

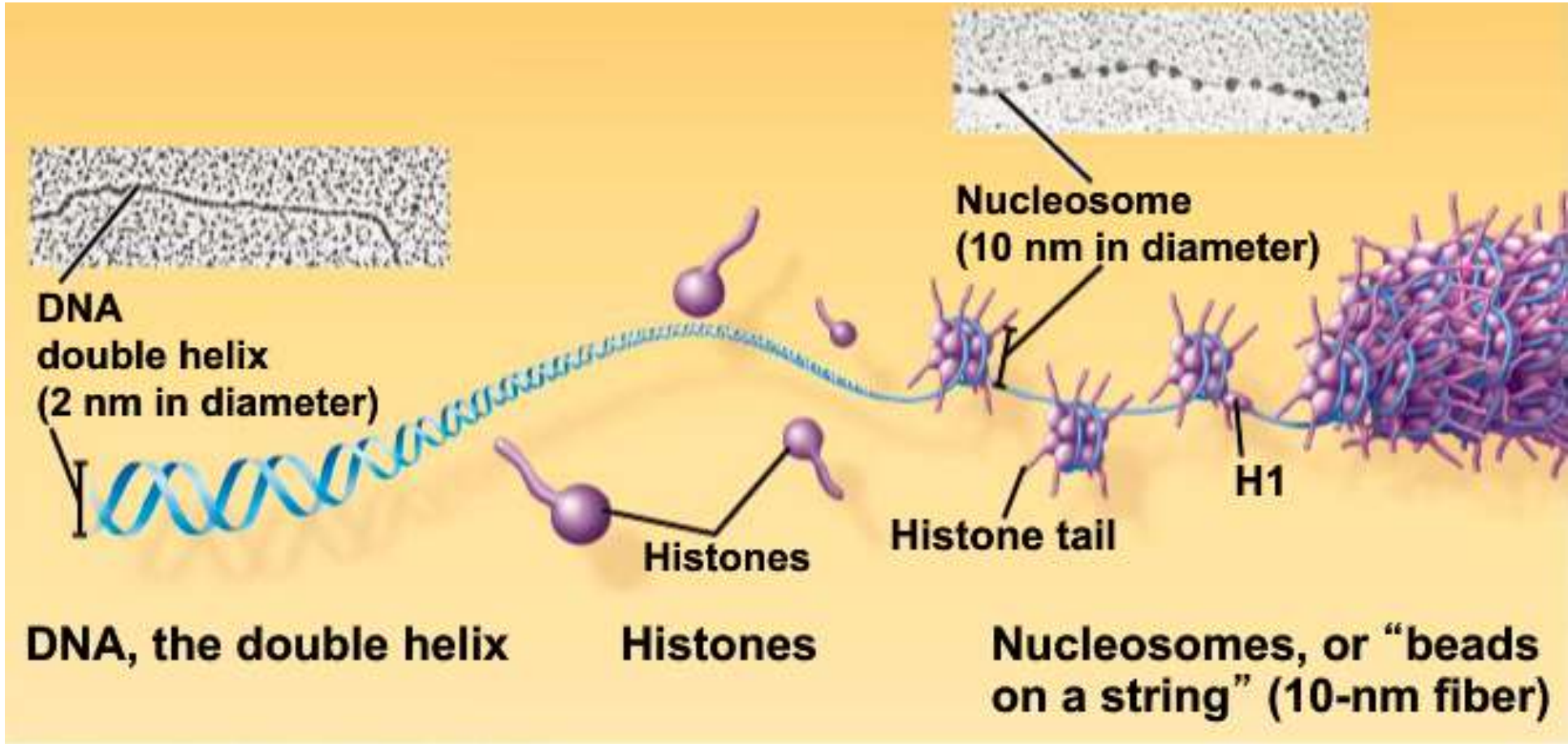
# History of Cytogenetics

True chromosome number  
established in 1956



*“ From their vantage through the microscope,  
the cytogeneticists’ view of the genome is still  
unrivalled in its scope, detail and color.”*

*Barbara J. Trask, 2002*



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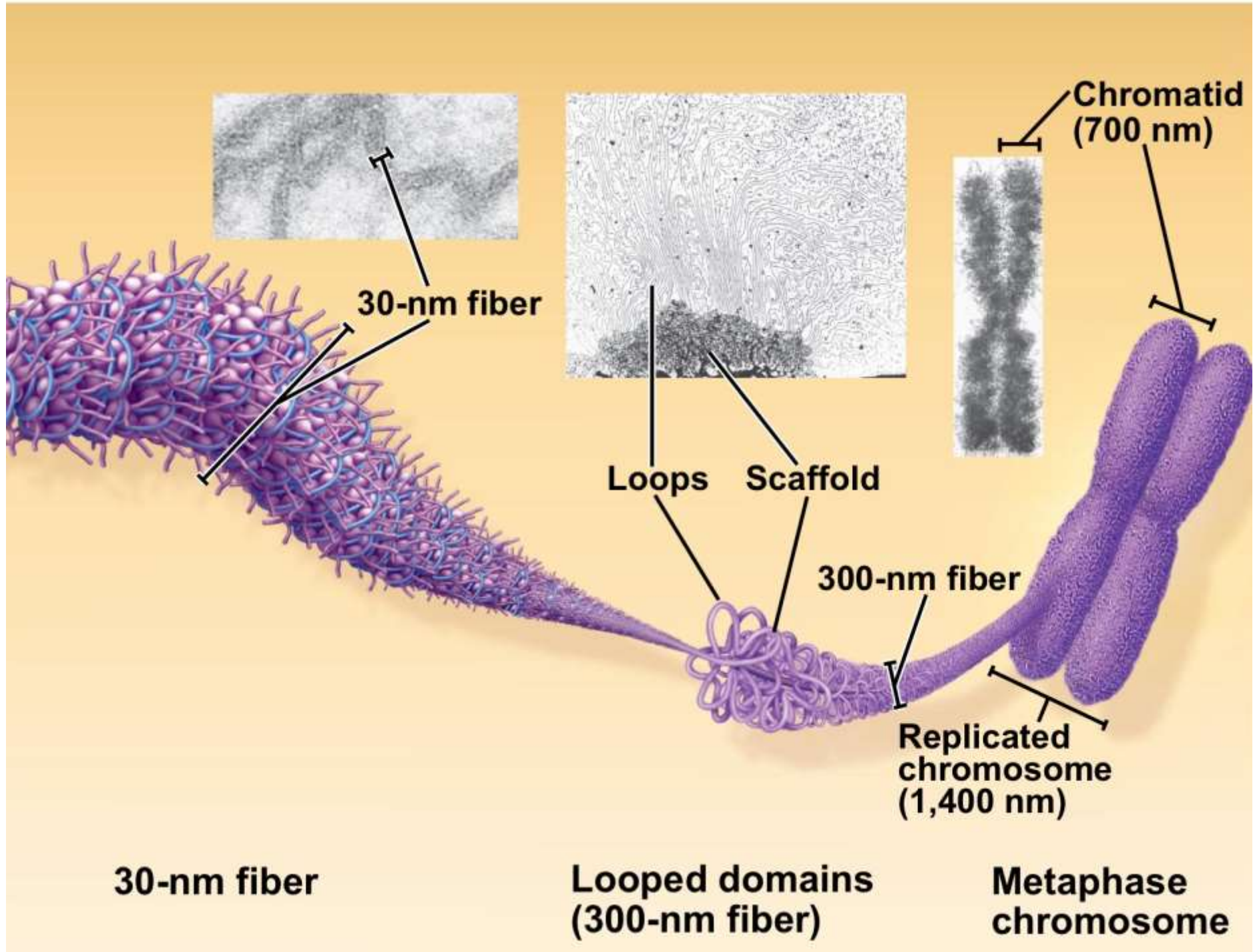
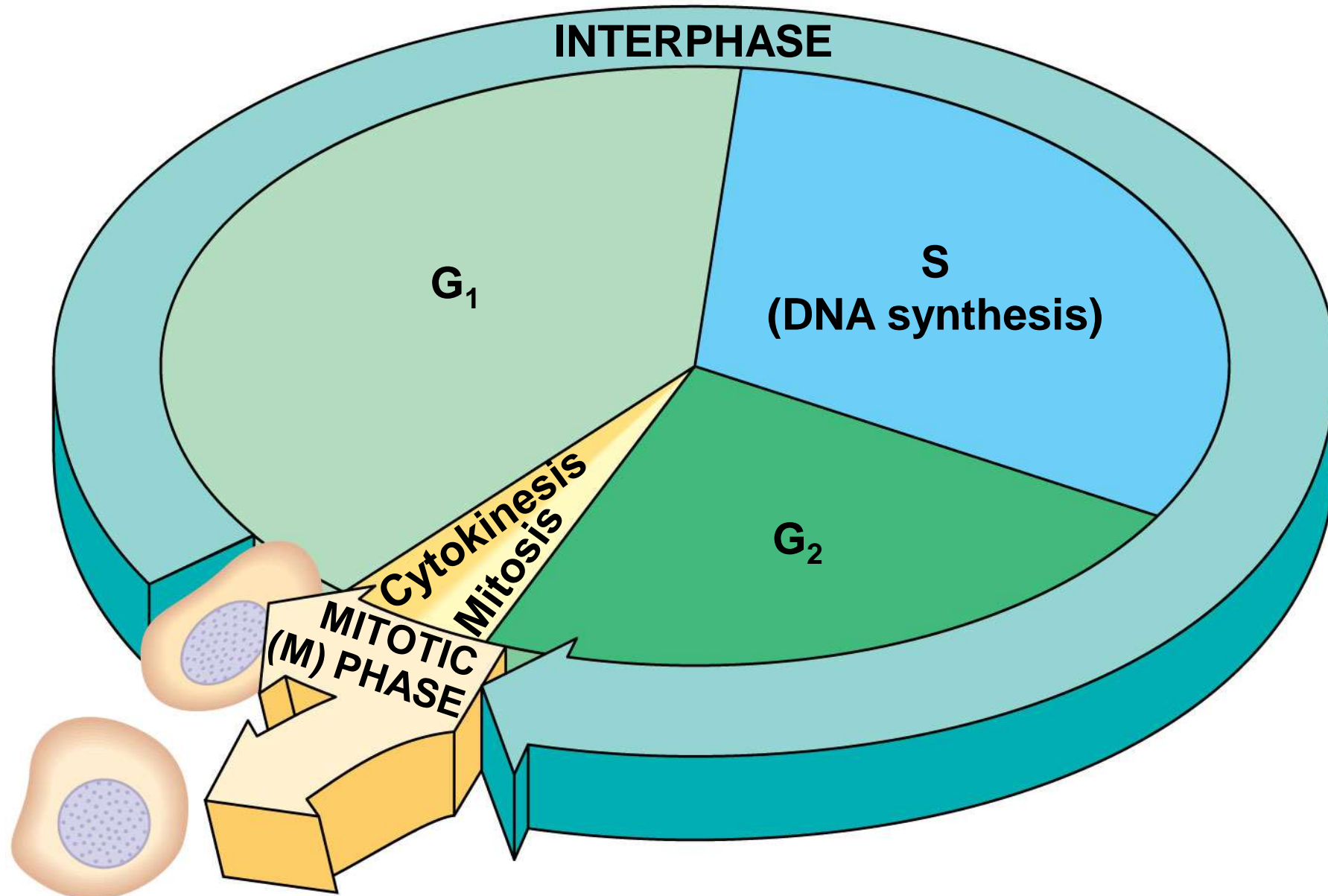
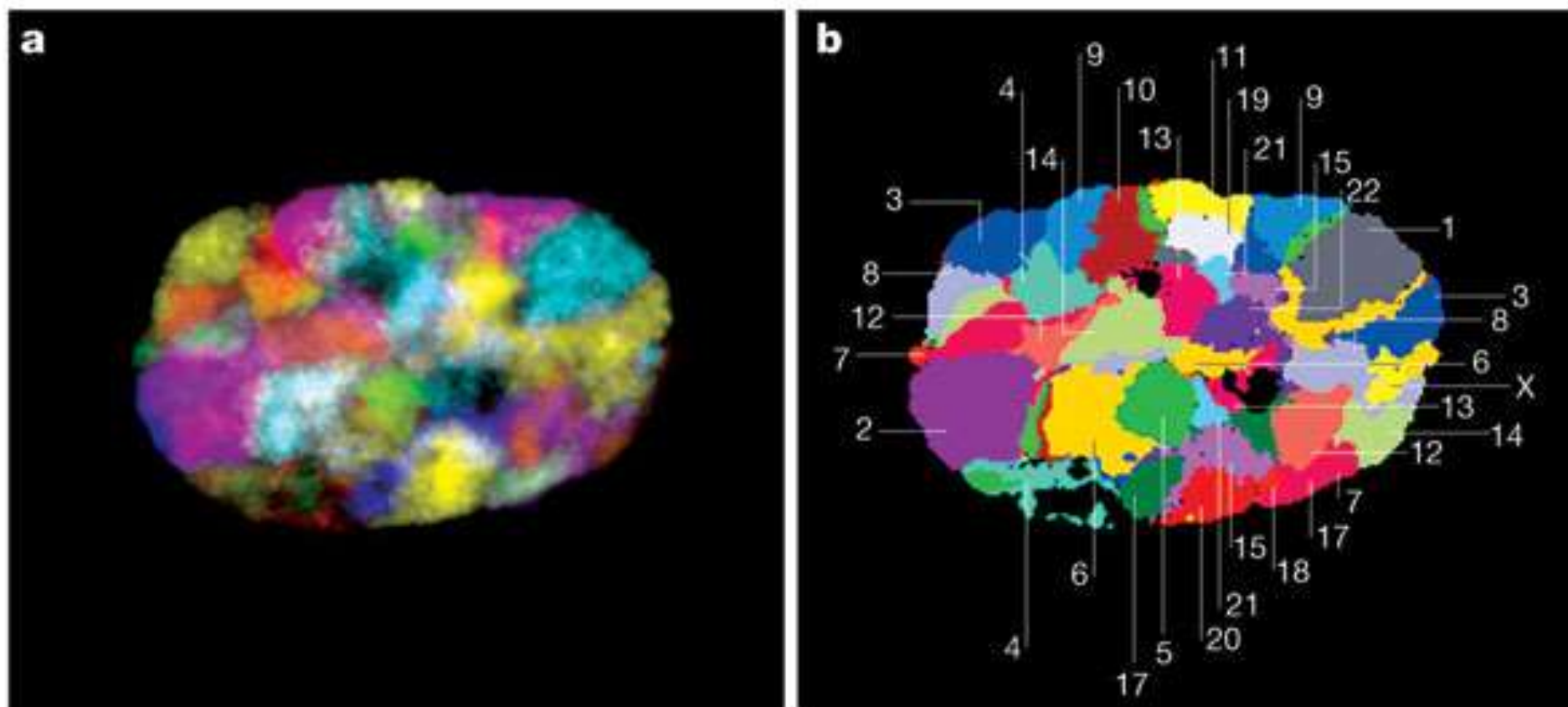
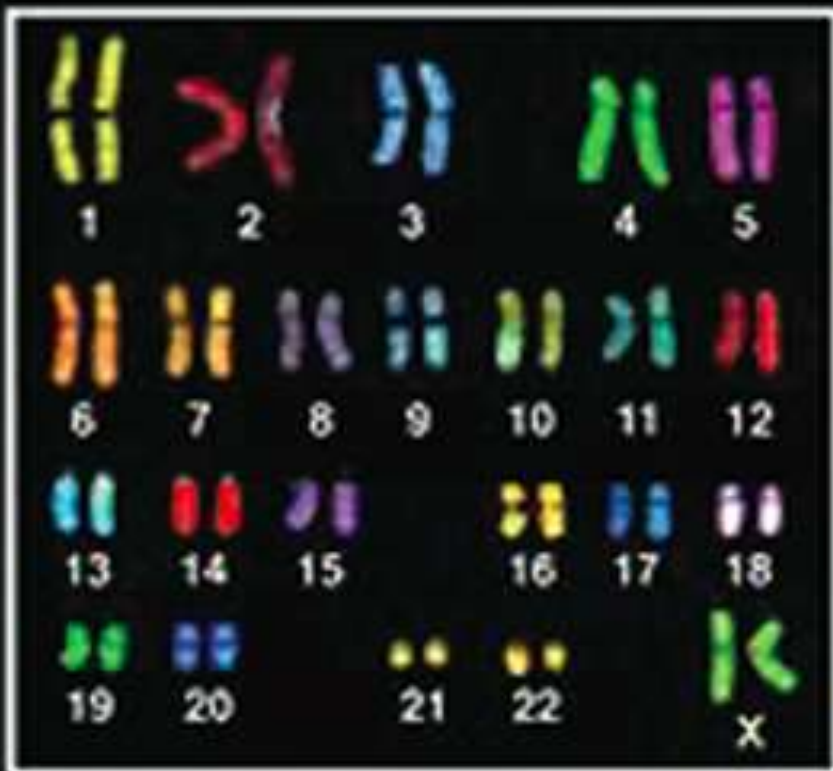
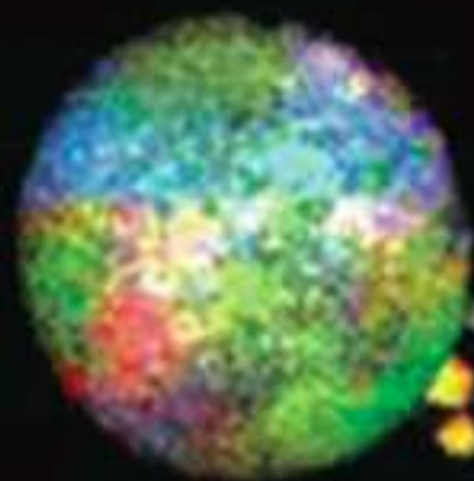


Figure 12.6

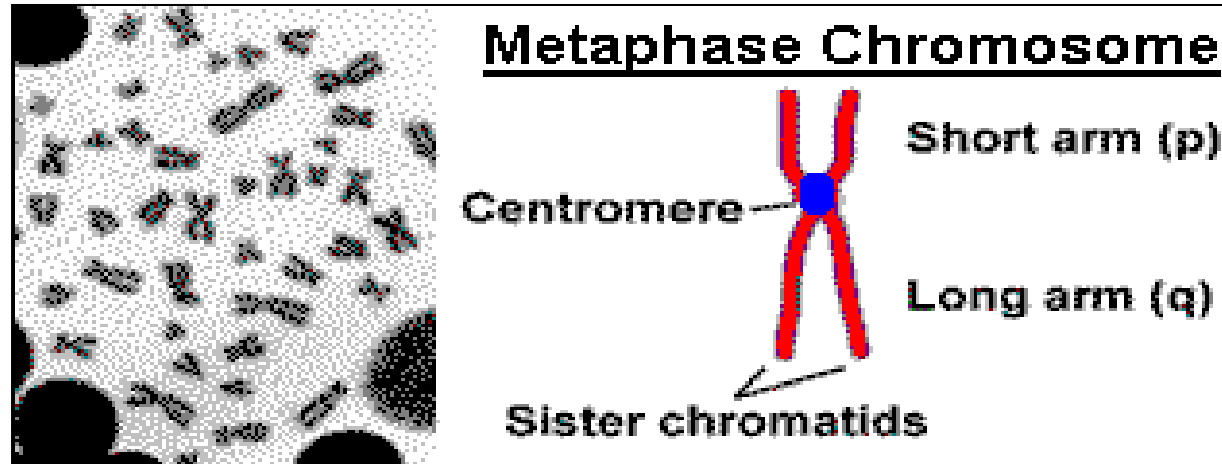




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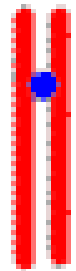
# Nomenclature of chromosomes



## Centromeric position and arm length



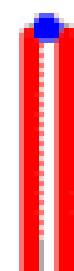
Metacentric



Submetacentric



Acrocentric



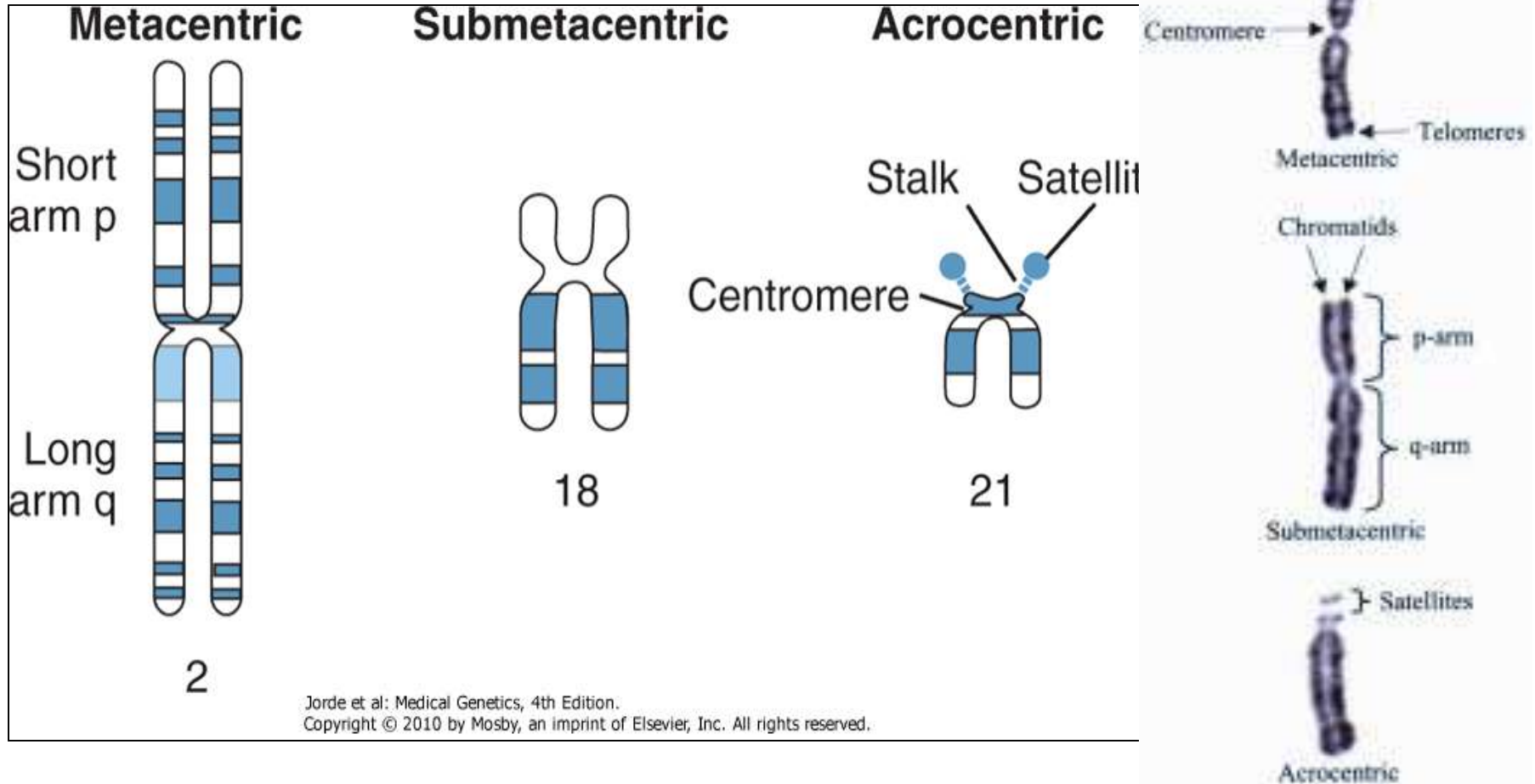
Telocentric

# Chromosome Shape

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

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

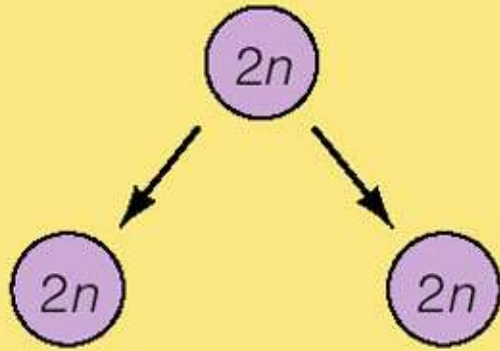
**Acrocentric** – centromere is placed near the end



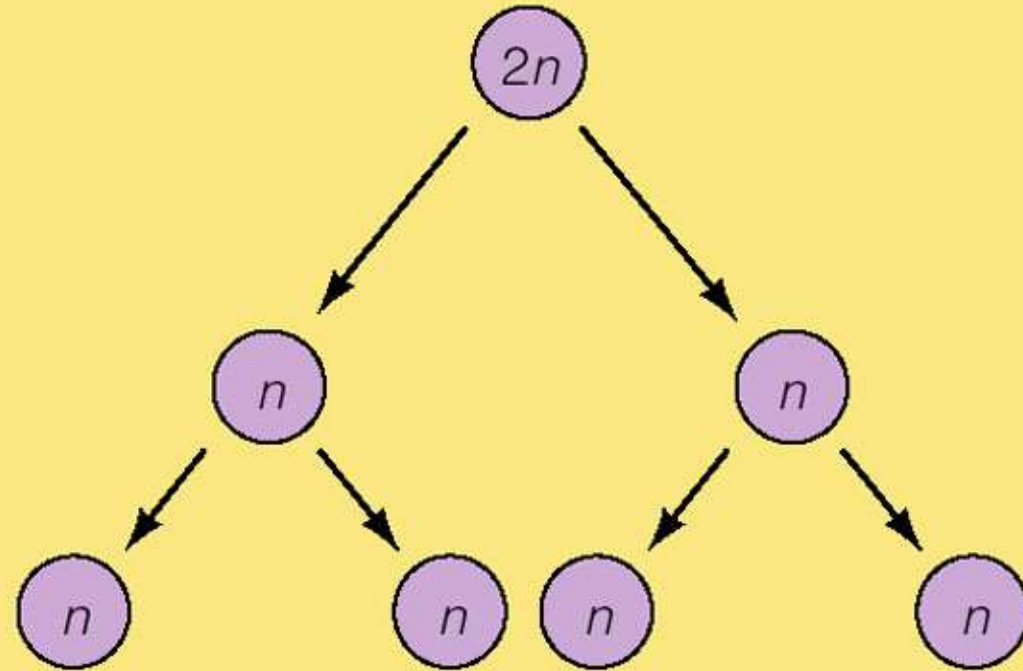
# Human chromosomes

- 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 (G<sub>1</sub>)**– 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 (G<sub>2</sub>)**– more cell growth; mitochondria divide; spindle precursors form

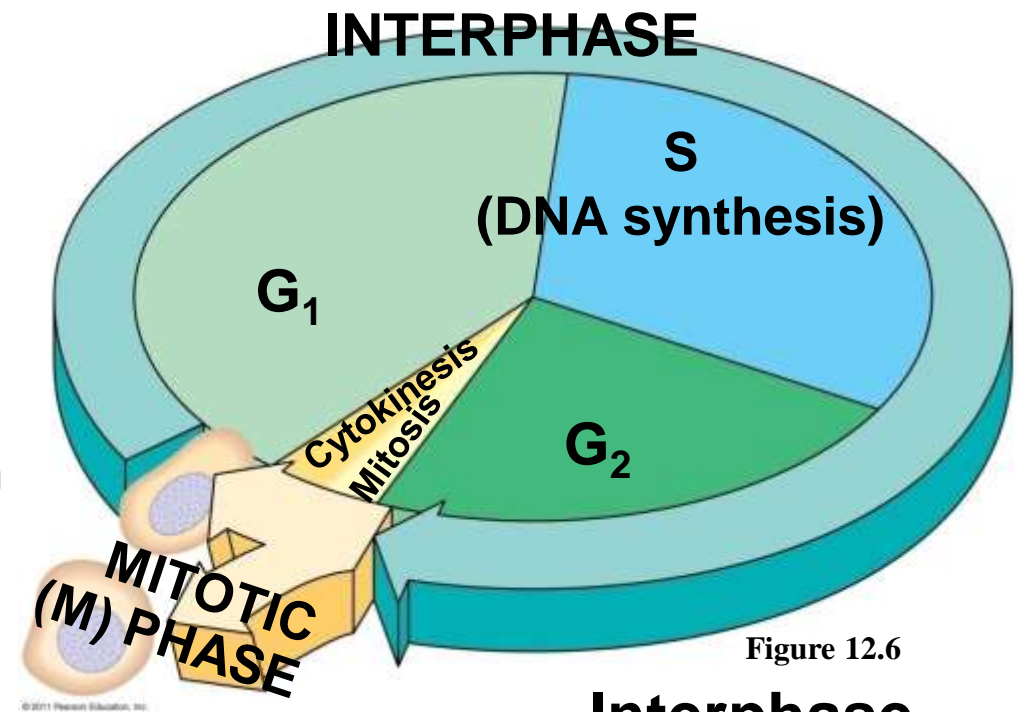
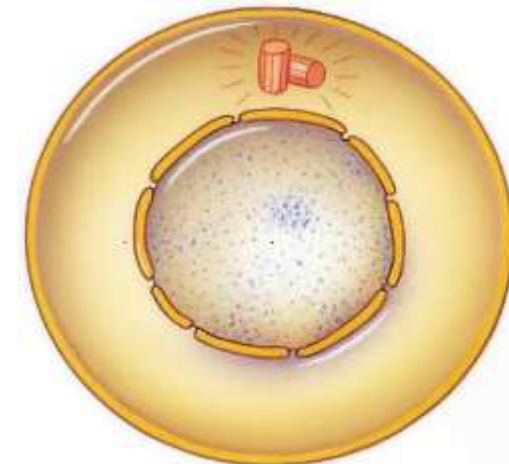


Figure 12.6

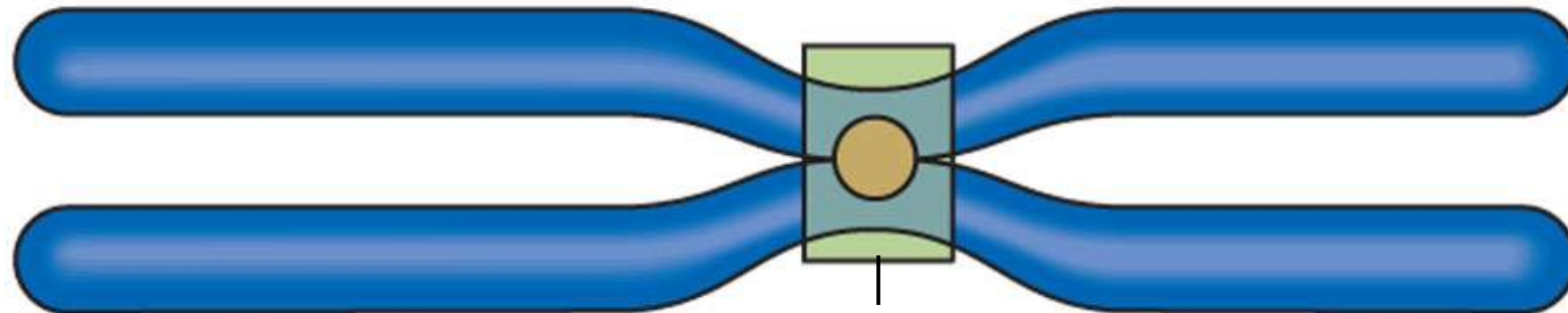
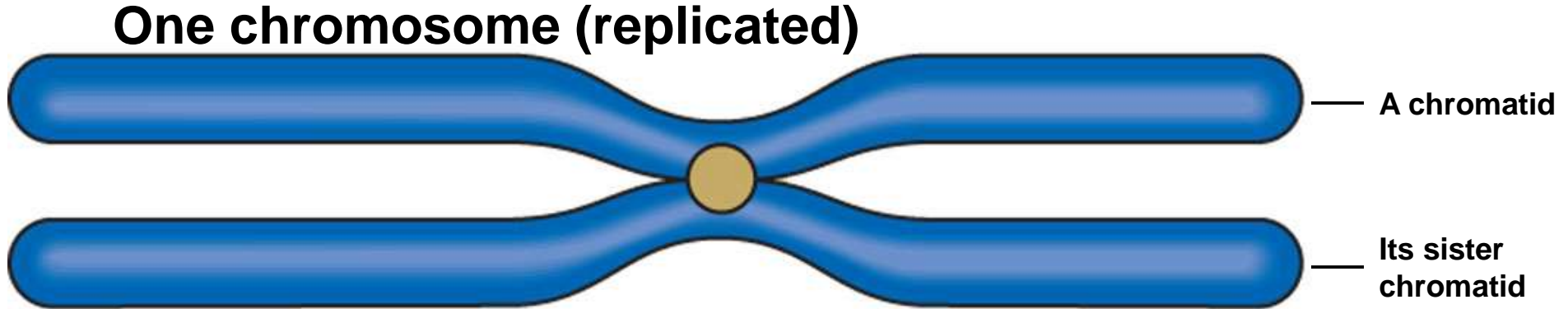
**Interphase**  
**18-24 hours**



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



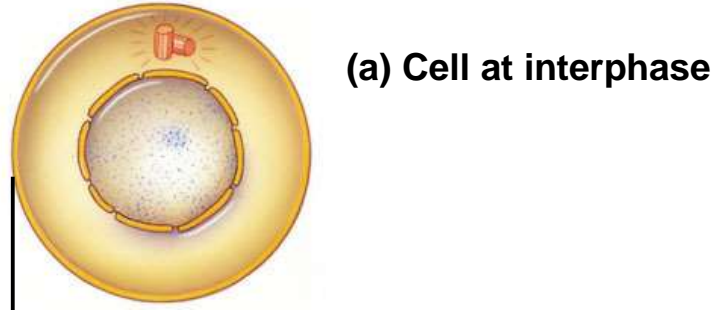
**One chromosome (unreplicated)**



**Centromere**

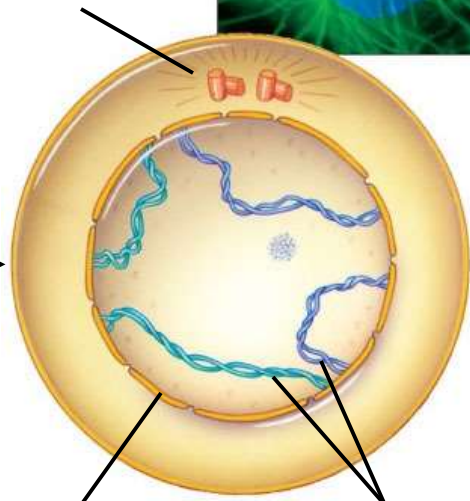
# Mitosis

- 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

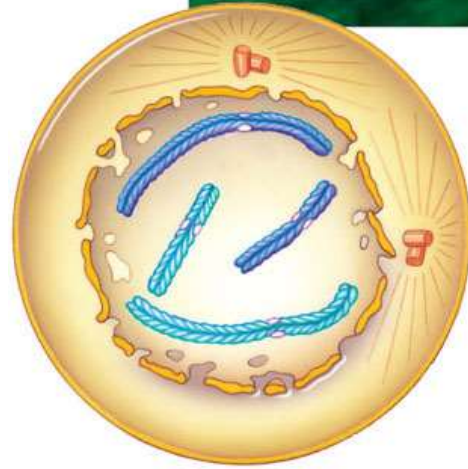
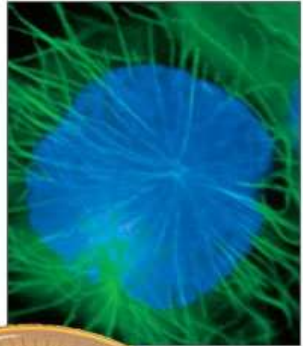
Pair of centrioles



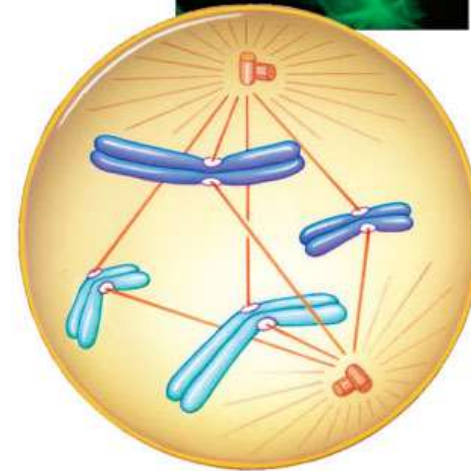
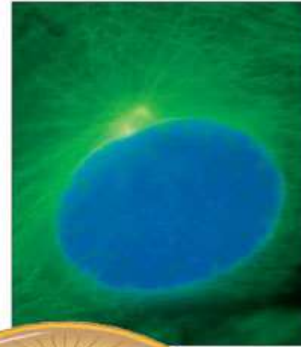
Nuclear envelope

Chromosome

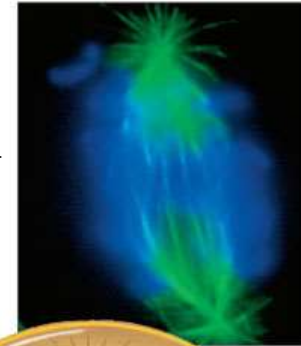
(b) Early Prophase



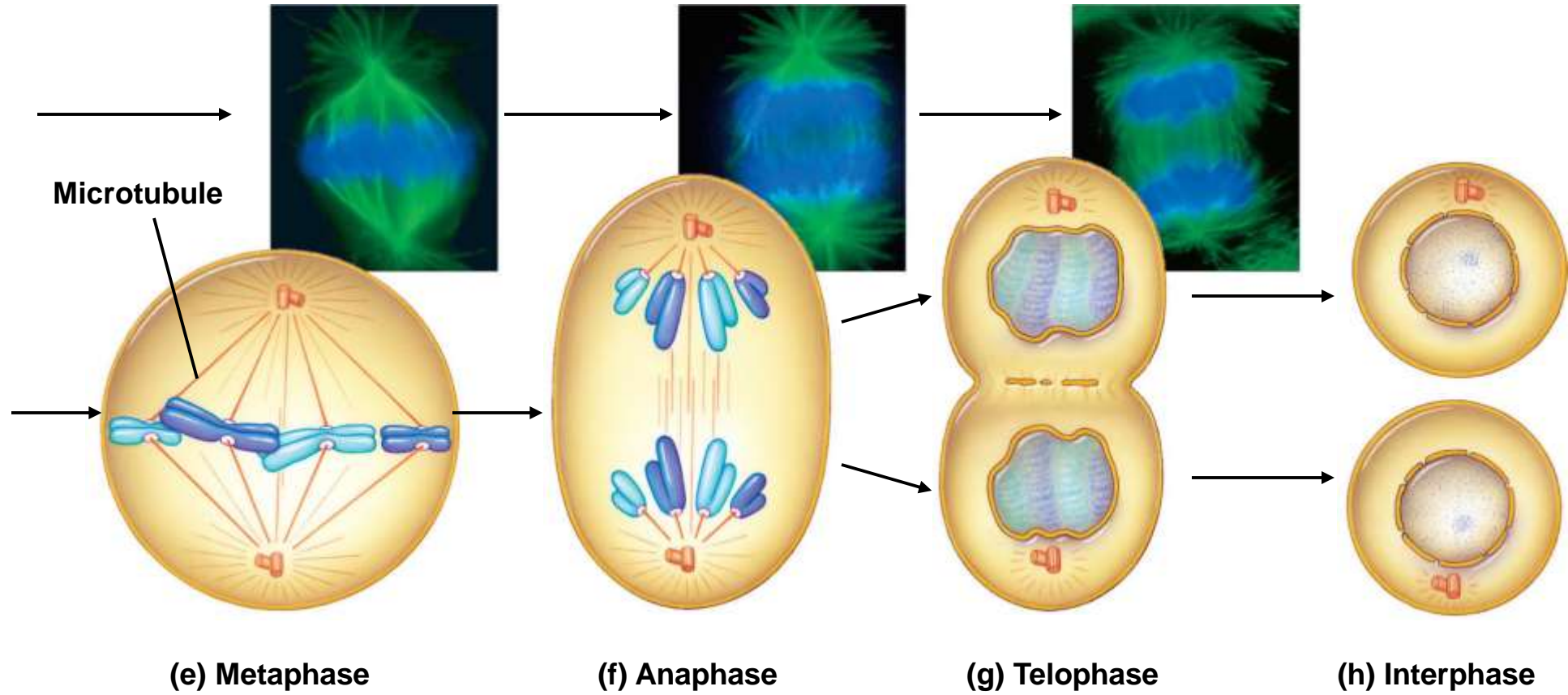
(c) Late Prophase



(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

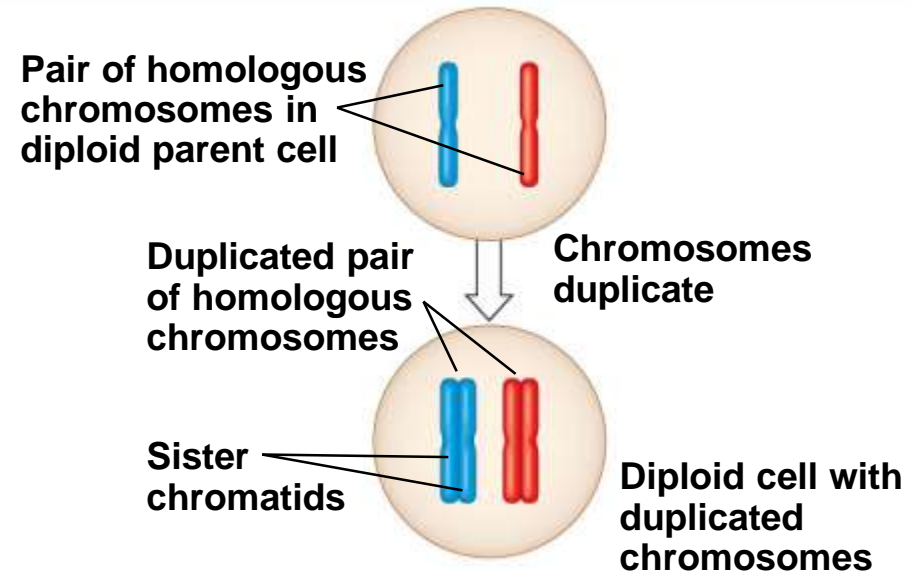


Figure 13.7-2

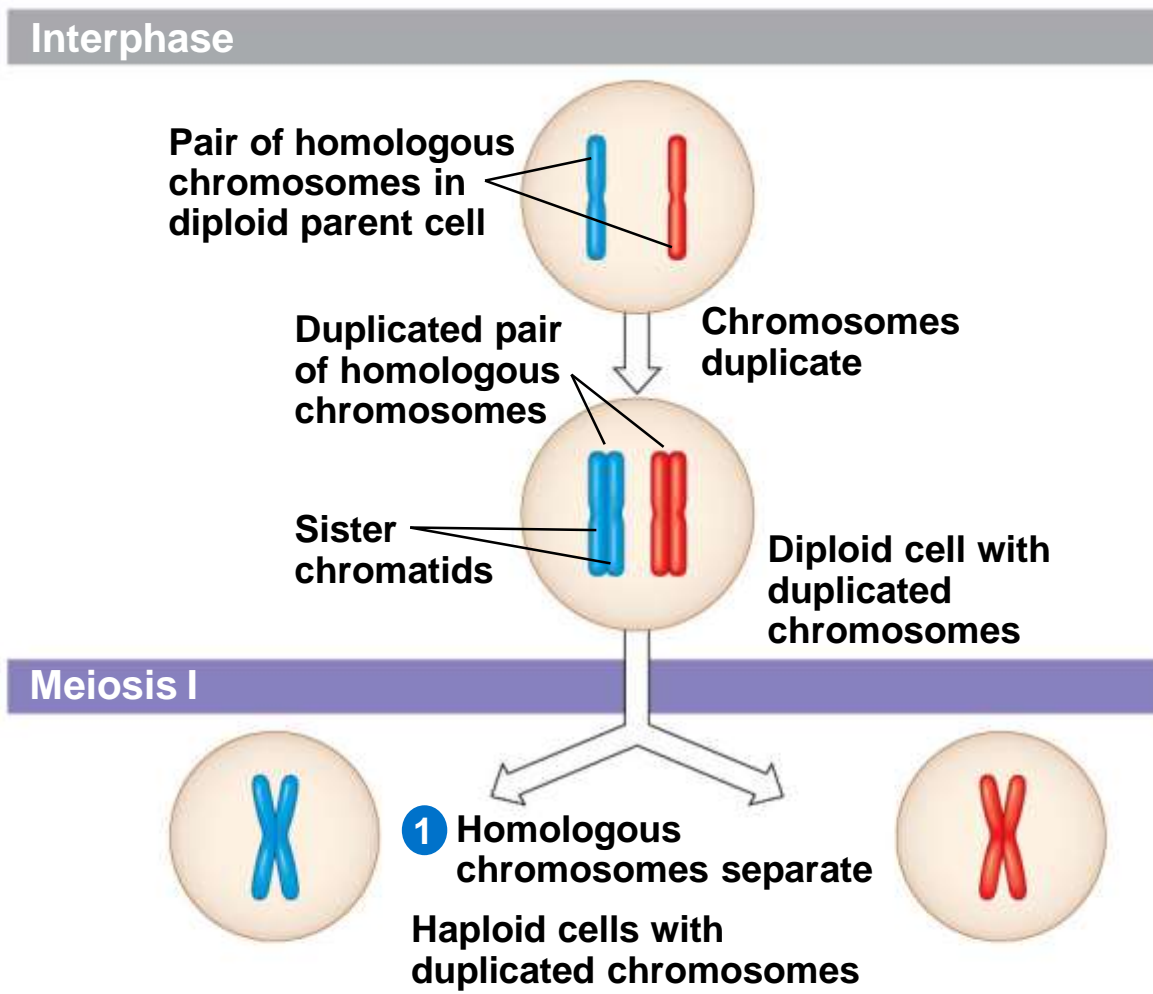
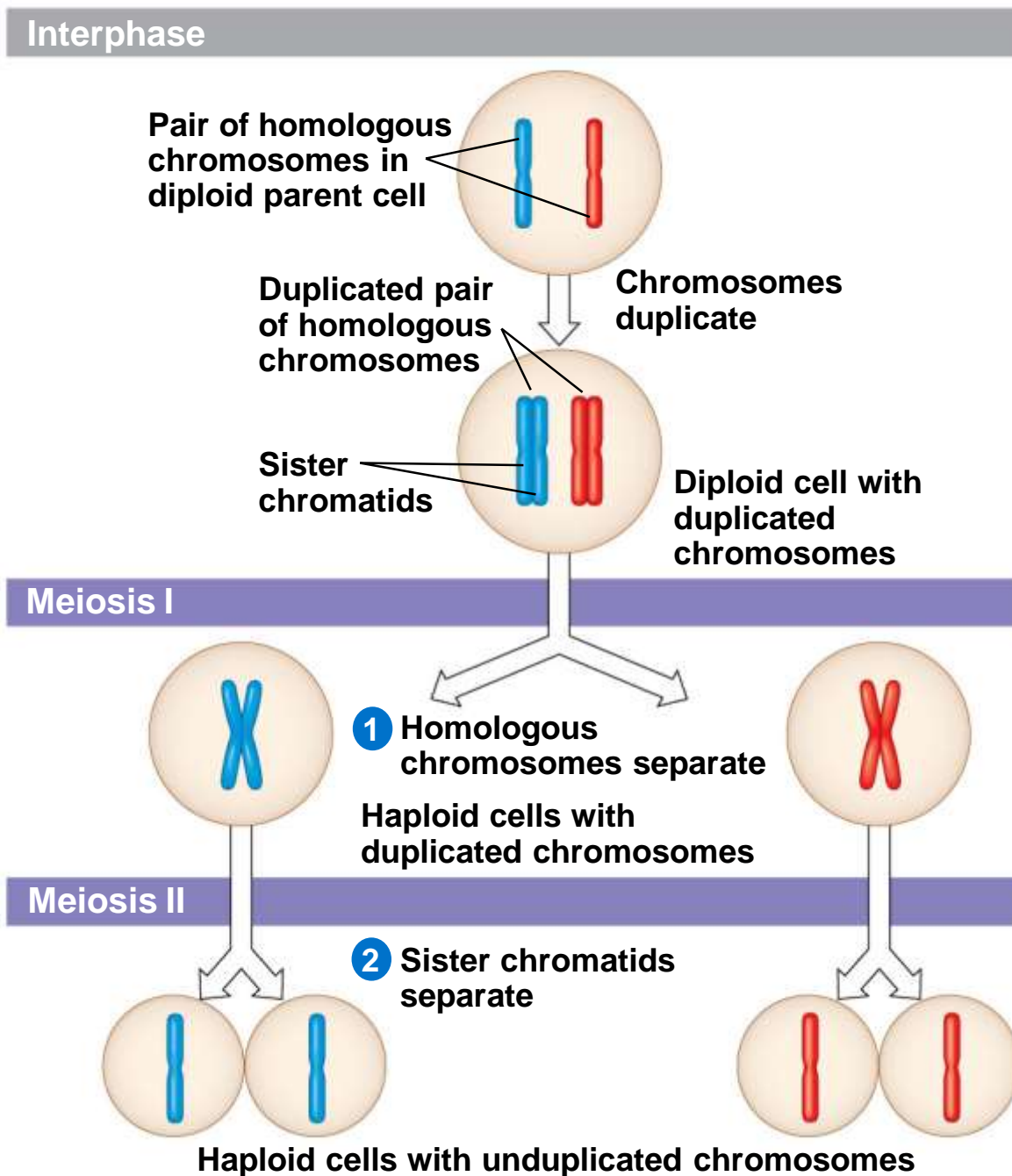
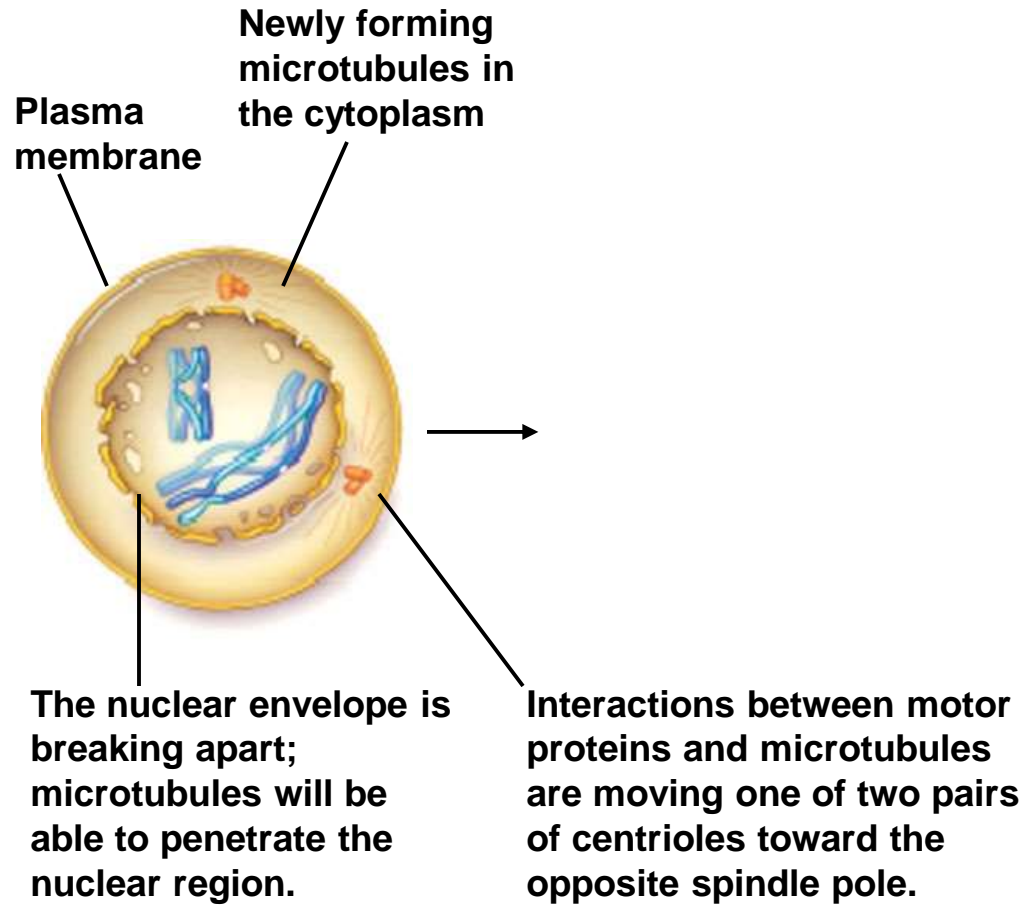


Figure 13.7-3

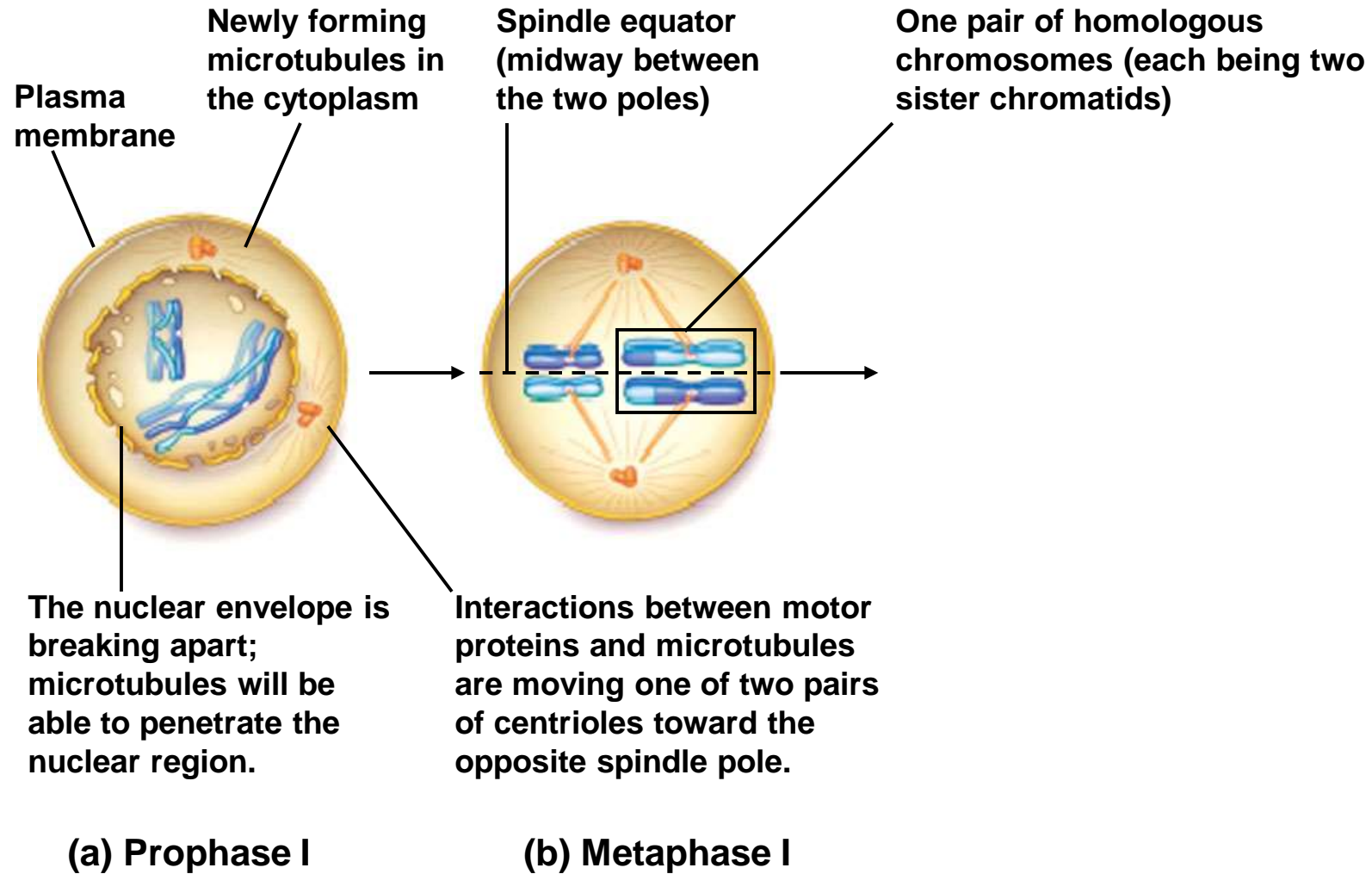


# Meiosis I

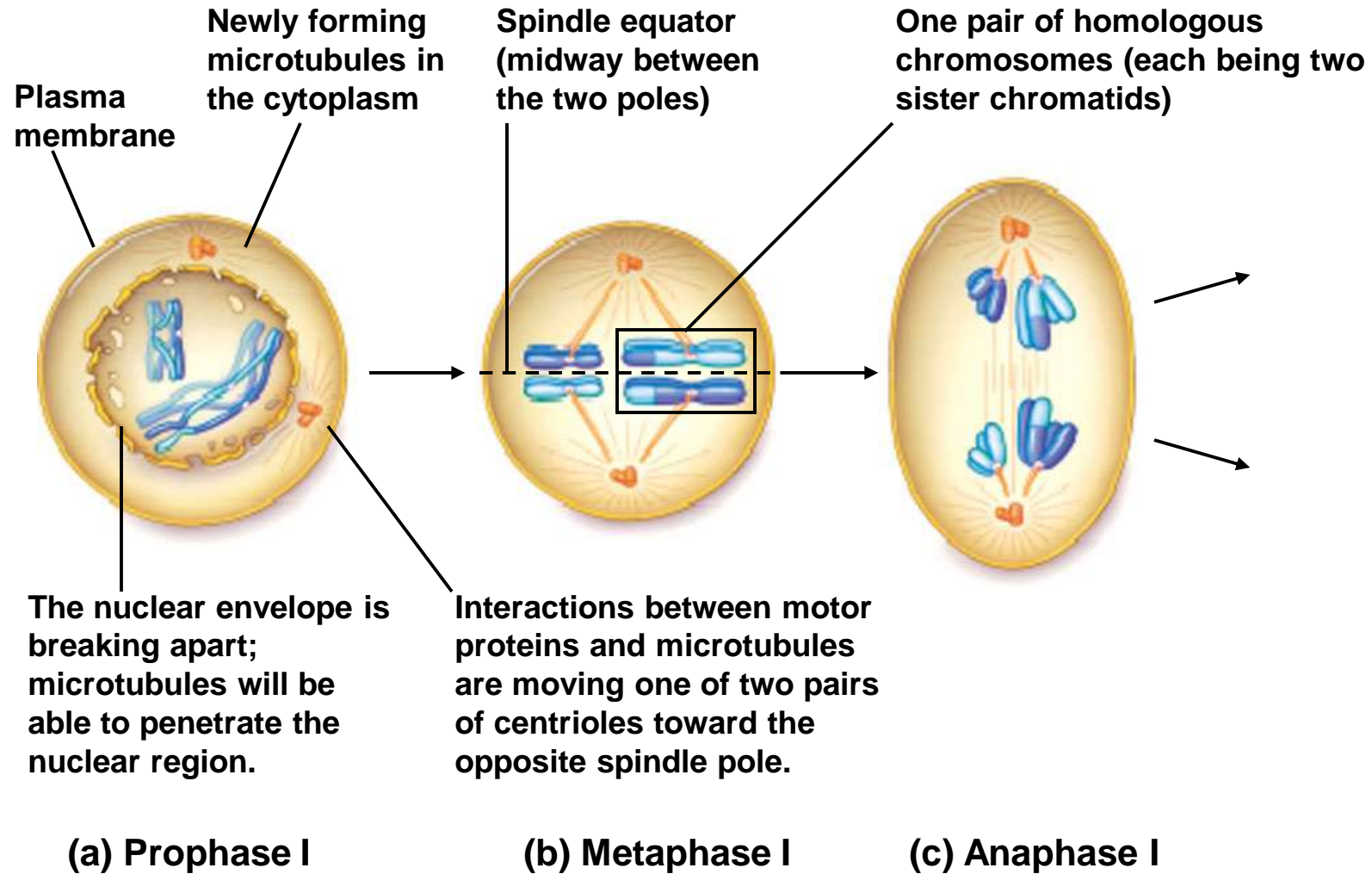


**(a) Prophase I**

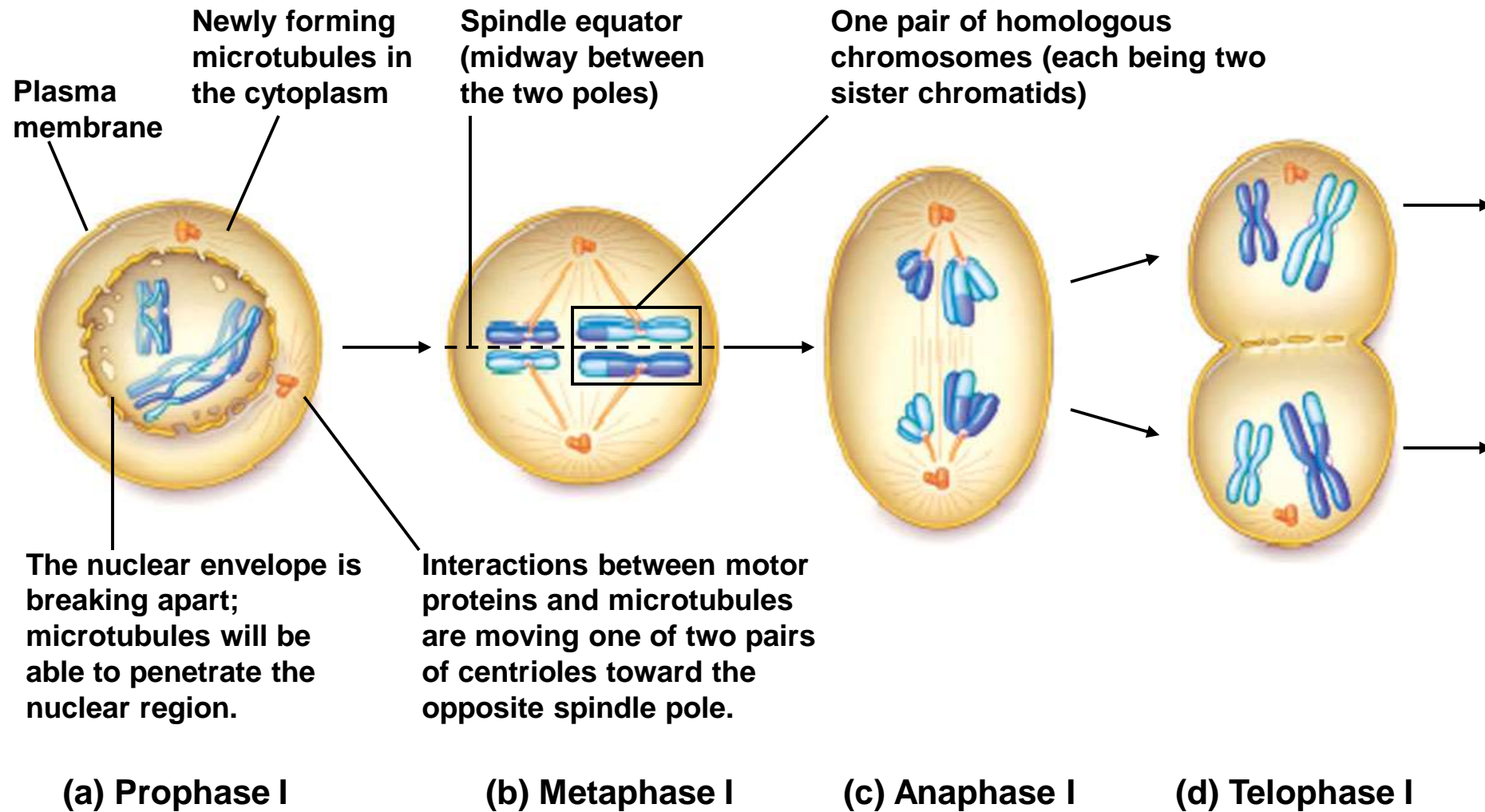
# Meiosis I



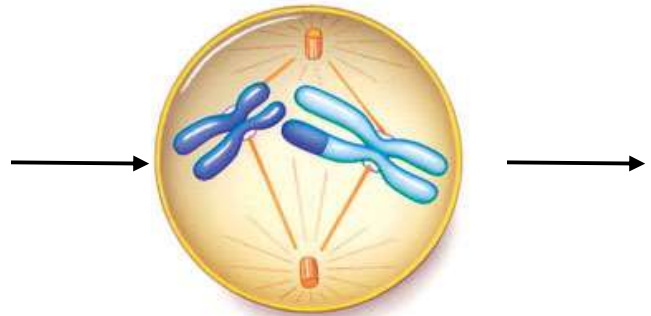
# Meiosis I



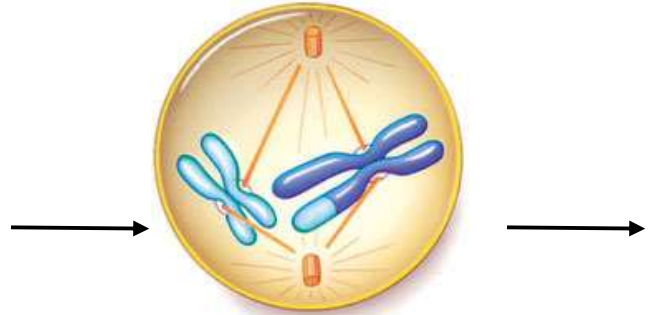
# Meiosis I



# Meiosis II

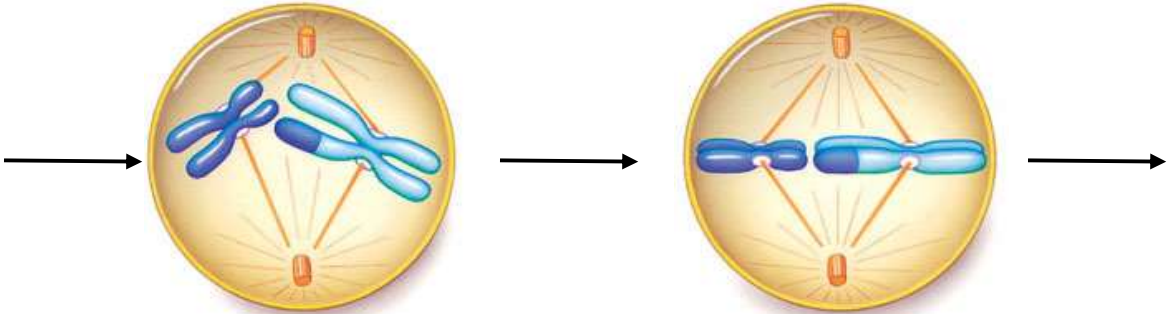


*There is no DNA replication  
between the two nuclear divisions.*

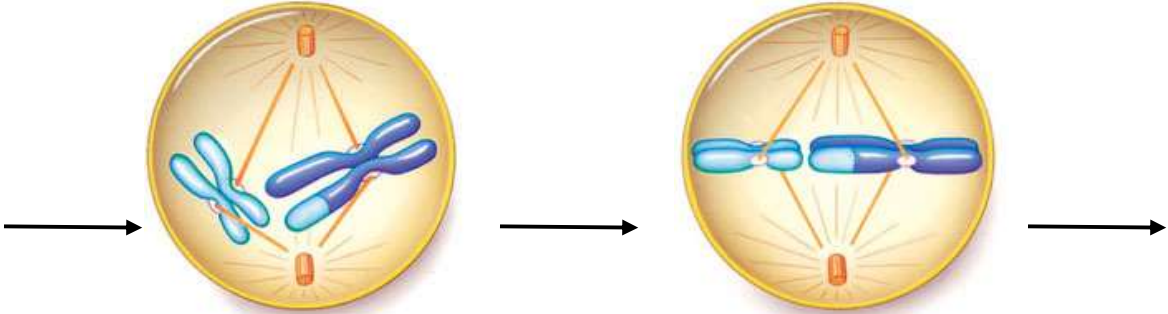


**(e) Prophase II**

# Meiosis II



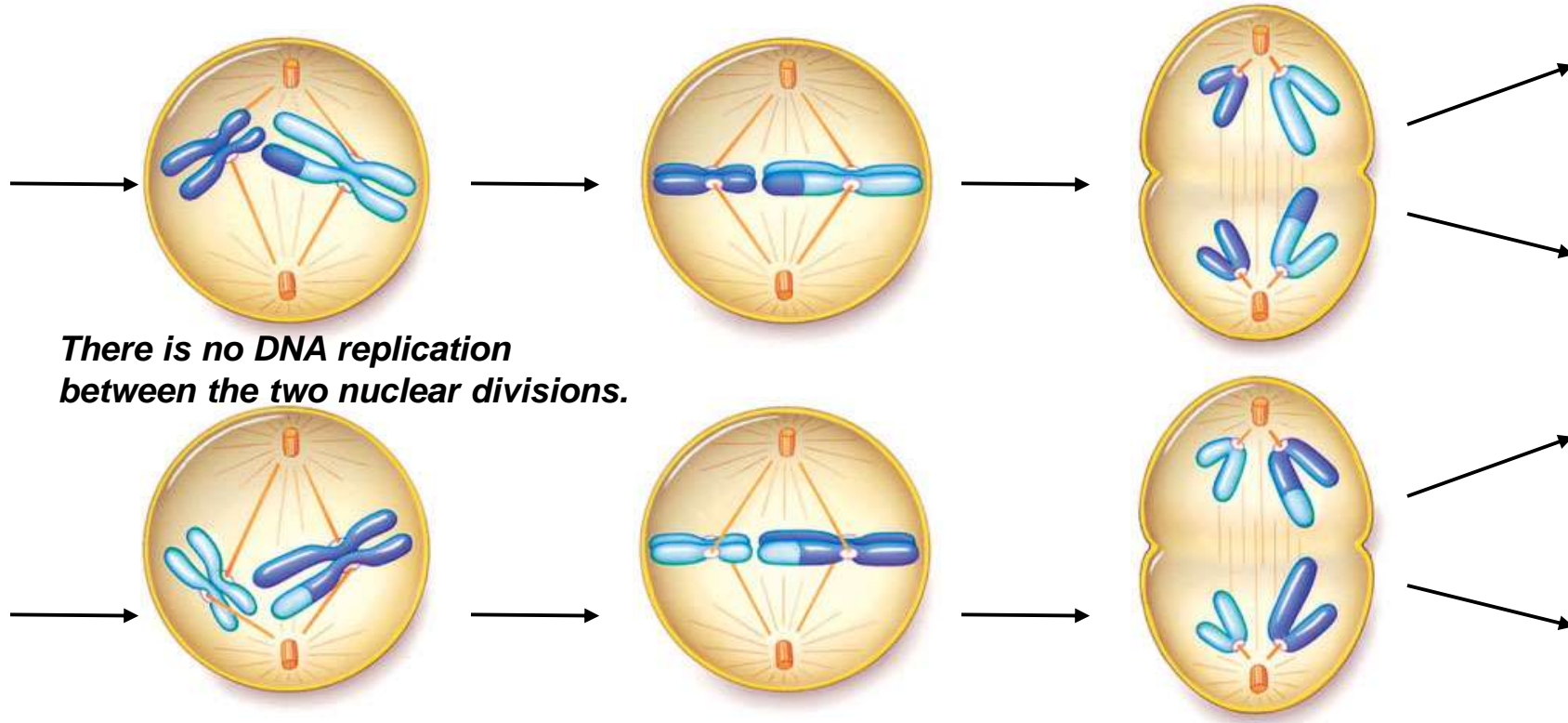
*There is no DNA replication between the two nuclear divisions.*



**(e) Prophase II**

**(f) Metaphase II**

# Meiosis II



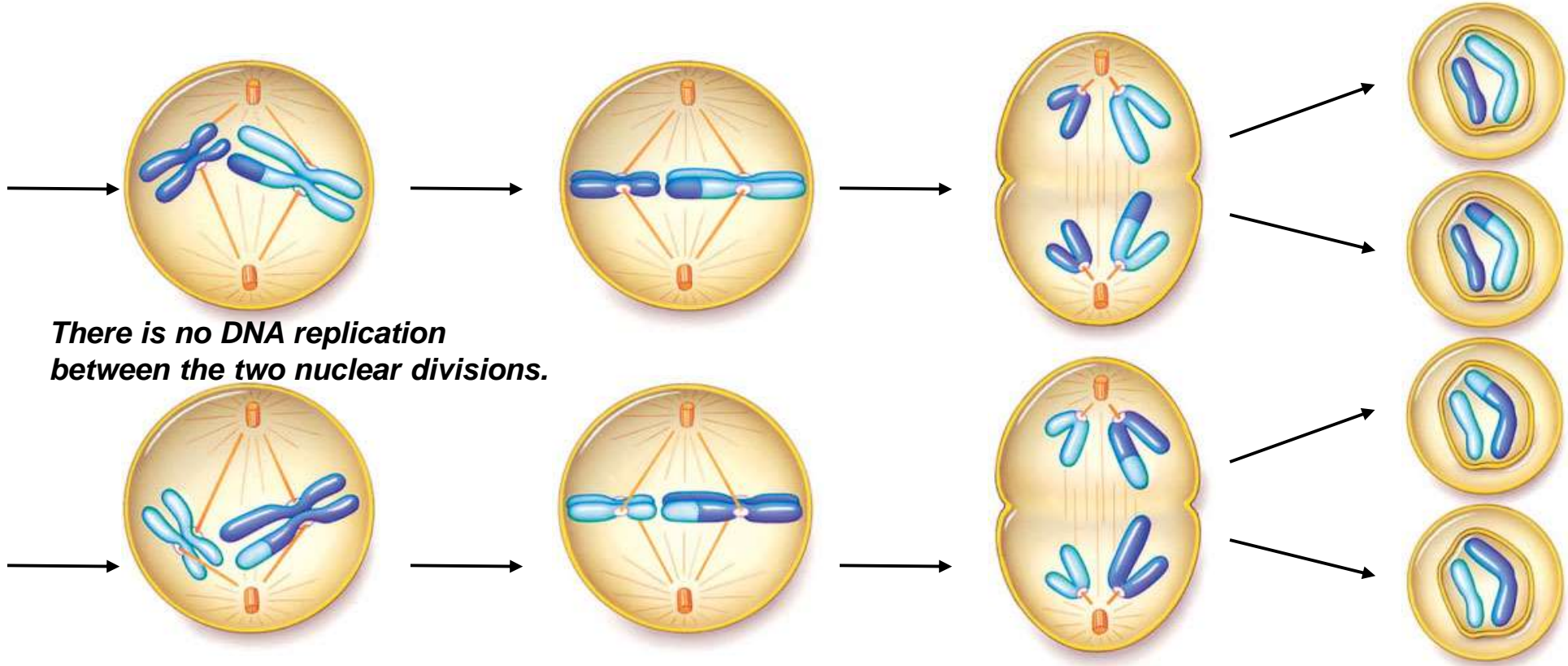
*There is no DNA replication between the two nuclear divisions.*

**(e) Prophase II**

**(f) Metaphase II**

**(g) Anaphase II**

# Meiosis II



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

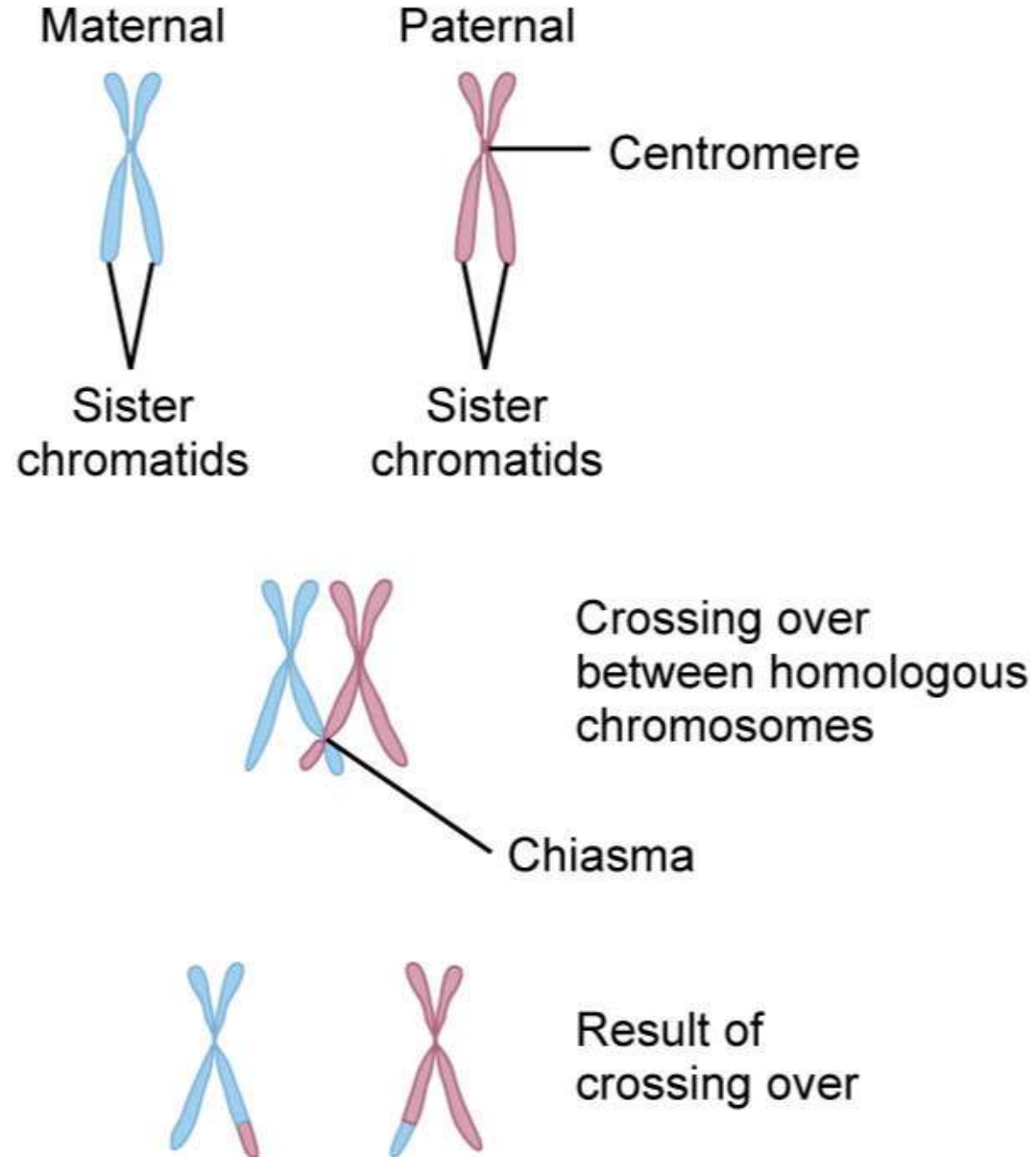
**(e) Prophase II**

**(f) Metaphase II**

**(g) Anaphase II**

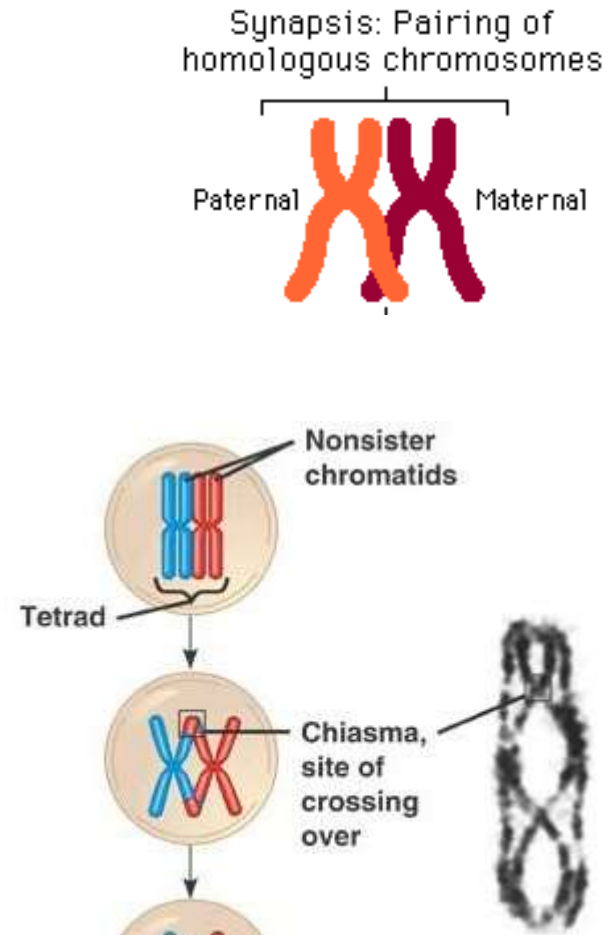
**(h) Telophase II**

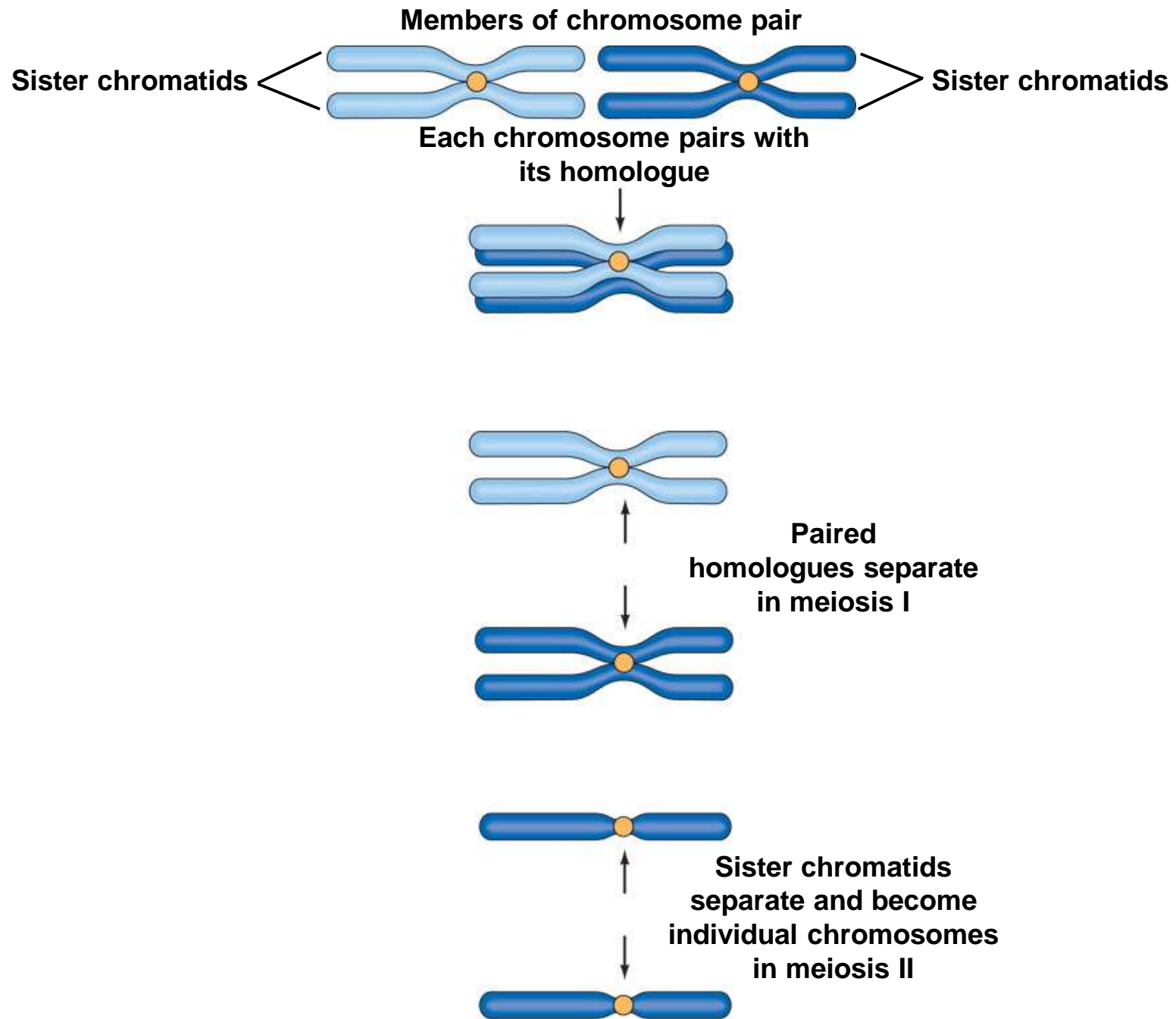
# Homologous chromosomes



# 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

