Chromosomes

Chapter 13

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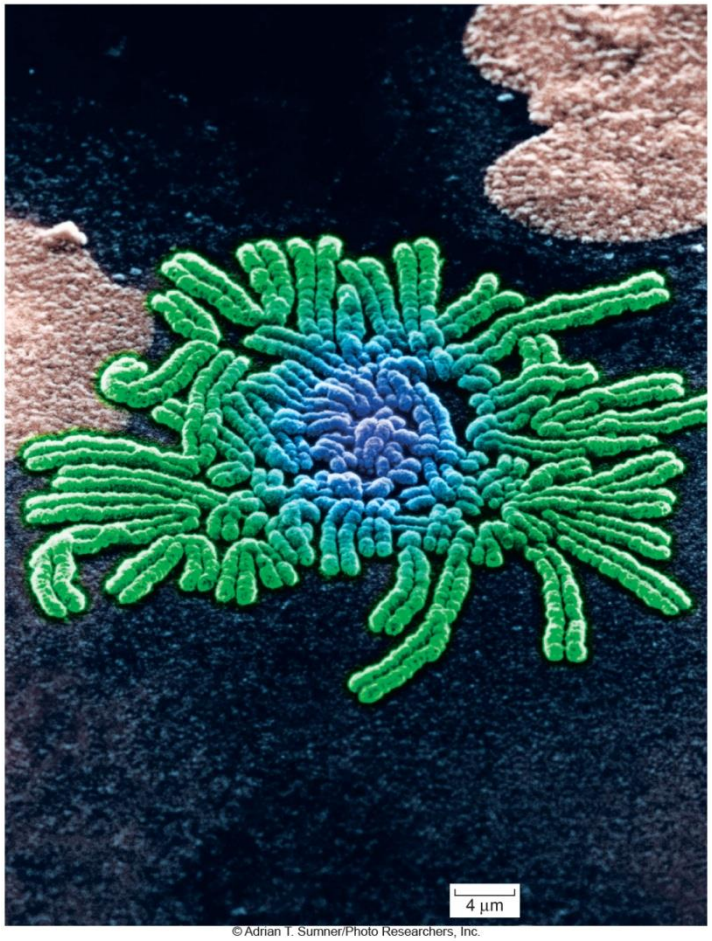

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http://commons.wikimedia.org/wiki/File:Carl_Correns.jpg

Chromosomal Theory of Inheritance

Carl Correns – 1900

- First suggests central role for chromosomes
- One of papers announcing rediscovery of Mendel's work

Walter Sutton – 1902

- Chromosomal theory of inheritance
- Based on observations that similar chromosomes paired with one another during meiosis

http://www.genetics.org/content/160/1/1/F1.expansion

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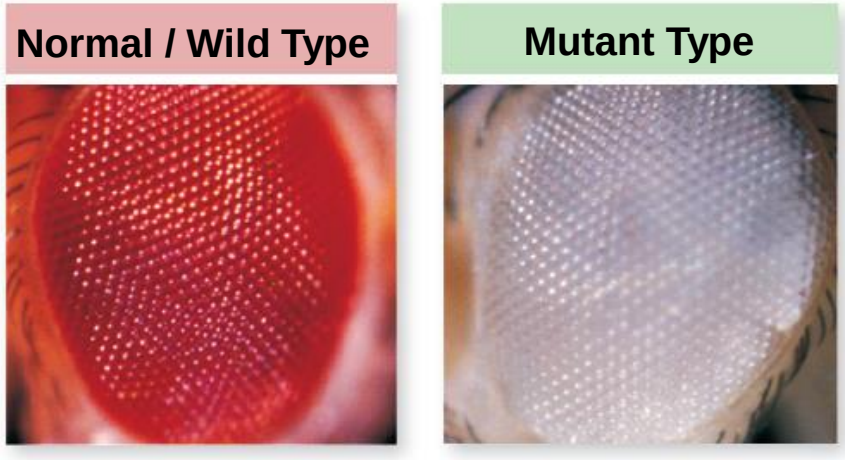

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Thomas Hunt Morgan – 1910

- Working with fruit fly, *Drosophila melanogaster*
- Discovered a mutant male fly with white eyes instead of red
- Crossed the mutant male to a normal red-eyed female
  - All F_1 progeny red eyed = dominant trait

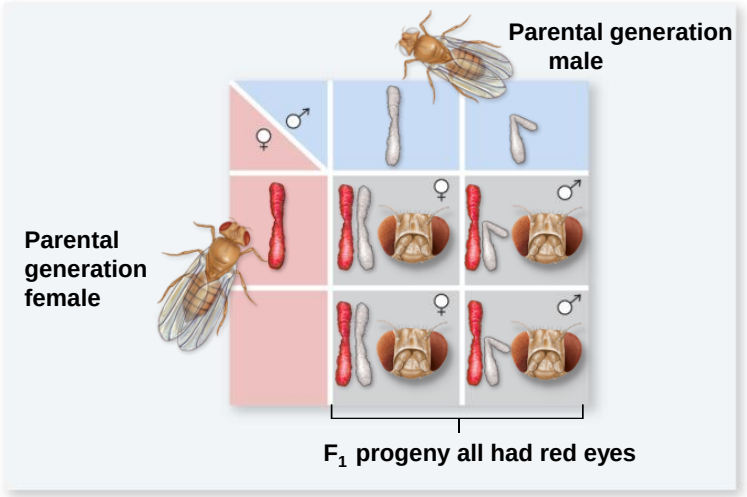
Nature Reviews | Genetics

http://commons.wikimedia.org/wiki/File:Drosophila_melanogaster.jpg

http://www.nature.com/nrg/journal/v2/n6/fig_tab/nrg0601_469a_F2.html

http://commons.wikimedia.org/wiki/File:Drosophila_melanogaster_laboratory_culture-vial.jpg

- Morgan crossed F_1 females x F_1 males



http://www.art.com/products/p14441303756-sa-i6716313/solvin-zankl-fruit-fly-wild-type-drosophila-melanogaster-lab-culture.htm

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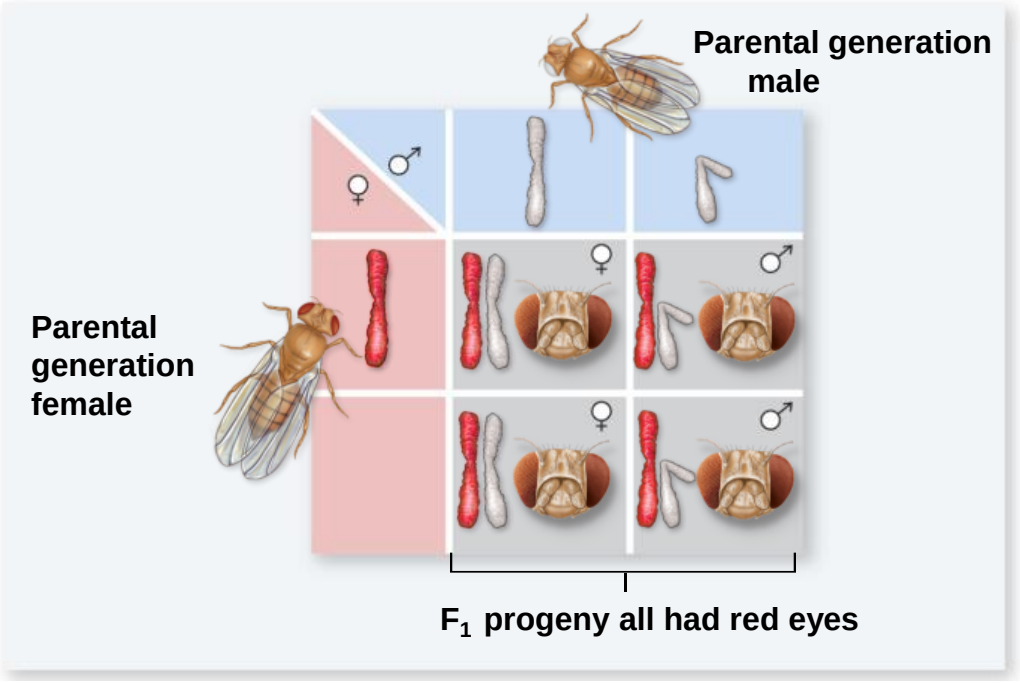
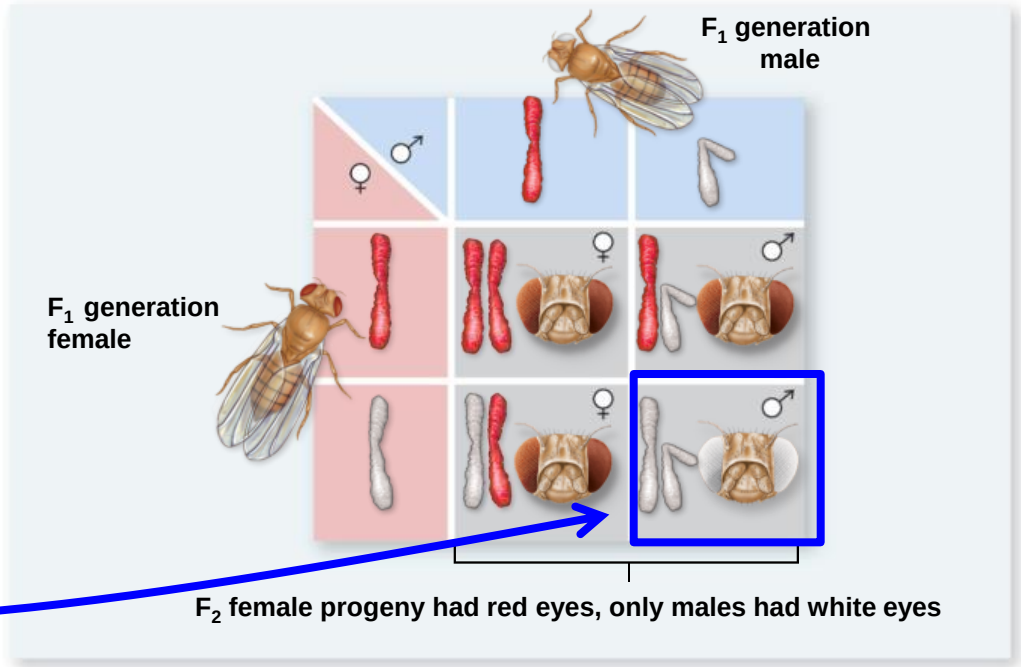
- Morgan crossed F_1 females x F_1 males
  - F_2 generation contained red and white- eyed flies
    - But all white-eyed flies were male





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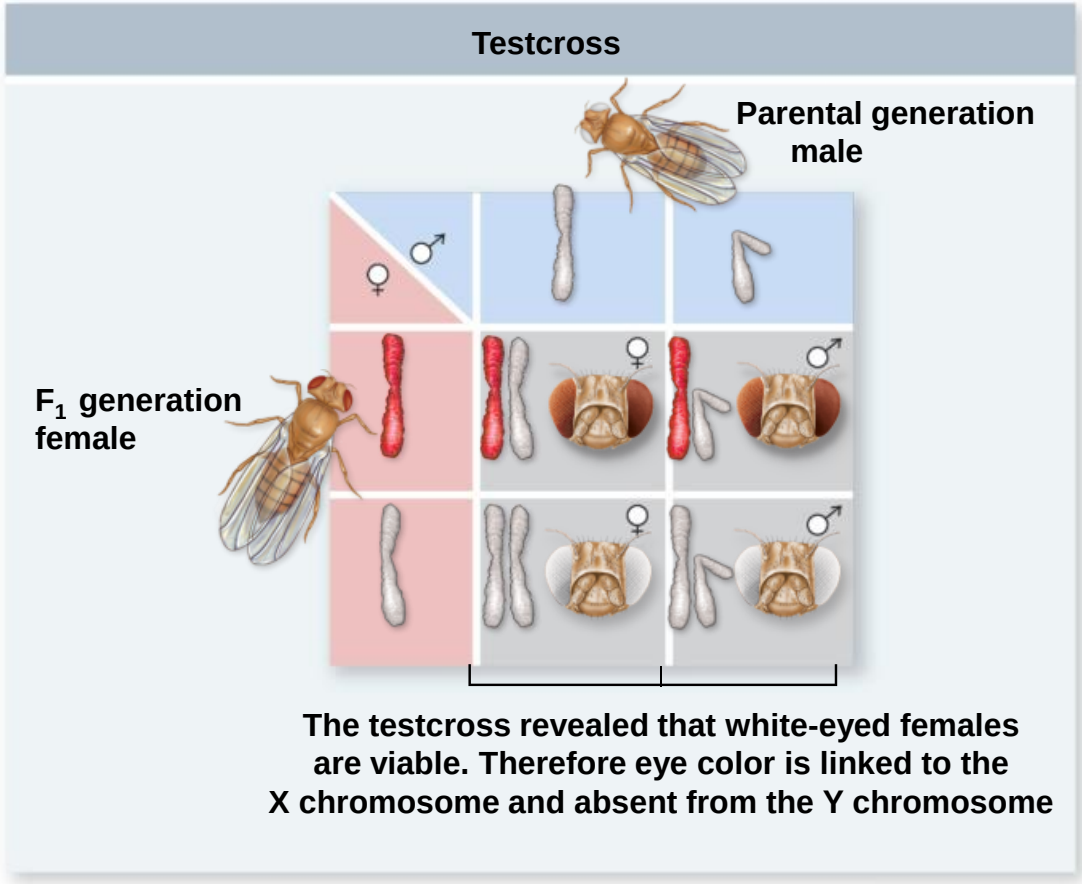
- Testcross of a F_1 female with a white-eyed male showed the viability of white-eyed females

- Morgan concluded that the *eye color gene resides on the X chromosome*



Sex Chromosomes

- **Sex determination** in *Drosophila* is based on the number of X chromosomes
  - 2 X chromosomes = female
  - 1 X and 1 Y chromosome = male

- Sex determination in humans is based on the presence of a Y chromosome
  - 2 X chromosomes = female
  - Having a Y chromosome (XY) = male

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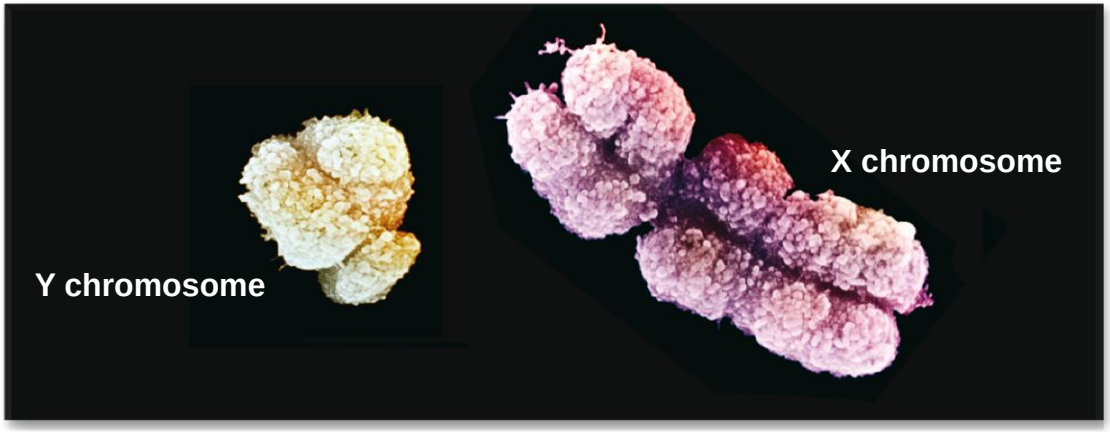

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35,000 ×

- Humans have 46 total chromosomes
  - 22 pairs are autosomes
  - 1 pair of sex chromosomes
  - Y chromosome highly condensed
    - Recessive alleles on male's X have no active counterpart on Y
  - "Default" for humans is female
    - Requires *SRY* gene on Y for "maleness"

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TABLE 13.1 Sex Determination in Some Organisms

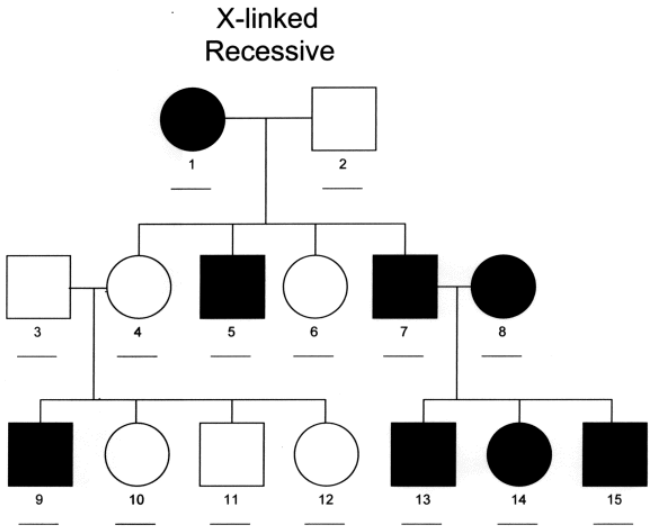
	Female	Male
Humans, *Drosophila*  	XX	XY
Birds 	ZW	ZZ
Grasshoppers 	XX	XO
Honeybees 	Diploid	Haploid

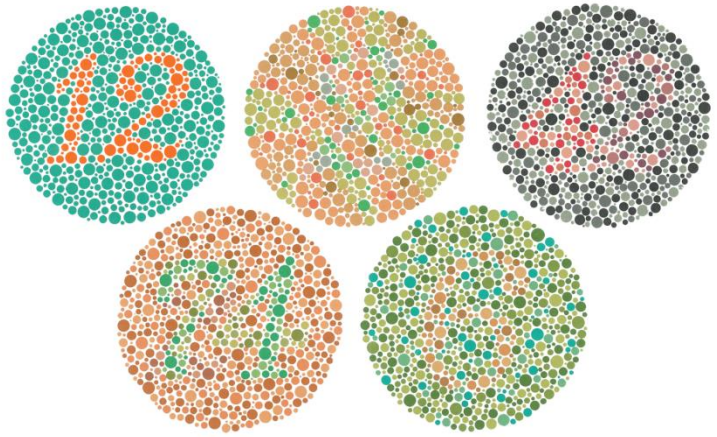
https://migrc.org/Library/SexlinkedRecessive.html

Sex Linkage

Can you determine the genotypes?

- Certain genetic diseases affect males to a greater degree than females
- **X-linked recessive alleles**
  - **Red-green color blindness**
  - Hemophilia

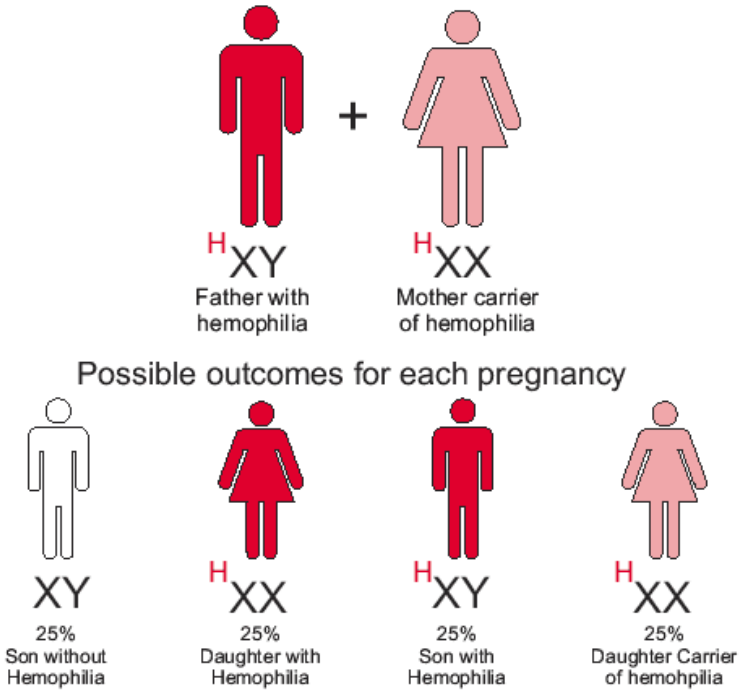




http://www.nationalguard.com/forums/showthread.php/26230-Help-with-Color-Blindness-and-joining-the-Military

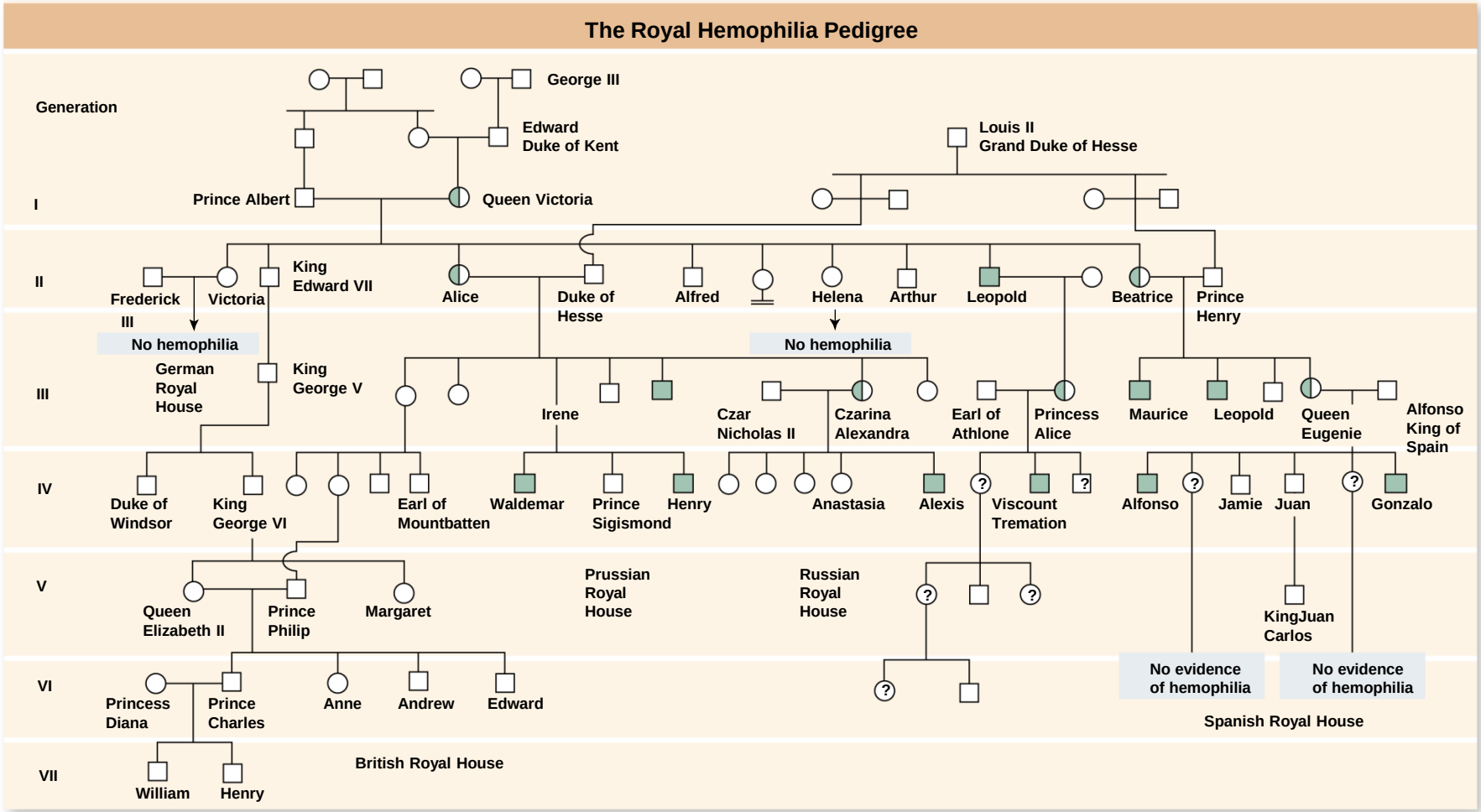
Hemophilia

- Disease that affects a single protein in a cascade of proteins involved in the formation of blood clots
- Form of hemophilia is caused by an X-linked recessive allele
  - Heterozygous females are asymptomatic carriers



http://www.hemophilia-information.com/hemophilia-genetics.html

- Allele for hemophilia was introduced into a number of different European royal families by Queen Victoria of England



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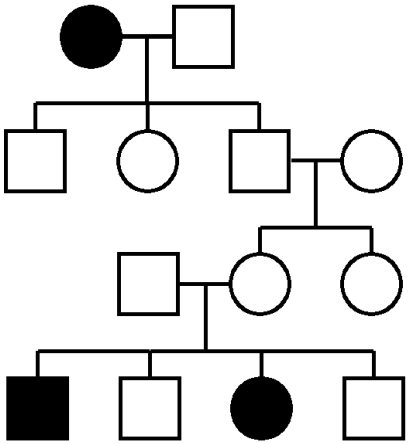
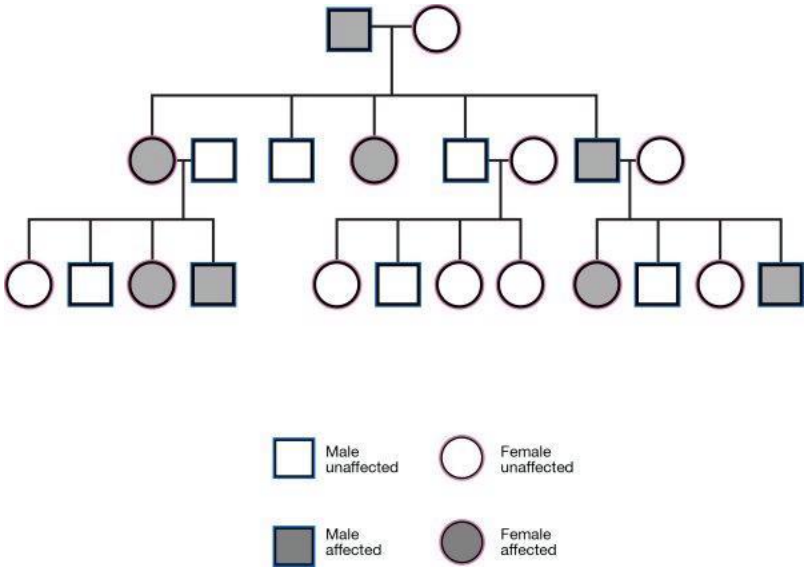
Autosomal Disease Pedigrees



Autosomal Recessive



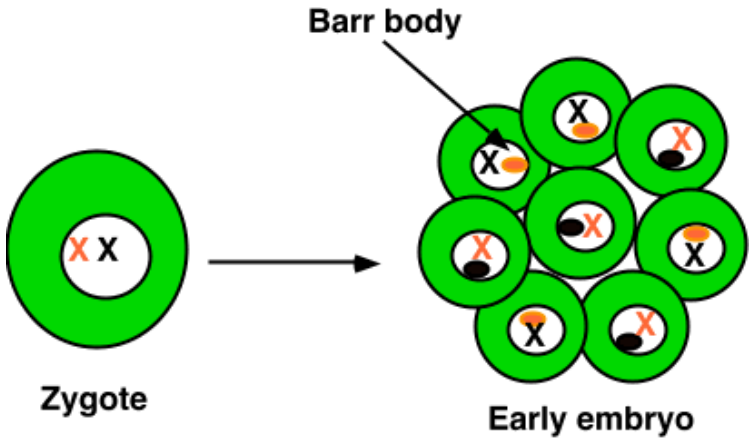
Autosomal Dominant

- faculty.clintoncc.suny.edu - 519 × 567 - More sizes
- www.genome.gov

Dosage compensation

Ensures an equal expression of genes from the sex chromosomes even though females have 2 X chromosomes and males have only 1

- In each female cell, 1 X chromosome is *inactivated and is highly condensed* into a **Barr body**
- Females heterozygous for genes on the X chromosome are **genetic mosaics**



http://bioserv.fiu.edu/~walterm/human_online/inheritance/introduction_to_genetics.htm

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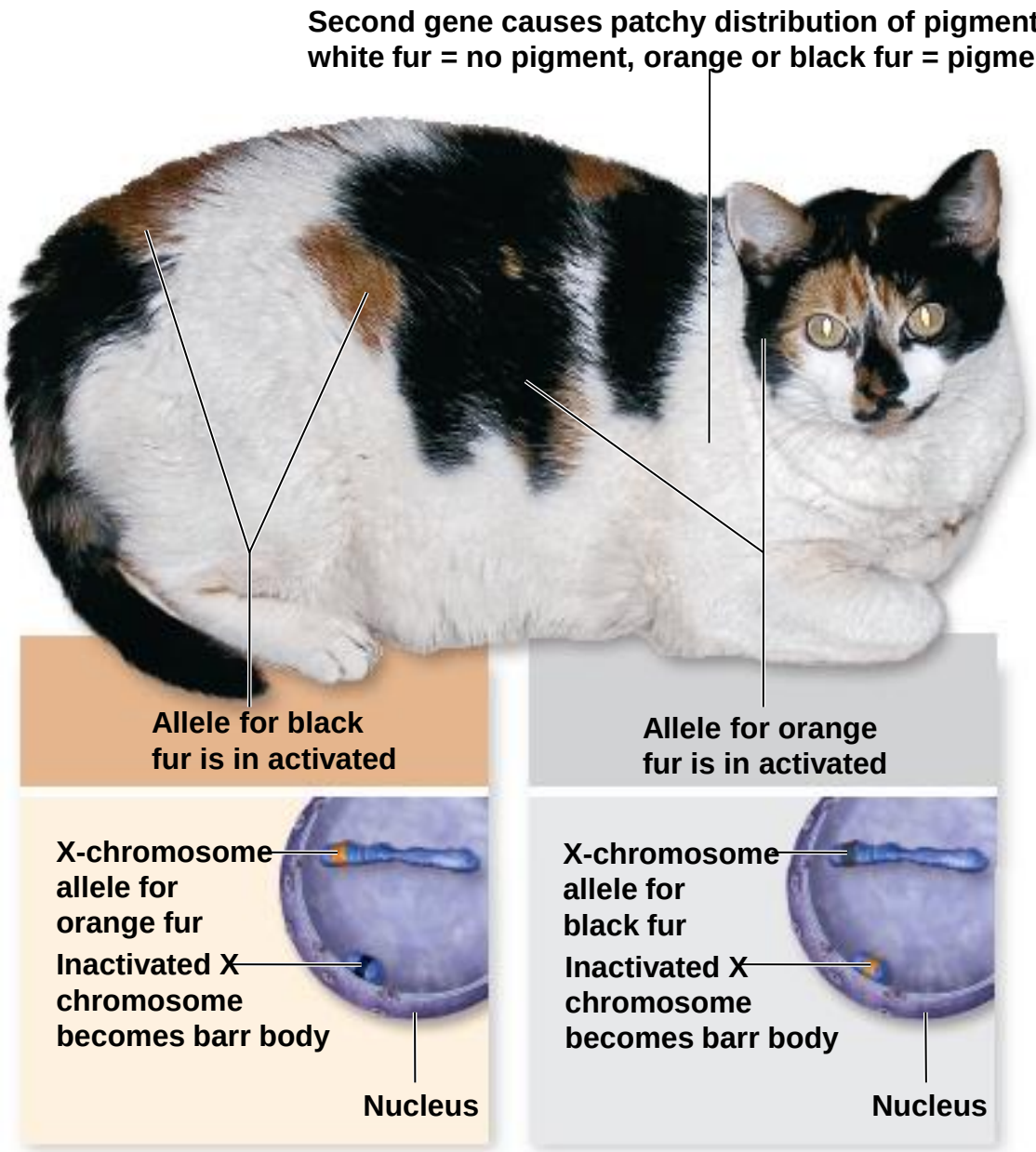
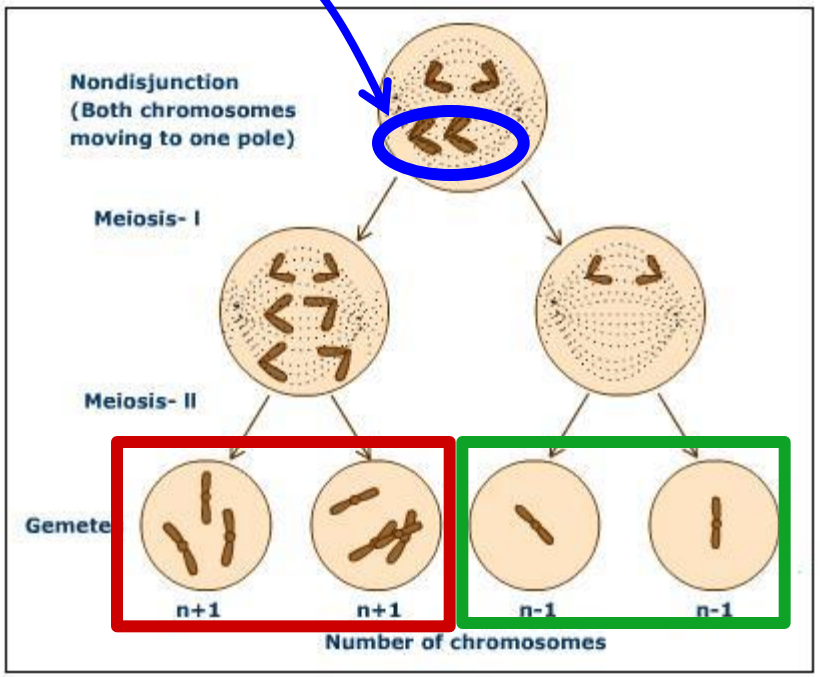


(top): © Kenneth Mason

- Calico cat
- X-chromosome inactivation in females

Nondisjunction

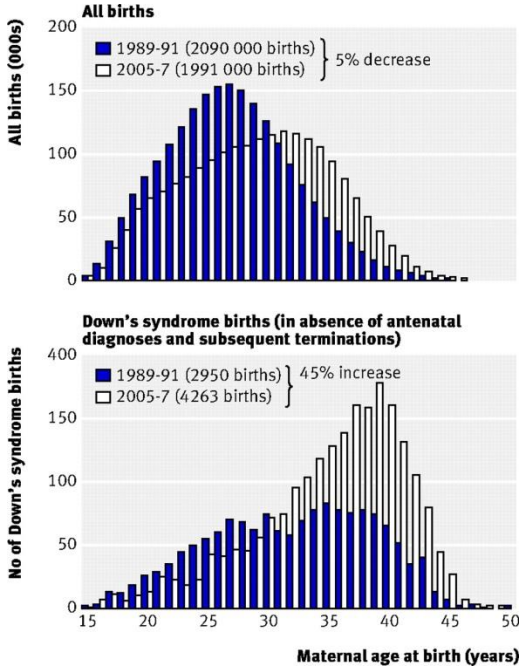
- Failure of homologues or sister chromatids to separate properly during meiosis



- **Aneuploidy** – gain or loss of a chromosome
  - **Monosomy** – loss
  - **Trisomy** – gain
  - In all but a few cases, do not survive

http://www.tutorvista.com/content/biology/biology-iii/human-genetics/genetic-diseases.php

- Smallest autosomes can present as 3 copies and allow individual to survive
  - 13, 15, 18, 21 and 22
  - 13, 15, 18 – severe defects, die within a few months
  - 21 and 22 – can survive to adulthood
  - **Down Syndrome – trisomy 21**
    - May be a full, third 21st chromosome
    - May be a translocation of a part of chromosome 21
    - Mother's age influences risk



http://www.bmj.com/content/339/bmj.b3794

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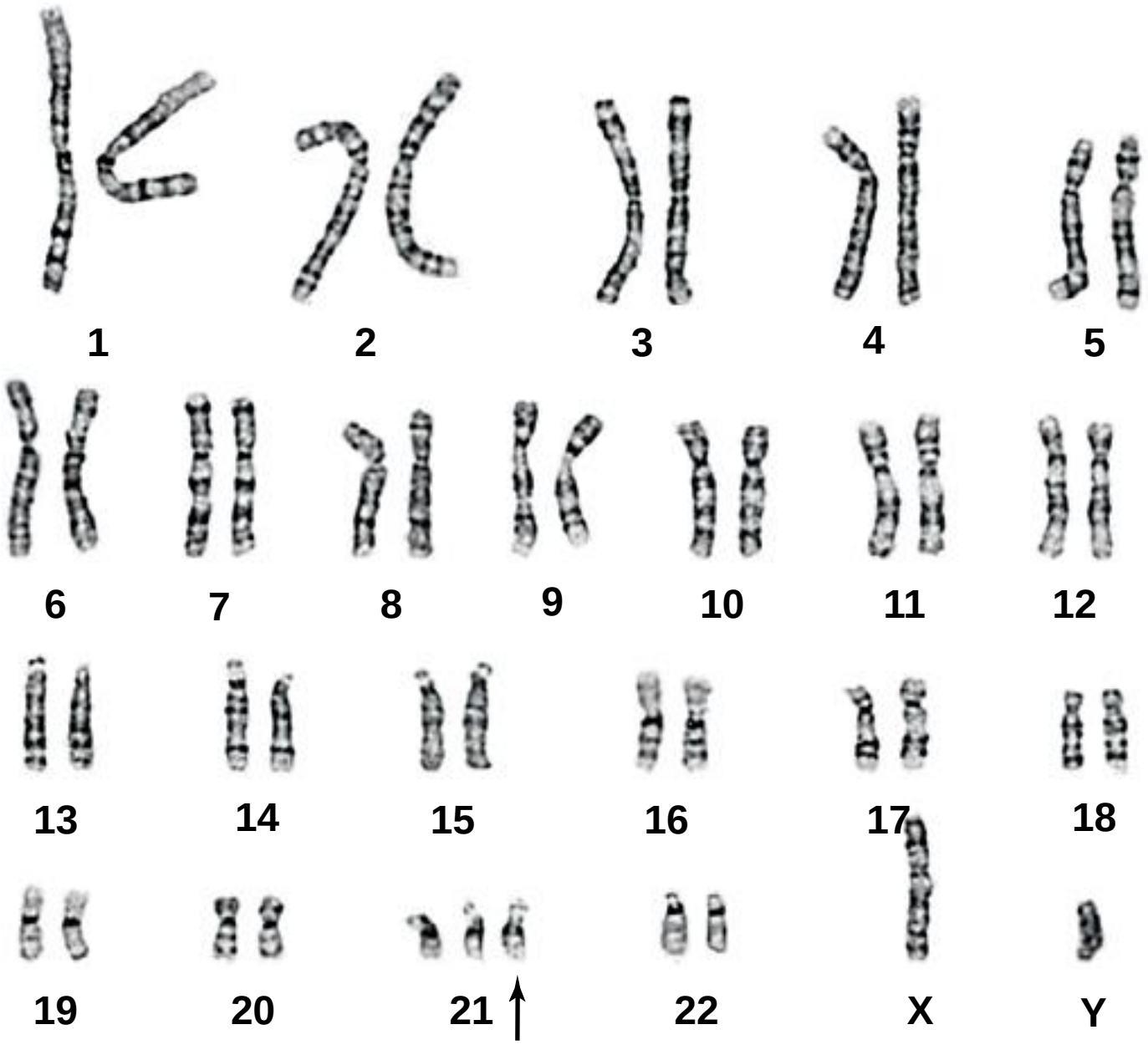

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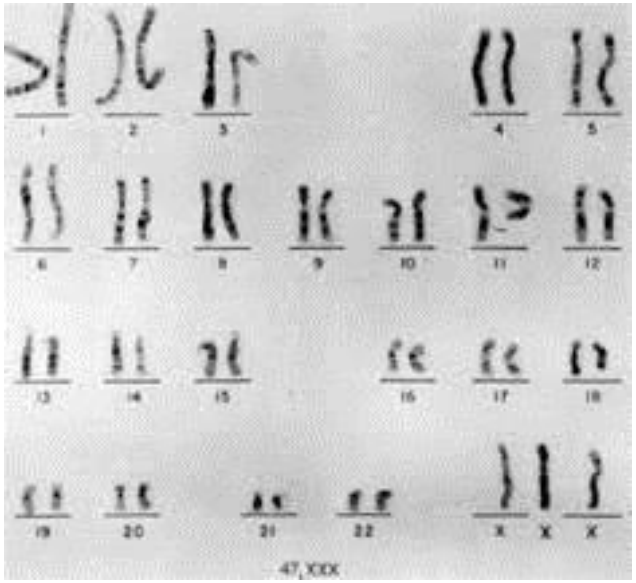
Nondisjunction of sex chromosomes

- Do not generally experience severe developmental abnormalities
- Individuals have somewhat abnormal features, but often reach maturity and in some cases may be fertile



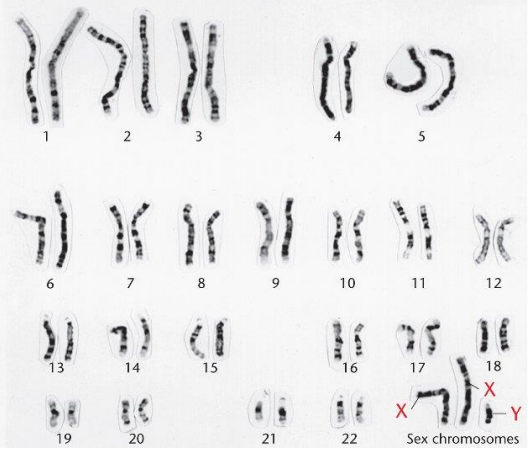
Because Barr bodies are formed from 2 of the three X chromosomes, Turner syndrome (XXX) females have only mild effects.

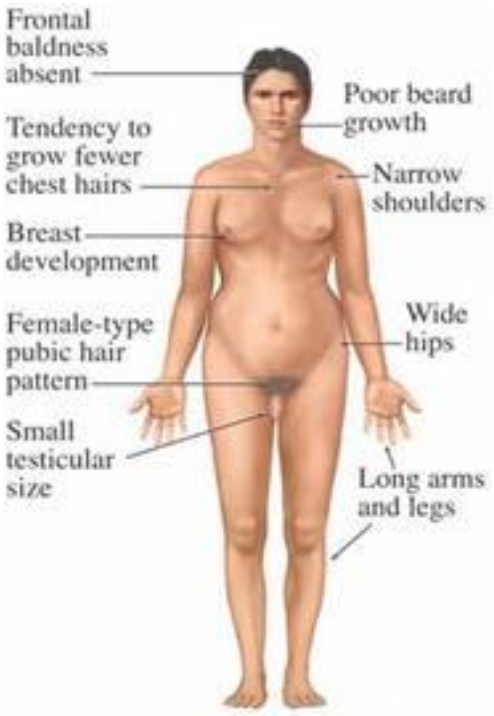
http://bio3400.nicerweb.com/Locked/media/ch07/Klinefelter_syndrome.html

Nondisjunction of sex chromosomes

- XXX – **triple-X females**
- XXY – males (**Klinefelter syndrome**)
- XO – females (**Turner syndrome**)
- OY – nonviable zygotes
- XYY – males (**Jacob syndrome**)

Klinefelter syndrome





http://cv-mrsdabbs.wikispaces.com/C+%26+GD--+Abnormal+Numbers+of+Sex+Chromosomes