

# Lecture 3: From Cells & Chromosomes to Heredity & Society

Section A: From Nature to Concepts

Genes & Society  
LSM3201 / GEK 1527

If you watch animals objectively for any length of time, you're driven to the conclusion that their main aim in life is to pass on their genes to the next generation.

- David Attenborough (Naturalist; Broadcaster)

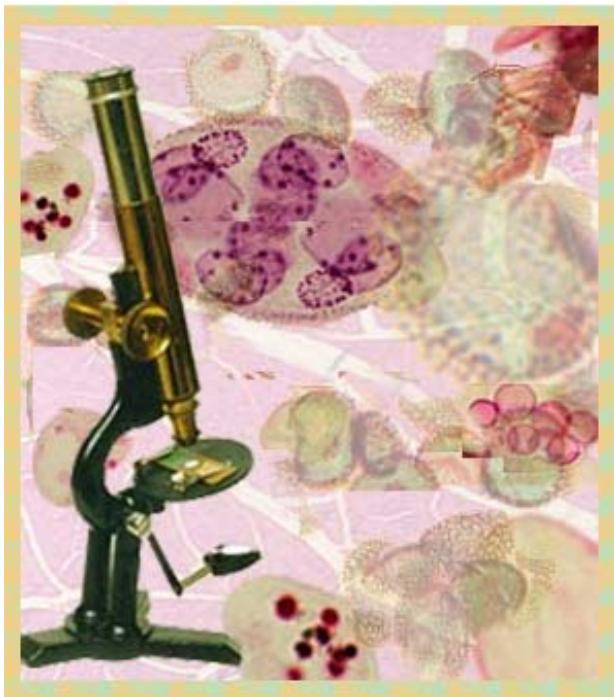


# Overview



- From Cells to Chromosomes
- Chromosomal Theory of Inheritance, Meiosis & Mitosis
- Heredity & Society
- Chromosomal Aberrations

# From Cells to Chromosomes



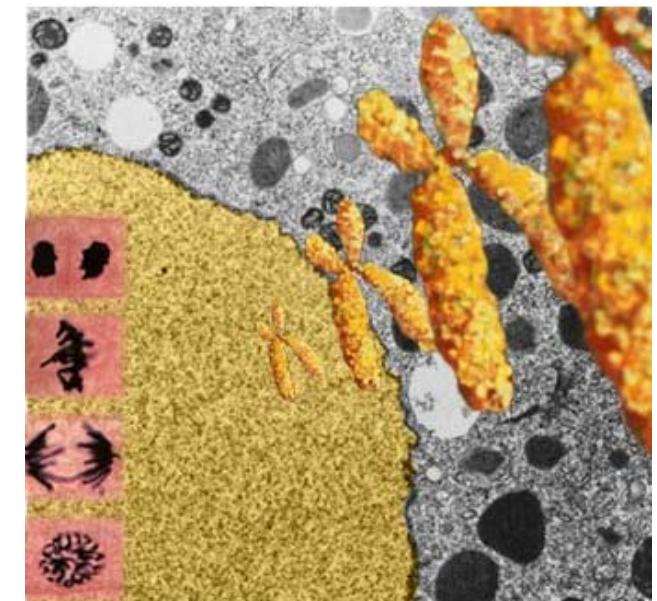
By 1830s, there was strong evidence that cells are the basic units of life.

1838: Schleiden & Schwann proposed Cell Theory that states “All living things are made of cells”

1858: Rudolph Virchow proposed that “every cell only arises from a preexisting cell”

1870s: Biological stains were developed that highlighted structures within cells — including thread-like chromosomes. Different organisms have different numbers of chromosomes, suggesting that they might carry information specific for each life form.

<http://www.dnaftb.org/6/>

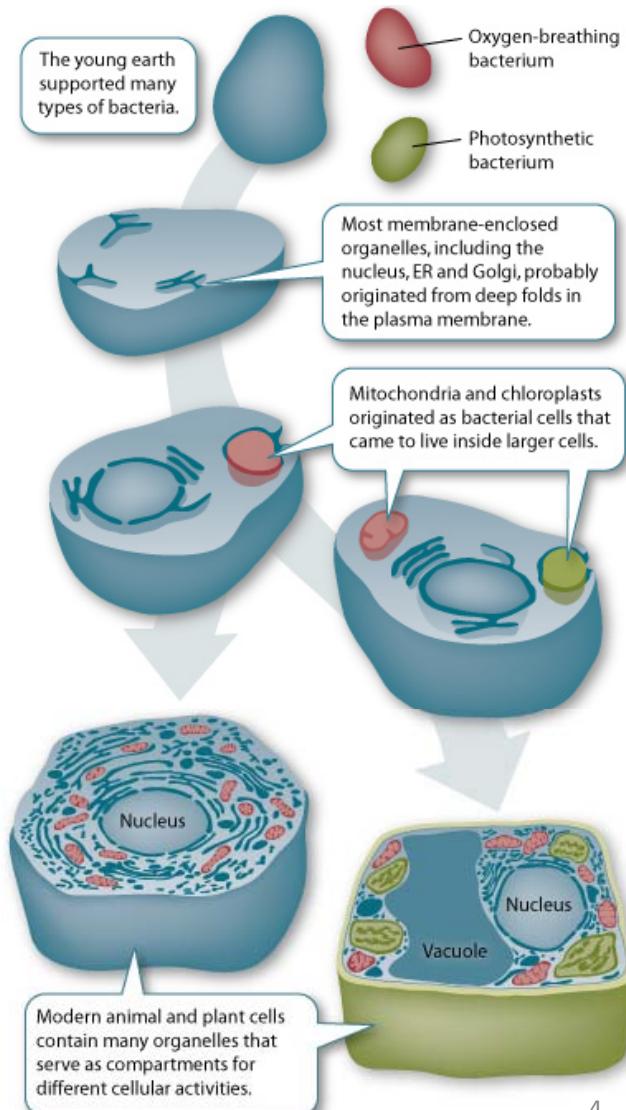


If cells are the basic units of life, they too must have a reproductive mechanism that maintains the proper chromosome number in each cell.

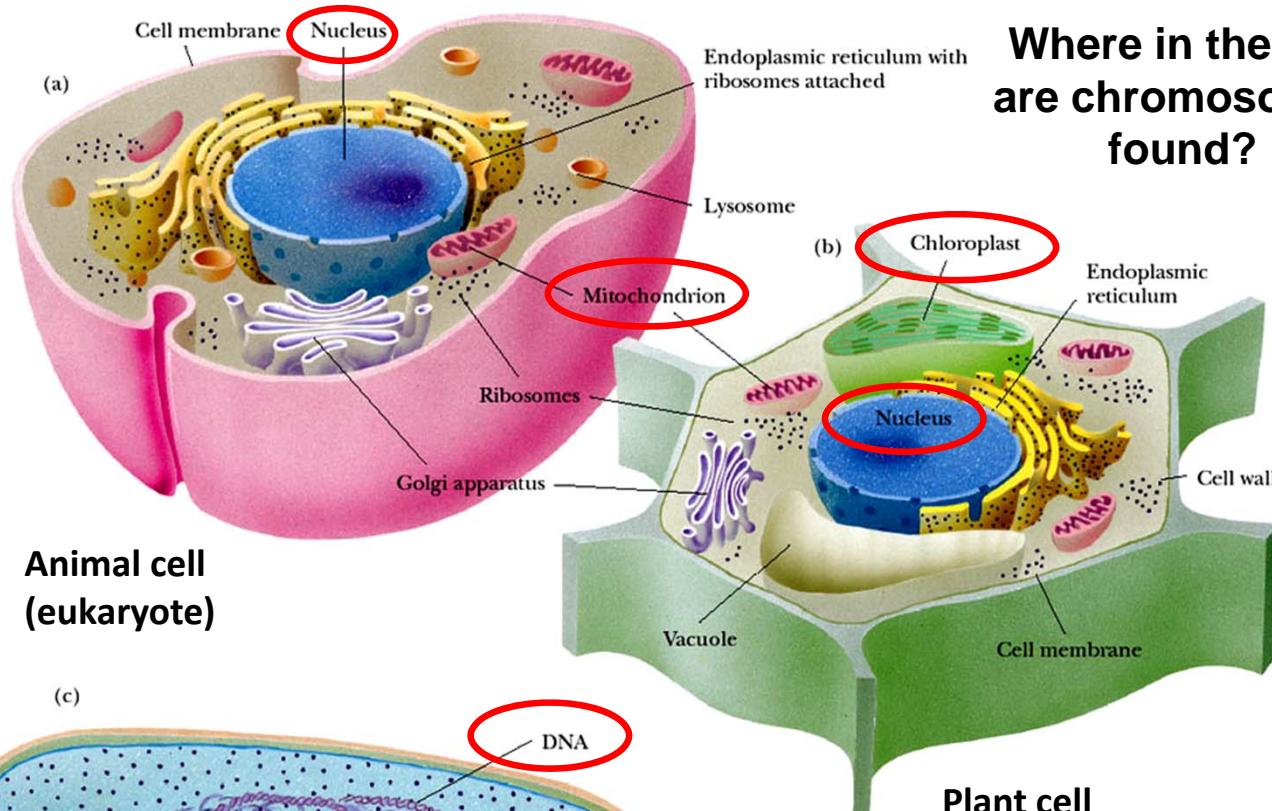
1882: Walther Flemming carefully documented the behavior of chromosomes during cell division (mitosis).

<http://www.dnaftb.org/7/>

## Endosymbiotic theory



## Where in the cell are chromosomes found?



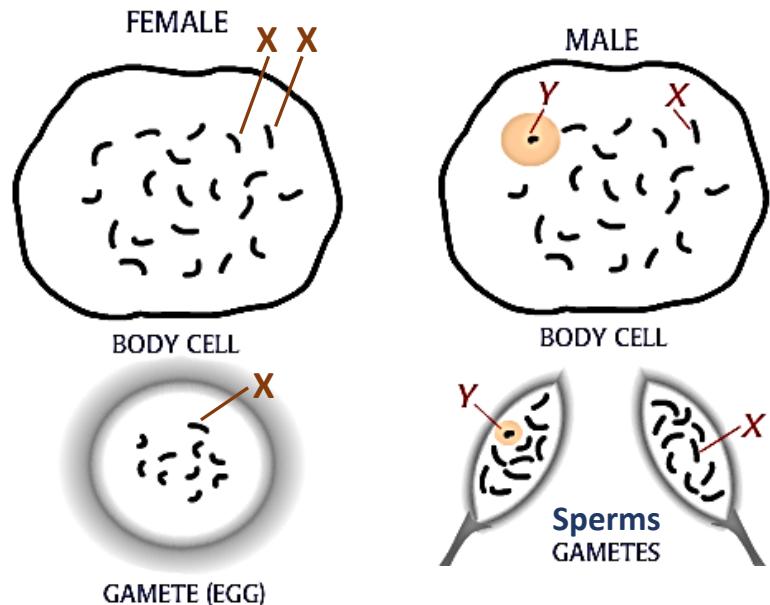
**Sex cells have one set of chromosomes;  
body cells have two sets.**



**1902:** Sutton & Boveri observed that meiosis halves the set of chromosome and randomly assorts homologous chromosomes into sex cells. The full chromosome number is restored when sperm and egg unite. This exactly mirrored the behavior of genes as deduced by Mendel.

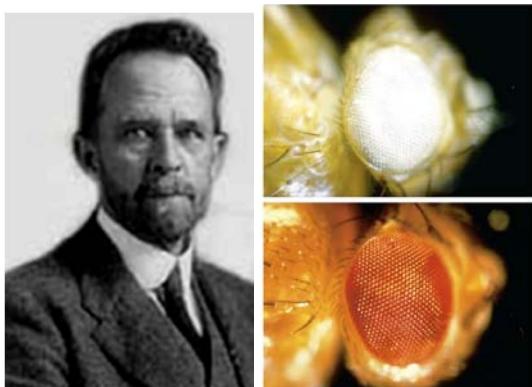
<http://www.dnafdb.org/8/>

**Specialized chromosomes determine sex**



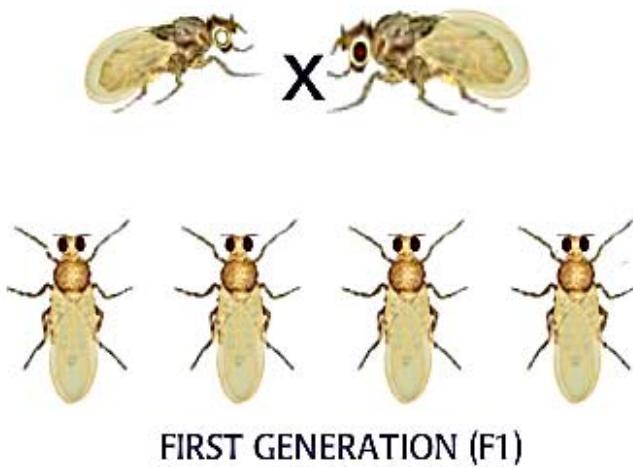
<http://www.dnafdb.org/8/>

**1905:** Stevens and Wilson observed an odd pair among the homologous chromosomes. One chromosome (X) was much bigger than the other (Y). In human beings, this mismatched pair of one X and one Y chromosome is seen exclusively in male cells. A matched pair of X chromosomes is found in female cells. Thus, XX chromosomes determine femaleness, and XY chromosomes determine maleness. Females produce only eggs with X chromosomes; males produce sperms with an X or a Y chromosome.



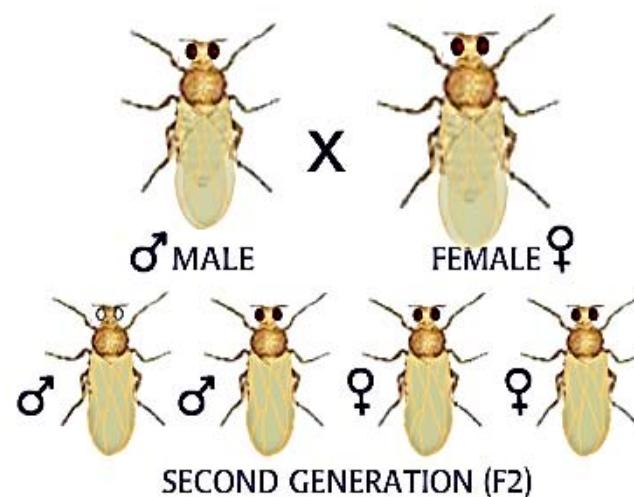
**1911: Thomas H. Morgan formulated Chromosomal Theory of Inheritance and Sex-linked inheritance.**

**Mendel's hereditary factors were found on chromosomes but  
Mendel's law is not always true.**

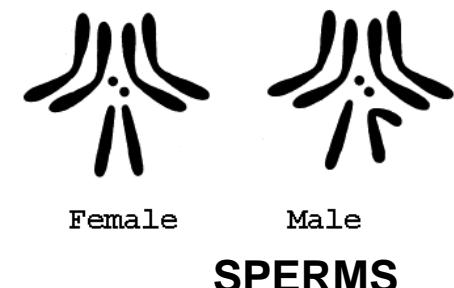


A cross between the mutant male and a red-eyed female produced only red-eyed offspring.

<http://www.dnaftb.org/10/>



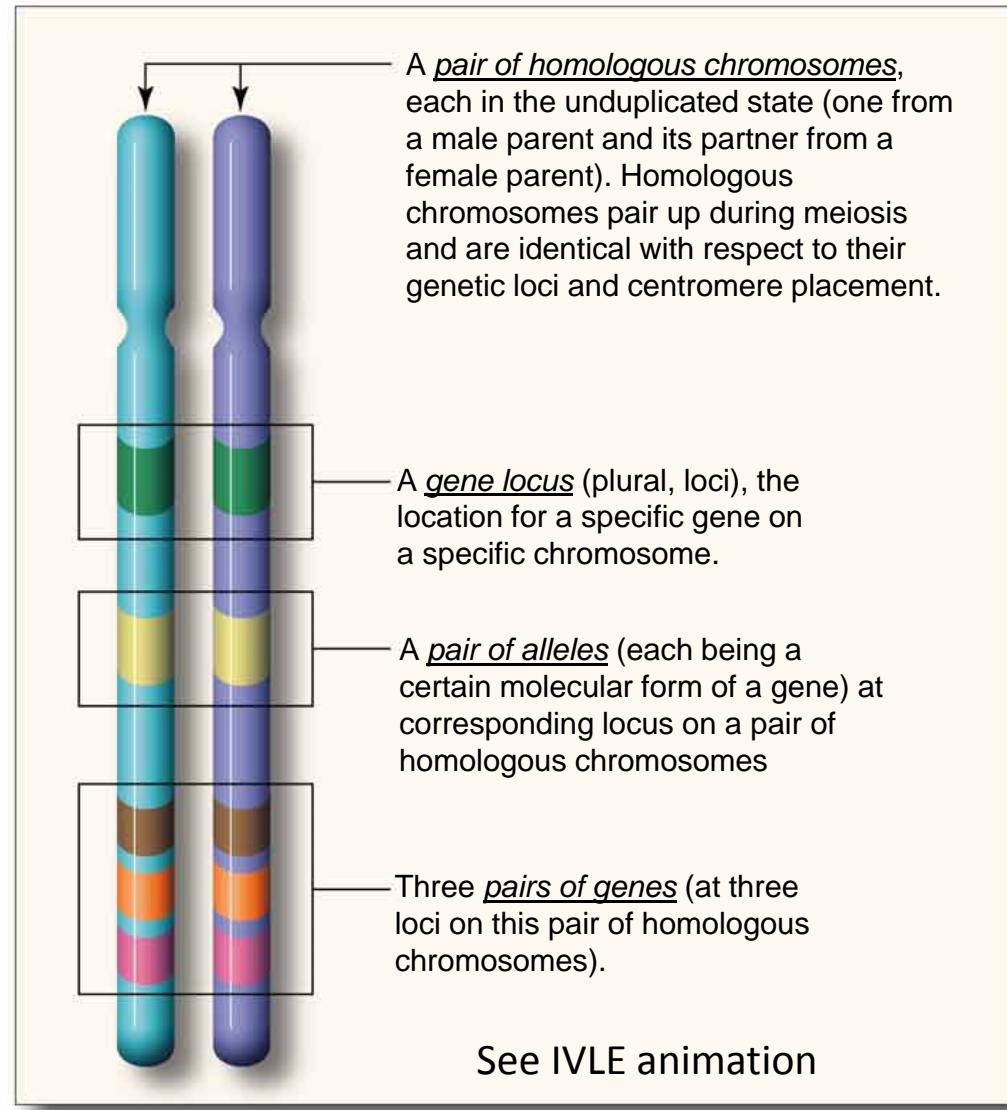
White-eyed mutants reappeared in the following generation — the classic pattern of a recessive trait. However, the white-eyed trait was seen exclusively in males of the second generation.



<b>E</b>	$X^R$	$Y$
<b>G</b>	$X^R$	$X^R Y$
<b>S</b>	$X^W$	$X^W Y$

They concluded that white-eyed is a sex-linked recessive trait. The gene for eye color must be physically located on the X chromosome.

# Mendelian Hereditary Factors On Morgan's Chromosomal Theory



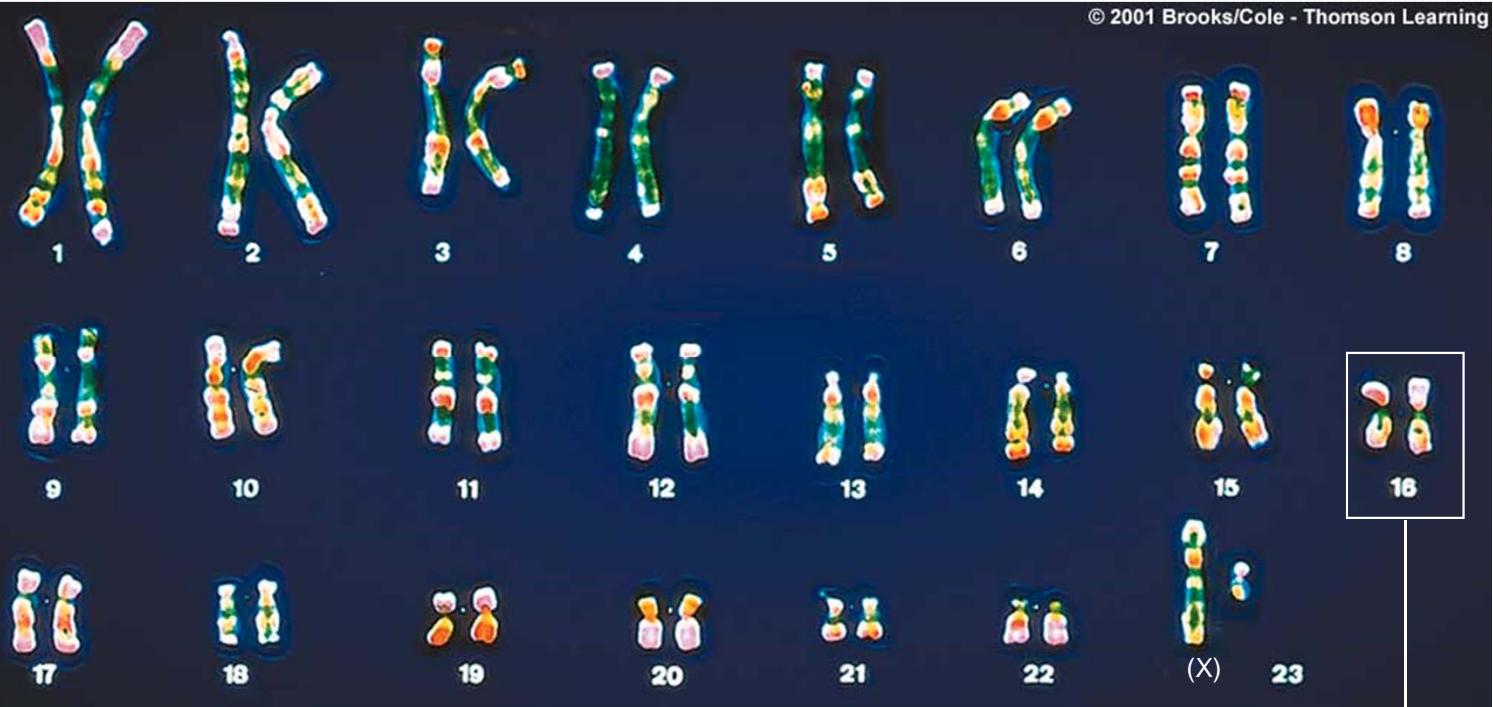


TABLE 2.1

The Haploid Number of Chromosomes for a Variety of Organisms

Common Name	Scientific Name	Haploid Number
Black bread mold	<i>Aspergillus nidulans</i>	8
Broad bean	<i>Vicia faba</i>	6
Cat	<i>Felis domesticus</i>	19
Cattle	<i>Bos taurus</i>	30
Chicken	<i>Gallus domesticus</i>	39
Chimpanzee	<i>Pan troglodytes</i>	24
Corn	<i>Zea mays</i>	10
Cotton	<i>Gossypium hirsutum</i>	26
Dog	<i>Canis familiaris</i>	39
Evening primrose	<i>Oenothera biennis</i>	7
Frog	<i>Rana pipiens</i>	13
Fruit fly	<i>Drosophila melanogaster</i>	4
Garden onion	<i>Allium cepa</i>	8
Garden pea	<i>Pisum sativum</i>	7
Grasshopper	<i>Melanoplus differentialis</i>	12
Green alga	<i>Chlamydomonas reinhardtii</i>	18
Horse	<i>Equus caballus</i>	32
House fly	<i>Musca domestica</i>	6
House mouse	<i>Mus musculus</i>	20
Human	<i>Homo sapiens</i>	23
Jimson weed	<i>Datura stramonium</i>	12
Mosquito	<i>Culex pipiens</i>	3
Mustard plant	<i>Arabidopsis thaliana</i>	5
Pink bread mold	<i>Neurospora crassa</i>	7
Potato	<i>Solanum tuberosum</i>	24
Rhesus monkey	<i>Macaca mulatta</i>	21
Roundworm	<i>Caenorhabditis elegans</i>	6
Silkworm	<i>Bombyx mori</i>	28
Slime mold	<i>Dictyostelium discoideum</i>	7
Snapdragon	<i>Antirrhinum majus</i>	8
Tobacco	<i>Nicotiana tabacum</i>	24
Tomato	<i>Lycopersicon esculentum</i>	12
Water fly	<i>Nymphaea alba</i>	80
Wheat	<i>Triticum aestivum</i>	21
Yeast	<i>Saccharomyces cerevisiae</i>	16
Zebrafish	<i>Danio rerio</i>	25

22 pairs of autosomes (non-sex chromosomes)

1 pair of sex chromosomes

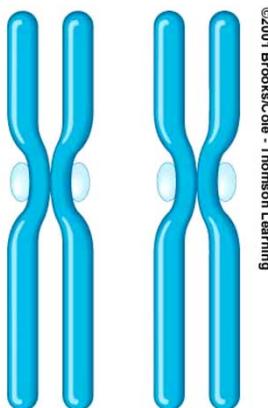
XX = female

XY = male

An organized complete set of chromosomes of a cell or individual that is usually visualized during certain phase of cell division is referred to as karyotype.

Learn genetics > Heredity & Traits > Make a karyotype

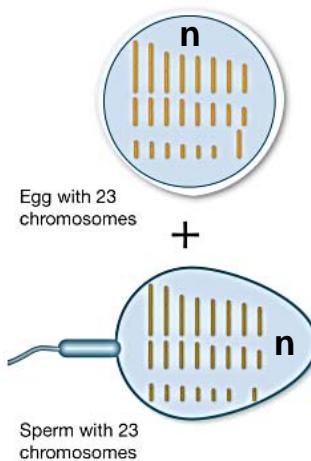
one pair of duplicated chromosomes





# Chromosomal Inheritance in Human Reproduction

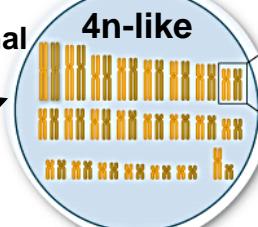
**Fertilization**  
 $n + n = 2n$



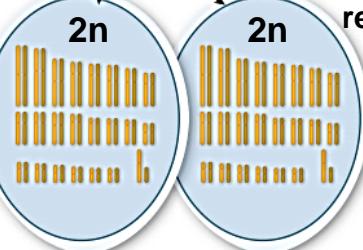
**Chromosomal replication**

**Mitosis**  
 $2n = 2n + 2n$

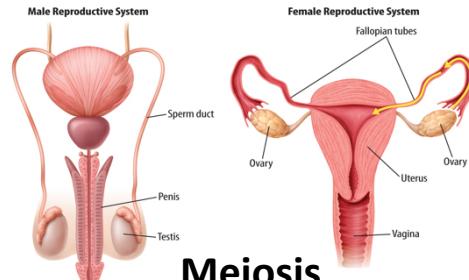
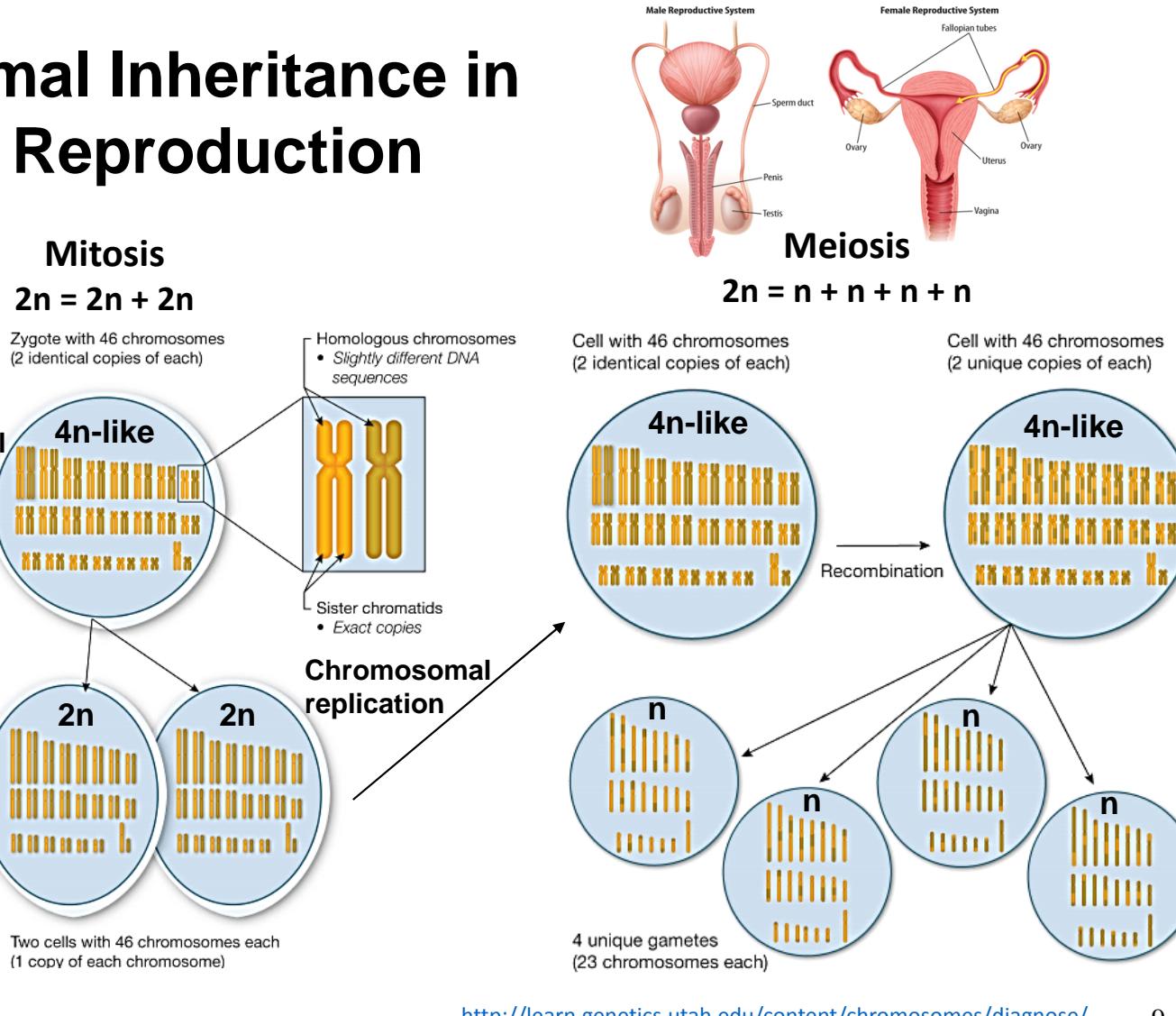
Zygote with 46 chromosomes  
 (2 identical copies of each)



**Chromosomal replication**

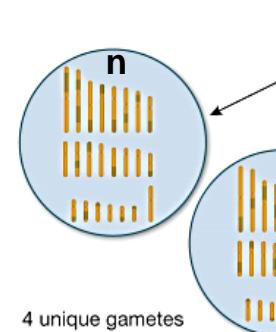
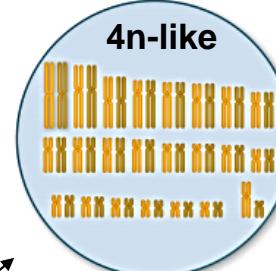


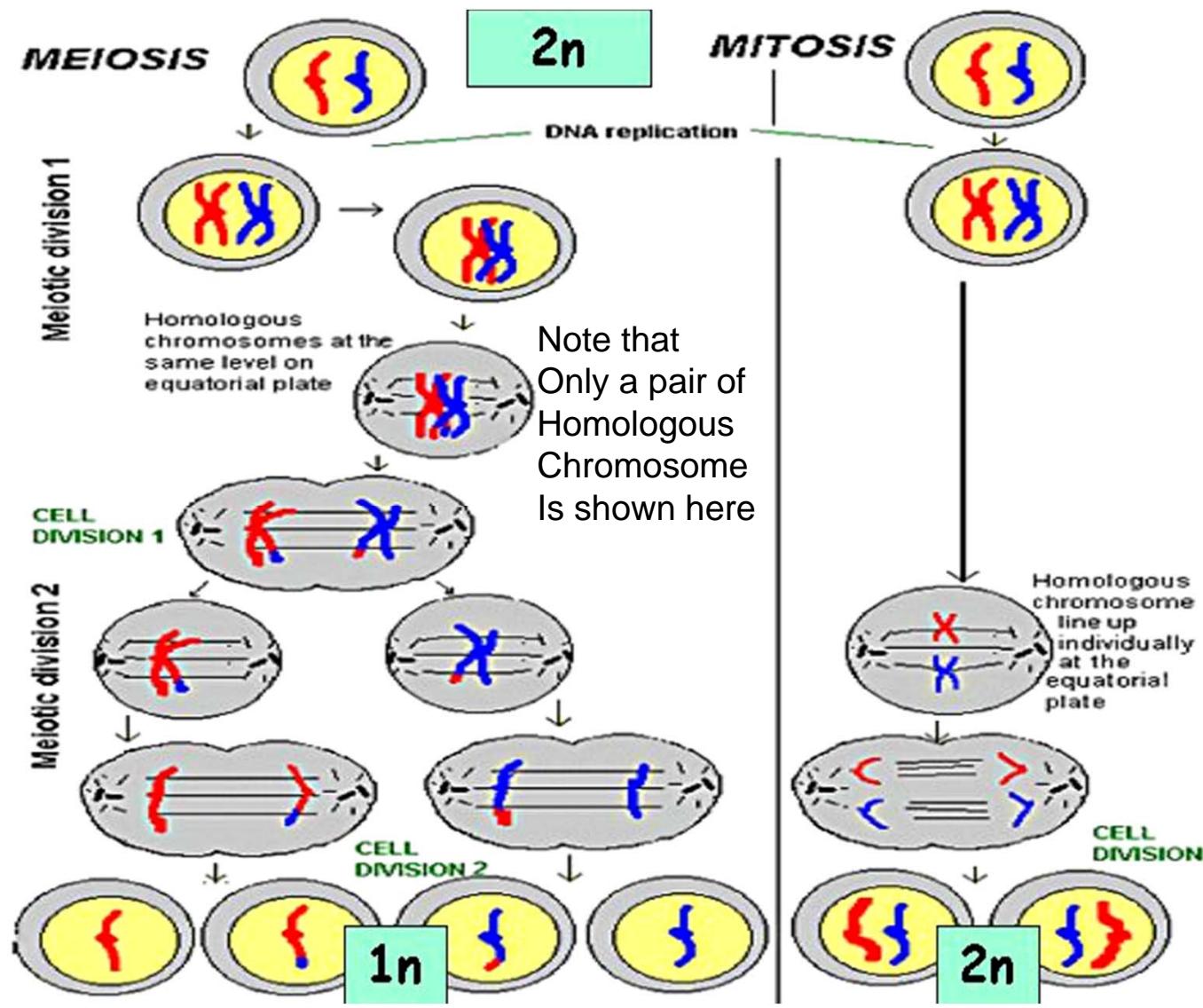
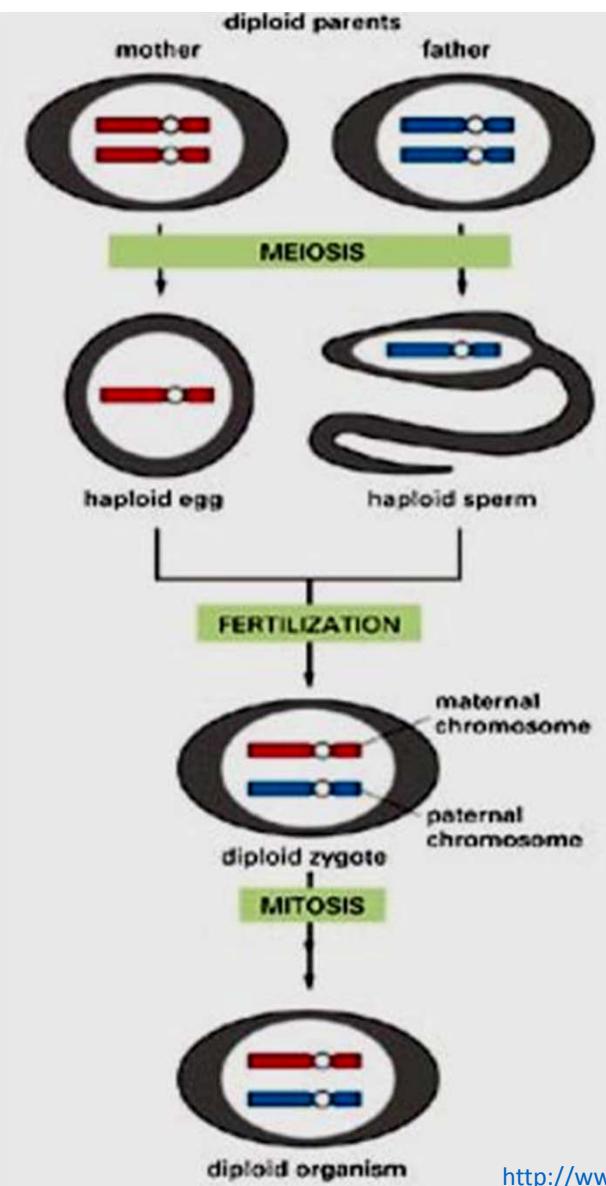
Two cells with 46 chromosomes each  
 (1 copy of each chromosome)



**Meiosis**  
 $2n = n + n + n + n$

Cell with 46 chromosomes  
 (2 identical copies of each)





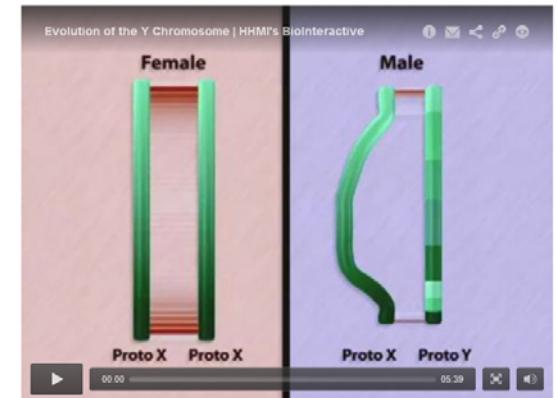
# Sexual reproduction: Meiosis & Homologous Recombination

- Provide genetic variations that can be advantages in changing environments (including host-pathogen arm race) so that the species will not be easily wipe out.
- Reduced (50%) the chance of passing deleterious mutation to offspring (unlike asexual reproduction which passes 100% to offspring; how is this compared to selective inbreeding?)
- Sexual reproduction requires homologous chromosomes that can serve as a back-up copy of genes just in case one copy experience a deleterious mutation. What about the Y-chromosome that has no homologous chromosome? Would it deteriorate over time in the population?

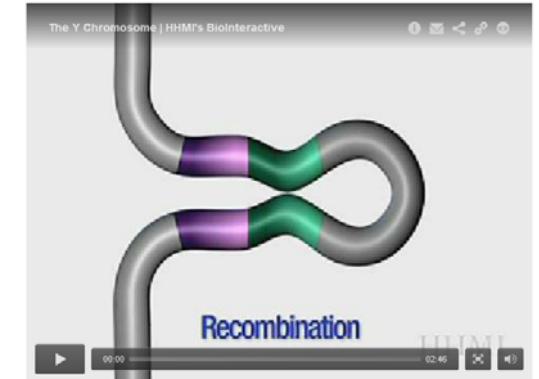
<http://www.hhmi.org/biointeractive/evolution-y-chromosome>

<http://www.hhmi.org/biointeractive/y-chromosome>

## Evolution of the Y Chromosome



## The Y Chromosome



# Heredity & Society

D Tour of the Basics

D What is Heredity?



**WHAT IS HEREDITY?**

Why do children look like their parents?  
Why do brothers and sisters resemble each other?  
This is because we "inherit" traits from our parents.  
The passing of traits from parents to child is the basis of heredity.

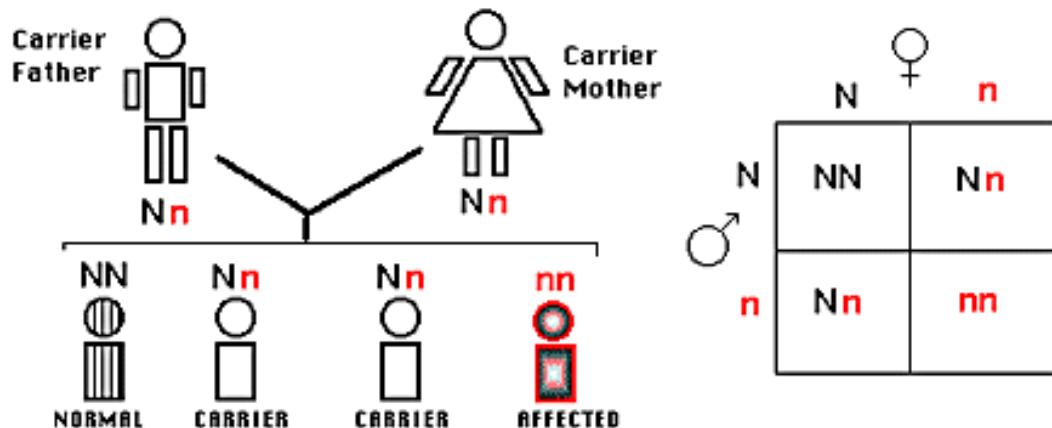
Previous | Next

<http://learn.genetics.utah.edu/content/inheritance/intro/>

**Freedom was conditioned by man's physical body, heredity, and environment.**

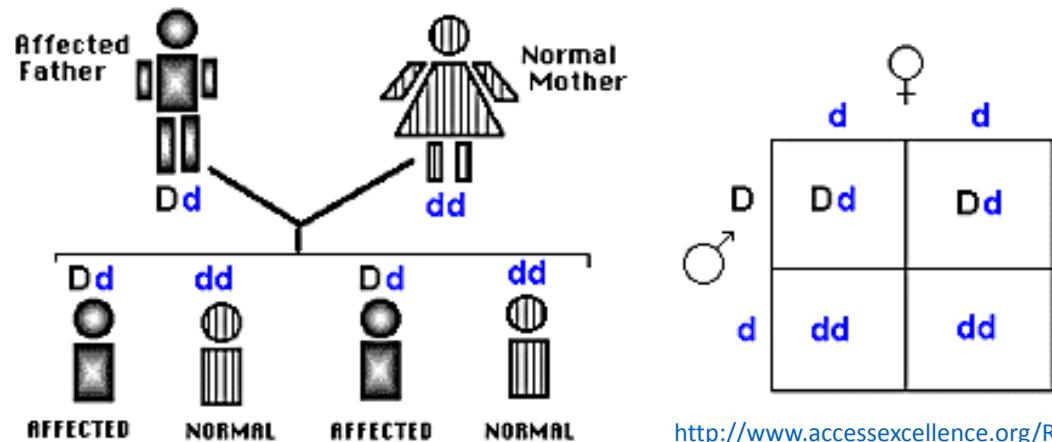
**- Kenneth Scott Latourette (Historian)**

## Recessive inheritance



The term "recessive allele" refers to an allele that causes a phenotype (visible or detectable characteristic) that is only seen in homozygous genotype (an organism that has two copies of the same allele; recessive trait) and never in a heterozygous genotype.

## Dominant inheritance



Dominant trait refers to a genetic feature that hides the recessive trait in the phenotype of an individual. A dominant allele causes the phenotype to be seen in a heterozygous (Aa) genotype.

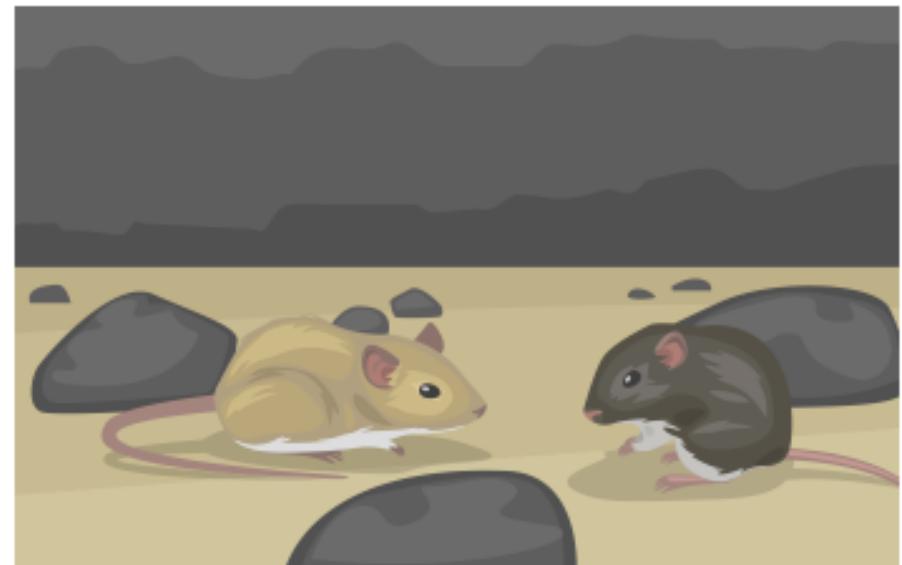
## Dominant phenotypes are not always more common than recessive phenotypes

Eye color is influenced mainly by two major genes, with minor contributions from few others. People with light eyes tend to carry recessive alleles of the major genes; people with dark eyes tend to carry dominant alleles. In Scandinavia, most people have light eyes—the recessive alleles of these genes are much more common here than the dominant ones.



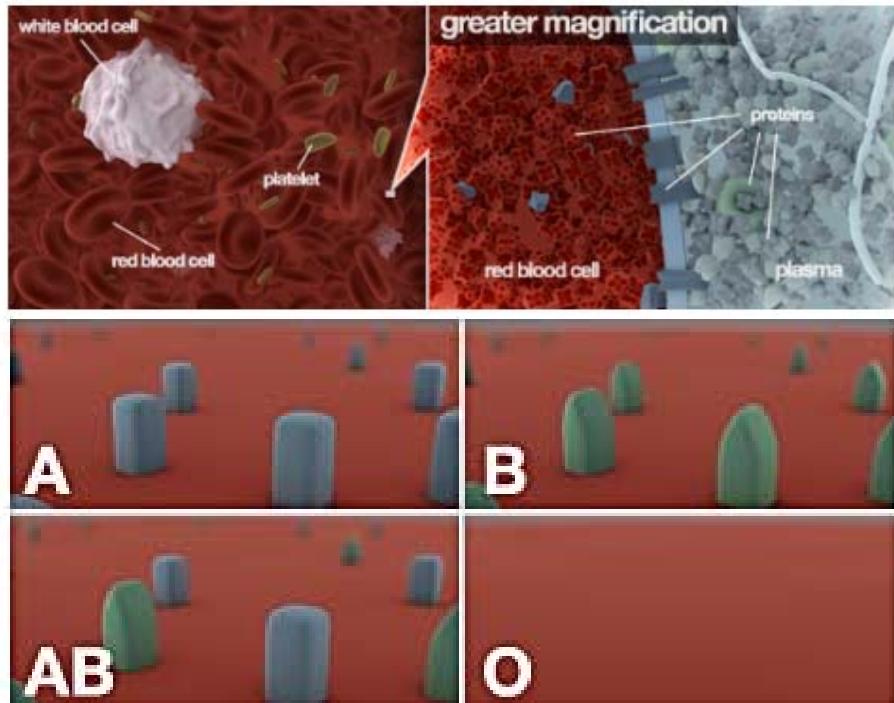
## Dominant alleles are not better than recessive alleles

The dark-fur allele is dominant, and the light-fur allele is recessive. When mice live in a habitat filled with dark rocks, dark fur is “better” because it makes the mice less visible to predators. But when mice live in a habitat filled with light rocks and sand, light fur is “better.” It’s the environment that matters, not whether the allele is dominant or recessive.



# Genes & Blood Groups

The ABO gene is found on chromosome 9

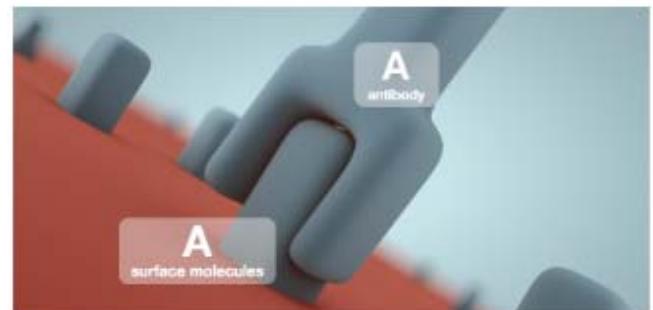


father	mother		
	A	B	O
A	AA	AB	AO
B	BA	BB	BO
O	OA	OB	OO

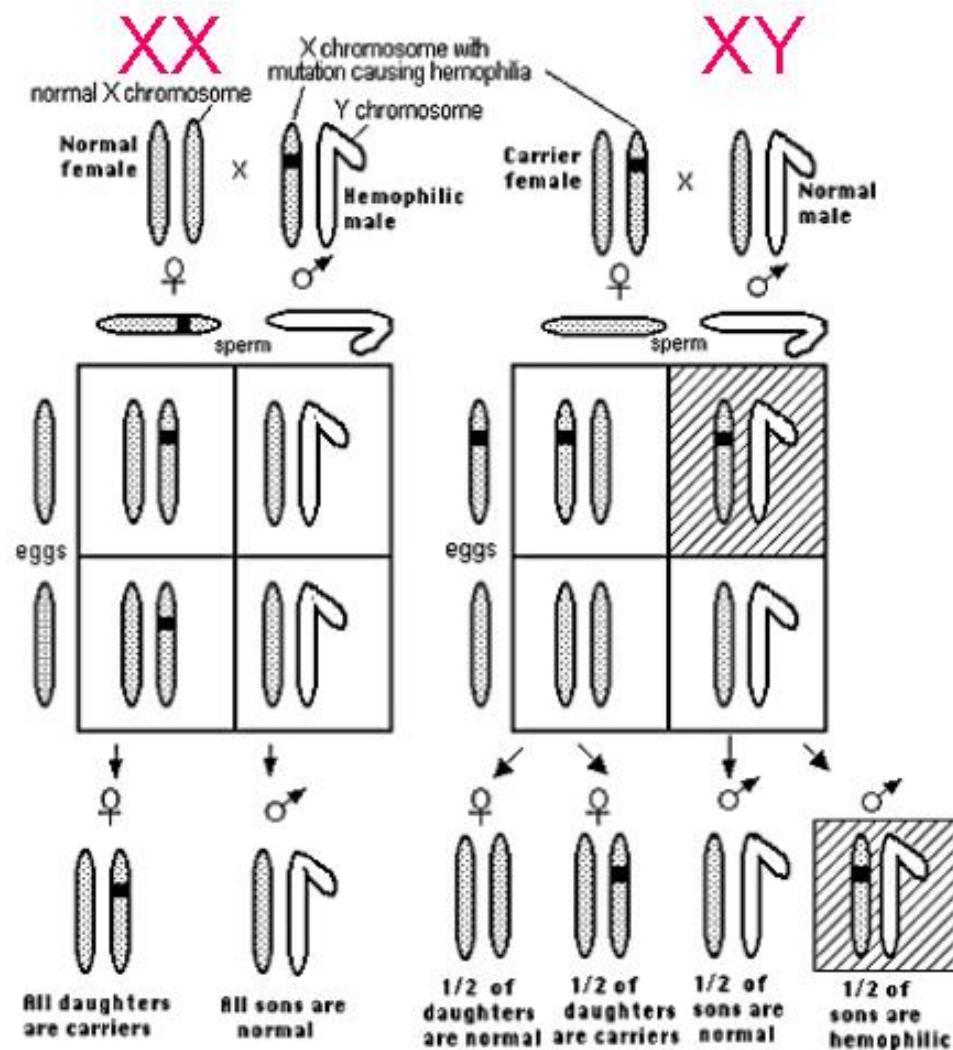
alleles    blood type

**A+A = A**  
**A+O = A**  
**A+B = AB**  
**B+B = B**  
**B+O = B**  
**O+O = O**

## When Blood Types Mix



blood type	red blood cell surface molecules	plasma antibodies
type A	A only	B only
type B	B only	A only
type AB	A & B	neither
type O	neither	both



## Inheritance of Hemophilia

<http://www.accessexcellence.org/RC/VL/GG/x-linked.php>

## Human X-linked Inheritance

### Example of traits

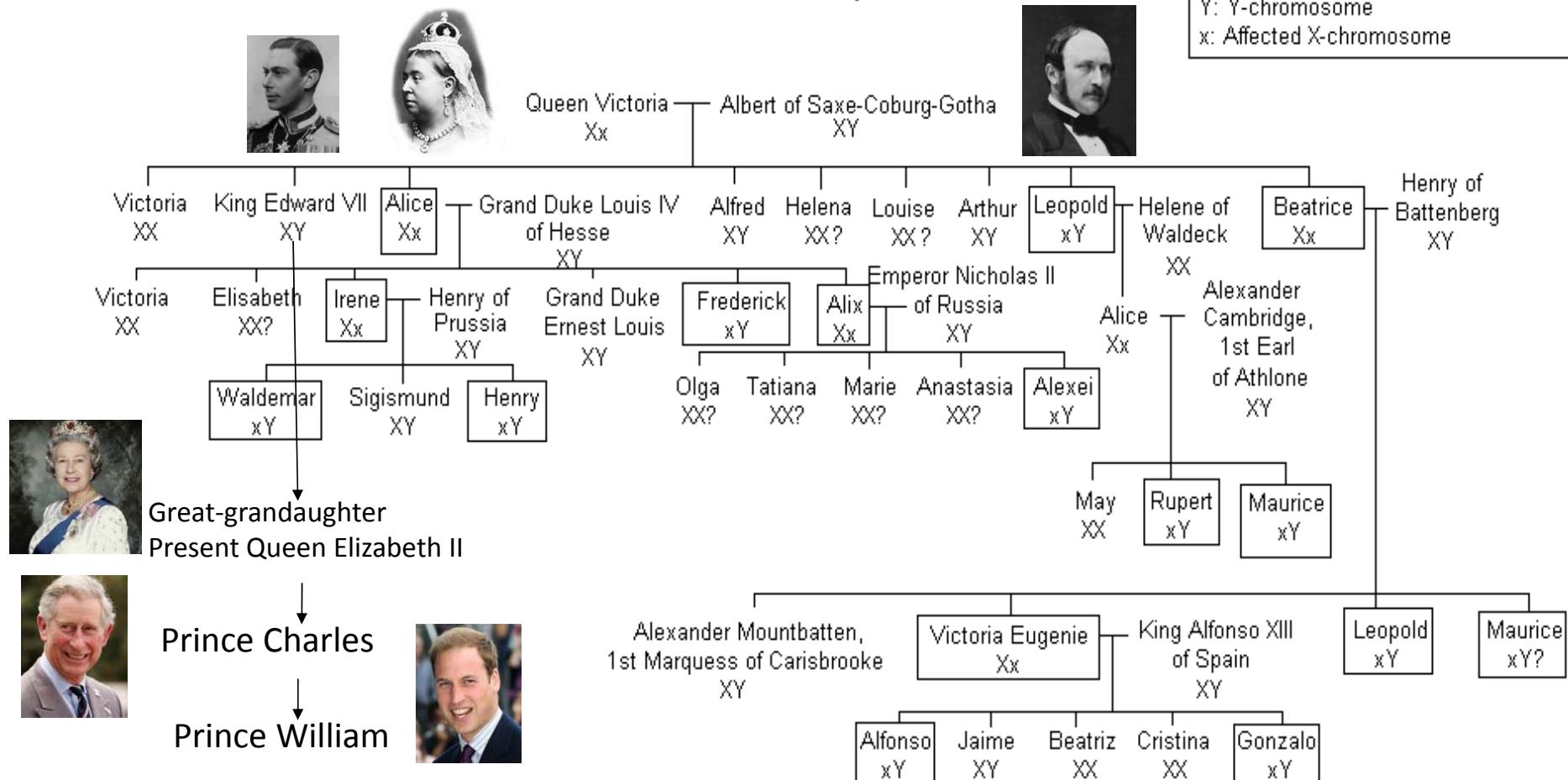
**Red-green Color Blindness:** Absence of (functional) red or green photoreceptor. Insensitivity to red or green light.

**Hemophilia A or B:** Absence of (functional) clotting factor VIII or IX. Blood clotting disorder. Frequent bleeding episodes.

**Duchenne Muscular Dystrophy:** Absence of (functional) protein dystrophin. Progressive life-shortening disorder characterized by muscle degeneration and weakness.

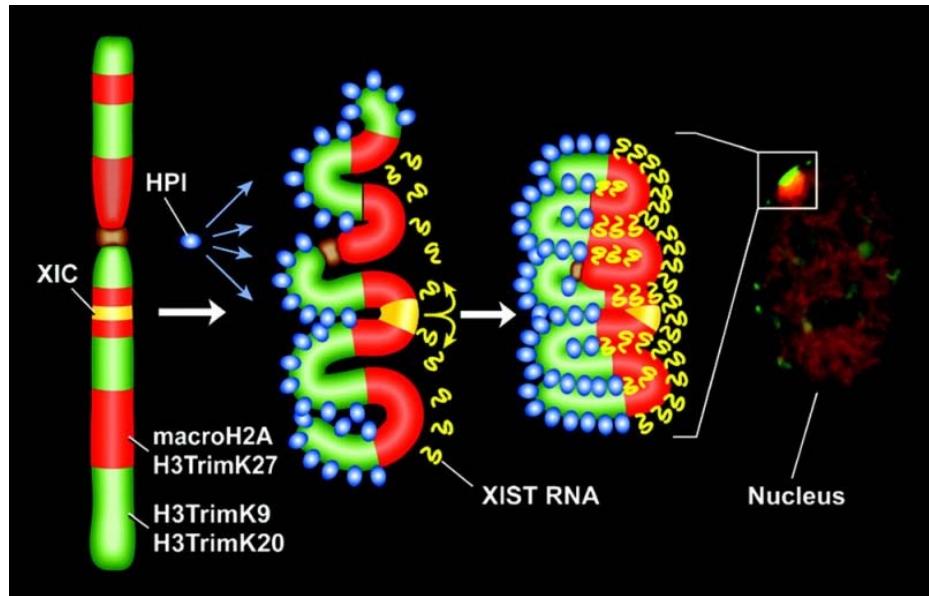
**Complete Androgen Insensitivity Syndrome (CAIS):** Absence of (functional) androgen receptor. XY-female.

# The British Haemophilia Line



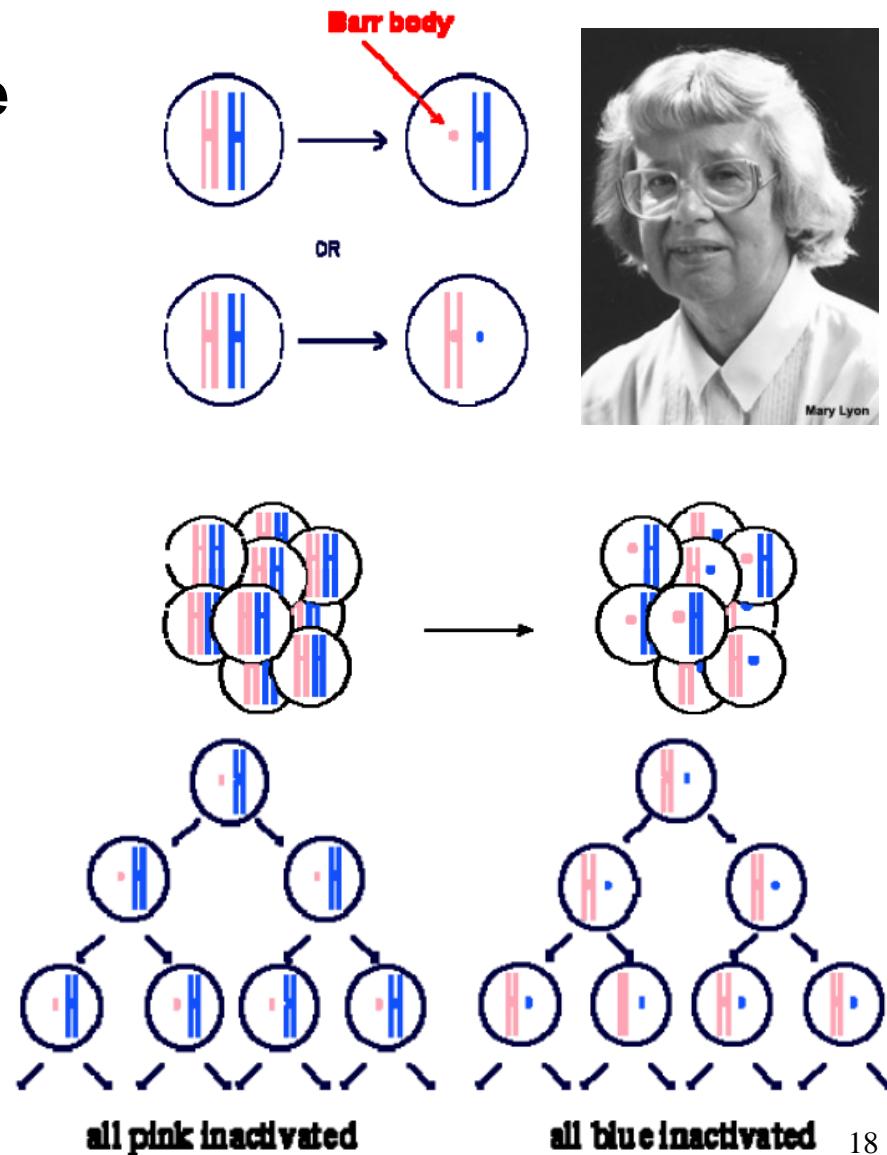
[http://en.wikipedia.org/wiki/File:Haemophilia\\_family\\_tree.GIF](http://en.wikipedia.org/wiki/File:Haemophilia_family_tree.GIF)

## X-inactivation to control gene dosage

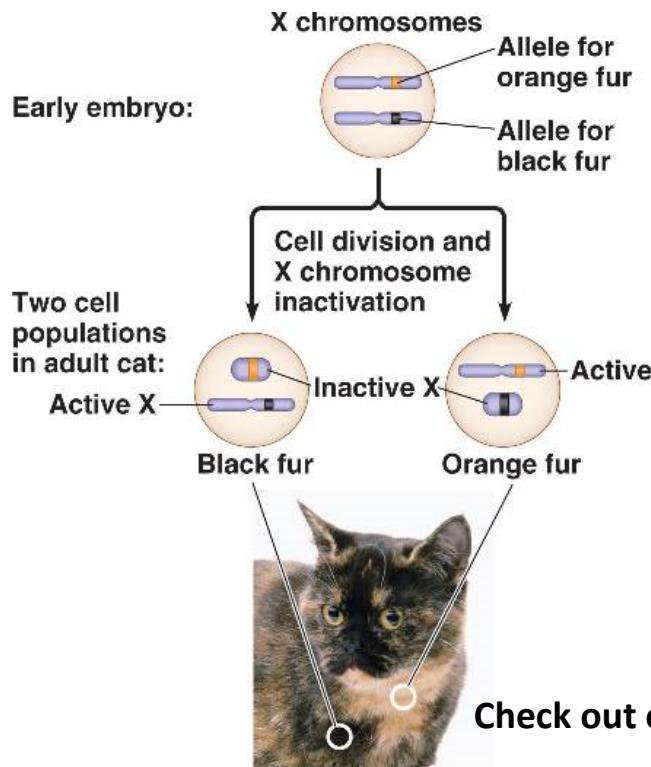


<http://www.ncbi.nlm.nih.gov/pubmed/15574503>

The X-inactivation center (XIC) gene on one of the X chromosome produces X-inactivated specific transcript (XIST) RNA and bind themselves on the X-chromosome. XIST initiate X inactivation and it is the methylation (aided by HPI) of that inactive (condensed) the X chromosome . The other x chromosome remains active. X-inactivation occurs randomly during embryonic development (12-16 days).

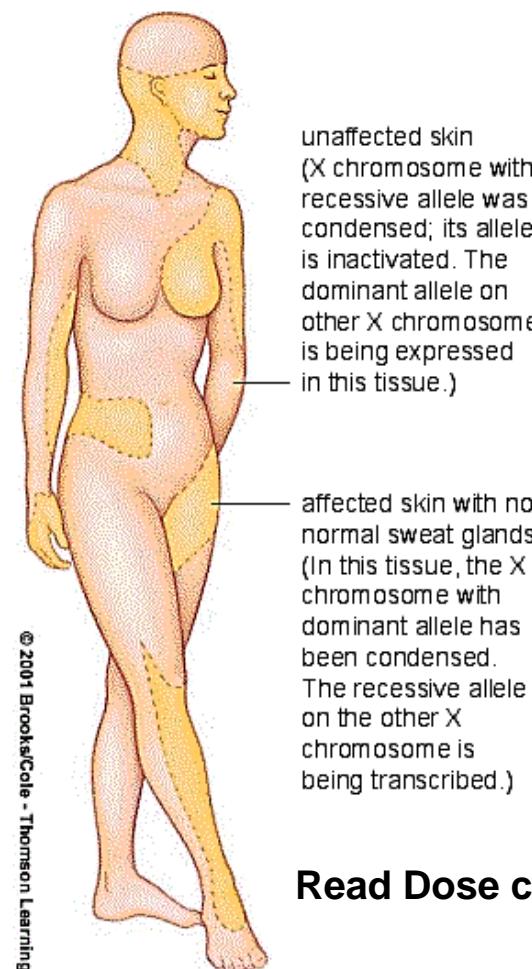


# Female mammals including humans are mosaic due to X-inactivation



<http://bio1151.nicerweb.com/Locked/media/ch15/X-inactivation.html>

A calico cat (is always female), where the random distribution of orange and black patches demonstrates X-inactivation.



Human anhydrotic dysplasia is a disease that results in the absence of sweat glands. Normal females are XX, heterozygous females are XX' and have patches of skin with sweat glands and patches of skin without sweat glands. Females that are X'X' do not have sweat glands.

**Read Dose compensation & X-inactivation**

# Chromosomal Dynamics: Normal vs. Aberrations

*(abnormalities at the chromosomal level)*

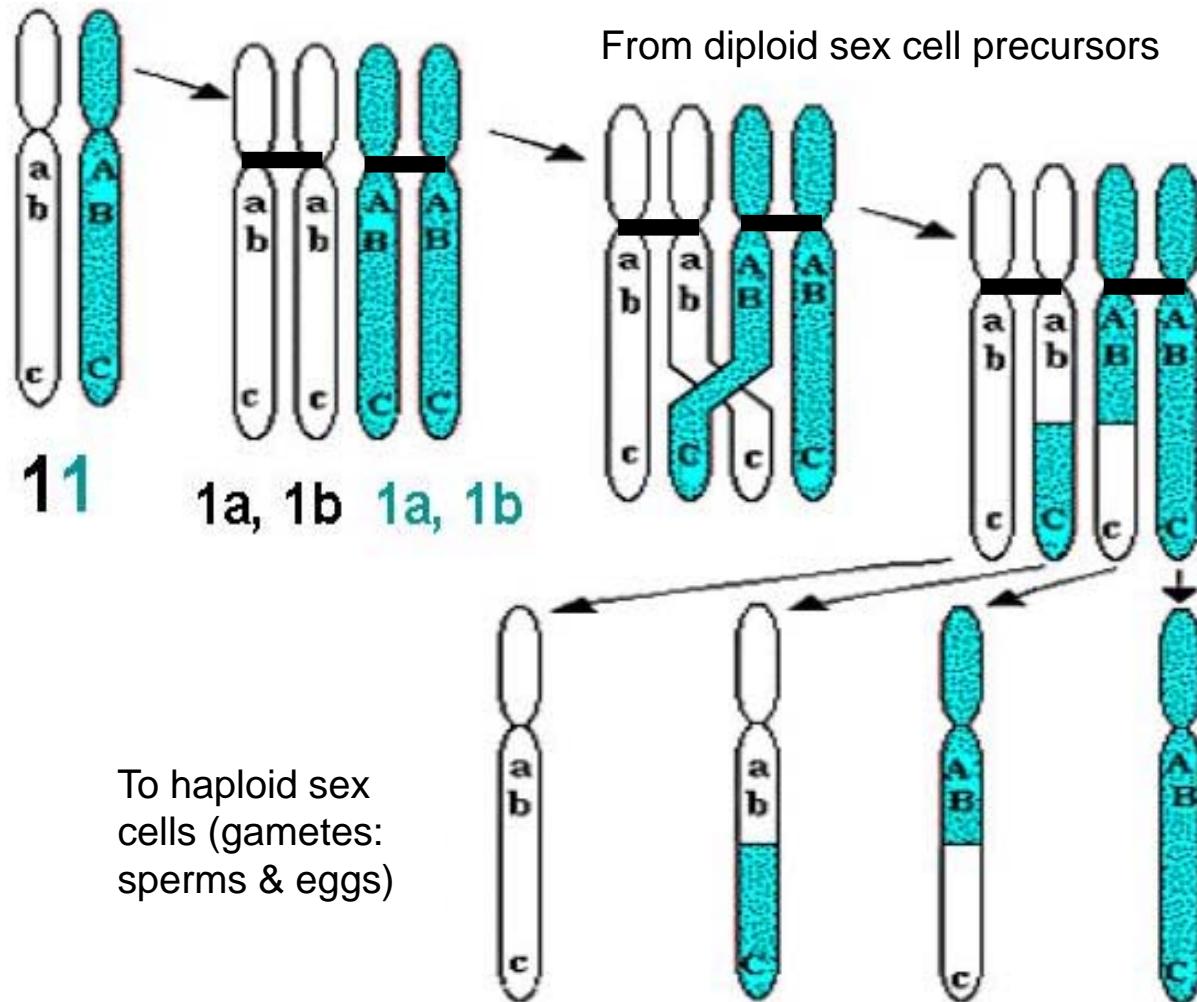
See additional reading material:  
Chromosomal aberration.pdf

Acceptance of one's life has nothing to do with resignation; it does not mean running away from the struggle. On the contrary, it means accepting it as it comes, with all the handicaps of *heredity*, of suffering, of psychological complexes and injustices.

- Paul Tournier (Physician; Author)

# Normal Homologous Recombination

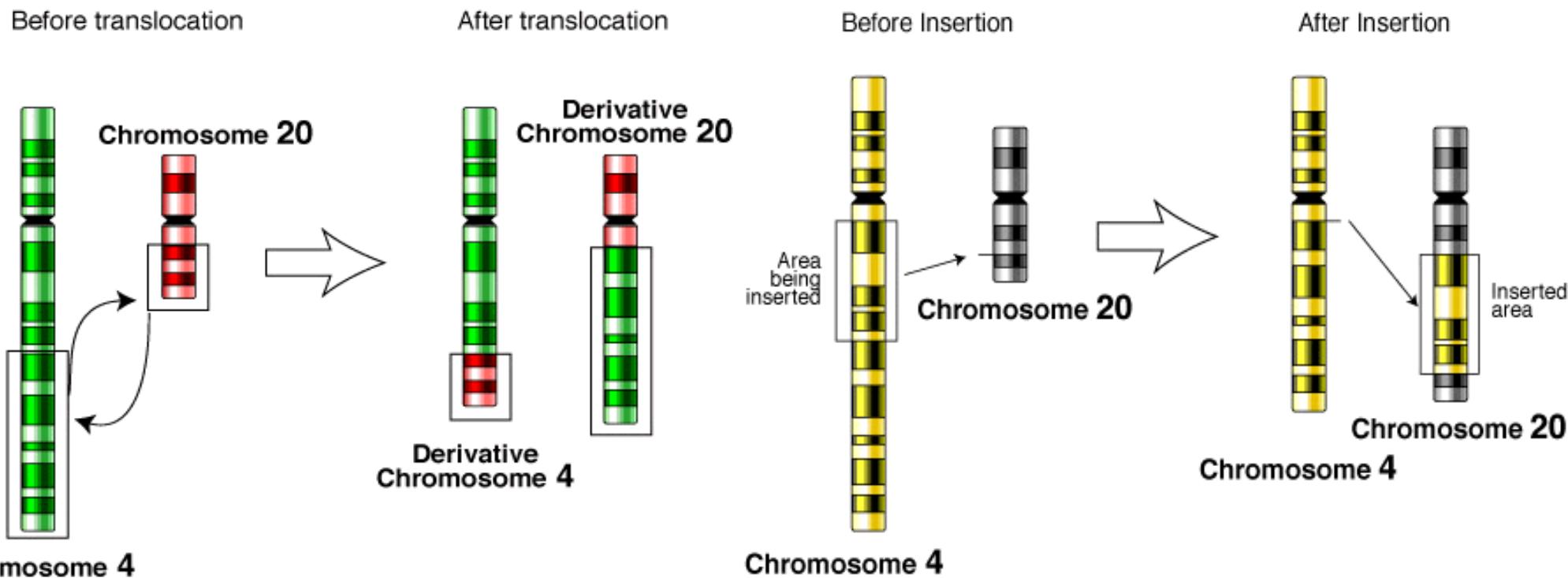
(occurs in testis and ovary)



**Crossing-over and recombination during meiosis**

# Translocation

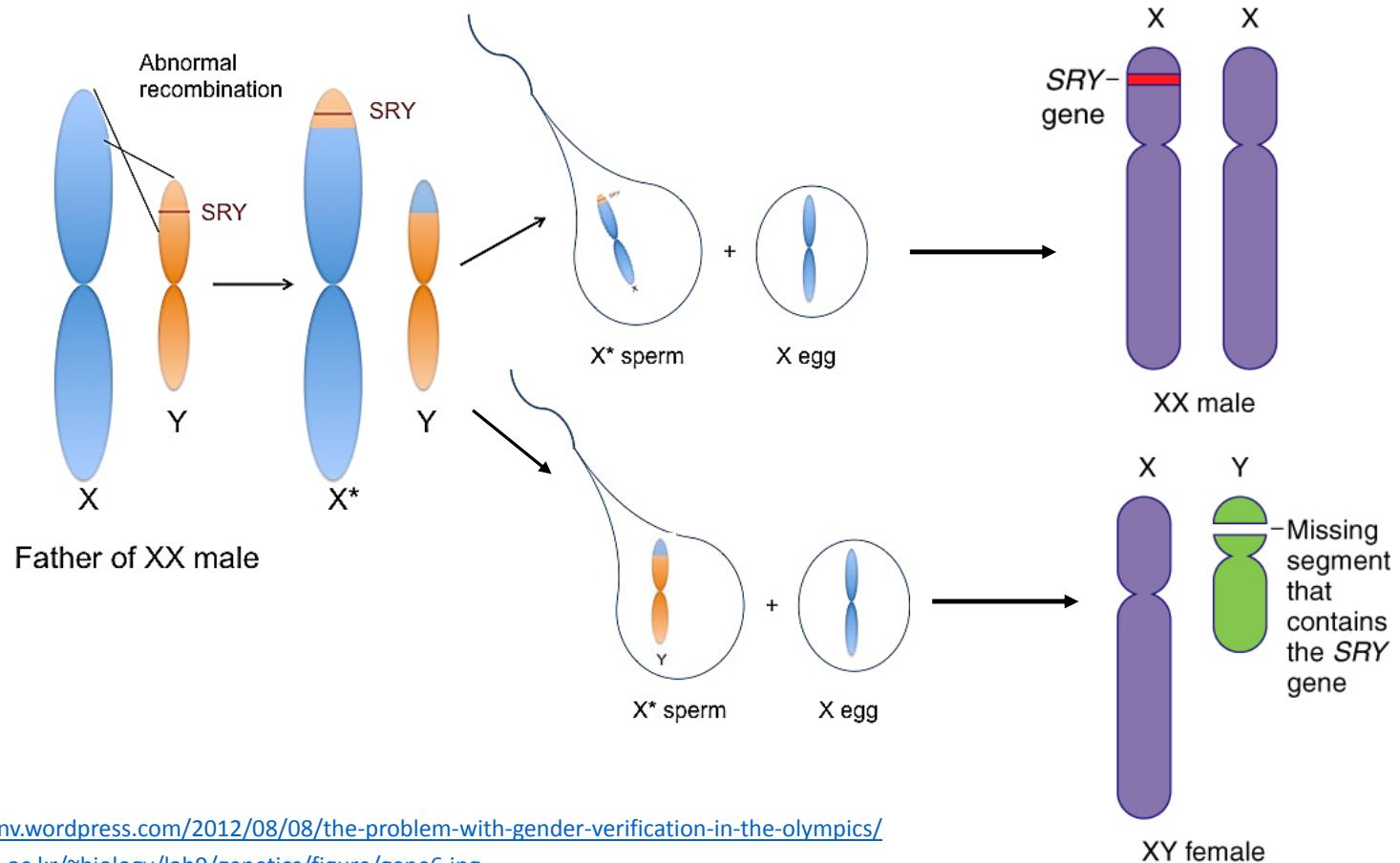
# Insertion



[http://en.wikipedia.org/wiki/Chromosomal\\_translocation](http://en.wikipedia.org/wiki/Chromosomal_translocation)

[http://en.wikipedia.org/wiki/Insertion\\_\(genetics\)](http://en.wikipedia.org/wiki/Insertion_(genetics))

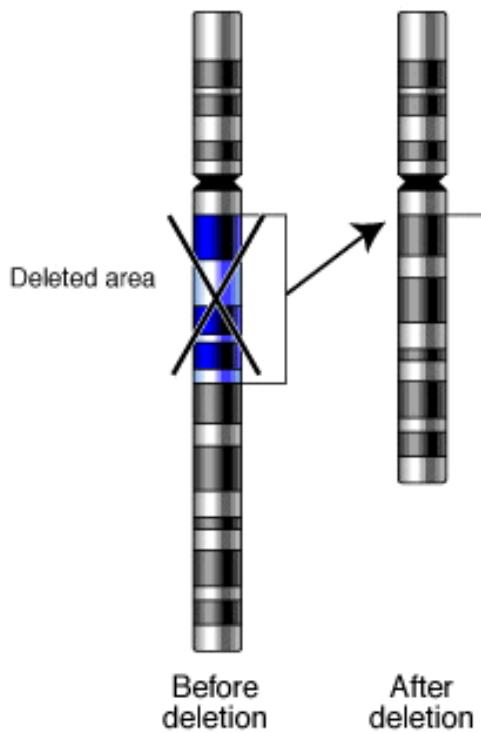
# Translocation of *sry* gene from Y-chromosome to X-chromosome causes XX-male and XY-female



<https://amasianv.wordpress.com/2012/08/08/the-problem-with-gender-verification-in-the-olympics/>

<http://iws.inha.ac.kr/~biology/lab9/genetics/figure/gene6.jpg>

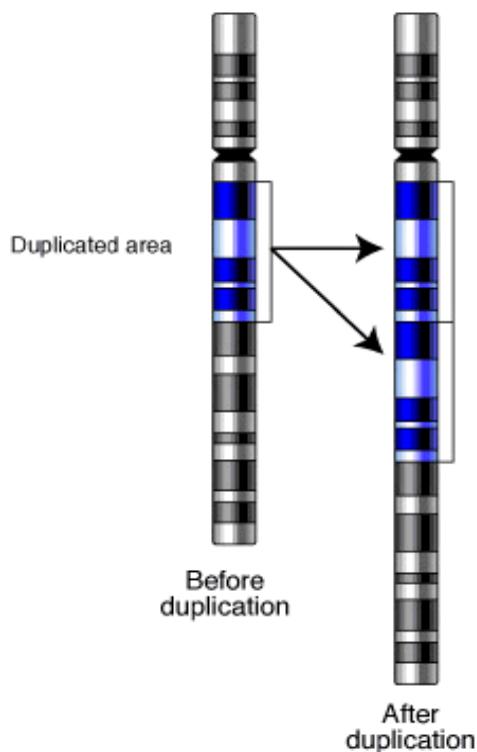
# Deletion



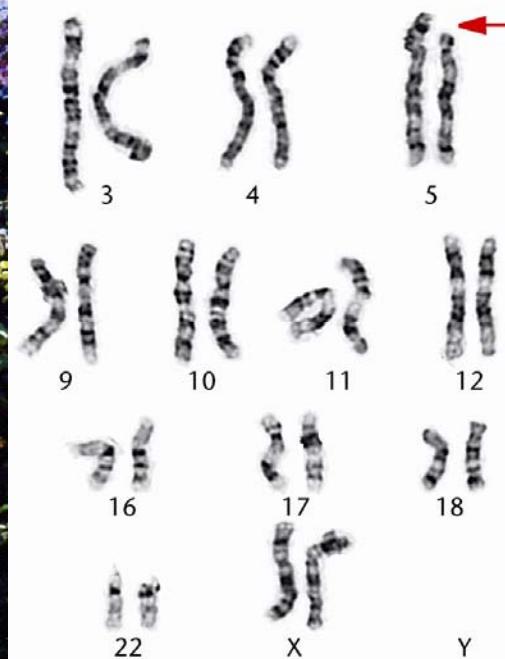
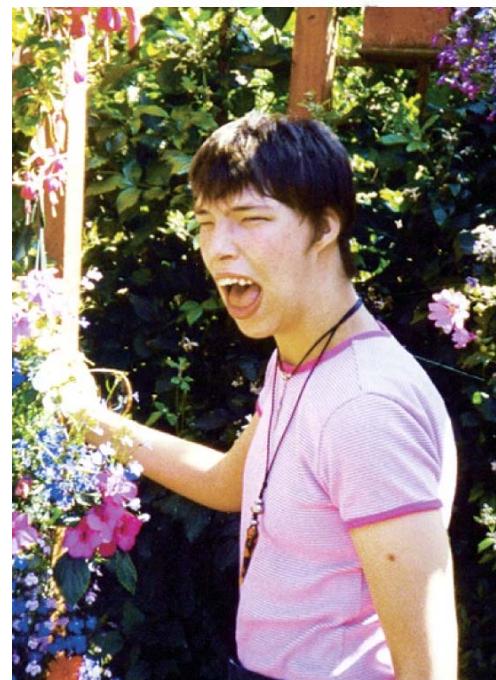
[http://en.wikipedia.org/wiki/Deletion\\_\(genetics\)](http://en.wikipedia.org/wiki/Deletion_(genetics))

<http://en.wikipedia.org/wiki/Genetics#mediaviewer/File:Gene-duplication.png>

# Duplication



A representative karyotype and a photograph of a child exhibiting cri-du-chat syndrome (46,5p-). In the karyotype, the arrow identifies the absence of a small piece of the short arm of one member of the chromosome 5 homologs.



Affected individuals have wide-set eyes, a small head and jaw and are moderately to severely mentally retarded and very short.

<http://learn.genetics.utah.edu/content/disorders/chromosomal/cdc/>

# When chromosome segregation fails.....

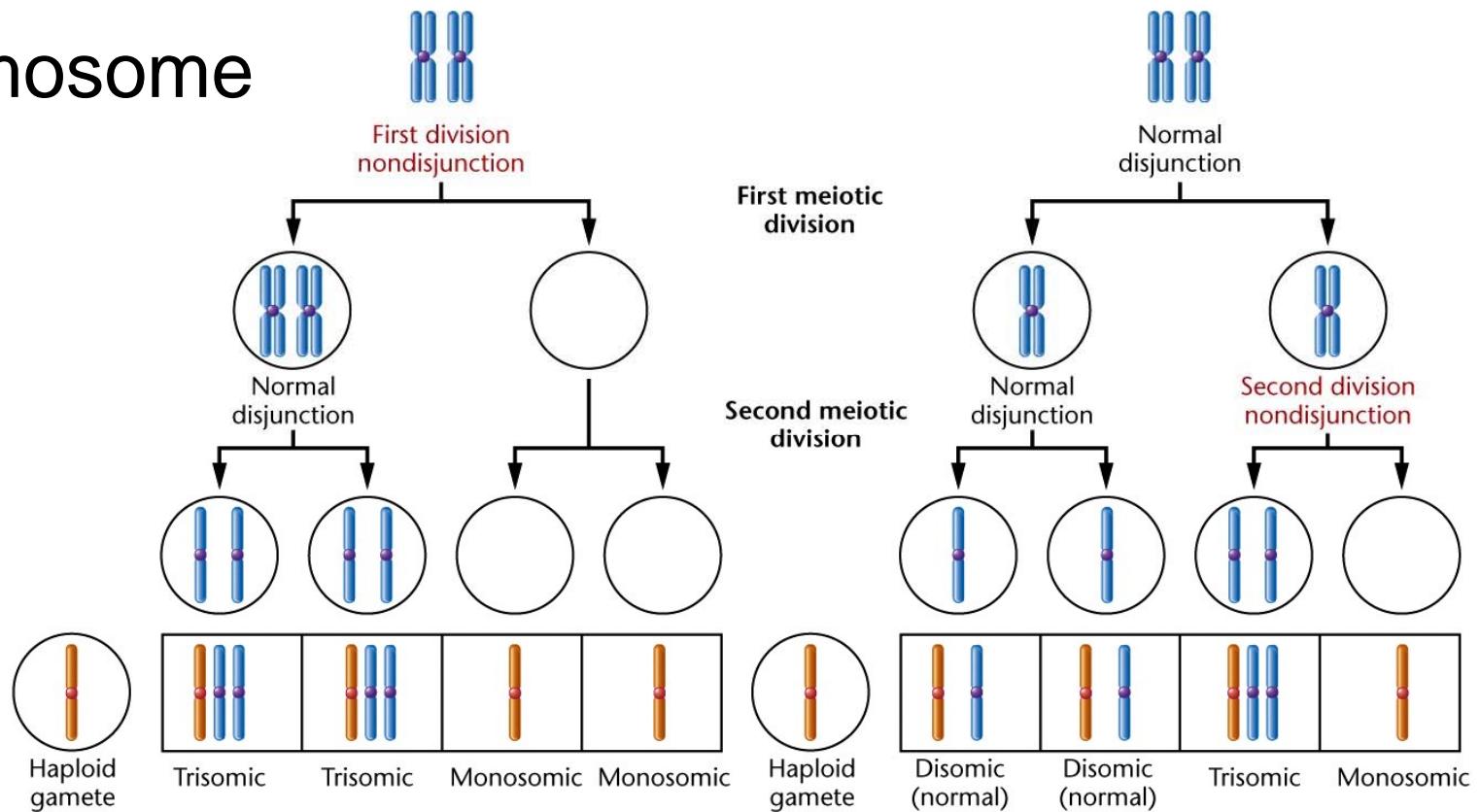
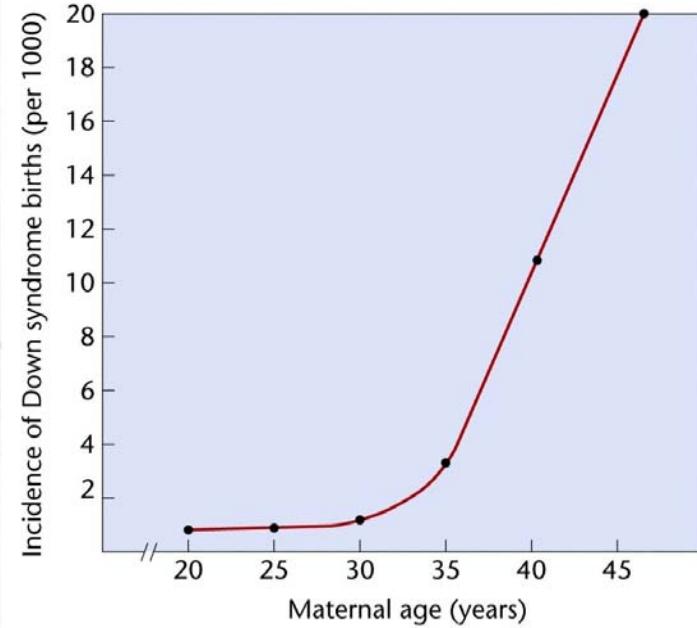
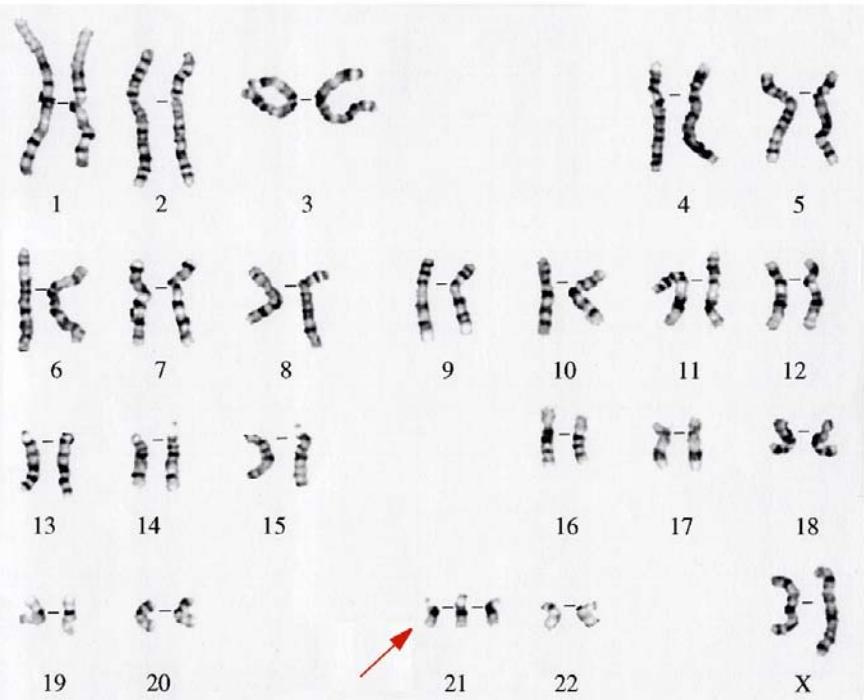


Figure 6-1 Essentials of Genetics, 6/e  
© 2007 Pearson Prentice Hall, Inc.

**Nondisjunction during the first and second meiotic divisions.** In both cases, some of the gametes that are formed either contain two members of a specific chromosome or lack that chromosome. After fertilization by a gamete with a normal haploid content, monosomic, disomic (normal), or trisomic zygotes are produced.

## Down syndrome: Trisomy 21

Three chromosome 21 are present, creating the 47,21+ condition.

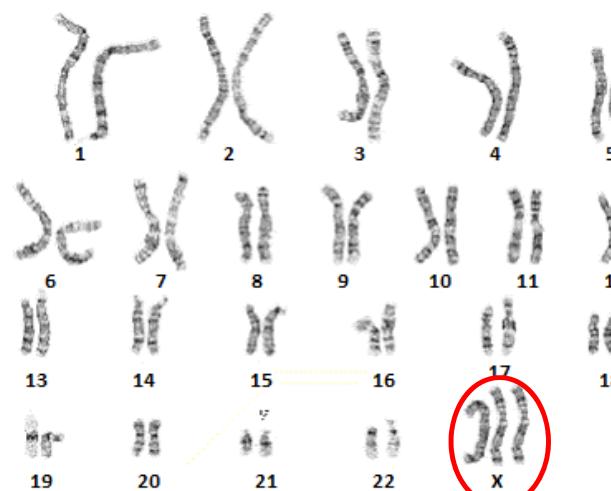


Characteristics include decreased muscle tone, asymmetrical skull, slanting eyes, short, flat faces and mild to moderate physical, psychomotor and mental retardation.

# Turner (XO), XXX & XXY (Klinefelter) Syndromes

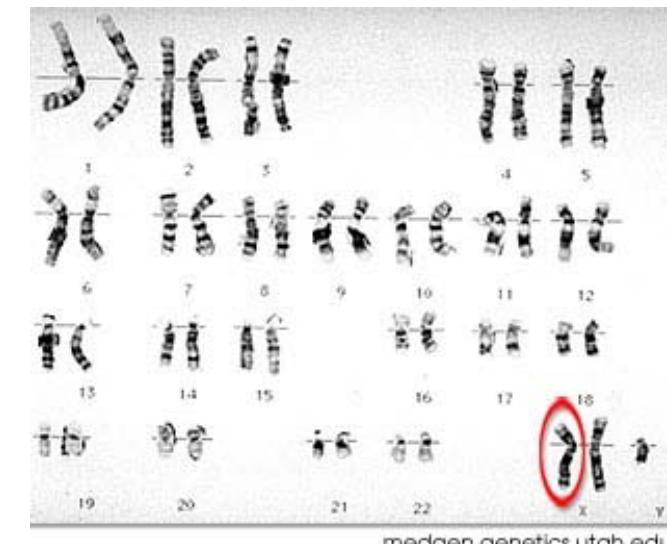


In Turner syndrome, female sexual characteristics are present but underdeveloped and short stature



<http://www.allthingsdiscussed.com/More/Karyotype-profile-of-chromosomes.php>

XXX girls tend to be tall and thin and are often shy. They have a higher incidence of dyslexia.



Men with Klinefelter syndrome are usually sterile, and tend to have longer arms and legs and to be taller than their peers. Speech delay & dyslexic.

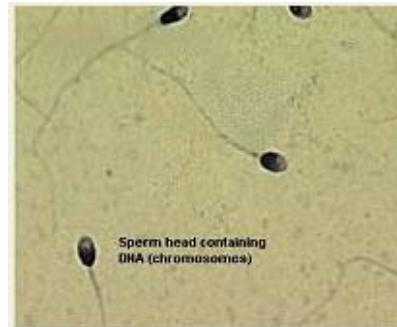
## What about X-inactivation?

# Sperm cells or Embryos Screening to reduced sex-linked diseases: Future direction?

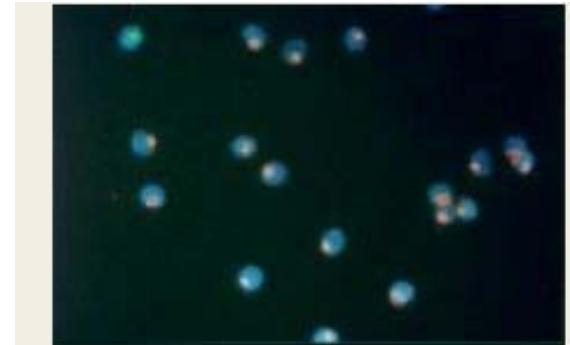
Reduced chance of getting a baby with sex-linked disease can be done by sorting sperm cells carrying X or Y chromosome (depending on the location of the disease allele).

This could be used for selecting sex of baby.  
But is this ethical?

<http://www.in-gender.com/Gender-Selection/MicroSort/faq/Sperm-Sorting.aspx>



Sperm Separation Instrument



FISH Analysis After XSort  
A pink spot identifies female  
sperm and a green spot  
identifies male sperm



FISH Analysis After YSort  
Male sperm contain a green spot  
and female sperm a red spot

# Summary of Key Concepts/Points



## 1. Chromosomal concepts

*Genes are found on specific locations (known as loci) on a chromosome, homologous chromosomes carries same genes (although not necessary same allele); non-homologous chromosomes carry different set of genes.*

## 2. Chromosomal Theory of Inheritance, Meiosis & Mitosis

*Mitosis and meiosis maintain the number of chromosomes in cells and organisms.*

## 3. Heredity & Society

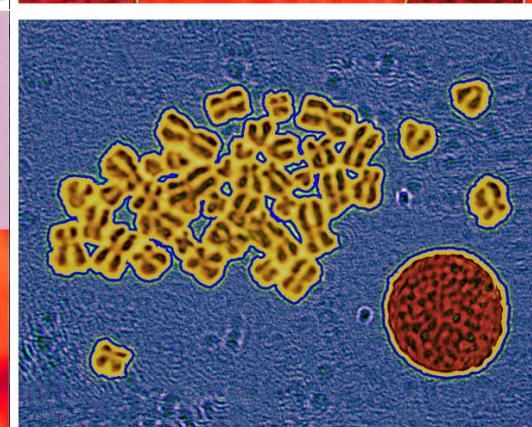
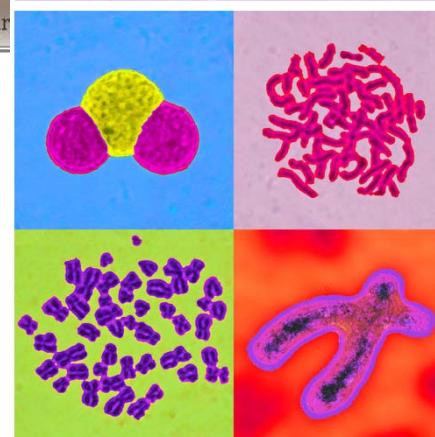
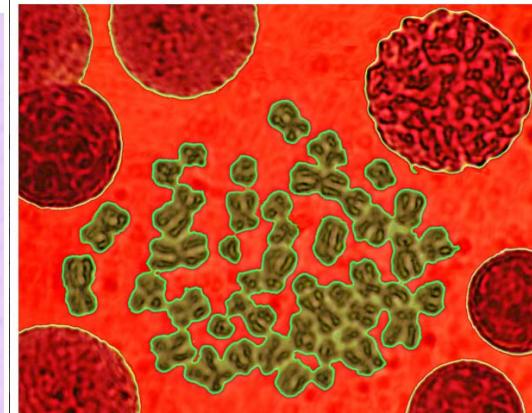
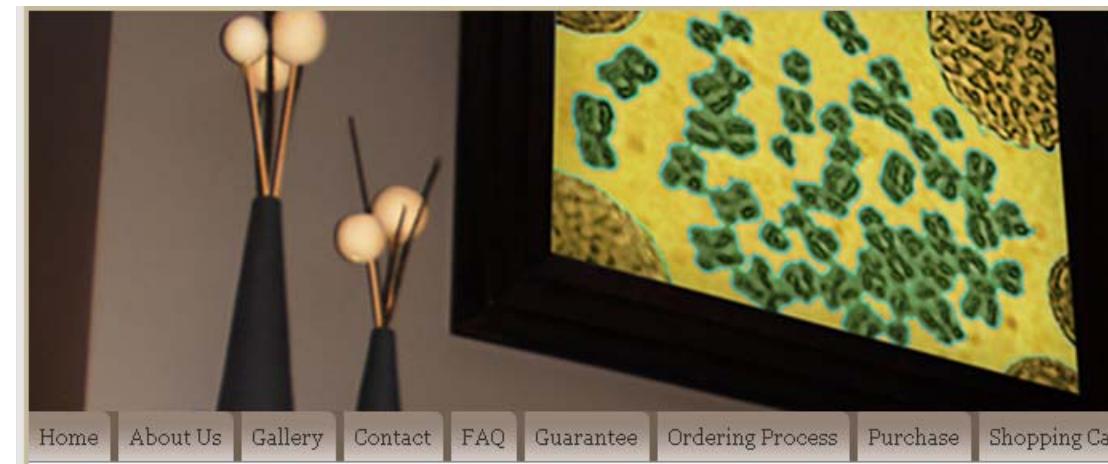
- Non sex-linked & sex-linked inheritance**
- Punnett square to determine possible genotypes for offspring**
- X-inactivation & mosaic female**

## 4. Chromosomal dynamics: normal vs aberrations

*Homologous recombination (normal) and chromosome aberrations (causes abnormal human genetic conditions)*

The image shows the front cover of a book titled "Females are MOSAICS" by Barbara R. Migeon. The title is at the top in large white letters. Below it, the subtitle "X Inactivation and Sex Differences in Disease" is written in smaller white text. The central part of the cover features a stylized human figure composed entirely of small, irregular yellow and white shapes, resembling a mosaic or puzzle pieces. The author's name, "Barbara R. Migeon", is printed at the bottom right in a small white font.

# Chromosomal Art



 **CELLPORTRAITS™**

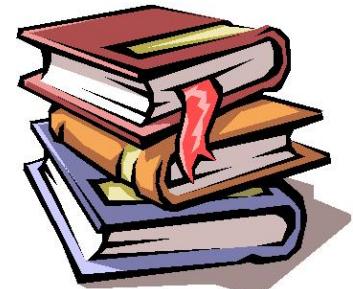
**Gallery**

<http://www.cellportraits.com/>

Refer to Supplemental material: DNA ART

<http://www.cellportraits.com/>

# Additional Enrichment Materials



- Deciphering the Genetic Language of Sex by David C. Page, M.D.
- Sexual Evolution: From X to Y by David C. Page, M.D.
- Dosage Compensation and X-inactivation
- Chromosomal Aberrations
- IVLE Animations : Animal Cell Division; Genetic Terminology; Meiosis; X-inactivation; ABO Blood Group
- Useful Weblinks
  - <http://www.dnafdb.org/#classical>
  - <http://learn.genetics.utah.edu/content/inheritance/intro/>
  - <http://learn.genetics.utah.edu/content/inheritance/patterns/>
  - <http://learn.genetics.utah.edu/content/inheritance/blood/>
  - <http://learn.genetics.utah.edu/content/disorders/chromosomal/>
  - <http://www.hhmi.org/bioInteractive/evolution-y-chromosome>
  - <http://www.hhmi.org/bioInteractive/y-chromosome>
  - <http://media.hhmi.org/bioInteractive/click/gendertest/gendertest.html>