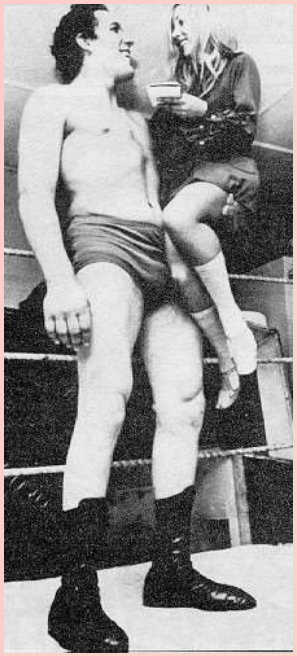
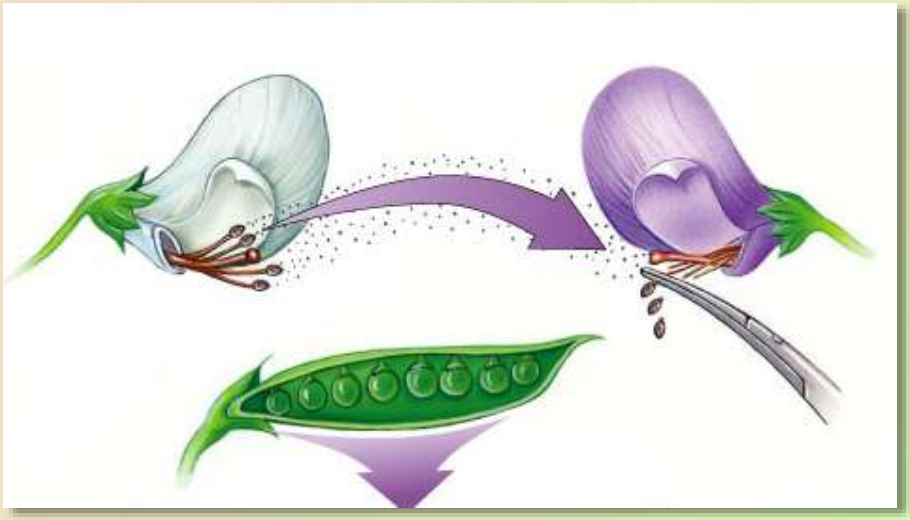
Beyond Mendel's Laws of Inheritance



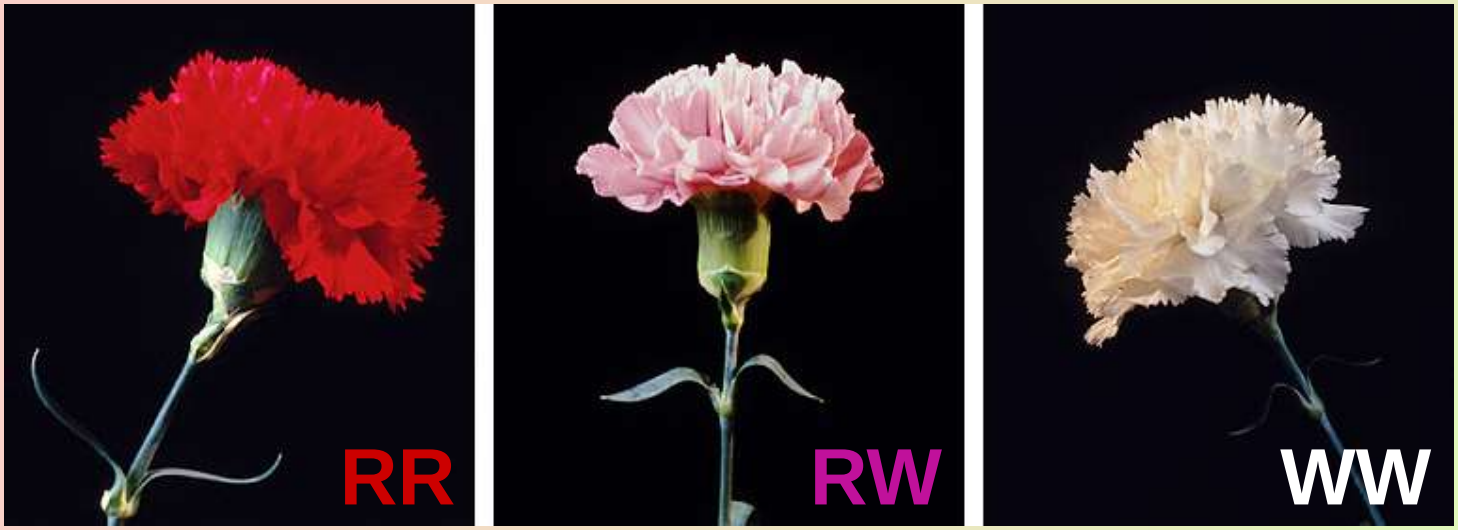
Extending Mendelian genetics

- Mendel worked with a simple system
  - peas are genetically simple
  - most traits are controlled by a single gene
  - each gene has only 2 alleles, 1 of which is completely dominant to the other
- The relationship between genotype & phenotype is rarely that simple



Incomplete dominance

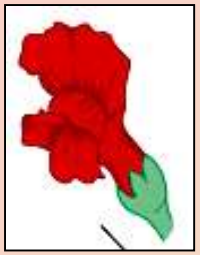
- Heterozygote shows an intermediate, blended phenotype
  - example:
    - RR = red flowers →RR
    - rr = white flowers →WW
    - Rr = pink flowers →RW
      - make 50% less color



Incomplete dominance

P

true-breeding red flowers

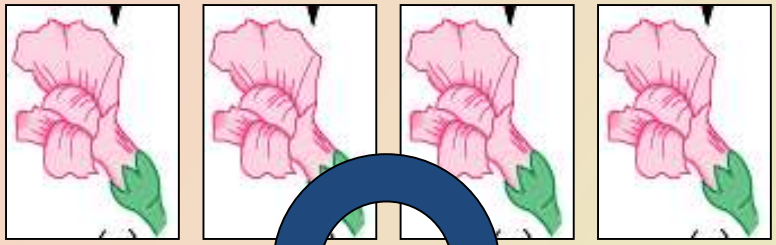


X

true-breeding white flowers

100% pink flowers

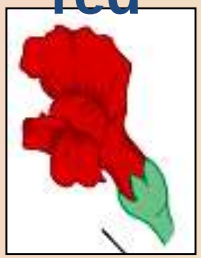
F_1 generation (hybrids)



100%

self-pollinate

25% red



50% pink

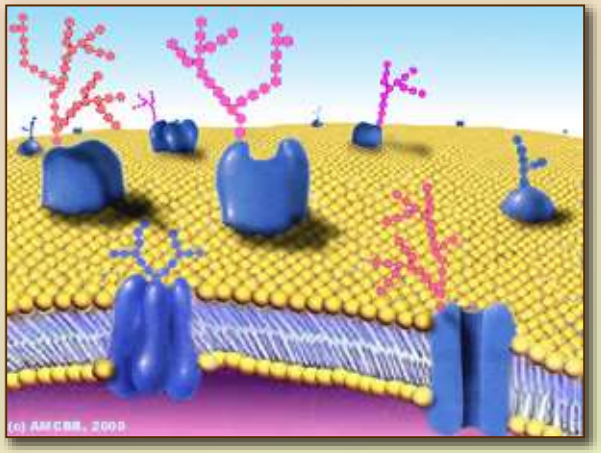
25% white

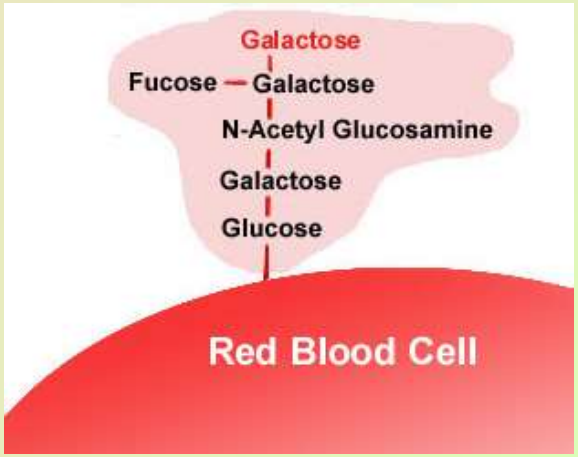
F_2 generation

1:2:1

Co-dominance

- 2 alleles affect the phenotype equally & separately
  - **not blende**d phenotype
  - human ABO blood groups
  - 3 alleles
    - I^A, I^B, *i*
    - I^A & I^B alleles are co-dominant
      - glycoprotein antigens on RBC
      - I^AI^B = both antigens are produced
    - *i* allele recessive to both





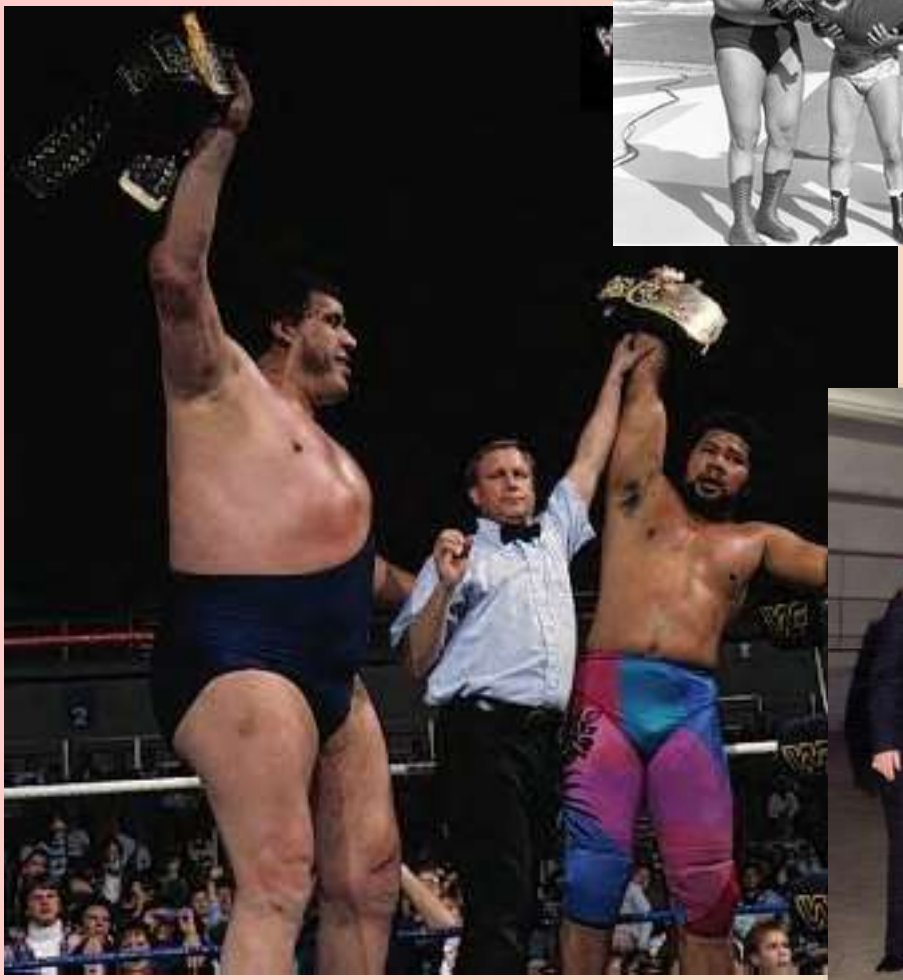
Genetics of Blood type

pheno-type	genotype	antigen on RBC	antibodies in blood	donation status
A	$I^A I^A$ or $I^A i$	type A antigens on surface of RBC	anti-B antibodies	___
B	$I^B I^B$ or $I^B i$	type B antigens on surface of RBC	anti-A antibodies	___
AB	$I^A I^B$	both type A & type B antigens on surface of RBC	no antibodies	**universal recipient**
O	$i\ i$	no antigens on surface of RBC	anti-A & anti-B antibodies	**universal donor**

Pleiotropy

- Most genes are pleiotropic
  - one gene affects more than one phenotypic character
    - 1 gene affects more than 1 trait
    - dwarfism (achondroplasia)
    - gigantism (acromegaly)

Acromegaly: André the Giant



Inheritance pattern of Achondroplasia

Aa x aa

dominant inheritance

	a	a
A	**Aa** dwarf	**Aa** dwarf
a	**aa**	**aa**

50% dwarf:50% normal or 1:1

Aa x Aa

	A	a
A	**AA** lethal	**Aa**
a	**Aa**	**aa**

67% dwarf:33% normal or 2:1

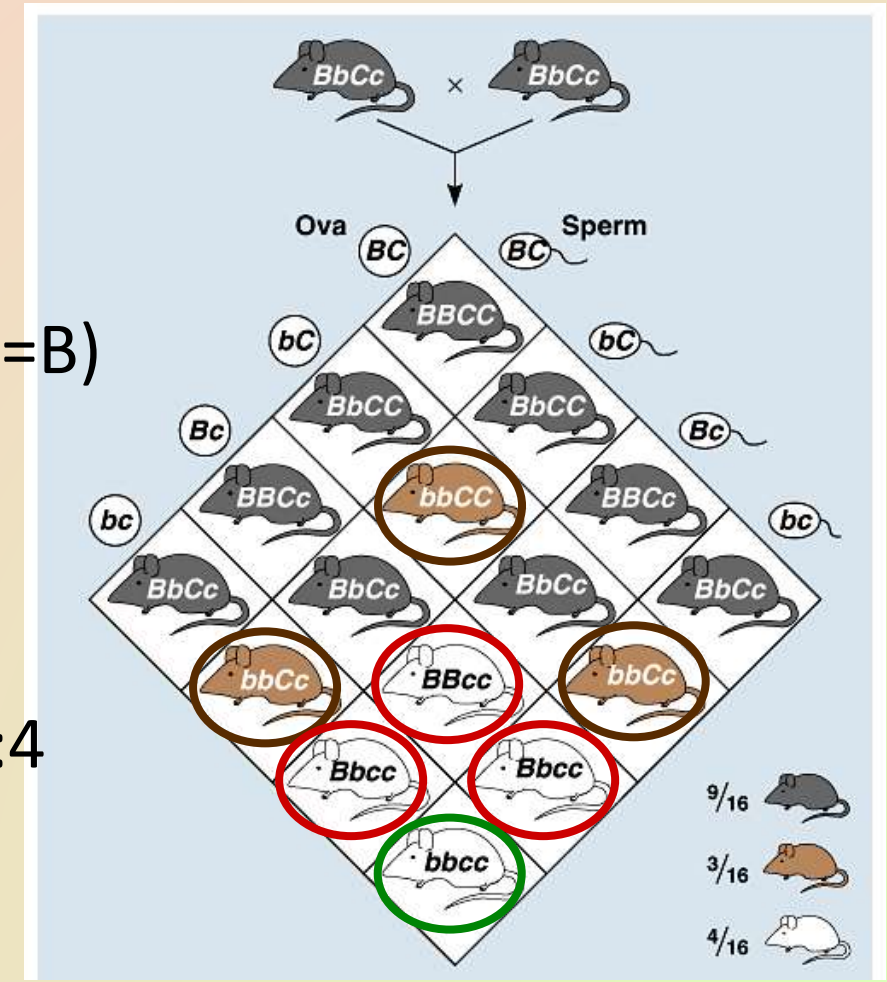
Epistasis

- One *gene* completely masks another *gene*
  - coat color in mice = 2 separate genes
    - C,c:
      pigment (C) or
      no pigment (*c*)
    - B,b:
      more pigment (black=B)
      or less (brown=b)
    - *cc* = albino,
      no matter B allele
    - 9:3:3:1 becomes 9:3:4

B_C_

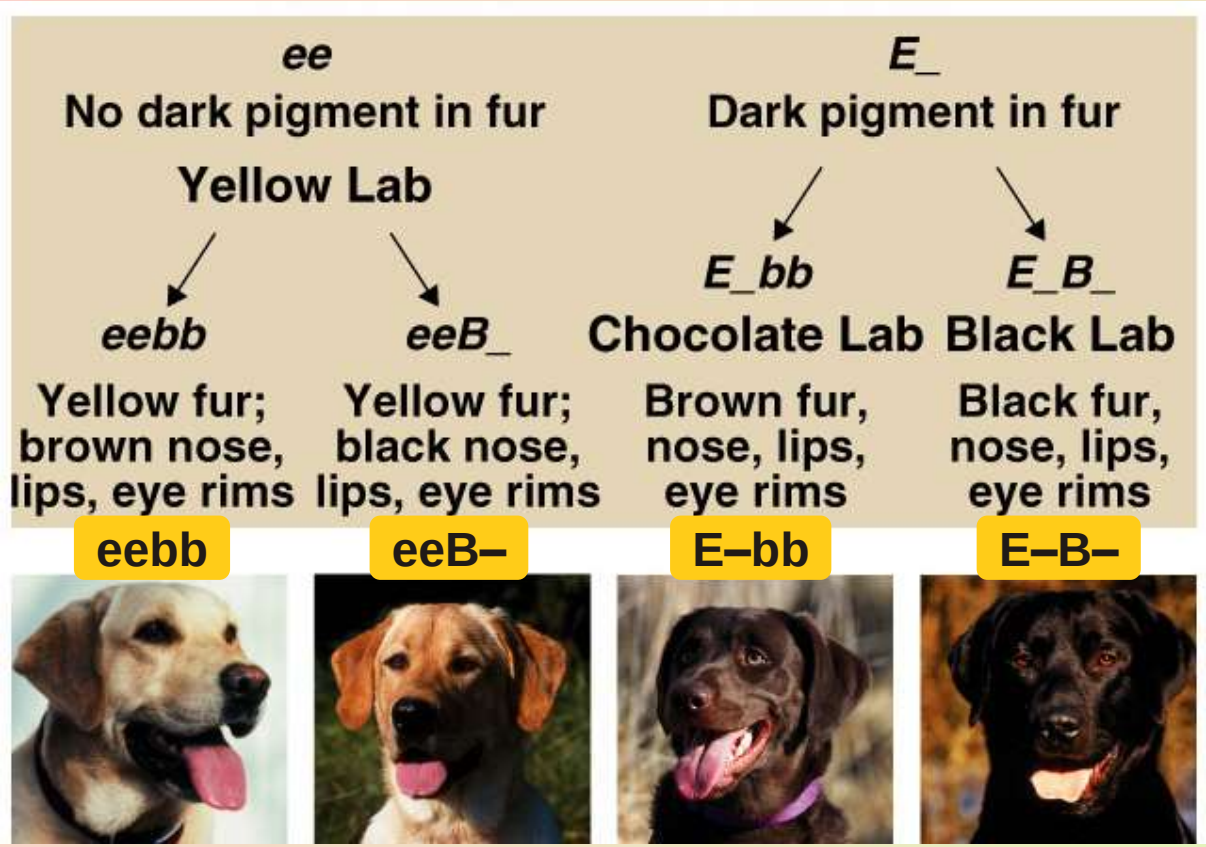
bbC_

_ _cc



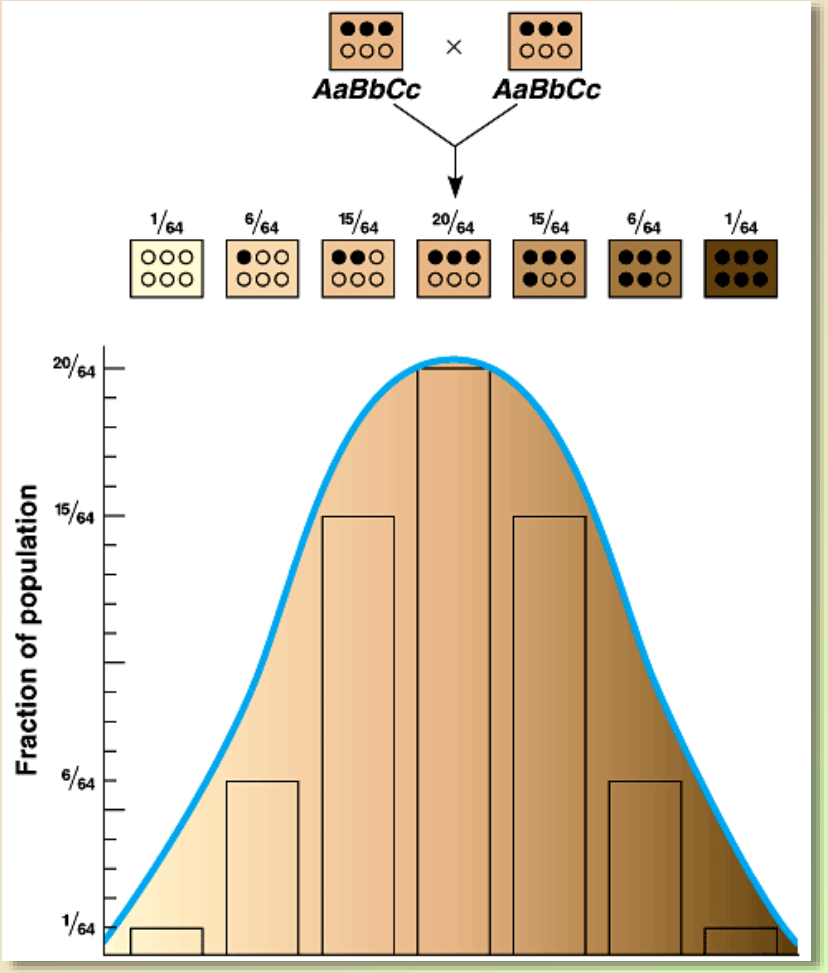
Epistasis in Labrador retrievers

- 2 genes: (E,e) & (B,b)
  - pigment (E) or no pigment (e)
  - pigment concentration: black (B) to brown (b)



Polygenic inheritance

- Some phenotypes determined by additive effects of 2 or more genes on a single character
  - phenotypes on a continuum
  - human traits
    - skin color
    - height
    - weight
    - intelligence
    - behaviors



Skin color: Albinism

- However albinism can be inherited as a single gene trait
  - aa = albino

albino Africans

melanin = universal brown color

Johnny & Edgar Winter

OCA1 albino

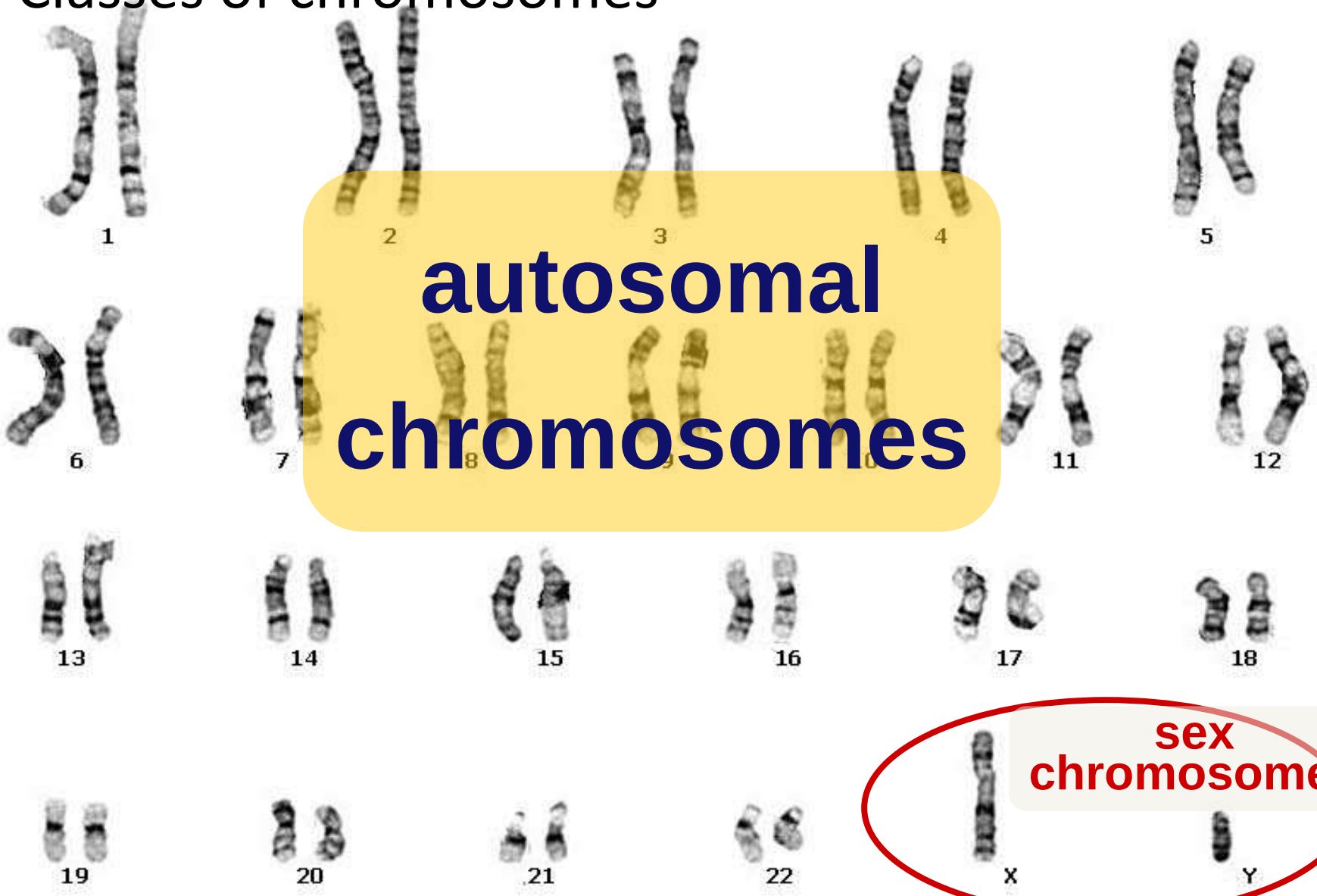
Bianca Knowlton

Sex linked traits

1910 | 1933

- Genes are on sex chromosomes
  - Discovered by T.H. Morgan at Columbia U.
  - *Drosophila* breeding
    - good genetic subject
      - prolific breeders/2 week generations
      - 4 pairs of chromosomes
      - XX=female, XY=male

Classes of chromosomes



autosomal chromosomes

sex chromosomes

Discovery of sex linkage

P

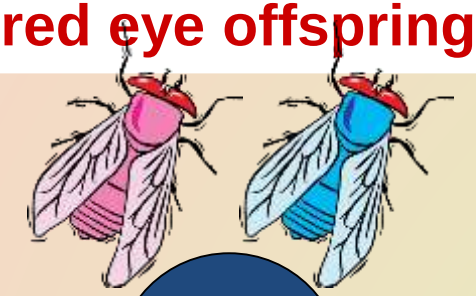
true-breeding red-eye female

X

true-breeding white-eye male

F_1 generation (hybrids)

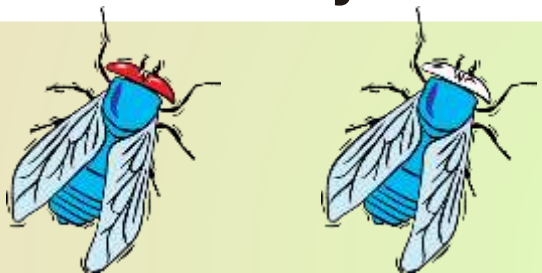
100% red eye offspring



F_2 generation

100% red-eye female

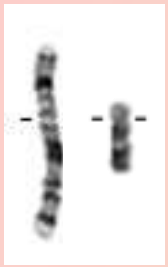
50% red-eye male
50% white eye male



Genetics of Sex

- In humans & other mammals, there are 2 sex chromosomes: X & Y
  - 2 X chromosomes
    - develop as a female: XX
    - gene redundancy, like autosomal chromosomes

  - an X & Y chromosome
    - develop as a male: XY
    - no redundancy



	X	Y
X	XX	XY
X	XX	XY

50% female : 50% male

Let's reconsider Morgan's flies…

x

x

	X^r	Y
X^R	X^RX^r	X^RY
X^R	X^RX^r	X^RY

100% red eyes

	X^R	Y
X^R	X^RX^R	X^RY
X^r	X^RX^r	X^rY

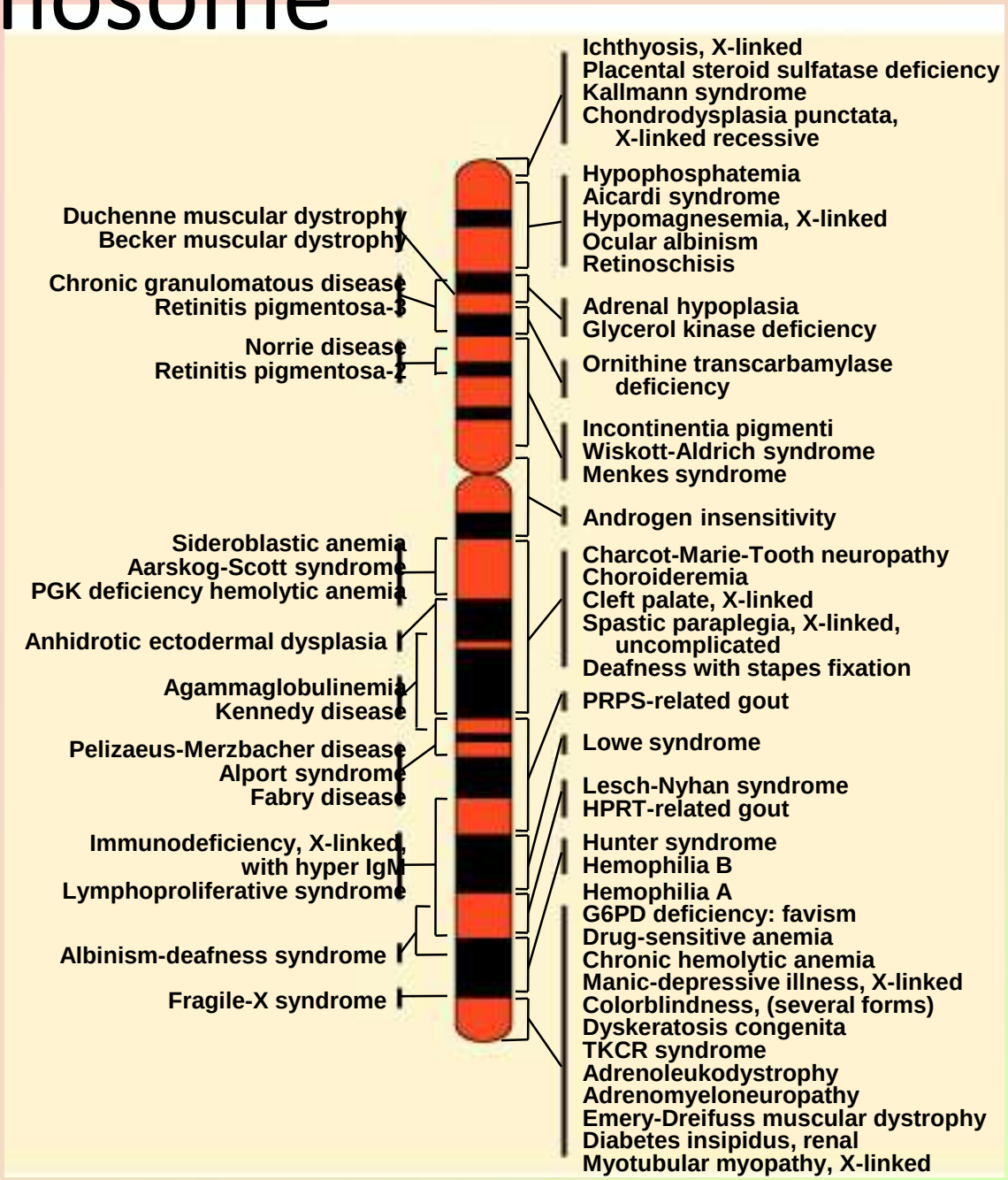
100% red females
50% red males; 50% white males

Genes on sex chromosomes

- Y chromosome
  - few genes other than SRY
    - sex-determining region: master regulator for maleness
    - turns on genes for production of male hormones
      - many effects = pleiotropy*!*
- X chromosome
  - other genes/traits beyond sex determination
    - mutations:
      - Hemophilia, Duchenne muscular dystrophy, color-blindness

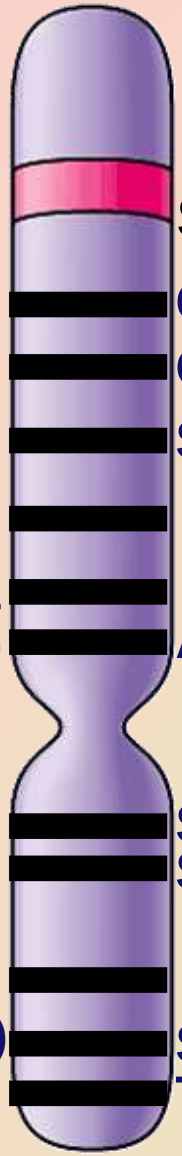
Human X chromosome

- Sex-linked
  - usually means "X-linked"
  - more than 60 diseases traced to genes on X chromosome



Map of Human Y chromosome?

< 30 genes on Y chromosome



Sex-determining Region Y (SRY)

Channel Flipping (FLP)

Catching & Throwing (BLZ-1)

Self confidence (BLZ-2)

note: not linked to ability gene

Devotion to sports (BUD-E)

Addiction to death & destruction movies (SAW-2)

Air guitar (RIF)

Scratching (ITCH-E)

Spitting (P2E)

linked

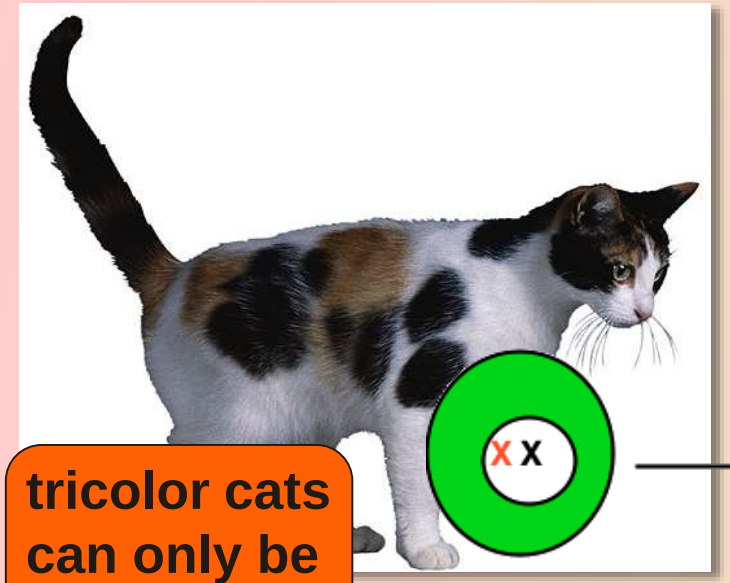
Inability to express affection over phone (ME-2)

Selective hearing loss (HUH)

Total lack of recall for dates (OOPS)

X-inactivation

- Female mammals inherit 2 X chromosomes
  - one X becomes inactivated during embryonic development
    - condenses into compact object = Barr body
    - which X becomes Barr body is random in each cell
      - patchwork trait = "mosaic"



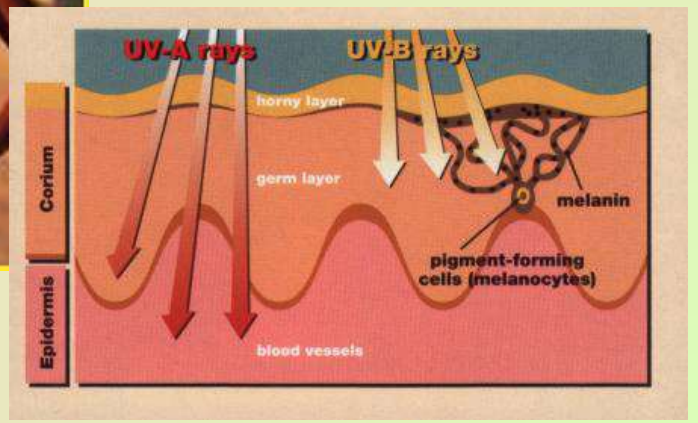
Environmental effects

- Phenotype is controlled by both environment & genes

Human skin color is influenced by both genetics & environmental conditions

Coat color in arctic fox influenced by heat sensitive alleles

Color of Hydrangea flowers is influenced by soil pH



Any Questions?

Review Questions

1. Three babies were recently mixed up in a hospital. After consideration of the data below, which of the following represent the correct baby/parent combinations?

Couple # Blood groups	I A and A	II A and B	III B and O
Baby # Blood groups	1 B	2 O	3 AB

A. I-3, II-1, III-2
B. I-1, II-3, III-2
C. I-2, II-3, III-1
D. I-2, II-1, III-3
E. I-3, II-2, III-1

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2. A mother with type B blood has two children, one with type A blood and one with type O blood. Her husband has type O blood. Which of the following could you conclude from this information?
   A. The husband could not have fathered either child.
   B. The husband could have fathered both children.
   C. The husband must be the father of the child with type O blood and could be the father of the type A child.
   D. The husband could be the father of the child with type O blood, but not the type A child.
   E. Neither the mother nor the husband could be the biological parent of the type A child.

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3. Vermilion eyes is a sex-linked recessive characteristic in fruit flies. If a female having vermilion eyes is crossed with a wild-type male, what percentage of the F_1 males will have vermilion eyes?
   A. 0%
   B. 25%
   C. 50%
   D. 75%
   E. 100%

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4. A recessive allele on the X chromosome is responsible for red-green color blindness in humans. A woman with normal vision whose father is color-blind marries a color-blind male. What is the probability that this couple's son will be color-blind?
   A. 0
   B. 1/4
   C. 1/2
   D. 3/4
   E. 1

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5. Red-green color blindness is a sex-linked recessive trait in humans. Two people with normal color vision have a color-blind son. What are the genotypes of the parents?

   A. *XcXc* and *XcY*
   B. *XcXc* and *XCY*
   C. *XCXC* and *XcY*
   D. *XCXC* and *XCY*
   E. *XCXc* and *XCY*

5. Red-green color blindness is a sex-linked recessive trait in humans. Two people with normal color vision have a color-blind son. What are the genotypes of the parents?

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D. *XCXC* and *XCY*
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6. A color-blind son inherited this trait from his
   A. mother.
   B. father.
   C. mother only if she is color-blind.
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7. In cattle, roan coat color (mixed red and white hairs) occurs in the heterozygous (*Rr*) offspring of red (*RR*) and white (*rr*) homozygotes. When two roan cattle are crossed, the phenotypes of the progeny are found to be in the ratio of 1 red:2 roan:1 white. Which of the following crosses could produce the highest percentage of roan cattle? *

   A. red × white
   B. roan × roan
   C. white × roan
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8. You think that two alleles for coat color in mice show incomplete dominance. What is the best and simplest cross to perform in order to support your hypothesis?
   A. a testcross of a homozygous recessive mouse with a mouse of unknown genotype
   B. a cross of F_1 mice to look for a 1:2:1 ratio in the offspring
   C. a reciprocal cross in which the sex of the mice of each coat color is reversed
   D. a cross of two true-breeding mice of different colors to look for an intermediate phenotype in the F_1
   E. a cross of F_1 mice to look for a 9:7 ratio in the offspring

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