STAT830

Statistical Methods in Bioinformatics

S1 Day 2018

Dept of Statistics

Contents

General Information

Unit convenor and teaching staff
Lecturer in charge
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Credit points
4

Prerequisites
Admission to MBiotech or MSc or MDataSc or GradDipBioTech or MBiotechMCom or MBioBus or MLabQAMgt or PGCertLabQAMgt or GradDipLabQAMgt or GradCertLabQAMgt or MConsBiol or GradDipConsBiol or MMarScMgt

Corequisites

Co-badged status

Unit description
This unit introduces the statistical and probabilistic concepts that are the basis for the study of bioinformatics. Topics include an introduction to probability and conditional probability, probability distributions, sampling distributions and an introduction to Markov processes. Particular attention is paid to how they relate to specific applications in the field of bioinformatics. A basic understanding of calculus will be an advantage.

Important Academic Dates

Information about important academic dates including deadlines for withdrawing from units are available at https://www.mq.edu.au/study/calendar-of-dates

Learning Outcomes

On successful completion of this unit, you will be able to:

Understand basic notions and fundamentals of Probability and Statistics.

Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.

Being able to apply probabilistic approach to genetic mapping and linkage analysis.

Understand basic properties of Markov Chains. Being able to recognise Markov processes and understand how they can be used in applications.

Be familiar with fundamental principles of statistical data modelling utilised in practice.

General Assessment Information

Tutorial Work

In the case when a student is unable to attend the tutorial due to unavoidable circumstances, the student must apply for special consideration via https://ask.mq.edu.au/

In the case of **a late submission** of a Tutorial Work**,** if no special consideration has been granted, 10% of the earned mark will be deducted for each day that a Tutorial Work, is late, up to a maximum of 50%. After 5 days, counted including weekends and public holidays, a mark of 0% will be awarded. NOTE: It is not the intention of this late penalty policy to cause a student to fail the unit when they have submitted their assignment no more than 5 days after the due date and they would have otherwise passed. In this case, if deductions for late submissions result in the final unit mark for a student being less than 50, when otherwise it would have been 50 or greater, the student's final mark will be exactly 50.

Test

You are permitted ONE A4 page of paper containing reference material printed or handwritten on both sides.

Calculators will be needed but must not be of the text/programmable type.

In the case when a student is unable to attend the test due to unavoidable circumstances, the student must apply for special consideration via https://ask.mq.edu.au/

Practical Test

The unit material distributed via iLearn is permitted. You may bring to the practical test a hard copy of your lecture and tutorial notes.

In the case when a student is unable to attend the test due to unavoidable circumstances, the student must apply for special consideration via https://ask.mq.edu.au/

Assessment Tasks

Name	Weighting	Hurdle	Due
Tutorial Work 1	10%	No	Week 3

Name	Weighting	Hurdle	Due
Test 1	20%	No	Week 5
Tutorial Work 2	10%	No	Week 7
Tutorial Work 3	10%	No	Week 9
Test 2	20%	No	Week 11
Practical Test	30%	No	Week 13

Tutorial Work 1

Due: **Week 3**
Weighting: **10%**

The worksheet will be handed out in the second hour of the tutorial and is to be submitted via iLearn site of the unit before 23:55 on the day of the tutorial.

For the tutorial work assessment conditions see **General Assessment Information**.

On successful completion you will be able to:

- Understand basic notions and fundamentals of Probability and Statistics.

Test 1

Due: **Week 5**
Weighting: **20%**

The Test will be held in the second hour of lecture time and will be 40 minutes long.

For the test conditions see **General Assessment Information**.

On successful completion you will be able to:

- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.

Tutorial Work 2

Due: **Week 7**
Weighting: **10%**

The worksheet will be handed out in the second hour of the tutorial and is to be submitted via iLearn site of the unit before 23:55 on the day of the tutorial.

For the tutorial work assessment conditions see **General Assessment Information**.

On successful completion you will be able to:

- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.

Tutorial Work 3

Due: **Week 9**
Weighting: **10%**

The worksheet will be handed out in the second hour of the tutorial and is to be submitted via iLearn site of the unit before 23:55 on the day of the tutorial.

For the tutorial work assessment conditions see **General Assessment Information**.

On successful completion you will be able to:

- Being able to apply probabilistic approach to genetic mapping and linkage analysis.

Test 2

Due: **Week 11**
Weighting: **20%**

The Test will be held in the second hour of lecture time and will be 40 minutes long.

For the test conditions see **General Assessment Information**.

On successful completion you will be able to:

- Being able to apply probabilistic approach to genetic mapping and linkage analysis.

Practical Test

Due: **Week 13**
Weighting: **30%**

The Practical Test will be held during tutorial time and will be 1 hour and 15 minutes long.

For the practical test conditions see **General Assessment Information**.

On successful completion you will be able to:

- Understand basic notions and fundamentals of Probability and Statistics.
- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.

- Being able to apply probabilistic approach to genetic mapping and linkage analysis.
- Understand basic properties of Markov Chains. Being able to recognise Markov processes and understand how they can be used in applications.
- Be familiar with fundamental principles of statistical data modelling utilised in practice.

Delivery and Resources

Classes

Lectures begin in Week 1. Tutorials begin in Week 2.

Students must attend two hours of lectures and two hours of tutorials per week.

The lecture notes will be made available on iLearn before the lecture.

Tutorial exercises will be set weekly and will be available on iLearn before the tutorial.

The timetable for classes can be found at https://timetables.mq.edu.au/2018/

iLearn

All unit related materials including lecture notes, tutorials and instructions for assessment tasks and administrative updates, will be posted on iLearn at

https://ilearn.mq.edu.au/login/MQ/

Software

The statistical software **R** will be used. This is a free software environment for statistical computing and graphics on UNIX platforms, Windows and MacOS and can be downloaded from the website

http://www.r-project.org/

Texts and materials

There is no required textbook for this unit.

Recommended reference sources:

1. W. P. Krijnen Applied Statistics for Bioinformatics using R, 2009.

http://cran.r-project.org/doc/contrib/Krijnen-IntroBioInfStatistics.pdf

2. S. Draghici Statistics and Data Analysis for Microarrays Using R and Bioconductor. Chapman & Hall/CRC Mathematical and Computational Biology, 2nd Edition, 2012.

3. P. N. Suravajhala. Your passport to a career in bioinformatics. New Delhi: Springer, 2013.

4. W. J. Ewens and G. R. Grant. Statistical Methods in Bioinformatics, an Introduction. Springer, 2005.

5. K. Lange. Mathematical and Statistical Methods for Genetic Analysis, Statistics for Biology and Health. Springer, 2002.

Unit Schedule

Weeks	Topics
W1	Introduction
W2	Discrete random variables and their characteristics
W3 - W5	Hardy-Weinberg Equilibrium (HWE); Departures from HWE; Statistical testing of HWE.
W6 - W7	Genetic mapping; Linkage analysis
W8-W9	Continuous random variables and their characteristics
W10 - W11	Hypothesis testing and its applications
W12-W13	Markov Chains and their applications

Policies and Procedures

Macquarie University policies and procedures are accessible from Policy Central (https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central). Students should be aware of the following policies in particular with regard to Learning and Teaching:

- Academic Appeals Policy

- Academic Integrity Policy
- Academic Progression Policy
- Assessment Policy
- Fitness to Practice Procedure
- Grade Appeal Policy
- Complaint Management Procedure for Students and Members of the Public
- Special Consideration Policy *(**Note:** The Special Consideration Policy is effective from 4 December 2017 and replaces the Disruption to Studies Policy.)*

Undergraduate students seeking more policy resources can visit the Student Policy Gateway (https://students.mq.edu.au/support/study/student-policy-gateway). It is your one-stop-shop for the key policies you need to know about throughout your undergraduate student journey.

If you would like to see all the policies relevant to Learning and Teaching visit Policy Central (https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central).

Student Code of Conduct

Macquarie University students have a responsibility to be familiar with the Student Code of Conduct: https://students.mq.edu.au/study/getting-started/student-conduct

Results

Results shown in *iLearn*, or released directly by your Unit Convenor, are not confirmed as they are subject to final approval by the University. Once approved, final results will be sent to your student email address and will be made available in eStudent. For more information visit ask.mq.edu.au.

Student Support

Macquarie University provides a range of support services for students. For details, visit http://students.mq.edu.au/support/

Learning Skills

Learning Skills (mq.edu.au/learningskills) provides academic writing resources and study strategies to improve your marks and take control of your study.

- Workshops
- StudyWise
- Academic Integrity Module for Students
- Ask a Learning Adviser

Student Services and Support

Students with a disability are encouraged to contact the Disability Service who can provide appropriate help with any issues that arise during their studies.

Student Enquiries

For all student enquiries, visit Student Connect at ask.mq.edu.au

IT Help

For help with University computer systems and technology, visit http://www.mq.edu.au/about_us/offices_and_units/information_technology/help/.

When using the University's IT, you must adhere to the Acceptable Use of IT Resources Policy. The policy applies to all who connect to the MQ network including students.

Graduate Capabilities

PG - Discipline Knowledge and Skills

Our postgraduates will be able to demonstrate a significantly enhanced depth and breadth of knowledge, scholarly understanding, and specific subject content knowledge in their chosen fields.

This graduate capability is supported by:

Learning outcomes

- Understand basic notions and fundamentals of Probability and Statistics.
- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.
- Being able to apply probabilistic approach to genetic mapping and linkage analysis.
- Understand basic properties of Markov Chains. Being able to recognise Markov processes and understand how they can be used in applications.
- Be familiar with fundamental principles of statistical data modelling utilised in practice.

Assessment tasks

- Tutorial Work 1
- Test 1
- Tutorial Work 2
- Tutorial Work 3
- Test 2
- Practical Test

PG - Critical, Analytical and Integrative Thinking

Our postgraduates will be capable of utilising and reflecting on prior knowledge and experience, of applying higher level critical thinking skills, and of integrating and synthesising learning and knowledge from a range of sources and environments. A characteristic of this form of thinking is

the generation of new, professionally oriented knowledge through personal or group-based critique of practice and theory.

This graduate capability is supported by:

Learning outcomes

- Understand basic notions and fundamentals of Probability and Statistics.
- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.
- Being able to apply probabilistic approach to genetic mapping and linkage analysis.
- Understand basic properties of Markov Chains. Being able to recognise Markov processes and understand how they can be used in applications.

Assessment tasks

- Tutorial Work 1
- Test 1
- Tutorial Work 2
- Tutorial Work 3
- Test 2
- Practical Test

PG - Research and Problem Solving Capability

Our postgraduates will be capable of systematic enquiry; able to use research skills to create new knowledge that can be applied to real world issues, or contribute to a field of study or practice to enhance society. They will be capable of creative questioning, problem finding and problem solving.

This graduate capability is supported by:

Learning outcomes

- Familiarity with special classes of discrete and continuous random variables and their distribution functions. Being able to evaluate probabilities of events, expected values and variances of random variables.
- Being able to apply probabilistic approach to genetic mapping and linkage analysis.
- Understand basic properties of Markov Chains. Being able to recognise Markov processes and understand how they can be used in applications.

Assessment tasks

- Test 1
- Tutorial Work 2

- Tutorial Work 3
- Test 2
- Practical Test

Changes from Previous Offering

Changes since First Published

Date	Description
21/02/2018	Duplicated entry for teaching staff.