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ADELPHI GENETICS FORUM

exploring heredity and society

Adelphi Review

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Adelphi Genetics Forum, 19 Northfields Prospect, London, SW18 1PE

Tel: 020 8874 7257 www.adelphigenetics.org

General Secretary: Mrs Betty Nixon

executiveoffice@adelphigenetics.org

Review Editor: Mr Robert Johnston
Charity No: 209258

EDITORIAL

One of the stated aims of the Adelphi Genetics Forum is to promote “education and communication with all interested individuals and groups”. One of the most significant groups is secondary Biology teachers which is why, in 2015, we held our first Teachers’ Conference aimed at improving their knowledge and understanding of modern genetics. This year marked the fifth such conference and it was our most successful yet with over 70 attendees. You can read a full account of the event on page 10 and look at the various presentations on our website.

Another of our aims is to promote “the study and understanding of “the historical origins and development of human heredity”. In this issue there are three articles which do exactly that. On page 4, we have the remarkable story of Lionel Penrose as told by his daughter **Professor Shirley Hodgson**, a current Trustee and leading geneticist in her own right.

Then on page 18, we have a unique insight into early genetic diagnoses by **Professor Ian Jackson** from his days at St Mary’s Hospital. It appears to be a story that has lain hidden for more than 40 years!

Finally on page 14, we have a report from the Progress Educational Trust of a meeting to mark the 100th anniversary of the birth of Baroness Mary Warnock, who led the original Human Fertilisation and Embryology Committee in the 1980s which shaped research law in this field in the UK and across the world.

Robert Johnston

My Life in Genetics

An Interview with Professor Shirley Hodgson
Trustee of the Adelphi Genetics Forum

Shirley and her father on her wedding day

My father, **Lionel Penrose**, was a hard act to follow!

Before I was born, he had spent seven years from 1931 working at the Royal Eastern Counties institution in Colchester, which housed about 1380 individuals who had been admitted because they were “mentally deficient” (the

contemporary phrase), although in some cases they had just been convicted of some felony. At the time, the “condition” designated as mental deficiency was poorly understood, and sometimes included poverty and alcoholism. This lack of understanding of the causes of this supposed single condition underpinned the concepts of eugenics. Lionel was tasked to identify the causes of mental deficiency. He set about examining all the patients in detail, arranging IQ tests and interviewing (and testing) their parents and 6629 siblings. He also took detailed family histories. His observations from this study were original and prescient. He noted that the parents of severely mentally deficient patients tended to have relatively normal IQs, whereas those of mildly affected individuals tended to have lower than average IQs, implying a multifactorial inheritance of the milder cases but monogenic genetic causes of severely affected individuals. This was further supported by the increased incidence of other affected patients in the sibships of severely affected patients. He found 63 cases of Down’s syndrome, and developed methods of quantifying the physical characteristics of this condition using palm and fingerprints which were measurable. His work showing that increased maternal age was associated with an increased risk of Down’s syndrome was pivotal and involved complex mathematical studies to show that it was maternal age rather than paternal age that was the issue. In addition, he showed that some, often young, mothers of Down’s syndrome children had some characteristics of Down’s syndrome, suggesting that they were mosaic for the condition.

Lionel was able to diagnose some cases of Tuberose Sclerosis (TS) amongst the patients, and he noted that the condition was inherited as an autosomal dominant trait characterised by variable penetrance. This phenomenon had not previously been well documented, and was ascribed to modifier genes. He also

calculated the mutation rate of this condition, predicated on the fact that many cases appeared to be de novo. He later contributed to the observation that increased paternal age was associated with increased mutation rates.

He identified some cases of phenylketonuria, recognised that it was an autosomal recessive condition, and developed an assay to measure phenylketones in the urine. He postulated that it might be possible to treat this condition with a low phenylalanine diet. His first attempts to do this were successful in reducing the level of phenylketones in the urine, but the diet was insufficient in calories to be sustainable, and he was informed that a fully adequate diet would be prohibitively expensive, so he had to give up this idea.

My family moved to Canada in 1939, where my father had two different jobs, and apparently had to write letters to himself from time to time! My mother became pregnant unexpectedly and I was born when she was 44. My father, concerned about the risk of Down's syndrome, rushed to the maternity ward to see me, examined my palms etc. and exclaimed to the astonished midwives "I don't THINK it's an imbecile!".

In 1945 my father was encouraged by J.B.S.Haldane to move to London to take up the Galton Chair of Eugenics at UCL. He agreed, but from the start he did everything in his power to change the name of the department from eugenics to human genetics, but due to bureaucratic issues he could not achieve this until 1963. He was completely opposed to the concept of eugenics, and at least he was able to change his notepaper immediately to remove any reference to eugenics, and change the title of the "Annals of Eugenics" (the departmental journal), to the "Annals of Human Genetics".

His opposition to eugenics was apparent to all who worked with him, and he told the eugenics society in a lecture he gave that "anyone who proposed to eliminate all those who did not belong to a pure stock could be considered to be a lunatic, and fortunately human lunatics are variable and do not have the same delusions". His opposition to eugenicists' ideas was based on their assumption of the superiority of certain types of people over others (noting that eugenicists perceived themselves as superior types), and clearly such judgements belittled the others, which he decried. He also explained that the proposed sterilisation of undesirable individuals by eugenicists would often be irrelevant for their aim, either because the individuals concerned were unlikely to have offspring, or because they had an autosomal recessive condition. He stated "to eliminate the gene for phenylketonuria would involve sterilising 1% of the population, and only a lunatic would advocate such a procedure to prevent the occurrence of a handful of harmless imbeciles".

The identification of the causes of mental deficiency did a lot to debunk the eugenic idea that alcoholism and poverty were inherited. Lionel was very fond of individuals with Down's syndrome and other people with such handicaps. He felt that such individuals should be respected and given the opportunity to fulfil their potential and contribute to society within their capabilities.

So what about my career? When I was about 16, I reluctantly gave up my desire to become a ballet dancer, because I wanted to "do good", so I became a doctor. However, I was emphatic that I could not become a geneticist because my father was so well known in the field. At the suggestion of a career advisor, I planned to become a GP because I was informed

that that was what female doctors did. However, I was sacked from a rather religious practice for prescribing the pill to unmarried women. Luckily, my ever helpful husband found an advertisement for a Locum position in clinical genetics at Guy's hospital working for Paul Polani, so I took that up, and was completely hooked! In order to take up the substantive post at Guys, I had to take membership, which I did, and returned to Guys.

In the late 1980s I became interested in the idea that cancer susceptibility could be inherited, and how this knowledge could help reduce the cancer burden in susceptible families. To my astonishment I found that my father had written a paper in 1948 which showed that the close relatives of women who had died of breast cancer had an increased risk of the condition (about a two-fold increased), but the idea that such a susceptibility could be inherited had taken some time to be generally appreciated.

I was impressed by the family cancer clinics run by Joan Slack (with Vicky Murday) which were being initiated in the 1980's. I moved to a consultant clinical genetics post at Addenbrooke's Hospital in 1990, and at about that time my husband (a gastroenterologist) had a patient with young onset colon cancer, who had already had uterine cancer. He noted that she resembled my cousin's wife Suzanna, and it turned out that his patient was her mother! After some hesitation I suggested that Suzanna go to see Joan Slack, as I suspected she might be at risk of the newly delineated condition Lynch syndrome. She did, and a screening protocol was planned, but as she was just pregnant with her third child, she put the screening off for a while. Tragically she developed ovarian cancer and later died.

I was immensely affected by this, and decided I must dedicate

the rest of my working life to cancer genetics. Eamonn Maher was a registrar in the department at that time, and we agreed to write a textbook, "A practical guide to human cancer genetics". This went some way to helping me towards my aim. I returned to Guy's as a Reader, where I developed the evolving S. E. Regional cancer genetics service, and did clinics for individuals with familial adenomatous polyposis and those with a family history of bowel cancer or Lynch syndrome at St. Mark's Hospital. Later when I went to St. George's to a chair in cancer genetics, I was able to help to develop the S.W. Regional cancer genetics service.

I was incredibly fortunate to receive support from Sir Walter Bodmer at the ICRF, to pursue research in cancer genetics. This enabled me to fund research fellows including Shehla Mohammed, Louise Izatt, Mark Tischkowitz, Ian Frayling, Andrew Beggs and Julian Barwell, who have all flourished and become important figures in cancer genetics services and research.

So what did I learn from my father that helped me develop my career? Persistence was I think a characteristic I inherited, and a desire to help people, which was a strong feature of my father's Quaker background. Also, I remember he was very keen to emphasise that I should never accept any ideas just because other people did, I must make my own judgement based on the evidence I had myself.

I am sad that my father did not live long enough to know about my work in cancer genetics. It would have been fun to hear his views.

Shirley Hodgson

Previous contributors to the *My Life in Genetics* series:

Published in the ***Adelphi Review:***

Dr George Burghel	**Issue 5**
Dr Helen Middleton-Price	**Issue 4**
Professor Nick Mascie-Taylor	**Issue 3**
Mr Robert Johnston	**Issue 2**
Dr Jess Buxton	**Issue 1**

Published in the ***Galton Review*:**

Professor Nicholas Wood	**Issue 15**
Professor Dallas Swallow	**Issue 14**
Professor David Galton	**Issue 13**
Professor Andrew Read	**Issue 12**
Professor Veronica van Heyningen	**Issue 11**
Professor Dian Donnai	**Issue 10**
Professor Philippa Talmud	**Issue 9**

Recent Advances in Genomics: A Fifth Teachers' Conference NOWGEN Centre, Manchester 28 June 2024

More than 70 secondary teachers attended this conference, run by the Adelphi Genetics Forum, aimed at updating their knowledge and understanding of some challenging topics in this field. The day was organised and chaired by our Librarian, **Robert Johnston,** who started the day by briefly describing the history, aims and activities of the Adelphi Genetics Forum.

The first speaker was our treasurer, **Professor Andrew Read** from the **Manchester Centre for Genomic Medicine**. His talk

was titled **'Principles of DNA Sequencing'** in which he described in detail the various methods from the original dideoxy sequencing through to the latest techniques including PacBio and Oxford Nanopore. This is a fast-moving field but Professor Read had all the information at his fingertips and was able to give lists of advantages and disadvantages for each system.

Following a coffee break, **Jessica Keen,** who is the **Pharmacy Lead at NHS North West Genomic Medicine Service Alliance,** spoke about **'The Implementation of Pharmacogenetics in the NHS'**. She discussed the importance of using genetic information from patients to ensure that they receive the most appropriate drug therapies, at the optimum dosage, to best treat diseases and prevent adverse reactions. She described various health conditions which have benefitted from such studies and which have also proved to be more cost-effective for the NHS.

The next speaker was **Dr Garrett Hellenthall** from **UCL Genetics Institute** who discussed **'Studying Historical Movements of Populations using DNA'**. He began by stating that studying the ancestral history of modern humans is difficult because it is impossible to accurately predict the dates of various events. However, as humans began to move out of Africa and across the globe, there were genetic consequences that can be seen in the genome.

Human genomes are 99.9% identical in sequence but that still means that 3 million nucleotides can vary between individuals. The greatest diversity in genomes can be found in Africa since humans have been there longest while more recently populated continents show less diversity as the population has evolved from a smaller founding group who have had less time to develop. Related individuals in any population

share segments of DNA but, as generations continue, the size of shared segments gets smaller. It is therefore possible to compare DNA from apparently unrelated people to see how long ago they were closely related. It then becomes possible to find correlations between the geography and genetics of populations.

After lunch **Andrew Walton**, the PhD student at UCL whom the Adelphi Genetics Forum is sponsoring, gave an interesting talk on his **'Thoughts on the Teaching of Genetics in Schools'**. He questioned whether the Mendel-based approach is the right

way or does it oversimplify the subject and encourage a belief in 'genetic determinism' ie the idea that our characteristics and abilities are predominantly genetically determined and thus immutable. He also suggested that miscommunicated genetics has

been used to boost the 'racial realist movement' and promote scientific racism. He suggested that the assertion that genetics is determinist and proves that race is a relevant biological category has its roots in the eugenics movement. He suggests that a better approach might be to stress the polygenic nature of many traits and the complexity of the Genotype-Phenotype relationship.

The next speaker was **Dr Rachel Thompson** from the **Centre for Human Genetics at the University of Oxford** who talked about the challenging subject of **'Ethical Issues of Genomics in Healthcare'**. Her role is linked to the Centre for Personalised Medicine and she began by considering some clinical examples which demonstrated the complexity of decision making with regard to consent for treatments, especially in children. She went on to consider the challenges of 'therapeutic misconception' where the boundaries between valuable research and clinical care become blurred.

The final talk was given by **Chris Watt**, **Principal Clinical Scientist at NHS Cancer Genomics in Manchester**. He spoke about **'Genomic Testing for Cancers'**. He explained the differences between Germline and Somatic variant testing and described some examples of each. He listed the various somatic cancer services carried out by the NHS and what they involve. Interpreting variants is challenging but national guidelines are available to ensure that different hubs follow the same procedures. He also described the recent development of using 'liquid biopsies' for earlier detection of cancers.

Copies of the presentations given can be found on our website at: https://adelphigenetics.org/events/teachers-conference-2024/

Robert Johnston

Progress Educational Trust
Mary Warnock at 100: The Architect of Embryo Law
17 April 2024

This PET event explored the life, work and legacy of Baroness Mary Warnock (1924-2019), marking the 100th anniversary of her birth.

Mary was a philosopher – and Patron of PET – and led the Government's Committee of Inquiry into Human Fertilisation and Embryology whose 1984 report went on to shape fertility and embryo research law, both in the UK and around the world.

A range of expert speakers, including current and former Chairs of the UK's fertility regulator – the Human Fertilisation and Embryology Authority (HFEA) – discussed the impact of the committee report, and of Mary's work more broadly. Those panellists who knew Mary also shared insights into Mary as a person The event attracted **151** attendees representing **81** organisations.

The first speaker, **Dr Duncan Wilson,** from the University of Manchester's Centre for the History of Science, Technology and Medicine, elaborated on the reasons why Baroness Warnock had such an influence on public policy. Baroness Warnock positioned herself (and bioethics more generally) as an intermediary between the public, professions, and politics, and was pragmatic when it came to developing guidelines for research. She sought to achieve compromise in areas where there was no obvious answer – exemplified by the 14-day rule, which was presented as both a scientific and philosophical

landmark. It helped also that Baroness Warnock was seen as an 'outsider', said Dr Wilson.

The next speaker, **Anna Mastroianni**, professor of Bioethics and Law at the Berman Institute of Bioethics at Johns Hopkins University, spoke about the influence Baroness Warnock had on US bioethics and fertility policy. While not necessarily direct, and difficult to spot in the text of policy documents (the anti-abortion movement in the USA held great sway), Baroness Warnock's work impacted on those sitting on committees, who recalled their engagement with the Warnock Report, and on public policy discourse more generally. The Warnock Report became a reference point for lawyers, scientists and policymakers involved in debates about reproductive policy and stood out among the many parallel efforts in ethics, law and policy at the time. Baroness Warnock highlighted the importance of cultivating public trust in the work scientists were doing, said Professor Mastroianni.

Next, **Baroness Ruth Deech**, crossbench peer in the House of Lords and former chair of the Human Fertilisation and Embryology Authority (HFEA), remembered Baroness Warnock as an assured and decisive person, whose philosophy (cultured at the University of Oxford) had a lasting, practical impact on the issues she was involved with. Her influence in Parliament was beneficial and marked, said Baroness Deech, while outside Parliament she promoted IVF laying the foundations for 30 years of progress in fertility and embryo research. She concluded by describing Baroness Warnock as the original quango queen and an established member of the Great and the Good, to the benefit of all of us.

Finally, **Julia Chain**, the current chair of the HFEA, spoke about Baroness Warnock's vision and commitment to public consultation in the context of the current review of the UK's fertility law.

She said the HFEA is building on Baroness Warnock's ability to achieve ethical consensus as it considers new issues that were not contemplated forty years ago. Baroness Warnock was a visionary of her time, said Chain, testament that her framework is still being used today – yet there is work to be done to ensure it remains updated for our time and beyond.

In the questions that followed the speakers were asked what we can learn from the UK amid the polarisation of debate in the USA, particularly following recent changes to abortion law, and, in turn, what lessons could the UK learn from the USA. Professor Mastroianni highlighted the importance of carrying out public consultations to help bring people along and to both recognise and be attentive to dissenting views. Chain said it was difficult since there is little consensus in the USA and rules are different from state to state, but she agreed with the importance of public consultation. Professor Mastroianni added that the USA lacks a venue where ideas can be shared in a respectful way.

The speakers were also asked if there is a need for another Warnock Report. Chain replied that the HFEA currently has a suitable committee structure and the mechanisms needed to enable discussions with the right people and has access to advice without needing a separate commission. On whether it is possible to have HFEA equivalent bodies communicating jointly across the world, Chain said the HFEA is a 'gold standard' for other countries and is willing to share best practice but international agreement on even less controversial issues would be impossible. Professor Mastroianni and Dr Wilson pointed out that both in the USA and Europe there is too much variance between states and countries, each wanting to do things differently, such that coordination would not work.

Finally, the speakers were asked if it remains important for non-specialists to be involved in discussions about future, complex technologies. Dr Wilson highlighted that is necessary and vitally important to have non-expert involvement and this was the core of what Baroness Warnock argued. Chain agreed, pointing out that at the HFEA those involved with law and policymaking need to be able to understand the technologies, so experts need to be able to explain them in lay terms. Felix Warnock added that his mother would often say she was well qualified to chair these committees specifically because she was a non-expert. The philosophical approach is to challenge expert opinions, he said, and ask tricky questions without necessarily having knowledge of the background subject matter. Indeed, he said, professionals need someone to identify issues and present questions.

As a result of attending the event the audience learned much more about Mary's life and work on the committee and her continuing influence in the field of assisted reproduction inside and outside the House of Lords, her impact on bioethics and the influence of her work outside the UK. PET was pleased that some of Mary's children attended the event and honoured that her son Felix Warnock accepted an invitation to join the panel.

This event brought home the importance of doing so and Baroness Warnock's lasting legacy in the field of fertility and embryo research. PET are grateful to the **Adelphi Genetics Forum** for providing funds that helped to make this such a successful event.

Dr Antony Starza-Allen
University of Surrey

A DNA Prenatal Diagnosis in the 1970s

By Ian Jackson, University of Edinburgh

In the late 1970s, when I was a PhD student, I carried out what is almost certainly the first DNA based prenatal diagnosis in the UK, and one of the first in the world, but it has never been documented.

During my undergraduate degree at Oxford I met a young Bob Williamson, then in his late 30s, who had just moved from Glasgow to St Mary's Hospital in London. His work towards isolating human genes, in particular disease genes (which had not yet been done) inspired me to apply for a PhD with him and I was accepted. The lab was full of very motivated students, postdocs and technicians. Work in the lab was coupled with an active social life. The Medical School bar was one floor below the lab and opened at 5pm, whilst Friday lunchtimes were spent in the local pub; a tradition Bob had brought down from Glasgow.

Peter Little had come to St Mary's from Ed Southern's Unit in Edinburgh and had success making a restriction site map of the human beta and gamma globin genes in collaboration with Dick Flavell in Amsterdam. It would be fair to say it took some time to get Southern blotting to work in St Mary's. Hybridisation was in workshop made Perspex boxes over a weekend, set up

after that Friday lunchtime in the pub but most efforts turned out to be completely black or completely blank. However, the focus was on developing the technology to clone human genes. Initially the technique was inefficient so the relevant

The St Mary's lab around 1979. Left to Right: Rob Elles, Raymond Dalgleish, Gill Annison, David Westaway, Ian Jackson, Mike Courtney, Peter Little. Foreground: mysterious column with pump on lab stool.

restriction fragments were enriched from milligrams of DNA using the Ed Southern designed "gene machine", electrophoresis in a wedding cake size cylinder of agarose running for days, or by differential retention on columns of mysterious matrices

powered by a pump balanced on a lab stool. Eventually the library making became more efficient and we were able to clone partially-digested DNA fragments into lambda phage vectors. At some point Tom Maniatis's human library arrived from Caltech so cloning normal human genes became relatively straightforward. But we were interested in disease genes.

Bob had a collaboration with Bernadette Modell, who ran the haemoglobinopathies clinic at UCH and had for a number of years characterised beta-thalassaemia patients. Painstaking hybridisation experiments had classified the diseases into beta-0, in which no beta-globin mRNA was detected or beta-plus, when some mRNA was present. As an aside I eventually cloned a beta-0 globin gene which on sequencing turned out to have a premature stop codon. My benchmate and flatmate David Westaway cloned a beta-plus which he found on sequencing to have an intronic mutation creating a new splice acceptor site.

Through the collaboration with Bernadette, I sat in on one or two of her clinics and it was inspiring to see how these very young patients were managing their care which involved many blood transfusions coupled with frequent injections of, and even overnight infusions of, deferoxamine to chelate out the iron overload caused by the transfusions. Bernadette had developed, with UCH colleagues, a method of sampling the cord blood of foetuses for prenatal diagnosis of haemoglobinopathies, which would give the mothers an option of termination if desired, but which carried quite a risk of causing miscarriage of unaffected foetuses.

At some point in 1978 Bob Williamson took a short sabbatical in Margaret Buckingham's lab in Paris and while he was away Bernadette phoned with a proposal. A couple in her care had a

child suffering from sickle-cell disease who had sadly died. The mother had very soon after become pregnant but did not want to go through the risky foetal blood sampling procedure. However, she was happy with, and consented to, an amniocentesis.

Y.W. Kan in San Francisco, had just published that the sickle-cell mutant beta globin gene (HbS) was found on a HpaI restriction fragment of 13kb, compared to either a 7 or 7.6kb for the non-mutant (HbA) allele. Bernadette's proposal was that we do a Southern blot on amniotic fluid cells to diagnose the genetic status of the foetus. However, it was not so straightforward. Kan had reported that HbA was occasionally carried on the 13kb fragment and the mutant HbS could also be sometimes found on the 7.6 kb fragment. In retrospect, and published soon after, it is clear that the sickle-cell mutation had occurred more than once, and on different haplotypes. I don't recall now whether Kan's 1978 paper in the Lancet reporting his DNA based prenatal diagnosis and the associated statistics had been published by this time. Nevertheless, I called Kan in California to ask his advice and he gave us their data on probabilities of the 13kb fragment carrying the sickle mutation.

For some reason I never thought to contact my supervisor, Bob, in Paris. No email then, of course; a letter would be slow and we didn't consider a phone call, despite my calling California. I did however discuss it with a postdoc., Emma Whitelaw, and we worked together.

We had blood DNA from the parents and, once we'd sourced HpaI "urgently" from the supplier and did a Southern blot (now working a bit more reliably) we could show both were heterozygous, 13kb + 7.6kb. We felt a lot more confident of a reliable diagnosis at this point. The amniotic fluid arrived in two 30ml tubes. I was surprised by the volume, and by the pellet of cells

once we'd spun them down. We isolated some tens of micrograms of DNA, enough for at least one backup should the first blot fail. Nevertheless, the first one gave a clear result, the foetus was also heterozygous for the two alleles like the parents, and was most likely heterozygous for HbS. We passed on the news to Bernadette who was able to give the mother some reassurance and we celebrated briefly (probably in the bar below the lab) and went back to cloning human genes!

My memory is that Bernadette had said before we did the analysis that the mother, despite consenting to the amniocentesis, had indicated that she would not request a termination of an affected foetus, given the recent death of her child. This somewhat reduced pressure to get the diagnosis right, although who knows what the mother's reaction would have been if we had indicated a high probability of a homozygous child. Several months later the child was born and haematology confirmed the heterozygous status.

Bernadette credits this demonstration of DNA diagnosis as convincing her obstetrician colleagues that it was worth developing chorionic villus sampling whereby foetal DNA could be obtained much earlier than by amniocentesis. A much earlier diagnosis is always preferable. In particular, diagnosis leading to termination before the mother is showing signs of pregnancy is culturally desirable in many communities, and once CVS and DNA analysis were established, prenatal diagnosis was more widely embraced.

My thanks to Bernadette Modell for helpful discussion and comments.

Ian Jackson
University of Edinburgh

CHASE Africa final report to the Adelphi Artemis Trust

The purpose of this project was to improve access to family planning information and services in marginalised rural communities of Eldama Ravine, Baringo County, Kenya, enabling people to recognise and realise their sexual and reproductive health rights. In line with the objectives of the Adelphi Artemis Trust, the project successfully assisted poorer communities in rural Kenya with a) the advancement of education in reproductive and sexual health, as a foundation for b) the provision of fertility control (non-coercive, voluntary family planning).

In the early stages of the project, the COVID-19 pandemic necessitated significant changes to the service delivery methods in line with rules and regulations, to limit the spread of the virus. As such, the original plan to run large-scale mobile health clinics attracting large crowds had to be revised. Instead, the focus shifted to home visits made by Community Health Workers (CHWs) and a referral system linked with health facilities and newly established 'safe spaces'. These were rented rooms in remote communities, used as a base by a nurse who attended each month.

This modification of the project was successful and proved highly effective for increasing the depth of people's awareness and understanding of modern contraceptives. This led to a high percentage of those reached with information choosing to try a modern family planning method. There were disadvantages that came with the suspension of the mobile clinics. These included a) a significant reduction in the overall number of people to hear family planning messages, b) a reduction in the capacity to pro-

vide a range of other health services, and c) a loss of anonymity for those women who were keen that a family planning intervention remained confidential. In contrast to the mobile clinics, women may know the staff at local health facilities and be more uncomfortable attending those facilities to seek contraception.

Another challenge faced throughout the project was the extremes of weather, from heavy rain to severe drought, which affected people's availability for engagement in discussion on the topic of family planning. In the project area, people are directly and immediately dependent on their local environment for food, water and fuel. Their activities are largely dictated by the weather and seasons, and periods of rain bring the urgency of growing crops, while dry periods bring the need to search for water. When people are pre-occupied with the challenging, time-consuming tasks of necessary daily subsistence, finding a suitable opportunity to be able to draw someone's attention to the matter of family planning and its benefits is a delicate act. Despite significant challenges, the project still managed to achieve good results, providing life-changing family planning information and services to thousands of individuals and families in Eldama Ravine.

Community Health Worker sharing SRHR information at Dandelion Safe Space

The overall numbers of services provided during the three year project were as follows: Number of people reached with family planning information: Actual - 37,569 (Projected - 102,000). Fewer people than we had anticipated were reached with family planning information due to the suspension of large mobile clinics, however, the level of awareness and understanding achieved by those who were reached was much greater due to the more personal, revised system of door-to-door visits. Number of contraceptive services provided to women: Actual - 13,180 (Projected - 9,720). More women than anticipated chose to use family planning, due in part, perhaps, to the higher level of understanding achieved with in-depth dialogue in people's homes, and in part due to the high demand for family planning created by people's particular desire to avoid pregnancy during the pandemic and drought.

The contraceptive services provided equated to 19,954 'Couple Years Protection' (CYP). Each Couple Year of Protection is a year's worth of protection from pregnancy. (This differs from the total number of contraceptive services provided, because some contraceptive methods last 1 month or 3 months, and some others last 3 years or 5 years). The number of first-time users of modern contraceptives was 7,384. Family planning information was shared with 118 people living with disabilities. 26 women living with disabilities chose to use a modern family planning method.

In addition to the family planning services, 5049 other primary healthcare services were also provided. These services included treatment of minor ailments, deworming, immunisations, HIV/AIDS testing and counselling, and cancer screening. Total cost of project: Actual £102,757 (Projected £161,721). The total project expenditure was reduced due to the suspension of large mobile clinics, and as such, the cost per family planning

service provided was lower and the revised service provision can be considered to have been very cost effective.

It is important, however, to remember the value of the large mobile health clinics, when they are permitted, for drawing people to a place where they have the opportunity to hear about family planning, and as a smoke screen to enable access to family planning for women whose family and friends do not support their choice to use modern contraceptives. Our intention is to remove all obstacles to accessing family planning, and large mobile health clinics continue to play their important role in that.

The project has been successful in reaching thousands of people in Eldama Ravine with family planning information, and increasing people's awareness and understanding of modern contraceptives, enabling them to make informed choices about their reproductive health. The project also reached thousands of women with family planning services, enabling them to action their choice to use contraceptives.

Behind the numbers is a real and tangible opportunity for change in the lives of each woman and her family. Each Couple Year of Protection (a year's worth of protection from pregnancy) costs on average £5.15 to deliver through this project. Such a small cost has the potential to make a huge difference to people's lives, and on behalf of the thousands of women and families reached through this project, we thank the Trustees of the Adelphi Artemis Trust for your support.

CHASE Africa

ADELPHI GENETICS FORUM

Conference 2024
The Royal Society - Wednesday, 16 October, 2024

Progress and challenges of implementing genomics into practice and society—the first 20 years

October 2024 marks 20 years since the publication of the completed sequence of the Human Genome Project (HGP) in *Nature*. The International Human Genome Sequencing Consortium's outstanding achievement gave rise to great hopes, with the expectation that it would enable 'researchers around the world to conduct even more precise studies of our genetic instruction book and how it influences health and disease'. How much of this prediction has been realised in the intervening years? This conference offers an opportunity to hear about some of the achievements, hurdles and failures of the intervening period from a variety of perspectives.

Speakers:

Anne-Ferguson Smith: *Epigenetic inheritance - models and mechanisms*
Bill Newman: *Implementing pharmacogenetics at scale in clinical practice*
Michael Parker: *The changing moral life of genetics and genomics since the Human Genome Project*
Andrew Read: *The Human Genome Project - 20 years on*
Fergus Shanahan: *No stool left unturned-why our microbiomes differ*
Steven Sturdy: *The fortunes of medical genomics: a quarter century of promise*
Clare Turnbull: *Genomics in population screening for cancer: opportunities, challenges and cautions*
Sarah Wynn: *The promise and challenges of genomics for patients and families affected by rare conditions*