EXAM 1

18 QUESTIONS, 50 MINUTES

CALCULATORS ALLOWED (AND HELPFUL)

NO CELL PHONES OR OTHER SUCH DEVICES

LECTURE 4: EXTENSIONS OF MENDELIAN GENETICS, II
JULY 11, 2011

Extensions of Mendelian Genetics (Chapter 5)

- Penetrance and Expressivity
  - Lethal Alleles
  - Pleiotropic Alleles
  - Multiple alleles at one locus
  - Genetic Interactions
    - Understanding Genetic Interactions With Epistasis

PENETRANCE

- Penetrance is the percentage of individuals with a particular genotype that express the expected phenotype.
  - Incomplete penetrance: Penetrance < 100%
  - e.g. human polydactyly
  - Trait is usually caused by a dominant allele.


PENETRANCE

- Sometimes people have the allele for polydactyly, but have normal numbers of digits.
- If we examined 42 people with an allele for polydactyly, we might get the following results:


38 4

Penetrance = 38/42

= 0.90

= 90%

EXPRESSIVITY

- Expressivity describes the degree to which a genotype is phenotypically expressed.
  - e.g. Polydactyly also exhibits variable expressivity:
    - individuals with an allele for polydactyly:
      - some have completely functional extra digits
      - some have only a small tag of extra skin.

EXPRESSIVITY

- Expressivity describes the degree to which a genotype is phenotypically expressed.
  - e.g. Neurofibromatosis is almost 100% penetrant but has variable expressivity:
    - Skin pigmentation defects on some individuals
    - Neural tumors on other individuals.

WHAT EXPLAINS INCOMPLETE PENETRANCE /VARIABLE EXPRESSIVITY?

- Other genes and/ or environmental factors are responsible for different degrees of both penetrance and expressivity.
  - e.g. A gene may encode an enzyme that produces a particular phenotype only within a limited temperature range.
  - At higher or lower temperatures, the enzyme may not function and the phenotype may not be expressed.
- Many characters exhibit incomplete penetrance and variable expressivity.

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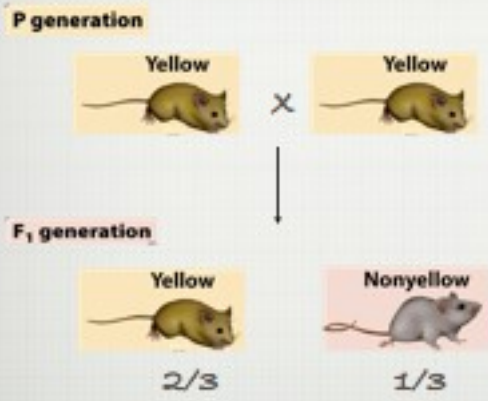
LETHAL ALLELES

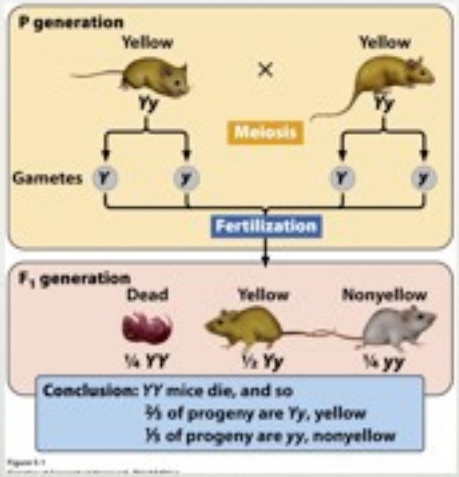
- cause death at an early stage of development, often before birth.
  - →Therefore some genotypes may not appear among progeny.
- e.g. the allele for yellow coat color in mice is lethal

LETHAL ALLELES

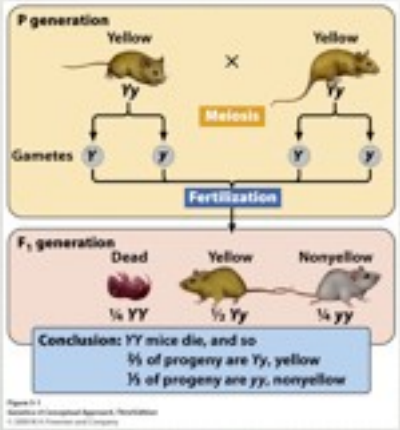


- No true-breeding yellow mice (all heterozygous)
- The ratio of yellow to nonyellow was 2:1
- Why? (What class is missing?)
- The allele for yellow is lethal when homozygous

LETHAL ALLELES



LETHAL ALLELES



- In this cross, the Y allele is pleiotropic: it has more than one phenotype.
- One is dominant; one is recessive
- What are these phenotypes and which is which?

PLEIOTROPY

- One mutation with multiple effects
- Most genes function many times in different tissues/organs
- A single mutation can thus affect many different systems

EXAMPLE: MARFAN SYNDROME

- Mutation in fibrillin gene, which encodes a connective tissue protein
- Functions in: lens of eye, blood vessel lining, bones, heart, all connective tissues

EXAMPLE: MARFAN SYNDROME

- Mutation causes: lens dislocation, aortic aneurism, long limbs, increased flexibility of joints

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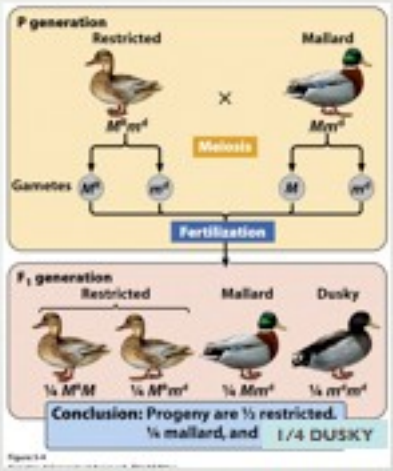
MULTIPLE ALLELES AT A SINGLE LOCUS

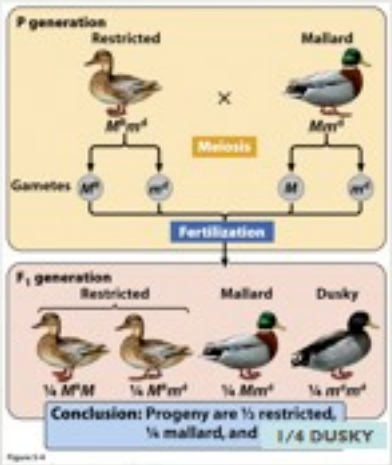
- Multiple alleles in a group
- Individuals have two alleles
- Example: Duck Feather Pattern
- M^R ☒ restricted pattern
- M ☒ wild-type mallard pattern
- m^d ☒ dusky pattern



MULTIPLE ALLELES AT A SINGLE LOCUS

- $M^R > M > m^d$

Genotype	Phenotype
M^RM^R	restricted
M^RM	restricted
M^Rm^d	restricted
MM	mallard
Mm^d	mallard
m^dm^d	dusky



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GENETIC INTERACTIONS

- What happens when a trait is affected by more than one gene?
  - These interactions can take a variety of forms
- Epistasis: when an allele of one gene modifies or masks the expression of another gene

EXAMPLE: ALBINISM AND COAT COLOR

Gene 1: B = Black Gene 2: A = normal

b = gray a = albino

EXAMPLE: ALBINISM AND COAT COLOR

AABB (black) x aabb (albino)

AaBb

(black)

EXAMPLE: ALBINISM AND COAT COLOR

AaBb (black) x AaBb (black)

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

F_2 RESULTS:

- 9 Black: 3 Grey: 4 albino
- Modification of 9:3:3:1 ratio - but progeny are still measured in 16th's, indicating two Mendelian genes

SO:

- The phenotype at the B locus is masked by the genotype at the A locus
- A is epistatic to B; or "albinism is epistatic to black/gray"

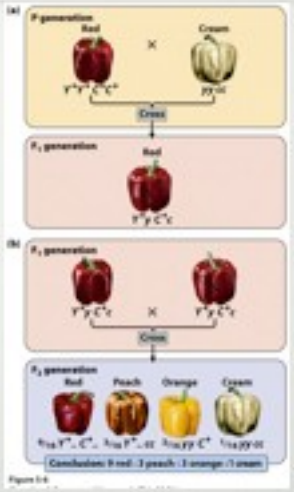
EPISTASIS

- Genotype ratios will still be 9:3:3:1
- Use the F2s to determine which phenotypes result from which genotypes

ANOTHER EXAMPLE: ADDITIVE EFFECTS

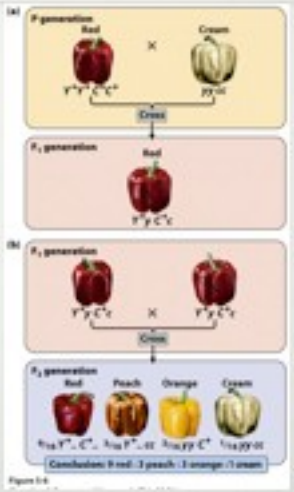
Fruit color in peppers

- Y and C loci both encode enzymes that produce pigments.
- The amount of these pigments determines the fruit color.



ANOTHER EXAMPLE: ADDITIVE EFFECTS

Genotype	Phenotype
$Y^+\_\ C^+\_$	red
$Y^+\_\ cc$	peach
$yy\ C^+\_$	orange
$yy\ cc$	cream



GENETIC INTERACTIONS: NOVEL PHENOTYPES

- Summer squash: shapes
- Cross true-breeding disc-shaped squash with true-breeding long squash
- F1 are all disc-shaped

GENETIC INTERACTIONS: NOVEL PHENOTYPES

- F2 ratios: 9 discs: 6 spheres: 1 long
- What do these ratios indicate?
- The F1s were presumably hets at both genes, so:

EXPECTED F2 GENOTYPES:

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

9 discs: A_B_

EXPECTED F2 GENOTYPES:

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

6 spheres: Add the two "3"
categories: A_bb and aaB_

EXPECTED F2 GENOTYPES:

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

1 long: aabb

SO:

- Having at least one copy of each dominant allele: disc shape
- Having only one copy of either dominant allele: sphere shape
- Being homozygous for both recessive alleles: long shape

FOR NEXT TIME:

- WE'LL FINISH "EXTENSIONS OF MENDELIAN GENETICS"
- READ CHAPTER 6: PEDIGREE ANALYSIS AND GENETIC MODEL ORGANISMS