Organization of the genetic material, chromosomes, chromatids

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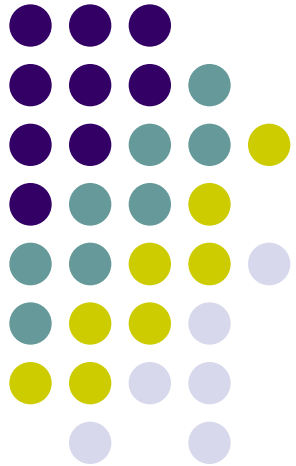
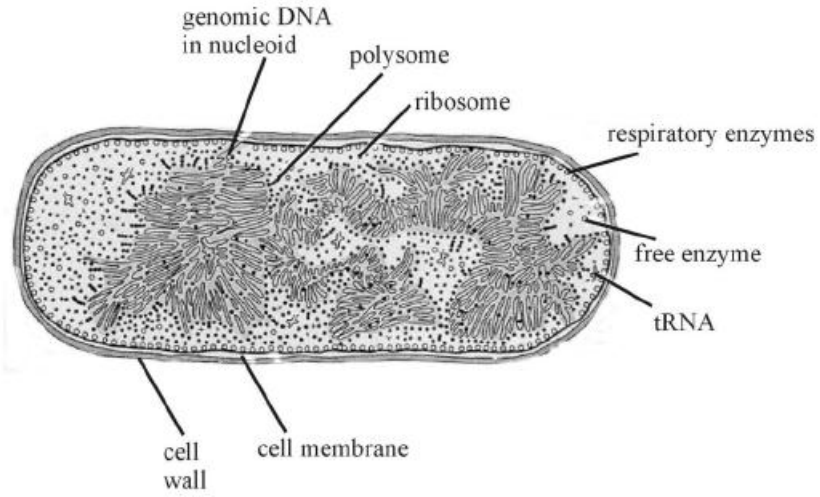
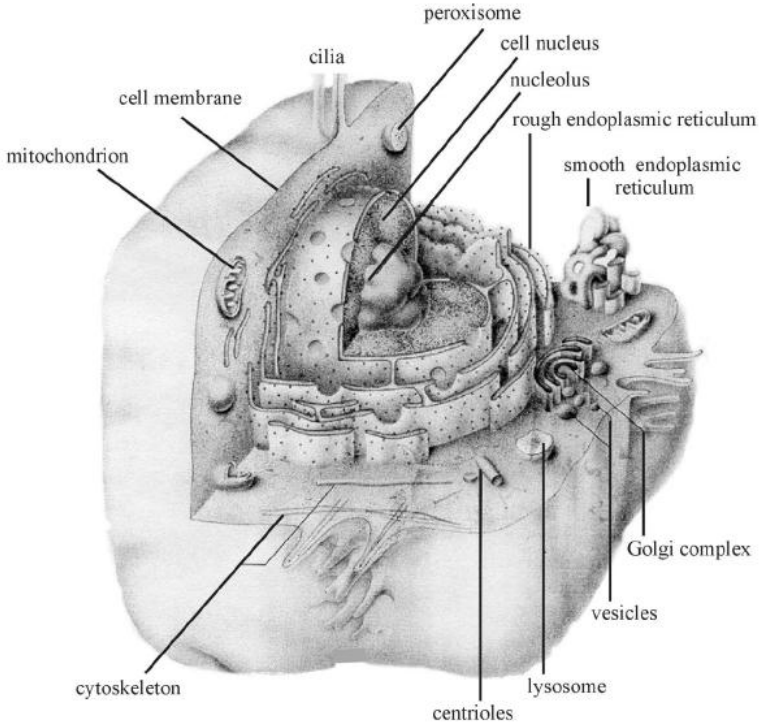


2011. 08. 11.

Location of the genetic material

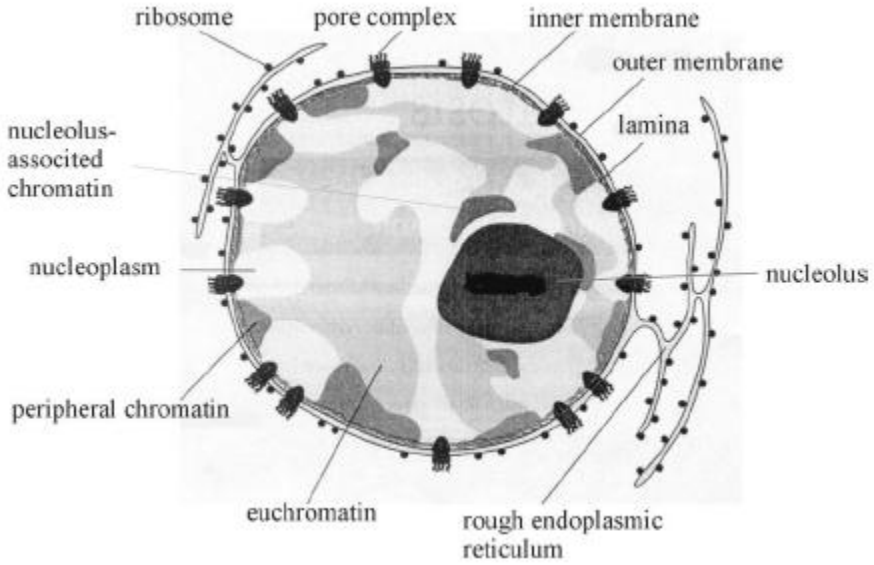
- Prokaryotes: in the cytoplasm → nucleoid
- Eukaryotes: in the nucleus → chromatin





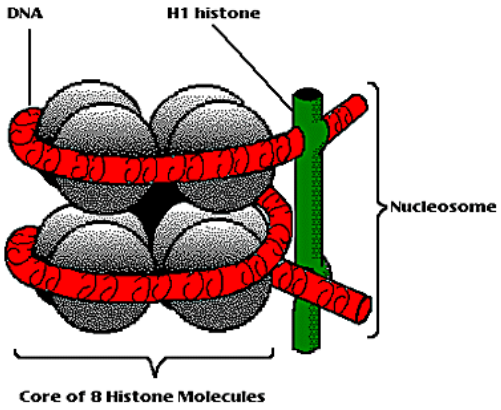
Chromatin

- only in eukaryotic cells
- in non-dividing cells (in the interphase)
- euchromatin: transcriptionally active
- heterochromatin: transcriptionally inactive
  - perinucleolar/nucleolus-associated
  - peripheral/marginal
  - diffuse

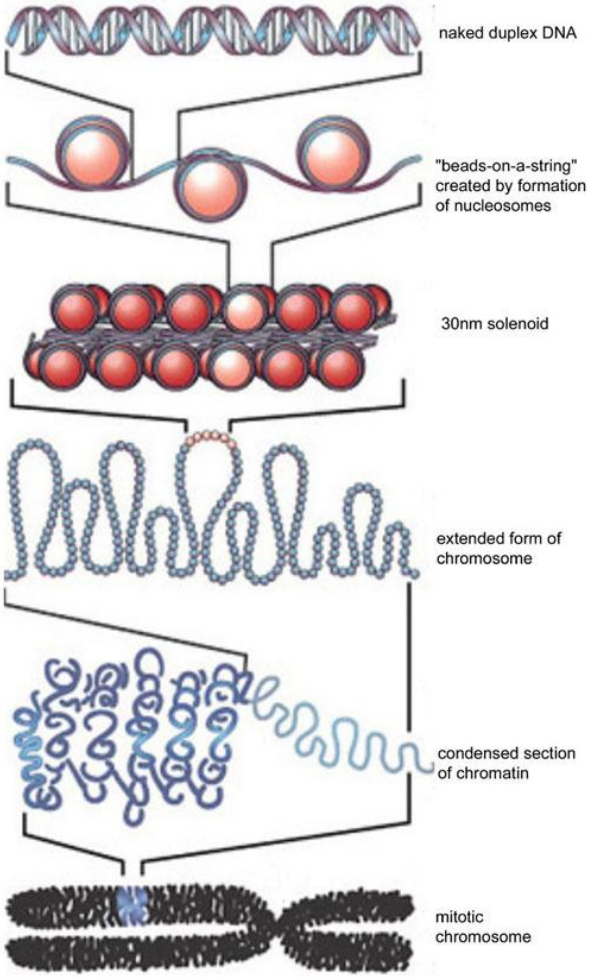


Chromatin organization

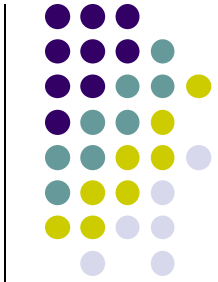
- condensation: chromatin becomes more compact
- levels:
  - DNA double helix
  - beads-on-a-string:
    - nucleosome: histone octamer + DNA
    - linker DNA
    - (chromatosome: nucleosome + H1 histone)
  - solenoid
  - looped domains
  - chromosome



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



The chemical composition of chromatin

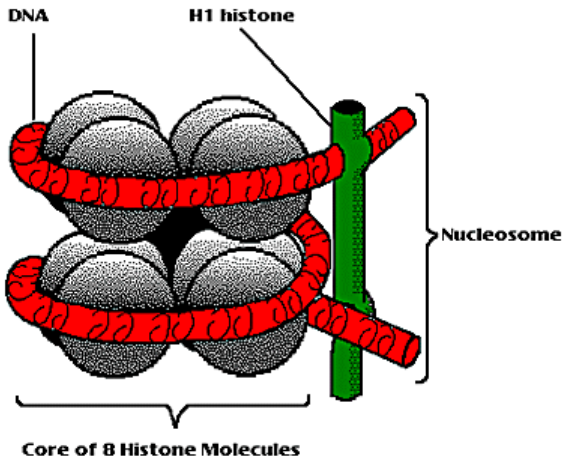


- DNA
- Proteins
  - Histones
  - Nonhistone proteins
- RNA
  - Pre-mRNA, mature mRNA
  - rRNA
  - tRNA …etc.
- inorganic ions
  - Mg^{++}
  - Ca^{++}

Proteins of chromatin I.

Histone proteins

- basic proteins (rich in Lysine, Arginine)
  - Nucleosomal histones (H2A, H2B, H3, H4)
    - octamer in nucleosome
  - H1 histone
    - outside the nucleosome
    - induces solenoid formation
- are highly conserved
- structural function
- regulation of gene expression
- chemical modifications
  - Phosphorylation → chromatin condensation
  - Acetylation → chromatin **de**condensation



Nucleosome

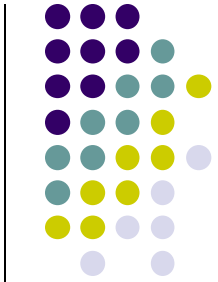
http://www.accessexcellence.com/AB/GG/nucleosome.gif

Proteins of chromatin II.

Nonhistone proteins

- tissue-specific expression
- different in structure
- different in function
  - Structural proteins (e.g. lamins)
  - Enzymes (e.g. DNA, RNA polymerases)
  - Transcription factors
  - Receptor proteins (e.g. steroid receptors)
  - Transport proteins (e.g. importin)
  - Chaperones (e.g. nucleoplasmin)

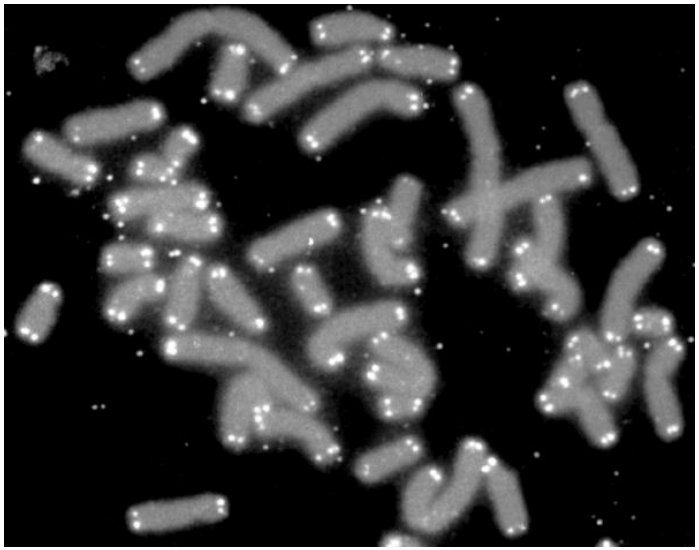
Basic genetic terms

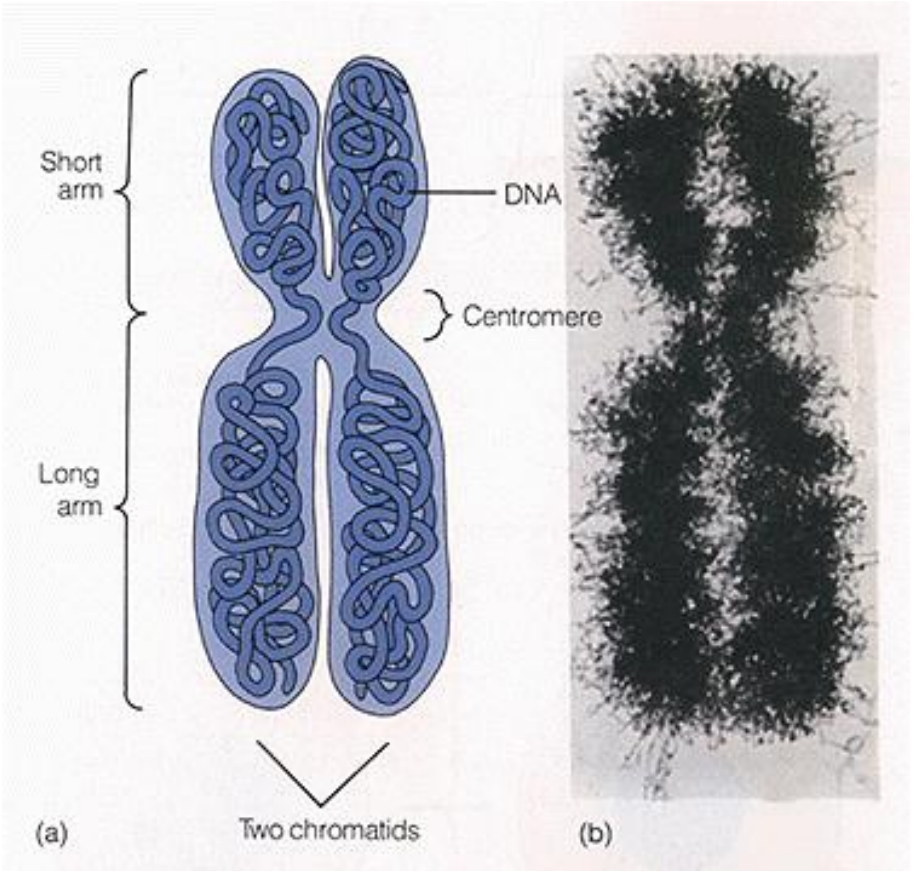


- **Gene**: region of DNA that codes for a protein
- **Alleles**: variant forms of a gene
- **Locus**: the site of a gene in a chromosome
- **Homologous chromosomes**: members of a chromosome pair
- **Somatic chromosomes/autosomes**: 1-22
- **Sex chromosomes**: X, Y
- **Homozygote**: carries identical alleles in a locus of homologous chromosomes
- **Heterozygote**: carries different alleles in a locus of homologous chromosomes
- **Genotype**: genetic constitution
- **Phenotype**: features that appear

Chromosomes

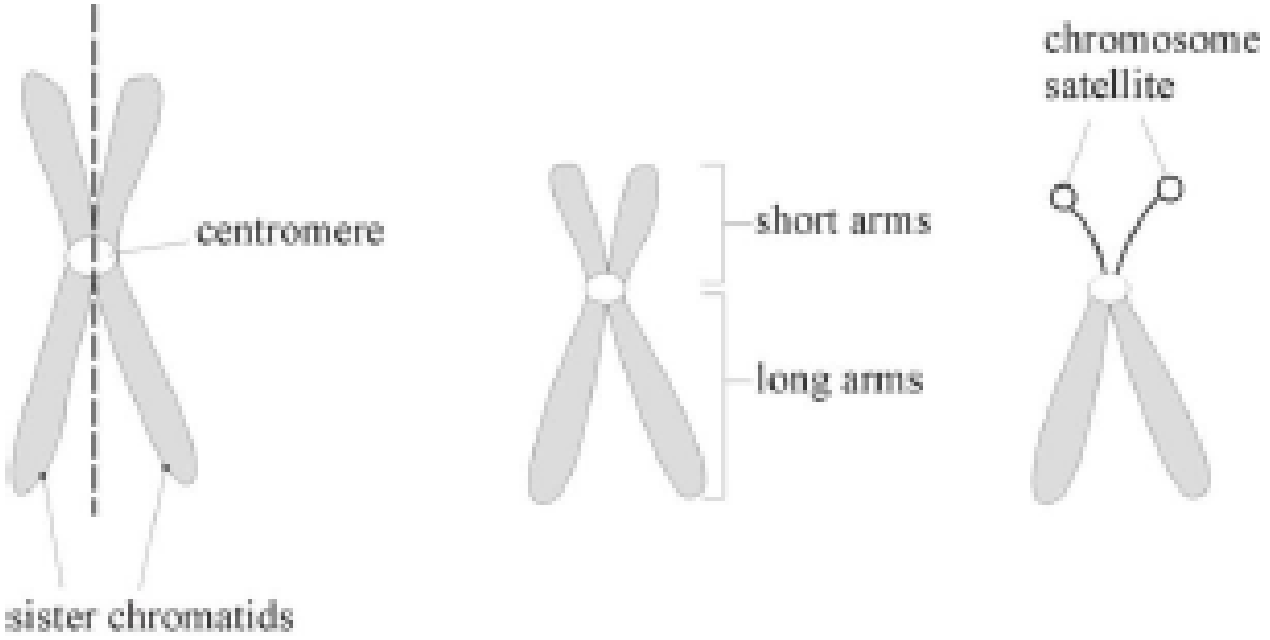
- chromatids
- centromere
- telomeres
- short (p) and long (q) arm





Chromosomes II.

- types (based on the position of the centromere):
  - metacentric
  - submetacentric
  - acrocentric



The normal human karyotype

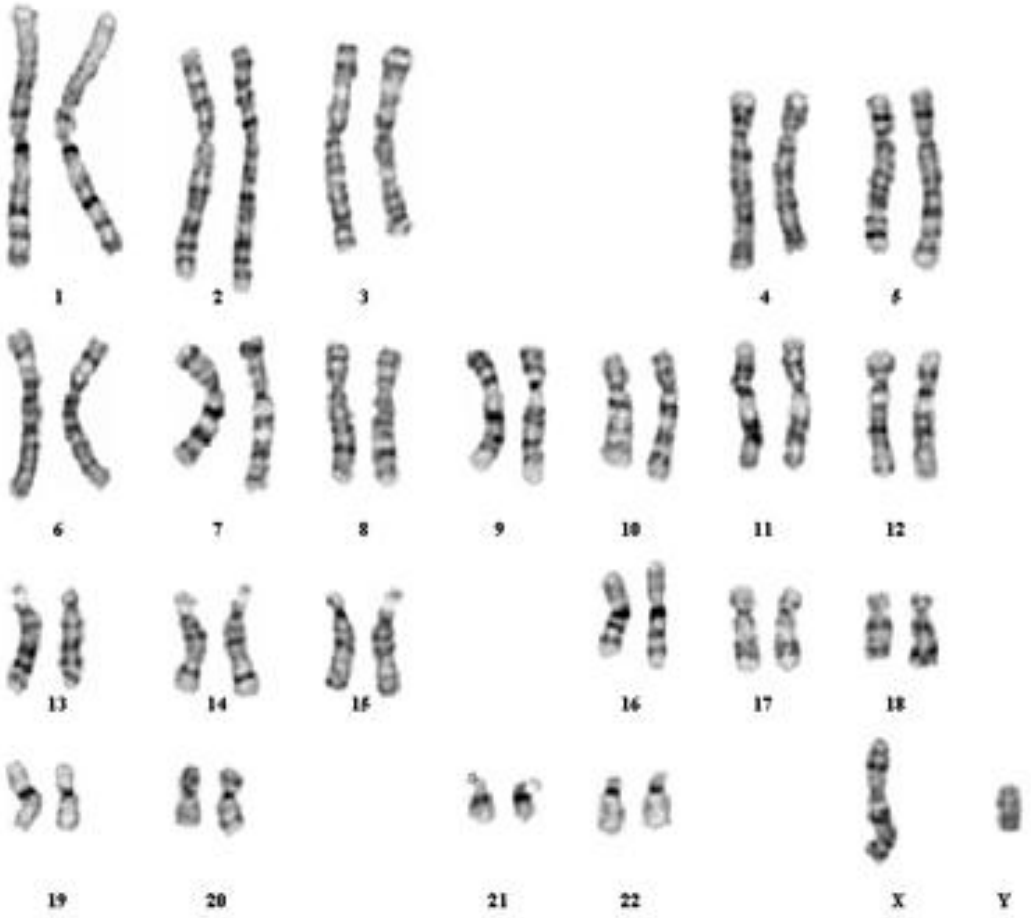
humans:

44 autosomes + 2 sex chromosomes

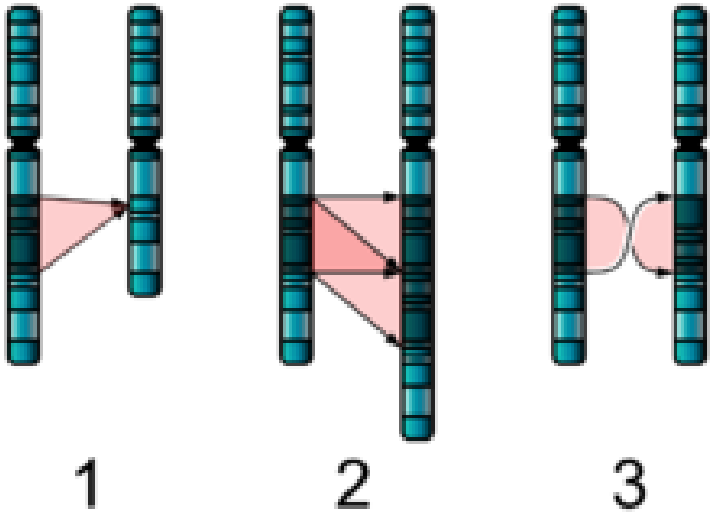
(altogether 23 pairs = 46 → 2n)

males: 44+XY

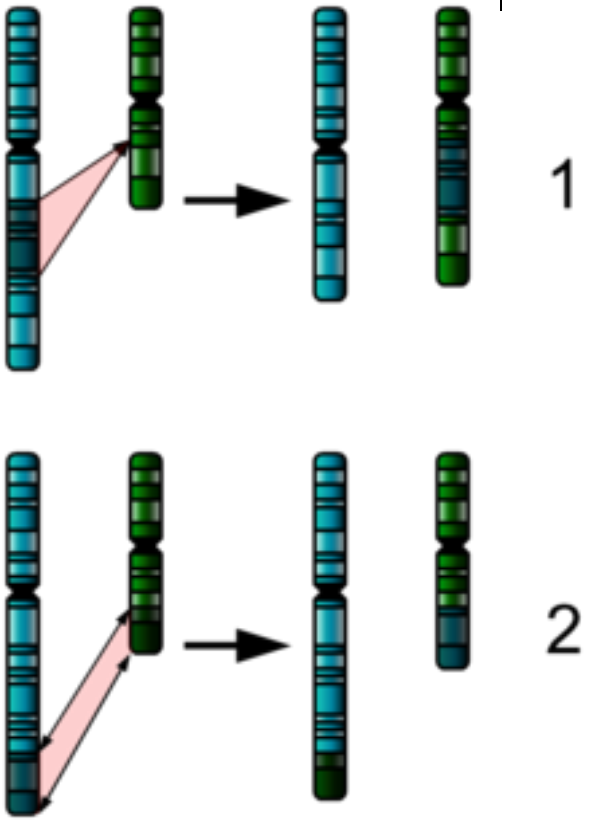
females: 44+XX



Structural chromosome abnormalities

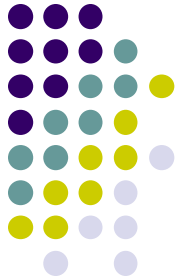


Single chromosome mutation
1)**Deletion** (e.g. Lejeune syndrome → cri du cat)
2)Duplication
3)**Inversion**



Two-chromosome mutations
1)Insertion
2)**Translocation**

Numerical chromosome abnormalities



- Polyploidy (e.g. triploidy, 3n) → **lethal**
- Aneuploidy
  - Monosomy
    - Normally Y chromosome in males
    - Turner syndrome (44 + X0)
  - Trisomy
    - Down syndrome (trisomy 21)
    - Patau syndrome (trisomy 13)
    - Edwards syndrome (trisomy 18)

Down syndrome (trisomy 21)