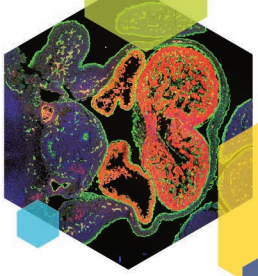
igc
INSTITUTE OF GENETICS & CANCER

UKRI Medical Research Council

THE UNIVERSITY of EDINBURGH

CANCER RESEARCH UK



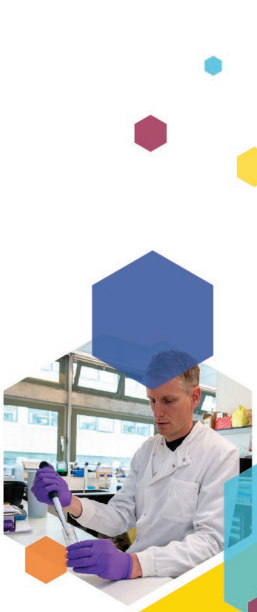
GRADUATE RESEARCH & TRAINING

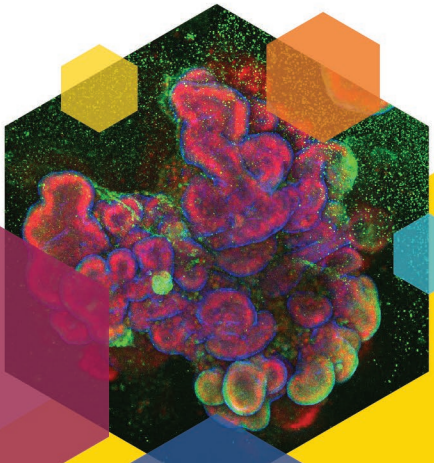
HANDBOOK 2026

Welcome

Graduate Research and Training Contacts

Staff Student Liaison Committee/Officer





GRADUATE RESEARCH & TRAINING

HANDBOOK 2026

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 post-docs) 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 of Genetics and Cancer is made up of centres, the MRC Human Genetics Unit (HGU) and the Cancer Research UK Edinburgh Centre (ECRC), each with their own themed graduate convenor. The Institute falls within the Institute of Genetics and Cancer (you will need to know this School affiliation when you apply for Transkills courses amongst other things), which 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 Kevin Myant (Director of Graduate Research and Training for the Institute of Genetics and Cancer). You will also have a thesis committee (set up at the start of your studies) which will be made up of your supervisors, an external advisor and a committee Chair. The chair will be decided at the beginning of your studies by the PG Convenors. The external advisor and the second supervisor for your studies will be decided by yourself and your principal supervisor. Formal issues (interruption of studies and so on) are dealt with by the Director of Graduate Research and Training and the College PG Office.

Director of Graduate Research and Training, **Professor Kevin Myant**

Deputy Director, CRUK Edinburgh Centre, Precision Medicine: **Professor Susan Farrington**

PGR Convener, MRC HGU: **Professor Ian Adams**

PGR Convener: **Dr Duncan Sproul**

Director of PG Studies, College of CMVM: **Professor Ruth Andrew**

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

PGR Administrator: **Alana Johnson**

Theme PGR Conveners

The IGC is organised into five different research themes, each consisting of around 15 research groups. The Postgraduate Conveners for each theme are listed below:

MRC Human Genetics Unit
Professor Ian Adams

Precision Medicine
Professor Susan Farrington

Therapy Discovery
Professor Kevin Myant

Biomedical Genomics
Professor Susan Farrington

Cell and Genome Biology
Dr Duncan Sproul / Professor Ian Adams

Disease Mechanisms
Dr Duncan Sproul

Epidemiology
Dr Duncan Sproul / Professor Kevin Myant

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 Officer is Dr. Dasa Longman.

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.

Dasa oversees the POGS induction events held during induction week for new PhD students, coordinates the 1st-year student journal clubs and organises 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 theme.

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 front-line 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 Kevin Myant - Director of Graduate Research & Training MRC HGU/Institute of Genetics and Cancer

Email Kevin.Myant@ed.ac.uk

Research Group

https://institute-genetics-cancer.ed.ac.uk/research/research-groups-a-z/myant-group

Professor Ian Adams - Graduate Convenor, MRC Human Genetics Unit

Email Ian.Adams@ed.ac.uk

Research Group

https://institute-genetics-cancer.ed.ac.uk/research/research-groups-a-z/adams-group

Professor Susan M Farrington - Deputy Director, CRUK Edinburgh Centre

Email Susan.Farrington@ed.ac.uk

Research Group

https://institute-genetics-cancer.ed.ac.uk/research/research-groups-a-z/farrington-group

Dr Duncan Sproul - Graduate Convenor

Email d.sproul@ed.ac.uk

Research Group

https://institute-genetics-cancer.ed.ac.uk/research/research-groups-a-z/sproul-group

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

Administration Support

Alana Johnson

Email student-admin@igc.ed.ac.uk

Location: CG.11

Alana manages 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, Alana provides 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.

Alana works 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 RESEARCH &TRAINING

HANDBOOK 2026

Induction Week

Monday 21st September	
09:30 - 10:00	PGR Welcome – Kevin, Ian, Susan, Duncan (E4.07)
10:00 - 11:00	Health and Safety Induction - Iain Kennedy (E4.07)
11:30 - 12:00	Health and Wellbeing - Andy Shanks (N5.01)
12:00 - 13:00	Equality and Diversity - Jennifer Lorigan (N5.01)
14:00 - 15:30	Good Practice in PhD Research - Maarten Van Den Anker, Ewa Ozga (E4.07)
15:30 - 17:00	Postgraduate Society (POGS) Social - POGS Team - Soft Seating NUCLEUS
Tuesday 22nd September	
09:00 - 12:00	HGU rotation project talks 1 (S1.16)
13:00 - 13:30	Edinburgh Innovations - Christina Starko (S1.14)
13:30 - 14:00	Library Services - Ruth Jenkins (S1.14)
14:00 - 14:30	EUSA Advice Session - EUSA Team (S1.14)
14:30 - 15:00	CMVM Welfare Advice - Faras Badani (S1.14)
15:30 - 16:00	PPIE Information Session - Jennifer Lorigan / Jenny Sharma (S1.14)
Wednesday 23rd September - HGU PhD Alumni Day	
Thursday 24th September	
11:30 - 12:30	Making the most of IT - Kenny Burn, Andrew Kirk (S1.14)
13:30 - 14:00	Bioinformatics Analysis core and advanced computational training- Graeme Grimes (MEC Computing Lab 1)
14:00 - 17:00	Good enough practices for scientific computing - Graeme Grimes (MEC Computing Lab 1)
Friday 25th September	
09:00 - 16:30	Intro to the comand line for genomics - Ewan McDowall (MEC Computing Lab 1)

Monday 28th September	
09:00 - 16:30	Project Organization management & data wrangling for genomics - Day 1 - Graeme Grimes (MEC Computing Lab 1)
Tuesday 29th September	
09:00 - 16:30	Data Wrangling for Genomics - Day 2 - Graeme Grimes (MEC Computing Lab 1)
Wednesday 30th September	
09:00 - 16:00	Introduction to Eddie - John ireland (MEC Computing Lab 1)
Thursday 1st October	
09:00 - 12:30	Intro to R and RStudio for Genomics - Day 1 - Jing Su (MEC Computing Lab 1)
13:30 - 16:00	HGU rotation project talks 2 (S1.14)
16:00 - 19:00	POGS Quiz - POGS Team (Nucleus)
Friday 2nd October	
09:00 - 12:30	Intro to R and RStudio for Genomics - Day 2 - Graeme Grimes (MEC Computing Lab 1)
13:30 - 17:00	HGU rotation project talks 3 (S1.15)
Monday 5th October	
09:00 - 16:30	Statistical thinking for Public Health, Simple Linear Regression - Hywell (MEC Computing Lab 1)
Tuesday 6th October	
09:00 - 16:30	Simple Linear Regression, Multiple linear regression for public health - Hywell (MEC Computing Lab 1)
Wednesday 7th October	
09:00 - 16:30	Intro to Python - Murray Wham (MEC Computing Lab 1)
Thursday 8th October	
09:00 - 14:00	Intro Git and GitLab - Murray Wham (MEC Computing Lab 1)
Friday 9th October	
09:00 - 14:00	Intro to Genome Browsers and IGV (MEC Computing Lab 1)

Monday 12th October HGU Rotation Programme (2026 Intake), 1st Rotation Project Start	
09:00 - 12:00	Reading and evaluating scientific literature - Ian Adams, Carolina Uggenti (E4.07)
14:00 - 15:30	TBC - Grzegla Kudla
Monday 19th October	
09:30 - 11:00	Scientific blogging - Rosie Free (E4.07)
14:00 - 15:30	Variant effect predictors - Ben Livesey (E4.07)
Thursday 22nd October	
16:00 - 18:00	POGS thesis writing workshop - POGS Team (E4.07)
Monday 26th October	
09:00 - 12:00	Degron tagging in human disease models: Opportunites and challenges - Andrew Wood (E4.07)
14:00 - 15:30	Critical Evalution Skills 1 - Carolina Uggenti (E4.07)
Monday 2nd November	
09:30 - 12:00	Biological Imaging - Ann Wheeler (E4.07)
14:00 - 15:30	Good Research Practice - Emma Hall (E4.07)
Monday 9th November	
10:00 - 11:00	Experimental Design Session 1 - Kevin Myant (E4.07)
14:00 - 15:30	Critical Evalution Skills 2 - Carolina Uggenti (E4.07)
Monday 16th November	
09:30 - 12:00	Experimental Design Session 2 - Kevin Myant (E4.07)
14:00 - 15:30	Next Generation Sequencing - Lee Murphy (E4.07
Monday 23rd November	
09:30 - 12:00	Analysing Imaging Data - James Iremonger (E4.07)
14:00 - 15:30	Critical Evalution Skills 3 - Carolina Uggenti (E4.07)

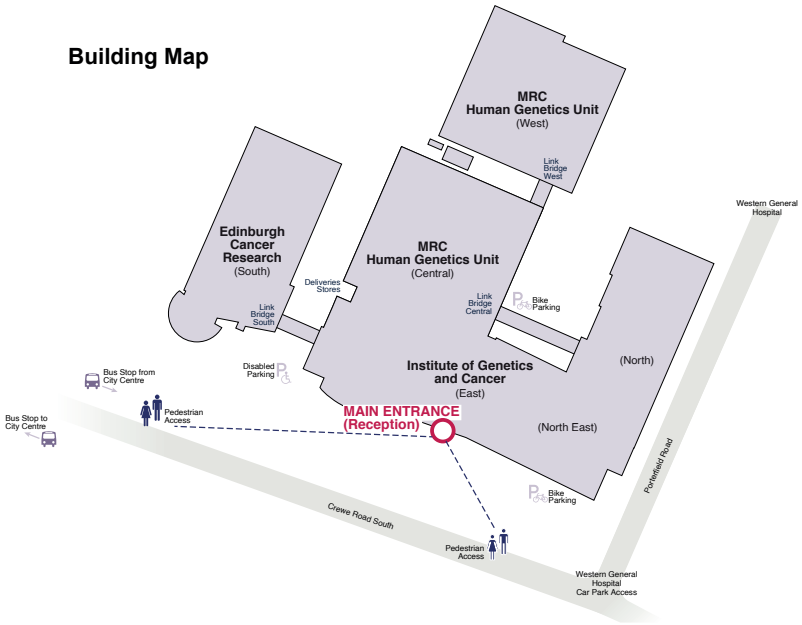
Monday 30th November	
10:30 - 12:00	Computational Cancer Genomics - Colin Semple (E4.07)
14:00 -15:30	Genome integrity disorders in the information age - Andrew Jackson (E4.07)
Monday 7th December	
09:00 - 12:00	Advanced proteomics and metabolomics - Alex Von Kreigsheim, Jair Macques Jnr (E4.07)
14:00 - 15:30	Critical Evalution Skills 4 - Carolina Uggenti (E4.07)
Friday 11th December - HGU students (2025 Intake) Christmas Talks E4.07	
Monday 14th December	
09:30 - 12:00	Experimental Model Systems - Cameron Wyatt, Laura Lettice (E4.07)
Monday 11th January - HGU Rotation Programme (2026 Intake), 1st Rotation Project Report Deadline	
Monday 11th January - HGU Rotation Programme (2026 Intake), 2nd Rotation Project Start	
10:00 - 11:00	Translating your research - Sarah Trewick, Kirsten McAuley (E4.07)
14:00 - 15:30	Critical Evalution Skills 5 Carolina Uggenti (E4.07)
Monday 18th January	
10:30 - 12:00	Drug Development - Neil Carragher, Stefan Symeonides (E4.07)
14:00 - 15:00	Biomedical data science - Catalina Vallejos (E4.07)
Monday 25th January	
09:00 - 11:30	Research Data Management Essentials - Jennifer Daub, Simon Smith (E4.07)
14:00 - 15:30	Critical Evalution Skills 6 Carolina Uggenti (E4.07)
Monday 1st February	
09:00 - 12:00	Genome Engineering - Andrew Wood (E4.07)
14:00 - 15:30	3D genome organisation. How to analyse it and determine its function? - Wendy Bickmore (E4.07)
Thursday 4h February	
16:00 - 18:00	POGS career event - POGS Team (E4.07)

Monday 8th February	
09:30 - 12:00	The genetics and genomics of ME/CFS, a complex disease - Chris Ponting (E4.07)
14:00 - 15:30	Critical Evalution Skills 7 Carolina Uggenti (E4.07)
Monday 15th February	
09:30 - 11:30	Alphafold modelling - Lukas Gerasimavicius (MEC Computing Lab 1)
14:00 - 15:30	RNA processing and gene regulation - Javier Caceres (E4.07)
Monday 22nd February	
09:30 - 12:30	Scientific graphics - Craig Nicol (E4.07)
14:00 - 15:30	Critical Evalution Skills 8 Carolina Uggenti (E4.07)
Monday 1st March	
09:30 - 12:30	Advanced Imaging Workshop - Ann Wheeler (E4.07)
14:00 - 15:30	Preventing inherited aneuploidies in the mammalian germline - Ian Adams (E4.07)
Monday 8th March	
09:00 - 10:30	Mechanisms of long - range gene regulation in development and disease - Hannah Long (E4.07)
11:00 - 12:30	Non-Coding mutations disrupt gene regulation and cause congenital defects - Laura Lettice (E4.07)
14:00 - 15:30	Critical Evalution Skills 9 Carolina Uggenti (E4.07)
Monday 15th March	
10:30 - 11:30	The role of epigenetics in human disease - Duncan Sproul (E4.07)
14:00 - 16:30	Critical Evalution Skills 10 Carolina Uggenti (E4.07)
Friday 19th March	
12:00 - 17:00	HGU Rotation Programme (2026 Intake), 2nd Rotation Project Talks (S1.14)
Monday 22nd March	
10:30 - 12:00	OH-MG, there is RNA in your DNA: causes and consequences of genome-embedded ribonucleotides - Martin Reijns (E4.07)
14:00 - 15:30	Chromatinopathies - Rebekah Tillotson (E4.07)

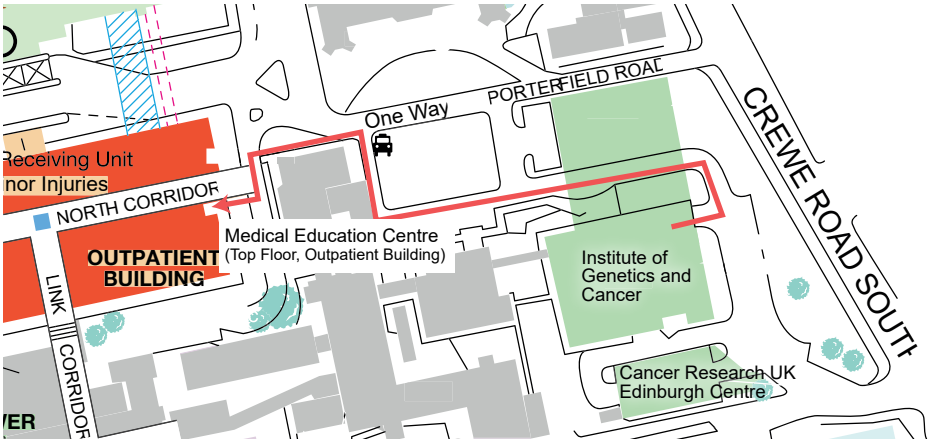
Friday 26th March - HGU Rotation Programme (2026 Intake), 2nd Rotation Project Report Deadline	
Monday 29th March - HGU Rotation Programme (2026 Intake), PhD Project Start	
Monday 7th June - HGU Rotation Programme (2026 Intake), 10 Week Report Deadline	
Monday 14th June	
14:00 - 17:00	HGU Rotation Programme (2026 Intake) 10 week report meeting (S1.16)
Tuesday 15th June	
14:00 - 17:00	HGU Rotation Programme (2026 Intake) 10 week report meeting (S1.16)
Wednesday 16th-18th June - POGS Retreat - Firbush - POGS Team	

Book your spot online!

Building Map

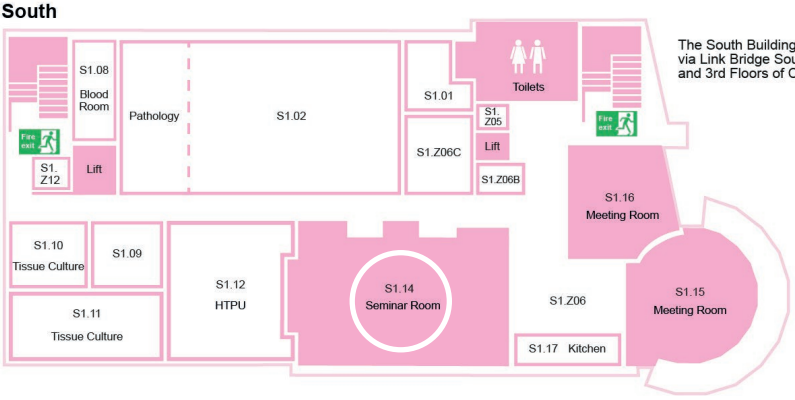


NHS Outpatients Building Computing Suite 1
Medical Education Centre, 3rd Floor

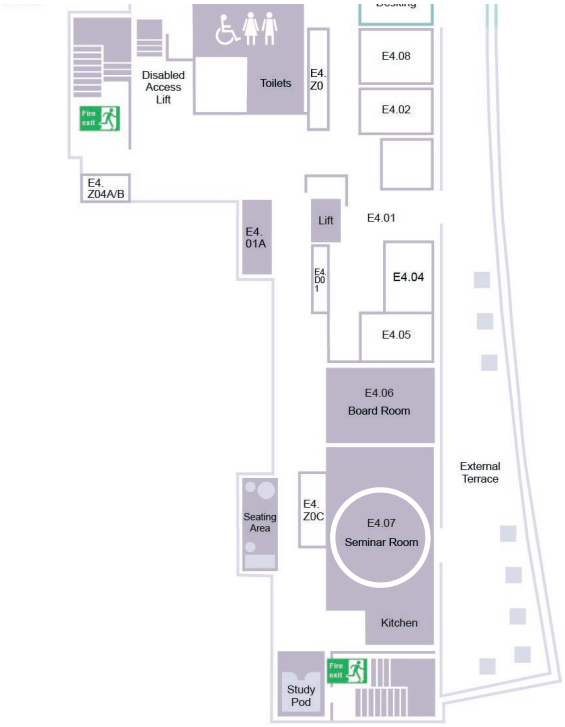


Institute of Genetics and Cancer
Cancer Research UK Edinburgh Centre South Seminar Room S1.14

First Floor
South



Institute of Genetics and Cancer



Assessment Guidelines
for all students

GRADUATE RESEARCH & TRAINING

HANDBOOK 2026

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 using the QR code below:

Assessment reviews and EUCLID

All students need to complete assessment reviews and upload these onto EUCLID to be signed off by your supervisors and the postgraduate director. Over the course of your studies, you will complete assessment reviews at the 10-week stage, the first year stage, the second year stage 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 year) or an additional meeting (e.g. at the end of the third year of three year funded PhD) would be helpful.

You should have at least one assessment review during each year of your studies.

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			✓	

We will send out the hand-in dates of these assessments to all students when they commence their studies.

Each assessment review will involve (i) the student writing a report of their progress to date and future plans (your scientific report) (ii) a meeting with the student's thesis committee or postgraduate director to discuss progress and future plans (iii) the student receiving written feedback on their progress and future plans (feedback report).

Once the student has completed their assessment review and has a copy of both their scientific report and the feedback report, the student should upload both their scientific report and the feedback report to EUCLID. If your scientific report document is too large to be uploaded to EUCLID, you will need to compress the file to an acceptable size. The EUCLID assessment form contains a number of questions, if these questions are already addressed in the uploaded scientific or feedback reports, then the information does not need to be re-entered on the EUCLID form, simply enter “See uploaded documents” into the EUCLID form. The EUCLID assessment form will automatically be sent to your supervisors and postgraduate director for sign-off. If your EUCLID submission does not contain an uploaded copies of both your scientific report and the feedback report, it will not be approved and will be returned to you for completion.

EUCLID will open once during each year of your studies to record your assessment reviews for that year. Therefore if you have more than one assessment review during the year, please upload all documents for those reviews at that time. Students on 3 year or 4 year PhD programmes are likely to have both their 10 week and 1st year assessment reviews in their first year and can upload all documents for both these assessment reviews to EUCLID together after their 1st year review. For students on any programme, any documents from interim or additional thesis committee meetings can also be uploaded onto EUCLID at the same time as your annual assessment review documents.

Students on the MRC HGU rotation programme will have their 10 week assessment review approximately 10 weeks after starting their chosen PhD projects (i.e. typically around 8-9 months after starting their studies).

10 week report assessment

At this stage, a meeting should be organised with your thesis committee and a thesis report collated which should be concise (1000 words excluding title, references, abstract or figure legends). At the meeting, the student should carry out a short (10-15 min) presentation outlining what they have done so far and their broad project plans to the thesis committee. Again, we are more interested in ensuring the student has settled in well and has a clear plan for what to do in their first year. Various training opportunities can also be discussed.

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).

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 (organised by students): 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 (decided by yourself and the principal supervisor) and a Chair. The external may be from the same building, but should be independent of the supervisors. The Chair will be decided by PGR Conveners at the start of your studies. For MScR and MD the roles of the chair and external are often combined.

The chair should be experienced and independent to your studies, they will be selected by the PGR team. The external can be an expert on the project area and can be recruited by the supervisor and student, but it shouldn't be someone from the supervisors group. This is the same for the second supervisor.

Scan QR code for Thesis Committee Guidance below:

General Information

GRADUATE RESEARCH & TRAINING

HANDBOOK 2026

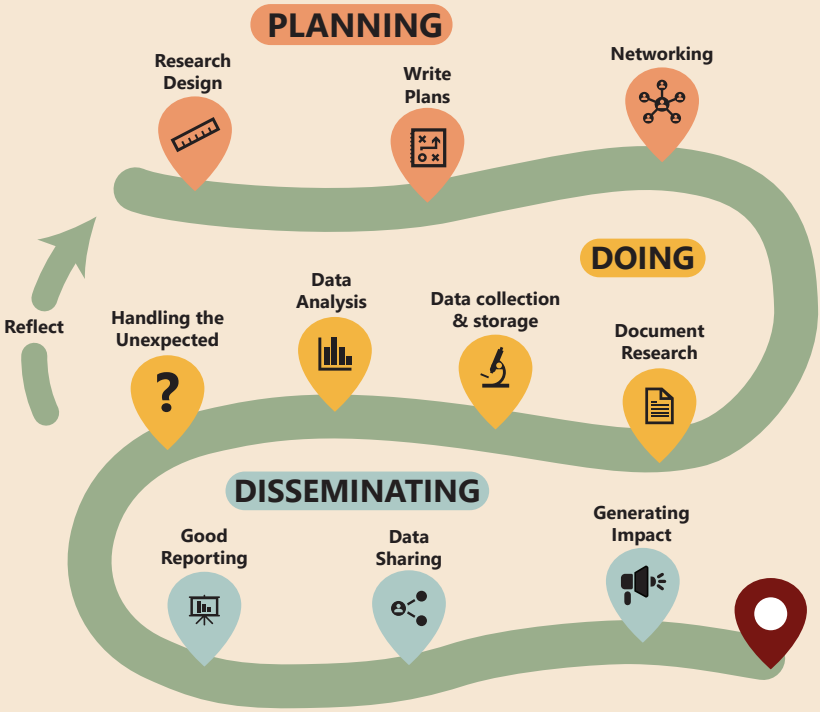
Good Research Practice Roadmap

Research Culture & Citizenship

Training & Skills Development

Ethics and Integrity

Open Research



This good research roadmap is designed to help researchers, particularly early career researchers, learn about and integrate good research practices into their work. It contains core themes relevant to all research stages, as well as stage-specific themes relevant to the planning, doing and dissemination of your work.. Use the QR code to access detailed explanations of each theme, including guidance on implementing good practices, and links to additional resources.

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).

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, the institute-wide ceilidh, a careers event, thesis writing workshop and several pub quizzes throughout the year! 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.

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

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) or one of the postgraduate conveners (Kevin, Ian, Susan, Duncan) 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.

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

Register with a GP/doctor

It is important to make sure that you look after yourself whilst studying, both physically and mentally, and that you know how to get medical assistance if you need it.

The University has provided information about registering with your nearest doctor, known as a General Practitioner (GP).

Further information can be found on the New Students website here:

https://www.ed.ac.uk/students/new-students/ready-university/top-6-tasks/register-doctor

THE UNIVERSITY of EDINBURGH

KEY BENEFITS OF A PUBLIC PGR PURE PROFILE

Recording your publications and research activities in Pure, will help you enhance the **visibility** of your research via the Edinburgh Research Explorer portal...

...which offers you a search engine optimised repository to increase **discoverability**...

...**share** your research, area of expertise and scholarly activities...

...while attracting **collaboration** with researchers from external institutions worldwide...

...gaining **recognition** of your research contributions....

... and giving you the opportunity to engage with research communities and build your **network**.

You can find further guidance and the public profile request form here: https://edin.ac/3Ssph7O

MENTAL HEALTH DURING YOUR PhD

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

47%

A study by the University of California, Berkeley, found nearly half of postgraduate students met criteria to classify them as depressed.[1]

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

IMPOSTER SYNDROME

7 in 10 people experience imposter syndrome throughout their careers[2]

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 no where 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/departmental 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

55% of PhD students are concerned about work-life balance[3]

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.[4] This is a fault with the system. Presenteeism is a common trait observed in academia, where peole 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 200 PhD graduates become full professors.[5] 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 JO@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 intervention 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 lack 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 (for example) may be a positive change you can make.

REQUEST COUNSELLING

Universities have counselling services that are available for you to use. This can be useful to help talk through your problems and also make sure 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 discussions to reduce workload temporarily or take time out.

TALK TO YOUR PEERS / POSTDOCS

It is highly likely that people around you have also experienced the stressful nature of a PhD. Reach out, if you feel able. Remember, postdocs have survived their PhD so may have some useful tips / 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. A good tip is to aim to get into the lab first thing in your day, before looking at other tasks like emails.

READ LITERATURE

There are a large amount of online resources available to help manage mental health and wellbeing. For example, the charity Mind, have a range of booklets available, from how to manage stress, to how to support others with mental health problems.

As a final note, if your PhD is heavily affecting your mental health and wellbeing, there is absolutely no shame in quitting and starting a new chapter in your life. There are plenty of successful people that quit their PhD! Put it this way - you might put up with an ill-fitting t-shirt for a while, but you wouldn't wear it for 4 years!!

REFERENCES

1. Graduate Student Happiness & Well-Being Report, 2014, University of California, Berkeley.
2. Sakulku, J. The Impostor Phenomenon. The Journal of Behavioral Science, 6(1), 75-97.
3. Graduate survey: A love–hurt relationship, Nature 550, 549–552
4. Guthrie, 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.

#TIMETOTALK
#RSCPOSTER

*ALTHOUGH I DID SURVIVE A PhD

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

igc INSTITUTE OF GENETICS & CANCER

UKRI Medical Research Council | THE UNIVERSITY of EDINBURGH | CANCER RESEARCH UK

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

igc INSTITUTE OF GENETICS & CANCER

UKRI Medical Research Council | THE UNIVERSITY of EDINBURGH | CANCER RESEARCH UK

Mental Health First Aiders

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

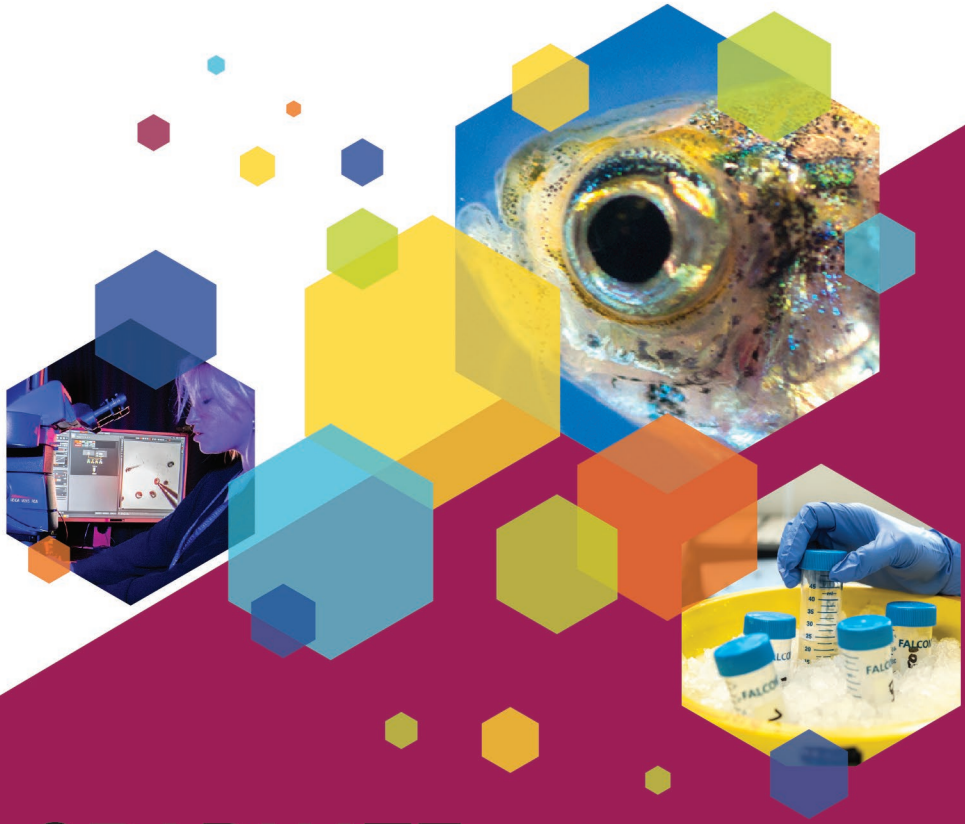
The IGC Mental Health First Aid team are here to help!

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 2026

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://registryservices.ed.ac.uk/academic-services/students/code-of-practice

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/

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

Research Ethics

General
www.pnas.org/content/86/23/9053.full.pdf

Image manipulation
www.jci.org/articles/view/21717/pdf

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

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/

The use of AI
https://www.ed.ac.uk/ai

MRC Human Genetics Unit 4 Year Programme

- Introduction to programme
- Projects available
- Rotation timeline

GRADUATE RESEARCH & TRAINING

HANDBOOK 2026

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 Ian Adams 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

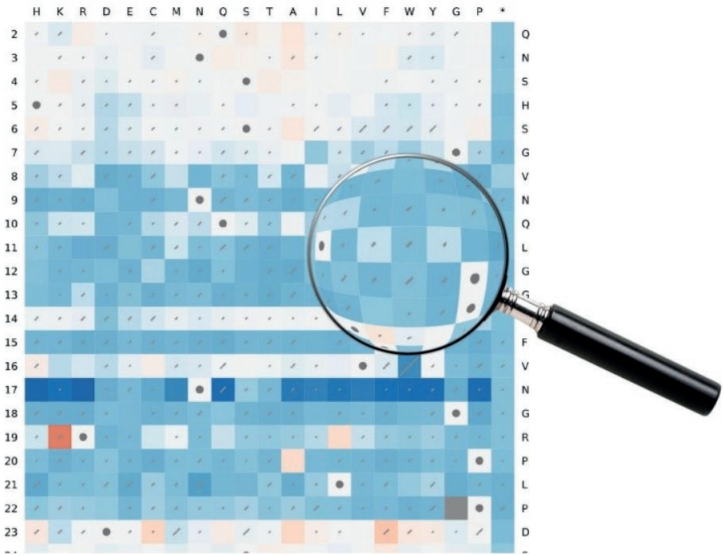
Supervisors will ask to meet up with you if there are any concerns.

Gene engineering with protein barcodes

Supervisor: Prof Grzegorz Kudla

Although genome sequencing is now routinely done in the NHS, diagnosis can be difficult because it is often unclear which mutations disrupt gene function and which are harmless. Our lab tries to solve this with Multiplex Assays of Variant Effects (MAVEs): experiments in which we construct thousands of mutated versions of a gene, put them into cells, and measure how each mutation affects the gene's function. We apply this approach to disease genes (for example, in insulin resistance, Parkinson's disease, and rare developmental diseases) and to genetic engineering, where we study how mutations change gene expression and RNA splicing.

MAVEs depend on being able to screen thousands of variants in one experiment, which is typically done using DNA barcodes. However, many mutations act at the protein level, for example by changing protein abundance or protein binding, and these effects are hard to capture with a DNA-level readout. To study them, we aim to develop a protein barcoding technology for MAVEs. Starting with a library of Green Fluorescent Protein genes available in the lab, you will design a collection of protein barcodes, integrate them into our genes, and analyse the results with quantitative mass spectrometry. The outcome will be a proof-of-concept protein barcode library and an assessment of how reliably the barcodes can be detected and quantified.



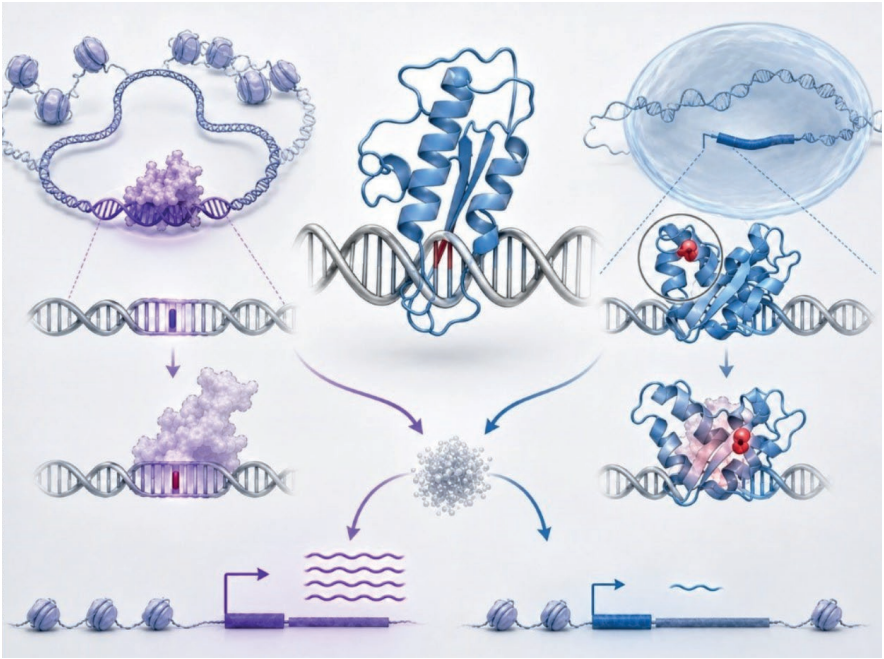
Decoding the Effects of Genetic Variation on Transcription Factors

Supervisors: Prof Joe Marsh

Transcription factors sit at a critical interface between the genome and gene regulation. Genetic variants can disrupt their function in many different ways: by changing the transcription factor protein itself, altering its interactions with DNA or other proteins, or modifying the regulatory sequences to which it binds. Understanding these effects is important for explaining how both coding and non-coding variants contribute to human disease.

In this rotation, you will investigate how genetic variation alters transcription factor function and gene regulation. The specific project can be tailored to your interests and background, and could involve analysing disease-associated coding or regulatory variants, using protein and DNA language models and other AI-based approaches to predict molecular effects, investigating large-scale functional datasets from multiplexed assays of variant effect, or integrating protein structure, human genetics and population data to identify functionally important regions.

Projects can range from primarily computational to close integration with experimental data and collaborators, and no previous computational experience is required. More broadly, the rotation will provide experience with emerging approaches for connecting variation across the genome to molecular mechanisms, gene regulation and human disease.

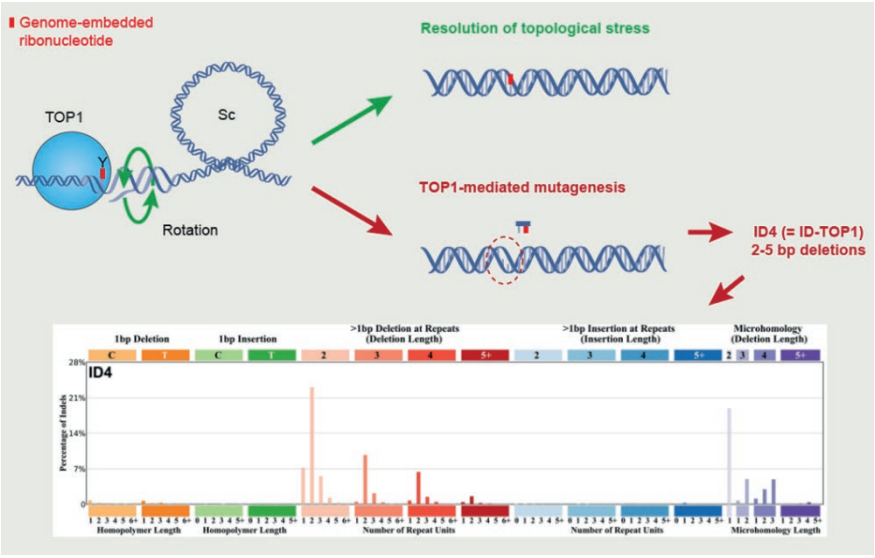


Understating mutagenesis in somatic cells

Supervisor: Prof Andrew Jackson & Dr Martin Reijns

Mutations drive phenotypic changes that underlie evolution, genetic disease and carcinogenesis. Understanding how, where and when changes to the DNA sequence occur is therefore important. Experiments that address this most commonly make use of highly proliferative cells, cultured in the lab. However, in relation to carcinogenesis it is of particular importance to understand the process of mutagenesis in somatic cells, which generally undergo no or limited cell divisions. We recently established the mechanism for an indel mutational signature that occurs in cancer and the germline. We showed this to be due to genome-embedded ribonucleotides and the action of topoisomerase 1, an enzyme that relieves topological stress, with mutations of this type more common in genes that are highly expressed, providing a basis for transcription-associated mutagenesis (Reijns, Parry, Williams et al. Nature, 2022).

Since this discovery, it has become clear that this signature also occurs in somatic cells, such as neurons. However, as ribonucleotides are largely thought to be incorporated during DNA replication in S-phase, our ongoing work aims to understand the similarities and differences in the mechanism that cause these mutations in non-dividing cells. Possible projects relating to this will involve a range of cell and molecular biology techniques, including flow cytometry, targeted DNA capture and short/long-read NGS, as well as bioinformatics analyses. Our investigations range from using cultured mammalian or yeast cells that are forced out of cycle to mouse models to study this in neurons and hepatocytes. Ultimately, we aim to understand how the interplay between DNA damage/repair, genome-embedded ribonucleotides and topological stress at highly transcribed genes may cause mutations in somatic cells, with implications for cancer and ageing.



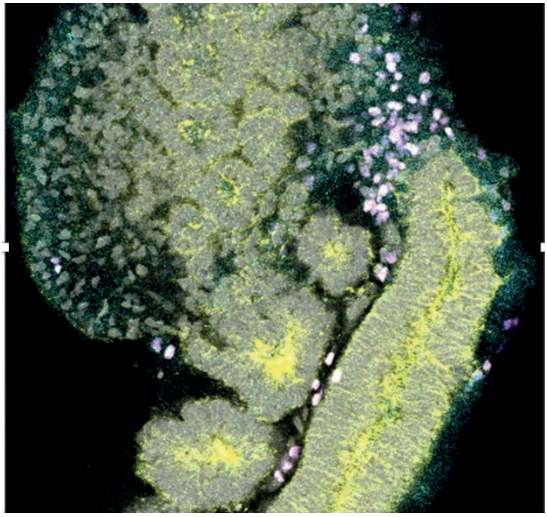
The Orgins of congenital melanocytic nevi using advanced human gastruloids

Supervisor: Prof Liz Patton, Dr Takuya Azami & Prof Martin Taylor

Melanocytes evolved to produce melanin — a UV-absorbing polymer that generates the diverse colour spectrum of our hair, eyes, and skin — and also have other roles, including in immune, auditory, heart and neuroendocrine systems that remain underexplored. Melanocyte biology has shaped human evolution and early human migrations by safeguarding skin health and supporting vitamin production. Skin colour remains highly relevant to the individual human experience, shaping societies worldwide. Melanocytes are an ideal model for studying cellular differentiation, as multiple lineages converge on a common cellular state defined by expressing the melanin biosynthetic pathway.

In collaboration with Martin Taylor (HGU), Takuya Azami (IGC) and Tatjana Sauka-Spengler (Stowers Institute), our team is generating first-of-its-kind lineage trajectories of zebrafish and human melanocytes, spanning embryonic to adult stages, using a zebrafish experimental model, human embryonic stem cells (hESCs) and gastruloids. Our cell-hierarchy maps will reveal how multipotent embryonic cells make fate decisions to become specified adult melanocytes that populate diverse tissues, and may guide therapeutic strategies for melanocytic diseases, including congenital melanocytic nevi and melanoma.

For this PhD project, the student will work closely with Dr Azami (Day-to-day supervisor) and Dr Patton to develop new human models of the melanocyte lineage origins and diversity using advanced cellular and gastruloid systems. For the first time, our lab has established melanoblast, precursor of melanocytes, in these gastruloids and are including trackable fluorescent reporters. Integrating high reproducibility and predictable gene expression patterns in these models and 2D melanocytes differentiation system from hESCs make them ideal for rapid screening of morphological phenotypes following different treatments, culture conditions, or genetic mutations, especially those that lead to congenital melanocytic nevi. These models offer a long-awaited opportunity to investigate previously inaccessible stages of post-implantation human development to understand the origins of congenital melanocytic disease.

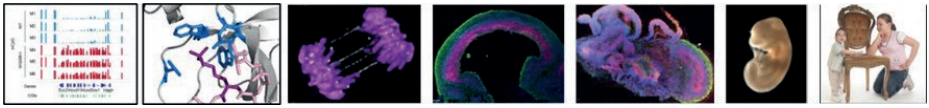


Stem cells in growth and ageing

Supervisor: Prof Andrew Jackson

Our lab works on single gene human disorders with extreme phenotypes, including the smallest people in the world. As well as discovering new genes, we are interested in what these conditions can tell us about biological and common disease processes. To do so we use multiscale approaches including biochemistry, informatics, cell biology and animal models to discover new pathways and processes.

We have made recent discoveries which link epigenetics and stem cell function with growth and ageing (Sarni et al Nat Genet 2026). We are developing cellular models to understand these links, and the mechanism by which DNA methylation alters stem cell output, that may shed light on stem cell plasticity, and exhaustion, with translational relevance to regenerative medicine.



Dissecting DNMT3B functions in Immunodeficiency-centromeric instability facial anomalies syndrome

Supervisors: Dr Duncan Sproul & Dr Hannah Long

Mutations in the DNA methyltransferase DNMT3B cause the recessive genetic condition Immunodeficiency centromeric instability facial anomalies syndrome type 1 (ICF1). DNMTs deposit the repressive epigenetic mark DNA methylation on the genome. However, it is challenging to connect altered ICF1 DNA methylation patterns to disease phenotypes because DNMT3B recruitment is mediated by multiple pathways.

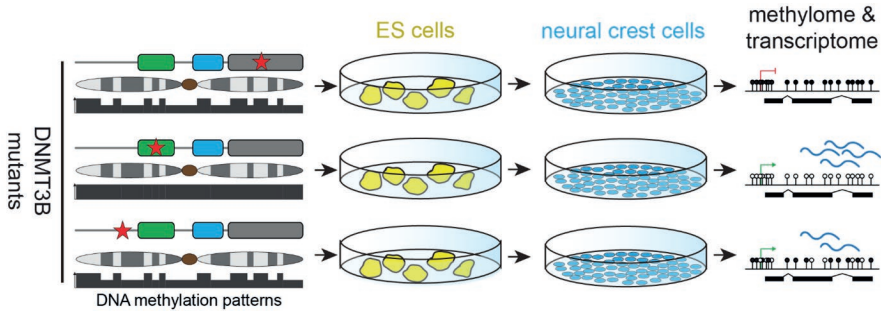
This project builds on our recent delineation of a pathway recruiting DNMT3B to constitutive heterochromatin (Taglini et al, 2024) to dissect the role of different DNMT3B functions in ICF1. We will generate human embryonic stem cell (hESC) lines with separation-of-function mutations targeting different DNMT3B recruitment pathways before characterising their epigenomes and transcriptomes compared to those with ICF1 mutations. As ICF1 is characterised by craniofacial phenotypes, we will also determine how these mutations impact differentiation of hESCs to cranial neural crest cells – a key cell type forming facial structures during development (Prescott et al, 2015; Long et al, 2020). This project lays the groundwork to understand the molecular basis of ICF1 patient phenotypes and ultimately design therapies. The major techniques used are: hESC culture, genome editing, Nanopore sequencing, CUT&Run, scRNA-seq and bioinformatics.

References:

Long et al 2020, https://doi.org/10.1016/j.stem.2020.09.001

Prescott et al 2015, https://doi.org/10.1016/j.cell.2015.08.036

Taglini et al 2024, https://doi.org/10.1038/s44319-024-00061-5 may cause mutations in somatic cells, with implications for cancer and ageing.



Investigating enhancer-associated RNA production at a craniofacial disease locus

Supervisor: Dr Hannah Long, Dr Nicola Carruthers & Dr Kirtsy Uttley

Rotation project only available during first semester

Many Mendelian disorders and complex disease traits, such as facial shape, are associated with DNA sequence changes in the non-coding, so-called 'dark' genome, often impacting regulatory switches called enhancers that are bound by transcription factors and turn gene expression on and off during development (Long et al, 2016). Human facial appearance is extremely diverse and sensitive to genetic perturbation – with around one third of birth defects exhibiting a facial difference. In the lab, we investigate gene regulatory mechanisms in facial progenitor cells called cranial neural crest cells (CNCCs) that we can differentiate in vitro from human embryonic stem cells – with a focus on how genetic alterations can shape facial differences in the population, drive evolutionary divergence and in extremes cause disease.

There are many exciting projects available in the lab on the themes of gene regulation, 3D genome organisation, craniofacial development and human genetic disease. In one project, we will explore the role of enhancer transcription in long-range gene regulation and disease. We will focus on a cluster of enhancers that are ablated in patients with an isolated craniofacial malformation called Pierre Robin sequence (PRS) (Long et al, 2020), and where we have observed ancient hominin variants can drive increased regulatory activity (Uttley, Jüllig et al, 2025, Development). Using a method called transient transcriptome sequencing (TT-seq), we have recently observed transcription from the two extreme long-range enhancer clusters at this locus in addition to a longer RNA transcript that extends >200 kb towards the SOX9 gene. Enhancer RNAs (eRNAs) have been shown to play important roles in enhancer-mediated gene regulation, with growing implications in disease, though their function remains poorly understood (Wang et al, 2026). Longer non-coding RNAs have also been implicated in gene regulation (Viswanathan et al, 2026). In this project we will confirm the cell-type specificity of the non-coding RNA (ncRNA) transcripts produced at the PRS disease locus, and explore their role in long-range gene regulation. We will examine RNA dynamics across the differentiation using digital PCR, and explore their function through genetic ablation and transient depletion. We will complement this with overexpressing ncRNA transcripts and examining effects on SOX9 expression. Leveraging edited cell lines in the lab, we can explore how alterations to the SOX9 locus impact ncRNA transcription. Together, this project will provide new insights into the role of transcription in long-range enhancer-mediated gene regulation and could provide new insight for understanding facial shape differences in the population, across evolution and in disease.

Technically, this project will involve culturing and differentiating human embryonic stem cells and cranial neural crest cells, genome engineering and imaging, RNA purification and molecular biology techniques such as digital PCR. Bioinformatic analyses will include analysis of available TT-seq data to identify other putative long enhancer-associated transcripts genome-wide in CNCCs.

References

Long HK, Prescott SL and Wysocka J. (2016) 'Ever-Changing Landscapes: Transcriptional Enhancers in Development and Evolution', Cell, 167(5), pp. 1170–1187.

doi: https://doi.org/10.1016/j.cell.2016.09.018

Long HK et al. (2020) 'Loss of Extreme Long-Range Enhancers in Human Neural Crest Drives a Craniofacial Disorder', Cell Stem Cell, 27(5), pp. 765-783.e14.

doi: https://doi.org/10.1016/j.stem.2020.09.001

Uttley K, Jüllig HJ, De Angelis C, Auer JMT, Ozga E, Bengani H, Long HK (2025) 'Neanderthal-derived variants increase SOX9 enhancer activity in craniofacial progenitors that shape jaw development'. Development 152 (21): dev204779.

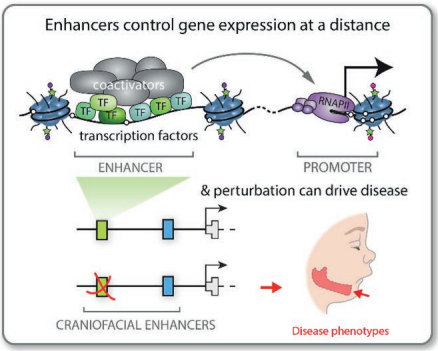
doi: https://doi.org/10.1242/dev.204779

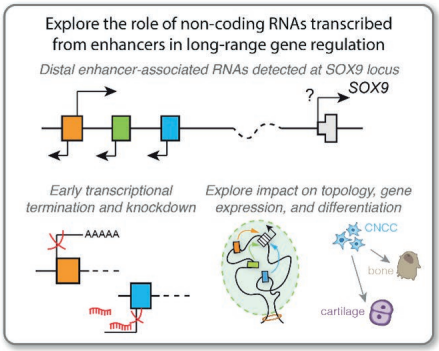
Wang Q., ten Dijke P and Fan C. (2026) 'Deciphering the regulatory landscape of enhancer RNAs in health and disease'. Sig Transduct Target Ther 11, 29.

doi: https://doi.org/10.1038/s41392-025-02436-z

Viswanathan K, Shaw P, Kiran M (2026) 'Mechanistic insights into the transcription-mediated roles of non-coding RNAs in gene regulation', Biochimica et Biophysica Acta (BBA) - Gene Regulatory Mechanisms,195169, ISSN 1874-939

doi: https://doi.org/10.1016/j.bbagrm.2026.195169





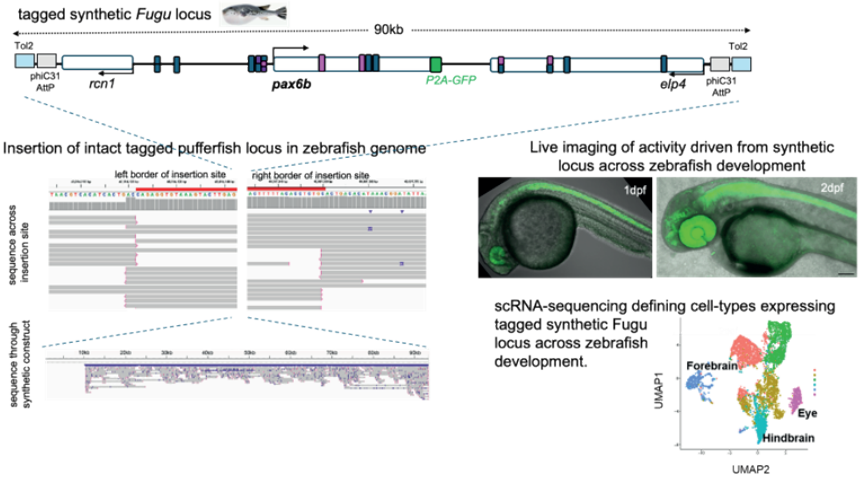
Dissecting complex regulatory landscapes in the non-coding genome

Supervisor: Prof Wendy Bickmore & Dr Shipra Bhatia

Genes with pleiotropic roles in development often reside in large, complex regulatory regions of the non-coding genome. Multiple enhancers in these regions can operate over huge genomic distances, ensuring precise spatial and temporal control of target gene expression. Disruption of enhancer-mediated gene regulation has been implicated in human disease. Our understanding of these disease mechanisms is hampered by the lack of information about the precise cell-type specific roles of each enhancer, as well as the rules defining the order of enhancers in the locus.

We address this knowledge gap by developing in vivo and ex vivo models to uncover the cell-type specific roles of developmental enhancers using PAX6 as a paradigm locus (Bhatia et al 2021, Uttley et al 2023). We combine high resolution live imaging, single cell omics and synthetic biology techniques in zebrafish and optic cup organoid models to define the cell-type specific functions of individual enhancers in the PAX6 locus.

The proposed project will focus on specific components of this effort, particularly the consequences of loss/re-arrangement/replacement of PAX6 enhancers in large synthetic constructs bearing the complete PAX6 regulatory domain. We have generated zebrafish transgenic lines bearing 90kbp synthetic bacterial artificial chromosomes (BACs) representing a compressed version of complete PAX6 regulatory domain. Selecting the cells where reporter gene expression is driven from these synthetic constructs, we have generated single cell resolution CAGE (sc5'RNA-sequencing) and accessible chromatin region (scATAC-sequencing) datasets. The rotation project will focus on analysis of these datasets to generate the first ever gene regulatory map for a synthetic regulatory landscape at single cell resolution in vivo. The subsequent PhD project will focus on uncovering the functional significance of these regulatory networks in establishing cellular identities.



Linking mutations in the chromatin remodeller ATRX to inflammation

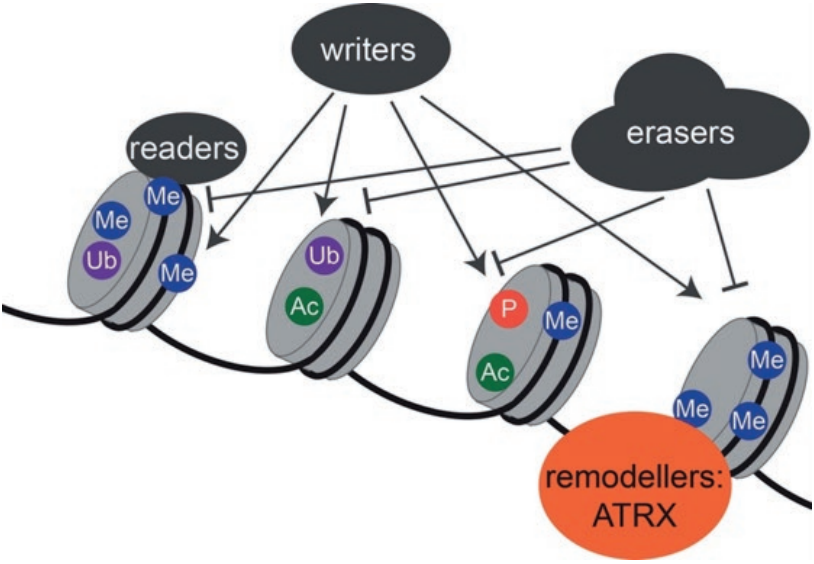
Supervisor: Dr Rebekah Tillotson & Prof Yanick Crow

Rare diseases are classified as those affecting <1 in 2,000 people. Collectively, they affect 1 in 17 people born in the UK.

The Tillotson laboratory studies a class of rare monogenic disorders caused by mutations in genes encoding chromatin factors. Chromatin factors write/read/erase epigenetic marks or remodel chromatin structure to regulate chromatin state and control transcription. The so-called “chromatinopathies” affect multiple tissues, always including the brain. Currently the Tillotson group is focused on a chromatinopathy called ATR-X syndrome, caused by hypomorphic mutations in the X-linked gene ATRX.

The Crow group studies rare monogenic disorders associated with inappropriate stimulation, or defective negative regulation of the type I interferon system. Recently, Crow’s team have discovered patients with ATR-X syndrome who also demonstrate type I interferon induced autoinflammation.

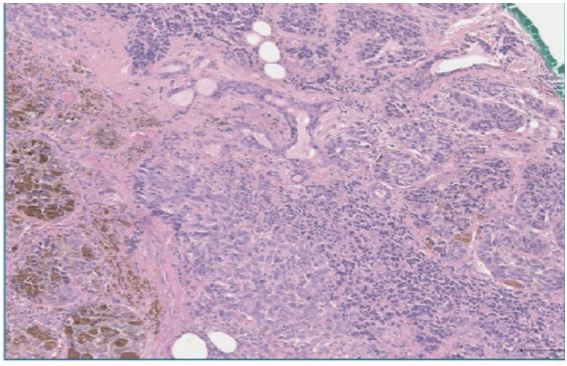
We are looking for a PhD student to investigate the mechanism underlying this newly discovered and unexpected link between ATRX dysfunction and innate immune system engagement. The project will use cellular and mouse models as well as patient-derived material, and involve experimental techniques such as CRISPR/Cas9 editing, transcriptomics (RNA-sequencing and qPCR), analysis of chromatin binding patterns, co-immunoprecipitation, western blotting, fluorescence microscopy, mouse phenotyping (neurobehavioural testing and pathology), as well as bioinformatic analysis.



What causes mucosal melanoma?

Supervisor: Prof Liz Patton

Melanoma in the skin is a deadly cancer caused by UV-light, but little is known about the origins and causes of melanoma in other non-UV light exposed cancers. While therapy has advanced rapidly for skin melanoma, there are no long-term effective therapies for rare melanomas, including mucosal melanoma in the nasal and oral cavities. Our lab has recently shown that canine oronasal mucosal melanoma is the same disease as in humans and have an ongoing canine clinical trial as a new companion species for drug discovery and treatment. Unlike in humans, where oronasal mucosal melanoma is a rare disease, it is common in the dog, providing an important cross species comparative oncology opportunity to discover the causative factors for oronasal mucosal melanoma in humans. In this PhD project, we will use single cell genomic technologies, spatial transcriptomics, and mucosa organoids in human and dogs to understand the origins and cause of mucosal melanoma. This work has the potential to help us understand how to prevent mucosal melanoma and may reveal new therapeutic targets that can be developed in our human and canine models



Understanding how cells protect replication forks

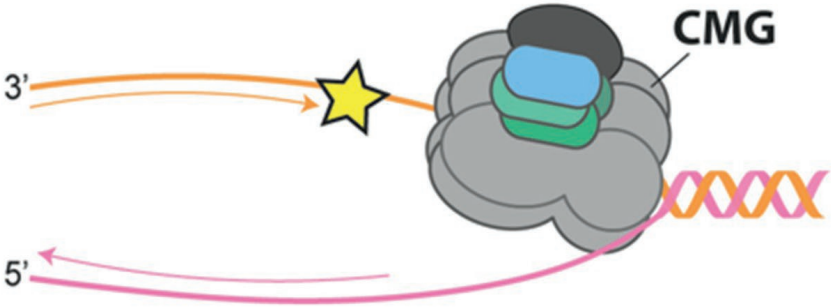
Supervisor: Dr Tom Deegan

ADNA replication stress is a major source of genome instability and is particularly relevant to cancer, where both the causes and consequences of replication stress can be amplified. Cells have evolved sophisticated mechanisms to stabilise and protect DNA replication forks when replication is perturbed, but many of the molecular mechanisms that control these responses remain poorly understood.

Our lab is interested in understanding how replication forks respond to stress, and how their structure and behaviour are regulated. In this project, we will take a biochemical approach, using reconstituted human replisomes in vitro to investigate the mechanisms that control replication fork stability and remodelling.

The aim is to develop a mechanistic understanding of these processes in a defined system, allowing us to ask questions that are difficult to address in cells. The project will focus on how key replication and DNA repair factors are recruited and regulated at stressed replication forks, with a particular interest in the role of RAD51 and fork remodelling.

This is an exciting opportunity to work on a relatively unexplored area of replication biology and to contribute to a broader effort in the lab to understand, and ultimately reconstitute, complex replication fork responses in vitro. The work will be relevant to fundamental questions in genome stability and may also provide insight into why defects in replication fork protection are strongly associated with cancer.



Mining the dark matter of the tumour genome: loss of heterozygosity in ovarian cancer

Supervisor: Prof Colin Semple & Dr Stuart Aitken

TFigure 1: The complex genome of HGSOC tumours. Structural variants are abundant across the tumour dataset (N=324), reflected in the frequencies of CNA variants (top panel), as well as translocations and complex events such as chromothripsis. Samples also show frequent whole genome duplication (WGD) across tumour stages and cohorts. Pathogenic germline variants are relatively enriched in known HGSOC susceptibility genes as expected.

Loss of heterozygosity (LOH) events are an established hallmark of tumour genomes, occurring when a cell that is originally heterozygous at a locus loses one of its two alleles at that locus, by any of three mutational mechanisms (Dutta and Schacherer, 2025). This can happen by simple deletion of one allele only (CN-loss LOH), by deletion of one allele accompanied by duplication of the remaining allele (CN-neutral LOH), or deletion accompanied by multiple duplications of the remaining allele (CN-amplification LOH). These events are common in tumour genomes, and are a major influence on their evolution, as well as a potential source of vulnerabilities that may underly new cancer treatments (Nichols et al, 2020).

LOH events are usually not studied in sufficient detail to discover the frequencies of CN-loss, CN-neutral or CN-amplification in a tumour type, or whether there have been functional impacts as a result, since ideally this requires substantial tumour cohorts with whole genome and transcriptome sequencing (WGTS) data. We have recently constructed a large (N=324) WGTS tumour cohort for high grade serous ovarian cancer (HGSOC) (Ewing et al, 2025), providing a unique opportunity to study the origins and impacts of LOH events at scale, in this structurally complex cancer of unmet need.

The student undertaking this project will gain skills in cancer genome bioinformatics.

Aims

1. Compile LOH regions and copy number alteration (CNA) calls from our WGTS cohort.

2. Use CNA calls to define the mutational mechanisms underlying LOH in HGSOC.

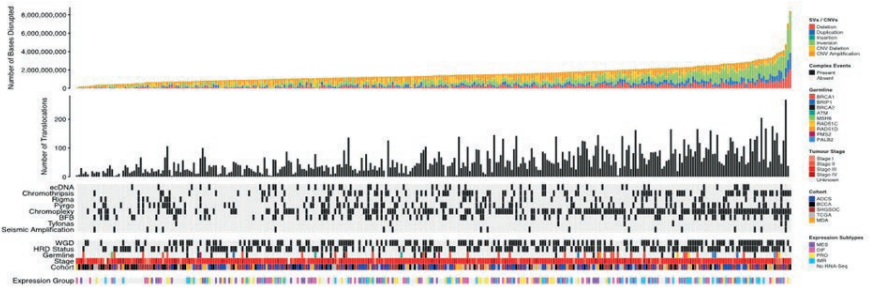
3. Assess effects on gene expression within LOH segments for cancer driver and bystander genes.

References

Dutta A, Schacherer J. The dynamics of loss of heterozygosity events in genomes. EMBO Rep. 2025 Feb;26(3):602-612.

Nichols CA, Gibson WJ, Brown MS, Kosmicki JA, Busanovich JP, Wei H, Urbanski LM, Curimjee N, Berger AC, Gao GF, Cherniack AD, Dhe-Paganon S, Paolella BR, Beroukhim R. Loss of heterozygosity of essential genes represents a widespread class of potential cancer vulnerabilities. Nat Commun. 2020 May 20;11(1):2517.

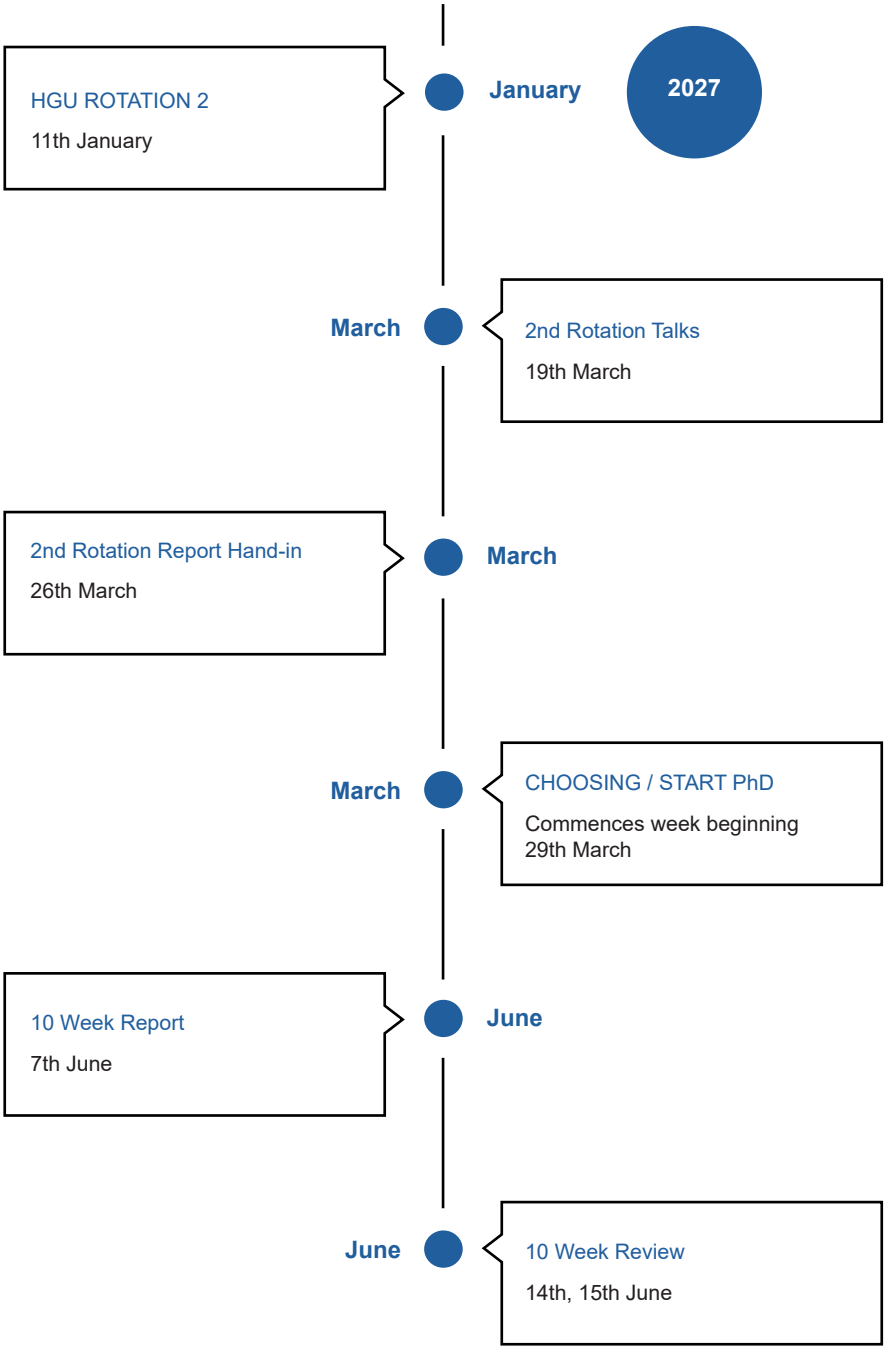
Ewing A, Meynert A, Silk R, Aitken S, Bendixsen DP, Churchman M, Brown SL, Hamdan A, Mattocks J, Grimes GR, Ballinger T, Hollis RL, Simon Herrington C, Thomson JP, Sherwood K, Parry T, Esiri-Bloom E, Bartos C, Croy I, Ferguson M, Lennie M, McGoldrick T, McPhail N, Siddiqui N, Glasspool R, Mackean M, Nussey F, McDade B, Ennis D; Scottish Genomes Partnership; McMahon L, Matakidou A, Dougherty B, March R, Carl Barrett J, McNeish IA, Biankin AV, Roxburgh P, Gourley C, Semple CA. Divergent trajectories to structural diversity impact patient survival in high grade serous ovarian cancer. Nat Commun. 2025 Jul 1;16(1):5586.


Number of Bases Disrupted
6,000,000,000
6,000,000,000
4,000,000,000
2,000,000,000
0
Number of Translocations
200
100
0
ecDNA
Chromothripsis
Rigma
Pyrgo
Chromoplexy
BFB
Tyfonas
Seismic Amplification
WGD
HRD Status
Germline
Stage
Cohort
Expression Group
SVs / CNVs
Deletion
Duplication
Insertion
Inversion
CNV Deletion
CNV Amplification
Complex Events
Present
Absent
Germline
BRCA1
BRIP1
BRCA2
ATM
PALB2
RAD51C
RAD51D
FANCI
FANCM
Tumour Stage
Stage I
Stage II
Stage III
Stage IV
Unknown
Cohort
ACCS
BCCA
OCTANE
TCGA
MDA
Expression Subtypes
MES
IMM
PRO
IMR
No RNA-Seq

HGU MRC DTP Rotation ProjectTalks
Project Pitches

Tuesday 22nd September - Room S1.16	
09:00 - 09:30	Prof Grzegorz Kudla - Gene engineering with protein barcodes
09:30 - 10:00	Prof Joe Marsh - Decoding the Effects of Genetic Variation on Transcription Factors
10:00 - 10:30	Dr Martin Reijns- Understanding mutagenesis in somatic cells
11:00 - 11:30	Prof Liz Patton - What causes mucosal melanoma?; The origins of congenital melanocytic nevi using advanced human gastruloids
11:30 - 12:00	Prof Martin Taylor - TBD
Thursday 1st October - Room S1.16	
14:00 - 14:30	Prof Andrew Jackson - Stem cells in growth and ageing
15:00 - 15:30	Dr Duncan Sproul - Dissecting DNMT3B functions in Immunodeficiency-centromeric instability facial anomalies syndrome
15:30 - 16:00	Dr Hannah Long - Investigating enhancer-associated RNA production at a craniofacial disease locus
Friday 2nd October - Room S1.15	
13:30 - 14:00	Prof Wendy Bickmore - Dissecting complex regulatory landscapes in the non-coding genome
14:00 - 14:30	Dr Rebekah Tillotson - Linking mutations in the chromatin remodeller ATRX to inflammation
14:30 - 15:00	Dr Tom Deegan - Understanding how cells protect replication forks

Training Timeline 2026 - 2027


HGU ROTATION 2
11th January
January
2027
March
2nd Rotation Talks
19th March
2nd Rotation Report Hand-in
26th March
March
March
CHOOSING / START PhD
Commences week beginning
29th March
10 Week Report
7th June
June
June
10 Week Review
14th, 15th June