



INSTITUTE OF  
GENETICS & CANCER



Medical  
Research  
Council



THE UNIVERSITY  
of EDINBURGH



CANCER  
RESEARCH  
UK



# GRADUATE RESEARCH & TRAINING

HANDBOOK 2022





# Welcome

Graduate Research and Training Contacts

Staff Student Liaison Committee/Officer



# GRADUATE RESEARCH & TRAINING

HANDBOOK 2022

## Welcome

We are delighted to welcome you to your postgraduate training programme at the Institute of Genetics and Cancer. On the following pages you will find information relating to the different programmes, timetable for the first 6 months, and the assessment timetable for the next 3 or 4 years.

As you probably know, we have a mixture of students on campus, some of whom are following a four year PhD programme with rotations and others who are starting a three year PhD in a specific lab, whilst others are studying for an MSc or MD. There are some teaching elements of the four year taught course that might be of interest to other students, for example covering different technologies, computer programming, aspects of clinical research and research ethics - these are shown in a detailed timetable. This teaching is only compulsory for the 4-year HGU students, but other students (and postdocs) are welcome to sign up and attend any sessions that you find useful; you might want to discuss your choice with your supervisor(s). We hope that the Graduate Research and Training environment will provide a useful framework for your studies. Please feel free to air your views, and to approach us about any issues you have, and help us to make the Institute a huge success!

## Graduate Research and Training contacts

The Institute is made up of three centres, the MRC Human Genetics Unit (HGU), the Centre for Genomics and Experimental Medicine (CGEM) and the Cancer Research UK Edinburgh Centre (ECRC), each with their own Graduate Convenor. The Institute falls within the School of Molecular, Genetic and Population Health Sciences (you will need to know this School affiliation when you apply for Transskills courses amongst other things), and the SMGPHS is within the College of Medicine and Veterinary Medicine or CMVM.

In the first instance you will mainly deal with your supervisors, Graduate Convenor or Nick Gilbert (Director of Graduate Research and Training for the Institute of Genetics and Cancer). You will also have a thesis committee (normally setup about 10 weeks into your PhD) which will be made up of your supervisors, an external advisor and a committee Chair. Formal issues (interruption of studies and so on) are dealt with by the Director of Graduate Research and Training and the College PG Office.

Head of School of MGPHS:  
**Professor Sarah Cunningham-Burley**

Director of Graduate Research and Training,  
Institute of Genetics and Cancer:  
**Professor Nick Gilbert**

Graduate Convenor, CRUK Edinburgh Centre:  
**Professor Val Brunton**

Graduate Convenor, MRC Human Genetics  
Unit: **Professor Nick Gilbert**

Graduate Convenor, Centre for Genomic &  
Experimental Medicine: **Dr Kathy Evans**

Director of PG Studies, College of MVM:  
**Professor Paddy Hadoke**

Staff Student Liaison Officer:  
**Dr Catherine Naughton**  
**Dr Dasa Longman**

Graduate Research and Training  
Administrator:  
**Pauline McDonald**

Graduate Research and Training Assistant:  
**Alana Johnson**

## Staff Student Liaison Committee

At the Institute of Genetics and Cancer we are committed to ensuring a high-quality student experience. To ensure we are able to deliver this, and to “maximise our students’ potential”, we encourage students to communicate their views and suggestions to help influence any required changes to policies and procedures. The Institute Staff Student Liaison Committee (SSLC) meets biannually to discuss matters of mutual concern of staff and students. The SSLC is composed of student and staff representatives, and we strongly encourage students at any stage of their graduate degree to consider joining the SSLC. The current Staff Student Liaison Officers are (SSLO) Dr. Catherine Naughton and Dr. Dasa Longman.



Catherine Naughton

Catherine is a senior research scientist in Professor Nick Gilberts laboratory in the MRC, Human Genetics Unit. She has over 15 years experience as a post-doctoral scientist and has mentored and supervised many PhD students.



Dasa Longman

Dasa is a Senior Scientist in the lab of Professor Javier Caceres, MRC HGU, and has many years experience of formal and informal mentoring of PhD and undergraduate students.

Catherine and Dasa together oversee the POGs induction events held during induction week for new PhD students, coordinate the 1st-year student journal clubs and organise the biannual SSLC meetings.

## What to do if things go wrong

If you have a problem with your project and/or supervisor, you should first try to resolve it between yourselves - it is important to keep lines of communication open where possible and not let things degenerate. If there is still a problem, then please seek advice - you should feel free to speak to your second supervisor, your thesis committee Chair, the Directors of the Graduate School or the PG Convenor for your building.

These conversations will be in confidence and a strategy will be devised to try and address any problems. Additional meetings of thesis committees can be arranged (subject to members’ availability) if the student and/or supervisors feel that this would help. If you are not happy with the outcome of frontline resolution (and on the rare occasions where a local resolution is not an appropriate early step) the University has procedures in place for dealing with complaints and the Institute of Genetics and Cancer adheres to these procedures rigorously. Details of these can be accessed through the CMVM Postgraduate Wiki which is also accessible from the Institute of Genetics and Cancer Graduate Research and Training web pages.

## Meet the Team: PG Directors



Professor Nick Gilbert - Director of Graduate Research & Training HGU/  
Institute of Genetics and Cancer

Email [Nick.Gilbert@ed.ac.uk](mailto:Nick.Gilbert@ed.ac.uk)  
Telephone 0131 651 8551 Location: C3.21

Research Group  
[www.ed.ac.uk/mrc-human-genetics-unit/research/gilbert-group](http://www.ed.ac.uk/mrc-human-genetics-unit/research/gilbert-group)



Dr Susan M Farrington - Graduate Convenor, CRUK Edinburgh Centre

Email [Susan.Farrington@ed.ac.uk](mailto:Susan.Farrington@ed.ac.uk)  
Telephone 0131 651 8632

Research Group  
[www.ed.ac.uk/cancer-centre/research/farrington-group](http://www.ed.ac.uk/cancer-centre/research/farrington-group)



Dr Kathy Evans - Graduate Convenor, CGEM

Email [Kathy.Evans@ed.ac.uk](mailto:Kathy.Evans@ed.ac.uk)  
Telephone 0131 651 8747 Location: N2.09

Research Group  
[www.ed.ac.uk/centre-genomic-medicine/research-groups/evans-group](http://www.ed.ac.uk/centre-genomic-medicine/research-groups/evans-group)

Students and staff should contact their local Centre PG Director for academic support.

### Administration Team



Pauline McDonald



Alana Johnson

Email [student-admin@jgc.ed.ac.uk](mailto:student-admin@jgc.ed.ac.uk)  
Telephone 0131 651 5771 Location: CG.11

Pauline and Alana manage the day-to-day administration of the Graduate Research and Training programme, and are based on the ground floor of the MRC Human Genetics Unit.

For queries related to Postgraduate Research and Training, Pauline and Alana provide support to prospective, on-programme and visiting students, as well as supervisors and academic staff. When appropriate, they will signpost students and staff to key central university services.

Pauline and Alana work closely with Centre PG Directors to enhance the Student Experience and oversee the following areas of work:

- Student Recruitment & Admissions
- Tier 4 Engagement & Monitoring process for international students
- Visiting student admissions
- Manage Graduate Research and Training website in liaison with PG Directors
- Coordinate teaching programme
- Organise student events e.g. Science at the Interface to Industry, Christmas lectures, John Inglis talks etc.
- Organise and minute Staff Student Liaison Committee (SSLC) / Postgraduate Studies Committee (PGSC)
- Manage Student Social Media Platforms

# Induction Week Teaching Timetable



**GRADUATE  
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# GRADUATE RESEARCH & TRAINING HANDBOOK 2022

## Induction Week

Monday 12th September	
10:00 - 10:30	PGR Director's welcome – Nick, Susan (Lecture Theatre)
10:30 - 11:00	Head of School welcome – Sarah Cunningham-Burley (Lecture Theatre)
11:15 - 12:00	Health & Safety – Iain Kennedy (Lecture Theatre)
Break for Lunch	
14:00 - 15:00	Director of Student Health and Wellbeing welcome – Andy Shanks (Lecture Theatre)
15:00 - 16:30	POGS TEAM: Buddy Session (Soft Seating Area of Nucleus)
Tuesday 13th September	
09:00 - 10:00	IT General Familiarisation – Kenny Burns & IT Team (MEC Computing Lab 1)
11:00 - 11:30	Sharlotte Patterson – CMVM Welfare Advisor (TBC)
Break for Lunch	
13:00 - 14:00	Student Disability and Learning Service – Jan Gardiner (Lecture Theatre)
14:00 - 15:30	Good Practice in PhD Research – Grace Alston and Lucy Scott (Lecture Theatre)
16:00 - 16:30	HGU students – introduction to 4 year PhD programme – Nick Gilbert (E4:07)
Wednesday 14th September (E4.07)	
09:00 - 10:00	Public Engagement Session (with comms) – Dee Davison
10:30 - 11:00	Institute of Academic Development – Louise MacKay
11:00 - 11:30	Library Services – Ruth Jenkins (TBC)
11:30 - 12:00	Equality and Diversity – Dee Davison and Peter Tennant
Thursday 15th September HGU PI Talks (S1.15)	
09:00	Andrew Jackson
09:15	Colin Semple
09:30	Joe Marsh
09:45	Martin Taylor
10:00	Pleasantine Mill
10:15	Luke Boulter
10:30	Liz Patton
Break	
11:00	Andrew Wood
11:15	Richard Meehan / Sisi Dimova
11:30	Tom Deegan

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11:45	Ian Adams
12:00	Ailith Ewing
12:15	Nick Gilbert
<b>12:30 - 14:00 (Waitrose Lunch with Group Leaders, S1 Break Out Area)</b>	
14:15	Hannah Long
14:30	Andy Badrock
15:00	Wendy Bickmore
<b>Break</b>	
15:45	Chris Ponting
16:00	Grzegorz Kudla
16:15	Chloe Stanton / Amy Findlay
<b>Friday 16th September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Intro to UNIX
<b>Monday 19th September (Online but students can use the MEC room if need be)</b>	
09:00 - 17:00	Open science and FAIR in practice
<b>Tuesday 20th September (Online but students can use the MEC room if need be)</b>	
09:00 - 17:00	Open science and FAIR in practice
<b>Wednesday 21st September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Intro to Eddie
<b>Thursday 22nd September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Ensembl genome browser
<b>Friday 23rd September (MEC Computing Lab 1)</b>	
09:00 - 12:30	UCSC genome browser
<b>Monday 26th September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Intro python
<b>Tuesday 27th September (MEC Computing Lab 1)</b>	
09:00 - 17:00	GIT
<b>Wednesday 28th September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Intro to R for genomics
<b>Thursday 29th September (MEC Computing Lab 1)</b>	
09:00 - 17:00	Intro to R for genomics
<b>Friday 30 September</b>	
09:30 - 16:30	Intro statistics

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<b>Monday 3rd October HGU Students Start Rotation Project 1 - E4.07</b>	
09:00 - 12:00	Good Research Practice - Helen Nickerson & Pauline Ward, Alison Meynert
<b>Thursday 6th October (E4.07)</b>	
09:00 - 11:00	Experimental design - Luke / Kevin
<b>Tuesday 11th October (E4.07)</b>	
11:00 - 12:00	Scientific Blogging - Lorna Campbell
<b>Wednesday 12th October (E4.07)</b>	
09:30 - 11:00	Reading and evaluating the scientific literature - Catherine Naughton & Dee Davison
<b>Thursday 13th October (E4.07)</b>	
14:00 - 17:00	Next Gen Sequencing Technology - Lee Murphy / Nick Gilbert
<b>Thursday 20th October (S1.14)</b>	
14:00 - 15:00	<b>Journal Club 1</b> - Emma Hall
<b>Monday 24th October (E4.07)</b>	
09:00 - 11:00	Experimental design - Kevin Myant / Luke Boulter
<b>Thursday 27th October (E4.07)</b>	
09:30 - 11:30	Biological Imaging - Ann Wheeler & Team
<b>Thursday 3rd November (E4.07)</b>	
09:00 - 12:00	Advanced Proteomics and Metabolomics - Alex von Kriegsheim, Jair Marques Junior & Roopesh Krishnankutty
<b>Monday 7th November (E4.07)</b>	
14:00 - 17:00	Experimental Model Systems - Alessandro Brombin & Ian Adams
<b>Thursday 10th November</b>	
09:30 - 12:30	Translating your research - Helen Nickerson & Sarah Trewick (E4.07)
14:00 - 15:00	<b>Journal Club 2</b> - Elias Friman (S1.14)
<b>Thursday 17th November (MEC Computing Lab 1)</b>	
10:00 - 12:30	Analysing Imaging data - Laura Murphy
<b>Monday 21st November (E4.07)</b>	
14:00 - 15:00	Drug Development - Neil Carracher & Stefan Symeonides
<b>Thursday 24th November</b>	
09:30 - 11:30	Super resolution imaging - Ann Wheeler & team (E4.07)
14:00 - 15:00	<b>Journal Club 3</b> - Emily Webb (S1.14)
<b>Friday 2nd December (South Seminar Room)</b>	
HGU Student Christmas Talks (2021 4 year HGU DTP Intake)	
<b>Monday 5th December (E4.07)</b>	
9:30 - 11:30	Genome Engineering - Andrew Wood & Pleasantine Mill

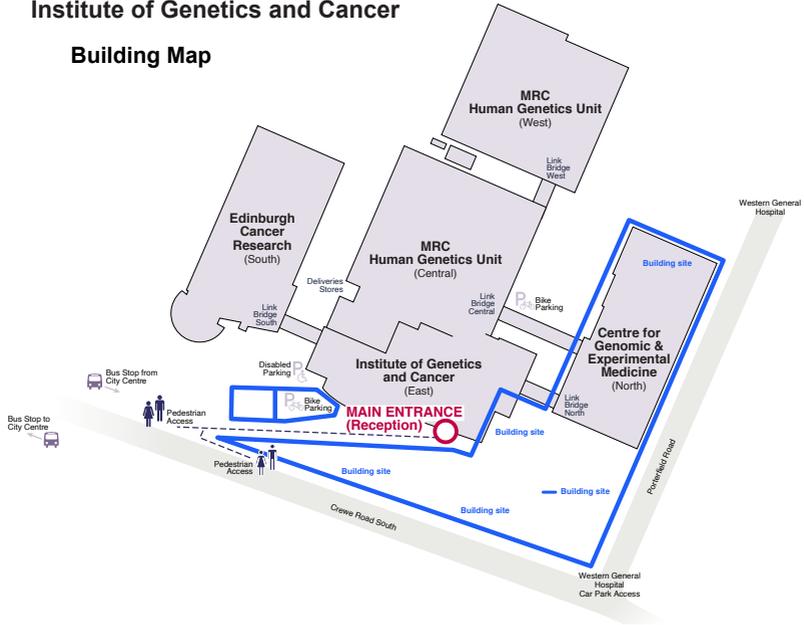
<b>Thursday 8th December (E4.07)</b>	
<b>HGU Rotation 1 talks</b>	
<b>Monday 19th December</b>	
<b>HGU Rotation Student Project Write-Up deadline</b>	
<b>Monday 2nd January</b>	
<b>Mini Induction week</b>	
<b>Monday 9th January (E4.07) HGU Students Start Rotation Project 2</b>	
09:00 - 12:00	Molecular ageing and senescence - Tamir Chandra
14:00 - 17:00	Computational Cancer Genomics - Colin Semple
<b>Monday 16th January</b>	
09:00 - 12:00	Molecular mechanisms of pathogenic mutations in proteins - Joe Marsh (E4.07)
14:00 - 15:00	<b>Journal Club 4</b> - Craig Anderon (S1.14)
<b>Monday 23rd January (E4.07)</b>	
10:00 - 12:00	Linking long-range enhancers and chromosome topology to human craniofacial development, evolution and disease - Hannah Long
14:00 - 17:00	Fundamentals of Size - Andrew Jackson
<b>Monday 30th January</b>	
09:00 - 12:00	Using biochemical reconstitution to understand fundamentals of genome replication - Tom Deegan (E4.07)
14:00 - 15:00	<b>Journal Club 5</b> - Laura Monaghan (S1.14)
<b>Monday 6th February (E4.07)</b>	
09:00 - 12:00	Establishing mechanisms underlying genetic associations with complex traits and diseases - Veronique Vitart / Chloe Stanton / Amy Findlay
14:00 - 17:00	Allele-aware functional genomics - Chris Ponting / Breeshey
<b>Monday 13th February</b>	
09:00 - 12:00	Genome architecture and congenital abnormalities - Laura Lettice (E4.07)
14:00 - 15:00	<b>Journal Club 6</b> - Fay Newton (S1.14)
<b>Monday 20th February (E4.07)</b>	
09:00 - 12:00	Using zebrafish models to study melanocyte stem cells and melanoma - Liz Patton
14:00 - 17:00	Innate immune signalling of self-nucleic acid in human disease - Yanick Crow
<b>Monday 27th February</b>	
10:00 - 11:00	Building biological tubes – can we use developmental biology to understand adult disease and cancer? - Luke Boulter (E4.07)
14:00 - 15:00	<b>Journal Club 7</b> - Sachin Wani (S1.14)

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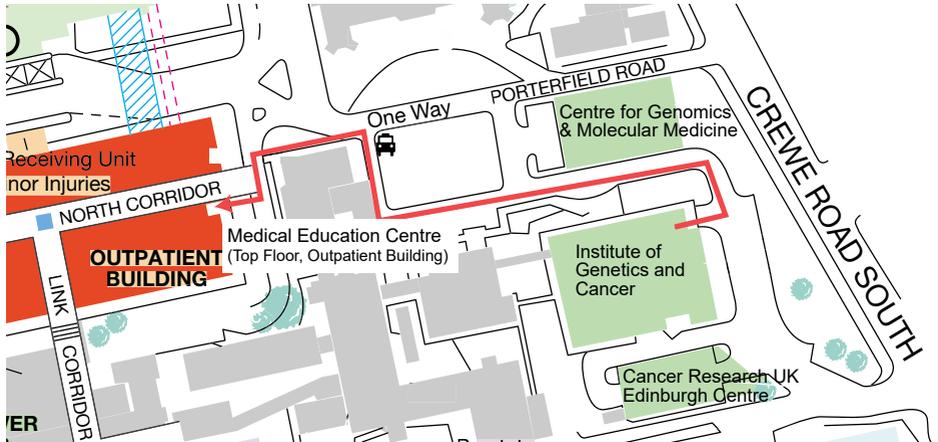
<b>Monday 6th March (E4.07)</b>	
09:30 - 12:00	The 3D genome and gene regulation - Wendy Bickmore
14:00 - 17:00	Post-transcriptional regulation of gene expression: From splicing to RNA quality control mechanisms - Javier Caceres
<b>Monday 13th March</b>	
09:00 - 12:00	Meiosis and Meiotic Chromosomes in the Mammalian Germline - Ian Adams (E4.07)
14:00 - 15:00	<b>Journal Club 8</b> - Lukas Tamayo Orrego (S1.14)
<b>Monday 20th March (E4.07)</b>	
09:00 - 12:00	Saturation mutagenesis - Greg Kudla
14:00 - 17:00	Degron technologies: engineering genomes, proteins and small molecules to make better models of human disease - Andrew Wood
<b>Monday 27th March (E4.07)</b>	
09:30 - 12:00	What role might epigenetics play in human disease? - Duncan Sproul
<b>Thursday 3rd April</b>	
HGU Rotation 2 talks	
<b>Friday 4th April</b>	
HGU Rotation student project write-up deadline	
<b>Monday 7th April</b>	
HGU PhD Starts	

**Institute of Genetics and Cancer**

**Building Map**

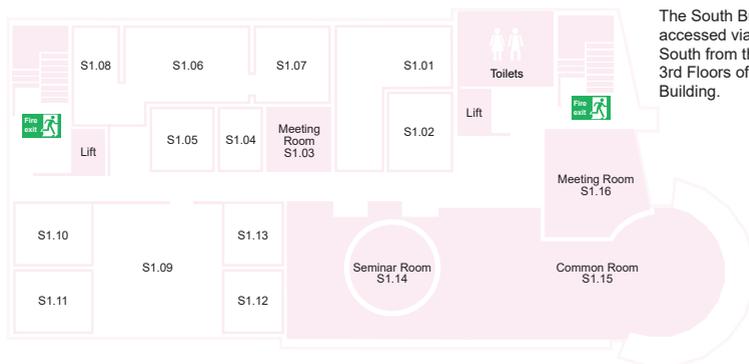


**NHS Outpatients Building Computing Suite 1  
Medical Education Centre, 3rd Floor**



**Institute of Genetics and Cancer  
Cancer Research UK Edinburgh Centre South Seminar Room S1.14**

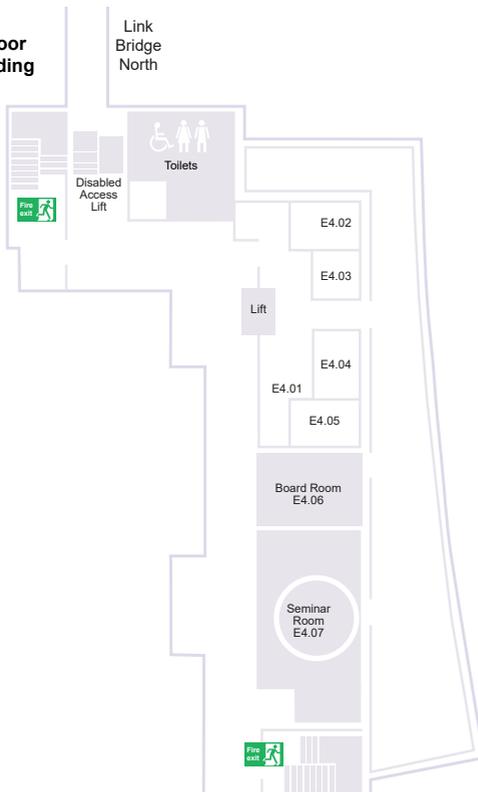
**First Floor  
South Building**



The South Building can be accessed via Link Bridge South from the 2nd and 3rd Floors of the Central Building.

**Institute of Genetics and Cancer**

**Fourth Floor  
East Building**



# Assessment Guidelines for all students



## **GRADUATE RESEARCH & TRAINING** HANDBOOK 2022

## Assessment Guidelines

### PhD, MD, MScR assessment guidelines

During the course of your studies you will regularly be assessed. This will comprise writing reports, attending and presenting at thesis committee meetings and completing an annual review on EUCLID. For part time students assessments should happen every year and follow this format.

In the Institute our assessments are based on the CMVM guidelines and further information can be found on the CMVM wiki (<http://edin.ac/2crLMTx>)

### Annual reviews on EUCLID

All students need to complete an annual review on EUCLID which will be signed off by you, your supervisors and postgraduate director. Over the course of your project you will complete an annual review to coincide with your 10-week, first year, second year and every subsequent year until you finish your studies. In some cases your thesis committee will decide that an interim meeting (e.g., half way through your second) or an additional meeting (e.g., at the end of the third year of a three year funded PhD) would be helpful. Please ensure your reports and feedback are uploaded onto EUCLID for sign-off. The online student portal (EUCLID) can also be used to record other important milestones in your training in Edinburgh and your supervisor may log individual meetings with you on this system.

### Student reports

As a guide these are the reports required for different programmes

	MSc by Research	3 year PhD	4 year PhD	MD
10 week report	✓	✓	✓	✓
6 month report	✓			
1st year report		✓	✓	✓
2nd year report		✓	✓	
3rd year report			✓	

### 10 week report

This report should be concise (1000 words excluding title, references, abstract or figure legends). As this report is being written at the beginning of your studies, we are most interested in what you plan to investigate over the next year. The report should include:

- Title, and the names of you and your supervisor.
- An abstract of less than 100 words.
- Introduction that provides sufficient background information for the reader to understand the proposal and that puts the scientific question(s) into context.
- A section that states the scientific question(s) that are being asked and the aims of the project.
- A short section on any progress made to date.
- A section describing your proposal for the next year's work.

- Figures can be added in any section to help describe the project or to show any data that you have obtained in the first few weeks of your project. Figure legends should provide succinct description of the figure.
- Reference List.

On completion, the report should be uploaded onto EUCLID and submitted to the Graduate Research and Training team: student-admin@igc.ed.ac.uk. Following submission you will be given feedback in the form of an email and/or meeting (depends on programme). This is also a good time to plan the composition of your thesis committee (see below).

### First Year Review:

- submitted at 9 month stage for PhD students
- submitted at 6 months for MScR students

The next assessment stage is the first-year review. This rigorous review is your opportunity to demonstrate your suitability to progress and will consist of three elements:

- a written report from the student
- a meeting with the student and thesis committee
- a written report by the thesis committee

**Student's written report:** The report should adopt a logical format and be of a high standard. It should be typed and free of typographical and grammatical errors. A clear statement of the aims of the project should be included in addition to a brief account of methods and their validation. Whilst it is recognised that at this stage students may not have substantial data, preliminary results should be documented and interpreted with a clear statement of intent as to immediate future studies (these might be expected to form the basis of discussion at interview). The text should be referenced as for a scientific paper and references listed at the end of the report. It is expected that the report should be around 5000 words. It should incorporate diagrams, figures and tables as necessary. Preliminary drafts of the report should be discussed with supervisors. It is often useful to ask your supervisor for an example report from a previous student. The student's report should be available to members of the thesis committee at least one week before the thesis committee meeting, allowing time for adequate consideration of the reports, and reports should be uploaded onto EUCLID.

**Thesis Committee Meeting:** This meeting will involve the student and thesis committee. The meeting is normally expected to include a short (10-15 minute) presentation by the student introducing the project, describing methodology and any preliminary results and identifying future studies. Students are strongly encouraged to rehearse with supervisors before the interview. You should expect the thesis committee to discuss specific points of content and organisation arising from the written report during the course of interview. You will have an opportunity to initiate a dialogue and, if necessary, raise matters of concern with the committee.

**Feedback:** The thesis committee should make an assessment of the student's written report, performance at interview and overall progress. The student should be informed of the committee's opinion during the meeting, they will then write a report, normally within one week of the meeting, summarising the assessment. Good and very good progress should be credited; any unsatisfactory aspects of performance should be clearly defined with an attempt to identify underlying reasons. It should make clear recommendations as to subsequent progress and action and be signed by all members of the committee. The student will have an opportunity to see the report, and be able to discuss strengths, weaknesses and any issues of concern with the chair in the absence of his/her supervisor(s). The student can also add comments before signing the report. An unsatisfactory report may be used for future discussions or as the basis for re-registering students for a different degree or in very rare cases discontinuing studies

(see outcomes). It is therefore essential that clear details of remedial action or the reasons for change in registration are documented. The signed thesis committee assessment should be uploaded onto EUCLID.

**Outcomes:** An initial recommendation will be made as to whether student progress is satisfactory or is inadequate in one or more aspects. In the case of inadequate performances a further recommendation from the thesis committee will be needed in terms of whether the student is (i) re-assessed or (ii) re-registered for a different degree, change in period of study or discontinued. In these cases it would be expected that students are totally unsatisfactory or severely deficient in several areas of their study.

### Second Year Review

The second-year report does not need to be as long as the first year report but should contain a clear indication of achievable plans for the following year and an outline plan for the thesis. As for the first year review the student should organise a meeting with the thesis committee who will also write a report. Your second-year report and assessment from the thesis committee should be uploaded onto EUCLID.

### Subsequent Reviews

For four year and continuing students there will be reviews every year until submission. Sometimes these will require a thesis committee meeting and this should be discussed with your supervisor.

### Final Year Talk

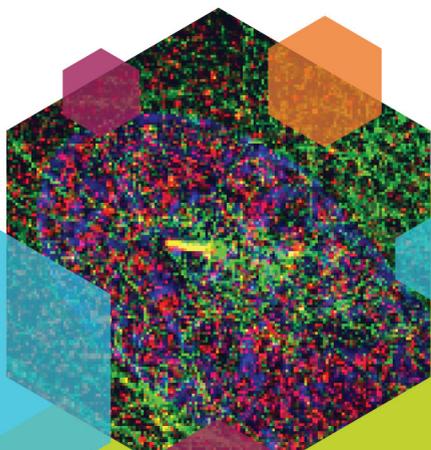
Students in their final year will be scheduled to give a talk to their centre. These are a fun opportunity to present to your friends and colleagues and should be seen as an opportunity to showcase your work. These will be organised by student admin and your graduate director.

### Thesis committee

The composition of the thesis committee will vary depending on your programme of study. It will comprise of your supervisors including a day to-day lab supervisor where appropriate, an external committee member and a Chair. The external may be from the same building, but should be independent of the supervisors. The Chair should be someone with experience of student supervision of at least Senior Lecturer level. For MScR and MD the roles of the chair and external are often combined.

# General Information

- Postgraduate transferable skills programme
- Social media
- POGS
- Social committee
- Annual/sick leave
- Health and Wellbeing
- Pastoral Support Committees
- Annual Student / Supervisor Structured Discussion



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## Postgraduate transferable skills programme - Institute of Academic Development (IAD)

[www.ed.ac.uk/institute-academic-development](http://www.ed.ac.uk/institute-academic-development)

The acquisition and development of generic research and transferable skills is an important part of postgraduate training. Courses covering a wide range of skills are available to postgraduate research students in the Graduate School of Medicine & Veterinary Medicine through the transferable skills programme. This programme concentrates on the professional development of postgraduates, providing courses directly linked to postgraduate study (e.g. Thesis Workshop, Good Practice and Academic Paper Writing) and future careers (e.g. Successful Career Strategies for PhD Students, Local GRADschools). The programme also provides information on other training opportunities for postgraduates.

Courses are free of charge to postgraduate students in the College of Medicine and Veterinary Medicine. The programme has been designed to be as flexible as possible so that each student can tailor the content and timing of the programme to their own requirements. Most courses are run several times each year and last for between half a day and a day.

## Workshops for postgraduate researchers by theme

The following workshops make up the core programme open to all postgraduate researchers, and are displayed by theme.

## Research Planning and Management

- Managing your Research Project
- Practical Project Management for Research Students
- Viva Survivor
- Innovation School
- Managing your Research Data

## Communication and Impact

- Designing Effective Slides
- Public Speaking, Networking and Engaging
- Poster Production
- Presenting made Easy – Presentation Techniques
- Presenting Made Easy – Delivering Presentations
- Presenting your Poster Pitch
- Research, Researchers and the Media, a hands on approach to communicating your research

## Writing and Publishing

- Academic writing peer review
- Beating Writers Block
- Developing a Writing and Publishing Strategy in the Internet Age
- Effective Writing: Grammar
- How to be your own best editor
- Is my writing 'Academic' Enough?
- Just Write
- Proof Reading
- Text, Coherence, Structure and Argumentation
- The Writing Process: Getting Started
- Writing a Literature Review
- Writing Abstracts
- Writing Clinic
- Writing for Publication
- Writing Retreat
- Writing Well: Language and Style
- Academic Writer – Creative Writer
- An Introduction to Copyright and Publishing
- This is what I do... and this is why it matters

## Digital and Library Skills

- Beginners Guide to Imaging
- Searching Literature and Managing Bibliographies
- Managing a Bibliography in Endnote
- Finding Academic Literature
- Social media for impact: strategy, connecting & metrics

## Statistics

- Statistical Consultancy 1:1 Session
- Introductory Statistics for Life Scientists

## Personal Effectiveness

- Conference and Events Organising
- Creating Effective Collaboration
- Creative Problem Solving for Researchers
- Imposter Syndrome: Why Successful people often feel like frauds
- Ease the Load – Feel good about your busy life
- How to be an Effective Researcher
- Mapping your Mind
- Seven Secrets of a Highly Successful Research Student
- Simply Assertive
- Speed Reading
- Teambuilding and Leadership Fundamentals
- Think Strategically Respond Rapidly
- Managing your Work, your Goals and Yourself

## Public Engagement

- Communications Toolkit for a Public Audience
- Dialogue: Public Engagement Beyond Public Lectures!
- Storytelling Techniques for Effective Communication
- Voice and Presentation Skills Workshop
- How to Design a Public Engagement Process
- Facilitation skills for public engagement
- An Introduction to Public Engagement

## Online learning

PhD student online training courses (topics include statistics; imaging; academic writing; and data management). Some you can do any time, and others run at specific times of the year.

- Statistics courses
- Imaging for scientists
- Academic writing
- Research Ethics and Integrity - an introduction
- Data management training
- Ready to research

The Edinburgh Local GRADschool is open to all PhD students in their final or penultimate year of study:

[www.ed.ac.uk/institute-academic-development/postgraduate/doctoral](http://www.ed.ac.uk/institute-academic-development/postgraduate/doctoral)

## Advice on using social media networks & confidentiality of information

Facebook, Twitter and other social media networks have changed the way we interact with each other and like them or not, they are a part of our society.

As some of you will carry out research where animals are involved, please ensure that you follow procedures to ensure our work continues to be ethical, credible and professional. Sharing images/discussions of animal work outside of the context of academic discourse is not appropriate. This not only applies to posts on social network sites but to informal discussions in the pub or on the bus.

## Please remember you must not post the following information:

- Scientific research information, analysis, results or any other information and /or images relating to your work.
- Location details of research buildings where animal work is carried out.

## Be mindful of your responsibilities

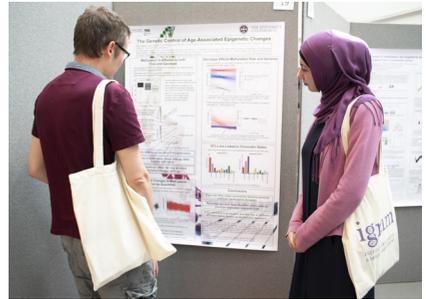
- Data Protection legislation - do not disclose other people's personal information without prior permission.
- Be aware that any posts you make in a professional capacity (even private posts) are subject to data protection and freedom of information and may need to be disclosed.
- University policies apply: Students must not post materials about their work and locations if doing so would carry a risk to themselves and especially to others, including the University as an organisation (see section 5 University policies).

[www.ed.ac.uk/website-programme/training-support/guidelines/social-media](http://www.ed.ac.uk/website-programme/training-support/guidelines/social-media)



## POGS

The Postgraduate Society (POGS) is a student-run committee open to the Institute students from all years and centres. Our aim is to improve the student experience, promote collaboration, provide support and have fun! By organising events throughout the year we bring students together, helping them develop skills and career perspectives. Our most popular events include the annual student retreat, Burns Night ceilidh, pub quiz, poster evening, and careers event. All students are welcome to take part so don't hesitate to come say hi!



POGS is jointly funded by the Institute and the Deanery, which means (almost) all of our events are completely free! Joining the POGS committee is a great way to get involved with the Institute community, and have your say on how events are run. Meetings are held approximately once a month, and we are always looking for new committee members. To get involved, contact us at: [pogs@igc.ed.ac.uk](mailto:pogs@igc.ed.ac.uk).



## Buddy Session by POGs

The buddy session is to provide new students with a pair of buddies who are in the final years of their PhD. The buddies are there to answer any Institute-related questions the new students have and provide help with any problems that might arise. At the buddy session small groups of students will be assigned to two buddies who will have your academic email address. The buddies will introduce themselves and get to know their students, share with them their email address and their offices so you will be able to find them anytime you want to speak to them. Follow up sessions will be organised by the buddies later in the year to catch up on students progress settling into the Institute.



**Best wishes, POGS**

## Vacation Leave

Students can take up to eight weeks' vacation time in a year, with agreement from their supervisor. There is no need to apply for an interruption of study when taking vacation leave.

## Sick Leave

The policies on sick leave are evolving and depend on your funder. Please check information from your funding organisation or contact your programme director or Student Admin for advice.

## Pastoral Support Committees

From September 2021 all students will be assigned a Pastoral Support Committee. This is completely independent of your thesis committee and will comprise two postdoctoral 'mentors' who will be based in different research teams and centres from you. The Pastoral Support Committee is there for to ask for advice, help, anything that you feel is not best addressed to your supervisor or thesis committee. The committee can meet as often as you like but at least once per year. Minutes won't be taken from the meetings but we will ask the committee to let us know when they have met.

Further information can be found on the IGC Graduate Research and Training website: <https://www.ed.ac.uk/institute-genetics-cancer/igc-graduate-research-and-training/information/student-pastoral-support-committees>

## Student Support

The Institute of Genetics and Cancer is a family, looking out for each other. We are excited that you are becoming part of our family. If you need any local support a good place to start is with you supervisor. They will understand your situation and will want to look out for you. Alternatively please contact student admin ([student-admin@igc.ed.ac.uk](mailto:student-admin@igc.ed.ac.uk)) or one of the postgraduate directors (Nick Gilbert, Val Brunton, Kathy Evans) and more information about different types of support is available at the back of this handbook.

Edinburgh university has lots of expertise in looking after students and a good place to start is the student Health and Wellbeing webpage: <https://www.ed.ac.uk/students/health-wellbeing>.

## Code of Practice

<https://www.ed.ac.uk/student-disability-service/staff/supporting-students/help-distressed-students>

<https://www.ed.ac.uk/files/atoms/files/copsupervisorsresearchstudents.pdf>

### **Annual Student / Supervisor Structured Discussion**

Many factors are important for a successful PhD. One of them is a good relationship between supervisor and student. To help this relationship it is important to be open and honest with each other and to manage each other's expectations. This is particularly important as the project progresses when there might be changes either due to the project or external factors. In our experience many problems can be avoided by having a dialogue, however this can be difficult for both parties, with neither wanting to put the other on the spot.

To overcome this problem, we would like all students and supervisors to have a “structured discussion” at the start of the PhD, and every year afterwards. In the first instance we will not monitor the results of the discussion, but instead ask students to let us know when you have had the meeting with your supervisor. In some cases, meetings will be online, in other cases face to face. Likewise for some PhD projects it might be relevant for all supervisors to participate, in other cases just the lead supervisor. Similarly, some topics will be relevant for different stages of your PhD.

<https://www.ed.ac.uk/institute-genetics-cancer/igc-graduate-research-and-training/information/annual-student-supervisor-structured-discussion>

# Student Health and Wellbeing

## Feeling Good App



The Foundation for Positive Mental Health is Working with the University of Edinburgh to provide free access to the Feeling Good App.

[www.ed.ac.uk/student-counselling/self-help/apps-podcasts-ted-talks-relaxation-recordings/feeling-good-app](http://www.ed.ac.uk/student-counselling/self-help/apps-podcasts-ted-talks-relaxation-recordings/feeling-good-app)

## Membership to the Consent Collective



Online support materials on consent, sex, gender, sexual harassment and relationships.

[www.consentcollective.com/edinburgh](http://www.consentcollective.com/edinburgh)

## Student Disability and Learning Support Service



Supports students with a range of health conditions, learning differences, disabilities and some temporary injuries.

[www.ed.ac.uk/student-disability-service](http://www.ed.ac.uk/student-disability-service)

## Student Counselling Service



Supports the mental health of all students at the University through short term counselling and referral to other support.

[www.ed.ac.uk/student-counselling](http://www.ed.ac.uk/student-counselling)

## Advice Place



Professional, impartial and inclusive service for all students at the University of Edinburgh.

[www.eusa.ed.ac.uk/support\\_and\\_advice/the\\_advice\\_place/](http://www.eusa.ed.ac.uk/support_and_advice/the_advice_place/)

## University Health Centre



NHS General Practitioners who rent premises from the University and offer full G.P. services to patients.

## Chaplaincy



The Chaplaincy is a safe and welcoming space for people of all faiths and none. Chaplaincy offers a range of support including Mindfulness, Yoga and the Listening Service.

[www.ed.ac.uk/chaplaincy](http://www.ed.ac.uk/chaplaincy)

## EUSA Mental Health and Wellbeing Society



Provides an informal and friendly space where students can learn more about the importance of mental wellbeing.

<https://www.eusa.ed.ac.uk/>

## SilverCloud



Online cognitive behavioural therapy.

[www.ed.ac.uk/student-counselling/what-is-silvercloud](http://www.ed.ac.uk/student-counselling/what-is-silvercloud)

## Togetherall



An online service offering self-help programmes, creative outlets and a peer support community monitored by mental health professionals.

[www.togetherall.com/en-gb/](http://www.togetherall.com/en-gb/)

If you would like to discuss student health and wellbeing or any of the resources above, please contact: [student-admin@igc.ed.ac.uk](mailto:student-admin@igc.ed.ac.uk)



WELLBEING  
AT IGC

# MENTAL HEALTH DURING YOUR PhD

AN OPINION PIECE BASED ON SOME RESEARCH I DID IN MY LUNCH HOURS AND MY ENTIRELY UNQUALIFIED EXPERIENCE\*



A study by the University of California, Berkeley, found nearly half of postgraduate students met criteria to classify them as depressed.<sup>1</sup>

## WHAT YOU MAY BE EXPERIENCING/FEELING (YOU ARE NOT ALONE, I PROMISE)

### IMPOSTER SYNDROME



Someone is going to figure out you don't belong here soon. You look good on paper, but passing that exam was a fluke. I don't have what it takes to [do these experiments, write a thesis, succeed in academia]. These are all classic signs of imposter syndrome. **Tip: reframe your thinking. Aim for progress not perfection.**

### NO MORE TICK BOXES



You got pretty good at doing essay and lab reports - they were all short term tasks. You also got good at figuring out what questions might be asked in exams. Now you have an open ended project, with the end nowhere in sight. You no longer have grades to tell you if you are doing a good job. Transitioning from this undergraduate mentality can be particularly tough. **Tip: break down your research into small, manageable goals.**

### FIRST TIME FAILING

You've always been the best student at school, and you did pretty well at university too. Now your science isn't working and everyone around you seems to be getting on just fine. These feelings can come about as at undergraduate level, experiments (believe it or not) are designed to work. **Tip: remember, you are at the forefront of scientific research - if it was easy it would already have been done!**



### ISOLATION / GUILT

Writing your thesis can be a particularly lonely, isolating task. This can also be coupled with feelings of guilt when going about your daily life as "you should be writing". Tips to manage this include still attending research group meetings/departamental seminars whilst writing. This can also be coupled with 'writer's block'. **Tip: when writing, start by making figures - it is far easier to write about what a figure means.**



### COMPETITIVE LANDSCAPE



Unfortunately, academia often fosters competition over collaboration, when it should be the other way around. This is made worse by the fact that often the only way to gauge how well you are doing is to compare yourself against others. **Tip: no two PhD projects are the same, so avoid comparing them.**

### THE WORK | LIFE STRUGGLE



There is an inherent culture of acceptance in academia of long work hours. In fact, 40% of academics report working more than 50 hours a week.<sup>4</sup> This is a fault with the system. Presenteeism is a common trait observed in academia, where people work long hours due to anxiety/stress, but are not being efficient in these long hours. **Tip: aim to be efficient inside normal working hours then focus on "you" time.**

A hard truth is only 7 in 10 PhD graduates become full professors.<sup>2</sup> During your PhD, make sure to work on other "soft skills" as well as doing your research. Like making a poster for an online Twitter competition for example...

## ARE THOSE AROUND YOU STRUGGLING? HERE ARE SOME POSSIBLE WARNING SIGNS



INCREASED DRINKING



INCREASED EATING



DECREASED EATING



WORKING LONG HOURS



BEING ABSENT



JOKING ABOUT SUICIDE



LOOKING DISHEVELLED

SELF-HARMING?  
SUICIDAL THOUGHTS?  
CALL SAMARITANS NOW  
ON **116-123**  
OR EMAIL [JD@SAMARITANS.ORG](mailto:JD@SAMARITANS.ORG)

## SOME WAYS TO HELP MANAGE YOUR MENTAL HEALTH AND WELLBEING



### SEEK MEDICAL ADVICE

Speak to a medical professional about how you are feeling. This may lead to interventions such as medication or counselling to help you manage your mental health.



### TAKE SOME TIME OUT

Taking a break can actually improve efficiency when you return to work. If you cannot justify taking a couple of weeks off, take a series of long weekends to get some time away.



### FOCUS ON YOU

It is a proven fact that levels of sleep can add to feelings of stress. Exercise can also work to alleviate stress. It may feel like you don't have time, but going for a walk at lunchtime (if accepted) may be a positive change you can make.



### REQUEST COUNSELLING

There are a large amount of online resources available to help manage mental health and wellbeing. The services have counselling services that are available for you to use. This can be useful to talk through your problems and also make local appropriate coping strategies are in place.



### TALK TO YOUR SUPERVISOR

It is not always possible, but if you feel you can approach your supervisor, discuss your mental health concerns with them. Other options include discussing to reduce workload temporarily or take time out.



### TALK TO YOUR PEERS / POSTDOCS

It is helpful to find people around you who also experienced the stressful nature of a PhD. Reach out if you feel alone. Remember, postdocs have completed their PhD so they have some world-class coping strategies.



### CREATE MANAGEABLE CHUNKS

If everything is overwhelming, try to break down your research into manageable tasks. It may help to do this in consultation with your supervisor. Good luck to you, and to get back to the things you love, before losing all of other tasks like animals.



### READ LITERATURE

There are a large amount of online resources available to help manage mental health and wellbeing. The services have counselling services that are available for you to use. This can be useful to talk through your problems and also make local appropriate coping strategies are in place.

Is a PhD ever, if your PhD is heavily affecting your mental health and wellbeing, there is absolutely no shame in getting out early a new chapter in your life. There are plenty of successful people that got their PhD & it was - you might get up with an 8:00 filing I don't for a while, but you wouldn't want it for 4 years!

## REFERENCES

1. Graduate Student Happiness & Well-Being Report, 2018, University of California, Berkeley.
2. Sakulku, L. The Impostor Phenomenon. *The Journal of Behavioral Science*, 6(1), 75-97.
3. Graduate survey: A love-hurt relationship. *Nature*, 500, 549-552.
4. Guitery, Susan, et al. Understanding mental health in the research environment: A Rapid Evidence Assessment. Santa Monica, CA: RAND Corporation, 2017.
5. The Scientific Century: Securing our future prosperity. *The Royal Society*, 2010.

A poster by Dr. Zoe Ayres (not a medical professional). Free to distribute.



#TIMETOTALK  
#RSCPOSTER

\*ALTHOUGH I DID SURVIVE A PhD



## Mental Health Portal

Do you want to learn about mental health and improve your resistance to stress, anxiety and other pressures?

Visit the IGC Mental Health Portal

- Tips to improve Resilience
- Free resources for EVERYONE
- Access at your own pace, in your own time, with no pressure or deadlines
- Can be used to support others as well as yourself

IGC Mental Health Portal: <https://edin.ac/mental-health-portal>



## Mental Health First Aiders

Are you concerned about your mental health or need someone to talk to?

The IGC Mental Health First Aid team are here to help!

The team are:

- Trained to provide a confidential listening service for ALL staff and students
- Able to signpost to a range of different free resources, proven to help

For a full list of the IGC Mental Health First Aid Team visit:

<https://edin.ac/mental-health-first-aiders>



# Useful Links



# GRADUATE RESEARCH & TRAINING HANDBOOK 2022

## Useful links

### General

College PG Office contacts

<https://www.ed.ac.uk/medicine-vet-medicine/postgraduate/contact-us/>

College PG research wiki (includes PG handbook, software available to students etc.)

<http://edin.ac/2crLMTx>

Code of Practice

<https://www.ed.ac.uk/institute-academic-development/postgraduate/doctoral/advice-support/regulations>

Assessment regulations

<https://www.ed.ac.uk/files/atoms/files/2019-postgraduateresearch.pdf>

Transferable skills training and support

[www.ed.ac.uk/schools-departments/institute-academic-development/postgraduate/doctoral](https://www.ed.ac.uk/schools-departments/institute-academic-development/postgraduate/doctoral)

### Searching the literature/ bibliographic management

A tool for running daily searches

<http://pubcrawler.gen.tcd.ie/>

A free online alternative to Endnote and Reference Manager

[www.zotero.org/](http://www.zotero.org/)

(note also that many journals have free apps for browsing abstracts).

### Research Ethics

General

[www.pnas.org/content/86/23/9053.full.pdf](http://www.pnas.org/content/86/23/9053.full.pdf)

Image manipulation

[www.jci.org/articles/view/21717/pdf](http://www.jci.org/articles/view/21717/pdf)

[www.cell.com/abstract/S0092-8674\(06\)00676-3](http://www.cell.com/abstract/S0092-8674(06)00676-3)

<http://jcb.rupress.org/content/166/1/11.full>

## Writing papers, giving talks

Advice on writing papers

[www.nature.com/nature/journal/v467/n7317/full/nj7317-873a](http://www.nature.com/nature/journal/v467/n7317/full/nj7317-873a)

How to give a good talk

[www.sciencedirect.com/science/article/pii/S1097276509007424](http://www.sciencedirect.com/science/article/pii/S1097276509007424)

How to give a bad talk

[www.sciencedirect.com/science/article/pii/S0960982299802929](http://www.sciencedirect.com/science/article/pii/S0960982299802929)

Useful advice ranging from lab techniques to giving talks and posters

<http://bitesizebio.com>

The Advice Place, Potterrow Reception, EUSA  
5/2 Bristo Square, Edinburgh EH8 9AJ

Tel: 0131 650 2656

<https://www.eusa.ed.ac.uk/>

### Advice Guides and Resources

Here you can read all of our advice guides. If you would like them in an alternative format, please contact us and we will do our utmost to accommodate this.

# MRC Human Genetics Unit 4 Year Programme

- Introduction to programme
- Projects available
- Rotation timeline



**GRADUATE  
RESEARCH  
& TRAINING**  
HANDBOOK 2022

## The first six months

The HGU PhD program is following an exciting and innovative format. You will spend the first 6 months on an intensive training period leading up to your final choice of PhD project. This period comprises taught courses, talks from individual group leaders about their work, teaching sessions on a variety of topics from technology to clinical research, journal club sessions which will give you a chance to hone your analytic and presentation skills, and 2 rotation projects. The detailed timetable can be found in the handbook.

The choice of rotation projects is up to you (available projects are listed at the end of this section) and you can approach any relevant group leader to discuss the projects. You will see that there is some time between rotations, giving you a chance to look around and choose a new lab. The only formal constraint is that you must spend time in 2 different labs. You may find, of course, that another student has already been accepted and that the PI is only willing to take on one student (as is normally expected of PIs). If this happens then try again in the next rotation period; if there is a real clash then Nick Gilbert will help but do please try and resolve things between yourselves in the first instance. Bear in mind that there is no formal requirement for you to choose a PhD project in a lab in which you have done a rotation project, the rotations are just a chance for you to try different labs and projects out.

Many of the group leaders welcome students coming to their lab meetings which is a good way of seeing life in labs other than the ones where you are doing rotation projects, but please be sure to make contact with the appropriate PI in advance.

## The PhD

After 2 rotations you will choose a PhD project. We will have individual meetings with you to discuss your choices in the event of any clashes. No supervisor will be able to take on more than one student, HGU students must choose projects within the HGU, but apart from this you can go to any lab within the available project section. It is up to you to discuss possible projects with PIs you are interested in; this is a dynamic process in which you should be fully engaged. Note that supervisors are not obliged to take you on, you need to ask whether they are willing, or whether they have other interested students and so on. If your research project involves the use of animals or human participants, work must not commence until the relevant Home Office project and personal licences have been awarded, and appropriate Local Ethical Approval Committee has been granted. We will not be producing PhD project outlines from supervisors. Rather, at the PhD 10 week stage (June) you will have to produce a short report that outlines the project that you will pursue. This will then be discussed and refined if necessary by your supervisors (more detailed guidelines are given under Assessment Procedures). You will then spend 3 years in the lab, winding up by April of your final year. You will then have a further 6 months to write up your thesis but remember it is imperative that you submit your thesis by the final university deadline of September of year 4!

We hope that this novel structure for PhD study will be as exciting for you as it has been for us to develop it. We will be asking for your feedback at several stages of the course - please feel free to air your views, and approach us about any issues you have, and help us to make the HGU PhD programme a huge success!

**Nick Gilbert**

### Lab Rotations

Each student will do 2 rotation projects of around 3 months. Contact details and summaries of research interests of eligible supervisors are all given in this booklet (note there are some people unable to take students for rotations, please check), and during the first week you will be hearing research talks by some of these PIs.

The choice of rotation projects is up to you - you are responsible for approaching potential supervisors to discuss their willingness to take you on and to jointly come up with a plan of work. Remember the project won't be formally assessed as part of your PhD, so make the most of your time to experience different techniques, and get a feel for life in different labs.

The only formal constraint is that you must spend time in 2 different labs. You may find, of course, that another student has already been accepted and that the PI is only willing to take on one student (as is normally expected of PIs). If this happens then try again in the next rotation period; if there is a real clash then one of us will intervene but do please try and resolve things between yourselves in the first instance. Bear in mind that there is no formal requirement for you to choose a PhD project in a lab in which you have done a rotation project, the rotations are just a chance for you to try labs out.

At the end of each rotation you have to write a report about your project, to be handed in by the end of the week after you finish in that lab. This should be in the format used by journals such as those on Biomedcentral, i.e. divided up into brief sections of background, results and conclusions and no longer than two sides of A4 (excluding figures).

This abstract should be submitted to the Institute of Genetics and Cancer PGSC by emailing:

[student.admin@igc.ed.ac.uk](mailto:student.admin@igc.ed.ac.uk)

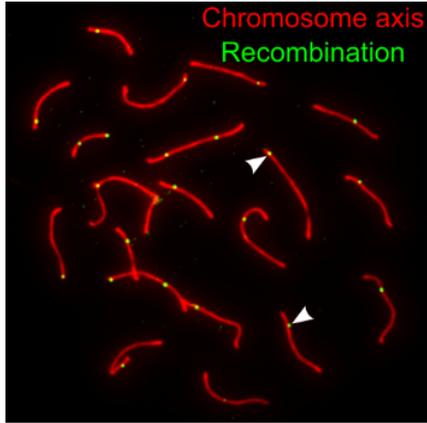
Supervisors will be asked for feedback on your performance in the lab and we will ask to meet up with you if there are any concerns. Towards the end of each rotation you will give a short assessed talk about your mini-project.

**How does chromosome structure regulate recombination and chromosome segregation in meiosis?**

**Supervisor: Dr Ian Adams**

**Rotation Oct-Dec  
Rotation Jan-March**

Pairing and recombination between homologous chromosomes during meiosis plays a key role in generating genetic variation, but defects in this process can cause the wrong number of chromosomes to be transmitted to the next generation. Each chromosome pair must have at least one crossover recombination event in order to segregate correctly, but it is not clear how each crossover then signals along the length of the chromosome to inhibit more crossovers from forming there. This project will investigate the role of the synaptonemal complex, a zipper-like protein structure that assembles between chromosome pairs, in regulating crossover recombination. We will



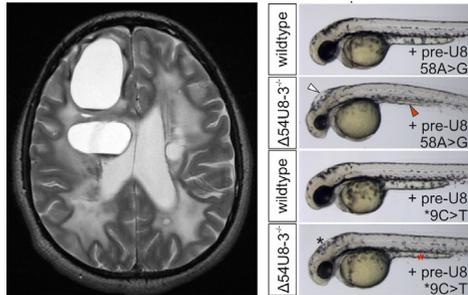
use gene-edited mice carrying mutations that interfere with synaptonemal complex assembly to perturb the structure of meiotic chromosomes, and super-resolution imaging to assess the effects on chromosome pairing, crossover recombination and chromosome segregation during meiosis.

**Exploring the role of the snoRNA U8 in human health and disease**

**Supervisor: Prof Yanick Crow**

**Rotation Oct-Dec  
Rotation Jan-March**

Evolutionarily conserved, ribosome biogenesis is required for protein production in all cells. Transcribed as a precursor, ribosomal RNA is processed and modified by small nucleolar RNAs (snoRNAs). This project will explore the role of snoRNAs in health and disease through a focus on the box C/D snoRNA U8, informed by our identification (Nat Genet 2016;48:1185-92) and characterisation (Am J Hum Genet 2020;106:694-706) of mutations in SNORD118, encoding U8, as the cause of a devastating Mendelian neurological disease. With an eye to translation, you will use both wet-lab and bioinformatic approaches to define the molecular and cellular biology of U8.

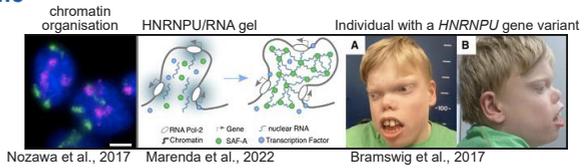


**How does a HNRNPU/RNA matrix regulate chromosome structure in health and disease?**

**Supervisor: Prof Nick Gilbert**

**Rotation Oct-Dec**

**Rotation Jan-March**



Chromatin is organised non-randomly in the nucleus. Subnuclear structures like the nucleolus, nuclear envelope, and other nuclear bodies, together with chromatin-associated protein complexes and chromatin-associated RNAs (caRNA) control the spatial organisation of chromosomes in three-dimensional (3D) space of the nucleus. Newly transcribed RNA interacts with HNRNPU (also called Scaffold-Attachment-Factor-A) to form a nucleoprotein gel that maintains structural stability of the genome. Through this process the HNRNPU/RNA gel decompacts chromatin and in its absence there is a radical change in chromosome structure. Abnormal spatial organisation of DNA is linked to many diseases like cancer, Huntington’s disease, progeria, and metabolic syndromes. Recently, mutations in HNRNPU have been linked to diseases with phenotypes including intellectual disability, autism spectrum disorder, and seizures. Interestingly, the effect of genome organisation and other nuclear processes in these diseases is not yet understood. Our hypothesis suggests that chromatin 3D mis-regulation disrupts normal nuclear processes such as gene expression and RNA export, leading to the disease phenotype.

To test this hypothesis we will make HNRNPU separation of function mutants in cell models to understand how HNRNPU/RNA can regulate chromosome structure. We will then make informative mutations in a transgenic mouse model using CRISPR editing to understand how these mutations function in a developmental context. This project will combine fundamental biology research with the translational potential of correcting HNRNPU function in human disease.

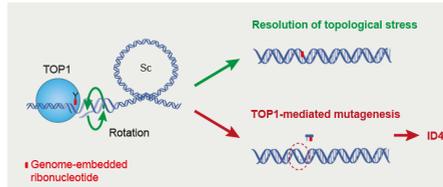
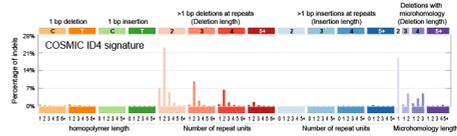
## Establishing the biological basis of cancer mutational signatures

Supervisor: Prof Andrew P Jackson

Rotation Oct-Dec  
Rotation Jan-March

Mutation underlies cancer development as well as evolution, so understanding how mutations are formed is important. Sequencing of cancer genomes has defined many mutational signatures, but for a substantial number of these the underlying mechanism is unknown. Linking such signatures to cellular pathways will be key to targeting future cancer therapy.

We recently established that the ID4 cancer signature is the result of Topoisomerase 1 activity at genome-embedded ribonucleotides (Reijns et al, 2022, Nature). To build on these findings, we are developing new methods to study mutational signatures using both mammalian cells and budding yeast. These methods aim to make use of novel reporters that allow magnetic



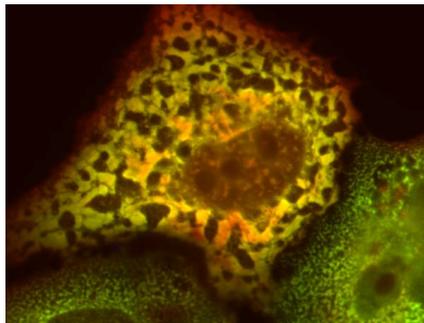
cell sorting combined with NGS and will be used to explore additional indel signatures and why ID4 differs between yeast and human. During a rotation you will have the opportunity to use molecular cloning, genome-editing, lentiviral transduction, mutation reporter assays, MACS, NGS library preparation and data analysis. As a PhD, this could develop into a wet/dry-lab project determining when and how mutations occurs in cancer.

## High-throughput discovery of disease mutations by in vivo deep mutational scanning

Supervisor: Dr Grzegorz Kudla

Rotation Oct-Dec  
Rotation Jan-March

Understanding which mutations lead to disease is a central goal of modern biology and medicine. Deep mutational scanning is a new approach that combines synthetic biology, next generation sequencing and computational analysis to systematically measure the effects of all possible mutations in a selected gene. So far, deep mutational scanning experiments were typically conducted in tissue culture. In collaboration with the Boulter and Khamseh labs, we will perform deep mutational scanning of tumour suppressor genes in an animal model of carcinogenesis, to explore the mechanisms of cancer formation in a physiological setting.



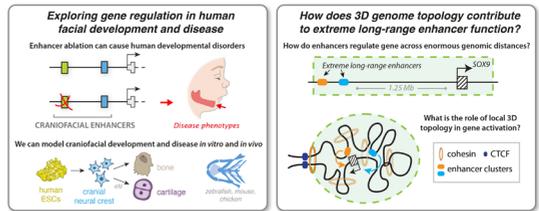
## Investigating long-range enhancers and 3D genome topology at a human craniofacial disease locus

Supervisor: Dr Hannah Long

Rotation Oct-Dec

Rotation Jan-March

Perturbation of gene regulation is central to many human genetic developmental disorders. In our lab, we study human facial development motivated by the wide diversity in facial appearance between individuals and the high frequency of birth malformations impacting the face. As our model, we focus on the SOX9 regulatory domain where non-coding mutations have been associated with facial dysmorphology. To explore mechanisms of disease, we leverage *in vitro* differentiation of facial progenitor (cranial neural crest) cells and have previously demonstrated that loss



of extreme long-range enhancers (Long et al, 2020) and perturbation of 3D genome topology (Chen\*, Long\* et al, in preparation) can be implicated in human disease. In this project, we will further explore the role of local 3D chromosomal structure in gene regulation. We will leverage human embryonic stem cells coupled with genetic engineering, reporter assays, genomics, and chromosomal imaging to explore the role of novel structural features in gene regulation across craniofacial development at loci implicated in craniofacial disease.

## Dissecting biological condensates during motile ciliogenesis

Supervisor: Dr Pleasantine Mill

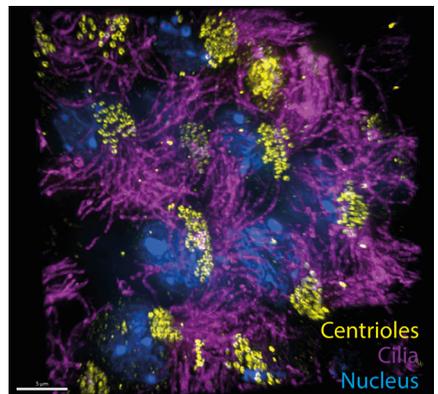
Day to Day supervisor: Emma Hall

Rotation Oct-Dec

Rotation Jan-March

Cilia are hair-like projections from the cell, vital for mammalian development. Primary cilia are immotile, and are essential for cell signalling, whereas motile cilia beat to produce fluid flow. Dysfunction of motile cilia can result in primary ciliary dyskinesia (PCD), characterised by recurrent airway infections and scarring, as well as infertility and hydrocephaly.

The Mill lab is developing novel therapeutic approaches including genome surgery for PCD. Understanding how, when and where we deliver these therapeutic approaches effectively requires detailed knowledge of motile cilia formation. Motile ciliogenesis involves the formation of 10-100s of cilia per cell. The initial step requires massive amplification of the centrioles, each of which then docks at the cell surface to nucleate a



Ependymal cilia (Magenta), nucleated by centrioles (yellow) lining the whole-mounted ventricles of the mouse brain

single cilium. Control of this process involves mysterious protein rich structures termed deuterosomes and fibrogranular material, but recent genetics suggests some redundancy in these processes. The student would learn dissection, immunofluorescence and high-resolution imaging techniques to characterise mouse models lacking these structures, to better understand the initial steps of motile ciliogenesis.

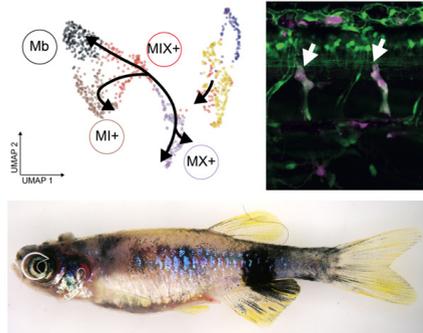
**Mapping the Genomic Landscape of the Melanocyte Stem Cell**

**Supervisor: Prof Liz Patton**

**Rotation Oct-Dec**

**Rotation Jan-March**

Melanocytes produce pigment, protect us from UV-irradiation, and are the lineage of origin for the deadly skin cancer, melanoma. Recently, we discovered that the transcription factor Tfap2b is specifically expressed in the melanocyte stem cell (McSC) in zebrafish, and that McSCs are the adult stem cell for multiple pigment cell types and nerve (associated) cells (PMID: 35021987). Excitingly, we find tfap2b+ cells in melanoma, suggesting an unappreciated stem cell state in melanoma. For this PhD project, we will use cellular genomics, imaging and fate mapping to understand how the Tfap2b transcription factor specifies the McSC and how it works with other transcriptional factors,



including the master melanocyte transcription factor, Mitfa, to shape the genome and cell fate of the McSC. This project has important implications for understanding how genomic mechanisms direct the fate of multi-potent stem cells, and how these stem cell pathways become dysregulated in melanoma.

**Long COVID: Phenotypes and Genotypes**

**Supervisor: Prof Chris P Ponting**

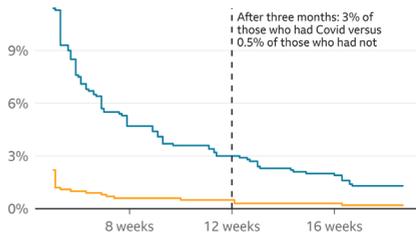
**Rotation Jan-March**

Around 200 million people worldwide are experiencing or have experienced Long COVID symptoms worldwide. 2.8% of the UK population have Long COVID, of whom 761,000 experience symptoms beyond a year following their initial SARS-CoV-2 infection. If we understood genetic risk factors for Long COVID then this could suggest candidate drugs for clinical trials and drug development opportunities.

In this project you will take advantage of soon-to-be released UK Biobank questionnaire data from over 161k individuals. Your aims will be to: (i) identify stratify people with Long COVID symptoms, and/or (ii) undertake initial genome-wide association analyses using SARS-CoV-2 infected individuals without these symptoms as controls. You would be working alongside Gemma Samms, a PhD student working on ME/CFS genetics.

**How long do coronavirus symptoms last?**

% of people **with** and **without** Covid experiencing symptoms



Source: ONS Coronavirus Infection Survey, 16 Sep 2021

*Figure. Most Long COVID symptoms have a prolonged probability of occurrence beyond 7 months.*

This rotation project is available in early 2023 when release of relevant UK Biobank data is expected.

## Origin and impact of germline mutations on regulatory sites in the human genome

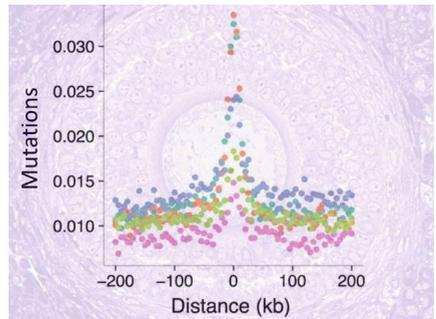
**Supervisors: Vera Kaiser & Colin Semple**

**Rotation Oct-Dec**

**Rotation Jan-March**

Inherited mutations occurring in the germline form the basis for biological evolution, but mutations do not occur randomly across the human genome. We have shown (Kaiser et al, Genome Res, 2021; see Figure) that transcription factor binding sites (TFBSs) that are active in the male germline are hotspots for particular mutations. Active regulatory sites (defined by ATAC-Seq in human spermatogonia) show elevated rates of deletion breakpoints and short insertions, causing a mutational cost to gene regulation. There is also thought to be substantial mutational input from the female germline and in early embryogenesis, but the mutational spectra at TFBSs active in these cells are unstudied. These studies are now feasible as a variety of chromatin data have recently been generated in human oocytes (Figure background) and early embryos.

We will test two main hypotheses relating to the germline origins and impact of human mutations. (i) TF binding events in the female germline and early embryo make distinct contributions to mutation patterns, involving mutational processes that contrast with those seen in the male germline. (ii) These patterns implicate different mutational and repair mechanisms, and cause different alterations to regulatory regions during evolution.

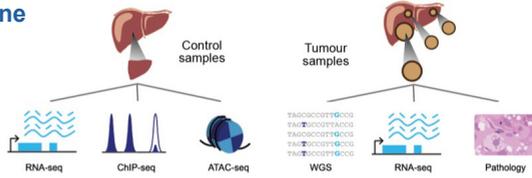


The project would best suit a bioinformatics student with an interest in the origin of germline variation or an enthusiastic biologist keen to develop bioinformatics skills.

## How does mutation impact gene expression in cancer?

**Supervisors: Stuart Aitken, Martin Taylor & Colin Semple**

**Rotation Oct-Dec**  
**Rotation Jan-March**



How do mutational loads at the regulatory regions of genes affect gene expression in tumours? This fundamental question in cancer genomics remains unresolved in spite of a decade of whole genome sequencing (WGS) studies of human tumours. We are studying early tumourigenesis in four different mouse strains treated with the same mutagen (Aitken et al, Nature, 2020), to produce abundant tumour samples (N=696) while controlling for the many confounding sources of variation seen in human tumour cohorts. In effect we have re-run tumour evolution many times under controlled conditions. This unique dataset benefits from WGS and RNAseq for tumours, plus in-depth characterisation of control samples targeted by the mutagen (see Figure).

The project would best suit a bioinformatics student with an interest in cancer or an enthusiastic biologist keen to develop bioinformatics skills.

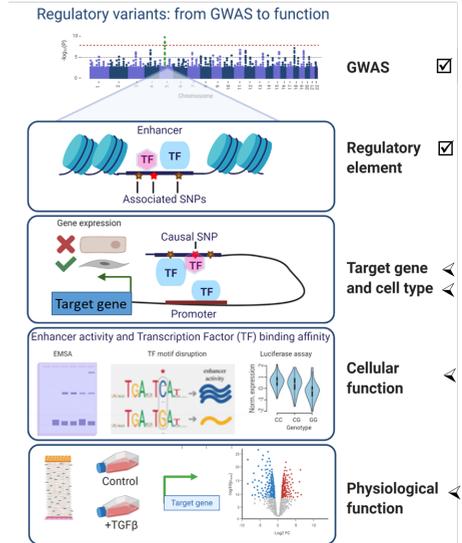
This project will determine the relationship between tumour mutational loads at known regulatory regions in the targeted cell type and the extensive dysregulated expression seen in the same tumours, to address the following questions. Are mutation loads correlated with expression changes? Are similar correlations seen in genetically distinct strains of mice or in tumours carrying different driver gene mutations? Are the expression patterns of particular genes or pathways disproportionately impacted by mutations?

**Functional analysis of a genetic association with the corneal disease Keratoconus within the SMAD3 gene**

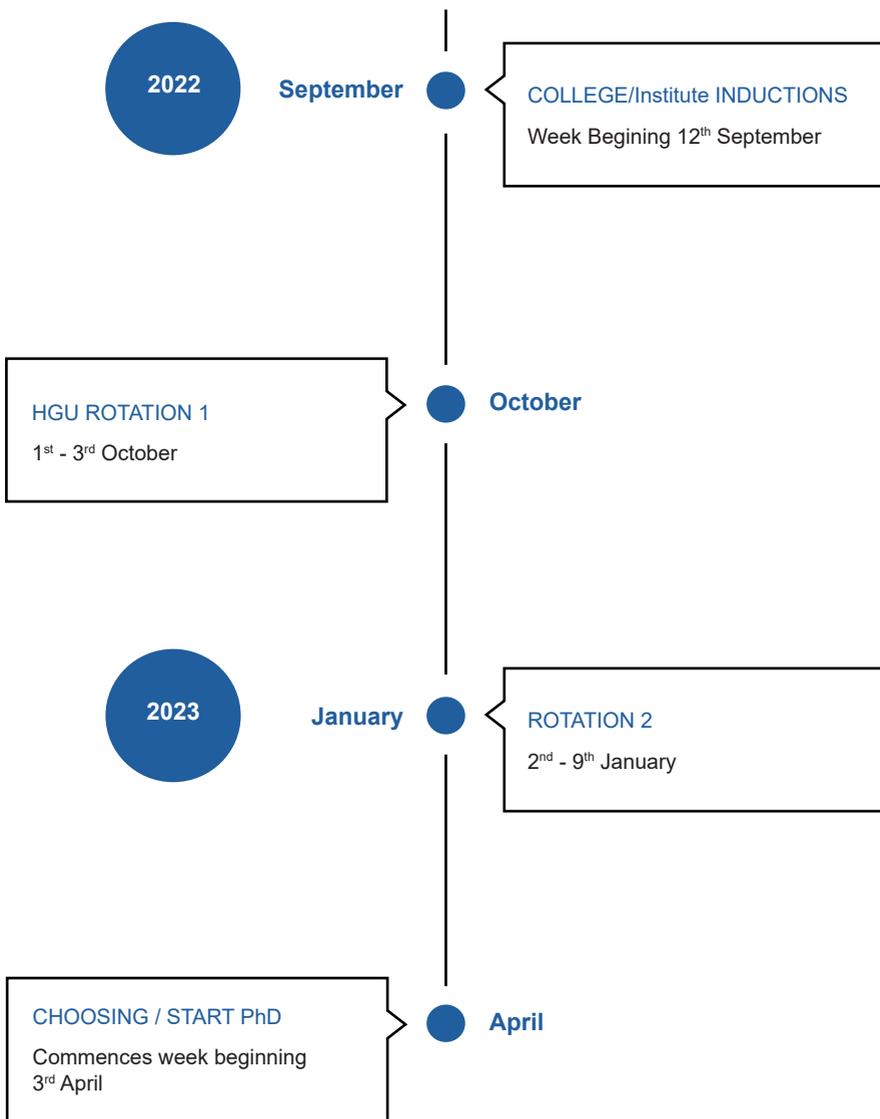
**Supervisor: Dr Veronique Vitart**

**Rotation Oct-Dec**  
**Rotation Jan-March**

Keratoconus is a multifactorial complex eye disease characterised by progressive weakening of the collagen-rich extra-cellular matrix in the cornea. A genetic risk locus of interest lies within an intronic regulatory region in SMAD3, a gene implicated in a rare systemic connective tissue disorder. Using cell type and allele specific assays, you will investigate how candidate causal variants might exert their effect. Hypothesised mechanism will be compared with that published for an independent regulatory locus affecting SMAD3, associated with coronary artery disease, as well as with that studied in the lab for another keratoconus risk loci. Spatio-temporal function for the regulatory region implicated could be also examined using animal models. This project has the scope to establish functional link with other risk loci implicated in keratoconus as well as to shed light on ECM regulatory pathway relevant to a broad range of diseases.



Training Timeline 2022 - 2023



# Student at the Institute?

## You have a new Facebook Group!

A closed group for announcements, course materials, discussions and a place to get to know your friends and colleagues.

Join by searching Facebook for OFFICIAL IGC Students or scan the QR code.



student-admin@igc.ed.ac.uk  
[www.facebook.com/groups/OFFICIALIGCStudents](http://www.facebook.com/groups/OFFICIALIGCStudents)

Find us on 



A new Facebook Group has been created for current on-programme students at the institute. This online space is a closed group and has been created specifically for students (not staff) for announcements, course materials, discussions and a place to get to know each other.

Join by searching Facebook for OFFICIAL IGC Students or visiting: [www.facebook.com/groups/OFFICIALIGCStudents](http://www.facebook.com/groups/OFFICIALIGCStudents)

### **Do I have to join the group?**

Yes. We hope the group will make life easier for everyone by having all the right information and people in the same place, reducing email traffic and providing a place for resources, questions and answers.

### **What if I'm not on Facebook or don't want to use my personal profile to join?**

That's ok – contact us and we can help you set up a new profile, just for life at the Institute.

### **What is a closed group?**

Only approved members of the group can see who the current members are and view posts in the group.

Anyone on Facebook can see the group's name and description, find it through search and request to join (requests are approved or declined by Administrators), but they can't see any of the content or members.

### **Who will be in the group and who moderates it?**

All postgraduate students on programme at the Institute.

Pauline and Alana are the Group Administrators with the Communications Manager as Moderator. Look out for group announcements from the Administrators – these flag key information. Join requests are approved by the Administrators, so no 'outsiders' will be able to join the group.

### **Can we say what we want?**

Although this is your group, remember that the group represents the Institute and we expect members to behave as professionally as they would in person on campus. Inappropriate posts will be moderated and removed.