Transgenesis and Gene Therapy: from the Animal to the Clinic

Code: 42891
ECTS Credits: 9

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Degree	Type	Year
Bioquímica, Biología Molecular y Biomedicina	OP	1

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Teachers

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Teaching groups languages

You can view this information at the end of this document.

Prerequisites

Graduate in the field of life sciences, for example:
Biology, Biochemistry, Biomedicine, Biotechnology, Pharmacy, Genetics, Medicine, Veterinary Medicine.

Objectives and Contextualisation

The student will gain insight into:

- Technologies used to generate transgenic animals overexpressing specific transgenes or mutant models with specific endogenous genes modified (knockout and knockin animal models).
- Application of the aforementioned technologies in biomedicine, biotechnology and livestock.

- Current legislation for animal experimentation
- Mouse anatomy and embryology in order to understand the embryonic development of organs and to analyze

morphological/anatomical abnormalities in genetically modified mouse models.
- *In vivo* and *ex vivo* gene therapy, including characteristics of the different types of vectors (viral and non-viral) used for gene transfer as well as their advantages and disadvantages, administration routes and applications of gene therapy in the treatment of hereditary and non-hereditary human diseases.

Learning Outcomes

1. CA11 (Competence) Draw on techniques for partially or wholly modifying living organisms to enhance pharmaceutical and biotechnological processes and products, or to develop new products in the field of biomedicine.
2. CA12 (Competence) Work as a team in the planning of transgenesis and gene therapy projects that address current challenges in the fields of biochemistry, molecular biology and biomedicine, demonstrating ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
3. CA12 (Competence) Work as a team in the planning of transgenesis and gene therapy projects that address current challenges in the fields of biochemistry, molecular biology and biomedicine, demonstrating ethical responsibility and respect for fundamental rights and duties, diversity and democratic values.
4. KA16 (Knowledge) Enumerate the molecular foundations of in vivo and ex vivo gene therapy in the field of biomedicine.
5. KA17 (Knowledge) Identify the anatomy and embryology of the mouse, as well as morphological alterations in genetically manipulated mice.
6. KA18 (Knowledge) Understand the different technologies for obtaining transgenic animals and for vivo and ex vivo gene therapy, as well as the ethical implications that this entails.
7. KA18 (Knowledge) Understand the different technologies for obtaining transgenic animals and for vivo and ex vivo gene therapy, as well as the ethical implications that this entails.
8. SA16 (Skill) Understand the different methodologies used to obtain transgenic animals through overexpression, blocking or modification of endogenous genes in a ubiquitous or tissue-specific manner.
9. SA17 (Skill) Appropriately use the different types of vectors used for gene transfer and therapy.
10. SA18 (Skill) Accurately analyse the molecular or physiological alterations of a transgenic animal, as well as the results of clinical gene therapy trials in humans in the field of biomedicine.

Content

PART 1. MOUSE MORPHOLOGICAL PHENOTYPING

By J. Ruberte, A. Carretero, M. Navarro and V. Nacher. Dept. Animal Health and Anatomy, UAB

1. Anatomic Terminology and Regions

2. Development and Placenta

3. Osteology

4. Arthrology and Miology

5. Cardiovascular System

6. Repiratory Apparatus

7. Digestive Apparatus

8. Urinary Organs

9. Male and Female Genital Organs

10. Nervous System

11. Visual Organ

12. Vestibulocochlear Organ

PART 2: TRANSGENIC ANIMALS AND GENE THERAPY

By F. Bosch and Verónica Jiménez.

Dept. Biochemistry & Molecular Biology, UAB

Part 2.1. Transgenic Animals:

1. Generation of transgenic animals by pronuclear microinjection.

2. Constitutive and conditional (tissue specific and/or inducible) Knockout/in animals.

3. Generation of *Knockout/in* animals by Genome Edition with *ZFNs, TALENs and CRISPR/Cas9*

4. Cloned animals by nuclear transfer. Aplications.

5. Consortia for genome mutagenesis and mouse phenotyping: Mouse Clinics.

6. Management of transgenic animal colonies. Current legislation on animal experimentation.

7. Aplications of transgenic animal technology in the study of diabetes, obesity, inherited diseases…

Part 2.2. Gene Therapy:

1. Introduction to the gene therapy field.

2. Characteristics of adenoviral vectors. Applications.

3. Characteristics of recombinant vectors derived from adenoassociated viruses. Applications in gene therapy for diabetes mellitus.

4. "Ex vivo" Gene Therapy: retroviral and lentiviral vectors. Applications.

5. Non-viral gene therapy. Applications.

6. Gene Therapy for Hereditary Diseases. Gene Therapy for Mucopolysaccharidosis (MPS).

7. *Invivo" genome editting*.

PART 3. INTRODUCTION AND DESIGN OF GENE THERAPY CLINICAL TRIALS FOR THE TREATMENT OF HUMAN DISEASES

By M. Chillon and A. Bosch, Dept. Biochemistry & Molecular Biology, UAB

Invited speaker: Ramon Alemany, Institut Català d'Oncologia (ICO)

1-Introduction to clinical trials. Factors to consider in the design of clinical trials of gene therapy. M Chillon

2-Development and production of vectors for clinical trials. M Chillon

3-Regulation on the use of Genetically Modified Organisms. Biosafety level and quality of production (GMP and GLP conditions). M Chillon

4-Adenoassociats vectors in clinical trials. Increased tissue specificity using pseudotyped AAV vectors and specific promoters. Immune response. A Bosch

5-Vectors derived from herpes simplex virus in clinical trials. A Bosch

6-Advantages and disadvantages of retroviral and lentiviral vectors in clinical trials. ABosch

7-Ongoing clinical trials for specific diseases: Haemophilia, ß-Thalassemia, Primary Immunodeficiencies, Cystic Fibrosis, Duchenne Muscular Dystrophy, lysosomal storage diseases, neurodegenerative diseases, blindness, cancer, etc. A Bosch, M Chillon

Activities and Methodology

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Lectures and lab practices	55	2.2	CA11, KA16, KA17, KA18, SA16, SA17, SA18, CA11
Type: Supervised			
Preparation of oral presentations and lab practices	44	1.76	CA11, KA16, KA17, KA18, SA16, SA17, SA18, CA11
Type: Autonomous			
Literature search and study for exams	120	4.8	CA11, CA12, KA16, KA17, KA18, SA16, SA17, SA18, CA11

Combination of lectures and laboratory practices and presentation of a project supervised by the teacher.

Theory 72%
Laboratory 11%
Supervised work 14%
Tutoring 3%

UAB Surveys

15 minutes of one class will be allocated for the response of the UAB institutional survey.

Annotation: Within the schedule set by the centre or degree programme, 15 minutes of one class will be reserved for students to evaluate their lecturers and their courses or modules through questionnaires.

Assessment

Continous Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Attendance and active participation in lectures	10%	0	0	CA11, KA16, KA17, KA18, SA16, SA17, SA18

Attendance to laboratory practices (part 1)	8%	0	0	KA17, SA18
Oral defence of selected papers (part 2)	32%	2	0.08	CA11, CA12, KA16, KA17, KA18, SA16, SA17, SA18
Theorical and practical tests (part 1 and 3)	50%	4	0.16	CA11, KA16, KA17, KA18, SA16, SA17, SA18

The evaluation of the module will be based on work done by students, attendance and class participation, practices, oral defence of a scientific paper and the grade of exams at the end of the course.

To be eligible for the retake process, the student should have been previously evaluated in a set of activities equaling at least two thirds of the final score of the course or module. Thus, the student will be graded as "No Avaluable" if the weighthin of all conducted evaluation activities is less than 67% of the final score.

Important: If plagiarism is detected in any of the works submitted, the student will fail the whole module.

This subject/module does not provide for the single evaluation system.

Bibliography

Biblography

Gene and Cell Therapy. Therapeutic and Strategies. 2nd Edition. Edited by Nancy Smith Templeton, 2000.
Molecular Medicine. Edited by R.J. Trent. 3rd Edition. Elsevier Academic Press. 2005.
DNA Pharmaceuticals. Formulation and Delivery in Gene Therapy.
DNA Vaccination and Immunotherapy. Martin Scheef. WiLey-VCH Verlay GmbH &Co.KgaA, 2005.
Gene Therapy Technologies, applications and regulations. From Laboratory to Clinic. Edited by Anthony Meager. John Wiley & Sons, LTD, 1999.
Gene Therapy. Therapeutic Mechanisms and Strategies. Edited by Nancy Smith Templeton, Danilo D Basic. Marcel Dekker, Inc, 2000.
Gene Therapy Protocols. 2nd Edition. Edited by Jeffrey R Morgan Humana Press, 2002.
Human Molecular Genetics 2. T Strachan & AP Read. John Wiley & Sons, Inc., 1999.
Molecular Biotechnology Principles and Applications of Recombinant DNA. Bernard R Glick and Jack J Pasternak. Washington ASM Press, 1994.
The anatomy of the laboratory mouse. M. J. Cook. Academic Press, 1965
A color atlas of sectional anatomy of the mouse. T. Iwaki, H Yamashita, T. Hayakawa. Braintree Scientific, Inc., 2001.
The atlas of mouse development. M. H. Kaufman. Academic Press, 1995.
Transgenic animals. Generation and use. L.M. Houdebine. Harwood Academic Publishers 1997.
Manipulating the mouse embryo. A laboratory manual. 3rd Edition. A Nagy, et al. Cold Spring Harbor Laboratory Press, 2003.
Mouse genetics and transgenics. A practical approach. Ed. IJ Jackson & CM Abbott. Oxford University Press, 2000.
Gene Targeting. A practical approach. 2nd Edition. Ed. AL Joyner. Oxford University Press, 2000.
Transgenesis Techniques. Principles and Protocols. Edited by: Alan R. Clarke. Humana Press. 2002. (2nd Edition).
Gene Knock-out Protocols. Edited by: Martin J. Tymms and Ismail Kola. Humana Press. 2001.
Embryonic Stem Cells. Methods and Protocols. Edited by: Kursad Turksen. Humana Press.2002.
HumanMolecular Genetics2. T. Strachan i A.P. Read. John Wiley & Sons, Inc., Publication. 1999.
Morphological Mouse Phenotyping: Anatomy, Histology and Imaging. J. Ruberte, A. Carretero and M. Navarro. Ed. Medica Panamericana, 2016.
X-Ray Annotation Mouse Atlas. J Ruberte et al.IMPC (Doctor Herriot SL), 2021.

Web links
Gene Therapy Clinical Trials Worldwide www.wiley.co.uk/genmed/clinical
Human Genome Project Information
www.ornl.gov/sci/techresources/human_genome/medicine/genetherapy.shtml
The anatomy of the laboratory mouse jaxmice.jax.org/library/notes/498.html
International Society for Transgenic Technologies
www.transtechsociety.org
Transgenesis en mamíferos
www.cnb.uam.es/~transimp/index2.html
EUMORPHIA
www.eumorphia.org
TBASE (The Transgenic/Targeted Mutation Database)
http://tbase.jax.org/
Database of Gene Knockouts
http://www.bioscience.org/knockout/knockhome.htm
BioMedNet Mouse Knockout Database
http://biomednet.com/db/mkmd

Specialized journals
Nature (www.nature.com)
Nature Medicine (www.nature.com/nm/)
Nature Biotechnology (www.nature.com/nbt/)
Nature Genetics (www.nature.com/ng/)
Proc. Natl. Acad. Sci. USA (www.pnas.org)
Journal Clinical Investigation (www.jci.org)
Cancer Gene Therapy (www.nature.com/cgt)
Current Gene Therapy (bentham.org/cgt)
Gene Therapy (www.nature.com/gt)
Gene Therapy & Molecular Biology www.gtmb.org/index_gtmb.html
Gene Therapy & Regulation www.vsppub.com/journals/jn-GenTheReg.html
Human Gene Therapy (www.liebertonline.com/loi/hum)
The Journal of Gene Medicine
www3.interscience.wiley.com/cgi-bin/jhome/10009391
Journal of Molecular Therapy
link.springer-ny.com/link/service/journals/00109
Journal of Controlled Release
www.sciencedirect.com/science/journal/01683659
Journalof Virology (jvi.asm.org)
Molecular Therapy www.sciencedirect.com/science/journal/15250016

Software

Not applicable

Groups and Languages

Please note that this information is provisional until 30 November 2025. You can check it through this link. To consult the language you will need to enter the CODE of the subject.

Name	Group	Language	Semester	Turn
(PLABm) Practical laboratories (master)	1	English	first semester	morning-mixed

(TEm) Theory (master)	1	English	first semester	morning-mixed