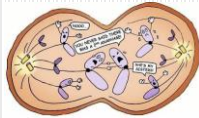


## Chromosomes Mitosis and Meiosis

Class 2  
SLE254 Genetics and Genomics  
Chapter 2 Concepts of Genetics (12<sup>th</sup> Ed)  
Pages 50-71 Ch2



## Terminology

• **Locus** –

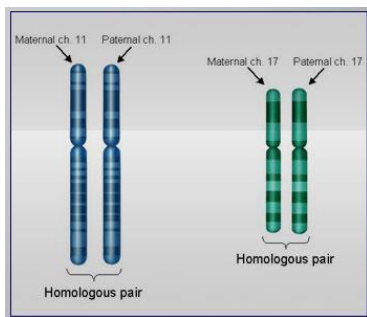
• **Allele** –

• **Genotype** –

• **Phenotype** –

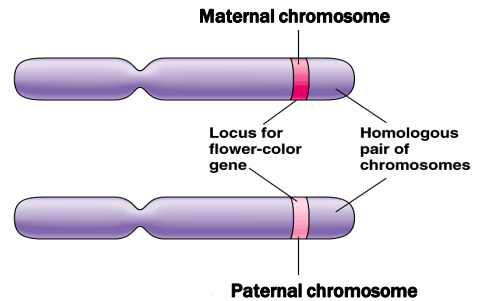


## Homologous Chromosomes



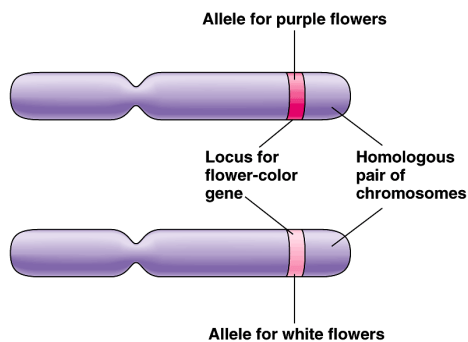
During meiosis homologs pair together

**Locus** – position of allele on a chromosome



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**Allele** – alternative DNA sequence at the same locus

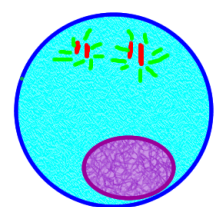


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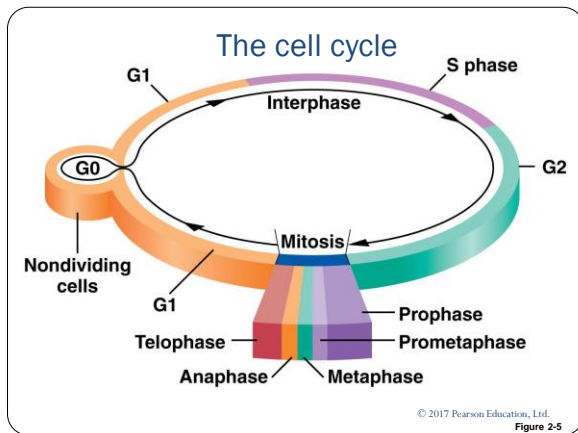
## Cell division and the cell cycle



Interphase



[http://www2.amsosol.edu.pe/class11/\\_11pengl/7th/science/images/cells.gif](http://www2.amsosol.edu.pe/class11/_11pengl/7th/science/images/cells.gif)  
[http://24-media.tumblr.com/tumblr\\_m9807gMVL1qbw86a1\\_400.jpg](http://24-media.tumblr.com/tumblr_m9807gMVL1qbw86a1_400.jpg)



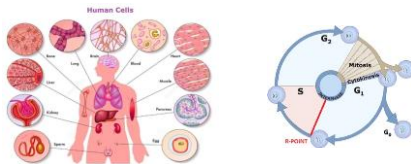
## Cell cycle 1. Interphase

- **Cell cycle**
  - Composed of **interphase** and **mitosis**
- **Interphase** includes
  - S phase: DNA is synthesized
  - Two gap phases (G1 and G2) (Figure 2-5)
- G0: Point in G1 phase where cells are nondividing, but a metabolically active state

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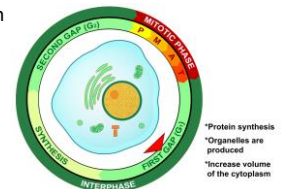
## Interphase

- **Interphase**: the period of **growth** between cell division
- Up to 90% of a cell's time in the normal cellular cycle may be spent in interphase
- The cell may appear to be dormant, however, biochemical activity is high during interphase



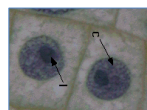
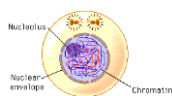
## Stages of Interphase: Gap 1

- **G<sub>1</sub> phase** – The period **prior to the synthesis of DNA**  
G = Gap 1 = first Gap
- For many cells, this phase is the major period of cell growth during its lifespan



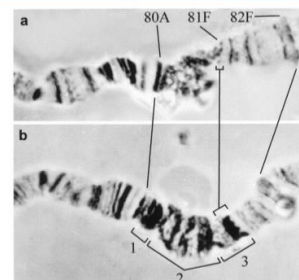
## Chromosomes in Interphase

- The DNA in a G<sub>1</sub> diploid eukaryotic cell is **2n**, meaning there are **two sets of chromosomes** present in the cell
- The genetic material exists in loose form **chromatin**
  - The structure of chromatin during interphase is optimised to allow easy access of transcription and DNA repair factors to the DNA
    - Not tightly coiled



## Interphase Chromatin

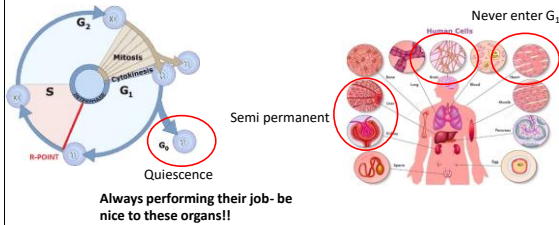
- Different levels of packing on various regions of the same chromosome
- **Euchromatin**: loosely packed region on chromatin, active transcription
- **Heterochromatin**: densely packed region on chromatin, inactivated



Eu = true; Hetero = different

## Stages of Interphase: $G_0$ phase

- A cell may **pause** in the  $G_1$  phase before entering the S phase and enter a state of dormancy called the  $G_0$  phase



## Stages of Interphase: $G_0$ phase

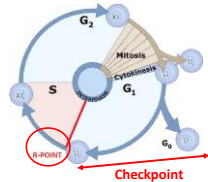
- Epithelial cells divide throughout life- rarely enter  $G_0$

Epithelia line tissues, organs: skin

Epithelium	Location	Characteristics
Simple cuboidal	Renal tubules, thyroid follicles	One layer of cells, cube-shaped
Simple columnar	Intestinal villi, liver	One layer of cells, taller than wide
Stratified squamous	Epidermis, oral cavity	Multiple layers, flat cells on top
Transitional	Urinary bladder	Multiple layers, can stretch
Glandular	Salivary glands	Secretory cells, often with ducts

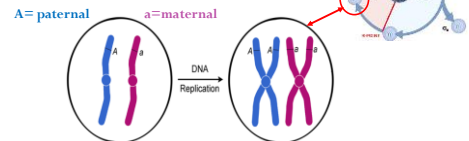
## Stages of Interphase: restriction (R) point

- The **restriction (R) point** is present at the end of  $G_1$  phase
- Depending on levels of nutrients, energy and external factors, cells must decide to enter the cell cycle or move into  $G_0$  phase



## Stages of Interphase: S phase

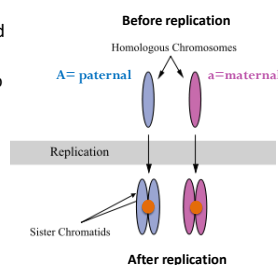
- S phase** – The period during which DNA is synthesised (**replicated**)
- The **S** represents **synthesis**
- Create exactly two identical semi-conserved chromosomes
- Detect and fix DNA damage



Doubling of DNA but the cell is still  $2n$ , meaning there are still two sets of chromosomes

## Structure of the chromosome

- Chromatid**
  - One strand of a duplicated chromosome
  - Joined by a centromere to its sister chromatid
- Sister chromatids**
  - Two chromatids joined by a common centromere
  - Each carries identical genetic information



## Structure of the chromosome

- Centromere**
  - Heterochromatic region of a chromosome to which microtubule fibres attach during cell division
  - Centromere location gives a chromosome its characteristic shape

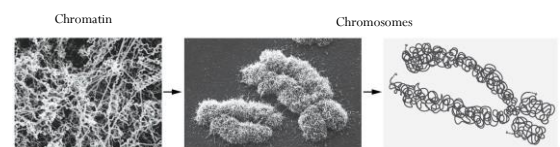
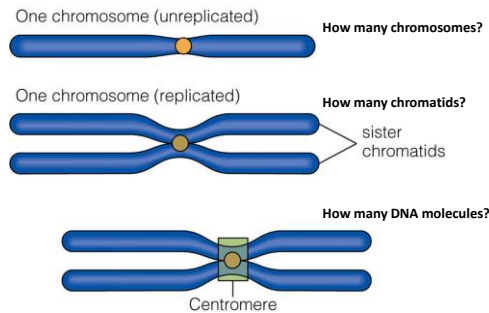


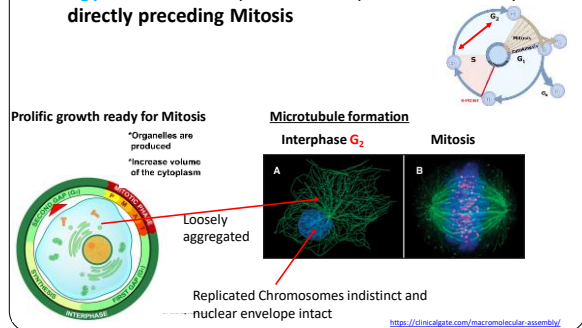
Figure 2.14

## Structure of the chromosome



## Stages of Interphase: $G_2$ phase

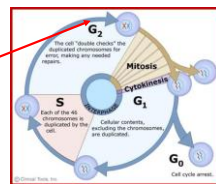
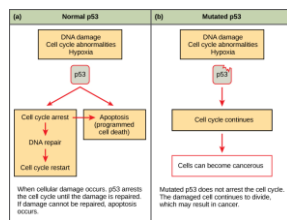
- $G_2$  phase – final subphase of Interphase in the cell cycle directly preceding Mitosis



## Stages of Interphase: $G_2$ restriction R point

- Final checkpoint before Mitosis

\*Serves to prevent the cell from entering mitosis with genomic DNA damage



p53 protein is the cell division 'police'

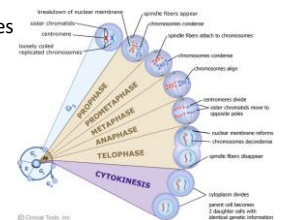
<https://openstax.org/c/biology/chapter/15-3-cancer-and-the-cell-cycle/>

## Cell cycle 2. Mitosis

- One diploid cell** divides to form **two diploid cells**
- Each cell has an exact copy of the genetic information contained in the parent cell

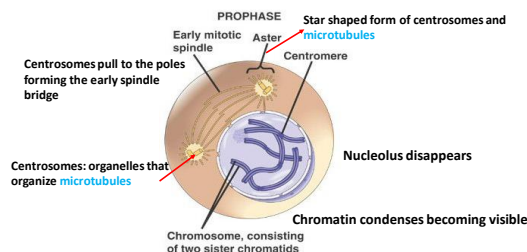
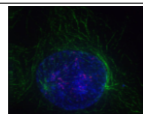
- Mitosis is divided into four stages

- Prophase
- Metaphase
- Anaphase
- Telophase



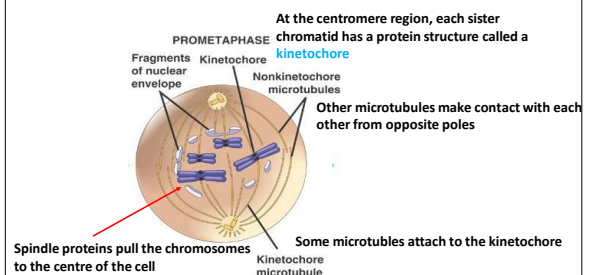
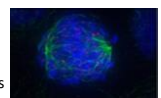
## Mitosis – Prophase

- Changes in the nucleus
- Changes in the cytoplasm



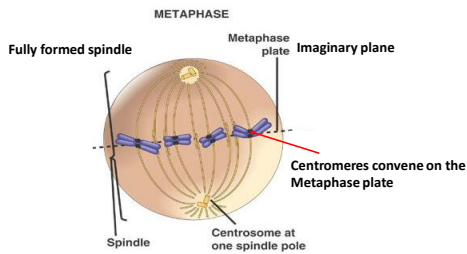
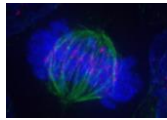
## Mitosis – Prometaphase

- Nuclear membrane breaks apart
- Spindle fibers attach to the condensed chromosomes
- Begin pulling the chromosomes to the center



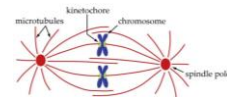
## Mitosis – Metaphase

1. Highly condensed chromosomes align in the middle of the cell



## Mitosis – Spindle checkpoint

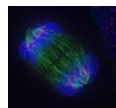
- Monitors the interaction between improperly connected kinetochores and spindle microtubules, and is maintained until kinetochores are properly attached to the spindle
- Monitors **kinetochore tension**:
  - When kinetochores are properly attached to **opposite spindle poles**, forces in the mitotic spindle generate tension



- **If activated**, the spindle checkpoint blocks anaphase entry
- **Deactivated**: correct orientation of sister chromatids

## Mitosis – Anaphase

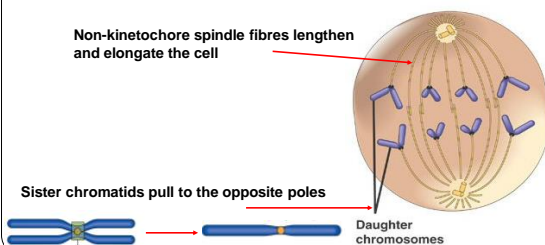
1. Chromosomes move to **opposite poles of the cell**
2. initiated by a protease known as **separase** which cleaves **cohesin**, a protein responsible for holding sister chromatids together



ANAPHASE

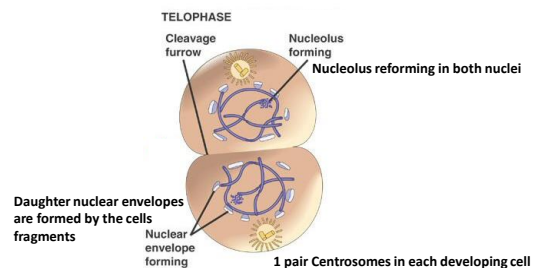
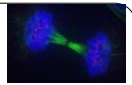
Non-kinetochore spindle fibres lengthen and elongate the cell

Sister chromatids pull to the opposite poles



## Mitosis – Telophase

1. Two **daughter nuclei** form in the cell
2. The chromosomes unwind back into **loose chromatin**



## Cytokinesis

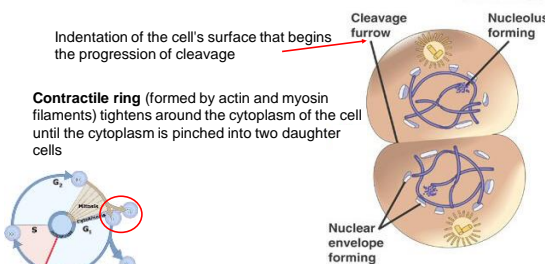
1. **Separate phase** to mitosis
2. **Cytoplasm divides** into two daughter cells



CYTOKINESIS

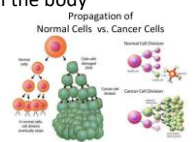
Indentation of the cell's surface that begins the progression of cleavage

**Contractile ring** (formed by actin and myosin filaments) tightens around the cytoplasm of the cell until the cytoplasm is pinched into two daughter cells



## Mitosis in growth and cell replacement

- Human somatic cells are genetically programmed to divide about 50-80 times
  - This limit allows growth to adulthood, and repairs such as wound healing
- Replicative exhausted cells undergo senescence, which is a cell cycle arrested state. They secrete proinflammatory factors to mediate their clearance from the body (macrophages)
- When it goes wrong?
  - **Cancer is a disease of the cell cycle**

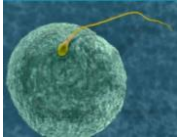


<https://www.slideshare.net/Lumidmed/Celw11/halmarks-of-cancer-sustained-proliferative-signaling>

## Meiosis: generation of genetic diversity

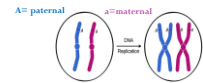
- **Meiosis:** A form of cell division that produces **four haploid cells** containing **only one copy** (paternal or maternal) of each chromosome

- Meiosis I
- Meiosis II



Humanas.tumblr.com  
<https://www.slideshare.net/catherinepatterson/cell-division-mitosis-and-meiosis-presentation>

## Terminology:



### 1. Homologous chromosomes

- Chromosomes exist in homologous pairs in diploid organisms
- **Maternal and paternal:** with identical gene loci (but often different alleles)
- **They physically pair during meiosis**

### 2. Assortment

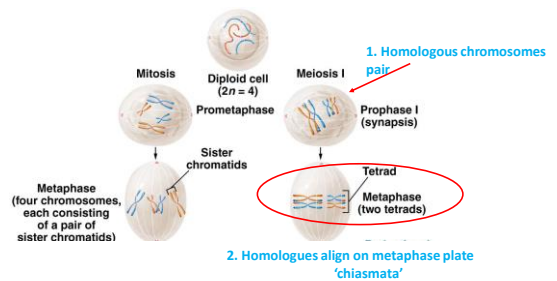
- Gametes receive random combinations of maternal and paternal chromosomes (generates genetic diversity)
- Result of **meiosis I**

## Meiosis:

### Two vital sources of genetic variation

- **Crossing over:** The exchange of chromosome segments between homologous regions (chromatids) in prophase I
- **Independent assortment** of maternal and paternal chromosomes in metaphase I

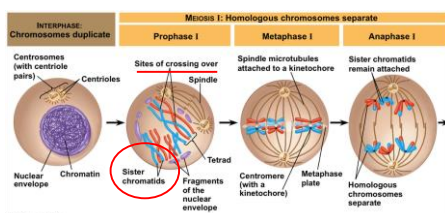
## Meiosis I. Differences between mitosis



## Meiosis I

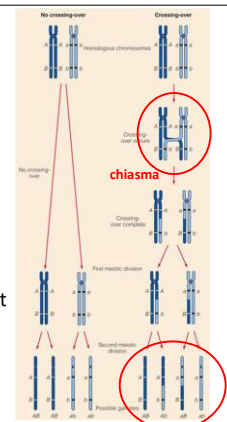
1. In meiosis I, members of a **pair of homologous chromosomes** physically associate

- **Crossing over occurs, which increases the genetic diversity in the gametes.**



## Prophase I. Crossing over

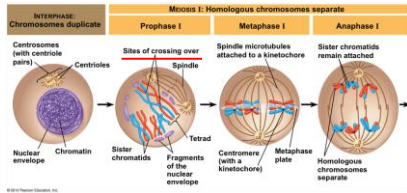
- Exchange of genetic material between homologous chromosomes that results in **recombinant** chromosomes
- Homologous regions (chromatids) break and then reconnect to the other chromosome
- Results in a new arrangement of maternal and paternal alleles on the same chromosome





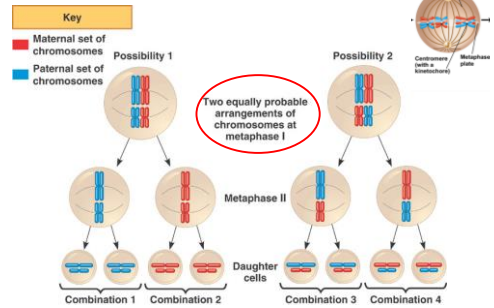
## Meiosis I. Independent assortment

Meiosis I produces cells that contain one duplicated member of each chromosome pair



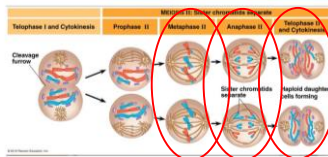
1. Homologous chromosomes line up **randomly** at **metaphase**
2. Random combinations of maternal and paternal homologues separate at **anaphase**

## Independent assortment



Does not occur (usually) in eukaryotic mitosis, only meiosis

## Meiosis II



### Metaphase II

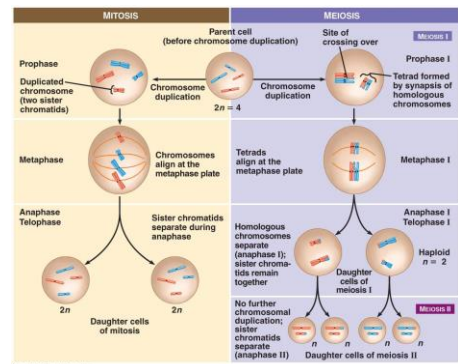
**Unpaired chromosomes** align at the cell's middle  
There is no synthesis (replication) of chromosomes before meiosis II.  
Therefore, only the remaining one replicated chromosome

### Anaphase II

**Centromeres divide** and daughter chromosomes move to opposite poles

**Meiosis produces four haploid cells (n)**  
**In humans, the haploid number is 23**

## Comparing mitosis and meiosis

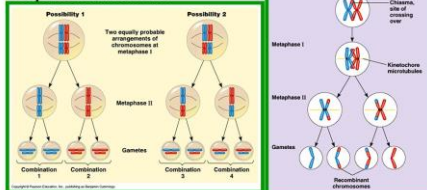


## Independent Assortment and Crossing Over Meiosis

How do we account for genetic variation?

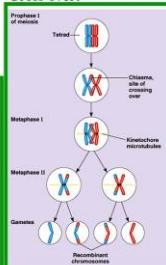
- \*Independent assortment
- \*Crossing over

### Independent Assortment:



<https://www.youtube.com/watch?v=-Zzp3mLycM>

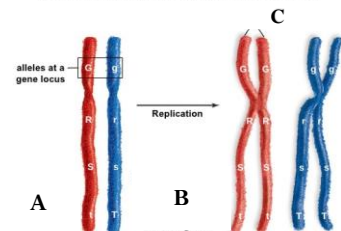
### Cross over:



## Quiz: What does A, B and C stand for?

### Homologous Chromosomes

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## Review questions

- What is the difference between chromatin and chromatids and at what stages in the cell cycle do they exist?
- At what stage of meiosis does crossing over and independent assortment occur? Do these events occur in mitosis, why?
- How many chromosomes are present at each cell cycle stage?
- How many chromatids?
- How many copies of the same gene (somatic cells chromosomes only)? (i.e. how many molecules of DNA?)
- What are the two main sources of genetic variation produced by meiosis?

## Prepare a glossary

Chromosome  
 Chromatin  
 Chromatid  
 Euchromatin  
 Heterochromatin  
 Cell cycle stages (Interphase, Mitosis, Meiosis, Synthesis, etc)  
 Centromere, spindle fibres, kinetochore  
 Independent assortment  
 Crossing over