Chromosomes and Karyotypes
Two Types of Chromosomes:

1. Autosomes
   - ALL chromosomes except the sex chromosomes
   - 22 pairs (Chromosomes #1-22)
Two Types of Chromosomes:

2. Sex Chromosomes:
   - 1 pair (human chromosome #23)
   - Determine the sex of an organism
     - In mammals & fruit flies XX is female, XY is male
     - In birds ZZ is male, ZW female
   - *FIX IN NOTE TEMPLATE
The only genetic difference between (human) males and females is this:

Females have two \( X \) chromosomes:

\[ \text{\underline{\underline{\underline{\underline{\text{X}}}}} \text{\underline{\underline{\underline{\underline{\text{X}}}}}}} \]

While males have one \( X \) and one \( Y \):

\[ \text{\underline{\underline{\underline{\underline{\text{X}}}}} \text{\underline{\underline{\underline{\underline{\text{Y}}}}}}} \]

The other 22 other pairs of chromosomes are the same.
Karyotypes

- A picture of the chromosomes in which the chromosomes arranged in matching (homologous) pairs
Karyotypes

- Arranged in size order from largest pair to smallest pair
- The sex chromosomes (X and Y) are usually the last pair, though they are not the smallest.
Normal Male
Karyotypes

- How are they used?
  - They are used for diagnosis of genetic abnormality based on the number of chromosomes.
  - They are used to determine the sex of an unborn child.
Karyotypes

- How are they prepared?
  - Cells are collected from a variety of sources:
    - Amniotic fluid via a pre-natal "amniocentesis"
    - Blood Sample
Karyotypes

- How are they prepared?
  - Sample of cells are allowed to continue dividing
  - Cells are stopped when in METAPHASE of MITOSIS.
Karyotypes

- A photograph of the chromosomes is taken and enlarged.
- A trained technician matches the chromosomes into the homologous pairs based on three characteristics:
  - Size
  - Banding
  - Centromere position
Chromosomal Disorders

- **Normal:**
  - Have 2 matching chromosomes for each of the 23 pairs

- **Aneuploidy:**
  - Having one more or one less of one of the chromosomes of the 23 pairs.
Chromosomal Disorders

- **Monosomy:** Missing one chromosome of one of the pairs
  - Turner’s syndrome; Monosomy 23
    - Missing one of the X chromosomes
    - Female who is X0 instead of XX
Chromosomal Disorders

Symptoms:

- Short stature
- Webbed neck
- Lack of secondary sex characteristics
- A hollow appearance to the chest
- Lack of menstruation
- Low hairline
- "Droopy" eyelids
Chromosomal Disorders

- **Trisomy**: An extra chromosome of one of the pairs
  - Down syndrome; Trisomy 21
  - Extra chromosome #21 (so, there are 3 chromosome #21)
Chromosomal Disorders

- Incidence
  - One of the most common chromosomal abnormalities
  - Frequency varies a lot according to the age of the mother.
    - The rate is only 1 in 2,000 for women 20 years old
    - In those 40 or older, it is 1 birth in 100.
Genetic Disorders

Symptoms:
- Small head, flattened in the back
- Broad, flat face
- Relatively small eyes, turned up at the outer corners
- Oversize tongue in a small mouth
- Single horizontal line across the palm, instead of the usual "head" and "heart" lines
- Short stature, with short limbs and stubby fingers
Genetic Disorders

- Kleinfelter’s Syndrome; Trisomy 23
  - Extra sex chromosome
  - Male who is XXY instead of XY
  - The most common sex chromosome abnormality in males
Genetic Disorders

Symptoms:

- Arm span exceeds height by more than an inch.
- No or very little body hair and no facial hair.
- High voice
- Minimal muscle growth in arms/legs
- Small testicles
- Breast Tissue (not just fat, but actually firm breasts)
- Low Testosterone Level