FOREWORD

INTRODUCTION

In 2014, four of Emeritus Prof. Peter (fondly known as PB) Beighton's past PhD students decided that they would like to honour him for his leadership and the influence that he had on their professional lives, and collaborated on a project to compile a Festschrift in his honour. They are Prof. Michael Hayden, now living in Canada, the first PhD graduate that PB supervised in 1979, together with Profs Jacquie Greenberg from the University of Cape Town (UCT), Alan Bryer from UCT and Groote Schuur Hospital (GSH), and Lawrence Stephen from the University of the Western Cape (UWC). Prof. Lawrence Stephen was the last PhD graduate that Prof. Beighton supervised before he officially retired in 1999.

Many colleagues who have worked over the past 5 decades and who continue to work with him were invited to contribute to this supplement to the SAMJ. The Festschrift includes the history behind Peter Beighton, who was born, grew up and trained in the UK and came to UCT in 1972. His legacy includes the impact that he made on those who trained under him in SA, as well as throughout the world. The Festschrift includes the history of genetics and current genetic practice in South Africa, as well as the influence that he has had on medical genetics in general, dental genetics and now genomics going into the new millennium. Tributes have been received from people all over the world, attesting to his outstanding leadership and mentorship, and highlight how he influenced the climate, growth and development of genetics at UCT, and scientific and technological fields in genetics and genomics over the past few decades.

This Foreword includes a message from Emeritus Prof. Stuart Saunders - former Head of Department (HoD) of Medicine at GSH in the 1970s when PB arrived at UCT - followed by some words from Emeritus Prof. J P van Niekerk, former Dean of the UCT Faculty of Health Sciences in the 1980s. Profs Greenberg, Bryer and Stephen conclude the Foreword, while Prof. Hayden introduces the legacy that Prof. Beighton has passed on.

Jacquie Greenberg, Alan Bryer, Lawrence Stephen, Michael Hayden

STUART SAUNDERS

Emeritus Professor and former Head of Department of Medicine (1970s), Groote Schuur Hospital, Cape Town, South Africa

I was examining at the University of the Witwatersrand (Wits) and staying with my good friend Tom Bothwell, my opposite number there. He told me one afternoon that he was going to a lecture on the genetics of bone disease. I said I would like to go with him and off we went. Peter Beighton, who was visiting the university, was the lecturer.

I thought he gave a brilliant lecture, and said to Tom that there was a vacant senior lectureship in human genetics in the Department of Medicine at UCT, and that I was going to ask Peter Beighton to apply for it. Tom's reply was they had a chair of genetics and he wanted Peter to apply for that. I said I would not act in those circumstances, but when I got home I persuaded UCT to upgrade the senior lectureship to a chair and told Tom that I was approaching Peter. Peter applied for the chair and I was delighted that he was appointed because I knew how outstanding he was. I

told Peter that the chair was in the Department of Medicine but that as soon as he wanted an independent department, I would ensure it manifested and that I would give him space, staffing and resources.

When the time came, the independent Department of Human Genetics was established with Peter as head.

The rest is history.

Peter developed a first-rate department, both at the clinical and research levels, and was an inspiring teacher. He was greatly helped by his charming wife, Greta, and by Molly Nelson - two very competent women. He became a good friend and was a loyal member of the university.

Peter established Human Genetics at UCT and made significant contributions to our understanding of the discipline. We are all deeply in his debt for what he did for medicine and for all the geneticists he inspired to follow his lead.

J P VAN NIEKERK

Emeritus Professor and former Dean of Faculty of Health Sciences (1980s), University of Cape Town, South Africa

Few people in academic careers have had the privilege of significantly influencing their field of endeavour. Peter Beighton is such an individual, and has had a major impact on genetics in SA and beyond.

Peter qualified in medicine at St. Mary's Hospital Medical School, University of London, in 1957. There followed a series of experiences including internships in London; national service as Captain in the Royal Army Medical Corps, Parachute Regiment, UK; and Medical Officer in the Parachute Regiment with the United Nations forces during the Democratic Republic of Congo crisis; postgraduate studies at the London School of Hygiene and Tropical Medicine, UK; epidemiological studies with the World Health Organization, International Medical Expedition to Easter Island, South Pacific; residency in Internal Medicine, St. Thomas' Hospital, London, UK; Research Fellow in Medical Genetics, Johns Hopkins Hospital, Baltimore, USA; and from 1970 to 1972, Senior Research Associate, Department of Orthopaedic Surgery, Wits, SA.

While in Johannesburg examining students, Dr Stuart Saunders, former Professor of Medicine and Vice-Chancellor of UCT, heard the young Beighton give a lecture and was so impressed that he set about creating a chair in human genetics at UCT, to which Peter was appointed in 1972. By then, Peter already had an impressive CV, with qualifications including two doctorates (MD, University of London 1968; PhD, Wits 1974). His publications were also varied and impressive, including 'The British expedition to the Air Mountains' in The Geographical Journal and 'Fluid balance in the Sahara', which was published in Nature.

Under his dynamic leadership, the department rapidly grew in size, scope and influence. The initial genetic clinics at many institutions and large-scale genetic epidemiological studies provided the basis for the development of cytogenetics and molecular laboratory facilities, including genetic counselling.

Peter's considerable contribution to genetics science can be gauged by the large number and the quality of his publications: 20 books, 34 book chapters and over 400 scientific papers. Some of the books are

major standard reference books, e.g. *Hypermobility of Joints* (4th edition; P Beighton, R Grahame, H Bird); *Gamut Index of Skeletal Dysplasias* (3rd edition; K Kozlowski, P Beighton) and *McKusick's Heritable Disorders of Connective Tissue* (5th edition; P Beighton, ed). His interest in history is reflected in two well-received books, including *The Person Behind the Syndrome* which he co-authored with his wife, Greta.

However, although the volume of his work is impressive, it is his collaboration with colleagues in different disciplines that led to the understanding and support of countless people with genetic problems. Roman Catholic nuns at a school for the deaf in Cape Town were completely charmed by his assistance that went beyond mere diagnosis. A physiotherapist at a school for disabled children in Kimberley said that his visits were a breath of fresh air and an inspiration. Peter often lectured at such institutions.

It has been said that the closest we can get to immortality is to have influenced for the good those who come after us. Prominent among his achievements was Peter's training of 14 clinical and scientific postgraduate students who obtained doctorates; of these, eight achieved full professorial status. Typical of such students was a colleague who rated himself as a very average student but was driven to success by Peter's belief in him and constant encouragement. Peter's insatiable sense of curiosity and adventure led him to many out-of-the-way places. His early days of playing rugby at the University of London later gave way to serious personal physical challenges, e.g. running marathons and orienteering. Despite setbacks, he still pushes himself to his mental and physical limits.

His many academic honours and awards include: the 1975 British Orthopaedic Association Robert Jones Gold Medal; the 1994 Fellowship of the University of Cape Town; and, in 2002, being the first recipient of the Order of Mapungubwe in Bronze, for outstanding achievements in medical genetics.

Uninterested in medical politics and management problems, Peter instead focused on building his department to becoming internationally recognised. He found a rich lode of genetic material in SA; he successfully mined and beneficiated it and has left an enviable legacy for his successors to build on.

JACQUIE GREENBERG

Emeritus Professor, Department of Human Genetics, University of Cape Town, South Africa

As a former employee for many years, postgraduate student and lifelong mentee of Peter Beighton (from 1972), I can honestly say that of all the colleagues I have ever worked with, PB was definitely the most influential and supportive academic I have ever known. I trust that our association will continue for many years to come. I can sincerely say that he greatly influenced the advancement of my career, by mentoring me as a professional and as a postgraduate student, guiding my research projects and encouraging collaborative publications over the past 40-odd years. It has been a pleasure and humble privilege to know and work with PB.

He was my HoD (1972 - 1999), my PhD supervisor (1986 - 1990) and is someone who has always been there for me as an academic leader and as a mentor, for >4 decades (1972 - 2016). He has demonstrated outstanding leadership in increasing the participation of young researchers in their chosen scientific and medical fields/careers; I know many of the significant numbers of diverse postgraduate students that he has trained and mentored to the completion of Masters, Doctoral and other professional medical degrees. I have also witnessed how his own research interests and passion for genetics have influenced and affected those who have worked with him. I can attest to his outstanding leadership as our HoD from 1972 to 1999, and how this affected the climate, growth and development of our faculty here at UCT. I have witnessed how PB was personally responsible for the significant increase in participation of young researchers in SA in their chosen medical, scientific and technological fields in medical genetics over this period.

Of the 14 PhD graduates that he supervised from the mid-1970s to 2000, five were women, at a time when women were generally not encouraged to do higher degrees. In fact, as I recall, when I joined UCT as a staff member in 1971, there were no female professors in the staff complement of about 10 academics in Anatomy and Physiology. I do believe that it is due to colleagues such as PB that today at UCT Faculty of Health Sciences, we number 22.

In addition, 11 of the 14 PhD graduates are clinical colleagues who PB mentored as postgraduate students and encouraged to come back to academia after they had qualified; some of whom had specialised previously and then graduated – with his support and under his supervision – with a PhD degree.

Of the three basic scientists who PB mentored and supervised to do a PhD, my story is probably the most remarkable. There are 20 years between my undergraduate and postgraduate degrees, during which time I left academia for 7 years to start a family. Had it not been for PB's support and encouragement to me as a member of his department, I would never have accomplished this. I have, in turn, supervised students and published many scientific papers in peer-reviewed journals, which has added to the pool of knowledge of genetics and medicine.

None of this would have happened had it not been for PB, who demonstrated that he had faith in me, and strongly supported and encouraged me to grow my career in genetics.

ALAN BRYER

Head of Division of Neurology, Department of Medicine, Groote Schuur Hospital and University of Cape Town, South Africa

Following an early publication in *Brain* on a family with adult-onset familial ataxia, co-authored by Derek Philcox (previous HoD of Neurology at GSH) and Prof. Beighton, I was persuaded by Dr Philcox to undertake a study of this group of disorders for a PhD project. At the time, a cluster of families with familial ataxia were known to be living in the Western Cape, with some affected members attending our neurology clinic. I realised that I would require some support in the field of genetics in order to undertake such a project. As a Wits undergraduate, I had not met Prof. Beighton before, nor had I any exposure to his lectures or teaching. As he was the co-author of the original paper, I decided to meet

with him to discuss the prospects of undertaking such a study. However, I took the decision to ask him to be my supervisor when I stood outside the door to his office prior to the appointment, before I had even met him. This was not a quick, irrational decision, but was based on what I saw on a board of honour mounted on the wall in front of his office. It contained a list of all the successful doctoral candidates who had been supervised by Prof. Beighton and the year in which they had qualified. All in all, there were 10 successful candidates on the board, and I thought at the time that anyone with that number of successfully supervised candidates clearly knew what he was doing and how to do it. My appraisal proved accurate

when I was fortunate enough to secure Prof. Beighton as supervisor of my project. To this day, I consider him to be the role model for the ideal PhD supervisor, and have tried to emulate some of his methods when I have supervised others. He had that special ability to keep a candidate motivated, on track and focused on the essential aspects of the study, allowing it to develop in an orderly, sequential manner with logical flow. He would always request a draft of the work done, in chapter format. This was returned in record speed, and supervisory meetings seldom took longer than 15 minutes. My draft was punctuated by his frequent comments, always written in pencil and never in red ink (perhaps not to alarm me). His comments were invariably on target and seldom required

extensive discussion, allowing the study to proceed smoothly. When my thesis was passed by all three eminent examiners without the need for a single correction, I attributed this to Prof. Beighton's steadfast stewardship as my supervisor. I was then able to take my place of honour in position number 11 on his board, to be followed by others who were also to have the privilege of his expertise and skill. Prof. Beighton has always been very encouraging, quick-witted with a delightful sense of humour without ever being disparaging of my efforts, and has been the source of inspiration allowing me to maintain my interest in neurogenetics for over 20 years. The tributes in this Festschrift are all testimony to the many achievements of an amazing man and his stellar career.

LAWRENCE X G STEPHEN

Head of Diagnostic Science Cluster, Faculty of Dentistry, University of the Western Cape, Cape Town, South Africa

By the time I met Peter Beighton, his reputation as an outstanding academic, mentor and scientist preceded him. The Faculty of Dentistry at UWC had been established a few years earlier and I consulted Peter regarding a family with dentinogenesis imperfecta that presented at the Department of Oral Medicine. This was the commencement of a long, interesting and fruitful association between the UWC faculty and Peter Beighton.

Peter Beighton has been a tremendous academic inspiration, and over the years has mentored several postgraduate MSc and PhD students from the Faculty of Dentistry. He was instrumental in bridging the divide that seemingly existed between the disciplines of dentistry and medicine, and this has resulted in several scientific publications. An important outcome of this association was the establishment of the combined dental (UWC)-genetic (UCT) clinic at Red Cross War Memorial Children's Hospital. Several of these postgraduates went on to become specialist orthodontists, maxillofacial surgeons, oral pathologists and HoDs at the Faculty of Dentistry.

Peter has always been patient, generous with his time and supportive in many aspects of my life. This was particularly true during the period of the merger between the Faculties of Dentistry of the Stellenbosch University and UWC. His two favourite phrases 'all problems have solutions' and 'management by objectives' have become the credos of my life.

As Peter Beighton's last PhD graduate before he retired in 2000, our association has evolved into a great friendship. I recall a particular experience when, at a congress of human genetics at the Kruger National Park, Peter had arrived the day before I did and had sighted the 'big five' game. On my arrival, he promptly informed me of the exact location of these amazing animals and unwittingly removed the adventure of tracking and spotting them. I have long since forgiven him as I realised that this was just his observant and generous nature.

To Peter, my friend and mentor, I salute you.