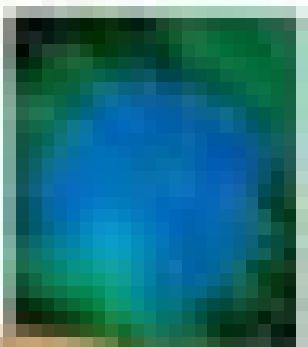




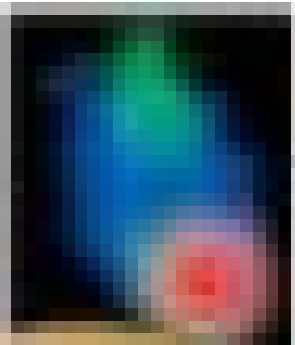
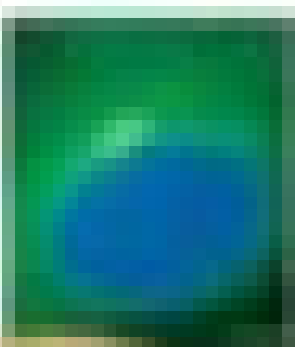
Light Intensity

Light



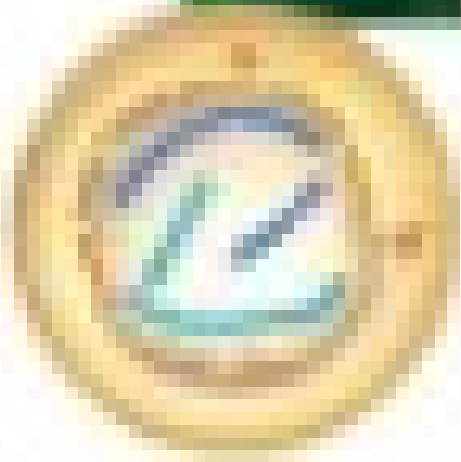
Light

Light



Light Intensity

Light Intensity



Light Intensity



Light Intensity

Light Intensity

Medical Genetics Course

Dr. Bilal Azab

The University of Jordan

School of Medicine

Department of Pathology, Microbiology and Forensic Medicine

Email: b.azab@ju.edu.jo

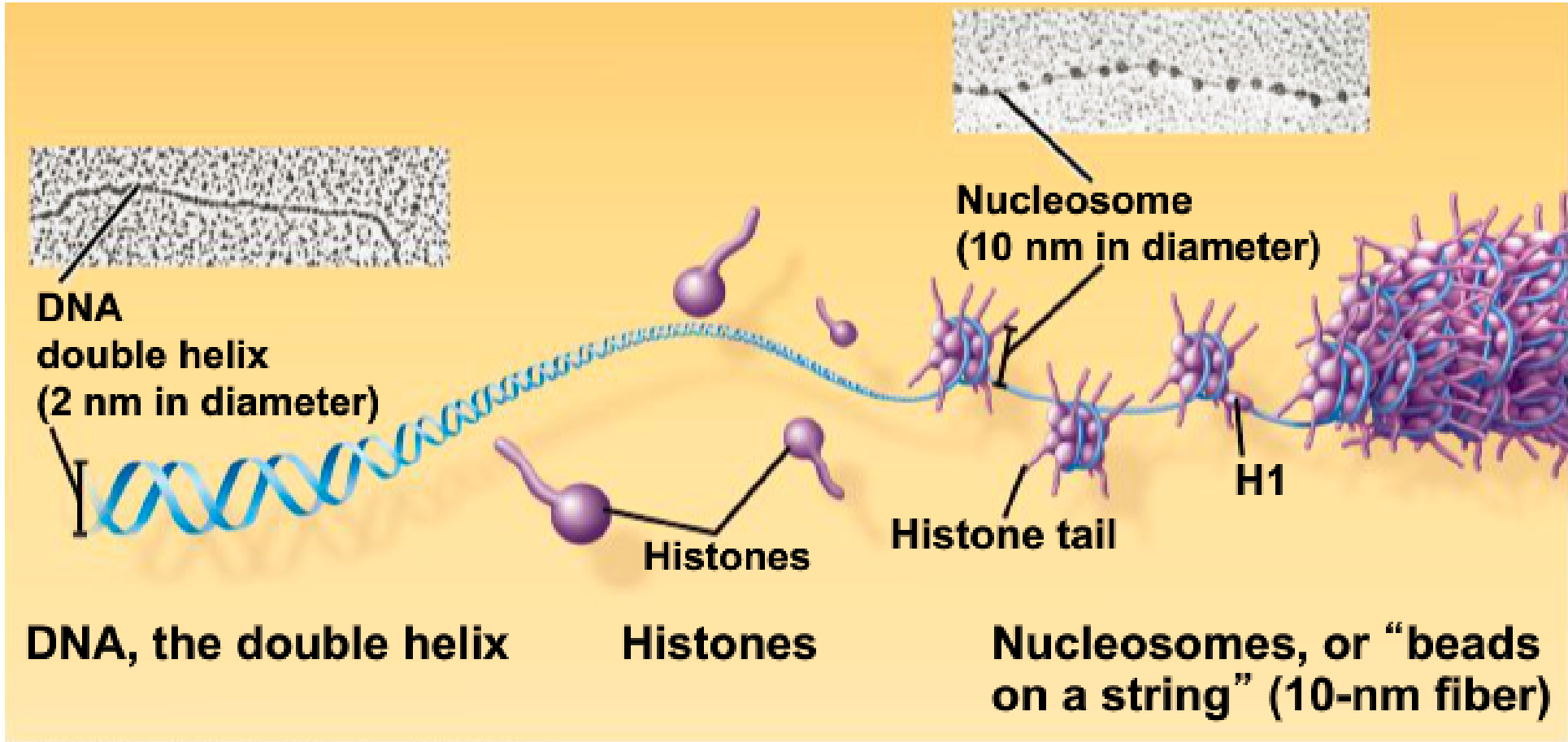
Lecture link: <https://youtu.be/IOovV7ZMn-I>

# History of Cytogenetics

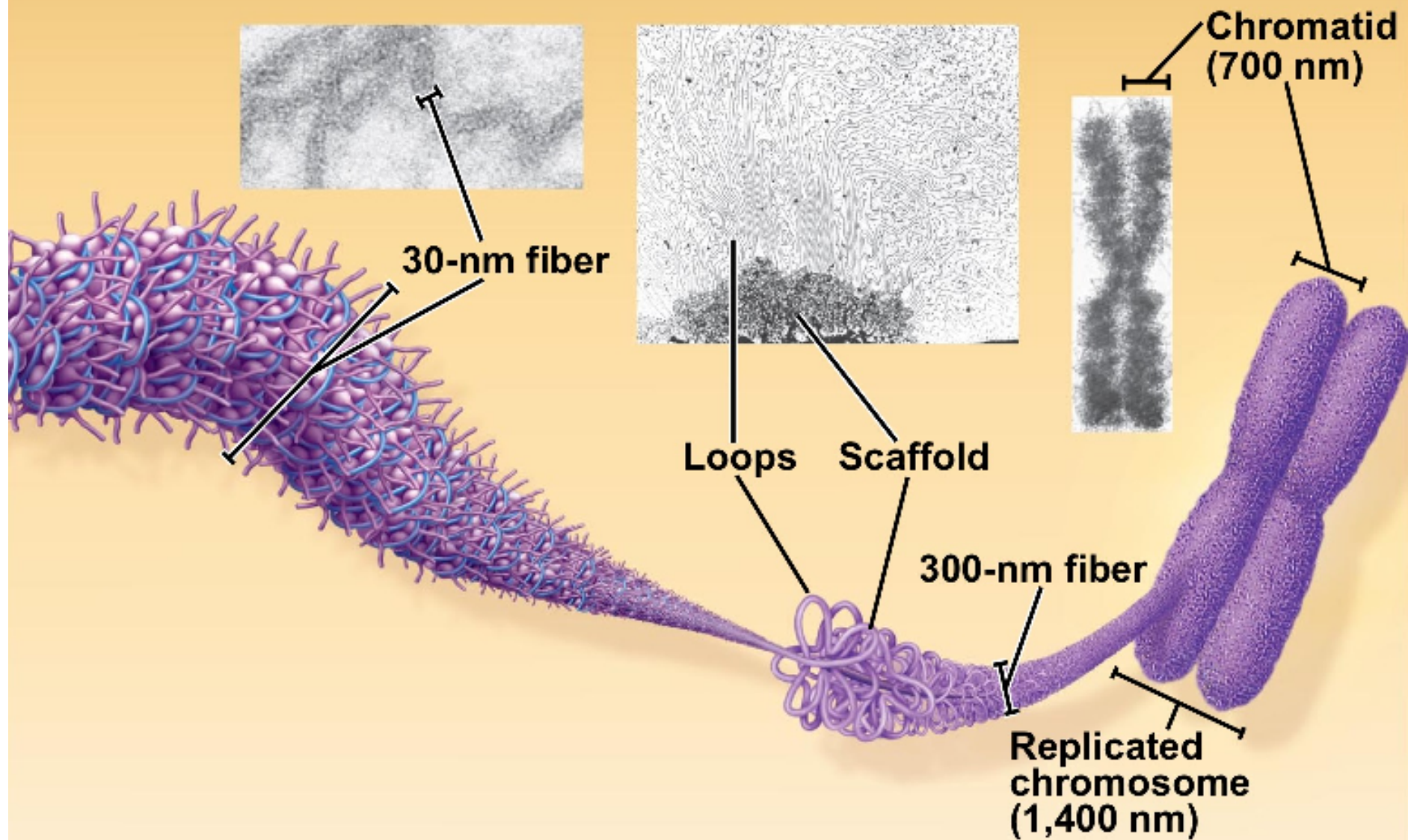
True chromosome number



*“ From their vantage through the  
microscope,  
the cytogeneticists’ view of the genome is*



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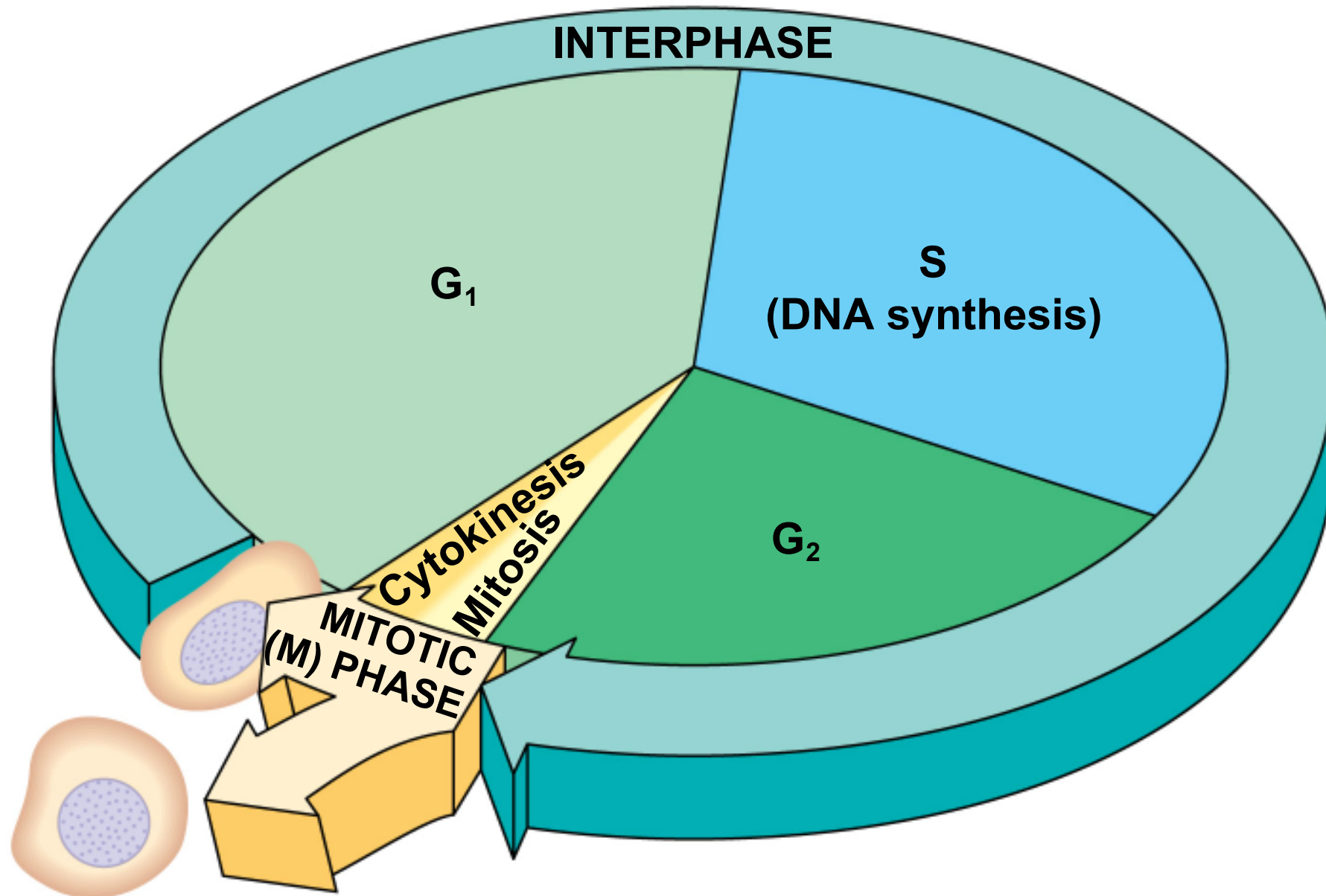


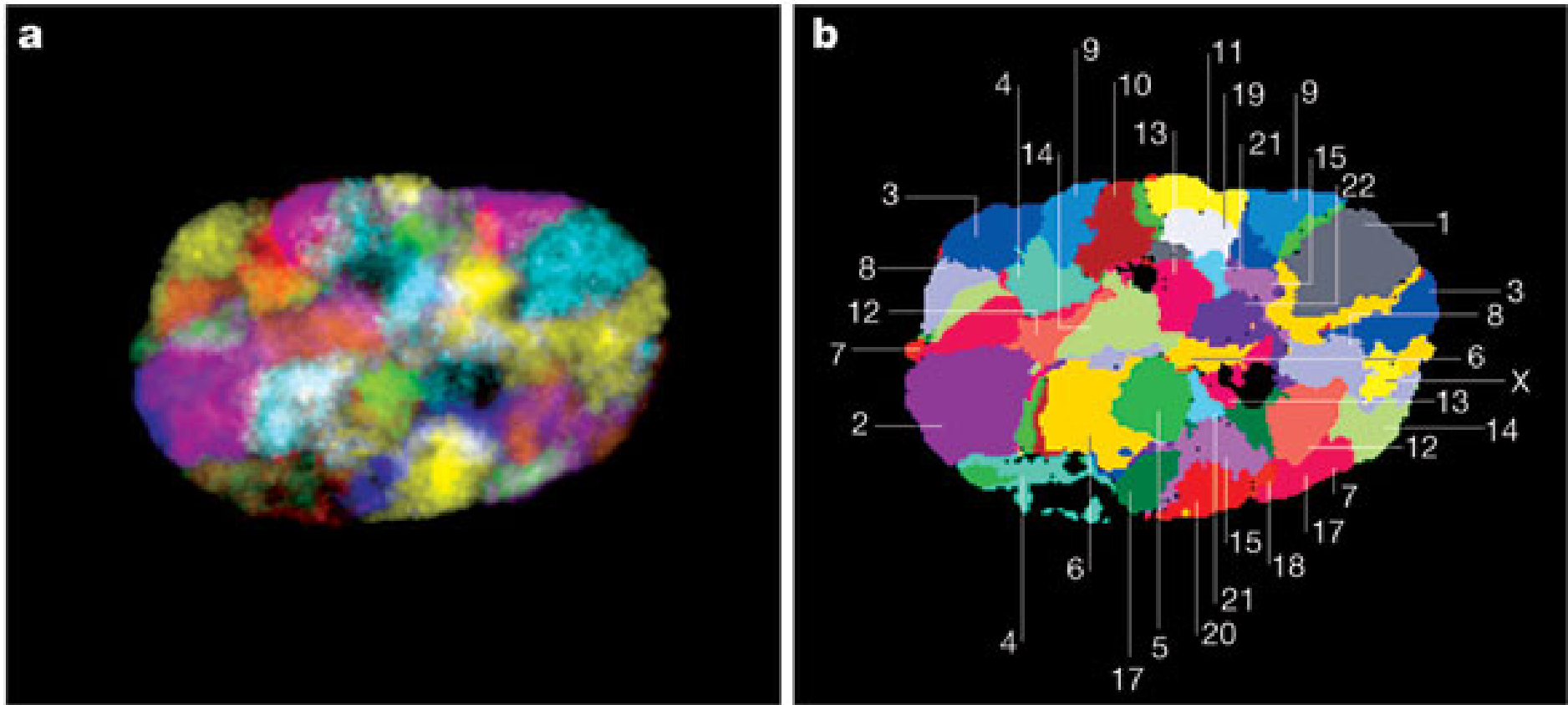
**30-nm fiber**

**Looped domains  
(300-nm fiber)**

**Metaphase  
chromosome**

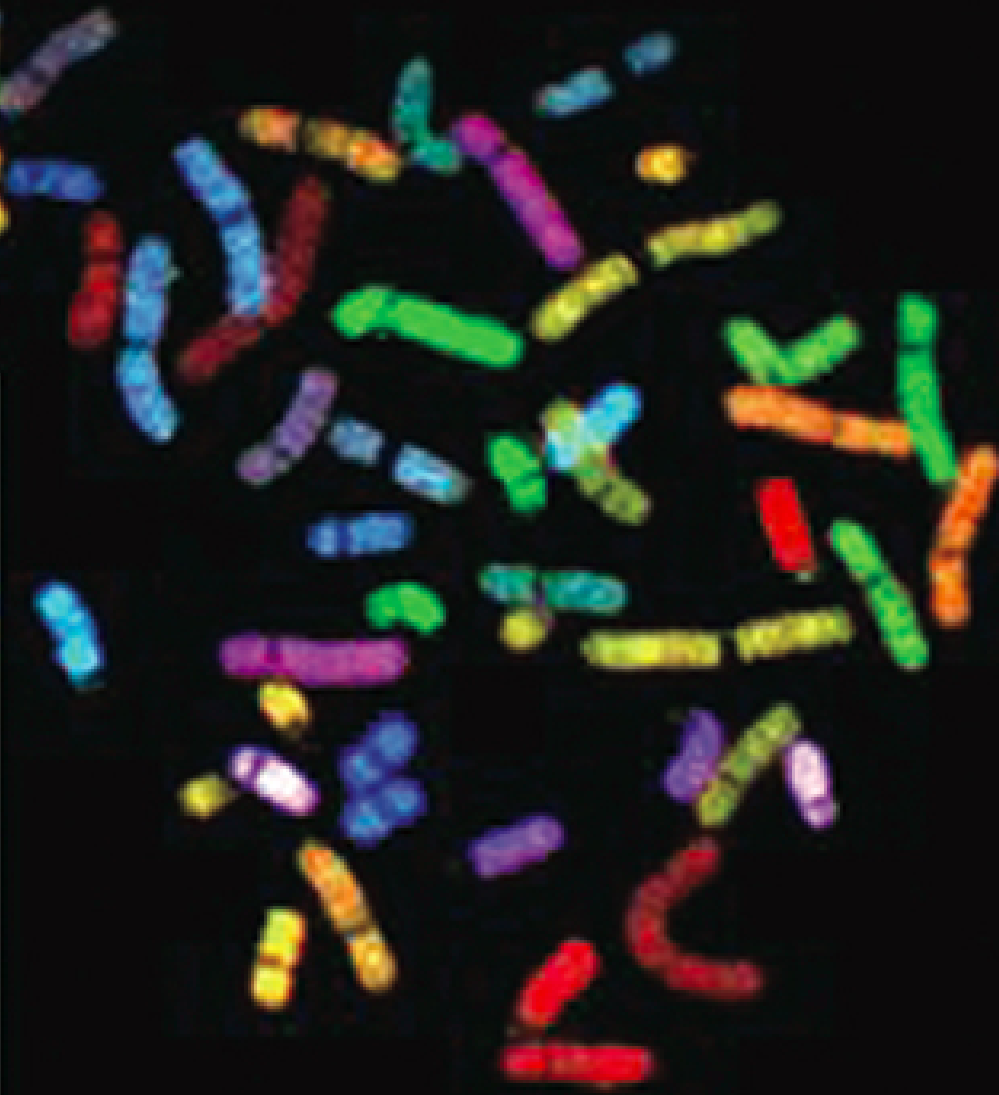
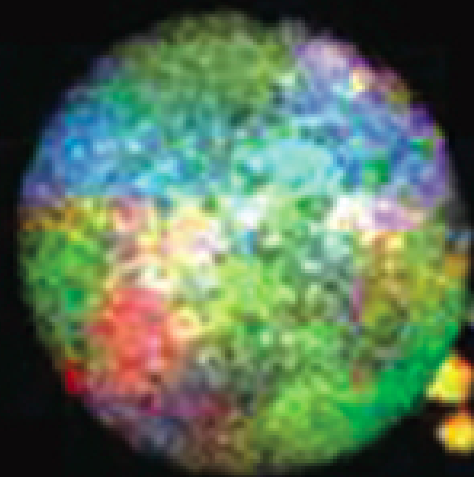
Figure 12.6





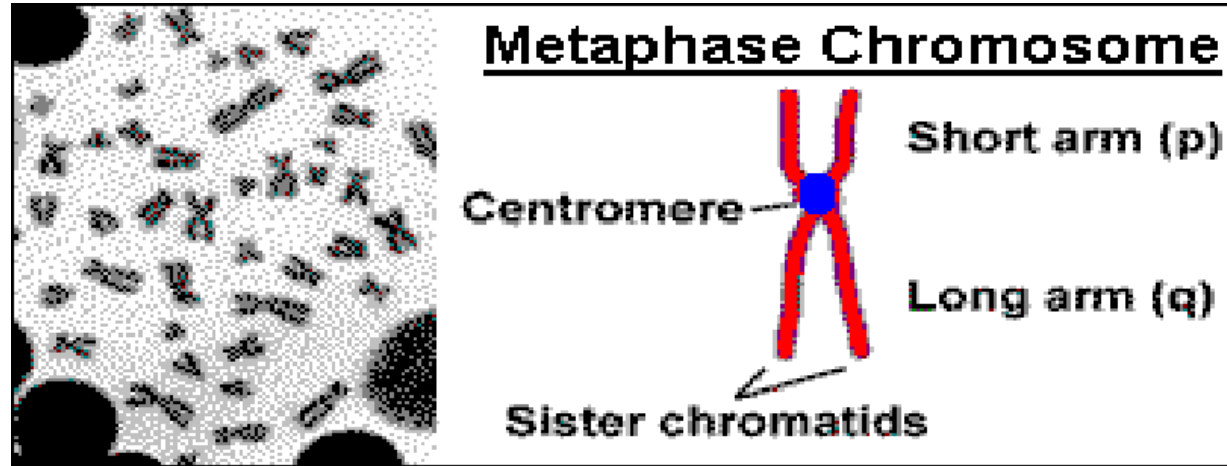
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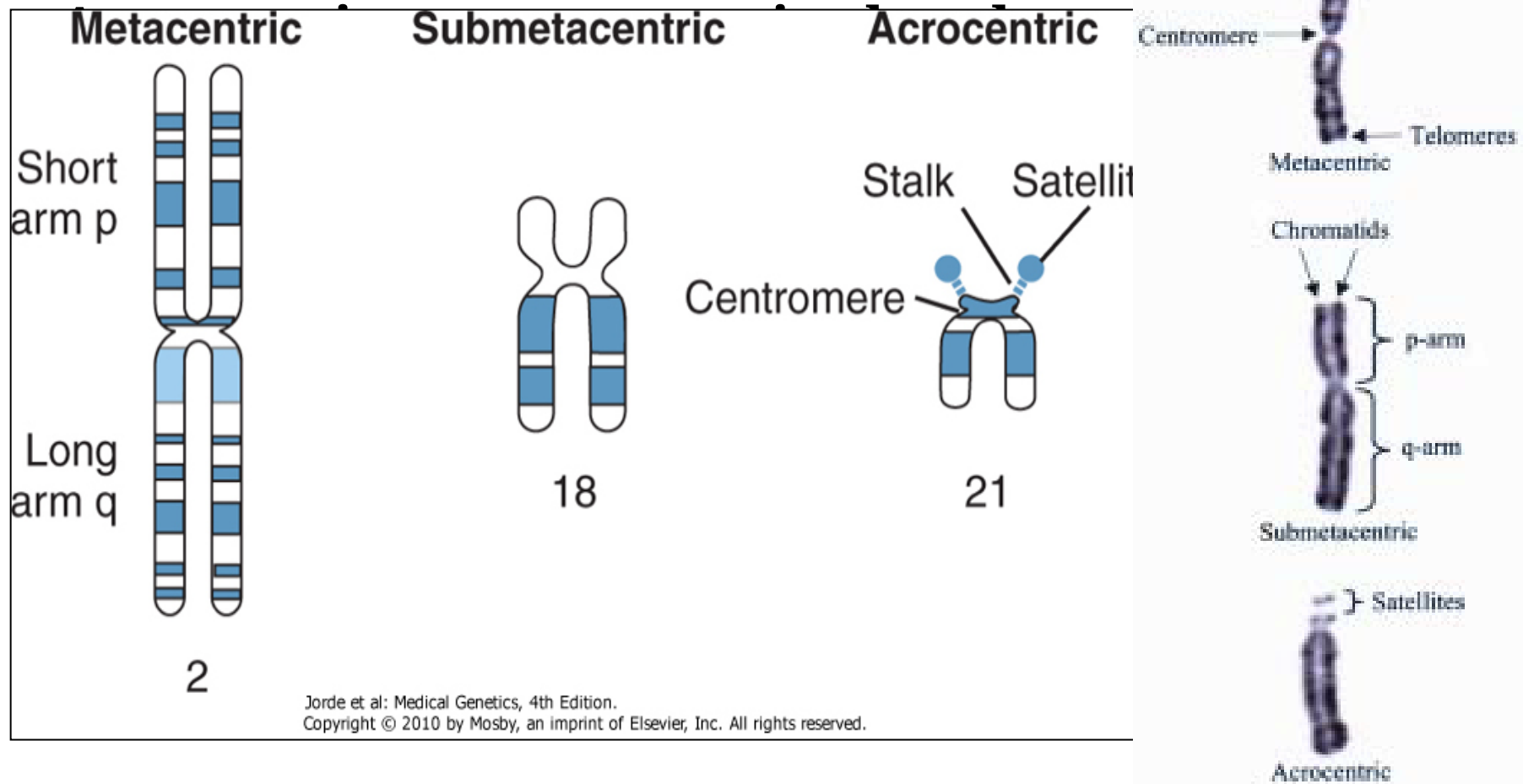
# Nomenclature of chromosomes



# Chromosome Shape

**Metacentric- centromere is located in the middle of chromosome**

**Submetacentric- centromere is displaced from the center**

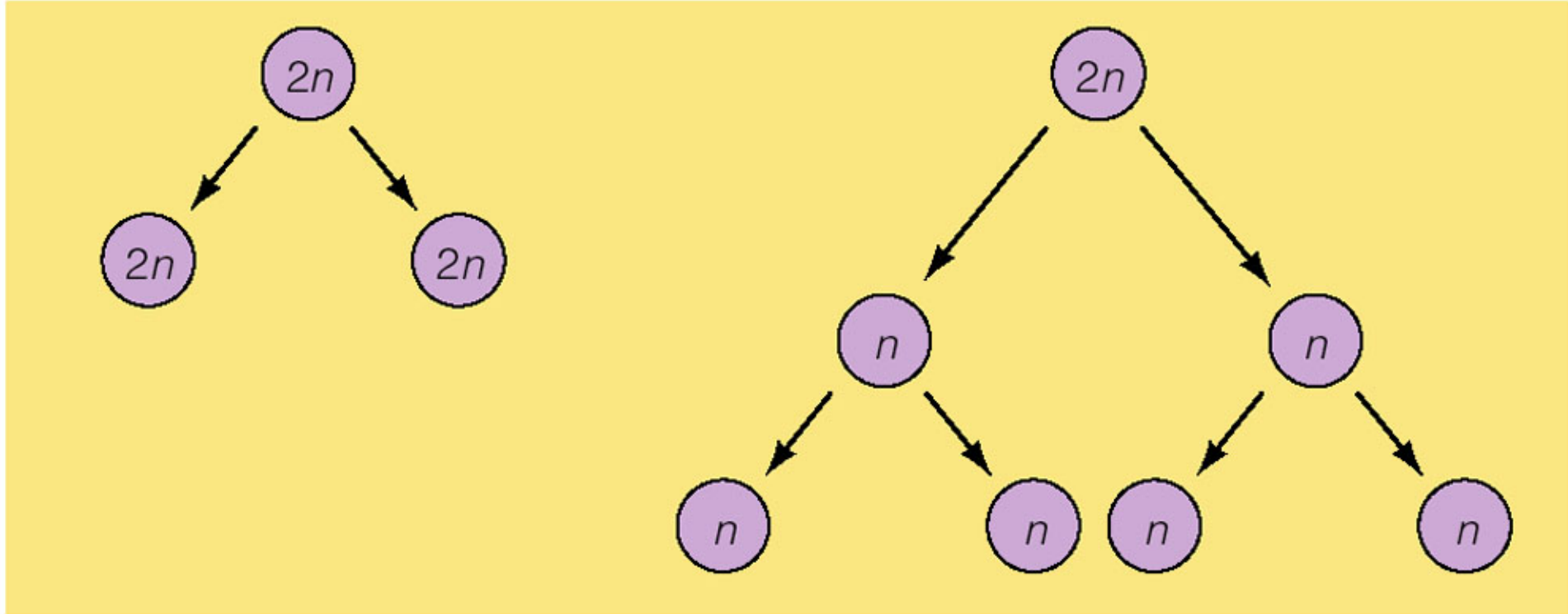


# Human chromosomes

- DNA and associated proteins are organized into chromosomes
- Human somatic cells are **diploid** and have 22 pairs of **autosomes** AND 1 set of **sex** chromosomes (XX or XY)= total of 46
  - Females XX
  - Males XY
- Germ cells are haploid and contain 22 chromosomes plus 1 sex chromosome (X or Y)

# Mitosis

# Meiosis



# Interphase

- **Gap 1 (G1)**– many cytoplasmic organelles are constructed; RNA, protein and other molecules are synthesized; cell almost doubles in size
- **Synthesis (S)**– DNA is replicated and chromosomes duplicate, forming 2 sister chromatids attached at the centromere
- **Gap 2 (G2)**– more cell growth; mitochondria divide; spindle precursors form

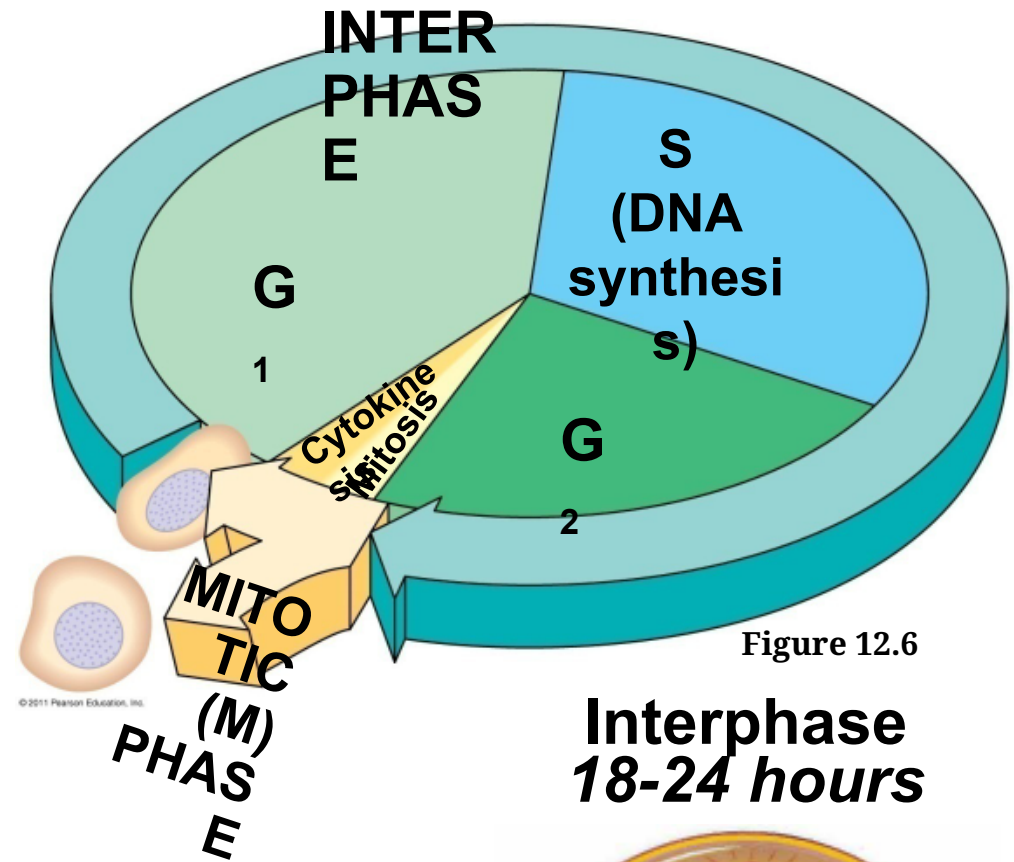
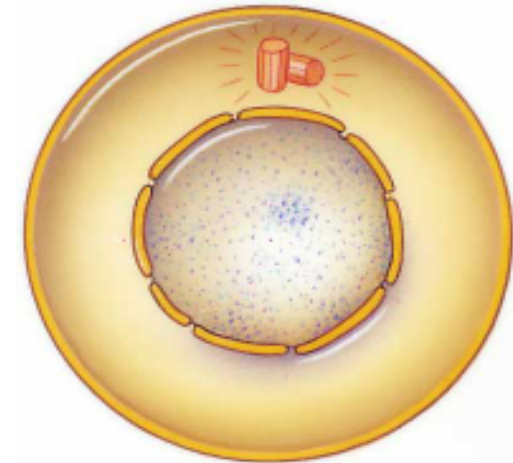


Figure 12.6

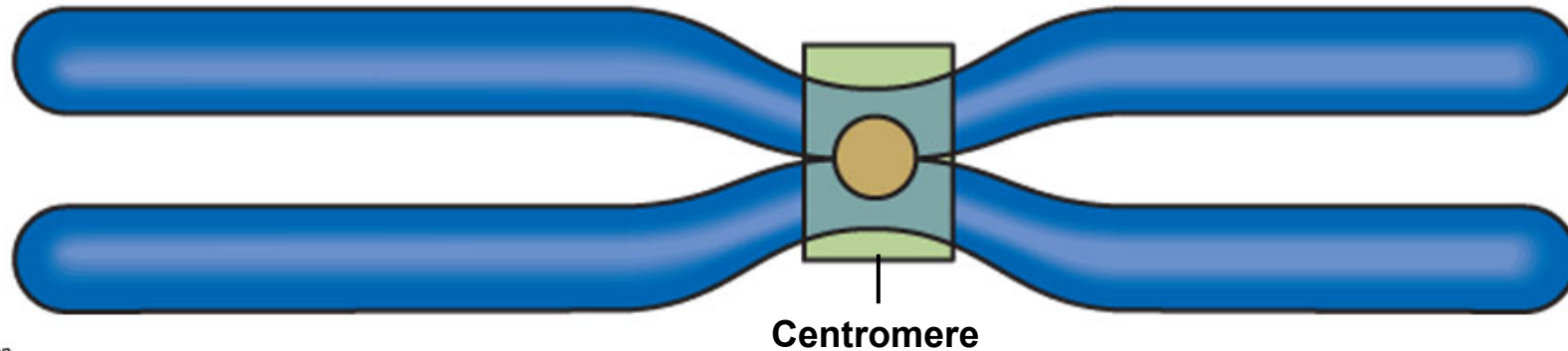
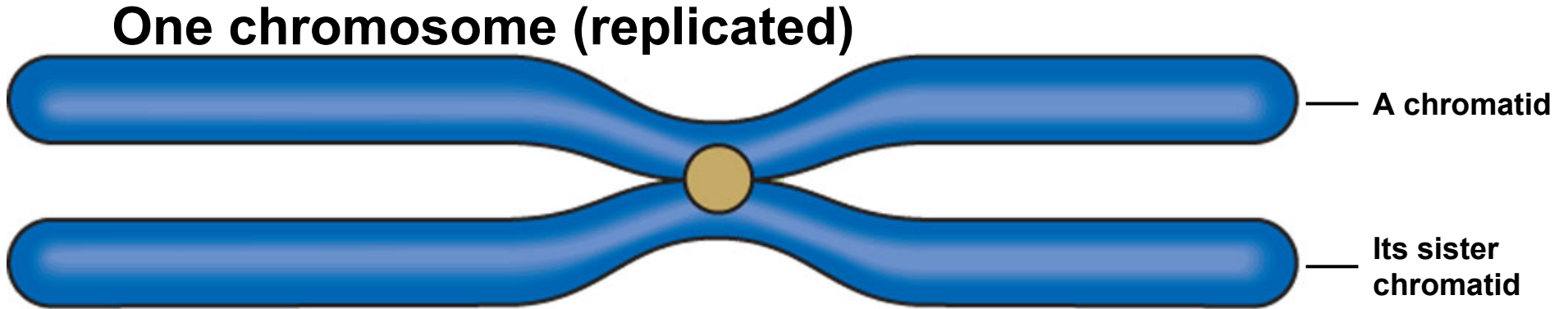
**Interphase**  
**18-24 hours**



**Fig.**  
**2.8a**

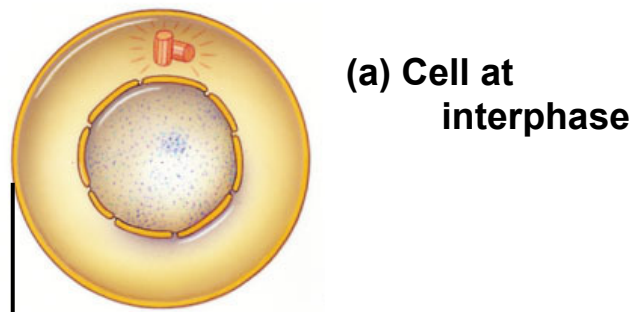


**One chromosome (unreplicated)**



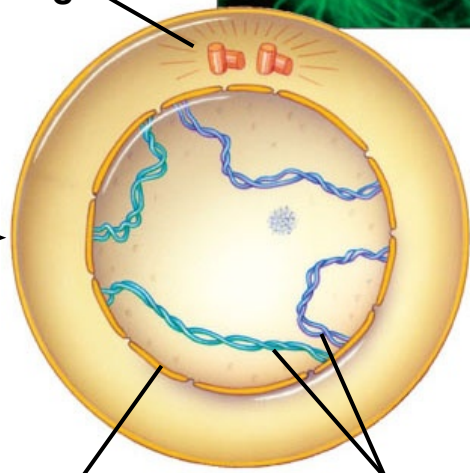
# Mitosis

- Produces identical daughter cells
  - (46 chromosomes)
- It must be accurate for cells to function properly
- Continuous process but divided into distinct steps:
  - Prophase
  - Metaphase
  - Anaphase
  - Telophase



Mitosis

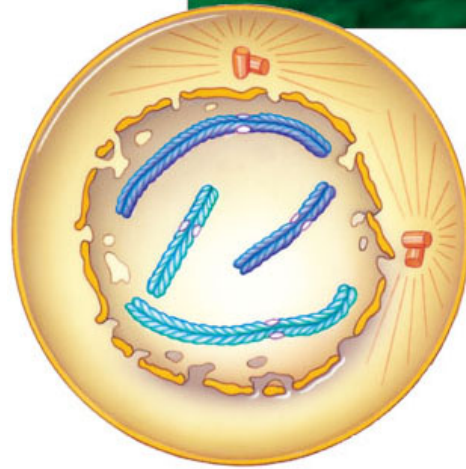
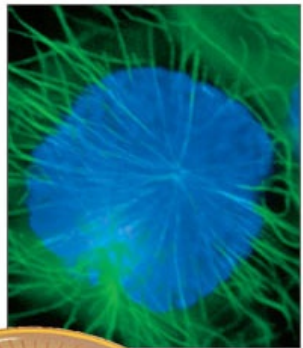
Pair of centrioles



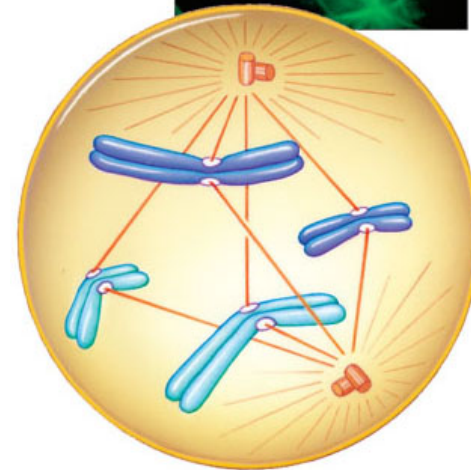
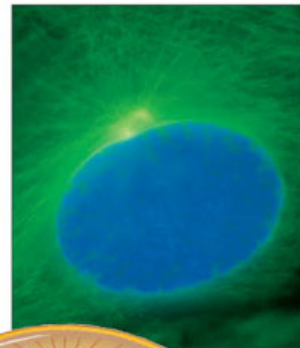
Nuclear envelope

Chromosome

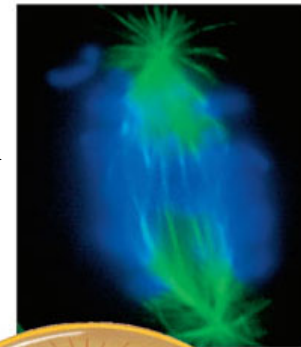
(b) Early Prophase



(c) Late Prophase

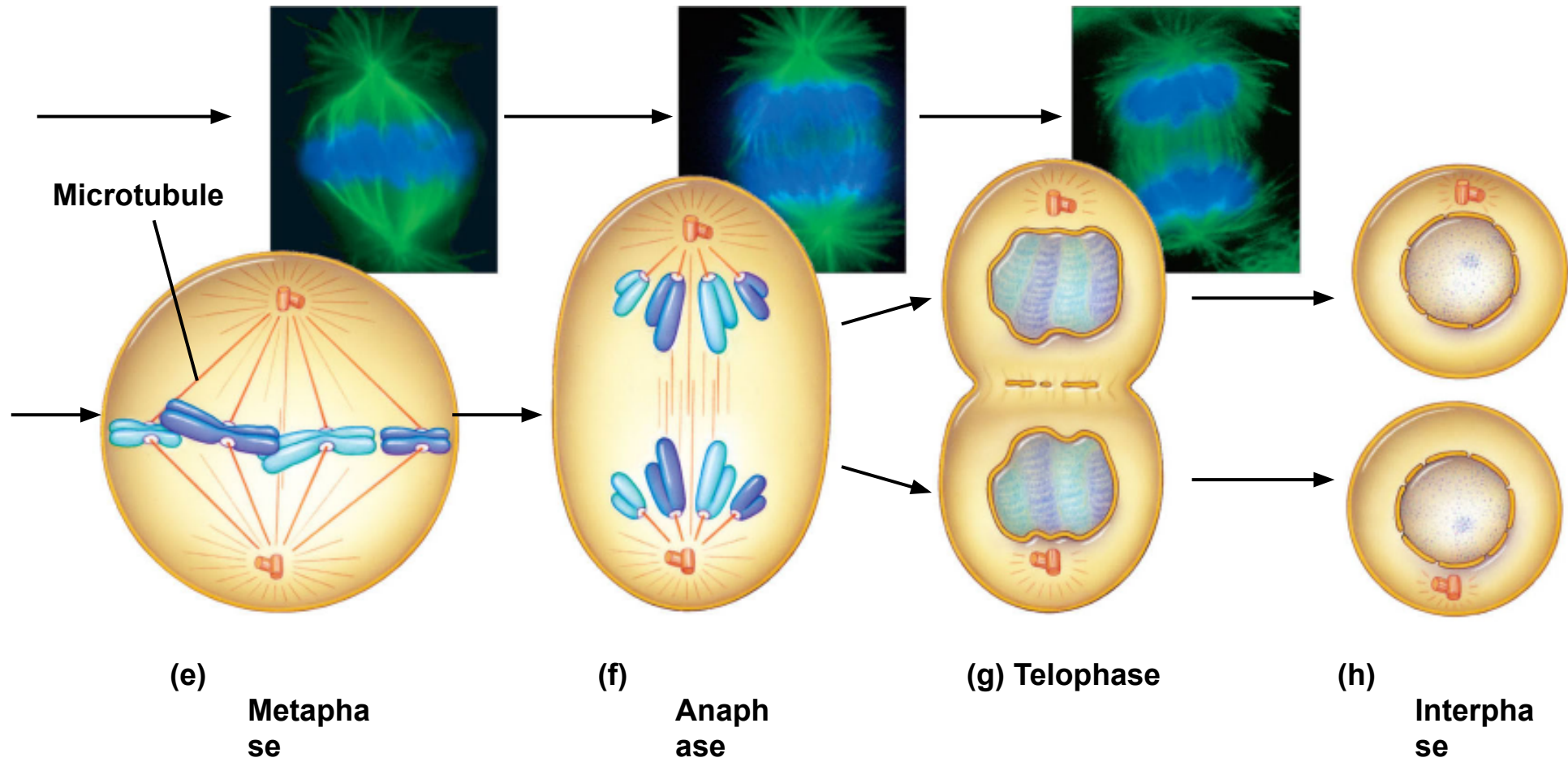


(d) Transition to Metaphase



Stepped  
Art





# The Stages of Meiosis

- After chromosomes duplicate, two divisions follow
  - Meiosis I (reductional division): homologs pair up and separate, resulting in two haploid daughter cells with replicated chromosomes
  - Meiosis II (equational division) sister chromatids separate
- The result is four haploid daughter cells with unreplicated chromosomes

Figure 13.7-1

## Interphase

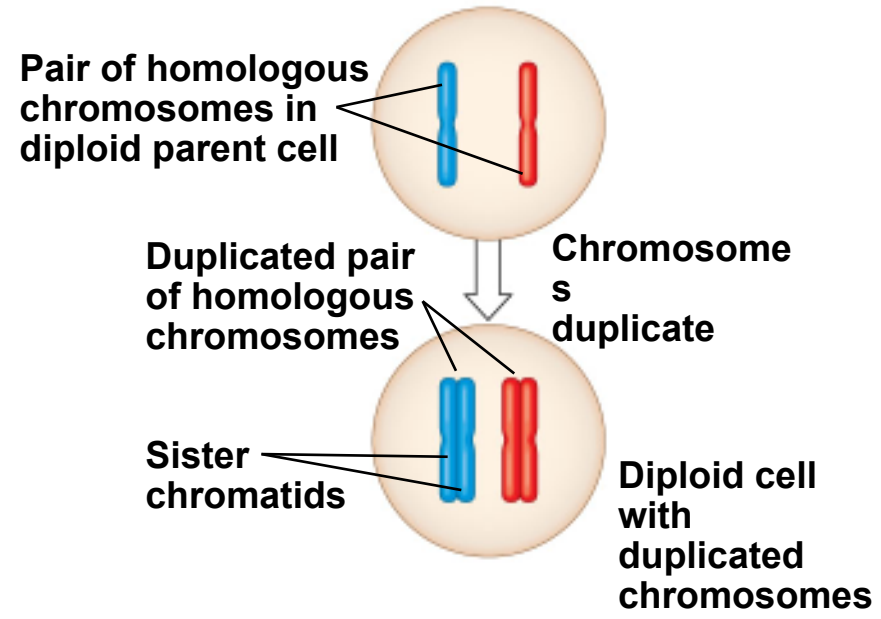


Figure 13.7-2

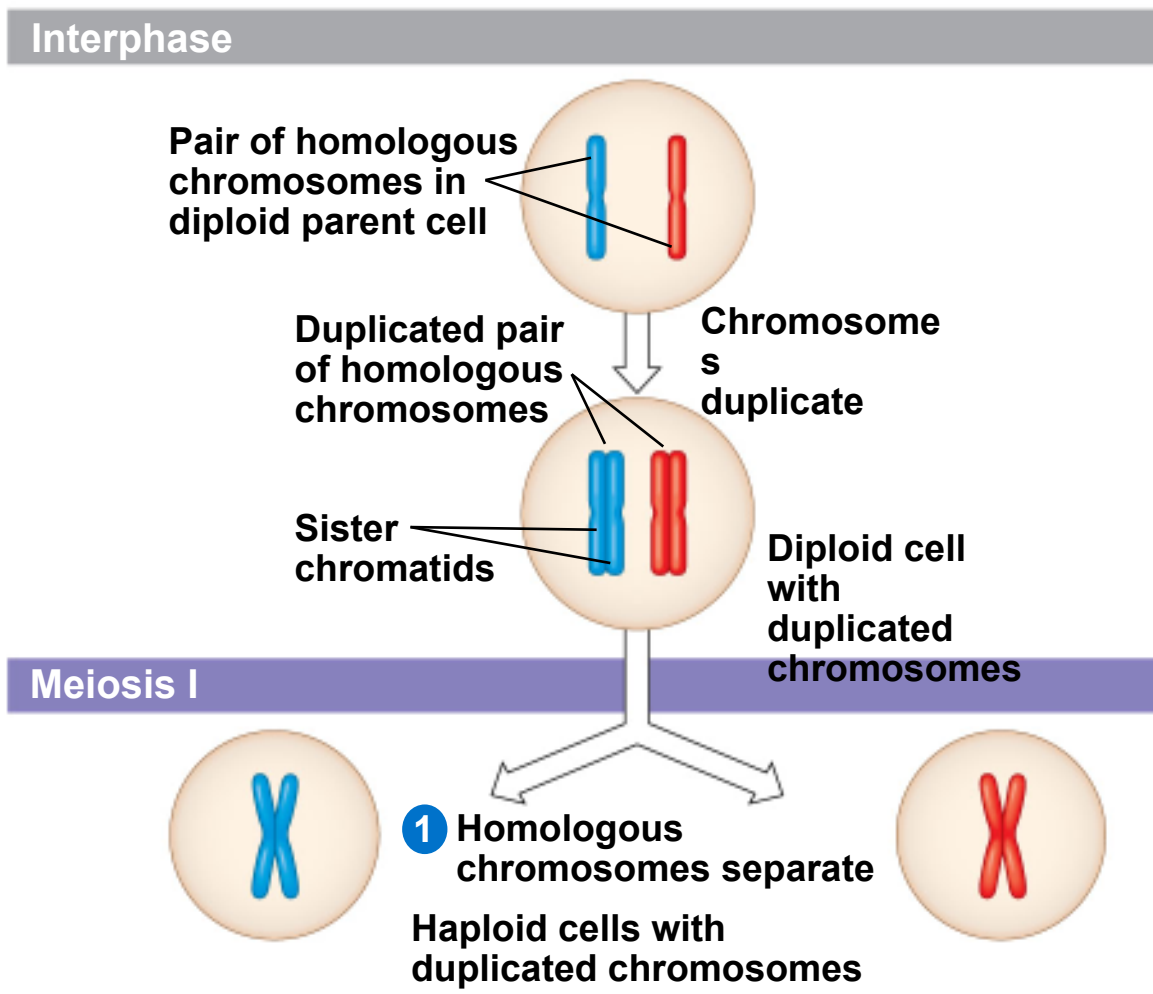
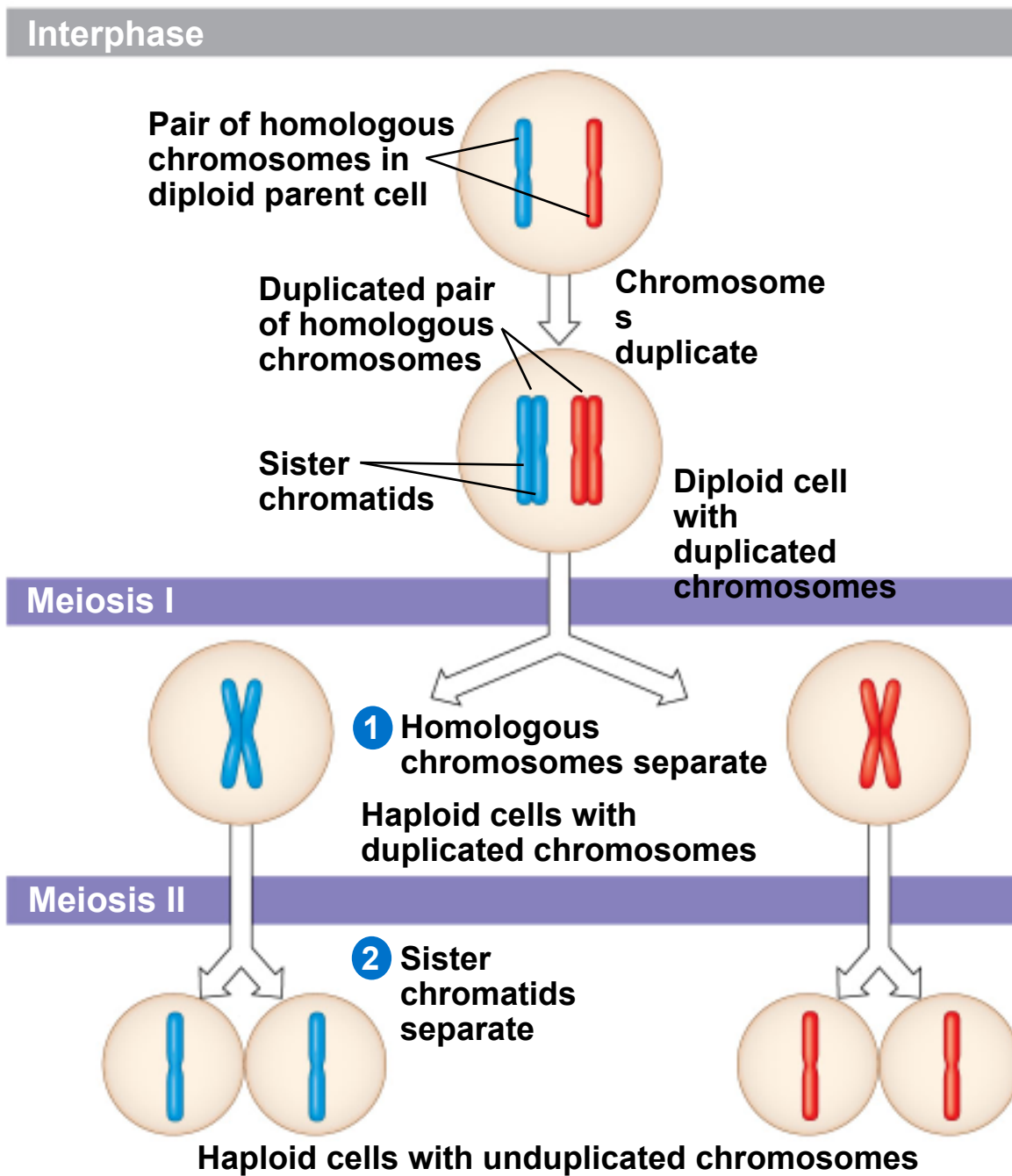
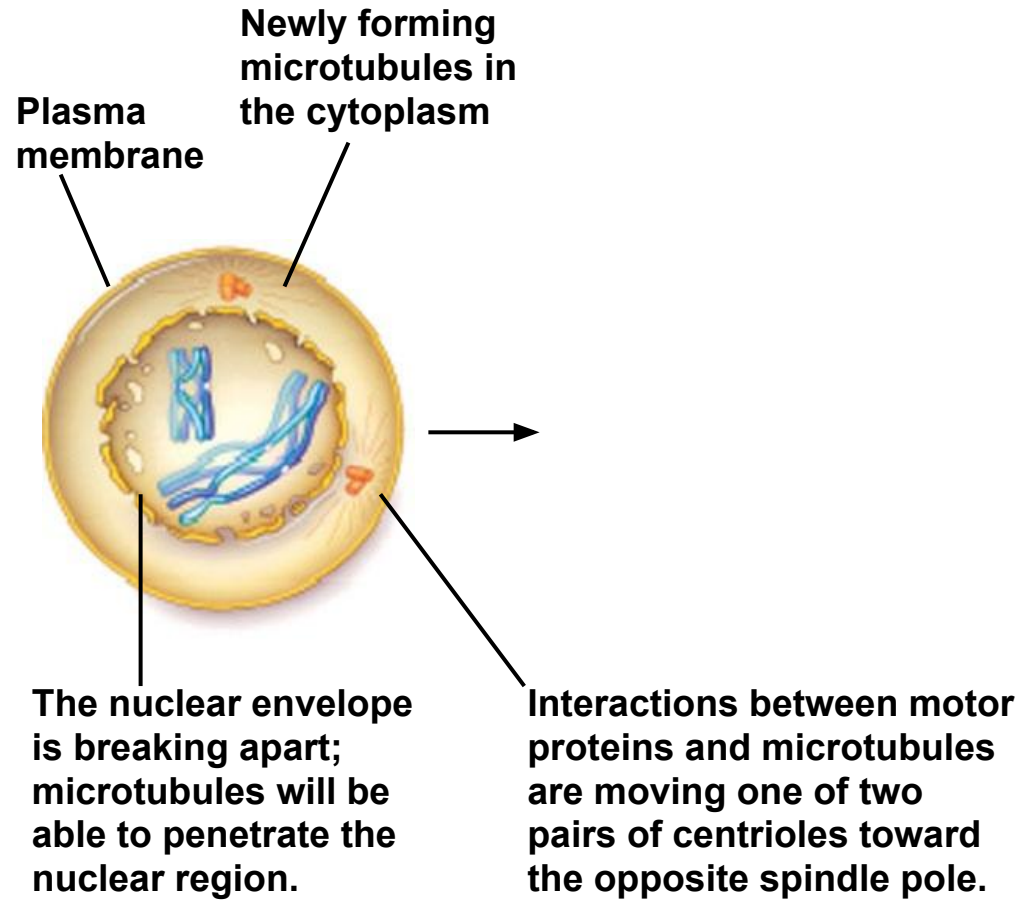


Figure 13.7-3

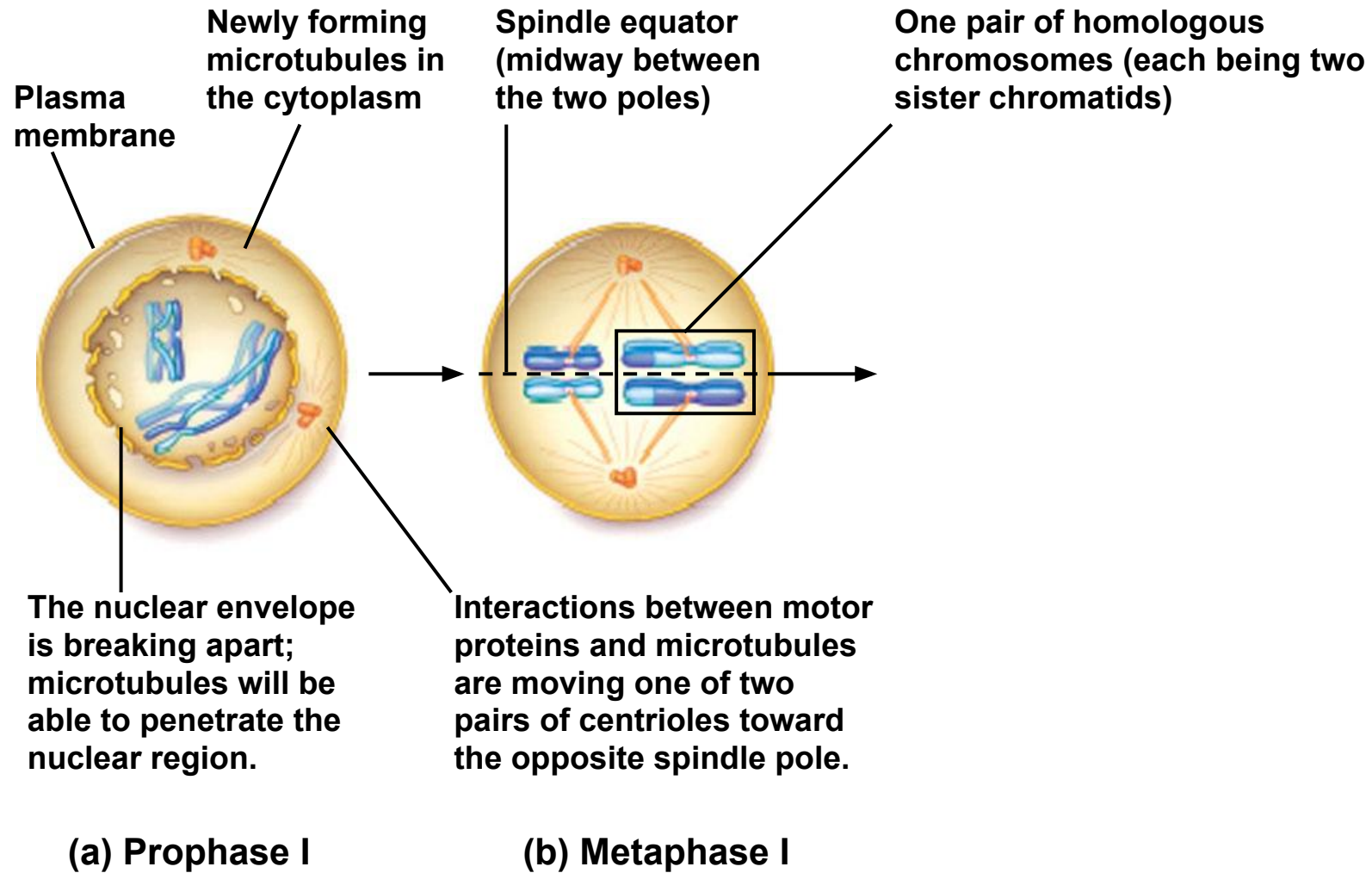


# Meiosis I

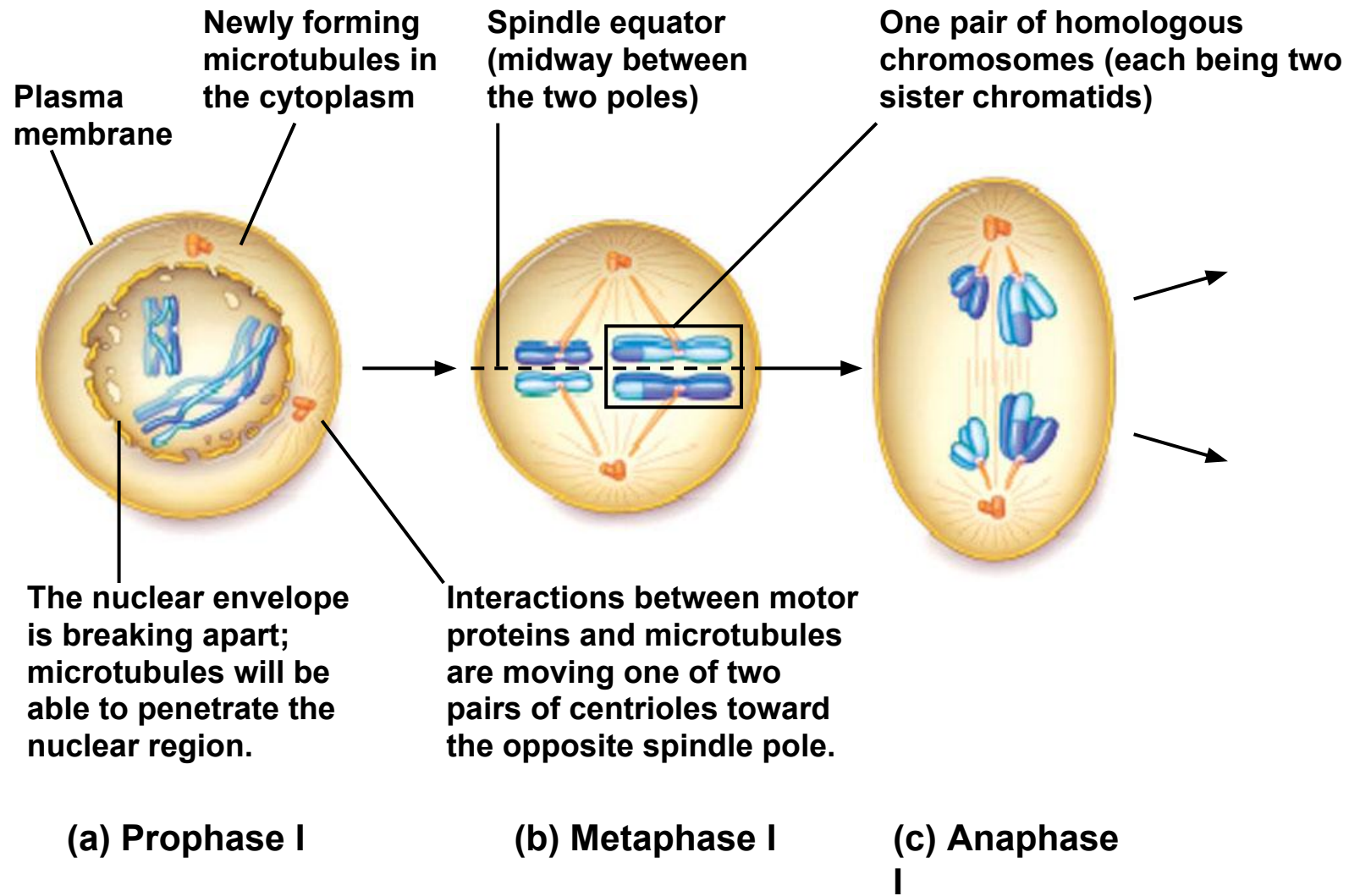


**(a) Prophase I**

# Meiosis I

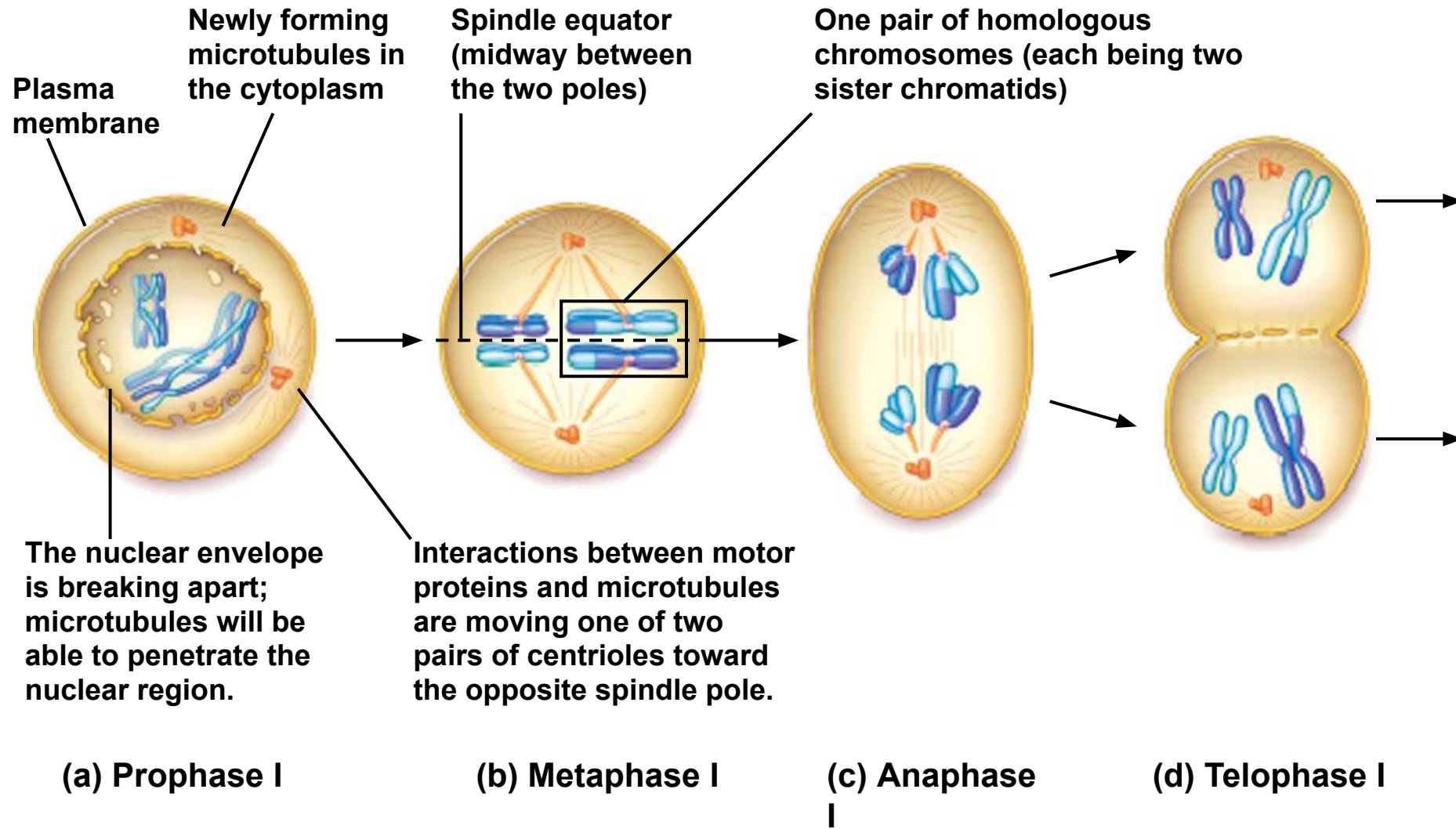


# Meiosis I

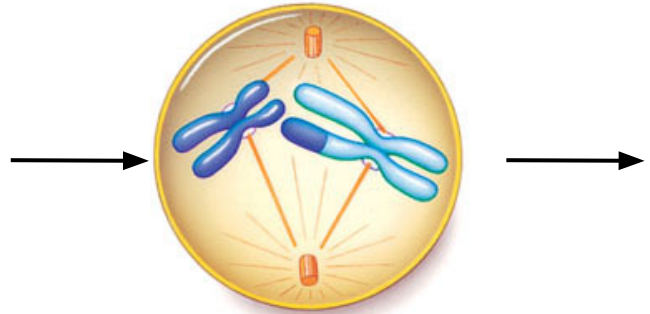




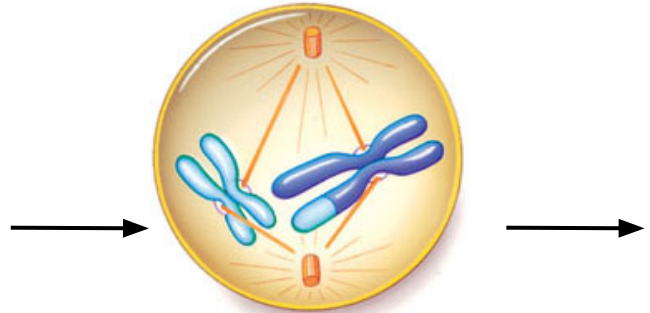
# Meiosis I



# Meiosis II

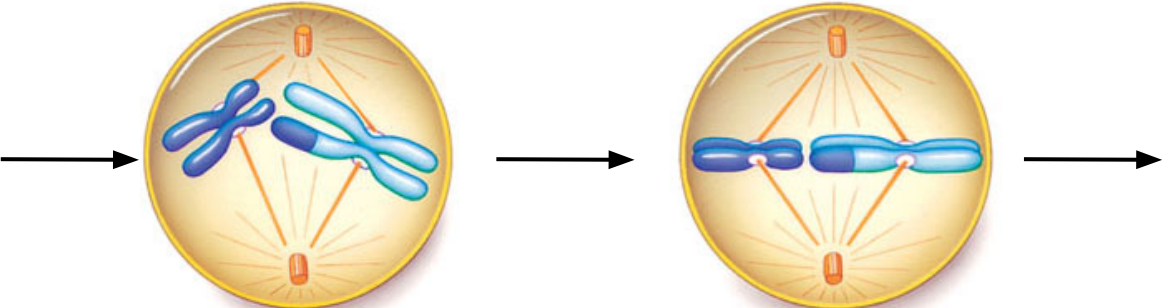


*There is no DNA replication  
between the two nuclear divisions.*

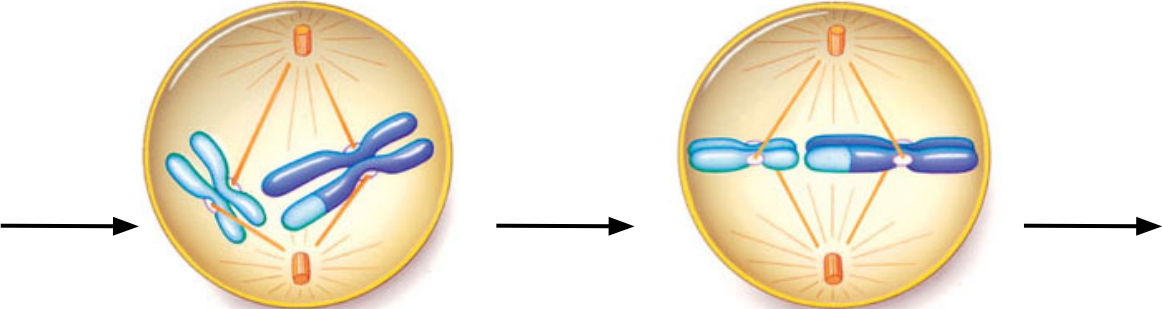


**(e) Prophase II**

# Meiosis II



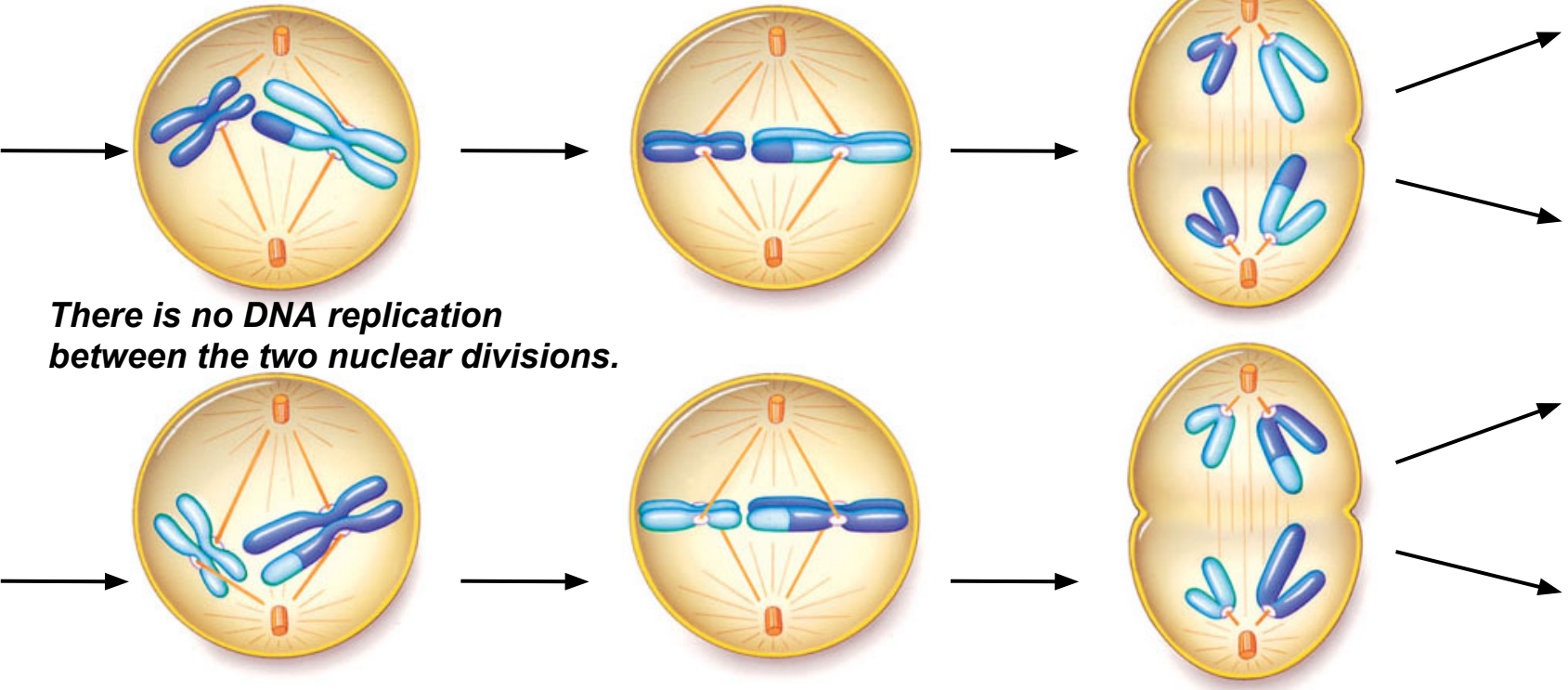
*There is no DNA replication between the two nuclear divisions.*



**(e) Prophase II**

**(f) Metaphase II**

# Meiosis II



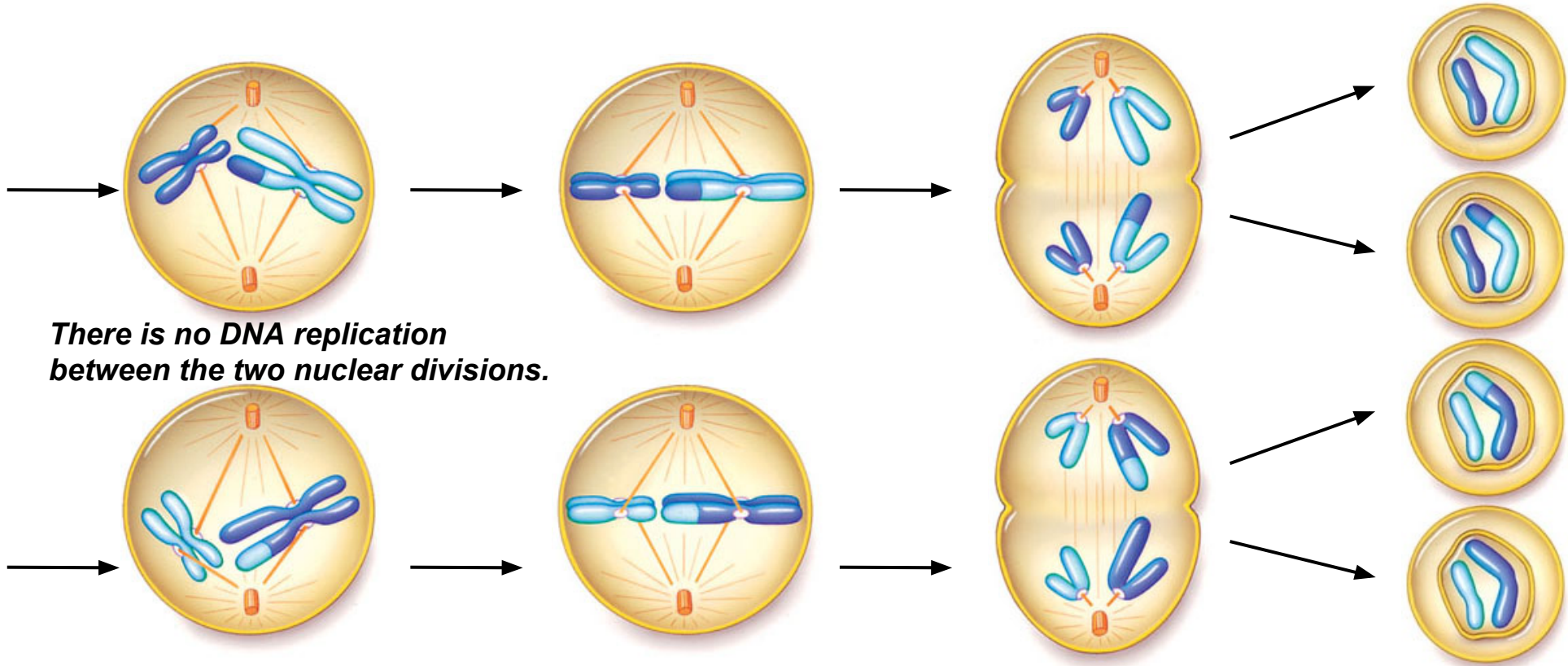
*There is no DNA replication between the two nuclear divisions.*

**(e) Prophase II**

**(f) Metaphase II**

**(g) Anaphase II**

# Meiosis II



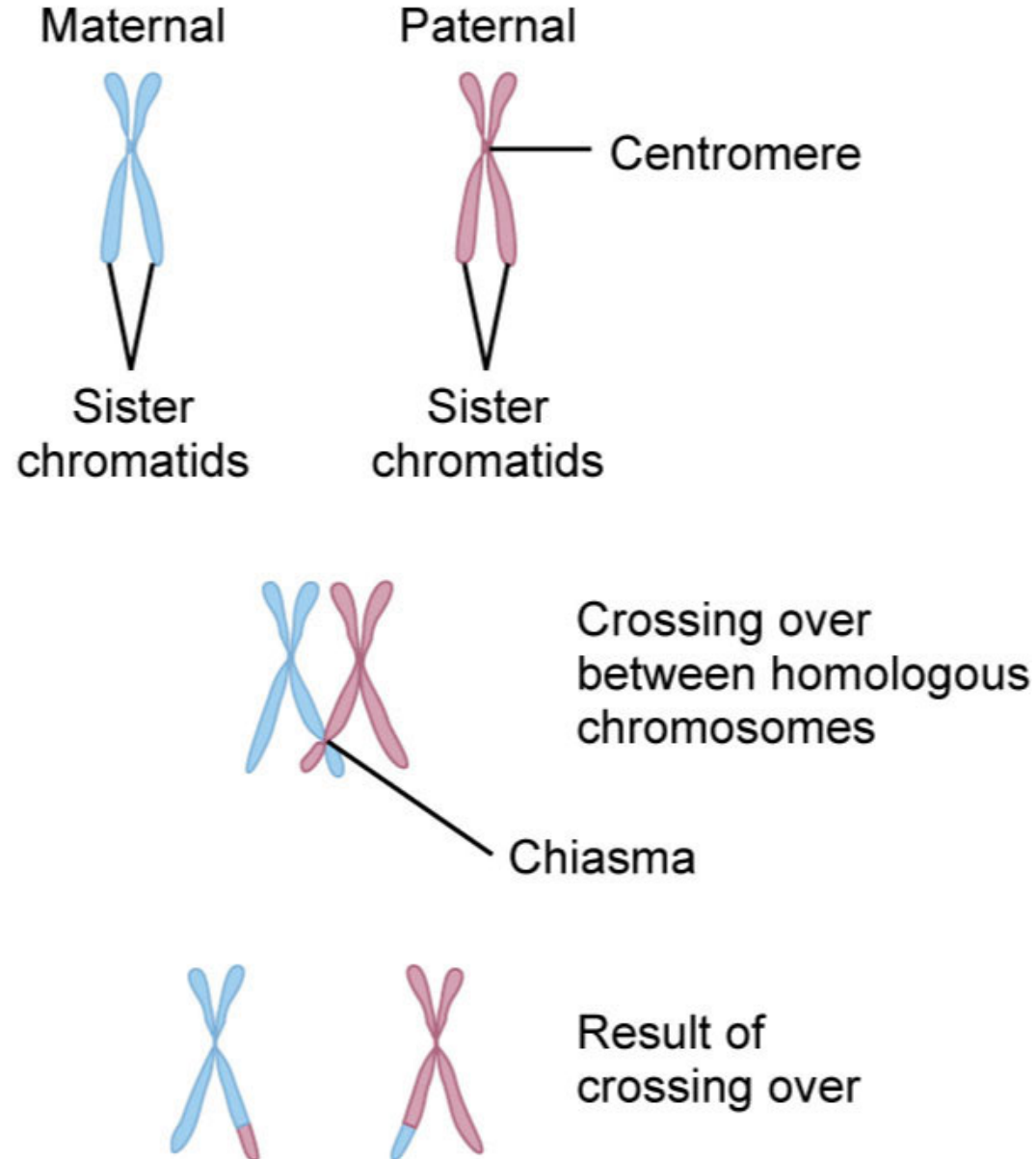
(e) Prophase II

(f) Metaphase II

(g) Anaphase II

(h) Telophase II

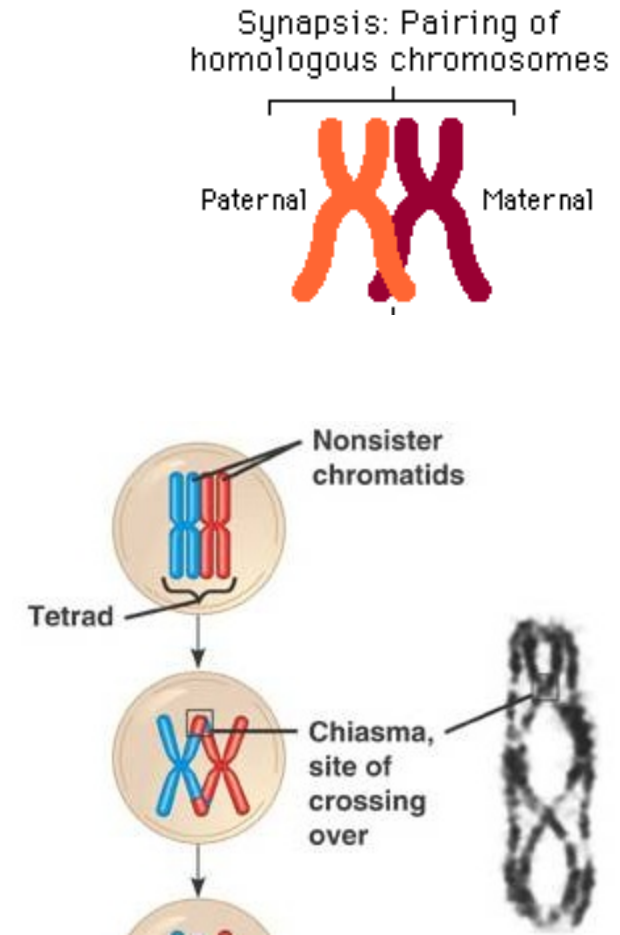
# Homologous chromosomes

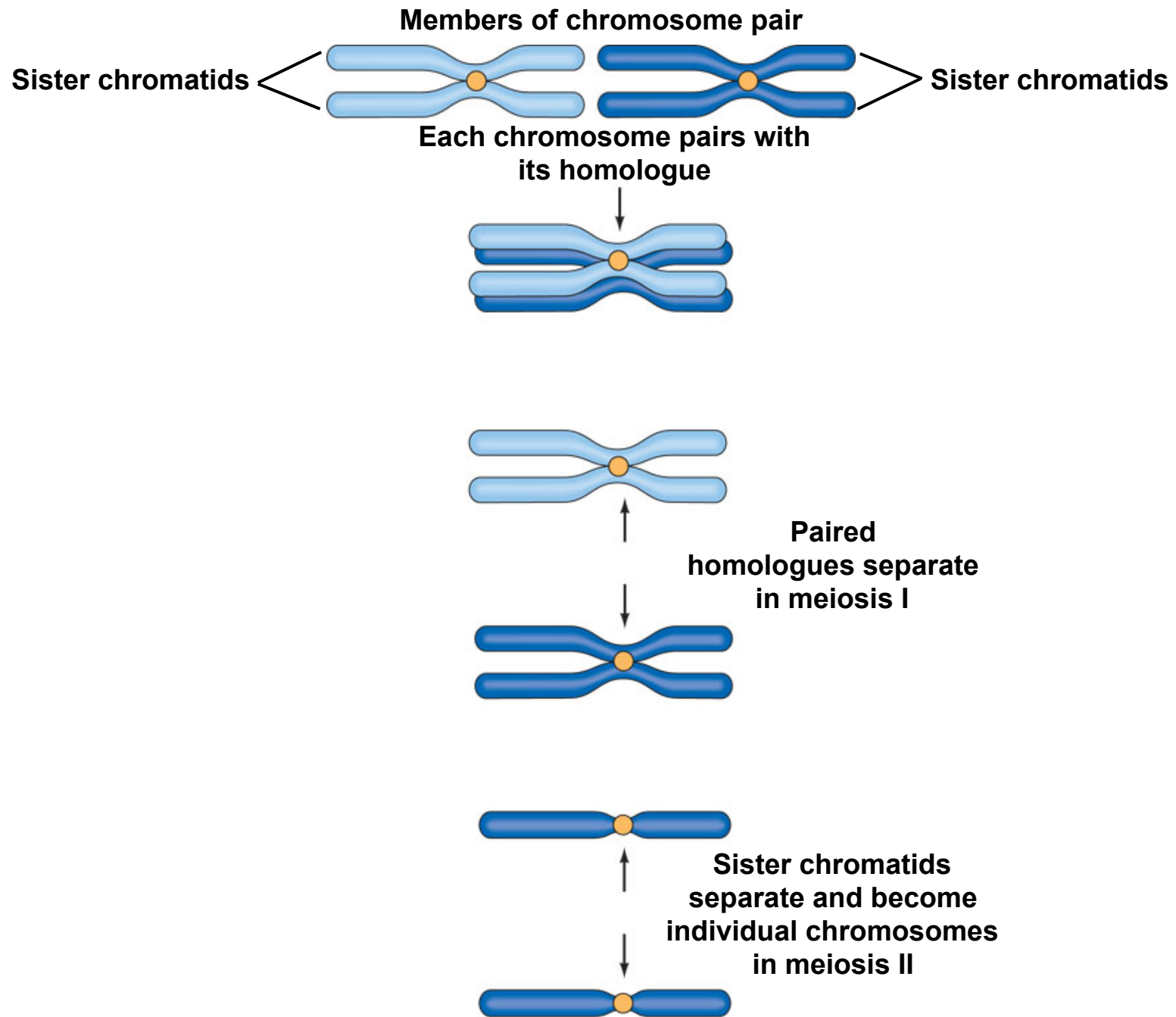




# Prophase I

- Leptotene
  - Replicated chromosomes align and begin to condense
- Zygotene
  - homologous chromosomes pair along entire length (**synapsis**)
  - synaptonemal complex forms
- Pachytene
  - Synapsis is complete and each pair of homologues is called a **tetrads (bivalent)**
  - **Crossing over** occurs (recombination at **chiasmata**)
- Diplotene
  - Homologous chromosomes separate some but remain bound at chiasmata
    - usually 2 chiasmata/chromosome, more frequent in females)
- Diakinesis
  - Further chromosome condensation; tetrads viable







# Genetic consequences of meiosis

- Reduction of chromosome number
- Diploid to haploid (essential for gametes)
- Random assortment of maternal and paternal chromosomes
  - genes on different chromosomes
  - maternal/paternal chromosomes
  - Number of possible chromosomal combinations =  $2^{23}$  or 8,388,608
  - Recombination between chromosome pairs increases the possible combinations
- Segregation of alleles
- Recombination/crossing-over
  - Allows new combinations of genes to be produced
  - Important for normal chromosome disjunction
  - Ensures genetic diversity

# Chromosome combinations: independent assortment

