ISAGREED

Mutation (point)

Modul no. 1: Animal Genetics

Martina Miluchová, Michal Gábor

Slovak University of Agriculture in Nitra

Faculty of Agrobiology and Food Resources

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED

Co-funded by the European Union

Mutation

Mutation is a sudden, heritable change appearing in an individual as the result of :

- change in the structure of a gene (**gene mutation**);
- changes in the structure of chromosomes (**chromosome mutation**);
- changes in the number of chromosomes (**genome mutation**).

ISAGREED

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

Co-funded by the European Union

Gene mutation

Gene = point mutation is a change in the DNA sequence that makes up a gene. That mutation caused change in the order or number of nucleotides in the DNA structure of a gene.

We distinguish mutations with an effect on:

- DNA sequence
- amino acid sequence
- reading frame
- function of the protein

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

Effect on DNA sequence

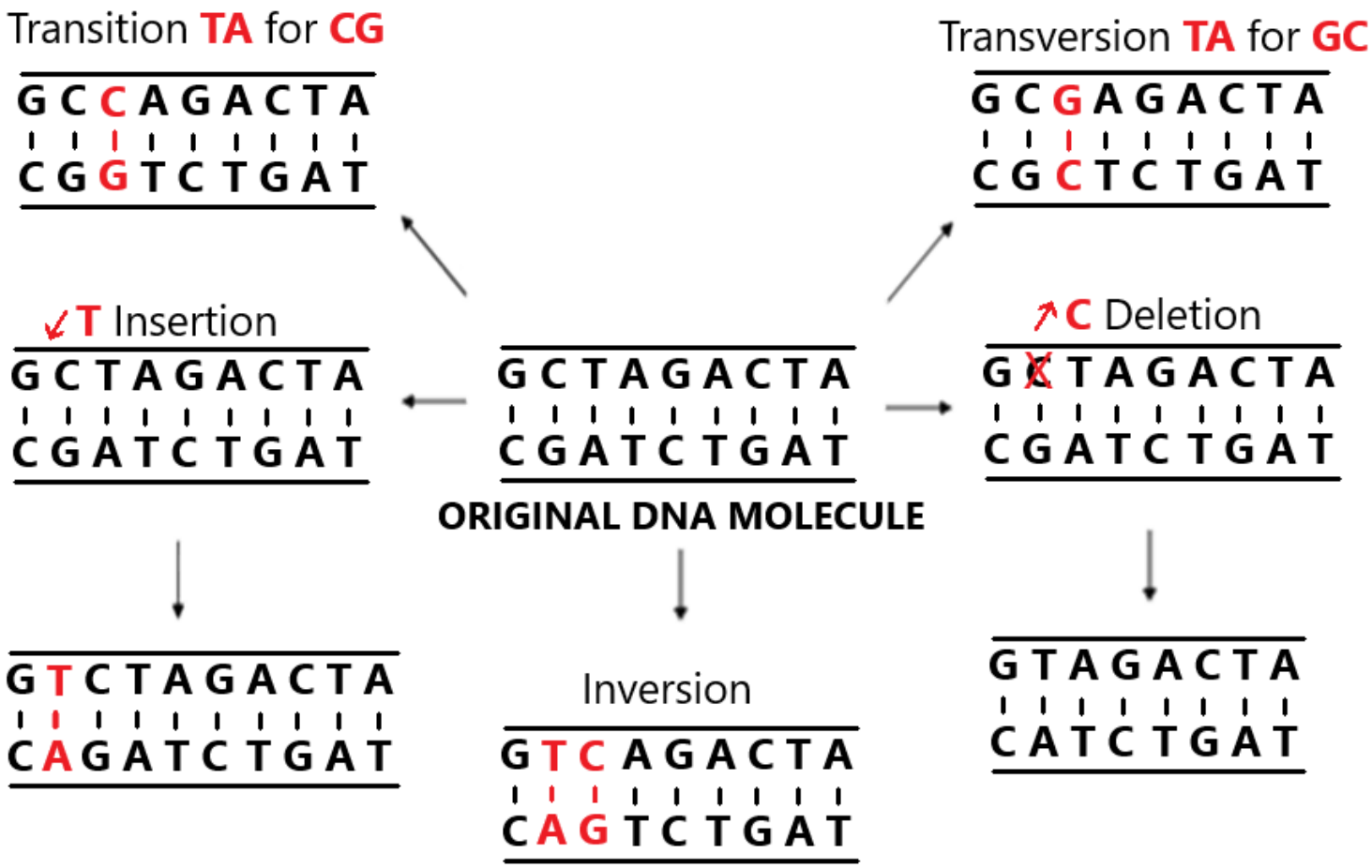
- **substitution** – a single nucleotide is substituted with (or exchanged for) a different nucleotide:
  - *transition mutation* – when a purine base is substituted for the other purine or one pyrimidine is substituted for the other pyrimidine
  - *transversion mutation* – when a purine replaces a pyrimidine (or vice versa)
- **inversion** – section of DNA is reversed
- **insertion** – add one or more extra nucleotides into the DNA
- **deletion** – remove one or more nucleotides from the DNA

Insertions and deletion can alter the reading frame of the gene.

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

Effect on DNA sequence



Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

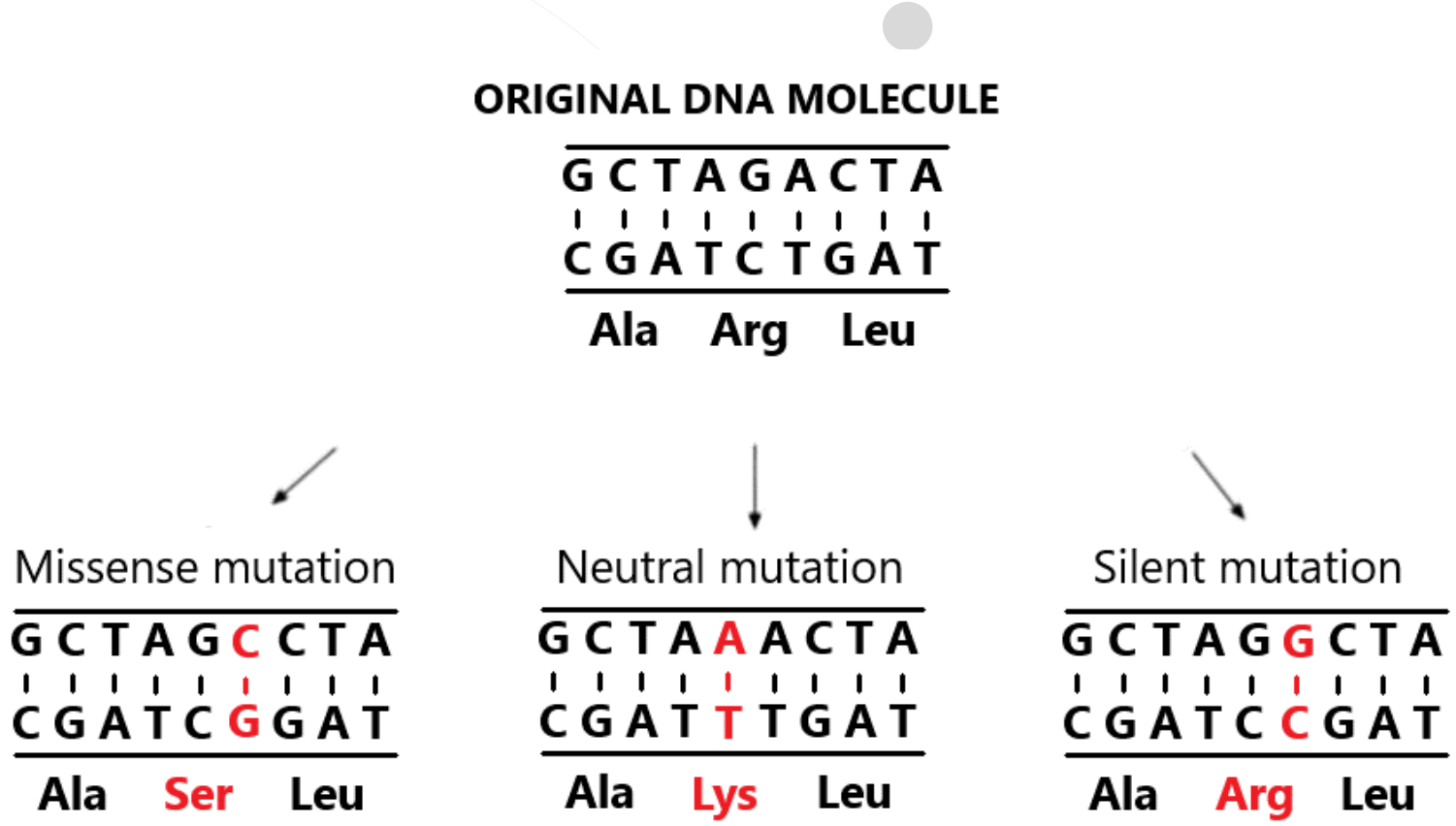
Effect on amino acid sequence

- ***missense mutations*** - a codon change causes a different amino acid to be inserted into the polypeptide chain
- ***neutral mutations*** - mutations cause one amino acid to be replaced with another amino acid with similar properties, so the change is less pronounced
- ***silent mutations*** - on the basis of the degeneracy of the genetic code, the inclusion of the same amino acid occurs despite a different codon

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

Effect on amino acid sequence



Erasmus+ project 2021-1-SK01-KA220-HED-000032068

Co-funded by the European Union

Effect on reading frame

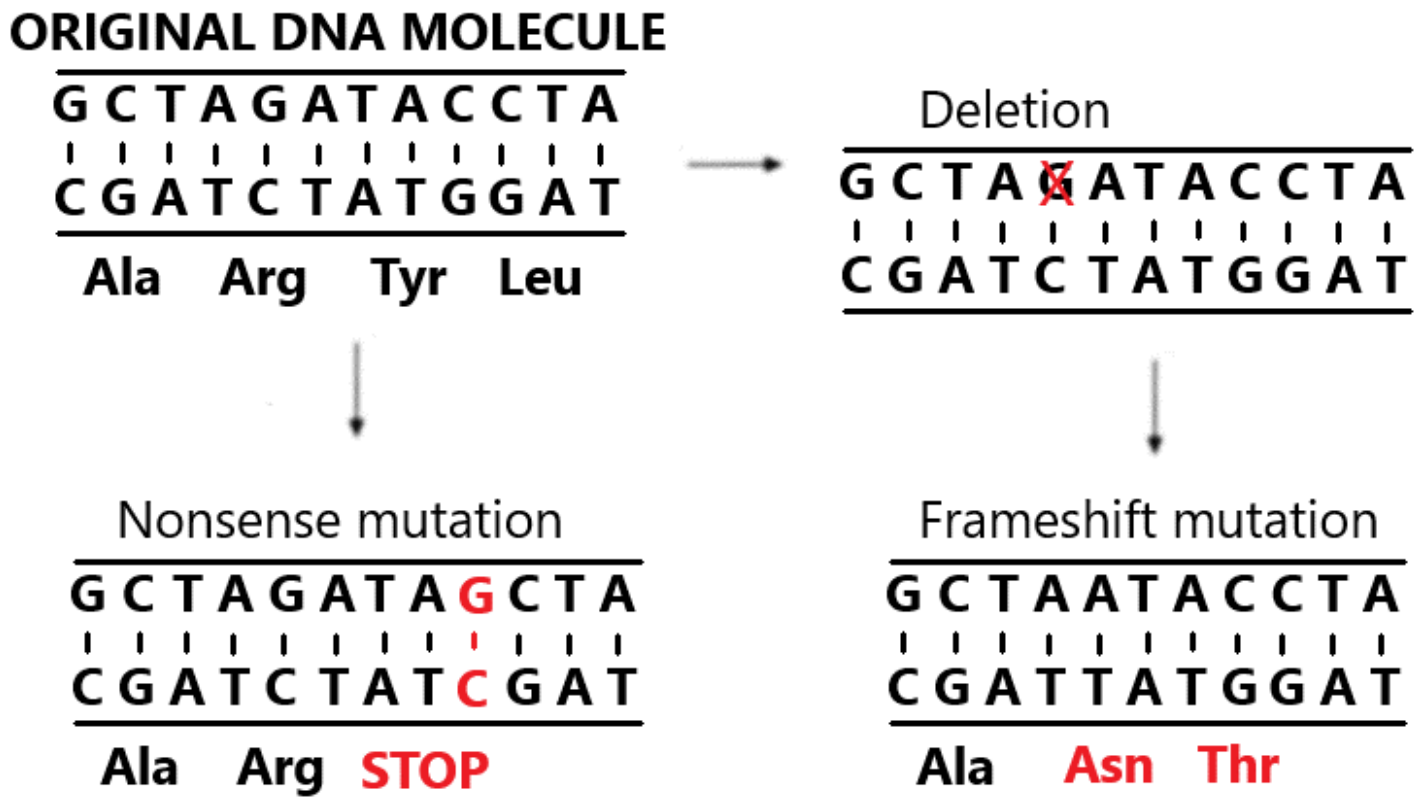
The **reading frame** is the way in which nucleotide sequences are interpreted into amino acid sequences.

- ***nonsense mutations*** - changing the codon for an amino acid to a STOP codon (UAG, UAA or UGA) causes premature termination of translation on the ribosome
- ***frameshift mutations*** - are a special type of mutations, the essence of which is the insertion or deletion of a number of nucleotides that is not a multiple of 3, leading to a shift in the reading of codons on the ribosome, resulting in the insertion of completely different amino acids from the site of the mutation

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

Effect on reading frame



Erasmus+ project 2021-1-SK01-KA220-HED-000032068

Co-funded by the European Union

Effect on function of the protein

- ***gain-of-function mutation*** **-** the mutated protein acquired new new properties, and therefore the phenotype of such a mutation is predominantly dominant

- ***loss-of-function mutation*** **-** the mutated protein is non-functional, but the second copy (normal allele) is usually sufficient to cover the needs of the organism, therefore this mutation is recessive.

- ***dominant negative mutation*** **-** the mutant allele has such a dominant phenotype that its product is antagonistic in expression to the product of the normal allele, resulting in loss of function despite the presence of one normal allele.

Erasmus+ project 2021-1-SK01-KA220-HED-000032068

ISAGREED
Co-funded by the European Union

ISAGREED

Partners:

Thank you for your attention!

This presentation has been supported by the Erasmus+ KA2 Cooperation Partnerships grant no. ***2021-1-SK01-KA220-HED-000032068 "Innovation of the structure and content of study programs in the field of animal genetic and food resources management with the use of digitalisation - Inovácia obsahu a štruktúry študijných programov v oblasti manažmentu živočíšnych genetických a potravinových zdrojov s využitím digitalizácie"****. The European Commission support for the production of this presentation does not constitute an endorsement of the contents which reflects the views only of the authors, and the Commission cannot be held responsible for any use which may be made of the information contained therein.*

Martina Miluchová

martina.miluchova@uniag.sk

Co-funded by the European Union