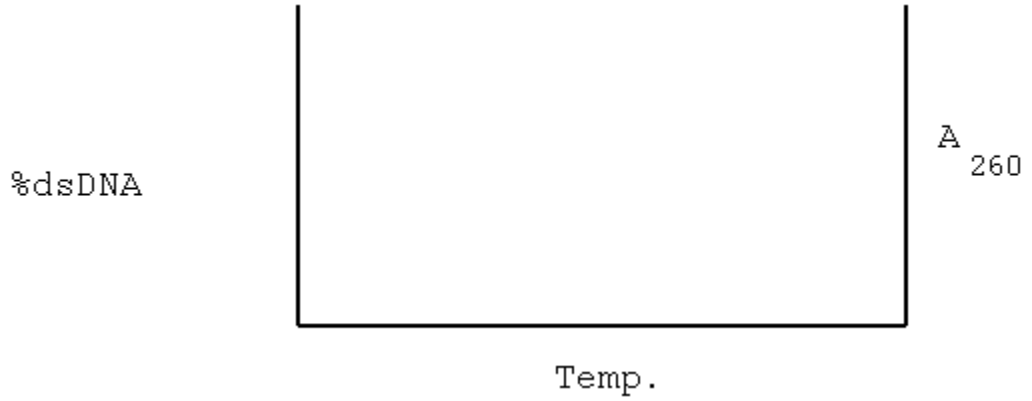


Human Y chromosome



Kirsch, Stefan & Muncn, Claudia & Jiang, Zhaoshi & Cheng, Ze & Chen, Lin & Batz, Christiane & E Eichler, Evan & Schempp, Werner. (2008). Evolutionary dynamics of segmental duplications from human Y-chromosomal euchromatin/heterochromatin transition regions. Genome research. 18. 1030-42. 10.1101/gr.076711.108

7. (10 points) Draw a melting curve for DNA using scales as shown below. On a single graph, compare curves for AT rich DNA, heterogeneous DNA and GC rich DNA. Indicate Tm for each. Make sure to include numbers for each of the two Y-axis scales. The X-axis scale can be assumed to be arbitrary.



8. (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.

9. (10 points)

a) Draw a diagram illustrating how chromosomes pair during meiosis in cells heterozygous for a reciprocal translocation. Feel free to use colors, shading or hatching to represent homologous chromosomes.

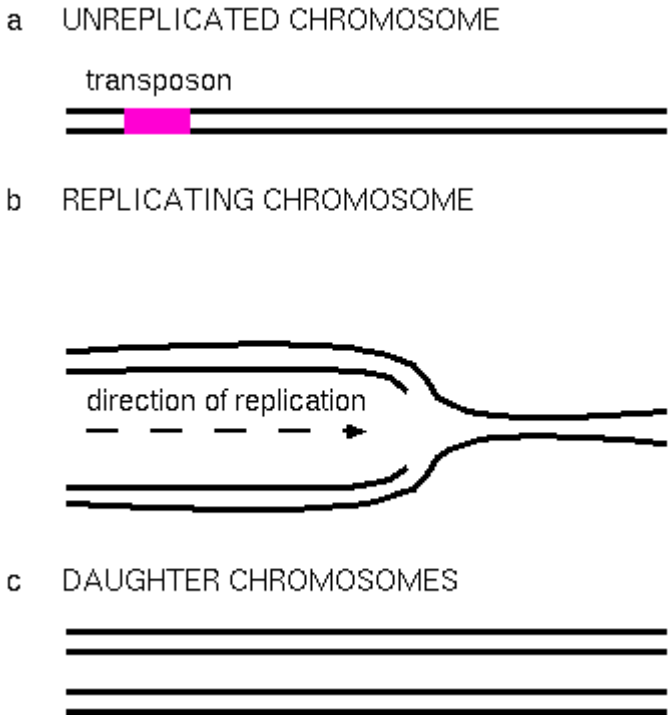
b) Draw a diagram illustrating the orientation of reciprocal translocation products from a), at metaphase, assuming an Alternate configuration.

10. (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.

11. (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 familes might also create reproductive barriers between populations within a species?

12. (5 points) Redraw the picture below, showing how transposition of a transposon during DNA replication can result in a net increase in the number of transposons.



13. (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?