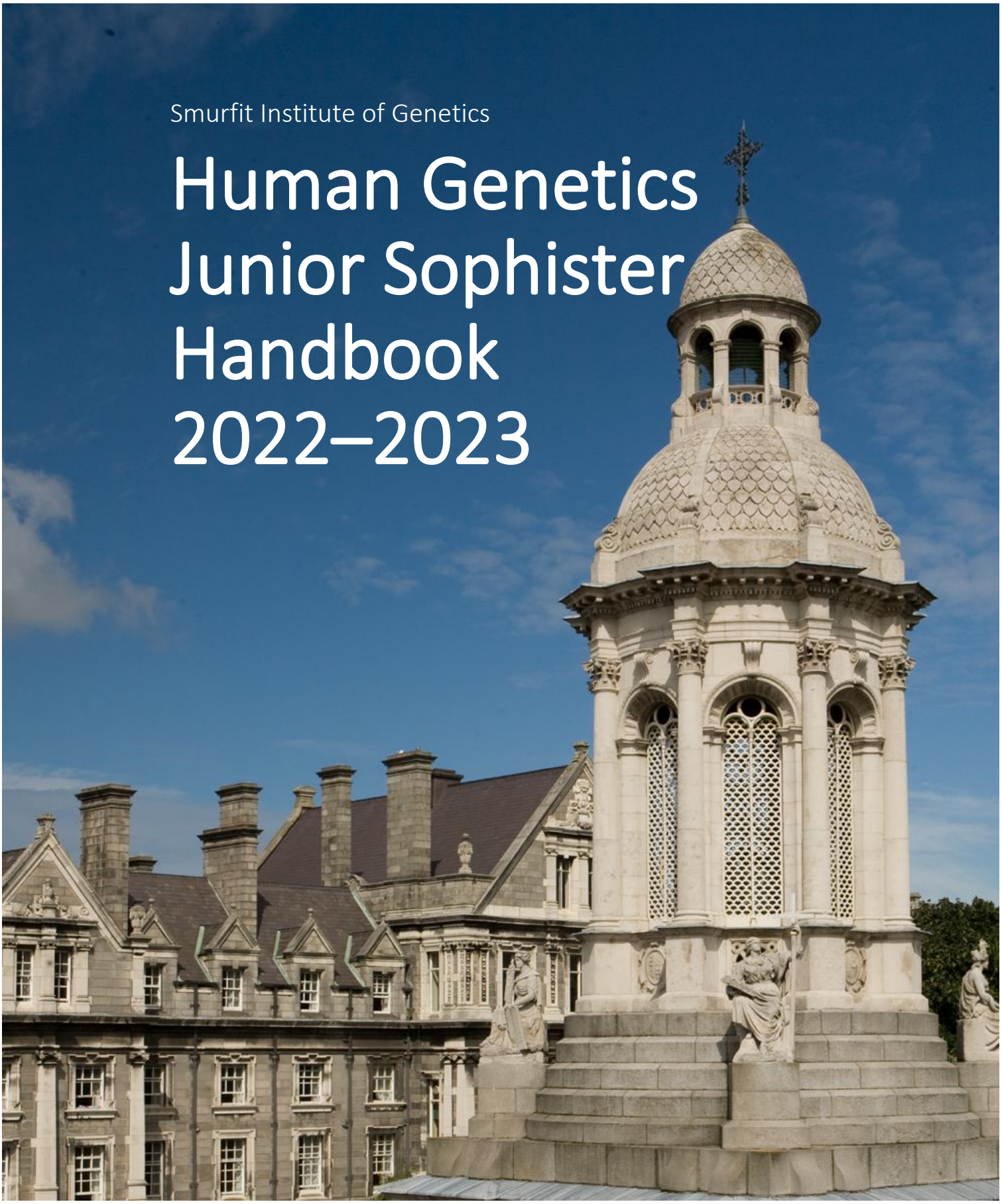
Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

Smurfit Institute of Genetics

Human Genetics Junior Sophister Handbook 2022–2023



labradoj@tcd.ie

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labradoj@tcd.ie

1. Introduction

It is a pleasure for me to welcome you to your Junior Sophister year in Human 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 Human 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.

A note on college life and COVID-19

In order to offer taught programmes in line with government health and safety advice, registered students are expected to be available to attend in-person teaching activities. Any request not to attend in person for exceptional reasons (such as travel restrictions or underlying health conditions) will be considered on a case-by-case basis by the relevant Head of School in consultation with College Health and there is no guarantee that these requests can be facilitated. It will depend on whether the programme learning outcomes and modes of assessment can be met through remote attendance. We would encourage all students to adhere to the safety protocols when on campus for in-person teaching activities or student club and society events, i.e., mask wearing, hand washing, cough etiquette and to maintain social distancing. https://www.tcd.ie/about/coronavirus/

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 Human 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 Human Genetics.

Human Genetics	
Semester 1 (S1)	**Semester 2 (S2)**
Core Modules	
GEU33015 Eukaryotic Molecular Genetics (5 credits)	GEU33215 Medical Genetics (5 credits)
GEU33007 Molecular Genetics Laboratory (5 credits)	GEU33025 Data Handling and Bioinformatics (5 credits)
GEU33075 Evolutionary and Population Genetics (5 credits)	GEU33035 Genetic Analysis of Nervous Systems (5 credits)
GEU33285 Science Structure Discussion and Presentation for Human Genetics (5 credits)	GEU33008 Analytical Genetics Laboratory (5 credits)
Open Modules Scenario I	
GEU33045 Genomics and Systems Biology (5 credits)	GEU33055 Developmental Genetics (5 credits)
Trinity Elective (5 credits)	BIU33250 Introduction to Immunology and Immunometabolism (5 credits) **OR** BIU33475 Basic Neurobiology (5 credits)
Open Modules Scenario II	
GEU33045 Genomics and Systems Biology (5 credits)	GEU33055 Developmental Genetics (5 credits)
BIU33150 Biochemistry for Biosciences (5 credits)	Trinity Elective (5 credits)
Open Modules Scenario III	
GEU33045 Genomics and Systems Biology (5 credits)	GEU33055 Developmental Genetics (5 credits)
Trinity Elective (5 credits)	Trinity Elective (5 credits)

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. Please note that attendance may be in person or in real-time online or lectures may be recorded depending on TCD policies relating to Covid-19.

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 Field Trip

A field trip takes place over two days on the week 27 (2nd – 3rd March 2023, 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.

2.6 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

2.7 Summer Vacation Research Seminars

The current rising Senior Sophister students will present their research seminars towards the end of Michaelmas term. You will be advised of the date and venue of these seminars and will be expected to attend.

2.8 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.

2.9 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)*

2.10 Prizes in Genetics

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

a) Leslie Bloomer Prize in Human Genetics 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.

2.11 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 2022-23). Please check the most recent version of the calendar for any updates (TCD Calendar):

"*58 ...*

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 credit-weighted 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 failed components of 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).

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 if you obtain a mark of < 35% in 3 or more modules you will be unable to progress 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.

2.12 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.
Photocopier – in the atrium of the Smurfit Institute.

2.13 Course coordinators:

Course Coordinator: Juan Pablo Labrador labradoj@tcd.ie
Executive Officer: Alicia Vega genetics@tcd.ie

Module code	Module	Coordinator	Email
GEU33007	Molecular Genetics Laboratory	Dr Conor Delaney	DELANEC9@tcd.ie
GEU33008	Analytical Genetics Laboratory	Prof Juan Pablo Labrador	LABRADOJ@tcd.ie
GEU33015	Eukaryotic Molecular Genetics	Prof Kevin Mitchel	Kevin.Mitchell@tcd.ie
GEU33025	Data Handling and Bioinformatics	Dr Karsten Hokamp	KAHOKAMP@tcd.ie
GEU33035	Genetic Analysis of Nervous Systems	Prof Juan Pablo Labrador	LABRADOJ@tcd.ie
GEU33045	Genomics & Systems Biology	Dr Kevin Daly	DALYK1@tcd.ie
GEU33055	Developmental Genetics	Prof Seamus Martin	MARTINSJ@tcd.ie
GEU33065	Plant and Microbal Genetics	Prof Frank Wellmer	WELLMERF@tcd.ie
GEU33075	Evolutionary and Population Genomics	Prof Russell McLauglin	mclaugr@tcd.ie
GEU33285	Science Structure, Discussion and presentation for Human Genetics	Prof Juan Pablo Labrador	LABRADOJ@tcd.ie
GEU33215	Medical Genetics	Prof Jane Farrar	gjfarrar@tcd.ie
BIU33150	Module Name Biochemistry for Biosciences	Prof D Nolan	denolan@tcd.ie
BIU33250	Module Name Introduction to Immunology & Immunometabolism	Prof Frederick J Sheedy	fsheedy@tcd.ie

BIU33475	Basics of Neurobiology	Prof Gavin Davey	Daveygdavey@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 GEU33015 Eukaryotic Molecular Genetics

1. Module Code **GEU33015**
2. Module Name Eukaryotic Molecular Genetics
3. Semester taught Semester 1
4. Contact Hours 24
5. Module Personnel Adrian Bracken; Sophia Kiang; Kevin Mitchell; Seamus Martin
6. Learning Aims The aim of this module is to introduce students to advanced concepts in the molecular genetics of eukaryotes. A major focus will be on the complexities of gene expression and its regulation. This will include transcription by RNA polymerase II, the role of the Mediator complex, and the processing steps involved in the maturation of pre-mRNAs: capping, polyadenylation and splicing. The regulation of gene expression and its critical importance in differentiation and development will be explored at the level of chromatin and nucleosome modifications, and in relation to the combinatorial interactions between transcription factors and *cis*-acting upstream regulatory elements such as enhancers. Approximately one-third of the course will explore recombinant DNA techniques used in gene expression and genome analysis.
7. Module content:

Week	Day & Time	Lecture Topic & Lecturer
8	Mon 9:00-10:00	mRNA transcription: RNA polymerase II; the core promoter; general transcription factors (Kiang)
8	Mon 10:00-11:00	Enhancers, Silencers and combinatorial control of gene expression: transcription factors. (Kiang)
8	Mon 14:00-15:00	Importance of gene regulation in differentiation, development and evolution. (Kiang)
9	Review week	
10	Tue 11:00-12:00	Processing of pre-mRNAs, capping, polyadenylation, splicing. (Kiang)
10	Tue 14:00-15:00	mRNA translation; localization and translational control. Non-coding RNAs (Kiang)
11	Mon 9:00-10:00	Non coding RNAs continued; RNA quality control (NMD) and mRNA turnover. (Kiang)
11	Mon 10:00-11:00	Tutorial 2: Online / Face to Face TBC (Kiang)
11	Mon 14:00-15:00	Chromatin and the nucleosome: the histone code hypothesis. (Bracken)
11	Tue 11:00-12:00	Histone modifications: writers, readers and erasers. (Bracken)
11	Tue 14:00-15:00	The 'Epigenome'; role of enhancers; genomic imprinting. (Bracken)
12	Mon 9:00-10:00	Protein folding and posttranslational modifications (Mitchell)
12	Mon 10:00-11:00	Protein localization and targeted degradation (Mitchell)
12	Mon 14:00-15:00	Recombinant DNA techniques: DNA-modifying enzymes (Mitchell)
12	Tue 11:00-12:00	Cloning vectors and strains (Mitchell)
12	Tue 14:00-15:00	PCR and cloning strategies (Mitchell)
13	Mon 9:00-10:00	Cloning by recombination: Gateway cloning (Mitchell)
13	Mon 10:00-11:00	Chromatin: local and higher order structure of chromosomes. (Bracken)
13	Mon 14:00-15:00	Polycomb repressor complexes and DNA methylation systems: roles In development and Disease. (Bracken)
13	Tue 11:00-12:00	Tutorial 1: Online (Bracken)
13	Tue 14:00-15:00	Tutorial 3: Online (Mitchell)
14	Mon 9:00-10:00	Generation of polyclonal and monoclonal antibodies and their applications (Martin)
14	Mon 10:00-11:00	Antibodies as diagnostic and research tools; therapeutic applications of antibodies (Martin)
14	Mon 14:00-15:00	Expressing recombinant proteins in bacteria, yeast and insect systems (Martin)
14	Tue 11:00-12:00	Expressing recombinant proteins in mammalian expression systems (Martin)
14	Tue 14:00-15:00	Making genomic libraries; recap of lecture material (Martin)
15	Revision Week	
16	Assessment Week	

TR060 – HUMAN GENETICS (Junior Sophister) Course Coordinator: Juan Pablo Labrador

labradoj@tcd.ie

NOTE: Venue LTEE3

8. Learning Outcomes: Upon successful completion of this module, students will be able to describe the critical features of gene expression and its regulation in eukaryotes. They will, for example, understand how pre-mRNAs are transcribed in the nucleus and processed to give mature mRNAs. Students will also have gained knowledge and an appreciation of the complexities of gene regulation at the level of chromatin and the nucleosome, and the binding of transcription factors to upstream *cis*-acting regulatory elements in DNA. In addition, students will have acquired an in-depth knowledge of the key recombinant DNA techniques that underpin molecular genetics analyses.

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.

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

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

11. Module Coordinator: Kevin Mitchell Kevin.Mitchell@tcd.ie

3.2 GEU33007 Molecular Genetics Laboratory

1. Module Code **GEU33007**
2. Module Name Molecular Genetics Laboratory
3. Semester taught Semester 1
4. Contact Hours 5 hours per week
5. Module Personnel Conor Delaney
David Noone (+ 3 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 14 Sep	Pr 1. Exploring gene regulation in E. coli: the lac operon p5	1 of 5
	Pr 2. Gene transfer and expression in plant cells p12 Transformation of Agrobacterium with pC-TAK1 p13	1 of 6
Fri 16 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 21 Sep	Pr 1. PCR amplification of lac operon sequences p9	3 of 5
	Pr 3. Exploring the genome and proteome of phage λ p17 Growth and isolation of phage λ p18	1 of 7
	Pr 2. Purification of Agrobacterium transformants p13	2 of 6
Fri 23 Sep	Pr 1. Agarose gel analysis of lac operon PCR products p10	4 of 5
	Pr 3. Recovery of precipitated λ virions p18	2 of 7
	Week 5	
Wed 28 Sep	Pr 3. Purification of λ virions on a CsCl step gradient p18	3 of 7
	Pr 2. Liquid culture of Agrobacterium (pC-TAK1) p13	3 of 6
	Pr 1. Sequence analysis of lac operon PCR products p10	5 of 5
	Pr 3. Extraction of phage λ DNA p19	3 of 7
Fri 30 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 5 Oct	Pr 4. Creating a library of λ genes in a plasmid vector p23 Isolation and gel analysis of pUC19 plasmid DNA 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
Fri 7 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 12 Oct	Pr 3. SDS-PAGE analysis of λ virion proteins p20	6 of 7
	Pr 4. Preparation of 'competent' E. coli cells and transformation with ligated pUC19-λ DNA p27	3 of 7
Fri 14 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 19 Oct	Pr 4. Isolation of putative pUC19-λ recombinant plasmids Restriction digestion of plasmid DNAs p27	5 of 7
Fri 21 Oct	Pr 4. Agarose gel analysis of digested plasmid DNAs p28	6 of 7
	Assignment of the First Lab Report – Due October 28th, 5pm	
Date	Project No. Page	Day
	Week 3	
Wed 14 Sep	Pr 1. Exploring gene regulation in E. coli: the lac operon p5	1 of 5
	Pr 2. Gene transfer and expression in plant cells p12 Transformation of Agrobacterium with pC-TAK1 p13	1 of 6
Fri 16 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 21 Sep	Pr 1. PCR amplification of lac operon sequences p9	3 of 5
	Pr 3. Exploring the genome and proteome of phage λ p17 Growth and isolation of phage λ p18	1 of 7
	Pr 2. Purification of Agrobacterium transformants p13	2 of 6
Fri 23 Sep	Pr 1. Agarose gel analysis of lac operon PCR products p10	4 of 5
	Pr 3. Recovery of precipitated λ virions p18	2 of 7
	Week 5	
Wed 28 Sep	Pr 3. Purification of λ virions on a CsCl step gradient p18	3 of 7
	Pr 2. Liquid culture of Agrobacterium (pC-TAK1) p13	3 of 6
	Pr 1. Sequence analysis of lac operon PCR products p10	5 of 5
	Pr 3. Extraction of phage λ DNA p19	3 of 7
Fri 30 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 5 Oct	Pr 4. Creating a library of λ genes in a plasmid vector p23 Isolation and gel analysis of pUC19 plasmid DNA 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
Fri 7 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 12 Oct	Pr 3. SDS-PAGE analysis of λ virion proteins p20	6 of 7
	Pr 4. Preparation of 'competent' E. coli cells and transformation with ligated pUC19-λ DNA p27	3 of 7
Fri 14 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 19 Oct	Pr 4. Isolation of putative pUC19-λ recombinant plasmids Restriction digestion of plasmid DNAs p27	5 of 7
Fri 21 Oct	Pr 4. Agarose gel analysis of digested plasmid DNAs p28	6 of 7
	Assignment of the First Lab Report – Due October 28th, 5pm	

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 5th; and the second report during the final practical on November 18th.

The first report is due at 17:00 Irish Time, October 28th 2022.

The second report is due at 17:00 Irish Time, December 2nd 2022.

11. Module Coordinator: Conor Delaney delanec9@tcd.ie
Matthew 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: 24 hours; In-person tutorials: 3.5 hours
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 10:00-11:00	Introduction to evolutionary and population genetics (Cassidy; in-person LTEE3)
3	Mon 14:00-15:00	Tutorial: Introduction (in-person LTEE3)
3	Mon 12pm onwards	Spectrum and mechanisms of DNA mutation I (McManus; pre-recorded)
3	Mon 12pm onwards	Spectrum and mechanisms of DNA mutation II (McManus; pre-recorded)
3	Mon 12pm onwards	Exogenous and structural causes of mutation (McManus; pre-recorded)
3	Mon 12pm onwards	Mutation and health (McManus; pre-recorded)
4	Mon 12pm onwards	Genetic variation and its detection I (McLaughlin; pre-recorded)
4	Mon 12pm onwards	Genetic variation and its detection II (McLaughlin; pre-recorded
4	Mon 12pm onwards	Alleles and genotypes: Hardy-Weinberg equilibrium (McLaughlin; pre-recorded)
4	Mon 12pm onwards	Alleles and genotypes: inbreeding (McLaughlin; pre-recorded)
4	Mon 12pm onwards	Effective population size (McLaughlin; pre-recorded)
4	Mon 14:00-15:00	Tutorial: Open office hours (McManus; in-person or online TBC) Human Genetics
4	Tues 15:00-16:00	Tutorial: Open office hours (McManus; in-person or online TBC) Genetics
5	Mon 12pm onwards	Genetic drift, fixation F-statistics and population structure (McLaughlin; pre-recorded)
5	Mon 12pm onwards	Linkage disequilibrium (McLaughlin; pre-recorded)
5	Mon 12pm onwards	Applied population genetics: complex traits and GWAS (McLaughlin; pre-recorded)
5	Mon 12pm onwards	Applied population genetics: population structure and PCA (McLaughlin; pre-recorded)
5	Mon 11:00-14:00	Project group tutorials (in-person/ Cassidy & Russell office)
6	Mon 12pm onwards	Evolutionary change in sequences: measuring evolution (Cassidy; pre-recorded)
6	Mon 12pm onwards	Evolutionary change in sequences: patterns and models (Cassidy; pre-recorded)
6	Mon 12pm onwards	Evolutionary change in sequences: the molecular clock (Cassidy; pre-recorded)
6	Mon 12pm onwards	Evolutionary change in sequences: molecular phylogenetics (Cassidy; pre-recorded)
6	Mon 12pm onwards	Evolutionary change in sequences: applied phylogenetics (Cassidy; pre-recorded)
6	Mon 14:00-15:00	Tutorial: Open office hours (McLaughlin; in-person or online TBC) Human Genetics
6	Tues 15:00-16:00	Tutorial: Open office hours (McLaughlin; in-person or online TBC) Genetics
7	Mon 12pm onwards	Genome evolution: gene duplication (Cassidy; pre-recorded)
7	Mon 12pm onwards	Genome evolution: concerted evolution (Cassidy; pre-recorded)
7	Mon 12pm onwards	Genome evolution: whole genome duplication (Cassidy; pre-recorded)
7	Mon 12pm onwards	Genome evolution: gene loss and base composition evolution (Cassidy; pre-recorded)

7	Mon 12pm onwards	Genome evolution: Transposition (Cassidy; pre-recorded)
7	Mon 11:00-14:00	Project group tutorials (in-person/ Cassidy & Russell office)
8	Mon 11:00-12:00	Tutorial: Open office hours (Cassidy; in-person or online TBC) Human Genetics
8	Mon 12:00-13:00	Tutorial: Open office hours (Cassidy; in-person or online TBC) Genetics
9	Study/Review week	
10	Thu 10:00-12:00	MCQ Exam venue (MAC LAB)
10	Thu 14:00-16:00	Project group tutorials (in-person)
12	Mon 9:00	Project hand-in deadline online submission 9:00am
12	Thu 12:00-15:00	Project presentation (Atrium at the Smurfit)
13	Fri 17:00	Reflective report online submission 5:00 pm

NOTE: Venue LTEE3 and Lecturer's office (more info from lecturers)

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 bias.
- **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%) and continual assessment (40%). The continual assessment is subdivided into a group poster project (35%), which includes an individual contribution mark, and a short reflective essay (5%).

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 be asked to present their poster, either online or at an in-person event.

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: Russell McLaughlin russell.mclaughlin@tcd.ie

labradoj@tcd.ie

3.4 GEU33285 Science Structure, Discussion and Presentation for Huma Genetics

1. Module Code **GEU33285**
2. Module Name **Science Structure, Discussion and Presentation for Human Genetics**
3. Semester taught Semester 2
4. Contact Hours 12
5. Module Personnel Adrian Bracken, Dan Bradley, Matthew Campbell, Lara Cassidy, Kevin Devine, 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:

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

NOTE: venue Dawson

- 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 11 **Mathematical Genetics I -** Networks/graph theory
- Week 12 **Mathematical Genetics II -** Game theory: Evolution of co-operation/altruism.
- Week 13 **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 6th);
15% tutorial paper presentation
15% of the module networks essay 1500 words (deadline Monday on week 14, November 28th at 5pm)

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

3.5 GEU33045 Genomics & Systems Biology

1. Module Code **GEU33045** (Open Module)
2. Module Name **Genomics & Systems Biology**
3. Semester taught 1
4. Contact Hours 24
5. Module Personnel Adrian Bracken, Carsten Kröger, Kenneth Mok, Kevin Daly
6. Learning Aims The aim of this module is to provide students with a general overview of methods used in the fields of genomics, proteomics and metabolomics and to explain how these methods are used for basic research, biotechnology, agriculture and medicine. To this end, a number of examples from work with diverse organisms (bacteria, fungi, plants, animals including humans) 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	Mon 17:00-18:00	Systems biology and -omics (Daly)
3	Tue 17:00-18:00	Sequencing technology: historical perspective (Daly)
4	Mon 17:00-18:00	History of the Human Genome Project (Daly)
4	Tue 17:00-18:00	2nd Generation Sequencing Technologies (Daly)
5	Mon 17:00-18:00	3rd Generation Sequencing Technologies (Daly)
5	Tue 17:00-18:00	Structural genomics: Reference Genomes and Alignment (Daly)
6	Mon 17:00-18:00	Comparative genomics and evolutionary genomics (Daly)
6	Tue 17:00-18:00	Population genomics and metagenomics (Daly)
7	Mon 17:00-18:00	Transcriptomics (Daly)
7	Tue 17:00-18:00	Functional genomics: Gene finding and annotation (Daly)
7	Wed 18:00-19:00	Functional genomics: the regulatory genome, annotation and networks (Daly)
8	Mon 17:00-18:00	Multi-omics and emerging technologies (Daly)
8	Tue 17:00-18:00	*Revision of material, discussion and answering student questions (Daly)*
9		**Study/Review week**
10	Tue 17:00-18:00	Bacterial genomes and comparative genomics (Kröger)
10	Tue 17:00-18:00	Functional genomics in bacteria (Kröger)
11	Mon 17:00-18:00	Introduction into the epigenome: histone and DNA modifications (Bracken)
11	Tue 17:00-18:00	Methods to analyse the epigenome; the ENCODE project (Bracken)
12	Mon 17:00-18:00	Cancer profiling and classification of tumour types (Bracken)
12	Tue 17:00-18:00	Using genomic information for the development of cancer therapies (Bracken)
13	Mon 17:00-18:00	Proteomics: Identify/characterise/quantify; Mass Spec and other technologies (Mok)
13	Tue 17:00-18:00	Quantitative proteomics; clinical proteomics (Mok)
13	Wed 18:00-19:00	Interaction/affinity proteomics; metabolomics introduction (Mok)
14	Mon 17:00-18:00	Metabolomics technologies (Mok)
14	Tue 17:00-18:00	*Revision of material, discussion and answering student questions (all lecturing staff)*
15		**Revision Week**
16		**Assessment Week**

NOTE: All lectures are prerecorded and available on Blackboard on the scheduled week. All tutorials will be delivered online live.
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 the difference between functional, comparative and structural genomics and will be familiar with the use of genomic technologies in fundamental and medical research, biotechnology and

agriculture. Students will be able to describe how genome sequences are being deciphered and annotated. They will further understand the difference between reductionist and systems 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 Kevin Daly dalyk1@tcd.ie

labradoj@tcd.ie

3.6 BIU33150 Biochemistry for Biosciences

1. Module Code BIU33150 (Open Module)
2. Module Name Biochemistry for Biosciences
3. Semester taught Semester 1
4. Contact Hours 20 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 –

Week	Lecture Topic & Lecturer
	Semester 1
Lect	
	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)
	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)
	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)
	Signalling
17	Signalling 1: Introduction to cell signalling & GPCRs (EC)
18	Signalling 2: G-Protein coupled Receptor (GPCR) regulation (EC)
19	Signalling 3: Receptor tyrosine kinases (RTKs)-PDGF and EGF (DZ/AD)
20	Signalling 4: RTK signalling – PKB and PDK1 (DZ/AD)

labradoj@tcd.ie

Lectures schedule and time table

All lectures will be delivered as follows with the accompanying lecture notes on blackboard : Module BIU33150.

Date of lects (on BB)	Lect No	Staff	Upload to BB by
1 Mon 12/09	1-2	AK	12/09
2 Mon 19/9	3-4	AK/KHM	19/09
3 Mon26/9	5-6	KHM	26/11
4 Mon 3/10	7-8	VK	3/10
5 Mon 10/10	9-10	MC	10/10
6 Mon 17/10	11-12	AB/DN	17/10
7 Reading week			
8 Mon 31/11	13-14	EC	31/10
9. Mon 7/11	15-16	DN	7/11
10 Mon 14/11	17-18	EC	14/11
11 Mon 21/11	19-20	AD/DZ	21/11

(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 assessments.
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 MCQ representing 16% of the marks for the course.

Complete details of the assessments, MCQs and end of term exam, will be posted separately on BB in BIU33150.
Sample MCQs and a sample paper will be available online.

11. Module Coordinator: Dr D Nolan

Email: denolan@tcd.ie
Phone: 8962455

Executive Officer: Úna Murphy
Email: MURPHYU1@tcd.ie
Phone: 018961608

4. Semester 2 Modules

4.1 GEU33215 Medical Genetics

1. Module Code **GEU33215**
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 model 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 16:00-17:0	Importance of medical genetics and history of developments in the field. Jane Farrar
29	Tue 16:00-17:00	Introduction to complex traits. Russell McLaughlin
29	Wed 16:00-17:00	Overview of spectrum of Mendelian and multifactorial disorders. Jane Farrar
29	Friday 17th May Bank holiday no lectures	
30	Mon 16:00-17:0	Quantitative traits and complex disorders. Russell McLaughlin
30	Tue 16:00-17:00	Identification of disease genes and disease mutations. Jane Farrar
30	Wed 16:00-17:00	Variance components and heritability Russell McLaughlin
30	Fri 16:00-17:00	Variant interpretation and ACMG guidelines. New trends in medical genetics. Jane Farrar
31	Mon 16:00-17:0	Introduction to pharmacogenomics. Jane Farrar
31	Tue 16:00-17:00	Genetic variation in cytochrome P450s and influence on drug response. Jane Farrar
31	Wed 16:00-17:00	Dominance, additivity and interaction. Russell McLaughlin
31	Fri 16:00-17:00	tutorial TBC
32	Mon 16:00-17:0	Notable pharmacogene variants with large effects. Tracking on polygenic pharmacogenetic traits - GWAS, high throughput analysis. Jane Farrar
32	Tue 16:00-17:00	Selection on complex traits and the liability scale for complex disorders. Russell McLaughlin
32	Wed 16:00-17:00	Selection on complex traits and the liability scale for complex disorders. Russell McLaughlin
32	Friday 7th April Bank holiday no lectures	

33	Monday 10th April Bank holiday no lectures	
33	Tue 16:00-17:00	Variants in pharmacogenes can influence disease risk. Actionable pharmacogenetic traits in clinical practice - the individualisation of medicine. Jane Farrar
33	Wed 16:00-17:00	Mapping complex disease genes I: GWAS and polygenic risk. Russell McLaughlin
33	Fri 16:00-17:00	Mapping complex disease genes II: missing heritability and rare variants Russell McLaughlin
TBC		Tutorial Jane Farrar
TBC		Tutorial Russell McLaughlin

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.

labradoj@tcd.ie

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 state-of-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: Examination After the end of semester 2

11. Module Coordinator Jane Farrar gjfarrar@tcd.ie

4.2 GEU33025 Data Handling and Bioinformatics

1. Module Code **GEU33025**
2. Module Name Data Handling and Bioinformatics
3. Semester taught Semester 2
4. Contact Hours 39 Hours
5. Module Personnel Karsten Hokamp (KH), Carsten Kröger (CK), Máire Ní Leathlobhair (MNL), 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, more specifically, approaches for the analysis of WGS data.
This module runs for 15 x 2 hours (combined lecture and practical sessions) plus a 5-hour lab practical and 2 x 2-hour in-class assessments in Semester 2.
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	Practical
22	**Wed 9:00-11:00**	Bioinformatics 1 – Biological Databases (FR, 2 hours)	
22	**Fri 11:00-13:00**	Bioinformatics 2 – Protein Resources & Function Prediction (FR, 2 hours)	
23	**Fri 11:00-13:00**	Bioinformatics 3 – Sequence Alignments (FR, 2 hours)	
23	**Fri 14:00-16:00**	Bioinformatics 4 – Multiple Alignments & Genome Browers (FR, 2 hours)	
24	**Wed 9:00-11:00**	Programming 1 – Variables and Loops (KH, 2 hours)	
24	**Wed 14:00-16:00**	Programming 2 – Input/Output, Branching (KH, 2 hours)	
25	**Wed 9:00-11:00**	Programming 3 – Lists, Tuples, Sets (KH, 2 hours)	
25	**Fri 11:00-13:00**	Programming 4 – Dictionaries (KH, 2 hours)	
26	**Mon 11:00-13:00**	Bioinformatics Assessment (FR, 2 hours)	
26	**Fri 11:00-13:00**	Programming 5 – Functions, System Commands (KH, 2 hours)	

26		TBC week 26 or 29	Biolab (CK, 5 hours)
28		**Study/Review week**	
29		TBC week 26 or 29	Biolab (CK, 5 hours)
29	**Thu 10:00-12:00**	Programming Assessment (KH, 2 hours)	
30	**Thu 11:00-13:00**	NGS Data Analysis 1 – Using R for bioinformatics (MNL, 2 hours)	
30	**Fri 11:00-13:00**	NGS Data Analysis 2 – Graphs & Data Visualization in R (MNL, 2 hours)	
31	**Thu 11:00-13:00**	NGS Data Analysis 3 – Markdown Notebooks in R (MNL, 2 hours)	
31	**Fri 11:00-13:00**	NGS Data Analysis 4 – Genome Assembly & Data Sharing (MNL, 2 hours)	
32	**Thu 11:00-13:00**	NGS Data Analysis 5 – NGS-based approaches for DNA (MNL, 2 hours)	
33	**Thu 11:00-13:00**	NGS Data Analysis 6 – NGS-based approaches for RNA (MNL, 2 hours)	
35	**Wed 17:00**	Report submission Wednesday, April 26th	

NOTE: Venue LTEE3

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 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 - Graphs & Data Visualization in R (MNL, 2 hours) This lecture will cover basic plotting in base R, data visualization with ggplot2 and introductory data visualization theory.

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 – NGS-based approaches for DNA (MNL, 2 hours) Students will be introduced to different high-throughput approaches to DNA sequencing including emerging real-time technologies such as Nanopore.

NGS Data Analysis 6 – NGS-based approaches for RNA (MNL, 2 hours) Students will be introduced to different high-throughput approaches to RNA sequencing including single-cell RNA-Seq methods.

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
ML1O. 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

10. Assessment Details: (1) A bioinformatics exam, (2) a Python programming exam and (3) a report based on NGS data analysis with an R programming component; each of the three parts will contribute ⅓ to the overall module (submission deadline Wednesday, April 26th 5 pm)

11. Module Coordinator: Karsten Hokamp kahokamp@tcd.ie

labradoj@tcd.ie

4.3 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 24
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 12:00-13:00	Experimental Genetics - Introduction - Structure and conservation of genes, nature of mutations and their effects on protein structure and function. (Labrador)
22	Wed 15:00-16:00	Experimental Genetics - Model organisms in genetic research . (Labrador)
22	Thu 9:00-10:00	Experimental Genetics - Experimental manipulation of animal genomes . (Labrador)
22	Thu 11:00-12:00	Experimental Genetics - Creation and use of transgenic animals to probe gene function in vivo. . (Labrador)
23	Tue 12:00-13:00	Developmental Neurogenetics - The purpose and design of genetic screens. . (Labrador)
23	Wed 15:00-16:00	Developmental Neurogenetics - genetic screens – mapping genes. . (Labrador)
23	Thu 9:00-10:00	Developmental Neurogenetics - Genetic analysis of neurogenesis . (Labrador)
23	Thu 11:00-12:00	Developmental Neurogenetics - Genetic analysis of axon guidance . (Labrador)
24	Tue 12:00-13:00	Developmental Neurogenetics - Flip lecture/ Tutorial. (Labrador)
24	Thu 9:00-10:00	Behavioral Genetics - Introduction to Behavioral Genetics - Cell organization and methods of cell biology. (Ramaswami)
24	Thu 11:00-12:00	Behavioral Genetics - Cell biology of neurons and synapse. Structures, electrical properties (Ramaswami)
25	Tue 12:00-13:00	Behavioral Genetics - Cell biology of neurons and synapse. Synaptic transmission and molecular determinants (Ramaswami)
25	Wed 15:00-16:00	Behavioral Genetics - Methods to study nervous systems (behavior, imaging, electrophysiology, anatomy) (Ramaswami)
25	Thu 9:00-10:00	Behavioral Genetics - Flip lecture/ Tutorial (Ramaswami)
25	Thu 11:00-12:00	Behavioral Genetics- Sensory circuits: vision; taste and smell Sensation; Transduction; Perception; Coding; (Ramaswami)
26	Tue 12:00-13:00	Behavioral Genetics - Behavioral Plasticity. (Ramaswami)
26	Wed 15:00-16:00	Behavioral Genetics - Learning and memory. (Ramaswami)
26	Thu 9:00-10:00	Behavioral Genetics- Sleep and Circadian Rhythms. (Ramaswami)
26	Thu 11:00-12:00	Behavioral Genetics – Flip lecture/ Tutorial – Final (Ramaswami)
26	Tue 12:00-13:00	Tutorial - (Ramaswami)
28		**Study/Review week**
30	**TBC**	Final written test

NOTE: LTEE3 excluding Thursday's 9:00-10:00 slot at **LTEE1**

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: Continuos assessment: class participation - TBC (50%) and test on week 30 TBC (50%)

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

labradoj@tcd.ie

4.4 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 week
5. 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 14:00-17:00 Tue 15:00-16:00	Drosophila husbandry, identification of phenotypes, setting up crosses Mendelian inheritance tutorial. P-elements tutorial Monohybrid, dihybrid crosses (Labrador)
23	Mon 14:00-17:00 Tue 15:00-16:00	Drosophila husbandry, identification of phenotypes, setting up crosses Non-Mendelian inheritance tutorial, Lethal mutations. (Labrador) Short Quiz 1: Monohybrid/dihybrid crosses
24	Tue 15:00-16:00	Segregation in Drosophila, identification of males carrying P-elements Lethal mutations , Sex linked inheritance. (Labrador) Short Quiz 2: Lethal mutations
25	Mon 14:00-17:00 Tue 15:00-16:00	Segregation in Drosophila - fly husbandry Sex linked inheritance , Linkage, recombination and mapping. Linkage and mapping tutorial (Labrador) Short Quiz 3: Sex linked inheritance
26	Mon 14:00-17:00 Tue 15:00-16:00	Segregation in Drosophila analysis of crosses Linkage and mapping (Labrador) Short Quiz 4: Linkage, Recombination and mapping
27	Mon 14:00-17:00 Tue 15:00-16:00	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	Mon 14:00-17:00 Tue 15:00-16:00	Segregation in Drosophila – Review of results P-element mapping in Drosophila Tutorial (Labrador) Fly husbandry
30	Mon 14:00-17:00 Tue 15:00-16:00	Analysis and review of P-element results Fly husbandry (Labrador)

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%. Lab report 50%

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

labradoj@tcd.ie

4.5 GEU33055 Developmental Genetics

1. Module Code **GEU33055** (Open Module)
2. Module Name Developmental Genetics
3. Semester taught Semester 2
4. Contact Hours 24
5. Module Personnel Seamus Martin, Adrian Bracken, 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 taught together 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 that dictate the specification of biological structures such as limbs and wings. How cell division is precisely regulated via the 'cell division cycle' will also be discussed in depth. The module will also introduce students to stem cell biology and how stem cells are programmed to undergo growth and differentiation.
7. Module content:

Week	Day & Time	Lecture Topic & Lecturer
22	Wed 11:00-12:00	Intro to development and developmental genetics-Overview (Martin)
22	Wed 14:00-15:00	Pluripotent Stem Cells (Bracken)
22	Thu 14:00-15:00	Multipotent Stem Cells 1 (Bracken)
23	Wed 11:00-12:00	Multipotent Stem Cells 2 (Bracken)
23	Wed 14:00-15:00	Cellular Reprogramming (Bracken)
23	Thu 14:00-15:00	The cell cycle: dissection in different experimental systems. Discovery of the cyclins and cyclin-dependent kinases (Martin)
24	Wed 11:00-12:00	The cell cycle: role of Cyclins and Cyclin-dependent kinases (Martin)
24	Wed 12:00-13:00	The cell cycle: how Cyclin activity is regulated by kinases and phosphates (Martin)
24	Thu 14:00-15:00	The cell cycle: role of Rb and CDK inhibitors. p53 and the DNA damage cell cycle checkpoint (Martin)
25	Wed 11:00-12:00	Introduction to *Drosophila* development and embryogenesis (Martin)
25	Wed 14:00-15:00	*Drosophila* development: anterior-posterior axis specification (Martin)
25	Thu 14:00-15:00	*Drosophila* development: role of maternal effect genes (Martin)
26	Wed 11:00-12:00	*Drosophila* development: activation and function of gap genes (Martin)
26	Wed 14:00-15:00	*Drosophila* development: activation and function of pair-rule genes (Martin)
26	Thu 14:00-15:00	*Drosophila* development: activation and function of segment polarity genes (Martin)
27	Wed 11:00-12:00	*Drosophila* development: dorso-ventral axis specification (Martin)
27	Wed 14:00-15:00	*Drosophila* development: homeotic selector genes and segment identity (Martin)
27	Thu 14:00-15:00	Tutorial: revision of material, discussion and student questions (Martin, Bracken)
28		**Study/Review week**
29	Wed 11:00-12:00	Organogenesis: genetic control of vertebrate limb development (Wellmer)
29	Wed 14:00-15:00	Organogenesis: genetic control of vertebrate limb development II (Wellmer)
29	Thu 14:00-15:00	Organogenesis: Genetic control of *Drosophila* eye development (Wellmer)
30	Wed 11:00-12:00	Principles of plant development (Wellmer)
30	Wed 14:00-15:00	Stem cells, cell fate determination and organogenesis in plants (Wellmer)
30	Thu 14:00-15:00	Tutorial: revision of material, discussion and student questions (Wellmer)

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 2

11. Module Coordinator Seamus Martin Email: MARTINSJ@tcd.ie

labradoj@tcd.ie

4.6 GEU33215 Medical Genetics

1. Module Code GEU33215
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 model 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 16:00-17:00	Importance of medical genetics and history of developments in the field. (Jane Farrar)
29	Tue 16:00-17:00	Introduction to complex traits. (Russell McLaughlin)
29	Wed 16:00-17:00	Overview of spectrum of Mendelian and multifactorial disorders. (Jane Farrar)
29	Friday 17th May Bank holiday no lectures	
30	Mon 16:00-17:0	Quantitative traits and complex disorders. (Russell McLaughlin)
30	Tue 16:00-17:00	Identification of disease genes and disease mutations. (Jane Farrar)
30	Wed 16:00-17:00	Variance components and heritability Russell McLaughlin
30	Fri 16:00-17:00	Variant interpretation and ACMG guidelines. New trends in medical genetics. (Jane Farrar)
31	Mon 16:00-17:0	Introduction to pharmacogenomics. (Jane Farrar)
31	Tue 16:00-17:00	Genetic variation in cytochrome P450s and influence on drug response. (Jane Farrar)
31	Wed 16:00-17:00	Dominance, additivity and interaction. (Russell McLaughlin)
31	Fri 16:00-17:00	No lecture / tutorial TBC
32	Mon 16:00-17:0	Notable pharmacogene variants with large effects. Tracking on polygenic pharmacogenetic traits - GWAS, high throughput analysis. (Jane Farrar)
32	Tue 16:00-17:00	Selection on complex traits and the liability scale for complex disorders. (Russell McLaughlin)
32	Wed 16:00-17:00	Selection on complex traits and the liability scale for complex disorders. (Russell McLaughlin)
32	Friday 7th April Bank holiday no lectures	
33	Monday 10th April Bank holiday no lectures	
33	Tue 16:00-17:00	Variants in pharmacogenes can influence disease risk. Actionable pharmacogenetic traits in clinical practice - the individualisation of medicine. (Jane Farrar)
33	Wed 16:00-17:00	Mapping complex disease genes I: GWAS and polygenic risk. (Russell McLaughlin)
33	Fri 16:00-17:00	Mapping complex disease genes II: missing heritability and rare variants (Russell McLaughlin)
TBC		Tutorial (Jane Farrar)
TBC		Tutorial (Russell McLaughlin)

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 state-of-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 gjfarrar@tcd.ie

labradoj@tcd.ie

4.7 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 24
5. Module Personnel Frederick Sheedy (FS), Jean Fletcher (JF), Michael Carty (MC), Ed Lavelle (EL), Richard Porter (RP), Luke O'Neill (LON), Emma Creagh (EC)
6. Learning Aims This module introduces students 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 importance of central energy and intermediary metabolic pathways and bioenergetics before considering how they are dysregulated in diseases like cancer and also how we can harness this knowledge for new immunotherapies.
7. Module content:

Week	Lecture Topic	Lecturer
22	Innate Immunity 1- Introduction to the Immune System	EC
22	Innate Immunity 2 – Innate Defences	FS
22	Innate Immunity 3 – Cellular Response to infection	FS
22	Innate Immunity 4 – PRR Signalling	FS
23	Innate Immunity 5 – Cytokines	FS
23	Innate Immunity 6 – DCs & Antigen Presentation	JF
23	Adaptive Immunity & Infection 1 – T-cell Receptor	JF
23	Adaptive Immunity & Infection 2– T-cell Activation & Differentiation	JF
24	Adaptive Immunity & Infection 3 - Effector T-cells	JF
24	REVISION TUTORIAL; INNATE IMMUNITY (*Venue/Time tbc, 2h*)	FS/EC
24	Adaptive Immunity & Infection 4 - B-lymphocytes & Plasma Cells	MC
25	Adaptive Immunity & Infection 5 – Antibodies	MC
25	Adaptive Immunity & Infection 6 – Infection & Covid-19	EL
25	Adaptive Immunity & Infection 7 - Vaccination	EL
25	Immunometabolism 1 – Intermediary Metabolism	RKP
26	Immunometabolism 2 – PPARs	RKP
26	Immunometabolism 3 – Nucleotide Metabolism	RKP
26	REVISION TUTORIAL; ADAPTIVE IMMUNITY (*Venue/Time tbc, 2h*)	JF/MC
27	Immunometabolism 4 – Cancer Cell Metabolism	RKP
27	Immunometabolism 5 – Immune Cell Metabolism	RKP
27	Immunometabolism 6 - Metabolites as Signalling Molecules	LON
27	Immunometabolism 7 – Applied Immunometabolism	LON
28	Reading Week	
29	*Online MCQ*	
30-33		
34	Revision Week	
35	Trinity Week	
36	Assessment	

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 key 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, recently released its 10th Edition which includes chapters on COVID-19 & vaccination. Students will be provided with discount codes for online versions and online/paper. This will be provided prior to commencement of the module.
Further reading will be given out by lecturers during the module.

10. Delivery: This Open module consists of 20 scheduled lectures which will be delivered online through Panopto (pre-recorded). These recordings will be released online in the Blackboard for the module on the Monday of the week they are scheduled for. 2 online Revision Tutorials will be held after the Innate Immunity & Adaptive Immunity Sessions. These will be done in-person, in suitable venues or online through Zoom, using Break-out rooms to divide the large class into smaller groups & PhD student moderators will lead discussion on topics relating to the lecture material identified by the class beforehand. The in-class MCQ will be made available on the Blackboard on week 29 (after Reading Week) & the end of Semester exam will also be online through the VLE

11. Assessment Details:
70% End of year examination, 30% in course assessed.
In course assessment: In-class end of module MCQ exam covering lecture material

12. Module Coordinator Emma Creagh ecreagh@tcd.ie
Executive Officer: Úna Murphy MURPHYU1@tcd.ie

labradoj@tcd.ie

4.8 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)
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
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. all text, including figure legends and tables; but excluding the Title, Index 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 6th, 2023), with the word count verified and included in the submitted version (see appendix n V).

- 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 legend (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 in the text 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 work must be bound (e.g. by ring binding), not stapled or clipped together. Read's print centre on Nassau Street (back of courtyard beside the Kilkenny shop) do binding quickly and cheaply.
- 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)

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).

Penalties:

Penalties 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.

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.

Gold medals 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 www.tcd.ie/academicregistry/exams /student-guide 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 PRIZES AND OTHER AWARDS, see also MISCELLANEOUS AWARDS (Calendar 2022/23)

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.

Appendices

I. Plagiarism

Plagiarism is regarded as a serious offence 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
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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 idea and fail to record its source.
All the above serve only as examples and are not exhaustive.
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 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 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 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 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.

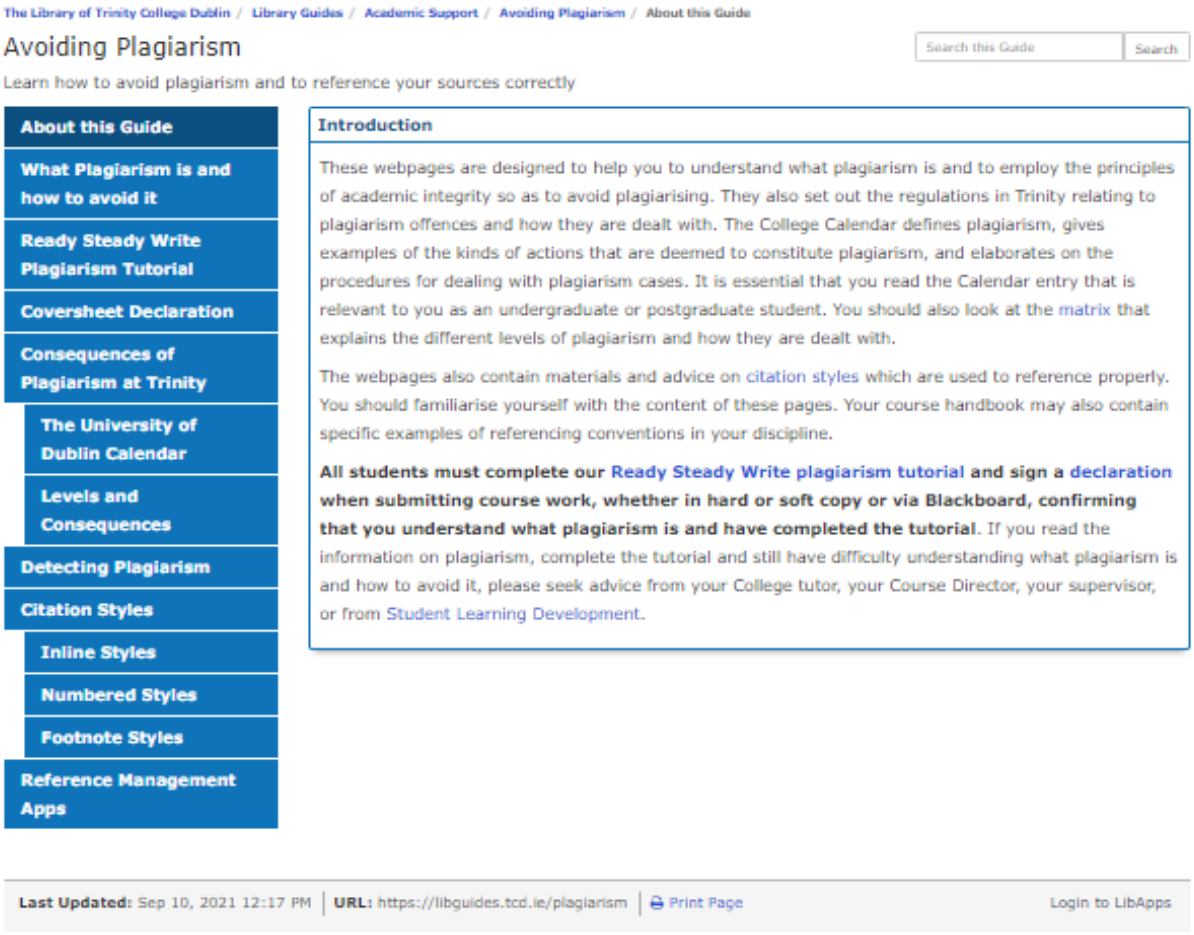
104 Provided that the appropriate procedure has been followed and all parties in §101 above are in agreement with the proposed penalty, the Director of Teaching and Learning (Undergraduate) should in the case of a Level 1 offence, inform the course director and where appropriate the course office. In 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 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.

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



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

(to be bound into your 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 (with the exception of the References section is: [ ]

Signed.....

Dated .....

VI. Important dates and timetable by semester

Module	Module name	type	Semester	week	submission date	Submission time
BIU33150	Biochemistry for Biosciences	MCQ 1 (24%)	Semester 1	8	TBC	TBC
GEU33075	Evolutionary and Population Genetics	MCQs (60%)	Semester 1	10	thu 3rd Nov	10:00-12:00
GEU33007	Molecular Genetics Laboratory	Lab report 1 (50%)	Semester 1	10	Fri 4th Nov	17:00
GEU33075	Evolutionary and Population Genetics	poster submission (0%)	Semester 1	12	Mon 14th Nov	09:00
GEU33075	Evolutionary and Population Genetics	poster presentation (35%)	Semester 1	12	Mon 17th Nov	12:00-15:00
GEU33075	Evolutionary and Population Genetics	Reflective essay (5%)	Semester 1	13	Fri 25th Nov	17:00
BIU33150	Biochemistry for Biosciences	MCQ 2 (16%)	Semester 1	13	TBC	TBC
GEU33007	Molecular Genetics Laboratory	Lab report 2 (50%)	Semester 1	14	Fri 2nd Dec	17:00
GEU33285	Science Structure, Discussion and Presentation for Human Genetics	networks essay 1500 words (15%)	semester 1	14	Mon 28th Nov	17:00
GEU33285	Science Structure, Discussion and Presentation for Human Genetics	written review 4000 words (70%)	semester 1	19	Fri 6th Jan	17:00
GEU33025	Data Handling and Bioinformatics	Bioinformatics Assessment (33.3%)	Semester 2	26	Mon 20th Feb	11:00-13:00
GEU33008	Analytical Genetics Laboratory	MCQ (50%)	Semester 2	27	TBC	TBC
GEU33025	Data Handling and Bioinformatics	Programming Assessment (33.3%)	Semester 2	29	Thu 16th	10:00-13:00
BIU33250	Introduction to Immunology & Immunometabolism	MCQS (30%)	Semester 2	29	TBC	TBC
GEU33035	Genetic Analysis of Nervous Systems	Test (50%)	Semester 2	30	TBC	TBC
GEU33008	Analytical Genetics Laboratory	Lab report (50%)	Semester 2	31	TBC	TBC

BIU33475	Basics of Neurobiology	continuous assessment (Literature Review) (30%)	Semester 2	33	14th April	TBC
GEU33025	Data Handling and Bioinformatics	Report submission (33.3%)	Semester 2	35	Wed 26th April	17:00
GEU33035	Genetic Analysis of Nervous Systems	class participation/essay (50%)	Semester 2	TBC	TBC	TBC

VII. Academic year structure 2022/32

Key Dates:

Orientation Week: Monday 05 September to Friday 9 September 2022
Study/Review Week: Monday 24 October to Friday 28 October 2022
Revision Week Semester 1: Monday 5 December to Friday 9 December 2022
Study/Review Week: Monday 6 March to Friday 10 March 2023
Revision Week Semester 2: Monday 17 April to Friday 21 April 2023
Trinity week: Monday 24 April to Friday 28 April 2023

2022/23 Teaching and Learning Weeks

UG continuing years / PG all years

Teaching Weeks	Academic Calendar Weeks	Week beginning	2022/23 Teaching and Learning Weeks
Michaelmas Teaching Term			
1	3	12-Sep-22	Teaching and Learning
2	4	19-Sep-22	Teaching and Learning
3	5	26-Sep-22	Teaching and Learning
4	6	03-Oct-22	Teaching and Learning
5	7	10-Oct-22	Teaching and Learning
6	8	17-Oct-22	Teaching and Learning
7	9	24-Oct-22	Study/Review
8	10	31-Oct-22	Teaching and Learning (Monday, Public Holiday)
9	11	07-Nov-22	Teaching and Learning
10	12	14-Nov-22	Teaching and Learning
11	13	21-Nov-22	Teaching and Learning
12	14	28-Nov-22	Teaching and Learning
Hilary Teaching Term			
1	22	23-Jan-23	Teaching and Learning
2	23	30-Jan-23	Teaching and Learning
3	24	06-Feb-23	Teaching and Learning (Monday, Public Holiday)
4	25	13-Feb-23	Teaching and Learning
5	26	20-Feb-23	Teaching and Learning
6	27	27-Feb-23	Teaching and Learning
7	28	06-Mar-23	Study/Review
8	29	13-Mar-23	Teaching and Learning (Friday, Public Holiday)
9	30	20-Mar-23	Teaching and Learning
10	31	27-Mar-23	Teaching and Learning
11	32	03-Apr-23	Teaching and Learning (Friday, Good Friday)
12	33	10-Apr-23	Teaching and Learning (Monday, Easter Monday)

Semester 1 (MT): 3-8,10-14
Semester 2 (HT): 22-27,29-33

VIII. 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 Range	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.

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

MyCareer: mycareerconnect.tcd.ie
TCD.Careers.Service
TCDCareers
www.tcd.ie/Careers/students/postgraduate
@TCDCareers
tinyurl.com/LinkedIn-TCD-Connecting

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