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

LIFS 4370 Human Genetics and Personalized Medicine
Fall semester, 2016-2017

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

Time and Venue: Tuesday & Thursday 10:30 - 11:50 Room 2502

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 writing and 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. Clinicians will be invited to give guest lectures and provide “bench to bedside” perspectives on medical genetics. 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 written report and 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]	60%	1, 2, 3
Data mining assignment [b]	15%	1, 2, 3
Group project oral presentation [c]	10% (group)	3, 4, 5
	5% (individual)	3, 4, 5
Group project written report [d]	10%	2, 3, 4, 5

a. Open-book, 2 hours.
b. Students will choose from a list of topics (available by October 4) and the assignment is due on December 1.
c. Groups of students will collaborate on the oral presentation. Students should form groups by September 20 and assignment of the time of presentation will be drawn on the same day.
d. The written report is due on December 6. The report should be no longer than 5 pages of text (12 pt font size, Times New Roman, single space, 1-inch page margins, inclusive of references) and 1 page of illustrations. The report should serve as a summary of the oral presentation.

Assessment rubrics

Group project oral presentation

	Needs improvement	Good	Excellent
Summarizes phenotypes of the disease	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	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	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.

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	01-09-2016	Overview of human genetics	Mak
	06-09-2016	How mutations alter function I	Mak
	08-09-2016	How mutations alter function II	Mak
2	13-09-2016	Imprinting	Mak
	15-09-2016	Mitochondrial defects	Mak
3	20-09-2016	Human population genetics I	Mak
	22-09-2016	Human population genetics II	Mak
4	27-09-2016	Technologies for genetic variation discovery I	Cheung
	29-09-2016	Technologies for genetic variation discovery II	Cheung
5	04-10-2016	Prenatal genetic screening: at the bench	Cheung
	06-10-2016	Prenatal genetic screening: at the clinic	Mak
6	11-10-2016	Case study I: unwanted reaction to specific food	Cheung
	13-10-2016	Case study I: unwanted reaction to specific food	Cheung
7	18-10-2016	Case study I: *group presentation*	Cheung
	20-10-2016	Case study II: failure to perceive the environment	Cheung
8	25-10-2016	Case study II: failure to perceive the environment	Mak
	27-10-2016	Case study II: *group presentation*	Mak
9	01-11-2016	Case study III: muscular dystrophy	Cheung
	03-11-2016	Case study III: muscular dystrophy	Cheung
10	08-11-2016	Case study III: *group presentation*	Cheung
	10-11-2016	Case study IV: susceptibility to cancer	Cheung
11	15-11-2016	Case study IV: susceptibility to cancer	Cheung
	17-11-2016	Case study IV: *group presentation*	Cheung
12	22-11-2016	Case study V: human with too much or too little fat	Mak
	24-11-2016	Case study V: human with too much or too little fat	Mak
13	29-11-2016	Case study V: *group presentation*	Mak