GENETICS

Meiosis

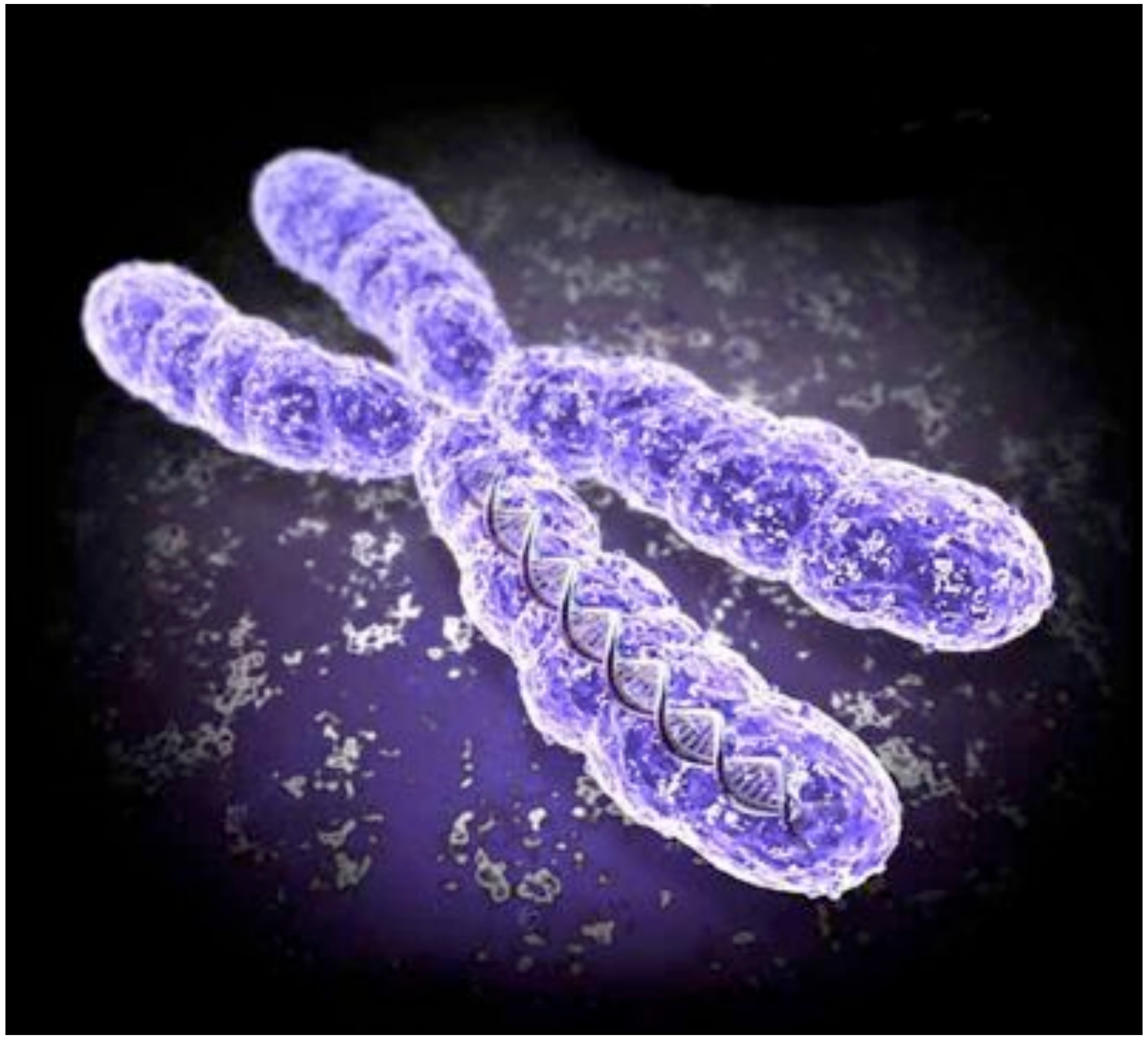
PREFACE

- **Genetics-** is the study of heredity and hereditary variation.
  - **Heredity-** aka inheritance, is the transmission of traits one generation to another.
    - Innate to heredity is the passing of similar traits and the generation of variation.
- There are important and practical applications that come from our understanding of heredity and hereditary variation.
- **Your understanding of genetics requires a comprehensive understanding of meiosis, a special case of cell division.**

Meiosis

I.

Main Idea: Parents pass chromosomes to their offspring, these chromosomes contain genes (the unit of heredity) that control traits.



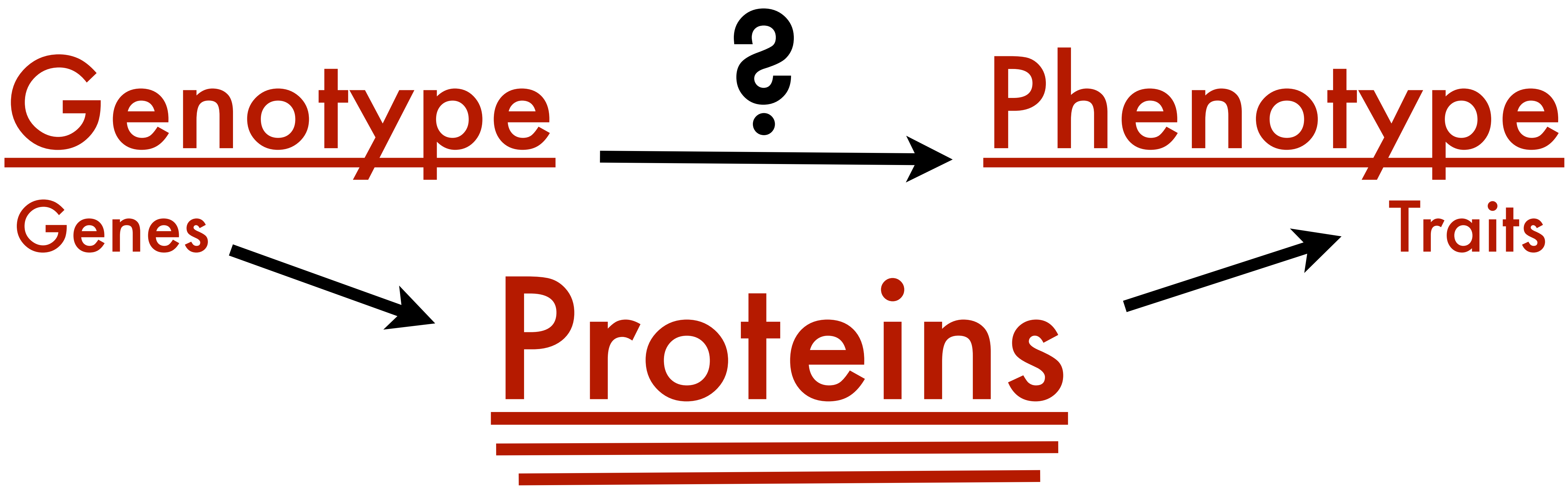
Remember this?

DNA → **Genes** → Proteins → Traits

OK, What exactly is a gene?

- (Basic Definition) A unit of inheritance that controls a phenotypic character.
- (Better Definition) A nucleotide sequence along a molecule of DNA that codes for a protein.
- **(Best Definition) A region of DNA that can be expressed to produce a final functional product that is either a polypeptide or an RNA molecule.**

Review



Proteins are the link between genotypes and phenotypes
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Review

Global Flow of Information

DNA → RNA → Protein

- The flow of genetic information involves two processes.
  - Transcription
  - Translation
- Together these two processes represent gene expression.

Inheritance of Genes

- **Gametes-** are the vehicles that carry genes from one generation to another.
  - Gametes are *haploid*, they carry only 50% a parent's genes (these genes are carried in 1 complete set of chromosomes).
    - **Germ cells** are diploid cells (they contain 2 copies of every chromosome) that undergo a special type of cell division that produces unique haploid cells...gametes
  - **Fertilization-** the union of gametes brings 1 set of chromosomes from each parent and produces a single unique cell called a *zygote* that is *diploid* .
- The *zygote* undergoes *mitosis* over and over again producing trillions of **somatic cells** (all diploid body cells except germ cells and gametes).

Inheritance of Genes

- **Asexual Reproduction-**
    - one parent
    - no fusion of gametes
    - daughter cells get all of parent's genes
    - daughter cells are identical* to each other and parent cell

} Mitosis

Binary Fission

* Unless of course a mutation occurs!

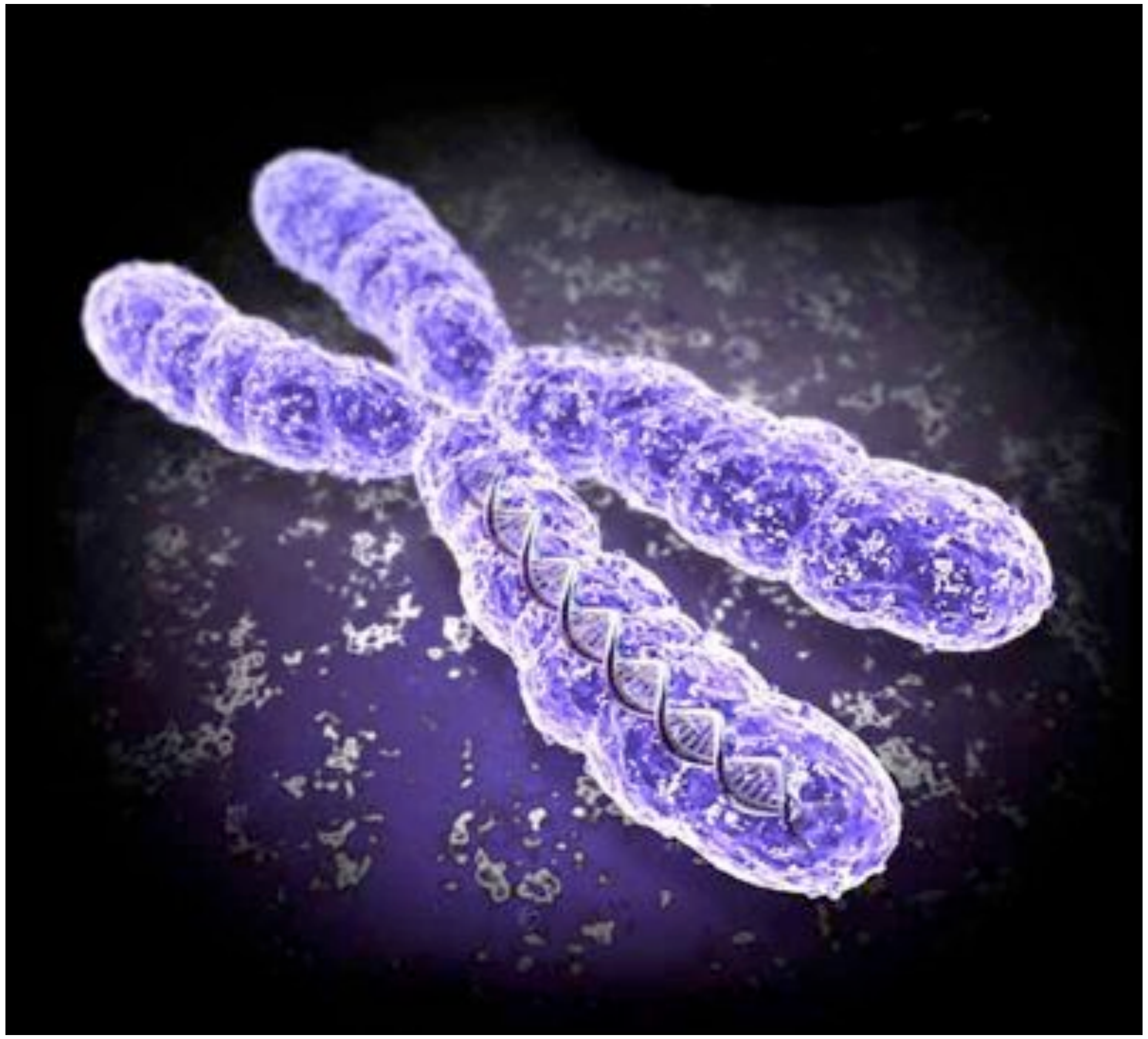
Inheritance of Genes

- **Sexual Reproduction-**
  - two parents
  - fusion of gametes
  - daughter cells get half of parent's genes
  - daughter cells are completely unique (one of kind cells)

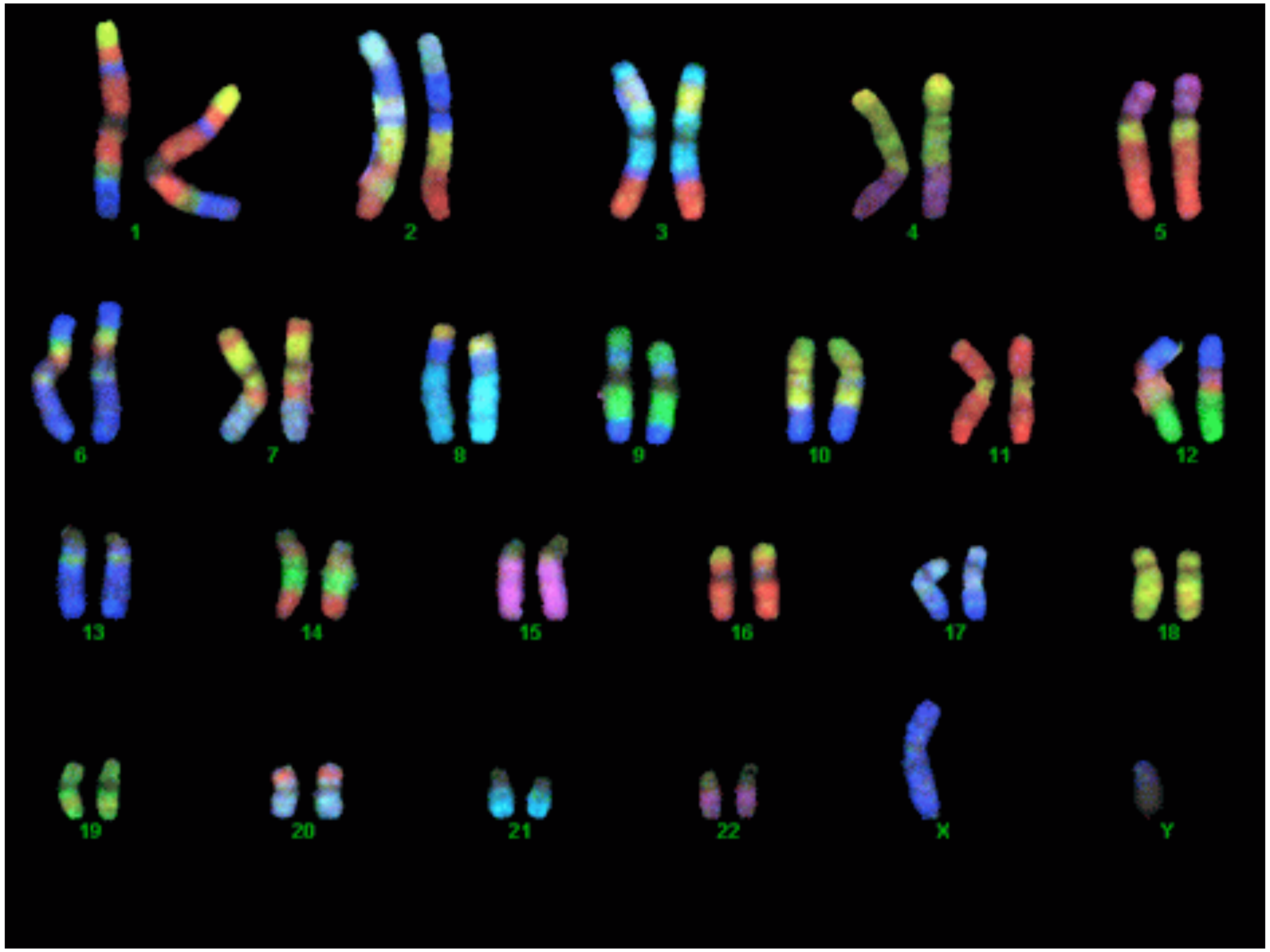
Meiosis

II.

Main Idea: Fertilization and meiosis alternate in life cycles regardless of variations in life cycles.

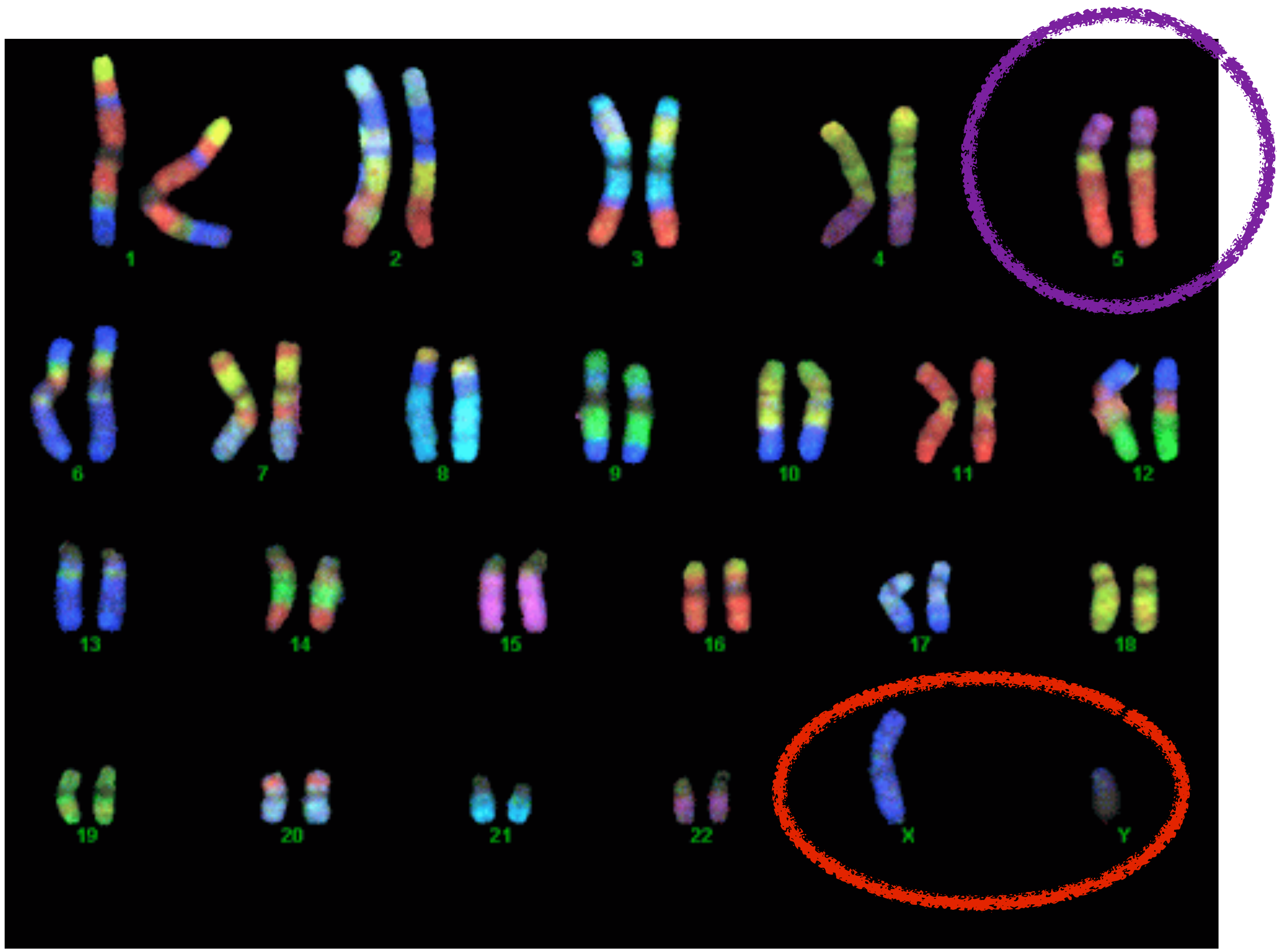


Closer look at Chromosomes



This is a **karyotype**, an ordered display of chromosomes. Notice the 23 types (pairs) are numbered and arranged from long to short. The position of the centromere and the colored banded patterns are also used when arranging the chromosomes.

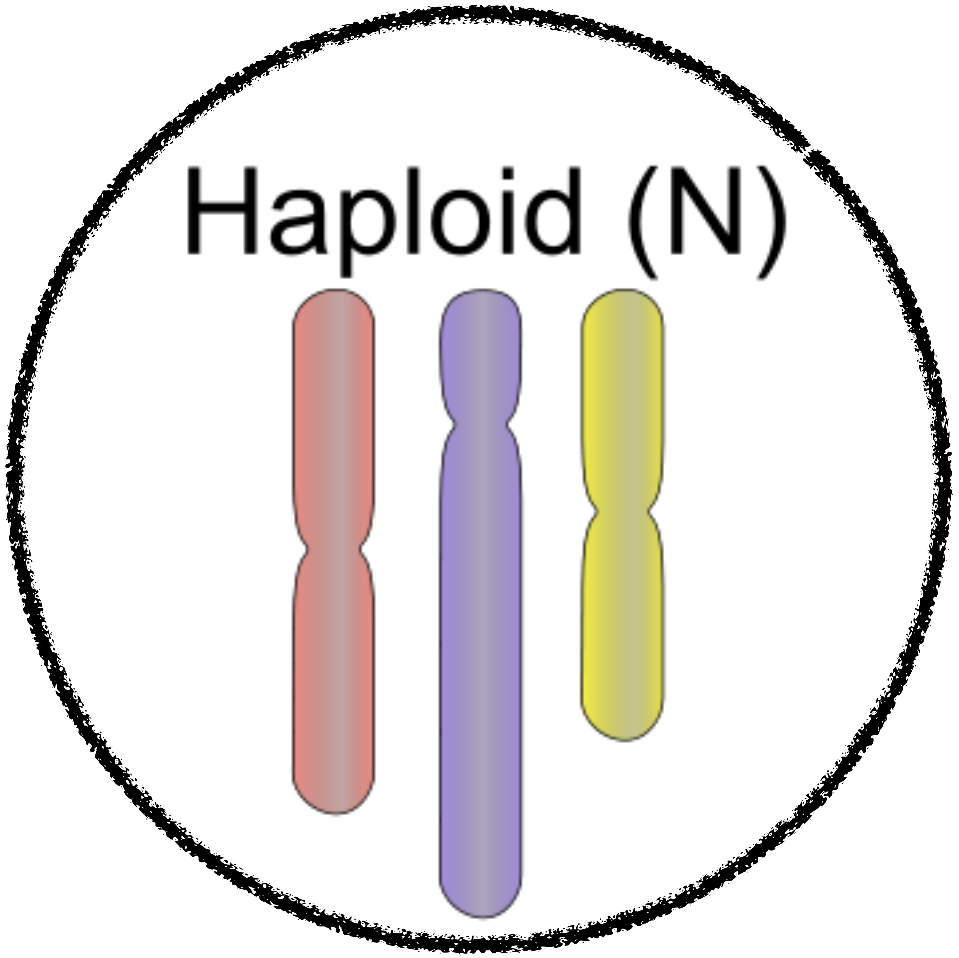
Closer look at Chromosomes



Homologous Chromosomes- chromosomes with same, length, centromere position and they carry genes controlling the same traits

Autosomes- the first 22 pairs chromosomes (all except sex chromosomes)

Sex Chromosomes- the X and Y chromosomes they are 23rd pair and they determine the sex of the offspring (XX=girl) (XY=boy)



Diploid

-symbol *2n*

-two sets of chromosomes (maternal & paternal sets)

-includes almost all cells

-humans *2n*=46

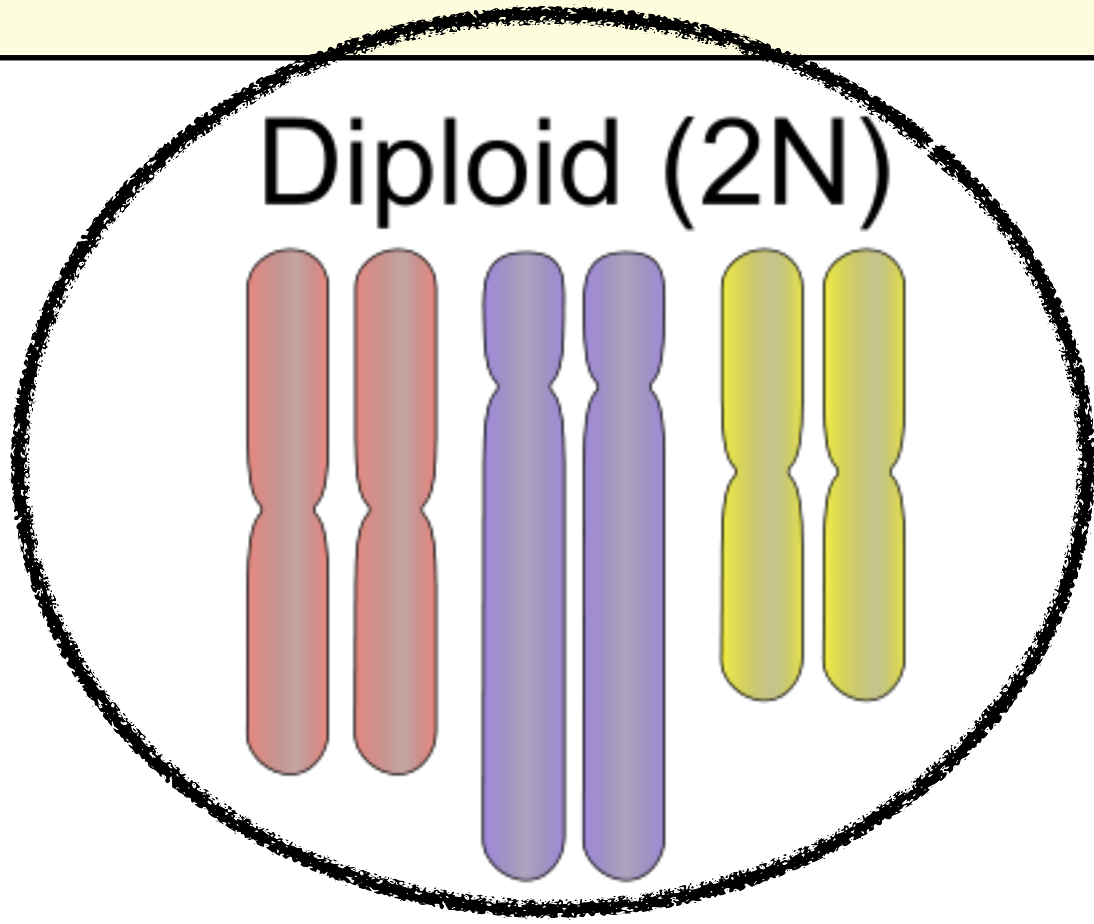
Haploid

-symbol *n*

-one set of chromosomes (maternal & paternal sets)

-only sperm and eggs

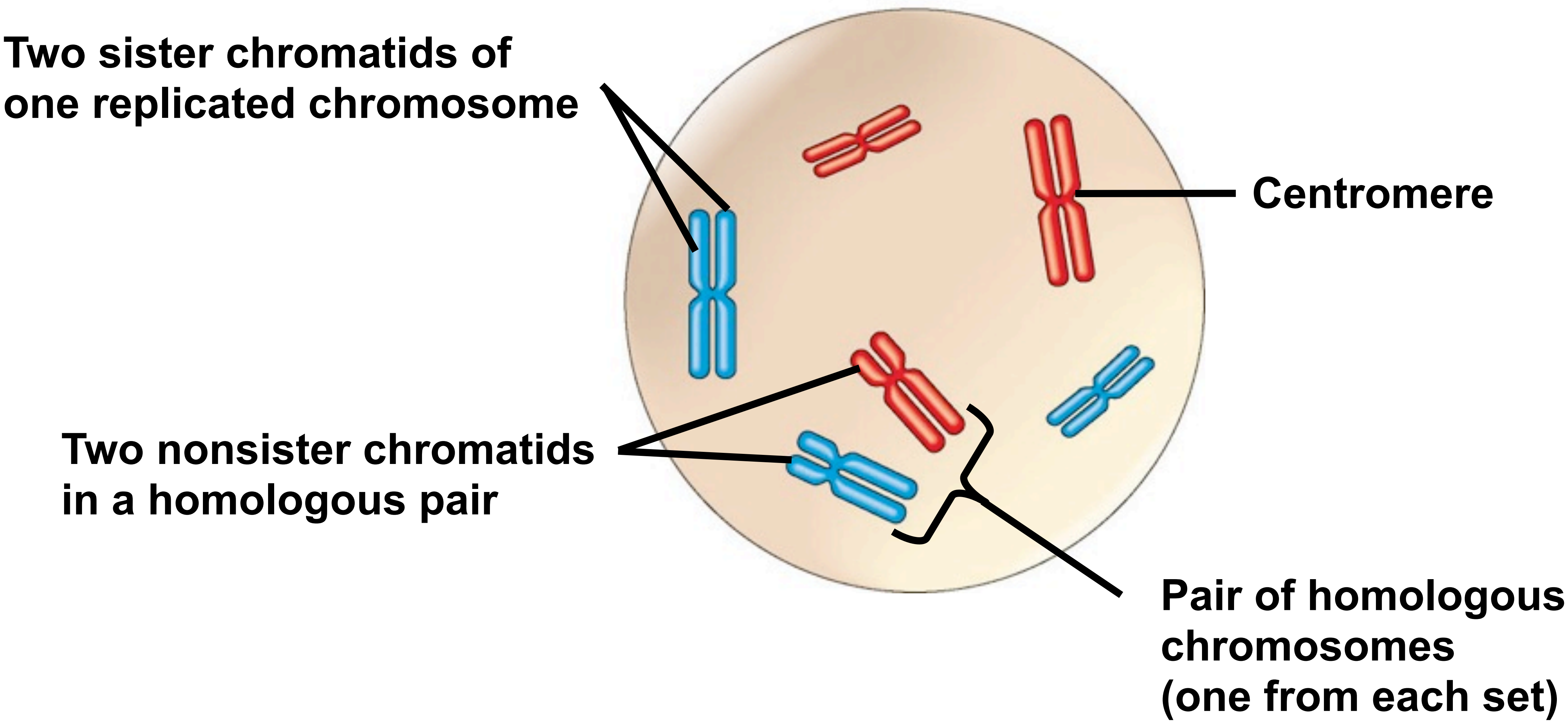
-humans *n*=23



Describing Chromosomes

Key

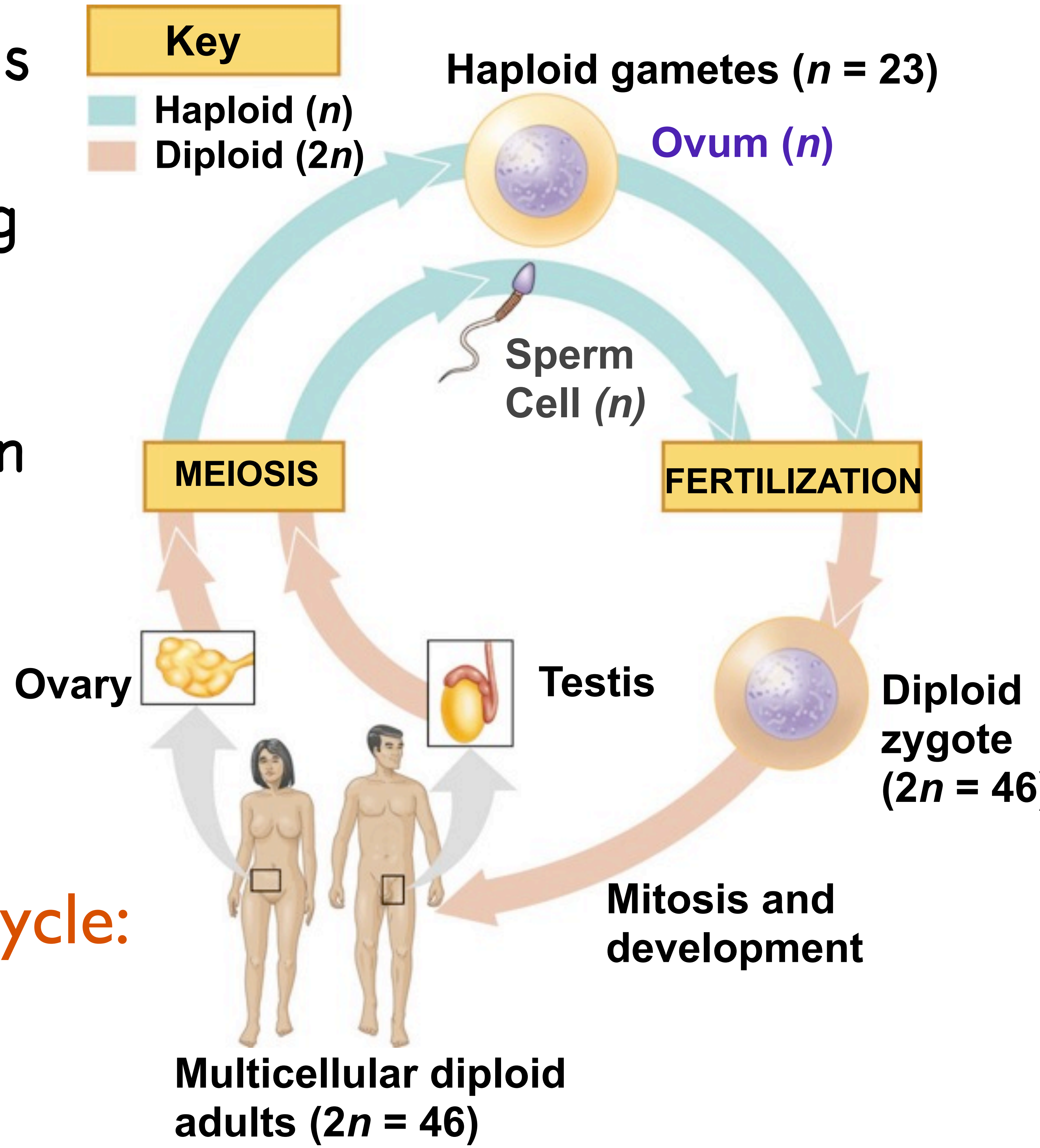
$2n = 6$ — Maternal set of chromosomes ($n = 3$); Paternal set of chromosomes ($n = 3$)



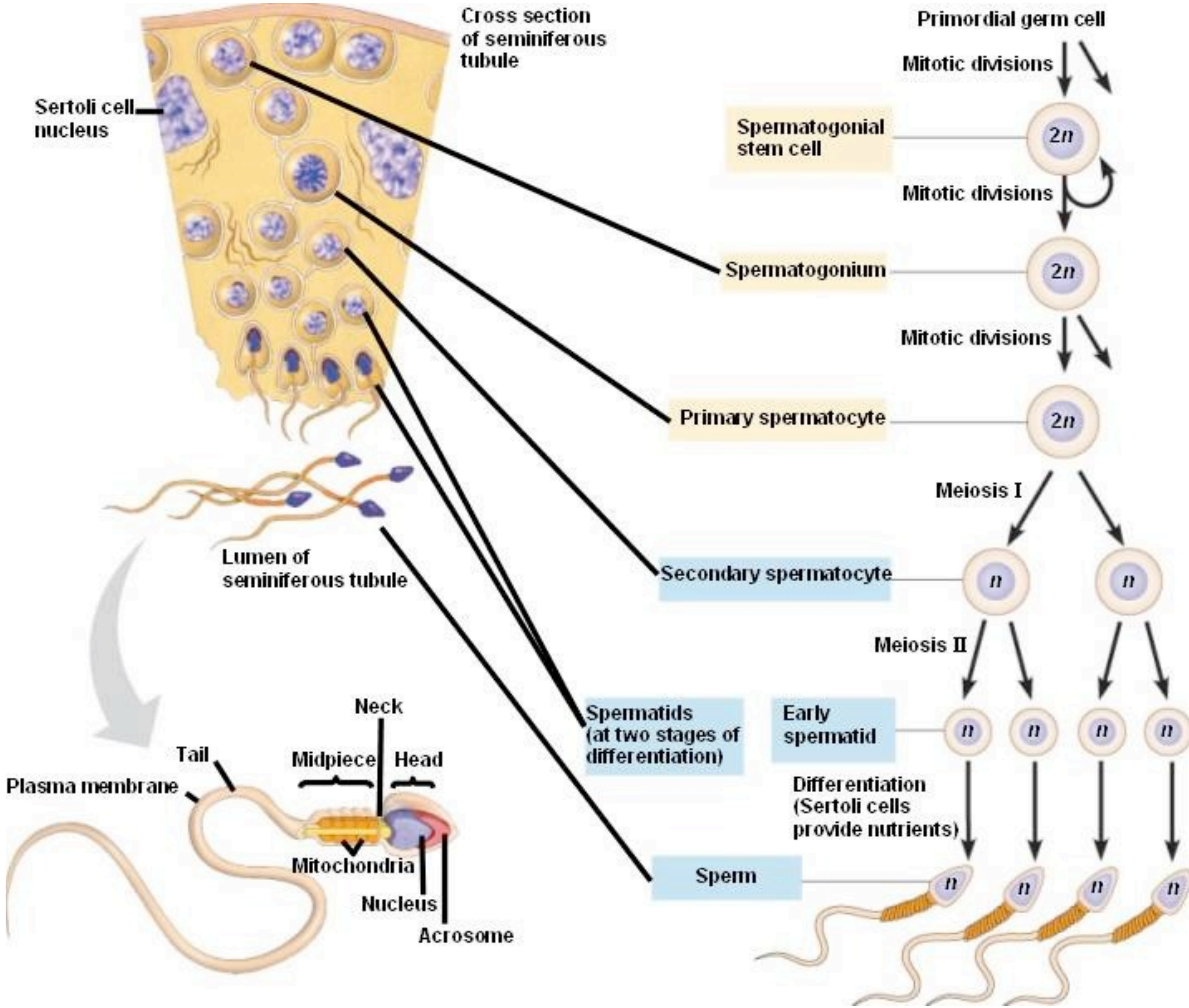
Behavior of Chromosomes

Fertilization and meiosis alternate in sexual life cycles, thus maintaining a constant number of chromosomes in each species from generation to generation.

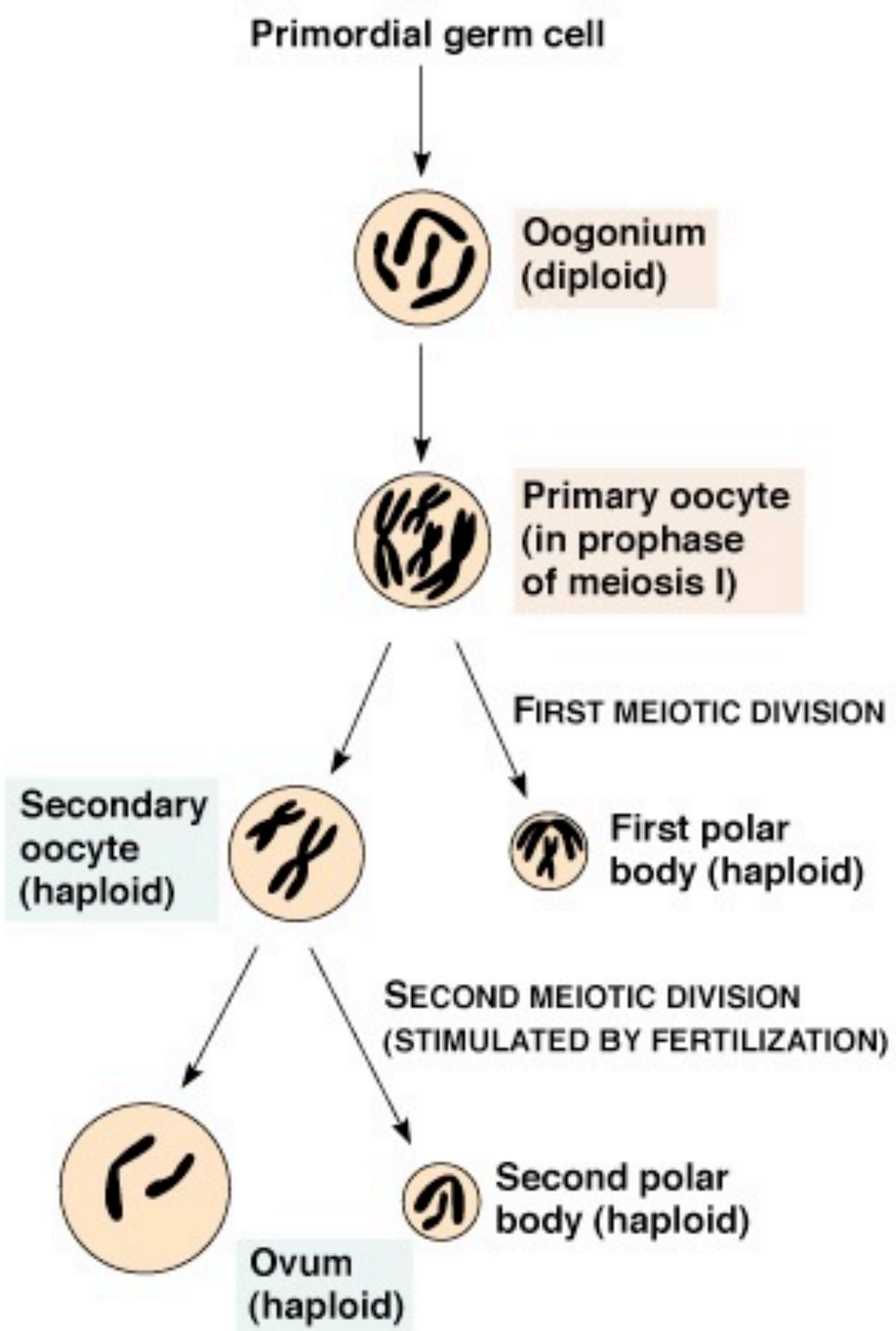
Human Life Cycle:

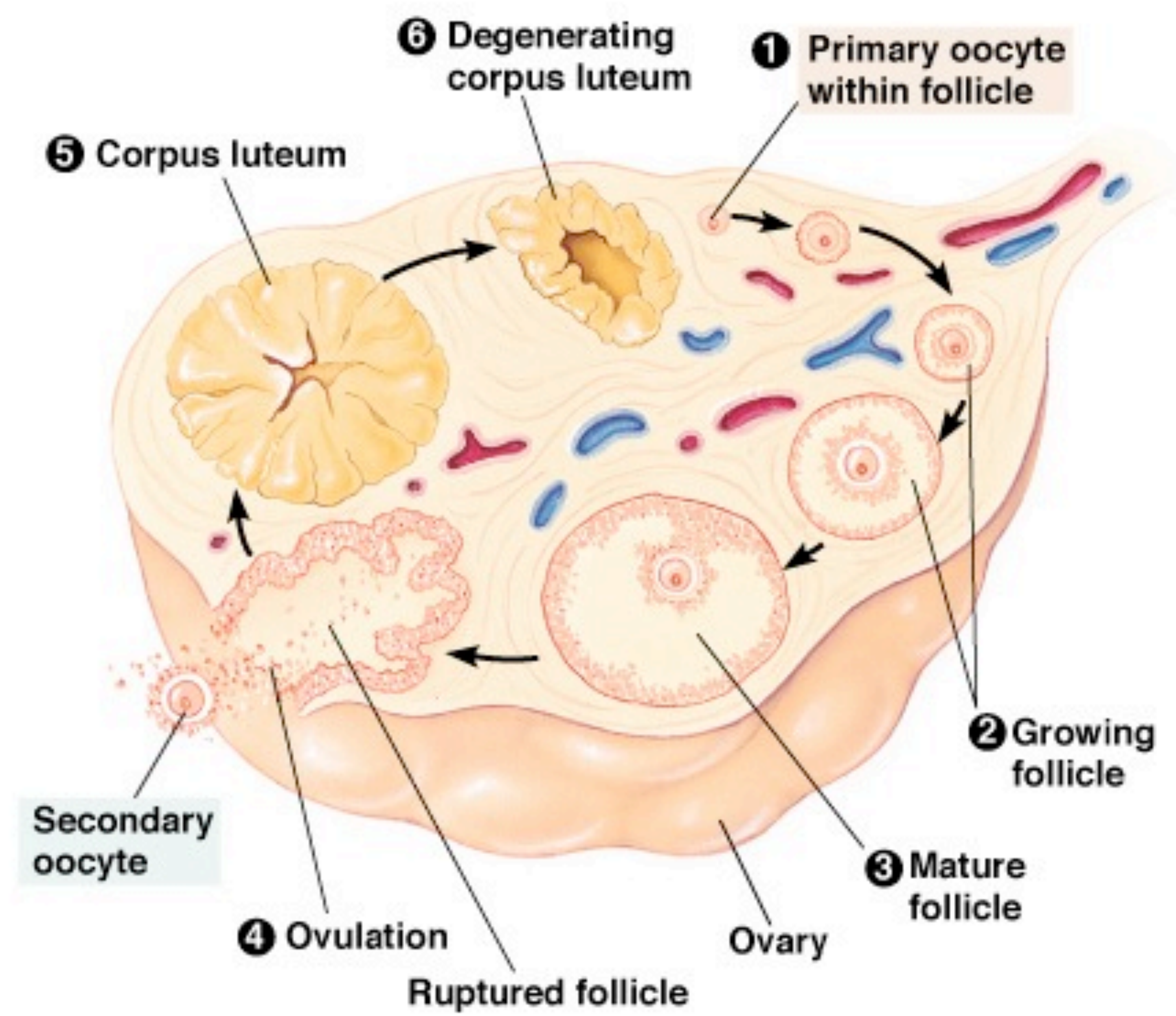


Review: Spermatogenesis



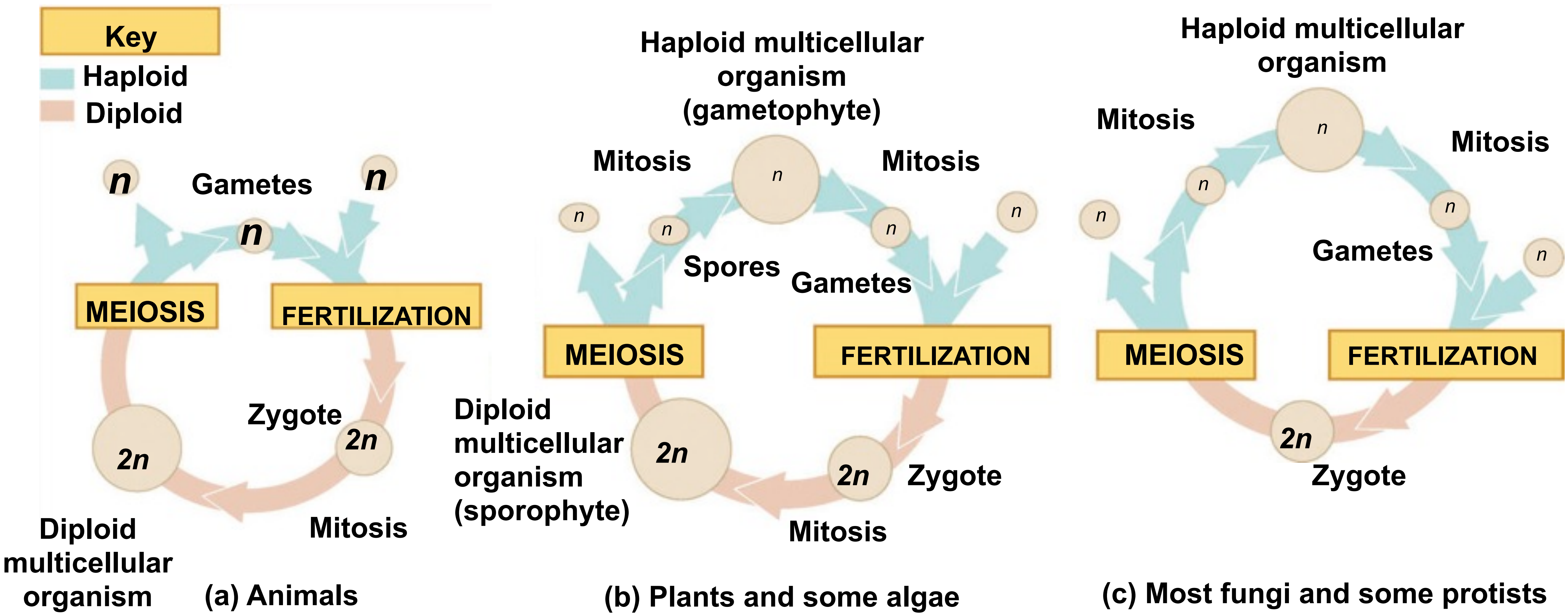
Review: Oogenesis



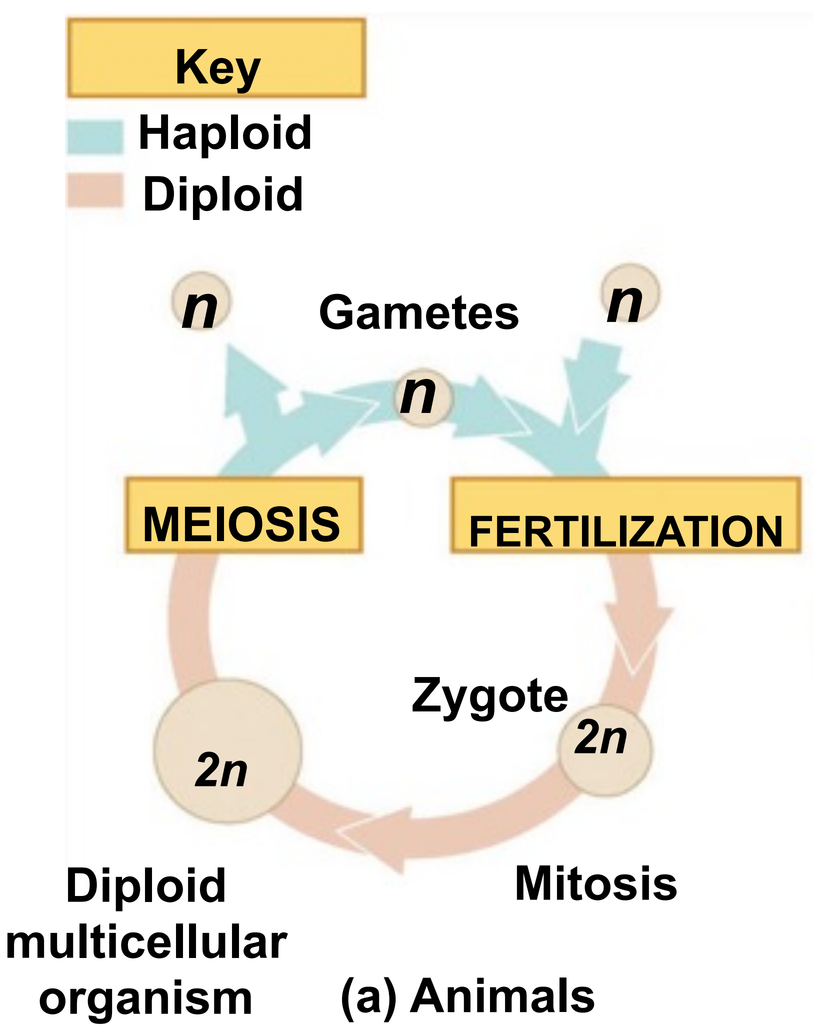


Variation of Sexual Life Cycles

- Alternation of fertilization and meiosis is common in all cycles
  - The timing of these two events differs in each cycle



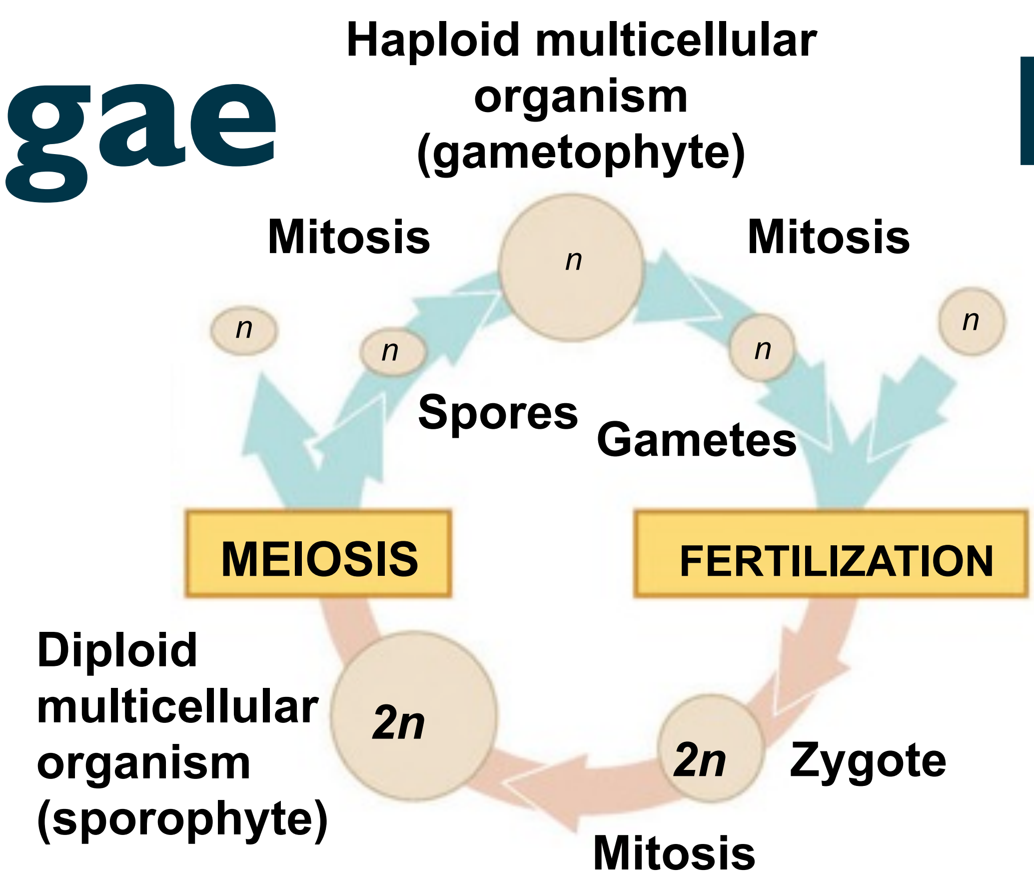
- Meiosis occurs only in germ cells
- Germ cells produce gametes
- Gametes are the only haploid cells
- Zygote grows mitotically into a multicellular diploid organism



Animal Life Cycle

- Exhibit *alternation of generations*-
  - Gametophyte- a multicellular haploid organism/stage
    - produces haploid spores
  - Sporophyte- a multicellular diploid organism/stage
    - produces haploid gametes

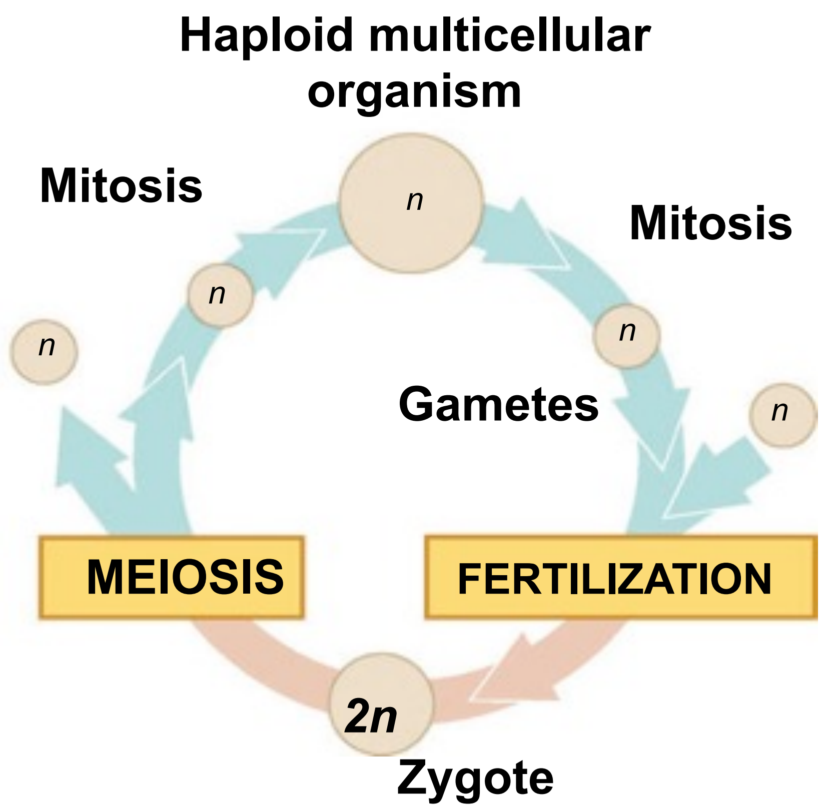
Plant/Algae Life Cycle



(b) Plants and some algae

- Meiosis occurs in zygote
- Meiosis does not result in gametes
- Meiosis produces haploid cells that mitotically produce a multicellular haploid organism
- Mitosis produces gametes

Fungi/Protists Life Cycles

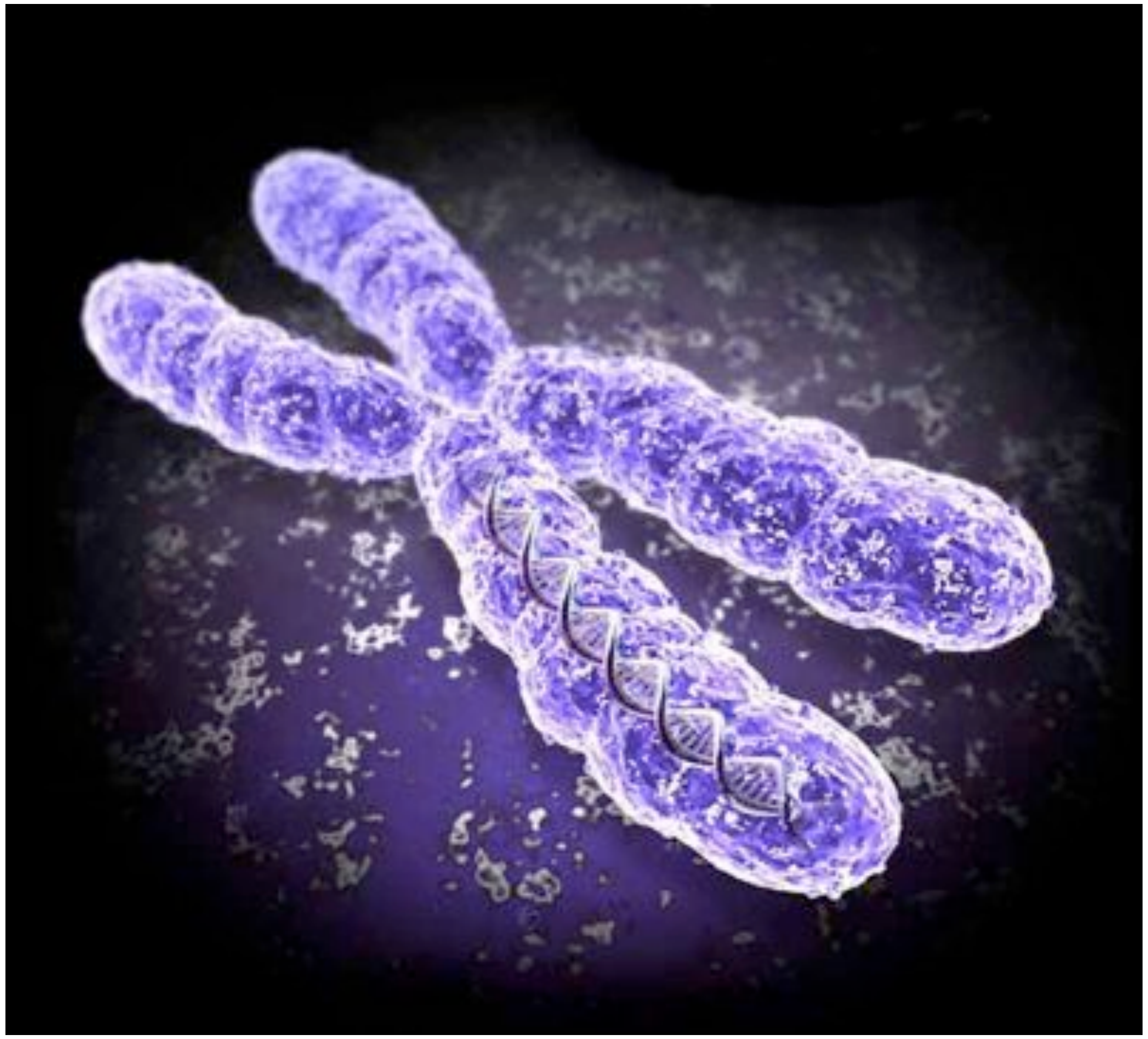


(c) Most fungi and some protists

Meiosis

III.

Main Idea: Meiosis reduces the number of chromosomes from 2 sets (diploid) to 1 set (haploid).

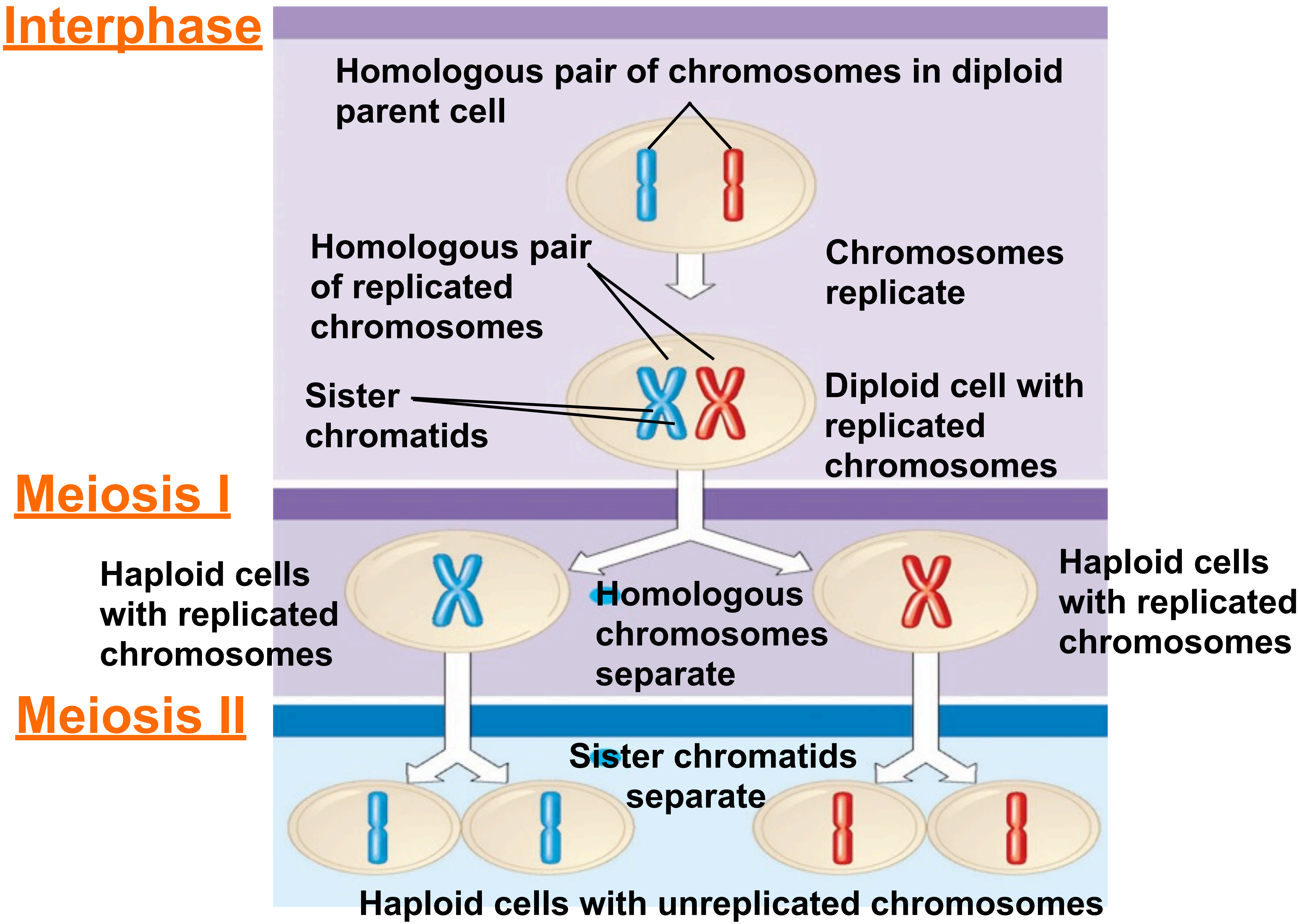


Mitosis and Meiosis

- There are many similarities and important differences between mitosis and meiosis.
  - *Look for comparisons as we examine each stage of meiosis in slides that follow.*
- In the meantime lets begin our examination of meiosis with this fundamental difference between the two processes.

Mitosis	**Meiosis**
Produces 2 identical diploid cells	Produces 4 unique haploid cells

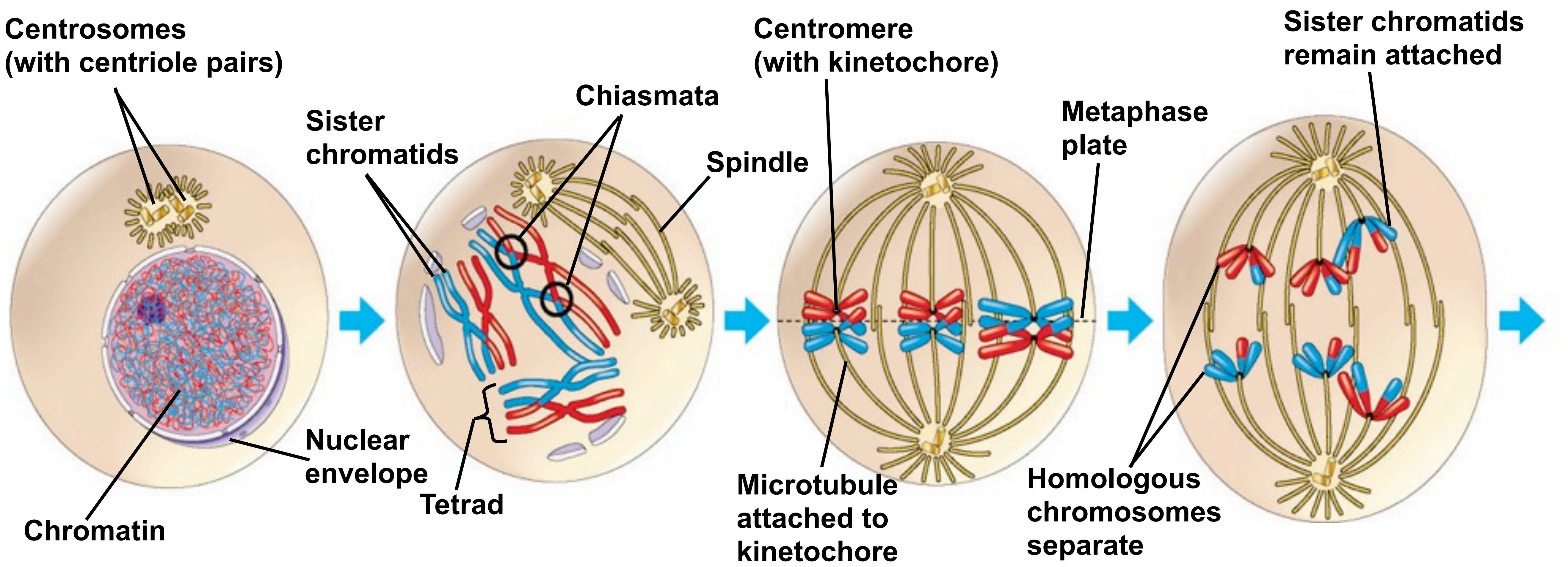
Meiosis Overview



Meiosis I

INTERPHASE	MEIOSIS I: Separates homologous chromosomes		
	PROPHASE I	METAPHASE I	ANAPHASE I

Homologous chromosomes (red and blue) pair and exchange segments; $2n = 6$ in this example

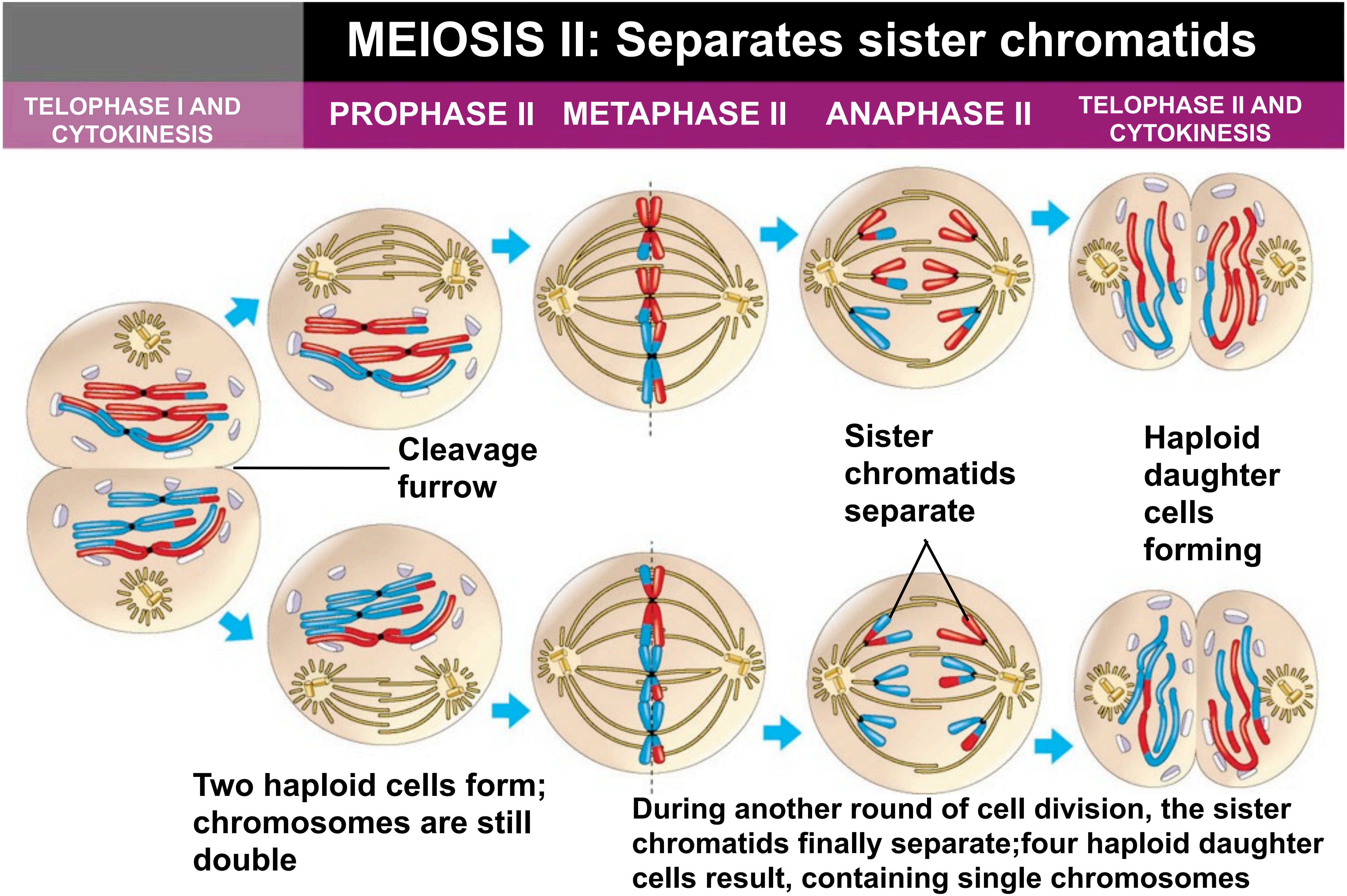


Chromosomes duplicate

Tetrads line up

Pairs of homologous chromosomes split up

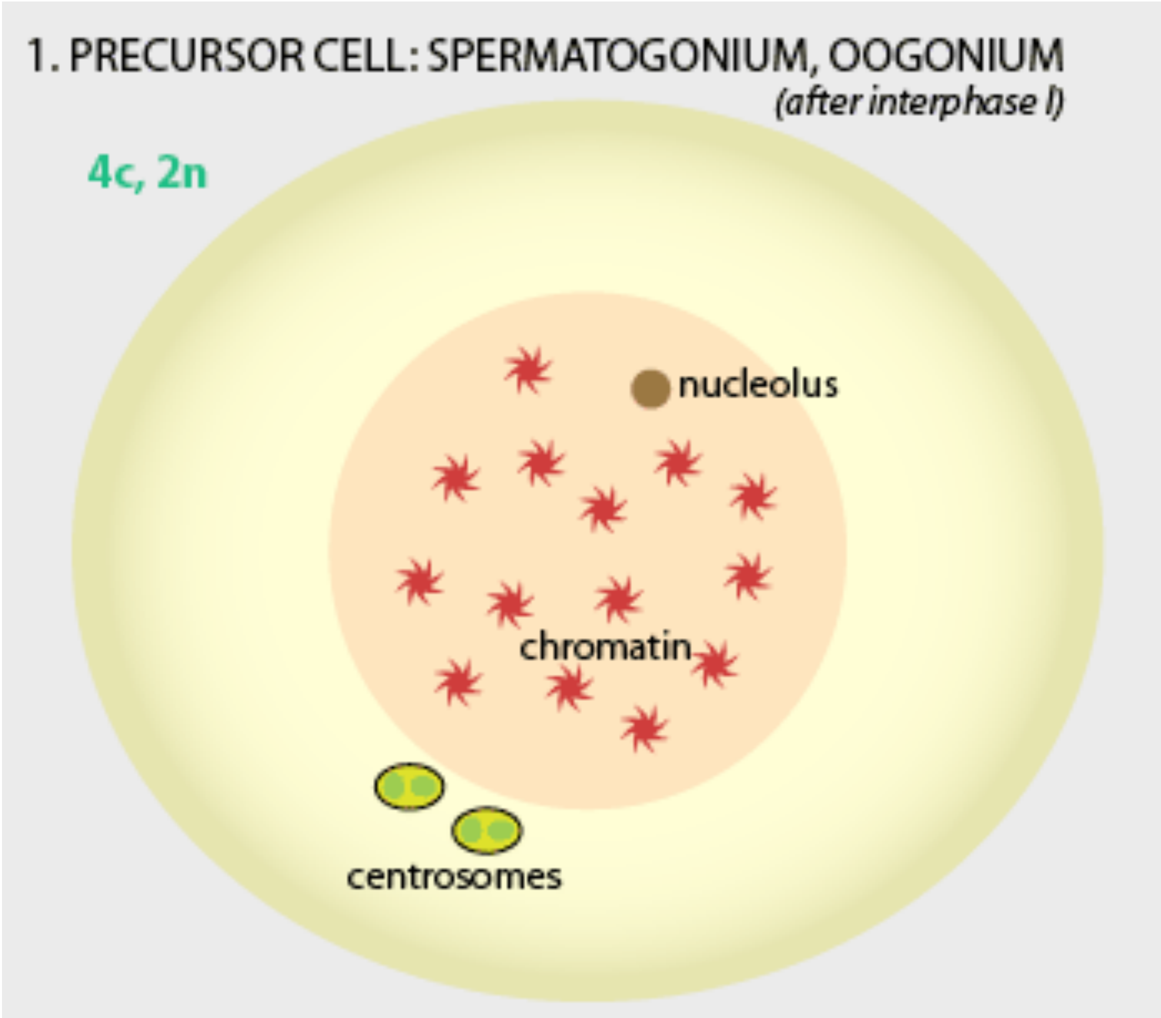
Meiosis II



MEIOSIS II: Separates sister chromatids
TELOPHASE I AND CYTOKINESIS
PROPHASE II
METAPHASE II
ANAPHASE II
TELOPHASE II AND CYTOKINESIS
Cleavage furrow
Sister chromatids separate
Haploid daughter cells forming
Two haploid cells form; chromosomes are still double
During another round of cell division, the sister chromatids finally separate;four haploid daughter cells result, containing single chromosomes

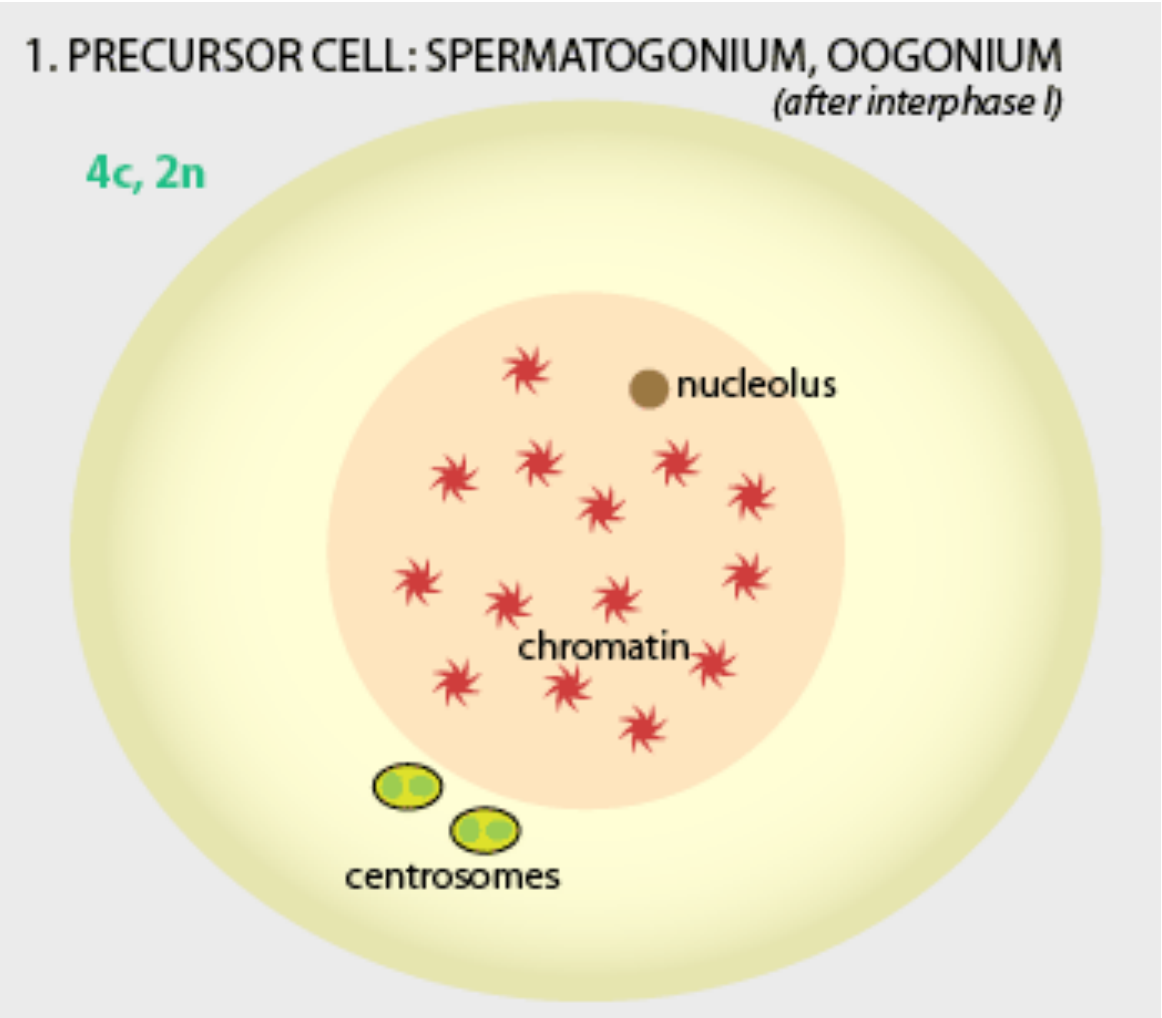
Meiosis: A Closer Look

- **Interphase G_1**
  - cell grows
  - organelles replicate
  - carries out destined functions
- **Interphase S**
  - replicates DNA
  - replicates centrosomes
  - cell grows
  - organelles replicate
  - carries out destined functions



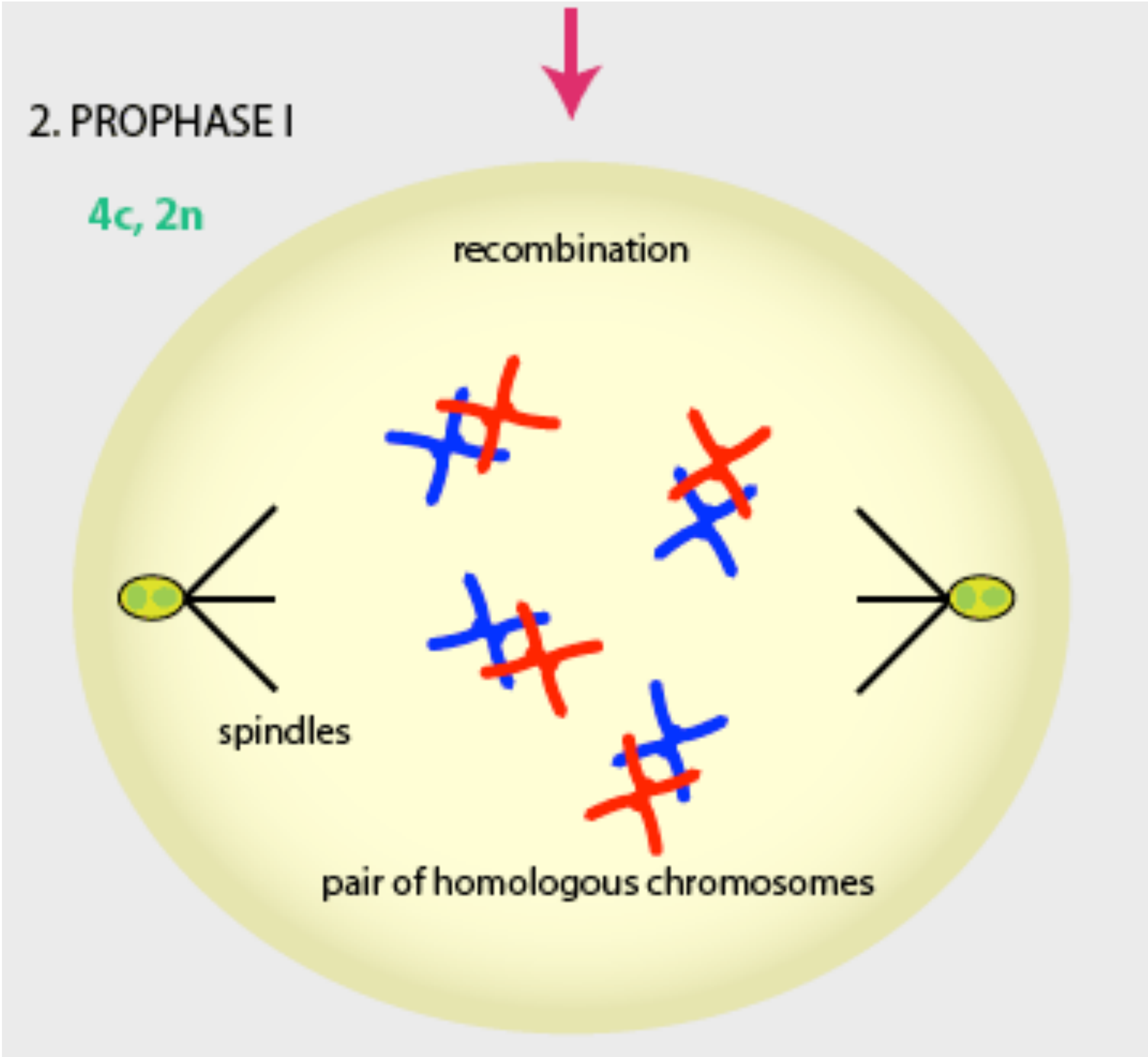
Meiosis: A Closer Look

- **Interphase G_2**
  - cell grows
  - organelles replicate
  - carries out destined functions
  - prepares for divisions
  - condenses chromatin into chromosomes (at the very end of G_2 or start of prophase)
- **Prophase I**
  - chromosomes begin to condense



Meiosis: A Closer Look

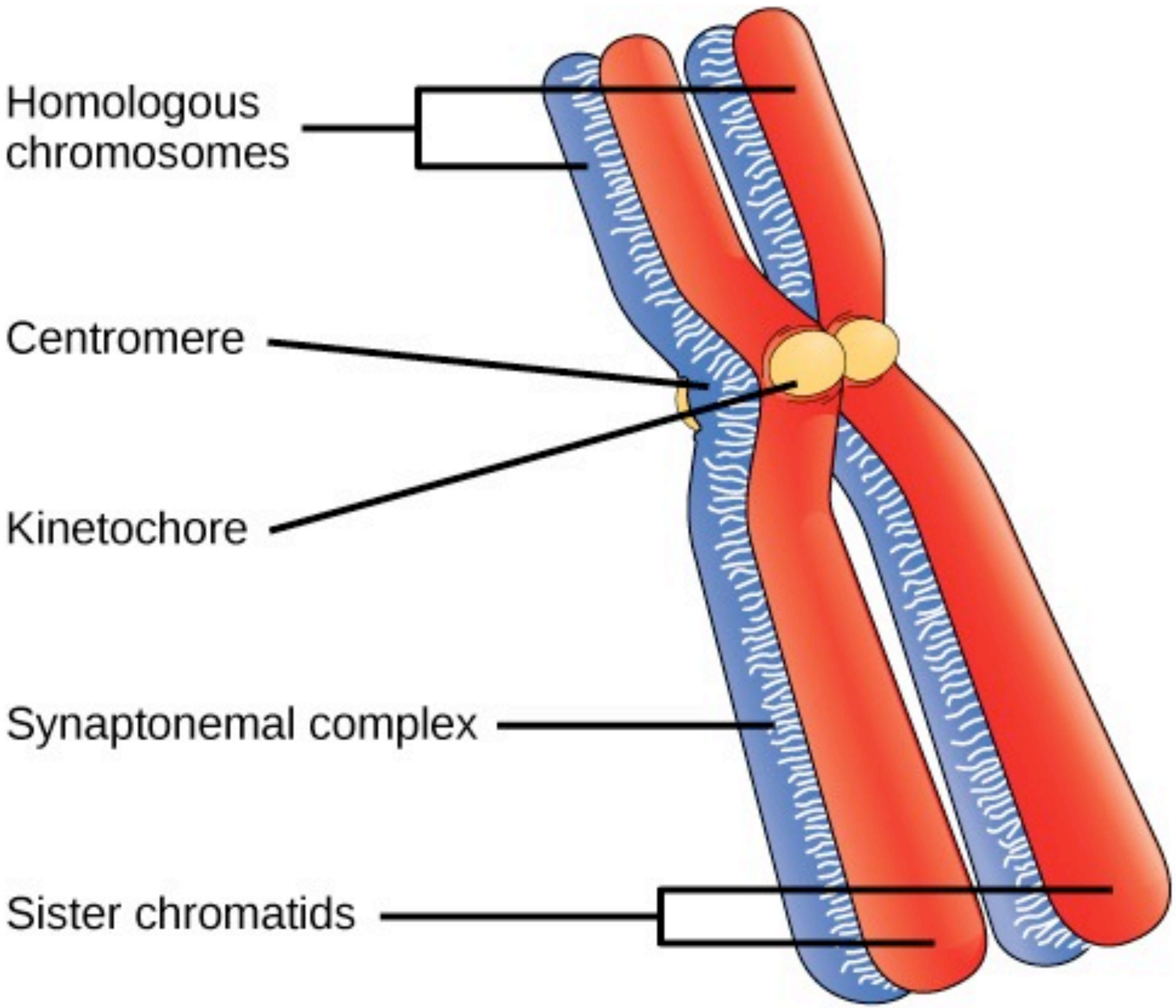
- **Prophase I (beginning)**
  - homologs pair up lengthwise
  - *synapsis** occurs, homologs connect at the synaptonemal complex
  - *crossing over** occurs, genetic exchange between corresponding segments of the chromosomes



- **Prophase I (middle)**
  - synaptonemal complex disassembles and homologs are loosely joined

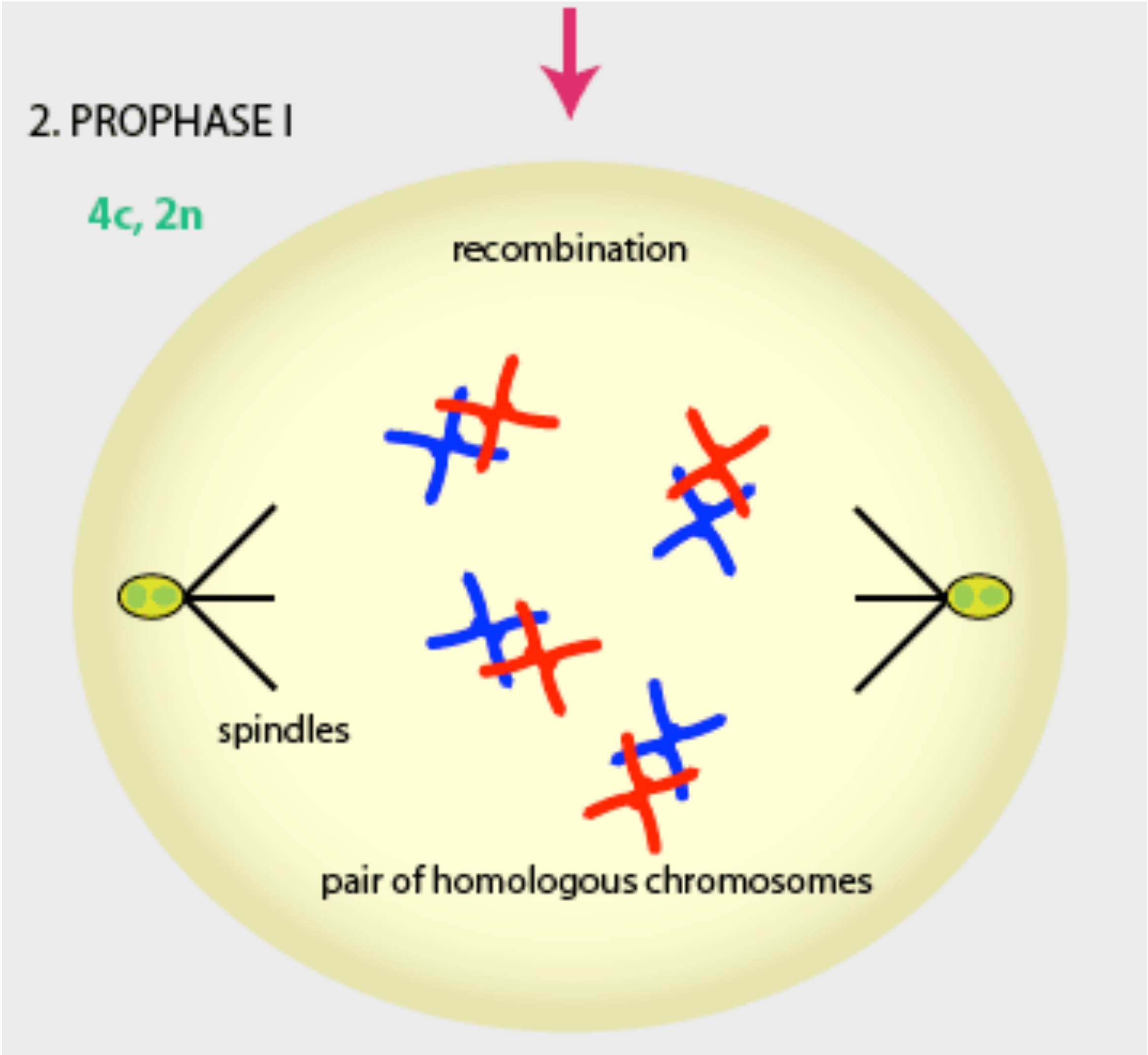
*unique to meiosis

Homologous Chromosomes Pairs (aka “tetrads”)



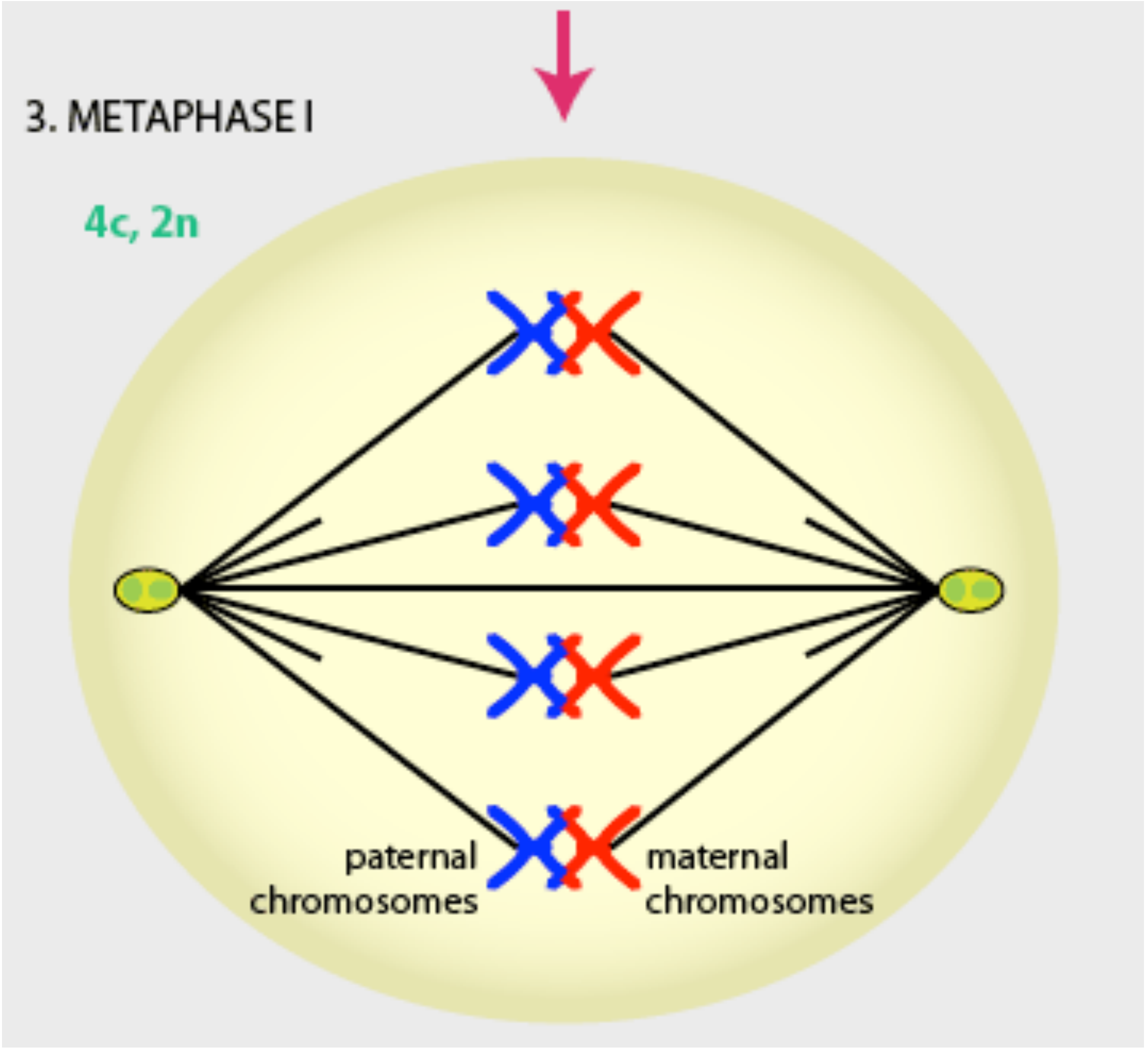
Meiosis: A Closer Look

- **Prophase I (middle)**
  - each chromosome has an "x" shaped region called the chiasma. It is the point where crossing over has occurred
  - nuclear envelope breaks down
  - centrosomes migrate
  - spindle fibers form



Meiosis: A Closer Look

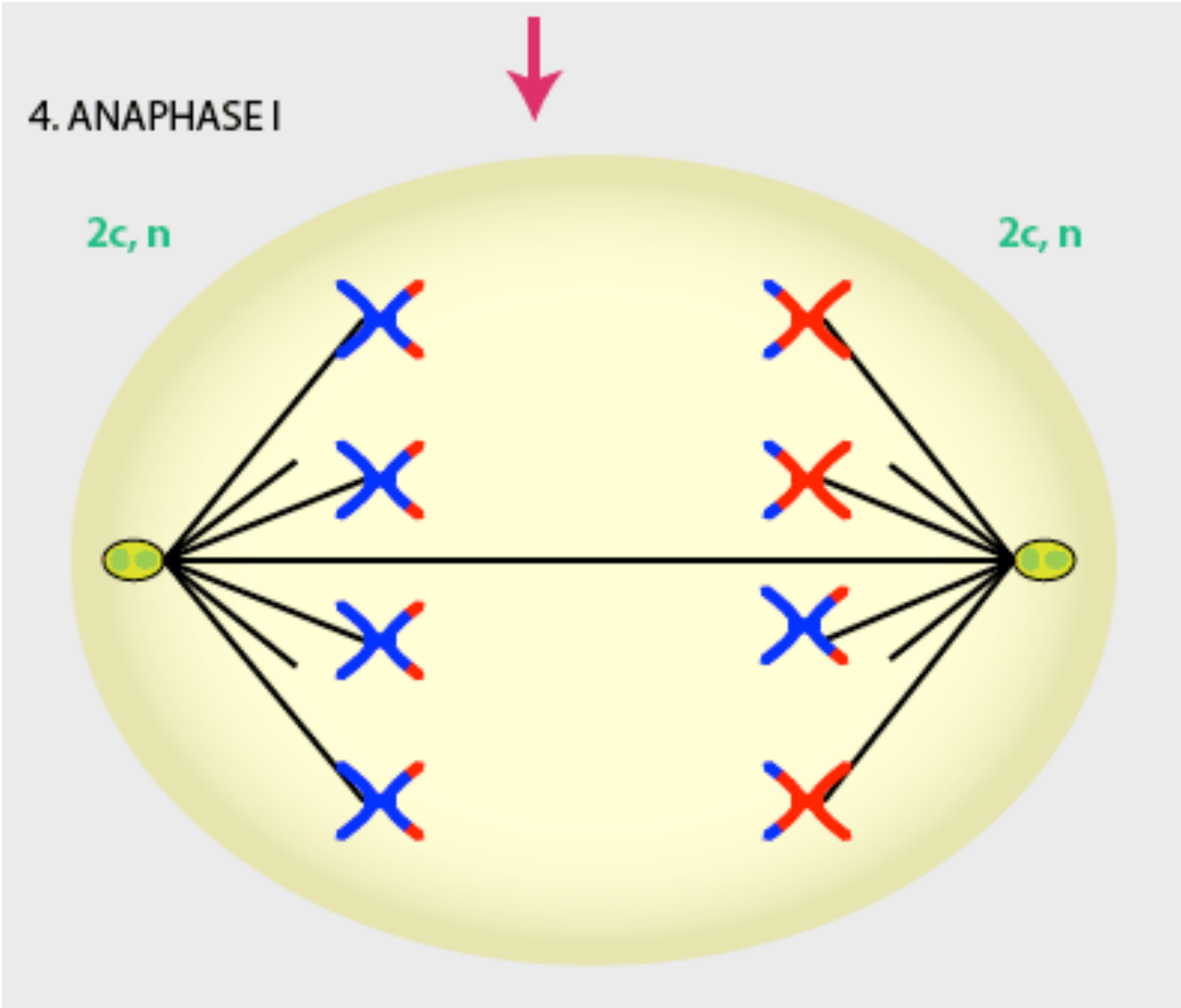
- **Prophase I (late)**
  - microtubules attach to kinetochores and move the homologous pairs toward the middle of the cell
- **Metaphase I**
  - homologous pairs (tetrads*) are now aligned at the metaphase plate
  - both chromatids of each pair are attached to spindles from opposite poles



*unique to meiosis

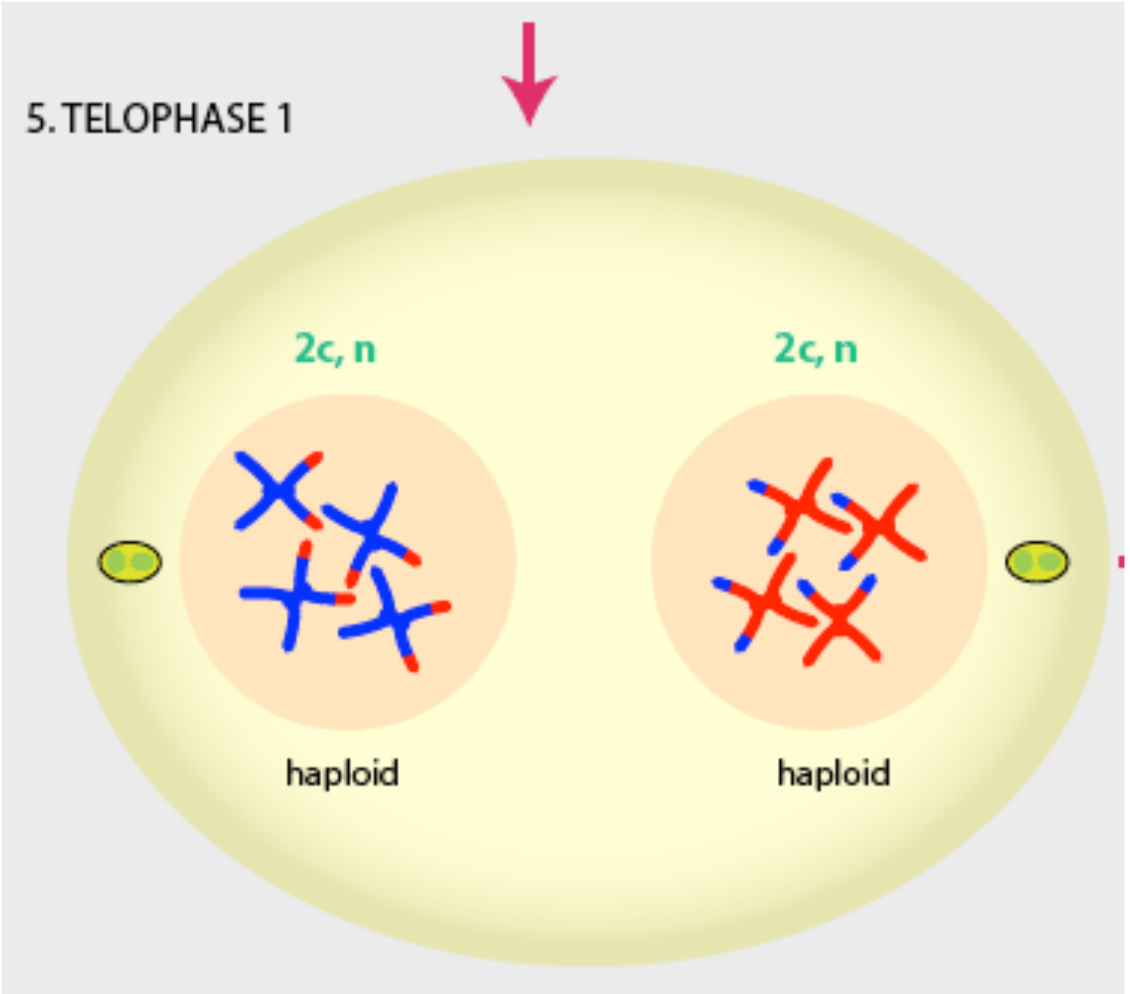
Meiosis: A Closer Look

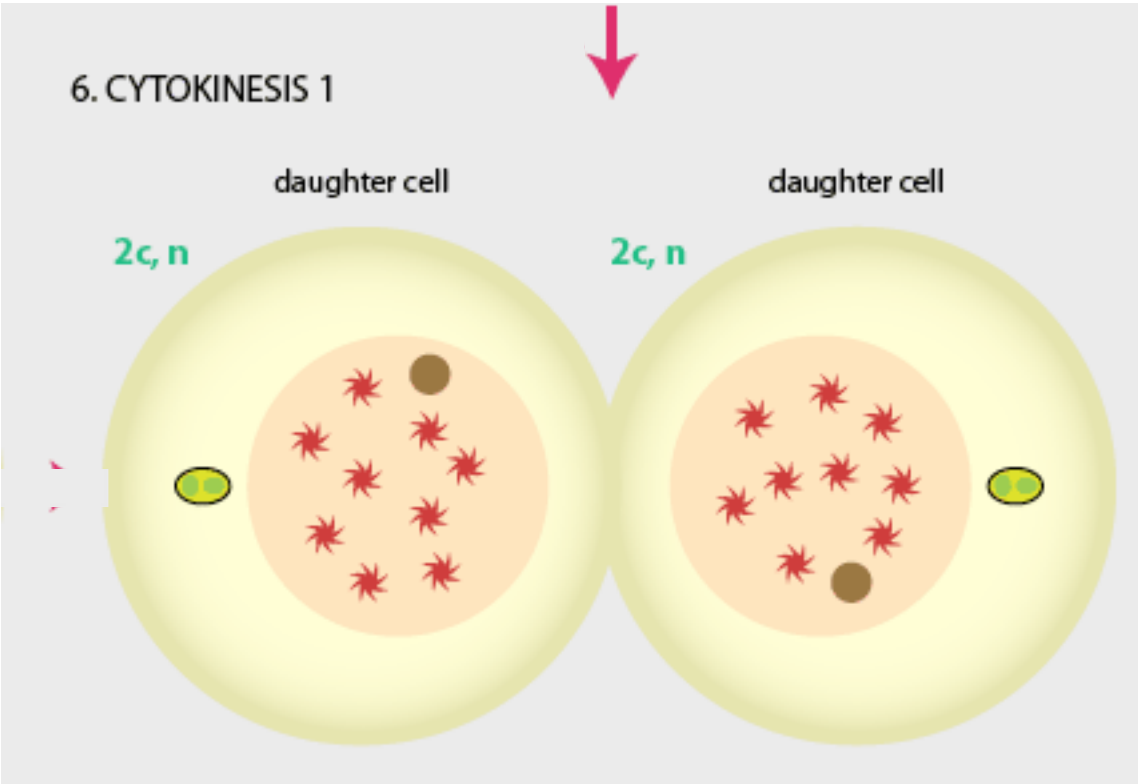
- **Anaphase I**
  - proteins holding homologs together break down
  - homologs separate and move toward each opposite pole
  - cohesion remains between sister chromatids and they move as a unit



Meiosis: A Closer Look

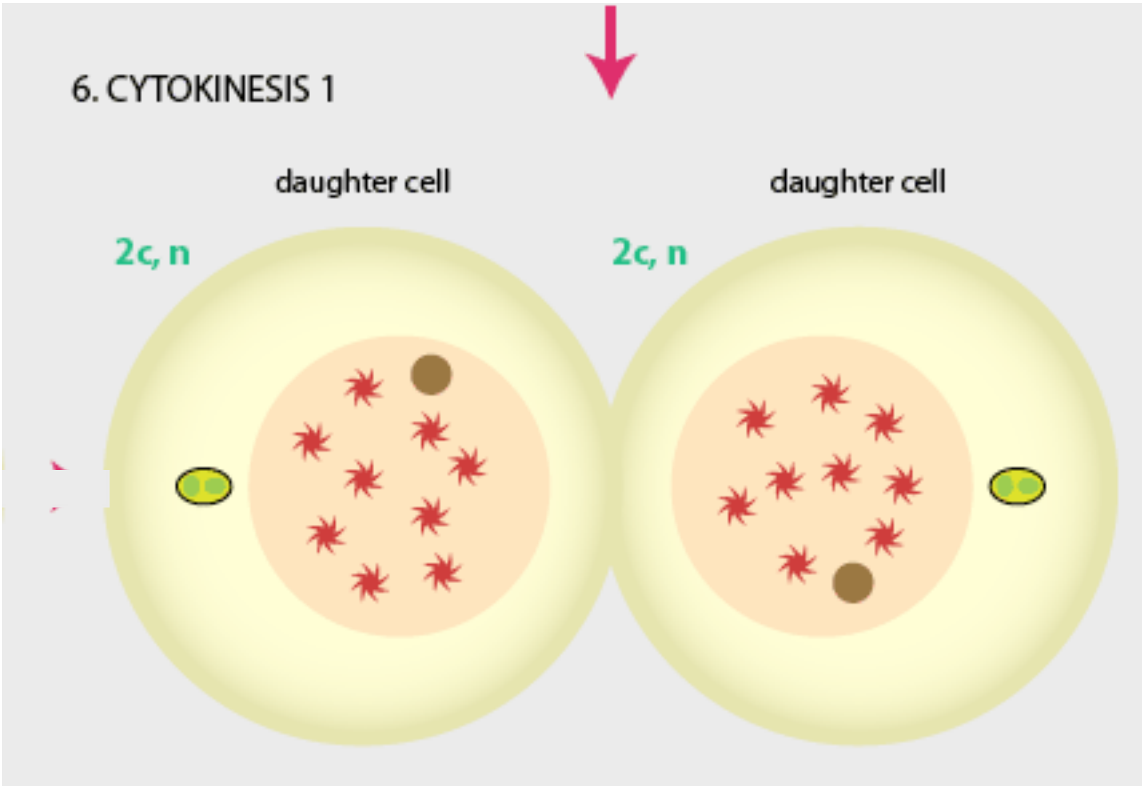
- **Telophase I & Cytokinesis**
  - each set has a haploid set of chromosomes but amount of DNA is still that of an normal cell
  - one of both sister chromatids contain regions of nonsister chromatid DNA
  - Cytokinesis begins before telophase I is complete





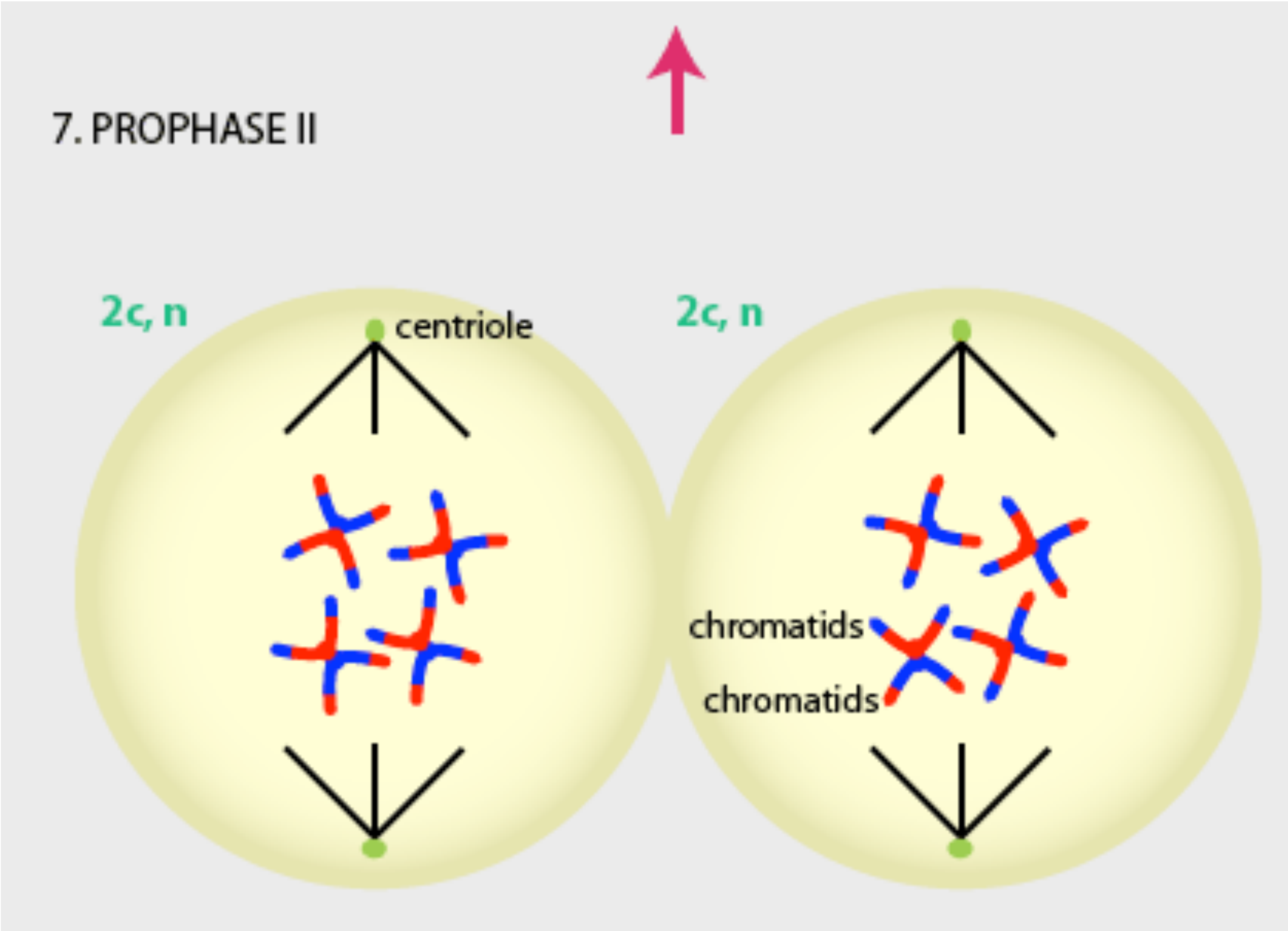
Meiosis: A Closer Look

- **Cytokinesis**
  - animal cells create cleavage furrows and plant cells create cell plates
  - no DNA replication between meiosis I and II
  - In some species the chromosomes de-condense and reform a nuclear envelope



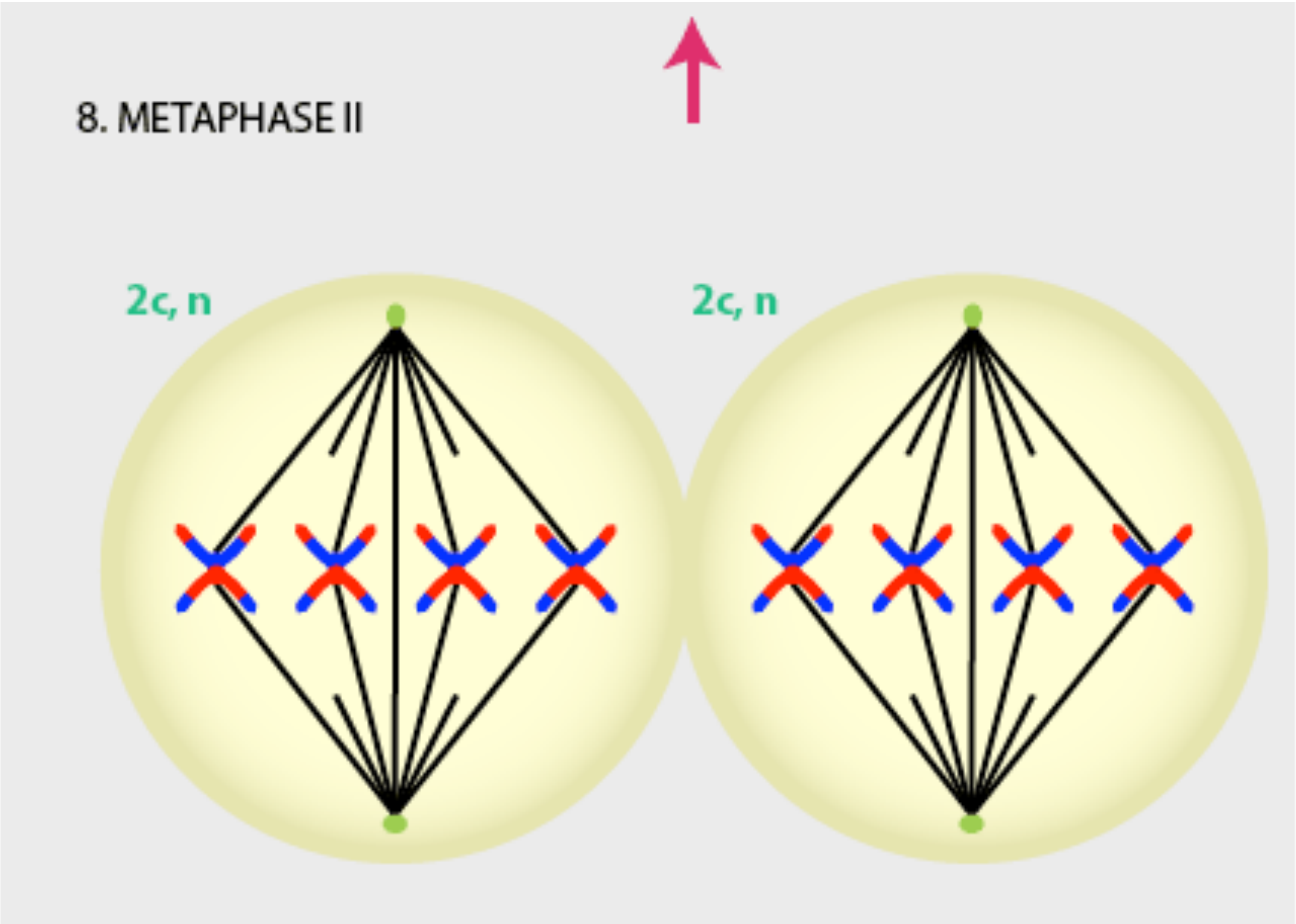
Meiosis: A Closer Look

- **Prophase II**
  - spindles form once again
  - microtubules attach to kinetochores and move the sister chromatids toward the middle of the cell



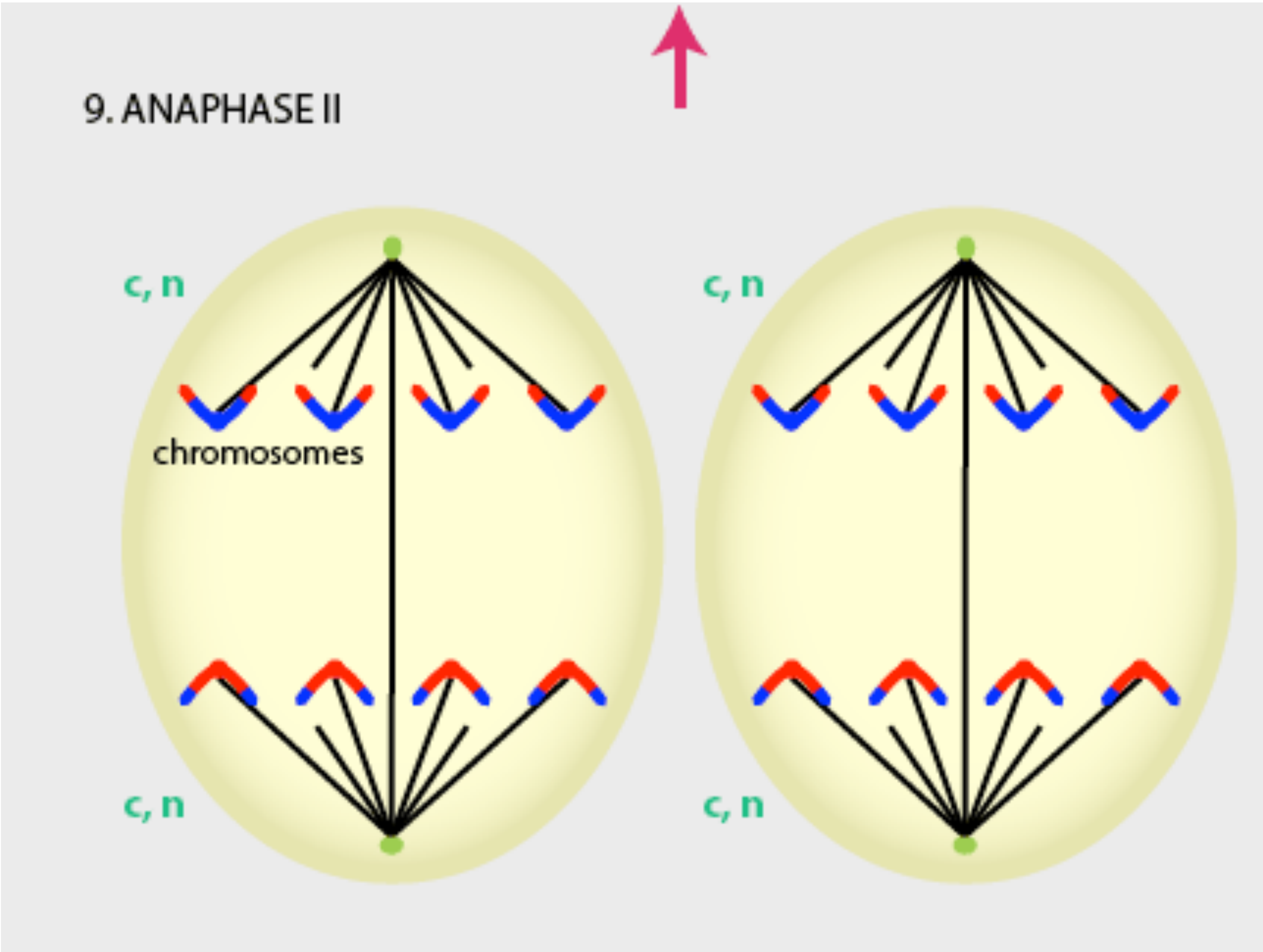
Meiosis: A Closer Look

- **Metaphase II**
  - sister chromatids are now aligned at the metaphase plate
  - each chromatid of each pair are genetically unique because of crossing over



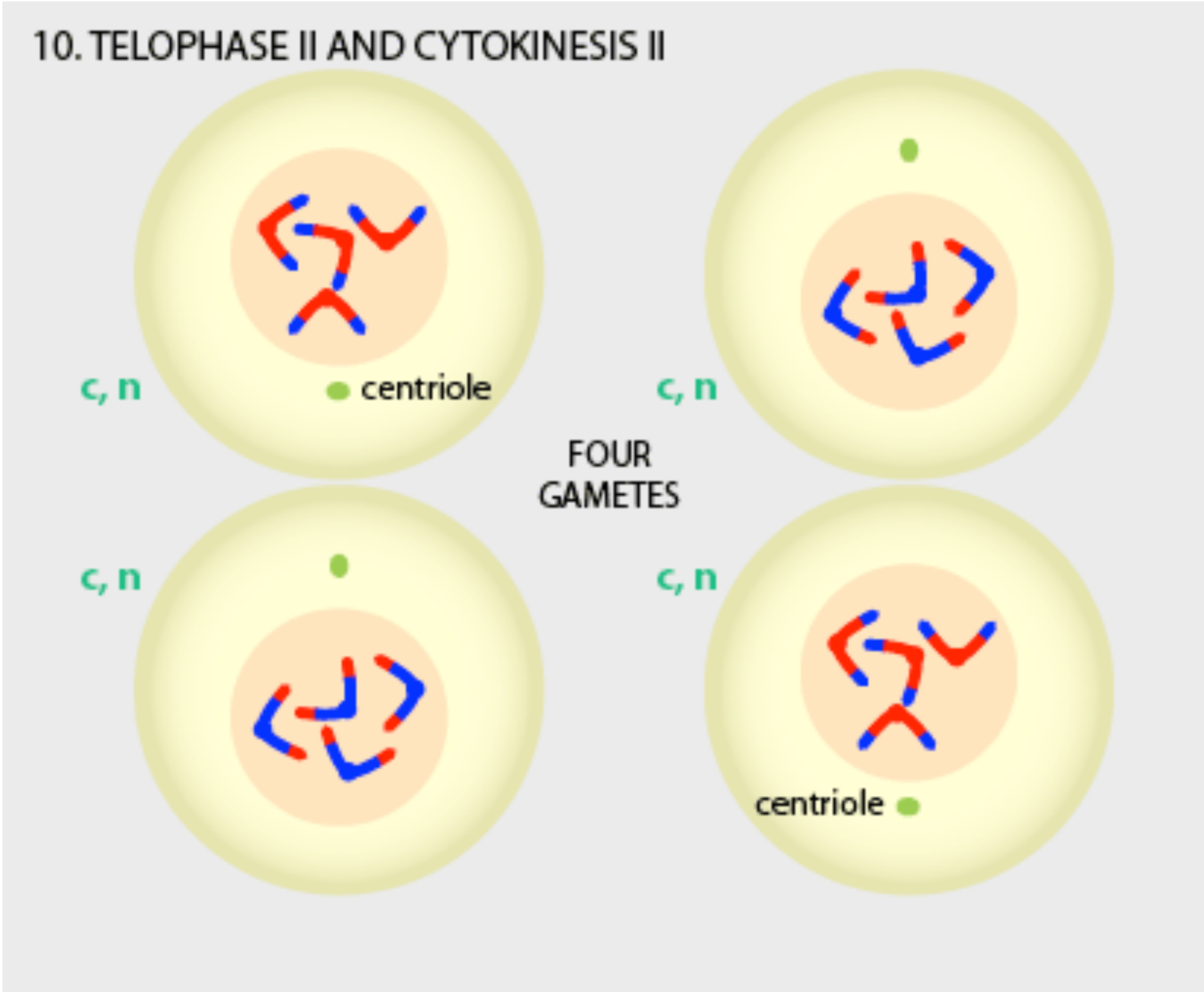
Meiosis: A Closer Look

- **Anaphase II**
  - proteins holding sister chromatids breakdown
  - chromatids separate and move toward opposite poles as individual chromosomes

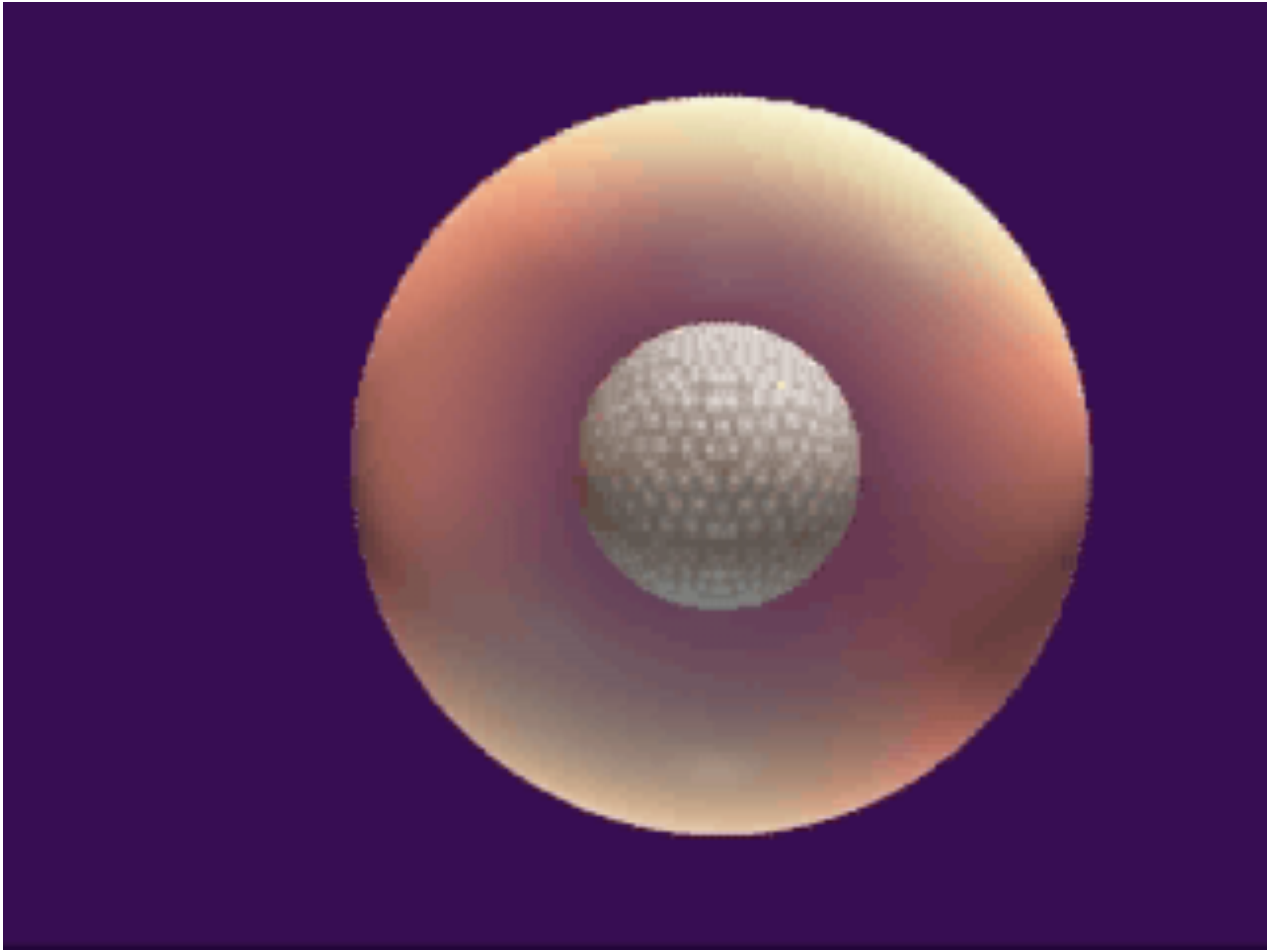


Meiosis: A Closer Look

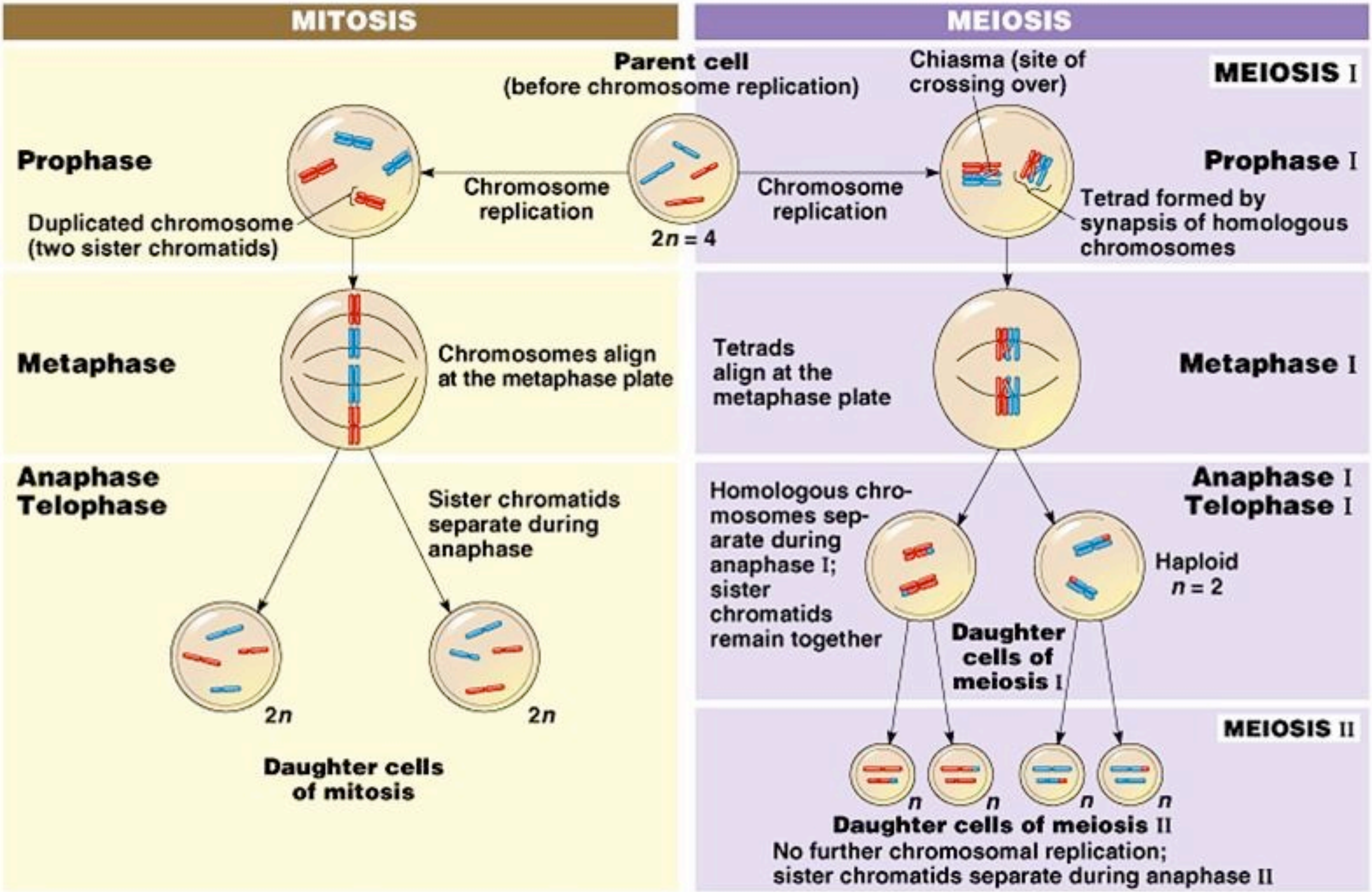
- **Telophase II & Cytokinesis**
  - chromosomes begin to de-condense
  - nuclear envelop reforms
  - cytokinesis results in 4 haploid cells
  - 4 cells are unique and haploid



Animation of Meiosis



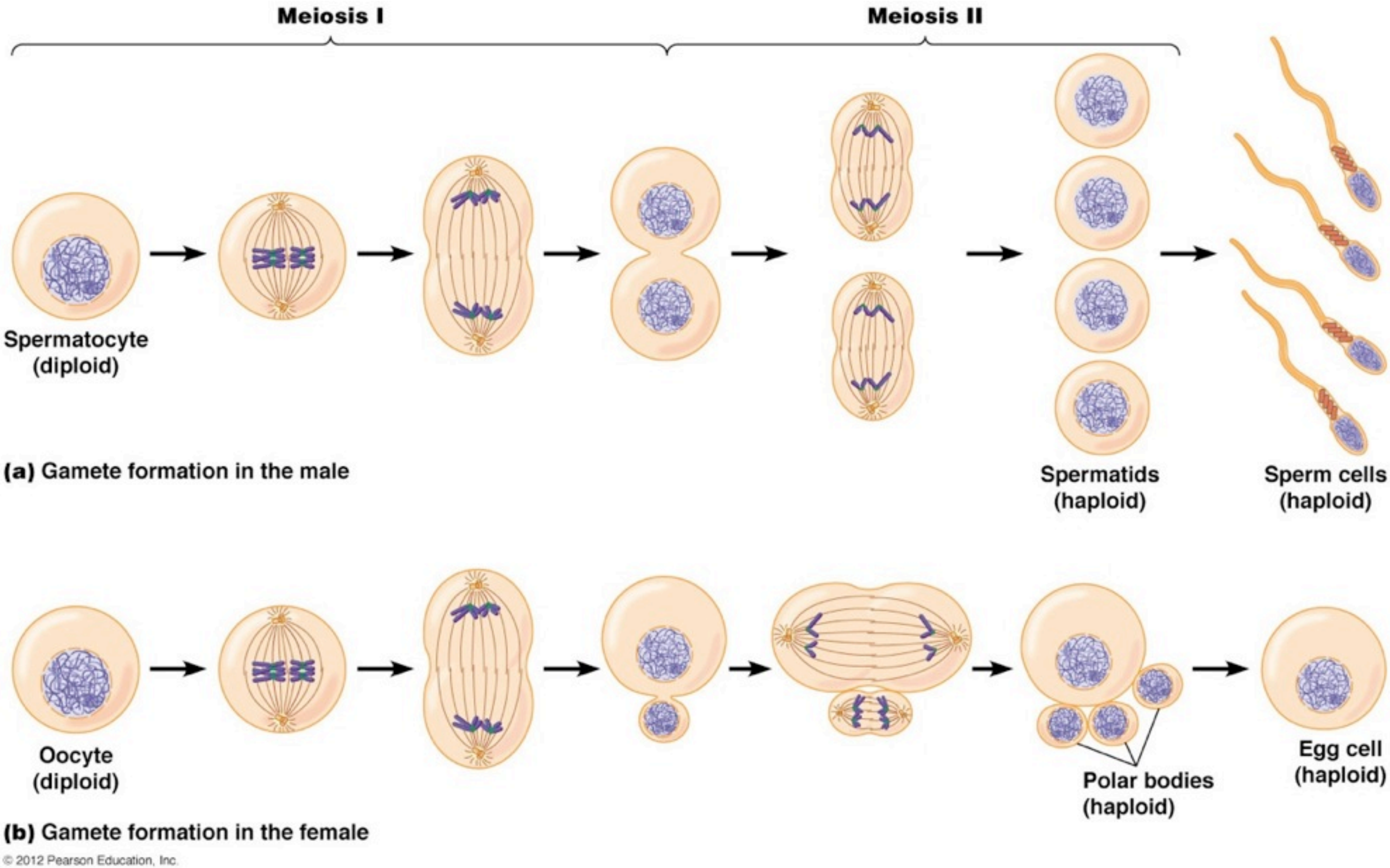
Illustrated Comparison



Summary: Mitosis/Meiosis

SUMMARY		
Event	**Mitosis**	**Meiosis**
DNA replication	Occurs during interphase before nuclear division begins	Occurs once, during the interphase before meiosis I begins
Number of divisions	One, including prophase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
Synapsis of homologous chromosomes	Does not occur	Synapsis is unique to meiosis: During prophase I, the homologous chromosomes join along their length, forming tetrads (groups of four chromatids); synapsis is associated with crossing over between nonsister chromatids
Number of daughter cells and genetic composition	Two, each diploid (2*n*) and genetically identical to the parent cell	Four, each haploid (*n*), containing half as many chromosomes as the parent cell; genetically nonidentical to the parent cell and to each other
Role in the animal body	Enables multicellular adult to arise from zygote; produces cells for growth and tissue repair	Produces gametes; reduces chromosome number by half and introduces genetic variability among the gametes

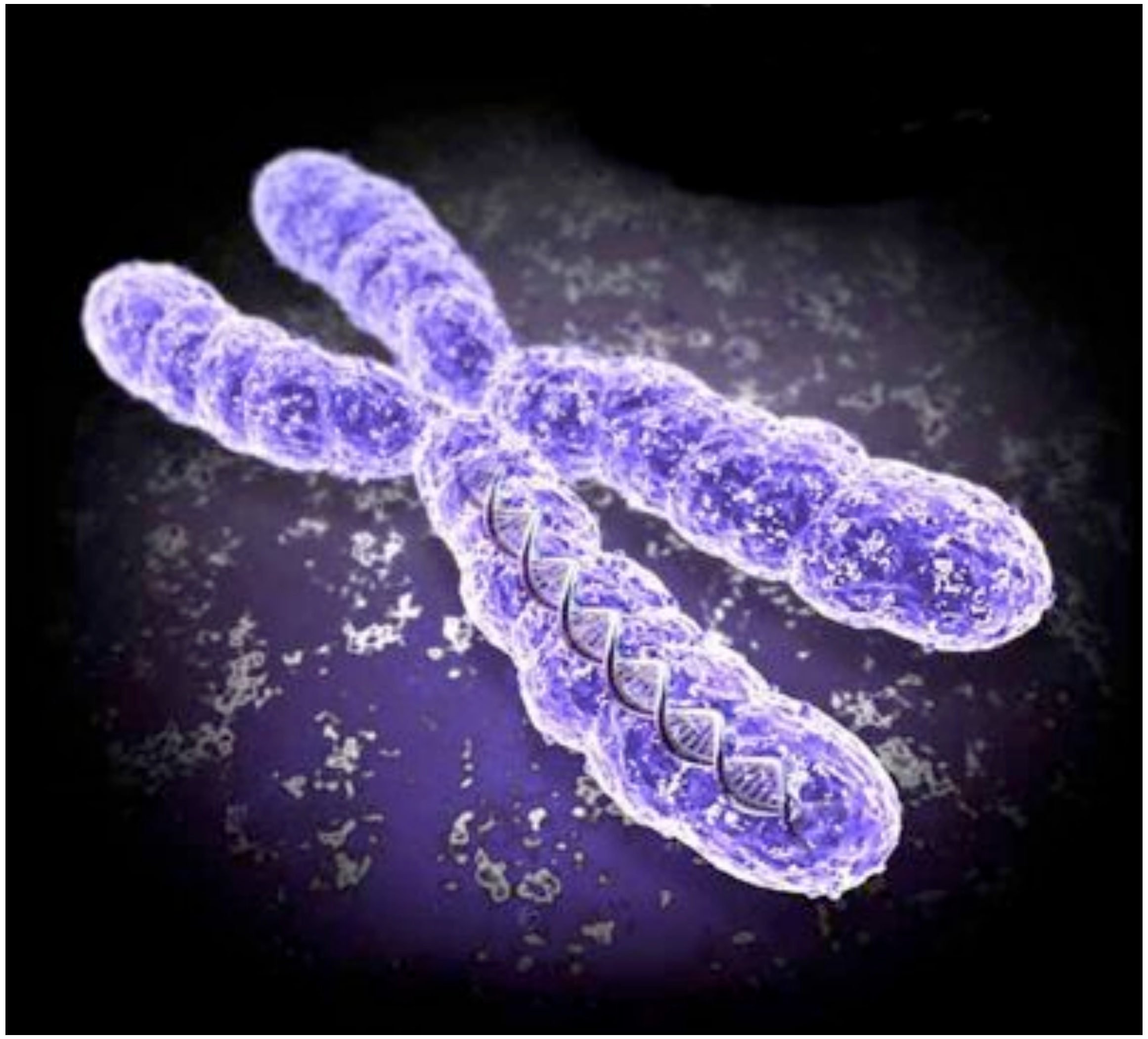
Production of Gametes



Meiosis

IV.

Main Idea: Meiosis creates the genetic variation associated with sexual reproduction.



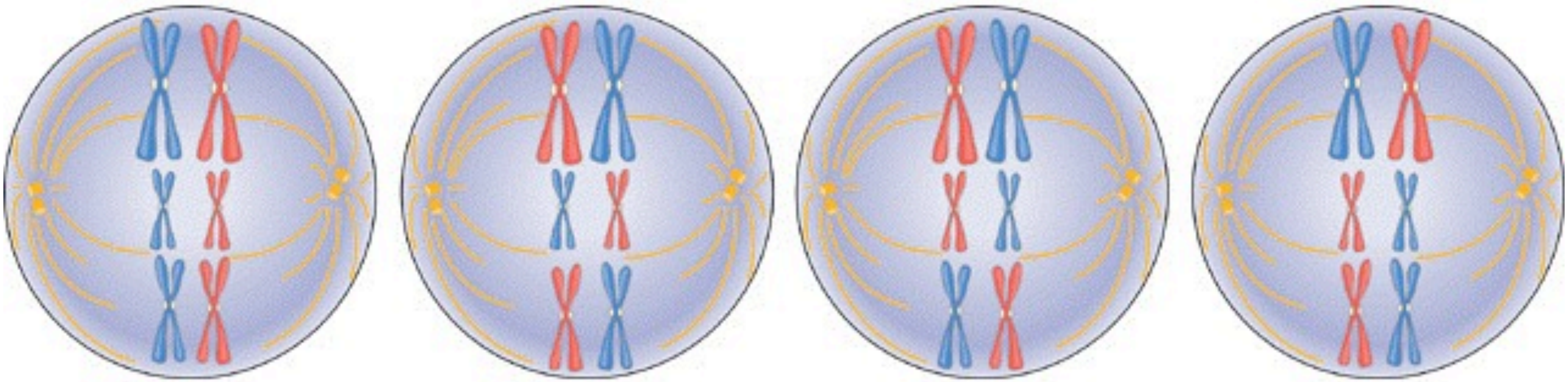
Evolution & Genetics

- Mutations created every allele, and will create every new allele in the future.
  - *recall alleles are different genes*
- Sexual Reproduction/Meiosis shuffles those alleles to produce unique individuals and variation in gene pools
- Meiosis plays therefor a direct role in evolution by providing the necessary variation for natural selection to work on.
- **Also, understanding meiosis provides an understanding of the mechanics behind inheritance of traits (genetics)**

Evolution & Genetics

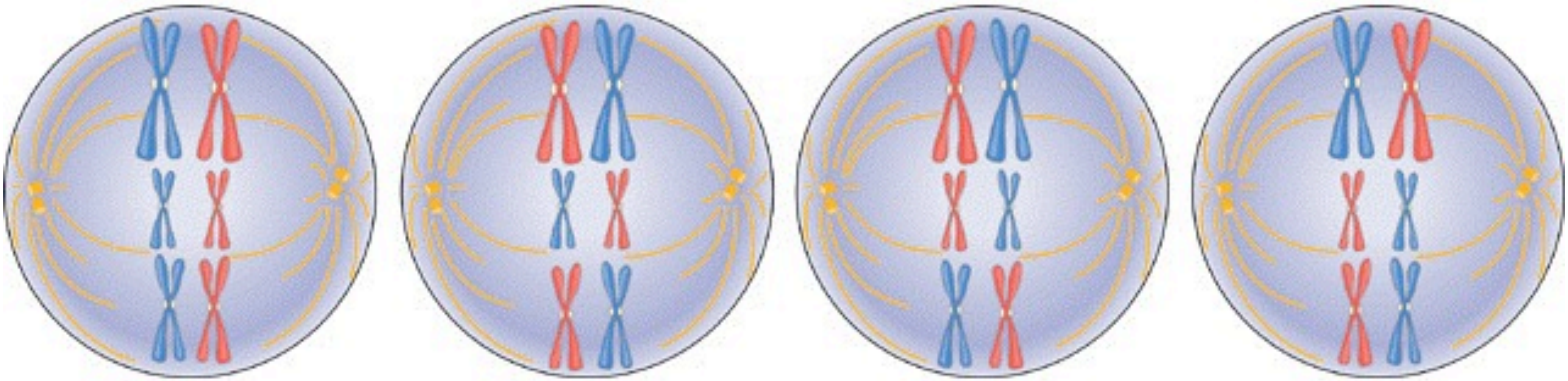
- The following slides will explore 3 mechanisms that generate variation in sexual reproduction.
  - independent assortment,
  - crossing over and
  - random fertilization
- This exploration is important for two reasons:
  - First, it illuminates the sources of variation that evolution requires.
  - **Secondly, understanding meiosis and these same mechanisms and that generate variation will provide the foundation for our understanding of inheritance (genetics).**

Independent assortment



- In metaphase 1, the homologous pairs (tetrads) align randomly at the cellular equator.
- The paternal and maternal chromosomes are randomly oriented towards one of the poles.
  - *Notice above 3 homologous pairs (tetrads) can arrange themselves in 4 different ways.*

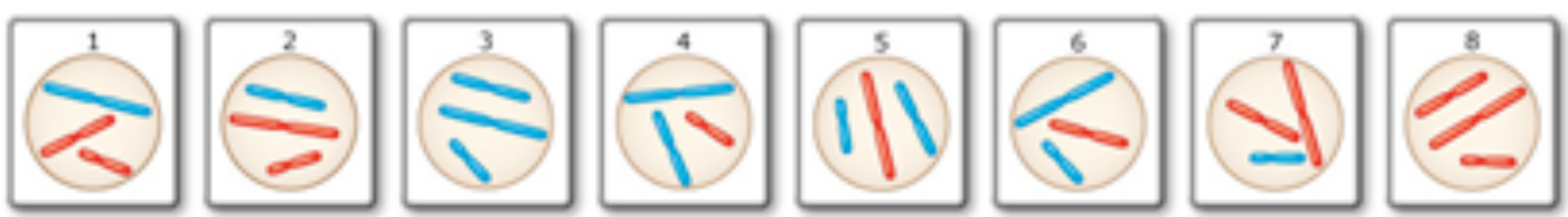
Independent assortment



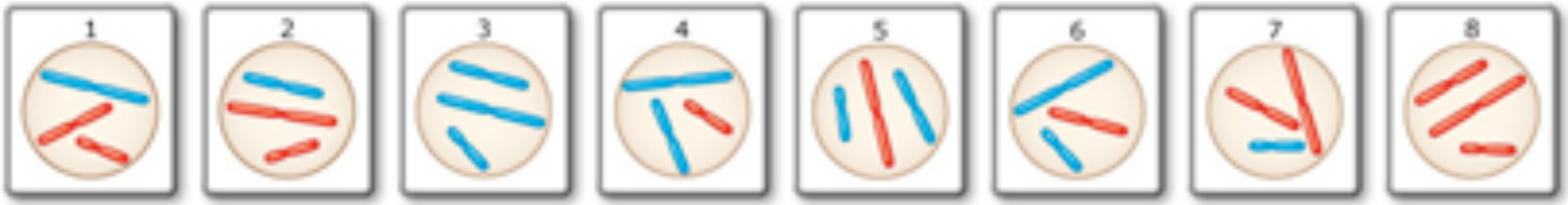
- Thus every gamete has a 50% chance of getting a paternal or maternal chromosome for each and number of chromosome pairs.

How many different gametes are produced if this example of meiosis goes to completion?

8



- Each daughter cell represents one outcome of all possible combinations of paternal and maternal chromosome combinations.



- Mathematically we can calculate the number of possible combinations by using the following equation:
  - $\mathbf{2^{n}}$ where *n* is the haploid number of the organism.

Humans:

...haploid number is 23, so 2^{23} = ~8.4 million

Bottom Line-

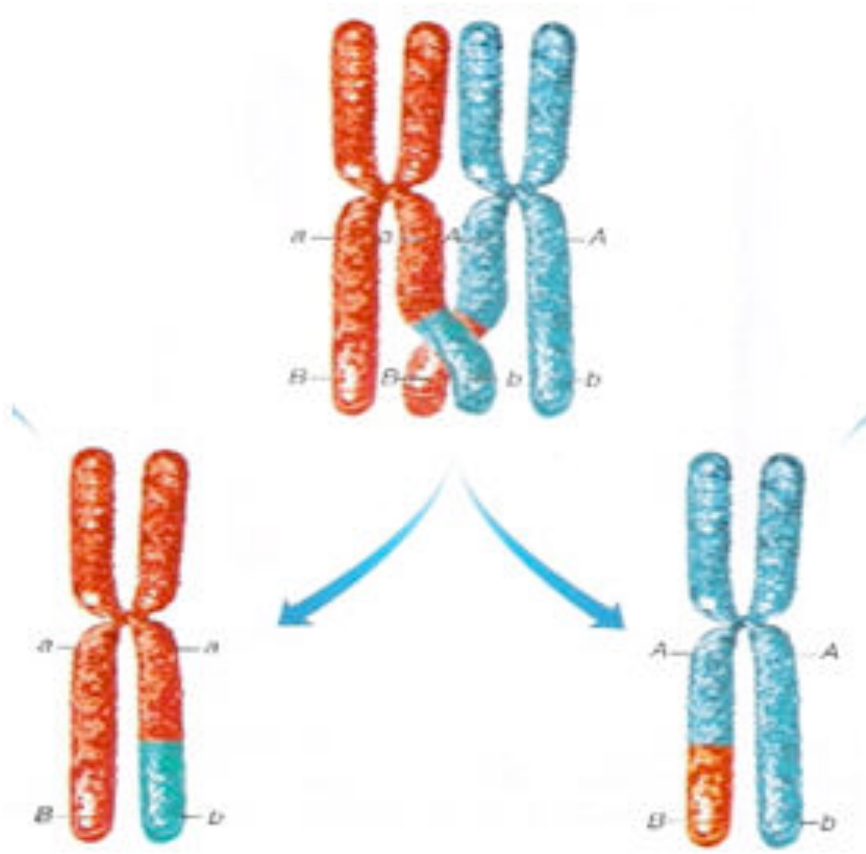
...humans can create ~8.4 million different gametes from independent assortment alone

Random Fertilization

- Although the process of ovulation is not random the actual oocyte that is ovulated is completely random.
- Although some sperm are more fit than others, the actual sperm that fertilizes the oocyte is completely random.
- This brings us back to the math... (8.4million)(8.4million)=
  - *...if any of the 8.4 million different oocytes could be fertilized by any of the 8.4 million different sperm than we could in produce ~ **70 trillion different zygotes**!*
- You can see that validity in the age old expression "you are one of a kind"
- But even this number does not tell the whole story of our truly unique nature, the fact is the actual number is far greater than 70 trillion.

Crossing Over

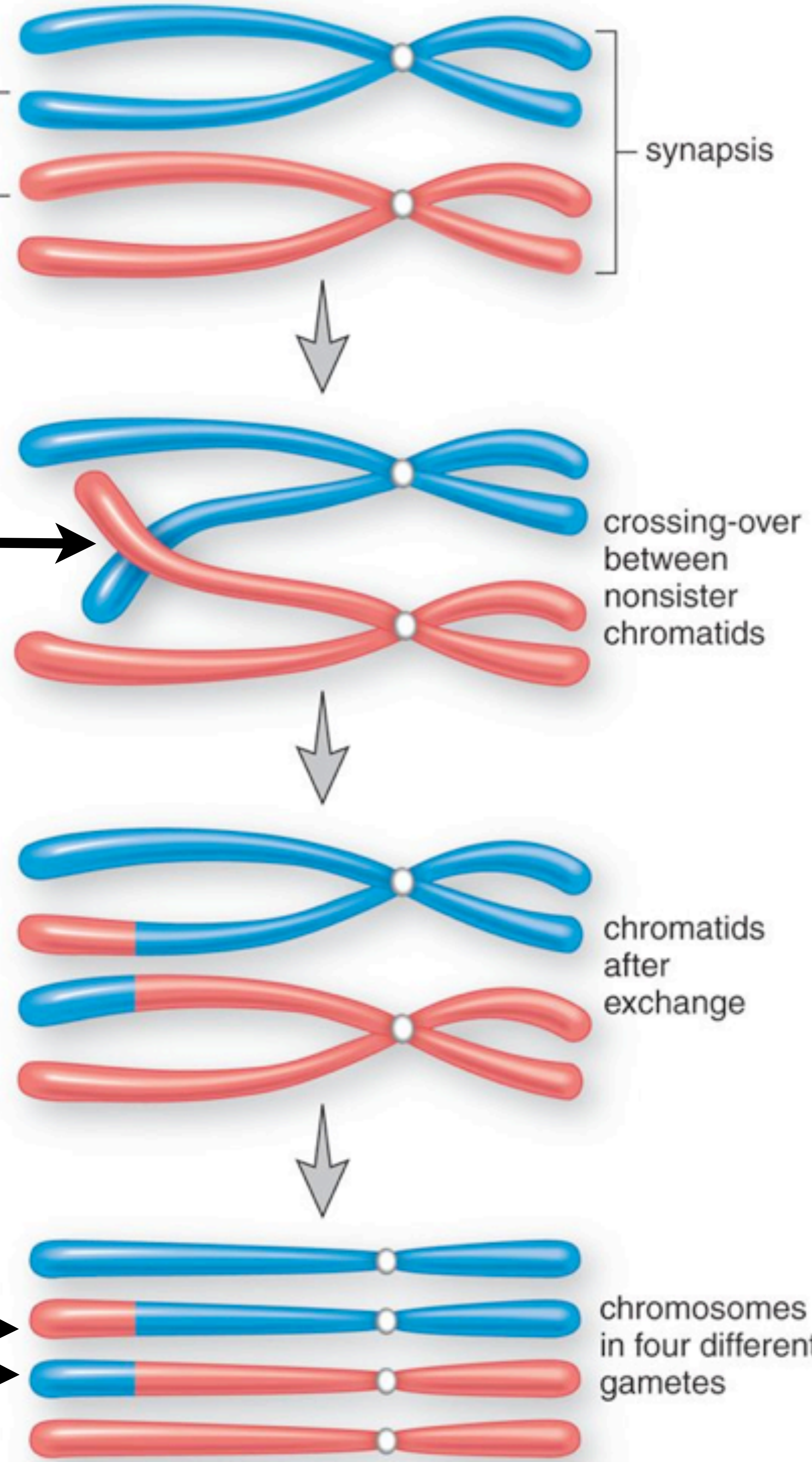
- Our examination of independent assortment leads us to believe that we inherit strictly paternal or maternal chromosomes but this is not the case.
- As a consequence of independent assortment paternal and maternal chromosomes exchange segments creating unique chromosomes different than the one we inherited from our parents.



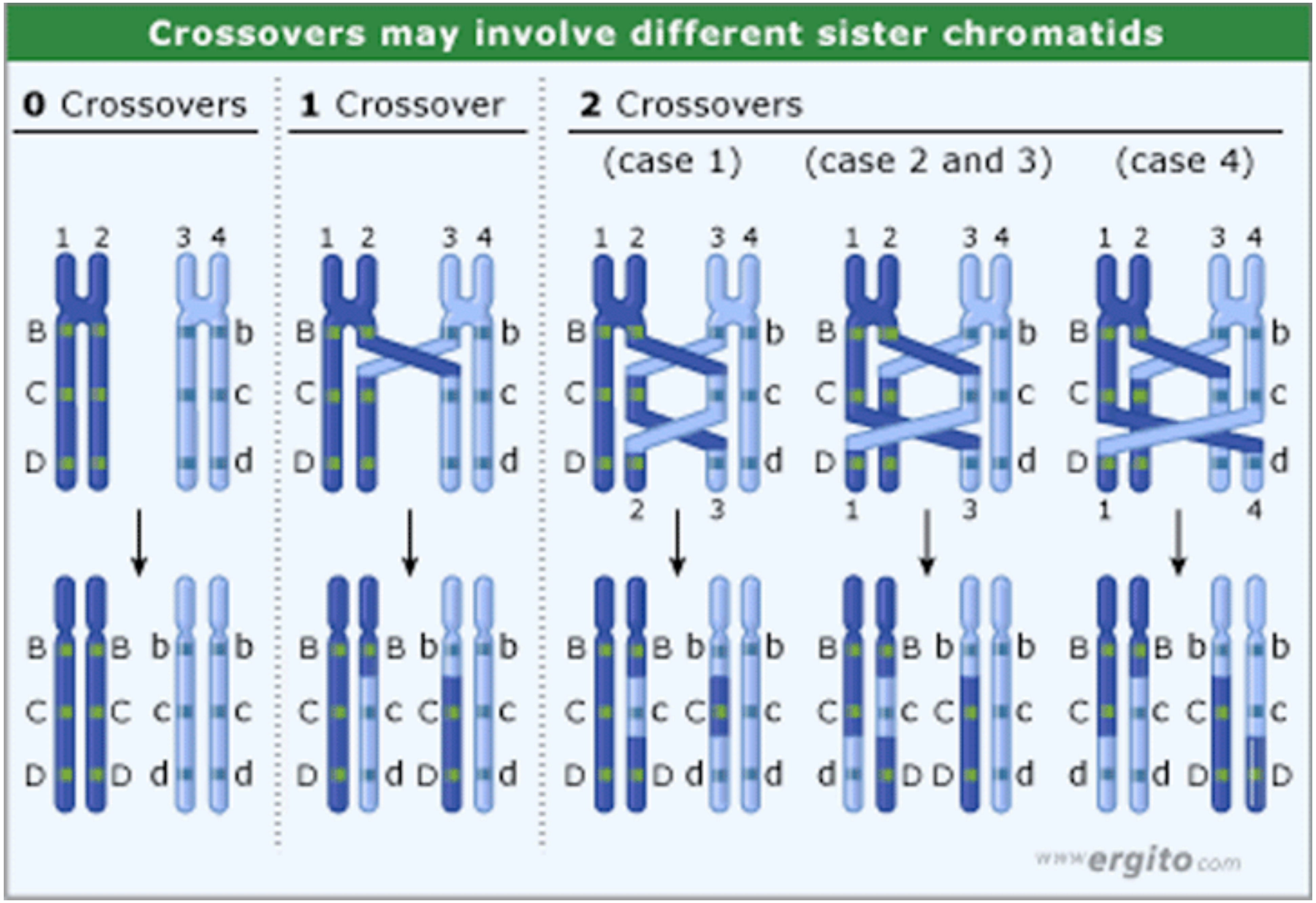
Crossing Over

Chiasma

Recombinant Chromosomes

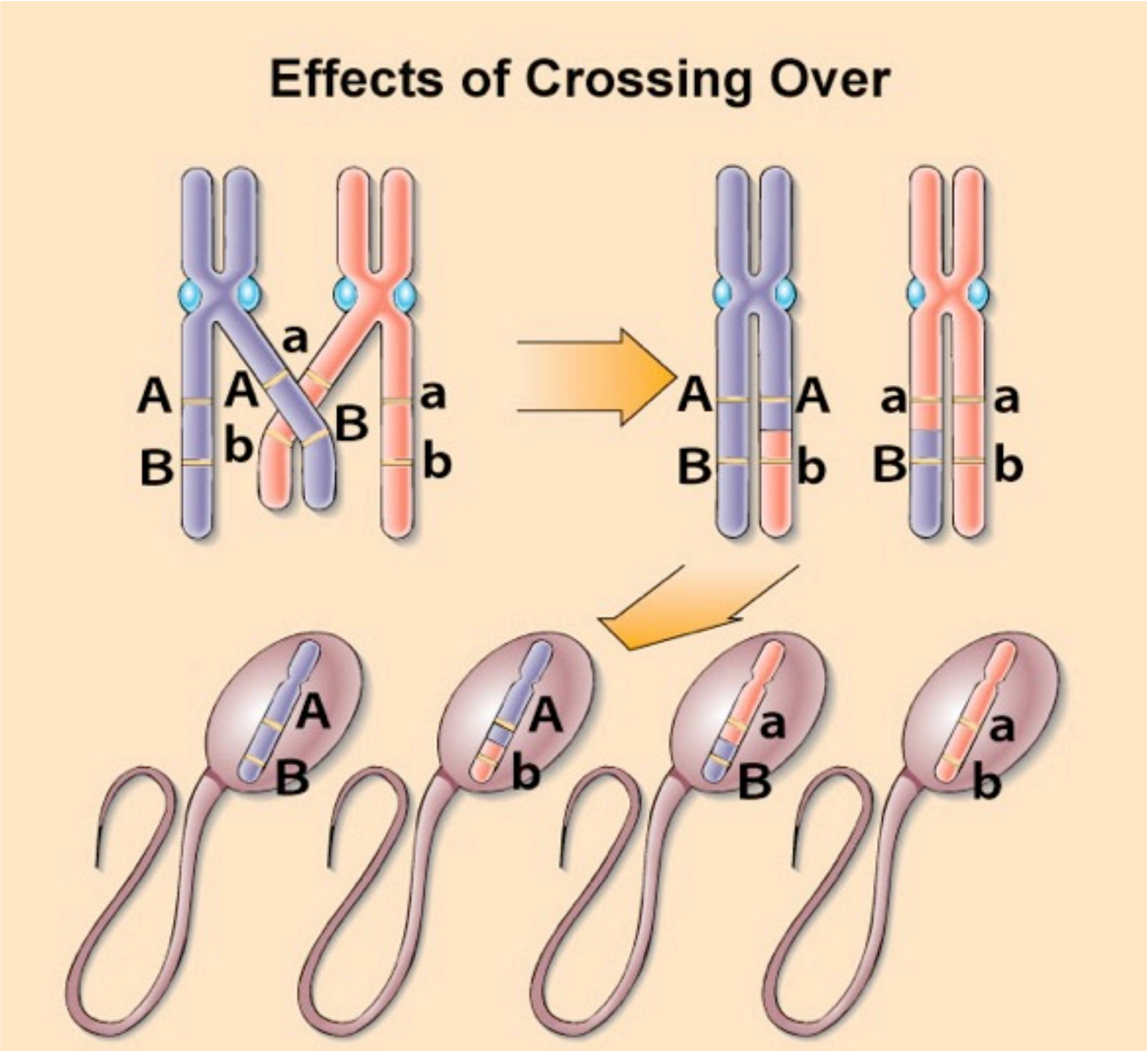


Crossing Over



Humans have on average 1-3 cross overs per chromosome pair, depending on the size of chromosome and the position of their centromere.

In this simple example crossing over has doubled the genetic variation possible.

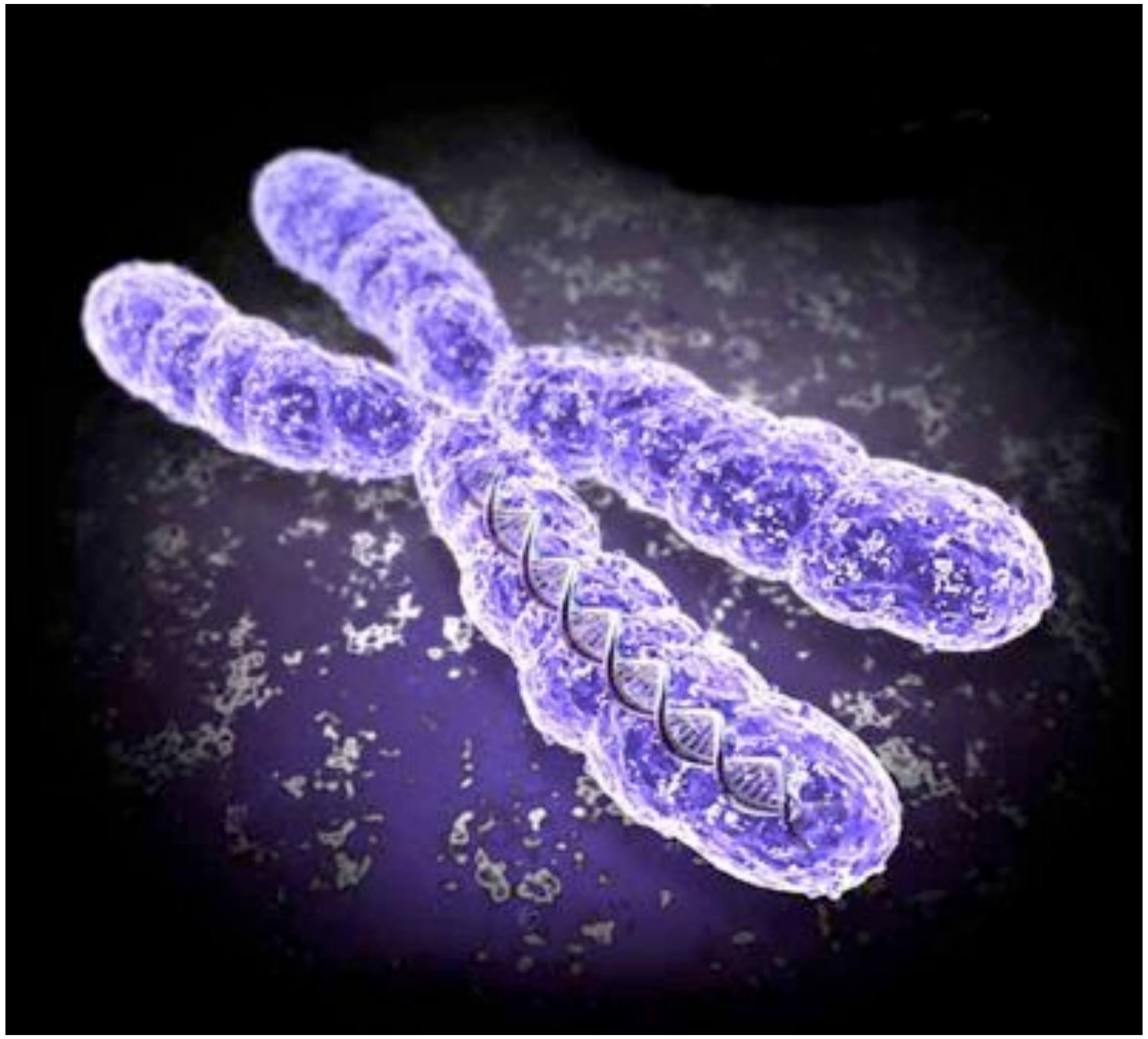


When we consider that crossing over takes place 1-3 times on 23 different pairs, multiply that by the variation that independent assortment and random fertilization create and you have an astronomically large number of possible individual offspring.

Transition to Genetics

V.

Main Idea: The principles of meiosis provide the foundation and framework for understanding the inheritance of traits.

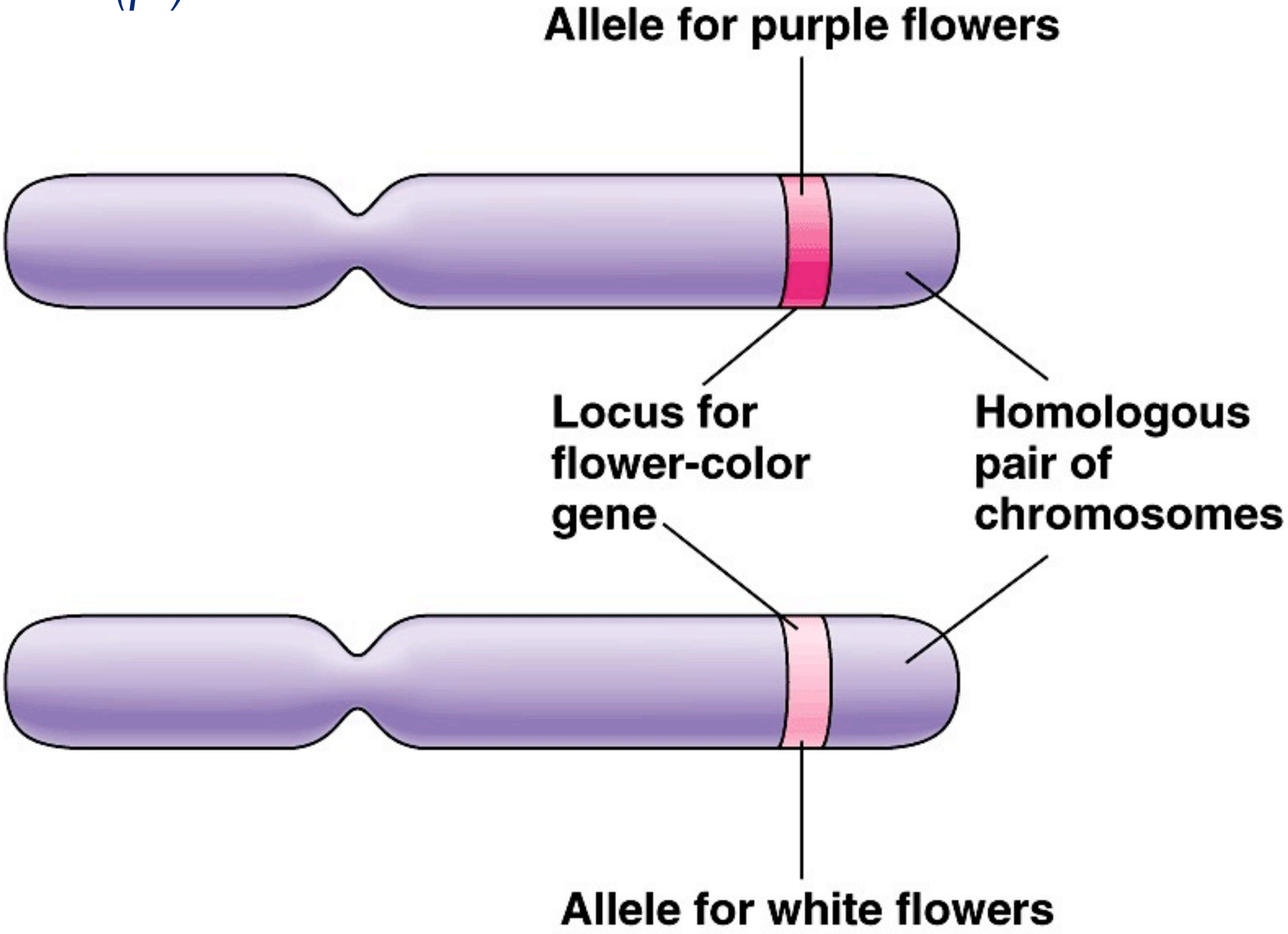


Alleles

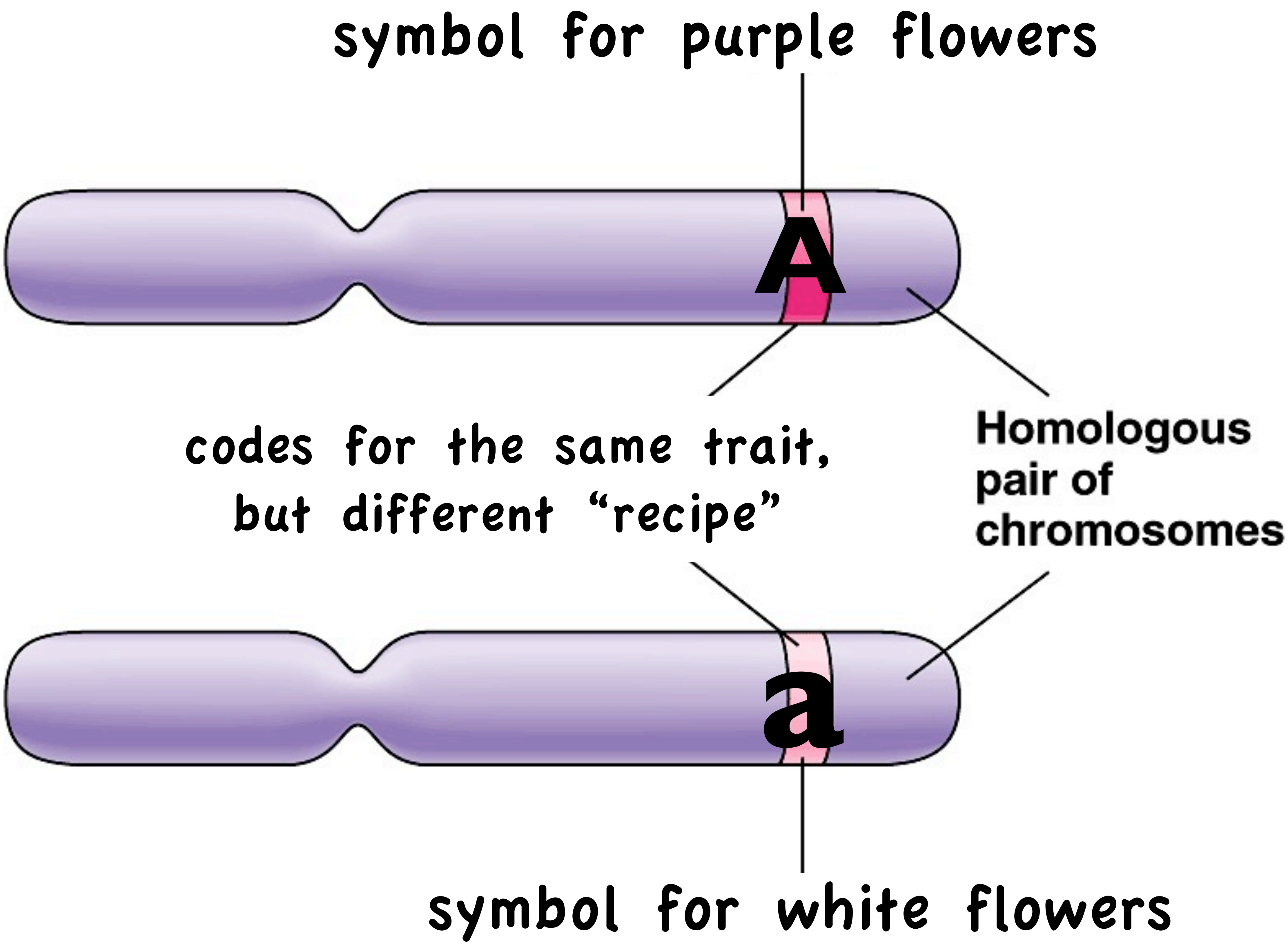
- Alleles are alternate forms of genes.
- Genes are strings of nucleotides that make up DNA.
- DNA wraps around proteins to form chromosomes.
- During reproduction parents donate chromosomes (carrying the alleles) that determine the traits of their offspring.

BUT, here is the key point! Parents do donate single alleles to their offspring rather single chromosomes. The chromosomes are packages of hundreds of alleles, to understand inheritance you have to understand the behavior of chromosomes, in other words meiosis!

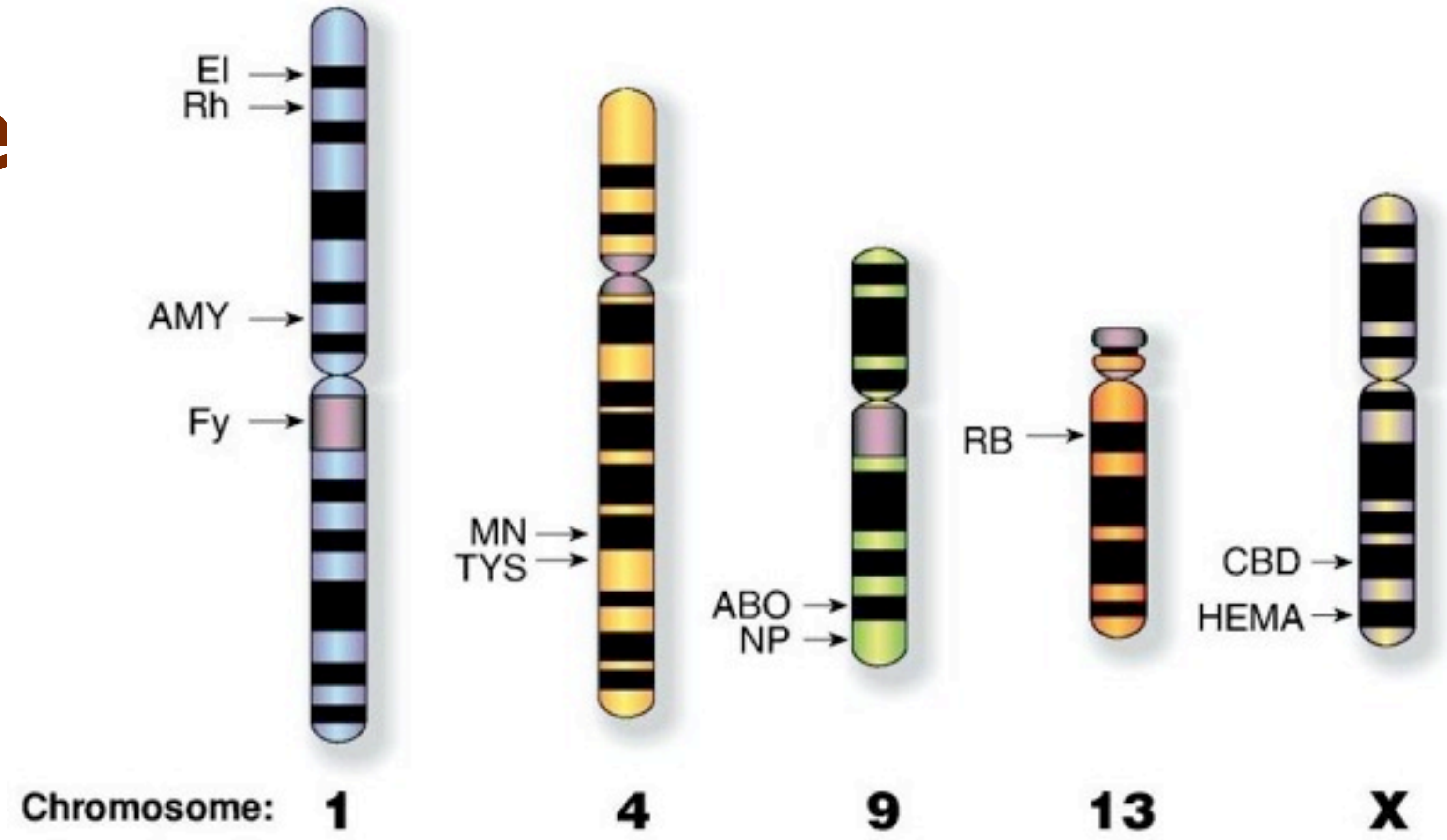
The location of an allele/gene on a chromosome is a ***locus*** *(sing.)* *or* ***loci*** *(pl.)*.



We can not see genes so we use symbolic letters to represent the genes we can not see.

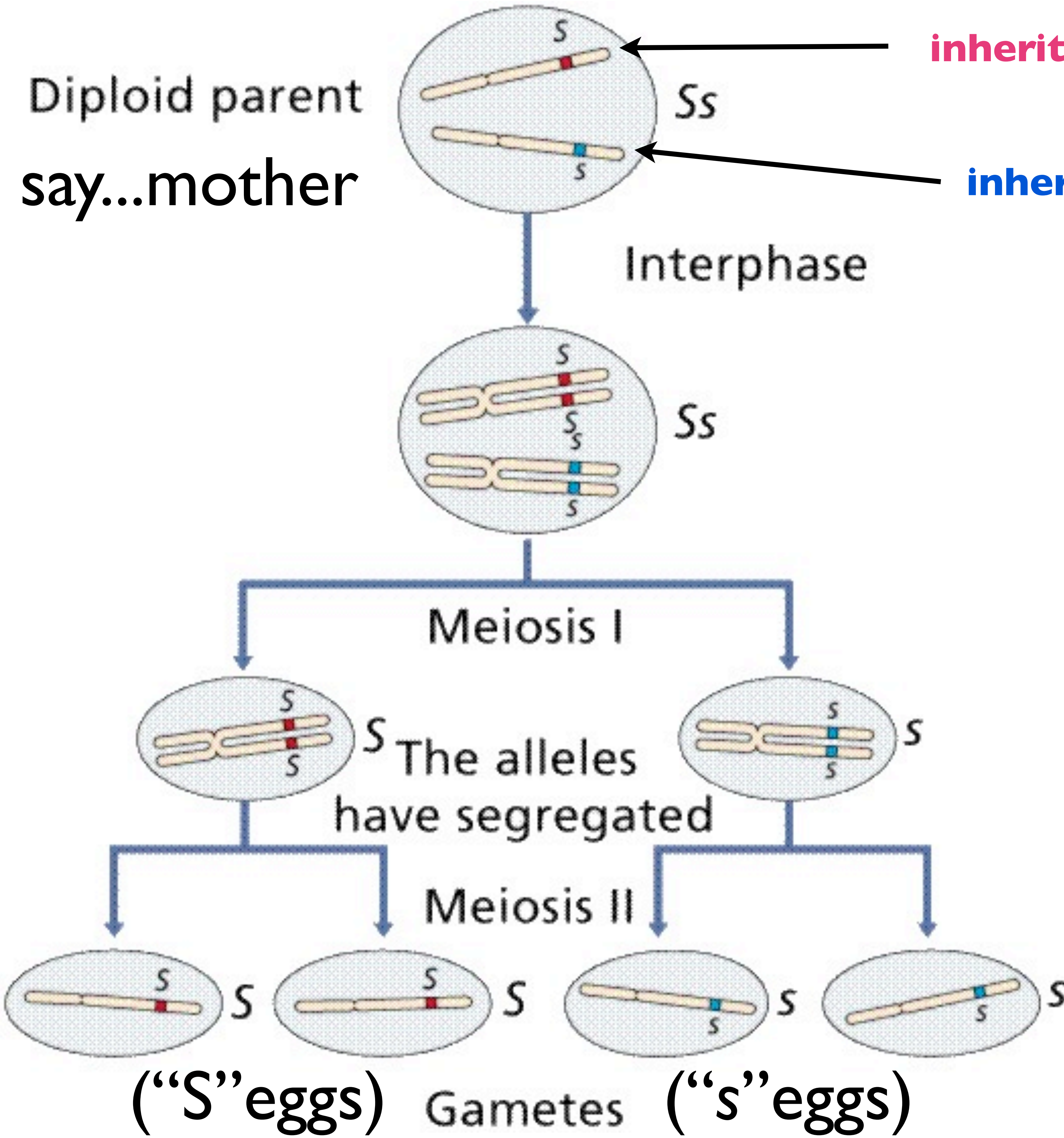


A Chromosome Carries Many Alleles/Genes



	GENE SYMBOLS
ABO	ABO blood type
AMY	Production of amylase enzyme
CBD	One form of colour blindness
EI	Shape of red blood cells
Fy	Duffy blood type
HEMA	Production of a blood clotting factor
NP	Structure of nails and kneecaps
Rh	Rhesus blood type
RB	Retinoblastoma (a cancer of the eye)
MN	MN blood type
TYS	Skin structure

Follow the chromosome, follow the traits!



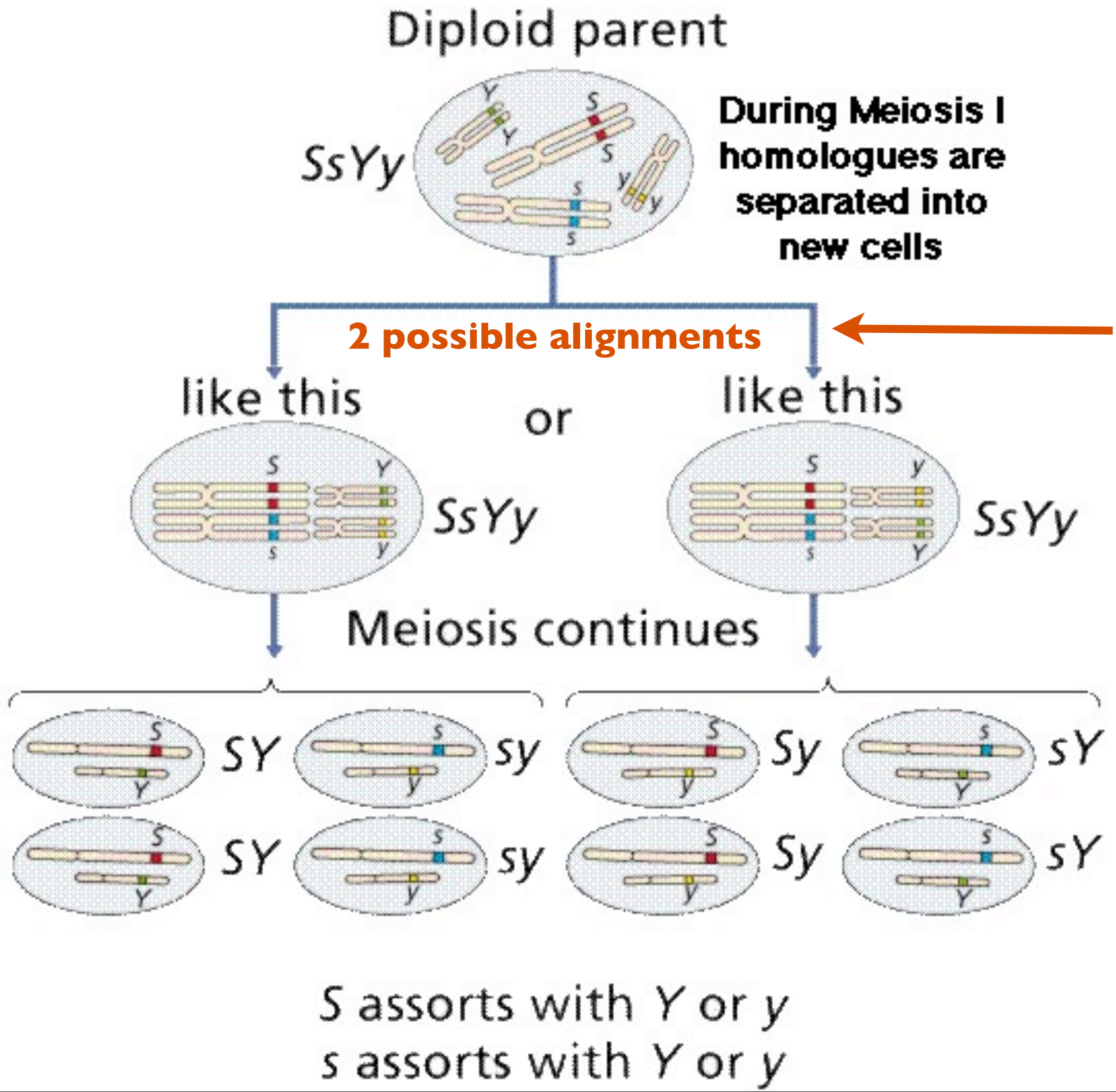
inherited from her mother

inherited from her father

Law of Segregation-
2 alleles in a pair must separate from each other into separate gametes

4 total gametes but only 2 possible kinds

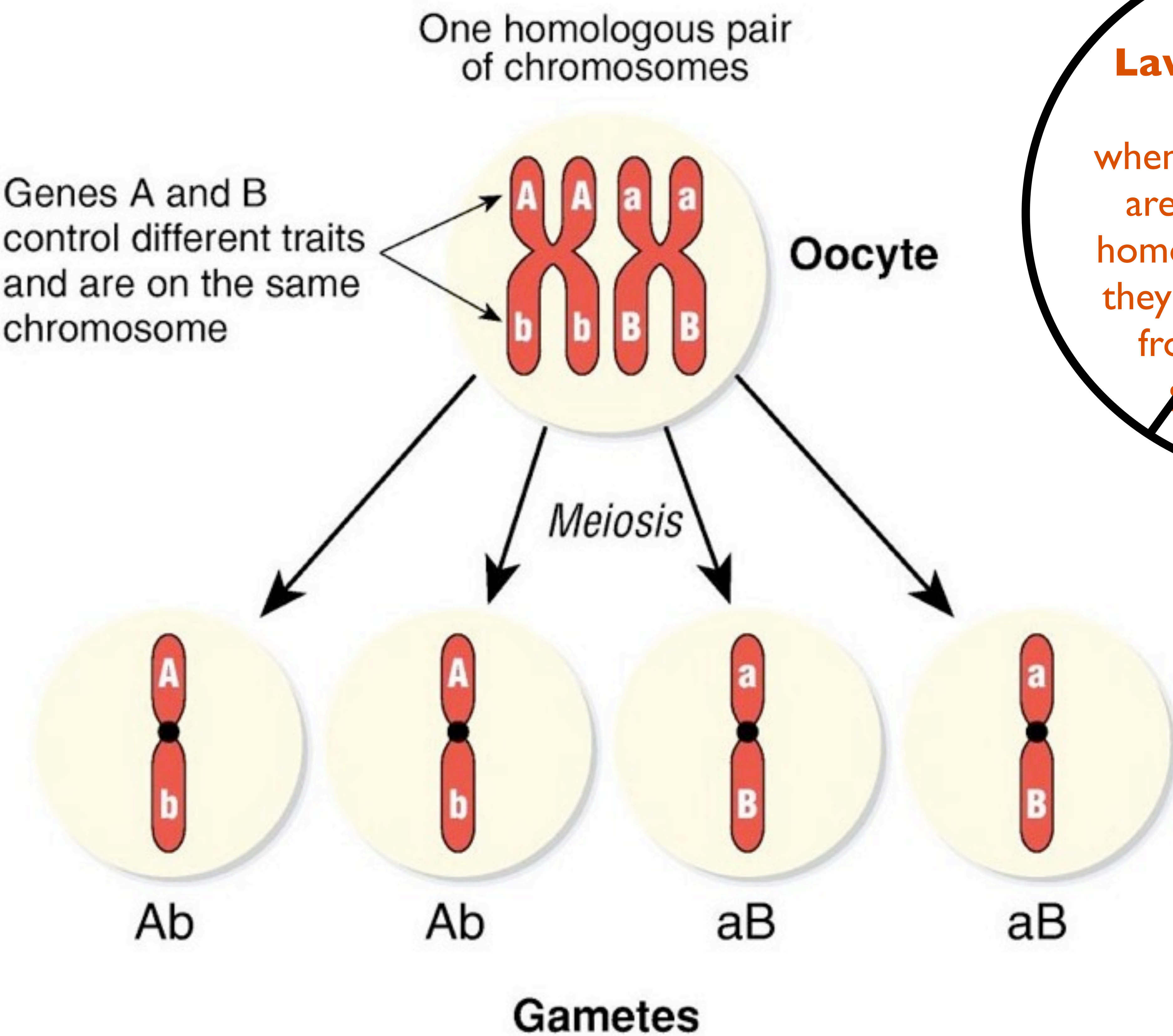
We Can Follow Two Traits Simultaneously



Law of Independent Assortment-
when a genes for two traits are on different pairs of homologous chromosomes they assort independently from each other upon gamete formation.

8 total gametes but only 4 possible kinds

We Can Follow Two Traits Simultaneously



~~**Law of Independent Assortment-**
when a genes for two traits are on different pairs of homologous chromosomes they assort independently from each other upon gamete formation.~~

WHY?

8 total gametes but only 2* possible kinds

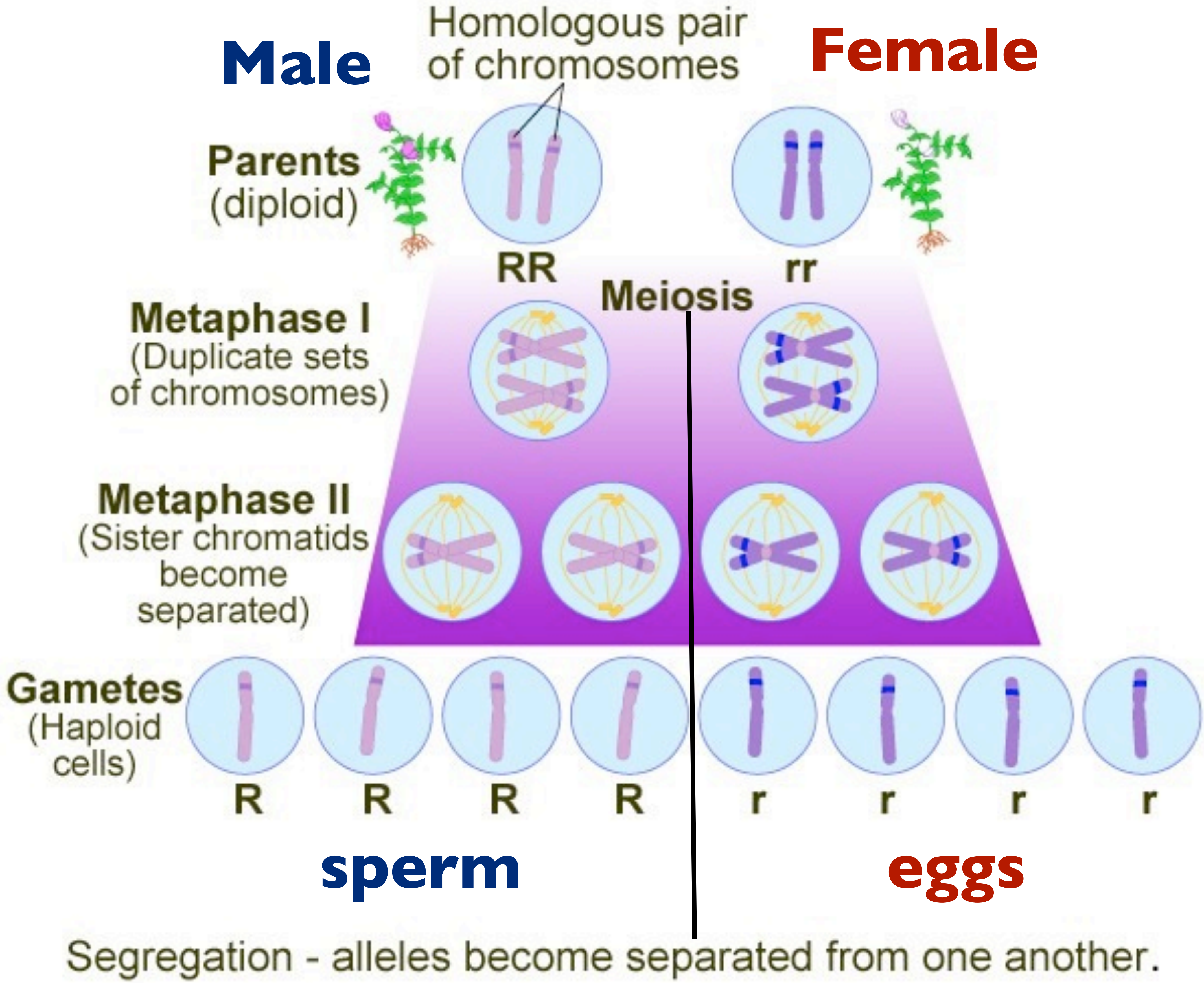
Draw the possible alignments in metaphase I.

What do all of the last three slides have in common?

We know the possible gametes, the different types of sperm or eggs that "could" be produced by the parent.

1.) We know that offspring result from the fusion of sperm with egg.

2.) **If** we know the types of possible sperm
3.) **and** we know the types of possible eggs
4.) **then** we can predict the possible offspring

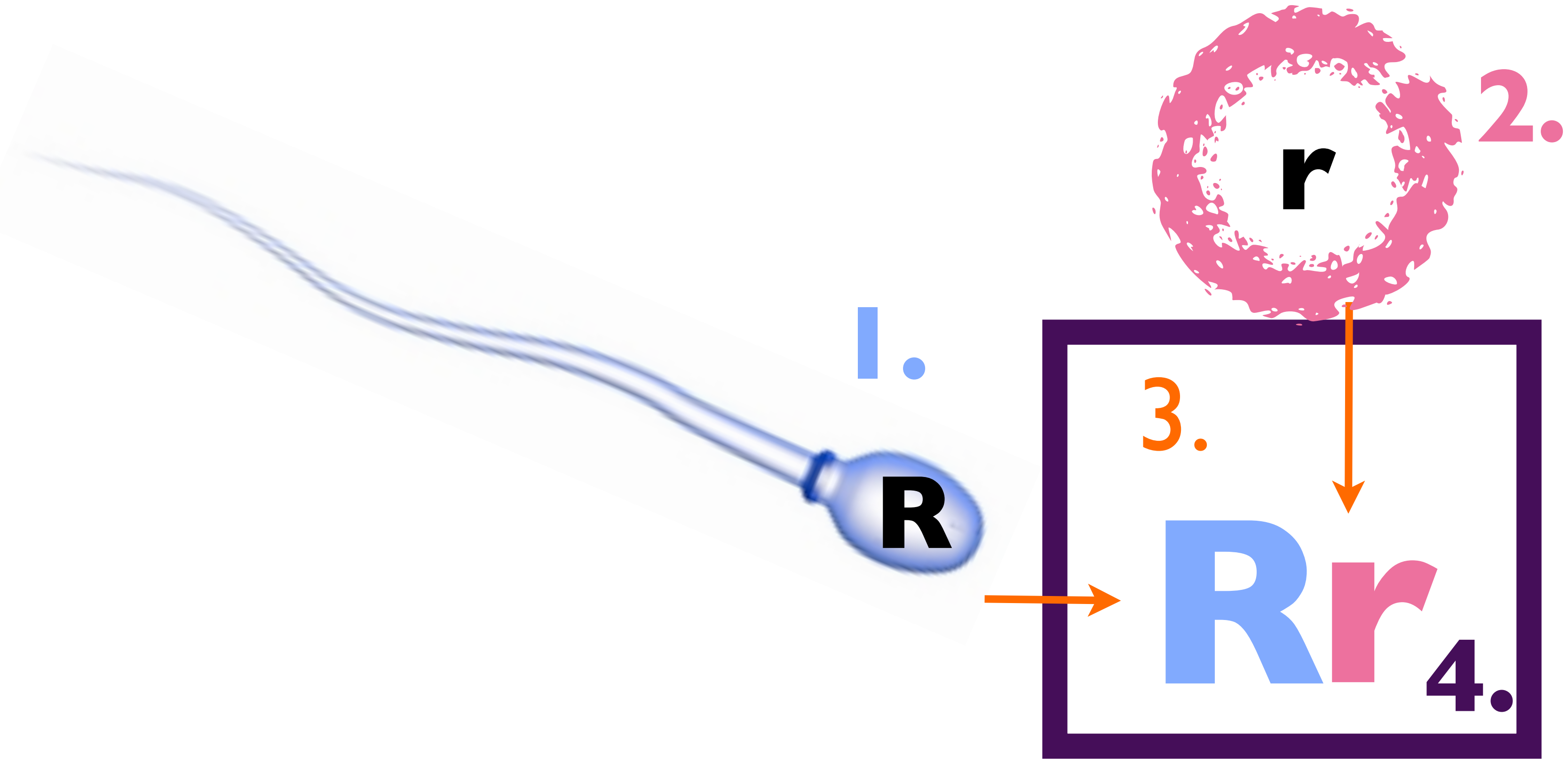

Male
Homologous pair of chromosomes
Female
Parents (diploid)
RR
rr
Meiosis
Metaphase I (Duplicate sets of chromosomes)
Metaphase II (Sister chromatids become separated)
Gametes (Haploid cells)
R R R R r r r r
sperm
eggs
Segregation - alleles become separated from one another.

1.) What are the possible sperm?

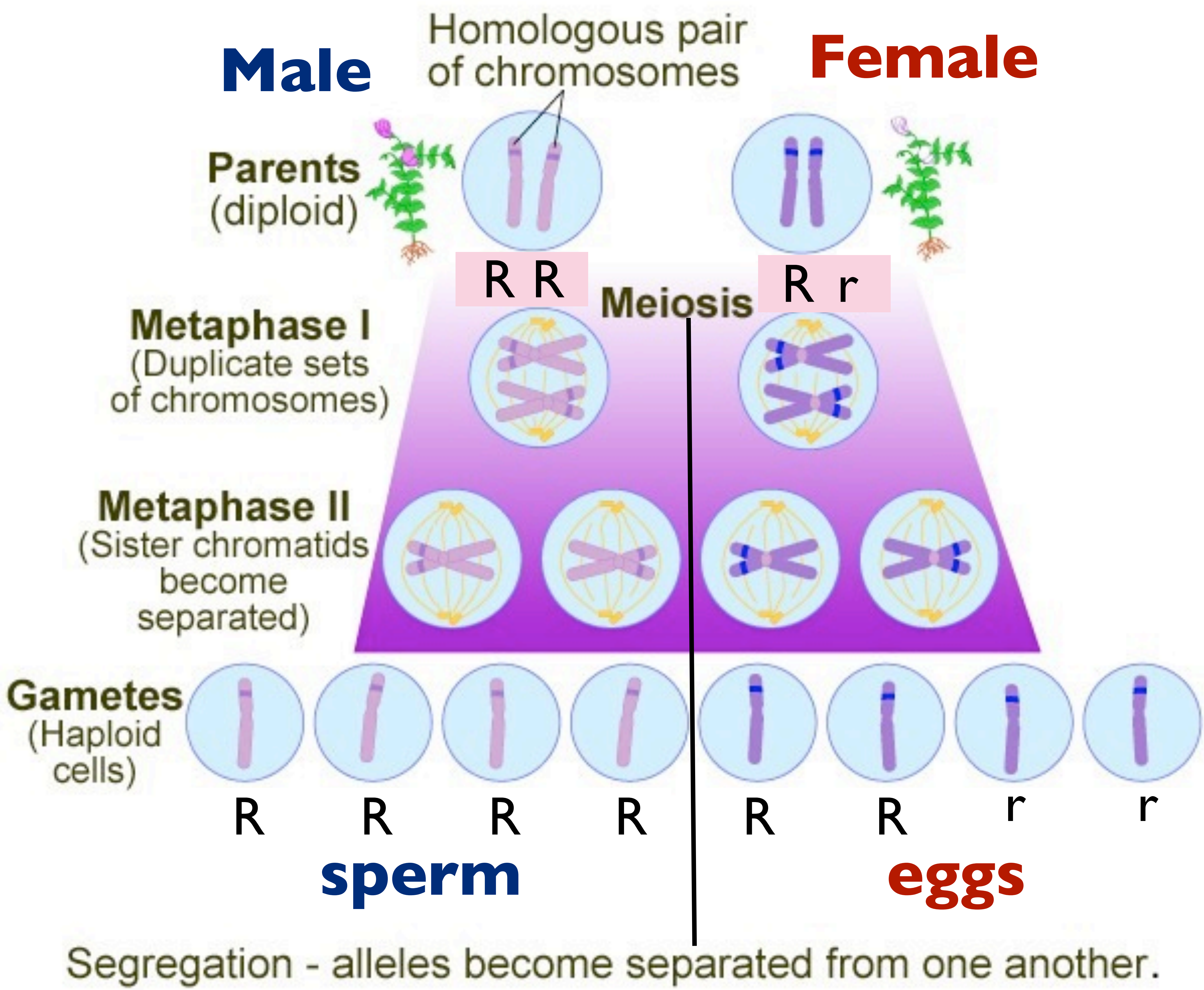
2.) What are the possible eggs?

3.) What are the possible fertilizations?

4.) What are the possible offspring?



What if we change it up?

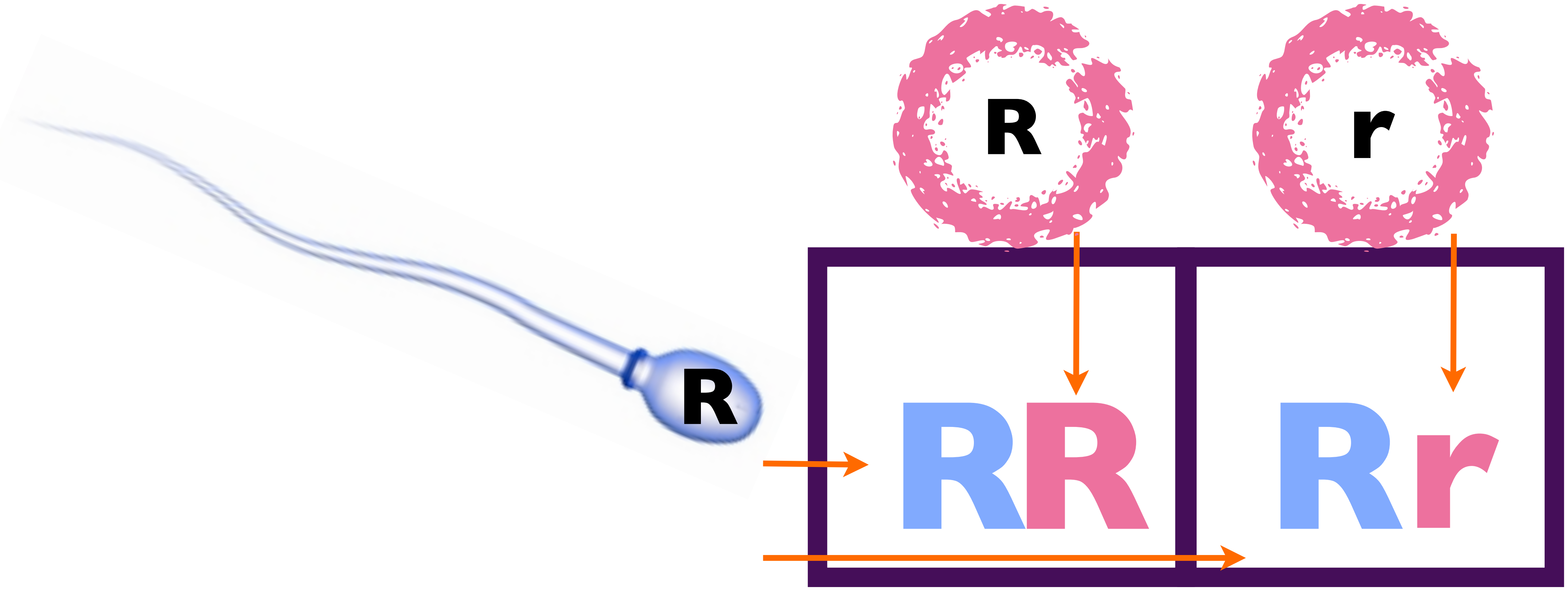


1.) What are the possible sperm?

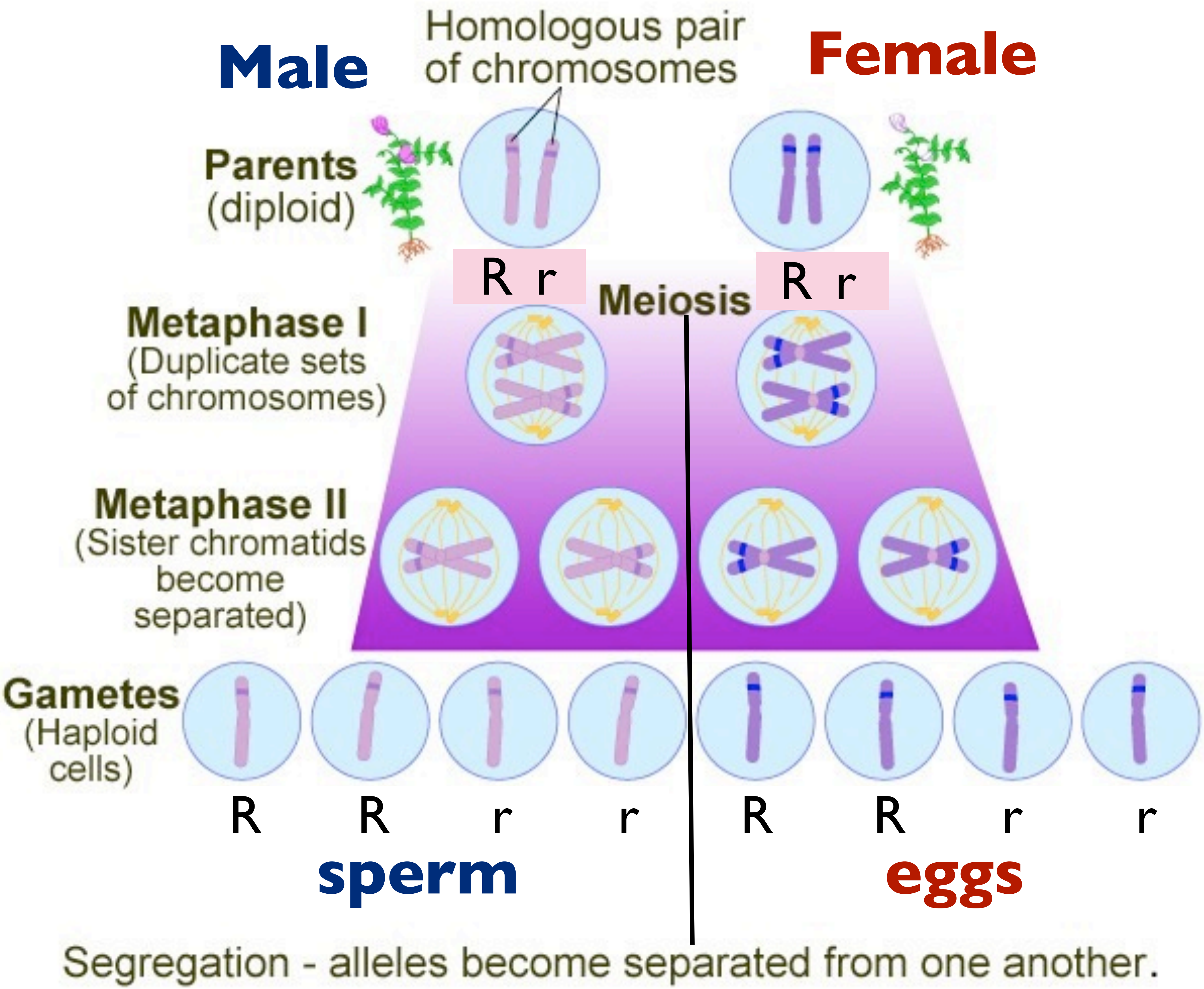
2.) What are the possible eggs?

3.) What are the possible fertilizations?

4.) What are the possible offspring?



What if we change it up again?

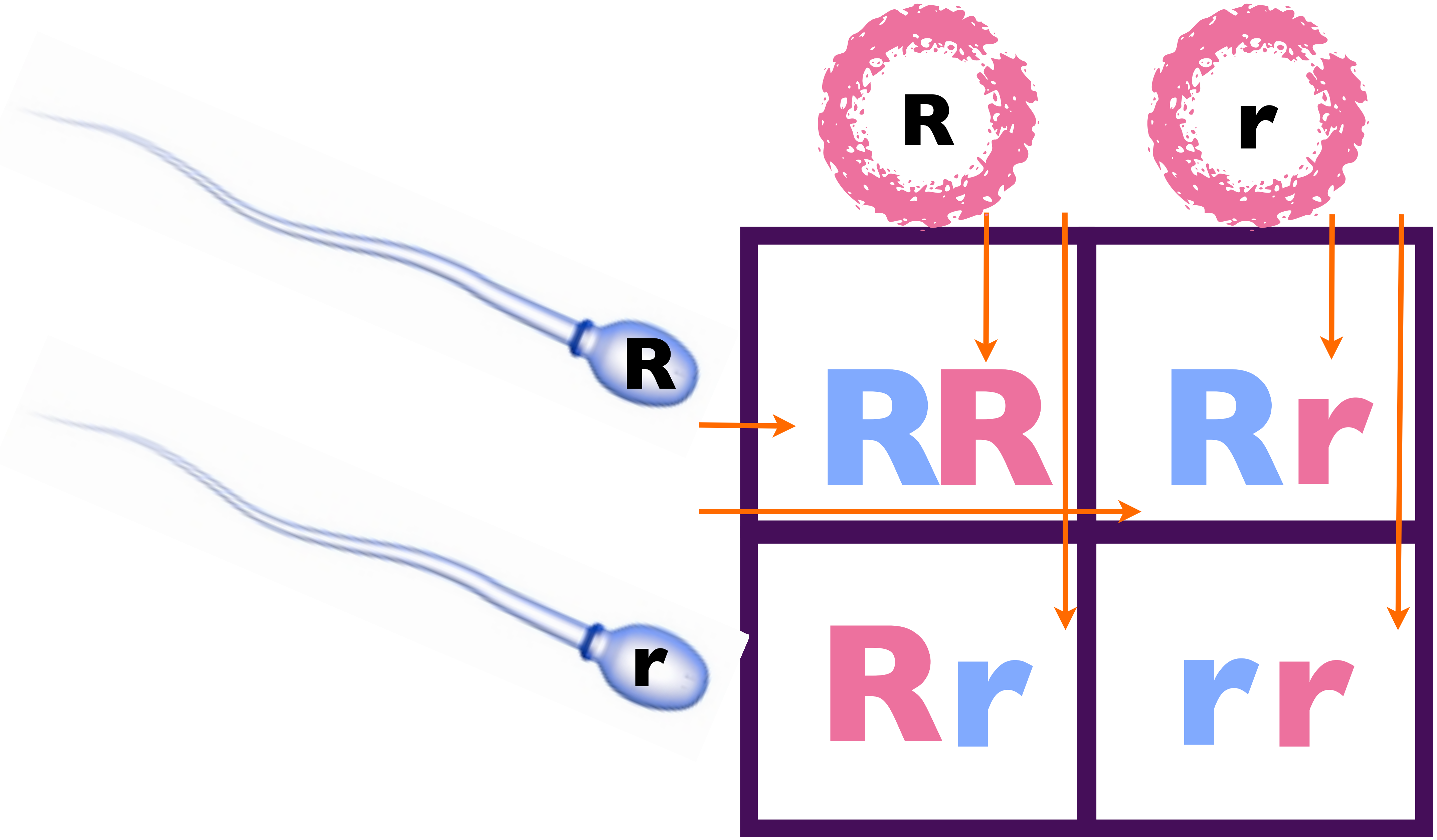


1.) What are the possible sperm?

2.) What are the possible eggs?

3.) What are the possible fertilizations?

4.) What are the possible offspring?



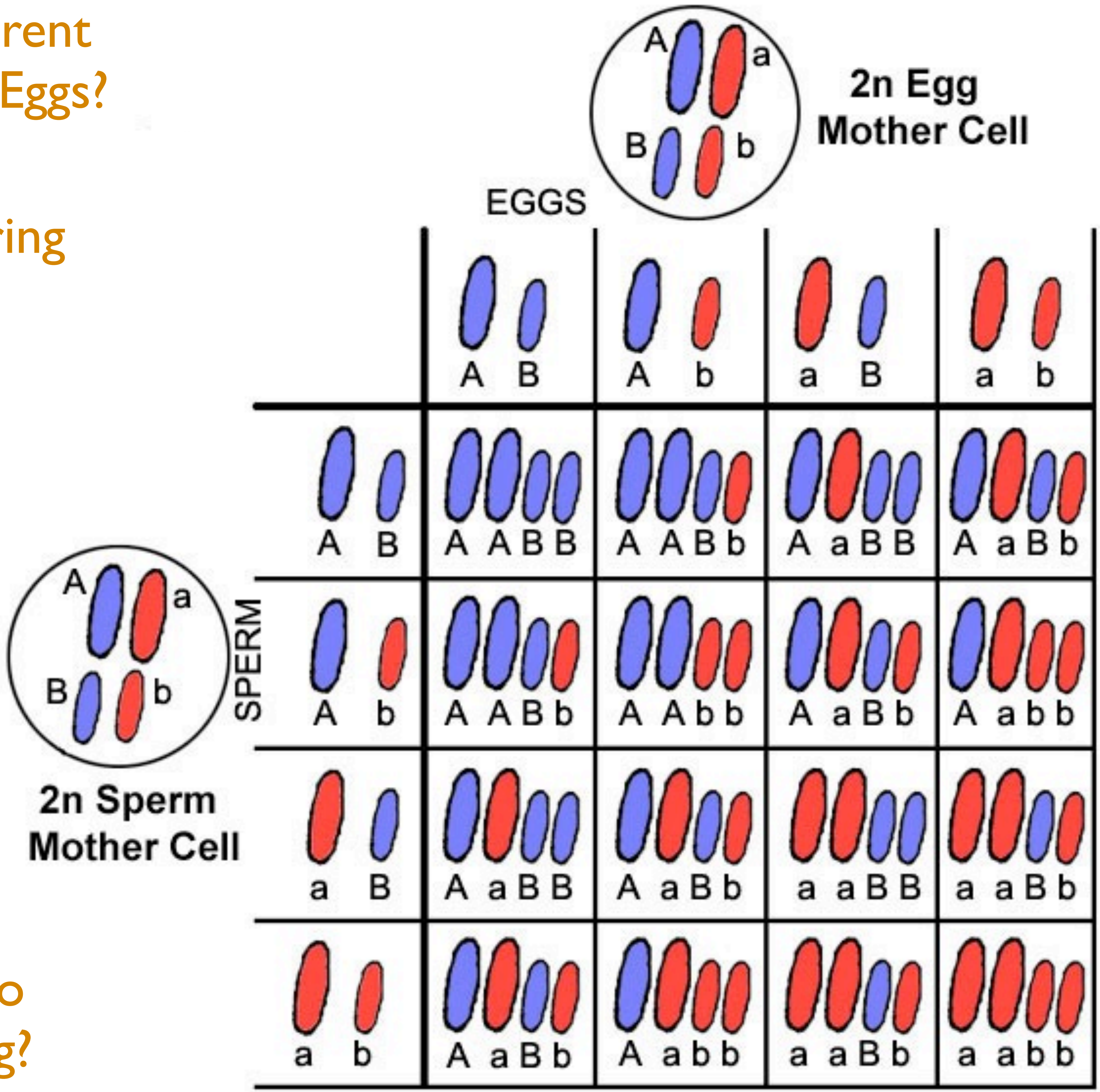
How many different types of sperm? Eggs?

How many offspring are depicted?

How many different offspring are represented?

What patterns do see in all gametes?

What patterns do see in all offspring?



In Summary

- **Genetics**- the study of *heredity* and hereditary variation.
- **Heredity**- the transmission of *traits* one generation to another.
- **Trait**- one or more detectable variants in a genetic *characteristic*.
- **Characteristic**- an observable feature that may vary among individuals.
- **Gene**- a discrete unit of hereditary information consisting of a specific nucleotide sequence in DNA that is responsible for characteristics.
- **Chromosome**- a cellular structure carrying genetic information (genes)

Transition to Genetics

BUT, here again is the key point! Chromosomes carry genes, genes control traits and genetics studies the transmission of these traits.

Understanding chromosomes transmission from one generation to another is an essential piece of knowledge in the "genetic puzzle"

It is amazing to think that our knowledge of genetics was born in a garden before we knew about chromosomes, genes and DNA.