





SASBi Newsletter

Bioinformatics @ WITS:

The Sydney Brenner Institute for Molecular Bioscience



Staff and Students @ the SBIMB

Prof. Michèle Ramsay: I am currently the Director of the Sydney Brenner Institute for Molecular Bioscience (SBIMB); South African Research Chair in Genomics and Bioinformatics of African Populations and Professor in the Division of Human Genetics, University of the Witwatersrand, Johannesburg. I hold a PhD in Human Molecular Genetics from Wits. My research interests include African population genetic and epigenetic diversity and their role in diseases exacerbated by adverse

lifestyle choices, including obesity and cardiometabolic diseases. I am PI of an NIH funded Collaborative Centre under the H3Africa Consortium for "Genomic and environmental risk factors for cardiometabolic diseases in Africans", past president of the African Society of Human Genetics, president of the International Federation of Societies of Human Genetics and I have recently been appointed as a member of the International Commission on the Clinical Use of Human Germline Genome Editing (convened by the UK Royal Academy of Science and U.S. National Academy of Medicine), a role that I am enjoying very much. Over the years I have supervised and mentored young scientists at Wits and across Africa and continue to be excited about developing genetic and genomic research and genetic services in African settings. What is the SBIMB? The Sydney Brenner Institute for Molecular Bioscience (SBIMB) named after the late Nobel laureate and Wits University graduate, Sydney Brenner, was launched in 2014. Our vision is to create a world-class research environment where top scientific and clinical researchers can conduct innovative and relevant biomedical and genomic research to address some of Africa's greatest health challenges and to develop the next generation of African scientists. The key areas of our research include genomics, population genetics, bioinformatics, biomedical informatics, molecular and cellular biology, computational and structural biology and epidemiology. If you could give advice to students who are new in the field, what would you say? Read a lot of science, read widely, talk to your peers, your mentors, family and friends about genomics and bioinformatics. Work with potential supervisors to develop a research project that interests you and is aligned with their research interests and programs. Although it is not always obvious to students, being a student is a magical time where you can explore and grow without the responsibilities that come with full-time employment. Hobbies

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Computational Resources @ WITS (ZA-WITS-CORE)

The Wits Core Cluster is a shared compute cluster between different academic units at Wits. The lead partners have been Electrical & Information Engineering, Physics and Wits Bioinformatics, with support from Computer Science and the Mathematical Sciences. The cluster is kindly hosted by the Wits ICT, and managed by the academic groups who own the cluster. The cluster management software allows us to share equipment so that we can minimise wasted CPU cycles, but also ensure the groups can get dedicated use of the resources that they put in at peak times.

- Nodes: 45 nodes with more than 1000 hyper-threaded cores.
- · Storage: More than 1PT.
- OS: CentOS 7.5 & Ubuntu 18.04.
- Software: gcc (GNU compiler collection), gdb, emacs, vi, MPICH/Open MPI, PYTHON (version 2.7 & 3.4), PERL and JAVA.
- Bioinformatics software: See list of installed softwares here.
- Databases: EMBOSS (genbank, swissprot, trembl).

Recent Activities @ SBIMB

- H3A/GSK ADME Collaboration Meeting
- 8-10 September 2019
- Precision Medicine in Africa 2-4 September 2019
- H3Africa CVD Working Group -In-Person Meeting 19-23 August 2019
- AWI-Gen-XHALE Workshop 8-12 July 2019







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and activities in your free time? I enjoy spending time with my family and close friends and travelling to interesting places – these can be just around the corner or the corners of the globe. Reading fiction is a great way to relax and recently I am harvesting vegetables, herbs and salads from our vegetable patch (it is spring after all!).

Researchers @ SBIMB

Institute for Molecular Bioscience (SBIMB) at the University of the Witwatersrand (Wits) in Johannesburg, South Africa. I am also a member of H3ABioNet, a Pan-African Informatics Network. As part of the network I am involved in the development and running of a number of bioinformatics teaching and training events, as well as the development of infrastructure and workflows to assist African bioinformaticists with analysing their data. On the research front, I am working on a number of projects aimed at using genetic variation in whole genome sequencing data to understand and Model the historical movements and interaction of populations in Africa. Fill us in on your career up to this point. I must admit that my career path has not been the most conventional in terms of an academic setting. I was always interested in the field of genetics and completed an undergraduate degree majoring in genetics and microbiology followed by an honours in human genetics at Wits. During my honours year, I realised that I did not really enjoy wet lab work and was lucky to be exposed to the field of bioinformatics through an introductory course we attended as part of the honours curriculum. I finished my honours degree and pursued a bioinformatics focused masters degree. After learning an entirely new set of skills and just about to finish my masters, I was offered a job as a junior lecturer at Wits Bioinformatics. Although a daunting task, I took the job in 2009. In 2013 I had the opportunity to join a National Institutes of Health (NIH, USA) funded project. I then took up the

pursued a bioinformatics focused masters degree. After learning an entirely new set of skills and just about to finish my masters, I was offered a job as a junior lecturer at Wits Bioinformatics. Although a daunting task, I took the job in 2009. In 2013 I had the opportunity to join a National Institutes of Health (NIH, USA) funded project. I then took up the position at Wits Bioinformatics which joined the SBIMB at Wits. Through the H3Africa Bioinformatics Network, I have been involved in developing and presenting several training events, including being involved in the development and implementation of the novel blended learning module used to teach introductory and intermediate bioinformatics courses across Africa as well as being involved in a number of research projects. If you could give advice to students who are new in the field, what would you say? The field of bioinformatics is very diverse and can be scary to enter and establish yourself in. The trick is to equip yourself with the necessary basic skills and then find an area of interest that you would like to pursue and focus your attention mastering the skills required to excel in that area. Bioinformatics is a rapidly developing field, so you need to be open to continuously improving your skills. If you were an animal - what would you be and why? Ever heard of a tardigrade or water bear? As the name suggests, they are micro-animals that live in water around moss and resemble bears under the microscope. They are thought to have survived all major extinction events and can survive in space! When did you first realise you love science/computers? I was fortunate enough to have access to a computer from a young age when I was still in primary school. Our first computer was a second hand XT with a whopping 128KB of RAM and a massive 10MB hard drive, running the DOS operating system on a screen with orange font and a command line interface. My brother and I broke and fixed this computer more times

Dr. Houcemeddine Othman: I am currently holding a postdoctoral position at the Sydney Brenner Institute of Molecular Bioscience for a new collaborative project, which aims to study the involvement of specific genes in the drug-response of individuals from African Populations. *Fill us in on your career up to this point.* I did my undergraduate studies in medical biotechnology, then I specialised in bioinformatics and computational biology. Particularly, I was interested in studying the effect of anti-cancer peptides from snakes and scorpion venom using computer simulation and molecular modelling. My work requires multidisciplinary abilities in biology and computer skills. *If you could give advice to students who are new in the field, what would*

you say? Always be curious about science; always free some time to learn new things; Science is

than I can remember and I guess this is where my interest in computers started.

collaborative, you will never make it alone. Some times research is exhaustive, you have to keep yourself mentally fit by having an active social life. If you could preach about something to other scientists, what would it be? Errors spread exponentially. If you make them early in your work, they will come to hunt you at the end. So there is no substitute solution besides doing good science. If you were an animal – what would you be and why? A crab, I will get to swim and sunbathe all day. If you could change one thing about yourself, what would it be? Being more focused and learn to avoid distraction in my work. What do most people not know about you? I am terrible in remembering things. What do you think you are best known for around the lab? I Am the only structural bioinformaticist in the Lab. Do you have a science joke to share? In God we Trust, all others bring data.







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Dr. Dhriti Sengupta: I am a bioinformatician and population geneticist, and my research interests include studying complex population structure, inferring demographic history as well as identifying selection signals spread across the genome. Presently, I am leading the AWI-Gen population structure study. The AWI-Gen South African dataset includes >5000 participants representing 8 main ethnolinguistic groups: Tsonga, Pedi, Zulu, Sotho, Tswana, Xhosa, Swazi and Venda; recruited from three South African provinces aimed at developing a nuanced understanding of population structure and demographic histories of South Eastern Bantu speakers. In addition, I am also involved in other projects such as the AWI-Gen GWAS and the H3A WGS study. Fill us in on your career up to this point: I have done my bachelors in zoology, masters in bioinformatics

and received my PhD in Bioinformatics from University of Calcutta, India in 2013. Since 2014, I have been working as a postdoctoral fellow at the Sydney Brenner Institute for Molecular Science, University of the Witwatersrand. If you could give advice to students who are new in the field, what would you say? The all-time clichés- Patience and hard work are must. And you will always love your work if you find the joy in learning new things:) Finally, please add comments to your programs otherwise you would spend almost similar time trying to remember why you wrote a particular line (I learnt it the hard way!!). What are your hobbies/activities you do in your free time? Planning to start a healthy life style... and finding excuses for not doing it... everyday! When did you first realise you love science/computers? Now if I look back, I guess I fell in love with computers while playing games like "Age of Empires". For science, I think it was a gradual slow and steady affair. Do you have a favourite computer language? Perl (I am almost a dinosaur now!!). Do you have a science joke to share? If I throw my manuscript into the forest, have I submitted it to Nature?

Students @ SBIMB

Ms. Natalie Smyth (PhD): I am currently a research assistant at the SBIMB BioBank. My project is aimed at understanding the genetic basis of extreme levels of high and low LDL-Cholesterol in African populations. Fill us in on your career up to this point. I started my studies at the University of Stellenbosch, where I did my BSc in Molecular Biology and Biotechnology. I then went on to do my honours in Plant genetics focusing on viroids in grapevines. I then completed my MSc in Plant Pathology, looking at the spatial and temporal distribution of aster yellows phytoplasma in grapevines in South Africa. I made the move up to Johannesburg in 2015 where I starting working at the SBIMB BioBank as a research assistant for the AWI-Gen project. I decided working in Human Genetics was more for me and I have subsequently started my PhD looking at the genetics behind high and low LDL-Cholesterol in African populations. If you could give advice to students who are new in the field, what would you say? Make sure the research projects you do really interest you and do not be afraid to ask questions and for help. What are your hobbies/activities you do in your free time? I do yoga and teach yoga classes in my free time. If you were an animal - what would you be and why? Elephant. They are loyal, smart, very protective of their loved ones and needless to say, other animals are pretty scared of them. When did you first realise you love science/computers? My parents bought me a mini microscope for my 11th birthday and I spent many many hours looking at many different things under the microscope and trying to describe what I was looking at. That was before I knew anything about genetics, or even cells. I fell in love with genetics when I first learnt what DNA was in grade 9 biology. I was very drawn to trying to understand the concept. Not sure I can explain why though:).

Mr. David Twesigomwe (MSc): My current project is about identifying variation in the CYP2D6 gene in African populations. This project is so interesting because we have the opportunity to mine new African datasets from various populations to provide insight into the pharmacogenomic variation landscape in Africa. Fill us in on your career up to this point. I did my BSc in Biomedical Sciences at Makerere University in Uganda from 2014 to 2017. In my second year, I got to know about Bioinformatics through a mentorship/consultation talk. I was so excited about the interdisciplinary nature of the field and started looking for training opportunities. The H3ABioNet Introduction to Bioinformatics Course (2017) was really helpful in this regard. I went on to do a year-long internship in Bioinformatics and Computational Biology at the Uganda Virus Research Institute, where I got hands-on training in a cutting-edge research environment. I moved to Wits University in January 2019 to start my MSc studies in Human Genetics. I am currently based at the Sydney Brenner Institute for Molecular Bioscience (SBIMB). If you could give advice to students who are new in the field, what would you say? Bioinformatics is a really exciting field, which brings the best out of you in terms of enthusiasm, teamwork, and

creativity among other things. Strive to benefit from the great wealth of information available today courtesy of the







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internet and also to have the right group of people around you to form a nice team. If you were an animal – what would you be and why? I wouldn't want to be an animal:). Do you have a science joke to share? That moment when your supervisor finds you watching a YouTube video... and you ask yourself why they didn't come two minutes earlier when your coding screen was on:).

Other People @ SBIMB

PIs, Staff and Administration

Name	Position
Prof. Scott Hazelhurst	Director of Bioinformatics
Prof. Chris Mathew	Distinguished Professor
Prof. Collen Masimirembwa	Visiting Professor
Dr. Ananyo Choudhury	Senior Scientist
Mr. Yusuf Ismail	Operations Manager
Mrs. Jocelyne Gayenga	Manager: Strategic Development
Ms. Mmatshepo Taunyane	Research Administrator
Ms. Cassandra Soo	BioBank Manager
Ms. Busisiwe Mthembu	BioBank Lab Assistant
Mr. Freedom Mukomana	Data Administrator
Dr. Tinashe Chikowore	Wellcome Trust Training Fellow
Dr. Ovokeraye Oduaran	Postdoctoral Fellow
Dr. Jean-Tristan Brandenburg	Postdoctoral Fellow
Dr. Abram Kamiza	Postdoctoral Fellow
Dr. Phelelani Mpangase	Postdoctoral Fellow

Active Research Projects

Our research teams are currently investigating some of the biggest and most compelling questions in modern science, with a focus on sub-Saharan African populations. These include:

- AWI-GEN
- H3ABioNet (H3Africa)
- · Cancer in African Populations
- GSK ADME
- · Albinism in Africa
- Autoimmune diseases
- · Microbiome Studies
- · Ophthalmological genetics
- · Cancer Treatment Pharmacogenetics

PhD Students

Name	Project Title			
Mr. Wenlong Carl Chen	The genetic aetiology of oesophageal cancer in the South African black populations.			
Mr. Evans Mathebula	Rheumatoid arthritis in black South Africans: Genomic susceptibility and Pharmacogenomic investigations.			
Mrs. Jenny Mary Mathew	Identification of expression quantitative trait loci (eQTLs) and their functional impact on ADME genes.			
Ms. Cassandra Soo	Genetic and environmental factors associated with cognitive decline and the dementia spectrum in an ageing South African population.			
Ms. Mahtaab Hayat	Investigating the genetic aetiology of breast cancer in black South African populations.			
Mr. Jorge da Rocha	Characterisation of key ADME gene variants in African populations through functional annotation and modelling.			
Ms. Surina Singh	Genetic associations with blood pressure and hypertension in African populations.			
Ms. Melanie Govender	Genetic associations with kidney disease in African populations.			
Ms. Michaella Hulley	Differential gene expression in exfoliation syndrome and exfoliation glaucoma in the conjunctiva of Black South Africans.			
Mr. Godfred Agongo	The effect of genetic variants, anthropometry and the environment on lipid profiles in adults in northern Ghana.			
Mr. Romuald Boua	Genetic variants and oxidative stress: Impact on the risk for atherosclerosis in the AWI-Gen study.			

MSc Students

Name	Project Title
Mr. Brian Kariithi	Evaluation of iPSC-derived hepatocyte model to predict functional effects of the
	CYP2D6*17 genetic variant in activation of tamoxifen to endoxifen.
Ms. Laura Cottino	A systematic assessment of the Copy Number Variation (CNV) landscape in ADME genes
	in Sub-Saharan African populations.
Mr. Runesu Bakasa	Is nanopore sequencing technology useful for confirming Huntington's disease diagnosis in African individuals?
Ms. Micaela Eisenberg	Investigation of the OCA2 gene control regions and their possible role in normal pigment variation.
Ms. Sebentile Mthimkulu	Telomere length association with Type 2 Diabetes in a sub-Saharan African Population.

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SASBi Update Letter from the President

Greetings!

It has been a year since the Council and I were elected at the SASBi-SAGS 2018 conference at Golden Gate National Park. During the past year, the Council has been very active, holding regular monthly teleconferences and trying to promote the Society. It has been a pleasure to see all the activity that is making our society more vibrant.

Two major upcoming events are workshops by Dr. Leonardo Mariño-Ramirez of the National Center for Biotechnology Information, title "Bioinformatics for the Biologist". The two workshops will be held on November 13 to 15, at Stellenbosch University, Stellenbosch, and on November 20 to 22, at the Turfloop campus, University of Limpopo, Mankweng (Polokwane). These workshops are made possible by the Fulbright Specialist Program, through the Embassy of the United States. Dr. Hugh Patterton (Stellenbosch) and I applied to the program and the Program was kind enough to fund the trip. This is very exciting and the response to the Stellenbosch workshop has been overwhelming with more than twice the number of applications than seats. Applications for the University of Limpopo workshop will be opening soon.

Another major achievement has been the bi-monthly Newsletter. We have consistently been publishing/circulating a newsletter on schedule. Again, this is very exciting since the Newsletter was designed to include a highlight of the bioinformatics at one of the South African universities each time. We want to encourage institutions to volunteer to produce a Newsletter and present their activities. Each Newsletter also features a section highlighting a few bioinformatics students at the institution that is producing that issue. We hope that this will help networking at the next SASBI-SAGS conference.

The Student Council has also been active and has organised several activities. Among them were participation in SciFest Africa 2019 in March and a joint symposium with the SA Society of Human Genetics at the Biennial Congress of the South African Society for Human Genetics in August 2019. Mahtaab, the president of the Student Council is currently on the organising committee for the ISCB Africa Student Council Symposium. The symposium will take place in November this year, in Kumasi, Ghana. Well done to the Student Council.

It is also a pleasure to note that our SASBi website (http://sasbi.weebly.com/) has been updated thanks to the hard work of Alisa Postma. We are considering moving to another platform that will permit easier updates and are awaiting some proposals by Werner Smidt. We will send out announcements as soon as we have a functional new site. Meanwhile, please visit the old site where you can also access archived copies of the newsletters.

Speaking of the SASBi-SAGS 2020 Conference, it will be held in the Stellenbosch region on 12 to 15 September 2020 (venue availability is driving the choice of date). Please diarize this already. The program will feature more integration between SASBi and SAGS topics, without parallel sessions. We wish to encourage more cross-talk and networking between the Societies and will try this approach. The SASBi and SAGS Councils have appointed a joint conference committee (table below), which is actively planning and preparing. We will be approaching potential donors for funding. More updates to follow.

SASBi-SAGS joint conference committee

	SAGS	SASBi
Co-Chairs	Clint Rhode	Gerard Tromp
Scientific Programme	Aletta van der Merwe	Ruben Cloete
Social Programme	Beatrix Coetzee (tentative)	Mahtaab Hayat
Sponsorships	Michael Wolf	Cedric Werely
Student Affairs	Kelvin Hull	Ncite da Camara

The paucity of venue availability is something for the Councils of both societies to consider for the 2022 joint meeting. It would behave the societies to establish a conference committee during, or soon after the conference to start planning (in the US some societies plan their annual meetings 3 to 4 years ahead).

Lastly, I will finish with an appeal. We are a small society, working hard to promote bioinformatics. Please encourage your colleagues in bioinformatics to join SASBi and help promote all things bioinformatic as well as be a benefit to bioinformaticians. The more members we have, the more volunteers will be available to help. We hope to institute travelling workshops and courses, but we need more bodies to make this possible.

Wishing everyone a fine end to the year!

Prof. Gerard Tromp







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Upcoming Events

Event: Symposium - Genetics of movement disorders in Africa

Date: 2 November 2019

Venue: STIAS, 10 Marais Road, Stellenbosch Central, Stellenbosch, South Africa

Info: Click here for more information.

Event: Conference - ISCB-Africa ASBCB 2019

Date: 11 - 15 November 2019 Venue: Kumasi, Ghana

Info: Click here for more information.

Event: Symposium - 3rd Student Council Symposium (SCS) Africa

Date: 11 November 2019 Venue: Kumasi, Ghana

Info: Click here for more information.

Event: Conference - Evolutionary Systems Biology

Date: 12 - 14 February 2020 (Bursary deadline: 19 Nov 2019)

Venue: Wellcome Genome Campus, UK *Info*: Click here for more information.

Event: Course - Mathematical Models for Infectious Disease Dynamics

Date: 24 February - 06 March 2020 (Bursary deadline: 14 November 2019)

Venue: Wellcome Genome Campus, UK *Info*: Click here for more information.

Event: Conference - Single Cell Biology

Date: 11 - 13 March 2020 (Bursary deadline: 2 January 2020)

Venue: Wellcome Genome Campus, UK *Info*: Click here for more information.

Event: Conference - Genomics of Brain Disorders

Date: 18 - 20 March 2020 (Bursary deadline: 7 January 2020)

Venue: Wellcome Genome Campus, UK Info: Click here for more information

Event: Conference - Genomics of Rare Diseases

Date: 25 - 27 March 2020 (Bursary deadline: 14 January 2020)

Venue: Wellcome Genome Campus, UK *Info*: Click here for more information.

Event: Conference - Proteomics in Cell Biology and Disease Mechanisms Date: 30 March - 01 April 2020 (Bursary deadline: 21 January 2020)

Venue: Wellcome Genome Campus, UK Info: Click here for more information.

Event: Conference - SASBi/SAGS 2020
Date: September/October 2020

Venue: Cape Town

Info: Click here for more information.

Event: Conference - International Congress on Human Genetics (ICHG)

Date: 01 - 13 March 2021 Venue: Cape Town

Info: Click here for more information.

SASBi-SC Update

Hi SASBi newsletter readers!

After a successful joint student symposium with the SASHG YRF in Cape Town, the SASBi-SC has taken a bit of a breather. Mahtaab, our chairperson, is on the organising committee for the ISCB Africa Student Council Symposium. This is set to take place on the 11th of November in Kumasi. Ghana. The ISCB Africa conference this year focuses on advancing research through data power: innovation, access, and opportunities relevant to Africa. The Student Symposium hopes to showcase this theme, too. We hope to get involved in a few activities in 2020. We'll report back in the next newslet-

The SASBi SC Team

Editorial Team

This issue was brought to you by...



Ms. Mahtaab Hayat PhD Candidate



Dr. Phelelani Mpangase Bioinformaticist

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