A picture of the chromosomes from a human cell arranged in pairs by size

- First 22 pairs are called autosomes
- Last pair are the sex chromosomes
- XX female or XY male
Karyotype Procedure

- 5 ml of blood is removed from the patient.
- If a fetus is being karyotyped amniotic fluid is removed from the amniotic sac which surrounds the fetus during development. This is done with the aid of a large syringe and ultrasound picturing. There are cells which have come off the fetus in this fluid.
- The white blood cells are removed from the blood or the living cells are removed from the amniotic fluid.
- These cells are then cultured in a medium in which they undergo mitosis. Mitosis is stopped at metaphase using chemicals.
- The cells are then placed onto a slide and spread out.
- They are viewed under a microscope which is specially adapted with a camera to take a picture of the chromosomes from one of the cells.
- Once the picture is taken and enlarged the chromosomes are cut out and arranged in pairs according to size and location of the centromere.
Karyotyping is the process by which doctors and geneticists take pictures of the chromosomes while the cell are undergoing mitosis.

The picture is then enlarged.

The picture of the chromosomes are then cut up so that each chromosome is removed. The chromosomes are matched up and attached to a paper according to size, banding patterns, & centromere position.

The chromosomes pairs are numbered from largest to smallest.

There are 22 pairs of chromosomes that are aligned first & which match up exactly. These are called autosomes & will code for human body characteristics.

Then the sex chromosomes are paired, in the female (XX) the chromosomes match and in the male (XY) the chromosomes do not match.
Boy or Girl?
The Y Chromosome Decides
Average Male

Average Female
Mutations
Mutations

- Changes in the genetic code
- Failure of DNA repair
- During fertilization these cause birth defects (genetic disorders) that can’t be cured
- During mitosis these cause cancer
Types of Mutations

- Mutations
  - Gene Mutations
  - Chromosominal
**Gene Mutations**

- Insertions or deletions (frameshift mutations), or changes of a single base (point mutations)
- This causes codon errors and affects amino acid translation

<table>
<thead>
<tr>
<th>The cow jumped over the moon.</th>
</tr>
</thead>
<tbody>
<tr>
<td>With a deletion of the &quot;w&quot; now becomes</td>
</tr>
<tr>
<td>The cojumpedover the moon.</td>
</tr>
</tbody>
</table>
Point Mutations vs. Frameshift

**Changing a base**

**original:**

\[
\text{AUG CAU GGC}
\]

**changed:**

\[
\text{AUG C\underline{CU} GGC}
\]

**Deleting or Inserting a base**

**original:**

\[
\text{AUG CAU GGC}
\]

**changed:**

\[
\text{AUG C\underline{UG} GGC}
\]

The codons have shifted!
Chromosomal Mutations

- Can cause death of the zygote / fetus
- Can cause sterility
- Most cause distinct abnormalities – many are very severe
- Affect physical & mental health
Types of chromosomal mutations

1. Duplications
2. Translocations
3. Deletions
4. Inversions
5. Changes in the numbers of chromosomes
Duplications

- Involves a chromosome that has a piece repeated
- Causes extra length (info) in the strand.
Translocation

- Transferring a piece of one chromosome to another chromosome
Deletions

- Omitting or losing a piece of a chromosome

Prader – willi syndrome
Inversion

- Attaching a piece of a chromosome backward
Changes in chromosome number

- Having more than two copies of each chromosome
- Can have from 3N to 5N of a chromosome including the sex chromosomes
Trisomy

- Having 3 copies of the chromosome instead of the pair (3N)
- Examples:
  1. Down’s Syndrome – trisomy 21
  2. Klinefelter’s - XXY
Edward’s Syndrome
Monosomy

- Having 1 copy of the chromosome instead of the pair
- Example:
  1. Turner Syndrome
Polysomy

- Having more than 3 copies of a chromosome.
- Example: not assigned a name but normally found only in sex chromosomes
  XXXXY
  XYYYY
### Disorders caused by a point mutation

<table>
<thead>
<tr>
<th>Fragile X Syndrome</th>
<th>Huntington’s Disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>The most common mental retardation disease besides Down Syndrome</td>
<td>A disease that shows symptoms late in life that is highly heritable</td>
</tr>
<tr>
<td></td>
<td>Degenerative nerve disease</td>
</tr>
<tr>
<td></td>
<td>Eventual death</td>
</tr>
</tbody>
</table>
Mutagens

- Chemicals or agents that cause copying errors during cell division

1. Exposure to radiation
2. Chemicals used in war
3. Chemicals in food preservatives
4. Viruses
Exposure to radiation can cause a multitude of chromosomal mutations.
Human Genetic Disorders

1. Turner’s Syndrome
2. Klinefelter’s Syndrome
3. Microcephaly
4. Marfan’s Syndrome
5. Prader-Willi Syndrome
6. Edward’s Syndrome
7. Epidermolysis Bullosa
8. Congenital Generalized Hypertrichosis
9. Cri du Chat
10. Achondroplasia
11. Gaucher’s Disease
12. Duchenne Muscular Dystrophy
13. Fragile X Syndrome
14. Neurofibromatosis
15. Huntington’s
16. Cystic Fibrosis
17. Cleft Palate
18. Polydactyly
19. Colorblindness
20. Hemophilia
21. Ichthyosis
22. Spina Bifida
23. Jacob’s Syndrome
24. Amyloidosis
25. Down’s Syndrome
26. Gastroschisis
27. Phenylketonuria (PKU)
28. Albinism
29. Tay-Sachs
30. Sickle Cell Anemia
31. Progeria
32. Cystic Fibrosis
33. Cleft Palate
34. Polydactyly
35. Colorblindness
36. Hemophilia
37. Ichthyosis
38. Spina Bifida
39. Jacob’s Syndrome
40. Amyloidosis
41. Down’s Syndrome
42. Gastroschisis
Albinism
Cleft Palate

Achondroplasia

Phenylketonuria Testing & diagnosed child
Dwarfism
- Bone elongation in dwarfs.
- Very painful procedure
Excess or deficits can result in obvious skeletal malproportions.

Twelve-year-old boy with pituitary gigantism measuring 6'5" with his mother. Not the coarse facial features and prominent jaw.

Picture 1. Gigantism and acromegaly. The author with a statue of Robert Wadlow, the "Alton Giant," who was the tallest person ever recorded. He measured 8 feet 11 inches at the time of his death.
Progeria

Sickle cell

Microcephaly

CGH

Duchenne Muscular Dystrophy

Polydactyly
Epidermolysis Bullosa
Prader Willi Syndrome
“The Cry of the Cat”