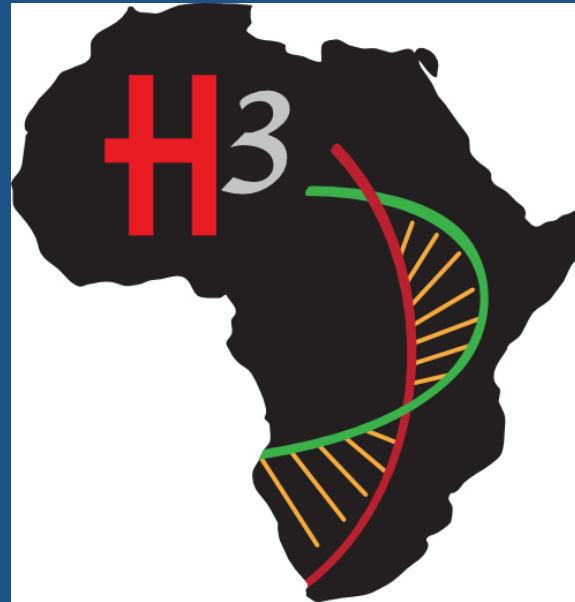


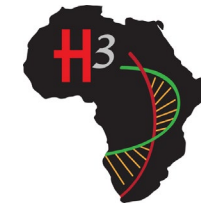
Human Heredity and Health in Africa (H3Africa)

Jennifer Troyer
Program Director - NHGRI



National Institutes of Health
Office of Strategic Coordination–The Common Fund

Working Group



FIC

- Barbara Sina
- Laura Povlich

NCI

- Stefanie Nelson

NEI

- Ellen Liberman

NHGRI

- Ebony Madden
- Allie Osgood
- Baergen Schultz
- Jennifer Troyer
- Ken Wiley

NHLBI

- Pankaj Qasba

NIAID

- Karen Lacourciere
- Sudha Srinivasan

NICHD

- Denise Russo

NIDCD

- Bracie Watson

NIDDK

- Paul Kimmel
- Afshin Parsa

NIEHS

- Bonnie Joubert
- Kim McAllister

NIMH

- Lora Bingaman
- Tara Dutka
- Geetha Senthil

NINDS

- Richard Benson
- Stacey Chambers

NLM

- Dan Gerendasy

OAR

- Stacy Carrington-Lawrence

OD

- Nicholas Leake
- Marie Nierras
- Danyelle Winchester

ORWH

- Rajeev Agarwal



NIH WG chairs

- Eric Green
- Roger Glass

IEC chairs

- Barry Bloom
- Kay Davies

Wellcome Trust

- Audrey Duncanson

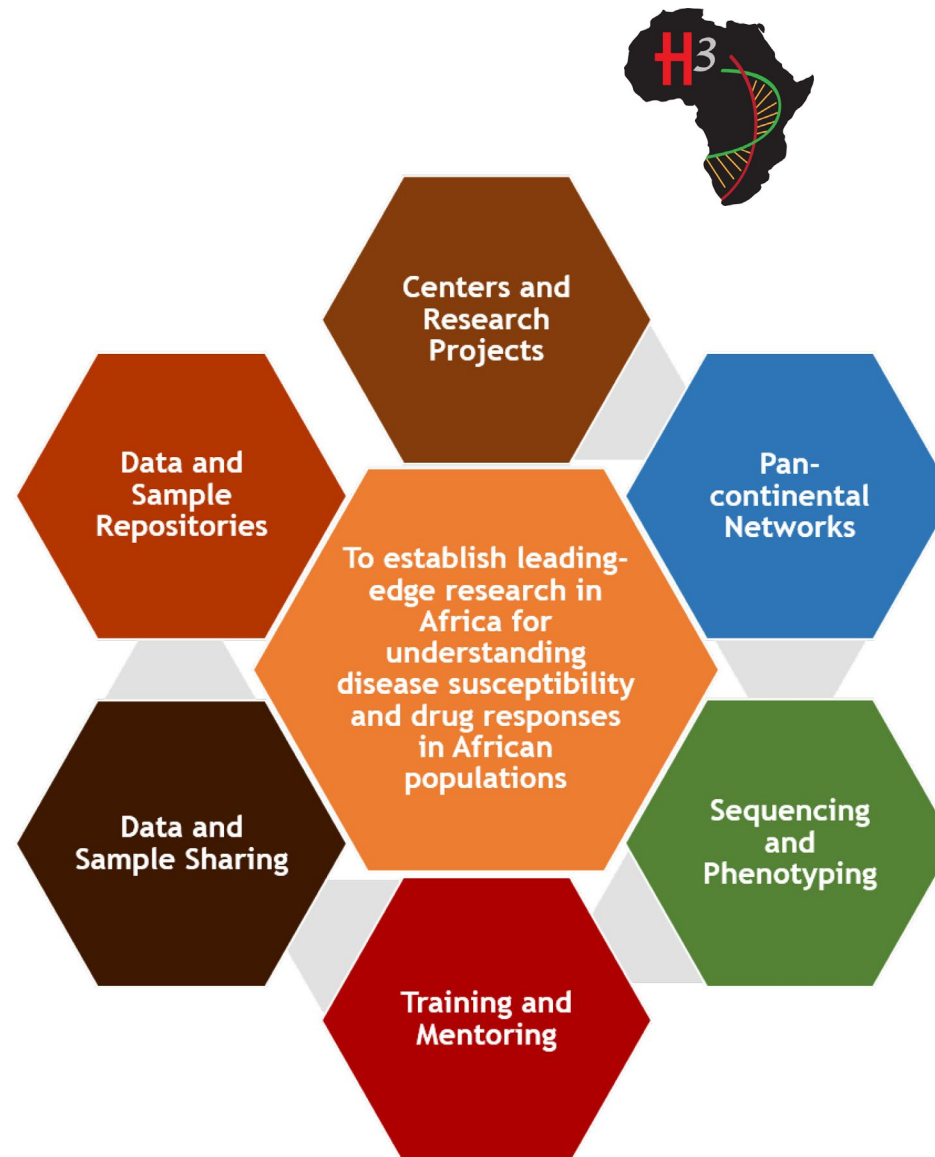
Science for Africa

- Jenniffer Maroa

Goal

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

H3Africa is developing capacity for health-related genomics research in Africa

By The H3Africa Consortium*†

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

RFA

August 2010



Awar

You
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END

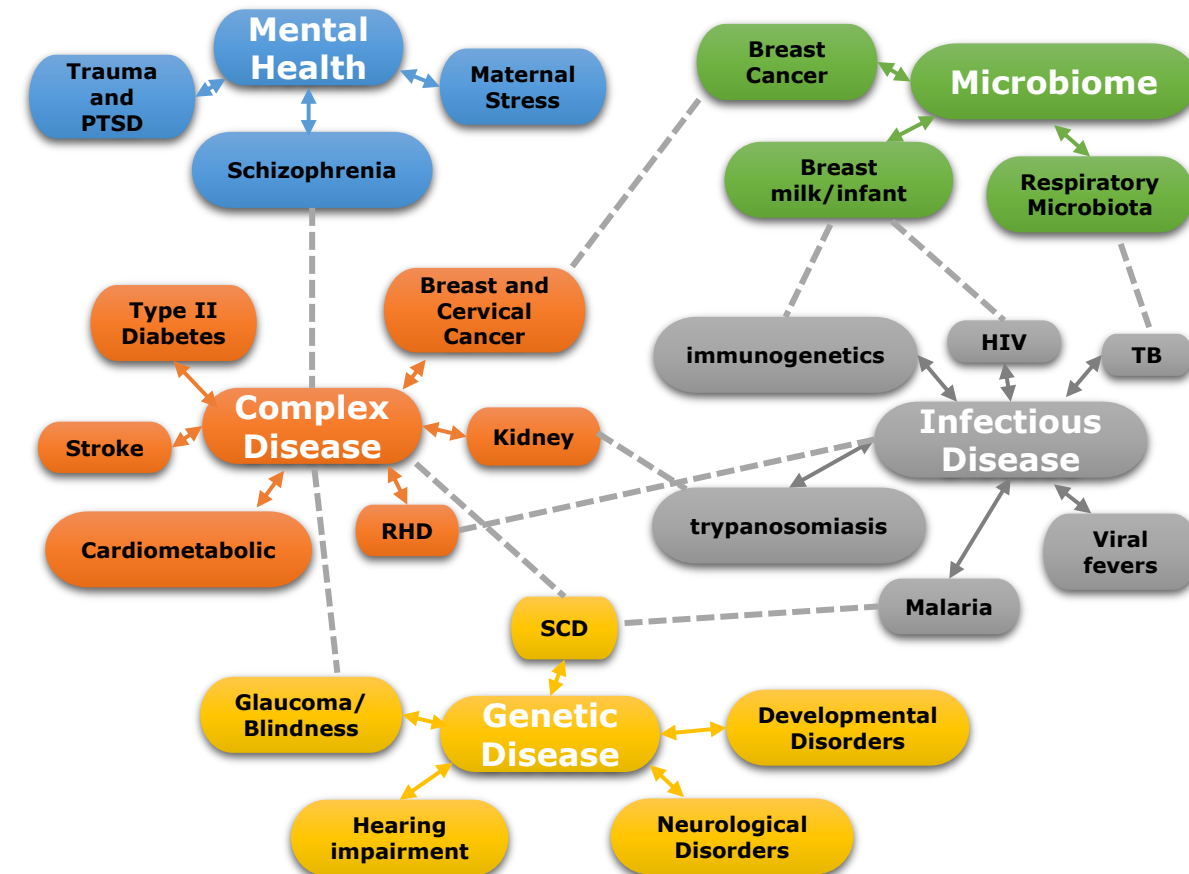
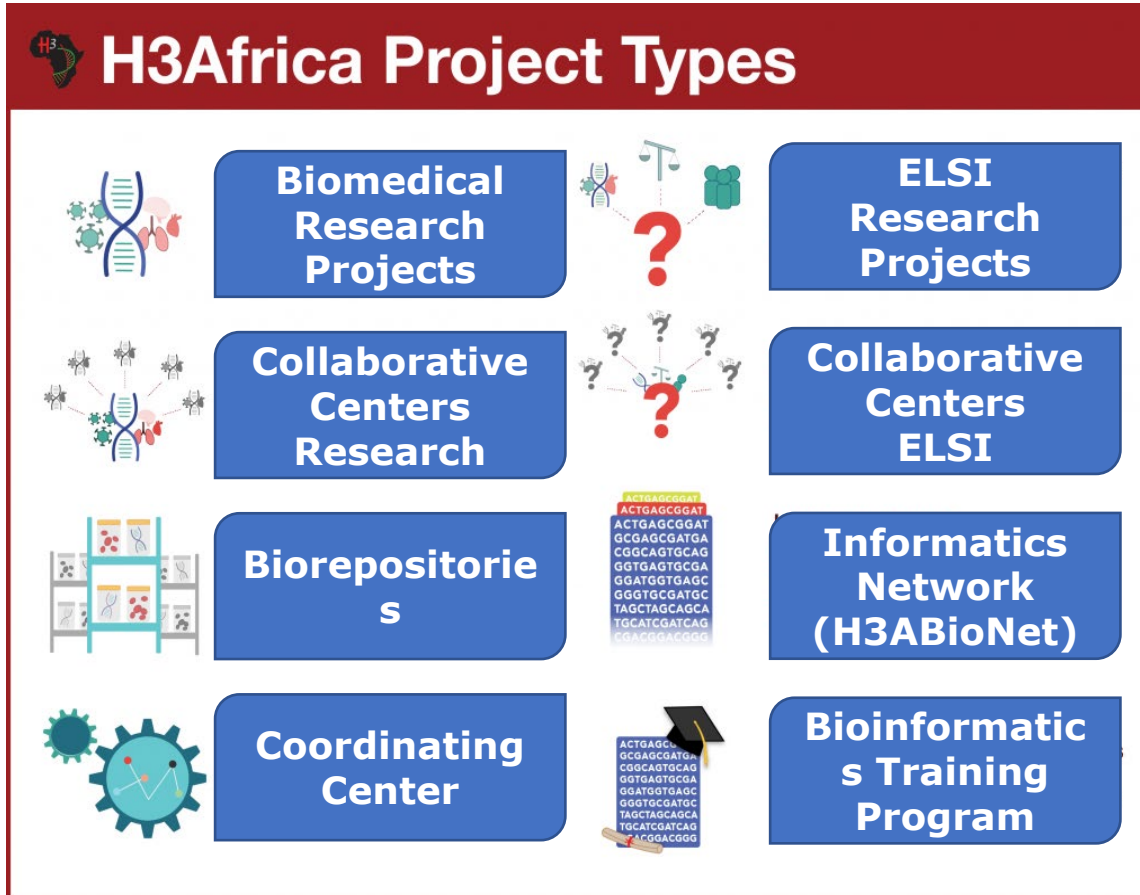
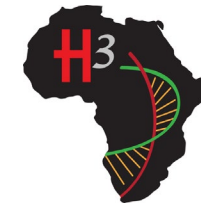
NCE

2022

9



Projects: overview



H3Africa Consortium



Steering Committee – PIs and funders

Working Groups – Project members and funders

- 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



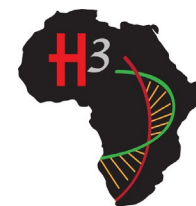
Policies and guidelines:

- 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

Research tools and products:

- Phenotype harmonization
- Cardiovascular disease harmonized data
- Training programs and trained personnel
- Population genetics studies
- ADME study
- 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
- Non-human 16S rRNA sequence data for microbiome
- Non-human full genome sequence data for microbiome

Qualitative data

- Focus groups and surveys
- Deliberative workshops

51 projects	>400 Trainees
100338 Participants recruited	>1000 Whole Exomes
50,000 samples genotyped	26 Core Phenotypes

Project outcomes



New neural network classification method for individuals ancestry prediction from SNPs data.

Soumare H, Rezgui S,
BioData Min. 2021 Jun
PMID: 34183066

Eyes of Africa: The Genetics of Blindness: Study Design and Methodology.

Olawoye O, Chuka-Okosa C, Akpa O, Realini T, Hauser M, Ashaye A.

BMC Ophthalmol. 2021 Jul 9;21(1):272. doi: 10.1186/s12886-021-02029-8.

PMID: 34242750 Free BMC article

Explanatory models for the cause of Fragile X Syndrome in rural Cameroon.

Kengne Kamga K, De Vries J, Nguifack S, Munung NS, Wenker A

J Genet Couns. 2021 Jun
PMID: 34145661

Prevalence and socio-demographic correlates of tobacco and alcohol use in four sub-Saharan African countries: a cross-sectional study of middle-aged adults.

Boua PR, Soo CC, Debpuur C, Maposa I, Nkoana S, Mohamed SF, Choma S, Oduro A, Asiki G, Micklesfield LK, Gómez-Olivé FX, Sorgho H, Mall S, Ramsay M: as members of AWI-Gen and the H3Africa Consortium.

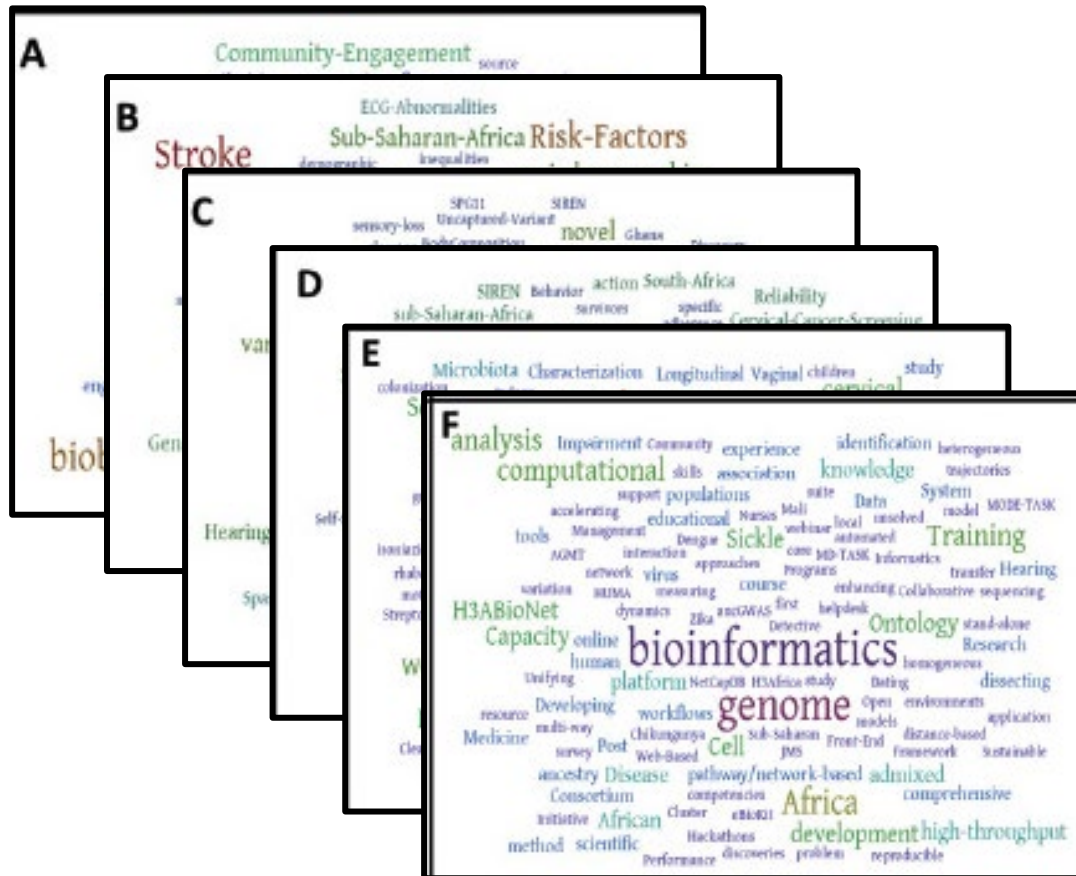
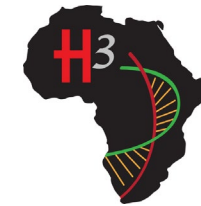
The role of causal knowledge in stigma considerations in African genomics research: Views of South African Xhosa people.

Matshabane OP, Campbell MM, Faure MC, Appelbaum PS, Marshall PA, Stein DJ, de Vries J.

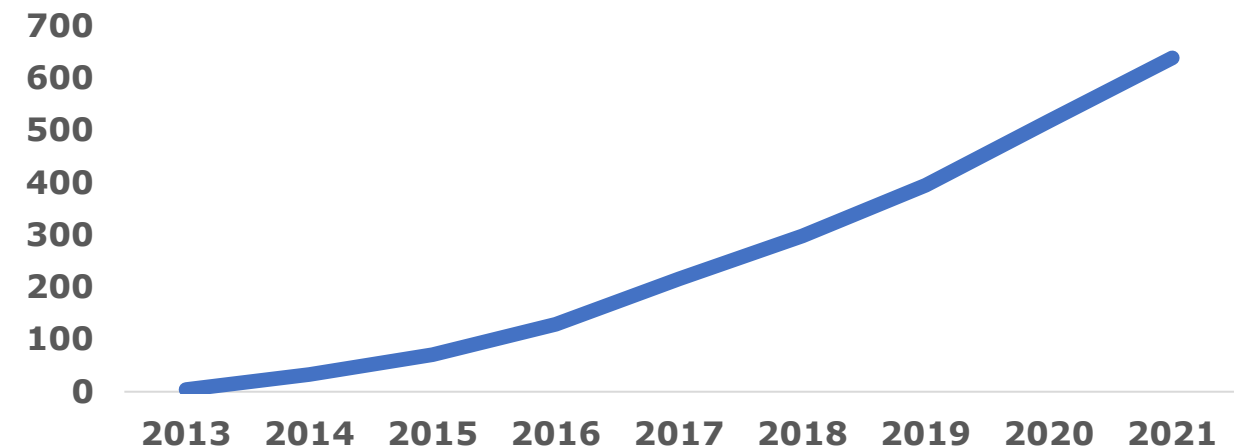
Soc Sci Med. 2021 May;277:113902. doi: 10.1016/j.socscimed.2021.113902. Epub 2021 Apr 7.

PMID: 33865094 Free article.




Publications



Cumulative H3Africa Publications

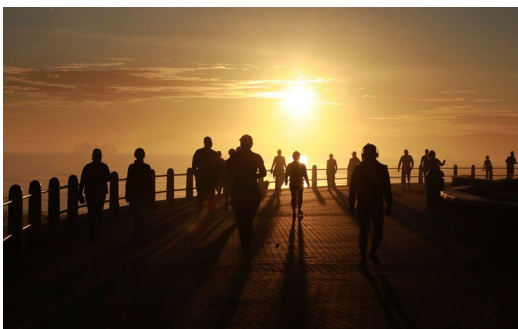


High-depth African genomes inform human migration and health

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

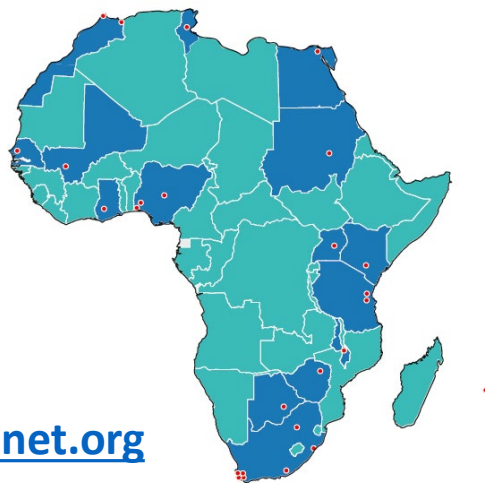
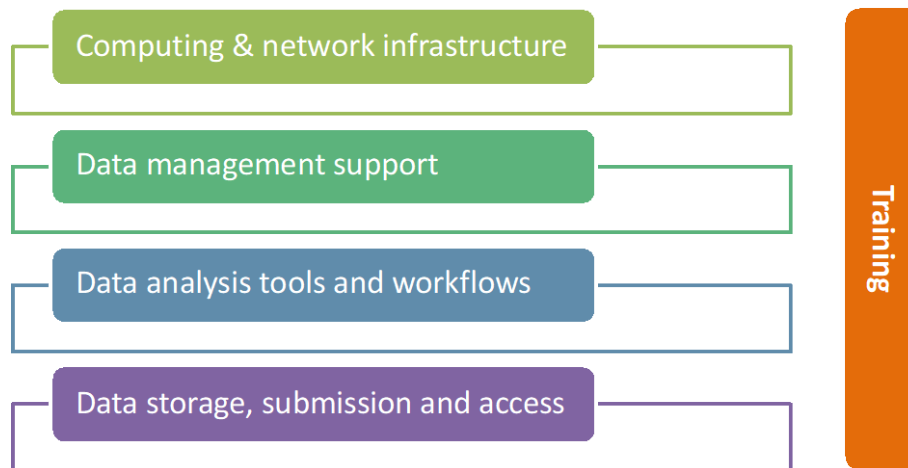
Africa's people must be able to write their own genomics agenda

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

- H3Africa genotyping array
- Data analysis and interpretation tools
 - HUMA
 - Genesis
- Machine learning applications
- Data integration and meta-analysis tools
- Statistical genetics tools for complex data
- Simulation tools
- Population specific reference graphs
- Bacterial pan genomes and GWAS
- Genome Detective –virus detection from NGS data
- 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



www.h3abionet.org

Developed expertise in:

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

Developed infrastructure:

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

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



Institute of Human Virology Nigeria



Project Lead
Dr. Alash'le Abimiku
aabimiku@ihvnigeria.org



Integrated Biorepository of H3Africa Uganda



Project Lead
Dr. Moses Joloba
mlj10@case.edu



Clinical Laboratory Services, South Africa



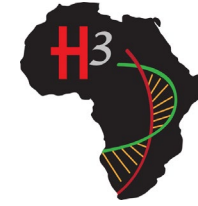
Project Lead
Dr. Elizabeth Mayne
elizabeth.mayne@NHLS.AC.ZA

Collectively developed

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



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

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 African Union

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:

- **Moses Joloba, Erisa Mwaka (Uganda)**
- **Mogomotsi Matshaba (Botswana)**
- **Nicky Mulder, Lynn Morris, Ambroise Wonkam, Elizabeth Mayne (South Africa)**
- **Seydou Doumbia (Mali)**
- **Leon Mutesa (Rwanda)**
- **Dwomoa Adu, Solomon Ofori-Acquah (Ghana)**

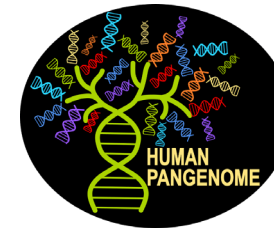


New Collaborations

- H3Africa Rare Disease Working Group and ClinGen
- Human Pangenome Reference Consortium
- Global Alliance for Genomics and Health (GA4GH)
- International Common Disease Alliance (ICDA)
- International HundredK+ Cohorts Consortium (IHCC)
- American Society for Human Genetics (ASHG)
- NHGRI DNA Day Events
- iHope Genetic Health (Genetic Alliance)



Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.



Looking Forward



NIH Funding:

- Harnessing Data Science for Health Discovery and Innovation in Africa (DS-I Africa)
 - 10 awards with 17 H3Africa Co-Is
- Polygenic Risk Methods in Diverse Populations (PRIMED)
 - 1 award with 6 H3Africa cohorts
- Other NIH funding opportunities
 - Multiple applications submitted to 17 different ICs, 10 awards so far



PRIMED
consortium

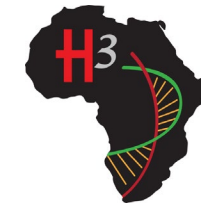
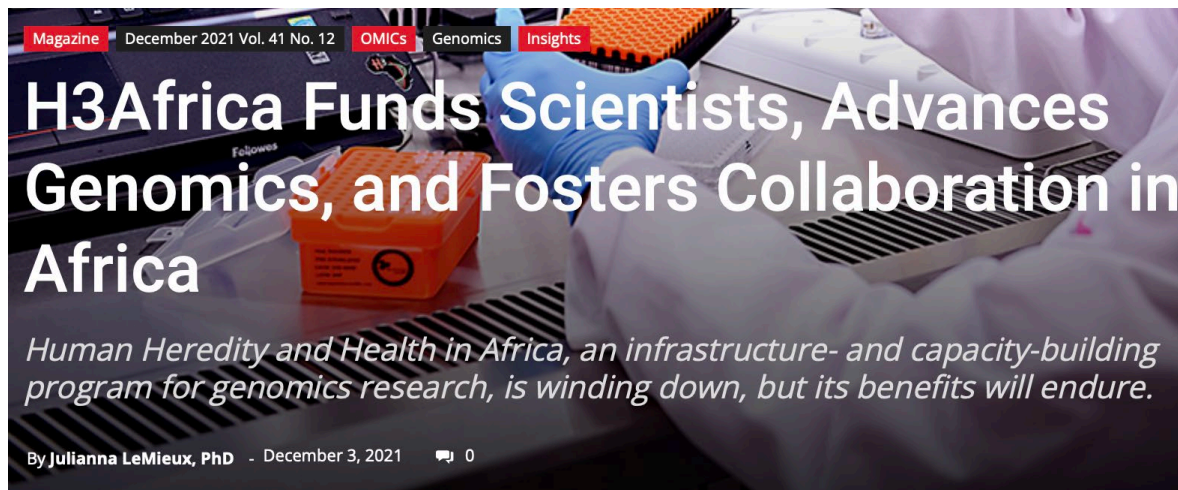


Other Funding:

- Wellcome Trust - African Population Cohort Consortium (APCC)
- International Common Disease Alliance (ICDA)
- World Bank Africa Centers of Excellence
- Bill and Melinda Gates Foundation



BILL & MELINDA
GATES *foundation*



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

12/28/2021



Ambrose 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.

[Ambrose Wonkam](#)

EDITORIAL | 10 February 2021

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

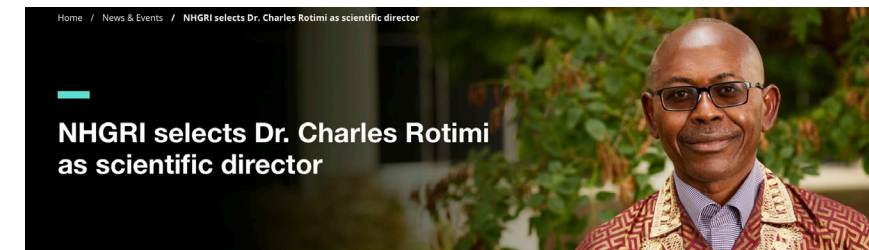
By re-committing to data sharing, researchers can fulfil the long-delayed promise of the Human Genome Project.

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](#) & [Adebawale A. Adeyemo](#)



Thank you - questions



LIFT AS YOU RISE

CELEBRATING BONGANI MAYOSI'S LEGACY

