Chapter 5 Topics

1. Animal Life Cycles
2. Structure of DNA in Eukaryotes and Prokaryotes!
3. Replication of DNA (copying of DNA)
4. Making proteins!
    - Transcription-going from DNA to mRNA
    - Translation-going from mRNA to protein

5. Gene regulation!
6. Sizing up the Genome
7. **Mutations**
8. Mitosis vs Meiosis (sexual reproduction)
9. Mendel and Punnett Squares
10. **Getting more real….**

7. Mutations!

Effects depend on where they are in body!

What are somatic mutations?

What are germline mutations?

How do effects differ depending on whether somatic or germline?

What causes mutations?

- Mistakes during replication or copying (many of these are fixed) (Ex. mitosis or meiosis)
- Environment (radioactive particles zooming thru us and UV radiation)

Not all mutations are harmful…maybe be neutral or beneficial.

Mutations are the **ultimate** source of all genetic variation among individuals in populations so super important to evolution (no variation, no evolution)!

(Remember Darwin really emphasized differences between individuals.)

May occur in coding region or in a non coding region.

May make a protein work better or work worse, make it more or less active, or change what it can do (a change in structure)

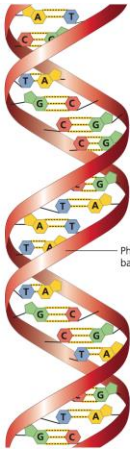
May change how much of the protein is made, where it is made (a change in regulation)

Here is a sequence of bases…(T=Thymine, G=Guanine etc..)

What are some things that could go wrong when it is being copied or replicated..

Think both small scale and big scale!

T G C A T T G C G T A G G C



Remember this is one half of the double helix.

Original T G C A T T G C G T A G G C

Mutation T G C A T T C C G T A G G C

T G C A T T T A G G C

T G C A T T C C G T A G G C

C C G

T G C A T T C C G T A G G C

T G C A T T T A G G C

Match each mutation with a term:

- Deletion
- Insertion
- Duplication
- Point mutation
- Inversion

T G C A T T G C G T A G G C

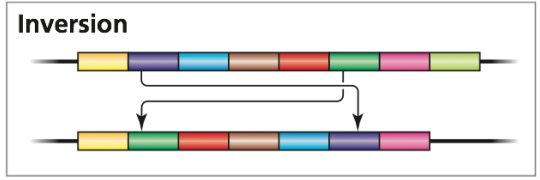
T G C **T G C** A T T G C G T A G G C

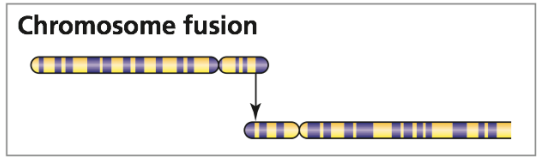
T G C A T T G C G T A G G C

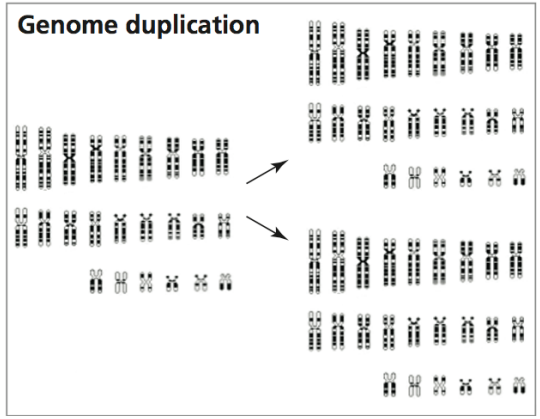
C G T A T T G C G T A G G C

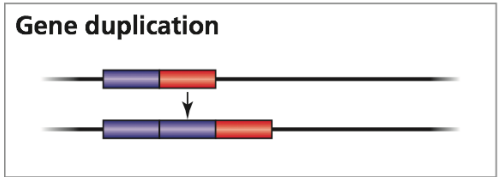
What do I mean by “big” and “small”?

- Point mutation is small
- Insertions can be big or small
- Deletions can be big or small
- Duplications can be big or small
- Inversions can be big or small









And….

- Two chromosomes can fuse
- You may have an extra chromosome
- You may lose a chromosome
- Your whole genome may be duplicated

Consider how a **single base insertion** affects the reading frame…..

T G C A T T G C G T A G G C C

T G C A T A T G C G T A G G C C

How might a **point mutation** affect the reading frame?

Remember how the “third position” is more flexible???

https://pmc.ncbi.nlm.nih.gov/articles/PMC8415378/

Just for fun!

Genome Res. 2021 Sep;31(9):1513–1518. doi: 10.1101/gr.271809.120

Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity

Jakob M Goldmann [1,2], Juliet E Hampstead [1,2], Wendy SW Wong [3], Amy B Wilfert [4], Tychele N Turner [4], Marianne A Jonker [5], Raphael Bernier [6], Martijn A Huynen [7], Evan E Eichler [4,8], Joris A Veltman [9], George L Maxwell [10], Christian Gilissen [1,2]

De novo mutations (DNMs) are drivers of genetic diversity and evolution and can also cause severe diseases, such as intellectual disability, autism, and schizophrenia (Veltman and Brunner 2012). The number of single nucleotide DNMs per individual genome ranges between 30 and 80 (Gilissen et al. 2014) and is correlated with the age of the parents at conception (Kong et al. 2012; Goldmann et al. 2016; Wong et al. 2016; Jónsson et al. 2017). Aging of fathers adds one DNM per year, while aging of mothers adds one DNM every four years. However, parental age at conception explains only part of the observed variation in DNM count between individuals, raising the possibility that other factors can affect the number of DNMs an individual carries. Such factors could be endogenous, such as genetic variation in genes involved in DNA repair (Goldberg et al. 2021), or could be of external origin, like ionizing radiation (Adewoye et al. 2015; Holtgrewe et al. 2018) and environmental pollutants (Ton et al. 2018; Beal et al. 2019). Studies of multi-offspring families have also suggested that the paternal age effect may differ significantly between families, where the mean yearly increase in DNMs per offspring with age of the fathers can vary from 0.2 to 3.2 DNMs per year (Rahbari et al. 2016; Sasani et al. 2019).

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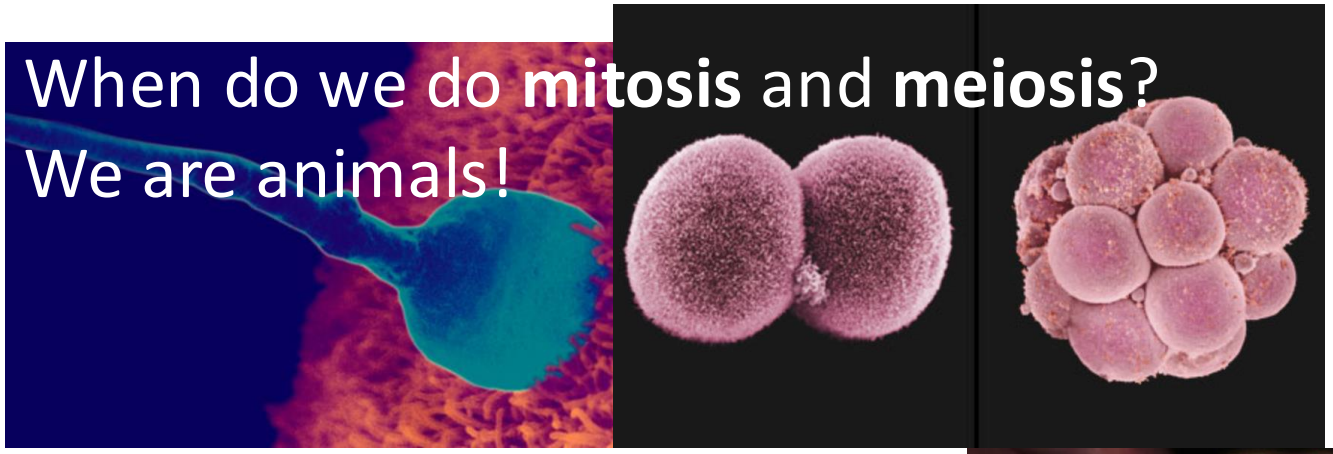
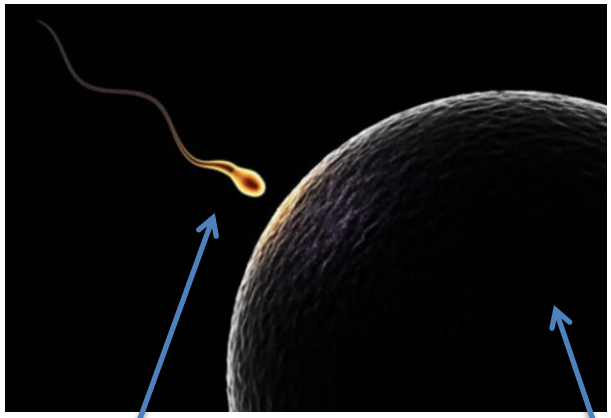
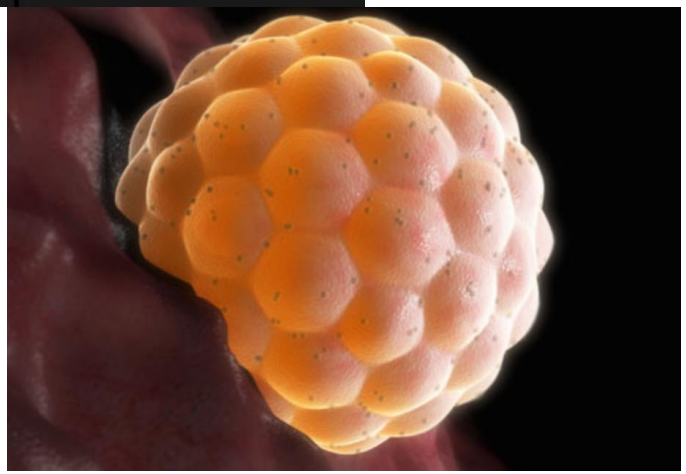
8. **Mitosis vs Meiosis (sexual reproduction)**
9. Mendel and Punnett Squares

When do we do **mitosis** and **meiosis**?

We are animals!





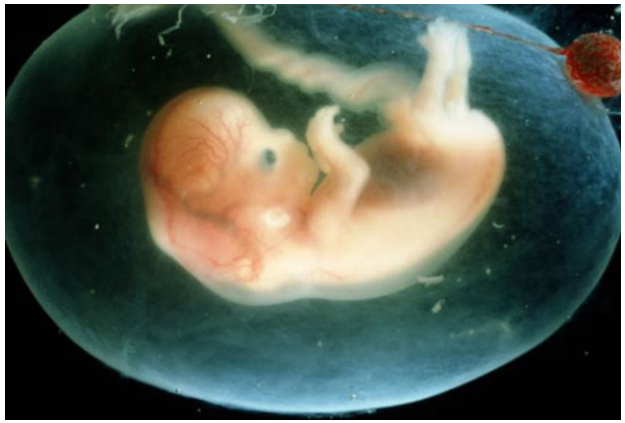


For your Review!

- Fertilization
- Zygote
- Mitosis
- Meiosis
- Gametes
- Haploid
- Diploid

Egg

Sperm



When do we do **mitosis** and **meiosis**?
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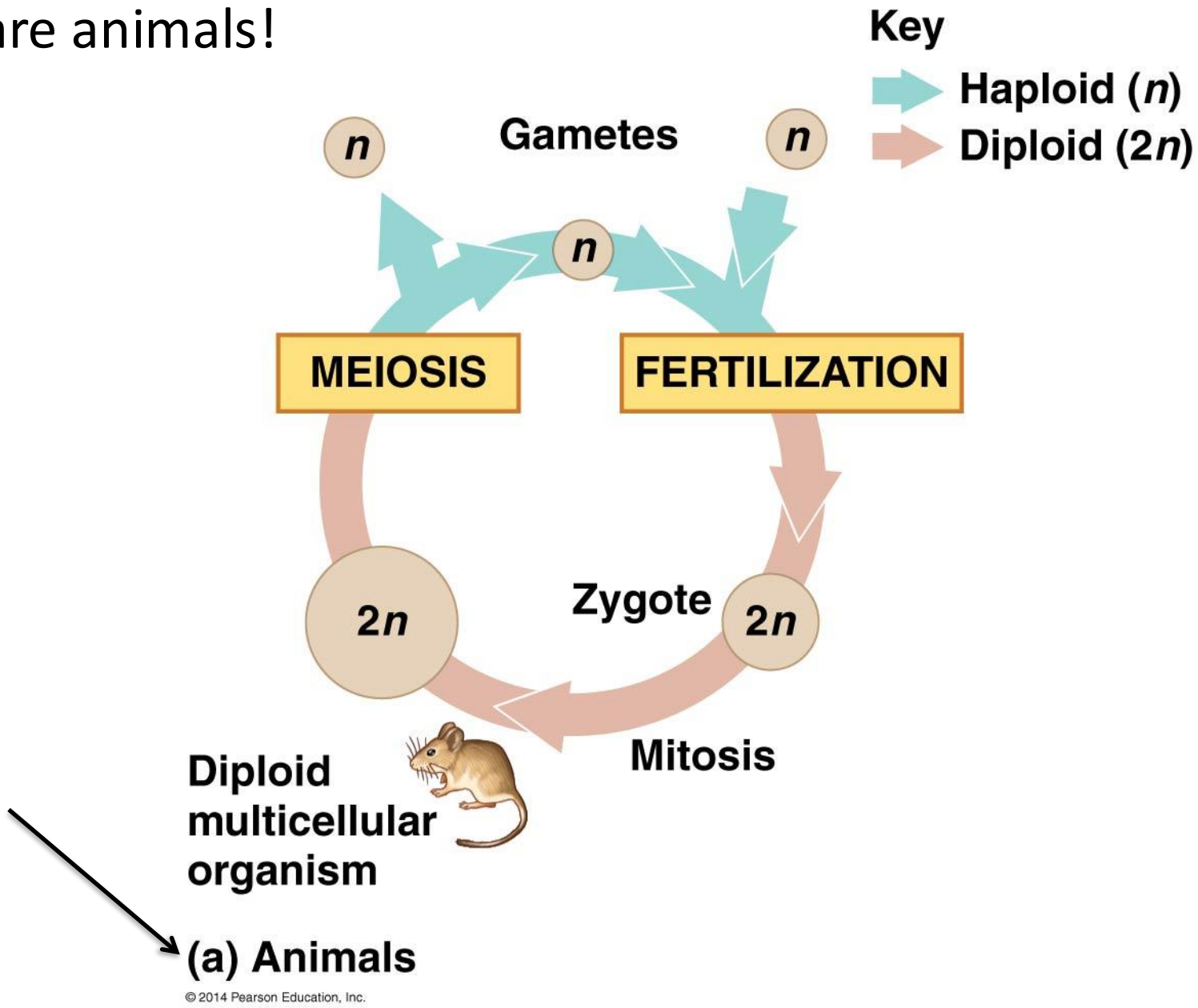
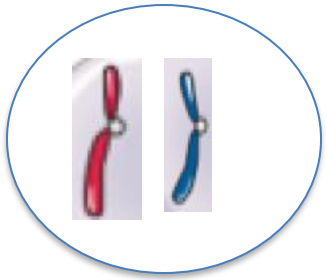


Mitosis

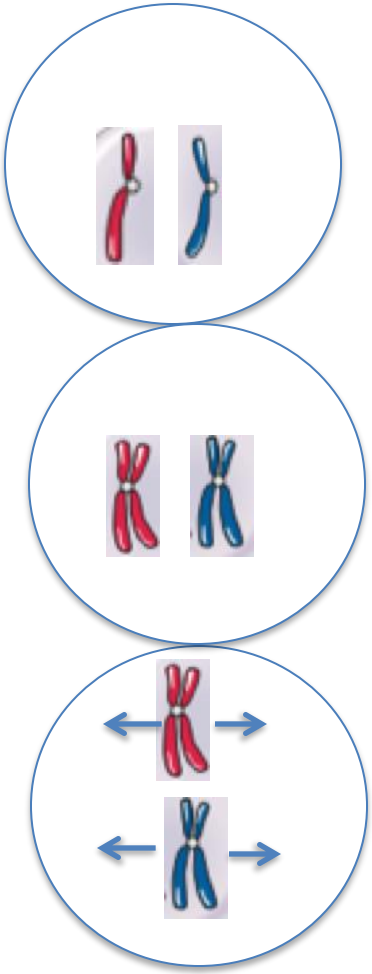
Meiosis



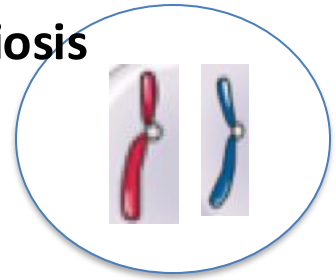
Once cell with one homologous pair of chromosomes!

We did Mitosis already…

Mitosis

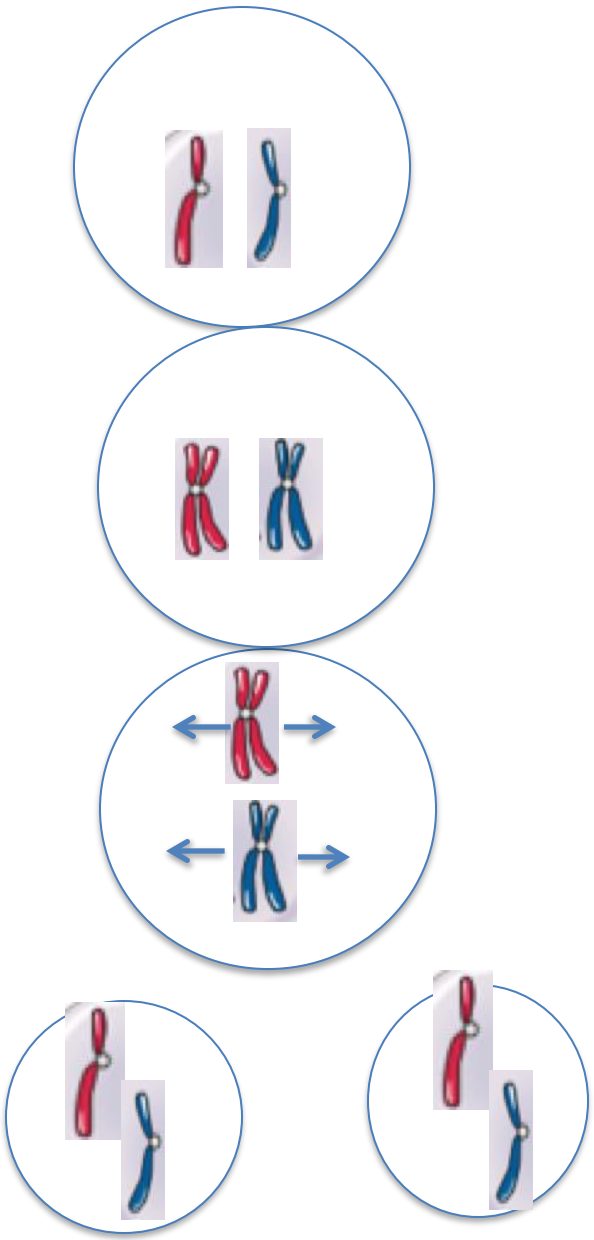


Meiosis

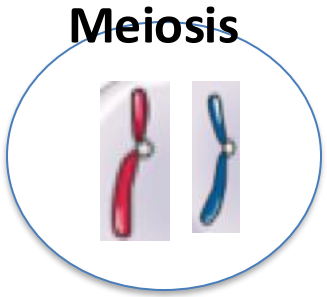


Are these sister chromatids separating from one another or homologous chromosomes?

Mitosis



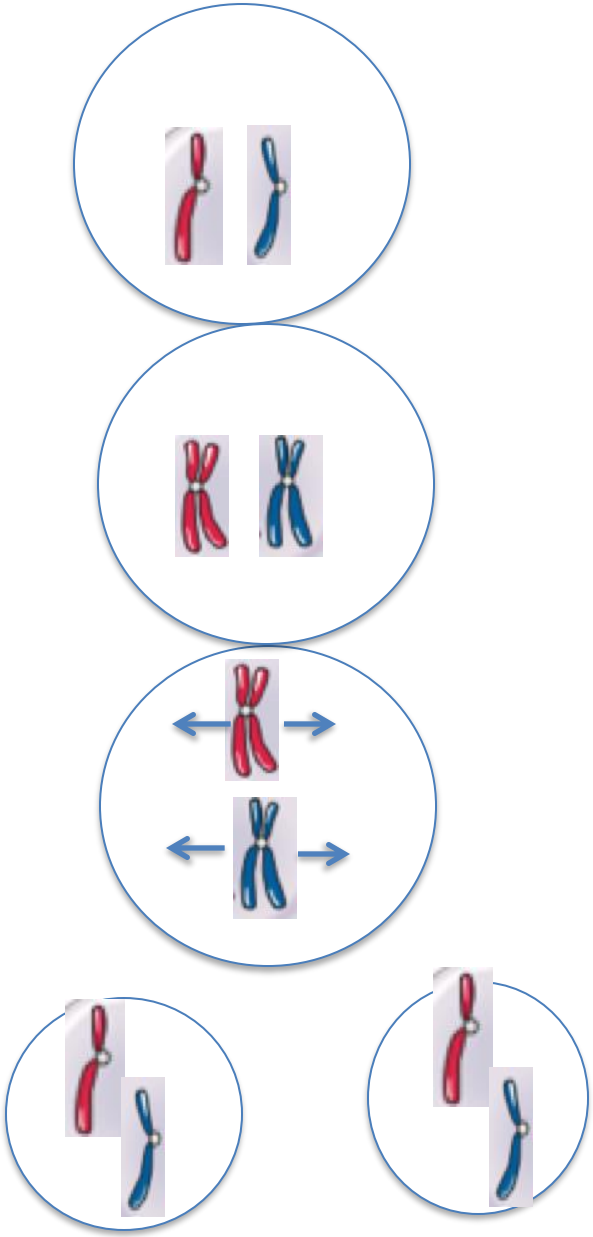
Meiosis



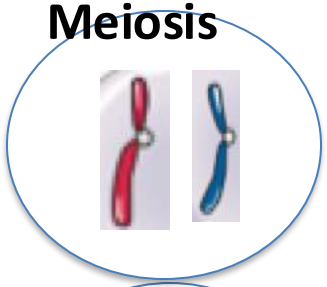
Mitosis
One cell to two cells
Diploid to diploid

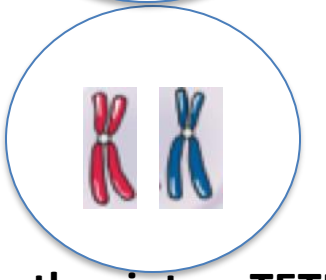
now lets make some gametes through meiosis

Mitosis

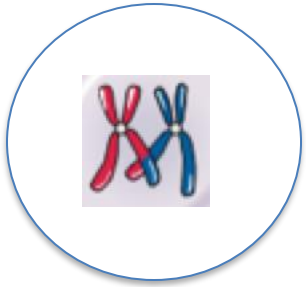


Meiosis

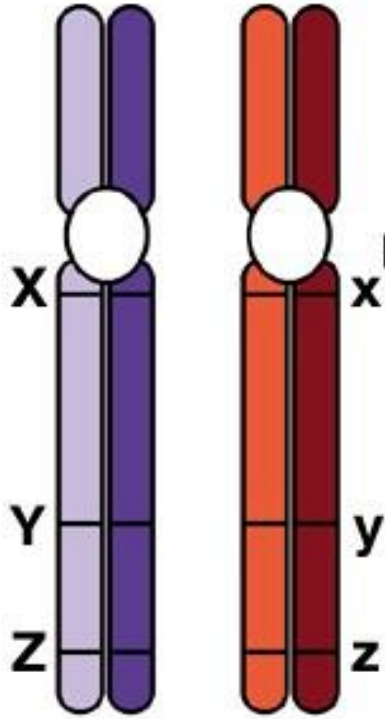




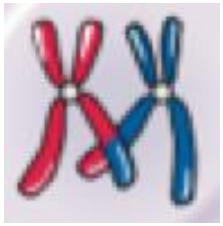
All 4 get together into a TETRAD to do what?????



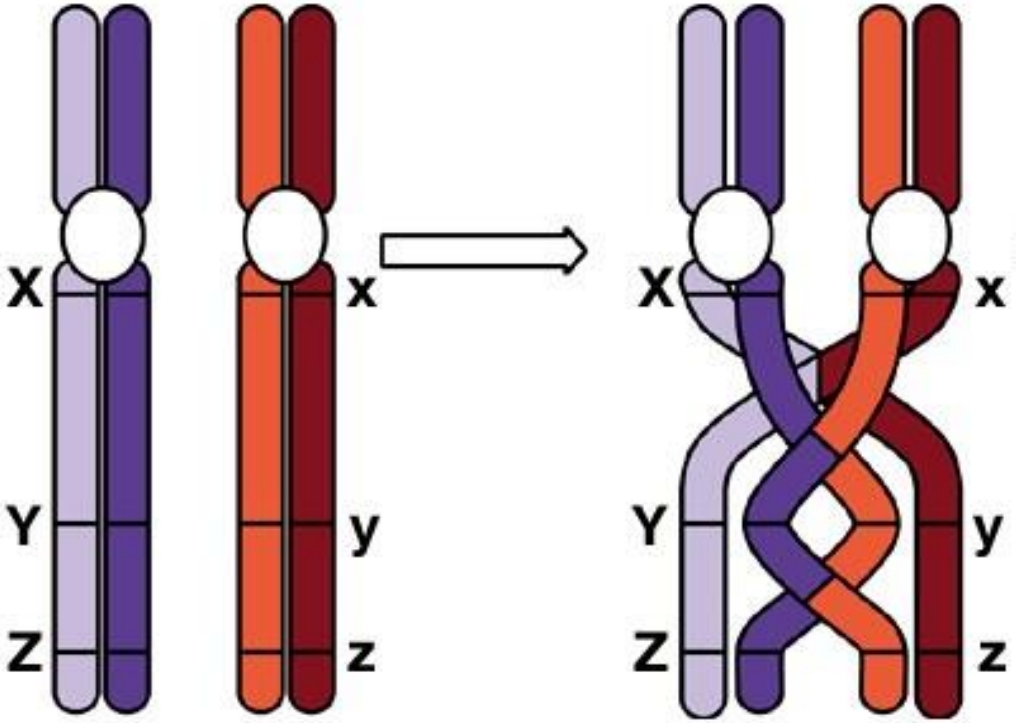
Crossing over during meiosis

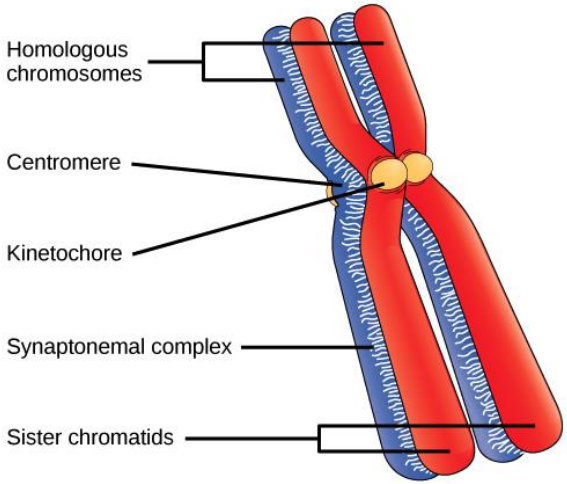


Crossing over=recombination

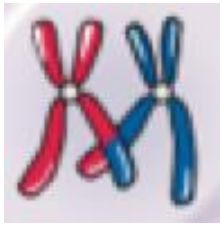


Crossing over during meiosis

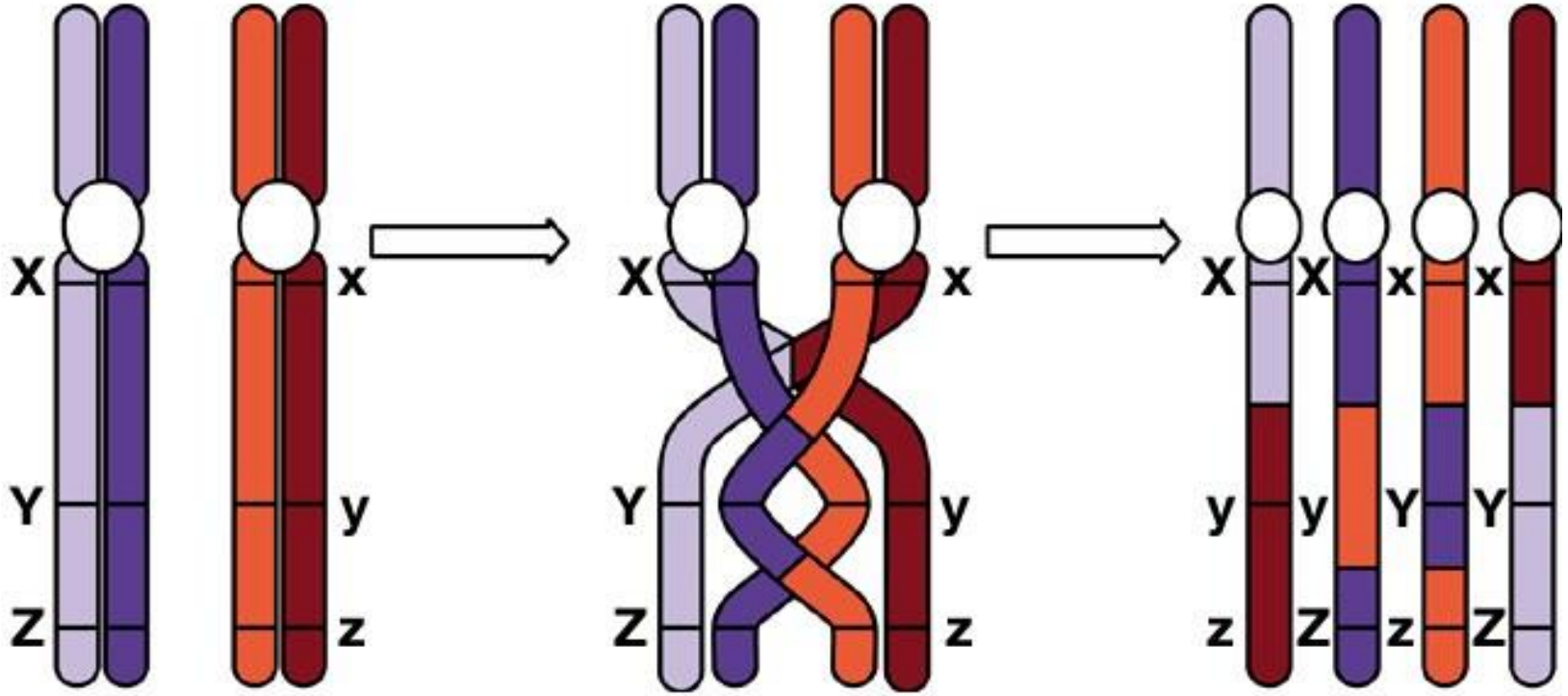




Crossing over=recombination

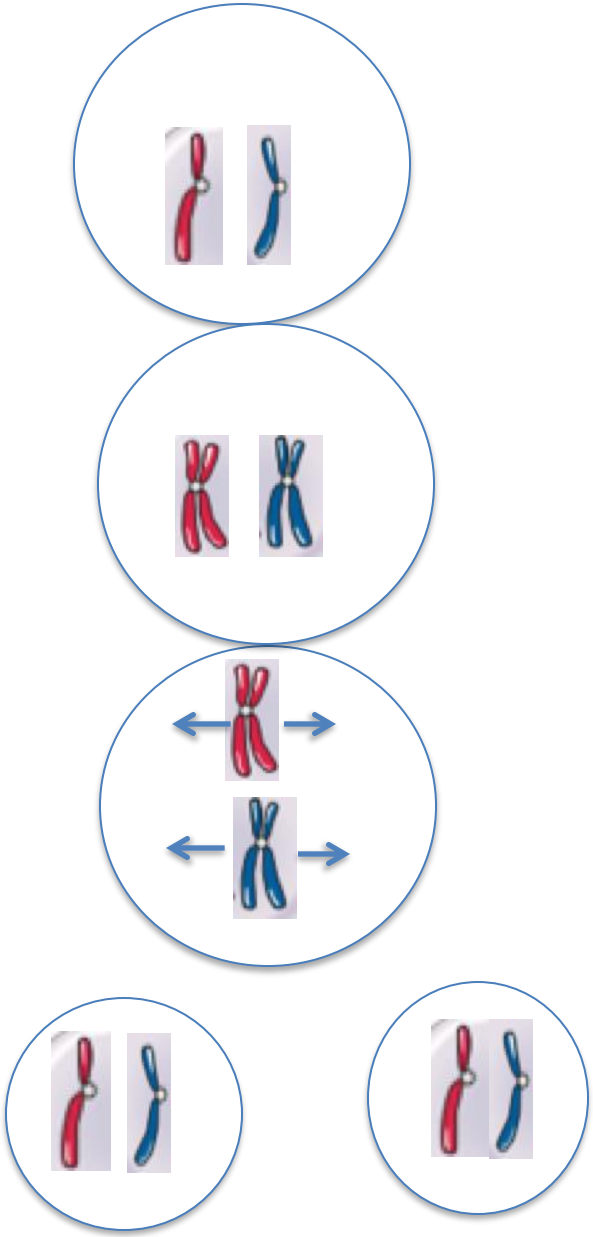


Crossing over during meiosis

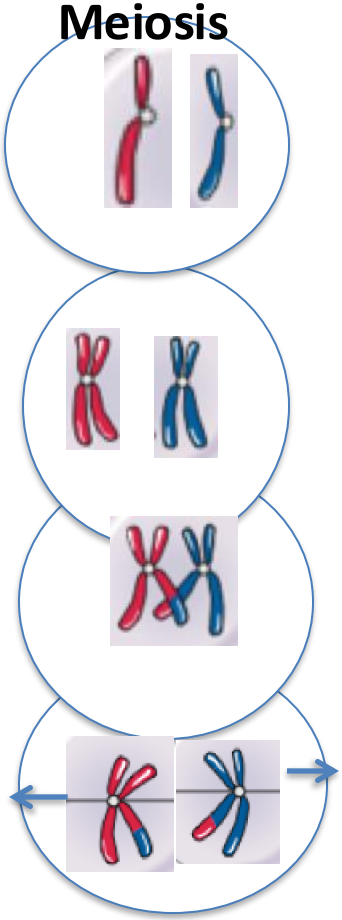


Crossing over=recombination

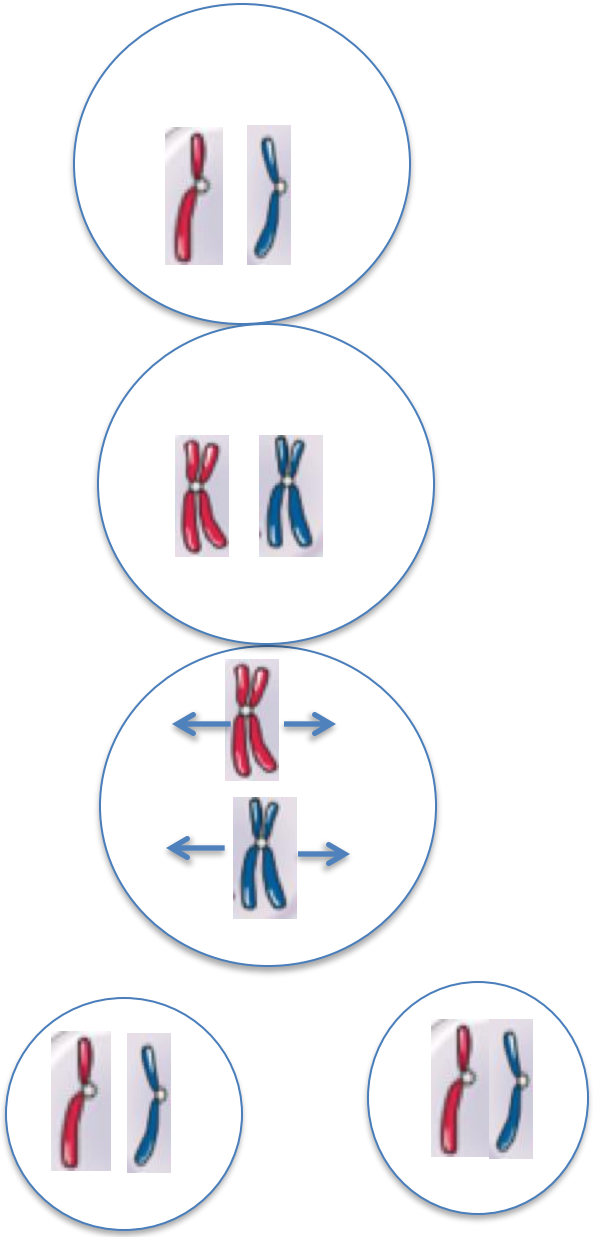
Mitosis



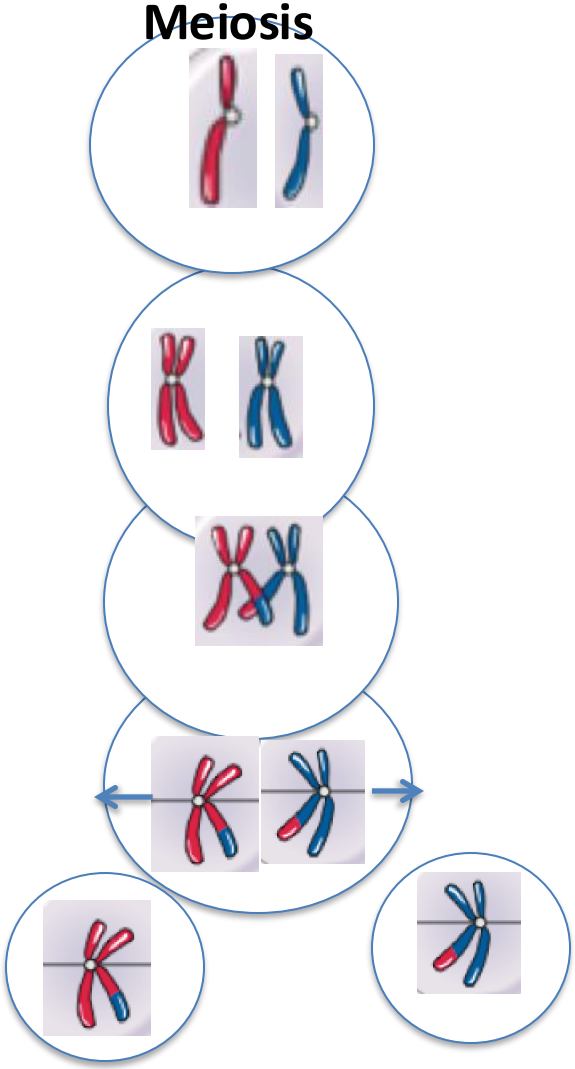
Meiosis



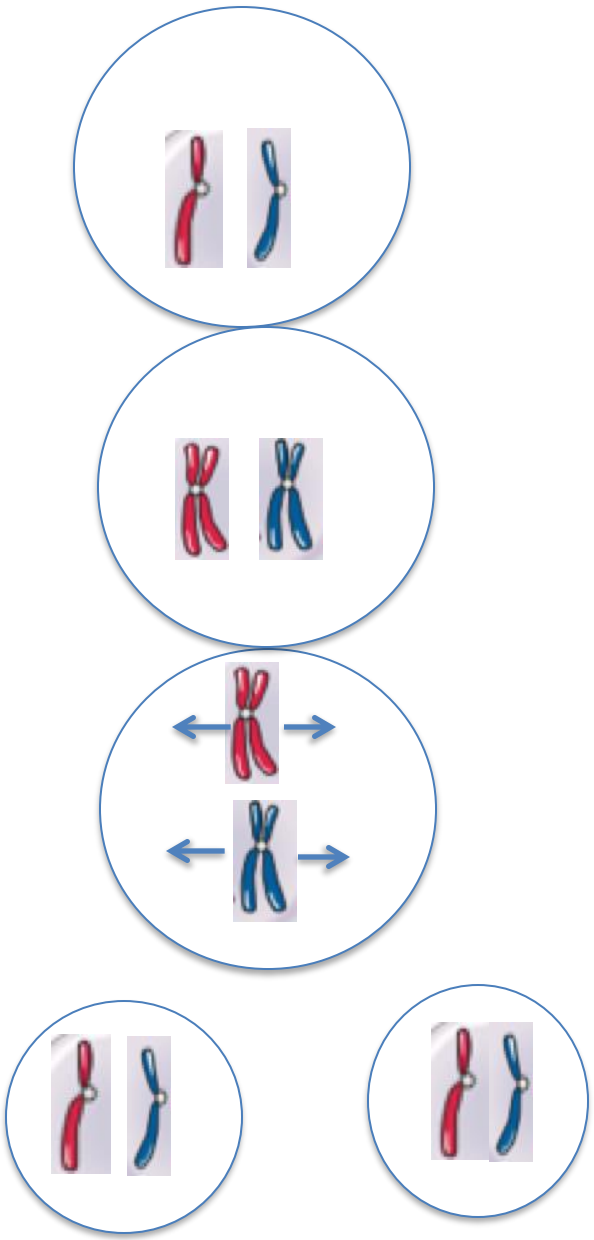
Mitosis



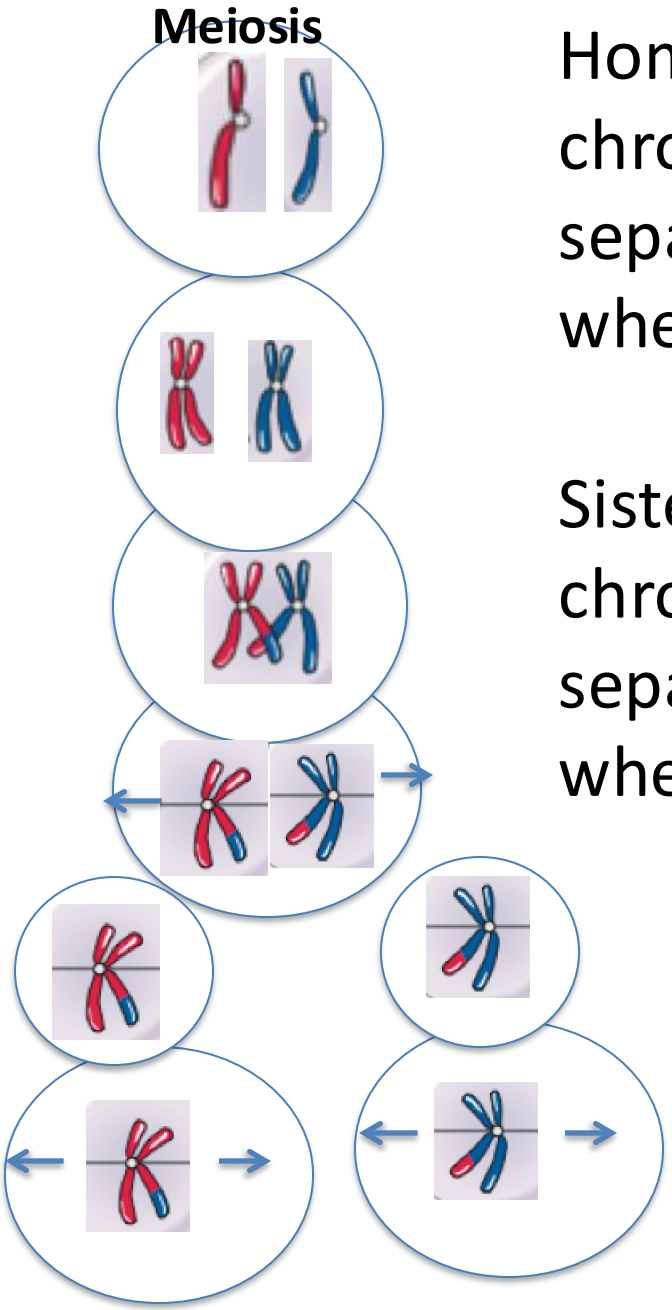
Meiosis



Mitosis



Meiosis



Homologous chromosomes separate where?

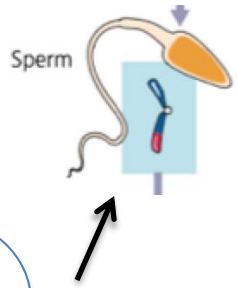
Sister chromatids separate where?

Mitosis

Meiosis

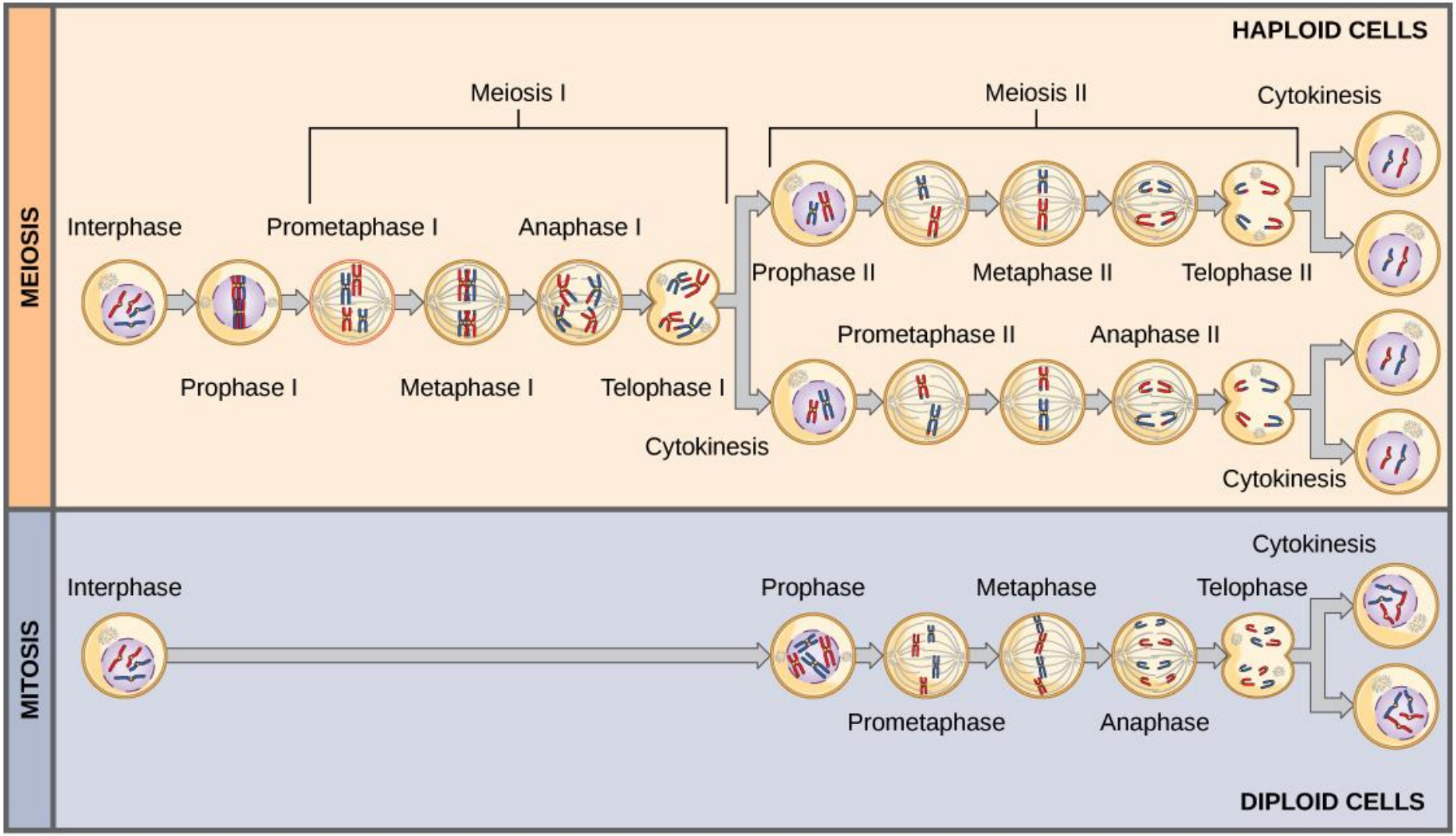
For Meiosis…
Homologous chromosomes separate where?

Sister chromatids separate where?



OVERVIEW!

Helpful? Not Helpful?



Remember that there are typically many other chromosomes all doing what we just showed with one chromosome simultaneously!

This is complicated and many things can go wrong!

Remember all the different kinds of “mutations”!

Ultimately ALL variation comes from MUTATION but….

Meiosis generates genetic variation IN EUKARYOTES by mixing up genomes.

It does this through

- Recombination (crossing over-swapping bits of one parent's chromosome with bits of other parent's chromosome)
- Independent assortment (idea that one parent's chromosomes as a group do not all head together to one "side" of cell and end up in a gamete together)
- Fusion of any individual random egg and individual random sperm (every gamete from a single individual is different)

Meiosis had to have evolved in Eukaryotes in order for them to sexually reproduce because to have egg and sperm come together and fuse you have to REDUCE these cells to the haploid state (n).

We assume it evolved in part from a modification of mitosis.

But we also know **the genes that are active during meiosis are the same genes** that are active during Horizontal or Lateral Gene Transfer in bacteria!!!

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