#### **APPROVED**

at a meeting of the Academic Council of NJSC «KazNU named after al-Farabi»
Protocol № 11 from 23.05.2025 y.

## Entrance exam program for the applicants to doctoral studies for a group of educational programs D081 – «Genetics»

### I. General provisions

- 1. The program was compiled in accordance with the Order of the Minister of Education and Science of the Republic of Kazakhstan dated October 31, 2018 No. 600 «On approval of the Model Rules for admission to training in educational organizations implementing educational programs of higher and postgraduate education» (hereinafter referred to as the Model Rules).
- 2. The entrance exam to doctoral studies consists of an interview, writing an essay and an exam on the profile of the group of educational programs.

| Block   | Points |
|---|--------|
| 1. Interview  | 30     |
| 2. Essay  | 20     |
| 3. Exam according to the group profile of the educational program | 50     |
| Total/pass  | 100/75 |

3. The duration of the entrance exam is 3 hours 10 minutes, during which the applicant writes an essay and answers the electronic examination ticket. The interview is conducted at the university premises before the entrance exam.

### II. Procedure for conducting the entrance exam

1. Applicants to doctoral studies for group of educational programs D081 – "Genetics" write a problem / thematic essay. The essay should be at least 250 words.

The purpose of the essay is to determine the level of analytical and creative abilities, expressed in the ability to build one's own argumentation based on theoretical knowledge, social and personal experience.

## 2. The electronic examination ticket consists of 3 questions.

Topics for preparing for the exam according to the profile of the educational program group:

Subject and tasks of modern genetics. Methodology of modern genetics. Using the laws of classical genetics in system analysis. Goals, objectives, prospects and methods of various areas of genetics (pharmacogenetics, environmental genetics, radiation genetics, medical genetics, oncogenetics, immunogenetics, etc.). Heredity,

inheritance, heritability. The classical approach from phenotype to genotype and molecular-genetic methods from genotype to phenotype. Mendel's laws. Chromosome theory of heredity of the Morgan school.

Model objects and their role in genetic research. Biological features of model objects and their role in genetic research. Major differences in the organization of cells in prokaryotes and eukaryotes. Application of knowledge about the life cycles of plants and animals in genetic experiments. Selection of a model object in accordance with the purpose and objectives of the study. Bioethical issues and risks. The problem of extrapolation to humans of the results of testing mutagens obtained using various model objects.

Hybridization. Monohybrid crossing. Dihybrid crossing. Unrelated crossing. Hybridological method of studying heredity. Prospects for using crossbreeding systems to obtain highly productive breeds of farm animals, varieties of cultivated plants and strains of microorganisms. Chemical and radiation mutagenesis as a method for increasing the diversity of starting material for hybridization.

*Mitosis, meiosis and cell cycle*. Features of the course of mitosis in polyploidy and polyteny. Spermatogenesis and oogenesis. Mechanisms of chromatin condensation and decondensation at different periods of the cell cycle. Formation of the fission spindle.

Structure and properties of chromosomes. Comparative analysis of prokaryotic and eukaryotic chromosomes and their properties. Euchromatic and heterochromatic regions of chromosomes. Chromosomal additions. Methods of interphase cytogenetics. Endogenous and exogenous sources of mutations. Gene, chromosomal and genomic mutations, their classification and examples of diseases associated with the occurrence of mutations. Mobile elements as a source of mutations. Induced and spontaneous mutagenesis. Cytogenetic research methods. Basic principles of cytogenetic analysis. Modern cytogenetic methods for testing the mutagenic activity of environmental factors and assessing its quality. Methods for differential staining of chromosomes.

*Karyotyping*. Genetic maps of chromosomes and their practical application. Diagnostic problems of fetal karyotyping. Sex chromatin.

Methods for localizing genes in chromosomes. Identification of localized genes. Linkage analysis and gene mapping. Methods used for genetic mapping.

Genetic methods of research in the field of human genetics. Genealogical method. The twin method and its application to assess the role of the environment in the manifestation and heritability of traits. Comparison of concordance between mono- and dizygotic twins. Molecular diagnostics.

Molecular genetic methods of analysis in genetic research. The use of molecular-genetic methods to study the mechanisms of genetic processes (gene expression, translation, transcription, repair, methylation and genomic imprinting, etc.), the action of individual genes and intergenic interactions. Methods for identifying factors in the development of carcinogenesis. Methods for molecular-genetic assessment of cancer. Basic stages of DNA isolation and purification. DNA sequencing.

Use of genetic methods to solve issues in the food industry, agriculture and medicine. Solving problems in the food industry, agriculture, medicine based on genetic methods. Genetic prerequisites for modern methods of artificial insemination in humans (IVF). Polymerase chain reaction (PCR) method: principle, stages, reaction components, varieties and equipment for PCR. Prospects and problems of using stem cells.

General principles and methods of genetic engineering. Review of enzymes used in genetic engineering. Methods of enzymology in genetic engineering. Genetic engineering of cultured mammalian cells. Methods for introducing DNA molecules into mammalian cells (hypertonic salt method, DEAE-dextran method, calcium phosphate method, etc.). Targeted DNA mutagenesis in vitro. Gene cloning. Methods for selecting hybrid clones of bacterial cells. Selective media, competent cells. Enzyme-linked immunosorbent assay (ELISA, ELISA).

General characteristics of vectors. DNA-based vector systems. Introduction of DNA molecules into cells. Methods for constructing recombinant DNA molecules. Methods for determining the nucleotide sequence of DNA. Vector systems based on animal and plant cells. Shuttle vectors, selective marker genes, transformation frequency.

*Methods for obtaining transgenic plants*. Obtaining crops with higher yields and resistance to pests. Modern methods of plant transformation. Crown galls, use of Ti-plasmids and Ri-plasmids to obtain transgenic plants, T-DNA. Plant viruses as vectors for genetic engineering. Application of transgenic plants.

*Methods for reconstruction of transgenic animals.* The feasibility and stages of obtaining transgenic animals. Technologies for the use of embryonic stem cells. Creation of transgenes with disabled genes, gene targeting.

*Prospects for gene therapy*. Ways to eliminate hereditary and non-hereditary diseases by transforming patient cells with "healthy" and regulatory genes. Bioethical problems of gene therapy

Modern achievements in the selection of cereal crops. Methods of chromosome engineering in wheat. Problems of hetero-, poly- and aneuploidy of plants. Chromosomal abnormalities on example of plants, in particular, cereals. Development of chromosome nomenclature. Discovery, study and use of aneuploids. Methods for creating a series of aneuploid lines of bread wheat. Schemes for obtaining aneuploids. Intervarietal chromosome substitution. Scheme for obtaining lines with intervarietal chromosome substitution. Replacement of a chromosome involved in reciprocal translocation in wheat. Genomic analysis of common wheat.

Methods used in genetic analysis of bacteria and bacteriophages. Clonal analysis. Selective media method. Fingerprint method

Genetic bioinformatics. Gene systematics. Genetic collections. Relationship between molecular genetics and bioinformatics. Conservation of biodiversity *in situ* and *ex situ*. Test forms and analyzer lines. Cell culture banks. Gene banks. 3D bioprinting. DNA markers in taxonomic studies. DNA barcoding methodology

*Radiation genetics.* Hereditary disorders due to radiation injuries. Routes of radionuclides entry into the body. Methods of radiation genetics. Nuclear medicine. "Peaceful" atom. Biodosimetry

Theories of evolution. Evolutionary ideas of J.-B. Lamarck. Driving forces of progressive evolution (gradation) and speciation. The inconsistency of Lamarck's ideas. The main driving forces of evolution according to Darwin. The main provisions of the theory by Charles Darwin. Basic provisions of the synthetic theory of evolution.

A population is an elementary unit of the evolutionary process. Population is the elementary unit of evolution. Genetic heterogeneity and polymorphism of the population. Genetic unity of the population. Hardy-Weinberg law. Conditions under which the Hardy-Weinberg equation is satisfied. Ecological and genetic variability of the population and methods of their analysis. DNA markers in population genetic studies.

*Genetic basis of evolution*. Variability: phenotypic, genotypic, paratypic, modification. Modifications. Norm of reaction. The concept of adaptive reaction norm.

*Elementary factors of evolution.* Factors of evolution that cause changes in the genotypic structure of the population. Natural selection and the meaning of probability and chance.

Biological and evolutionary significance of species. General concept of the type, characteristic of the difficulty in its universal application. Reasons for the increase in the number of species. Speciation. Instant speciation. Gradual and sympatric speciation.

The significance of geographic variability and its adaptive nature. Analysis of isolating mechanisms, their classification and significance. Examples of disruption of isolating mechanisms.

*Evolution of ontogeny*. The concept of ontogenesis. Features of ontogenesis in different organisms and its duration. Ontogenetic differentiation. Duration of ontogenesis. Integrity and stability of ontogenesis. Autonomy and embryonic ontogenesis.

Evolution of phylogenetic groups. The main forms of phylogeny: phyletic evolution, divergence, parallelism, convergence. The main directions of evolution are arogenesis and allogenesis. Forms of allogenesis (telogenesis, hypergenesis, hypogenesis, etc.) Typical change of phases of adaptationogenesis.

Directions and patterns of the evolutionary process. The relationship between micro- and macroevolution. The relationship between onto- and phylogeny. Muller-Haeckel law. The theory of phylembryogenesis by A.N. Severtsev. Biological and morphophysiological progress and regression in evolution. Recapitulation. Molecular evolution of genes.

Anthropogenesis. The place of man in the system of the animal world. Human ancestors. The main stages of the evolution of Homo. The main stages of development of Homo sapiens. The role of work and social lifestyle in human evolution. The influence of elementary evolutionary factors on the evolution of modern man.

*Biostatistics in genetics*. Statistical methods for interpreting data in genetic studies. Application packages for statistical processing of genetic data.

#### III. List of sources used

#### Main:

- 1. Omirbekova N.Zh., Zhunusbaeva Zh.K., Bersimbay R.I. Small workshop on general genetics: educational and methodological manual. Almaty: Kazakh University, 2017. 100 p.
- 2. Bisenbaev A.K. Molecular biology: a collection of problems and tests. Almaty: Kazakh University, 2021. 186 p.
- 3. Inge-Vechtomov, S. G. Genetics with the basics of selection: a textbook for students. higher textbook institutions / S. G. Inge-Vechtomov. 3rd ed. St. Petersburg: Publishing House N-L, 2015. 718 p.
- 4. Zhimulev I.F. General and molecular genetics: textbook for universities Novosibirsk: Siberian University Publishing House, 2017. 480 p.
- 5. Makrushin N. M., Plugatar Yu. V., Makrushina E. M., Goncharova Yu. K., Goncharov S. V., Shabanov R. Yu. Genetics: a textbook for universities: 2nd ed. St. Petersburg: Lan Publishing House, 2021. 404 p.
- 6. Molecular biology of the cell: in 3 volumes / B. Alberts, A. Johnson, D. Lewis, etc. M.-Izhevsk: Research Center "Regular and Chaotic Dynamics", Institute of Computer Research, 2013. 808 p.
- 7. Severtsov A.S. Theories of evolution: a textbook for universities / A.S.Severtsov. 2nd ed., rev. M.: Yurayt Publishing House, 2020. 384 p.
- 8. Medical genetics: textbook / ed. N. P. Bochkova. M.: GEOTAR-Media, 2014. 224 p.
- 9. Biyasheva Z.M., Lovinskaya A.V., Dauletbaeva S.B., Kalimagambetov A.M. Statistical methods in biology with software // Textbook for biol. specialties: Almaty Kazak University, 2019. 108 p.

# Supplementary:

- 1. Cytogenetic methods in genetic monitoring: educational method. allowance / S.Zh. Kolumbaeva, A.V. Lovinskaya, A.M. Kalimagambetov; KazNU named after. al-Farabi. Almaty: Kazakh University, 2018. 161 p.
- 2. Bochkov N.P. Clinical genetics: textbook. / N. P. Bochkov, V. P. Puzyrev, S. A. Smirnikhina; under. ed. N.P. Bochkova. 4th ed., add. and processed M.: Publishing house. group "GEOTAR-Media", 2013. 582 p.
- 3. Abilev S.K., Glazer V.M. Mutagenesis with the basics of genotoxicology. St. Petersburg: Nestor-History, 2015. 304 p.
- 4. Genetic basis of plant breeding. In 4 volumes. T. 4. Biotechnology in plant breeding. Genomics and genetic engineering. Minsk: Publishing House "Belarusian Science", 2014. 653 p.
- 5. Faller, D.M. Molecular Cell Biology: A Guide for Physicians / Gerald M. Faller, Dennis Shields; lane from English under general ed. acad. I. B. Zbarsky. Moscow: Binom-Press, 2012. 256 p.