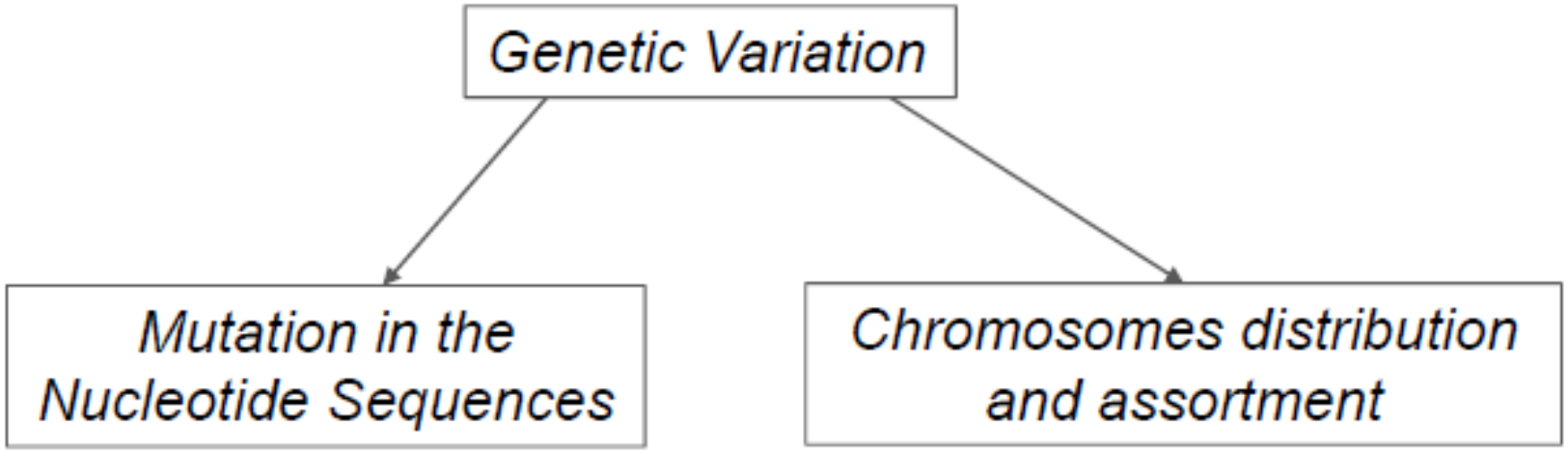
Section 4.2 – Genetic Variation

SBI3U

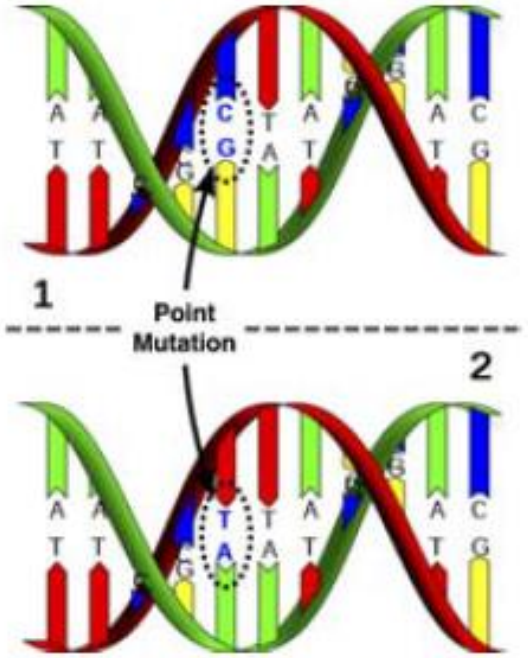
MRS. FRANKLIN

Genetic Variation



Genetic Variation – Nucleotide Sequence

Mutations are the original source of genetic diversity. The changes in genetic sequences create different versions of genes.



Reshuffling the different versions of genes during meiosis produces variations that result in a species having unique traits.

Genetic Variation – Chromosomes

Most of the variations arise from the behaviour of chromosomes during meiosis.

There are 3 main mechanisms that contribute to this diversity.

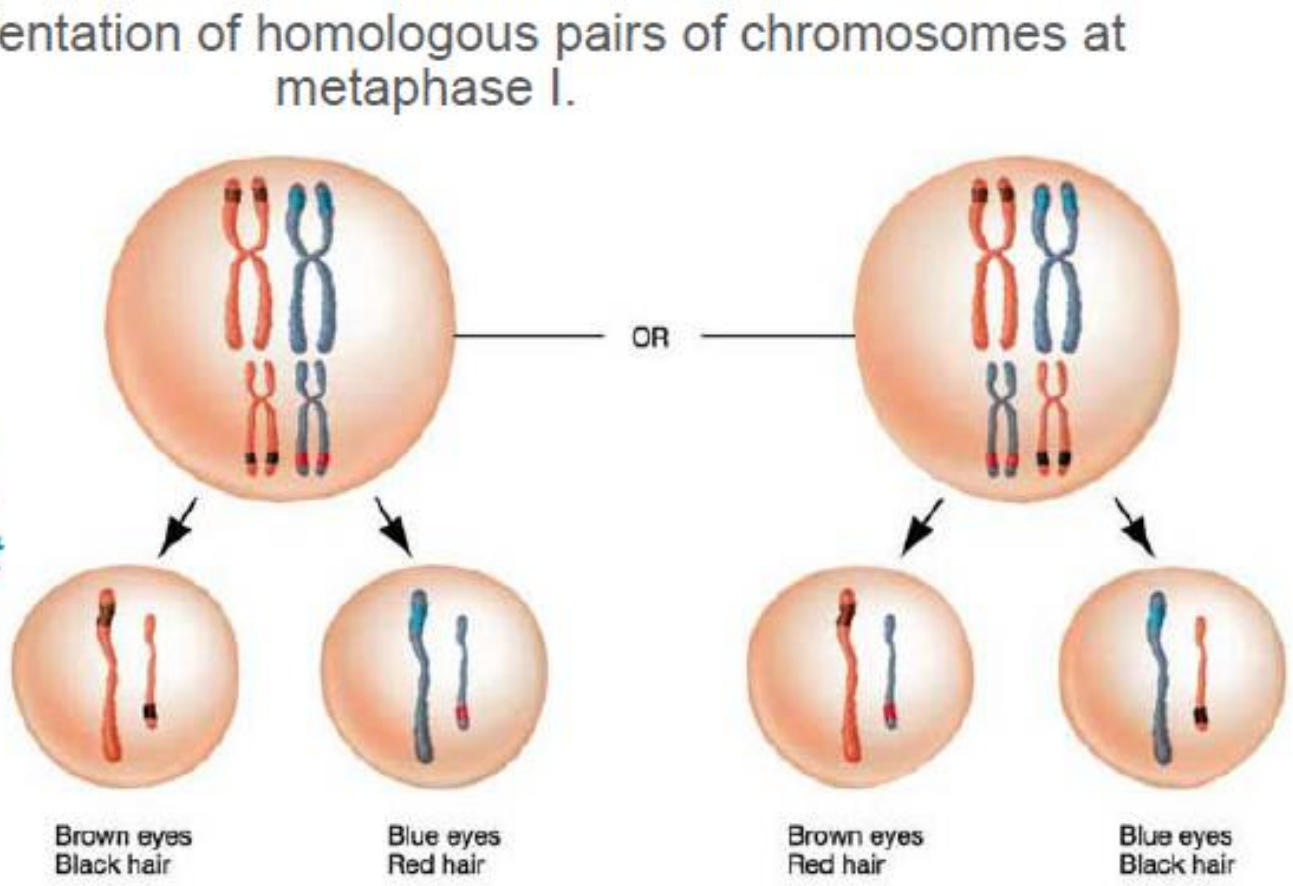
1) Independent Assortment of Chromosomes

2) Crossing Over

3) Random Fertilization

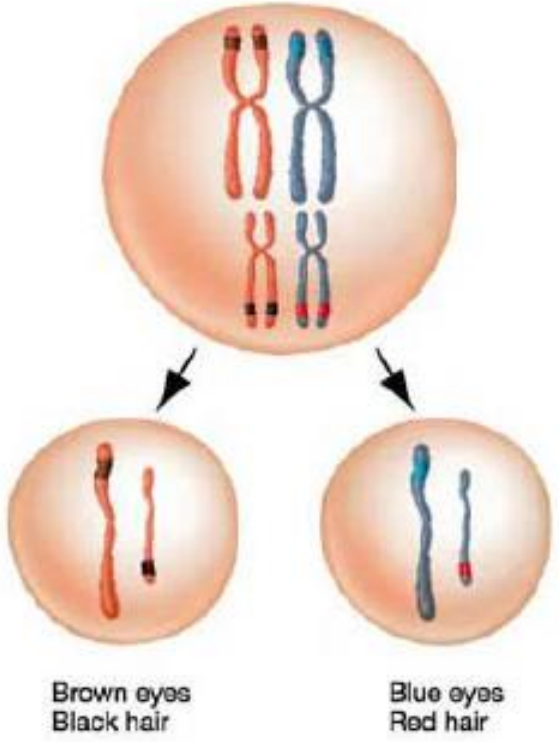
1) Independent Assortment

The orientation of homologous chromosomes is random. There is a 50/50 chance that a daughter cell will get the maternal or paternal chromosome.



1) Independent Assortment

Each homologous pair is positioned at the metaphase plate independently of other pairs.



Each haploid daughter cell represents 1 possible outcome.

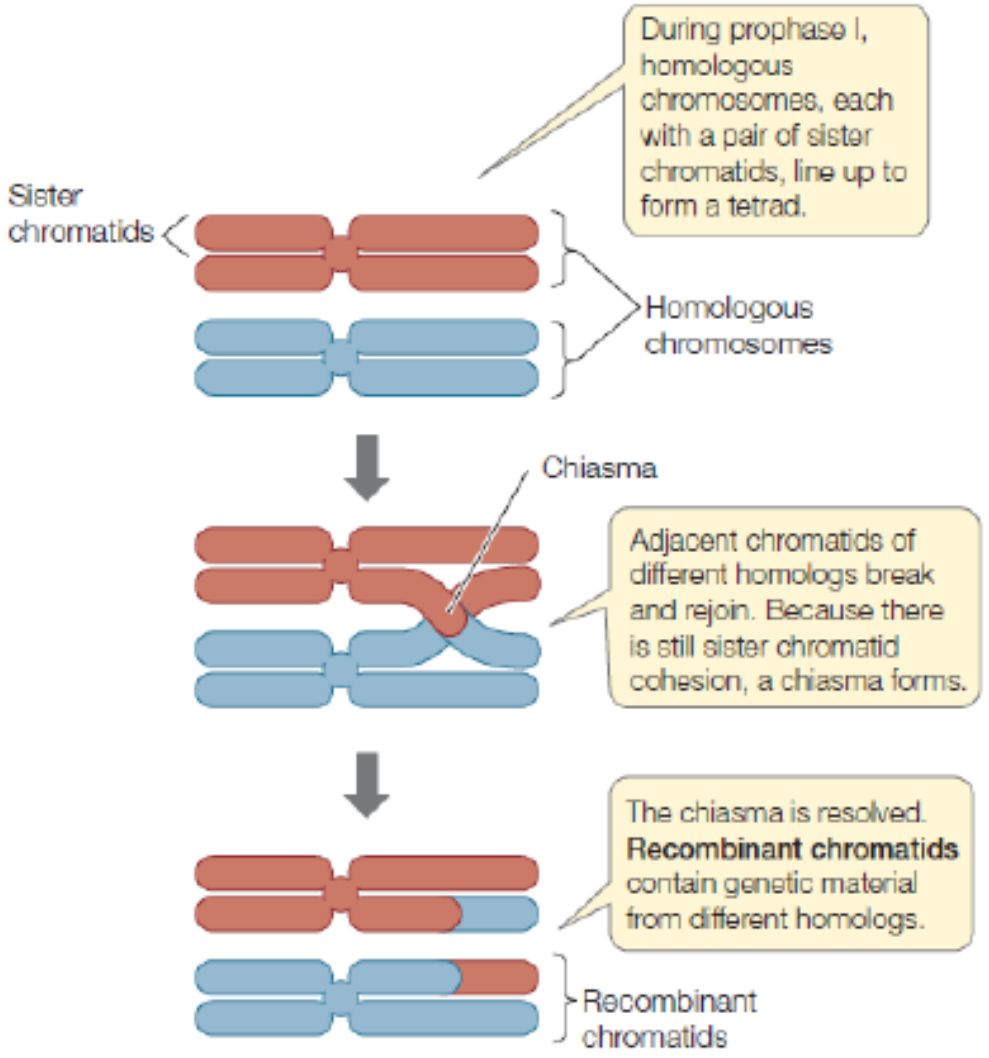
Number of possible chromosomal combinations is 2^n

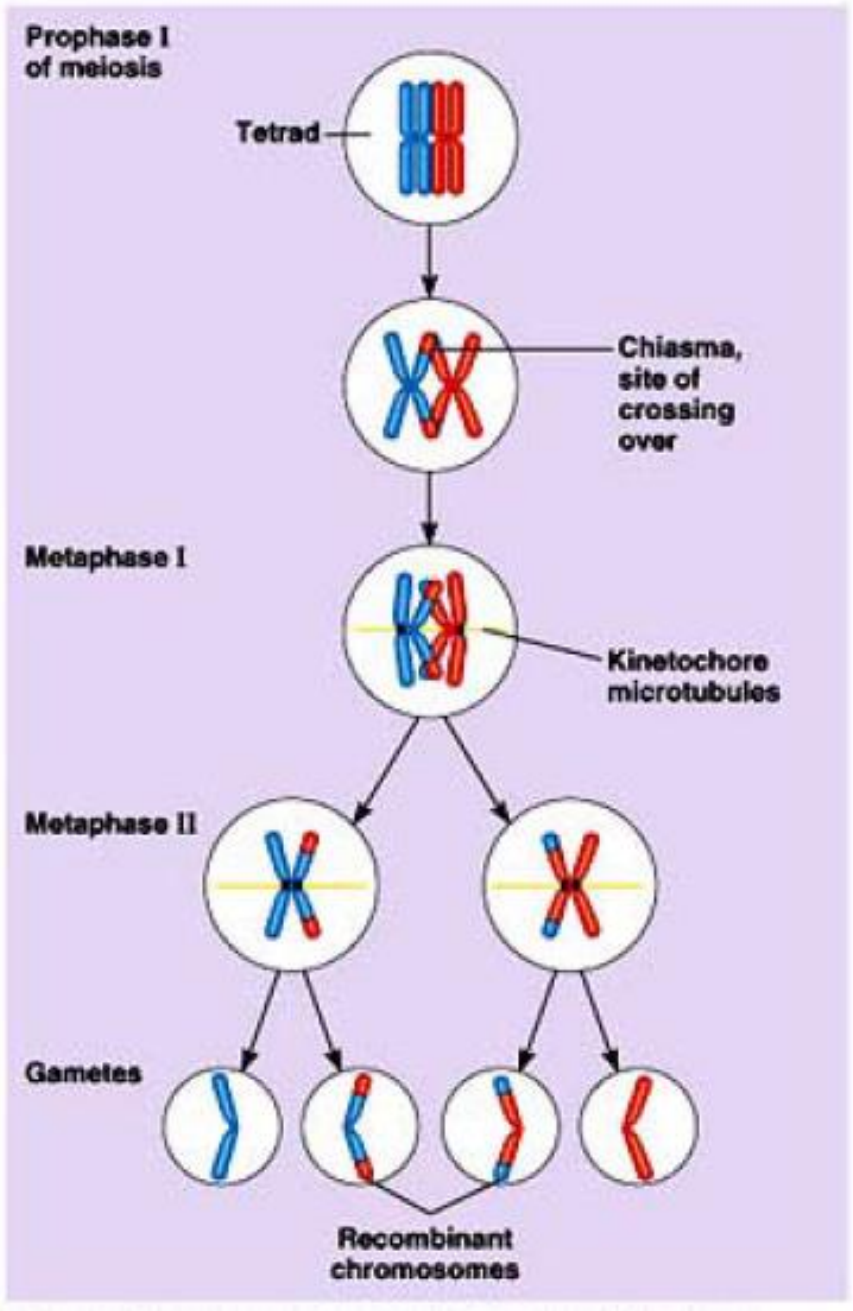
n = 23 (haploid number); $2^{23} = 8$ *million*

2) Crossing Over

The events of crossing over produce chromosomes with new combinations of maternal and paternal genes.

Crossing over is a random process and thus contributes to the genetic diversity in our population.

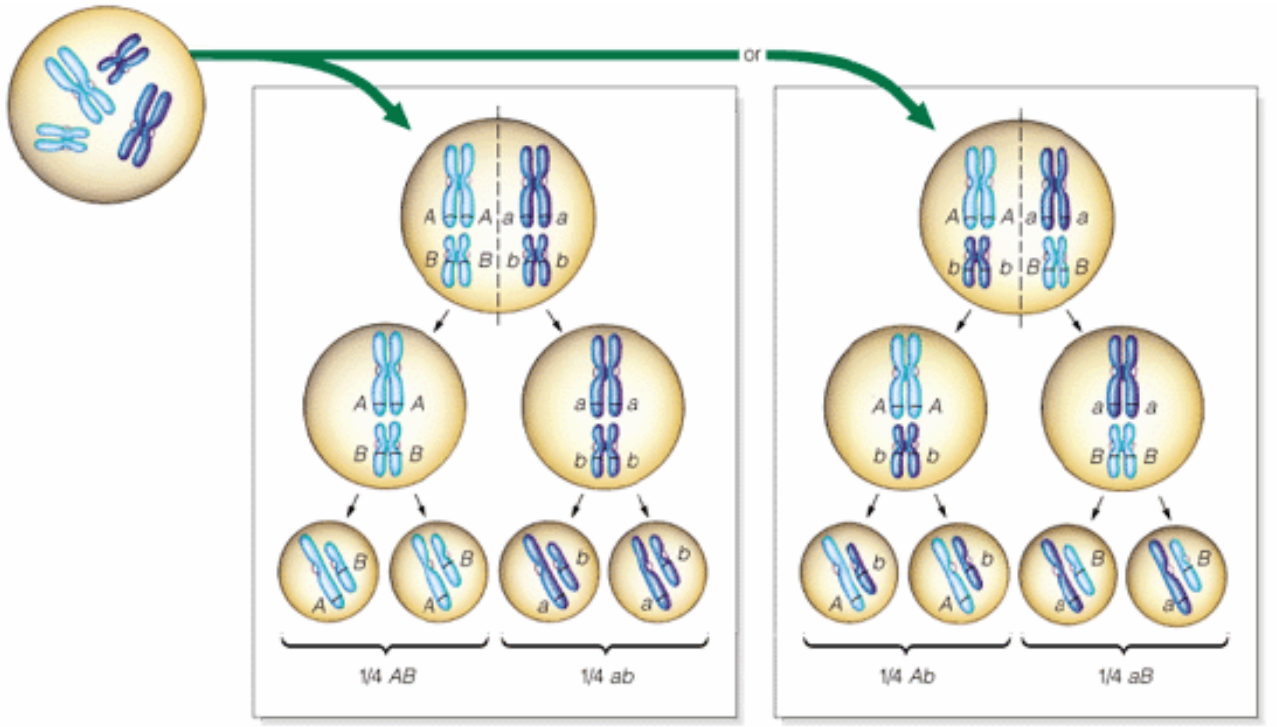




At metaphase II, chromosomes that contain recombinant chromatids can be oriented in different ways.

3) Random Fertilization

In humans, a female or male gamete represents one of the 8 million possible combinations.



The fusion of any one gamete will produce a zygote with one of 64 trillion diploid combinations.

Mistakes in Meiosis

Many genetic disorder in humans can be traced back to errors that occurs during gamete formation (meiosis).

Abnormalities an result from mistakes in the separation of chromosomes or incorrect exchange of information.

Two main types of mistakes that can occur:

1. Nondisjunction

2. Alteration of chromosome structure

1) Nondisjunction

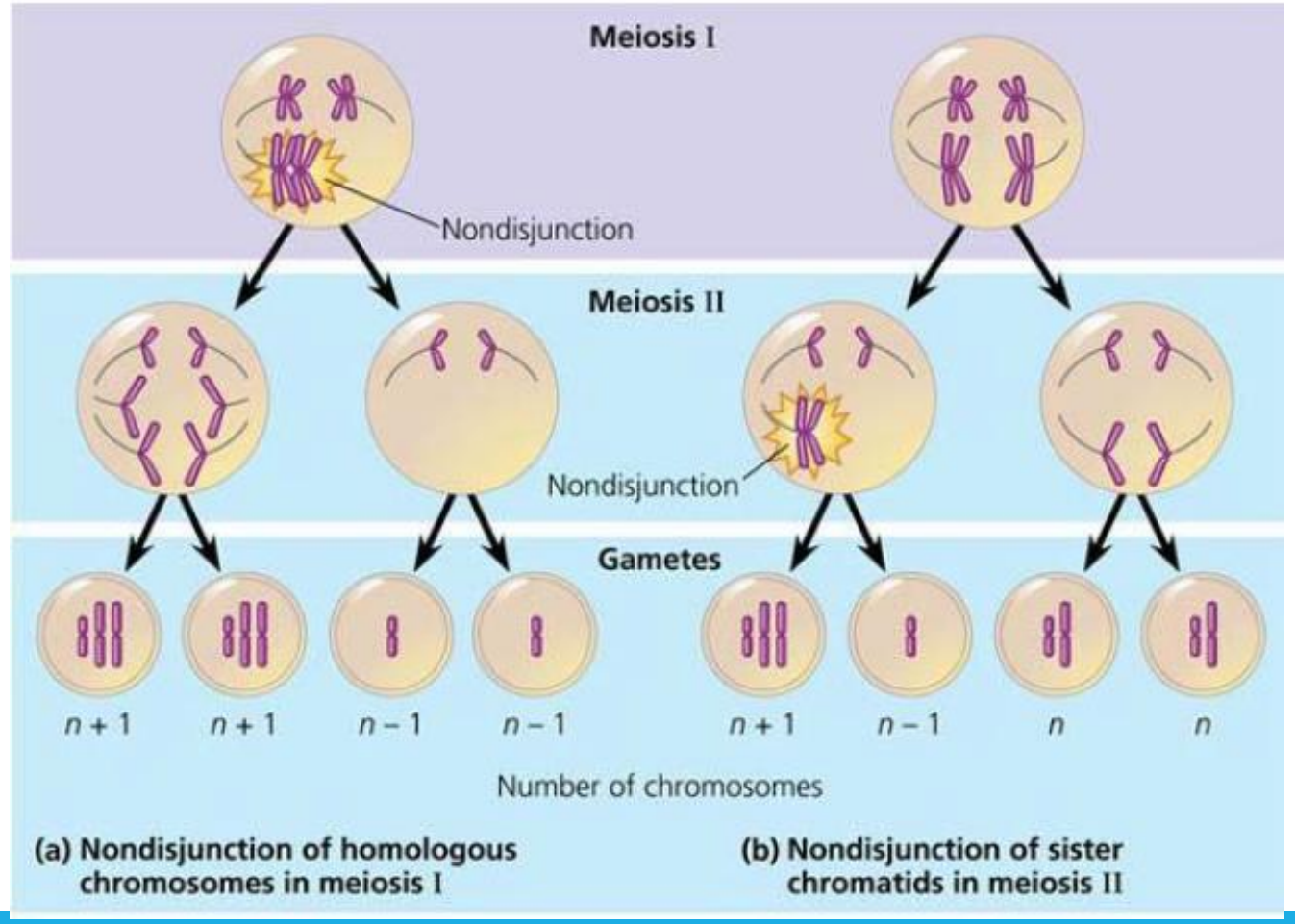
Occurs when:

a) Homologous pairs don't separate successfully during meiosis I

b) Sister chromatids do not separate during Meiosis II.

*** One gamete receives an extra copy of a chromosome and another gamete receives no copy.*

1) Nondisjunction



Monosomy: *missing a chromosome*

Trisomy: *contains an additional chromosome*

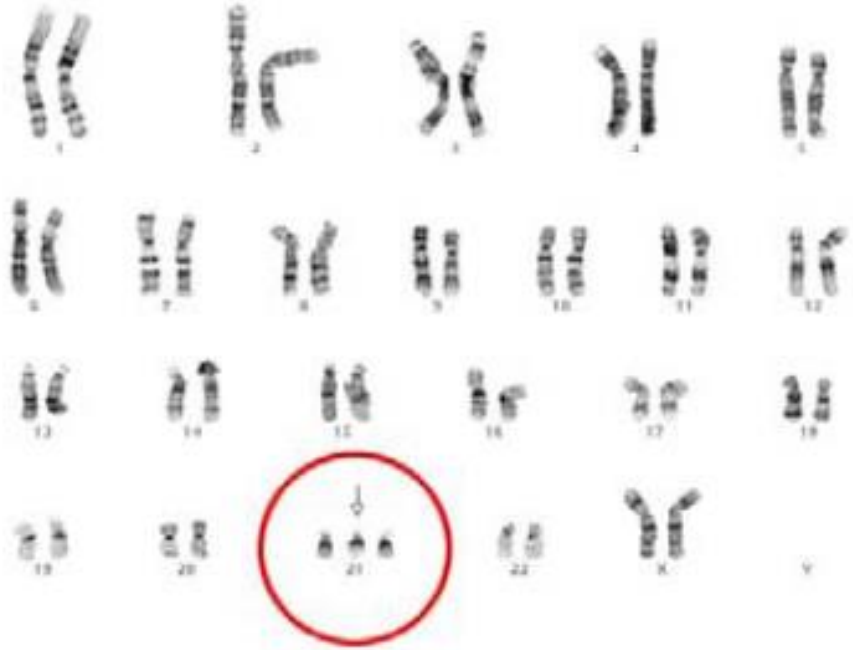
1) Nondisjunction

When the gametes fuse, if there is an abnormal number of chromosomes in one of the gametes, the zygote will become ***monosomic*** or ***trisomic,*** these are different forms of ***aneuploidy***.

Through mitosis, all cells in the body of that individual will contain that abnormal number of chromosomes. This can lead to many disorders and symptoms.

Autosomal Aneuploidy

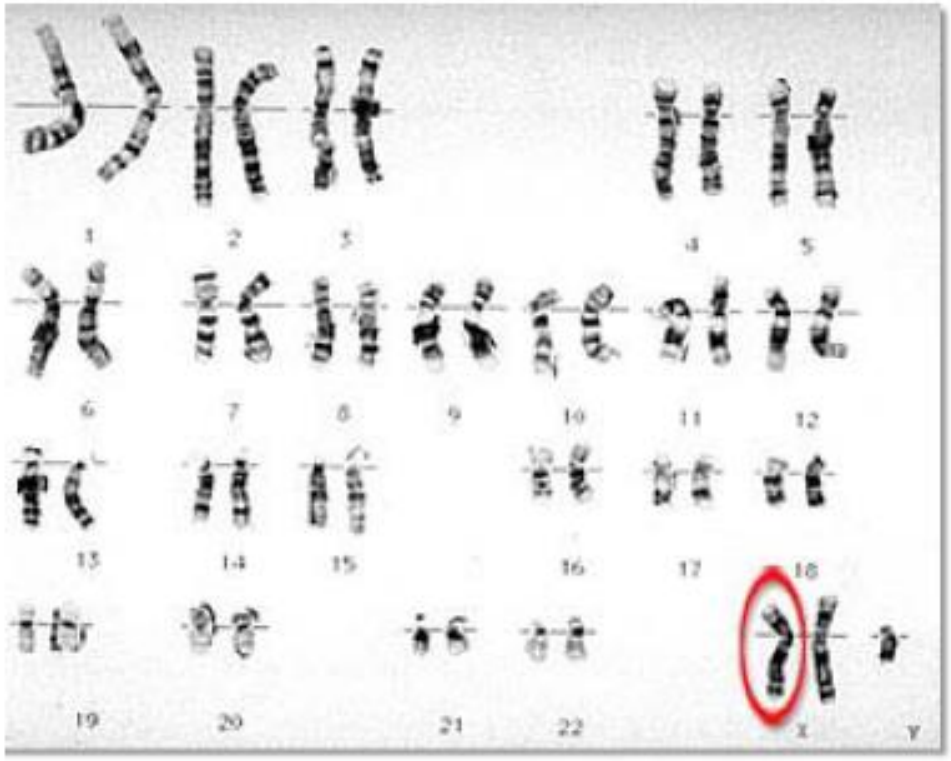
Down Syndrome: due to an extra chromosome 21 (Trisomy 21)



A somatic cell will contain 47 chromosomes.

Sex Chromosome Aneuploidy

Klinefelter Syndrome: an extra X chromosome in Males (**XXY**)



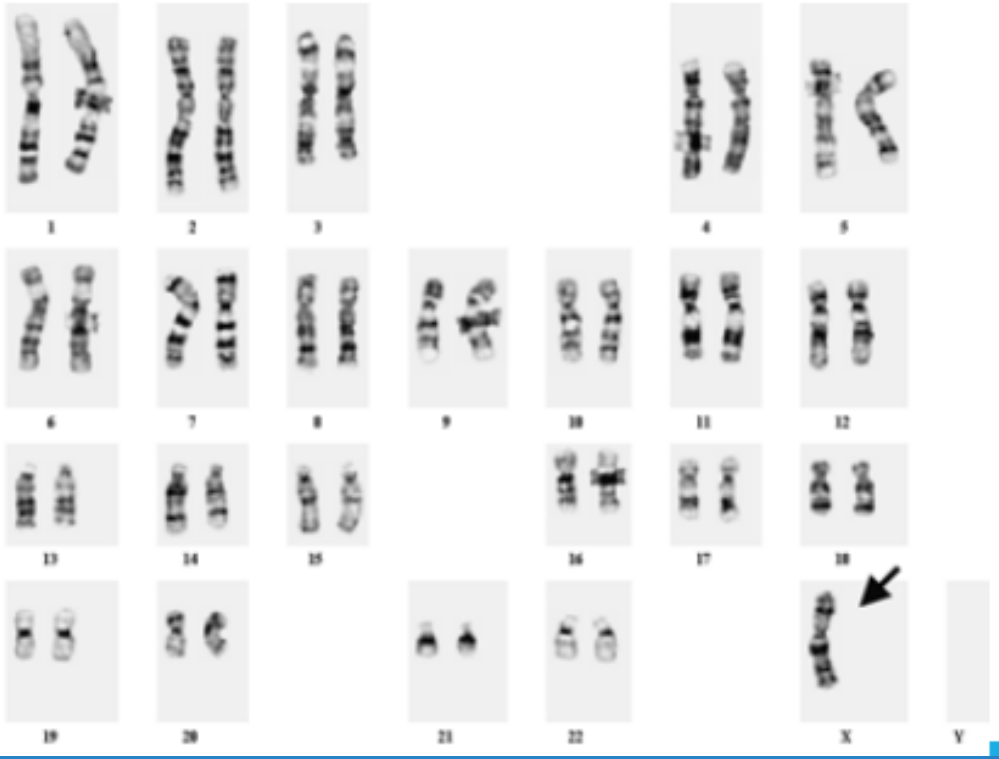
Symptoms:

Contain male sex organs but the testes are abnormally small and the man is sterile.

*The extra **X chromosome is inactivated**, but there can still be the development of breasts.*

Sex Chromosome Aneuploidy

Turner Syndrome: Females is missing an X chromosome **(XO)**, monosomy



Symptoms:

*They have female characterisitcs, they are **sterile** because the sex organs do not mature.*

Sex Chromosome Aneuploidy

Triple X (XXX): females have an extra X chromosome

Symptoms:

These females are healthy and cannot be distinguished form other XX females.

2) Alteration of Chromosome Structure

A breakage in the chromosome can result in 4 different structural changes:

1)Deletion

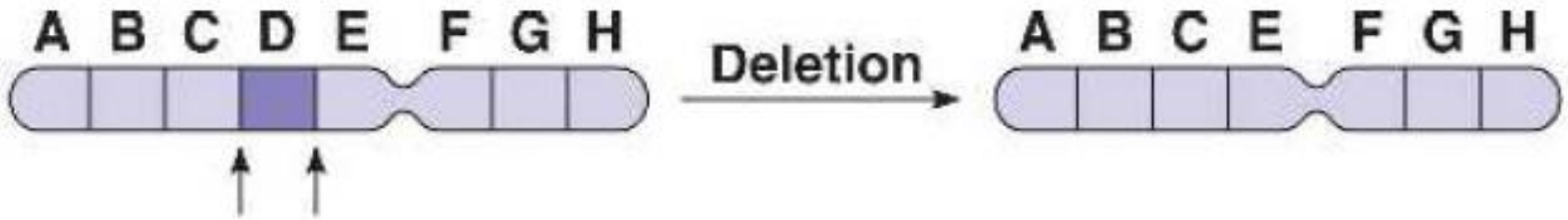
2)Duplication

3)Inversion

4)Translocation

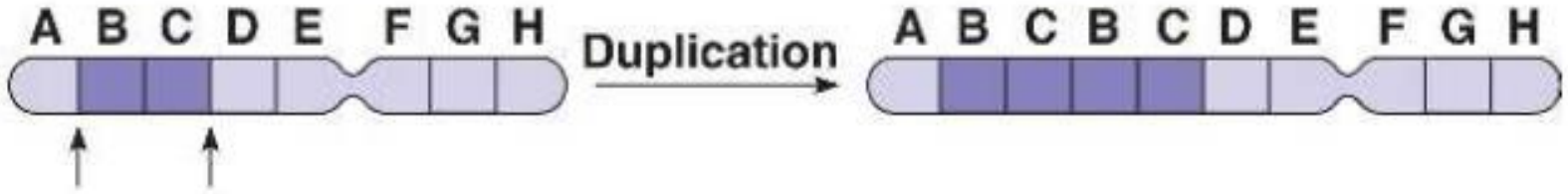
A) Deletion

A chromosomal fragment lacking a centromere is lost. The affected chromosome is missing genes



B) Duplication

A set of genes is duplicated and repeated on a chromosome.Thus causing there to be 2 sets of the same genes.



C) Inversion

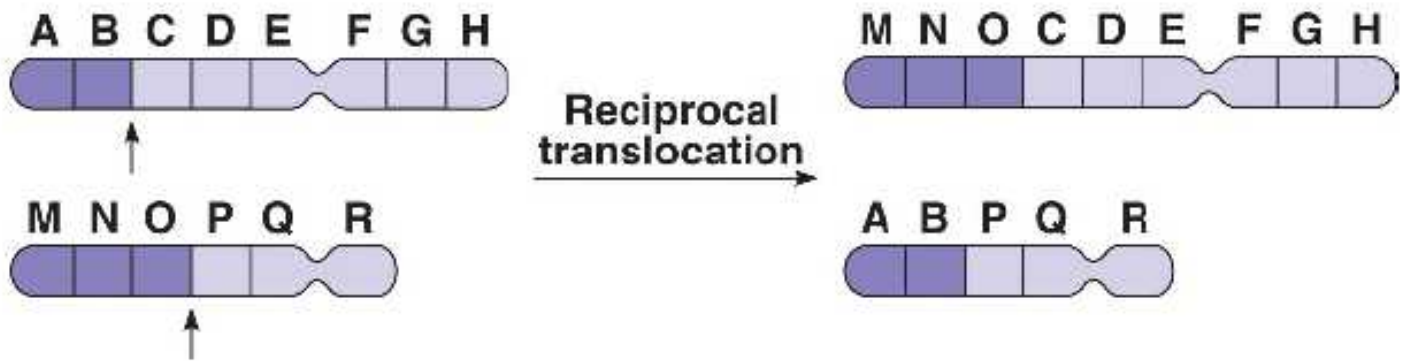
A chromosomal fragment can reattach to the original chromosome but in reverse order.

D) Translocation

A segment moves from one chromosome to a nonhomologous chromosome

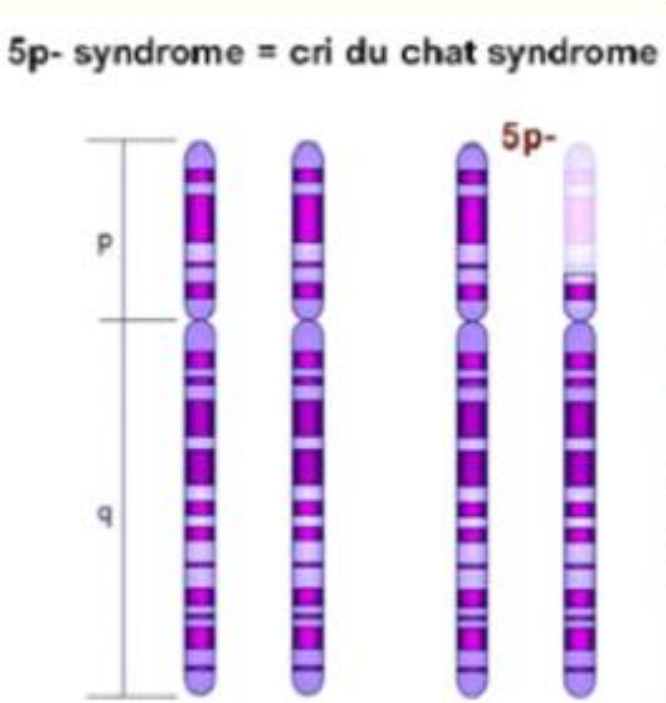
Reciprocal translocation: both chromosomes exchange informaiton

Nonreciprocal: the other chromosome does not exhcange information.



Example: Deletion

Cri du Chat: Deletion on chromosome 5

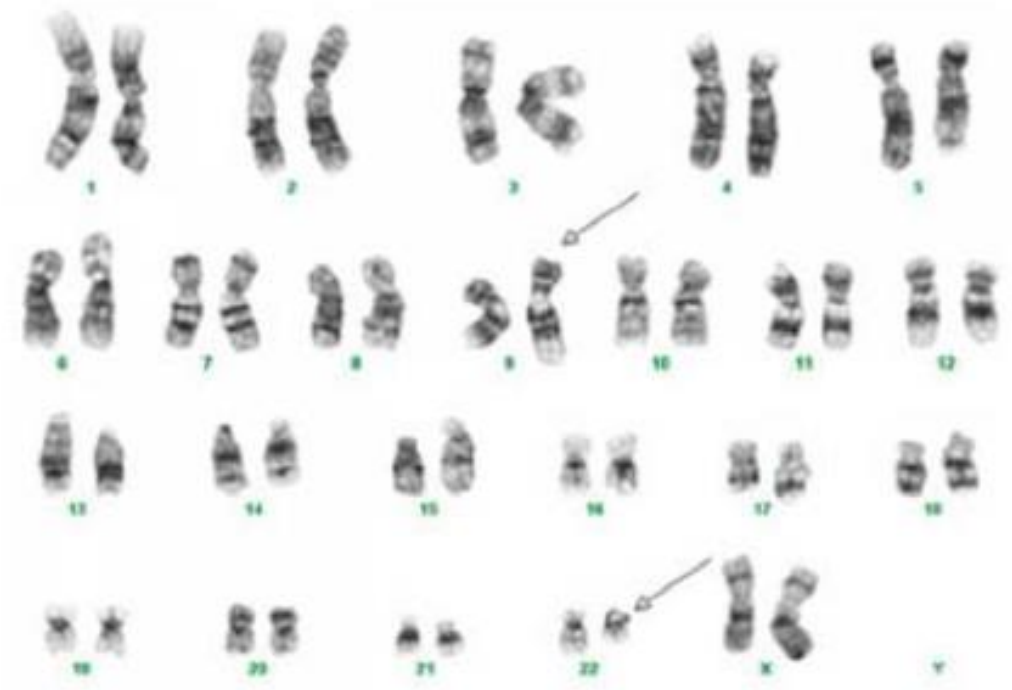


Symptoms:

The child is mentally disabled, small head with unusual facial features. Children with this disorder generally die in infancy or early childhood.

Reciprocal Translocation

Chronic Myelogenous Leukemia (CML): Reciprocal translocation between chromosome 22 and chromosome 9. The chromosome 22 is abnormally short.



Checking for Understanding

1. Which of the following correctly describes the changes in chromosome structure?

A) *inversion:* part of one chromosome becomes attached to another chromosome.

B) *deletion:* a piece of chromosome is inverted

C) *duplication*: a complete chromosome is copied

D) *translocation*: part of a chromosome is copied

E) *translocation*: part of one chromosome becomes attached to another chromosome.

Homework

Textbook: p. 181 # 10, 11 & 13