PLNT3140 INTRODUCTORY CYTOGENETICS
FINAL EXAMINATION
January 21, 2022

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 (total 2708 cM) 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) 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?

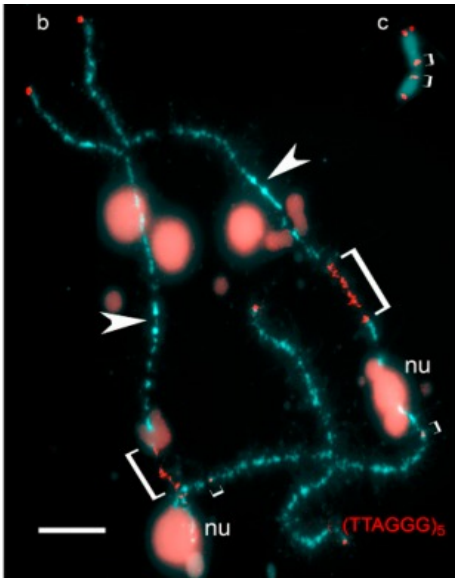
Hints:
1. Show as many amino acids as possible
2. Use a `fixed font such as Courier` to show RNA or amino acid sequences

3. (10 points) Suppose you have agreed to review a manuscript for a journal. The authors report a new mutant for the Histone H2a gene. The mutation results in an inactive H2a protein which prevents formation of a histone core particle. When homozygous H2a mutants are crossed with the wild type, the progeny segregate 3:1 for the wild type to mutant phenotype.

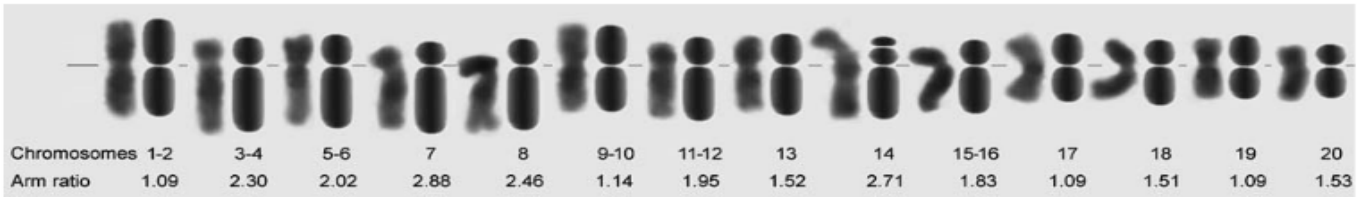
Would you accept or reject the paper? Briefly state your reasons.

4. (5 points) In some bird species, oocytes are arrested in diplotene of meiosis, during which they partially uncoil to allow transcription to occur. These so-called lampbrush chromosomes allow for high-resolution imaging of chromosomes. In the figure below, the bright region indicated by the brackets indicates hybridization of a telomeric FISH probe to a site within the chromosome arm. Arrowheads show the positions of centromeres. The inset c) shows the condensed metaphase chromosomes, also hybridized with telomeric probe.

Propose a genetic mechanism by which telomeric sequences occur within a chromosome.



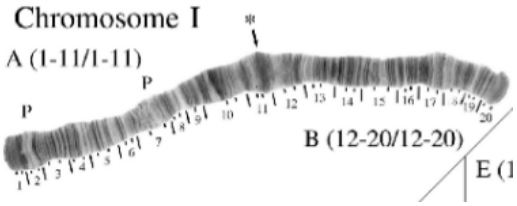
5. (5 points) On which chromosome would you expect to find ribosomal RNA genes?



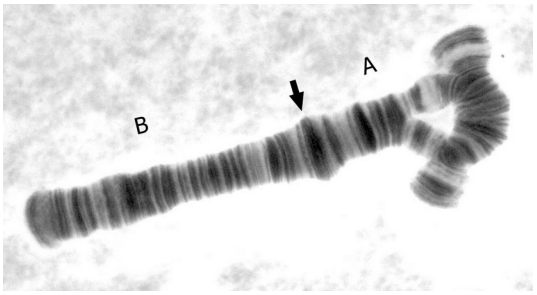
6. (5 points) Multi-slect.

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

7. (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

8. (10 points, multiple choice) The 20 chromosomes of the oilseed plant Camelina sativa have been arranged in a circle to facilitate easy comparison of homologous blocks of sequences between chromosomes. In this example, homologous sequences shared by both chromosomes 3 and 7 are shown. The density of repetitive sequences is shown in the outer (red) circle, while the density of genes is shown in the inner circle (blue).

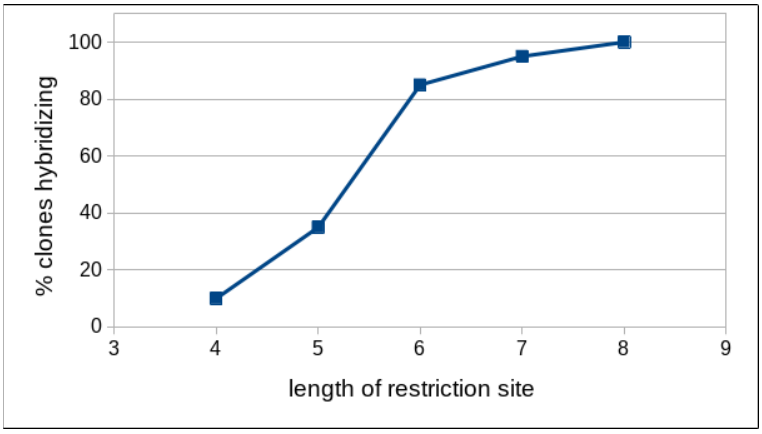
Based on data in the map, select one of the following for each statement below:

- SUPPORTED - consistent with the map, with some direct evidence
- CONSISTENT- consistent with the map, but no direct evidence from the map
- INCONSISTENT - contradicted by map

a) Gene rich regions tend to have a high repeat density
b) Chromosome 7 is derived from part of chromosome 3
c) The intergenic regions in chromosome 3 have lost copies of repetitive sequences that are present in chromosome 7
d) The intergenic regions in chromosome 7 have gained copies of repetitive sequences that are not present in chromosome 3
e) Camelina sativa is an allotetraploid

9. (10 points) Five libraries of genomic DNA fragments were constructed using DNA cut with five different restriction enzymes.

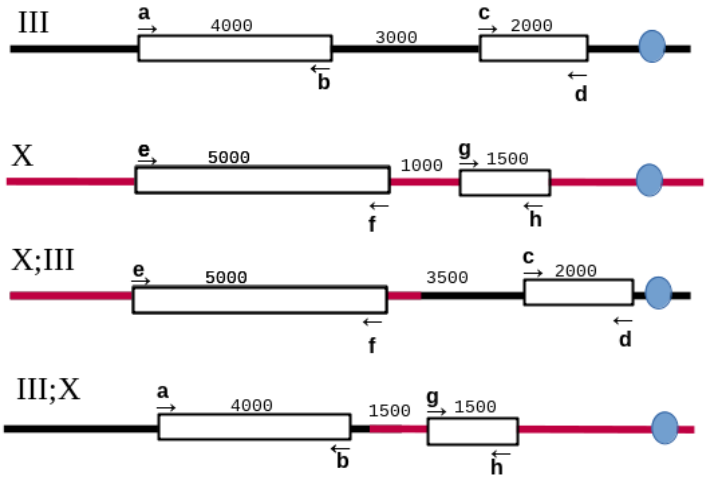
Enzyme	Recognition Sequence	length of rest. seq.	avg. insert size
TaqI	TCGA	4	256
MboII	GAAGA	5	1024
BamHI	GGATCC	6	4096
AbeI	CCTCAGC	7	16384
NotI	GCGGCCGC	8	65536



Each of the libraries was probed with a sequence from a 300 bp middle repetitive sequence, and the percentage of clones hybridizing was estimated for each library. For example, a library of TaqI fragments would have an average insert size of 256 bp. When the library of TaqI fragments was probed, about 10% of the clones showed hybridization. When the library of MboII fragments was probed, about 35% of the clones showed hybridization, and so on.

Briefly explain why these results were seen.

10. (10 points) The map shows two genes on chromosomes III and X, and PCR primers available for these genes. The goal is to detect individuals carrying the translocations. Choose pairs of primers that would give distinct bands for each of the two normal and two translocated chromosomes.



a) III
b) X
c) X;III
d) III;X
e) null control*

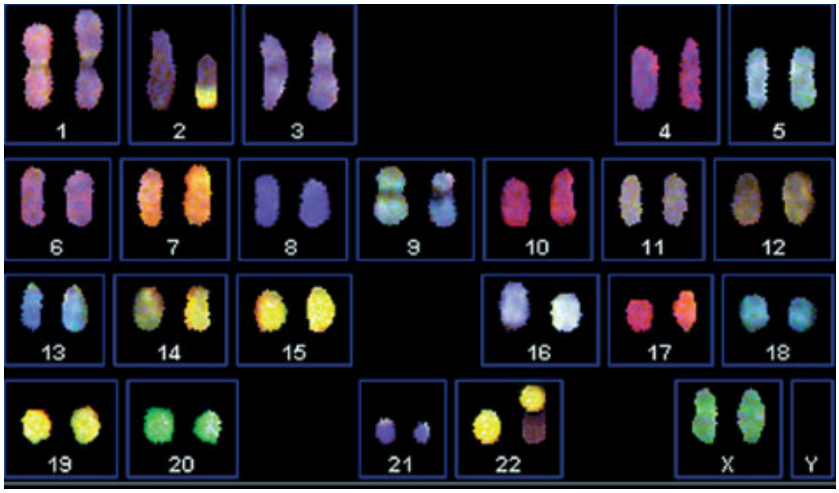
* - choose any primer combination that would not give any bands

11. (10 points, multiselect)

In maize, the Ds element inserted into the C locus, inactivating that gene, and giving yellow kernels. When the yellow maize (cc) was crossed with a line bearing the Ac locus, the kernels displayed blue sectors. Check all that apply:

a) The Ac element carried a wild type C allele.
b) The Ds element carried a gene for transposase.
c) The Ac element carried a gene for transposase.
d) In some kernels, excision of the Ds element early in kernel development resulted in large blue sectors.
e) In some kernels, excision of the Ds element early in kernel development resulted in small blue sectors.

12. (10 points) The spectral karyotype below shows a set of human chromosomes. What sex is the person from which the chromosomes were imaged? What is the other important finding that is apparent from this data?



13. (10 points) Given the following cases,

a) diploid which has undergone a autopolyploidization to autotetraploid
b) two diploids which have hybridized to form an allotetraploid

match the following statements

i) C_0t curve will not change
ii) C_0t curve will shift to the right
iii) Genome size (bp) will stay the same
iv) Genome size (bp) will double
v) homeologous pairing will occur in meiosis