Karyotype

• Typical human body (somatic) cell has 46 chromosomes
• A display of the all 46 chromosomes is called a karyotype
Karyotype – Homologous Chromosomes

- Each person has 2 twin chromosomes called homologous chromosomes
  - Homologous chromosomes are two chromosomes of the same type
    - They are the same size, shape, and have the same genes
  - Where do you think each came from?
    - One chromosome from Mom
    - One chromosome from Dad
Homologous Chromosomes

- Each homologous chromosome carries the same sequence of genes controlling a characteristic, but different versions
  - Example:
    - Eye color is located on the same place, but may be different versions (blue, brown, hazel)
  - Different from sister chromatids because the material is genetically different

Figure B-11: Homologous Chromosomes

Homologous chromosomes contain DNA that codes for the same genes. In this example, both chromosomes have all the same genes in the same locations (represented with colored strips), but different ‘versions’ of those genes (represented by the different shades of each color).

Homologous Chromosomes Consist of 4 sister chromatids
Chromosomes

- Humans have 23 homologous pairs of chromosomes
- **Human body cells (diploid)** have 22 pairs of autosomes and 1 pair of sex cells (23 pairs or 46 chromosomes)
  - The 23rd pair are called **sex chromosomes**
- **Human sex cells**, gametes, (haploid) have **22 autosomes** and **1 sex cell**.

![Karyotype](image)

Autosomes are inside the red box and labeled with numbers. The sex chromosomes are outside of the red box and given letters X and Y. This karyotype is from a male because males have one X and one Y while females have two X chromosomes.
Autosome vs. Sex Chromosome

- Autosomes are chromosomes that are NOT a sex-determining chromosome
- Most chromosomes are autosomes
  - You receive:
    - 22 autosomes and 1 sex chromosome from Mom
    - 22 autosomes and 1 sex chromosome from Dad
- Sex chromosomes are chromosomes needed for determining the sex of the individual

Sex Chromosomes

- All homologous chromosomes look the same except for the sex chromosomes
  - Males have XY
  - Females have XX
Genetic Disorders – Abnormal Chromosome Numbers

- Chromosomal abnormalities often result from Nondisjunction.
  - Nondisjunction is the failure of chromosomes to separate properly during meiosis
  - Can result in:
    - Cells that have too many chromosomes or too few chromosomes
      - Trisomy & Monosomy

<table>
<thead>
<tr>
<th>Name of Abnormality</th>
<th>Chromosome Affected</th>
<th>Description of Abnormality</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down Syndrome, or Trisomy 21</td>
<td>#21</td>
<td>47 chromosomes; mental retardation with specific characteristic features; may have heart defects and respiratory problems</td>
</tr>
<tr>
<td>Edwards’ Syndrome, or Trisomy 18</td>
<td>#18</td>
<td>47 chromosomes; severe mental retardation, very characteristic malformations of the skull, pelvis, and feet, among others; may die early infancy</td>
</tr>
<tr>
<td>Patau Syndrome, or Trisomy 13</td>
<td>#13</td>
<td>47 chromosomes; abnormal brain function that is very severe, many facial malformation; usually die in early infancy</td>
</tr>
<tr>
<td>Turner’s Syndrome</td>
<td>Single X in Female (XO)</td>
<td>45 chromosomes; in females only; missing an X chromosome; do not develop secondary sex characteristics; are infertile</td>
</tr>
<tr>
<td>Klinefelter’s Syndrome</td>
<td>Extra X in Male (XXY)</td>
<td>47 chromosomes; in males only; sterile, small testicles; otherwise normal appearance</td>
</tr>
<tr>
<td>XYY Syndrome</td>
<td>Extra Y in Male (XYY)</td>
<td>47 chromosomes; in males only, low mental ability, otherwise normal appearance</td>
</tr>
<tr>
<td>Triple X Syndrome</td>
<td>Extra X in Female (XXX)</td>
<td>47 chromosomes; sterility sometimes occurs, normal mental ability</td>
</tr>
</tbody>
</table>

**Genetic Disorders Caused by an Abnormal Chromosome Number**

- **Trisomy** is an abnormality in which a cell has an extra chromosome or section of a chromosome. The cell contains 47 chromosomes instead of 46.
- **Monosomy** is an abnormality in which a cell is missing a chromosome. The cell will contain 45 chromosomes instead of 46.
Trisomy 21 – Down’s Syndrome
If you look at the space where chromosome 21 sits, you notice that there are 3 (tri) chromosomes instead of 2 (a pair). Is this person male or female? How do you know?

Monosomy X – Turner Syndrome
Notice that there is only 1 C chromosome. Which means that there are 45 chromosomes and not 46. Male or female?