



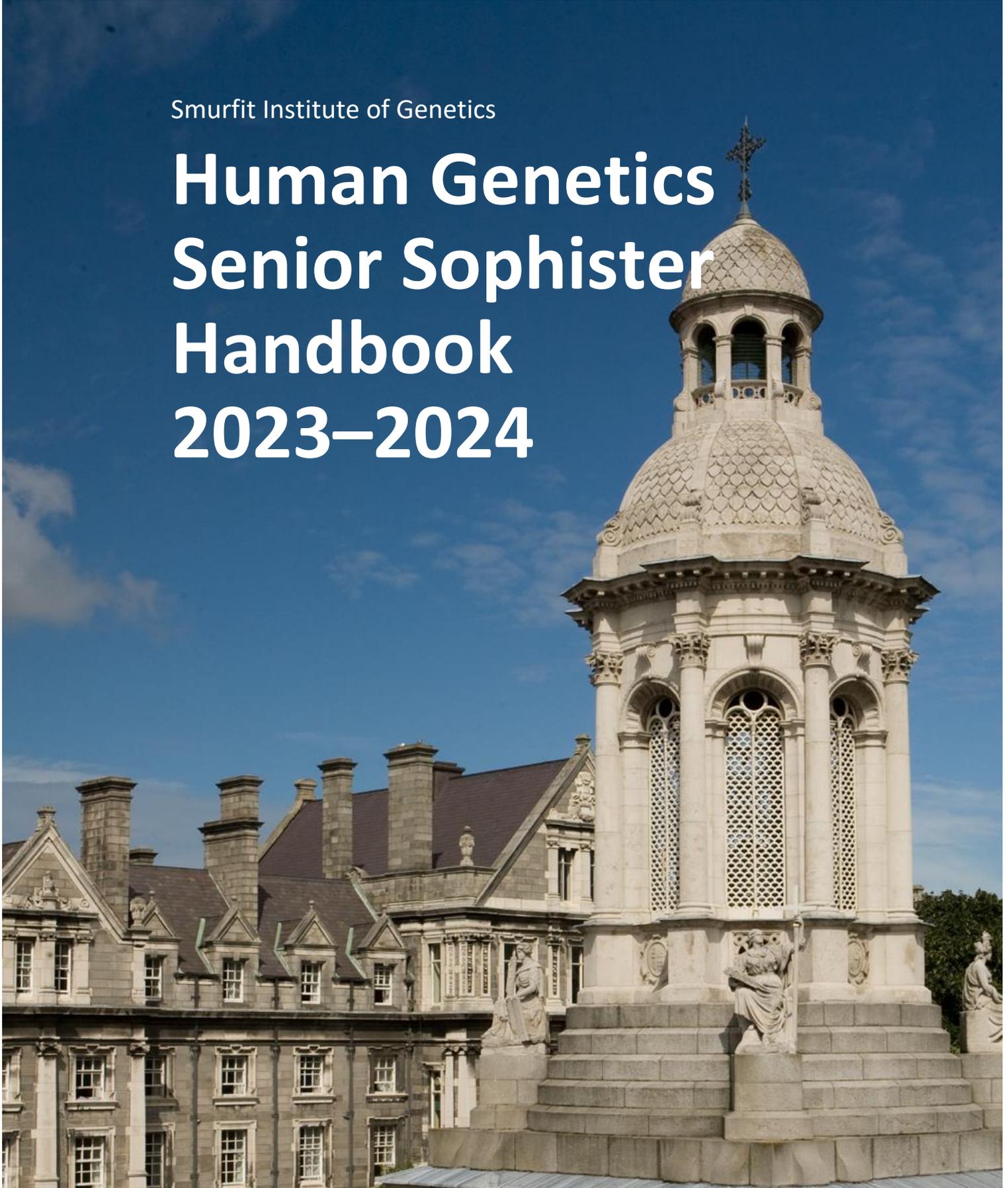
Trinity College Dublin

Coláiste na Tríonóide, Baile Átha Cliath

The University of Dublin

Smurfit Institute of Genetics

Human Genetics Senior Sophister Handbook 2023–2024



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Course administration: Ms. Alicia Vega genetics@tcd.ie

1. Introduction

It is a pleasure for me to welcome you to your Senior Sophister year in Genetics. As you know, our Department is relatively young (established in 1958) considering the age of the university. Nevertheless, in its short history it has been responsible for big discoveries in the field of Genetics. Our graduates have gone on to positions of great influence over the world and we look forward to seeing where this degree will take you when you finish in May 2024.

Our philosophy has a strong focus on the individual and in developing your analytical as well as technical skills. It will be important for you to learn a range of subjects but also to think critically and be able to apply your knowledge to problems. We also believe that communication skills are an essential part of your education and you will develop these skills through a literature review as well as a capstone project. These projects continuously prove to be really rewarding for students. It will also give you an opportunity to work side by side with our amazing scientists (PhD students and postdocs).

While the year ahead will be incredibly busy, please remember to make time for yourself and to continue in your extra-curricular activities, be they sporting or otherwise. Make sure you set realistic goals and aspirations and please reach out to the lecturing staff if you have any issues during the year, we're here to support you. Your success this year is also our success!

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 on the College webpage. However, your personal tutor will be able to guide you should you require any such support.

I wish you the best of luck in your final year and will look forward to seeing you all about the department in the year ahead.

Best wishes,

Prof Matthew Campbell, Head of Genetics.

YOU ARE ADVISED TO READ THE FOLLOWING CAREFULLY AND TO KEEP IT FOR REFERENCE THROUGHOUT YOUR MODERATORSHIP YEAR.

2. Programme Structure/Overview

The Senior Sophister course is divided into:

- 4 Core Modules
- Capstone Project

The Moderatorship Examination includes assessment of the Research Project, the Review, Problems in Genetics, the Papers and Viva of the final examination. The year counts for a total of 60 ECTS credits which are allocated as follows:

| Human Genetics | |
|---|------------------------|
| Semester 1 (S1) | Semester 2 (S2) |
| Core Modules | |
| GEU44208 Medical Genetics in the Era of Precision Medicine (10 credits) | |
| GEU44009 From Individuals to Populations to Species: Development, Behavior, Population Genetics and Evolution (10 credits) | |
| GEU44010 Dealing with Data in Genetic Research (10 credits) | |
| GEU44011 Molecular and Cellular Genetics (10 credits) | |
| Capstone Project | |
| GEU44012 (20 credits) | |

(For your information, when consulting papers from previous years, the exam structure was changed for lecture modules in 2016-17. For 2011-2016 papers 1 and 2 were similar (examining the same modules) and papers 2 and 3 were similar. Up to 2011, Papers 1 and 3 were similar, and Papers 2 and 4 were similar. What was previously paper 6 is now paper 4, having been paper 5 for a few years. Problems were formerly assessed during the summer exams as paper 5, and then as paper 4 up to 2022. Therefore, while the content is largely the same and you should use past exams as a reference, please note that the structure has changed for academic year 2022/2023.

You can find past exam paper following the link

<https://www.tcd.ie/academicregistry/exams/student-guide/>

3. Time Management

Over the course of this year you will have various ongoing assignments in addition to attending lectures and studying the material for the final exams. In particular, the Review and Project contribute significantly to your overall assessment. Generally, an ECTS unit corresponds to 20-25 hours of *overall* commitment by a student. It is very important to try and balance the effort you give to all these commitments. You may also have to adjust to an irregular work schedule as demanded by the experiments you are carrying out in the course of your project.

4. Modules

4.1 GEU44208 Medical Genetics in the Era of Precision Medicine

1. **Module Code:** GEU44208

2. **Module Name** Medical Genetics in the Era of Precision Medicine

3. **Semester taught** Semester 1

4. **Contact Hours** 34 hours + 5 guest lectures

5. **Module Personnel** Prof Jane Farrar, Prof Matthew Campbell

6. **Learning Aims** The study of genomes, and predominantly but not exclusively the human genome, is radically altering health care today and will do so even to a greater extent in the future. The module aims to provide an overview of the burgeoning field of molecular medicine/precision medicine and the genetic information that underpins this field and incorporates basic and applied aspects of medical genetics.

A key focus of the module will be to illuminate how genomic information is currently being utilised in medicine. Topics covered will include current disease diagnostics using genetic methodologies and information, the interpretation of genetic information and provision of information to patients in a clinical setting. The clinical trial process and pharmacogenomics will also be briefly covered.

Genomic information as a driver of novel therapeutic development for a range of disorders will be outlined with powerful examples in the clinic or in preclinical development. The multivalent aspects of genomic medicine including development of therapies for Mendelian and multifactorial diseases will be outlined. Identification of disease targets and development of targeted therapies from gene replacement therapies to gene editing therapies will be reviewed. Ethical debates regarding genetic information will be discussed, as will issues such as somatic versus germline therapies, among others. The student will be provided with a comprehensive overview of this truly powerful and rapidly expanding field.

7. **Module content:** Programme of lectures

| Week | Day & Time | Lecture Topic & Lecturer |
|------|--------------------|---|
| 4 | Mon 12:00-13:00 | MC: Vascular and Nervous system development; A common evolution |
| 4 | Tue 11:00-12:00 | JF: Introduction to medical genetics in the era of precision medicine |
| 5 | Mon 12:00-13:00 | MC: The blood-brain and blood retina barriers |
| 5 | Tue 11:00-12:00 | JF: Molecular tools for development of cell and gene therapies |
| 6 | Mon 12:00-13:00 | MC: The retinal vasculature |
| 6 | Tue 11:00-12:00 | JF: Challenges and successes in gene therapies |
| 7 | Mon 12:00-13:00 | MC: The genetics of age-related macular degeneration (AMD) |
| 7 | Tue 11:00-12:00 | JF: CNS disorders: target identification & therapeutic development |
| 8 | Mon 12:00-13:00 | MC: AMD; Therapies and therapeutic target identification |
| 8 | Tue 11:00-12:00 | JF: CNS disorders: therapeutic development for MND |
| 9 | Review Week | |
| 10 | Tue 12:00-13:00 | JF: Systemic targets for gene therapy, challenges & progress. |
| 10 | Wed 9:00-10:00 | MC: Alzheimer's Disease. |

| | | |
|----|-----------------------------|---|
| 10 | Wed 10:00-11:00 | JF: Haemophilia A & B: Current status of gene therapies |
| 10 | Thu 9:00-10:00 | MC: Alzheimer's Disease: Therapies and therapeutic target identification |
| 10 | Thu 10:00-11:00 | JF: Tutorial |
| 11 | Mon 12:00-13:00 | MC: Tutorial |
| 11 | Tue 12:00-13:00 | JF: Ex vivo and in vivo gene editing therapies |
| 11 | Wed 9:00-10:00 | MC: Neuropsychiatric conditions and the blood brain barrier |
| 11 | Wed 10:00-11:00 | JF: Oligonucleotide therapies: Chemistries, MOAs |
| 11 | Thu 9:00-10:00 | MC: The genetics of 22q11 deletion syndrome |
| 11 | Thu 10:00-11:00 | Guest lecture – TBC |
| 12 | Mon 12:00-13:00 | MC: Autosomal dominant leukoencephalopathies and treatment options |
| 12 | Tue 12:00-13:00 | JF: Oligonucleotide therapies for Duchenne muscular dystrophy & spinal muscular atrophy |
| 12 | Wed 9:00-10:00 | MC: 22q11 deletion syndrome: The lived experience |
| 12 | Wed 10:00-11:00 | JF: Duchenne muscular dystrophy: minigenes as therapies |
| 12 | Thu 9:00-10:00 | Guest lecture – TBC |
| 12 | Thu 10:00-11:00 | Guest lecture – TBC |
| 13 | Mon 12:00-13:00 | MC: Multiple sulfatase syndrome: The lived experience |
| 13 | Tue 12:00-13:00 | JF: Defining targets, designing therapies: current status for ocular disease |
| 13 | Wed 9:00-10:00 | MC: Principles of Human genetics, lecture 1 |
| 13 | Wed 10:00-11:00 | JF: Principles of Human genetics, lecture 2 |
| 13 | Thu 9:00-10:00 | Guest lecture – TBC |
| 13 | Thu 10:00-11:00 | Guest lecture – TBC |
| 14 | Mon 12:00-13:00 | MC: Principles of Human genetics, lecture 3 |
| 14 | Tue 12:00-13:00 | JF: Principles of Human genetics, lecture 4 |
| 14 | Wed 9:00-10:00 | MC: Principles of Human genetics, lecture 5 |
| 14 | Wed 10:00-11:00 | JF: Principles of Human genetics, lecture 6 |
| 14 | Thu 9:00-10:00 | MC: Tutorial |
| 14 | Thu 10:00-11:00 | JF: Tutorial |
| 28 | -----Study/Review Week----- | |
| 36 | Assessment Week | |

NOTE: venue LTEE3

A number of Thursday slots have been reserved for guest lectures – TBC – In total there will 2 or 3 guest lectures but the slot times are still to be confirmed

8. Learning Outcomes:

Students will gain an understanding the aspects of precision medicine including the identification of disease targets and the development of innovative therapies.

9. Recommended Reading List:

10. Assessment Details: Assessment is exclusively by a 3 hours end of year exam.

11. Module Coordinator: Prof Jane Farrar Jane.Farrar@tcd.ie

4.2 GEU44009 From Individuals to Populations to Species: Development, Behavior, Population Genetics and Evolution

1. Module Code: GEU44009

2. Module Name: From Individuals to Populations to Species: Development, Behaviour, Population Genetics and Evolution

4. Contact Hours: 40 lecture hours

5. Module Personnel: Prof Dan Bradley, Dr Kevin Mitchell, Prof Aoife McLysaght, Dr J. Pablo Labrador

6. Learning Aims: This module builds on the knowledge from earlier academic years and encompasses core concepts in genetics to develop a deeper conceptual and specific knowledge and understanding of the interplay of development, heritability and evolutionary processes.

The molecular evolution lectures will consider various aspects of evolution covering large-scale genomic events down to small changes in genes and regulatory sequences. These will be discussed in the context of speciation, adaptation, the evolution of sex and sex chromosomes, the evolution of development (morphological evolution), and fundamental patterns of genetic variation arising through mutation and selection.

The population genetics lectures will explore evolutionary concepts in a more recent timeframe, specifically looking at human population genetics. These lectures will consider human adaptive evolution, the migratory paths of ancient modern humans as illustrated by patterns of genetic diversity, the contributions and legacy of archaic humans, and regional diversity and adaptations in human populations.

The development lectures will focus on the genetics of neural and neuronal specification. There will be special emphasis on the generation of diversity in the nervous system and a focus on the spinal cord and axon guidance.

Finally, this module will consider the genetics of behaviour and explore this in terms of how it is shaped by the interplay of evolution and development. These lectures will consider how organisms are adapting to their environment and how evolution shapes that, and how development realises that. These lectures will encompass fundamental concepts of heritability and association studies and expand into the genetics of complex traits including intelligence, sexuality and personality. The lectures will also consider how all of these concepts can be used to understand the genetics of neurodevelopmental disorders.

7. Module content:

| Week | Day & Time | Lecture Topic & Lecturer |
|-------------------|-----------------|---|
| Semester 1 | | |
| 3 | Tue 9:00-10:00 | Ape and human phylogeny (Bradley) |
| 3 | Tue 10:00-11:00 | " |
| 3 | Tue 14:00-15:00 | Human evolution within Africa (Bradley) |
| 3 | Tue 15:00-16:00 | " |
| 3 | Wed 9:00-10:00 | Whole Genome Duplication (McLysaght) |
| 3 | Wed 10:00-11:00 | Speciation (McLysaght) |
| 4 | Tue 9:00-10:00 | Evolution of duplicated genes (McLysaght) |

| | | |
|-------------------|-----------------------------|--|
| 4 | Tue 10:00-11:00 | " |
| 5 | Tue 9:00-10:00 | Archaic <i>Homo</i> species and their ancient genomes (Huerta-Sanchez) |
| 5 | Tue 10:00-11:00 | " |
| 5 | Wed 9:00-10:00 | De novo gene evolution (McLysaght) |
| 5 | Wed 10:00-11:00 | Adaptive conflict and evolutionary antagonisms (McLysaght) |
| 6 | Tue 9:00-10:00 | Archaeological genomes and the peopling of Europe (Bradley) |
| 6 | Tue 10:00-11:00 | " |
| 6 | Wed 9:00-10:00 | EvoDevo (McLysaght) |
| 6 | Wed 10:00-11:00 | " |
| 6 | Thu 11:00-12:00 | Outlying genome regions: the Y chromosome; recently selected regions (Bradley) |
| 6 | Thu 12:00-13:00 | " |
| 7 | Tue 9:00-10:00 | Genome evolution and mutation (McLysaght) |
| 7 | Tue 10:00-11:00 | Sex chromosome evolution (McLysaght) |
| Semester 2 | | |
| 22 | Wed 9:00-10:00 | Neural induction (Labrador) |
| 22 | Wed 10:00-11:00 | " |
| 22 | Thu 9:00-10:00 | Introduction to heritability (Mitchell) |
| 22 | Thu 10:00-11:00 | " |
| 23 | Wed 9:00-10:00 | Neuroectoderm patterning and neural precursor specification (Labrador) |
| 23 | Wed 12:00-13:00 | " |
| 23 | Thu 9:00-10:00 | Genetics of Intelligence (Mitchell) |
| 23 | Thu 10:00-11:00 | " |
| 24 | Wed 9:00-10:00 | Diversity generation in the nervous system (Labrador) |
| 24 | Wed 10:00-11:00 | " |
| 24 | Thu 9:00-10:00 | Personality (Mitchell) |
| 24 | Thu 10:00-11:00 | " |
| 25 | Wed 9:00-10:00 | Spinal cord as a model for neuronal specification (Labrador) |
| 25 | Wed 10:00-11:00 | " |
| 25 | Thu 9:00-10:00 | Sexuality (Mitchell) |
| 25 | Thu 10:00-11:00 | " |
| 26 | Wed 9:00-10:00 | Guiding axons to their targets (Labrador) |
| 26 | Wed 10:00-11:00 | " |
| 26 | Thu 9:00-10:00 | Neurodevelopmental disorders (Mitchell) |
| 26 | Thu 10:00-11:00 | Energy balance (Mitchell) |
| 27 | Wed 10:00-11:00 | TBC (Labrador) |
| 28 | -----Study/Review Week----- | |
| 36 | Assessment Week | |

NOTE: venue LTEE3

8. Learning Outcomes: Students completing this module will develop a strong sense of the interplay of genetics, evolution and development on the scales of species differences, individual variation and developmental stochasticity. This enriches the conceptual insights and cultivates a deeper understanding of how genetics forms a common language for understanding all aspects of biological diversity.

9. Recommended Reading List: Primary literature (journal articles) will be referenced in the lecture notes.

10. Assessment Details: Assessment is exclusively by a 3h end-of-year exam. The exam will be structured as 4 either/or questions (one from each lecturer) of which the student must answer 3.

11. Module Coordinator: Prof Aoife McLysaght

Email: aoife.mclysaght@tcd.ie

4.3 GEU44010 Dealing with Data in Genetic Research

1. **Module Code** **GEU44010**

2. **Module Name** **Dealing with data in genetic research**

4. **Contact Hours** 16 + 2 introduction tutorial

5. **Module Personnel** Dr Russell McLaughlin, Dr Lara Cassidy, Prof Dan Bradley, Dr J. Pablo Labrador

6. **Learning Aims** This module will explore data science in genetics as it stands in the 21st century, covering multiple layers of abstraction from the fundamentals of computer science to high-level statistical models used to relate data to biology. Through a taught component, students will learn how genetic data are represented in a computer, how the problem of data manipulation and processing is optimised and structured into algorithms, how these algorithms are chained into analytical pipelines and the form taken by the outputs, from file format specifications to model-based representations of error and uncertainty.

Students will gain applied experience at each of these levels of abstraction and will become familiar with some of the most commonly-used academic software in genetics and genomics. This taught component will be evaluated through continual assessment, supplemented with an examination presenting analytical problems in genetics drawn from the diversity of subject areas taught in their undergraduate programme. Students will also gain experience in synthesis and meta-analysis of data across studies through the submission of a literature review. Upon completion of the module, students will understand the relevance of data science in genetics and will be equipped with a highly-transferrable skillset that enables them to structure a problem algorithmically, manipulate commercial and academic software for their own purposes, and relate the outputs of their approach back to the biological question.

7. **Module content:** Programme of lectures/practicals –

| Week | Day & Time | Lecture Topic |
|------|------------------------------------|--|
| 3 | Thu 9:00-11:00 | How to write a literature review on a genetics/human genetic topic (Labrador & Cassidy) |
| 4 | Thu 9:00-11:00 | From data to discovery I: a critical look at scientific literature (McLaughlin) |
| 5 | Thu 9:00-11:00 | The data deluge: how to handle data using computers (McLaughlin) |
| 6 | Thu 9:00-11:00 | Thinking algorithmically: sequence alignment (McLaughlin) |
| 7 | Thu 9:00-11:00 | Back to biology: annotation and variants (Cassidy) |
| 9 | <i>-----Study/Review Week-----</i> | |
| 10 | Thu 11:00-13:00 | Starting into statistics: analytical descriptors of genotype data (Cassidy) |
| 11 | Thu 11:00-13:00 | Data within data: extracting more from what you have (Cassidy) TBC |
| 12 | Thu 11:00-13:00 | Where statistics meets art: data visualization and hypothesis testing (Bradley) |
| 13 | Thu 11:00-13:00 | From data to discovery II: working together, sharing data and resources (McLaughlin & Cassidy) |
| 15 | <i>-----Study/Review Week-----</i> | |
| 16 | <i>Assessment Week</i> | |

NOTE: venue LTEE3

Content Description

From data to discovery I: a critical look at scientific literature

In this lecture we will introduce the module as a journey from raw data -- numbers, text and ones and zeros -- to discoveries presented in the scientific literature. The module will take students through many of the levels of abstraction required to understand this journey, as well as offering some hands-on practical application of the concepts presented in the lectures. In this lecture, we will begin by presenting some published data and critically evaluating its presentation and merit in the literature, and asking the question, "How did the authors get here?" This forms the basis for the lectures that will follow.

The data deluge: how to handle data using computers

We will now begin to think deeply about the nature of data -- how it is generated in genetics, how it is represented and how it is stored. This immediately brings us into the domain of computer science, so this lecture will describe the fundamentals of how computers are designed and how they work, and, using this knowledge, we will learn about how computers are harnessed to do the bidding of data scientists. We will discuss how data science is applied to answer questions in biology using statistical approaches applied to genetic data.

Thinking algorithmically: sequence alignment

Fundamental to most applications of data science is the idea that a problem can be solved with an algorithmic approach using structured data. As most genetic data begins with DNA sequence, we will look at this concept using an example of a famous and centrally important algorithm: Smith-Waterman alignment. This algorithm provides an optimal solution to the alignment of two DNA sequences (or any character sequence), given a set of alignment parameters. We will explore in-depth how the algorithm works and various implementations of the approach. The lecture will also introduce more modern approaches that depend on data compression to align the many billions of sequence reads that come from a typical next-generation sequencing experiment to the three gigabase human reference genome.

Back to biology: annotation and variants

Now that we have aligned DNA sequences, this lecture will begin to explore how we can use this structured data to return to the biological questions we set out to answer. How do we extract biological information from DNA sequences? We will look at methods that can be adopted to annotate sequences as genic or regulatory, as well as approaches for discovering and genotyping variation present in an individual's genome. Systems for expressing uncertainty are an important component of these methods; these will be discussed along with systematic ways to represent and interrogate resulting data using file format specifications and open-source software.

Starting into statistics: analytical descriptors of genotype data

With a set of variant calls for the genome of an individual or group of individuals, we can answer a lot of biological questions -- from demography to disease. But an intimate understanding of the dataset in hand is crucial to avoid snags that can introduce bias or lead to entirely false conclusions. In this lecture we will examine ways that genotype data can be interrogated to better understand it, including statistical approaches to summarize genotypes and variants in a meaningful way. We will also contrast summary measures that operate on a regional (locus-specific) or global (genome-wide) level to reveal patterns and structure within our data.

Data within data: extracting more from what you have

Often raw data needs some level of processing before it can be usefully analyzed to answer biological questions. But even processed data can still contain useful information embedded within it which can be extracted using specialist approaches. In this lecture we will look at methods that can be used to reveal and harness useful structure within genotype data to answer deeper and richer biological questions.

Where statistics meets art: data visualization and hypothesis testing

Data visualization is one of the most important components of completing a scientific study. This begins with early visualization of raw or unanalyzed data to conduct “sanity checks” and ensure data are clean and representative of the study. From here, the task of the analyst is to decide on an approach to statistically explore their data and choose appropriate visualization strategies. This lecture will provide a refresher in many of the concepts that are required for this process and describe the journey through visualization and hypothesis testing to arrive at a scientific conclusion.

From data to discovery II: working together, sharing data and resources

Now that we have analyzed our data, conducted statistical tests and generated figures that support our conclusions, we will take a look back over the process and discuss dos and don'ts of working in large and data-rich collaborative environments, principles of data and resource sharing, and concepts associated with developing analytical pipelines and procedures for reproducibility and open science.

Problems in Genetics

Problems in Genetics incorporate genetics knowledge alongside a genetical approach to thinking about biological questions. This is where you put your learning into action. The type of problems you may be faced with is broad and diverse and may include experimental design, interpretation and analysis of experimental results, and quantitative analysis of genetic data. By its very nature, this is not something where you can learn the answers, but you can train yourself to better recognise a productive approach (this is where practice on past questions is very useful).

Problems in Genetics is examined in the summer exams, and past exam questions will be a valuable inspiration for study. Assessment of this section will take place during assessment week. The assessment will take place over 3.5 hours and you will need to answer all questions (typically 12-15 problems). The problems will be of a varied nature and degree of difficulty and may be based on material or concepts from both the Fresher and Sophister years.

These will test your ability to explain data, handle evidence and solve problems. You may bring notes, photocopies, the 'Introduction to Genetic Analysis' by Griffiths et al. textbook and a calculator to this exam. For practice, you may find useful problem sets in many genetics textbooks. Data will often be taken from published papers.

Review

You should arrange via e-mail to meet with your review supervisor during the first week of term to discuss the review topic and to seek their advice regarding the published literature.

Review Presentation

Your literature review *must not* exceed **4,000 words** (i.e. all text, including Figure legends and Tables; but excluding 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 on time (dates listed *under 7. deadlines and dates to remember*), with the **word count** verified and included in the submitted version.

The objective is to bring the reader up-to-date on the subject under review. You should

therefore consult major texts to understand the historical aspects of the subject, and then the most recent major review articles to discover the state of the field at the time those reviews were written. You should critically review the major papers in the field during the previous ~5 years and try to define the interesting questions that remain to be answered. The emphasis of your review should be on the most recent work in the field, *i.e.*, approximately the past 5 years.

Each review topic is assigned by a member of staff, your review supervisor. You should meet with them soon after your topic is assigned, and they may give you some advice on how to start. Your supervisor will be willing to discuss the outline of your review and to advise you on its structure, but they will not read or edit draft versions of your review.

Other Review requirements:

WARNING: Your review must not contain material produced via a process of “copying” or “cutting and pasting” of text from any source: this is plagiarism. Also, the “paraphrasing” of text (*i.e.* changing few words in a sentence or paragraph) constitutes plagiarism. It can be readily detected by computer-based searches of your submitted work. For more information on plagiarism please see the appendix.

Plagiarism is a serious offence. Any submitted work (*e.g.* your Review) that contains plagiarized content will be marked punitively and may even be awarded a mark of 0%.

The review must be your own work; you may not use generative AI (ChatGPT or similar) to write the review. Note that your supervisor will be able to gauge your level of understanding of the subject matter in your meetings with them, and this may be taken into account in their assessment of the final review.

1. The title of your work and your name should be on the front cover.
2. All pages must be numbered and the work must be bound.
3. The work should be divided into Abstract; Introduction; Main text to be organized in subsections with headings, according to topic; Conclusion(s); References.
4. If figures and tables are included, they must be numbered. If they are not original, then the source must be cited.
5. Each Figure and Table must be accompanied by an explanatory legend (the text of all legends must be **included** in the overall document **word count**). If you have included a figure or table that you did not draw yourself, you must write your own legend and cite the source in the legend, *e.g.* “figure from (Jones et al 2018)”.
6. References must be presented in the following way (a reference system based on numbers, rather than author names, must not be used). You can use software such as EndNote or Mendeley to help with reference formatting.

In the text of the review: When citing a single author: (Behan, 2006); two authors: (Watson and Crick, 1953); three or more authors: (Lander *et al.*, 2001). Do not include author initials in the in-text citations.

In the References section (Bibliography), *each reference* should contain the full list of author names with initials, the full article title, and the journal details, using the ‘minimum punctuation’ style shown in the following example. If there are more than 5 authors, you may list the first one followed by “*et al.*”.

Watson JD, Crick FHC (1953) Molecular structure of nucleic acids; a structure for deoxyribose nucleic acid. *Nature* 171: 737-738

7. References to websites should not be used as a substitute for the primary published literature in the field under review and therefore should be cited only in exceptional cases. If you want to refer to a website, give the address in the text, not in the references list.

8. You must include a signed statement concerning **Plagiarism**.
9. You must also include a signed statement giving the overall document **word count**.
10. **Submission is through Blackboard** using the Turnitin system to screen for possible plagiarism. The results will be made available to you.

Review Assessment

The review will be assessed taking into account the following criteria:

- Difficulty of topic
- Scientific content
- Clarity of thinking / Comprehension of the subject
- Ability to write scientifically
- Structure of the review
- Awareness of the recent literature on this subject
- Presentation

8. Learning Outcomes:

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

- Critically evaluate the presentation and visualisation of data in scientific papers
- Describe the journey from data to discovery in genetics
- Describe the nature of the various types of data used in genetics
- Identify and explain the function of the various components of a modern computer
- Describe different DNA sequencing technologies in technical detail
- Implement the Smith-Waterman alignment algorithm and describe other alignment/assembly algorithms
- Dissect the components of modern genomic file format standards (SAM, VCF)
- Explain various methods for extracting biological information from aligned sequences
- Analyse large genotype datasets using a suite of statistical methodologies
- Use advanced algorithms to turn genotype data into a richer set of haplotypes
- Describe strategies for data visualisation in genetics
- Chain algorithms into pipelines and build data analysis scripts
- Share data and code via online repositories such as Github
- Write a literature review

9. Recommended Reading List: There is no specific reading list for this module.

10. Assessment Details:

- **Continual assessment assignments** (2 credits total): provided weeks 4, 6, 10 and 12; due weeks 4, 8, 12 and 14
- **Problems in genetics examination** (4 credits total). 3.5h end-of-year exam
- **Literature review** (4 credits total)

11. Module Coordinator

Dr Russell McLaughlin

Email: mclaugr@tcd.ie

4.4 GEU44011 Molecular and Cellular Genetics

1. **Module Code** GEU44011
2. **Module Name** Molecular and Cellular Genetics
3. **Semester taught** Semester 1 and Semester 2
4. **Contact Hours** 30
5. **Module Personnel** Prof Seamus Martin, Prof Adrian Bracken, Prof Mani Ramaswami

6. Learning Aims This module will deepen the student's understanding of a range of core concepts in molecular and cellular genetics, including: chromatin organization and regulation, the non-coding genome, epigenetic control of gene expression (Chromatin Biology and Epigenetics). The module will also overview protein structure, post-translational modifications of proteins, the diverse impacts of mutation on protein function, as well as cellular organization and organelle function (General concepts in molecular biology). We will explore cell signaling and the activation of gene expression programmes; genetic conservation and divergence of a key cellular process (programmed cell death) from lower to higher organisms will also be covered in detail (Genetic control of Programmed Cell Death). This module will also examine genetic mutation and disease by looking at cancer and how this arises, spanning from the discovery of cancer-promoting genes (oncogenes and tumor suppressor genes) and how the latter drive tumor formation, as well as how mutations affecting genes that play a role in the regulation of the epigenome contribute to the development of cancer. We will also explore cancer therapy via targeting specific cancer-associated mutations (precision oncology), as well as recent developments in targeting chromatin-remodeling proteins in cancer (Cancer Genetics). Overall, this module on Molecular and Cellular Genetics will cover a broad sweep of molecular biology, illustrating key principles of how genes are expressed, how they exert their functions, how the protein products of genes are folded, post-translationally modified and degraded, and how gene mutations lead to the subversion of these functions to provoke disease.

7. Module content: Programme of lectures

| Week | Day & Time | Lecture Topic & Lecturer |
|-------------------|------------------------------------|---|
| Semester 1 | | |
| 7 | Fri 11:00-12:00 | Recap of general concepts in cell and molecular biology (Prof. Seamus Martin) Lecture 1. Protein structure, diversity, function, post-translational modifications and effects of mutation. Cellular organelles and their primary functions |
| 7 | Fri 12:00-13:00 | Recap of general concepts in cell and molecular biology (Prof. Seamus Martin) Lecture 2. Cell signaling and the activation of gene expression programmes, with examples from signals that promote cell division, cell differentiation, cellular activation and secretion (in inflammation) and cell death. |
| 8 | Fri 11:00-12:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 1 . Chaperone systems (small hsps, hsp40, hsp70 and hsp60/chaperonins) involved in protein translation, translocation (ER and mitochondria); stress management and signaling (hsp90) |
| 8 | Fri 12:00-13:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 2. Chaperone systems (small hsps, hsp40, hsp70 and hsp60/chaperonins) involved in protein translation, translocation (ER and mitochondria); stress management and signaling (hsp90) |
| 9 | -----Study/Review Week----- | |
| 10 | Tue 9:00-10:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 3. Proteosomal; lysosomal and autophagy systems |

| | | |
|------------|-----------------|---|
| 10 | Tue 10:00-11:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 4. Proteosomal; lysosomal and autophagy systems |
| 10 | Fri 11:00-12:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 5. RNA chaperone systems. How they were discovered and mechanisms by which they work with some examples. |
| 10 | Fri 12:00-13:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 6. RNA chaperone systems. How they were discovered and mechanisms by which they work with some examples. |
| 11 | Tue 9:00-10:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 7. Biological condensates (RNP granules, which are modulated by both protein and RNA chaperones) |
| 11 | Tue 10:00-11:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 8. Prions/amyloids in mammals and yeast (which also are modulated by chaperones and cell stress signaling pathways). |
| 11 | Fri 11:00-12:00 | Proteostasis (protein handling) (Prof. Mani Ramaswami) Lecture 9. Prions/amyloids in mammals and yeast (which also are modulated by chaperones and cell stress signaling pathways). |
| 11 | Fri 12:00-13:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 1. Introduction & the Discovery of Oncogenes |
| 12 | Tue 9:00-10:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 2. The Discovery Tumour Suppressor Genes |
| 12 | Tue 10:00-11:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 3. Cancer Pathways and Cancer Evolution |
| 12 | Fri 11:00-12:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 4. Chromatin regulator genes are mutated in cancer and the relevance of DNA methylation. |
| 12 | Fri 12:00-13:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 5. Precision Oncology: Targeting Oncogenes, Cancer Pathways, and non-genetic mechanisms |
| 13 | Tue 9:00-10:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 6. The EZH2 Polycomb gene in cancer and newly approved EZH2 inhibitor therapy. Oncohistones |
| 13 | Tue 10:00-11:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 7. Treating cancers with loss of SWI/SNF genes – a powerful lesson from Drosophila genetics. |
| 13 | Fri 11:00-12:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 8. Chromatin regulator genes in leukaemia and new therapies. |
| 13 | Fri 12:00-13:00 | Cancer Genetics (Prof. Adrian Bracken) Lecture 9. The non-coding genome in cancer |
| Semester 2 | | |
| 22 | Mon 11:00-12:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 1: Why cell death control is essential to multicellularity Cell death control: necrosis versus apoptosis, pathology and disease implications. |
| 22 | Mon 12:00-13:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 2. Recognition and removal of apoptotic versus necrotic cells from tissue. Cell death as a driver of inflammation: DAMPs & IL-1 family members. |
| 22 | Fri 9:00-10:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 3: Identification of genes involved in programmed cell death Genetic screens in C. elegans. Genetics screens in Drosophila |
| 22 | Fri 10:00-11:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 4: Genetics of mammalian PCD Molecular conservation of PCD control from nematodes to humans |
| 23 | Mon 11:00-12:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 5: Caspases as key players, their role and function. |
| 23 | Mon 12:00-13:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) |

| | | |
|----|-----------------------------|---|
| | | Lecture 6: Routes to caspase activation: intrinsic, extrinsic and CTL attack |
| 23 | Fri 9:00-10:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 7: Bcl-2 family genes and the intrinsic pathway to cell death in mammals |
| 23 | Fri 10:00-11:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 8: The CTL pathway to PCD in mammals. Death receptors and the extrinsic pathway. |
| 24 | Fri 9:00-10:00 | Genetics of Programmed Cell Death (Prof. Seamus Martin) Lecture 9: Cell Death and Disease |
| 24 | Fri 10:00-11:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 1. Revision of Histone and DNA Modifications |
| 25 | Mon 11:00-12:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 2. Chromatin regulators in development and human developmental disorders |
| 25 | Mon 12:00-13:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 3. DNA methylation in development and disease |
| 25 | Fri 9:00-10:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 4. Polycomb group proteins in development and cellular identity |
| 25 | Fri 10:00-11:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 5. Genome organization in development and disease |
| 26 | Fri 9:00-10:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 6. Long non-coding RNAs |
| 26 | Fri 10:00-11:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 7. X-chromosome inactivation and genomic imprinting |
| 26 | Fri 14:00-15:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 8. Transgenerational epigenetic inheritance. |
| 26 | Fri 15:00-16:00 | Chromatin biology and Epigenetics (Prof. Adrian Bracken) Lecture 9. The non-coding genome in disease. |
| 28 | -----Study/Review Week----- | |
| 36 | Assessment Week | |

NOTE: Venue LTEE3

8. Learning Outcomes: Upon successful completion of this module, students will have a deeper understanding of the mechanisms regulating protein as well as RNA folding via dedicated chaperones and protein degradation via the proteasome or autophagy. They will also learn about diseases that can result from the accumulation of insoluble protein/RNA aggregates (biological condensates). They will have acquired advanced knowledge concerning how gene expression can be regulated through modification of chromatin (i.e. epigenetic control), by addition of methyl and other groups to histone proteins as well as DNA, and how epigenetic control influences cell fate and development. Students will also acquire cutting-edge knowledge concerning how cell death is regulated in a programmed manner, how this influences many aspects of immunity, and how dysregulated cell death can lead to cancer and other diseases. They will be able to describe experimental approaches that have been used to identify genes that regulate cell death in *C. elegans*, *Drosophila melanogaster* and in mammals, as well as approaches that are used to explore gene therapy *in vitro* and *in vivo*. They will have acquired knowledge concerning how oncogenes and tumor suppressor genes were discovered, the cellular signaling pathways these genes regulate, how such oncogenes are targeted therapeutically in 'precision oncology', and how epigenetic regulators can play a role in the development of cancer.

9. Recommended Reading List: Specific papers from the primary scientific literature will be cited by individual lecturers during their course and students are encouraged to source these papers themselves.

10. Assessment Details: Assessment is exclusively by a 3 hours end of year exam.

11. Module Coordinator : Prof. Seamus Martin Email: martinsj@tcd.ie

4.5 GEU44012 Capstone Project

For those of you considering a further career in research, your Project will be quite an important first step. It is a chance for you to develop your skills in the lab and demonstrate your aptitude for research to your supervisor, who will likely be an important source of letters of reference for you in your further career.

Normally, you are expected to start your research project shortly after Study Week in Semester 1 (dates listed *under 7. deadlines and dates to remember*). As usual, some projects will be entirely computational/analytical. You should contact your supervisor by e-mail at the start of term and arrange to meet with them to discuss the project and background reading material. Thus, prior to arriving in the lab, you should have already become familiar with the relevant background literature on the research topic; material that should prove useful when writing the Introduction section of your report.

You must finish all experimental work and computer analysis on the date specified (dates listed *under 7. deadlines and dates to remember*), and submit the write-up on or before the deadline.

Supervision: A member of the lecturing staff will be the supervisor of your project. In many cases this member of staff will assign a postdoctoral fellow or PhD student in their lab to assist you on a day-to-day basis. However, the member of staff is your primary supervisor. They are responsible for the scientific direction of your project, and it is important that you discuss the progress of your project with them regularly.

Your project will be marked by your supervisor and a second member of the academic staff.

Time commitment: Every project is unique and the time commitment can vary. As students, it is ultimately up to you to decide how much time to spend on your project, but it is wise to consider this in the context of the ECTS weighting and the other demands on your time. The project is worth 20ECTS. A rough rule-of-thumb is that you should expect about 20-25 hours of effort by the student per ECTS. So that works out as 300-375 hours for your project. Note, this includes all your reading, writing-up, and actual lab work.

If for argument's sake we assume 300 hours overall, and then presume that 1/3 of the time is spent on the write up, then that leaves 200 hours. Over the course of 10 weeks that equates to 20 hours per week working on the project: primarily lab work, analysis, and literature research.

Research seminar: You must give a 10-minute seminar on your research project to a group including the members of your lab and at least one other lab. **This will count for 5% of your project grade.**

The intention is that you produce 8-10 slides, but not more than this. This is not intended as a comprehensive run-through of everything you have done in the lab (it is possible that you will do a longer presentation with your supervisor as a normal part of being in the lab). The purpose of this is to get you thinking about the overall picture of your project without getting lost in the nitty gritty.

For your presentation you should try to address the following points: (1) What is my question and why is it interesting?; (2) What have others done?; (3) What have I done?; (4) What does it mean?

You should email a copy of your presentation to your supervisor and cc genetics@tcd.ie (Alicia) at any time during week 26. This will be made available to the external examiners as part of their evaluation of your project.

Drafts: You should discuss the overall structure of your project report with your supervisor, before you start writing it in detail, and certainly at least 1 month before the submission deadline. Your supervisor will be willing to discuss the outline of your report and to advise you on its structure. They will also be willing to discuss the details of specific Figures or Tables that you want to include in the Results. However, your supervisor will not read or edit draft versions of your report.

Project report presentation: Your Project Report must not exceed **6,000 words** (*i.e.* all text excluding the Figure legends, Tables 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 on time, with the word count verified and included in the submitted version.

The report should be written following the structure of a scientific paper. It should be composed of an Abstract (250 words maximum), Introduction, a section on Materials and Methods, a section on Results, and a Discussion. It should have a Reference list with full references (use the referencing system and style specified above for the Review). It must be typed, paginated and bound. In summary a competent scientist should be able to repeat your experiments or your analysis having read your report. Your report will be marked down if it is not presented well.

In summary:

WARNING: Plagiarism is a very serious offence. Any submitted work (e.g. your Project Report) that contains plagiarized content will be marked punitively and may even be awarded a mark of 0%.

1. The title of your work and your name should be on the front cover.
2. The following page should be a Table of Contents.
3. All pages must be numbered.
4. The work should be subdivided into the following sections:
Abstract; Introduction; Materials and Methods; Results; Discussion; References
[Results and Discussion may be combined where appropriate]
5. Figures and tables must be numbered.
6. Each figure and table must be accompanied by an explanatory legend.
7. References **must** be presented using the same style indicated above for your Review.
8. **Submission is through Blackboard** using the Turnitin system to screen for possible plagiarism. The results will be made available to you.
9. You must include a signed statement concerning **Plagiarism** and **word count**. (Appendix IV)

You should write your project report with the non-expert geneticist in mind, *i.e.*, don't assume intimate knowledge of either the general field or the specifics of your project on the part of the reader. **Remember that your report won't just be read by your project supervisor, but also by a second member of staff and by the external examiner** – and they will not know anything about the project except what you tell them in your report. They won't know what you were trying to do, unless your report explains it.

In writing up your project you must use the scientific literature as your model.

The Abstract is very important because it should summarize (in not more than 250 words) the rationale for doing the experiments, what your main results were, and what you conclude from them. Your Introduction (which should not exceed 3,000 words), should introduce the research area, but it should also introduce your specific project. What was the aim of the project? What questions were you trying to answer? Were you testing an hypothesis? Why was your particular experimental

strategy chosen over alternative ways of answering the same question? The Introduction should end with a formal declaration of the specific aim(s) of the research that was undertaken.

Your Materials and Methods should resemble those of a journal article, with perhaps a little more detail. You should not, however, devote 10 pages with explicit details of every solution you made up; reference to standard manuals will suffice.

Your Results section should be written as in a scientific paper, describing the rationale and design of experiments as you go along and not merely presenting data. **The Results section should not just be a collection of Figures or Tables with no text.** It should talk the reader through the experiments you did, why you did them, and what these experiments show. Figures and Tables should be referred to in the text.

The Discussion section is where you discuss what your results mean, and how they fit into the field. Do they support previous work? Contradict it? Did you answer the questions you set out in the Introduction?

In your writing you should try to display yourself as a scientist. Show your knowledge of the field and the place of your project in the field. Describe the design of the project showing how you expected it to produce useful results. If there is an hypothesis be sure to state it clearly. If there are puzzling conflicts in the published literature that you set out to resolve, make sure you explain these. Describe your experimental results adequately and clearly. Do not include trivial data. Accuracy, clarity and orderliness are essential. Interpret and explain your data. Give your own ideas. Make your own judgments. Be thoughtful, critical, original and constructive. Avoid pedantry. None of these hints is easy to realize - one good idea is worth a thousand pages. Make sure you emphasize what you believe to be the most important discoveries and ideas.

Assessment of the Project: The project will be assessed taking the following criteria into account:

- Difficulty of project
- Understanding of literature/project
- Clarity of thinking
- Ability to design experiments
- Ability to analyse and discuss experiments
- Commitment, effort and behaviour in the laboratory
- Ability to work independently
- Presentation and content of write-up

It is important to emphasize that marks are not allocated solely on the basis of the experimental results obtained – *i.e.* there is NOT a direct relationship between the quantity of results obtained and marks awarded. Instead it must be evident, during your stay in the laboratory and in the write-up of your project, that you have read the literature, formulated a hypothesis, designed appropriate experiments to test this hypothesis (to include all appropriate controls), written all experimental details in you laboratory notebook, interpreted the results and evaluated them to decide if they are consistent with your hypothesis and that you were capable of bringing a “problem-solving” approach to bear on difficulties encountered with experiments. The assessment will include an evaluation of how well you performed ALL these tasks.

*****Please note: we won't be able to disclose any marks in advance of main exams because all marks are subject to approval by the External Examiner and cannot be made available in advance of the final examiners' meeting. The same rule applies for any deferred exams***.**

5. Safety

You must understand that safety is an issue of paramount importance in the department – we take it very seriously – you are REQUIRED to take it seriously also.

You should already have a copy of the Science Faculty Safety Manual and a statement on safety issues within the department is appended to this document (Appendix V). You are required to read these documents before you start laboratory work. The department will give you a short course on aspects of general safety, safety in handling biological, chemical and radioactive materials and on the extent to which you as undergraduates can handle such materials. You must act at all times in a manner that protects your own safety and the safety of all others in the department. You must **NEVER do experiments ALONE** in a laboratory! The laboratory PI is responsible for all aspects of safety within the laboratory. If you are in any doubt about equipment usage or procedures, please consult a more senior person or one of the technicians. Prof. Tony Kavanagh is the departmental safety officer. Prof. Dan Bradley is the GMO safety officer. Ms. Brenda Campbell is the Fire Safety officer. You must consult them on matters of safety and report any incident immediately.

Obviously be guided by your supervisor, the technical staff and experienced researchers. Take care of the equipment and only use it if you know how! Laboratories are by their nature potentially dangerous, so please act thoughtfully and defensively. Please remember you are responsible not only for your own safety but the safety of other i.e. other researchers, our technical staff and the cleaning staff who keep the Smurfit Institute so well – do not endanger others by your actions.

As a matter of general safety, it is important that you do not wear laboratory gloves when touching door handles, bannisters etc.. If it is necessary for you to go between two rooms wearing gloves, then either keep one hand ungloved, or ask someone to come with you to open the doors.

6. Reading the literature

There is a huge literature in genetics. You will be given references to important recent papers in your lectures, which are an authoritative selection. You are expected to read as many of these as possible – if you have problems with them talk about them with your lecturers and you will also find that the research fellows and graduate students will be helpful. Reading and talking about original research papers is an important way of learning how to do science. You should also read reviews, for example the papers in *Nature Reviews Genetics*. Make a point of looking at *Nature* and *Science* each week.

You should aim to be familiar with any major discoveries in genetics reported in the literature during the year (which we may not have had the chance to include in our lectures). Examiners are often impressed by students who are familiar with the recent literature on a topic and incorporate references to it into exam answers, provided that it is relevant.

7. Deadlines and dates to remember

Please note all dates are provisional and may be subject to change.

| | | |
|-------------------------------------|-------------------|---|
| <u>Review submission deadline:</u> | Semester 1 | Tuesday 31st of October 2023 |
| <u>Project lab work to start:</u> | Semester 1 | Tuesday 31st of October 2023 |
| <u>Project lab work to end:</u> | Semester 2 | Friday 16th February 2024 |
| <u>Project research seminar</u> | Semester 2 | <u>Between</u> 19th Feb-1st March 2024 |
| <u>Project submission deadline:</u> | Semester 2 | Monday 4th March 2024 |

- **All work should be submitted to Blackboard by the dates above**
- **Work submitted LATE WILL BE PENALISED by a 5% reduction in mark per day, or part thereof, that the assignment is late.**

Provisional dates for Moderatorship exams 2024

| | | |
|----------|----------------------------|-------------|
| GEU44010 | Mon 29 th April | 10.00-13.30 |
| GEU44208 | Tue 30 th April | 10.00-13.00 |
| GEU44009 | Thurs 2 nd May | 10.00-13.00 |
| GEU44011 | Fri 3 rd May | 10.00-13.00 |

| | |
|----------------------------|------------------------------------|
| Reception to meet externs: | Wed 15 th May (evening) |
| Vivas | Thu 16 th May |
| Results posted | Fri 17 th May |

8. External examiner

New External Examiner for the Genetics degree will be appointed.

9. Your career ahead

You should start thinking now about what you want to do after Moderatorship. There are many openings for geneticists. If you are aiming to do postgraduate research (MSc or PhD), you should aim for a 1st or a II.1 to have the best choice of a place to study, and to qualify for certain scholarships (e.g. Irish Research Council scholarships). Students with II.2s have also been accepted in recent years for research degrees here and abroad. If you are planning to apply for postgraduate courses in the USA you must prepare for and sit the GRE (Graduate Record Examination) during the Michaelmas term, and you may also need to submit a Visa application several months in advance of travel.

A TCD's Careers Office staff will talk to the class on a date to be arranged. Information on the careers advisory service is provided under Appendix VI of this booklet.

If you require written references: you should obtain these from your Research Project supervisor in the first instance. If you worked in a laboratory during the summer, the head of that lab would be a good second referee. If an additional reference is required, ask your Review supervisor.

Your referees will also be happy to give you **advice on how to present your CV** and how to write cover letters.

And finally ...

You will have a challenging year ahead. Others have found it really worthwhile. All the staff wish you the best of luck.

Prof Matthew Campbell

Head of Department

APPENDICES

I. *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 |

II. 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.

III. *Declaration to include on review*

UNIVERSITY OF DUBLIN

TRINITY COLLEGE

SCHOOL OF GENETICS AND MICROBIOLOGY

SMURFIT INSTITUTE OF GENETICS

DECLARATION FOR REVIEW

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

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

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

Signed.....

Dated

The **word count** of this document (excluding the References section) is:

Signed.....

Dated

IV. Declaration to include on project

UNIVERSITY OF DUBLIN

TRINITY COLLEGE

SCHOOL OF GENETICS AND MICROBIOLOGY

SMURFIT INSTITUTE OF GENETICS

DECLARATION FOR PROJECT

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** or **AI generated**.

Signed.....

Dated

The **word count** of this document (excluding the References section and figure legends) is:

Signed.....

Dated

V. *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 evacuation 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.

| | |
|---|---|
|  | Gas under pressure Symbol: Gas cylinder |
|  | Explosive Symbol: Exploding bomb |
|  | Oxidising Symbol: Flame over circle |
|  | Flammable Symbol: Flame |
|  | Corrosive Symbol: Corrosion |
|  | Health hazard/Hazardous to the ozone layer Symbol: Exclamation Mark |
|  | Acute toxicity Symbol: Skulls and Crossbones |
|  | Serious health hazard Symbol: Health hazard |
|  | Hazardous to the environment Symbol: Environment |

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/

VI. 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.

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

VII. Academic Year Structure 2023/24

Academic Year Calendar 2023/24

| Assessment Commence Week | Week beginning | 2023/24 Academic Year Calendar | | Term / Semester |
|-----------------------------|-------------------|---|---|--|
| | | UG continuing years / PG all years | UG new first years | |
| 1 | 28-Aug-23 | Reassessment * (Semesters 1 & 2 of 2022/23) | | ←Midwinter term begins/Semester 1 begins |
| 2 | 04-Sep-23 | Orientation (Postgraduates, Visiting & Erasmus); Marking/Results | | |
| 3 | 11-Sep-23 | Teaching and Learning | | ←Midwinter teaching term begins |
| 4 | 18-Sep-23 | Teaching and Learning | Orientation (JF UG) | |
| 5 | 25-Sep-23 | Teaching and Learning | Teaching and Learning | |
| 6 | 02-Oct-23 | Teaching and Learning | Teaching and Learning | |
| 7 | 09-Oct-23 | Teaching and Learning | Teaching and Learning | |
| 8 | 16-Oct-23 | Teaching and Learning | Teaching and Learning | |
| 9 | 23-Oct-23 | Study/Review | Study/Review | |
| 10 | 30-Oct-23 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | |
| 11 | 06-Nov-23 | Teaching and Learning | Teaching and Learning | |
| 12 | 13-Nov-23 | Teaching and Learning | Teaching and Learning | |
| 13 | 20-Nov-23 | Teaching and Learning | Teaching and Learning | |
| 14 | 27-Nov-23 | Teaching and Learning | Teaching and Learning | |
| 15 | 04-Dec-23 | Revision | Revision | |
| 16 | 11-Dec-23 | Assessment * | Assessment ** | ←Midwinter term ends Sunday 17 December 2023/Semester 1 ends |
| 17 | 18-Dec-23 | Christmas Period - College closed 22 December 2023 to 1 January 2024 Inclusive | Christmas Period - College closed 22 December 2023 to 1 January 2024 Inclusive | |
| 18 | 25-Dec-23 | | | |
| 19 | 01-Jan-24 | | | |
| 20 | 08-Jan-24 | Foundation Scholarship Examinations ^ | Foundation Scholarship Examinations ^ | |
| 21 | 15-Jan-24 | Marking/Results | Marking/Results | ←Hilary Term begins/Semester 2 begins |
| 22 | 22-Jan-24 | Teaching and Learning | Teaching and Learning | ←Hilary teaching term begins |
| 23 | 29-Jan-24 | Teaching and Learning | Teaching and Learning | |
| 24 | 05-Feb-24 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | |
| 25 | 12-Feb-24 | Teaching and Learning | Teaching and Learning | |
| 26 | 19-Feb-24 | Teaching and Learning | Teaching and Learning | |
| 27 | 26-Feb-24 | Teaching and Learning | Teaching and Learning | |
| 28 | 04-Mar-24 | Study/Review | Study/Review | |
| 29 | 11-Mar-24 | Teaching and Learning | Teaching and Learning | |
| 30 | 18-Mar-24 | Teaching and Learning (Monday, Public Holiday) | Teaching and Learning (Monday, Public Holiday) | |
| 31 | 25-Mar-24 | Teaching and Learning (Friday, Good Friday) | Teaching and Learning (Friday, Good Friday) | |
| 32 | 01-Apr-24 | Teaching and Learning (Monday, Easter Monday) | Teaching and Learning (Monday, Easter Monday) | |
| 33 | 08-Apr-24 | Teaching and Learning | Teaching and Learning | |
| 34 | 15-Apr-24 | Revision | Revision | ←Hilary Term ends Sunday 21 April 2024 |
| 35 | 22-Apr-24 | Trinity Week (Monday, Trinity Monday) | Trinity Week (Monday, Trinity Monday) | ←Trinity term begins |
| 36 | 29-Apr-24 | Assessment * | Assessment * | |
| 37 | 06-May-24 | Marking/Results (Monday, Public Holiday) | Marking/Results (Monday, Public Holiday) | |
| 38 | 13-May-24 | Marking/Results | Marking/Results | |
| 39 | 20-May-24 | Marking/Results | Marking/Results | |
| 40 | 27-May-24 | Research | Research | ←Trinity Term ends Sunday 2 June 2024/Semester 2 ends |
| 41 | 03-Jun-24 | Research (Monday, Public Holiday) | Research (Monday, Public Holiday) | |
| 42 | 10-Jun-24 | Research | Research | |
| 43 | 17-Jun-24 | Research | Research | |
| 44 | 24-Jun-24 | Research | Research | |
| 45 | 01-Jul-24 | Research | Research | |
| 46 | 08-Jul-24 | Research | Research | |
| 47 | 15-Jul-24 | Research | Research | |
| 48 | 22-Jul-24 | Research | Research | |
| 49 | 29-Jul-24 | Research | Research | |
| 50 | 05-Aug-24 | Research (Monday, Public Holiday) | Research (Monday, Public Holiday) | |
| 51 | 12-Aug-24 | Research | Research | |
| 52 | 19-Aug-24 | Research | Research | |

* Note: additional/corrigency days may be required outside of the formal assessment/assessment weeks.
 - Note: It may be necessary to hold a small number of JF examinations/assessments outside of semester 1.
 ^ Note: It may be necessary to hold some examinations/assessments in the preceding week.

Key Dates:

| | |
|----------------------------------|--|
| Orientation Week: | Monday 04 September to Friday 8 September 2023 |
| Study/Review Week: | Monday 23 October to Friday 27 October 2023 |
| Revision Week Semester 1: | Monday 4 December to Friday 8 December 2023 |
| Study/Review Week: | Monday 4 March to Friday 8 March 2024 |
| Revision Week Semester 2: | Monday 15 April to Friday 19 April 2024 |
| Trinity week: | Monday 22 April to Friday 26 April 2024 |

VIII. Progression and Awards

Refer to Calendar General Regulations.

IX. Graduate Attributes

The Trinity Graduate Attributes represent the qualities, skills and behaviours that you will have the opportunity to develop as a Trinity student over your entire university experience, in other words, not only in the classroom, but also through engagement in co- and extra-curricular activities (such as summer work placements, internships, or volunteering).



The four Trinity Graduate Attributes are:

- To Think Independently
- To Act Responsibly
- To Develop Continuously
- To Communicate Effectively

Why are the Graduate Attributes important?

The Trinity Graduate Attributes will enhance your personal, professional and intellectual development. They will also help to prepare you for lifelong learning and for the challenges of living and working in an increasingly complex and changing world.

The Graduate Attributes will enhance your employability. Whilst your degree remains fundamental, also being able to demonstrate these Graduate Attributes will help you to differentiate yourself as they encapsulate the kinds of transversal skills and abilities, which employers are looking for.

How will I develop these Graduate Attributes?

Many of the Graduate Attributes are 'slow learned', in other words, you will develop them over the four or five years of your programme of study.

They are embedded in the curriculum and in assessments, for example, through undertaking independent research for your final year project, giving presentations and engaging in group work.

You will also develop them through the co-curricular and extra-curricular activities. If you help to run a club or society you will be improving your leadership skills, or if you play a sport you are building your communication and team-work skills.