Genetics: Genetic variation in African populations

More than three million new genetic variants are uncovered in one of the most extensive studies of high-depth-sequenced African genomes reported to date. This study, published this week in *Nature*, provides insights into ancient migrations along the routes of Bantu-speaking populations.

Despite Africa's central role in the origin of modern humans, our knowledge of the diversity represented in African populations has been sparse. Zané Lombard and colleagues from the H3Africa Consortium address this imbalance by performing whole-genome sequencing analyses of 426 individuals, representing 50 ethnolinguistic groups, including previously unsampled populations, to explore the breadth of genomic diversity across Africa. The authors show that these newly discovered variants were found mostly among newly sampled ethnolinguistic groups. They identified new evidence for natural selection in and around 62 genes associated with viral immunity, DNA repair and metabolism. They observed complex patterns of ancestral mixing within and between populations, alongside evidence that Zambia was a likely intermediate site along the routes of expansion of Bantu-speaking populations. These findings improve the current understanding of migration across the continent, and identify responses to human disease and gene flow as strong determinants of population variation.

The authors emphasize the necessity for a broader characterization of African genomic diversity — including more individuals and from additional populations — for a more comprehensive understanding of human ancestry and to improve health research.

CONTACTS

Zané Lombard

National Health Laboratory Service, and School of Pathology, Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, SOUTH AFRICA Email: zane.lombard@wits.ac.za; Tel: +2711-489-9208

Neil Hanchard

Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, UNITED STATES OF AMERICA

Email: hanchard@bcm.edu Tel: +1713-798-0393

Adebowale Adeyemo

Center for Research on Genomics and Global Health, National Human Genome Research Institute, National Institutes of Health, Bethesda, UNITED STATES OF AMERICA

Email: adeyemoa@mail.nih.gov Tel: +1301-594-7501

The DOI number for this publication will be 10.1038/s41586-020-2859-7. Once published online at 1700 Central African Time (CAT) /1600 London time (BST) / 1200 US Eastern Time on 28 October 2020, it will be available at the following URL: https://www.nature.com/articles/s41586-020-2859-7

Acknowledgements:

The Human Heredity and Health Africa (H3Africa) Consortium is supported by the Office of Strategic Coordination (The Common Fund) of the Office of the Director, US National Institutes of Health (NIH), in partnership with The Wellcome Trust and the African Academy of Sciences (AAS)/Alliance for Accelerating Excellence in Science in Africa (AESA). WGS in H3Africa cohorts was supported by a grant from the National Human Genome Research Institute, National Institutes of Health (NIH/NHGRI) U54HG003273. The African Collaborative Center for Microbiome and Genomics Research (ACCME) is funded by NIH/NHGRI grant U54HG006947. The AWI-Gen Collaborative Centre is funded by NIH grant

U54HG006938. The Exploring Perspectives on Genomics and Sickle Cell Public Health Interventions was funded by NHGRI/NIH grant U01HG007459. The Clinical and Genetic Studies of Hereditary Neurological Disorders in Mali study was funded by the NHGRI/NIH grant U01HG007044. The Collaborative African Genomics Network (CAfGEN) is funded by the National Institute of Allergy and Infectious Diseases (NIAID) of NIH and the NHGRI of the NIH (U54AI110398). 'TrypanoGEN: an integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis', was funded by the Wellcome Trust (099310/Z/12/Z). L.R.B. was supported by the CERCA Programme/Generalitat de Catalunya and by the Spanish Ministry of Economy and Competitiveness, through the 'Severo Ochoa Programme for Centres of Excellence in R&D' 2016–2019 (SEV-2015-0533). N.M. (principal investigator), S.A., G.B., G.W., J.K., Y.J.F., T.O., O.F., E.A., S.H., G.M., M. Mbiyvanga, A.B., S.K.K., E.R.C. and A. Moussa are funded by the NIH H3ABioNet grant under award number U24HG006941.