

STAT730 Statistical Methods in Bioinformatics

S1 Day 2017

Dept of Statistics

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General Information

Unit convenor and teaching staff Lecturer Nino Kordzakhia nino.kordzakhia@mq.edu.au Room 611, L6, E7A Wally's Walk TBA

Credit points 4

Prerequisites Admission to MRes

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 the Probability theory in DNA sequencing 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

DISRUPTION TO STUDIES NOTIFICATION

It is a student's responsibility to notify the University of their circumstances. All students of the University have the right to provide notification of a Disruption to Studies that has affected an assessment.

To be eligible for Special Consideration, a student must notify the University of a serious and unavoidable disruption no later than five (5) working days of the assessment task date or due date. For notifications made more than five (5) working days after the assessment task date or due date, an additional assessment task may not be possible (refer Disruption to Studies Outcomes Schedule). All Disruption to Studies notifications are to be made online via the University's <u>Ask MQ</u> system and must include supporting documentary evidence. Notifications of Disruptions to Studies after 5 days will still be assessed, however they are more likely to have a remedy of Withdrawal without Academic Penalty applied if they are deemed serious and unavoidable.

IMPORTANT - FINAL EXAMINATION

Should you need to apply for *Disruption to Study* for your final examination due to unavoidable circumstances, you must make yourself available for the week of **July 24 – 28, 2017**. If you are not available at that time, there is no guarantee an additional examination time will be offered. Specific examination dates and times will be determined at a later date.

Assessment Tasks

Name	Weighting	Hurdle	Due
Tutorial work 1	10%	No	Week 3
Test	20%	No	Week 8
Tutorial work 2	10%	No	Week 11
Final Examination	60%	No	University exam timetable

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 handed in at the end of the tutorial.

In the case when a student is unable to attend the tutorial due to unavoidable circumstances, the

student must apply for *Disruption to Studies*.

On successful completion you will be able to:

• Understand basic notions and fundamentals of Probability and Statistics.

Test

Due: Week 8 Weighting: 20%

The Test will be held in the tutorial time and will be one hour long.

The test conditions will be similar to that of the final examination.

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 11 Weighting: 10%

The worksheet will be handed out in the second hour of the tutorial and is to be handed in at the end of the tutorial.

In the case when a student is unable to attend the tutorial due to unavoidable circumstances, the student must apply for *Disruption to Studies*.

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.

Final Examination

Due: University exam timetable Weighting: 60%

A three-hour final examination for this unit will be held during the University Examination period.

You are permitted ONE A4 page of paper containing reference material printed or handwritten on both sides. The page will not be returned at the end of the final examination.

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

You are expected to present yourself for examination at the time and place designated in the University Examination Timetable. The timetable will be available in Draft form approximately eight weeks before the commencement of the examinations and in Final form approximately four weeks before the commencement of the examinations at

https://students.mq.edu.au/study/exams-and-results/exam-timetables

The University has general rules that will apply to students in every exam they sit

https://students.mq.edu.au/study/exams-and-results/examinations

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 the Probability theory in DNA sequencing 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 http://www.timetables.mq.edu.au

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 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	Lecture Topics
W1	Introduction
W2	Discrete random variables and their characteristics
W3 - W5	Hardy-Weinberg Equilibrium (HWE); Departures from HWE; Statistical testing of HWE.
W6	Statistical problems in DNA sequencing
W7	14/04/17 - Public Holiday

Session 1 Recess: 17/04/17 - 30/04/17		
W8	Continuous random variables and their characteristics	
W10 - W11	Hypothesis testing and its applications	
W12	Markov Chains and their applications	
W13	Review	

Policies and Procedures

Macquarie University policies and procedures are accessible from <u>Policy Central</u>. Students should be aware of the following policies in particular with regard to Learning and Teaching:

Academic Honesty Policy http://mq.edu.au/policy/docs/academic_honesty/policy.html

Assessment Policy http://mq.edu.au/policy/docs/assessment/policy_2016.html

Grade Appeal Policy http://mq.edu.au/policy/docs/gradeappeal/policy.html

Complaint Management Procedure for Students and Members of the Public <u>http://www.mq.edu.a</u> u/policy/docs/complaint_management/procedure.html

Disruption to Studies Policy (in effect until Dec 4th, 2017): <u>http://www.mq.edu.au/policy/docs/disr</u>uption_studies/policy.html

Special Consideration Policy (in effect from Dec 4th, 2017): <u>https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policies/special-consideration</u>

In addition, a number of other policies can be found in the Learning and Teaching Category of 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/support/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 <u>eStudent</u>. For more information visit <u>ask.m</u> <u>q.edu.au</u>.

Student Support

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

Learning Skills

Learning Skills (<u>mq.edu.au/learningskills</u>) 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 <u>http://www.mq.edu.au/about_us/</u>offices_and_units/information_technology/help/.

When using the University's IT, you must adhere to the <u>Acceptable Use of IT Resources Policy</u>. 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.
- 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
- Tutorial work 2
- Final Examination

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 the Probability theory in DNA sequencing 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
- Tutorial work 2
- Final Examination

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

Assessment tasks

- Test
- Tutorial work 2
- Final Examination

Changes from Previous Offering