GENETICS

Agriscience 1

OBJECTIVE

The student will investigate and understand common mechanisms of inheritance and protein synthesis. Key concepts include:

- d) prediction of inheritance of traits based on the Mendelian laws of heredity;
- e) genetic variation (mutation, recombination, deletions, additions to DNA);
- h) use, limitations, and misuse of genetic information; and
- i) exploration of the impact of DNA technologies.

Do you have . . .

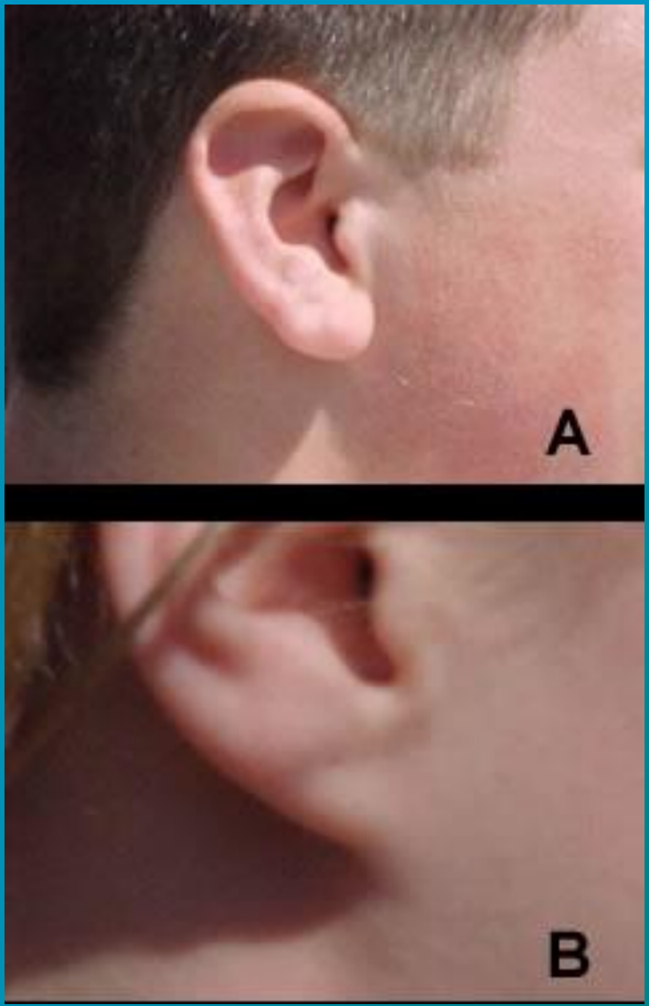
- A widow' s peak?

YES

NO

Do you have . . .

- Attached earlobes



NO

YES

Do you have . . .

- The ability to roll your tongue?

Do you have . . .

- A hitchhiker’s thumb?

YES

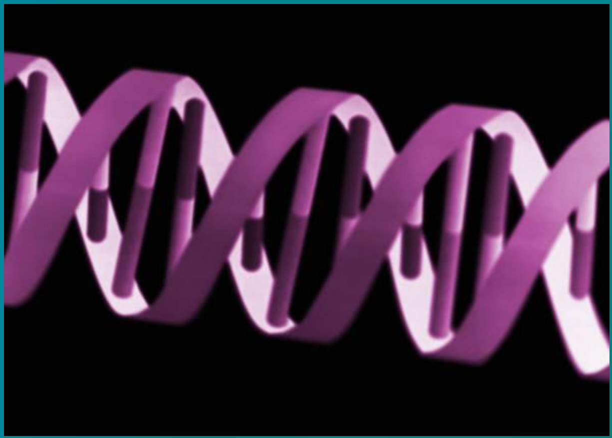
NO

Reflection

- Why do some people and animals have certain traits that others do not?
- Why are some of these traits more common than others?

DNA

- DNA is your instruction. It is the genetic blue print. DNA is made of small nucleotides connected together.
- FUN FACT: Every human is 99.9% genetically the SAME!!!
- BUT it's the proteins we make that makes EACH of us DIFFERENT!



So what makes the difference??

Genes

- Small segments of DNA are called genes. Genes are then used to make proteins. These genes wind together to form chromosomes.

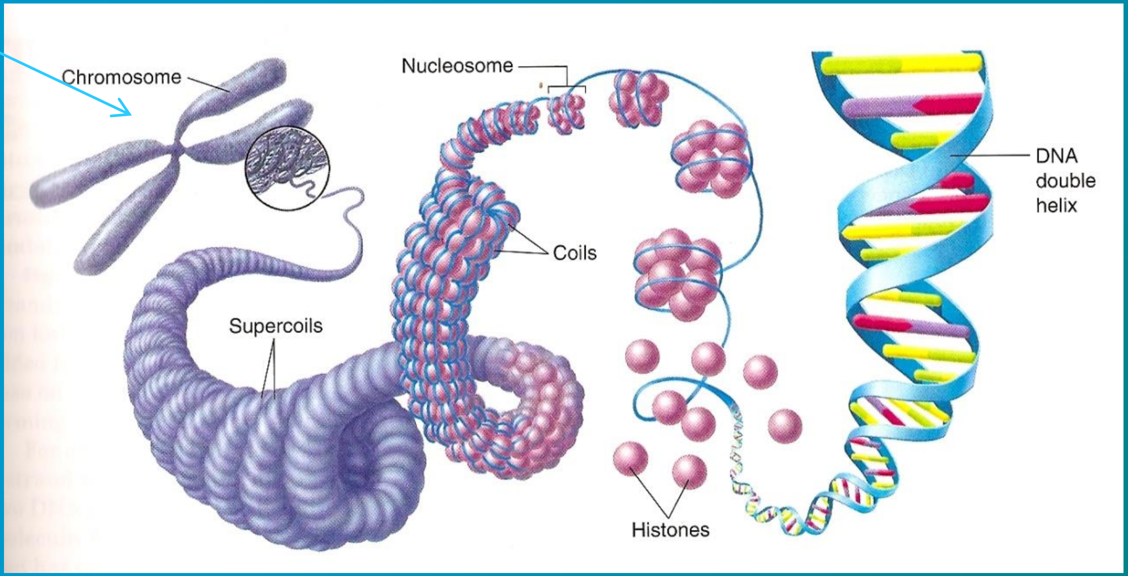
- How do we acquire specific genes?

VOCABULARY TERMS

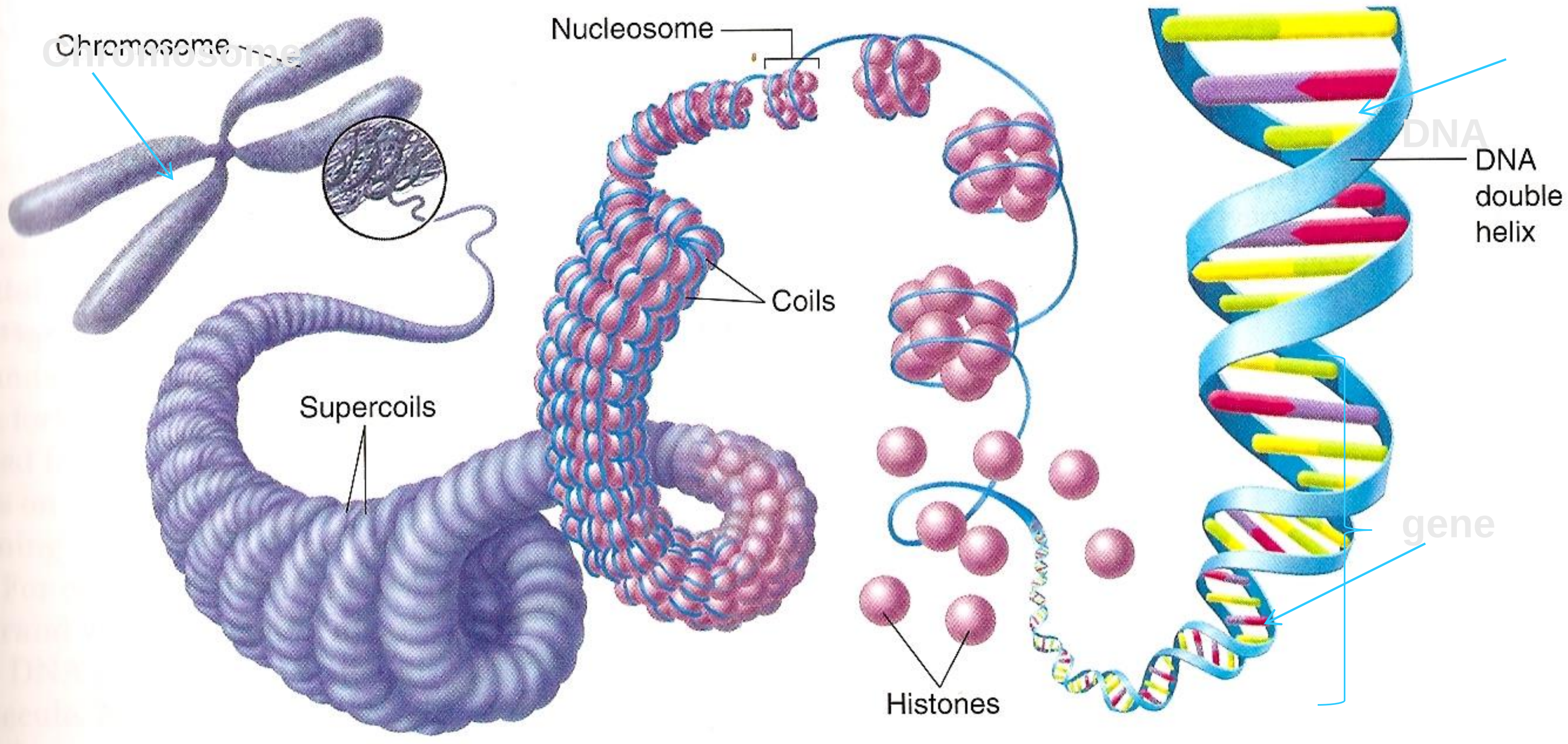
INHERITANCE or **HEREDITY**- The genetic transmission of characteristics from parent to offspring, such as hair, eye, and skin color.

Chromosomes

- During cell division, DNA winds up to form chromosomes. Chromosomes are MADE of genes (small pieces of DNA) and proteins. **Chromosomes** are genes that you see!



Big Picture



Chromosome
Chromosome
Nucleosome
Coils
Supercoils
Histones
DNA
DNA double helix
gene

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

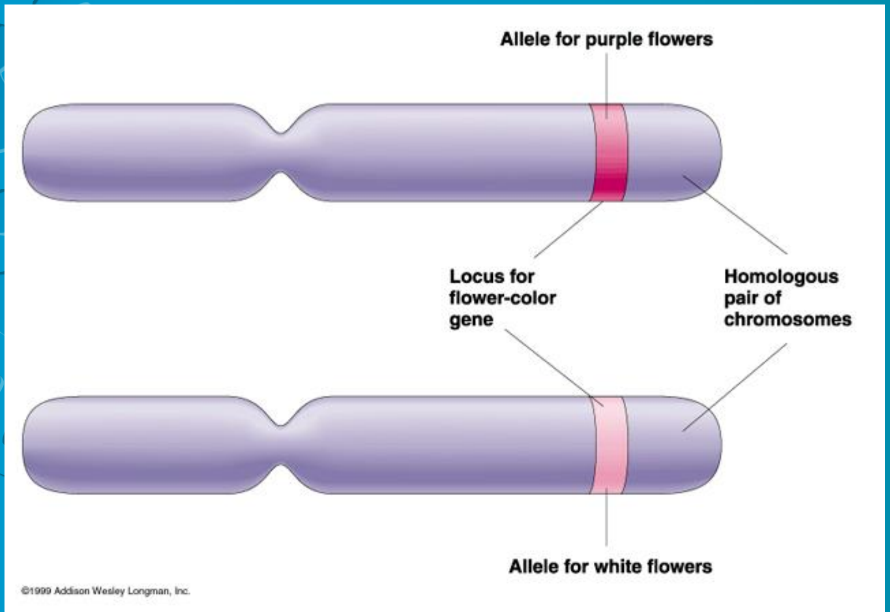
Vocabulary Terms

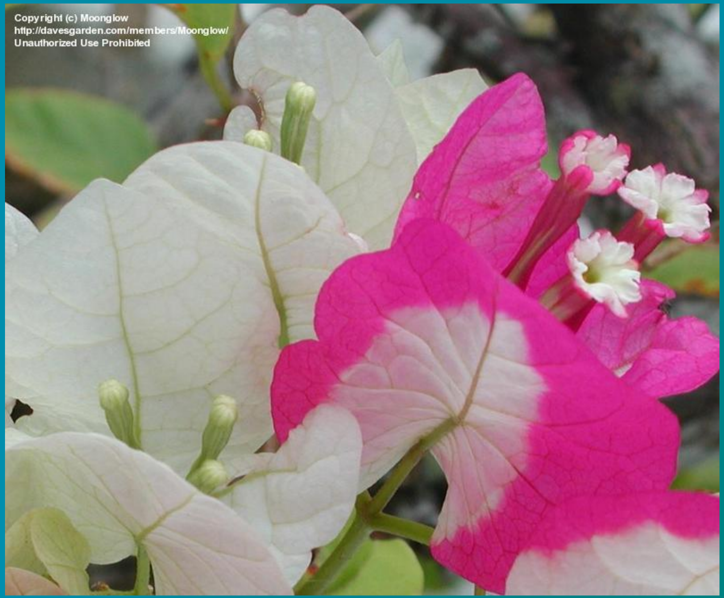
- POLYGENIC INHERITANCE- a trait controlled by two or more genes that may be on the same or on different chromosomes

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

Key Terms

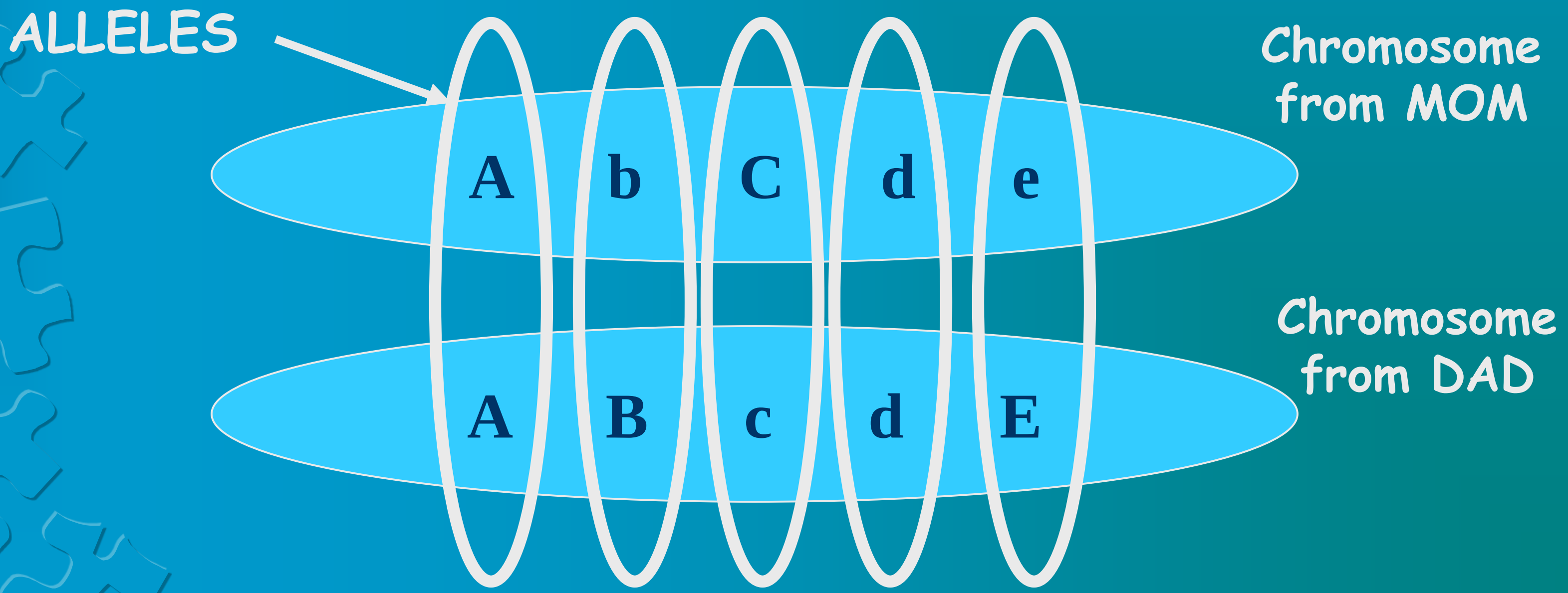
- **Trait** — characteristic that is inherited; can be either dominant or recessive

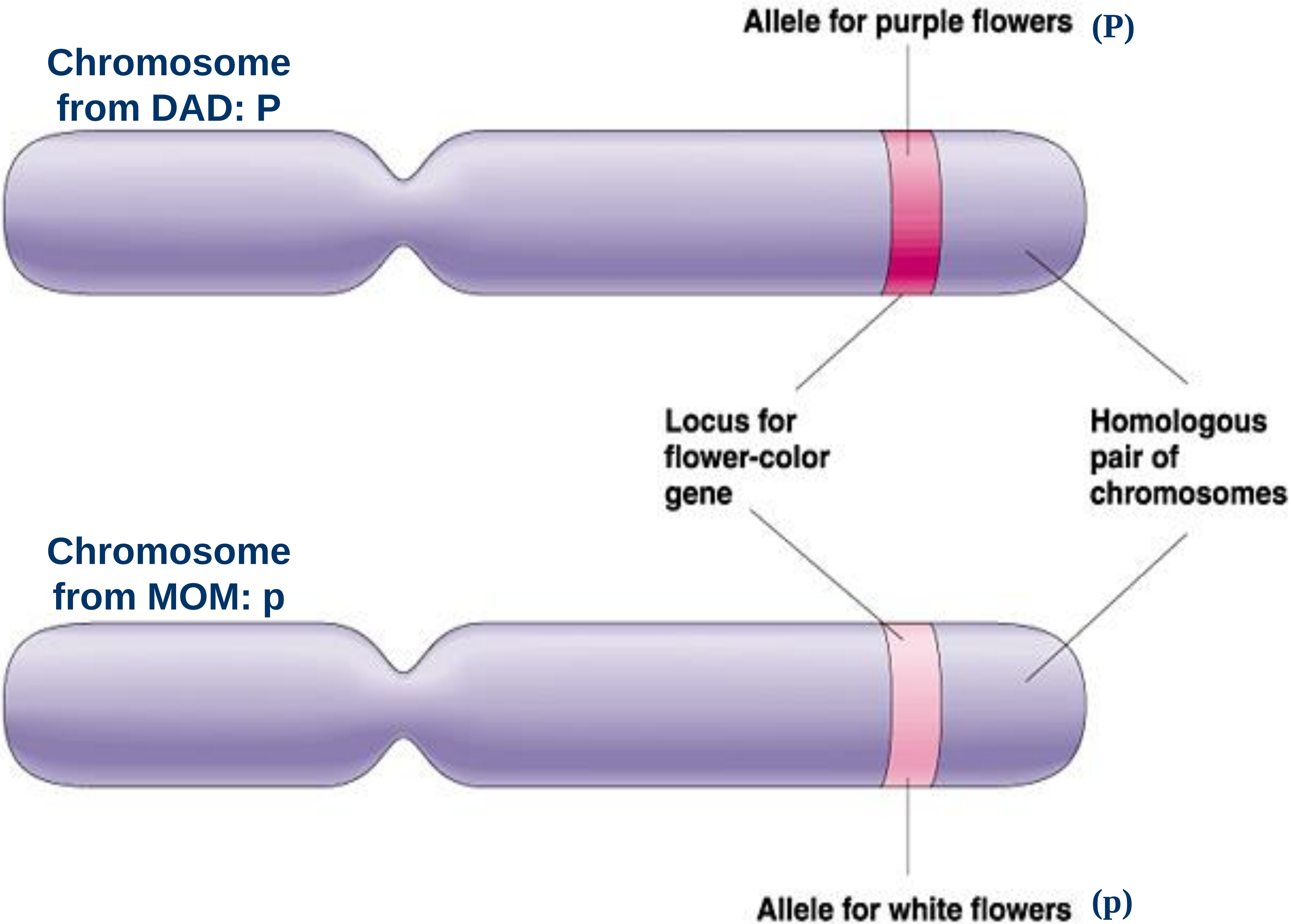




VOCABULARY TERMS

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




Chromosome from DAD: P
Allele for purple flowers (P)
Locus for flower-color gene
Homologous pair of chromosomes
Chromosome from MOM: p
Allele for white flowers (p)

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 ***<u>BOTH</u>*** genes are recessive

What are Recessive Genes?

- Horned is recessive to polled.

Dominant: PP or Pp

Recessive: pp ONLY

What are Recessive Genes?

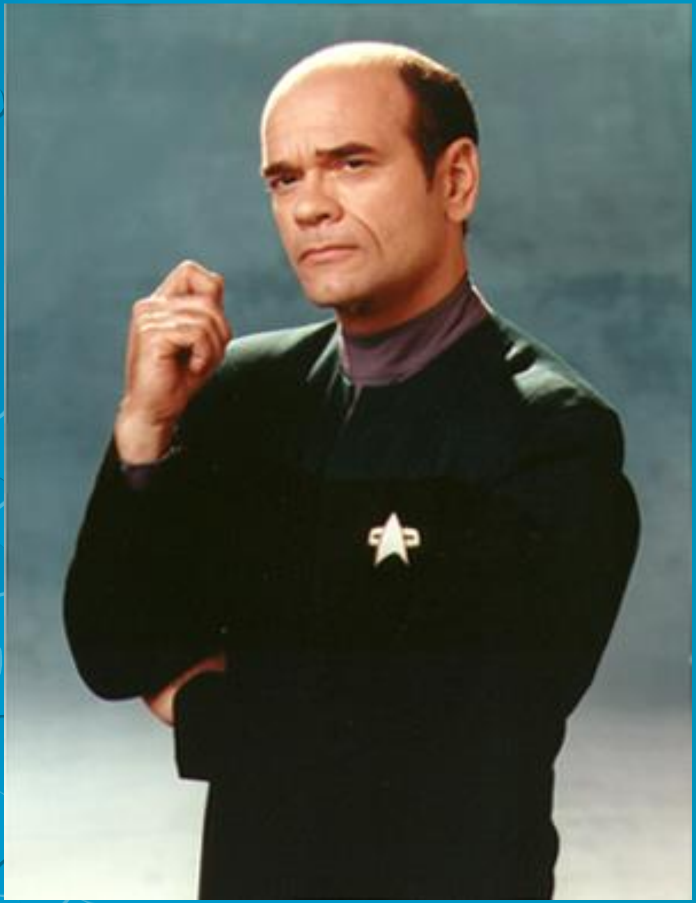
Black wool is recessive to white wool.

Dominant: WW or Ww

Recessive: ww ONLY

What are Recessive Genes?

- Dwarfism is recessive to average size.



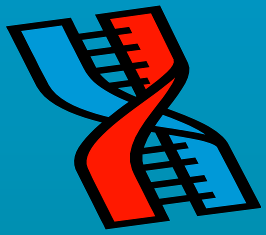
Dominant: DD or Dd

Recessive: dd ONLY

What are Recessive Genes?

- Albinism (Albino) is recessive to pigmented.

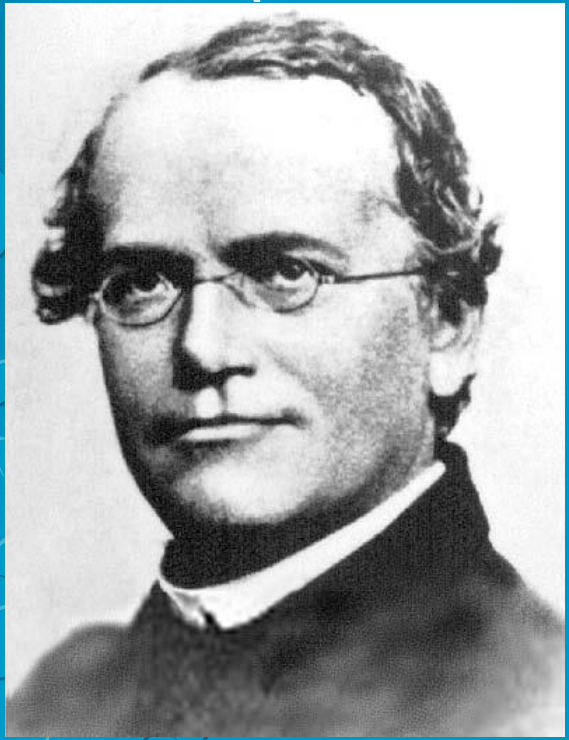
What makes an organism the way that it is?



- <u>NATURE vs. NURTURE</u>
  - 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.

Historical Background

- Gregor Mendel (born 1822) is known for his experiments with sweet pea plants.



Gregor Mendel

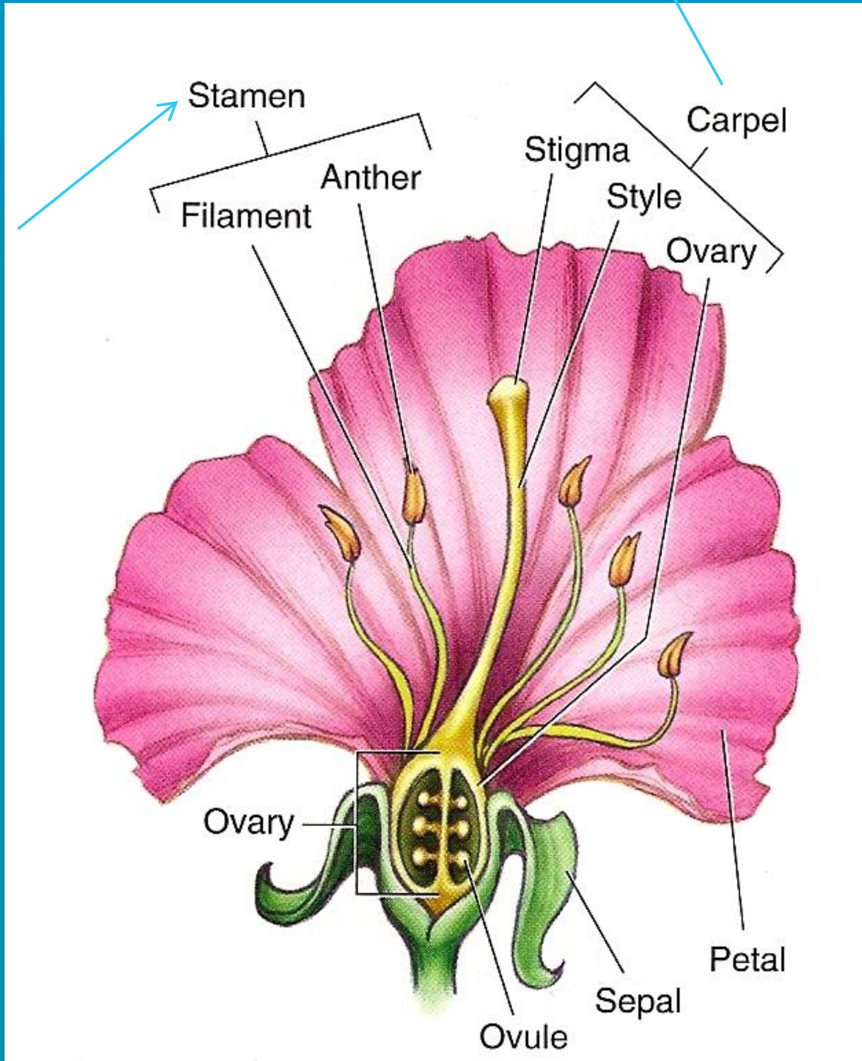
- Grew up in small region of Austria that is now part of the Czech Republic
- Many remember him as an Austrian monk

- The Father of Genetics

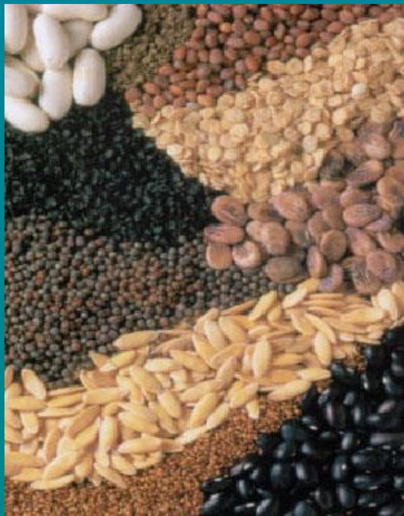
- *Flower Sex Organs*
  - *Stamen*
  - *Anther*
  - *Carpel*
  - *Ovary*

Female

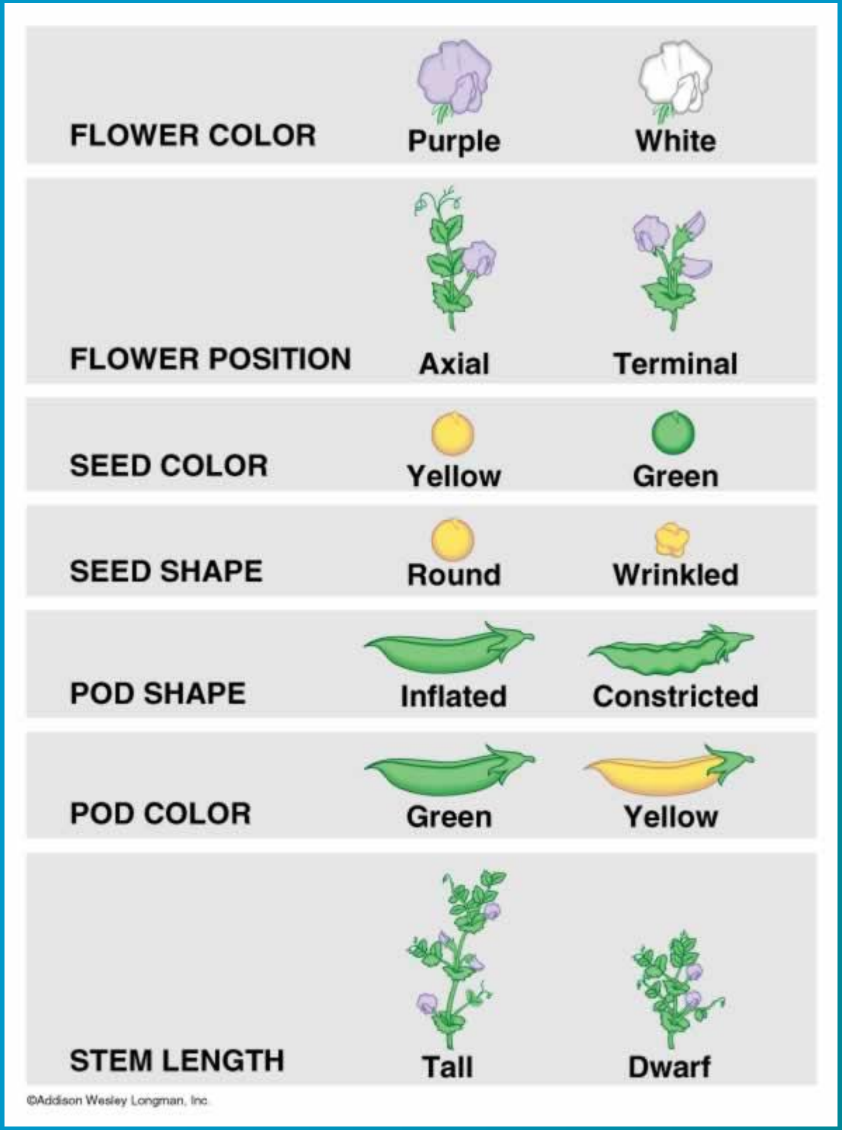
Male



- Flowers reproduce sexually. One flower contains sperm cells in its pollen and egg cells in its ovaries. Male and female sex cells must join in fertilization to create a tiny flower embryo inside a seed.



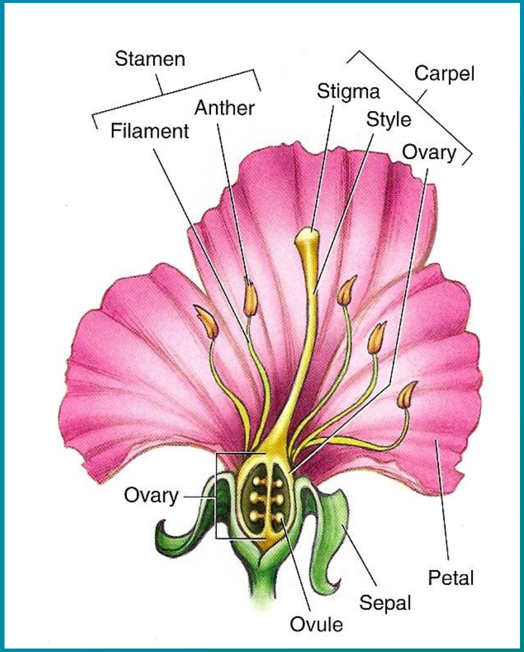
Mendel noticed . . .



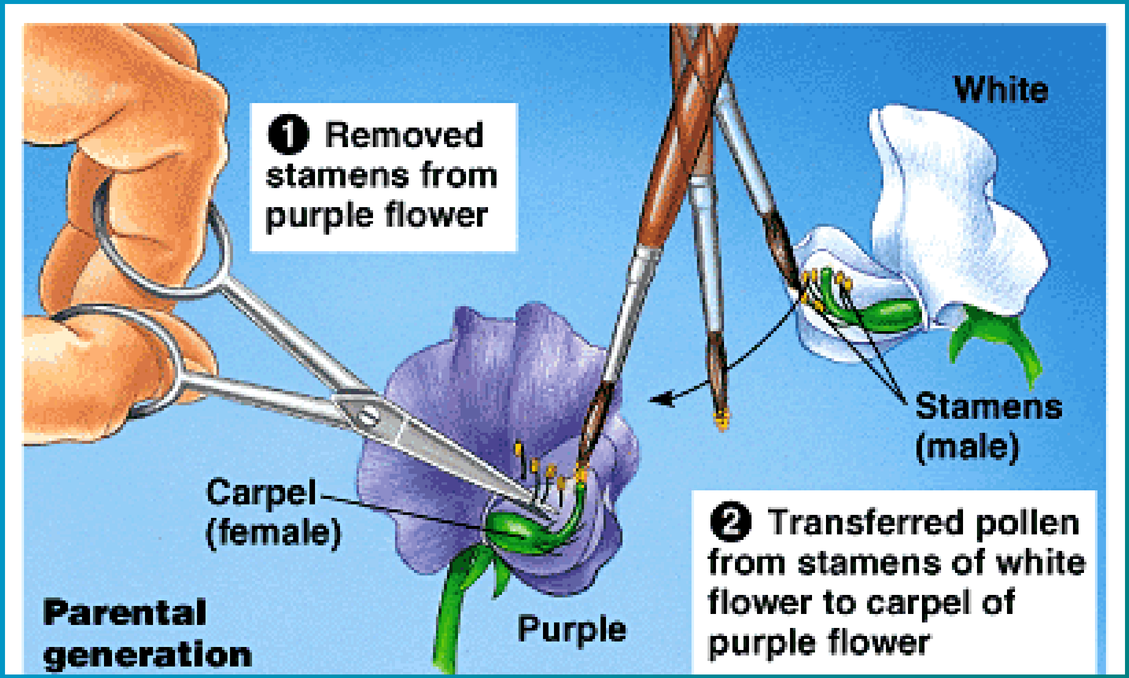
Determining Genotypes and Phenotypes

Trait	Alleles	
Seed Shape	Round (R ___)	Wrinkled (rr)
Seed Color	Yellow (Y ___)	Green (yy)
Seed coat color	Gray (W ___)	White (ww)
Pod Shape	Smooth (S ___)	Wrinkled (ss)
Pod Color	Green (G ___)	Yellow (gg)
Flower Position	Axial (A ___)	Terminal (aa)
Plant Height	Tall (T ___)	Short (tt)

- Pea flowers self-pollinate, which means pollen from the anther drops into the ovary and fertilizes the egg cells.

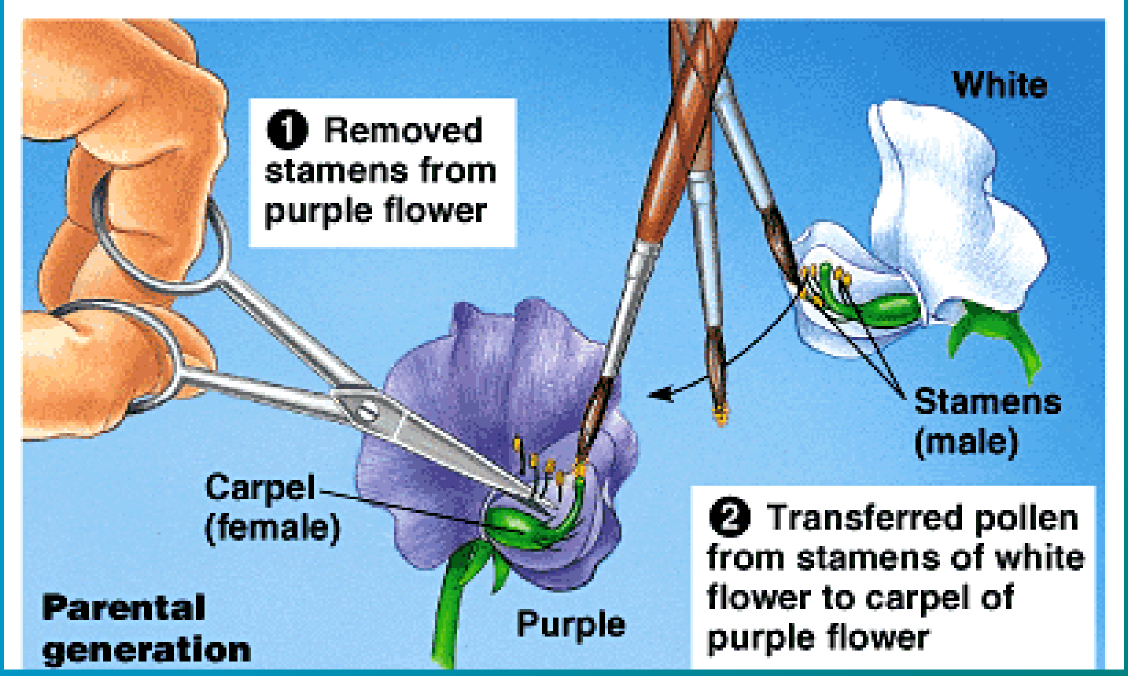


- In order to perform a controlled experiment, Mendel removed the anther from one plant so he could control pollination.



- Mendel started with true-breeding plants, which produce offspring identical to themselves.
  - Example: purple flowered plants produce offspring with purple flowers

- Mendel took pollen from a white flowered plant to fertilize a purple flowered plant. Mixing plants with two different traits creates a <u>hybrid</u> plant.



- **Traits are determined by specific sequences of DNA called genes.
  - Example: pea plants have a gene for flower color, pea plants have a gene for plant height, pea plants have a gene for seed shape.
  - What other genes do pea plants have?

- **There are different possible forms of the same gene called alleles.
  - Example: pea plants can have a purple allele or a white allele for flower color.
  - pea plant can have a tall allele or a short allele for plant height.
  - pea plants can have a round allele or a wrinkled allele for seed shape.

Question…..

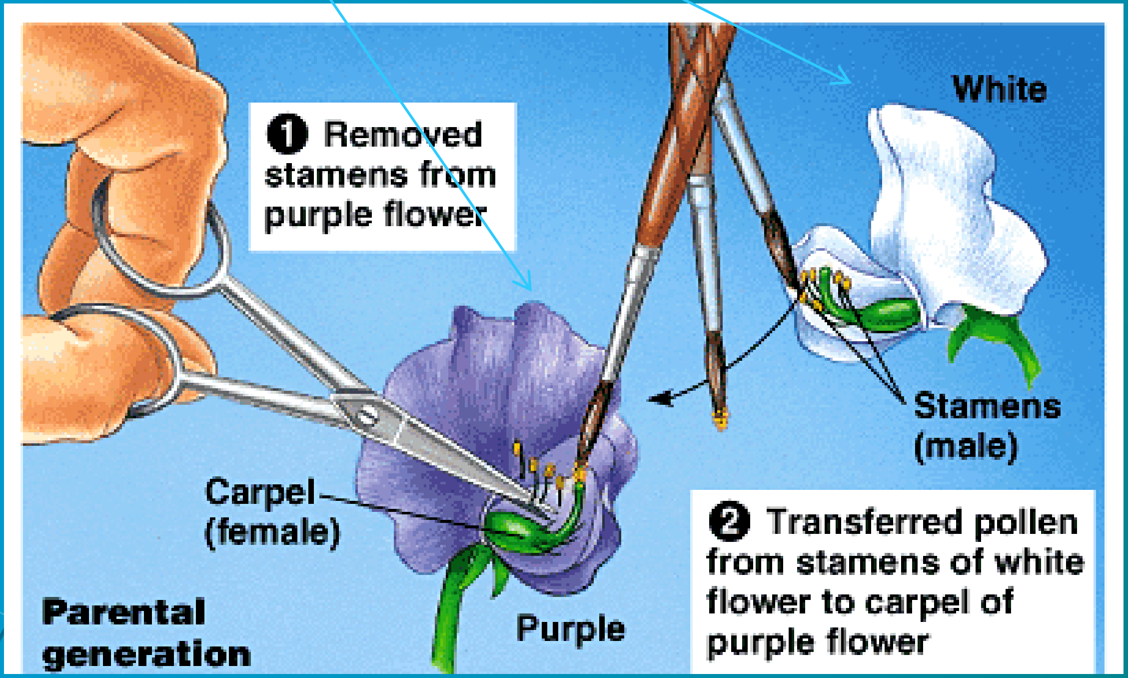
- Question:
  - How is an allele different from a gene? Give an example of each.

Genotype vs. Phenotype

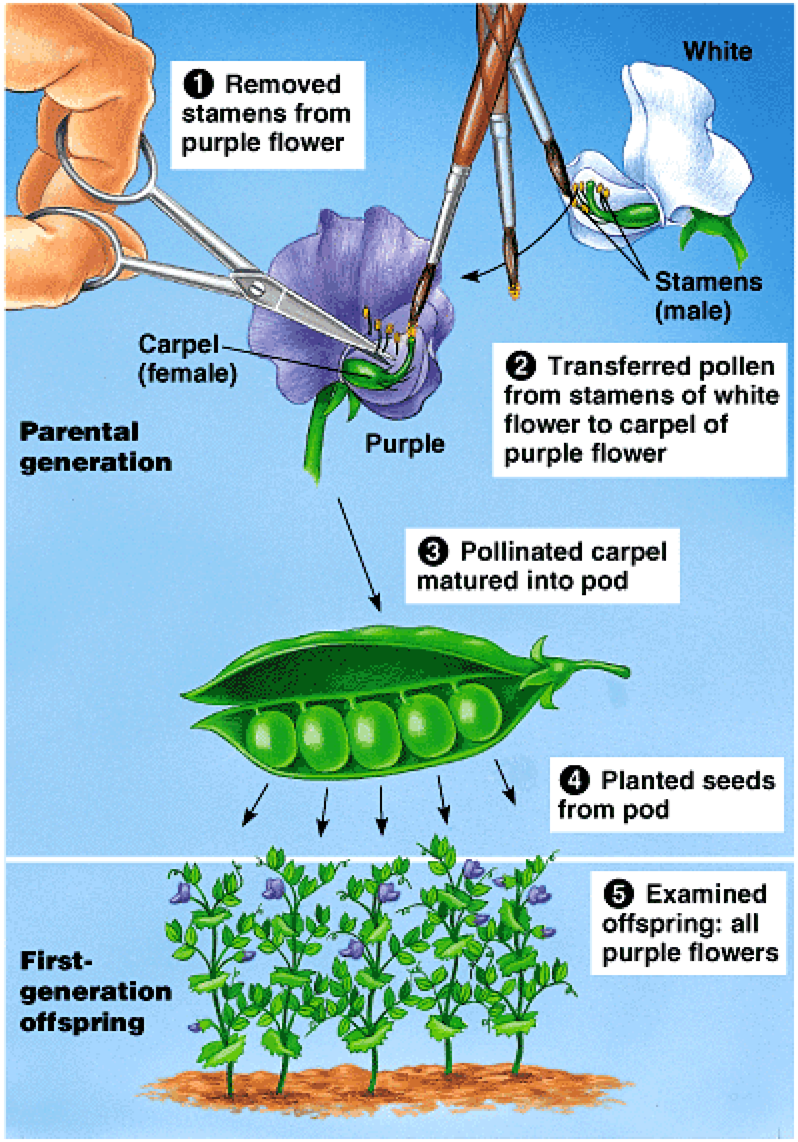
- Genotype = genetic makeup of an individual (think "gene = genotype")
  - Example: the genotype for a pure tall pea plant would be TT
- Phenotype = the expression of the genotype or the physical characteristics that you can see (think "physical = phenotype")
  - Example: If the genotype for height in a pea plant is tt, then you will see a short pea plant.

Remember Mendel's cross?

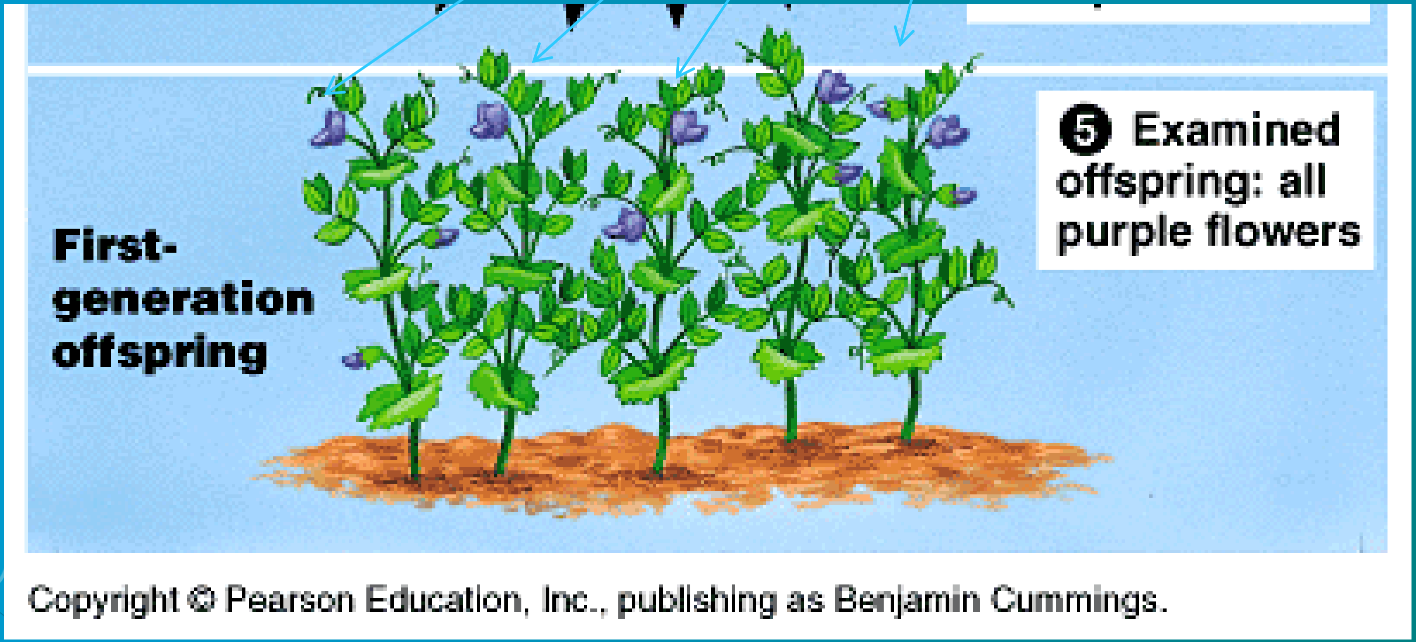
- What color do you think the offspring flowers will be?



- The hybrid plants only showed the trait of one parent plant—they were all **PURPLE**!



- The purple allele is DOMINANT over the white allele because the hybrid plants were all purple.



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.

	P	P
p	Pp	Pp
p	Pp	Pp

Homozygous-recessive

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

Heterozygous - hybrid

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

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

	R	R
W	RW	RW
W	RW	RW

Homozygous

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_AI_B	I_BI_O
I_O	I_AI_O	I_OI_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.

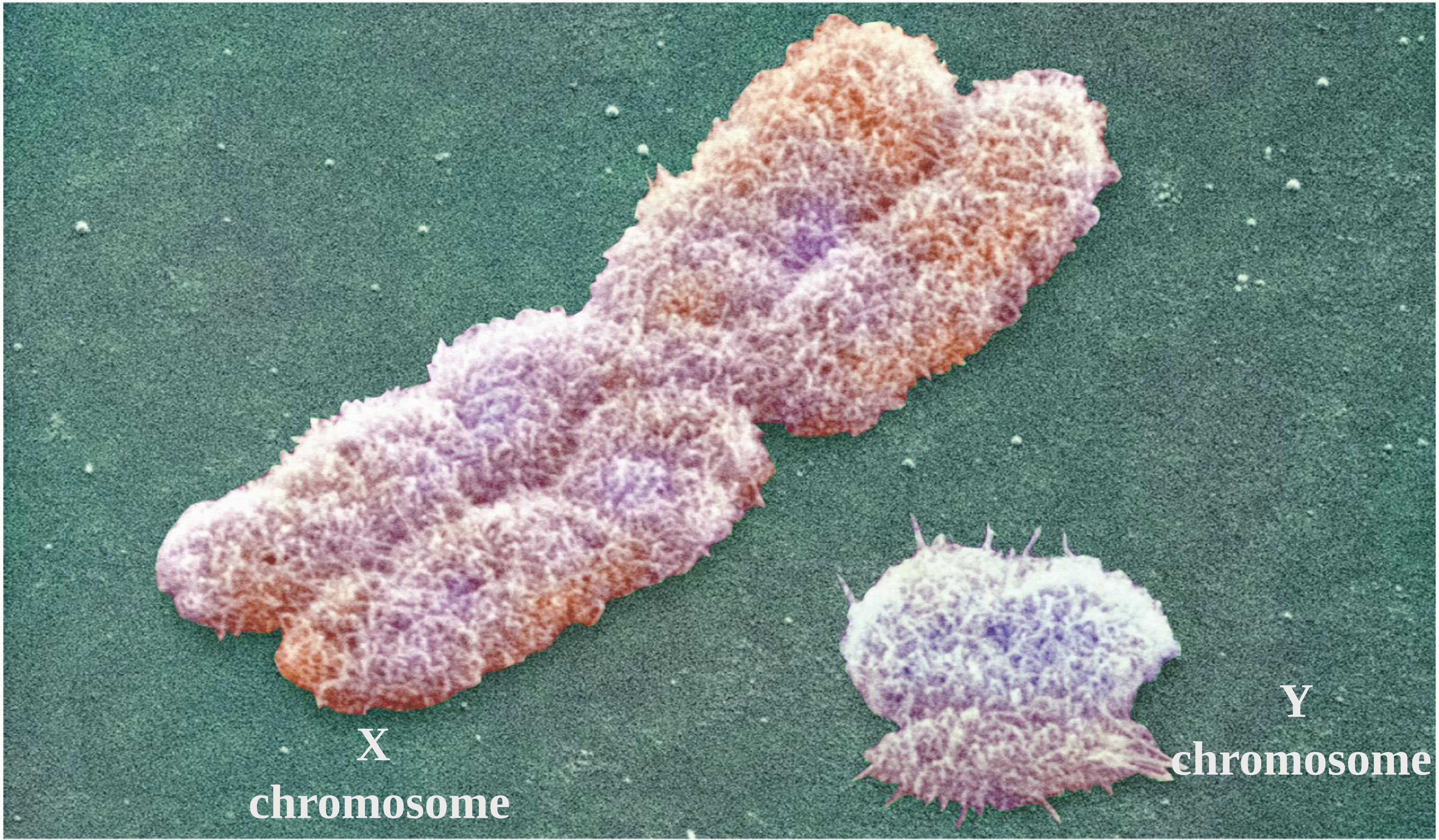
Incomplete Dominance

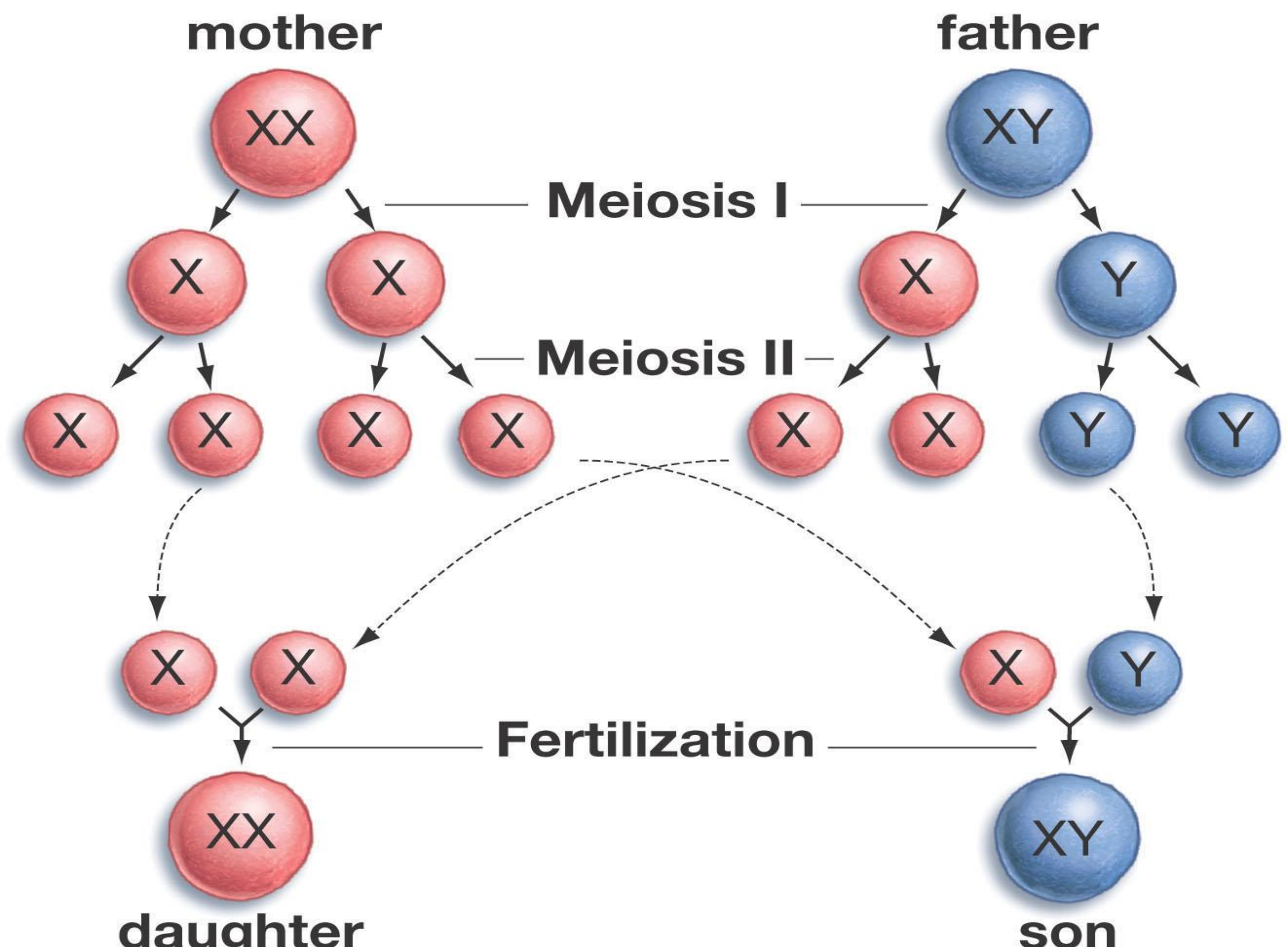
Ex.

Stop (Go To Lesson 2 & 3)

Sex-Linked Traits

Boy or Girl? The Y Chromosome "Decides"




mother
father
XX
XY
Meiosis I
X
X
X
Y
Meiosis II
X
X
X
X
X
X
Y
Y
X
X
X
Y
Fertilization
XX
XY
daughter
son

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?

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

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

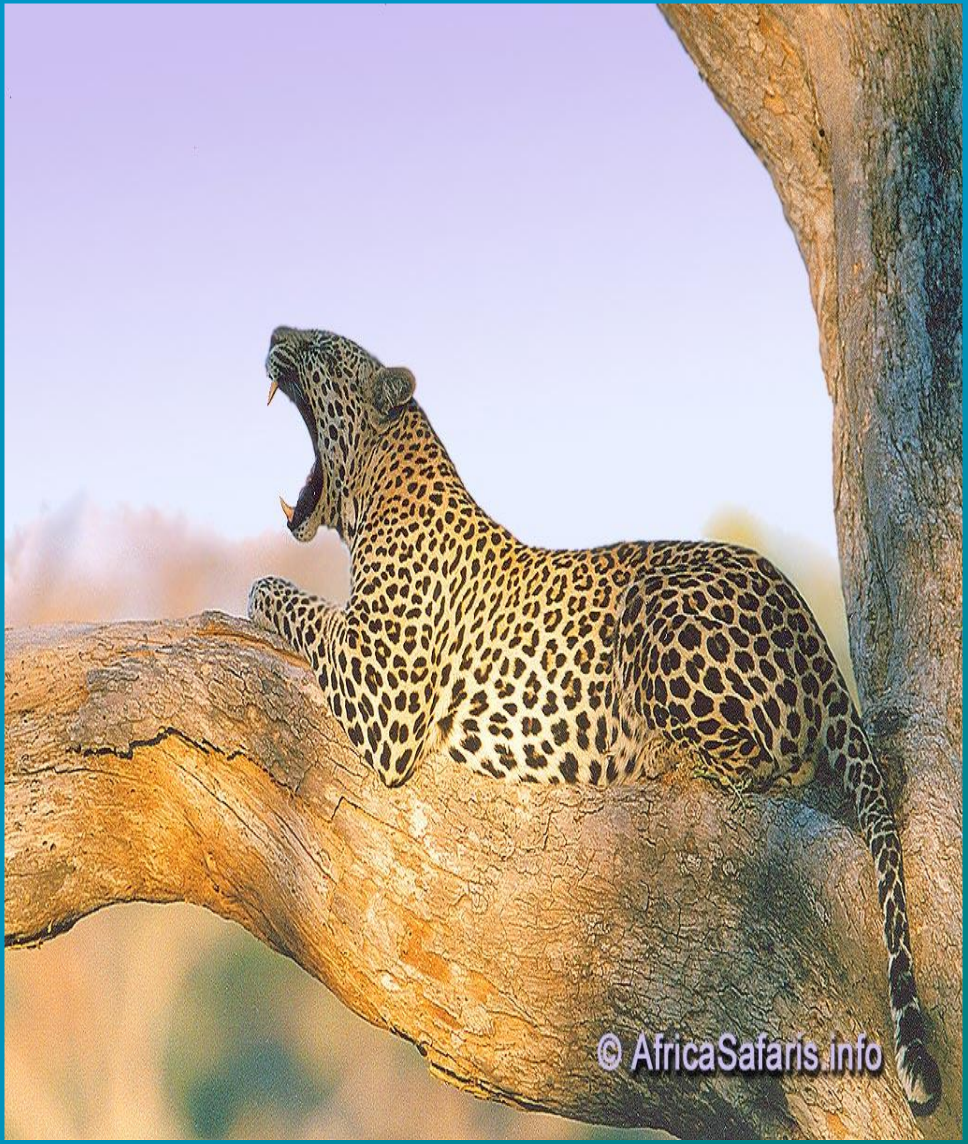
- Genetically diverse populations are **more likely to survive** changing environments.
- Greater variation within the species makes a population **better suited to adaptation** to changes in the environment.

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

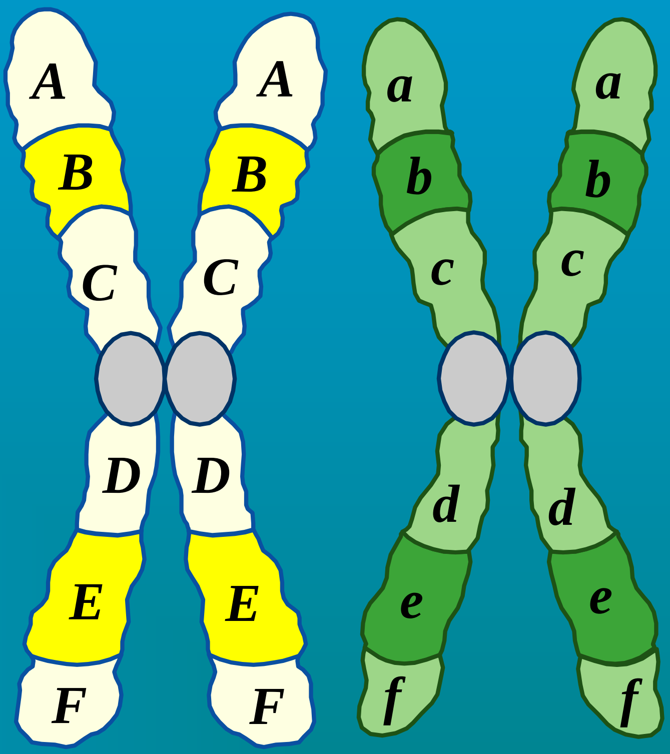
Crossing-over

•the physical exchange of chromosomal material between chromatids of homologous chromosomes.

•Result: Generation of new combinations of genes (alleles).

RECOMBINATION

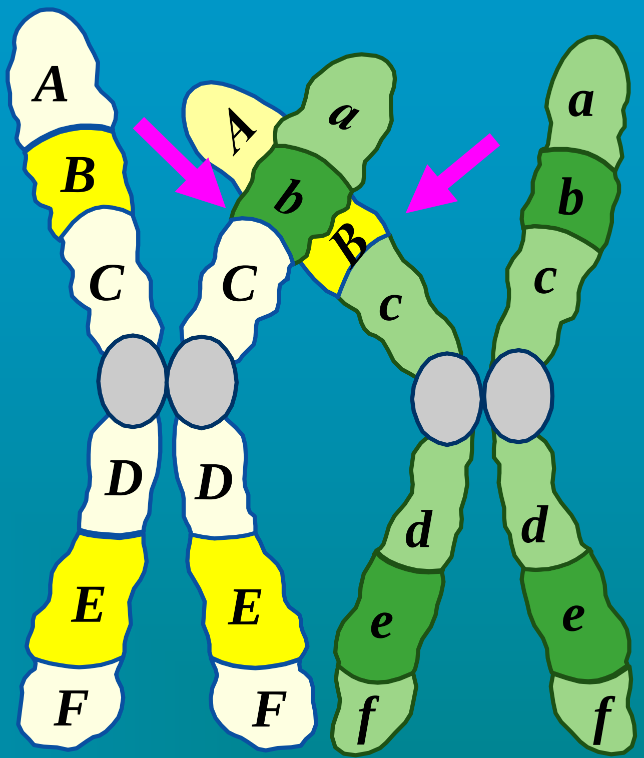
- Occurs in prophase of meiosis I
- Generates diversity



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

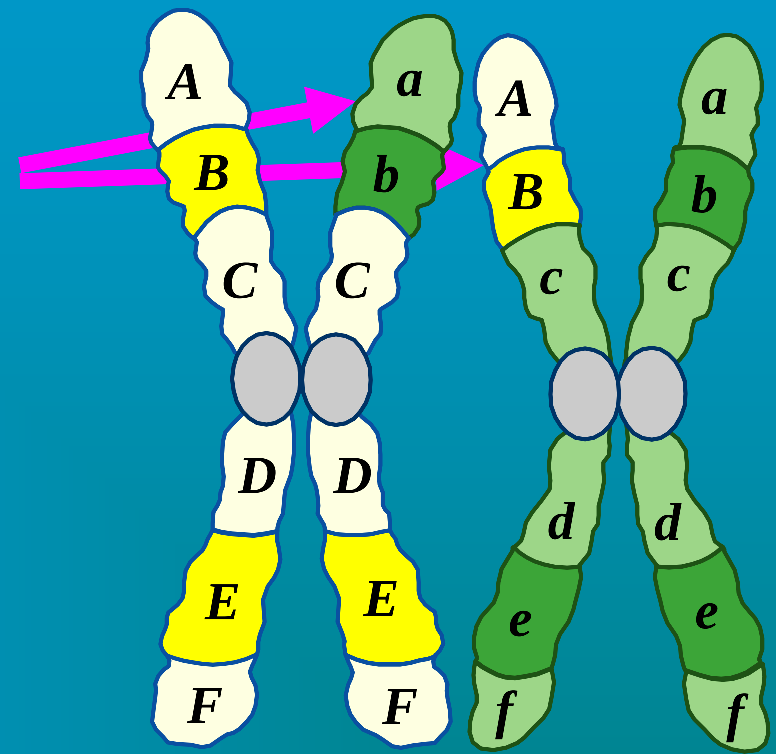
RECOMBINATION

Letters denote genes
Case denotes alleles

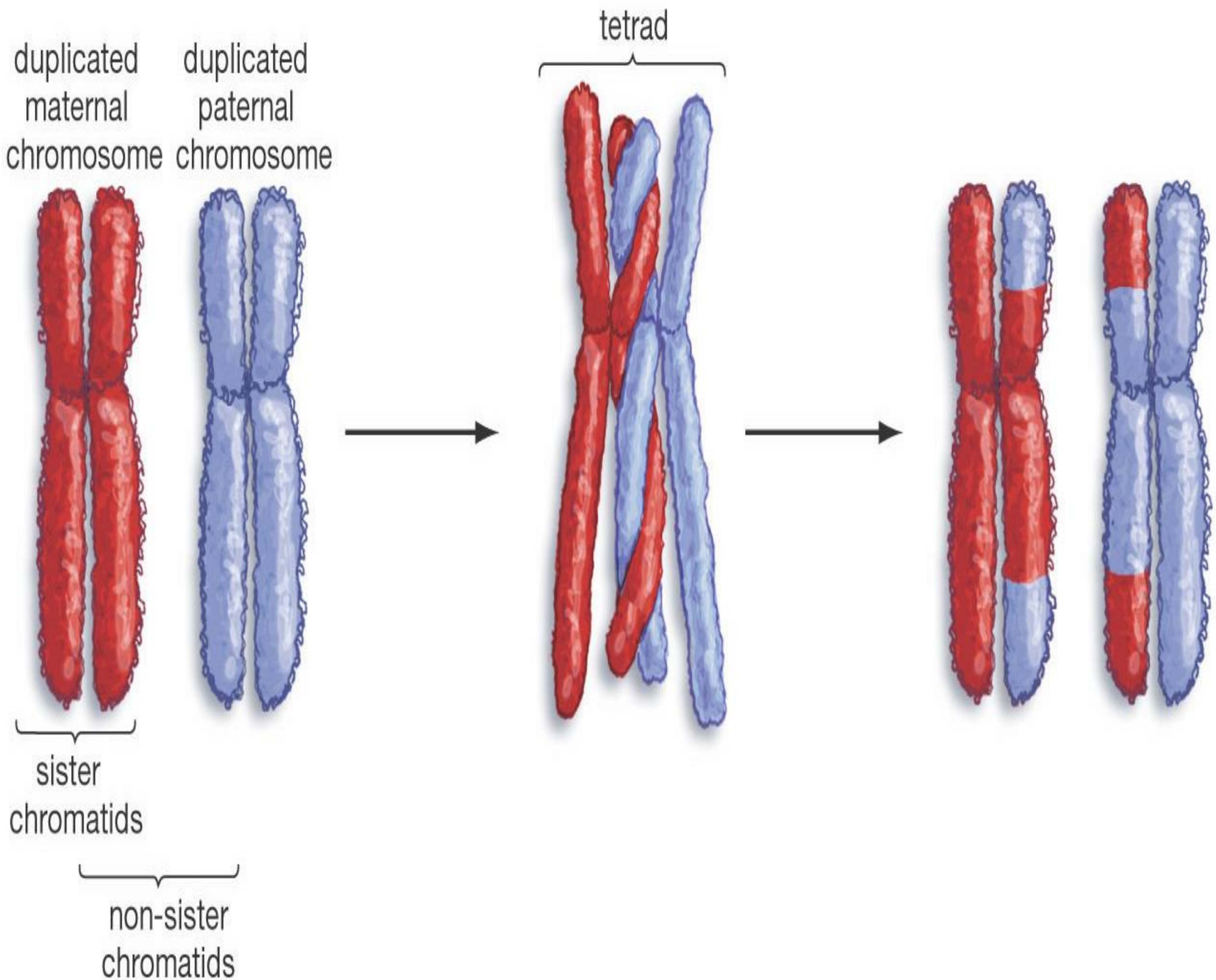


RECOMBINATION

Alleles have crossed over to produce new gene combinations



Exchange of parts of non-sister chromatids.



Genetic Diversity

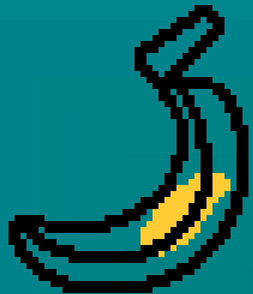
- Sometimes entire chromosomes can be added or deleted, resulting in a genetic disorder such as Trisomy 21 (Down syndrome).

Genetic Diversity

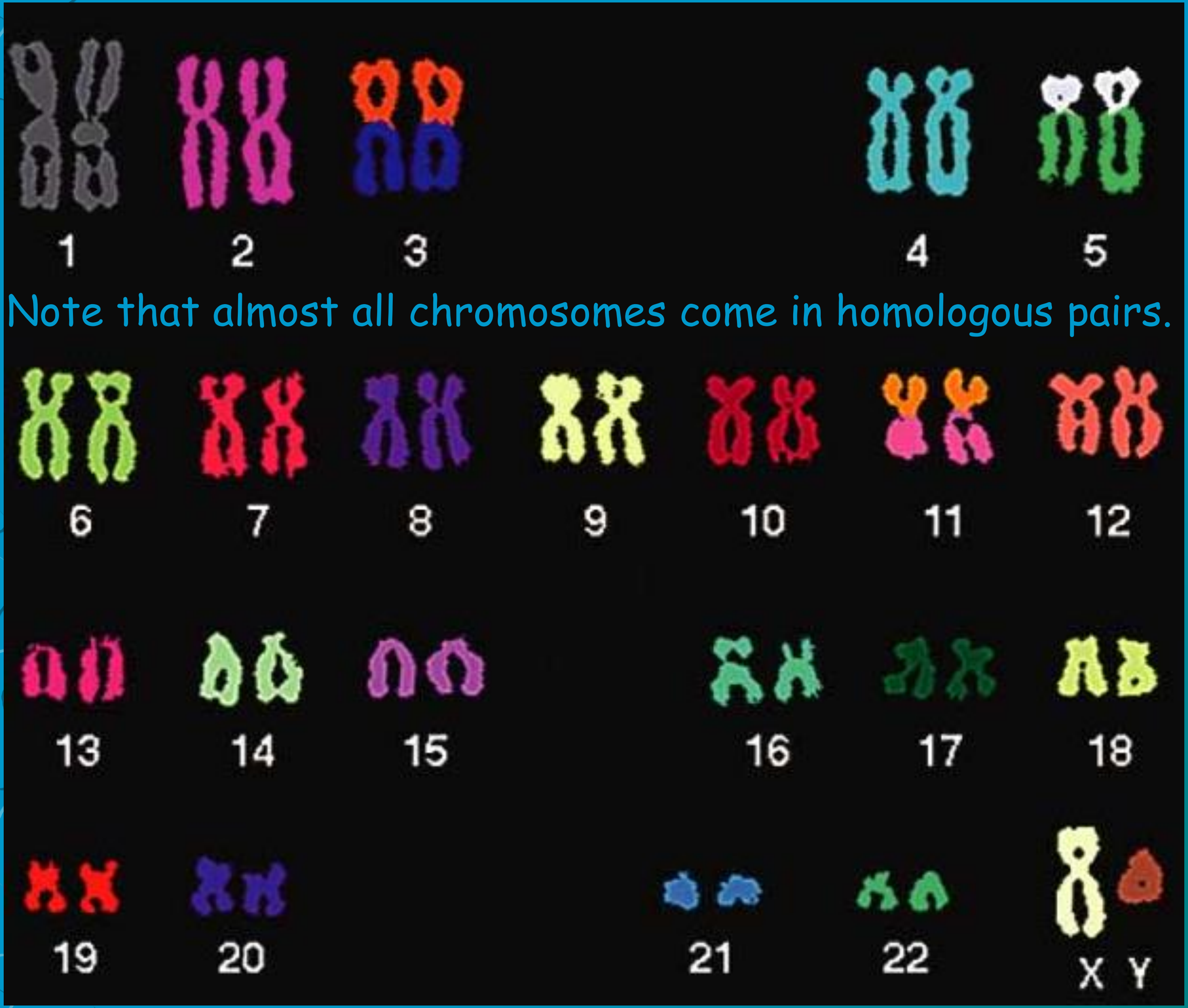
- Chromosomal Errors
- **NONDISJUNCTION:** the failure of chromosomes to separate properly in meiosis. Gametes with extra or too few chromosomes result.
- Can cause diseases such as Down's Syndrome.

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



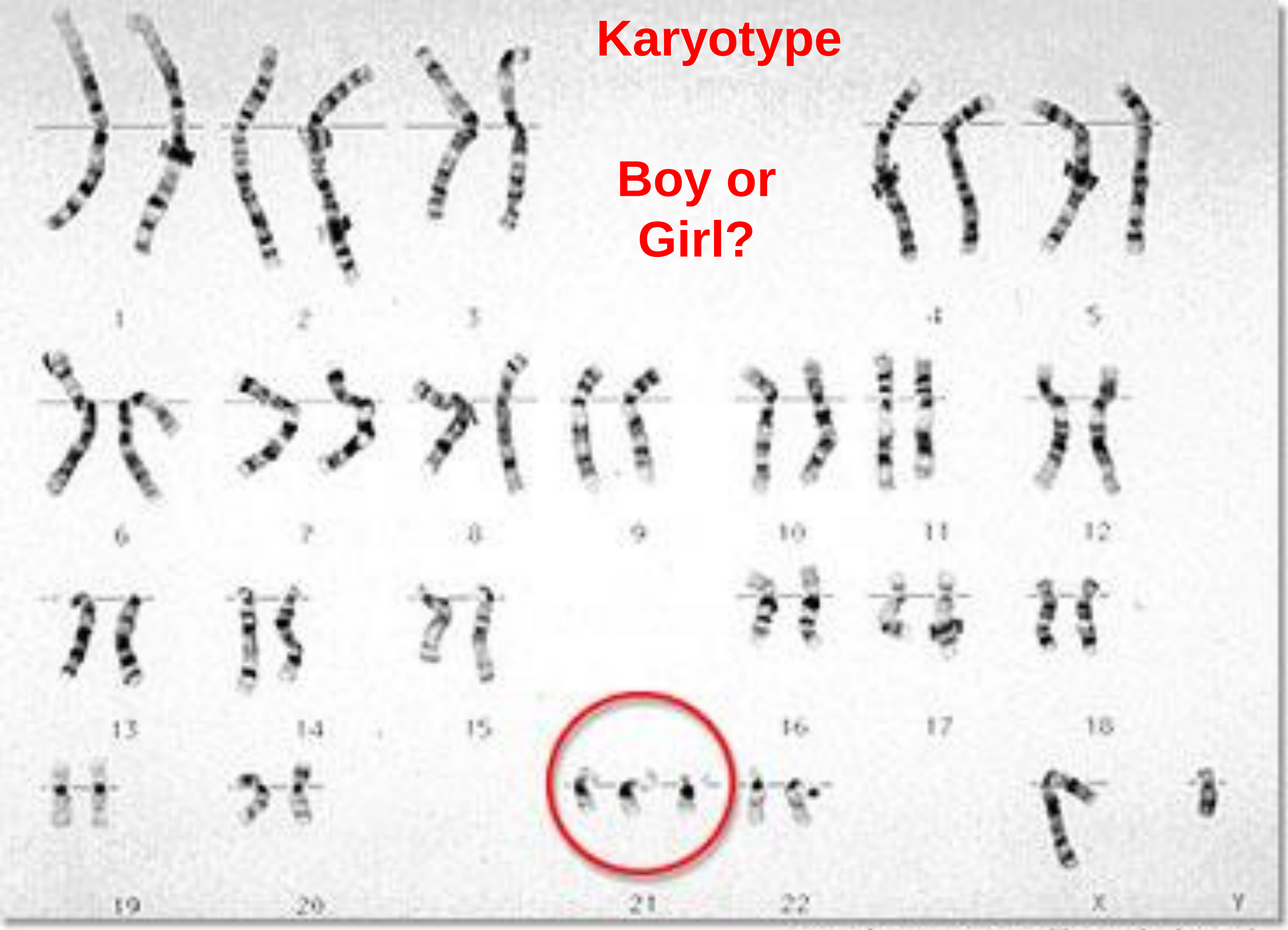
A Karyotype is an Informative, Arranged Picture of Chromosomes At Their Most Condensed State



Boy or girl?

Karyotype

Boy or Girl?



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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

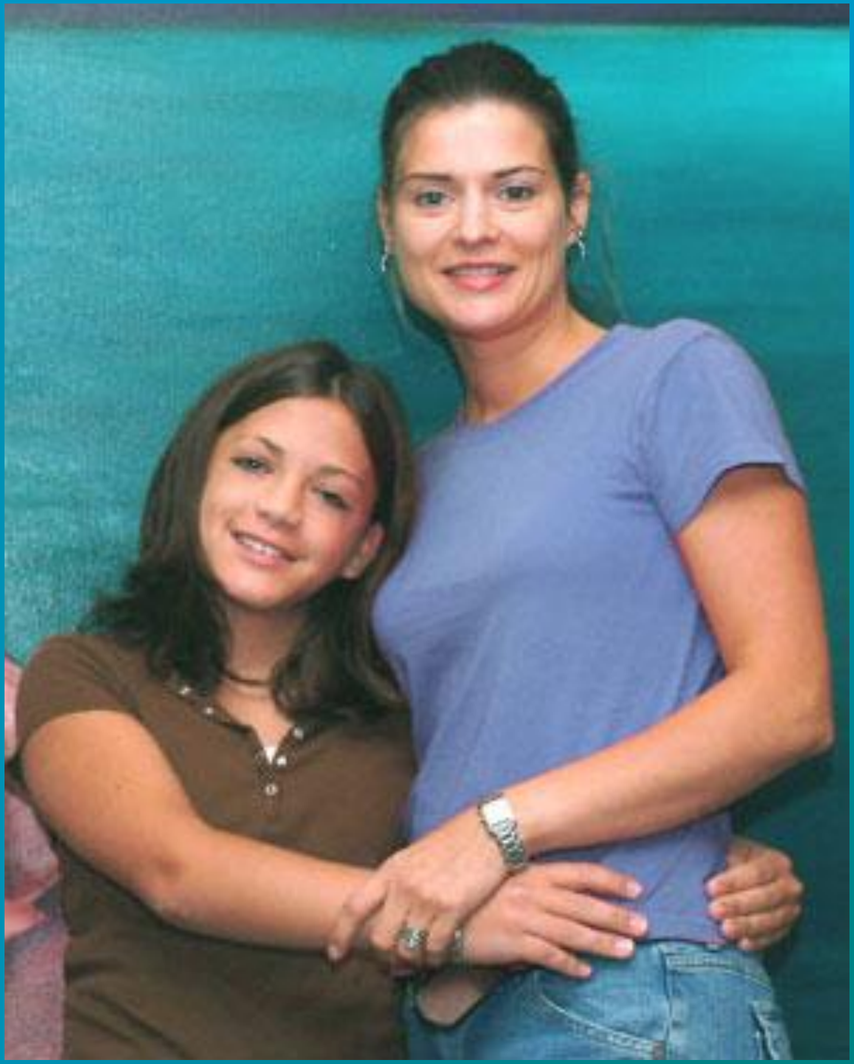
Turner's syndrome

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

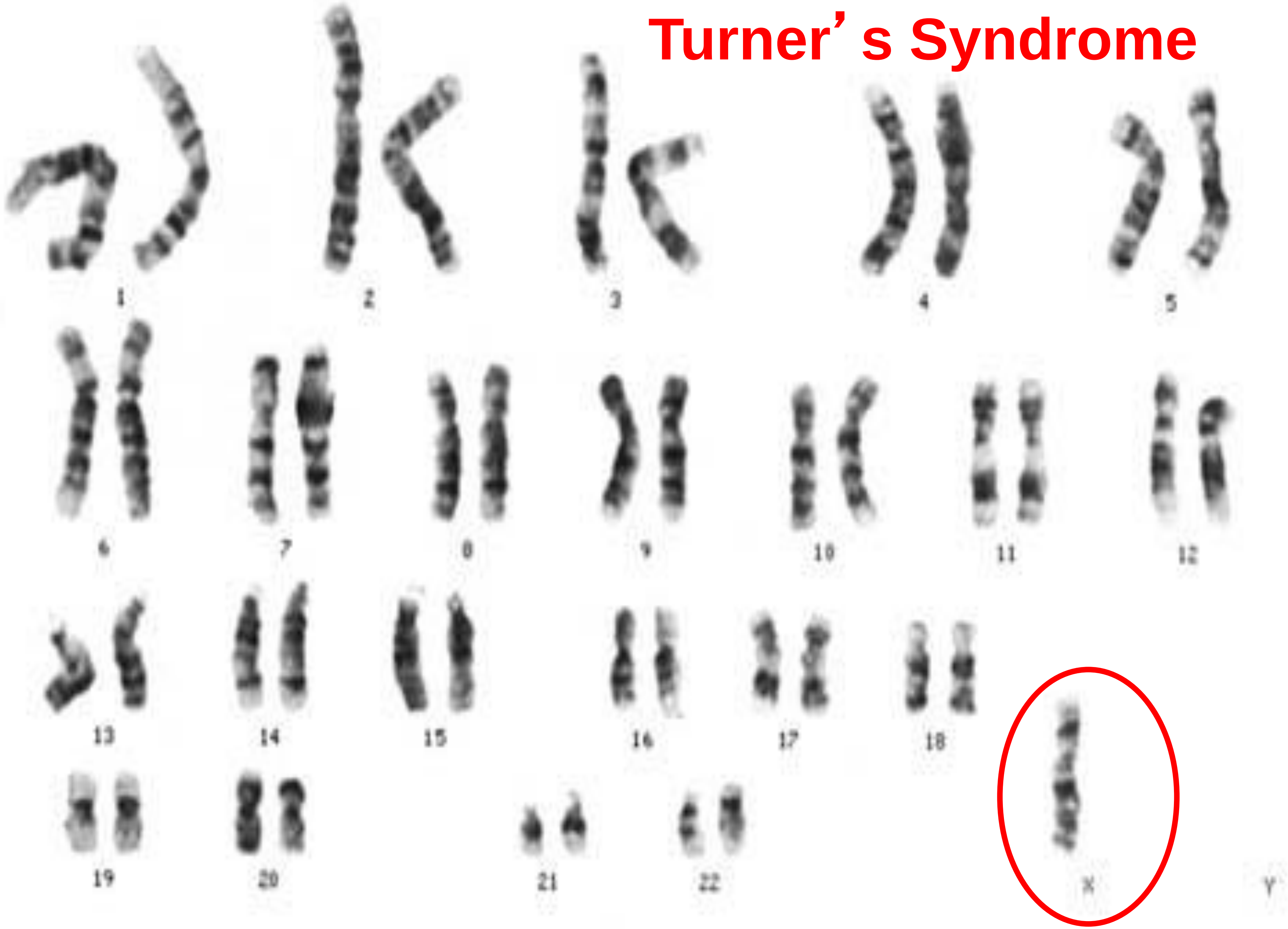
Genetic Diseases

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.

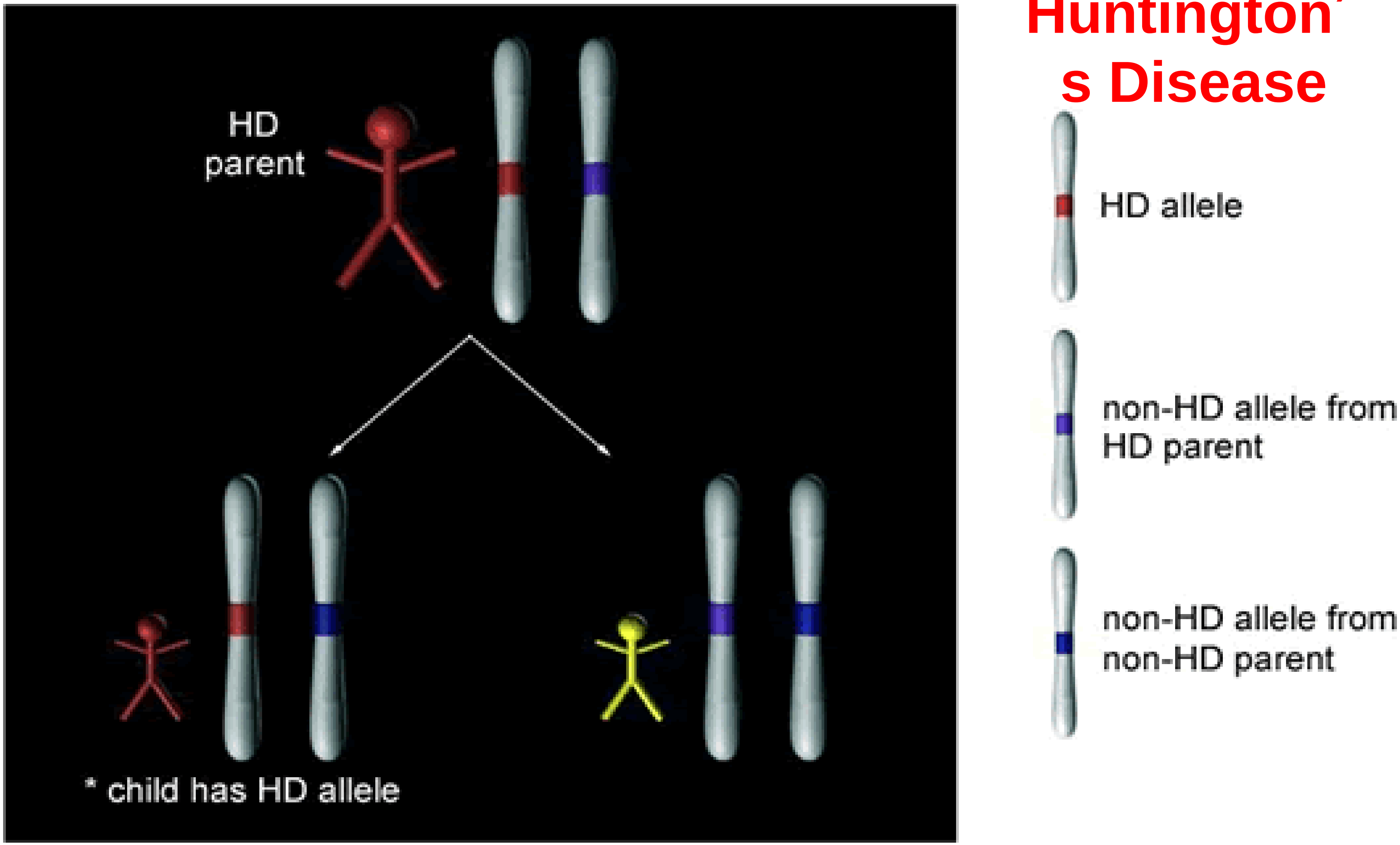
Genetic Diseases

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.

Huntington’s Disease

Figure C-2: Risk for child of HD individual



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.

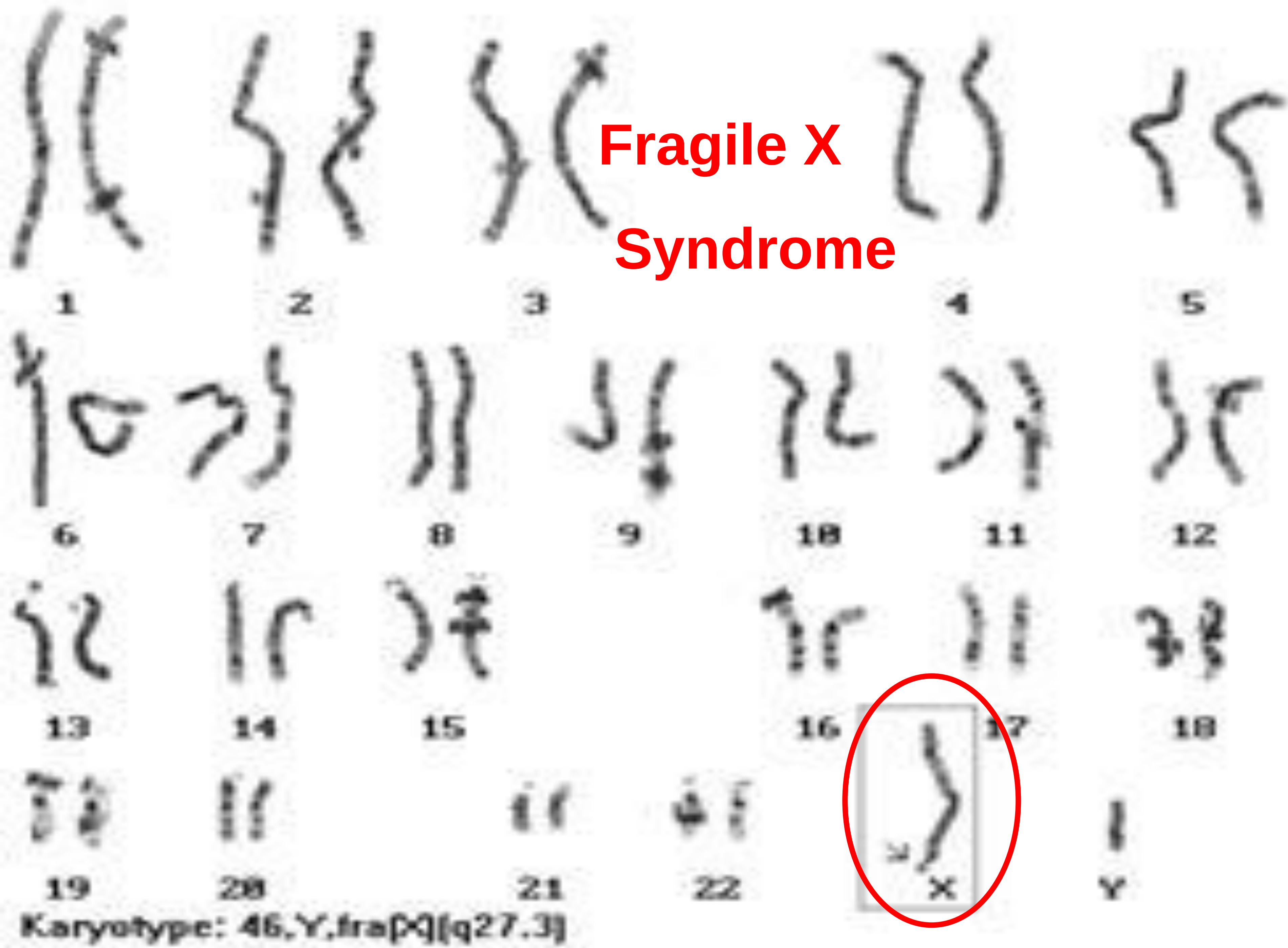
Genetic Diseases

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.

Fragile X Syndrome

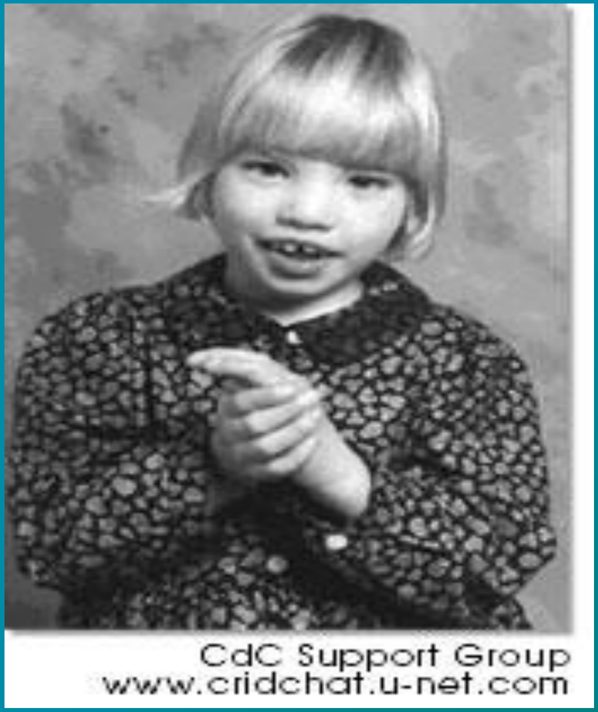


Genetic Diseases

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.

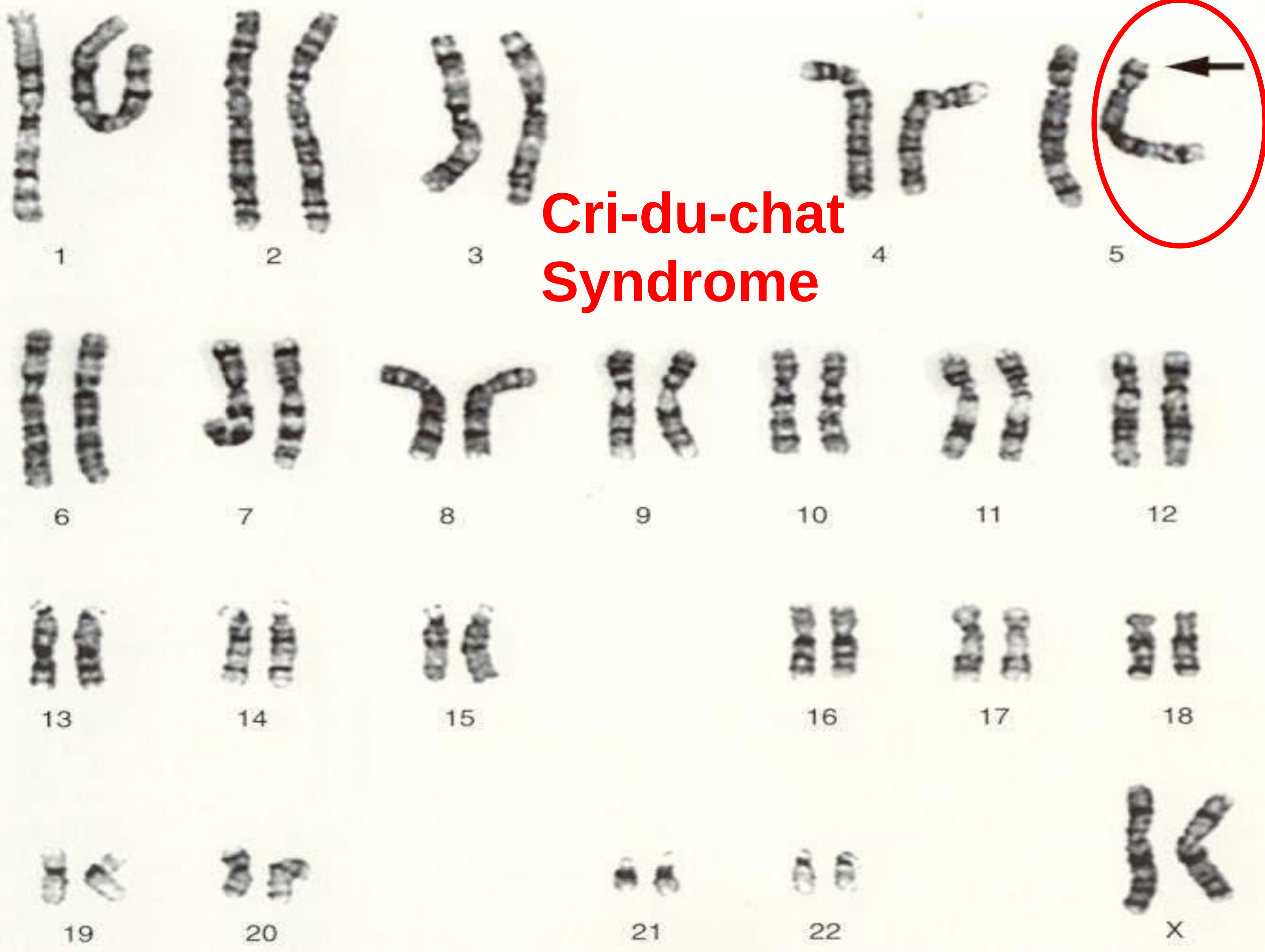


Genetic Diseases

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

Cri-du-chat Syndrome



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.

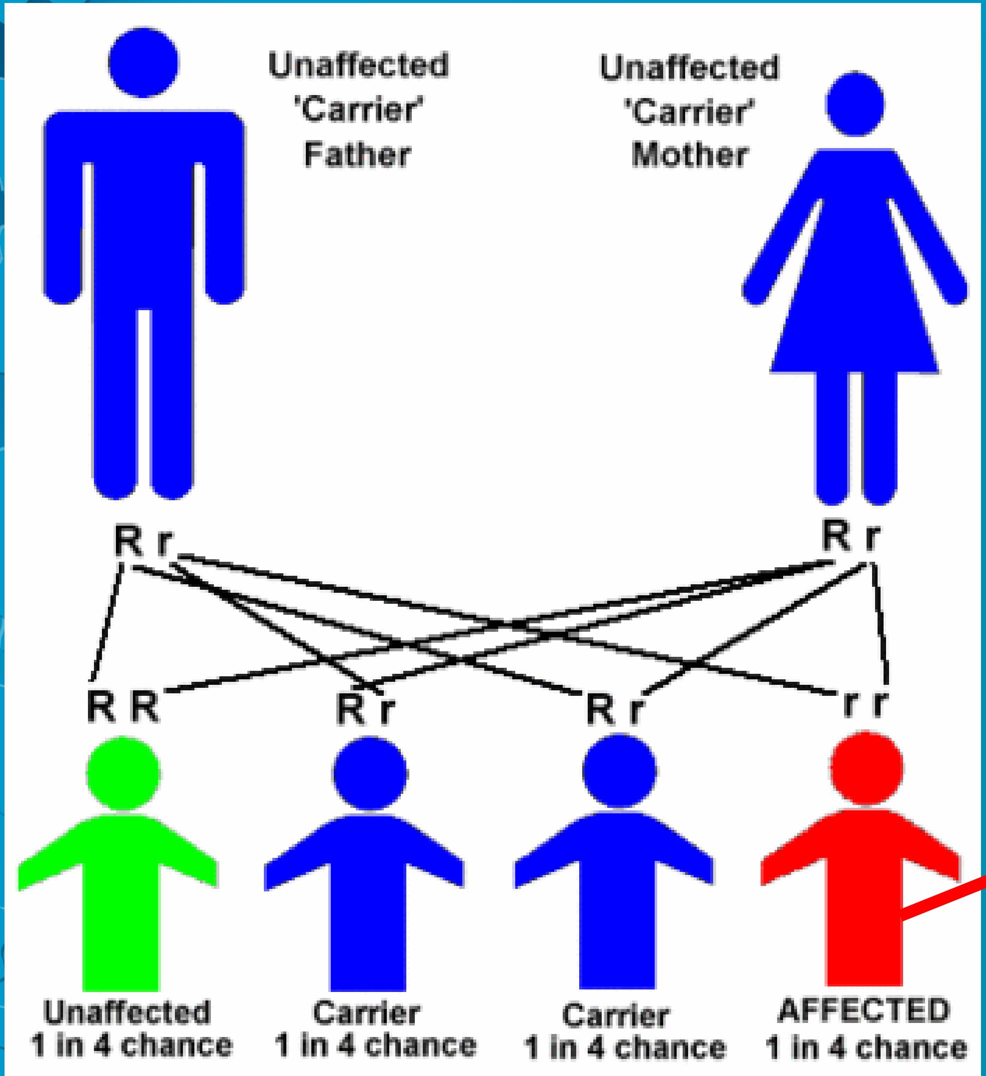
Genetic Diseases

Tay-Sachs Disease

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

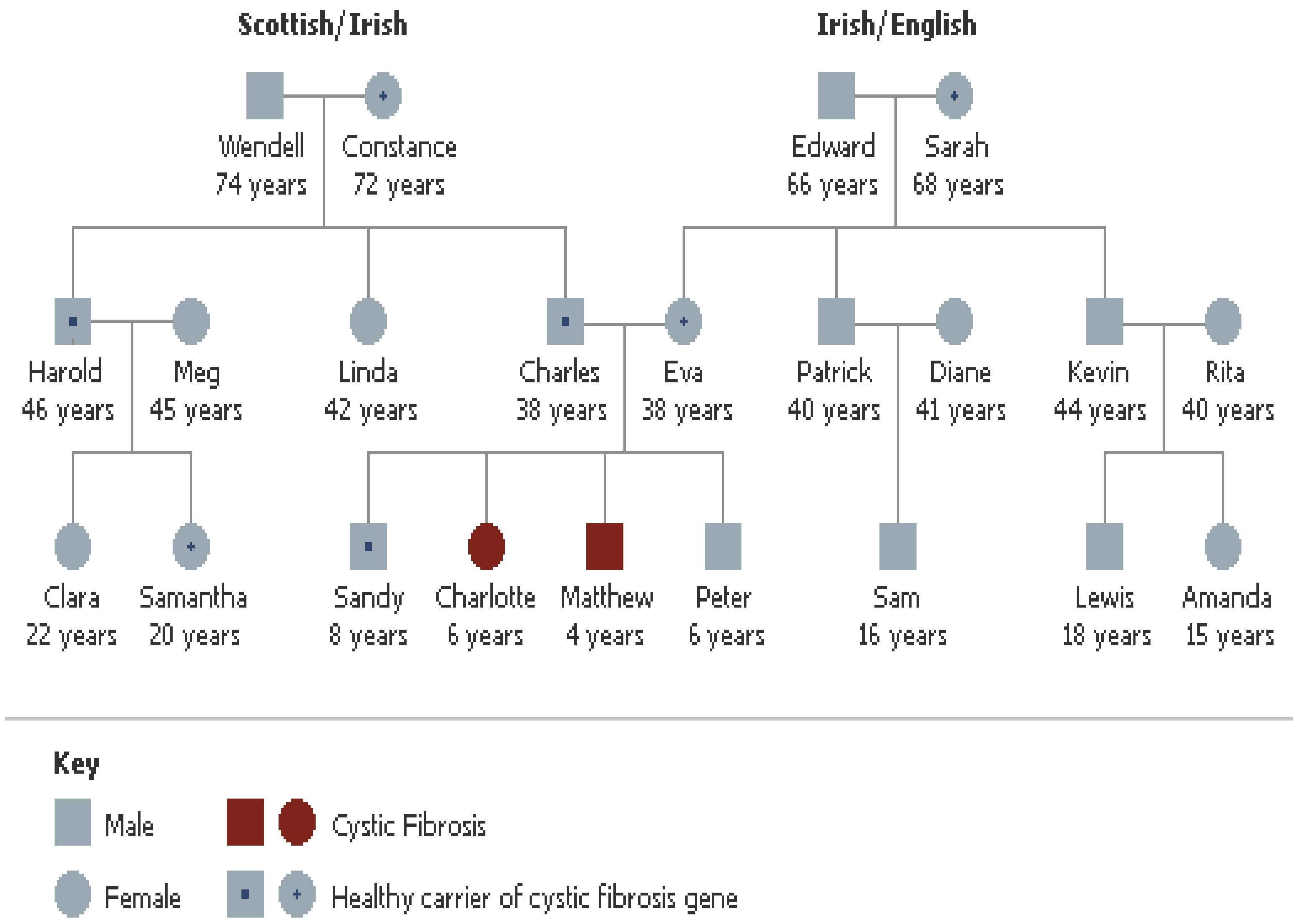
Genetic Diseases

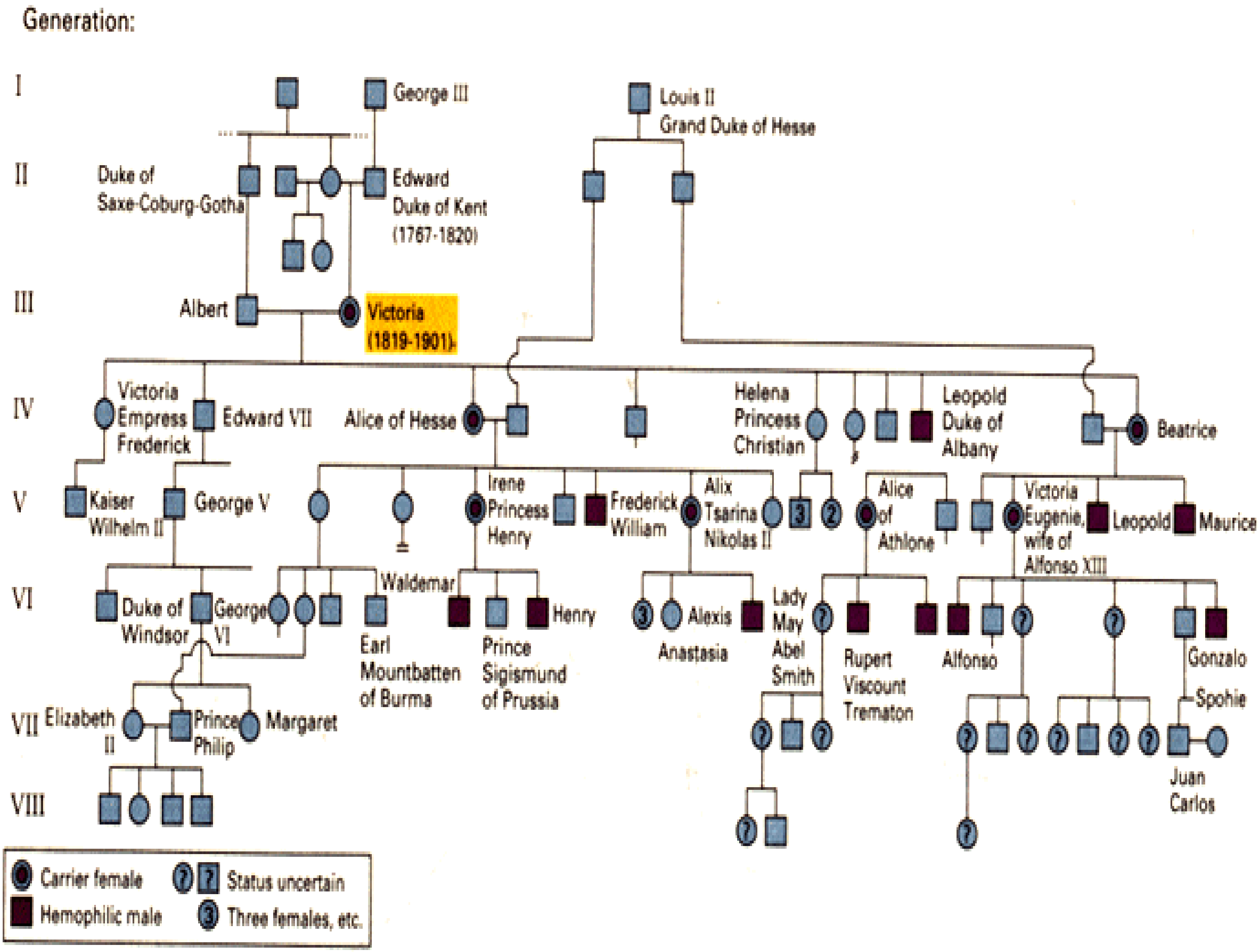
Tay-Sachs Disease



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.


Scottish/Irish
Irish/English
Wendell 74 years
Constance 72 years
Edward 66 years
Sarah 68 years
Harold 46 years
Meg 45 years
Linda 42 years
Charles 38 years
Eva 38 years
Patrick 40 years
Diane 41 years
Kevin 44 years
Rita 40 years
Clara 22 years
Samantha 20 years
Sandy 8 years
Charlotte 6 years
Matthew 4 years
Peter 6 years
Sam 16 years
Lewis 18 years
Amanda 15 years
Key
Male
Female
Cystic Fibrosis
Healthy carrier of cystic fibrosis gene



Generation:
I
II
III
IV
V
VI
VII
VIII
George III
Louis II
Grand Duke of Hesse
Duke of
Saxe-Coburg-Gotha
Edward
Duke of Kent
(1767-1820)
Albert
Victoria
(1819-1901)
Victoria
Empress
Frederick
Edward VII
Alice of Hesse
Helena
Princess
Christian
Leopold
Duke of
Albany
Beatrice
Kaiser
Wilhelm II
George V
Irene
Princess
Henry
Frederick
William
Alix
Tsarina
Nikolas II
Alice
of
Athlone
Victoria
Eugenie,
wife of
Alfonso XIII
Leopold
Maurice
Duke of
Windsor
George
VI
Waldemar
Earl
Mountbatten
of Burma
Prince
Sigismund
of Prussia
Henry
Alexis
Anastasia
Lady
May
Abel
Smith
Rupert
Viscount
Trematon
Alfonso
Gonzalo
Spohie
Elizabeth
II
Prince
Philip
Margaret
Juan
Carlos
Carrier female
Hemophilic male
Status uncertain
Three females, etc.

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