

Human Genetic Concepts for Health Care (3 credits)

Fall 2022 BIOL 666

Instructor: Dr. Yuliya Dobrydneva

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Office hours : TBA by appointment only

Class meets on Zoom Thursdays 7.30 -10.00 pm

Catalogue Description: Principles of genetically determined diseases with emphasis on clinical aspects of these diseases, genetic counseling, and laboratory methods used in human genetics. Record-enhancer and extended studies students preparing to enter Med- or Dental Schools are welcome and may find the course especially useful.

Course Objectives:

The course will integrate knowledge of genetic principles and framework of genetically determined human diseases with a special emphasis on the pathophysiological aspects of monogenic and multifactorial diseases. Students will learn to identify genetic factors, such as mutations and chromosomal abnormalities causing human disease; describe how these genetic changes underline pathophysiological mechanisms of disease; describe clinical correlations with underlying genetic defects; compare and contrast different modes of disease inheritance and manifestations.

Pre-requisite: B.S. degree completed or enrollment in accelerated Master's program. At least one Cell or Molecular Biology undergraduate course. *Not available to students who have taken BIOL 572.* Students in BIOS/BIOL program will be able to count either this course, either BIOL572 Human Genetics, but not both of them.

Text: Recommended, but not required: Medical Genetics by Ian D. Young, Oxford core texts, any edition.

- Additional reading: Thompson & Thompson Genetics in Medicine, 8th Edition
- Please stay tuned for additional reading as it may be posted on Bb. Announcements with updates will be posted regularly!

Where to find course materials: Content of all of your weekly works, including lectures, quizzes and other assignments will be posted on Bb under a tab “**Weekly sessions**”. Each week has its own folder (week 1, week 2 etc).

All the course materials, including lectures, assignments, assessments, quizzes, tests, discussions for each week will be placed in appropriate weekly sessions folder. All you need is in the weekly folder.

Important: All course communications will be conducted via Bb announcements. Please be sure that your mailbox is enabled to receive email messages from Bb.

- Please check Bb announcements regularly, at least twice a week.
- Not seeing the announcement is not an excuse for late/missing work.
- Please refer to a computer center for help!

Course evaluation and Grading:

Assessments, weighted evenly: include quizzes, discussions, essays, presentations etc	20%
Exam I	20%
Exam II	20%
Exam III	20%
Final Exam (cumulative)	20%

- All exams, assessments and quizzes are administered via Bb only! **Please be sure that you are proficient in basic functionality of Bb**
- All exams and quizzes are open book, open notes, open everything, and untimed
- Final exam will be administered during the finals week (TBA), according to the university schedule.
- Important: **Exams are not repeatable. When you sign up for this class, you are committing to come to the class for exams.**

Grading:

- Students will receive a letter grade based on a 100-pointscale.
- An A+ is not awarded as a final grade.
- **No extra credit is allowed in this course!**

Letter grades for the course will be based on an overall % for the course and assigned as follows:

>92 A
>90 A-
>87 B+
>84 B
>80 B-
>77 C+
> 70 C
> 60 C-
F below 60

Grades will not be rounded up. Therefore, if you receive a grade of 79.999, your grade will be C+, not a B-.

Expectations:

- Let Dr. Dobryднеva know of your needs and constraints as early as possible **prior** to the assignment due dates.
- Notify Dr. Dobryднеva during the first week of the semester regarding course schedule conflicts due to military obligations and/or religious observances.
- **Any notifications beyond the first week of classes or after the exam/quiz due date has passed may not be approved and grade of zero will be entered.**
- Students are expected to maintain an atmosphere of professionalism, mutual respect, positive attitude and academic integrity towards course faculty and fellow classmates, and to follow the university Honor code.

LATE POLICY: PLEASE READ CAREFULLY!

- Students should make every effort to submit/complete assignments on time.
- **Any assignment that is not turned in on time, without prior arrangements with the instructor, may result in a zero grade for this assignment**
- Any arrangements to extend a deadline for an assignment must be made **prior** to the deadline.
- No extension will be granted after the deadline.
- Each student is allowed maximum of two (2) extensions per semester. Other requests may not be granted and grade of zero will be entered
- If requesting an extension, please provide medical/employment documentation to be considered
- Any assignment/assessment that has not been completed by the end of the semester will receive a grade of zero.
- Note that there are no breaks/vacations in the online course.
- In the event of major holidays course will proceed as planned

Required Equipment –Since this is an online course and all assessments are complete through the computer, you need to have a reliable internet connection.

Technology Requirements: *It is a student's responsibility to ensure access to a reliable internet for quizzes, exams and lectures.*

- **Hardware:** You will need access to a Windows or Macintosh computer with at least 2GB of RAM and access to a fast and reliable broadband internet connection (e.g., cable, DSL). A larger screen is recommended for better visibility of course material. You will need speakers or headphones to hear recorded content and a headset with a microphone is recommended for the best experience. For the amount of Hard Disk Space required, when taking a distance education course, consider and allow for:
 1. the storage amount needed to install any additional software and
 2. space to store work that you will do for the course.

If you consider the purchase of a new computer, please go to [Patriot Tech](#) to see recommendations.

- **Software:** Many courses use Blackboard as the learning management system (LMS). You will need a browser and operating system that are listed compatible or certified with the Blackboard version available on the [myMason Portal](#). See [supported browsers and operating systems](#). Login to [myMason](#) to access your registered courses. Some courses may use other learning management systems. Check the syllabus or contact the instructor for details. Online courses typically use [Acrobat Reader](#), [Flash](#), [Java](#), and [Windows Media Player](#), [QuickTime](#) and/or [Real Media Player](#). Your computer should be capable of running current versions of those applications. Also, make sure your computer is protected from viruses by downloading the latest version of Symantec Endpoint/Anti-Virus software for free [here](#).

- Students owning Macs or Linux should be aware that some courses may use software that only runs on Windows. You can set up a mac computer with Boot Camp or virtualization software so Windows will also run on it. Watch [this video](#) about using Windows on a Mac. Computers running Linux can also be configured with virtualization software or configured to dual boot with Windows.
Note: If you are using an employer-provided computer or corporate office for class attendance, please verify with your systems administrators that you will be able to install the necessary applications and that system or corporate firewalls do not block access to any sites or media types.

○ **Schedule Fall 2022 Genetics of Human disease Biol 666**
Schedule may be subject to change! Please stay tuned for the announcements.

Week	Dates	Assignment
Week 1	August 25	Week 1 Introduction Bb discussion
Week 2	September 1	Genes and mutations Weekly quiz
Week 3	September 8	Hemoglobin Weekly quiz
Week 4	September 15	Cystic fibrosis Weekly quiz
Week 5	September 22	Exam 1
Week 6	September 29	Multifactorial disease Weekly quiz
Week 7	October 6	Alzheimer Weekly quiz
Week 8	October 13	Diabetes Weekly quiz
Week 9	October 20	Exam 2
Week 10	October 27	Mitochondrial disease Weekly quiz
Week 11	November 3	Primary immune deficiency Weekly quiz
Week 12	November 10	Pharmacogenomics Weekly quiz
Week 13	November 17	Exam 3
Week 14	December 1	No new material Reading day
Week 15 Finals week	December 8	Final Exam (TBA)

