



## 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



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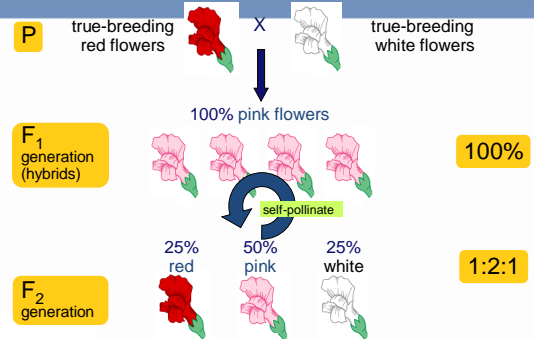
## Incomplete dominance

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



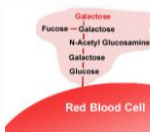
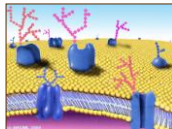
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## Incomplete dominance



## Co-dominance

- 2 alleles affect the phenotype equally & separately
  - not blended phenotype
  - human ABO blood groups
- 3 alleles
  - I<sup>A</sup>, I<sup>B</sup>, i
  - I<sup>A</sup> & I<sup>B</sup> alleles are co-dominant
    - glycoprotein antigens on RBC
    - I<sup>A</sup>I<sup>B</sup> = both antigens are produced
  - i allele recessive to both



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## Genetics of Blood type

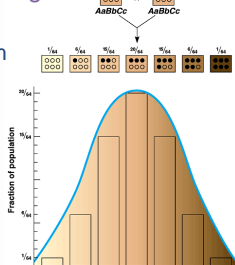
phenotype	genotype	antigen on RBC	antibodies in blood	donation status
A		antigens on surface of RBC	antibodies	—
B		antigens on surface of RBC	antibodies	—
AB		antigens on surface of RBC	antibodies	
O		on surface of RBC	antibodies	

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## 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



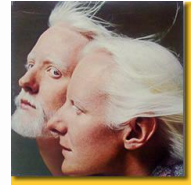
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## Skin color: Albinism

- Albinism is inherited as a single gene trait
  - aa = albino



albino Africans



melanin = universal brown color

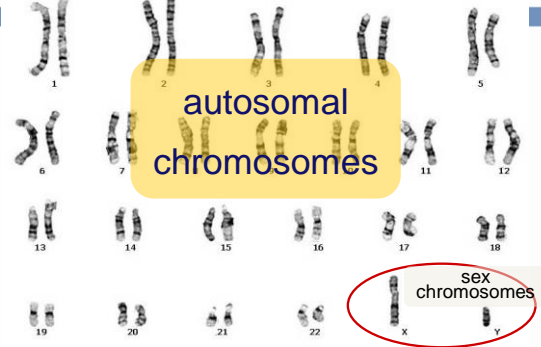


## Sex linked traits 1910 | 1933

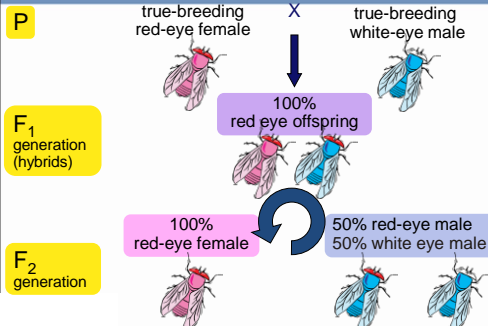
- Genes are on sex chromosomes
  - as opposed to autosomal chromosomes
  - first discovered by T.H. Morgan at Columbia U.
  - Drosophila* breeding
    - good genetic subject
      - prolific
      - 2 week generations
      - 4 pairs of chromosomes
      - XX=female, XY=male



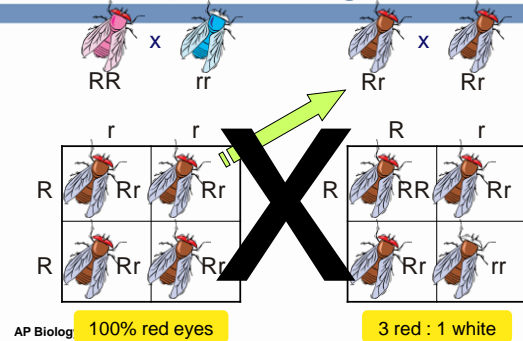
## Classes of chromosomes



## Discovery of sex linkage



## What's up with Morgan's flies?



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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

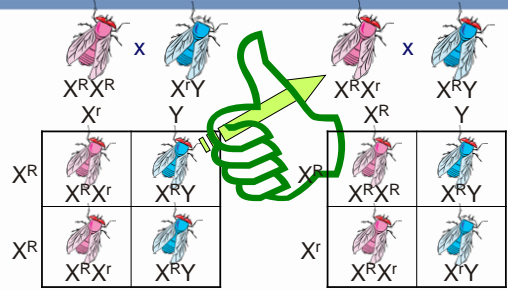


	X	Y
X	XX	XY
X	XX	XY

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50% female : 50% male

## Let's reconsider Morgan's flies...



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100% red eyes

100% red females

50% red males; 50% white males

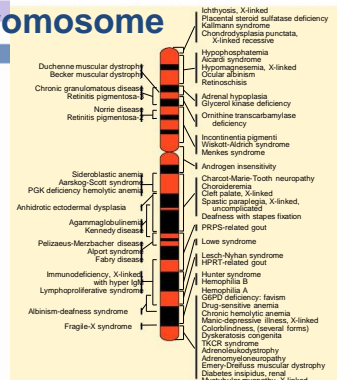
## Genes on sex chromosomes

- 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

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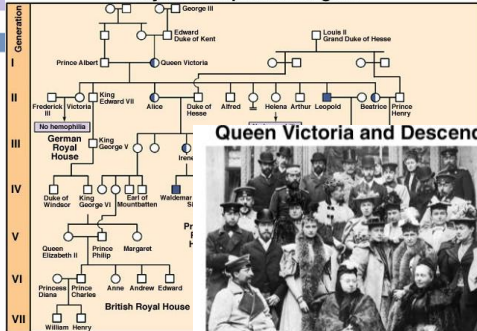
## Human X chromosome

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



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## Royal Hemophilia Pedigree



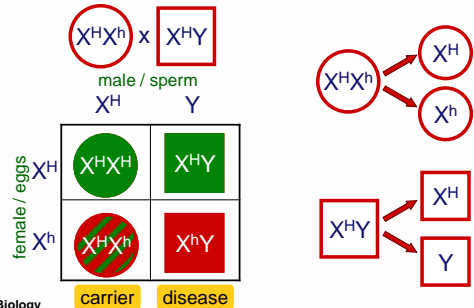
## Queen Victoria and Descendants



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## Hemophilia

sex-linked recessive

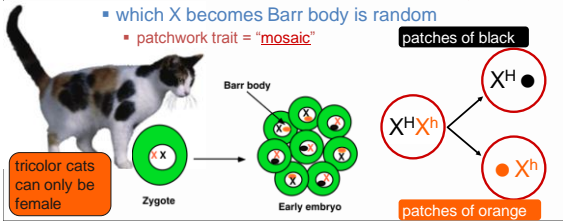


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carrier disease

## 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"



## 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



Color of Hydrangea flowers is influenced by soil pH



Coat color in arctic fox influenced by heat sensitive alleles