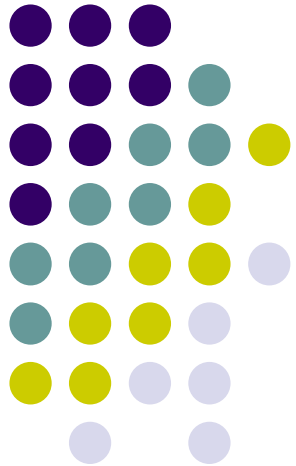
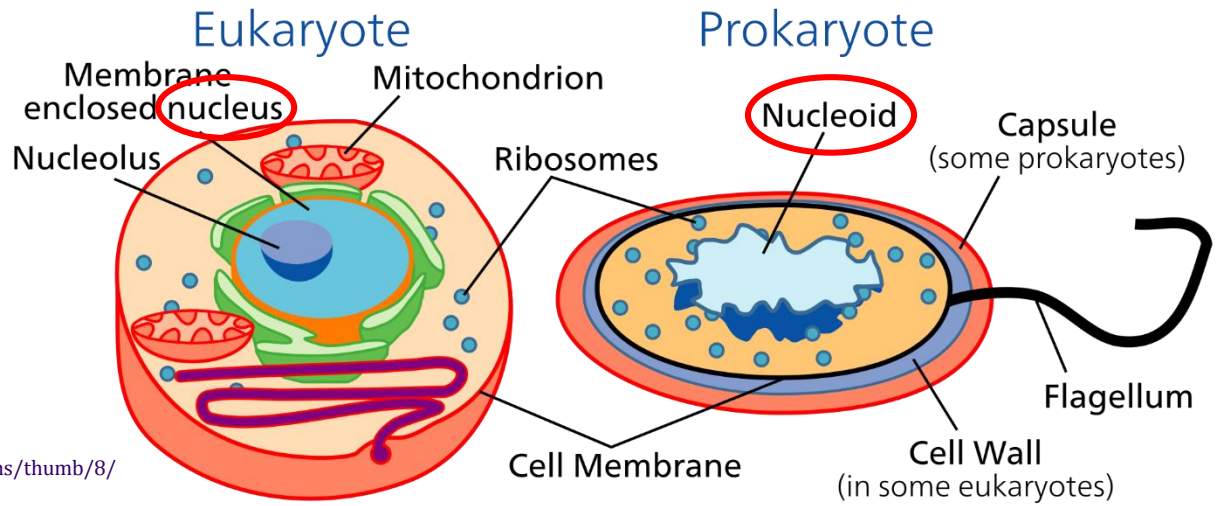
Organization of the genetic material, chromosomes, chromatids

Judit Varga



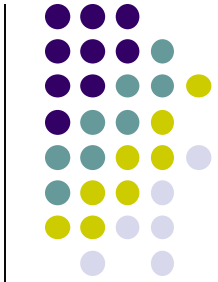
Location of the genetic material

- genetic material: DNA
- prokaryotes:
  - DNA is „naked”
  - in the cytoplasm → nucleoid
- eukaryotes:
  - DNA binds to proteins → **chromatin**
  - in the nucleus



https://upload.wikimedia.org/wikipedia/commons/thumb/8/83/Celltypes.svg/2000px-Celltypes.svg.png

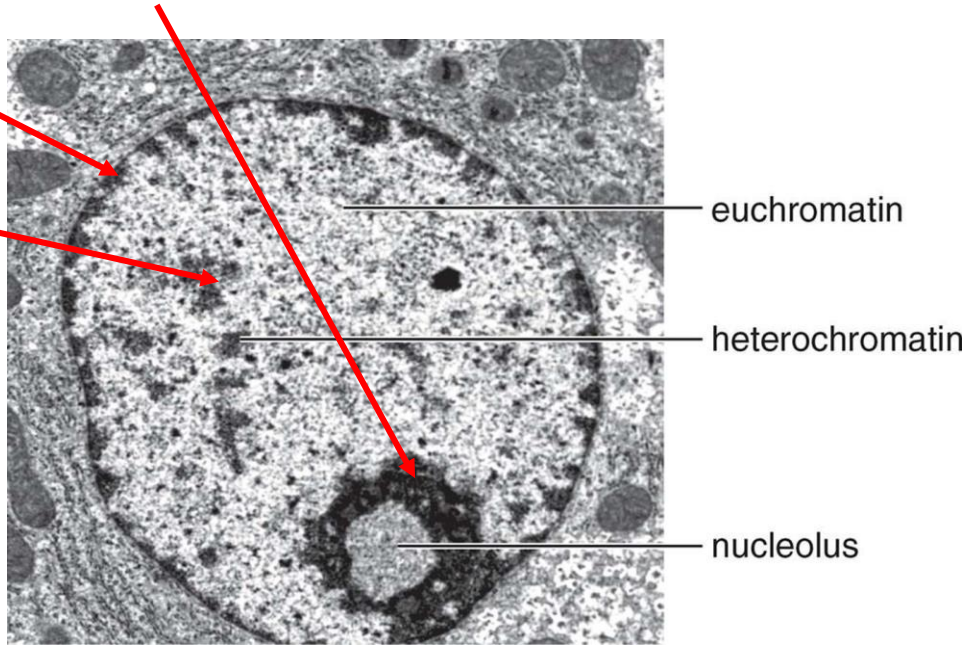
Chemical composition of chromatin



- only in eukaryotic cells
- chromatin = DNP (deoxiribonucleoprotein)
- DNA + proteins + RNA + ions
  - DNA: genetic material
  - proteins
    - histones: structural proteins
    - nonhistone proteins: many types
      - enzymes → DNA polymerases, RNA polymerases
      - regulatory proteins → transcription factors
  - RNA: different types
  - ions (e.g. Mg^{++}, Ca^{++}): stabilization

Types of chromatin

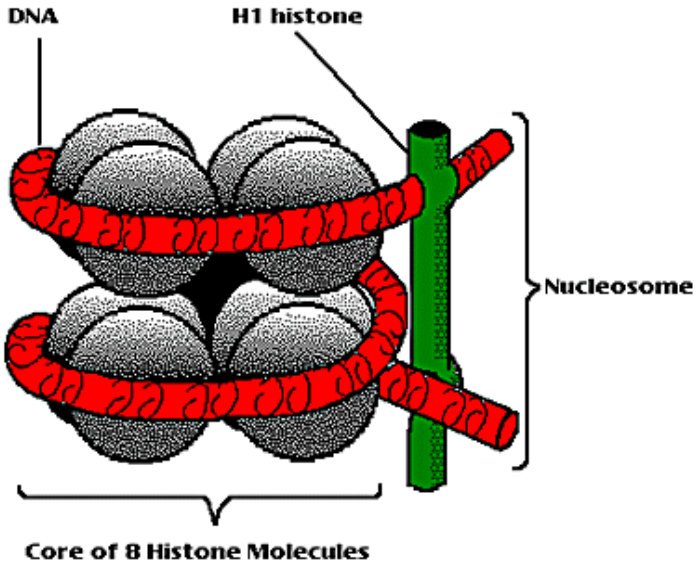
- euchromatin: transcriptionally active
- heterochromatin: transcriptionally inactive
  - perinucleolar/nucleolus-associated
  - peripheral/marginal
  - diffuse



http://schoolbag.info/chemistry/mcat_biochemistry/mcat_biochemistry.files/image140.jpg

Chromatin organization

- DNA: 2 m ↔ nucleus: 10 µm
- levels:
  - euchromatin:
    - DNA double helix
    - beads-on-a-string:
      - nucleosome: histone octamer + DNA
      - linker DNA
  - heterochr.:
    - solenoid
    - looped domains
    - chromosome (during cell division)
- condensation:
  - chromatin becomes more compact
  - euchromatin → heterochromatin



Nucleosome

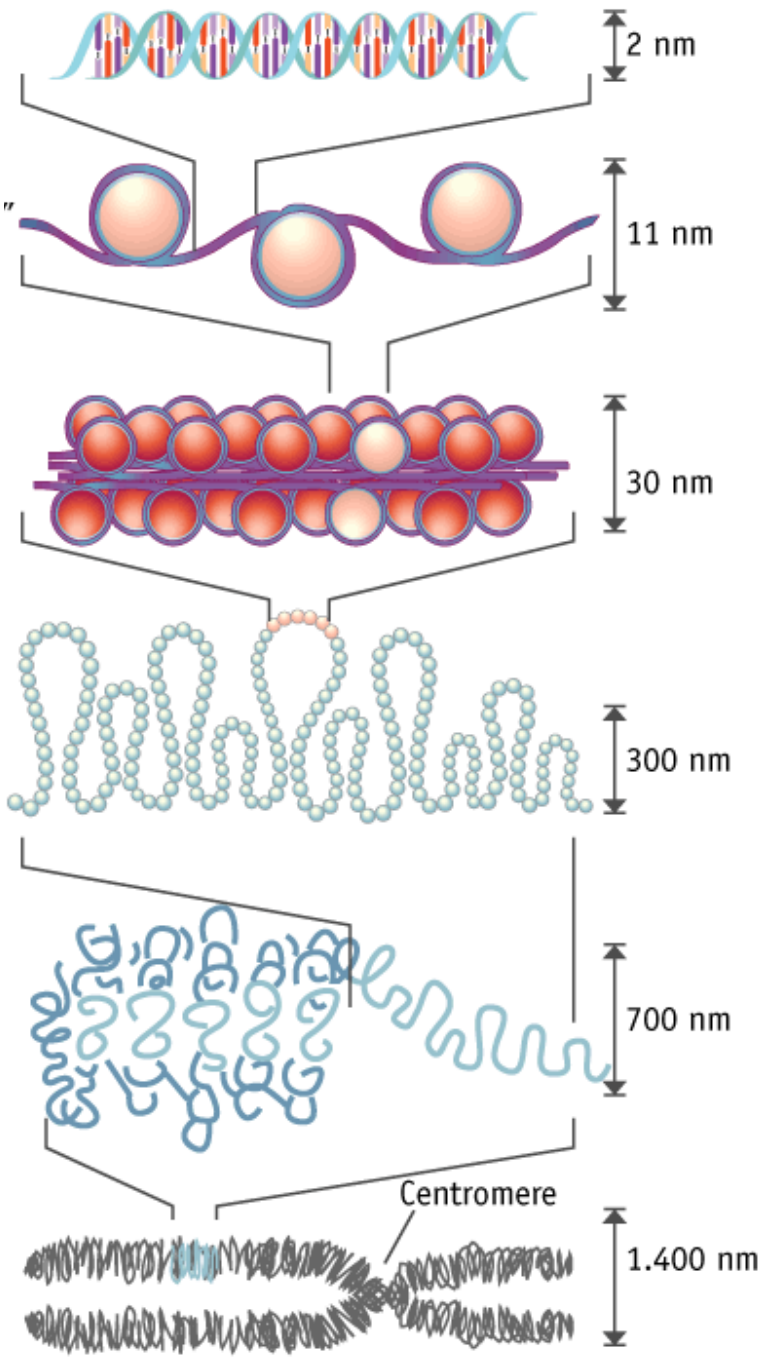
http://www.accessexcellence.com/AB/GG/nucleosome.gif

double helix

„beads on a string”

solenoid

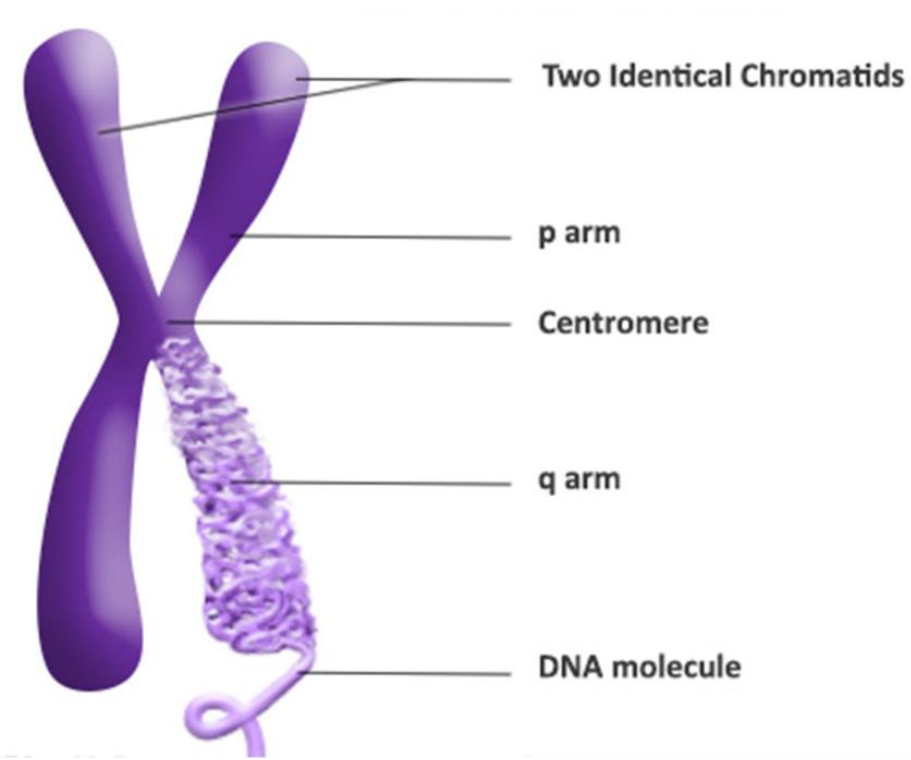
chromosome



http://www.medinfo.net/research/~/media/5EEB8DBAA4D94610AFFC85A4834F2A2A.ashx

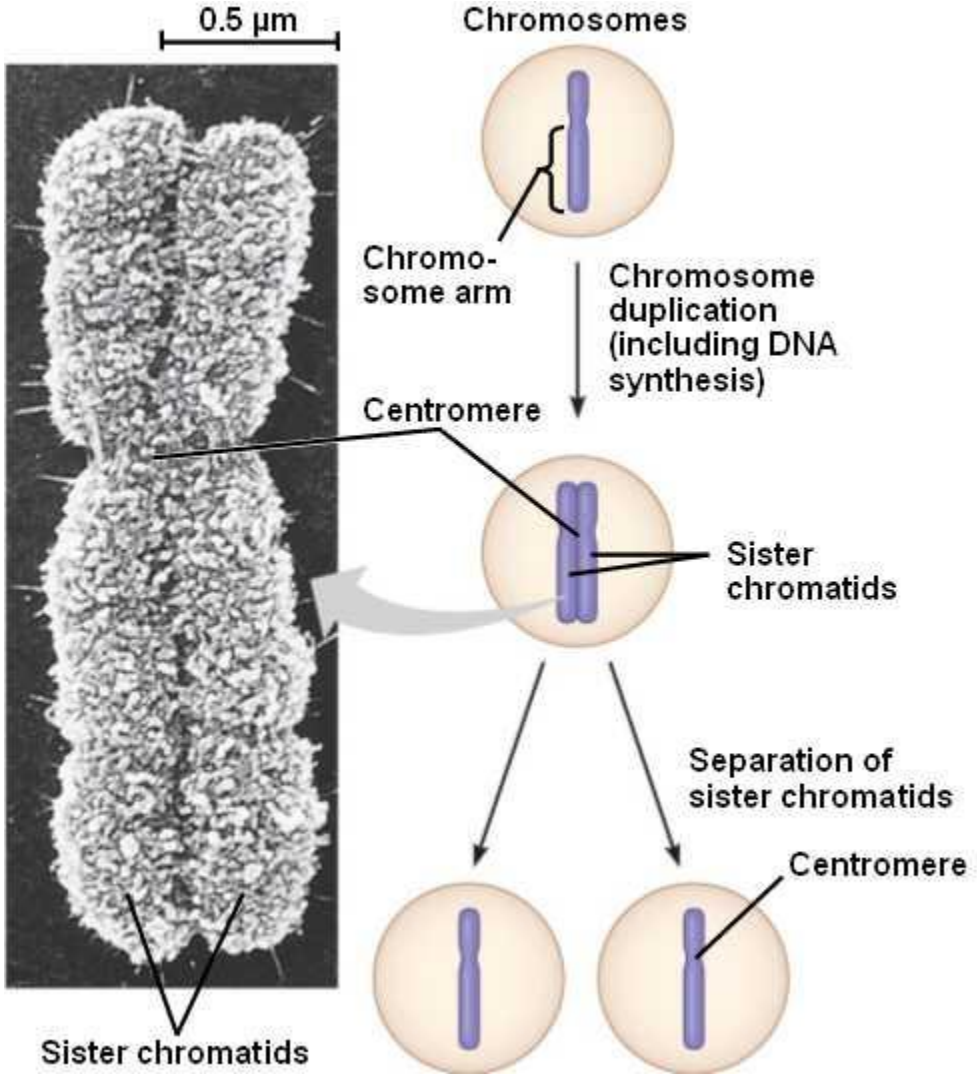
Chromosome structure

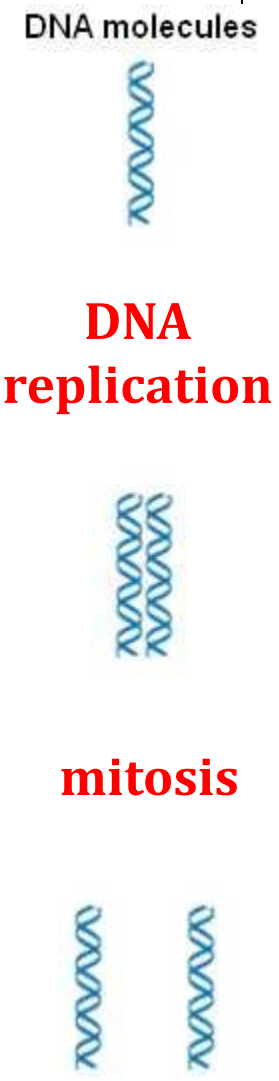
- cell division: equal distribution of DNA → chromosomes form
- 2 chromatids/chromosome → are identical
- 1 DNA molecule/chromatid
- centromere, kinetochore
- telomeres
- short (p) and long (q) arm



http://www.passmyexams.co.uk/GCSE/biology/images/structure-chromosome.jpg

Chromosome structure II.

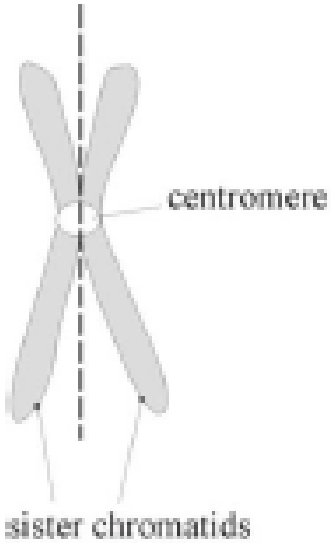




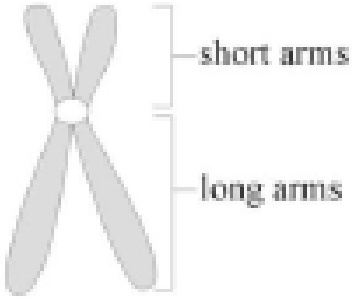
http://bio1151.nicerweb.com/Locked/media/ch12/12_04ChromosomeDuplicat.jpg

Types of chromosomes

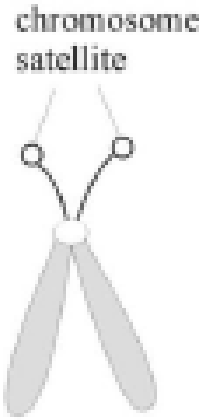
- based on the position of the centromere:



metacentric

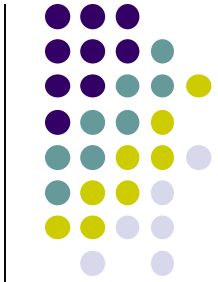


submetacentric



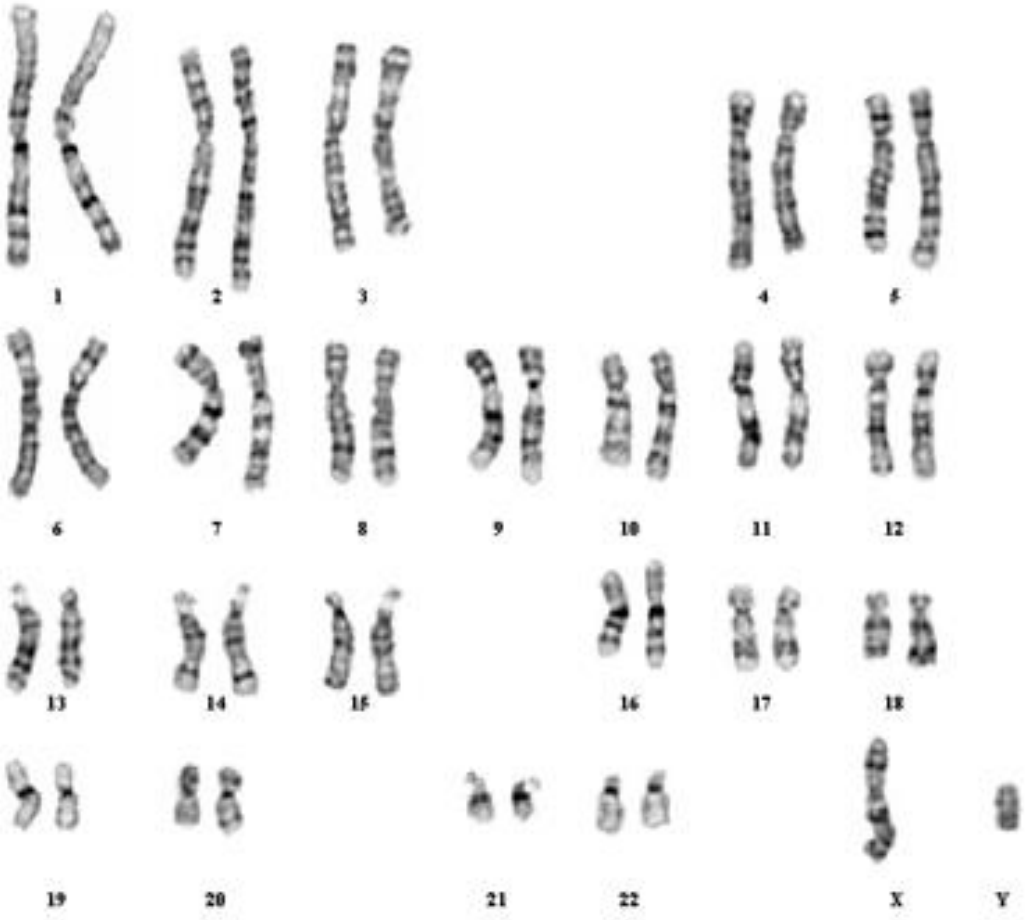
acrocentric

The normal human karyotype

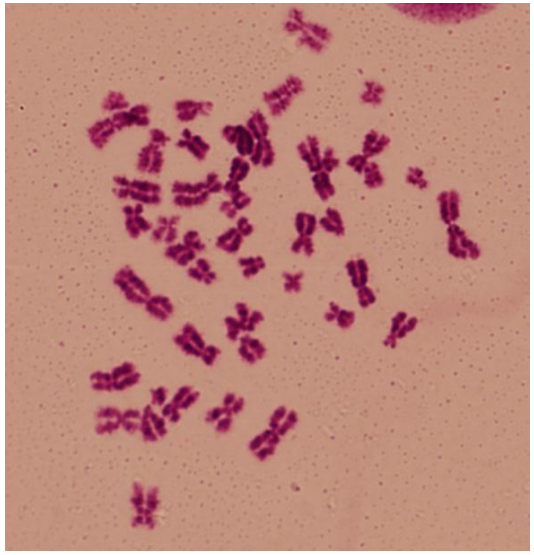


- karyotype: all chromosomes of a cell

- humans:
  - somatic cells → diploid (2n)
    - 23 pairs / 46 chromosomes
    - 1 pair = 2 homologous chromosomes
    - 44 autosomes + 2 sex chromosomes
    - males: 46,XY
    - females: 46,XX
  - gametes/germ cells → haploid (n)
    - 23 chromosomes
    - sperm: 23,X or 23,Y
    - oocyte: 23,X



Denver system

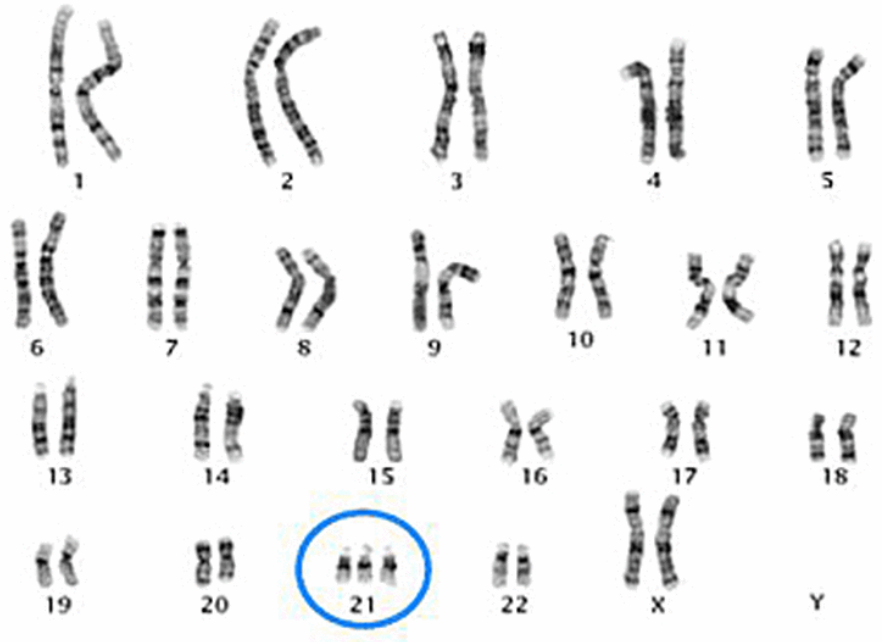


Giemsa staining

Karyotype abnormalities

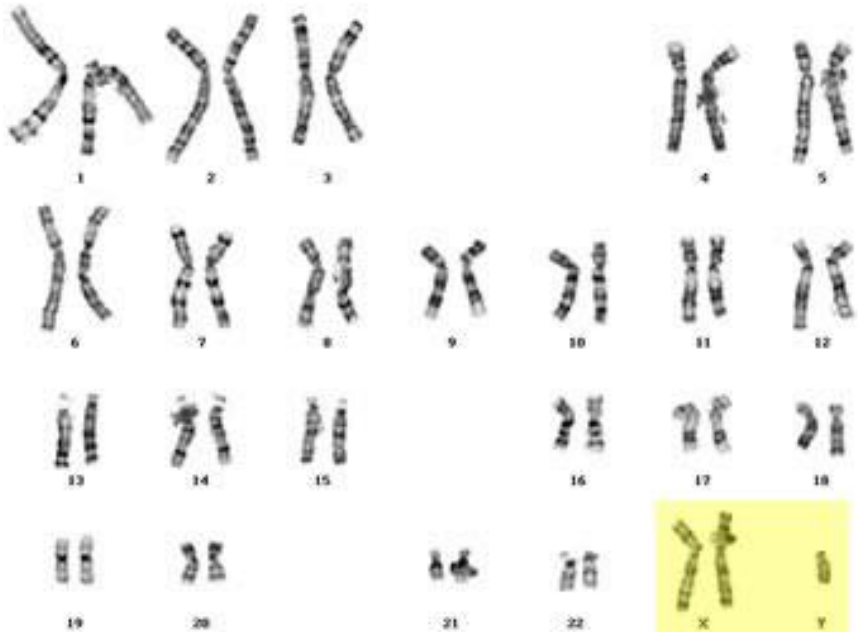
1. structural (deletion, translocation, etc.)
2. numerical

Down syndrome (trisomy 21)

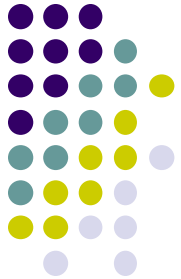


http://downsyndromereport.weebly.com/uploads/2/6/8/5/26858943/137697.gif?726

Klinefelter syndrome (47,XXY)



http://oncofertility.northwestern.edu/sites/oncofertility/files/legacy_files/karyotype.jpg



Thank you for your attention! ☺