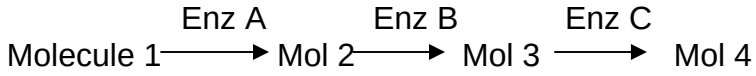
CLASS NOTES FOR CH 13, DAY 2: EPISTASIS

Gene Interactions produce new phenotypes
Genes can work together in a pathway.



Combine what we know about pathways and mutants with Mendelian crosses.

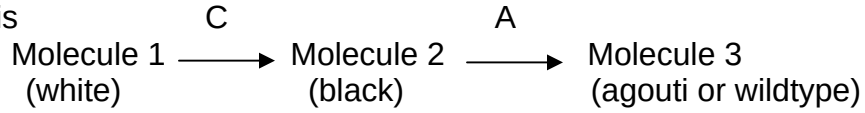
Epistasis
The interaction of 2 or more genes to control a single phenotype.

[Example: Comb shape in chickens
Walnut comb is dominant for both loci; Rose comb is dominant for R and recessive for p (R/_ p/p); Pea comb is dominant for P and recessive for r (r/r P/_); and single comb is recessive for both (r/r p/p)
Note this is different from complementation such as body color in fruit flies.

Problem: R/r P/p X R/r P/p
a) What is the phenotype of these individuals?
b) What are the gametes produced?
c) Do the Punnett square.
d) What is the proportion of the different combs?]

Recessive Epistasis
Example: Mouse coat color

Probably pathway is



A mutation in Enzyme A will accumulate molecule 2 and the mouse is black.
A mutation in Enzyme C will accumulate molecule 1 and the mouse is white.
If there's a mutation in Enzyme C, genotype of Enzyme A is irrelevant.

Do A/a C/c X A/a C/c (double heterozygous cross)

Coat color in rodents:
A/_ C/_ is agouti (9)
a/a C/_ is black (3)
a/a c/c and A/_ c/c are albino (3+1 or 4)

<u>Recessive epistasis shows 9:3:4 ratio with a double heterozygous cross</u>

Similar to coat color in Labrador retrievers:

Black lab is B/_ E/_

Yellow lab is b/b e/e or B/_ e/e

Chocolate lab is b/b E/_

Probable pathway is

$$\text{Mol 1 (yellow)} \xrightarrow{E} \text{Mol 2 (chocolate)} \xrightarrow{B} \text{Mol 3 (black)}$$

How many black, chocolate and yellow labs if two labs (each B/b E/e) produce 2 litters of 8 puppies each?
[Ans: 9 black puppies, 3 chocolate lab puppies and 4 yellow lab puppies]

Dominant Epistasis

Example: Summer squash color

Probable pathway is white $\xrightarrow{\text{inhibitor W}}$ green $\xrightarrow{Y}$ yellow

Gene Y converts green to yellow; yy keeps green

Gene W is <u>inhibitor</u> for enzyme to convert white to green;

When W/_, keeps color white; w/w allows green

Summer squash color

W/_ Y/_ (9) or W/_ y/y (3) is white squash

w/w Y/_ (3) is yellow squash

w/w y/y (1) is green squash

<u>Dominant epistasis gives a 12:3:1 ratio in a double heterozygous cross</u>

Dominant and Recessive Epistasis

Hypothetical flower color

$$\text{Orange 1} \xrightarrow{R} \text{Orange 2} \xrightarrow{\text{Inhibitor S}} \text{Red}$$

R/_ S/_ (9), r/r S/_ (3) and r/r s/s (1) are orange

R/_ s/s (3) are red

<u>Dominant and recessive epistasis will give 13:3 ratio in a dihybrid cross</u>

Epistasis of Duplicate Genes
Gene or genotype at 1 locus can produce phenotype identical to gene or genotype at another locus.

Duplicate recessive epistasis:
Example: Sweet pea flower color:

Probable pathway is white 1 $\xrightarrow{C}$ white 2 $\xrightarrow{P}$ purple

Flowers which are C/_ P/_ (9) are purple
Flowers which are C/_ pp (3), c/c P/_ (3) or c/c p/p (1) are white

Duplicate recessive epistasis shows 9:7 ratio in a dihybrid cross
AND all F2 whites breed true while 1/9 purple breed true.

Duplicate dominant epistasis:
Example: Fruit shape in shepherd's purse plant

Probable pathway is heart-shaped $\xrightarrow{\text{Inhibitor of H}}$ heart-shaped $\xrightarrow{\text{Inhibitor of N}}$ narrow

Plants that are H/_ N/_ (9), H/_ n/n (3) or h/h N/_ (3) are heart-shaped
Plants that are h/h n/n are narrow

Duplicate dominant epistasis shows 15:1 ratio with double heterozygous cross

Essential Genes and Lethal Alleles
If a gene is essential for life, a mutation in that gene is lethal.
May be recessive or dominant.

Recessive lethal allele:
Lethal if get 2 copies

Mice normally agouti. The yellow mutation, A^y, is lethal if A^y/A^y
Punnett square of A/A^y X A/A^y gives 2/3 A/A^y and 1/3 A/A
Yellow mutation is large deletion of chromosome that puts
Promoter of another region *Raly*, next to coat color gene.
Probably loss of *Raly* that is lethal.

Dominant lethal allele:
Lethal if only get a copy.
Hard to study unless organism first reaches reproductive age.

Example is Huntington's disease.

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Gene Interactions produce new phenotypes
Combine what we know about mutations in pathways with Mendelian crosses.

Epistasis
The interaction of 2 or more genes to control a single phenotype.

Recessive Epistasis
<u>Shows 9:3:4 ratio</u> with double heterozygous cross
Coat color in rodents:

A/A C/C is agouti; a/a c/c and A/_ c/c are albino; a/a C/_ is black
Do Punnett square of two A/a C/c individuals to get 9:3:4 ratios
Probable pathway is

albino $\xrightarrow{C}$ black $\xrightarrow{A}$ agouti molecule

Dominant Epistasis
<u>Shows 12:3:1 ratio</u> with double heterozygous cross
Summer squash color
W/_ Y/_ or W/_ y/y is white squash; w/w Y/_ is yellow squash; w/w y/y is green

Probable pathway is white $\xrightarrow{\text{inhibitor W}}$ green $\xrightarrow{Y}$ yellow

Epistasis of Duplicate Genes
Gene or genotype at 1 locus can produce phenotype identical to gene or genotype at another locus.

Duplicate recessive epistasis:
Sweet pea flower color:
Dihybrid cross gives 9:7 ratio (purple to white)
AND all F2 whites breed true while 1/9 purple breed true.
Probable pathway is white 1 $\xrightarrow{C}$ white 2 $\xrightarrow{P}$ purple

Duplicate dominant epistasis:
Fruit shape in shepherd's purse plant
Shows 15:1 ratio with double heterozygous cross (heart-shaped to narrow fruit)

Probable pathway is heart-shaped $\xrightarrow{\text{Inhibitor of H}}$ heart-shaped $\xrightarrow{\text{Inhibitor of N}}$ narrow

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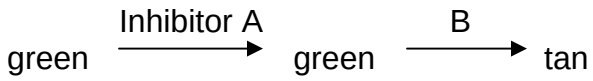
HOMEWORK FOR CHAPTER 13, DAY 2

1. Define the terms epistasis and duplicate epistasis.

2. In corn, the kernel color may be white, purple or red and is controlled by 2 genes, I and P. Corn that is I_ P_ is red; corn that is I_ pp is purple and corn that is iiP_ or iipp is white.
A dihybrid cross shows a ratio 9 red: 3 purple: 4 white
a) Which type of epistasis does this show?
b) What is the probable pathway involved?

3. A metabolic pathway in a plant seed is as follows:



a) What would be the expected ratios of green seeds to tan seeds in a dihybrid cross?
b) A total of 320 seeds from this cross are counted. How many of these seeds are green and how many are tan on average according to your calculations?

4. In cats, dd is lethal, with the kittens dying before birth and so are never seen.
a) Draw the Punnett Square of Dd x Dd.
b) What is the ratio of DD : Dd : dd?
c) Mutations in essential genes may be lethal. What are essential genes?
d) Think of an example of an essential gene.

OVERHEAD EXAMPLE OF EPISTASIS: LABRADOR RETRIEVERS

Labs can be black, chocolate or yellow with coat color determined by 2 genes.

Black true breeding lab, BBee is crossed with a yellow true breeding lab, bbEE.
F_1 generation is BbEe. Two F_1 dogs are crossed.

	BE	**Be**	**bE**	**be**
BE	**BBEE**	**BBEe**	**BbEE**	**BbEe**
Be	**BBEe**	**BBee**	**BbEe**	**Bbee**
bE	**BbEE**	**BbEe**	**bbEE**	**bbEe**
be	**BbEe**	**Bbee**	**bbEe**	**bbee**

What is the ratio of colors in labs?

What type of epistasis is involved?

What is the likely metabolic pathway involved?