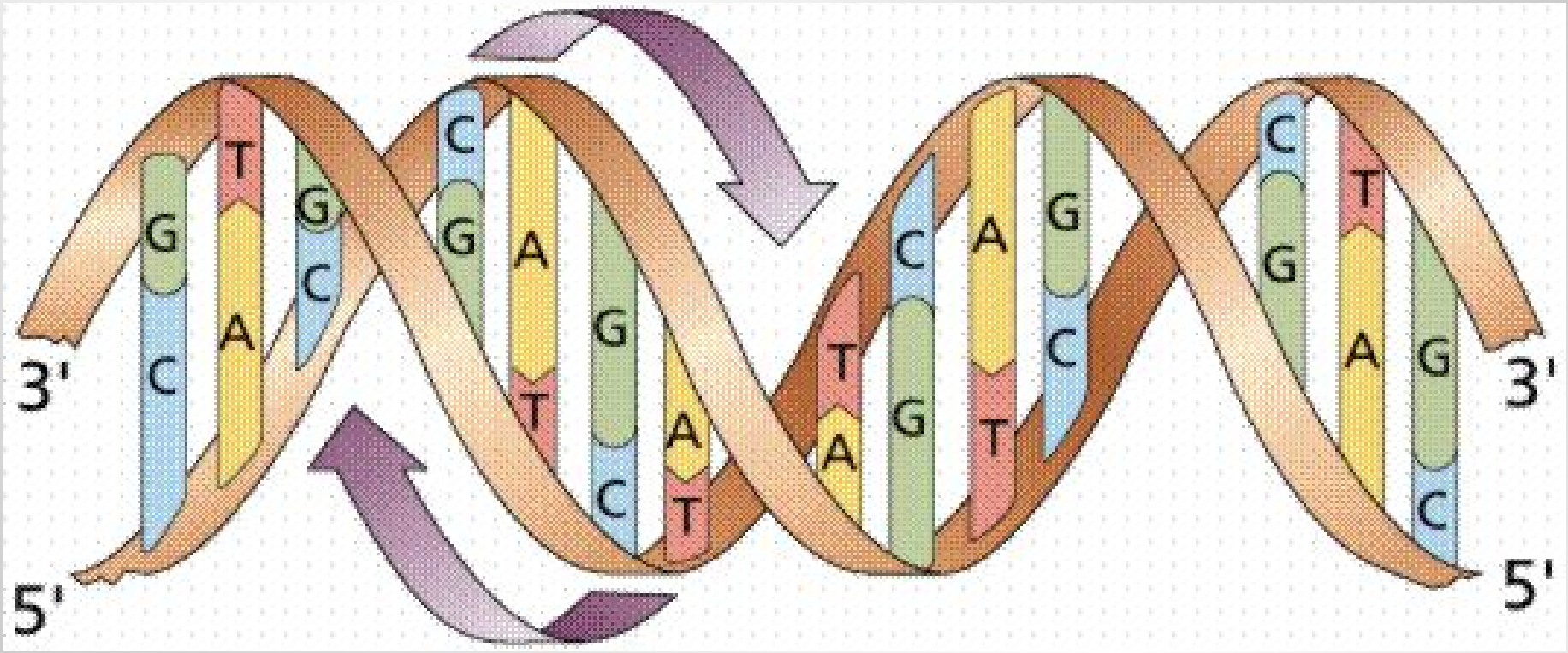
Notes PG#1: Mitosis, Meiosis & Chromosomal Disorders

EQ1: What is the difference between Mitosis and Meiosis?

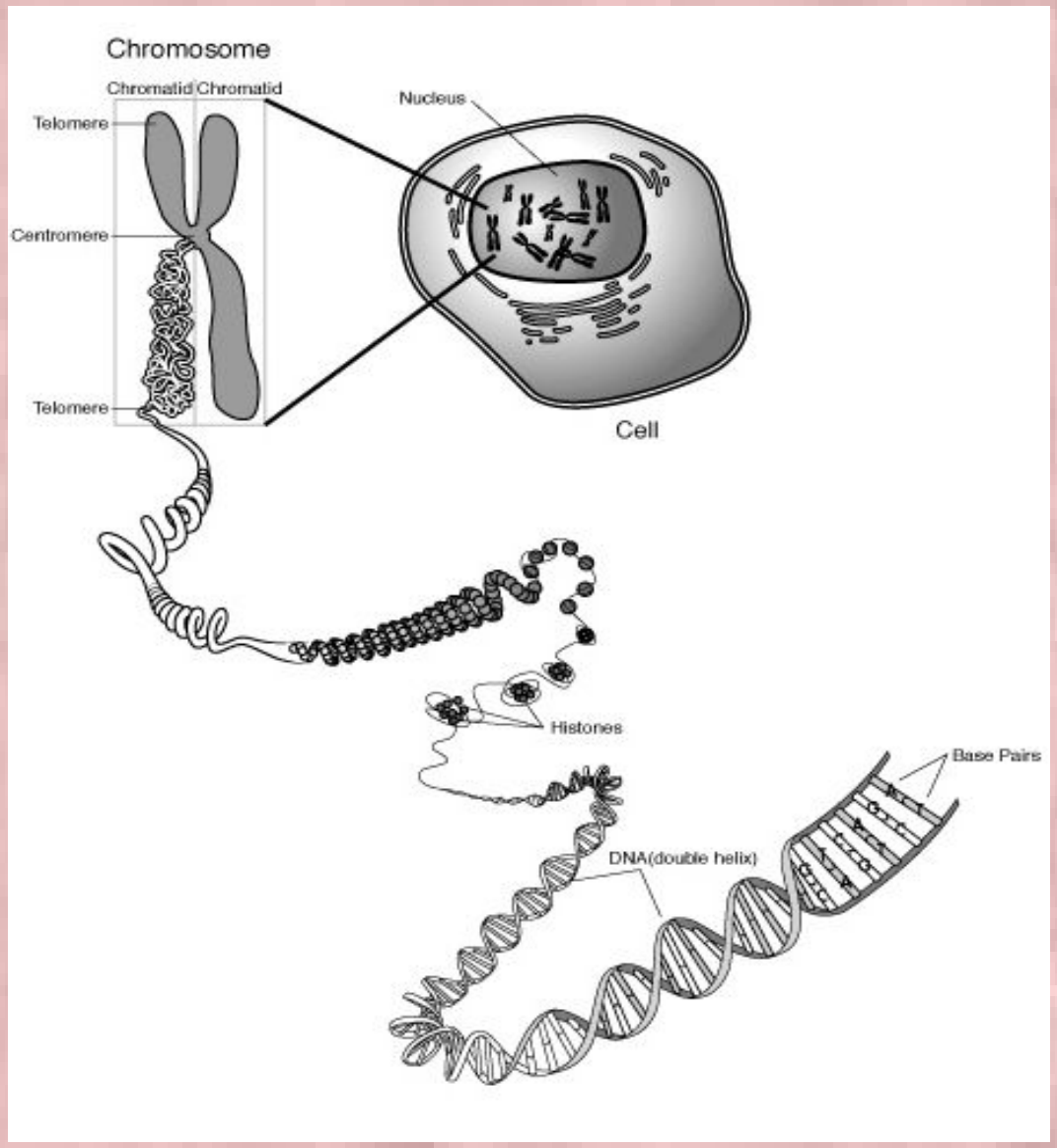
EQ2: What is the difference between haploid and diploid?

DNA is the molecule that transfers hereditary information from one cell to the next.

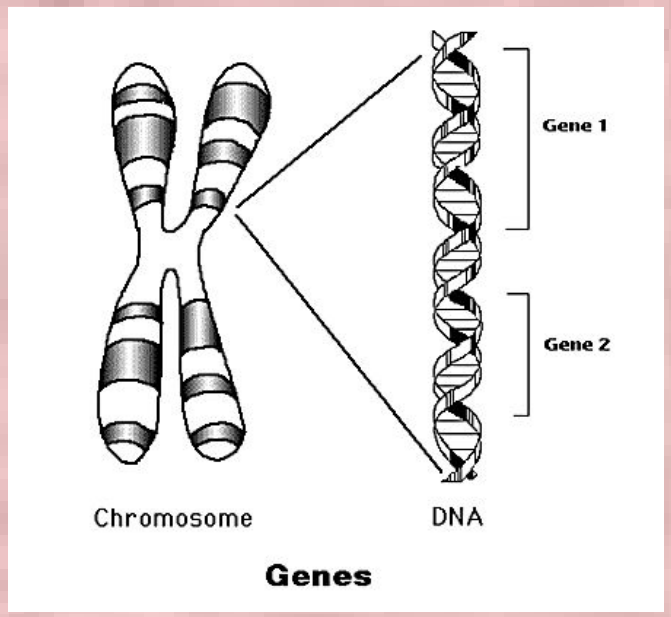


This is a model of the double helix shape of DNA.

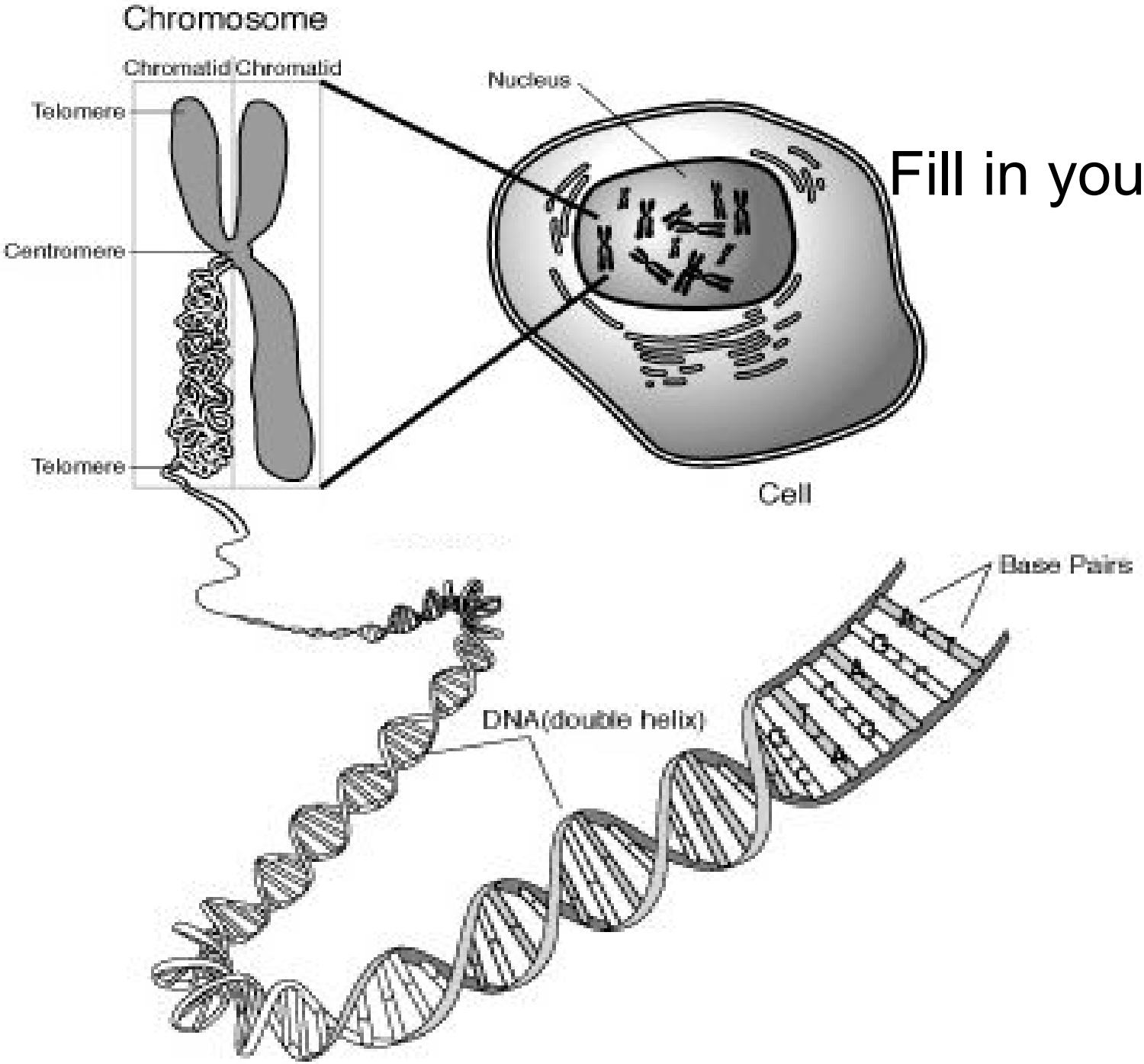
When a cell is dividing, DNA winds up tightly and forms chromosomes in the nucleus of the cell. A Copy of DNA is needed in all cells.



A specific portion of the DNA is called a gene, which has 1 piece of genetic information.
Ex. Hair color is a gene.



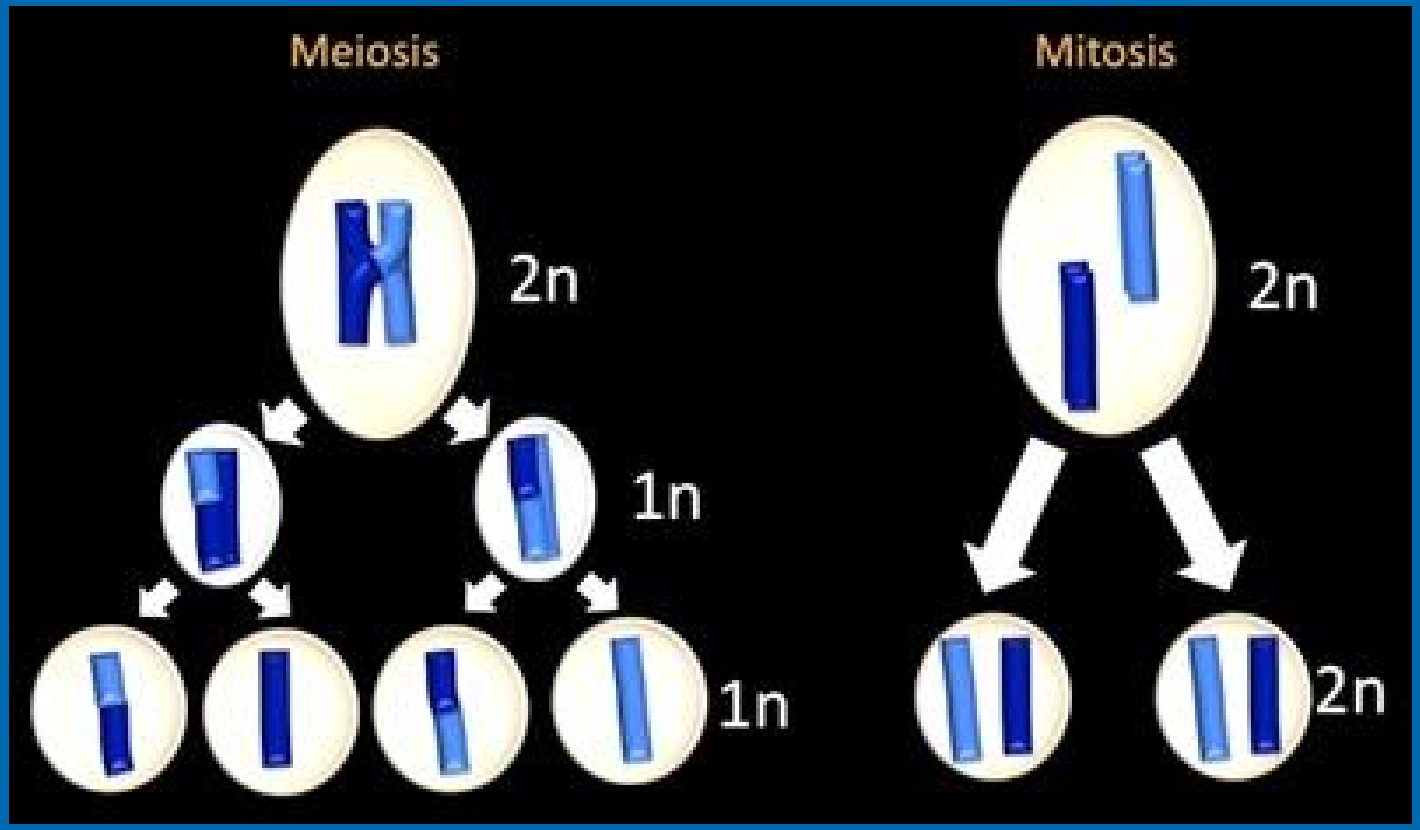
The genes are contained within the chromosome.



Fill in your diagram!

Mitosis & Meiosis

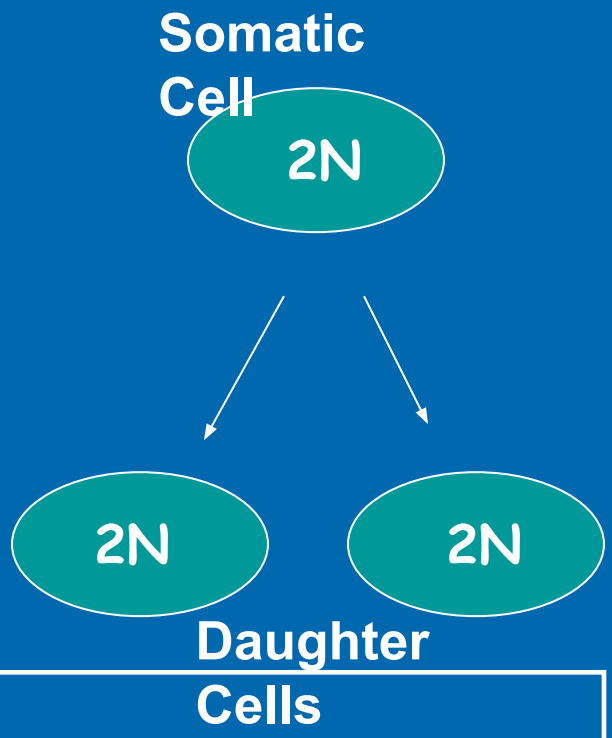
- Mitosis purpose: **Copy** cell & DNA inside, ex. heal wounds
- Meiosis purpose: Make genetically **unique** gametes that can combine with other parent gamete ex. Make babies



MITOSIS

DRAW:

When somatic (body) cells reproduce themselves the process is called MITOSIS.



Main purposes of mitosis:

1. Growth
2. Repair
3. Asexual reproduction (bacteria)

Mitosis leads to the production of two *"daughter cells"*.
Each daughter cell has the same number of chromosomes as the parent cell.

- All cells made by Mitosis are diploid, containing 2 copies of each chromosome (2N).

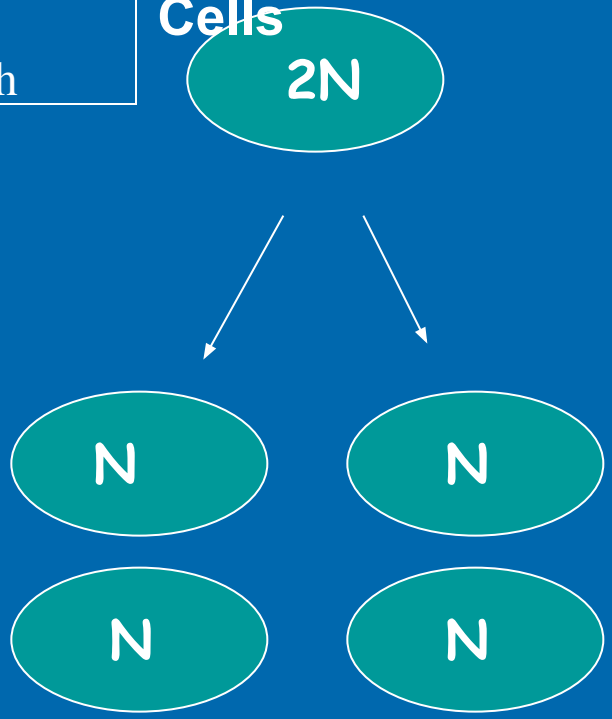
MEIOSIS

DRAW:

When gamete (sperm or egg) cells reproduce themselves the process is called MEIOSIS. Meiosis reduces chromosome # in half.

- Each cell made by Meiosis contains only 1 copy of each chromosome (n).

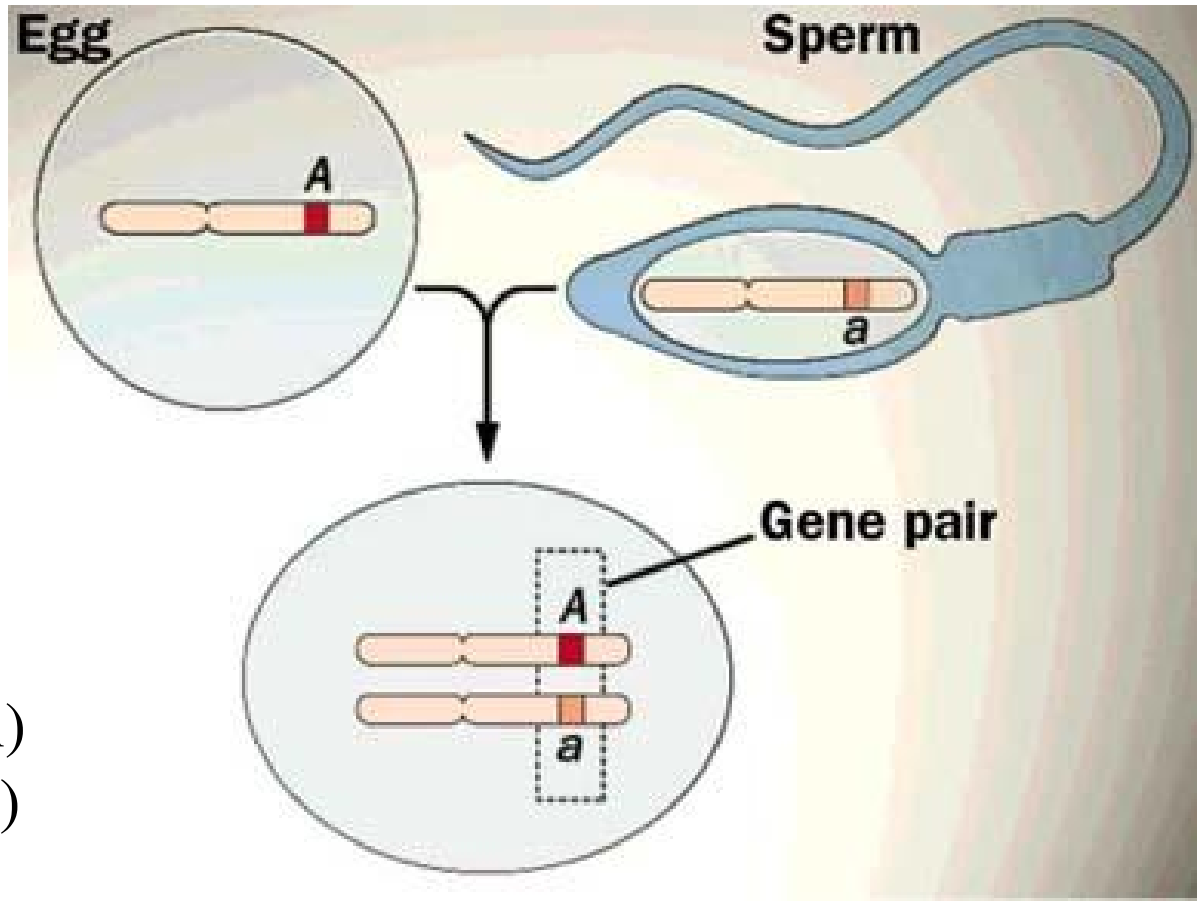
Making Gamete Cells



During meiosis, a single diploid cell divides and produces FOUR genetically different haploid cells.

In Meiosis crossing over occurs, where the chromosomes next to each other (homologous chromosomes) swap genes, this increases genetic variety in the potential offspring.

Gametes (n)
Made by
Meiosis



Somatic Cells (2n)
Reproduce (clone)
through Mitosis

In animals…

Somatic cells (cells of the body) are ***diploid***. This means that each cell has two chromosomes of each type. They are in PAIRS. One from mom and one from dad. These are the cells used to make karyotypes. Made through MITOSIS.

⇒ Biologists use "**2N**" to symbolize ***diploid***.

Gamete cells (egg, sperm) are ***haploid***. This means that each cell has only ONE of each type of chromosome. *Why?* Made through MEIOSIS.

⇒ Biologists use "**N**" to symbolize ***haploid***.

Mitosis or Meiosis???

- 1. Asexual Reproduction? _____
- 2. Results in 2 identical cells? _____
- 3. Results in 4 non-identical cells? _____
- 4. Produces gametes? _____
- 5. Results in variation? _____
- 6. Results in haploid cells? _____
- 7. Used for growth and repair? _____
- 8. Skin cells? _____
- 9. Sperm cells? _____
- 10. Makes 2N cells? _____

Sex Chromosomes: A single pair of chromosomes that determines the gender of the organism. Always put last, pair #23.

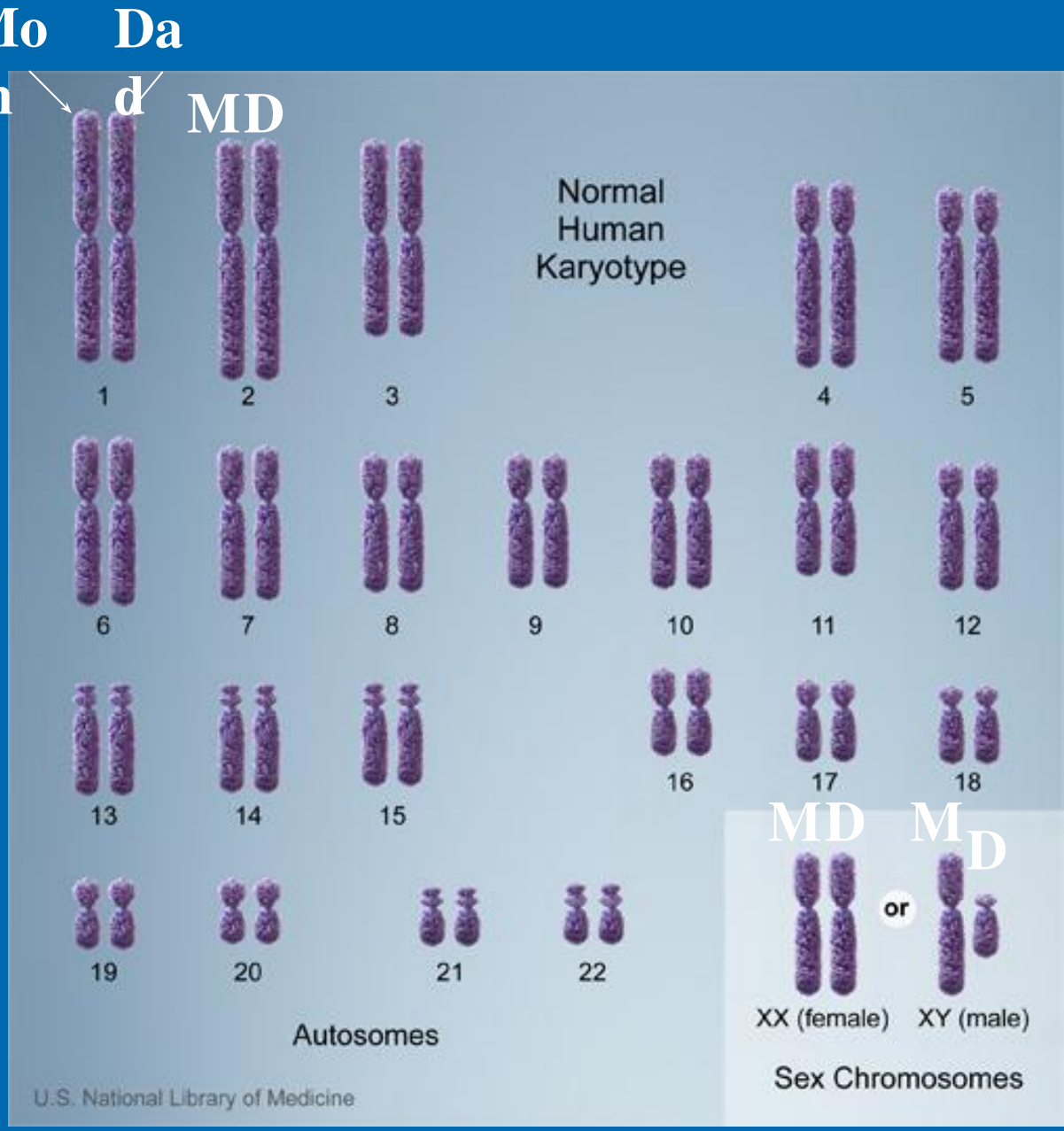
-Female = XX

-Male =XY

-Do NOT always match by size- XY!

Autosomes: All other chromosomes not involved in gender determination. Pairs #1-22, Put from largest to smallest.

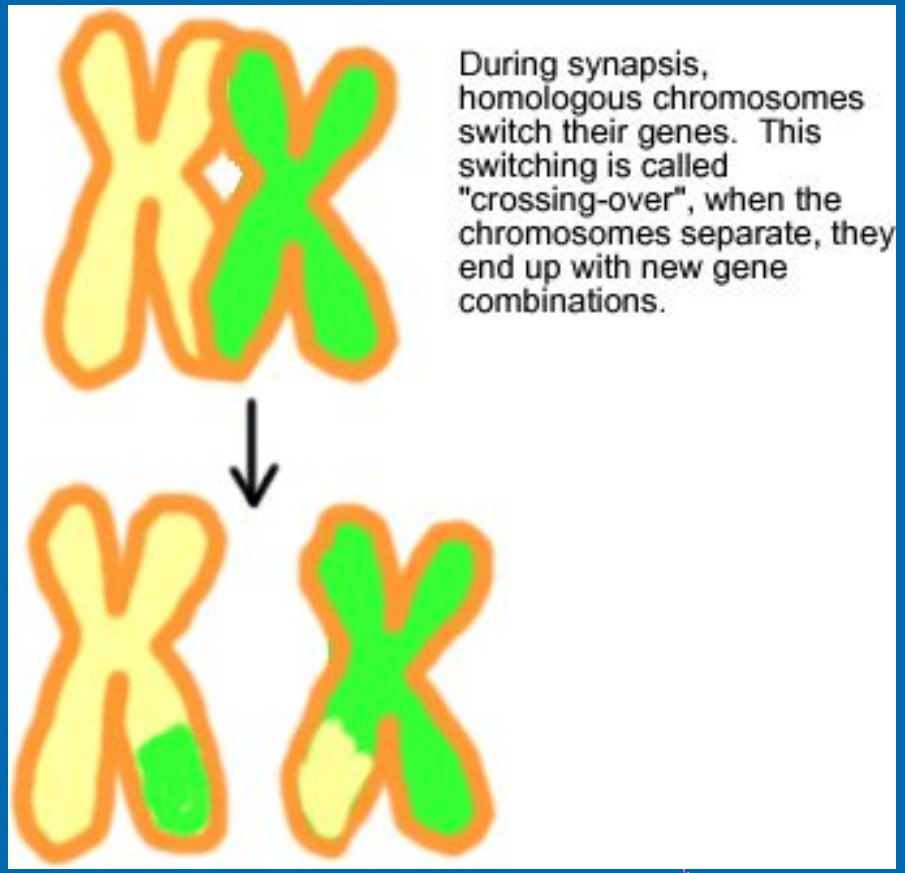
-Matched by same size, *like socks!*



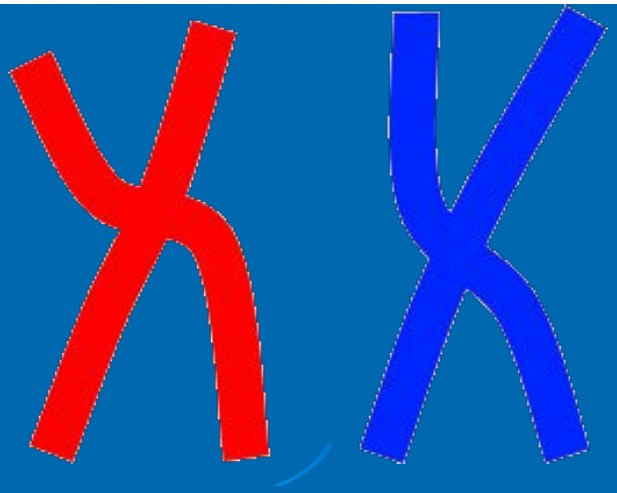
Arts & Crafts: Karyotyping

No, the chromosomes in your cells are not usually neatly paired up from 1 to 23. First, scientists have to stain the cell and view it with a powerful microscope. They then take a photograph of the chromosomes in the cell, print it out, cut out the chromosomes and find the matching pairs by same size. They then line them up from largest to smallest with the XX or XY at the end.

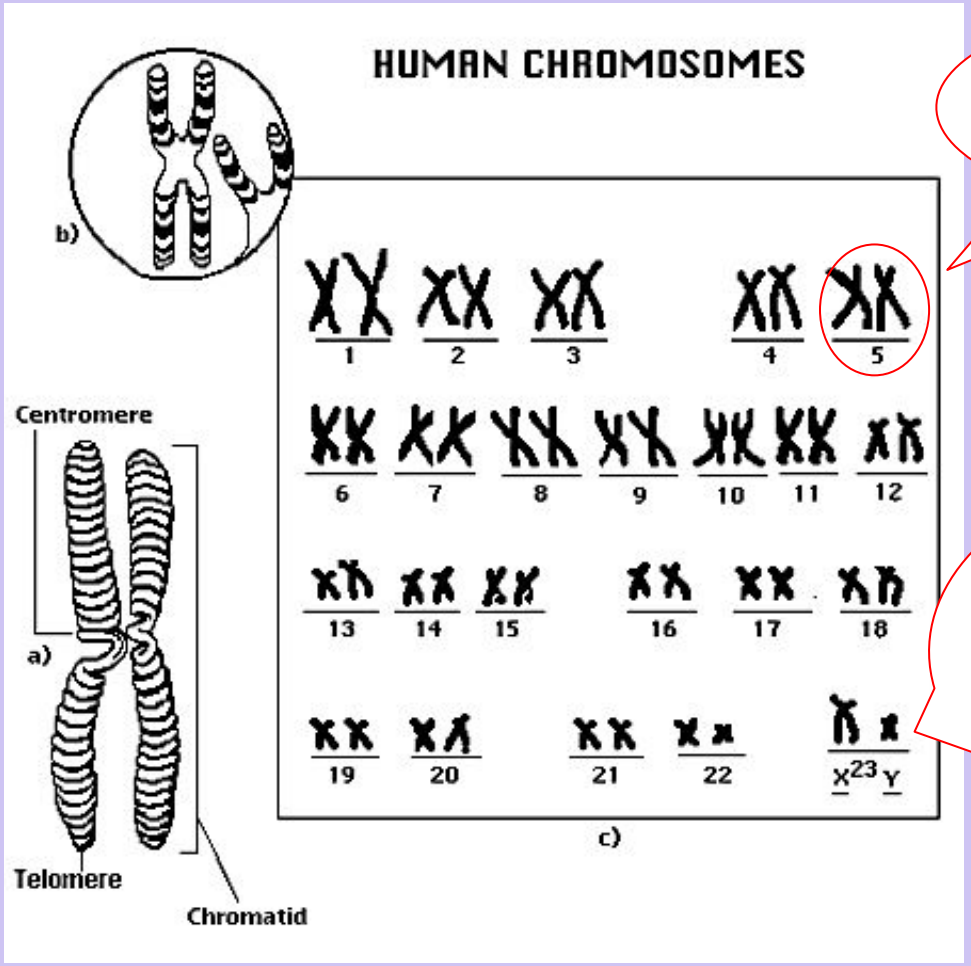
- Meiosis: Crossing Over: Homologous Chromosomes swap (switch) some genes during MEIOSIS only. This creates variety in the gametes.



- This is why kids who have the same two parents don't have to look identical, or even alike. Or, conversely, why two siblings might look exactly like their sibling- if they had the same cross-overs.



A **karyotype** is a picture showing the arrangement of a full set of human chromosomes.

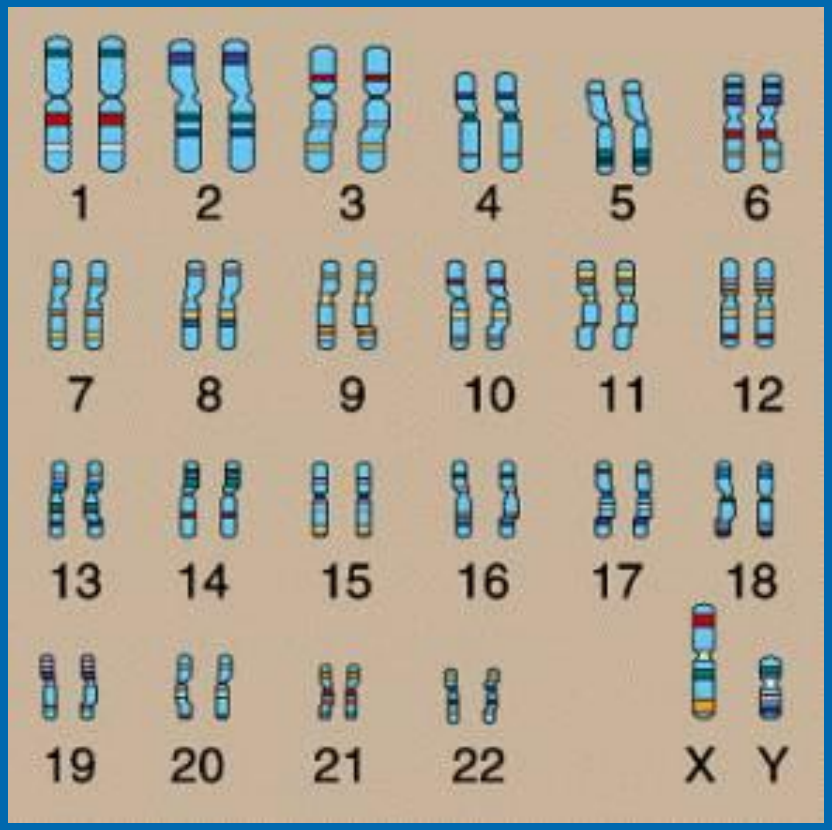


-Humans have 46 (23 pairs- half from mom, half from dad) chromosomes

-Each pair is called "*homologous*," meaning "same" genes

Practice

- Is this karyotype from a male or female human?



Practice

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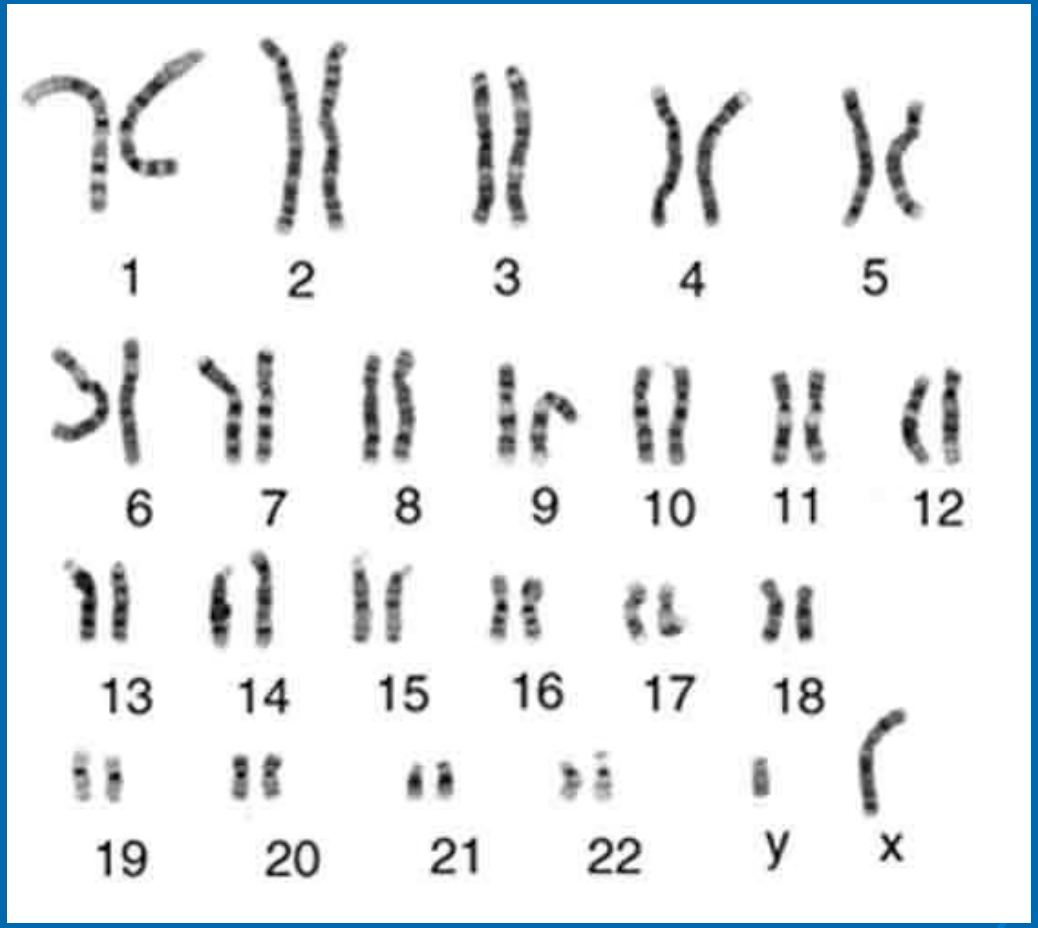

Practice

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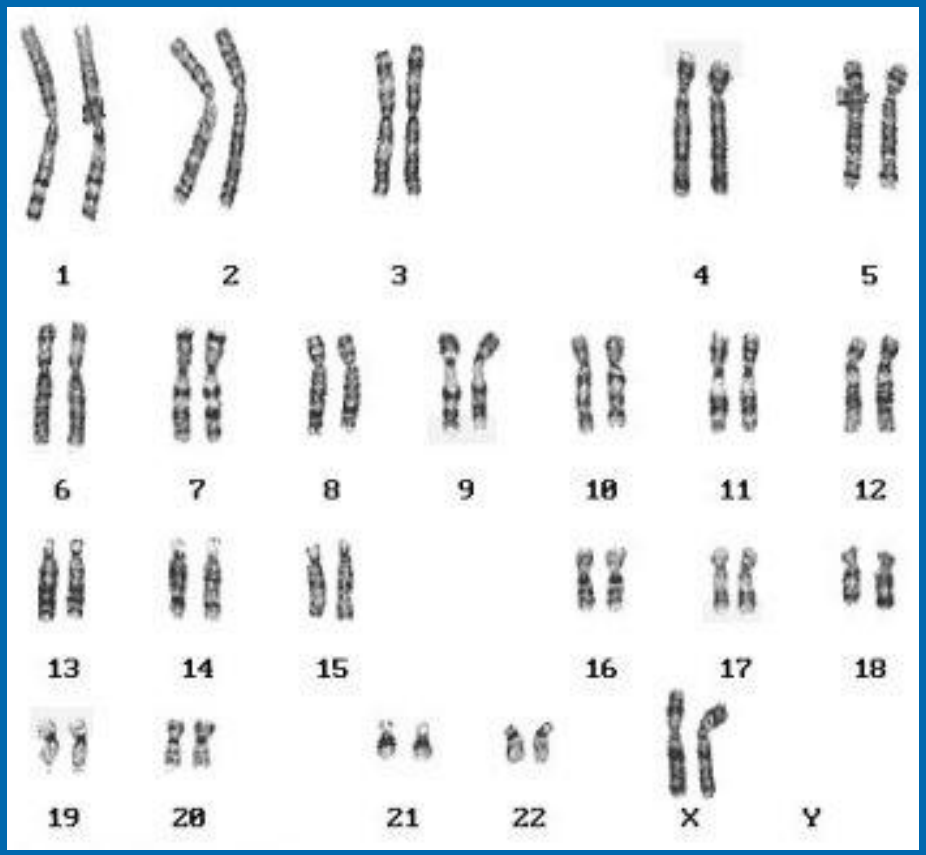
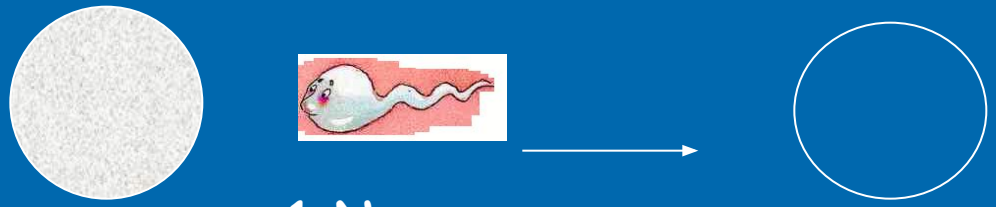


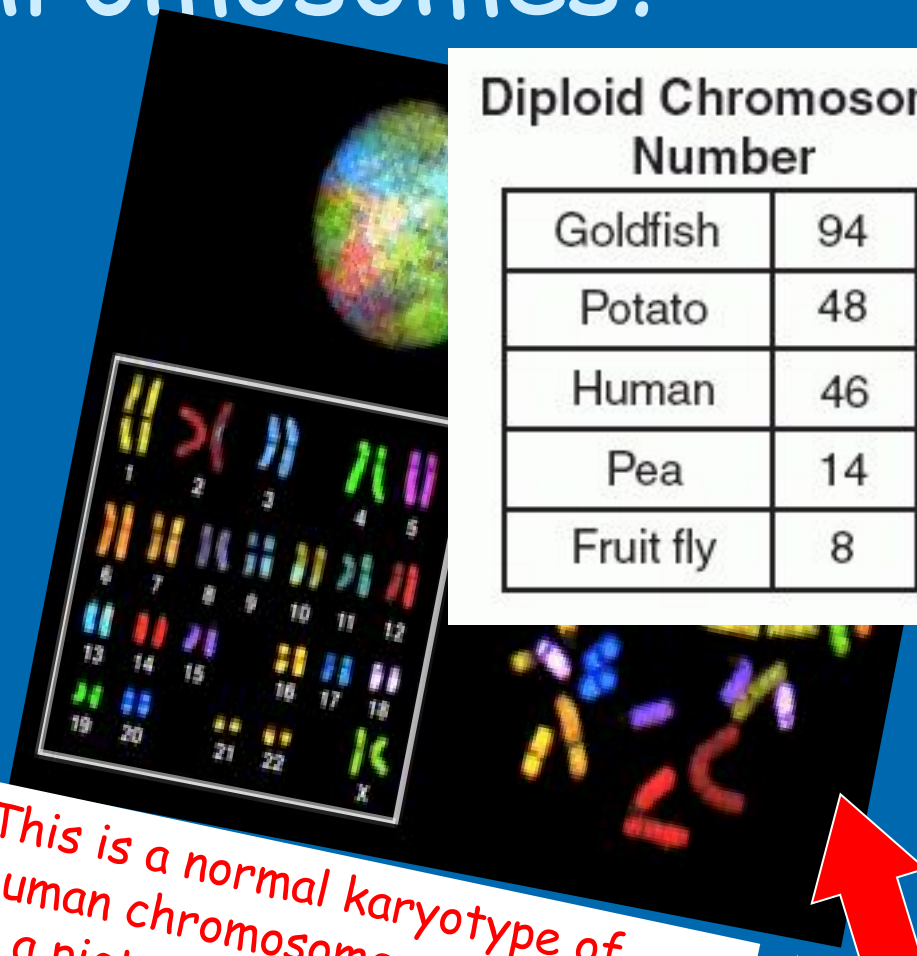
Fertilization for a diploid offspring

Upon fertilization, a "N" sperm meets a "N" egg and a zygote (2N) is formed.



What are chromosomes?

- *Humans* have 23 **pairs (2n=46)** of chromosomes, with one chromosome from each parent. The chromosomes are coiled up DNA.
- Under normal conditions all of the chromosomes are inherited intact and in pairs.



Diploid Chromosome Number

Goldfish	94
Potato	48
Human	46
Pea	14
Fruit fly	8

This is a normal karyotype of human chromosomes. A karyotype is a picture of chromosomes lined up to look at and compare.

4 Chromosomal Disorders

- Deletion: when cells go through meiosis, portions of the chromosome are lost.
- Inversion: when cells go through meiosis, parts of the chromosome are flipped.
- Translocation: when cells go through meiosis, parts of the chromosomes stick together and switch.
- Non-disjunction: when cells go through meiosis the chromosomes don't separate correctly and either too many or not enough are passed on.

Down Syndrome

1 in 31,000 births

46 chromosomes
XY=97%
XX=3%

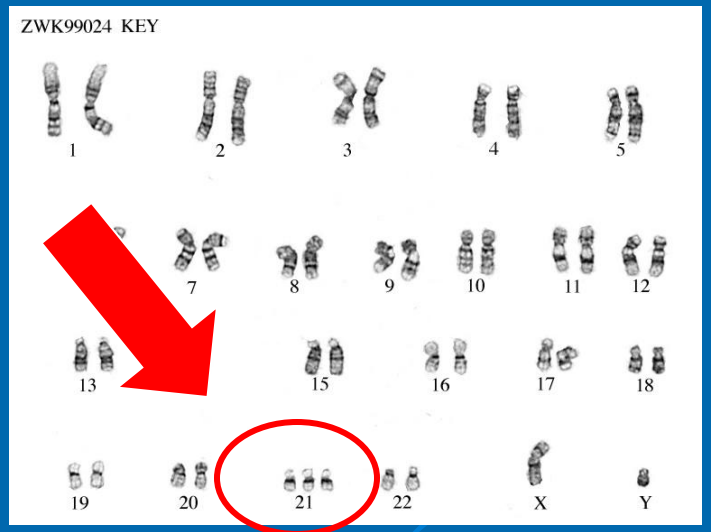
#14/21 Translocation

Down Syndrome: Trisomy

90-95% of Downs cases
1 in 1,250 births

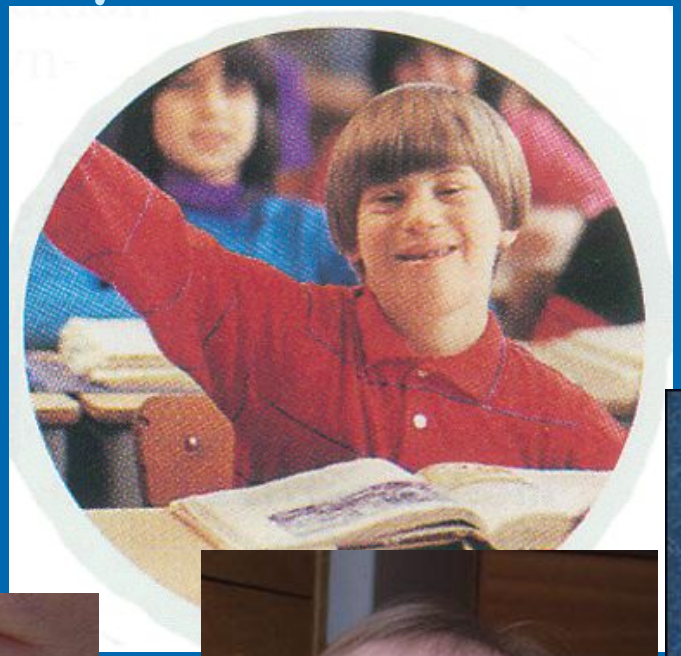
47 chromosomes
XY or XX

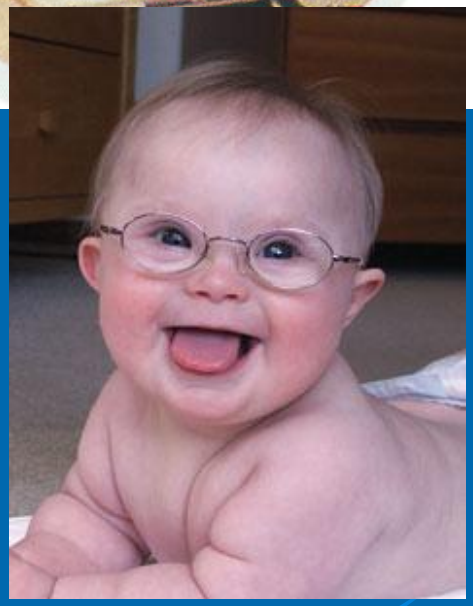
#21 Trisomy Non-disjunction



Down Syndrome

Short, broad hands
Stubby fingers
Rough skin
Impotency in males
Mentally retarded
Small round face
Protruding tongue



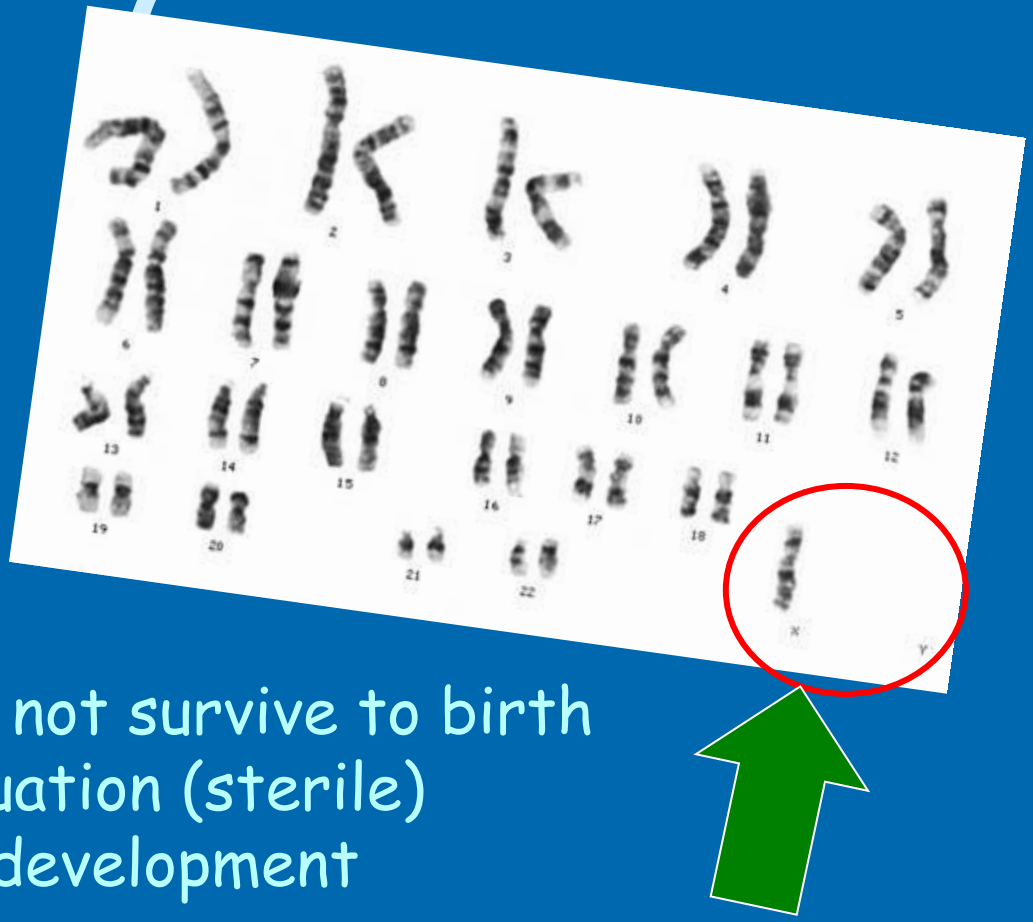


Non-disjunction: Turners Syndrome

1 in 5,000 births

45 chromosomes X only

#23 Monosomy
Nondisjunction



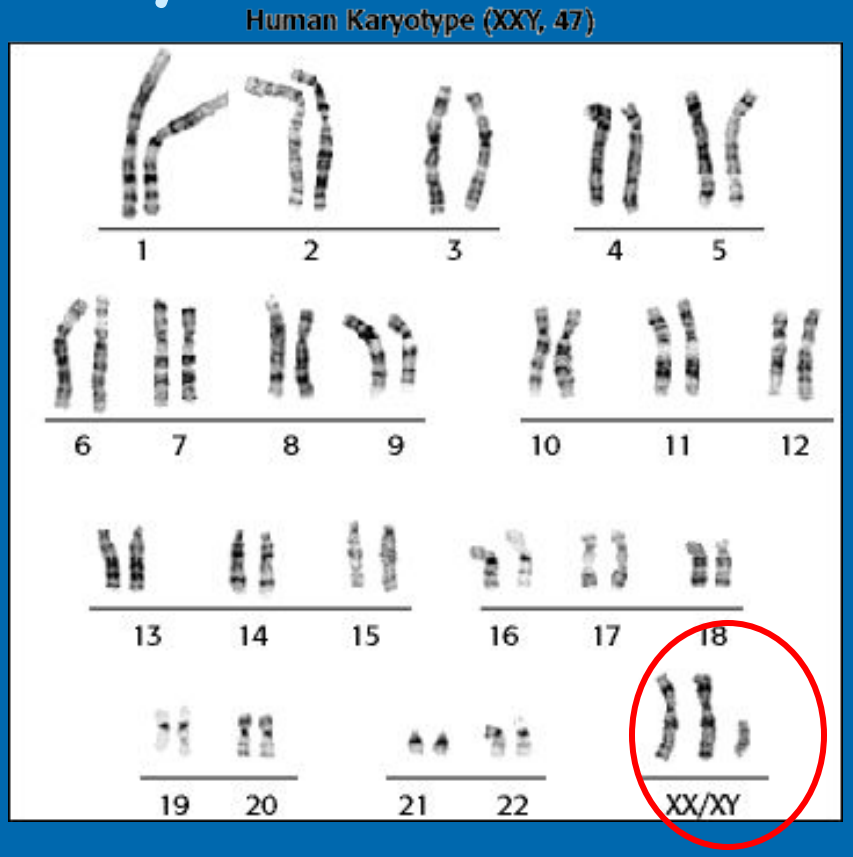
Symptoms:
96-98% do not survive to birth
No menstruation (sterile)
No breast development
Narrow hips
Broad shoulders and neck

Klinefelter Syndrome

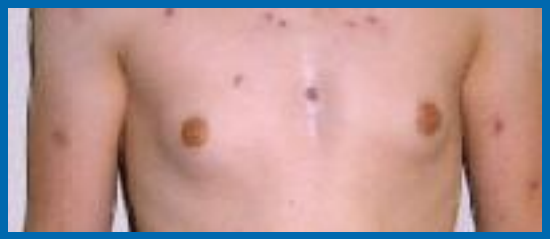
1 in 1,100 births

47 chromosomes
XXY only

#23 Trisomy
Nondisjunction

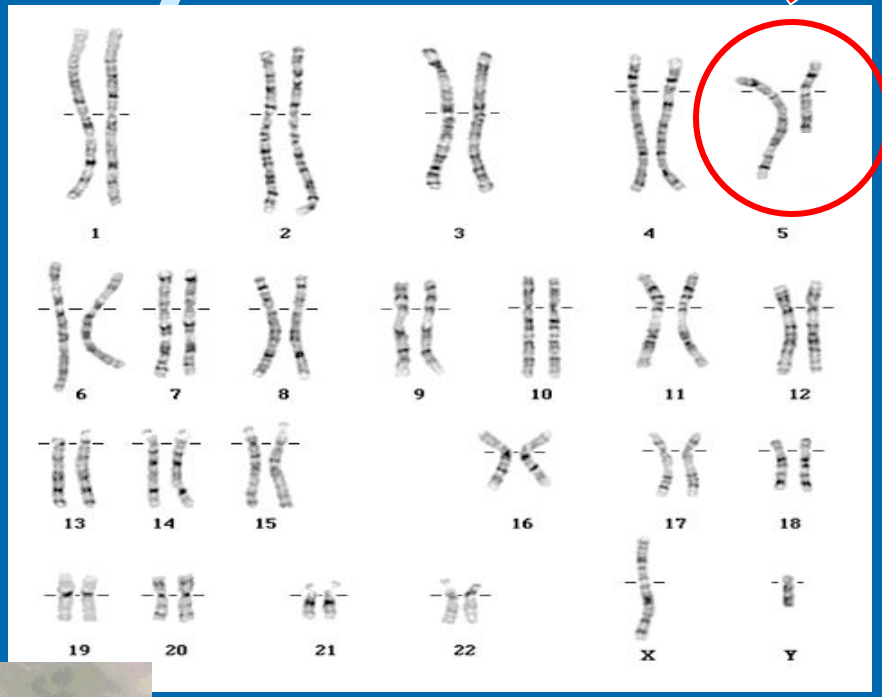


Scarce beard
Longer fingers and arms
Sterile
Delicate skin
Low mental ability
Normal lifespan

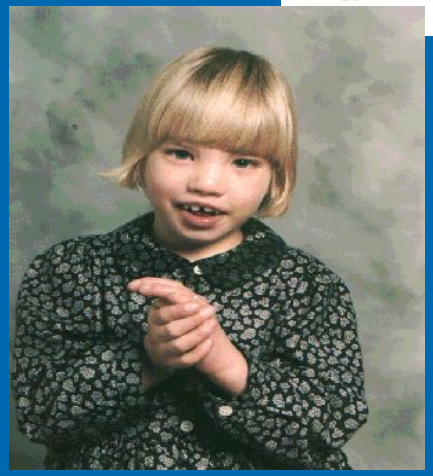


Deletion: Cri-Du-Chat Syndrome

- 1 in 216,000 births
- 46 chromosomes
- #5 Deletion of lower arm



Symptoms:
Moon-shaped face
Heart disease
Mentally retarded
Malformed larynx
Normal lifespan

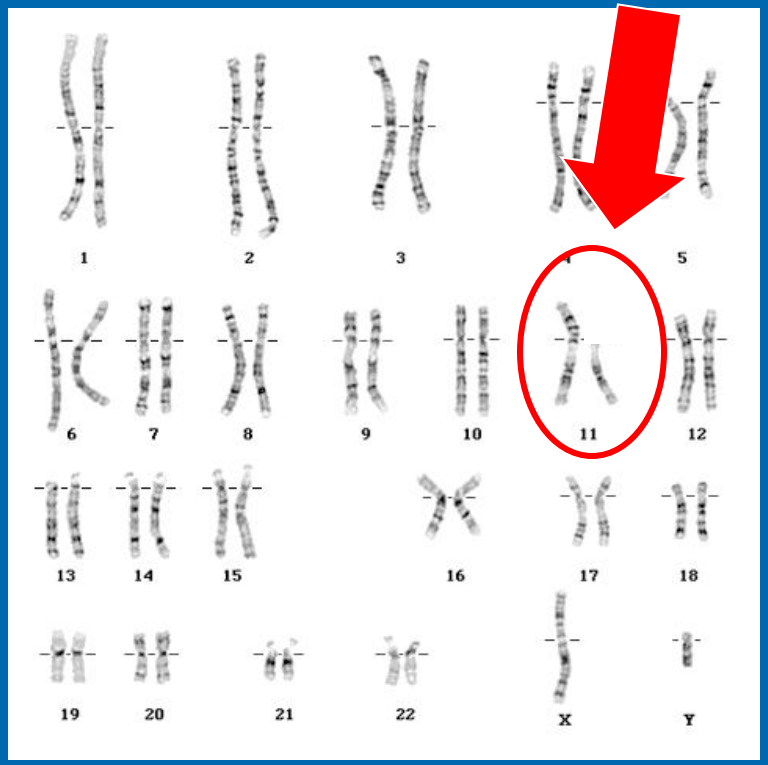


Deletion: Aniridia-Wilms Tumor Syndrome

1 in 50,000,000 births

46 chromosomes
XY or XX

#11 Deletion of upper arm



Symptoms:
Mentally retarded
Growth retarded
Blindness
Tumors on kidneys
Short lifespan

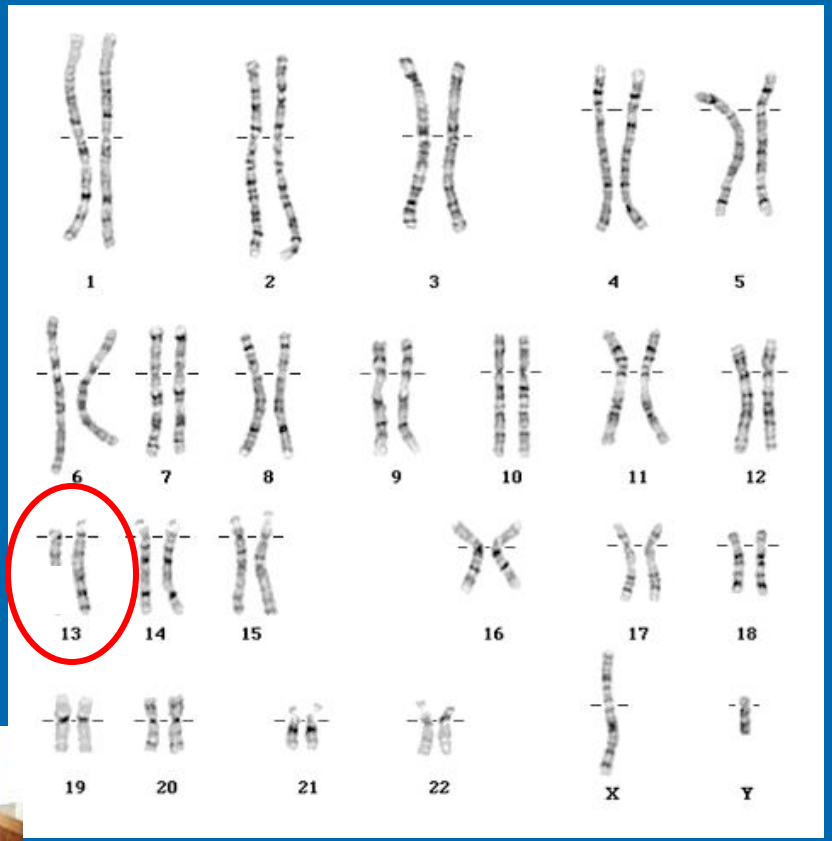
Thirteen Q Deletion Syndrome

1 in 500,000 births

46 chromosomes
XY or XX

#13 Deletion of lower arm

Mentally retarded
Deformed face
No thumbs
Heart disease
Short lifespan



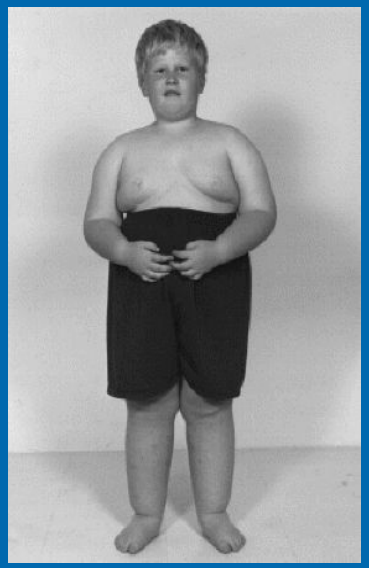
Deletion: Prader-Willi Syndrome

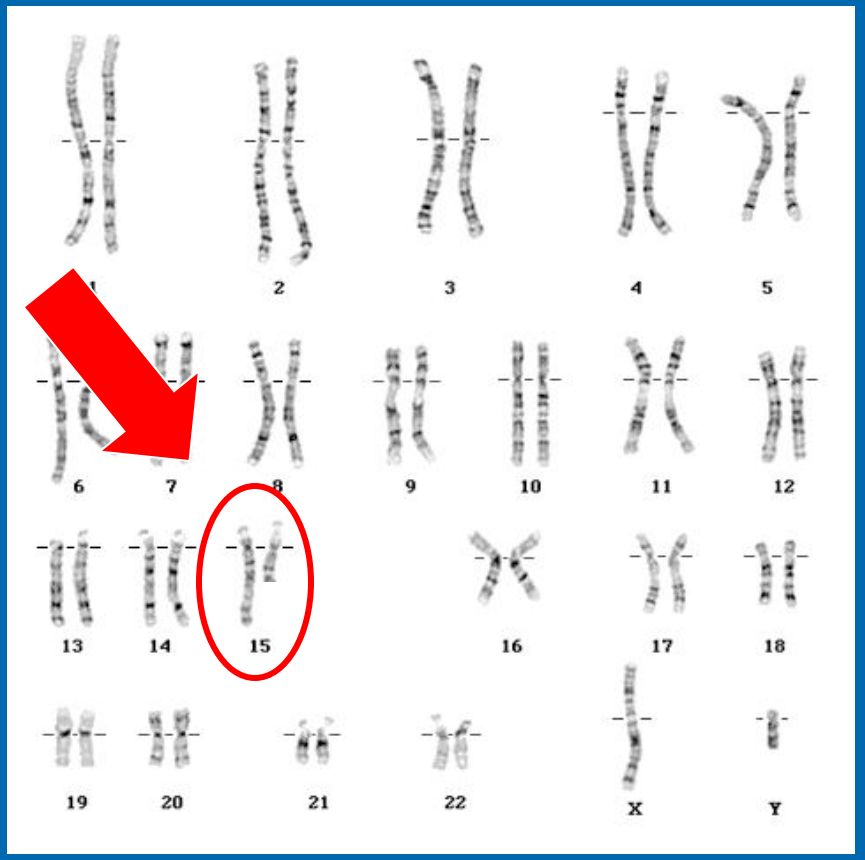
1 in 5,000,000 births

46 chromosomes
XY=97%
XX=3%

#15 Deletion of lower arm

Small bird-like head
Mentally retarded
Respiratory problems
Obesity
Short lifespan





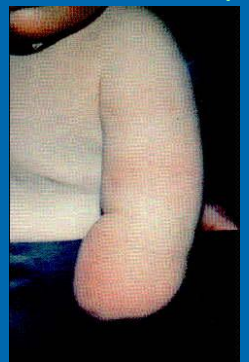
Eighteen Q Deletion Syndrome

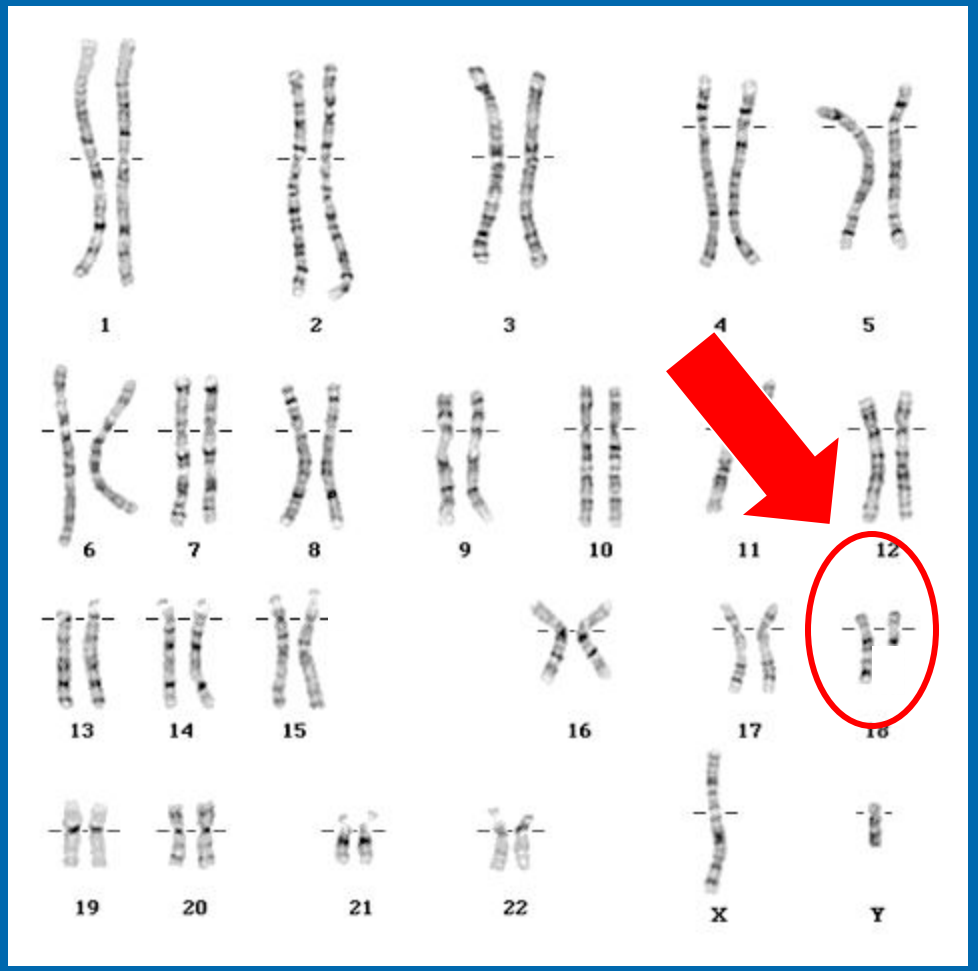
1 in 10,000,000 births

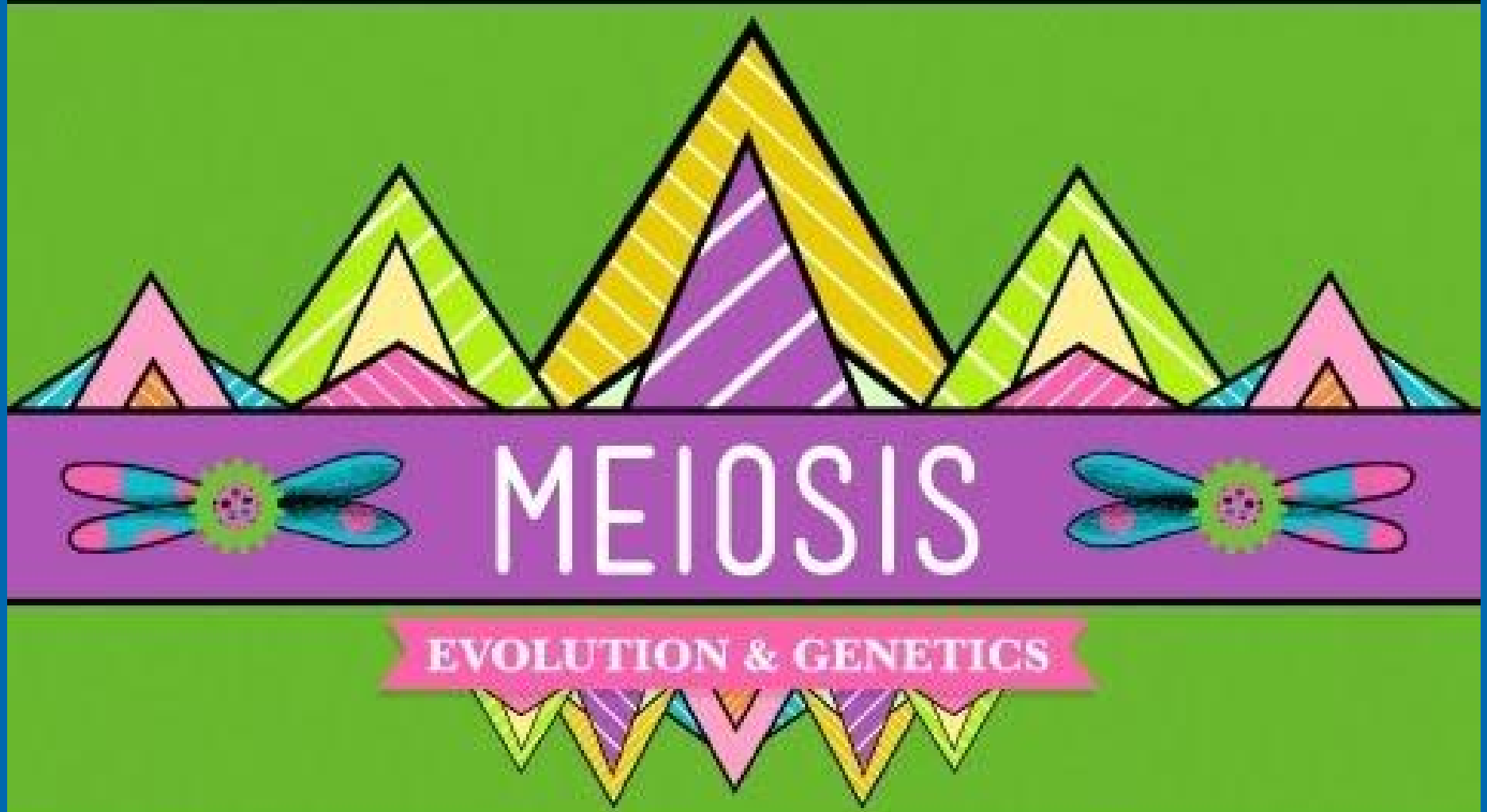
46 chromosomes
XY or XX

#18 Deletion of lower arm

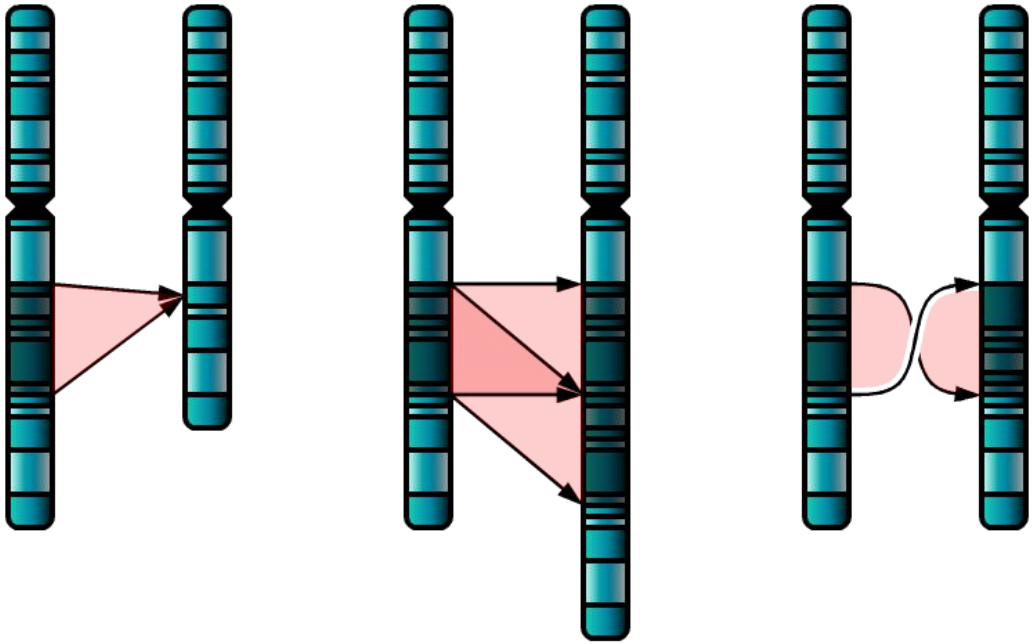
Mentally retarded
Heart disease
Abnormal hands and feet
Large eyes
Large ears
Normal lifespan






MEIOSIS
EVOLUTION & GENETICS

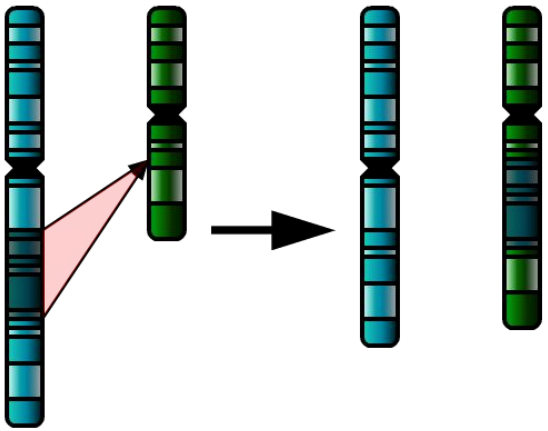
Single Chromosome Disorders



1. Deletion
   - Genetic material is missing
2. Duplication
   - Genetic material is present twice
3. Inversion
   - Genetic material is "flipped"

Two Chromosome Disorders

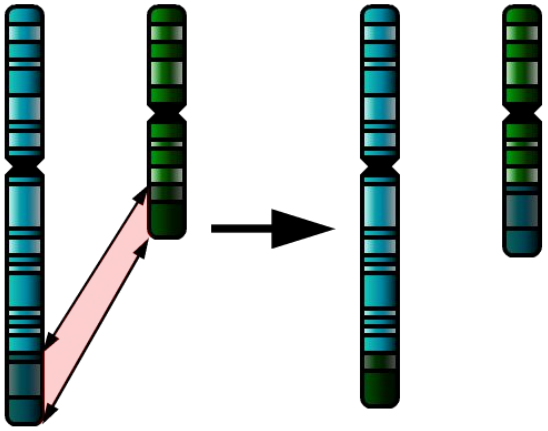
(Both types are called "translocation")



1

Insertion

- Genetic material is **added** from another chromosome

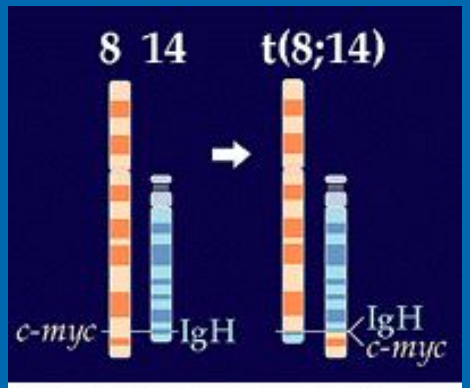


2

Translocation

- Material is **swapped** with another chromosome

Translocation: Burkitt Lymphoma



In Burkitt lymphoma, Myc, which is normally found on chromosome 8, is transferred to chromosome 14. This is known as chromosome translocation and can be characteristic of a cancer type. [image credit: Gregory Schuler, NCBI, NLM, NIH.]

Translocation of the *Myc* gene on chromosome 8

Normal Myc genes control cell growth and division

- Translocated *Myc* genes don't function properly
- Leads to cancer of the lymph nodes

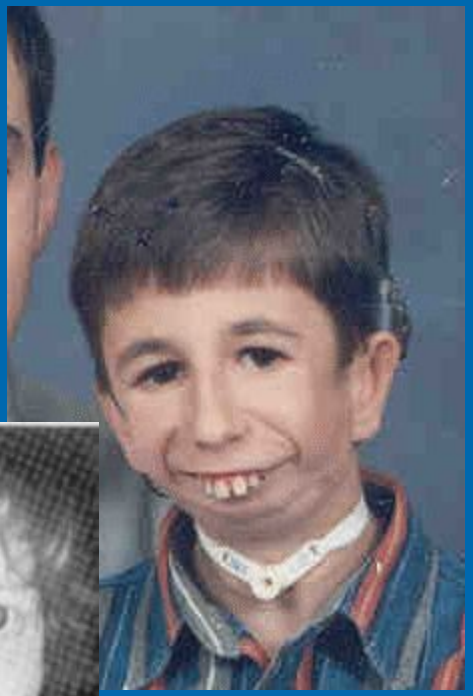
Cat-Eye Syndrome

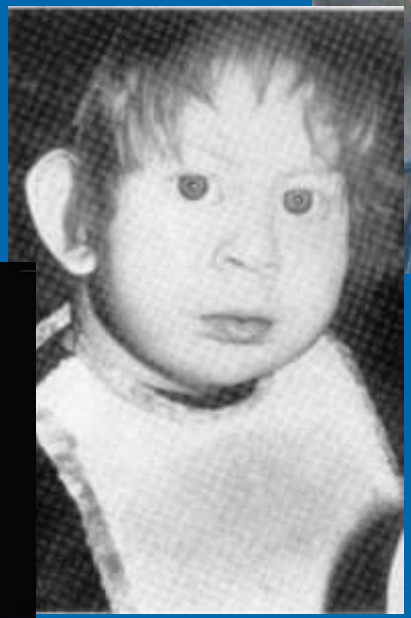
1 in 1,000,000 births

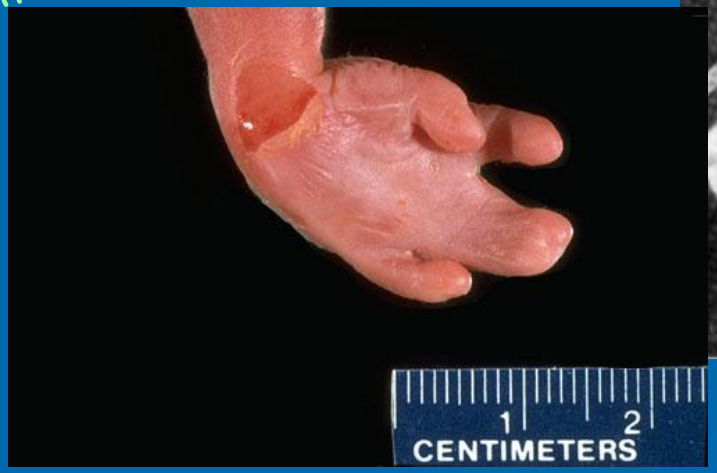
46 chromosomes
XY or XX

#22 Deletion of bottom arm

Fused fingers and toes
Mentally retarded
Small jaw
Heart problems
Normal lifespan





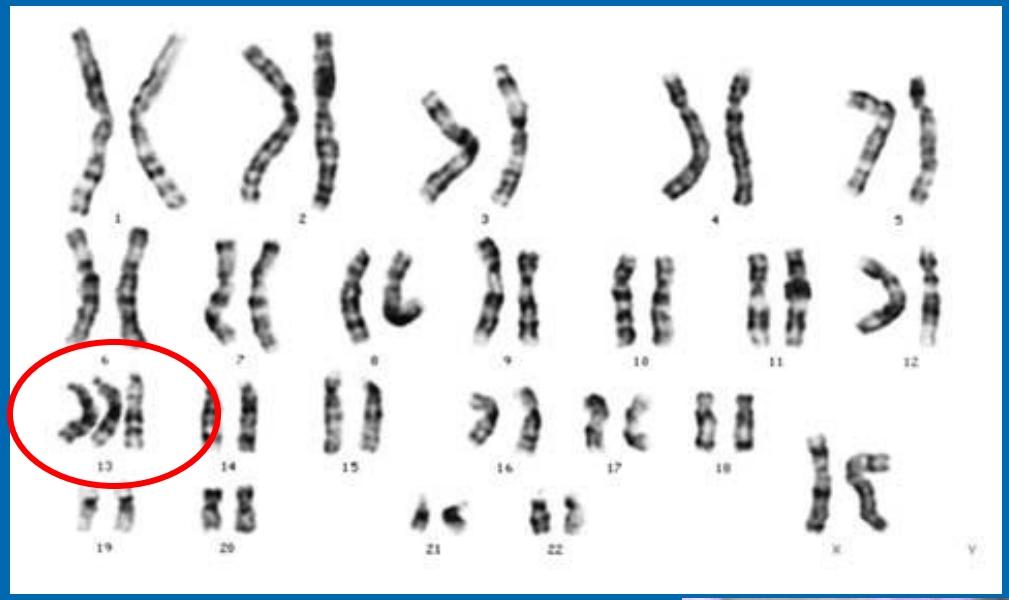


Patau's Trisomy Syndrome

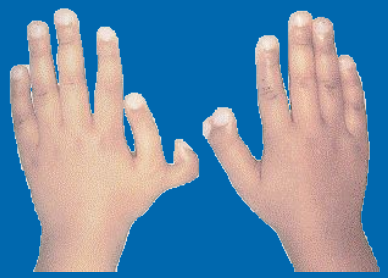
1 in 14,000 births

47 chromosomes
XY or XX

#13 Trisomy
Nondisjunction



Small head
Small or missing eyes
Heart defects
Extra fingers
Abnormal genitalia
Mentally retarded
Cleft palate
Most die a few weeks after birth



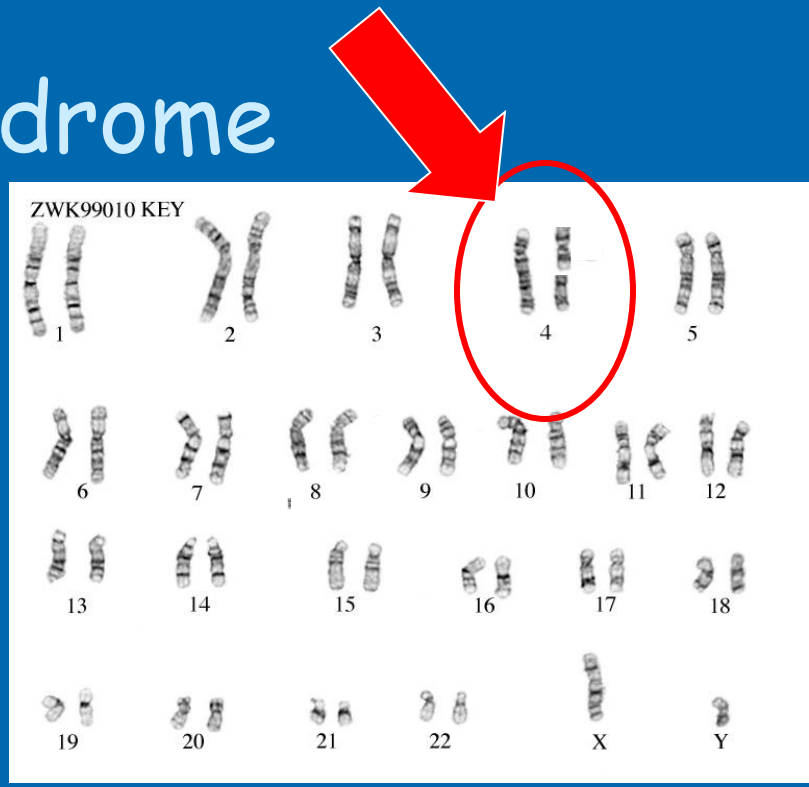
Four-Ring Syndrome

1 in 10,000,000 births

46 chromosomes
XY or XX

#4 Inversion

Cleft palate
Club feet
Testes don't descend
Short lifespan



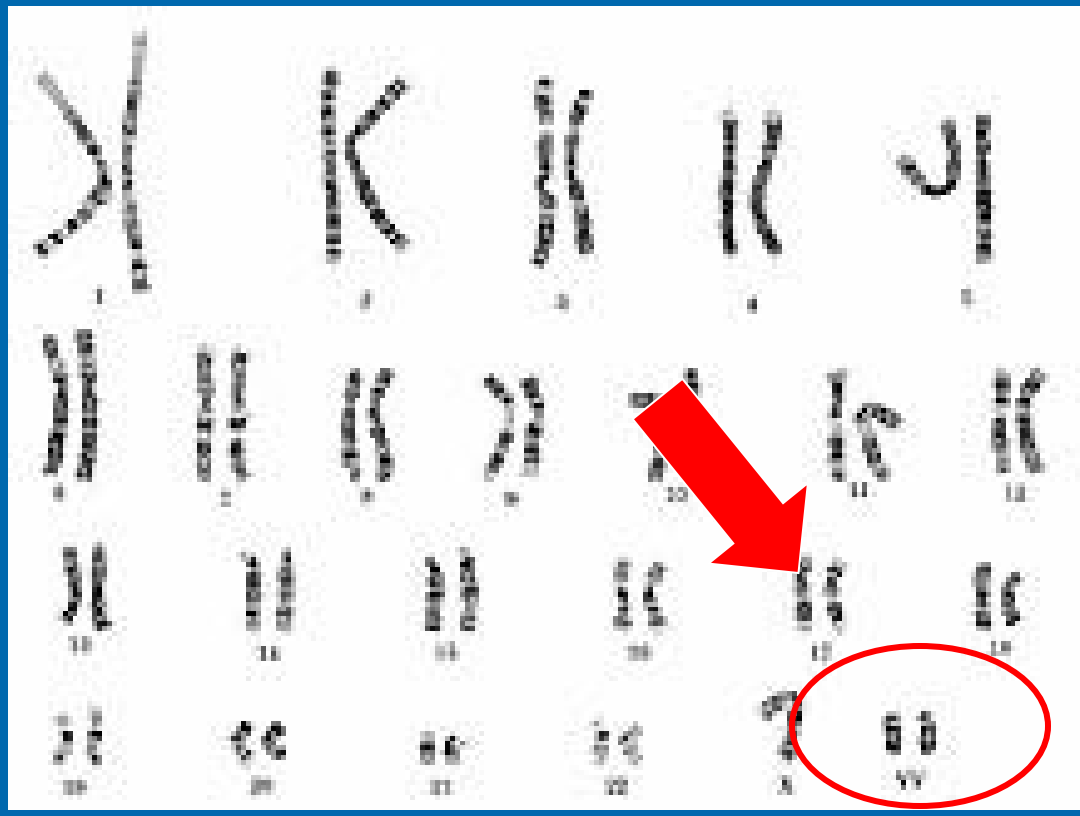
Jacob's Syndrome

1 in 1,800 births

47 chromosomes
XYY only

#23 Trisomy
Nondisjunction

?



Normal physically
Normal mentally
Increase in testosterone
More aggressive
Normal lifespan

Triple X Syndrome

Normal physically
- Sometimes taller

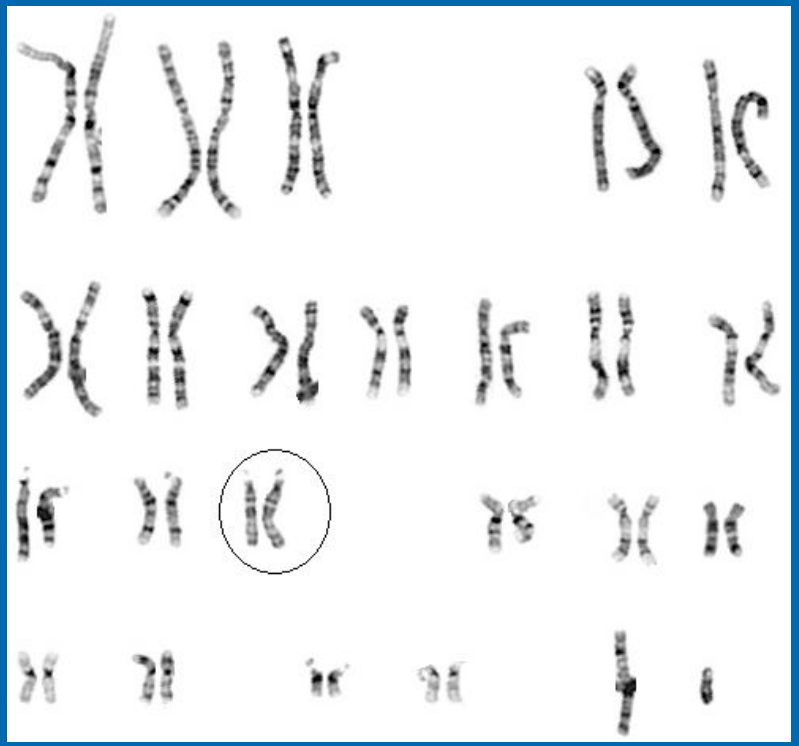
Normal mentally
- Inc. risk of retardation

Fertile

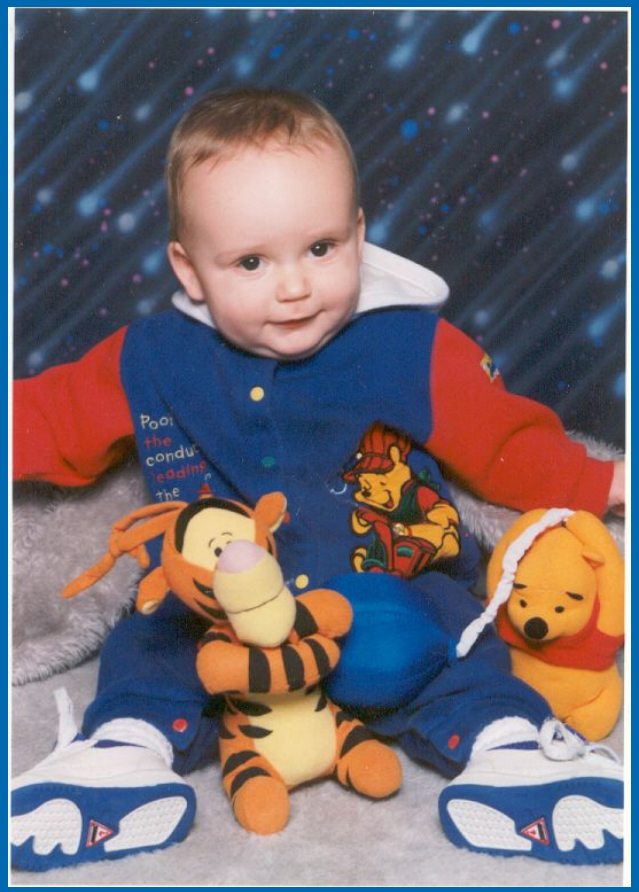
Other Types of Inherited Genetic Disorder…

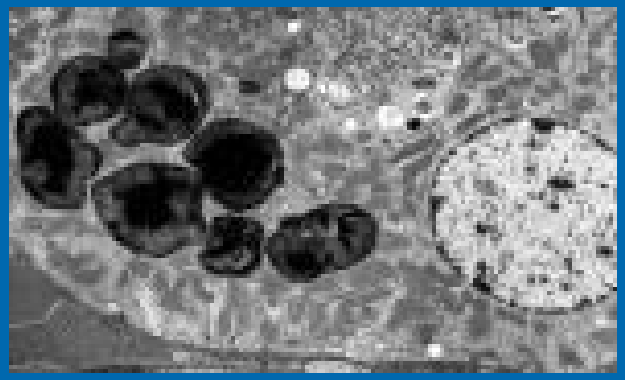
Tay-Sachs

Multiple kinds of mutation on Chromosome 15



- ***Tay-Sachs***
  - incidence rate of infantile Tay-Sachs=
    - Ashkenazi Jewish groups, French Canadian & Cajun: 1 in 30 are carriers
    - other groups: 1 in 300 are carriers





- accumulation of lipids on brain
- brain malfunction; death by age 5

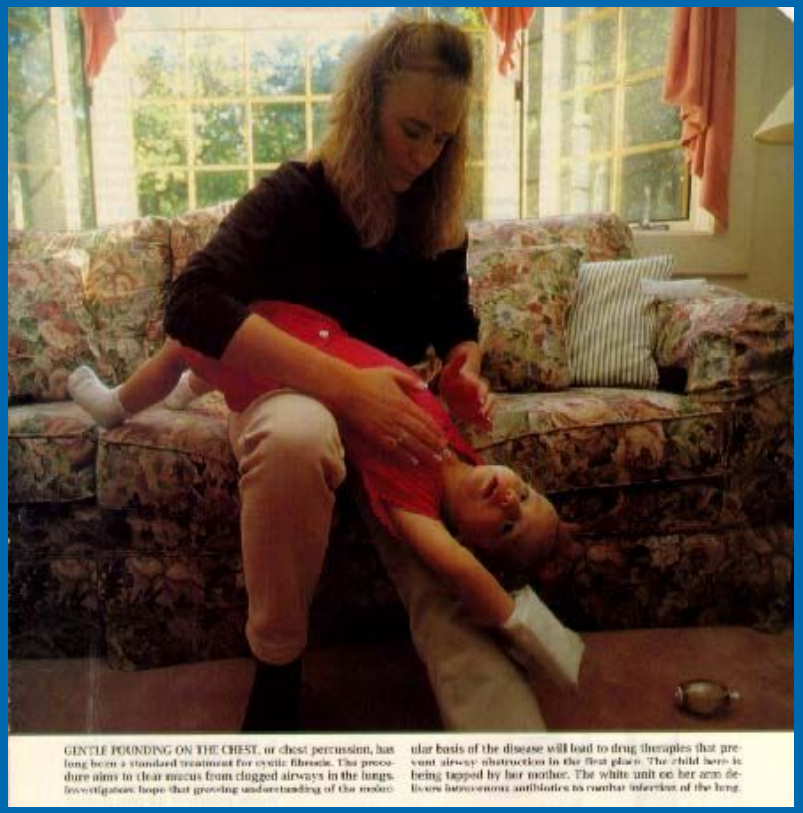
Sex-linked: X chromosome

Dystrophin and utrophin are a similar size and have comparable modular architecture. This similarity means that utrophin can sometimes substitute for dystrophin, so providing a potential route for therapy for muscular dystrophy sufferers.

- ***Duchenne Muscular Dystrophy***
  - 1 boy in every 3,000
  - progressive weakening of muscles and loss of coordination
  - more common in males

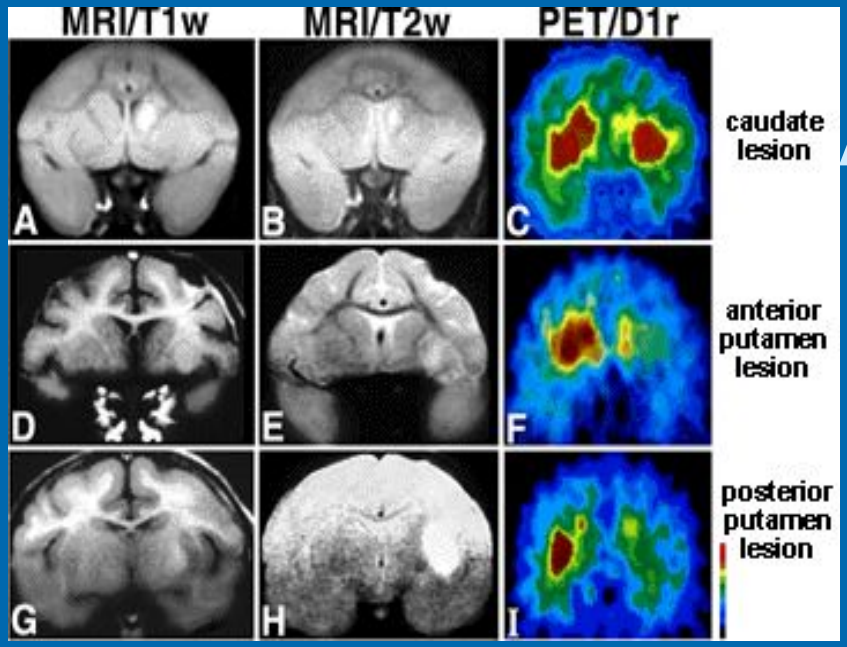
Cystic Fibrosis

- 1 in 31 is carrier in U.S.
- Single gene mutation
- mucus in lungs, pancreas, digestive tract and other organs
- *most common* lethal genetic disease in U.S.



GENTLE POUNDING ON THE CHEST, or chest percussion, has long been a standard treatment for cystic fibrosis. The procedure aims to clear mucus from clogged airways in the lungs. Investigators hope that growing understanding of the molecular basis of the disease will lead to drug therapies that prevent airway obstruction in the first place. The child here is being tapped by her mother. The white unit on her arm delivers intravenous antibiotics to combat infection of the lung.

- heterozygote may be resistant to typhoid fever



Autosomal Dominant

- *Huntington's*
  - incidence: 1/10,000 in U.S.
  - nervous system degenerates
  - late onset
  - may not show symptoms until past reproductive age

Karyotype Lab “Quiz” Tomorrow

- Karyotyping for a disease
- Partners
- 4 diseases represented, pick 2 karyotypes, cut and match. Once matched up, answer the questions about your two karyotypes.