Course title <English>	統計遺伝学基礎　I Introductory Genetics-Statistics I	Affiliated department, Job title,Name	Graduate School of Medicine Professor,YAMADA RIYOU

Target year	Professional degree students	Number of credits	2	Course offered year/period	2017/Irregular, First semester

Day/period	月６	Class style	Lecture	Language	Japanese and English

[Outline and Purpose of the Course]

Genetic information is to be used in clinics as a part of personalized medicine and precision medicine. The information is based on research results in genome and omics studies. Genetic tests are the portal of the data usage. This lectures, I and II, covers 4 typical genetic tests and focuses on statistical backgrounds of the tests so that the participants understand the validity and ambiguity of the results and feel ready to provide information on the genetic risk in clinical settings.

It is important to understand the above-mentioned items. However the field of genetic testings is rapidly changing and the students who finish these lectures should be able to follow the progress. One of the most useful skills to keep themselves updated for the progress is that they can generate simulated data sets and perform statistical methods on them by themselves. In order to accelerate this goal, the textbook of the lectures are written with "R" language, a statistical programming language.

This year's lectures provided by the unit of statistical genetics are most suitable for the beginners, compared to the lectures last year on statistical thinking and the lectures next year on mathematics for statistical genetics.

This lecture is open to all graduate students as a part of Kenkyuka-oudan programs.

[Course Goals]

Statistical backgrounds in the four cases ([A} Cases, below) should be understood with the concerning points ([B] Concerning points, below) and the results of genetic tests should be described in your own wordings.

[A] Cases
- Mendelian traits
- Cancer syndromes such as BRCA1/2
- Complex genetic traits (SNP-based)
- Sub-typing diseases with expression profiles

[B] Concerning points
- Difference between classical and NGS-era genetic diagnosis
- Patients have to select an option among very extreme options, e.g., prophylactic maspectomy.
- DTC (Direct-To-Consumer) genetic testing kits
- Selection of biomarkers for clinics and its validation

[Course Schedule and Contents]

The contents are covered by I in the first semester and II in the second. 1 and 2 will be the topics of I and the rest, II. However some of 3 and 4 might be handled in I and some of 1 and 2 might be in II.

1. Mendelian traits
Pedigrees

Continue to 統計遺伝学基礎　I (2)↓↓↓

統計遺伝学基礎　Ⅰ (2)

Genotypes and Phenotypes
NGS and Disease-responsible mutations

2. Cancer syndromes such as BRCA1/2
Basics
Risk assessment
Decision tools

3. Complex genetic traits (SNP-based)
Genetic models
Population, cohort
SNP 2x3 table and association tests
Multi-locus model

4. Transcriptome ~ Sub-typing diseases with expression profiles
Molecular profiling for cancers ~ Prognostic infromation
Differential expression analysis
Clustering and heatmap
Supervised clustering
Validation for its clinical use

[Class requirement]

Basics of biology and genetics are desirable but not compulsory if willing.
Needs a laptop PC with WiFi access. Knowledge/skill in computation is not necessary but the participants without them will need to spend time to get familiar at home along the course.
Participation to both I and II is desirable but not compulsory.

[Method, Point of view, and Attainment levels of Evaluation]

Discussion in the classes
Homeworks
Exam on the last day

[Textbook]

ryamada 『StatGenet2017Text: Use of Genetic Data in Clinics (English Edition)』（Kindle）ISBN: B01MRQM1CG（You can generate the textbook in html format: See http://d.hatena.ne.jp/ryamada22/20161120）

[Reference books, etc.]

（Reference books）
山田　亮『遺伝統計学の基礎』（オーム社）ISBN:978-4-274-06822-5

Continue to 統計遺伝学基礎　Ⅰ (3)↓↓↓

統計遺伝学基礎　Ⅰ (3)

（Related URLs）

https://statgenetkyotou.moodlecloud.com/course/view.php?id=17(Login with name=guestsan and password=guestsan)

[Regarding studies out of class (preparation and review)]

Usage of R in daily activities is highly recommended to facilitate the understainding of the course.
Homework every week.

（Others (office hour, etc.)）

English with some Japanese.

*Please visit KULASIS to find out about office hours.