**SYLLABUS**

**Fall semester 2020-2021 academic years**

**On the educational program “7M05109-Genetics”**

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| **Discipline’s code** | **Discipline’s title** | **Independent work of students (IWS)** | **No. of hours per week** | | | | | **Number of credits** | **Independent work of student with teacher (IWST)** |
| **Lectures (L)** | **Practical training (PT)** | | **Laboratory (Lab)** | |
| MMK 6307 | Molecular Mechanisms of Heredity and Variability | 98 | 15 | 30 | | 0 | | 5 | 7 |
| Academic course information | | | | | | | | | |
| **Form of education** | **Type of course** | Types of lectures | | | Types of practical training | | Number of IWS | | Form of final control |
| Combined | Theoretically | analytical lecture  make a slide and analyze | | | performance of tasks | | 6 | | Writing exam in IS Oqylyq |
| Lecturer | Saparbaev Murat | | | | | |  | | |
| e-mail | [\*\*murat.saparbaev@gustaveroussy.fr](mailto:**murat.saparbaev@gustaveroussy.fr) | | | | | |
| Telephone number | +33 142115404 | | | | | |

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| **Academic presentation of the course** |

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| **Aim of course** | **Expected Learning Outcomes (LO)**  As a result of studying the discipline the student will be able to: | **Indicators of LO achievement (ID)**  (for each LO at least 2 indicators) |
| **The learning objective here is for Ph.D. students to gain an understanding of the major DNA repair pathways in eukaryotic cells, highlighting proteins involved in the cellular responses to ionizing radiation or anti-cancer chemotherapy.** | **LO 1.** To provide a solid understanding of the concepts and scientific methods of modern genetics as it applies to humans. | **ID1.1.** Consider, summarize, and debate the efficacy and ethics of modern genetic/genomic testing from the perspective of understanding what genes are.  **ID1.2.** Consider, summarize, and debate the efficacy and ethics of modern genetic/genomic testing from the perspective of understanding how genes inherited, how they result in traits or human disease, and how genomic analyses are performed & interpreted. |
| **LO 2.** To develop a better appreciation of the power and the limitations of a genetics-centric view of human biology and disease. | **ID2.1.** Comprehend a given current affairs article, news story, or documentary related to human genetics (e.g. genomics testing, human disease, human evolution);  **ID2.2.** Confidently summarize the major points to family, friends, and colleagues including conveying the issue’s significance (including any ethical concerns) and explaining the underlying genetics and molecular biology. |
| **LO 3.** To develop conceptual skills to address questions in genetics research and clinical practice | **ID3.1.** Ability to work in international, international scientific teams, to be politically correct in any non-standard situations.  **ID3.2.** Ability to develop creative individual abilities, adherence to cultural and ethical values for solving world outlook problems in the field of biology; possession of socially significant ideas about a healthy environment, readiness for various types of communication to address environmental and other security issues. |
| **LO 4.** To develop critical thinking with regard to news reports of advances in genetics and their social implications. | **ID4.1.** Appreciate the origins of many fundamental biology and human health breakthroughs in genetic studies of model organisms.  **ID4.2.** Apply the scientific method to design a hypothetical, straightforward genetics experiment to test an unresolved scientific question and/or to appraise/rebut a claim based on scientific results (or the lack thereof). |
| **Prerequisites** | "Biochemistry-1В22", "Genetics-Gen3419", "Molecular biology-1В118", "Special Practical for human cytogenetics-SPCytGch3505". "General and molecular genetics-1B210", "Basics of Development genetics -OGR 3506 ", "Genetic engineering-GI 3418". | |
| **Post requisites** | Doctoral students can use this knowledge in solving specific scientific problems defense of the thesis | |
| **Information resources** | **Main literature:**   1. Benjamin Lewin. Genes VIII: AND Molecular Biology of the Gene Prentice Hall, 2007. 2. Griffiths AJF, Gelbart WM, Miller JH, et al. Modern Genetic Analysis.New York: W. H. Freeman; 1999. 3. Brown T. A. Genomes 3. 3rd edition. Garland Science: New York, 2006. 711p. 4. Brown T. A. Gene Cloning and DNA Analysis: An Introduction/ Blackwell Publishings,2010. 5. W.A. Bickmore. Chromosome Structural analysis: A Practical Approach/ Oxford University Press*,* UK, 1999, 234 pp 6. Sambrook J. et al. Molecular Cloning: A Laboratory Manual (3- Volume Set)/ CSHL Press,2012. 7. Strachan, T. Human Molecular Genetics. 3rd ed. New York, NY: Garland Science, 2003.   **Internet-resources:**  <http://study.com/academy/subj/science.html>  <https://www.khanacademy.org>  https://www.nature.com/scitable/topics | |

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| **Academic policy of the course in the context of university moral and ethical values** | **Academic Behavior Rules:**  All students have to register at the MOOC. The deadlines for completing the modules of the online course must be strictly observed in accordance with the discipline study schedule.  ATTENTION! Non-compliance with deadlines leads to loss of points! The deadline of each task is indicated in the calendar (schedule) of implementation of the content of the curriculum, as well as in the MOOC.  **Academic values:**  - Practical trainings/laboratories, IWS should be independent, creative.  - Plagiarism, forgery, cheating at all stages of control are unacceptable.  - Students with disabilities can receive counseling at e-mail \*\*\*\*\*\*\*@gmail.com. |
| **Evaluation and attestation policy** | **Criteria-based evaluation:**  Homework (preparation for a practical class) – 35%;  Selection of literature sources on the research topic - 10  Writing a literary review - 15;  Exams (including Midterm exam) – 40%. TOTAL – 100%  **Summative evaluation:**  Your final score will be calculated using the formula  Final score=(MT1+MT2)/\*0.6+0.1MT+0.3 FC  Below are the minimum estimates in percentage terms:  95% - 100%: А 90% - 94%: А-  85% - 89%: В+ 80% - 84%: В 75% - 79%: В-  70% - 74%: С+ 65% - 69%: С 60% - 64%: С-  55% - 59%: D+ 50% - 54%: D- 0% -49%: F |

**CALENDAR (SCHEDULE) THE IMPLEMENTATION OF THE COURSE CONTENT:**

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| weeks | | Topic name | LO | ID | amount of hours | | Maximum score | Form of Knowledge Assessment | The  Form of the lesson  / platform |
| **Module 1 DNA damage and mutagenesis** | | | | | | | | | | |
| 1 | L.1.«Chemical and physical alterations in primary structure of nucleic acids and their consequences on heredity and variability in the population». | | LО 1 | ID 1.1. | | 2 |  |  | Video lecture  in ZOOM | |
| PT1. (i) Nature of spontaneous DNA damage and their role in mutagenesis.  (ii) Nature of oxidative DNA damage and their role in aging and cancer. | | LО 1 | ID 1.2. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 2 | L.2. Role of environmental factors in the genome stability and mutational variability. | | LО 2 | ID 2.1. | | 2 |  |  | Video lecture  in ZOOM | |
| PT 2. (i) Nature and mechanisms of chemical mutagens and carcinogens.  (ii) Role of ultraviolet and ionizing radiation in DNA damage and mutagenesis. | | LО | ID 2.2. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 3 | L.3. « Mechanisms of cellular response to DNA damage in bacteria and eukaryotes their role in the maintenance of genome stability ». | | LО 1 | ID 1.1. | |  |  |  | Video lecture  in ZOOM | |
| PT 3. (i) DNA damage response in bacteria and its role in mutagenesis.  (ii) DNA damage responses in eukaryotes and their role in mutagenesis. | | LО 2 | ID 2.1. | |  | 10 |  | in the auditorium traditionally | |
| IWSP 1 Consultation on the implementation of IWS 1. | | LО 4 | ID 4.1. | |  |  |  | Univer system | |
| IWS 1. | | LО4 | ID 4.1. | |  | 25 | Logic task |
| **Module П Molecular mechanisms of DNA repair** | | | | | | | | | | |
| 4 | L.4. “Molecular mechanisms of DNA repair of non-bulky lesions in cellular genome”. | | LО 3 | ID 3.1. | | 1 |  |  | Video lecture  in ZOOM | |
| PT 4. (i) Principles of base excision repair and role in genome stability.  (ii) DNA glycosylases: their substrate specificities and role in mutagenesis. | | LО 1 | ID 1.1. | |  | 10 |  | in the auditorium traditionally | |
| 5 | L.5. “Molecular mechanisms of nucleotide incision repair for oxidative DNA base damage and its role in cellular protection against genetoxic stress”. | | LО 2 | ID 2.1. | |  |  |  | Video lecture  in ZOOM | |
| PT 5. (i) Principles of nucleotide incision repair pathway.  (ii) AP endonucleases: their substrate specificities and biological roles. | | LО 1 | ID 1.1. | |  | 10 |  | in the auditorium traditionally | |
| IWSP 2 Consultation on the implementation of IWS2 | | LО 3 | ID 3.1. | |  |  |  | Univer system | |
| IWS2  Make a structural and logical diagram of the read material | | LО 3 | ID 3.1.  ID 3.2. | |  | 25 | Logic task |
| 5 | MT 1 | | LО 1 | ID 1.1. | |  | 100 |  |  | |
| 6 | L.6. “Molecular mechanisms of direct DNA damage reversal in mutations prevention and active epigenetic reprogramming”. | | LО 1 | ID 1.1. | | 2 |  |  | Video lecture  in ZOOM | |
| PT 6. (i) Mechanisms of direct repair of ultraviolet and alkylation DNA damage.  (ii) Oxidative DNA demethylases and epigenetic reprogramming. | | LО 1 | ID 1.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 7 | L.7. “Molecular mechanisms of bulky DNA lesions and their role in mutagenesis and human genetically inherited diseases”. | | LО 1 | ID 1.1. | |  |  |  | Video lecture  in ZOOM | |
| PT 7. (i) Principles of nucleotide excision repair in bacteria: role in mutagenesis and cell survival.  (ii) Principles of nucleotide excision repair in eukaryotes: role in protection against chemical carcinogens and UV | | LО 5 | ID 5.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 8 | L.8. “Molecular mechanisms of homologous recombination and their role in DNA replication and chromosome breakage repair”. | | LО 1 | ID 1.1. | | 2 |  |  | Video lecture  in ZOOM | |
| PT 8. (i) Role of RecA and Rad51 proteins in homologous recombination and repair.  (ii) Role of homologous recombination in maintenance of genetic variability in population and avoidance of deleterious mutations. | | LО 1 | ID 1.1. | |  | 10 | Analysis | in the auditorium traditionally | |
| IWSP 3 Consultation on the implementation of IWS 3.1 | | LО4 | ID 4.1. | |  |  |  | Univer system | |
| IWS 3.1 | | LО4 | ID 4.1. | |  | 20 | Logic task |
| 9 | L.9. “Molecular mechanisms of non-homologous end joining (NHEJ) and their role in protection against ionizing radiation and immune response”. | | LО 1 | ID 1.1. | |  |  |  | Video lecture  in ZOOM | |
| PT 9. Principles of non-homologous end joining (NHEJ)  (ii) Repair of double strand breaks generated by ionizing radiation and immune system”. | | LО 1 | ID 1.1. | | 2 | 10 | Analysis | in the auditorium traditionally | |
| 10 | L.10. “Molecular mechanisms of DNA replication errors and their role in the maintenance of genome stability and low rate of spontaneous mutations”. | | LО 1 | ID 1.1. | | 2 |  |  | Video lecture  in ZOOM | |
| PT 10. (i) Principles of mismatch repair in bacteria.  (ii) Principles of mismatch repair in eukaryotes and their role in cancer. | | LО 1 | ID 1.1. | |  | 10 | Analysis | in the auditorium traditionally | |
| IWSP 4 Consultation on the implementation of IWS 3.2 | | LО4 | ID 4.1. | |  |  |  | Univer system | |
| IWS 3.2 Make a structural and logical diagram of the read material | | LО4 | ID 4.1. | |  | 15 | Problem task |
| IWSP 5 Consultation on the implementation of IWS 4 | | LО 3 | ID 3.2. | |  |  |  |
|  | IWS 4 Make a structural and logical diagram of the read material | | LО 3 | ID 3.2. | |  | 15 | Problem task | Univer system | |
| 10 | МТ (Midterm Exam) | | LО 1 | ID 1.1. | |  | 100 |  | in the auditorium traditionally | |
| **Module П Molecular mechanisms of DNA repair** | | | | | | | | | | |
| 11 | L.11. “Molecular mechanisms of action of poly(ADP-ribose) polymerases (PARPs) in DNA strand break signaling and repair: their role in genome stability and transcription regulation”. | | LО 1 | ID 1.1. | |  |  |  | Video lecture  in ZOOM | |
| PT 11. (i) Poly(ADP-ribose polymerases (PARP): structures and functions.  (ii) Poly(ADP-ribose polymerases (PARP): chromatin remodeling and epigenetic regulation. | | LО 1 | ID 1.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 12 | L.12. “Molecular mechanisms of aberrant DNA repair and their role in mutagenesis and genetic evolution”. | | LО 2 | ID 2.1. | | 1 |  |  | Video lecture  in ZOOM | |
| PT 12. (i) DNA glycosylase mediated aberrant error prone repair and its role in cancer and aging.  (ii) Aberrant DNA repair and evolution of CpG islands in warm blood animals. | | LО 2 | ID 2.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| IWSP 6 Consultation on the implementation of IWS3 | | LО 2 | ID 2.1. | |  |  |  | Univer system | |
| IWS 5 Make a structural and logical diagram of the read material | | LО 1 | ID 1.1. | |  | 25 | Problem task |
| 13 | L.13. “Molecular mechanisms of inter-strand DNA crosslinks (ICLs) repair and their role in the genome stability, mutagenesis and age-related diseases”. | | LО 1 | ID 1.1. | | 1 |  |  | Video lecture  in ZOOM | |
| PT 13. Fanconi anemia, a major cellular system to detect and repair inter-strand DNA crosslinks (ICLs).  (ii) Alternative DNA glycosylase dependent repair of inter-strand DNA crosslinks (ICLs). | | LО 1 | ID 1.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 14 | L.14. “DNA repair and mutagenesis in vertebrate mitochondria”. | | LО 1 | ID 1.1. | | 1 |  |  | Video lecture  in ZOOM | |
| PT 14. (i) Mutational patterns in mitochondrial genome and role of oxidative stress and aging.  (ii) Mitochondrial DNA replication and repair: evidence for asymmetric DNA strand inheritance | | LО 1 | ID 1.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| 15 | L.15. Mutational signatures in cancer and aging. | | LО 1 | ID 1.1. | | 1 |  |  | Video lecture  in ZOOM | |
| PT 15. (i) Mechanisms of mutagenesis in cancer.  (ii) Passenger and driver mutations in cancer. | | LО 1 | ID 1.1. | | 1 | 10 | Analysis | in the auditorium traditionally | |
| IWSP 7 Consultation on the implementation of IWS 3.1 | | LО 5 | ID 5.1. | |  | 25 |  | Univer system | |
| IWS 6 Make a structural and logical diagram of the read material | | LО4 | ID 4.1. | |  |  | Analysis |
|  | MT 2 | | LО4 | ID 4.1. | |  | 100 |  | in the auditorium traditionally | |

[Abbreviations: QS - questions for self-examination; TK - typical tasks; IT - individual tasks; CW - control work; MT - midterm.

Comments:

- Form of L and PT: webinar in MS Teams / Zoom (presentation of video materials for 10-15 minutes, then its discussion / consolidation in the form of a discussion / problem solving / ...)

- Form of carrying out the CW: webinar (at the end of the course, the students pass screenshots of the work to the monitor, he/she sends them to the teacher) / test in the Moodle DLS.

- All course materials (L, QS, TK, IT, etc.) see here (see Literature and Resources, p. 6).

- Tasks for the next week open after each deadline.

- CW assignments are given by the teacher at the beginning of the webinar.]

**Dean \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Zayadan B.K.**

**Chairman of the**

**Faculty Methodical Bureau \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Nazarbekova S.T.**

**Head of the Department \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Lovinskaya A.B.**

**Lecturer** \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ **Saparbaev M.K.**