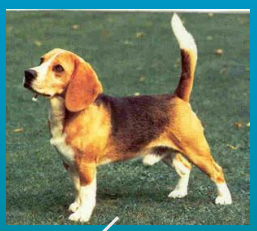
GENETICS

Biology

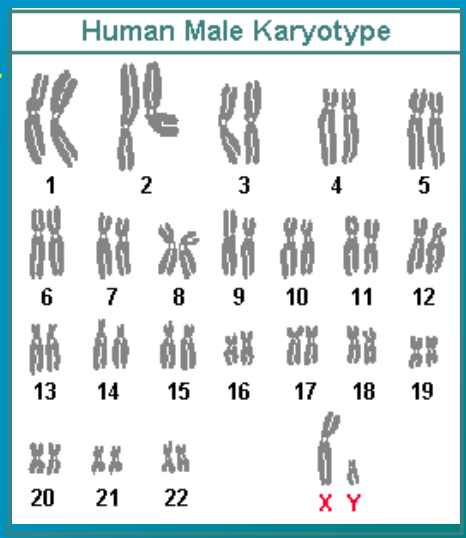
VOCABULARY TERMS

INHERITANCE or **HEREDITY**- The genetic transmission of characteristics from parent to offspring, such as hair, eye, and skin color.



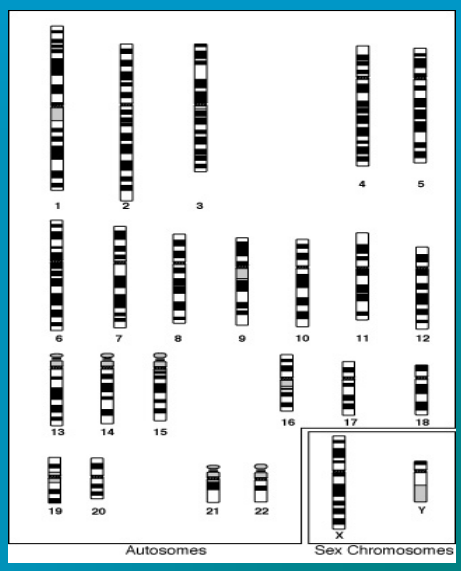
VOCABULARY TERMS

HOMOLOGOUS CHROMOSOME- A pair of matching chromosomes in an organism, with one being inherited from each parent.



VOCABULARY TERMS

AUTOSOME- A chromosome that is not a sex chromosome.



VOCABULARY TERMS

GENOTYPE- the genes present in the DNA of an organism.

There are *always 2 letters* in the genotype because (as a result of sexual reproduction)

1 gene from MOM + 1 gene from DAD = 2 genes (2 letters) for offspring

VOCABULARY TERMS

Now, it turns out there are 3 possible GENOTYPES:

1. 2 capital letters (like "TT")
2. 1 of each ("Tt")
3. 2 lowercase letters ("tt").

Since WE LOVE VOCABULARY, each possible combo has a term for it.

VOCABULARY TERMS

HOMOZYGOUS: GENOTYPE has 2 capital or 2 lowercase letters (ex: TT or tt)

("homo" means "the same")

Sometimes the term "PUREBRED" is used instead of homozygous.

VOCABULARY TERMS

HETEROZYGOUS: GENOTYPE has 1 capital letter & 1 lowercase letter (ex: Tt)

("hetero" means "other")

A heterozygous genotype can also be referred to as ***HYBRID***.

VOCABULARY TERMS

Let's Summarize:

Genotype- genes present in an organism

(usually abbreviated as 2 letters)

- TT = homozygous = purebred
- Tt = heterozygous = hybrid
- tt = homozygous = purebred

VOCABULARY TERMS

- PHENOTYPE- how the trait physically shows-up in the organism; it is the observable traits present in an organism

  What the organism *LOOKS* like

- Examples of phenotypes: blue eyes, brown fur, striped fruit, yellow flowers

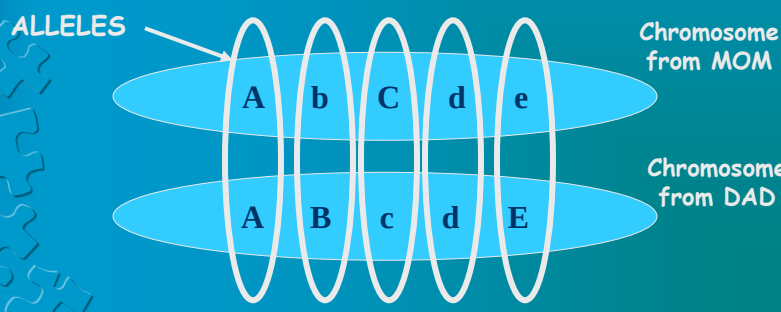
VOCABULARY TERMS

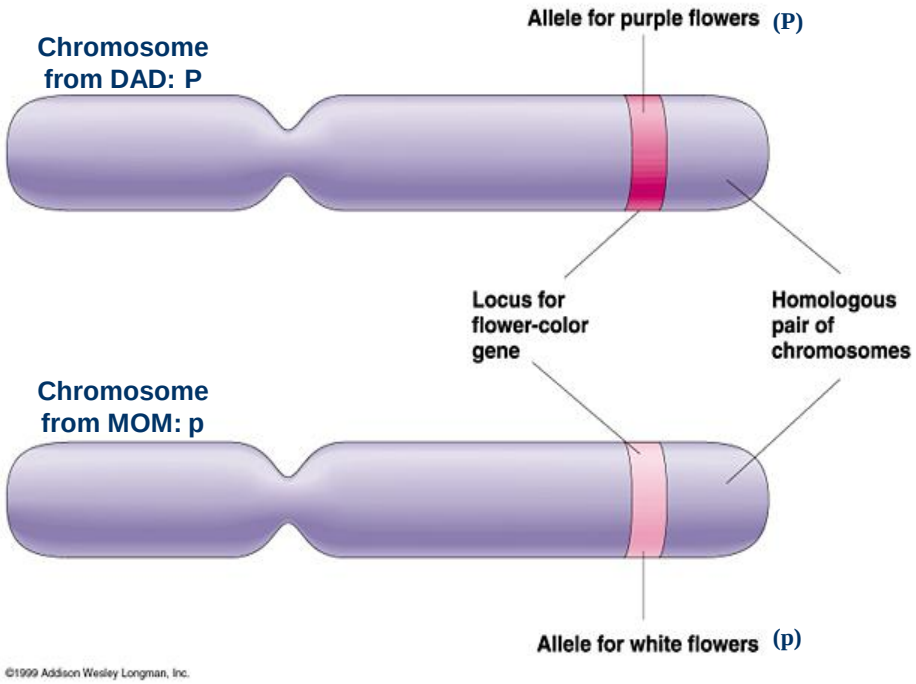
- POLYGENIC INHERITANCE- a trait controlled by two or more genes that may be on the same or on different chromosomes

- Examples of polygenic inheritance: eye color, skin color, and blood group

VOCABULARY TERMS

- ALLELES- alternative forms of the same gene. Alleles for a trait are located at corresponding positions on homologous chromosomes called loci.





VOCABULARY TERMS

- When 1 allele masks (hides) the effect of another, that allele is called **DOMINANT** and the hidden allele is called **RECESSIVE**.

VOCABULARY TERMS

- Dominant alleles are represented by a CAPITAL letter
- Recessive alleles are represented by a LOWERCASE letter

What are Dominant Genes?

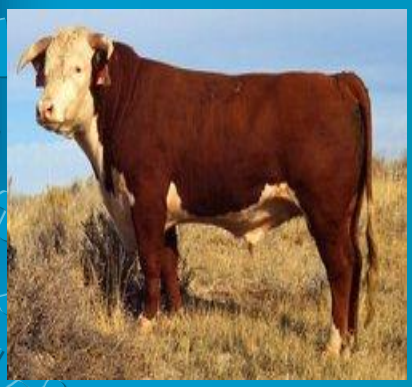
- Dominant Genes = one gene overshadows the other
- Angus Cattle: black is dominant, red is not

Dominant: BB or Bb

Recessive: bb ONLY

What are Dominant Genes?

Hereford: white face is dominant

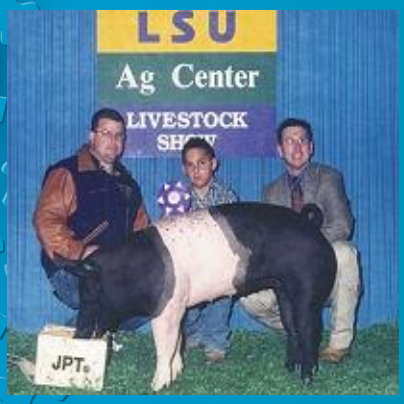


Dominant: WW or Ww

Recessive: ww ONLY

What are Dominant Genes?

- Hampshire Hog: white belt is dominant



Dominant: WW or Ww

Recessive: ww ONLY

What are Recessive Genes?

- The gene that is overshadowed by a dominant gene
- Recessive genes can only express themselves if ***BOTH*** genes are recessive

What are Recessive Genes?

- Horned is recessive to polled.

Dominant: PP or Pp

Recessive: pp ONLY

What are Recessive Genes?

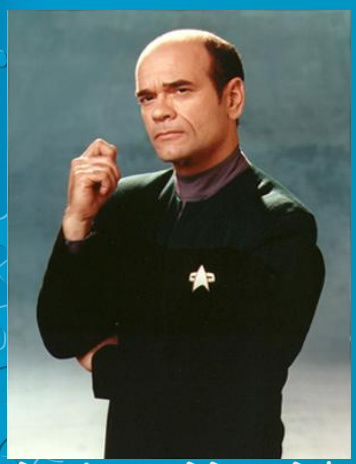
Black wool is recessive to white wool.

Dominant: WW or Ww

Recessive: ww ONLY

What are Recessive Genes?

Dwarfism is recessive to average size.



Dominant: DD or Dd

Recessive: dd ONLY

What are Recessive Genes?

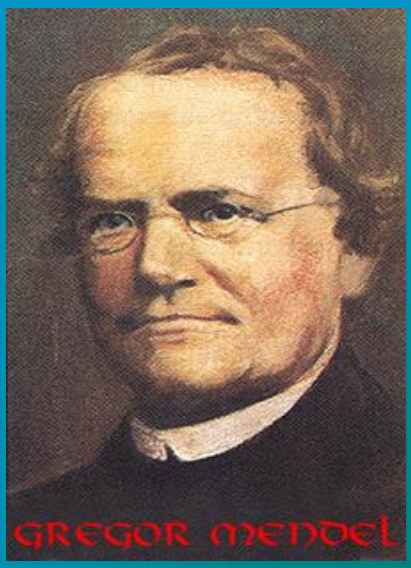
- Albinism (Albino) is recessive to pigmented.

What makes an organism the way that it is?

- NATURE vs. NURTURE
  - Traits that are expressed through genes can be inherited.
  Characteristics that are acquired through environmental influences, such as injuries or practiced skills, cannot be inherited.

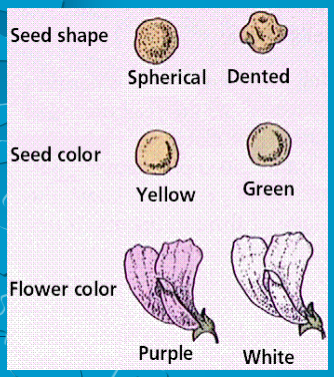
Gregor Mendel (1822-1884)

- Austrian monk
- Called the "Father of Genetics" for his study of the inheritance of 7 traits in pea plants.



Gregor Mendel (1822-1884)

- The traits that Mendel chose to study were easily observable in 2 distinct forms.



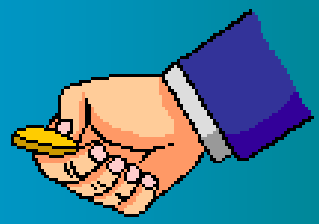
EX.: Stem Height - tall vs. short
Pod Shape - round vs. wrinkled
Flower Color - white vs. purple
Seed Color - green vs. yellow

Gregor Mendel (1822-1884)

- The significance of Mendel's work was not recognized until the turn of the 20th century
- Its rediscovery prompted the foundation of genetics.

Genotype Symbol	Genotype Vocabulary	Phenotype
TT	homozygous DOMINANT or purebred tall	tall
Tt	heterozygous or hybrid	tall
tt	homozygous RECESSIVE or purebred short	short

- Geneticists apply mathematical principles of probability to Mendel's laws of heredity in order to predict the results of simple genetic crosses



- Mendel's laws of heredity are based on his mathematical analysis of observations of patterns of the inheritance of traits.
- The laws of probability govern simple genetic recombinations.
- To see this we use a Punnett Square

PUNNETT SQUARES

- To complete a Punnett square, we use a letter to represent each allele.
- We represent the dominant allele with a capital letter, and the recessive allele is given the same letter but in lowercase.

PUNNETT SQUARES

- For the pea plant flowers:
  dominant: purple color = P
  recessive: white color = p.
- If both parents are purebred, then the purple colored parent must be *PP* and the white colored parent must be *pp*.

How can we predict these results?

Homozygous-dominant

We complete the possible combinations.

Homozygous-recessive

	P	***P***
p	***Pp***	***Pp***
p	***Pp***	***Pp***

These results show that all the F_1 (1st filial generation) offspring are all purple colored hybrids.

	P	***P***
p	*Pp*	*Pp*
p	*Pp*	*Pp*

100% purple offspring

We can use another Punnett square to predict the F_2 (2nd filial generation) offspring.

Heterozygous - hybrid

Heterozygous - hybrid

	P	p
P	PP	Pp
p	Pp	pp

The results are always mathematically the same, a 3:1 ratio with 75% purple & 25% white offspring

Heterozygous - hybrid

Heterozygous - hybrid

	P	p
P	PP	Pp
p	Pp	pp

Phenotypic ratio 3:1
Genotypic ratio 1:2:1

CODOMINANCE

- Not all alleles are dominant and recessive.
- Some alleles are equally strong and neither are masked by the other.
- Alleles which are equally strong are said to be *"codominant"*.

CODOMINANCE

- When both alleles are present, they are both expressed in the phenotype.
- The hybrid is a blend of both alleles.
- When expressing codominant alleles, both alleles are represented by **different capitalized letters**.

CODOMINANCE: F_1 GENERATION

Homozygous

Homozygous

	R	R
W	RW	RW
W	RW	RW

CODOMINANCE: F_1 GENERATION

100% pink offspring

	R	R
W	RW	RW
W	RW	RW

CODOMINANCE: F_2 GENERATION

Heterozygous

Heterozygous

	R	W
R	$R\,R$	RW
W	RW	WW

CODOMINANCE: F_2 GENERATION

Heterozygous

A 1:2:1 ratio with
25% red, 50% pink &
25% white offspring

Heterozygous

	R	W
R	$R\,R$	RW
W	RW	WW

CODOMINANCE: IN HUMANS

Blood Type:

phenotypic ratio

1:1:1:1

1 type A

1 type B

1 type AB

1 type O

	I_A	I_O
I_B	$I_A I_B$	$I_B I_O$
I_O	$I_A I_O$	$I_O I_O$

CODOMINANCE: IN HUMANS

Blood Type:

A & B are equally strong.

O is recessive.

$I_A I_O$ is Type A

$I_B I_O$ is Type B

$I_A I_B$ is Type AB

$I_O I_O$ is type O

INCOMPLETE DOMINANCE

- *Incomplete dominance* is a situation in which both alleles are equally strong and both alleles are visible in the hybrid genotype.
- When an intermediate phenotype occurs and no allele dominates, incomplete dominance results.

INCOMPLETE DOMINANCE

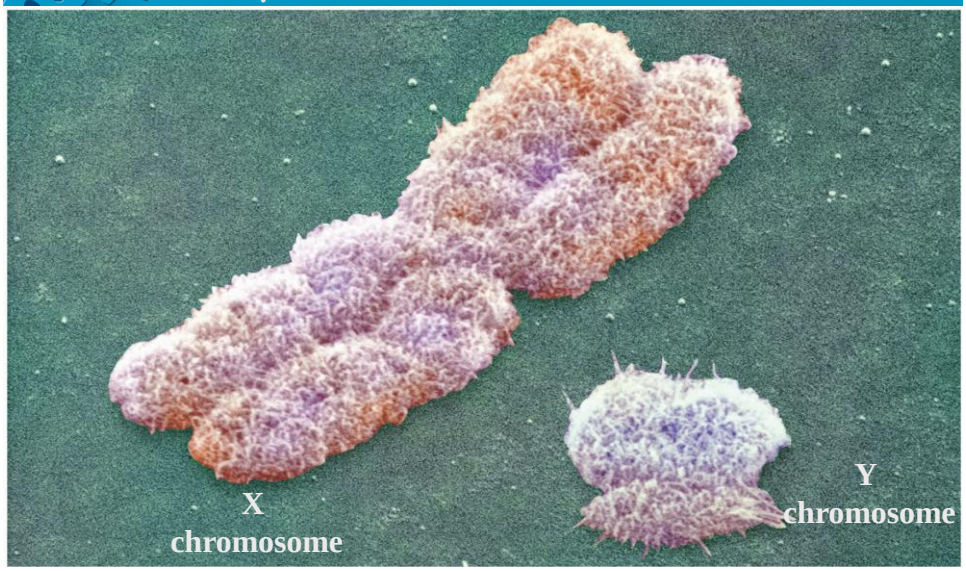
EX.

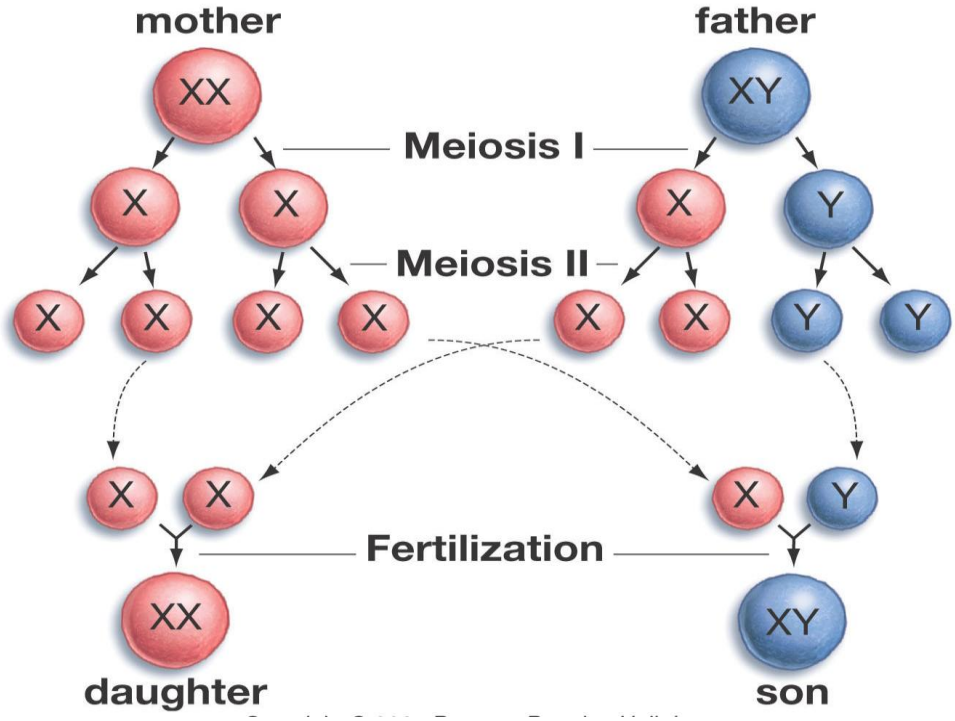
INCOMPLETE DOMINANCE

EX.

SEX-LINKED TRAITS

Boy or Girl? The Y Chromosome "Decides"





What are Sex Linked Traits?

- In 1910, Thomas Morgan discovered traits linked to sex chromosomes in fruit flies.
- Some genes are attached to the X and Y chromosomes
- EXAMPLE: In humans, colorblindness and baldness are found on the X chromosomes

What are Sex Linked Traits?

- In Men, traits expressed anytime present
- In Women, must have two genes to show trait
- Children inherit baldness from their mothers

Punnett Square: What sex will the offspring be?

	X	Y
X	X X	X Y
X	X X	X Y

50% chance of a male or a female child.

Baldness is carried by the mother

	X	Y
X_B	$X X_B$	$X_B Y$
X	X X	X Y

Phenotype:

25% bald males

25% bald carrier females

25% not bald males

25% non-carrier females

If Dad is bald, will you be bald?

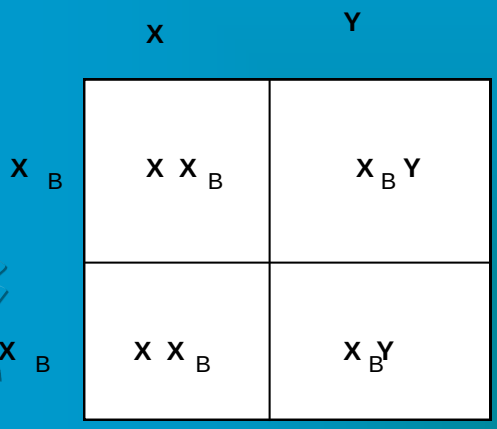
	X_B	Y
X	$X X_B$	X Y
X	$X X_B$	X Y

Phenotype:

0% bald males

100% bald carrier females

What if Mom is bald?



Phenotype:

100% carrier females

100% bald males

GENETIC DIVERSITY

- The sorting and recombination of genes in sexual reproduction results in a great variety of gene combinations in the offspring of any 2 parents.
- Do you look EXACTLY like your brothers & sisters?

Genetic Diversity

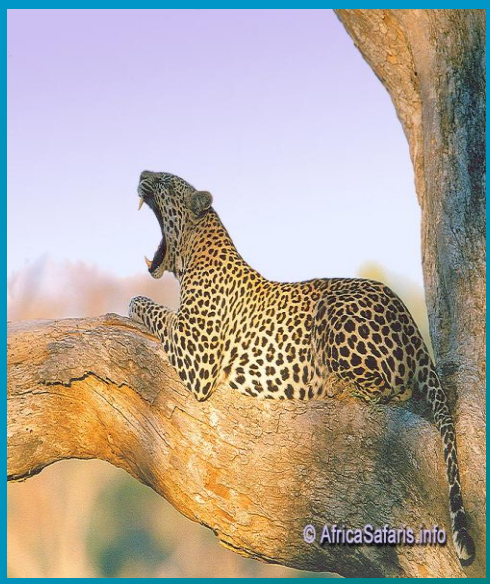
- Genetically diverse populations are more likely to survive changing environments.
- Greater variation within the species makes a population better suited to adaptation to changes in the environment.

Genetic Diversity

- Leopard populations around the world are in danger because of inbreeding.

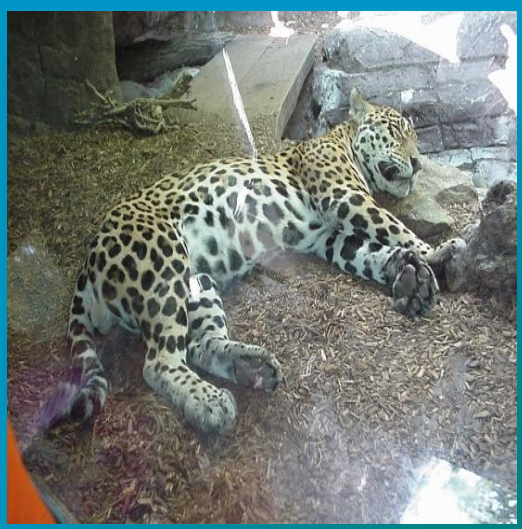
GENETIC DIVERSITY

- There is very little genetic variation between any 2 individuals.



GENETIC DIVERSITY

- This makes them VERY susceptible to disease & will likely lead to their extinction.



Genetic Diversity

- Recombination and mutation provide for genetic diversity.
- Inserting, deleting, or substituting DNA bases can alter genes.
- An altered gene in a sex cell may be passed on to every cell that develops from it, causing an altered phenotype.

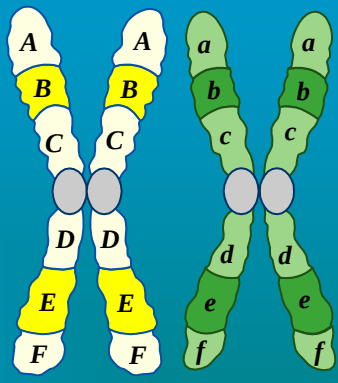
Recombination

Crossing-over

- The physical exchange of chromosomal material between chromatids of homologous chromosomes.
- Result: Generation of new combinations of genes (alleles).

RECOMBINATION

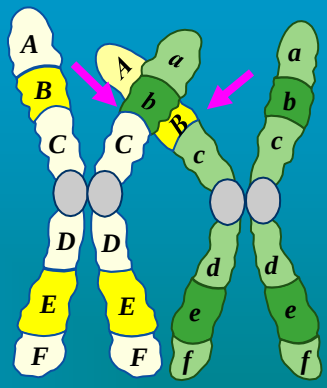
- Occurs in prophase of meiosis I
- Generates diversity



Creates chromosomes with new combinations of alleles for genes A to F.

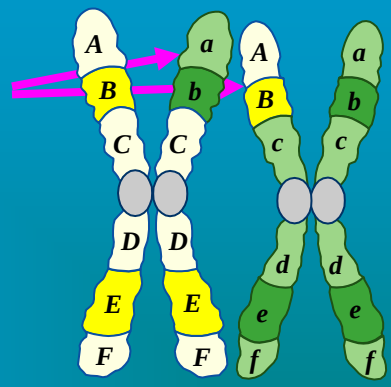
RECOMBINATION

Letters denote genes
Case denotes alleles

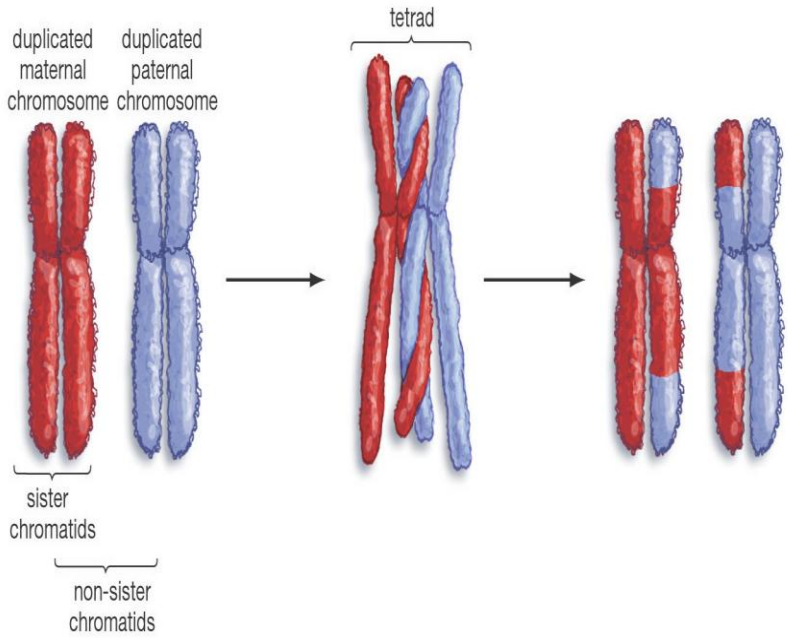


RECOMBINATION

Alleles have crossed over to produce new gene combinations



Exchange of parts of non-sister chromatids.



GENETIC DIVERSITY

- Sometimes entire chromosomes can be added or deleted, resulting in a genetic disorder such as Trisomy 21 (Down syndrome).

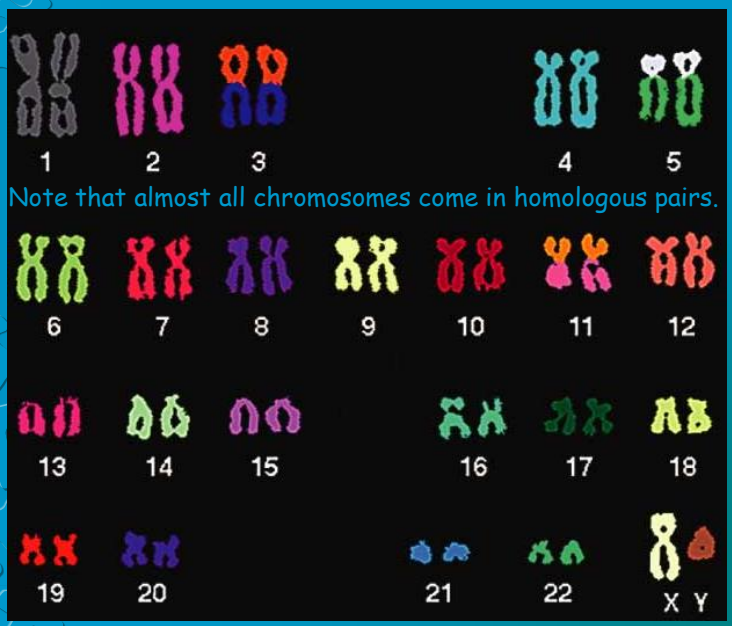
GENETIC DIVERSITY

- Chromosomal Errors
- NONDISJUNCTION: the failure of chromosomes to separate properly in meiosis. Gametes with extra or too few chromosomes result.
- Can cause diseases such as Down's Syndrome.

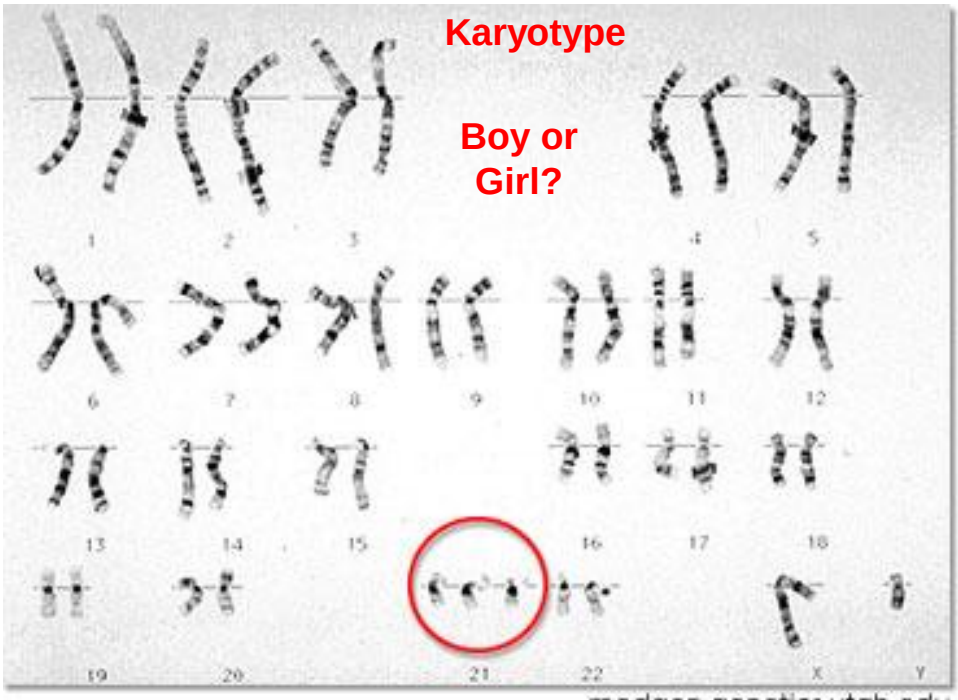
GENETIC DIVERSITY

- Chromosomal Errors
- **POLYPLOIDY:** organisms with entire extra sets of chromosomes
- Results in the death of the fetus in animals
- Often occurs in plants and causes the fruits and flowers to be larger. EX.: bananas, lilies

A Karyotype is an Informative, Arranged Picture of Chromosomes At Their Most Condensed State



Boy or girl?



Genetic Diseases

Turner's Syndrome

- Turner's syndrome is a genetic disorder affecting only females, in which the patient has one X chromosome in some or all cells; or has two X chromosomes but one is damaged.

Genetic Diseases

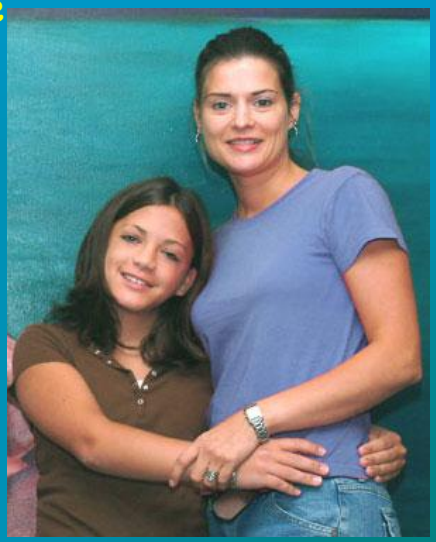
Turner's syndrome

- Signs of Turner syndrome include:
  - short stature,
  - delayed growth of the skeleton,
  - shortened fourth and fifth fingers,
  - broad chest,
  - and sometimes heart abnormalities.

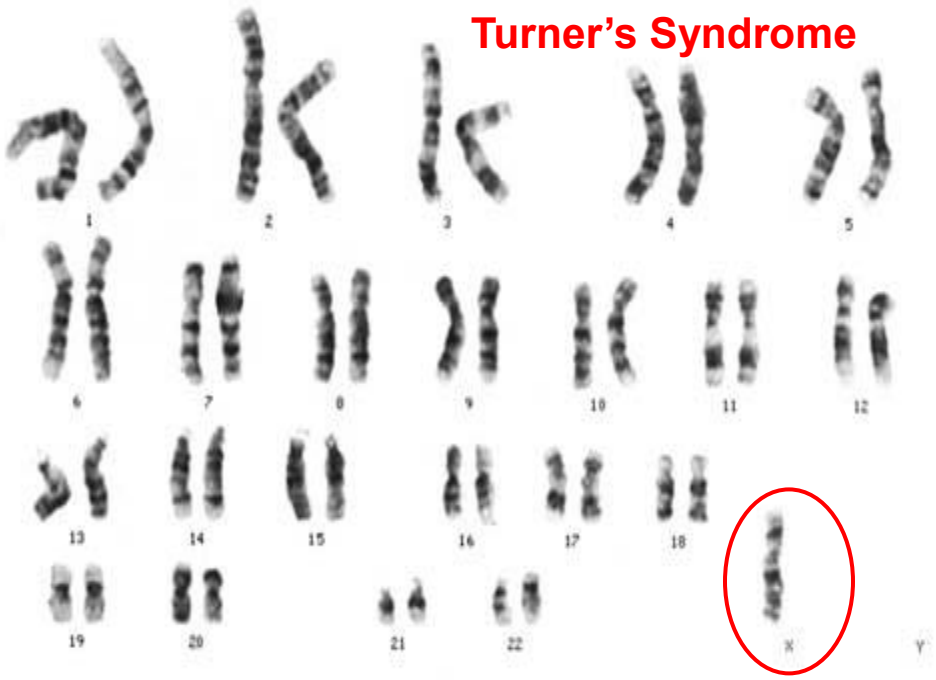
Genetic Diseases

Turner's syndrome

- Women with Turner syndrome are usually infertile due to ovarian failure.
- Diagnosis is by blood test (karyotype).



Turner's Syndrome



Genetic Diseases

Huntington's Disease

- Huntington's disease (HD) is an inherited disorder caused by the degeneration of certain nerve cells in the brain.
- The gene for Huntington's disease is codominant.
- HD causes bizarre involuntary movements and loss of intellectual abilities (dementia).

Genetic Diseases

Huntington's Disease

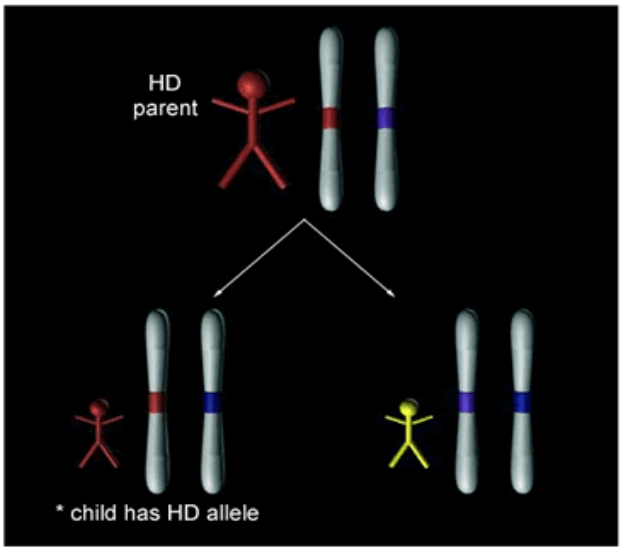
- The condition begins most often in mid-adulthood and progresses slowly to death.

Genetic Diseases

Huntington's Disease

- The identification of the codominant gene for HD now makes it possible to determine who will develop this disease by examining their DNA from a blood sample in the laboratory.

Figure C-2: Risk for child of HD individual



Huntington's Disease

HD allele

non-HD allele from HD parent

non-HD allele from non-HD parent

Each child has 1 in 2 chance of inheriting the non-HD allele. This is a 50% risk.

This diagram shows how HD may or may not be passed from parent to child. The HD allele is the gene that causes HD, and the non-HD allele is the alternative gene that does not cause HD.

www.stanford.edu

Genetic Diseases

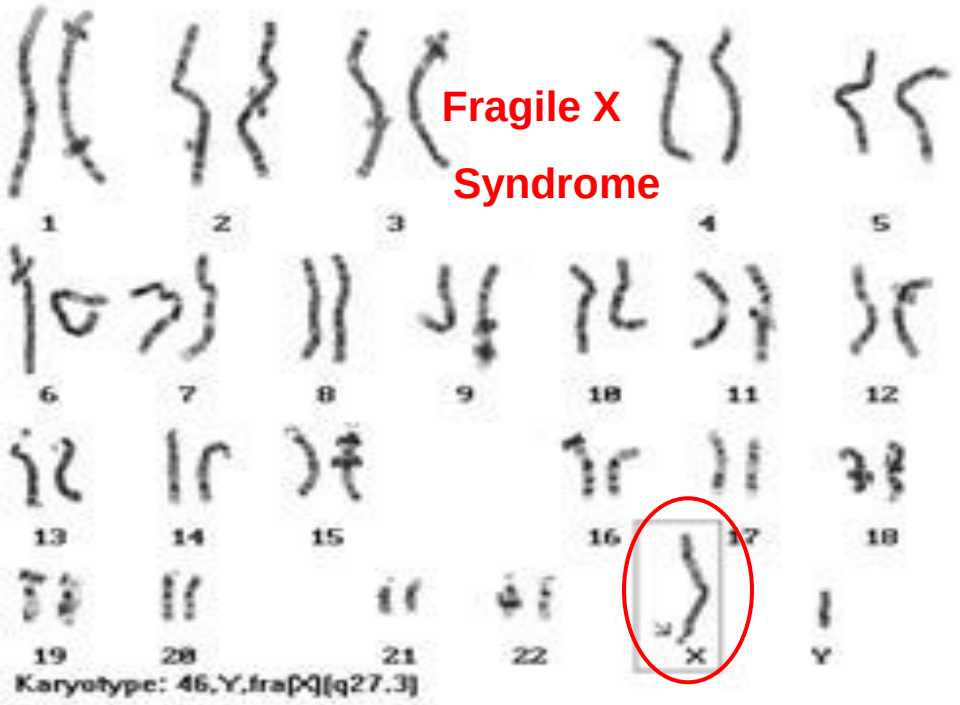
Fragile X Syndrome

- An inherited disorder caused by a defective gene on the X-chromosome.

Genetic Diseases

Fragile X Syndrome

- Symptoms of Fragile X Syndrome:
  - mental retardation,
  - Enlarged testes,
  - and facial abnormalities in males
  - and mild or no effects in females.
- It is the most common inherited cause of mental retardation.

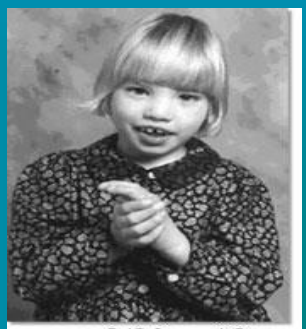


Genetic Diseases

Cri-du-chat Syndrome

- Cri-du-chat Syndrome is a rare genetic disorder due to a missing portion of chromosome # 5.

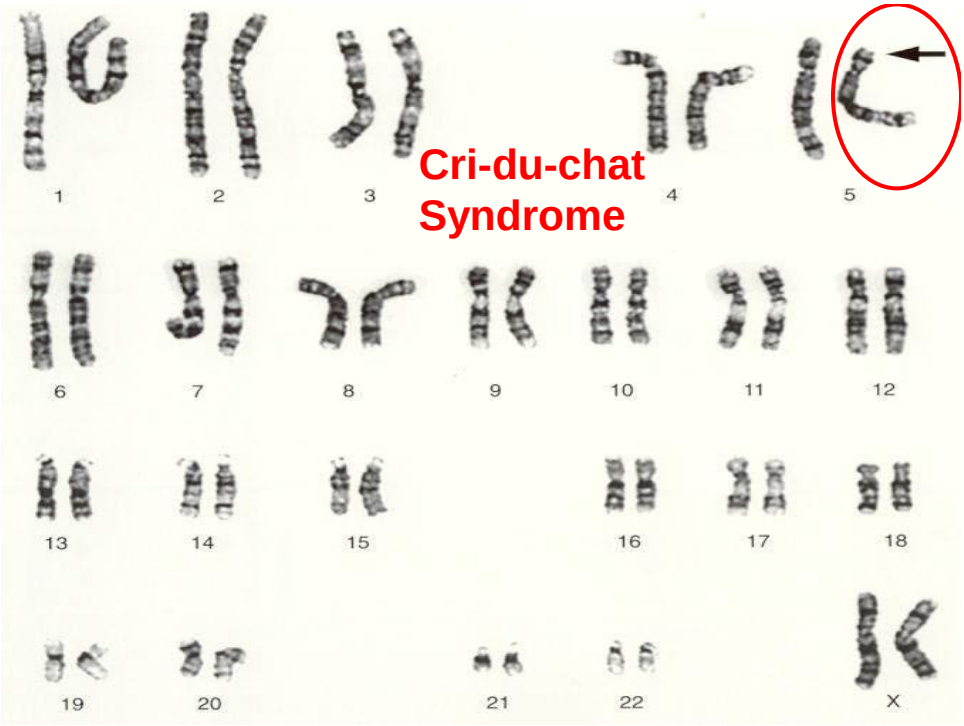
Its name, meaning *cat cry* in French, is from the distinctive mewing sound made by infants with the disorder.



Genetic Diseases

Cri-du-chat Syndrome

- The disorder is characterized by:
- distinctive facial features,
- small head size,
- low birth weight,
- weak muscle tone,
- a round face,
- epicanthal folds,
- low set ears,
- facial asymmetry
- severe mental retardation is typical



Genetic Diseases

Tay-Sachs Disease

- A hereditary disease that affects young children almost exclusively of eastern European Jewish descent, in which an enzyme deficiency leads to the accumulation of fat in the brain and nerve tissue.

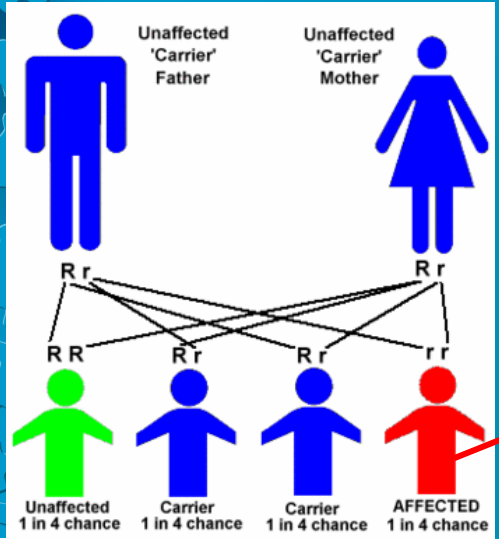
Genetic Diseases

Tay-Sachs Disease

- Tay-Sachs results in:
  - mental retardation,
  - convulsions,
  - blindness,
  - and ultimately death.

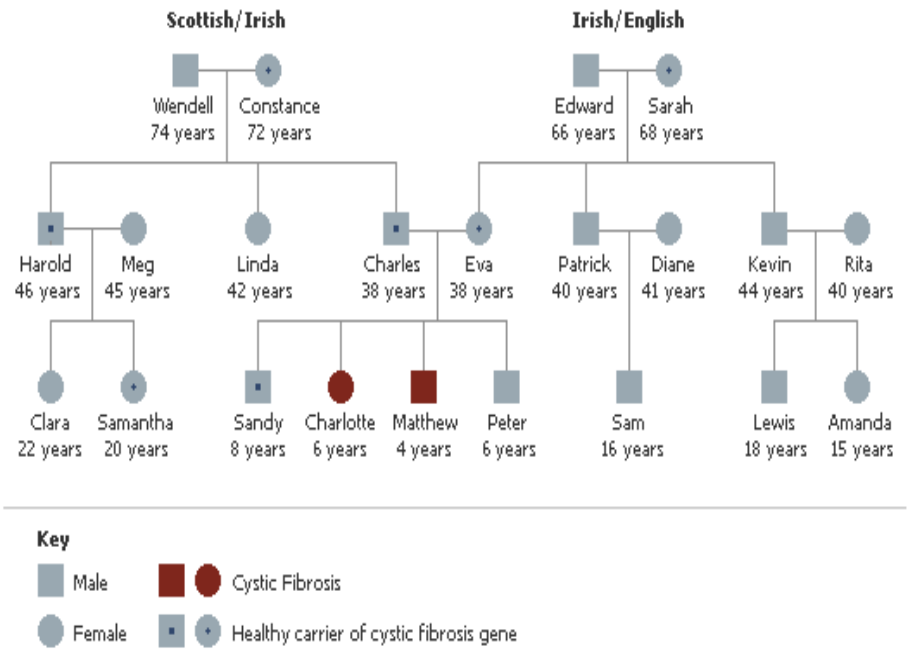
Genetic Diseases

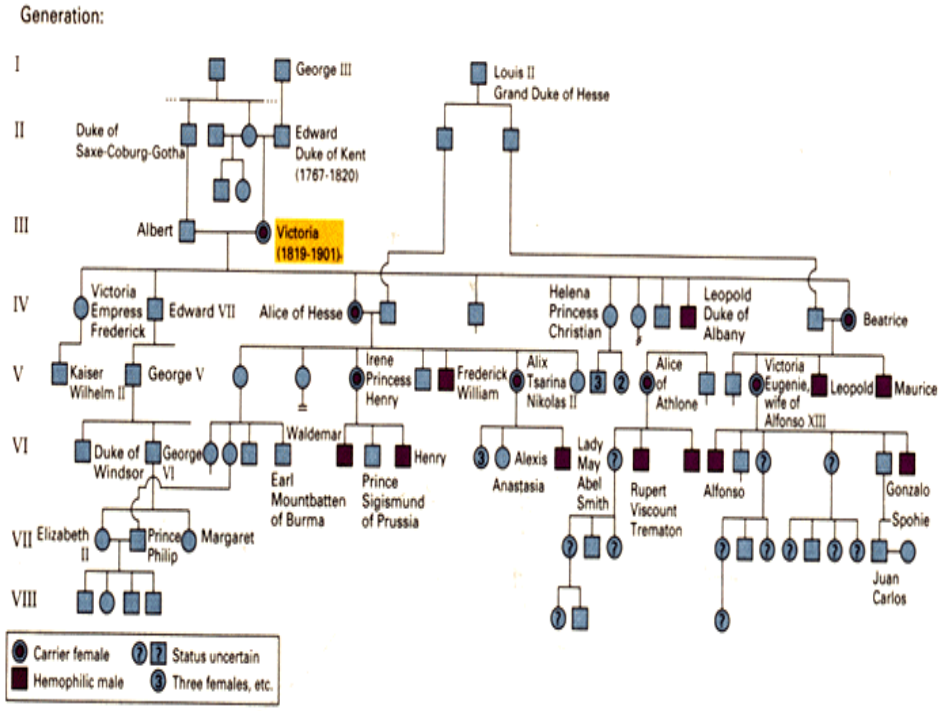
Tay-Sachs Disease



Pedigrees

- Pedigree charts show a record of the family of an individual.
- It can be used to study the transmission of a hereditary condition.
- It is particularly useful when there are large families and a good family record over several generations.





ETHICAL & MORAL CONCERNS

- The potential for identifying and altering genomes raises practical and ethical questions.

Ethical & Moral Concerns

- Eugenics, a pseudo-science of selective breeding of humans, was a movement throughout the twentieth century, worldwide as well as in Virginia, that demonstrated a misuse of the principles of heredity.

Ethical & Moral Concerns

- Eugenics is a dangerous idea that subtly promotes racism.
- Hitler was a proponent of eugenics and tried to create a "superior" race known as the Aryans.

ETHICAL & MORAL CONCERNS

- Cloning is another morally charged issue facing us today.
- *Cloning* is the production of genetically identical cells and/or organisms.

ETHICAL & MORAL CONCERNS

- Dolly was famous all over the world because of the way she was born, in 1996. She was the world's first cloned mammal.

Dolly the sheep 1996 - 2003

ETHICAL & MORAL CONCERNS

- Other cloned animals