

Introduction to Medical Genetics: Human karyotype

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At the end of this resource, you should be able to

- Describe karyotype
- Explain how karyotype can be done
- Describe chromosome classification



Topics

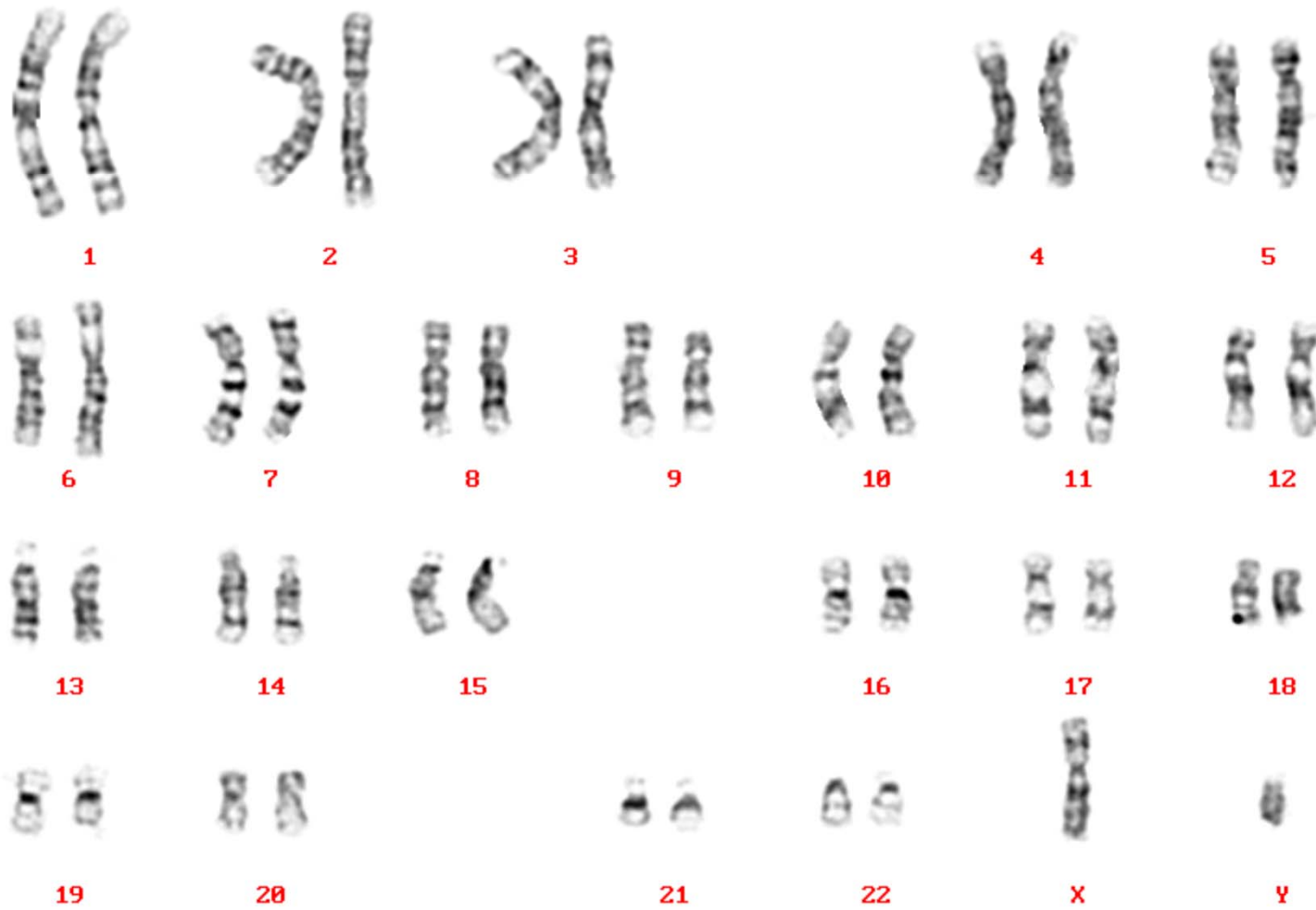
- What is human karyotype?
- Chromosome banding
- Chromosome classification

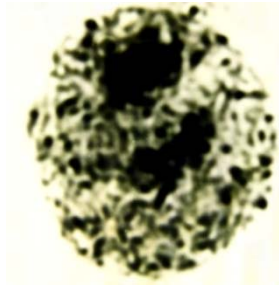


Human Karyotype

- Full set of **paired homologous chromosome (autosomes)** and **sex chromosome**
- From a mitotic cell arranged according to **descending order in size**
- Human have:
 - **22 pairs of autosomes + 1 pair of sex chromosome**
- 23 pairs or 46 chromosomes
- Human **Male** Karyotype – 46, XY
- Human **Female** Karyotype – 46, XX







Dividing cell



Prophase



Metaphase spread



Metaphase chromosome

Chromosome banding

- Chromosomes are tiny and requires contrast to visualize
- Metaphase chromosome are suitable for visualization
- **Colchicine** is used to accumulate metaphase chromosome
- Staining is carried out using Giemsa or Quinacrine mustard



Chromosome banding

- Staining is enables chromosome identification and structural analysis
- Based on staining chemical chromosome banding can be G, Q or R banding
- Giemsa staining is widely used, produces dark and light **G bands**
- Giemsa staining can be visualised using light microscope
- Quinacrine mustard staining produce bright and dim **Q bands**
- Quinacrine mustard staining requires fluorescence microscope.
- **R bands** are the reverse of G bands



Chromosome classification

- Based on **centromere position** human chromosome can be classified into 3 types
- **Metacentric** – centromere in or near the center ($p = q$)
- **Submetacentric** – centromere off the center ($p \neq q$)
- **Acrocentric** – centromere near one end (substantially small with satellite attached by stalk) Human chromosome 13, 14, 15 and 21
- **Telocentric** – not found in human, centromere at one end (only one arm)



Summary

- **Human Male Karyotype** - full set of paired homologous chromosome and sex chromosome (46, XY)
- **Chromosome banding** -based on staining procedure chromosome banding can be G, Q or R banding
- **Chromosome classification** - based on centromere position human chromosome can be classified into 3 types, metacentric, submetacentric and acrocentric

