A New Age for African-Driven Genomics Research

Human Heredity and Health in Africa (H3Africa)

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The advent of the human genome sequence affords researchers the ability to further unravel the molecular mechanisms that contribute to the development of communicable and noncommunicable diseases (NCD) [1,2]. Although utilization of the human genome in both basic and clinical sciences has been a benefit to discovering causal or associated variants that underlie disease, there is an underrepresentation of ancestral diversity and admixed populations in genomic data utilized by researchers [3-6]. This poor diversity undermines the full benefit of population health improvement that can be derived from genomics research [7,8]. Although it is important to improve ancestral diversity in genomic data, it is paramount to expand and diversify all levels of the scientific workforce. To help address this issue, the National Institutes of Health and The Wellcome Trust, with assistance from the African Society of Human Genetics, launched the Human Heredity and Health in Africa (H3Africa) Consortium. The H3Africa Consortium is an internationally funded initiative dedicated to facilitating a contemporary research approach to the study of genomics and environmental determinants of common diseases, with the goal of improving the health of African populations. Over the past 5 years, the H3Africa Consortium has empowered African scientists to develop and use genetics and genomics as a platform to better understand the underpinnings of both communicable diseases and NCD on the African continent. By contributing to the development of the expertise needed among African scientists and establishing networks of African investigators, the H3Africa Consortium has demonstrated the importance of supporting African-led genomics research [9,10].

To celebrate the 5-year anniversary of this momentous program, this special issue of *Global Heart* highlights the research that has been led by African scientists who are associated with this initiative.

This special issue of *Global Heart* endeavors to advance our understanding of cardiovascular disease (CVD) and NCD that disproportionately affect African populations. The articles presented in this special issue highlight research from principal investigators from the Eighth H3Africa Consortium and the Ninth Annual African Society of Human Genetics meetings held in Dakar, Senegal, and other investigators in this area of research. The articles range from basic and biomedical sciences to infrastructure and capacity building, with a focus on improving delivery of care to populations in Africa. For example, Adeoye et al. [11] examine the growing burden of stroke by exploring the prevalence and prognoses associated with various electrocardiographic abnormalities among African acute stroke patients encountered in a multisite, cross-national, epidemiologic study. Moreover, Xu et al. [12] highlight the significant opportunities and critical challenges for extending stroke genomics research to the African ancestry population. Wonkam et al. [13] investigate the 22q11.2 deletion syndrome in patients with congenital heart defects in Cameroon, highlighting the prevalence of this microdeletion among sub-Saharan African populations. Kodaman et al. [14] describe the collection and use of exome-wide data from a Ghanaian population to test for heterogeneity in the correlation between plasminogen activator inhibitor-1 genotype and 4 CVD risk factors (body mass index, triglycerides, mean arterial pressure, and fasting glucose), with the hypothesis that loci involved in the relationship between plasminogen activator inhibitor-1 and other risk factors will also modify their correlational structure.

Although genomic research is advancing, there is a need to also include sex differences in genomic research. Gómez-Olivé et al. [15] characterize regional and male/female sex differences in the awareness of hypertension in Africa. Barfield and Boyce [16] provide a brilliant commentary articulating the importance of using sex as a biological variable in addition to ancestry when conducting genomic studies.

Some of the challenges in fully utilizing genomics in both basic and biomedical sciences involve the need for individuals with expertise in informatics, as well as the infrastructure to carry out genomic-related initiatives. From the beginning, H3Africa was designed to provide the necessary infrastructure and capacity building to address this need on the African continent. To highlight the importance of capacity building and infrastructure to perform informatics, Mulder et al. [17] describe how infrastructure support and human capacity, through H3ABioNet, have significantly contributed to the establishment of African scientific networks, data analysis facilities, and training programs. It also shows how H3ABioNet has impacted genomics and bioinformatics in Africa. In addition, Brown and Tastan Bishop [18] describe the contributions of structural bioinformatics to drug discovery, focusing particularly on the analysis of nonsynonymous single nucleotide polymorphisms.

The articles presented in this special issue of *Global Heart* highlight not only the importance of supporting African researchers, but also how their contributions are improving the field of genomics as a whole. For example,

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Ibrahim et al. [19] discuss the importance of using ancestral African genomes from large populations in east Africa to elucidate mechanisms that are believed to underlie the development of some CVD traits during the transition from traditional (i.e., hunter-gatherer societies) to modern lifestyles. Bentley and Rotimi [20] explore the mismatch between serum biomarkers and hyperlipidemia among individuals of African ancestry by investigating the presence of interethnic differences in the biological relationships underlying serum lipids. Finally, Landouré et al. [21] discuss observed neurological complications in Malian patients with sickle cell disease, the most commonly inherited Mendelian disorder in the world.

The articles that comprise this special issue of *Global Heart* include original research articles, reviews, and commentaries. Each article addresses a specific topic of importance for African or African ancestry populations. Practitioners, patients, policy makers, and researchers who seek to understand CVD and NCD in African populations will find this special issue to be a valuable resource. Furthermore, this issue will help advance the science of cardiovascular medicine, genomics, and population health for African and African ancestry populations while also celebrating the monumental contributions of H3Africa in igniting genomics research in Africa.

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