MOLECULAR & GENETIC DIAGNOSTICS

ACADEMIC YEAR 2017 2018 SYLLABUS

Welcome to Molecular & Genetic Diagnostics!

Please, remember that it is very important to read and understand the syllabus. It is the best source of information about the subject and it contains all the information you need to follow the course over the term.

Remember that it's your responsibility to read and understand all the material on the syllabus.

1. SUBJECT DESCRIPTION

Degree:	Biotechnology
Course:	Molecular & Genetic Diagnostics
Area:	Genetics
Department:	Molecular Biology & Biochemical Engineering
Academic Year:	2017-18
Term:	First Semester
Total Credits:	6
Year:	3rd year
Type of Course:	Optative Subject
Course Language:	English

Teaching Model	
a. Lectures (General Background):	70%
b. Labs and experimental procedures:	15%
c. Guided Academic Activities:	15%

2. SUBJECT COORDINATOR

Name:	Nicolás Prados Dodd
Course:	Experimental Sciences
Area:	Genetics
Department:	Molecular Biology & Biochemical Engineering
Category:	Collegiate Assistant Professor (Profesor Asociado)
Tutorials:	Mondays-Tuesdays 14:00 to 15:00 (confirm in advance)
	It is possible to arrange different time or day.
	Online through the "Aula Virtual"
Office:	Building 22, 2nd floor, office num 2
e-mail:	ngpradod@upo.es
Telephone:	n/a

3. GENERAL INFORMATION

3.1. Description and objectives

At the end of the course, the student will be able to:

- Appreciate the actual available molecular diagnostic tests
- Be able to assess scientific reports about molecular diagnostic tests
- Understand some basics about genetic counselling

3.2. Contributions to the training plan

Optative subjects are multidisciplinary, in this case, we have the statistics theory of a diagnostic test, medicine, molecular biology and engineering.

This subject allows the student to achieve a basic concept of Molecular Diagnostics which is one of the most important applications of modern biotechnology. Many big companies are investing millions of euros to develop cost-effective and better tests. The sequencing and interpretation of our genome for less than 1000€ is a medicine landmark that is going to be reached in the next years and will revolutionize modern medicine.

This subject lets the students acquire enough knowledge about this area to help them decide if they are interested to specialize in this area during their professional career.

3.3. Recommended or required prior knowledge

- General knowledge of Genetics, Molecular Biology and Biochemistry.
- Good English.
- Managing bibliography and computers. It will be essential to use the "Aula Virtual".

4. Skills

4.1. Degree skills developed in the course

- Understanding the scientific method
- Use of the scientific literature so you can perceive clearly recent advances and future developments
- Be able to develop the ability to acquire, interpret and analyse information related to molecular diagnostics.
- The capacity to solve problems related to similar aspects to the solutions provided during the subject.

4.2. Specific skills

- Understanding of the theory of diagnostic testing
- Recognize the uses and clinical applications of modern molecular diagnostics
- Search of the necessary information to do a genetic counselling or interpret a molecular test
- Ability to describe in clear language the implications of the result of a test to someone without a genetic or molecular background.
- Due to the multidisciplinary characteristics of optative subjects, many of the skills overlap with the general subjects of the degree described elsewhere.

5. Course contents

The Theory Topics are divided in Lessons. Depending on the number of modules they will take longer.

Lesson 1 Molecular Diagnostics of Infectious diseases

- 1A Theory of Diagnostic Tests
- 1B Quantifying DNA/RNA: PCR
- 1C Non PCR amplifications for quantification
- 4D Sequencing, next generation sequencing and next-next generation sequencing: applications in infectious diseases

Lesson 2 Cytogenetics

- 2A Karyotype
- 2B FISH
- 2C CGH
- 2D Sequencing

Lesson 3 DNA Profiling

Lesson 4 Mendelian Disorders

- 4A Pedigrees
- 4B Databases OMIM/GENETESTS
- 4C Strategies diagnosing molecular disorders
- 4D Genotyping molecular disorders
- 4E Linkage: indirect analysis of monogenic disorders

Lesson 5 Molecular testing of non-mendelian monogenic disorders

- 5A Mosaicism
- 5B Mitochondrial disorders
- 5C Imprinting disorders

Lesson 6 Complex disorders

Lesson 7 Cancer

6. Methodology and resources

The tandem "student-professor" through the "coincident work" represent the 30% of the total time scheduled for this subject. This 30% is distributed as follows: Basic Teachings (70%), Lab Sessions (15%) and Guided Activities (15%).

It is expected that the 60% of the time dedicated to a subject is student autonomous work. The last 10% is dedicated to evaluation tasks.

6.1. Basic Teachings

The course includes up to 24 hours of classes (on site) where we will develop the basics of the subject

For the development of the basic teachings assigned classrooms will be used, with up to 20 people and equipped with computer for video projection, allowing presentations with images and animations for the presentation of concepts. In addition, all classrooms have slates for the detailed explanation of those concepts requiring additional explanations or detail.

6.2. Lab Sessions

Throughout the course, part of theoretical knowledge will be developed in 3 lab sessions:

- Case report: Interpretation of a real case of molecular testing and genetic counselling. It will be supervised by Nicolás Prados and it will be instructed in a computer classroom.
- Molecular testing of cystic fibrosis.
- Visit of a clinical molecular testing facility and interpretation of arrays.

These two sessions will be imparted by an assigned professor and may be changed due to availability of the external facility. Hands-on lab sessions are conducted primarily in the student's laboratory of Genetics, equipped with the necessary instrumental to perform different practices. Laboratories also have audio visual, simulation tools and consumables necessary for practice.

6.3. Guided Activities (Homework)

Throughout the course different homework will be distributed to all students. Some will be common and other will be personalized to each student. They will have a time limit to be returned by the "Aula Virtual".

1. Virtual Lab at the Howard Hughes Institute: a virtual lab session of a immunoassay and a sequencing test
2. Wikipedia edition of terms related to Molecular Diagnostics
3. Newsroom: search and review of subject-related news in the media

4. Gene counselling case.

6.4. Virtual Classroom (Aula Virtual)

All information regarding the subject will be distributed through this tool. The student should visit this website regularly for further information, outlines, hand-outs, and links to other sites of interest

7. Assessment

7.1. Personal assessment

Since the attendance and participation are very important for the qualifications of this subject, the possibility of knowing who is who from the beginning is necessary for the normal development of the course.

For that reason and in order to make possible the continuous assessment, the students are kindly requested to provide an ID-photo at the beginning of the semester. You can post your picture directly through the virtual classroom in your profile or, if you prefer, you can send it to your professor by any other means if you do not want to share your picture.

You have to understand that without this tool it would not be possible to take into consideration your public contributions to the course.

7.1. General evaluation

The different activities of the course will generate points. All points are added to calculate the final score:

- “Sobresaliente”: 90 or more points
- “Notable”: 70 or more points
- “Aprobado”: 50 or more points

Matrícula de Honor: 1 will be distributed to the student with more points.

It is possible to get more than 100 points:

- Content assessment: 90 points max
- Homework assessment: 20 points max
- Lab Sessions: 15 points max

IMPORTANT: THERE WILL BE NO FINAL EXAM IN FEBRUARY.

If a student does not reach 50 points, there will be a final exam in July where all the content of the subject will be evaluated.

There will be no partial exams to increase the

7.2. Content Assessment

Focussing on the continuous evaluation of the student, there will be six assessments of the Basic Content. They will consist on an Assignment on the Virtual Classroom. They can be done at the University facilities or at home. There is no restriction in the use of any available resource. There will be 25 hours to finish the assignment and it will be scheduled from Thursday 15:00 hours to Friday 15:00 hours. The assignment will be scheduled the following week after the end of the lesson evaluated. There will be no exceptions unless it is a holiday (it would be moved to the next week). The assignment is designed to be completed in one hour, the virtual classroom will provide a two hours' window to finish the assignment once started. The student is responsible to have a working computer and internet connection. The assignment should be done alone. Plagiarism is not allowed, but, as it is not possible to control if the assignment is done alone, in groups or copying previous answers, suspicions of plagiarism will be under marked. Each assignment will be scored up to 15 points.

These assignments substitute the final exam in February. There will be no partial exams to increase the final score. If a student wants to improve his mark, he must renounce the actual score and sit the July exam.

7.3. Homework assessment

There will be up to four student works proposed to the students. Some works will be general (the same for all the students) and other will be personalized. Examples of this work will be:

- HHI Virtual Lab
- Wikipedia edition
- Gene counselling case
- News comments

Each homework will be scored up to 5 points.

7.4. Lab Session assessment

Each session will be evaluated depending on the attendance to the session and/or a test depending on the characteristics of each one. They will be scored up to 5 points. The details will be advanced in each case.

7.5. Extra points

In some situations, extra points may be awarded (up to 5 points):

- Active participation in class or in the Virtual Classroom
- Outstanding answers in the assignments
- Exceptional homework. Note that clarity and concision is awarded. Extensive work of thousands of words will be under scored.

8. Literature

Because it is a very fast developing field no general text is recommended as they are all outdated. During the development of the course specific reviews, articles or websites will be described for tracking specific topics.

The following list is just an example.

- Trent, R.J. "Molecular Medicine". 3a ed. Elsevier, 2005
- Serre, J.L. "Diagnostic Techniques in Genetics". John Wiley & Sons, 2006
- Buckingham, L y Flaws, M.L. "Molecular Diagnostics: Fundamentals, Methods and Clinical Applications" F. A. Davis Company, 2007

Bibliografía complementaria (Monografías)

- Bradley, J., Johnson, D. y Pober, B. "Medical genetics" 3a ed. Blackwell, 2006
- Day, I.N.M., Humphries, S.E. "Genetics of Common Diseases. Future therapeutic and diagnostic possibilities". BIOS Scientific Publishers. Oxford, 2002
- Elles, R. y Mountford, R. "Molecular Diagnosis of Genetic Diseases". Humana Press, 2a ed., 2002
- "Emery and Rimoin's Principles and Practice in Medical Genetics". Churchill Livingstone, 5a ed., 2006
- Jeffery, S., Booth, J. y Myint, S."Molecular Diagnosis". BIOS Scientific Publishers, 2000
- Jorde, L.B., Carey, J.C., Bamshad, M.J. y White, R.L. "Genética Médica". Elsevier- Mosby, 3a ed., 2004
- Killeen, A.A. "Principles of Molecular Pathology". Humana Press, Totowa, New Jersey, 2004
- Korf, B. "Human genetics and genomics". Blackwell Publishing, 2006
- Kumar, D. "Genomics and clinical medicine". Oxford University Press Inc, USA, 2007
- Leonard, D.G.B. "Diagnostic molecular pathology". Saunders, 2003
- McKinlay Gardner, R.J. y Sutherland, G.R. "Chromosome Abnormalities and Genetic Counseling". Oxford University Press, 3a ed., 2003
- Nussbaum, R.L., McInnes, R.R., Williard H.F. "Genética en Medicina". Thompson and Thompson 5a ed. Ed Masson, 2004
- Pasternak, J.K. "An introduction to human molecular genetics: mechanisms of inherited diseases". Recurso electrónico, 2005
- Patrinos, G.P. y Ansorge, W. "Molecular Diagnostics". Elsevier, 2005
- Ross, D.W. "Introduction to molecular medicine". Springer, cop. 3a ed., 2002
- Sanjurjo, P. y Baldellou, A. "Diagnóstico y tratamiento de las enfermedades metabólicas hereditarias". Ergón, 2006
- Sudbery, P. "Genética molecular humana". Pearson Educación, 2a ed., 2004.
- Young, I.D. "Medical Genetics". Oxford University Press, 2005