

DISCIPLINE FILE
 Academic year 2026–2027

1. Data about the programme

1.1.	“CAROL DAVILA” UNIVERSITY OF MEDICINE AND PHARMACY
1.2.	FACULTY OF MEDICINE
1.3.	DEPARTMENT 7 PEDIATRICS
1.4.	DISCIPLINE: MEDICAL GENETICS
1.5.	DOMAIN OF STUDY: HEALTH – Sectorally regulated within the European Union
1.6.	STUDY CYCLE: LICENCE
1.7.	STUDY PROGRAMME: MEDICINE – ENGLISH MODULE

2. Data about the discipline

2.1.	Name of the discipline in the educational plan: Genetics				
2.2.	Discipline code: DF I 6S2M				
2.3.	Discipline type (FD/SD/CD): FD				
2.4.	Discipline regimen (MD/OPD): MD				
2.5.	The holder of the course activities: Professor Dragos Cretoiu Associate Professor Viorica Elena Radoi Lecturer Radu Ioan Ursu Lecturer Ina Ofelia Focsa				
2.6.	The holder of the seminar activities: Professor Dragos Cretoiu Associate Professor Viorica Elena Radoi Lecturer Radu Ioan Ursu Lecturer Ina Ofelia Focsa Assistant Professor Rodica Mirela Dragotoiu Assistant Professor Cerasela Diana Paraschiv Assistant Professor Laura Elena Dordea-Leonte Assistant Professor Florina Raicu Assistant Professor Traian Grozescu Assistant Professor Ionut-Grigore Basaraba				
2.7. Year of study	I	2.8. Semester	II	2.9. Type of evaluation (E/C)	E

3. Total estimated time (hours/semester of didactic activity and self-preparation/study)

I. Academic training (teaching, practical application, assessment)						
3.1. Nr hours/week	3	From which:	3.2. lecture	1	3.3. seminar/ laboratory	2
3.4. Total hours of educational plan	42	From which:	3.5. lecture	14	3.6. seminar/ laboratory	28
Evaluation (number of hours): 2						
II. Self preparation/study						

Time allocation		hours
Study of course materials, textbooks, books, study of the recommended minimal bibliography		20
Additional research in the library, research through the internet		8
Performing specific activities for preparing projects, laboratories, elaborating reviews or other tasks		10
Preparation for presentations or assessments, preparation for the final examination		16
Tutoring		2
Other activities		2
3.7. Total individual study hours		58
3.9. Total hours per semester (3.4.+ 3.7.)	100	
3.10. Number of credits	4	

4. Preconditions (where applicable)

4.1. of curriculum	No mandatory prerequisites (year I discipline); correlated disciplines, taken in parallel or subsequently according to the curriculum: Biochemistry, Cell and Molecular Biology.
4.2. of competences	Basic knowledge of biology; use of computers and electronic resources; academic learning and communication skills.

5. Conditions (where applicable)

5.1. to conduct the lecture	Lecture hall with video projector and sound system; access to the faculty e-learning platform; lecture materials available on the discipline's learning management system (Google Classroom). Attendance at lectures is mandatory, under the conditions of the UMFCF Regulation on students' professional activity in force.
5.2. to conduct the seminar / laboratory	Attendance at practical classes is mandatory; excused absences, exemptions from make-up and the make-up/repetition of activities are governed by the UMFCF Regulation on students' professional activity in force. Room with projector/computer, microscopes; access to databases (OMIM, ClinVar, gnomAD, GeneReviews, HGNC) and to anonymised test reports (karyotype, CMA, NGS) for interpretation exercises; observance of ethics and data confidentiality.

6. Learning outcomes

Knowledge	Skills	Responsibility and autonomy
Explains the structure and organisation of the human nuclear and mitochondrial genome (DNA, RNA, chromatin,	- Takes a family history, constructs and interprets three-generation pedigrees and recognises inheritance patterns. - Calculates the recurrence risk for monogenic diseases, for common	Observes the ethical and deontological norms and the legislation on confidentiality, genetic data protection and informed consent in medical genetics.

chromosomes, genes) and the gene–protein–phenotype relationship. • Describes DNA replication and repair, gene expression and its regulation, including epigenetic mechanisms. • Classifies genomic variation and mutations (SNV, indel, CNV, structural variants, repeat expansions) and explains their functional consequences and genotype–phenotype correlations. • Characterises Mendelian and non-Mendelian inheritance patterns, multifactorial inheritance and the principles of population genetics. • Explains the principles of clinical cytogenetics, oncogenetics, developmental genetics and mitochondrial disease. • Describes the principles, indications and limitations of genetic testing methods (karyotype, FISH, CMA, PCR, Sanger sequencing, NGS – panel/WES/WGS) and of genetic screening (prenatal, newborn, carrier). • Explains the principles of pharmacogenomics, of therapies for genetic diseases and of personalised medicine.	chromosomal abnormalities and, empirically, for multifactorial diseases. • Interprets, at an introductory level, a karyotype (ISCN nomenclature) and a FISH/CMA/NGS test report, including ACMG pathogenicity classes and the meaning of variants of uncertain significance (VUS). • Formulates the indication for genetic consultation and testing and chooses the method appropriate to the clinical situation. • Integrates clinical, pedigree and laboratory data into basic diagnostic reasoning. • Uses electronic biomedical resources (OMIM, ClinVar, gnomAD, GeneReviews) and standardised terminology (ISCN, HGVS).	• Communicates genetic information clearly and empathetically, adapted to the patient's health literacy, and works effectively in a multidisciplinary team. • Recognises the limits of own competence and the situations that require referral to a medical geneticist. • Self-assesses performance, identifies knowledge gaps and plans continuing learning in a rapidly evolving field.
---	---	---

7. Course objectives (aligned with the learning outcomes)

7.1. General objective	To build a solid theoretical and practical foundation in medical genetics and genomics, needed to understand the molecular and hereditary mechanisms of disease, to recognise the patient with a genetic disorder and to integrate genetic testing, screening and genetic counselling into medical practice.
7.2. Specific objective	• O1. To explain the organisation and functioning of the human genome at the molecular, chromosomal and epigenetic level. • O2. To apply the patterns of inheritance and the concepts of penetrance, expressivity and heterogeneity to the estimation of familial risk. • O3. To recognise and interpret the common chromosomal abnormalities and copy-number variants, as well as their mechanisms of origin. • O4. To argue the choice of genetic testing methods (from karyotype to genome sequencing) in common clinical situations and to interpret a test report at an introductory level. • O5. To integrate concepts of oncogenetics, pharmacogenomics and personalised medicine into basic clinical reasoning. • O6. To identify the particular features of mitochondrial disease, of multifactorial diseases and of developmental abnormalities. • O7. To know the principles of prenatal, newborn and carrier screening and of genetic counselling. • O8. To demonstrate ethical behaviour and appropriate communication in situations with genetic implications.

8. Contents

8.1. Lecture	Teaching methods	Observations
Introduction to genetics	Lecture; discussion	Lecture 1
Structure of nucleic acids Structure of DNA and RNA and their differences; conformational types and non-canonical DNA forms – implications in pathology	Interactive lecture; guided questions; short case studies; video resources	Lecture 2
DNA replication and repair DNA damage repair mechanisms and associated disorders; the mitochondrial genome	Lecture; illustrative cases (genomic instability syndromes)	Lecture 3
Organisation of the human genome Chromatin, chromosomes, coding and non-coding genome; the gene – structure and function	Lecture; illustration with photomicrographs and diagrams	Lecture 4
Gene expression and epigenetics Transcription, RNA processing, translation, the genetic code; regulation of gene expression; DNA methylation, histone modifications, non-coding RNAs, genomic imprinting, X-chromosome inactivation	Lecture; analysis of diagrams; examples of epigenetic diseases	Lecture 5

Genomic variation and mutations SNV, indel, CNV and other structural variants, repeat expansions; mechanisms of origin; effects on transcript and protein (synonymous, missense, nonsense, frameshift and splicing variants); functional consequences (loss/gain of function, haploinsufficiency, dominant-negative effect); HGVS nomenclature	Lecture; applied items	Lecture 6
Methods of genetic and genomic analysis Karyotype, FISH, chromosomal microarray (CMA), PCR/QF-PCR/MLPA, Sanger sequencing, next-generation sequencing (NGS: gene panels, exome – WES, genome – WGS), long-read sequencing, methylation analysis; principles of interpretation (ACMG classes, VUS, secondary findings)	Lecture; demonstrations on public databases (OMIM, ClinVar, gnomAD)	Lecture 7
Monogenic and non-Mendelian inheritance Autosomal dominant, autosomal recessive, X-linked, Y-linked and mitochondrial inheritance (heteroplasmy, threshold effect); penetrance, expressivity, genetic heterogeneity, mosaicism, anticipation, uniparental disomy, genomic imprinting	Lecture; case studies with pedigrees	Lecture 8
Clinical cytogenetics Numerical and structural chromosomal abnormalities; microdeletion/microduplication syndromes (copy-number variants); karyotype–phenotype correlations	Lecture; presentation of anonymised laboratory reports	Lecture 9
Developmental genetics and dysmorphology Developmental and signalling genes; malformation, deformation, disruption, dysplasia; syndrome, sequence, association; teratogenesis; sex determination and disorders of sex development (DSD)	Lecture; guided discussion of dysmorphology cases	Lecture 10
Oncogenetics Proto-oncogenes, tumour suppressor genes, DNA repair genes; the multistep model and the two-hit hypothesis; hereditary cancer predisposition syndromes; somatic vs germline testing; targeted therapies	Lecture; case analysis	Lecture 11
Multifactorial inheritance and population genetics Common diseases of the adult – threshold model, heritability, empirical recurrence risk; Hardy-Weinberg equilibrium, founder effect, consanguinity; genetic ancestry vs race and ethnicity; genome-wide association studies (GWAS) and polygenic risk scores – limits of transferability between populations	Lecture; discussion of association studies	Lecture 12

Genetic screening and diagnosis Carrier screening; prenatal screening (first-trimester combined screening; non-invasive screening by analysis of cell-free DNA in maternal plasma – cfDNA/NIPT, noting the predominantly placental origin of the “fetal” fraction) and prenatal diagnosis (chorionic villus sampling, amniocentesis, prenatal CMA and exome); preimplantation genetic testing (PGT); newborn screening for inborn errors of metabolism (the phenylketonuria example: enzyme defect, biochemical confirmation, dietary therapy); predictive testing	Lecture; cases with critical discussion	Lecture 13
Genetic counselling, personalised medicine and ethics Genetic consultation and counselling – principles (deepened in LP 13); pharmacogenomics; principles of therapy of genetic diseases (dietary therapy, enzyme replacement therapy, antisense oligonucleotides, gene therapy, genome editing); ethical and legal aspects – principles (informed consent, secondary findings, testing of minors, genetic data protection; deepened in LP 11)	Lecture; counselling role-play	Lecture 14
Recent bibliography – minimum (mandatory): • The discipline's lecture materials (Google Classroom), updated yearly. • Cohn R, Scherer SW, Hamosh A, editors. Thompson & Thompson Genetics and Genomics in Medicine. 9th ed. Elsevier; 2023. ISBN 978-0323547628 – chapters corresponding to the lecture topics. • Turnpenny PD, Ellard S, Cleaver R. Emery's Elements of Medical Genetics and Genomics. 16th ed. Elsevier; 2021. ISBN 978-0702079665 – chapters corresponding to the lecture topics. Supplementary (optional) bibliography: • Jorde LB, Carey JC, Bamshad MJ. Medical Genetics and Genomics. 7th ed. Elsevier; 2026. ISBN 978-0443115165. • Strachan T, Read AP. Human Molecular Genetics. 5th ed. Garland Science; 2019. ISBN 978-0367002503. • Firth HV, Hurst JA. Oxford Desk Reference: Clinical Genetics and Genomics. 2nd ed. Oxford University Press; 2017. ISBN 978-0192519825. • Pyeritz RE, Korf BR, Grody WW, editors. Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics. 7th ed. (multi-volume series). Academic Press/Elsevier; 2018–2024. • Milunsky A, Milunsky JM, editors. Genetic Disorders and the Fetus: Diagnosis, Prevention and Treatment. 8th ed. Wiley-Blackwell; 2021. ISBN 978-1119676935. • Carey JC, Battaglia A, Viskochil D, Cassidy SB, editors. Cassidy and Allanson's Management of Genetic Syndromes. 4th ed. Wiley-Blackwell; 2021. ISBN 978-1119432678. Guidelines and nomenclature: • Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the ACMG and the AMP. Genet Med. 2015;17(5):405-424. • Riggs ER, Andersen EF, Cherry AM, et al. Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the ACMG and ClinGen. Genet Med. 2020;22(2):245-257. • Hastings R, Moore S, Chia N, editors. ISCN 2024: An International System for Human Cytogenomic Nomenclature. Cytogenet Genome Res. 2024;164(Suppl 1):1-224.		

• den Dunnen JT, Dalgleish R, Maglott DR, et al. HGVS recommendations for the description of sequence variants: 2016 update. Hum Mutat. 2016;37(6):564-569. Curricular references (international harmonisation): • Massingham LJ, Nuñez S, Bernstein JA, et al. 2022 Association of Professors of Human and Medical Genetics (APHMG) consensus-based update of the core competencies for undergraduate medical education in genetics and genomics. Genet Med. 2022;24(10):2167-2179. • Skirton H, Lewis C, Kent A, Coviello DA; Members of Eurogentest Unit 6 and ESHG Education Committee. Genetic education and the challenge of genomic medicine: development of core competences to support preparation of health professionals in Europe. Eur J Hum Genet. 2010;18(9):972-977. • Electronic resources: OMIM (omim.org); GeneReviews (ncbi.nlm.nih.gov/books/NBK1116); ClinVar; gnomAD; Orphanet.		
8.2. Laboratory/ practical lesson	Teaching methods	Observations
Meiosis; the cell cycle Crossing-over and genetic recombination, consequences of meiosis, nondisjunction	Interactive discussion	LP 1
Gametogenesis Spermatogenesis, oogenesis; abnormalities of female and male gametogenesis	Interactive discussion	LP 2
Sex chromatin and X-chromosome inactivation Exercises and mini-cases: number of Barr bodies as a function of the number of X chromosomes (46,XX; 45,X; 47,XXY; 47,XXX); mechanism and exceptions of inactivation; skewed inactivation and manifesting carriers of X-linked diseases; Y chromatin (F body – historical significance)	Item solving; mini-case	LP 3
The normal karyotype Image-based exercises: identification of chromosomes on a G-banded karyogram and assembly of the karyotype; steps of lymphocyte culture and slide preparation (demonstration); principles of FISH and of chromosomal microarray (CMA); writing a normal karyotype in ISCN 2024 nomenclature	Work on images and sample reports; portfolio: commented report	LP 4
Chromosomal abnormalities Exercises on karyograms and reports: identification of numerical (aneuploidies, polyploidies) and structural abnormalities (deletions, duplications, inversions, translocations, ring chromosomes, isochromosomes) and writing the ISCN formula; mosaicism; recognition of a copy-number variant (CNV) on a CMA report; calculation of the reproductive risk of balanced translocation carriers (segregation); recognition on photographs of the phenotype of the frequent chromosomal syndromes (Down, Turner, Klinefelter, Edwards, Patau, common microdeletions)	Work on images and sample reports (karyotype, FISH, CMA)	LP 5

The pedigree Exercises: standardised symbols; family history and construction of a three-generation pedigree from a simulated interview; identification of indicators for referral to genetic consultation	Case study; pedigree construction exercises	LP 6
Inheritance of normal and pathological traits Pedigree exercises: recognition of the criteria of autosomal dominant, autosomal recessive, X-linked, Y-linked and mitochondrial inheritance (including heteroplasmy and threshold effect); Mendel's laws applied to crosses and to the ABO/Rh blood groups; recurrence-risk calculation on cases (haemoglobinopathies, phenylketonuria, Duchenne muscular dystrophy)	Pedigree cases; risk calculation; mini-quiz	LP 7
Molecular genetics techniques I Demonstration and exercises: DNA extraction and quality control; principle and interpretation of a PCR reaction (conventional, real-time) and of electrophoresis; restriction enzymes; reading a QF-PCR (aneuploidies) and an MLPA (deletions/duplications) result; reading a Sanger electropherogram and annotating the variant (HGVS); indications and limitations	Demonstration (PCR workflow); interpretation of sample results	LP 8
Molecular genetics techniques II Presentation and exercises: the next-generation sequencing (NGS) workflow from sample to report (library, sequencing, alignment, variant calling and filtering); choosing the appropriate test on clinical scenarios (gene panel, exome/WES, genome/WGS, long-read sequencing, CMA, methylation analysis); searching a gene or variant in OMIM, ClinVar, gnomAD and GeneReviews	Interactive presentation; demonstrations on public databases	LP 9
Interpretation of genetic test reports Exercises on anonymised reports: karyotype (ISCN), CMA, QF-PCR/MLPA, Sanger and NGS sequencing; application of the ACMG pathogenicity classes; interpretation of variants of uncertain significance (VUS) and of secondary findings; negative or uninformative results and residual risk, in relation to the clinical indication and the limits of the method; formulating the answer to the patient and to the referring physician	Work on anonymised reports; cases; portfolio	LP 10
Bioethics in medical genetics Case analysis and debate: drafting and discussing an informed consent form for genomic testing; decisions on predictive testing of minors, incidental/secondary findings and the right not to know, confidentiality within the family, genetic data protection and non-discrimination; simulated genetic consultation (role-play) with feedback	Interactive discussion; role-play; cases	LP 11

Population genetics and multifactorial diseases – applications and risk calculation Exercises: calculation of the carrier frequency from disease incidence (Hardy-Weinberg, autosomal and X-linked) and of the risk for couples; effect of consanguinity on risk; application of empirical-risk tables to cases of common diseases and malformations; critical reading of a Manhattan plot (GWAS) and of a polygenic risk score or direct-to-consumer test report	Item solving; critical discussion	LP 12
Genetic counselling and prenatal diagnosis Cases: establishing the indication for genetic consultation and testing; risk calculation with Bayes' theorem (carrier of an X-linked disease, residual risk after a negative test); interpretation of a first-trimester combined screening and of a cfDNA/NIPT result (high risk vs diagnosis, detection rate, false positives) and choice of the diagnostic test (chorionic villus sampling or amniocentesis; prenatal CMA/exome); preimplantation genetic testing (PGT) and carrier-screening scenarios; a counselling case in hereditary cancer predisposition (BRCA or Lynch); communicating results	Cases; critical discussion	LP 13
Revision and practical examination	Test; feedback	LP 14
Recent bibliography – minimum (mandatory): • The discipline's practical-class materials (Google Classroom), updated yearly. • Cohn R, Scherer SW, Hamosh A, editors. Thompson & Thompson Genetics and Genomics in Medicine. 9th ed. Elsevier; 2023. ISBN 978-0323547628 – chapters corresponding to the practical-class topics. Supplementary (optional) bibliography: • Turnpenny PD, Ellard S, Cleaver R. Emery's Elements of Medical Genetics and Genomics. 16th ed. Elsevier; 2021. ISBN 978-0702079665. • Hastings R, Moore S, Chia N, editors. ISCN 2024: An International System for Human Cytogenomic Nomenclature. Cytogenet Genome Res. 2024;164(Suppl 1):1-224. • Richards S, et al. ACMG/AMP standards and guidelines for the interpretation of sequence variants. Genet Med. 2015;17(5):405-424; Riggs ER, et al. ACMG/ClinGen technical standards for constitutional copy-number variants. Genet Med. 2020;22(2):245-257. • Electronic resources: OMIM, GeneReviews, ClinVar, gnomAD, Orphanet.		

9. Evaluation

Activity type	9.1. Evaluation criteria	9.2. Evaluation methods	9.3. Percentage in the final grade
9.4. Lecture	Understanding of the fundamental concepts; their application to problems (inheritance patterns, recurrence risk, choice and interpretation of genetic tests); clinical integration of the notions; correctness of answers.	Written examination, 50 multiple-choice items, 50 minutes, centralised assessment.	85%
9.5. Seminar/ practical activity	Active participation; interpretation of karyograms, pedigrees and test reports; problem solving; quality of the portfolio (pedigree, karyotype/CMA/NGS report interpretation, mini case analyses); observance of ethics.	Short tests and formative assessment during the semester, without weight in the final grade; written practical examination in week 14 (LP 14): 30 multiple-choice items in 30 minutes, drawn from the same item bank for all series, graded with whole marks from 1 to 10.	15%
9.5.1. Individual project (if applicable)	Not applicable separately; integrated into the seminar portfolio.	—	—
9.6. Minimum performance standard			
• Fulfilment of the attendance obligations and, where applicable, of the make-up/repetition of practical activities, under the UMFCF Regulation in force, and submission of the practical-class portfolio. • Minimum grade 5 in the practical examination, a condition for sitting the written examination (one practical re-examination before the examination session for students with a grade below 5): interpretation of a simple pedigree; recognition of a normal karyotype and of the common numerical abnormalities; basic interpretation of a genetic test report. • Minimum grade 5 in the written examination. • Final grade = $0.85 \times \text{written examination grade} + 0.15 \times \text{practical examination grade}$, rounded to the nearest whole number.			

Date of completion:
16.10.2026

Signature of the course holder

Signature of the laboratory holder

Date of approval by the Department Council:

Signature of the Department Director