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Genomics and bioinformatics capacity in Africa: no continent is left behind

Gerald Mboowa, Ivan Sserwadda, and Dickson Aruhomukama

Abstract: Despite the poor genomics research capacity in Africa, efforts have been made to empower African scientists to get involved in genomics research, particularly that involving African populations. As part of the Human Heredity and Health in Africa (H3Africa) Consortium, an initiative was set to make genomics research in Africa an African endeavor and was developed through funding from the United States' National Institutes of Health Common Fund and the Wellcome Trust. H3Africa is intended to encourage a contemporary research approach by African investigators and to stimulate the study of genomic and environmental determinants of common diseases. The goal of these endeavors is to improve the health of African populations. To build capacity for bioinformatics and genomics research, organizations such as the African Society for Bioinformatics and Computational Biology have been established. In this article, we discuss the current status of the bioinformatics infrastructure in Africa as well as the training challenges and opportunities.

Key words: Africa, bioinformatics, genomics, Uganda.

Résumé : En dépit de la faible capacité en matière de recherche en génomique en Afrique, des efforts visant à habiliter les scientifiques africains à s'impliquer en recherche génomique, particulièrement celle visant des populations africaines, ont été faits. Le consortium "Human Heredity and Health in Africa (H3Africa)" est une initiative visant à faire de la recherche génomique en Afrique un travail véritablement africain et il a été mis sur pied grâce à un financement conjoint du United States' National Institutes of Health Common Fund et du Wellcome Trust. H3Africa cherche à encourager l'emploi d'une approche contemporaine en recherche par des chercheurs africains et de stimuler l'étude des déterminants génomiques et environnementaux de maladies communes. Le but de ces efforts est d'améliorer la santé des populations africaines. Pour augmenter la capacité de recherche en matière de bioinformatique et de génomique, des organisations comme l'African Society for Bioinformatics and Computational Biology (Société africaine de bioinformatique et de biologie computationnelle) ont été mises sur pied. Dans cet article, les auteurs présentent l'état actuel de l'infrastructure bioinformatique en Afrique de même que les défis et opportunités en matière de formation. [Traduit par la Rédaction]

Mots-clés : Afrique, bioinformatique, génomique, Uganda.

Introduction

Genomic technologies are developing at an unprecedented rate and continue to improve the potential to diagnose, treat, and elucidate the underpinnings of disease and health (Mulder et al. 2016). This is being achieved partially because of the reduced costs of DNA sequencing, evidenced by the acquisition of different genome sequencing platforms distributed on each continent. It is equally imperative for modern-day medical service providers in Africa to be knowledgeable in the applications and roles of genomics. Therefore, building capacity and expanding genomics training and research in Africa is key. Many institutions in Africa have developed bioinformatics training programs both at the degree and

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non-degree levels. The training programs are providing opportunities to researchers on the continent to utilize these skills in order to address health challenges among the native populations. A critical mass of trained individuals is yet to be attained. There is, however, an imbalance in the distribution of training institutions on the continent. Other challenges affecting bioinformatics training include funding and internet connection issues.

Historical perspective of bioinformatics in Africa

In Africa before the year 2010, bioinformatics and genomics training was largely done by small groups at very few institutions and heavily relied on visiting experts from Europe and America. In some cases, a number of individuals had to travel to the Western world to pursue graduate bioinformatics training; however, on their return to the continent, they were struck by the reality of the lack of computational infrastructure to translate their acquired knowledge and skills to build capacity. Out of frustration, a number of them would return to the Western world where they would get access to these resources as well as attractive remuneration. The inception of the African Society of Human Genetics (AfSHG) in 2003 (Morris 2010; Ndiaye Diallo et al. 2017; Rotimi 2004) subsequently led to the establishment of the Human Heredity and Health in Africa (H3Africa) initiative in 2011 funded through the National Institutes of Health (NIH)'s Common Fund and the Wellcome Trust (Adoga et al. 2014; Morris 2010). The AfSHG has since then seen the growth of member societies in African countries such as Cameroon (Cameroonian Society of Human Genetics (CSHG)), Democratic Republic of Congo (Congolese Society for Human Genetics (CoSHG)), Egypt (National Society of Human Genetics (NSHG)), Mali (Malian Society of Human Genetics (MSHG)), Rwanda (Rwandan Society of Human Genetics (RSHG)), Senegal (Senegalese Society of Human Genetics (S2GH)), South Africa (Southern African Society for Human Genetics (SASHG)), and Tanzania (Tanzania Society of Human Genetics (TSHG)). To ensure sustainable genomics capacity in Africa, H3Africa has built infrastructure and continues to train a critical mass of researchers who also have analytical skills. It has also established bioinformatics nodes in different countries in Africa, biobanks/biorepositories, and degree training programs. There is no doubt that the initiative has greatly boosted genomic endeavors on the continent.

Bioinformatics degree training has now extended to institutions in major regions of Africa; eastern, northern, southern, and western. Individuals who do not have access to this formal graduate training at their institutions, but have internet connection, are able to pursue online bioinformatics training from a number of platforms, such as H3ABioNet's "Introduction to Bioinformatics (IBT)" (Gurwitz et al. 2017), and many related workshops delivered by H3AbioNet, which is a pan African bioinformatics network for H3Africa. This initiative has stimulated and strengthened African collaborative partnerships within the continent as well as with high-income countries in the postgenomics era, ensuring that available resources can be effectively utilized laying the first major steps to guide precision public health on the continent. Precision public health is an emerging focus of public health that complements the development of precision medicine, utilizing advances in new technologies and knowledge unlocked through big data to better target public health efforts within populations (Weeramanthri et al. 2018). Efavirenz (EFV) is a first generation non-nucleoside reverse transcriptase inhibitor of HIV-1 and is one of the preferred component of the first line treatment regimen of HIV infection worldwide (Rakhmanina and van den Anker 2010; World Health Organization et al. 2006). In 2007, an African genomics scientist aided in the HIV public health program by showing that *CYP2B6* gene variant carried by many Zimbabweans slowed their ability to break down EFV. Consequently, the drug accumulated in the bloodstream, leading to hallucinations, depression, and suicidal tendencies (Nordling 2017; Nyakutira et al. 2008). After more than 8 years of H3Africa, the continent is no longer simply seen as a source of DNA but rather an important scientific collaborator.

Genome sequencing projects

Many countries in the developed world have undertaken massive genome sequencing projects aimed at improving human health. These include whole-genome sequencing projects led by Human Longevity, Inc. of San Diego, California. Human Longevity has already sequenced 25 000 human genomes and aims to sequence 100 000 genomes per year and to have one million genomes sequenced by 2020. The strategy for mining these data is to identify ways to understand how an individual's genes affect his or her health (McBride 2016). The All of Us research program in the United States is an ongoing genomic-focused NIH project aiming at collecting and analyzing health and lifestyle data from ≥1 000 000 volunteers in the US, including people with diverse gender, ethnic, and geographic backgrounds. Researchers will take into account the individual differences in lifestyle, environment, and human biology to uncover paths toward delivering precision medicine (National Institutes of Health n.d.). The United Kingdom's (UK) 100 000 Genome Project sequenced 100 000 human whole-genomes with the ultimate goal of making human whole-genomes part of regular UK's National Health Service (NHS) records (Marx 2015; Wheway et al. 2019). The Israeli government has set its target to sequence 100 000 volunteers for their health database project over the coming five years since 2018 (Gilmore 2018). In India, the first phase of the initiative, called the Genome India Project, is set to sequence and catalog a total of 10 000 Indians. The Genome India – Human Genome Cataloging Project aims at making predictive diagnostic markers available for some priority diseases,

including cancer (BioTecNika 2019). In China, 11 670 human genome sequences representative of the Han Chinese population from the CONVERGE project provided information that was used to understand origins in the Han Chinese when analyzed with genomic data from other human populations (Gao et al. 2020). Others include the Saudi Human Genome Program aiming at sequencing 100 000 human genomes by 2020 to conduct world-class genomics-based biomedical research in the Saudi population (Saudi Genome Project Team 2015); the UAE Human Genome Project aiming at establish the first national genetic database for future research, lending support to decision-makers as they set plans and strategies for the future of the healthcare sector (Dubai 10X 2018). The biggest of all these human genome sequencing endeavors is one from China known as the Precision Medicine Initiative, which is a 15-year project whose worth is US$9.2 billion, and its major goal is to sequence 100 000 000 human genomes through 2030 (Stark et al. 2019). A detailed global catalog of presently ongoing national genomic-medicine initiatives is beyond the scope of our article but described in Stark et al. (2019).

The African continent has taken major strides too. These include the formation of the H3Africa Consortium. However, African governments must be ready and willing to support the initiative at different levels including formulating policies that strengthen the collaborations between African genomics researchers and their Western counterparts. This would be best achieved at the continental union level through the African Union. In that respect, local governments must support their scientists so that they can retain them and attain critical numbers to catch up with the rest of the world.

The endorsement of next-generation sequencing technologies by the World Health Organization for the detection of mutations associated with drug resistance in Mycobacterium tuberculosis complex (World Health Organization 2018) is both an application and example that calls for a paradigm change in the training requirements for graduates, and even for the current human resource working at the disease diagnostic centers, to embrace the genomic evolution. The skills required to generate, interpret, and disseminate this big data require knowledge of genomics and bioinformatics. To date, whole-genome sequencing has been generally performed in research laboratories where results are not necessarily packaged in a clinician-friendly format. The implication of this technological evolution is that both clinicians and laboratory personnel need to embrace the technology either through their formal training or enroll for "on-job" related training programs. Over time, bioinformatics was mainly restricted to settings with sophisticated scientific resources. Nevertheless, numerous low- and middle-income nations have recently made strides in genomics and bioinformatics training as well as their application (Karikari 2015). Current improvements in research funding, infrastructural support, and capacity building continue to shape bioinformatics into an important discipline in Africa. The contributions of these are leading to the establishment of world-class research facilities, biorepositories, training programs, and scientific networks as well as funding schemes to improve studies involving human disease and health in Africa (Karikari 2015). Various health-related applications continue to emerge from genomic activity for some infectious diseases, particularly those endemic in Africa (Mbaye et al. 2019). Also, the arrival of next-generation diagnostics, drugs, and vaccines as well as surveillance tools could be enhanced by improving the nature and degree of participation of African scientists in genomic and postgenomic inquiry (Mbaye et al. 2019).

Bioinformatics training programs

Training and education in the basics of bioinformatics and genomics could begin at the level of high schools. This would improve the teaching of other subjects such as genetics, evolution, and biochemistry at the next level, whereas more advanced training could be done at the undergraduate and graduate levels (Karikari and Aleksic 2015). An example is an "Introduction to Bioinformatics" course that is offered to undergraduate students at the University of the Western Cape by the South African National Bioinformatics Institute (SANBI). This program includes Comparative Genomics, Introduction to Biological Databases, and Phylogenetics to better prepare medical students for applications of genomics/bioinformatics and data-driven medicine (Brazas et al. 2014). There is a growing demand for bioinformatics qualifications and applications from across Africa (Machanick and Tastan Bishop 2015), and health professionals must be knowledgeable about genomics so as to proficiently use the outcomes of genomics research successfully (Collins et al. 2003). African institutions should aim at training highly competent scientists who will utilize the technologies to address African health and disease-related challenges to change the imbalance of global trends in genomic studies. There are a number of graduate training programs in Africa (Table 1) though mainly in South Africa. Other institutions offering bioinformatics-related training in Africa include the Kenya Medical Research Institute (KEMRI), Kenya Wellcome Trust Research Programme (KWTRP), International Centre for Insect Physiology and Ecology (ICIPE) – Kenya, the Biosciences eastern and central Africa – International Livestock Research Institute (BecA–ILRI) Hub – Kenya, and the MRC/UVRI Uganda Medical Informatics Centre (UMIC), a UK Medical Research Council funded initiative. The African Bioinformatics Education Committee (ABEC) was created from an H3ABioNet degree development workshop held in Gaborone, Botswana, in March 2014 with the aim of providing general support for setting up and monitoring Bioinformatics degree programs in Africa (H3ABioNet n.d.). The

Table 1. Graduate bioinformatics training programs in Africa.

Bioinformatics program	University	Country
M.Sc. in Bioinformatics*	University of the Western Cape	South Africa
M.Sc. in Bioinformatics and Computational Molecular Biology†	Rhodes University	South Africa
M.Sc. in Bioinformatics	University of Pretoria	South Africa
M.Sc. in Bioinformatics	Covenant University	Nigeria
M.Sc. in Bioinformatics	University of Sciences, Techniques, and Technologies of Bamako	Mali
M.Sc. in Molecular Biology & Bioinformatics	Jomo Kenyatta University of Agriculture and Technology	Kenya
M.Sc. in Bioinformatics	Pwani University	Kenya
M.Sc. in Bioinformatics‡	Future University	Sudan
M.Sc. and Ph.D. in Bioinformatics§	Makerere University	Uganda
M.Sc. in Bioinformatics‖	University of Malawi	Malawi
M.Sc. and Ph.D. (by research thesis) in Bioinformatics¶	University of Cape Town	South Africa

*https://www.uwc.ac.za/Faculties/NS/SANBI/Pages/Training.aspx.
†http://www.h3abionet.org/component/attachments/download/186.
‡http://www.h3abionet.org/component/attachments/download/188; http://www.h3abionet.org/component/attachments/download/175.
§https://breca.mak.ac.ug/.
‖https://training.h3abionet.org/curriculum_development_wg/wp-content/uploads/2014/03/University_of_Malawi_COM_Bioinformatics_Curriculum_19_10_2015_proposal.pdf.
¶http://www.cree.uct.ac.za/sites/default/files/image_tool/images/47/2018/UCT_2019_applicant_guide.pdf.

H3ABioNet's recommended Bioinformatics curriculum degree program guidelines can be viewed. For example, in Uganda, an initial investment has been made to provide the computational infrastructure to facilitate the training (Table 2) and genome sequencing platforms (Table 3).

Massive open online courses (MOOCs)

Currently, genomics and bioinformatics learning occurs beyond classrooms as most of the learning is via simplified online resources and activities, which leaves the classrooms to become dedicated to small-group learning interactions (Brazas et al. 2014). The online platforms are very popular at the moment and include H3ABioNet (https://www.h3abionet.org/), Coursera (https://www.coursera.org/), Udacity (https://www.udacity.com/course/tales-from-the-genome–bio110), edX (https://www.edx.org/course?search_query=bioinformatics; https://www.edx.org/course?search_query=genomics), FutureLearn (https://www.futurelearn.com/), the Kahn Academy (http://www.khanacademy.org), Class Central (https://www.classcentral.com/), and MOOCs list (https://www.mooc-list.com/). These offer a range of basic and advanced courses in bioinformatics, genomics, genetics, computing, statistics, and modeling (Loman and Watson 2013). Recently, online learning initiatives have become progressively comprehensive in their selection of courses and sophisticated in their presentation. This has led to the recent announcements of a number of consortia and startup activities that have made the promise to make university education on the internet free of charge a real possibility (Searls 2012). Also, one can now explore the unlimited potential for obtaining comprehensive bioinformatics/genomics training with currently existing free video resources. David B. Searls presents a bioinformatics curriculum in the form of a virtual course catalog, together with editorial commentary, and an assessment of strengths, weaknesses, and likely future directions for open online learning in this field. H3ABioNet has developed a portal for the African Genomic Medicine curriculum and related resources that ensure a linkage of the H3ABioNet portal to the existing online curricula management systems in various training institutions within African universities and colleges with provision of "train-the-trainer" initiatives in genomic medicine; this has thereby increased awareness and adoption of the genomic medicine curricula made possible (Nembaware et al. 2016).

Genomic research in Africa

The genomic era is now a reality (Collins et al. 2003). No continent should be left behind regarding the "omics" revolution. Genome-wide association studies exploited these technological developments in large case-control studies, with unprecedented success (Price et al. 2015) and rapidly unraveled the role of host genetic factors in pathogenesis of common diseases (Manolio et al. 2008). Despite the findings from genomic studies having universal clinical impact, these studies have mainly focused on Caucasians (Popejoy and Fullerton 2016). It has been revealed that only 3% of the global genome-wide association studies that link genetic traits to patterns in health, disease, and drug tolerance had been performed on Africans, compared with 81% on Caucasians (Popejoy and Fullerton 2016), yet Africa is both home to modern humans (Chan et al. 2019) and is inhabited by the most genetically diverse people in the world (Birney and Soranzo 2015). Major efforts must be made to pledge that advances in genomics are applied to

Table 2. Genomics and bioinformatics research groups in Uganda.

Research group	Research focus	Funders/Collaborators	Computation capacity
TrypanoGEN	Human genetic susceptibility to African trypanosomiasis	Wellcome Trust and National Institutes of Health	COVAB – 16 TB storage and 1024 GB DDR3 RAM linked to CIRDES; Burkina Faso – 144 TB storage and 128 GB DDR3 RAM
Nurturing Genomics and Bioinformatics Research Capacity in Africa (BRECA)	Genomics and bioinformatics graduate training programs	National Institutes of Health – Human Heredity and Health in Africa (H3Africa): Global Health Bioinformatics Research Training Program (U2R)	8 Mac workstations (21-inch) – 8 GB memory, 2.7 GHz Intel Core i5, 1 TB hard drive; 4 MacBook Pro (15-inch) – 16 GB memory, 2.2 GHz Intel Core i7, 250 GB hard drive, supported by servers with upto 40 TB storage and 500 GB RAM
Collaborative African Genomics Network (CAfGEN)	Mechanistic insights to pediatric HIV and HIV–TB disease progression	National Institute of Allergy and Infectious Diseases (NIAID), the Botswana and the Uganda Children's Clinical Centers of Excellence, Makerere University and the University of Botswana, and Baylor College of Medicine, Houston	8 Mac workstations (21-inch) – 8 GB memory, 2.7 GHz Intel Core i5, 1 TB hard drive; 4 MacBook Pro (15-inch) – 16 GB memory, 2.2 GHz Intel Core i7, 250 GB hard drive, supported by servers with upto 40 TB storage and 500 GB RAM
Uganda Medical Informatics Centre (UMIC)	Human research capacity in bioinformatics and computational genomics	UK Medical Research Council (MRC), Wellcome Trust Sanger Institute / University of Cambridge	64XP blade server, with 2048 cores and 16 TB RAM, virtualization capacity of 4X HP servers, with 20 cores and 256 GB RAM each. Storage capacity of up to 256 TB usable disk space. Back up capacity of 348 TB. Networking capacity of 10 GbE switches and MX104 routers as well as regional connectivity of up to 1 Gbs
The African Center of Excellence in Bioinformatics & Data-Intensive Sciences	Bioinformatics and data-intensive sciences	National Institute of Allergy and Infectious Diseases and the Office of Cyber Infrastructure and Computational Biology (NIH/NIAID/OCICB)	24 high-end workstations with local NUCs and server connection; 6 virtual reality workstations and 8 GPU processors; high performance computing servers with 1500 TB ZFS storage; cluster of 16 nodes and 32 cores/node; minimum 8 GB/core (512 GB/node) and 1 high memory node (2 TB+); connectivity on RENU network: 100 GB ethernet ToR with internet connectivity: 20 GB; 25 laptop computers with 16 GB RAM, 512 GB, processor i7 CPU 2.9 GHz

health improvement of even people living in low- and middle-income countries; however, this remains an important contemporary challenge (Pang 2002).

The huge storehouse of data produced by the Human Genome Project is being used to change every aspect of biological research and biomedicine. In the past eight years, more than US$100 million has been invested in projects to boost genetic research involving African populations by international research-funding organizations (Nordling 2017). There are a number of African-based genome sequencing initiatives that have generated thousands of African genomic datasets. These include TrypanoGEN (http://www.trypanogen.net/), MalariaGEN (https://www.malariagen.net/), the Collaborative African Genomics Network (CAfGEN) (Mboowa et al. 2018), and other human genome/exome sequencing projects that

Table 3. Genome sequencing platforms in Uganda.

Institution	Genome sequencing platforms
Uganda Virus Research Institute (UVRI)	iSeq 100 Sequencing System; Illumina MiSeq Next Generation Sequencer; Oxford Nanopore's MinION Sequencing; Sanger ABI 3500 Genetic Analyzer
TB Supranational Reference Laboratory (SRL)	Illumina MiSeq Next Generation Sequencer
Makerere University, Department of Immunology and Molecular Biology	Illumina MiSeq Next Generation Sequencer; Applied Biosystems SeqStudio Genetic Analyzer
Makerere University, Infectious Diseases Institute (IDI), Core Laboratory	Sanger ABI 3500 Genetic Analyzer
MBN Clinical Laboratories	Sanger ABI 3500 Genetic Analyzer
Joint Clinical Research Centre (JCRC)	Sanger ABI 3730XL Genetic Analyzer

are extensively described in Bope et al. (2019). The African Genome Custom Chip has been central to bringing these initiatives together to ensure that they capture all the genetic variation identified from these genomic sequencing projects. This was to ensure that all the common human genetic variation on the continent can be represented on the same SNP chip. This offers a great boost to the future of genomic research on the continent while utilizing array genotyping technology. For Africans to gain from their genome data, capacity must be built on the continent and necessary strategies laid to retain it. The recent endeavors in genomic research in Africa have already identified 2 700 000 previously unrecorded SNPs (Nordling 2017), which have potential implications on human origin, disease patterns, health, and drug tolerance.

A generation of graduate-level well-trained African genomic researchers is needed to chart the impact of these newly identified variations among the populations on the continent. The implication of this is that major biomedical research questions relevant to the African population can be addressed by research scientists working in Africa (Karikari and Aleksic 2015). The microarrays chip is being utilized to economically screen thousands of Africans to identify and understand genetic predispositions to complex traits and common infectious diseases in Africa, achieved by comparing allele frequencies across the genomes of large numbers of disease-specific cases and controls. This alleviates the limitations of non-economical sequencing to identify disease-associated alleles. Inadequate knowledge and understanding of the genomics of pathogens and their respective hosts has been identified as the major challenge in the control of infectious diseases in Africa (Folarin et al. 2014). Understanding the African human genomes, those of continental pathogens and inter-population genetic variability would result in radical advances in the management of the common infectious diseases on the continent and in medicine at large. The achievements of H3Africa span over a range of research areas aimed at understanding the genetic diversity of African populations and the genetic underpinning of disease susceptibility. Most of the H3Africa-funded research projects are still ongoing and their outcomes are yet to be published and include the following:

- Genomic and environmental risk factors for cardiometabolic disease in Africans
- Eye of Africa: The genetics of blindness
- Genomic characterization and surveillance of microbial threats in West Africa SickleGenAfrica: Sickle Cell Disease Genomics Network of Africa
- Center for Research on the Respiratory Microbiota of African Children (ReMAC)
- Stroke Investigative Research & Educational Network (SIREN)
- Collaborative African Genomics Network (CAfGEN)
- H3Africa Kidney Disease Research Network
- African Collaborative Center for Microbiome and Genomics Research (ACCME)
- Burden, spectrum, and etiology of type 2 diabetes in sub-Saharan Africa
- TrypanoGEN: An integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis
- The RHDGen Network: Genetics of rheumatic heart disease and molecular epidemiology of *Streptococcus pyogenes* pharyngitis

Bioinformatics and genomics in Uganda

In Uganda, bioinformatics was initially mainly done by individual groups at Makerere University and the Uganda Virus Research Institute (UVRI). It was the dawn of H3Africa that awarded four genomics projects to research teams at Makerere University, Uganda's premier research institution. These included the Collaborative African Genomics Network (CAfGEN), Integrated Biorepository of H3Africa Uganda (IBRH3AU), Integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis (TrypanoGen), and Nurturing Genomics and Bioinformatics Research Capacity

in Africa (BReCA). These four named projects have continued to support each other in so many ways, creating a perfect demonstration of synergy that was fostered by H3Africa on the continent. Another important outcome of the collaborative genomic initiative in Uganda is a genome resource to enable insights into human population history and genomic discovery in Africa. It has led to what is so far Uganda's largest genome-wide association study. This project incorporated whole-genome sequencing data from 1978 individuals from rural Uganda, providing evidence of geographically correlated fine-scale population substructure (Gurdasani et al. 2019). The country has since seen great development in bioinformatics with the introduction of the H3Africa Consortium and immensely benefited from H3Africa initiatives. Present at UVRI is a bioinformatics node for H3ABioNet as well the Uganda Medical Informatics Centre (UMIC) that has influenced the growth of bioinformatics in Uganda through offering pieces of training and computational resources. In 2017, the country hosted the ISCB/ASCB Annual Conference at UVRI in Entebbe (Rafael et al. 2017), and two years later (2019) it took a step to create a stable footing for genomic medicine in the future by starting its first graduate degree program in Bioinformatics whose approval required having built a critical number of bioinformatics scientists to take lead in the training of the students. This is another aspect that has demonstrated the sustainability of genomics and bioinformatics initiatives in the country even beyond the H3Africa Consortium.

Challenges and opportunities in genomic research

As a substantial number of African scientists, including skilled bioinformaticians, continue to leave the continent in search of better job opportunities that offer more competitive salaries, a strain is created on local training and research institutions where the need for bioinformatics expertise is on the rise (Atwood et al. 2015; Field et al. 2006; Ranganathan 2005). The CAfGEN has established sustainable genomics research programs in both Uganda, Botswana, and Eswatini via long-term training of Ph.D. students from these countries at Baylor College of Medicine, Houston, Texas (Mlotshwa et al. 2017). The main challenge facing genomic researchers and clinicians is limited resources (Helmy et al. 2016). High-throughput sequencing (HTS) generates short-read sequences that are used to build the sequences of whole-genomes using complex computer algorithms that require substantial computational processing capabilities (El-Metwally et al. 2013). The genomics reads obtained from most platforms require respective reference genomes as a substrate for mapping alignment prior to variation discovery (Price et al. 2015), annotation, and successful interpretation of genomic data alone (Glenn 2011). High costs are involved in setting up and subsequently maintaining a genomics sequencing facility. This remains a challenge affecting applications of HTS in low-income countries. The estimated cost of purchasing sequencing instruments alone so as to establish an HTS facility range between US$100 000 and US$700 000 (Glenn 2011). Other costs incurred in the process include obtaining skilled personnel, setting up the necessary infrastructure/computational facilities, cost of long-term data storage, buying of reagents, and instrument maintenance (Glenn 2011). This is not readily affordable for most universities and research institutions in low- and middle-income countries; however, it can be in part solved through fostering international collaborations. For example, in Rwanda, the Department of Genetics was opened in 2005 with a grant from Coopération Universitaire au Développement from Belgium and the National University of Rwanda (Mboowa and Sserwadda 2019; Uwineza and Mutesa 2015).

Bioinformatics in Africa faces a challenge of slow, unstable, and expensive internet connection services. But through international collaborations, local institutions have tried to solve the above- mentioned challenges. For example, at Makerere University, the Research and Education Network for Uganda (RENU) offers stable internet connection via ethernet and Wi-Fi. The bandwidth is 360 Mbps and keeps increasing annually, making it a fast, reliable service that can be used for transfer and processing of large data sets. RENU regionally connects to UbuntuNet alliance, which is internationally connected to other research education networks in Africa, the Americas, and Europe. RENU offers a platform that promotes collaboration between higher education institutions. Researchers, lecturers, students, and managers in member institutions freely connect with their peers nationally, regionally, and globally, and they are able to share research experiences across the world via live videoconferencing, webinars, and skype calls at very good speeds.

Comprehension of genomics research studies, quality of research ethics review processes, and understanding of the implications of broad consent, secondary analyses of shared data, dissemination of results, and incidental findings to communities (Mulder et al. 2017) continue to undermine genomic developments in Africa. Additional challenges with genomics research in Africa include the inability to have genomics-knowledgeable individuals constituting some ethical review boards or committees. The costs of running and maintaining computing infrastructure are often very high for most individual institutions to meet, especially due to an unstable electricity supply. African genomic researchers are sometimes unwilling to share both genomic data generated (Mulder et al. 2017) and computational resources, limiting timely analyses that can offer immediate discoveries.

In Africa, government policies have not prioritized research funding for genomics. However, the government of South Africa has taken an exemplary initiative to fund genomic research through the South African National Research Foundation; this has boosted

national development and is the number one funder for South Africa in genomics research (Karikari et al. 2015; Mulder et al. 2016). The development of training programs is one of the most vital aspects of advancing genomics in low- and middle-income countries. Methods of training bioinformaticians and genomics scientists have been introduced by taking a good number of researchers to overseas organizations who return to train others in Africa (Chimusa et al. 2015). This approach offers a fairly cost-effective approach to training and introducing HTS technologies on the continent.

Workshops and seminars

Workshops and seminars offer short-term training sessions; these target those individuals who require basic knowledge in bioinformatics and genomics, but they are also appropriate for those who want to refresh or enhance their skills in these fields. Short-term trainings form a foundation for professional graduate programs. In Uganda, for example, these courses have evolved into a fully-fledged graduate program (https://www.breca.mak.ac.ug/). A primer bioinformatics workshop at Makerere University (conducted on 5–6 June 2017) funded by a University of Georgia, Boston University, and Case Western Reserve University collaboration was attended by over 40 graduate students; UC Davis Bioinformatics Core & Alliance for Global Health and Science ran a weeklong (24–28 July 2017) combination of bioinformatics lecture series and practical sessions on RNAseq analysis (Henry Wheeler Center for Emerging and Neglected Diseases n.d.). A number of other groups have also organized onsite bioinformatics and genomics works such as Viral Bioinformatics and Genomics (23–28 June 2019) at Makerere University funded by Wellcome Trust; Genome Campus Advanced Courses and Bioinformatics & Next Generation Sequencing Techniques Short Course (5−21 February 2018) funded by The Africa Center of Excellence in Materials, Product Development & Nanotechnology (MAPRONANO ACE) (Wamai 2018). Between 9 May and 4 August 2017, a total of 56 graduate students signed up for an intense three-month online H3ABioNet course "Introduction to Bioinformatics (IBT_2017)"; unfortunately, a less than 15% completion success rate was seen. This low pass rate in IBT_2017 was partially due to the fact that this course was rigorous and ran for three months, making it hard for the students who are already enrolled in their own full-time courses to complete all the necessary IBT_2017 requirements to obtain a minimum mark of 60% in the assessments overall alongside submitting at least 90% of practical assignments by the relevant course due dates. At Makerere University, "Introduction to Bioinformatics" has been taken in iterations during 2018 and 2019, but this same year included a new "16SrRNA Microbiome Intermediate Bioinformatics" course (H3ABioNet n.d.) alongside "Introduction to Bioinformatics", targeting specifically those students who had done the earlier two iterations of "Introduction to Bioinformatics".

Funding, collaborations, and partnerships

In 2017, the NIH Common Fund and Fogarty through the new Global Health Bioinformatics Research Training Program awarded grants to four institutions in Africa to support the H3Africa Initiative. The awards are meant to provide five years of support totaling up to $5 million for interdisciplinary training for bioinformatics scientists at a network of research sites across Africa. The training will help develop skills to lead integrative teams to solve significant global health problems in Africa that are important to Africans (Fogarty International Center @ NIH 2017). The institutions included Covenant University (Nigeria) in partnership with University of Bamako (UB), Mali; Kwame Nkrumah University of Science and Technology (KNUST), Ghana; University of Sciences, Techniques and Technologies of Bamako, Mali; International Centre of Insect Physiology and Ecology (ICIPE), Kenya; and Makerere University, Uganda.

Perspective

Entrusting HTS analyses to professional bioinformatics scientists may be beneficial but enabling individual researchers to perform their own analyses of genomic and other large-scale HTS data is a more sustainable model. This may increase innovation since the respective researchers have a deeper insight into the biological question under investigation. Although Africa is the second largest continent by geographic area and is the second most populated continent in the world, it has the highest rate of population growth (https://www.un.org/en/sections/issues-depth/population/) and continues to register an ever-increasing incident of noncommunicable diseases and emerging and re-emerging infectious diseases. In 2017, the World Health Organization projected that deaths from noncommunicable diseases were likely to rise globally by 17% over the next 10 years; the region would experience a 27% increase, that is 28 million additional deaths from these conditions which are projected to exceed deaths due to communicable, maternal, perinatal, and nutritional diseases combined by 2030. In some African countries, such as Mauritius, Namibia, and Seychelles, noncommunicable diseases cause over 50% of all reported adult deaths. This implies that noncommunicable diseases will soon be a leading cause of ill health, disability, and premature death on the continent, and it will have an adverse impact on socio-economic development (https://www.afro.who.int/health-topics/noncommunicable-diseases). This situation will require a better understanding of the roles of lifestyle, environment, and human host genetic factors in relation to noncommunicable diseases besides infectious diseases. HTS technologies and their application stand to play a pivotal role in this new era; therefore, national/

regional governments and their respective educational and health sectors must be ready to embrace this new reality.

Lastly, with respect to the notion that modern humans descended from Africans, the African genome has much to offer in our understanding of genetic contribution in health and diseases. African nations still have more to benefit from the collaborative opportunities with higher-income nations through sharing their experiences from the lessons learned during the large-scale population-level genomic studies they have undertaken previously.

Conclusions

The H3Africa Consortium has been a successful endeavor and now happens to be in its final funding phase. We envisage that for the sustainable application of genomics technologies in Africa, respective governments must show commitment through the continental African Union's New Partnership for Africa's Development (NEPAD) Agency, the eight African sub-regional bodies as seen from other western counterparts, or the African Academy of Sciences (AAS).

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Competing interests

The authors have declared that no competing interests exist.

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