

39.314 INTRODUCTORY CYTOGENETICS FINAL EXAMINATION

December 4 , 2001

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.

1. (10 points) ESTs are cDNA clones for which partial sequence is available, usually from a single sequencing reaction. This question distinguishes between what ESTs can tell you, and what they can't. Complete the sentence for ANY 5 of the following:
 - a) Map position: An EST, by itself doesn't tell you the map position of a gene, but it can be used to find the position by _____.
 - b) Amino acid sequence: An EST tells you some of the amino acid sequence of a protein, if you can determine _____.
 - c) Locations of introns: An EST can tell you the location of some of the introns if you also know _____.
 - d) Gene function: An EST can't tell you the function of gene from the sequence alone, but that sequence can be used to infer function if _____.
 - e) Gene copy number: If a gene exists as a multigene family, a large EST population can tell you a minimal estimate of copy number but _____.
 - f) Gene expression: ESTs can tell you whether or not a gene is expressed in a given tissue or developmental stage, but if you want to know _____ the ESTs must be used in gene array experiments.

- 2) (5 points) What is the difference between a kinetochore and a centromere?

- 3) (10 points) Define ANY 5 of the following:
 - a) Metacentric chromosome
 - b) Acrocentric chromosome
 - c) p and q arms (human nomenclature)
 - d) transposition
 - e) translocation
 - f) chromosomal satellite

- 4) (5 points) Use a diagram to show why the presence of a tandem duplication of part of a chromosome can lead to both a duplication and a deletion.

- 5) (10 points) Suppose that a plant heterozygous for a reciprocal translocation is crossed to one with the normal karyotype. What will be the ratio of 'normal : translocation heterozygote : translocation homozygote'?

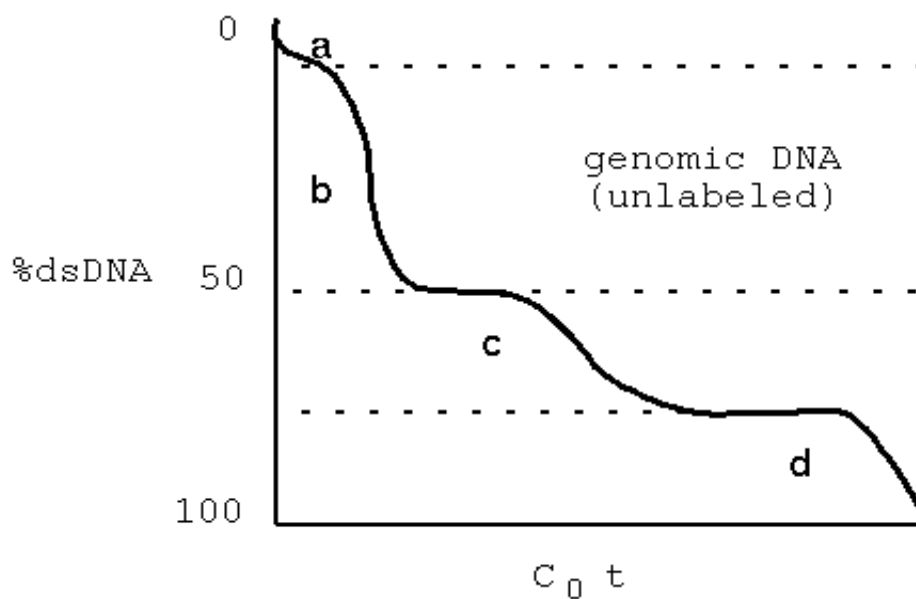
6) (15 points)

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

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

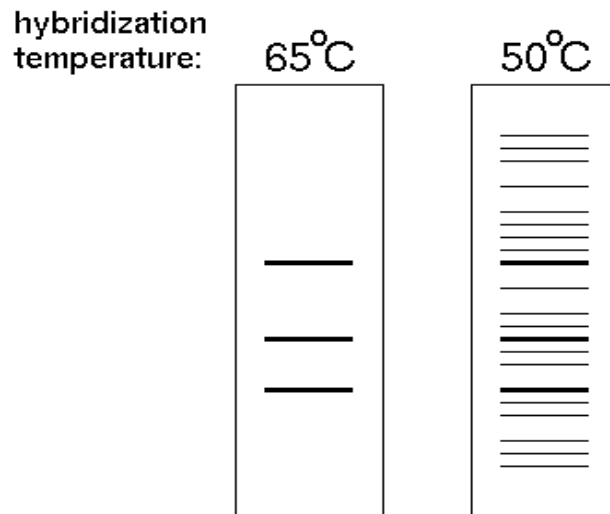
7) (15 points) The following C_0t curve was obtained using DNA from a eukaryotic genome:



- For each of three fractions **a**, **c** and **d**, identify that fraction, and give an example of a gene or a type of sequence that might be found in these fractions.
- It is observed that fraction **a** reanneals almost instantly. How might you go about purifying fraction **a** ?
- Fraction **a** was purified, and run on a gel under non-denaturing conditions. Ethidium bromide staining showed that a major band was seen that appeared to migrate at 150 bp. However, when the same DNA was run under denaturing conditions, the band migrated at 300 nucleotides in length. Explain this result.

8) (10 points)

A southern blot of mouse genomic DNA was hybridized with a mouse cDNA clone at 65°C, showing three bands. The blot was washed and re-hybridized with the same probe, but this time at 50°C with dozens of bands now visualized. It is well-known that at high temperatures, close-to perfect match is required for two sequences to hybridize. The lower the temperature, the more mismatches are tolerated.



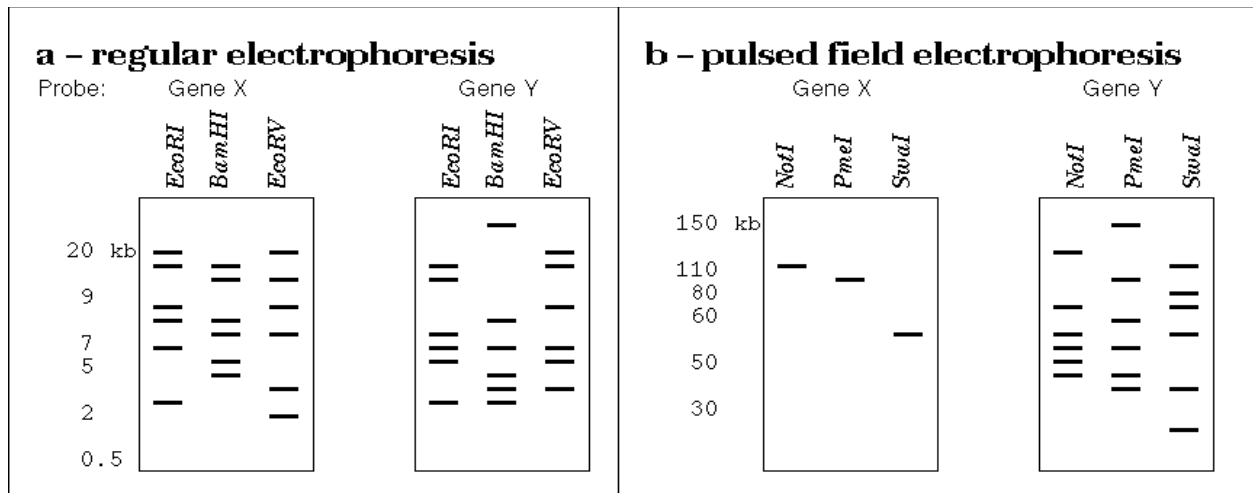
What do these results tell you about the evolution of this sequence in the rodent genome?

9) (10 points) The *Brassica napus* genome is amphidiploid, containing two homoeologous genomes (AACC) which are represented in *B. rapa* (AA) and *B. oleracea* (CC). Suppose you wanted to assemble complete contigs, consisting of partially overlapping BAC clones spanning the entire length of each chromosome. Would it be easier to do this in the amphidiploid *B. napus*, or in the diploid species *B. rapa* or *B. oleracea*? Explain your reasoning.

10. (10 points) To study the organization of two genes, X and Y, in the rice genome, restriction digests were performed using enzymes listed in the table below. None of these enzymes cuts within either gene. Southern blots of these digests, using either X or Y as probes, are shown.

a) Based on southern blots shown in **a**, estimate the number of copies of X and Y per haploid genome in rice.

b) To learn more about where the genes are located in rice, pulsed field electrophoresis (**b**) was done using DNA digested with enzymes that cut at 8 bp recognition sequences. Explain the difference between Gene X and Gene Y, with respect to their organization in the genome.



Enzyme	Recognition Sequence	Avg. distance between sites (bp)
<i>EcoRI</i>	gaattc	4096
<i>BamHI</i>	ggatcc	4096
<i>EcoRV</i>	gatatc	4096
<i>NotI</i>	gcggccgc	65536
<i>PmeI</i>	gtttaaac	65536
<i>SwaI</i>	atttaaat	65536