BB2920-C17

Genetics Name: Solutions

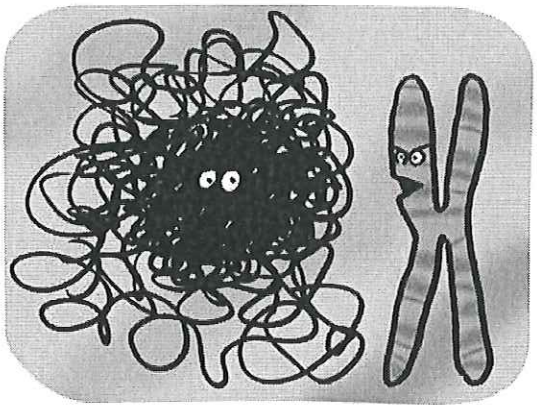
Exam 2

Professor Farny

Instructions:
Do not open this exam until instructed to do so.
You will have 60 minutes to complete the exam.
You may not leave the examination room during the exam.
Phones, tablets, computers, and any other electronic device are strictly prohibited. They must be completely out of sight for the entirety of the exam.

You may use a calculator. Phones, tablets, laptops, etc. may not be used as calculators. It must be a separate, regular calculator.

Helpful hint! Even if a problem doesn't ask for it, it can be very helpful to quickly sketch out a simple Punnett square or a schematic of the chromosome for linked alleles in order to visualize the problem!



Dude, mitosis starts in five minutes...
I can't believe you're not condensed yet.

http://www.promega.com/

Punnett Square showing standard dihybrid cross:

paternal gametes \ maternal gametes	**AB**	**Ab**	**aB**	**ab**
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Question:	possible points	points received
1	18	
2	18	
3	16	
4	16	
5	10	
6	8	
7	8	
8	6	
Total:	100	Score:
Bonus!	4	Final score

Question 1 (18 points): Examine the cross below. Identify the parental type offspring and recombinant offspring by placing a P or an R, respectively, in the boxes to the left of each offspring category. On the line to the right of each offspring category, give the genotype of those offspring.

A pure-breeding mouse with brown fur and rounded ears (B/B · R/R) is crossed with a pure-breeding mouse with white fur and pointed ears (b/b · r/r). The F1 generation from the parental cross are then testcrossed. The ratio of the offspring from the F1 testcross are as follows:

BR/br × brbr

P or R:	phenotype:	# of offspring	genotype:
R	brown fur and pointed ears	11	Br / br
P	brown fur and rounded ears	73	BR / br
R	white fur and rounded ears	14	bR / br
P	white fur and pointed ears	67	br / br

total 165

1 ea

1 ea

Bb RR

Are these genes linked? (Yes) No (circle one)

a) In a single sentence, explain your response (WHY do you think there are linked or unlinked?)

2 Parental and recombinant offspring categories are not found in equal proportions.

b) If yes, calculate the distance between these loci (show your work):

4

$$\frac{\text{recombinants}}{\text{total}} \times 100\% \quad \frac{(11+14)}{165} = \sim 15\% \text{ recombinant}$$

$$= 15 \text{ m.u.}$$

c) As stated in the problem above, the B/B alleles result in a mouse with brown fur, and the b/b alleles result in a mouse with white fur. Based on the information from your F1 testcross, do you think the B allele is haplosufficient or haploinsufficient? Briefly explain your reasoning.

4

Haplosufficient - because Bb individuals are brown (not a lighter brown or intermediate phenotype) So one dominant copy of B is enough for the fully brown phenotype.

Question 2 (18 points)

2n = 14

The Tasmanian devil has a total of 14 chromosomes in its somatic cells in G1. For each of the situations below, circle the ploidy of the cell, the number of genomes present ("n") and write in the ***total number*** of chromosomes present.

each circle = 0.5

	Ploidy	Genomes	Chromosomes
a) somatic cell in prophase:	haploid (diploid)	n 2n (4n)	28
b) meiocyte in prophase I:	haploid (diploid)	n 2n (4n)	28
c) mature gamete:	(haploid) diploid	(n) 2n 4n	7
d) meiocyte in prophase II:	(haploid) diploid	n (2n) 4n	14
e) somatic cell in telophase:	haploid (diploid)	n (2n) 4n	14
f) somatic cell in G1	haploid (diploid)	n (2n) 4n	14

g) Which pair, homologous chromosomes or sister chromatids, has the more identical DNA sequence? (Assume for this question that you are considering the pairs before crossing-over occurs).

2 sister chromatid

h) Which pair, homologous chromosomes or sister chromatids, is separated during anaphase II?

2 Sister Chromatid

i) Among which pair, homologous chromosomes or sister chromatids, does crossing over occur?

2 homologous chrom.

j) How many unique gametes can a Tasmanian devil make ONLY considering the process of independent assortment? Show your work.

3 pts

$2^n = 2^7 = 128$

2^{14} (1 pt)

Question 3: (16 points) A man with a specific unusual genetic trait marries an unaffected woman and they have children. Possible pedigrees for this family are shown below, but the presence or absence of the trait in the children is not given. For each type of inheritance, give the genotype of the father (using A or a as allele designations) AND indicate the expected genetic status of the offspring by filling in the symbols for the children as appropriate.

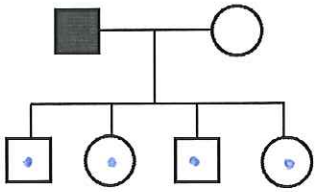
Affected males or females should be shaded in: ■ ●

Carrier males or females should be indicated by placing a dot in the middle of the symbol: ⊙ ⊡

IMPORTANT NOTES: The mother is NOT affected and is NOT a carrier. The disease allele(s) exist only in the father. Assume that the perfect expected ratio is obtained: for example, only half of one sex of the offspring is expected to be affected, shade only one of the two symbols for that sex; if all of one sex are carriers, indicate carrier status for all of that sex of offspring. If you are stuck, start by identifying the father's and mother's genotypes and sketch a Punnett square!

a) Autosomal Recessive

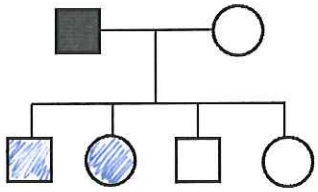
father's genotype: aa



all are carriers
none are affected

b) Autosomal Dominant

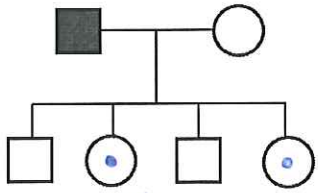
father's genotype: Aa



no carriers,
½ males & females affected
(AA is acceptable only IF your pedigree matches your genotype! - all offspring would be affected)

c) X linked Recessive

father's genotype: $X^a y$

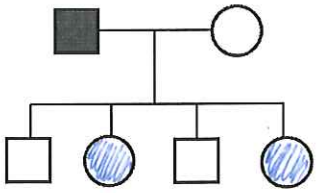


$X^a X^A$

all females are carriers
all males unaffected

d) X linked Dominant

father's genotype: $X^A y$



$X^A X^a$

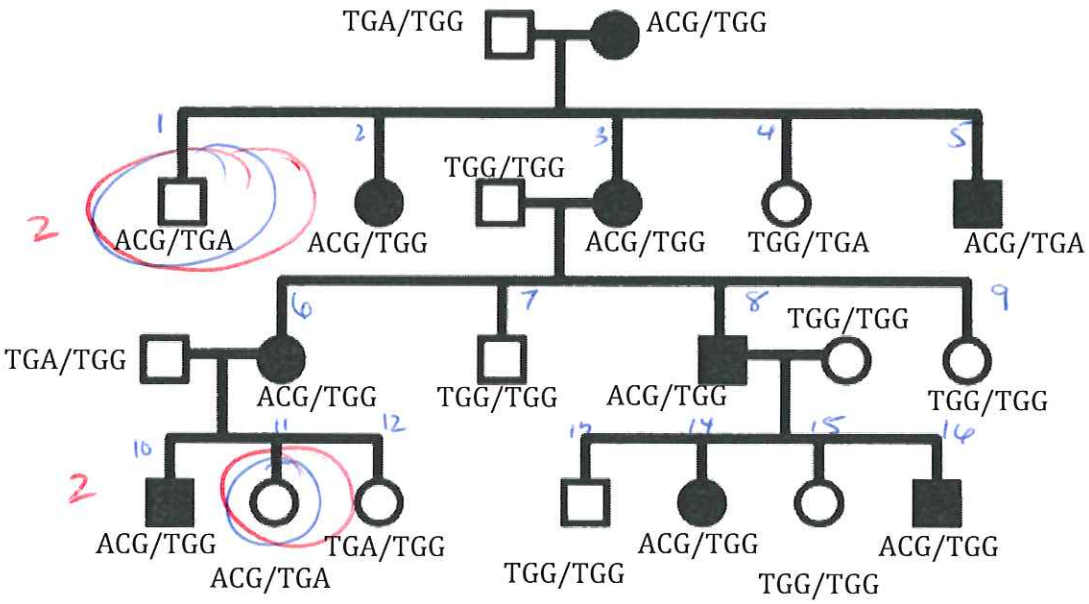
all females affected
all males unaffected

* note for X-linked it is not possible for a male to be a carrier.

Question 4 (16 points) – Below is a pedigree for a family affected by Huntington's disease. Adjacent to each family member is the haplotype for a small SNP series on Chr 4. The disease allele and the SNPs *are linked*.

There are three possible haplotypes at this SNP locus:

haplotype designation:	Sequence:
ACG	A C A G C A T C A T T A C G T
TGA	T C A G G A T C A T T A C A T
TGG	T C A G G A T C A T T A C G T



2 a) What is the inheritance pattern? Auto Dom.

b) Based on the information from the pedigree, sketch the chromosomes of the two parents in generation I in the space below. Use the allele designations H and h for the Huntington's gene alleles.

6 (1.5 per chrom.)

h TGA | h TGG

H ACG | h TGG

(2+2) c) Circle any recombinant individuals in the pedigree.

d) Based on this pedigree, what is the distance between the SNP locus and the disease locus? Show your work.

4 16 offspring, 2 recomb.

2/16 = 1/8 or 12.5% ⇒ 12.5 map units

Question 5 (**10 points, 1 pt each, no partial credit)** Match the terms to the correct descriptions (each definition will match only once):

Write the letter of corresponding the definition here ↓

1. co-dominance F
2. pleiotropy I
3. epistasis J
4. polygenic inheritance C
5. expressivity H
6. complementation A
7. haplosufficient D
8. penetrance E
9. recessive lethal mutation G
10. incomplete dominance B

A. When mutations in two different genes result in the same phenotype, crossing these two mutants will result in a wild-type phenotype

B. The dominant allele is haploinsufficient, causing the phenotype of the heterozygote to be intermediate to the phenotypes of the two homozygotes

C. Multiple genes contribute to a single phenotype

D. A single dominant wild-type allele confers enough of a gene dosage to result in a wild-type phenotype

E. The percentage of individuals with a given allele that display the associated phenotype

F. A gene that has multiple alleles, two of which can be expressed simultaneously in the heterozygote

G. An allele that causes the death of the organism in the homozygous recessive state

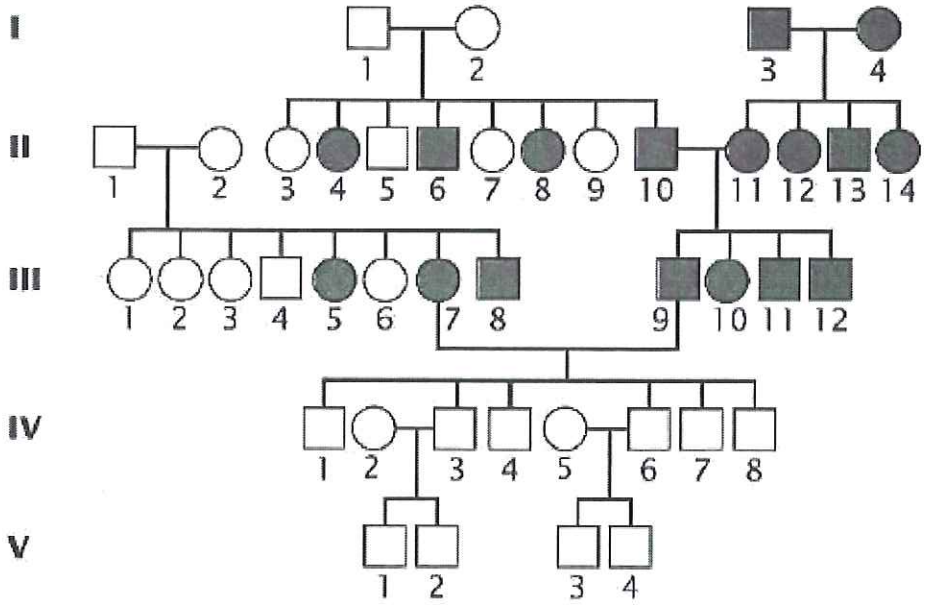
H. The degree to which a given allele is expressed at the phenotypic level

I. A single gene that affects more than one phenotype

J. The effect of a gene interaction where the genotype of one gene masks the effects of a gene at another locus

Question 6 (8 points)

Below is an extended, multi-generational pedigree a family with congenital blindness.



a) Consider ONLY generations I, II and III: What is the most likely inheritance pattern? A.R. autosomal recessive

b) What is the most likely explanation for the phenotypes of offspring in generations IV and V? Is this consistent with your suggested inheritance pattern above? Briefly explain.

Complementation!

Individuals I-1 and I-2 are carriers for a mutation. Individuals I-3 and I-4 are affected by a mutation that is in the same gene (allelic!) as individuals I-1 and I-2.

Persons II-1 and II-2 are carriers for a mutation in a different gene that still results in blindness.

When persons III-7 and III-9 mate, their mutations will complement, resulting in all seeing offspring.

Question 7: (8 points) Sickle cell anemia is an autosomal recessive condition caused by a single mutation within the oxygen-carrying protein hemoglobin. Interestingly, individuals with the mutant hemoglobin allele are protected against infection from the malaria parasite. While sickle cell disease is highly dangerous and can be lethal, the mutant hemoglobin allele is found at a very high frequency in populations in many locations where malaria is a constant health threat. Heterozygotes for the sickle cell allele do not have symptoms of sickle cell disease.

Is this an example of polygenic inheritance, incomplete dominance, pleiotropy, complementation or epistasis? Explain your response.

- Pleiotropy - because the hemoglobin gene controls both hemoglobin shape and malaria sensitivity.
- polygenic inheritance, complementation, and epistasis are not relevant here.
- A case can be made for incomplete dominance of the hemoglobin allele at the molecular level, that results in partially sickled cells that don't cause disease but do prevent malaria infection.

Question 8: Journal Club (6 points): In the articles we read for Journal Club 2 by Huang et al. and Horn et al.:

a) What region of the TERT gene was found to have mutations in many melanomas? promoter

b) (Circle one) These mutations **increased/decreased** the expression of telomerase in melanoma cells.

c) The function of telomerase is (circle one):

A. to maintain cells in a G0 state
B. to prevent the conversion of normal cells into cancer cells
C. to maintain the ends of linear chromosomes
D. to ligate Okazaki fragments on the lagging strand

d) Based on the pedigree below, the most likely inheritance pattern for the inherited melanoma is:

Auto. Dom.

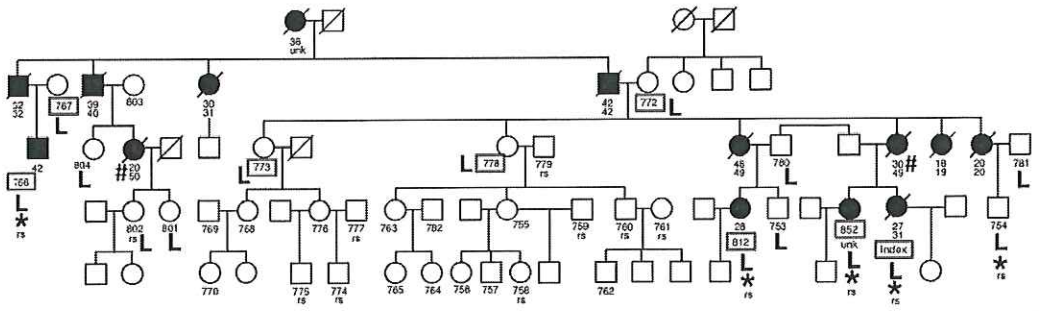


Fig. 1. Pedigree of melanoma-prone family. Four generations were affected by melanoma (solid symbols; circles represent females, and squares represent males). After linkage analysis carried out on 15 family members (L), HTS was performed on four affected and four unaffected individuals (boxed samples). A mutation in the *TERT* promoter was identified in all affected members and one unaffected individual (stars). Strikethrough symbols indicate deceased individuals. Two-digit numbers are age at onset of melanoma and age at death; Unk, unknown; Rs, rs2853669 observed in heterozygous form; three-digit numbers, DNA available; #, affected by other cancers; and index, index patient.