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1. Introduction

It is a pleasure for me to welcome you to your Junior Sophister year in Genetics. Our Department is relatively young (established in 1958) considering the age of the Trinity College. Nevertheless, in its short history it has been responsible for big discoveries such as the location of the gene behind retinitis pigmentosa and incubator of spin out companies such as IdentiGEN or Genable. Our graduates have gone on to positions very influential positions all over the world.

In your first year you will be exposed to core concepts of Genetics and more specialized subject be explored in the following year. Our philosophy has a strong focus on developing analytical skills. It will be important for you to learn different subject but also to think critically and be able to apply your knowledge to the solution of problems. We also believe that communication skills are an essential part on the education of our graduates and students are required to write scientific reviews and make oral presentations we have a module each year dedicated to improving your written and oral presentation.

You are probably aware of all the different activities that the University provides you beyond the normal academic program. I encourage you to participate in different activities and societies as this is a unique experience in your university years. In this respect, students from our department run the Genetics Society.

Finally, I would also like to make you aware that College offers to you a wide range of support services (personal, financial, career and academic). All this information is available in the College webpage. However, your personal tutor will be able to guide you should you require any such support.

With best wishes for the year ahead,
Juan Pablo Labrador,
JS Genetics Coordinator and
Director of Teaching and Learning, School of Genetics and Microbiology.

2. General Information

2.1 JS course structure

The Junior Sophister year is comprised of modules totaling 60 European Credit Transfer System credits (ECTS). There are 40 credits of core modules compulsory for all Genetics students and 20 credits of open modules and Trinity Electives spread equally over two semesters in the academic year. The different possible combinations are described in the following module structure table for Genetics.

| Human Genetics | | | | |
|--|--|--|--|--|
| Semester 1 | Semester 2 | | | |
| Core N | Modules | | | |
| MIU33302 - Molecular genetics I - Regulation of gene expression (5 credits) | GEU33301 Bioinformatics (5 credits) | | | |
| GEU33007 Molecular Genetics Laboratory (5 credits) | GEU33303 - Molecular genetics II - Genome structure and dynamics (5 credits) | | | |
| GEU33075 Evolutionary and Population Genetics (5 credits) | GEU33008 Analytical Genetics Laboratory (5 credits) | | | |
| GEU33285 Science Structure, Discussion and Presentation for Genetics (5 credits) | GEU33035 Genetic Analysis of Nervous Systems (5 credits) | | | |
| Open Modu | lles Scenario I | | | |
| GEU33045 Genomics and Systems Biology (5 credits) | GEU33215 Medical Genetics (5 credits) | | | |
| Trinity Elective (5 credits) | GEU33055 Developmental Genetics (5 credits) OR BIU33250 Introduction to Immunology and Immunometabolism (5 credits) OR BIU33475 Basis Neurobiology (5 credits) | | | |
| Open Medu | BIU33475 Basic Neurobiology (5 credits) les Scenario II | | | |
| GEU33045 Genomics and Systems Biology (5 credits) | GEU33215 Medical Genetics (5 credits) | | | |
| BIU33150 Biochemistry for Biosciences (5 credits) | Trinity Elective (5 credits) | | | |
| Open Modules Scenario III | | | | |
| GEU33045 Genomics and Systems Biology (5 credits) | GEU33215 Medical Genetics (5 credits) | | | |
| Trinity Elective (5 credits) | Trinity Elective (5 credits) | | | |

Please consider your Open module/Trinity Elective pattern choices carefully as changes will not be permitted. If necessary, please seek advice from the relevant Course Advisor

2.2 Lectures courses and attendance

Our aim during the Junior Sophister year is to provide you with a thorough grounding in the fundamentals of modern Genetics so that you will be well prepared for the challenges of the Senior Sophister year. We therefore expect you to attend all lectures and in addition, some modules have strict attendance policy with direct consequences as a student may be deemed non-satisfactory.

It is necessary to constantly be aware that timetable may be subject to change if external conditions and circumstances demand this. Please be advised that the schedule might slightly vary subject to Lecturers availability. Any changes will be communicated via Blackboard announcement/email.

2.3 Attendance at seminars

In addition to the lecture courses there are weekly departmental seminars scheduled for 1 p.m. on Fridays in the atrium (these will be announced a few days ahead of time). Whilst it is not compulsory to attend these seminars you are strongly recommended to do so.

2.4 Communication

Announcements will be made by emailing you at your **tcd.ie email address**. You must read this mailbox regularly or set it up to forward to an account that you do read. Your primary contact for each module is the module coordinator and for general queries the Course Coordinator.

2.5 Summer Vacation Research Experience outside Ireland

We encourage Junior Sophisters to gain experience working in a research laboratory during the summer vacation. Each year the Department awards 6 travel bursaries (4 Bill Vincent Awards, David McConnell award and James Watson award) on the basis of performance in the Senior Freshman exams, to enable students to carry out a vacation research project outside of Ireland. Prof. Matthew Campbell will advise interested students about placements in various research labs. However, it is the student's responsibility alone to arrange: air travel, Visas (where appropriate), work permits, and any insurance requirements. These arrangements should be made as far in advance as possible from the departure date – preferably in early January

2.6 Behaviour in the Department

We expect high standards of personal behaviour in the Department consistent with its professional status. Please do not invite students from other Departments or friends into the Smurfit Institute, and when you are in the building please keep the noise down. Alcohol and smoking are absolutely forbidden. Students are not permitted to go on the roofs of the buildings.

2.7 Safety

Please make sure that you have received and have read the Science Faculty Safety Manual. Remember also that you are responsible for your own safety and that you have a responsibility not to endanger others by your actions. (*Refer to Appendix IX Safety Statement*)

2.8 Prizes in Human Genetics

The following prizes are awarded annually to students who have excelled during the Junior Sophister year:

- a) <u>Leslie Bloomer Prize in Human Genetics</u> awarded to the best qualified student in Human Genetics (based on overall JS assessment results) who in addition wishes to carry out research in the summer prior to entering the Senior Sophister year.
- **b)** Mcclintock Prize in Human Genetics awarded to Sophister student who has excelled in oral presentation of a subject of his/her choice within the field of Human Genetics. This prize is awarded based on presentations made on the field trip.

<u>Gold medals</u> are awarded by the Board to candidates of the first class who have shown exceptional merit in assessments for their honours bachelor degree. To be eligible, candidates must pass each year which counts towards their degree result, on the basis of a single annual attempt (which includes deferrals), and achieve the overall degree mark specified for their programme, which is set at 75 per

cent or above. See https://www.tcd.ie/academicregistry/exams/gold-medals/ for individual programme thresholds.

Various studentships, scholarships, exhibitions, and other prizes are awarded to students on the results of honour and other examinations, provided that sufficient merit is shown. Monetary awards are sent directly to prize winners unless otherwise stated under the regulations for the particular prize. For details see Part D – Prizes and other awards (Calendar 2023/24)

A book prize is awarded to each candidate obtaining an annual result of an overall first class honours grade in an honour course. These prizes are not awarded in the Senior Sophister or final year. Book prizes may be collected from the Academic Registry by the award holder in person.

2.9 Passing the Junior Sophister Year

It is important that you aim to achieve high grades in your continuous assessments and exams because 30% of the marks obtained in the JS year will contribute directly to your Senior Sophister BA Moderatorship grade. Also, when **project and review topics for Senior Sophister year** are assigned next year, students with higher marks in JS year will tend to get their higher preference choices of topic.

The current regulations are included below (calendar 2023-24). <u>Please check the most recent version of the calendar for any updates (TCD Calendar):</u>

Progression regulations: Bachelor programmes

59 Some programmes with professional accreditation have received a derogation from specific regulations on progression by the University Council. The relevant programme entry provides these details. See www.tcd.ie/teaching-learning/academicaffairs/ug-prog-award-regs/derogations /byschool.php. In order to rise with their class, students must obtain credit for the academic year by satisfactory attendance at lectures and tutorials and by carrying out, submitting and sitting the required assessment components. In addition, students must pass the year by achieving, at a minimum, an overall creditweighted average pass mark for the year (40 per cent or 50 per cent, as per programme regulations) and either: (a) accumulate 60 credits by achieving at least the pass mark in all modules or (b) pass by compensation. All modules and components within modules are compensatable (except in particular professional programmes where compensation does not apply). To pass a year by compensation, in programmes that locate the

pass mark at 40 per cent, a student must

achieve the pass mark in modules carrying a minimum of 50 credits and obtain a module mark of at least 35 per cent in any remaining module(s). A student may accumulate a maximum of 10 credits at qualified pass where the mark lies between 35-39 per cent. To pass a year by

compensation, in programmes that locate the pass mark at 50 per cent, a student must achieve the

pass mark in modules carrying a minimum of 50 credits and obtain a module mark of at least 45 per cent in any remaining module(s). A student may accumulate a maximum of 10 credits at qualified pass where the mark lies between 45-49 per cent.

60 Progression is on an annual basis. Within a year students may carry failed modules from one semester to the next but not from one academic year to another; that is, they will not be able to rise to the next year of their programme until they have successfully

completed the preceding year(s). Students who have not passed their year are required to present for reassessment when: (a) they obtain in excess of 10 credits at qualified pass (i.e. marks between 35-39 per cent where the pass mark is 40 per cent; or 45-49 per cent where the pass mark is 50 per cent); (b) they fail any module (i.e. achieving marks below 35 per cent where the pass mark is 40 per cent; or below 45 per cent where the pass mark is 50 per cent); (c) they do not obtain an overall pass mark for the year; (d) any combination of (a) - (c) occurs.

61 If a student has achieved both fail and qualified pass grades at the first sitting or has exceeded the 10 credit limit allowed for compensation and is not

permitted to rise with their year, they must present for reassessment in all modules for which they obtained a fail and/or a qualified pass.

62 Different modalities of assessment to the first sitting are permitted in the reassessment session, as determined by the programme.

- 63 The same progression and compensation regulations as outlined above apply at the reassessment session. The overall credit-weighted average for the academic year will be calculated using the most recent marks achieved.
- **64** Students who fail to satisfy the requirements of their year at the reassessment session are required to repeat the year in full (i.e. all modules and all assessment components).
- **65** Students are permitted to repeat any year of an undergraduate programme subject to not repeating the same year more than once and not repeating more than two academic years within a degree course, except by special permission of the University Council.
- **66** The maximum number of years to complete an undergraduate degree is six years for a standard four-year programme and seven years for a five-year programme.

There is one reassessment session which is held at the beginning of Michaelmas term. Students are assessed in all failed modules from both semesters during the reassessment session. Students are not permitted to repeat successfully completed assessments or examinations in order to improve their performance. In exceptional circumstances such as illness, if a student does not attempt exams at the end semester, they can defer until the reassessment examining period. Applications to defer exams should be made to the Senior Lecturer's Office via your tutor.

You are strongly advised to submit all the lab reports during the year, and not to miss the continuous assessment tests, because <u>if you obtain a mark of < 35% in 3 or more modules you will be unable to progress</u> to the Senior Sophister year (you'll have to repeat the JS year or leave College). If you fail to submit coursework on time, you will get a mark of zero for it.

2.10 Textbook

We recommend Introduction to Genetic Analysis by Anthony J. F. Griffiths, Susan R. Wessler, Sean B. Carroll, and John Doebley (current edition is 12th edition, 2020).

Department's web address: http://www.tcd.ie/Genetics
Keys – from the Genetics Executive Officer, €10 deposit.

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

2.11 Course coordinator

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

Executive Officer: Alicia Vega genetics@tcd.ie

| Module code | Module | Coordinator | Email |
|-------------|---|---|------------------------|
| MIU33302 | Molecular genetics I - Regulation of gene | r genetics I - Regulation of gene Prof Kevin Mitchell | |
| | expression | Prof Carsten Kroger | Carsten.Kroeger@tcd.ie |
| GEU33301 | Bioinformatics | Dr Karsten Hokamp | KAHOKAMP@tcd.ie |
| GEU33303 | Molecular genetics II - Genome structure | Prof Frank Wellmer | WELLMERF@tcd.ie |
| | and dynamics | | |
| GEU33007 | Molecular Genetics Laboratory | Prof Matthew Campbell | CAMPBEM2@tcd.ie |
| GEU33008 | Analytical Genetics Laboratory | Prof Juan Pablo Labrador | LABRADOJ@tcd.ie |
| GEU33035 | Genetic Analysis of Nervous Systems | Prof Juan Pablo Labrador | LABRADOJ@tcd.ie |
| GEU33045 | Genomics & Systems Biology | Dr Michael Dolan | MJDOLAN@tcd.ie |
| GEU33055 | Developmental Genetics | Prof Seamus Martin | MARTINSJ@tcd.ie |
| GEU33075 | Evolutionary and Population Genomics | Prof Lara Cassidy | CASSIDL4@tcd.ie |
| GEU33285 | Science Structure, Discussion and presentation for Human Genetics | Prof Juan Pablo Labrador | LABRADOJ@tcd.ie |
| GEU33215 | Medical Genetics | Prof Jane Farrar | Jane.Farrar@tcd.ie |
| BIU33150 | Biochemistry for Biosciences | Prof D Nolan | denolan@tcd.ie |
| BIU33250 | Introduction to Immunology & | Dr Aisling Dunne | aidunne@tcd.ie |
| | Immunometabolism | | |
| BIU33475 | Basics of Neurobiology | Prof Gavin Davey | gdavey@tcd.ie |

NOTE: Minor changes to that described in this JS Genetics Handbook may be undertaken during the academic year and will be notified through email to students.

3. Semester 1 Modules

3.1 MIU33302 Molecular genetics I - Regulation of gene expression

1. Module Code MIU33302

2. Module Name Molecular genetics I - Regulation of gene expression

3. Semester taught4. Contact Hours30 hours

5. Module Personnel Kevin Mitchell, Carsten Kroger, Mani Ramaswami, Adrian Bracken

6. Learning Aims This module will examine the principles and processes of regulation of gene expression, in prokaryotes and eukaryotes. It will be anchored around the problems that organisms have to solve in order to survive. These include modulating the cellular economy and maintaining homeostasis in response to dynamically changing internal and external states. This is crucial in microbes for adapting to environmental change and in multicellular organisms for coordinating cellular differentiation and development and regulating cellular, tissue- level and organismal physiology. The module will cover mechanisms and principles of regulation of transcription, chromatin regulation, gene regulatory motifs and networks, epigenetics, mRNA splicing, mRNA turnover, translation, non-coding RNA functions, and protein folding and localisation.

7. Module content (subject to change):

| Week | Date & Time | Lecture Topic & Lecturer |
|------|---------------------------|--|
| 3 | Tue, 10 th Sep | Cellular cognition (regulating the cellular economy) (KM/CK) |
| | 12:00-13:00 | |
| 3 | Thu, 12 th Sep | Gene regulatory motifs (KM) |
| | 15:00-16:00 | |
| 3 | Fri, 13 th Sep | Gene regulatory networks (KM) |
| | 10:00-11:00 | |
| 4 | Tue, 17 th Sep | Dynamical systems and landscapes (KM) |
| | 12:00-13:00 | |
| 4 | Thu, 19 th Sep | Group project discussion (KM/CK) |
| | 15:00-16:00 | |
| 4 | Fri, 20 th Sep | Regulation of transcription in eukaryotes - I (MR) |
| | 10:00-11:00 | |
| 5 | Tue, 24 th Sep | Regulation of transcription in eukaryotes - II (MR) |
| | 12:00-13:00 | |
| 5 | Thu, 26 th Sep | Splicing as regulatory mechanism (MR) |
| | 15:00-16:00 | |
| 5 | Fri, 27 th Sep | RNA quality control (NMD) and mRNA turnover (MR) |
| | 10:00-11:00 | |
| 6 | Tue, 1 st Oct | Regulation of transcription in prokaryotes - I (CK) |
| | 12:00-13:00 | |
| 6 | Thu, 3 rd Oct | Regulation of transcription in prokaryotes - II (CK) |
| | 15:00-16:00 | |
| 6 | Fri, 4 th Oct | Regulation of transcription in prokaryotes - III (CK) |
| | 10:00-11:00 | |
| 7 | Tue, 8 th Oct | Tips on presentations (KM) |
| | 12:00-13:00 | |
| 7 | Thu, 10 th Oct | Group project meetings (KM/CK) |
| | 15:00-16:00 | |
| 7 | Fri, 11 th Oct | Group project meetings (KM/CK) |
| | 10:00-11:00 | |
| 8 | Tue, 15 th Oct | Chromatin biology - I (AB) |
| | 12:00-13:00 | |

| Chromatin highery II (AD) |
|--|
| Chromatin biology – II (AB) |
| |
| Chromatin biology – III (AB) |
| |
| Review Week |
| Non coding RNAs I (CK) |
| |
| Regulation of translation – I (CK) |
| |
| Non coding RNAs II (KM) |
| |
| Regulation of translation – II (KM) |
| |
| Protein folding and posttranslational modifications (KM) |
| |
| Protein localization (KM) |
| |
| Review / discussion (KM/CK) |
| |
| Group Presentations (KM/CK) |
| |
| |

NOTE: Venue Moyne lecture theatre

8. Learning Outcomes:

At the end of this module, students should:

- Have a working knowledge of the mechanisms of regulation of gene expression in Pro- and Eukaryotes.
- Understand the principles underpinning cellular cognition, homeostasis, regulation of the cellular economy, and multi-cellular development
- Be able to deploy this knowledge and understanding to address novel problems involving these topics.
- Know how to independently research a topic in the scientific literature.
- Have experienced the dynamics of a group project and presentation.

9. Recommended Reading List:

Anthony J.F. Griffiths et al. Introduction To Genetic Analysis. 12th edition. New York, NY: W.H. Freeman & Company, 2020.

Further reading on specialist topics will be provided during the presentation of the module.

10. Assessment Details:

50% continuous assessment (group project (50%) – group presentation.

50% end of semester exam (1.5-hour exam paper at the end of semester 1).

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

Group project:

Students will be placed into groups of four or five and given a topic related to the principles of the module. They will be expected to research the literature on this topic and prepare a short presentation to the class, using Powerpoint or similar software. The goal is for students to learn how to independently explore and assess the scientific literature, to work in a group towards a common goal, to get some experience in communicating effectively, and to deepen their understanding of the principles of the module by exploring a particular topic in detail. The group exercise will be assessed on the basis of the presentation.

End of semester exam:

The students will be asked to answer 1 essay-type question and 10 short answer questions (3-5 sentences).

11. Module Coordinators: Kevin Mitchell (Kevin.Mitchell@tcd.ie) Carsten Kröger (Carsten.Kroeger@tcd.ie)

3.2 GEU33007 Molecular Genetics Laboratory

1. Module Code GEU33007

2. Module Name Molecular Genetics Laboratory

3. Semester taught4. Contact Hours5 hours per week

5. Module Personnel Kevin Mitchell Mike Dolan

David Noone (+ 4 demonstrators)

6. Learning Aims This comprises a set of robust experiment-based projects in molecular genetics that have given us consistently good results in the Junior Sophister class environment for many years. The central theme of the module is: gene expression and its regulation.

Aims: To provide hands-on training and experience of widely used experimental strategies and techniques in molecular genetics/ molecular biology, which include: the isolation and purification of genomic and plasmid DNA; the polymerase chain reaction (PCR); the use of agarose and polyacrylamide gel electrophoresis in the analysis of DNA, RNA and proteins; genetic transformation of *E. coli*; gene cloning and analysis in plasmid vectors; *lacZ*, GUS and GFP reporter gene assays.

7. Module content: Programme of laboratory practicals

| Date | Project No. | Page | Day |
|------------|---|------------|--------|
| | Week 3 | | |
| Wed 11 Sep | Pr 1. Exploring gene regulation in <i>E. coli</i> : the <i>lac</i> operon | p5 | 1 of 5 |
| | Pr 2. Gene transfer and expression in plant cells Transformation of <i>Agrobacterium</i> with pC-TAK1 | p12 p13 | 1 of 6 |
| Fri 13 Sep | Pr 1. Induction of the lac operon: measuring lacZ expression Alpha-complementation and its significance in lacZ | p7 | 2 of 5 |
| | Week 4 | | |
| Wed 18 Sep | Pr 1. PCR amplification of <i>lac</i> operon sequences | p9 | 3 of 5 |
| | Pr 3. Exploring the genome and proteome of | p17 | 1 of 7 |
| | phage Growth and isolation of phage λ | p18 | |
| | Pr 2. Purification of <i>Agrobacterium</i> transformants | p13 | 2 of 6 |
| Fri 20 Sep | Pr 1. Agarose gel analysis of <i>lac</i> operon PCR products | p10 | 4 of 5 |
| | Pr 3. Recovery of precipitated λ virions | p18 | 2 of 7 |
| | Week 5 | | |
| Wed 25 Sep | Pr 3. Purification of λ virions on a CsCl step gradient | p18 | 3 of 7 |
| | Pr 2. Liquid culture of <i>Agrobacterium</i> (pC-TAK1) | p13 | 3 of 6 |
| | Pr 1. Sequence analysis of <i>lac</i> operon PCR products | p10 | 5 of 5 |
| | Pr 3. Extraction of phage λ DNA | p19 | 3 of 7 |
| Fri 27 Sep | Pr 2. Transformation of leaf cells via Agrobacterium | p13 | 4 of 6 |
| | Pr 3. Restriction digestion of phage λ DNA | p19 | 4 of 7 |
| | Week 6 | | |
| Wed 2 Oct | Pr 4. Creating a library of λ genes in a plasmid vector Isolation and gel analysis of pUC19 plasmid DNA | p23 p24 | 1 of 7 |
| | Pr 4. Restriction digestion of pUC19 | p25 | 1 of 7 |
| | Pr 3. Gel analysis and processing of λ DNA digests | p20 | 5 of 7 |

labradoj@tcd.ie

| | | | <u>oradoj@tcd.</u> |
|------------|--|--------|--------------------|
| Fri 4 Oct | Pr 4. Gel analysis and processing of pUC19 digests | p25 | 2 of 7 |
| | Pr 4. Ligation of pUC19 and λ DNA fragments | p26 | 2 of 7 |
| | Week 7 | | |
| Wed 9 Oct | Pr 3. SDS-PAGE analysis of λ virion proteins | p20 | 6 of 7 |
| | Pr 4. Preparation of `competent' <i>E. coli</i> cells and transformation with ligated pUC19-λ DNA | p27 | 3 of 7 |
| Fri 11 Oct | Pr 4. Inspection of transformation plates and inoculation of miniprep cultures | p27 | 4 of 7 |
| | Pr 3. Interpretation of SDS-PAGs of virion proteins | p21 | 7 of 7 |
| | Week 8 | | |
| Wed 16 Oct | Pr 4. Isolation of putative pUC19-λ recombinant plasmids Restriction digestion of plasmid DNAs | p27 | 5 of 7 |
| Fri 18 Oct | Pr 4. Agarose gel analysis of digested plasmid DNAs | p28 | 6 of 7 |
| | Assignment of the First Lab Report | | |
| | Week 9 | | |
| | Study Week (21 – 26 Oct) | | |
| | Week 10 | | |
| Wed 30 Oct | Pr 5. Exploring light-induced gene expression in plants | p30-31 | 1 of 4 |
| | Differential regulation of rRNA gene expression | | |
| | Pr 2. Detection of transgenes in regenerated leafy shoots a. Detection of the the <i>Kan^r</i> gene by PCR b. Detection of the GUS gene via reporter activity | p14 | 5 of 6 |
| Fri 1 Nov | Pr 5. Agarose gel analysis of rRNAs | p31 | 2 of 4 |
| | Pr 2. Agarose gel analysis of <i>Kan^r</i> PCR products | p15 | 6 of 6 |
| | Pr 4. Sequence analysis of pUC19-λ clones | p28 | 7 of 7 |
| | Week 11 | | |
| Wed 6 Nov | Pr 5. Exploring light-induced gene expression in plants: 2. Regulation of RUBISCO synthesis | p32-33 | 3 of 4 |
| | Pr 5. Exploring light-regulated gene expression in plants: 3. Regulatory properties of the <i>RbcS</i> promoter | p34-36 | 3 of 4 |
| Fri 8 Nov | Pr 5. Interpretation of RUBISCO gels Discussion: Regulation of gene expression by light | p33 | 4 of 4 |
| | Assignment of the Second Lab Report | | |
| | Week 12 | | |
| | Lab Report writing (13 - 17 Nov) | | |
| | | L | |

NOTE: Labs will run on Wed 14:00-17:00 and Fri 15:00-17:00. Venue Biolab

8. Learning Outcomes: On successful completion of the module, students will be able to:

- isolate and purify genomic and plasmid DNA;
- assemble the reagents required to amplify DNA using the polymerase chain reaction (PCR);
- analyse DNA and RNA using agarose gel electrophoresis;
- make protein extracts and analyse them using SDS-polyacrylamide gel electrophoresis;
- transform E. coli and Agrobacterium using plasmid DNA;
- grow, isolate and purify bacteriophage;
- clone DNA fragments in plasmid vectors;
- use Agrobacterium to introduce genes into the plant nucleus;
- perform *lacZ*, GUS and GFP reporter gene assays.
- 9. Recommended Reading List: published primary research papers and reviews

10. Assessment Details:

Students prepare two Laboratory Reports of equal weighting.

Students are notified of the first Laboratory Report during Week 6 on October 2th; and the second report during the final practical on November 8th.

The first report is due at 17:00 Irish Time, October 25th 2024.

The second report is due at 17:00 Irish Time, November 29th 2024.

11. Module Coordinator: Mike Dolan MJDOLAN@tcd.ie

Matt Campbell CAMPBEM2@tcd.ie

3.3 GEU33075 Evolutionary and Population Genetics

1. Module Code GEU33075

2. Module Name Evolutionary and Population Genetics

3. Semester taught Semester 1

4. Contact Hours Online: 23 hours; In-person tutorials: 7 hours 15 minutes

5. Module Personnel Lara Cassidy, Russell McLaughlin, Ross McManus

6. Learning Aims This module provides an in-depth exploration of genetic variation, from its origins to its evolutionary consequences. The information in DNA is not always transmitted accurately from one generation to the next. DNA sequences can change spontaneously by the process of mutation and inaccurate DNA repair, resulting in genetic variation (polymorphism) within populations. Variable sites at different positions in the genome get shuffled into new combinations by the process of genetic recombination that occurs during sexual reproduction. Whether a particular allele survives for a long time in a population or goes extinct depends on the evolutionary forces acting on the population. If a new allele is advantageous to the population, Darwinian natural selection will tend to increase its frequency in the population; alternatively, if the new allele is disadvantageous natural selection will tend to eliminate it. However, selection is only one of several evolutionary processes that change allele frequencies within populations over generations. In this module, students will learn about the origin of genetic variation, its distribution within populations and long-term changes brought about by evolutionary processes.

7. Module content

| Week | Day & Time | Lecture Topic & Lecturer |
|------|---------------------------|---|
| 3 | Mon 9 th Sept | Introduction to evolutionary and population genetics (Cassidy; in-person |
| | 10:00-11:00 | venue: LTEE3) |
| 3 | Mon 9 th Sept | Tutorial: Introduction (Cassidy and McLaughlin; in-person venue: LTEE3) |
| | 14:00-16:00 | |
| 3 | Mon 9 th Sept | RMM L1: Spectrum and mechanisms of DNA mutation I (McManus; pre-recorded) |
| | 8 am onwards | |
| 3 | Mon 9 th Sept | RMM L2: Spectrum and mechanisms of DNA mutation II (McManus; pre-recorded) |
| | 8 am onwards | |
| 3 | Mon 9 th Sept | RMM L3: Exogenous and structural causes of mutation (McManus; pre-recorded) |
| | 8 am onwards | |
| 3 | Mon 9 th Sept | RMM L4: Mutation and health (McManus; pre-recorded) |
| | 8 am onwards | |
| 4 | Mon 16 th Sept | RMcL L1: Genetic variation and its detection I (McLaughlin; pre-recorded) |
| | 8 am onwards | |
| 4 | Mon 16 th Sept | RMcL L2: Genetic variation and its detection II (McLaughlin; pre-recorded |
| | 8 am onwards | |
| 4 | Mon 16 th Sept | RMcL L3: Alleles and genotypes: Hardy-Weinberg equilibrium (McLaughlin; pre-recorded) |
| | 8 am onwards | |
| 4 | Mon 16 th Sept | RMcL L4: Alleles and genotypes: inbreeding (McLaughlin; pre-recorded) |
| | 8 am onwards | |
| 4 | Mon 16 th Sept | RMcL L5: Effective population size (McLaughlin; pre-recorded) |
| | 8 am onwards | |
| 4 | Tue 17 th Sept | Tutorial RMM L1-L5: (McManus); in-person venue: LTEE3) |
| | 14:00-17:00 | |
| 5 | Mon 23 rd Sept | RMcL L6: Genetic drift, fixation F-statistics and population structure |
| | 8 am onwards | (McLaughlin; pre-recorded) |
| 5 | Mon 23 rd Sept | RMcL L7: Linkage disequilibrium (McLaughlin; pre-recorded) |
| | 8 am onwards | |
| 5 | Mon 23 rd Sept | RMcL L8: Applied population genetics: complex traits and GWAS |
| | 8 am onwards | (McLaughlin; pre-recorded) |
| 5 | Mon 23 rd Sept | RMcL L9: Applied population genetics: population structure and PCA |
| | 8 am onwards | (McLaughlin; pre-recorded) |

| | | <u>labradoj@tcd.le</u> |
|----|---|---|
| 5 | Mon 23 rd Sept 9:00-11:00 | Tutorial RMcL L1-L5: McLaughlin; in-person venue: LTEE3 |
| 5 | Mon 23rd Sept 13:00-17:00 | Poster Tutorial in-person venue: Lara / Russell office TBC |
| 6 | Mon 30 th Sept 8 am onwards | LC L1: Evolutionary change in sequences: measuring evolution (Cassidy; pre-recorded) |
| 6 | Mon 30 th Sept 8 am onwards | LC L2: Evolutionary change in sequences: patterns and models (Cassidy; pre-recorded) |
| 6 | Mon 30 th Sept 8 am onwards | LC L3: Evolutionary change in sequences: the molecular clock (Cassidy; pre-recorded) |
| 6 | Mon 30 th Sept 8 am onwards | LC L4: Evolutionary change in sequences: molecular phylogenetics (Cassidy; pre-recorded) |
| 6 | Mon 30 th Sept 8 am onwards | LC L5: Evolutionary change in sequences: applied phylogenetics (Cassidy; pre-recorded) |
| 6 | Mon 30 th Sept 9:00-11:00 | Tutorial RMcL L6-L9: McLaughlin; in-person venue: LTEE3 |
| 7 | Mon 7 th Oct 8 am onwards | LC L6: Genome evolution: gene duplication (Cassidy; pre-recorded) |
| 7 | Mon 7 th Oct 8 am onwards | LC L7: Genome evolution: concerted evolution (Cassidy; pre-recorded) |
| 7 | Mon 7 th Oct 8 am onwards | LC L8: Genome evolution: whole genome duplication (Cassidy; pre-recorded) |
| 7 | Mon 7 th Oct 8 am onwards | LC L9: Genome evolution: gene loss and base composition evolution (Cassidy; pre-recorded) |
| 7 | Mon 7 th Oct 8 am onwards | LC L10: Genome evolution: Transposition (Cassidy; pre-recorded) |
| 7 | Mon 7 th Oct 9:00-11:00 | Tutorial LC L1-L5: Cassidy; in-person venue: LTEE3 |
| 7 | Mon 7th Oct 14:00-17:00 | Poster Tutorial in-person venue: Lara / Russell office TBC |
| 8 | Thur 17 th Oct 9:00-11:00 | Project group tutorials in-person venue: LTEE3 |
| 9 | | Study/Review week |
| 10 | Tue 29 th Oct 15:00-16:00 | Tutorial LC L6-L10: Cassidy; in-person venue: LTEE3 |
| 11 | Mon 4 th Nov 9:00 | Project hand-in deadline online submission 9:00am |
| 12 | Wed 13 th Nov 14:00-17:00 | Project presentation (Atrium at the Smurfit) |
| 12 | Fri 15 th Nov 17:00 | Reflective report online submission 5:00 pm |
| 14 | Thu 28 th Nov 10:00-12:00 | MCQ Exam venue (MAC LAB) |

NOTE: Groups for tutorials will be facilitated by course coordinator.

Description of each Lecture

• Introduction to evolutionary and population genetics (Cassidy)

This lecture introduces molecular evolution and provides historical context, including ideas presented by Lamarck, Weissmann and Darwin. Core principles, including selection and neutral mutations, are also introduced.

- Spectrum and mechanisms of DNA mutation I (McManus)
- Spectrum and mechanisms of DNA mutation II (McManus)

These two lectures explore the different ways that DNA can change (mutate) between individuals, leading to genetic variation within a population and a substrate on which evolution

acts. Types of DNA mutation are described, along with the molecular biological mechanisms bringing about these changes.

• Exogenous and structural causes of mutation (McManus)

This lecture extends our view of the origin of mutation beyond molecular mechanisms to include exogenous (eg environmental) factors and structural changes.

Mutation and health (McManus)

The consequences of mutation on human health are discussed.

- Genetic variation and its detection I (McLaughlin)
- Genetic variation and its detection II (McLaughlin)

These two lectures describe in detail some core methodologies and technologies used in the identification and study of genetic variation in DNA. Methods include polymerase chain reaction, electrophoresis, Sanger sequencing, whole-genome SNP genotyping and next-generation sequencing. Technologies are also compared and contrasted for their relative advantages and constraints in the context of conducting population genetic studies on genetic variation.

• Alleles and genotypes: Hardy-Weinberg equilibrium (McLaughlin)

In this lecture, we define the expected relationship between allele and genotype frequencies in an idealized population, and the interpretation when observation deviates from expectation. Statistical methods for assessing the probability of observed data under a null model are detailed, and the conclusions that can be drawn from the results of these tests are discussed.

• Alleles and genotypes: inbreeding (McLaughlin)

The concept of inbreeding (mating between related individuals) is introduced and our model for expected genotype frequencies under various inbreeding scenarios is updated. This forms the basis of ensuing lectures.

• Effective population size (McLaughlin)

In this lecture we state how populations are modelled as idealized populations with effective sizes that can – sometimes counterintuitively – be far from actual census sizes. We discuss the various parameters that affect effective population size and their consequences in the population, including resilience in agriculture and animal breeding.

• Genetic drift, fixation, F-statistics and population structure (McLaughlin)

Here we explore genetic drift (the random long-term change in allele frequencies) and the ultimate long-term fate for all genetic variation: either loss or fixation. We explore the role of parameters such as population size on defining the rate of fixation of an allele in a population. We then set a statistical framework for the analysis of variance of genotype frequencies in a population and their use in defining the fixation index, a useful tool for identifying genetic structure (non-random patterns) in populations.

Linkage disequilibrium (McLaughlin)

This lecture introduces the new concept of linkage disequilibrium – the statistical correlation of adjacent alleles – and its uses in modern genomics.

- Applied population genetics: complex traits and GWAS (McLaughlin)
- Applied population genetics: population structure and PCA (McLaughlin)

In these final three lectures, we apply some of the techniques and methods that have been discussed in previous lectures to understand the genetic basis of traits in a population and genomic descriptors of population differentiation. We explore the design and typical execution of a genome-wide association study (GWAS), from its motivation to the statistical considerations in controlling false positives and technical bias. We qualitatively discuss the application of principal component analysis (PCA) in delineating population structure and

controlling GWAS, and finish with a tour of modern, population-scale genome sequencing studies such as the UK Biobank.

• Evolutionary change in sequences: measuring evolution (Cassidy)

In this lecture we learn how biological entities are compared on the molecular level and introduce the concept of molecular homology. In part two, we discuss the evolutionary forces that cause related sequences to diverge through time. We define a substitution event and introduce the molecular clock and neutral theory.

• Evolutionary change in sequences: patterns and models (Cassidy)

In this lecture we consider evolution as a series of substitution events and introduce substitution models, which can be used to describe the patterns and rates of sequence evolution. We discuss the assumptions of these models and violations to them. The concept of functional constraint is introduced, as well as the Ka/Ks ratio method for detecting the action of purifying and positive selection on protein-coding sequences.

• Evolutionary change in sequences: the molecular clock (Cassidy)

The concept of the molecular clock and its applications are explored in more detail. We learn how to calibrate a molecular clock with an outgroup and use it to estimate species divergence times. The core assumption of the clock - a constant rate of substitution – is explored and the causes of non-constant rates of evolution across lineages are introduced.

• Evolutionary change in sequences: molecular phylogenetics (Cassidy)

This lectures explains the construction of phylogenetic trees from sequence data to describe the evolutionary relationships between species. The aims of the field of phylogenetics are outlined and students are given a refresher in phylogenetic terminology and the concept of reproductive isolation. Methods for reconstructing and rooting trees are described in turn. Methods for assessing tree reliability are also given.

Evolutionary change in sequences: applied phylogenetics (Cassidy)

This lecture provides examples of real-world applications of phylogenetics. This includes the characterization of the last common ancestor of all life and the closest living relatives of cetaceans (whales and dolphins). Viral phylogenetics is also introduced.

• Genome Evolution: Gene Duplication (Cassidy)

This lecture answers the question: 'where do new genes come from?' The mechanisms and evolutionary consequences of gene duplication are discussed. In part two, several examples of gene duplication driving phenotypic evolution are provided (globins, olfactory receptors and primate opsins).

• Genome Evolution: Concerted evolution (Cassidy)

This lecture defines and explores concerted evolution, the process which results in the non-independent evolution of genes that are closely related in sequence. Duplicated genes within a species may remain similar due to the horizontal spread of mutations by unequal crossing over and gene conversion. Both mechanisms are discussed in turn. The concerted evolution of rRNA repeats, globin genes in primates, and ribonucleases in ruminants are given as examples.

Genome Evolution: whole genome duplication (Cassidy)

The concept of polyploidy is defined and its incidence in the natural world explored. The mechanisms and evolutionary consequences of true polyploidy are discussed. In part two, we learn about the process of diploidization, whereby a polyploid species reverts to a diploid state through structural rearrangements of the genome and gene loss.

Genome Evolution: gene loss and base composition evolution (Cassidy)

This lecture covers two distinct topics. In part one, we learn about the mechanisms and consequences of gene loss. We ask why a single copy gene may be lost from a species and cover

several examples of gene loss in humans and other species. In part two, we learn about base composition - the proportions of the four bases within DNA. We ask why GC content differs across species and discuss both mutational and selection-based hypotheses for this variation. We also look at GC and AT skews on the two strands of the DNA helix, as well as codon usage hias

Genome Evolution: transposition (mobile genetic elements) (Cassidy)

Transposition is discussed in this lecture – the process by which genetic elements are transferred "horizontally" between organisms rather than the more conventional "vertical" inheritance of genetic material from parent to offspring. Types of transposable elements are introduced and their prevalence in nature outlined. The evolutionary consequences of transposition are also discussed.

8. Learning Outcomes

On completion of this module, students will be able to:

- Describe and explain the origin of genetic variation through mutation
- Describe and explain the consequences of genetic variation in health and disease
- Identify suitable technological approaches to the detection and study of different types of genetic variation
- Describe the relationship between population genotype and allele frequencies under panmixia and inbreeding
- Define effective population size (N_e), identify population parameters that influence N_e and describe practical consequences of low N_e even in large census populations
- Specify the long-term evolutionary trajectories of alleles under genetic drift and define fixation
- Define and quantitatively describe population structure using F-statistics
- Define linkage disequilibrium and describe its use in population genomics
- Explain the design of genome-wide association studies for complex traits and their use of population genetic principles to control bias
- Discuss modern population-scale genome sequencing efforts to understand human health and disease
- State the neutral theory of molecular evolution
- Describe the nature and consequences of evolutionary change in DNA and protein sequences, including codon usage bias
- Define the relationship between mutation accumulation and evolutionary time and its use in the molecular clock
- Construct phylogenetic trees based on evolutionary relationships between related sequences
- Define homolog, paralog, ortholog and ohnolog and describe concerted evolution of paralogous genes within species
- Describe large-scale evolutionary changes to genetic sequences, including transposition, gene and genome duplication and gene loss

9. Recommended Reading List

Introduction to Genetic Analysis, 11th ed. (Griffiths, Wessler, Carroll, Doebley) – chapters 15-18 Principles of Population Genetics, 4th ed. (Hartl & Clark)

10. Assessment Details

This module will be graded through an invigilated MCQ examination (60%) on 28th Nov (MAC Lab) and continual assessment (40%).

The continual assessment is subdivided into:

Group poster project (35%) submission on 4th Nov and presentation on 13th Nov, which includes an individual contribution mark, and a short reflective essay (5%) submission 15th Nov.

For the MCQ, each student will receive a random set of questions that capture the entire breath of the module. These questions will be designed to test for both quick recall and reasoning skills. Students may be asked to solve mathematical problems, interpret tabular data and phylogenetic diagrams. There will be no negative marking.

For the group project, students will be placed in groups of 3-4 individuals and asked to produce a scientific poster that addresses a specific research question that is currently relevant or under debate in the field of evolutionary and population genetics. Please note that this is not the general format of a scientific poster, which typically is a presentation of a specific research project carried out by an individual or team. The assessment poster is better thought of as a mini-literature review. Students will present their poster at an in-person event in the Smurfit Institute of Genetics.

When you have completed your poster, each group member will write a succinct reflective report that critically reflects on their involvement in the project and evaluate what was produced by your team.

11. Module Coordinator: Lara Cassidy (cassidl4@tcd.ie)

3.4 GEU33085 Science Structure, Discussion and Presentation for Genetics

1. Module Code GEU33285

2. Module Name Science Structure, Discussion and Presentation for Human Genetics

3. Semester taught 14. Contact Hours 12

5. Module Personnel Adrian Bracken, Dan Bradley, Matthew Campbell, Lara Cassidy, Mike Dolan, Jane Farrar, Seamus Martin, Kevin Mitchell, Juan Pablo Labrador, Aoife Mc Lysaght, Russell Mc Laughlin, Mani Ramaswami, Frank Wellmer.

6. Learning Objectives: The module has the following learning objectives: (1) discussion of the design and implementation of genetic analysis of biological phenomena relevant to human genetics; (2) discussion of genetic analysis in cell and animal models - their strengths and weaknesses; (3) discussion of mathematical genetics; (4) discussion of how to research, design and write a literature review on a human genetics subject; (5) to write a 4,000 literature review on an assigned human genetics topic; (6) discussion of science communication: how to assemble and present a talk on a human genetics topic. Students will make a 10 minute presentation on a paper/review each week (week 4-7) within the topic of the tutorial.

7. Module content: Programme of tutorials

| Week | | Programme of Tutorials GEU33285 |
|------|---------------------------|--|
| 3 | Tue 10 th Sept | Review I: How to write a literature review on a genetics/human genetic topic |
| | 14:00-16:00 | (Labrador & Cassidy) LTEE3 |
| 3 | Wed 11 th Sept | Module introduction (Labrador) |
| | 10:00-11:00 | |
| 3 | Wed 11 th Sept | How to make an oral presentation (Labrador) LTEE3 |
| | 11:00-12:00 | |
| 3 | Thu 12 th Sept | Review I: How to write a literature review on a genetics/human genetic topic |
| | 9:00-11:00 | (Labrador & Cassidy) LTEE3 |
| 3/4 | One to one meeting | Review II: Supervisor and student meet to discuss the topic and how to structure |
| | with supervisor | the review. (student to reach out supervisor and organise meeting) |
| 4 | Wed 18 th Sept | Human Genetic Analysis I: In search of human disease genes (Labrador & |
| | 9:00-12:00 | Ramaswami) |
| 5 | Wed 24 th Sept | Human Genetic Analysis II: Understanding gene function (Labrador & |
| | 9:00-12:00 | Ramaswami) |
| 6 | One to one meeting | Review III: Supervisor and student meet to discuss review structure produced by |
| | with supervisor | the student. |
| 6 | Wed 2 nd Oct | Human Genetic Analysis III: Studying mechanisms (Labrador & Ramaswami) |
| | 9:00-12:00 | |
| 7 | Wed 9 th Oct | Human Genetic Analysis IV: Emerging methodologies in elucidating the molecular |
| | 9:00-12:00 | basis of human disease (Labrador & Ramaswami) |
| 9 | | Reading Week |
| 10 | Wed 30 th Oct | Mathematical Genetics I: Networks/graph theory (Bradley & Mc Lysaght) |
| | 10:00-12:00 | |
| 12 | Wed 13 th Nov | Mathematical Genetics II: Game theory: Evolution of co-operation/altruism |
| | 10:00-12:00 | (Bradley & Mc Lysaght) |
| 13 | Wed 20 th Nov | Mathematical Genetics III: Bayesian statistics (Bradley & Mc Lysaght) |
| | 10:00-12:00 | |
| 14 | Mon 25 th Nov | Essay Assignment submission |
| | 9:00 | |
| 14 | Wed 27 th Nov | Assignment Feedback (Bradley & Mc Lysaght) |
| | 10:00-12:00 | |
| 19 | Fri 3 rd Dec | Assignment written review submission |
| | 17:00 | |

NOTE: Venue: lectures Dawson/LTEE3

- Week 3 **Review I:** How to write a literature review on a genetics/human genetics topic: Discussion under the following headings: Topic / focus, structure of review, themes, progression of ideas, literature survey, search databases, references.
- Week 3: **How to make an oral presentation:** Discussion of how to prepare a talk, focusing on the topic, judging your audience, slide preparation and the general do's and don'ts of preparing and giving an oral presentation. Discussion will be centred on "Giving a seminar: Suggestions for Graduate Students" by John Roth.
- Week 3/4 Review II: Discussion of the review topic assigned to each student and review with the supervisors. This is arranged by the supervisor / student at their mutual convenience.

Week 4 Human Genetic Analysis I: In search of human disease genes. Concepts discussed:

Understanding how mutations affect proteins in ways that determine whether they generate recessive or dominant phenotypes. Connecting terms – hypomorph, loss of function, amorph, null, neomorph, gain of function to different types of recessive and dominant phenotypes) Penetrance and expressivity

- Weeks 5 **Human Genetic Analysis II: Understanding gene function** Concepts discussed: somatic cell hybrids (rodent/huma),YAC, Cosmids and other techniques used classically (probes, southern northern). Discussion on pre-mutations, variable penetrance and semi-dominance How does one go from gene to understanding to treatment?
- Weeks 6 **Human Genetic Analysis III: Studying mechanisms** Discussion on mechanisms to understand gene function, use of model organisms, genetics, biochemical essays, cell biology. Understanding pathways. Focus on FMR1: functions and activities of dFMR1 in flies and mouse. What proteins does it bind? What RNAs does it bind and regulate?
- Week 7 Human Genetic Analysis IV: Emerging methodologies in elucidating the molecular basis of human disease Discussion of mouse and drosophila model of FMR1 Fragile X therapies (Reading will be provided before each tutorial)
- Week 10 Mathematical Genetics I Networks/graph theory
- Week 11 Mathematical Genetics II Game theory: Evolution of co-operation/altruism.
- Week 12 Mathematical Genetics III Bayesian statistics.
- Week 14 Mathematical Genetics IV Assignment Feedback
- **8. Learning Outcomes:** Students will have the following skills: (1) to research and write a scientific literature review; (2) to research, prepare and give a scientific oral presentation; (3) to design and perform a genetic analysis of a biological phenomenon relevant to human genetics; (4) to choose an appropriate model organism in which to study specific biological phenomena; (5) to perform mathematical genetic analysis.

9. Recommended Reading List:

Tutorials in weeks 4-7 will be based on the following reviews:

Nat Rev Genet. 2015 16(5): 275–284 Genetic linkage analysis in the age of whole-genome sequencing. Jurg Ott, Jing Wang and Suzanne M. Leal

Adv Exp Med Biol 2020;1236:1-38. A History of Mouse Genetics: From Fancy Mice to Mutations in Every Gene. María J García-García

How to make a scientific presentation: "Giving a seminar: Suggestions for Graduate Students" by John Roth.

Reading material for the Mathematical genetics tutorials will be presented during the first tutorial.

10. Assessment Details: 70% Written review, 4,000 words (deadline, Friday of week 19, January

3rd);

15% tutorial paper presentation

15% of the module networks essay 1500 words (deadline Monday on

week 14, November 25th at 5pm)

11. Module Co-ordinator Juan Pablo Labrador labradoj@tcd.ie

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

3.5 GEU33045 Genomics & Systems Biology

Module Code
 Module Name
 GEU33045 (Open Module)
 Genomics & Systems Biology

3. Semester taught 14. Contact Hours 24

5. Module Personnel Mike Dolan, Adrian Bracken, Carsten Kröger, Kenneth Mok

6. Learning Aims The aim of this module is to equip students with a comprehensive understanding of the methods used in the fields of genomics, proteomics and metabolomics and how these methods are used for basic research, biotechnology, agriculture and medicine. To this end, several applications from work in diverse organisms (bacteria, fungi, plants, animals including humans) in addition to specific diseases and disorders (Schizophrenia and Cancer) will be presented. The module further introduces students to the field of systems biology and outlines how systems biology differs from the classic reductionist approach used in biology.

7. Module content:

| Week | Day & Time | Lecture Topic & Lecturer |
|------|--|---|
| 3 | Tue 10 th Sept 17:00-18:00 | Introduction to Genomics and Systems Biology (Dolan) |
| 3 | Wed 11 th Sept 17:00-18:00 | History of DNA Sequencing I (Dolan) |
| 4 | Mon 16 th Sept 17:00-18:00 | History of DNA Sequencing II: The Human Genome Project (Dolan) |
| 4 | Tue 17 th Sept 17:00-18:00 | Modern Day DNA sequencing I: 2nd Generation Sequencing Technologies (Dolan) |
| 5 | Mon 23 rd Sept 17:00-18:00 | Modern Day DNA sequencing II: 3rd Generation Sequencing Technologies (Dolan) |
| 5 | Tue 24 th Sept 17:00-18:00 | Structural and Comparative Genomics (Dolan) |
| 6 | Mon 30 th Sept 17:00-18:00 | Genomic Architecture of Schizophrenia (Dolan) |
| 6 | Tue 1 st Oct 17:00-18:00 | Transcriptomics: Revealing Gene Expression (Dolan) |
| 7 | Mon 7 th Oct 17:00-18:00 | Profiling RNA expression in the Schizophrenic Brain (Dolan) |
| 7 | Tue 8 th Oct 17:00-18:00 | Regulation of Gene Expression (Dolan) |
| 7 | Wed 9 th Oct 17:00-18:00 | Single-cell and spatial Transcriptomics (Dolan) |
| 8 | Mon 14 th Oct 17:00-18:00 | Single-cell dissection of the human brain in health and disease (Dolan) |
| 8 | Tue 15 th Oct 17:00-18:00 | Revision of material, discussion and answering student questions (Dolan) - tutorial |
| 9 | | Study/Review week |
| 10 | Tue 29 th Oct 17:00-18:00 | Bacterial genomes and comparative genomics (Kröger) |
| 11 | Mon 4 th Nov 17:00-18:00 | Functional genomics in bacteria (Kröger) |
| 11 | Tue 5 th Nov 16:00-17:00 | Introduction into the epigenome: histone and DNA modifications (Bracken) |
| 11 | Tue 5 th Nov 17:00-18:00 | Methods to analyse the epigenome; the ENCODE project (Bracken) |
| 12 | Mon 11 th Nov 17:00-18:00 | Cancer profiling and classification of tumour types (Bracken) |

| 12 | Tue 12 th Nov 17:00-18:00 | Using genomic information for the development of cancer therapies (Bracken) |
|----|---|--|
| 13 | Mon 18 th Nov 17:00-18:00 | Proteomics: Identify/characterise/quantify; Mass Spec and other technologies (Mok) |
| 13 | Tue 19 th Nov 17:00-18:00 | Quantitative proteomics; clinical proteomics (Mok) |
| 14 | Mon 25 th Nov 17:00-18:00 | Interaction/affinity proteomics; metabolomics introduction (Mok) |
| 14 | Tue 26 th Nov 16:00-17:00 | Metabolomics technologies (Mok) |
| 14 | Tue 26 th Nov 17:00-18:00 | Revision of material, discussion and answering student questions (all lecturing) |
| 15 | | Revision Week |
| 16 | | Assessment Week |

NOTE: Venue LTEE3

8. Learning Outcomes: Upon successful completion of this module, students will be able to describe experimental approaches used in the fields of genomics, proteomics and metabolomics. They will understand how to leverage these methods to analyze complex biological systems and questions. Students will be able to evaluate the applications of these techniques in biological sciences and discuss case studies involving specific diseases and disorders. Finally, they will be able to differentiate between systems biology and traditional reductionist approaches in biology.

9. Recommended Reading List: none

10. Assessment Details: One 1.5-hour exam paper at the end of semester 1

11. Module Coordinator Mike Dolan MJDOLAN@tcd.ie

3.6 BIU33150 Biochemistry for Biosciences

***Last year schedule and module descriptor. New timetable and details will be provided on Blackboard. Please use this information as a guide. All lectures are recorded and available to students who have Timetable clashes**

Module Code BIU33150 (Open Module)
 Module Name Biochemistry for Biosciences

3. Semester taught4. Contact Hours20 hours

5. Module Personnel Amir Khan (AK), K.H. Mok (KHM), Vincent kelly (VK), Martin Caffrey (MC), Andrei Budanov (AB), Derek Nolan (DN), Emma Creagh (EC), Aisling Dunne (AD), Daniela Zisterer (DZ).

6. Learning Aims This module follows on from the biochemistry/cell biology component of the "Molecules to Cells" BIU22201 module of year 2. The aim is to provide Junior Sophister students of other disciplines with the grounding in biochemistry necessary to (i) understand biology at a molecular level, (ii) form a mechanistic view of biological processes and (iii) appreciate the pathobiochemical basis of disease. The module covers four major themes in biochemistry: Proteins and Nucleic Acids, Membranes, Cytoskeleton and Signalling. The module will be assessed through a combination of in course assessment and an individual end of term exam.

7. Module content: Programme of lectures and practicals –

| Lecture | Lecture Topic & Lecturer | | |
|---------|--|--|--|
| | Dowt A. Drotteine and Muslais Asida | | |
| 1 | Part A - Proteins and Nucleic Acids | | |
| 1 | Amino acids and peptide bond (AK) | | |
| 2 | Structures , motifs and folds (AK) | | |
| 3 | Structure and mechanism: serine proteases (AK) | | |
| 4 | Spetrophotometry of biomolecules (KHM) | | |
| 5 | Protein folding and pathologies (KHM) | | |
| 6 | The proteome (KHM) | | |
| 7 | DNA, chromatin & the nucleus (VK) | | |
| 8 | RNA structure, folds & function (VK) | | |
| | Part B – Membranes | | |
| 9 | An introduction to cellular and model membranes (MC) | | |
| 10 | Membrane composition and therapeutic approaches (MC) | | |
| 11 | The Synthetic & Assembly Mechanisms for Membrane Proteins that Form | | |
| | Specific Topologies (DN) | | |
| 12 | Membrane transport of small molecules, specificity, mechanisms, energy | | |
| | Coupling (DN) | | |
| | Part C – Cytoskeleton | | |
| 13 | Structure of tubulin and microtubules and the Assembly / Disassembly of | | |
| | Microtubular Structures (EC) | | |
| 14 | Microtubular motors, types, mechanism of movement, regulation, | | |
| | physiological roles (EC) | | |
| 15 | Introduction to actin and the actin cytoskeleton (DN) | | |
| 16 | F-actin nucleation & pathologies associated with actin cytoskeleton (DN) | | |
| | Part D – Signalling | | |
| 17 | Introduction to cell signalling & GPCRs (EC) | | |
| 18 | G-Protein coupled Receptor (GPCR) regulation (EC) | | |
| 19 | Receptor tyrosine kinases (RTKs)-PDGF and EGF (DZ/AD) | | |
| 20 | RTK signalling – PKB and PDK1 (DZ/AD) | | |

Lectures schedule and time table

All lectures will be delivered online as follows with the accompanying lecture notes on blackboard: Module BIU33150. Lectures will be released 2 per week starting on week 1 of the teaching term (Monday 11th Sept). Subsequent lectures will be released 2 per week each following Monday. Lectures are often divided into part A , C etc on BB that combine to give 50 min overall for the lecture. Sampel MCQs and exam Qs are available on line in BB.

| Date of lects (on BB) | Lect No | Staff | Upload to BB by |
|-----------------------|---------|--------|-----------------|
| 1 Mon 11/09 | 1-2 | AK | 11/09 |
| 2 Mon 18/9 | 3-4 | AK/KHM | 18/09 |
| 3 Mon 25/9 | 5-6 | KHM | 25/09 |
| 4 Mon 2/10 | 7-8 | VK | 2/10 |
| 5 Mon 9/10 | 9-10 | MC | 9/10 |
| 6 Mon 16/10 | 11-12 | DN | 16/10 |
| 7 Reading week | | | |
| 8 Mon 30/10 | 13-14 | EC | 30/10 |
| 9 Mon 6/11 | 15-16 | DN | 6/11 |
| 10 Mon 13/11 | 17-18 | EC | 13/11 |
| 11 Mon 20/11 | 19-20 | AD/DZ | 20/11 |

(1-11 week of teaching term)

- 8. Learning Outcomes: On completion of this module, the student will be able to:
 - Recall and comprehend key knowledge and concepts of the hierarchy of polypeptide structure and the forces that stabilize the three-dimensional shape of proteins
 - Explain the link between a protein structure and its biological activity, and with appropriate examples, how human diseases arise from a deviation in structure
 - Appreciate the principles of spectrophotometry and its applications to biomolecules
 - Understand the concept of the proteome and its importance in disease
 - Integrate key concepts about nucleic acid structure and function
 - Demonstrate an understanding of the biochemical processes of nucleic acids in the cell
 - Recall and integrate key knowledge and concepts concerning the role of lipids in membrane structure and function
 - Describe how model membranes are formed and their applications
 - Describe how an understanding of membrane composition and structure can be used in the design of vaccines, antibiotics and beta-blockers.
 - Demonstrate a knowledge of the biosynthesis of membrane proteins, including the mechanisms of insertion and transport to their various locations.
 - Explain the types of membrane transport and how this process is coupled to energy and assayed.
 - Describe the structure of microtublules, their assembly and disassembly and their polarity.
 - Describe the structure of microtublule motors and the processes of directed vesicle transport and cytoplasmic streaming.
 - Describe the structure of monmeric actin and how it is assembled into filaments
 - Explain how actin nucleation is linked to pathological states.

- Describe the general principles of G-protein coupled receptor (GPCR) signalling and its regulation, the initial discovery of G-proteins linked to cyclase, the functional effects of cAMP and the activation of GPCR-linked signal-activated phospholipases.
- Discuss Receptor Tyrosine Kinase (RTK) signalling and details of MAP kinase cascades, using PDGF and EGF as examples. Explain RTK and PI3K pathways in the context of PKB (Akt) and PDK1 signalling.
- Describe the principles of steroid hormone receptor signalling mechanisms.
- **9. Recommended Reading List:** A reading list will be given out by lecturers during the module.
- 10. Assessment Details: 60% End of year examination, 40% in course assessed.

In course assessment: Two online MCQ assessemnts.

The MCQs will be structured as follows:

There will be two MCQs per lecture, giving 40 MCQs for the course.

The first MCQ will be after reading week and will cover the first 12 lectures (so 24 MCQs) representing 24% of the marks for the module.

The second MCQ will be in week 12, i.e. the last week of the term and will cover the material contained in the eight lectures after reading week. There will be 16 MCQs representing 16% of the marks for the course.

11. Module Coordinator: Dr D Nolan denolan@tcd.ie

4. Semester 2 Modules

4.1 GEU33100 Field Trip

Module Code GEU3100
 Module Name Field Trip

3. Semester taught 24. Contact Hours 18

- **5. Module Personnel** Adrian Bracken, Dan Bradley, Matthew Campbell, Lara Cassidy, Mike Dolan, Jane Farrar, Seamus Martin, Kevin Mitchell, Juan Pablo Labrador, Aoife Mc Lysaght, Russell McLaughlin, Mani Ramaswami, Frank Wellmer.
- **6. Learning Objectives:** The field trip takes place over two days on the week 27 (27th 28th Feb 2025, Hilary Term). It is a great opportunity for staff and students to meet scientifically and socially in an informal setting. Each student is expected to present a short (10 minute) presentation on paper related to their review chosen together with their supervisor.

7. Module content:

| Week | | Lecture Topic & Lecturer |
|------|----------------------------|--------------------------------------|
| 26 | Wed 19 th Feb | Field trip tutorial (Labrador) LTEE3 |
| | 12:00-13:00 | |
| 27 | Thurs 27 th Feb | Student presentations |
| | 10:00-18:30 | |
| 27 | Fri 28 th Feb | Student presentations |
| | 10:00-15:00 | |

- **8. Learning Outcomes:** At the end of this module, students should be able to deliver a professional presentation on a scientific subject.
- **9. Recommended Reading List:** Individually suggested by supervisors and student personal research.
- **10. Assessment Details:** 10 min presentation on week 27 (27th 28th Feb 2025). Attendance and presentation are **compulsory**.
- 11. Module Coordinators: Juan Pablo Labrador labradoj@tcd.ie

4.2 GEU33303 Molecular genetics II - Genome structure and dynamics

1. Module Code GEU33303

2. Module Name Molecular Genetics II

3. Semester taught 2

4. Contact Hours 23 (5 ECTS)

5. Module Personnel Prof. Frank Wellmer, TBD

6. Learning Aims The aim of this module is to introduce students to fundamental concepts in Molecular Genetics. By focusing on plant and microbial genetics, we want to highlight the overlap between these seemingly disparate biological systems. The microbial genetics component will focus on critical regulatory aspects of the gene expression machinery (transcription and translation), and genome replication (DNA replication, homologous recombination, mutagenesis and DNA repair). In relation to plant genetics, students will be introduced to major topics such as the structure and evolution of the nuclear genome, the importance of model plants, including Arabidopsis thaliana, light-regulated gene expression, hormone receptors and signal transduction systems. The evolutionary origins of plant cell organelles (chloroplasts and mitochondria) via endosymbiosis involving ancestral microbes will also be explored as will be the many facets of plant-microbe interactions including plant immunity and symbiosis.

7. Module content:

| Week | Date & Time | Lecture Topic & Lecturer |
|------|----------------------------|--|
| 22 | Tue 21 st Jan | DNA replication I: Linear and circular genomes; replication strategies; origins of |
| | 11:00-12:00 | replication (TBD) |
| 22 | Tue 21 st Jan | DNA replication II: regulation of initiation at oriC; role of DnaA and |
| | 14:00-15:00 | hemimethylation (TBD) |
| 22 | Thurs 23 rd Jan | Homologous recombination I: Holliday, Meselson-Radding and Szostak models |
| | 15:00-16:00 | (TBD) |
| 22 | Fri 24 th Jan | Homologous recombination II: RecA, SSB, RecBCD, RuvA, B, C; Horizontal gene |
| | 16:00-17:00 | transfer (TBD) |
| 23 | Tue 27 th Jan | DNA Mutagenesis: chemical, deamination, base-analogues, chromosomal |
| | 14:00-15:00 | integration through homologous DNA, transposons (TBD) |
| 23 | Thurs 30 th Jan | DNA Repair systems: Mismatch base repair; Base excision repair; nucleotide |
| | 15:00-16:00 | excision repair; SOS response; Non-homologous end-joining; Error Prone DNA |
| | | Polymerases (TBD) |
| 24 | Tue 4 th Feb | Tutorial: revision of material, discussion and answering student questions (TBD) |
| | 11:00-12:00 | |
| 24 | Tue 4 th Feb | Introduction: why plant research is important (Wellmer) |
| | 14:00-15:00 | |
| 24 | Thurs 6 th Feb | Models for plant research and nuclear genome structure (Wellmer) |
| | 15:00-16:00 | |
| 25 | Tue 11 th Feb | Evolution of the nuclear genome and sex determination in plants (Wellmer) |
| | 11:00-12:00 | |
| 25 | Thurs 13 th Feb | Discovery and mutagenic potential of transposable elements (Wellmer) |
| | 15:00-16:00 | |
| 26 | Fri 23 rd Feb | Regulation of transposon activity (Wellmer) |
| | 14:00-15:00 | |
| 28 | | Review Week |
| 29 | Wed 12 th Mar | The plastid genome and plastid evolution (Wellmer) |
| | 16:00-17:00 | |
| 29 | Thurs 13 th Mar | The genetic toolbox for analyzing plant biology: the making of transgenic plants |
| | 14:00-15:00 | (Wellmer) |
| 29 | Fri 14 th Mar | Tutorial: revision of material, discussion and answering student questions |
| | 14:00-15:00 | (Wellmer) |
| 30 | Wed 19 th Mar | The genetic toolbox for analyzing plant biology: genetic screens (Wellmer) |
| | 16:00-17:00 | |

| 30 | Thurs 20 th Mar | The genetic toolbox for analyzing plant biology: mutant analyses (Wellmer) |
|----|----------------------------|--|
| | 14:00-15:00 | |
| 30 | Thurs 20 th Mar | Understanding signal transduction in plants (Wellmer) |
| | 15:00-16:00 | |
| 30 | Fri 21 st Mar | Understanding the response pathway for light (Wellmer) |
| | 14:00-15:00 | |
| 31 | Wed 26 th Mar | Understanding the response pathway for the hormone ethylene (Wellmer) |
| | 16:00-17:00 | |
| 31 | Thurs 27 th Mar | Plant-microbe interactions (Wellmer) |
| | 14:00-15:00 | |
| 31 | Thurs 27 th Mar | Plant-pathogen interactions: an evolutionary arms race (Wellmer) |
| | 15:00-16:00 | |
| 33 | Thurs 10 th Apr | Tutorial: revision of material, discussion and answering student questions |
| | 14:00-15:00 | (Wellmer) |

- 8. Learning Outcomes: Upon successful completion of this module, students will be able to describe the critical regulatory features of the gene expression systems in prokaryotic microbes and eukaryotic cell organelles (chloroplasts and mitochondria). They will understand how DNA is replicated, how it can be recombined via homologous recombination, altered by mutagenic processes, and repaired. Students will also have acquired knowledge of key topics in plant genetics, such as light-regulated gene expression, hormone receptors and signal transduction, and the genetic basis of plant immunity.
- 9. Recommended Reading List: Anthony J.F. Griffiths et al. Introduction To Genetic Analysis. 12th edition. New York, NY: W.H. Freeman & Company, 2020.

Further reading on specialist topics will be provided during the presentation of the module.

10. Assessment Details: One 1.5-hour exam paper at the end of semester 2

11. Module Coordinators: Frank Wellmer (<u>wellmerf@tcd.ie</u>)

Alastair Fleming (fleminal@tcd.ie)

4.3 GEU33301 Bioinformatics

Module Code
 Module Name
 Semester taught
 Contact Hours
 GEU33301
 Bioinformatics

 Semester 2
 Ontact Hours
 Hours

5. Module Personnel Dr Karsten Hokamp (KH), Dr Carsten Kröger (CK), Dr Máire Ní Leathlobhair (MNL),

Dr Fiona Roche (FR)

6. Learning Aims This module is taught in combination with the Microbiology department and contains web-based bioinformatics, Python programming and a data handling component. Lectures will be held in computer labs to enable a hands-on approach. The bioinformatics component provides a practical introduction to the use of commonly used bioinformatic databases and tools with a focus on web-based applications. Students will become familiar with accessing biological sequence databases and exploring various sequence analysis tools to understand evolutionary relationships and how this can help to draw protein functional and structural inferences. The Python programming component introduces students to computer programming in Python using bioinformatics-related examples and exercises. It will be carried out within an internal JupyterLab environment. The data handling part contains a biolab component in which samples will be prepared for whole-genome sequencing (WGS). The combined lectures and practicals cover basic techniques for processing next-generation sequencing data and working with the statistical software R.

This module runs for 15×2 hours (combined lecture and practical sessions) plus a 5-hour lab practical and 2×2 -hour in-class assessments in Semester 2.

Students are required to bring their own laptop for the practical sessions.

As learning aims of this module students will

- learn how to search a range of biological databases
- understand how proteins are annotated and classified
- get to know which tools can be used to explore unknown sequences of interest
- master the concept of sequence alignment and homology searching
- understand the process of programming
- acquire basic Python programming skills
- gain a thorough understanding of next-generation sequencing data
- be familiar with the steps and tools required to process, map and visualise NGS data
- become familiar with the specifics of NGS data analysis
- understand how to enhance NGS results with annotation data
- isolate genomic DNA from E. coli bacteria
- prepare a DNA library for Nanopore sequencing
- carry out WGS analysis of a bacterial genome

7. Module content: Programme of lectures and practicals

| Week | Day & Time | Lecture Topic & Lecturer |
|------|--------------------------|--|
| 22 | Mon 20 th Jan | Bioinformatics 1 – Biological Databases (FR, 2 hours) |
| | 14:00-16:00 | |
| 22 | Fri 24 th Jan | Bioinformatics 2 – Protein Resources & Function Prediction (FR, 2 hours) |
| | 11:00-13:00 | |
| 23 | Mon 27 th Jan | Bioinformatics 3 – Sequence Alignments (FR, 2 hours) |
| | 14:00-16:00 | |

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|----|--------------------------|--|-------|
| 23 | Fri 31 st Jan | Bioinformatics 4 – Multiple Alignments & Genome Browers (FR, 2 hours) | |
| | 14:00-16:00 | | |
| 24 | Fri 7 th Feb | Programming 1 – Variables and Loops (KH, 2 hours) | |
| | 14:00-16:00 | | |
| 25 | Mon 10 th Feb | Programming 2 – Input/Output, Branching (KH, 2 hours) | |
| | 14:00-16:00 | | |
| 25 | Fri 14 th Feb | Programming 3 – Lists, Tuples, Sets (KH, 2 hours) | |
| | 14:00-16:00 | | |
| 26 | Mon 17 th Feb | Bioinformatics Assessment (FR, 2 hours) | |
| | 11:00-13:00 | | |
| 26 | Mon 17 th Feb | Programming 4 – Dictionaries (KH, 2 hours) | |
| | 14:00-16:00 | | |
| 26 | Fri 21 st Feb | Programming 5 – Functions, System Commands (KH, 2 hours) | |
| | 11:00-13:00 | | |
| 28 | Study/Review we | eek | |
| 29 | Tue 11 th Mar | Biolab Practical (CK, 5 hours) ** | |
| | 11:00-12:00 & | | |
| | 14:00-18:00 | | |
| 29 | Thu 13 th Mar | Programming Assessment (KH, 2 hours) | |
| | 10:00-12:00 | | |
| 29 | Fri 14 th Mar | NGS Data Analysis 1 – Using R for bioinformatics: Part I (MNL, 2 hours) | |
| | 11:00-13:00 | | |
| 30 | Thu 20 th Mar | NGS Data Analysis 2 – Using R for bioinformatics: Part II (MNL, 2 hours) | |
| | 11:00-13:00 | | |
| 30 | Fri 21 st Mar | NGS Data Analysis 3 – Graphs & Data Visualization in R (MNL, 2 hours) | |
| | 11:00-13:00 | | |
| 31 | Thu 27 th Mar | NGS Data Analysis 4 – Markdown Notebooks in R (MNL, 2 hours) | |
| | 11:00-13:00 | | |
| 32 | Thu 3 rd Apr | NGS Data Analysis 5 – Introduction to Machine Learning in R (MNL, 2 hours) | |
| | 11:00-13:00 | | |
| 33 | Thu 10 th Apr | R Assessment (MNL, 2 hours) | |
| | 11:00-13:00 | | |

NOTE: Venue LTEE3 and MAC Lab.

Description of each Lecture:

Bioinformatics 1 - Biological Databases (FR, 2 hours) This lecture covers how bioinformatics data are stored and organised with a focus on resources provided at NCBI and the EBI. Students will learn about the different types of data and tools found within these resources, delving deeper into the Gene and Ensembl databases.

Bioinformatics 2 - Protein Resources and Function Prediction (FR, 2 hours) This lecture describes how protein sequence data are stored, annotated and classified. Students will also learn about computational methods used for predicting protein function and be introduced to protein resources, such as UniProt and InterPro.

Bioinformatics 3 - Sequence Alignment (FR, 2 hour) This lecture introduces the concept of pairwise sequence alignment, the process of comparing two sequences to determine if they are evolutionarily related to one another. The lecture will also explore the BLAST algorithm, which compares single sequences against a database of sequences to search for significantly similar sequences.

Bioinformatics 4 - Multiple Sequence Alignment & Genome Browsers (FR, 2 hours) This lecture extends alignment approaches to the alignment of multiple sequences and discusses its many

^{**}Biolab 3 Carsten Kroger session on Tue 11th March.

applications including its role in inferring function and structure and identifying genetic variants from sequence comparison. Students will also be introduced to genome browsers and learn how to interactively explore and visualise biological data in the context of the genome with a specific focus on comparative genomics.

Programming 1 – Variables and Loops (KH, 2 hours) This lecture covers string variables, string formatting, as well as built-in functions and methods for strings. This is followed by the use of 'while' and 'for' loops for repeated application of programming steps.

Programming 2 – Input/Output, Branching (KH, 2 hours) This lecture deals with ways of reading experimental data from files into a Python script and how to store results generated by a script in a file. It also introduces ways of making decisions in a Python script through branching. With this additional skill set students will be able to write more elaborate scripts and tackle processing of sequence data in Fasta format.

Programming 3 – Lists, Tuples, Sets (KH, 2 hours) Lists are a very common feature in data science. They are represented in Python through various types of iterable variables, which are covered in this lecture together with their built-in functions and methods.

Programming 4 – Dictionaries (KH, 2 hours) This lecture is dedicated to dictionaries, a collection of key-value pairs, which enables students to implement a DNA translation script.

Programming 5 – Functions, System Commands (KH, 2 hours) With the introduction of functions, the repertoire of programming skills is expanded so that scripts can be written more efficiently. System commands will enable the execution of external programs from within script.

Practical – Isolation of gDNA and Nanopore Sequencing (CK, 5 hours) In this practical, the students will learn how to isolate genomic DNA from Escherichia coli bacteria and how to quality-control the DNA integrity to be suitable for whole genome sequencing. One DNA library will be prepared for Oxford Nanopore sequencing.

NGS Data Analysis 1 – Using R for Bioinformatics (MNL, 2 hours) The last part of the module deals with the analysis of Next-Generation Sequencing (NGS) data. This lecture will introduce students to how the statistical software R can be used to manipulate and analyse NGS and other data. Students will learn about data structures, data manipulation and subsetting, as well as functions and packages in R.

NGS Data Analysis 2 – Using R for Bioinformatics: Part II (MNL, 2 hours) Building on Part I, this lecture will continue to explore how we can use R for the analysis of NGS data. Students will explore more complex data manipulation methods and learn about specialized bioinformatics packages in R. The session will also cover statistical analysis, enabling students to apply these skills to their own research projects.

NGS Data Analysis 3 - Markdown Notebooks in R (MNL, 2 hours) This lecture covers the use of R Markdown Notebooks as an integrated way to perform and report analysis of data. Students will learn about the structure of R Notebooks, how to use Markdown to format text and how to create and customize code blocks within Notebooks.

NGS Data Analysis 4 - Genome Assembly & Data Sharing (MNL, 2 hours) Students will learn about different ways to assemble a genome de novo from short and long read sequencing data. The lecture will also involve discussion of recent ambitious large scale sequencing projects making genome assemblies publicly available and the principles of data sharing.

NGS Data Analysis 5 – Introduction to Machine Learning in R (MNL, 2 hours) This lecture will introduce students to the basics of machine learning using R. Students will learn about key machine learning concepts and techniques, including supervised and unsupervised learning.

8. Learning Outcomes: On successful completion of the module students should be able to:

MLO1. Query a range of bioinformatic databases

MLO2. Apply tools to investigate unknown sequences

MLO3. Carry out sequence alignment and homology searching

MLO4. Visualise biological data through a genome browser

MLO5. Approach programming tasks in a structured way

MLO6. Write Python scripts following good coding practice

MLO7. Integrate external programs and use functions within Python scripts

MLO8. Solve entry-level bioinformatics problems using Python scripts

MLO9. Assess the quality of NGS data

ML10. Apply bioinformatics tools for processing NGS data

ML11. Visualise NGS data through genome browser resources

ML12. Carry out core NGS downstream analyses resulting in genome assemblies

ML13. Integrate external annotation data with analysis results through Python scripts

ML14. Competency to isolate bacterial DNA and to prepare a Nanopore sequencing reaction

9. Recommended Reading List:

The Biostar Handbook, 2ndEdition (https://www.biostarhandbook.com/)

Bioinformatics, 4th Edition, John Wiley and Sons Ltd, by Andreas D. Baxevanis, Gary Bader, David Wishart

Bioinformatics and Functional Genomics, 3rd Edition, 2015, Wiley Blackwell, Jonathan Pevsner A Critical Guide to BLAST, TK Attwood

https://www.mygoblet.org/about-us/publication/critical-guide-blast

How to Think Like a Computer Scientist, Learning with Python 3 (RLE), 2012 edition, by Peter Wentworth, Jeffrey Elkner, Allen B. Downey, and Chris Meyers

(https://openbookproject.net/thinkcs/python/english3e/)

10. Assessment Details: (1) A bioinformatics exam 33%

(2) a Python programming exam 33%

(3) R homeworks and an R programming exam 34%.

In case of an overall fail mark for this module, the failed components

need to be repeated during the reassessment period.

11. Module Coordinator: Dr Karsten Hokamp <u>kahokamp@tcd.ie</u>

4.4 GEU33035 Genetic Analysis of Nervous Systems

1. Module Code GEU33035

2. Module Name Genetic Analysis of Nervous Systems

3. Semester taught Semester 2

4. Contact Hours 30

5. Module Personnel Juan Pablo Labrador, Mani Ramaswami

6. Learning Aims The module is focused on understanding how experimental genetics are used to manipulate genes in organisms to address problems in biology. Areas covered are 1) Experimental Genetics: structure and conservation of genes, nature of mutations and their effects on protein structure and function, model organisms in genetic research and experimental manipulation of animal genomes. 2) Developmental Neurogenetics: the purpose and design of genetic screens, genetic analysis of neurogenesis and genetic analysis of axon guidance 3) Behavioral Genetics: cell organization and methods of cell biology, cell biology of neurons and synapses, creation and use of molecular reporters of specific gene or cell activity, methods to study nervous systems, sensory circuits, sensation; transduction; perception; coding; behavior, learning and memory, sleep and circadian rhythms.

7. Module content: Program of lectures

| Week | Day & Time | Lecture Topic & Lecturer |
|------|---------------------------------|---|
| 22 | Tue 21st Jan 12:00- | Experimental Genetics - Introduction - Structure and conservation of genes, nature of mutations |
| | 13:00 | and their effects on protein structure and function (Labrador) |
| 22 | Wed 22nd Jan | Experimental Genetics - Dominance and germ line transmission (Labrador). |
| | 15:00-16:00 | |
| 22 | Thu 23rd Jan | Experimental Genetics - Transgenesis and the Gal 4 system (Labrador). LTEE1 |
| | 9:00-10:00 | |
| 22 | Thu 23rd Jan 11:00- | Experimental Genetics - Experimental manipulation of animal genomes. (Labrador) |
| | 12:00 | |
| 22 | Thu 23rd Jan 12:00- | Developmental Genetics - The purpose and design of genetic screens (Labrador) |
| | 13:00 | |
| 23 | Tue 28th Jan 12:00- | Developmental Genetics – Mapping Genes (Labrador) |
| | 13:00 | |
| 23 | Wed 29th Jan | Developmental Genetics - From phenotypes to genes, midline screen (Labrador) |
| | 15:00-16:00 | |
| 23 | Thu 30th Jan | Developmental Genetics - From genes to pathways, modifiers screens (Labrador) |
| | 11:00-12:00 | |
| 23 | Thu 30th Jan | Neurogenetics/ Behavioral Genetics Overview (Ramaswami) |
| | 12:00-13:00 | |
| 24 | Tue 4th Feb | Building blocks – 1: Neurons, Ion channels, Synapses (Ramaswami) |
| | 12:00-13:00 | |
| 24 | Wed 5th Feb | Building blocks – 2: Neurons, Ion channels, Synapses (Ramaswami) |
| | 15:00-16:00 | |
| 24 | Thu 6th Feb | Cells to perception: visual system and some experimental methods (Ramaswami) |
| | 11:00-12:00 | |
| 24 | Thu 6th Feb | Genetics and biology of taste and smell (Ramaswami) |
| | 12:00-13:00 | |
| 25 | Tue 11 th Feb 12:00- | Cellular Basis for Memory (Ramaswami) |
| | 13:00 | |
| 25 | Wed 12 th Feb | Genetic analysis of memory in Drosophila (Ramaswami) |
| | 15:00-16:00 | |
| 25 | Thu 13 th Feb | Circadian Rhythms (Ramaswami) |
| | 11:00-12:00 | |
| 25 | Thu 13 th Feb | TBC |
| | 12:00-13:00 | |

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|----|---------------------------------|------------------------|
| 26 | Tue 18 th Feb | TBC |
| | 11:00-13:00 | |
| 26 | Tue 18 th Feb | TBC |
| | 12:00-13:00 | |
| 26 | Wed 19 th Feb | TBC |
| | 15:00-16:00 | |
| 26 | Thu 20 th Feb 11:00- | TBC |
| | 12:00 | |
| 26 | Thu 21 st Feb | TBC |
| | 12:00-13:00 | |
| 27 | Tue 25 th Feb | TBC |
| | 11:00-12:00 | |
| 27 | Tue 25 th Feb | TBC |
| | 12:00-13:00 | |
| 28 | | Study/Review week |
| 29 | Mon 10th Mar | TBC Final written test |
| | 15:00-16:00 | |
| 29 | Mon 10th Mar | TBC |
| | 16:00-17:00 | |
| 30 | Wed 19 th Mar | TBC |
| | 14:00-16:00 | |
| 31 | Mon 24th Mar | TBC |
| | 10:00-11:00 | |
| 31 | Mon 24th Mar | TBC |
| | 11:00-12:00 | |
| 31 | Mon 24th Mar | TBC |
| | 14:00-15:00 | |
| 31 | Mon 24th Mar | TBC |
| | 15:00-16:00 | |
| | · · | |

NOTE: Venue LTEE3 // week 22 Thursday 9:00-10:00 LTEE1

Lecture times are subject to change within the hours reserved for the module on the above timetable.

8. Learning Outcomes: Upon successful completion of this module, students will be able to understand and describe how model organisms are used in genetic research and common technologies and methods employed to genetically modify organisms. Students should also understand the basis of genetic screens and mapping. They will be able to explain epistasis through the analysis of different genetic interactions in neurogenesis and axon guidance. Students will become familiar with the cell biology of neurons and synapse as well as methods to probe synaptic activity. Students will also learn about circuitry underlying perception.

9. Recommended Reading List: Anthony J.F. Griffiths; Susan R. Wessler; Sean B. Carroll; John Doebley. Introduction To Genetic Analysis. New York, NY: W.H. Freeman & Company, 2015

10. Assessment Details: Continuous assessment: class participation – Poster presentation

(Monday Feb 24^{th)} TBC (50%) and test on week 29 TBC (50%)

11. Module Coordinator Juan Pablo Labrador labradoj@tcd.ie

4.5 GEU33008 Analytical Genetics Laboratory

1. Module Code GEU33008

2. Module Name Analytical Genetics Laboratory

3. Semester taught Semester 2

4. Contact Hours 5 hours per week5. Module Personnel Juan Pablo Labrador

6. Learning Aims

This module is a practical module that introduces the fundamentals of genetic analysis and the use of *Drosophila melanogaster* as a genetic model organism. The module will cover different aspects of model organisms handling and segregation. Virtual crosses are employed to understand Mendelian genetics and non-Mendelian inheritance including segregation, recombination, gene mapping, lethal genes and sex-linked inheritance.

7. Module content: Programme of lectures

| Week | Day & Time | Lecture Topic & Lecturer | | |
|-----------------|---|---|--|--|
| 22 | Mon 20 th Jan 16:00-18:00 Tue 21 st Jan 15:00-17:00 Wed 22 nd Jan 16:00- 18:00 | Drosophila husbandry, identification of phenotypes, setting up crosses Mendelian inheritance tutorial. P-elements tutorial Monohybrid, dihybrid crosses (Labrador) | | |
| 23 | Tue 28 th Jan 15:00- 17:00 Wed 29 th Jan 16:00- 18:00 Drosophila husbandry, identification of phenotypes, setting up crosses Non-Mendelian inheritance tutorial, Lethal mutations. (Labrador) Short Quiz 1: Monohybrid/dihybrid crosses | | | |
| 24 | Segregation in Drosophila, identification of males carrying P-elements Lethal mutations , Sex linked inheritance. (Labrador) Short Quiz 2: Lethal mutations | | | |
| 25 | Tue 11 th Feb 15:00-17:00 Wed 12 th Feb 16:00- 18:00 | Segregation in Drosophila - fly husbandry Sex linked inheritance , Linkage, recombination and mapping. Linkage and mapping tutorial (Labrador) Short Quiz 3: Sex linked inheritance | | |
| 26 | Tue 17 th Feb 15:00-17:00 Wed 18 th Feb 16:00- 18:00 Segregation in Drosophila analysis of crosses Linkage and mapping (Labrador) Short Quiz 4: Linkage, Recombination and mapping | | | |
| 27 | Mon 24 th Feb 14:00-17:00 Tue 25 th Feb 15:00-17:00 Wed 26 th Feb 16:00- 18:00 | Fly husbandry -Set up crosses if required Review Mendelian and non-Mendelian inheritance (Labrador) Final Quiz: Mendelian and non-Mendelian inheritance, lethal mutations, linkage and mapping (date TBC) | | |
| 28 | | Reading Week | | |
| 29 | | No lectures | | |
| <mark>30</mark> | Mon 17 th Mar 14:00-17:00 | No lecture St Patrick day | | |

| Tue 18 th Mar Segregation in Drosophila – Review of results P-element mapping in Drosophila Tutorial (Labrador) Fly husbandry | | P-element mapping in Drosophila Tutorial (Labrador) |
|--|--|---|
| 31 | Analysis and review of P-element results Fly husbandry (Labrador) | |
| 32 | Mon 31 st Mar 14:00-17:00 Wed 1 st April 16:00- 18:00 | Extra slot |

NOTE: Experiment weeks may be subject to change as we are working with live animals. An updated experiment, MCQs and lab report timetable will be provided at the start of the module.

Venue MAC LAB and BIOLAB3

8. Learning Outcomes: Upon successful completion of this module, students will be able to understand how model organisms are used in genetic research in a laboratory setting. Students should be able to set-up crosses and plan experiments using *Drosophila melanogaster*. Students will be able to design crosses and analyse ratios in the progeny. Mendelian inheritance ratios through the analysis of monohybrid, dihybrid and trihybrid crosses involving two or 3 chromosomes should be understood. Students will appreciate non-Mendelian inheritance involving sex linked loci or linked loci in autosomes or sex chromosomes.

9. Recommended Reading List:

Anthony J.F. Griffiths; Susan R. Wessler; Sean B. Carroll; John Doebley. Introduction To Genetic Analysis. New York, NY: W.H. Freeman & Company, 2015

Greenspan, R.J. (2004). Fly pushing: the theory and practice of Drosophila genetics, 2nd ed. (Cold Spring Harbor, N.Y.: Cold Spring Harbor Laboratory Press).

10. Assessment Details: MCQs/Final MCQ 50% Week 27. Lab report 50% submission deadline

Friday week 30/31 (TBC) 5pm

11. Module Coordinator Juan Pablo Labrador labradoj@tcd.ie

4.6 GEU33055 Developmental Genetics

Module Code
 Module Name
 GEU33055 (Open Module)
 Developmental Genetics

3. Semester taught Semester 2

4. Contact Hours 24

5. Module Personnel Prof. Seamus Martin, Prof. Adrian Bracken, Prof. Frank Wellmer

6. Learning Aims This module aims at introducing students to fundamental concepts in developmental genetics and to experimental approaches that are used to study development. To this end, the module takes a comparative approach: the development of different organisms (insects, vertebrates, plants) will be examined to demonstrate differences and commonalities in the genetic mechanisms controlling morphogenesis. Students will be introduced to important developmental control mechanisms, including morphogens, spatial signaling via asymmetric distribution of signaling molecules, signal transduction cascades that culminate in differential gene expression and the activation of homeotic selector genes which dictate the specification of biological structures such as limbs and wings. We will also explore the factors governing cell division, a key biological process that is used to generate huge numbers of new cells during development as well as for tissue turnover and repair, and we will examine how cell division is precisely regulated via the 'cell division cycle'. The module will also introduce students to stem cell biology and will examine how stem cells are programmed to undergo growth and differentiation.

7. Module content:

| Week | Day & Time | Lecture Topic & Lecturer | |
|------|--|---|--|
| 22 | Wed 22 nd Jan | Intro to development and developmental genetics-Overview (Martin) | |
| | 11:00-12:00 | | |
| 22 | Wed 22 nd Jan | Pluripotent Stem Cells (Bracken) | |
| | 14:00-15:00 | | |
| 22 | Thu 23 rd Jan | Multipotent Stem Cells 1 (Bracken) | |
| | 14:00-15:00 | | |
| 23 | Wed 29 th Jan | The cell cycle 1 (Martin) | |
| | 11:00-12:00 | | |
| 23 | Wed 29 th Jan | Cellular Reprogramming (Bracken) | |
| | 14:00-15:00 | | |
| 23 | Thu 30 th Jan | Multipotent Stem Cells 2 (Bracken) | |
| | 14:00-15:00 | | |
| 24 | Wed 5 th Feb | The cell cycle 2 (Martin) | |
| | 11:00-12:00 | | |
| 24 | 24 Wed 5 th Feb The cell cycle 3 (Martin) | | |
| | 14:00-15:00 | | |
| 24 | Thu 6 th Feb | The cell cycle 4 (Martin) | |
| | 14:00-15:00 | | |
| 25 | Wed 12 th Feb | Introduction to <i>Drosophila</i> development and embryogenesis (Martin) | |
| | 11:00-12:00 | | |
| 25 | Wed 12 th Feb | Drosophila development: anterior-posterior axis specification (Martin) | |
| | 14:00-15:00 | | |
| 25 | Thu 13 th Feb | Drosophila development: role of maternal effect genes (Martin) | |
| | 14:00-15:00 | | |
| 26 | Wed 19 th Feb | Drosophila development: activation and function of gap genes (Martin) | |
| | 11:00-12:00 | | |
| 26 | Wed 19 th Feb | Drosophila development: activation and function of pair-rule genes (Martin) | |
| | 14:00-15:00 | | |

| | | <u> </u> | | | |
|----|--------------------------|--|--|--|--|
| 26 | Thu 20 th Feb | Drosophila development: activation and function of segment polarity genes (Martin) | | | |
| | 14:00-15:00 | | | | |
| 27 | Wed 26 th Feb | Drosophila development: dorso-ventral axis specification (Martin) | | | |
| | 11:00-12:00 | | | | |
| 27 | Wed 26 th Feb | Drosophila development: homeotic selector genes and segment identity (Martin) | | | |
| | 12:00-13:00 | | | | |
| 27 | Wed 26 th Feb | Tutorial: revision of material, discussion and student questions (Martin, Bracken) | | | |
| | 14:00-15:00 | | | | |
| 27 | Wed 26 th Feb | Extra slot – no lecture scheduled | | | |
| | 15:00-16:00 | | | | |
| 28 | | Study/Review week | | | |
| 29 | Wed 12 th Mar | Organogenesis: genetic control of vertebrate limb development (Wellmer) | | | |
| | 11:00-12:00 | | | | |
| 29 | Wed 12 th Mar | Organogenesis: genetic control of vertebrate limb development II (Wellmer) | | | |
| | 14:00-15:00 | | | | |
| 30 | Wed 19 th Mar | Extra slot – no lecture scheduled | | | |
| | 11:00-12:00 | | | | |
| 31 | Wed 26 th Mar | Organogenesis: Genetic control of <i>Drosophila</i> eye development (Wellmer) | | | |
| | 11:00-12:00 | | | | |
| 31 | Wed 26 th Mar | Principles of plant development (Wellmer) | | | |
| | 14:00-15:00 | | | | |
| 32 | Wed 26 th Mar | Stem cells, cell fate determination and organogenesis in plants (Wellmer) | | | |
| | 11:00-12:00 | | | | |
| 32 | Wed 26 th Mar | Tutorial: revision of material, discussion and student questions (Wellmer) | | | |
| | 14:00-15:00 | | | | |

NOTE: Venue LTEE3

8. Learning Outcomes: Upon successful completion of this module, students will be able to describe fundamental mechanisms underlying the control of development in animals and plants. They will understand how cell division is tightly regulated in higher organisms via a dedicated set of enzymes (called the cyclins and cyclin-dependent kinases) and how deregulated cell division can lead to cancer. They will have acquired knowledge to outline key events during development including embryogenesis and the formation of organs and will be able to describe how these events are regulated at a molecular level. Students will be familiar with the biology of stem cells and how these cells are programmed to undergo growth and differentiation. They will be able to describe experimental approaches that are used by researchers to dissect developmental processes and mechanisms.

9. Recommended Reading List:

Anthony J.F. Griffiths; Susan R. Wessler; Sean B. Carroll; John Doebley. Introduction To Genetic Analysis. New York, NY: W.H. Freeman & Company, 2015.

10. Assessment Details: One 1.5-hour exam paper at the end of semester 211. Module Coordinator Prof. Seamus Martin Email: MARTINSJ@tcd.ie

4.7 GEU33215 Medical Genetics

1. Module Code GEU33215 (Open Module)

2. Module Name Medical Genetics

3. Semester taught Semester 2

4. Contact Hours 18

5. Module Personnel Jane Farrar, Russell McLaughlin,

6. Learning Objectives The module provides an introduction to core concepts in medical genetics highlighting the importance and power of genetic information in the era of genomic medicine and the impact of such information for all of us. Learning objectives include: (1) discussion of the genetic basis of single gene disorders (Mendelian) and complex disorders; (2) overview of the history of medical genetics; (3) insights into key developments in medical genetics up to 2020 including state-of-art technologies and novel innovative therapies; (4) discussion of the key technologies and methodologies currently used to elucidate the genetic basis of human traits; (5) discussion of the individualisation of medicine and the important roles of genetic information in disease diagnosis, prognosis and the design and choice of therapy. In summary, the module provides an introduction to:

- The genetic basis of mendelian & complex disorders
- Genetic technologies & methodologies used to elucidate the genetic basis of human traits
- The exploitation of genomic data in diagnosis, prognosis & treatment of disease
- The individualization of medicine using genetic information

7. Module content: Programme of lectures

| Week | Day & Time | Lecture Topic & Lecturer | | |
|-----------------|--|---|--|--|
| 29 | Mon 10 th Mar | Importance of medical genetics and history of developments in the field. (Jane Farrar) | | |
| | 17:00-18:00 | | | |
| 29 | Wed 13 th 17:00- | Introduction to complex traits. (Russell McLaughlin) | | |
| | 18:00 | | | |
| 29 | Fri 15 th 16:00- | Overview of spectrum of Mendelian and multifactorial disorders. (Jane Farrar) | | |
| | 17:00 | | | |
| 29 | Fri 15 th 17:00- | Quantitative traits and complex disorders. (Russell McLaughlin) | | |
| | 18:00 | | | |
| <mark>30</mark> | <mark>Mon 17th Mar</mark> | Bank holiday -no lectures | | |
| | 17:00-18:00 | | | |
| 30 | Tue 18 th Mar | Identification of disease genes and disease mutations. (Jane Farrar) | | |
| | 17:00-18:00 | | | |
| 30 | Wed 19 th Mar | Variance components and heritability (Russell McLaughlin) | | |
| | 17:00-18:00 | | | |
| 30 | 30 Fri 21 st Mar Variant interpretation and ACMG guidelines. (Jane Farrar) | | | |
| | 16:00-17:00 | | | |
| 30 | Fri 21 st Mar | New trends in medical genetics. (Jane Farrar) | | |
| | 17:00-18:00 | | | |
| 31 | Mon 24 th Mar | Introduction to pharmacogenomics. (Jane Farrar) | | |
| | 17:00-18:00 | | | |
| 31 | Tue 25 th Mar | Genetic variation in cytochrome P450s and influence on drug response. (Jane Farrar) | | |
| | 17:00-18:00 | | | |
| 31 | Wed 26 th Mar | Dominance, additivity and interaction. (Russell McLaughlin) | | |
| | 17:00-18:00 | | | |
| 30 | Fri 28 th Mar | Notable pharmacogene variants with large effects. Tracking on polygenic pharmacogenetic | | |
| | 16:00-17:00 | traits - GWAS, high throughput analysis. (Jane Farrar) | | |
| 30 | Fri 28 th Mar | Selection on complex traits and the liability scale for complex disorders. (Russell McLaughlin) | | |
| | 17:00-18:00 | | | |
| 32 | Mon 31 st Mar | Selection on complex traits and the liability scale for complex disorders. (Russell McLaughlin) | | |
| | 17:00-18:00 | | | |

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|----|---|---|--|--|--|
| 32 | Tue 1 st Apr 17:00- 18:00 | Variants in pharmacogenes can influence disease risk. Actionable pharmacogenetic traits in clinical practice - the individualisation of medicine. (Jane Farrar) | | | |
| 32 | Wed 2 nd Apr 17:00-18:00 | Mapping complex disease genes I: GWAS and polygenic risk. (Russell McLaughlin) | | | |
| 32 | Fri 4 th Feb 16:00- 17:00 | Mapping complex disease genes II: missing heritability and rare variants (Russell McLaughlin) | | | |
| 32 | Fri 4 th Feb 17:00- 18:00 | Tutorial (Jane Farrar) | | | |
| 33 | Mon 7 th Feb 17:00-18:00 | Tutorial (Russell McLaughlin) | | | |
| 33 | Tue 8 th Feb 17:00-18:00 | TBC | | | |
| 33 | Wed 9 th Feb 17:00-18:00 | TBC | | | |
| 33 | Fri 11 th Feb 16:00-17:00 | TBC | | | |
| 33 | Fri 11 th Feb 17:00-18:00 | TBC | | | |

NOTE: Venue LTEE3.

Description of Lectures:

Jane Farrar

History of and importance of medical genetics: Topics including disease prevalence (congenital, Mendelian and multifactorial disorders including cancer), the practical value of genetic information, inter alia, the role of genetic counseling and the potential of emerging new therapeutics based on knowledge of disease etiology, are outlined.

Overview of spectrum of Mendelian and multifactorial disorders: A reminder on nomenclature is given including transition, transversion, missense, nonsense, frameshift, dynamic, dominant-negative, haploinsufficiency etc. A brief description of phenotypes associated with autosomal dominant, recessive and X-linked disorders is provided, emphasising that the majority of such conditions are clinically significant with many being highly distressing, serious and life-threatening. The genetics underpinning multifactorial disorders and methodologies utilised to elucidate these genetic factors. Identification of disease genes and disease mutations. An overview of the development and utilisation of methodologies to identify disease causing genes and mutations. Application of genome sequencing in the era of genomic medicine.

Variant interpretation and ACMG guidelines. New trends in medical genetics: In the era of big data from whole genome sequencing (WGS) interpretation of novel genetic variants is key; innovative methods for interpretation. Use of the American College of Medical Genetics (ACMG) guidelines in clinical genetics. Targeted therapies directed by genetic information – current status for gene therapy, gene editing etc.

Introduction to pharmacogenomics. Genetic variation between individuals greatly influences drug response, drug efficacy and / or drug toxicity and indeed is relevant to all of us - on average every human carries a number of actionable pharmacogenetic variants. Such genetic information is leading to the individualisation of medicine and has enormous ramifications for medicine and drug development. An introduction to methods used to identify genetic variants for monogenic and polygenic pharmacogenetic traits will be discussed. Genes encoding drug metabolising enzymes (DMEs) will be introduced including Phase I and Phase II DMEs. The significant effects of genetic variants in the

thiopurine methyltransferase (TPMT) gene (a Phase II DME) on response to major chemotherapeutic and immunosuppressant drugs will be outlined.

Genetic variation in cytochrome P450s and drug response: The family of genes encoding cytochrome P450 enzymes (Phase I DMEs) will be introduced including CYP2C9 and CYP2D6, among others. Genetic variants in these DME genes influence metabolism of drugs, including commonly used medications such as warfarin and codeine, among others. A summary of the studies underpinning the now actionable pharmacogenomic outcomes associated with warfarin and codeine therapies will be provided. Genetic information can be utilised to target the optimal therapy at the right dose to the correct patient population.

Notable pharmacogene variants with large effects. Tracking on polygenic pharmacogenetic traits - GWAS, high throughput analysis. Genetic variants that can cause catastrophic drug responses will be discussed, as will how these are becoming actionable in clinical practice. Methods used to identify genetic factors that influence he risk of an adverse drug response between individuals will be reviewed. Parallels between such pharmacogenomic studies and the use of GWAS, WGS to decipher the genetic determinants of complex disorders will be highlighted.

Variants in pharmacogenes influence disease risk. Actionable pharmacogenetic traits in clinical practice - the individualisation of medicine:

While pharmacogenetic traits underpin much of the variance between individuals in response to medications, genetic variants in pharmacogenes can also influence disease risk. Many DMEs metabolise not just exogenous compounds (therapeutics) but also endogenous compounds, and therefore it is not surprising that genetic variants in these genes can contribute to disease risk - the mounting evidence that genetic variants in pharmacogenes can impact disease risk will be reviewed. Actionable outcomes from pharmacogenomic studies will be discussed highlighting that genetic factors greatly impact how each of us responds to medication with enormous implications for medicine and drug development. An overview will be provided of pharmacogenetic traits that have obtained FDA/EMA backing with actionable outcomes in clinical practice. The barriers that have hindered translation of pharmacogenetic information from the laboratory into clinical practice will be discussed.

Russell McLaughlin

Introduction to complex traits: In this lecture we define the concept of complex traits, framed against classical Mendelian genetics and alternative models of genetic architecture. This lecture also explores the problem of defining phenotypes in the presence of clinical and genetic heterogeneity.

Quantitative traits and complex disorders: This lecture sets the statistical framework for following lectures, defining basic population parameters including mean, variance and distribution, using a worked example of human height. The liability scale is introduced as a prototype for modelling complex disorders as quantitative traits.

Variance components and heritability: Here we demonstrate how phenotypic variance can be decomposed into genetic and non-genetic components, allowing the estimation of heritability (the fraction of phenotypic variance due to genetic variance). Methods for estimating heritability are discussed, including worked examples with inbred plant lines and monozygotic twins.

Dominance, additivity and interaction: Our genetic model for complex traits is further decomposed to explicitly define variance conferred by dominance effects, gene-gene and gene-environment interactions and additive effects of trait-increasing alleles. Methods for heritability estimation using non-twin pedigree data are defined.

Selection on complex traits and the liability scale for complex disorders: This lecture discusses the practical application of statistical models for complex traits, explaining models of variance components established in previous lectures through the lens of plant and animal breeding. The equivalence between this and the liability scale for complex disease is delineated.

Mapping complex disease genes I: GWAS and polygenic risk: Having established the overall model for complex diseases, this lecture now lays out a key method used to understand the genetics of complex disease: the genome-wide association study (GWAS). We also describe and explain how GWAS data can be used to understand the genetic architecture of complex diseases (e.g., polygenic/non-polygenic) and to estimate the fraction of overall heritability explained by common variants.

Mapping complex disease genes II: missing heritability and rare variants: This lecture defines the missing heritability problem along with explanations and solutions for understanding the genetic architecture of disease in the post-GWAS era, specifically in the context of rare variants. We shift our focus from GWAS and rare single-nucleotide variants to some other types of genetic variation that are likely to play a major role in the aetiology of complex diseases. The lecture takes a dive into a worked example of the role of repeat expansions in the neurodegenerative disease amyotrophic lateral sclerosis.

8. Learning Outcomes:

On completion of the module students will:

- Have gained an overview of the history of medical genetics and understand the current stateof-the art in this rapidly changing and exciting field
- Have knowledge regarding the genetic basis of mendelian and complex disease
- Have gained insights into methods of analyses used in medical genetics; linkage studies, genome-wide association studies (GWAS), next generation sequencing (NGS), data analyses tools etc
- Understand the genetics of polygenic traits, the concept of heritability and methods to estimate
 it
- Have insights into emerging themes in personalized medicine that influence drug design, drug choice and drug response and the importance of genetic information for medicine and drug development
- Understand the relevance of medical genetics to diagnosis, prognosis and treatment of disease

9. Recommended Reading List:

'Genetic and Genomics and Medicine', (Garland Science) - Tom Strachan, Judith Goodship and Patrick Chinnery

'Introduction to Genetic Analysis' (Macmillan Education) Griffiths, Wessler, Carroll & Doebly

Primary references will be provided in lecture materials

10. Assessment Details: One 1.5-hour exam paper at the end of semester 2

11. Module Coordinator Jane Farrar gifarrar@tcd.ie

4.8 BIU33250 Introduction to Immunology & Immunometabolism

1. Module Code BIU33250 (Open Module)

2. Module Name Introduction to Immunology & Immunometabolism

3. Semester taught Semester 2

4. Contact Hours 22

5. Module Personnel Aisling Dunne (AD), Jean Fletcher (JF), Michael Carty (MC), Ed Lavelle (EL), Luke

O'Neill (LON)

6. Learning Aims This module introduces to the basic components and function of the immune

system – the molecules, cells, tissues and organs that make up the immune system. It will illustrate the immune responses to infection. Additionally, it will introduce students to the principles of integrated metabolism and bioenergetics before considering how they are dysregulated in inflammatory diseases and also

how we can harness this knowledge for new immunotherapies.

7. Module content:

| Lecture # | Lecture Topic | Lecturer |
|-----------|--|----------|
| 1 | Innate Immunity 1- Introduction to the Immune System | AD |
| 2 | Innate Immunity 2 – Innate Defences | AD |
| 3 | Innate Immunity 3 – Cellular Response to infection | AD |
| 4 | Innate Immunity 4 – PRR Signalling | AD |
| 5 | Innate Immunity 5 – Cytokines | AD |
| 6 | Innate Immunity 6 – DCs & Antigen Presentation | JF |
| 7 | Adaptive Immunity & Infection 1 – T-cell Receptor | JF |
| 8 | Adaptive Immunity & Infection 2– T-cell Signalling | |
| 9 | Adaptive Immunity & Infection 3 – T Effector T-cells | JF |
| | Revision Tutorial: Innate Immunity | |
| 10 | Adaptive Immunity & Infection 4 - B-lymphocytes & Plas | MC |
| 11 | Adaptive Immunity & Infection 5 – Antibodies | |
| 12 | Adaptive Immunity & Infection 6 – Infection & Covid-19 | EL |
| 13 | Adaptive Immunity & Infection 7 - Vaccination EL | |
| 14 | Immunometabolism 1 - Metabolites as Signalling Molec LON | |
| 15 | Immunometabolism 2 – Applied Immunometabolism LON | |
| 16 | Immunometabolism 3 – Immunometabolism & disease | AD |
| | Revision Tutorial: Adaptive Immunity | TBC |

8. Learning Outcomes:

On successful completion of this module students will be able to:

- Identify cells, receptors and soluble component of the innate immune system and how they function to eliminate pathogen.
- Define how an adaptive immune response is initiated and how different types of adaptive immune responses are used to eliminate particular pathogens.
- Identify how the immune system can cause disease and how it can be exploited therapeutically.
- Recall the principals of central energy and intermediary metabolic pathways and appreciate their importance in cellular function.
- Apply knowledge on cellular metabolism to diseases including cancer and inflammation.

9. Recommended Reading List:

The recommended text for this module is Janeway's Immunobiology published by Norton's Books, currently in its 10th Edition.

Further reading will be given out by lecturers during the module.

10. Delivery

This Open module consists of 16 scheduled lectures (plus 2 tutorials) which will be delivered either online through Panopto (pre-recorded) or face to face. Any recordings will be released online in Blackboard and students will be notified beforehand. The in-class MCQ will be made available on the Blackboard after Reading/revision Week 1 at a flexible slot. The end of Semester exam will be an inperson, written exam paper.

11. Assessment Details:

60% End of year examination, 40% in course assessed.

In course assessment: In-class end of module MCQ exam covering lecture material End of year exam; 3 essay questions, answer 2 – specific instructions will be given on paper.

12. Module Coordinator Dr Aisling Dunne

Email: aidunne@tcd.ie Phone 01-8961612

Executive Officer: Úna Murphy

Email: MURPHYU1@tcd.ie

Phone: 018962437

13. Module Website https://tcd.blackboard.com/webapps/login/

4.9 BIU33475 Basics of Neurobiology

***Last year schedule and module descriptor. New timetable and details will be provided on Blackboard. Please use this information as a guide. All lectures are recorded and available to students who have Timetable clashes**

1. Module Code BIU33475 (Open Module) Module)

2. Module Name Basics of Neurobiology

3. Semester taught Semester 2

4. Contact Hours 16 hours (16 Lectures)5. Module Personnel Gavin Davey & David Loane

6. Learning Aims: This module focuses on chemical transmission between neurons, how neurotransmitters are classified and identified and describes typical and atypical neurotransmitters and their functions in the brain. It considers mechanisms in which abnormal neurotransmission gives rise to common neurological & psychiatric disorders.

7. Module content: Programme of lectures and practicals –

| Week | Lecture Topic & Lecturer | | | | |
|--------|--|--|--|--|--|
| Semest | Semester | | | | |
| 22 | Intro: cell types in the brain and their functions; NT types; NT criteria (GD) | | | | |
| 22 | Techniques for studying neurotransmission (GD) | | | | |
| 22 | Acetylcholine release & exocytosis (GD) | | | | |
| 23 | Biogenic Amines I (GD) | | | | |
| 23 | Biogenic Amines II & brain disorders (GD) | | | | |
| 23 | Glutamatergic neurotransmission systems | | | | |
| 24 | GABAergic neurotransmission systems (GD) | | | | |
| 24 | Atypical Neurotransmitters I (GD) | | | | |
| 24 | Atypical Neurotransmitters II (GD) | | | | |
| 25 | Brain lipids, gangliosides & lipid mediators (GD) | | | | |
| 25 | Intracellular trafficking & signalling (GD) | | | | |
| 25 | Inborn metabolic diseases of the brain (GD) | | | | |
| 26 | Inborn metabolic diseases of the brain (DL) | | | | |
| 26 | Neurobiology of schizophrenia & autism (DL) | | | | |
| 27 | Neurobiology of mood and anxiety disorders (DL) | | | | |
| 27 | Neurobiology of addiction (DL) | | | | |

8. Learning Outcomes:

On completion of this module, the student will be able to:

- Describe the cell types in the brain and common techniques that enable chemicals with neurotransmitter-like properties to be identified
- Understand the criteria that need to be satisfied in order for a chemical to be classified as a neurotransmitter
- Develop a knowledge of the biogenic amines (acetylcholine, dopamine, noradrenaline, adrenaline, serotonin) and the properties that allow them to be classified as neurotransmitters
- Develop a knowledge of glutamate and GABA and the properties that allow them to be classified as neurotransmitters
- Develop a knowledge of atypical neurotransmitters (NO, CO, D-serine, neuropeptides, purines) and the properties that allow them to be classified as neurotransmitters

• Develop a knowledge of how dysfunctional neurotransmitter systems give rise to common neurological & psychiatric disorders

9. Recommended Reading List:

Basic Neurochemistry (Siegal, Albers, Brady, Price) Academic Press, 7th Edition. (6th Edition is online free at https://www.ncbi.nlm.nih.gov/books/NBK20385/?term=basic%20neurochemistry)
Principles of Neural Science by Eric Kandel , James Schwartz , Thomas Jessell , Steven Siegelbaum , A.J. Hudspeth

10. Assessment Details: Examination (70% written examinations; 30% continual assessment).

11. Module Coordinator: Dr Gavin Davey

Email: gdavey@tcd.ie Phone: 018968408

Executive Officer: Gabrielle Mc Cabe

Email: GAMCCABE@tcd.ie

Phone: 018964195

5. Review Instructions

Please note: due to the length and number of reviews to be assessed, we may not be able to return marks to you before the annual exams in May.

Format

Your review must not exceed 4,000 words (i.e. <u>all</u> text excluding in the title page, index/table of contents, figure legends, tables, plagiarism declaration, appendix and the references section). It must be typed in Times New Roman 12 point font, with a line spacing of 1.5. It must be submitted not later than Friday, on week 19 (January 5th, 2023), with the <u>word count verified and included in the submitted version (see appendix n V)</u>.

- The work should be divided into: Abstract; Introduction; Main text in sections according to topic; Conclusion; References.
- Pages must be numbered.
- Figures and Tables: Each Figure/Table must be numbered (Figure 1, etc.). Figures must have a <u>legend</u> (text attached to the Figure that explains what it shows). Each Figure/Table must be referred to from a sentence in the main text, to tell the reader when to look at it. You are allowed to copy Figures and Tables from papers or websites, provided that the legend cites the source clearly ("Figure taken from Smith et al., 2011").
- Citations: When the text refers to a published paper, the citations <u>in the text</u> must use a format like these examples:
 - XYZ was observed (Behan, 2011). For papers with 1 author.
 - XYZ was observed (Behan and Murphy, 2011). For papers with 2 authors.
 - XYZ was observed (Behan et al., 2011). For papers with 3 or more authors.
 - Most references are cited at the ends of sentences like this (Behan et al., 2011). However, it is sometimes more useful to write something like Behan et al. (2011) found that XYZ was not true.
 - Do not use a citation system based on numbers.
 - Do not include the initials of the authors in the citations in the main text.
- References section: The references section (also called the bibliography) is the list of papers that have been cited in the text. It appears at the end of the review. It gives more details of the papers that have been cited: complete list of authors (initials and surname); year of publication; title of the article; journal name; volume number; page numbers (first and last). Example:
 - Behan M, Cahill S, Murphy C (2011) The plastid genome of higher plants. Nature Reviews Genetics 103: 56-58.
- References to websites should not be used as a substitute for the primary published literature in the field under review and should only be cited if there is no published paper as an alternative. If you need to cite a website, put the address (URL) directly in the text of as a footnote, not in the references section.
- The review's title and your name should be on the front cover.
- Make sure you are aware of College policies regarding plagiarism (Appendix I, II and III) and complete the 'Ready, Steady, Write' online tutorial.
- Review submission is online and you must upload an electronic copy of your review to *Blackboard* before the deadline.

Declaration information

Please create a **separate** word document to accompany your essay or review submission which contains **your Name & Student Number** and the following **DECLARATION** information;

I have read and I understand the plagiarism provisions in the General Regulations of the University Calendar for the current year, found at: http://www.tcd.ie/calendar
I have also completed the Online Tutorial on avoiding plagiarism 'Ready, Steady, Write', located at http://tcd-ie.libguides.com/plagiarism/ready-steady-write

The statement should be signed (electronic signature will suffice) and dated. A template is also available (Appendix n V)

The review must be your own work. Note that your supervisor will be able to gauge your level of understanding of the subject matter in your meetings with them, and this may be taken into account in their assessment of the final review.

6. Assessments guidelines:

The overall Junior Sophister results will represent 30% of your final moderatorship grade. Modules are assessed by continuous assessment and/or by examination. The distribution scheme of marks between papers continuous assessment and practical work varies with each module and it is specified within its description. Specific exam dates as well as submission deadlines are also specified for each module and it is vital that you submit on time.

- Word limit: Word limit is strictly enforced. However, a margin of 10% above the limit is allowed and there is no penalty below. **Shorter, concise answers are encouraged**. Limit may not apply for some LENS students.
- Time: The allocated time includes submission of the answers. A 5-10min lateness will be allowed without penalty. Some LENS students are allowed extra time and, although module coordinator will be aware, the student should make sure required accommodations are taken into account.
- Submission: Blackboard is the preferred submission platform. Submissions are allowed in the system past the deadline (they are flagged in red as late in Blackboard).

<u>Penalties</u> are applied when time or length are exceeded, and it only applies to the time or length beyond the allowed margin.

- Exceeding the word limit: Marker will consider when grading the ability of the student to adhere to the specified limit.
- Exceeding the time limit: Open book exams, 5% grade reduction/15min.
 Assignments, review 5% grade reduction/day.

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

No penalties will apply if there are mitigating circumstances or the student has been allowed to exceed the limits. The onus is on the student to provide evidence of mitigating conditions. These incidences should be approved on a case-by-case basis by the Course Coordinator for consistency.

Mitigating circumstances are well documented technical problems. LENS students' particular circumstances or sickness properly justified.

Appendices

I. Plagiarism

Plagiarism is regarded as a <u>serious offence</u> by the University and could result in censure by the Junior Dean. Proven instances of plagiarism will result in heavy penalties.

A full statement of the College's position on plagiarism can be found in the College Calendar and is reproduced (Calendar 2022-23) here:

Plagiarism

96 General

It is clearly understood that all members of the academic community use and build on the work and ideas of others. It is commonly accepted also, however, that we build on the work and ideas of others in an open and explicit manner, and with due acknowledgement.

Plagiarism is the act of presenting the work, form of words or ideas of others as one's own, without due acknowledgement.

Plagiarism can arise from deliberate actions and also through careless thinking and/or methodology. The offence lies not in the attitude or intention of the perpetrator, but in the action and in its consequences.

It is the responsibility of the author of any work to ensure that he/she does not commit plagiarism.

Plagiarism is considered to be academically fraudulent, and an offence against academic 48 Calendar 2022-23

integrity that is subject to the disciplinary procedures of the University.

97 Examples of Plagiarism

Plagiarism can arise from actions such as:

- (a) copying another student's work;
- (b) enlisting another person or persons to complete an assignment on the student's behalf;
- (c) procuring, whether with payment or otherwise, the work or ideas of another;
- (d) quoting directly, without acknowledgement, from books, articles or other sources, either in

printed, recorded or electronic format, including websites and social media;

- (e) paraphrasing, without acknowledgement, the writings of other authors;
- (f) using another person's form of words without quotation marks (this constitutes plagiarism even if the student provides a reference to that person or their work).

Examples (d) and (e) in particular can arise through careless thinking and/or methodology where students:

- (i) fail to distinguish between their own ideas and those of others;
- (ii) fail to take proper notes during preliminary research and therefore lose track of the sources from which the notes were drawn;
- (iii) fail to distinguish between information which needs no acknowledgement because it is firmly in the public domain, and information which might be widely known, but which nevertheless requires some sort of acknowledgement; (iv) come across a distinctive methodology or

All the above serve only as examples and are not exhaustive.

idea and fail to record its source.

98 Plagiarism in the context of group work Students should normally submit work done in co-operation with other students only when it is done with the full knowledge and permission of the lecturer concerned. Without this, submitting work which is the product of collaboration with other students may be considered to be plagiarism.

When work is submitted as the result of a group project, it is the responsibility of all students in the group to ensure, so far as is possible, that no

TR060 – GENETICS (Junior Sophister)

work submitted by the group is plagiarised. In order to avoid plagiarism in the context of collaboration and group work, it is particularly important to ensure that each student appropriately attributes work that is not their own.

99 Self plagiarism

No work can normally be submitted for more than one assessment for credit. Resubmitting the same work for more than one assessment for credit is normally considered self-plagiarism. 100 Avoiding plagiarism

Students should ensure the integrity of their work by seeking advice from their lecturers, tutor or supervisor on avoiding plagiarism. All schools and departments must include, in their handbooks or other literature given to students, guidelines on the appropriate methodology for the kind of work that students will be expected to undertake. In addition, a general set of guidelines for students on avoiding plagiarism is available on http://libguides.tcd.ie/plagiarism. 101 If plagiarism as referred to in §96 above is suspected, in the first instance, the Director of Teaching and Learning (Undergraduate), or their designate, will write to the student, and the student's tutor advising them of the concerns raised. The student and tutor (as an alternative to the tutor, students may nominate a representative from the Students' Union) will be invited to attend an informal meeting with the Director of Teaching and Learning (Undergraduate), or their designate, and the lecturer concerned, in order to put their suspicions to the student and give the student the opportunity to respond. The student will be requested to respond in writing stating his/her agreement to attend such a meeting and confirming on which of the suggested dates and times it will be possible for them to attend. If the student does not in this manner agree to attend such a meeting, the Director of Teaching and Learning (Undergraduate), or designate, may refer

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the case directly to the Junior Dean, who will interview the student and may implement the

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie

procedures as referred to under CONDUCT AND COLLEGE REGULATIONS §2.

102 If the Director of Teaching and Learning (Undergraduate), or designate, forms the view that plagiarism has taken place, he/she must decide if the offence can be dealt with under the summary procedure set out below. In order for this summary procedure to be followed, all parties attending the informal meeting as noted in §101 above must state their agreement in writing to the Director of Teaching and Learning (Undergraduate), or designate. If one of the parties to the informal meeting withholds his/her written agreement to the application of the summary procedure, or if the facts of the case are in dispute, or if the Director of Teaching and Learning (Undergraduate), or designate, feels that the penalties provided for under the summary procedure below are inappropriate given the circumstances of the case, he/she will refer the case directly to the Junior Dean, who will interview the student and may implement the procedures as referred to under CONDUCT AND COLLEGE REGULATIONS §2.

103 If the offence can be dealt with under the summary procedure, the Director of Teaching and Learning (Undergraduate), or designate, will recommend one of the following penalties:

- (a) Level 1: Student receives an informal verbal warning. The piece of work in question is inadmissible. The student is required to rephrase and correctly reference all plagiarised elements. Other content should not be altered. The resubmitted work will be assessed and marked without penalty;
- (b) Level 2: Student receives a formal written warning. The piece of work in question is inadmissible. The student is required to rephrase and correctly reference all plagiarised elements. Other content should not be altered. The resubmitted work will receive a reduced or capped mark depending on the seriousness/extent of plagiarism; (c) Level 3: Student receives a formal written
- (c) Level 3: Student receives a formal written warning. The piece of work in question is inadmissible. There is no opportunity for resubmission with corrections. Instead, the student is required to submit a new piece of

TR060 – GENETICS (Junior Sophister)

work as a reassessment during the next available session. Provided the work is of a passing standard, both the assessment mark and the overall module mark will be capped at the pass mark. Discretion lies with the Senior Lecturer in cases where there is no standard opportunity for a reassessment under applicable course regulations.

104 Provided that the appropriate procedure has been followed and all parties in §101 above are in agreement with the proposed penalty, the Director of Teaching and Learning (Undergraduate) should in the case of a Level 1 offence, inform the course director and where appropriate the course office. In the case of a Level 2 or Level 3 offence, the Senior Lecturer must be notified and requested to approve the recommended penalty. The Senior Lecturer may approve, reject, or vary the recommended penalty, or seek further information before making a decision. If the Senior Lecturer considers that the penalties provided for under the summary procedure are inappropriate given

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the circumstances of the case, he/she may also

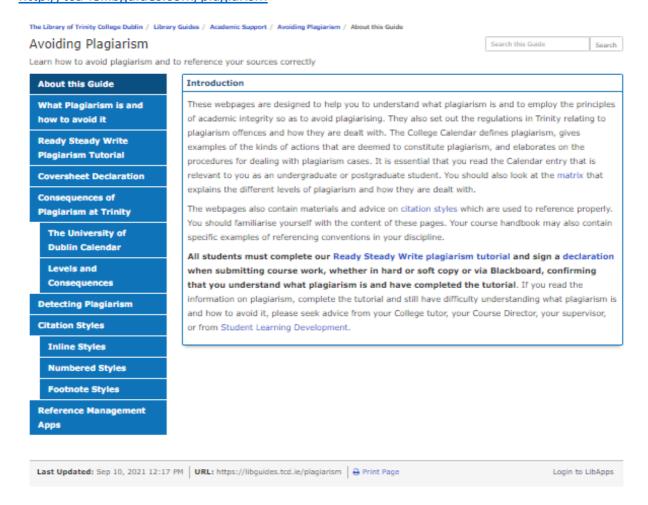
refer the matter directly to the Junior Dean who will interview the student and may implement the procedures as referred to under CONDUCT AND COLLEGE REGULATIONS §2. Notwithstanding his/her decision, the Senior Lecturer will inform the Junior Dean of all notified cases of Level 2 and Level 3 offences accordingly. The Junior Dean may nevertheless implement the procedures as referred to under CONDUCT AND COLLEGE REGULATIONS §2. 105 If the case cannot normally be dealt with under the summary procedures, it is deemed to be a Level 4 offence and will be referred directly to the Junior Dean. Nothing provided for under the summary procedure diminishes or prejudices the disciplinary powers of the Junior Dean under the 2010 Consolidated Statutes. [1] UG: Calendar Part II, General Regulations, Academic Progress, Paragraphs 96 and following.

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Course Coordinator: Juan Pablo

II. Online central repository

All students are required to access the online central repository in which all information and resources on plagiarism have been consolidated. This facility explains what plagiarism is, and how it can be avoided. The central repository is being hosted by the Library and is located at http://tcd-ie.libguides.com/plagiarism



- Plagiarism Policy https://www.tcd.ie/teaching-learning/academic-policies/assets/plagiarism-mar2020.pdf
- Calendar, Part III, General Regulations & Information, Section I 'Plagiarism'
 https://www.tcd.ie/calendar/graduate-studies-higher-degrees/complete-part-III.pdf

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III. Ready Steady Write Plagiarism Tutorial

All students are required to complete the Ready Steady Write plagiarism tutorial, a resource developed by the Centre for Academic Practice and eLearning (CAPSL) at Trinity College Dublin, to help you understand and avoid plagiarism and develop your academic writing skills and academic integrity. www.tcd.ie/Library/support/plagiarism/story.html

Plagiarism can occur in many forms, for example copying another student's work, or quoting directly from published sources without acknowledgement, or using as your own slightly modified versions of the published work of others. Thus, in writing essays or other project work you are warned against copying verbatim, or copying and making minor modifications to, phrases, sentences, paragraphs, sections or illustrations from other published work.

Students and staff have access to Turnitin computer software (see Appendix IV) that can readily detect plagiarism. The Department will use this sensitive anti-plagiarism tool to screen essays and other forms of formal assessed work and Turnitin reports can be used as evidence if plagiarism is suspected. Accordingly, you are strongly recommended to synthesize your own language at all times. A full statement of the College's position on plagiarism can be found in the College Calendar

IV. Turnitin - Blackboard

Turnitin is an online software program that aids plagiarism prevention. It allows students and lecturers to check students' work for academic integrity by searching for text that is improperly cited or potentially plagiarised. Once uploaded to Turnitin, assignments are compared to millions of books, journal articles, web pages and student papers, identifying any unoriginal material within the essay. The software then creates an Originality Report which highlights and quantifies unoriginal content.

For more information, see http://tcd-ie.libguides.com/plagiarism/detecting-plagiarism and to access the student training tutorial, see http://www.turnitin.com/en_us/training/student-training

V. Declaration to be included on literature review TRINITY COLLEGE DUBLIN THE UNIVERSITY OF DUBLIN

SCHOOL OF GENETICS AND MICROBIOLOGY

SMURFIT INSTITUTE OF GENETICS

DECLARATION FOR REVIEW

I have read and I understand the plagiarism provisions in the General Regulations of the University Calendar for the current year, found at http://www.tcd.ie/calendar.

I have also completed the Online Tutorial on avoiding plagiarism 'Ready Steady Write', located at http://tcd-ie.libguides.com/plagiarism/ready-steady-write.

I declare that this Review does not contain material which has been **PLAGIARISED**.

| Signed | |
|---|-----------------------------|
| Dated | |
| The word count of this document the References section is: | nent (with the exception of |
| Signed | |
| Dated | |

VI.Important dates and timetable by semester

| Module | Module name | Туре | Semester | week | submission date | Submission time |
|-----------|--|--|------------|-------|----------------------------|------------------------------|
| BIU33150 | Biochemistry for Biosciences | MCQ 1 (24%) | Semester 1 | ТВС | ТВС | ТВС |
| GEU33007 | Molecular Genetics Laboratory | Lab report 1 (50%) | Semester 1 | 9 | Fri 25th Oct | 17:00 |
| GEU33075 | Evolutionary and Population Genetics | poster submission | Semester 1 | 11 | Mon 4th Nov | 09:00 |
| GEU33075 | | poster presentation (35%) | Semester 1 | 12 | Wed 13th Nov | 14:00-17:00 |
| GEU33075 | Evolutionary and Population Genetics | Reflective essay | Semester 1 | 12 | Fri 15th Nov | 17:00 |
| MIU33302 | | Group presentations | Semester 1 | 13 | Tue 19 th Nov | 11:00-12:00 & 14:00-15:00 |
| MIU33302 | Molecular genetics I | Group presentations | Semester 1 | 13 | Thu 21 st Nov | 15:00-16:00 |
| MIU33302 | Molecular genetics I | Group presentations | Semester 1 | 13 | Fri 22 nd Nov | 10:00-11:00 |
| GEU33285 | Science Structure, Discussion and Presentation for Hum Genetics | One essay 1500 words (15%) | semester 1 | 14 | Mon 25th Nov | 17:00 |
| GEU33075 | Evolutionary and Population | MCQs (60%) | Semester 1 | 14 | Thu 28th Nov | 10:00-12:00 |
| GEU33007 | Molecular Genetics Laboratory | Lab report 2 (50%) | Semester 1 | 14 | Fri 29th Nov | 17:00 |
| BIU33150 | Biochemistry for Biosciences | MCQ 2 (16%) | Semester 1 | ТВС | ТВС | ТВС |
| GEU33085 | Science Structure, Discussion and Presentation for Genetics | written review 4000 words (70%) | Semester 2 | 19 | Fri 3 rd Jan | 17:00 |
| GEU33301 | IRiointormatics | Bioinformatics Assessment (33%) | Semester 2 | 26 | Mon 17 th Feb | 11:00-13:00 |
| GEU33008 | Analytical Genetics Laboratory | MCQ (50%) | Semester 2 | 27 | Tuesday or Wednesday | ТВС |
| GEU33301 | IRIOINTORMATICS | Programming Assessment (33%) | Semester 2 | 29 | Thu 13 th Mar | 10:00-12:00 |
| GEU33008 | Analytical Genetics Laboratory | Lab report (50%) | Semester 2 | 30/31 | (TBC) | 17:00 |
| BIU33250 | Introduction to Immunology & Immunometabolism | MCQS (30%) | Semester 2 | 29 | ТВС | ТВС |
| GEU33035 | Genetic Analysis of Nervous Systems | Test (50%) | Semester 2 | 29 | Mon 10th Mar | 15:00-16:00 |
| GEU33035 | Genetic Analysis of Nervous Systems | class participation/essay (50%) | Semester 2 | 31 | ТВС | ТВС |
| BIU33475 | Basics of Neurobiology | continuous assessment (Literature Review) (30%) | Semester 2 | 33 | April (TBC) | ТВС |
| GEU333301 | Bioinformatics | Report submission (34%) | Semester 2 | 35 | Thu 10 th April | 11:00-13:00 |

Guidelines on Awarding Grades for Examinations and Essays Answers in the Sophister Years

Note that these guidelines are for use as a general reference. Differences may occur between disciplines.

| | Mark | Criteria |
|------|--------|--|
| I | 90-100 | IDEAL ANSWER; showing insight and originality and wide knowledge. Logical, accurate and concise presentation. Evidence of reading and thought beyond course content. Contains particularly apt examples. Links materials from lectures, practicals and seminars where appropriate. |
| | 80-89 | OUTSTANDING ANSWER; falls short of the 'ideal' answer either on aspects of presentation or on evidence of reading and thought beyond the course. Examples, layout and details are all sound. |
| | 70-79 | MAINLY OUTSTANDING ANSWER; falls short on presentation and reading or thought beyond the course, but retains insight and originality typical of first class work. |
| II-1 | 65-69 | VERY COMPREHENSIVE ANSWER; good understanding of concepts supported by broad knowledge of subject. Notable for synthesis of information rather than originality. Sometimes with evidence of outside reading. Mostly accurate and logical with appropriate examples. Occasionally a lapse in detail. |
| | 60-64 | LESS COMPREHENSIVE ANSWER; mostly confined to good recall of coursework. Some synthesis of information or ideas. Accurate and logical within a limited scope. Some lapses in detail tolerated. |
| II-2 | 55-59 | SOUND BUT INCOMPLETE ANSWER; based on coursework alone but suffers from a significant omission, error or misunderstanding. Usually lacks synthesis of information or ideas. Mainly logical and accurate within its limited scope and with lapses in detail. |
| | 50-54 | INCOMPLETE ANSWER; suffers from significant omissions, errors and misunderstandings, but still with understanding of main concepts and showing sound knowledge. Several lapses in detail. |
| III | 45-49 | WEAK ANSWER; limited understanding and knowledge of subject. Serious omissions, errors and misunderstandings, so that answer is no more than adequate. |
| | 40-44 | VERY WEAK ANSWER; a poor answer, lacking substance but giving some relevant information. Information given may not be in context or well explained, but will contain passages and words, which indicate a marginally adequate understanding. |
| F-1 | 35-39 | MARGINAL FAIL; inadequate answer, with no substance or understanding, but with a vague knowledge relevant to the question. |
| | 30-34 | CLEAR FAILURE; some attempt made to write something relevant to the question. Errors serious but not absurd. Could also be a sound answer to the misinterpretation of a question. |
| F-2 | 0-29 | UTTER FAILURE; with little hint of knowledge. Errors serious and absurd. Could also be a trivial response to the misinterpretation of a question. |
| U.G | | Ungraded |

Faculty of Engineering, Mathematics and Science - Guidelines on Marking, last modified 2007.

VII. Academic year structure 2024/25

The University of Dublis

Academic Year Calendar 2024/25

| Anadomis Calendar Week | Week beginning | | | | | |
|---|--|---|---|---|--|--|
| | | UG continuing years / PG all years UG new first years | | | | |
| 1 | 26-Aug-24 | Reassessment * (Semesters 1 & 2 of 2023/24) | | ←Michaelmas Term begins/Semester 1 begins | | |
| 2 | 02-Sep-24 | Orientation (Postgradusts, Visiting & Erasmus); Marking/Results | | | | |
| 3 | 09-Sep-24 | Teaching and Learning | | +Michaelmas tracking term begins | | |
| 4 | 16-Sep-24 | Teaching and Learning | Orientation (JF UG) | | | |
| 5 | 23-Sep-24 | Teaching and Learning | Teaching and Learning | | | |
| 6 | 30-Sep-24 | Teaching and Learning | Teaching and Learning | | | |
| 7 | 07-Oct-24 | Teaching and Learning | Teaching and Learning | | | |
| | 14-Oct-24 | Teaching and Learning | Teaching and Learning | | | |
| 9 | 21-0ct-24 | Study/Review | Study/Review | | | |
| 10 | 28-Oct-24 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | | | |
| 11 | 04-Nov-24 | Teaching and Learning | Teaching and Learning | | | |
| 12 | 11-Nov-24 | Teaching and Learning | Teaching and Learning | | | |
| 13 | 38-Nov-24 | Teaching and Learning | Teaching and Learning | | | |
| 14 | 25-Nov-24 | Teaching and Learning | Teaching and Learning | | | |
| 15 | 02-Dec-24 | Revision *1 | Revision *1 | | | |
| 16 | 09-Dec-24 | Assessment #1 | Assessment #1 ~ | ←Michaelmas term ends Sunday 15 December 2024/Semester 1 ends | | |
| 17 | 16-Dec-24 | | | | | |
| 38 | 23-Dec-24 | Christmas Period - College closed 24 December 2024 to 1 January 2025 Inclusive | Christmas Period - College closed 24 December 2024 to 1 January 2025 Inclusive | | | |
| 19 | 30-Dec-24 | 24 Describer 2024 to 134 mary 2023 House | 24 December 2004 to 2 2015thy 2023 include | | | |
| 20 | 06-lan-25 | Foundation Scholarship Examinations A | | | | |
| 21 | 13-lan-25 | Marking/Results | Marking/Results | ←Hlary Term begins/Semester 2 begins | | |
| 22 | 20-lan-25 | Teaching and Learning | Teaching and Learning | ←Hlary teaching term begins | | |
| 23 | 27-lan-25 | Teaching and Learning | Teaching and Learning | | | |
| 24 | 03-Feb-25 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | | | |
| 25 | 10-Feb-25 | Teaching and Learning | Teaching and Learning | | | |
| 26 | 17-Feb-25 | Teaching and Learning | Teaching and Learning | | | |
| 27 | 24-Feb-25 | Teaching and Learning | Teaching and Learning | | | |
| 28 | 03-Mar-25 | Study/Review | Study/Review | | | |
| 29 | 30-Mar-25 | Teaching and Learning | Teaching and Learning | | | |
| 30 | 17-Mar-25 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | | | |
| 31 | 24-Mar-25 | Teaching and Learning | Teaching and Learning | | | |
| 32 | 31-Mar-25 | Teaching and Learning | Teaching and Learning | | | |
| 33 | 07-Apr-25 | Teaching and Learning | Teaching and Learning | | | |
| 34 | 14-Apr-25 | Revision (Friday, Good Friday) | Revision (Friday, Good Friday) | 6-Hillary Term ends Sunday 20 April 2025 | | |
| 35 | 21-Apr-25 | Assessment *2 (Monday, Easter Monday) | Assessment *2 (Monday, Easter Monday) | ←Trinity Term begins | | |
| 36 | 28-Apr-25 | Trinity Week (Monday, Trinity Monday) *2 | Trinity Week (Monday, Trinity Monday) *2 | | | |
| 37 | 05-May-25 | Marking/Results (Monday, Public Holiday) | Marking/Results (Monday, Public Holiday) | | | |
| 38 | 12-May-25 | Marking/Results | Marking/Results | | | |
| 39 | 19-May-25 | Marking/Results | Marking/Results | | | |
| 40 | 26-May-25 | Research | Research | ←Trinity Term ends Sunday 1 June 2025/Semester 2 ends | | |
| 41 | 02-Jun-25 | Research (Monday, Public Holiday) | Research (Monday, Public Holiday) | | | |
| 42 | 09-Jun-25 | Research | Research | | | |
| 43 | 16-Jun-25 | Research | Research | | | |
| 44 | 23-Jun-25 | Research | Research | | | |
| 45 | 30-Jun-25 | Research | Research | | | |
| 46 | 07-Jul-25 | Research | Research | | | |
| 47 | 14-Jul-25 | Research | Research | | | |
| 48 | 21-Jul-25 | Research | Research | | | |
| 49 | 20-Jul-25 | Research | Research | | | |
| 50 | 04-Aug-25 | Research (Monday, Public Holiday) | Research (Monday, Public Holiday) | | | |
| 51 | 11-Aug-25 | Research | Research | | | |
| 52 | 18-Aug-25 | Research + | Research + | | | |
| * Note: ad | ditional/contingency day | s may be required outside of the formal assessment/rea | sssessment weeks. | | | |
| | * Note: It may be necessary to hold a small number of # examinations/assessments outside of semester 1. | | | | | |
| A Note: It i | may be necessary to hol | d some examinations/assessments in the preceding wee | sk. | | | |
| | | usion: 09-Dec-24 to 14-Dec-24 inclusive (contingency d | | | | |
| * Note: semester 2 Assessment session: 22-Apr-25 to 02-May-25 inclusive (includes Council approved contingency dates: 29-Apr-25 to 02-May-25) | | | | | | |
| | Note: the academic year structure is due to be reviewed during 2024/25 - any changes will be notified should Council approve any change. | | | | | |

Learning Section 2017/2017

Key Dates:

Orientation Week:Monday 02 September to Friday 6 September 2024Study/Review Week:Monday 21 October to Friday 25 October 2024Revision Week Semester 1:Monday 2 December to Friday 6 December 2024

Study/Review Week:Monday 3 March to Friday 7 March 2025Revision Week Semester 2:Monday 14 April to Friday 18 April 2025Trinity week:Monday 21 April to Friday 25 April 2025

VIII. Prizes and other awards

RONALD A. FISHER PRIZE IN GENETICS The Ronald A. Fisher prize in genetics is awarded annually to a Sophister student who has excelled in oral presentation of a topic of his/her own choice within the field of genetics. The prize was established by a gift from George Dawson who founded the Department of Genetics in 1958 and led it until 1987. Sir Ronald Fisher, Baldwin Professor of Genetics at Cambridge (1943-57), was acknowledged as a leading authority on genetical theories of natural selection and statistics and was a major influence on George Dawson, a student at Cambridge. The prize will be awarded on the advice of the Head of the Department of Genetics. Value, €191

DAWSON PRIZE IN GENETICS This prize was founded in 1990 by colleagues and friends of George Dawson, member of staff (1950-87) and Professor of Genetics (1967-87). He founded the department in 1959 and started a programme of summer research for rising Senior Sophisters. The prize will be awarded, on the advice of the Professor of Genetics, to the student of genetics who has distinguished himself/herself in the Junior Sophister examinations.

Value, **€316**

A.W.B. VINCENT SCHOLARSHIPS IN GENETICS AND HUMAN GENETICS These scholarships are awarded to students in genetics and human genetics, selected by competition, to facilitate research internships at institutions for two to three months in the summer of their Junior Sophister year. William Vincent, LL.D. (h.c.), (1919-2012) established these scholarships in 1975 and funded them for 40 years. His family and friends contributed to ensure they would continue after his death. Dr Vincent was president of the American Irish Foundation from 1972 to 1987 when it merged with the American Ireland Fund of which he became vicechairman. He was a generous supporter of many charitable projects in Ireland. Value, €1500

IX. Safety statement

To ensure the health and safety of everyone in the Genetics department, we will share with you the departmental Safety Statement. We ask you to read and abide by the rules given in this Safety Statement. Please note that the failure to comply with the procedures outlined in the departmental Safety Statement may result in disciplinary action.

The following rules are of particular importance for undergraduate students who enter laboratories and lecture theatres and must be followed at all times:

- Students are not allowed to enter laboratories unless they are authorized to do so.
- Students are not permitted to work in laboratories unsupervised.
- Students must follow the instructions of laboratory supervisors at all times.
- Eating and drinking is not permitted in laboratories and lecture theaters.
- Smoking is strictly prohibited in all campus buildings.
- Students must wear a suitable laboratory coat while working in a laboratory.
- Safety glasses must be worn in laboratories when there is access/use of chemicals or potential exposure to biological agents containing aerosols. Those wearing spectacles for vision correction must wear Pulsafe glasses which are placed over the normal spectacles.
- For laboratory work, wear long trousers or skirts and shoes with non-slip soles that fully cover your feet. Open-toed sandals, flip-flops, high heels, ballet-style, crocs and canvas shoes/runners are not permitted.
- For laboratory work, long hair must be properly tied back and adequately restrained.
- No loose hanging jewellery or headphones are permitted in the laboratory.
- Gloves must be worn and changed as required in all laboratory environments involving the use of chemicals or biological agents.
- Coats, bags or personal belongings must not be left on lab benches or anywhere where they could cause an obstruction.
- Students should not congregate at the entrance to a lab or lecture theatre or at building entrances.
- If any glass apparatus/container/pipette breaks while in use, inform a member of staff immediately.
- In the event of a fire alarm, students must leave the institute immediately following the evaculation routes outlined in the departmental safety statement.
- Ensure caps are replaced on all containers with chemicals when an experiment is completed.
- If you come into direct contact with chemicals, inform a member of staff immediately.
- Familiarize yourself with the location of first aid kits, safety showers and eye wash stations in the laboratory you are working in.
- Students must familiarize themselves with the European Standard Chemical hazard symbols shown below.



If you have any concerns about Health and Safety in the department, please contact the departmental Safety Officer (currently Prof. Frank Wellmer) or the Head of Department (currently Prof. Matthew Campbell).

For further information on Health and Safety, see the website of the College Safety office at www.tcd.ie/estatesandfacilities/health-and-safety/

X. Careers Advisory Service

What do you want to do? How will you get there? We are here to support you in answering these and other questions about your career.

Junior and Senior Fresh Students

Get Involved: Remember that your course of study, extra-curricular activities, voluntary and part-time work all provide opportunities for developing skills and gaining an insight into your career preferences. In your Senior Fresh year, look out for short-term internship opportunities.

MyCareer: Log in to MyCareer to keep abreast of jobs, study and careers events of interest to you.

Junior Sophisters

Attend class seminar: Typically this takes place in Hilary term and includes information on applying for work experience and internships and postgraduate study.

Get work experience: The programme of summer work experience and internships is particularly relevant to Junior Sophisters. Personalise your MyCareer profile to receive email alerts tailored to your preferences.

MyCareer: Log in to MyCareer to keep abreast of jobs, study and careers events of interest to you.

Finalists and Senior Sophisters

Meet Employers and/or Explore Further Study: You may have decided to seek employment directly after graduation and many employers visit Dublin to actively seek out talented graduates. For others, further study may be their preferred option. Your

MyCareer dashboard will keep you informed.

Find Jobs: Personalise your MyCareer profile to receive email alerts tailored to your interests.

Attend class seminar: Typically this takes place in Michaelmas term and includes information on applying for postgraduate study and jobs.

GradLink Mentoring: An opportunity to get advice and support from a Trinity graduate.

Drop-In CV/ LinkedIn Clinics: We also provide support at a practical level, helping you to improve your applications, which will benefit you in securing your future, whether in employment or further study.

Practice Interviews: A practice interview tailored to the job/ course of your choice with practical feedback.

MyCareer: Log in to MyCareer to keep abreast of jobs, study and careers events of interest to you.

MyCareer

An online service that you can use to:

- Apply for opportunities which match your preferences - vacancies including research options
- Search opportunities- postgraduate courses and funding
- View and book onto employer and CAS events
- Submit your career queries to the CAS team
- Book an appointment with your Careers Consultant

Simply login to MyCareer using your Trinity username and password and personalise your profile.

Careers Advisory Service
Trinity College Dublin, 7-9 South Leinster Street, Dublin 2
01 896 1705/1721 | Submit a career query through MyCareer.



Opening Hours

During term: 9.30am - 5.00pm, Monday - Friday
Out of Term: 9.30am - 12.30pm & 2.15 - 5.00pm, Monday - Friday