**Семинар № 1 на тему «Знакомство с направлениями работы кафедры», проводимый в рамках кружка «Генетик» кафедры молекулярной биологии и генетики**

Семинар проходил в соответствии с нижеизложенными пунктами (на рус.):

1. Возникнув как биохимия нуклеиновых кислот, **молекулярная биология** пережила период бурного развития собственных методов исследования, которыми теперь отличается от биохимии. К ним, в частности, относятся методы генной инженерии, клонирования, искусственной экспрессии и нокаута генов. Поскольку ДНК является материальным носителем генетической информации, молекулярная биология значительно сблизилась с генетикой, и на стыке образовалась молекулярная генетика, являющаяся одновременно разделом генетики и молекулярной биологии. Так же, как молекулярная биология широко применяет вирусы как инструмент исследования, в вирусологии для решения своих задач используют методы молекулярной биологии. Для анализа генетической информации привлекается вычислительная техника, в связи с чем появились новые направления молекулярной генетики, которые иногда считают особыми дисциплинами: биоинформатика, геномика и протеомика.

2. **Дата основания кафедры:** Кафедра Дарвинизма и генетики основана в 1948 году. С открытием специальности молекулярная биология в 1987 году кафедра переименована как генетики и молекулярной биологии, а в 2011 году, в связи с нововведениями в университете, название кафедры изменено на кафедра молекулярной биологии и генетики.

**Заведующая кафедрой:** д.б.н., профессор Айташева З.Г.

3. С начала организации кафедры выпущено более 700 специалистов. Аспирантуру на кафедре успешно окончили более 70 человек. Свыше 100 выпускников, аспирантов и докторантов кафедры имеют ученые степени докторов и кандидатов биологических наук, а с 2008 г. - также ученую степень Ph.D.

Многие выпускники кафедры являются видными учеными и организаторами науки, директорами и руководителями подразделений НИИ, опытных и селекционных станций, преподавателями ВУЗов. **Наши выпускники** заслужили высокий авторитет своими трудами среди зарубежных специалистов (в Великобритании, Египте, Израиле, США, Франции, ФРГ и др. странах).

4. **Кафедра сотрудничает** с научными центрами и учебными заведениями стран ближнего и дальнего зарубежья. В целях расширения международного сотрудничества преподаватели кафедры проходят научные стажировки в ближнем и дальнем зарубежье, приглашаются зарубежные ученые для чтения лекций и проведения практических занятий.

5. Кафедра уделяет большое внимание преподаванию дисциплин, имеющих большое значение и развитие, как в глобальном, так и на республиканском уровне: молекулярная биология, биотехнология, генетика. **Обучение** ведется в модели непрерывного образования на трех языках с использованием элементов мультимедийного пространства.

6. На кафедре имеются следующие **лаборатории**: учебно-исследовательская лаборатория «Мутагенеза», лаборатория общей генетики и селекции, лаборатория хромосомной инженерии, учебная лаборатория им. академика Н.Л. Удольской, лаборатория большого практикума по генетике, лаборатория генофонда линий дрозофилы, лаборатория цитогенетики человека, лаборатория биохимии и биотехнологии.

7. В настоящее время на кафедре работает на постоянной основе 26 человек.

Руководитель кружка
PhD, и.о. доцента Жусупова А.И. 26 сентября 2014 г.

**Семинар № 3 на тему «Human genome project and postgenomic era», проводимый в рамках кружка «Генетик» кафедры молекулярной биологии и генетики**

Семинар проходил в соответствии с нижеизложенными пунктами (на англ.):

1. The Human Genome Project (HGP) was the international, collaborative research program whose goal was the complete mapping and understanding of all the genes of human beings. All our genes together are known as our "genome."

2. The HGP has revealed that there are probably about 20,500 human genes. The completed human sequence can now identify their locations. This ultimate product of the HGP has given the world a resource of detailed information about the structure, organization and function of the complete set of human genes. This information can be thought of as the basic set of inheritable "instructions" for the development and function of a human being.

3. The International Human Genome Sequencing Consortium published the first draft of the human genome in the journal *Nature* in February 2001 with the sequence of the entire genome's three billion base pairs some 90 percent complete. A startling finding of this first draft was that the number of human genes appeared to be significantly fewer than previous estimates, which ranged from 50,000 genes to as many as 140,000.The full sequence was completed and published in April 2003.

Upon publication of the majority of the genome in February 2001, Francis Collins, the director of NHGRI, noted that the genome could be thought of in terms of a book with multiple uses: "It's a history book - a narrative of the journey of our species through time. It's a shop manual, with an incredibly detailed blueprint for building every human cell. And it's a transformative textbook of medicine, with insights that will give health care providers immense new powers to treat, prevent and cure disease."

4. Information may no longer be a bottleneck to understanding and tackling complex genetic diseases such as cancer. For those who cut their research teeth not so very long ago, before PCR was thought of by Kary Mullis, this is truly an amazing position to be in. To think that effective management of cancer is potentially just a time and money issue equation, where new disease causation data needs to be converted into novel targeted or personalized therapies, how can we eliminate drug discovery economics as an overriding limiting factor?

5. The genomic era has brought with it a basic change in experimentation, enabling researchers to look more comprehensively at biological systems. The sequencing of the human genome coupled with advances in automation and parallelization technologies have afforded a fundamental transformation in the drug target discovery paradigm, towards systematic whole genome and proteome analyses. In conjunction with novel proteomic techniques, genome-wide annotation of function in cellular models is possible. Overlaying data derived from whole genome sequence, expression and functional analysis will facilitate the identification of causal genes in disease and significantly streamline the target validation process. Moreover, several parallel technological advances in small molecule screening have resulted in the development of expeditious and powerful platforms for elucidating inhibitors of protein or pathway function. Conversely, high-throughput and automated systems are currently being used to identify targets of orphan small molecules. The consolidation of these emerging functional genomics and drug discovery technologies promises to reap the fruits of the genomic revolution.

Руководитель кружка
PhD, и.о. доцента Жусупова А.И. 28 ноября 2014 г.

**Семинар № 4 на тему «Gene ontology», проводимый в рамках кружка «Генетик» кафедры молекулярной биологии и генетики**

Семинар проходил в соответствии с нижеизложенными пунктами (на англ.):

1. From a practical view, an ontology is a representation of something we know about. “Ontologies” consist of a representation of things that are detectable or directly observable, and the relationships between those things. There is no universal standard terminology in biology and related domains, and term usages may be specific to a species, research area or even a particular research group. This makes communication and sharing of data more difficult. The Gene Ontology project provides an ontology of defined terms representing gene product properties. The ontology covers three domains: - cellular component, the parts of a cell or its extracellular environment; - molecular function, the elemental activities of a gene product at the molecular level, such as binding or catalysis; - biological process, operations or sets of molecular events with a defined beginning and end, pertinent to the functioning of integrated living units: cells, tissues, organs, and organisms.

2. Each GO term within the ontology has a term name, which may be a word or string of words; a unique alphanumeric identifier; a definition with cited sources; and a namespace indicating the domain to which it belongs. Terms may also have synonyms, which are classed as being exactly equivalent to the term name, broader, narrower, or related; references to equivalent concepts in other databases; and comments on term meaning or usage. The GO ontology is structured as a directed acyclic graph, and each term has defined relationships to one or more other terms in the same domain, and sometimes to other domains. The GO vocabulary is designed to be species-neutral, and includes terms applicable to prokaryotes and eukaryotes, single and multicellular organisms.

3. GO is not static, and additions, corrections and alterations are suggested by, and solicited from, members of the research and annotation communities, as well as by those directly involved in the GO project. For example, an annotator may request a specific term to represent a metabolic pathway, or a section of the ontology may be revised with the help of community experts. Suggested edits are reviewed by the ontology editors, and implemented where appropriate.

4. Genome annotation is the practice of capturing data about a gene product, and GO annotations use terms from the GO ontology to do so. The members of the GO Consortium submit their annotation for integration and dissemination on the GO website, where they can be downloaded directly or viewed online using AmiGO. In addition to the gene product identifier and the relevant GO term, GO annotations have the following data:

- The reference used to make the annotation (e.g. a journal article)
- An evidence code denoting the type of evidence upon which the annotation is based
- The date and the creator of the annotation

5. There are a large number of tools available both online and to download that use the data provided by the GO project. The vast majority of these come from third parties; the GO Consortium develops and supports two tools, AmiGO and OBO-Edit.

AmiGO is a web-based application that allows users to query, browse and visualize ontologies and gene product annotation data. In addition, it also has a BLAST tool, tools allowing analysis of larger data sets, and an interface to query the GO database directly.

OBO-Edit is an open source, platform-independent ontology editor developed and maintained by the Gene Ontology Consortium. It is implemented in Java, and uses a graph-oriented approach to display and edit ontologies.

Руководитель кружка
PhD, и.о. доцента Жусупова А.И. 26 декабря 2014 г.

**Семинар № 5 на тему «Prospects on human disease study and prevention», проводимый в рамках кружка «Генетик»**
**кафедры молекулярной биологии и генетики**

Семинар проходил в соответствии с нижеизложенными пунктами (на англ.):

1. Common diseases such as type 2 diabetes and coronary heart disease result from a complex interplay of genetic and environmental factors. Recent developments in genomics research have boosted progress in the discovery of susceptibility genes and fueled expectations about opportunities of genetic profiling for personalizing medicine. Personalized medicine requires a test that fairly accurately predicts disease risk, particularly when interventions are invasive, expensive or have major side effects.

2. Recent studies on the prediction of common diseases based on multiple genetic variants alone or in addition to traditional disease risk factors showed limited predictive value so far, but all have investigated only a limited number of susceptibility variants. New gene discoveries from genome-wide association studies will certainly further improve the prediction of common diseases, but the question is whether this improvement is sufficient to enable personalized medicine.

3. There is considerable anticipation of future improvements in disease prevention and treatment following recent advances in genomics. One aspect of genomics that is receiving considerable interest is epigenetics - the regulatory processes that control the transcription of information encoded in the DNA sequence into RNA before their translation into proteins. Programmed developmental changes and the ability of the genome to register, signal, and perpetuate environmental cues are subsumed under the epigenetic banner

The epigenome records a variety of dietary, lifestyle, behavioral, and social cues, providing an interface between the environment and the genome. Epigenetic variation, whether genetically or environmentally determined, contributes to inter-individual variation in gene expression and thus to variation in common complex disease risk. Interventions based upon epigenetic agents, including DNA methyltransferase inhibitors and histone deacetylase inhibitors, have been in clinical use for many years, but their role outside treatment of specific cancers is not established.

Epigenetic therapies will only be fruitful if epigenetic mechanisms are causally related to the disease being treated. Evidence linking epigenetic variation to specific disease phenotypes to date is lacking. Epidemiological approaches can be applied to help separate causal from non-causal associations.

4. There is an increasing body of evidence to demonstrate that epigenetic patterns are altered by environmental factors known to be associated with disease risk (e.g., diet, smoking, alcohol intake, environmental toxicants, stress); however, an important question remains to be resolved in defining which epigenetic changes are a secondary outcome of either exposure or disease, and which lie on the causal pathway linking the two. Without proven causality, interventions to prevent or treat common complex diseases based upon epigenetic mechanisms will not be fruitful. Conversely, regardless of causality, defining a robust prospective relationship between epigenetic patterns and phenotypic traits may have application in diagnostics or in identifying high-risk individuals for non-epigenetic-based interventions.

5. Substantial improvements may only be expected if we manage to understand the complete causal mechanisms of common diseases to a similar extent as we understand those of monogenic disorders. Genomics research will contribute to this understanding, but it is likely that the complexity of complex diseases may ultimately limit the opportunities for accurate prediction of disease in asymptomatic individuals as unraveling their complete causal pathways may be impossible.

Руководитель кружка
PhD, и.о. доцента Жусупова А.И. 30 января 2014 г.