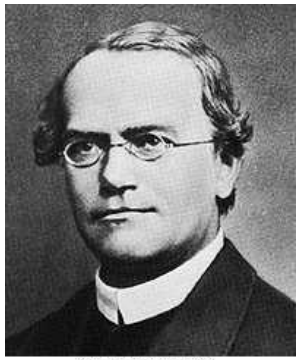
IMPACT OF GENETIC CONDITION ON FAMILIES

Lutfi Nurdian A

Mendelian Theory

Gregor Johann Mendel

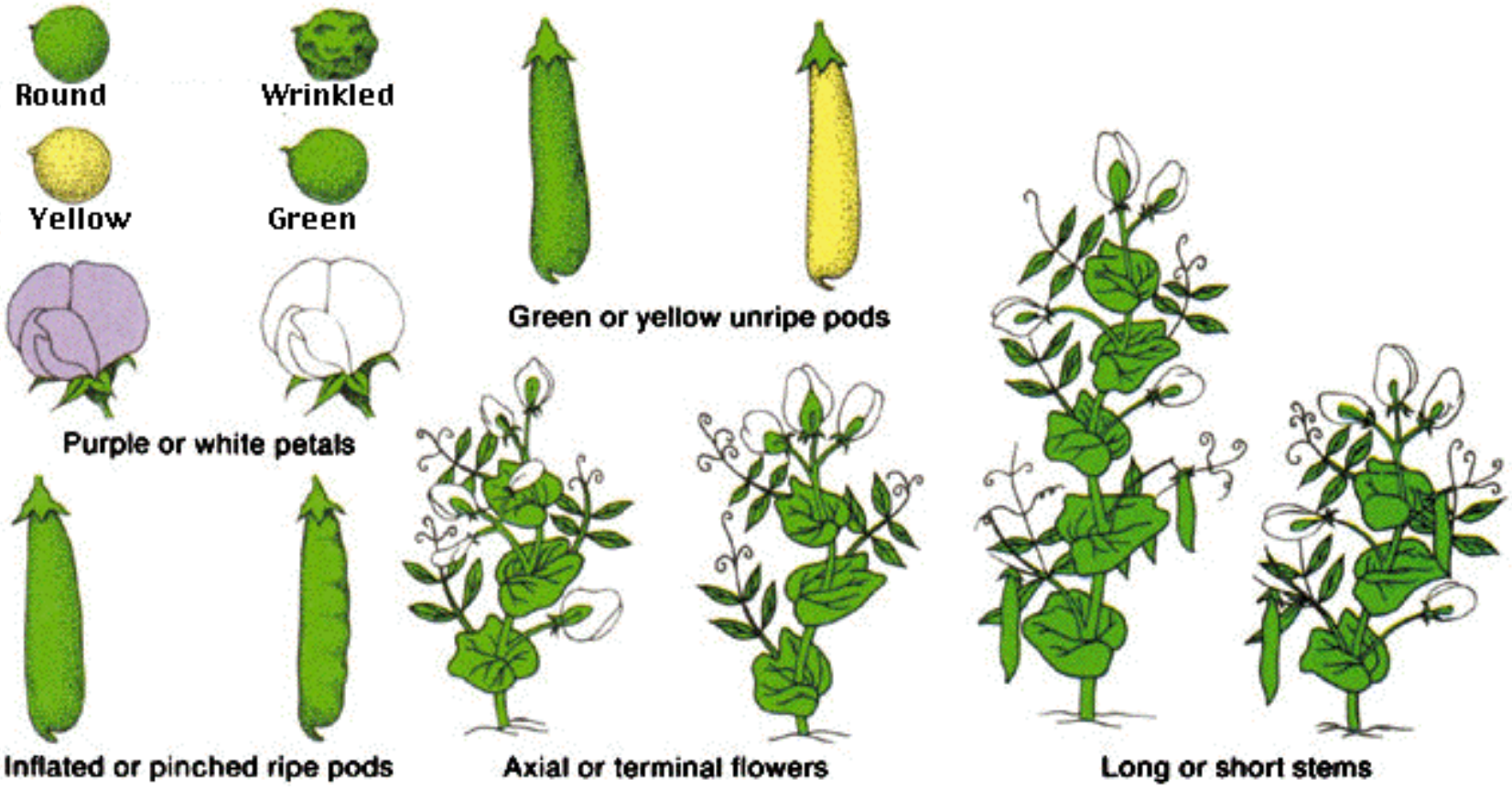
- Austrian Monk, born in what is now Czech Republic in 1822


Gregor Mendel

- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.
- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a "blending" process and the offspring were essentially a "dilution"of the different parental characteristics.

Mendel's peas

- Mendel looked at seven traits or characteristics of pea plants:



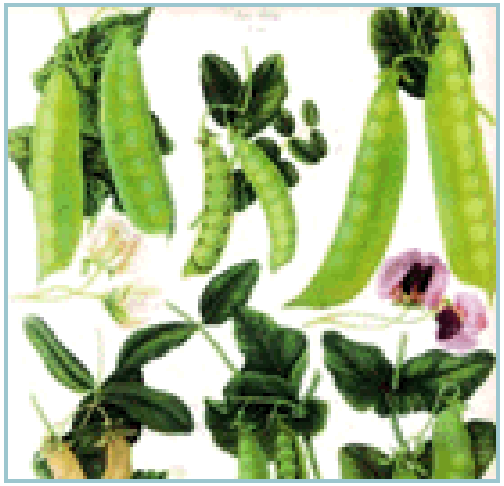
- In 1866 he published *Experiments in Plant Hybridization*, (*Versuche über Pflanzen-Hybriden*) in which he established his three Principles of Inheritance
- He tried to repeat his work in another plant, but didn't work because the plant reproduced asexually! If…
- Work was largely ignored for 34 years, until 1900, when 3 independent botanists rediscovered Mendel's work.

- Mendel was the first biologist to use Mathematics – to explain his results quantitatively.
- Mendel predicted

  The concept of genes

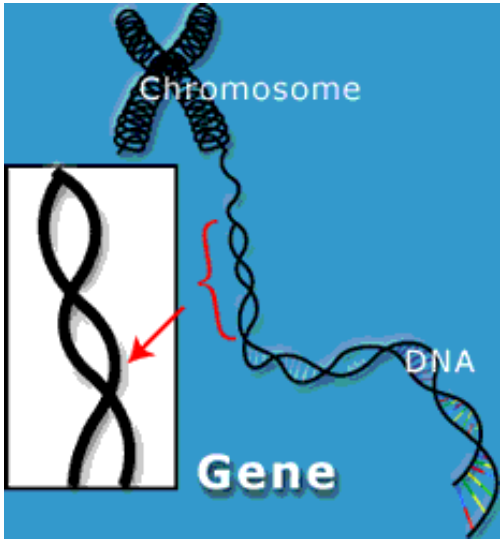
  That genes occur in pairs

  That one gene of each pair is present in the gametes



Genetics terms you need to know:

- **Gene** – a unit of heredity; a section of DNA sequence encoding a single protein
- **Genome** – the entire set of genes in an organism



- **Alleles** – two genes that occupy the same position on homologous chromosomes and that cover the same trait (like ‘flavors’ of a trait).
- **Locus** – a fixed location on a strand of DNA where a gene or one of its alleles is located.

- **Homozygous** – having identical genes (one from each parent) for a particular characteristic.
- **Heterozygous** – having two different genes for a particular characteristic.

- **Dominant** – the allele of a gene that masks or suppresses the expression of an alternate allele; the trait appears in the heterozygous condition.
- **Recessive** – an allele that is masked by a dominant allele; does not appear in the heterozygous condition, only in homozygous.

- **Genotype** – the genetic makeup of an organisms
- **Phenotype** – the physical appearance of an organism (Genotype + environment)

- **Monohybrid cross**: a genetic cross involving a single pair of genes (one trait); parents differ by a single trait.
- **P** = Parental generation
- $\mathbf{F_1}$ = First filial generation; offspring from a genetic cross.
- $\mathbf{F_2}$ = Second filial generation of a genetic cross

Monohybrid cross

- Parents differ by a single trait.
- Crossing two pea plants that differ in stem size, one tall one short

  T = allele for Tall

  t = allele for dwarf

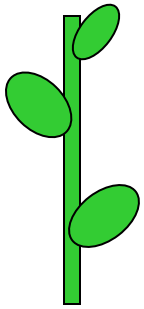
  TT = homozygous tall plant

  t t = homozygous dwarf plant

T T × t t

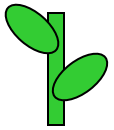
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



T T × *t t*

(tall) (dwarf)

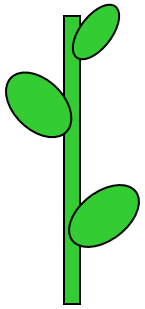


↓

F_1 generation
is heterozygous:

T *t*

(all tall plants)



Using a Punnett Square

STEPS:

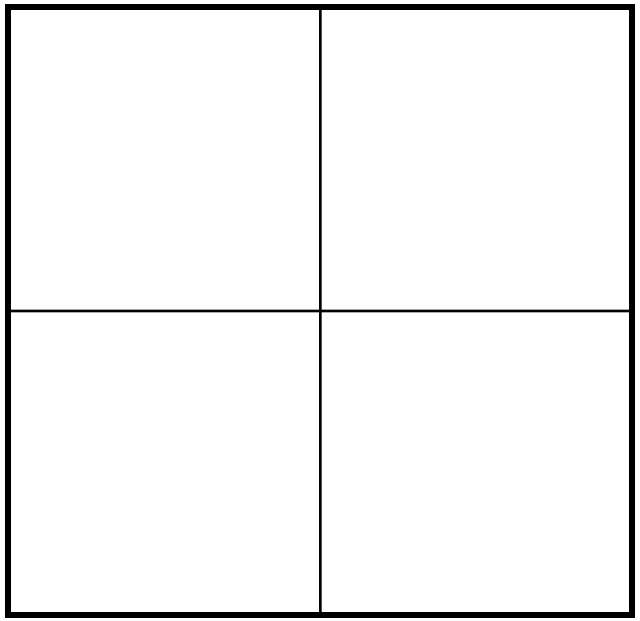
1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

Parent genotypes:

TT and *t t*

Cross

T T × ***t t***



Punnett square

4. **"split" the letters of the genotype for each parent & put them "outside" the p-square**
5. determine the possible genotypes of the offspring by filling in the p-square
6. summarize results (genotypes & phenotypes of offspring)

T T × ***t t***

	T	**T**
t	**T *t***	**T *t***
t	**T *t***	**T *t***

Genotypes:
100% T t

Phenotypes:
100% Tall plants

Monohybrid cross: F_2 generation

- If you let the F1 generation self-fertilize, the next monohybrid cross would be:

T *t* × **T *t***

(tall) (tall)

	T	***t***
T	**T T**	**T *t***
t	**T *t***	***t t***

Genotypes:

1 TT= Tall

2 T*t* = Tall

1 *tt* = dwarf

Genotypic ratio= 1:2:1

Phenotype:

3 Tall

1 dwarf

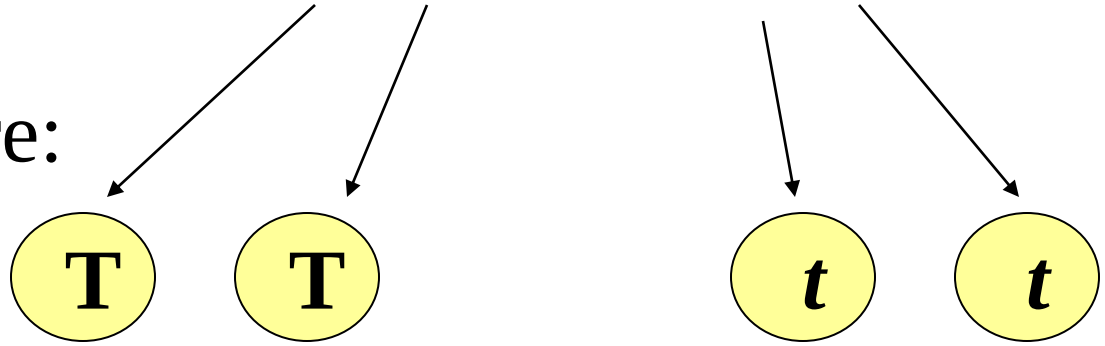
Phenotypic ratio= 3:1

Secret of the Punnett Square

- Key to the Punnett Square:
- Determine the gametes of each parent…
- How? By "splitting" the genotypes of each parent:

If this is your cross **T T** × ***t t***

The gametes are:



Once you have the gametes…

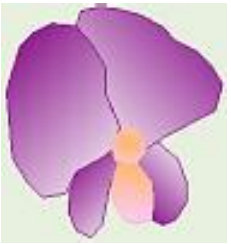
T **T** × ***t*** ***t***

	t	***t***
T	**T *t***	**T *t***
T	**T *t***	**T *t***

Another example: Flower color

For example, flower color:

P = purple (dominant)



p = white (recessive)

If you cross a homozygous Purple (PP) with a homozygous white (*pp*):

P P × *p p*

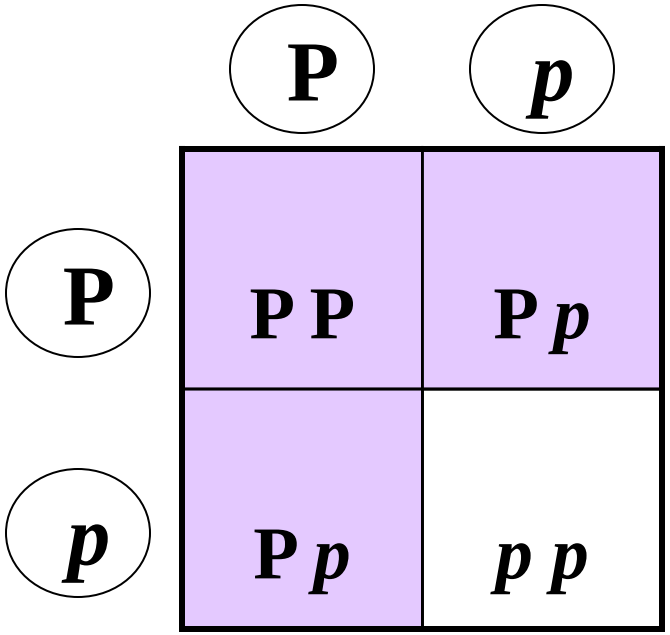
↓

P *p*

ALL PURPLE (P*p*)

Cross the F1 generation:

P *p* × P *p*



Genotypes:
1 PP
2 Pp
1 pp

Phenotypes:
3 Purple
1 White

Mendel's Principles

- **<u>1. Principle of Dominance</u>**:

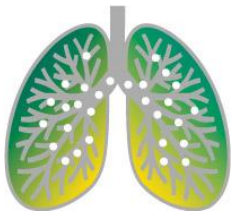
  One allele masked another, one allele was dominant over the other in the F_1 generation.

- **<u>2. Principle of Segregation</u>**:

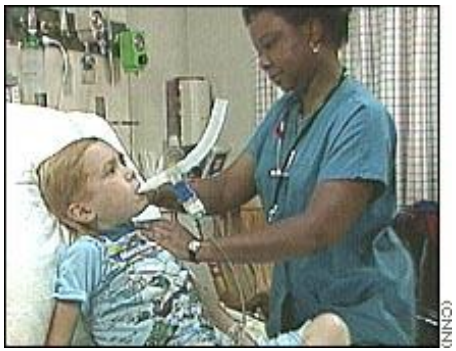
  When gametes are formed, the pairs of hereditary factors (genes) become separated, so that each sex cell (egg/sperm) receives only one kind of gene.

Human case: CF

- Mendel's Principles of Heredity apply universally to all organisms.
- Cystic Fibrosis: a lethal genetic disease affecting Caucasians.
- Caused by mutant recessive gene carried by 1 in 20 people of European descent (12M)
- One in 400 Caucasian couples will be both carriers of CF – 1 in 4 children will have it.



- CF disease affects transport in tissues – mucus is accumulated in lungs, causing infections.



Inheritance pattern of CF

IF two parents carry the recessive gene of Cystic Fibrosis (*c*), that is, they are heterozygous (C *c*), one in four of their children is expected to be homozygous for *cf* and have the disease:

C C = normal
C *c* = carrier, no symptoms
c *c* = has cystic fibrosis

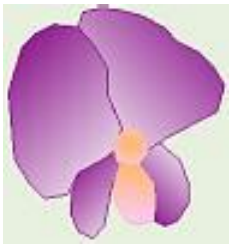
	C	***c***
C	**C C**	**C *c***
c	**C *c***	***c c***

Dihybrid crosses

- Matings that involve parents that differ in **<u>two</u>** genes (two independent traits)

For example, flower color:

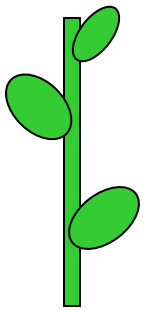
P = purple (dominant)



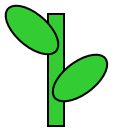
p = white (recessive)

and stem length:

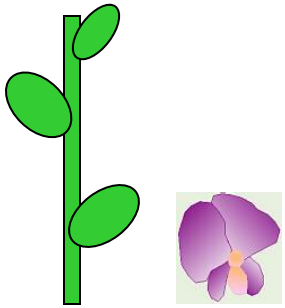
T = tall



t = short

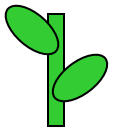


Dihybrid cross: flower color and stem length



TT PP × *tt pp*

(tall, purple) (short, white)



Possible Gametes for parents

T P and *t p*

	tp	*tp*	*tp*	*tp*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

$$T\textit{t}\ P\textit{p} \times T\textit{t}\ P\textit{p}$$

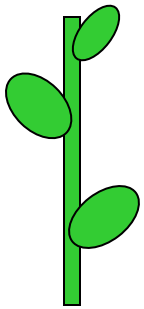
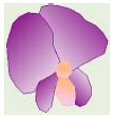
(tall, purple) (tall, purple)

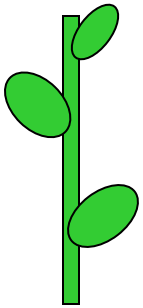
Possible gametes:
TP Tp tP tp

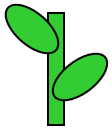
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

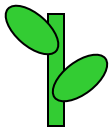
Four phenotypes observed
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall  purple 

3 Tall  white 

3 Short  purple 

1 Short  white 

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

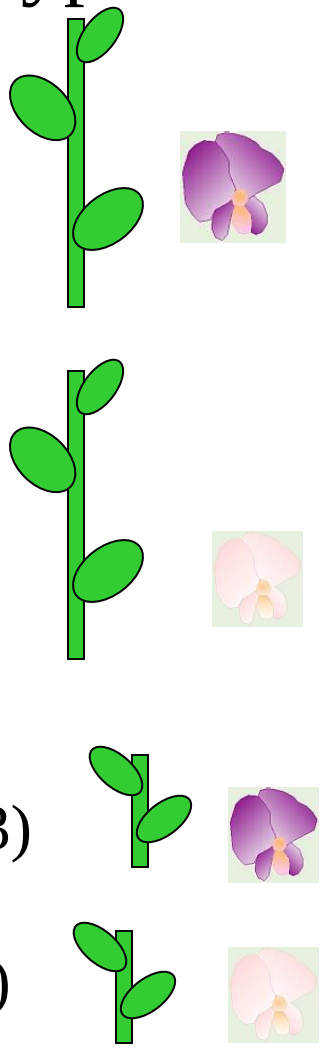
Phenotype Ratio = 9:3:3:1

Dihybrid cross: 9 genotypes

Genotype ratios (9):

Four Phenotypes:

1	TTPP	Tall, purple (9)
2	TTP*p*	
2	T*t*PP	
4	T*t*P*p*	
1	TT*pp*	Tall, white (3)
2	T*tpp*	
1	*tt*PP	Short, purple (3)
2	*tt*P*p*	
1	*ttpp*	Short, white (1)



Principle of Independent Assortment

- Based on these results, Mendel postulated the **3. Principle of Independent Assortment**:

  "Members of one gene pair segregate independently from other gene pairs during gamete formation"

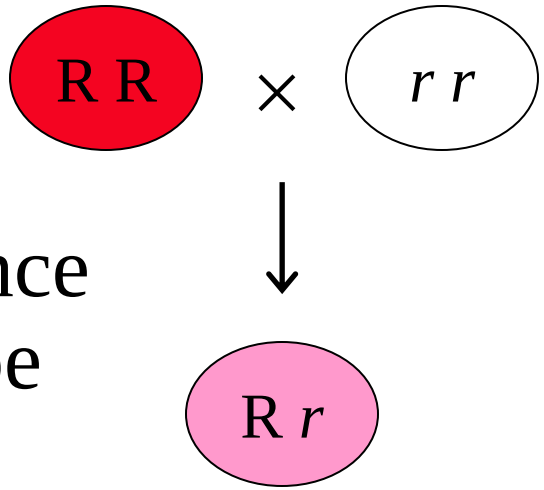
Genes get shuffled – these many combinations are one of the advantages of sexual reproduction

Incomplete Dominance

Snapdragon flowers come in many colors.

If you cross a red snapdragon (RR) with a white snapdragon (rr)

You get PINK flowers (Rr)!



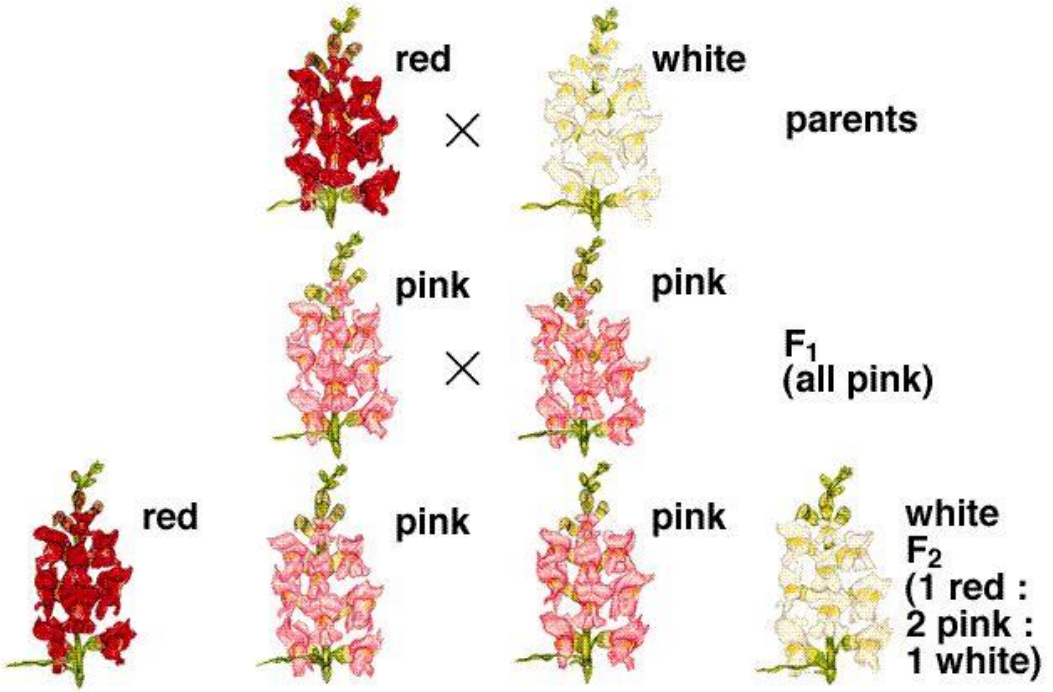
Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete dominance

When F1 generation (all pink flowers) is self pollinated, the F2 generation is 1:2:1
red, pink, white

	R	*r*
R	**R R**	**R *r***
r	**R *r***	***r r***

Incomplete Dominance



MULTIPLE ALLELES

Genes that have more than two alleles

Genes and their alleles

- About 30% of the genes in humans are **di-allelic**
  They exist in two forms, (they have two alleles)
- About 70% are **mono-allelic**, they only exist in one form and they show no variation
- A few are **poly-allelic** having more than two forms.

The ABO blood system

- Controlled by a **tri-allelic gene**
- **6 genotypes**

- The alleles for **antigens** on the surface of the red blood cells
- Two of the alleles are **codominant** to one another and both are **dominant** over the third

- Allele $\mathbf{I^A}$ produces antigen **A**
- Allele $\mathbf{I^B}$ produces antigen **B**
- Allele **i** produces **no** antigen.

The ABO blood system

Genotypes	Phenotypes (Blood types)
$I^A I^A$	A
$I^A I^B$	AB
$I^A i$	A
$I^B I^B$	B
$I^B i$	B
ii	O

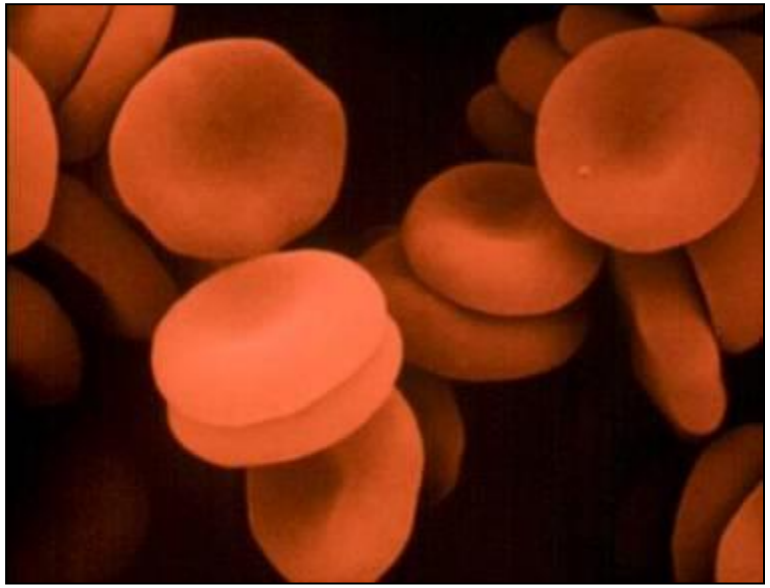
Note:

- Blood types A and B have two possible genotypes – homozygous and heterozygous
- Blood types AB and O only have one genotype each.

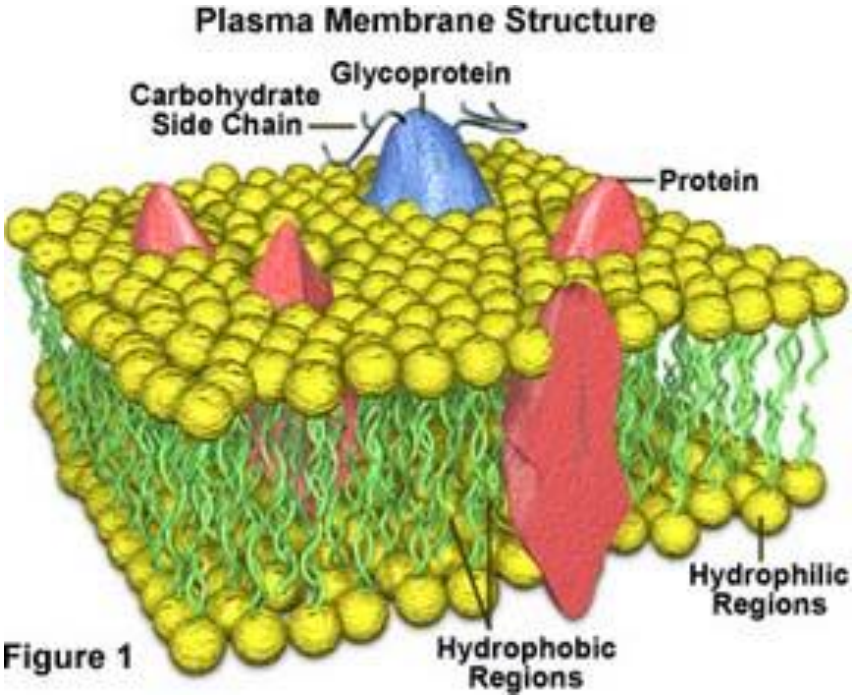
Blood types and transfusions

- Blood types vary and your immune system recognises your own blood type = **self**
- Other blood types = **non-self**
- If a blood, which is incompatible with your body, is transfused it will result in the **agglutination** of the foreign red blood cells.

Antigens

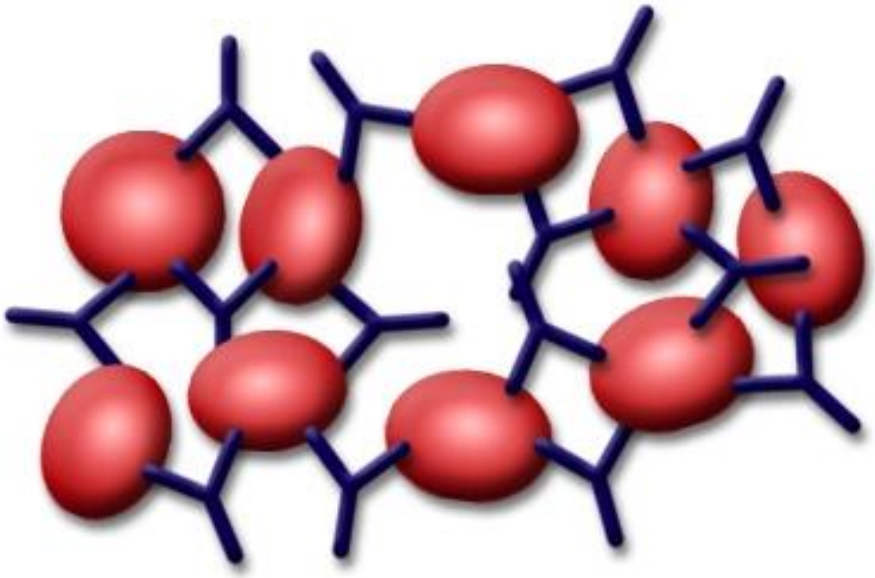


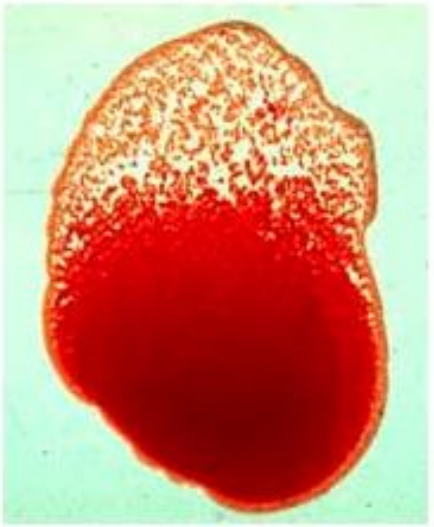
encarta.msn.com/.../Erythrocytes.html



www.okc.cc.ok.us/.../Plasma_Membrane.htm

Agglutination





www.vet-lyon.fr/.../ENV_immuno_1A/immun1-04.htm.

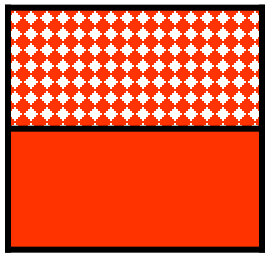
Blood types and transfusions

- Type A people produce antibodies to agglutinate cells which carry Type B antigens
  Recognised as **non-self**
- The opposite is true for people who are Type B
- Neither of these people will agglutinate blood cells which are Type O
  Type O cells do not carry any antigens for the ABO system
  Type O cells pass incognito
- What about type AB people?

Donor-recipient compatibility

Recipient

Donor \ Type	A	B	AB	O
A				
B				
AB				
O				



= Agglutination

= Safe transfusion

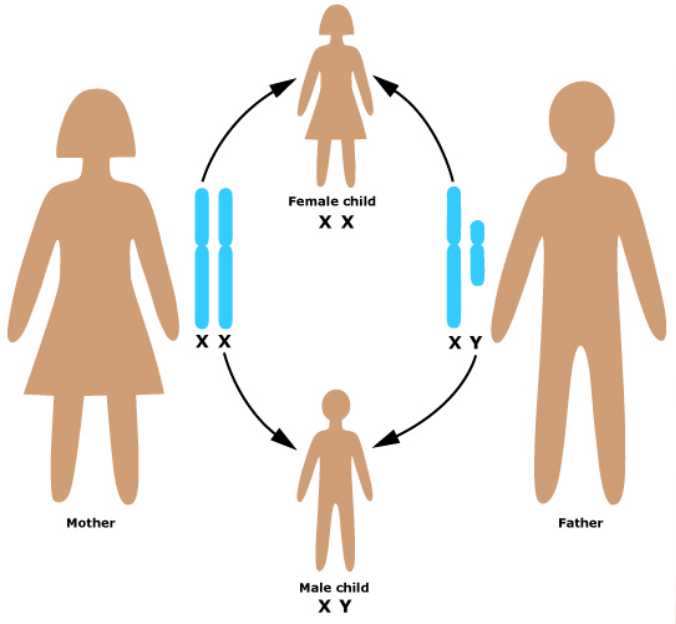
Note:

- **Type O blood** may be transfused into all the other types = the **universal donor**
- **Type AB blood** can receive blood from all the other blood types = the **universal recipient**.

SEX LINKED INHERITANCE

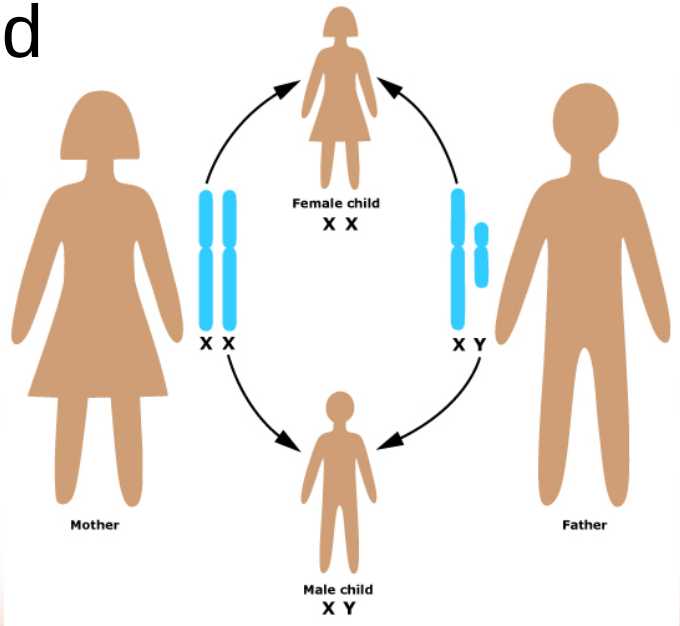
SEX DETERMINATION

- The sex of an individual is determined by the sex chromosomes contributed to the zygote by the sperm and the egg



SEX DETERMINATION

- An egg can donate an X
- A sperm can donate an X or Y
- Therefore the sperm determines the sex of a child

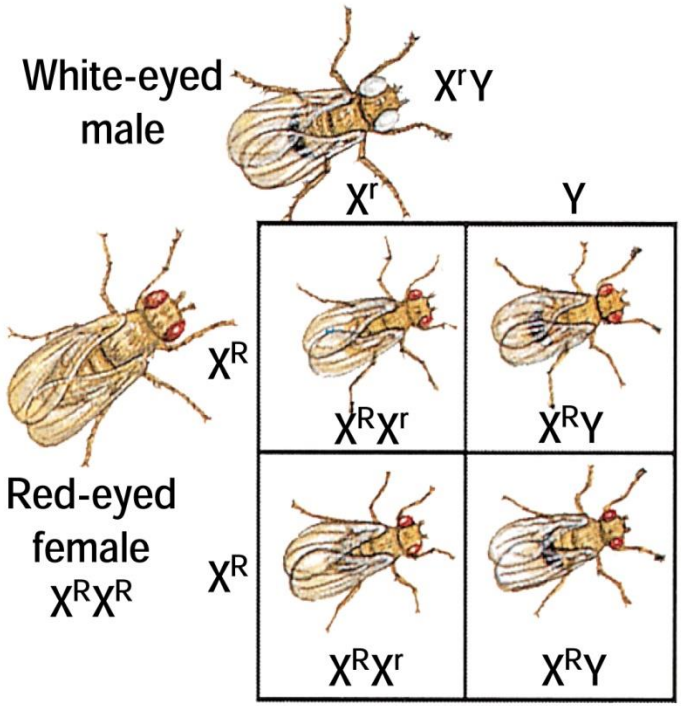


SEX-LINKED INHERITANCE

- Using fruit flies as test subjects, Thomas Morgan studied eye colour using simple monohybrid crosses.
- Red eyes (R) are dominant over white eyes (r).

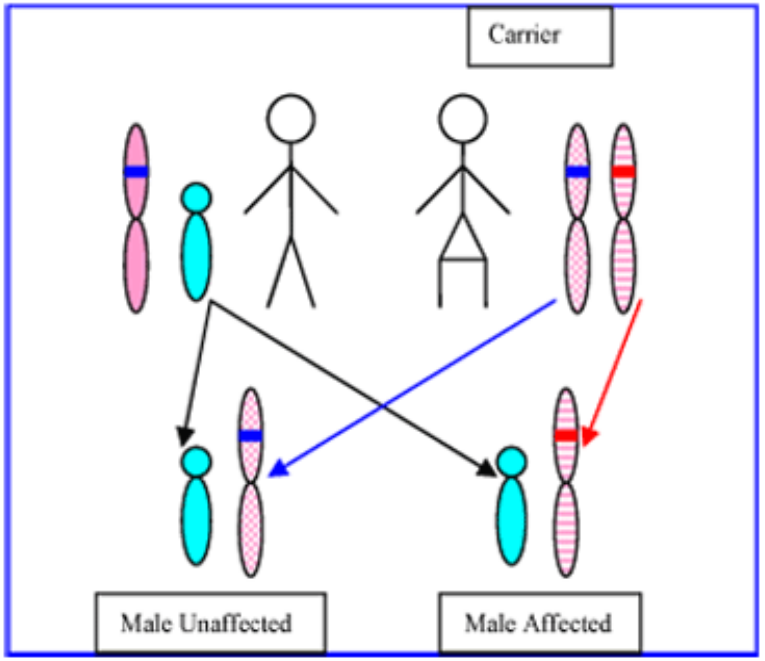
SEX-LINKED INHERITANCE

- When he crossed purebred white-eyed males with red-eyed females, he was unable to produce a female with white eyes.
- He concluded that the gene must be located on the X chromosome.



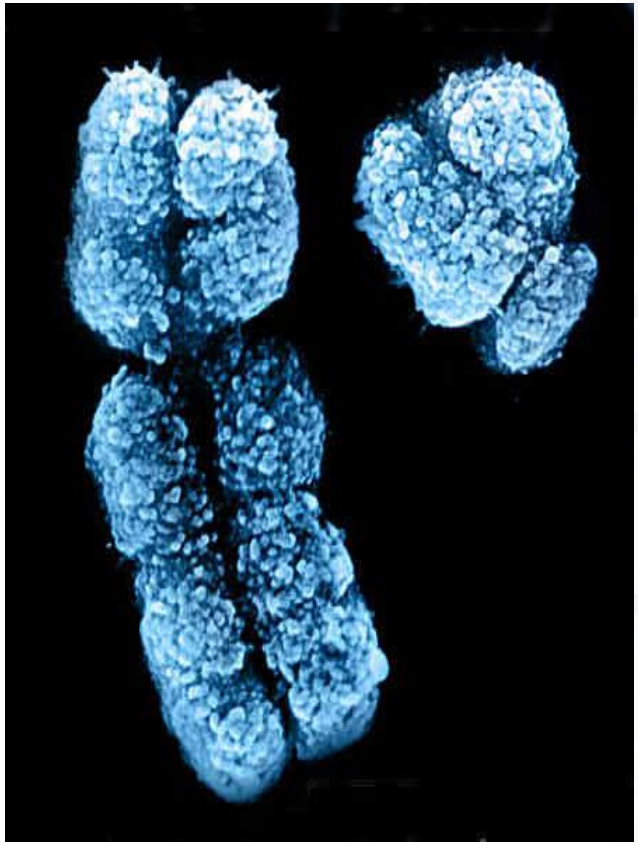
SEX-LINKED INHERITANCE

- Some traits are located on the sex chromosomes, so the inheritance of these traits depends on the sex of the parent carrying the trait.



SEX-LINKED INHERITANCE

- Most known sex-linked traits are X-linked (carried on the X chromosome). This is probably because the X chromosome is much larger than the Y chromosome.

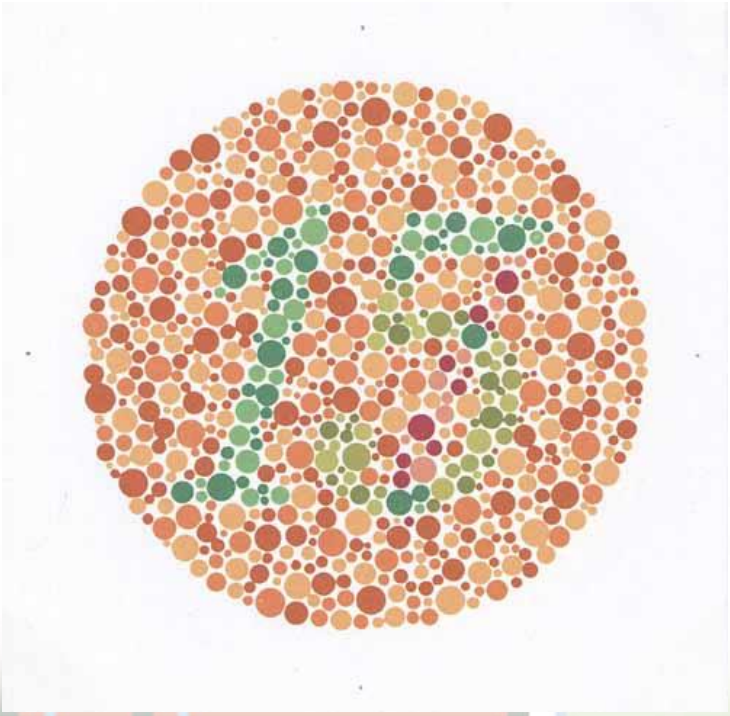


SEX-LINKED DISORDERS

- Some sex-linked traits are associated with disorders.
- Most are found on the X chromosome, Y-linked disorders are rare.
- Males are at a much greater risk for inheriting sex-disorders because they only inherit one X, so if the X has the allele for the disorder, they will suffer from the disorder.
- Recessive **lethal** X-linked traits result in death.

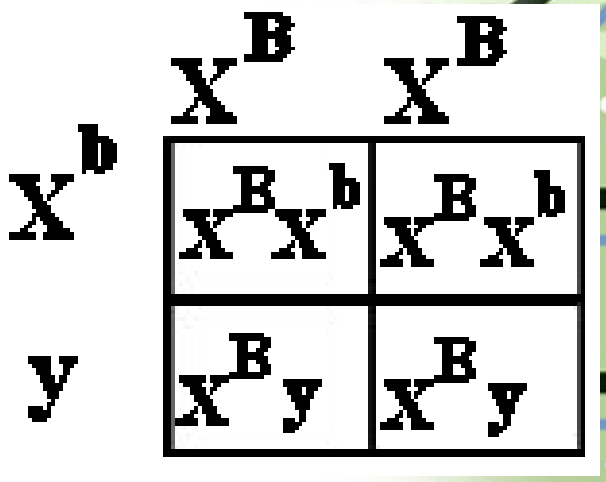
EXAMPLES OF SEX-LINKED TRAITS and DISORDERS

- Male pattern baldness, red-green colour blindness, myopia, night blindness, hemophilia

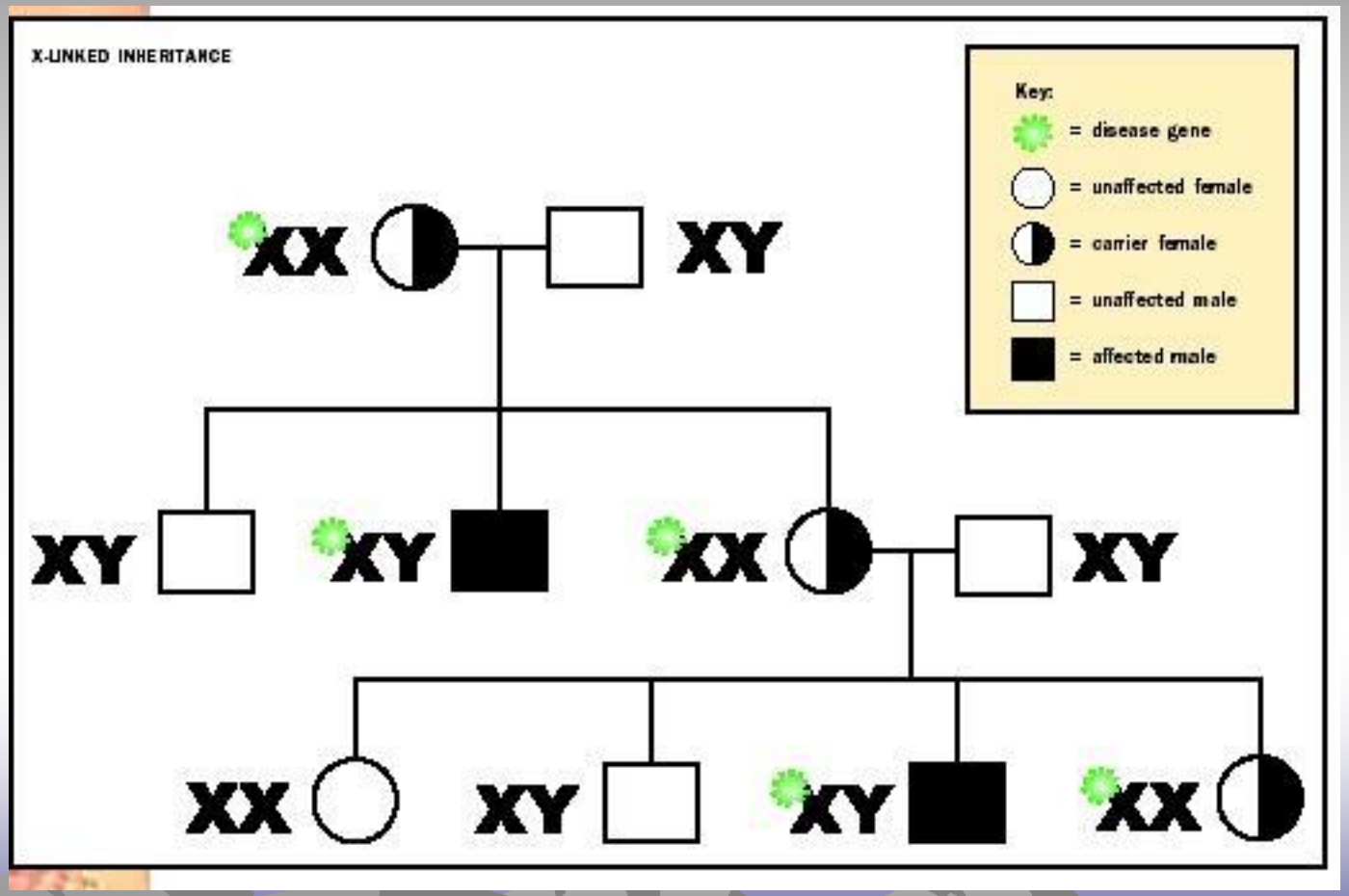


SEX-LINKED INHERITANCE

- Punnett squares are used to predict the outcome of sex-linked inheritance.
- Assume the trait is X-linked unless told otherwise!
- Most disorders are recessive, some are dominant, the question will tell you.
- A “carrier” is a female who is heterozygous for the trait.

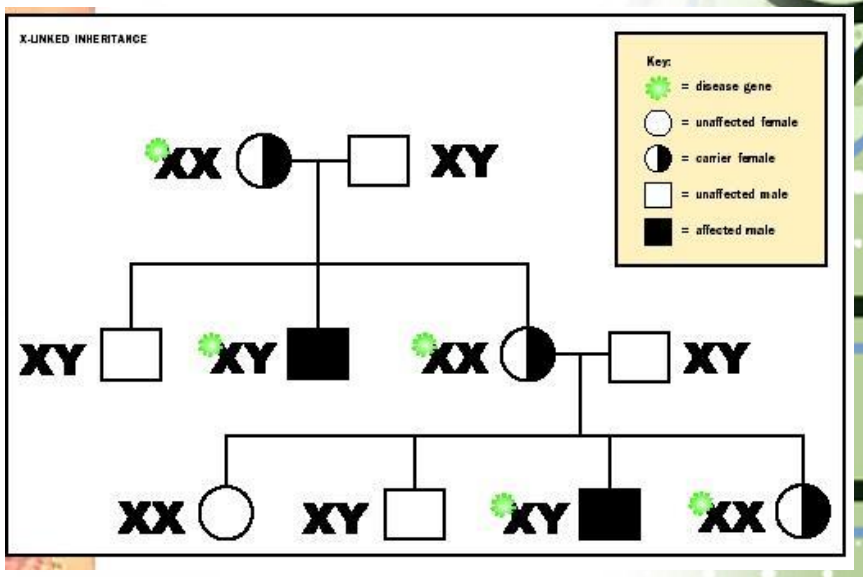


Patterns of Inheritance



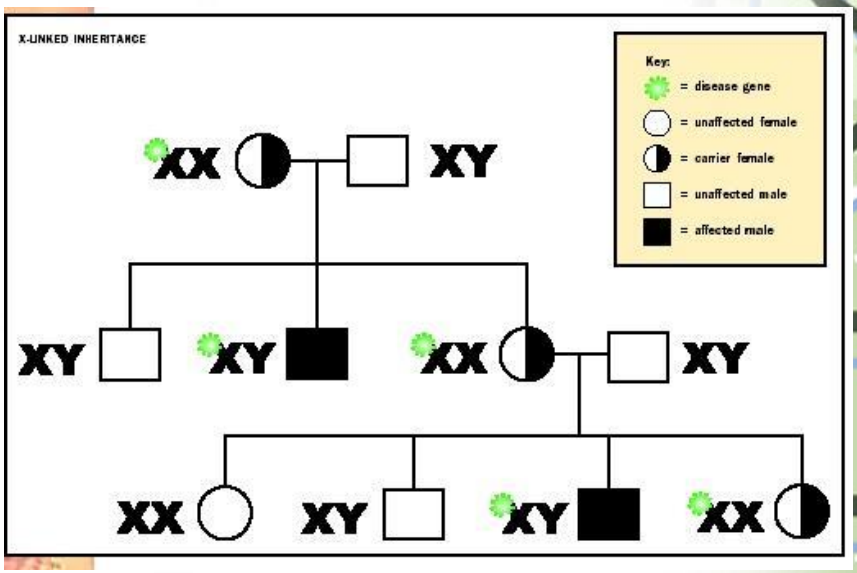
Pedigrees

- A pedigree is a genetic family tree that shows how prevalent a trait is in a family unit from generation to generation.
- They are often used to track the expression of genetic conditions and disorders.



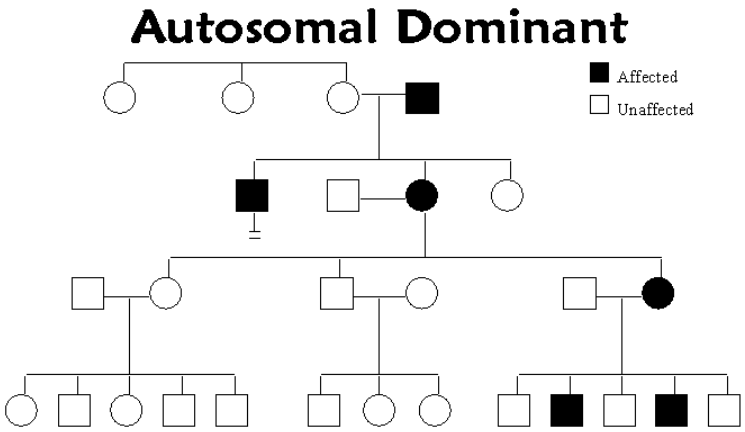
Pedigrees

- Squares represent males and circles females.
- A coloured in shape means that person has the trait in question.
- A half coloured in shape means that they are carrying an allele for a recessive trait.



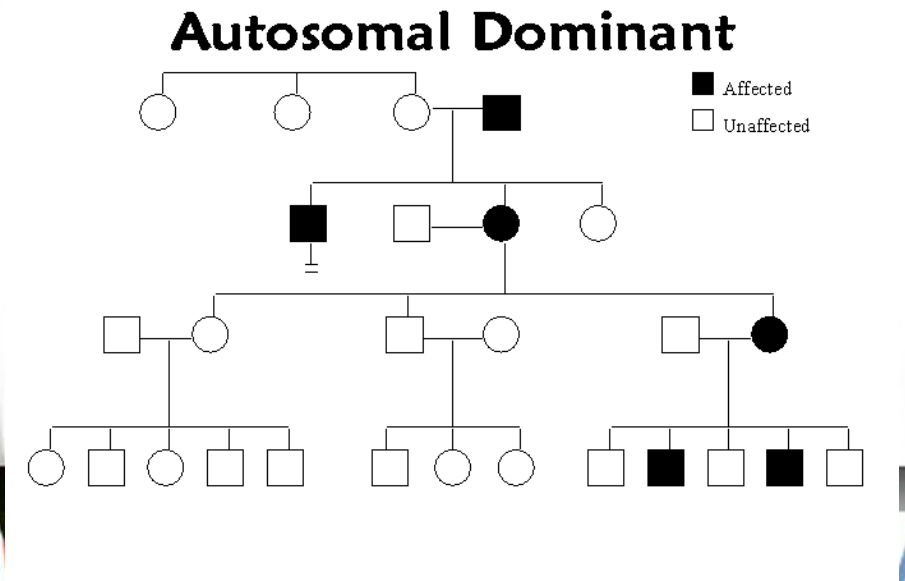
Autosomal Dominant Inheritance

- Autosomal means not on the sex chromosomes.
- Refers to those situations in which a single copy of an allele is sufficient to cause expression of a trait.



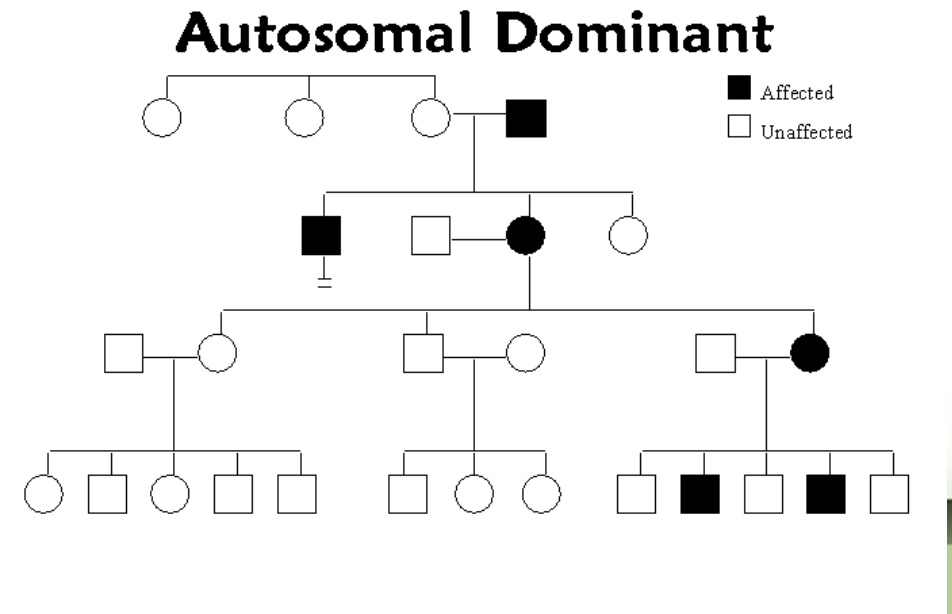
Autosomal Dominant Inheritance

- 1. Every affected person should have at least one affected parent.
- 2. Males and females should be equally often affected.
- 3. An affected person has at least a 50% chance of transmitting the dominant allele to each offspring



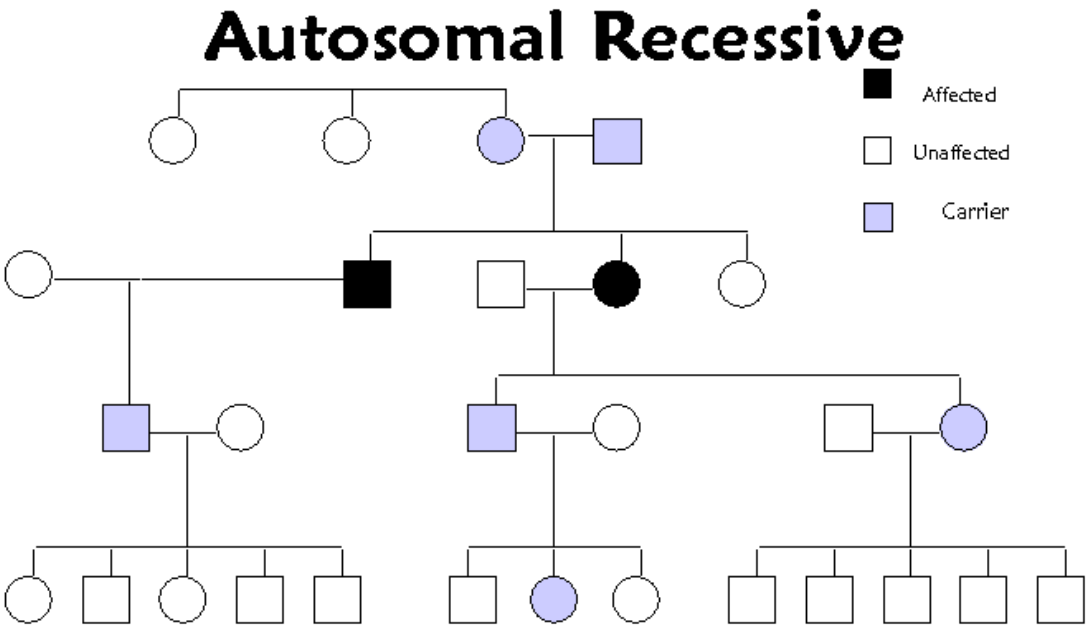
Autosomal Dominant Inheritance Examples

- Progeria (caused by a mutation) in which the person ages very rapidly. They die before they can reproduce.
- Huntington's Disease in which the central nervous system starts to break down around the age of 30.



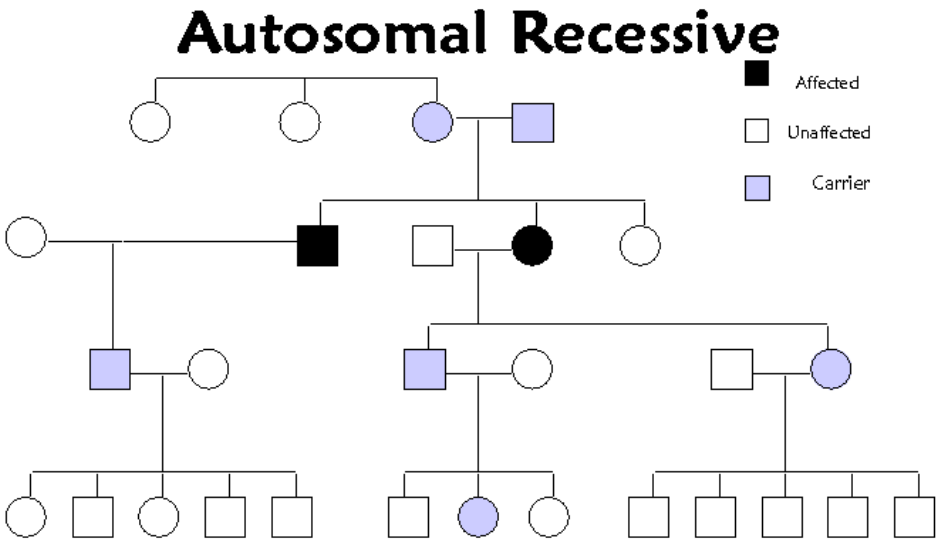
Autosomal Recessive Inheritance

- Refers to those situations where two recessive alleles result in a trait being expressed



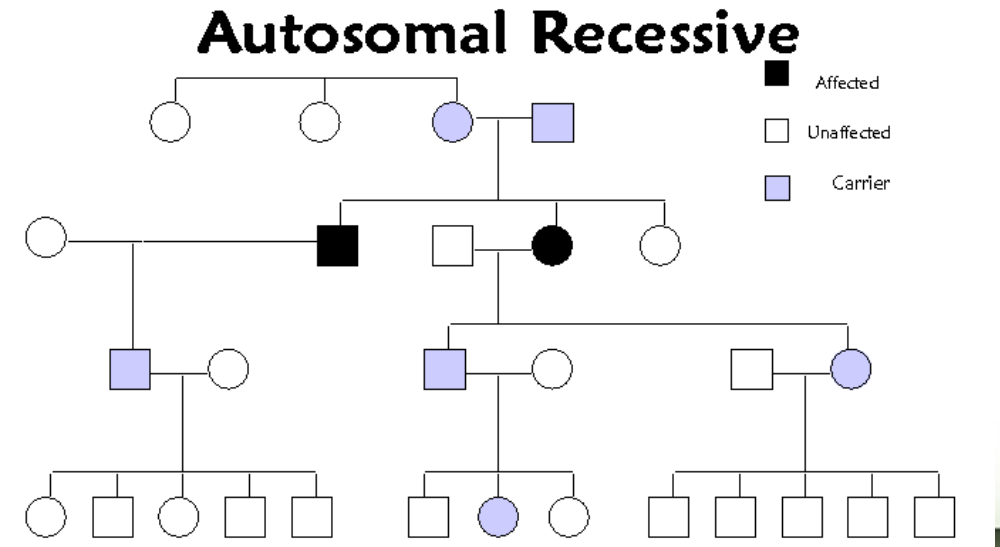
Autosomal Recessive Inheritance

- 1. An affected person may not have affected parents. Parents would be carriers.
- 2. Affects both sexes equally. Can appear to skip generations.
- 3. Two affected parents will have affected children 100% of the time.



Autosomal Recessive Examples

- Albinism is a genetic condition which is the loss of pigment in hair, skin and eyes.
- Tay Sachs is a genetic disorder which is a build up of fatty deposits in the brain, eventually proving to be fatal

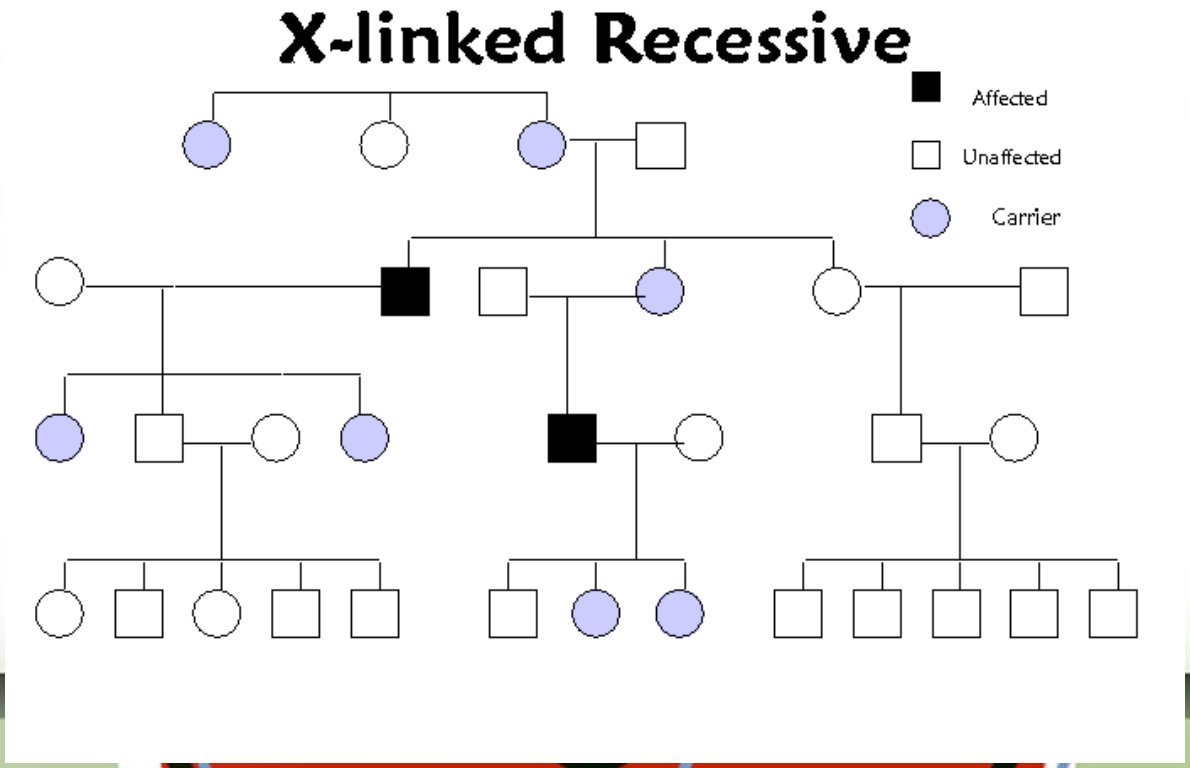


Codominant Inheritance

- Sickle cell Anemia is a codominant condition/disorder in which there is a defect in hemoglobin, an important protein in red blood cells.
- An individual homozygous for sickle cells suffers from blood clots to important organs, anemia and usually dies prematurely.
- An individual heterozygous for normal and sickle cells does not suffer the full disorder, but some red blood cells still have defective hemoglobin.
- In certain areas of the world this is an advantage. Malaria is caused by a protist that prefers normal blood cells. If some of your blood cells are damaged, you are less likely to become a host! (Heterozygous Advantage)

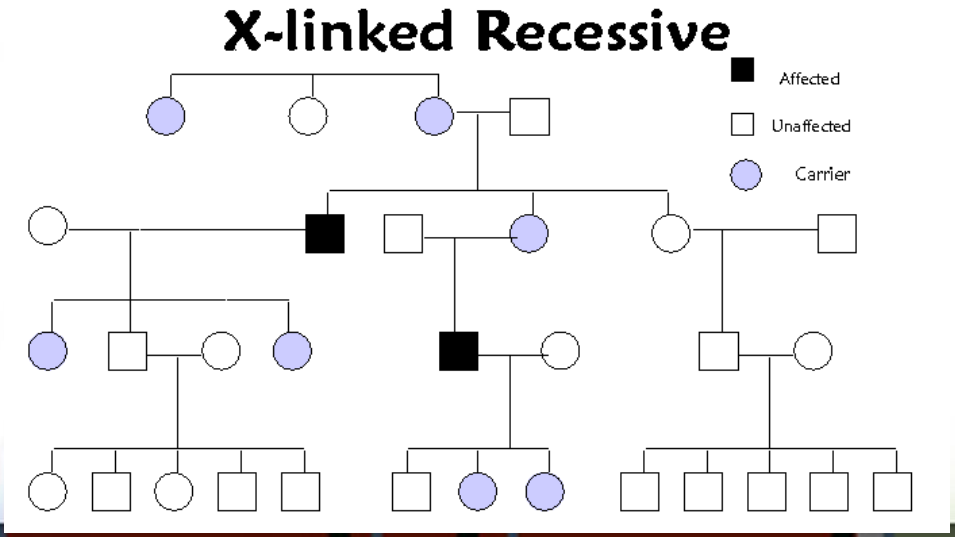
X – linked Recessive Inheritance

- Refers to those situations where a recessive allele on the X chromosome can lead to a trait/condition or disorder



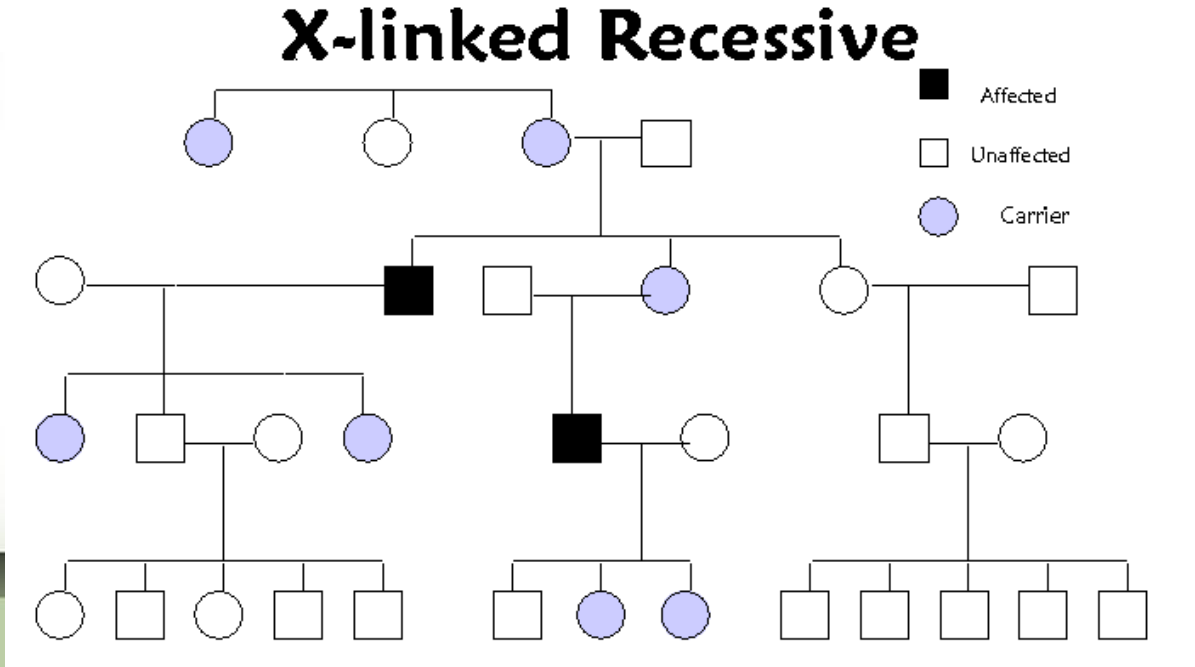
X – linked Recessive Inheritance

- Males are affected more often than females. Ratio of 8:1.
- Affected males will transmit the allele to all daughters, but not to sons.
- Homozygous recessive females can arise only from matings in which the father is affected and the mother is affected or a carrier.



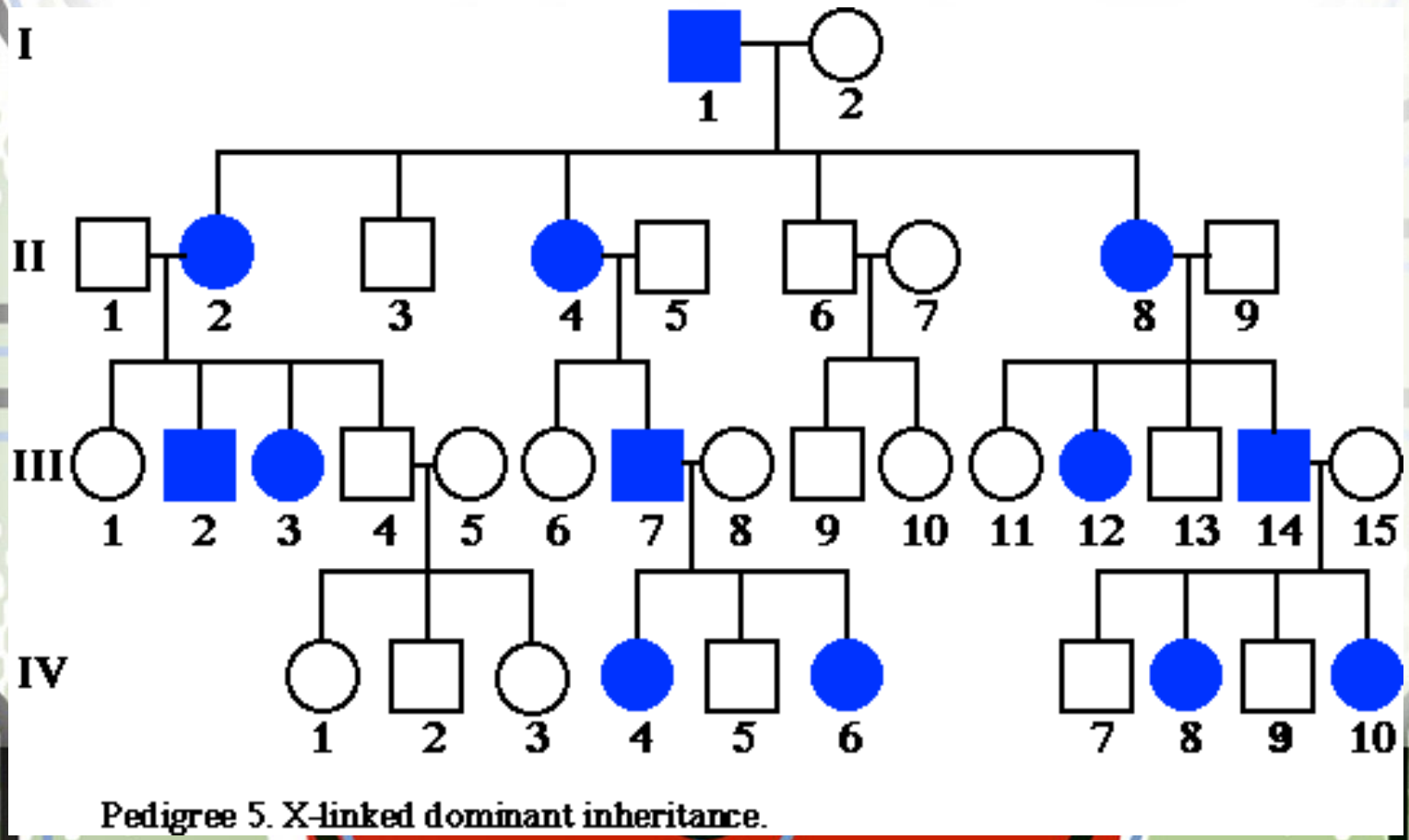
X – linked Recessive Disorders

- Hemophilia which is the inability of the blood to clot properly.
- Duchenne Muscular Dystrophy which causes progressive and degenerative muscle weakness.



X – Linked Dominant Inheritance

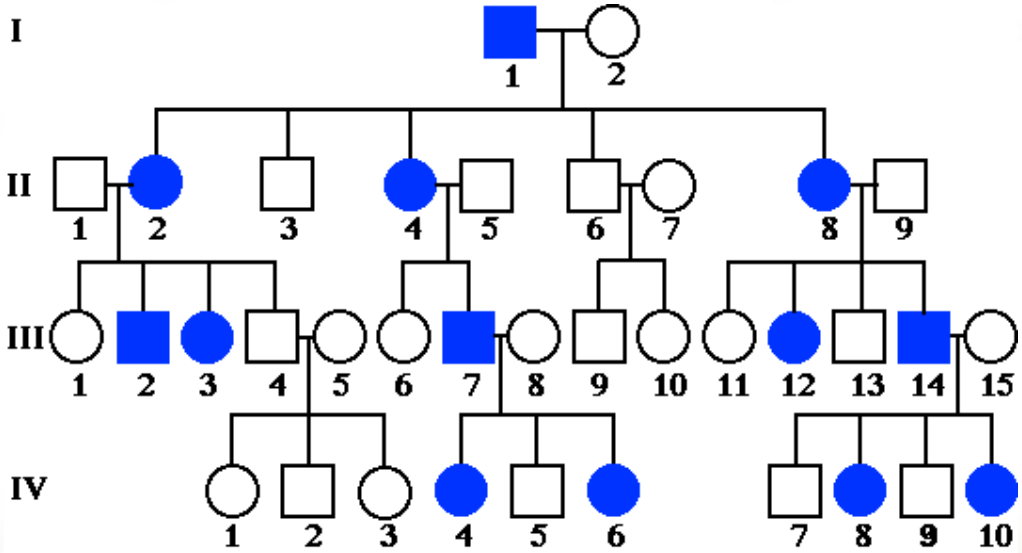
- Refers to situations where a single dominant allele on the X chromosome can lead to a trait/condition.



Pedigree 5. X-linked dominant inheritance.

X – Linked Dominant Inheritance

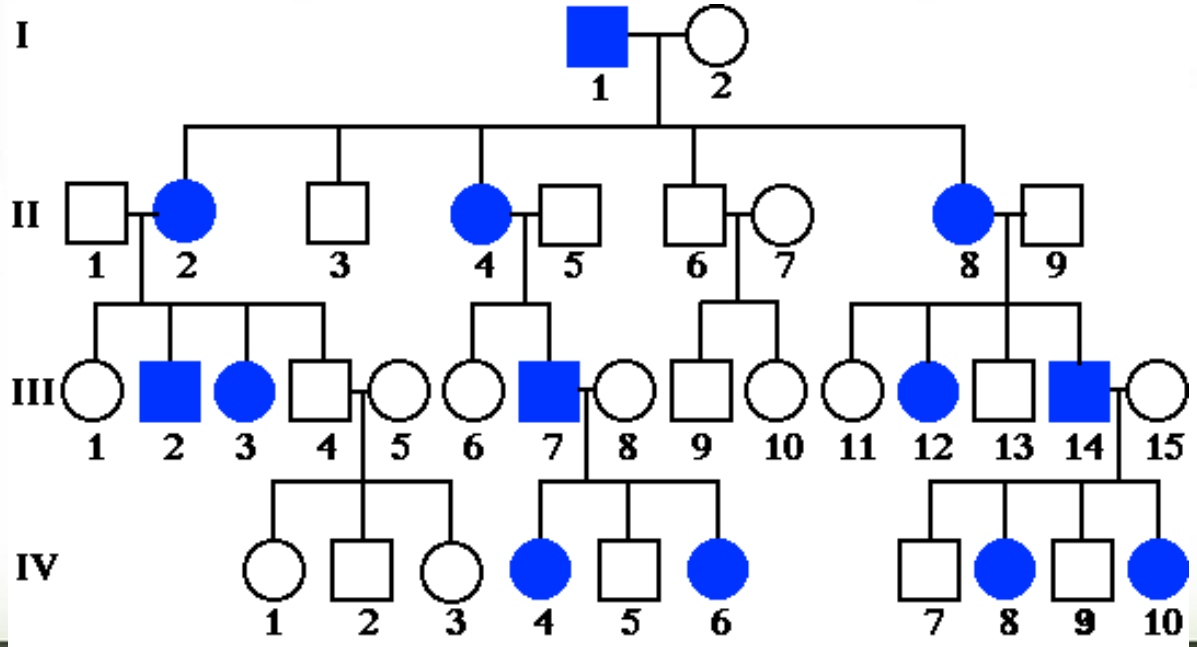
- 1. Twice as many females are affected as males.
- 2. Usually half the children of an affected female will be affected, regardless of sex.
- 3. All the daughters of an affected male will be affected but none of the sons.



Pedigree 5. X-linked dominant inheritance.

X – Linked Dominant Example

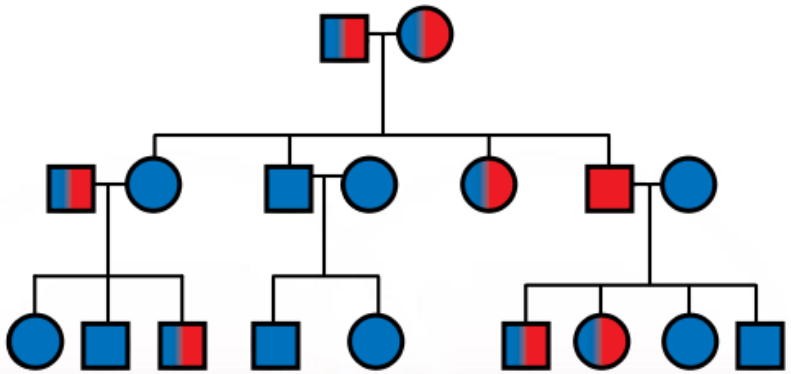
- Vitamin D resistant rickets which can lead to bone deformities, particularly in the lower limbs (bowed legs).



Pedigree 5. X-linked dominant inheritance.

PEDIGREES

- Chart showing genetic relationships between members of a family
- Squares represent males, circles females
- Colour shows infected person, ½ shaded shows carrier



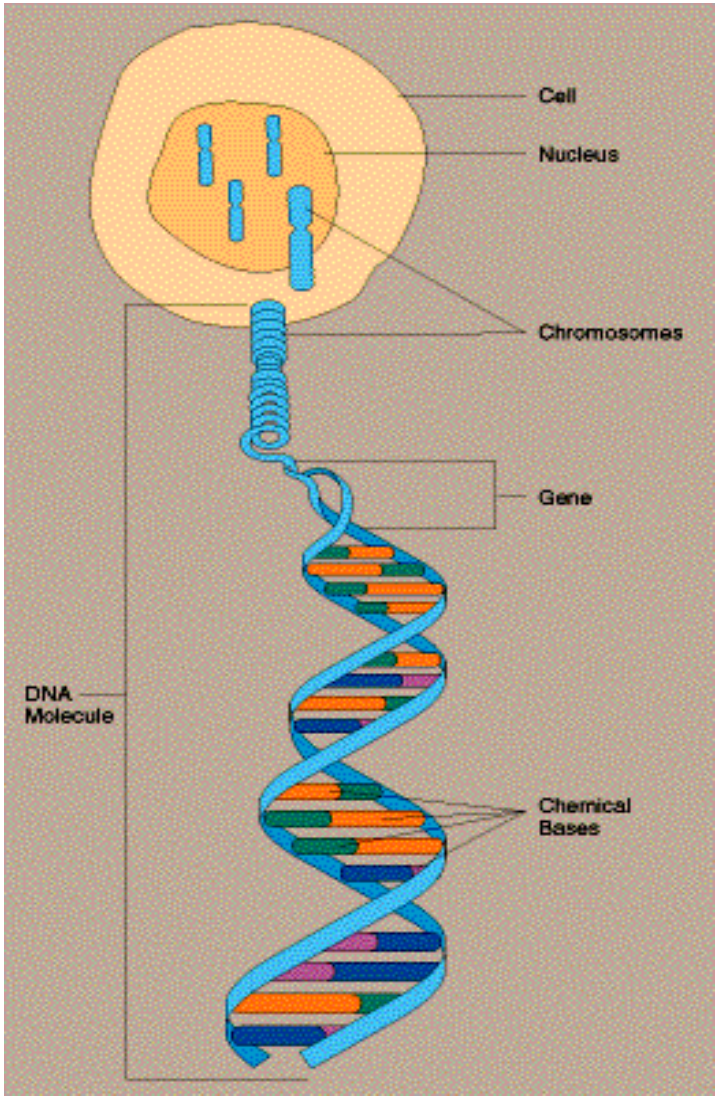
Chromosomal Aberrations

Basic definitions

- Chromosomes, DNA and genes
- Chromatides, and centromere
- Arms of a chromosome (p and q)
- Karyotype
- Autosomes and sex chromosomes
- Genotype and phenotype

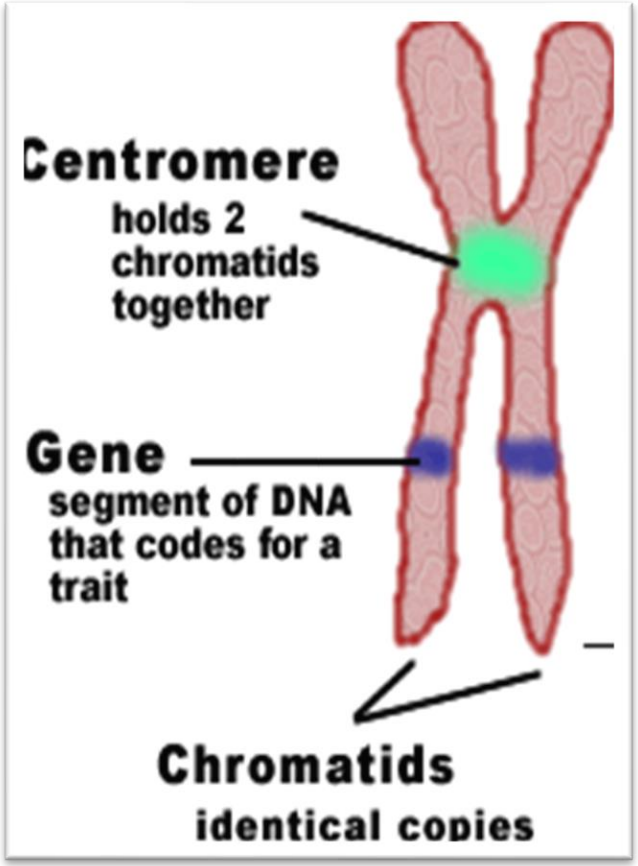
Human Chromosomes

- The **chromosome** carries the genetic information.
- composed of **deoxyribonucieic acid (DNA)** on framework of protein .
- Segments of DNA molecules comprise the genes; the units of heredity.

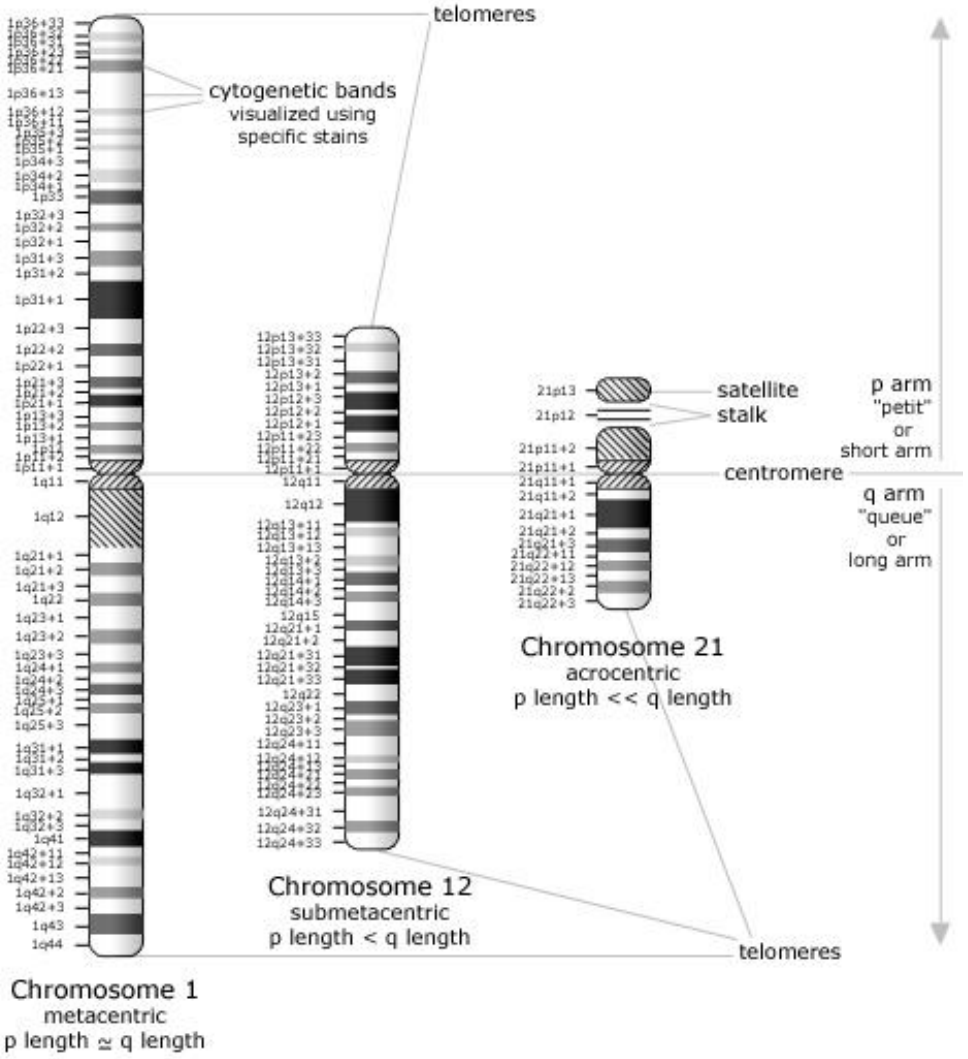


Chromosomes

- During cell division, the chromosome can be seen to consist of 2 parallel strands; the **chromatids**, held together at one point, the **centromere.**



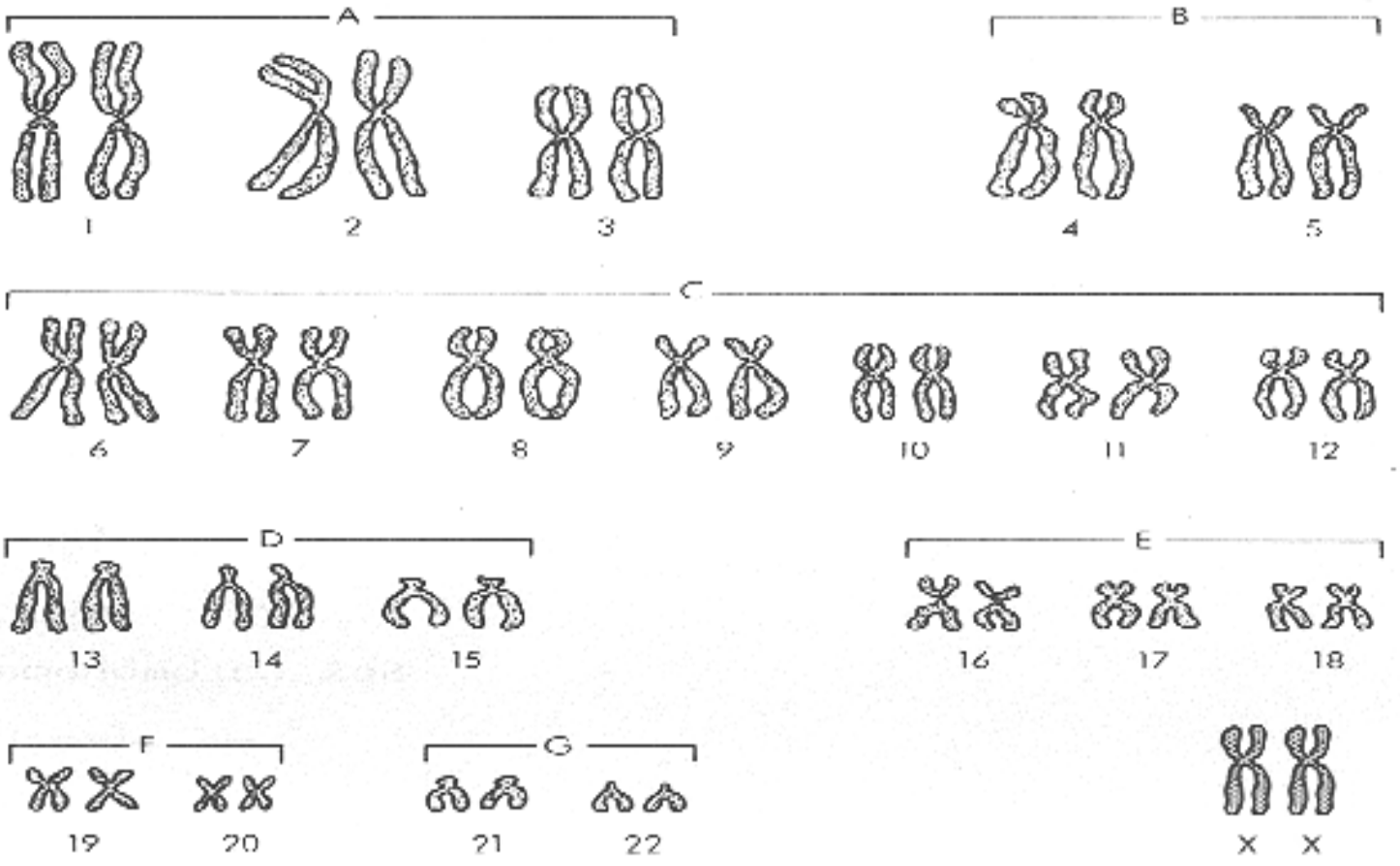
Human Chromosomes



Karyotype

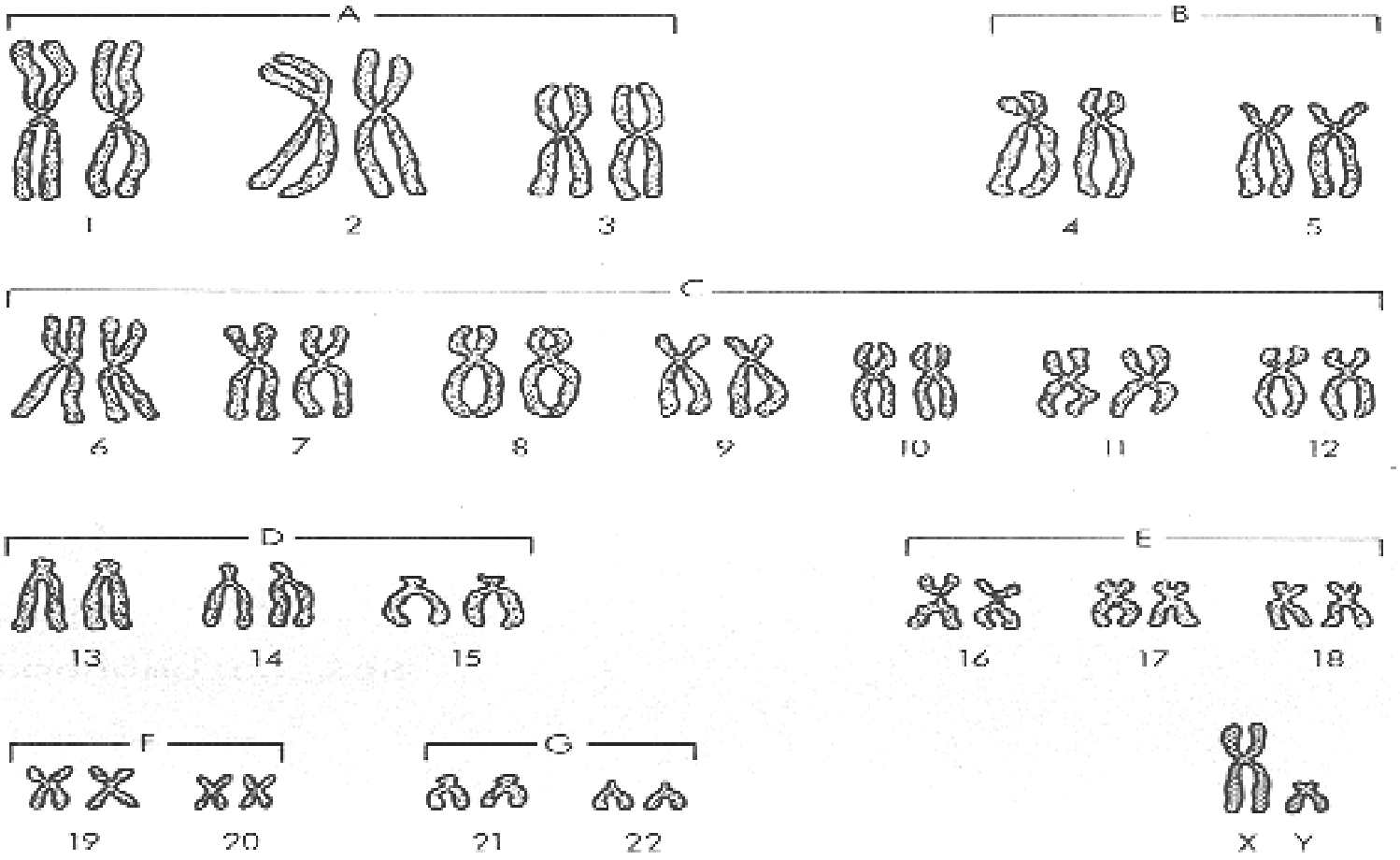
- It is the set of chromosomes of an individual.
- It is the systematized arrangement of the chromosomes of a single cell.
- In the human cell, there are 46 chromosomes or 23 pairs (diploid number); of these 23 pairs, 22 are similar in both sexes and are called the ***autosomes.*** The remaining pair is called ***sex chromosomes*** : XX in the female cells and XY in the male cells .
- Chromosomes are arranged in groups A to G according to their shape & size.

Karyotype of a normal female



Normal Female

Karyotype of a normal male



Normal Male

Chromosomal Abnormalities

Chromosomal Abnormalities

- Chromosomal abnormalities are either numerical or structural.
- They are a very common cause of early spontaneous miscarriage.
- Usually, but not always, cause multiple congenital anomalies and learning difficulties.

Chromosomal Aberrations (abnormalities)

- **Structural Aberrations**
  - Deletion
  - Duplication
  - Inversion
  - Translocation
- **Numerical Aberrations (abnormalities)**
  - Polyploidy: Multiple of the haploid (> Diploid)
  - Aneuploidy: Abnormal number

Structural abnormalities

1) **Deletion** : loss of a portion of a chromosome
2) **Duplication** : extra piece of a chromosome. .
3) **Inversion** : fragmentation of a chromosome followed by reconstitution with *a* section inverted.
4) **Translocation** :

the transfer of a chromosome or a segment of it to a non-homologous chromosome

Mutations

What Are Mutations?

- Changes in the nucleotide sequence of DNA
- May occur in somatic cells (aren't passed to offspring)
- May occur in gametes (eggs & sperm) and be passed to offspring

Are Mutations Helpful or Harmful?

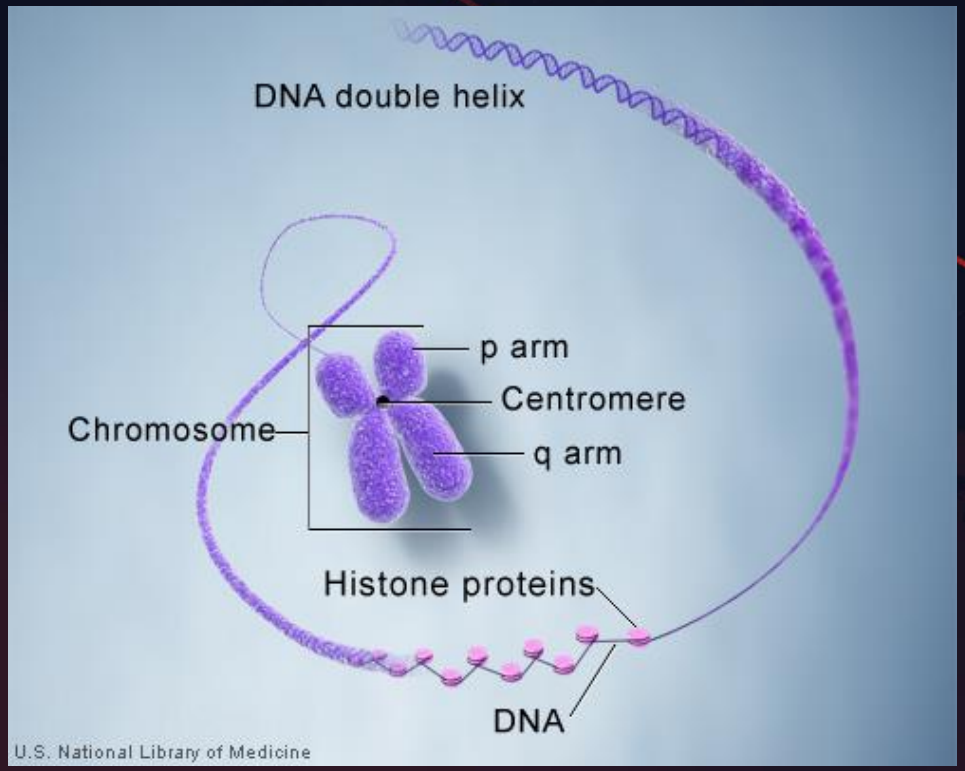
- Mutations happen regularly
- Almost all mutations are neutral
- Chemicals & UV radiation cause mutations
- Many mutations are repaired by enzymes

Are Mutations Helpful or Harmful?

- Some type of skin cancers and leukemia result from somatic mutations
- Some mutations may improve an organism's survival (beneficial)

Quick Review: What is a chromosome?

A chromosome is a DNA molecule that is tightly coiled around proteins called histones, which support its structure, to form a thread-like structures.



Types of Mutations

Chromosome Mutations

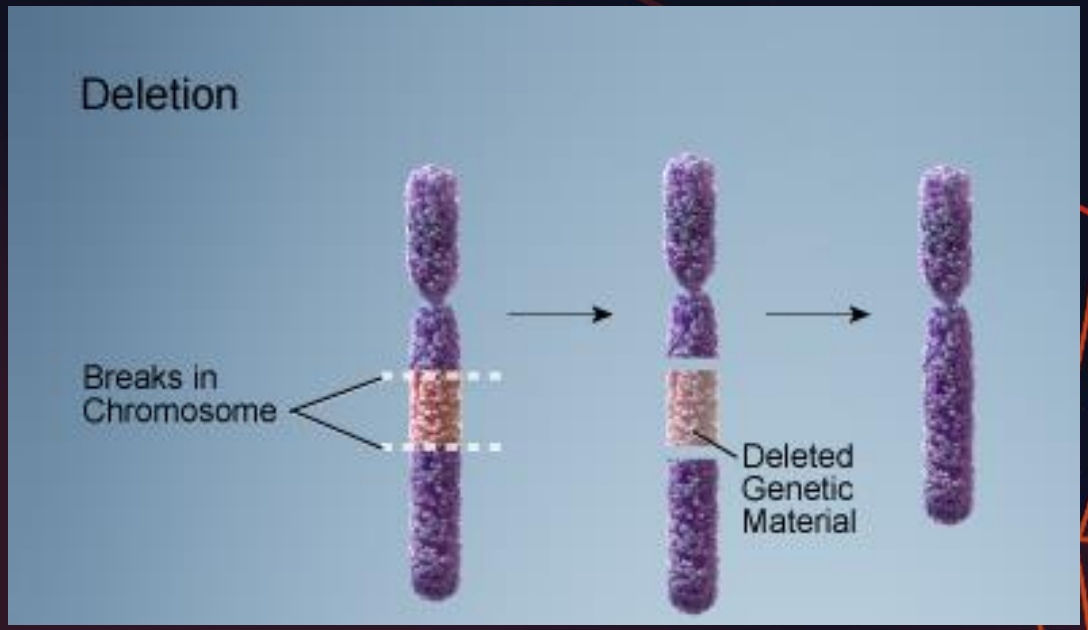
- May Involve:
  - **Changing the structure** of a chromosome
  - The **loss or gain** of part of a chromosome

Chromosome Mutations

- Five types exist:
  - Deletion
  - Inversion
  - Translocation
  - Nondisjunction
  - Duplication

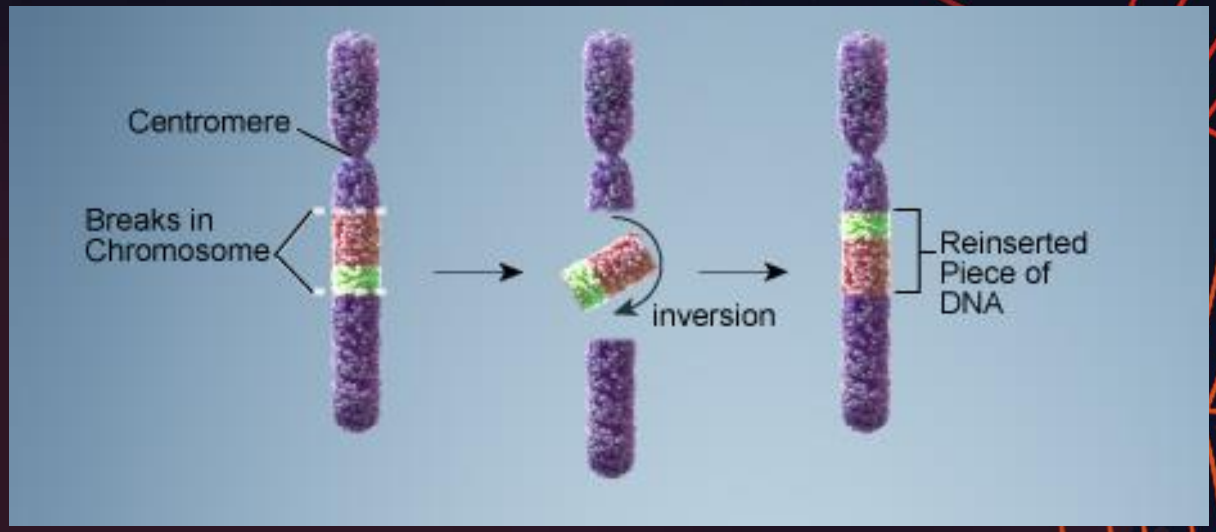
Deletion

- Due to **breakage**
- A **piece** of a chromosome is **lost**



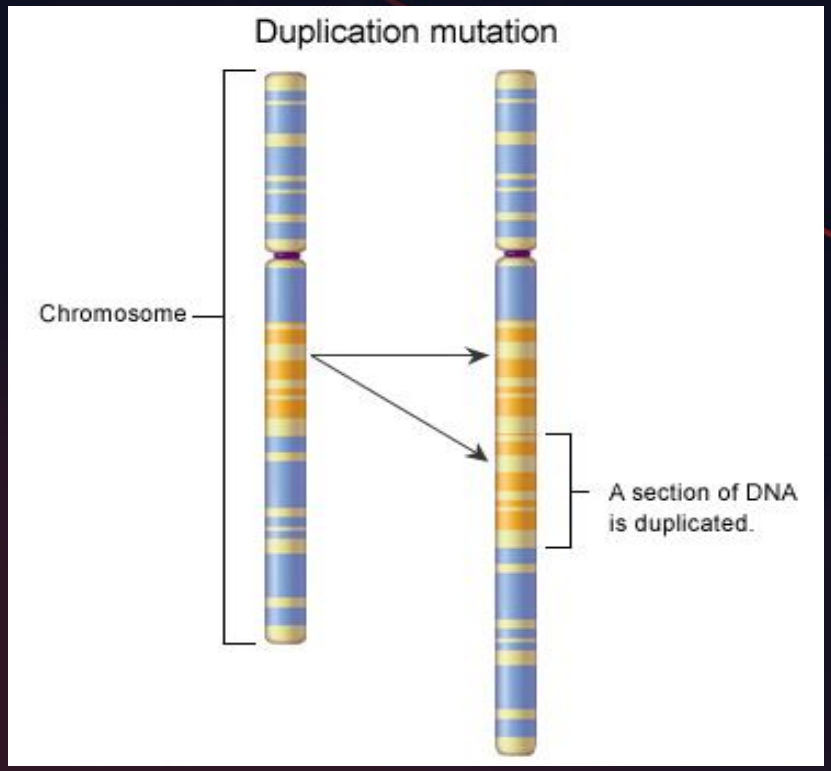
Inversion

- Chromosome segment **breaks off**
- Segment flips around **backwards**
- Segment **reattaches**



Duplication

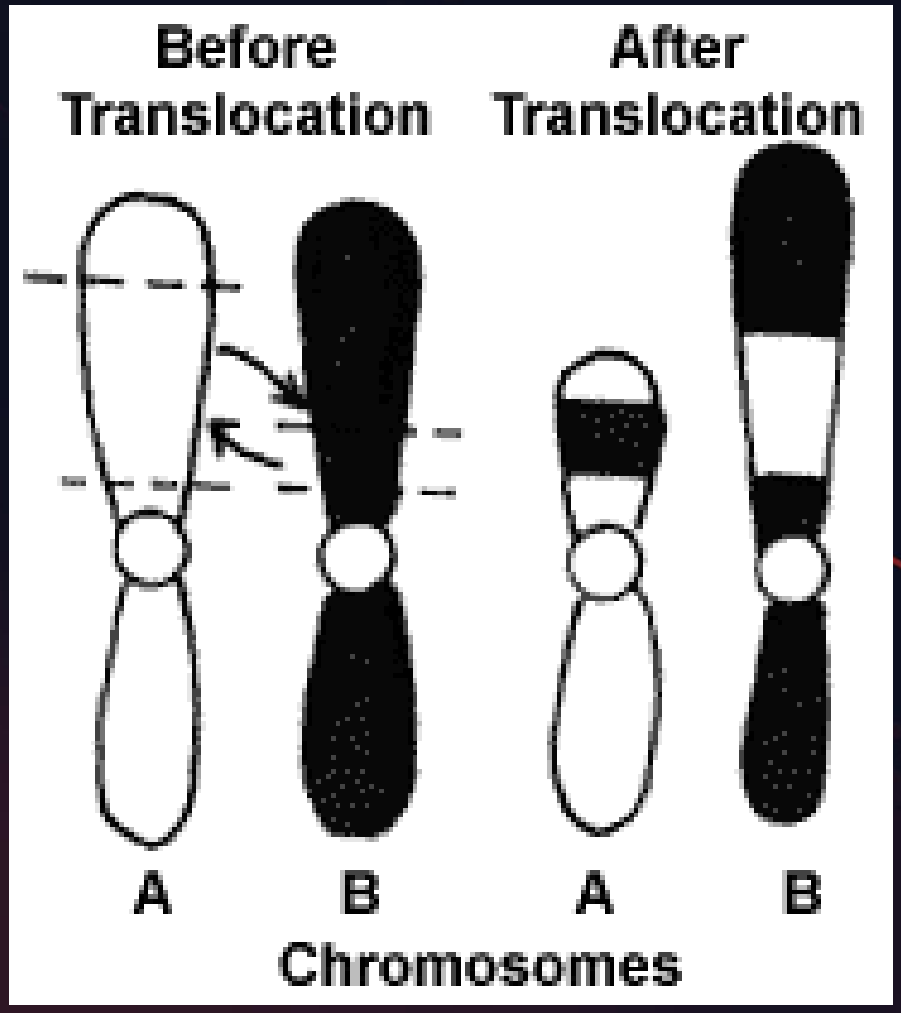
- Occurs when a gene **sequence is repeated**



Translocation

- Involves **two chromosomes** that are **NOT** homologous
- **Part** of one chromosome is **transferred to another** chromosome

Translocation



Nondisjunction

- **Failure** of chromosomes **to separate** during meiosis
- Causes gamete to have **too many** or **too few** chromosomes
- **Disorders:**
  - **Down Syndrome** – three 21st chromosomes
  - **Turner Syndrome** – single X chromosome
  - **Klinefelter's Syndrome** – XXY chromosomes

Down Syndrome

Down syndrome (DS or DNS), also known as **trisomy 21**, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is typically associated with physical growth delays, characteristic facial features and mild to moderate intellectual disability.

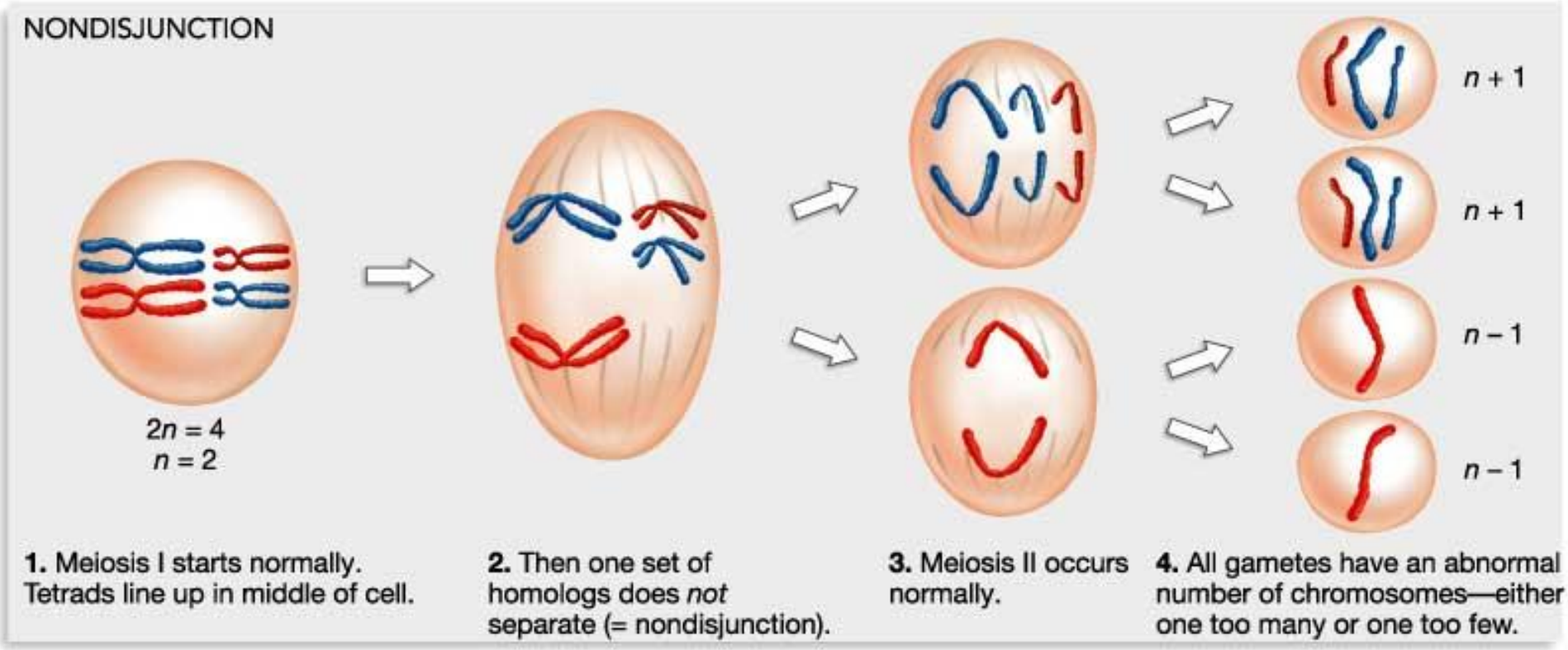
Turner Syndrome

A condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.

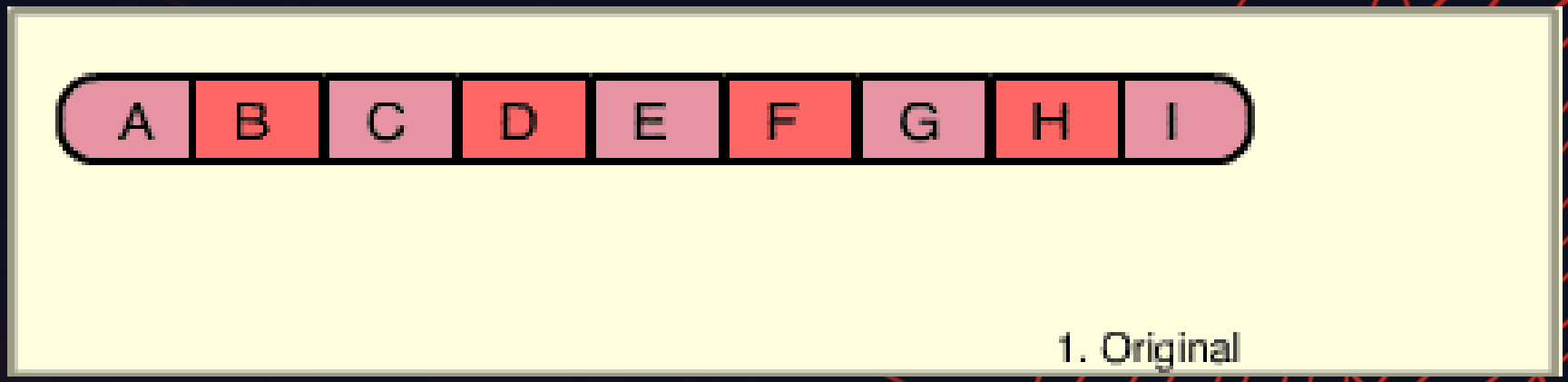
Klinefelter's Syndrome

A genetic disorder that affects males.

Klinefelter's syndrome occurs when a boy is born with one or more extra X chromosomes. Most males have one Y and one X chromosome. Having extra X chromosomes can cause a male to have some physical traits unusual for males such as weaker muscles, greater height, poor coordination, less body hair, and sterility


NONDISJUNCTION
2n = 4
n = 2
n + 1
n + 1
n – 1
n – 1
1. Meiosis I starts normally. Tetrads line up in middle of cell.
2. Then one set of homologs does not separate (= nondisjunction).
3. Meiosis II occurs normally.
4. All gametes have an abnormal number of chromosomes—either one too many or one too few.

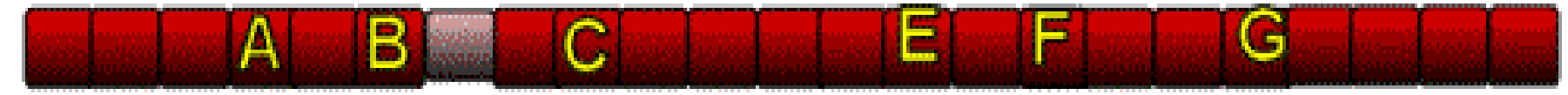
Chromosome Mutation Animation



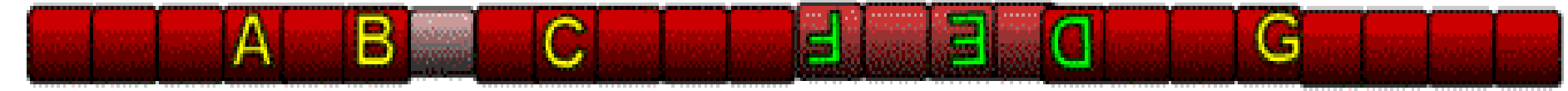
Original Chromosome

Duplication

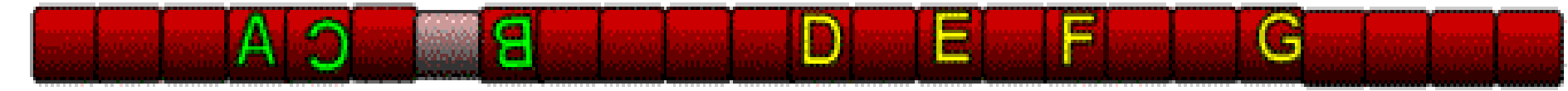
Deletion



Inversion



Inversion



Gene Mutations

- Change in the **nucleotide sequence** of a gene
- May only involve a single nucleotide
- May be due to **copying errors**, **chemicals**, **viruses**, etc.

Types of Gene Mutations

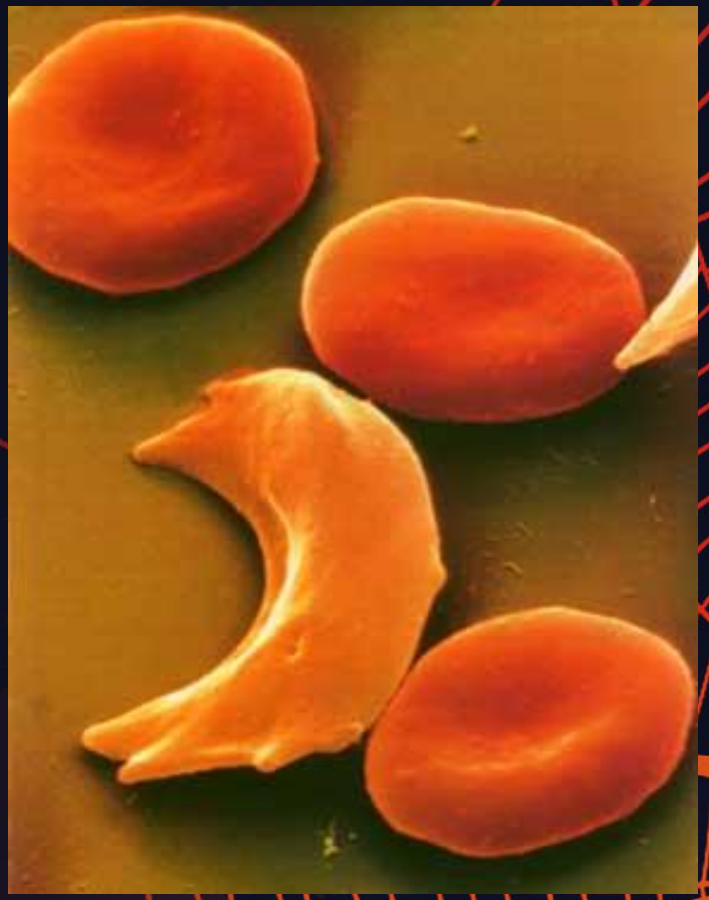
- Include:
  - Point Mutations
  - Substitutions
  - Insertions
  - Deletions
  - Frameshift

Point Mutation

- Change of a **single** nucleotide
- Includes the deletion, insertion, or substitution of **ONE** nucleotide in a gene

Point Mutation

- **Sickle Cell disease** is the result of one nucleotide substitution
- Occurs in the **hemoglobin gene**



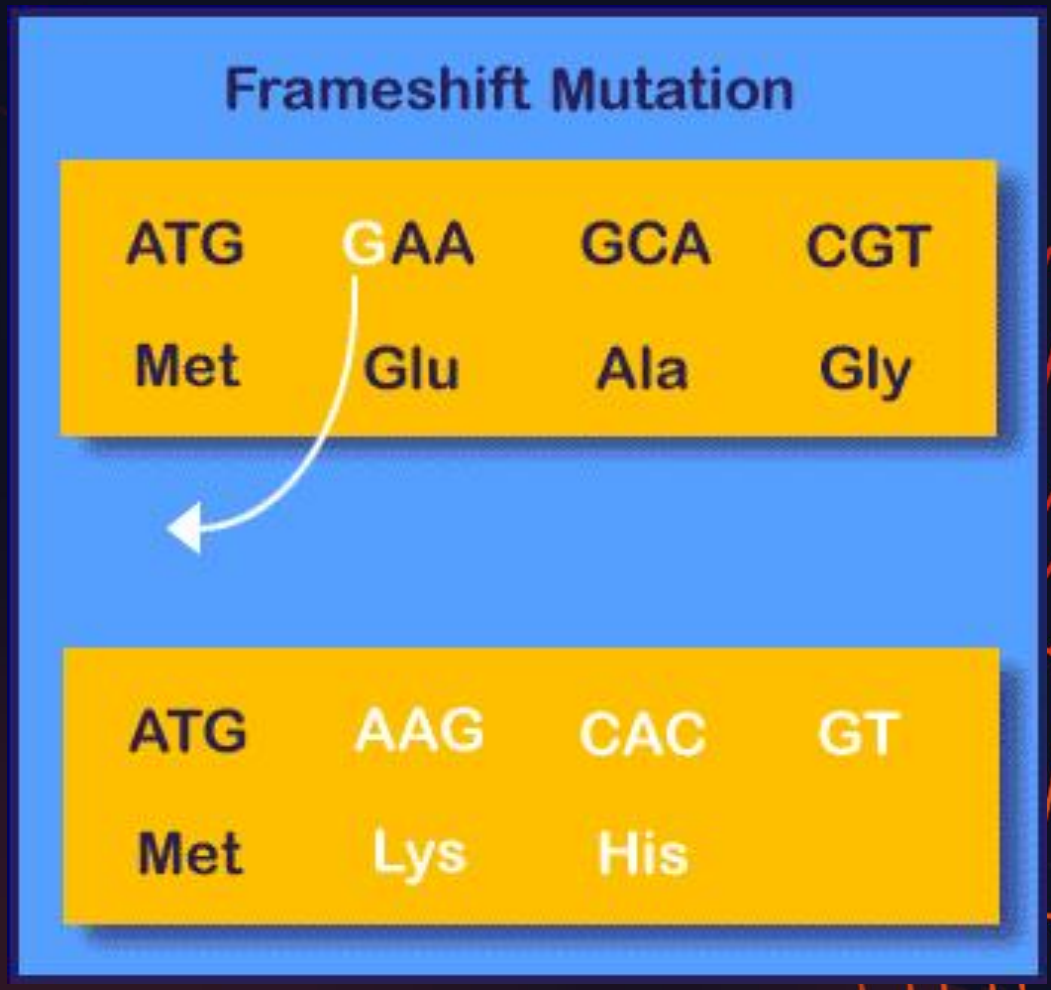
Frameshift Mutation

- **Inserting or deleting** one or more nucleotides
- Changes the "**reading frame**" like changing a sentence
- **Proteins** built **incorrectly**

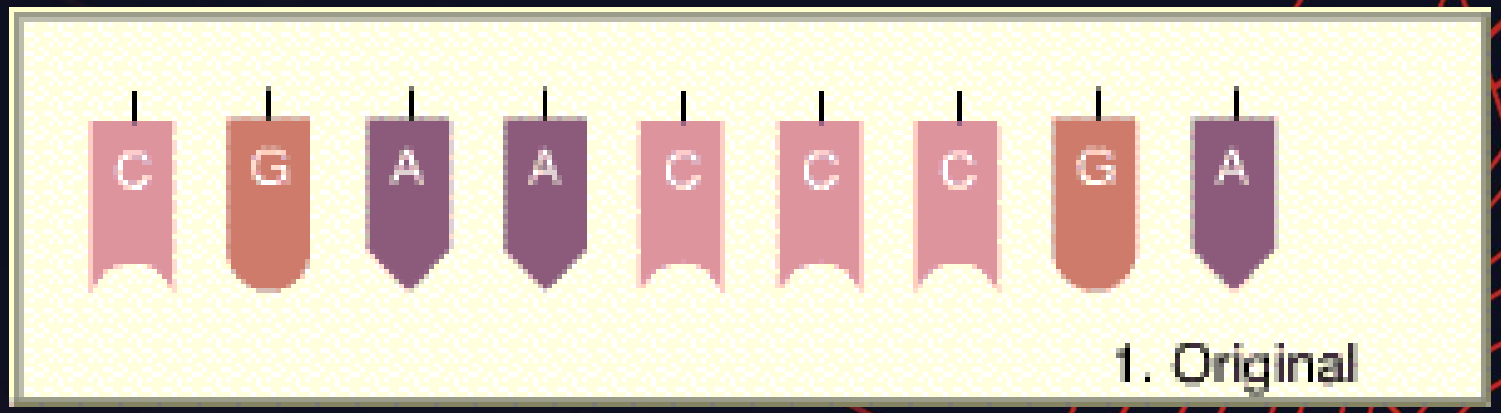
Frameshift Mutation

- Original:
  - The fat cat ate the wee rat.
- Frame Shift (“a” added):
  - **The fat c**a**a tet hew eer at.**

Amino Acid Sequence Changed

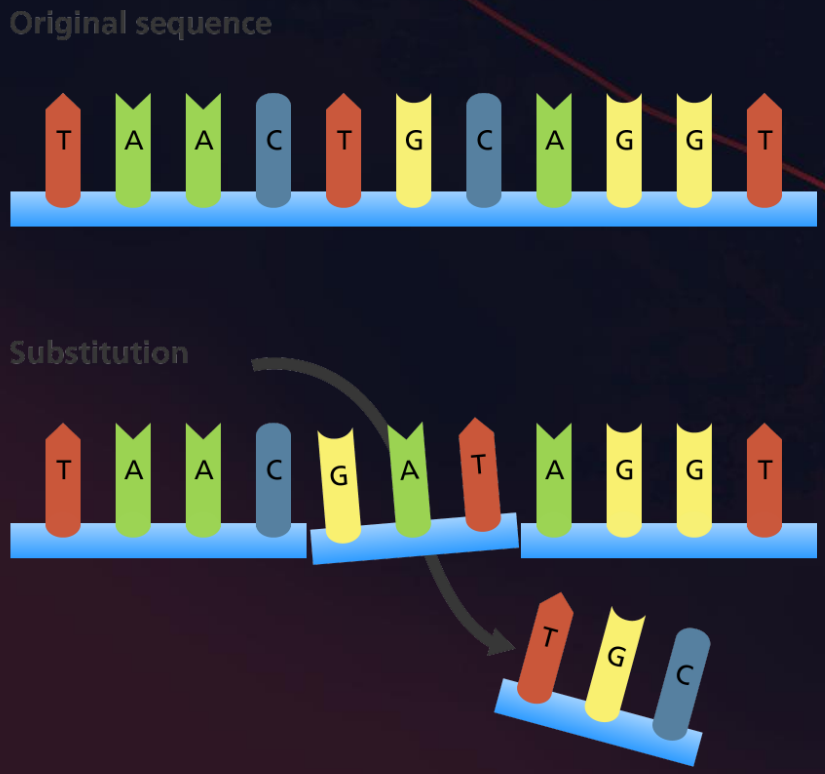


Gene Mutation Animation



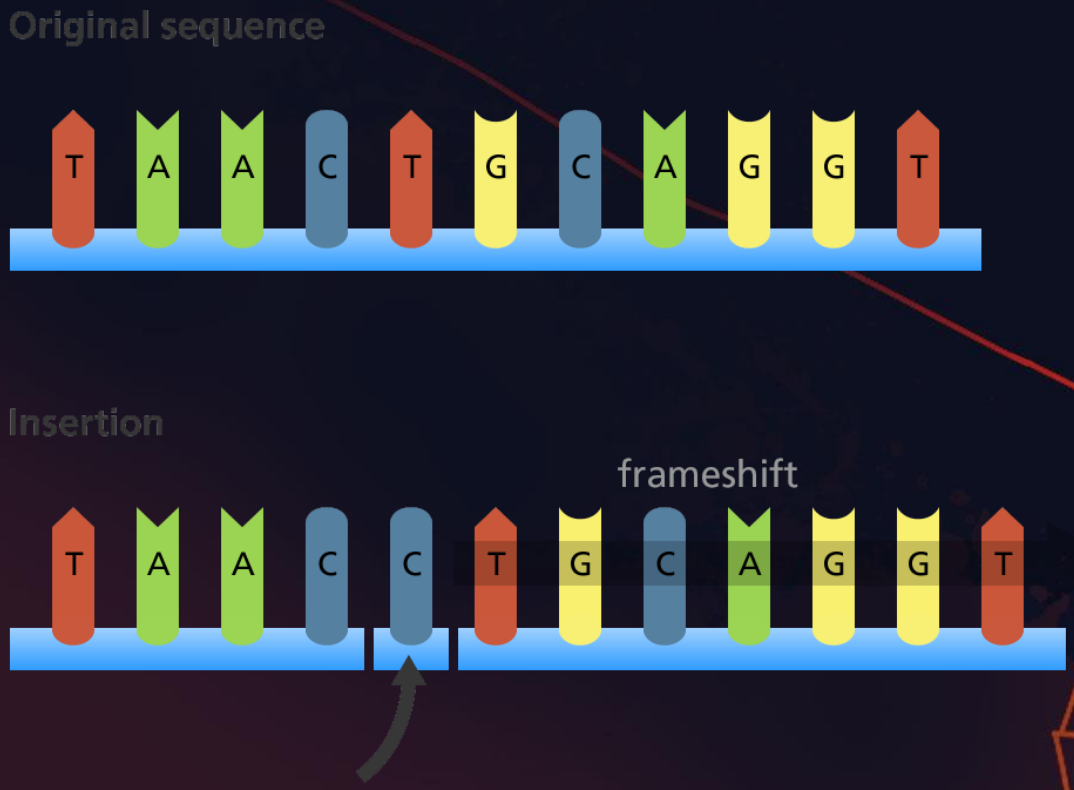
Substitution Mutation

A **substitution** is a **mutation** that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G)



Insertion Mutation

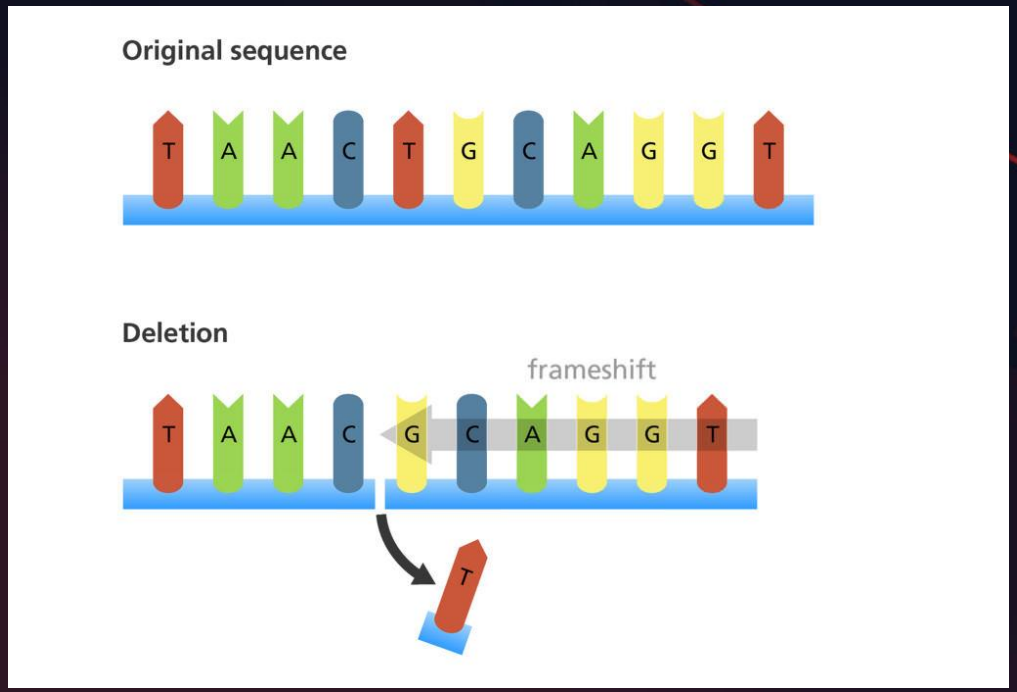
The addition of one or more nucleotide base pairs into a DNA sequence



Deletion Mutation

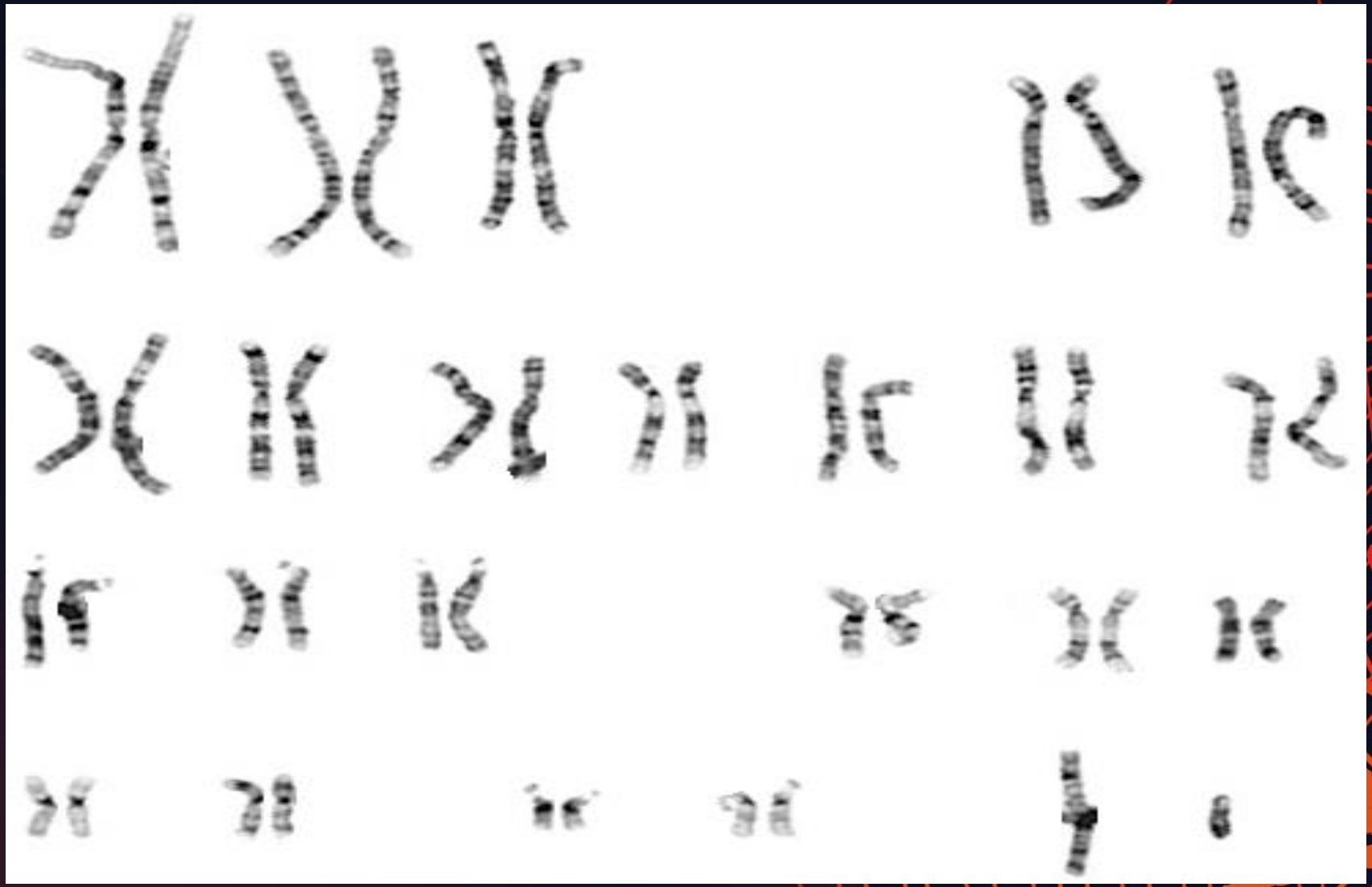
A part of a chromosome or a sequence of DNA is lost during DNA replication.

Any number of nucleotides can be deleted, from a single base to an entire piece of chromosome



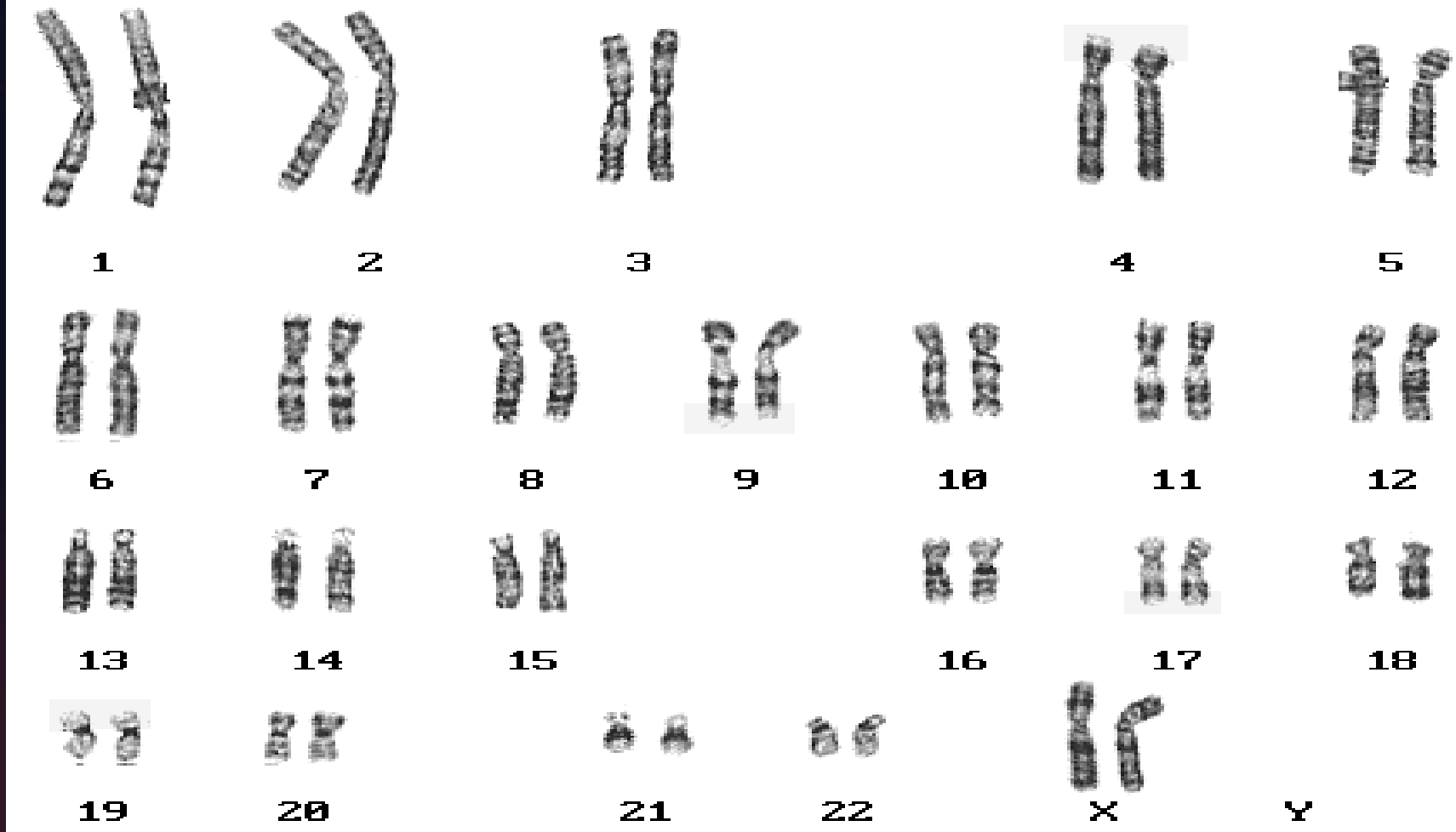

Substitution
Insertion
Deletion
Original sequence
TGGCAG
TGGCAG
TGGCAG
Mutated sequence
TGGTAG
TGGTATCAG
TGGG

Normal Male



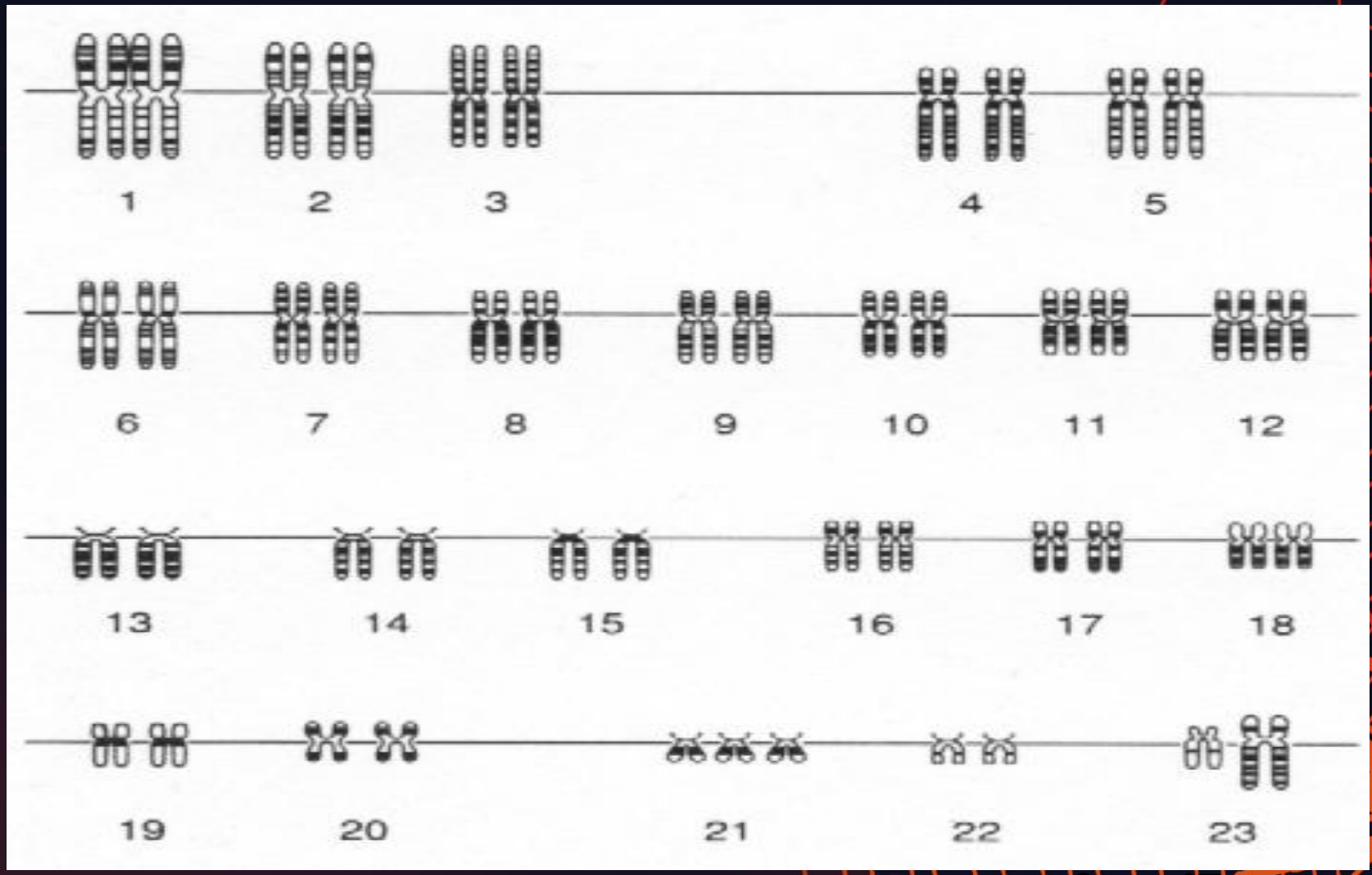
2n = 46 XY

Normal Female



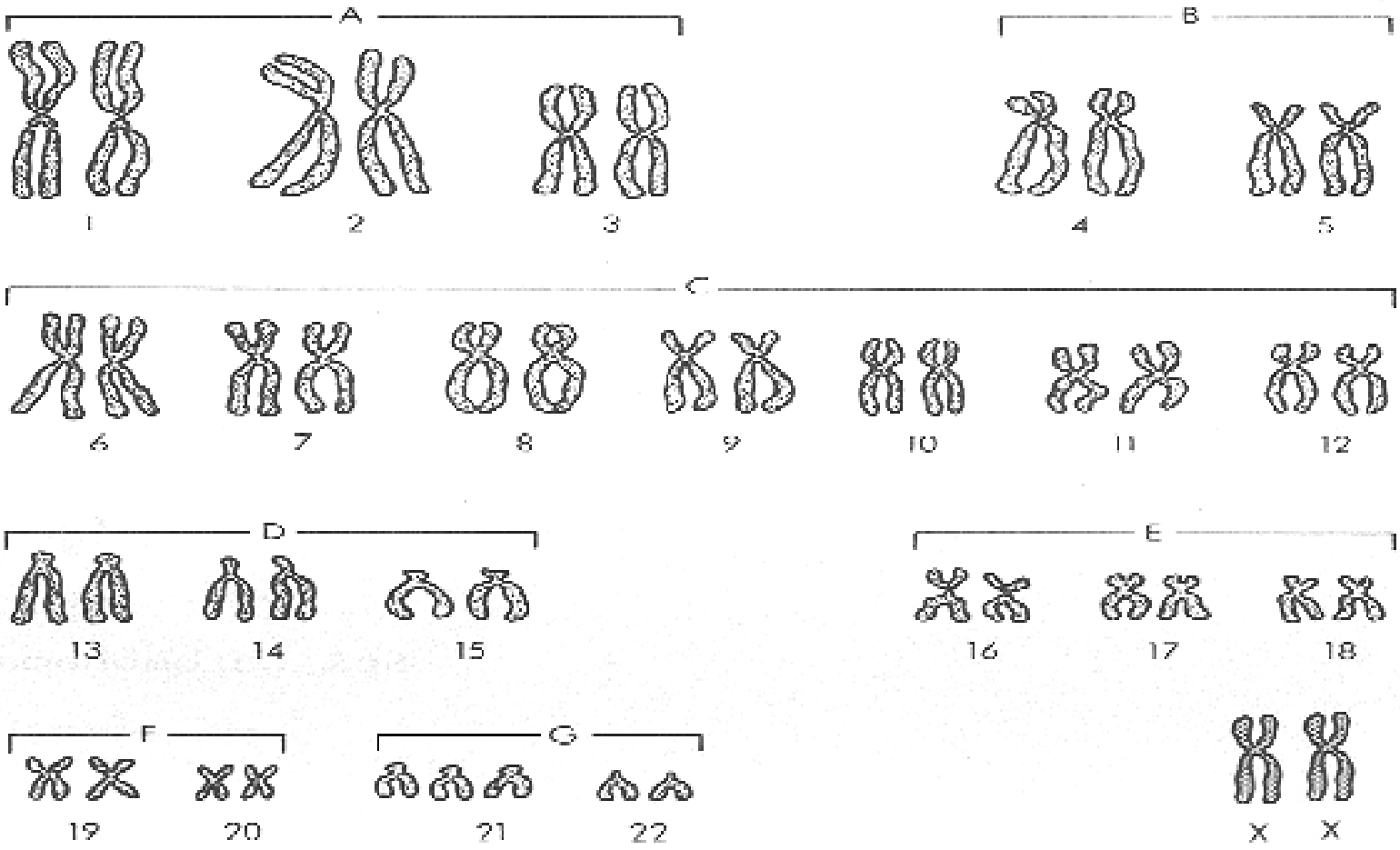
2n = 46

Male, Trisomy 21 (Down's)



2n = 47

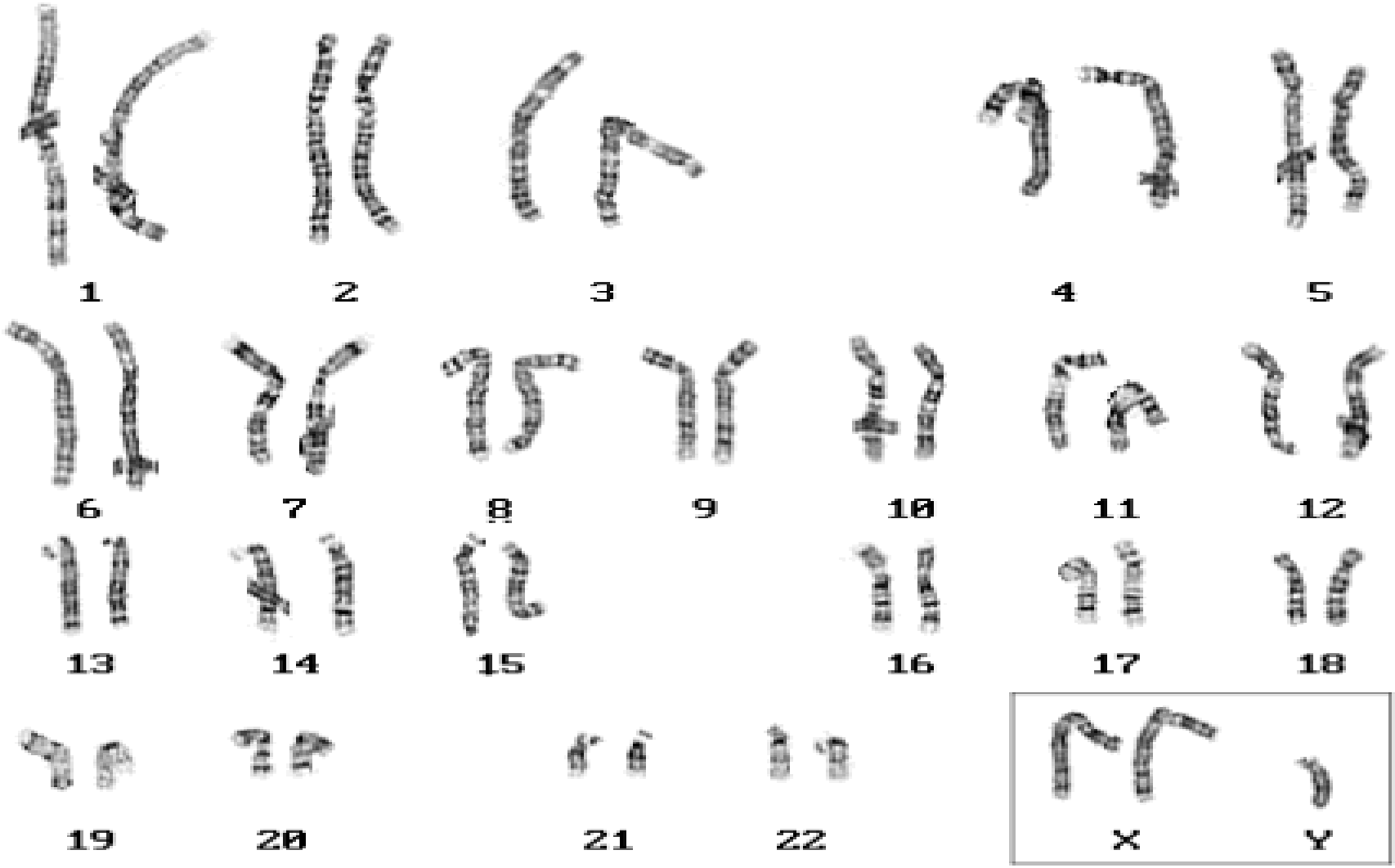
Female Down's Syndrome



Down Syndrome

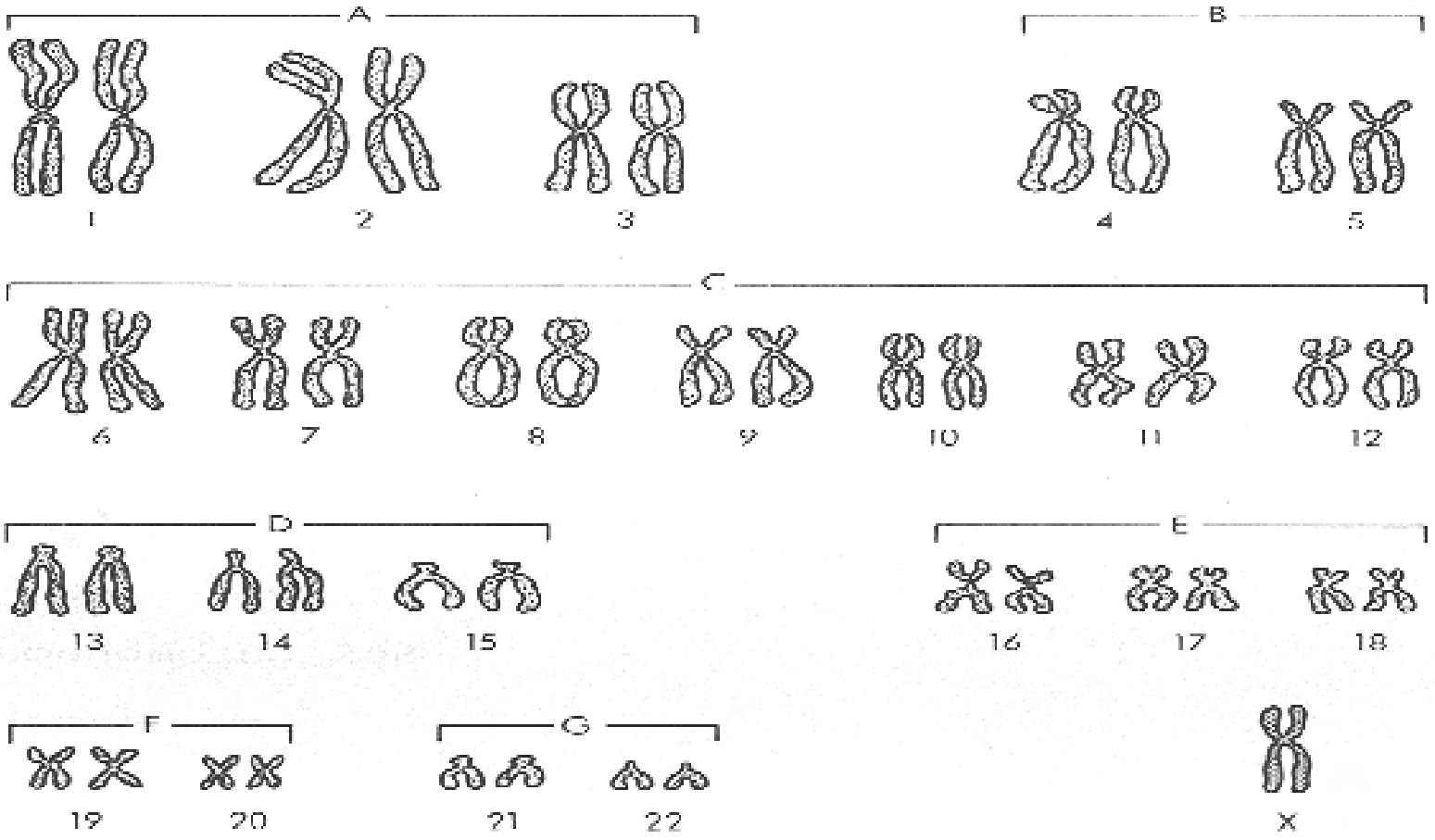
2n = 47

Klinefelter's Syndrome



2n = 47

Turner's Syndrome



Turner's Syndrome

2n = 45

Thank you…..