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Galton Institute

*Exploring Human Heredity*

# The Galton Review

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# EDITORIAL

Two of the aims of the Galton Institute are stated as:
"Promoting and supporting education and communication with regard to human heredity" and "encouraging the study and understanding of its historical origins and development".

It was with these in mind that Council decided to run an essay competition for 6th form students to mark the 150th anniversary of the publication of Gregor Mendel's findings on inheritance in peas in the *Proceedings of the Natural History Society of Brünn.* It was, of course, this work which defined the laws we now refer to as Mendelian inheritance.

The students were limited to 1,000 words on "any aspect of the life, work and/or legacy of Gregor Mendel". The marking team, led by Dr Elena Bochukova, spent many hours reviewing the entries. They considered scientific content, use of language and originality. Eventually, the winner was identified as Harry Pendlebury from St Mary's College in Liverpool. The judges were particularly impressed with his novel approach and mature use of language. The winning essay is reproduced on page 4.

The Institute's third aim is to "stimulate and inform public debate on the social and ethical implications of the study of human heredity and its relevance to human well-being". One of the main ways it promotes this is by giving financial support to various organisations wishing to run conferences on relevant subjects. In this edition, there are reports from two such events: the European Molecular Biology Laboratory Symposium and The European Human Behaviour and Evolution Association Conference.

Finally, I should like to draw your attention to the theme of our annual conference in November which will be *Environmental Factors in Gene Regulation*. It promises to be a fascinating programme and includes the delivery of the 100th Galton Lecture by Dame Linda Partridge on *Nutrition and Lifespan*. The full programme can be found on page 16 and our website along with information on how to register for tickets.

Robert Johnston

MENDEL ESSAY PRIZE 2016

The winning essay:

# The life, work and legacy of Gregor Mendel

Abstract:

The importance of 19th-century pioneering genetic research by Gregor Mendel is considered, referring also to the potentially useful but unachieved convergence of Mendel's specific and controlled plant breeding experiments with Darwin's observation-based conclusions about inheritance and variation. The potential benefits of their collaboration, had it occurred, are considered.

For most of human civilisation, the idea of inherited characteristics was implicitly understood visually. Humans observed that progeny – any kind, their own, plants and animals – commonly and broadly resembled parents. Nobody knew how it happened, it just "was". Any knowledge worth the name was purely observational.

By the mid-19th century, Biology was beginning to pass the "stamp-collecting" stage of mere observation and classification of species. This was partly due to Charles Darwin's ground-breaking publication "On the Origin of Species"[i] which implicitly suggested the existence of "agents" of inheritance. Much later, these were given the name we know now, "genes". This happened when mathematical analysis of breeding observations began to inform Biology via the efforts of Gregor Mendel and later, others.

Mendel, a monk living in Moravia, now in the Czech Republic, studied and eventually taught in the University of Brünn. His genetic studies originated from an interest in bees, and their agricultural usefulness. What might be learned about improving their breeding ability and effectiveness as plant pollinators? Before he started investigating pea plants, Mendel was a keen beekeeper; unfortunately the monastery forced him to remove them, as they were annoying visitors![ii] He turned to mice, to better understand animal heredity, although his abbot was negative about monks studying the results of animal sexual reproduction and behaviour.[iii]

His research into pea genetics was in fact incidental and a substitute for his normal work, but over the years he carried out thousands of carefully controlled selective breeding experiments on them. From 1856 he established the general observations that for each characteristic studied – such as plant height, pod shape or colour, seed shape or colour, and also flower position and colour[iv] – there appeared to be only two "invisible factors", thought by Mendel to be contributed, one from each parent plant, that affects the appearance of the next generation. His main results were made public in 1865[v].

Summarising, he determined that given the propensity of a trait to be expressed or not, it was possible to predict, with some mathematical accuracy, what the progeny of any one chosen cross would produce, regarding the percentage of plants having two or possibly more of the characteristics investigated.

Mendel also suspected from these analyses that there were two and only two "factors" determining each specific characteristic, what we now call the "phenotype". Observations also suggested that these "factors" were involved in a hierarchy of importance or "dominance" versus "recessivity", suggesting when and how either observed trait would appear in the progeny. He defined these two particular terms, and was the first biological scientist to so do. His conclusions were reinforced from his detecting recurring mathematical ratios, which later became predictable within large samples, for the likely appearance of specific characteristics in a future generation of plants. This was if he had recorded what the parent plants looked like and what their own parents had been like as well, otherwise the ratios would not work.

Mendel died in 1884 and the significance of his work was unrealised for almost 20 years. Most biologists still accepted the "blending inheritance" hypothesis, in that traits of a later generation would generally seem to be an integration of some of those of its parent generation. This would have implied that results of inheriting characteristics were not at all discrete or quantifiable. However, by 1900 Mendel's papers were rediscovered, and his experimental results with peas were rapidly verified by other biologists, in particular by Hugo de Vries, Carl Correns and William Bateson. Bateson himself lectured frequently advising other biologists to conduct very large and extended breeding experiments on organ-

isms including animals and plants, even before he became aware of Mendel's work.

There was still debate about quantisation extents in gene expression to give the "phenotype" (the words "gene" and "genetics" had by then been invented by Bateson). Then, later geneticists – and all before the importance, let alone structure, of DNA had been discovered – gradually integrated both approaches, the "biometric approach" of "fuzzy/non-quantifiable interaction of characteristics" and the Mendelian "genotypic approach" to practical heredity. Much later it became understood that genes often acted not just unitarily but in combinations, producing empirically-observed continuous variation, as well as fully discrete phenotypes.

Before Mendel, understanding of inheritance could not be called a science. Everything Man did to selectively-breed anything was observational and, as a result, somewhat unpredictable. For example, it took the Mayan culture about 9,000 years to modify generations of wild teosinte grasses to resemble the direct ancestor of *Zea mays* (sweet corn).

if Mendel and Charles Darwin had seen each other's research and could have corresponded, genetics might now be decades ahead of today's already advanced position, and a much maturer science. In the late 19th century, organic chemistry advanced sufficiently to be able to characterise nucleic acids, now known as DNA and RNAs. Thus, biochemical understanding of inheritance would also have developed earlier.

Neither scientist invented the word "gene" as we understand the term today. However, Mendel came closest, 50 years before the term emerged, to identifying that entity as a "factor". He showed statistically that genes exist and are quantifiably transmissible according to easily understood laws. Mendel's work is still important today as it is the stepping-stone to how modern genetics is understood.

Endnotes:

[i.] On the origin of species ref: Darwin, C. (1859), *On the Origin of Species by Means of Natural Selection, or the Preservation of Favoured Races in the Struggle for Life*, (1st ed.) (London: John Murray)

ii. http://mendelmuseum.muni.cz/en/g-j-mendel/mendeluv-vcelin *The Masaryk University Mendel Museum.*
iii. Henig, Robin Marantz (2000). *The Monk in the Garden: The Lost and Found Genius of Gregor Mendel, the Father of Genetics,* Boston: Houghton Mifflin. ISBN 978-0395-97765-1. pp. 15–17.
iv. Henig, Robin Marantz (2000). *The Monk in the Garden: The Lost and Found Genius of Gregor Mendel, the Father of Genetics*, Boston: Houghton Mifflin. ISBN 978-0395-97765-1. pp. 78–80.
v. Mendel, G J. (1865) *Experiments in plant hybridisation*, read to Brno natural History Society http://www.esp.org/foundations/genetics/classical/gm-65.pdf

Harry Pendlebury
St Mary's College, Liverpool


Harry Pendlebury with our editor Robert Johnston and Nicola Addy, Head of Biology at St Mary's College

All the entries were of a very high standard and the results were as follows:

1st Harry Pendlebury – St Mary's College, Liverpool
2nd Laura Cooper – Stockport Grammar School
3rd Laura Fitzgerald – Withington Girls' School (Manchester)

2015 Workshop on Fertility Preferences in Asia

(in preparation for the volume *'Family Demography in Asia: A Comparative Analysis of Fertility Preferences'*)

Asian Population Association

Scientific Committee on Fertility Preference

The Workshop on Fertility Preferences in Asia was held on July 27th 2015, alongside the 3rd APA International Conference in Kuala Lumpur, Malaysia. The objective of this satellite workshop was to produce a series of national reports concerning the 'state-of-the-art' of research on fertility preferences in countries across Asia. It was the initial step towards the preparation of the book: *Family Demography in Asia: A Comparative Analysis of Fertility Preferences,* which is scheduled to be published in 2016 by Edward Elgar Publishing.

The session focused on fertility preferences in countries across Asia, and was attended by authors and contributors to the book from 16 countries. There were 18 presentations on Asian countries, which were divided into several sub-sessions according to their geographic locations. These were East Asia: China, Taiwan, Japan and Mongolia; Central Asia; West Asia: Palestine and Jordan; South Asia: India, Bangladesh, Nepal and Sri Lanka; South East Asia: Indonesia, Papua New Guinea, Singapore, Cambodia, Timor Leste, Philippines, Myanmar and Malaysia. In these presentations, the authors presented the overview of their findings on country-level fertility preferences in 5 main divisions i.e.; background, data sources available and measurements used, results of national survey, evidence of regional surveys where available, and policy implications.

Most of the country's quantitative data were available from government's national health surveys. The total fertility rates (TFR) was found to be either declining or stagnating with time, except in case of Mongolia, where TFR has consistently increased from 1.9 in 2005 (below replacement level) to 3.1 in 2014. The authors found that factors like employment and labour market status in case of

Taiwan, education level of women in case of India and Sri Lanka, belief systems in Timor Leste, and nature and adequacy of family policy systems in several countries were some of the determinants of fertility preferences in Asia. It was concluded that strengthening existing family policy systems in several countries, disseminating knowledge about assisted reproductive technology in Japan, enhancing education especially among ethnic minority groups in Sri Lanka, part-time work arrangements for mothers in Singapore, and focus on regional and sub-regional policies in India will be good policy implications for respective countries.

The objective of the workshop was to assimilate all the country-specific findings into one book. Therefore book planning was one of the most important elements of the entire workshop, as it gave directions on how the book should be structured. After long discussions, it was decided that the first part of the book will deal with thematic fertility preferences issues such as labour migration, civil conflicts, son-preferences and more. Also, emphasis was decided to be placed on contextual factors of the fertility preferences.

Therefore, the workshop was not just a great learning opportunity for the participants, but also boosted in giving a concrete direction to the book. The convergence of different cultures in a beautiful setting of Kuala Lumpur made this workshop even more enjoyable.

Priyanka Raj
National Institute of Technology (MANIT), Bhopal, India

The organisers of this workshop are grateful to the Galton Institute for a grant of £1,000.

We hope you have all had an opportunity to look at the new Galton Institute website at **www.galtoninstitute.org.uk** and would be grateful for any feedback you may have.

You can email **executiveoffice@galtoninstitute.org.uk** with any comments you have.

Thank you for your help and interest.

## The European Human Behaviour and Evolution Association (EHBEA)

## Evolutionary Developmental Biology: Current Debates

The Programme Organizers: Professor Clark Barrett, Professor Tom Dickins and Dr Willem Frankenhuis

On 28 March 2015, a satellite meeting was convened in Helsinki prior to the annual EHBEA conference. The theme of this meeting was the role of development in evolutionary biology, with especial emphasis upon behavioural biology. There has been much discussion about how to conceptualize the relationship between development and evolution and this has gathered some momentum in recent years. The ambition of this meeting was to reflect the diverse issues at play within evolutionary developmental studies at the same time as highlighting key areas of future growth. To that end a similarly diverse set of speakers were invited to give their views on these matters, based on their own theoretical and empirical work.

Clark Barrett (UCLA) opened the meeting with a talk on Open reaction norms and human flexibility. Barrett argued that in biology, the concept of a reaction norm typically implies a closed mapping function: a curve that maps between a range of environmental conditions and a range of phenotypic outcomes, both of which are in some sense pre-specified. However, many of the things humans learn and the environments they face, are evolutionarily novel in many ways, never having been encountered by prior generations. Empirically, humans and other animals often (though not always) deal with such novelty adaptively. Generally, adaptive responses, shaped by selection, must be due to the fitness success of phenotypes produced in the past, which poses something of a puzzle: closed reaction norms generally involve mapping functions from environments to phenotypes that occurred over stretches of past evolutionary time. As a possible resolution to this puzzle Barrett introduced the concept of an open reaction norm, which can include both open (i.e. unspecified) and closed (specified) parameters. He outlined how such reaction norms might evolve and how to conceptualize them theoretically, using as an example, his recent work on children's cultural learning about danger.

The next speaker was Ben Dickins (Nottingham Trent University) who discussed (Epi)mutational dynamics and bet hedging. His focus was upon new phenotypes, which arise through changes in sequence context maintained by epigenetic marks such as histone modifications. Dickins made clear that epigenetic changes underlie cell differentiation, and noted that their contribution to inter-individual variation in multicellular organisms has attracted attention. This is because it connects gene regulation, which is responsive to environmental variation, to changes in populations. In microbes, mutations are a significant source of adaptive and non-adaptive variation, yet this variation is not simply random and can also be responsive to the environment. More generally DNA replication and repair vary in efficiency between lineages and mutation rates and biases cause evolution directly and through interaction with selection or drift. Dickins presented an examination of these dynamics that he argued can be used to guide our understanding of the interaction between variation-generating mechanisms and evolution.

Sinead English (Oxford University) discussed the use of Information as a loom to weave development and evolution. Her opening position was that natural selection shapes the developmental processes that construct the phenotypes, which, in turn, are under selection. She then went on to present a framework in which this interplay between development and evolution can be conceptualized by considering information transmission within and across generations. English showed how this perspective sheds light on major questions in evolutionary biology such as how individuals integrate different inputs for development (genes, parents, the environment) and the conditions under which non-genetic versus genetic mechanisms of inheritance evolve. A key component determining the reliability of information is the autocorrelation of the environment across generations. English described a meta-analysis on the empirical support for adaptive maternal transmission of information which illustrated how rarely studies have considered this autocorrelation. Thus, she argued, to move the field forward we need more theory-driven studies and data-driven theory.

Developmental changes in aggression and body size: an evolutionary perspective was the focus of Tim Fawcett's (University of Bristol) talk. Fawcett claimed that during development, humans and other social animals must learn how to navigate a dynamic, interactive environment in which the consequences of

behaviour depend critically on how others perceive and respond to them. Given the uncertainty inherent in such situations, natural selection will tend to favour rules for adjusting behaviour flexibly in response to social feedback from previous interactions. These rules should generate ontogenetic changes in behaviour as individuals gain experience and gradually adapt to their personal social circumstances. Fawcett has been exploring whether or not this evolutionary perspective can help us to understand observed patterns of behavioural development, using childhood aggression as an example. He presented a state-dependent evolutionary model that predicts developmental trajectories of aggression as a function of physical strength and information state, as individuals interact and learn about the consequences of their behaviour. He then analysed the developmental relationship between body size and physical aggression in humans, using a longitudinal cohort study of boys growing up in the Canadian province of Québec. Both the model and the data showed a strengthening association between relative size and aggressive behaviour across ontogeny, consistent with the hypothesis that individuals gradually learn about their physical capabilities through interacting with their peers. In the human data, after controlling for age-related changes, weight was the strongest predictor of aggressive behaviour: boys who were heavier than average for their age and height were more likely to get into fights or physically attack other children. These findings hint at the potential of evolutionary theory to shed light on the interplay between physical and behavioural development.

Emma Flynn (Durham University) spoke about *Developmental niche construction*. Niche construction is the modification of components of the environment through an organism's activities. A core claim is that humans modify their environments mainly through ontogenetic and cultural processes, and it is this reliance on learning, plasticity and culture that lends human niche construction a special potency. Flynn, an educational psychologist, constructed her talk to facilitate discussion between researchers interested in niche construction and those interested in human cognitive development by highlighting some of the related processes. She discussed the transmission of culturally relevant information, how the human mind is a symbol-generating and artefact-devising system, and how these processes are bi-directional, with infants and children both being directed, and directing, their own development. She then reflected upon these in the light of four approaches: natural pedagogy, activity theory, distributed cognition and situated learning. She concluded by offering three future directions; two involving the use of new tech-

niques in the realms of neuroscience and modelling, and the third suggesting exploration of changes in the effects of niche construction across the lifespan.

The concluding talk was from Daniel Nettle (Newcastle University) on *Developmental plasticity in the European starling: Empirical observations and evolutionary interpretations*. Nettle discussed how in altricial birds' experience during the first few days of life can affect many different behavioural traits in adulthood. Nettle and his colleagues study such effects using cross-fostering in the European starling, and he reviewed recent findings with respect to boldness, food motivation, impulsivity, expectation of reward, and flight performance. He noted that it is widely suggested that responsiveness to early developmental inputs is adaptive. Nettle outlined the prevailing adaptive explanations for such responsiveness, (the 'weather forecast' model, in which a stressful ontogeny is suggested to carry information about the adult environment, and the 'making the best of a bad job' model, in which adult behaviour represents a compensation for phenotypic limitations resulting from development). He then suggested a non-adaptive alternative interpretation of some of the empirical findings. Nettle concluded by considering what needs to be demonstrated to show that a developmental response is adaptive, and how we discriminate between competing adaptive interpretations of the same phenomenon.

The meeting was well attended and there was much discussion during the day and at the social event afterwards. This is testament to the high quality of the presentations and we thank our speakers for their investment in the process and for their clear and thought provoking talks. It was heartening to see colleagues from the various corners of our multidisciplinary subject interacting in such an animated fashion, and the satellite meeting clearly acted as an excellent stimulus package. For this gratifying situation we must also thank EHBEA, the Galton Institute and the Journal of Evolutionary Psychology (now Evolution, Mind and Behaviour) for financial support. We also owe a debt to Anna Rotkirch and her team in Helsinki for all the organizational work establishing rooms, refreshments and a social event thereafter. Without them none of this would have happened.

Tom Dickins
Professor of behavioural science at Middlesex University

The Galton Institute made a grant of £1,000 to this satellite meeting.

17th European Molecular Biology Laboratory PhD Symposium

Just By Chance? Randomness and Variability Shaping Biology

Heidelberg, 22 – 24 November 2015

The 2015 EMBL PhD symposium, as always organised by the EMBL first-year PhD students, tackled one of the most challenging questions in the life sciences. The matter of how intricate structures and systems are produced with great precision despite, or perhaps even because of, the stochastic nature of the underlying chemical processes, is central to a thorough understanding of biology.

Each day of the symposium highlighted a different aspect of this topic. The first day focused on proteins, structures and networks, the second on regulatory systems from cells to organisms, and the third on genetic variation and inheritance. These diverse and multifaceted topics were covered by twelve scientific expert talks approaching the issue from different perspectives, ranging from computational biology, biophysics and structural biology to genetics, evolution and developmental biology. Keynote lectures were given by Peter Fraser (Babraham Institute), Michael Levine (Princeton University) and Laura Landweber (Princeton University). An additional three speakers focused on inherent challenges faced by researchers and the interplay between science and the society. They addressed the issue of communicating science to a general public (Zoë Gamble, science communicator), publication ethics (Irene Hames, publishing consultant) and ethical considerations arising with new technologies (Jeantine Lunshof, researcher and bioethicist at UMCG).

The expert lectures were complemented by twelve short talks and two poster sessions in which early-career researchers, in particular PhD students, presented their work. These presentations were selected from abstracts submitted by the participants. In total, 201 early-career researchers and scientists attended the symposium, coming from 43 research institutes and universities in 18 different countries.

Overall, the 17th EMBL PhD Symposium was highly successful - its greatest strengths being the intermingling of different fields, and of well-established and early-career scientists. Participants and speakers were actively engaged in discussions, not only during the lectures and talks, but in smaller groups also during black board sessions, coffee breaks, lunch and dinner. These discussions involved current and future scientific questions, as well as the framework in which science is conducted, and life as a researcher.

The Conference Grant of £1,000 from the Galton Institute was an invaluable contribution to the symposium. It was used for the travel expenses of scientific experts from different fields and countries. We (the organising committee) are deeply grateful for this support. For pictures and more details of the PhD Symposium, please visit our website and social media channels.

Yonca Ural Blimke
PhD student, EMBL

Official website http://phdsymposium.embl.org/symp2015/
Facebook https://www.facebook.com/EMBL.PhD.Symposium/
Twitter @EMBLPhDSymp, #JustByChance
Instagram @phdsymposium

## Post doctoral travel grant

The Galton Institute has introduced a post doctoral travel grant, available to outstanding post doctoral researchers, normally within 6 years of receiving a doctoral degree, working in the field of genetics.

The Fellowship, which will be 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.

THE GALTON INSTITUTE

Conference 2016

## Environmental factors in gene regulation

To be held 16 November, 2016 in
the Wellcome Trust Lecture Hall at The Royal Society,
Carlton House Terrace, London

Speakers and topics:

Professor Sir Peter Ratcliffe, FRS
*Oxygen sensing and hypoxia signaling*

Professor Akhilesh Reddy
*Regulation of circadian clocks by redox homeostasis*

Dr Julie Gibbs
*Circadian regulation of innate immunity and inflammation*

Professor Jonathan Seckl, FRSE
*Early environmental regulation of glucocortoid receptor gene expression*

Professor Michèle Ramsay
*Fetal exposure leads to altered gene expression*

The 100th Galton Lecture:

Professor Dame Linda Partridge, FRS
*Nutrition and lifespan*

Professor Anne Ferguson-Smith
*Parental nutrition can modify gene expression in the offspring*

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Admission is free, but strictly by ticket available from:

The General Secretary at: executiveoffice@galtoninstitute.org.uk