The AWI-Gen Study, 2012-2022

by Stuart Ali (AWI-Gen Project Manager and Investigator)

The AWI-Gen Study aims to understand genetic and environmental risk factors for obesity, diabetes and hypertension in Africans (1). AWI-Gen is the Africa Wits-INDEPTH Partnership for Genomic Research, which is a 10-year study (2012 to 2022) by the University of the Witwatersrand (Wits) and the International Network for the Demographic Evaluation of Populations and Their Health (INDEPTH). The PI is Michèle Ramsay (Wits) and the Co-PI (2012 to 2017) was Osmon Sankoh (INDEPTH). The AWI-Gen study operates at six research centers in four

Map of Africa showing the study areas where the AWI-Gen study operates

African countries: Burkina Faso, Ghana, Kenya, and South Africa. At the end of recruitment for Phase 1 in August 2016, AWI-Gen had recruited 10,700 volunteer participants between the ages of 40 to 60 years.

The AWI-Gen study explores how genes, the environment, and lifestyle affect health – in particular obesity and cardiovascular disease. In doing so we hope to better understand the determinants of health and the impact of different lifestyles and behaviours.
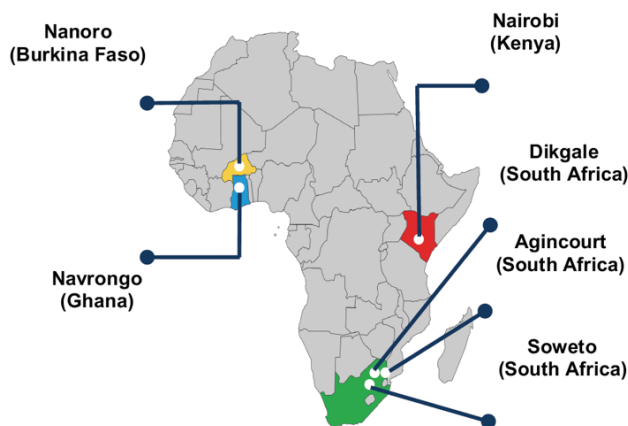
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Once we have more knowledge on the genetic contributions to obesity, diabetes and hypertension, we hope to be able to calculate genetic risk scores to identify individuals at high risk. This could lead to changes in public health policies and new approaches to prevention and treatment.

In the first phase of the study (2012-2017) (2), AWI-Gen made some important discoveries. In East and South Africa, a large proportion of individuals suffer from hypertension, and whilst many of them are taking medication, their hypertension is not properly controlled (3). We have also discovered that obesity is very common in East and South Africa, and that women are at higher risk of obesity (4). In contrast, people in West Africa tend to have lower levels of hypertension (3) and lower levels of obesity (4).

In the second phase (2017-2022) we will revisit the participants recruited for the first stage in order to examine how their health, behaviours and exposures have changed over time. The study will also capture new data on exposure to pollutants, respiratory health, sleep, frailty and life stressors.

By understanding what is responsible for differences in obesity and hypertension in different African regions, we hope to uncover new insights that could help improve the health of Africans.

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ACEGID Obtains International Accreditation for MSc and PhD in Molecular Biology and Genomics by AQAS, Germany

by Onikepe Folarin

The African Centre of Excellence for Genomics of Infectious Diseases (ACEGID) was established with a mandate to build capacity in the field of genomics as it relates to infectious diseases. This gave birth to the development of curricula for master's and doctorate degrees in Molecular Biology and Genomics (MBG) at Redeemer's University, Nigeria. The curricula were designed to comply with world class standards, hence the need for international accreditation.

International accreditation for MSc. and PhD in MBG is important to ACEGID for true demonstration of excellence and to provide assurance that the academic quality meets or exceeds international standards. In addition, the accreditation will provide better visibility and international recognition to the students and university at large. In order to achieve this, the Agency for Quality Assurance Through Accreditation of Study Programs (AQAS e.V) in Germany was consulted. AQAS e.V is a member of the European Quality Assurance Register for Higher Education (EQAR) and European Association for Quality Assurance in Higher Education (ENQA). AQAS e.V is an accrediting

agency that accredits programs and higher education institutions in Germany, Europe and worldwide.

The accreditation process was officially initialised by the Accreditation Commission (AC) on 14/15 May 2018 and followed three major stages; contract and agreement, self assessment and an expert site visit. The complete process spanned a period of 9 months.

AQAS experts interacting with graduate students in the Molecular Biology and Genomics programmes.

The evaluation by the experts was quite positive, especially regarding facilities and resources - both human and capital - available to run postgraduate degrees in MBG in the university. In addition, the experts were impressed by Professor Christian Happi's leadership of ACEGID, as well as the team of lecturers who shared in the vision of building capacity to a critical mass of young African scientists in the field of genomics.

On December 10, 2018, the AC announced the award of international accreditation for the programmes of MSc. and PhD. in Molecular Biology and Genomics at Redeemer's University. This award of international accreditation demonstrates international standards, improvement of academic quality and public accountability of the programmes. The international accreditation of these programmes at Redeemer's University by AQAS further confirms the university as a centre of excellence, and serves to improve the visibility of the university, thereby attracting greater numbers of students, especially international students, for admission to these programmes.

New Frontiers in Data-Driven Science at the Heart of Africa

by Daudi Jjingo

The African Centers of Excellence in Bioinformatics and Data Intensive Sciences (ACEDIS) was launched on March 21st, 2019 at Makerere University in Uganda. This effort involved key personnel from BReCA, the Ugandan H3Africa Bioinformatics training programme, who worked tirelessly with multiple partners towards the establishment of ACEDIS.

Housed at the Infectious Disease Institute (IDI) on the Makerere campus, the center is hailed as the first of its kind in East Africa. It is a centre for Computational Biology and Big Data analysis with a dedicated high-performance computing cluster, a tele-learning centre,

collaborative spaces and a virtual reality facility for the latest 3-D pedagogical approaches. The centre has also been designed to support research and development in Artificial Intelligence, Machine Learning, and the delivery of Masters, PhD and post-doctoral level training programs in bioinformatics. With data analysis capacity of this scale and sophistication, the centre will be a major platform for data-driven research and discovery through innovation and computation.

Contributions to this project have come from the US Government's National Institute of Allergy and Infectious Diseases and its Office of Cyber Infrastructure and Computational Biology (NIH/NIAID/OCICB), supported by the Foundation for the NIH (FNIH) and its private partners, who have made generous in-kind donations of equipment, software and hardware infrastructure. Uganda's Infectious Disease Institute (IDI) and Makerere University's Colleges of Health Science (CHS) and Computing and Information Science (CoCIS) have provided specialized space, as well as technical staff to facilitate the center.

Conference Seeks to Advance Women in Global Health

By Karin Zeitvogel

A woman health worker in rural India was gang-raped "to teach her a lesson" after she promoted contraception, family planning and education for girls. Women working on polio vaccination campaigns have been abducted

and killed. And one of the world's few women health ministers was asked during congressional testimony if she was married or single. The male lawmaker asking the question said the answer would indicate to him and his colleagues if the minister had ever had "a good night". Those were some of the stories heard by the 900, mainly female attendees at the second Women Leaders in Global Health (WLGH) conference held in London in November, 2018. Despite women making up around three-quarters of the global health workforce, often serving on the frontlines of a health crisis or providing unpaid care for a family member, they hold less than a quarter of global health leadership positions, noted former Fogarty trainee Dr. Soumya Swaminathan, who is now WHO Deputy Director-General for Programmes.

Change has to start at the base of the pyramid and cover every level of caregiver and recipient, conference-goers were told. "We need leaders and excellent foot soldiers," said Dr. Wafaa el-Sadr, a professor at Columbia University and member of Fogarty's advisory board. "Everyone within an organization needs to be transformed." All women need access to quality health care, and organizations need to revisit their messaging about women's health, said Dr. Joanne Liu, the international president of Médecins Sans Frontières (MSF). "If we don't improve access to health care for women, we're always going to be running behind a train that is moving faster," Liu said. Three-quarters of patients who go to MSF hospitals are women seeking care for a pregnancy-

related issue or to give birth, she added. "But we never talk about that. We talk about the war-wounded, about Ebola, never about women coming in to deliver and being safe and well cared for."

The conference, part of the Women Leaders in Global Health Initiative, was hosted by the London School of Hygiene and Tropical Medicine. The 2019 WLGH conference is set to take place in November in Rwanda. Read more

Article adapted from the Fogarty International Center newsletter, January-February 2019

Mentorship Training in LMICS Needs Increased Support

by Shana Potash

Whether helping young scientists shape their careers, conduct ethical research, or define a work-life balance, mentors are instrumental in nurturing future generations of global health researchers. But in low- and middle-income countries (LMICs), formal mentoring is not often adequately supported by institutions or included in formal training programs. To encourage LMIC organizations to strengthen mentoring and institutionalize the practice, Fogarty Scholars and Fellows program faculty and alumni produced a new publication to serve as a guide.

Mentoring in Low- and Middle-Income Countries to Advance Global Health Research , a supplement to the *American Journal of Tropical Medicine and Hygiene*, offers recommendations, case studies and an overview of toolkits to help design mentorship programs tailored to LMICs. The publication was inspired by a series of "Mentoring the Mentor" workshops hosted in LMICs by faculty of Fogarty's Global Health Program for Fellows and Scholars. It was edited by Dr. Craig Cohen, Co-director of the University of California Global Health Institute in San Francisco. More than 40 leaders in global health from around the world contributed to develop and publish the special issue.

"Great mentors are not born. Mentoring skills, like any other, must be developed," Fogarty Director Dr. Roger I. Glass and Dr. Flora Katz, director of the Center's extramural programs, note in the preface to the supplement. "It is our hope this collection of articles will provide a stimulus for increased funding to fill this critical need." Read more

Article adapted from the Fogarty International Center newsletter, January-February 2019

PUZZLE Of HIV: Predicting Drug Resistance and Understanding the Mutation Mechanism

by Olivier Sheik Amamuddy, Margaret Nabatanzi, Nigel Bishop and Özlem Tastan Bishop
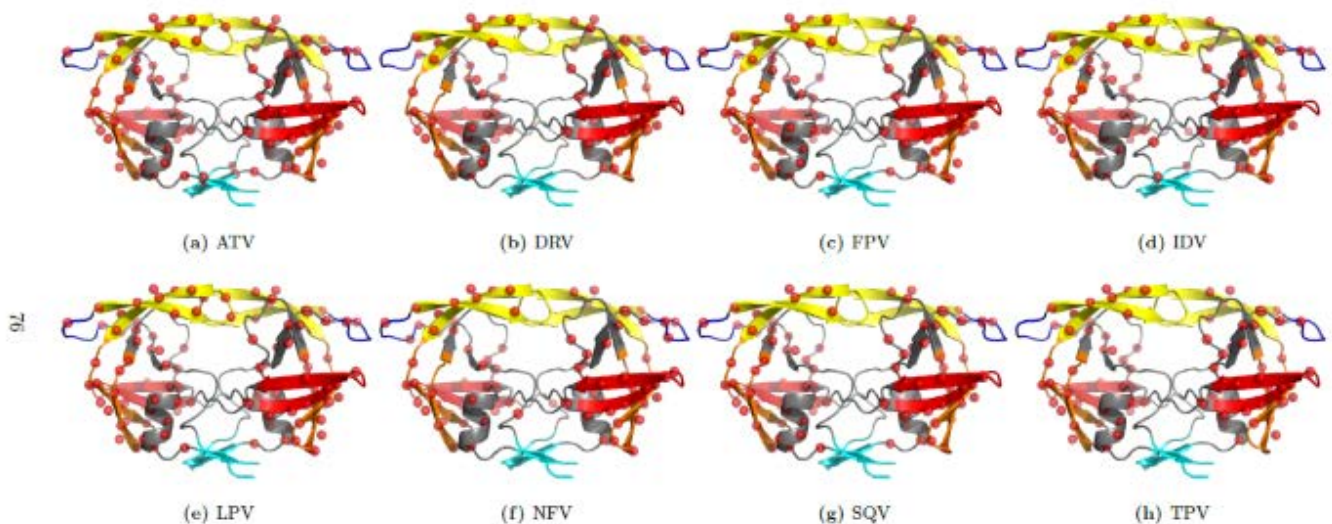
Mapping of the variation positions onto 3D structures, for each of the FDA-approved HIV protease inhibitors ATV, DRV, FPV, IDV, LPV, NFV, SQV and TPV used for the drug resistance ensembles. The colored cartoon representations depict the fulcrum, elbow, ap, cantilever *and* interface, *while the variation positions are depicted as red spheres. Note that even though single spheres are shown, some positions comprise multiple residue variations, some of which are validated drug resistance mutations (major and accessory DRMs) from the 2017 update. Variations from the susceptible ensembles are not shown, for clarity (Taken from Olivier Sheik Amamuddy's PhD thesis)*

In the last few years, Research Unit in Bioinformatics (RUBi) at Rhodes University has been tackling HIV-1 drug resistance related problems; on one hand by searching ways to improve the performance of drug resistance prediction for HIV protease and reverse transcriptase, on the other hand by developing approaches to understand resistance mechanism of HIV protease against eight existing drugs.

Antiretroviral (ARV) resistance is a great concern for many reasons including the use of sub-optimal regimens. Thus, predicting resistance to ARVs before and during any treatment is important. This problem is usually treated as one of classification, since in a clinical context it is normally sufficient to predict the effectiveness (or not) of a given ARV. However, we approached it as an artificial neural networks (ANN) regression problem, thereby making full use of all available data and so potentially improving the predictive accuracy of the model (*Sheik Amamuddy et al. 2017, BMC Bioinformatics*). The results obtained show that our model is at least comparable to that of other methods, and for most ARVs is a definite improvement.

We developed a web application, HIV-1ResPredictor (https://hiv1respredictor.rubi.ru.ac.za/), that provides access to the ANNs to predict antiretroviral drug resistance in patients infected with HIV-1 subtype B in order for them to be more widely and easily utilized. We believe that beside personalized medicine, this

application has uses in clinical practice environments as well as public health, potentially reducing cases of treatment failure and cross-resistance, and their associated morbidities, mortality, and resistance-associated costs. This work was done by Margaret Nabatanzi as part of her MSc degree.

In another effort, we developed an approach to understand the mechanism of early drug resistance against 8 HIV protease ARVs and, for the first time, showed how mutations, even when far from the active side of protease (Figure 1), are changing the structural behavior of the protein (*Sheik Amamuddy et al. 2018, Scientific Reports).*These findings have great importance in novel drug design research and development against HIV.

RUBi would like to invite H3Africa to use the webserver and to collaborate by sharing subtype C data, which will enable a focus specifically on African populations in these approaches.

ACEGID, Redeemer's University Confirmed Yellow Fever Virus in Samples of Unknown Pathogen Using the Next-Generation Sequencing Technique

By *Onikepe Folarin*

Yellow fever is an acute mosquito-borne viral haemorrhagic fever, once common in Nigeria but largely absent for decades until 2017 when the Nigerian Center for Disease Control (NCDC)

reported an outbreak [78 (2.4%) of 3295 samples confirmed positive as of 16-December 2018]. After a cluster of undiagnosed febrile patients in Edo State, Nigeria were retrospectively identified by clinicians, thirteen (13) patient samples were sent to the African Centre of Excellence for Genomics of Infectious Diseases (ACEGID), Redeemer'sUniversity, Ede, Nigeria from the Irrua Specialist Teaching Hospital (ISTH), Irrua, Edo State, Nigeria for unbiased metagenomic sequencing to determine the etiology of the fever.

At ACEGID, whole genome sequencing method and RT-qPCR was carried out within four (4) days of sample receipt and established the presence of Yellow fever virus (YFV) as the cause of the fever in nine patient samples by RT-qPCR and assembles four genomes by sequencing. Phylogenetic analysis was used to compare sequences from the 2018 outbreak to previous sequences from Nigeria and other parts of the world and the sequences from the 2018 outbreak were more related to sequences from other West African countries than to earlier Nigerian sequences showing that the YFV responsible for the 2018 outbreak in Edo State does not descend directly from the Nigerian YFV outbreaks of the past century but is instead part of the broader diversity of current YFV in West Africa. We immediately reported our findings to the Nigerian Centre for Disease Control (NCDC) who declared an outbreak of Yellow fever in Edo State the following day based partly on our findings. We also immediately shared our pre-publication data on **Virological**:

http://virological.org/t/yellow-fever-outbreak-in-nigeria-2018/274 to facilitate open science and open data sharing and to help the global scientific community combat the outbreak. A paper reporting our findings is also in preparation.

Talking to Women in Bioinformatics

By Busisiwe Mlotshwa

We interviewed Professor Zané Lombard of the Human Genetics Division at the University of Witwatersrand (Wits), Johannesburg, South Africa, to get her take on being a female scientist in the field of Bioinformatics.

Professor Zané Lombard, Associate Professor, Division of Human Genetics, University of the Witwatersrand, Johannesburg, South Africa.

Busisiwe: Tell us about yourself, your background, and how you came to land your current role.

Prof. Lombard: So, umm, I've been in the division of Human Genetics for a while now. I completed my PhD in Bioinformatics and Human Genetics here at Wits University in 2007, then went to work for the National Health Laboratory Service as a medical scientist. I returned to Wits in 2012, teaching Bioinformatics, and this position has actually been a great opportunity to return to research.

Bioinformatics wasn't really a career choice when I started studying, so I wasn't really aware of it during my undergrad. My undergraduate major was in Biochemistry, as were my Honours and master's degrees. When I joined Michele Ramsey's group at Wits, she was involved in starting up the Bioinformatics network, and offered me a PhD in Bioinformatics, which I found really scary (laughs), as I had no background in it, but also really interesting. I thought it was great opportunity to work with Michele at SANBI (South African National Bioinformatics Institute) where I received formal training. **All** (she stresses) scientists working in the field need to know some bioinformatics, especially because our datasets can be so different.

Busisiwe: Is your job what you thought it would be when you started?

Prof. Lombard: No, things are never what you imagine them to be. I'm now heading research and development, which is a more research-focussed position and what I wanted when I

came back to the division of Human Genetics. But there is lots of administrative work and teaching as well, which distract from research. Unexpectedly, managing small groups of people is harder than I thought it would be, but I consider it part of my career growth to encompass new responsibilities.

Busisiwe: What have been the opportunities in this career path, for women in Africa, as you've made your way up the ladder of success? Do you feel these opportunities have changed - become more favourable or less so - for women?

Prof. Lombard: The field is definitely changing, with a lot of female role models. Science is actually pretty female dominated in some fields, but Bioinformatics was very male dominated when I joined. There are more opportunities now than there were a few years ago, with organizations like H3ABioNet and AESA creating opportunities for all scientists but especially female scientists in Africa. Sometimes, though, you have to create those openings for yourself in order to pursue your career passion. In South Africa, there are a number of opportunities for students at postgraduate level but developing a career in Bioinformatics is still a struggle because there are so few faculty positions available.

Busisiwe: How do you see your role changing in the next 10 years?

Prof. Lombard: Well, as you become more senior, your role does change. An increased administrative role can remove you from the exciting part of analysing data and wrestling with answers, but I think it comes full circle again. Increasing seniority also provides you with access to more resources so you can build a team to help you with the admin end of things so that you can return to research and have more of a focus on research and engaging results with your team. I'm anticipating that my role will give me the opportunity to become more focussed on research and data analysis. I think whatever your responsibilities, it's important to be true to yourself; identify which things you enjoy in your career and what sparks joy for you and make sure you get to do that. Nurture your passion. Always keep an eye on the ball and don't move away too much from the things that you love doing.

Busisiwe: One of your current projects is Deciphering Developmental Disorders in Africa (DDD-Africa). Can you elaborate on the goals (and progress) of the project?

Prof. Lombard: This is a new project (excitement in her voice); we received funding just over a year and a half ago, from H3Africa (through the NIH National Institute of Mental Health) to investigate developmental disorders in African children. One of the main undiagnosed conditions in children with developmental disabilities is developmental delay; the limited number of diagnostic tests available often don't provide answers to parents and in 80% of the cases we see, the cause of the disability is not identified. Our sites are in Johannesburg, Kinshasa and the DRC and we aim to collect 500 trios, perform WES on the samples and subject the data to

bioinformatics analyses to see if we can arrive at a diagnostic answer. We also want to see if we can implement this approach as a diagnostic tool in the future for genetic testing. As far as progress is concerned, we're about one-third of the way through recruitment, which should be complete beginning next year. We aim to begin analyses of sequencing data towards the end of this year. Ultimately, we want to make sure that we improve the lives of our patients and their parents so I'm excited about setting up testing and sequencing locally and using technology to impact local lives.

Busisiwe: Many women feel intimidated at the thought of holding down a high impact career and having a family. What social obligations go along with a job in your field and how do these impact your private life?

Prof. Lombard: (long breath) It's hard; this is definitely one of the difficulties that women in science face. It feels like you have to make an impossible choice between your career and private life because you need to work very hard in order to advance your career but at the same time, you shouldn't neglect your personal life. Most often, it's women who take on the role of primary caregiver at home and they juggle this role with the demands of their career. My social responsibilities to the field include being part of various committees, giving reviews, attending conferences and so on. Travel is a big part of my position so finding a balance is challenging; having a good support system in place - family and friends - helps a lot. It's difficult to do it all but it's not

impossible. Having both a career in science and family is hard work but I encourage women everywhere not to give up but to work on finding a way to make it work for them.

Busisiwe: What's the most important leadership lesson you've learnt and how has it proven invaluable?

Prof. Lombard: I think there are a couple that come to mind. I heard this quote at a conference - The best is the enemy of good enough. Sometimes in your career, you can become paralyzed because you feel you're not "the best" you could be; the feeling paralyzes you so you can't achieve anything. With what you already have, the knowledge, opportunities and resources available to you, I feel you should move forward to the next stage of your career. One day you'll get to that "best place" in your career.

I also feel that education and training are really important in advancing oneself in life. Nelson Mandela said that 'everyone can rise above their circumstances and achieve success if they are dedicated to and passionate about what they do.' I second that sentiment; I think it's really essential to be passionate and educate yourself and be a constant learner in life.

Busisiwe: What advice would you give to students who are new in the field (and even those like me who are not so new)?
Prof. Lombard: Opportunities have been far and few in-between but are becoming more available. Keep swimming (laughs), move

forward and take up opportunities that present themselves to you. I think we always regret more the things that we **don't** do, compared to the things we do, so look for openings to develop yourself. Also, think about what you can bring back to the community where you grew up so you can give opportunities to others who desire to follow a similar career path.

Busisiwe: Scientists don't make the best managers. What's your response to this statement?

Prof. Lombard: An important point! In my experience, we're really well-trained in writing results but aren't given formalized training in management. You really feel this when you get to a point in your career when you're expected to know how to manage people and be a good leader. I think this is a real skillset that one shouldn't be expected to just 'have'. We need more management and leadership training as soft skills because people don't intuitively know how to develop these abilities. This may be an area for H3Africa to think about organizing future workshops in as an essential tool for career progressiveness.

Busisiwe: Thank you so much for taking the time to share these snippets about yourself and your career with myself and the readership of the H3A newsletter, Prof. Lombard.

Prof. Lombard: Thank you, Busi, and best of luck for the final push in your PhD!

We continued the conversation with Dr. Liberata Mwita, a young bioinformatician working with Professor Julie Makani in the Sickle Cell Program at Muhimbili University of Health and Allied Sciences in Dar es Salaam, Tanzania.

Dr. Liberata Mwita, Bioinformatician, Sickle Cell Program, Muhimbili University of Health and Allied Sciences, Dar es Salaam, Tanzania.

Busisiwe: Can you tell me a little about yourself and how you became a bioinformatician, because it's still a pretty much unexplored field here in Africa?

Dr. Mwita: Yes, yes, it is. I have a background in molecular biology and biotechnology and my master's was in biotechnology. In the beginning, I didn't have any idea about what

bioinformatics is! During my master's program, I had the opportunity to spend some months at the University of Pretoria (UP) and I had to perform some bioinformatics analyses as part of my research. So, I took a short course in bioinformatics, found it really interesting and decided to continue my education with a PhD in bioinformatics. I also realized that there are very few bioinformaticians in my country and we really need local expertise, which also weighed into my decision to pursue a career in Bioinformatics.

Busisiwe: I assume that at that time next-generation sequencing hadn't yet become a widespread technique in exploring genomes?

Dr. Mwita: (laughs) Not really, no. I started my PhD in 2013, at which time the current techniques in next-generation sequencing simply weren't available. The technology keeps improving all the time. I was fortunate in that there were several professors at UP whom I considered experts in bioinformatics and who had access to various resources; generally, I think there are more resources to perform bioinformatics research in South Africa as compared to many other African countries. I am lucky because I had the opportunity to work and learn in an area where facilities were available, unlike in my country. At present, you cannot conduct a PhD in bioinformatics wholly in Tanzania, you must look for additional resources outside the country.

Busisiwe: Hmm, I was about to ask if you've always had a strong interest in Bioinformatics but that guess you've pretty much answered that question!

Dr. Mwita: No, not really, not from the beginning but I think my interest was sparked by the actual opportunity to learn about it and do it. Growing up, no, I had no idea and no interest in the field. I consider my entry into the field more by chance and God's planning rather than my own planning. But since developing that initial interest and joining the field, I've grown in it and find it an exciting career path. We still have a scarcity of local expertise in bioinformatics, both in Tanzania and Africa as a continent.

Busisiwe: I've read about the efforts of different African countries to create interest in STEM, and especially in females, but it appears that more effort is required, and we haven't yet achieved gender equality in participation in science-based careers. Growing up, were you always interested in science?

Dr. Mwita: (laughing) In science, oh, yes! I was raised by medical doctors, so science was always considered important at home. No matter what area, I was always interested in developing a career in science. I agree that young people, and particularly women, may think that science is very difficult and prefer to pursue careers in other fields. But I think there are now many efforts to inspire the younger generation, particularly women, to undertake science subjects. Hopefully in 10-20 years we'll have a lot more women in science.

Busisiwe: Tell us about your most important role models, both in your career and private life, and how they've contributed to your being where you are today.

Dr. Mwita: My role models in my career began with my two supervisors in my master's degree. I was fortunate to continue being mentored by the same supervisors into my PhD. They're both men; (laughing) I don't know whether that's good thing in this context! What I learnt from them is hard work, discipline, patience in mentoring, how to plan my time effectively and so many other good qualities. They've been with me through the processes of learning to research, to publish, and have basically helped me grow into the field. They're also family people and value family time so that's another thing I picked up from them.

I've also learnt a lot from Professor Julie Makani since joining the Sickle Cell Program. So, I moved from being mentored by men to being mentored by a woman, but I think the qualities they've taught me and fostered in me are the same. I love the way Prof. Makani juggles various activities - lecturing, performing research, managing several grants and still being a wife and a mother. I think I learnt a lot from these three individuals and I'm trying to follow their steps.

In my private life, I consider my parents, particularly my mother, to be my role models. From my youth, they've taught me to work hard. I've learnt that whatever opportunities you get, work hard and try to achieve your goals.

Busisiwe: So, you briefly alluded to this, work-life balance; it's a phrase that's frequently heard in the workplace these days but it's difficult to achieve, especially for women in science. What do you do to try and live a balanced life?

Dr. Mwita: I think that is my daily struggle to balance. I always try to plan ahead, have a timetable, plan my tasks but also be flexible. Things don't always go according to plan! Sometimes you have a lot of work and deadlines to meet but a family member may fall ill. Prioritize, so you get the most important things done, and try to stick to the plan, but also build flexibility into your plan. You can find yourself concentrating on work to the detriment of your private life and social responsibilities, or you can find yourself so focussed on your private life that you fail to deliver at work. My three key points are to plan ahead, prioritize, but maintain flexibility. When things don't go according to the plan, it's important to have a plan B; accept what is happening and try to do better.

Busisiwe: With respect to building your career, what would you do differently if given the opportunity?

Dr. Mwita: Performing bioinformatics research in a developing country is still challenging. Expertise is scarce and other resources, like high performance computing centres, are also absent or few. Most research relies on grants, but we don't have much support from our local

governments, yet we still have to forge ahead in research. Given the opportunity and considering the field that I'm in, I wish we had more long and short local courses in bioinformatics in our local universities, as well as access to technical resources, so that we can build expertise to support research. I think we can get our governments to support local research by better communicating and demonstrating the value of research – how it can change people's lives and offer solutions to local problems.

Busisiwe: Tell us about your current project; what sparked your passion for this research topic?

Dr. Mwita: I have several projects but my main work at the Sickle Cell Centre, which is one of the H3ABioNet nodes, is to support research investigations that have genetic data and require interpretation. We work with GWAS data from individuals with Sickle Cell Disease (SCD) and our aim is to find genetic variants that account for the different phenotypes we see in our patients. SCD is a genetic disorder with different clinical manifestations. I've also just begun investigating genetic variants associated with anaemia, so this is a very new project. Prior to this, I was working with a team looking at genetic variants associated with liver function in SCD. From December, I'll also be working as a lecturer in the department of Pharmaceutical Microbiology and we're interested in incorporating a bioinformatics component in the master's curriculum, which is one way to build local expertise.

Busisiwe: What advice would you give to students considering entering this field?

Dr. Mwita: I think the most important thing is to have an interest in what you're doing; it's very difficult to work hard and achieve success in something that you don't like! The key to being successful in any field is to nurture your passion, and team hard work with discipline. Also, be open to learning through the process; qualities like time management and patience in the face of unexpected obstacles are important because research can be frustrating.

Busisiwe: What do you perceive to be the greatest obstacle(s) right now to African scientists performing the same calibre of research in the field of Bioinformatics as counterparts in more developed areas of the world?

Resuming after a brief break in communication…

Dr. Mwita: Ah, I thought the internet had disappeared (laughing)! Or that electricity had gone off at your end! These are still valid challenges in much of Africa.

I think one of the main obstacles for us is building local expertise. Also, we have to train and **retain!** Other challenges include computational resources such servers to store data; to purchase and maintain these resources is expensive. Funding is a common problem for many researchers as well; sometimes you wish to continue previously begun research but have no funds. You may have to restructure your

research in order to comply with the key interests of potential funders. Funding at the level of local government could help alleviate some of these constraints.

Busisiwe: Is there anything else you would like to share with our readers?

Dr. Mwita: I think H3Africa is doing great work in genomic research of common diseases with the goal of improving the health of Africans. Local researchers need to keep performing research despite the obstacles that we encounter in our local settings.

Another break in communication and we end the interview using the chat tool…

Dr. Mwita: Oh dear, the sound has gone – more African challenges!

Busisiwe: (my turn to laugh) Indeed! Dr. Mwita, I'm a bioinformatics student with the University of Botswana so it's really encouraging to hear what you've gone through and overcome. Thanks for sharing your journey with me.

Dr. Mwita: Thank you so much, Busisiwe. I'm glad to be a part of this. All the best for your PhD studies. Bye!

Standardizing the Data We Collect!

By Lyndon Zass, Judit Kumuthini, Katherine Johnston & Alia Benkhala

Over the past few years, the Human Heredity and Health in Africa's (H3Africa) Bioinformatics Network (H3ABioNet)

(www.h3abionet.org) has advocated the importance of standardising the data we collect and how we report this data. Standardising data collection does not only benefit future users of the data, but also allows current users to produce quality, FAIR (Findable, Accessible, Interoperable, Reproducible) (https://fairsharing.org/) data which can be not only immensely valuable for research purposes but a key driver for sustainability as well.

This has led to the development of several support resources which can be locally employed by the H3Africa consortium and its members in both current and future research endeavours. Adapting collection measures from the Phenotypes and Exposures (PhenX) toolkit, the H3ABioNet has developed the H3Africa Standard Case Report Form (CRF), which outlines a list of recommended and harmonized phenotypes which should be captured by the H3Africa projects as well as how these phenotypes should be captured.

While the Standard CRF can be employed specifically for the collection of study participants' phenotype data, the H3ABioNet has also expanded on this work and developed domain-specific harmonized data dictionaries for the collection and reporting of participant-, study- and experiment-level information. The domain-specific data dictionaries focus on specific fields of research within the H3Africa consortia, outlining recommended data elements that expand on certain sections within the Standard CRF. Unlike the Standard CRF, which can be used across domains within

the H3Africa, these data dictionaries are recommended for use within the domain for which they were designed. Thus far, the H3ABioNet has developed data dictionaries for

Excerpt of standard Case Report Form (CRF)

two domains – stroke and kidney disease research.

To ensure interoperable data collection and reporting, the recommendations outlined by both the Data Dictionaries and Standard CRF have been mapped to existing biomedical ontologies. Therefore, these tools allow researchers to collect phenotype data which are comparable and interoperable within the consortium (Standard CRF) as well as data which are comparable and interoperable beyond the consortium within specific research fields (Data Dictionaries). Importantly, work is continuing in both the Standard CRF and Data Dictionaries projects in order to refine and expand on specific phenotypes and domains for the benefit of the H3Africa consortium. The H3ABioNet encourages the consortium to reach

out to these projects if they wish to contribute to these efforts.

What to standardize in your data?

Both the Standard CRF and the Data Dictionaries can be directly implemented in the REDCap platform, for data collection and management. These tools can be downloaded from the H3ABioNet website, under the *Data Standardization* page. For more information, contact Katherine Johnston (katherine.johnston@uct.ac.za), Dr Alia Benkahla (alia.benkahla@pasteur.tn) or Dr Judit Kumuthini (judit.kumuthini@cpgr.org.za). Additionally, in order to request assistance in the employment of the aforementioned tools, visit the H3ABioNet Helpdesk, accessible via the H3ABioNet website.

"…reap the maximum benefit from project data…"

Ultimately, by standardizing the way in which we collect and report data, the H3ABioNet aims to empower and support the H3Africa consortium to reap the maximum benefit from their project data by adding scientific and ethical value and thus contributing to the sustainability of current research efforts.

Quiz Competition: mGenAfrica in Collaboration of Metro South Department of Education (Western Cape, Cape Town)

Rolanda Julius, Vicky Nembaware, Verena Ras, Kagisho Montjane, Paballo Chauke

mGenAfrica is an innovative internet-based platform and mobile application which aims to increase awareness and interest in careers in genomics and related fields and improve learners' knowledge in life sciences. The platform was started by the Pan African Bioinformatics Network for H3Africa (H3ABioNet) and is supported by various stakeholders and collaborators including the H3Africa Coordinating Centre (H3ACC), H3Africa working groups (Community Engagement and Education & Coordinated Training); the Sickle Africa Data Coordinating Center (SADaCC), the Wellcome Centre for Infectious Diseases Research in Africa (CIDRI) and the Metro South Department of Education (MSED)

mGenAfrica was officially launched on Saturday, 18 August 2018 in the New Lecture Theatre, Upper Campus, University of Cape Town through a High School Quiz Competition. The platform promotes activities such as quizzes, freely accessible learning material including videos, profiles of staff working in life sciences and live chat sessions, a career corner, translation corner and online competitions. We also have an active Facebook and twitter page where learners can conduct scheduled live chat sessions with researchers. Researchers can sign up as volunteers to chat live to learners via this website: https://mgen.h3abionet.org/. After signing up, the researchers' availability will then be captured on a calendar.

Over 340 high school learners represented by 39 schools participated in the first ever mGen Africa Quiz competition. On arrival, learners accompanied by their teachers were treated to coffee and muffins and received a goody bag packed with essential stationery. The welcome address was by Prof Nicola Mulder, Division of Computational Biology, UCT who advised and encouraged the school learners about careers in genomics. The quiz heats kicked off with several teams competing against each other online.

This was followed by a tasty lunch during which learners were free to visit several exhibition stalls. Exhibitors included Biosafety South Africa, Wellcome Centre for Infectious Diseases Research in Africa (CIDRI-Africa), GeneMap and H3Africa. Learners were seen with several exhibitor paraphernalia including branded pens, lanyards and stickers. UCT Radio's DJ Tau Setho, was the MC and entertained learners and teachers with a dance-off. The final round saw learners compete for top prizes which included scientific calculators, solar panelled backpacks, headphones with the grand prize being a laptop.

Gr 12 school learners competing in the mGenAfrica Quiz competition, University of Cape Town, South Africa

Recently, mGenAfrica was showcased at the Sci-Fest in Grahamstown Eastern Cape Province, South Africa. The 2019 festival, took place from 6-12 March with the theme "Discover your element" which celebrates the International Year of the Periodic Table of Chemical Elements as proclaimed by the United Nations. Over 60 000 school children of all ages, grades, races, genders and other diversity markers were in attendance to hear about different science careers. mGenAfrica had an exhibition stand where pupils were taught about genetics, bioinformatics and the newly created mobile app.

In the near future, other activities are being planned, including inclusion of Genomics into the national High School curriculum, exhibition booths to engage with mGenAfrica applications at science expos and museums and laboratory training for teachers. In addition, the Department of Education is keen to expand the quiz competition to the entire province.

Content adapted from the following sources:
https://www.news.uct.ac.za/article/-mgenafrica-sparking-interest-in-careers-in-genomics
https://mesh.tghn.org/articles/project-report-mgenafrica/

Software Carpentry - Teaching Basic Computing Skills to Young Researchers

By Ziyaad Parker

H3ABioNet hosted its first Software Carpentry workshop at the University of Cape Town, targeting young researchers aiming to learn basic computing skills in Linux, Git and Python. Although the workshop was only two days long, it provided students the opportunity to gain new knowledge and practice their skills. The first day consisted of Linux, Git tutorials and exercises while second day focused mainly on Python tutorials and exercises.
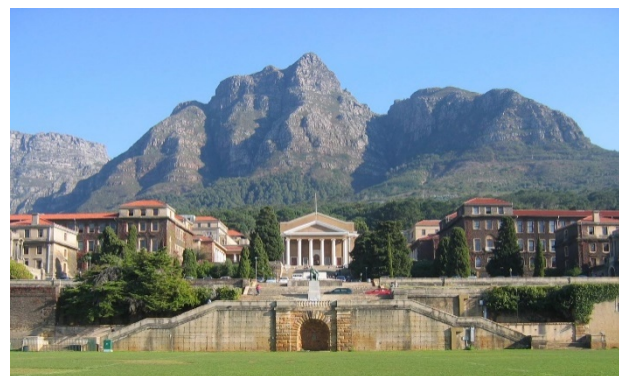
The workshop was headed by Ziyaad Parker (H3ABioNet), who previously attended training both Software Carpentry and FAIR (Findable, Accessible, Interoperable, Reproducible) Data Carpentry Instructor workshops. Lyndon Zass (H3ABioNet) was a helper who had also previously attended the Software Carpentry Instructor Training. The vision of the Carpentries for H3ABioNet is to have trainers in every H3ABioNet node, so when the need for Carpentry workshops arises from students across Africa there are resources locally available.

The workshop at the University of Cape Town consisted of just under 20 students with 4 instructors - Samar Elsheikh, Kenneth Babu, Mamana Mbyivanga and Ziyaad Parker. Male and female instructors were represented and all were positive role models. The teachings were conducted using the Carpentry teaching materials created by other content creators and instructors for previous workshops. The lessons consisted of hands-on practical sessions where the instructors explained the concepts and topics. The students were provided time to do an example and ask questions. Students worked directly from Github, Bash and Python. Additionally, common tools, workflows and the open science culture were communicated clearly in these workshops. One of the challenges was that two days was too short. Another challenge was that students from other nodes applied and wanted to attend, but due to funding shortage it was not possible for everyone to attend.

More detailed information about the content of the workshop can be found on the following website: https://h3abionet.github.io/2018-10-30-SWC-course/

If you are interested in having a Software, Data or FAIR Carpentry Training done in your region. Please fill in the following form: https://redcap.h3abionet.org/redcap/surveys/?s=9HEKACHPC9

The 12th H3Africa Consortium Meeting: An Extraordinary Experience

by Busisiwe Mlotshwa

The 12th H3Africa Consortium Meeting convened on the scenic grounds of the Radisson Blu Hotel in Kigali, Rwanda from September 16 – 23, 2018. This was an extraordinary meeting incorporating a joint session with the African Society for Human Genetics (AfSHG) and concluding with a Wellcome Genome Campus workshop on Next Generation Sequencing Analysis for Monogenic Diseases in African Populations.

The first day of the meeting, September 16th, commenced with a split session; Fellows Day took place at the College of Medicine and Health Sciences, University of Rwanda, whilst the workshop on Environmental Health in Africa was held at the Kigali Convention Centre (KCC). Professor Leon Mutesa gave the welcome address at Fellows Day, encouraging the fellows with highlights of his journey to becoming director of the Center for Human Genetics at the University of Rwanda. He provided snippets of advice that both young and established researchers clearly identified with:

Fellows' Day participants assembled for a group picture at the College of Medicine and Health Sciences, University of Rwanda.

Fellows' oral presentations were well-received and covered a gamut of topics reflecting the diversity of projects within the consortium. The day concluded with a Fellows' dinner back at KCC.

Day two of the meeting, September 17th, led straight into a mix of Working Group and PI Presentations sessions. The Biorepositories presented updates on their continued development and extended services. PI presentations covered updates on mostly established projects such as the Collaborative African Genomics Network (CAfGEN) and RHDGen: The Genetics of Rheumatic Heart Disease Network for H3Africa.

September 18th, the third day of the meeting, had a decidedly bioinformatics theme to it, with presentations focussing on topics such as data submission and biospecimen cataloguing as well as updates from various projects involved in expanding bioinformatics research capacity in Africa. The day ended with reports from the Working Groups, the newest of these being the Rare Disease Working Group led by Prof. Zanè Lombard. At the dinner that evening, Prof. Raj Ramesar delivered a fitting, and moving, tribute to the late Prof. Bongani Mayosi.

The joint AfSHG and H3Africa session on September 19th provided H3Africa researchers with the opportunity to showcase their projects to a larger audience. A panel discussion addressing the sustainability of genomics and human genetics in Africa gave participants much food for thought as the session closed. Participants also had the opportunity to go on various tours of Kigali throughout the meetings and visit several of the local markets.

September 20th covered another split session; the H3Africa Community Engagement workshop was well-attended by study
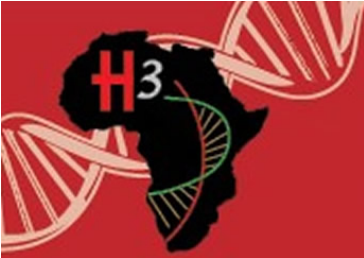
coordinators and researchers in ethics while Day 2 of the ASHG meeting opened with a workshop on the challenges in publishing from Africa. The open forum held over lunch, Establishing a '2020 Vision' for Genomics, attracted the interest of many fellows from both H3Africa and ASHG. The ASHG meeting wrapped up on September 21st with a helpful workshop on career opportunities for young African geneticists in the morning, and a strategic planning session in the late afternoon focussed on building skills and resources for genomics research in Africa.

The Wellcome Genome Campus workshop on Next Generation Sequencing Analysis for Monogenic Diseases in African Populations was held at the BPN, Kigali over September 22 - 23rd. Participants included researchers at all career stages. Workshop material was extensive, covering the basics of human genetics for those new to the field, but also expanding into variant analysis pipelines and interpretation of genetic variants.

"This is my first time attending an H3Africa Consortium Meeting; the keynote speakers were all brilliant…"

"The entire program was well organized."

"We had a great academic exchange."

In conclusion, the H3Africa Fellows would like to thank the funders and organizers (Dr.

Michelle Skelton and the H3ACC, Prof. Leon Mutesa and his team of cheerful volunteers), for arranging yet another successful and memorable conference.

Presentation and Poster Award Recipients from the 12th H3Africa Consortium Meeting

H3Africa 12th Consortium Meeting H3Africa Prize Giving: Laura Povlich (Fogarty/NIH), ErdawTachbele (3rd prize oral presentation), Janet Peace Babirye (1st prize poster presentation), Gaone Retshabile (2nd prize poster presentation, tied), Harriet Nankya (2nd prize poster presentation, tied), Segun Fatumo (2nd prize oral presentation), Busisiwe Mlotshwa (1st prize oral presentation) and Rolanda Julius (H3Africa training coordinator)

Training Corner by the ECTWG

In this edition of the H3Africa Newsletter, the training corner is highlighting some achievements by the H3Africa trainees and researchers. Congratulations!

Dr Tinashe Chikorowe
Tinashe was recently awarded a Wellcome Trust International Training Fellowship for his project: Characterization of gene-lifestyle interactions associated with obesity-related traits in African populations.

Dr Tinashe Chikorowe, Wits University

"Anyone who has ever made anything of importance was disciplined."— Andrew Hendrixson

Prof. Lynn Morris (Immunoglobulin project) Prof. Morris was awarded The World Academy of Sciences (TWAS) Prize in Medical Sciences for 2018. This is in recognition of her pioneering studies on the neutralizing antibody response to HIV infection that has provided fundamental insights for HIV vaccine development.

Prof. Lynn Morris being congratulated by Professor Felix DapareDakora, President of the African Academy of Sciences

Prof. Morris was also inducted as a new Fellow of the African Academy of Sciences (AAS) at an AAS General Assembly Meeting, held on 10-11 December 2018 in Pretoria. AAS Fellows are Africans who may live in or outside the continent and are elected by other AAS fellows based on achievements that include their publication record, innovations, leadership roles and contribution to policy.

Graduations

Dr Simone Richardson (Immunoglobin project) Simone recently obtained her PhD degree from the University of the Witwatersrand. Simone is actively involved in examining the impact of genetic diversity in the antibody constant regions on Fc effector function as part of the NICD's H3Africa project.

Simone with supervisors Professor Lynn Morris and Dr Nono Mkhize at her graduation

It's never perfect, so just submit the darn thing, already." - Steven L. Taylor

Bernardin Ahouty (TrypanoGEN) Congratulations to Bernardin who recently obtained his PhD in genetic epidemiology on 08 December 2018 at the University Felix Houphouet-Boigny of Abidjan (Ivory Coast). Bernardin affiliated with the TrypanoGEN project and was supervised by PI: Assoc. Prof. Enock Matovu and Co-PI in Ivory Coast: Prof. Mathurin Koffi

Bernardin at his graduation

(Please send us your achievements or awards for inclusion in this section!)

Publications from the Consortium

2018-2019:

Correction: The burden of dyslipidaemia and factors associated with lipid levels among adults in rural northern Ghana: An AWI-Gen sub-study.
Agongo et al. PMID:30811505

Model framework for governance of genomic research and biobanking in Africa - a content description.
Yakubu et al PMID:30714023

The Collaborative African Genomics Network (CAfGEN): Applying Genomic technologies to probe host factors important to the progression of HIV and HIV-tuberculosis infection in sub-Saharan Africa.
Mboowa et al.PMID:30714022

Data resource profile:
Cardiovascular H3Africa Innovation Resource (CHAIR).
Owolabi et al.PMID:30535409

Sociodemographic, socioeconomic, clinical and behavioural predictors of body mass index vary by sex in rural South African adults-findings from the AWI-Gen study.
Wagner et al. PMID:30499746

Developing reproducible bioinformatics analysis workflows for heterogeneous computing environments to support African genomics.
Baichoo et al. PMID:30486782

The burden of dyslipidaemia and factors associated with lipid levels among adults in rural northern Ghana: An AWI-Gen sub-study.
Agongo et al.PMID:30485283

Importance of human demographic history knowledge in genetic studies involving multi-ethnic cohorts.
Kulohoma BW.PMID:30430140

Gender differences in sociodemographic and behavioural factors associated with BMI in an adult population in rural Burkina Faso - an AWI-Gen sub-study.
Boua et al.PMID:30311555

Genomic and environmental risk factors for cardiometabolic diseases in Africa: methods used for Phase 1 of the AWI-Gen population cross-sectional study.
Ali et al.PMID:30259792

Relation Between Religious Perspectives and Views on Sickle Cell Disease Research and Associated Public Health Interventions in Ghana.
Dennis-Antwi et al. PMID:30171429

Demographic, socio-economic and behavioural correlates of BMI in middle-aged black men and women from urban Johannesburg, South Africa.
Micklesfield et al. PMID:30079826

Socio-demographic and behavioural determinants of body mass index among an adult population in rural Northern Ghana: the AWI-Gen study.
Nonterah et al.PMID:29992851

Sociodemographic and behavioural factors associated with body mass index among men and women in Nairobi slums: AWI-Gen Project.
Asiki et al.PMID:29966508