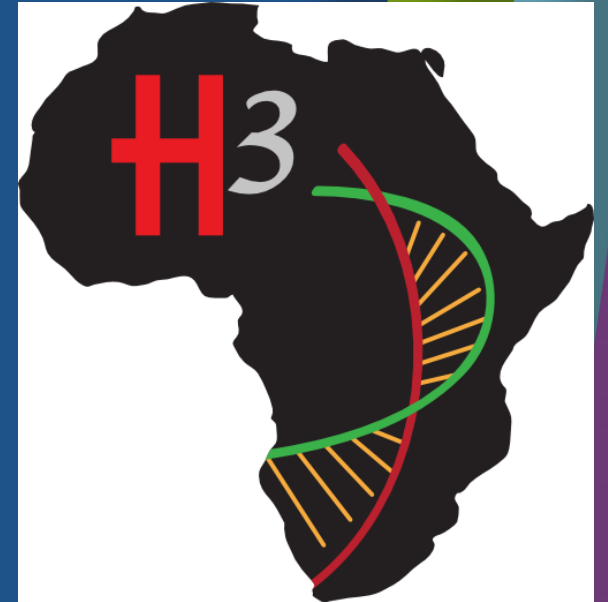


Human Heredity and Health in Africa (H3Africa)



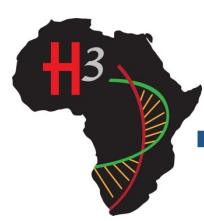
Jennifer Troyer, Program Director – NHGRI

On behalf of NHGRI program, grant, review, and administrative staff: Jane Peterson, Mark Guyer, Jeff Struewing, **Ebony Madden**, Ken Wiley, **Iman Martin**, Quynh Nguyen, Laura Skow, Gabe Batzli, Harry Wedel, Baergen Schultz, Allie Osgood, **Bri Foster**, **Jake Baroch**, Chris Darby, Diane Patterson, Jill Saletta, **Anneliese Galczynski**, **Devon Bumbray-Quarries**, **Donna Morris**, **Deanna Ingersoll**, **Kim Ferguson**, **Sean Thompson**, **Priscilla Crockett**, **Edith Dehaut**, Ken Nakamura, **Keith McKenney**, **Rudy Pozzatti** and the trans-NIH working group



National Institutes of Health

Office of Strategic Coordination – The Common Fund



Trans-NIH H3A Working Group

Chairs: Eric Green, NHGRI and Roger Glass, FIC

FIC

- Barbara Sina
- Laura Povlich

NIEHS

- Bonnie Joubert
- Kim McAllister

NCI

- Stefanie Nelson

NEI

- Ellen Liberman

NIA

- Stacy Carrington-Lawrence (OAR)
- Damali Martin (NCI)

NHLBI

- Pankaj Qsaba

NIAID

- Karen Lacourciere
- Sudha Srinivasan

NICHD

- Denise Russo

NIMH

- Tara Dutka
- Geetha Senthil

NINDS

- Richard Benson
- Stacey Chambers

NLM

- Dan Gerendasy

NIDCD

- Bracie Watson

NIDDK

- Paul Kimmel
- Afshin Parsa

OD

- Marie Nierras
- Nick Leake
- Danyelle Winchester

Wellcome Trust

- Audrey Duncanson

Science for Africa

- Rosalyn Wamuyu

Independent Experts

- **Barry Bloom - chair**
- **Kay Davies - chair**
- Rex Chisholm
- Charmaine Royal
- Ruth Chadwick
- Phillip Awadalla
- Ayo Oduola
- Solomon Nwaka
- Catherine Hoyo
- Francine Ntoumi
- Eimear Kenny
- Moffat Nyirenda
- Fred Nakwagala
- Carlos Bustamante
- Val Sheffield

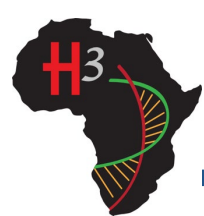


To facilitate an African-based research approach to the study of genomic and environmental determinants of common diseases with the goal of improving the health of African populations

- **Capacity development**
–human and research infrastructure
- Building **collaborative networks**

Enabling the genomic revolution in Africa



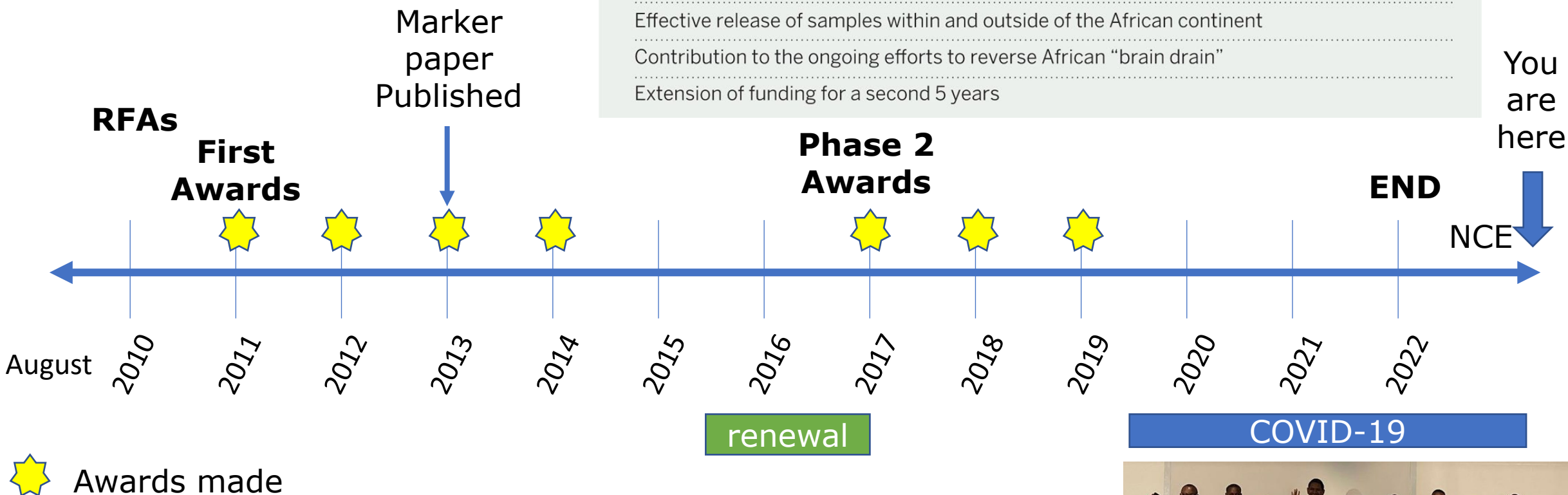


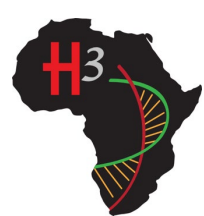
Timeline



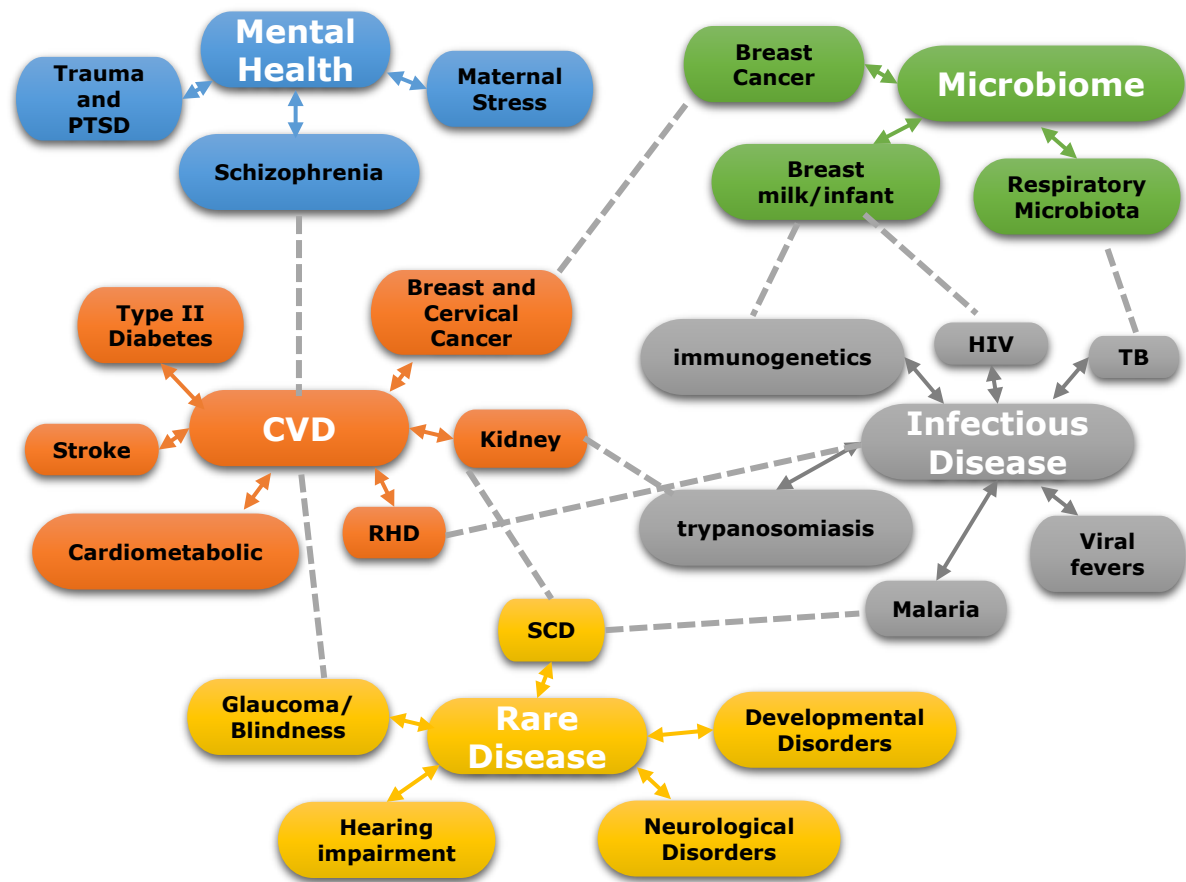
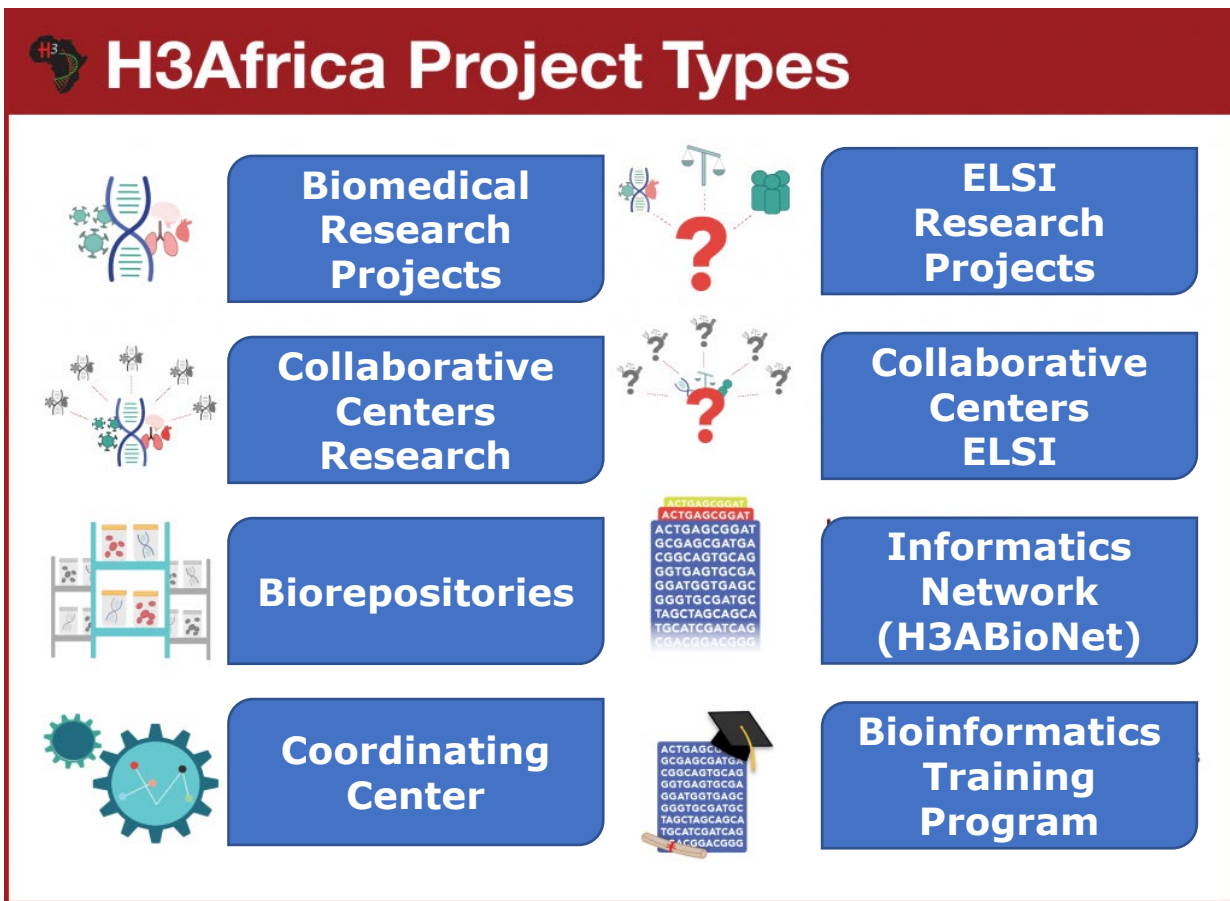
Measures of success for the 5-year H3Africa program

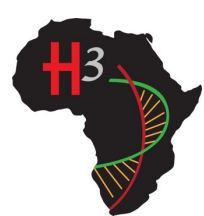
- Publication in high-impact journals with African lead and senior authors
- Increased availability of funding for biomedical and genomics research in Africa
- Effective operation of a pan-African bioinformatics network
- Regular data release
- Establishment of one or more full-scale biorepositories
- Effective release of samples within and outside of the African continent
- Contribution to the ongoing efforts to reverse African "brain drain"
- Extension of funding for a second 5 years





Projects: overview





H3Africa Consortium

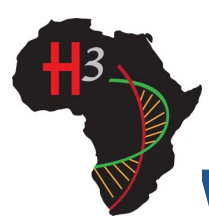


Steering Committee Working Groups

- Ethics and Community Engagement
- Data and Biospecimen Sharing
- Publications
- Biorepository
- Sustainability
- Education and Coordinated Training
- Phenotype Harmonization
- Genome Analysis
- Communication and Outreach
- *Cardiovascular Disease*
- *HIV/AIDS*
- *Mental Health*
- *Environmental Health*
- *Rare Disease*

Independent Expert Committee





WG outcomes

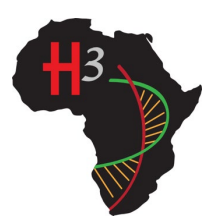


Policies and guidelines:

- H3Africa Guidelines for Community Engagement
- H3Africa Guidelines for Informed Consent
- Framework for African Genomics and Biobanking
- H3Africa Data Sharing, Access & Release Policy
- H3Africa Data and Biospecimen Access Committee Guidelines
- H3Africa Publications Policy
- H3Africa Biorepository Submission Documents
- Feedback of Findings Guidelines

Research tools and products:

- Phenotype harmonization
- Cardiovascular disease harmonized data
- Training programs and trained personnel
- Population genetics studies
- Pharmacogenomics/HLA typing/blood traits
- H3Africa genotyping array



Project outcomes

Phenotype data (associated with genotype data)

- Demographic information
- Anthropometric data
- Disease and health related phenotype data

Genomic data, human and pathogen

- Sequence data (whole genome, exome, targeted)
- Genotyping array data (H3Africa chip)
- Epigenetic data
- Transcriptomic data

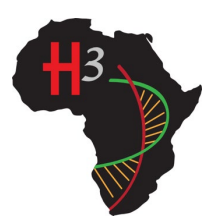
Microbiome sequence data

- Patient/sample phenotypes
- 16S rRNA
- Metagenomic data

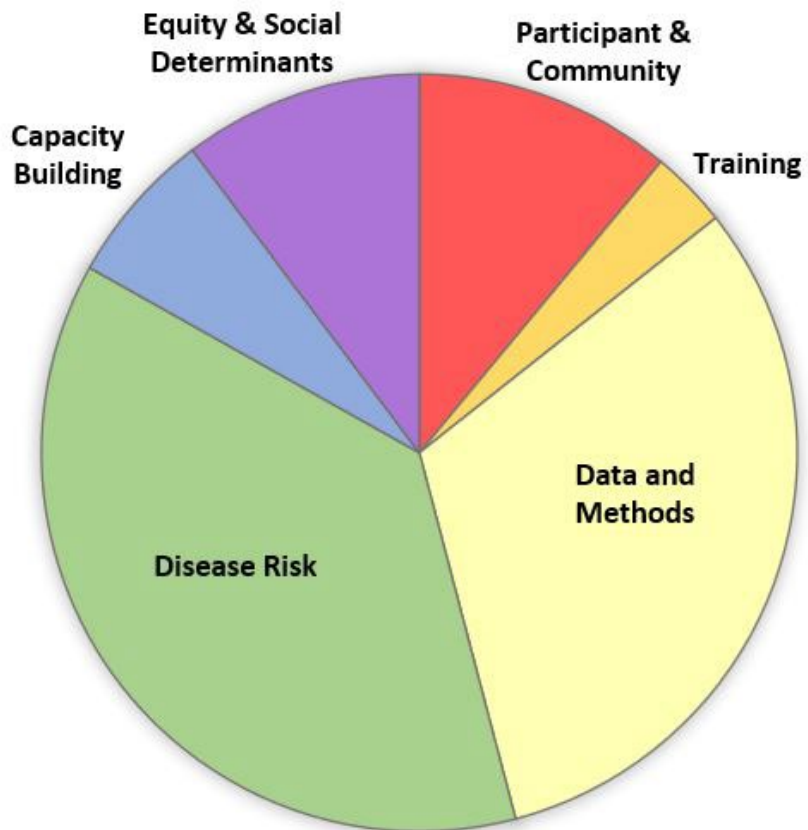
Qualitative data

- Focus groups and surveys
- Deliberative workshops

>700 Publications	434 International Presentations
51 Projects	>400 Trainees
100338 Participants recruited	>1000 Whole Exomes
50,000 Samples genotyped	26 Core Phenotypes



Publications



> [Bioinformatics](#). 2023 Jan 1;39(1):btac791. doi: 10.1093/bioinformatics/btac791.

> [J Stroke Cerebrovasc Dis](#). 2023 Mar;32(3) doi: 10.1016/j.jstrokecerebrovasdis.2023.106

Are there differences in attitudes towards disclosure of stroke? A qualitative study using SIREN-SIBS genomics study

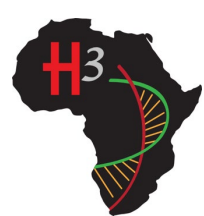
SysBioIPGWAS: simplifying post-GWAS analysis through the use of computational technologies and integration of diverse omics datasets

Oyedunni Arulogun ¹, Michelle Nichols ², Cc Onoja Akpa ⁴, Fred S Sarfo ⁵, Albert Akpalu Morenikeji Komolafe ⁹, Lukman Owolabi ¹⁰, Joshua Akinyemi ¹³, Godwin Ogbole ¹⁴, Ber Yaw Mensah ⁵, Osahon Jeffery Asowata ¹³, Arti Singh ⁵, Philip Adebayo ¹⁷, Donna Arnel Philip Ibinaieye ⁸, Wisdom Oguike ⁸, Chimdir Okechukwu Ogah ¹⁶, Rufus Akinyemi ²⁰, Br

Oluwadamilare Falola ¹, Yagoub Adam ¹, Olabode Ajayi ², Judit Kumuthini ², Suraju Adewale ¹, Abayomi Mosaku ¹, Chaimae Samtal ³, Glory Adebayo ^{1 4}, Jerry Emmanuel ^{1 5}, Milaine S S Tchamga ⁶, Udochukwu Erundu ⁷, Adebayo Nehemiah ⁷, Suraj Rasaq ⁷, Mary Ajayi ⁷, Bola Akanle ^{1 8 9}, Olaleye Oladipo ^{1 8 9}, Itunuoluwa Isewon ^{1 5 9}, Marion Adebiji ^{1 7 9}, Jelili Oyelade ^{1 5 9}, Ezekiel Adebiji ^{1 5 9 10}




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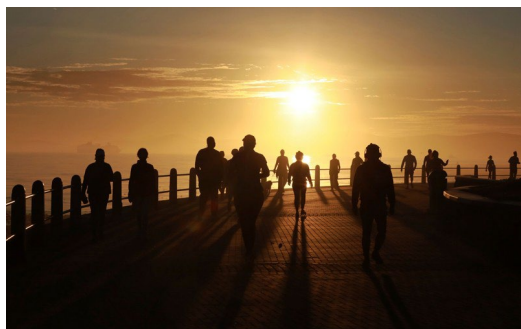
Article | [Open Access](#) | Published: 28 October 2020

High-depth African genomes inform human migration and health

Ananyo Choudhury, Shaun Aron, Laura R. Botigué, Dhriti Sengupta, Gerrit Botha, Taoufik Bensellak, Gordon Wells, Judit Kumuthini, Daniel Shriner, Yasmina J. Fakim, Anisah W. Ghoorah, Eileen Dareng, Trust Odia, Oluwadamilare Falola, Ezekiel Adebisi, Scott Hazelhurst, Gaston Mazandu, Oscar A. Nyangiri, Mamana Mbiyavanga, Alia Benkahla, Samar K. Kassim, Nicola Mulder, Sally N. Adebamowo, Emile R. Chimusa, Donna Muzny, Ginger Metcalf, Richard A. Gibbs, TrypanoGEN Research Group, Charles Rotimi, Michèle Ramsay, H3Africa Consortium, Adebowale A. Adeyemo , Zané Lombard  & Neil A. Hanchard  -Show fewer authors

Nature 586, 741–748(2020) | [Cite this article](#)

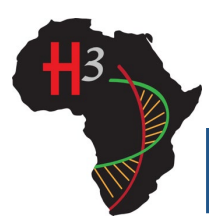
21k Accesses | 586 Altmetric | [Metrics](#)



EDITORIAL · 28 OCTOBER 2020

Africa's people must be able to write their own genomics agenda

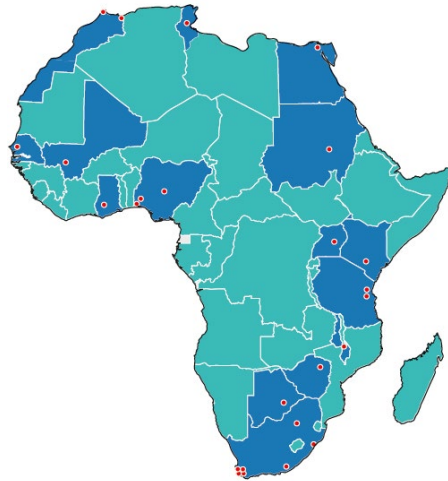
Genomics on the continent is finally getting the attention it deserves from international donors – but more funding needs to come from national and regional sources, too.



Resources – H3ABioNet

Local Expertise in:

- Genomic data analysis
- Workflow development
- Data management
- REDCap clinical databases
- Data harmonization



Local Infrastructure:

- 135TB genomic data stored
- 1.2 PB available for storage
- 80TB genomic data transferred
- 3432 cores for processing
- 18 computing facilities
- Containerized workflows

- H3Africa genotyping array
- Statistical genetics tools for ancestrally complex data
- Simulation tools
- **Population specific reference graphs**
- Genome graph tool – African pan-genome
- **SOPs and Docker containers for H3Africa pipelines**
 - Whole genome/exome NGS analysis
 - 16S rRNA diversity analysis
 - GWAS analysis
 - SNP imputation
 - *RNASeq*

Computing & network infrastructure

Data management support

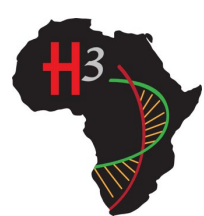
Data analysis tools and workflows

Data storage, submission and access

Training

www.h3abionet.org

H3Africa		H3Africa Data & Biospecimen Catalogue					
Human Heredity & Health in Africa							
H3Africa Chip Design Study	H3AChip	Control	EGAS00001002976	DUO:0000006	348	151	116
				DUO:0000027			
				DUO:0000024			
				DUO:0000019			
				DUO:0000021			



Resources–Repositories

H3Africa Biorepositories are ISBER compliant and play a critical role in supporting the collection and storage of African specimens that are linked to diseases relevant to African populations



Collectively developed



- LIMS and catalog of samples
- Regional distribution, training & support
- Effective, affordable, & reliable regional courier shipping routes



Institute of Human Virology Nigeria



Integrated Biorepository of H3Africa Uganda



Clinical Laboratory Services, South Africa



Alash'le Abimiku



Moses Joloba



Elizabeth Mayne



Training

New or enhanced graduate degree granting Bioinformatics Programs at:

- **Makerere University** (Kampala, Uganda) Ph.D. and M.A.
- **Pwani University** (Kilifi, Kenya) M.A.
- **Covenant University** (Ota, Nigeria) Ph.D. and M.A.
- **Kwame Nkrumah University of Science and Technology** (KNUST; Kumasi, Ghana) Ph.D. and M.A.
- **University of Sciences, Techniques, and Technologies of Bamako** (USTTB; Bamako, Mali) Ph.D. and M.A.
- **Muhimbili University of Health and Allied Sciences** (MUHAS, Tanzania) M.A.
- >100 graduate students supported in these programs

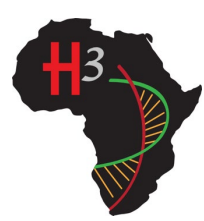


100s of graduate students, postdocs, and young investigators trained within H3Africa research projects

Thousands of people trained through Introduction to Bioinformatics Training distance learning class (H3ABioNet)

Many other courses, workshops, and webinars run by H3ABioNet, Working Groups, and the Coordinating Center

- Materials and recordings available on the H3Africa Website, H3ABioNet Website, and the H3Africa YouTube Channel



H3Africa Emerging Leaders



Simone Sterne

Immunogenetics
University of the
Witwatersrand,
South Africa

H3Africa PI: Lynn Morris



Segun Fatumo

Computational Genomics
MRC/UVRI and LSHTM
Uganda

**H3Africa PIs: Nicky
Mulder, Leon Mutesa**



Tinashe Chikowore

Genomics of CVD
Channing Division of
Network Medicine/Harvard
USA

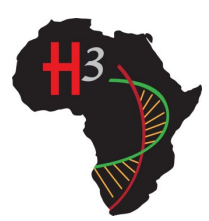
**H3Africa PIs: Michele
Ramsay and Zane Lombard**



Yosr Hamdi

Precision Oncology
Lead: Genome Tunisia
Institut Pasteur de Tunis
Tunisia

**H3Africa PIs: Nicky
Mulder, Alia Benkahla**



COVID Response

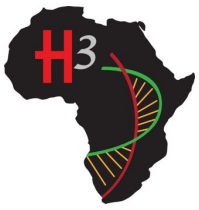
Christian Happi (Nigeria) African Center of Excellence for Genomics of Infectious Diseases (ACEGID): Sequencing of first African SARS-CoV-2 genome and sequencing of samples from + patients for the African Union/Africa CDC

Tulio de Oliveira (South Africa) Centre for Epidemic Response and Innovation: Leads the Network for Genomic Surveillance in South Africa, responsible for identification and characterization of Omicron

International and domestic response to COVID-19:

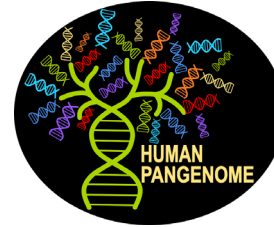
- **Biorepository PIs and staff, community engagement networks**
- **Erisa Mwaka (Uganda)**
- **Mogomotsi Matshaba (Botswana)**
- **Nicky Mulder, Lynn Morris, Ambroise Wonkam (South Africa)**
- **Seydou Doumbia (Mali)**
- **Leon Mutesa (Rwanda)**
- **Dwomoa Adu, Solomon Ofori-Acquah (Ghana)**





Collaborations

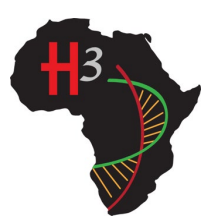
- Clinical Genome Resource (ClinGen)
- Human Pangenome Reference Consortium
- Global Alliance for Genomics and Health (GA4GH)
- International Common Disease Alliance (ICDA)
- International HundredK+ Cohorts Consortium (IHCC)
- African Society of Human Genetics (AfSHG)
- American Society for Human Genetics (ASHG)
- Regional Prospective Observational Research in Tuberculosis (RePORT)
- iHope Genetic Health (Genetic Alliance)
- African Population Cohorts Consortium (APCC)
- Undiagnosed Disease Network International (UDNI)



Harnessing Data Science for Health Discovery and Innovation in Africa (DS-I Africa)



- NIH Common Fund Program started in 2021
- Goal: to advance data science and related innovations in Africa to create an ecosystem that can begin to provide local solutions to countries' most immediate public health problems through advances in research.
- H3Africa investigators involved in DS-I Africa awards
 - Nicola Mulder, Michelle Skellton, Keymanthri Moodley, Christian Happi, Michelle Ramsay, Stephen Tollman, Ambroise Wonkam, Alash'le Abimiku, Tulio De Oliveira, Clement Ademabowo, David Katetee, Moses Jaloba, Stephen Obaro, Derrick Fauts, Scott Hazelhurst



Other Funding

NIH Funding:

- Polygenic Risk Methods in Diverse Populations (PRIMED)
1 award with 6 H3Africa cohorts
- Other NIH funding opportunities
Applications submitted to 17 different ICs, 21 awards so far



Other Funding:

- Wellcome Trust
- World Bank Africa Centers of Excellence
- Bill and Melinda Gates Foundation
- African Union/Africa CDC
- South Africa Medical Research Council (SAMRC)
- USAID



H3 Africa Looking Forward

COMMENT | 31 January 2023

Could Africa be the future for genomics research?

Funds for a major genomics programme in Africa will run dry this year. A chance to address global inequity in health-related genomics by building on the success of this initiative must not be missed.

[Zané Lombard](#) & [Guida Landouré](#)

World Renowned Geneticist and Sickle Cell Disease Expert Takes Helm of Genetic Medicine Department at Johns Hopkins

12/28/2021



Ambroise Wonkam, M.D., Ph.D., D.Med.Sc. Credit: University of Cape Town, South Africa



COMMENT | 10 February 2021

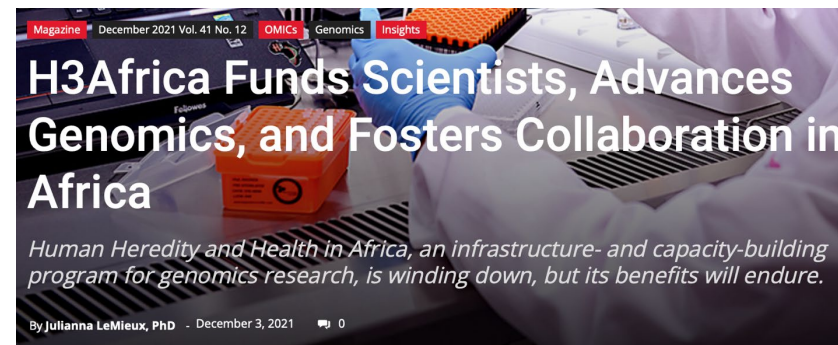
Sequence three million genomes across Africa

Capture the full scope of variation to improve health care, equity and medical research globally.

[Ambroise Wonkam](#)

The next 20 years of human genomics must be more equitable and more open

By re-committing to data sharing, researchers can fulfil the long-delayed promise of the Human Genome Project.



NEWS AND VIEWS | 10 February 2021

From one human genome to a complex tapestry of ancestry

In the 20 years since the first drafts of the human genome were made public, an explosion in genome sequencing has revealed how our evolutionary history and health can be understood by analysing the diversity in our genomes.

[Charles N. Rotimi](#) & [Adebowale A. Adeyemo](#)



[nature](#) > [nature africa](#) > [news](#) > [article](#)

NEWS | 26 February 2023

Plan for network of Genomics Centres of Excellence across Africa

Closing the gap on access to genomics technologies.

Thank you - questions



LIFT AS YOU RISE

CELEBRATING BONGANI MAYOSI'S LEGACY

