

Beyond Mendel

(chapters 14 and 15)

- Mendel published studies of exactly seven traits in peas
- All traits showed two phenotypes, one dominant, one recessive (either-or inheritance)
- The *mechanisms* Mendel discovered are universal.
 - "particulate" inheritance
 - parents with two alleles, gametes with one allele
 - segregation during reproduction
- We now know how all those relate to what chromosomes do in meiosis.
- However, we can also now explain many patterns of inheritance that Mendel could not.

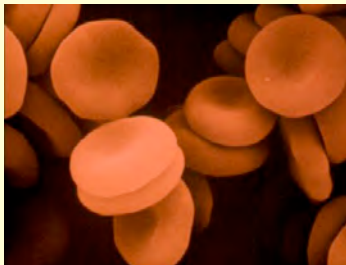
Incomplete dominance

- Phenotype of heterozygote is intermediate between phenotype of two homozygotes
- This is still different from blending inheritance



Multiple Alleles

- Some genes have more than just two alleles - Example: ABO blood types

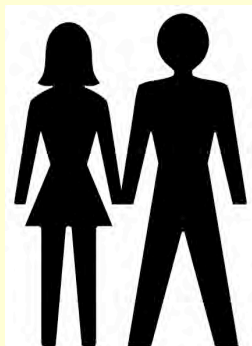


Conditional Expression

- Some genes are only expressed (i.e., affect your phenotype) under certain conditions.



Conditional expression in humans



- Secondary sex characteristics



Pleiotropy

- Pleiotropic genes have not just one effect, but many different effects

Pleiotropy

• Example: Sickle Cell Anemia

Caused by a single defective gene

- Anemia
 - Shortness of breath
 - Dizziness
 - Headache
 - Coldness in the hands and feet
 - Pale skin
 - Chest pain
- Sudden pain throughout the body
- Hand-foot syndrome: pain, swelling, fever
- Splenic crisis
- Difficulty fighting infections
- Pulmonary artery hypertension
- Delayed growth and puberty
- Eye problems
- Stroke
- Gallstones



Test Cross



• What is the genotype?

Test Cross

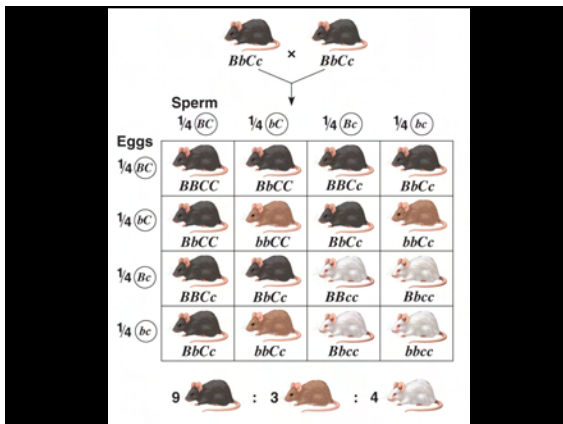


X



Epistasis

- One genetic locus affects expression of alleles at a second locus
- Example: albinism
- Aside: how do genes work?



Sex-linked Genes

- In mammals, females have two X chromosomes while males have an X and a Y
- The Y chromosome has few functional genes (Why?)
- Genes on the X chromosome will show distinctive patterns of inheritance
- In most cases, by "sex-linked," we mean "x-linked recessive," (locus is physically on the X chromosome)

- In mammals, females have two X chromosomes while males have an X and a Y
- Other sex-determining systems:
 - ZW females, ZZ males (birds, butterflies)
 - XX females, XO males (grasshoppers, roaches)
 - Diploid females, haploid males (ants, bees, wasps)

Linkage

- Two genes on the same chromosome are said to be linked.
- Linked genes also show distinctive patterns of inheritance

Independent assortment

Two equally probable arrangements of chromosomes at metaphase

Metaphase II

Gametes

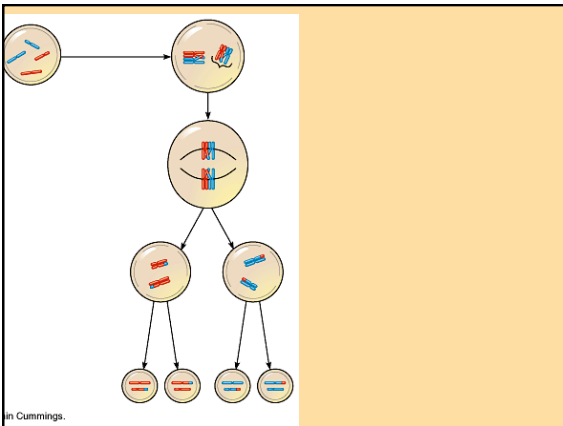
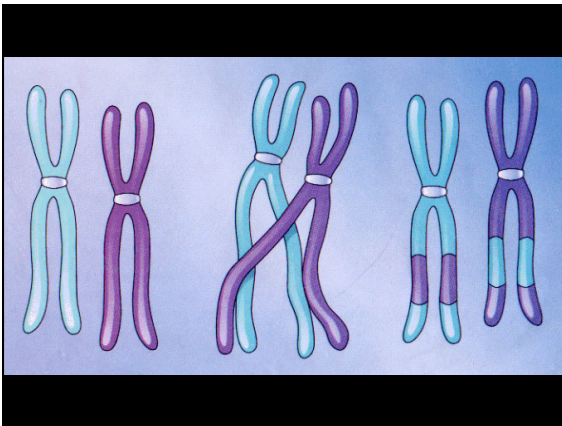
Combination 1, 2, 3, 4

P Generation (homozygous)
 Wild type (gray with normal wings) $b^+ b^+ vg^+ vg^+$ × Double mutant (black with vestigial wings) $b b vg vg$

F₁ dihybrid (wild type) (gray with normal wings) $b^+ b vg^+ vg$ × **Double mutant** (black with vestigial wings) $b b vg vg$

TESTCROSS

Offspring of testcross	Wild type $b^+ b vg^+ vg$	Black-vestigial $b b vg vg$	Gray-vestigial $b^+ b vg vg$	Black-normal $b b vg^+ vg$
Expected (independent assortment)	575	575	575	575
	Parental phenotypes		Recombinant phenotypes	
Observed	965	944	206	185



A Short Lesson in Probability

- All probabilities can be expressed as a number between 0 (never happens) and 1 (happens always).
 - Probability (heads) = 0.5



Probability

To find the probability of two or more independent events happening *together*, multiply the individual probabilities.
 P (three heads in a row) = $0.5 \times 0.5 \times 0.5 = 0.125$ or $1/8$

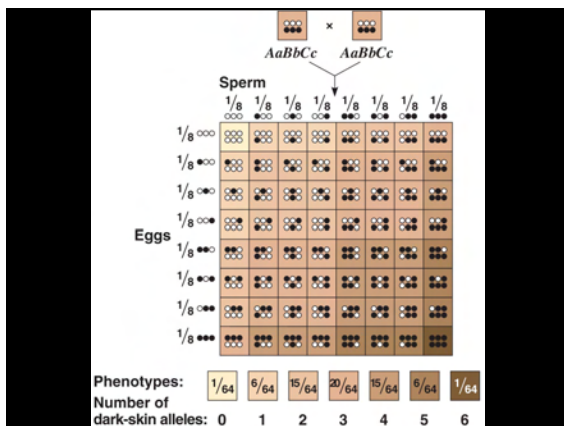


Probability

- If there are two or more ways to get to the same result, add the probability of each way.
- Examples: (dice, gamete formation)

Polygenic traits

- Traits affected by alleles at many genes.
- Polygenic traits show a continuous distribution of phenotypes
- Examples: height in humans, skin color in humans
- There are more of these polygenic traits than there are simple mendelian traits



Did Mendel cook his data?

- In peas the number of chromosomes is $2n = 14$
- Mendel studied exactly 7 traits
- Each of those traits is on a different chromosome
- Therefore, Mendel never had to explain the patterns you get with linked genes.
- What are the chances of picking seven traits, one each from seven chromosomes?