African Integrative Genomics: Implications for Human Evolution and Disease

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Africa is thought to be the ancestral homeland of all modern human populations. It is also a region of tremendous cultural, linguistic, climatic, and genetic diversity. Despite the important role that African populations have played in human history, they remain one of the most understudied and underrepresented groups in human genomics studies. The goal of this project is to characterize genomic and gene expression variation in Africans and to correlate that variation with detailed information about normal variable anthropometric traits, as well as traits that play a role in cardiovascular function, nutrition, metabolism, immune function and other traits related to adaptation to diet (bitter taste perception, lactose tolerance, glucose tolerance, and fat tolerance). We will also study the genetic history of African populations and correlations between linguistic and genetic diversity. In addition, we will characterize the gut microbiome (the collection of microbial species present in fecal matter) which can play a major role in metabolism, nutrition, and susceptibility to disease. These data will be integrated in order to identify genetic, environmental, and microbial factors that play a role in variable traits related to both health and disease. A comprehensive knowledge of patterns of variation in African genomes is critical for a deeper understanding of human genetic diversity, the identification of functionally important genetic variation, the genetic basis of adaptation to diverse environments and diets, and the origins of modern humans. Furthermore, a deeper understanding of African genomic variation will provide the necessary foundation for powerful and efficient genome-wide association and systems biology studies to identify coding and regulatory variants that play a role in phenotypic variation including disease susceptibility. By comparing populations with similar genetic ancestry but living in distinct environments (e.g. urban vs. rural, hunter-gatherers vs. pastoralists vs. agriculturalists) we will be able to distinguish both genetic and environmental factors that contribute to variability in traits of relevance both for health and disease. An additional goal is to train African scientists in the latest genomic techniques and bioinformatics analyses and to assist with building capacity for genomics research in Africa.