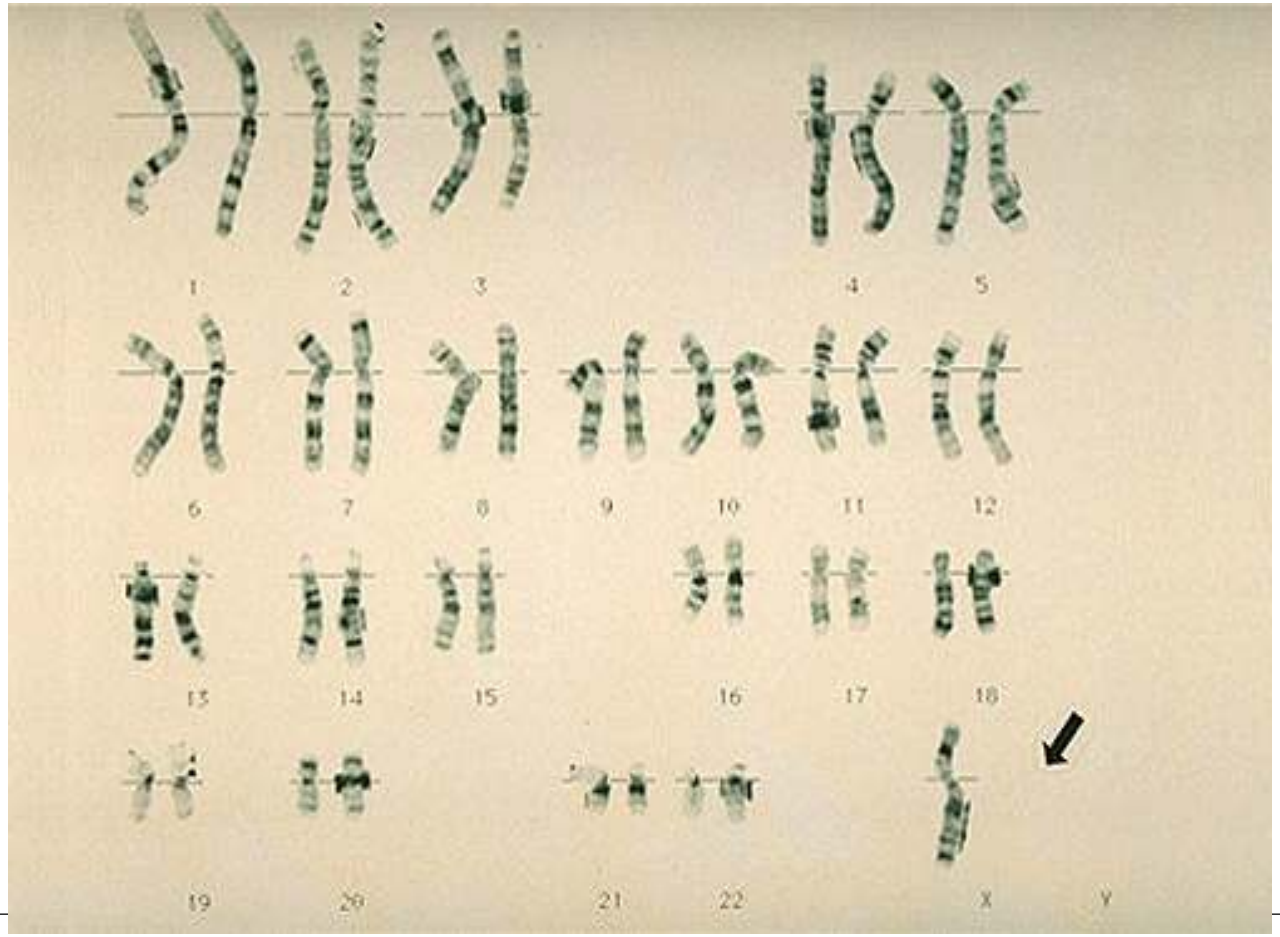
Chromsome Disorders and Chromosome Mutations

Today's assignment

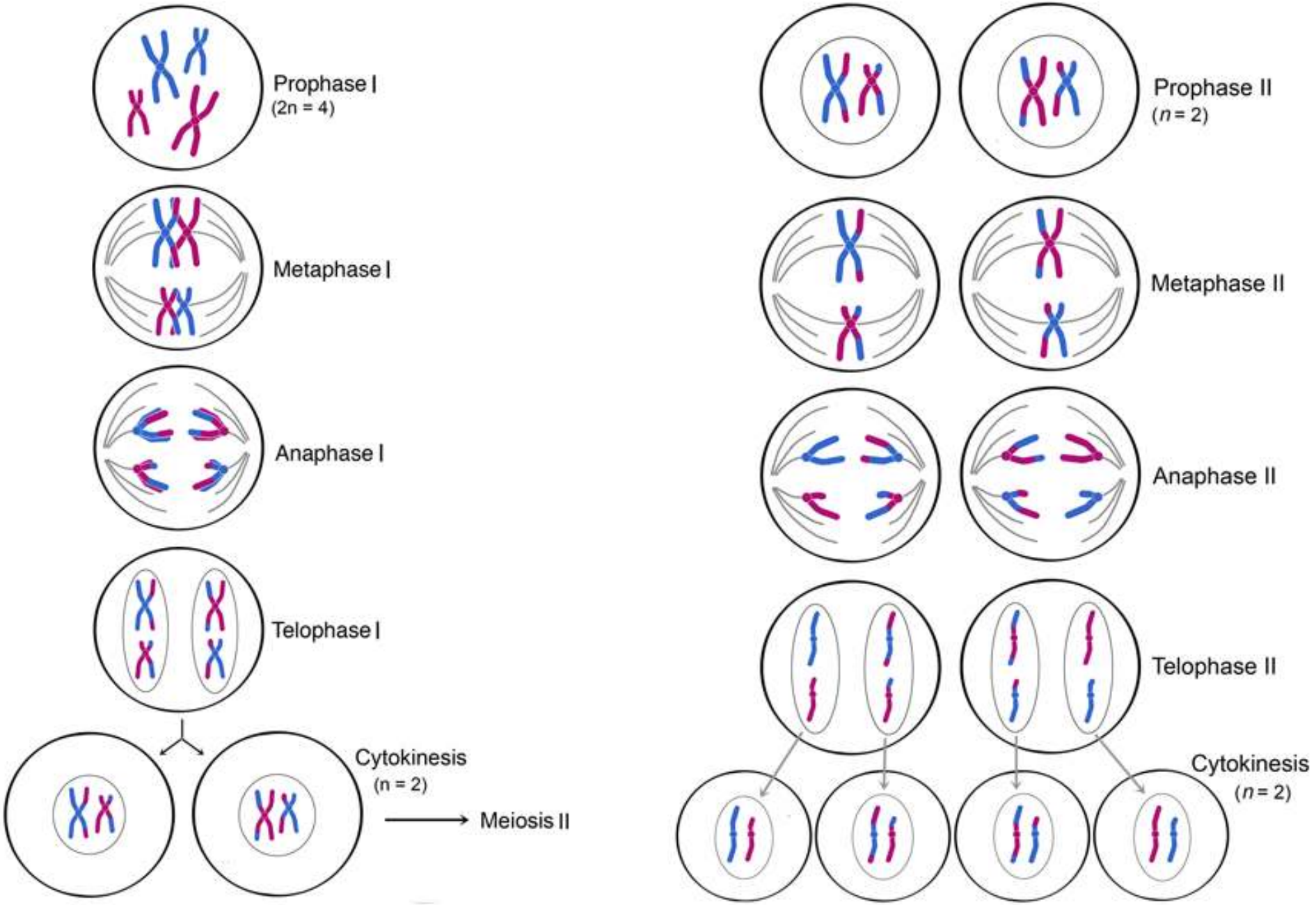
- As you go through this powerpoint, read the information. You will find a few situations where you are asked to write something down. Do this on a sheet of paper. When you finish, put your paper in the "turn in" bin.

Each **Chromosome** is important to the development of a normal organism.



- Can problems with meiosis occur?

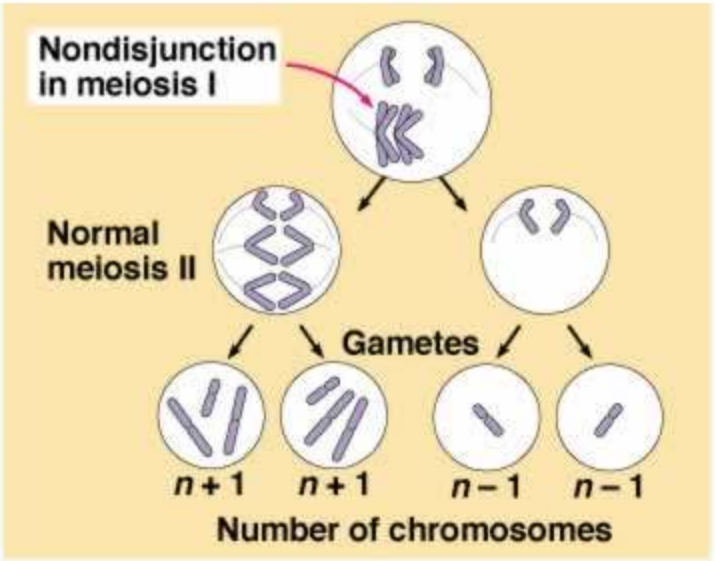
- Brainstorm: look at the picture of meiosis on the next slide and identify a place where an error can occur.
- 1. Write you ideas on your paper about where an error could occur.

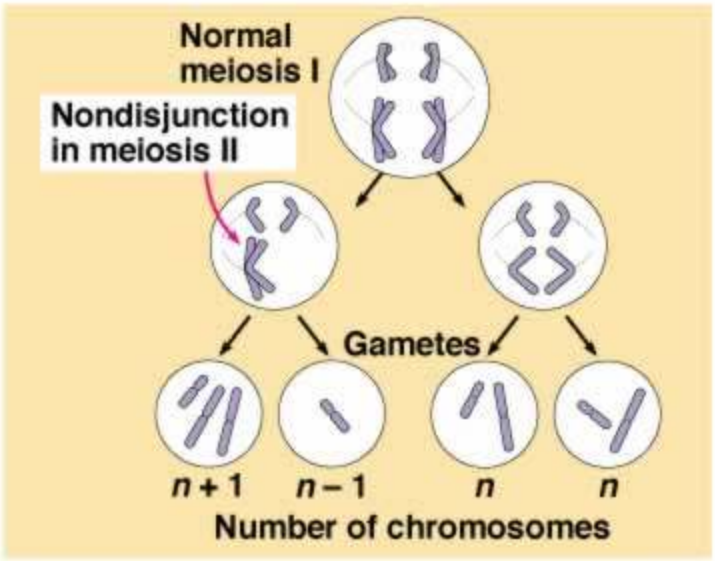

Prophase I
(2n = 4)
Metaphase I
Anaphase I
Telophase I
Cytokinesis
(n = 2)
Meiosis II
Prophase II
(n = 2)
Metaphase II
Anaphase II
Telophase II
Cytokinesis
(n = 2)

How do errors happen in meiosis?

Nondisjunction

- Sometimes homologous chromosomes fail to separate correctly. Sometimes sister chromatids fail to separate properly.
  - Result: Some cells end up missing chromosomes and others end up with too many chromosomes.





Does this cause problems?

- Yes
- Missing chromosomes causes errors with normal development in an organism
- Extra chromosomes causes errors in development also

Many genetic disorders are caused by chromosome problems

- Down Syndrome: Trisomy 21- an individual has 3 copies of chromosome 21
- Patau Syndrome: Trisomy 13- an individual has 3 copies of chromosome 13
- Edwards Syndrome: Trisomy 18- an individual has 3 copies of chromosome 18
- Klinefelter Syndrome: A male has an extra X chromosome (XXY)
- Turner Syndrome: A female is missing an X chromosome (X 0)
- Triple X (XXX): A female has 3 X chromosomes
- XYY: A male has an extra Y chromosome

Can other problems happen with chromosomes?

- Any ideas?

- Problems can happen with crossing over

- What could happen?
- 2. Write your ideas about how errors in crossing over could occur on your paper.

Changes in the DNA

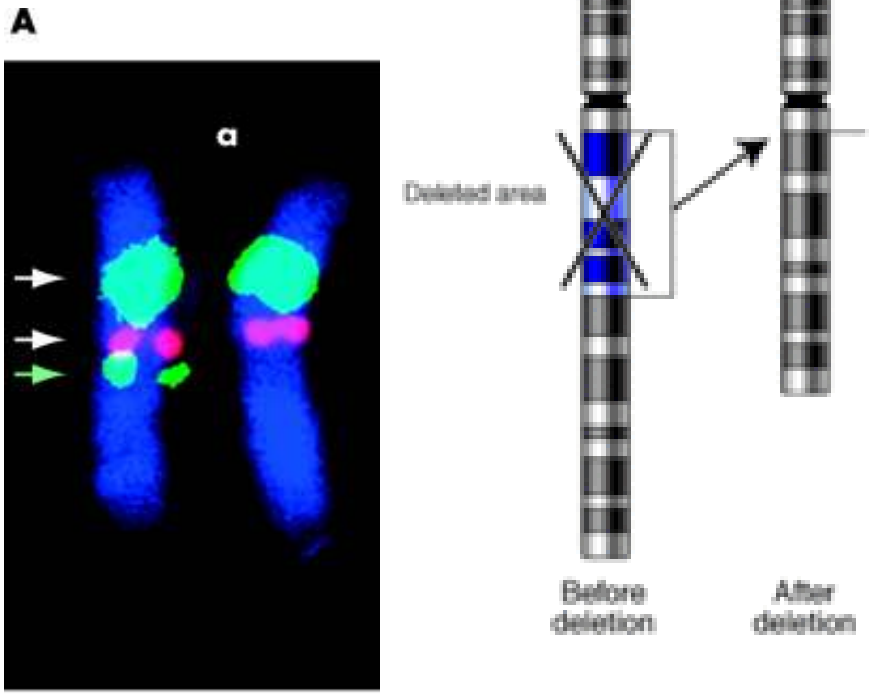
In general, mutations are changes in the DNA code.

Chromosomal <u>Mutations</u>: alterations in chromosomes

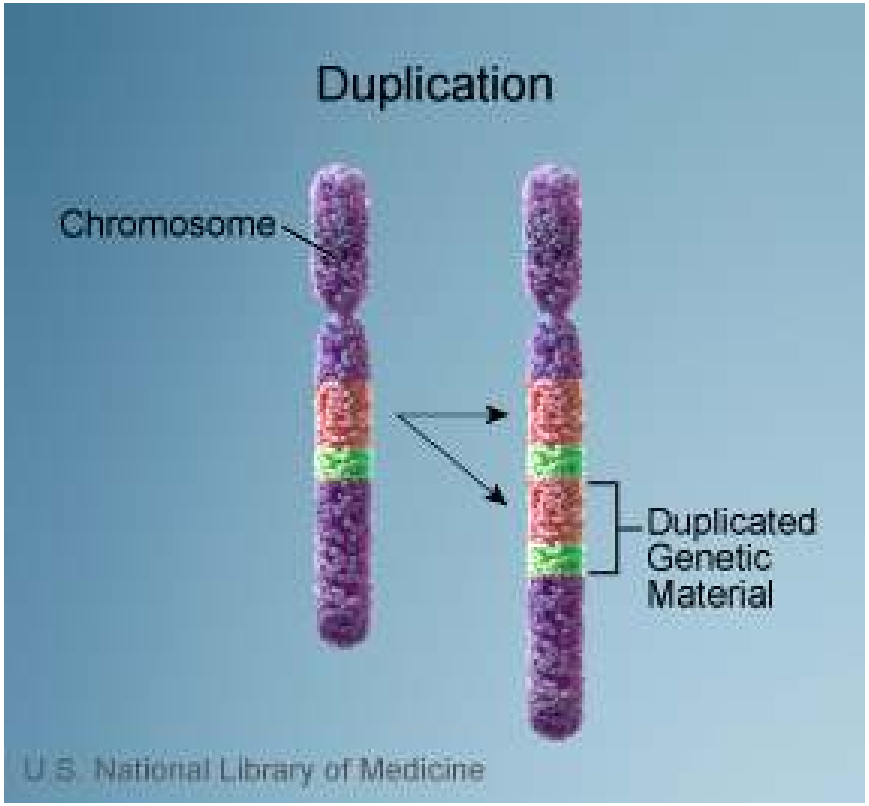
Sometimes during crossing over, segments of chromosomes can break off and get lost or get placed in the wrong location.

Why can it be a bad thing if the DNA gets changed?

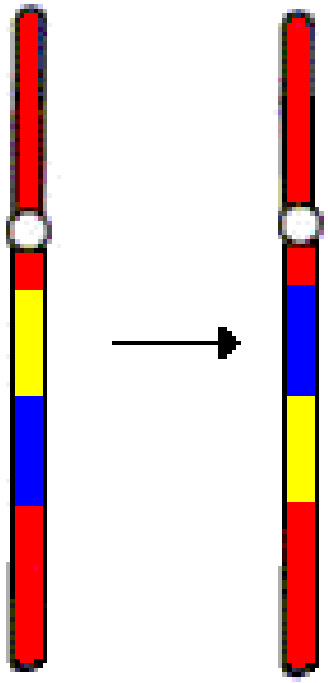
- **<u>Deletion</u>** is a process in which a chromosome is damaged as fragments break off the chromosome and are lost.



- **<u>Duplication</u>** occurs as fragments attach to the homologous chromosomes.

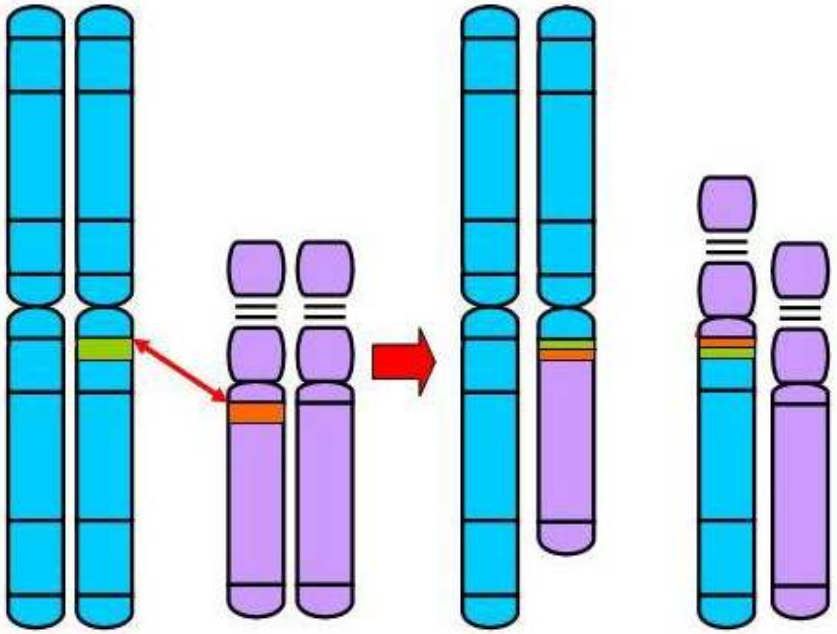


- **Inversion** happens when fragments of the original chromosome attach in reverse order.



Paracentric Inversion

- **Translocation** occurs when a fragment may join a non-homologous chromosome.



Other less common abnormalities

- **Cri-du-chat (cat cry) syndrome** (deletion on chromosome 5): Affected children have a cat-like, high-pitched cry during infancy, mental and physical disabilities . About 1 in 20,000 to 50,000
- **Prader-Willi syndrome** (deletion on chromosome 15): Learning disabilities and short stature. About 1 in 10,000 to 25,000
- **22q11 deletion syndrome (**DiGeorge or velocardiofacial syndrome) (deletion on chromosome 22): About 1 in 4,000. Can cause heart defects, cleft lip/palate, immune system abnormalities, characteristic facial features and learning disabilities. 50-percent chance of passing this chromosomal abnormality on to their offspring.
- **Wolf-Hirschhorn syndrome** (deletion on chromosome 4): Mental disabilities, heart defects, poor muscle tone and seizures. It affects about 1 in 50,000

Research Time

- Go to this website learn.genetics.utah.edu
- In the "Human Health" Category click on the "Genetic Disorders" link, then click the link to Chromosome Abnormalities.
  There you will find a list of disorders. Choose TWO disorders and on your paper do this for each disorder.
  - Describe how this disorder is cause by a chromosome error.
  - Describe the symptoms of the disorder.
  - Describe any treatments available for the disorder.

That’s it for today!

Today's activity

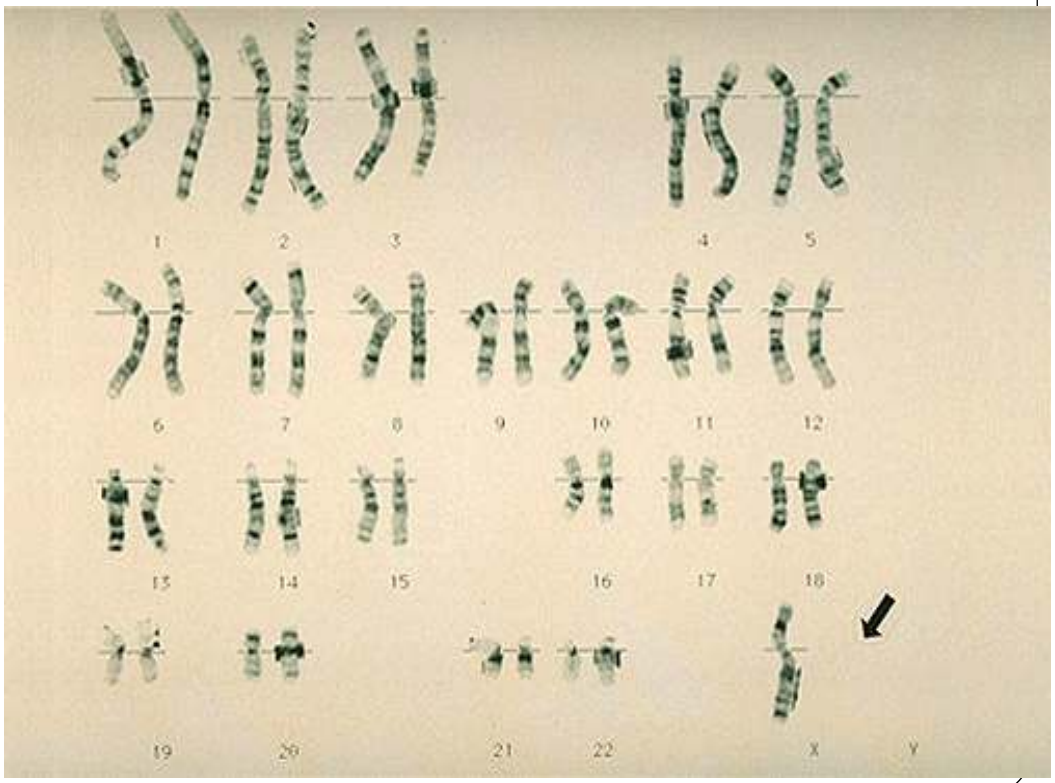
- Today you will be playing the role of a doctor. Your patient has come to you reporting problems that relate to chromosome disorders.

- You goal will be to correctly identify the chromosome disorder found in your patient.

Here's what you need

- Grab a Karyotype Lab sheet
  - You can work with 1 partner of your choice if you would like to. Each person needs their own sheet.
- Read the patient scenarios and choose a patient to work with- grab the lettered chromosome sheet.
- Each chromosome sheet has 1 set of chromosomes numbered 1-22. Your job is to find the matching pair to each chromosome on your sheet. Number them when you find the match. Look closely at the banding patterns to make sure you get the right matches! Don't forget, each person has sex chromosomes. Find and label those as well.
- Once you have identified all the chromosomes, you will be able to assess what chromosome disorder your patient is dealing with. Use the previous slide that mentions the genetic disorders to make the correct diagnosis. Then use that information to answer the questions on your lab sheet.
- Good luck and have fun being a doctor!

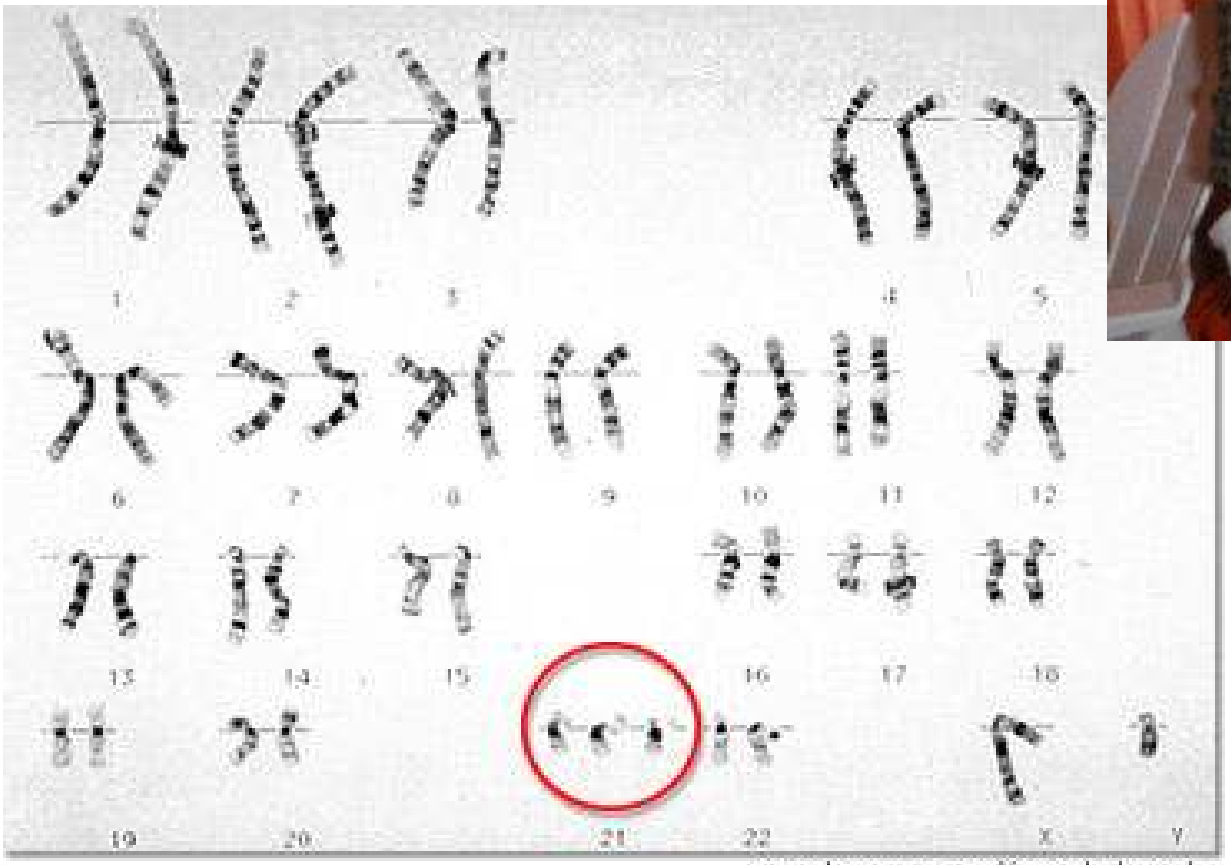
- **<u>Monosomy</u>** is a condition in which a diploid cell is missing a chromosome. Most embryos will not survive if missing a chromosome.
  - EX: Turner Syndrome



Down Syndrome:

Trisomy 21 an individual has 3 copies of the 21st chromosome

(A 3rd chromosome on the 21st pair)



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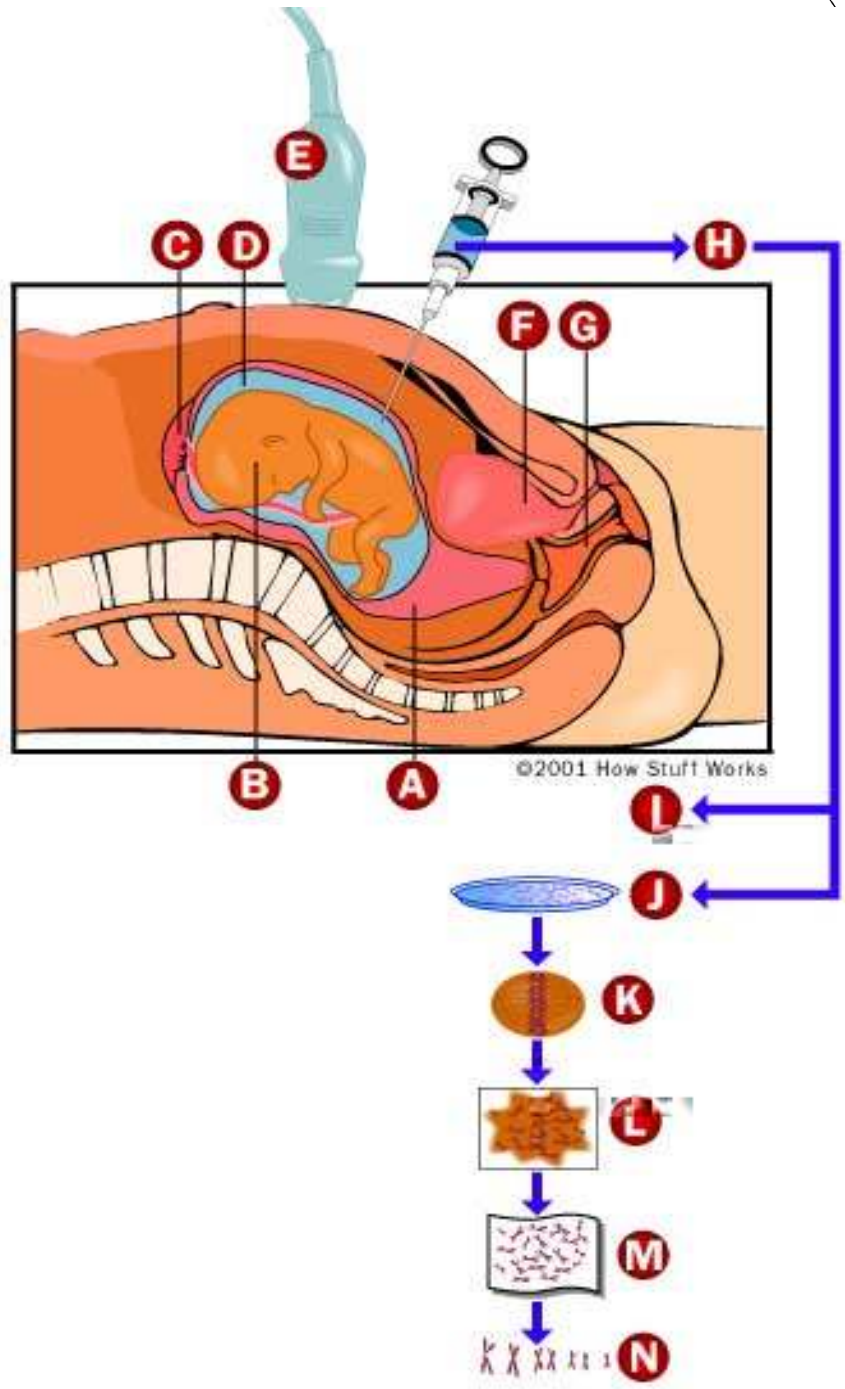
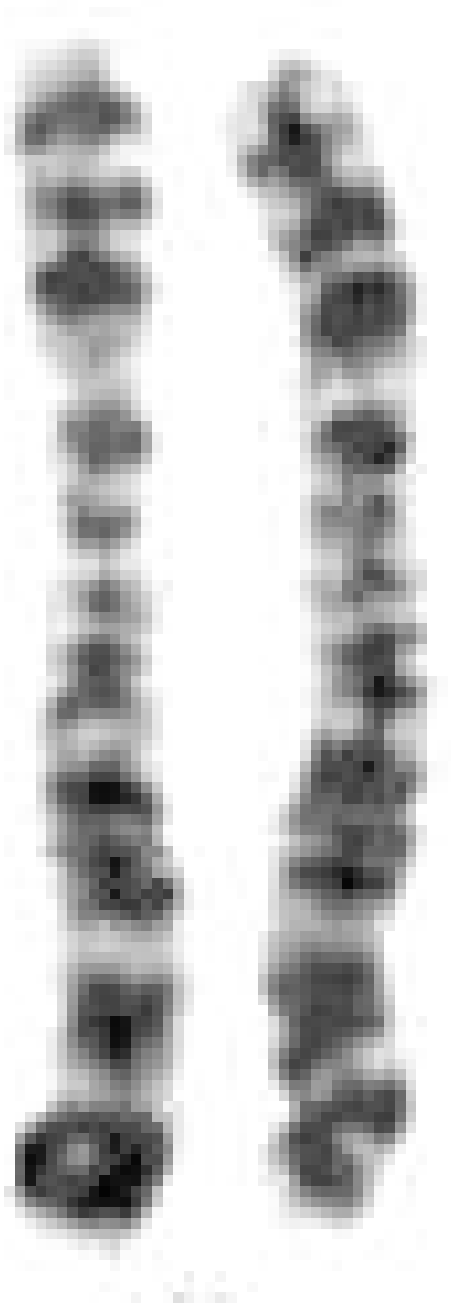
Common chromosome abnormalities

- Trisomy 13 and 18 – more severe physical and mental disabilities. Most live less than 1 year.
- Klinefelter syndrome (XXY): boys with lower than normal testosterone levels

In Amniocentesis, amniotic fluid is tested for chromosome problems.

The normal frequency of down syndrome is 1 in 1500. For couples who are older than 30 it is 1 in 1000. In couples 35-40 it is 1 in 750. In couples older than 45 the risk is 1 in 16.





Use clues like, Banding Pattern, Length and Centromere location to determine which chromosomes have been selected