The science of inheritance Genetics

The central dogma of MCB:

| DNA | $4 \underset{3}{3}$ |
| :---: | :---: |
| RNA | $\stackrel{3}{3}$ |
|  | $\stackrel{\text { ¢ }}{\text { ¢ }}$ |



## Basic terms I.




## Chromosome = a continuous piece of DNA

- 22 pairs of numbered autosomes + one pair of sex chromosomes, X and Y / humans
- each parent contributes one chromosome to each pair (homologous chromosomes)
- condensed (well stainable) bodies in dividing cells $\leftrightarrow$ more relaxed chromatin, occupying „chromosome territories" in the nucleus of interphase cells
+ circular chromosomes / dsDNA in mitochondria



## Basic terms II.

Gene $=$ the unit of inheritance, a DNA segment $\rightarrow$ a functional (m)RNA $\rightarrow$ a protein / / a feature
Allele = the variant of a gene (normal / wild type or mutant)
Genetic locus = the exact location of a gene on a chromosome (eg. 7q21.13 for EPO)

Genotype = the complete genetic makeup
Phenotype = the observable characteristics / traits
(how we look like / function)


## Basic terms III.

Diploid (2n) cell / organism = contains two homologous copies of each chromosome (except $X \& Y$ in males $=$ hemizygous situation)

Haploid $(\mathrm{n})=$ having only one complete set of chromosomes, half of the normal, diploid number

Homozygous = having identical genes in corresponding genetic loci of homologous chromosomes

Heterozygous = having different alleles in corresponding genetic loci of homologous chromosomes

## Basic terms IV.

The strength of genes = their ability to express the coded property in the phenotype:
Dominant = a single copy / heterozygosity is enough for its manifestation (eg. gain-of-function / mutation, haplo-insufficiency, dominant negative mutation, temporal or spatial alterations in the expression of a gene)

Recessive = manifestation only in homozygotes
(eg. partial / complete loss-of-function mutation)
Incomplete dominance = the feature's manifestation is halfway between the extremes coded by the alleles

Codominance = both coded extremes are equally manifested

## Meiosis in females and in males I



## Meiosis in females and in males



## Tools of genetics

## - pedigrees

## Common symbols:

Male
Mating
Parents and
children:
boy; 1 girl
(in order of birth)
of sex indicated
Affected individuals

