

## GENETICS

**Study Programme: MIMV    Curricular Year: 2<sup>nd</sup>    Semester: 4<sup>th</sup>    Compulsory    Credits: 5.0 ECTS**

**Lecturer(s):** António J. Freitas Duarte (CCP) and Victor Diogo Alves (R)

### 1. Contact hours:

**Lectures - 28 h, Practicals - 28 h, Total - 56 h**

### 2. Objectives:

Acquire the basic principles and methodologies on molecular, cyto, mendelian and population genetics. To know the mechanisms that determine the transmission of hereditary traits. To know the molecular mechanisms of genetic diseases, including cancer. To provide a working knowledge of genetics of disease, including the principles of population genetics and of a broad range of inherited disorders in animals and the fundamental breed genetic diseases. To develop competence in obtaining, recording, and interpreting patient history and pedigree information. To provide an understanding of the methodologies used in cytogenetic and molecular genetics. To know how to interpret the normal and aberrant cariotype of domestic species. Use of OMIA, OMIM and other bibliographic resources to present a monographic work related with genetic disease, gene therapy, genetic resistance, etc.

### 3. Programme:

**Theoretical:** Medicine and Genetics: Molecular basis of Normal and Pathological disorders. Molecular Genetics. Genetic Polymorphism. Genome Analysis. Genetic linkage analysis. Genealogy analysis. Cytogenetic basis of pathological disorders. Single gene disorders. Immunogenetics. Familial disorders not due to a single gene. Introduction to Population Genetics.  
**Practical:** Segregation and Genealogy analysis. Linkage Analysis. Probabilities and Evaluation of the carrier status in animal breeding. Paternity exclusion testing and animal identification. Cytogenetics. Population genetics. Seminars "Genetics and Disease".  
Monography on a genetically determined clinical disorder. The students must perform a genetic analysis of a clinical situation. Whenever possible, they must focus on the disorder's molecular mechanisms including its physiopathology. A Seminar is organized with oral presentations.

### 4. Bibliography:

Nicholas, F.W. (2010). *Introduction to Veterinary Genetics*, 3<sup>rd</sup> Ed., Wiley-Blackwell.

Nicholas, F.W. (1989). *Veterinary Genetics*. Oxford Univ. Press.

Korf, B. R. (2000). *Human Genetics - A Problem-Based Approach*. Blackwell Science, Malden, USA.

Griffiths, A.J.F. et al. (2020). *An Introduction to Genetic Analysis*. 12<sup>th</sup>. Ed., WH Freeman and Co, USA.

### 4. Assessment:

Written examination (80% of final classification, including theoretical and practical evaluation).

Monography (20%, evaluated in its written form and during its oral presentation).