39.314 INTRODUCTORY CYTOGENETICS
FINAL EXAMINATION

December 4 , 2003
Time: 1:00 p.m. to 3 p.m.
Location: 134 Agriculture

This examination consists of questions totalling 100 points, and is worth 35% of the final grade.

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1) (20 points) For <u>any two</u> of the following, explain whether it would be better (ie. faster, cheaper, more accurate) to use genomic approaches (eg. reassociation kinetics, mapping with molecular markers, contig assembly, genomic sequencing) or cytogenetic methods (eg. staining, banding, FISH, microdensitometry)

a) measurement of genome size
b) detection of chromosomal abnormalities (eg. translocations, inversions)
c) determination of chromosome number
d) determination of ploidy level

In what ways are the methods you chose better than other possible approaches?

2) (10 points) A cross was made between a plant that is trisomic for a dominant marker with a plant that is diploid for the recessive allele, as illustrated below:

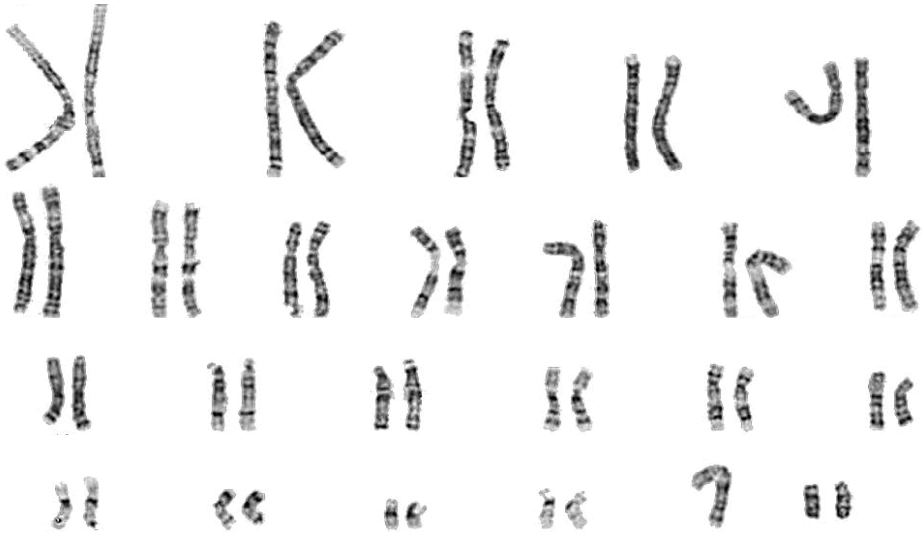
```
                          AAA      x      aa
                                   |
                                   v
F1 progeny           AAa                      Aa
                      | self                    | self
                      v                         v

Gametes       AA  2A  2Aa  a                A    a
```

Show the genotypes, and their respective ratios, for all possible progeny. What is the phenotypic ratio of A_ to aa F2 progeny?

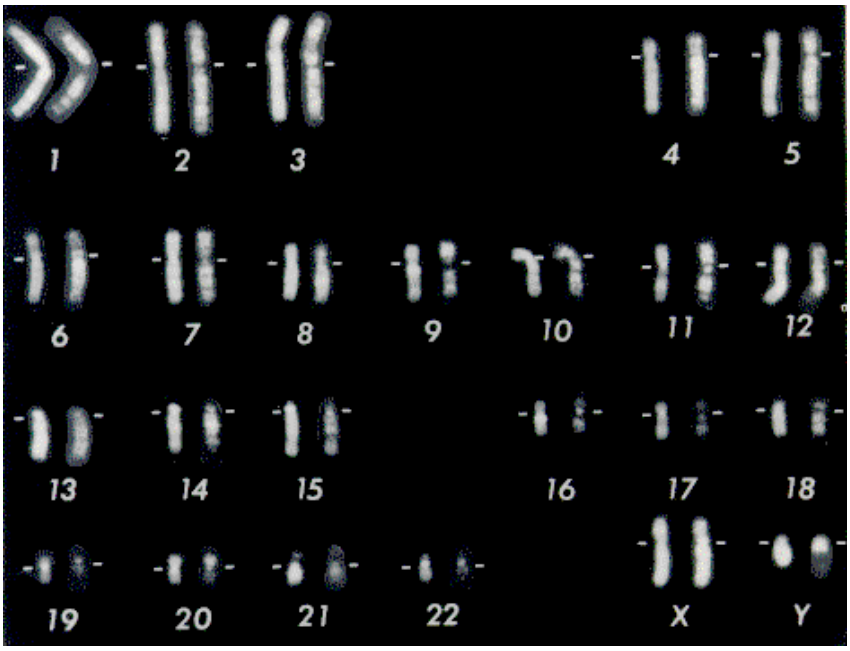
3) (5 points) In the context of <u>genetic mapping</u>, explain what this equation means:

$$N = \frac{(1-P)}{(1-\frac{2d}{G})}$$

4) (5 points) The human karyotype below is from an aneuploid individual. What is the aneuploidy?



5) (15 points) We know that eukaryotic genomes are predominantly middle repetitive sequences, interspersed among single copy DNA, throughout the genome. For example, in-situ hybridization of human chromosomes with an L1 family probe gives the following result:



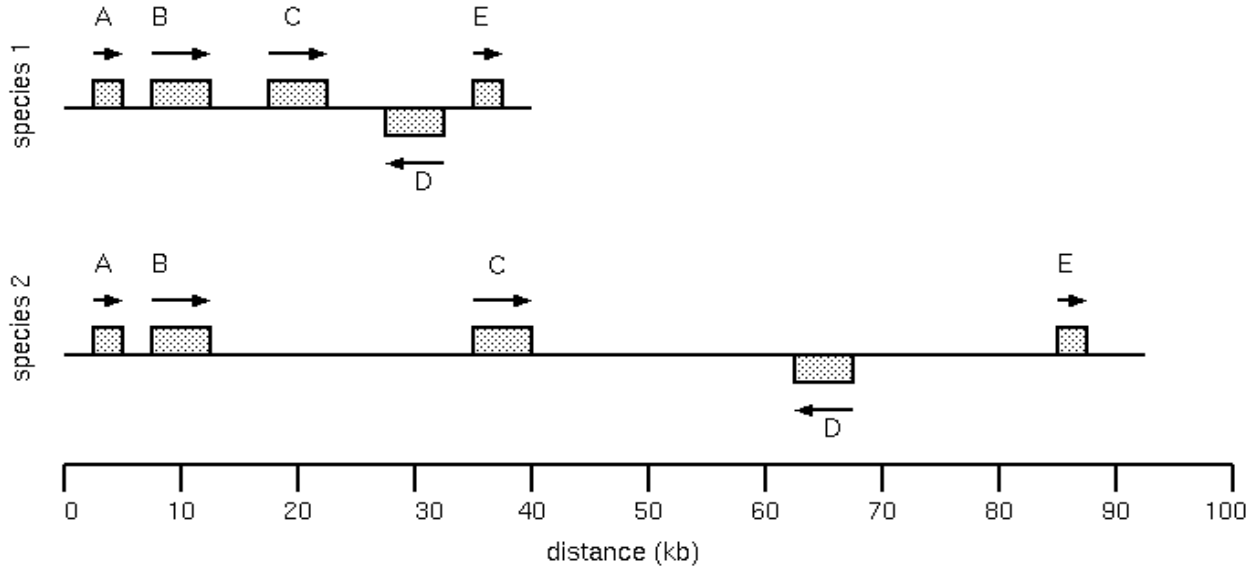
right - hybridization with L1 probe; left - DAPI staining

In light of this data, why don't all chromosomes pair with all other chromosomes? Give at least 3 reasons.

6) (20 points) RAPD markers for two loci, A and B are tested for genetic linkage by crossing two parents and selfing the F1s. One of the original parents was homozygous for the presence of a band (+) for both markers , while the other parent was homozygous for the absence of a band (-), for both markers. Rewrite the table, indicating, for the four phenotypic classes, the expected ratios of progeny in a segregating F2 population. Assume that"tightly-linked", means that no recombination is detected between A and B loci, and that A and B are linked in coupling.

Marker A	Marker B	unlinked	tightly-linked, in coupling + + − − −−−− x −−−− + + − −
+	+		
+	-		
-	+		
-	-		

7. (15 points) The figure below shows a chromosomal region from two different species. The location and directions of transcription for 5 genes are shown. Describe possible evolutionary processes that may have occurred since the divergence of these two species. What are the implications for the evolution of genome size?



8. (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