## Mendel \& the Gene Idea, Part II Chapter 14, pp. 262-285

## Lecture Outline

- Laws of probabilities govern Mendelian inheritance
- Beyond Mendel... complex inheritance patterns
- Incomplete dominance
- Codominance and multiple alleles
- Epistasis
- Many human traits follow Mendelian inheritance patterns
- Pedigree analysis
- Carriers
- Pleiotropy
- Inherited disorders



## Allele: flower color

Purple $x$ white
$\mathrm{F}_{2}$ phenotype ratio $=3: 1$
Law of segregation


## Dihybrid

Allele: seed color and shape

## Yellow/round $x$ green/wrinkled

$\mathrm{F}_{2}$ phenotype ratio $=9: 3: 3: 1$
Law of independent assortmen

# Concept 14.2 <br> The laws of probability govern Mendelian inheritance 

What is the probability that $2 r$ (wrinkled seed) alleles will be present in BOTH gametes at fertilization?

- Use the multiplication rule (a.k.a. product rule)


For heterozygotes... What is the probability that the $R$ (round seed) allele will come from the egg $(R r)$ ? What is the probability it will come from the sperm $(r R)$ ?

- These are mutually exclusive events $\rightarrow$ use the addition rule (a.k.a. sum rule)


Concept 14.3 - Inheritance patterns are often more complex
than predicted by simple
Mendelian inheritance (complete dominance)

## Incomplete dominance



## Codominance and multiple alleles

| (a) The three alleles for the ABO blood groups and their <br> carbohydrates |  |  |  |
| :---: | :---: | :---: | :---: |
| Allele | $\boldsymbol{I}^{\boldsymbol{A}}$ | $\boldsymbol{I}^{\boldsymbol{B}}$ | $\boldsymbol{i}$ |
| Carbohydrate | A $\triangle$ | B $\bigcirc$ | none |


| (b) Blood group genotypes and phenotypes |  |  |  |  |
| :---: | :---: | :---: | :---: | :---: |
| Genotype | $I^{A} I^{A}$ or $I^{\boldsymbol{A}} \boldsymbol{i}$ | $I^{B} I^{B}$ or $I^{B} \boldsymbol{i}$ | $I^{A} I^{B}$ | ii |
| Red blood cel appearance |  |  |  |  |
| Phenotype (blood group) | A | B | AB | 0 |

## Epistasis

- A type of gene interaction in which the alleles of 1 gene masks the effects of a dominant allele of another gene
- E.g., coat color in mammals
- The epistatic gene determines whether pigment is deposited on the hair shaft


## Epistasis



## Concept 14.4: Many human traits follow Mendelian patterns of inheritance

## Pedigree analysis

| $\square$ Male | $\square$Affected <br> male | $\square \_\bigcirc$ | Mating |
| :--- | :--- | :--- | :--- |
| Female | Affected <br> female | $\square \square \square \square$ |  |
| Offspring, in <br> figi. <br> firth order <br> (first-born on left) |  |  |  |



Fig. 14-15
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Fig. 14-15

| 1st generation |
| :--- |
| (grandparents) |


| 2nd generation |
| :--- |
| (parents, aunts, |
| and uncles) |
| 3rd generation |
| (two sisters) |

Attached earlobe


Suppose the couple in the second generation decide to have 1 more child. What is the probability that the child will have a widow's peak and attached earlobes?
A. $5 / 16$
B. $3 / 16$
C. $11 / 16$
D. $5 / 8$
E. $1 / 8$

Hint: consider the rules of independent assortment and multiplication


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Independent assortment = dihybrid cross
Multiplication rule (3/4 x $1 / 4=3 / 16$ )


## Cystic fibrosis

- carriers
- pleiotropic gene



## Dominantly inherited disorders

Huntington's disease


Since the human genome was sequenced in 2003, there has been the development of over 2,000 different genetic tests available to prospective parents. If one parent tests positive and the other tests negative for a recessive allele associated with cystic fibrosis, what is the probability that their first child will have the disorder? What is the probability that their first child will be a carrier?
A. Have disorder: 100\%; Carrier: 0\%
B. Have disorder: $0 \%$; Carrier: $100 \%$
C. Have disorder: 0\%; Carrier: 50\%
D. Have disorder: 100\%; Carrier: $100 \%$
E. Have disorder: 50\%; Carrier: 50\%

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C. Have disorder: 0\%; Carrier: 50\%
D. Have disorder: 100\%; Carrier: 100\%
E. Have disorder: 50\%; Carrier: 50\%

## Chap 14, Lecture Review I

- Explain Mendel' s 2 laws of inheritance and how his pea plant experiments that led to these conclusions.
- Describe how to the laws of probability govern Mendelian inheritance patterns.
- Use a Punnett square to predict the results of a monohybrid and dihybrid cross and to state the phenotypic and genotypic ratios of the F2 generation
- Explain how phenotypic expression in the heterozygote differs with complete dominance, incomplete dominance, and codominance


## Chap 14, Lecture Review II

- Define and give examples of pleiotropy and epistasis
- Review how a pedigree analysis
- determines whether a character of interest is dominant or recessive
- predicts phenotypes of future generations
- Describe why recessively inherited disorders are common in human populations.
- Explain why lethal dominant genes are much rarer than lethal recessive genes.

