

## 39.314 INTRODUCTORY CYTOGENETICS FINAL EXAMINATION

December 16 , 2005

Time: 9:00 a.m. to 11:00 a.m.

Location: 229 EIT, Seats 142-157

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

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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 readable. If I can't read it, I can't give you any credit.
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1. (20 points) Fill in the blanks.

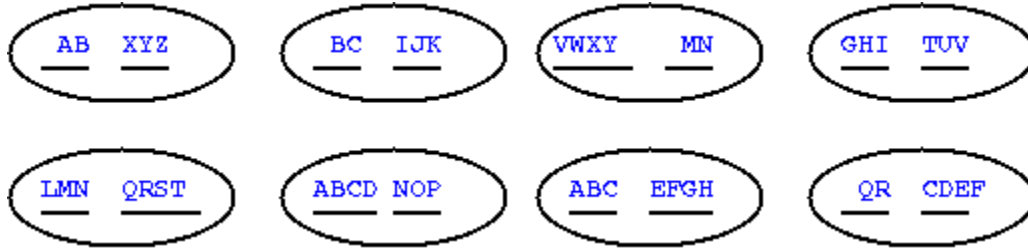
\_\_\_\_\_ i \_\_\_\_\_ can generate both deletions and duplications in a single event. Double-stranded breaks in a chromosome can also generate deletions, because one of the resultant chromosomal fragments will be lacking a \_\_\_\_\_ ii \_\_\_\_\_ and therefore cannot segregate reliably. Introduction of alien chromosomes to crop species through crosses with \_\_\_\_\_ iii \_\_\_\_\_ can also cause deletions in some or all of a chromosome. Once duplications and deletions exist, their effects on chromosome pairing in heterozygotes can result in new duplications and deletions occurring in subsequent generations as a result of \_\_\_\_\_ iv \_\_\_\_\_ cycles. Ring chromosomes also can cause deletions and duplications. There is no fundamental reason why ring chromosomes shouldn't be able to replicate and segregate normally. However, if \_\_\_\_\_ v \_\_\_\_\_ occurs, there will almost certainly be a duplication and deletion.

Another mechanism for chromosomal evolution is transposition. The first transposon discovered was the Ds element, which is flanked by \_\_\_\_\_ vi \_\_\_\_\_. Normally, a Ds element will stay at a fixed chromosomal location. However, if a Ds-bearing maize line is crossed with a maize line containing the Ac element, \_\_\_\_\_ vii \_\_\_\_\_. This is possible because the Ac element carries a gene for \_\_\_\_\_ viii \_\_\_\_\_ which is lacking in Ds. In the presence of Ac, Ds mobilization caused \_\_\_\_\_ ix \_\_\_\_\_ of the colorless phenotype to colored. The earlier the excision of Ds occurs during kernel development, the \_\_\_\_\_ x \_\_\_\_\_ the colored sectors will be.

2. (10 points) In classical Mendelian genetics, the distances between loci are based on the frequency of recombination. The closer two loci are, the more likely it is that they co-segregate.

An alternative approach is Radiation Hybrid (RH) mapping. Briefly, cells from one species (eg. human) are subjected to ionizing radiation, which causes double-stranded breaks in chromosomal DNA, randomly distributed across the genome. Treated cells are fused with mouse cells. In many

cases, a mouse cell will incorporate one or more broken human chromosomal fragments into mouse chromosomes, while most human chromosomes are lost. Several hundred clonal cell lines are established, each with one or more human fragments. Some examples of clones are illustrated below (only human fragments are shown):



To use these cell lines for mapping, PCR-based markers of any kind can be used to score for presence or absence of loci. The closer together two loci are on the chromosome, the more frequently one will see bands for both loci. In the example above, A and B are found on three different fragments, while both A and D are both found on only one fragment. We would conclude that A is closer to B than it is to D.

- a) (5 points) Would the RH distances calculated be the same as distances calculated by Mendelian genetics? Why or why not?
- b) (5 points) Mendelian genetics requires the presence of two different alleles at each locus. Is this also true of RH markers? Explain.

3. (15 points)

The new world cotton species *Gossypium hirsutum* has a 2n chromosome number of 52. The old world species *G. thurberi* and *G. herbaceum* each have a 2n number of 26. Hybrids between these species show the following pairing arrangement at Metaphase I:

Hybrid	Pairing Arrangement
<i>G. hirsutum</i> X <i>G. thurberi</i>	13 small bivalents + 13 large univalents
<i>G. hirsutum</i> X <i>G. herbaceum</i>	13 large bivalents + 13 small univalents
<i>G. thurberi</i> X <i>G. herbaceum</i>	13 large univalents + 13 small univalents

- a) What is the probable x number of the *Gossypium* species?
- b) Explain the origin of *G. hirsutum* in evolutionary terms. Use diagrams to support your explanation. How would you test your interpretation?

4. (5 points) In the table below, the genetic lengths are given for all human chromosomes, with the exception of Y. Why are no results shown for the Y chromosome?

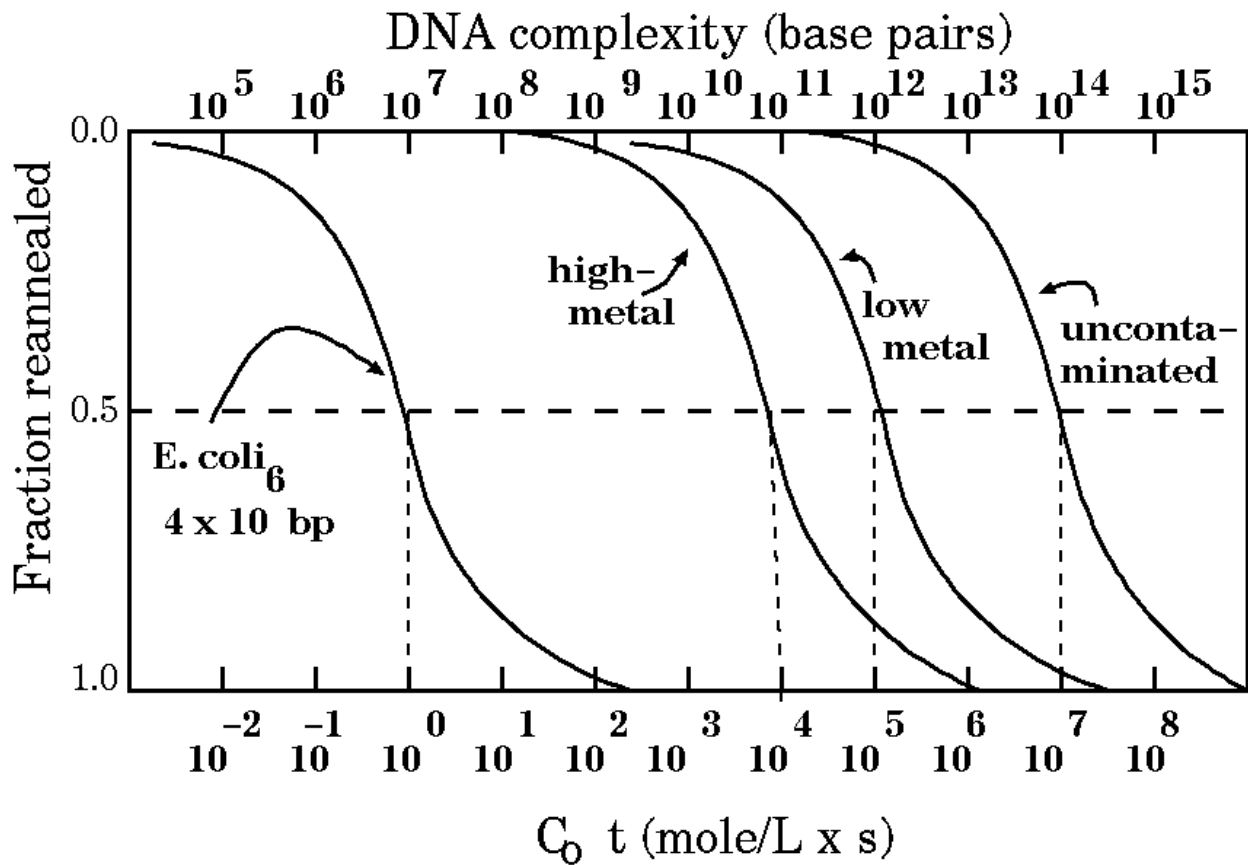
Physical and Genetic lengths of human chromosomes					
	Physical map (Mb)	Genetic map (cM)			Number of markers
		Male	Female	Sex Avg.	
1	282	195	345	270	468
2	252	190	325	257	407
3	225	161	276	218	369
4	205	147	259	203	302
5	199	151	260	206	334
6	191	138	242	190	293
7	169	128	230	179	246
8	158	108	210	159	247
9	150	117	198	158	193
10	146	134	218	176	256
11	153	109	196	152	260
12	153	136	207	171	239
13	100	101	156	129	175
14	87	94	142	118	161
15	87	103	155	129	125
16	106	108	150	129	151
17	89	109	162	135	181
18	89	99	143	121	158
19	69	93	127	110	120
20	59	75	122	98	141
21	30	47	76	62	67
22	31	49	83	66	66
X	156		179	179	177
TOTAL	3191	2591	4460	3615	5136

5. (10 points) What would be the problems associated with using fluorescent in-situ hybridization for estimating the distance between two loci on a chromosome? If you had to do it, which stage of meiosis would you choose, and why?

6. (15 points) Many species of bacteria can not be grown in pure culture, making them very difficult to study. This means that it is often difficult to estimate the number of bacterial species, and their relative abundance, in natural settings such as soil. If we make some simplifying assumptions, we can use  $C_0t$  analysis to get an idea of the number of species in a soil sample. These assumptions are

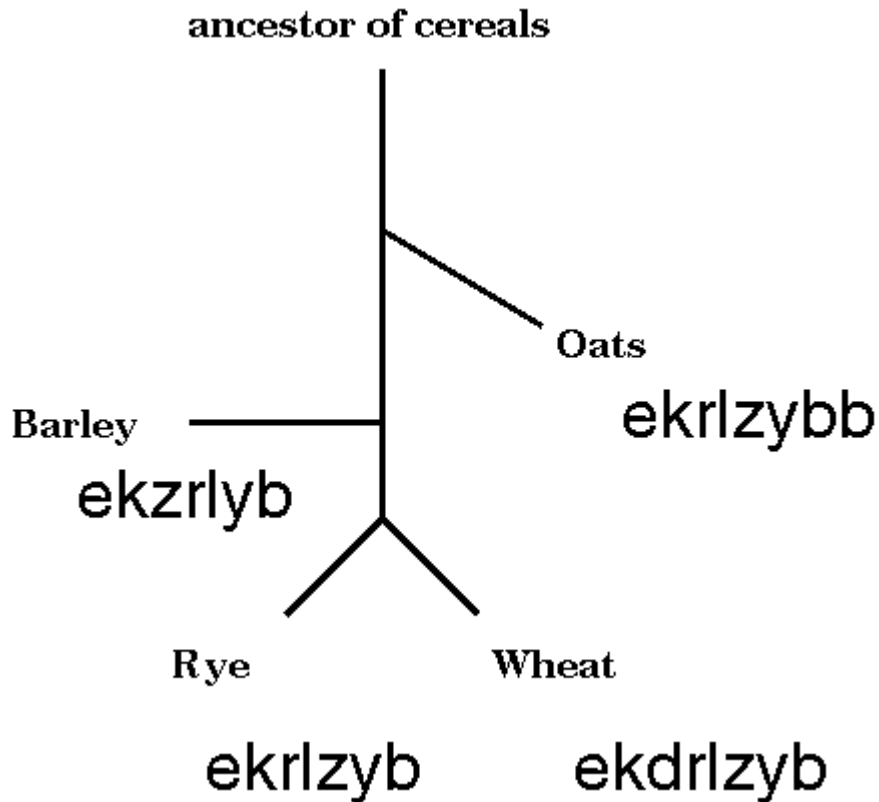
- all bacterial genomes are about the same size, using *E. coli* as the standard.
- all bacterial genomes are distinct from one another ie. they do not cross hybridize.

In an experiment designed to assess the ecological effects of heavy metal pollution on bacteria, bacteria were purified from soil samples at sites with high levels of metal contamination, low levels of contamination, or from uncontaminated soil. DNA was isolated from each bacterial sample, and  $C_0t$  analysis was done, using purified *E. coli* DNA as a reference standard. The results are shown below.



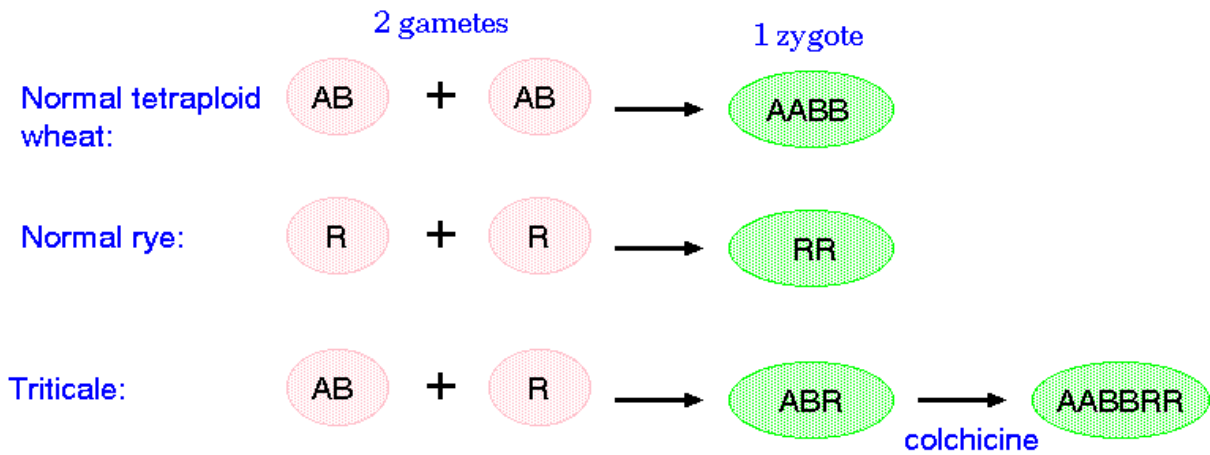
Briefly describe the differences between bacterial populations in uncontaminated, low metal and high metal soils.

7. (10 points) Just by looking at any pair of sequences or chromosomes, it is impossible to determine evolutionary history. For example, it is not possible to distinguish between an insertion in one sequence versus a deletion in another. However, when examples are available from several species whose evolutionary histories are known, it is possible to reconstruct the sequence of events that have led to the modern day chromosomes that we observe. The figure below shows the relationships between four cereal species. For each species, the order of a set of loci on a chromosome is indicated.



Reconstruct the order of mutational events, (eg. insertion, deletion, inversion, duplication etc.) and show the order of these loci in the common ancestor of the four cereals.

8. (15 points) The synthesis of Triticale from wheat and rye is illustrated below:



- What is the purpose of the colchicine step?
- What problems would arise if it were omitted?
- At what stage in development must colchicine be administered to the haploid plants?