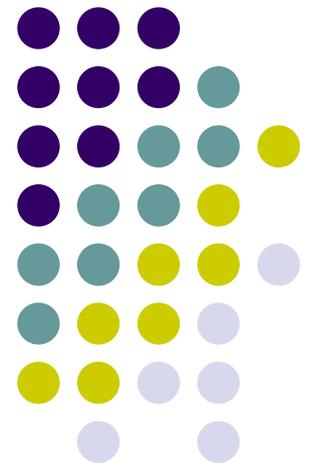


# Organization of the genetic material, chromosomes, chromatids

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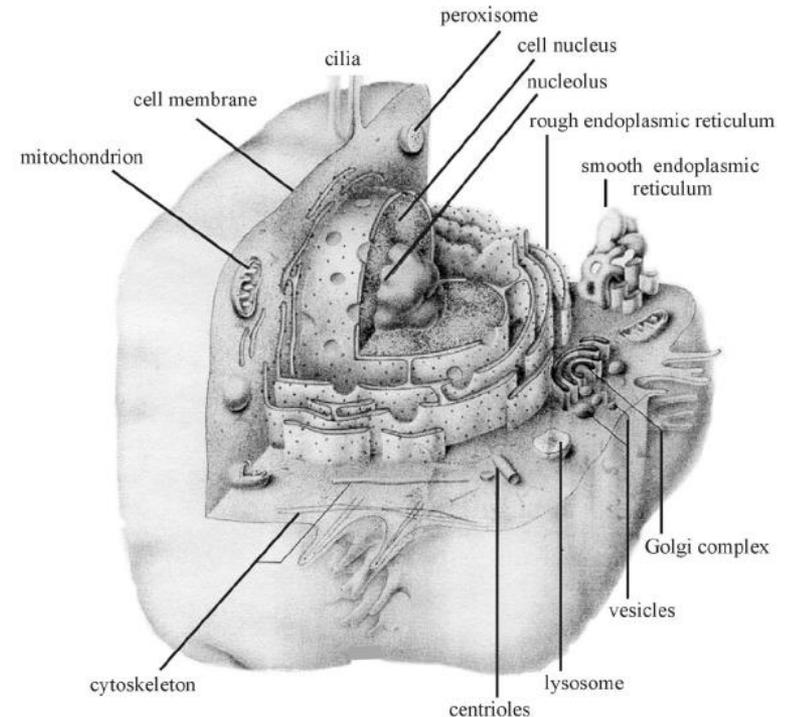
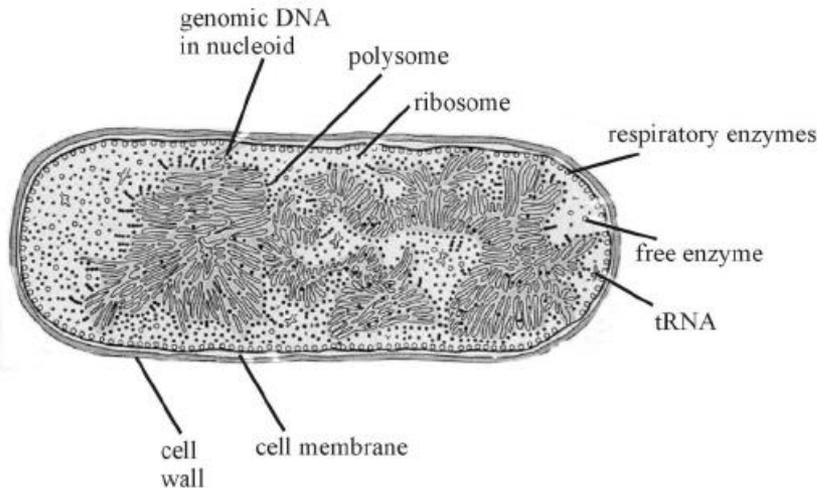
Oktavia Tarjanyi M.D.  
Gergely Berta M.D.





# Location of the genetic material

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



# Why do eukaryotes have chromatin?

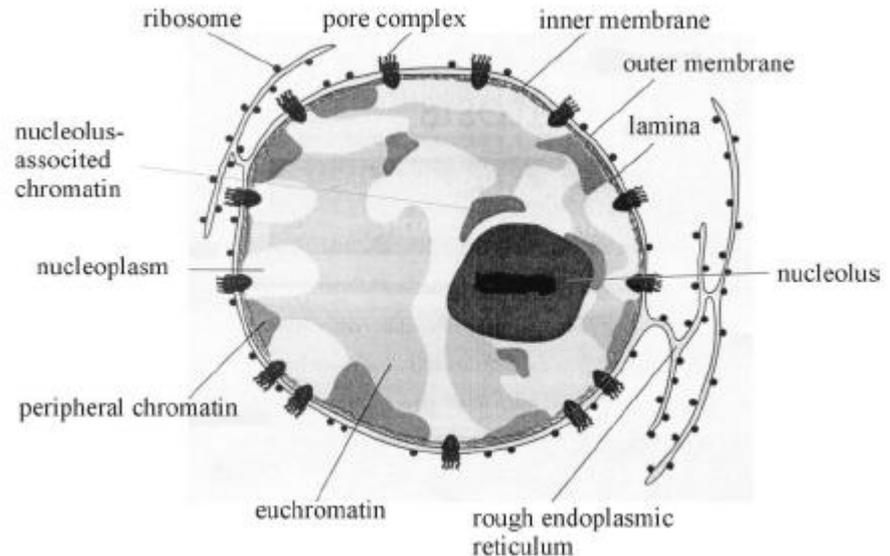


- Chromatin is a packaged form of the DNA into a smaller volume to fit in the cell
- Proteins of the chromatin strengthen the DNA and allow mitosis and meiosis because they allow the formation of the chromosomes.
- Proteins of the chromatin serve mechanisms to control gene expression.

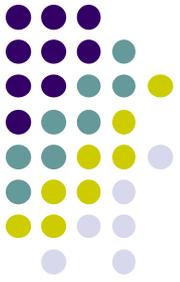
# Chromatin



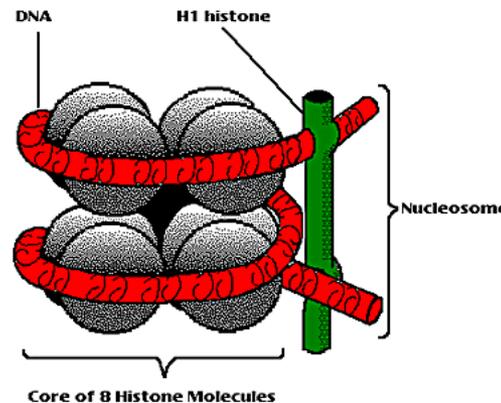
- only in eukaryotic cells
- in non-dividing cells (in the interphase)
- euchromatin: transcriptionally active
- heterochromatin: transcriptionally inactive
  - perinucleolar/nucleolus-associated
  - peripheral/marginal
  - diffuse



# Chromatin organization

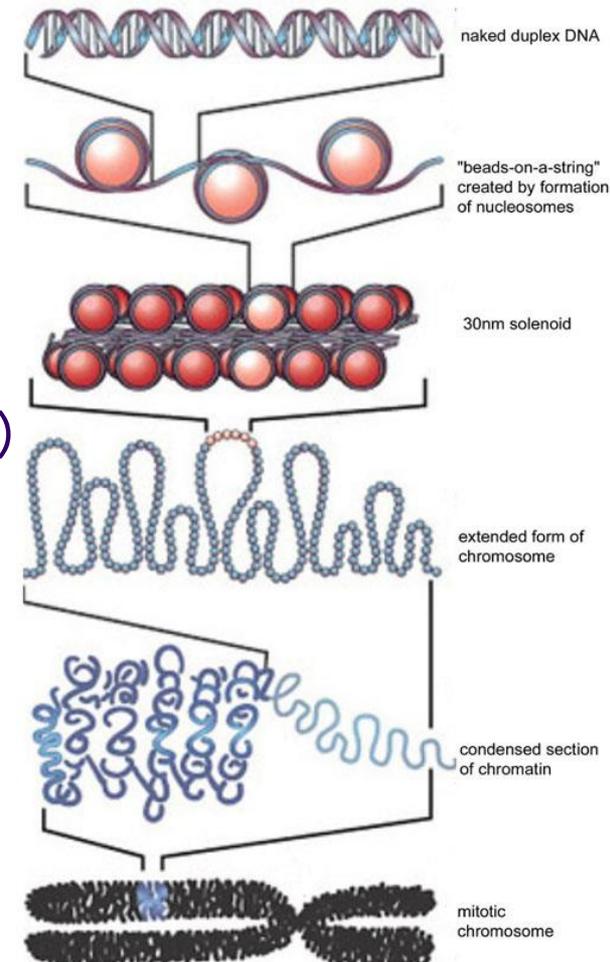


- condensation: chromatin becomes more compact
- levels:
  - DNA double helix
  - beads-on-a-string:
    - nucleosome: histone octamer + DNA
    - linker DNA
    - (chromatosome: nucleosome + H1 histone)
  - solenoid
  - looped domains
  - chromosome

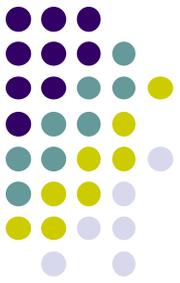


Nucleosome

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



# The chemical composition of chromatin



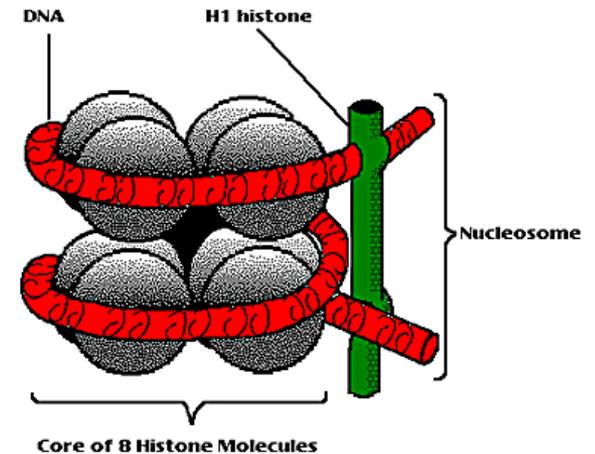
- DNA
- Proteins
  - Histones
  - Nonhistone proteins
- RNA
  - Pre-mRNA, mature mRNA
  - rRNA
  - tRNA ...etc.
- inorganic ions
  - $Mg^{++}$
  - $Ca^{++}$



# Proteins of chromatin I.

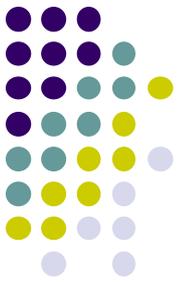
## Histone proteins

- 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>

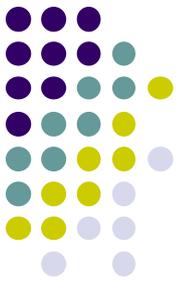


# Proteins of chromatin II.

## **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)

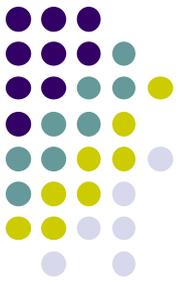
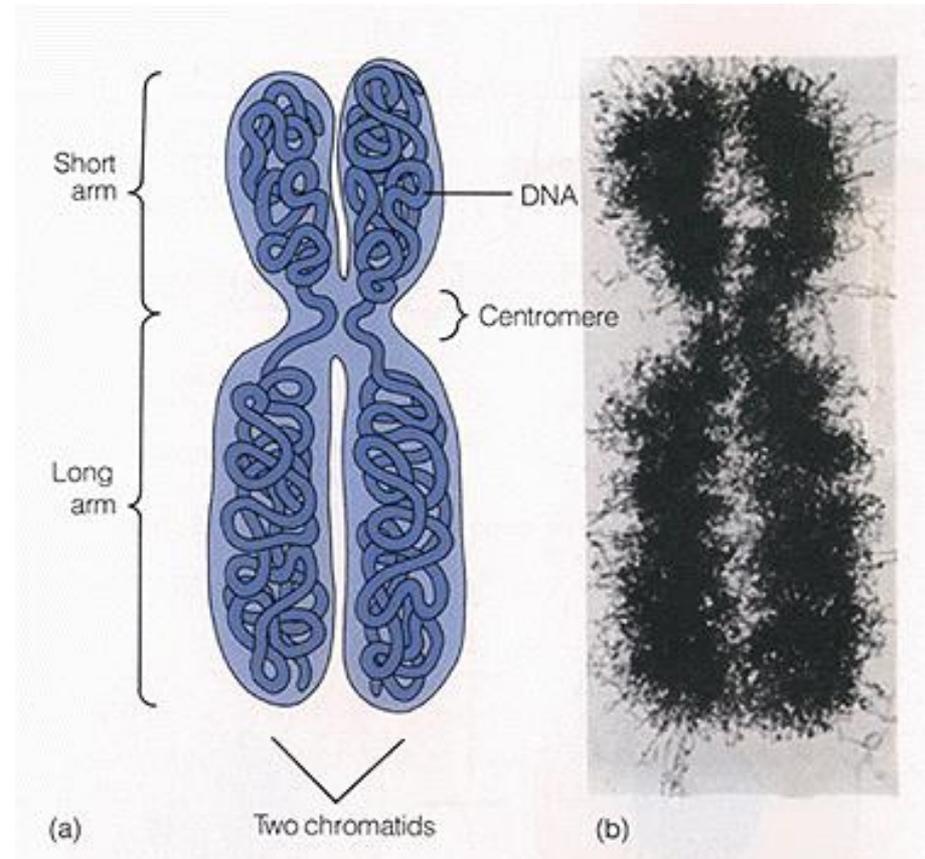
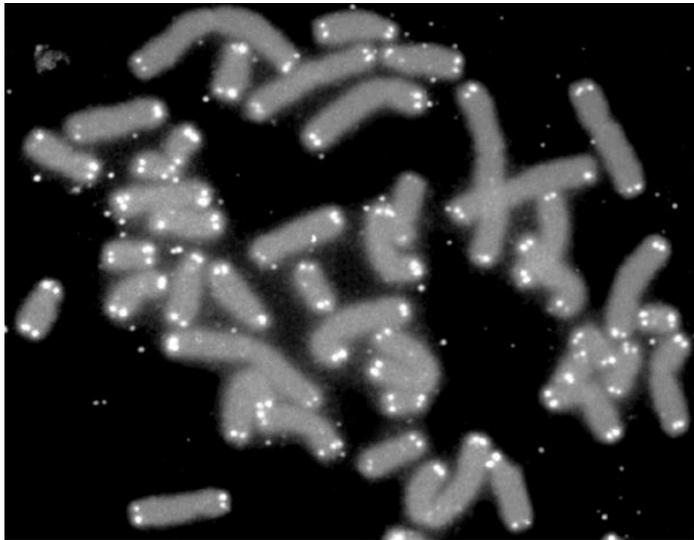
# Genetic terms associated with chromosomes



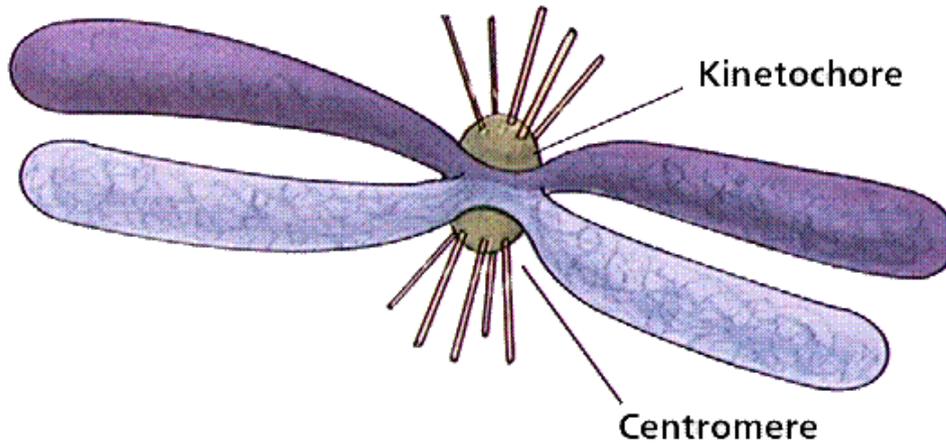
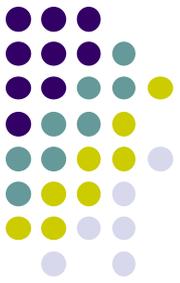
- **Gene:** region of DNA that codes for a protein
- **Locus:** the site of a gene in a chromosome
- **Homologous chromosomes:** members of a chromosome pair
- **Somatic chromosomes/autosomes:** 1-22
- **Sex chromosomes:** X, Y

# Chromosomes

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

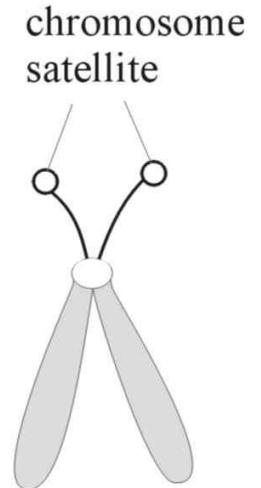
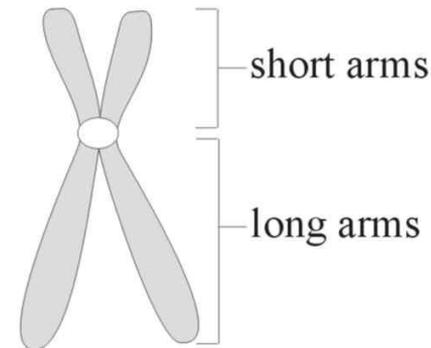
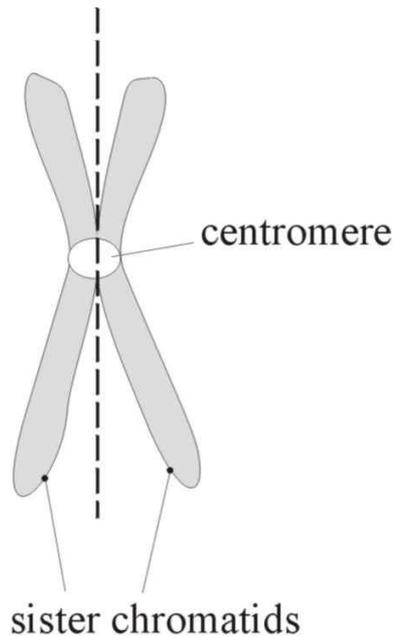


# Chromosomes II.

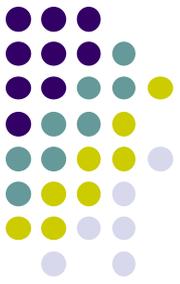


- types (based on the position of the centromere):

- metacentric
- submetacentric
- acrocentric

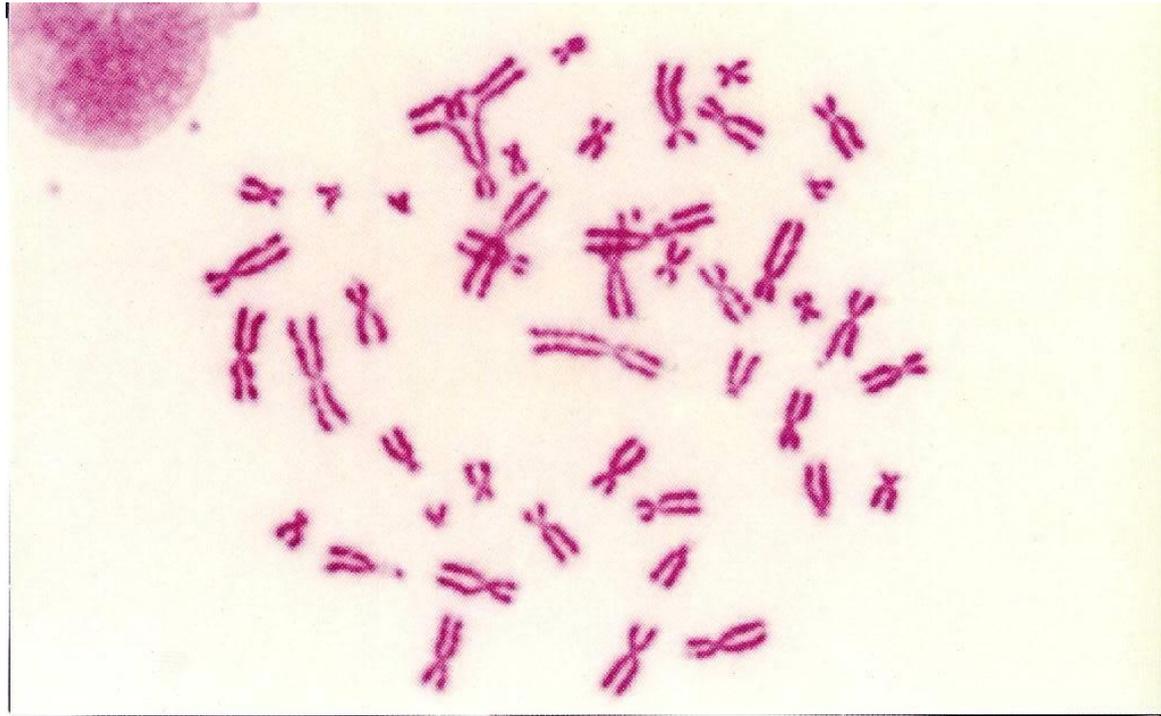


# Karyotype



- The set of all chromosomes deriving from a cell's nucleus is called karyotype.

The number of the chromosomes is different in various species. For example humans have 46, dogs have 78, cats have 38.





# The normal human karyotype

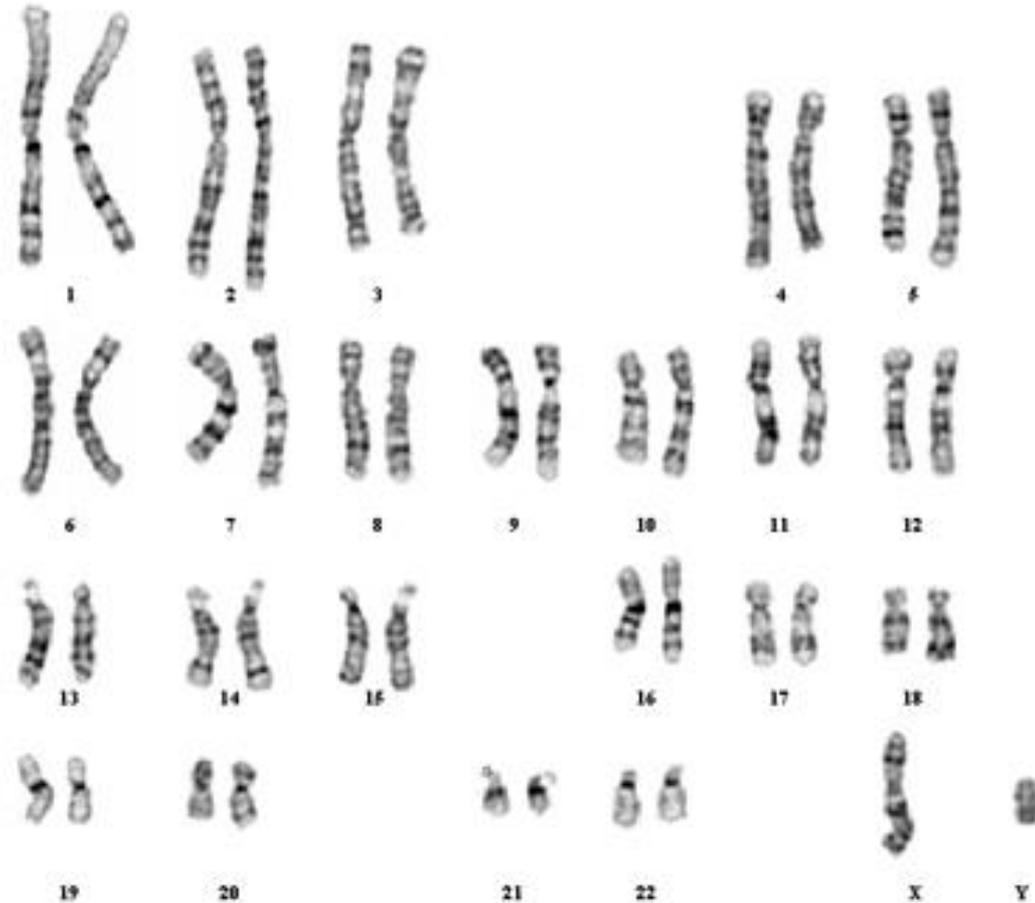
## humans:

44 autosomes + 2  
sex chromosomes

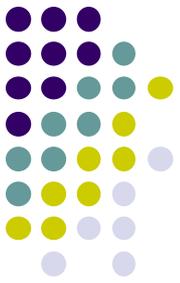
(altogether 23  
pairs = 46 → 2n)

**males:** 44+XY or  
46, XY

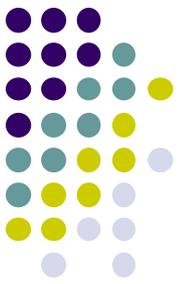
**females:** 44+XX  
or 46, XX



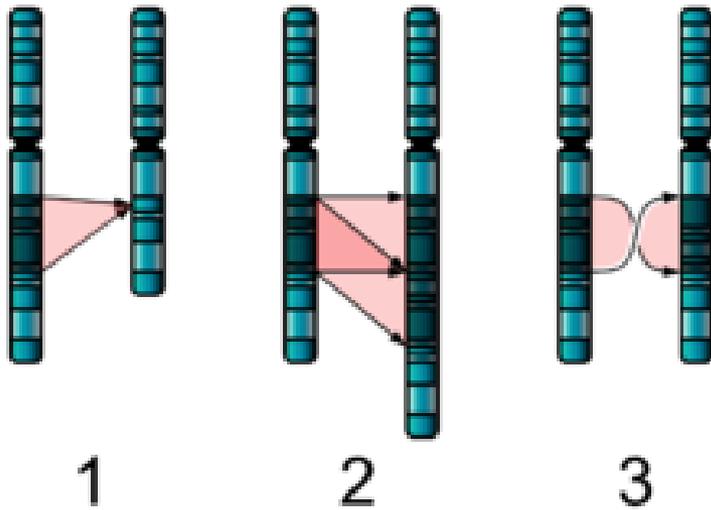
# Diploid / haploid cells



- Sexually reproducing species have somatic cells (body cells), which are diploid [ $2n$ ] having two sets of chromosomes, one from the mother and one from the father.
- Gametes, reproductive cells (egg, sperm), are haploid [ $n$ ]: they have one set of chromosomes.
- Gametes are produced by meiosis of a diploid germ line cell.
- When a male and a female gamete merge (fertilization), a new diploid cell (fertilized egg) is formed. From this cell a new organism develops.



# Structural chromosome abnormalities

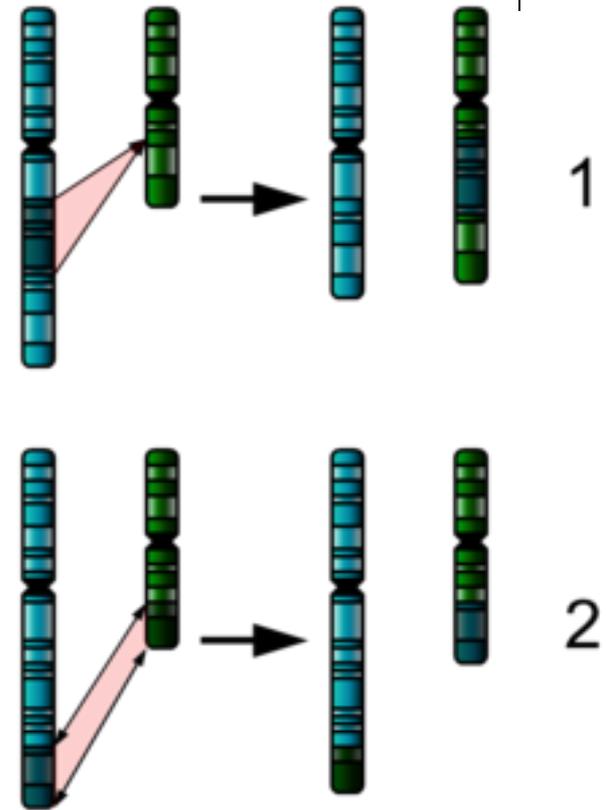


Single chromosome mutation

1) **Deletion** (e.g. Lejeune syndrome → cri du cat)

2) Duplication

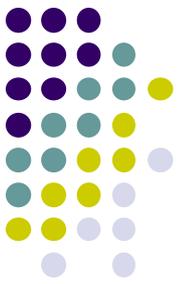
3) **Inversion**



Two-chromosome mutations

1) Insertion

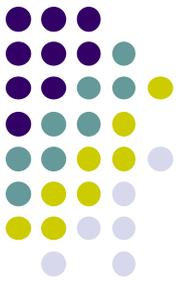
2) **Translocation**



# 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)



# FISH

