Sean L Sellars
Emeritus Professor of Otolaryngology, University of Cape Town, South Africa

Severe childhood deafness is a profound personal tragedy and a major social burden. Among the many avenues of clinical and scientific exploration that Prof. Peter Beighton and his Department of Human Genetics pursued over the years, subsequent to his appointment to the Chair in 1972, was that of profound congenital and childhood deafness. My personal association with the incumbent of the new Chair of Human Genetics prompted the idea of conducting a comprehensive genetic survey at the two Schools of the Deaf in Cape Town. With the forthright drive and determination of Prof. Beighton this came about and these schools welcomed this extra intrusion into their uniquely special world. Additional arrangements for itinerant visits to the two dedicated Schools for the Deaf in Worcester were later established, and over the years that followed, a mutually beneficial and remarkable relationship, indeed friendship, developed between these special schools in the Cape and the investigatory team members involved in these survey visits.

The children were assessed class by class, with their own teacher in attendance. A clinical history and any relevant information concerning past illnesses and family disorders were documented with the help of an attendant teacher and nurse. Otorhinolaryngological and general medical examinations and, where possible, audiometry, were carried out and specific morphological anomalies as well as pathological abnormalities were recorded. Syndromic and other classifiable complexes, as well as family occurrences, were invariably recognised on initial presentation. A number of immediate benefits for these children were realised. Appropriate and prompt referral for hospital assessment and attention could be and was readily made for children with cardiac and other medical abnormalities, for middle ear disease and eye disorders, and for 'at risk' families that could be considered in need of genetic counselling.

Over the next 10 years, recurrent surveys were made and additional visits to other schools for the deaf were included. Within a decade, 16 schools for the profoundly deaf and 3 schools for the hard-of-hearing had been visited in SA. Composite analysis of the 3 064 children examined over the years at these 19 schools was published, and a subsequent publication reported the findings from 5 460 hearing-impaired children seen in 28 special schools. In later years, scientific studies into the genetics of deafness have been undertaken by clinicians and scientists, whom Prof. Beighton has personally enthused, inspired and informed. Under his guidance, their work on this important disorder has been variously published, much to the enhancement of the international reputation of the Division of Human Genetics at UCT.

Special tribute is made to one extraordinary man, responsible in his speciality of human genetics for uplifting both SA clinical healthcare and the academic wellbeing of the students and staff who have come under his guidance. Many years ago, that great South African, General Smuts, in his delivery in Oxford of the Rhodes Memorial Lecture on 2 November 1929, stated, 'It is ever the hallmark of genius to initiate points of view which are not a flash in the pan but burn with a steady brilliance, and to launch ideas whose fruitfulness increases with time and which carry their own immortality.'

For those who have had the privilege to work with him, Emeritus Prof. Beighton has done just this.

Herman Hamersma
Otology and Neurotology, Private Practice, Flora Clinic, Roodepoort, Gauteng, South Africa (formerly Professor of Ear, Nose and Throat, University of Pretoria, South Africa)

At the 1973 South African Medical Association Congress in Cape Town, I presented a paper on my experience: 'Total facial nerve decompression in patients with osteopetrosis.' I suggested that a future genetic study was indicated, but stated that I had no experience with genetics.

Immediately after my talk, I was approached by Prof. Sean Sellars, Head of Ear, Nose and Throat at UCT. He introduced me to Prof. Peter Beighton, the newly appointed Head of the Department of Human Genetics, who had attended my talk. Sean told me that Peter had a special interest in sclerosing bone dysplasias. Peter expressed a desire to be involved in the study that I had mentioned, and I gladly accepted his collaboration. At that stage there was no internet, and all the literature research that I had undertaken involved paging through the *Index Medicus*.

Peter Beighton subsequently visited me in Pretoria and saw 15 patients in my consulting rooms. He could immediately differentiate them into two groups, i.e. dominant and recessive forms of osteopetrosis. Peter then contacted an expert on bone dysplasias, Prof. Jurgen Spranger of Mainz, who referred him to a 1967 publication of Dr Hansen in a paediatric textbook, where he had proposed the name 'sclerosteosis' for osteopetrosis with syndactyly. When I visited Cape Town in October 1973, we saw more cases together at Groote Schuur Hospital (GSH).

In January 1974, Peter was informed about a sclerosteosis patient in Albertina who had been operated on for increased intracranial pressure by a neurosurgeon in Pretoria. Apparently, she had taken a turn for the worse and we believed that we should go and see her. However, the Suez crisis made it impossible to get enough petrol to drive so far over the weekend and Peter arranged for a small airplane from the UCT Flying Club to take us to Albertina, together with nursing sister Lecia Durr (now Bartmann, her tribute on page S125). Fortunately, the patient was still alive, but nearly totally blind, and she died not long afterwards. The opportunity to examine her at that penultimate stage of her illness provided valuable insights into the pathogenesis of the condition.

When we departed from the local airport on our return flight, the monoplane lifted up from the runway but a sudden crosswind caused the pilot to oversteer and the right wheel touched a gravel heap! Fortunately, the left wheel was still intact and seemed to retract, but we saw brake fluid oozing from the upper surface of the right wing. We could not radio for help because the radio could not transmit at low altitude and so we continued towards Cape Town. Peter Beighton sat in front next to the pilot and kept chatting to cheer him up. The pilot was as white as a sheet! Lecia and I were in the back seat.

After half an hour, the radio in the plane came alive and a pilot from a South African Airways Boeing at cruising altitude over the Orange Free State told our pallid pilot that he had disturbing news for us, i.e. we had left the right wheel in Albertina! We spent the next half hour thinking about the impending belly landing at Cape Town Airport.

After we came over the mountains at Sir Lowry's Pass, the control tower instructed our pilot to do a fly past so that they could see whether the left wheel was retracted, prior to attempting the belly landing. Fortunately, it was retracted and they directed us to a distant area of grass next to a runway where two fire engines were waiting for us. There, we belly landed successfully on the grass in a sea of foam. That was the fastest stop we had ever done in our lives.

Medical research can be dangerous! See the photograph!

Kazimierz Kozlowski

Emeritus and Senior Staff Radiologist, The Children's Hospital at Westmead, Sydney, Australia

Accidental encounters are sometimes followed by lasting links. I met Prof. Peter Beighton at a congress of the International Skeletal Society in Germany in 1975, at which he gave a talk on osteogenesis imperfecta. He was an excellent speaker and I was impressed by the quality of his paper as well as his presentation. We met again the following year in India, where we were invited overseas speakers at a congress of paediatric genetics. This meeting confirmed that PB and I shared a common interest in genetic bone disorders.

I was invited to visit Cape Town in 1978 where I spent two very productive weeks, reviewing radiographs of skeletal dysplasia patients, with PB. It was a cornucopia of most interesting material. Owing to the efficiently organised Department of Human Genetics, four collaborative articles were written during that time and we successfully delineated three previously unrecognised disorders: osteoglophonic dysplasia,[1] spondyloepimetaphyseal dysplasia with joint laxity (SEMDJL)[2] and distal osteosclerosis.[3]

In 1989, I had the pleasure to receive PB for a short academic visit to Sydney where he was accommodated in the Children's Hospital guest room. I remember his astonishment when he opened the door to find a woman lying in the bed! The situation was quickly resolved; it was not an Australian custom of VIP reception, but a wrong key.

We were co-authors of a book, *Gamut Index of Skeletal Dysplasias*, published by Springer-Verlag in 1984. Further editions were published in 1995 and 2001.[4] In the years that followed, our paths crossed at congresses around the world. By 2015, we had collaborated on more than 20 papers in the medical literature. Personally, I am heavily indebted to PB for editing the grammar and syntax of my own medical articles written in the English language. I have happy memories of our long-lasting friendship and professional interaction.

1. Beighton P, Cremin BJ, Kozlowski K. Osteoglophonic dwarfism. Pediatr Radiol 1980;10(1):46-50.
2. Beighton P, Kozlowski K. Spondylo-epi-metaphyseal dysplasia with joint laxity and severe progressive kyphoscoliosis. Skel Radiol 1980;5(4):205-212.
3. Beighton P, Macrae M, Kozlowski K. Distal osteosclerosis. Clin Genet 1980;18(4):98-304.
4. Kozlowski K, Beighton P. Gamut Index of Skeletal Dysplasias. 3rd ed. London: Springer-Verlag, 2001.

Ingrid Winship

Executive Director for Research, Melbourne Health; and Professor of Adult Clinical Genetics, University of Melbourne, Australia

A defining moment in my career was during Peter Beighton's lecture at the University of Natal in 1974. As a high school student planning a career in medicine, I attended this lecture with my father, Bill, and by the end of the hour I had decided to become a clinical geneticist. I began reading genetics in earnest. Later, as a medical student, I assisted him with field work for a research project and when I qualified and it was time to choose a medical discipline, it was quite straightforward. Peter welcomed me into the department and gave me all the opportunities required to create a career. He led by example; his work ethic and his drive were infectious. I recall child abuse allegations being laid on the parents of a child with epidermolysis bullosa. Incensed by the injustice, I described this to Peter, suggesting this as a situation worthy of publishing.[1] His response was supportive – that I should have the first draft on his desk by 16h00 that day – which I did! Peter's passion and enthusiasm inspired others to do the same. Working with Peter was fun, informative and constructive. He placed trust in his team and expected us to deliver, while empowering and providing the resources for success.

When I was invited to move to New Zealand to establish a new clinical genetic service, Peter encouraged me to seize the opportunity. His earlier insistence on maximising learning and making contacts when travelling had allowed me to visit major clinical services around the world. In this way, I had observed how genetic services were conducted at Great Ormond Street, and in Manchester, Cardiff, Glasgow and Vancouver, which enabled me in the challenge, at 36 years of age, of setting up a successful genetics department de novo, in Auckland, New Zealand.

Peter, thank you for what you have taught me in knowledge and resourcefulness, and also in self-belief, tenacity, alacrity and compassion.

1. Winship IM, Winship WS. Epidermolysis bullosa misdiagnosed as child abuse. S Afr Med J 1988;73(6):369-370.

Claudette Medefindt
Head of Science, Retina South Africa, Johannesburg, South Africa and Deputy President, Retina International

In 1979, Prof. Beighton met with a small group of patients affected by the genetic eye condition retinitis pigmentosa. He encouraged them to form a support group and the South African Retinitis Pigmentosa Society was born.

After many years of growth and change, the organisation evolved into Retina South Africa – a powerful voice in advocacy and research support to find treatments for all forms of retinal genetic conditions.

Prof. Beighton was also instrumental in the establishment of the research programme at UCT to look into the genetics of retinal degeneration, and the first grant of R15 000 from the renamed Retinitis Pigmentosa Foundation in 1992 saw the beginning of a dynamic partnership between UCT and Retina South Africa that endures to this day.

In 1994, the team identified the first unique SA gene causing a dominant form of retinitis pigmentosa and many successes followed. The project has earned recognition for its work in the genetics of retinal degeneration in SA. The team continues to be at the forefront of what is now a multidisciplinary international research initiative using gene therapy, stem cells, Optobionics, artificial vision and pharmaceutical interventions to find treatments for retinal blindness.

Prof. Beighton was always a ready source of advice and encouragement to Retina South Africa and served on their Scientific and Medical Advisory Board for many years.

Without his support, encouragement and vision, we doubt if the retinal patient movement in SA would have been established or grown to the point where it is today – a strong and respected component of Retina International, the international support group movement.
Thank you Prof. Beighton for your vision and commitment to fighting retinal blindness.

Sol Zieff
Specialist Paediatrician, Christiaan Barnard Memorial Hospital, Cape Town, South Africa (formerly Senior Paediatrician, Red Cross War Memorial Children's Hospital, Cape Town, South Africa)

Dear Peter,
I thank you most sincerely for your letter regarding your re-retirement after an incredibly long innings. The idea of a Festschrift would be an apt tribute highlighting your illustrious career over four decades. Before your arrival, human genetics in Cape Town was truly a Cinderella branch of medicine, the cytogenetics laboratory being housed in the nether regions of the Department of Physiology under Pauline Webb's direction.

It was only after you assumed your position as HoD that clinical genetics made its mark. At the Red Cross Children's Memorial Hospital, the weekly clinic soon expanded to two. They were essentially dysmorphology clinics. Patient attendance increased dramatically to the extent that after 2 years, I was able to present an analysis of the first 200 patients at the 5th Annual Congress of the South African Genetic Society, in Stellenbosch, in February 1974.

Chromosomal anomalies accounted for most of the pathology seen and I do recall our taking wagers on the probable diagnosis where it was not clinically apparent – do I still have a few beers owing to me?

Your contribution to orthopaedic genetics remains legendary and recognised universally, while your ability to impart seemingly abstruse clinical material in a comprehensible form endeared you to medical students and colleagues in the Faculty of Medicine, as well as to the participants in the UCT Summer School programme who returned year after year to access the font of advances in genetics.

The biographical monographs on *The Man Behind the Syndrome* and *The Person Behind the Syndrome* were clearly a labour of love (yours and Greta's).

The richly deserved accolades that you have accumulated over the years will remain unchallenged for many more years.

May I wish you and Greta continued good health to enjoy the fruits of your labours, especially the grandchildren, now that you have the time to indulge them.

Very sincerely, Sol Zieff.

Peter Beighton and Greta, 1970s.

Peter Bonafede
Medical Director, Providence Arthritis Center, Portland, Oregon, USA

I first met Prof. Beighton (PB) as a medical student, shortly after he became the chairman of the UCT Department of Human Genetics, in 1972. His presentation to our medical school class made the subject exciting. He didn't bore us with Mendelian inheritance, but made the topic come alive, for instance, with his speculation as to the origins of the Tuareg nomads and the mystery behind the statues on Easter Island. It was an easy decision then as to where I wished to spend the 6 months before doing my National Service in 1976. I learnt an immense amount from PB during this period and also in the second 6 months after National Service, in 1977. I learned basic genetics as well as skills in medical writing, public speaking, running a department and some of life's important lessons.

Prof. Beighton was and is the most important mentor in my career and I still quote and live by many of his maxims.

PB helped me select a topic for an MD and without his encouragement and sage advice I may never have graduated. However, I eventually did as his first graduating doctoral student in 1978, together with Mike Hayden: mine an MD and Mike's a PhD (see page S7). I have not made a career out of genetics, but my time with PB and my graduating with a doctoral degree made a large difference in career opportunities, such as obtaining work in London, England, and training further, firstly at GSH in Internal Medicine and then in Rheumatology in the USA at Oregon Health and Sciences Center, where Prof was known by reputation. I have met rheumatologists and orthopaedists from multiple countries who are familiar with PB and his work.

Many of what I call the 'Beighton Rules' still influence my decisions. In no particular order, firstly, there is the 'law of diminishing returns', which we used to decide whether any activity was worth the time and effort that would be involved. This law has helped me make decisions as to where I should place my energy, particularly with limited time and resources both in work and life. It helped us decide what topics I should pursue and where I should concentrate my efforts, both then and now. The next useful law is the importance of team work and getting on with others when deciding on employability. It is vital that personnel for all positions, from those doing basic work through to professionals and heads of department (HoDs), work as a team because, if not, even the brightest and highest qualified will likely be unsuccessful and create discord. The last rule that I will quote is Prof emphasising 'be careful with whom you collaborate'. What an example he was! If we had something of merit that we wished to publish, I would do the first draft. By the next day, he would have corrections and comments back to me and within a week we would submit the article to the journal. This productivity was exceptional and emulated by few. I will admit that I haven't always chosen partners wisely and there are potential publications that never got further than the first draft. I am sure that I have assimilated many other 'rules' that I have not named.

Prof and his department were very productive from the beginning of his tenure but he also knew how to make projects great adventures. For instance, when we went to evaluate deaf persons for possible genetic diseases at the School for the Deaf in Worcester, we would stop in at Bainskloof Hotel on our way home. To earn the anticipated fabulous meal and wonderful wine in the evening, we would run down Bainskloof pass for 5 miles where Greta, Prof's wife, would pick us up and take us back to the hotel. We were fortunately allowed to shower before dinner! And then there were the days when Prof would tell me that the tide was low at Milnerton beach and we should go for a run. During these 5- to 10-mile runs we would not only talk about work, but rugby, beer and a wide range of other topics of interest. This exercise started my recreational running and I eventually ran more than 30 marathons and ultramarathons. Thanks Prof.

I am now a rheumatologist in Oregon, USA, and PB's influence still touches me. His first foray into skeletal dysplasias was his interest in Ehlers-Danlos syndrome, a condition characterised by joint hypermobility, as well as other manifestations depending on the category. Patients with hypermobility tend to have more aches and pains, including fibromyalgia, an enigmatic pain syndrome that usually presents to rheumatologists. Additionally, because of their joint laxity and the risk of recurrent joint dislocations it is more common for them to develop premature osteoarthritis. The Beighton Score is universally used to assess the presence of joint hypermobility, a scoring system that is practical and easy to measure at the bedside. Prof first used this score in his early population studies in SA and has told me that he never intended that it be used as a clinical tool in patient care. It is with pride that when I see a patient with joint hypermobility I quote the Beighton Score and have it documented in the medical record. I have also told a number of colleagues of my connection to Prof, and have shown a few privileged ones my autographed book, *Hypermobility of Joints*, a gift to me from Prof.

It is now 38 years since I worked in the Department of Human Genetics, UCT, and it was a most productive and enjoyable time in my practice of medicine. I have many wonderful memories and could recount numerous additional anecdotes. I can truly say that PB was a major leader in the development of the field of human genetics in SA and his influence is felt worldwide. I was very fortunate to be able to learn and work under him and am proud to have played a very small role in his department in the early days.
Thank you, Prof. Beighton.

Thank you Janet Bonafede and Caroline McCulley, MD, for critical review of the manuscript.

George Gericke
Clinical Head: Genetics Unit, AMPATH National Pathology Group, Centurion, South Africa

Who is Peter Beighton? A man's man, characterised by honesty, integrity, humility, diligence and a love of exuberant laughter; highly adventurous and of great courage, racing the death-defying Isle of Man motorcycle event; walking across the Sahara with a Tuareg, recording urine osmolality along the trip; and being a military medical officer with the UN in the (then) Belgian Congo.

He is also fun-loving, laughing at himself and telling the next story with great amusement. During a visit to Australia, perhaps jet-lagged and somewhat tired after a city-per-day whirlwind lecture tour, he began one of the evening lectures by enthusiastically proclaiming, 'I am delighted to be in Perth,' when somebody from the back of the hall shouted, 'Brisbane you fool!'

He had an exceptional lecturing talent, interlaced with humour. Once, at Tygerberg Hospital, where, in his last slide, an orthopaedic surgeon stood next to a man with a skeletal dysplasia, he asked, 'Is the Professor of Orthopaedics a giant, or is the patient a dwarf?'

He is also known for being a straight talker: he would come running into my office in his socks carrying a big coffee mug, and convey a very clear message: 'I like your ideas, I am glad you came' before disappearing without waiting for a reply, or, 'You messed up this paper, you will have to rewrite it' (actually in stronger language than rendered here). You always knew exactly where you stood, which was pleasantly different from the Afrikaner reticence I experienced in the academic world before.

When a particular staff member with a generally known fondness for dogs, with whom he allegedly frequently clashed, was absent one morning at a journal club meeting, he wanted to know, 'Where is … ?' When told that the person was sick, he said, 'What … distemper?'

During a meeting chaired by the very formal Prof. Willie van Niekerk, University of Stellenbosch obstetrics professor and feared by most living humans, PB's new digital watch started playing a loud tune. One can imagine firstly Van Niekerk's knotted eyebrows when his important meeting was disrupted, and secondly those same eyebrows raising in amazement when PB could not stop the sounds, promptly took it off his arm and offered it to the professor, saying, 'Here, you fix it!'

He remained an intrepid supporter. I gave a plenary lecture on epigenetic mechanisms involved in transgenerational stress transmission during war situations at a multidisciplinary meeting on 'The Cost of War' in Liverpool a few years ago. PB graciously attended as he happened to be in the UK at the same time. At the end of the presentation there was what could possibly have been interpreted as a muted response from the eclectic academic audience. PB was the ever-enthusiastic supporter, who suddenly jumped up with blazing eyes and with a sweep of his arms proclaimed loudly, 'What a magisterial presentation!'

In later years, we tried to meet at least once a year over a cup of tea to talk and reminisce. Some years ago, just before one of our meetings his house caught fire and only one room was left untouched, without water and electricity. As soon as I started making sympathetic comments he quickly stopped me by forcefully indicating, 'No sympathies George, this is a real character-building exercise'.

When I was at the University of Pretoria, sometimes agonising over certain academic or collegial matters which worried me at the time, I phoned him for advice and my problems vanished when he simply said, 'Don't take anything too seriously George, life's a game, life's just a game!'

Peter Beighton, a truly extraordinary man, larger than life, now rewriting one of his books, still supervising doctoral students and busy with his nth doctorate himself, deserves to be nominated the most outstanding player of the century in the Genetics World Cup. He was lucky to have been supported (and I suppose tolerated, like all of us men are, it seems) in his case by an extraordinary woman such as Greta. And the best stories have not even been told!

Denis L Viljoen
Chairman for The Foundation for Alcohol-Related Research and Extraordinary Professor, Department of Obstetrics and Gynaecology, Faculty of Health Sciences, Stellenbosch University, South Africa

Peter Beighton is my mentor and friend. We met on a golf course late on a Wednesday afternoon after work for a nine-hole game arranged by a mutual friend, now a prominent paediatric surgeon. The date was mid-year 1982, and my colleague warned me that PB was the eccentric English-born Head of Human Genetics at the UCT Medical School, who loved being outdoors and played golf erratically. From the very beginning of our relationship PB held true to all these observations and many more. Life was never dull around PB. Immediately after our golf game, during which he had sprayed golf shots into trees, water, bunkers and bushes with total abandon, he offered me a post as an MRC Fellow in Human Genetics at UCT. I had plans for private paediatric practice but readily accepted this challenging position.

PB had an 'I can' attitude to research, academic writing and teaching that was refreshing and inspiring. He gathered a team of young, gifted medical practitioners and scientists, all of whom have subsequently become leaders in the fields of medical genetics and closely allied specialities, and who have prominent positions in major departments throughout the world. These include Jack Goldblatt, Mike Hayden, George Gericke, Ingrid Winship, Colin Wallis, Gillian Wallis, Rajkumar Ramesar, Lawrence Stephen, Jacqueline Greenberg and many others. Most wrote doctoral theses under his expert guidance, and these efforts produced several hundred original manuscripts and more than a score of books. My own association with PB lasted 16 years and produced 80 papers and an MD thesis.

A few aphorisms and incidents have to be told to commemorate PB's incredibly productive publishing career and a few eccentric, erratic and bewildering behaviours. All his associates will remember a lightning-quick mind, ability to think 'on his feet' in order to alleviate awkward situations and a very mischievous humour. For instance, if a projector failed at an international meeting during his presentation, PB's advice was immediately to call the attention of the chairman of the session and seat himself, apparently without a care in the world, while the head of the session tried, usually unsuccessfully, to fix the errant instrument. Similarly, if a contentious question was posed, usually by an aggrieved 'expert' who was having his or her academic limelight 'stolen' during a PB presentation, his advice was to request the chairman of the session to adjudicate in the matter to avoid any further rancour. Several such manoeuvres were very much part of PB's armamentarium.

Several amorphisms come to mind: 'the law of diminishing returns' was helpful in thesis and manuscript preparations. Another helpful amorphism was 'time spent in reconnoitring is never wasted.' This advice to check the presentation arena, its foibles and obstacles and instruments such as laser pointers, microphones, projectors and lighting switches, was invaluable.

PB has many interests, but many have been curtailed due to advancing years, e.g. marathon-running, gymnasium attendance, cross-country sorties, cycling, bird-watching, coin and medal collections, battlefield and Victorian history, visiting remote islands and more. However, through all these disparate activities, one constant companion has been his faithful, doting, beloved wife Greta Beighton. Without her, I believe PB would have never achieved his prodigious outputs of writing and activity. May both have many more happy and productive years together.