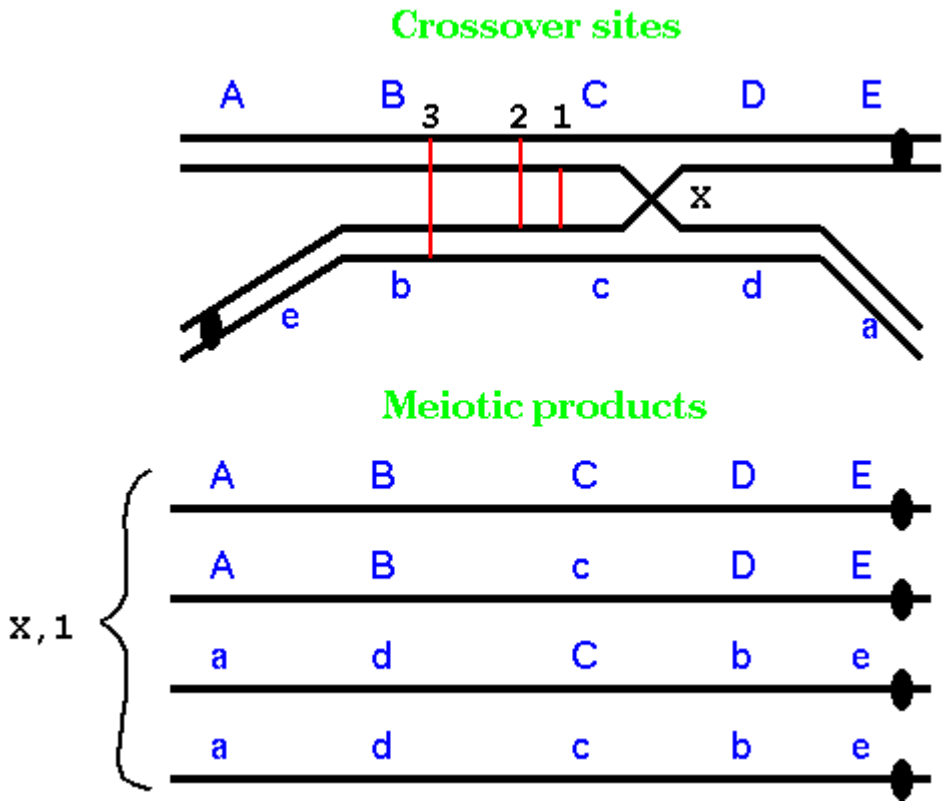
PLNT3140 INTRODUCTORY CYTOGENETICS
FINAL EXAMINATION

December 12, 2023, 1:30 pm - 3:30 **E2-130 EIT**

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.

1.) (10 points) Chromosome pairing in a heterozygote for an inversion is shown below. (For simplicity, an inversion loop is avoided by drawing the termini unpaired.) Consider a set of possible double crossovers. In all cases, one crossover event occurs at position X. The example shows meiotic products resulting from a second crossover at site 1. Draw both of the meiotic products if the second crossover site was at either 2 or at 3.



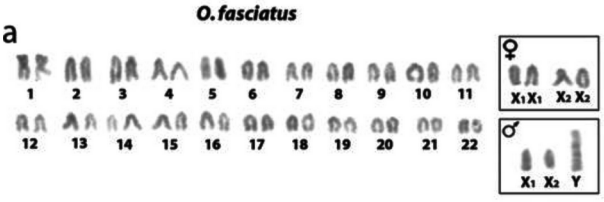
2. (5 points) In the table below, the genetic lengths are given for all human chromosomes, with the exception of Y. Why are no results shown for the Y chromosome?

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

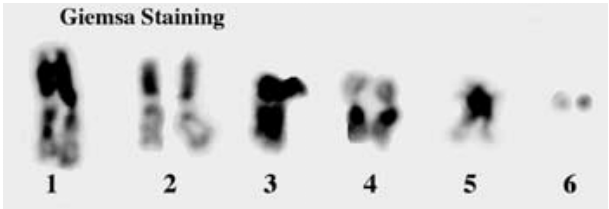
3. (10 points) Karyotypes of the fish *Oplegnatiathus* and the fruit fly *Scaptodrosophila* are shown below.

For which of the two species would it be more difficult to create a chromosome painting kit that distinguishes each chromosome with a unique fluorescent spectrum.? Explain your reasoning, based on what you know about about the steps that go into creating such a kit.

A *Oplegnatiathus fasciatus*



B *Scaptodrosophila hibisci*



4. (10 points) The accompanying table lists the lengths of chromosomes in the rat (*Rattus norvegicus*).

Loc	*Type*	*Name*	*RefSeq*	*INSDC*	*Size (Mb)*
Nuc	Chr	1	NC_005100.4	CM000072.5	282.76
Nuc	Chr	2	NC_005101.4	CM000073.5	266.44
Nuc	Chr	3	NC_005102.4	CM000074.5	177.7
Nuc	Chr	4	NC_005103.4	CM000075.5	184.23
Nuc	Chr	5	NC_005104.4	CM000076.5	173.71
Nuc	Chr	6	NC_005105.4	CM000077.5	147.99
Nuc	Chr	7	NC_005106.4	CM000078.5	145.73
Nuc	Chr	8	NC_005107.4	CM000079.5	133.31
Nuc	Chr	9	NC_005108.4	CM000080.5	122.1
Nuc	Chr	10	NC_005109.4	CM000081.5	112.63
Nuc	Chr	11	NC_005110.4	CM000082.5	90.46
Nuc	Chr	12	NC_005111.4	CM000083.5	52.72
Nuc	Chr	13	NC_005112.4	CM000084.5	114.03
Nuc	Chr	14	NC_005113.4	CM000085.5	115.49
Nuc	Chr	15	NC_005114.4	CM000086.5	111.25
Nuc	Chr	16	NC_005115.4	CM000087.5	90.67
Nuc	Chr	17	NC_005116.4	CM000088.5	90.84
Nuc	Chr	18	NC_005117.4	CM000089.5	88.2
Nuc	Chr	19	NC_005118.4	CM000090.5	62.28
Nuc	Chr	20	NC_005119.4	CM000091.5	56.21
Nuc	Chr	X	NC_005120.4	CM000092.5	159.97
Nuc	Chr	Y	NC_024475.1	CM002824.1	3.31
MT	Chr	MT	NC_001665.2	AY172581.1	0.016313
	Un	-	.	-	88.16
TOTAL					2870.2063

a) Using the Clark and Carbon formula, calculate the number of BAC clones needed to ensure a 99% chance of finding at least one clone for any given gene. Assume that the BAC library has an average insert size of 100 kb.

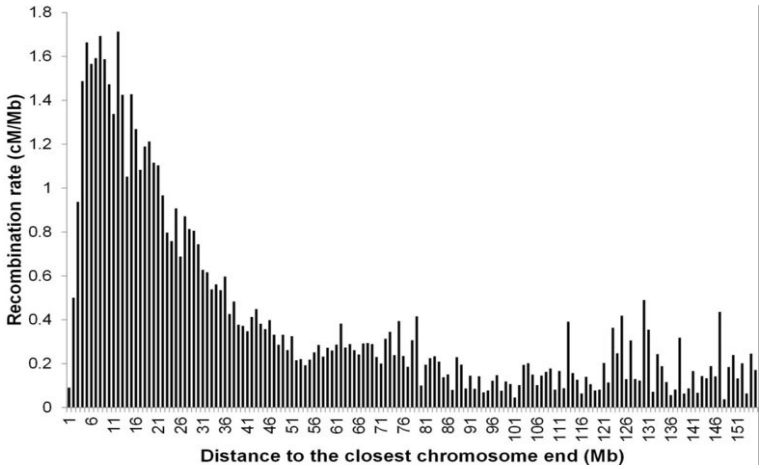
$$N = \frac{\ln(1-P)}{\ln(1-f)}$$

b) Suppose that you didn't care about getting a complete genomic library, but rather were only interested in getting genes from chromosome 18. (Assume flow cytometry is not an option.) Does that make any difference to your cloning strategy? Explain why or why not.

5. (5 points) When haploids are produced in species such as wheat (1N = 21), one often sees that some chromosomes are partially paired with other chromosomes during meiosis. In principle, there is only one copy of each homologous chromosome. Why is pairing possible, in such an instance?

6. (10 points)

a) A high density linkage map of the pig (*Sus scrofa*) genome was made consisting of 38,599 SNP markers covering a genome whose total genetic length (averaged among several crosses) is about 2000 cM. The physical length of the genome is 2334 Mb. What is the average length, in Mb, of 1 cM in the pig genome?



b) The accompanying graph was made by measuring the recombination frequency every million bases along each of the 18 chromosomes. The graph shows pooled results for all positions on all chromosomes. What is the most important conclusion that can be drawn from these results?

Data from Tortereau F et al. (2012) A high density recombination map of the pig reveals a correlation between sex-specific recombination and GC content. BMC Genomics 201213:586 https://doi.org/10.1186/1471-2164-13-586.

7. (10 points) A contig was constructed from six overlapping BAC clones labeled **a**, **k**, **j**, **p**, **r** and **x**. In a chromosome walk, the order of clones in the contig can be determined by hybridizing BAC-end probes from one clone with each of the other clones. Draw a diagram illustrating the overlap of the six BAC clones in the contig. Notation: 1 and 2 refer to probes from either end of a BAC. For example, **a1** and **a2** are end probes from BAC clone **a**. Minus (-) means that a probe did not hybridize to any of the six clones.

BAC end probe	hybridizes to BAC clone
a1	j
a2	x
k1	-
k2	r
j1	r
j2	a
p1	x
p2	-
r1	k
r2	j
x1	a
x2	p

8. (10 points) In many species, transposons are scattered across the entire genome. There are usually so many that just about any gene should be within a short distance (in cM) from a copy of a transposon. One might think that transposons could be used as molecular markers for genetic mapping. Give at least 2 reasons why transposons cannot be used for genetic mapping.

9. (15 points) In a cross between two *Arabidopsis* lines, A and B, a map of one chromosome was constructed using a set of co-dominant markers. An excerpt of the mapping data for this cross is shown in panel I. At each locus, the marker is scored as being homozygous for the allele from parent A, homozygous for the allele from parent B, or heterozygous. The order of loci shown in the table corresponds to the order of those loci on the chromosome.

a) What is the predicted ratio for seeing A, H or B, at any given locus?

b) In cross II, parent A was crossed with another *Arabidopsis* line, C. Thus, the expected phenotypes would be either A, H or C. In this cross, the mapping data look similar to that found in cross I. However, all loci distal to g3883 exhibit only the A phenotype, in all progeny. What is a simple explanation for this result?

c) Based on your answer to b, what cross could you do to test your hypothesis? What would be the results if you hypothesis was true?

I. A x B

```
        segregating progeny ------------------->

marker/
map posn.
g6844   HHAAAAABHHBAAAHBHHHHABHHHABBAHHBHAHHBAAHHA
g3843   HHAAAAABHHBAAAHBHHHHABHHHAHBAHHBHAHHBAABAA
g2616   HHAAHHHBHHBAAAHBHHHABHHHHHHHBBHBHHAHHHHHHH
m210    HHAHHBHHHHHAAAHHBHHHAHHAHAHHHAABHHAHBHABAA
g6837   HHAABHHAHBHHBAAHBHHHAHHAHAHHHAABHHAHBHABAA
g10086  AHHHAAHHHAHBHHBAHHHHAHHAHAHHHAAHHHAHBHHBAB
g4564a  HAAHHBHHHHHAAAHHBHHHAHHAHAHHHAAHHHAHBHHBAA
g3845   HAHHHBHHHAHAAAAHBHHAHBAHAHHHAAHHHAHBHHHAHH
g4539   AHHHAAHHHAHBHHHAHHHAAHBAHAHHHHAHHBAHBHHHAH
m557    HAHHHBHHHAHAAAAHBHHAAHBAHAHHHHAHHBAHBHHHAH
g3883   HAHHHBHHHAHAAAAHBHHHAHBAHAHHHHAHHBAHBHHHAH
g19833  HAHAHBHHAHAAAAHBHHAHBAHHHHAHHHABAHHAHHBBAH
g19838  HAHHAHBHHAHAAHBHAHHBAAHBHHAHHBAAHHHHABHHAH
m272    HAHAHBHHHAHAAHBHAHHBAAHBHHAHHBAAHHHHABHHAB
g4513   HAHAHBHHHAHAAAAHBHAHHHBAAAHBHHAHBBAAHHHHAB
```

II. A x C

```
        segregating progeny ------------------->

marker/
map posn.
g6844   HHAAAAACHHCAAAHCHHHHACHHHACCAHHCHAHHCAAHHA
g3843   HHAAAAACHHCAAAHCHHHHACHHHAHCAHHCHAHHCAACAA
g2616   HHAAHHHCHHCAAAHCHHHACHHHHHHHCCHCHHAHHHHHHH
m210    HHAHHCHHHHHAAAHHCHHHAHHAHAHHHAACHHAHCHACAA
g6837   HHAACHHAHCHHCAAHCHHHAHHAHAHHHAACHHAHCHACAA
g10086  AHHHAAHHHAHCHHCAHHHHAHHAHAHHHAAHHHAHCHHCAC
g4564a  HAAHHCHHHHHAAAHHCHHHAHHAHAHHHAAHHHAHCHHCAA
g3845   HAHHHCHHHAHAAAAHCHHAHCAHAHHHAAHHHAHCHHHAHH
g4539   AHHHAAHHHAHCHHHAHHHAAHCAHAHHHHAHHCAHCHHHAH
m557    HAHHHCHHHAHAAAAHCHHAAHCAHAHHHHAHHCAHCHHHAH
g3883   HAHHHCHHHAHAAAAHCHHHAHCAHAHHHHAHHCAHCHHHAH
g19833  AAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAA
g19838  AAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAA
m272    AAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAA
g4513   AAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAAA
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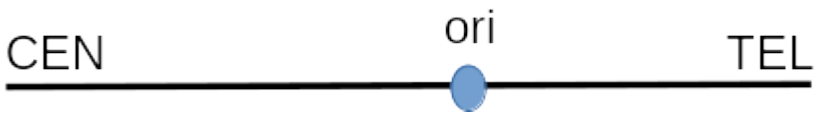
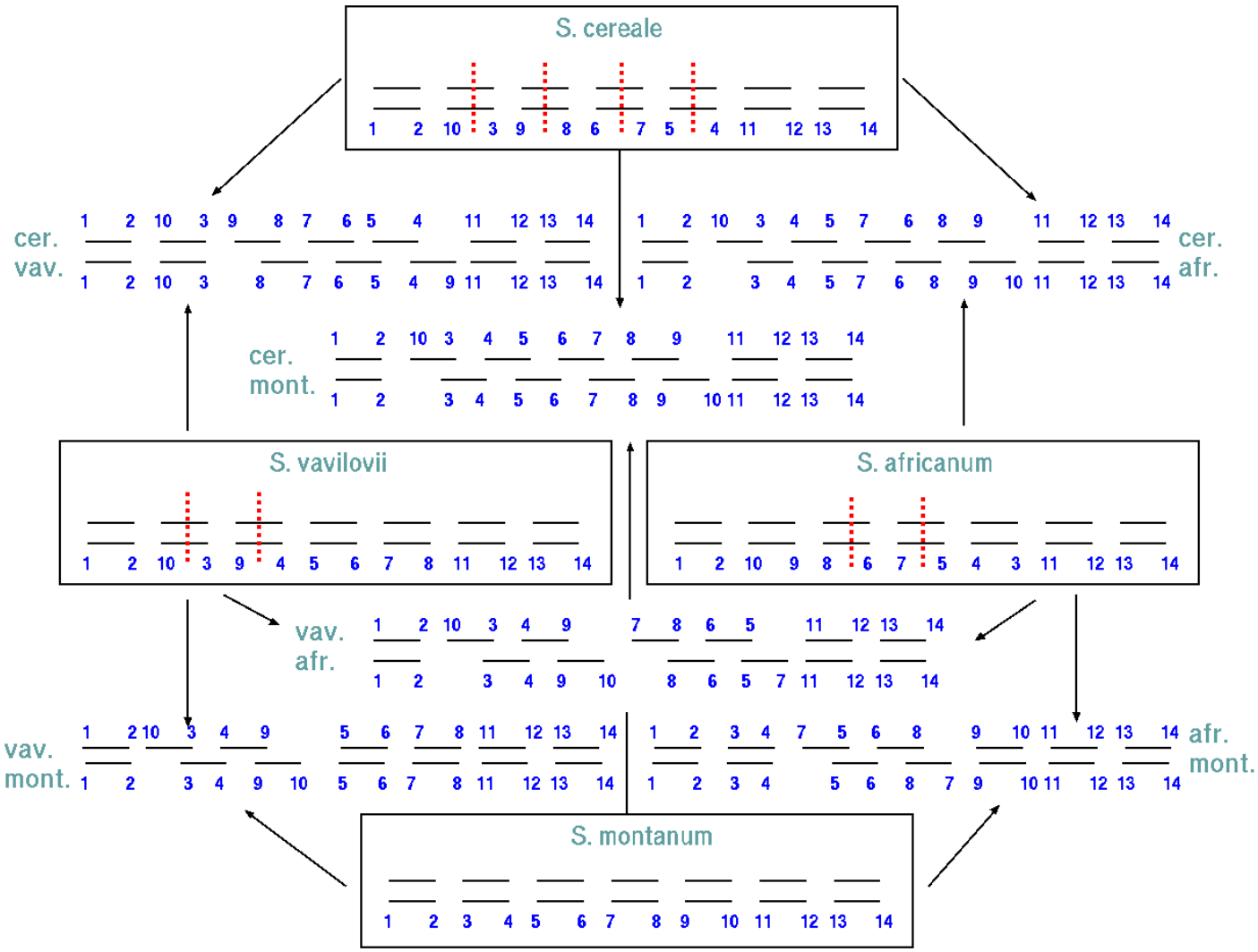
10. (5 points) Two samples of DNA were isolated from a human male donor, one from blood, and one from sperm. Using equal masses of DNA, C_0t curves were done for each sample. What differences, if any, would you expect to see between the two C_0t curves?

11. (5 points) The term telocentric is a bit of a misnomer. It implies something like this:

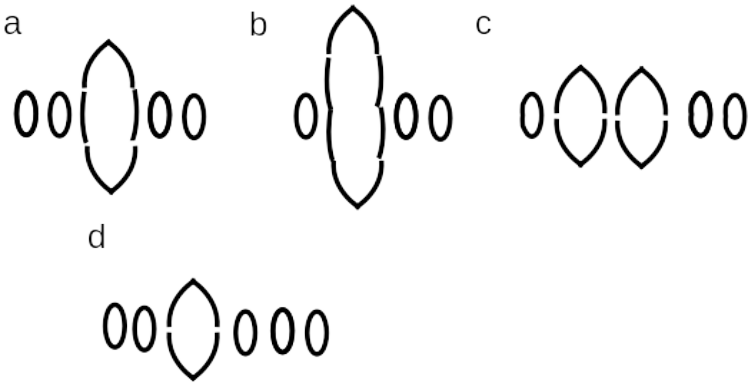


What is the problem with this chromosome? Draw a more realistic diagram depicting a telocentric chromosome.

12. (10 points) In class we described chromosome evolution using interspecific crosses of Secale species.



For each interspecific cross listed i - v, match observed meiotic pairing patterns (a - d). Some patterns maybe seen in two or more crosses. The chromosomes shown are in no particular order.



Answer in this order:

i) afr/mont

ii) vav/mont

iii) vav/afr

iv) cer/vav

v) cer/aft

13. (15 points) This question refers to data on the next page. Several inversions were found in polytene chromosomes of the dipteran midge Axarus varvestris. Inversion heterozygotes are shown for four chromosomes exhibiting inversions (Fig. 3). Arms are named using the letters A - G. Centromeres are indicated by arrows. For inversions on arms A, C, F and G, Tables 2 - 4 show the frequencies of larvae homozygous for the standard chromosome (AA), heterozygous for the inversion (Aa), or homozygous for the inversion (aa) at three locations (Fig. 1) on two different sampling dates. If the population was in Hardy-Weinberg equilibrium for each inversion, we would expect to see AA, Aa and aa progeny in the proportions of $p^2 : 2pq : q^2$, where q is the frequency of the inverted chromosome and frequency of the standard chromosome p is equal to 1-q. For example, from a sample of 90 larvae, given an allele frequency q = 0.44 of the C inversion, the expected number of aa larvae would be $0.44^2 \times 90 = 17.4$*. (Note: "p" in the table is NOT the same p as in the equation above, but rather refers to the probability of seeing these ratios by chance if the population was in HW equilibrium).

* - *The expected numbers of progeny in the table differ slightly from those predicted by the given value of q. The differences are small enough as to be attributable to rounding errors, for our purposes just ignore those differences.*

State whether the data on the next page would be informative regarding each of the following components of evolution:

a) Mutation
b) Migration
c) Selection
d) Random drift
e) Non-random mating

Briefly explain your reasoning. Hint: Take particular note of allele frequencies q for the different inversion.

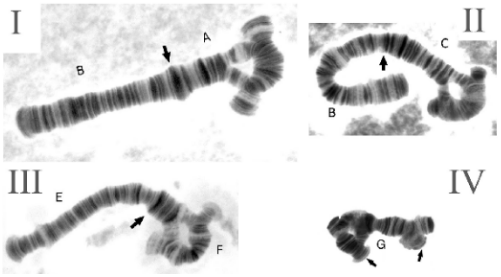


Figure 3.

Fig. 1. The three Connecticut River sites where larvae of the genus *Axarus* were collected.

River flows from North (top) to South (bottom)

Table 2. Data and calculations for Hardy–Weinberg equilibrium for the Putney, Vermont, collections.

Date collected	Inversion	*n*	AA	Aa	aa	*q*	G_{adj}	*p*
2 Aug. 2001	Obs. A_{1-5}	90	79	11	0	0.06		
	Exp. A_{1-5}		79.34	10.33	0.34		0.70	0.70
	Obs. C_{1-6}	90	30	41	19	0.44		
	Exp. C_{1-6}		28.34	44.33	17.34		0.50	0.78
	Obs. F_{13-20}	90	70	20	0	0.11		
	Exp. F_{13-20}		71.11	17.78	1.11		2.47	0.29
	Obs. G_{2-7}	90	59	31	0	0.17		
	Exp. G_{2-7}		61.67	25.66	2.67		6.43	0.04*
8 Dec. 2001	Obs. A_{1-5}	88	85	3	0	0.02		
	Exp. A_{1-5}		85.03	2.95	0.03		0.05	0.98
	Obs. C_{1-6}	88	27	41	20	0.46		
	Exp. C_{1-6}		25.64	43.72	18.64		0.34	0.84
	Obs. F_{13-20}	88	66	22	0	0.13		
	Exp. F_{13-20}		67.38	19.25	1.38		3.12	0.21
	Obs. G_{2-7}	88	45	43	0	0.24		
	Exp. G_{2-7}		50.25	32.49	5.25		14.03	<0.01*

Note: Obs., observed; Exp., expected; AA, standard arrangement homozygote; Aa, inversion heterozygote; aa, inversion

Table 3. Data and calculations for Hardy–Weinberg equilibrium for the Hinsdale–Northfield collections.

Date collected	Inversion	*n*	AA	Aa	aa	*q*	G_{adj}	*p*
23 July 2001	Obs. A_{1-5}	96	87	9	0	0.05		
	Exp. A_{1-5}		87.21	8.58	0.21		0.43	0.81
	Obs. C_{1-6}	96	37	47	12	0.37		
	Exp. C_{1-6}		38.13	44.74	13.13		0.24	0.89
	Obs. F_{13-20}	96	70	26	0	0.14		
	Exp. F_{13-20}		71.76	22.48	1.76		4.05	0.13
	Obs. G_{2-7}	96	58	38	0	0.20		
	Exp. G_{2-7}		61.76	30.48	3.76		9.39	0.01*
10 Nov. 2001	Obs. A_{1-5}	113	105	8	0	0.04		
	Exp. A_{1-5}		105.14	7.72	0.14		0.28	0.87
	Obs. C_{1-6}	113	45	56	12	0.35		
	Exp. C_{1-6}		47.16	51.68	14.16		0.79	0.67
	Obs. F_{13-20}	113	84	29	0	0.13		
	Exp. F_{13-20}		85.86	25.28	1.86		4.24	0.12
	Obs. G_{2-7}	113	47	66	0	0.29		
	Exp. G_{2-7}		56.64	46.73	9.64		27.87	<<0.01*

Note: See note to Table 2 for details.

Table 4. Data and calculations for Hardy–Weinberg equilibrium for the Shepherd's Island collections.

Date collected	Inversion	*n*	AA	Aa	aa	*q*	G_{adj}	*p*
7 Aug. 2001	Obs. A_{1-5}	101	100	1	0	0.00		
	Exp. A_{1-5}		100	1	0		0.00	1.00
	Obs. C_{1-6}	101	59	30	12	0.27		
	Exp. C_{1-6}		54.22	39.56	7.22		5.53	0.06
	Obs. F_{13-20}	101	75	26	0	0.13		
	Exp. F_{13-20}		76.67	22.65	1.67		3.81	0.15
	Obs. G_{2-7}	101	50	51	0	0.25		
	Exp. G_{2-7}		56.44	38.12	6.44		17.44	<<0.01*
1 Dec. 2001	Obs. A_{1-5}	83	82	1	0	0.01		
	Exp. A_{1-5}		82	0.99	0		0.00	1.00
	Obs. C_{1-6}	83	52	28	3	0.20		
	Exp. C_{1-6}		52.48	27.04	3.48		0.11	0.95
	Obs. F_{13-20}	83	65	18	0	0.11		
	Exp. F_{13-20}		65.98	16.05	0.98		2.16	0.34
	Obs. G_{2-7}	83	44	39	0	0.23		
	Exp. G_{2-7}		48.58	29.84	4.58		12.06	<0.01*

Note: See note to Table 2 for details.