Human Karyotypes

- Birth defects (2-3% human pregnancies)
- Genetic defects
- Chromosomal abnormalities
- Amniocentesis: embryonic (fetal) cells

Lab # 5

- Karyotyping
- Dermatoglyphics

Karyotype Preparation

- Cells (from blood, amniotic fluid, etc) are grown in vitro to increase their number
- Cell division is then arrested in metaphase
- Cells are centrifuged and lysed to release chromosomes
- Chromosomes are stained, photographed, and grouped by size and banding patterns
Identifying Features of Chromosome

- Chromosome size
- Position of centromere
- Banding pattern

Human Karyotype (Male)

Normal Female Karyotype
Chromosomal Abnormalities

- More than 80 types
- Can determine sex of unborn+
- Blood, hair, other tissues
- Embryonic and fetal tissues
- Frequency: 1 in 160 newborn infants

Chromosomal Abnormalities

- Normal chromosomal number: 46
- Irregular Number
  - Polyploidy (e.g., 23 + 23 + 23)
  - Aneuploidy (23 + 22)
- Structural Modification
- Missing pieces

Translocation
Duplication

Cri du chat

Down Syndrome

- Langdon Down (1828-1896)
### Sex Chromosome Abnormalities

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Gender</th>
<th>Syndrome (if any)</th>
<th>Physical Traits</th>
</tr>
</thead>
<tbody>
<tr>
<td>XXY, XXYY, XXXY</td>
<td>male</td>
<td>Klinefelter syndrome</td>
<td>Sterility, small testicles, breast enlargement</td>
</tr>
<tr>
<td>XYY</td>
<td>male</td>
<td>No syndrome</td>
<td>Normal male traits</td>
</tr>
<tr>
<td>XO</td>
<td>female</td>
<td>Turner syndrome</td>
<td>Sex organs don't mature at adolescence, sterility, short stature</td>
</tr>
<tr>
<td>XXX</td>
<td>female</td>
<td>Metafemale</td>
<td>Mental retardation, limited fertility</td>
</tr>
</tbody>
</table>

### Female Sex Chromosome Abnormalities

- **Turner Syndrome (XO)**
  - ![Image of a girl with Turner Syndrome]
  - Sex organs don't mature at adolescence, sterility, short stature

- **XXX (Metafemales)**
  - ![Image of a girl with XXX (Metafemales)]
  - Mental retardation, limited fertility
Male Sex Chromosome Abnormalities

- XXY (Klinefelter Syndrome)

Male Sex Chromosome Abnormalities

- XYY
- Richard Speck

Dermatoglyphics

- derma = skin
- glyphe = carve
- friction ridge formations: palms, soles, fingers, toes
Dermatoglyphics

- 3rd and 4th month of fetal development
- forensic significance
- 2000 year old Egyptian mummy
- primates, arboreal marsupials

Classification

- Arches
- Loops
- Whorls

Triradius

- Target Wheel
- Convex Circle
- 4. Loop
- 3. Triradius
Arch

Loop
- 1 triradius (delta)
- Radial loop
- Ulnar loop
- No triradius

Whorl
- 2 triradii
British Frequencies

- loops -70%
- whorls- 25%
- arches-5 %

- whorls more common in the first and fourth digits (35%); radial loops more common on the right than left hand

Pattern Intensity Index

- Count number of triradaii on 10 digits (0 to 20)

Pattern Size

- Count no. of ridges on 10 digits (arches= 0)
Anthropology

- Bushmen: arches common
- Chinese: whorls frequent
Inheritance

- polygenic or multifactorial inheritance

Medical Genetics

- Down's Syndrome
- Turner's Syndrome