The module: Molecules, Genes and Diseases (MGD)
Session 6
Lecture 11
Duration: 1 hour

Inheritance of Genes

Mitosis and meiosis, genotypes and phenotypes

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Human Heredity Chapters 2, 3, 4, 5
Lippincott's Illustrated Reviews: Cell and Molecular Biology Chapter 20
Complete and review questions started in work session 6.

For more detailed instruction, any question, cases need help please post to the group of session.

Intended learning outcomes of Lecture 8

At the end of this lecture you should be able to:

- ► Describe the process and role of mitosis **(LO.1)**
- ► Describe the process and role of meiosis. **(LO.2)**
- ► Distinguish clearly between genotype and phenotype. **(LO.3)**
- ► Explain how environmental factors have an influence on both phenotype and genotype. **(LO.4)**
- ► Distinguish clearly between gene and allele. **(LO.5)**

Reminder:

(LO.1)

☼ In humans, chromosomes exist in pairs.

☼ Most human cells, called somatic cells, carry 23 pairs, or 46 chromosomes, but certain cells, such as sperm and eggs, carry only one copy of each chromosome and have 23 unpaired chromosomes. **Why?**

☼ ♂ have one pair of chromosomes that are not completely matched.

☼ Members of this pair are known as **sex chromosomes** and are involved in sex determination.

☼ There are two types of sex chromosomes: [X and Y] ♂ carry X & Y chromosomes, and ♀ carry XX chromosomes.

☼ All other chromosomes are known as **autosomes**.

☼ Chromosomes carry genetic information that ultimately determines the structure, shape, and functions of the cell.

☼ This genetic information is contained in the sequence of nucleotide subunits in DNA and organized into genes.

KEEP IN MIND AS YOU READ

- Many genetic disorders alter cellular structure or function.
- Gaucher disease affects lysosomal function.
- Cancer is a disease of the cell cycle.
- Meiosis maintains a constant chromosome number from generation to generation.

Describe the process and role of mitosis

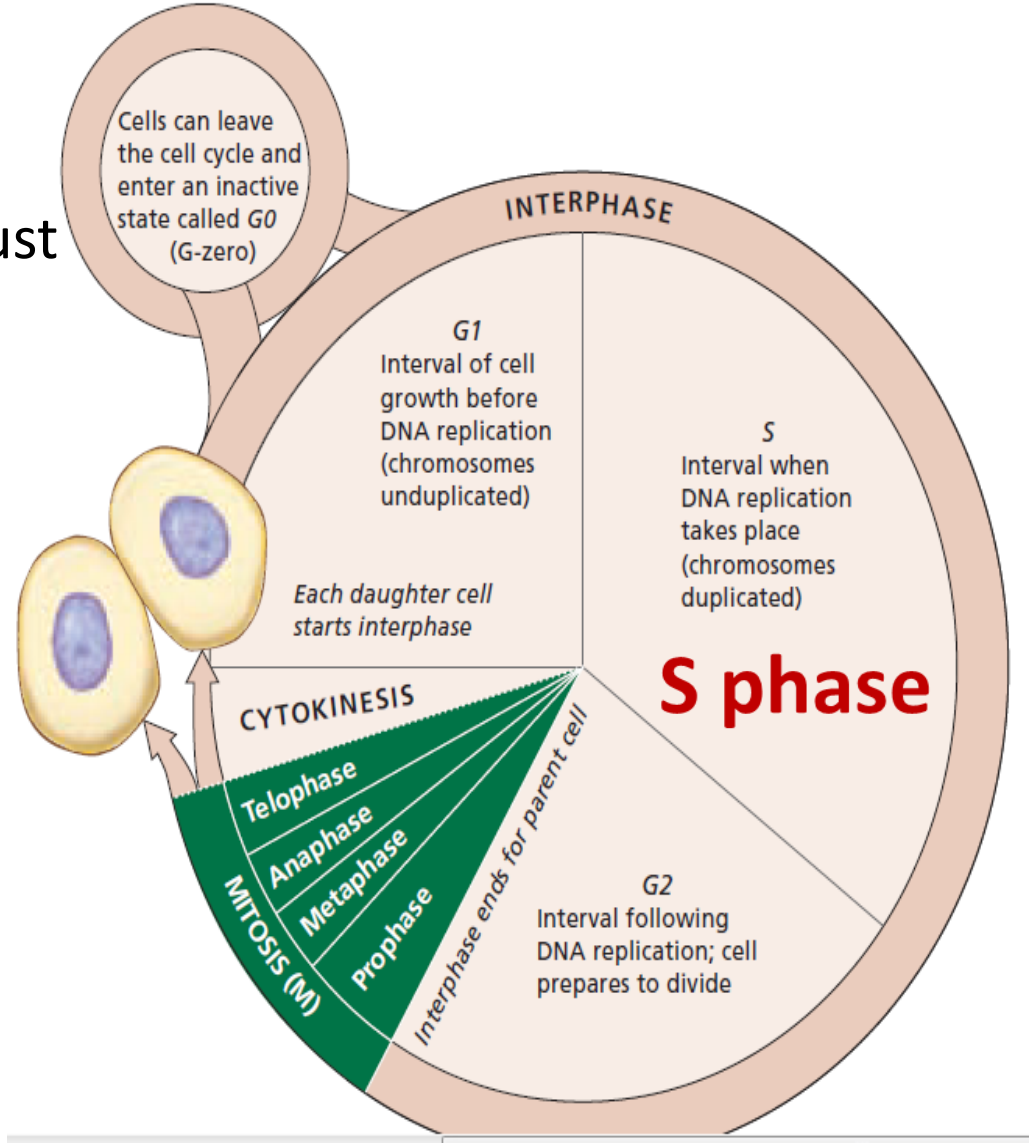
(LO.1)

When an organism requires additional cells, either for **growth** or to **replace** those normally lost, new ones must be produced by cell division, or proliferation.

Before a cell can divide all genetic information is **duplicated** during the **S phase** of the cell cycle.

Each chromosome duplicates, creating the classical X-shape;

each chromosome now consists of two identical sister **chromatids** that are touching in a structure called the **centromere**.



☺Spotlight on . . .

Cell Division and Spinal Cord Injuries

Many highly differentiated cells, such as those of the nervous system, do not divide. They move from the cell cycle into an inactive state called G0 (G-zero). The result is that injuries to nervous tissue, including the spinal cord, cause permanent loss of cell function and paralysis.

(LO.1)

Spina bifida (SB) is one of the most common and most complex birth defects involving the nervous system.(Module?) It occurs with a frequency of 1-2 per 1,000 births in the United States, but is higher in other populations. Spina bifida is one of a group of disorders called neural tube defects. The neural tube forms early in embryonic development and gives rise to the brain and the spinal cord. Neural tube defects occur during days 17 and 30 of development. **(Module?)**

Research has shown that nutrition, especially the amount of folate (Module?) in the diet has a significant impact on the frequency of SB.

Mitosis (nuclear division)

Is a continuous process and can be divided into **five** phases based on progress made to a specific point in the overall nuclear division.

After completion of nuclear division, **cytokinesis** occurs, involving **cytoplasmic** division and resulting in the formation of **two** separate daughter cells from the one parent cell.

☺ **When cells escape from the controls that are part of the cell cycle, they can become cancerous.**

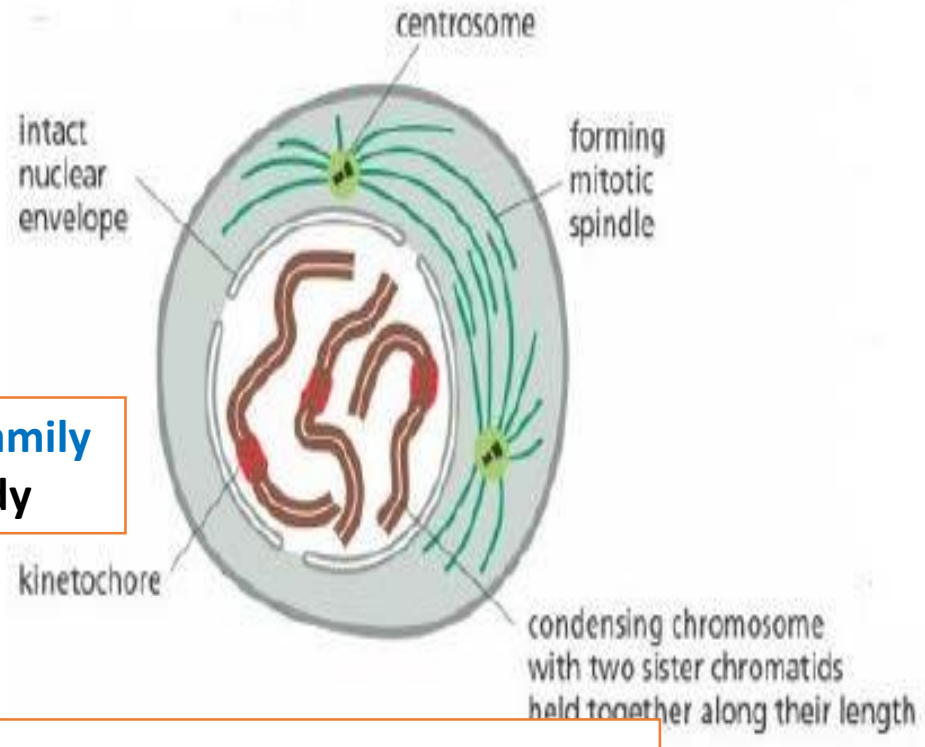
(LO.1)

1. Prophase

- In prophase, the nuclear envelope remains intact.
- Specialized protein complexes, called kinetochores, form and associate with each chromatid.

- Mitotic spindle microtubules will attach to each kinetochore.

The microtubules of the cytoplasm disassemble and then reorganized on the surface of the nucleus to form the mitotic spindle.



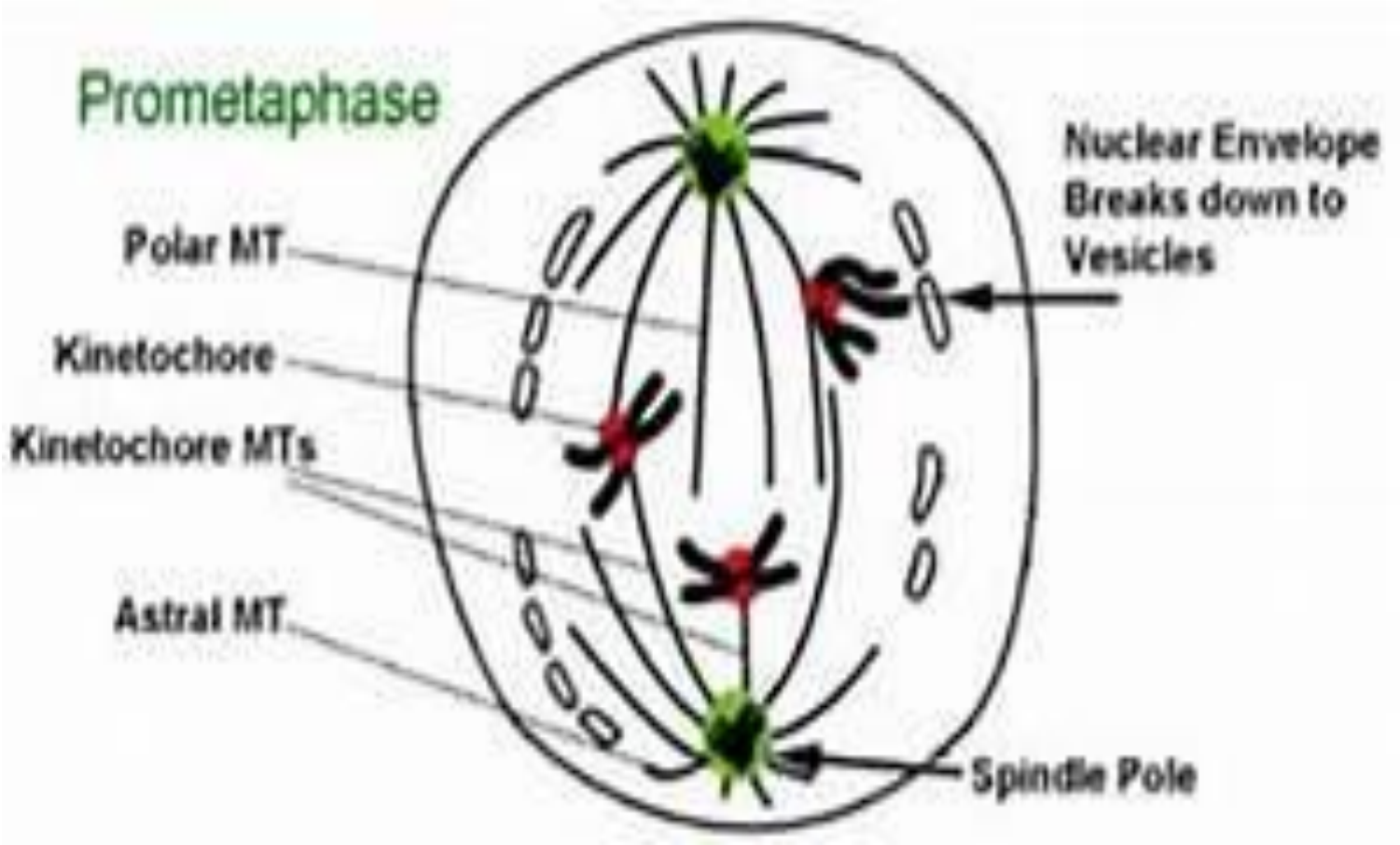
Aurora kinase family (Role)? Self Study

- Two centriole pairs push away from each other by growing bundles of microtubules forming the mitotic spindle.
- The nucleolus disassembles in prophase.

☺ Elevated expression of the aurora kinase family has been observed in many human tumors. Inhibitors of aurora kinases are being assessed as anticancer therapies.

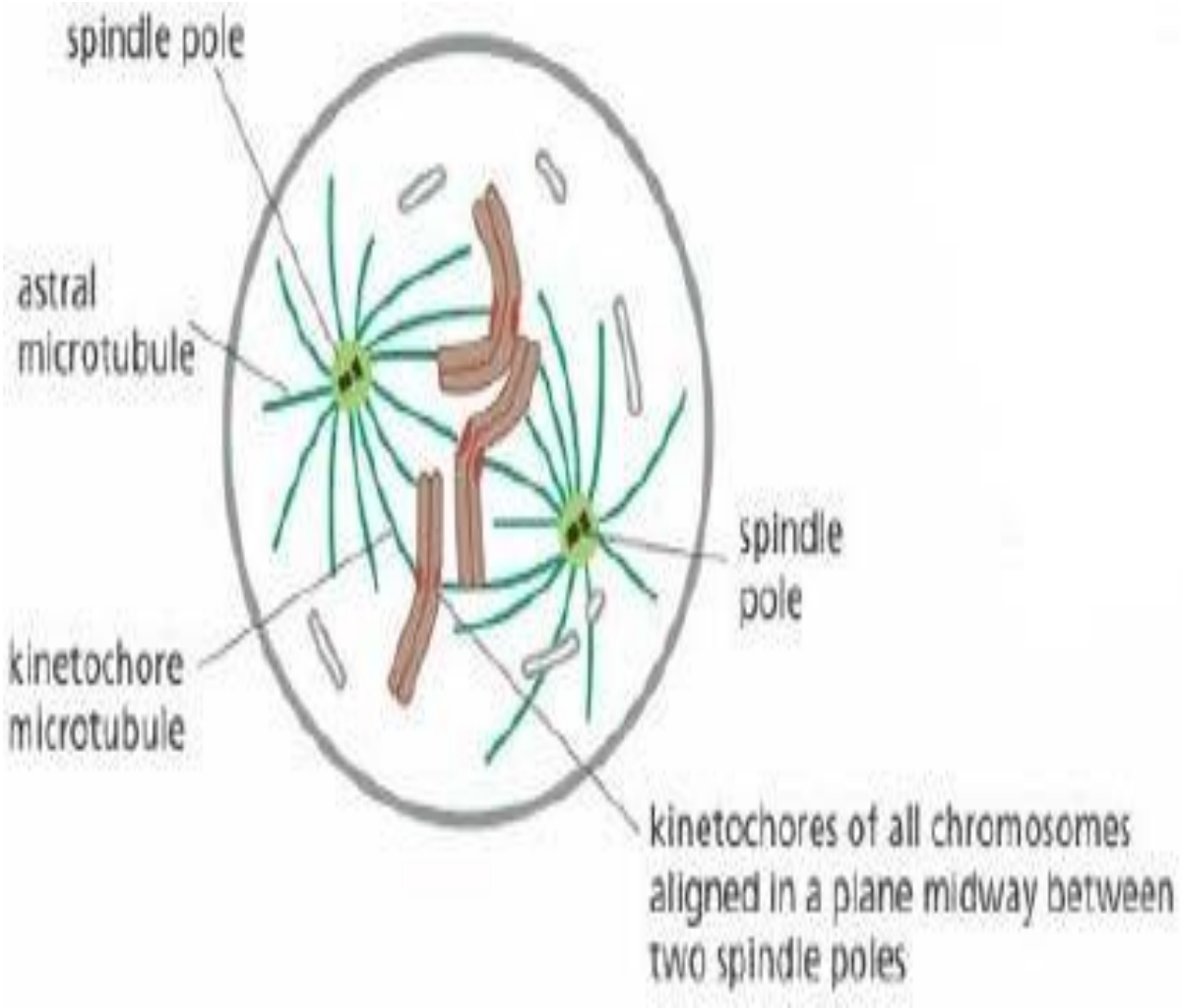
2. Prometaphase

- The disassembly of the nuclear envelope marks the beginning of prometaphase.
- Spindle microtubules bind to kinetochores and chromosomes are pulled by the microtubules of the spindle.



3. Metaphase

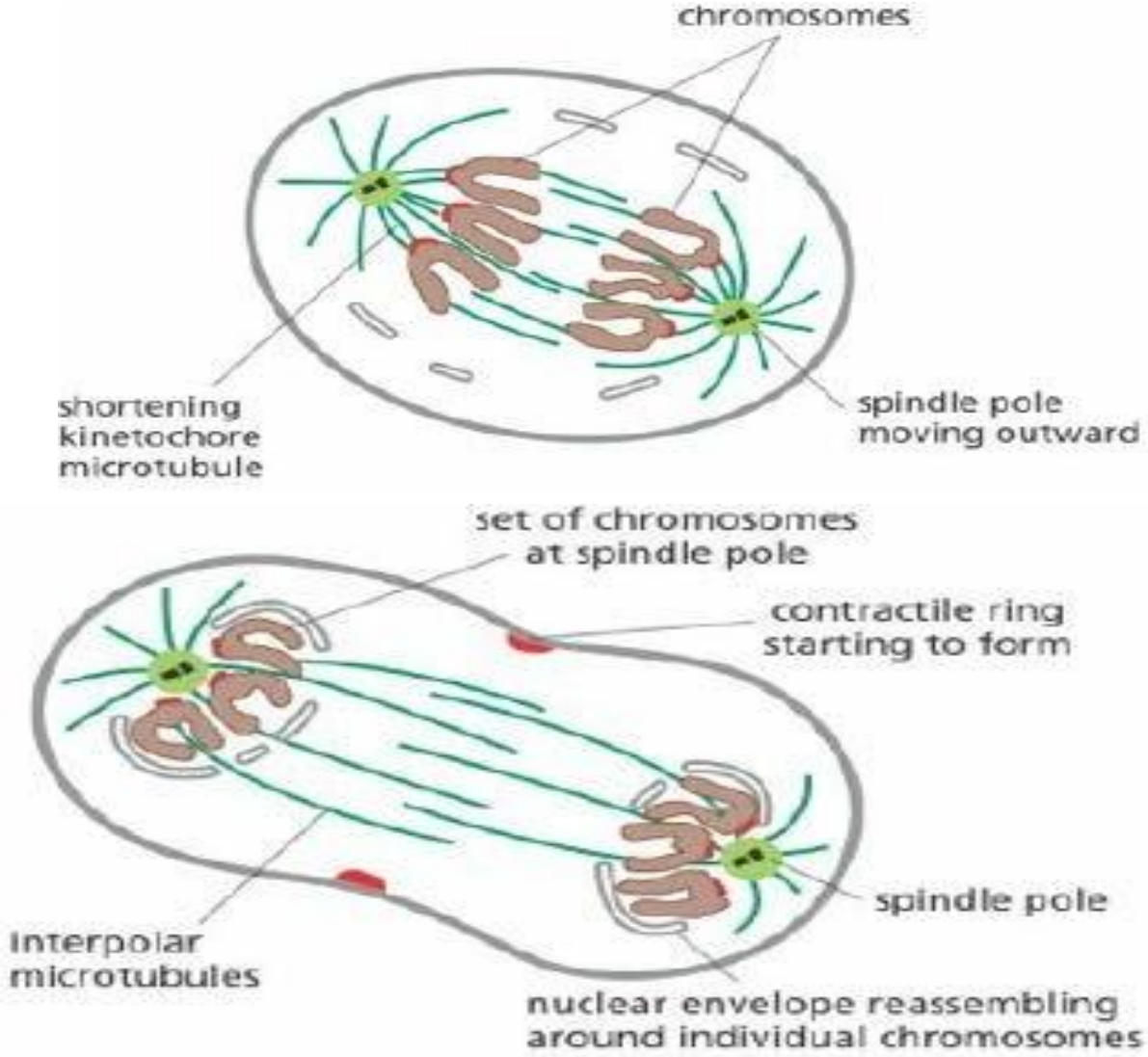
- Chromatids aligned at the **equator** of the spindle, halfway between the two poles.
- The aligned chromatids form the metaphase plate.
- **Karyotype** analyses used to determine the overall chromosome composition and structure most often require cells in metaphase.



(LO.1)

4. Anaphase

- The mitotic poles are pushed further apart as a result of polar microtubules elongation.
- Each centromere splits in two and paired kinetochores also separate.
- Sister chromatids migrate toward the opposite poles of the spindle.



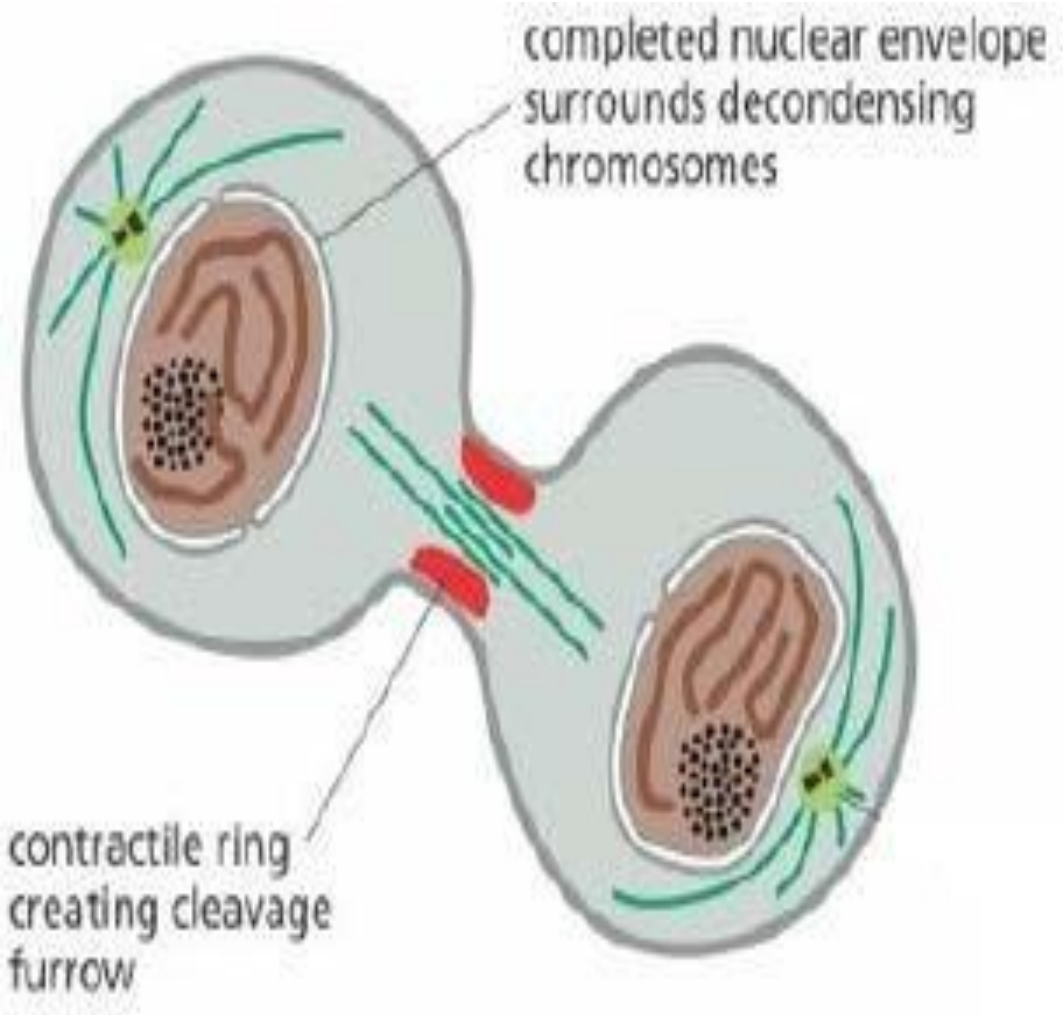
5. Telophase

- Characterized by kinetochore microtubule disassembly and mitotic spindle.
- Nuclear envelopes form around each of the two nuclei containing the chromatids.
- The chromatids decondense into dispersed chromatin and nucleoli reform in the daughter nuclei.

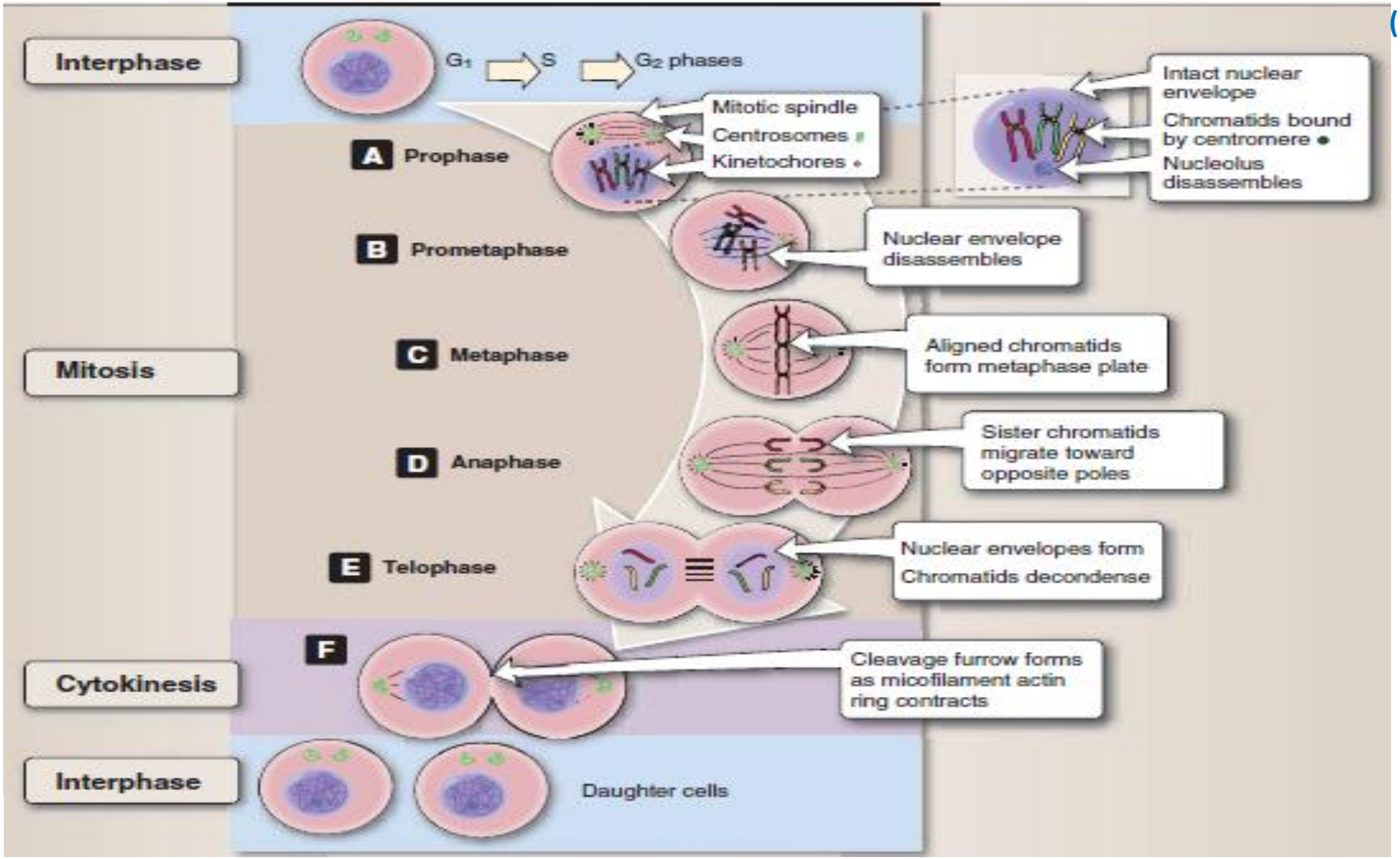
(LO.1)

Cytokinesis

The cytoplasm is divided in two by a contractile ring of actin and myosin filaments. Contraction of this actin-based structure results in the formation of a **cleavage furrow** that is seen beginning in anaphase. The furrow deepens until opposing edges meet and create two daughters, each with one nucleus.



(LO.1)


Interphase
G1
S
G2 phases
Mitotic spindle
Centrosomes
Kinetochores
Intact nuclear envelope
Chromatids bound by centromere
Nucleolus disassembles
A Prophase
B Prometaphase
Nuclear envelope disassembles
Mitosis
C Metaphase
Aligned chromatids form metaphase plate
D Anaphase
Sister chromatids migrate toward opposite poles
E Telophase
Nuclear envelopes form
Chromatids decondense
F
Cytokinesis
Cleavage furrow forms as micofilament actin ring contracts
Interphase
Daughter cells

Describe the process and role of meiosis. (LO.2)

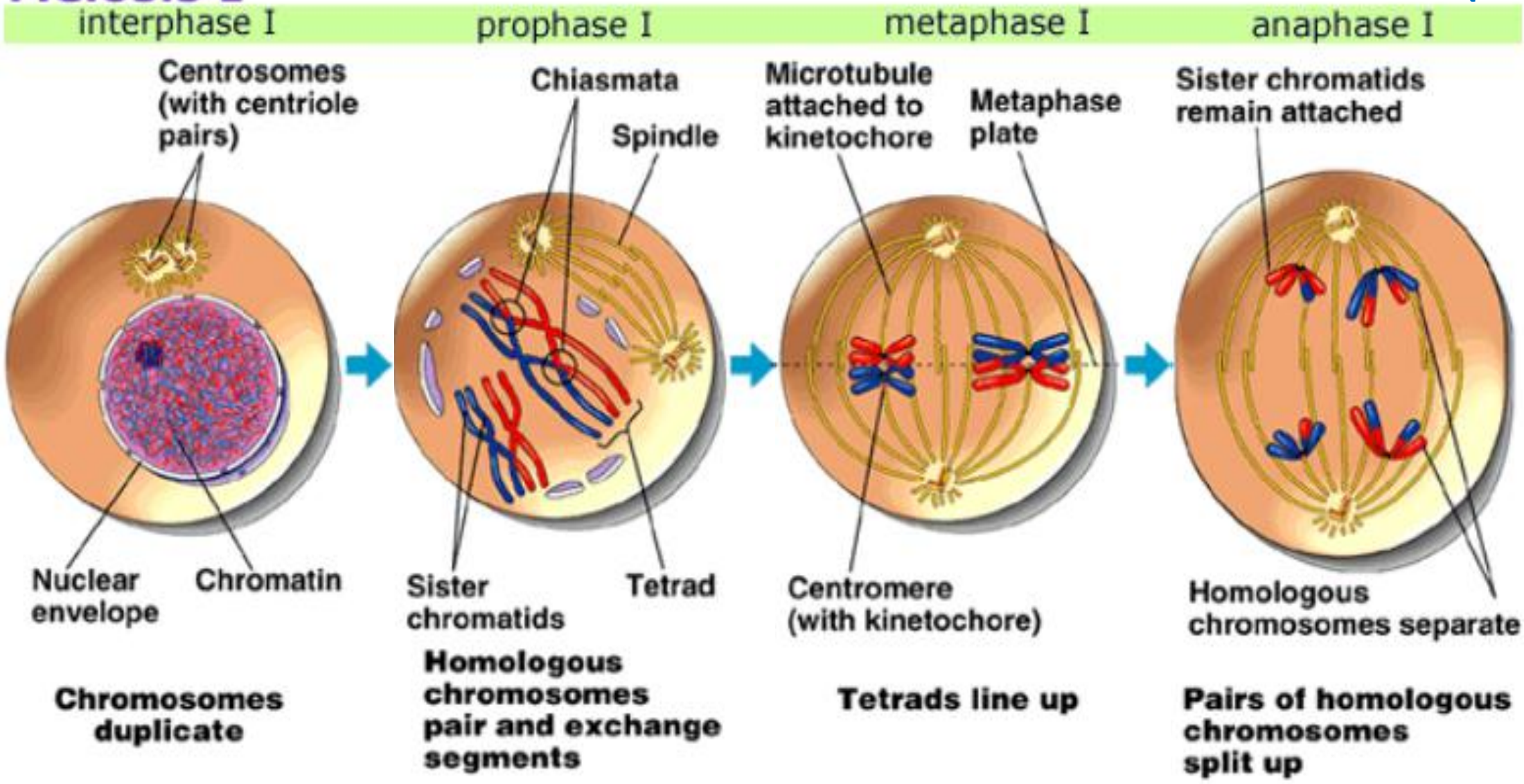
Meiosis is a relatively long-drawn-out procedure, taking very much longer than any mitosis. As gametes are haploid, genetic information must be halved during **spermatogenesis** and **oogenesis** **(which will be covered in detail in reproductive module).**

The main consequences of meiosis are that it **maintains** a constant number of chromosomes from generation to generation and generates **genetic diversity**, which is achieved in two ways: **independent assortment** of chromosomes (during meiosis I) and **crossing-over**.

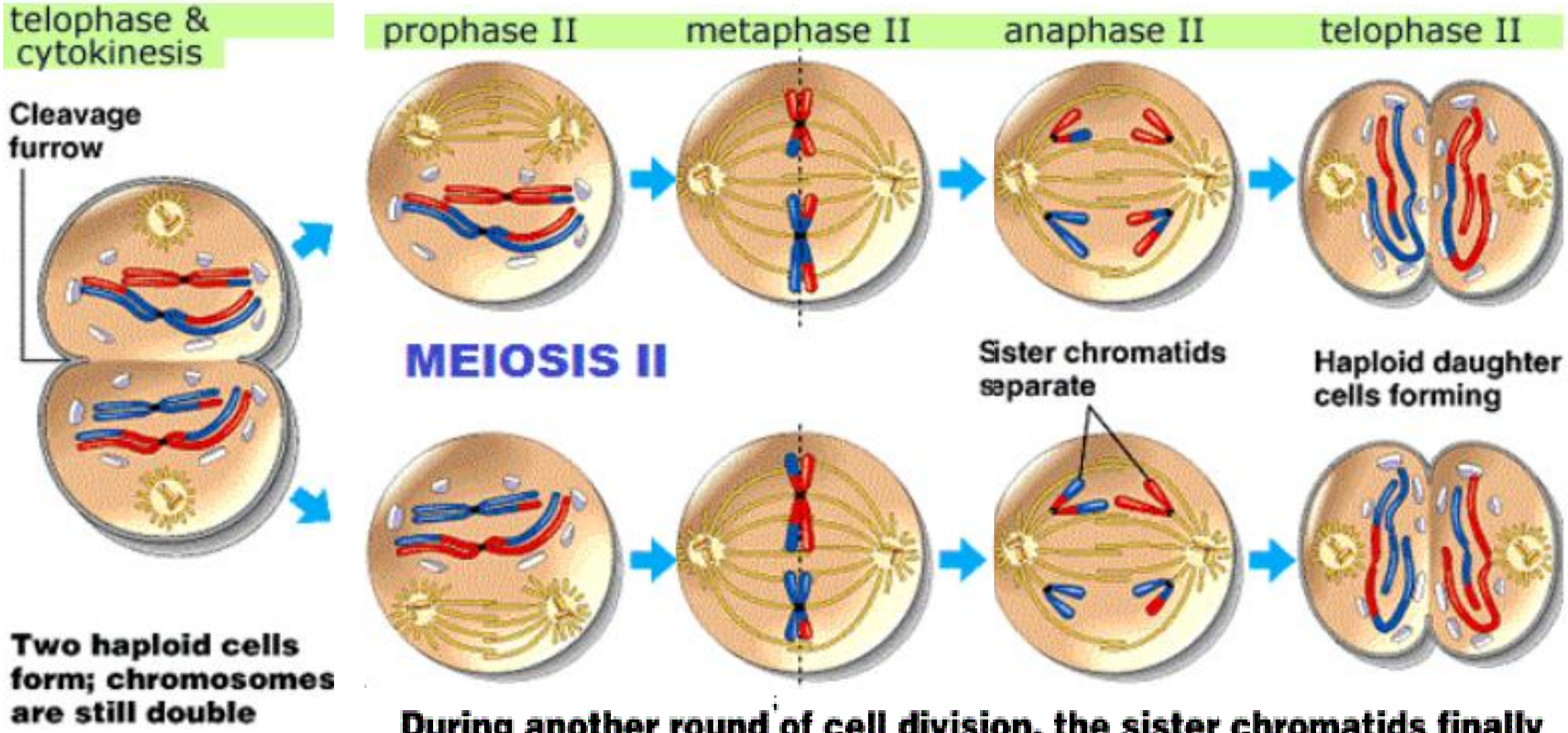
- During **meiosis I** the homologous chromosomes of each chromosome pair are divided.
- During **meiosis II** the chromatids of each chromosome are divided.

Meiosis I

(LO.2)



(LO.2)



During another round of cell division, the sister chromatids finally separate; four haploid daughter cells result, containing single chromosomes

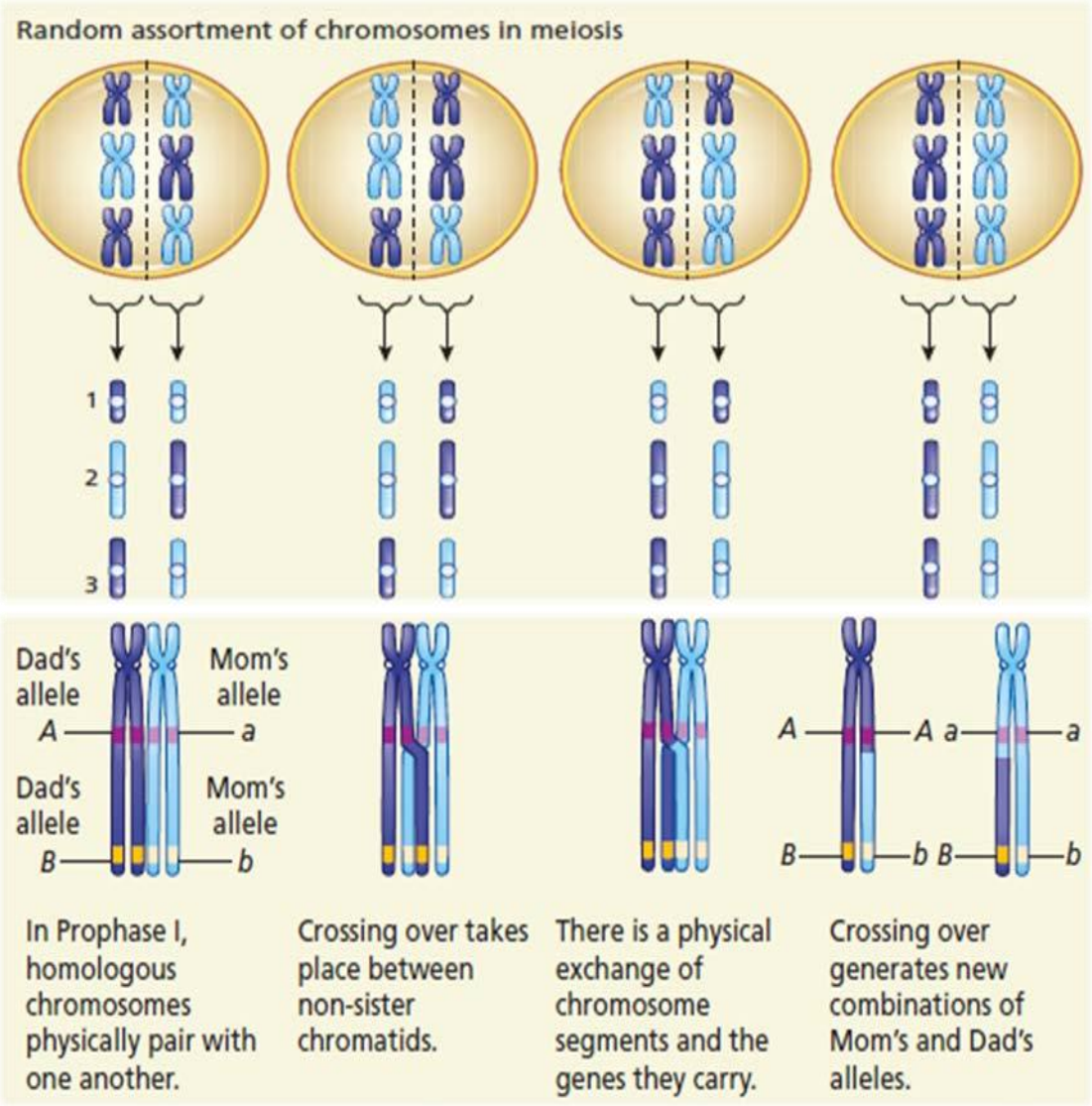
(LO.2)

Crossing over

The pairing of the maternal and paternal chromosomes is accompanied by recombination, a process in which an exchange of DNA occurs between two identical or very similar nucleotide sequences.

It occurs during the prophase of the first meiotic division. This recombination typically results in a physical swapping of homologous segments from the maternal and paternal chromosomes, an event known as crossing-over.

Crossing-over multiplies the already huge number of different gamete types produced by independent assortment.



(LO.2)

Mitosis

One round of replication and **one** round of division.

Two daughter cells per cycle.

Daughter cell genetically **identical**.

Chromosome number of daughter cells

Same as that of parent cell (2n).

Occurs in **somatic** cells.

Occurs throughout life cycle.

Used for growth, repair, and sexual reproduction.

Meiosis

One round of replication and **two** rounds of division (meiosis I and meiosis II).

Four daughter cells per cycle.

Daughter cell genetically **different**.

Chromosome number of daughter cells **half**

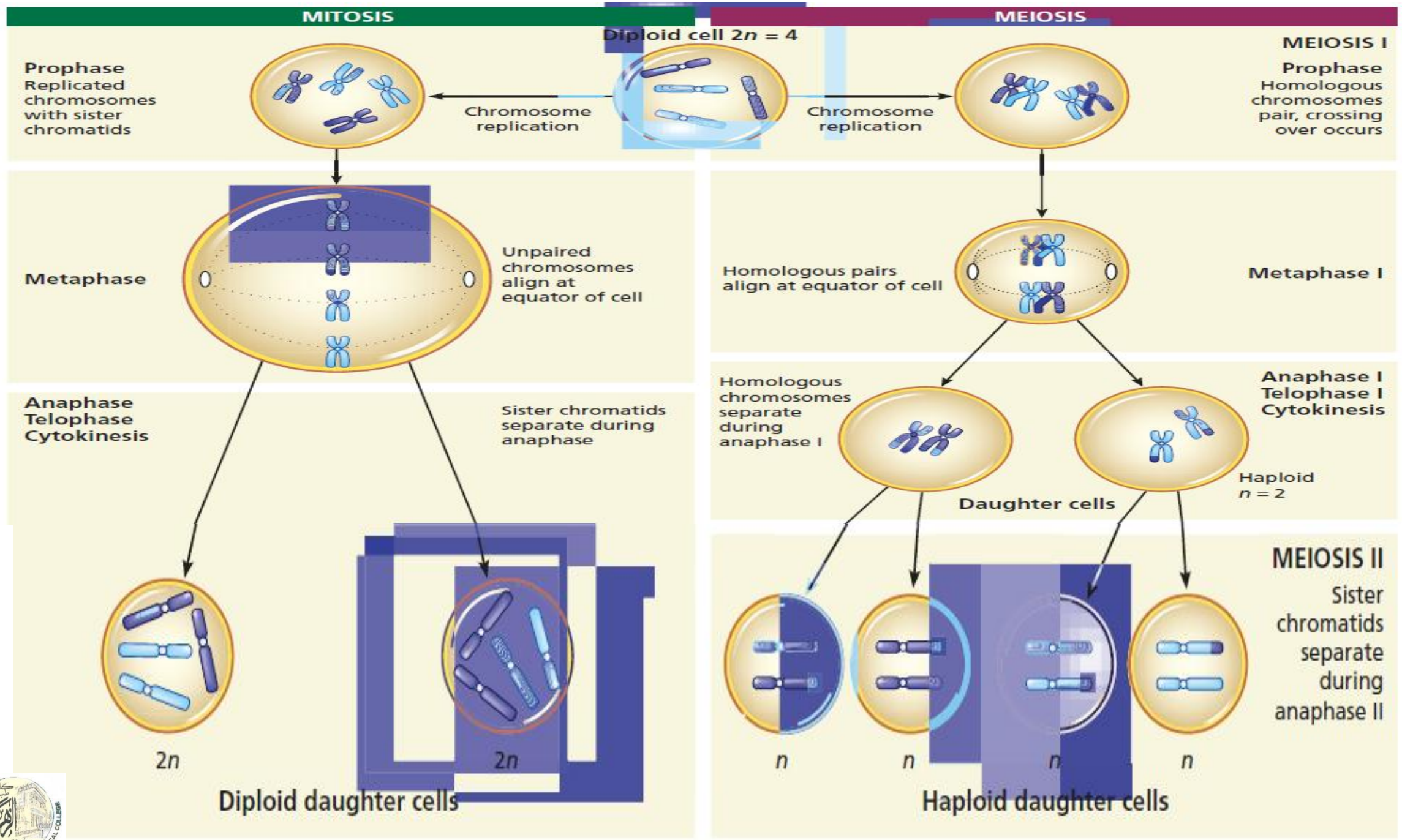
that of parent (1n).

Occurs in **germline** cells.

In humans, completes after sexual maturity

Used for sexual reproduction, producing new combinations.

(LO.2)


MITOSIS
MEIOSIS
Diploid cell 2n = 4
Prophase
Replicated chromosomes with sister chromatids
Chromosome replication
Chromosome replication
MEIOSIS I
Prophase
Homologous chromosomes pair, crossing over occurs
Metaphase
Unpaired chromosomes align at equator of cell
Homologous pairs align at equator of cell
Metaphase I
Anaphase
Telophase
Cytokinesis
Sister chromatids separate during anaphase
Homologous chromosomes separate during anaphase I
Anaphase I
Telophase I
Cytokinesis
Haploid
n = 2
Daughter cells
MEIOSIS II
Sister chromatids separate during anaphase II
2n
2n
n
n
n
n
Diploid daughter cells
Haploid daughter cells

Distinguish clearly between genotype and phenotype.

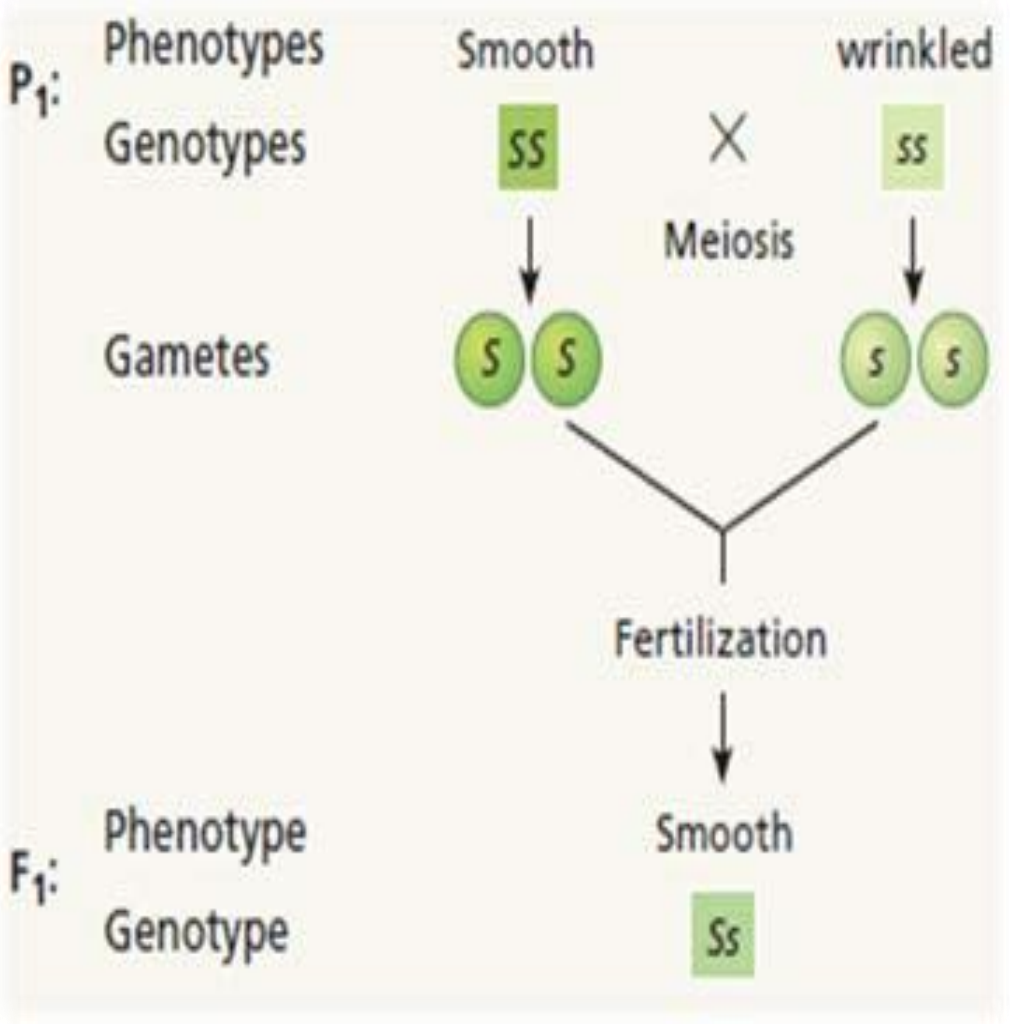
(LO.3)

Genotype: the genetic make-up of an individual (either as a whole or for one specific genetic locus). It depends upon the hereditary information that was given to an individual by their parents.

- ✓ It is fixed at the moment of fertilization and, barring mutation, is unchanging.

Phenotype: all observable characteristics of an individual or the observable trait as a result of the genetic make-up of one (or more) specific genetic locus (loci). It depends upon genotype and the influence of the environment.

- ✓ It is variable and undergoes continuous change throughout the life of the organism.

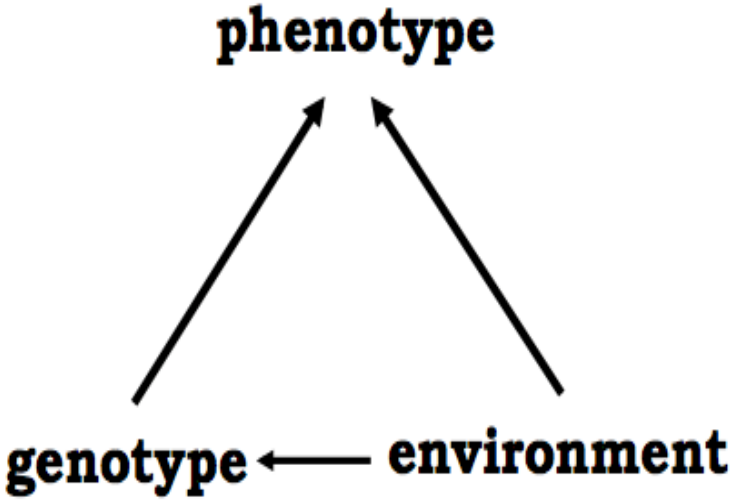


(LO.4)

Explain how environmental factors have an influence on both phenotype and genotype:

Multifactorial traits:

Traits that result from the **interaction** of one or more **environmental** factors and two or more **genes**.



Multifactorial traits have several important characteristics:

- Several genes control traits.
- Each gene controlling the trait **contributes** a small amount to the phenotype.
- Environmental factors **interact** with the genotype to produce the phenotype.

Human height, for example, is a multifactorial trait; controlled by several genes, and environmental factors make significant contributions to variations in its expression.

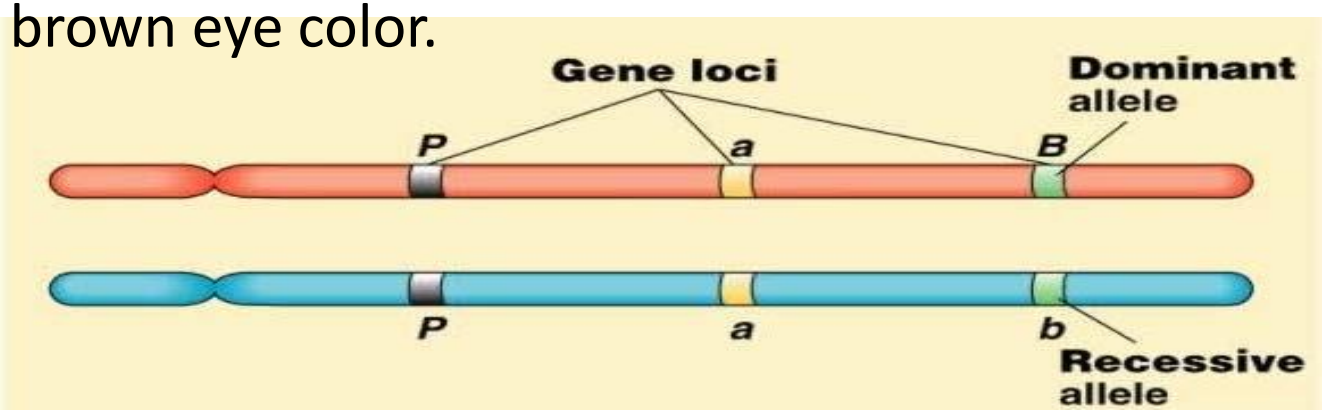
Multifactorial inheritance underlies many human traits and diseases such as obesity, diabetes, hypertension & heart-related problems, arthritis or even breast cancer.

Distinguish clearly between gene and allele

(LO.5)

Gene: The fundamental unit of heredity and the basic **structural** and **functional** unit of genetics. It is a length of DNA on a chromosome that contains the code for a protein (or RNA).

Allele: an alternative form of a gene; each individual has **two** alleles for every gene, which can either be the same or different. For example, a chromosome may carry a gene for eye color. One copy of the chromosome may carry an allele for blue eyes, while the other carries an allele for brown eye color.



(LO.5)

The two alleles of homologous chromosomes can be divided into:

- ✓ **Homozygous**: Both alleles of a gene are the same
- ✓ **Heterozygous**: Two different alleles of a gene
- ✓ **Hemizygous**: Only one allele of a gene represent in the genes on X chromosome in male

Locus: a specific position on a chromosome.