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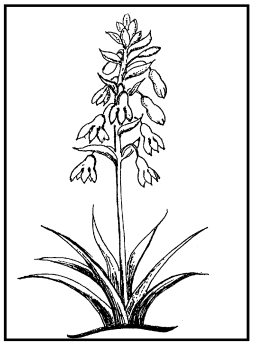

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The Galton Institute

NEWSLETTER

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Contents

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Galton Institute Conference 2014

Genetics in Medicine

Held 4 November, 2014 at the Royal Society in London

This year's Galton Institute Annual Conference was dedicated to Genetics in Medicine and covered diverse topics, such as genetic testing and gene therapy, as well as the application of cutting-edge genomic technologies to cancer and metabolic diseases. It was an inspiring overview of the current application of genetics in clinical care today.

The first session, chaired by Professor Dian Donnai, started with an inspirational talk from **Professor Sir John Burn** (Newcastle University). He is medical director and head of the Institute of Human Genetics, and lead clinician for the National Health Service North East.

In his lecture, entitled ***Overview of Genetic Medicine,*** Professor Burn succeeded in walking us through genetics, and its transformation and 'evolution' into human genetics, then clinical genetics, followed by genetic medicine and more recently into genomic medicine. He reminded the audience that it was indeed Galton himself who in 1889 introduced the concept of regression to the mediocrity (mean) to become the core of regression analysis, and also the idea of polygenic inheritance and mathematical relationship between genotype and phenotype. Galton used the example of height, and the genetic advances to date have clearly demonstrated that over 180 genetic variants are involved in the genetic makeup of human height.

Professor Sir John Burn

Professor Burn then talked about the advance in the predictive markers for risks of developing genetic diseases. The example of the genotype-driven anticoagulant Warfarin dosing, where

recently published results from a randomized trial achieved successful genotype-guided dosing based on 3 individual single nucleotide polymorphisms (SNPs).

He envisaged the future of genetic medicine and talked about the application of nanotechnologies to DNA testing and presented work close to his heart; the development of a Nanowire DNA detection chip, applying nanowires, (metallic or semiconducting particles) in pioneering novel assays for fast, accurate and affordable DNA testing. These exciting new technological advances will ensure accessibility of genetic testing technologies through the world, not restricting it to the world's richest countries.

Professor Andrew Hattersley, FRS (University of Exeter) followed with his lecture on ***Using Genetics to improve care in Diabetes***, presenting his research on the genetics of diabetes and translating it into clinical practice.

Professor Andrew Hattersley, FRS

He began by talking about work from his laboratory on gene discovery success in neonatal diabetes (NND). Within NND, 45% of these diagnoses represent a transient form, 45% permanent diabetes, and 10% are due to syndromic pancreatic dysplasia.

Together with Professor Frances Ashcroft's team in Oxford, they were able to demonstrate that heterozygous activating mutations in the gene KCNJ11, encoding the Kir6.2 subunit of the ATP-sensitive potassium (KATP) channel cause neonatal diabetes. They hypothesized that in the patients carrying these activating mutation, the channel could be closed by an ATP-independent mechanism (e.g. by sulfonylureas) and insulin secretion might be restored. Indeed, when sulfonylureas in tablet form replaced insulin injections in these patients, they achieved far better glucose control, which also had a lasting effect. This represents a true personalized therapy for ~50% of the patients. This also led to change in the current guidelines for diabetes testing for patients <6months of age. This testing is now offered in >70 countries.

Professor Hattersley discussed the implications of the fast adoption of genetic diagnostic tests, also leading to new gene discoveries and genetic stratification of diabetes. He described the move from a traditional scenario, where selective late testing of individuals genes based on clinical features is being replaced by an early non-selective investigation, involving comprehensive genetic analysis of multiple genes by next-generation sequencing (NGS). This change in the paradigm for genetic testing is already bearing fruits as novel genetic discoveries further the mechanistic insights into diabetes and pancreatic function.

In the second scientific session, chaired by the Galton Institute former President, Professor Sir Walter Bodmer, FRS, we heard lectures by two distinguished female clinical researchers, Professor Nazneen Rahman (The Institute of Cancer Research) and Professor Sadaf Farooqi (University of Cambridge).

Professor Nazneen Rahman

Professor Nazneen Rahman, talk entitled ***Genetics in cancer and treatment***, presented her work in using science to make a difference for the cancer patients.

She described the two types of cancer genes: genes carrying somatic mutation, and the cancer predisposition genes, where germline mutations confer highly or moderately increased risks of cancer. There are over 100 cancer predisposition genes (CPGs), explaining 2-3% of cancers, and they have clear clinical utility, including improved diagnostics, familial cancer prevention, and optimized tailored therapies. In the UK, the National Health Service only tests for half of these genes, and access is restricted. In 2013 a research programme, called Mainstream Cancer genetics (MCG) programme, generously funded by the Wellcome Trust and directed by Professor Rahman, was initiated to

lay the foundations for an accessible genetic testing for anyone with cancer.

This initiative is a joint venture between The Institute of Cancer Research, The Royal Marsden Hospital, the Wellcome Trust Centre for Human Genetics (Oxford) and Illumina.

The major aims are identification of the CPGs in an accredited NGS testing laboratory setting, making the genomic medicine in cancer genetics a reality. The current gene panel tests 97 genes, 260 Genome-wide association (GWAS) SNPs and 24 fingerprinting SNPs. Professor Rahman took us through the sequencing and analytical challenges that the programme is facing and its attempts in setting a high standard for such initiatives. The genetic interpretation of the detected genetic variants is particularly complicated, and all variants should be considered innocent until proven guilty and deemed pathogenic mutation. In order to increase the confidence of the results, the programme has generated whole exome sequencing data on 1000 people from the 1958 British Birth Cohort, which has been made publically available (www.icr.ac.uk/genetic-resources). Making the causal relationship between a genetic variant and disease is the next hurdle in interpreting the functional significance of a variant. Interpretation goals for genetic testing are to make the processes automated, evidence-based and dynamic, where variants are triaged into clear clinical management categories. Custom-made functional assays need to be developed to study the mechanism of action for each variant found.

Towards the end of her lecture Professor Rahman discussed the economic benefits of Predictive Genetic testing in healthy individuals compared to Medical Genetic testing in diseased individuals, and how this changing paradigm is dependent on successful and timely integration of multiple expertises to apply technological advances into mainstream oncology services.

Professor Sadaf Farooqi followed, presenting her talk on ***Genetics and Obesity***. Starting with a reminder that obesity seen in population is largely environmentally driven, but with an underlying importance of genetic factors. Her laboratory studies the extreme phenotype of severe childhood onset obesity in a cohort of >5,000 patients, Genetics of Obesity Study (GOOS, www.goos.org.uk). Using the tractable approach of studying the extremes of body weight in humans, they have been successful in assigning roles for multiple genes in the energy homeostasis, achieving insights into mechanism of disease, which lead to drug discovery and benefits to patients (both diagnostic cally and interventionally).

Professor Sadaf Farooqi

Historically, the first studies to point to the brain regions controlling body weight, the hypothalamus and the brainstem, have been performed in rodent models. Also in experimental animals, it was discovered that fat acts as an endocrine organ, and nutritional signals from the gut are also transmitted in to the brain. Professor Farooqi and Professor Stephen O'Rahilly, through the discovery of gene mutations in patients with obesity, demonstrated that Leptin, a hormone secreted by white adipose tissue is a pivotal regulator of energy balance in humans. In the brain, leptin modulates the function of regions, involved in food reward as shown by fMRI imaging of patients. These studies illuminate the biological basis of food reward with complex behavioural regulation. The commonest highly penetrant obesity gene is MC4R, encoding the melanocortin receptor 4, accounting for approximately 5% of the genetic mutations in GOOS. In addition to their severe obesity, these patients paradoxically also have low blood pressure, disproportionate to their level of obesity.

Recently, the team was able to show that it was indeed Leptin, which is the key link to blood pressure regulation. The gene discovery program, combined with data from animal models is now building a picture of the brain control of energy balance, where these processes are regulated by molecules acting in several hypothalamic areas, the arcuate and the paraventricular nuclei in particular.

Recently, as part of the UK10K consortium, Professor Farooqi's team has undertaken whole exome sequencing in 1000 patients from the GOOS cohort. When they analysed

the genes with evidence for involvement in body weight controls in rodents, they discovered that nearly all of these >60 genes carry genetic variants in obesity patients. Understanding the individual contribution of each gene to the disease will be the next challenge. Several of the genes investigated (SIM1, SH2B1) show very strong links between behaviour and obesity, and these will be further investigated.

Looking into the future, she concluded her talk with her plans to look into the mirror extreme phenotype – extreme leanness in a large population cohort. Integrating the genetic findings from both phenotype extremes in an integrated approach will hopefully shed more light on body weight regulation in humans.

After lunch, in a session chaired by Professor Philippa Talmud, we heard the lecture of **Professor Bobby Gaspar** (Institute of Child Health, University College London) entitled ***Gene therapy***.

His research focuses on gene therapy for monogenic disorders of the bone marrow.

Professor Gaspar started his lecture with the history of the gene therapy in paediatric diseases such as haemophilia and Fanconi anemia. He then went on to explain why bone marrow diseases represent particularly good targets for gene therapy, focusing on the known cell populations, which could also be manipulated *ex vivo*.

He discussed the development of gene therapy for severe combined immunodeficiency (SCID), X-linked SCID and Adenosine deaminase (ADA) SCID. The X-linked SCID was the first condition to be treated using gene therapy. In 1999 it was demonstrated that the growth of the lymphocytes is blocked. Applied gene vectors introducing the gene for the interleukin receptor 2 gamma (IL2RG) under the control of viral promoters were used on the first ten

Professor Bobby Gaspar

patients to be treated at Great Ormond Street Hospital. Although initially the clinical outcome was promising, there were side effects to the therapy. Four patients in France and one in the UK developed leukaemia. At the core of this was the genome integration of the constructs near an oncogene, driven by a powerful long terminal repeats (LTRs) in the viral promoters. Work to improve the vectors led to development of novel LTR-free constructs to contain internal mammalian promoters and the results encouragingly demonstrate that these are safer. The biochemical defect in ADA SCID was also shown to be correctable by reintroducing the ADA gene and 42 patients treated in USA, UK and Italy show 100% survival rate.

Professor Gaspar talked of other disorders where gene therapy has been applied, including lysosomal storage disease, a diverse group of disorders with complex systemic and central nervous system pathologies, where functional copies of the defective enzymes have been introduced. In the case of the X-linked adrenoleukodystrophy and metachromatic leukodystrophy, haemopoetic stem cell gene therapy with lentiviral vectors is used. The bone marrow cells are corrected *ex vivo* and reintroduced in the patient.

Concluding his talk with an overview of the gene therapy translational research process, he also described the future direction in the field, which focuses on the application of gene editing technologies (Zn-finger proteins, TALENs, and CRISPR/Cas9 nucleases) to repair the defective genes, carrying a promise of accurate and safe *ex vivo* gene manipulation.

The 2014 Galton Lecture was delivered by **Professor Andrew Wilkie, FRS** (University of Oxford), entitled ***Lionel Penrose and the paternal age effect for mutations – sixty years on.***

In 1912 Wilhelm Weinberg noted that sporadic cases of achondroplasia (dwarfism) were more often seen in the last-born than first-born child, this is one of the first examples of the recognition of parental age in genetic disease. It was Lionel Penrose, the Galton Chair at the Galton Laboratory at University College London, who in 1954 first suggested that paternal age is a key factor to the phenomenon, using the example of Down syndrome. It is now accepted that the father's age is of pivotal significance for the development of certain disorders such as achondroplasia. The maternal age link to cranio-

synostosis (premature fusion of the cranial sutures) the focus of Professor Wilkie's own research, was also first suggested by Penrose in 1938.

In the 1960s, working on the craniosynostosis condition Apert Syndrome (AS), Eric Blank noted a paternal age effect, maternal age effect and birth order, which seemed all to have an effect upon initial investigation. Using correlation and partial correlation methods, the only significant factor was determined to be the paternal age effect. Human spermatogenesis and DNA replication errors were the longstanding hypothesis behind this effect.

Professor Wilkie's own work has conclusively demonstrated that it is in fact a selfish selection that drives this accumulation of mutated gene copies. He recounted the story behind this ground-breaking discovery in his study of the genetics of AS, which occurs in 1:65,000 live births. The condition almost always arises by new mutations. In 1995, he discovered the gene defective in AS, the fibroblast growth factor receptor 2, FGFR2. The two most common mutations affect two adjacent CpG island encoding Ser252 and Ser253, and act in a gain of function mechanism by increasing the binding affinity of the receptor to its growth factor, and the downstream signalling cascades. Both FGFR2 gene and FGFR3 gene (mutated in achondroplasia) have a >500-fold elevated rate of mutation over the background rate in the human genome. His laboratory confirmed the paternal origin of all AS cases (147/147). Other genes have also been linked to paternal age effect, including HRAS (Costello syndrome), PTPN11 (Noonan syndrome), RET (Multiple endocrine neoplasia 1 and 2). Studying the AS mutation levels at position 755, Professor Wilkie was able to demonstrate a selective advantage of the pathogenic FGFR2 mutations. This provided the conclusive evidence of selection mechanism behind the mutation rate, rather than a copy-error hypothesis, and described a selfish spermatogonial selection process. Similar spectrum of mutations is seen somatically in specific cancers, such as testicular tumours, where FGFR3 mutations were found.

Professor Andrew Wilkie, FRS

Some of the work in the lab is currently trying to map the occurrence of these mutations in testes. In conclusion, same mutations in germ cells cause both selfish clones and birth defect, whereby the mutation which is beneficial to the testis is harmful to the organism.

Looking into the future for the next 60 years, Professor Wilkie predicted a comprehensive catalogue of the mutations at each position in the human genome, which would broaden our view on consequences from human evolution and disease. Improving early genetic testing and interventions would facilitate complete integration of genetics in medical care.

In her presentation of the Galton Plate to Professor Wilkie, Professor Veronica van Heyningen, FRS, President of The Galton Institute, thanked him for his elegant work and presentation.

Professor Sir David Weatherall, FRS (University of Oxford) concluded the conference with a paper entitled ***Summing up: what we have learned from genetics for medical care.***

He started by describing the global rate of birth defects. A recent report, published by the March of Dimes Foundation found that an estimated 7.9 million children (6% of total births worldwide) are born with a birth defect of genetic or partially genetic origin, and 3.3. million of these children die each year. Five diseases account for 25% of this number, namely congenital heart defects, neural tube defects, Down syndrome, glucose-6-phosphate dehydrogenase (G6PD) deficiency, and the haemoglobin disorders (thalassemia and sickle-cell disease). This global health burden is affecting especially severely the middle- and low-income countries, altogether accounting for 93.7% of birth defect cases. Poor maternal health, poverty, and greater frequency of consanguineous marriages are recognized significant risk factors.

It is difficult to document the frequency of genetic disease accurately. Recently, the first comprehensive global study of the hemoglobinopathies, causing deficiencies in structure

or function of haemoglobin, has documented the disease burden and epidemiology. Professor Weatherall shared his personal experience in the inherited disorders of haemoglobin over the past nearly 50 years. The development of prevention screening by measuring the rate of haemoglobin synthesis, and the relationship between maternal and foetal haemoglobin made possible the prenatal diagnosis of the disorders. Other successes of early screening include the use of amniocentesis to test for chromosomal disorders, prevention of Rh haemolytic disease and the control of metabolic disease by neonatal screen. The outcomes of successful screening programmes include improved management and survival.

Professor Sir David Weatherall, FRS

He recounted his work in Cyprus on the β-thalassemia, providing one of the early examples of population control. Raising the population awareness to the disease and its prevention by prenatal diagnosis used any available help, including the unexpectedly strong support and assistance from the Greek Orthodox Church. In Sri Lanka, the efforts the Oxford-Sri Lanka North/South partnership has put their efforts into initiating the National Thalassaemia programs for education and screening, as well as building a central reference laboratory and a National Thalassaemia Centre.

In the summary of his presentation, Professor Weatherall recognized the major progress in the control and understanding of Mendelian Disease, the increased knowledge of the mechanisms and potential value of biomarkers in cancer, as well as the valuable contribution of genetics to the control of communicable disease. Looking into the future, he predicted that more progress should be expected towards defining the genetic component of common disorders by realizing the value of rare phenotypes and applying better phenotypic definitions in future GWAS studies, and continuous development of partnerships between countries to improve disease prevention and management.

Report by Elena Bochukova (University of Cambridge)

The meeting was organised and chaired by Professor Dian Donnai (Manchester University) and Professor Philippa Talmud (University College London)

THE GALTON INSTITUTE
Conference 2015

To be held at The Royal Society on Wednesday, 11 November, 2015

The topic is *MATE CHOICE* and the
Galton Lecturer is Professor Alan Bittles
Other details will be announced on the website and in the next Newsletter

Admission free but strictly by ticket
From: betty.nixon@talk21.com

British Society for Population Studies

Annual Conference 2014

This year's BSPS annual conference was held at the University of Winchester, with attendance surpassing 300 for the first time ever. The venue itself was splendid, and BSPS were favoured with three days of bright and sunny weather, always a bonus. Over the course of the Conference, 186 submitted papers were presented in 40 strand sessions, with 6 sessions running simultaneously in each time slot. This year saw a particularly lively poster session, with over 50 posters on display, attracting much comment and discussion. Training sessions were offered on *How to analyse UK Census Flow data: Wifi and Excel*, and *How to create and compare demographic projections for local planning & estimate the children from new housing*. Additionally, a PhD workshop gave graduate students an opportunity to present and discuss their planned dissertations with senior academics. BSPS is very grateful to all who gave their time and expertise to bring these special sessions to Conference, and to all those who organised strands.

Two plenary sessions attracted large audiences. David Satterthwaite (International Institute for Environment and Development – IIED) presented on the theme of *Can a finite planet support an urbanizing world?* Eilidh Garrett (University of St. Andrews) spoke on *Historical Demography: past, present and future, a genealogist's view*.

The BSPS website at www.bsps.org.uk has the full Conference programme (including all the paper abstracts) available to download as a PDF. Abstracts are also presented separately there by strand, with contact details of presenters if further information is required.

The 2015 Conference will be at the University of Leeds on 7th -9th September. BSPS hopes to see you there.

Plenary 1: "Can a finite planet support an urbanizing world?" - David Satterthwaite

In the first plenary of the conference David Satterthwaite challenged us to imagine what the ideal, sustainable city might look like. The rapid pace of urbanization in recent years in the developing world has indicated that the future is irrefutably urban. On the one hand, he argued, this trend holds much promise: cities have the advantage of providing compact and agglomeration economies where opportunity and talent can be fulfilled, and resources can be pooled to reduce risk arising from natural disasters, climate change or poverty. On the other hand, however, cities as the centres of population growth and the concentration of wealthy groups can often – though not inevitably – be heavily polluting and more burdensome in their environmental impact.

Having laid out the problem, Dr Satterthwaite's plenary was structured in three parts. In the first part of the talk, he presented data on green house gases (GHG) emissions for several cities across the world to illustrate their differential environmental burden and highlighted the shortcomings of existing indicators to track emissions. With the richest 2% of the world contributing 50% of the world's GHGs, high-consumption lifestyles are a root cause of high GHG emissions. Larger, less dense and automobile-dependent cities, such as Washington, DC, and Denver, Colorado, are some of the worst culprits in their GHG emissions. This does not imply, however, that all rich cities are equally culpable and inherently unsustainable. Oslo, Copenhagen and Porto Alegre provide positive counterexamples of compact, wealthy cities with comparatively low GHG emissions per capita. The sustainable level most cities ought to aim towards is around 2 tonnes/person/year – a level that unfortunately very few cities including the positive examples mentioned currently meet. When interpreting the GHG per capita indicator, Dr Satherthwaite stressed the importance of being aware of the underlying factors used to compute it. He emphasized the need for more detailed statistics, such as those that distinguish between consumption- and production-based emissions, to enable a thorough judgement of the environmental impact of cities.

Given that most cities across the world currently fail to meet sustainability standards, where might we find a positive template to guide us?

In the second part of his talk, Dr Satterthwaite provocatively revealed insights on planning sustainable cities from the most unlikely of places – the world's largest urban slum in Mumbai, Dharavi. With a staggering population of 400,000 within two square kilometres, he argued that Dharavi provides several valuable lessons to city planners in its com-

pact use of space, recycling-intensive and renewable-energy focused diverse economies, and high levels of community participation. Through a series of photographs, anecdotes and local perspectives gained through fieldwork and projects there, Dr Satterthwaite spoke with an intimate familiarity of the ways in which Dharavi is pioneering but also the ways in which it is flawed. Its most serious challenges, such as poor sanitation, lack of toilets and poor occupational and ecological health, need urgent redress and he emphasized the salient role of local stakeholders and a federation of slum residents in organizing for change. Ultimately, while Dharavi remains an imperfect example, it is one that encapsulates a number of challenges that similar contexts in the developing world are already facing or will do so in the near future.

Dr Sattherthwaite concluded his talk by setting out the goals cities ought to target to enable 'a finite planet to support an urbanizing world'. Cities, he underlined, must seek to cut GHG emissions by facilitating infrastructure and encouraging lifestyle changes that decouple a 'high lifestyle' from 'high energy consumption' one. They must pool their resources more efficiently to facilitate disaster risk reduction, poverty reduction and a more equitable provision of resources. Cities more urgently than ever need to generate methods to adapt and mitigate the risks of climate change. Tackling all these issues, he acknowledged, is no easy task but one that requires greater autonomy and resources to be entrusted to city governments, plus the growing participation and recognition of local stakeholders and residents.

Plenary 2: Historical Demography: Past, present and future, a genealogist's view - Eilidh Garrett

Delegates were treated to an engaging discussion on historical demography by Dr Eilidh Garrett of the University of St Andrews who started her talk by acknowledging the wide array of people and institutions which have helped shape her career, giving the audience a potted 'genealogy' of her own work in the past few decades. She particularly thanked Dr Alice Reid, a fellow historical demographer at the University of Cambridge, with whom she has a close working relationship spanning the last 20 years.

The main thrust of Dr Garrett's lively talk was to welcome a breaking down of barriers within the field of historical demography and the resulting dialogue between different areas of the discipline, both within the UK and internationally. Dr Garrett welcomed this new and exciting era for historical demography, one which follows the recent celebration of the Cambridge Group for the History of Population and Social Structure's 50th anniversary, a group she has long been associated with. After these celebrations, She relayed with pleasure that the European Society for Historical Demography would meet for the first time, a clear marker of the beginning of an international dialogue for historical demography.

The changes this new era has brought have largely been heralded, in the UK at least, by the arrival of two new datasets which Dr Garrett drew upon to illustrate her talk. She also drew comparisons with other existing datasets such as the North Atlantic Population Project. However it was data from the UK that was at the forefront of her talk.

To introduce genealogy, Dr Garrett talked of the popular television show, 'Who do you think you are?'. This program has raised the profile of genealogy, encouraging a diverse group of amateur genealogist to begin researching their family tree. Dr Garrett emphasised that it is this popularity which has prompted organisations to digitise individual records and place them online: for a small fee, amateur and professional genealogists alike can access these records instantly. Her talk went on to discuss how the ready availability of these data is welcome to historical demographers who build up from these individual records to explore trends and patterns in the population, much as demographers do with contemporary data.

However, despite the advances in technology which have eased conducting this kind of research, Dr Garrett emphasised that historical demography is hampered so long as those working in this field remain 'boxed' into their own sub-disciplines, defined by their use of different data sources, methods or even their interest in different spaces or times. Indeed she joked that on arrival at Cambridge she was known as the 'census woman' who looked at snapshots in time, unlike proper historical demographers who link baptisms, marriages and burials to reconstitute families.

To help illustrate the importance of talking to fellow historical demographers and demonstrate how this can help enhance our understanding of different historical events, Dr Garrett used the example of the fertility transition in Europe. This example also served to introduce the Integrated Census Microdata (I-CeM) held at the University of Essex. She discussed how the team has painstakingly collated census data for Britain between 1851 and 1911, harmonising the data over time to consistent and detailed

geographies.

Dr Garrett showed the audience how sub-registration district-level data can help us understand the fertility transition in a way that previous notable attempts such as the Princeton surveys or work by Robert Wood were unable to do. She talked of how these studies, using either county-level or registration district-level data were not detailed enough to help explain why or where the fertility transition began. However, using sub-registration district-level data from I-CeM, she showed the audience how differences in fertility rates between areas devoted to textile or mining industries might help explain the fertility transition.

For the final section of the plenary, Dr Garrett introduced another new dataset which plays a similar role in breaking down the 'boxes' of historical demography: the 'Digitising Scotland' project will digitise 24 million Scottish record images of births, marriages and deaths since 1855, and has the potential to be a great tool for demographic research. She talked enthusiastically about the prospect of such a rich data source where deaths will be coded to ICD-10 codes and variables such as occupation will be comparable across time. To illustrate the value of this dataset, Dr Garrett showed the audience a graph of 'deaths per day' in Scotland for 1950 and 1951 using data from the pilot study. As cause of death are coded to ICD-10, this dataset can help us understand an epidemic in Scotland during this time, as well as following its progression through the country because the data is geo-referenced.

Dr Garrett concluded her inspiring talk by thanking genealogists and records offices who have prompted the establishment of two new data sources with the potential to bring historical demography to the forefront of population studies. It is hard not to agree with her conclusions and share in the excitement for a new era of historical demographic research as we look forward to the insight into our past which this will bring.

Thanks for plenary reports to: Ridhi Kashyap (University of Oxford) – David Satterthwaite

Fran Darlington
(University of Leeds) - Eilidh Garrett

BSPS would like to thank **The Galton Institute** for their invaluable financial support again in 2014. This helps to defray the plenary speakers' expenses and the bursaries for student members.

BOOK REVIEW

***The Forgotten Brummie: the life and legacies of Sir Francis Galton*, by David Allen**

Pub. David Allen (2014), ISBN 978-1500305925 pp 162

This is a lively and enjoyable book to read covering the multiple contributions that Francis Galton made to African exploration, meteorology, statistics, mechanical inventions (Galton whistle, Galton Board etc.), heredity, psychology and of course introducing the notion of eugenics. One has to be a sort of polymath oneself to write learnedly about all these topics if one wants to do justice to Francis Galton's endeavours. This book does just this and gives an excellent layman's account of the field in the first 64 pages of the book, which is an accomplishment in itself.

Thereafter the interests of the author come more to the fore. He is an industrialist and was financial director of Cadbury Ltd in Bourneville, Birmingham and explores the social, economic and political aspects of Galton's ideas on eugenics. He defines eugenics as 'the scientific study of the biological and social factors which improve or impair the inborn qualities of human beings and of future generations'; there are many other simpler definitions of eugenics that Galton advanced but this is perhaps the most controversial one so providing lots of material to write about!

Starting with Galton's thesis that people's advantageous mental abilities are inherited (from his book *Hereditary Genius*) implies that mental disabilities are also inherited and what should society and politicians do about the latter group. Basing ideas on Darwin's views on natural selection about survival of the fittest Galton suggested that the mentally fit should be encouraged to procreate, whereas the mentally unfit (unintelligent, idle, feckless, sociopathic etc.) should be discouraged from doing so. This is a gross oversimplification which is no doubt why politicians subsequently took up and modified these ideas to suit their own purposes with so much enthusiasm.

This book, appears to align more with the position of State care combined with individual responsibility for the unfit. Thus he deplores the evolution of the Welfare State built by the post-war Labour government of 1945 that gradually introduced

Unemployment Benefits, Family Allowances, Housing Benefits, Child Allowances, Home help, Meals-on-Wheels. etc. which led to an enormous national debt. The Welfare State made it financially advantageous for some people to be out of work rather than to take a regular job; or to have a large family than to work because they could live off child benefits; or for young women to have multiple pregnancies (often with different fathers) rather than trying to become more self-sufficient; or to provide State support for heavy smokers to have expensive medical treatments even if they refuse to stop smoking.

These are all complex socio-political issues and a large number of people come into the story including: George Bernard Shaw, Sydney Webb, H G Wells, Marie Stopes, Maynard Smith, Julian Huxley, Aldous Huxley, etc. If the reader has momentarily forgotten who they were and their contributions, the author provides a very helpful mini-biography of each - those for Vilfredo Pareto, Montague Norman, A Gobineau and O Verschuer were particularly useful for me. I had not forgotten them, I never knew of them.

The last point that the author makes is that Francis Galton has been forgotten in Birmingham – 'The Forgotten Brummie'- well he is not forgotten in London. There is a very active Galton Institute (see our website) with a highlight being an annual Galton Lecture given by such eminent men as John Maynard Keynes, J H Edwards FRS, Sir David Weatherall FRS, Sir Walter Bodmer FRS. etc. The last two presidents of the Galton Institute have been J S Jones FRS, Sir Walter Bodmer FRS and Veronica van Heyningen FRS is our current one. University College London also holds quite frequent seminars on Galton's and Pearson's contribution to statistics. Perhaps the author could make the occasional trip to London to come to one of our Annual Symposia.

David Galton
David is Professor of the Department of Metabolism and Genetics at Bart's Hospital, London and a Trustee of The Galton Institute.

From DNA to Social Minds

Report of conference held by the Department of Psychology at the University of York
30 June—1 July, 2014

The role of genetics in human social behaviour has become a topic of considerable interest and importance in recent years. However, biological scientists and social scientists are rarely in the same room discussing the issues that arise at the nexus of their respective fields. The goal of the event was to attempt to bridge this gap by drawing together academics from each of these research traditions, and to this end the organising committee managed to attract a stellar set of keynote speakers from the fields of molecular and behavioural genetics, personality neuroscience, and experimental social psychology. The conference delegates also represented a broad range of disciplines, including evolutionary psychology, behavioural genetics, personality, social psychology, social neuroscience, and anthropology.

David Skuse (University College London) provided an excellent opening keynote on the role of oxytocin in human social behaviour (with a focus on face emotion recognition). Philip Corr (City University) followed up with a tour de force overview of personality neuroscience research, and also highlighted a number of the challenges that behavioural scientists will face if they wish to integrate biological and personality data successfully.

Essi Viding (UCL) started Day 2 with an inspiring keynote on the darker side of human social behaviour: the genetics of psychopathy and conduct disorder. The final key note of the conference was delivered by Constantine Sedikidis (University of Southampton) who provided an outstanding talk on the topic of self-knowledge and self-enhancement (as well as a crash-course on Ancient Greek philosophy!). In addition to their key note presentation, our speakers also engaged in a lively and thoughtful panel session at the end of Day 1 discussing: "Where next for biosocial sciences?".

We also saw ten high quality oral presentations from delegates over the two days on topics including affective neuroscience, hormones and behaviour, face perception, the evolution of cooperation, and gene-environment interplay on human intelligence, as well as 18 posters of similar breadth and interest.

All in all, the event fuelled a large amount of thought in the domain of social genetics and related fields and provided a forum for discussion that often doesn't find its place in more traditional academic conferences.

Gary Lewis
Department of Psychology
University of York

This conference was part sponsored by **The Galton Institute** with additional financial support from the European Human Behaviour and Evolution Association.

European Human Behaviour and Evolution Association

Conference 2014

With the support of the Galton institute, 2014 saw Bristol hosting the 2014 European Human Behaviour and Evolution Association conference. The venue, At-Bristol, was a fantastic springboard and the organisers received well-deserved thanks (and flowers).

As always, what stood out was the wide range of approaches represented by the 200+ delegates: Evolutionary psychologists, cultural evolutionists, human behavioural ecologists, evolutionary biologists, developmental psychologists, and an array of scientists who simply seek to understand human behaviour using evolutionary principles. EHBEA is a wonderful illustration of the benefits of bringing together different approaches – beautifully illustrated by Young Investigator Prize winner Willem Frankenhuis's plenary, which showed us how combining developmental and evolutionary sciences can lead to fascinating new insights.

The keynotes were equally diverse: Russell Gray started us off with the centrality of language to understanding human history, cognition and culture; Annette Karmiloff-Smith led us towards rapprochement in the great 'domain specific' vs 'domain general' debate; Martie Haselton gave us the latest on the lively debate surrounding changes in women's mate preferences across the ovulatory cycle; Daniel Hruschka asked the question – what proximate cues tells us to engage in costly giving? While Samir Okasha tackled a more 'ultimate' question – how does kin selection versus multi-level selection help us understand the evolution of social behaviour?

Discussions - in the post-talk Questions and Answers, during the breaks, and on Twitter - were lively. Much of the focus was on classic evolutionary questions: how humans choose partners; why they cooperate; how childhood learning shapes us; how we ended up with language. There were sessions with a practical focus too – the 'Brainjuicer' talk on how scientific understanding can inform business will have been of interest to scientists contemplating their 'pathways to impact' as they seek research funding in an increasingly impact driven climate. And there were broader questions about how we should be (evolutionary) scientists: from the scourge of 'p-hacking' - the massaging of data to generate statistically significant results (and when it is or isn't cricket to suggest another scientist has done it), to the use of Amazon Mechanical Turk to recruit participants for studies. In particular, discussions concentrated on whether we've jumped from frying pan to fire in an effort to get more 'representative' samples than the 'standard' undergraduate from Western, Educated, Industrialized, Rich and Democratic (WEIRD) societies. We can expect to hear much more about both of these are issues.

We had some fantastic contributions from our student presenters: congratulations were due to Julien Barthes for winning the student presentation competition with his talk "Male homosexual preference: Where, when, why?" and to Jeanne Bovet for the best student poster entitled "Men prefer women with late expected age at menopause.". As always EHBEA was a fun and fascinating four days. We can't wait for EHBEA in Helsinki in 2015.

Dr. Katherine Cross,
University of St. Andrews

EHBEA would like to thank **The Galton Institute** who helped support this conference with a grant of £1,000.

Canadian Conference on Epigenetics: Epigenetics, Eh!

23-27 June, 2014
London, Ontario, Canada

The 2nd Canadian conference on Epigenetics was held in Western University in London, Ontario, organised by ***Melissa Mann, Nathalie G. Bérubé, David I Rodenhiser Thomas A. Drysdale, Christopher L. Pin and Victor Han***.

The focus of the conference was on Biochemical and Clinical applications. There were very informative talks by various researchers over 4 days, many relevant to my research-work, and I mention a few highlights .

The conference started with the plenary speaker: ***Hiroyuki Sasaki***. He first talked about the early years of his research career and the problems he faced with his mouse models of disease because of imprinting. He explained his fascination for the mechanism of imprinting.

He also talked about *Piwi* RNA, which are male germline-specific and maintain methylation at the ICR of

Rasgrf1 (Science). He showed some recent findings from his lab, where they have used post-bisulfite adaptor tagging (PBAT) to study the methylation status of postnatal sperm stem cells. His lab revealed large partially methylated domains similar to those found in placenta and cancer cells but not in somatic cells. They also discovered a high level of non-CG methylation in neonatal prospermatogonia, and stage-specific differentially methylated regions, both potentially important for the regulation of stem cell properties and differentiation.

On the second day, ***Dr Matthew Lorincz*** talked about transcriptional regulation of transposons. His lab demonstrated a role for DNA methylation in controlling expression of ERVs. Both ERV1 and ERV2 classes of retrovirus are marked with H3K9me3, with Setdb1 playing a very important role in the process of establishing these marks on the elements.

In addition, ***Dr. Carolyn Brown*** who is an expert in the area of X chromosome inactivation, explained how dosage compensation is associated with X chromosome inactivation. Her lab focuses on *XIST* RNA, DNA sequences and interacting proteins that establish silent chromatin. Using an inducible transgene for *XIST*, they dissected the region of RNA necessary for localization and silencing, as well as the recruitment of proteins and heterochromatin modifications. She also showed the importance of DNA sequence in the spread of silencing using DNA methylation to discover genes subject to, or escaping from silencing. These latter are often autosomal material shifted onto the X chromosome and X- linked transgenes.

There were poster sessions on the second and third day of the conference at which I also presented my own recent paper on two new classes of genes which share some features with imprinted genes and are controlled in part by DNA methylation (*Development*, Jan 2014). I was very happy to see people showing interest in our work and got good feedback and advice that can help me shape my future work and career.

Overall, the conference was a great experience as it provided me with a chance to present my work at an international meeting and to learn about new developments in other relevant areas in the field of epigenetics. It also offered me the opportunity to network with many leading scientists globally.

I would like to thank the Genetics Society and **The Galton Institute** for providing me with this great opportunity.

Avinash Thakur
University of Ulster

Junior Scientist Travel Grant

The Galton Institute has entered into an agreement with The Genetics Society to provide support of £500 toward each of three travel bursaries per annum organised and administered by the Genetics Society.

These £750 bursaries are given, on a competitive basis to outstanding students working for a PhD on a topic relevant to the mission of the Institute to allow them to attend appropriate conferences. Reports of their use of the bursaries will be placed in the Newsletter.

Details and application form can be found on the Genetics Society website.

Integrating the genome with the phenome

Annual meeting of the Bloomsbury Centre for Genetic Epidemiology and Statistics, in conjunction with the South of England Genetic Epidemiology Group

The Bloomsbury Centre for Genetic Epidemiology and Statistics (BCGES, http://bcges.lshtm.ac.uk) is a joint Research Centre of University College London (UCL), the London School of Hygiene and Tropical Medicine (LSHTM) and Birkbeck, University of London. In 2014 its annual scientific meeting was held in conjunction with the South of England Genetic Epidemiology Group, an ad hoc colloquium of researchers from institutes in London, Cambridge, Oxford, Bristol, Cardiff and elsewhere. The meeting, on the theme of "Integrating the genome with the phenome", was held at the Institute of Child Health, UCL with the support of The Galton Institute on 8 July 2014.

Following the successes of genome-wide association studies in mapping genes affecting single traits, a major new challenge is to integrate these results with the wealth of additional phenotype data becoming available. This includes identification of genes affecting multiple traits, integration of data from multiple "-omics" technologies, and delineation of relationships between genes, intermediate biomarkers and disease. Each of these areas has the potential to improve our knowledge of the molecular mechanisms of disease. Methods to deal with this wealth of data are under active development and are not widely known by applied researchers. This meeting aimed to promote dialogue between methodological and applied researchers, and

to open up discussions on how best to develop the analysis methods for these challenging new data.

Angelica Ronald (Birkbeck) opened proceedings by describing how genetic studies of multiple psychotic dimensions in adolescence could shed light on the developmental origins of schizophrenia. Delilah Zabaneh (UCL) then described the UCLEB consortium, a group of prospective cohort studies with detailed phenotyping on cardiovascular biomarkers, and showed how different sources of genomic annotation were being used to prioritise genetic associations for follow-up work. Matthew Silver (LSHTM) explained how in the Gambia, the season in which a child is conceived affects its later health outcomes, and described how epigenetics could explain this effect by mediating the seasonal variation in maternal nutrition. Marco Scutari (UCL) closed the morning by explaining Bayesian networks and their use in mapping quantitative trait loci for multiple traits simultaneously.

After lunch, Chris Wallace (Cambridge) described several projects in her group, using statistical methods to identify shared genetic control of related traits such as autoimmune diseases. David Balding (UCL) reviewed the concept of kinship and how it has been redefined in the wake of high throughput genotyping which allows precise calculations of kinship for nominally unrelated individuals. Mike Weale (Kings College London) presented methods for using genomic annotation data as prior information to sharpen statistical inference about genetic association. Finally, Jonathan Marchini (Oxford) described a general statistical model for assessing association with multiple correlated traits, allowing for relatedness between study subjects, and presented several analytical and computational advances that are implemented in his software.

The meeting was attended by 200 delegates who welcomed the range of topics covered. A number of posters were also contributed and helped to create a lively atmosphere during lunch and the subsequent drinks reception.

Frank Dudbridge

Professor of Statistical Genetics
London School of Hygiene and Tropical Medicine

Thanks go to **The Galton Institute** for helping to fund this conference.

Genetics, Genomics and Global Health: Inequalities, Identities and Insecurities

Conference Centre, University of Sussex, 18 July 2014

On Friday 18th July 2014 an interdisciplinary one-day conference brought together experts from the fields of policy, research, industry, foundations, journalism, and non-governmental organisations at the University of Sussex for the 4th Annual Global Health Conference on "Genetics, Genomics and Global Health –Inequalities, Identities and Insecurities". It was co-organised by the University of Sussex Centre for Global Health Policy, the Wellcome Trust, Brighton and Sussex Centre for Global Health Research, the Centre for Bionetworking with support from the European Research Council, the Global Health Working Group of the British International Studies Association, and the Galton Institute. Following a keynote and plenary panel on 'Genetics, Genomics and Global Health' participants divided into groups to debate specific topics –including global health gaps, genetic privacy, global health security, molecular diagnostics, genetic identities and bioinformation economies. The general format was for invited experts to give short presentations, followed by wider discussion with the audience. The diversity of disciplines represented coupled with the theme ensured lively and thought-provoking discussion and a number of key points emerged from the day.

The key points to emerge from the meeting are summarised below:

1. Genetics and genomics could prove transformational for global health in the coming decades by generating new opportunities for diagnosing, treating and managing a number of communicable and non-communicable diseases. However, at least two critical barriers remain for low- and middle-income countries: (1) comparatively little research focuses upon locally relevant diseases, taking into local genetic variation and conditions in low-income countries; (2) the high cost of many technologies –which are principally developed through private sector and commercial involvement – makes access to most of these technologies prohibitively expensive for low-income countries.

2. A decade after the first human genome was successfully sequenced, we are gaining a much more nuanced and fine-tuned picture about which diseases genetic and genomic information may help address in future. It is also becoming clear, however, that the impact of genetics and genomics on global health will not be uniform; rather it will likely vary across different diseases as well as different areas of global health – such as humanitar-

ian biomedicine, population health, and global health security.

3. Realizing the potential global health benefits of genetic and genomic information will require difficult balances to be struck in the years ahead, including: (1) between the commercial interests driving the advancement of new health technologies versus protecting the privacy of people's genetic and genomic information; and (2) between investing in new capacity for genetic and genomic technologies in low-income countries versus spending on more affordable but well-established technologies with a proven track record in improving global health, as well as on the wider social determinants of health.

4. The impact of genetics and genomics on global health will depend not just on the technologies themselves, but also on the wider social, political, and economic contexts in which new technologies are adopted and/or adapted. In particular, the process of producing genomic information will likely have profound implications for the way in which health care data will be structured and formatted in future, so as to better facilitate its triangulation with genetic data. That process will have potentially far-reaching social consequences – consequences for which societies are not yet fully or even well prepared.

5. Despite the likely long-run benefits of genetics and genomics for global health, history cautions that the introduction of new technologies are also often accompanied by new risks. Here there are dangers that new information about life at the genetic level could be put to nefarious use, or that even well intentioned scientific research on lethal diseases could lead to an accidental release of a dangerous pathogen. Harnessing genetic and genomic knowledge for the advancement of global health will thus have to navigate carefully between realizing the social benefits whilst minimizing possible new dangers.

We are particularly grateful to **the Galton Institute** which supported the conference with a grant of £1000. This funded the attendance of the keynote speaker, Professor Andrew Lakoff, Associate Professor of Sociology, Anthropology and Communication, University of Southern California and 20 Masters and PhD students at the meeting.

Melanie Newport
Professor in Infectious Diseases and Global Health, Brighton and Sussex Medical School

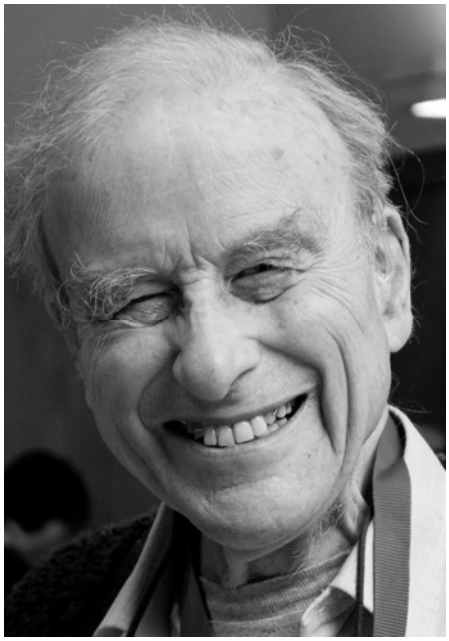
OBITUARIES

Dr Harry Stopes-Roe
27 .3.24. - 11.5.14.

Harry was a long term member of the Galton Institute and its predecessor the Eugenics Society. He was also the only child of the dominant birth-control pioneer Marie Stopes and her second husband Humphrey Roe a businessman who had distinguished himself as a pilot in the First World War.

Marie Stopes had characteristically strong and idiosyncratic views on how Harry was to be brought up: no books as they might give him second-hand and thus second rate thoughts; no trousers until he was eleven, as they gave heat in the wrong places and similarly no bicycle riding. This allied to the letter she dictated to her husband absolving her from her marriage vows as he could not satisfy her says much about her personal sexuality and the milieu in which Harry was brought up. Her first marriage had been annulled on grounds of non-consummation.

Harry was educated privately until he went to Charterhouse. He read physics at Imperial College London and gained a PhD in philosophy from St John's College, Cambridge. He moved to Birmingham as a lecturer and senior lecturer in Science Studies which neatly included his twin interests of science and philosophy. It was whilst there that he stared to devote his life to establishing humanism as a realistic alternative to traditional religions. He was rigorous in his thinking, worrying over precise phraseology. He was involved in the debates on the City of Birmingham's 'Agreed Syllabus for Religious Education' in 1975; this was the first time an attempt had been made to include non-religious approaches to living, such as humanism, in a multi-faith model of religious education. He helped devise the BHA's policy for education covering both religious and non-religious ways of living or 'lifestances' as he liked to call them, having debated for hours at the World Humanist Congress in Buffalo Illinois on the exact wording of a definition of humanism. The result was largely his: "humanism is a democratic and ethical life stance, which affirms that human beings have the right and responsibility to give meaning and shape to their own lives."


Dr Harry Stopes –Roe

Harry was active in the International Humanist and Ethical Union and

his work for the British Humanist Association was honoured in 2005 by being made a vice-president having previously been chairman.

This gives only a partial picture of Harry. His mother so objected to his marrying Mary Wallis daughter of the scientist and inventor Sir Barnes Wallis on the grounds that she was short-sighted and therefore eugenically impure that she refused to come to the wedding and largely cut him out of her will. Nonetheless Harry supported his mother's memory particularly when there were objections to his mother's depiction on a postage stamp, saying that her critics did not understand the views prevalent in the 1920's and that his mother had a strong sense of duty to the poor. Harry was intellectually rigorous and honest as well as being a charming human being, all characteristics which his mother should have applauded. That Harry was able to surmount his upbringing and present such a rounded and sensible personality is to his credit and doubtless to his wife's support.
He is survived by his wife, Mary, a retired psychologist at the University of Birmingham and two sons and two daughters.
GV 27.07.14.

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Dr Tim Black, CBE
7.1.37. - 11.12.14.

We note with regret the death of Dr Tim Black a family planning pioneer who co-founded Marie Stopes International, one of the world's largest family planning organisations which provides a range of health care services, including family planning advice, vasectomies and abortions in 40 countries around the world thereby helping six million couples each year.

In 1975 Tim Black and Phil Harvey put up money to buy the lease of Marie Stopes's famous Mothers' Clinic in Whitfield Street, London, reopening it as Marie Stopes International (MSI). Prior to 1975 the Galton Institute (the Eugenics Society as it was then known) had administered this clinic in the seventeen year period since the death of Marie Stopes - as required by a clause in her Will. The Institute's association with Marie Stopes International continued through the many projects we helped fund in the 40 countries where they worked.

Although Dr Black was not a member of the Institute our long association with him through Marie Stopes International means we remember him with both appreciation and admiration.
BN 7.1.15.

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Dr Anthony Smith
30.3.26. – 7.7.14.

Anthony was a member of The Galton Institute for 41 years and a Trustee for six of these. Anthony was best known as a bestselling author, broadcaster, adventurer, balloonist, rafter and former *Tomorrow's World* presenter. He published some 30 books, the best know of these was his best-selling work *The Body* (later renamed *The Human Body*) which sold over 800,000 copies.

Anthony was the first Briton to fly a balloon across the Alps in 1963 and led The Sunday Times Balloon Safari expedition flying from Zanzibar to East Africa and across the Ngorongoro crater the previous year.

In January 2011 Anthony set out from the Canary Islands on a home-made raft to cross the Atlantic to Eleuthera in the Bahamas. He had recruited three other volunteers and assembled a raft made of yellow gas pipes, topped with a small hut, a sail ballooning from a telegraph pole and with a foot pumped computer for communications. Whilst on the raft, in mid-ocean, Anthony celebrated his 85th birthday.

After arriving in St Maarten in the Caribbean he recruited another crew and set sail in April 2012, finally reaching Eleuthera 24 days later after being washed up in a storm on the beach. Amazingly, it was the same beach as the *Jolly Boat*, a lifeboat from the SS Anglo Saxon which was sunk by the Germans *in 1940*, was washed up on. Anthony had secured the return of this boat in 1997 from the Mystic Seaport Museum in Connecticut and ensured its display in the Imperial War Museum in London.

Anthony is survived by two sons, three daughters and a grandson. BN 7.1.15.

THE NEW A-LEVEL CURRICLUM
IS IT FIT FOR PURPOSE?

The wait is over. Biology teachers across England and Wales now know what they must prepare to teach for the new A-level curriculum, starting in September 2015.

Every five years A-level specifications change. This time, however, owing to government intervention (essentially Michael Gove), both content and format of A-levels are changing. Why A-levels must change so frequently is perplexing. Secretaries of State like to leave their mark and in the Sciences there are bound to be new topics which need to be included at the expense of others. Nevertheless, the more cynical members of the teaching profession see such changes as a way of selling new textbooks and the now essential software that accompanies them. Certainly, the publishing companies benefit when these changes appear.

So what's new in A-level Biology? I'm going to consider the two areas of change: content and format.

Firstly, how has the content changed and has it changed for the better? There are plenty of topics

which were on the syllabus when I started teaching A-level Biology in 1974 and which are still there today. Most of the biochemistry, cytology and ecology have barely changed and the same questions still come up on the exam papers after all these years.

At the Galton Institute, however, we are especially interested in the genetic components of the new specification. Here, there is good news to report. Dihybrid crosses and autosomal linkage have made a welcome return to the Mendelian section, while Genetic Drift finally makes an appearance alongside Natural Selection. The biggest changes, however, concern updating the section on DNA and biotechnology. Epigenetics gets a mention, as does methylation of DNA and acetylation of histones. For the first time, genetic fingerprinting uses terms such as VNTRs. Even genomics has a walk-on part.

Ofqual, who formulate these changes, are to be congratulated. Some teachers, understandably, are wary of change and are concerned with how they will approach some of these demanding topics and how they can afford to offer worthwhile practical work to their students. There is also more mathematical content which is excellent training for the budding career scientists but many 'average' A-level Biology students might well be put off by this.

This leads me to a concern shared by many Biology teachers: assessment of practical work in all the Science subjects. The new scheme will involve candidates undertaking 12 prescribed practicals over two years. These will be assessed by the teachers themselves, resulting in an overall Pass or Fail grade for practical work. In other words, it will not contribute to the actual grade, which will be entirely externally assessed by examination. There is no requirement for students to plan practical work and no way of discriminating the A* student from the grade E student. I should be astonished if less than 100% of candidates achieve 'Pass' for their practical work.

This lack of discrimination is a serious concern. The idea that in these practical subjects, practical ability is not contributing to the final grade seems absurd. It would be like aptitude for drawing or painting not counting in A-level Art!

A number of organisations have expressed their grave concerns regarding this, including BERG (Biology Education Research Group) and SCORE (Science Community Representing Education) but so far the DfE seem remarkably obdurate.

I have further misgivings with the format of the new curriculum. At present, almost all A-level students start their two year courses studying FOUR subjects. At the end of the first year, they sit exams (AS) and achieve a grade for each. University offers, however, are usually for THREE subjects and candidates usually drop one subject (and cash-in that AS grade), continuing with the other three. These AS marks then contribute to the full A-level.

Mr Gove didn't like this. He wanted all the assessment to be at the end of the two year course because he believed that this is a better measure of ability (despite university science courses being assessed over three years as modules). That is why he had already abandoned January module exams. So from September 2015, in ALL A-level subjects, candidates will start their four subjects, but if they sit AS exams at the end of the first year, these will NOT contribute to the full A-level. In other words, they are a waste of time and money except when a candidate is sure which subject is to be dropped. Unfortunately, this does not happen. Candidates typically work hard in all four subjects and then AFTER the AS results, decide which one to drop. This new system will work only if candidates know which subject they intend to drop from day one. Of course, candidates could just sit AS exams in all their subjects, as they do now, and then choose which one to drop. Surely these exams would be good practice? But why would students prepare thoroughly for exams which don't count for anything? There are also significant entry-fee implications for schools. The absurdity is of course that the man behind all this is no longer in post!

Ofqual have done a good job updating the content of A-level Biology. It seems madness to me, however, to detach practical work from the grade assessment. This, along with the change to AS exams, means that this new curriculum does not do the job. These changes were driven by political expediency and not educational merit. The end result is a great disappointment to most committed teachers.

Robert Johnston
Fellow of the Galton Institute

THE GALTON INSTITUTE

TEACHERS' CONFERENCE

Genetics in the 21st Century
30 June, 2015

To be held at:
Nowgen Centre
29 Grafton Street
Manchester M13 9WU

This is a genetics up-date day for A-Level Biology Teachers in the light of the changes to the curriculum

Admission free but strictly by ticket
From: betty.nixon@talk21.com