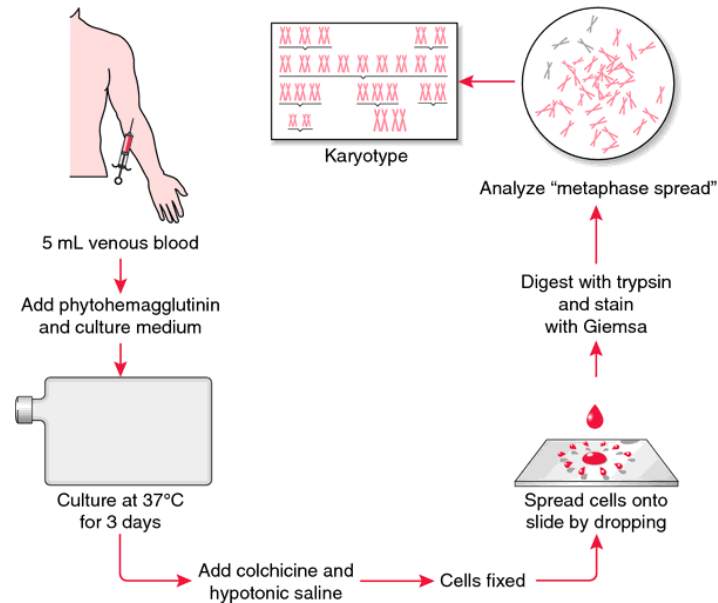


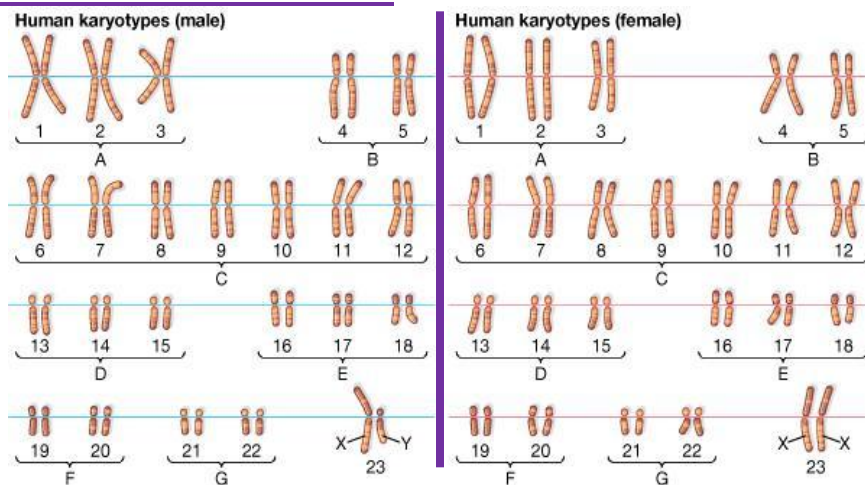


**Karyotyping:**

- **Definition:** a laboratory technique used to analyze chromosomes in order to look for any major chromosomal anomalies which may cause a genetic condition. It is the complete set of metaphase chromosomes in a cell.
- **When to do karyotype?**
  - ✓ Indeterminate gender (male or female?).
  - ✓ Dysmorphisms.
  - ✓ Developmental delay or mental retardation.
  - ✓ Delayed puberty.
  - ✓ Infertility or repeated spontaneous abortions.
- **Process of karyotyping (notice that the sample is mainly taken from blood lymphocytes):**

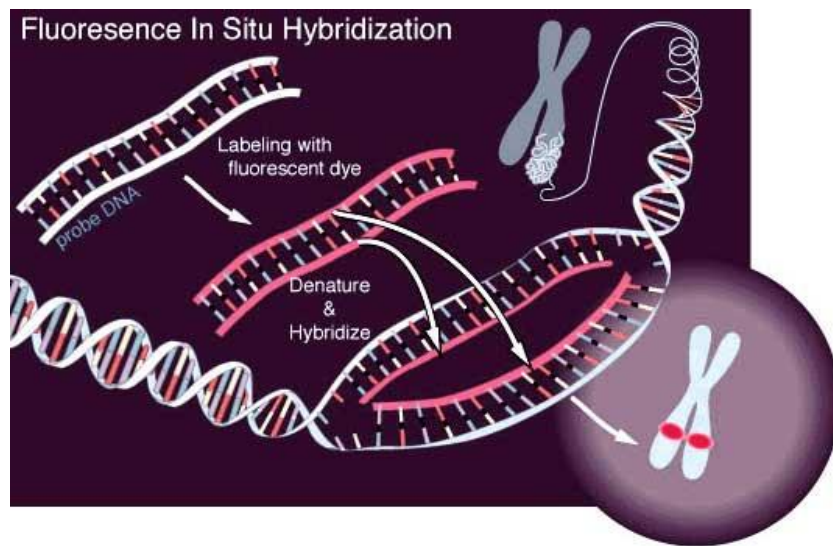


**Classification of human chromosomes:**



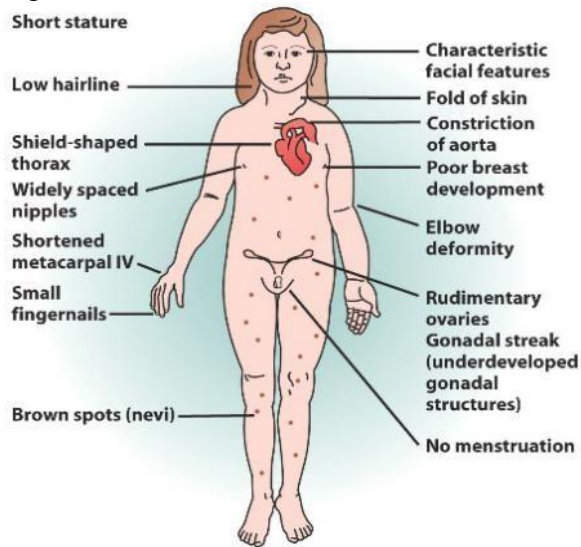
**Chromosomal staining:**

<b>G-banding</b>	Staining metaphase chromosome with Giemsa to show AT-rich regions.
<b>Q-banding</b>	Fluorescent dye showing AT-rich regions.
<b>C-banding</b>	Treated with acid and base then stained with giemsa
<b>R-banding</b>	Reverse banding
<b>FISH (Fluorescence In Situ Hybridization)</b>	Use of fluorescent tags to label cellular DNA thus visualizing the chromosome. The use of multicolor FISH probes can determine the number of chromosome copies in interphase nuclei.



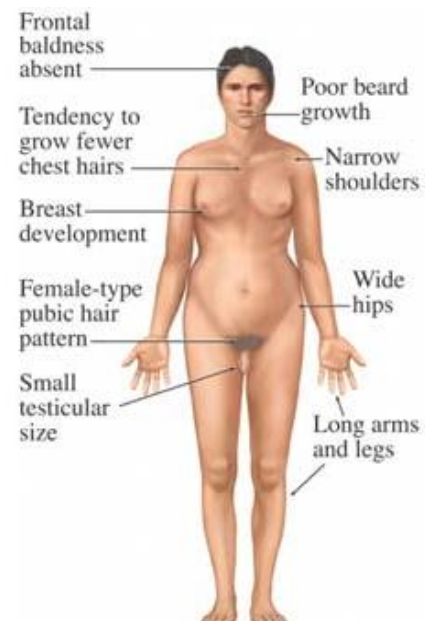
- **Turner syndrome:**

- **Monosomy X (only one X chromosome is present).**
- **Clinical features:**
  - ✓ Short stature.
  - ✓ Webbed neck.
  - ✓ Shield chest with widely spaced nipples.
  - ✓ Ovarian dysgenesis which causes delayed puberty and infertility.
  - ✓ Cardiac defects (e.g. coarctation of the aorta).



- **Klinefelter syndrome (XXY):**

- **It is the most common cause of male hypogonadism and infertility.**
- **Clinical features:**
  - ✓ Tall stature with long extremities.
  - ✓ Hypogonadism.
  - ✓ Gynecomastia.
  - ✓ Shyness or aggression.

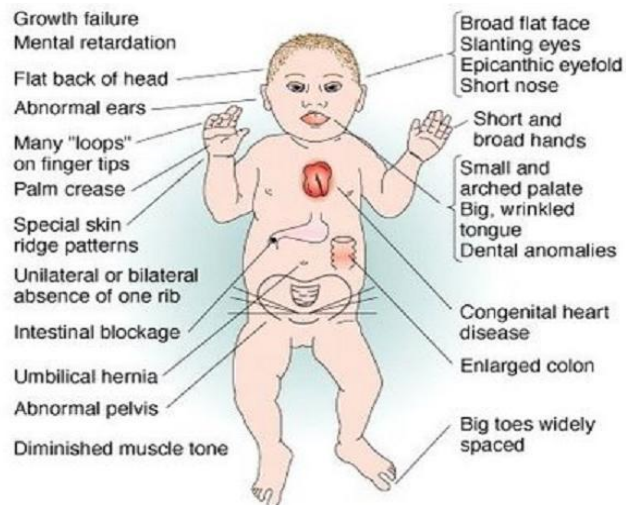




- **Down syndrome:**

- **Trisomy 21 in which the risk is increase with advanced maternal age.**

Clinical features	Complications
<b>Craniofacial features:</b> <ul style="list-style-type: none"> <li>• Brachycephaly.</li> <li>• Epicanthal skin folds.</li> <li>• Upslanting palpebral fissures.</li> <li>• Brushfield spots.</li> <li>• Protruding tongue.</li> </ul>	<b>Leukemia</b>
<b>Hypotonia.</b>	<b>Early Alzheimer's disease</b>
<b>Mental retardation.</b>	<b>Obstructive sleep apnea</b>
<b>Musculoskeletal features:</b> <ul style="list-style-type: none"> <li>• Clindodactyly.</li> <li>• Single palmar crease.</li> <li>• Wide space between first and second toes.</li> </ul>	<b>Hypothyroidism</b>
<b>Gastrointestinal features:</b> <ul style="list-style-type: none"> <li>• Duodenal atresia.</li> <li>• Hirschsprung's disease and omphalocele</li> <li>• Pyloric stenosis.</li> </ul>	<b>Cataracts</b>
<b>Cardiac features (40%):</b> <ul style="list-style-type: none"> <li>• Endocardial cushion defects (most common)</li> </ul>	<b>Glucoma</b>



- **Edward's syndrome:**

- **Trisomy 18 which is more commonly seen in females.**
- **95% of cases die within first year of life** ☹️
- **Clinical features:**
  - ✓ Mental retardation.
  - ✓ Hypertonia.
  - ✓ Small facial features.
  - ✓ Clenched hands with overlapping digits.
  - ✓ Rocker-bottom feet.

