PROFESSOR SIR PETER DONNELLY

Doctor of Science, *honoris causa*

Peter Donnelly is foremost amongst researchers whose work has underpinned the genomics revolution of the last two decades. His fundamental insights into modern genomics/genetics have elucidated a new understanding of the genome and its role in biology and human disease and enabled countless other research projects. He was central in developing the methods underpinning genome-wide association studies (GWAS), as well as leading many of the pioneering GWAS that have transformed our understanding of the genetic basis of disease. Recently he has pioneered the use of whole genome sequencing in clinical medicine. Genomics is beginning to have an enormous impact on clinical medicine with great promise to revolutionise diagnosis, patient care and the development of therapies. Peter's work has been central to these advances.

Peter did his BSc (Hons) in mathematics and statistics at the University of Queensland before taking up a Rhodes Scholarship to study for his doctorate at the University of Oxford. A statistician by training, he made considerable contributions to that field before broadening his interests to apply rigorous statistical approaches to genetic data. His work is wide-ranging. A short list includes:

(i) Fundamental contributions to developing the mathematical/statistical methods underpinning much 21st century population and disease genetics research. Early work on the coalescent revolutionised population genetics modelling. Several of these methods are exceptionally highly cited and have received significant awards.

(ii) Key insights into understanding variation in human populations and its consequences. Peter has been a leader of ground-breaking large-scale genomics studies and the development of the mathematical methodologies which underpin them, such as (a) *Genomic variation*: Peter has been central in developing many of the most important data resources in the world for studying human genetics; (b) *Human disease*: Peter led the Wellcome Trust Case Control Consortium (WTCCC) studies, which won numerous awards; and its follow-up, WTCCC2: 15 GWAS studies across >65,000 samples, 11 papers in *Nature/Nature Genetics*; and (c) *Human demography*: The 'People of the British Isles' study is widely considered a landmark study in human genetics. With Stephen Leslie (University of Melbourne), Peter was central to producing the first fine-scale genetic map of a human population, with major insights into UK history and demography. Peter went on to perform a similar ground-breaking study of the Iberian Peninsula.

(iii) With colleagues Peter has produced fundamental insights into genomic organisation with implications for our understanding of disease, population variation, and speciation. They developed the first methods to infer recombination hotspots (the major means of shuffling genetic information between generations) from genomic data and went on to identify ~30,000 hotspots across the human genome (~15 previously known from painstaking laboratory studies). They then produced genetic maps with massively finer resolution than before, making a fundamental contribution to human disease research. In a ground-breaking advance they identified in humans a DNA sequence motif associated with hotspots (the first for any species) and then co-discovered PRDM9, the protein that defines the locations of hotspots in humans and mice (a major discovery in mammalian biology that opened up an entirely new field of research). Peter was the first to characterise the role of PRDM9 as a mechanism underlying speciation (only known speciation gene in mammals and the first to be identified in any vertebrate).

Peter's leadership has significantly shaped the field of genomics. He led or co-led major international efforts including the HapMap Analysis Group; he was on the steering committee for the 1,000 Genomes Project; pioneered using genome-wide genetic variation to study human diseases, through GWAS (chaired the landmark WTCCC, its successor WTCCC2, and Oxford's WGS500 project). For UK Biobank, a project with massive impact, Peter led the programme to design a genotyping array, and to collect, QC, and release genome-wide SNP data on all 500,000 participants. He was a founding board member of the Australian Research Council Centre of Excellence for Mathematical and Statistical Frontiers (ACEMS), of which the University of Melbourne is the lead stakeholder.

The impact of Peter's work is immense. His papers have over 8,000 citations per year for each of the last 6 years. Since 2004 he has 27 senior/joint-senior author papers in *Science*, *Nature* (3 covers), or *Nature Genetics.* Peter has also had major impact more broadly on the public understanding of science.

Peter has played a central role as a supervisor and mentor to a new generation of leaders in the field, including a core of outstanding statistical geneticists at the University of Melbourne. He worked closely with Prof. David Balding FAA, Director of Melbourne Integrative Genomics (MIG), early in Prof. Balding's career. Prof. Stephen Leslie, Dr Damjan Vukcevic, Dr Davis McCarthy (MIG) and Dr Rory Bowden (Head of the Centre for Genomics at WEHI) were all students and/or postdocs in Peter's research group at Oxford. Peter continues to mentor and collaborate with this group of University of Melbourne researchers and this is bearing fruit as Melbourne becomes an international centre for excellence in statistical genomics.

Peter has won many major awards and was knighted by the Queen in 2019 for his services to human genetics. In short, Peter is a visionary scientist, a deep thinker, a great leader and a committed and inspirational mentor.