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<tr><td>Course title</td><td colspan="5">Molecular Basis of Cellular Function and Dysfunction</td></tr>
<tr><td>Course code</td><td colspan="5">GEMD-101</td></tr>
<tr><td>Course type</td><td colspan="5">Required</td></tr>
<tr><td>Level</td><td colspan="5">Undergraduate</td></tr>
<tr><td>Year / Semester</td><td colspan="5">Year 1, Semester 1</td></tr>
<tr><td>Teacher’s name</td><td colspan="5">Constantinos Voskarides</td></tr>
<tr><td rowspan="3">ECTS</td><td rowspan="3">13</td><td colspan="4">Teaching Periods per Week</td></tr>
<tr><td>Large Group Learning</td><td>Small Group Learning</td><td>Laboratories & Skills</td><td>Clinical Practice</td></tr>
<tr><td>8</td><td>6</td><td>2</td><td>4</td></tr>
<tr><td>Course purpose and objectives</td><td colspan="5">The aim of the course is to enable the students to develop an in-depth and well-rounded understanding of the molecular basis of cellular biology and the correlation between its disturbance and disease, as well as the potential for therapeutic intervention.
Overall, the student will, by the end of the course, be able to:
• Describe the function of cellular components and explain how dysfunction can cause disease
• Describe the structure and function of the human genome and interpret the significance of genomic variation in the aetiology of disease
• Explain the regulation of gene transcription and translation and outline how these processes can be disturbed
• Outline the process which converts a translated polypeptide into a functional protein and relate disruption to disease
• Describe the process of cell division and proliferation and explain its regulation
• Explain the molecular and cellular events that underlie the neoplastic process
• Outline the potential therapeutic uses of the different types of stem cells
• Describe the principles of gene therapy, discuss therapeutic strategies and outline their limitations
• Discuss the concept of development of novel and targeted therapies, tailored to individual patients</td></tr>
<tr><td>Learning outcomes</td><td colspan="5">At the end of the course the student will be able to:

Knowledge
1. Describe the basic structure of the human cell</td></tr>
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	2. Outline the structure and function of the cell membrane and cell junctions 3. Outline the basic structure and key functions of cellular organelles and the cytoskeleton 4. Discuss, with examples, the implications of dysfunction of cellular organelles 5. Outline the process of cell signalling and provide examples of how this process might be manipulated for therapeutic purposes 6. Outline the different types of cell death and discuss the role of programmed cell death 7. Describe the structure and function of telomeres and their role in the aging cell 8. Outline the metabolism of nucleotides and explain their role in the synthesis of DNA 9. Describe the structure of DNA and compare intragenic and extragenic DNA 10. Describe the basic structure of the human gene 11. Discuss the concept of genetic variation and discuss its role in the aetiology of disease 12. Describe the classification of genetic variants in terms of its effect on gene function and functional implications for the encoded polypeptide 13. Define the modes of inheritance of genetic disease 14. Describe the basic structure of the human chromosome 15. Describe structural variants of the human chromosome and outline their clinical relevance 16. Describe the PCR method and the basis of sequencing techniques such as Sanger and next generation sequencing and discuss clinical applications 17. Outline basic techniques for visualizing the human chromosomes and describe the basis of molecular cytogenetic techniques such as array comparative genomic hybridization and discuss clinical applications 18. Outline the process of transcription and discuss its regulation 19. Outline the role of transcriptional regulation in human development 20. Define epigenetic regulation and outline the different mechanisms 21. Discuss the role of non-coding RNAs in health and disease 22. Outline the process of RNA splicing and discuss how its disturbance can cause disease 23. Outline the process of translation and discuss its regulation 24. Outline post-translational modifications and discuss their significance and clinical correlates 25. Define the levels of protein structure and correlate with protein function providing examples 26. Briefly outline techniques to study the transcriptome and proteome 27. Describe mitotic cell division 28. Describe the cell cycle and its regulation 29. Discuss the dysregulation of the cell cycle and the clinical consequences 30. Outline the process of DNA replication

	31. List the potential errors during DNA replication and discuss their potential implication if they remain uncorrected 32. Outline how the cellular machinery identifies and corrects DNA errors 33. Discuss the clinical implications of genetic defects in the DNA-correcting machinery of the cell and the clinical implications of DNA transposition 34. Outline the key events in the neoplastic process 35. Distinguish between driver and passenger mutations 36. Define oncogenes and describe how they contribute to the development of neoplasia 37. Explain how the different types of tumour suppressor genes prevent the development of neoplasia 38. Define Knudson's two-hit hypothesis 39. Discuss the implications of germline mutations in genes that relate to the neoplastic process 40. Describe the clinical features of the main cancer predisposition syndromes (such as Lynch syndrome and BRCA-associated breast and ovarian cancer) and explain their genetic basis 41. Describe the significance of cancer cell cultures 42. Define gene therapy and outline the different types of methodological approaches 43. Briefly outline gene cloning and gene editing techniques and discuss their research and clinical applications 44. Outline current applications and limitations of gene therapy 45. Define stem cells and compare and contrast embryonic and somatic stem cells 46. Briefly outline the process of somatic stem cell reprogramming to induce pluripotency and discuss the applications of this technique 47. Describe how genetic disorders can be cured by stem cell and tissue engineering approaches 48. Outline how stem cell technology and precision gene therapy can be united to treat genetic diseases 49. Illustrate how targeted treatments can be tailored to the specific mutations causing genetic disease 50. Define personalised medicine 51. Discuss how genetic studies in populations can contribute to the management of common diseases *Skills* 52. Interpret a basic electropherogram 53. Use appropriate symbols to construct a genogram depicting the inheritance of genetic disease 54. Interpret the notation used in genetic reports and discuss the significance of results

	55. Explain the limitations of genomic risk profiling and the pitfalls of direct-to-consumer genetic testing *Professional competencies* 56. Discuss the implications of variable access and utilisation of genetic testing 57. Outline the process of obtaining informed consent for genetic testing and list pitfalls of the testing process 58. Explain how the association between genetic variants and disease phenotypes is established through genome-wide association studies 59. Discuss the ethical challenges associated with the therapeutic use of stem cells 60. Outline the patient safety considerations during the development of novel therapies using gene therapy and gene editing techniques as examples

Prerequisites	None	**Required**	None

Course content	• Structure and function of the cell • Organisation and function of the genome • Genomic variation and its significance • Cell division and its regulation • Principles of neoplasia • Stem cells and gene therapy • Personalised medicine
Teaching methodology	Lectures – normally two face-to-face, three on-line p/week Tutorials – two case-based learning small group sessions, two expert-led class discussions/debates Flipped classroom activities Community and/or hospital and/or laboratory visits each week, relating to the case of the week Student centred learning/self-study
Bibliography	Required textbooks/reading

Authors	Title	Edition	Publisher	Year	ISBN
Turnpenny, Ellard and Cleaver	Emery's elements of medical genetics and genomics	16th	Elsevier	2020	9780702079665
Alberts et al	Molecular Biology of the Cell	7th	W. W. Norton & Company	2022	978-0393884821
Jorde et al	Medical Genetics	6th	Elsevier	2019	9780323597371

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<tr><td></td><td>Recommended textbooks/reading
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<tr><th>Authors</th><th>Title</th><th>Edition</th><th>Publisher</th><th>Year</th><th>ISBN</th></tr>
<tr><td>Nussbaum et al</td><td>Thompson & Thompson Genetics in Medicine</td><td>8th</td><td>Elsevier</td><td>2015</td><td>9781437706963</td></tr>
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<tr><td>Assessment</td><td>The course will be assessed at the end of Semester 1 with a Summative Final Examination consisting of Single Best Answer MCQs (SBAs) and Short Answer Questions (SAQs).</td></tr>
<tr><td>Language</td><td>English</td></tr>
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