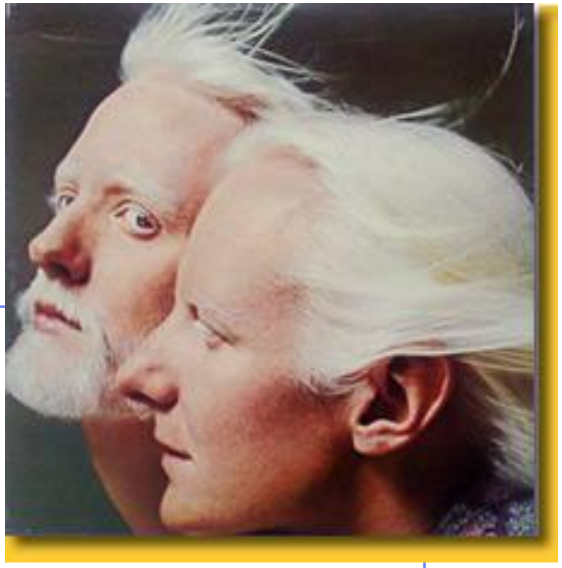
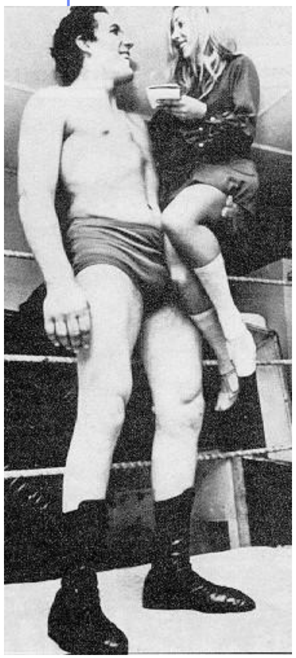


Chapter 14.

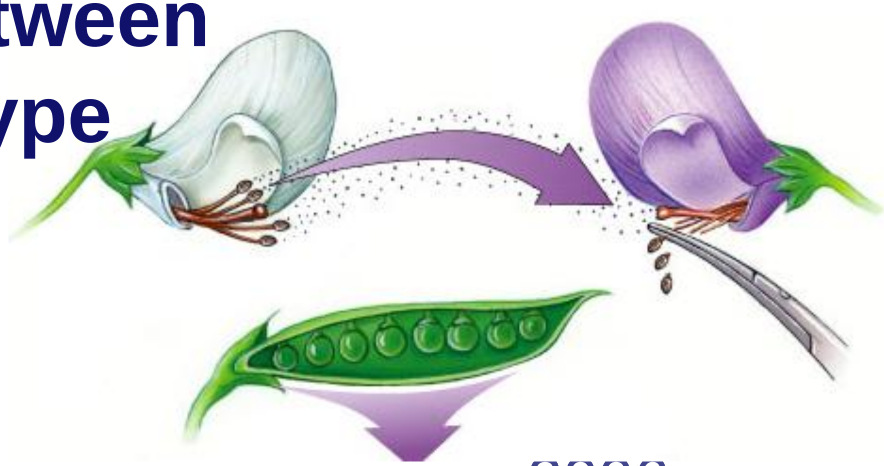
Beyond Mendel's Laws of Inheritance



Modified from Kim Foglia

Extending Mendelian genetics

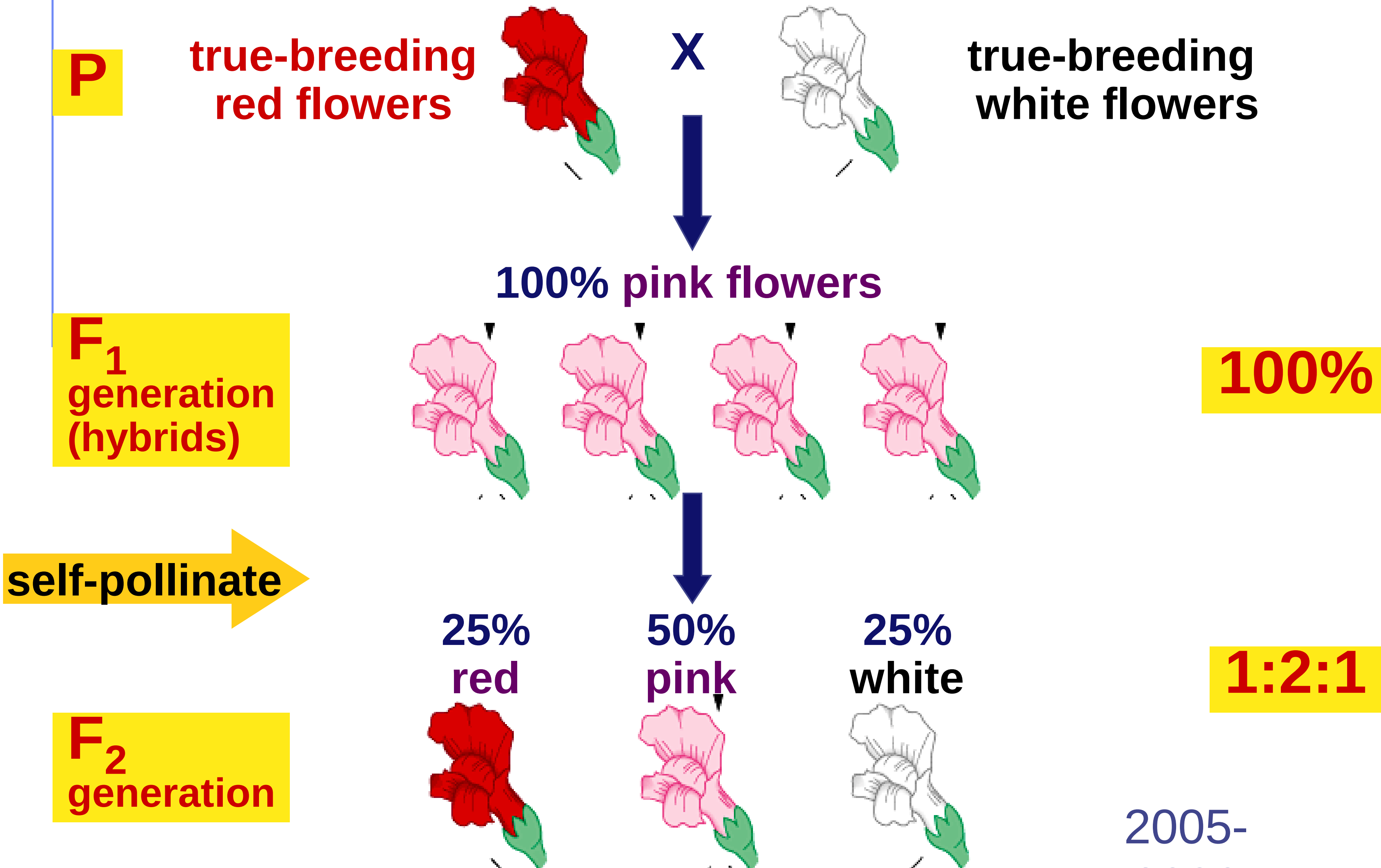
- **Mendel worked with a simple system**
  - **peas are genetically simple**
  - **most traits are controlled by a single gene**
  - **each gene has only 2 alleles, 1 of which is completely dominant to the other**
- **The relationship between genotype & phenotype is rarely that simple**



Incomplete dominance

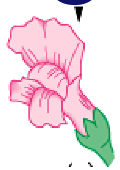
- **Heterozygotes show an intermediate phenotype**
  - **RR = red flowers**
  - **rr = white flowers**
  - **Rr = pink flowers**
    - **make 50% less color**

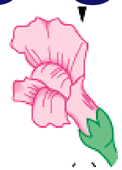
Incomplete dominance

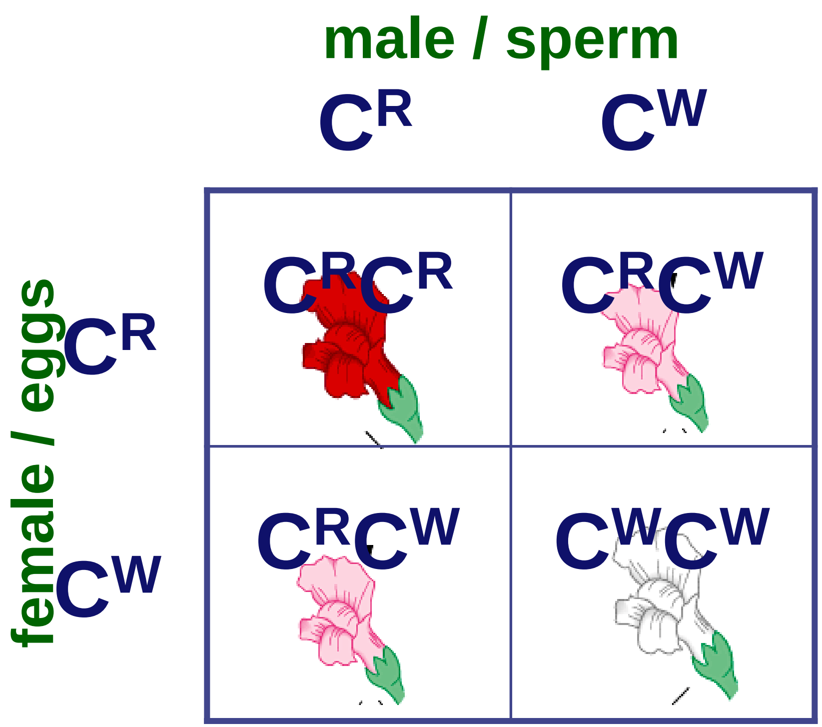


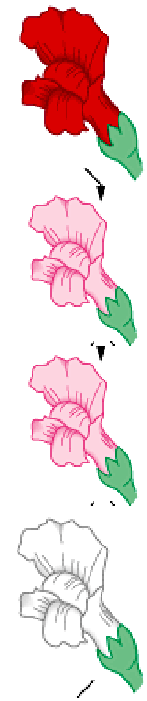
Incomplete dominance

$C^R C^W$ x $C^R C^W$







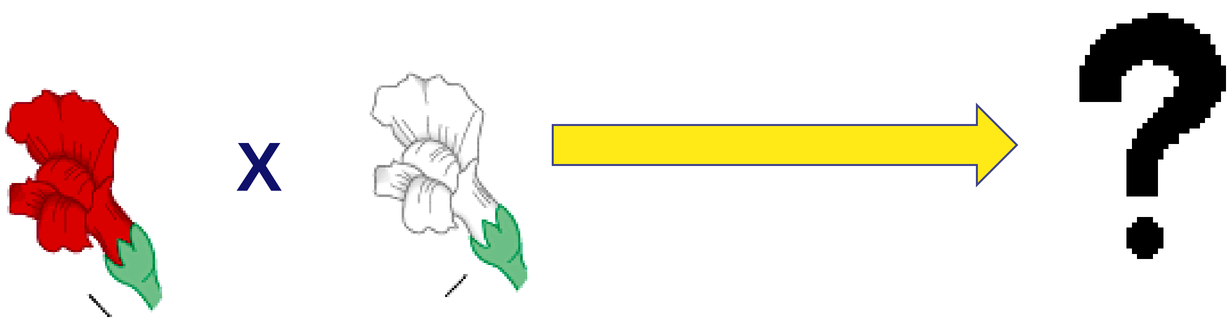


	% genotype	% phenotype
$C^R C^R$	25%	25%
$C^R C^W$	50%	50%
$C^R C^W$		
$C^W C^W$	25%	25%
	1:2:1	1:2:1

Wednesday, April 2nd

Please consider the following:

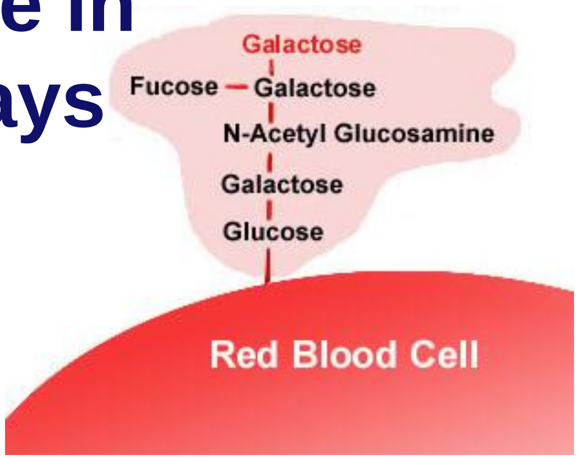
- *A carnation that is red is crossed with a carnation that is white. What are the genotypes of these flowers if they exhibit* ***incomplete dominance****?*
- *What would the phenotype(s) be of the F_1 offspring generation from this cross?*

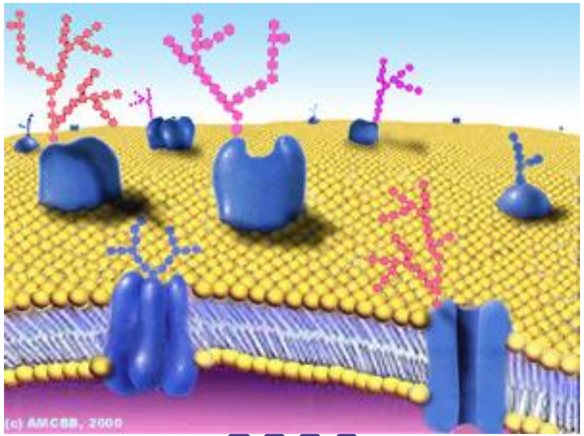


Today I will **explain** incomplete dominance.
I will **differentiate** between incomplete dominance & codominance.
I will **calculate** the probability of inheriting various blood types using known genotypes.

Co-dominance

- 2 alleles affect the phenotype in separate, distinguishable ways
  - ABO blood groups
  - 3 alleles
    - I^A, I^B, i
    - both I^A & I^B are dominant to i allele
    - I^A & I^B alleles are co-dominant to each other
  - determines presences of oligosaccharides on the surface of red blood cells





Blood type

genotype	phenotype	phenotype	status
$I^A I^A$ $I^A i$	type A	type A oligosaccharides on surface of RBC	—
$I^B I^B$ $I^B i$	type B	type B oligosaccharides on surface of RBC	—
$I^A I^B$	type AB	both type A & type B oligosaccharides on surface of RBC	universal recipient
$i\ i$	type O	no oligosaccharides on surface of RBC	universal donor

1901 | 1930

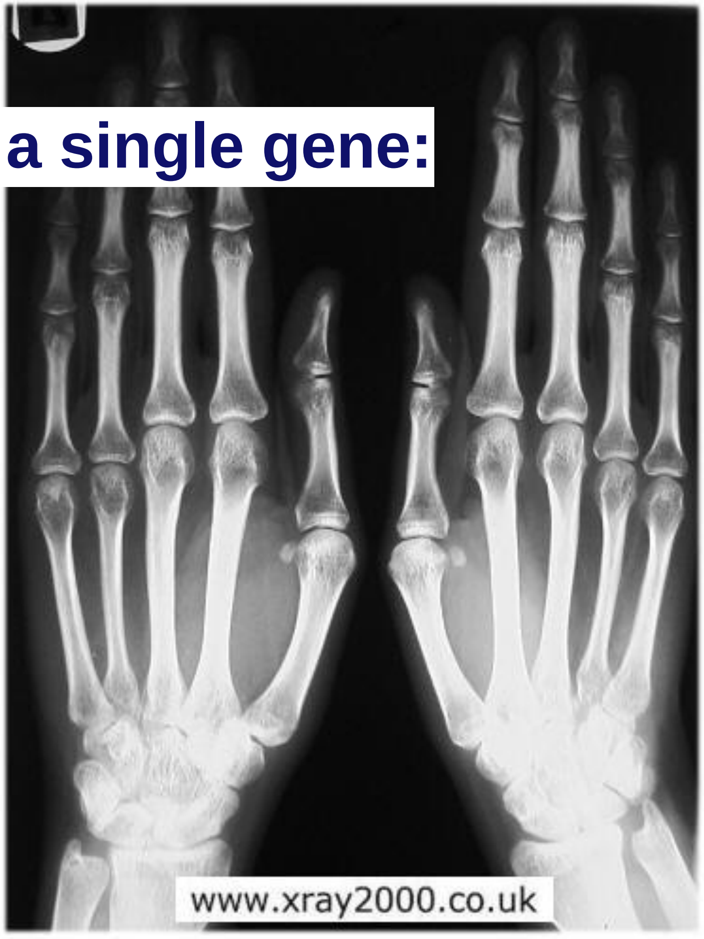
Blood compatibility

- **Matching compatible blood groups**
  - critical for blood transfusions
- **A person produces antibodies against oligosaccharides in foreign blood**
  - wrong blood type
    - donor's blood has A or B oligosaccharide that is foreign to recipient
    - antibodies in recipient's blood bind to foreign molecules
    - cause donated blood cells to clump together
    - can kill the recipient

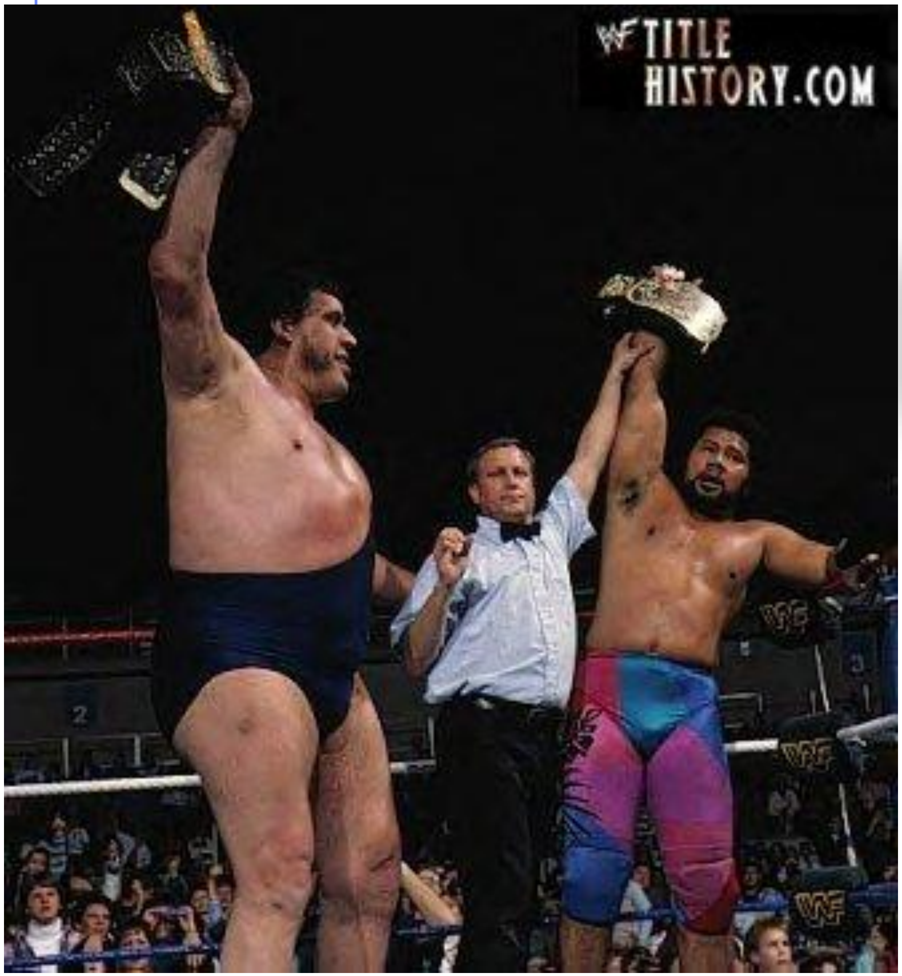
Karl Landsteiner
(1868-1943)

Pleiotropy

- **Most genes are <u>pleiotropic</u>**
  - one gene affects more than one phenotypic character
    - wide-ranging effects due to a single gene:
    - dwarfism (achondroplasia)
    - gigantism (acromegaly)



Acromegaly: André the Giant

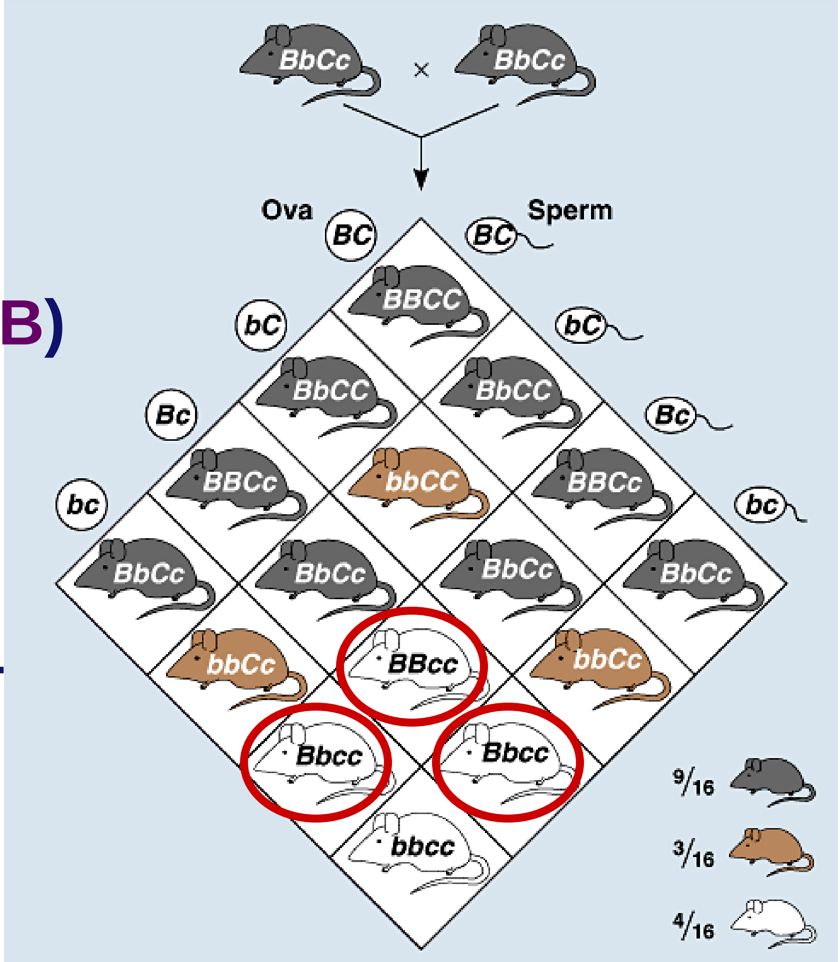


Pleiotropy

- It is not surprising that a gene can affect a number of organism's characteristics
  - consider the intricate molecular & cellular interactions responsible for an organism's development
    - cystic fibrosis
      - mucus build up in many organs
    - sickle cell anemia
      - sickling of blood cells

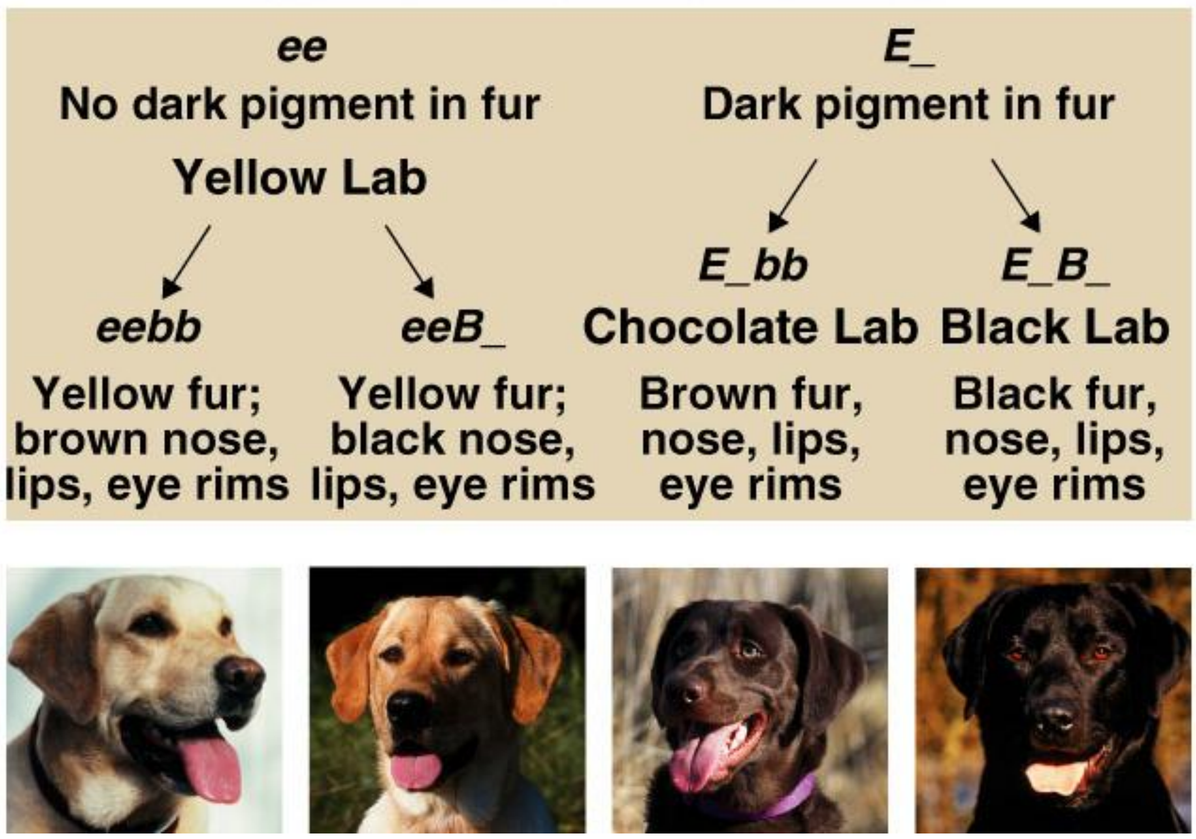
Epistasis

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



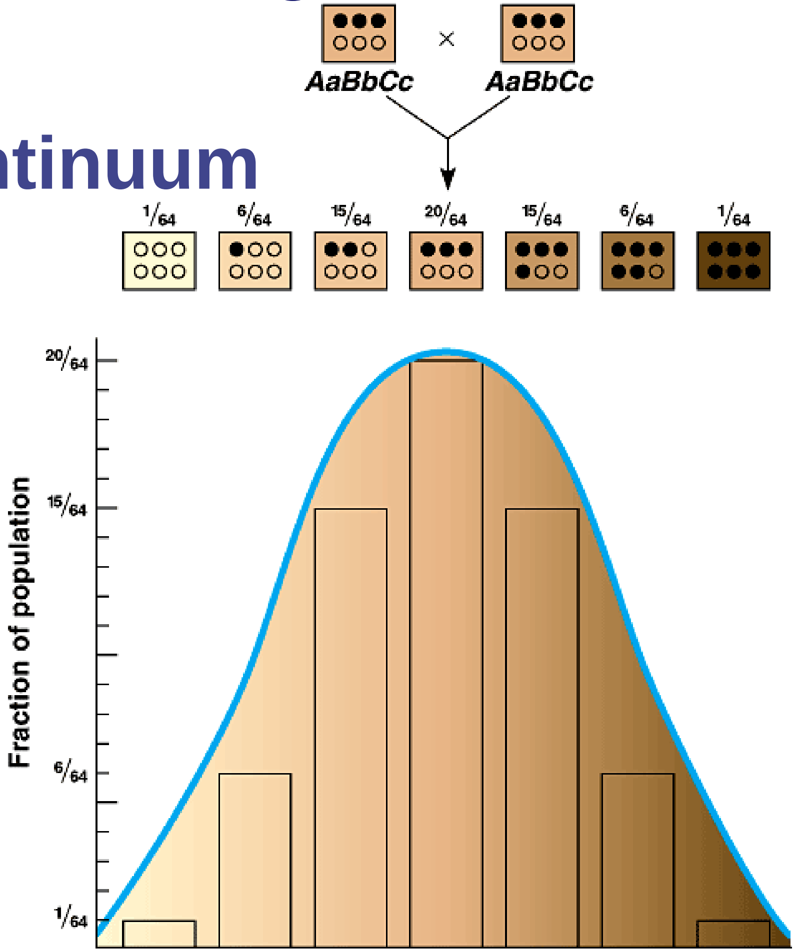
Epistasis in Labrador retrievers

- 2 genes: E & B
  - pigment (E) or no pigment (e)
  - how dark pigment will be: 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
    - eye color
    - intelligence
    - behaviors



Albinism

albino Africans

Johnny & Edgar Winter

Thursday, April 3rd

QUESTION TO PONDER

Describe *epistasis*. Provide an example from class that was discussed yesterday.

- Please take out your practice problems from Wed.
- Today we will discuss *Sex-linkage* before embarking upon HARDY-WEINBERG population genetics.

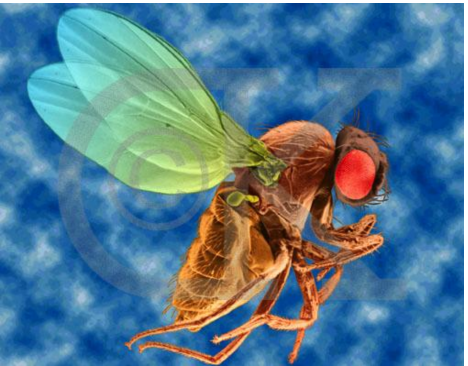
It all started with a fly…

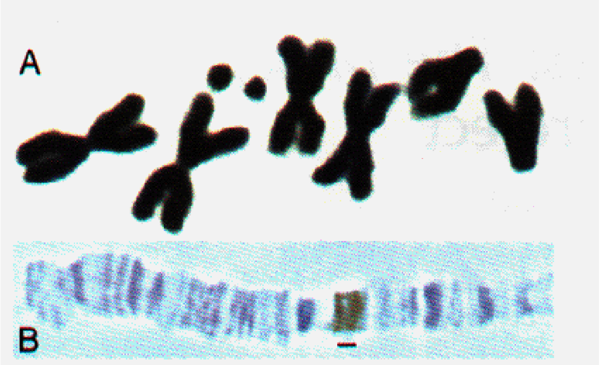
- **Chromosome theory of inheritance**
  - **experimental evidence from improved microscopy & animal breeding led us to a better understanding of chromosomes & genes beyond Mendel**
    - ***Drosophila* studies**

A. H. Sturtevant in the *Drosophila* stockroom at Columbia University

Thomas Hunt Morgan

- embryologist at Columbia University
  - 1st to associate a specific gene with a specific chromosome
  - *Drosophila* breeding
    - prolific
    - 2 week generations
    - 4 pairs of chromosomes
    - XX=female, XY=male





Morgan's first mutant…

- **Wild type fly = red eyes**
- **Morgan discovered a mutant white-eyed male**
  - **traced the gene for eye color to a specific chromosome**

Discovery of sex linkage

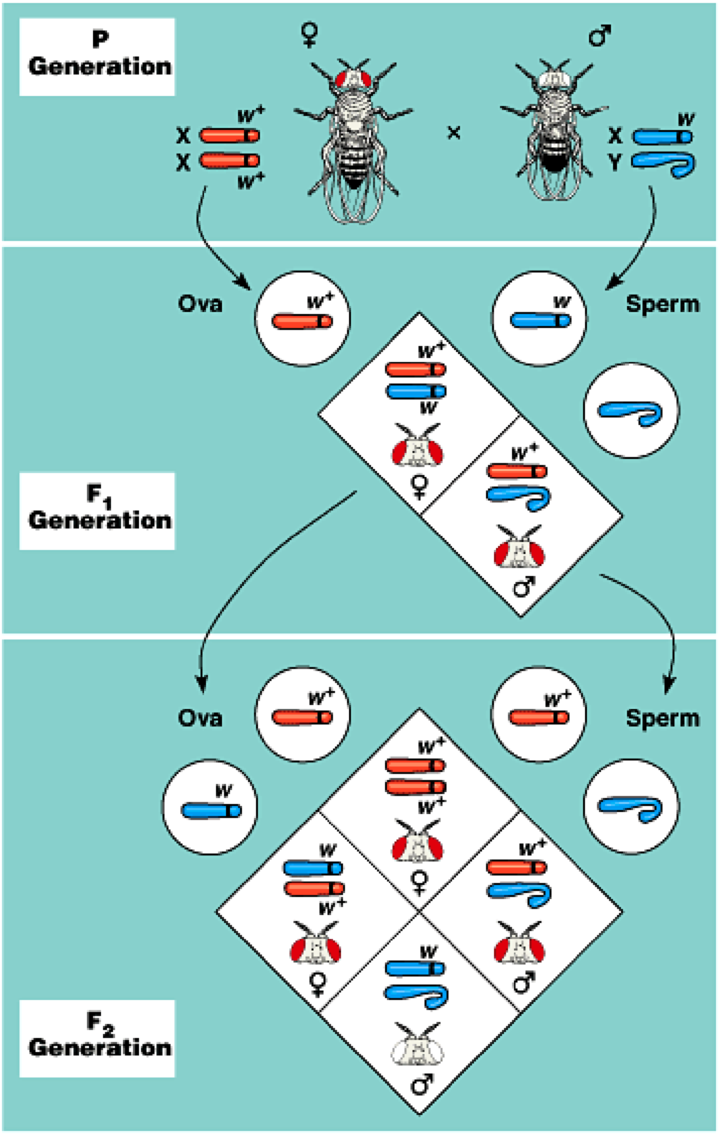
red eye female **x** white eye male

all red eye offspring

75% red eye female **x** 25% white eye male

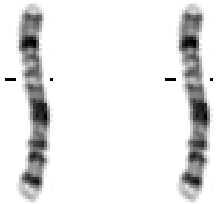
How is this possible?

Sex-linked trait!

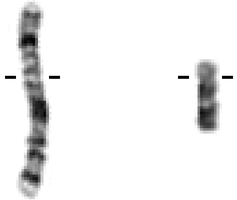


Sex-linked traits

- Although differences between women & men are many, the chromosomal basis of sex is rather simple
- In humans & other mammals, there are 2 sex chromosomes: X & Y
  - 2 X chromosomes develops as a female: XX

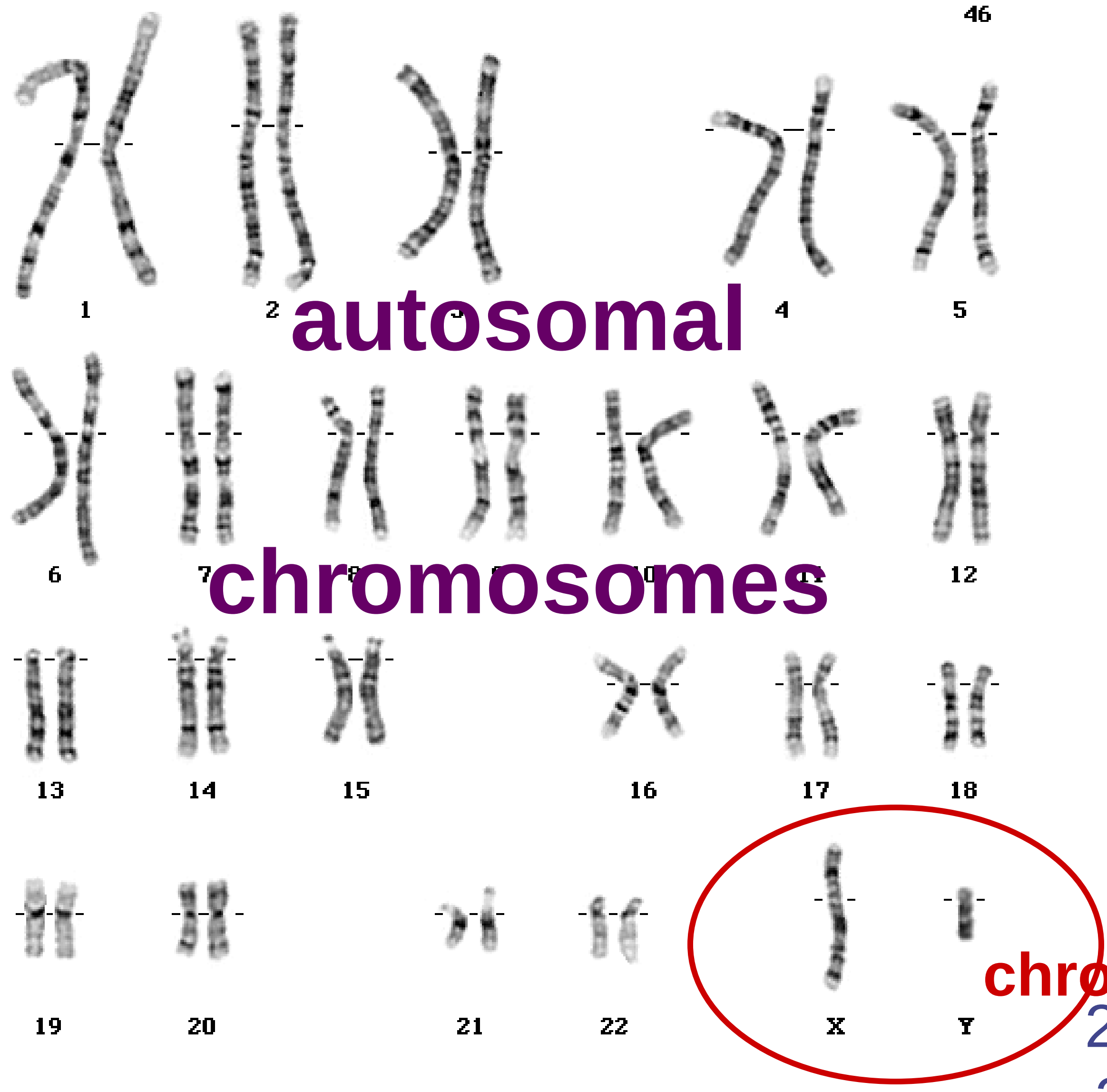
    

    - redundancy
  - an X & Y chromosome develops as a male: XY

    - no redundancy

Sex chromosomes



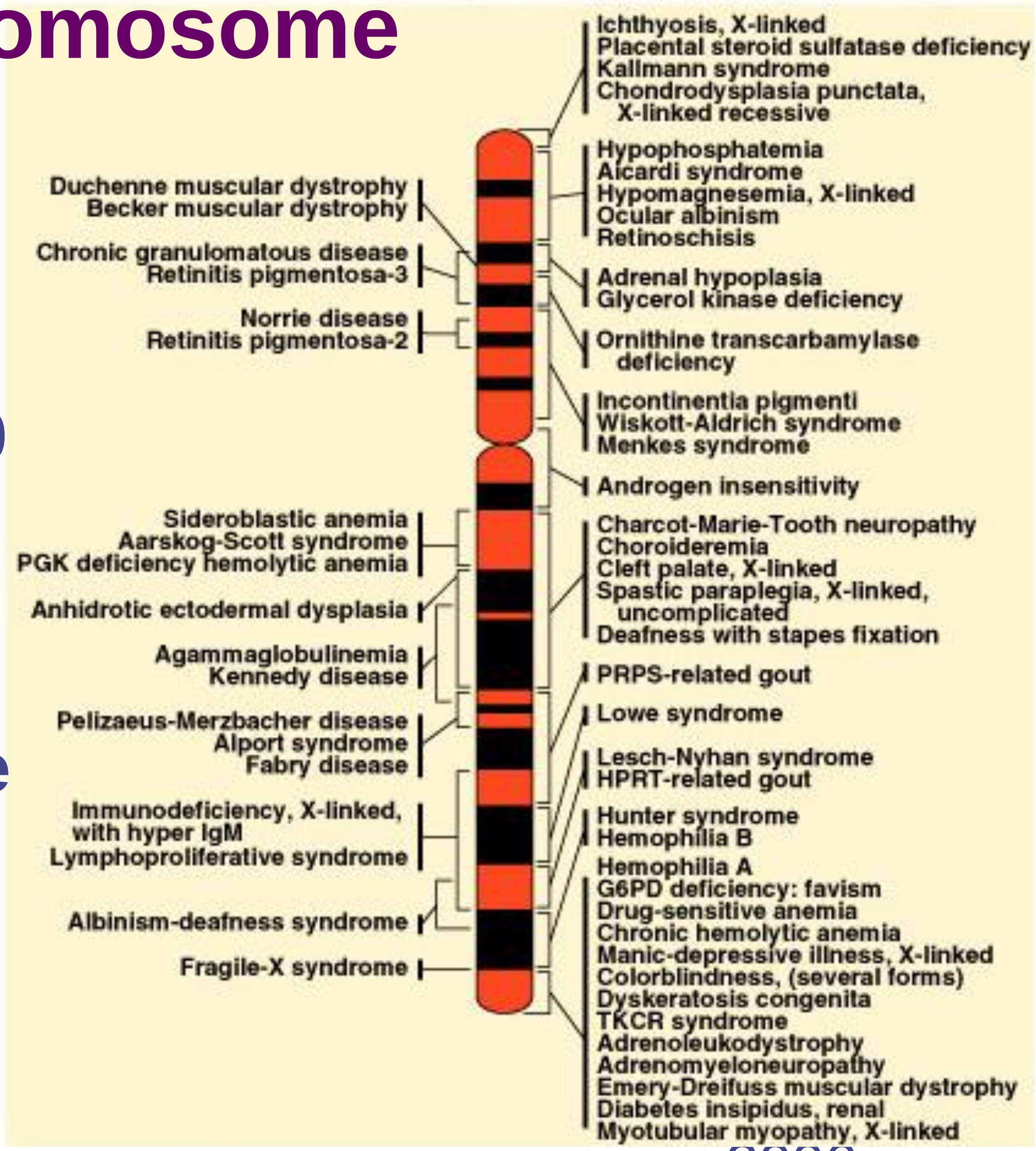
sex chromosomes

Genes on sex chromosomes

- Y chromosome
  - SRY: sex-determining region
    - master regulator for maleness
    - turns on genes for production of male hormones
      - pleiotropy!
- X chromosome
  - other traits beyond sex determination
    - hemophilia
    - Duchenne muscular dystrophy
    - color-blind

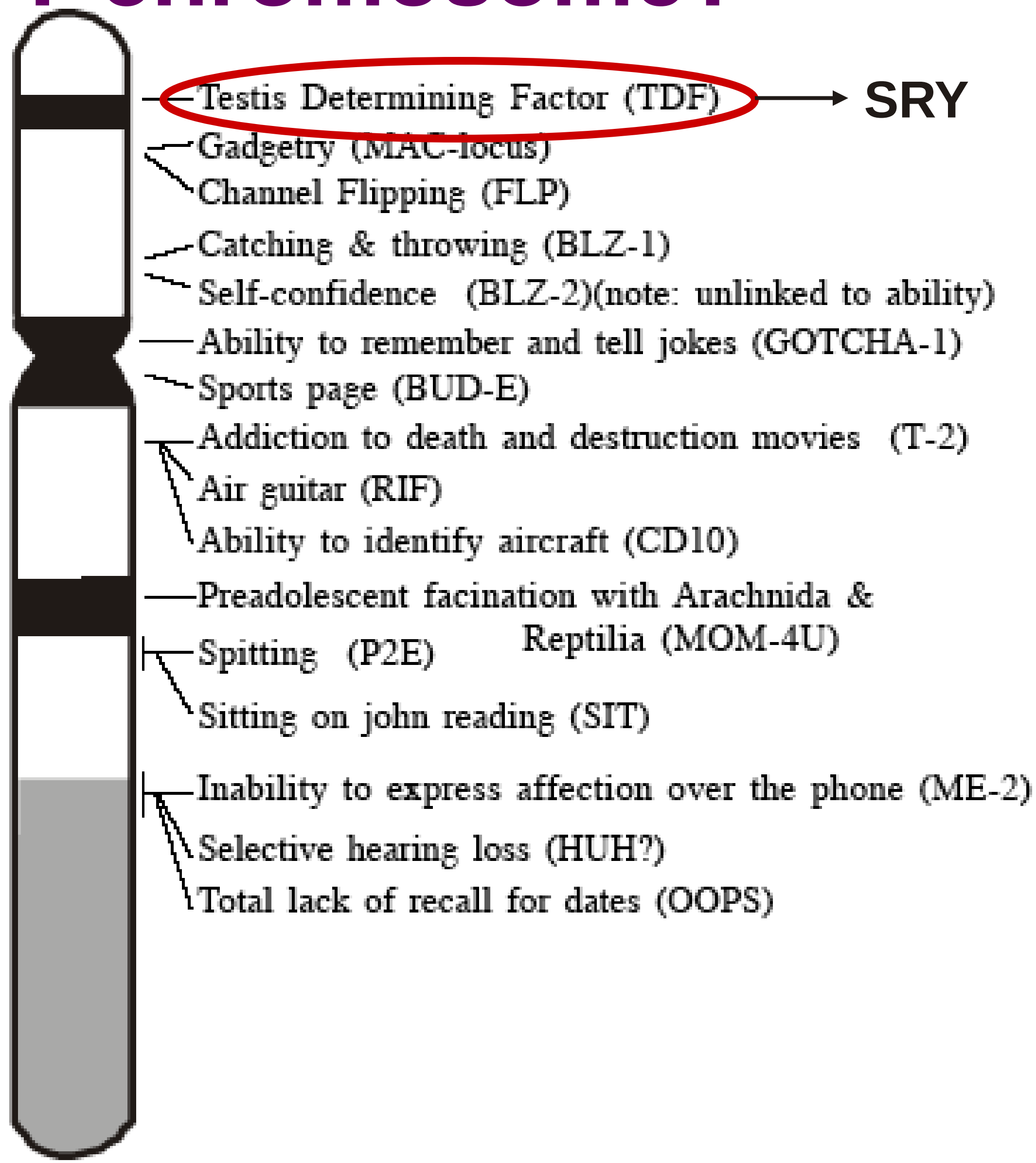
Human X chromosome

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

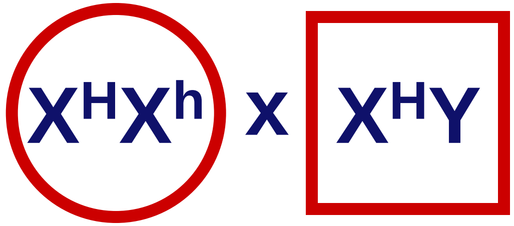


Map of Human Y chromosome?

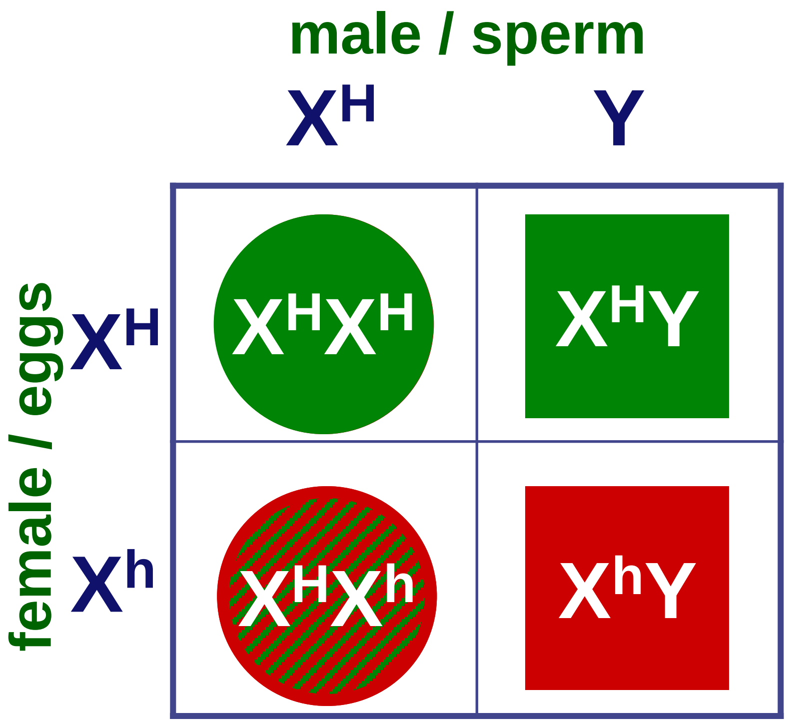
- < 30 genes on Y chromosome

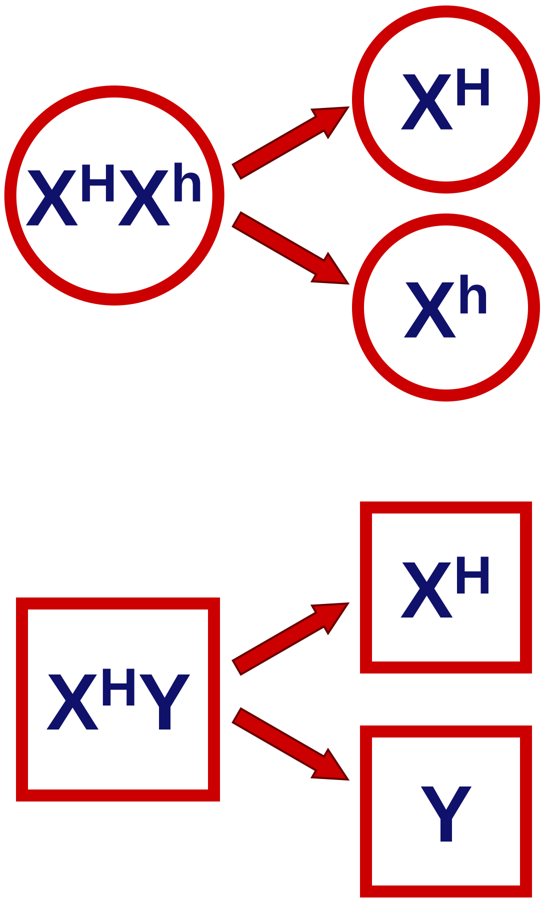


Sex-linked traits



sex-linked recessive





Sex-linked traits summary

- **X-linked**
  - **follow the X chromosomes**
  - **males get their X from their mother**
  - **trait is never passed from father to son**
- **Y-linked**
  - **very few traits**
  - **only 26 genes**
  - **trait is only passed from father to son**
  - **females cannot inherit trait**

Friday, April 4th

Please take out your lab activity #8, Population Genetics/Evolution.

- Mrs. Talley has a note card and PTC for you.
- You will need to move to your classroom you are testing **pronto**. Remember to explain what you are there to do, and what PTC is.
- Record the number of tasters AND non-tasters.

A. Perform a cross between a mother who is heterozygous type A blood with a type O father. What is the chance they will have a child with:

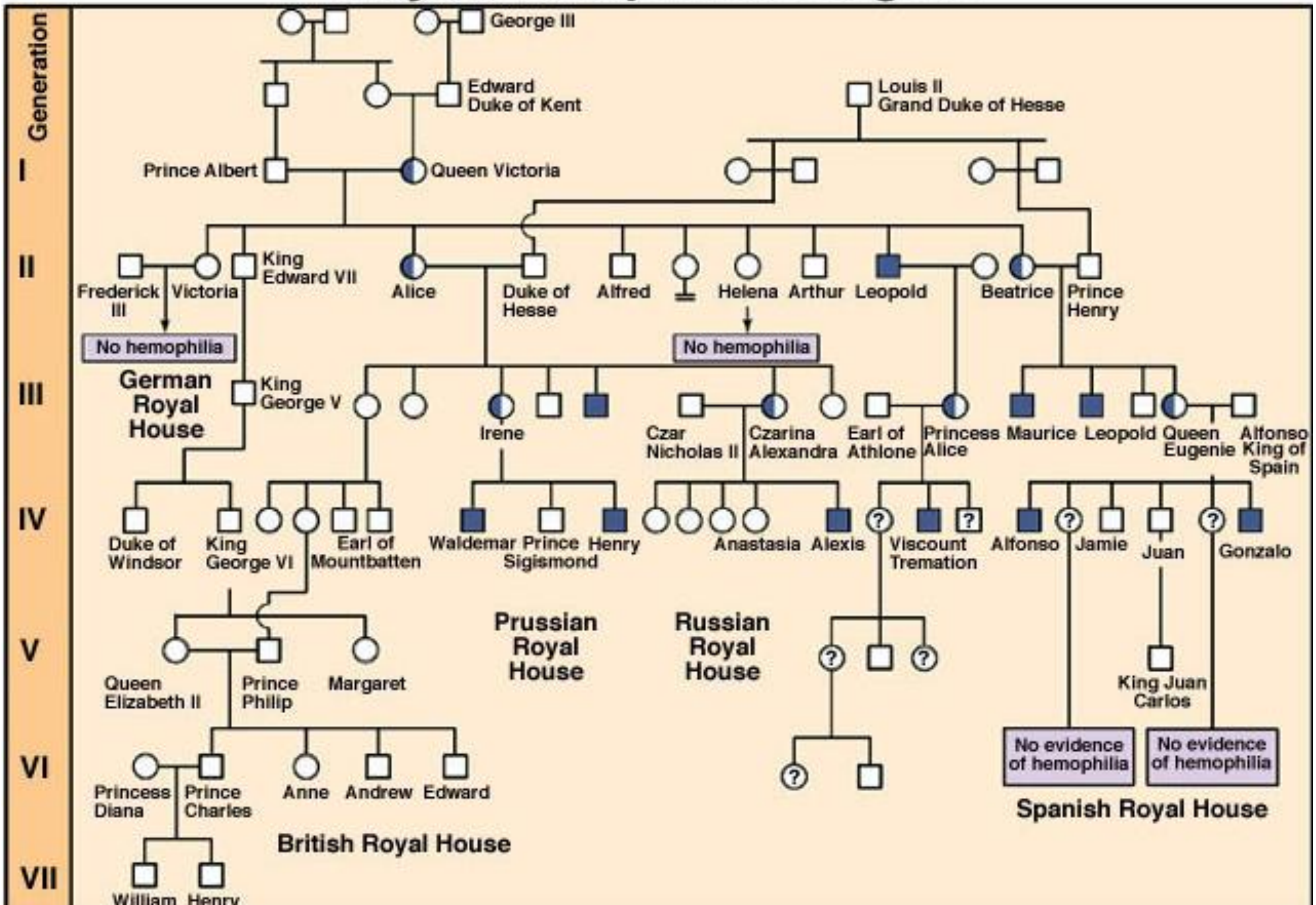
Type **A** blood? Type **B**? Type **AB**? Type **O**?

B. Cross a woman who is homozygous dominant for hemophilia with a man who has hemophilia. Recall, hemophilia is **sex-linked recessive**.

Predict the chance that they have a daughter **with** hemophilia/a son with hemophilia?

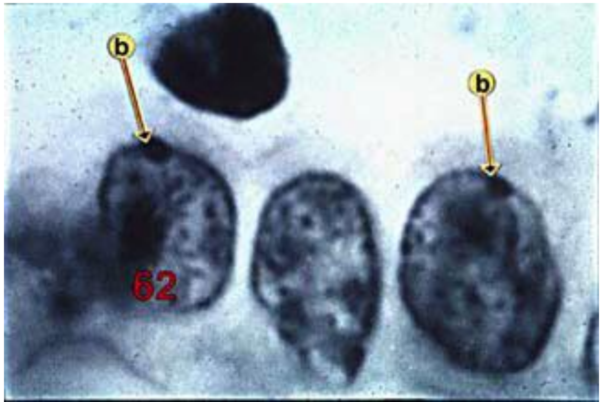
Queen Victoria and Descendants

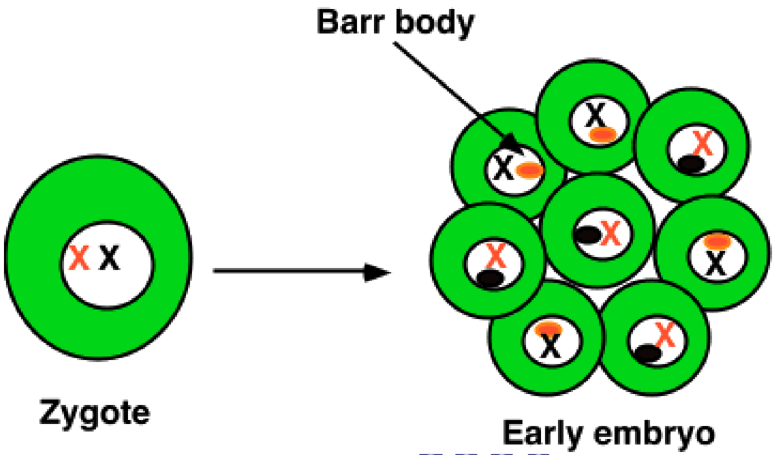
Royal Hemophilia Pedigree



X-inactivation

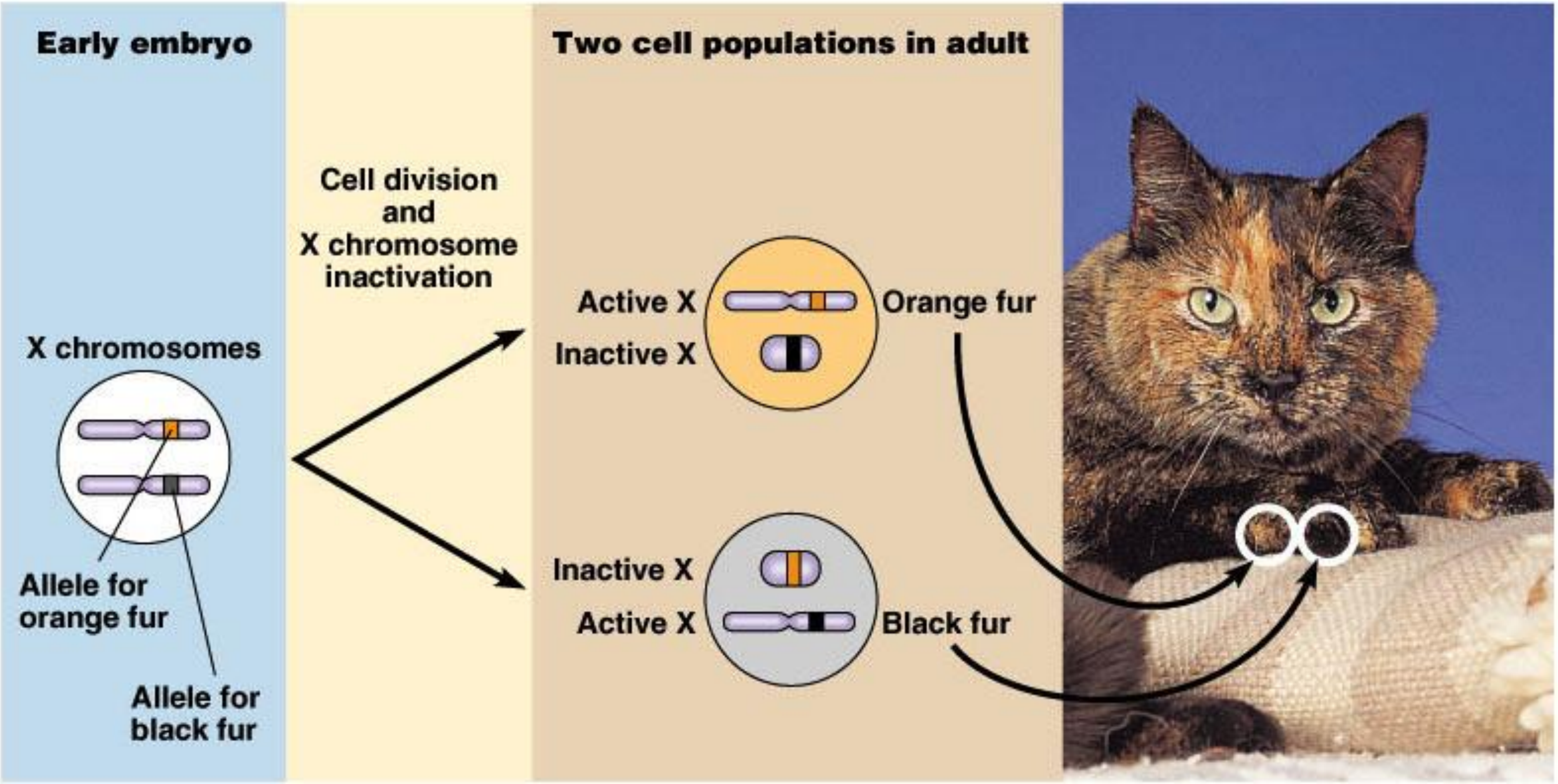
- Female mammals inherit two X chromosomes
  - one X becomes inactivated during embryonic development
    - condenses into compact object = Barr body





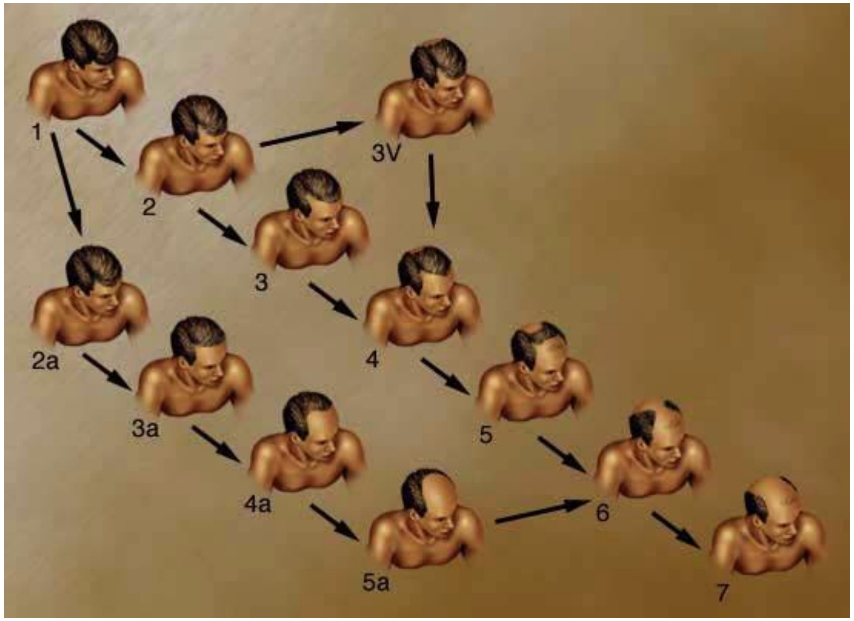
X-inactivation & tortoise shell cat

- 2 different cell lines in cat



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



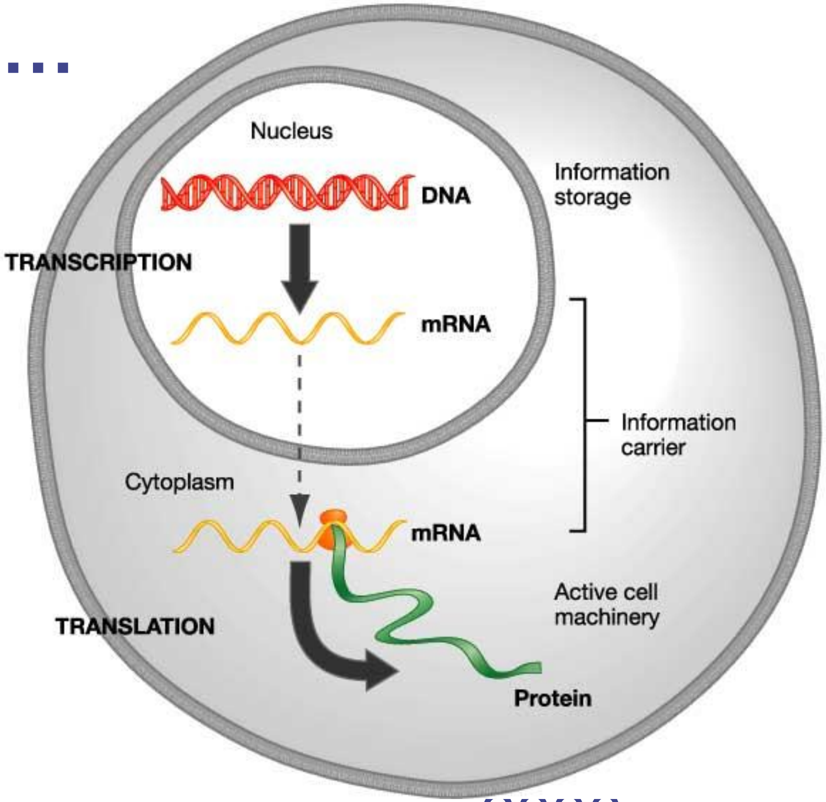
2006

Mechanisms of inheritance

- **What causes the differences in alleles of a trait?**
  - yellow vs. green color
  - smooth vs. wrinkled seeds
  - dark vs. light skin
  - Tay sachs disease vs. no disease
  - Sickle cell anemia vs. no disease

Mechanisms of inheritance

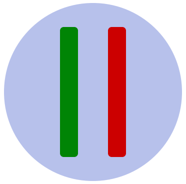
- **What causes dominance vs. recessive?**
  - **genes code for polypeptides**
  - **polypeptides are processed into proteins**
  - **proteins function as…**
    - **enzymes**
    - **structural proteins**
    - **hormones**



How does dominance work: enzyme

= allele coding for functional enzyme (green)

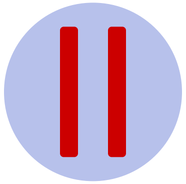
= allele coding for non-functional enzyme (red)



= **50% functional enzyme**
- sufficient enzyme present
- normal trait is exhibited
- NORMAL trait is DOMINANT

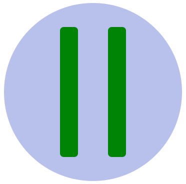
Aa

carrier



= **100% non-functional enzyme**
- normal trait is not exhibited

aa

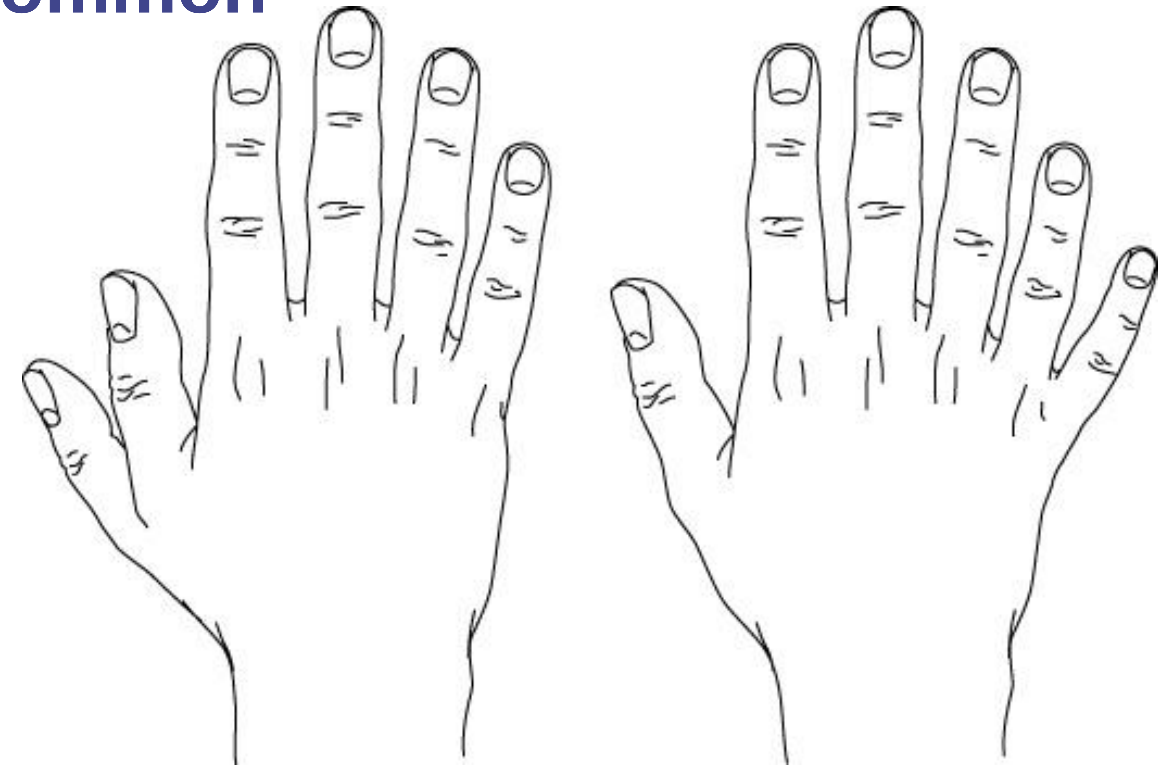


= **100% functional enzyme**
- normal trait is exhibited

AA

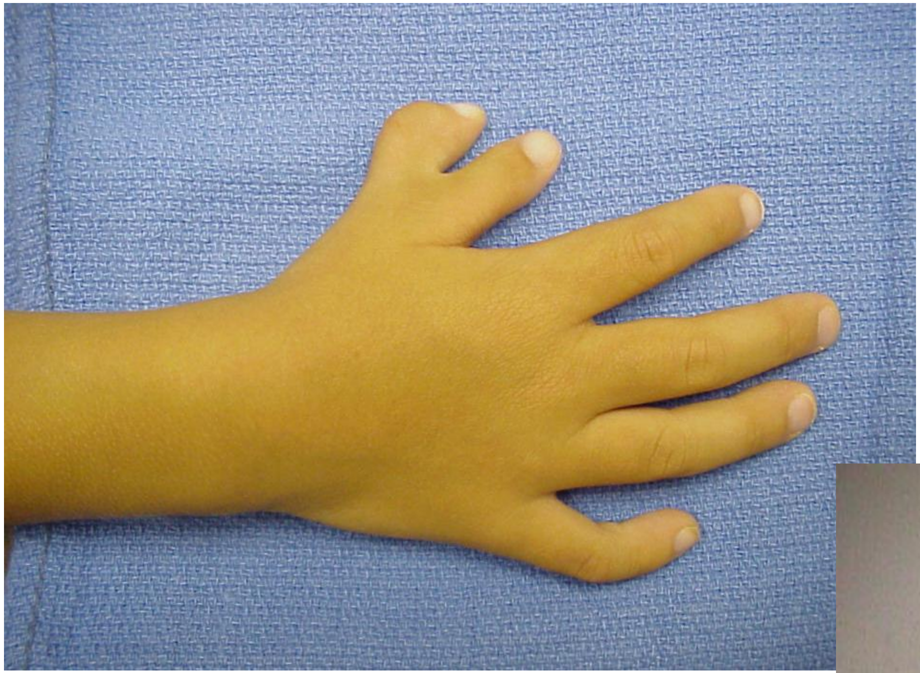
Prevalence of dominance

- **Because an allele is dominant does not mean…**
  - it is better
  - it is more common



Polydactyly: dominant allele

Polydactyly



individuals are born with extra fingers or toes

dominant to the recessive allele for 5 digits

recessive allele far more common than dominant

- → 399 individuals out of 400 have only 5 digits
- → most people are homozygous recessive (aa)

Hound Dog Taylor