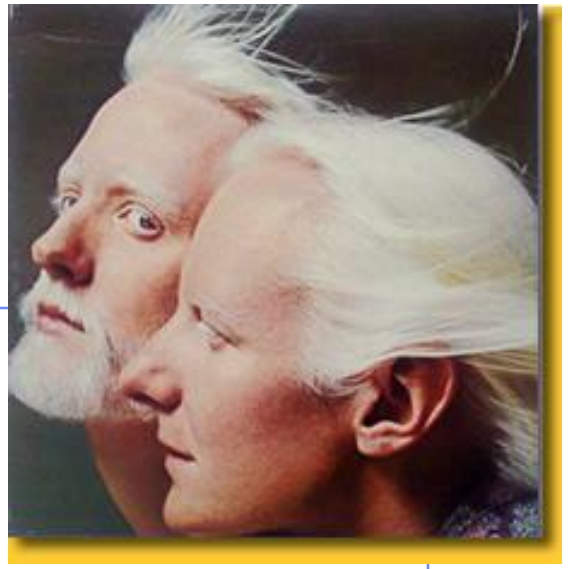
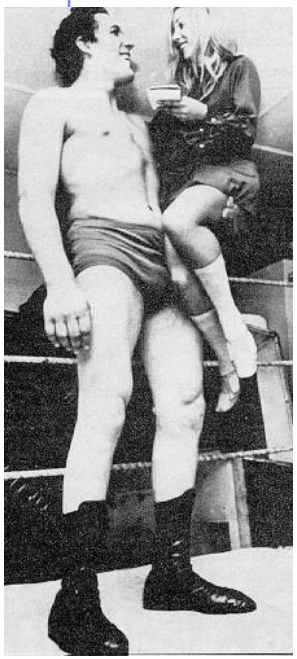
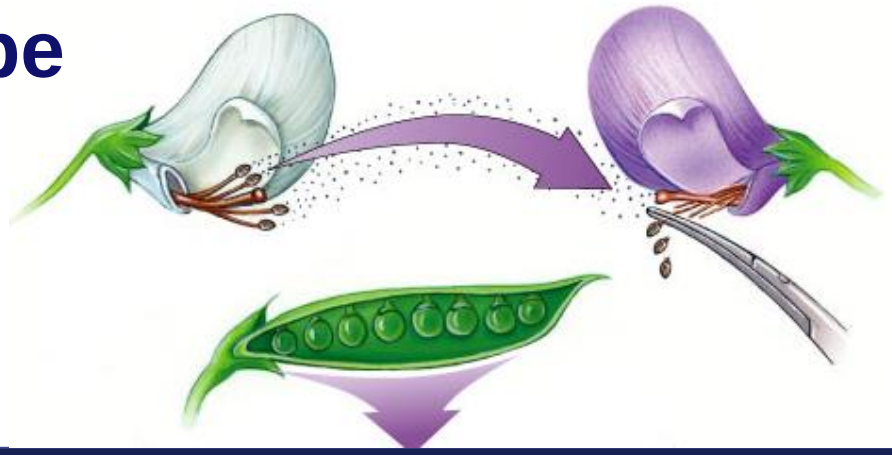


Beyond Mendel's Laws of Inheritance



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

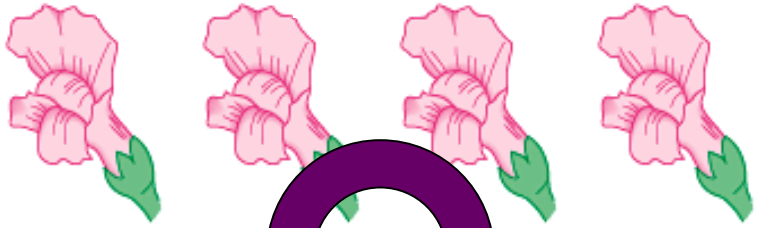
true-breeding red flowers

X

true-breeding white flowers

100% pink flowers

F_1 generation (hybrids)



self-pollinate

F_2 generation

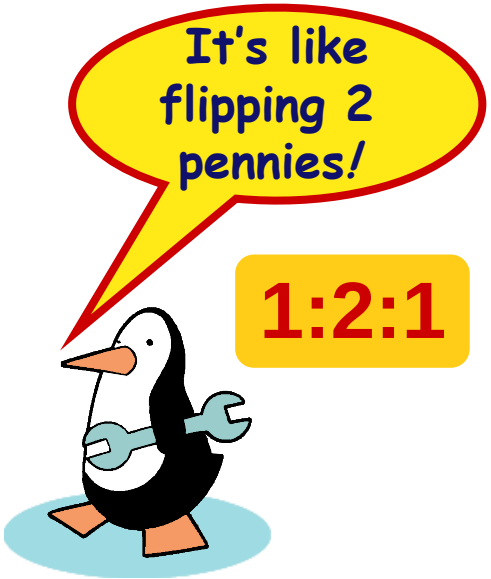
25% red

50% pink

25% white

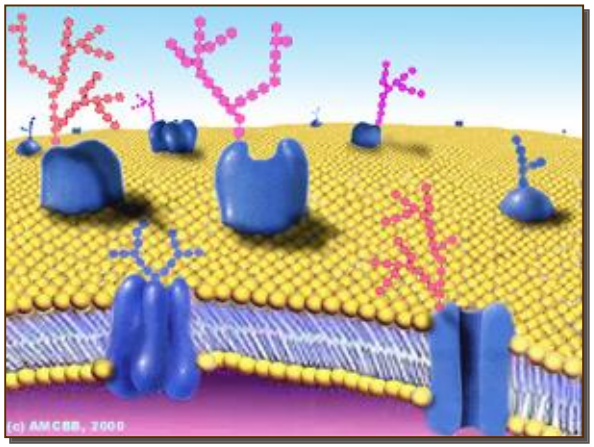
100%

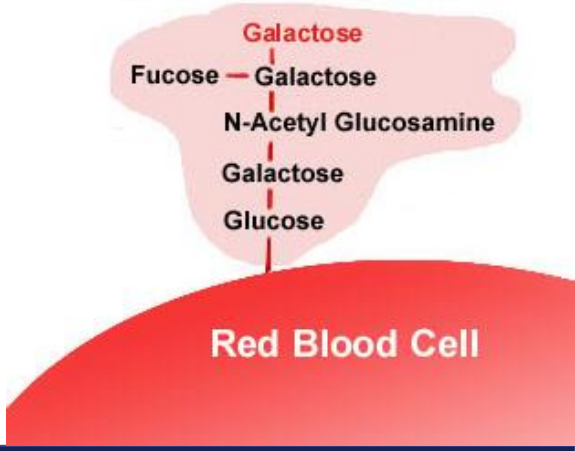
1:2:1



Co-dominance

- **2 alleles affect the phenotype equally & separately**
  - **not blended 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**





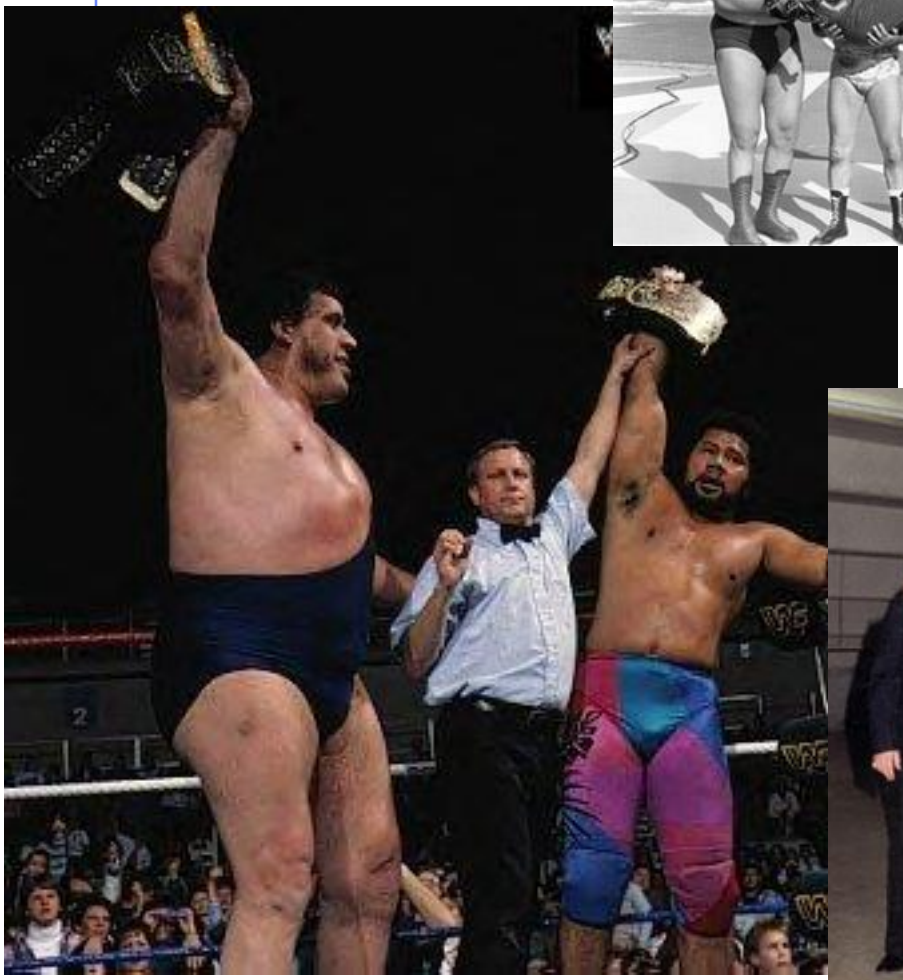
Genetics of Blood type

pheno-type	genotype	antigen on RBC	antibodies in blood	donation status
A		antigens on surface of RBC	antibodies	___
B		antigens on surface of RBC	antibodies	___
AB		antigens on surface of RBC	antibodies	
O		on surface of RBC	antibodies	

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

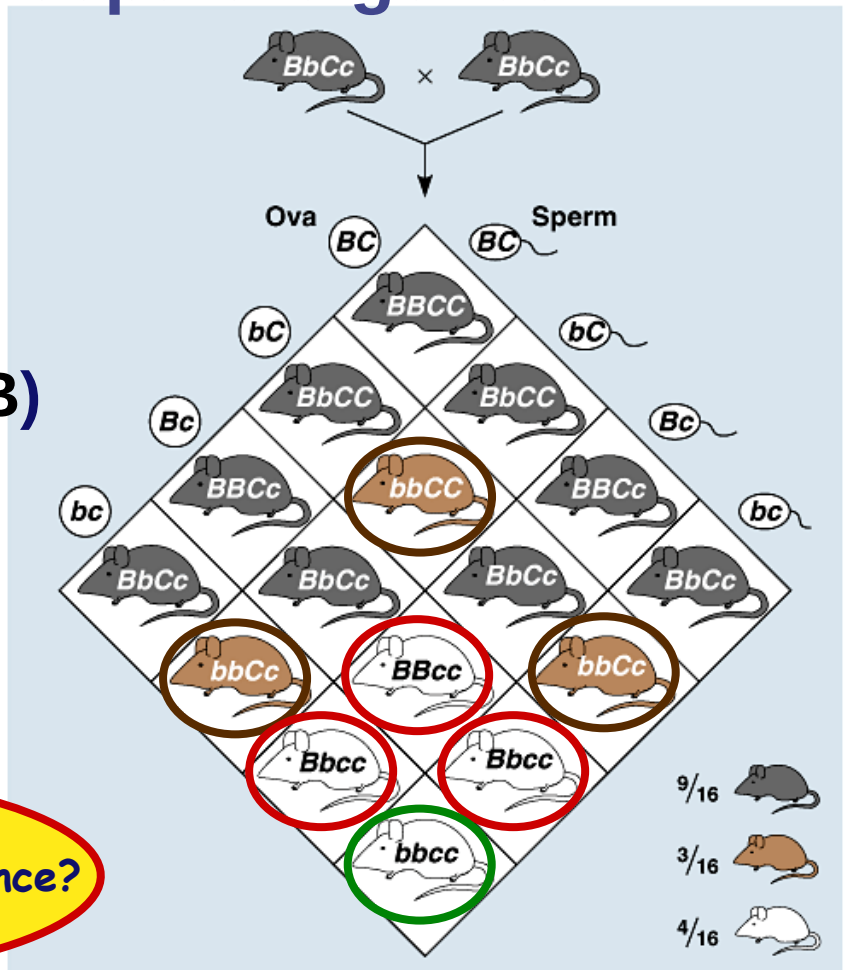
Epistasis

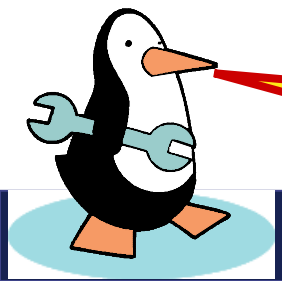
- One *gene* completely masks another *gene*
  - coat color in mice = 2 separate genes
    - C,c:
      pigment (C) or
      no pigment (*c*)
    - B,b:
      more pigment (black=B)
      or less (brown=b)
    - *cc* = albino,
      no matter B allele
    - 9:3:3:1 becomes 9:3:4

B_C_

bbC_

_ _cc

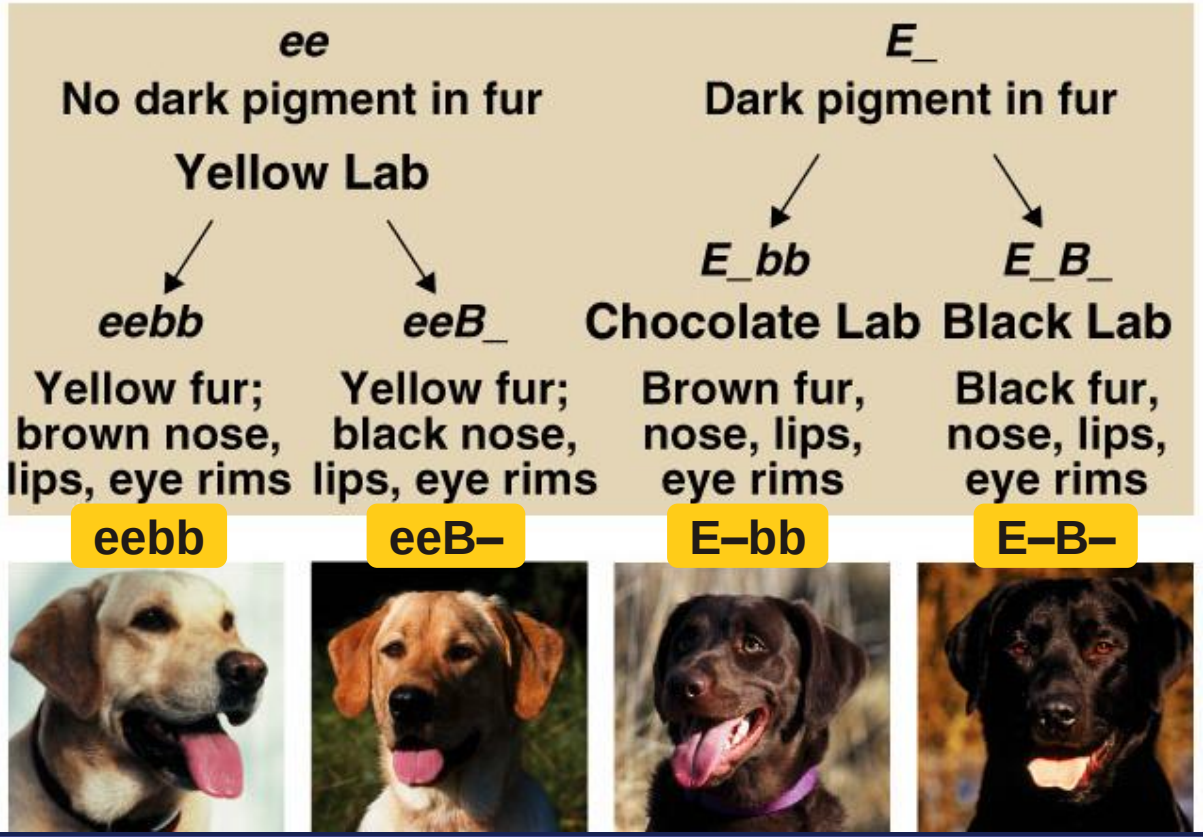




How would you know that difference wasn't random chance? Chi-square test!

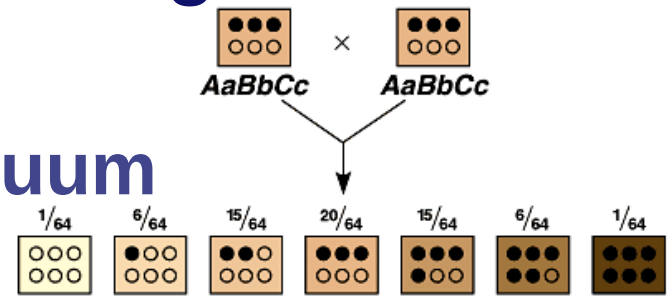
Epistasis in Labrador retrievers

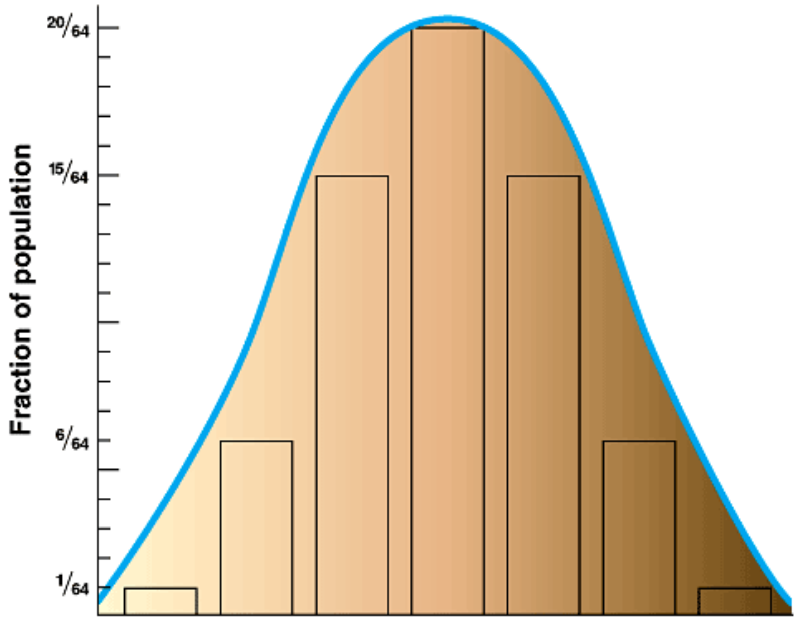
- 2 genes: (E,e) & (B,b)
  - pigment (E) or no pigment (e)
  - pigment concentration: black (B) to brown (b)



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

tyrosine → enzyme → melanin

albinism

Johnny & Edgar Winter

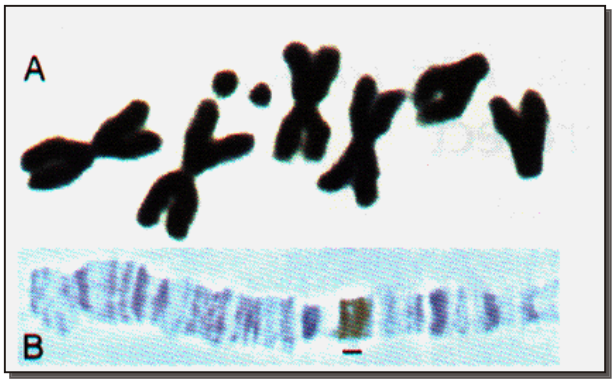
OCA1 albino

Bianca Knowlton

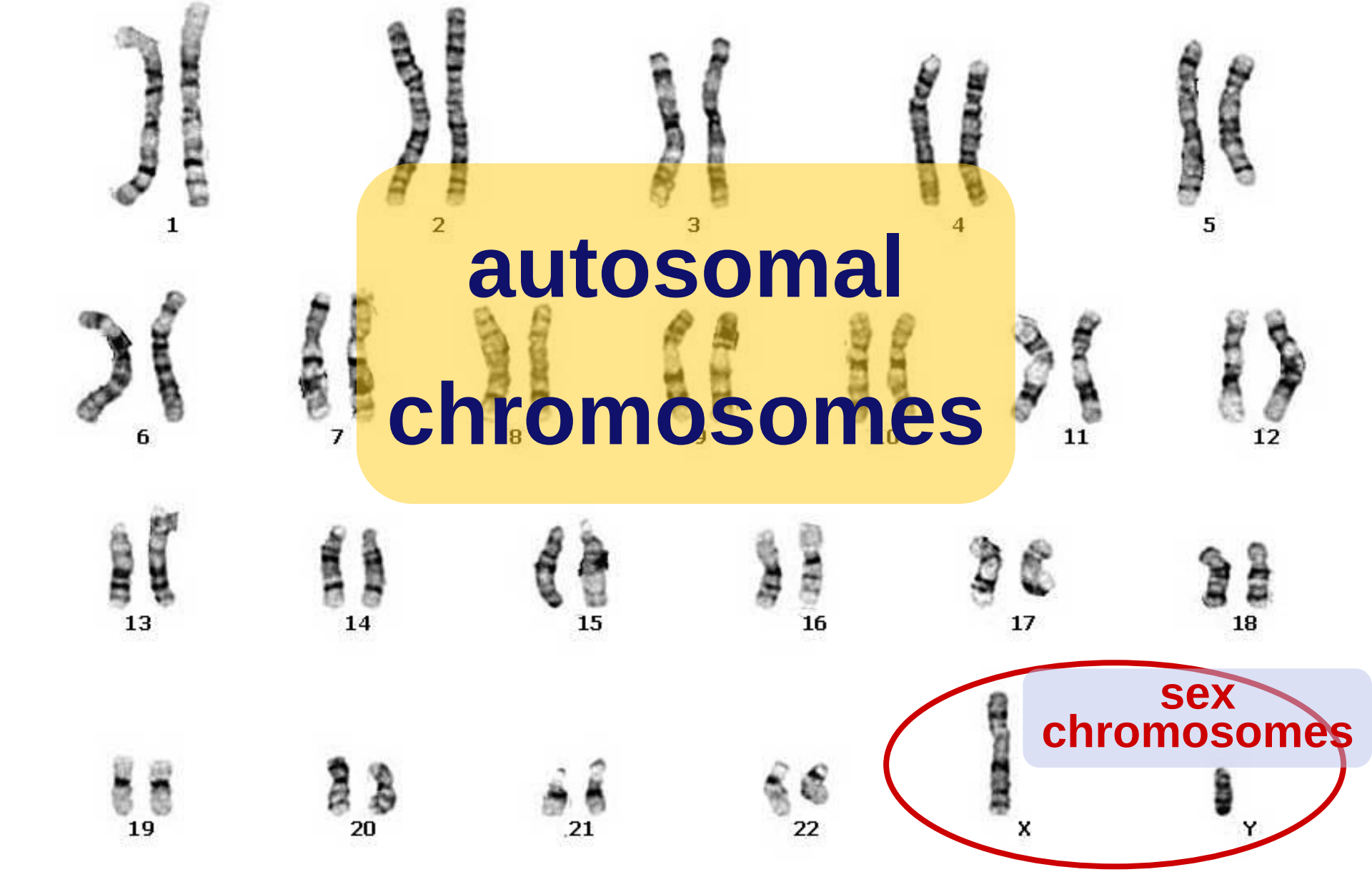
1910 | 1933

Sex linked traits

- Genes are on sex chromosomes
  - as opposed to autosomal chromosomes
  - first discovered by T.H. Morgan at Columbia U.
  - *Drosophila* breeding
    - good genetic subject
      - prolific
      - 2 week generations
      - 4 pairs of chromosomes
      - XX=female, XY=male



Classes of chromosomes



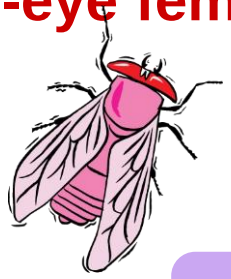
autosomal chromosomes

sex chromosomes

Discovery of sex linkage

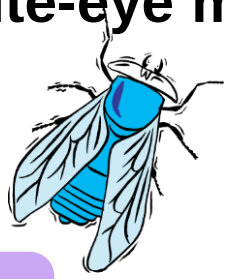
P

true-breeding red-eye female



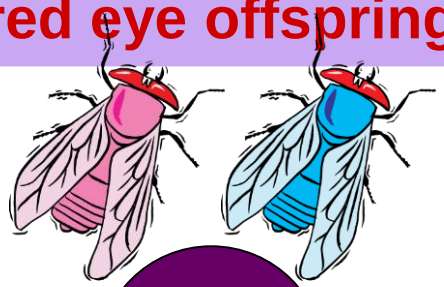
x

true-breeding white-eye male



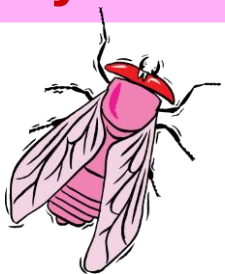
F_1 generation (hybrids)

100% red eye offspring

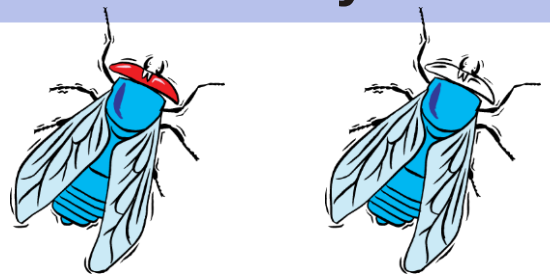


F_2 generation

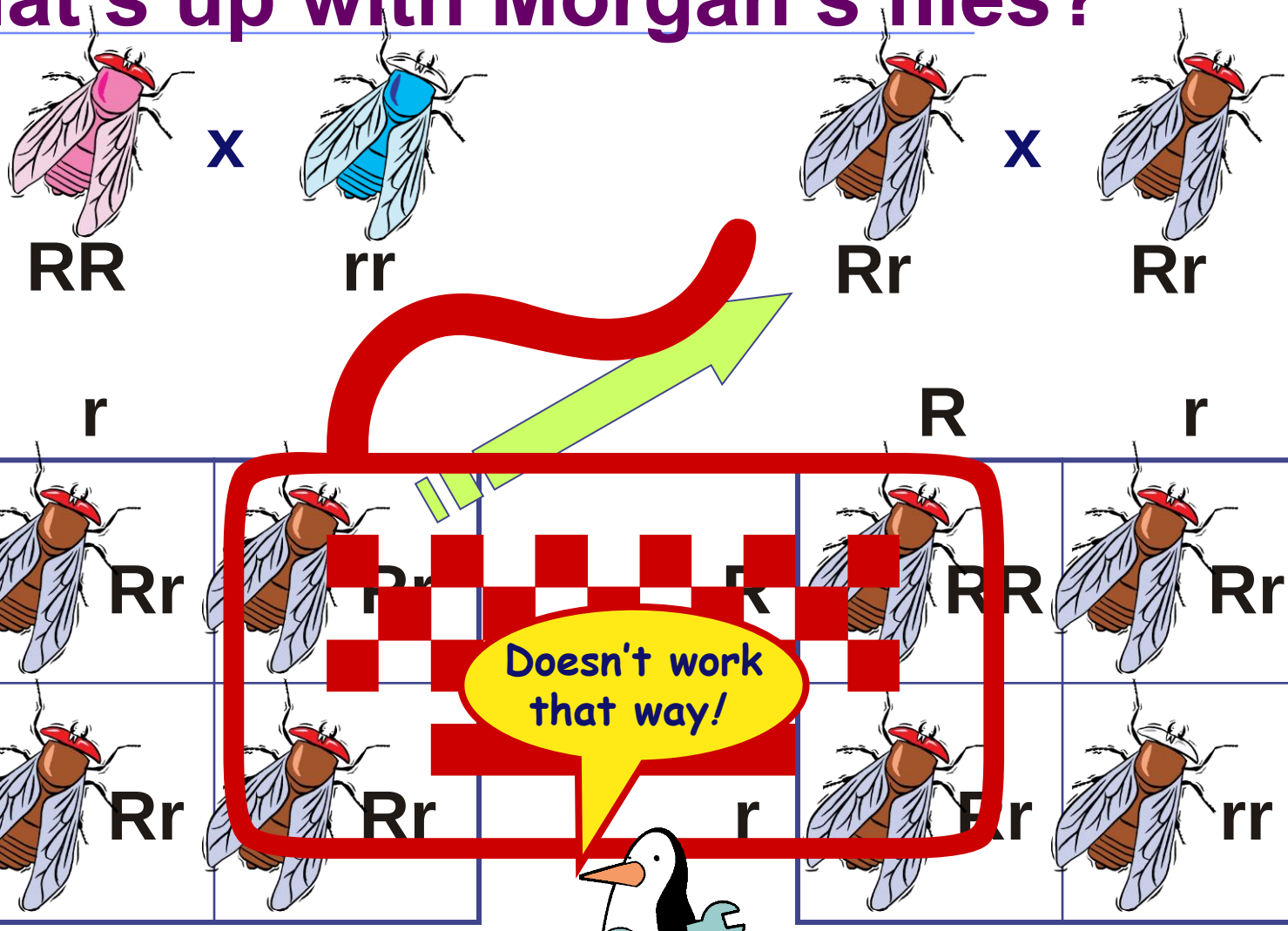
100% red-eye female



50% red-eye male
50% white eye male



What's up with Morgan's flies?



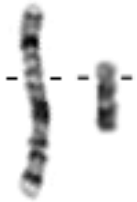
100% red eyes

3 red : 1 white

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

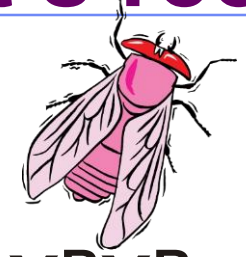


    - develop as a male: XY
    - no redundancy

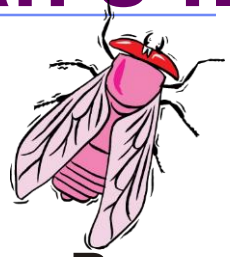
	X	Y
X	XX	XY
X	XX	XY

50% female : 50% male

Let's reconsider Morgan's flies…

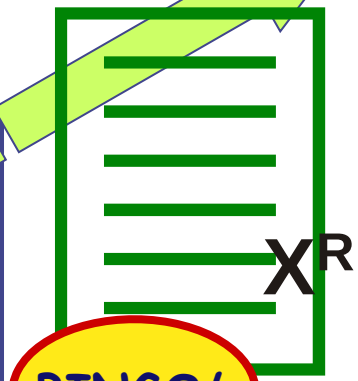


X^RX^R x X^rY

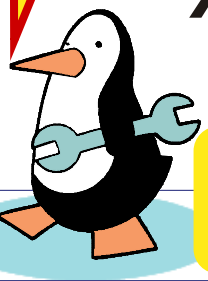


X^RX^r x X^RY

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



BINGO!



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

100% red eyes

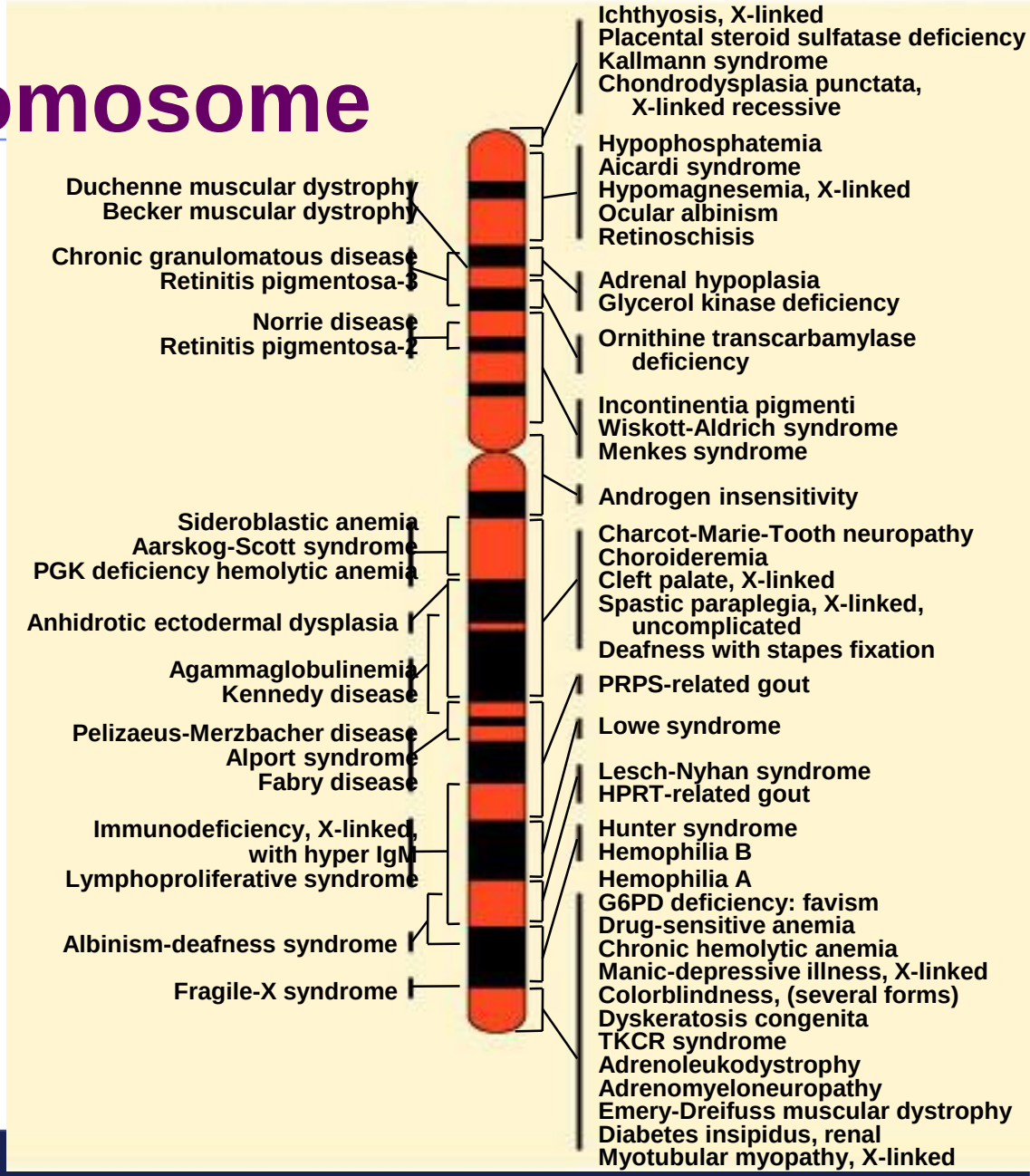
100% red females
50% red males; 50% white males

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)

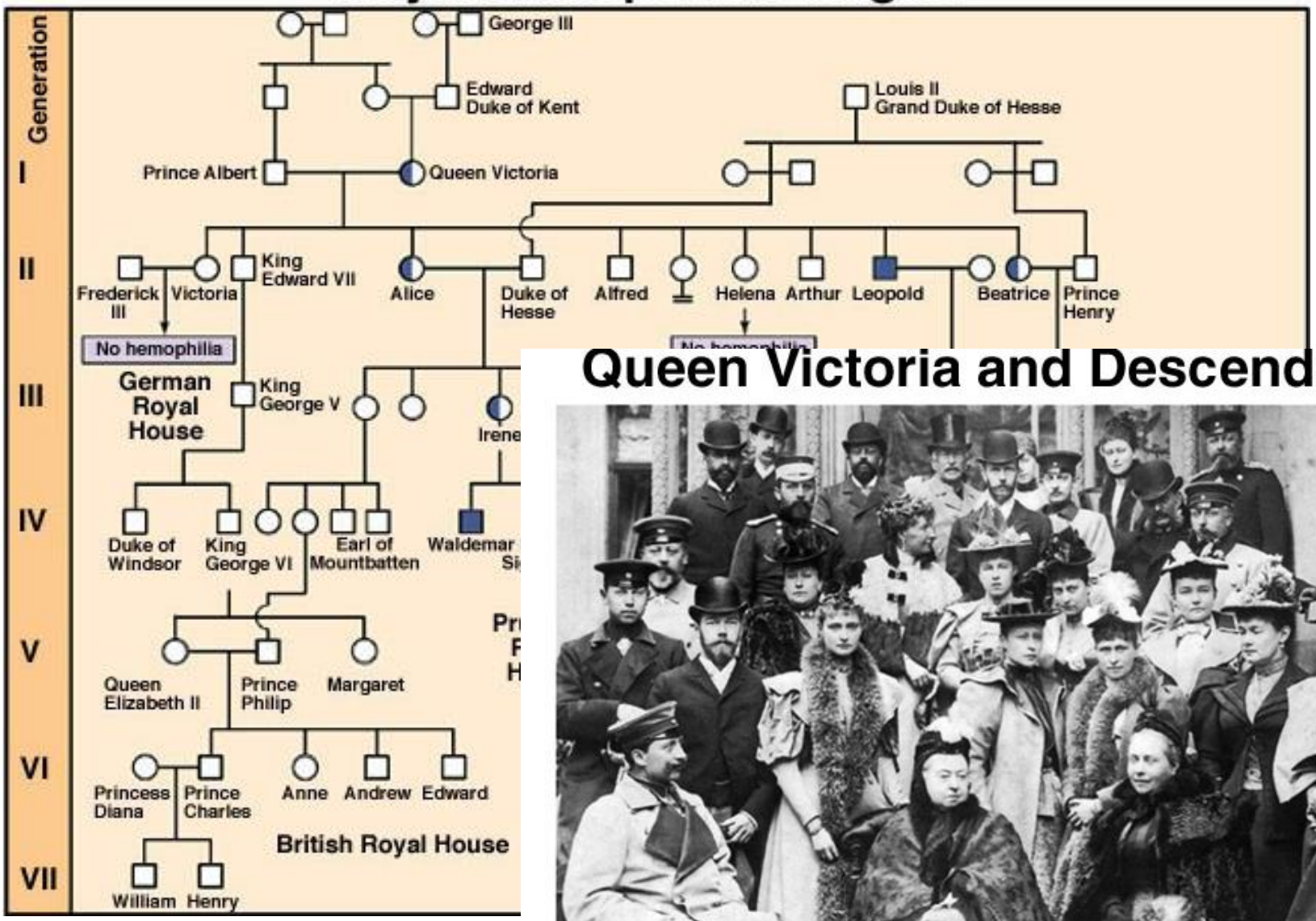
linked

Inability to express affection over phone (ME-2)

Selective hearing loss (HUH)

Total lack of recall for dates (OOPS)

Royal Hemophilia Pedigree

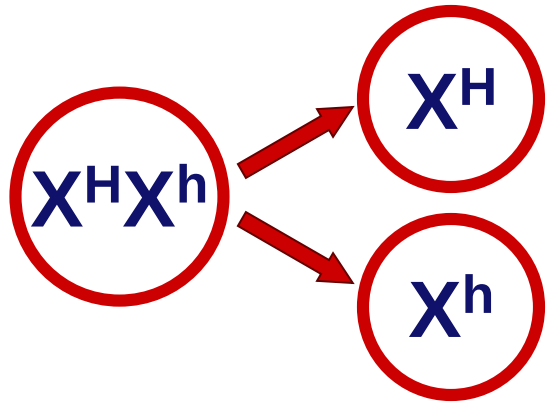


Queen Victoria and Descendants

Hemophilia

sex-linked recessive

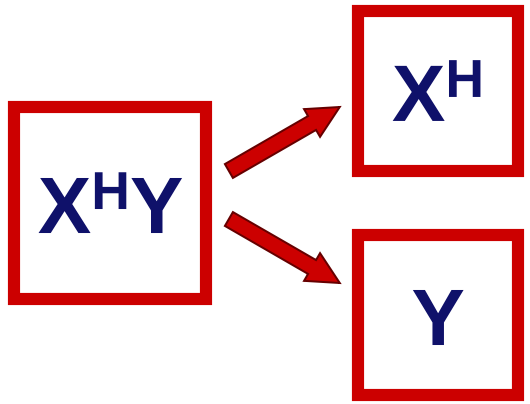
X^HX^h x X^HY



male / sperm

female / eggs	X^H	Y
X^H	X^HX^H	X^HY
X^h	X^HX^h	X^hY

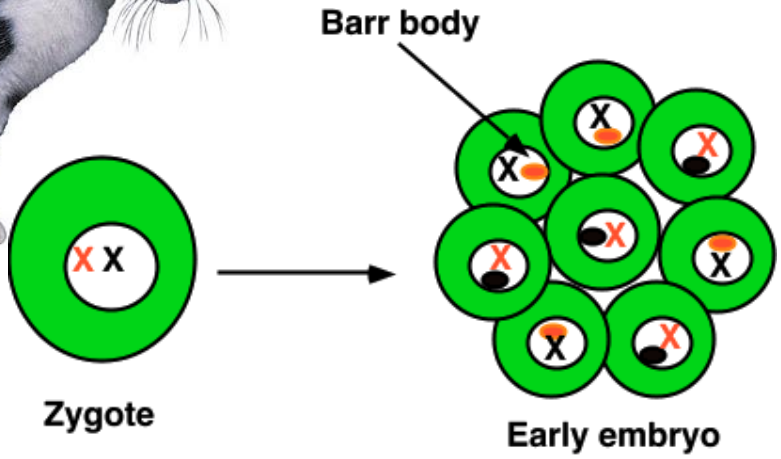
carrier disease

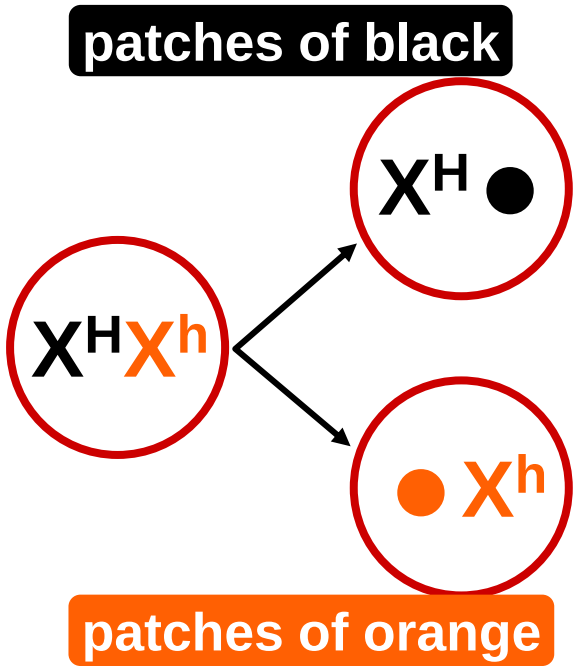


X-inactivation

- 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
      - patchwork trait = "mosaic"

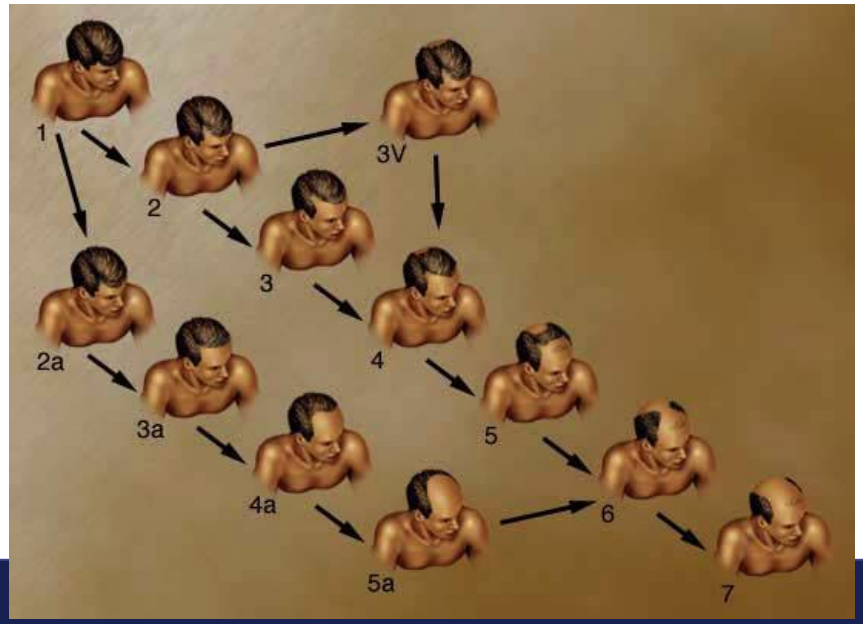
tricolor cats can only be female





Male pattern baldness

- **Sex influenced trait**
  - autosomal trait influenced by sex hormones
    - age effect as well = onset after 30 years old
  - dominant in males & recessive in females
    - B_ = bald in males; bb = bald in females



Environmental effects

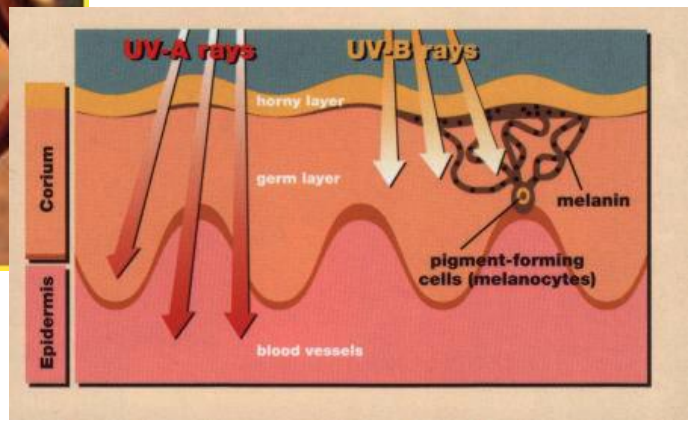
- **Phenotype is controlled by both environment & genes**

Human skin color is influenced by both genetics & environmental conditions

(a)

Coat color in arctic fox influenced by heat sensitive alleles

Color of Hydrangea flowers is influenced by soil pH



Any Questions?

Review Questions

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%

4. **Barring in chickens is due to a sex-linked dominant gene (*B*). The sex of chicks at hatching is difficult to determine, but barred chicks can be distinguished from nonbarred at that time. To use this trait so that at hatching all chicks of one sex are barred, what cross would you make?**

A. barred males × barred females
B. barred males × nonbarred females
C. nonbarred males × barred females
D. nonbarred males × nonbarred females

5. 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