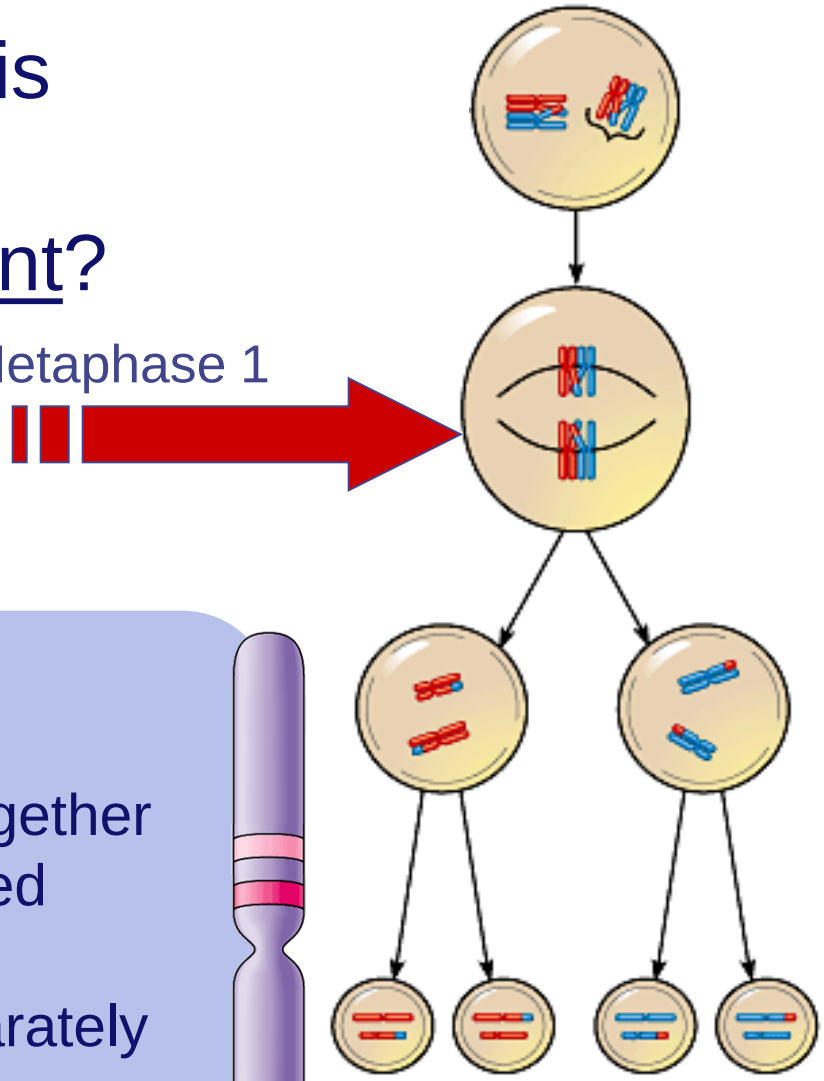
Mendel's 2nd law of heredity

- Law of **independent assortment**
  - different **loci** (genes) separate into gametes independently
    - non-homologous chromosomes align independently
    - classes of gametes produced in equal amounts
      - YR = Yr = yR = yr
    - only true for genes on separate chromosomes or on same chromosome but so far apart that crossing over happens frequently

Law of Independent Assortment

- Which stage of meiosis creates the law of independent assortment?

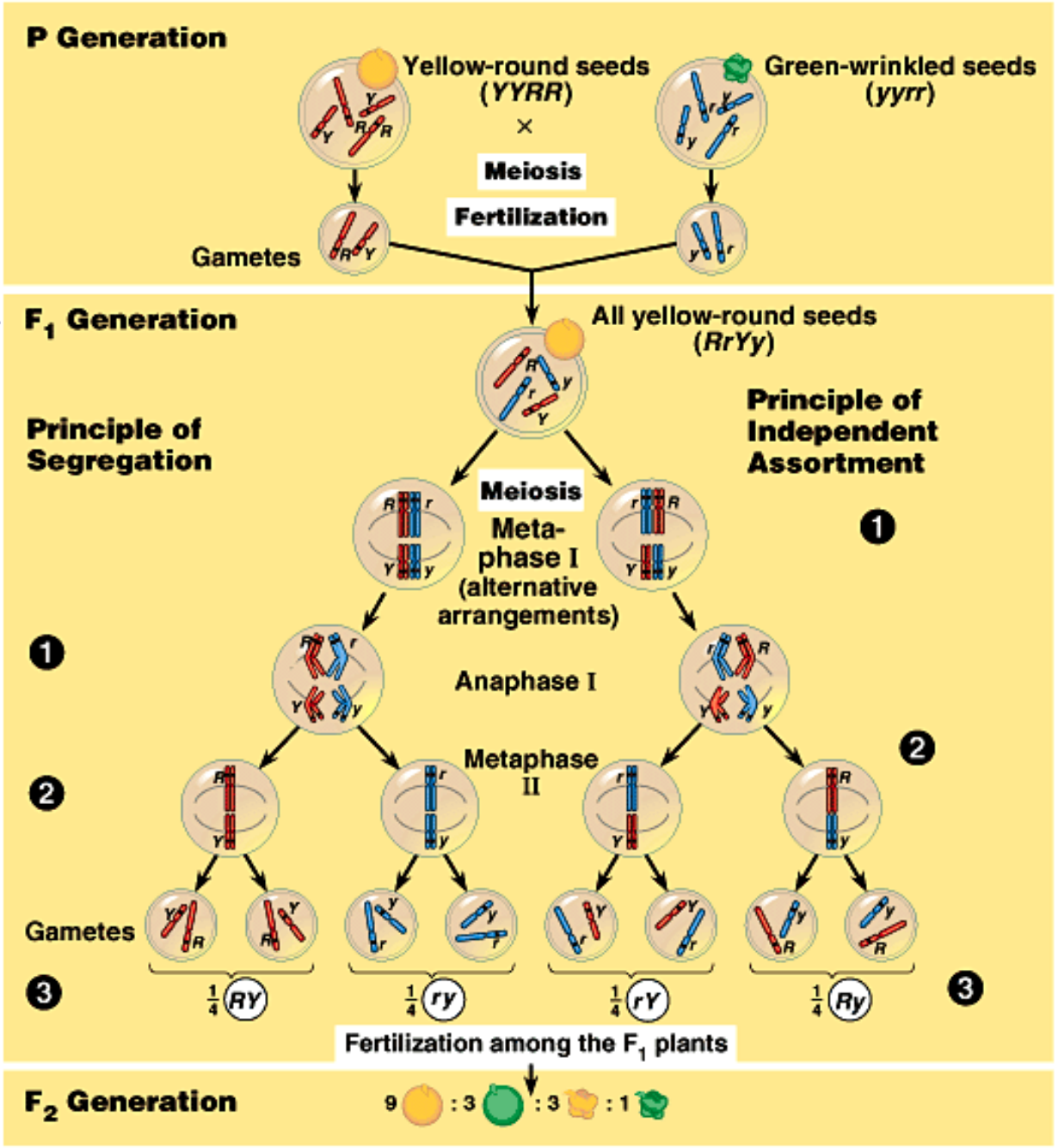


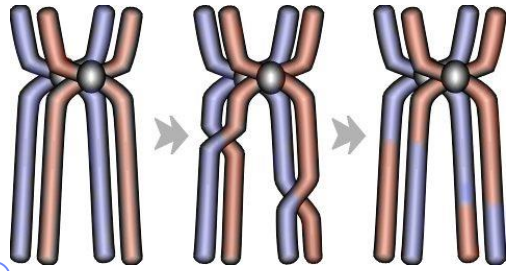
EXCEPTION

- If genes are on same chromosome & close together
  - will usually be inherited together
  - rarely crossover separately
  - "linked"

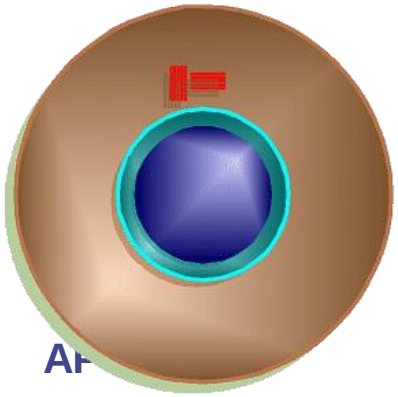
The chromosomal basis of Mendel's laws…

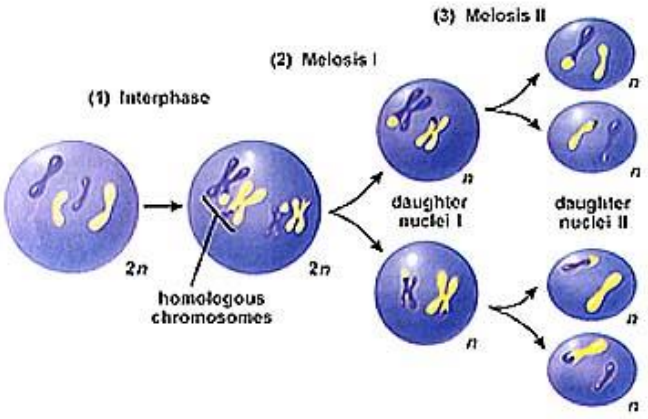
Trace the genetic events through meiosis, gamete formation & fertilization to offspring





Errors of Meiosis
Chromosomal Abnormalities
Concept 15.4





Modified from Kim Foglia

Friday, April 10th

Have you ever heard of the disease called **Tay-Sachs**? Please watch this video clip.

- Is the disease autosomal or sex-linked?
- Is the disease dominant or recessive?
- How does this disease differ from Down syndrome?

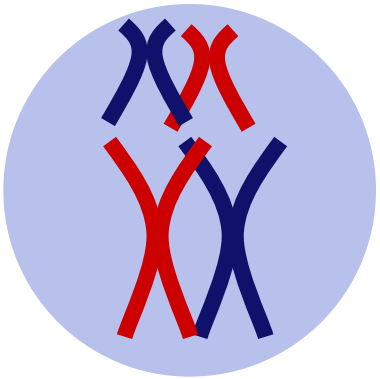
Today I will…

1. ***Describe*** disjunction.
2. ***Explain*** trisomy and ***provide*** an example of a condition linked to this error.
3. ***List*** various disorders associated with chromosomal abnormalities.

Review: Mendel's laws of heredity

- **Law of segregation**
  - **monohybrid cross**
    - single trait
  - each **allele** segregates into separate gametes
    - established by Metaphase 1
- **Law of independent assortment**
  - **dihybrid (or more) cross**
    - 2 or more traits
  - **genes** on separate chromosomes assort into gametes independently
    - established by Metaphase 1

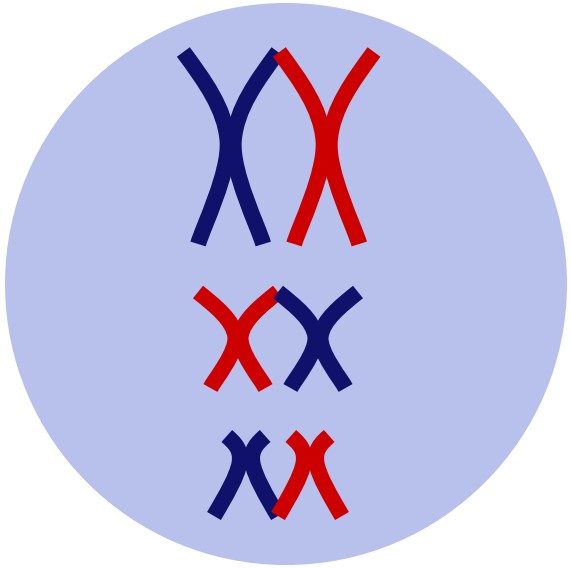
EXCEPTION
- linked genes



metaphase1

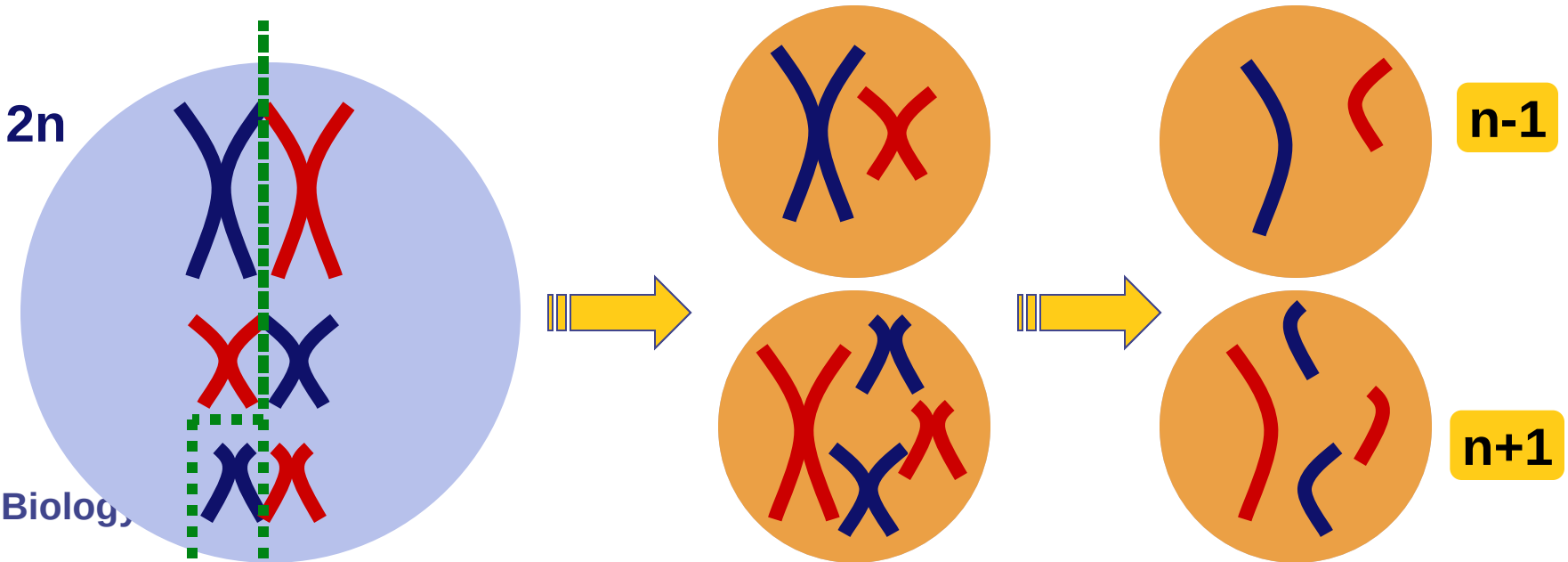
Chromosomal abnormalities

- Incorrect number of chromosomes
  - nondisjunction
    - chromosomes don't separate properly during meiosis
  - breakage of chromosomes
    - deletion
    - duplication
    - inversion
    - translocation

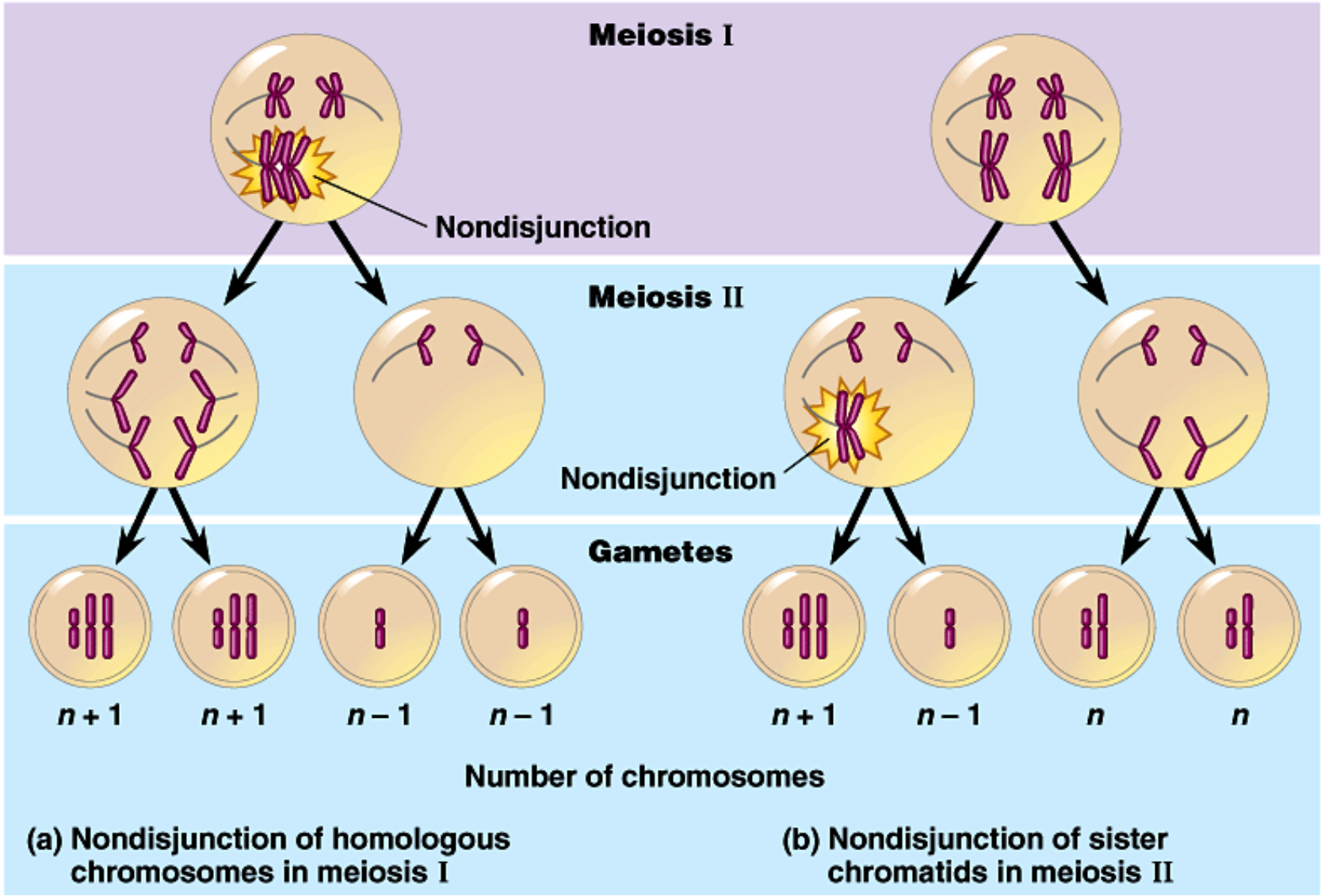


Nondisjunction

- Problems with meiotic spindle cause errors in daughter cells
  - homologous chromosomes do not separate properly during Meiosis 1
  - sister chromatids fail to separate during Meiosis 2
  - too many or too few chromosomes

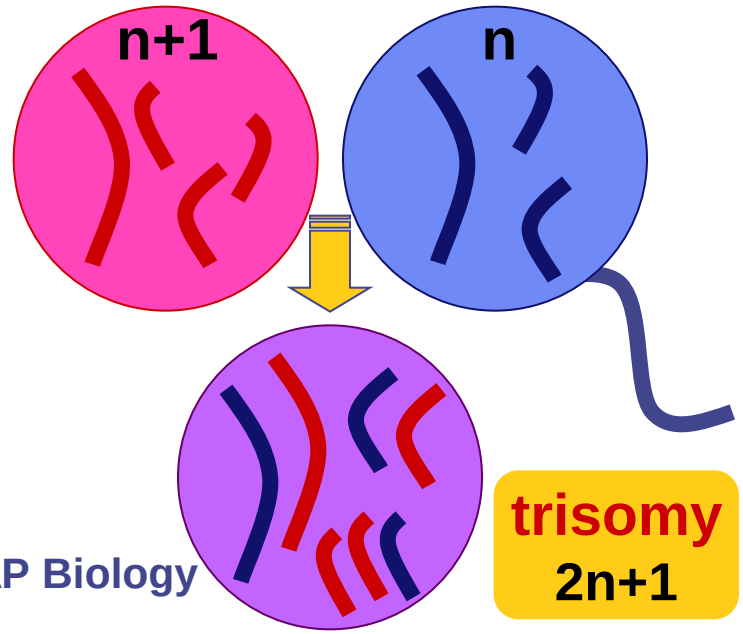


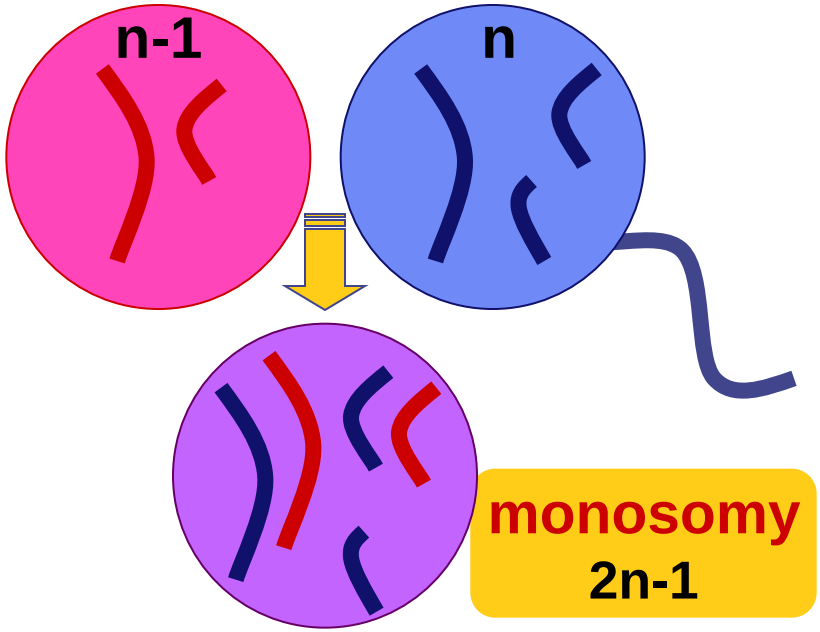
Alteration of chromosome number



Nondisjunction

- Baby has wrong chromosome number
  - trisomy
    - cells have 3 copies of a chromosome
  - monosomy
    - cells have only 1 copy of a chromosome



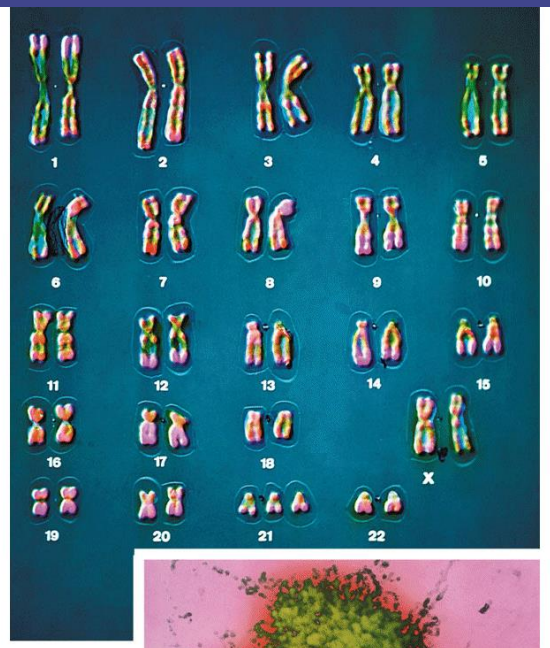


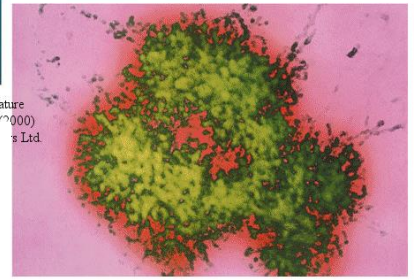
Human chromosome disorders

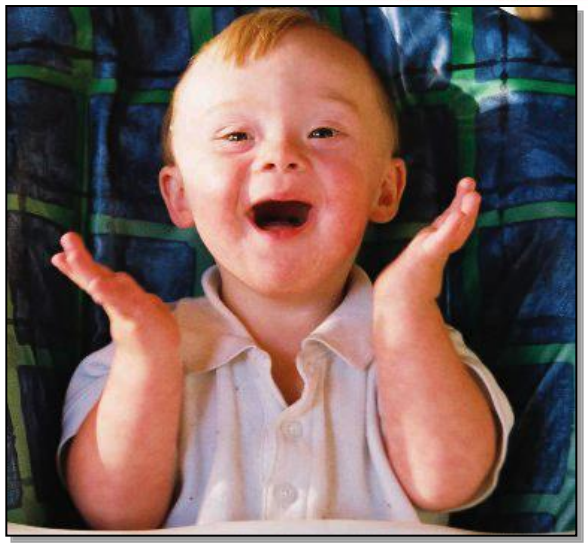
- **High frequency in humans**
  - most embryos are spontaneously aborted
  - alterations are too disastrous
  - developmental problems result from biochemical imbalance
    - imbalance in regulatory molecules?
- **Certain conditions are tolerated**
  - upset the balance less = **survivable**
  - characteristic set of symptoms = **syndrome**

Down syndrome

- Trisomy 21
  - 3 copies of chromosome 21
  - 1 in 700 children born in U.S.
- Chromosome 21 is the smallest human chromosome
  - but still severe effects
- Frequency of Down syndrome correlates with the age of the mother

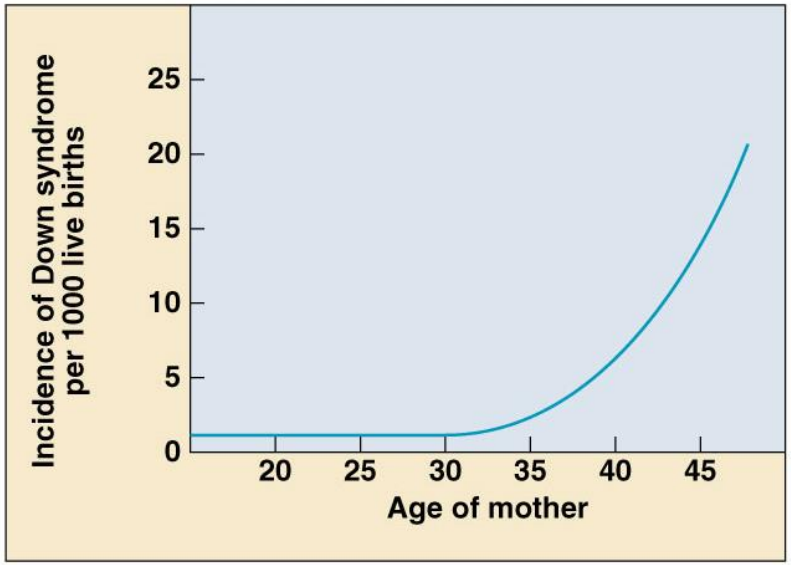






Down syndrome & age of mother

Mother's age	Incidence of Down Syndrome
Under 30	<1 in 1000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12

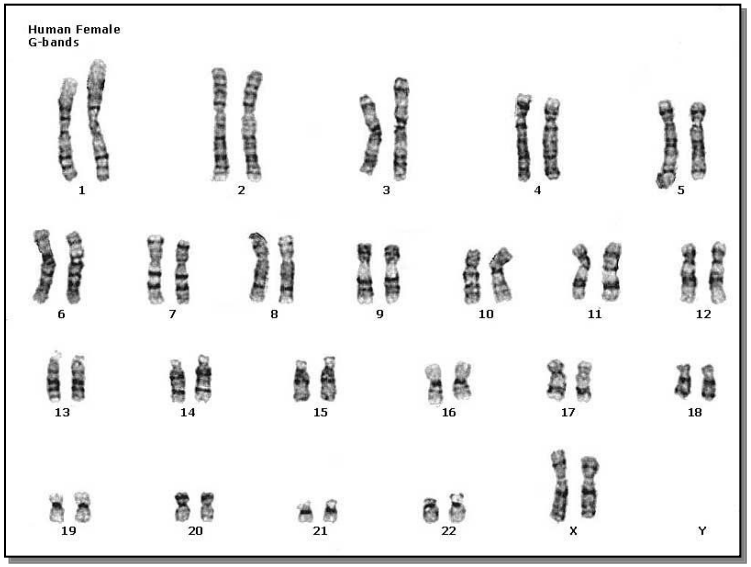


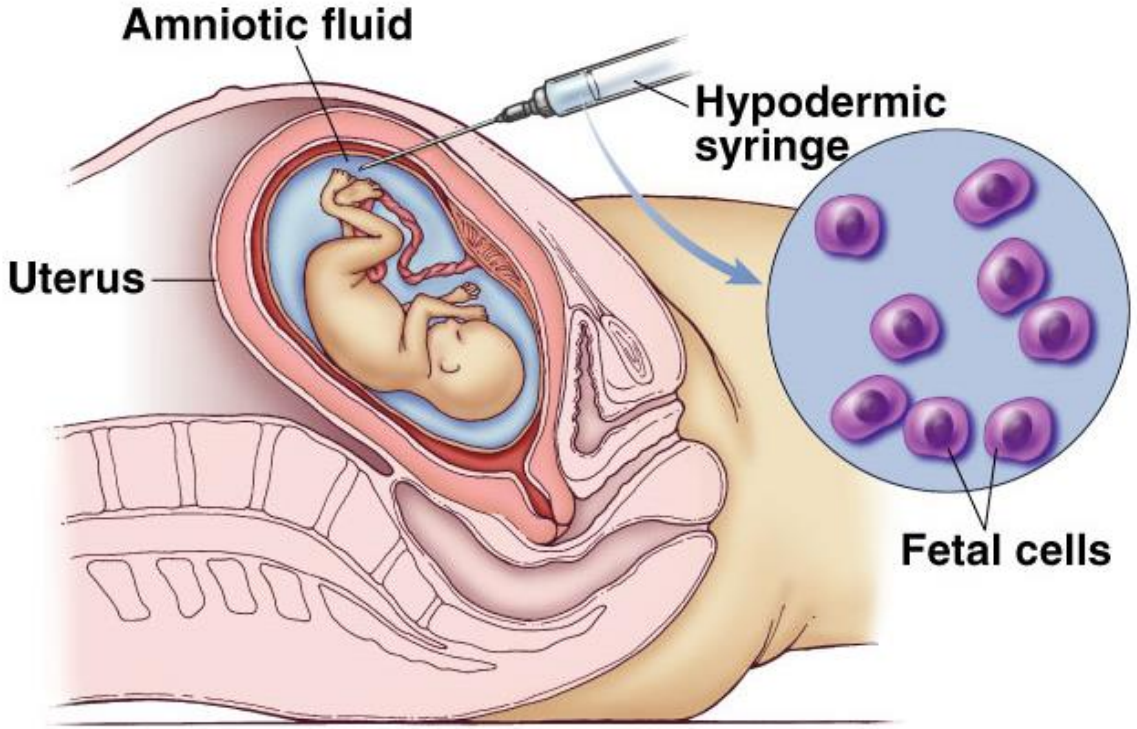
Rate of miscarriage due to amniocentesis:

- **1970s data**
  0.5%, or 1 in 200 pregnancies
- **2006 data**
  <0.1%, or 1 in 1600 pregnancies

Genetic testing

- **Amniocentesis in 2nd trimester**
  - sample of embryo cells
  - stain & photograph chromosomes
- **Analysis of karyotype**

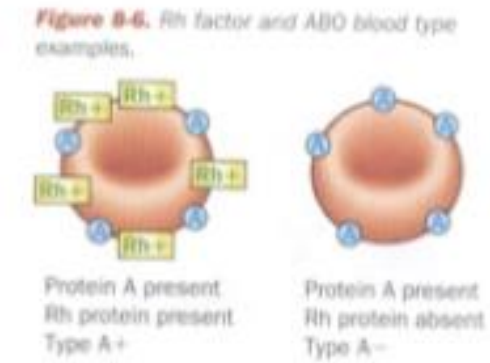




Rh factor: How is this important?

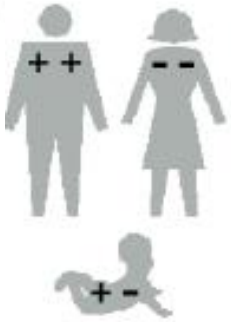
Rh Factor

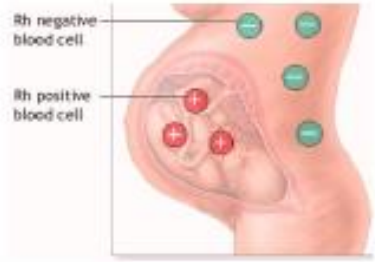
If you are Rh+, your blood cells have the Rh antigen and you do NOT make antibodies against yourself. If, however, you are Rh- your immune system will treat the Rh antigen on someone else's blood as foreign and produce Rh antibodies.

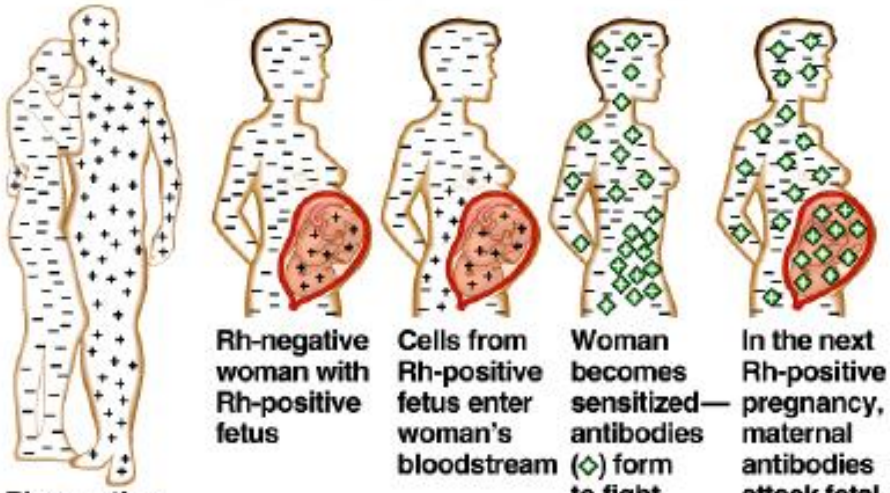


Rh Pregnancy Problems

A mother with Rh- blood is at risk in her second pregnancy if her first child is Rh+. The first child produces cells with the Rh marker which mingle with the mother's blood during the trauma of birth. The mother's immune system produces antibodies against the Rh marker. First kid is OK because he's born by this time.



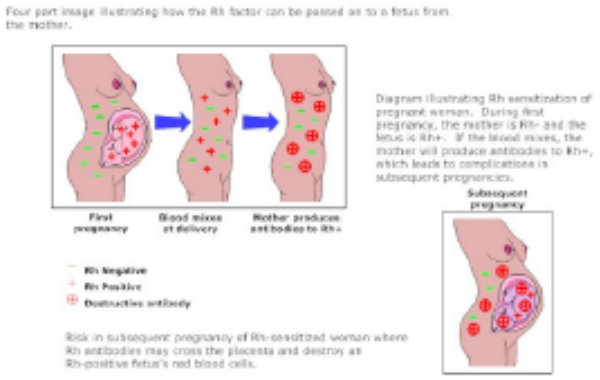




Rh Pregnancy Problems (cont.)

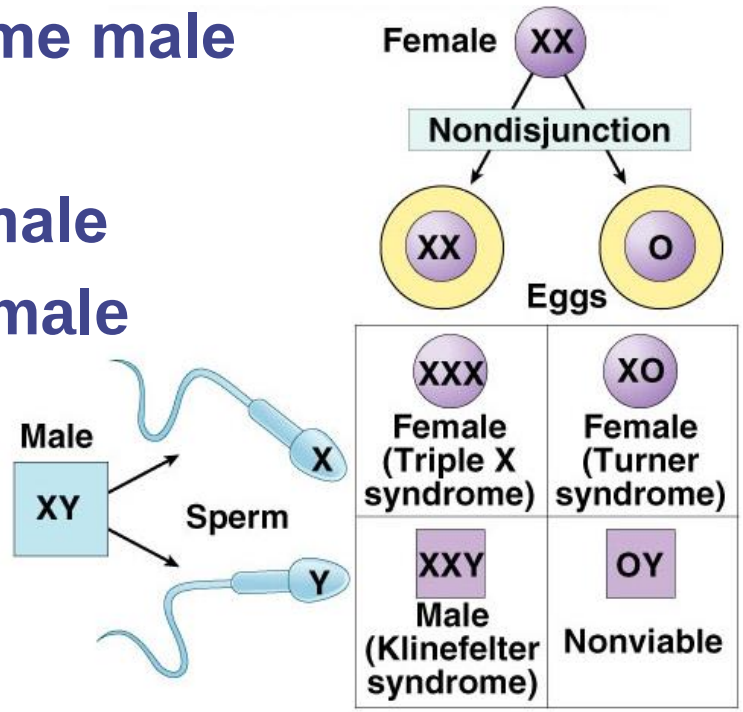
If the second fetus is Rh+, the mother's antibodies will attack the child's red blood cells. These antibodies can cross the placenta, whereas the antibodies for type A, B blood cannot. The mothers can be given a shot of Rh antibodies after a Rh+ birth to help out.

HOW DOES THIS HELP?



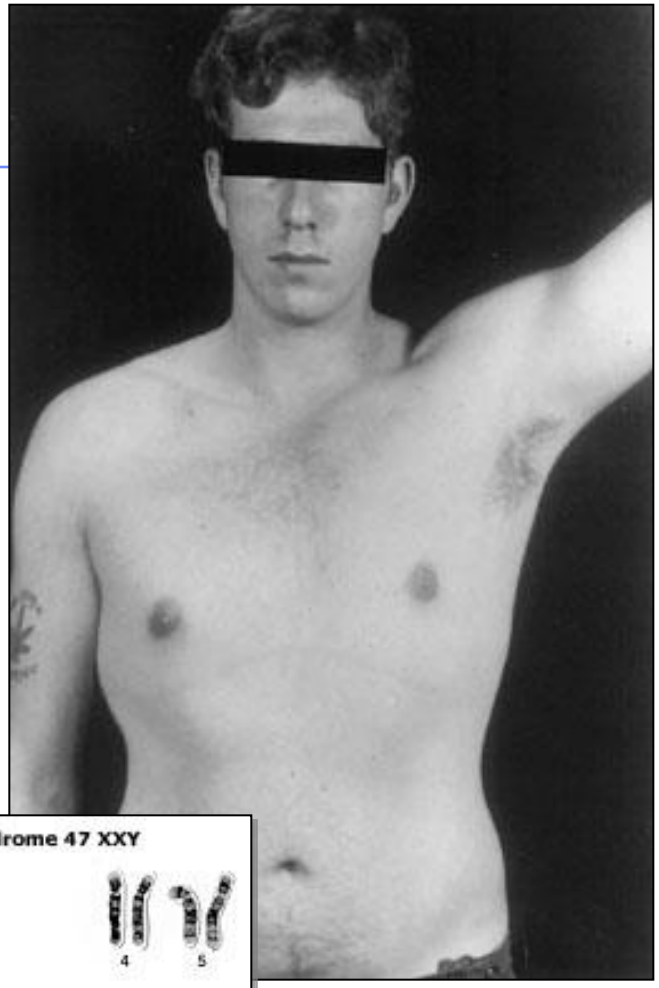
Sex chromosomes abnormalities

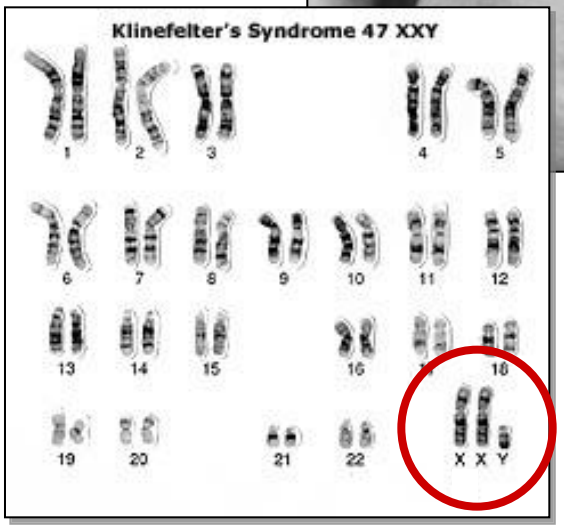
- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct syndromes in humans
  - XXY = Klinefelter's syndrome male
  - XXX = Trisomy X female
  - XYY = Jacob's syndrome male
  - XO = Turner syndrome female



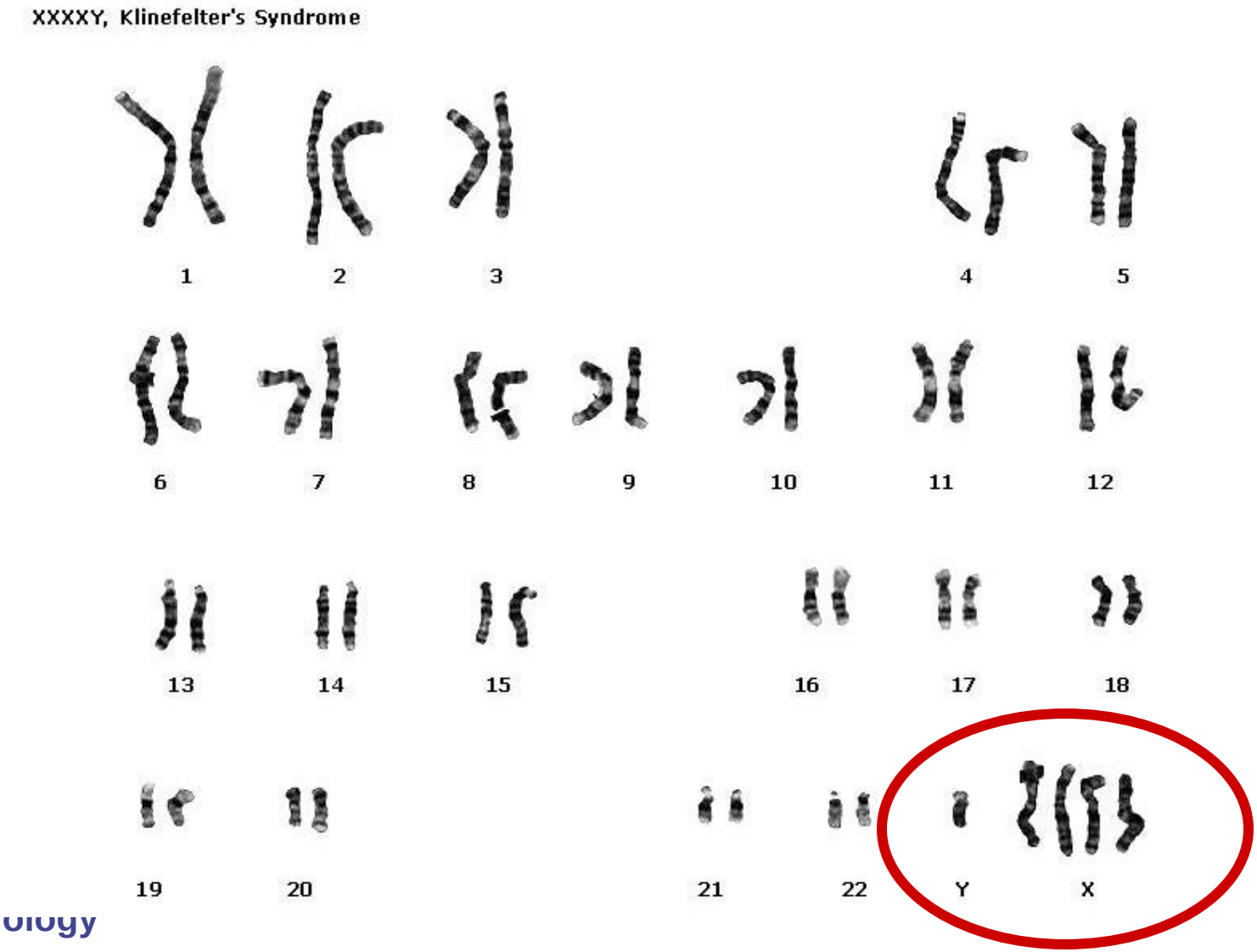
Klinefelter's syndrome

- XXY male
  - one in every 2000 live births
  - have male sex organs, but are sterile
  - feminine characteristics
    - some breast development
    - lack of facial hair
  - tall
  - normal intelligence



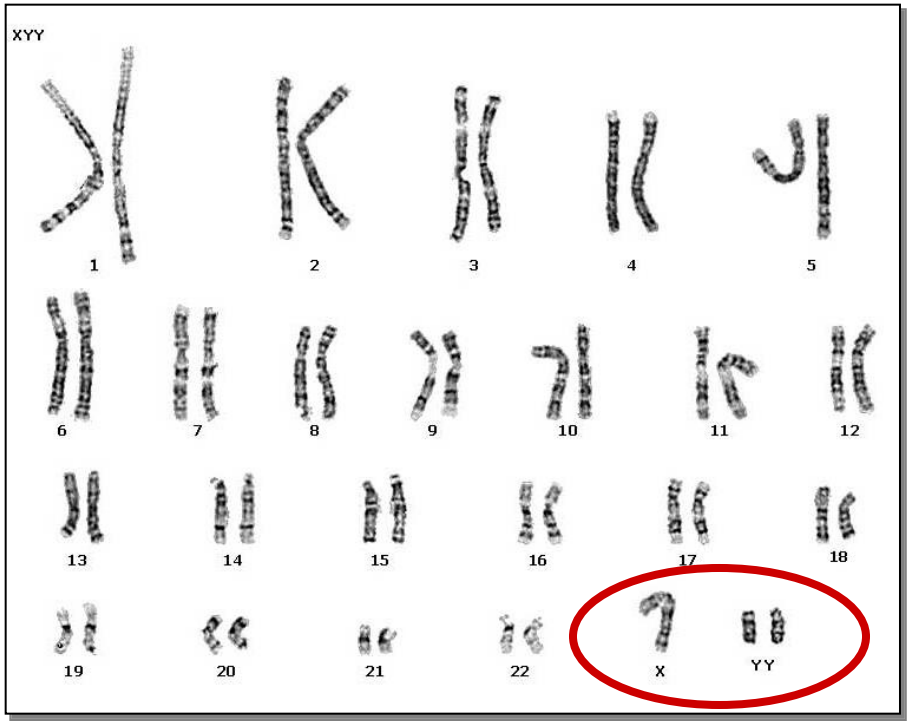


Klinefelter's syndrome



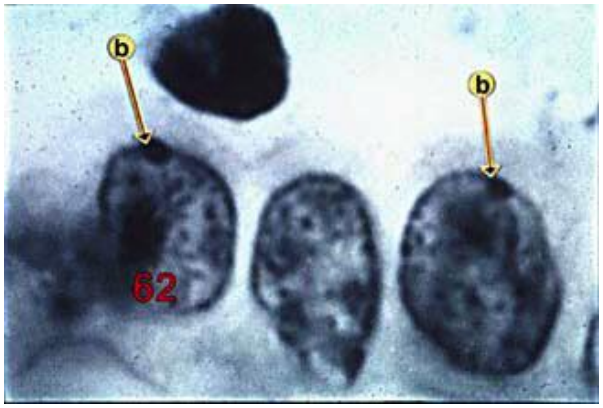
Jacob's syndrome male

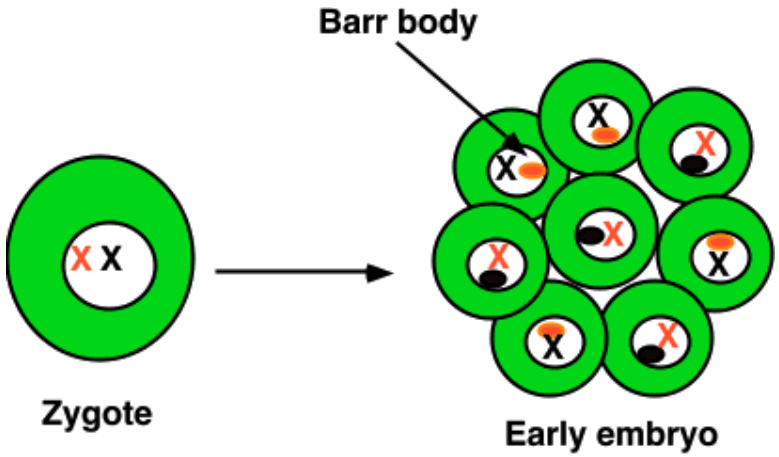
- **XYY Males**
  - 1 in 1000 live male births
  - extra Y chromosome
  - slightly taller than average
  - more active
  - normal intelligence, slight learning disabilities
  - delayed emotional maturity
  - normal sexual development



Trisomy X

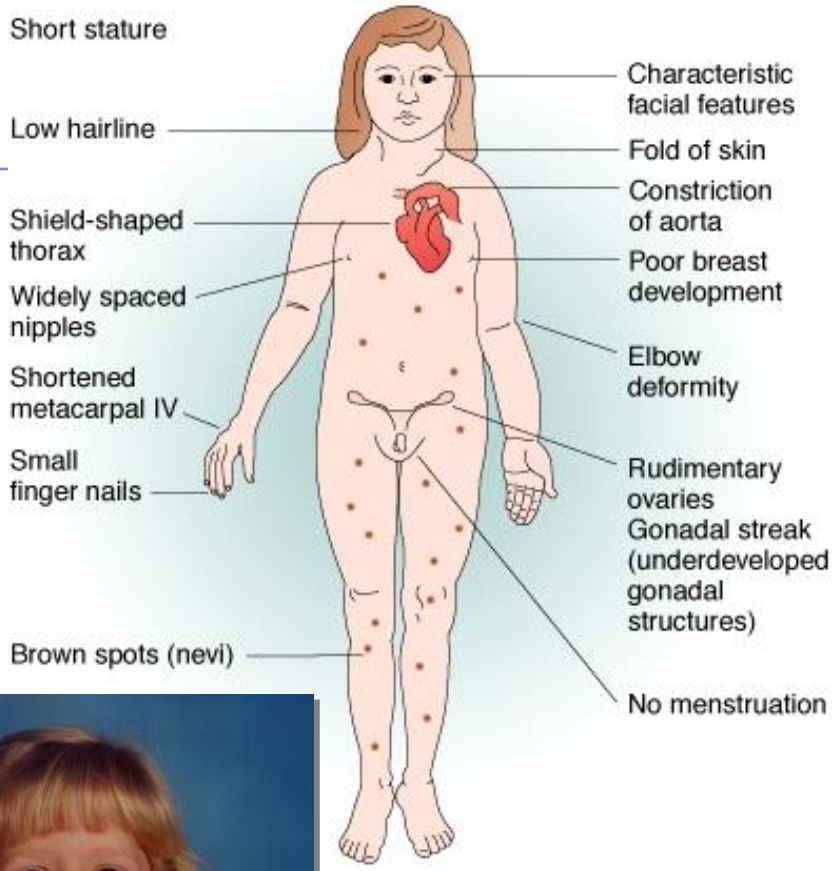
- XXX
  - 1 in every 2000 live births
  - produces healthy females
    - Why?
    - Barr bodies
      - all but one X chromosome is inactivated





Turner syndrome

- **Monosomy X or X0**
  - 1 in every 5000 births
  - varied degree of effects
  - webbed neck
  - short stature
  - sterile



Monday, April 13th

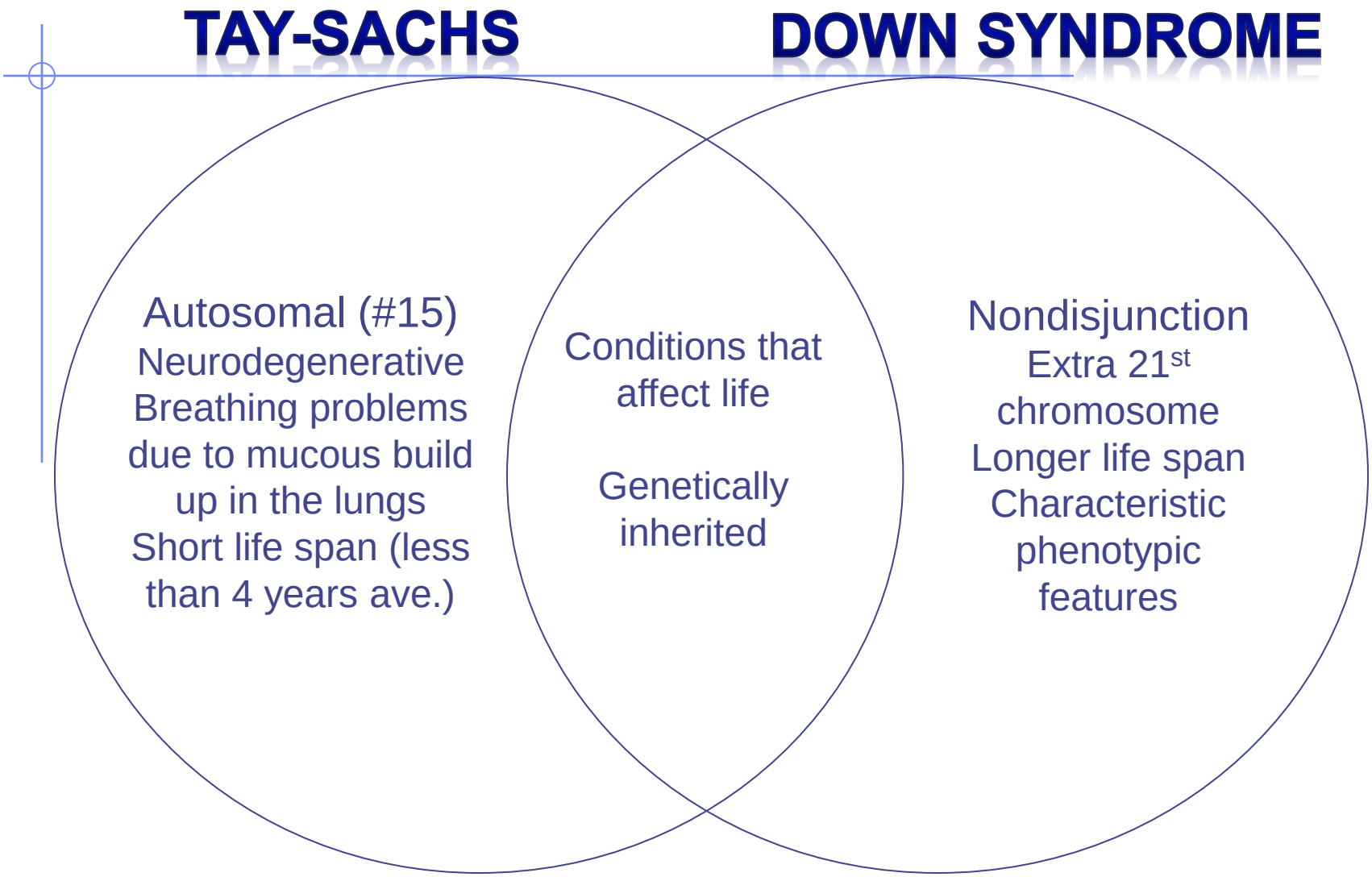
Happy Monday! Okay, we have some work to do, so let's get started…

QUESTION TO PONDER

*Please take out your chapter 15 reading guide. On the back, set up a Venn diagram to compare and contrast **Down Syndrome** with **Tay-Sachs Disease**.*

Today I will…

1. ***Compare*** various genetically inherited conditions.
2. ***Contrast*** the four main types of chromosomal breakage.


TAY-SACHS
DOWN SYNDROME
Autosomal (#15)
Neurodegenerative
Breathing problems due to mucous build up in the lungs
Short life span (less than 4 years ave.)
Conditions that affect life
Genetically inherited
Nondisjunction
Extra 21st chromosome
Longer life span
Characteristic phenotypic features

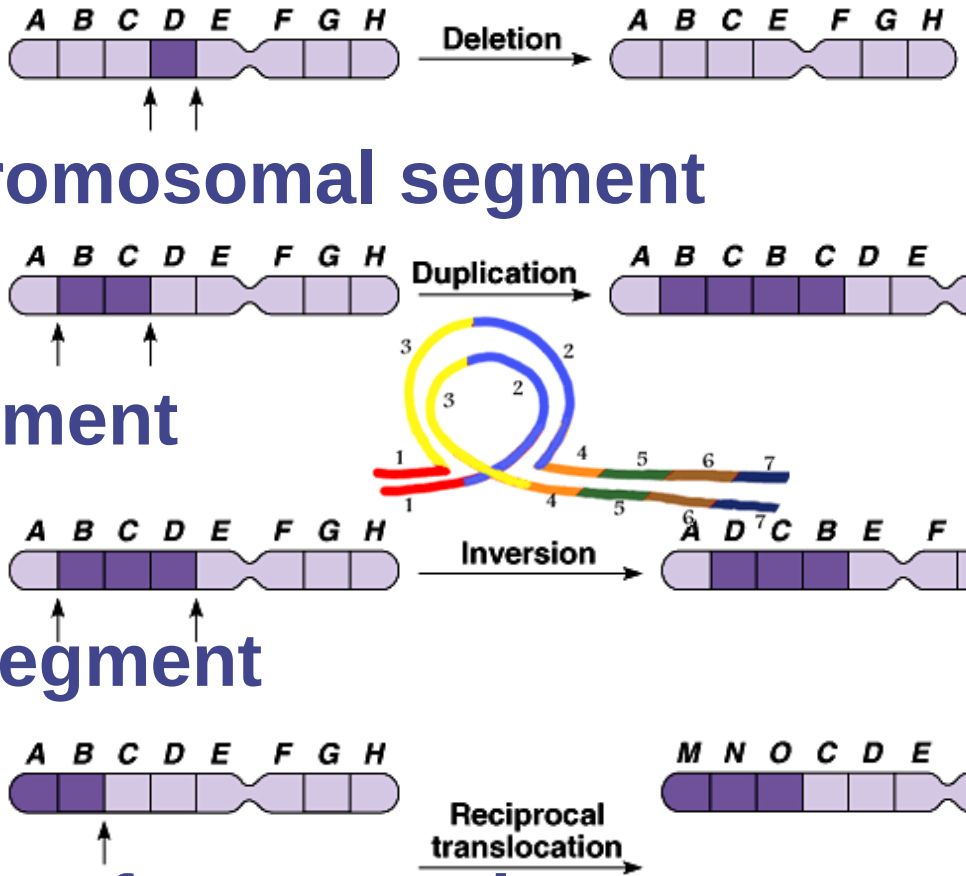
Changes in chromosome structure

error of replication

- **deletion**
  - loss of a chromosomal segment
- **duplication**
  - repeat a segment

error of crossing over

- **inversion**
  - reverses a segment
- **translocation**
  - move segment from one chromosome to another



Tuesday, April 14th

Good morning! Today you will prepare for your **UNIT 5, GENETICS**, exam.

- Please be certain you have a study guide.
- Please complete your "Pink Packet of FUN"…word problems!

*The multiple choice portion is **WEDNESDAY**.*

*The essay portion is **THURSDAY**.*

I will **compare** and **contrast** various terms and inheritance patterns associated with heredity. I will **distinguish** amongst various genotypes and phenotypes, using mono- and dihybrid crosses.

Essay ideas…

1. Choose **two** genetic conditions and compare and contrast. How are each of these inherited? Symptoms?
2. Summarize the work of Gregor Mendel. State the laws of inheritance (segregation vs. independent assortment)
3. Describe the stages of meiosis cell division in humans. Explain crossing over and nondisjunction events.
4. Contrast the work of Mendel and Morgan.