

## DNA & Genes

## The Human Genome and Inheritance

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What is the human genome?

Simply, the different types of sequences that make up the total DNA of a human cell

- 3 billion base pairs
- about 22 000 genes
- Only 2 % of the DNA encode proteins
- Genes include exons and introns
- 50 % repeated sequences (called "junk DNA")

- 23 chromosome pairs  $\rightarrow$  46 chromosomes
- 44 autosomes, 2 sex chromosomes
- X and Y -chromosomes
- $\cdot XX \rightarrow female$
- $\cdot XY \rightarrow Male$



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Chromosomes carrying the same genes are called homologous



# Mutations

- Heard of those mutants in Hollywood movies? Well, this is what it is
- Alterations in DNA sequence
- Some are normal DNA variation
- Caused by chemical and physiological agents and errors in DNA replication
- Cells can repaire some mistakes
- If not repaired, changes in DNA sequence are made permanent by DNA replication



## Point mutations:



### Single base mutations:

- 1. Missense mutation: leads to an amino acid change
- 2. Silent mutation: does not change the amino acid
- 3. Nonsense mutation: causes premature stop-codon



- Frameshift mutations:
  - insertion/deletion dublication
  - translocation
  - →Altered reading frame
    → Severe impacts on protein structure

### Passing on the genetic information:



- Information passed on in the sexual reproduction
- Needed for new characteristics to develop
- Offspring recieve genes by inheriting chromosomes

Two important terms...

Phenotype: The outlook of an organism

Genotype: The genetic information written in DNA

Phenotypes



GCCAAGAATGGCTCCCACCT GGCTCTCAGACATTCCCCTGG TCCAACCCCCAGGCCATCAAG ATGTCTCAGAGAGGCGGCTAG ACACCCAGAGACCTCAAGTGA CCATGTGGGAACGGGATGTTT CCAGTGACAGGCA







### All somatic cells • 23 chromosome pairs

- (46 chromosomes)
- Diploid cells, 2n



Sperm cell
23 chromosomes
Haploid cell, n



Egg cell • 23 chromosomes • Haploid cell, n



#### A chromosome pare:



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- · A locus
- An allele

#### <u>Mitosis</u>

- Division of somatic cells
- Products two daughter cells from one parent cell
- The number of chromosomes does not change
- DNA duplicates before entering the mitosis
- MITOSIS DNA Replication Cell Division

Takes 1-2 hours

#### Meiosis

- Only in gamete formation
- One diploidic parent cell produces four haploid gametosytes
- Mature gametocytes have 23 chromosomes (n)







Crossing-over and recombination during meiosis

 Chromatids change parts between homologous chromatids during the meiosis

 Causes redistribution of heridary material between the homologous chromosomes

- → number of genes doesn't change
- → new allele combinations are formed



# Inherited diseases

- DNA mutations are significant in development of diseases
- Inherited diseases are caused by mutations passed from a parent to a offspring
- Monogenic diseases: disease is caused by one mutation in one gene
- Multifactionial diseases: disease is caused by interaction of different mutations and environmental factors

• Mendelian inheritance: Presence or absence of the phenotype depends on the genotype at a single locus

- Dominant character: only one allele needed to cause the phenotype (heterozygous)
- **Recessive** character: both allels needed to cause the phenotype (homozygous)





#### Autosomal dominant inheritance:



#### Autosomal recessive inheritance:



X-chromosome linked recessive inheritance:



#### X-chromosome linked dominant inheritance:



