

**Medical Genetics**

Code: 101970  
ECTS Credits: 6

Degree	Type	Year	Semester
2500890 Genetics	OB	3	2

**Contact**

Name: Immaculada Ponsa Arjona  
Email: Imma.Ponsa@uab.cat

**Use of Languages**

Principal working language: catalan (cat)  
Some groups entirely in English: No  
Some groups entirely in Catalan: Yes  
Some groups entirely in Spanish: No

**Teachers**

Rosa Miró Ametller  
Vicenç Català Cahís  
Maria Angels Rigola Tor

**Prerequisites**

It would be appropriate to have passed and achieved the competences corresponding to the subjects: "Cell Biology and histology", "Human Genetics", "Cytogenetics" and "Genetics and Reproduction".

**Objectives and Contextualisation**

The main objectives are:

Know the genetic basis of the main diseases with a base or genetic component.  
Relate the genetic dysfunction with the pathological phenotype.  
Perform the genetic interpretation of the diagnosis, prognosis, prevention and therapy of the most frequent genetic pathologies in the human population.  
Understand the distribution of genetic-based diseases in a population taking into account their origin.  
Analyze genetically the probands-family relationship that facilitates the offer of a genetic counseling.

**Competences**

- Apply knowledge of theory to practice.
- Appreciate the importance of quality and a job well done.
- Assume ethical commitment
- Be able to analyse and synthesise.
- Be sensitive to environmental, health and social matters.
- Describe epigenetic mechanisms.
- Describe the genetic bases of the development and control of genic expression.
- Describe the organisation, evolution, inter-individual variation and expression of the human genome.

- Design and interpret studies associating genetic polymorphisms and phenotypical characters to identify genetic variants that affect the phenotype, including those associated to pathologies and those that confer susceptibility to human illnesses or those of other species of interest.
- Develop analysis, synthesis and communication strategies to transmit the different aspects of genetics in educational settings.
- Measure and interpret the genetic variation in and between populations from a clinical, conservational and evolutionary perspective, and from that of the genetic improvement of animals and plants.
- Perceive the strategic, industrial and economic importance of genetics and genomics to life sciences, health and society.
- Perform genetic diagnoses and assessments and consider the ethical and legal dilemmas.

## Learning Outcomes

1. Apply knowledge of theory to practice.
2. Appreciate the importance of quality and a job well done.
3. Assume ethical commitment
4. Be able to analyse and synthesise.
5. Be sensitive to environmental, health and social matters.
6. Describe the clinical consequences derived from epigenetic control mechanisms.
7. Describe the genetic and environmental causes of congenital defects.
8. Describe the role of genetic variation in the human species in the diagnoses, prevention and treatment of illnesses.
9. Describe the structure and variation of the human genome from a functional, clinical and evolutionary perspective.
10. Enumerate and describe the different techniques for analysing DNA polymorphisms that can be applied to studies of genetic variation associated to human pathologies.
11. Evaluate and interpret genetic variation in human populations and from a clinical and evolutionary perspective.
12. Explain how knowledge of human genetic variation is applied to personalised medicine, pharmacogenomics and nutrigenomics.
13. Interpret scientific publications, and solve problems and example cases in the fields of human and cancer genetics.
14. Prepare communication proposals in educational settings regarding the importance of the transfer of genetic progress to clinical practice.
15. Recognise the strategic importance of genetic progress in the field of human health, especially applications of the genomic to personalised medicine, pharmacogenomics and nutrigenomics.

## Content

1. General principles
  - 1.1. Basic introduction to genetic diseases
  - 1.2. Predisposition or genetic susceptibility concept
  - 1.3. Genetic counseling
2. Chromosome aberrations
  - 2.1. Autosomal trisomies
  - 2.2. Alterations of sex chromosomes
  - 2.3. Chromosome microdeletions and microduplications
3. Neuromuscular diseases
  - 3.1. Muscular dystrophies: definition and classification
  - 3.2. Duchenne and Becker muscular dystrophies
  - 3.3. Other muscular dystrophies
  - 3.4. Myotonic dystrophy
  - 3.5. Spinal muscular atrophy

4. Mental and behavioural disorders
  - 4.1. Intellectual Disability
  - 4.2. Fragile X syndrome and associated diseases
  - 4.3. Rett syndrome
  - 4.4. Autism
  - 4.5. Schizophrenia
5. Central nervous system diseases
  - 5.1. Huntington's disease
  - 5.2. Alzheimer's disease
  - 5.3. Parkinson's
  - 5.4. Primary tumors of the central nervous system
6. Skeletal and connective tissue diseases
  - 6.1. Osteogenesis imperfecta
  - 6.2. Achondroplasia
  - 6.3. Marfan syndrome
  - 6.4. Sarcoma
7. Craniofacial diseases
  - 7.1. Craniosynostosis
8. Dermatological diseases
  - 8.1. Albinism
  - 8.2. Skin cancer
9. Ophthalmological diseases and deafness
  - 9.1. Color vision deficiency
  - 9.2. Deafness
10. Cardiovascular diseases
  - 10.1. Hypertension
11. Gastrointestinal diseases
  - 11.1. Celiac disease
  - 11.2. Colon cancer
12. Respiratory diseases
  - 12.1. Lung cancer
13. Metabolic diseases
  - 13.1. Alterations in glucose metabolism: lactose intolerance
  - 13.2. Alterations in lipid metabolism: Hypercholesterolemia
  - 13.3. Alterations in amino acid metabolism: Phenylketonuria
14. Hematological diseases
  - 14.1. Hemoglobinopathies
  - 14.2. Haemophilia
  - 14.3. Leukemia and Lymphoma
15. Urogenital diseases
  - 15.1. Polycystic kidney
  - 15.2. Kidney cancer
  - 15.3. Bladder cancer
  - 15.4. Prostate cancer
16. Endocrinological diseases
  - 16.1. Diabetes mellitus
  - 16.2. Breast cancer

## Methodology

Lectures: Systematic exhibition of the subject's programme, giving relevance to the most important concepts. The student acquires the basic scientific knowledge of the subject in theory classes, which will complement the personal study of the exposed themes.

Problem based learning (PBL): Students will work in small groups, under the teacher supervision, on specific problems during 3 sessions of 2 hours each one for each case, and a total of 2 cases. At the end of each case, the work will be exposed to the rest of the classmates.

In general, the platform for communication and material transfer used will be Moodle.

## Activities

Title	Hours	ECTS	Learning Outcomes
Type: Directed			
Lectures	34	1.36	3
Problem based learning (PBL)	12	0.48	1, 3, 5, 4, 2
Type: Supervised			
Face-to-face and virtual tutorials	6	0.24	1, 3
Work production	18	0.72	1, 3, 5, 4, 2
Type: Autonomous			
Documentation search	17	0.68	
Evaluation	6	0.24	
Reading of texts	19	0.76	4
Study	38	1.52	4

## Assessment

The competences of this subject will be evaluated through: exams, group work and public presentations.

The evaluation system is organized in two modules, each of which will have a specific weight assigned in the final qualification:

- Problem Based Learning Module (ABP) (25%). The aspects that will be taken into account for the qualification will be: the interest and the quality of the work demonstrated throughout the development of the case for each one of the students and the group, and the final presentation. In the case of repeating students, if the qualification of this learning evidence in previous courses was equal to or greater than 5, they can renounce to repeat the activity and this qualification will be used to calculate the global grade of the subject. Due to the characteristics of the activity, this learning evidence is not recoverable.
- Written test module (75%). There will be two exams, each one corresponding to a half of the subject taught in lecture sessions. Exams will be test type with four multiple-choice options.

Evaluation activities	Recoverable	Value
1st part	Si	37,5%
2nd part	Si	37,5%
PBL case 1	No	12,5%
PBL case 2	No	12,5
Total*		100%

\*Requirements to calculate the global qualification

The weighted average of the scores will be applied based on qualifications equal to or greater than 4 in each of the partial exams.

In order to pass the subject, it will be necessary to obtain a global qualification equal to or greater than 5.

#### Recovery exam / retake process

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

The partial exams can be recovered when the qualification of the exam has been less than 4. The student can do the recovery exam only of the partial suspended.

If the student has to recover both partial exams, the score obtained corresponds to 75% of the qualification of the subject, disappearing the requirement of having to obtain a score greater than or equal to 4 of each of the partial exams. This score will be used to calculate the global qualification along with the other evaluation activities.

Students who wish to improve the qualification of one or both partial exams may do it in the recovery exam, previously renouncing to the qualification obtained in the corresponding partial exam.

#### Copy and plagiarism

All forms of plagiarism in any evaluation activity and/or copying in an exam are reasons for being awarded an immediate suspend the subject.

### Assessment Activities

Title	Weighting	Hours	ECTS	Learning Outcomes
Exams	75%	0	0	11, 8, 9, 7, 6, 12, 15
Problem based learning (PBL)	25%	0	0	1, 3, 5, 10, 13, 14, 4, 2

## Bibliography

- Bain, Barbara J. Haemoglobinopathy Diagnosis. 2n ed. Editorial Blackwell Science, 2006
- Donalson P, Daly A, Ermini L i Bevitt D. Genetic of complex disease. Editorial Garland Science, 2016
- Epstein R.J. Human Molecular Biology. Editorial Cambridge University Press, 2003
- Farreras-Rozman. Medicina interna. Editorial Elsevier, Vol, 1 i 2, 2009
- Firth H.V. i Hurst J.A. Oxford Desk Reference Clinical Genetics. Editorial Oxford University Press, 2005
- Gardner R.J.M. i Sutherland G.R. Chromosome abnormalities and Genetic Counseling, 5a ed. Oxford Monographs on Medical Genetics. Editorial Oxford University Press, 2011
- Harper P. S. Practical genetic counselling, 7a ed. Editorial Hodder Arnold, 2010
- Jorde L.B., Carey J.C. i Bamshad M.J. Genética Médica, 5a ed. Elsevier, 2016
- Korf B.R. i Irons M.B. Human Genetics and Genomics, 4ª ed, Editorial Wiley-Blackwell, 2013
- Klug V.S. et al. Conceptos de Genética, 10ª ed, Editorial Pearson, 2013
- Lee, R.J., Abramson, J.S I Goldson, R.A. Case studies in cancer. Ed. W. W. Norton & Company. 2019
- Nussbaum R. L., McInnes R.R. i Willard H.F. Thompson and Thompson Genética en Medicina, 8a ed. Editorial Elsevier, 2016
- Read A. i Donnai D. New Clinical Genetics. Editorial Scion Publishing Ltd, 2011
- Rimoin D.L. Emery and Rimoin's principles and practice of medical genetics, 5a ed. Editorial Churchill Livingstone. Vol 2-3, 2006
- Rooney D.E. i Czepulkowski B.H. Human cytogenetics : constitutional analysis : a practical approach. 3a ed. Oxford University Press 2001
- Rooney D.E. i Czepulkowski B.H. Human cytogenetics : malignancy and acquired abnormalities : a practical approach. 3a ed. Oxford University Press 2001
- Salvador J. i Carrera JM. Síndromes congénitos malformativos. Colección de Medicina Materno-Fetal. Editorial Masson, 1995
- Scriver Ch.R., et al. The Metabolic & molecular bases of inherited disease, 8th ed. Editorial MacGraw-Hill, NewYork, 2001
- Stracher T., et al. Genetics and Genomics in Medicine, 1st ed. Garland Science, 2014
- Weinberg, R.A. The Biology of Cancer. Garland Science, 2a ed. (2014)
- Wright A. i Hastie N. Genes and common diseases: Genetics in Modern Medicine. Editorial Cambridge University Press, 2007