Division of Life Science
The Hong Kong University of Science and Technology

LIFS 4370 Human Genetics and Personalized Medicine
Fall semester, 2018-2019

Instructors: Dr. Ho Yi MAK (E-mail: hym@ust.hk) (course coordinator)
Dr. Tom CHEUNG (E-mail: tcheung@ust.hk)

Time and Venue: Monday & Wednesday 12:00 - 13:20 Room 4620

Course Description
Credits: 3
Pre-requisite: LIFS 2210 or LIFS 3140

Many variations in the human population such as hair thickness, tolerance to milk in adults, high blood cholesterol and susceptibility to certain types of cancer can be determined by genetic factors. This course will cover the principles and up to date technologies for the discovery and analysis of human genetic variations. The application of basic scientific knowledge in a clinical setting will be discussed.

Intended Learning Outcomes
On successful completion of this course, students are expected to be able to:

1. Interpret the mode of inheritance of genetic traits and diseases based on family history and genomic data.
2. Explain and differentiate the technologies used for the diagnosis of diseases in a clinical laboratory setting.
3. Evaluate genetic data published in international journals and assess their relevance to specific human diseases.
4. Work as a member of a group to gather relevant information and devise strategies to solve a family medical mystery.
5. Present the findings of the group project in oral presentations.

Teaching approach
The primary delivery mode of the course will be lectures and small group discussions, supplemented with the use of personal response systems and videos. Basic concepts in genetics will be reviewed, followed by case studies of human genetic variations. Students are required to work in teams to apply their skills in database mining, literature review and analysis to solve a family medical mystery. The results will be summarized in a group oral presentation, in which peer participation will be expected.

Assessment scheme

Assessment Task	Percentage	Intended Learning Outcomes assessed
Final written exam [a]	65%	1, 2, 3
Data mining assignment [b]	20%	1, 2, 3
Group project oral presentation [c]	10% (group)	3, 4, 5
	5% (individual)	3, 4, 5

a. Open-book, 3 hours.

b. Students will choose from a list of topics (available by October 3) and the assignment is due on December 5. The report should be no longer than 4 pages of text (12 pt font size, Times New Roman, single space, 1-inch page margins, inclusive of a maximum of 15 references) and 1 page of illustrations (e.g. screen shots of the NCBI Variation Viewer).

c. Groups of students will collaborate on the oral presentation. Students should form groups by September 19 and assignment of the time of presentation will be drawn on the same day.

Assessment rubrics

Data mining assignment

	Needs improvement	Good	Excellent
Summarizes phenotypes of the disease **5%**	Does not consult publicly available database or primary literature relevant to the disease.	Reviews publicly available database or primary literature relevant to the disease.	Reviews publicly available database or primary literature relevant to the disease and identifies potential gap in knowledge, e.g. prevalence in ethnic groups.
Summarizes variant level evidence **5%**	Does not consult publicly available database.	Reviews one or more publicly available database.	Reviews one or more publicly available database and shows clear logic in relating the disease with genetic variations.
Summarizes gene level evidence **5%**	Failure to relate gene function to the disease.	Correctly relate gene function to the disease.	Correctly relate gene function to the disease and explore additional genes that may cause the same disease.
Appropriate use of language **5%**	Direct copying of passages without citing the source. Recurrent typographical or grammatical errors.	Mostly appropriate incorporation of source material by paraphrasing. Occasional typographical or grammatical errors.	Appropriate incorporation of source material by paraphrasing. No typographical or grammatical errors.

Additional assessment rubrics will be discussed at the beginning of the course.

Student learning resources

Course material will be drawn from the primary literature and the following reference books.

"The Human Genome: A User's Guide, 3rd edition" by Julia E. Richards and R. Scott Hawley

"Human Evolutionary Genetics, 2nd edition" by Jobling *et al*

Course schedule

Week	Date	Topic	Instructor
1	3-Sep-18	Overview of human genetics	Mak
	5-Sep-18	How mutations alter function I	Mak
2	10-Sep-18	How mutations alter function II	Mak
	12-Sep-18	Mitochondrial defects	Mak
3	17-Sep-18	Human population genetics	Mak
	19-Sep-18	Imprinting	Mak
4	24-Sep-18	Technologies for genetic variation discovery I	Cheung
	26-Sep-18	Technologies for genetic variation discovery II	Cheung
5	1-Oct-18	Public holiday	
	3-Oct-18	Prenatal genetic screening: at the bench	Cheung
6	8-Oct-18	Case study I: failure to perceive the environment	Mak
	10-Oct-18	Case study I: failure to perceive the environment	Mak
7	15-Oct-18	Case study I: *group presentation*	Mak
	17-Oct-18	Public holiday	
8	22-Oct-18	Case study II: unwanted reaction to specific food	Cheung
	24-Oct-18	Case study II: unwanted reaction to specific food	Cheung
9	29-Oct-18	Case study II: *group presentation*	Cheung
	31-Oct-18	Case study III: muscular dystrophy	Cheung
10	5-Nov-18	Case study III: muscular dystrophy	Cheung
	7-Nov-18	Case study III: *group presentation*	Cheung
11	12-Nov-18	Case study IV: susceptibility to cancer	Cheung
	14-Nov-18	Case study IV: susceptibility to cancer	Cheung
12	19-Nov-18	Case study IV: *group presentation*	Cheung
	21-Nov-18	Case study V: human with too much or too little fat	Mak
13	26-Nov-18	Case study V: human with too much or too little fat	Mak
	28-Nov-18	Case study V: group presentation	Mak