<table>
<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">Dr 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
2. Define the levels of protein structure and correlate with protein function providing examples
3. Outline the basic routes of protein trafficking inside the cell</td></tr>
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	4. Outline the structure and function of the cell membrane and cell junctions 5. Outline the basic structure and key functions of cellular organelles and the cytoskeleton 6. Discuss, with examples, the implications of dysfunction of cellular organelles 7. Outline the process of cell signalling and provide examples of how this process might be manipulated for therapeutic purposes. 8. Describe the structure of DNA and chromatin and compare intragenic and extragenic DNA 9. Describe the basic structure of the human gene 10. Discuss the concept of genetic variation and discuss its role in the aetiology of disease 11. Describe the classification of genetic variants in terms of its effect on gene function and functional implications for the encoded polypeptide 12. Define the modes of inheritance of genetic disease and apply probabilities' principles 13. Describe the basic structure of the human chromosome 14. Describe structural variants of the human chromosome and outline their clinical relevance 15. Outline the process of transcription and discuss its regulation 16. Define epigenetic regulation and outline the different mechanisms 17. Discuss the role of non-coding RNAs in health and disease 18. Outline the process of RNA splicing and discuss how its disturbance can cause disease 19. Outline the process of translation and discuss its regulation 20. Outline post-translational modifications and discuss their significance and clinical correlates 21. Briefly outline techniques to study the transcriptome and proteome 22. Describe mitotic cell division 23. Describe the cell cycle and its regulation 24. Discuss the dysregulation of the cell cycle and the clinical consequences 25. Outline the process of DNA replication 26. List the potential errors during DNA replication and discuss their potential implication if they remain uncorrected 27. Outline how the cellular machinery identifies and corrects DNA errors 28. Discuss the clinical implications of genetic defects in the DNA-correcting machinery of the cell and the clinical implications of DNA transposition 29. Outline the different types of cell death and discuss the role of programmed cell death 30. Describe the structure and function of telomeres and their role in the aging cell, and the main aging theories 31. Outline the key events in the neoplastic process and metastasis, and the main involved signalling pathways 32. Distinguish between driver and passenger mutations 33. Define oncogenes and describe how they contribute to the development of neoplasia 34. Explain how the different types of tumour suppressor genes prevent the development of neoplasia 35. Define Knudson's two-hit hypothesis

	36. Discuss the implications of germline mutations in genes that relate to the neoplastic process 37. 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 38. Define personalised medicine 39. Discuss how genetic studies in populations can contribute to the management of genetic diseases 40. Describe the basis of sequencing techniques such as Sanger and next generation sequencing and discuss clinical applications 41. 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 42. Interpret a basic electropherogram 43. Use appropriate symbols to construct a genogram depicting the inheritance of genetic disease 44. Interpret the notation used in genetic reports and discuss the significance of results 45. Explain the limitations of genomic risk profiling and the pitfalls of direct-to-consumer genetic testing 46. Discuss the implications of variable access and utilisation of genetic testing 47. Outline the process of obtaining informed consent for genetic testing and list pitfalls of the testing process 48. Discuss core issues in bioethics and ethical considerations regarding the use of genetics in health care 49. Discuss ethical issues associated with genetic testing, with particular reference to autonomy, consent, confidentiality 50. Explain how the association between genetic variants and disease phenotypes is established through genome-wide association studies (GWAS) 51. Define gene therapy and outline the different types of methodological approaches 52. Briefly outline gene cloning and gene editing techniques and discuss their research and clinical applications 53. Outline current applications and limitations of gene therapy 54. Define stem cells and compare and contrast embryonic and somatic stem cells 55. Briefly outline the process of somatic stem cell reprogramming to induce pluripotency and discuss the applications of this technique 56. Describe how genetic disorders can be cured by stem cell and tissue engineering approaches 57. Outline how stem cell technology and precision gene therapy can be united to treat genetic diseases 58. Illustrate how targeted treatments can be tailored to the specific mutations causing genetic disease 59. Discuss ethical questions in medical scientific research and challenges associated with stem cell research/therapy 60. Outline the patient safety considerations during the development of novel therapies using gene therapy and gene editing techniques as examples

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<tr><td>Prerequisites</td><td>None</td><td>Required</td><td>None</td></tr>
<tr><td>Course content</td><td colspan="3">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</td></tr>
<tr><td>Teaching methodology</td><td colspan="3">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</td></tr>
<tr><td>Bibliography</td><td colspan="3">Required textbooks/reading
<table>
<tr><th>Authors</th><th>Title</th><th>Edition</th><th>Publisher</th><th>Year</th><th>ISBN</th></tr>
<tr><td>Turnpenny, Ellard and Cleaver</td><td>Emery’s elements of medical genetics and genomics</td><td>16th</td><td>Elsevier</td><td>2021</td><td>9780702079665</td></tr>
<tr><td colspan="6">Library access link:
https://search.ebscohost.com/login.aspx?direct=true&AuthType=ip,sso&db=nlebk&AN=1805966&site=eds-live&custid=s1098328&ebv=EB&ppid=pp_Cover</td></tr>
<tr><td>Alberts et al</td><td>Molecular Biology of the Cell</td><td>7th</td><td>W. W. Norton & Company</td><td>2022</td><td>978-0393884821</td></tr>
<tr><td>Jorde et al</td><td>Medical Genetics</td><td>6th</td><td>Elsevier</td><td>2019</td><td>9780323597371</td></tr>
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Recommended textbooks/reading
<table>
<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 colspan="3">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>
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Language	English