Obituary

Professor Sue Povey (1942- 2019).

Former Editor-in-Chief of the Annals of Human Genetics, died January 11th 2019

Professor Dallas Swallow and Professor Veronica van Heyningen

Professor Sue Povey had a distinguished research career at University College London. She was also editor-in-chief of this journal (from 2000-2007) but had an even longer-term involvement with the Annals, acting as a member of the editorial board for more than 25 years. Many readers will be familiar with aspects of her life and work. Some of you will have collaborated with her over the years, but perhaps may not be aware of the breadth of her contributions, which ranged over a wide spectrum of topics.

Sue qualified with degrees in Genetics and Medicine at Cambridge and UCL. Her clinical training included work with Save the Children Fund in Algeria, which she found rewarding but challenging. Her plan had been to specialize in paediatrics, but genetics had grabbed her interest while she was still a student. An overland student trip in a Landrover to India had resulted in her coauthoring a paper on the genetics of leprosy. That experience, together with a short research stint in Harry Harris' lab at UCL, inspired her to decide that her future lay in human genetics research. After completing her clinical training, she joined Harry Harris's group at the MRC Human Biochemical Genetics Unit, which formed part of the Galton Laboratory, UCL, as a staff member.

Sue's first successes involved exploiting the enzyme detection systems, developed by Harry Harris and David Hopkinson among others, in the context of somatic cell genetics. Starch gel electrophoresis (described by a former student as electrophoresis in mashed potato) allowed the distinction of enzymes originating from different species, as well as different individuals. Using somatic cell hybrids, in collaboration with Walter Bodmer's group in Oxford, Sue contributed to mapping many human genes to individual chromosomes. This approach led her to coauthoring, with many groups from all over the world, more than 60 papers mapping different genes, before the human genome was fully sequenced. Collaborating with John Evans' group in Edinburgh she evaluated the stability of long term human cell lines in culture. An extremely practical outcome of this was the recognition that many cell lines used for research had been taken over by other more strongly growing cell lines, including the now well-known cell line HeLa. This discovery was not popular, sometimes resulting in the need for reinterpretation of experimental data, some of which had already been published; but led to the all important routine of regular checking of the identity of cell lines, and better cell culture practices.

Her group was quick to adopt the new DNA technologies as soon as they appeared, which were then applied to a variety of genetic problems. A remarkable early research contribution, involving both enzyme marker and DNA detection, came out of her

collaboration with Sylvia Lawler and others, and led to the understanding of the genetic origin of hydatidiform moles and ovarian teratomas.

Despite not practising as a clinician, Sue kept up her registration until quite recently and was motivated throughout her career by a strong interest in human disease and the desire to help patients and their families. Together with Peter Cook, she was able to explain her own niece's liver disease that sadly took her life at the age of 13, ascribing it to deficiency of alpha-1-antitrypsin. As well as delivering early diagnostics for this gene, she went on to contribute significant research in this area, and the genomic DNA that Gavin Kelsey in her group had cloned was used to make the first transgenic sheep (Tracey) that produced this protein in its milk.

As DNA technology advanced, the pace of mapping the human genome speeded up. Sue went to every Human Gene Mapping Workshop meeting, where she worked ferociously. She immediately recognised the vital need for proper gene nomenclature and gene annotation. She took over from Phyllis McAlpine as chair of the International Human Genome Organisation (HUGO) Gene Nomenclature Committee (HGNC) in 1996, until her official retirement in 2007, when her colleague, Elspeth Bruford became coordinator. THE HGNC activities were initially funded by the US Department of Energy, but Sue was successful in gaining further funding from the US National Institutes of Health, as well as the UK Medical Research Council and the Wellcome Trust, some of which still fund the HGNC to this day. This enabled her to build up a team of postdocs and programmers for gene naming during the incredibly intensive, but exhilarating, time of the Human Genome Project (HGP). This was a challenging and controversial activity. Names and acronyms had to follow strict guidelines and be unique. Many genes had been named first in Drosophila and had been given names that reflected the mutant phenotypes which led to their discovery: for example, Sonic hedgehog, Lunatic Fringe etc. Sue had to explain patiently to the community that some of these names just could not be used for human genes; there was no way that one could explain to a parent that their child carried a mutation in Lunatic Fringe!

Under Sue's leadership over 20,000 human genes were given a name, including many on her favourite chromosome 9, and she was delighted to be an author on the publication of the sequencing of that chromosome as part of the HGP. Sue's attachment to chromosome 9 had begun after being approached by John Osborne to collaborate in the study of the disease tuberous sclerosis complex (TSC). They were able to show linkage to the gene controlling the ABO blood group system on that chromosome. However, not all families showed this linkage, but with additional DNA markers linkage to chromosome 16 was demonstrated in some families. The race was then on to identify the two different TSC genes. Sue was always ready to collaborate, despite the challenge of intense international rivalries, and her group was one of eight different labs that co-authored the paper identifying the gene on chromosome 9 in 1997. Her involvement with TSC continued until her death. She set up and managed the TSC variation database, an invaluable international resource, now run by her collaborator Rosemary Ekong. In recent years she worked closely with the Human Variome Project and made a huge and much appreciated contribution to developing ethical guidelines for maintaining confidentiality, while also allowing data to be shared for the benefit of other patients, diagnostics and research.

Sue Povey had a helpful and cooperative approach to life and science. She showed an unusually high level of concern for and care of friends and colleagues in need, making her house, her time and much more, available - always rising to the occasion. She could spend hours of her time solving other people's problems, and willingly took on committee and advisory work. But she always stood firm in defence of her principles, taking on a whole committee if necessary. She was a key member of the Human Genome Organisation (HUGO) from 1996 until recently, and a driver in several other organisations, relating both to her work and her personal life. After her official "retirement" she acted as Chairman of the Board of Governors at a school for children with learning difficulties.

Sue was strongly supportive of her staff and students, giving them opportunities not afforded to other young researchers. To enable them to go to international meetings, she would often travel at very low cost herself, despite the inconvenience, or indeed pay out of her own pocket. Several of these students became full professors, or took leading roles in their careers. Many were female. She herself had been brought up by her medical mother to believe that women could achieve whatever career they wanted but should not learn to sew!

Sue was unfailingly dedicated to her work, but allowed time for her love of birds, flowers, gardening, hill walking and walking with dogs. She rarely took proper holidays, though sometimes visited colleagues in Romania, Australia, USA, Iceland and elsewhere, combining scientific commitments with exploring the countryside. One holiday to the Galapagos was, she said, the experience of a lifetime.

She will be much missed by her friends and family as well as her extended scientific family.

Margaret Susan Povey, known as Sue
Born 24 April 1942
Died 11 January 2019