

# GENETICS SOCIETY NEWS

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- How genetic linkage was discovered

The Genetics Society News is edited by Margherita Colucci and items for future issues can be sent to the editor by email to [newsletter@genetics.org.uk](mailto:newsletter@genetics.org.uk).

The Newsletter is published twice a year, with copy dates of July and January.



## A word from the editor

# Welcome to Issue 85



Welcome to the latest issue of the Genetics Society Newsletter!

In this issue, “change” is the keyword.

Through a series of interviews, we explored the changes from their undergraduate role and the career evolution of the past years Summer Studentship grant winners. Where are they now? What impact that research experience had on them? Find out more in “Genetics Society Summer Studentship - Share your story, Part 1”, page 23.

Big changes happened in the Society too. While we say goodbye to our previous president, Laurence Hurst, and thank him for all his great work and dedication to the society, we have welcomed our new President, Anne Ferguson-Smith. Anne is the Pro-Vice-Chancellor for Research and the Arthur Balfour Professor of Genetics at the University of Cambridge. She is an expert on genomic imprinting and focuses on mammalian developmental geneticist and epigeneticist. Please, go to page 22 to know more about her and what she hopes to achieve over the next 3 years.

Finally, there are aspects in research and, more generally, in the scientific

world that cry for change: with Dr Stuart Ritchie, we explore misconduct and fraud in science and the solutions that “open science” proposes, talking about his latest book “Science fictions: how fraud, bias, negligence and hype undermine the search for truth”.

I would like to draw your attention to the opportunity of contributing to the special issue of Heredity. In July 2022, this special issue will be celebrating Mendel’s 200th birthday with short essays, reviews and research articles on “exceptions” to Mendel’s laws. We would like to invite you to submit your piece (more information on page 20).

Looking forward to this special issue, we propose “How genetic linkage was discovered” by Antonio Marco (page 39): focusing on Mendel’s laws of heredity, this is an overview of the discovery of genetic linkage in which William Bateson, the founder of The Genetics Society, was also involved.

Enjoy!

Best wishes,  
Margherita Colucci

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**The Genetics Society Journals**

*Heredity*

[www.nature.com/hd](http://www.nature.com/hd)

Editor-in-Chief: Prof Barbara Mable

Heredity Editorial Office, University of Glasgow,  
Graham Kerr Building, Glasgow, G12 8QQ, Scotland

*Genes and Development*

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

Editor: Dr Terri Grodzicker

Genes & Development, Cold Spring Harbor Laboratory Press,  
500 Sunnyside Boulevard, Woodbury, New York, 11797, USA

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# Meetings Announcements

More detailed information and links to event websites can be found at [www.genetics.org.uk/events\\_categories/conferences/](http://www.genetics.org.uk/events_categories/conferences/)

## Genetics Society Carer's Award

In recognition of carer's responsibilities, an award of (up to) £60/day will be made available to enable members and selected speakers to attend Genetics Society scientific meetings and events (including virtual events). Awardees can spend this money as they think will best support their attendance. Applications can be made through the mysociety portal.

## Functional Regulatory Genomics and Disease

**Date:** 15-17 November, 2021

**Location:** Hadyn Ellis Building, Maindy Road, Cardiff University, Cardiff

**Website:** [genetics.org.uk/events/functional-regulatory-genomics-disease](http://genetics.org.uk/events/functional-regulatory-genomics-disease)

**Info:** Complex heritable conditions such as obesity, diabetes, alcohol abuse, schizophrenia and depression affect a significant proportion of the UK population.

It has become clear over the past five years that the genetic basis of these diseases does not primarily affect regions of the human genome that code for proteins but may affect the much larger part of the genome, called the regulatory genome, that controls where, when and by how much the expression of genes are turned on and off.

However, compared to protein coding sequences, which make up only 1.5% of the human genome, the regulatory genome remains poorly understood.

Thus, the focus of this conference will be to invite international leaders in the interdisciplinary study of the genetics of disease, the exploration of the regulatory genome and the in-depth biology of its activity and function, to interact and discuss their work.

By doing so it is hoped that we can reach a consensus on how we might coordinate international interdisciplinary efforts to understand the role of the regulatory genome in health and disease.





More detailed information and links to event websites can be found at

**[www.genetics.org.uk/events\\_categories/external-meetings](http://www.genetics.org.uk/events_categories/external-meetings).**

We will happily include any announcements for genetics-based meetings in this section.

Please send any items to **[theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)**.

### **CRISPR and Beyond: Perturbations at Scale to Understand Genomes (Virtual Conference)**

**Date:** 1-3 September, 2021

**Deadlines:** 20 July, 2021 (Abstract and Bursary);  
23 August, 2021 (Registration)

**Location:** Online

**Website:** <https://coursesandconferences.wellcomeconnectingscience.org/event/>

**Info:** The development of CRISPR/Cas-based technologies and DNA synthesis make it possible to modulate genomes with relative ease. These tools can help us understand how genetic variation influences phenotype and thereby answer long-standing questions in biology that impact human health, laying the foundations for precision medicine for heritable diseases and cancer treatment.

This rapid advancement in gene-editing technology enables us to begin to understand the functional implications of natural and disease-related human genetic variation. This year's programme will cover approaches that modulate the genome and its context at scale, from single nucleotides and genes to hundreds of growth environments.

We will discuss (i) assays that focus on individual nucleotides in coding and non-coding regions to understand the effects of single mutations; (ii) focused- and genome-wide randomization methods that assess the influence of changing sequence structure and content; (iii) genome-wide knock-out, knock-down, and upregulation experiments to measure the phenotype when a gene is perturbed; (iv) interaction screens to uncover context-specificity of effects; (v) methods and applications of gene editing (vi) single cell readouts and other emerging technologies. Computational approaches are integral to all these topics, and will be covered by invited speakers, as well as sought for in submitted abstracts.

### **Virus Genomics and Evolution (Virtual Conference)**

**Date:** 13-15 September, 2021

**Deadlines:** 20 July, 2021 (Abstract and Bursary); 6 September, 2021 (Registration)

**Location:** Online

**Website:** <https://coursesandconferences.wellcomegenomecampus.org/our-events/>

**Info:** This meeting will provide a multidisciplinary forum for scientists interested in the genomics and evolutionary analysis

of viruses and will address the fundamental questions of viral origins, transmission and pathogenesis.

Sessions will run online in afternoons for Europe and Africa | mornings for the Americas, enabling participation of evolutionary biologists, bioinformatics and public health practitioners from across the globe.

This year's meeting will focus on virus evolution, epidemics and outbreaks and emerging viral infections and zoonoses. We will also explore new technologies to analyse viruses and large datasets and discuss diagnostic methods to improve public health.

### **The 14th Annual Royal Marsden Breast Cancer Virtual Meeting: Hot Topics in Breast Cancer**

**Date:** 8 October, 2021

**Location:** Online

**Website:** <https://www.royalmarsden.nhs.uk/14th-annual-royal-marsden-breast-cancer-virtual-meeting-hot-topics-breast-cancer>

**Info:** We are pleased to announce that we will be holding the 14th Annual Royal Marsden Breast Cancer Meeting on Friday 8th October 2021.

As we continue to work through the implications of the COVID pandemic on how we all work and meet professionally, we plan to hold the meeting again this year as a virtual event, but with a live panel for both question and answers, and for the popular MDT part of the meeting. We aim to provide an update on what is challenging and controversial in the management of patients with breast cancer, and this year will focus on 3D surface breast imaging, management of oligo-metastases, with updates on optimal cancer genetics testing and recent developments in medical oncology treatments. We will also have a special session on geriatric assessment and what that can mean for improving breast cancer services. The meeting is open to all professionals who are involved in looking after women with breast cancer, including clinicians and senior nurses.

**CRISPR-Cas: From Microbiology to Biomedicine****Date:** 2-4 November, 2021**Deadline:** 10 September, 2021**Location:** Sede Antonio Machado, in Baeza (Andalusia, Spain)**Website:** <https://www.unia.es/oferta-academica/formacion-continua/oferta/item/workshops-current-trends-in-biomedicine-2>**Info:** Biomedicine is a prime concern at the Universidad Internacional de Andalucía (UNIA). In this context, the University continues in 2021 the programme “Current Trends in Biomedicine”, started in 2004.

The purpose of these workshops is to promote and improve international cooperation and scientific exchanges on the Biomedicine field, thus promoting and facilitating scientific interaction, specially between Andalusian researchers and the international scientific community. The conferences will be held over a three-day period at UNIA's head office, Sede Antonio Machado, in Baeza (Andalusia, Spain) and the working language will be English. Sede Antonio Machado is in Plaza Santa María, in the historic city centre, facing the Cathedral. The head office has all the facilities required to host this series of workshops: computer room, library, classrooms, and meeting rooms. It also has a hall of residence where those attending the conferences will be accommodated. Thus, social interaction and informal discussions will be facilitated.

**Bioinformatics for Immunologists (Virtual)****Date:** 27 September - 1 October, 2021**Location:** Online**Website:** <https://coursesandconferences.wellcomeconnectingscience.org/event/>**Info:** This course – organised jointly by Wellcome Connecting Science and EMBL-EBI – will provide participants with an introduction to a range of bioinformatics resources and approaches applicable to immunological research. The resources introduced during the course will cover a variety of data types, from genomic and proteomic data to computational models, biological pathways, and reaction information.

Participants will gain experience of the analysis pipelines for NGS experiments relevant to immunology and will be led through an exploration of this data to identify information of interest. They will also learn how data from several sources can be integrated to provide a wider view of their research, thereby enabling them to be more confident users of their own data and that from public sources.

**Visions III: Star Gazing into the Galaxy of Animal Genetics and Genomics****Date:** 3-4 November, 2021**Deadline:** 10 September, 2021**Location:** Iowa State University Campus**Website:** <https://www.animalgenome.org/share/visions/>**Info:** In keeping with previous Visions conferences hosted by the Animal Breeding, Genetics and Genomics Group at Iowa State University, this conference will explore and discuss the challenges and opportunities that the future provides in animal genetics and genomics. In particular the conference will explore new technologies and the data they offer, their impacts for animal improvement and animal source foods in the developed and developing world and the role scientists play in public acceptance. Talks will be followed by meaningful discussion and audience participation. We encourage students, academics, and industry personnel to attend.**Mitochondrial Medicine – Therapeutic Development (Virtual Conference)****Date:** 30 November - 2 December, 2021**Deadline:** 5 October 2021 (Abstract, Bursary), 23 November 2021 (Registration)**Location:** Online**Website:** <https://coursesandconferences.wellcomeconnectingscience.org/event/>**Info:** Owing to the ongoing situation with covid-19, this Wellcome Connecting Science conference will be organised as a virtually. Mitochondrial disorders have emerged as a major cause of inherited human disease. Although the past decade has seen major advances in our understanding of their genetic basis and the underlying pathology, these findings have yet to translate into new therapies. There is a growing appreciation that new treatments will only emerge through a concerted collaboration between clinicians, laboratory scientists and the life sciences industry, based on a firm understanding of the disease mechanisms. This conference will build new partnerships that harness our understanding of the disease mechanisms, accelerating the pace of effective treatments for mitochondrial diseases. This year's meeting will include pre-clinical models for mitochondrial disease, an update on validation and outcome measures for clinical trials, a discussion on the importance on data sharing for precision medicine, and guidance on regulations for therapeutic development from a range of international agencies.

We will bring together leaders in the field of translational mitochondrial medicine, with a programme designed to engage and inspire the next generation of mitochondrial researchers. The meeting attracts international participants interested in mitochondrial diseases, working in molecular genetics, biochemistry, pathology, and clinical medicine.

More detailed information and links to courses websites can be found at [genetics.org.uk/events\\_categories/training-courses](https://genetics.org.uk/events_categories/training-courses)

We will happily include any announcements for genetics-based or statistical analysis training courses and workshops in this section. Please send any items to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk).

### RNA-seq Data Analysis (Virtual Workshop)

**Date:** 24-27 August, 2021

**Location:** Online

**Website:** <https://genomics.ed.ac.uk/services/rna-seq-data-analysis>

**Info:** RNA sequencing (RNA-seq) is quickly becoming the method of choice for transcriptome profiling. Nevertheless, it is a non-trivial task to transform the vast amount of data obtained with high-throughput sequencers into useful information. Thus, RNA-seq data analysis is still a major bottleneck for most researchers in this field. The ability of correctly interpreting RNA-seq results, as well as knowledge on the intrinsic properties of these data, are essential to avoid incorrect experimental designs and the application of inappropriate analysis methodologies. The aim of this workshop is to familiarise researchers with RNA-seq data and to initiate them in the analysis by providing lectures and practicals on analysis methodologies. In the practicals Illumina-generated sequencing data and various widely used software programs will be used.

### Prenatal Genetics Short Course 2021 (Virtual)

**Date:** 13 September-2 November, 2021

**Location:** Online

**Website:** <https://www.guysandstthomasevents.co.uk/>

**Info:** Recent technological advances are shaping testing and counselling options in prenatal genetics.

This skills-based course is designed to give midwives and other health professionals working in a prenatal testing setting tools to help them to enable patients to access appropriate onwards referral. These include obtaining a family history, identify high risk family histories, approaches to genetic testing, consent taking and counselling skills associated with providing information and results.

The course will provide the background scientific knowledge needed to understand the theory behind the application of genetics in a prenatal setting. The scientific content will be complemented with case-based examples and practical exercises to connect the scientific theory to the application in your day to day practice.

Participants will be equipped with the basics of prenatal genetic counselling and managing complex scenarios as well as an update on new technologies. They will also gain skills in taking a family history and assessing when a referral on to clinical genetics is indicated.

### Genetic Risk Assessment Course for Advanced Practice Practitioners and Advanced Breast Cancer Risk Assessment Workshop

**Date:** 10 -12 September, 2021

**Deadline:** 9 September, 2021 (Registration)

**Location:** Online

**Website:** <https://web.cvent.com/event/7c434250-ba27-4bc6-91e1-e8a40fa6ddaa/summary>

**Info:** The National Association of Nurse Practitioners in Women's Health (NPWH) is pleased to present this new course that will increase advance practice clinicians knowledge of and skills related to genetics, cancer risk assessment, carrier screening, and prenatal genetic screening. It will help prepare health care providers to proactively assess their patient's genetic risks, offer genetic screening when indicated and improve cancer prevention strategies and positive reproductive outcomes.

### Genetic Analysis of Population-based Association Studies

**Date:** 20-24 September, 2021

**Deadline:** 8 September 2021 (Registration)

**Location:** Online

**Website:** <https://coursesandconferences.wellcomeconnectingscience.org/>

**Info:** This advanced course aims to give researchers involved in genetic disease studies a firm grounding in the use of the latest statistical methods and software for analysis of genetic association studies. This includes both small-scale disease-specific studies and large-scale collaborative projects including those that can be used for analysis of multiple complex traits such as UK Biobank.

The course will cover both theoretical and practical aspects of the design and analysis of such studies. Each topic will include a lecture followed by a practical session in which state-of-the-art statistical software will be applied to relevant datasets. The practical sessions will illustrate the ideas presented in the lectures. All the software used will be freely available so that skills learned can be applied after the course.

The programme will also include seminars from internationally renowned researchers from the field of complex disease genetics, along with opportunities for participants to discuss their own research projects with course instructors and with each other.

The Genetics Society helps support several sectional interest groups by providing meeting sponsorship. We currently have 18 groups who organise sectional interest meetings with the organizers and dates of any forthcoming meetings are listed below. If you are interested in any of these areas, please contact the relevant organiser. This information is also available at: [www.genetics.org.uk/events\\_categories/sectional-interest-groups/](http://www.genetics.org.uk/events_categories/sectional-interest-groups/)

Groups who wish to be considered for sectional interest group status should contact Scientific Meetings Secretary, Prof Stefan Hoppler ([s.p.hoppler@abdn.ac.uk](mailto:s.p.hoppler@abdn.ac.uk)) in the first instance.

### Archaea group

**Contacts:** Malcolm White ([mfw2@st-andrews.ac.uk](mailto:mfw2@st-andrews.ac.uk)) and Thorsten Allers ([thorsten.allers@nottingham.ac.uk](mailto:thorsten.allers@nottingham.ac.uk))

### British Yeast Group

**Contacts:** Janet Quinn ([janet.quinn@newcastle.ac.uk](mailto:janet.quinn@newcastle.ac.uk)), Simon Whitehall ([simon.whitehall@newcastle.ac.uk](mailto:simon.whitehall@newcastle.ac.uk)), Julian Rutherford ([julian.rutherford@newcastle.ac.uk](mailto:julian.rutherford@newcastle.ac.uk))

### C. elegans

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### Evolutionary Genetics and Genomics

**Contacts:** Frank Jiggins ([fmjl001@cam.ac.uk](mailto:fmjl001@cam.ac.uk))

### Fly South-West

**Contacts:** James Hodge ([James.Hodge@bristol.ac.uk](mailto:James.Hodge@bristol.ac.uk))  
**Website:** <http://www.bristol.ac.uk/phys-pharm-neuro/events/fly-meetings/>

### Genetics Society Pombe Club

**Contacts:** Jacqueline Hayles ([jacqueline.hayles@crick.ac.uk](mailto:jacqueline.hayles@crick.ac.uk))

### London Fly Meetings

**Contacts:** Nic Tapon ([nic.tapon@crick.ac.uk](mailto:nic.tapon@crick.ac.uk)), Isabel Palacios ([mip22@cam.ac.uk](mailto:mip22@cam.ac.uk)), Giorgio Gilestro ([g.gilestro@imperial.ac.uk](mailto:g.gilestro@imperial.ac.uk))

### Mammalian Genes, Development and Disease

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### British Meiosis Meeting

**Contacts:** James Higgins ([mm2020@leicester.ac.uk](mailto:mm2020@leicester.ac.uk))

### Mammalian Genetics and Development

**Contacts:** Nick Greene ([n.greene@ucl.ac.uk](mailto:n.greene@ucl.ac.uk)), Andrew Copp ([a.copp@ucl.ac.uk](mailto:a.copp@ucl.ac.uk)), Cynthia Andoniadou ([malito:cynthia.andoniadou@kcl.ac.uk](mailto:malito:cynthia.andoniadou@kcl.ac.uk))

### UK Cilia Network

**Contacts:** Pleasantine Mill (<https://www.ed.ac.uk/mrc-human-genetics-unit/research/mill-group>), Toby Hurd (<https://www.ed.ac.uk/mrc-human-genetics-unit/research/hurd-group>) and Andrew Jarman (<https://www.ed.ac.uk/discovery-brain-sciences/our-staff/research-groups/andrew-jarman>)

### e-ACTG (edinburgh Alliance for Complex Trait Genetics)

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### Arabidopsis

**Contacts:** Peter Etchells ([Peter.Etchells@durham.ac.uk](mailto:Peter.Etchells@durham.ac.uk))

### Telomere Network UK (TeN)

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### Northern Bioinformatics User Group (Northern BUG)

**Contacts:** Jarek Bryk ([j.bryk@hud.ac.uk](mailto:j.bryk@hud.ac.uk))

### Ecological Genetics Group

**Contacts:** Gemma Beatty and Thom Dallimore ([genetics@britishecologicalsociety.org](mailto:genetics@britishecologicalsociety.org))

### Population Genetics Group

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### London Human Genetics Network

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# Heredity

## PODCAST

Hear directly from the experts, wherever you are  
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### Recent episodes:

#### **A pest with potential**

A scourge to agriculture, flour beetles may be the best model system you're not using

#### **Tales from the field**

Revisit some of the best fieldwork experiences shared on the Heredity Podcast

#### **Maternal matters**

Venture into the world of maternal effects – a form of inheritance that goes beyond genes

#### **Reversing sex**

What happens when a developing lizard embryo receives conflicting sex determination signals?

#### **When less is more: adaptive loss of function**

There are many ways to break a gene, and that's not always a bad thing

#### **History according to mice**

Etched into mouse genomes lie clues to the ancient movements of human populations

Listen online or download today: [\*\*nature.com/hdy/podcast\*\*](https://www.nature.com/hdy/podcast)

Heredity



The journal covers a broad range of topics within the field of genetics and therefore papers must address conceptual or applied issues of interest to the journal's wide readership. We encourage submissions on any study system but there should be a take-home message that focuses on broad general lessons that can be extended beyond single organisms.

**The journal particularly encourages submissions in the following areas:**

- population genetics/ genomics
- molecular evolution and phylogenetics
- functional genomics, transcriptomics, metabolomics and proteomics
- genome architecture
- epigenetics
- ecological genetics
- evolutionary genetics
- conservation genetics
- applied genetics
- quantitative genetics
- adaptation genomics
- crop and livestock genetics/ genomics

*Heredity's* original articles cover new theory and primary empirical research that offers novel insights, using the latest advances in technological and analytical tools. We have recently added a computer notes category, for which we invite submissions describing software packages that would be of interest for genetic analyses. The journal also encourages submission of reviews, mini-reviews and proposals for special issues on current topics.

## Editorial Board

*Heredity* has a small but diverse team of Associate Editors <https://www.nature.com/hdy/editors>, whose expertise spans the full range of the journal remit. We also have a dedicated editorial assistant, who is funded by the Genetics Society <http://www.genetics.org.uk/> and provides a direct communication link between authors, reviewers and editors. With this small team, we strive for a personalised approach to the publishing experience, which helps us to provide thorough, constructive and timely peer review.

## Fees and Open Access

Authors don't pay colour page charges and publishing is free, unless full Open Access is selected as an option. We also encourage use of Green Open access, by depositing author accepted manuscripts to institutional open access depositories; papers become free access 6 months after publication in print. Springer Nature also supports submission of manuscripts to preprint servers, prior to submission to *Heredity*.

## Reaching a Wider Audience

*Heredity* authors have the option of being featured in the *Heredity* podcast <http://www.genetics.org.uk/news/heredity-podcasts/>, which is presented twice per month by James Burgon. To more widely disseminate their research, *Heredity* authors also now have the option of writing a blog-type article in the *Nature Ecology & Evolution* community Behind the Paper channel <https://natureecoevocommunity.nature.com/> to accompany their formal paper published in *Heredity*.

**Nature Research Ecology & Evolution Community**

## Journal Metrics

Article metrics such as number of downloads, citations and online attention are available from each article page, and provide an overview of the attention received by a paper.

**The 2020 peer review performance metrics for *Heredity* are shown below:**

- Average time to decision without external review – 6 days
- Average time to decision following external review – 58 days
- Average time to secure reviewers – 20 days
- Average time for return of reviews – 18 days
- Articles published online within approximately 13 days and in print within 2 months.
- Over 51,000 recipients in receipt of the monthly electronic table of contents alert.

The *Heredity* website has over 57,000 page views per month.

# Heredity News



*Jinliang Wang stepped down as Associate Editor for Heredity*

After many years of diligent and valuable service, Jinliang Wang stepped down as Associate Editor for *Heredity* at the end of 2020 but continued with following up on manuscript revisions until mid-way through 2021. We would like to sincerely thank him for his contributions. His expertise in both theory and empirical work, ranging from conservation through selective improvement of domesticated species, reflects the full spectrum of the remit of the journal. Jinliang is a theoretical population geneticist who first moved from China to the UK in 1997 to do a postdoc at the University of Edinburgh and then joined the Zoological Society of London's Institute of Zoology in 2000. We already miss him greatly but we were pleased to welcome Matthew Hartfield to replace him (from September 2020). Matthew is a NERC Independent Research Fellow based at the Institute of Evolutionary Biology at The University of Edinburgh. He completed his PhD at Edinburgh followed by postdocs in Montpellier, Toronto, and Aarhus. His research uses theoretical and computational methods to investigate the evolution of reproductive modes, and how they interact with genetic selection. He also combines theory with empirical work and has very broad-ranging interests.

*His expertise in both theory and empirical work, ranging from conservation through selective improvement of domesticated species, reflects the full spectrum of the remit of the journal*



*Matthew Hartfield, new Associate Editor for Heredity*

We also welcomed two other new members to our Associate Editor team in September 2020. Sam Banks is a Professor in the Research Institute for the Environment and Livelihoods at Charles Darwin University (CDU), Darwin, Australia. He did his PhD at Monash University and, although a terrestrial mammalogist by preference, 'went marine' as a postdoc at Macquarie University before moving to the Australian National University and then CDU in 2018. A core research interest is the intersection of disturbance ecology and population genetics, specifically how fire regimes influence population dynamics and genetic diversity, as well as the broader application of genetic tools to mammal conservation in northern Australia. Lindsey Compton did such a fantastic job as Guest Editor of the special issue entitled "**Plant Quantitative Genetics: from Theory into Practice**" that we invited her to join as an official Associate Editor. Lindsey is a Lecturer in Genetics in the School of Biosciences at the University of Birmingham. Her work focuses on understanding the genetic basis of complex traits in crops and the application of this knowledge for improving crop breeding. Lindsey's research group uses a combination of statistical genetics and multi-omics approaches to dissect agriculturally important traits (e.g. abiotic stress resistance) into their underlying genetic components in a variety of organisms, but particularly the autotetraploid crop potato. She is particularly interested in genomic adaptations to the autopolyploid state, including meiotic chromosome behaviour, and how to address the resulting complexities in genetic analyses, including Quantitative Trait Locus mapping and association studies.

September 2020 was also when we announced the winners of our first annual prize for the best student paper published in *Heredity*. The winner was Donald McKnight, who did a fantastic presentation at the virtual PopGroup meeting "in" Liverpool in January and has also featured in our *Heredity* podcast.



## Genetics Unzipped – the Genetics Society podcast

Get the most fascinating genetics stories direct to your ears

How did bats evolve their amazing abilities to fly and echolocate? Is there a gene for being a Very Good Dog? And do you have genetic superpowers hidden within your DNA? The answers to all these questions – and much more – can be found in the latest series of Genetics Unzipped, The Genetics Society's podcast.

Over the past six months we've covered topics ranging from RNA vaccines to ageing, giants to genome editing, and conservation to canine genetics. We've heard how human activities have shaped species, and how scientists are using genetic information to do everything from solving wildlife crimes to developing better drugs. And we've also looked back on the extraordinary and controversial life of the great geneticist JBS Haldane and heard the true story of Dolly the Sheep from Bill Ritchie, the embryologist who cloned her.

Over the coming months we'll be discovering how researchers are studying the genetics behind life-limiting

conditions like chronic pain and ME/CFS, delving into the origins of life, and exploring the story of one of the most famously-named genes, Sonic Hedgehog.

Presented by award-winning science writer and broadcaster Kat Arney and produced by First Create The Media, Genetics Unzipped is released every other Thursday and alternates in-depth interviews with rich storytelling episodes.

Find Genetics Unzipped on Apple Podcasts, Spotify or wherever you get your podcasts, or head to [geneticsunzipped.com](https://geneticsunzipped.com) to check out our extensive archive and full transcripts. You can also follow us on Twitter @GeneticsUnzip or email [podcast@geneticsunzipped.com](mailto:podcast@geneticsunzipped.com) with ideas for stories and guests. And if you're already an avid listener, please do spread the word and take a moment to leave a rating or a review on Apple Podcasts to help more people discover the show.



*Genetics Unzipped is presented by award-winning science writer and broadcaster Kat Arney and produced by First Create The Media.*



# Honorary Secretary's Notices

**Kay Boulton** . The Roslin Institute, University of Edinburgh (secretary@genetics.org.uk)

## Committee changes and elections

### Upcoming Committee Vacancies

There are no unfilled vacancies upcoming for 2022

The following posts will become available on 1st January 2023:

**Policy Officer**, to replace Rebecca Oakey

**Corporate Genetics and Biotechnology**, to replace Alison Bentley

**Evolutionary, Ecological and Population Genetics**, to replace Jason Wolf.

### Medal and Prize Lecture Announcements

The 2021 Prize lecture and medal awards will be held in Cambridge on 14th October. Keep an eye out for invitations. The event will be available remotely and hopefully in person.

## Medal Nominations Genetics Society Medal

The Genetics Society Medal is an award that recognises outstanding research contributions to genetics. The Medal recipient, who should still be active in research at the time the Medal is awarded, will be elected annually by the Committee on the basis of nominations made by any individual member of the Society. Those making nominations must be members of the Genetics Society, but there is no requirement for the nominee to be a member, nor any restriction on nationality or residence. Neither current members of the Committee nor those who have retired from office in



the past four years may be nominated for the award. The recipient will be invited to deliver a lecture at a Genetics Society meeting, where the medal will be awarded, in the year following their election.

### Call for Nominations

Nominations are now being invited for the **2023** Genetics Society Medal. To make a nomination, please confirm that your candidate is willing to be nominated, then forward a two-page CV of the candidate, together with a list of their ten most important publications, plus a one-page letter of recommendation outlining why you feel their contributions to the field have been outstanding.

**These documents must be submitted electronically to the Honorary Secretary of the Genetics Society, Kay Boulton, by 30th April, 2022 at: [secretary@genetics.org.uk](mailto:secretary@genetics.org.uk).**

## Mary Lyon Medal



This award, named after the distinguished geneticist Mary Lyon FRS, was established in 2015 to reward outstanding research in genetics to scientists who are in the middle of their research career.

The Mary Lyon medal will be awarded annually, and the winner will be invited to present a lecture at one of the Genetics Society scientific meetings.

### Call for Nominations

Nominations are now being invited for the **2023 Mary Lyon Medal**. To make a nomination, please confirm that your candidate is willing to be nominated, then forward a two-page CV of the candidate, together with a list of their ten most important publications, plus a one-page letter of recommendation outlining why you feel their contributions to the field have been outstanding.

**These documents must be submitted electronically to the Honorary Secretary of the Genetics Society, Kay Boulton, by 30th April, 2022 at: [secretary@genetics.org.uk](mailto:secretary@genetics.org.uk).**

## Balfour Lecture

The Balfour Lecture, named after the Genetics Society's first President, is an award to mark the contributions to genetics of an outstanding young investigator.

The Balfour Lecturer is elected by the Society's Committee on the basis of nominations made by any individual member of the Society. The only conditions are that the recipient of the award must normally have less than 10 years' postdoctoral research experience at the time of nomination, and that

any nomination must be made with the consent of the nominee. Exceptions to the 10 year limit will be made if the nominee has taken a career break for child or other caring responsibilities.

Those making nominations must be members of the Genetics Society, but there is no requirement for the nominee to be a member, nor is there any restriction on nationality or residence.

### Call for Nominations

Nominations are now being invited for the **2023 Balfour Lecture**. To make a nomination, please confirm that your candidate is willing to be nominated, then forward a two-page CV of the candidate, together with a list of their ten most important publications, plus a one-page letter of recommendation outlining why you feel their contributions to the field have been outstanding.

**These documents must be submitted electronically to the Honorary Secretary of the Genetics Society, Kay Boulton, by 30th April, 2022 at: [secretary@genetics.org.uk](mailto:secretary@genetics.org.uk).**

## JBS Haldane Lecture

The JBS Haldane Lecture recognises an individual for outstanding ability to communicate topical subjects in genetics research, widely interpreted, to an interested lay audience.

This speaker will have a flair for conveying the relevance and excitement of recent advances in genetics in an informative and engaging way.

The annual open lecture will be delivered on a topic, and in a place, agreed with the Genetics Society. In addition to delivering the Lecture, the recipient will receive an honorarium of £1000 and a three-year membership of the Society.

### Call for Nominations

Nominations are now being invited for the **2023 JBS Haldane Lecture**. To make a nomination, please confirm that your candidate is willing to be nominated, then forward a two-page CV of the candidate, together with a list of their ten most important publications, plus a one-page letter of recommendation outlining why you feel their contributions to the field have been outstanding.

**These documents must be submitted electronically to the Honorary Secretary of the Genetics Society, Kay Boulton, by 30th April, 2022 at: [secretary@genetics.org.uk](mailto:secretary@genetics.org.uk).**

## Sir Kenneth Mather Memorial Prize

The Sir Kenneth Mather Memorial Prize of £150 rewards a BSc, MSc or PhD student of any UK University or Research Institution who has shown outstanding performance in the area of quantitative or population genetics.

The prize is awarded annually and pertains to a project report, dissertation or thesis submitted during the academic year in question. The winner will be invited to present their work, usually at the Genetics Society sponsored "Pop Group" meeting.

### Call for Nominations

Nominations for the 2020/21 Sir Kenneth Mather Memorial Prize should be submitted to The Genetics Society electronically via the website, before October 1st, 2021. To be eligible for nomination, as a condition of their course, theses/dissertations/project reports are required to be submitted by the student to the nominating University or Institution between 1st September 2020 and 31st August 2021. Nominators should supply their Genetics Society membership number on the application form.

Nominations will be assessed by a panel of two people with experience in the area of quantitative/population genetics, one from the University of Birmingham, and the other nominated by the Genetics Society. Nominations should include a cover letter from the proposer, a CV of the nominee and an electronic copy of the student's project report, dissertation or thesis. **Please note, the Genetics Society does not accept self-nominations for this award.**

# Local Ambassadors

The Local Ambassadors act as key liaisons between the membership and the Society's Office and Committee, helping to recruit new members, publicising the Society's scientific meetings and other activities, and providing feedback from the membership on matters of professional concern.

The Society normally appoints only one local ambassador per company, institution or department, but exceptions can be made when there are semi-autonomous subdivisions containing a substantial number of members or potential members.

Should you wish to volunteer as a Local Ambassador, or if existing Ambassadors wish to update their contact details, please contact the Honorary Secretary, Kay Boulton, by e-mail at:

**[secretary@genetics.org.uk](mailto:secretary@genetics.org.uk)**







**Location**

Oxford  
Oxford  
Oxford  
Oxford  
Oxford  
Oxford  
Plymouth  
Reading  
Salford  
Sheffield  
Southampton  
St Andrews  
Stirling  
Stirling  
Stoke-on-Trent  
Sunderland  
Swansea  
York

**Institute**

University of Oxford (Zoology)  
University of Oxford (Plant Sciences)  
University of Oxford (Plant Sciences)  
University of Oxford  
University of Oxford (John Radcliffe Hosp)  
Oxford Brookes University  
Oxford Brookes University  
University of Plymouth  
University of Reading  
University of Salford  
University of Sheffield  
University of Southampton  
University of St Andrews  
University of Stirling  
University of Stirling  
Staffordshire University  
University of Sunderland  
Swansea University  
University of York

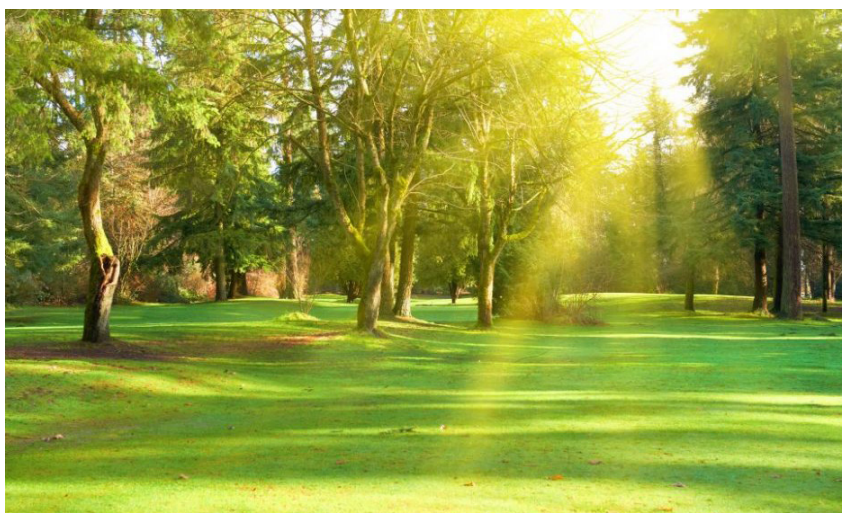
**Local ambassador**

Dr S E Kearsey  
Professor Liam Dolan  
Dr Niloufer Irani  
Professor Jonathan Hodgkin  
Professor Andrew O M Wilkie  
Dr Ravinder Kanda  
Dr Paul Potter  
Dr Mairi Knight  
Dr Louise Johnson  
Professor Geoff Hide  
Dr Jon Slate  
Dr Mark A. Chapman  
Professor Mike Ritchie  
Hoang Anh Nguyen  
Dr Mario Vallejo-Marin  
Dr Gavin McStay  
Dr Timothy Barrow  
Dr Claire Morgan  
Dr Sean T. Sweeney

# Life Membership of The Genetics Society

Have you reached the age of retirement (65), but wish to continue with your involvement in the Society? If so, and you are a current ordinary member, then you might consider applying to become a Life Member of the Society.

Life members will continue to receive notices and remain eligible to vote in the Society AGM but will not be required to pay further subscriptions. Recipients of the Genetics Society Medal will also be offered Life Membership. Should you require additional information about becoming a Life Member, please contact The Genetics Society Office ([theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)).



A Genetics Society Workshop

# Communicating Your Science

**We are accepting applications for the 2022 workshop, but expect it to be held later in the year (April, 2022). Dates and venue to be confirmed. Please check the website for latest information.**

An important part of science is getting your results and ideas across to others, through papers, presentations, theses, grant proposals, conversations and interviews. Your audience may include specialists in the field, those from other disciplines, industry, or the general public. How can you best communicate your science?

Working together with others on the course, you will learn how to structure stories, bridge disciplines, simplify concepts and communicate effectively with a range of audiences. You will also get in-depth tutoring and practice in one of three streams: storytelling and public talks, developing hands-on demonstrations or multimedia (podcasts).

## Speakers and Tutors include

### Erico Coen

(Author and Professor of Genetics, John Innes Centre, Norwich)

### Helen Keen

(Multi-award winning writer and performer)

### First Create The Media

(Led by award-winning writer and broadcaster, **Kat Arney**)

### Alison Woollard

(2013 Royal Institution Christmas Lecturer and Professor of Genetics, University of Oxford)

## Workshop Organiser

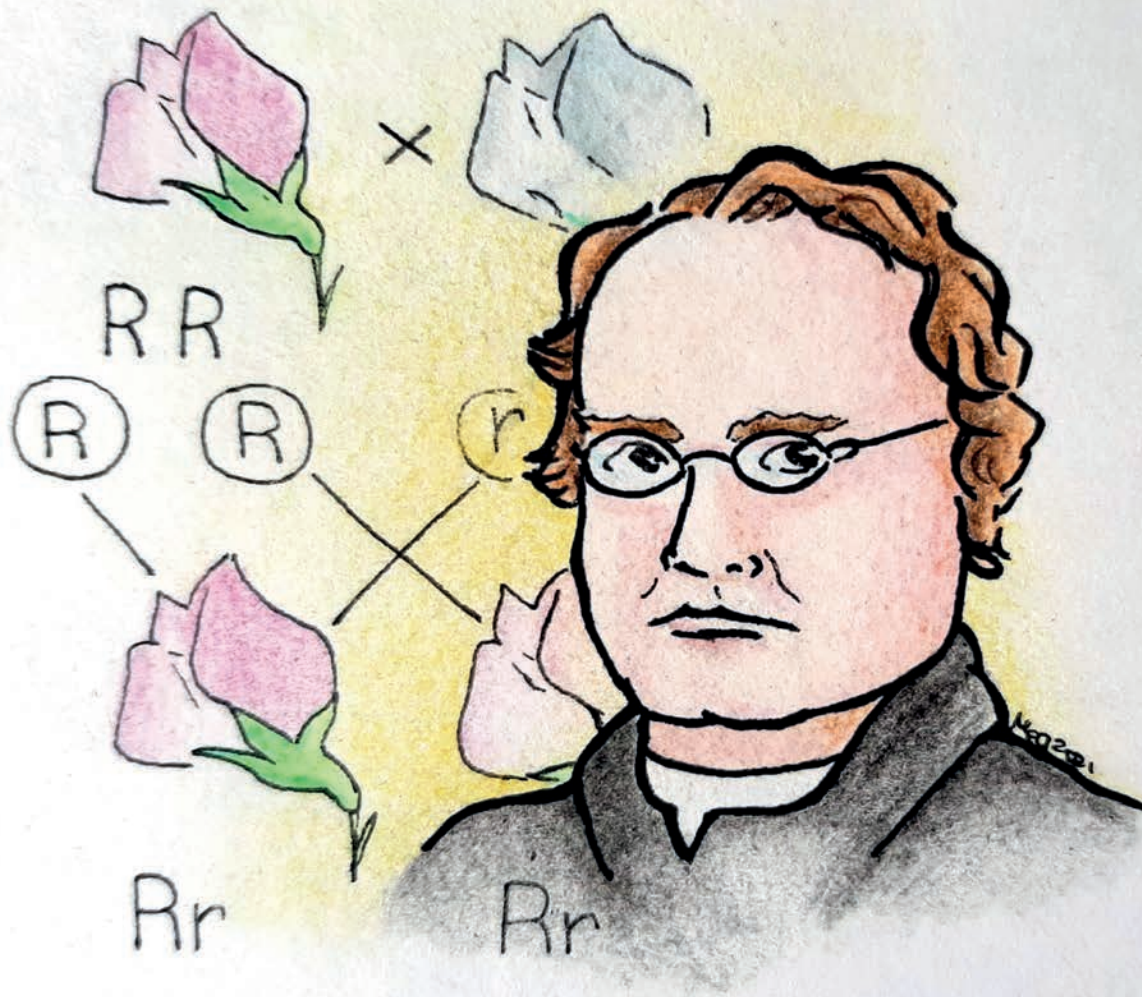
**Jonathan Pettitt** (University of Aberdeen)

**This workshop is open to PhD students and postdoctoral researchers working in genetics and related areas. The Genetics Society will cover costs of travel, accommodation and meals for all successful applicants.**

**The workshop is postponed until Easter 2022.  
Applications are open, deadline 14th February 2022**

<http://www.genetics.org.uk/grants/comm-your-sci>





# Call for Papers:

## Special issue of *Heredity* in honour of Mendel's 200th birthday

Mendel (born 22 July, 1822) is best known for his three laws of inheritance:

- 1) Law of independent assortment
- 2) Law of dominance
- 3) Law of segregation

However, the field of genetics has been enriched by considering exceptions to these laws. Examples include: epigenetics and genomic

imprinting, epistasis, paramutation, gene conversion, meiotic drive, social effects, symbiosis and the microbiome, quantitative genetics, cultural inheritance.

*Heredity* is seeking submissions of short essays, reviews, perspectives or research articles that address how Mendel's ideas have fueled development of understanding of such "exceptions".

The Genetics Society is sponsoring the special issue and would welcome representation across their membership.

Please send expressions of interest (by Aug. 20, 2021) to [heredity-journal@glasgow.ac.uk](mailto:heredity-journal@glasgow.ac.uk) with "Mendel" in the subject line. Full papers will be due by Dec. 20, 2021.

**Heredity**

 **the genetics society**



# Northern Bioinformatics User Group

Dr Jarek Bryk . University of Huddersfield



The Northern Bioinformatics User Group (<https://northernbug.github.io>) is a network of computational biologists and bioinformaticians (current and aspiring) in the north of England and a user-driven meeting that aims to build an engaged community of researchers to discuss ways to analyse or exploit large-volume data in biology. Modelled after the Scotland-based Next Generation Bioinformatics User Group (<http://nextngenbug.org>), Northern BUG was initiated at the University of York in May 2018 and, after four meetings, became a Sectional Interest Group, supported by the Genetics Society in September 2019.

Since then, we organised two meetings, at the University of Hull on 9th September 2019 and at the University of Leeds in 24 January 2020, with next meetings, originally planned for May at the University of Liverpool and for September at the University of Manchester, suspended until the end of the SARS-CoV-2 pandemic.

The meetings are small (50-75 participants) on a single day and single track, allowing for participants from the area stretching from Liverpool to Hull and from Sheffield to Newcastle to go to and back from the meetings on the same day. We are trying to treat them not as conferences but rather user groups, emphasising the more informal, open and collaborative nature of the meetings.

The Hull Northern BUG began with a short organising session, where

the Steering Committee was selected and an organising structure was established: Northern BUG has now a Slack channel (<https://northernbug.slack.com>), a Google Group page (<https://groups.google.com/forum/#!forum/northern-bug>) and a public website hosted at GitHub (<https://northernbug.github.io>). Dr Mark Dunning from the Bioinformatics Core Facility at the University of Sheffield then gave an overview of an offshoot initiative of the Northern BUG, a workshop for trainers (both core facilities-based and academic) held at the University of Sheffield on 19th July 2019 with the purpose of understanding similarities in current bioinformatics education across the region, standardise learning outcomes, and to share teaching load between institutions.

Research talks were delivered on medical genomics (Dr Lucy Stead, University of Leeds), on treatment-resistant glioblastomas, ancient DNA and big data archeology (Eleanor Green, BioArCh, University of York) and were complemented by an overview of the state-of-the-art in genome assembly (Dr John Davey, University of York) and Dr Chris Collins' (University of Hull) presentation on their 5000-core high performance computing cluster.

In the meantime, poster authors presented a series of 1-minute talks, briefly describing their projects and encouraging participants to learn more about them during the poster session.

The Leeds Northern BUG, organised by Dr Lucy Stead from Leeds, introduced two new sessions during the meeting: a coding clinic, matching up participants with bioinformatics questions to volunteers who can help them, and an in-depth best practices tutorial, the first

one on ChIP-sequencing, delivered by Dr Ian Donaldson (University of Manchester) and Dr Iros Barozzi (Imperial College London).

The meeting followed with a series of research talks on the analysis of carryover sequences in RAD-seq data (Haeyam Taiy from the University of Huddersfield), on the translation of long non-coding RNAs (Isabel Birds from the University of Leeds) and on bacterial GWAS (Dr Alexander Predeus from the University of Liverpool), among others. A poster session was held during the lunch break and the after-lunch session was enlivened by an opinionated talk by Dr Alastair Droop from the Wellcome Trust Sanger Centre on essential methods for modern bioinformatics: containerization, workflow management and code repositories. Dr Connor Meeham (University of Bradford) then delivered an overview of the genomic complexities of the West African strain of *Mycobacterium tuberculosis* and, finally, Dr Jarek Bryk (University of Huddersfield) presented his idea for a future grant application for comments and suggestions from the audience.

Both meetings concluded in nearby pubs, a crucial networking activity for the participants, who overwhelmingly agreed that the informal and flexible format of the meetings and its support for early career researchers are working very well and have already established Northern BUG as an important and useful focal point for the research community in the north.

We are all - organisers and participants - looking forward to resuming the Northern BUG meetings in the future.

*In this issue of the Newsletter, we have six feature pieces. The first feature is an interview of our outgoing and incoming Presidents, Professor Laurence Hurst and Professor Anne Ferguson-Smith, on what inspired them to become geneticists and their time as President.*

*The next article is based on the interview with the author of “Science Fictions: how fraud, bias, negligence and hype undermine the search for truth”: Dr Stuart Ritchie told us more about scientific misconduct and its wide impact.*

*The third piece is a series of interviews: “Genetics Society Summer Studentship - Share your story” tells the experiences of the past grant winners. Industrious Science features an interview with Jaqueline Palma, CEO at CircaGene and PetGenoma.*

*Then, we have an overview of the discovery of genetic linkage with the article “How genetic linkage was discovered” by Antonio Marco.*

*The final feature offers insights on the exceptional work of the Committee on Mutagenicity of Chemicals in Food, Consumer Products and the Environment (COM) and its impact.*

## Presidential handover

The Genetics Society president oversees the entire running of the Society and is in post for 3 years, which is enough time to make quite an impact! We spoke to our outgoing President, Laurence Hurst about his time in the Society, and to our current President, Anne Ferguson-Smith, about what she hopes to achieve over the next 3 years.



### Prof. Laurence Hurst

*What are the best memories you have of your role as President in these past three years?*

I have so many! It was a privilege to be the President during such a special time for the Society. Perhaps a highlight

was being on the stand at the Chelsea Flower Show and the opportunity to explain genetics to anyone willing to listen. On a lighter note, a memory that will be hard to expunge from memory is being woken by the hotel's fire alarm on a dark wet Edinburgh winter's morning to be greeted on a damp pavement by fellow committee members in their dressing gowns and slippers.

More generally, while the centenary came with many great moments, it is the interactions with the committee and the members that will really stick with me. It has been a pleasure to work with such a collaborative and talented group.

That so many volunteers work so hard to help run the society restores one's faith in humanity.

*What do you see as your biggest achievement whilst President?*

Not making an idiot of myself too often.

*As new president, you found yourself immediately immersed in the preparations for the Centenary of the*

*Genetics Society - How would you describe that period, how was it for the society to reach this milestone?*

For me at least the period running up to and through the Centenary year was very enjoyable. It should be no secret that the preparations for the Centenary were all down to the prior President, Wendy Bickmore, and her team. I had the easiest job – just turn up and speak. This being said, I much enjoyed doing some background reading on the early history of the Society. I hope we went some way to putting Edith Saunders, the mother of British Plants Genetics, in her rightful place, not least as the moving force behind the Society.

I think it is also worth looking back to see that there was no inevitability to the survival of the Society. For example, with moves to dissolve it about five years after it was founded, the Society very nearly became a footnote in history. That it has persisted is testament to all the people over the last 100 years that have tirelessly given of their time. It remains unusual amongst learned societies in being run by

volunteers. Perhaps this is part of its success as these same people have been invested in ensuring that the society adapt and grow. In turn, the Centenary has been, I think, an important part of the Society's evolution and has given a new impetus. People with deeper history with the Society assure me that there is a new energy about, which I could well believe.

### *How has the Genetics Society evolved in these years?*

As much as anything the Centenary provided the opportunity to pause, take a long view of the Society and ask where it might be going. I think perhaps we have changed on two fronts. When I took over the Society was strongly focused on academic geneticists. The Centenary forced us to think about that. While academic genetics remains at our core, we have taken the opportunity to explore more what we can do in terms of public engagement and, to a lesser extent, engagement with industry. Support for the younger members in exploring the diversity of career opportunities that are open to them is a welcome aspect of this new focus. In this regard, we are rediscovering the ethos of the founders – they very much encouraged the society to engage with experts outside of academia.

### *What is the next challenge you are preparing for after these great and intense three years?*

I suspect it will be a challenge to step back from what seems to have become a second family. I don't think I'll miss the lengthy zoom meetings, however.

### *What is going to be your relationship with the society?*

There if you need me. Otherwise, it's over to Anne....



## **Prof. Anne Ferguson-Smith**

### *What inspired you to pursue a career in genetics?*

Actually, I grew up around genetics because my father (who has just retired at 89) is a medical geneticist who has integrated genetics research with clinical genetics in the NHS; and my mother worked in the diagnostic cytogenetics lab. I recall when I was very small, my father showed me a metaphase chromosome under the microscope. It was all alone, away from the rest of the metaphase spread – I can still see it – and when he explained that chromosomes were tiny and inside all our cells and carried the code that made our cells work, I was blown away and excited that I could actually see this very powerful X-shaped thing. Later, when I was an undergraduate studying molecular biology at the University of Glasgow, the molecular genetics syllabus was truly exceptional – way ahead of its time – and what I learnt still has a huge influence on my research today. Importantly, our genetics lectures were excellent. In particular, Dave Sherratt was inspirational.

### *How did you become involved with The Genetics Society?*

I joined the Genetics Society a few

years after I started my own lab, and then was elected onto the Committee in Area A (Gene structure, function and regulation). This was early in the post-genomic era and was at a time when many genetics departments had disappeared or merged with other departments. Others may disagree, but it seemed to me that the academic discipline was suffering a bit because so much attention had been focused on generating sequence information. It was an interesting transitional time and I was really glad to be part of The Genetics Society because it reaffirmed how much we still needed fundamental genetics, quantitative approaches, model systems and model organisms, and needed to continue to explore and connect genetics across scales. Of course, we have now moved on and genomics is fully integrated into all aspects of modern genetics in very positive ways.

### *What do you feel is your USP as President?*

I am keen for genetics to be a bigger part of society's vocabulary. Now, more than ever before, genetics is part of everyday life with people being aware of DNA and genomes, the transmission of traits and genetic disorders, and gene-environment interactions. We therefore have a real opportunity to champion the discipline even more and reach out to the wider community. Although we are a so-called 'learned society', I'd like us to engage the wider public more. We are currently conducting a survey to better understand the public's perception of genetics, and this will provide a framework upon which we can plan further engagement.

I'd also like us to try more actively to widen participation and encourage the next generation who might not be thinking that genetics is for them, to experience it – perhaps via our very

successful summer studentship scheme but in other ways too.

*What are you enthusiastic about in your new role?*

In the short term, after having been shut away from in-person interactions with genetics colleagues at our meetings and events I am looking forward to seeing everyone again, and participating in stimulating Society activities.

I also am very much looking forward to the publication of our special issue of *Heredity* in July next year which will celebrate the 200th anniversary of the birth of Gregor Mendel. We are inviting essays around exceptions to Mendel's laws.

*What are your ambitions for the future of the Genetics Society?*

Since our centenary has now passed

and the Society has so successfully celebrated its past 100 years, now is the time to focus on looking forward. What should our priorities be as a Society, what kind of a difference can we make and what should the next 10 or 20 years look like for us? And in 98 years from now, what will the Society be celebrating at its next centenary?

Importantly, it is useful to think about how discipline is evolving and what can the Genetics Society do to continue to encourage fundamental conceptual advances in the field, communicate their impact, and facilitate the effective application of genetics to the global challenges we face? Genetics is even more embedded in all aspects of the life sciences with major contributions to make. For example, it has a profound role to

play in solutions linked to saving our planet and of course it is central to our understanding of the behaviours of pathogens, and health within populations..... the list can go on and on.

*If you had to give one piece of advice to early career researchers, what would it be?*

Two things...

First, stay curious, keep asking questions and always be a student.

Second, and this may sound a bit odd, but try and develop a relationship with your research that allows you to 'listen' to what it is telling you and let that guide your journey.

## A chat with Dr Stuart Ritchie - Exploring "Science fictions: how fraud, bias, negligence and hype undermine the search for truth"

By Margherita Colucci and Dr Stuart Ritchie

Recently, the amount of misleading news and discordant scientific research reached a new peak with claims and news on COVID-19 research. This may not come as a surprise, considering not only the exceptionality of the situation, but also the less exceptional records of fraud and negligent mistakes in science. Nevertheless, the topic is more complex than we think, and it is intertwined with many factors. Precisely, fraud, bias, negligence and hype, as it is brilliantly explained in "Science Fictions: how fraud, bias, negligence and hype undermine the search for truth" by Dr Stuart Ritchie.

Dr Ritchie is a lecturer at the Social Genetic and Developmental Psychiatry Centre at King's College London, interested in human cognitive behaviour and development and their interactions with genetics and neurodevelopment. Currently, he is focusing on researching and building the open science approaches described in his book. Specifically, he is exploring pre-registration methods: "Usually, pre-registration is considered for new data collection rather than for a data set that already exists, for example, if you're doing an analysis of UK Biobank data" he explained during our interview

"I've been trying to work out what is the best way to do that [pre-registration], also looking back through the literature to ascertain how reliable it is."

The circumstances that led to write this book, as Dr Ritchie explains in the preface, are connected to his experience as PhD student, when the failed replication of an influential research in psychology was rejected by the very same journal that published the study in the first place. At the time, this episode was perfectly framed by the debunking of other research papers (for example, the 'Coping with Chaos' published in *Science* by the social



*“The person who got me into science, from right at the very start, when I was an undergraduate student, was Richard Dawkins. I know that’s really a cliché thing to say, well, his books made me think ‘oh okay this science thing is worth it!’”*

psychologist Diederik Stapel), which raised concerns on the current state and integrity of scientific practice. However, Dr Ritchie’s interest in this topic have deeper roots.

“One of the reasons I was interested in [replicating] that study” he explained “is that I was interested in the skeptics scene, the scientific skeptics who investigate paranormal beliefs. I don’t think I would have replicated or tried to replicate that paper if I hadn’t had a pre-existing interest in the paranormal and this type of weird beliefs. It is strange that we have a whole subfield in psychology - a very minor subfield, of course - where people are making claims that are impossible according to physics.

And then, when we tried to replicate this paper and the scientific journal rejected us, I started to apply the skepticism to science itself, to the scientific process. And lots of other people did too: that’s where the replication crisis really kicked off”.

Touching such sensitive and controversial aspects of science, Dr Ritchie said that the book was welcomed by very different reactions. Numerous people contacted him to share their stories: “A lot of people said to me things like ‘I know X person at X University, who I am sure committed fraud’ or ‘I know that this person engaged in p-hacking [reanalysing the same data until statistically significant results are selectively obtained]; I used to be in the lab of someone who did this, I know this, I’ve seen it’” (many of these stories were

not further investigated). Even more stories and experiences were shared when, half-way through writing the book, he asked on Twitter for further contributions or information: “It was a real eye-opener, because it fits with that survey I mention in the book, which says that 10% of scientists say that they have themselves committed fraud, but 14% say that they know someone who has committed fraud. That fits with my experience where lots and lots of people I knew were saying they knew someone who has committed fraud, and so I think it made me realize that fraud, or the suspicion of fraud is a lot more common than we would like to think”.

But not everyone was so collaborative. On one side there were people that disagreed on divulging this research, fearing public’s mistrust in science.

On the other side, there were those who held extreme views and proclaimed that nothing of the current system should be salvaged and that there is need to ‘start from scratch’: “My book doesn’t go that far: my book suggests to try several strategies, to change things sensibly. There are serious problems in the current scientific system, but we don’t actually know what is the fix that would work.”.

There are also people who criticized the reformers in science. For example, Dr Ritchie recalls that when Science published several replication studies in psychology research, those who were against the replication attempts argued that the studies were low-quality,

not good faith replication work, in other words, just a way to ‘bully’ their colleagues.

The book analyses diverse episodes of research misconduct in all scientific fields, covering a vast time scale (from Samuel Morton’s measurements of human skulls in the 1840s to the unsupported claims in the 2017 book “Why we sleep” by the neuroscientist and Berkeley Professor Matthew Walker, for example). This broad overview allowed Dr Ritchie to explore how scientific fraud, its causes and its prevention may have changed through the years.

“There was never a golden age: if you look through history of science you will find all sorts of biases and historical frauds. [Fraud] is not a new problem, but I think that the modern academic publishing system, although it clearly comes with some benefits - such as being able to disseminate your papers to the world - it makes the situation worse under certain aspects. There’s some evidence suggesting that researchers have to have more papers on their CVs than they would have 10-20 years ago. You can’t just assume that all these papers are bad, but I think that you simply cannot have enough time to spend on really high quality work if you’re constantly under pressure to publish more and more and more papers - so this is one aspect that I think is possibly getting worse. I don’t think that this push for longer CVs benefits science.”

This is closely linked to competitiveness in publishing, summarised by the well-known phrase “publish or perish”. Dr Ritchie shared his thoughts on the role of competitiveness in scientific progress: “I think there’s a balance: I think hyper competitiveness is probably bad for science because science relies not only on coming up with ideas, but also checking

that those ideas relate to reality, test them against real data, and slowly build up your replicated, evidence-based research. However, if you focus too much on just replications, you will miss out on innovation. We need to create an environment where both ‘exploratory research’ and new ideas are praised. I think that, at the moment, we do research that’s basically exploratory, but then we write it up as if it was confirming a theory that we already had and predicted from the start. I think we need to decouple those: I think we need to have people who are coming up with new ideas - clearly labeled as exploratory research - and then other people who are trying to do confirmatory research, making theories. Both are equally important aspects of science and should be rewarded equally. Now we kind of do one versus the other”.

Dr Ritchie continued: “Other problems that I cover in the book, such as biases, negligent mistakes, and scientific fraud, have been around for as long as science has been around. However, I feel that in the last 10 years, people are talking more about problems like the replication crisis, and we’ve reached the point where we have the technology to deal with it. So I think that it’s much easier to be transparent and more thoughtful about open science now than before”. Dr Ritchie explained how favouring this type of discussions helps in bringing awareness to the problem. Universities are also positively contributing, creating open science focused workshops for PhDs, which may hopefully become a requirement soon.

And what happens when fraudulent science reaches the wider public? “For instance, take the story of masks in the coronavirus pandemic. There were scientists and politicians, in March [2020], saying to not wear masks. And I remember a tweet from an epidemiologist who said, ‘I’m at an

epidemiology conference. I’m in a room with 100 people, none of them are wearing masks. If scientists and epidemiologists don’t wear a mask, then you don’t need one either’. And I remember thinking that it was an amazingly strong statement. Now, of course, the evidence has changed, and we know masks are actually useful. So, I think that making strong statements and making science an unquestioned truth is actually a really bad idea and can backfire. It is important to give people a better idea on how science works”. There are many ways of bringing awareness to these issues: “At the very end of the book I suggest to have a look at what other scientists are saying about any given piece of research, to see what the general discussion is. Usually, you see a paper published in a journal and that’s it! You don’t see anyone else’s opinion on that. I think social media is a really great place to go to find other scientists talking about science. I think just seeing the general discussion around the paper is really important”.

And science communication may have a part in this, but needs to have an additional focus: from explaining scientific theories to explaining how science research and the scientific process work: “Science communication has not emphasized enough the actual process of how science is done. I think that if the general public is aware of it, science would not be perceived as some infallible truth. [Science] is a best guess, it’s been checked by a few people, there’s always the possibility that something you’re reading is not certain. So, I think science communication could do a better job of explaining, not just what the knowledge is in the scientific papers, but how that knowledge came about, also injecting a dose of skepticism”.

More possible solutions are investigated in the last chapters of the book (Part III: Causes and Cures). As not all solutions can fit every situation, Dr Ritchie highlights how different strategies can accommodate different research fields: “I think that researchers can pick and choose aspects and tools for open science transparency to make their research better.

We’re supposed to have a system that can do a much better job of stopping biased research getting into the literature, correcting mistakes, uncovering fraud before it gets out into the world. Imagine, for instance, if the people who were the peer reviewers of the article on autism and the MMR vaccine by Andrew Wakefield in 1998 had looked in a bit more detail into the children’s medical records and found that they were actually all falsified in the paper. Imagine how different the world would be. We would have way less new vaccine skepticism. At the moment, in the coronavirus pandemic, we’re worried about never getting these vaccines and worried that people won’t take them. The system is failing us in a really conspicuous way, and we can’t just accept this as normal, we have to do something to improve it.”

And as there are always new examples of ‘bad science’, Dr Ritchie revealed that there will be possibly a new chapter in the next year edition of the book, covering COVID-19 research. There has been really low-quality science, such as some articles retracted from the Lancet and the New England Journal of Medicine with questionable data and authors’ involvement. However, there have been good examples too: “the vaccine research has been amazingly transparent. They’ve been really upfront and in the last few weeks we’ve seen some amazing results from the vaccine studies. This makes me feel more optimistic”.

# Genetics Society Summer Studentship

## Share your story, Part 1

*The Summer Studentship Grant aims to support vacation research by undergraduate geneticists.*

*Since 2013, research projects have been successfully completed, and students have shared their results during the Summer Studentship workshop ... what next?*

*The grant winners shared their stories with us: what they gained from their experience, how they progressed in their careers and what impact the Genetics Society programme had on their career paths. This is the first of a two part feature: eight interviews are reported here, but there are more to come in the next issue!*

### Alberto Echevarría-Poza



My name is Alberto Echevarría-Poza and I am a passionate Spanish biotechnologist. In the summer of 2018, I was very lucky to receive a studentship from the Genetics Society to do a summer internship at the John Innes Centre in Norwich while I was still taking my undergraduate degree in Spain.

*Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

During my project in Professor Alison M. Smith's laboratory and under Dr. David Seung's great supervision, we studied natural variation in amylose content in

*Arabidopsis thaliana*. Amylose is the minor component of starch, yet it plays a great role in its properties. We studied several ecotypes of *Arabidopsis* that were predicted to have deleterious mutations in the only gene encoding the enzyme responsible for amylose synthesis. We surprisingly found that naturally occurring *Arabidopsis* plants can have a big variation in their amylose content, or even have no amylose whatsoever! We all still wonder what the role of amylose in nature can be.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

The best part of the project to me was how nicely Alison, David, and everyone else in the lab treated me. I could really feel everyone's trust, and, even if I was just a newcomer and had no experience in the field, they let me do all the exciting experiments of the project. Since the very first day, they made me understand that I was not there just to get some lab experience while speculating with something new for other lab members nor to just lend somebody a hand with some random experiments; David and Alison had carefully designed a brief project that I could complete during my intern-

ship and that would give great publishable results (and they were published indeed!). David and Alison were always patient with me and very encouraging. They really could not have treated me better!

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The entire experience was amazing! I learned a lot in the lab, and everyone made me feel like I was at home. I also had the opportunity to meet many other brilliant students and scientists and to make lots of friends. I really cannot recommend it enough.

*What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

Apart from feeling much more confident in the lab and learning to think as a scientist, the studentship also helped me develop my presenting skills, since all scholars were invited to a conference by the Genetics Society where we presented our projects and results to each other. The entire experience was of great help to then complete my undergraduate degree, but not only for that, because developing a critic way of thinking

and learning how to present one's ideas is always of great value in life regardless of what you end up doing.

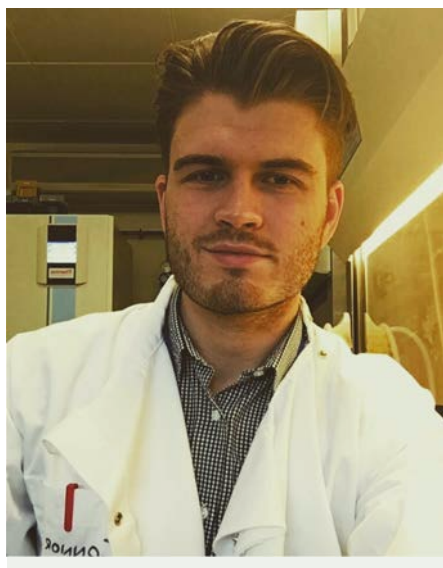
*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I feel like the internship really helped me finish making my mind up, and after completing my undergraduate degree, I returned to the UK to start a PhD at the University of Cambridge. I must confess I cannot wait for the pandemic to give us a break and go and greet Alison, David, and the rest of the lab gang at the John Innes Centre where I did my Genetics Society project.

*What are you planning for your next steps in your career and education?*

I still have some time until I complete my PhD and decide what to do next, but I really hope I always continue doing something related to science and I can teach younger people what others patiently taught me so together we all make the world a better place where to live.

## Connor Ross



I'm Connor, a final year MRC-DTP PhD student at Wellcome-MRC Cambridge Stem Cell Institute, University of Cambridge, working in the laboratory of Professor Jenny Nichols. I obtained a First-Class BSc (Hons) from the University of Aberdeen (with a year spent at the University of Guelph, ON, Canada) in Human Embryology and Developmental Biology. Currently, my main focus of research revolves around understanding the roles of WNT signalling in early human embryo development and naïve embryonic stem cells.

*Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

When I was an undergraduate student, I had the opportunity to work in the lab of Professor Stefan Hoppler, who was a lecturer on my course. I worked on validating downstream targets of Gata4 in embryoid bodies, derived from mouse ES cells to study heart development. We identified a series of genes which appear to be downstream of Gata4 and may play important roles in heart development of mice and humans.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

The best part of this project was working with mouse embryonic stem cells and generating embryoid bodies (EBs). At around D6 following the formation of EBs, patches of contractile cardiomyocytes are clearly visible.

Even more interestingly, when Gata4 is overexpressed, you could see more patches of beating cells, indicating that Gata4 when overexpressed generates higher numbers of cardiomyocytes.

I also had the opportunity to learn qRT-PCR, which is now a technique I routinely use in my PhD research.

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

Overall, the experience was fantastic and most worthwhile. The opportunity to be offered a summer studentship position massively boosted my CV and subsequent application for a PhD position here at the University of Cambridge. Having the research experience and access to grant money significantly assisted in obtaining a PhD position, effectively bypassing the requirement to do a Masters degree. I would absolutely recommend to any undergraduate student to seize this opportunity if it presents itself. It is a fantastic opportunity to get in the lab, get hands on and do some of your own research. It's also a great way to network and make new friends from different fields or disciplines.

*What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

Whilst undertaking this summer project working with mouse and human ES cells, it prompted me to read the literature surrounding the field and where we are today. A lot of the earlier work using human and mouse stem cells is well recognized. This knowledge massively boosted my exam answers far beyond the spectrum of material we taught in the lectures. Furthermore, the techniques I learnt here also assisted with courses that required a theoretical background. Overall, the summer studentship was undeniably a significant contributing factor to me obtaining a First-Class degree in a niche field and a PhD position in Cambridge.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

Since finishing the studentship programme, I was accepted into a well-established MRC



doctoral training programme PhD student-ship. I recently authored a review which was published in Nature Communications (Origin and functions of the yolk sac in primate embryogenesis) and have several first author and co-authorship manuscripts in preparation. I feel without the opportunity offered by the Genetics Society for a summer studentship, my ability to obtain a position at Cambridge might have been trickier.

### *What are you planning for your next steps in your career and education?*

Following the completion of my PhD, I intend to remain in research and obtain a fellowship to continue with my interest for WNT signalling in human development and disease. This will more than likely commence within the UK. Alternatively, a position in a clinical embryology and stem cell environment may also be under consideration.

## Elise Georges



Hi, I'm Elise Georges! I completed the Genetics Society Summer Internship in 2018. It took place during my last summer before I graduated from the University of Paris-Saclay with a BSc in Biomedical Science. Today, I am a PhD student at Queen Mary University of London.

### *Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

My project focused on the use of RNAseq to characterize the expression of H3K27me3 demethylases in Acute Myeloid Leukaemia derived cell lines and uncover their transcriptome to define clusters of genes in leukaemic pathways.

### *What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

The most exciting part was first on a professional level. I started the internship being confident in the lab but not knowing much about bioinformatics. I learnt a lot throughout the summer. From basic coding languages to writing scripts and run software to analyse data... I've had the chance to put my hands on many different aspects of bioinformatic analysis and dive into it. I am very grateful for the knowledge I was able to acquire. Then, on a more personal level, I have received a remarkable mentorship from Douglas Vernimmen, and I was able to thrive with his dedicated and very supportive supervision.

### *What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

I would definitely recommend this programme to students interested in pursuing a research career. The experience, the skills that I've gained and the incredible people I was able to meet throughout my

internship are just a few examples of the numerous valuable things I was able to take back with me after that summer. This studentship gave me the opportunity to have a professional experience abroad and get an overview of how research is conducted here in the UK compared to France.

### *What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

It meant a lot to me to be part of this as the opportunity of doing research abroad is extremely valuable at any point of a professional career. I developed both technical and soft skills, and I also met my current PhD supervisor while I was doing my internship in Douglas Vernimmen's lab, thanks to this studentship. I found that being fluent in English and having some bioinformatic experience on the CV opened many doors and probably helped my PhD application to go through.

I would like to thank the Genetics Society for allowing me to come over to the Roslin Institute and have an amazing experience abroad including the final summer workshop where I could present my work among other amazing students from all over Europe. The conference took place in Edinburgh during the last few days of August, and each student gave a short talk about their project. We also had the chance to have various lectures from PIs, post-docs and PhD students about their experience doing research in academia. This conference concluded the studentship and was definitely one of the highlights. Meeting the others, networking with them and having debates of what Science is or should be... All this provided invaluable insights and definitely shaped me as a young scientist.

### *Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path. What are you planning for your next steps in*

### *your career and education?*

Following the summer studentship, I applied for a PhD position at the Barts Cancer Institute in London. I don't work in epigenetics anymore but in epitranscriptomics, which are still very related fields. I now study enzymes playing a role in RNA decay and their impact on haematopoietic homeostasis and leukaemogenesis. My project is mostly based on in vivo and in vitro work but from time to time I am able to use my bioinformatic skills to analyse data I have generated. I also found myself very keen on spreading my knowledge through teaching and presentations, a feeling I started to have when given the chance to present my work during the final studentship conference.

Overall, the programme gave me the opportunity to gain bioinformatic skills that opened many doors and helped me understand a broader range of biology research, the opportunity to discover the haematology field that I ended up enjoying greatly and getting a PhD in, and finally the opportunity to realize that I liked doing research as much as transmitting and spreading the knowledge that we, scientists, have.

### **Gem Flint**



Hello! My name is Gem and I was a Genetics Society summer studentship holder in 2019 between my second and third years of university. I am currently a fourth year MSci Biomedical Sciences student at the University of Southampton. My main research interests are pharmacology, biochemistry, genetics and nucleic acid biology.

### *Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

In the summer of 2019, I undertook my summer studentship project at the University of Southampton research laboratories under the supervision of Dr James Dillon. My project aimed to investigate the implication of the mgl-2 receptor in the model organism *C. elegans* response to noxious stimuli. I achieved this by using genetic knockouts, genetic crossing and positive allosteric modulators to assess the receptors importance in *C. elegans* aversive behavior to 1-octanol.

### *What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

When I completed the studentship, I had just finished my second year of my undergraduate studies so at that point I didn't have the opportunity to conduct independent research before. The project enabled me to complete 8-weeks of hands-on research experience that I had yet to encounter through my degree. It was so exciting to get the opportunity to do so and I loved every minute of it. Prior to completing the project, I had thought I would like to do a PhD after my bachelors and masters studies were over and to pursue a career in research, however, I was unsure as I hadn't had any research experience.

The studentship confirmed to me that a career in research was definitely one I wanted to pursue, and a PhD was the next step I

should take to achieve that.

### *What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The studentship was an invaluable experience from start to finish. Getting the opportunity to conduct your own research is just the tip of the iceberg. It provided me with a great insight into what it's like to conduct research at a university environment-including all the lab meetings and journal clubs that come with it. Particularly, the workshop at the end of the 8-week research period was an incredible experience. It provided me my first chance to communicate my research to an academic audience as well as to meet and get to know fellow studentship holders. I was also able to hear about the amazing work my peers had conducted in their own studentships, listen to a series of interesting talks from the society academics and take part in many fun activities. The summer workshop was an amazing opportunity for an undergraduate student and one I will always look back on with fond memories. I would highly recommend the Genetics Society summer studentship for any student looking to gain research experience as it provided me with incredible opportunities both inside and out of the laboratory.

### *What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

The studentship project helped me develop transferable skills such as taking an observation from the laboratory and evaluate existing literature to derive a possible explanation for the observed result.

This is a skill that has been key for my degree; especially when writing both my dissertation and thesis. Also, I became a more independent thinker throughout the project and was more able to analyse the data collected and choose what direction I wanted

my project to go, with the help of my supervisor. The presentation to academics and peers at the summer workshop was also the first time I presented my work to an academic audience and this experience was invaluable. I have also been able to put into practice what I learnt from this experience in the presentations I have completed as part of my degree and consequently have done well.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I have almost completed my MSci in Biomedical Sciences and will be graduating this July. I am delighted to say that I have accepted a PhD position at Imperial College London within the Institute of Chemical Biology, co-funded by the EPSRC and CRUK and starting in October. The project is investigating the link between G-quadruplex DNA and PARP to overcome resistance to PARP inhibitors. Although this is an area slightly different to what I completed my summer studentship in, the skills and experience gained from the studentship led me to want to do a PhD in the first place and gave me my first taste for research. For these reasons, I am extremely grateful that I had the opportunity of taking part in the studentship, which helped to shape the direction I chose for my career and future.

*What are you planning for your next steps in your career and education?*

I will begin my PhD at Imperial College London in October 2021 and will continue this over the next four years. At the moment, my ambition is to then go onto post-doctoral research with my ultimate goal to lecture at university level alongside completing my own research.

I am open to new experiences, however, and look forward to seeing where my PhD takes me!

## Güniz Göze Eren



Hello, my name is Güniz Göze Eren. I am from Turkey. I was awarded with the Genetics Society Summer Studentship in 2017 during the summer of the 3rd year of my Molecular Biology and Genetics bachelor

*Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

I studied elephant brains! The African elephant's cerebellum, to be more precise. African elephants have the largest cerebellum compared to the other mammals and more than 97% of their neurons are found in this section of the brain. My project aimed to identify the neurodevelopment genes that give rise to this outstanding brain.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

I have mainly used comparative genomics tools to identify those genes. I have

compared several cerebellar development genes in the Elephantidae family to find out evolutionary changes that gave rise to the enlargement of cerebellum in African elephants. And the surprisingly Elephantidae family consists of phenotypically diverse animals, from as small as elephant shrews to manatees.

It was very exciting to work with a wide range of animal genomes.

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The best part of the Genetics Society Summer Studentship was to be part of an amazing research community.

I have conducted my summer internship at the Zoology Institute of Cambridge University and for someone at the beginning of a research career like me, the chance to meet these brilliant researchers conducting curiosity-driven research and sharing their passion and motivation gave me a huge motivational boost.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I am currently studying my PhD at the Caesar neuroethology institute on the predatory behaviour of cannibalistic nematode *Pristionchus pacificus*. After my bachelors, I decided to study brains in more detail, so I have completed a MSc in Neural and Behavioural sciences at the Tübingen University in Germany.

The Genetics Society Summer Studentship and my precious mentor, Dr. Stephen Montgomery, gave me the chance to take the first steps into my favourite research topic, neuroscience, and thanks to them I can follow my (research) dream.



**Isabel Esain Garcia**

Hello! I'm Isabel Esain, a 23-year-old scientist from Spain. I studied my undergraduate degree in Biochemistry at Imperial College London and carried out research at the Universities of Harvard (US) and Cambridge (UK). I am currently doing my PhD at the University of Cambridge working on epigenome engineering and DNA secondary structure.

*Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

My project focused on developing a new *in vitro* strategy to inform of *in vivo* genome editing success.

CRISPR has revolutionised the world of genome editing, one inescapable truth however, is that editing strategies must be adapted and tailored to the cellular context in which the target resides. Strategies that work in one cell type often do not work in another. This gap in methodology was stopping CRISPR from reaching its full potential. My goal was to develop an inexpensive high-throughput, reproducible, data-driven genome editing design platform.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

The most exciting part of the project was working on a novel concept with the potential to have a positive impact in the world, in a highly collaborative and multi-disciplinary group.

From a technical perspective, I had the chance to learn lots of new techniques from molecular biology and genetics to fluid dynamics, as well as putting the skills I learnt during my undergraduate degree into practice. I worked at the Cancer Research Institute (CRUK) in collaboration with the Department of Physics (Cavendish Laboratory), both are part of the University of Cambridge.

From a personal perspective, I couldn't have been luckier with the group I worked with. The atmosphere in the lab was highly supportive and everyone made doing science so much fun. My supervisor, Dr Alasdair Russell, is absolutely brilliant. He was always sharing his passion for science and he inspired me to pursue a PhD after finishing my degree. It was a real privilege to work with all the members of the lab and I will always be thankful to them for giving me the chance to grow as a scientist and as a person.

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The highlight of the experience was getting to share my passion with driven researchers from all over the world. Feeling useful within the scientific community in a highly collaborative environment and working together towards a common goal to make the world a better place. I believe that the future of medicine will rely on genetic methods for personalised treatments and precision therapies.

Being part of this studentship was crucial for my scientific career and without a doubt I would recommend it to anyone interested in research and with curiosity for science. Getting involved in the studentship will give you the chance to explore new interests and getting a wider insight into a field that you might be considering for your future career. I found the studentship intellectually stimulating and I would definitely repeat the experience.

*What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

The Studentship had a transformative impact on how I do Science. Not only it gave me the chance to develop technical, personal and soft skills, crucial for today's fast-changing environment, but also it taught me how to work under pressure, deal with failure and success and find creative solutions to problems.

Generating ideas and applying them to develop something new can be a challenging process, it requires a lot of scientific decision-making and experimental design. I realised how important it is not only to be an independent and open-minded researcher but also how crucial effective teamwork is. Working with others to achieve a collective goal and having everyone's perspectives to benefit the team is key for success in science, and this is something I experienced first-hand. I think collaborating with others is one of the most beautiful things in Science, and thanks to this Studentship I had the chance to collaborate with other researchers from Cambridge (Physics Department, Cavendish Laboratory), and with international collaborators from Germany and the US.

In addition, having the chance to present my research at the Genetics Society Studentships Conference in Edinburgh (UK) with other motivated scientists showed me how important science communication is. All the skills I gained during this Studentship gave me the confidence to finish my



undergraduate degree at the highest level and helped me to become the scientist and person I am today.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I am currently pursuing a PhD in Medical Science at the University of Cambridge.

My work focuses on developing a new targeted approach to investigate DNA secondary structure and performing epigenome engineering to obtain mechanistic insight into gene expression regulation in cells. Understanding the role of chromatin structure on gene activity presents a new strategy for cancer therapeutics.

Participating in the Studentship programme had a huge impact on my career. It allowed me to discover the world of genome editing and inspired me to pursue a PhD.

*What are you planning for your next steps in your career and education?*

Science is my passion and becoming a scientific professor in academia has always been my ultimate goal.

However, having worked on genome editing for a few years has also made me realise how important science policy is and how crucial it is to understand the ethical and social consequences of our research.

Thanks to the Genetics Society, I had the chance to participate at the 'Voice of the Future', a national discussion hosted at the UK Parliament between scientists from different fields around urgent matters in science policy.

This experience gave me a wider insight into how important policy-making is and how the politics of scientific matters work. With my motivation and experience, I hope to have a positive impact on society through science.

## James Sanders



Hello, my name is James Sanders. I started my education in Genetics at the University of Glasgow as an undergraduate before moving on to a Master of Research at the University College London in Synthetic Biology, focusing on building computational pipelines and machine learning models to engineer aminoacyl-tRNA synthetase (aaRS) enzymes for incorporation of non-canonical amino acids (ncAAs). I am now a first year PhD student with the Cai Lab at the University of Manchester, continuing to work on expanding the genetic code expansion now in synthetic yeast. I had the great fortune of being a summer student with the Open Bioeconomy Lab at the University of Cambridge, funded by the Genetics Society, working on optogenetic switches and antibiotic-free plasmid stability systems as a means of reducing the cost of molecular biology research in low-resource contexts.

*Could you give a brief overview to sum up the topic of the Summer Studentship project you undertook?*

Optogenetic switches are a fantastic means of gene induction, provided spatiotemporal

control of gene expression through widely available and cheap LEDs; however, they are poorly characterised. I hoped to create a datasheet for a set of optogenetic switches to allow researchers to select the genetic part most suitable for their needs. This involved assembling genetic constructs from basic 'parts' as well as adapting a test-bed platform to facilitate high-throughput light induction of these recombinant systems.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

I was very excited to have hands-on experience while working with a small, dedicated and driven team of synthetic biologists. What impacted me most was the interdisciplinary experience which had me designing genetic constructs, prototyping 3D printable devices, understanding C++ code within embedded software and soldering PCB boards.

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The real-world experience of a wet lab project gave me an insight into the potential biology holds for tackling today's pressing problems but also a reality check on the stubbornness of biology to do what you want sometimes (not everything works first try!) I would highly recommend a summer studentship to anyone contemplating it as a means of getting a taste of academic research before committing to longer projects.

*What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

I gained many technical skills within molecular biology but also learnt a great deal on project management and communica-

tion. The interdisciplinary nature of the project also instilled confidence in going beyond my comfort zone as a biologist.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I am now a first year PhD student with the Cai Lab at the University of Manchester, working to expand the genetic code of synthetic yeast to incorporate non-canonical amino acids. Since the summer studentship, I have completed a Masters in Synthetic Biology at the University College London, gaining a myriad of bioinformatic skills during the global pandemic. The studentship was excellent preparation for taking on a longer PhD research project. It taught me not only to feel comfortable in the lab environment but also how to structure my research and data sustainably.

*What are you planning for your next steps in your career and education?*

I look to continue in my PhD studies and hopefully contribute some useful research to the academic literature

## Katy Walsh



My name is Katy Walsh, I'm 24 years old and I'm currently studying for a PhD in Cardiovascular Sciences at the University of Manchester. Last year, I received a first class MSci degree in Genetics from the University of Glasgow.

*What was the most exciting part of the project (regarding the topic, the techniques, and/or your overall and personal experience in completing the project)?*

My summer studentship project was at the University of Leeds in their cardiovascular department.

The aim of the project was to determine whether small, specifically times fluctuations in maternal glucose levels contributed to aberrant foetal growth. Offspring from diabetic pregnancies are more likely to be born large for gestational age (LGA) and have a greater risk of developing cardiovascular disease later in life. Continuous glucose monitoring of diabetic females suggested that small changes in the maternal glucose correlated with LGA offspring.

We aimed to mimic these changes in an ex vivo placental model and determine the impact of this on miRNA expression.

*What was the best impression of the experience? What meant for you to be part of this studentship, would you suggest it to other fellow students and why?*

The most exciting part of the project for me was working with human tissue, as it made the study seem directly translatable to clinical work. After training, I was allowed to be present during non-emergency caesarean sections in order to collect fresh placental tissue. Discussing the study with these women and asking for their consent was a great way to see who this work would benefit.

*What skills and experiences did you gain? Do you think these were helpful for concluding your degree and how?*

The summer studentship was a great way to gain more practical experience in a subject I would not have otherwise had to opportunity to work on. After the studentship, I really enjoyed meeting the other studentship students and hearing about everyone else's

experience. It was also a good opportunity to practise presenting my work, which I have found incredibly beneficial as I have progressed onto a PhD.

Obviously, I gained a lot of technical laboratory skills, and generated a lot of data I was taught to analyse in an effective and efficient manner. I also learnt more personal skills including presentation and communication. I think my greatest takeaway from the studentship was a greater understanding of working in academia, and the opportunities available if I chose to continue studying after my undergraduate.

*Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.*

I am now studying a PhD at the University of Manchester, on their 4-year BHF PhD programme. I chose this as the initial MRes year allowed me to find my specific area of interest within cardiovascular disease and provided a proper training of the skills and attitude required to be successful in research.

The studentship made me realise I wanted to continue in cardiovascular research and cemented my goal of becoming a lecturer in cardiovascular health in the future.

*What are you planning for your next steps in your career and education?*

I will spend other 3 years in Manchester, where I will continue studying the vascular element of Alzheimer's disease, and I hope that after successfully defending my thesis, I will be able to apply for post-doctoral positions in USA.



Are you working in industry? Have you completed an internship? Do you have a story on your working experience in genetics to share? Then ...

### **We want to hear from you!**

This new series would like to give scientists (at any level) a space to share their experiences (short or long!) outside academia, to talk about their career journey, and, why not, to inspire early career scientists with suggestions and tips.

If you would like to be the next "industrious bee", please contact **Margherita Colucci** at [newsletter@genetics.org.uk](mailto:newsletter@genetics.org.uk).





## A day in the work-life of a Chief Executive Officer, Jackeline Palma



By Jackeline Palma and Margherita Colucci

Industrious Science explores the work experiences of Genetics Society members' in the genetic field working outside academia. These interviews aim to look into career paths, evaluating various aspects of the transition from student roles.

In this issue, Jackeline talks about her journey from university studies to CEO of two genetic testing companies, and gives precious insights into the start-up world. Jackeline's experience gives a great overview of the entrepreneur role in genetics - I am sure her advice will be inspiring to postgraduates looking for a career in this field.

**Margherita:** Thinking about your educational and career path ... When/How did you realise that moving into industry was the right choice for you?

**Jackeline:** I realised that to make an innovative change in the direct-to-customer DNA testing field and to propose a game-changer technology like our patented FHE encryption, I needed to be an entrepreneur. I had to start my own company and move to industry.

Our patent is the first of its type in the genetic industry scenario. It allows us to encrypt the information and analyse the data without opening the encryption - all is secured and private.

**M.:** What was the most appealing aspect of transitioning from academia to industry?

**J.:** The first call was to focus on my transferable skills: I wanted to challenge myself on a different level and explore my abilities in an industrial setting with real-world application.

**M.:** Did you always have a clear idea in your mind about your future path?

**J.:** The honest answer is no. However, I was always committed to conservation biology. Initially, I thought that this would lead me to work in the conservation research field and to study specific traits and genes for animal breeding. I have never thought that I would have ended

Fact

File

I originally studied Biology, graduated from the University

National Autonomous of Mexico (UNAM). Later, I completed my diplomas and postgraduate studies in Human Applied Genetics; I was Project Manager for In-Vitro-Diagnosis and Medical Devices (IVD) and sequencing companies.

Before starting my entrepreneurship journey, I worked on a BARDA (Biomedical Advanced Research and Development Authority) project in the USA.

I wanted to provide world-class Digital Health Services to protect and improve Public Health both at population and individual levels with the addition of privacy and protection of DNA Data. So, I became the co-founder of CircaGene.





up working under my own start-ups/ companies.

**M.: Throughout your career path, you gained extensive experience in different roles. Please, tell us about your professional evolution: how did you start, which challenges did you face?**

**J.:** I started as a female biologist in a developing country with little support in science research, I think that these were enough challenges to start with!

I undertook a double major at university, in marine biology and in genetics. I was keen to combine both fields, but it was soon clear that this wasn't a viable option – I even remember my academic assessor telling me: “Your proposed dissertation is suitable for a Doctorate degree, not for a bachelor, so no” – He did say no!

I initially struggled to keep a job in biology research as the opportunities were few and grants limited. Therefore, I decided to start a career portfolio to develop skills needed across industries (e.g. certifications in Project Management, Education, Marketing and Masters in Business Administration).

I completed a master degree in education, and I was a biology teacher for a while. I completed a bachelor degree in international marketing to experience Marketing and Business related roles, hoping that, one day, I could apply these acquired skills in the biological field!

It is funny to think that all the experiences and jobs I had (in education, communication, sales, marketing, project management, and genetics) contributed to my positive learning curve, and I am now consistently applying all these skills to my entrepreneur role. At the time, I saw this under a negative perspective, but now I appreciate that my career path prepared me to become an entrepreneur and inventor.

**M.: What experiences/career**

**decisions and interests led you to CircaGene?**

**J.:** I believe that curiosity is the key as well as the push to change the world. For example, I worked in a company where I had many new ideas to offer, the motivation to change some experiments, to innovate, but when it was not possible to do any of these in this company, I gained my own scientific awareness and asked myself how I could do all of this myself.

Also, bad career experiences made me want better working ethics as a boss, manager and leader. I was subject to discrimination and harassment because of my background: I wanted to change this and starting my own company was one way to stop this behaviour.

Before CircaGene, I was leading and managing budgets of over \$52 Million dollars, but even then, my results and success did not translate in promotion.

**M.: What were the challenges as a new start-up?**

**J.:** The first challenge was to communicate our value offer. Scientists tend to share higher-level technical information, therefore, I had to re-learn how not to be so technical.

The second biggest challenge was to raise funds, like any other start-up. New start-ups must fight to acquire credibility.

For example, I had to pitch in front of a huge audience (200+ persons, and only 12 female entrepreneurs) who were not convinced of the usefulness of DNA analysis for prevention. It was a very intimidating setting. So, be brave, Jacky, and be a Brave Biologist changing the world of DNA!

**M.: Which are the set of skills / experiences you think were the most valuable in building your start-up?**

**J.:** Confidence, analysis skills, growing resilience and being a doer! Learning to deal with different people and their

personalities is also helpful and a big challenge.

**M.: Evaluating your experience so far, focusing on your current role at CircaGene ... Could you describe your typical working day?**

**J.:** A typical workday includes planning and focus on goals/objectives that need to be achieved.

We exchange a lot of emails, and, at the moment, all meetings are online, which also prompts and helps in contacting partners, providers, suppliers and potential sales, for example.

I also run the R&D department: the research team is working all the time to develop all new kits, update the databases, and including all new discoveries in our reports.

There are also very long days, especially when there are not enough human resources available - no two days are the same and time management is essential.

**M.: Which are the most exciting projects you are working on at the moment?**

**J.:** I am working on genetic testing kits that are the first in the world to target autism and oral health. I am also planning original projects on cancer prevention and better tools for mental health.

I am very excited about our new series of webinars on DNA privacy and DNA analysis accuracy, it will be super engaging and educative.

**M.: How does a project idea begin and how is it led to completion? How many sectors and collaborators are involved?**

**J.:** We want to complete millions of projects!!! We founders are biologists, but, as a start-up, each project idea has to pass through an evaluation step where the market needs, finances and resources are considered. Thankfully, we have super intelligent people and a couple of the most brilliant genius scientists collaborating with us. There are also

volunteers helping us in the market research: they are assessors interested in the ethics behind our business, so they want to be part of it although they have no monetary investment - this is a huge asset for me.

These amazing people are integral part of the business because they care. Mentorship is like salt in the medieval time, extremely expensive and labour intensive to harvest and a specific amount is necessary to preserve the business.

**M.: Do you have contact with clients? Do you need to explain some aspects of genetics to them? How are clients supported to understand the genetics behind their results?**

**J.:** All of our results are delivered by an expert who explains the analysis process and results and recommends specialists' appointments.

Our kits also includes a Genetic Counsellor appointment, where the genetics behind the results is explained (including videos and animations) with the opportunity to ask questions too.

The team behind each analysis includes several experts: geneticists, computational biologists, physicians, counsellors, even phycologists if needed.

**M.: What would you say is the best part of your job? What are you passionate about your job?**

**J.:** Biology is the study of life and is therefore the broadest subject/field you can study. I like that Biology encompasses everything, from molecular research to animals. Following this passion and curiosity, I created one company for Pet Genomics and one for Human Genetics!! I am so excited to help optimise not only the human health, but also the wellbeing of the humans' "best friends": dogs.

Working in the healthcare field as a biologist is so rewarding: I feel I can contribute to change lives, helping prevent, cure and treat illnesses for both

humans and dogs.

**M.: Which are the current challenges in your field?**

**J.:** In my opinion, education is the main challenge. Specifically, it is of vital importance to inform people on the massive benefits of Genetic Testing and its advantages not only at an individual level, but also its impact on communities and, therefore, on the population and on the economy (prevention is more cost effective compared to treatments in the longer term).

Due to other DTC companies that have been inaccurate in their results and sold data to third parties without authorisation, it is now a challenge to regain people's confidence and trust in the science behind the tests, to make them aware of the clinical implications of the genetic analysis and the vast benefits that DTC-GT offers to all of us, including our pets.

It is important to demonstrate that it is possible to do genetic analysis respecting privacy, to improve the standards of diagnoses and to change the way personalised medicine is currently done.

**M.: Your point of view and experiences can help many students in having a clearer idea of a career... Any suggestions for postgraduates who are considering their future options? Your determination and your experiences make you a great example, especially for women in STEM and industry - what advice would you give?**

**J.:** Work with what you have, first... grow what you don't. Become a creator, a leader; never a victim of your circumstances.

Our ideals are just guiding stars; the journey is never over.

As a woman in STEM and from a minority group, I am a huge advocate on giving opportunities to people who want to demonstrate what they are capable of. All paths are valid, from decades in the

lab, to change of careers: it is the ability and willingness of taking care of the first steps that matters.

**M.: What to expect from the consulting and start-up world and how to prepare for the possible challenges?**

**J.:** Becoming a consultant requires a lot of persistence and, sometimes, networking skills. Hard work and ability to face uncertainty are expected both in a consulting role and in start-ups.

We need to prepare mentally, emotionally and financially. As scientist, we focus on acquiring knowledge, skills, but we rarely consider the interpersonal skills. You have to develop an emotional stamina for the job!

When I attend leadership courses I learn about the history of leadership, about big thinkers, but ... does anyone have any practical advice? How to overcome issues with employees, how to advise a team mate who is having x and y issues?

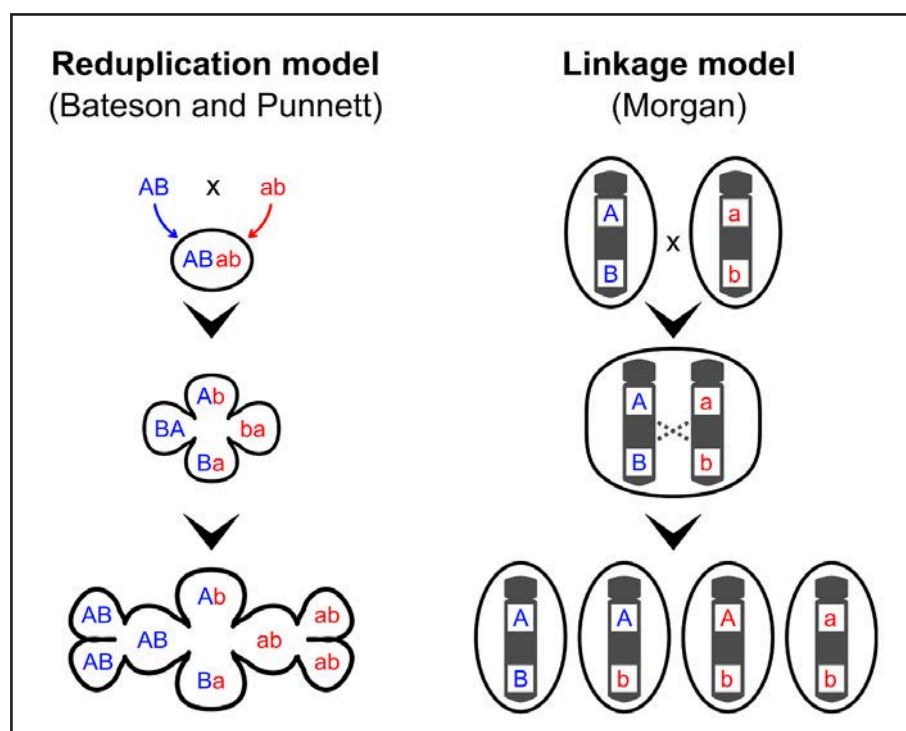
We need tips and examples from experienced people, as we may not have the time to learn all first-hand. Therefore, I am opening a new network to promote mentorship, focusing on various skills and entrepreneurship (<https://www.worldmexicannetwork.com>).

**Thank you very much, Jackeline, for the interesting insights into the start-up world.**

**I am looking forward to hear about other inspiring experiences - If you would like to be the next "industrious bee", please, contact me at [newsletter@genetics.org.uk](mailto:newsletter@genetics.org.uk)**

# How genetic linkage was discovered

Antonio Marco (University of Essex)



Reduplication and Linkage models to explain the coupling and repulsion of gametes.

Source: Marco A. (2013). Chromosomes and reduplication: how genetic linkage was discovered.

Genetic linkage doesn't need an introduction to members of the Genetics Society. It is a basic concept that we learn very early in our careers. However, it took some time and a few arguments between scientists until it was well characterised. Here I briefly outline how genetic linkage was discovered.

In 1900 Hugo de Vries, Carl Correns and Erich Tschermak re-discovered Mendel's laws of heredity. It was actually Correns who proposed the term 'law', yet he also described the first exception to one of these laws: the law of independent assortment.

In his re-discovery paper, he wrote that there are exceptions to this law and sometimes characters were linked or conjugated [1,2]. Plant breeders already knew that there were correlated characters, but they didn't have a convincing explanation for it. In a follow up paper, Correns further developed the idea of conjugated characters, identified examples from Mendel's own work, and associated it to the old idea of correlated characters [2]. He also differentiated between complete linkage, in which two characters were always together, and partial linkage, in which two

characters tended to appear together but in varying degrees. In parallel, Walter Sutton and Theodor Boveri were working on their chromosome theory, suggesting that hereditary material is hosted physically in chromosomes. Correns quickly accepted Boveri's ideas and proposed a chromosomal model of genetic linkage [2]. However, Correns' model did not have extensive support as the experimental evidence was still scarce.

By 1900 William Bateson was already a respected scientist working in animal evolution and mutation, and he proposed the discrete transmission of hereditary characters, similar to Mendel's postulates [3]. After the re-discovery of Mendel's laws, he not only gave full credit to his predecessor, but also published an eloquent defence of Mendel's laws in a work that included an English translation of Mendel's works: "Mendel's Principles of Heredity" [4]. Bateson reported to the Royal Society in 1901 the experiments he did in poultry (experiments he started in 1898) and in various plants in collaboration with botanist Edith Rebecca Saunders.

The work in plants was particularly important as Bateson and Saunders found evidence of some characters being inherited together, as Correns had suggested. Bateson, Saunders and a recently appointed fellow, Reginald Punnett, reported what it is considered the first empirical confirmation of linkage in plants [5].

Examples of linkage in animals soon appeared. For instance, the siblings Naomi and J. B. S. Haldane, together with student A. S. Sprunt found linkage in mice. Unfortunately, Sprunt

*A mechanistic explanation of linkage was still missing. Correns had already proposed that linkage was due to factors (genes) being hosted in chromosomes, but the chromosomal theory was not yet widely accepted.*

died in the Great War and J.B.S. Haldane, fearing that he may be killed soon as well (he served as a lieutenant in France), drafted a preliminary report while at the trenches. The paper became the first confirmation of linkage in mammals [6]. By that time, linkage was being studied in great detail in another animal which will change the genetics field for good: *Drosophila*.

A mechanistic explanation of linkage was still missing. Correns had already proposed that linkage was due to factors (genes) being hosted in chromosomes, but the chromosomal theory was not yet widely accepted. For instance, Thomas Hunt Morgan, an American embryologist, did not believe that chromosomes carried the genetic information [7]. He started to work on the transmission of characters in a new model organism that his colleague William Castle suggested: the fruit fly, or *Drosophila melanogaster*. After he discovered the first fly mutant (*white*) and confirming that the mutation was consistently associated with the X chromosome, he changed his mind [8]. Now inspired by Boveri's ideas, Morgan and his team, the so-called fly room, embraced the idea of chromosomal genetic linkage, and started to work tirelessly to systematically characterize and quantify genetic linkage in the fly. Only ten years had passed since Mendel was re-discovered.

Bateson and Punnett, however, were still reluctant to accept that chromosomes hosted the genes.

Instead, they proposed that the cellular divisions giving rise to gametes would be asymmetrical, having an impact in how genetic factors are transmitted [9]. For instance, if two gametes of genotypes AB and ab form a new zygote, the new individual will produce new gametes by duplication (cell division). But due to asymmetric divisions, there will be more gametes AB and ab than Ab and aB. Hence, A and B are coupled (linked) alleles. They called this model 'reduplication'. But Morgan and his students (Alfred Sturtevant, Calvin Bridges and Hermann Muller) had already collected dozens of *Drosophila* mutants and quantified recombination rates that they used to map genes into the chromosomes. The chromosomal-linkage model seemed to be consistent with their experiments. The findings were summarized in the now classic "The Mechanisms of Mendelian Heredity" [10]. Bateson eventually accepted chromosomal linkage, but he warned: "promising though it is, must be tried by tests on a scale far wider than experience of *Drosophila* provides before we are able to assess its value with confidence" [11]. Interestingly, one consequence of the Bateson-Morgan debate was the creation of the American journal *Genetics* [12].

Another discussion point was the meaning of non-additive genetic distances. Morgan and his group measured genetic distances as the frequency of recombination events. Thus, if we have three consecutive genes in a chromosome, the distance

between the first and the third should be the same as the sum of the distances between the first and second, and the second and third. But that was not the case, as the distance between the first and third was always smaller. The fly room attributed that to more than one recombination event. Castle, instead, proposed a model of three-dimensional organization of genetic factors [13]. After some bitter arguments it was settled that linkage and recombination was consistent with multiple crossing-over events between chromosomes. The last bit to the whole story was to empirically demonstrate that crossing-over was actually the cause of genetic recombination. This last piece of the puzzle came, once again, from plants. A junior Barbara McClintock and her PhD student Harriet Creighton showed that recombination in corn was indeed a consequence of crossing-over events between chromosomes [14].

Punnett eventually accepted the chromosomal theory of genetic linkage. But how had he and Bateson originally missed the connection between linkage and chromosomes? Punnett wrote decades after the debate was over:

"The answer is Boveri. We were deeply impressed by his paper [...] and felt that any [...] breakage and recombination was forbidden. For to break the chromosome would be to break the rules." [15]

Linkage was finally characterized, but investigations in this new field that combined genetics and chromosomes (cytogenetics) did not stop there. Eventually, McClintock demonstrated that crossing-over was not the only way chromosomes had to exchange information. She showed, indeed, that bits and pieces with genetic information were moving in and out of the chromosomes. But this is a different, yet fascinating, story.



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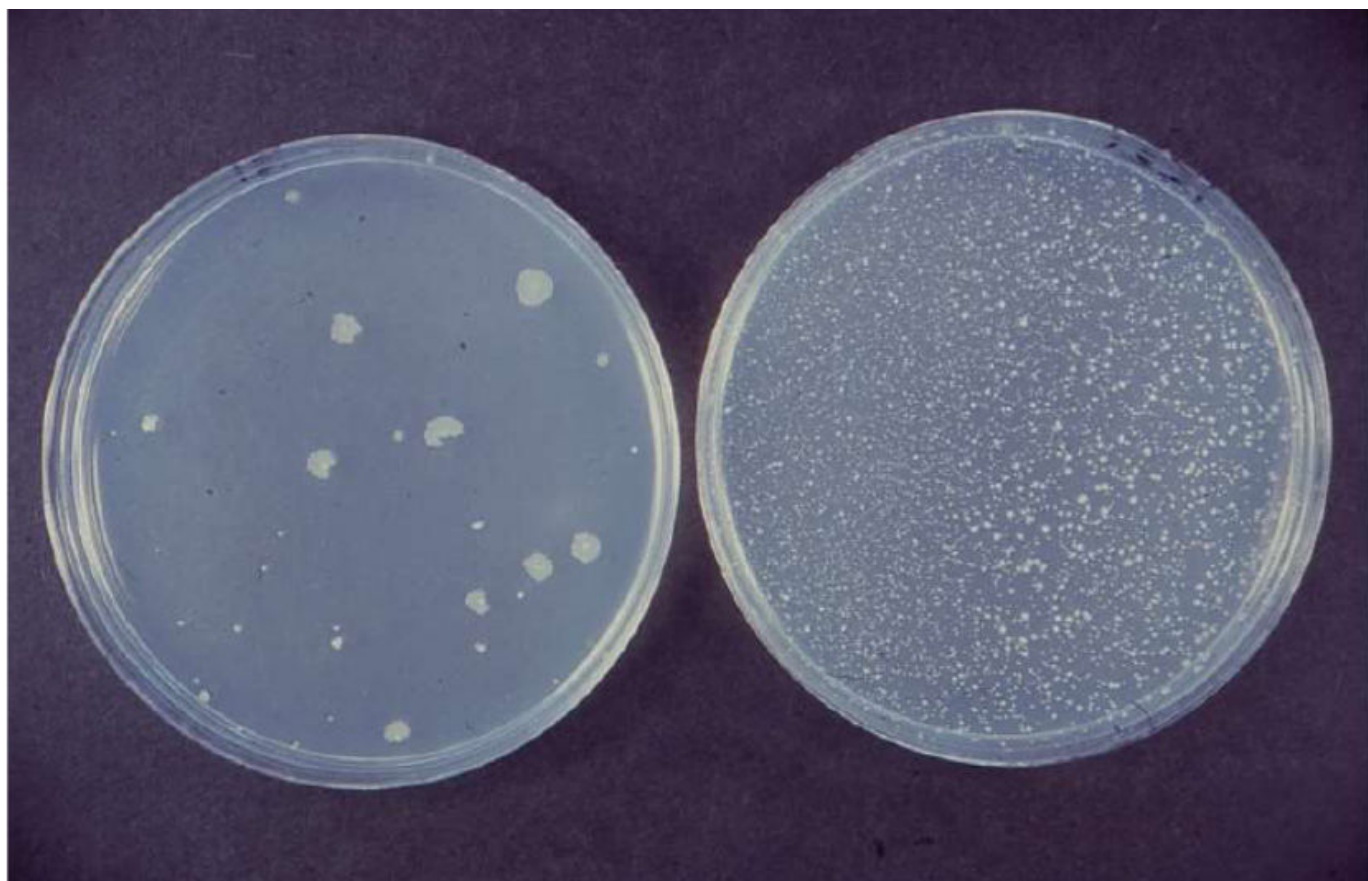
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# Committee on Mutagenicity of Chemicals in Food, Consumer Products and the Environment (COM)

David Lovell (St George's University of London)



*The Ames Salmonella test, the 'work horse' of genotoxicity since 1974. Left: control plate with natural revertants; Right, treated plate with high number of revertants suggesting chemical causes mutations*

For over 40 years, the Committee on Mutagenicity of Chemicals in Food, Consumer Products and the Environment (COM) Expert Committee has been providing expert advice to

government departments and agencies on issues related to mutagenicity. The COM assesses and advises on mutagenic risks to humans, on important general principles or new scientific discoveries

in connection with mutagenic risks and co-ordinates with other bodies concerned with the assessment of mutagenic risks making recommendations for mutagenicity testing. I have been chairing this

*The COM was established in its present form in 1978 following concerns developing in the late 1960's and 1970's that many chemicals, including pesticides, food additives and drugs, were likely to possess the ability to induce mutations.*

committee for the last 8 years, my term ended on 31st March 2021 and Professor Gareth Jenkins of Swansea University is the new Chair. The COM is a non-statutory body. It is jointly sponsored by the Department of Health and Social Care (DHSC) and the Food Standards Agency (FSA). Its Scientific Secretariat is led by Public Health England (PHE, which will be part of UK Health Security Agency from October 2021) together with the Food Standards Agency (FSA). The COM forms part of a triumvirate with its sister expert committees on Carcinogenicity (COC) and Toxicity (COT).

The COM was established in its present form in 1978 following concerns developing in the late 1960's and 1970's that many chemicals, including pesticides, food additives and drugs, were likely to possess the ability to induce mutations.\* A letter was sent to the then Department of Health and Social Security (DHSS) in 1970, pointing out the possible dangers from mutagenic chemicals. This led to the formation of the DHSS Sub-Committee on Mutagenicity. The interest of Professor Bryn Bridges of the MRC Cell Mutation Unit at Sussex University in mutations led to him publishing in 1973 a 'three tier' scheme for mutagenicity testing which subsequently developed into the basic approach used by many regulatory agencies [1,2].

The 1970s was a time of intense activity in the development of tests for mutations (which led to the emergence of genetic toxicology). One milestone was Bruce Ames' 1973 paper [3] with the succinct title "Carcinogens are Mutagens". Ames

had built on the work of Heinrich Malling who, in 1971, had developed methods for studying metabolism using isolated liver preparations, called an S9 mix, so allowing mammalian metabolism to be explored *in vitro/ex vivo* [4]. Ames developed a quick and, at the time, cheap test which combined the test chemical, the S9 mix and specialised bacterial strains of *Salmonella* and *E. coli* lacking the ability to grow on agar plates deficient in an essential amino acid, histidine or tryptophan. This test detected 'revertant colonies' resulting from mutations which allowed the bacteria to survive on the deficient agar. His early work showed a high association between chemicals which were bacterial mutagens and the results of animal tests for carcinogenicity.

Ames's work led to the development of a plethora of other short-term tests using a range of endpoints – gene mutations, chromosomal damage, and marker systems showing indications of DNA damage - in various organisms - rodents, fruit flies, bacteria, fungi and plants as well as a set of *in vitro* test systems. Bridges' paper introduced a strategy into the testing process and he was a member of the DHSS sub-committee from 1972 (which turned into the COM in 1978) and became its Chair in 1983 when it took on its current role.

A distinction is made in the field of genetic toxicology between mutations and genotoxicity. A mutation is an event which is passed from generation to generation in an organism, such as a hereditary defect, or through in cell division to a daughter cell, such as in cancer.

Genotoxicity is damage to the genetic material such as chromosome breaks or damage to DNA which may subsequently lead to a transmissible mutation or lead to the death of the cell without it being transmitted to the next generation. Increasing appreciation on the role of genes in cancer has been reflected in the role of the genotoxicity short term tests and *in silico* methods for predicting and providing evidence for events in cancer. The tests are also used to identify potential mutagens in substances or chemicals before they are introduced on to the market thereby protecting the human population from the induction of mutations causing inherited conditions.

Mutagenicity has several axioms: an agent that is genotoxic *in vitro* is presumed to be genotoxic *in vivo*; one that is genotoxic in somatic cell is presumed to be genotoxic in germ cells; a genotoxic chemical is assumed not to have a threshold. Experimental evidence is needed to override these axioms and the COM provides advice on such evidence. Identifying an agent as a genotoxic hazard has traditionally been a binary decision. Increasingly, however, there is interest in developing quantitative dose-response genotoxicity data for risk assessment.

The COM published in 1981 the DHSS Guidelines [5] to help bring some order into the field. This explained the background to mutagenicity, the various testing approaches and was influential, together with the newly formed UK Environmental Mutagenicity Society (UKEMS), in developing clear guidance for the conduct and interpretation of the tests [6]. This work had a major impact on the evolution of the international regulatory environment. It helped the development of a series of OECD Guidelines for genotoxicity tests; over 20 have been produced and the COM continues to have appreciable influence in the OECD activities.

The 1981 Guidance was updated in 1989

[7] and again in 2000 [8] when it incorporated an *in vitro* micronucleus test for clastogenicity/aneuploidy and *in vivo* testing using transgenic animal models. A further revision in 2011 [9] specified in more detail the three-stage strategy. A new revision has just been completed and will be published in 2021. This reviews new tests in development and modifies advice, based upon experience, on how methods should be used. The series of updates have rationalized the battery of core tests needed and provide a clear strategy for testing for the three endpoints: gene mutations, chromosomal damage and aneuploidy.

A key aspect of COM's guidance relates to the 3R's (Replacement, Reduction and Refinement) with respect to animal use. The current strategy requires *in vitro* and *in silico* testing methods to be used as much as possible, only moving on to *in vivo* tests when these are considered necessary and where the test is relevant to the chemical and endpoint being investigated. The EU's Cosmetics Directive, where the assessment of cosmetic ingredients using animals is not permitted, has created a spur to develop alternative methods that can provide safe products without animal testing. It is worth noting that the Ames test is the alternative test method *par excellence* having now been used for over 45 years in genetic toxicology. One of the original OECD 1983 guidelines was No.471 Bacterial Reverse Mutation Test [10] which includes the

'Ames Test', and which has remained unchanged.

Bryn Bridges was followed as Chair of the COM by Professor James (Jim) Parry (University College, Swansea) from (1993 to 2001) a fungal geneticist with wide experience in genetic toxicology and then by Professor Peter Farmer of the University of Leicester (2001 - 2012) who is an expert on DNA adducts and myself from (2013-2021). Many researchers in the field have been members of COM and many, especially in the early years of the committee, will also have been members of the Genetics Society.

At the practical level, the Committee is made up of the Chair and 12 members, including the Chair of the COC as *ex officio* and 2 lay members. Members are scientists in academia or with experience in organizations with expertise in the science, test methods and regulatory requirements associated with genetic toxicology and mutagenicity. This mix of members is primarily why the COM is somewhat unusual in being an Advisory Non-Departmental Public Body (ANDPB) - not the catchiest of acronyms - which is a committee of independent experts rather than a government departmental committee.

**Currently, the COM meets formally 3 times a year (at present, through virtual meetings). Minutes and papers are uploaded to the COM Website (<https://www.gov.uk/government/>**

**[organisations/committee-on-mutagenicity-of-chemicals-in-food-consumer-products-and-the-environment/about#meetings](https://www.gov.uk/government/organisations/committee-on-mutagenicity-of-chemicals-in-food-consumer-products-and-the-environment/about#meetings)) and past papers from 2001-2013 can be found at the National Archive**

**<http://webarchive.nationalarchives.gov.uk/20131102020211/http://www.iaacom.org.uk/papers/index.htm>).**

**Meetings are open to members of the Public who can attend by applying in advance of the meeting. All members are expected to act as individuals, not as representatives. Members do not receive any remunerations for their work. They must declare any relevant interests and not participate where these may conflict with matters being discussed.**

The consequences of Brexit are challenging for COM. Many regulations have been agreed internationally with European organisations such as the European Chemicals Agency (ECHA), the European Food Safety Authority (EFSA) and the European Medicines Agency (EMA) having responsibility. UK scientists played important roles in the development of EU Guidelines, making important contributions and interventions to ensure that the approaches taken were based upon evidence and good science. It is not yet clear how the changes involved with the UK's exit from the EU will ensure that UK expertise continues to influence such scientific debates. Almost certainly there will be a need for more expert advice to the relevant UK Government Departments and Agencies taking on these roles,

In some ways the COM's work develops slowly. Agreeing new OECD guidelines or revisions to existing ones is a slow process and depends on the mutual agreement of many countries. However, it is likely that the field of genetic toxicology will now change more rapidly. Scientific advances made in areas such as the -omics, whole genome screening (WGS),

*Vacancies for memberships of Committees such as the COM occur from time to time, and I encourage interested people to consider applying for these roles. Members find the committee interesting, intellectually rewarding but also an important use of their scientific skills to the benefit of the wider community both nationally, and because to the COM's high regard, internationally.*



machine learning / AI, *in silico* and other modelling methods will become integrated into the regulatory environment. Investigations in area such as nanomaterials, 3D-tissue models and possible epigenetic effects will require that new techniques will need to be integrated. The paradigm shift in moving from identifying genetic damage as a binary present/absent hazard identification to the use of the data as part of a quantitative risk assessment will probably occur in the relatively near future.

Vacancies for memberships of Committees such as the COM occur from time to time, and I encourage interested people to consider applying for these roles.

Members find the committee interesting, intellectually rewarding but also an important use of their scientific skills to the benefit of the wider community both nationally, and because to the COM's high regard, internationally. Hopefully, this brief overview of the COM will pique your interest.

\* For an indication of the interest that was developing see <https://www.nature.com/articles/245355a0.pdf> Many of the key players in this initial stage are also mentioned in Sobel's review of radiation and genetic damage and in the abstracts of "Fifty Years of Genetics" a proceedings of the 160th Meeting of the Genetics

Society in 1969 <https://www.nature.com/articles/hdy1969100.pdf>.

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The Genetics Society receives several requests from members each year to sponsor meetings in the field of genetics. These meetings are usually one-off meetings with an ad hoc organising committee and may be partly sponsored by another Society. In this issue, we have reports from Angelica Ronald and Karoline Kuchenbaecker.

# London Genetics Network meeting

December 4th 2020

Angelica Ronald, Karoline Kuchenbaecker (Birkbeck)

The aims of the London Genetics Network are:

1. to increase London-based collaboration between researchers interested in human genetics
2. to leverage London's expertise by fostering knowledge exchange
3. to develop practical and online resources for training and development in statistical and computational genetics
4. to support early career researchers through a range of new opportunities and research prizes

We held our launch meeting in December 2020 which was generously funded by the Genetics Society.

The enthusiasm with which the Network was received was overwhelming. We had over 300 registrations from over 30 different institutions across London and the surrounding regions, 19 poster submissions and viewing numbers hovered between 150-175 viewers throughout the day. In

addition, we have received many messages of encouragement from individuals, including many renowned genetics researchers, such as Professor Robert Plomin, KCL, and Dr Nick Luscombe, Crick Institute.

The day kicked off with superlative keynote talks by Professor Thalia Eley (King's College London) and Dr Nathan Skene (Imperial).

The schedule of speakers included talented early career researchers Dr Oliver Pain (King's College London), Dr Olga Gianakopoulou (UCL), Dr Kaitlin Samocha (Sanger) and Dr Conrad Iyegbe (KCL). After lunch we were treated to a methods talk by Professors Frank Dudbridge (Leicester) and Andrew McQuillin (UCL). A future directions session offered broad visions from Professors Aroon Hingorani (UCL), Sir Mark Caulfield (QMUL) and Angelica Ronald (Birkbeck).

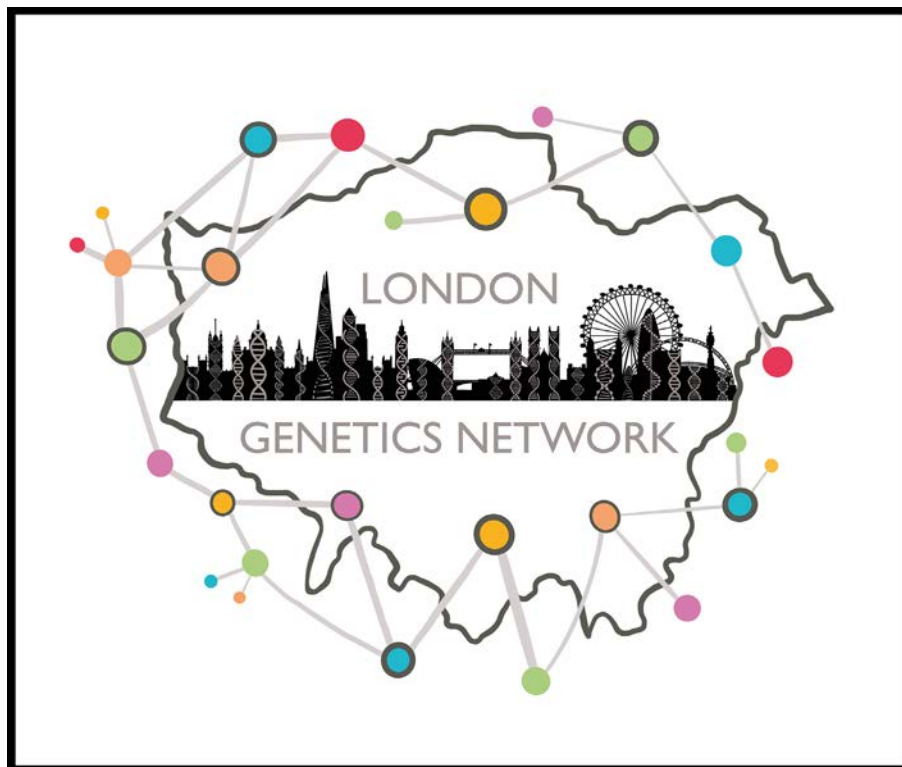
The chairs, Dr Emma Meaburn (Birkbeck), Dr Yalda Jamshidi (St George's), Dr Nick Luscombe (Crick Institute) and Dr Karo-

line Kuchenbaecker (UCL) -- all chosen for their own knowledge and breadth in the field -- rose to the challenge of eloquently introducing speakers and keeping them to time despite the remote Zoom 'airwaves'. =

The speakers, chairs and committee members included some of London's most influential genetics researchers, representing most of London's key institutions for genetics research.

The human genetics content touched upon, amongst other things, common and rare genetic variation, physical and mental health, behavioural genetics, ancestrally diverse populations, typical and atypical development, phenotyping, disease prediction, sequencing, single cell genomics, drug development, GWAS and transcriptomics.

The posters were judged by an independent committee, Dr Alvina Lai (UCL), Professor Andrew McQuillin (UCL) and Professor Robert Plomin (KCL). The prizes were £100 John Lewis vouchers each. In their presentation, the committee noted the



[www.gel.bbk.ac.uk/london-genetics-network/](http://www.gel.bbk.ac.uk/london-genetics-network/)

high quality and wide scope of the posters. We would like to congratulate the 3 poster winners, Elena Arciero (Sanger), Albert Henry (UCL) and Chloe Austerberry (UCL) for their excellent posters.

As per the Society remit, we were able to offer 3 carer awards to cover the cost of preschool childcare to attend the meeting. These were selected randomly from a hat. We congratulate Dr Chloe Wong (KCL), Laura Havers (Birkbeck) and Dr Zhanna Balkhiarova (Surrey) for their carer awards and were delighted they were able to attend the meeting.

In terms of other highlights, we announced a One-New-Contact challenge at the start of the day. Participants were encouraged to make at least one new contact using the direct messenger function in Zoom. Our 'strategy meeting' at the end of the day,

with everyone unmuted, allowed us to capture live feedback from attendees and to brainstorm new activities within the network and to conduct polls on what people would find useful.

Finally, we would like to thank Sandra Howgate, our brilliant illustrator; our meeting ECR rep Chloe Austerberry; PhD student Aislinn Bowler who took on the role of meeting coordinator with aplomb and kept Zoom running smoothly throughout.

#### Facts and figures

- 300 registrations from over 30 different institutions across London and the surrounding regions, 19 poster submissions, 11 talks, 3 poster prizes
- Viewing numbers 150-175
- Twitter impressions: 111 total tweets,

which had the potential to reach 172,014 people

- Questions from the audience throughout the day: 39
- Feedback showed 100% of participants rated the meeting as "very good" or "excellent"

#### Example Tweets

"Super cool & insightful research presented today on the influence of human genetics on diseases, how to leverage that info for prediction, personalised medicine & future directions of genetics research at #LGN2020 Great organisation by @KKuchenbaecker et al! #Genomics #genetics"

"This is such a great idea I now want to start a Amsterdam Genetics network"

"Sounds wonderful if you ever want to make it a London-Amsterdam Genetics network let us @NTRscience know!"

"Congratulations to all!! Great posters and great meeting! :)"

#### Example Feedback comments

"Getting to know other London-based genetics researchers and active efforts from organizers to highlight the works from early-career researchers and carer[s]" (best bits)

"My favorite part of the meeting was the diversity of subjects that still all seemed relevant to behavioral genetics in general. Nice choice in terms of presenters!"

"Wish we could have all had drinks after!"

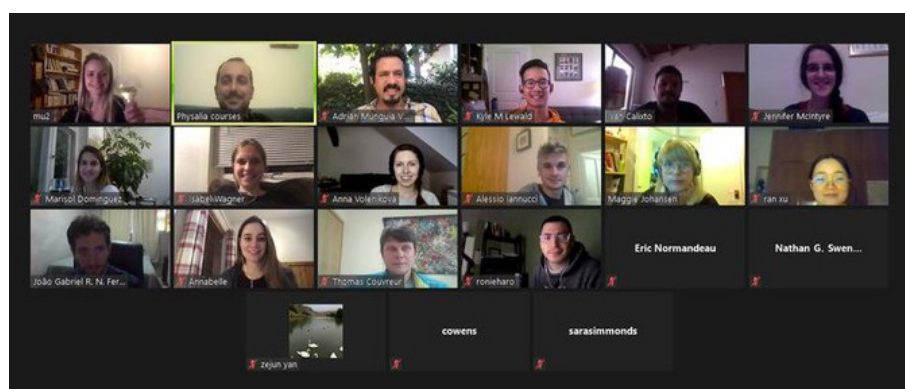
The organisers successfully applied to become a Sectional Interest Group (SIG). To know more, please, check our website at [https://genetics.org.uk/events\\_categories/sectional-interest-groups](https://genetics.org.uk/events_categories/sectional-interest-groups).

The Genetics Society Training Grants are available to enable members to go on short training courses in the area of Genetics research. In this issue, we have reports from Annabelle de Vries, Emma Kenyon, Hollie Marshall and Marc Ciosi.

## Eukaryotic Genome Assembly Using PacBio and Hi-C by Physalia-courses

9th -14th of November 2020

Annabelle de Vries (University of Warwick)



The course on Eukaryotic Genome Assembly using PacBio and Hi-C by Physalia-courses, provides an introduction to *de novo* genome assemblies using PacBio data from raw data to a fully assembled genome.

In five days, course leaders Dr. Marcela Uliano-Silva, Senior Bioinformatician at the Wellcome Sanger Institute, and João Gabriel Ferreira, Biophysics PhD candidate at the Universidade Federal do Rio de Janeiro and Bioinformatician analyst at Bio Bureau Biotechnology, taught us about the basics with theoretical lectures, explanation of the bioinformatics and how to interpret the analyses. We were also given hands on assignments in groups to assemble the genome of a given organism. In my case, *Vanessa atalanta*, or the red admiral.

With longer and higher quality reads it becomes easier to map whole genomes, which can be used for all kinds of research. One sequencing technique is Pacific Biosciences (PacBio) HiFi sequencing, which allows for whole genome high accurate long reads which can be used for *de novo* genome assembly. PacBio also provides Long Reads (CLR) sequencing which is less accurate than HiFi but provides even longer reads. Both techniques and how to use these datasets for genome assembly were discussed during the course.

The week started with a short introduction to everyone and their work, followed by a lecture introducing concepts of genomics and more specific difficulties of genome assembly, like repeats. There was also a lecture to

introduce working with Linux to prepare us for the bioinformatics during the week. The second day was about K-mer analysis, which can help determine genome size, repeat content and heterozygosity of the sequenced data. We performed the K-mer analysis on our group organism. This was followed by another lecture on the several programs and tools that can be used for the genome assembly, such as Canu, Falcon, Flye. During the practical assignment we used HiFi data of *Vanessa atalanta* and used both HiCanu and HiFiasm to assemble the genome and observe the differences in the assemblies. On the third day, we discussed purging of haplotigs and covered polishing of the genome. To evaluate purging and assembly quality, the tool Merqury was used. The next day, we learned how to scaffold genomes with Hi-C using the software SALSA2. This was followed by polishing of the assemblies and curation of the genome using PacBio CLR data. The outcomes for our groups were presented on the final day. It was interesting to see the findings of other groups and how they explained figures and findings in their own words.

This course was originally to be held in Berlin, but it was held online with 23 participants on a Zoom call working



from different countries and institutes across the world. The course leaders were very excited to get us all involved and to network, even though it was an online event.

Working in smaller groups allowed us to interact more and to engage in the course. Whenever someone had difficulties with understanding certain aspects, we were able to discuss this and go through it for our group's dataset. Whenever we had questions, the course leaders were very helpful in guiding us through the bioinformatics and answering our questions on interpreting our results. We were also encouraged to try to analyse our own data, with time for questions about our own research which was often discussed in the group to show examples of how you might cope with similar problems or datasets.

For my research I will be looking at DNA from historical plant collections. DNA degrades under poor environmental conditions and over time, which makes it more fragmented and deaminated. We therefore need to have a reference genome for the assembly of the short-fragmented DNA sequences. There is no genome sequenced for a relative species, so I will do a de novo genome assembly of the species I'm working on. The course leaders were very helpful in advising about the PacBio sequencing technique I would probably want to use and provided insights in performing the assembly of the plant genome.

It was a great course with excellent course leaders who inspired the participants to actively participate. The course ended with a virtual drink and question moment, reflecting on what we discussed over the week. We went through a lot of information, but it fully prepared us to perform genome assemblies using our own data.

If you are interested in a genome assembly using PacBio data, I highly recommend this course.

## WGCAC Next Generation Sequencing Bioinformatics Course

19th -24th October 2020

Emma Kenyon (University of Sussex)

This course was aimed at scientists who want to analyse their own Next Generation Sequencing (NGS) results. The course contained both lectures and hands on practical learning. The latter of these saw participants split into groups and put into breakout rooms with tutors. The course started with a broad overview of the types of NGS platforms available and the type of sequencing experiments each are best suited for. The course then goes on to teach the participant how to use Linux as all the packages used on the course are open source allowing the scientist to perform bioinformatics on their data without costly software. This is particularly important for myself as I am trying to get the best value for money for the publicly funded charity that awarded my fellowship. The course then goes on to discuss data formats and repositories before teaching participants to do read alignments, variant calling, SV calling, RNA-Seq and ChIP-Seq. Finally on the last afternoon the participants got the chance to participate in a group project aligned to their particular research interests. In my case this was looking at differential gene expression in RNA-seq data from wild type and mutated organisms. This gave participants the chance to go through everything they had learned and create a pipeline to analyse the NGS data they had been given.

The course was originally a residential at the Wellcome Trust Campus but was switched to a virtual course. This had advantages over being on site in that each participant had a copy of all the software they needed

for the course installed on their own computer as a virtual machine to be used at a later date rather than using the computers at the venue. This means the participants own computers are now set up to analyse their NGS data in future. The disadvantage of not being on site was that it was more difficult to have discussions with other participants and the instructors during the breaks and of course no catering. As the participants on the course were from all over the world and from diverse biological backgrounds, this had the potential to result in some very interesting conversation, but this was reduced due to the virtual format and with participants being in different time zones.

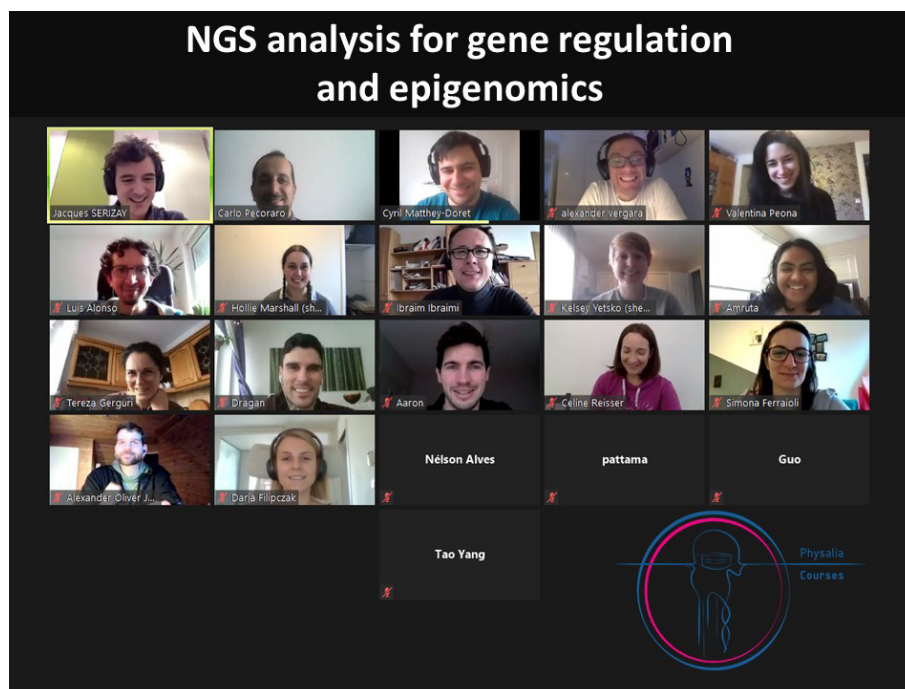
In addition to the lectures and hands on practical sessions the course also had two research lectures with invited speakers giving talks on how they used NGS in their research. The first of these was from a research scientist at EMBL-EBI who is using NGS to research mammalian regulatory evolution. The second was from a clinical scientist based at the Cambridge University Hospitals NHS trust who gave a case study-based lecture about using NGS to understand microbial genomics in clinical practice. I thought both talks were very good for the audience which was made up of both fundamental and clinical scientists and some such as myself who have interest in both areas.

In summary I thought the course was excellent and would recommend it to anyone who plans to analyse their own NGS data.

# NGS Analysis for Gene Regulation and Epigenomics

11th -15th January 2021

Hollie Marshall (University of Edinburgh)



The Next Generation Sequencing (NGS) analysis for gene regulation and epigenomics course covers the field of regulatory genomics. Specifically, training includes the bioinformatic analysis of ChIP-Seq, ATAC-Seq and Hi-C data. This course was due to be held at the Freie Universität in Berlin however it was moved online this year due to the COVID-19 pandemic.

Each day consisted of around 1/3 lectures and 2/3 practical sessions (along with a good number of needed coffee breaks). The lecture component was particularly informative and aimed to give a background on the types of data

we would later analyse in the practical session. This background information consisted of a historical look at the techniques, showing how they were developed and have evolved over time along with an intricate description of the wet lab methods used to generate the data. This was super useful as it allowed participants to fully understand the nuances of the data which is vital for carrying out an appropriate analysis.

After the background lectures we moved to the practical components. Using high performance computing resources from Amazon we could access pre-made bioinformatic environments

to analyse example data. The course lecturers walked participants through each step of the analysis, giving custom recommendations and explaining the purpose of every line of code. As the lecturers came from non-model organism backgrounds, they were able to point out analysis considerations throughout for non-model organism data, which was a particular highlight.

Throughout the course participants also had access to a slack group. This enabled us to ask questions outside of the 1-7pm course times which the lecturers could answer for us the next day. They also provided a google doc for us to introduce ourselves and swap emails to try to emulate the networking we would have done in person.

In terms of course content we covered Hi-C analysis, ChIP-Seq analysis, ATAC-Seq analysis, Snakemake, Nextflow, key Bioconductor packages for NGS analysis and how to integrate multiple omics datasets. For Hi-C we learned all of the steps from raw data to interaction map creation (who knew paired ended data had to be aligned in single-end mode!) along with the identification of chromatin loops and TADs/interaction domains.

For ChIP-Seq we discovered the pros and cons of using a pre-designed Nextflow pipeline. Nextflow pipelines are simple to use, you can download a pipeline and 'plug in' your raw data. The pipeline then carries out all steps from quality control to ChIP peak calling.

Whilst these pre-made pipelines make analysis simple and provide reproducible outputs, they aren't always the best idea when working with non-model organism data. This is because these pipelines may be designed for use with, say, mammalian data which will need different parameter settings compared to insect data.

We also learnt how to make our own easy to use reproducible pipelines which would carry out a similar function to a Nextflow pipeline (i.e. you just give the pipeline an input file) but which contain our own custom analysis. Snakemake provides a simple workflow management system which allows the user to define blocks of analysis (which can be python, R or bash) within a single file. There are loads of benefits to using Snakemake and I would highly recommend anyone who carries out any type of bioinformatic analysis to look into it.

The last large component of the course was ATAC-Seq analysis. For me personally this was the most informative part of the course. It turns out there are a plethora of considerations needed when analysing ATAC data and a simple out of the box solution will definitely not work for everyone. The biggest thing I have learnt is that choosing the right peak caller is vital. The standard MACS2 peak caller works by defining peaks based on coverage differences whereas the YAPC peak caller uses peak shape. Whilst YAPC is more sensitive and generally better when using heterogenous data (i.e data from the whole body of an organism), if you don't have deep enough coverage it can give a lot of false positive calls.

As a non-model organism biologist working on multiple arthropod species (from bumblebees to daphnia to mealybugs) it is absolutely vital I fully

understand the nuances of the different data types I work with. There is no 'one size fits all' pipeline which can give biologically meaningful results for non-model species. This course has helped me understand these data types meaning I can now make informed analysis decisions for my projects. I have also come away with some extra skills in Snakemake which will make my code more accessible to others. As someone who is passionate about open and reproducible science this was a big bonus. Thank you very much Genetics Society for the grant to attend this course, I now have the confidence to tackle my data knowing I'm making the right analysis decisions!

# Analysis Of Genetic Association Studies

Online course. 11th -15th January 2021

Marc Ciosi (University of Glasgow)

Huntington disease (HD) is an autosomal dominant neurodegenerative disorder caused by the expansion of a genetically unstable CAG repeat in the *HTT* gene.

Longer alleles are associated with more severe disease with inherited CAG repeat length variation accounting for ~50% of the variation in the age at onset. The expanded CAG repeat

is genetically unstable in the soma. Somatic instability is allele length- and age-dependent and highly expansion-biased.

I have recently developed a highly sensitive massively parallel sequencing approach to genotype HD alleles and quantify the very low levels of somatic expansions associated with expanded HD alleles in blood DNA (Ciosi et al.

2018 *Protocol Exchange*). I am currently applying this approach to quantify somatic expansions in large numbers of HD patients for whom detailed clinical data are available.

Using that approach I have recently shown that the amount of somatic expansions of HD alleles is associated with HD severity (Ciosi et al. 2019 *EBioMedicine*). The next phase of my

project will be to perform genome-wide association studies (GWAS) to identify the genetic modifiers of somatic expansion, that I predict will also be modifiers of HD disease severity. For this project, I needed to improve my theoretical and practical knowledge in relationship with GWAS.

To support me in this, the Genetics Society Training Grant provided funding to enable me to attend the online course “Analysis of genetic association studies” organised by the department of health data science of the University of Liverpool at the end of February. The four-day course provided guidance on how to undertake the statistical analysis of a genetic association study, with a particular focus on GWAS. It provided an overview of the key statistical issues to be aware of when analysing genetic association studies, and an introduction to software for conducting the analyses. The course was structured to include a combination of short lectures and computer practicals to ensure that attendees gain hands-on experience of analysing genetic association datasets.

The main topics covered by the course were the following:

1. Introduction to Linux and R
2. Introduction to format of genetic data and technicalities associated with the use of large files.
3. SNP calling
4. Genotype quality control
5. Analyses of association
6. Population stratification
7. Genotype imputation
8. Meta-analysis

The course has not only improved my ability to use genome-wide SNP data in my current research but has also given me the practical basis to confidently plan future GWAS.

The skills I acquired on this course are directly applicable to my project identifying the trans-acting genetic modifiers of somatic expansion of HD alleles and disease severity in HD which will hopefully help identify novel therapeutic targets for what remains an incurable and devastating disorder. I am looking forward to sharing my new

expertise with other members of my research group as well as collaborators who are investigating the genetic modifiers of somatic expansion in related repeat expansion diseases.

I am very thankful for the financial support granted by the Genetic Society to help expand my skills set in genetics.

To finish with I would like to thank all the organisers, speakers and tutors that have made that week a great week of training and learning:

Prof Andrea Jorgensen  
(University of Liverpool)

Dr Anna Auer-Fowler  
(University of Liverpool)

Dr Ravi Girikematha Shankar  
(University of Liverpool)

Dr James Cook  
(University of Liverpool)

Prof Andrew Morris  
(University of Manchester)

*The skills I acquired on this course are directly applicable to my project identifying the trans-acting genetic modifiers of somatic expansion of HD alleles and disease severity in HD which will hopefully help identify novel therapeutic targets for what remains an incurable and devastating disorder.*



*These reports are from researchers who the Genetic Society has funded (up to £1500) to undertake a field-based genetic research project, the results from which would be suitable for publication in the Society's journal Heredity.*

# Fieldwork at Lake Xochimilco: eDNA metabarcoding for Mexico's iconic wetland freshwater fauna

November 2020 - April 2021

Alejandro Maeda-Obregon (University College London)



Mexico, the southernmost country of North America, is the world's 2nd in ecosystem diversity and home to more than 10% of the planet's biodiversity. Mexican aquatic systems are remarkably diverse in freshwater species assemblages, with amphibians and freshwater fish presenting high degrees of endemism of 60% and

49%, respectively. Alarmingly, more than half of Mexico's amphibian species and 40% of freshwater fish species are registered in IUCN's Red List Threatened categories, with the country's wetland ecosystems considered severely impacted and imperilled.

An example of the delicate ecological situation is the Trans-Mexican Volcanic Belt ecoregion, a volcanic mountain range with high-altitude plateaus located in the country's centre, thriving with endemic terrestrial and freshwater fauna. It is also the winter home to migratory species like the Monarch butterfly. The endorheic Valley of Mexico Basin is located in one of the central plateaus, a valley with important biological diversity and one of the world's largest metropolis: Mexico City.

Originally, a lake complex dominated the Valley of Mexico Basin, consisting of five shallow water bodies (Zumpango, Xaltocan, Texcoco, Xochimilco and Chalco). With the establishment of the Spanish Colony in 1521, the lake complex underwent a continuous draining process to allow floods control and make available land for construction. These modifications continued through several centuries, with most of the lakes completely gone by the 20th century. One of the few remnants is Lake Xochimilco (LX), a World Heritage Site and Ramsar



Wetland.

LX is a complex system of canals and lagoons where ancient traditions and customs still survive, home for endemic amphibians and fishes such as Tlaloc's leopard frog and the mexclapique fish. And among them, the axolotl is worldwide famous! The axolotl is a paedomorphic salamander, sexually maturing without losing its characteristic gills (and smile!), permanently living underwater and capable of regenerating almost any part of its body. Regrettably, different ecological pressures such as water pollution, uncontrolled urban expansion, and human-mediated introduction of alien fish species have all contributed in combination to the severe native populations decline over the last decades. According to Dr Luis Zambrano (a collaborator), LX is a highly fragmented and heterogeneous system, causing traditional survey methods not to be suitable anymore. Therefore, my PhD aims to employ an Environmental DNA (eDNA) framework to assess the current status of native biodiversity and elucidate their spatial interactions with exotic species and water pollutants.

The Heredity grant facilitated my fieldwork during the winter months in Mexico City. I spent evenings (axolotls are more active at this time of the day) collecting water samples at different spatial points across LX paired with traditional fishing surveys and measuring water parameters. During the survey, field technician Ana Soler identified caught freshwater specimens and took small tissue samples from specimens to build the genetic reference database (necessary for assigning eDNA sequences at species level via a bioinformatics pipeline), with native specimens subsequently released. As the field conditions during the night were not safe, I transported the water samples in a coolbox back to the accommodation. Filtration was performed using a peristaltic pump and Sterivex filters that were subsequently preserved using absolute ethanol molecular grade and frozen. Organic pollutants and heavy metals were measured using a photometer.

After extracting the DNA from the water filters and using universal primers to amplify mtDNA markers, I will build up a genomic library using next-generation sequencing

and bioinformatics pipelines for metabarcoding analysis. In combination with the water parameters and pollutants data, I will then use different approaches to investigate the biological and ecological relationships of native fauna. Additionally, I will employ a novel eDNA approach using a hypervariable mtDNA marker to detect wild axolotl populations' genetic structure.

With the pressure of the biodiversity crisis both in the country and the world, I hope that the project's results will benefit the analysis and conservation of LX's native fauna through a novel and non-invasive eDNA approach. Findings can potentially influence the construction location of new refuges for native biodiversity and conserved by locals. Also, the novel population genetics approach will be crucial to the conservation of the axolotl while expanding the current eDNA framework's frontiers!

Doing research fieldwork back in my country is as exciting as it's challenging. The mixed conditions of being out in the wetland while also getting through the chaos and traffic of Mexico City can complicate the streamlining of protocols dealing with eDNA. The ongoing COVID-19 pandemic interrupted fieldwork and access to laboratories while restricting social interaction with our collaborators. Nevertheless, despite the adverse circumstances, there were huge rewards, such as establishing collaboration with locals while strengthening relationships with our Mexican peers.

I want to thank the Genetics Society for the funds and making the fieldwork feasible and to my supervisors, friends and collaborators from Mexico for their ongoing support during these challenging times.

## The role of habitat selection and local adaptation in the population structure of an apex predator

October - November 2020

Isabel Salado (Doñana Biological Station, Spanish National Research Council)

Understanding the microevolutionary processes that generate genetic population structure among demes within a species is essential in order to understand diversification processes and to address specific conservation issues.

Population structure depends on the extent of gene flow, which in turn is related to dispersal capabilities of a species. Large mammalian carnivores are animals with high mobility, able to overcome topographic barriers and long geographical distances. The grey wolf (*Canis lupus*) is one of the most mobile large carnivores, and may disperse over distances up to 900 km. In addition, wolves can occupy very diverse habitats, from the Arabian desert to the frozen Arctic, taiga forests or Mediterranean scrublands. However, despite this amazing dispersal capability and generalist behaviour, previous studies have reported genetic differentiation among wolf populations at a continental scale, probably associated with differences in habitat characteristics. However, it is still not clear whether ecological processes affect wolf population structure at a finer scale. If dispersing wolves favour habitat types similar to those where they were born, dispersal and population dynamics would be biased and fine population genetic structure could emerge, as suggested by the “Matching Habitat Choice” hypothesis.



Isabel Salado taking a picture of a wolf scat in the middle of a trail

In Europe, wolf population numbers have recovered in the last decades.

However, the Iberian wolf population, in the southern limit of its distribution, is not expanding and remains isolated from other European wolf populations since the mid-19th century or earlier. Iberian wolves are distributed through a diverse range of habitats, from well preserved mountain habitats with

abundant wild ungulates, to highly humanized agricultural areas with very limited forest cover. We aim to understand the ecological processes that are limiting dispersal of Iberian wolves through assessing their current genetic population structure.

To achieve this, we are performing a non-invasive genetic monitoring. We have been collecting fresh faeces from

*Large mammalian carnivores are animals with high mobility, able to overcome topographic barriers and long geographical distances.*





Wolf scat on top of a broom (*Cytisus* sp) at 1650 m above sea level in Avila (Central Spain)



Fieldwork team during the Coronavirus pandemic: from left to right – Carles Vilà, Alberto Fernández, Laurentino García and Isabel Salado

different wolf packs (family groups) across most of the distribution area of the species in Iberia. Wolves use paths and trails to move through the territory and tend to leave scats at intersections as marking behaviour. We have been surveying these places in different areas in Spain and collected just a small portion of the excrement to avoid interfering with the marking behaviour of the species.

In October and November 2020 thanks to funds from the Heredity Fieldwork Grant I was able to visit two additional field areas along the edge of the main wolf distribution range in the Iberian Peninsula, the Central System Mountains. In this region, habitat is very patchy; wild mountain areas are interspersed with areas heavily used by livestock. Also, wolf packs are scattered and fluctuating in numbers every year due mainly

to anthropogenic disturbances and persecution. To collect the samples, we needed to drive long distances to check several areas with and without recent evidences of wolf presence. Fortunately, we successfully sampled at least 12 potential wolf packs, collecting a total of 25 wolf-like faeces.

Back in my institution, I have already started to extract the DNA from some faecal samples. To genotype these faeces, I will use a multilocus genotyping protocol based on next generation sequencing approaches that I have been optimizing as part of my thesis. A better understanding of the mechanisms controlling gene flow between wolf populations will shed light on possible dispersal and colonization patterns, which can help understand what are the limitations for the expansion of the Iberian wolf population at a time when wolves

are expanding across Europe. This knowledge can be of great importance in the design of landscape genetics models that might be applied in the development of conservation actions for this apex predator.

I would like to thank the Genetics Society for the funding which made this fieldwork campaign possible. I would like to thank my supervisors, Dr Carles Vilà and Dr Jennifer A. Leonard, for their guidance and for convincing me I was capable of leading this work. I also express my immense gratitude to my fieldwork colleagues, particularly Dr Alberto Fernández, Laurentino García and Marta Portolà, who helped me to collect the samples and shared together fascinating discussions which will be very useful to interpret the final results of this work.



*This studentship supports vacation research by undergraduate geneticists interested in gaining research experience in any area of genetics by carrying out a research project over the long vacation, usually prior to their final year. In this issue, we have reports from Ersi Christodoulou.*

# Can Artificial Intelligence Be Used to Solve Today's Biological Problems?

Ersi Christodoulou (University of Cambridge)

**During summer I had the opportunity to be part of a very exciting project in the lab of Dr. Ben Steventon at the Department of Genetics, Cambridge.**

## BACKGROUND OF PROJECT

The lab is focused on studying the Presomitic Mesoderm (PSM), the tissue that undergoes somitogenesis and leads to the formation of the vertebrae column. It has been well characterised in the past how differences in gene expression exist along the length of the zebrafish PSM, with *tbxta*, *tbx16* and *tbx24* expressed from posterior to anterior. The lab aimed to characterise the interactions between these three genes as well as FGF and Wnt, by also finding the numerical values of 24 parameters, where each parameter describes one interaction. Doing this would give insights on how gene expression is coordinated with cell movements in this complex developmental process. They started using a live modelling approach, where a Markov chain Monte Carlo model was used to predict the values of these

parameters. In order to further get an unbiased fitting of these parameters directly to 3D image data they then decided to use a neural network approach.

The way that a neural network works is that it is initially trained using simulated data with random parameters in order to learn how to map a set of parameters that can lead to a specific pattern. Then this is evaluated using real data to see if the neural network can predict those parameters from a given pattern. **The handling and analysis of that real data was where my project was focused.**

## METHOD AND DATA CREATION

After initial training on IMARIS and familiarisation with the shape of the PSM, training in creating cell tracking was undertaken. A movie of a zebrafish tailbud growing over time is the initial stage and the steps taken to properly create tracking data are:

- **Tracking:** Parameters such as the tracking algorithm and maximum distance, have to be inputted in order for IMARIS to predict how these spots will move at every frame and create a track for every cell.
  - **Filtering:** At each frame, tracks that were not part of the PSM would have to be removed manually.
  - **Removal of somites:** As somitogenesis is ongoing, every few frames new somites would form. These would also have to be removed manually as this is now a different tissue and those cells' movements are no longer representative of the PSM and thus relevant to the project
- The final product would be a movie where the PSM is getting shorter and thinner over time and this data would be exported to an excel file.
- However, the target of the project was to create variation in these tracks. In order to do so, 3 different parameters were changed.
- **Segmentation:** Here it has to be decided what size each spot (which represents a nucleus) will be and the tailbud is segmented.

- Firstly, the initial segmentation varied from 4 to 5 to 6 microns and this led to a difference in the initial 3D pattern of the PSM. (Figure 1)
- The next two variables would specifically affect the way that the cells would move:
- Choice of an algorithm by which IMARIS would predict how the cell would move in the next frame. When Brownian motion was selected, the program assumed that the cell can practically move into any direction, while when Autoregressive motion was selected, then the cell would be more likely to keep moving in the same direction as it did before. (Figure 2)
- The maximum distance is the distance that a cell is allowed to move from its previous position in Brownian motion or from its expected position in Autoregressive motion. A range of values was also employed here from 5,6,7.5 and 10 microns. (Figure 3)

The reason why variation was required was because this exists in nature, whether this is between different developmental stages or different zebrafish. It is thus important for a neural network to be able to cope with this in cell movement. It is interesting to think of this variation as a list of different handwritten digits. All 5s or 6s will look different, yet a handwriting recognition neural network should still be able to identify them as a 5 or 6.

## OUTLOOK

Experiencing the significance of interdisciplinarity in biology and how data science and mathematics can be used to solve an important biological problem was eye opening. Learning to work across boundaries has now

become a personal mission that will allow me to evolve into an independent and skilled scientist. The way that the Genetics Society has chosen to

support students during the summer allows others like me to discover their true scientific interests and start their journey to a research career.

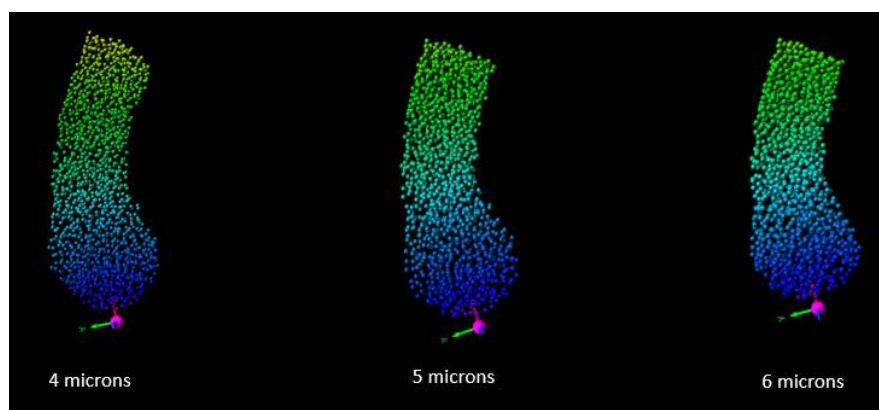


Figure 1: Variation in segmentation size

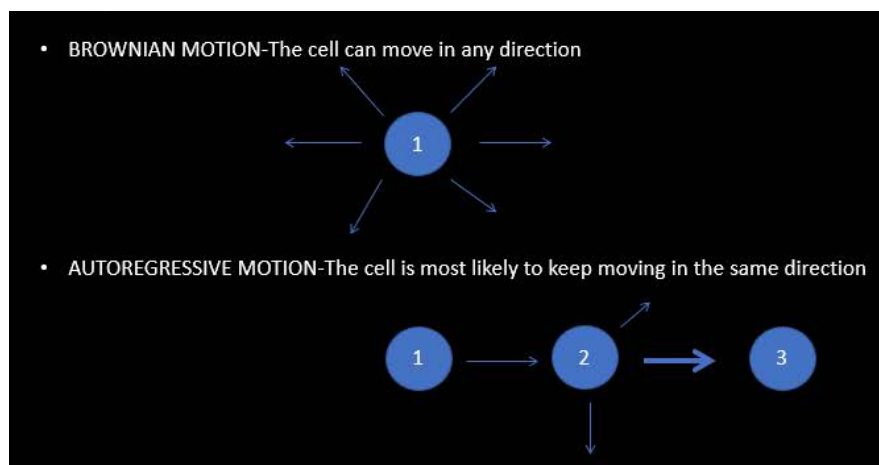


Figure 2: Variation in the algorithm predicting cell movement - Autoregressive vs Brownian

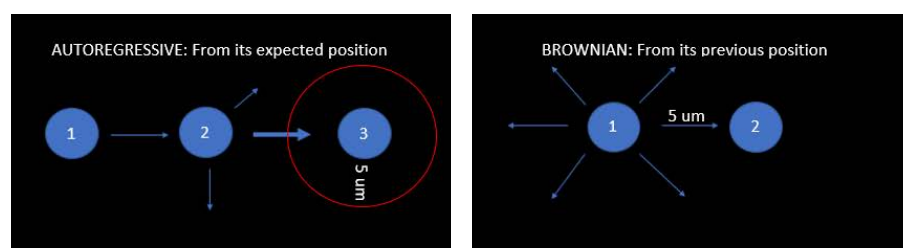


Figure 3: Variation in maximum distance

To apply for any of our grant schemes, instructions and downloadable funding application forms are available from the drop down Funding tab on the Genetics Society website [www.genetics.org.uk](http://www.genetics.org.uk)

## One-off Meeting Sponsorship

### Purpose

Sponsorship of genetic themed meetings not organised directly by the Genetics Society.

The Genetics Society receives several requests from members each year to sponsor meetings in the field of genetics. These meetings are usually one-off meetings with an ad hoc organising committee and may be partly sponsored by another Society. The guidelines below indicate a review process for applications and the conditions that must be met for the award of Genetics Society sponsorship.

Please note: these grants are to be used towards the organisation of a meeting and not towards the attendance of a meeting, but should include a Carer's Award to allow those with responsibilities to attend. Meetings should be based in, or have strong links with, the UK.

### Review of applications

Members may apply at any time of the year and we encourage members to submit at least three months in advance of their event to allow the application to be reviewed. Applications will be sent to the Scientific Meetings Secretary for review at the end of each month.

### Apply for a Genetics Society Grant

Once you have logged in to the mySociety membership portal, please select "Me and the GS" followed by "Grants" from the options at the top of the page, and then choose the one-off meeting sponsorship award. If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

The application will be circulated to the full committee for review. The review will cover suitability of the meeting for Genetics Society sponsorship and level of support requested. The committee will be asked to respond within two weeks and the Society aims to respond to requests within four – six weeks.

### Conditions of sponsorship

Several levels of sponsorship are possible. We can sponsor a specific Genetics Society lecture for a few hundred pounds or a Genetics Society session for up to about £1,000. As major sponsor of an entire meeting we can potentially sponsor several thousand pounds but the costs need to be well justified. The major sponsorship levels for the last 12 months have been between £1,000 and £2,000. Applications should include a figure to cover the anticipated uptake of the Carer's Award. The committee judges all applications and even if approved, may not agree to fund the full amount applied for.

### Diversity guidelines

Ensure a good balance between established and new investigators on the Speaker list. Ensure that there is an attempt for broad geographical representation where possible

### Publicity

The sponsorship of the Genetics Society must be mentioned in all pre-meeting publicity (e.g. posters, flyers, website) and in the meeting programme. If the Genetics Society is the major sponsor the meeting should be advertised as a "Genetics Society-sponsored meeting".

Use of the Gen Soc logo in publicity materials.

Details of the programme of the meeting and registration forms should be sent as far in advance as possible to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk), for inclusion in the Society's newsletter and on the website.

The meeting organisers agree to make details of how to apply for Genetics Society membership available to non-members attending the sponsored meeting. Meetings that receive maximum sponsorship will be expected to offer a discounted registration fee to Genetics Society members, to encourage non-members to join the Society at the same time. New members may then attend at the discounted rate, once confirmation of their application for membership of the Genetics Society has been received from the Society's Office.

### Meeting Report

A short report on a meeting that receives sponsorship of £1000 or more, for possible publication in the newsletter and on the website, should be sent to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk) within one month of the conference taking place.

### Use of Sponsorship

Genetics Society sponsorship may be used at the organiser's discretion, but budget travel and accommodation options should normally be insisted upon. Any unused grant should be returned to the Genetics Society. The Society will not be responsible for any losses incurred by the meeting organisers.

An invoice for the grant awarded should be submitted to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk). The grant may be claimed in advance of the meeting and no longer than one month after the meeting.

## Sectional Interest Groups

### Purpose

Sponsorship of research meetings on particular themes. Funding is available for genetics research communities who wish to run regular series of meetings. Current examples include Arabidopsis, E-ACGT (Edinburgh Alliance for Complex Trait Genetics), POP Group (Population Genetics Group) and the C. elegans Group. Meetings should be based in, or have strong links with, the UK.

- Members may submit Sectional Interest Group (SIG) applications at any time of the year, and we encourage submissions at least three months in advance of the proposed event to allow the application to be reviewed. Applications will be sent to the Scientific Meetings Secretary for review at the end of each month.
- The application will be circulated to the full committee for review. The review will cover suitability of the meeting for Genetics Society sponsorship and level of support requested.
- The committee will be asked to respond within two weeks and the Society aims to respond to requests within four – six weeks.

**All applications for funding should be submitted using the online application form:**

### Apply for Sectional Interest Group Funding

Once you have logged in to the mySociety membership portal, please select “Me and the GS” followed by “Grants” from the options at the top of the page, and then choose the Sectional Interest Group award. If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

The award of Genetics Society support will be subject to review of applications by the committee and subject to the following conditions:

### Publicity

The sponsorship of the Genetics Society must be mentioned in all pre-meeting publicity (e.g. posters, flyers, website). It should also be acknowledged in the meeting programme booklet. It is understood that wherever possible, the meeting should be advertised as ‘A Genetics Society Meeting’. However, where the Society’s financial contribution support is only partial, and where this formula of words would conflict with the interests of other sponsors, it is acceptable for the meeting to be advertised as a ‘Genetics Society-Sponsored Meeting’.

### Use of the Gen Soc logo in publicity materials.

Details of the programme of the meeting should be made available to all Genetics Society members via the Society’s newsletter, and an electronic copy should be sent as far in advance as possible to the newsletter editor, at the latest by the advertised copy date for the newsletter preceding the close of registrations for the meeting. The same details will appear on the Genetics Society website. This information should include the programme of speakers, the topics to be covered, plus details of how to register for the meeting.

If the meeting is advertised on the Internet, then a link to the Genetics Society website ([www.genetics.org.uk](http://www.genetics.org.uk)) should be included.

### Diversity guidelines

A brief statement, indicating how you have addressed the diversity guidelines or explain why you could not conform to the guidelines will be required.

Appropriate representation of women as invited Speakers is required, and will be monitored by the Society. Organizers must Ensure a good balance between established and new investigators on the Speaker list. Ensure that there is an attempt for broad geographical representation where possible

### Meeting Report

A report on the meeting, once it has taken place, should be submitted for publication in the newsletter, which is the official record of the Society’s activities. This should be sent as soon as possible after the meeting to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk), and should include brief factual information about it (where and when it took place, how many people attended and so on), together with a summary of the main scientific issues covered.

### Use of Funds

Genetics Society funds may be used to support speaker travel, accommodation, publicity or any other direct meeting costs, at the organisers’ discretion, but must include a carer’s award to allow those with responsibilities to attend. It is understood that budget travel and accommodation options will normally be insisted upon. Any unused funds should be returned to the Society. The Society will not be liable for any financial losses incurred by the meeting organisers. Any profits should be retained solely for the support of similar, future meetings, as approved by the Society.

A written invoice for the agreed amount of Genetics Society sponsorship should be forwarded to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk), no later than one month after the meeting date. Funds may be claimed in advance of the meeting, as soon as the amount of support has been notified in writing.

### Registration Fees

Meeting organisers may levy a registration charge for attendance at the meeting as they see fit. However, it is understood that Genetics Society members will be offered a substantial discount, so as to encourage non-members wishing to attend to join the Society at the same time. The meeting organisers agree to make available to non-member registrants full details of how to apply for Genetics Society membership, such as appear on the website and in the newsletter, and may charge such persons the same registration fee as charged to members, upon confirmation from the Society’s Office that their application and remittance or direct debit mandate for membership fees has been received.

### Other Sponsorship

The meeting organisers are free to apply to other organisations for sponsorship of the meeting, as they see fit. However, organisations whose policies or practices conflict with those of the Genetics Society should not be approached. In cases of doubt, the officers of the Genetics Society should be consulted for advice.

### Continued Support

For those groupings holding their first such meeting with Genetics Society support, it is understood that the Society’s support for future meetings of the series will be decided on the basis of the success of the first meeting, including adherence to all of the conditions listed above. The first meeting is hence supported on a pilot basis only.

### Nominated Meeting Co-ordinator

The meeting organisers will nominate a responsible person who will liaise with the Genetics Society on all matters relating to the meeting, and whose contact details will be supplied to the Society’s Office. This person will inform the Society if he/she resigns or passes on his/her responsibility for the meeting or series to another person, whose contact details shall also be supplied.



## Heredity Fieldwork Grants

### Purpose

to support field-based genetic research.

Grants of up to £1,500 are available to cover travel and accommodation costs associated with a field-based genetic research project.

The research should produce results that would typically be suitable for publication in the Society's journal *Heredity*.

### Eligibility Criteria

- All students are eligible to apply for this grant immediately after they join the Genetics Society.
- Other applicants (i.e. PI's and Co-I's) must have been members of the Genetics Society for at least one year before applications can be accepted.
- Funding for students is primarily targeted to those at post-graduate level. However, in exceptional circumstances we will consider applications from students who are required to complete a fieldwork study in their final undergraduate, or MSc by Research year.
- Applicants other than PI's and Co-I's are required to submit a supporting letter from their supervisor who must be a current Genetics Society member.
- A maximum of one Heredity Fieldwork Grant per individual per two years will be awarded.
- Only one application per research group will be funded in any one year.
- The applicant must be completing the fieldwork themselves.
- Recipients of these grants must submit a short report within two months of completion of the project that may be included in the Genetics Society newsletter.
- These grants are open to all members of the Genetics Society; however, priority will be given to applications from those with a UK base wishing to undertake fieldwork in the UK or overseas, and to non-UK-based students wishing to undertake fieldwork in the UK.

### Eligible costs:

The scheme is intended to support costs directly associated with the field component of a research project, such as travel and accommodation expenses, consumables for pre-processing (e.g. sample collection, storage, RNA extraction).

### Costs associated with lab procedures, e.g. downstream molecular analysis or equipment are not eligible.

The scheme is not intended to cover the costs of salaries for those engaged in fieldwork, or to fund attendance at conferences. However, it is recognised that in some circumstances, e.g. for health and safety or practical reasons, an assistant, and/or particular equipment may be required. If a field assistant is required, their expenses will be considered if they are a member of the Genetics Society.

Equipment essential for the gathering of data in the field may be eligible, but should not total more than one third of the funds applied for.

### How to apply:

Applications should be made online via the Genetics Society Grants application site. Once you have logged in to the mySociety membership portal, please select "Me and the GS" followed by "Grants" from the options at the top of the page, and then choose the Heredity Fieldwork Grant.

If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

Deadlines are quarterly (midnight on: 1 February, 1 May, 1 August, 1 November).

### Apply for a Heredity Fieldwork Grant

A complete application should contain the following:

- A completed online GS Funding Application Form.
- If the applicant is a postgraduate, undergraduate, or MSc by Research student, a supporting statement from their supervisor, who should also be a member of the Genetics Society, is required. This statement should be uploaded via the online application form before the deadline.

The Genetics Society aims to notify the decision within one month of application deadlines. However, applications should be submitted at the earliest opportunity, and at least 3 months in advance of the start date of the fieldwork.

### Join the Genetics Society

The Heredity Fieldwork Grant is funded by income from the journal *Heredity*

## Genes and Development Summer Studentships

### Purpose

To support vacation research by undergraduate geneticists.

Grants are available to provide financial support for undergraduate students interested in gaining research experience in any area of genetics by carrying out a research project over the long vacation, usually prior to their final year.

Awards will be made to the host institution. The studentship includes:

- Up to £750 to cover justifiable expenses incurred by the host laboratory.
- A stipend to cover 8 weeks subsistence during the studentship.

The student must be able to attend a workshop that will take place in Oxford, in early September 2021, providing an opportunity for all students to get together, discuss their findings, make new friends and start to develop their professional contact network. If necessary, a Carer's Award will be available to allow those with responsibilities to attend the Summer School.

Undergraduate students who wish to do vacation research projects are encouraged to seek a PI to sponsor them and to develop a project application with the sponsor.

### Eligibility Criteria

- The project should be realistic and achievable by a student within an eight-week time frame for completion prior to the last week in August.
- Applications must be made by Principal Investigators (PI) at Universities or Research Institutes, NOT by the named student.
- Please note that only one application per lab group / per applicant may be submitted.
- The application must be for a named undergraduate student, preferably from another institute or university, and is not transferable.
- Both the PI and the named student must be members of the Genetics Society.
- Extension of honours projects or early starts for PhD students are not eligible.
- Recipients cannot hold these awards in conjunction with other summer studentships, i.e. summer studentships cannot be used to part-fund a project.
- There are no restrictions concerning the nationality of the student, and the student does not have to attend a UK university, nor does the studentship need to take place within the UK. However, students MUST attend the Summer School as follows:
- Students must be available to participate in the summer school held in Oxford, early September 2021.
- Travel within the UK to Oxford will be reimbursed.
- Travel to the UK cannot be funded.
- Accommodation, transport to the venue and meals will be included for the duration of the summer school.
- Students will be asked to write a short report (around 800 words) within two months of completion of the project that may be included in the newsletter.
- 

- Applications MUST include the following:
  - project outline
  - project plan (including student training needs)
  - student CV
  - student statement
  - reference letters

### How to apply

Once you have logged in to the mySociety membership portal, please select "Me and the GS" followed by "Grants" from the options at the top of the page, and then choose the Summer Studentship award.

If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

A panel from the Genetics Society committee will review applications including both information on the student and the proposed project. Feedback on unsuccessful applications will not be provided.

A panel from the Genetics Society committee will review applications including both information on the student and the proposed project. Feedback on unsuccessful applications will not be provided.

### Other conditions:

- Recipients cannot hold these awards in conjunction with other summer studentships, i.e. it cannot be used to part-fund a project.
- Students of these grants will be asked to write a short report (around 800 words) within two months of completion of the project that may be included in the newsletter.
- Students are expected to attend a workshop in early September, where they will report on their project and participate in group activities.

The Genetics Society Summer Studentship grants are funded by income from the journal Genes and Development

## Conference Grants

### Purpose

The purpose of these grants is to support the attendance of Genetics Society “junior scientist” members at conferences on research in Genetics. The scheme has two main streams: (A) to support attendance at meetings organised directly by the Genetics Society or sponsored by the Society as a Sectional Interest Group; and (B) to support attendance at non-Genetics Society meetings.

### Eligibility Criteria

- Scheme (A) is open to undergraduate, Masters and PhD students and to postdoctoral scientists within six years of their PhD viva. Scheme (B) is open to PhD students and postdoctoral scientists within six years of their PhD viva (but not undergraduate or Masters students). (Scientists who obtained their PhD more than six years ago are not eligible for these schemes.)
- Scheme (B) is open to members with a UK base wishing to attend conferences outside of the UK and to non-UK-based members wishing to attend a conference in the UK. We regret that we cannot consider applications from bases outside the UK for conference attendance outside the UK.
- Supervisors providing support letters must be current members of the Genetics Society and should include their membership number in the supporting letter. This supporting letter must be uploaded along with the online application before the deadline.
- Grant recipients will be asked to write a short report that may be published in the Genetics Society Newsletter.
- A maximum of one grant per two years will be awarded per applicant.

(A) Grants to assist with travel and accommodation (but not registration) costs to attend Genetics Society or Sectional Interest Group meeting. Grants up to £150 are available for travel and essential overnight accommodation to attend any of the Genetics Society’s own bi-annual meetings and those of our Sectional Interest Groups. The most economic form of travel should be used.

### How to apply

For Genetics Society and Sectional Interest Group meetings (e.g., Fly South West, GARNet, E-ATCG), applications should be submitted online before the registration deadline of the meeting.

Once you have logged in to the mySociety membership portal, please select “Me and the GS” followed by “Grants” from the options at the top of the page, and then choose the Junior Scientist Conference Grant award. If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

There is no limit to the frequency that grants can be awarded for attending Genetics Society meetings.

The supervisor’s supporting statement must be uploaded along with the online application before the deadline. Supervisors writing support letters must be current members of the Genetics Society and should include their membership number in the student application.

Carer’s Award. In recognition of carer’s responsibilities, an award of (up to) £60/day will be made available to enable members and selected speakers to attend Genetics Society scientific meetings and events. Awardees can spend this money as they think will best support their attendance. Applications can be made through the mysociety portal.

(B) Travel, accommodation and registration cost at other (non-Genetics Society) meetings.

Grants of up to £750 are available to attend conferences in the area of Genetics other than Genetics Society or Sectional Interest meetings. Applications should be submitted in time for one of our bi-monthly deadlines (1st day of February, April, June, August, October and December) and should be made by logging into your membership account. Once you have logged in, please select “Me and the GS” followed by “Grants” from the options at the top of the page, and then choose the Junior Scientist Conference Grant. Note that the conference you are applying for must take place AFTER the application deadline.

Up to three Conference grants per year will be co-sponsored by the Galton Institute and will provide up to £1,000. Applicants for a prestigious Galton co-sponsored award should request between £750 and £1,000 in support and explain how their work conforms to the mission of the Galton Institute. The Galton co-sponsored award is only open to registered PhD students who will take up the award before their PhD graduation date. If unsuccessful for the Galton co-sponsored award, applications will be automatically considered for a standard stream B grant for which a maximum of £750 can be awarded.

## Training Grants

### Purpose

To support attendance at short training courses.

Grants of up to £1,000 are available to enable members to go on short training courses in the area of Genetics research, e.g. those run by Edinburgh Genomics, MRC Harwell, and Wellcome Genome Campus. In some cases, longer courses or visiting another laboratory for training may be allowed. Eligible expenses include travel, accommodation, subsistence and tuition fees.

### Eligibility Criteria

- A maximum of one Training grant per individual per two years will be awarded.
- Only one application from any research group will be funded in any one year.
- Open to those with a UK base wishing to attend training courses within and outside of the UK and to non-UK-based students wishing to attend a training course in the UK. We regret that we cannot consider applications from bases outside the UK for training course attendance outside the UK.
- When a relevant course is available in the UK, a detailed explanation is required of why the applicant should be funded to attend a similar/ the same course abroad.

- Recipients of these grants must submit a short report within two months of completion of the project, for possible inclusion in the Genetics Society newsletter.

### How to apply

Applications should be made online via the Genetics Society Grants application site. Once you have logged in to the mySociety membership portal, please select “Me and the GS” followed by “Grants” from the options at the top of the page, and then choose the Training Grant. If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

Deadlines are quarterly (@midnight on: 15 February, 15 May, 15 August, 15 November).

A supporting statement from the applicant’s supervisor, who must be a current member of the Genetics Society, should be uploaded via the online application form before the quarterly deadline. However, if the applicant is a named investigator (PI or Co-I), this is not necessary. The Genetics Society aims to notify the decision within one month of applications. Applications should be submitted at the earliest opportunity, and at least 3 months in advance of the start date of training.





## Public Engagements Grants

Grants are available to members of the Genetics Society to cover costs associated with travel and materials for public.

Due to the ongoing COVID-19 pandemic, we will be accepting proposals for public engagement activities which are delivered virtually.

Grants are available to members of the Genetics Society to cover costs associated with travel and materials for public engagement activities relevant to Genetics.

A two-tier system is in operation, allowing both small and larger scale projects to be assessed:

- Applications for Tier 1 will be considered for small activities, costing up to £1000.
- Applications for Tier 2 will be considered for larger activities, costing from £1-5000.

Successful applicants must:

- acknowledge Genetics Society support at their activity or event
- feature the Genetics Society logo in any new promotional items produced

The Society possesses a useful stock of publicity material (e.g. pop-up banners, leaflets) which you are welcome to use, by arrangement. Where possible, applications should be submitted at least three months in advance of the project start date. A decision would normally be expected within four weeks.

If you have any queries regarding the application process or are experiencing any difficulty with your submission, please contact [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)

Application deadlines are @ midnight on 1st March and 1st September. Please note that the Society takes no responsibility for risk assessments or public liability issues related to any event or activity. These must be completed according to established practice at the host institution.



## New Virtual Conference Grant

With many conferences continuing to move to an online format the Genetics Society have launched an additional scheme of our conference grant, to support members who wish to attend virtual conferences on research in Genetics.

Awards of up to £300 are available to PhD students and postdoctoral scientists within six years of their PhD viva to cover registration costs for virtual conferences.

**The grant is available to:**

- UK-based members to enable attendance at a genetics-related virtual conference based either in the UK or elsewhere
- non-UK-based members to attend a UK-based Genetics related virtual conference

More information about the grant, including instructions on how to apply can be found on our website.

The next deadline for this award is on 1st August 2021, applications should be submitted through mySociety.

Please send any enquiries to [theteam@genetics.org.uk](mailto:theteam@genetics.org.uk)



# Contacting the Genetics Society

**Members and potential members can contact the Genetics Society membership team in the following ways:**

**By phone:**  
0203 793 7850

**By email:**  
[TheTeam@genetics.org.uk](mailto:TheTeam@genetics.org.uk)

**By post:**  
The Genetics Society, 1 Naoroji Street, London, WC1X 0GB

**The Genetics Society offers a wide range of benefits to its members including:**

- Access to generous grants
- Discounted rates for attendance at prestigious Genetics Society meetings
- A biannual newsletter via post
- Free online access to the Society's journal *Heredity*

**Thank you for your support!**

If you are interested in joining the Society, if you are a current member and have any queries about your membership subscription, or if you would like to advise us of a **change of name**, address or **membership status**, please contact the membership team.

If you are looking for an easy way to manage your membership payment and wish to set up an annual Direct Debit, a simple form can be downloaded from the Genetics Society website at <http://bit.ly/2aLRIOF>. Please complete and return the original to the membership team by post at the address above. Postgraduate and full members paying by Direct Debit will receive a discount of £5 off their annual fee.

# ***Heredity*** has a new look: a new front cover every month!

We are accepting figures/pictures/photos from authors that have their articles accepted in the journal.  
Please contact the editorial office to receive the details!

## Heredity

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