

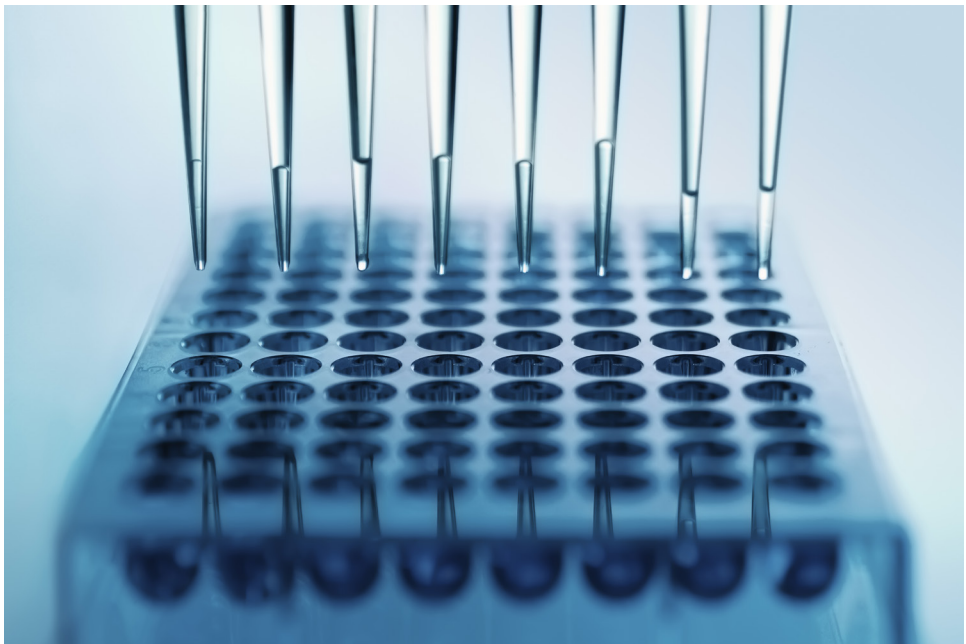
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**Galton**  
Institute

*Exploring Human Heredity*

# The Galton Review



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## EDITORIAL

Every autumn, we return to the palatial splendour of the Royal Society in St. James', London for our annual conference. Council aims to produce as varied a series of conferences as possible, so after 'Genome Editing' in 2018, this year we are learning about British ancestry in 'New Light on Old Britons'. The conference is the brainchild of **Professor David Coleman** and he has persuaded an impressive field of speakers to attend. I hope you can come. Details of the conference can be found on the back page.

Meanwhile, in June we welcomed almost 60 secondary teachers of Biology to NOWGEN in Manchester, a very modern venue, quite different from the Royal Society. This was the third of our conferences for teachers, a now biennial event aiming to update teachers' knowledge and understanding of fast-moving issues in genetics. Topics covered by a range of experts included Epigenetics, CAR-T cells and Bioinformatics. A full report can be found on page 4.

On page 7, the latest in our series 'My life in Genetics' looks at the career of our Vice-President, **Professor Dian Donnai, CBE** from Manchester. She has played such a major role in Human Genetics in this country and around the world and her story makes for absorbing reading. There are even one or two surprises.

As always, we also have reports from various events sponsored by the Galton Institute in recent months or from students who have received a travel grant. Venues could hardly be more widespread and include Cambridge, Baltimore, Warsaw and Kigali in Rwanda. I hope you enjoy reading about them.

**Robert Johnston**

**Recent Advances in Genetics:  
a Third Teachers' Conference  
26 June 2019  
Nowgen Manchester**

Once again, this was a well-attended event attracting almost 60 teachers, mainly from the North of England. The event was chaired by **Robert Johnston** and also present was the President of the Galton Institute, **Professor Veronica van Heyningen**.

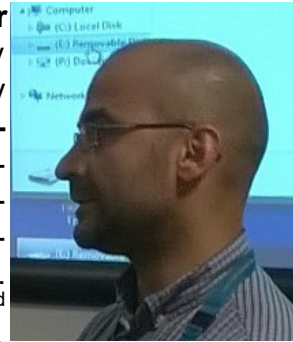
**Robert Johnston** opened proceedings with a brief account of the aims and activities of the Galton Institute and a look at its history and that of Francis Galton.

The first speaker was **Professor Andrew Read** (University of Manchester), the Treasurer of the Galton Institute. He spoke at length on **Epigenetics**, beginning with the obvious example of X inactivation. He went on to consider DNA methylation, histone modification and chromatin conformation. He believes that "if you want to understand how cells function, you must understand gene regulation and that requires an understanding of epigenetics".

**Dr Emma Woodward** (Manchester University NHS Foundation Trust) was next to speak on **Precision Genomics for Cancer**. She began by considering the spectrum of variants that can occur in a population and then concentrated on those that increase risk for breast cancer. The best known and studied are the BRCA1/2 genes which are both relatively common and, if present, cause a significant increase in risk. It is now possible to calculate a Polygenic Risk Score, enabling clinicians to decide which might be the best drugs to administer. There has been significant success with PARP inhibitors and such precision medicine is clearly the future

for treatment of such cancers.

The last talk before lunch was given by **Dr George Burghel** (Manchester University NHS Foundation Trust) who gave a lively and animated account of the **100,000 Genomes Project**. He introduced the subject by describing the Human Genome Project and thought that it “changed everything” and opened the way for genomics. The key has been the development of 2<sup>nd</sup> and 3<sup>rd</sup> generation sequencing which have revolutionised the speed of such work and



Dr George Burghel

dramatically reduced the cost. Such large-scale studies have begun to dramatically improve diagnosis and prediction of rare diseases since the variants linked to such conditions have been identified. However there are still plenty of challenging issues to address including counselling and ethical considerations.

After lunch **Dr David Thybert** (Earlham Institute, University of East Anglia) gave his thoughts on **Genomic Approaches to Studying Evolution**. He explained how Neutral Evolution and Natural Selection can both fix alleles in a population and showed how it is possible to compare genomes between and within species to identify which processes are taking place. He described his work on rodents such as the Deer Mouse and the Spring Mouse and said that studying such species is like using evolution as a “natural laboratory”. It also enables researchers to discover the molecular mechanisms behind the functioning of the genome.

**Dr Nuria Martinez-Cibrian** (Manchester University NHS Foundation Trust) spoke next on **T-cell Therapy and Cancer**. She concentrated her enthusiastic and well-illustrated lecture on the use of Chimeric Antigen Receptor T cells (CAR-T cells) to treat aggressive cancers. She described how the patient’s own T

-cells are removed, infected with retrovirus carrying the anti-CD19 gene, expanded in the laboratory then returned to the patient, all within 35 days. Success has been significant but there have also been concerns over toxicity effects which are being addressed. The two most common are Cytokine Release Syndrome and Neurotoxicity. Both can be serious but management of them is improving all the time.



Dr Nuria Martinez-Cibrian

The final speaker was **Professor Simon Hubbard** (University of Manchester) whose talk was entitled **What is bioinformatics and how do you become a bioinformatician?** He described how the science of Bioinformatics has evolved since the 1980s and had been driven by work in protein structure and DNA sequencing. Nowadays, there is so much of this work being done that “we’re drowning in data” and there is a desperate need for more bioinformaticians. Much of the work involves assembling the long chains produced so quickly by modern third generation DNA sequencing so the exons, genes, promoters etc. can be identified. Professor Hubbard described the somewhat unconventional route his career has followed and stressed that a good bioinformatician isn’t by necessity an outstanding mathematician but mustn’t be scared of statistics and should be comfortable handling data.

This was an intense day for the teachers who attended but feedback from them included: “an excellent and inspiring day”, “has helped me develop my knowledge and understanding” and “would recommend ALL Biology teachers to attend. I can’t wait for the next one!”

**Robert Johnston**

## **My Life in Genetics – an Interview with Professor Dian Donnai, CBE, Vice-President of the Galton Institute**

### **What first inspired you to follow a medical career?**

I attended a tiny girls' Grammar School in Shropshire where they didn't even offer chemistry or physics and no-one had ever gone on to study medicine. No-one in my family had been to university either so it was an unlikely beginning. However I was admitted to an orthopaedic hospital for some bone surgery when I was 13yrs old and felt the hospital environment was the most fascinating place I had ever seen. To begin with, I considered a career in physiotherapy but to be honest was a little put off by the gymnastics!

I owe a great deal to my headmistress who suggested that I think about medicine and, in addition to studying botany and zoology at the girls' school, she arranged with the local boys' grammar school for me to study chemistry and physics after I had completed O levels. I had to get from a standing start to A level in 18 months, cycle two miles each way for each lesson and promise not to use the toilets. The chemistry master even called me Charlie since he had never taught a girl and wasn't going to start then! My A level studies coincided with the discovery of the genetic code and I avidly read the *New Scientist*.



Professor Dian Donnai

I decided I wanted to go to medical school in London but my choice was limited by the application forms where most asked detailed questions about what your father did for a living (not your

mother) and whether he was a doctor, lawyer or vicar (mine was a labourer). St Mary's Hospital Medical School was the one which didn't ask these questions and so I chose to apply there and I was lucky to be offered the major entrance scholarship.

### **What was it about Human Genetics that made you move into this field?**

After medical school and more postgraduate exams, I began to specialise in paediatrics but my husband, who was training in obstetrics and gynaecology, first got a post in Sheffield then Manchester and we eventually had 2 children so my career was interrupted and I did short term paediatric and general practice jobs.

However, I was always interested in finding the underlying causes of disease and congenital malformations. Two cases stick in my mind – a baby born at St Mary's in London who died shortly after birth with multiple abnormalities. Usually such babies had an autopsy but little else but I pushed for more investigations and arranged for a skin biopsy to be taken and the cells cultured at Guys Hospital for cytogenetic studies which revealed deletion of part of the short arm chromosome 4 and was reported in the literature as the second case ever of Wolf-Hirschhorn syndrome. I also remember the professor of paediatrics in Sheffield not believing it was me (a locum in the casualty department) who had diagnosed a case of C1 esterase inhibitor deficiency on the basis of the clinical signs and proven it by further investigations.

There were no clinical training posts in genetics in the mid-1970s so I applied in Manchester for a post advertised as 'married woman wanted for clinical assistant sessions in genetics' – they wouldn't get away with that now! At first I worked two half days a week but after a year three national training posts in clinical genetics were established in England and Wales, and Manchester was allotted one to which I was appointed. As soon as I started in genetics, I knew I had found 'my home' in medicine and feel so lucky to have been in the specialty from early on.



## **How much has the subject changed in your time?**

The short answer is that scientifically genetics has changed out of all recognition but for the patients and families the problems and worries are the same. I see myself primarily as a physician for families with rare diseases. They want to know what the problem is, why it happened, whether there is anything that can be done about it and whether it will happen again. Clinical Genetics is very much multidisciplinary team work. It is also a very international specialty since we deal with families with extremely rare diseases.

When I first started in the specialty many disorders could not be diagnosed and hadn't even been described or delineated. At first, in addition to seeing patients in my clinic, I collaborated with others to describe syndromes which hadn't been written about in the literature. These then became the focus for research using new technologies to try and identify the underlying genetic mechanisms. Gradually in the UK, genetic centres were established in each region and, from the handful of people in the 1980s now the Manchester Centre for Genomic Medicine employs around 300 clinicians, genetic counsellors, nurses, scientists, bioinformaticians, researchers and administrative staff.

With modern technology, around half of rare conditions affecting growth and development of children can now be diagnosed but there is still a great deal to be discovered. The new knowledge about underlying genetic causes has opened up the possibility of treatments in some cases and clinical trials are in progress for an increasing number of genetic disorders. Because my field of work concerns developmental disorders in children I haven't mentioned genetic diseases of adult life, but progress is similarly rapid in cancer genetics, cardiac genetics and neurogenetics.

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

As well as my own subspecialty of dysmorphology (development disorders and congenital malformations) I have had a particular interest in public engagement and involvement in genetics since I

feel it is vitally important that everyone knows about the scientific possibilities as well as being able to participate in the debate about how advances should be utilised and the ethical issues that may result. In the early 2000s following the genetics White Paper 'Our Inheritance, Our Future' we developed one of the so-called genetics knowledge parks in Manchester (Nowgen) which focussed on the non-technical aspects of genetics; health economics, public engagement as well as teaching and training to engage other medical specialists – the beginning of 'mainstreaming'. I also recognised the importance of strategic developments and engagement with politicians and policy makers and have sat on many committees both in the UK and internationally.

### **Who have been the greatest influences on your work?**

I have been fortunate to interact with many eminent doctors and scientists in my career, but my values as a doctor were shaped by Professor Tom Oppé, the professor of paediatrics at St Mary's Hospital in London who taught me the value of listening to parents and taking account of their lives outside the hospital and the importance of research to improve knowledge and treatments. In genetics in Manchester, I've had a long and productive professional relationship with Professor Andrew Read; we have had many research and teaching collaborations and are now writing the 4<sup>th</sup> Edition of our text book *New Clinical Genetics* (Scion Publishing Ltd).

### **What do you think will be the greatest challenges for Human Genetics in the coming years?**

I will limit myself to the clinical applications of genetics where there are plenty of challenges! Whenever a discovery is made there are endless promises of how it will result in better treatments but of course this does not apply to all conditions and indeed it is hard to see how many of the developmental disorders in children which affect the structure of the brain and other organs could ever be treated in a curative sense. Therefore one big challenge is managing parents' expectations. However understanding the genetic basis of disease and the pathways involved are powerful tools which

may help effective clinical management of complications such as epilepsy for example. For some conditions though, such as cystic fibrosis, spinal muscular atrophy and muscular dystrophy, knowledge of the precise genetic variation has already led to effective and curative treatments for some patients. The problem here is the cost and it is the role of the clinicians in combination with patient organisations to engage with health providers to argue the benefits of such revolutionary treatments. The same arguments also apply to genetic-based cancer and neurological treatments amongst many others.

### **What do you think is the main role of the Galton Institute?**

The Galton Institute has an important role in public engagement and education in all aspects of genetics. The annual flagship Galton Institute Conferences held at the Royal Society with their very diverse audiences and sheer breadth of subject matters are rightly extremely popular. Topics have included *Genetics in Medicine*; *Environmental factors in gene regulation* and *Surveying Galton's Legacy*. Other initiatives include a series of booklets written for lay people on a variety of topics largely relating to applications of genetics in medicine and several very successful courses for teachers on modern genetics and genomics. The GI also supports young scientists with travel grants and small conferences where the topics are relevant and where support is needed to make the conferences as accessible as possible to younger attendees.

### **Finally, please tell us one thing about yourself that is not widely known and may surprise people.**

When I was a medical student, to earn some pocket money, I was a film extra. Several of us took part in films being made at Pinewood and Shepperton Studios. Our pay was around £4 per day but we were provided with lunch which was a bonus. We could also earn an extra 50p for special action which meant you were on camera for a longer time. I was on screen with Sophia Loren and Gregory Peck in *Arabesque* and with James Robertson Justice and Hattie Jacques in *Doctor in Clover!*

## **African Society of Human Genetics Conference held in conjunction with the 12<sup>th</sup> H3Africa consortium meeting**

September 2018 - Kigali Convention Centre, Rwanda

This event's objective was to bring together brilliant scientists from around the world to share their research while promoting young researchers to continue to promote human genetics in Africa. The main theme for the conference was focused on "*Building skills and resources for genomics, epigenetics and bioinformatics research for Africa*".

The conference gathered together 435 participants from 38 countries and was officially opened by the Rwandan Hon. Minister of Health, Dr Diane Gashumba. There were tributes to two eminent geneticists who recently passed away, Professors Bongani Mayosi and Luigi Luca Cavalli-Sforza.

There were two plenary sessions, one on Human Genome Project (by **Eric Green**, NIH) and another one on genetic and epigenetic modulation of the acute chest syndrome in sickle cell disease (by **Solomon Ofori-Acquah**, Ghana). There were 9 conference sessions including sessions on 1) African population genetics; 2) Bioinformatics training and young researchers; 3) H3Africa consortium showcase; 4) Epigenomics and functional genomics; 5&6) Genomics of rare diseases (Part I & II); 7) Host genetics and infectious disease; 8) Ethics and genomics in Africa; and 9) Complex disease genomics. Invited speakers included prominent researchers in all these areas including **Neil Hanchard, Lucien Koulischer, Nicola Mulder, Guida Landoure, Michele Ramsay, Christian Happi, Jantina de Vries, Elizabeth Marincola, Monica Uddin, Youssef Idaghdour, Jacob Souopgui, Caroline Tiemessen, Marlo Moller, Aime Lumaka, Rhokaya Ndiaye, Paulina Tindana, Adebawale Adeyemo, Anatole Laleye**, and **Dezheng Huo**. The format of the conference was

a series of presentations, both oral and poster by young and established scientists. A total of 138 abstracts were submitted and after being reviewed and graded, 134 were selected. Of these, 23 were oral presentations and the remaining 111 were poster presentations.

The conference was a great success and after all the sessions, young researchers had a panel discussion with the senior researchers where they were advised on how to enhance their careers by:

- (i) Reaching mentors
- (ii) Never giving up, keep trying, no matter what
- (iii) Keep applying for grants
- (iv) Find anything that can be a research project
- (v) Realizing that a dream will enhance our work
- (vi) Stick with your dream and try to achieve it
- (vii) Your mentor does not always have to be a physical person, but may be inspiring books that can guide you to reach your dreams

The following are some of the areas that were also discussed: (i) Building capacity for pharmacogenomics; (ii) Opportunities to showcase younger scientists to motivate others; (iii) Opportunities for training and volunteering; (iv) Empowering more women to speak out on their views; (v) Organizing a conference specifically for students to give them opportunities to address their projects; and (vi) Grants and sponsorship for the younger generation.

In addition, two successful panel discussions occurred: “Translational genomics” facilitated by **Debo Adeyemo, Ghada Youssef El Kamah, and Ambroise Wonkam**; and “Strategic Planning” for the AfSHG including resources for genomics, epigenetics and bioinformatics research for Africa facilitated by **Charles Rotimi and Sonia Abdelhak**.

At the end of the conference, the best presentations given by young researchers were evaluated by a committee and winners

were selected for awards which included 10 subscriptions to *Nature Genetics* generously offered by the Chief Editor (Myles Axton), books and four full scholarships to attend the 2<sup>nd</sup> Winter Institute in Statistical Genetics at New York University Abu that will take place in January 2019 (offered by Youssef Idaghdour).

We are very grateful for financial support from the **Galton Institute** which supported the attendance of two African early career researchers; one fully supported for oral presentation and another one partially supported for a poster presentation at the AfSHG 2018 meeting.

**Leon Mutesa**  
**Chair of Local Organizing Committee**

### **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/>

**British Society for Population Studies Conference  
10-12 September 2018, University of Winchester**

In 2018, BSPS welcomed the participation of the Netherland Demographic Society (NVD). Members of NVD organised two of the sessions and arranged the opening day plenary. BSPS is particularly grateful to **Leo van Wissen** for facilitating this initiative.

Over the course of the two days, 162 submitted papers were presented across six simultaneous sessions. Additionally, there were training sessions on Popgroup, the three UK Census Longitudinal Studies, and an introduction to handling large datasets in *R*. An early career mentoring session in a speed dating format proved very popular. The usual lively poster session saw joint winners. The student poster prize went to **Rishita Nandagiri** from LSE, for *"They Know everything" the role of Community Health Workers in abortion access* and the senior prize went to **Dariya Ordanovic** from ESRI Spain, **Diego Ramiro-Fariñas** from the Spanish National Research Council (CSIC) and **Francisco Vici-ana** from Junta de Andalucia for *Spatial variations in mortality due to heat waves in southern Spain*, a very topical subject.

Abstracts of all papers presented can be found on the BSPS website at:

<http://www.lse.ac.uk/social-policy/research/Research-clusters/british-society-for-population-studies/annual-conference>

Plenary sessions were from **Professor Renske Keizer** (Erasmus University) & **Professor Danny Dorling** (University of Oxford). Reports of these plenaries follow:

**Plenary 1:** Professor Renske Keizer (Erasmus University) - *Father's role in the development of children's diverging destinies: families, inequality and social policies*

Professor Keizer outlined the research interest in the role of the father in the intergenerational transmission of inequality with illustrative examples from her ongoing study of parent-child play in the Dutch context. Professor Keizer opened her talk with the example of the income gap in children's standardised reading scores and noting that research interested in the intergenerational transmission of these differences has tended to focus on the role of the mother, but with the scarce evidence available on the role of the father suggesting an even larger educational gradient for fathers than for mothers. She pointed out that the scarcity of evidence is startling considering father involvement has become more polarised by social class, suggesting that fathers (can) play a substantial role in development of children's diverging destinies. Professor Keizer further argued that understanding the role of father involvement in the process of intergenerational transmission could inform policies aimed at weakening the intergenerational transmission of social inequalities.

Research interest in fatherhood is relatively recent and Professor Keizer argued that traditionally, research had seen mothers as the main carer of children and fathers primarily as the family provider, with the associated assumption that fathers would be less reliable sources of research information regarding their children. Two societal developments explain the more recent increase in attention to fatherhood. Firstly, the increase in divorce rates and the related concern about the difference in child outcomes between 'intact' and 'fatherless' families. This meant that more researchers interested in the outcomes of children of separated parents began to include fathers with shared care or contact in their research design and data collection. Secondly, the increase in women's employment outside the home has led to increased societal acceptance of involved fatherhood.

Professor Keizer noted that her study's research interest is not only in documenting child outcomes but also in understanding the mechanisms underlying the influence of involved fatherhood on child development. She highlighted four mechanisms of how fa-



thers influence their children's development, interacting in ways that were different from mothers. These were: 1) 'rough and tumble' physical play, 2) socialization, 3) signalling danger, and 4) language. As examples, she mentioned that while mothers tend to engage in imaginary play with their children, fathers tend to engage in more physical play, and that fathers tend to use more advanced language in speaking to their children than do mothers (more complex sentences, posing open questions etc). However, Professor Keizer stressed that it is difficult to disentangle whether these are sex, gender or role differences. In addition, while evidence suggests that father involvement is linked with positive child development, there are strong selectivity effects with increasing polarisation as higher educated parents tend to have more stable relationships and careers, while those with lower levels of education have higher divorce/separation rates and more precarious employment, which can constrain the time available for father-child interaction. Thus, the benefits from father involvement in parenting might have increasingly become a higher social class privilege.

In the final part of her presentation, Professor Keizer showed some illustrative video clip examples from her own ongoing research focused on the role of play in transmission of inequality. The research involved observations of parent-child interactions as well as questionnaires with a total of 100 families with children aged 3 years old in the Netherlands, including both higher and lower educated parents. Showing three video clips from the study, each of a parent who had been instructed by the researcher to play with their child without using toys, Professor Keizer highlighted both the gender and the social class differences found in the study. While in the first clip, a highly educated mother suggested a game of hide and seek with her child, in the clips of father interactions both the more highly educated father and the father with lower levels of education engaged in very similar physical 'rough & tumble' play. Based on the observational data, Professor Keizer suggested that the social class differences were smaller than might have been expected from the literature, but

that the questionnaire and interview data suggested that the frequency of such father-child interactions was higher among the higher educated. Highlighting that in the Dutch policy context where the 1.5-earner model is common, the relatively short parental and paternity leave, and with parental leave not arranged by law but by collective agreements and largely unpaid, Dutch policy increases social class differences in time spent on involved fathering, because not all can afford to take leave in order to be more involved. Professor Keizer concluded that while the social context for parenting/father involvement differs between countries, the conclusion from the talk was that the policy challenge is to create attractive policy measures for lower income families which encourage father involvement in order to potentially weaken the intergenerational transmission of social inequalities.

**Plenary 2:** *Linking mortality to the past: solving the geographical problems*– **Professor Danny Dorling** (University of Oxford)

From the outset, Dorling acknowledged that he would not be directly answering the questions set out in his abstract regarding UK and sub-national mortality rates. Instead his 2018 talk would be aiming to reach out to an early career researcher who would engage with the issues outlined and continue to build on existing research in this field. He also took time to acknowledge the contributions made by several researchers to recent research projects on which he had worked.

Throughout the talk, audience members were invited to consider thought-provoking graphs, charts and images. Dorling first focussed on issues surrounding caring, support and possible contributions to mortality and showed the audience a graph comparing the Local Authority District life expectancy gap during the time of the recent Labour government to today's government. Dorling suggested a sequence of events had taken place from the late

2000s onwards that had an impact on this, including the cutting of support for older and disabled people. He also asserted that if an institution blames 'flu, as was done between December 1952-March 1953, they tend to continue to use it as an excuse and contrasted the blaming of 'flu from 2013-March 2018 with a chart of known flu epidemics from 1969/70-1999/00.

In 2016, the Office for National Statistics released their annual mid-year estimates and Dorling commented that no-one noticed that 52,000 more people had died, asserting that whilst death certificates increasingly record the cause of death as dementia and Alzheimer's, 'flu is recorded less often.

Dorling also turned his attention to infant mortality rates, noting that whilst Scotland now has a lower infant mortality rate than England, the UK as a whole has dropped in the neonatal mortality rankings of European Union countries from a rank of 7 in 1990 to 19 in 2015. He suggested a need to look at the provision of midwives and maternity services and noted that at present around 30% of midwives in London are European Union but not UK citizens. After touching upon the impact public transport and infrastructure can have upon public health, Dorling stated that he thought that in the future we'll look back on this period and the issues he had discussed like we do employment in the 1980s and will question the regional variation in health outcomes and mortality rates.

Thanks to Jenny Chanfreau (LSE) and Sarah Garlick (University of Liverpool) for the plenary reports. Thanks are also due to the **Galton Institute**, who again awarded a grant to BSPS which helped to cover some of the Conference expenses. As ever, BSPS is very grateful to them.

**BSPS Secretariat**

## **Genetic Alliance UK Annual Conference 2018**

25 September, 2018 at Amnesty International, London

This important event saw 180 people from the genetic field come together for thought-provoking presentations, practical workshops and Q&A sessions. For the first time the event was also streamed live to those not able to attend in person, and interactive software was introduced to increase audience participation. The event attracted 55 patient organisations representatives, 8 researchers and HCP, 14 industry representatives and 10 policy makers.

With the greatest number of people in attendance, and more contributing and joining online – this was the most successful Genetic Alliance UK Annual Conference to date. Presentations covered a variety of issues and topics that affect the patient and wider genetic, rare and undiagnosed community.

We welcomed **Professor Dame Sue Hill**, Chief Scientific Officer for England, who gave an update to the community on *'the implementation of genomics in the NHS'*, followed by **Professor Jill Clayton-Smith**, Consultant Clinical Geneticist and leader of the EU Network for Rare Congenital Malformations and Intellectual Disability on *'how the implementation of genomics is changing the relationship between clinicians and their patients'*. **Dr Amy Hunter** and **Rosa Spencer-Tansley** from Genetic Alliance UK presented their stark findings from their research project: *'Living with a rare condition: the effect on mental health'*. Three patient organisations, gave presentations on the current challenges that each faces as an organisation. **Clare Dickson for ALD Life**, **Jan Fowler for SoftUK** and **Aisling McMahon from the Cystic Fibrosis Trust** all spoke and then took part in a lively panel session discussion with the floor and those online. All of these presentations were followed by lively discussions and Q&As.

Following a networking lunch and Genetic Alliance UK's AGM where the charity's new strategy was launched, delegates were invited to choose to attend two of the five scheduled workshops. These included:

- (i) Building a strong case for support – run by the Genetic Alliance UK fundraising team.
- (ii) Do you need a patient registry? – run by **Mary Bythell**, Head of Rare Disease Registration, Public Health England, **Jeanette Aston**, Rare Disease Data Liaison, Public Health England, **Angela Stringer**, Duchenne Muscular Dystrophy Registry Curator from Action Duchenne and **Rebecca Cosgriff**, Registry Lead, Cystic Fibrosis Trust.
- (iii) Producing patient leaflets and information - **Dr Claire Andersen**, Information Officer, Unique, **Ciaran Scott**, Clinical Trials Officer, AKU Society and **Nick Sireau**, Chairman of Trustees, AKU Society.
- (iv) Managing online communities - **Lauren Roberts**, Director of Support, Genetic Alliance UK
- (v) Working with the media - **Vivienne Parry OBE**, Head of Engagement, Genomics England

Feedback from the conference demonstrated how useful the day-long activity was with 93% feeling better informed, 92% having a greater feeling of being part of the patient community and 91% feeling more empowered to support their community.

The conference was supported by the **Galton Institute** and Genetic Alliance UK, and the patient organisations they represent, are incredibly grateful.

**Emma Damian-Grint**  
**Director of Fundraising and Communications**

### **Grants for conferences and workshops**

The Galton Institute makes awards of up to £1000 to help meet the cost of organising and running conferences or workshops on topics relevant to the Institute's aims. We will under special, exceptional circumstances increase funding up to a maximum of £2,000, if the request is well justified.

Full details of the grants can be found on our website at:  
<http://www.galtoninstitute.org.uk/grants/grants-for-conferences-or-workshops/>

## **5<sup>th</sup> International Conference of the Polish Society for Human and Evolution Studies**

**24-26 September 2018, Warsaw**

This conference was held at the Centre of New Technologies of the University of Warsaw. It was jointly organized by the Polish Society for Human and Evolution Studies, Museum and Institute of Zoology of the Polish Academy of Sciences and the Centre of New Technologies of the University of Warsaw. The whole conference (including all talks, posters and discussion panel) was held in English.

There were 69 attendees registered to the conference. 42 of them were women and 27 men. 46 attendees were affiliated to Polish institutions, 14 came from the Czech Republic, 3 from Germany, 3 from Russia, and 1 each from Israel, Hungary and United Kingdom. 55% of the attendees were students in Masters and Doctoral programmes.

During the three days of the conference there were four plenary talks given by the world experts in their fields. In reproductive ecology. **Professor Grazyna Jasienska** spoke on "Evolutionary perspective on women's reproduction and health", in human ethology **Dr Jan Havlicek**, on "MHC, mate choice and reproduction", in cultural evolution **Professor Russell Gray** on "The cultural evolution of religion" and in molecular anthropology **Dr Wolfgang Haak** on "Europe's genetic prehistory".

There were also twenty two regular talks covering a wide range of topics such as perception of traits in mate choice and social behaviour contexts, origin and mechanisms of rituals and moral judgements, evolution of language and music, reproductive strategies in primates and many more. There were also seventeen post-

ers presented throughout the conference with a dedicated poster session during the first day of the meeting. All student talks and posters were subjected to competition for two awards. The vote was cast by the conference attendees on the last day. The best student talk award went to Yitzchak Ben Mocha for his “Humans conceal sex across cultures. But why?”, while the best student poster award went to Aleksandra Poniewierska, Aleksandra Szczepańska and Mateusz Kościcki for “A synthetic view of the origins of language”.

On the second day of the conference a discussion panel was held on public outreach and science communication on subjects concerning human evolution and the associated challenges in times of fake news and pseudoscience. The panel was titled “Talking ‘bout evolution sounds like a whisper” and was led by **Konrad Talmont-Kaminski** – a cognition researcher. The panellists were **Bogusław Pawłowski**, the president of the Polish Society for Human and Evolution Studies, **Barbara Pietrzak**, a member of the Spokesmen of Science initiative, and **Michał Misiak**, a doctoral student in evolutionary psychology..

Thanks to the support from the **Galton Institute** we were able to invite three distinguished international plenary speakers. The support was acknowledged during the conference opening with the presentation of a slide outlining Galton Institute’s mission.

This conference was a great opportunity for researchers from seemingly distant scientific fields to meet and exchange ideas during the scientific sessions as well as during the evening social events of the conference. Such an interdisciplinary environment is always welcome.

More about the conference can be found on its website:  
[www.ptnce2018.pl](http://www.ptnce2018.pl)

**The Galton Institute collaborates with the Genetics Society to provide support towards three travel bursaries each year which are organised and administered by the Genetics Society. These grants are available to students working towards a PhD on a topic relevant to the aims of the Institute. We print below three reports received from grant recipients.**

**Grant report for Junior Travel Grant  
Lucy Dunbar**

In February 2019, I attended the 42nd annual Association for Research in Otolaryngology (ARO) Midwinter Meeting, held in Baltimore, Maryland. The meeting aims to bring together over 2000 students, researchers, clinicians and audiologists to communicate the latest developments in the auditory field. With dozens of talks, countless posters and numerous networking and social events crammed into four and a half days, there was a constant buzz of activity.

I was given the opportunity to present my DPhil research in the form of a poster, which took place during the 'Genetics of Hearing Loss' session. My project aims to characterise a mouse mutant with a severe, early-onset and progressive hearing loss phenotype, which is caused by a mutation in the *Cln2* gene. The poster not only represented the first time I have been invited to present



my research at a conference, but also the first time that the *Cln2* gene has been reported in relation to mammalian hearing loss. Although I stood by my poster for several hours, time flew by and I had a number of helpful discussions with various attendees who wanted to ask questions, suggest future ideas for my research and generally learn more about this novel deafness-related gene. Interestingly, a podium presentation later on in the meeting also focused on the role of *Cln2*, but this time in zebra fish. I felt that, by discussing the same gene in different model systems, my poster and the presentation complemented each other well and have furthered our understanding of the role of *Cln2* in the auditory system.

The data I presented on my poster is currently being put together into a manuscript that our team hopes to publish this summer, working in collaboration with researchers based in Europe and the US. As many of our collaborators and co-authors were also present at the ARO meeting, we had the chance to get together between sessions to discuss outstanding questions and our future research plans. Additionally, it was great to be able to meet in person, as opposed to just communicating exclusively via email!

Due to the size and nature of the meeting, the presentation and poster sessions covered a broad range of topics including the auditory cortex, speech perception, tinnitus and hair cell regeneration. Additionally, there were plenty of sessions relevant to the genetics of the mammalian auditory system, with interesting talks by **Jonathan Bird** and **Leonardo Andrade** on auditory hair cell development and maintenance. Both of these were informative and relevant to both my own research and that of my wider lab group at MRC Harwell. Additionally, the presidential symposium that opened the meeting, given by **Professor Karen Steel** from King's College London, highlighted the increasing prevalence of age-

related hearing loss and the need to address this.

After talks had finished for the day and posters had been browsed, the activities carried on into the evening with various networking events, workshops and talks by special guests. One such event that stood out for me was the science communication workshop, in which attendees were filmed giving an 'elevator pitch'-style talk describing their research to a non-specialist. We received feedback on our talks and filmed them repeatedly to become comfortable with communicating our research – something that will inevitably become useful when talking to members of the public, scientists in different fields and potential collaborators in the future. A less challenging evening event was the spARO social – a gathering organised for student and young post-doc attendees to meet in an informal setting away from the conference venue. The evening proved very popular, as it gave those of us new to the field (and indeed academic research in general), a chance to chat about everything from our favourite talks and posters of the day, to opinions of life in academia and our plans for the future.

In summary, this meeting was the perfect place to learn about the latest developments in auditory research, build relationships with other researchers and, importantly, to present the hard work that my team and our collaborators have put into our upcoming manuscript. I would like to express my gratitude to the Genetics Society and the **Galton Institute** for making this valuable trip possible. Thank you so much!

**Lucy Dunbar**  
**Medical Research Council Harwell**

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## Grant report for Junior Travel Grant Pro Chatzikyriakou

I attended the *Conference Systems Epigenetics Event* which is a relatively new, four-day meeting and provides a platform to scientists whose main focus is basic and biomedical research in the field of cancer epigenetics, bioinformatics and system biology. Present were many distinguished scientists in the field of epigenetics who shared the latest findings from their labs.

**Professor Geneviève Almouzni** started the conference giving the EMBO Keynote Lecture presenting and summarising data on histone variants and chaperones in relation to cancer. **Professor Pamela Munster** from UCSF spoke about the interplay of epigenetic regulation and immune system and how this can be harnessed in cancer immunotherapies. **Professor Martin Fussenegger** from ETH Zurich talked about the fascinating topic of metabolic prostheses that can manipulate cell metabolism while **Professor Jonathan Chubb** of UCL explored transcriptional bursting as a means of regulating gene expression. The meeting had a very good balance with many other scientists presenting their most up-to-date findings. Many researchers presented novel techniques for probing the epigenome while new analytical and bioinformatic techniques were highlighted. This was a good opportunity not only to see what is happening in other labs around the world at the moment but importantly to have a taste of where the field is going in the future.

The conference had a high number of delegates presenting their data in poster form. The two poster sessions were very busy and led to interesting discussions. As part of it, I had the chance to present my poster which described my findings on the *DNA methylation of pheochromocytomas and paragangliomas* and was se-

lected for a flash presentation as well. During the sessions, I received incredibly valuable feedback and suggestions from delegates. This also provided the opportunity to establish a new collaboration that will provide complementary data to further understand the degree of epigenetic deregulation in this disease.

Illumina hosted a very interesting workshop on the utility of their methylation array technologies in cancer research. As part of this, **Professor Andreas von Deimling** from DKFZ talked about classifying tumours based on the levels of promoter methylation and how this can be used for patient stratification and to inform treatment decisions, exemplified by MGMT promoter methylation. **Professor Manel Esteller** of IDIBELL explored the use of the same technology in cancer research and the applications in clinical oncology, including how it can be employed in carcinomas of unknown origin to identify the primary tumour.

The meeting had a very rich non-scientific part as well. The first day ended with the theatrical performance 'Epigenetic Human Simulation' followed by a panel discussion on gender and diversity in science. This was designed to provide a forum where the challenges that minority groups in science face can be addressed. Delegates were invited to engage in the discussion, sharing their experiences and ways to support and encourage everyone in the science community to have equal opportunities. On a different day, another open discussion event took place, this time on business ethics in research. Guests from industry and academia, along with invited social scientists, shared their views on ethical issues arising from the interchange between innovative research findings and the market.

The organisers had planned a very busy social programme. Each day they offered a selection of tasty local delicacies and at the end of the day drinks provided an opportunity for delegates to wind down discussing the highlights of the talks and network. Overall, this was a very successful conference that covered a variety of

cutting-edge topics in epigenetics, systems biology and cancer. I am incredibly grateful to the Genetics Society and the **Galton Institute** for this award that allowed me to attend this meeting, present my data, receive useful feedback and expand my network of peers sharing similar interests. I highly recommend the conference to anyone who is interested in systems epigenetics, cancer and precision medicine and I hope I will meet some of you next year!

**Pro Chatzikyriakou**  
**King's College London**

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### **Grant report for Junior Travel Grant** **Milan Muso**

This year's Diabetes Keystone meeting brought experts from around the world to the mountain village of Whistler in Canada. Walking into the local conference centre, with its huge gates and majestic stone fireplace, felt akin to walking into great hall of Moria from Tolkien's Middle-Earth, with a sense of the importance of the upcoming event.

With around 10% of people diagnosed worldwide, diabetes has reached epidemic proportions and is still not fully understood. In his keynote lecture, **Mark McCarthy** introduced the complex task of deciphering over 400 loci identified by genome-wide association studies (GWAS) to identify the underlying mechanisms of diabetes. To me, the most promising solutions to this problem included **Melina Claussnitzer's** high-throughput screens of non-coding regulatory elements and the novel "GWAS in a dish" approach for effectively identifying causal genes and risk variants.

A major theme of the meeting was the heterogeneity of diabetes. Whereas **Leif Groop** presented his work identifying up to 5 distinct subtypes of diabetes, Mark McCarthy stressed the continuity of these subclasses using the “palette-model” of complex disease. **George L King** showed how inter-individual differences in the susceptibility to diabetic complications can be used to reveal protective mechanisms and **Lee M Kaplan** focused on the heterogeneity of response to therapy. Other themes included the epigenomics of diabetes, with **Maike Sander** showing how fasting remodels  $\beta$ -cell chromatin, and the discussion of new potential drugs, such as tissue-specific and long-acting insulins, and the exciting concept of a glucose-responsive insulin. Thought-provoking findings were presented on the effect of insulin in the brain.

In my poster entitled “*Testing the WARS2 gene and its 3’UTR SNP as functional candidates for waist-hip ratio regulation*”, I demonstrated our approach to trace GWAS loci to their functional mechanisms. I received many suggestions for future experiments and discussed both the limitations and greater implications of our findings. The less formal afternoon programme, that the Keystone meetings are well regarded for, allowed me to discuss broader scientific questions over lunch and during a long walk in the beautiful Canadian countryside.

I am extremely grateful to the Genetics Society and the **Galton Institute** for funding me to attend this Conference. I have gained new insights for both my project and metabolic disease in general, met many great researchers and had my first experience of presenting at an international conference. Last but not least, the meeting helped me to clarify my thoughts about the future directions of my postdoctoral research.

**Milan Muso**  
**University of Oxford**

**Call for funding proposals  
for  
Grants from the Artemis Trust of  
The Galton Institute**

The Artemis Trust is wholly owned by the Galton Institute. It was established in 2016 and evolved from the Birth Control Trust, founded in 1977, which had extremely narrow objectives. The wider ranging objectives of the Artemis Trust are:

- to help preserve and protect the physical and mental health of people, particularly but not only those from poorer communities, in particular by:
- Assisting in the provision of fertility control and other measures to improve reproductive and sexual health; and
- Advancing education in all aspects of reproductive and sexual health.

**Maximum Grant: £15,000 p.a.**

**Details and application form:**

**[www.galtoninstitute.org.uk](http://www.galtoninstitute.org.uk)**

# **Galton Institute Conference 2019**

## ***NEW LIGHT ON OLD BRITONS***

**The Royal Society  
Wednesday 30 October, 2019**

### **SPEAKERS:**

**Professor Nick Ashton:**

The climate, palaeogeography and early human settlement of Britain over the last million years

**Professor Ian Barnes and Dr Selina Brace**

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

**Dr Lara Cassidy**

The genomic history of Ireland

**Professor Sir Barry Cunliffe, CBE, FBA**

The 'Celts' in Britain – a romantic fiction?

**Professor Turi King**

Genetics and history: how DNA can be used as a window onto the past

**Professor Chris Stringer, FRS and Dr Silvia Bello**

The First Britons: bones and behaviour

*Admission is free but strictly by ticket from:*

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