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Meeting Report

Inauguration of the Tanzania Society of Human Genetics: Biomedical Research in Tanzania with Emphasis on Human Genetics and Genomics

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Abstract. Human genetics research and applications are rapidly growing areas in health innovations and services. African populations are reported to be highly diverse and carry the greatest number of variants per genome. Exploring these variants is key to realize the genomic medicine initiative. However, African populations are grossly underrepresented in various genomic databases, which has alerted scientists to address this issue with urgency. In Tanzania, human genetics research and services are conducted in different institutions on both communicable and noncommunicable diseases. However, there is poor coordination of the research activities, often leading to limited application of the research findings and poor utilization of available resources. In addition, contributions from Tanzanian human genetics research and services are not fully communicated to the government, national, and international communities. To address this scientific gap, the Tanzania Society of Human Genetics (TSHG) has been formed to bring together all stakeholders of human genetics activities in Tanzania and to formally bring Tanzania as a member to the African Society of Human Genetics. This article describes the inauguration event of the TSHG, which took place in November 2019. It provides a justification for its establishment and discusses presentations from invited speakers who took part in the inauguration of the TSHG.

INTRODUCTION

Africa, referred to as the cradle of mankind, is the birthplace of modern humans who have lived in the continent for over 300,000 years.[1,2] Consequently, this evolutionary history has led to various genetic characteristics that are specific to the African populations as elucidated from human genome projects such as the International HapMap,[3] the 1,000 genomes project,[4] and the African genome variation project.[5] Insights from these projects have highlighted the richness of human genetic variation in African populations including haplotype diversity and complex patterns of population structure compared with other populations.[6] However, African populations remain understudied and underrepresented.[7–9] A recent meta-study revealed that most genome-wide association studies related to health have been focused on European populations (88%), whereas only 3% of the studies have been performed in Africa.[10] This is because of a number of reasons including limited funding, human resource, and required infrastructure.[11]

Globally, it is now agreed that without proper understanding of the genomes of African populations, important questions in genomic medicine and biology cannot be answered. This is because of the genetic structure of African populations that offers among other things, low linkage of genetic variants, hence making it easier to identify potential disease-causing variants. It is known that the occurrence, susceptibility, and resistance to many disorders/diseases, both rare and common, are influenced by two key factors: environmental and genetic, both being highly diverse in Africa. It is therefore important to understand the genetic factors that contribute to human diseases in Africa to transform medical diagnosis, prevention, and treatment.

According to the WHO, eight million children are born each year with serious birth defects that are of genetic or partially genetic origin, 90% of which occur in low- or middle-income countries.[12] An example of this is sickle cell anemia, which is the most common monogenic disease, with more than 300,000 children born each year globally, majority of who reside in sub-Saharan Africa (SSA).[13] However, in Africa, because of the high prevalence of communicable diseases such as HIV infections, tuberculosis, and malaria, studies of genetic conditions are often overlooked because of the limited resources. Most countries in SSA lack clinical and diagnostic facilities that are needed to address disease conditions that are genetic in nature. This issue requires significant investment in the areas of knowledge, research, human resources, equipment, policy, and adequate infrastructure.

To date, there have been various initiatives to address this gap. The African Society of Human Genetics (AfSHG, https://www.afshg.org/) is a remarkable network that was founded in

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2003, aimed at equipping the growing scientific community in the continent on matters pertaining to human genetics.[14] The main objective of the AfSHG is to facilitate the congregation of scientists in the field of human genetics and genomics in Africa, to interact, network, and collaborate (https://www.afshg.org/about/). The inaugural meeting of the AfSHG was themed "Biomedical research in Africa with emphasis on genetics" and took place in Accra, Ghana, in December 2003.[14] Since then, 11 conferences have taken place throughout Africa.[15–19] Apart from conferences, the AfSHG is a mother organization to country-specific human genetics societies across Africa, providing them necessary support in driving the genetics agenda in the continent.

Tanzania is among the African countries lagging behind in implementing the human genetic/genomics agenda. To date, there is no formal database of genetics-related/influenced diseases in the country. However, some of genetic studies involving samples from Tanzania or conducted in the country include the Human Heredity and Health in Africa (H3A) studies in diabetes,[20] tuberculosis,[21] albinism,[22] sickle cell disease (SCD),[23,24] and malaria.[25] To address this gap, the Tanzania Society of Human Genetics (TSHG) was formed in 2017. This article describes the inauguration event of the TSHG that took place in November 2019. It provides a justification for its establishment and discusses presentations from invited speakers who took part in the inauguration event.

ESTABLISHMENT AND ROLES OF THE TSHG

The TSHG was conceptualized in 2017 and became the eighth society in Africa after those in Cameroon, the Democratic Republic of the Congo, Egypt, Mali, Rwanda, Senegal, and South Africa. The TSHG was formed with a mission to coordinate human genetics research and related activities in Tanzania to generate knowledge and recommendations for the prevention, diagnosis, and treatment of genetic diseases and promotion of health. Consequently, the TSHG advocates for research and training of a workforce to hasten the development of services and products centered on genetic technologies. Other activities the society engages in include consultancy in health and genetics intersection as well as generating awareness about the genetics of human beings as related to human health.

The society seeks to promote knowledge generation through research and education with an ultimate goal of improving human health. Genetic research being a resource-intensive arena is imperative to consolidate resources and human capacity among actors such as government and private institutes, including those in research, academia, civil society, and the private sector. By achieving these goals, the TSHG hopes to bring attention to and help facilitate the development of solutions to public health burden of many rare and common genetic disorders in Tanzania.

INAUGURATION OF THE TSHG

The TSHG was officially inaugurated on November 28, 2019, concurrently with the Tanzania Health Summit in Dodoma, Tanzania. The inauguration attracted more than 200 stakeholders from various parts of the country representing the health and allied sciences fields. The meeting was themed "Biomedical research in Tanzania with emphasis on human genetics and genomics" and was officially launched by the Deputy Minister for Health, Community development, Gender, Elderly and Children.

The meeting highlighted the status of research and application of human genetics in Tanzania as well as lessons learnt from other countries in Africa. Members of the society and stakeholders took advantage of the forum to strategize potential collaborations, establish partnerships, and work toward driving the agenda of improving diagnosis and treatment of genetic disorders in the country.

The keynote speaker, Professor Collen Masimirembwa, gave highlights on "Pharmacogenetics in Precision Medicine in African populations: opportunities and challenges for the research bench to patient bedside experience." Although Africa has made great strides in biobanking and bioinformatics through major funded projects such as H3Africa[26] and H3AbioNet,[27] the capacity for wet laboratory skills and platforms to generate genetic data has remained limited, hampering progress for implementation of clinical pharmacogenetics. This is reflected by the fact that most published genomic studies involve shipping samples to developed countries for analysis. Through the formation of societies such as the TSHG, Africa is moving in the right direction to ensure timely development to support the transition from laboratory research to patient solutions.

Professor Julie Makani, pioneer of SCD research in Tanzania, presented on the genetics of SCD in Tanzania and efforts undertaken to address the most common genetic condition in the continent. Sickle cell disease is among the earliest genetic diseases discovered over 100 years ago.[28] It however still continues to pose the largest burden in SSA when compared with the rest of the world and has the lowest childhood survival rate in Africa.[29] To manage this, collaborative efforts such as SickleInAfrica[30] and SickleGenAfrica (http://sicklegenafrica.com/) have been established. Genetic counseling is among the SCD resources that can be used for other genetic conditions. In the developed countries and in India, genetic counseling has proven to reduce SCD burden. However, the formal delivery of this service is still lagging behind in many African countries. In Tanzania, for example, SCD counseling is delivered by a well-trained clinician, that is a medical doctor or nurse. With increasing demand of this service in Africa, more initiatives are being developed to address the gap. Through SickleInAfrica, there have been ongoing genetic counseling trainings in partnership with the University of Cape Town and other stakeholders. In addition, support for genetic counseling research is increasingly being provided by partners such as the NIH.

Dr. Deus Ishengoma, an expert in infectious diseases, spoke on "Human genetics and the epidemiology of infectious diseases: Building the capacity for genomics and bioinformatics in Africa." Infectious diseases have represented a major health problem, both in terms of morbidity and mortality. To build capacity for African researchers to contribute to the evolving genetic and genomic research, various initiatives have been launched. These initiatives include the program on the Developing Excellence in Leadership and Genetics Training for Malaria Elimination in SSA and the Pan-African Malaria Genetic Epidemiology Network founded by the Plasmodium Diversity Network Africa.[31] Through such programs, various scientific leaders have been trained to access and analyze genomic data from human, parasites, and vectors to

understand the key interactions between them that will significantly contribute to how we treat and subsequently eliminate malaria in the continent.

Prof. Ben Hamel, a clinical geneticist, discussed "Clinical genetic case studies in Moshi." Potential roles of case reports include recognition of new diseases or diseases which are new in a particular age of ethnic group, recognition of new features or a new mutation, detection of drug side effects, identification of a specific pathophysiology of a disease, and describing its management in resource-limited settings. Also, these reports serve as a starting point for further research and educating healthcare workers.[32] In previous studies conducted by his team, more than eight case reports have been published since 2012, four of which investigated syndromes, two skeletal dysplasias, and two neurological disorders.[33–40] The presentation highlighted the need to improve diagnostic skills and therefore the importance of reporting such rare genetic disorders.

The formation of the TSHG is timely as it can contribute to the genomic medicine agenda to improve genetic disease diagnosis, perform disease risk assessment, and improve drug safety and efficacy as well as direct therapy. The TSHG and the government at large were advised to initially invest in biobanking of well-defined disorders, for which relevant research questions can be addressed. In addition, investment in clinical oncogenetics will be of direct benefit to patients with hereditary forms of cancer such as breast, ovarian, and colorectal cancers, among many others.

Among the remarkable addresses delivered at the launching program was the patient perspective. Sharifa Mohammed Mbarak, a mother to two children with an unknown rare genetic condition, founder, and chairperson of Ali Kimara Rare Disease Foundation (AKRDF), exhibited a perspective of a patient living with a rare genetic disease. The presentation highlighted the struggles of patients and their families to find the right diagnosis, treatment, and care for people living with rare genetic conditions. The lack of services targeting individuals with rare genetic diseases in the country was highlighted. Losing a child to a rare disease stimulated the founding of AKRDF, whose vision is to become the voice of all children affected by rare genetic disease in Tanzania.

Dr. Faustine Ndugulile, the deputy Minister for Health, in his inaugural guest of honor speech, echoed the importance of forming the TSHG and assured a full support by the government of Tanzania. The vision and objectives of the society are in line with the agenda of the government of Tanzania. Therefore, the deputy minister stressed, the importance of using genetic knowledge and skills to find solutions for Tanzanians with genetic diseases.

The inaugural meeting of the TSHG demonstrated clearly that the coming together of scientists, patient groups, and policy makers in Tanzania has the potential to influence the direction and impact of genetic research and targeting specific diseases in the country. Various diseases that are genetic in origin continue to burden Africa, and it is only when these diseases are addressed in the right channels, a positive impact can be anticipated. Understanding the human genome and variation will require tremendous support from the government and the scientific world to symbiotically build capacity in populations that are currently underrepresented in basic and complex genetic studies.

CONCLUSION

Through collaboration between different stakeholders in Tanzania, AfSHG, and other collaborators, the TSHG was successfully formed and inaugurated. The existence and current activities of the TSHG as well as its potential impact at the national level in Tanzania need further encouragement from local and international communities. We envision the TSHG as an organization that will build a strong platform for scientists working in genomic and genetic issues in Tanzania. Information on the TSHG is available at http://www.tshg.or.tz/.

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