

# GENOMICS AND TRANSLATIONAL MEDICINE

## STUDY PLAN 2019-2020

**Coordinated by Dr Josep M Llovet** ICREA Professor, IDIBAPS, University of Barcelona. Director of the Liver Cancer Program, MSSM (New York). Professor of Medicine, Department of Medicine, Faculty of Medicine, University of Barcelona and **Dr Roser Pinyol**, Liver Cancer Translational Research Laboratory, BCLC Group, IDIBAPS - Hospital Clínic. Associated Professor, Department of Medicine, Universitat de Barcelona.

### GENERAL INFORMATION

Subject Name	Genomics and Translational Medicine
Code	573669
Type	Compulsory
Teaching	First semester
Coordinator	Prof. Josep M Llovet, Dr Roser Pinyol
Contact Details	jmllovet@clinic.cat, rpinyol@clinic.cat
ECTS credits	8

### OBJECTIVES

The purpose of this subject is to provide students with scientific, conceptual, methodological and practical knowledge about translational medicine. Specifically, the genomics and epigenomic bases will be assessed, as well as their applications in translational research. These concepts will be integrated with the study of the signaling pathways involved in various diseases and their functional role.

The overall objective is to provide a scientific basis for the design and implementation of translational research, and the knowledge related to relevant methods, techniques and applications in biomedicine.

### COMPETENCES TO BE GAINED DURING THE STUDY

#### Generic

G1: Capacity for learning and responsibility (capacity for analysis and synthesis, to adopt global perspectives and to apply the knowledge acquired/capacity to take decisions and adapt to new situations).

G2: Learning skills that are necessary to undertake further research studies with a high degree of autonomy.

#### Specific

S1: Understand the basic, clinical and therapeutic principles of different pathologies

S2: Learn the procedures and methodologies used in translational studies

S3: Become familiar with the development of biomedical research and learn the basic tools for translational research

S4: Ability to explain the basic molecular principles underlying pathologies, ability to understand the role of genes in human cancer and the basic concepts of oncogenes and targeted therapies.

## THEMATIC BLOCKS

### 1. Basic Principles

- Introduction
- Role of epigenetics in human diseases
- Personalized medicine in oncology
- Molecular pathology in oncology
- Principles of genetic engineering
- Immunology and cancer
- Principles of experimental Design
- Bioinformatics and Managing of Big Data

### 2. High throughput technologies

- Gene expression
- SNP array, CNVs and GWAS
- Exome sequencing
- Methylome analysis
- Single Cell Genomics
- Proteomics

### 3. Signaling pathways

- Signaling pathways.
- Resistance to molecular therapies
- TGF-beta signaling in liver cancer

### 4. Genomics in cancer

- Molecular classification of hepatocellular carcinoma
- Targets for therapies in pancreatic cancer
- miRNA in human diseases and digestive cancer
- Angiogenesis: Drugs & mechanisms of resistance
- Colorectal cancer: genetics and genomics
- Molecular classification of breast cancer
- Molecular therapies and immunotherapy in melanoma
- Immunotherapies in cancer
- Role of adult stem cells in cancer

### 5. Genomics in other diseases

- Next generation sequencing in hematological diseases
- Pluripotent cells in translational medicine: recent advances and open problems
- Chronic Hepatitis C: from genotyping to therapies
- Liver portal hypertension and fibrosis
- Inflammatory disease / Crohn's disease
- Inflammatory bowel disease (IBD)
- Translational medicine in Alzheimer disease
- Translational medicine in Parkinson disease
- Translational research in psychiatric disorders
- Genomics in Multiple Sclerosis
- Genomics in autoimmune encephalitis.
- Genomics in autoimmune diseases
- Endocrinology
- Translational medicine in renal diseases
- Fetal and perinatal translational medicine
- Translational research in cardiovascular diseases
- System biology in lung diseases

### 6. Trial design and Biomarkers

- Innovation in translational medicine
- Translational medicine: implications in trial design
- Statistical principles for clinical trials
- Trial design and innovation.
- From Bench to Spin off

## METHODOLOGY

Total training hours: 8 credits ECTS x 25h/credit = 200h

- a) Face-to-face training (72h): Lectures and Seminars
- b) Home training (128h): Individual and group work

## EVALUATION

**Evaluation criteria:** 50% of the final score will depend on the attendance and active participation in class. The remaining 50% will be obtained through a written exam. The written exam will be based on a multiple option test. To pass the subject, students will have to fulfill three requisites: Attendance-score  $\geq 20/50$ , exam-score  $\geq 20/50$ , and overall score (attendance + exam)  $\geq 50/100$ .

**Reevaluation:** In case of failing the ordinary evaluation (overall-score  $\leq 50/100$ ), students that have a minimum of 1/3 of the exam questions correct will have the chance to be re-evaluated. For that, they will need to present a

critical appraisal of 3 scientific articles in front of an evaluation committee. The re-evaluation final score will never get over 50 points.

## REFERENCES

### Books

- Handbook of Translational Medicine. Edicions de la Universitat de Barcelona, Barcelona-B-16.985-2016. ISBN: 978-84-475-4030-3. Ed: Josep M Llovet.
- Translational Medicine: The Future of Therapy?  
Autors: James Mittra and Christopher-Paul Milne  
Data: Apr 17, 2013
- Genomic and Personalized Medicine, Second Edition: V1-2  
Autors: Geoffrey S. Ginsburg and Huntington F Willard PhD  
Data: Nov 29, 2012
- Translational Medicine and Drug Discovery  
Autors: Bruce H. Littman MD and Rajesh Krishna PhD FCP  
Data: Oct 15, 2014

### Articles

- Albani S, Prakken B. The advancement of translational medicine-from regional challenges to global solutions. *Nat Med.* 2009;15:1006-9.
- Berger B, Peng J, Singh M. Computational solutions for omics data. *Nat Rev Genet.* 2013;14(5):333-46.
- Garraway LA, Lander ES. Lessons from the cancer genome. *Cell.* 2013;153(1):17-37.
- Hanahan D, Weinberg RA. Hallmarks of cancer: the next generation. *Cell.* 2011;144(5):646-74.
- Heyn H, Esteller M. DNA methylation profiling in the clinic: applications and challenges. *Nat Rev Genet.* 2012;13(10):679-92
- McGranahan N, Swanton C. Biological and therapeutic impact of intratumor heterogeneity in cancer evolution. *Cancer Cell.* 2015;27(1):15-26.
- Pardoll DM. The blockade of immune checkpoints in cancer immunotherapy. *Nat Rev Cancer.* 2012;12(4):252-64
- Rezza A, Sennett R, Rendl M. et al. Adult stem cell niches: cellular and molecular components. *Curr Top Dev Biol.* 2014;107:333-72.
- Schulze K, Imbeaud S, Letouzé E, Alexandrov LB, Calderaro J, Rebouissou S, et al. Exome sequencing of hepatocellular carcinomas identifies new mutational signatures and potential therapeutic targets. *Nat Genet.* 2015;47(5):505-11
- Sia D, Hoshida Y, Villanueva A, Roayaie S, Ferrer J, Tabak B, et al. Integrative molecular analysis of intrahepatic cholangiocarcinoma reveals 2 classes that have different outcomes. *Gastroenterology.* 2013;144(4):829-40.
- Vogelstein B, Papadopoulos N, Velculescu VE, Zhou S, Diaz LA Jr, Kinzler KW. Cancer genome landscapes. *Science.* 2013;339(6127):1546-58.
- Wan L, Pantel K, Kang Y. Tumor metastasis: moving new biological insights into the clinic. *Nat Med.* 2013;19:1450-64.