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Trefor Jenkins (1932–2025): Pioneer of human genetics in southern Africa

Professor Trefor Jenkins was one of the most influential figures in the development of the field of human genetics in southern Africa. His career, spanning over four decades, fundamentally transformed the landscape of genetic research, clinical services and education across the region. Through his pioneering work at the University of the Witwatersrand (Wits) and the South African Institute for Medical Research (SAIMR) [now integrated into the National Health Laboratory Service (NHLS)], Jenkins established the foundation for modern genetic studies in South Africa while contributing significantly to our understanding of human population diversity and genetic diseases on the continent.

Early career and academic foundation

Jenkins began his career in medicine in the United Kingdom, qualifying in 1956, before embarking on a journey that would take him to Rhodesia (now Zimbabwe) and eventually South Africa. His initial work as a mine medical officer provided him with early exposure to the diverse populations of southern Africa, an experience that would later inform his genetic research. Jenkins taught and did research in the field of human genetics and founded genetic counselling clinics and diagnostic and research laboratories throughout his career.¹

With the support and encouragement of Professor Phillip Tobias, Jenkins' transition to human genetics came in 1969 when he took up the positions of Head of the Human Sero-Genetics Unit at the SAIMR and part-time lecturer in human genetics in the Department of Anatomy (Wits). This marked the beginning of his systematic approach to establishing human genetics as a distinct discipline in southern Africa.^{2,3} Together with Tobias, he conducted field research on local communities, motivated by his curiosity about population history and why some diseases, like the sickle cell trait, were prevalent in some community groups he encountered as a doctor working in Zimbabwe.

Institutional leadership and development

Jenkins' appointment as the first Professor of Human Genetics at Wits in 1974 transformed the department to excel in research, clinical services and education. He introduced genetic services in Johannesburg, which later expanded to other regions. At first, the services involved chromosome studies, and later he introduced serogenetic markers, including blood groups and serum proteins, for diagnostic purposes and research.

The South African Medical Research Council, in conjunction with the SAIMR and Wits, awarded Jenkins an extramural research unit and appointed him as Director of the Human Ecogenetics Research Unit between 1977 and 1993. Funding from this source supported much of the research conducted in the department. In the early 1980s, in line with international trends, Jenkins turned his attention toward integrating the 'new genetics' based on the use of DNA technology to enhance diagnosis and research. His department became a regional centre of excellence, attracting researchers and clinicians from across the globe and stimulating international collaborations that elevated the profile of genetic research in African populations.

Through a generous endowment from Philip von Wielligh, Jenkins initiated a lecture series that ran for over two decades and enabled him to invite prominent international experts to visit the department to share their knowledge and expertise. This provided an exciting and enabling environment for cutting-edge research.

Following his retirement, Jenkins was called back to service to play a major role in the establishment of the Institute for Human Evolution (later renamed the Evolutionary Studies Institute) at Wits and he acted as Interim Director from 2004 to 2009. During this time, he co-edited a book with Philip Bonner and Amanda Esterhuysen titled *A Search for Origins: Science, History and South Africa's 'Cradle of Humankind'*, which was published in 2007.⁴

Research contributions and scientific impact

Jenkins' research portfolio was remarkably diverse, reflecting the complex genetic landscape of southern Africa. He published and collaborated on over 300 papers and three books, demonstrating his prolific contribution to the scientific literature. His work encompassed several key areas that were particularly relevant to the African context.

One of his most significant contributions was in population genetics, where he studied the genetic diversity and relationships among southern African populations.⁵ His research provided crucial insights into the origins, migrations and genetic affinities of various ethnic groups in the region. This work was particularly important given the unique position of southern Africa as a crossroads of human migration and the home of some of the world's oldest human populations.

Blood group genetics formed another major area of Jenkins's research. This research contributed to understanding population relationships and migration patterns in southern Africa, which provided the foundation that was later refined with patterns of variation at the molecular level using DNA markers.

His team's work led, among other contributions, to new insights into the origins and affinities of the peoples of sub-Saharan Africa. These findings were crucial for understanding human evolutionary history and the genetic basis of population diversity in Africa.

The second broad theme of his research was the identification of the molecular DNA variants that cause Mendelian traits, including sickle cell anaemia and albinism, that disproportionately affect African populations. The research of the students and staff he guided also uncovered the mutational profiles for thalassaemia, cystic fibrosis, spinal

muscular atrophy, Huntington's disease, myotonic dystrophy, Duchenne muscular dystrophy, fragile X syndrome, several rare skin diseases and other genetic conditions in South African families.⁶ The knowledge gained informed the genetic services offered by the Department and ensured their relevance in the South African context.

In a seminal review article⁷, published in the *Journal of Medical Genetics* in 1990, Jenkins provided a comprehensive overview of the state of medical genetics in South Africa. This publication became a landmark reference that documented the progress made in the field and outlined future directions for genetic research and clinical services in the country.

Educational innovation and legacy

Beyond his research contributions, Jenkins was a keen educator who revolutionised human genetics teaching in southern Africa. Jenkins also pioneered an undergraduate teaching project in medical ethics at Wits, demonstrating his commitment to ethical practice in genetics research and clinical care.

His educational interests led him to increase the contribution of genetics to the undergraduate medical student curriculum, while his enthusiasm for research led him to encourage his staff, colleagues and clinicians to pursue higher degrees in the field. This approach created a multiplier effect, as his students and mentees went on to establish genetic services and research programmes throughout the region and further afield.

Jenkins' commitment to education extended beyond formal university settings. He recognised the importance of translating genetic knowledge into practical clinical applications. He was a superb speaker who enthralled audiences with his humour, enthusiasm and storytelling ability in many diverse situations.

Jenkins' work gained international recognition, and he became a respected voice in global genetics communities. His research provided African perspectives on human genetic diversity, challenging Eurocentric assumptions and contributing to a more inclusive understanding of human genetics. He was awarded honorary degrees by the Universities of South Wales, the Witwatersrand and Cape Town for his contributions to science and education.

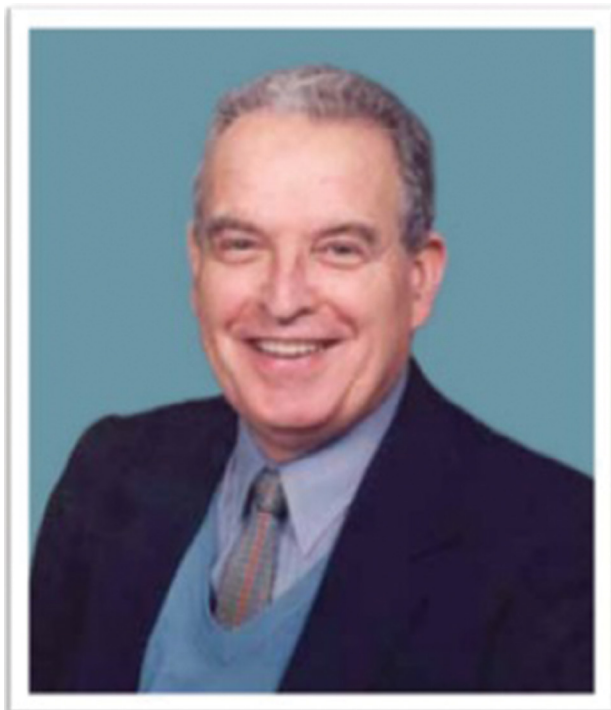


Image: ASSAf

"Prof Trefor Jenkins: A good man in Africa."¹

His community engagement expanded when he started the South African Inherited Disorders Association in 1973. This association initiated genetic support groups in the community, supported research, and raised public awareness of genetic disorders and birth defects. In addition, he was instrumental in founding the Southern African Society for Human Genetics in 1986, creating a space for geneticists to engage and be updated with the most recent developments in the field of human genetics.

Jenkins was also one of the founding members of the Academy of Science of South Africa (ASSAf) which was inaugurated in 1996. In 2003, he was awarded the first ASSAf Science-for-Society Gold Medal, the highest accolade ASSAf confers in recognising outstanding achievement in scientific thinking for the benefit of society.

Impact on contemporary genetic research

The foundation laid by Jenkins continues to influence contemporary genetic research in southern Africa. His emphasis on studying Indigenous African populations has proven prescient, as modern genomic studies have revealed the exceptional genetic diversity present in African populations. Current research building on his work has shown that African populations harbour the greatest genetic diversity of any continental group, reflecting humanity's African origins.

Modern whole-genome sequencing studies of African populations continue to build on the population genetic frameworks established by Jenkins and his colleagues. These contemporary studies validate many of his earlier insights about population relationships and migration patterns while providing unprecedented detail about the genetic history of African peoples.

Medical ethics, human rights and standing up against apartheid

The tragic death of Steve Biko in 1977⁸ stands as one of the most significant cases in medical ethics, highlighting the intersection of professional responsibility, political pressure and human rights. The subsequent legal and professional proceedings, including the work of academics like Trefor Jenkins, Phillip Tobias, Frances Ames and another group of doctors including Dumisani Mzamane, Yosuf Veriava and Tim Wilson, exposed deep-seated racism within South African medicine and raised fundamental questions about medical ethics under authoritarian regimes.

Through their collaboration on 'The Steve Biko Affair: A Case Study in Medical Ethics', Jenkins and co-author Graham McLean provided a critical analysis that continues to influence medical ethics education worldwide.^{9,10} They used the Biko case to highlight the moral dilemmas that challenged the medical profession's fundamental principles. The case became a landmark study in medical ethics, revealing how political influence can undermine professional responsibility. It also contributed to the development of international guidelines for medical professionals working in situations of political conflict or oppression and helped establish principles for maintaining medical neutrality and professional independence, even under extreme political pressure.

Jenkins was a founding force behind the establishment of the *Steve Biko Centre for Bioethics* at Wits in 2007, where a swathe of pertinent issues continues to be debated. He became deeply involved in the destigmatisation of HIV/Aids, advocating to make it a notifiable condition in medical practice in South Africa. Later, Jenkins worked closely with the then Dean of the Faculty of Health Sciences, Max Price, and Yosuf Veriava to put together a submission to the Truth and Reconciliation Commission on behalf of the faculty. There is now a plaque at the entrance of the Wits Medical School that states: "The Faculty reaffirms its rejection of racism and other violations of human rights."

The contribution of Jenkins' work as a medical geneticist, ethics educator and moral analyst demonstrates the importance of medical professionals taking broader social responsibilities seriously. His analysis of the Biko affair, combined with his scientific work challenging racist ideologies, represents a comprehensive commitment to using medical knowledge and professional influence in service of human dignity and social justice.

Conclusion

Trefor Jenkins' career represents a transformative period in the history of human genetics in southern Africa. Through his visionary leadership, he established the institutional framework, research programmes and educational initiatives that continue to shape genetic research and clinical practice in the region today. His work demonstrated the critical importance of studying human genetic diversity in African populations, both for understanding fundamental aspects of human evolution and for addressing health disparities that disproportionately affect African communities.

Jenkins' legacy extends far beyond his immediate research contributions. He created a sustainable model for genetic research and clinical services that could thrive in resource-limited settings while maintaining international standards of excellence. His emphasis on education and capacity building ensured that his influence would continue through subsequent generations of African geneticists and clinicians.

The foundation he established continues to be relevant in the genomic era, as researchers increasingly recognise the importance of including diverse populations in genetic studies. Jenkins' early recognition of the genetic wealth represented by African populations has proven prophetic, as contemporary genomic research continues to reveal the exceptional diversity and evolutionary significance of African genetic heritage.

His career stands as a testament to the impact that dedicated individuals can have on entire scientific disciplines, particularly in areas where such disciplines are still developing. Through his combination of rigorous science, institutional building and educational innovation, Trefor Jenkins created lasting change that continues to benefit both the scientific community and the populations of southern Africa.

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