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!

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)
- Or a combination of these two!! (UV exposure during replication=risky)

Not all mutations are harmful…may 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 within a species.)

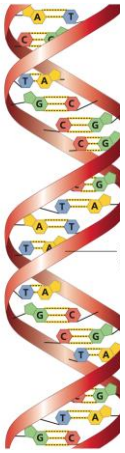
May occur in coding region or in a non coding region.

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 of the the protein).

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

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

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



Think both small scale and big scale!

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

Remember when you see a string of bases like this it is one half of the double helix.

Match each mutation with a term:

- Deletion
- Insertion
- Duplication
- Point mutation
- Inversion

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

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

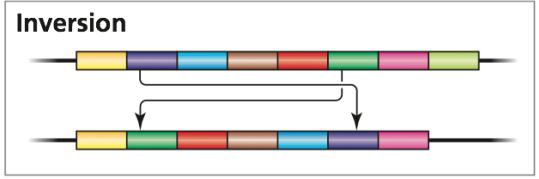
T G C A T T G C G T A G G C

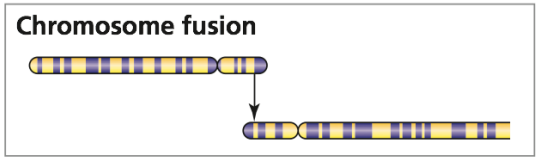
C G T A T T G C G T A G G C

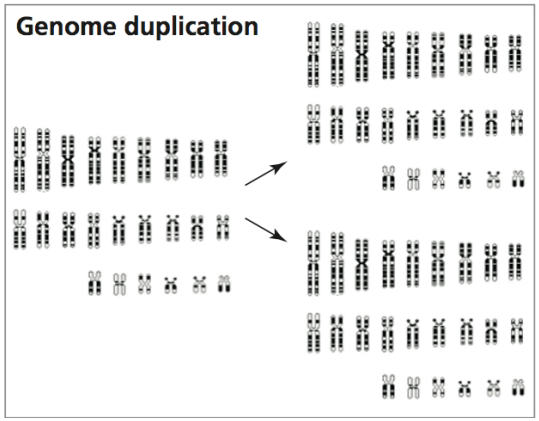
What do I mean by "big" and "small" below?

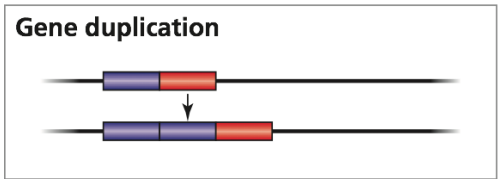
- Point mutations are small
- Insertions can be big or small
- Deletions can be big or small
- Duplications can be big or small
- Inversions can be big or small

Get it?









And on an even bigger scale….

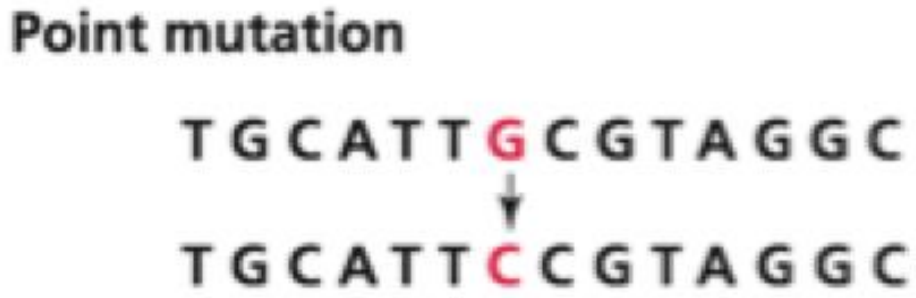
- Two chromosomes can fuse
- You may have an extra chromosome
- You may lose a chromosome
- Your whole genome may be duplicated

- 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 (wobble)???

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

Just for fun but its good practice to read hard stuff!

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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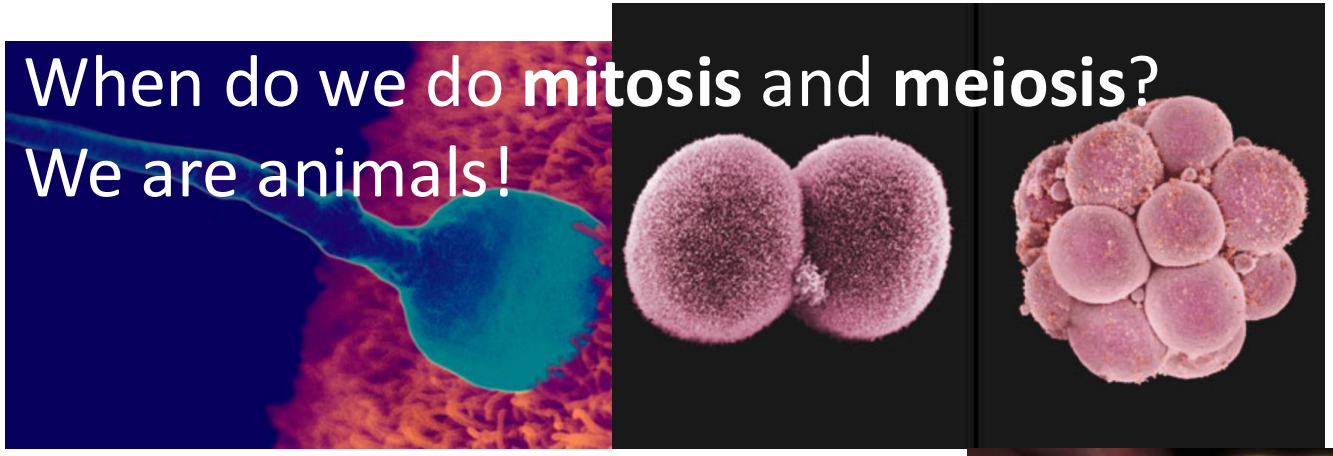
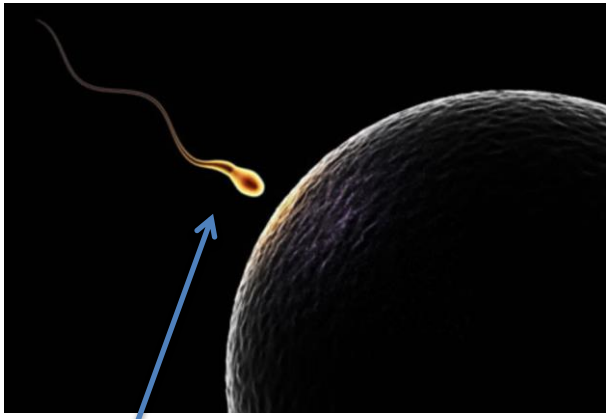
When do we do **mitosis** and **meiosis**?
We are animals!





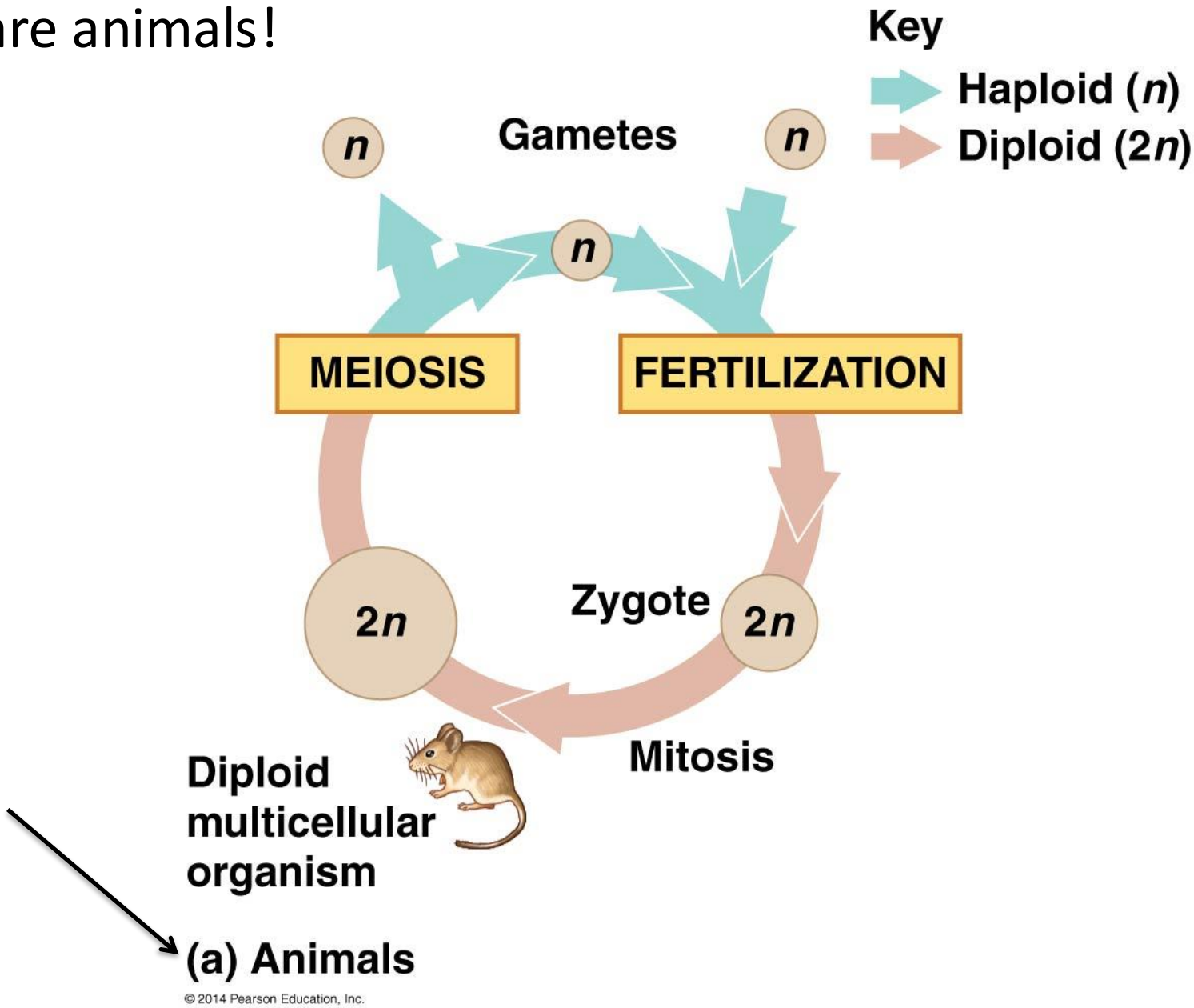
For your Review!

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

Egg

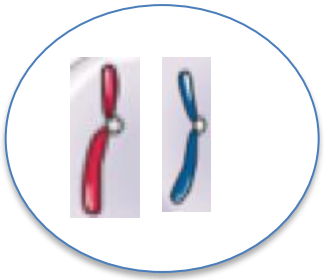
Sperm

When do we do **mitosis** and **meiosis**?
We are animals!



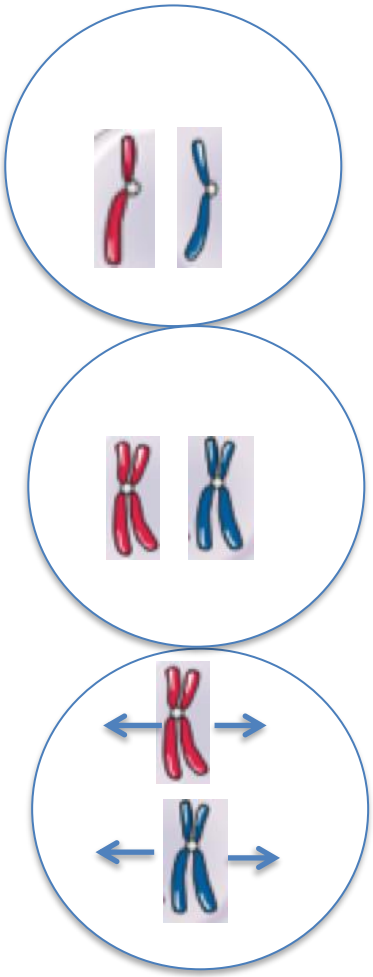
Mitosis

Meiosis

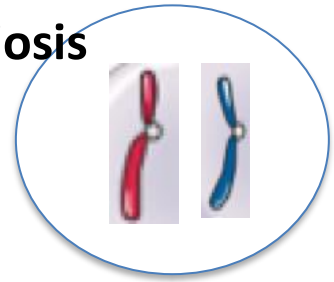


Once cell with one homologous pair of chromosomes!
We did Mitosis already…

Mitosis

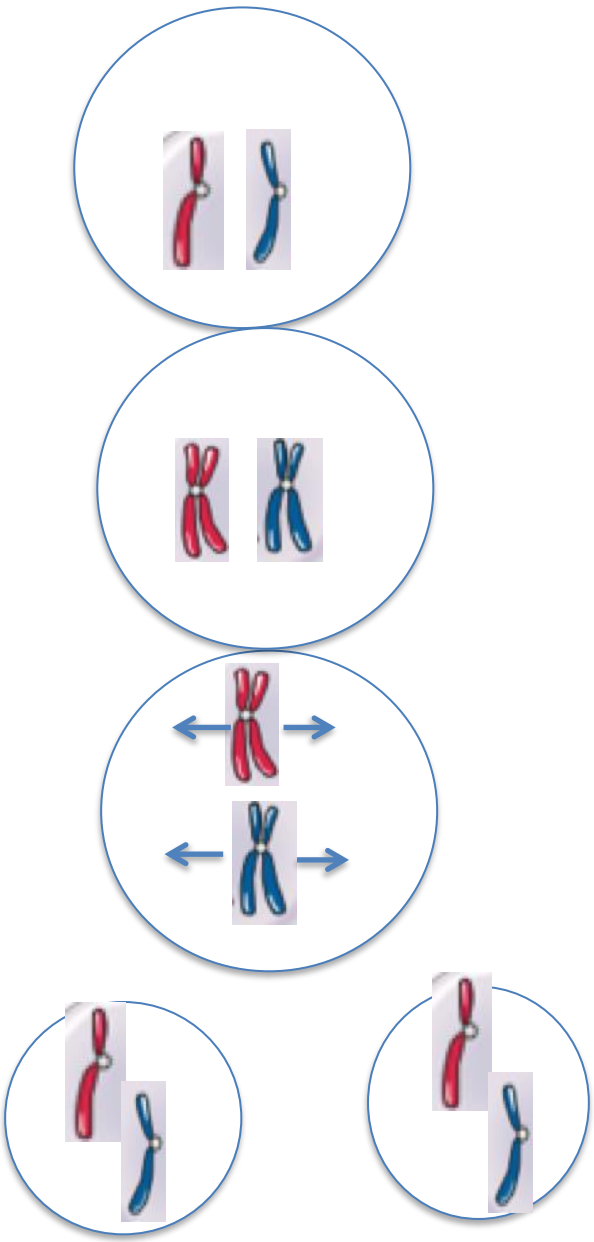


Meiosis

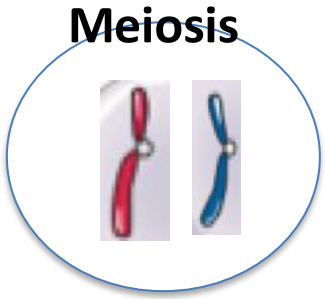


Are these sister chromatids separating from one another or homologous chromosomes?

Mitosis



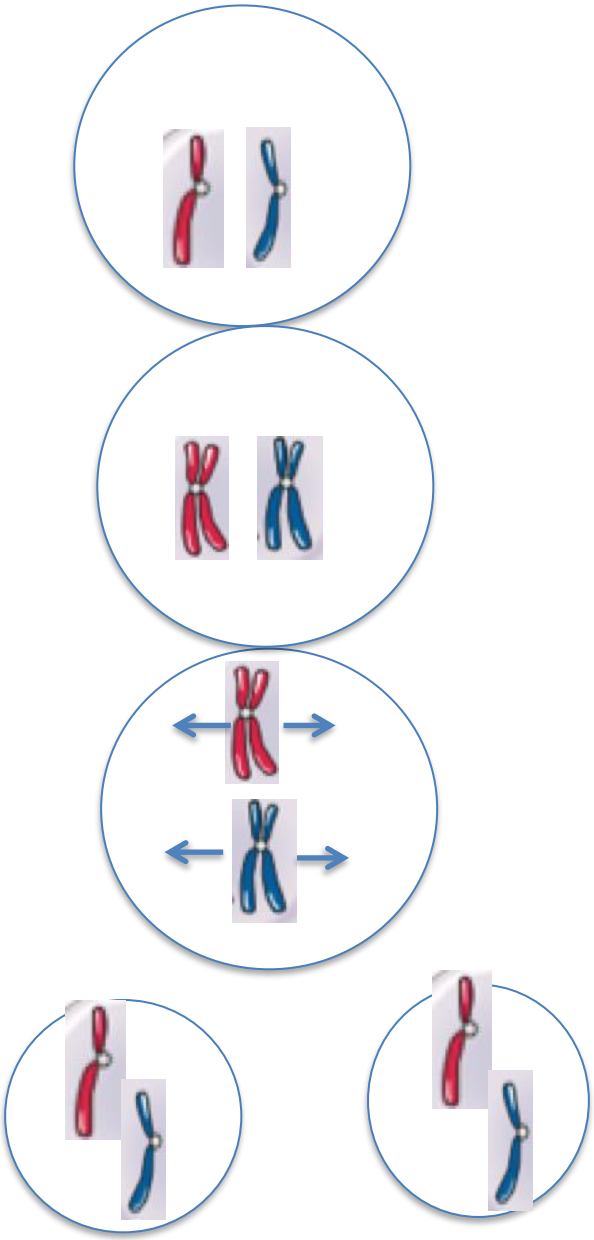
Meiosis



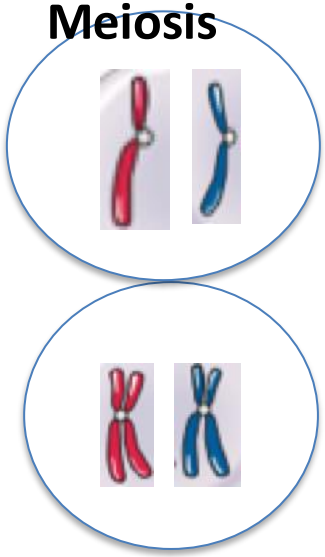
Mitosis
One cell to two cells
Diploid to diploid

now lets make some **gametes** through meiosis

Mitosis

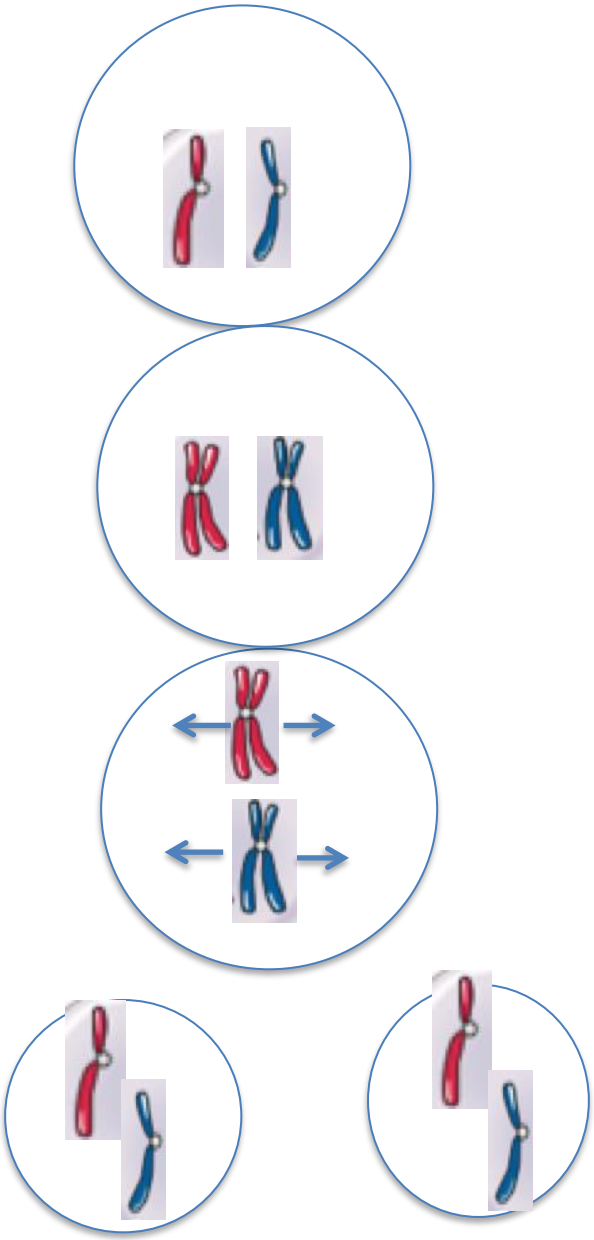


Meiosis

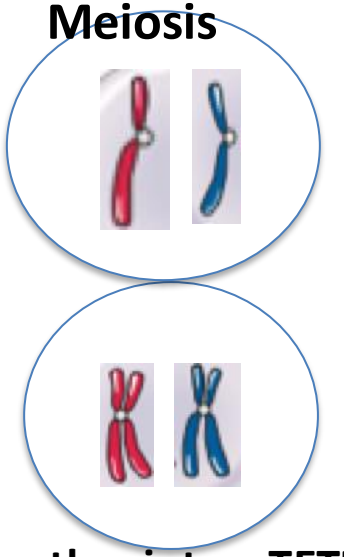


Now what?

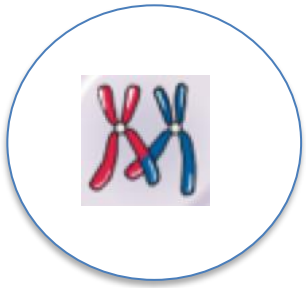
Mitosis



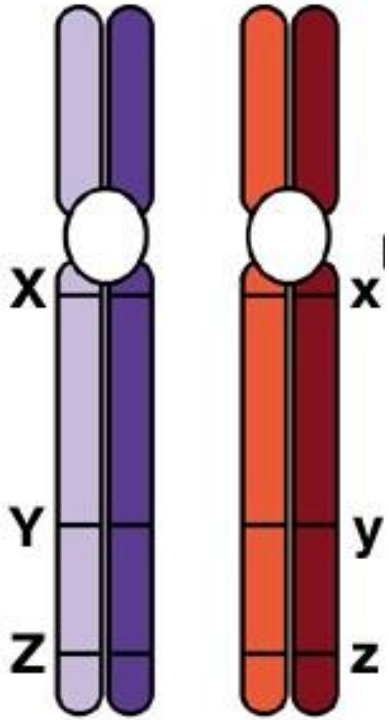
Meiosis



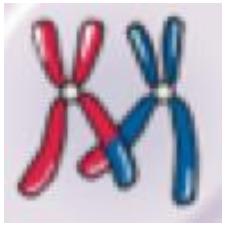
All 4 get together into a TETRAD to do what?????



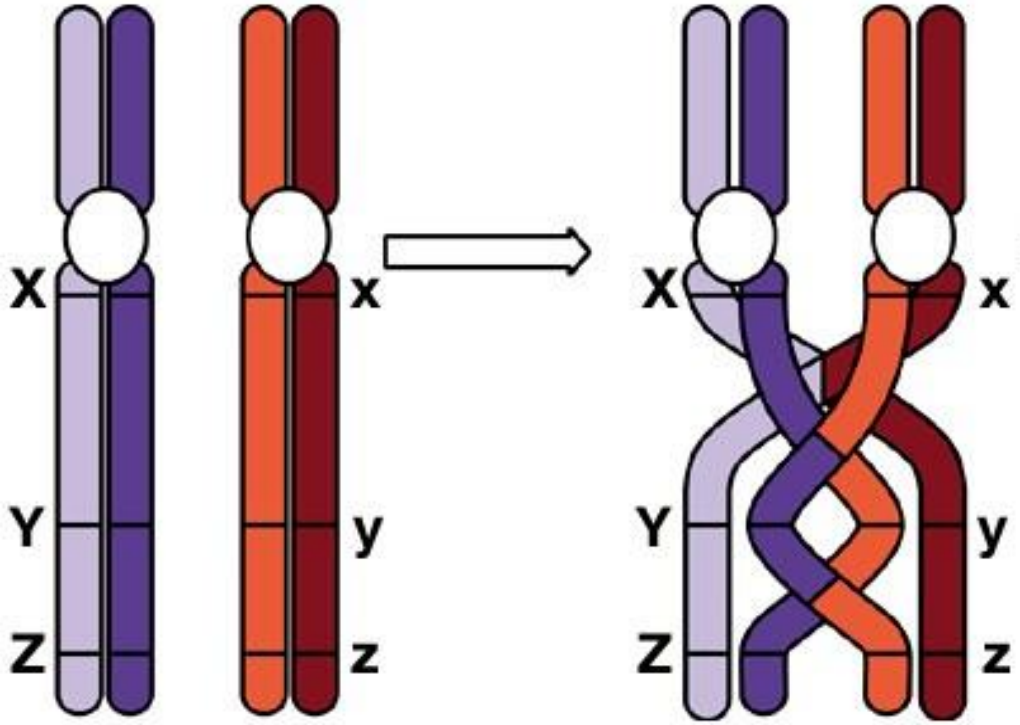
Crossing over during meiosis

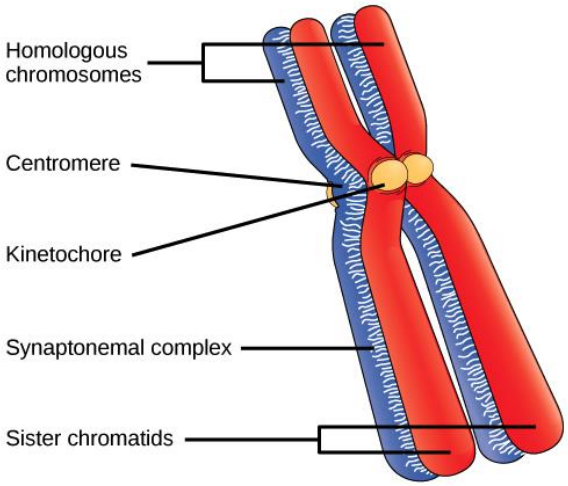


Crossing over=recombination

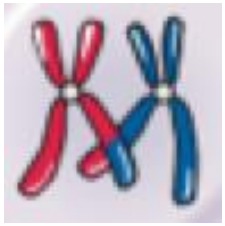


Crossing over during meiosis

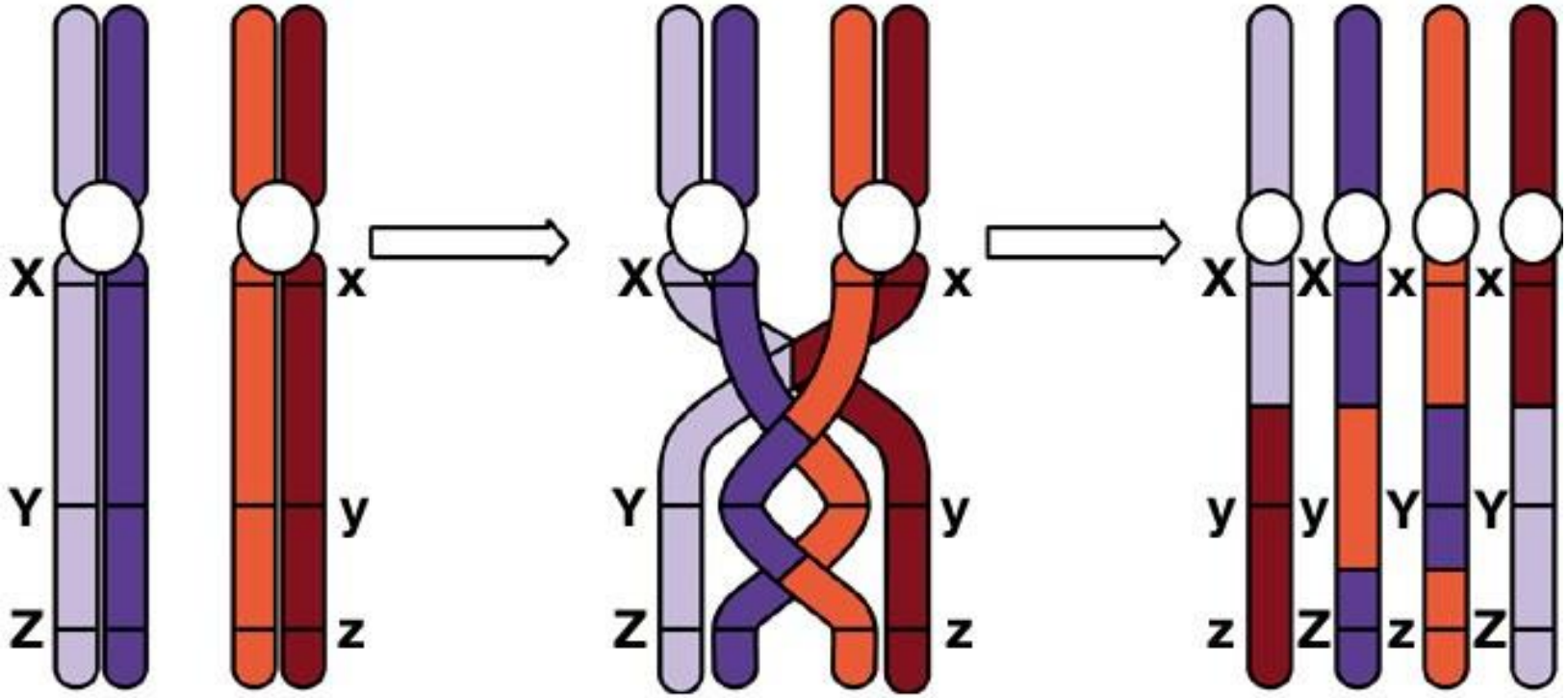




Crossing over=recombination

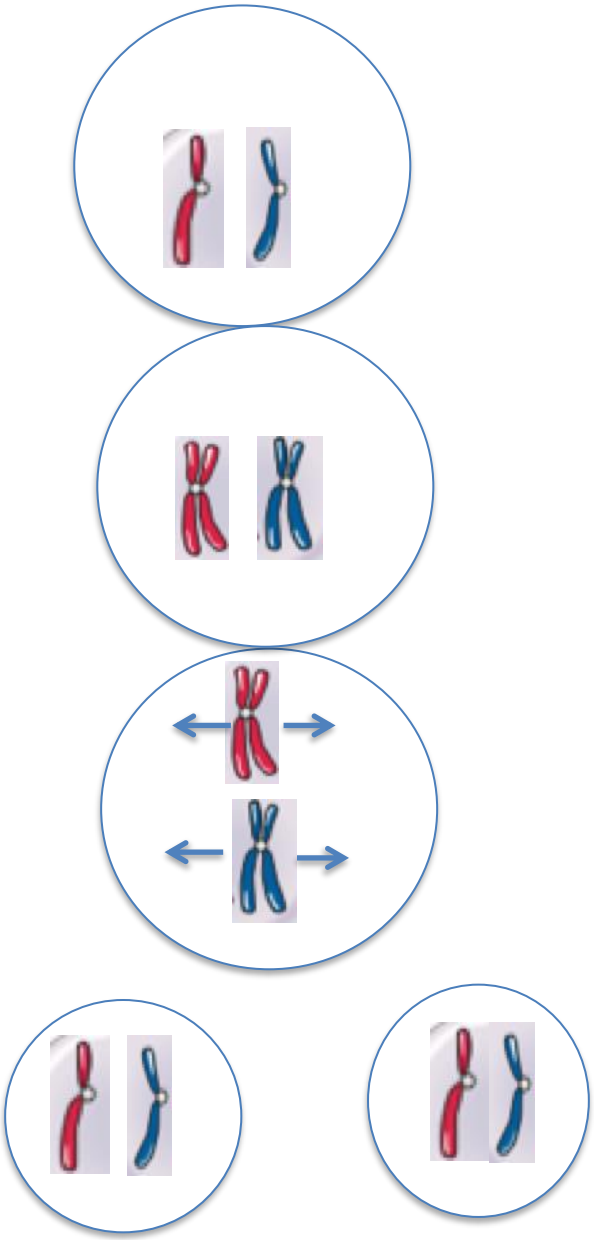


Crossing over during meiosis

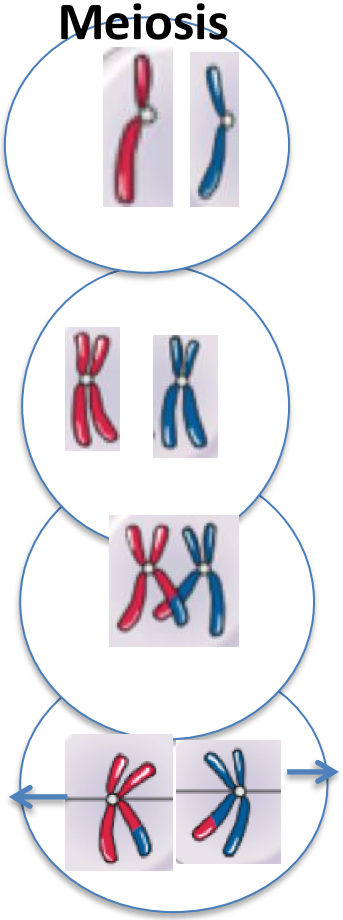


Crossing over=recombination

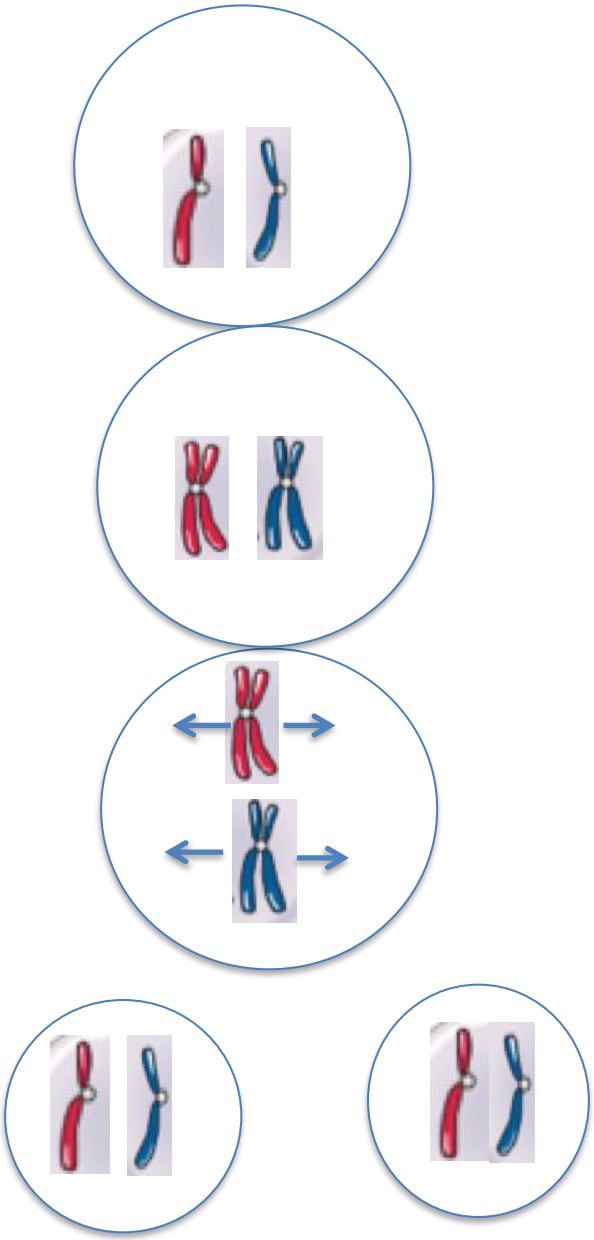
Mitosis



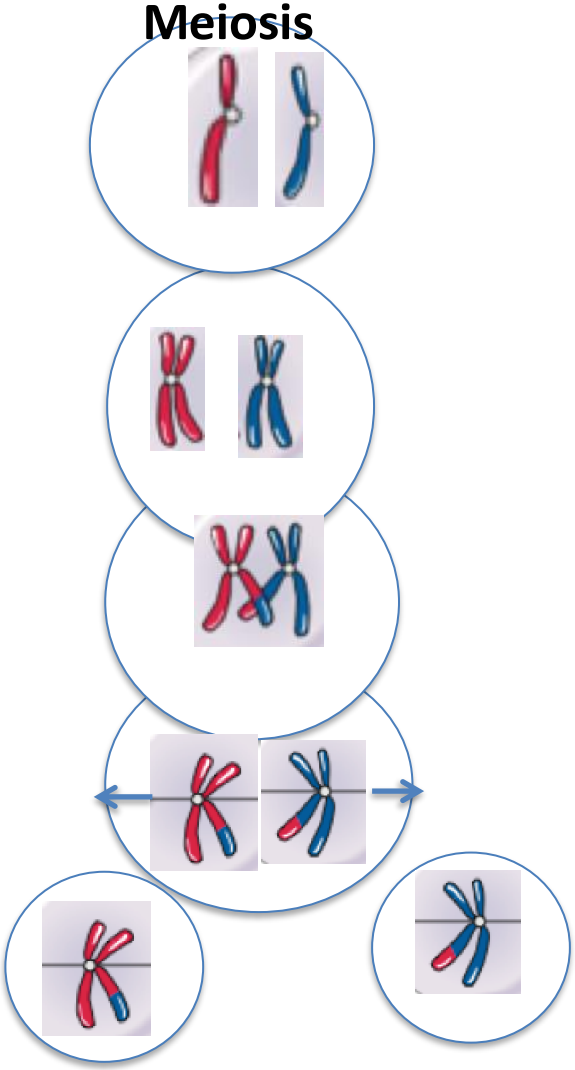
Meiosis



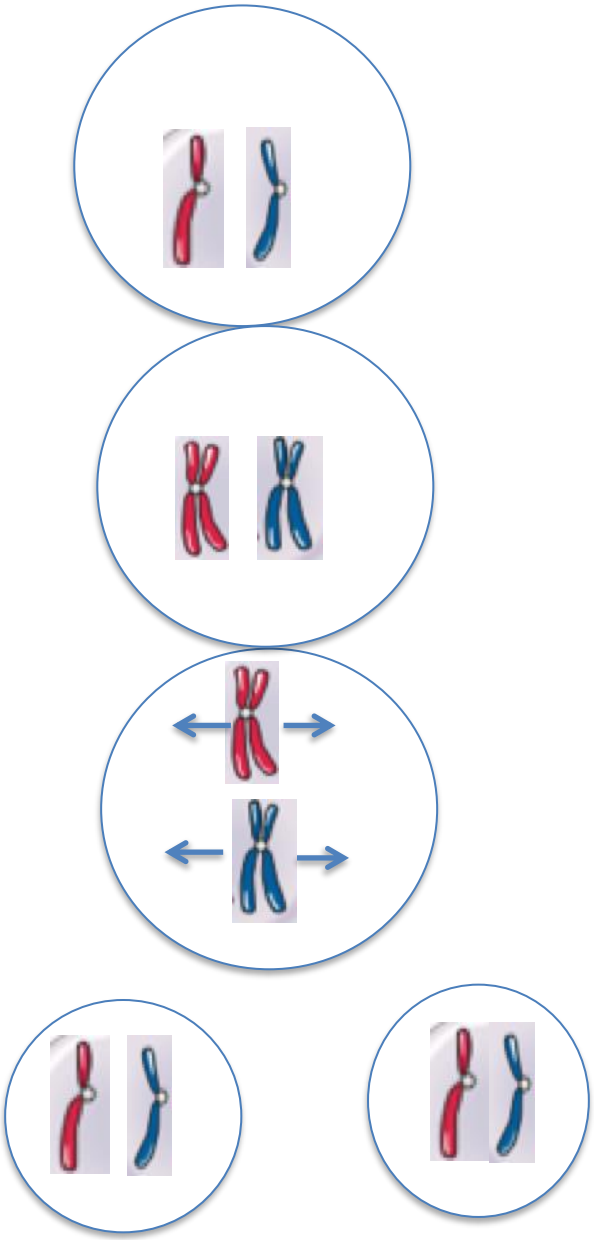
Mitosis



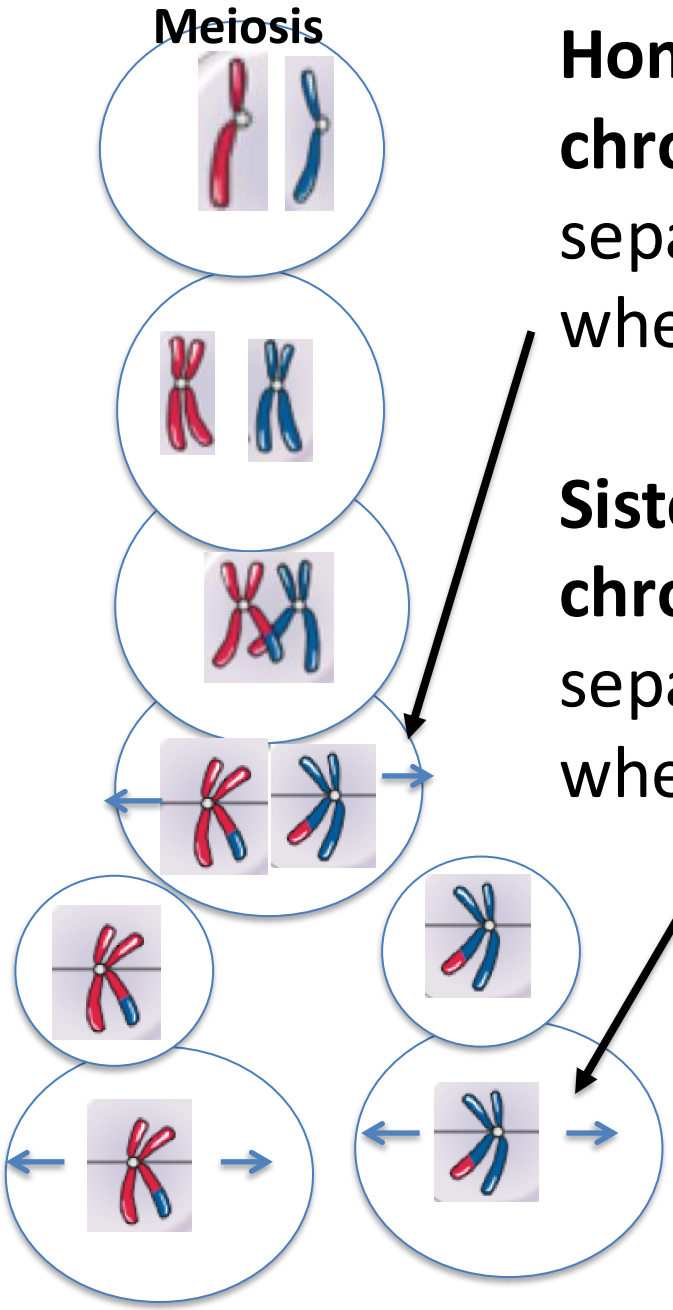
Meiosis



Mitosis

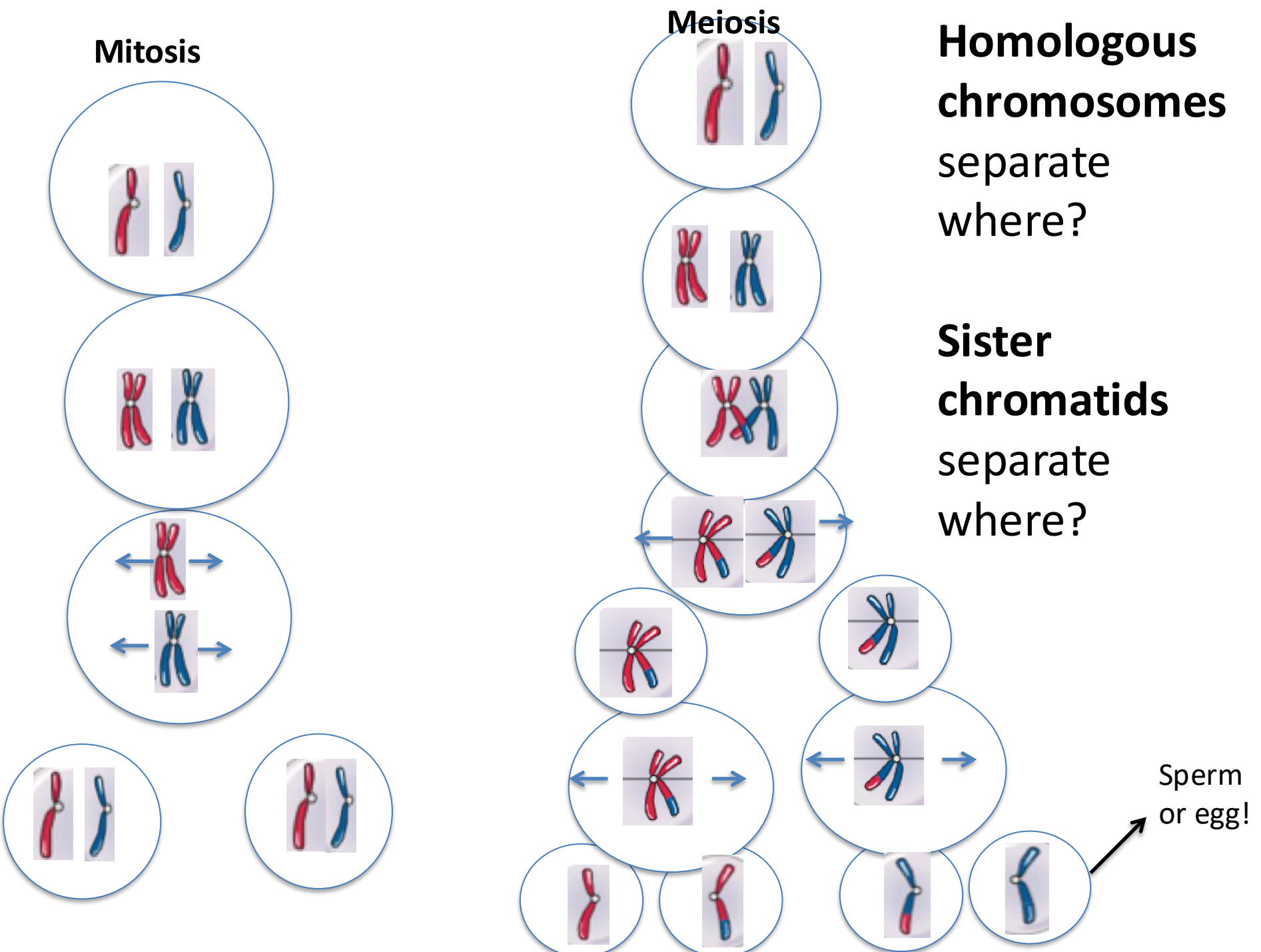


Meiosis



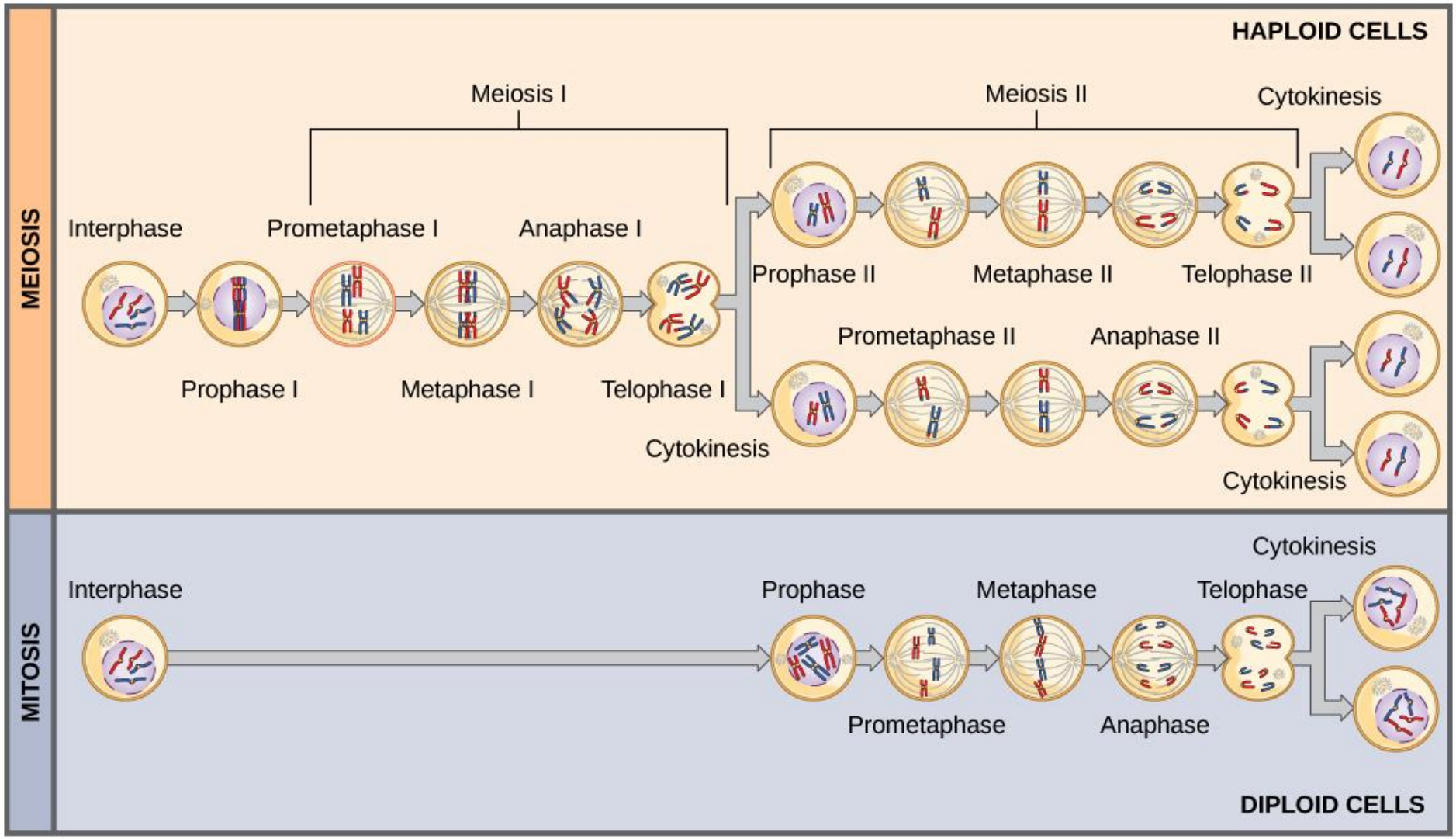
Homologous chromosomes separate where?

Sister chromatids separate where?


Mitosis
Meiosis
Homologous chromosomes separate where?
Sister chromatids separate where?
Sperm or egg!

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 many different kinds of mutations can occur when DNA is replicating (copying itself) during both mitosis and meiosis.

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!!!

In other words those genes are highly conserved.

How cool is that!

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