Division of Human Genetics



Celebrating 50 years 1973-2023









Unravelling Genetics, Enriching Lives: Celebrating 50 Years of HumGen



UNIVERSITY OF CAPE TOWN GROOTE SCHUUR and RED CROSS HOSPITALS

MRC Genomic and Precision Medicine Research Unit

Division of Human Genetics

Department of Pathology

Faculty of Health Sciences
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10th October 2023

Dear Past and Present Members of the Division of Human Genetics.

As we gather to commemorate the 50th Anniversary of our Division, I find myself filled with both pride and humility. Our journey together, spanning five decades, has been nothing short of extraordinary in this post-human genome project era.

My own connection with the Division traces back to a pivotal moment in December 1988 when Professor Peter Beighton interviewed me. I accepted the role as head of the Human Genetics Research Laboratory, embarking on this remarkable journey officially in January 1989, fresh from completing my Ph.D. It has been a continuous privilege to immerse myself in the captivating realm of Human Genetics/Genomics. This journey has allowed me to engage in groundbreaking research, undertake inspiring international visits to leading Human/Medical Genetics facilities, and contribute to transformative discoveries. Among these, my sabbatical at the University of Chicago Medical Center in the mid-90s stands out, where I unraveled the gene/mutation responsible for colorectal cancer in our South African cohort. Returning home with this knowledge, we translated it into a diagnostic/predictive test, eliminating the need for unnecessary colonoscopies in mutation-negative individuals....and the rest is history. https://www.youtube.com/watch?v=v7PWr5Oy3jo&t=1s

Our Division has consistently shone as a beacon of dedication and excellence in the field of Human and Medical Genetics. Over these five decades, we've not only seen our numbers grow but, more importantly, witnessed our unwavering commitment to pushing the boundaries of knowledge in our domain. Human Genetics lies at the heart of modern medicine, and our Division has played an instrumental role in dissecting the intricate tapestry of genetic influences on health and disease. From pioneering research to compassionate clinical care, we've improved the quality of life for countless individuals.

Our noteworthy contributions extend to understanding genetic diversity, particularly among South African populations. This work has not only expanded our scientific horizons but also paved the way for more equitable healthcare solutions. By embracing cutting-edge technologies like Next Generation Sequencing, Bioinformatics, Big Data analytics, and Artificial Intelligence, we've emerged as leaders in utilizing these tools for the betterment of healthcare and research. We've also empowered the next generation, equipping them with the skills needed to navigate the complexities of modern genetics and genomics, resulting in record numbers of applicants for our postgraduate programs.



Additionally, our Division's legacy of successful graduates occupying leadership positions nationally and internationally fills us with immense pride. On this occasion, it's fitting to acknowledge the crucial funding support from UCT, the National Research Foundation (NRF), the Medical Research Council (SAMRC), Retina South Africa, and international agencies. These organizations have played a pivotal role in enabling our cutting-edge research and providing bursaries to our students.

We've had the privilege of organizing several local and international conferences, with the most recent being the highly successful International Congress of Human Genetics held in Cape Town in February of this year (www.ichg2023.com). It revolved around the theme of '...coming home,' and what made it particularly exhilarating was the substantial international participation, especially among younger researchers. Out staff and students truly shone at this meeting.

While we celebrate our Golden Anniversary, let us not forget the challenges we've faced along the way. Limited resources and the unwavering commitment of our staff to provide exceptional clinical services, even in the face of adversity, are truly inspiring. It's this resilience and dedication that define our Division and set us apart.

In closing, I want to extend my profound gratitude for your unwavering support and tireless efforts in advancing our mission. As we move forward, let us continue to embody the values of compassion, innovation, and dedication that have been the cornerstone of our Division's success. Leading such an exceptional team fills me with immense pride, and I eagerly anticipate the continued growth and impact of our Division in the years to come.

With warm regards and heartfelt congratulations,

Yours sincerely

Raj S Ramesar

Professor and Head: Division of Human Genetics

Director: MRC Genomic and Precision Medicine Research Unit



Department of Pathology

Faculty of Health Sciences, Observatory, 7925, South Africa Head of Department: Professor Komala Pillay

Sr Admin. Officer: Adielah van der Schyff Tel: +27 (0)21 406-6757

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29 September 2023

Dear Prof Ramesar and the Division of Human Genetics

RE: 50th anniversary celebration

I am writing to extend my heartfelt congratulations to you and the entire Division of Human Genetics at the University of Cape Town as you celebrate your 50th anniversary. This significant milestone not only acknowledges the legacy of excellence that the division has cultivated but also highlights the invaluable contributions that have been made to the fields of medical genetics, research, and healthcare.

The journey of human genetics has been one of profound discovery and transformative impact on both diagnostic and research fronts. As the head of the Department of Pathology, I am acutely aware of the critical role that genetics plays in the realm of diagnostics. Over the past five decades, the division has played a pivotal role in advancing our understanding of human genetics and how it influences health and disease.

The Division's commitment to conducting research among neglected populations, such as indigenous African populations is particularly impressive. By focusing on the genetic aspects of diseases prevalent in these populations, you made a profound impact on global health disparities. Furthermore, it is essential to recognize the Division's incorporation of Bioinformatics and Big Data analytics, as well as Artificial Intelligence in the field of Human Genetics and Genomics. These efforts have not only enhanced the precision and efficiency of genetic analyses but have also driven change in education and training.

Your efforts towards capacity building are commended as you continue to equip future scientists and pathologists with the skills to harness these technologies effectively. The division is ensuring that the next generation of pathologists/specialists is well-prepared to navigate the complexities of modern genetics and genomics.

As we celebrate this momentous occasion, I want to express my appreciation for the collective dedication of you and your team in advancing the frontiers of human genetics. The work has not only elevated the reputation of the University of Cape Town but has also enriched the global scientific community. I look forward to witnessing the continued growth and impact of the division in the years to come.

Once again, congratulations and may your future endeavours be as fruitful and enlightening as your past achievements.

Yours sincerely,

Professor Komala Pillay Head of Department







Department of Medicine

Active in clinical service, health education and research

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> Tel: +27 21 406 6200 Fax: +27 21 448 6815 Email: Ntobeko.Ntusi@uct.ac.za Website: www.medicine.uct.ac.za

Ntobeko A. B. Ntusi BSc (Hons) MBChB FCP(SA) MD DPhil Cert Cardiol Phys SA Professor of Medicine

Chair and Head, Department of Medicine, University of Cape Town and Groote Schuur Hospital Director, SAMRC Extramural Unit on Intersection of Noncommunicable Diseases and Infectious Diseases Principal Investigator, Hatter Institute for Cardiovascular Research in Africa, University of Cape Town Principal Investigator, Cape Universities Body Imaging Center, University of Cape Town Collaborating Investigator, Wellcome Centre for Infectious Diseases Research in Africa Director, Cardiovascular Magnetic Resonance Congress of South Africa Editor-in-Chief, South African Heart Journal Associate Editor, Circulation



10 October 2023

Professor Raj Ramesar Head: Division of Human Genetics Department of Medicine University of Cape Town

Dear Raj,

It is a great honour to write in my capacity as the Chair and Head of the Department of Medicine at the University of Cape Town and Groote Schuur Hospital to extend my warmest and sincere congratulations to you and colleagues and students in Division of Human Genetics at the University of Cape Town on the momentous occasion of your 50th anniversary. This milestone is a testament to the enduring commitment and unwavering dedication of your Division in advancing the field of Human and Medical Genetics on this continent and beyond.

Half a century ago, the establishment of the Division of Human Genetics marked a significant moment in the history of the Department of Medicine, with the appointment of Professor Peter Beighton, as the first Full Professor of Human Genetics. Over the years, we have witnessed the incredible growth and evolution of this discipline under his leadership until 1999. This was followed by further growth under your inspired leadership and stewardship since then. I wish to acknowledge the formidable challenges you have faced, particularly concerning limited resources. The commitment of your staff to providing excellent clinical services, even in the face of adversity, is nothing short of inspiring. The pursuit of excellence in patient care and teaching and training, often against daunting odds, reflects the core values that have guided your Division throughout its rich history.

One of the most remarkable aspects of your Division's journey has been its unwavering focus on clinical diagnosis through the Medical Genetics Unit. Your Division's dedication to serving patients in need of specialised genetic evaluation and counselling has been a beacon of hope for countless individuals and their families. The impact of your clinical work extends far beyond the laboratory, as you provide solace and guidance to those grappling with complex genetic conditions.

Moreover, the advancements in diagnostic technologies over the years have been a game-changer for patient care. The improved precision and speed of genetic analyses have not only enhanced our ability to diagnose genetic disorders but have also paved the way for personalised treatment strategies. Your Division's utilisation and promotion of these cutting-edge technologies, through the many multidisciplinary teams you work in, has undoubtedly led to improved patient care outcomes, offering hope and better quality of life to those affected by genetic conditions.

As we celebrate this momentous occasion, I wish to express my profound appreciation for your Division's resilience, dedication, and remarkable contributions to our Department and the field of Medicine. Your work

has been a source of pride for our institution and has set a standard of excellence that continues to inspire us all. In the face of challenges, you have demonstrated that a steadfast commitment to patient care, clinical excellence, and innovation can yield profound results. Your Division's legacy is a testament to the power of science and compassion in improving the lives of individuals and communities.

Congratulations once again on this remarkable 50th anniversary. May your Division's journey continue to be marked by progress, compassion, and unwavering dedication to the betterment of healthcare and the human condition.

Yours,

Ntobeko Ntusi

M.B. A. Marsi

Professor of Medicine and Chair & Head of Department University of Cape Town & Groote Schuur Hospital

Message from the Head of the Clinical Genetics Unit

Associate Professor Karen Fieggen

I am Karen, a paediatrician and medical geneticist who has been grateful to call UCT Humgen my work home for the last 20 years (scary thought...). I first encountered the division as a junior doctor looking to fill a gap while waiting to get into paediatrics (my first love – I haven't given up my baby stethoscope yet) in the late 1980's. I was invited by Prof Beighton to join genetics and "see the world". Not sure if Albertinia and Springbok count as the world but the work was definitely stimulating and interesting, even though I thought then that it was a bit too depressing for me . So many unanswered questions, so few treatment options. However, the genetic bug had bitten, and I returned after completing my paeds training and working as a general paediatrician, a bit older even if not wiser but much more appreciative of the holistic care that we could offer in medical genetics.

Since that time, I have been so fortunate, working with amazing people from diverse backgrounds at every level and in every area of the discipline. I have learnt so much, grown both personally and professionally and been constantly humbled by the compassion and passion for the care and health of those affected by genetic disorders shown by every person I have encountered in Humgen.

In those 20 years, so much has changed and watching the genomic revolution has been thrilling! There is surely no speciality in medicine that has changed more in 2 decades than ours. Diagnosis, prognosis and treatment approaches have been elucidated, redirected, and accelerated. My only sadness is that still, for most of my patients, the cost of much of this remains out of their reach. However, having witnessed the remarkable growth of African genomics in the last decade, I am hopeful that collectively we in Africa can and will develop solutions in genetics and genomics that will be of benefit to all. not just the well-resourced. What a true privilege to be able to say that I cannot imagine a place I would rather be than right here, right now than celebrating 50 years of UCT Humgen!



Message from the Convenor of the Genetic Counselling Programme

Associate Professor Tina-Marié Wessels

I am Tina-Marié Wessels and I have been the course convener of the MMedSc in Genetic Counselling at UCT since 1 April 2014. I obtained my MSc(Med) in Genetic Counselling and my PhD at the University of the Witwatersrand in 1996 and 2013 respectively.

When the lecturer's position in Genetic Counselling at UCT was advertised in 2013, I was excited at the opportunity to join a world class university. It was a relatively "young" programme, and I thought it would be a new challenge to help grow the field. At that stage I did not know how much I would learn and how many opportunities for growth and personal development I would have.

Prof Jacquie Greenberg, who can be regarded as the grandmother of genetic counselling in the Division, together with other colleagues, laid the foundation for a programme of excellence. UCT attracted my predecessor, Pat Craig who together with Prof Greenberg started the first formal training programme in 2004. After much motivation and persistence, the division was finally successful in creating a permanent lecturer's position which enabled me to move to the Cape. We are very proud to say that by the end of 2023, we would have graduated 25 genetic counsellors. The UCT program has a number of firsts, including the first PhD qualified genetic counsellor, Zandre Bruwer and the first African genetic counsellor, Malebo Malope.



UCT's graduates have gone on to make major contributions to the field both in private and state practice. Although it is regrettable that many of our graduates have had to take up positions in other countries, we are very proud to know that our graduates are highly sought after. Even to the extent that Kalinka Popel has been appointed as the course convener of the genetic counselling degree in Ghana, and Katryn Fourie as the course convener at the University of the Witwatersrand. We also contributed to Oman's genetic counselling services. Dr Bruwer was the first masters level genetic counsellor in the country, and our programme assisted them with training more genetic counsellors, enabling them to develop their own degree in Genetic Counselling.

The genetic counselling programme has always been committed to providing services to all in South Africa and as a result have been dedicated to training other health care professionals in medical genetics and genetic counselling. We have offered occasional courses to both South African health care professionals as well as health care professionals in the rest of Africa. This has contributed to increasing the workforce that will make the translation of genomics research into

health care possible. We are committed to provide genetic counselling to all individuals and their families in South Africa and will continue to train Master's level genetic counsellors, other health care professionals in the realm of genetics and to perform quality research to ensure services are evidence based.

In Memory

Emeritus Professor Peter Beighton 1934 - 2023

It is with profound sadness that we paid tribute in 2023 to the remarkable life of Emeritus Professor Peter Beighton, our founder. We, in the Division of Human Genetics, honour a remarkable leader and mentor who helped introduce and transform the field of Human Genetics in South Africa.

Professor Beighton's academic journey began with his medical training at St Thomas' Hospital in London, where he laid the foundation for his exceptional career. From there, he went on to the United States to work under the tutelage of the Godfather of Medical Genetics, Dr. Victor McKusick, a legendary figure in medical genetics (of OMIM and other fame), at Johns Hopkins Hospital/University in Baltimore. This pivotal experience, and working with the leaders in the field of human and medical genetics, deepened his understanding of the field and ignited a passion within him that would shape the course of his life's work. Fuelled by an insatiable curiosity and keen observational eye, Professor Beighton's thirst for knowledge led him to explore uncharted territories. He undertook groundbreaking research unravelling the mysteries of genetics in the most remote corners of the world e.g. the Sahara Desert. His quest for understanding then took him to enigmatic geographical locales such as Easter Island, Tristan da Cunha, and St Helena where he expanded the boundaries of his scientific endeavours. In 1973, Professor Beighton's exploratory journey brought him to South Africa, where he joined the University of Cape Town (UCT) and Groote Schuur Hospital. Under the guidance of Professor Stuart Saunders (Head of the Department of Medicine at that time, and subsequently the vice Chancellor of UCT), he established the Department of Human Genetics within the Department of Medicine at UCT.



Throughout his illustrious career, Professor Beighton's dedication and expertise led to an astonishing body of work. With over 430 peer-reviewed publications to his name, 34 chapters in books, and 20 monographs and editions, his research resonates across the scientific landscape, continuing to shape our understanding of inherited disorders of the skeleton and connective tissues. His contributions have become the cornerstone of our field, enlightening countless researchers and inspiring generations to come. Beyond his scientific achievements, Professor Beighton was a compassionate and supportive mentor to all who had the privilege of working alongside him. His guidance, wisdom, and belief in the potential of staff and students have empowered us to push the boundaries of our own capabilities. He nurtured an environment of collaboration and intellectual curiosity, fostering a community of scholars driven by a shared purpose to unlock the mysteries of the human genetic code.

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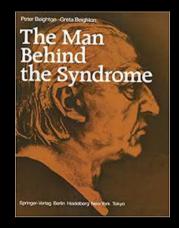
In Memory

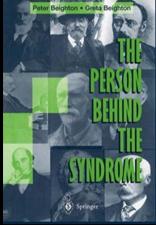
Emeritus Professor Peter Beighton 1934 - 2023

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Professor Beighton's tireless efforts were acknowledged and celebrated throughout his remarkable journey. His accolades included the Gold Medal of the British Orthopaedic Association, the President's Medallion of the South African Orthopaedic Association, the Smith & Nephew literary award, and the Silver Medal of the South African Medical Research Council. In 2002, he was the inaugural recipient of the esteemed Order of Mapungubwe - bronze, bestowed upon him in recognition of his lifetime achievements as a scientist and his unparalleled research into human genetics generically and inherited disorders of the skeleton specifically. We not only mourn the loss of an extraordinary scientist and visionary leader but we also celebrate a life well-lived and the enduring legacy of Professor Peter Beighton. Throughout his remarkable life, Professor Beighton Peter Beighton and his wife Greta Beighton shared an interest in the history of medical genetics and have published two unique volumes of brief biographies of people for whom genetic syndromes have been named. Prof and Greta were always a team. Sadly, Greta died in Cape Town on 29 May 2017, and this took a toll on Professor Beighton.

His impact has reached far beyond the confines of our institution, extending into the very fabric of scientific exploration and the hearts of all those who have been touched by his wisdom and guidance. May his indomitable spirit and insatiable thirst for knowledge continue to inspire us as we carry his legacy forward, advancing the frontiers of Human Genetics in his honour.











UCT Division of Human Genetics



Milestones



1973

- Official start of Department of Human Genetics
- Description of "The Beighton Score" (now internationally used) to assess joint mobility by Prof. Peter Beighton

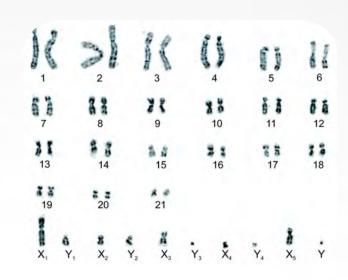


1972

- Prof. Peter Beighton has First Chair in Human Genetics (Dept of Medicine) at UCT
- Cytogenetics Unit moves from Physiology Department into Human Genetics
 - Routine chromosome analysis on blood samples is introduced

1976

 Ronald Smart is one of two Genetic Counsellors registered with the South African Medical and Dental Council



1980s

Cytogenetics unit introduces semi-automated karyotyping system

1978

- First multiracial Huntingtons Disease clinic at GSH
- First MD doctoral graduate and first PhD graduate at the Division

1983

 Prof. Peter Beighton is the Director of the newly established SAMRC
 Research Unit for Skeletal Disorders



L to R: Dr Rebecca Felix (first PhD student on Colorectal Cancer Project) on field trip in Wupperthal with Prof. Raj Ramesar and Sr **Ursula Algar (Surgery)**

1990s

- Cytogenetic unit introduces fluorescence in situ hybridisation (FISH)
 - First Colorectal Cancer outreach trip to the Northern Cape, which eventually became a multi-disciplinary effort including the Pink Drive
 - · Establishment of DNA biorepository

1984

 Human Genetics **Molecular Laboratory Opens**

1987

• DMD/BMD diagnostic

service implemented by Dr Robea Ballo

· Establishment of the **Inherited Retinal Disorders Research** Group funded by Retina **South Africa**

UCT joins international genetic research project

The Departments of Human Genetics and Psychiatry at UCT have joined an international research project into the genetic origins of Bipolar Affective Disorder - commonly known as

Disorder - commonly known as manic depression.

The research project, funded by the pharmaceutical compa-ny, Sandoz Pharma Lld of Swit-zerland, will contribute to growing research into the ge-netic basis of psychiatric disor-ders.

netic basis of psychiatric disor-ders.

Dr Raj Ramesar of UCT's
Department of Human Genetics
and Dr Francois Daubenton of
the Department of Psychiatry
visited Sandoz Pharma in Ba-sel, Switzerland, earlier this year

"What has become blatantly clear is that diseases which appear outwardly similar differ at the fundamental etiological level - and are likely to warrant distinct therapeutic and menagement modalities."

"For this reason it is vital that a strong research basis be maintained which aims at identifying the genetic basis of disease in South African populations."

Dr. Ramesar says that while considerable effort has gone into determining the molecular genetic basis of Mendelian diseases (those that follow simple patterns of inheritance, and



Seen in Basel. Switzerland, are members of the international Seen in Basel, Switzerland, are members of the international team undertaking genetic research into psychiatric disorders, from the left, Dr Francois Daubenton of UCT's Department of Psychiatry, Dr Raj Ramesar of UCT's Department of Euman Genetics, Dr John Kelsoe of the Department of Psychiatry, University of California, San Diego, and Dr Hermann Lübbert, Head of the Genome Centre, Sandoz, Switzerland.



1996

 Establishment of Neuropsychiatric **Genetics Group:** "Genetics of Bipolar Disorder" with Funding from Novartis, **Switzerland**

1991

 Establishment of computer-based database

> Ophthalmic Genet. 2002 Mar;23(1):61-5. doi: 10.1076/opge.23.1.61.2207.

A computer-based register for inherited retinal dystrophies in Southern Africa

M T Rebello 1, L J Greenberg, R S Ramesar

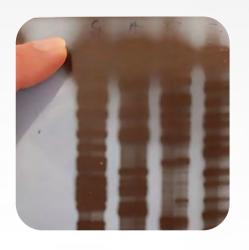
Affiliations + expand

PMID: 11910560 DOI: 10.1076/opge.23.1.61.2207

1998

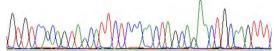
 Joint Honours programme launched (Cell Biology and Human Genetics) which eventually grew to become the BSc (Med) **Honours Program**

- Switch from radioactive (P32)-labelled sequencing to automated flourescent sequencing (first ABI sequencer acquired)
- Establishment of Colorectal Cancer Research Group
 PI: Prof. Raj Ramesar





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2001

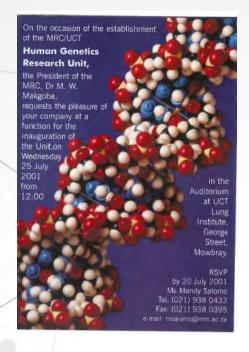
 Cytogenetics unit incorporated into the National Health Laboratory Service (NHLS)

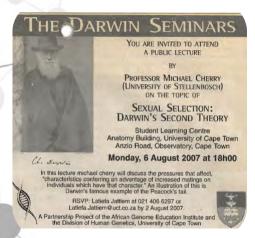


2000

- Prof. Peter Beighton retires
- Prof. Raj Ramesar is the new Head of Division







- Establishment of SAMRC Human Genetics Research Unit Director: Prof. Raj Ramesar
- First graduated Genetic Counsellors
- Division Moves from Anatomy Building to IDM
- First Darwin Seminar
 for the publichighly popular

2002

 Emeritus Prof. Peter Beighton receives the Order of Mapungubwe (Bronze) from President Mbeki



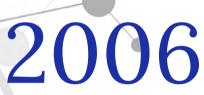
2004

 MSc Genetic Counselling Course started- Convenor: Prof. Jacquie Greenberg



Pioneers: (From left) Assoc Prof Gonda Perez, with MSc students in genetic counselling, Brenda Kruger and Frieda Basson, at an oathtaking ceremony for the four students on the course.





NHLS acquires
 CytoVision, a
 semi-automated
 karotyping system



• Establishement of Pharmacogenomics Research

Prof. Raj Ramesar receives
 Alan Pfifer Award for
 Community work relating to

Group
PI: Prof. Collet Dandara



Group



2007

 Cytogenetics and molecular diagnostics moved from the medical campus to the C21 NHLS unit at GSH







- First Learners' Open Day
- Establishment of H3Africa





L to R: Prof. Raj Ramesar, Prof. Michele Ramsay (Past President of AFSHG) and Prof. Charles Rotimi (Founding President of AFSHG)

2011

• First PhD in genetic counselling at Division in the **Division: Dr Zandre Bruwer**



• First Joint AFSHG and SASHG conference

 Honours in Forensic Genetics introduced at **UCT**



Next Generation
 Sequencing platform established

2013

- First whole genomes sequenced on African soil
- Induced pluripotent stem cells (iPSCs) introduced to study SCA7

2016

AFSHG
 headquarters shift
 to Division of
 Human Genetics at
 UCT



- · First Black African Genetic Counsellor in South Africa: Malebo Malope
- **Establishment of Sickle Africa Data Coordinating Center** (SADaCC)
- · Establishment of Individual Findings in Genetic Research in Africa (IFGeneRA) Project
- Establishment of Hearing **Impairment Genetics Studies in Africa (HI-GENES Africa)**

PI of all three of the above: Prof. **Ambroise Wonkam**



2019

- DNA microarray: the first "next generation" technology introduced into the scope of the diagnostic genetic service
- Formation of the African **Pharmacogenomics** Consortium/ Network (APN) PI: Prof. Collet Dandara



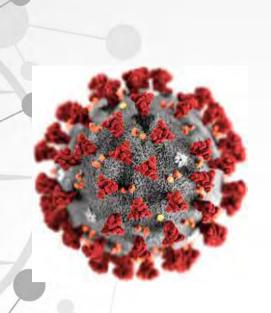
2018

- · Establishment of **Precision and Genomic Medicine** SAMRC Extramural **Research Unit** Director: Prof. Raj Ramesar
- · Launch of GeneMAP **Research Centre** PI: Prof. Ambroise Wonkam









Prof. Ambroise
 Wonkam receives
 Alan Pfifer Award



2022

• Establishment of SAMRC
Extra-Mural Unit:
Platform for
Pharmacogenomics
Research and Translation
Director: Prof. Collet
Dandara





- Prof. Ambroise Wonkam receives SAMRC Gold Scientific Merit Award
- Establishement of Public Understanding of Big Data in Genomics Medicine in Africa (PUBGEM-Africa)
 PI: Prof. Ambroise Wonkam



2023.....coming home





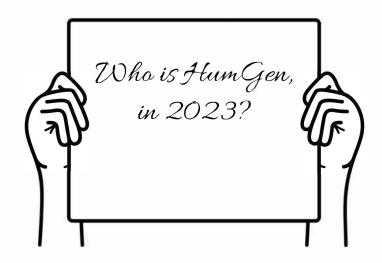
• HumGen Attendees at the International Congress of Human Genetics at the CTICC in Cape Town, organised by the African Society of Human Genetics and the Southern African Society of Human Genetics

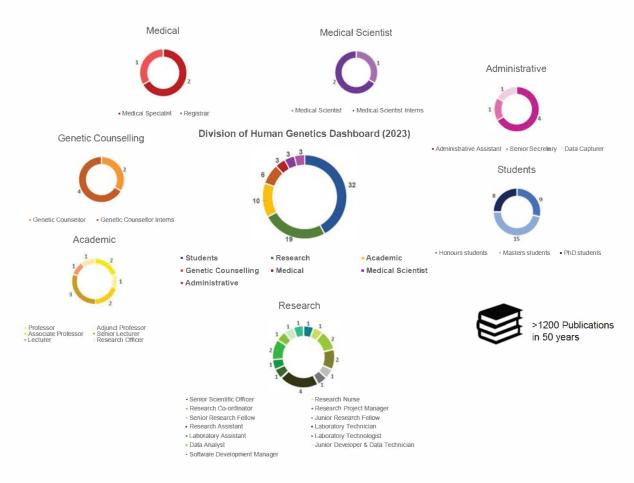


• Celebration of DNA Day

• Prof. Ambroise Wonkam receives HUGO African Prize









Hum Gen In the News

Surgeon dedicates award to team

f 🔰 🕓 🗗 in 🤠 🖼 🛨

Africa: UCT Sickle Cell Genetics Study a First for Africa

Wall**A**frica

College honours Beighton

How to become a genetic counsellor



Predisposed to

advise on genes

UCT joins international genetic research project



MEDISense

The human genome is the key to infinite doors

MONDAY

UCT graduate makes genetic breakthrough

The promise of the African genome project



International consortium, including UCT's Geneticists, identifies

Gene research breakthrough offers hope to osteoporosis sufferers worldwide



Mail&uardian

SA researchers snare blindness gene

f y in O

UCT researchers make major

blindness discovery

mGenAfrica: Introducing learners to careers in STEM



South Africa at coalface of genetics research



Stem cell technology aids disease research



UCT finds vital clue to cause of

sight affliction



I dead in the builtroom assiss in Memphis by r Alden on Assess IS





Rare Disease Day



Social Responsiveness



Learners Open Day



End of Year Functions



Spring Day









HUMGEN ALUMNI – WHERE ARE THEY NOW?

Aisha Pandor

Where are you/what are you doing now?

I'm an entrepreneur and founder of a software platform called SweepSouth. I'm really excited to be exploring again in the biotech space, in precision medicine specifically.

Time in the Division (Years and Position)

2006 to 2012 (Hons, Masters and PhD student)

What group were you in?

Retina group

Favourite memory of your time in the division

It was generally such a positive experience, and what sticks out were the great relationships and conversations with wonderful, smart people who really care about the work they're doing.

Alison September

Where are you/what are you doing now?

I am a professor at UCT within the Division of Physiological Sciences

Time in the Division (Years and Position)

I started in the Division the year we had our first democratic elections Feb 1994, as a research assistant in the Retina Project and completed all my postgraduate qualifications within the Division, in the Anatomy building. I worked with both Profs Greenberg and Ramesar. I left after my PhD qualifications in 2003.

What group were you in?

Retina

Favourite memory of your time in the division

We had the most amazing working environment and it was so much fun! Prof Beighton's legendary seminars were epic memories, conferences with the SASHG team, end of year parties were a blast and Raj spontaneously delivering pizzas when we were doing an all-nighter in the labs bring back fond memories.

I congratulate the team Human Genetics on reaching this 50th milestone! Many, many achievements are noted during this pioneering journey at UCT and I believe many more events will form part of the future history at UCT and SA. I salute you all! When you join HUMGEN you remain a HUMGENNER for life, its after all in your genes.





Anna-Susan Marais

Time in the Division (Years and Position)

I never worked for UCT Humgen but with Humgen. Many good years with wonderful memories.

I was a genetics nurse with the Department of Health, Genetic Services. We conducted annual genetic screening clinics in the rural towns of the Western Cape Province from 1982 to 1995. Professor Peter Beighton, Denis Viljoen and many other geneticists accompanied us on these clinics. We took the genetics service to the patient / client.

Favourite memory of your time in the division

We had wonderful experiences, saw many interesting cases and beautiful places, laughed a lot, ate too much – we had awesome memorable times.



Cedrik Ngongang

Where are you/what are you doing now?

Currently based in Washington DC, USA/Medical Geneticist for n-Lorem Foundation, a company developing and providing Antisense Oligonucleotides for experimental treatment of individuals with ultrarare mutations

Time in the Division (Years and Position)

2013 - 2018, Registrar, and Research Officer

What group were you in?

HumGen group: Clinical Unit, and Wonkam group (Hearing Impairment)



• Celia van der Merwe

Where are you/what are you doing now?

I'm currently Director of the NeuroDevelopmental Variability Initiative (NDV) at the Broad Institute.

Time in the Division (Years and Position)

I was a postdoc at HumGen for 3 years (2016-2018).

What group were you in?

Psychiatric Genetics

Favourite memory of your time in the division

My favourite memory was all the laughs and chats we had while sharing an office space with Heidi, Lisa and Shareefa.



Where are you/what are you doing now?

Currently working as a Laboratory Technician for SADaCC.

Time in the Division (Years and Position)

5 Years

Favourite memory of your time in the division

Getting to know people of different backgrounds.

Clair Engelbrecht

Where are you/what are you doing now?

I emigrated to the UK in 2021. I worked in the NHS as a specialist GC for 2.5 years. Currently I'm working as a genetic counsellor at a private fertility clinic in London, UK. Here I provide Genetic counselling to couples undergoing PGT-A, PGT-SR and PGT-M. I may also see patient for carrier Genetic testing or other general indications. Myself and another UCT alumni have opened a private online genetic counselling practice in the UK (evolvegenetics.co.uk)

Time in the Division (Years and Position)

I was at UCT for my MSc Med. This was 2 years. I then moved to Tygerberg to do my internship.







What group were you in?

Genetic counselling. Worked as part of the SCDO working group. I worked part time for Retina SA, through UCT, during my studies.

Favourite memory of your time in the division

The great friends I made. Tina Wessels was a great teacher and leader. Always kind and patient. Very much a part of how I developed into the GC I am today.

Delesa Damena Mulisa

Where are you/what are you doing now?

Currently, I am a postdoc at the Laboratory of Malaria immunology and Vaccinology (LMIV), NIAID, NIH, Bethesda, USA.

Time in the Division (Years and Position)

PhD student (2017-2021)

Favourite memory of your time in the division

I'm always proud to be part of division of Human Genetics at UCT where I learned the science of human genome and its incredible potential to address real world medical problems. I never forget all the great and welcoming people in Human genetic family. You all are exceptionally incredible !!! Of course, the beauty of Cape Town is extra-ordinary and can't be seen anywhere!!!



• Danielle Smith

Where are you/what are you doing now?

A molecular diagnostics specialist in Cape Town, working as a private consultant.

Time in the Division (Years and Position)

2008 - 2015 (Hons, MSc, PhD, Postdoc)

What group were you in?

Cancer group for Honours, then Neurogenetics for MSc onwards.

Favourite memory of your time in the division

Lunches in the tearoom, chair ballet, soup day and drinks at the MAC



Denis Awany

Where are you/what are you doing now?

I'm currently a Bioinformatics postdoctoral research fellow at the South African Tuberculosis Vaccine Initiative (SATVI), in the Faculty of Health Sciences at UCT.

Time in the Division (Years and Position)

I was a PhD student in the Division, between 2017 and 2021.

What group were you in?

The Genetics & Genetic Epidemiology research group.

Favourite memory of your time in the division

My favourite memory of the division is the African Day events in the division, when people could show their diversities, from dress codes to foods.



Where are you/what are you doing now?

I am retired from Groote Schuur and the clinical Genetics team but still wear my nurses cap part time in the private sector.

Time in the Division (Years and Position)

After many years of being a critical care nurse at Grotties , I decided I needed a change of pace and environment. After a quick tutorial on chromosomes and genes from an ICU colleague I made my way over to Health Sciences to meet Professor Peter Beighton. I was so taken with his charm, intelligence and knowledge, and also by his warmth and I knew this would be my home for years to come. For the next 20 years I worked in this wonderful environment of human genetics side-by-side with Professor Beighton and other wonderful medical and nursing colleagues as well as being introduced to a world of Cyto and molecular genetics.



Favourite memory of your time in the division

Exposure to academia and an amazing scientific world, forging wonderful working relationships and friendships.



Donald Basel

Where are you/what are you doing now?

I am in America and work at the Medical College of Wisconsin where I am Professor Pediatrics, Division Chief: Medical Genetics and Medical Director of Genetics: Children's Wisconsin.

I'm also Director of the Center of Excellence - National Organization for Rare Disorders, Director of the Diagnostic Center of Excellence, Undiagnosed Disease Network, and Associate Director: Undiagnosed and Rare Diseases, Mellowes Center GSPMC.

Time in the Division (Years and Position)

I was a trainee fellow under Peter Beighton in 1998/1999

What group were you in?

Clinical

Favourite memory of your time in the division

There are many: Outreach program within the Western Cape with the wonderful nurses, being challenged to a stair climb race by Prof Beighton, Social gatherings with Raj, Jackie and other lab and members of the department.



Where are you/what are you doing now?

After my PhD which I completed in 2014 with a thesis on Huntington disease, I left Cape Town for the "big bad city" (i.e. Johannesburg J). I joined the Division of Human Genetics at the University of the Witwatersrand in 2015 as a Claude Leon-funded postdoctoral fellow under the mentorship of Prof Amanda Krause, one of the few researchers in the country who shares a keen interest in the genetics of HD and HD-like disorders. Over the next five years, I worked on several research projects including HD phenocopies, inherited breast cancer, and developmental disorders. I also completed the required internship and registered with the Health Professions Council of South Africa as a Medical Biological Scientist in Human Genetics.



In 2020, I was appointed Senior Medical Scientist at the Division and then in 2022, moved up to Principal Medical Scientist. I currently have a significant diagnostic mandate a focus on the application of genomic technologies (chromosomal microarray and next-generation sequencing) to molecular diagnostics within the National Health Laboratory Service. I have maintained my interest in HD genetics and inherited cancers, and have supervised (and continue to supervise) postgraduate student projects under these topics. Concurrent with my service role, I continue to



expand my skills and knowledge in the analysis and interpretation of DNA variation (both structural and sequence level).

On a personal level, I happen to be married to a fellow alumnus of the Division – we were not in the lab at the same time, as he was a few years ahead and there are three or four members of the Division (past and current) who deserve some credit for this relationship. We will forever be grateful to them!

Time in the Division (Years and Position)

2007: BSc (Med) Honours Human Genetics 2008 - 2010: MSc (MED) Human Genetics

2011-2014: PhD Human Genetics

What group were you in?

My Honours was with the Retina group, supervised by Drs Lisa Roberts and George Rebello. My MSc and PhD were with the Neurodegenerative disease group, under Prof Jacquie Greenberg.

Favourite memory of your time in the division

I met, and made friends with some of my favourite people to date! My fondest memories are probably around the group lunches, as this most often ended up being the funnest and funniest hour (actual tears because we were laughing so hard), but occasionally there were deep life-changing conversations around some fundamental aspect of life OR genetics.

Also, every 'eureka' moment I had in the lab when something challenging FINALLY worked (I see you SSCP gels).

• Frieda Loubser

Where are you/what are you doing now?

Genetic Counsellor in Cape Town: Private Practice

Time in the Division (Years and Position)

2004 - 2013 First Genetic Counsellor (one of two to do the MSc Genetic Counselling course), worked as GC on contract posts at GSH, RXH.

What group were you in?

Genetic Counselling

Favourite memory of your time in the division

2010 world cup soccer: good memories of playing mini soccer at the department.



Gasnat Shaboodien

Where are you/what are you doing now?

I am A/Prof Gasnat Shaboodien and I am the Group leader and Director of Cardiovascular Genetics (CVG) at the Cape Heart Institute (CHI) at UCT.

Time in the Division (Years and Position)

I joined the division from January 2002 to December 2010 where I worked as a research assistant on the Cancer project with Prof Raj Ramesar. I then switched to the Cardiovascular Genetics group, that was also part of HumGen, where I did my PhD with Prof Bongani Mayosi.

What group were you in?

Cancer, Cardiovascular Genetics.

Favourite memory of your time in the division

Hmmm, so many! The division of Human Genetics treats everyone like family. Even to this day, I can just pick up the telephone and call up Raj to ask for a quick chat/advice and he's there; or if I need help in the lab, I know they would be willing to assist! I often get my old colleagues where we rehash the good old days...even thinking about it now makes me smile. My favourite memories would have to be playing dressing-up for Heritage day and Spring day, and I'll throw in Halloween just for fun! I am sure those blackmail pictures are still floating around in the division somewhere! Happy 50th to the Division of Human Genetics! May you go from strength to strength and continue to inspire the next generation of scientists!

Gemma Louise Carvill

Where are you/what are you doing now?

I'm an Assistant Professor and lead my own lab of about 12 people at Northwestern University in Chicago. We focus on the genetic etiologies of the epilepsies, using cutting edge genomic approaches to identify new genetic etiologies, and we use patient-specific cellular models to study pathogenic mechanisms. I also co-direct the Adult Epilepsy Genetic Program, and teach the first year medical students all things genetics. Learn more about our work and the fun we have in lab (a HumGen tradition being continued – we are big on fun) here: https://sites.northwestern.edu/carvilllab/

Time in the Division (Years and Position)

2005: Bsc Hons student 2006-2010: PhD student

What group were you in?

Neurodevelopmental disorders with Dr Rene Goliath





Favourite memory of your time in the division

- 1. Chair Ballet
- 2. Lunches at the end of the hall and the many giggles, so much so that we were occasionally told to hush
- 3. That cold room and all the ridiculous 'equipment' that we wore to go in there.
- 4. The MAC NSFW!
- 5. The highly collaborative nature and supportive environment at HumGen, in particular Lisa Roberts and Alvera Vorster and how they had endless patience with the trainees and were so giving with their time and expertise.

George Rebello

Where are you/what are you doing now?

I am currently at JDJ Pathology in KZN where I am helping to setup a Molecular/Genomic laboratory. We are keen to offer private precision medicine services, probably starting with molecular Cytogenetics and Pharmacogenomics.

Time in the Division (Years and Position)

I started in Humgen in Dec 1997 and primarily worked in the retina group. I had the pleasure of being part of the original RP17 group who worked like mad things to isolate (localise as it turns out) the RP17 gene. I worked with Raj on setting up the original bioinformatics training in Humgen and was involved in the move from Anatomy to WBN. I stayed in Humgen till 2008 when I ventured into private enterprise for a while. I have since returned to my ancestral home



(Humgen) and have been working with Raj on a number of projects, helping a number of students to finish "old" projects. I have worked with Raj on the Colorectal Cancer Project for a few years now and I am always excited to see how "cutting edge" the work in the Division is.

Favourite memory of your time in the division

My favourite memory is a bit complicated and spreads over a number of years: Humgen has always felt like a part of the family, my daughters had Humgenners at baby-showers, birthdays, and all the other landmarks in their lives. We went on flower-trips, had braais, attended functions at the MAC, and generally did a whole lot of stuff together. The staff, led by Raj and Jacquie Greenberg were always there to help out and share the load and the joy of living in Cape Town. My daughter's Godmother is a Humgenner (Lisa Roberts), my best friends are people I met through the lab, my best memories of the last 26 years often involve people from Human Genetics who shared so many experiences; Chair ballet with Skollie (the very respectable Janine Scholefield), Jolly hockey sticks (equally respectable Gemma Carvill), Lisa (serious scientist and leading retinal researcher) and the others from the "zone". Crazy photo sessions with (the now very respectable Prof) Gasnat Shaboodien. Dinner with Jacquie and Ivan where PB (Prof Peter Beighton) often told revealing and hilarious stories.

Dinner at the home of Raj and Jenny (the best cook in Humgen), where I had the pleasure of meeting some of the leading scientists in the world who came to visit, and were always treated like part of the family. and literally hundreds of times we just had group tea and cake to celebrate "jelly babies" or a degree or some other life event with my family – Human Genetics.

It has been my pleasure and joy to be associated with Raj who has always been a steadfast and supportive mentor and friend and has always led the charge to "enjoy the moment", no matter how challenging the current "crisis" may be.

Heidi Paulse

Where are you/what are you doing now?

Currently working in the Department of Obstetrics and Gynaecology – PA to the HOD

Time in the Division (Years and Position)

Worked in the Division of Human Genetics from Jan 2015 – June 2022 as the Senior Secretary

What group were you in?

Admin group but assisted all the various groups

Favourite memory of your time in the division

Loads of memories especially all the Divisional functions – Great Gatsby function stands out the most.



Where are you/what are you doing now?

I am a manager at Allan Gray, one of the largest asset managers in SA.

Time in the Division (Years and Position)

Mid 2012 – Start 2017, PhD student

What group were you in?

Cancer

Favourite memory of your time in the division

The friends I made in the Division were priceless.

The opportunity to live in Switzerland for the Next Generation Scientist Program. UCT Indoor Olympics at the MAC.

We would always find time to celebrate the little things too – soup day in Winter, Diversity day, secret Santa etc. I always thought the Learners Open Day was a great milestone for giving back and





getting young minds excited about science. The Darwin seminars and having world renowned scientists visit were also notable.

Irene Appolis

Where are you/what are you doing now?

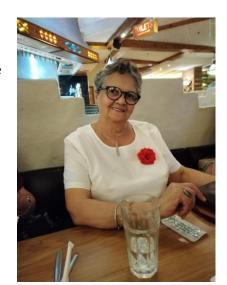
I'm living in Pinelands Grove retirement complex. I moved in year 2020. It was the time with epidemic of Covid. I settled nicely. We have lots of activities. Linedancing. Keep fit classes. Fellowship. Bible discussions. Knitting. Card games. Walks to Howard centre.

Time in the Division (Years and Position)

I spent 35 years in cytogenetics and three years in animal unit as laboratory assistant.

What group were you in?

My work load was assembling water distillers. Sterilized all apparatus. Worked in the dark room glazing prints. Preparing and sterilizing work for tissue culture and blood culture. Handling case dockets which was regarded as strictly confidential.



Favourite memory of your time in the division

My favorite memories was when Dr Raj Ramasar joined the department. Then became head of department.

Jacquie Greenberg

Where are you/what are you doing now?

I am retired and live in Pinelands Grove Retirement Village in the Western Cape. SA

Time in the Division (Years and Position)

When PB came and started the Division of HumGen in 1972, Ronnie & Pat Smart, Fran Bayley, Glynnis Schutte and I joined him and started the Cytogenetics Lab. From there I moved around the Division from Late Onset Genetic Diseases (Huntington's Disease) & the Inherited Ataxias (Spinocerebellar Ataxia), to Retina, to Muscular Dystrophy (Duchenne Muscular Dystrophy) as well as Genetic Counselling. I became Emeritus Professor in 2018.



Favourite memory of your time in the division

For me that has to be when PB gave his inaugural lecture in 1972 and said: " If you want to see the world, join Genetics". Three of us got up and went down to sign up with him and the rest is history. Needless to say, I did get to see the world thanx to PB & Genetics and loved every moment of the journey.

Janet Lyons-Lewis

Where are you/what are you doing now?

I work for an American-headquartered biotechnology company (Amgen) as a Global Study Manager, project managing investigator-led clinical research studies in leukemia (both adult and paediatric) in EMEA and APAC regions. I have worked here for 7 years. Before that I worked in a leading London Hospital Trust, managing the haematology clinical trials portfolio in multiple myeloma and lymphoma.

Time in the Division (Years and Position)

I was an Honours student in HumGen lab in 2006, under supervision of the wonderful Jacquie Greenberg and worked on a project investigating Huntington's disease, and then worked for a very short time as a Research Assistant with Jacquie in 2007 until April, when I then left South Africa to move to London, UK. I am still in London now, 16 years later!!



Favourite memory of your time in the division

Memories from my time in HumGen are both happy and nervous! Nervous in that I feared tests and exams(!) as part of the Honours degree, but happy in that the people in the HumGen department welcomed us students with open arms and friendly faces. I loved the family-feel of the department, the dress up days, I remember Lisa and Gemma 'suiting up' in what seemed like 100 layers before going into the -80 freezer to get samples. I also remember the legendary CT wind/rain whipping horizontally on the walk from the car park up to the med school buildings and being soaking wet for the reminder of the day. I also remember toward the end of the year unscheduled power failures beginning to happen (often disrupting a western blot or PCR run!)... in what I guess was the start of loadshedding.

Janine Blanckenberg

Where are you/what are you doing now?

I'm a Senior Scientist in the Sequencing Assay Development Department at Roche, Cape Town. We develop new sequencing reagents and kits.

Time in the Division (Years and Position)

I was in HumGen from 2003 to around 2007. I completed my Honours and Masters in HumGen. I started my PhD in HumGen but the Cardio Genetics group later moved to The Hatter Institute.

The Cardio Genetics group.

What group were you in?



Favourite memory of your time in the division

There are so many good memories J I remember there being a real good vibe in HumGen. There was always lots of food for birthday celebrations and get-togethers. Gasnat Shaboodien and I at one time used to collect "birthday money" each month from colleagues to buy the birthday person flowers on their birthday. Janine "Skollie" Scholefield and I were PhD students at the same time in the Division. When there were phone calls for "Janine" this would lead to many more questions from our side to figure out which Janine they were looking for. And, of course, chair ballet!

Janine Scholefield

Where are you/what are you doing now?

After completing my PhD in Human Genetics at the University of Cape Town (UCT), I spent three years as a Nuffield Medical Fellow at the University of Oxford. Since returning to South Africa, I have specialised in cellular modelling of disease establishing induced pluripotent stem cell (iPSC) research at the Council for Scientific and Industrial Research (CSIR). My research interest lies within in developing physiologically relevant cellular models of disease using advanced technologies, including stem cells, genome engineering, and microscopy, especially within the unique context of the diverse sub-Saharan African genetic background. Using these techniques, the team's goal is to promote incorporation of pre-clinical models in global drug development pipelines. I currently hold the position of Research Group Leader of the Bioengineering and Integrated Genomics Group (BIGG) at the CSIR.



I further hold a Senior Honorary lectureship position in the Department of Human Biology at UCT, and an Honorary lectureship position at the Department of Human Genetics at Wits. In addition, I am the Editor-in-Chief of the Springer Nature publication Gene Therapy.

Time in the Division (Years and Position)

2000: Hons Cell Biology and Genetics 2004-2005: Masters in Human Genetics 2006-2009: PhD in Human Genetics

What group were you in?

Neurodegenerative diseases – having worked on both Huntington's disease and SCA7, under the guidance of Prof Jacquie Greenberg

Favourite memory of your time in the division

- The time we had to present to Eric Lander.
- The time we had to take J Craig Venter on a tour of the lab.
- The time we had load shedding in the lab in 2007 and played chair ballet waiting for the PCR machines to come back online.
- The time Prof Beighton said to me after advising on one of his projects, "You're either going to win the Nobel prize or be an associate professor." I have yet to accomplish the latter, but I appreciated the complement.

In all seriousness I assume you're looking for a pivotal moment, I experienced in the division? If so, it was walking down the stairs when I asked my mentor and supervisor why we do what we do in genetics, given the power of the science in the context of the usage of the samples we acquire. I asked, "why?".

She responded, "Why not?"

It was a defining moment for me to extricate myself from the intricacies of what we can do and what we should do, given our privilege of knowledge. She was testing me to ensure I understood the importance of the communities we serve. Her response stays with me in all the work I do, in ensuring I constantly ask myself and now, my own team, to consider the potential impact of our research with respect to the people whose lives we seek to positively impact.

Jantina De Vries

Where are you/what are you doing now?

I am an associate professor in Bioethics in the Department of Medicine at UCT and have recently founded The Ethics Lab, in which we seek to apply knowledge from the African humanities to the ethics of new and emerging health technologies.

Time in the Division (Years and Position)

I was a postdoctoral fellow in HumGen for 2 years, from Feb 2011 to Feb 2013

What group were you in?

I worked on the ethics of genetics research in Africa, building on work I had done during my PhD.

Favourite memory of your time in the division

At one point, Prof Ramesar organised a 'suggestion box' where people could submit reflections about their experience of working in the Division. Although some of the notes and conversations that flowed from those suggestions were challenging, I felt that the willingness to have difficult discussions to be really inspiring. I loved my time in HumGen and still collaborate with people in the division.



Jason Bosch

Where are you/what are you doing now?

Postdoc at the Institute of Microbiology of the Czech Academy of Sciences

Time in the Division (Years and Position)

Master of Science in Medicine (MSc (Med)) in Human Genetics, 2012-2013

What group were you in?

Genetic Deafness (Ambroise Wonkam)

Favourite memory of your time in the division

I had many great experiences during my time in HumGen, however my favourite memory is not a specific event. I had a disappointing end to my honours degree which left me demotivated in science. But my time in HumGen and under Ambroise's supervision reignited my passion for science and helped me build a solid foundation for my scientific career.

Jennie Slee

Where are you/what are you doing now?

I am now a clinical Geneticist at Genetic Services of WA in Perth (Perth Australia - tho could be Scotland or South Africa near Polkinghorne). I do adult, prenatal and cancer genetics (despite my years of paediatric training - don't do paed genetics)

I have been in that position since 1998 - doing my training in Cape Town and Perth.

Time in the Division (Years and Position)

At Human Genetics UCT I was medical officer for 12 months - 1990 - and what a great year it was. I wanted to do another year - but Prof Beighton had seen through me by then!

Favourite memory of your time in the division

I think my favourite memory was actually getting the job in the first place. I applied when I was doing another year in paediatrics at Cecilia Makiwane in the Ciskei under the fantastic leadership of David Power. I had a reply that I had an interview with Prof Beighton in Cape Town. I was terrified. I had never had to do an interview before - just apply and wait for a yes/no - so this was serious stuff! I arranged an appointment to coincide with me transporting a child with Fallots and Fallot spells (including one on the plane where I had to insist I sit next to the child and not a distance away - because the flight hostesses could manage all in flight medical emergencies?) from CMH to RXH. I got ushered into the large room with the big mahogany desk at the back, with Prof Beighton facing the door behind the desk. The look was intimidating - and the British Colonel voice heightened it. The interview proceeded well - but I knew I was a goner when Prof said the job was a highly sought





after position. Prof's words to me was that if I wasn't a South African national - I would need to be brilliant (which I wasn't) or of international renown (which I wasn't) to get the position. I had no chance - so I relaxed and let the Slee humour take over. Out his window was the incredible Table Mountain which I was seeing for the first time - so I said to Prof words to the effect of "If I stood stark naked on Table Mountain, I would make the news in both Cape Town and Perth....does that count as international renown?". I got the job! (and I never stood naked anywhere - though others might have a different story - its not true).

Another memory for both Prof and I was after I got called up to GSH Medical Director - or whatever his title was. I had been accused of going up for a second plate of ice cream at the res - and this was actually a serious telling off!! I had to report back to Prof about the reason for the call to the headmasters office. I think he was somewhat pleased! He gave me a great reference on my return to OZ - which I will have to find - alluding to my exploits and long lunches at the pub etc. I actually asked Prof why he gave me the job when I saw him in 2017 - when we out for dinner and only a little lubricated - he did not say a word! A memory I don't have a photo of was Lyn Peterson causing exploding glass from the microwave as she heated up a medium she was using.

PS I'll bet they never employed another Aussie! Having said that - I stayed in touch with Prof and Jackie - and saw them (and Robea - and Michelle Babaya) in 2017/2018 when in RSA with my children.

Katryn Fourie

Where are you/what are you doing now?

I am currently working at the NHLS and WITS as the genetic counselling manager as well as the course coordinator for the MSc Med Genetic Counselling program.

Time in the Division (Years and Position)

2 years, I believe 2014 and 2015.

What group were you in?

Genetic Counselling

Favourite memory of your time in the division

I always enjoyed the teaching blocks. I also have a very clear memory of my first clinical meeting, with Nakita presenting a case, and I remember thinking: What language is she even speaking? I have no idea what any of these words mean! Luckily, I did after the teaching blocks.



Kelly Loggenberg

Where are you/what are you doing now?

After many years in the UK I am currently the in-house genetic counsellor at Next Biosciences which is a genetic testing laboratory with a focus on reproductive genetics. I also work in private practice.

Time in the Division (Years and Position)

Genetic counselling student from 2005 – 2007.

What group were you in?

Counselling

Favourite memory of your time in the division

Certainly the lasting friendships from my time in Division, particularly with the other GC 'students' who have all achieved great things in our profession.



Where are you/what are you doing now?

I am currently 2 weeks away from handing in my dissertation for the completion of my MSc in Quantitative Genetics & Genome Analysis from the University of Edinburgh. My dissertation is building a regression modelling method to Forecast the Genetic Effects of non-European populations using European Individuals. I will be staying in Edinburgh after my degree to find a job specialising in data analysis and public health research.

Time in the Division (Years and Position)

BMedSci (Hons) in Forensic Genetics student in 2021

What group were you in?

Wonkam Group: HiGeneS (Hearing impairment)

Favourite memory of your time in the division

Endless days of lab work in the Ramesar Lab with my classmates. I do miss looking out and seeing Table Mountain and the sunshine everyday. Although a long day, I absolutely loved the final presentation day. When we got to see what everyone had done, and to see where my class is now makes me immensely proud. And the writing retreat to prepare my thesis for publication with the Wonkam Group. Which has gone on to be a pre-print. In addition, presenting my work at two symposiums and winning best presenter for this work. I was immensely proud of the work we did and how it could impact communities. Really has inspired me to do what I do now.





Kita Magangana

Where are you/what are you doing now?

I am an assistant Administrator.

Time in the Division (Years and Position)

Started working from the 1st Nov 2019.

What group were you in?

I am under the Wonkam group.



Lauren Watson

Where are you/what are you doing now?

I'm currently living in the UK. After leaving UCT I worked as a postdoc and junior research fellow at the University of Oxford, before making the move to the UK Civil Service in 2021. I am currently working as the Emerging Technologies Team Lead in the Department of Health and Social Care.

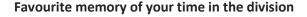
Time in the Division (Years and Position)

I was a Hum Gen student from 2006-2012 (Honours, Masters and PhD) and postdoc from 2013 to 2014 $\,$

What group were you in?

I completed my Honours and Masters under the supervision of the late Prof Bongani Mayosi in what was then the cardiac research group,

and my PhD and postdoc with Prof Jacquie Greenberg in the neuro research group (the 'A' team!) working on spinocerebellar ataxia.



HumGen has always been such a warm, welcoming place. I loved working together with the whole Division to put on our open days for high school learners, and helping host the joint SASHG/AfSHG conference in 2011 was also a highlight.

Lara Holtes



Where are you/what are you doing now?

I am in the final stage of my MSc Medical Biology at Radboud University in Nijmegen, the Netherlands. I will start my PhD in September 2023 at the Radboudumc in the blindness genetics group. My PhD project is part of a larger collaboration with University College London and we will be investigating the novel disease mechanism for autosomal dominant retinitis pigmentosa type 17 and exploring therapeutic approaches.

Time in the Division (Years and Position)

Honours student 2020

What group were you in?

Retina group

Favourite memory of your time in the division

My favourite memory of my honours year is when we finally got into the lab to work on our research projects after all the COVID lockdowns.



Where are you/what are you doing now?

I am currently working at the Research Office, UCT in Mowbray. I am an Administrative Officer in the Data Analytics and Reporting cluster.

Time in the Division (Years and Position)

I started working at Human Genetics in 2004-2013

What group were you in?

Secretary, Admin Group

Favourite memory of your time in the division

There were so many good memories but I remember the support we would receive especially with events or in fact in any work situation. Thank you Prof Raj Ramesar, Emeritus Prof Jacquie Greenberg, Assoc/Prof Karen Fieggen, Prof Collet Dandara, Ms Adielah van der Schyff and other staff and students. It was an honour to be part of the HumGen family.





• Lerato Majara

Where are you/what are you doing now?

I am a postdoctoral research fellow in the Martin Lab at the Broad Institute of Harvard and MIT.

Time in the Division (Years and Position)

Was in the division form 2015 – 2021

What group were you in?

Was part of the Psychiatric Genetics Group.

Favourite memory of your time in the division

Favourite memory was the 2015 end of year function, which was also Prof Jacquie Greenberg's farewell.



• Liezel Smith

Where are you/what are you doing now?

I joined the Division of Molecular Biology and Human Genetics, in the Department of Biomedical Sciences, Stellenbosch University. I am a senior researcher and Research Manager for the DSI-NRF Centre of Excellence for Biomedical TB Research and the SAMRC's Centre for Tuberculosis Research.

Time in the Division (Years and Position)

I was part of HumGen from March 2005- Aug / Sept 2006

What group were you in?

Psychiatric Genetics - I worked with Jonathan Savitz on Bipolar Disorder

Favourite memory of your time in the division

My favorite memories are the countless socials and tea breaks where everyone sat around the tea table. I treasure the memories and all the good friends made.



Mary Mufford

Where are you/what are you doing now?

Postdoctoral Research Fellow in the Department of Psychiatry and Mental Health

Time in the Division (Years and Position)

2016 BSc.Med.Hons, 2017-2018 MSc.Med, 2019-July 2022 PhD

What group were you in?

Psychiatric Genetics

Favourite memory of your time in the division



Lunch in the tea room, everyone comes together every day regardless of position or group and we would have the best chats and share snacks. I miss this so much! Also, on one of the Learner's days that I was involved in, we did a forensics crime scene for the kids and it was so much fun to get dressed up and come up with fun crimes and ways for the kids to solve them while learning. Also, all of the other fun extra activities were such a blast e.g. monthly birthday celebrations, spring and winter celebrations, Africa Day and Women's Day celebrations and of course, the end-of-year functions, where everyone got together and we came up with different ways to have fun and celebrate the achievements over the year. HumGen has the most social and fun working environment that I have ever experienced. It was work mixed with play, so it was so productive because you love to come to work!

Maryam Fish

Where are you/what are you doing now?

I am a postdoctoral fellow at KRISP (University of KwaZulu-Natal)

Time in the Division (Years and Position)

1 year as an Honours student

What group were you in?

X-linked intellectual disability (with Dr Rene Goliath)

Favourite memory of your time in the division

Learning new molecular genetics techniques in the laboratory



Maithili Sashindranath

Where are you/what are you doing now?

I am in Melbourne, Australia. I work at Monash University. I am leading a research group in vascular biology, and also managing a clinical project in melanoma imaging.

Time in the Division (Years and Position)

2003- BSc Hons with Dr George Rebello

What group were you in?

Retina group

Favourite memory of your time in the division

Staying up all night in the lab when my lit review was due- I drank several cups of Jacquie's brewed coffee and when she found me in the morning, she asked me to show me her hands, and they were trembling like leaves! I wasn't a coffee drinker until that night!

Margie Granger

Where are you/what are you doing now?

Enjoying retirement and like in true HumGen spirit, I enjoy travelling abroad.

Time in the Division (Years and Position)

Departmental assistant - retired in 2001.

Favourite memory of your time in the division

The 17 years at hum gen were the best years of my working career. I felt that hum gen was like a family. I still remember my first day and how Prof Beighton welcomed me warmly to the department.





Marelize Immelman (previously Swart)

Where are you/what are you doing now?

Currently I am employed as entry level medical scientist in the Human Genetics laboratory, NHLS Groote Schuur Hospital. I am responsible for the microarray testing.

Time in the Division (Years and Position)

I was in the Division between 2010 and 2016. I did MSc (Med) in Human Genetics from 2010 to 2011 and then continued with PhD in Human Genetics both degrees were supervised by Prof Dandara.

What group were you in?

Prof Dandara's research group.

Favourite memory of your time in the division

All the fun times at the MAC club and the fun train trip we took to Kalk bay. We also hosted wonderful Learner Open Days and Hons techniques courses

Maryam Bibi Rumaney

Where are you/what are you doing now?

After graduating I worked as the HIV/TB laboratory specialist at Medecins Sans Frontieres. I was based at the Cape town office, with frequent travel to the sites where I worked on projects in various global locations. Thereafter I worked for the African Cancer institute at Stellenbosch University where I was involved in research. As a mother I wanted more flexibility to be able to still practice my science and be a mother to my children. I decided to freelance and go into consultancy work. Taking my love for global health and my experience with Medecins Sans Frontieres, I was able to become a self-employed researcher offering my services to a whole host of different industries. (www.mbrumaney.co)

Time in the Division (Years and Position)

I hold a MSc in Human Genetics, from 2012 - 2015.

What group were you in?

I worked on sickle cell disease and was part of the Wonkam research group,

Favourite memory of your time in the division

My journey of science has indeed been an adventure. I am grateful for my time at HumGen, and for the people that I have met during my stay at the lab. My fondest memories are the friendships that were created and the support that we gave each other through our years as post-graduate students on this path of life-long-learning.



Maureen Akinyi

Where are you/what are you doing now?

I am currently in Houston, Texas, U.S.A, at Baylor College of Medicine and I'm halfway through my second postdoc since I wanted to get some experience/training in high throughput analyses before starting my own lab in the very near future. I mostly work on splicing related analyses, and we use both biochemical and large-scale transcriptomic methods, specifically eCLIP, to globally map the functional effects of RNA-binding proteins on different types of RNA.

Time in the Division (Years and Position)

I was in HumGen from 2008-2011 (About 3.5 years)

What group were you in?

I started off in the cancer group for my Honours and then moved to the Retina group for my Masters.

Favourite memory of your time in the division

I have soo many fond memories of being in HumGen, best place to be for a budding scientist! But if I must choose, it would be the many evenings we spent talking about anything and everything at the postgrad MAC club, lab mates, supervisors and students became friends and that made HumGen feel like home!

Megan Campbell

Where are you/what are you doing now?

I'm currently a PhD student in the department of psychiatry and a GINGER research fellow at the Broad

Time in the Division (Years and Position)

3 years (2018 to 2020); Honours and MSc student

What group were you in?

Psychiatric Genetics

Favourite memory of your time in the division

I think my favourite memory was racing the office chairs down the passage one evening while we waited for our PCR to finish (accomplices won't be named).





Megan van Zyl

Where are you/what are you doing now?

I am currently a second year registrar in Ophthalmology at Tygerberg Hospital - doing my MMed on Primary Congenital Glaucoma combining my medical and genetics backgrounds

Time in the Division (Years and Position)

Honours student 2009

Favourite memory of your time in the division

Working with kind-hearted individuals and having coffee breaks with Nakita Lang



• Michelle Parker

Where are you/what are you doing now?

I'm still in research, and studying Fetal Alcohol Spectrum Disorders at Stellenbosch University.

Time in the Division (Years and Position)

1 year. I did my Honours in the division in 2003. I was also based there in 2007 for a short while as part of Prof Mayosi's group.

What group were you in?

Cancer genetics

Favourite memory of your time in the division

Probably the fruit salad club!



Monica Araujo

Where are you/what are you doing now?

Genetic counsellor and lecturer at the Division of Human Genetics at the NHLS and Wits University. Course coordinator for the MSc (Med) in Genomic Medicine at Wits.

Time in the Division (Years and Position)

Two years (2017 and 2018) as a Genetic Counselling Masters student. I also began working on the Inherited Retinal Disorders (IRD) project under the supervision of Professor Raj Ramesar and Professor Jacquie Greenberg in September 2017. I worked for the IRD project until I left Cape Town in December 2018.



What group were you in?

Counselling and Retina.

Favourite memory of your time in the division

Throughout my internship, I faced many challenging and emotionally difficult personal and professional situations. I moved away from home to pursue my MSc, and I experienced many first during my time at UCT. With the support and guidance of the staff members and the love and understanding of my colleagues, I was able to learn and grow in a supportive environment. I admired everyone's enthusiasm for the work that they were doing and for genetics in general.

Mzwandile Mbele

Where are you/what are you doing now?

Currently I am a Senior Research Officer at Hair and Skin research laboratory (HSR), UCT, based at Groote Schuur Hospital. I am involved in wound healing and frontal fibrosing alopecia genetics research.

Time in the Division (Years and Position)

I was with Human genetics in 2005 to 2006, when I came to do honours in genetics and I left when I was starting my Masters -I was under Bongani Mayosi group which was involved in cardiovascular genetics.

What group were you in?

Cardiovascular Genetics

Favourite memory of your time in the division

I remember HumGen as a good lab with their member who treat everyone as a family. At my time at HumGen as a student when I could not wait for my supervisor or co-supervisor to assist me, all HumGen staff members could easily help, without any difficulties. All lab members were like the family to me and even now I am still in contact with them.

Nicki Tiffin

Where are you/what are you doing now?

Now I am a Professor at the South African National Bioinformatics Institute (SANBI) at the University of the Western Cape. I work in health data integration: human health genomics, epidemiology, health informatics, and ethics and governance for health data.

Time in the Division (Years and Position)

I was at Human Genetics from 2005 to 2007 as a bioinformatics postdoc.





• Priyum Koonjul

Where are you/what are you doing now?

Director Business Development, Medical Devices, National Research Council (NRC), Montreal, Canada

Time in the Division (Years and Position)

1999-2000, Post Doc with Jacquie Greenberg and Raj Ramesar

What group were you in?

Retina group

Favourite memory of your time in the division

Shopping with the girls while waiting for sequencing gel to set :)



• Reinette Weideman

Where are you/what are you doing now?

I'm living in Vredehoek, Cape Town, with my husband and our dog, Rumi. I am the South African Country Head for hear.com, a company that provides hearing care through an online-first model.

Time in the Division (Years and Position)

2011-2013, Honours and Masters student

What group were you in?

First with Jacqui looking at SCA, then with Raj looking at Bipolar Disorder pharmacogenetics.

Favourite memory of your time in the division

Memories of us all having lunch together in the team room and just connecting



René Goliath

Where are you/what are you doing now?

I left the division of human genetics in 2010 when our family relocated to Johannesburg. Soon after I embarked on a Masters degree in Genetic Counselling through a "first of its kind" joint collaboration between the University of Pretoria and UCT. I spent a highly fulfilling time practising as a genetic counsellor at Steve Biko Academic Hospital, University of Pretoria, School of Medicine, Faculty of Health Sciences - after which time I relocated back to Cape Town from where I am now running my own genetic counseling private practice at www.drgoliathgenetics.com

Time in the Division (Years and Position)

1990 to 2010 - Chief Medical Technical Officer, Senior Medical Scientist, Head of the Molecular Genetic Diagnostics Laboratory (NHLS), Principal investigator of research projects involving

Duchenne's muscular dystrophy and Intellectual disabilities, Course convenor for the BSc honours program (UCT)



Retinal, Duchenne Muscular Dystrophy, Molecular Diagnostics

Favourite memory of your time in the division

It has to be the time I was pursuing a Masters degree in Molecular Genetics. I discovered a novel gene locus for Retinitis Pigmentosa, a retinal disorder which causes blindness. The discovery made international headline news and enjoyed media coverage in the UCT MONDAY PAPER, Sunday Times, and the Groote Schuur News Publication. Professor Greenberg and I appeared on the Good Morning South Africa SABC Television program for an interview. The defining moment for our achievement was when the finding was accepted for publication in the International Human Molecular Genetics peer-reviewed journal. The discovery prompted the then Head of Department, Prof Beighton to encourage me to upgrade my studies to a PhD degree - which I duly completed as a result.



Rizqa Sulaiman-Baradien

Where are you/what are you doing now?

I am a Medical Geneticist. I am currently an Attending (Consultant) Medical Geneticist for Shared Health Manitoba and the University of Manitoba in Winnipeg, Canada.

Time in the Division (Years and Position)

I did my registrar training with Professor Ambroise Wonkam and Professor Karen Fieggen from October 2016-October 2020

Favourite memory of your time in the division

Some of my favorite memories were the social gatherings in the clinical team. We had many memorable meals, tea parties and celebrations and Prof Fieggen's home.



Sameeh Karstens

Where are you/what are you doing now?

I am currently at Groote Schuur hospital – NHLS lab where I am pursuing the HPCSA Internship to be become a qualified Medical Biological Scientist.

Time in the Division (Years and Position)

May 2022 – July 2023; Research Assistant/Lab Technician

What group were you in?

Ramesar Lab staff member

Favourite memory of your time in the division

I don't exactly have a favourite memory, however one aspect of my time in the division that I truly enjoyed was the very welcoming and friendly staff members. Coming out of University and this research lab being my first full-time job, seemed quite intimidating – but with the help of the staff and my manager Mr. Faghri February who guided me, I was able to find my feet and eventually contribute to the work that was being done there. Along with the great environment the Ramesar lab brought, the opportunity to work closely with colleagues from diverse backgrounds and areas of expertise was incredibly rewarding. This sense of teamwork and mutual support was undoubtedly a highlight of my experience.

oh yes, Gatsby day was fun too!



• Sanna Simpson (Sanne Colijn while at humgen).

Where are you/what are you doing now?

I am currently back in the Netherlands, where I am from. I am with my husband (who is from South Africa) and we have a daughter together and another little one on the way. I work at a bank as internal accounts manager.

Time in the Division (Years and Position)

I was with humgen from 2013 till 2016, for my bachelor and my masters degree. I worked on bipolar and suicide in the bipolar and forensic pathology group. The forensic pathology was a new collaboration which I started with Prof. Ramesar and Dr. Maistry.



What group were you in?

Pathology/ Psychiatric Genetics

Favourite memory of your time in the division

Being from a different country I have so many good memories from my time in SA. It's very hard to pick a favorite. When I look back at my time in humgen I am just super happy that everyone received me as a new family member at the department. Everyone is so kind and helpful, and I am just very grateful for my time at UCT. I made lots of friends that I still speak to even though I live on the other side of the world.

Sarah Owens

Where are you/what are you doing now?

I am currently living in Cape Town with my husband and two children. I am a global worker with Urban Tribes and on staff at Peoples Church in Woodstock.

Time in the Division (Years and Position)

I was a Fogarty International Clinical Research Fellow in Prof Ramesar's cancer genetics lab from 2008-2009, and 2010-2011.

Favourite memory of your time in the division

Some of my favorite memories of my time in the division are: a trip to the Northern Cape with Ursula to connect with families participating in our cancer genetics project, mentoring younger students, and presenting our research at the AfSHG/SASHG conference in 2011.



Shrinav Dawlat

Where are you/what are you doing now?

NHLS Groote Schuur Hospital (Inherited Metabolic Disease)

Time in the Division (Years and Position)

9 months as a Research Assistant

What group were you in?

Ramesar Lab Staff (helping out Akshay with lab manager duties in Ramesar Lab)

Favourite memory of your time in the division

Working with Akshay during the lockdown, and getting the lab in shape.



• Sinead Cameron-Mackintosh

Where are you/what are you doing now?

I started and run a private genetic counselling practice called Simply Genetics in Kwa-Zulu Natal.

Time in the Division (Years and Position)

I was in the Division from 2020 to 2023

What group were you in?

I was in the Clinical Genetics group as a genetic counselling student then intern

Favourite memory of your time in the division

A favorite memory was Rare Disease day in 2021, the group went for a fun run/walk on the sea point promenade.



• Stephanie Benjamin

Where are you/what are you doing now?

I'm a Senior Stability Scientist at Roche Diagnostics Cape Town, previously KAPA Biosystems (Responsible for testing the shelf life of manufactured products)

Time in the Division (Years and Position)

2015 -2016 NRF Intern on Rare disease projects and performing DNA extractions and filing of samples for Retina.

2016- BSc Honours (Rare diseases);

2017 - August 2017 Research Assistant in Retina group

What group were you in?

Various groups; mainly Retina and Rare Diseases.

Favourite memory of your time in the division

Lunch times in the tea-room, I remember being new to the Division but also Cape Town as a whole which is known for it's close cliques and I just passively sat in the conversation and laughed when applicable, and eventually was accepted into the social aspect of the group Iol. This eventually became after work Friday drinks at the MAC club too.



Where are you/what are you doing now?

Currently Senior Medical Scientist / Lecturer in Chemical Pathology (joint UCT/NHLS), responsible for the Inherited Metabolic Disorders (IMD) genetic diagnostic referral laboratory

Time in the Division (Years and Position)

1 year (2004 – honours)

What group were you in?

Retina group

Favourite memory of your time in the division

Tea times 😂





Tichaona Savanhu

Where are you/what are you doing now?

I am based in Johannesburg and I'm starting a new job next month as the Regional Performance Director for Sub-Saharan Africa for Ad Dynamo (a digital media sales business)

Time in the Division (Years and Position)

Honours Student in 2004

What group were you in?

Cancer - HNPCC

Favourite memory of your time in the division

I learnt so much in a very short space of time. I think my favourite memory was how supportive my supervisor Alvera Vorster was day to day.

Timothy Spracklen

Where are you/what are you doing now?

Postdoctoral research fellow at the Children's Heart Disease Research Unit / Cape Heart Institute, University of Cape Town

Time in the Division (Years and Position)

2013-2016; BSc(Med)(Hons) and MSc student

What group were you in?

Ramesar group - Cisplatin project

Favourite memory of your time in the division

I will always cherish the fun socials we had and the end-of-year wrap up presentations. I have fond memories of the Care HumGen initiative, I think it was a great idea!





Zandre Bruwer

Where are you/what are you doing now?

In South-Africa, working as a genetic counsellor on NeuroDev study at Red Cross Children's hospital, and Part-time genetic counsellor at Pathcare

Time in the Division (Years and Position)

Years: 2007 until 2011

Position: MSc Genetic counselling student, PhD student, Genetic

counsellor

What group were you in?

Counselling, Cancer

Favourite memory of your time in the division

Everyone was made to feel special on a birthday or special occasion - cake/treats were always set out to celebrate every occasion in the division.



Where are you/what are you doing now?

I joined the Division of Clinical Pharmacology in 2019. For my Fogarty HATTP-funded PhD thesis (supervised by A/Prof P Sinxadi (UCT), Prof G Maartens (UCT) and Prof D Haas (Vanderbilt University), I conducted a series of genome-wide association studies in a subset of participants enrolled in the ADVANCE clinical trial (NCT03122262), investigating how genetic differences influence blood concentrations of modern antiretroviral drugs, dolutegravir and tenofovir, and how they affect weight gain associated with these drugs. Currently a pursuing a 2-year postdoctoral fellowship at the University of Pennsylvania. Investigating the association of polygenic risk scores and drug-induced liver injury in the AIDS Clinical Trials Group cohort.



Time in the Division (Years and Position)

I was in the Division from 2015-2018 in the PGx group doing a Masters degree. I graduated with an MSc (Med) Human Genetics from UCT in 2018

Favourite memory of your time in the division

My favourite memory was taking part in the Amazing Race games and our year end functions.

