FEBRUARY 2022 | ISSUE 86

GENETICS SOCIETY NEWS

In this issue

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.

The Newsletter is published twice a year, with copy dates of February and August.

Diversity and Inclusivity in genetics, piecing together data from across the globe, articles from page 32

the geneticssociety

A word from the editor...

Welcome to Issue 86

Margherita Colucci,
Newsletter Editor
newsletter@genetics.org.uk

Welcome to the latest issue of the Genetics Society Newsletter!

In this issue we are exploring diversity and inclusivity in genetic research. Feature articles cover studies on genetic diversity in underrepresented populations, research that aim to make data and information more accessible to families and communities, and research careers in countries with limited resources. It is a complex and varied topic, and the following articles provide a thoughtful glimpse into many issues and fascinating research topics:

"Human variation is an interesting phenomenon observable at many levels of our existence, but perhaps most fascinating at the molecular level. Nothing attests to each human's uniqueness as well as the DNA. […] Many of the underrepresented populations exhibit the greatest genetic diversity. Their genomes have evolved in response to their unique environment and lifestyle choices, therefore, this inequity means that vast amounts of genetic variants are missed since they are either absent or present in low frequencies in the European population"
(Oyewumi Akinpelu, page 32)

"[P]recision medicine seems to be closer than ever. However, not all populations will benefit equally. Nor will the architecture of diseases be fully understood without the adequate representation of all human diversity"
(Maria Jose Palma Martínez, page 35)

We continue to explore the career evolution and impact of research experience on the past years Summer Studentship grant winners: students shared their experience and hopes with us in "Genetics Society Summer Studentship - Share your story", page 42. For more interviews, see issue 85.

The Newsletter is growing! Be prepared for new material, from "picture of research" to book reviews and scientific games! Would you like to be part of this change? For more information see page 58.

If you have any comments or suggestions for future themes or articles, please send them to me: newsletter@genetics.org.uk

Enjoy!

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Web: www.genetics.org.uk

The Genetics Society Journals
Heredity
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
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

Functional Regulatory Genomics and Disease

Date: 11-13 April, 2022

Location: The Royal College of Surgeons, Edinburgh

Website: https://genetics.org.uk/events_categories/GS-conferences

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.

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 to support their attendance. Applications can be made through the mysociety portal.

Genetics of Reproduction

Date: 18th November, 2022

Location: Royal Society London (in person and hybrid options)

Info: Reproduction is the process whereby organisms pass on their genetic information to the next generation. Reproductive strategies are highly varied and success is measured not only by the number of offspring produced but also the reproductive success of the next generation. Mammals (and some cartilaginous fish and reptiles) have evolved a strategy whereby fertilised eggs are retained inside the female, the embryo receives nourishment from the mother via a placenta and the offspring a born alive. Further maternal resources in the form of milk are exclusively provided by the mother alongside high quality maternal care. This intimate and bidirectional relationship between the mammalian mother and her offspring increases the chances of offspring surviving to adulthood to reproduce but places a substantial burden on the mother.

Direct nurturing by the mother also creates an excellent opportunity for exploitation both by the offspring she carries and cares for, and by the male parent. While these adaptations have led to the global success of mammals, they are also linked to highly common pregnancy complications which impact the health of the mother, her offspring and, in some cases, her offspring's offspring.

This meeting will cover establishment of the mammalian germline, embryonic and placental development and common pregnancy complications finishing with a lively debate on the evolution of pregnancy led by invited speakers.

External Meetings Diary

More detailed information and links to event websites can be found at **https://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**

Marine Biological Association postgraduate conference 2022 – Turn of the tide: Marine organisms in an ocean of change

Date: 20-22 April, 2022
Deadlines: 20 July, 2021 (Abstract and Bursary); 23 August, 2021 (Registration)
Location: Redmonds Building, Brownlow Hill, Liverpool
Website: https://mba2022.org/wellcomeconnectingscience.org/event/
Contact: theteam@mba2022.org
Info: The 18th MBA non-profit postgraduate conference is hosted by Liverpool John Moores University. It will be the first hybrid MBA conference offering both in-person and online access to all three days of the event! Online registration to the streamed event is open to all ages and free. We have a main topic all about past, present and future change at the species level (e.g., genetic, behavioural) and/or ecosystem level (e.g., trophic ecology, hydrology).
For this topic, the changes can have different abiotic/biotic and natural/human caused drivers.
There are 3 main sub-themes for postgraduate speakers to apply for, based on the location of their research: estuary/coastal environments, the Open Ocean (Pelagic and Demersal environments), abyssal/deep Sea environments.

3rd Epigenetics Conference: from Mechanisms to Disease #3EPI22

Date: 19-22 May, 2022
Deadlines: 25 Mar 2022 (Poster Submission); 25 Mar 2022 (Registration Deadline)
Location: Cancun, Mexico
Website: https://coursesandconferences.wellcomeconnectingscience.org/event/
Contact: admin@fusion-conferences.com
Info: The DNA of eukaryotic cells is organized into chromatin. Chromatin is the template of life. It is the vector of genetic information and it is at the core of fundamental processes such as gene regulation, stem cell fate determination and cancer. The study of chromatin structure and function and the study of mechanisms of disease and their diagnostic and therapeutic approaches are mostly covered by separate meetings. However, the boundaries among these various disciplines are disappearing and now, the meeting on "Epigenetics: from mechanisms to disease" will bring together scientists studying chromatin architecture, epigenetics, stem cell biology cancer and other diseases. The talks will cover a broad range of topics, including chromosome organisation, long-range interactions, chromatin assembly, stem cell regulation/differentiation, RNA-based mechanisms, transcription regulation, DNA methylation and hydroxymethylation, as well as the use of genome engineering technologies, single-cell approaches and cutting edge epigenomics and imaging approaches. Speakers have been selected to broadly reflect lessons learned from a variety of model organisms and experimental approaches. Young scientists will be able to present their work through a large number of short talks selected from submitted abstracts as well as through poster presentation. The program has also allocated ample time for exchanging ideas and discussing novel hypotheses at the end of each session, as well as time for informal interactions and networking.

Courses

More detailed information and links to these and other courses can be found at **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**.

Generalised Additive Models In R; A Data-Driven Approach To Estimating Regression Models (Virtual Course)

Date: 14-18 February, 2022
Location: Online
Website: https://www.physalia-courses.org/courses-workshops/
Info: Most of the statistical methods you are likely to have encountered will have specified fixed functional forms for the relationships between covariates and the response, either implicitly or explicitly. These might be linear effects or involve polynomials, such as x + x2 + x3. Generalised additive models (GAMs) are different; they build upon the generalised linear model by allowing the shapes of the relationships between response and covariates to be learned from the data using splines. Modern GAMs, it turns out, are a very general framework for data analysis, encompassing many models as special cases, including GLMs and GLMMs, and the variety of types of splines available to users allows GAMs to be used in a surprisingly large number of situations. In this course we'll show you how to leverage the power and flexibility of splines to go beyond parametric modelling techniques like GLMs.

Conditional Transgenics

Date: 14-15 February, 2022
Location: Advance at MRC Harwell, Harwell Campus, Oxfordshire, OX11 0RD
Website: https://www.har.mrc.ac.uk/training/courses/conditional-transgenics/
Contact: training@har.mrc.ac.uk
Info: Transgenic mice are an important resource to understand gene function, regulation, and expression. This course is designed to introduce the principles behind conditional genetic modifications, experimental design and analysis.

NGS analysis for gene regulation and epigenomics (virtual)

Date: 24 February - 4 March, 2022
Location: Online
Website: https://www.physalia-courses.org/courses-workshops/course59b/
Info: This course will introduce biologists and bioinformaticians to the field of regulatory genomics. We will cover a broad range of software and analysis workflows that extend over the spectrum from the best practices in the quantitative analysis of ChIP-seq and ATAC-seq data to the analysis of the chromatin 3D structure (such as A/B compartments, chromatin loops or TADs). This course will help the attendees gain accurate insights into local and spatial regulatory functions of the chromatin.

Introduction To Genome-Wide Association Studies (GWAS) (Virtual Course)

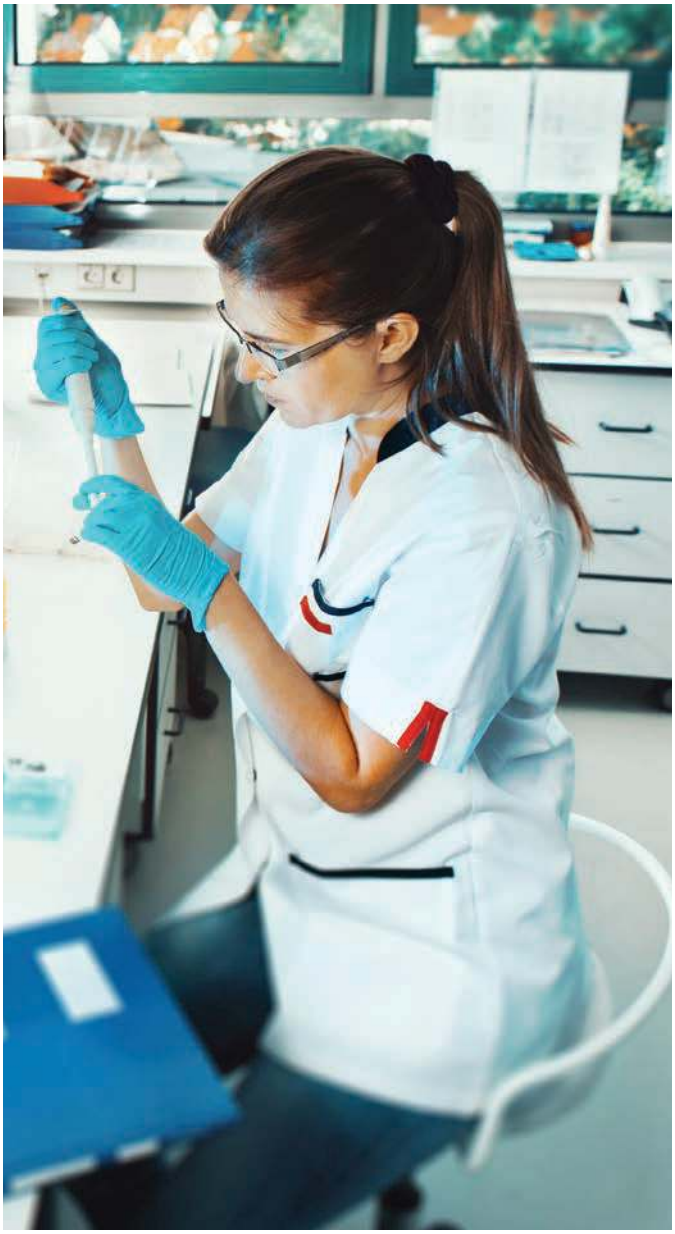
Date: 9-13 May, 2022
Location: Online
Website: https://www.physalia-courses.org/courses-workshops/course49/
Info: This course will introduce students, researchers and professionals to the steps needed to build an analysis pipeline for Genome-Wide Association Studies (GWAS). The course will describe all the necessary steps involved in a typical GWAS study, which will then be used to build a reusable and reproducible bioinformatics pipeline.

Advanced Mouse Genetics

Date: 16-17 May, 2022
Location: Advance at MRC Harwell, Harwell Campus, Oxfordshire, OX11 0RD
Website: https://www.har.mrc.ac.uk/training/courses/advanced-mouse-genetics/
Contact: training@har.mrc.ac.uk
Info: This course explores more complex genetics topics, including multiple allele crosses, complex disease models and will inform breeding strategies and experimental design rather than give a complete guide to the molecular biology. Our team of experienced trainers will support you to examine how different modification lead to different breeding strategies, the importance of genetic consistency and integrity, different modes of inheritance and troubleshooting of reproducibility.

DNA to Disease: Genetics for Beginners

Date: 6 June, 2022
Location: Advance at MRC Harwell, Harwell Campus, Oxfordshire, OX11 0RD
Website: https://www.har.mrc.ac.uk/training/courses/dna-to-disease-genetics-for-nonbiologists/
Info: What is a gene? What is DNA? How do these make proteins? What goes wrong and causes disease in the human body? We will look at the genetic code, how this helps protein formation as well as how errors in DNA can lead to disease.



SECTIONAL INTEREST GROUPS 8

The Genetics Society helps support several sectional interest groups by providing meeting sponsorship. We currently have 16 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/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) in the first instance.

Archaea group

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

British Yeast Group

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

C. elegans

Contacts: Steven Nurrish (s.nurrish@ucl.ac.uk), Michalis Barkoulas (m.barkoulas@imperial.ac.uk)

Evolutionary Genetics and Genomics

Contacts: Frank Jiggins (fmjl001@cam.ac.uk)

Fly South-West

Contacts: James Hodge (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)

London Fly Meetings

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

Mammalian Genes, Development and Disease

Contacts: Rosalind John (johnrm@Cardiff.ac.uk), David Tosh (d.tosh@bath.ac.uk), David Allard (d.allard@exeter.ac.uk), Keith Vance (k.w.vance@bath.ac.uk), Karin Malik (k.t.a.malik@bristol.ac.uk)

British Meiosis Meeting

Contacts: James Higgins (mm2020@leicester.ac.uk)

Mammalian Genetics and Development

Contacts: Nick Greene (n.greene@ucl.ac.uk), Andrew Copp (a.copp@ucl.ac.uk), Cynthia Andoniadou (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)

Contacts: Chris Haley (chris.haley@roslin.ed.ac.uk), Josephine Pemberton (j.pemberton@ed.ac.uk)

Arabidopsis

Contacts: Peter Etchells (Peter.Etchells@durham.ac.uk)

Telomere Network UK (TeN)

Contacts: Alessandro Bianchi (a.bianchi@sussex.ac.uk), Nicola Royle (njr@leicester.ac.uk), David Lydall (d.a.lydall@ncl.ac.uk)

Northern Bioinformatics User Group (Northern BUG)

Contacts: Jarek Bryk (j.bryk@hud.ac.uk)

Ecological Genetics Group

Contacts: Gemma Beatty and Thom Dallimore (genetics@britishecologicalsociety.org)

Population Genetics Group

Contacts: Andrea Betancourt (aabt@liverpool.ac.uk), Stuart Wigby (S.Wigby@liverpool.ac.uk), Robert Fitt (Robert.Fitt@liverpool.ac.uk), Diana Carolina Erazo Quintero (D.Erazo-Quintero@liverpool.ac.uk), Vicencio Oostra (Vicencio.Oostra@liverpool.ac.uk), Evelyn Taylor-Cox (E.Taylor-Cox@liverpool.ac.uk), Ilik Saccherir (saccheri@liverpool.ac.uk), James Hall (J.P.J.Hall@liverpool.ac.uk), Sam Whiteford (hlswhit2@student.liverpool.ac.uk), Matthew Kelbrick (Matthew.Kelbrick@liverpool.ac.uk)

London Human Genetics Network

Contacts: www.londongeneticsnetwork.com , Professor Angelica Ronald (a.ronald@bbk.ac.uk), Dr Karoline Kuchenbaecker (k.kuchenbaecker@ucl.ac.uk)

Heredity PODCAST

Hear directly from the experts, wherever you are and on whatever device...

Recent episodes:

Genetic purging
A look at the surprising benefits of inbreeding in small, endangered populations

The best of 2021
Highlights from the past year of the Heredity Podcast

Surprising little lemurs
Prepare for the unexpected, as a 'simple' mutation rate study contends with the scandalous sex lives of grey mouse lemurs

A genomic approach to oceanography
Clues to ancient ocean currents lie hidden within the genomes of migratory marine species like the green turtle

Resisting the Tilapia Lake Virus
A natural outbreak of an emerging disease provides ideal conditions to study a major threat to the sustainable aquaculture of the third most important farmed fish

Why's that fish transparent?
Aquaculture meets model systems and gene editing in a quest to identify the gene responsible for turning red sea bream transparent!

Listen online or download today: **nature.com/hdy/podcast**

Heredity

the geneticssociety

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 to accompany their formal paper published in *Heredity*.

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

Annual prize for the best student-led paper in Heredity

We are pleased to announce the winners of the second annual prize for the best student-led paper in Heredity.

Each year Heredity awards a prize for the best paper led by a PhD, Master's or undergrad student. Papers can be considered up to three years after the original degree/ project completion. For this year's prize we considered 36 papers, accepted between 1 Aug, 2020 and 30 July, 2021.

The quality of papers was very high but the main award for this year goes to Allie Graham, from the University of Utah for the paper "Adaptive introgression of the beta-globin cluster in two Andean waterfowl".

We would like to thank all of the authors for submitting their research to Heredity and specifically congratulate the three finalists.

Armando Arredondo (Université de Toulouse) was selected lst runner up for the article "Inferring number of populations and changes in connectivity under the n-island model"; this was also the best theory paper led by a student. Joanna Denkena from the Helmholtz Zentrum München has been selected as 2nd runner up with the article "Region-level epimutation rates in *Arabidopsis thaliana*".

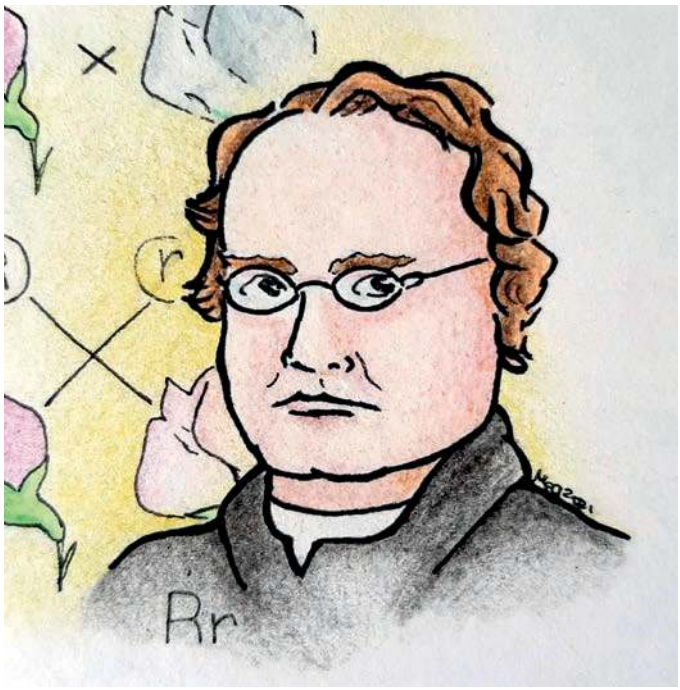
You can find all student led papers on the Heredity website under collections:
https://www.nature.com/hdy/collections

Graham AM, Peters JL, Wilson, RE et al. (2021) Adaptive introgression of the beta-globin cluster in two Andean waterfowl. Heredity 127, 107–123 https://doi.org/10.1038/s41437-021-00437-6

Arredondo A, Mourato B, Nguyen K et al. (2021) Inferring number of populations and changes in connectivity under the n-island model. Heredity 126, 896–912 https://doi.org/10.1038/s41437-021-00426-9

Denkena J, Johannes F, Colomé-Tatché M (2021) Region-level epimutation rates in Arabidopsis thaliana. Heredity 127, 190–202 https://doi.org/10.1038/s41437-021-00441-w

Heredity Special Issue in Honour of Mendel's 200th birthday



Gregor Mendel (born 20 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. The Genetics Society is sponsoring a special issue which will coincide with the bicentenary in July and will include short essays, reviews, perspectives and research articles that address how Mendel's ideas have fuelled development of understanding of such "exceptions".

genetics
unzipped
the Genetics Society podcast

Genetics Unzipped is back for 2022

Genetics Unzipped podcast, The Genetics Society's podcast, is back for a fourth year with even more fascinating stories and interviews from the world of genetics.

We'll be exploring what happens when maths and molecules collide to establish the underlying patterns of life, looking back on the work of leading scientific figures such as D'Arcy Thompson and Alan Turing. Coming right up to the cutting edge, we'll be finding out why researchers are so excited about mysterious packages of genetic material known as exosomes and how we can harness them for health. And we'll also be grappling with the darker side of genetics, addressing the legacy of eugenics and racism that extends from the history books through to today.

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** to check out our extensive archive and full transcripts. You can also follow us on Twitter @GeneticsUnzip or email **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

Following restructuring of Executive Committee roles in line with evolving policies, a new position has been created: "**Officer for Membership and Partnership**".

The responsibilities of the Membership and Partnership Officer will include ensuring continuing growth of Society membership, the appointment of and communications with Ambassadors, and communications with Society partners. More details can be found on the website.

Application/nominations for the role should be submitted by the 31st March 2022 to the Honorary Secretary, Kay Boulton via email: secretary@genetics.org.uk.

Applicants/nominees and nominators should be members of the Society and hold an academic post in a UK university or research institute.

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

- **Policy Officer**
  to replace Rebecca Oakey
- **Evolutionary, Ecological and Population Genetics**
  to replace Jason Wolf.

Details and responsibilities of these roles are supplied on the Society website. Applicants/nominees and nominators should be members of the Society and hold an academic post in a UK university or research institute.

Applications should be submitted by 30th June 2022 to the Honorary Secretary, Kay Boulton via email: **secretary@genetics.org.uk**.

Medal and Prize Lecture Announcements

The 2022 Prize Lecture and Medal awards are as follows:

Genetics Society:	Robin Lovell-Badge, Francis Crick Institute, London
Mary Lyon Medal:	Irene Miguel-Aliaga, MRC, LMS
Balfour Lecture:	Sam Behjati, Wellcome Sanger Institute, Cambridge
Sir Kenneth Mather Memorial Prize:	Robert Hillary, University of Edinburgh
JBS Haldane Lecture:	Mike Fay, Royal Botanic Gardens, Kew
Mendel Medal awards:	Davor Solter and Azim Surani, Gurdon Institute, University of Cambridge

Genetics Society Medal 2022

Robin Lovell-Badge, Francis Crick Institute, London

Robin Lovell-Badge is a senior group leader and head of the Laboratory of Stem Cell Biology and Developmental Genetics at the Francis Crick Institute.

He obtained his PhD in embryology at University College London (UCL) in 1978, carrying out mouse stem cell and embryo research with Martin Evans. It was during his postdoc in the Genetics Department at the University of Cambridge where he began to consider genetic approaches. An EMBO Long Term Fellowship in Paris allowed him to develop methods for studying gene function and regulation, via embryonic stem cells and transgenic mice. He then established his independent laboratory in 1982 at the Medical Research Council (MRC) Mammalian Development Unit, UCL, directed by Anne McLaren. It was here that he began to study sex determination, combining embryology with molecular genetics and mutation studies in the mouse to test and find candidates for the Y-linked testis determining gene. In 1988 he moved to the MRC National Institute for Medical Research (which was incorporated into the Francis Crick Institute in 2016), becoming Head of Division in 1993

It was in 1990, in collaboration with Peter Goodfellow's lab, that Robin identified *Sry/SRY* as a new candidate for the testis determining gene in mice and humans. He went on to prove that *Sry* was the gene and the only one on the Y chromosome required to initiate testis rather than ovary differentiation. Subsequent work by Robin's lab and others have identified and tested the function of many other relevant genes and established many of the genetic pathways involved in the initiation and maintenance of gonadal sex.

At the same time as finding *Sry/SRY*, Robin's lab also discovered the first members of the *Sox* gene family. He went on to show, using genetic methods, the importance of *Sox2* for pluripotency in the early embryo, and for *Sox1*, *Sox2*, *Sox3* and *Sox9* for the development of the central nervous system, the pituitary, and for stem cells in these systems. In addition to being of fundamental interest, Robin's work is of clinical relevance, providing better diagnosis and understanding of the etiology of disorders of sex differentiation and of disorders affecting the CNS and pituitary. Robin was elected a member of EMBO (1993), a Fellow of the Academy of Medical Sciences (1999), the Royal Society (2001), the Royal Society of Arts (2002), the Royal Society of Biology (2011), the American Association for the Advancement of Science (AAAS) (2018), and the Galton Institute (2018). He has received the Louis Jeantet Prize for Medicine (1995), the Amory Prize (1996), the Feldberg Foundation Prize (2008), the Waddington Medal of the British Society for Developmental Biology (2010), and the ISSCR Public Service Award (2021). He was awarded a CBE in the 2018 New Year's Honours List.

Robin was a Distinguished and is now a Special Visiting Professor at the University of Hong Kong (where he has also been a visiting professor since 1996); an Honorary Professor at UCL, (since 2003), and a Visiting Professor at King's College London (since 2016). He is President of the Institute of Animal Technology.

Robin is also very active in both public engagement and policy work, notably around stem cells, genetics, human embryo and animal research, and in ways science is regulated and disseminated.

Robin will present his lecture and receive his medal at the upcoming Society Scientific meeting "Genetics of reproduction" on 18th November at the Royal Society, London.

Mary Lyon Medal

Irene Miguel-Aliaga, MRC, LMS

Irene Miguel-Aliaga is Professor of Genetics and Physiology at Imperial College London, and MRC Investigator at the MRC London Institute of Medical Sciences.

Irene has an interest in the crosstalk between organs – in particular, how and why the intestine communicates with other organs, such as the brain. Her lab was one of the first to tackle the study of the brain-gut axis using the powerful genetics of *Drosophila*: work that they have now extended to mouse and human models.

Irene and her team discovered that the brain-gut axes of males and females are very different, and that these intestinal sex differences impact food intake, gamete production and tumour susceptibility. They have also investigated how the intestine senses nutrients, revealing unexpected roles for metal sensing in the regulation of feeding and growth. Irene trained as a biochemist in Barcelona, Spain and she received her PhD in Genetics from the University of Oxford (UK). She investigated how neurons develop during postdoctoral work at Harvard (USA), Linköping University (Sweden) and NIMR (now Crick Institute, UK).

Irene was the recipient of an ERC Starting Grant and currently holds an ERC Advanced Grant. She was elected to the EMBO YIP programme in 2012, to EMBO in 2017 and to the Academy of Medical Sciences in 2019. She was also awarded a Suffrage Science Women in Science award in 2018.

Irene will receive her medal and present her lecture at the Society Mendel's 200th Birthday celebration, RHS Wisley, Surrey, on 20th July, 2022.

JBS Haldane award

Mike Fay, Royal Botanic Gardens, Kew

Mike graduated from the University College of Wales, Aberystwyth, in Genetics and Plant Breeding, and then carried out research on genetic resources in clover for his PhD at the Welsh Plant Breeding Station (now Institute of Biological, Environmental & Rural Sciences (IBERS), Aberystwyth University). After two years working for the Commonwealth Agricultural Bureaux International as a Scientific Information Officer, he moved to Kew (where he still works) in 1986, as Head of the Micropropagation Unit. In 1995, he established a program in Conservation Genetics in the Jodrell Laboratory. Subsequently, he became Head of Genetics, and is now Senior Research Leader in Conservation Genetics and Molecular Ecology.

For many years, his research has focused on the application of genetic and other data in the formulation of conservation management plans, working extensively with Natural England and other organisations. Major projects include population genetics of orchids and studies of polyploidy, hybridization and apomixis in endemic whitebeams and rowans (*Sorbus* species).

He has published > 220 publications in peer-reviewed journals, and he is joint author of *Plant of the World – An Illustrated Encyclopedia of Vascular Plants* (2017). He has been Chair of the Orchid Specialist Group of the Species Survival Commission of IUCN since 2006 and Chief Editor of the Botanical Journal of the Linnean Society since 2008.

His interest in genetics extends beyond his professional life, and he is a keen amateur genealogist.

Balfour Lecture

Sam Behjati, Wellcome Sanger Institute, Cambridge

Sam Behjati is a Group Leader / Wellcome Intermediate Fellow at the Wellcome Sanger Institute and Honorary Consultant Paediatric Oncologist at Addenbrooke's Hospital, Cambridge. Originally from Germany, he read medicine at the University of Oxford. Subsequently he underwent academic clinical training in London and obtained his PhD from Cambridge. He started his current position at Sanger in October 2018.

Sam's research focuses on the origins of childhood cancer. In one line of enquiry, he uses somatic mutations as barcodes of human development which enable the reconstruction of the embryology of tissues. He has applied this approach to study normal tissue development, most notably, of the placenta, and to childhood cancers. For example, in Wilms tumour, his analyses showed that many seemingly sporadic tumours arose from developmentally acquired clonal expansions. In another line of research, he studies single childhood cancer cells, through the direct comparison of cancer with developmental cells, to place childhood cancers on trajectories of human development.

Sam's work has been recognised by several awards such as the Pezcoller Foundation-EACR Rising Star Award, the Robert J. Arceci Innovation Award of the St Baldrick's Foundation and the Science / SciLifeLab Prize for Young Scientists. He is an EMBO Young Investigator and has recently been awarded a Wellcome Senior Research Fellowship.

Sir Kenneth Mather Memorial Prize 2020/2021

Robert Hillary, University of Edinburgh

I am delighted to have received the Sir Kenneth Mather Memorial Prize. I want to extend my gratitude to the Genetics Society and the University of Birmingham. I would also like to thank my brilliant PhD supervisors Dr Riccardo Marioni, Dr Kathryn Evans, Prof Craig Ritchie, Prof Ian Deary and thesis chair Prof Caroline Hayward who nominated me for this award. I feel very fortunate to have had such supportive supervisors and mentors to guide me through my PhD and beyond.

I embarked on a Wellcome-funded PhD programme in Translational Neuroscience at the University of Edinburgh. My PhD thesis was titled: 'A multi-omics approach to understand the role of plasma proteins in cognitive ageing and dementia'. The overarching aim of my work was to determine whether blood-based molecular markers can predict dementia risk. First, I performed genome- and epigenome-wide association studies on the levels of over 400 blood proteins measured in either The Lothian Birth Cohort 1936 or Generation Scotland. I applied these data to causal analysis methods and found a small number of blood proteins whose levels might causally associate with dementia risk. Second, I showed that an existing blood-based predictor of mortality termed 'DNAm GrimAge' robustly associated with multiple measures of brain health but did not associate with the incidence of Alzheimer's disease. I am now working as a postdoctoral researcher at the Institute of Genetics and Cancer, Edinburgh and hope to pursue a fellowship to continue my work in molecular epidemiology and common disease.

Robert presented his work and received his award at the 2022 Pop group SIG meeting in January this year.

Mendel Medal 2022

Azim Surani, Gurdon Institute, University of Cambridge

Azim Surani received a PhD at Cambridge University in 1975 under Sir Robert Edwards (Nobel laureate, 2010), working on early mammalian development. After moving to the Animal Research Station Cambridge in 1979, he continued to investigate if both parental genomes are essential for mammalian development, which led to Genomic Imprinting in 1984.

Genomic Imprinting has been pivotal for advances in epigenetics and mammalian development, showing that parental chromosomes retain a memory of their origin with heritable DNA methylation tags, which regulate the expression of parental alleles with a role in mammalian development, growth, behaviour and human diseases. The integration of the imprinting cycle into the mammalian germline cycle allows for epigenetic resetting, including the erasure and reestablishment of imprints. Genomic imprinting was critical for the evolution of placental viviparity, with a fundamental impact on strategies for mammalian development.

Surani was elected Marshall-Walton Professor at Cambridge University in 1992 and subsequently Director of Germline and Epigenetics Research in 2013 at the Gurdon Institute. He elucidated the hitherto unknown genetic basis for mammalian germ cell specification in mice and humans and the mechanisms regulating the unique germline epigenetic program. He is also studying in vitro models of early human development, the germline and in vitro gametogenesis.

He is a Fellow of the Royal Society and the Academy of Medical Sciences, and his awards include the William Bate Hardy Prize, a Royal Medal for mammalian development, Rosenstiel Award for epigenetic regulation of gene expression in mammals, ISSCR McEwen Award for Innovation, and the Canada International Gairdner Award for genomic imprinting and epigenetics.

Davor Solter, Emeritus Director, Max-Planck Institute of Immunbiology and Epigenetics, Freiburg, Germany

Davor Solter, M.D. (1965), Ph.D. (1971) both from the University of Zagreb, Croatia. Assistant and Associate Professor in the Departments of Anatomy and Biology, University of Zagreb Medical School 1966-1973. In 1973 moved to the Wistar Institute, Philadelphia and became Member and Professor in 1981 as well as Wistar Professor at the University of Pennsylvania. In 1991 he was appointed Member of the Max-Planck Society and Director of the Max-Planck Institute of Immunobiology in Freiburg. From 2008-2014 Research Director, Institute of Medical Biology, A*STAR, Singapore and Professor, Duke-NUS Graduate Medical School. Currently Visiting International Professor Siriraj Center for Excellence in Stem Cell Research, Mahidol University Medical School, Bangkok.

He was and is a member of numerous editorial and advisory boards and of the American Academy of Arts and Sciences, EMBO and Academia Europea. In 1998 received March of Dimes Prize in Developmental Biology for pioneering the concept of imprinting, in 2007 Rosenstiel Award (shared with Mary Lyon and Azim Surani) for discovery of imprinting and in 2018 Canada Gairdner International Award for discovery of imprinting.

Davor Solter contributed significantly to many areas of mammalian developmental biology, namely: differentiation of germ layers; the role of cell surface molecules in regulating early development; biology and genetics of teratocarcinoma; biology of embryonic stem cells; imprinting and cloning. His current research interest focuses on genetic and molecular control of genome reprogramming and of activation of the embryonic genome.

2023 Medal, Lecture and Award Nominations

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. In addition to delivering the Lecture, the recipient will receive an honorarium of £1000.

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.

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 of their election.

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 Genetics Society and Mary Lyon Medals, and the Balfour and JBS Haldane Lectures. 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. 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 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 June, 2022 at: secretary@genetics.org.uk.

Sir Kenneth Mather Memorial Prize

In a change to previous years, The Sir Kenneth Mather Memorial Prize of £500 rewards a BSc or MSc student of any UK University or Research Institution who has shown outstanding performance in the area of quantitative or population genetics within the current academic year. The prize is awarded annually and pertains to a project report or dissertation 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. 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.

Bruce Cattanach Prize

A new award for 2023, the Bruce Cattanach Prize is awarded annually by Mouse News Letter Ltd for an outstanding PhD thesis related to the use of *in vivo* animal models. Bruce tirelessly nurtured and encouraged junior scientists in the use of animal models. The £500 prize money is to be spent by the recipient to advance their science interests and career. The recipient will be invited to present their work at a Genetics Society Scientific Meeting. Nominations will be assessed by a panel of two people from Mouse News Letter Ltd.

Edith Rebecca Saunders Award

A new award, the REdith Rebecca Saunders Award is awarded by The Genetics Society for an outstanding PhD thesis in any genetics research area. The £500 prize money is to be spent by the recipient to advance their science interests and career. The recipient will be invited to present their work at a Genetics Society Scientific Meeting. Nominations will be assessed by a panel of two Society Committee members.

Call for Nominations

Nominations for the 2021/22 Sir Kenneth Mather Memorial, Bruce Cattanach, and Rebecca Saunders Prizes should be submitted to The Genetics Society electronically via the website, before October 31st, 2022. To be eligible for nomination, as a condition of their course, dissertations/project reports/theses are required to be submitted by the student to the nominating University or Institution between 1st September 2021 and 31st August 2022. Nominators should supply their Genetics Society membership number on the application form. 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 these awards.

Watch this space for more announcements about a new post-doc award and a science communication writing prize!

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).

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.

Ambassadors receive a welcome pack with information about the role and materials to assist with distribution of information.

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.**

Location	Institute	Local ambassador
Aberdeen	University of Aberdeen	Professor Anne Donaldson
Aberystwyth	Aberystwyth University	Dr Dylan Wyn Phillips
Bangor	Bangor University	Dr Alexander Papadopulos
Bath	University of Bath	Dr Araxi Urrutia
Belfast	University of Ulster	Dr Declan McKenna
Birmingham	University of Birmingham	Dr Lindsey Compton
Bournemouth	Bournemouth University	Dr Anna Mantzouratou
Brighton	University of Sussex	Dr Alessandro Bianchi
Bristol	University of Bristol (SOMs)	Professor Patricia Kuwabara
Cambridge	University of Cambridge (Dept of Zoology)	Dr Howard Baylis
Cambridge	University of Cambridge (Dept of Plant Sciences)	Dr Ian Henderson
Cambridge	University of Cambridge (Dept Phys, Dev, Neuro)	Dr Bénédicte Sanson
Cambridge	University of Cambridge (Sainsbury Laboratory)	Philip Wigge
Cambridge	University of Cambridge (Dept of Genetics)	Hansong Ma
Canterbury	Canterbury Christ Church University	Dr Simon C. Harvey
Cardiff	University of Wales College of Medicine	Dr Timothy Bowen
Cardiff	Cardiff University	Dr William Davies
Coventry	University of Warwick	Dr Jose Gutierrez-Marcos
Dublin	University College Dublin	Professor Oliver Blacque
Dublin	Trinity College Dublin	Dr Alastair Fleming
Dundee	James Hutton Institute	Dr Isabelle Colas
Dundee	University of Dundee	Professor Michael JR Stark
Durham	Durham University	Dr David Doupé
Edinburgh	MRC Human Genetics Unit	Professor Ian Jackson
Edinburgh	The Roslin Institute	Dr Douglas Vernimmen
Edinburgh	Institute of Evolutionary Biology	Dr Jarrod Hadfield
Edinburgh	SRUC	Professor Eileen Wall
Essex	University of Essex	Dr Antonio Marco
Exeter	University of Exeter	Dr Sarah Flanagan
Exeter	University of Exeter	Dr Ben Longdon
Glasgow	University of Glasgow	Dr Iain Johnstone
Glasgow	University of Glasgow	Dr Kevin O'Dell
Guildford	University of Surrey	VACANT
Harpenden	Rothamsted Research	Dr Adriana Maria Torres-Ballesteros
Harwell	MRC Harwell	Dr Akanksha Bafna
Hatfield	University of Hertfordshire	Dr Maria Braoudaki

Location	Institute	Local ambassador
Hinxton	Wellcome Trust Sanger Institute	VACANT
Huddersfield	University of Huddersfield	Dr Martin Carr
Hull	University of Hull	Dr David Lunt
Kent	University of Kent	Dr Mark N Wass
Lancashire	Edge Hill University	Dr Paul Ashton
Leeds	University of Leeds (School of Biology)	Dr Andrew Peel
Leicester	University of Leicester	Dr Ed Hollox
Liverpool	University of Liverpool	Dr Tony Plagge
Liverpool	University of Liverpool	Dr Peter Walley
Liverpool	Liverpool John Moores University	Dr Craig Wilding
London	Imperial College London (South Kensington)	Dr Michalis Barkoulas
London	Imperial College London (Silwood and Ascot)	VACANT
London	UCL Institute of Neurology	Professor E M C Fisher
London	King's College London	Professor Simon Hughes
London	St George's University of London	Dr Yalda Jamshidi
London	Kingston University	Dr Francesca Mackenzie
London	Queen Mary and Westfield College	Professor Richard A Nichols
London	UCL Department of Genetics, Evolution and Environment	Professor Andrew Pomiankowski
London	Royal Veterinary College	Dr Claire Russell
London	The Natural History Museum	Prof. Harald Schneider
London	Francis Crick Institute	Dr James Turner
London	University of Westminster	Dr Emanuela Volpi
London	Royal Botanic Gardens, Kew	Dr Alexander Papadopulos
London	UCL Institute of Ophtalmology	Rosa Correra
Manchester	University of Manchester	Dr Catherine Walton
Manchester	University of Manchester	Dr Reinmar Hager
Newcastle upon Tyne	University of Newcastle (Biol Sci)	Dr Maxim Kapralov
Norwich	University of East Anglia	Dr Tracey Chapman
Norwich	John Innes Institute	Professor Enrico Coen
Nottingham	University of Nottingham (University Park Campus)	Professor John Brookfield
Nottingham	University of Nottingham (Sutton Bonnington Campus)	Dr Richard Emes
Nottingham	University of Nottingham (Queen's Medical Centre)	Dr Helen Miranda Knight
Oxford	University of Oxford (Centre for Neural Circuits & Behaviour)	Professor Stephen F Goodwin
Oxford	University of Oxford (Zoology)	Dr S E Kearsey
Oxford	University of Oxford (Plant Sciences)	Professor Liam Dolan
Oxford	University of Oxford (Plant Sciences)	Dr Niloufer Irani
Oxford	University of Oxford	Professor Jonathan Hodgkin
Oxford	University of Oxford (John Radcliffe Hosp)	Professor Andrew O M Wilkie
Oxford	Oxford Brookes University	Dr Ravinder Kanda
Oxford	Oxford Brookes University	Dr Paul Potter
Plymouth	University of Plymouth	Dr Mairi Knight
Reading	University of Reading	Dr Louise Johnson
Salford	University of Salford	Professor Geoff Hide
Sheffield	University of Sheffield	Dr Jon Slate
Southampton	University of Southampton	Dr Mark A. Chapman
St Andrews	University of St Andrews	Professor Mike Ritchie
Stirling	University of Stirling	Hoang Anh Nguyen
Stirling	University of Stirling	Dr Mario Vallejo-Marin
Stoke-on-Trent	Staffordshire University	Dr Gavin McStay
Sunderland	University of Sunderland	Dr Timothy Barrow
Swansea	Swansea University	Dr Claire Morgan
York	University of York	Dr Sean T. Sweeney

Professor Joy Delhanty

Professor Joy Delhanty studied zoology at University College London (UCL) where she also gained her PhD in genetics. She worked at the Galton Laboratory in UCL with Lionel Penrose and gained her professorship there in 1998. She was a director of the UCL Centre for Preimplantation Genetic Diagnosis in the Institute for Women's Health and was a supervisor to 25 PhD students.

By Dr. Anna Mantzouratou *FIBMS*

My mentor and PhD supervisor Prof. Joy DA Delhanty passed away on the 1st of October 2021. Although I got my PhD way back in 2008, I would always go back to her for guidance and advice and we kept regular contact discussing our common research interests. I am missing her and her guidance very much. She was a member of the Genetic Society and we were frequently talking about interesting items on the society's newsletter so it seemed an appropriate platform to share my experience about Joy.

Joy was a pioneer in cytogenetic investigations, researching tirelessly the origins of aneuploidy in human foetuses, embryos and gametes. Her publication on the first cytogenetically detected triploidy in a foetus (Penrose and Delhanty, 1961) makes for an excellent insight into the beginnings of her research and how far the field of cytogenetics has come thanks to pioneering work like this one. Hence moving forward to 1993 when Joy and her team published the first results of preimplantation genetic diagnosis for X-linked disorders (Delhanty *et al*, 1993) and laying the foundation of a whole new diagnostic field for human preimplantation embryos.

I got the privilege to join Joy's team in 2002. In our meetings Joy would not say much, as I was doing enough talking for the both of us (!), but she would listen very carefully and will always come out with the exactly right thing to say or the right advice to give. I never understood how she did it but she had an amazing ability to remember relevant studies that were published years ago whenever I had a question! She would just flick through her paper drawer and magically pull out the paper and give it to me. It only took her a few seconds, and there was me who mostly managed to remember what I had for breakfast! When I managed to get my lectureship job she gave me her favourite coplin jar as a present and made me promise I will look after it; once a cytogeneticist, always a cytogeneticist!

I feel so lucky I was in Joy's team, I admired her and I am grateful for all the opportunities she gave me. She always protected and guided her PhD students and there are many of us out there all doing amazing research. I hope this article conveys to the society's members how excellent Joy was as a person and as a scientist. I could not be there for her funeral to say goodbye in person but I offer this message to her in the hope her energy will continue to guide my science...

References

- *Delhanty JD, Griffin DK, Handyside AH, Harper J, Atkinson GH, Pieters MH, Winston RM. Detection of aneuploidy and chromosomal mosaicism in human embryos during preimplantation sex determination by fluorescent in situ hybridisation, (FISH). Hum Mol Genet. 1993 Aug;2(8):1183-5. doi: 10.1093/hmg/2.8.1183. PMID: 8401499.*
- *Penrose LS, Delhanty JD. Triploid cell cultures from a macerated foetus. Lancet. 1961 Jun 10;1(7189):1261-2. doi: 10.1016/s0140-6736(61)92766-0. PMID: 13734147.*

"Dear Joy, I would like to thank you for all the guidance and advice you gave me, helping my journey to independence and provide a beacon of light so I can find my way. I will always be grateful."

Professor William G Hill OBE FRS FRSE (1940 – 2021)

Professor Hill was an eminent quantitative and population geneticist and recipient of the Society "Centenary Year" Mendel Medal (2019). Bill was also a gentleman, a friend and a respected colleague of many Society members and beyond.

By Kay Boulton

As a tribute to Bill we have collected many precious anecdotes and memories and share a selection here. more can be found on our website: https://genetics.org.uk/medals-and-prizes/genetics-society-medals-and-lectures/mendel-medal/mendel-medal-2019-william-g-hill/

If you have any photographs, memories or anecdotes of Bill that you would like to share, please send to Kay Boulton secretary@genetics.org.uk and they will be added to the website.

My own memories of Bill are of the kindest and most supportive person. When I first met Bill he was chairing a steering group and I was merely taking minutes. I had no idea of his stellar fame, and I know he had commented to Steve Bishop on how refreshing that had been.

We hardly talked about science, but chatted about our families and shared farming backgrounds. Later, Bill supported my change of career by providing references for the Quantitative Genetics MSc and later my PhD at Edinburgh.

I was thrilled that Bill was awarded the Mendel Medal by the Genetics Society in 2019, and that I was able to organise the event. My everlasting memories of Bill will be as Jarrod and Andrea describe next – a master of mumble, surprise, with a leaning tower of papers on his desk. I feel honoured to have known Bill and been able to call him a colleague and friend.

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By Jarrod Hadfield

I moved to Edinburgh soon after Bill's retirement. In those first few years I must have asked him hundreds of questions, all of which he answered with patience and from an encyclopaedic knowledge of the literature. Even in his 70's he would devour the latest advances in quantitative genetics - his desk was like an arcade coin pusher, with an improbable overhang of recently printed papers. He loved to discuss these advances also, but you had to work hard to understand him. He mumbled and chuckled in equal measure, and in the 15 years I knew him, the only vowels I heard him pronounce were the e in Ne and the a in Va. He used these a lot, and equations would be spoken mid-sentence without any real indication that he had switched from English to Maths. Ne, Va, LD, h-squared were like the names of his children, and always used as if the listener had the same level of familiarity and affection. He found quantitative genetics 'endlessly fascinating' and still without answers to the big questions. Why, for example, was heritability concentrated around 0.25 and not uniform from 0 to 1? He always said the answers would be simple and obvious once we knew them, and so he kept chipping away. Although he never did find the mother lode, plenty of substantial nuggets were found along the way: Hill-Robertson interference, of course, but many others too. Bill was probably the single most important person in my academic development. He taught me a huge amount of population and quantitative genetics, but more importantly he inspired me. His small eyes in a large face still sparkled when he talked about things he'd been studying for over 50 years. Bill is gone, but his legacy will outlive us all.

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By Andrea Doeschel-Wilson

My first encounters with Bill were in the early 2000s at the weekly quantitative genetics journal club in Edinburgh, which Bill attended diligently. For novices in quantitative genetics like me, this was a gold mine to get the insight opinion of giants like Bill about the latest QG studies. We were quite nervous presenting the papers, all too aware of our ignorance in the midst of a genius. But Bill never put us to shame. Instead he generally pretended to sleep through the presentations, and ever so gently planted a few key questions or comments when he 'woke up' to set us in the right direction. I cherish my memories of Bill as the

humble genius with the highest qualities of a proper English gentleman.

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By Sue Brotherstone and Ian MS White

Bill used to have a steady stream of high profile visitors from home and abroad and they would sometimes join in the departmental coffee break. The conversation ranged freely but usually took on a strong scientific slant whenever Bill was present.

After Bill retired he moved to a shared office and his conversation started to include other matters: politics, religion, the state of the world.
But above all else, he liked to discuss football. During the World Cup, the frequency of emails between Bill and Dan Gianola rose to an unprecedented level. It is possible that they were discussing genetics but we suspect that the correspondence was more to do with goal averages, or the probability that Ecuador would survive against Brazil.

Occasionally we could observe a great mind at work. When deep in thought, Bill would sit motionless, staring into the middle distance. Then a long sigh, and we knew that another genetically knot had been untied, another problem solved.

Over the years, we learned more from casual conversation with Bill (and his visitors) than from any number of formal lectures or seminars. He will be much missed, both as a colleague and a friend.

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By Jean-Luc Jannik

A paper from the year I was born!

Hill, W.G., and A. Robertson. 1966. The effect of linkage on limits to artificial selection. Genet. Res. 8(3): 269âEUR”294

The Atlas Computer, on which the simulations were done, was one of the world's first supercomputers, in use from 1962 until 1971. Atlas' capacity promoted the saying that when it went offline, half of the United Kingdom's computer capacity was lost.

"A computer simulation study has been made of selection on two linked loci in small populations, where both loci were assumed to have additive effects on the character under selection with no interaction between loci"

In gratitude to you Bill!

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By Daniel Gianola

I met Bill Hill for the first time in 1976, during the first world conference in quantitative genetics in Ames, Iowa. Of course, I knew who he was, as I had struggled with some of his Biometrics on design of selection experiment when I was a PhD student. At some point, during one of the evenings, some of us were chatting in a sort of university lounge or dorm.

Bill pointed out that there was nothing to drink. I had a bottle of mundane whisky (Red Label) and I brought to the meeting (just in case), to make the conversation livelier.

I saw him again in 1982 and eventually became friends. I spent two months in Edinburgh in 1998, and he kindly shared his office with me during our stay. At that time, he was Dean of Biological Sciences, but made time for discussion. One of the points which was perplexing to Bill (and even more to me, an ignoramus of population genetics and of other matters) was how in the world Sewall Wright had seen that the Fokker-Plack equation could be used to solve the problem of the equilibrium distribution of allelic frequencies under some idealized conditions.

I was very fortunate during the Edinburgh period, because Laurie and Priscilla Piper were there on sabbatical, and we socialized a lot with the Piper and Hill couples. It was a great time, and I had quite a few pints with Bill and Laurie.

Hill has been, and will continue to be, one of the scientists whom I have admired the most. His contributions were monumental, and his contributions to discussions were always milestones. A brilliant and a super-fast mind, coupled with a powerful intuition were his most notable assets as a scientist. He was always humble, and aggressive intellectually but not scary. Last but not least, Bill was generous and hospitable and had an admirable sense of integrity as a person.

His legacy is extremely important to us. Hill's papers and volumes will continue providing much and needed guidance to both young and seasoned scientists.

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By Professor Geoff Simm

I first met Bill in the early 1980s, when I arrived in Edinburgh to do a PhD and attended classes in the MSc in animal breeding that Bill co-directed and taught on. Later, in the East of Scotland College of Agriculture (later part of the Scottish Agricultural College and now Scotland's Rural College) I ran a number of farm animal breeding research projects and Bill provided regular, highly-valued advice to me and a growing number of colleagues working on these projects – and we jointly supervised many great MSc and PhD students, many now leaders in their fields. It is notable that genetics in Edinburgh, under Bill's leadership and Alan Robertson's before him, involved a two-way street between theory and practice. Both had farming interests and were passionate about application of

science in livestock breeding – which has been to the great benefit of breeders and breeding organisations here and abroad – Bill provided direct technical support to several. The challenges in application have often stimulated developments in theory.

Bill was the principal flag bearer of Edinburgh's longstanding international reputation in genetics for over 30 years. Despite this he always gave very generously of his time, experience and considerable intellect to students and colleagues – something that was and remains hugely appreciated by both groups! He wore his brilliance lightly, liked people (most of them anyway!) and saw the humorous side of many situations.

Rosemary and Bill were fantastic hosts to generations of students and visiting scientists coming to Edinburgh, and helped make it such a great place to study and work.

It has been a great honour to call Bill a colleague and friend - he will be sorely missed.

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By Dr Victor Olori

On the broad stairs of Ashworth labs, King's building, he turn to me sharply and said "Don't call me SIR, just Bill". For a young African student fresh out of Nigeria, this was another rude shock. How can I address my mentor, my supervisor, a most senior professor, dean of college and this giant of a man as "Just Bill"? But I soon came to know, that was the nature of the man. Kind, humble and willing always to come down to your level with joy to help. Although it was the name of Douglas Falconer through his book that first brought the University of Edinburgh into my radar in Faraway Nigeria, It was Bill Hill that brought me to study in Edinburgh. Yet I first encountered the man in Guelph, Ontario, at the 5th World Congress on Genetics Applied to Livestock Production. Having just journeyed From Lagos via Frankfurt and experiencing serious jet lag for the first time, seeing and talking to Bill was the only thing I could remember from the opening ceremony of the WCGALP in August 1994.

From that day till my latest visit to Bill's office before the onset of the ravaging COVID and forced incarceration of people, Bill was always there listening, probing and helping to solve problems concerning the theory and practice of animals breeding. He patiently nurtured and imparted knowledge into me throughout my study days. Long after graduating as one of Bills numerous students, I was lucky be in and around Edinburgh throughout my career to date which meant I have continued to have close association and collaborations with Bill. It is therefore particularly painful to hear of his transition even though we all know death is inevitable. This is one loss too much and the only consolation is that Bill has left behind a great legacy. He lives on in his children and family but also in all his academic children collaborators and friends. Not surprised that even in death, he has chosen to continue to further the cause of knowledge with his body.

I pray that the Lord will grant Rosemary and his family the fortitude to bear his loss. Rest in Peace Sir W. G. Hill, for you are indeed a Knight of the genetics and animal breeding round table.

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By Carlos Lopez-Fanjul de Arguelles

I met Bill for the first time in 1968, as a lecturer in the Diploma Course at the Institute of Animal Genetics in Edinburgh. He impressed me so much that, after finishing the course, I asked him to be the supervisor of my Ph. D. thesis. Susan Hayter and myself were the first Bill's PhD students, finishing in 1972. Sadly, Susie died a few years afterwards and Bill usually referred to me as his older student alive.

Under joint authorship, I presented some preliminary results of my thesis work in the Population Genetics Group meeting in Bangor, December 1971, which Bill could not attend. After my talk was finished and the chairman asked for questions, Kenneth Mather took the occasion to launch a furious attack, not particularly to my paper but to the Edinburgh group, one of the many episodes of the vivid debate carried out in the time between the confronted views of the Edinburgh and Birmingham schools. Before I could utter a single word, Dick Lewontin who was then visiting in Edinburgh, made a conclusive defence, as Bill undoubtedly would have done if present. After the session finished, I was comforted by Alan Robertson who said to me: "you have been well understood". I still don't know what he meant by this but, from them onwards, Bill and Alan's work had been the main source of inspiration of my research along 40 years, and, much more important, I have always enjoyed with gratitude their friendship and generous advice.

Bill was the model of the scientist to imitate. Rest in peace.

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By Bruce Walsh

Bill will indeed be missed. His legacy as a titan in the field of quantitative genetics is secure, and the impact of his work will only grow over time. For example, a rather (at the time) obscure paper he did with Alan Robertson on the impact of tight linkage on selection response in an animal breeding context – the so-called Hill-Robertson effect –

now underlies the population-genetic analysis of genomic data (selection at nearby sites makes a region more neutral). Our two textbooks on quantitative genetics (Lynch & Walsh 1998; Walsh and Lynch 2018) would not have been possible with the guidance (and careful review!) from Bill. In particular, Bill prodded us (well, mainly me) for years with "Keep writing" on the second volume, and read the entire manuscript (several times), keeping us from a making a number of stupid mistakes. Much of this volume covers material that can directly be traced back to Bill.

As impressive as he was as a scholar, Bill was more impressive as a person. One of my fondest memories of Bill was from the Illinois long-term selection meeting in 2002. My wife (a PhD in linguistics, not biology) accompanied me and mentioned that she had the most delightful conversation about the on-going world cup with a "fellow from England". They made a friendly wage on the US team's chance in a match. Bill (unlike his namesake gambling company) lost the bet and very graciously bought her a drink. On relating this interaction, she asked me if I had ever heard of a "Bill Hill"! That was typical Bill. Very genuine as well as very generous with his time, just as happy to spend time talking to a new graduate student as with a senior Professor (indeed, he was likely more happy to spend time with the student!).

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By Han Mulder

I visited Bill for the first time in spring 2005 as a PhD-student from Wageningen University to discuss with him opportunities for a sabbatical in autumn 2005. So I stayed from September until December in Edinburgh. He was already retired but with a great and sharp mind with respect to almost everything, but especially quantitative genetics. It was in the beginning still a bit of search for a topic that we both liked and were able to see the light. Finally, I started simulations about genetic variation in environmental variance. Step by step we were trying to understand the results and this led to what became my biggest contribution to quantitative genetics so far: prediction of breeding vales and selection responses with genetic heterogeneity of environmental variance (Genetics 175:1895-1910). I could not have done it without him. Later when I was back in Wageningen I continued corresponding with Bill on email distance and also that worked. Later I wrote with him the review Hill and Mulder 2010 (Genet. Res. 92:381-395), for which I stayed a couple of days with him in his house. He was so energetic, unbelievable. Every evening, we spent some time at Kings buildings to make the most work hours as we could. This review is now one of my most cited papers. When I became assistant professor and decided to develop my research line about genetics of environmental variance, I visited Bill regularly (2013, 2014, 2015, 2018), which resulted in one extra paper (Mulder, H. A., W. G. Hill, and E. F. Knol. 2015. Heritable environmental variance causes non-linear relationships between traits: application to birth weight and stillbirth of pigs. Genetics 199:1255-1269). He became a mentor for me and I enjoyed every minute speaking with him. Also, in between, we at least emailed a couple of times a year.

One of the things that we have in common is interest in farming, so he always talked with me about his farm as I did with respect to my father's farm.

Reading his papers is always inspiring, so his early papers on de novo mutations were a great inspiration for my recent work on de novo mutations and their impact on quantitative traits (H. A. Mulder, S. H. Lee, S. Clark, B. J. Hayes and J. H. van der Werf. 2019. The Impact of Genomic and Traditional Selection on the Contribution of Mutational Variance to Long-Term Selection Response and Genetic Variance. Genetics early online).

Bill's greatest contributions to animal breeding:

I think his greatest contributions are his research on maintenance of genetic and environmental variance, so this includes things like genetics of Ve, maintenance of genetic variance, e.g. contribution of DNM, but also the variability in relationships which is of great importance for predicting accuracy of genomic prediction.

Bill as a person:

- Very friendly, helpful and gentle
- Great and sharp mind
- Interest in family life
- Hard working, workaholic

We will miss him as being one of the giants of quantitative genetics. It was an honour to work with him, probably as one of the last PhD-students and younger generation scientists. Without him, I would never have developed so much interest in quantitative genetics of Ve.

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By Armando Caballero

I started working with Bill as a postdoc in 1990. I had visited him a few months before and the first time I spoke with him I thought that I would not be able to understand him in my whole life. Fortunately, when I arrived at the Institute, he installed me for a few months in an office shared with a predoctoral student of Chinese nationality, whose thesis supervisor had been the recently deceased Alan Robertson. Every time Bill came into the office and said something to me, I would assent to everything without

understanding anything, and when he left, my office mate repeated to me what he had said with a Chinese accent that I could understand much better. I was able to survive the first month in that way until I started understanding him little by little. Later, they told me that sometimes the British themselves could not understand him, which left me less worried. What it was to be initially a two-year stay turned out to be almost a seven-year one. I was extremely lucky to be able to work with him for so long because this allowed me to achieve a theoretical background that I could have never got otherwise. Bill had an infinite capacity for work. Despite his numerous obligations of all kinds, he had time to talk to me almost every day, even if only for a few minutes, which encouraged me to work harder. Bill maintained a pleasant work environment due to his kindness and his sense of humour, and is scientific capacity was incredible, both in knowledge and in deductive and mathematical ability. Any questions I had were resolved instantly or he would tell me where I could find the solution. That made it very easy to work in a difficult field such as quantitative genetics. His way of doing science and his human quality are the fundamental principles that I learned from him and that I try to teach to my own students.

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By Peter Keightley

One thing I remember very well from doing my PhD with Bill from 1986-1989 (nominally part time as a Research Assistant) was that he would come into the office I shared with Sue Brotherstone almost every morning before the coffee break (which was in Alan Robertson's office along the corridor) and ask the same question: "What have you discovered?". This was not because he expected anyone to actually discover something every day, instead I think it was a nice way of opening the conversation about where we were with my project. We would then talk about what results I had produced and what I might do next. He took an incredibly keen interest, but I never felt that it was too much and he was happy for me to go my own way with it. For example, my project was part of a grant on the impact of new mutations under directional selection and was never intended to be about stabilizing selection, but ended up focusing a lot on that.

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By Karin Meyer

Bill was an amazing person. His contributions to quantitative genetics were enormous, spanning the great breadth from theoretical developments to the implementation of livestock improvement programmes.

A specific area of Bill's interest was the estimation of genetic parameters. For instance, he made geneticist aware of the need for covariance matrices to be positive definite and the effects of sampling errors on response to selection on indices. Moreover, while he tended to stay clear of the computational side, he championed the uptake of maximum likelihood based estimation fitting linear mixed models. Edinburgh has been at the forefront of making REML the standard method for variance component estimation in quantitative genetics -- no doubt to a substantial extent due to Bill's involvement.

Bill was known for his unfailing dedication and rigorous approach to science. In addition to his own seminal contributions, he had a profound impact through his numerous postgraduate students and postdocs. Bill had the uncanny knack of pushing his students to realms of genetics and statistics they would have never imagined or reached otherwise. In addition, his utmost interest in and emphasis on scientific papers instilled sound publication habits. Truly invaluable gifts which shaped the careers of many.

I am privileged to have had Bill as supervisor and mentor for my Ph.D. and a postdoc. "I am not interested in how fast you can compute the wrong answer" was a no-nonsense, characteristic comment of Bill's which has become a fond memory.

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By Roel Veerkamp

My first getting to know Bill was in August 1990, even without meeting him. As a young naive student from Wageningen, coffee table talk in Roslin was that Bill must have read and edited all the papers for the WCGALP that year. The first time we met was at the journal club, where Bill always had a preferred seat at the corner. Discussions were always special when Bill was there, and that was most of the time. When I gave al seminar about my thesis work (crosses between mice lines), he raised his eyebrows when I called the preliminary analysis "Jan Boerenfluitjes", but was awake during the whole seminar.

After finishing my MSc, I started working at SRUC. An interesting combination, because Bill Hill was not only one of my PhD supervisor, he was also partner in a project that aimed to develop new selection indices for UK dairy cattle. Given all the politics and organizations involved, it costed quite some time to get final industry support for the project. I never forget the first project meeting. When a technical director confused us when explaining what he expected from the project, Bill politely told him that he was at the wrong project meeting. Bill had a strong interest and involvement in

"Bill was a giant of quantitative genetics and animal breeding and an important figure in population genetics. Less well known are his important impacts in conservation genetics…"

practical animal breeding, and as a PhD supervisor we spent a lot of time discussing dairy cattle genetics. I was amazed how much time he took, also to discuss other things, even when he was Dean of Biological Science.

Bill was the only permanent member of the WCGALP permanent committee. In 2010 during the Leipzig conference, Bill hinted to me that it was time to take responsibility to organise WCGALP in the Netherlands. So we did and organised a bid in Auckland. In 2017 he politely declined to become a member of our WCGALP advisory board ("it is time for a new generation"), and over time it became clear that his health made it unlikely for him to participate in 2022 in Rotterdam. Now he will be missed even more, although many of us will recollect and benefit from his incredible contribution to our scientific field.

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By Professor Richard Frankham

I was deeply saddened by Bill Hill's passing. Bill was a giant of quantitative genetics and animal breeding and an important figure in population genetics. Less well known are his important impacts in conservation genetics, with the following three being especially notable. First, his single sample method for estimating effective population size from linkage disequilibrium predominates in the discipline. Second, his prediction of response to selection from new mutations is important with regard to conservation of genetic diversity and evolutionary potential. Third, the Hill-Robertson effect has attained enhanced prominence in the genomic era in the context of linked selection effects. In addition to his scientific contributions, Bill was a very pleasant and civilized person.

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By Frank Nicholas

My wife, Jan, and I met Bill and Rosemary soon after we arrived in Edinburgh late in 1971. Bill was then in full flight as a young lecturer. Although my PhD supervisor was Alan Robertson, Bill was equally supportive throughout my candidature, not only scientifically but also socially. Indeed, he and Rosemary introduced us to their solicitor, Mr Cochrane, who enabled Jan and me to purchase a delightful Victorian cottage in Haddington, where we lived for most of our all-too-short stay in Scotland.Like me, Bill had grown up on a farm. When my parents came to the UK to visit us in 1973, Bill arranged for my parents to visit his parents on their farm – a magical meeting of farmers from the opposite ends of the world.

Perhaps the most important lesson I learned under Alan's and Bill's mentorship was to understand what it is like not to understand! I was forever struggling to understand concepts that Alan and Bill understood instinctively. Although that lesson was a bit down-heartening at the time, neither Alan nor Bill ever showed any signs of despair! Importantly, I have since come to appreciate that understanding what it is like not to understand is a valuable attribute for any teacher.

Within a few months of our arrival, Bill got me involved in a project that had nothing to do with my thesis work, but which I found just as interesting. Under his never-ending encouragement, my propensity for attention-to-detail (pedantry?) was put to good use in checking every line of algebra, making the occasional suggestion here and there, and doing all the calculations in what became my first Edinburgh paper, published soon after I returned to Sydney.

I remain forever grateful for Bill's tolerance, mentorship and friendship.

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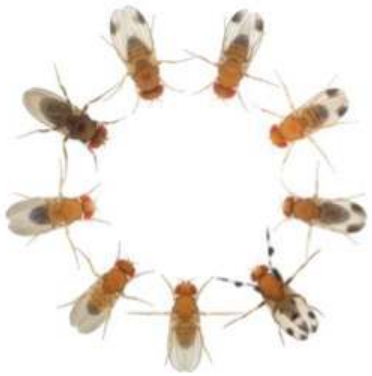
By Adam Eyre-Walker

Bill was my PhD supervisor during my studies at Edinburgh University during the early 1990a. Given that I had dropped out of a PhD after just 10 days the previous year, Bill took quite a chance taking me on. I then proceeded to start working on DNA sequence analysis a field he was not involved in! However, he very generously allowed me to pursue my own path and supported me throughout my studies. He was insightful, generous with his time and efficient - give him a manuscript and you would comments back within 24 hours. I am very grateful to him for supporting me early in my career and allowing me to find my own path.

The 12th South-West Fly meeting

Dr James Hodge . University of Bristol



After a break over 22 months due to Covid, it was great to be able to be back at University of Bristol on the afternoon of 17th November for South-West Fly meeting (SWFM) discussing the latest *Drosophila* research. Veteran SWFM speaker and supporter, Professor Helen White-Cooper of Cardiff University discussed the latest tools, sequencing, and expression data available through next generation sequencing, AI, web tools with curation from the expert fly community. More specifically the talk was on the FlyCellAtlas: What can you learn from chopping adult flies into little bits and then sequencing RNA from their nuclei. Helen gave a live workshop on the free online tool, so everybody can interrogate the expression pattern of their favourite fly gene or tissue namely the testis for Helen. Next Dr Edgar Buhl spoke about a new project he is researching at Bristol University with James Hodge, human geneticists and paediatricians using flies to model rare childhood neuropathies. Essentially the Bristol Children's Hospital clinicians see a lot of kids with rare genetic diseases, which they have no idea what they are or how to treat them. As a means to diagnosis, treatment and understanding the disease, they are now sequencing their patients, and then the group make the equivalent mutation in the fly gene and looking for resulting neuropathic defects. The group has also started the first gene therapy in Bristol, for diseases like Amyloid Lateral Sclerosis. Continuing on the fly disease modelling theme, Eilish Mackinnon from Dr Owen Peters' lab at the Dementia Research Institute at Cardiff University (DRI@CU) talked about the glial role of *small wing/ PLCG2* in modifying amyloid-associated phenotypes in *Drosophila.* This was an elegant set of PhD thesis experiments where a human gene identified by GWAS for Alzheimer's disease (AD) performed by DRI@CU was knocked down in flies yielding AD-like effects of shortening life, eye degeneration and climbing defects. Many of these phenotypes were brought about through changing human amyloid processing. Strikingly phenotypes were due to misexpression in glia as opposed to neurons, with microglia now being thought to be key culprits in the progression of AD pathology. In fact, correcting small wing expression could rescue secreted human amyloid ß defects. Eilish has just used CRISPR to make a conserved PLCG2 AD mutations into the fly small wing gene. Although humans don't have wings and flies don't get Alzheimer's disease, ingeniously, the Cardiff group can monitor neurodegeneration live in the fly wing as well measuring any associated axonal trafficking defects by imaging GFP in the long sensory neurons of the fly wing in AD model flies.

After a tea and coffee break, Dr Paul Langton from Professor Eugenia Piddini's lab at the University of Bristol talked about the role of Xrp1 and proteotoxic stress in Minute cell competition. He introduced the concept of winner and loser cells in terms of their growth, and importance in stem cell division as well as increased cell competition of cancer cells. His work takes advantage of clonal analysis in flies and ability to image live, count and measure the size of cells in developing discs. This allows you to study lethal genes, cell autonomy and boundary effects. Orthologues were looked at in human cells lines and manipulated pharmacologically. The last talk was from a recent postdoc, Dr Meg Stevens of Dr Benjamin Housden's lab at the University of Exeter. They have been identifying novel drugs to treat neurofibromatosis type 1 tumours using combinatorial or synthetic lethal screening in *Drosophila* cells. This has involved performing a range of genetic and drug screens in the context of different diseases using *Drosophila* and human cells, using CRISPR to make analogous mutations in the genes of the two species. Drugs tended to translate well between the models, although there are challenges and differences of feeding the flies the drug as opposed to the cell cultures let alone finding the effective dose and treatment regime in a whole mammal. The high throughput capability and genetic tractability of flies will hopefully be leading to us being able to find more effective treatments in the future. It was great to be able to continue the discussion of fly research in person for the first time in nearly 2 years, and over pizza and drinks generously provided by sponsorship by the Genetics Society, the next meeting is 9th March, 2022, please email **james.hodge@ bristol.ac.uk** if you want to be added to the SWFM special interest group and attend future meetings.

In this issue of the Newsletter, we have seven feature pieces. In the first article, Oyewumi Akinpelu evaluates how diversity is accounted in genetic research. The next article by Maria Jose Palma Martínez gives an overview of the Mexican Biobank Project and its impact. The third article brings the personal story and experience of Prof. Raúl Godoy-Herrera in genetic research affected by limited funding in Chile. Then, Dr Kat Arney reflects on accessibility of genetic research with her guests on the Genetics Unzipped podcast. The fifth piece is a series of interviews: "Genetics Society Summer Studentship - Share your story, Part 2" tells the experiences of the past years grant winners. In this issue, Industrious Science feature the first student-focused interview with laboratory scientist Adriana Macko, at Illumina. Finally, as part of the Newsletter's new section, Dr Michela Leonardi demonstrates how a complex genetic model can become a table game.

Evaluating diversity in genetic research

By Oyewumi Akinpelu

Human variation is an interesting phenomenon observable at many levels of our existence, but perhaps most fascinating at the molecular level. Nothing attests to each human's uniqueness as well as the DNA. Although only 0.1% of the 3 billion base pairs that contribute to our genome varies, it accounts for a wealth of information that has aided advances in various sub-fields of science such as medicine, pharmacogenetics, and forensic science.

Since the completion of the Human Genome Project in 2003, the advent of sequencing technologies such as Next-Generation Sequencing and Whole Genome Sequencing has made reading the sequence of nucleotides in the human genome easier and faster. The result has been tremendous discoveries that have helped improve human health outcomes [1,2]. However, despite the growth experienced in the field of genetics and genomics over the last few decades, there is a chink in the armour - about 84% of the world population do not benefit optimally from the breakthroughs being achieved [3].

THE PROBLEM

It is widely known that the majority of the databases currently used for drawing inferences in genetic research are skewed towards the European population [4,5]. As of 2018, approximately 78% of the individuals included in genome-wide association studies (GWAS) were of European ancestry with the closest following population being those of Asian ancestry at 10%.

The African, Hispanic, and other populations were represented at 2%, 1% and < 1% respectively.

Many of the underrepresented populations exhibit the greatest genetic diversity. Their genomes have evolved in response to their unique environment and lifestyle choices, therefore, this inequity means that vast amounts of genetic variants are missed since they are either absent or present in low frequencies in the European population [6–8]. This is the case for the G1 and G2 variants of the apolipoprotein L1 gene found in high frequencies in those of African ancestry.

These variants which are thought to confer resistance to sleeping sickness, also increase the risk of kidney disease in individuals of this population by 7-to-10 fold [9,10].

The status quo of limited diversity has the potential to stall the progress being recorded in the field of genetics because it impedes the understanding

Increasing diversity in genomic research is not only helpful to the underrepresented populations, but also of great value to people of all backgrounds.

of the overall genetic structure of diseases and can worsen current health disparities [4,7]. Furthermore, the wide gaps could create a lag in the outlook of precision medicine, since using estimates of genetic risk derived from one population for another population could result in inaccuracies [1].

For example, the common causative allele, ΔF508, responsible for > 70% of Cystic Fibrosis cases in the European population, accounts for only 29% of the cases found in people of African ancestry. The absence of the latter information in the past led to several missed diagnoses [11,12].

Understanding of genetic composition of a population helps determine potential drug efficacy as well as the likelihood of adverse reactions, therefore, if not effectively addressed, the lack of diversity in genetic studies could prove dangerous [7,13]. For example, a gene variant found in Zimbabweans slows down their ability to metabolise an antiretroviral drug, efavirenz, and for about 20% of the population that have two copies of the variant, the drug accumulates in their bloodstream and results in hallucinations, depressions, and suicidal tendencies [13,14]. Similarly, in 2008, an antimalarial drug combination (chlorproguanil-dapsone) had to be withdrawn because a mutation resulting in G6PD deficiency among Sub-Saharan Africans had not been properly accounted for [15].

Increasing diversity in genomic research is not only helpful to the underrepresented populations, but also of great value to people of all backgrounds. A notable instance is a study that enabled the development of new drugs that can help in the treatment of heart diseases globally. The study which found that some individuals of African ancestry carry a nonsense mutation on the PCSK9 gene that causes low levels of LDL cholesterol in their blood provided insights into the link between the gene and cholesterol levels [16–18].

In response to the seriousness of the situation and the amount of work that needs to be done, many initiatives such as the 'All of Us' programme have been launched in recent years.

There have also been responses such as the Hispanic Community Health Study focused on building databases specific to the underrepresented groups.

RESPONSES OUT OF AFRICA

Some significant factors that have contributed to the wide gaps in the data include a lack of funding, facilities and indigenous capacity in the Low-to-Middle-Income countries whose inhabitants constitute a large percentage of the underrepresented populations [17].

Within Africa, a consortium called the Human Heredity and Health in Africa (H3Africa) has been addressing this issue since 2012.

Funded by the Wellcome Trust and the US National Institutes of Health, the consortium manages three biobanks that support over 50 projects. It aims to bridge the gap in African genomic data and provides support, training and resources that equip African scientists to carry out genomic studies [13,19,20]. A recent study supported by H3Africa carried out whole genome sequencing of 426 individuals from 13 African countries and identified more than three million genetic variants, thus further highlighting the wealth of diversity to be unearthed in this population [21,22]

Another organisation, 54gene, a genomics lab in Lagos, Nigeria, has garnered a lot of attention in the two years since its inception. The organisation launched a biobank to help address the gap in the genomic databases by collecting blood, tumour tissue and saliva samples from people of African heritage. It has also partnered with Illumina to sequence the DNA in the samples collected and has high hopes of discovering novel genetic variants responsible for cardiovascular diseases, diabetes, and cancer in the African population.

The ultimate goal is to help provide information that would enable individuals of African ancestry to have equal opportunities to understand their risk of developing a disease and thus make decisions to improve their health outcomes [23,24].

Also, contributing immensely to building genomic capacity in the continent is the African Centre of Excellence for Genomics of Infectious Diseases (ACEGID).

This organisation which focuses on the genomics of Infectious diseases, played a huge role during the COVID-19 pandemic, releasing the first genome sequence of the virus from Africa to the scientific community [25].

MOVING FORWARD

Between 2009 and 2018, there was only an increase of about 19% in the level of diversity among the candidates included in GWAS studies; and a majority of the increase came from the Asian population [7]. While even the smallest

hints of progress are valued, if we are going to fully harness the benefits of our genome, we cannot afford to continue to move at a slow pace. It is therefore imperative that more scientists get on involved and that collaborations are established to prevent the isolation of databases. The goal should be to bridge the gaps between all the available databases used globally to draw more robust inferences and conclusions, that can benefit all of us.

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The Mexican Biobank Project

María José Palma Martínez and Andrés Moreno-Estrada (National Laboratory of Genomics for Biodiversity, LANGEBIO -Advanced Genomics Unit, CINVESTAV)

Nowadays, thanks to technological advances and a better understanding of the genomic variants, precision medicine seems to be closer than ever. However, not all populations will benefit equally. Nor will the architecture of diseases be fully understood without the adequate representation of all human diversity [1].

The genomic revolution has decreased the cost of whole-genome sequencing. Despite this, developing countries still find it hard to obtain enough funds to get whole genome sequences of their populations. So, genotyping is a cheaper way to conduct population studies that partially cover the diversity in the country.

The usage of arrays has facilitated the conduction of Genome-wide association studies (GWAS) [2]. However, the representation of individuals with non-European ancestry is very low. In 2019, the number of individuals with Hispanic ancestry included in the GWAS catalog constituted slightly more than 1%. On the other hand, 78% of the individuals are European [1]. This disparity highlights the need to increase the number of association studies in non-European populations since GWAS findings may not be valid for other populations. In order to increase studies in non-European populations, it is necessary to generate whole-genome data for those. Multiple initiatives around the globe are increasing their representation, some of them in Latin America. In addition, to the low representation of these populations, the history of the Latin American population is of special interest to GWAS due to its recent admixture process and complexity that this fact involves.

The Mexican Biobank project (MXB) is a collaborative initiative committed to reduce the currently existing bias in medical genomics [3] by increasing the representation of diverse populations (http://www.mxbiobankproject.org).

The MXB is an international effort funded by CONACYT Mexico, the UK Research Councils (RCUK), and the Newton Fund.

This project is screening the largest nationwide Biobank in Mexico, comprising of 40,000 DNA samples from all across the Mexican territory collected as part of a periodical health survey carried out by the Instituto Nacional de Salud Pública (ENSA 2000) [4].

The project is currently undertaking the genetic profiling of 6,059 individuals through the Multi-Ethnic Global Array (MEGA), containing 1.8M SNPs. It is also incorporating whole-genome sequencing data for 550 individuals.

In the near future, the MXB will also include measured antibody titers on a panel of common circulating pathogens in the Mexican population. In summary, the MXB includes genomic data and health-related information that allow the study of genetic variation specific to the Americas and its possible role in the development of diseases of interest that affect the populations living in present-day Mexico.

Multiple projects using MXB data are under development, including the large-scale profiling of the Mexican population through the analysis of autosomal ancestry, Identical By Descent (IBD) segments and uniparental markers. Other important projects are related to the detection of variants associated with phenotypes of interest. For instance, one of these projects aimed to boost the statistical power of association studies in Latin American populations through the improvement of imputation performance using a Native American reference panel. Genotype imputation is an important step to increase statistical power in association studies conducted on understudied populations. This study was conducted using 50 whole genomes from the MXB and 84 publicly available genomes. The results showed the need to increase the sequencing of Native American genomes in order to improve the imputation performance in Latin American populations [5].

In addition, this study highlights the importance to have a reference panel with representation from the diverse Native American populations showing that it is not enough to include samples from a particular region to benefit all populations in Latin America.

Latin Americans are not a homogeneous ancestry group, important differences in the population composition of each country can be found. Even, inside of each country population stratification is significant.

Thus a big and diverse panel is needed to benefit all the population in Latin America.

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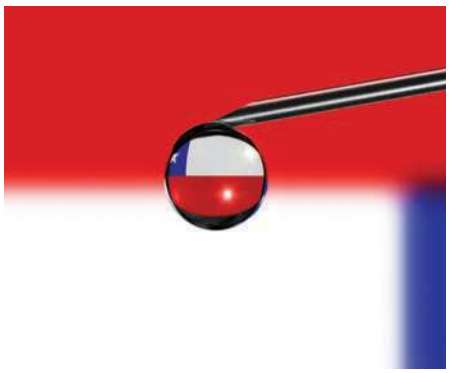
Conducting research in Chile with limited funding

By Professor Raúl Godoy-Herrera (Universidad de Chile)



According to UNESCO, Chile spends 0.38 % of its GDP on research and development, while the UK spends 1.7 %. This budget shortage in Chile, makes it difficult to carry out scientific research publishable in mainstream journals.

These financial problems also affect creation in the Fine Arts, Music and Literature. How have we dealt with the scarcity of resources? First, by turning our attention to biological problems that: (i) matter to the international community of biologists, but that have not received sufficient attention and (ii) do not require a large budget for their study. In this context, we have focused on the larval and adult *Drosophila* behaviour in nature. Second, building bridges with foreign scientists with similar interests.

Third, by seeking grants in Chile and abroad to access laboratories in foreign universities.

In November 1972, at the First Latin American Congress of Genetics held in Sao Paulo, Brazil, I presented my studies on excavation of the nutrient medium by *Drosophila* larvae [1]. In that time, I was an undergraduate student. My teachers, Professors Danko Brncic, Eduardo del Solar and Susi Koref-Santibañez, insisted that I had to go to some Anglo-Saxon university to continue my studies. I was committed to the Popular Unity government of President Salvador Allende and the decision to go abroad was extremely difficult for me. In September 1973 the guns spoke and the painful coup d'état against the Popular Government took place. Things became difficult for me.

It was only in 1977 that I was able to finish writing and publish some of my results on the larval digging behavior. They appeared in Behavior Genetics [2]. The following year, I published another study demonstrating the genotypic basis of the burrowing behaviour of larval *D. melanogaster* [3]. In 1971 and 1973, with Dr Eduardo del Solar, I published two studies, in Spanish, on the distribution of eggs by females of *D. melanogaster* and *D. funebris* [4,5]. With these publications in/on my CV, in 1979, I wrote to Professor Aubrey Manning, Department of Zoology, University of Edinburgh, and Professor Kevin Connolly, Department of Psychology, University of Sheffield. I asked if they would be interested in accepting me as a student. I had read some papers authored by these scientists, raising in me a great interest in working with them. I got a positive response from both professors, which made me very happy. Eventually, I was awarded a British Council Scholarship which enabled me to come to the laboratory of the late Professor Kevin Connolly. There was a stimulating working environment and I met excellent people such as the then graduate student Matthew Cobb, now Professor at the University of Manchester. I also met the recently deceased Dr Barrie Burnet from the Department of Genetics, University of Sheffield.

As a result of the collaboration with Professor Connolly and Dr Burnet, we published a number of papers in well-known journals such as Heredity and initiated a UK-Chile programme of collaborative research and teaching in Behavioural Genetics. The program benefited a number of Chilean students. The collaboration lasted no less than 10 years. In 1988, I obtained a fellowship from The Royal Society and was able to meet Professor Aubrey Manning in Edinburgh. At that time, I spent days including weekends studying the behaviour of larval and adult *Drosophila* in nature. I had understood that it was essential to investigate the behaviour of these organisms in the wild to link properly the evolution and genetics of larval and adult behaviour with ecology of *Drosophila* breeding sites.

Our focus was larval digging behaviour, pupation sites selection, egg-laying sites selection, food preferences of larvae and adults, and social relationships between adults beyond reproduction. Santiago, the capital of Chile, is surrounded by a diversity of fruit orchards and native vegetation. We took a bus to access the orchards. The problems investigated aroused and continue to arouse the interest of medical students, Teachers of biology, MSc and PhD students. Periodically, I receive e-mails from academics and students from prestigious foreign universities, asking for specific aspects of our research and/or requesting information on the behaviour of larvae and adults of isolates of Chilean *Drosophila* species. To all of them we answered in a timely manner and tried to support them in their interesting studies.

When I look back on how I started researching the behaviour of larval and adult *Drosophila* in the wild, I feel that the best reward I have received is the interest, support and joy of my students, and letters and messages from professors and students from abroad. I would like to conclude by saying that budgetary constraints slow down creation in science, the fine arts and the humanities, but they do not stop it. Whoever is interested in a scientific problem will find ways to overcome budgetary obstacles. In this task, the family support and the help of undergraduate and postgraduate students interested in understanding nature is essential.

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Genetics Unzipped – Genes for all: Making sure everyone benefits from genetics research

By Dr Kat Arney

Adapted from Genetics Unzipped podcast (July 15, 2021) Episode S4.14

Powering up African genetics research. Charles Rotimi is the director of the Centre for Genomics and Global Health within the NIH National Human Genome Research Institute at Bethesda, Maryland in the US, and a distinguished NIH investigator. He's also the founder of the African Society of Human Genetics and the driving force behind a major genomics project called Human Heredity and Health in Africa, or H3Africa, which he helped to establish ten years ago and is now wrapping up.

Kat: So tell me about the founding of the H3Africa. What made you realise that there was a project of this kind of scale and scope that needed to happen to understand the diversity of genomes in Africa? We say Africa, it's not a country, it's a continent. It's incredibly diverse. What was the drive and the impetus that started that project?

Charles.: I think again, I'll take us back a little bit to the HapMap. During the HapMap projects, for me, I was extremely conscious of the fact that I was one or two or maximum three persons of African ancestry who were present in those discussions. So to me, that stopped with me and I did not, want that to continue.

I wanted to create opportunity for African ancestry individuals to be a player in the genomic world. And also I was quite concerned that the genomic revolution may not benefit Africa if African scientists and the community members do not adequately participate in genomics. So that led us to the formation of the African society of human genetics, which actually created for the first time the forum for geneticists across Africa, and also non geneticists but interested in doing research in Africa, to come together and to create a forum for that discussion. So under the Africa study of human genetics we started, um thinking about what can we do to make sure that the genomic revolution did not fly over Africa physically. So we started thinking about doing an African genome project which subsequently developed into what we call H3Africa today.

K.: So tell me a bit more about some of the work that was being done under H3Africa and some of the insights that came out of it. What did you actually discover along the way?

C.: So we all know that we all, as human beings, if we trace our roots far back enough we all end up in Africa, so again, that contributed to the scope, wide scope of human genetic variation across Africa. So recently, as a result of sequencing over 18 ethno-linguistic group across Africa, especially among those populations that have not been a part of HapMap, and also thousand genome projects, we were able to make discoveries of 3 million new variants that is currently not in public databases. So again, just highlighting the critical importance of the systematic sampling of African genomes to fully understand human genetic variation.

One of the things that enabled us to make this kind of discovery is that when humans migrated out of Africa about a hundred thousand years ago, that small group of humans that migrated, only took a subset or the variation that excited on the African continent.

So there are part of our human genetic inheritance that can only be studied in Africa populations, again just because of that evolutionary history. So we also make a discovery in terms of the part of our genome that has been under selection as a result of our ability to adapt to different environment. And we make discoveries about selective

protective mechanisms in our genome. It gives things like viral infection, nematodes, infections, and even autoimmune response and pigmentation. So those were all very, very novel signs that H3Africa was indeed delivering on this Initiative. The medical aspects is going to come very soon because we have now genotype close to 60,000 individuals. And the analysis of that data set is ongoing for various diseases like kidney disease, sickle cell, stroke, uh, which is a major problem. So all of those are being analysed and we do hope that we'll make some novel discoveries that will really make us to understand how genetics interact with the environment to increase disease risk or resistance. Yeah.

K.: Were there any things that were particularly surprising to you along this journey?

C.: Yes, for me one of the main surprises, which was a very pleasant surprise, was the fact how ready African scientists were to take responsibility for this project. So I had suspected for some time, but it made it very clear that the issue is really not lack of training or lack of the ability to do this kind of work. It was really a issue of opportunity. And so the way African investigators rallied around this project and made it their own was absolutely a wonderful surprise for me. And a very good one because it really led to the success that it enjoys today.

K.: So with all the information that's coming out, the insights that you're getting, the tools that you're being able to build to understand the genomes of all sorts of different African people. How do we turn all these insights into benefits, into better health? Because Africa is a very broad group of very different countries, including some of the very poorest communities on earth. And there are all sorts of health pressures there. You know, you have food scarcity, you have all sorts of diseases, you have war and conflict. And here in the in the UK, we're talking about genomics in the NHS and all these sort of very expensive, fancy tests and things like that. How do we really make genomics and all this research really useful for the public health benefit of people living in Africa.

C.: I look at it as making incremental progress. I'll give you an example of where genomics has indeed helped quite in a very practical sense. And that is, if you remember the 2014 Ebola epidemic, there was a need to be able to sequence the pathogen in a timely manner. Part of the success story about the ability of African scientists to be able to do that work was the fact that H3Africa has indeed enabled laboratories to be set up that has genome sequencing capabilities. And again, with some additional funding from the world bank. So the scientist in Nigeria and other parts of Africa were able to do sequencing of the Ebola virus and able to track it as he was mutating and also as it was spreading across different parts. [...]

K.: So you set up H3Africa, 10 years ago, we're now in 2021, where would you like to see the field of African genomics in 2031? Just take a leap 10 years into the future. What's it like, what's your vision?

C.: I would just, I would like to see it without boundaries. I just went to see it blow up and grow in any direction that is beneficial across Africa. I want it to be a part of the economic development. I want it to be a part of the scientific development infrastructure. I also would like Africa to be a part of the process of creating a global good, not just recipients of global good. You know, there is a lot about African genome that if we understand it well, it will benefit everybody, not just Africans, because again, that is the root of human evolutionary history. [...]

Sharing family health histories. Dr Laura Koehly is a senior investigator at the National Human Genome Research Institute, with a special interest in helping people unlock the information hidden in their family health histories, particularly focusing on underserved and less privileged communities.

Laura: [...] we can think of family health history as a clinical tool. It's a genomic tool that's used in a clinical setting to do genetic risk assessment. [...] We see that in particular, in the context of families with inherited cancer syndromes it can also be used to personalise health care. For example, individuals with a strong family history will be referred in for screenings often more frequently and at an earlier age. And it can also impact lifestyle recommendations that clinicians might give to patients with a strong family history. But none of that can happen if patients come into the clinical setting without information.

So if they don't know their family health history, then these personalised healthcare services

can't occur. And what we found from our work is that in particular, those in younger generations do not know a fair amount of their family health history. [...] So our Mexican heritage family members who were ages 18 to 30 did not know about 30% of their family health history. Participants who identified as black in these age groups from 18 to 30 did not know about 30% of their family health history in contrast to our non-Hispanic white participants in those same age groups who did not know about 15% of their family health history. So there was definitely a disparity in family history knowledge. And when we looked at older generations, there was no disparity in family health history knowledge.

So our older family members regardless of their racial identity, did not know about 15% of their family health history. So what this suggested to us is that if we could get families to talk about their family health history and to share information about their family health history, we might be able to reduce this knowledge disparity within these minority populations.

Kat: So tell me a bit about the project that you're working on now, who are the people that you're working with, the families, where are they and what are you doing?

L.: [...] In 2015, we developed a workbook. Our goal was really to increase genomic literacy by helping individuals understand how their family health history fit into their increased risk of disease. So the workbook focuses on five disease contexts. [...] And so those who receive the workbook can look at their family health history, and they can fill in the algorithm to evaluate whether or not they are at increased risk of disease. Importantly, [...] it's not just about genetics, it's also about lifestyle and behaviour. [...] And we did not have a large representation in terms of diversity within the sample.

So our participants were primarily white and we were interested, given some of the results we had found in our previous project, whether or not the tool would have meaning and salience to other populations. And so we actively engaged under resource black communities in the Washington DC and Baltimore area to evaluate the workbook and receptivity to the workbook and identify whether we had to tailor aspects of the workbook specifically to the communities that we were working in [...]

K.: Laura's work with the community in Baltimore helping them to uncover their health inheritance has made her reflect on her own role in her family's history too.

L.: Our younger generations do not know a fair amount of information about their family health history. So, you know, often the keepers of that information are the older generation. And you know, I was actually thinking about this last week where my my uncle passed away last week and he's my father's brother. And it was this realisation suddenly that I am now, me and my siblings and my cousins, are now the oldest generation that's living in my father's family. And there's so much that I'm like, oh my gosh. Now I'm playing point in my family. So if my nieces and nephews and, you know, my cousins, children need information in terms of their health history, they're relying on us and I'm not sure we know everything, right? So this family share tool is an opportunity for families to have these conversations with older generations and archive and curate that information. So it's there for younger generations, because at some point the younger generation is going to be the oldest generation, right.

Responsible genomics research with tribal and indigenous communities. Dr Sara Hull is a bioethicist who has worked at the National Institutes of Health for more than 20 years, helping researchers make sure their work is done ethically and doesn't cause harm to the people involved. She has a particular interest in working with indigenous and tribal populations in the US - people with whom the scientific community doesn't have the greatest track record.

Sara: In these studies that we conducted and that many others conducted, we learned that a majority of the public is comfortable with allowing researchers to have fairly broad discretion about future uses, but a substantial minority, 15% - 20%, sometimes higher would say, no. [...] I became concerned about who we were leaving out of research when we had those policies that allowed for broad sharing and for broad future use. And that's actually where I took a dramatic pivot and started really paying attention to the concerns expressed by populations. It wasn't a coincidence that many of those individuals were members of communities that have

historically been excluded from research who weren't realising the benefits of the research. And that made me rethink some of our standard policies, and think about the potential negative impacts they might have on research with some of other populations who are experiencing the greatest health disparities, the greatest burden of disease.

Kat: So when you started to look at these populations who were being excluded or perhaps self excluding from this kind of research? Can you give me some examples of where things have perhaps gone wrong in the past that would make these tribes and these populations very suspicious of scientists turning up wanting to do stuff with them.

S.: So there's, there is a long history of many examples of research that you may have heard referred to as helicopter research. The idea is that researchers come in with their research questions, kind of fly into communities, extract data and samples and bring those away, do their research, publish on it and never returned to the community for those benefits to be realised [...] The research isn't well tailored to be translated into tangible benefits for the community. And in fact, many times has caused stigma and harm to those tribes based on the way that it's reported out in the literature and in the press.

Perhaps the best known example that's been written about quite a bit in the literature I refer to as the Arizona State University case, you'll also hear it referred to as the Havasupai case. It concerns a tribe in Arizona whose biospecimens were collected for use in type two diabetes research, but that were retained and used for secondary research on a number of topics that were quite stigmatising and harmful to the interests of the tribe. And this resulted in a lawsuit and a settlement where ultimately the tribe was able to receive the specimens that had been collected back from the researchers symbolising the end of the research and the ability to handle the specimens respectfully in accordance with the views of the tribe [...]

In my conversations with tribal populations. What I hear consistently simply is that the community wants to hear what the research uncovered and how it might be relevant to, and hopefully ideally beneficial to the community, but even just learning about the science and understanding that it's sometimes takes time to translate results. I've never heard from any individual or tribal group, that there is an interest in receiving results related to genetic ancestry. In fact, if anything, I've heard the opposite, that there's concern that participating in research might lead to genetic information that gets conflated with information about tribal identity or membership. And there are sometimes questions about how we will keep this information separate, but there has been no interest in the receipt of those kinds of results. In the context of tribal research that I've heard.

K.: Why is it important that we do get a diversity of populations of ethnically diverse of tribal groups involved in genetic research in this kind of genetic and health research?

S.: Well, in general, we want those groups to realise the benefits of our investment in research. And this gives us a pathway to close the health disparities gap. And when we're talking about tribal communities, I mean we do see health disparities in other racial and ethnic minorities, but tribal populations in the United States are distinct in part because tribal membership is not really a racial or an ethnic group, it's a political status. Tribes have a special relationship with the federal government and we're obligated to uphold commitments that I can say pretty confidently have not been honoured in the past. We're supposed to deliver healthcare and deliver access to other benefits in exchange for the land that we acquired through colonisation. And there have been persistent health disparities in indigenous populations since colonisation and many of these can be attributed to the social conditions that were created by colonisation. And so to break out of these patterns, we really need to look at the mirror and figure out what it is that we need to be doing differently as a matter of justice.

Genetics Society Summer Studentship

Share your story, Part 2

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 previous 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 final of a two part feature: seven interviews are reported here, and if you would like to know more, please take a look at Issue 85!

Kinga Kołodziej

Kinga Kołodziej – 4th Year PhD student (in neurogenetics) at the University of Leicester, Genetics and Genome Biology department. Keen Genetic enthusiast.

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

The project that I undertook for my Summer Studentship was involving yeast *Saccharomyces cerevisiae* and high throughput screening to identify novel genes that regulate chemotherapeutic resistance.

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)?

It was exciting to be using the "PHENOS" pipeline (PHENotyping On Solid media), which was being developed at the time. Moreover, working on a project that was investigating chemotherapeutic resistance genes, made it feel like I was contributing to something important, since cancer is such a growing issue and current treatments can fail. Research to increase our understanding of the mechanisms behind why it may be the case, and what should be done to improve the effectiveness of therapies, or what alternatives should be offered to current treatments, felt worthwhile. At the time, I was volunteering at a local hospital for the Anthony Nolan Cord Blood Programme, helping blood cancer patients increase their chance of finding a matching donor, as I wanted to make a difference in someone's life. I felt like my studentship project and the volunteering supplemented each other well.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

It really meant a lot to me when I was awarded my studentship. It was my very first "grant application", and I knew that these studentships are highly competitive. So being successful in applying, and knowing that someone believed in me and my capabilities as well, was a really nice confidence boost that I needed at the time. The experience itself was great! I was part of a really nice research group, I was well supported not only by my supervisor, but also the PhD student that I worked with at the time, which really gave me an opportunity to learn a lot. Besides doing lab practicals that were part of my BSc course, this was my very first experience of working in a real lab setting. It helped to reassure me that genetic research is the career path I want to focus on.

"I felt like my studentship project and the volunteering supplemented each other well."

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

During this project I gained experience in performing basic lab techniques, such as DNA extraction and purification, PCR, gel electrophoresis, high-throughput growth phenotyping and Next Generation Sequencing.

I also had a chance to attend a conference in Edinburgh, and present my research findings in front of fellow recipients of the studentship. It was my first experience of presenting in a setting different than a classroom for assessment purposes. It was definitely much more fun, as it gave me an opportunity to meet other like-minded individuals who shared my passion and enthusiasm for science, and who were equally ambitious and driven. Being able to share our results from our own mini-projects felt more exciting and a far more personal experience, than talking about someone else's research, which is what I was used to during my first year!

I thoroughly enjoyed working in the lab and conducting active research, however working with yeast was my least favourite aspect of the whole placement. So ironically the most valuable lesson I have drawn from this studentship is the choice of model organism I would rather avoid working with in the future, if possible haha.

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.

The aspect of this studentship I feel most grateful about that I will forever appreciate, is the fact that this opportunity has opened numerous doors for me. At the time I was applying for various placements, as I was interested in undertaking a Year in Industry as part of my degree. And thanks to having this Genetic Society Summer Studentship Award, and the additional lab experience this early within my career, I got an offer for a placement in Oxford. I believe it was THE item on my CV that made me stand out from the crowd and made me a strong candidate. Having secured a placement at Oxford BioMedica, and working as part of the Cell Engineering Group there, I have gained even more lab experience (this time working with tissue culture and lentiviral vectors) prior to returning for my final year of my Medical Genetics BSc. I have then undertaken an experimental project for my dissertation (working with fruit flies and researching neurodegeneration), which further cemented my eagerness towards academic research. Following my graduation, I have been offered a PhD position (under supervision of Prof. Flaviano Giorgini and Prof. Charalambos Kyriacou), funded by the MRC, in the lab that I have done my final year project. My PhD project was initially somewhat of a continuation, as it was built on the foundations set out in my dissertation work, although the focus of the project has steered in a different direction since then, based on the data I obtained in preliminary screens. I still work with *Drosophila melanogaster* as a model organism to investigate genetic modifiers and mechanisms in neurodegeneration. And I absolutely love my project!

I truly believe that I was able to join the MRC IMPACT DTP programme for my PhD, because my accumulated lab experience was an advantage I had over fellow graduates, making me a strong candidate. Additionally, working with so many different model organisms helped me make informed decisions about my PhD project choices and I am really proud of my journey, as all this hard work started to pay off with visible, measurable outcomes! It was my determination and hard work that got me to the point I am today, but I really think that the Genetic Society Summer Studentship Award, is what made it all possible, so even though it happened so early on, it had a real impact on my career progression.

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

Actually, as a matter of fact, I am hoping to supervise a student on a studentship placement in the Summer of 2022 – with help of my supervisor who will put the application forward, and a young aspiring scientist keen to undertake a project in the summer, I would love to be able to go "full circle" and give back to the scheme, by helping that individual kick-start their scientific career by supervising their work in the lab, just like I was helped only a few years ago. It would allow me to gain some supervising experience and I might even be involved in drafting the project to get that additional grant-writing experience; an essential skill for researchers nowadays! I'm really hoping that the student will be able to secure Genetic Society funding in order to achieve that, as it would have a sentimental meaning to me, bringing back good memories.

I am still working towards completing my PhD, so I have several months of solid work in the lab, followed by write-up period to produce my thesis, and contribute to a publication, but in terms of long-term plans, I definitely hope to continue with a research career in genetics. I find genetics to be absolutely fascinating, and having had the chance to work both in a University setting, as well as industry, I realised that my heart belongs to academia, and that's where I intend to stay. I would like to inspire young scientists-to-be, just like I was inspired when I was only starting my journey. Therefore, my next steps after my PhD will be to do a post-doc, keep working on my portfolio, contributing to research and building my publication record, attend and present at conferences, continue building my network of contacts, form collaborations, continue developing my skillsets, accumulate considerable amount of experience and who knows, maybe one day I will be able to start a research group of my own?

Kristan Piroeva

My name is Kristan Piroeva and I was given the opportunity to take part in the Genetics Society Summer Studentship in the summer of 2019. I recently graduated from the University of Essex with a first-class honours in BSc Genetics for which I was also awarded the Royal Society of Biology Student Award for achieving the highest year mark in the Department of Life Sciences. I am currently a MPhil Genomic Medicine student at the University of Cambridge.

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

Topic: Nucleosome repositioning in chronic lymphocytic leukaemia (CLL)

My project focused on epigenetic changes in chronic lymphocytic leukaemia (CLL) patients – a relatively common type of blood cancer among the Western world population which is characterised with a high number of abnormal B cells and a variable course of disease. The mutation status of the IGHV pathway is used to differentiate between patients with slow disease progression – mutated IGHV (M-CLL), and patients with more aggressive disease – unmutated IGHV (U-CLL), but the biological mechanisms behind these differences are still poorly understood. We were particularly interested to see whether nucleosome repositioning takes place in M-CLL and U-CLL patients and whether CTCF binding is affected within each of the two CLL subtypes – eventually leading to deregulated gene expression.

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)?

Every single aspect accompanying the project was really exciting and extremely interesting to me as it was the first "real" project that I got the chance to work on.

Back then I was new to the bioinformatics field but was really motivated to progress into this area. Thanks to my supervisor – Dr Vladimir Teif, and his PhD student – now Dr Chris Clarkson, I significantly enhanced my bioinformatics skills and by the end of the project I was confident in dealing with genomics data and performing unsupervised computational analysis.

However, the feeling of being part of a research lab was surely the best part of doing research in my opinion. I really enjoyed discussing science and communicating ideas for our projects not only with the lab members of Teif lab but with the members of other collaborating labs as well. The weekly lab meetings were my absolute favourite as we used to gather at the nearby cafeteria and have coffee and cake while going over newly published science articles.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

It really impressed me how the Genetics Society gives students the opportunity to get involved in research and, more specifically - gives them the freedom to choose the area in which to work. At the workshop in Edinburgh, I met and communicated with students from different science backgrounds and in this way, I gained insight into many diverse research areas. The studentship most certainly opens your eyes – it helps you decide whether science and research are your thing or there are other areas in which you might have a greater interest.

Personally, the whole experience was most certainly invaluable, highly motivating, inspiring, and eye-opening! I definitely recommend it to other fellow students as I think it is a great way of pursuing your interests and setting up the path towards a future career. Hopefully in the future the Genetics Society will be able to offer not 30 but 300 studentships each summer!

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

I gained an impressive set of skills during the studentship – from bioinformatics, coding, analytical and statistical skills, to graphical, science communication, writing and presentation skills. I learnt how to be creative, how to think and how to be a scientist!

However, as a person whose biggest fear was public speaking, one of the most valuable skills that I gained during the studentship was how to present with confidence and on top of that - to enjoy it. Over the summer my supervisor asked me to present several times to overcome that fear, however, the workshop in Edinburgh was definitely the turning point! The talks and motivation from the organisers' side along with the excellent presentations given by fellow students completely changed my mindset and made me realise that anxiety was not in my favour at all. This experience undoubtedly helped me ace my final year project presentation as well as any other presentation after.

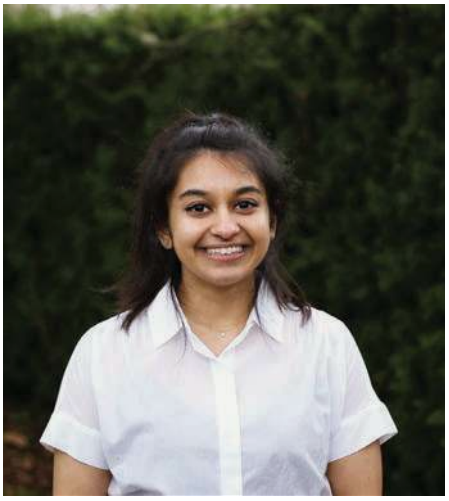
Tell us about where you are now, the progress in your career since the Studentship programme and what impact it had on your career path.

After completing the Genetic Society Summer Studentship, I became a part-time research assistant at the Teif lab supported by the Undergraduate Research Opportunity Programme (UROP) of the University of Essex which gave me the chance to further expand my project and look at other epigenetic changes in CLL patients. The manuscript is in preparation and hopefully not too long from now the work will be ready to publish.

Undoubtedly, the Genetic Society Summer Studentship gave me the start needed. Since then, I've been more than motivated to follow the science path and progress. My determination to keep growing in the Genomics field and the support and encouragement of my supervisor resulted in my application to the MPhil Genomic Medicine programme at the University of Cambridge. Here, I slightly shifted from the epigenomics field but gained lots of new knowledge and skills which allowed me to work with 100,000 Genomes Project data.

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

My main goal is to follow my passion for genomics and cancer research and keep growing in these fields. I'm definitely looking forward to seeing where my interests would take me. Hopefully one day I'll have the opportunity to apply all I have learnt throughout the years in my country and help more people diagnosed with cancer and other disorders benefit from the power of genomics, similar to the way 100KGP is helping people and families here in the UK.



Nikita Telkar

I'm Nikita Telkar, a current PhD Candidate in Medical Genetics at the University of British Columbia in Vancouver, Canada. I completed my BSc in Biomedical Genetics from Newcastle University, and my MSc in Genetics of Human Disease from UCL. My specific interests in genetics lie in cardiovascular, developmental, population and computational genomics.

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

I undertook my Summer Studentship project at the Newcastle Institute of Genetic Medicine in 2016, under the supervision of Professor Susan Lindsay, and it involved mapping the WNT5A protein in the human embryonic gut. Using immunohistochemistry, I detected the presence of the protein in the human tissue of a Carnegie Stage 20 (50 days post-conception) embryo, the first instance worldwide. Using the MAPaint software, I annotated the expression throughout the oesophagus, intestines and the anus, and I transformed the 2D expression patterns in a 3D video version using the Amira software. An unexpected finding was WNT5A's expression in the embryonic heart and kidney, which had not been reported before. My contribution is still available on the Human Developmental Biology Resource (HDBR) website, which is managed by the MRC and Wellcome Trust.

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)?

Having been given an opportunity to lead my own research project, and to have been awarded funding for it, was incredible. I think the most exciting part was knowing that I was working with human tissue as opposed to animal or plant, but even more so that it was embryonic human tissue. I had only learnt a little about *embryonic* development during my classes till that moment, and physically seeing those microscopic sections was mind-blowing – the embryo I was working with was only about 1cm long!

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

The best part about being awarded the studentship was the confidence and reassurance that I received that the field I had chosen (i.e., pursuing my degree in genetics) and the one that I wholeheartedly loved, was one that I was actually good at.

It gave me the opportunity to meet the other awardees of the Studentship for that year at the Workshop in Edinburgh: students of my caliber and as enthusiastic about their research as I was. I would undoubtedly recommend applying for the Studentship to any student, because the immediate achievement it gave and long-term exposure of the Award stays with me still every day.

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

I learnt a phenomenal amount about human embryonic development and genetics, more so than I would have if it had not been for this project. Funnily enough, my supervisor for the project turned out be the module leader for a core module that I undertook during the academic year after the Studentship (in 3rd Year). The knowledge and information that I had gained during my project helped me incredibly to understand the undeniably complex module – my course mates discussed how that module was the most difficult of our final year. The project gave me an in-depth view about a research area that I was already curious about and turned out that I was actually good at!

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.

The Studentship was the highlight of my undergraduate course at Newcastle professionally, and the doors it opened and still opens is undeniable. My poster for the project won an award at Newcastle's University-wide Research Scholarships Exhibition, as well as in the Faculty of Medicine. My report of the same was selected to be published in Genetics Society's January 2017 Newsletter. The Studentship was instrumental in getting my MSc acceptances from UCL and Imperial College London (of which I chose UCL). I got to work with my first-choice project/supervisor for my MSc project, which was in association with the Wellcome Sanger Institute. It involved assessing the correlation of cholesterol markers across diverse ancestries and building polygenic risk scores (PRSs) leading to a second-author publication in Nature Communications. And currently, tying all my experience together, my PhD project is in human developmental genetics. I'm investigating novel small RNA species in the normative human placenta, and how they correlate with inherent biological traits (such as sex and ancestry) and also to DNA methylation patterns. As such, I am developing a streamlined workflow for the optimization, processing, and analysis of low-quality RNA-seq data. My previous knowledge in embryonic genetics helped me tremendously in understanding placental biology, which can be quite an abstract organ to understand.

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

I view my PhD as the step in my career where I want to learn the use of as many computational tools and to gain as many skills as possible. My aim upon graduating is to work with a company that incorporates ancestry and underrepresented populations in genetic studies. There are so many populations that exist that still don't have their own version of a reference genome, and given that allele frequencies can vary widely between differing populations, accounting for ancestry or ethnicity is paramount any time the results from any genetic study are considered. Building a reference genome for the population that I belong to, the South-Asian population, is my ultimate goal. Founding a diagnostic company is also in the cards, and would be very cool.

Oluwaseyi A Pearce

Hi, I'm Seyi Pearce, a second year PhD student at the University of Sheffield, studying the interactions between ageing and Parkinson's disease. I graduated with a First-Class Honours in Biomedical Science (Bsc) from the University of Sheffield as well. During the second year of my undergraduate degree, I was privileged to have my Summer Studentship funded by the Genetics Society. This enabled me gain valuable laboratory skills which went a long way in helping secure my PhD.

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

The glucocorticoid receptor (GR) belongs to the nuclear receptor family which controls many cellular processes. GR maintains genome stability and is implicated in disease as reduced genome stability is crucial in tumour formation and ageing. My Summer Studentship project was focussed on identifying mechanisms by which GR maintained genome stability.

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 remember being excited when I received the email confirming funding for my Summer Studentship. I remember being eager to get into the laboratory and experience the day-to-day realities of scientific research. I found my research topic to be very interesting. I enjoyed working with zebrafish, particularly because of their genetic potential and optical transparency which allowed me to take incredible images. An added bonus is that the group I was working with (the van Eeden group) was filled with approachable people who were equally interested in their respective research topics. I thorough enjoyed my Summer Studentship, from the laboratory skills I gained which I still use today in my PhD, to the friends I made.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

One of the best things about this experience was meeting other students in Edinburgh at the Summer conference organised by the Genetics Society. Presenting my research topic helped me build my communication skills which is very important for any research project. Likewise, learning about the various research topics funded that year was very informative. The conference was a great avenue to make connections, friends and learn about possible paths to obtaining a PhD. I definitely would suggest this Summer Studentship to fellow students.

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

I was able to build on my organisation, scientific communication (both written and verbal) and perseverance, among others. Without a doubt, these were helpful in completing my undergraduate degree as I was able to handle my work-load better during the following academic year and improve the quality of my final year dissertation.

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 a PhD. After completing my Summer Studentship, I began my third and final year of Biomedical Science. I graduated with First-Class Honours and was successful in securing this PhD soon after. Having been successful at securing competitive external funding for this Summer Studentship, as well as the accompanying laboratory experience, made me competitive during my PhD application. Likewise, I was able to decide to continue with postgraduate research thanks to my experience during this Studentship.

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

I plan on pursuing a career in industry after completing my PhD.

Reuben James

My name is Reuben James; I am a molecular entomologist currently undertaking a PhD project in the Hogenhout lab at the John Innes Centre, Norwich. I undertook my undergraduate Bsc Biochemistry and Genetics degree at Swansea University, which is also where I undertook my summer studentship project under supervision from Dr Miranda Walker neé Whitten.

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

My summer studentship project was to use molecular techniques to identify gregarine parasites in insects. Gregarine parasites are protozoan, single cellular organisms distantly related to the harmful *Plasmodium* and *Trypanosoma* parasites. They are mostly harmless, but the family now includes the potentially lethal *Cryptosporidium*. As insects such as meal worms are being increasingly investigated as sustainable food sources, we deemed it important to assess which parasites exist in these insects, and what they may produce.

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)?

For me the most exciting part of the project was being able to dive into an area of research that I had little experience in, but was increasingly intrigued by. I was able to learn about the importance of molecular entomology in both medicine and food science, whilst also learning important skills which I have used throughout my career since. Further, it was exciting to develop my scientific mind and take control of my own project, backing up my ideas and suggestions with good reasoning. The project also acted as a good introduction to molecular biology techniques such as PCR, microscopy, and mass spectrometry.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

I would certainly suggest this studentship to fellow students, especially those who are unsure if they want a career in research. The studentship allows one to steer their own project in a direction of their choosing whilst also learning what it is like to work with other people in a lab environment. The workshop at the end of the studentship allows one to present their project to their peers and connect with others in a similar career stage to themselves. For me, this studentship allowed me to explore an area of science that has since been the focus of my career. I gained confidence in my innovative and investigative abilities, as well as in presenting my work and networking with others.

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

The techniques and skills I learned during my summer project allowed me to have a more fulfilling and successful final year research project, with better time management and organisation. This allowed me to undertake my project whilst learning and revising my other modules efficiently. Further, the molecular techniques I had used continued to come in useful throughout my final year, enabling me to get more out of my final year project than if I had had to spend the first month training in these areas.

"After the studentship programme I undertook my final year project with the same supervisor looking at symbiont-mediated RNAi as a control method for pest insects. This allowed me to further my interest in molecular entomology as well as gene-editing techniques."

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.

After the studentship programme I undertook my final year project with the same supervisor looking at symbiont-mediated RNAi as a control method for pest insects. This allowed me to further my interest in molecular entomology as well as gene-editing techniques. Fortunately, I managed to complete the research for my final year project before the COVID-19 pandemic prevented further lab time. The studentship project and my final year project provided me with the skills and experience I needed to start my PhD at the John Innes Centre in Norwich on the optimization of tools to study gene function in pest invertebrates, under the supervision of Professor Saskia Hogenhout, and I am now 8 months into my first year. More specifically, I am looking at ways to facilitate CRISPR-Cas gene-editing in the green peach aphid, whose strange reproduction cycle makes this a difficult task. It is fair to say that without the studentship and guidance from Miranda, I would not have developed my interest in molecular entomology, and it is highly unlikely that I would have ended up in the fortunate position I am now in.

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

After my PhD I hope to take what I learn about gene-editing in insects and apply it to the study of insect genetics and parasite-vector interactions. There are many disease vectors such as the Tsetse Fly and the Kissing Bug, which may be better protected against if more was understood about their interactions with their harmful parasites. Further, the study of insect-plant and insect-phytopathogen interactions is something I would be interested in as there is much yet to be explored in this area, which has a big impact on agriculture worldwide.

Stanley Liang

My name is Stan and I'm a Canadian student who has recently graduated with a degree in Biochemistry from the University of Victoria. Since then, I have transitioned my focus to neuroscience research. I'm particularly interested in understanding the molecular genetics involved in learning and memory.

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

For my Summer Studentship at Kings College London, I undertook a project to optimize immunohistochemistry protocols for the detection of members of the imprinted gene network in developing mice embryos in the Charalambous lab. Unlike other genes, the expression of imprinted genes is unique in that the genes are only expressed from one allele and their expression is dictated by whether the gene was inherited from the mother or the father (eg. The gene *Zac1* is only expressed from the paternal allele). Ultimately, my project aims were to map the expression of some of these imprinted genes during embryonic development.

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 for me was the responsibility I was offered and the international experience. Prior to the Genetics Society summer studentship, I did not have any experience with real research. In our courses, the laboratory projects were always expected to produce a result. There was a deep sense of accomplishment and triumph when I was finally able to generate usable data after troubleshooting numerous failed experiments. After gathering my data, I prepared my presentation for the Genetics Society Summer Student conference. It was the first time I would need to present in front of a large crowd full of people! I remember during my presentation I was so nervous and terrified... But in the end, I was extremely proud of the presentation I gave and ultimately what I did for my project.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

I honestly believe that the short time I spent in London for the studentship was a life-changing experience. Not only I obtained new scientific knowledge, but I also underwent tremendous personal growth. Furthermore, this was a great opportunity to make friends with like-minded people; many of whom I continue to stay in touch with.

My advice for my fellow students is: Do not pass on opportunities/challenges that will facilitate development in some domain of life. And personally, I think that the Genetics Society summer studentship is exactly the type of opportunity to facilitate growth while providing a wonderful experience. All the great things I learned during my project has stuck with me and helped me generously in my career goals.

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

I think the most valuable skill I developed during my project was how to think critically about new information and how to frame scientific questions. The critical thinking skills I developed really helped me to excel in my final year of undergraduate studies, specifically, when dealing with more abstract concepts. Furthermore, the laboratory time really provided me with the extra boost of confidence that other undergraduate students did not get.

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.

Recently, I received a scholarship to study neuroscience in San Diego (California) and investigate the mechanisms behind Fetal Alcohol Spectrum Disorder. Interestingly, the knowledge I acquired during my GS studentship played a role in securing this position – to say the least, it has been a serendipitous arrangement. If I had not studied imprinted genes during my GS Studentship programme, I would not have the knowledge to compose my current research project.

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

After I complete my master project, I plan on pursing medical school. Ultimately, I hope to continue a career in translational research. I'm particularly interested in bridging the gap between basic science and patient care. From estimates, it takes roughly eight years for bench science to be applicable inpatient care! I'm interested in helping to facilitate and accelerate that transition by pushing patient-oriented research in my future field.

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)?

For me the most exciting part of the project was being able to dive into an area of research that I had little experience in, but was increasingly intrigued by. I was able to learn about the importance of molecular entomology in both medicine and food science, whilst also learning important skills which I have used throughout my career since. Further, it was exciting to develop my scientific mind and take control of my own project, backing up my ideas and suggestions with good reasoning. The project also acted as a good introduction to molecular biology techniques such as PCR, microscopy, and mass spectrometry.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

I would certainly suggest this studentship to fellow students, especially those who are unsure if they want a career in research. The studentship allows one to steer their own project in a direction of their choosing whilst also learning what it is like to work with other people in a lab environment. The workshop at the end of the studentship allows one to present their project to their peers and connect with others in a similar career stage to themselves. For me, this studentship allowed me to explore an area of science that has since been the focus of my career. I gained confidence in my innovative and investigative abilities, as well as in presenting my work and networking with others.

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

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Tumie Ntereke

Hi there, I'm Tumie! I was born and raised in Botswana and moved to the UK for higher education. I am currently a second year PhD student at the University of Leicester and am based at the Leicester Cancer Research Centre. My research aims to determine the putative use of exosomal cargo, a source of nucleic acid signatures, as a blood-based biomarker in breast cancer patients. I am an avid reader and incredibly passionate about diversifying the Academy to propagate equity in research and its findings.

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

My summer project explored the causative mutation of a condition called Eosinophilia and I was based in Dr. Ed Hollox's lab at the University of Leicester. Dr. Hollox and his collaborators had recruited a family known to be affected by the condition, and collected blood samples. Whole exome sequencing was carried out to identify the causative mutation. Prior to my project, 7 functional, and potentially causative variants were identified, by applying hard filters to the sequencing data. I was able to design primers, prepare the new samples for sequencing and through analysis of the sequencing data, propose a causative variant of eosinophilia. Upon validation of our findings, this novel work was written up and was recently published.

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 think the entire experience was very exciting for me, especially learning new techniques in the lab. I still remember the first day I got to make an agarose gel; it was the most intense 4 minutes of my life watching it go in the microwave, praying it didn't explode. The project also gave me a taster to the bioinformatics aspect of genomic research which I thought was fascinating. Another highlight was the workshop in Edinburgh which gave me a chance to meet my colleagues and learn about the various things they had been up to in their respective labs that summer. The social events also gave us time to network and it was a great environment to foster friendships with likeminded people.

What meant for you to be part of this studentship, would you suggest it to other fellow students and why?

I would definitely recommend the studentship to anyone interested. The Genetics Society was the first learned society I had ever joined and the summer studentship my first ever grant! This turned out to be quite a pivotal factor in moulding my journey after that summer. Being able to experience research outside lecture rooms and practical classes really shifted my perception of academia. I can only hope that it could be an encouraging avenue for students who might not be are not quite sure of what they want to do after completion of their undergraduate degree.

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

I gained some amazing, and incredibly useful transferable skills. As I undertook the studentship in the penultimate summer leading to my graduation, I was able to directly utilise skills such as time management and qPCR analysis which I had gained from the studentship and applied them during my final year lab project. Because the methods I used were similar to those I had learned over the summer, I hit the ground running. The entire experience made me appreciate the collaborative nature of research. To think it started with patients attending a consultation in clinic then went on to the "wet" lab bench aspect was mind blowing to me. I also think the collaborative nature of research is often underplayed in lecture theatres during undergrad but teamwork really does make the dream work!

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.

Upon completion of my undergraduate degree in Medical Genetics, I had my eyes set on doing a PhD. I had really enjoyed the clinical relevance of my studentship project and knew I wanted to stay in the translational research domain. As such, I applied for various programmes but ended up undertaking a project with Professor Jacqui Shaw working on exploring exosomal cargo for utility in the liquid biopsy in breast cancer.

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

Well, if we take it back to Edinburgh 2017, Dr Kat Arney gave a talk about her journey into science communication. I still think about how whilst she was talking, I had my "Aha" moment. I've always loved bench science, but I also appreciate the need for it to be disseminated in a digestible manner to non- scientists, especially with translational research which has a direct impact on patients. The public need to be aware of cancer risk factors, symptoms and the possible impact of research on their diagnosis, treatment and disease outcome. I belong to an ethnic minority and I know a lot of people in my community tend to prefer to take in information from people who look like them, and have had a similar background and life experience. Ideally, my next step is to secure a postdoctoral position, whilst also cultivating my various digital science communication platforms.

A day in the work-life of a laboratory scientist intern, Adriana Macko

Industrious Science

By Adriana Macko and Margherita Colucci

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

This issue brings you the first student-focused interview of the series, with Adriana talking about her internship experience at Illumina. Adriana reflects on her student and intern roles, considering the advantages of her experience - I am sure her advice will be inspiring to students looking for intern roles.

Margherita: Thinking about your educational and career path … What brought you to Biomedical science/what sparked your interest, and what do you enjoy the most in this field?

Adriana: On the trivial side, I've spent a lot of my childhood in the hospitals as I've been suffering from eczema for a quite long time. The medical environment encouraged me to be curious and ask doctors many questions about the disease itself. Eventually, doctors became my friends. They would kindly explain the physiological response to the disease in simple terms.

My interest in medicine grew more each year. However, I've noticed that I was keen to explore medicine from a more detailed perspective, I developed a passion for the molecular angle of it. I was not certainly interested in the ways to cure disease as in prescribing medicines. This approach seemed to me to be a short-term solution for addressing the symptoms. Instead, due to my experience as patient, I wanted to find a medical long-term solution. I think this part of my childhood determined my interest and curiosity in science and in the understanding of the root causes of the disease at a molecular level and its possible prevention.

Fact File

Originally from Lithuania, I moved to the UK to broaden my horizons and dive into studies of Biomedical Sciences at Royal Holloway, University of London. After my second year of studies, I took a year of absence to complete a one-year internship at Illumina, the company known for its sequencing that allows researchers to ask virtually any question related to the genome, transcriptome, or epigenome of any organism. In the past 10 months I have been working as laboratory scientist intern and I am testing my potentiality as a scientist.

M.: How did you decide to get an internship experience? Do you have specific career ideas, it was an unexpected opportunity, or the option of an year in industry was already included in your course, maybe the reason why you chose your course in the first place?

A.: Illumina internship was an unexpected opportunity. The idea of industrial work experience came when I was preparing for short summer placements that were organized by my university. I was not aware of opportunities outside academia. I accidentally came across offers for one-year internships while getting ready for CV workshops and looking for tips online. I was extremely thrilled when I found out that many companies organize internships for students who are mid-way through their degrees! My expectations were not high, because I failed an interview at my university for a summer placement. Nonetheless, I've decided to apply for internships outside university, but without great expectations. I took it as a challenge to get out of my comfort zone, and a chance to gain more experience at attending interviews. And certainly, the application process becomes more enjoyable when motivated by curiosity, and not by competitiveness. Honestly, I did not believe that a generic student might get an internship in a world-recognized company.

M.: Do you think universities are doing enough to support the option of a year in industry?

A.: From my perspective, the universities offer the option of a year in industry, however the search for an internship and the application process is not very supported.

I can't talk for all universities, as yet my university did a minimum to introduce the industrial internships. My lecturers were supportive and happy when I announced that I got an offer for an internship, however, I have never heard anyone talking about the possibility to get one. The industrial side of science is not empathized as much as the academic one and, as a student, I did not realize how important is the role of science in industry.

M.: How did university prepare you for this internship?

A.: As I mentioned, students had a chance to attend a workshop focusing on the CV and cover letter. I shall say that the most valuable experience was an interview followed by a detailed personal feedback. This was useful for my Illumina application process. University's career & Employability Services kindly answered my further questions regarding assessment day.

When talking about skills required for a lab-based internship itself, I believe that the most important ones are basic laboratory working techniques. Fortunately, my course involved a fair amount of laboratory practice where I had a chance to become a confident laboratory user. Eventually, I noticed that writing a lab report after the practice sessions was one of the most efficient methods to gain experiment planning and analytical skills that were handy during my internship.

M.: How many of your course mates went for a similar path?

A.: As far as I know, my course mates seem to continue with the 3rd year of the studies without any internships this year. Unfortunately, many short summer placements were cancelled due to the pandemic. Being part of a big course and not knowing everyone individually, I might not have all the information on this.

M.: What do you think you will do next after this experience and after completing your degree?

A.: This internship gave me an amazing opportunity to meet many scientists and ask about their career paths. Pleasant conversations provided detailed insights into postgraduate studies and inspired me to strive for more knowledge. Illumina's environment definitely sparked my interest in the field of genomics! Being genetics my primary focus, I never thought that there were other areas that I would enjoy exploring even more. For the time being, I aim to progress in postgraduate studies in the area of genomics. I think further studies would fulfil my current desire to know more and lead to more

"The assessment day was stressful to a certain point. Illumina created a great atmosphere and, overall, it was an exciting experience. However, knowing that you're being observed is quite a terrifying experience to everyone perhaps. "

specific knowledge that I could apply later when going back to industry roles.

M.: Evaluating your experience at Illumina so far… Could you describe a typical working day in a few lines?

A.: There is no typical day! I might have some routine tasks, but my work at Illumina is dynamic and fast-paced: there is always something new going on. The projects and daily work may vary dramatically: one day I might plan my own experiment and work on it, another day I might be dragged into a completely new project and help other scientists. In a day I would attend collaborative meetings, plan further project steps, work on them and analyse or present my results.

M.: What are you passionate about this experience, what is the best part for you? And the worst/ most difficult?

A.: My passion for science went rocking this year! Being a university student and having loads of material to learn is sometimes overwhelming and it can be difficult to see the bigger picture of science. Working in the industry brings a realization that science on a big scale can actually change people's life. From a social perspective, my favourite part is meeting new people. Seeing the passion in people's eyes is the best motivation to strive for a career on the same path. Learning about different backgrounds brings so many new perspectives: I believe that one of my most interesting discoveries was to realise that science is extremely multidisciplinary and there are more options than to be a "scientist". I find it fascinating how science can be employed within businesses.

The only difficulties I faced were due to the pandemic. Although I gained experience, I haven't seen Illumina at its full capacity. Meeting people virtually does not build the same bonds as in person.

M.: How was the training organized (if any)? How did you prepare for this?

A.: I'm lucky because my team includes the "application trainers", so I've been taught by official trainers! I've received lots of training and became an independent and confident lab user. In terms of preparation, our trainers are really supportive and send all essential materials before training. Apart from the scientific background, there is no specific preparation needed to attend the training. But the knowledge of NGS technologies is always a plus!

M.: What do you think this experience is going to bring to your curricular experience (do they complete each other; would this industry experience e give you a better understanding of the curricular material)? What skills did you gain?

A.: Experience at Illumina will be beneficial in my third year of studies. I widened my knowledge about cancer genomics and general knowledge of genetics. I find fascinating how much and how fast you can learn through experience and not just from textbooks. I've gained technical laboratory skills as well as transferable skills. My learning journey has begun from performing basic laboratory technician tasks, such as serial dilutions and concentration calculations, to confidently planning certain aspects of my projects or designing the experiment and presenting the acquired data to other team members.

Furthermore, I am excited to take the modules related to genomics and bioinformatics next year. I believe it would be satisfying to attend mentioned lectures and have background knowledge already! But my mindset towards science has changed since the beginning of the internship and I think that this will help me to understand faster other concepts taught at university and from a different perspective. I will approach some concepts differently, from a more practical than the theoretical perspective and this should improve my performance.

M.: Your point of view and experiences may help many other students in having a better idea on how to approach an internship and complete the first steps towards a career in genetics… What to expect as laboratory scientist intern?

A.: First and foremost, to be treated as a member of the team. You will have responsibilities and an internship needs to be treated as a proper job. However, as an intern, you need to be ready to learn a lot. I believe your curiosity defines how much valuable experience you will gain from the internship. Aside from the valuable lab experience, you also start to live according to the company's values. You will embrace communication, collaboration, and flexibility in your everyday life.

M.: How was the process to find an internship opportunity?

A.: It is not difficult if you are very motivated to find one. I shall say do not hesitate to email companies or research leaders if you are interested in the job they are doing. It's important to start looking at a specific time when internships are offered (around November – February).

M.: What was the hardest part in securing it and which set of skills / experiences you think were valued the most?

"As a student, be an open book and absorb as much from other people as you can. Also, it might sound boring, be yourself in the application, because this is what people like."

A.: The assessment day was stressful to a certain point. Illumina created a great atmosphere and, overall, it was an exciting experience.

However, knowing that you're being observed is quite a terrifying experience to everyone perhaps. Each task focused on a specific set of skills. For example, the majority of tasks focused on team working and communication skills, which are crucial working in a company. When applying to a scientific role, it is important to secure your calculation abilities – it is worth remembering basic concentration formulas and calculations.

M.: Any suggestions on how to find (even websites suggestions) and to secure a great internship opportunity? What do you know now and wish you knew before applying?

A.: There are many websites such as (prospects.ac.uk; uk.indeed.com; glassdoor.co.uk; targetjobs.co.uk). If there is a particular company where you would love to work, I suggest checking its website or its recruiting sites. LinkedIn is a great place to find opportunities too, I would definitely recommend it to students who don't have a profile yet! Also, I want to highlight that time is important in the application process.

Thus, the majority of internships would be offered for second-year students, in the middle of their degree, and missing a deadline due to the lack of awareness is truly painful.

Besides that, the advice I would give to the 'past' self is to not be focusing on only one field of science. My mindset was fixed on a particular aspect of science, even though I didn't had a chance to explore any others. I learned that any opportunity is valuable. As a student, be an open book and absorb as much from other people as you can. Also, it might sound boring, be yourself in the application, because this is what people like.

M.: What do you think that university should do to better introduce students to the "industry side of science"?

A.: I think it would be useful to give a few workshops or seminars about the different paths that students might take during or after their degrees. Studying biomedical sciences and other life sciences doesn't lead to academia all the time. There is a new world of industrial science that is as great as academia. And it is important for universities to support and encourage students to explore the diversity of scientific career paths. Additionally, career events with alumni interns would be beneficial and inspiring for the first or second-year students.

In the email, you write this "I would love to tell other undergraduates how one year of internship outside academia might shape one as a person and as a professional". It would be great to know more, please, feel free to expand on this!

When I applied for an internship, I expected to get only laboratory experience. I could not imagine that one year outside academia could shape me both as a person and as a professional practitioner. I enjoyed a transition from university to working environment – from life where every minute filled with study pressure to a 9-5 work routine. Work itself is intellectually fulfilling and there is a pleasant feeling of achievement after the end of each workday. This year has been a wonderful balance of getting knowledge and experience in an industrial working environment, as well as shifting the priorities to focus on self-development. Moving to a new city and joining a new company during the pandemic improved my interpersonal skills, I met a diversity of open-minded people that keep inspiring me since day one. Even though getting practical experience is invaluable, I cannot emphasize enough how much I appreciate the wisdom I acquired from these fantastic people.

Thank you very much, Adriana, for the interesting insights into your internship experience!

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

Industrious Science

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**.

A Genetics Society Workshop

Communicating Your Science

Applications are now open for the 2022 workshop, to be held at Chicheley Hall, 25-27th April 2022. Please see the website for further details and how to apply: https://genetics.org.uk/grants/comm-your-sci/

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

Helen Keen
(Multi-award winning writer and performer)

First Create The Media
(Led by award-winning writer and boadcaster, **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 UK travel, accommodation and meals for all successful applicants

Applications are open, deadline midnight, 28th February 2022

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

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The Newsletter is growing... and needs you!

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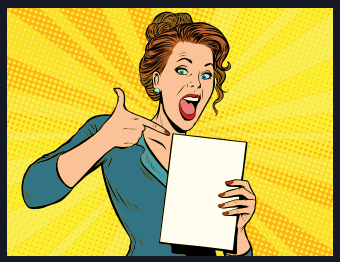

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Climate Change, The Board Game

By Dr Michela Leonardi (University of Cambridge)

It is a truth (almost) universally acknowledged that past climatic fluctuations heavily influenced the distribution (1,2) and evolution of animals and humans, both in terms of genetic patterns (including geographic structuring) (3-7) and adaptation (8).

At the same time, given the complex interactions between climate, natural selection, species distribution, and demography it may be hard to effectively communicate how this kind of influences work.

To do so, I created "Climate change – the board game", a free educational resource that, while playing a game, shows how species migrate, evolve or go extinct as a response to climate changes.

The inspiration behind the game

Climate Change – The board game is based on the research performed by the Evolutionary Ecology Group, Cambridge, within the ERC Consolidator Grant 647787 "LocalAdaptation".

The group developed continuous paleoclimatic reconstructions for the last 800,000 years (9-10) and climate-informed spatial genetic models (CISGeM) that use such paleoclimate to reconstruct past population sizes, local movements, and range expansions leading to the genetic patterns observed (3,4).

How does it work

Each player is a species, living in a world where the climate may change every now and then. Every species has its own DNA and collects mutations through time, that potentially allow it to adapt to new habitats. The game explores in a rigorous but simple way complex scientific concepts such as adaptation, evolution, speciation and extinction. In the near future, this website will contain additional material to help better understand the science behind the game. There is also the opportunity to explore the effects on animal species of the current human-induced climate changes with respect to the natural ones.

The game is for 2 to 5 players and is suitable for people aged 8 and older. The approximate duration is 30 minutes. It has been designed as

an educational resource for schools, but can be played also with family or friends.

The materials and the rules can be freely downloaded from the website to be printed at home, or the game can be played online for free (see website).

Useful links

Game website: https://michelaleonardi.netsons.org/climate-change-board-game/

ERC Consolidator Grant 647787 "LocalAdaptation": https://cordis.europa.eu/project/id/647787

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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, David Sherratt, and Adam Price.

London Genetics Network meeting

December 3rd 2021

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 annual meeting on the 3rd of December 2021 which was generously funded by the Genetics Society.

The enthusiasm with which the Network was received was overwhelming. We had over 230 registrations from over 30 different institutions across London and the surrounding regions, many poster submissions and viewing numbers hovered between 90-115 viewers throughout the day. In addition, we have received many messages of encouragement from individuals. In terms of Twitter impressions: there were 233 total tweets, which had the potential to reach 31,680 people #LGN2021.

The day kicked off with an informative keynote talk by internationally renowned scientist Professor Gudrun Moore (UCL). The schedule of speakers included talented early career researchers Dr Anna Gui (Birkbeck), Dr Wikus Barkhuizen (UCL), Dr Kaili Rimfeld (KCL), Baihan Wang (UCL) and Dr Qinqin Huang (Sanger). After lunch we were treated to a keynote talk by Professor Segun Fatumo (LSHTM) and excellent talks from Dr Jonathan Coleman (KCL), Dr Yalda Jamshidi (St Georges) and Dr Greg Findlay (Crick). Professor Angelica Ronald (Birkbeck) concluded the day with feedback and plans for an in-person social in 2022. The speakers showcased London's scientific excellence as well as diversity in terms of ethnic background and gender. 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, multi-ancestry GWAS, intergenerational transmission, biobanking, the African genome, behavioural genetics, consanguinity, pre-natal and infant development, large-scale phenotyping, ascertainment biases and saturation genome editing.

The posters were judged by an independent committee, Dr Nathan Skeen (Imperial), Professor Inga Prokopenko (Surrey) and Dr Margherita Malanchini (QMUL). The prizes were £100 Waterstones vouchers each. In their presentation, the committee noted the high quality and wide scope of the posters. We would like to congratulate the 3 poster winners, Daniel S Malawsky (Sanger), Dr Vignesh Kartik Chundru (Sanger) and Max Tomlinson (KCL) 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 using R code. We congratulate Dr Marta Futema (St George's), Dr Julia Zollner (St George's) and Dr Saskia Hagenaars (KCL) for their carer awards; we were delighted they were able to attend the meeting.

Finally, we would like to thank Sandra Howgate, our brilliant illustrator; our meeting ECR rep Dr Ryan Arathimos (KCL); PhD student Kai Lim (UCL) our network Communications Manager (UCL).

Example Feedback comments

"I would like to congratulate you on such a great annual meeting! I really enjoyed the variety and quality of talks."

" I think the idea of carer award is great! I feel more meetings/conferences should implement such initiative to support those with careering responsibilities."

"An excellent #LGN2021 meeting – thanks to @KKuchenbaecker and @Gelironald for their fantastic leadership, @kxlim for keeping everything running smoothly all day, and @RyanArathimos for ensuring ECRs were front and centre!"

"Thanks for the amazing opportunity offered by the London Genetics Network @LdnGeneNet

... Really interesting discussion and look forward to more research coming up in diverse ancestries"

"Thank you for organising such an amazing conference! All the talks were really high quality and super interesting!"

https://www.londongeneticsnetwork.com/annualmeeting/

Find us on Twitter @LdnGeneNet

UK Rice Research Consortium Annual Meeting

10–11th November 2021

Prof. Adam Price (University of Aberdeen)

The 4th Annual UK Rice Research Consortium (UKRRC) meeting was held on the 10th and 11th of November 2021. It was a hybrid (face to face and online via Zoom) meeting held at the Aberdeen Science Centre. The target attendees were academic group leaders and advanced researchers working on rice (UKRRC also holds an annual early career researchers event). It was entitled "Rice and Climate Change" and was timed to run concurrently with COP26. There were 26 live delegates and 30 joined online. The meeting was opened by a short address from Professor Karl Leydecker, Senior Vice-Principal of the University of Aberdeen. The event was split into two science sessions.

The first sessions was "Greenhouse Gas Emissions, Resource Use Efficiency and Climate Impacts in Rice" and was started by Professor Tapan Adhya of Kalinga Institute of Industrial Technology, India addressing the meeting online.

His talk "Sustainability of rice production system: in clash with the changing environment" gave a brilliant background to the importance of rice and the context of climate change both in terms of mitigation and adaptation. There were nine live presentations from the UKRRC community.

The second session was "Genetics and Genes for Adaptation to Climate Change" and was started by an online presentation from Dr Amelia Henry of the International Rice Research Institute who presented "The trait development pipeline for rice breeding". This highlighted the climate change relevant traits, the genetic sources of genes and the breeding pipelines being used to exploit them. That was followed by one online and three further live presentations from the UKRRC community. In addition to scientific presentations there were three discussion sessions on "Planning UKRRC's future", "Funding Opportunities" and "Collaboration: Mapping and Joint Planning".

Funding was received from the Rank Prize Fund, the University of Aberdeen and the Genetics Society. Of particular interest to the Genetics Society were presentations about genetic mapping heat stress in a MAGIC population (University College London), mapping in a Vietnamese rice diversity panel (Earlham Institute), and photosynthesis traits in *Oryza glaberrima* (Nottingham), plus talks on specific genes *AXR4* (Nottingham), *SUMO* (Durham) and nematode resistance genes (Aberdeen).

Live delegates were offered a £50 travel bursary if they used an alternative to flying for attendance and six people claimed that fund.

Overall, the meeting was a great success, allowing some quality face to face dialogues (for some the first time since COVID started) and interaction with online participants, even in the discussion sessions. Feedback was very good and only comments were some challenges with background noise in the live room being fed into the live video stream for online participants and a couple of comments about mediocre hotel experience.

Journey of a Molecular Detective

20th September 2021

Lidia Arciszewska and David Sherratt (University of Oxford)

110 participants were present in person at this 'hybrid Symposium', the first in-person event in the recently completed striking New Biochemistry building. Furthermore, the Symposium was streamed live to ~120 on-line registrants all over the globe. ~65 of the in- person participants were ex-students and -postdocs who Prof David Sherratt's trained throughout his career spanning 50 years, many travelling from the USA and Europe. The Symposium was divided into four sessions encompassing the stages of David Sherratt's career, Sussex, Glasgow and two for Oxford. The presentations reflected the path of Prof Sherratt's scientific interests beginning with plasmid biology, through site- specific recombination and transposition, and chromosome biology; bacterial chromosome segregation, replication and repair then to molecular machines.

The presentations, all with a strong genetics focus, were from an ex-student (Claudio Stern), ex-postgraduates (Gordan Dougan, Lorraine Symington, Colin Stirling, Stephen Bell, Rodrigo Reyes-Lamothe, Anjana Badrinarayanan), and ex-postdocs and collaborators (Jan Löwe, Dale Wigley, FX Barre, Christian Lesterlin, Neil Johnson, Steve Kowalczykowski).

Topics of the talks spanned developmental biology and genetics of twins (Claudio Stern); to bacterial pathogenicity (Dougan); complex systems biology (Johnson); and to diverse aspects of chromosome biology (other speakers).

There were stimulating discussions and the overall event demonstrated the feasibility and success of 'hybrid events'. Furthermore, postgraduates and early career researchers had the opportunities to meet the speakers and experience the career trajectories of an international group of scientists. The Symposium was followed by a reception in the Department and 72 participants (including students) then enjoyed an excellent dinner at Trinity College. Overall, the Symposium was an excellent occasion for celebrating David Sherratt's long career, with outstanding presentations. It provided the opportunity for ex-lab members to get to know each other, or to re-kindle past friendships. We are indebted to the Genetics Society for the one-off grant, which helped make this meeting a huge success.

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 Carla Canedo Ribeiro, Declan Morrissey, Fenella R Wood, Michael O'Leary, and Rebecca Whitla.

Introduction to Nanopore Sequencing and Data Analysis

20th-21st October 2021

Carla Canedo Ribeiro (University of Kent)

Farming of pigs (*Sus scrofa domesticus*) and cattle (*Bos taurus*) has a very significant impact on the economy, culture, and the environment at a global level. As the leading source of animal protein, these two species alone produce approximately 65% of the meat and over 95% of the milk consumed worldwide.

To maintain productivity to high enough levels to sustain the raising global demand for food without worsening the environmental impact of breeding, genomic selection strategies have been introduced. These make use of genotyping and/or sequencing methods to study the genotypes of specific animals (intended parents) and attempt to predict the phenotype of their offspring. However, these strategies typically fail to take into account the underlying fertility of the intended parents due to the complexity of this trait, but also due to their current technical limitations which renders them unable to detect many forms of chromosomal rearrangements (like reciprocal translocations) which are known to be important causes of sub-fertility in both humans and farm animal species.

Indeed, an infertile boar or bull carrying a chromosomal rearrangement, the semen of which is used for artificial insemination, can lead to a loss of up to £2 million to an enterprise due to the number of animals that will not be borne as a result. Even though the semen is commonly analysed for parameters such as volume, morphology, and motility, infertility linked to a chromosomal abnormality cannot be detected in this way either. Chromosomal analysis in this field is currently performed by employing fluorescence in situ hybridization (FISH), however, this is a laborious technique requiring additional effort for the breeder and whose limited resolution might lead to missing some chromosomal abnormalities.

To improve on this resolution and develop a methodology that allows for comprehensive chromosome abnormality screening and genomic selection at the same time, I aim to develop a new approach using long-read genome sequencing, particularly Oxford Nanopore (ONT), to assess the genotype and discover chromosomal rearrangements among breeding populations of both pigs and cattle.

With this in mind, the Nanopore online workshop would be the most suitable training for advancing this project. This was a two-day course delivered by Oxford Nanopore experts. The course aims to provide a comprehensive overview of Nanopore sequencing, including planning for experiments and hands-on examples of the application of this technology. The course is divided into two main sessions of 3.5 hours each in the mornings. The first session offers an introduction to Nanopore sequencing and covers in detail the wet-lab part of the technology (handling a flow cell and sequencing). The second session is focused on data analysis and offers an introduction to long-read data. It provides a workshop on command line fundamentals and includes guidance on how to correctly perform data analysis and quality assessment, demonstrates the dedicated software and workflows, and supports the learner by providing further bioinformatics resources and guides.

I am looking forward to applying the knowledge acquired in my project, as well as sharing it with the team as well collaborators. I am also very grateful for the financial support granted by the Genetic Society to help expand and improve my skills related to bioinformatics. I would like to thank all the workshop organisers and who provided it from the Oxford Nanopore Technologies team.

Ultraconserved Element library preparation, target enrichment and hybridisation of anthozoans

6–4th July 2021

Declan Morrissey (National University of Ireland Galway)

Figure 1: (L-R) Jessica Gordon, Dr Michelle Taylor, and Declan Morrissey

In June 2021, I visited Dr Michelle Taylor's lab at the University of Essex to learn how to construct libraries from octocoral DNA and how to do target enrichment and hybridisation of Ultraconserved Elements (UCEs). After a year of yo-yo lockdowns in the Republic of Ireland, travelling out of the country was a surreal experience.

In my luggage were 96 DNA extractions of deep-sea corals sourced from around the world over the past two years. Luckily, Ireland and England are both part of the Common Travel Area and even post Brexit there are no political hurdles to travelling. At the University of Essex, we had twice-weekly antigen testing for COVID19. This testing, compounded by social distancing and mandatory mask use inside, made the workplace feel very safe.

My day-to-day training was with Jessica Gordon, a PhD student using Single Nucleotide Polymorphisms derived from UCEs for population genetics of *Acanella arbuscula*, a common Atlantic deep-sea coral. She has prepared approximately 400 individuals using this method and has perfected it. She passed on all the tips and tricks she picked up and ensured that I knew how to avoid the common pitfalls. One of the major skills I learned throughout this training period was how to upscale processes into a 96-well plate. I am used to working with a lower volume of samples so the experience of using the multichannel pipettes was invaluable. Furthermore, I gained experience planning an indexing system for my libraries and how to clean and size select DNA with magnetic beads. This training period has given me both the skills and confidence to start preparing future libraries in-house. During our downtime, it was great to hang out with other octocoral researchers. Jess and I streamed a "specimen tour" on her twitch account (DeepSeaJess) where we talked about all the different corals we found in Dr Taylor's lab. After a year of isolation, it was nice to meet new people again (Figure 1).

The libraries generated during this training period are currently being sequenced and the data generated will be used for a chapter of my PhD and, in time, multiple journal articles. My research focuses on the biodiversity and evolution of the bamboo corals (Class Anthozoa: Family Isididae: Subfamily Keratoisidinae, Figure 2). This exclusively deep-sea group is currently undergoing a major redescription due to its complicated taxonomic history which used unreliable morphological characters.

This has led to polyphyly at the species and genus levels. Furthermore, octocorals gather mutations in the mitogenome at a much slower rate compared to other animals due to

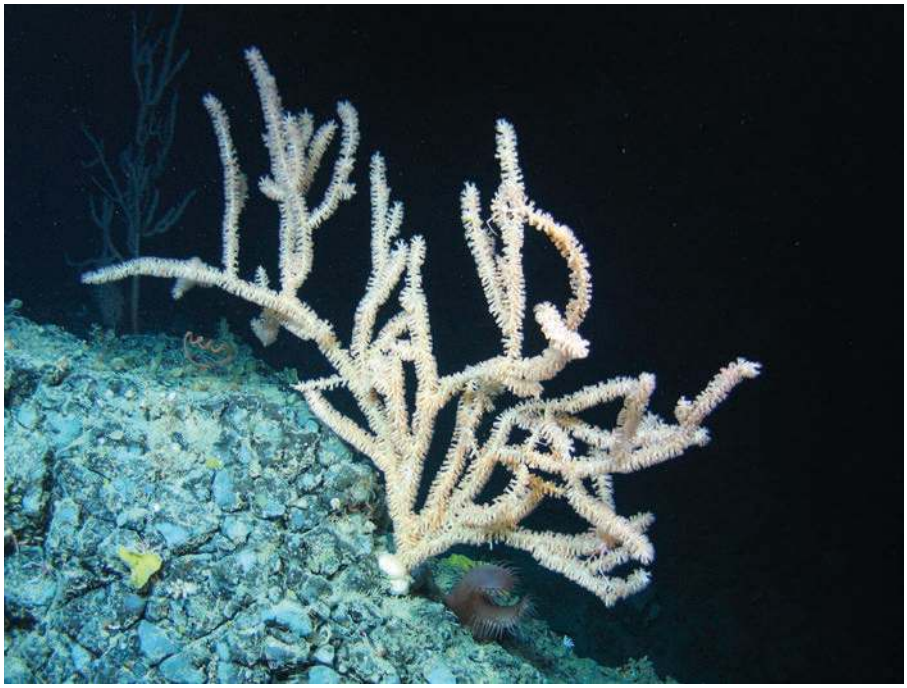

Figure 2: A bamboo coral of the genus Keratoisis from the North Porcupine Bank at 1301 m.

the active mismatch repair gene *MutS*. This has limited the use of DNA barcoding for discriminating between species and reliably inferring evolutionary relationships. Using UCEs, I will target thousands of loci across the nuclear genome. The large amount of genetic information gathered from this method will allow me to genetically delimit species and then we can start to look for corresponding morphological homologs that can better delimit and be used to describe new species and genera in the subfamily.

As UCEs can be used to answer evolutionary questions from population structure to paleo phylogenomics, the method has and will continue to revolutionise the field of anthozoan evolution. Therefore, by learning how to successfully carry out this protocol, I have widened the scope of the research my lab and I are capable of pursuing. I am very grateful to the Genetics Society UK for funding this training period and to Dr Taylor and Jessica Gordon for all their help and support!

Declan Morrissey is funded by the Irish Research Council Postgraduate Scholarship GOIPG/2019/3682. The consumables and reagents used during this training period were funded by an NUI Galway Thomas Crawford Hayes Fund award and the Irish Research Council. The bamboo coral image was taken during research cruise CE18012 aboard the RV Celtic Explorer with the ROV Holland I. Many octocorals used in this training period were gathered on ship time funded by Science Foundation Ireland (SFI) and the Marine Institute under Investigators Programme Grant Number SFI/15/1A/3100, co-funded under the European Regional Development Fund 2014 – 2020 awarded to Professor Louise Allcock (National University of Ireland Galway).

Physalia Courses - Adaptation Genomics

6–4th July 2021

Fenella R Wood
(University of Aberdeen)

For my PhD research, I aim to increase our understanding of spurdog (*Squalus acanthias*), a small species of shark, in the Northeast Atlantic using genomic and stable isotope approaches, particularly in relation to their aggregation behaviour and population structure. The aggregating nature of this species makes them vulnerable to capture by commercial fisheries, and although they can no longer be targeted, they are still caught regularly as by-catch contributing to their Endangered status. So far, population structure of spurdog in the Northeast Atlantic have only been investigated with a small set of microsatellites which did not reveal a clear structure. Using SNPs derived from Restriction Site Associated DNA sequencing (RADseq) will allow me to not only get a better representation of genetic connectivity but will further enable analysis of adaptive loci. This might reveal adaption patterns of interest in spatial conservation management as well as be an important consideration when estimating the impact of large bycatch events on the gene pool.

To perform these analyses, RADseq will be used to provide tens of thousands of polymorphic loci across the whole genome.

The Genetics Society provided funding for me to attend a course entitled 'Adaptation Genomics' provided by Physalia courses. Usually held in Berlin, Germany, the course was delivered online using a combination of zoom, slack and github. Course leaders were Dr Claire Mérot (Université Laval, Canada), Dr Anna Tigano (University of New

Hampshire, USA), and Dr Yann Dorant (Université Laval, Canada). This course covered the topic in great detail, from an introduction to bioinformatics through to structural variants. The course was delivered over five days, with each day's topic advancing in complexity. Here is a breakdown of some of the topic covered.

DAY 1
Next Generation Sequencing (NGS) data handling.

DAY 2
Population structure and confounding factors.

DAY 3
Outlier detection and environmental associations.

DAY 4
Accounting for structural variants.

DAY 5
Functional approaches.

This course came highly recommended by fellow colleagues within our field of conservation and population genetics. A reputation that obviously had a far reach throughout Europe, where most of the attendees were based, but also attendees from Mexico, Chile, USA, Hong Kong, and Australia. It was a great opportunity to hear about the other research projects occurring globally that will also be applying these analytical methods. The support offered by the course tutors was invaluable, especially with the variety of background knowledge the attendees had. I benefitted from further practice using the Unix command line and working through examples of how to perform the analyses in preparation to receive my own RADseq data.

I look forward to applying these analytical approaches to my own research. Attending 'Adaptation Genomics' has been invaluable to the progression of my own PhD research. As someone new to the field of population genomics and to many of the bioinformatic analysis approaches, this course provided by Physalia has been an asset to my development and progression within my own PhD. Of which I wouldn't have been able to benefit from without the support of the Genetics Society Training Grant. I would like to thank the Genetics Society for their help in supporting my training and development as a researcher within the field of conservation genetics.

Experimental Evolution 7th edition: bringing theory and practice together

2–5th November 2021

Michael O'Leary (University College London)

Experimental evolution is the practice of attempting to recreate wild evolution in a laboratory setting. It is the use of laboratory experiments or controlled field manipulations to explore and identify evolutionary dynamics. Often it involves model organisms adapting to new environmental conditions and exploring the evolutionary dynamics that facilitated this change. My PhD topic centres heavily upon these principles so being a junior researcher, I was keen to explore these ideas further and to widen the scope of my knowledge of the subject area.

It was suggested to me that I attend the Experimental Evolution training module, which after a one-year hiatus due to COVID-19 was due to be held for its 7th annual edition at the *Ecole Normale Supérieure (ENS)* in Paris, France in association with the Vienna Graduate School of Population Genetics. The course ran from 2nd – 5th of November 2021.

The four-day training course was run by a consortium of world-leading researchers in their respective fields such as Luis-Miguel Chevin (CEFE, Montpellier), François Mallard (ENS Paris), Christian Schlötterer (VetMed University, Vienna), Maud Tenaillon (DYGAP, Moulon), Olivier Tenaillon (IAME,

Paris), and Henrique Teotónio (ENS Paris). The course was attended by 25 students. 14 of these first- and second-year masters students from the

ENS and the remaining 11 were PhD students like myself, from an array of different institutions from countries such as Germany, Holland, Austria, Switzerland and the United Kingdom.

The itinerary of the course consisted mainly of lectures encompassing many different aspects of experimental evolution. The course was aimed at introducing us to the experimental approaches employed by researchers, to test hypotheses about natural selection and genetic drift, and to enable us to estimate parameters about standing genetic variation within populations. During the course we discussed the use of several model organisms to address specific questions, ranging from asexual microbes to sexual metazoans, about domestication, adaptation to novel environments and extinction.

During the course there were a varied range of compelling talks, predominantly focussing on population genetic processes and phenotypic evolution, with topics such as maintenance of genetic variation, developmental evolution, host-pathogen coevolution, phenotypic plasticity and transgenerational effects, evolution of sex and recombination explored, among others. Of notable interest to myself were François Mallards talk on "Predicting multivariate phenotypic evolution" and a truly enthralling lecture by Henrique Teotónio regarding the history of the field of Experimental evolution, which as a junior scientist relatively new to the field, I found extremely engaging.

Of particular interest to myself was the talk given by L.M. Chevin entitled "Adaptation and extinction risk in changing environments". Within this talk Dr Chevin detailed many aspects of species adaptation and responses to rapid environmental change, he also devoted significant time within his talk to touch on the evolutionary theory of "*Bet-Hedging*" the phenomenon that I am conducting my thesis work upon. I found this talk extremely conceptually significant and it really gave me a much better understanding of my topic area and the current research that is currently underway in the field of species adaptation to change.

I am extremely grateful for the training grant I received from the Genetics society which contributed to the costs of my travel, accommodation and living costs. Without their generosity. I would not have been able to attend such a fascinating and career building event.

Conservation Genomics training course by Physalia-courses

27–30th September 2021

Rebecca Whitla (Oxford Brookes University)

The Conservation Genomics course from Physalia-courses ran on Zoom, and over four days attendees learned how to perform different population genomics analyses and how these can be used to inform conservation practises. The course was run by Dr Evelyn Jensen (Newcastle University, U.K.), Dr Catherine Cullingham (Carleton University, Can.), with Rachel Grey (Newcastle University, U.K.) and Dr Carlo Pecoraro (co-founder of Physalia-courses). Each day was a mix of lectures and practical sessions, with practical exercises running in a Linux environment and R.

After quick introductions from the course leaders and attendees, we started with a general introduction to conservation genomics study design, data collection techniques, and file types we may encounter during analysis. For the practical session we checked the quality of our sequences using FastQC and trimmed adaptor sequences, aligned our sequences to a reference genome, filtered our reads and detected SNP variants. All practicals were performed with subsets of polar bear genomic data collected from a previous study, and so we could discuss the use of each analysis and real-world implications of the results. The hands-on practical sessions were typically done in small groups (within

We looked at estimating effective population sizes using linkage, heterozygosity and coancestry calculation methods on polar bear populations in Canada. We also discussed strengths and weaknesses of each method, and the effect population structure will have on these analyses. All of these methods were performed using NeEstimator v2.

Zoom 'break-out rooms'), where attendees could network, discuss tasks and troubleshoot issues they might be having. This was helpful as people with more experience with the Linux command line and R could help out others.

Day two included tutorials and practical exercises on genotype and SNP filtering using the Linux Command Line, and population structure analyses, such as PCA and DAPC analyses in R and demonstrations of STRUCTURE. There were discussions of strengths and weaknesses of programmes used and of alternatives that are available.

On day three we focused on identifying adaptive loci using environmental correlation and outlier detection methods. To identify how the polar bear loci are associated with environmental variables we performed an RDA.

Outlier analysis was also performed using R packages OutFlank and PCAdapt, and results from all three methods were compared.

We also looked at estimating effective population sizes using linkage, heterozygosity and coancestry calculation methods on polar bear populations in Canada. We also discussed strengths and weaknesses of each method, and the effect population structure will have on these analyses. All of these methods were performed using NeEstimator v2.

Day four focused on diversity and relatedness analyses. Our first lecture was on genetic diversity within populations and the different techniques that can be used to calculate this. For the hands-on session we estimated individual heterozygosity using plink1.9 within the Linux command line, and examined the data in R using the hierfstat package. Observed heterozygosity, expected heterozygosity and the inbreeding coefficient were also identified for different populations of polar bears.

We also had a lecture on relatedness, and a practical investigating relatedness among a sample of 400 polar bears. We also determined which estimator of relatedness was most appropriate for the data we have. We then performed a simulation to determine how well the SNP data we have can distinguish between relationship categories (eg. parent-offspring, sibling, etc) and therefore how useful the data is. This was performed in R using the relatedness package.

The course ended with a discussion on tips and tricks for installing software and troubleshooting, and a chat about everyone's research goals. For my PhD project I will be using whole genome sequencing to identify genomic signatures of range expansion and contraction in UK Butterfly Species. I will study three species: museum specimens of the Black Veined White, Aporia crataegi, which has undergone range contraction and subsequent extinction in the UK; fresh specimens of the Wood White, Leptidea sinapis, a species that is currently undergoing range contraction; and fresh samples of the Orange Tip, Anthocharis cardamines, a species currently experiencing range expansion. This course has helped develop my knowledge on available bioinformatics tools, and has helped me consider which analyses to use in the future.

The leaders for this course were knowledgeable and helpful, and the course itself was very informative as it taught population genetic theory, how to execute analyses bioinformatically, and the rationale for choosing specific packages and analyses. I would recommend this course for anyone considering using these types of analyses.

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 around Scotland: Combinatorial view on eyebright speciation?

Yanqian Ding (The University of Edinburgh)

The mysterious story behind rapid speciation always triggers evolutionists' interests. Many speciation genomics studies revealed an important role of old genetic variants in facilitating rapid speciation and adaptive radiation. Old variants, often derived from hybridisation, providing a good exiting fuel for rapid speciation. This process refers to 'combinatorial speciation', is a newly summarized genetic mechanism of reassembly old variants leading to new species.

British eyebright (*Euphrasia*) contains recent rapid speciation across the UK, which has massive hybridisation among different species. Besides, it's a semi-parasite plant, which means host plants will contribute to its variation. The previous whole-genome studies showed low species-level differentiation with only a few genomic outlier regions. This provides a good plant model system to study combinatorial speciation. Thus, in my project, I chose two species with different mating system, aiming to disentangle the mystery from phylogeographic history, mating system and local adaptation.

Supported by the Heredity Fieldwork Grant, I was able to collect the samples across all over Scotland, which providing sufficient data for future analyses. With the help of my colleagues, we collected in total 500 fresh plant samples (dried in silica) from 22 sites across all the Scotland, including 3 sites from Outer Hebrides. It includes two species: *Euphrasia micrantha* and *E. arctica* and the hybrid between the two: *E. arctica* x *E. micrantha*. *E. micrantha* is highly selfing while *E. arctica* is a mixed mating species, it's variable but contains larger flowers, indicating high proportions of outcrossing. (Sampling map details: www.google.com/maps/d/edit?mid=1jmSGmftBqJaenWiEJSN9lPlNzjnJbioP&usp=sharing)

As the two of the most widely distributed species in Scotland, *E. arctica* and *E. micrantha* have lots of sympatric distributions, however, during the field trip, I noticed how different the local habitat varies from species to species, or some places even varies from population to population. For example, *E. micrantha* is more commonly co-occur with common heather *(Calluna vulgaris)*, the latter is distributed in more acid soil, in heather land. While *E. arctica* prefers grassland. Thus, I also collected 36 soil samples from above sites in the field, especially those area distributed both *E. arctica* and *E. micrantha*. This will be tested for soil types, pH and nutrients. Seeds were also been collected in the late September for future common garden experiment, which allows us to test local adaptation from different soils and host plants.

As a semi-parasite plant, different host plants also provide them opportunities to keep species difference within sympatric distribution. For example, the following photo shows how stunning the two species distributed as neighbors but with significant different traits.

Currently I am doing DNA extraction from all the samples. Whole genome resequencing will be applied for future population genetics and phylogeograhic studies. The patterns and mechanisms of population genetic structure will be assessed through a diverse array of methods, including Bayesian clustering, approximate Bayesian computation to simulate possible demographic models. In particular, we hope to explore through the spatial scale of genetic structure, the colonization history and the relationship among mating system, genetic diversity and population genetic structure to identify the divergence associated with variants. We expect the results of this project to contribute to the understanding of combinatorial view of speciation.

Bioinformatics in Schools

Stevie Anne Bain (University of Edinburgh)

Our Bioinformatics in Schools project designs and delivers curriculum-linked, hands-on bioinformatics workshops for secondary school biology classes. Bioinformatics – a discipline combining aspects of biology, computing, mathematics and statistics – is now an essential element of modern biology. We use case studies based on DNA sequences to highlight its importance in topics such as genetics and evolutionary biology. Additionally, our senior level workshop uses Raspberry Pi computers to introduce pupils and teachers to coding.

Over the last 18 months, the COVID-19 pandemic has meant that we cannot conduct our school visits as we normally would. However, we consider public engagement with genetics to be crucial in the fight against misinformation and inequality. Therefore, we used our Genetics Society Public Engagement Award to develop an online bioinformatics workshop for teachers. This provided an opportunity for teachers to engage with researchers and get hands-on experience of bioinformatics. Our workshops took place after the school day to avoid any disruption to school timetables. To support this interactive online workshop, we sent each participant a Bioinformatics in Schools Resource Package containing handouts for all our activities, a bioinformatics textbook and a complete Raspberry Pi kit preloaded with our workshop materials (Fig 1). This allowed teachers to fully participate in all activities during the workshop and then use our educational materials with their own classes.

"In addition to presenting our own resources, we have also invited researchers with bioinformatics expertise to present case studies of their research"

We piloted this teacher workshop at the Midlothian Science Festival in October 2020 and have since held events for teachers across Scotland. So far, we have reached 57 teachers and 54 schools. A benefit of our online format is increased ease of participation, particularly for teachers in remote locations such as the Highlands and Islands. In addition to presenting our own resources, we have also invited researchers with bioinformatics expertise to present case studies of their research. One such case study involved the use of sequencing to investigate SARS-CoV2 genome evolution. In these workshops, teachers have the opportunity to ask questions and share their experiences with their peers and with researchers. This, in turn, allows us to further develop our resources to maximise their benefits for schools. Positive teacher feedback showed that these workshops were a great success:

"I thought it was very exciting because we were able to investigate something."

"Fantastic, relevant, interesting, fun and pitched at the perfect level."

This online workshop and accompanying resource package have allowed us to reach a large number of teachers across the country during difficult times. We are excited to continue with our program of online public engagement and thank the Genetics Society for their vital contribution.

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.

Eukaryotic mRNA processing

Bertille Montibus (King's College London)

Cold Spring Harbor Laboratory is a world leading centre for research and education which organises every year conferences on different topics regrouping the world leaders as well as junior researchers. For the 13th time (first time virtually), they organised this year the "Eukaryotic mRNA processing" conference. I selected this conference as my post-doctoral research is centred on the regulation of 3'end processing of mRNA molecules through a process called alternative polyadenylation. Thanks to the genetics society conference grant I was able to attend this conference.

It was really stimulating to listen about the latest developments in the field, most of them corresponding to unpublished work. This conference also gave me the opportunity to learn optimisation and usage of cutting-edge technologies to study the different aspects of transcription and mRNA processing.

I particularly enjoyed the sessions on alternative splicing and 3'end processing. I was especially inspired by a talk given by Anna Fiszbein, a young group leader at Boston University, studying the regulation of mammalian expression programs. She presented a project where her group have developed bioinformatics pipelines to extract new information from publicly available RNA-sequencing datasets. They also showed that some of the regions of genes annotated as exons can alternatively be used as an internal exon or the first exon of alternative transcripts of the same gene. This phenomenon can contribute to transcript diversity from each gene.

Her project elegantly showed that it is still possible to identify new mechanisms of gene regulation by using resources that are already available in the public domain. My project is also aiming to use datasets from the public domain to identify new aspects of gene regulation, and following this talk, I am planning to apply similar approaches for some of the questions we are trying to answer in the group.

I was also lucky to present and discuss about my work during the poster sessions (1 hour every day). The posters were displayed in an online gallery, giving enough time for people to look at the posters. In parallel, there was a dedicated slack channel for interactions about projects. I had a few questions on my poster and some suggestions that are directly beneficial for the future directions of the work.

I would have preferred to attend the conference in person, but I have found that as an early career researcher, it was easier to discuss with the speakers/poster presenters online. Indeed, having a dedicated slack channel made it easier to post a message/directly ask a question to the speakers, without having to worry about the time constraints of the conference.

Finally, I would like to thank the Genetics society for its support and for the great opportunity to attend this conference which will impact the future direction of my project.

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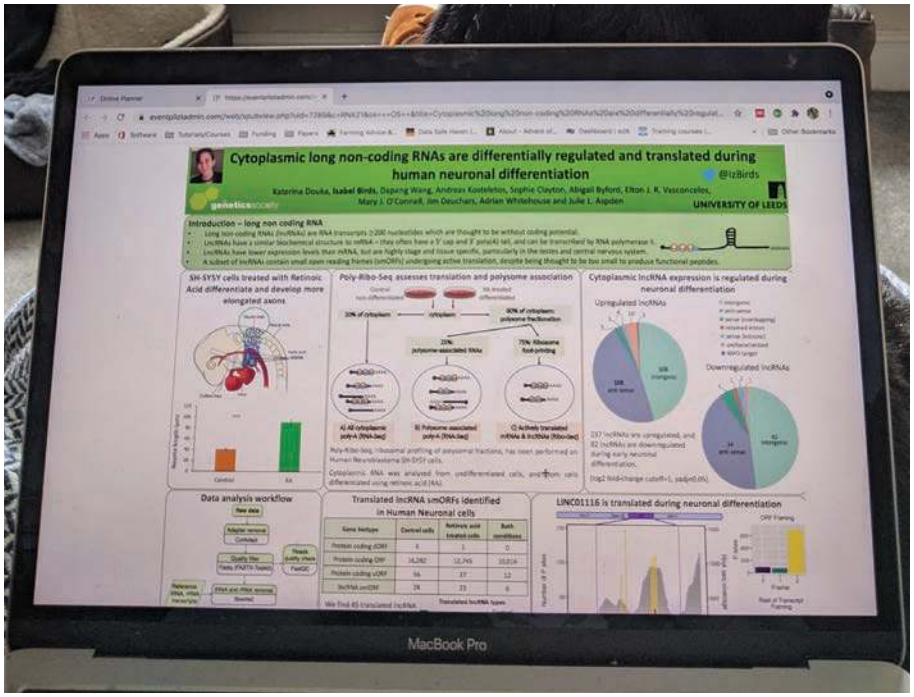
The annual meeting of the RNA Society, 2021

Isabel Birds (University of Leeds)



The annual meeting of the RNA Society is one of the largest conferences in RNA biology, and for the second year running it took place online. Last year the organisers only had a couple of months to pivot from in person to online, so it was interesting to see how the conference changed with more time to plan.

One large change was running the conference in half days over two weeks, to cater for as many time zones as possible. I particularly appreciated the recording of the sessions – this meant I could enjoy the sunny bank holiday weekend without missing out on any talks! Being able to go back and note down missed references or details is also massively helpful – I hope that the practice of sharing presentations will continue as when we return to in person meetings.

I really appreciated the number of ECR centred events organised by The RNA Society Jr Scientist Committee, including a biotechnology careers panel, social events and a networking and job jamboree. The committee even created a new podcast to mark the occasion – the Ribozone! All of these chances to network and get input from people pursuing different careers are more impactful than ever as I approach the end of my PhD.

The conference was a also great opportunity to share our preprint (now accepted into the RNA journal) with the wider RNA community. As a computational biologist I don't spend much time talking about wet lab work, so pulling out the important aspects to discuss on my poster was a really useful experience.

I was quite nervous about the online judging of posters, but found it to be a better experience than at in person conferences. Having a set time meant there were no worries of having missed the judge, and it was more productive to be able to have a conversation in a quiet environment. The only downside of the online platform was that it was difficult to visit other posters while keeping an eye on my own during the live sessions.

Unsurprisingly the bioinformatics session was my favourite. I also really appreciated the chance to hear about a broader range of subjects, not just the specialised areas our lab works in. The keynotes from Irene Bozzoni and Nobel prize winner Jennifer Doudna were particular highlights.

Thanks to the Genetics Society for enabling me to take part!

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 Alice Lomas, Ashley Wong, Barney Hill, Cameron Dhruv Bhimsingh, Chloe Tucker, Danilo Negro, and Drew Farr.

Investigating the effect of CHD4 point mutations on the interaction with NuRD and ChAHP complex proteins

Alice Lomas (University of Bristol)

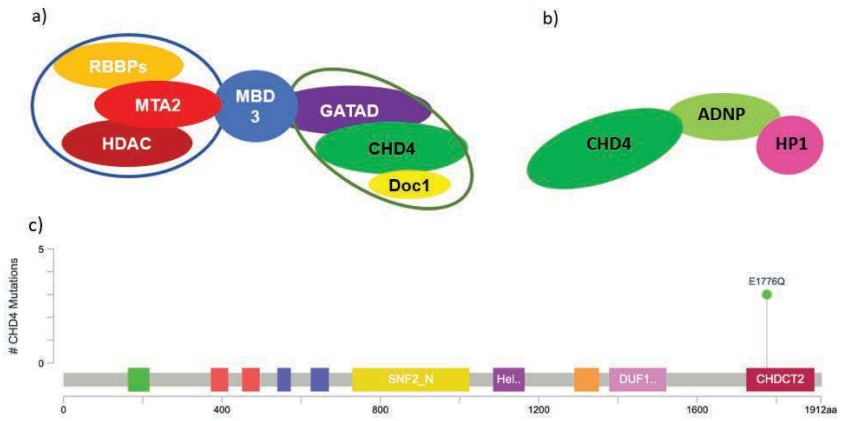


Figure 1 *a) NuRD complex with CHD4 in green. Interaction with MTA2 examined via immunoprecipitation b) ChAHP complex, interaction with ADNP examined via immunoprecipitation. c) Schematic representation of domain structure of CHD4 with the location of the mutations investigated labelled, figure produced from cBioPortal (3, 5, 6)*

Introduction

CHD4 (chromodomain helicase DNA binding protein) is a chromatin remodeling protein responsible for histone sliding (1). CHD4 forms part of the NuRD and ChAHP complexes (Figure 1). The Nucleosome Remodeling and deacetylation (NuRD) complex is important in the regulation of transcription(2). NuRD is also vital in determining stem cell fate. CHD4 is also found in the ChAHP complex where it interacts with ADNP. The ChAHP complex is involved in regulation of chromatin configuration (3).

The specific mutations investigated were E1776V and E1776Q mutations. These mutations have been identified in patients with CHD4-associated neurodevelopmental disorders. In addition, these specific point mutations have also been found in some bladder cancers(4). E1776V and E1776Q mutations are found in a conserved domain that is known to be necessary for NuRD association(5).

In this study the effect of the mutants on CHD4 interaction with NuRD and ChAHP complex proteins was investigated.

Methods

Flag-tagged CHD4 was mutated using PCR to produce CHD4 with E1776V and E1776Q mutations respectively. The mutated CHD4 fragments were then inserted into PiggyBac plasmids and cloned into *E.Coli*. Once purified the plasmids were sequenced to ensure the correct mutations had been introduced. The mutant CHD4 plasmids were transfected into BC8 cells, a mouse embryonic stem cell line. These cells express WT CHD4, however this is not flag-tagged. The cells were grown in selection media containing inhibitors MEKi and GSK3i, ensuring the stem cells remained in an undifferentiated state.

The cells were grown up and harvested to make nuclear extract used for IP-western blots. For the initial western blot nuclear extract was made from cells both treated with and without doxycycline. The PiggyBac plasmid used in transfection is dox inducible, so addition of doxycycline should induce expression of transfected CHD4. An IP Western blot to pull down for Flag

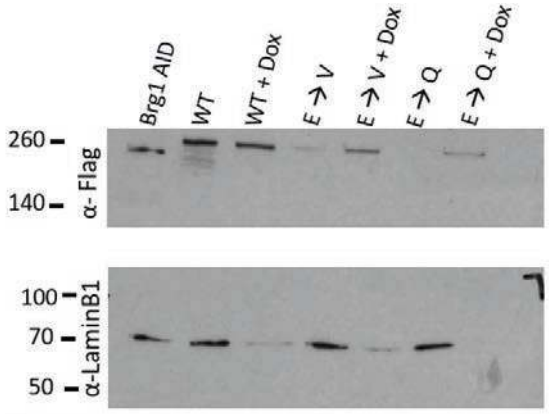


Figure 2 Immunoprecipitation for Flag and LaminB1 for nuclear extract produced from cells with and without the addition of doxycycline. Brg1 AID cell line used as a positive control for Flag.

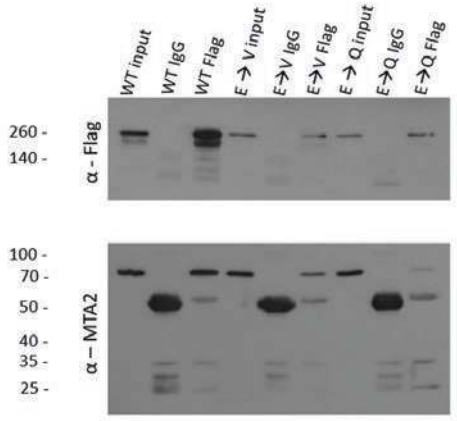


Figure 3 Immunoprecipitation using IgG, pulling down for Flag and MTA2.

and MTA2 (figure 1a) to examine the effect of mutants on interaction with the NuRD complex was completed. Interaction with the ChAHP complex was investigated through a further IP Western pulling down for ADNP (figure 1b). IgG coated magnetic beads were used in the IP Western blot and a control of each sample incubated with IgG was loaded.

Results and discussion

Figure 2 shows WT and mutant flag-tagged CHD4 is successfully expressed in BC8 cells. Dox induction is clear in the E1776Q mutation. However, in WT and E1776V expressing cells the dox system appears leaky. This did not present an issue in further experiments since they did not rely on depletion on CHD4 expression. The α-LaminB1 loading control differs between the samples so these results may not be an accurate reflection of CHD4 levels.

Figure 3 shows that the E1776Q mutation causes a dramatic drop in interaction with MTA2 protein. There may be some effect seen in E1776V mutant, however from this single Western blot it is difficult to tell. Reprobing with MBD3 was subsequently attempted to try to elucidate which protein the mutation affects interaction with. However, the IgG bands disrupted the MBD3 bands. Future experiments probing more NuRD complex proteins would be interesting.

The IP-Western for ADNP (figure 3) shows that for both the E1776V and E1776Q mutants there is a large decrease in interaction with ADNP. Interestingly the E1776V mutation affects interaction with the ChAHP complex but not the NuRD complex.

These mutations help to consolidate that the domain which contains the mutations is involved in NuRD interaction. In addition, it suggests this domain also functions in ChAHP interaction. Furthermore, these mutations will provide a useful genetic tool to dissect CHD4 function in future experiments.

Conclusion

The E to Q mutation appears to affect the interaction of CHD4 with MTA2, suggesting there is some effect on the ability of the NuRD complex to be formed with this mutation. There may be some effect seen in the E to V mutation, but further investigation would be necessary.

Both the E1776V and E1776Q mutations affect the interaction of CHD4 with ADNP and therefore are likely to affect function of the ChAHP complex. This also helps to elucidate that this domain is involved in ChAHP binding as well as NuRD binding. The effect of the mutations on interaction with ADNP is especially interesting as ADNP syndrome shares similarities with the symptoms associated with these CHD4 mutants (8,9) This may go some way to give reason for the neurodevelopmental disorder seen in patients with E1776V and E1776Q mutations.

Acknowledgements

Thank you to the whole Henrich lab who made this experience so enriching and enjoyable!

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The Structure and Composition 5' untranslated regions (5'UTRs) in Neurodegenerative Disorders (NDs)

Ashley Wong (University of Oxford)

Introduction

Neurodegenerative disorders (NDs) are a group of disorders characterised by the progressive loss of structure and function of neurons[1]. Since the pathogenesis of most NDs features the aggregation of misfolded proteins[2], patterns in the regulation of implicated genes may guide the identification of biomarkers and suitable treatments.

The 5' untranslated regions (5'UTRs) of mature mRNA transcripts are known to play a role in the regulation of gene expression, particularly by modulating translation[3]. For example, upstream start codons (uAUGs) can form upstream open reading frames (ORFs), overlapping ORFs or N-terminal extensions, all of which may attenuate the rate of translation of the coding sequence (CDS). Another layer of complexity is added by the dependence of initiation at particular uAUGs on the local sequence context. This means that leaky scanning (where a scanning ribosome passes a uAUG without initiating translation), re-initiation, and internal ribosome entry sites can all influence translation initiation at the uAUG and thereby that at the CDS[4].

The 5'UTRs of ND genes are therefore expected to be a good control set, making them a useful point of comparison for other diseases: since the majority are age-related, they should not possess any features that immediately set them apart.

Methods

Owing to the lack of an existing database of ND genes, a list was curated by combining PanelApp[5] Neurodegenerative disorders – adult onset (v.2.31) Green genes with those indicated as pathogenic or likely pathogenic at least six times on ClinVar (downloaded 29.06.2021)[6].

The list of ND names used to query ClinVar was produced manually using MeSH synonyms. Here, a combination of gene lists was used because PanelApp omits all non-adult onset NDs. This resulted in a list of 329 ND genes and 18,002 non-ND genes.

Then, R was used to extract information about the following features from MANE Select sequence data (v.0.95)6: 5'UTR length, GC proportion, codon bias, number of introns, intron length, (CGG)4 repeats, number of uAUGs, Kozak strength, distance between the end of the uORF from the CDS, uORF length.

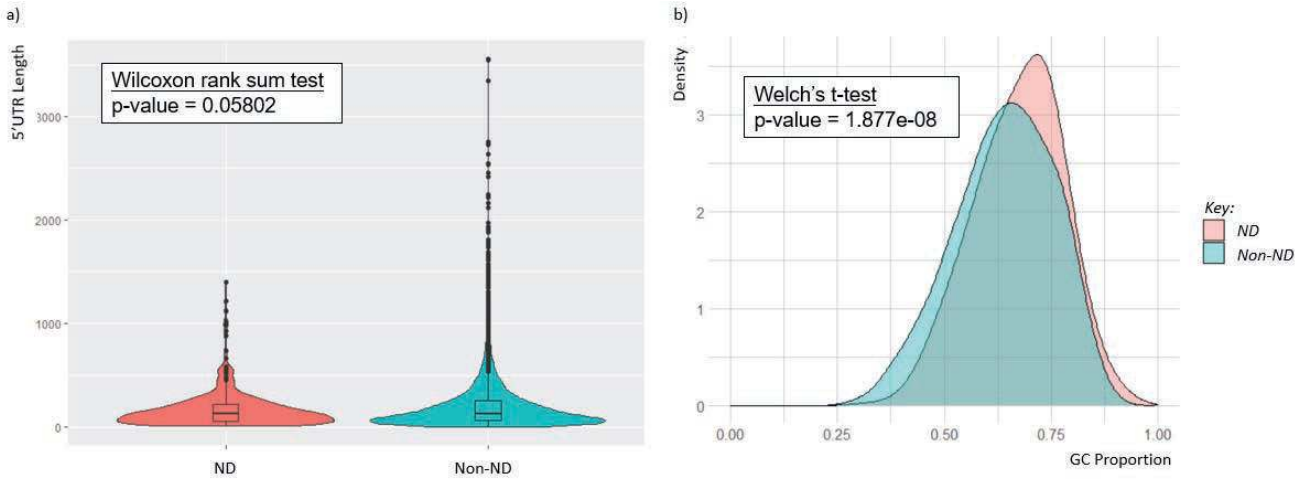

Figure 1: *(a) Violin plot with boxplot overlaid of 5'UTR length in ND and non-ND gene transcripts. The difference between the medians was short of statistical significance. (b) Density plot of the GC proportion in the transcripts of ND and non-ND gene transcripts. Welch's t-test found that the two means were significantly different.*

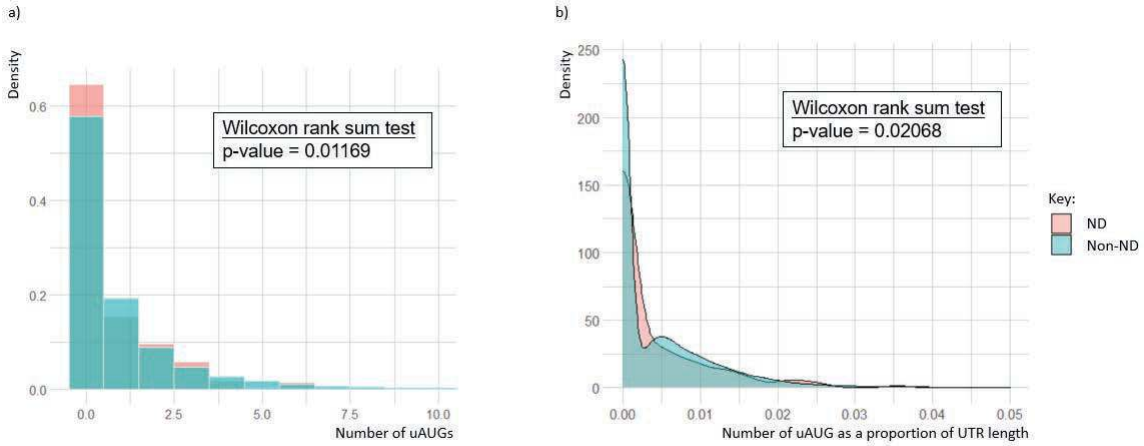

Figure 2: *The number of uAUG (a) prior to correcting for UTR length; (b) as a proportion of UTR length.*

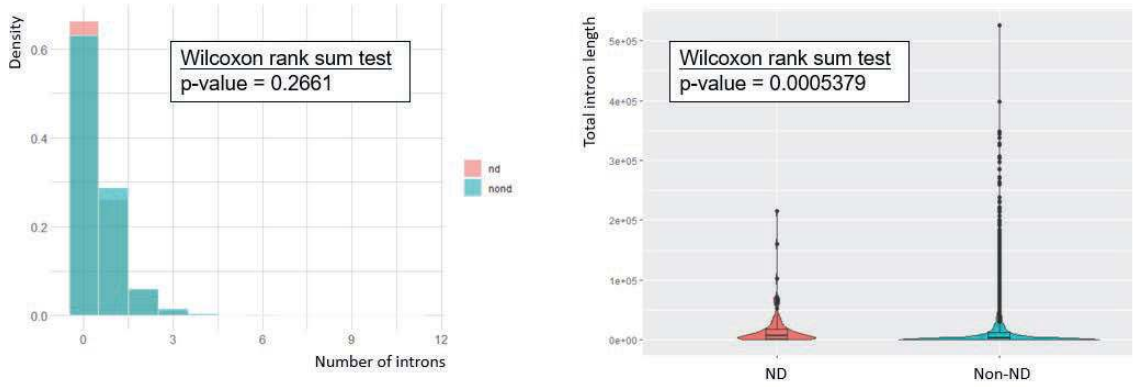

Figure 3: *(a) Bar graph of umber of introns. (b) Violin plot with overlaid boxplot of total intron length of ND vs. non-ND genes.*

Results and Discussion

The proportions of transcripts without 5'UTRs was not significantly different between ND and non-ND gene transcripts. In both cases, the proportions were very low (0.0092 and 0.014, p = 0.63 from 2p test), indicating the importance of 5'UTRs, perhaps owing to their role in gene regulation.

The difference in median length of 5'UTRs for ND compared to non-ND gene transcripts (Figure 1a) was just short of statistical significance (129 and 136, respectively, with p = 0.058 as calculated by a Wilcoxon rank sum test). Yet, the mean GC content as a proportion of UTR length is higher for ND genes (Figure 1b). A higher GC content is associated with increased formation of secondary structures, which usually repress translation. This suggests that the translation of ND gene mRNA may be more tightly regulated, though secondary structure data is needed to validate this.

ND 5'UTRs have more upstream start codons than non-ND ones. This remains true after correcting for UTR length, as shown by the density plot in Figure 2. uAUGs can lead to translation initiation before the start of the coding sequence, decreasing overall protein expression.

As is evident in Figure 3(a), the number of introns decreased exponentially for both groups of genes. There are ongoing debates about the functional consequences of introns. However, human housekeeping genes tend to have fewer and shorter introns relative to tissue-specific genes whereas tissue-specific genes tend to have more numerous and longer introns[7]. ND genes have higher median total intron length (Figure 3(b)), suggesting that they may generally be more tissue-specific, as would be expected.

The rest of the features that were probed as part of the project were not significantly different between the two groups. It would be interesting to see whether other adult-onset disease sets share similar characteristics; any differences between putative disease sets would be interesting to explore. More data on secondary structure would be very valuable for understanding mechanisms of translation regulation. Further, while an attempt to predict translated uORFs was made by looking for canonical start and stop codons in the transcript sequences, it has been demonstrated that translation may be initiated at non-canonical start codons (e.g. CUG)[8], so more experimental data on translated ORFs would be very informative on how 5'UTRs modulate translation.

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I am immensely grateful for Dr Nicky Whiffin and Nechama Tatz-Wieder for teaching me so much through their incredible supervision and mentorship, and the Genetics Society for funding this studentship.

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Identification of cell-types associated with latent factors inferred from phenome-wide GWAS summary statistics

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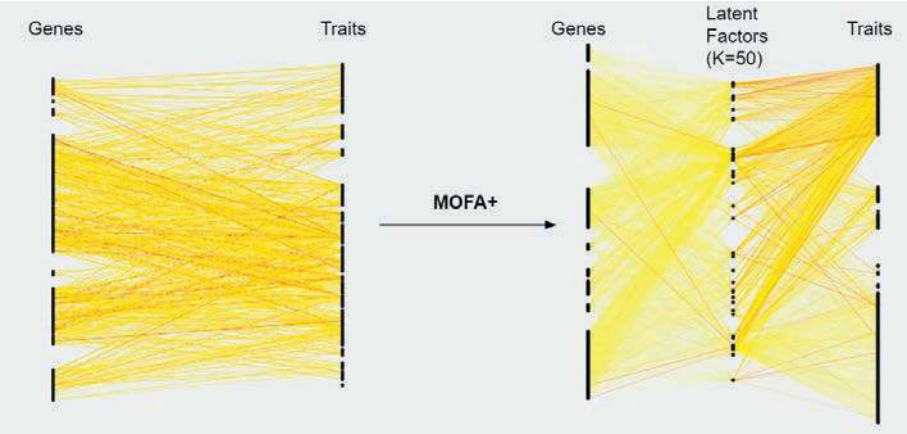
Introduction

In the last two decades genome wide association studies have accelerated the discovery of significant loci-trait relationships at an unprecedented rate [1]. Despite this there have been challenges translating these discoveries into clinical advances due to the polygenicity and pleiotropy of complex traits, in which many loci affect a single trait and many traits are affected by a single loci [2]. One explanation for this is that some traits may have many contributing genetic factors, some of which may be shared across multiple different traits.

Recently there have been efforts such as GWAS ATLAS [3], to collect and standardise GWAS summary statistics in order to develop understanding of the shared genetic structure of these complex traits. We investigated the application of MOFA+ [4], a dimensionality reduction algorithm, in order to infer 50 latent factors over more than 1700 GWAS ATLAS traits. Once we had inferred these genetic components we sought to characterise them through scRNA-sequencing, generating associations between cell-types and components.

Methods

GWAS were gathered from ATLAS at gene-level and filtered by European ancestry with a sample size of greater than 5000. Once filtered, we inferred latent factors using MOFA+. MOFA (Multi-Omics Factor Analysis) is an R library for low-dimensional representation of genomics data using computationally efficient variational inference, which in our case allowed for in-memory computation given its support for handling sparse data. We decided to use K=50 latent factors for our final model as a trade-off between the total variance explained and the interpretability of factors.



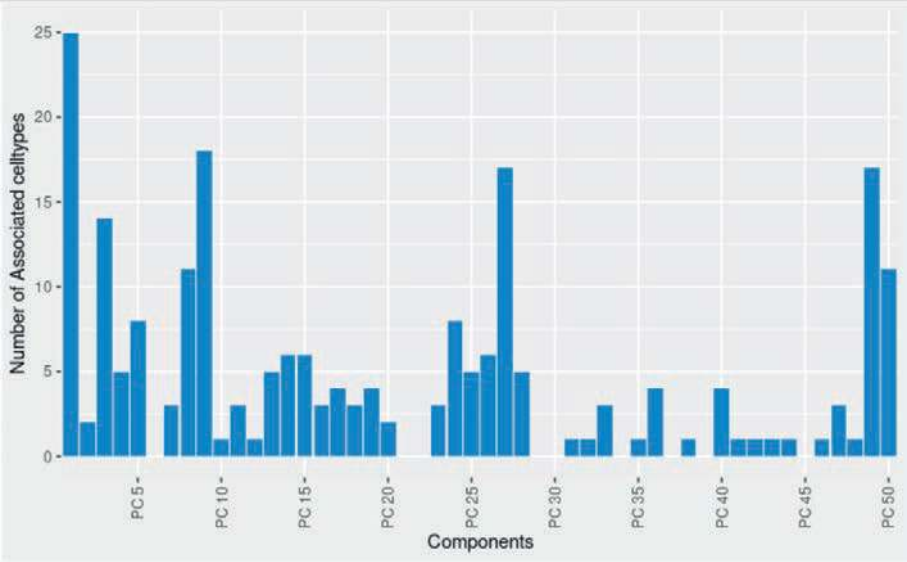
For the characterisation of the MOFA factors we gathered and processed the Descartes dataset, a single-cell atlas of gene expression gathered from human fetal samples covering 77 cell types [5]. We then tested associations between every cell type's gene expression and MOFA factor gene associations using a linear model filtering for p and q-values greater than than 0.005.

Results

In total the 50 latent factors explained 34.66% of the variance of the GWAS. These factors showed strong diversity with 60 different cell-types being associated with at least one factor. For example the top 3 factors for Alzheimer's Disease were factor 49 (32.14% weighting), factor 40 (11.20% weighting) and factor 39 (8.75% weighting). Factor 49, comprising Neurodegenerative traits with the top ones being "Schizophrenia/ Bipolar disorder", "Age started wearing glasses or contact lenses" ,"Cognitive performance" and "Schizophrenia". The most associated celltype for this factor was inhibitory neurons (p=4.28e-12). Factor 40 (11.20%) interpreted as Heart and Lung health with its top traits being "Resting heart rate", "Pulse rate (automated reading)", "FEV1" (forced expiratory volume), "FVC" (Forced vital capacity). The most associated celltype for this factor was Cardiomyocytes (p=3.20e-08).

Finally factor 39 (8.75% weighting), interpreted as a Depression and Anxiety factor had the top traits: "Anxiety - Recent worrying too much about different things", "Depression - Recent feelings of depression", "Anxiety - Recent inability to stop or control worrying", "Anxiety - Recent trouble relaxing" although this factor has no significant cell type associations.

The results of our approach to the dimensionality reduction of the phenome



can be compared to the results of Tanigawa, Y., Li, J., Justesen, J.M. *et al* (2019) in which they applied TSVD to 2,138 UKBiobank phenotypes, inferring 100 latent factors. In this study 41.9% of variance in the GWAS were explained by the 100 factors, comparable to our value of 34.6%.

In conclusion our work presents a new way in which to investigate the shared structure of a wide range of traits with associated cell type annotations and performance comparable with c urrent methods.

Acknowledgements

I am incredibly grateful for the mentorship and support of Brian Schilder, Dr Nathan Skene and the Neurogenomics lab.

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Computational Identification of Conserved Non-Coding Elements with Potential to Act as *Cis*-Regulatory Elements in East-African Cichlids

Cameron Dhruv Bhimsingh
(The Earlham Institute: Haerty Gr)

In the late 70's King & Wilson (1975) [1] laid the foundation for investigations into gene regulation finding an approximately 99% protein-coding sequence similarity between Chimpanzees and Humans. This also carried, with great similarity, for other vertebrates despite the vast morphological, behavioural, and phenotypic differences between species[2].

Recent technological advances have enabled researchers to delve into the genomes of numerous species and allude to the effects of differential gene expression/regulation on phenotype evolution[3]. Importantly, computational approaches facilitate exploration into non-coding genomic regions which could harbour 'cis' -Regulatory Elements (CREs) that modify gene expression[4] (Figure 1). Evidence suggests that Cis-Regulatory Elements (CREs) under purifying selection are key players in phenotype regulation[5] via transcriptional gene regulation[6] as enhancers[6],[8], promoters[9], and silencers[10]. Owing to this, CREs are prime candidates as drivers of organismal change forming the focal point of this investigation.

The Cichlidae family of teleost fish are invaluable to the field of comparative genomics. Comprised of

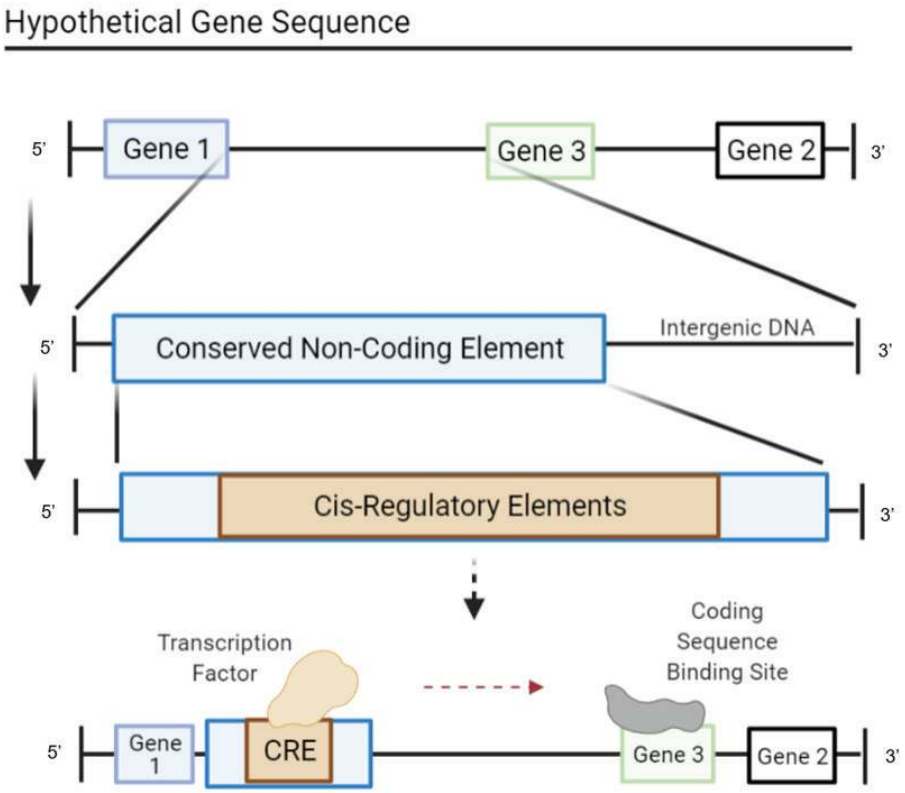

Figure 1: The spatial relationship between conserved non-coding elements (CNEs) and cis-regulatory elements at a hypothetical gene sequence. *Whereby CNEs are located within non-coding DNA and CREs housed within these evolutionarily CNEs demonstrate how CREs may impose a regulatory effect, either up or downstream, on a selection of genes by manipulation of TFBS.*

c. 2285 predominantly East African species[11],[12] and having explosively diversified into numerous adaptive radiations over a relatively brief evolutionary period[13],[14] the family has birthed a plethora of unique phenotypes including; jaw morphologies, body colour, body shape, behaviour, and adaptations to predation[12],[15],[16],[17],[18].

Utilising this model system (Figure 2) within a computational analysis based on evolutionary constraint- which is the notion that functional non-coding elements evolve slower than flanking DNA sequences because of the effects of purifying selection[20], this investigation tests the hypothesis that divergence at non-coding regulatory regions could be a key contributor to cichlid phenotypic diversity by predicting CNEs with potential to act as CREs[20]. T

his allows for the study of CNE evolution in highly adapted species radiations through analysis of highly conserved non-coding elements (hCNEs) and accelerated non-coding elements (aCNEs) as well as identification of species-specific functions of CNEs (based on gene proximity). This work may also allude to the role CNEs play in wider vertebrate evolution and transcriptionally regulated human disorders/disease.

Materials & Methods

Each of the six cichlid species genomes used in this investigation were downloaded In FASTA format from online genomic repositories to the Earlham Institute's High Performance Computing Cluster. Following this, these genomes were then computationally processed and analysed following use of the tools/pipeline in Figure 3.

The process of detecting CNEs with potential to act as CREs began with the generation of a multiple species' alignment file (.MAF) which mapped evolutionarily conserved bases within each haplochromine species to the

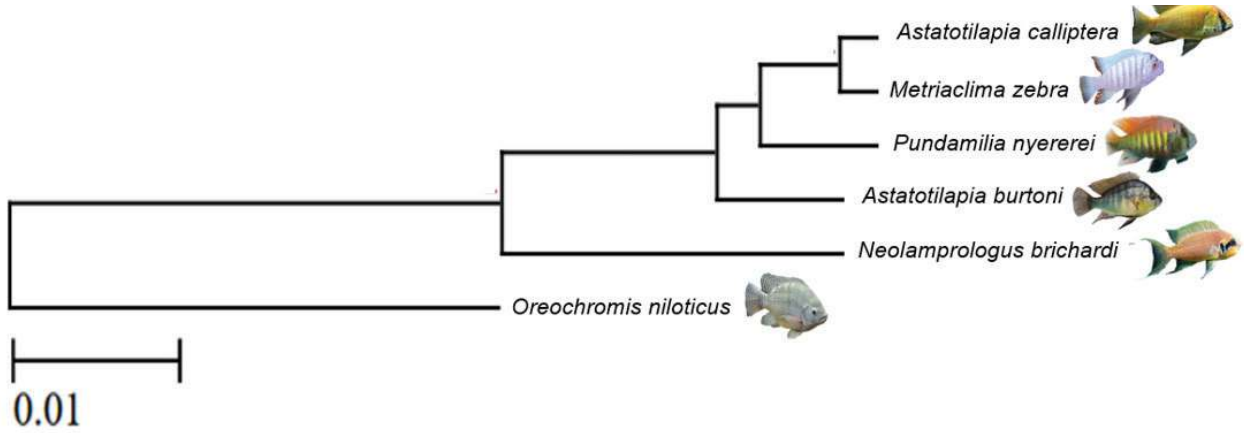

***Figure 2: A Phylogenetic tree as Estimated by PhyloFit Based on neutral evolutionary sites representing the evolutionary relationship between the investigated 5 haplochromine cichlids and ancestral reference species- the Nile Tilapia**. Constructed using ETEToolKit[19] using the NCBI Taxonomy database.*

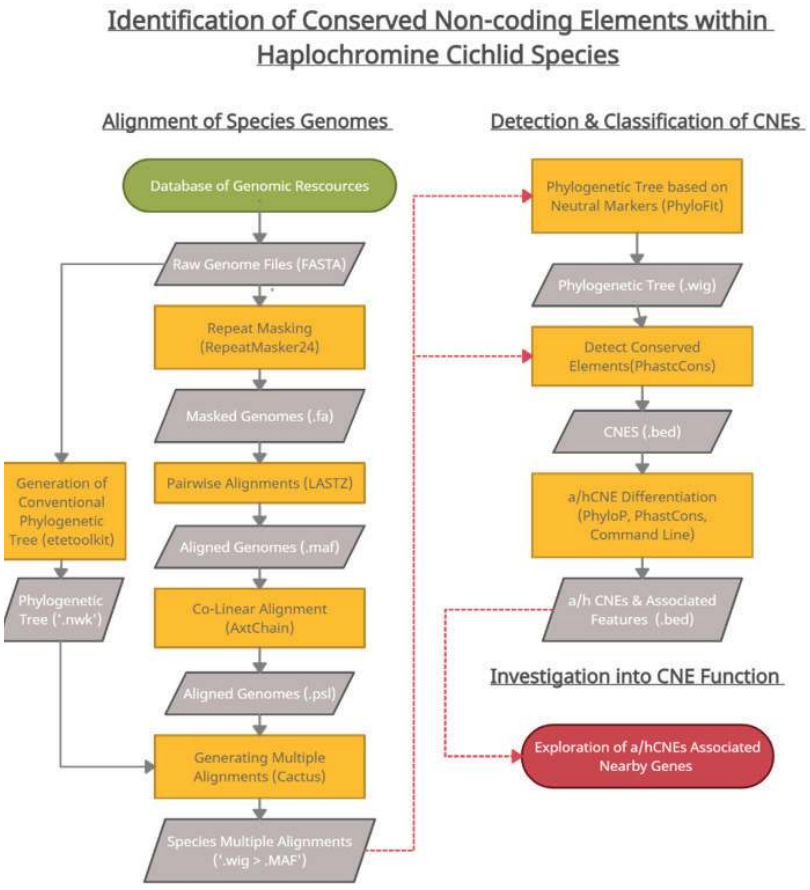

***Figure 3: Simplified bioinformatic pipeline used to analyse East-African cichlid genomes in the identification and investigation of Conserved Non-Coding Elements acting as cis-regulatory elements.** Start= green, End= red, processes= yellow (program used in brackets), inputs/outputs = grey (file format in brackets).*

reference species *O. niloticus*. This then facilitated detection of evolutionarily conserved elements from *O. niloticus* to each query haplochromine species and enabled their intersection with known annotations in the reference genome. Following this the multiple alignment file was then split by species to allow for the detection of aCNEs (PhyloP: altsubscale >1, p -val <0.05) which may have cis-regulatory function.

Results & Discussion

A total of 273,455 CNEs covering 77.49 Mb (7.7%) of the reference genome were identified, <1% (502) of which were found to be highly conserved across the six species multiple alignments. This left the majority of CNEs as candidate aCNEs (272,953).

The overwhelming majority of these were found in intronic regions (lncRNA introns & protein coding introns); regions within 5kb of a known promoter; intergenic regions and UTRs (Figure 4).

These incidences could reflect biological relevance which has previously been demonstrated with 3'UTRs harbouring miRNA binding sites involved in gene expression regulation[21],[22]. Additionally, these observations could be explained by background selection acting on exons being extended to these

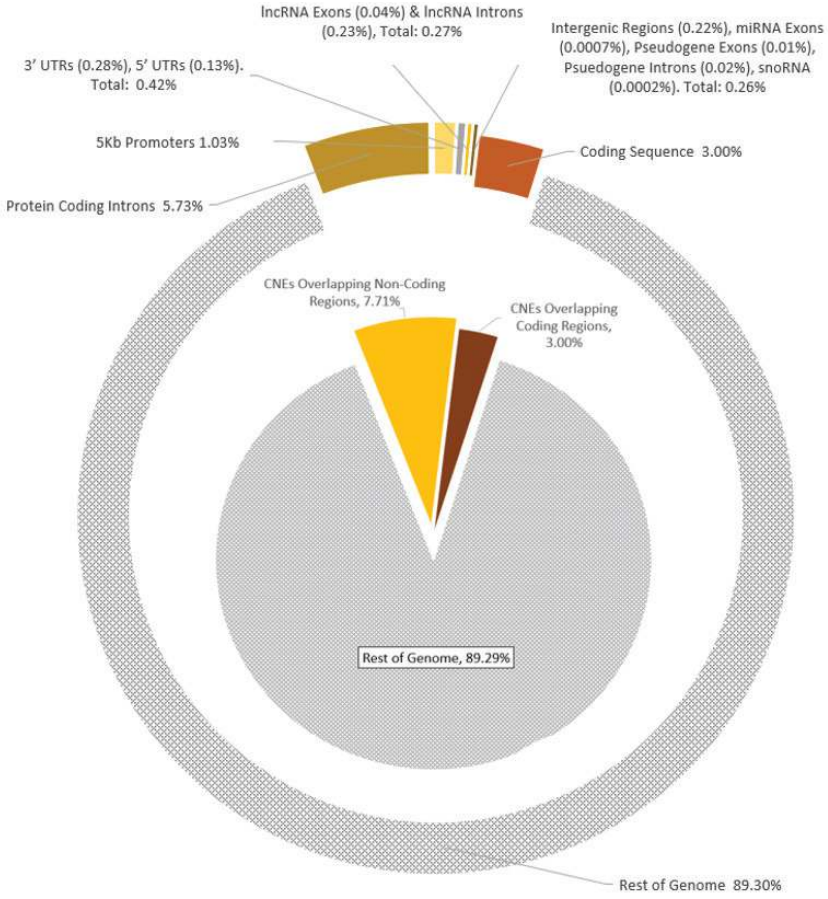

Figure 4: Identified conserved non-coding elements (CNES) following qualitative filtration for CNE length and overlap with associated genome annotation. *Internal pie chart shows the total no. of identified CNES and their grouping by association with annotations in the reference genome. External doughnut chart reflects CNES sub-classifications of parent pie charts classes. Labelling follows (Associated genome annotation, Percentage coverage of ref. genome) format.*

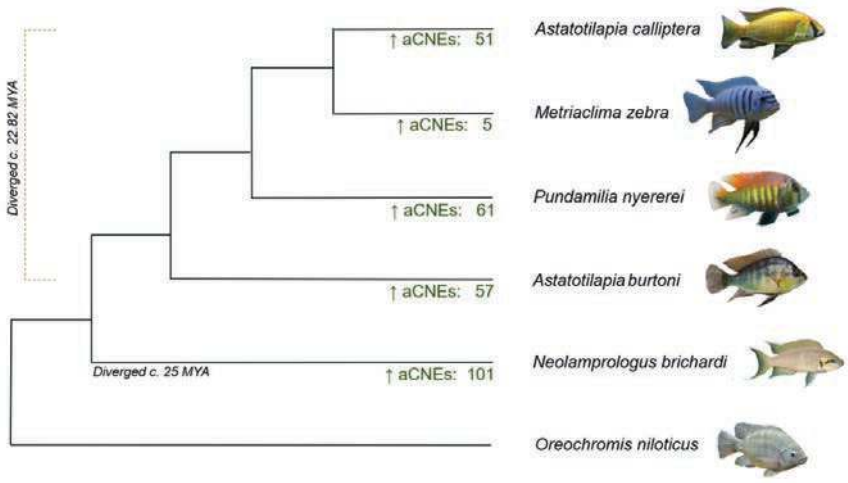

Figure 5: No. of aCNEs occurring within each of the five haplochromine lineages generated from pairwise multiple alignment files derived from a 6-way multiple species alignment. *Total number of aCNEs identified in that lineage is shown in green.*

regions as previously demonstrated in *Drosophila*[23]. Alternatively, these may reflect CNEs around unannotated exons[24],[25] or regulatory elements[26],[27] however, combined, this likely suggests that CNEs found at these regions are more likely to have gained cis-regulatory function than other regions resulting in evolutionary conservation.

The investigation sought to shine a light on those CNEs which demonstrated accelerated lineage dependant evolution (Figure 5) within some of these annotation regions. By looking into aCNEs found near genes of known ontology this investigation proposes a selection of CNEs which demonstrate increased likelihood to have cis-regulatory function (Table 1).

Conclusions & Future Work

In predicting these CNEs, particularly aCNEs with potential to act as CREs, the data produced by this investigation serve as a resource for further studies in cichlids; it's benefit compounded by the development of the pipeline used to identify them, which lends itself to larger scale investigations which may include more species and span larger evolutionary timescales.

This work has been implemented at EI where work has begun to apply this pipeline to a data set 27 teleost species which may trace lineage specific evolutionary history of CNEs across many millions of years encompassing diverse developmental and phenotype-regulating genes.

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Table 1: A subset of aCNEs found near genes of known ontology in Teleost Fish and mammal alike. *Each gene corresponds with a particular aCNE found within the give feature in the reference annotation.*

Species	Feature	Gene	Role in Cichlids
Astatotilapia burtoni	5kb Promoter	gabarapb	Role in the Regulation of the central nervous system
		fbg	Role in Biasing sex-gene expression, particularly sperm competition.
		ddx54	Implicated as a driver of male pregnancy in Syngnathidae
	5' UTR	plod3	Role in lens formation of the eye
		nfxl1	Role in B cell Activation
Astatotilapia calliptera	5kb Promoter	brms1	Implicated as a cancer metastasis suppressor
		CAMTA2	Implicated in cardiac growth
		cnga3	Roles in olfactory signalling and olfactory signalling.
	3' UTR	FABP2	Implicated in the modulation of cell growth and proliferation
	5kb Promoter	clul1	Involved in pigmentation patterning
Metriaclima zebra	5kb Promoter	xpnpep2	Role in collogen degradation
Neolamprologus brichardi	5kb Promoter	ANK2	Role in the localization and membrane stabilization of ion transporters
		cth1	Involved in cleavage, blastula, and gastrula stages of carp embryogenesis
		tmem88b	Implicated in the regulation of the circadian clock
		ppp2r5b	Role in brain development
		ddit4l	Implicated in apoptosis regulation
		DCLK2	Implicated in establishing hippocampal organisation
		KCNAB3	Role in rod/cone eye development
		OTUD7A	Suggested tumour suppressor
		tmprss5	Implicated in numerous physiological and pathological processes
		TAFA1	Role in regulating expression of proteins predominantly expressed int the central nervous system.
		trpc5a	Implicated in calcium channel activity
Pundamilia nyererei	3' UTR	FABP2	Role in cytosolic transport of long-chain fatty acids

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Reproductive isolation among European populations of *Philaenus-spumarius*

Chloe Tucker (The John Innes Centre/ The University of Sussex)

Introduction

Xylella-fastidiosa, a bacterial plant pathogen that affects many crop species, was recently introduced to continental Europe and could be extremely detrimental if it reached the UK. Main vectors include polyphagous xylem feeders such as *Philaenus-spumarius*, but current control strategies are extreme, and little is known about their ecological impact (1).

Phylogenetic inference using mitochondrial genetic data splits European populations of *P.spumarius* into three lineages: Western, Southern-central, and North-eastern (2). In the UK, the North-eastern and Western lineages dominate the North-west and South-east respectively. The Southern-central lineage may have traits that increase *X.fastidiosa* transmission, thus it is necessary to find the likelihood of mixture between populations.

Here I carried out mating trials to determine the degree of prezygotic reproductive isolation (RI) between three populations of *P.spumarius* from England, Scotland, and Italy. Two genetic markers, a fragment each of the mitochondrial cytochrome c oxidase subunit 1 (CO1) and the nuclear elongation factor1α (ELF1α) were used to infer relationships and divergence between populations.

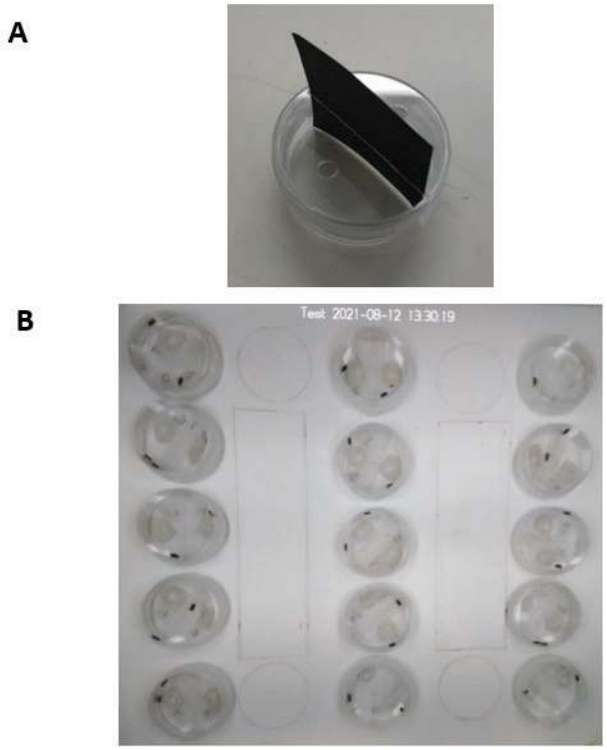

Figure 1: Experimental setup to test behavioural reproductive isolation between European populations *Philaenus-spumarius*.

Pairs of *Philaenus-spumarius* were introduced into modified 6cm Petri dishes and recorded with a Raspberry Pi Zero camera in a PHCbi MLR-352H-PE growth cabinet. **(A)** Dishes had a paper divider to separate individuals that was removed at the start of the experiment after being placed in the growth cabinet. This was done to ensure that the mating experiment effectively started at the same time for all couples. **(B)** A maximum of 15 trials were carried out at once.

Methods

P.spumarius nymphs were collected between April and July 2021 by collaborators and reared in controlled conditions to mature in July. Intra-population mating was allowed to mimic the natural conditions experienced by populations.

15 mating trials were performed for each of the 9 population combination pairs, (15*9=135 mating pairs). Pairs of opposite sexes in 6cm Petri dishes were placed in a PHCbi MLR-352H-PE growth cabinet at 18°C with LED panels (Figure 1). Behaviour was recorded for 4 hours via a Raspberry Pi Zero camera. Student's t-tests were used to analyse the earliest latency among mated couples.

For 5 individuals of each population and sex, DNA was extracted, and PCR was performed for each marker (3, 4). The product was purified, Sanger sequenced and curated manually to call ambiguous bases and remove low-quality sequences. Sequences were aligned with translatorX using MAFFT and a maximum-likelihood tree was inferred with IQ tree (model HQY+F+I).

Results

Of 135 trials, there were only 23 mating events. Couples of the same population had shorter latency than those of differing populations, with means of 77.22mins and 121.24mins respectively,

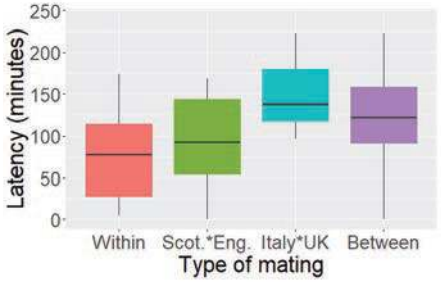

Figure 2: Mating latency among European populations of *Philaenus-spumarius*.

Pairs of English (Eng.), Scottish (Scot.), and Italian (Italy) *Philaenus-spumarius* were observed for 4 hours and latency recorded. Latency is the time from the start of each trial till the first mating instance, pairs that did not mate are excluded. Within refers to pairs of the same population, between refers to pairs where individuals were from different populations. UK refers to data from both Scottish and English populations. * indicates mating events between individuals of the given populations.

but this result was not statistically significant (Student's t-test; t=1.4989, p=0.1522, N=19). Scottish and English bugs preferred each other to Italian bugs with latency means of 92.48mins and 149.99mins respectively (Figure 2), but again this was not statistically significant (Student's t-test; t= 1.7459, p= 0.1114, N=12).

Amplification of both markers was successful, however, sequencing of ELF1α was unsuccessful. The maximum-likelihood model did not separate the three populations with the CO1 marker.

Discussion

These data suggest behavioural isolation, as insects preferred intra-population mating, and a geographic trend, as UK bugs preferred each other to Italian bugs. However, bugs mated infrequently, thus the sample size was too small to assess significance.

The CO1 maximum-likelihood tree did not clearly cluster individuals by population, which conflicts with literature (2). The multiple alignment showed little variation in the CO1 region sequenced, suggesting that the region is insufficient to infer phylogenetic relationships. Additionally, errors would have a disproportionate effect. Therefore, future research should use the region outlined in Rodrigues, et.al., 2014, or the whole mitochondrial genome (2). However, little difference between the lineages would suggest that the UK might be more vulnerable to outbreaks due to high migration rates. The North-eastern lineage is more distantly related than the other lineages, so it is surprising that UK bugs preferred each other to Italian bugs, suggesting factors beyond historical geographic separation are driving RI (2).

Further experiments are necessary to confirm these results. For example, the propensity to mate may be increased by changing the experimental setup or performing trials at a different time. Higher activity has been observed at >20°C instead of 18°C. Moreover, data was recorded over the summer and some populations or sexes may prefer to mate in Autumn (5).

My results indicate that reliable inference of genetic divergence requires using another more variable CO1 region (e.g. as in Rodrigues, et.al., 2014 (2)), or using high-throughput methods that provide more data (e.g. whole mitochondrial genome sequencing, reduced representation sequencing, or whole-genome resequencing).
My results suggest partial behavioural RI among the populations. It may be beneficial to determine the driver of RI, be it genetic drift due to geographical separation, or local adaptation to different types of habitat or host plants. Together with understanding the ecology of *P.spumarius*, this may help us control the main vector *X.fastidiosa*. Overall, this study suggests that the evolutionary history and population structure of vectors should be considered when controlling vector-transmitted plant pathogens.

Acknowledgements

Thanks to the Entomology and Insectary Platform (Victor Soria-Carrasco, Mike Darrington, Anna Jordan, and Susannah Gill) and the Saskia Hogenhout lab (Roberto Biello and Sam Mugford) at the John Innes Centre, collaborators who collected insects (Maria Saponari and Katherine Lester) and the Genetics Society for a great summer!

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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**

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
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, 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 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. 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
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) 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, 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, 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

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

Are you an undergraduate student who is thinking about a career in genetics, or curious to know what that might involve?

Are you an undergraduate student who is thinking about a career in genetics, or curious to know what that might involve?

We have grants available to support you to gain experience in any area of genetics by doing a research project during the summer holiday prior to the final year of your degree. The Genetics Society is committed to supporting and promoting equality and diversity. We welcome applications for the summer studentship programme from all sections of the community regardless of sex, gender identity, sexual orientation, ethnicity, disability, creed, caring responsibilities, or age.

The summer studentship grant includes:

- £300/week allowance for up to 8 weeks
- Up to £750 to cover host laboratory expenses
- A place on the 2-3 day expenses paid* student Summer School Workshop
- If you need it, a Carer's Award is also available to cover child care or other responsibility costs while you do the research and attend the summer school.

*Only travel within the UK can be funded

You must be able to attend the UK based Summer School workshop at the end of the 8 weeks research project (exact dates will be confirmed). The workshop provides an opportunity for all summer students to get together, discuss their research, and start to develop a professional contact network. All costs associated with attendance will be funded by The Society, including caring costs if you need them.

What you need to do next:

- Find a research area of genetics that you are curious about and match that to a research group leader in a University or research institution of your choice. Detailed help with all of this can be found in the FAQ's section on our website.
- With your research group leader, work out what your project will be about. More details about how to do this are provided in the FAQ section on our website.
- Both you and the group leader must be members of the Genetics Society.
- Complete the application form with the help of the research group leader.
- Submit your application before 31st March 2022.

FAQ and full details of conditions and how to apply are available on our website: https://genetics.org.uk/grants/summer-studentships/

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
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

Application deadlines are @ midnight on lst March and lst 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.

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.
- Scheme (C) is open to members with a UK base wishing to attend virtual conferences organised both within or outside the UK, and to non-UK-based members wishing to attend a virtual conference based in the UK. We regret that we cannot consider applications from bases outside the UK for conferences organised 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.

View upcoming Genetics Society Scientific and Sectional Interest Group meetings.

(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 (Scheme B) 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.

(C) Registration costs for (non-Genetics Society) virtual meetings.

Grants of up to £300 are available to cover registration costs for virtual conferences.

The virtual conference grant (a temporary measure during the pandemic situation, to be reviewed on a six monthly basis) 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

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

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.

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

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 https://genetics.org.uk/membership/direct-debit-mandate/. 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.

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