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Galton Institute

Exploring Human Heredity

The Galton Review

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EDITORIAL

2019 has so far been a year of considerable upheaval in the UK but here at the Galton Institute we have been trying to continue the good work of previous years and in this issue of the *Galton Review* we will, among other things, be considering the work of Council.

This is a group of talented people with a diverse range of expertise in various fields related to the aims of the Institute. On page 6 is the first in a series of interviews with members of Council in which we find out what has been the driving force in their career and what it is about genetics which fascinates them. The first interviewee is **Professor Philippa Talmud** of UCL, Vice-President of the Galton Institute. Her story makes for absorbing reading.

On page 12, the **President and Professor Dallas Swallow** pay tribute to **Professor Sue Povey** who died in January. Her obituary paints a picture of a talented geneticist with strong principles and obvious charisma. She will be remembered, among many other things, as a major contributor to the Human Genome Project.

On page 16 is the report from **Dr Versha Prakash** from Royal Holloway, University of London, who received the Institute's Postdoctoral Travel Award and went to Denmark to work on genome editing therapies for rare genetic diseases.

We have two major conferences coming up this year. In June, there is the third biennial Teachers' Conference in Manchester with an opportunity for secondary teachers to update their knowledge on various topics ranging from Epigenetics to Bioinformatics. The list of speakers is impressive. Then at the end of October is the Annual Conference at the Royal Society and this year's theme is 'New Light on Old Britons' and we will consider the ancestry of the British population. **Professor David Coleman** has brought together a list of outstanding speakers with the Galton Lecture being presented by **Professor Sir Barry Cunliffe** on 'The Celts in Britain – a romantic fiction?'; it promises to be another exceptional conference.

Details of both conferences can be found on page 28 and if you would like tickets please go to 'Future Events' on our website.

Robert Johnston

Galton Institute Council

Council Meeting 20.3.19.

The Galton Institute Council consists of fifteen Fellows, acting as Trustees, five of whom are officers. The full list of Council members, along with a brief biography of each, can be found on the website. Each member of Council is an expert in his or her own field and is a member of one or two sub-committees dedicated to a particular role.

Council meets three times a year, in March, June and November in the impressive surroundings of the Society of Antiquaries in Burlington House, Piccadilly. Typically, the meetings involve deliberations on finances, membership, past and future conferences, grant applications and publications. Members of Council are also Trus-

tees of the Galton Institute Artemis Trust and responsible for administering the funds of the Trust in accordance with the Trust Deed.

Recently, discussions have also taken place on how to expand our activities to increase public awareness of what we do.

We thought you might be interested to find out more about our Trustees and, to this end, are running a series of interviews with them, the first of which with Professor Philippa Talmud appears on the next page.

Council Meeting 20.3.19.

Picture on facing page: L to R: Professor Veronica van Heyningen, Professor John Beardmore, Professor Dian Donnai, Professor Andrew Read, Dr Lesley Hall and Dr Paul Hurd.
Picture Above: L to R: Professor Philippa Talmud, Professor David Coleman, Mr Robert Johnston, Dr Jess Buxton, Professor David Galton, Professor Dallas Swallow and Mrs Betty Nixon.

My Life in Genetics – an Interview with Professor Philippa Talmud, Vice-President of the Galton Institute

You grew up in South Africa, what brought you to the UK?

The Nationalist Party came into power in 1948 so I knew no other system of government, other than apartheid. I came from a family very opposed to the regime, but not actively involved in the struggle against apartheid. Even so, I vividly remember our house being raided by the plain-clothed secret police, a bunch of heavies who went through our house with a fine toothed comb. Luckily the mildly subversive literature that we did have was hidden in a storeroom, which they never found. It is upsetting, even now, to think back to that period, and recall the extent of the discrimination against anyone who did not have a white skin. But apartheid didn't stop there. It penetrated every way of life. As a repressive regime it also restricted all forms of arts and culture and free speech, so living under such a system was extremely oppressive for everyone. It was really dangerous to defy the government and could result in imprisonment and 90 days incarceration without trial! At that time it seemed that the only way change would come about was by bloody revolution. Thankfully this never occurred but it took someone like Nelson Mandela to bring about the peaceful transition. My mother, a single parent by then, encouraged all three of her children to go abroad, as it seemed that South Africa had no future. I graduated with a BSc (Hons) from University of Cape Town and like many of my contemporaries I left immediately for the UK, with plans to study for a PhD. I registered at UCL to work with Professor Dan Lewis who had managed to get funding for me from the Wellcome Trust.

What was it about Human Genetics that convinced you to move into this field?

It was some time into my career that I changed to work on Hu-

man Genetics. When I first thought of doing a PhD the research that excited me most was in microbial genetics, particularly fungal genetics. Fungi such as *Coprinus lagopus*, the organism that formed the basis of my research, was an excellent test organism because of the monoploid and diploid phases in its life cycle, and the fact that the reproductive cycle was so short, so experimental results were obtained quickly. My research was related to ageing and examining mutations that could be introduced into the organism by replacing naturally occurring amino acids that are essential for growth, with synthetic amino acid analogues that affected the reliability of DNA synthesis and repair. But after more than 10 years working in the field (and my best paper being published in Nature!) I started to get restless, and I decided to make a move. Human Genetics had changed a great deal from the days of family pedigree studies and lod scores, to the 'new genetics' using DNA variants as genetic markers in either population or family studies. I basically had to retrain, learning how to do a Southern blot and how to pour sequencing gels, a steep learning curve! After a short spell working on Brittle Bone Disease I started working on the genetics of heart disease.

What areas of Human Genetics have you been involved with at UCL?

In 1983 I went for an interview to work with Steve Humphries at St Mary's Hospital Medical School. Steve's lab was working on heart disease, from two points of view. Firstly Steve had a special interest in the monogenic dominant disorder Familial Hypercholesteroleamia (FH) (for which Brown and Goldstein were to receive the Nobel Prize in Medicine or Physiology in 1986), but the lab also researched the more common form of heart disease, which was considered to have both polygenic and environmental determinants.

In 1991 the lab moved to UCL, before an interim period at the Sunley Research Centre, at Charing Cross Hospital. So essentially all my research at UCL over the last 25 years has been in the field of heart disease. From the start I studied genes involved in cholesterol and triglyceride metabolism, namely genes determin-

ing the apolipoproteins, enzymes and receptors involved in these pathways. Using common genetic variants we carried out association studies of healthy individuals identifying common variants that showed statistically significant differences in cholesterol or triglyceride levels. However, association *per se* was not enough to confirm causality, and together with co-workers we carried out functional studies on specific genetic variants to validate that these genetic associations had a sound functional basis. The variants we tested were in gene promoters, signal peptides or affected enzyme activity or receptor binding sites. A more difficult aspect of the research was to examine potential environmental exposures that could affect heart disease risk, so as to get a more complete picture of this multifactorial disease. The obvious factors were smoking and alcohol intake, and we published several papers looking for smoking-genotype interactions. Unfortunately, data on environmental measures is less accurate than genotype as it often relies on questionnaire data filled out by study participants, which are notoriously inaccurate. A better measure would have been of blood levels of smoking or alcohol by-products.

What has been the main focus of your work in recent years?

Our research was influenced by two major breakthroughs in technology. Firstly the development of gene chips that made it possible to examine anything from a few hundred to a million genetic variants in a single sample in one experiment, and secondly, the genome wide association studies (GWAS), looking at association of genetic variants evenly spaced across the whole genome. Furthermore, UCL was particularly fortunate in having several large population studies of healthy individuals who had been followed for anything from ten to fifty years, with rich data on disease status, and biochemical and life style information. These included the Whitehall II study of civil servants, the two large birth cohorts, and the Northwick Park Health Study of individuals drawn from GP practices across the UK. Together these provided us samples from more than 17,000 individuals with long time follow up data.

Another big breakthrough came in the form of the development of statistical algorithms to study such big data. The change for us was huge. Prior to this we had worked primarily on our own, researching a few genes at a time with occasional collaborations. Suddenly this all changed and collaboration became the name of the game. And so we embarked on an extremely exciting time with very lively weekly meetings with colleagues from several different UCL departments brain-storming the huge amount of data that became available from a 50,000 SNPs (single nucleotide polymorphism) in these 17,000 samples. This was an extremely productive, exciting and rewarding time with many novel association found and subsequently confirmed by replication in other study samples, The results from the Wellcome Trust coronary heart disease (CHD) GWAS was published soon after and shocked the heart disease community by identifying a completely novel gene of unknown function as the biggest CHD GWAS hit. We could now include these variants in our studies. The beauty of this period for me was that we all were so involved in the research. The study of many variants at one time led to the development of a gene score to determine the combined effect of inheriting several of these risk/ protective CHD alleles.

In addition, one of the conundrums of FH, the monogenic form of heart disease that the lab had worked on for many years, was that although mutations in three genes had been identified as disease-causing, they could only account for disease in ~ 40% of patients. Using this new gene score approach, combining data of the top ranking LDL cholesterol level variants, we were able to show that at least 80% of those with clinical FH but with no mutation had, in fact, co-inherited these risk alleles and had a polygenic, and not monogenic, form of hypercholesterolaemia.

Subsequently as data pooling led to larger and larger studies and international collaborations with often as many as 500 co-authors per publication, the research was taken over by a few key individuals, often from abroad, and the excitement, involvement and hands on analysis faded. My retirement coincided happily with this period.

Which colleagues have had the greatest influence on your work?

When I joined Steve Humphries at St Mary's, Professor Bob Williamson was head of department. Bob was a slightly controversial figure but he ran a department that was open and casual, while still demanding very high standards of researchers. Bob's legacy in Human Genetics influenced many labs, as his colleagues and students went on to head genetics departments in the UK and abroad based on Bob's ethos. Steve Humphries was one of these and I think that Steve ran a lab that was stimulating and at the same time was extremely supportive, resulting in a great productive atmosphere to work in, which I have always felt was important.

Another person who influenced the way I think about research, but who I only met late in my career is Professor Uta Frith. Uta is a developmental psychologist working in cognitive neuroscience at UCL. When Uta became an FRS she decided to make use of the Royal Society as a place for women to network and she started the 'Science and Shopping' group. It was a monthly get together of women from all branches of science who would meet and chat about anything from their scientific interests to child care and even shopping!! Uta taught me the importance of networking, but also the value of mentoring and helping younger students and colleagues with career development.

What do you consider to have been the greatest achievements in Human Genetics in your time?

There have been several key developments that have made ground breaking changes that influenced my research particularly, and also moved the whole field forward. The first I think was the development of the Polymerase Chain Reaction which enabled geneticists to study small, discreet fragments of DNA for variant analysis or cloning etc. Then the functional studies such as luciferase assays for promoter investigation and the yeast two hybrid system which enabled us to look at protein DNA interactions. The SNP chip and whole genome scans radically changed the way

population genetics could be studied and identified potential new drug targets. Then shortly after came high throughput sequencing which has identified many novel genetic variants. Together with development of statistical algorithms for analysis of big data, this has revolutionised disease research. What might prove to be the biggest breakthrough is the development of gene editing using CRISPR/Cas9 which is on the verge of clinical trials to correct mutations in monogenic diseases. Human Genetics is poised to see the benefit of this major breakthrough on a very wide range of diseases including cancers.

What do you think will be the greatest challenges for geneticists in the coming years?

Whole genome sequencing has led to the identification of hundreds of novel genetic variants. Many of these are clearly functional but there still remain even more that are defined as 'variants of uncertain significance'. In order to use these in family studies, to identify family members at risk of the disease, it is essential that their functionality be determined. This is a major task ahead. Then of course the introduction of gene editing in a therapeutic setting opens many ethical and scientific questions.

What role has the Galton Institute played in furthering research into Human Genetics?

In my mind the role of the Galton Institute is to promote knowledge dissemination. The annual Galton Institute meeting, held at the Royal Society, brings world renowned researchers together to discuss their ground breaking work on a wide range of topics. The Galton Institute small grants enable many smaller meeting on various aspects of genetics to be held. We help fund at least 8-10 meetings a year, which is important for the exposure of results and ideas. Through the Genetics Society we support students who want to attend major meetings abroad which they would not have funding to do otherwise. So I think the Galton Institute fills a niche not filled by other charities.

Finally, please tell us one thing about yourself that is not widely known.

Before moving to the field of human genetics, I considered giving up research. I had reached an impasse. So in 1980-81 my then husband and I, with our two small children, went to live in the South of France for nearly a year. I have always felt that work-life balance was important and I wasn't happy with the way my life was going. This might have been a cop-out but it was an idyllic time, although not exactly providing an answer to my professional dilemma! When we came back to the UK and I had my third child, I was at a loss what to do. I seriously considered going to study medicine. It had always been on my mind, but the idea of going back to study for a further 5 years didn't sound all that appealing with three young children. A friend put me in touch with Bob Williamson who had connections with labs working in Human Genetics in London and through him I started the next phase of my career in Human Genetics, and ultimately CHD research. The result led to an exciting, and extremely worthwhile career in research.

OBITUARY

Margaret Susan Povey, Human Geneticist, born 24 April 1942; died 11 January 2019

Molecular geneticist and member of the Galton Laboratory, who was a leading contributor to the Human Genome Project

In 2003 the Human Genome Project (HGP) published the complete sequence of human DNA. Sue Povey, who has died aged 76, contributed greatly to this international collaborative project with her team at University College London, her work as a molecular geneticist having started much earlier, in the late 1960s. She

was motivated throughout by a strong interest in people and disease.

At the outset she exploited newly developed enzyme detection systems that revealed differences between individuals and among species, allowing her to solve a number of longstanding puzzles. One was mapping the chromosomal location of human genes, initially by family studies, which use inheritance patterns across the generations, like Gregor Mendel's pioneering work, to identify closely linked genes. Later mapping used human-mouse hybrid cell culture. She co-authored more than 60 gene-mapping papers (several with us) before the human genome was fully sequenced. The exponentially growing chromosome maps provided critical landmarks for speedier mapping with new DNA technologies, which Sue was quick to adopt.

A remarkable early achievement, initially with enzyme technology, subsequently with DNA, was her contribution to the understanding of the origin of two types of gynaecological tumours: hydatidiform moles, products of abnormal conception with unbalanced paternal chromosomal contributions, and ovarian teratomas, which arise as a result of faulty egg development. More mundane, but extremely practical, was the recognition, through genetic marker analysis, that many cell lines used for research had been taken over by other fast growing cancer cell lines, such as HeLa. Sue was also able to link her own niece's liver disease, which took her life at the age of 13, to deficiency of alpha-1-antitrypsin. Sue went on to contribute significant research in this area, and also delivered early molecular diagnostics to other families.

As DNA technology advanced, the pace of disease-gene mapping accelerated. Sue attended and contributed to every Human Gene Mapping Workshop between 1975 and 1991. Her early work in this field made her appreciate the vital need for precise gene naming and annotation. She took over from Phyllis McAl-

pine as chair of the International Human Genome Organisation (HUGO) gene nomenclature committee in 1996, continuing until her official retirement in 2007. Repeated success in obtaining US and UK funding enabled Sue to build up a team of postdoctoral researchers and programmers for gene-naming during the incredibly intensive, but exhilarating, time of the HGP.

After establishing maps with some genes assigned to each chromosome, work began on searching for the positions of disease-associated genes. In 1985 Sue began to map the complex disease tuberous sclerosis (TSC). She soon succeeded in linking TSC to the ABO blood group, which in turn was assigned to chromosome 9. However, further analysis showed that the disease in some families mapped to chromosome 16. The race was now on to identify two different TSC genes. Although Sue was aware of international rivalries, she was always ready to collaborate, so her group was one of eight different labs that, in 1997, co-authored the paper identifying the gene on chromosome 9. As diagnostic results accumulated, she set up and managed the TSC variation database, an invaluable international resource for interpretation of molecular genetic results, and remained involved until her death. In recent years she made massive contributions to developing ethical guidelines for maintaining confidentiality while also allowing genetic disease data to be shared for the benefit of other patients, diagnostics and research.

Professor Sue Povey*

**Image: Wellcome Trust History of Modern Biomedicine project*

Born in Leeds, Sue was the daughter of Jack Povey and his wife, Margaret (nee Robertson). Jack was an RAF intelligence officer during the Second World War, who went on to set up the physics department at St Michael's College, now Mount St Mary's Catholic High School, in Leeds. Margaret was the first woman to graduate from Leeds Medical School, becoming a paediatrician and running a maternity hospital in Leeds. From her, Sue gleaned that women can pursue any career they wish, but should avoid learning to sew. From Notre Dame Collegiate School in Leeds, she went to Girton College, Cambridge, and in 1964 graduated in genetics. Three years later she qualified in medicine at UCL. After clinical training in Liverpool and Huddersfield, she spent a year with the Save the Children Fund in Algeria.

Her decision to become a research scientist was triggered by an earlier overland trip to India, which yielded her first paper on the genetics of leprosy, and by a stint in the laboratory of the renowned human geneticist Harry Harris, whose group at the MRC Human Biochemical Genetics Unit at UCL she joined as a staff member in 1970. She remained at UCL for the rest of her career, becoming Haldane professor of human genetics in 2000.

Unassuming at first sight, Sue could be fierce in defence of her principles, taking on a whole committee if necessary. To enable her staff and students to attend international meetings, she would often travel at very low cost herself. Several of her students, many of them women, are now professors or in leading professional roles. Holidays were generally extensions of work trips to interesting countries where she could walk and enjoy the fauna, flora and terrain with colleagues.
Sue is survived by her brother, Phil, and nephew, Ian.

Veronica van Heyningen
Dallas Swallow

A version of this obituary previously appeared in The Guardian, to whom we are grateful.

Galton Institute Postdoctoral Travel Award 2019 Report

Research project: Development of CRISPR/Cas genome editing as a treatment for Ataxia Telangiectasia
(**www.actionforat.org/crispr/**)

My research is focused on developing genome editing based therapies for the genetic disease called Ataxia Telangiectasia (A-T). A-T is a rare disease caused by defects on the ATM gene. The ATM gene is responsible for producing the ATM protein, lack of which causes neurodegeneration in the cerebellum, immunodeficiency and a high risk of cancers. Currently, there is no cure for A-T.

The question that I am trying to address in my research is whether repair of the faulty ATM gene using CRISPR-Cas genome editing is capable of restoring the protein deficiency, especially in human haematopoietic stem cells (HSCs) that have the potential to form an entire immune system. If feasible, this approach could be beneficial in alleviating the immunodeficiency aspect of A-T. HSC genome editing has already seen success in various other diseases including sickle cell disease, haemoglobinopathies and some forms of severe combined immuno-deficiencies

So far into my research, I have carried out genome editing of the ATM gene in cells taken from A-T patients. However, the cell types of importance are human blood stem progenitor cells (CD34+ cells). Human CD34+ cells can be purified from the umbilical cord blood that is donated by mothers soon after childbirth. These cells are extremely resourceful but they are difficult to obtain, grow, and require use of specialized reagents and expertise. These cells are also very expensive, if purchased commercially.

In order to learn how to purify and to carry out genome editing in CD34+ cells, I chose to work with Dr Rasmus Bak. Established as a Fellow at Aarhus Institute of Advanced Studies and an Associate Professor at Aarhus University in Denmark, Dr Bak is

renowned for his research experience in developing and applying novel strategies for genome editing in human cells. In the past few years, Dr Bak has developed tools to carry out highly efficient and robust genome editing in human blood stem cells.

The two primary goals of my visit were:

To learn how to purify the cells from fresh umbilical cord blood. I had the opportunity to observe this procedure in the lab with a trained research technician who routinely carries out this procedure. It is demanding and time consuming, lasting approximately 5-6 hours. This was carried out using a specialised commercial kit, handling and preparation of which is critical to ensure optimal results. I now understand the requirements for processing and handling umbilical cord blood to purify viable stem cells.

To carry out genome editing in CD34+ cells. I had the opportunity to follow through an entire week-long genome editing experiment. The experiment was carried out by a postdoctoral researcher, who has extensive training and experience in working with purified CD34+ cells. I had the chance to follow through their optimised protocols and data analysis methods. All of these methods are directly applicable to my research.

Apart from the planned experiments, I also had the opportunity to participate in an on-going *in vivo* genome editing experiment involving the use of laboratory mice. I worked alongside a senior researcher who had extensive experience in animal work. I learnt how to dissect mice and prepare samples to analyse the outcomes of genome editing using flow cytometry and droplet digital PCR. Although not part of my existing project, genome editing in A -T mouse models will form the next stages of my current research.

Apart from lab work, I had a chance to present my research in a seminar where I received constructive feedback and several improvisations for my work.

Overall, my visit was very fruitful. Most importantly, I had a chance

to work as part of an inspiring and thriving research group, with whom I can consult throughout the project. I am confident that using the techniques learnt in Dr Bak's lab, I can now progress to edit *ATM* in wild-type CD34+ cells and then we shall try to obtain very rare *ATM*-mutated cells from patient-derived umbilical cord cells for comparison.

I am very grateful to Dr Bak and his research team for being so generous with their time, expertise and hospitality and I thank the **Galton Institute** for making this research visit possible.

Dr Versha Prakash
Postdoctoral Research Associate,
Royal Holloway University of London

Postdoctoral travel grant

The Galton Institute is seeking applications for our postdoctoral travel grant, available to outstanding postdoctoral researchers, normally within 6 years of receiving a doctoral degree, working in the field of genetics.

The Fellowship, which is up to £6,000, aims to support visits to carry out research into aspects of human inheritance in laboratories abroad 'to enrich the research experience and help develop the scientific career of the Fellow'. The duration of the Fellowship needs to be well justified and requests for up to 6 months will be considered. Applications will also be considered for attendance at advanced, intensive, high quality laboratory-based courses, e.g.: at Cold Spring Harbor, Woods Hole and similar centres.

Full details of the grant can be found on our website at:
http://www.galtoninstitute.org.uk/grants/galton-institute-postdoctoral-travel-grant/

Bridging The Gaps
Interdisciplinary Approaches in Life Sciences

European Molecular Biology Laboratory PhD Symposium
October, 2017 Heidelberg

This international PhD Symposium focused on understanding and analysing living systems by means of different scientific disciplines. The conference was organised by first-year PhD students of EMBL and was mainly aimed at PhD students, but was open to everyone. It attracted researchers of all stages, and from different countries, who were interested in interdisciplinary research in the life sciences. A total of 170 participants met in the conference facilities of the Advanced Training Center at EMBL. Over the course of three days, lectures from 15 invited speakers together with short and flash talks, selected from abstracts submitted by participants, were given.

After opening the meeting by the organisers, a welcome speech was given by **Dr Ian Mattaj**, Director General of the EMBL, who highlighted the long tradition of the EMBL PhD Symposia and its origins in the curriculum of the PhD education at EMBL, giving students the opportunity to gain experience in sponsor acquisition, assembling of a scientific programme and handling of the complete logistics behind a conference.

The following scientific programme was set under a comprehensive theme for each of the three days of the symposium: “Evolving Life, Modifying Life and Visualising Life”. “Evolving Life” was opened with the keynote lecture, held by **Dr Jason Chin** from the MRC-LMB in Cambridge, speaking about his research on reprogramming of the genetic code in cells and animals, and applications of this technique for chemical biology. The topic was further

discussed during Dr Chin's blackboard session parallel to the lunch break. The next lecture was given by **Angela Relogio** from the Charité Medical University of Berlin, who talked about her work on circadian rhythms and the importance of time in biological systems. Following a lecture from the science journalist **Franziska Badenschier** from Science Media Centre Germany, on how to communicate with journalists about scientific topics, **Janet Kelso** from the Max Planck Institute for Evolutionary Anthropology in Leipzig, spoke about her research on comparative genomics to analyse mixing between modern humans and Neanderthals. The last lecture of the first day was given by **Professor Barbra di Ventura**, from the University of Freiburg, about her work on optogenetic tools to manipulate and analyse dynamics in living systems. The day was closed by blackboard sessions with Franziska Badenschier, **Professor Dorothea Fiedler** (FMP Berlin), Professor Barbara di Ventura and **Dr Eva Haenssler** (QIAGEN) allowing discussions in smaller groups.

The second day with the topic "Modifying Life", started with a lecture from **Dr Christoph Merten** from EMBL about microfluidics and applications of the techniques for compound screenings and for diagnostics. This was followed by a keynote lecture given by **Professor Dorothea Fiedler**, who talked about the relevance of inositol pyrophosphates in signalling and how to use chemical tools to investigate them. This lecture was followed by a talk on 4D-bioprinting of different tissues given by **Dr Mikael Garcia**, project manager at the French start-up company Poietis. Next, **Carla Fehr** from University of Waterloo, Canada, talked about women in the Scientific Community and how hierarchical structures developed in recent times. The next lecture was given by **Professor Ernst Stelzer** from the Goethe University in Frankfurt, about engineering and applying state-of-the-art microscopy techniques, and in particular the use of light sheet microscopy for imaging of organismal development. This second day was closed with a panel discussion about career perspectives in the life sci-

ences with panelists from industry, start-up companies, science communication and academia.

The last day of the symposium was held under the theme "Visualising Life" and started with **Professor Suliana Manley** from EPF Lausanne, who talked about advances in super-resolution microscopy. The subsequent keynote lecture was given by **Professor Michael K. Rosen**, from the University of Texas Southwestern Medical Center, addressing the physical mechanisms of phase separation and liquid demixing and the relevance for the organisation of cells. **Dr Arnaud Gautier** from École Normale Supérieure in Paris, talked about developing tools to turn on fluorescence of protein tags via small molecules with high temporal precision. Subsequently, **Dr Fiametta Ghedini** from Spotify talked about the use of comics for science communication. Next, **Dr Lori Passmore** from the MRC-LMB in Cambridge gave a lecture about the use of cryo-electron microscopy to understand polyadenylation of mRNA. Finally, **Professor Carla Fehr** talked about "The pleasures and perils of researching across disciplinary boundaries". The scientific programme was concluded with blackboard discussions with Dr Gautier, Prof. Rosen, Dr Passmore and Dr Ghedini before the symposium was closed with an award ceremony for posters and short talks.

During these three days, a diverse programme was offered, ranging from scientific talks to science communication and philosophical discussions. All these talks emphasised the power of bridging disciplines to reach a common goal: Understanding life.

Thanks to the commitment of all those involved and the kind support of our sponsors and supporting grants, including the generous **Galton Institute** Conference Grant, the conference was an insightful and scientifically highly valuable meeting.

Maximilian Beckers
Annika Brosig
Isabell Schneider
EMBL

Genetic Alliance UK
Successful Partnerships Conference 2016

This is the flagship event in Genetic Alliance UK's calendar and is an excellent opportunity to bring our members together to hear about our achievements for the previous year, what's going on in the research and policy world relating to genetics and to announce plans for the year ahead.

126 people attended the event, over 88 of them representing the patient group members, with other attendees being researchers, clinicians, industry, trustees and staff.

Presentations covered a variety of issues: **Dr Gina Radford**, Deputy Chief Medical Officer discussed the impact of genomics on healthcare; **Sarah Rickwood** from IMSHealth covered the new challenges of Orphan Drugs in Europe. With the focus of our conference being partnerships, we heard from **Jan Mather**, Chair of Behcet's Patients Centres on their partnerships with clinicians. Our members were also at the centre of our event: **David White** from Cavernoma Alliance UK, **Heather Band** from Batten Disease Family Association and **Gillian Thomas**, a carer for a patient with myeloma, shared their experiences of partnerships with a variety of stakeholders such as researchers, patients and policy makers.

Feedback of the conference through an evaluation survey to all delegates was positive; all of the speakers were rated 'Good or Excellent' by 96% of attendees while 92% said they would be interested in attending the conference next year.

Comments from attendees included: "This was wonderful. Thank you for giving us the opportunity to meet up and talk to each other" and "Good programme, with a nice diversity of talks, both patient group examples and policy points." Positive feedback was

also received on our conference pack, which included a booklet containing all the information for the day, including names of attendees and also our annual report and accounts for 2015-16, which may also be accessed at:
https://www.geneticalliance.org.uk/media/2503/genetic-alliance-uk-annual-report-2015-2016.pdf

The conference was supported by the **Galton Institute**, for which the charity is very grateful.

Mariana Campos
Genetic Alliance UK

University of Cambridge's Centre for Research into the Arts, Social Sciences and Humanities (CRASSH)

Mapping Morality in Global Health

26-27 July 2018

This conference, jointly organised by academics from the University of Cambridge and Oslo, had originally been conceived of as an opportunity to explore concerns around the growing popularity of 'effective altruism', especially as applied to global health funding. Of particular concern was the oft-deployed tagline of "simple solutions to complex problems". To those of us involved in doing either the work of public health or the work of closely observing it, the promise of a "simple solution" to performing efficacious and morally robust global health, seemed incongruous with the unavoidably complex and variable moral, political-economic and technical landscapes of global health. As effective altruism is not the only moral framework deployed in global health that utilises a simplified rendering of these complex and diverse moral landscapes, it was necessary to expand the scope of the conference beyond consequentialist ethics. Ultimately, the conference was realised as a forum in which the complexity, diversity and nuance of the moral

landscapes of global health might be re-asserted and the processes and consequences of particular “flatland” renderings of it be properly examined, be they consequentialist, humanitarian, religious or otherwise.

The conference was a success, both in terms of its popularity and also in its stated ambition of reasserting complex moral landscapes relative to reductionist moral paradigms of global health. Thirty speakers from nine countries took part in the conference, which included leading epidemiologists, anthropologists, historians, law scholars, sociologists, economists, and political scientists. In addition to the speakers, an audience of 25 people registered for the full two days of the conference with a number of others, mostly students, attending individual sessions. The conference fuelled numerous impassioned yet considered discussions, as speakers and audience members attempted to trace the contours of a range of moral features in global health. Whilst it is not possible to catalogue all of these discussions here, or do justice to even the most prominent ones, it is still worth mentioning a few recurring themes and a particular tone to the discourse that proved invaluable in developing a meaningful rendering of the moral landscapes of global health.

The conference commenced with a paper by anthropologist **Professor Peter Redfield** on the fungibility of human life. The paper and discussion which followed set a particularly productive tone, one that focused on the relationships between different moral renderings of situations rather than attempting to assert the rightness or wrongness of any particular moral framework. Throughout the conference this constructive, non-combative approach to moral discourse was further bolstered by a recurring focus on empirical nuance rather than rhetorical absolutes. In a number of papers this was manifest by a focus on historical continuities, showing the shared origins, and subsequent divergences,

of different contemporary moral frameworks. This approach was perhaps clearest in a paper by **Professor Jean-Paul Gaudillier** tracing the linear, rather than revolutionary, emergence of the 'Global Burden of Disease' (GBD) metric in the 1990s and the reductionist approach to health policy it appeared to herald. For the most part, however, nuance was established in the conference's papers through an attention to the multi-layered nature of moral discourse in global health enterprises. These overlapping, sometimes contradictory, layers were perhaps best illustrated through comparisons of the papers of the law scholars present, which dealt with the legal, political, or policy-driven rhetoric of global health, and those of the anthropologists, sociologists and historians, which gave accounts of the messy playing out of the global health initiatives on the ground, which often appeared to diverge, or even be entirely independent of the high-level rhetoric. Even within detailed empirical accounts of singular events, however, a multiplicity of the moral paradigms being enacted or referenced was evident. For example, in **Dr Sophie Roborgh's** account of the activist doctors of the 2011- 2013 Egyptian uprising, who could seamlessly slip between the roles of rock-throwing activist and politically neutral medical volunteer, tending to injuries on both sides. Collectively, this tone and these foci created detailed, realistic renderings of the complex moral landscapes of global health.

The importance of this kind of re-assertion of the complexity of moral landscapes in the face of increasingly default, reductionist models, was perhaps best illustrated by **Professor Margaret Sleebom-Faulkner's** paper on Non-Invasive Prenatal Testing (NIPT). In her paper, Professor Sleebom-Faulkner describes different countries' diverse, culturally-grounded, views of reproduction, life and disability and how they relate to particular moral concerns around NIPT and its application in screening for chromosomal abnormalities and elective abortions. The paper also tracked a blanching of these fundamental concerns with a preva-

lent utilitarian approach to NIPT policy, which Sleebom-Faulkner attributes to governments' inabilities to engage with such diversity and complexity of moral concerns within their countries. This blanching, and as such erasure, of some of the most significant moral discussions available to society is obviously of great concern. Some of the more practical unintended consequences of not properly attending to the messiness and complexity of health initiatives were captured in papers such as **Claire Wendland's** keynote address, which described the rollout of a programme in Malawi aimed at improving birth outcomes by increasing the involvement of men. Not only was the effect of this initiative on birth outcomes unclear, but in some of its manifestations appeared to create far more taxing, disempowering, and complex moral worlds for both expectant mothers and fathers. Finally, **Ruth Jane Prince' s** paper discussing the varied moral, political and economic landscapes of the Universal Health Coverage (UHC) movement in Africa, arguably the most significant burgeoning global health initiative at present, flagged the urgent need to engage with such complex moral landscapes going forwards.

There were a number of interesting papers and important discussions tackling issues outside of complex moral landscapes and consequentialist ethics. Notably, there were two panels which exclusively examined North-South global health collaborations. The papers in these panels covered topics ranging from the scientific ethos of Global South researchers whose career trajectories are largely determined by Western funding priorities (**Ferdinand M. Okwaro),** to questions of ownership and authority (**Jenny Thornton**) and the role of trust in contemporary transnational collaborations (**Angeliki Kerasidoou**), to the unwitting facilitation of foreign research projects through the performance of "deviant" research ethics by local actors (**Emmanuelle Roth**).

The conference demonstrated that whilst what might be seen as "reductionist" or simplified moral frameworks have clear utility within global health, it is crucial that they are understood in relation to complex, multi-layered, moral, political, cultural and economic landscapes. The conference also demonstrated the usefulness of a multidisciplinary approach when attempting to develop such detailed, realistic renderings of these complex environments. Whilst some disciplines might struggle to speak to each other directly, the mix of perspectives provides a multi-dimensionality to these issues which reveals significant features of these landscapes that might otherwise go unseen. These insights and the discussions that yielded them, as well as the conversations and collaborations they have subsequently seeded, would not have been possible without the support of the College of Research into the Arts, Social Sciences and Humanities (CRASSH), The Institute of Medical Ethics (IME), the **Galton Institute,** and the University of Oslo.

A copy of the program and the abstracts for all of the papers is available at: **http://www.crassh.cam.ac.uk/events/27450**

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Ancient DNA and the changing structure of the prehistoric British population: from the Mesolithic to the Bronze Age

Professor Sir Walter Bodmer, FRS
The genetic structure of the populations of the British Isles

Professor Turi King
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