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19th H3Africa Meeting Fellows Agenda

H3Africa Fellows Professional Development 29 May -1 June 2022 Transcorp Hilton Hotel Abuja, Nigeria	
DAY 1: 29 May	
INTRODUCTION TO SINGLE CELL GENOMICS USING R Bauchi Meeting Hall Trainer(s): Collins M. Morang'a – Bioinformatician (WACCBIP); Vincent Appiah – HPC Manager (WACCBIP); Dr Lucas Amenga-Etego – Senior Research Fellow (WACCBIP)	GENOMIC EPIDEMIOLOGY Ogun/Nassarawa Meeting Hall Trainer: Anita Ghansah
08:30	Bus departs to Transcorp Hilton
09:00 – 09:30	Arrival and Registration
09:30 – 09:45	Welcome address
09:45 – 10:30	Presentation: Steps involved in processing raw scRNA-seq data
10:30 – 10:45	Practical: Login and familiarize with RStudio Server
10:45 – 11:00	TEA/COFFEE BREAK
11:00 – 13:30	Practical: QC of Single cell data – Visualization
	Practical: SCT transformation and data integration steps
	Practical: PCA and Cluster Visualizations - UMAP
13:30– 14:15	LUNCH
14:15 – 15:00	Article discussion : Approaches used in cell identification or annotation
15:00 – 17:30	Practical: Cell annotation - Manual and reference guided
	Practical: Differential gene expression – DESeq2
17:30	Closing remarks and announcements
18:30	FELLOWS DINNER
20:00	Bus departs to Hawthorn Suites
DAY 2: 30 May NATURE MASTERCLASS WEBINARS	
Plateau meeting hall Facilitators: Nature Portfolio editors, Rolanda Julius	
07:30	Bus departs to Transcorp Hilton
08:00 – 08:15	Arrival and registration
08:15 – 08:30	Welcoming and introductions
08:30 – 12:00	Webinar 1
12:00 – 13:00	LUNCH
13:00 – 15:30	Webinar 2
	Announcements
Lagos Meeting Hall	
15:30 – 18:45	Plenary sessions

20:00 – 22:00	WELCOME RECEPTION (FOUNTAIN AREA)	
22:00	Bus departs to Hawthorn Suites	
DAY 3: 31 May PI PRESENTATIONS; FELLOWS SPEED TALKS; NATURE MASTERCLASS ABSTRACT REVIEWS		
Lagos meeting hall		
Chair: Barbara Sina		
08:00	Bus departs to Transcorp Hilton	
09:30 – 12:00	One-on-One with Nature Portfolio Editors	Kaduna Boardroom
12:00 – 13:30	Plenary sessions	
13:30 – 14:30	LUNCH	
Fellows Speed Presentations Group 1 (45 min)		
14:30 – 14:33	David Twesigomwe	Characterisation of pharmacogene allelic variation in African populations and development of a novel diplotype calling algorithm.
14:33 – 14:36	Nadja Louw	The role of copy number variants in the aetiology of developmental disorders in South Africa - a whole exome sequencing study
14:36 – 14:39	Osahon Asowata	Risk scoring model for predicting Hypertension among indigenous Africans: Findings from the SIREN study
14:39 – 14:42	Oumou Traore	De novo variant in PAX3 gene in a Malian family with Waardenburg syndrome type 1
14:42 – 14:45	Ousmane Doumbia	Study of the Nuclear Organization of Telomeres in Urinary Circulating Cells in Bladder Cancer
14:45 – 14:48	Raylton Chikwati	Menopause, HIV and Cardiometabolic Disease Risk: A Cross-Sectional Analysis on Midlife Women from Kenya and South Africa
14:48 – 14:51	Ruth Nanjala	Evaluating the accuracy of genotype imputation in the Major Histocompatibility Complex (MHC) region in selected African populations
14:51 – 14:54	Shahiid Kiyaga	Comparative genomics, antibiotic resistance and virulence determinants of clinical Pseudomonas aeruginosa strains in Kenya
14:54 – 14:57	Shola Able-Thomas	Participant withdrawals from an observational longitudinal study of the respiratory microbiota of Gambian children during a pandemic

14:57 – 15:00	Wisdom Akurugu	Identification of HPAS-associated SNPs in children on corticosteroid treatment
15:00 – 15:15	Q & A	
15:15 – 17:30	Plenary sessions	
17:30	COFFEE/TEA BREAK	
19:00	DINNER	
22:00	Bus departs to Hawthorn Suites	
DAY 4: 1 June PI PRESENTATIONS; FELLOW SPEED TALKS; NATURE MASTERCLASS ABSTRACT REVIEWS		
Lagos meeting hall		
Chair: Alash'le Abimiku		
08:00	Bus departs to Transcorp Hilton	
09:30 – 12:00	One-on-One with Nature Portfolio Editors	Kaduna Boardroom
12:00 – 13:00	Plenary sessions	
13:00 – 14:00	LUNCH	
14:00 – 14:15	Plenary Talk: Thomas Kariuki	Science for Africa Foundation
Fellows Speed Presentations Group 2 (40 min)		
14:15 – 14:18	Grace Adjo Larbie Gafa	What do research participants want and what should they be given? Feedback of Genetics Research Findings in the H3Africa Kidney Disease Research Network
14:18 – 14:21	Harriet Nankya	Stakeholders’ perspectives on community engagement for genomics research in Africa – a literature review
14:21 – 14:24	Henrietta Ifechukwude Monye	Attitudes towards genetic testing and the associated factors among adult eye clinic patients in Ibadan, Nigeria
14:24 – 14:27	Isaac Emmanuel Omara	Genetic Diversity of Bundibugyo ebolavirus from Uganda and the Democratic Republic of Congo
14:27 – 14:30	Mamadou Sangare	Structural characterization of phosphatidylserine decarboxylase a promising therapeutic target and prediction of its potential inhibitors
14:30 – 14:33	Maria Magdalene Namaganda	Impact of next-generation sequencing (NGS) on HIV-1 drug resistance testing among patients experiencing virological failure at the time of therapy switching in Uganda
14:33 – 14:36	Melaku Tilahun	Popular perception on genomic studies and hereditary diseases in Ari Community, South Omo Zone, Ethiopia
14:36 – 14:39	Modibo K Goita	Profile of individual telomere length of a rare chromosomal

		abnormality in Chronic Myeloid Leukemia in Mali.
14:39 – 14:42	Nana Yaa Achiaa Karikari Agyemang	Apolipoprotein L1 variants and Preeclampsia
14:42 – 14:45	Nicholas Opoku	Comprehensive Benchmarking of Copy Number Variants Detection Tools Using Whole-Exome Sequencing
14:45 – 14:55	Q & A	
14:55 – 15:20	Plenary talk: Grace Ajuwon	Tools for Enhancing Visibility of Your Research and Citations
Fellows Speed Presentations Group 3 (40 min)		
15:20 – 15:23	Alfred Ssekagiri	QuasiFlow: A Nextflow Pipeline for Analysis of NGS-based HIV-1 Drug Resistance Data
15:23 – 15:26	Andrews Frimpong Adu	Benchmarking of Long-read Assembly Tools for Analyzing African Genomes
15:26 – 15:29	Carlyne Fiona Tchouaket Ngueyi	Impact of COVID-19 on the Sputum Conversion Rate and Adherence of Tuberculosis patients to Directly Observed Treatment at some Diagnosis and Treatment Centres of the Littoral and South West Regions of Cameroon within 2019 and 2021: A hospital-based retrospective study
15:29 – 15:32	Cheick Abdel Kader Cisse	Clinical and genetic aspects of osteogenesis imperfecta in a Malian family
15:32 – 15:35	Diana Gladys Kolieghu Tcheumeni	Prevalence of advanced HIV disease in people living with HIV on antiretroviral therapy at the Buea Regional Hospital and co-infection rates of Mycobacterium spp. and Cryptococcus spp.
15:35 – 15:38	Edwin Moses Appiah	Gastric microbiome predictive model for gastric carcinogenesis
15:38 – 15:41	Elvis Twumasi Aboagye	Novel MARVELD2 Variant Associated with Post-lingual Autosomal Recessive Non-Syndromic Hearing Impairment in Consanguineous Ghanaian Family
15:41 – 15:44	Ester Acen	Vitamin D binding protein gene polymorphism and free serum bioavailability among tuberculosis patients and household contacts
15:44 – 15:47	Fatoumata Gnine Fofana	Docking of human band 3 anion transporter proteins with their Plasmodium falciparum

		interactors based on short linear motifs
15:47 – 15:50	Ntui Vincent Ntui-Njock	Current status of molecular markers of Plasmodium falciparum resistance to Sulphadoxine-Primethamine (SP) and the effectiveness of IPTp-SP among pregnant women in Mutengene, Southwestern, Cameroon.
15:50 – 16:00	Q & A	
16:00 – 16:30	TEA/COFFEE BREAK	
Sustainability Session		
Chairs: Alash'le Abimiku & Dwomoa Adu		
16:00 — 18:30	Plenary sessions	
Achievements, awards,dedication and closing		
Chair: Ambroise Wonkam, Zane Lombard & Michelle Skelton		
18:30 — 19:00	Plenary sessions	
19:30	DINNER (Pool BBQ)	
22:00	Bus departs to Hawthorn Suites	
DAY 5: 2 June EXCURSIONS (Optional), Bus departs Transcorp Hilton Departures		

Workshops

Introduction to single cell genomics using R computational workshop

Introduction

A cell represents the basic unit of biology and contains a blueprint of the genome. This blueprint is normally transcribed to produce RNA transcripts, often referred to the transcriptome that determines/reflects the cell type and ultimately its function. The transcriptome of single cells is almost always lost in studies that conduct bulk or population level analysis of RNA from individuals. Thus, deeper insights into the biological functions of individual cells at tissue/organ/organism level can be gained from detailed understanding of genomic, epigenomic and transcriptional variation at single cell level. In the past 5-10 years there has been a rapid technological advancement in single cell sequencing and computational methods/technologies that permit detailed analysis of RNA-Sequencing data at single cell level (Linnarsson & Teichmann, 2016; Macaulay & Voet, 2014). This workshop seeks to introduce fellows to the computational approaches used to analyse single cell data in R.

Course Description

In this course we will focus on the introduction to single cell genomics and will instruct participants on how to process and analyse single cell data. The computation part of the workshop will focus on using Seurat (Hao et al., 2021) package in R/RStudio. A working knowledge in R is required for this course, but R server accounts with data and pre-installed packages will be provided during the course. Additionally, the fellows will be expected to spend 2 -3 hours prior to the course to go through the course reference article.

Learning Objectives

- Understand the steps involved in processing raw single cell sequencing reads.
- Compute QC metrics of scRNA-Seq data and assess the data quality.
- Perform transformation and integration of the scRNA-Seq data.
- Compute PCA, Manifold Learning and explore visualization of single cell data.
- Understand the approaches for Cell Identification & Annotation.
- Perform differential gene expression in various cell types.

Target Audience:

- H3A Fellows (Number of participants is not limited)
- Fellow with basic working knowledge in R and RStudio.

Certificates

After completing the workshop, participants will receive a certificate.

Workshop Requirements

- Attendee own laptop
- Room set up with projector and screen
- Good internet connection
- Article for Discussion (Mattia Forcato, Oriana Romano, and Silvio Bicciato. Computational methods for the integrative analysis of single-cell data, Briefings in Bioinformatics, Volume 22, Issue 3, May 2021, bbaa042, <https://doi.org/10.1093/bib/bbaa042>)

Genomic Epidemiology

- A brief review of basic concepts in human genetics and a summary of recent advances in genomic technology, with an emphasis on their relevance to genetic epidemiology.

- Concepts and topics related to single genes: Mendelian inheritance, modes of inheritance, a description of the characteristics of complex genetic traits, the Hardy–Weinberg principle, the genetic code, basic gene structure, types of genetic markers including single nucleotide polymorphisms (SNPs) and copy number variants (CNVs), and the concept of identity by descent (IBD).
- Concepts and topics related to multiple genes: genetic linkage, recombination, linkage disequilibrium, haplotypes, and tag SNPs.
- The overview of developing genomic technology describes genome-wide SNP genotyping platforms, Sanger and next-generation (massively parallel) DNA sequencing methods, and exome sequencing.
- Different study designs for investigating common and rare genetic variants are summarized.

Nature Masterclass

Workshop Description

The Nature Masterclasses virtual workshop in scientific writing and publishing is designed to offer accessible training for busy, early- to mid-career researchers in the natural sciences. Through a combination of interactive group webinars, pre-workshop activities and an individual Abstract Review session for each participant, all led by Nature Portfolio editors with relevant scientific expertise, the workshop will help participants to effectively communicate their research and provide them with a unique opportunity to discuss their scientific writing with an editor. Participants register directly for the workshop in order to receive a unique link to join the workshop. This link will be sent in the registration confirmation email.

Handout Materials

Each participant will receive a Participant Guide after they have registered for the workshop which contains the workshop agenda, trainer biographies and pre-workshop activities. They will also receive a Resources booklet containing some background information on the topics covered in the webinars which they will be able to download during and after the webinars. Participants will be able to access recordings of the webinars for 30 days after the sessions have taken place and will also receive a watermarked copy of the slide presentations which will be accessible during and after the webinars.

Certificates

After completing the workshop, participants will receive a Nature Masterclasses Certificate of Training, signed by Dr Magdalena Skipper, Editor-in-Chief, Nature.

Abstract Review

Each participant will be allocated an individual Abstract Review session with a Nature Portfolio editor. During the session, the editor will talk through their advice for an unpublished abstract, of up to 250 words, which has been submitted on behalf of the participant before the workshop. The session will last about 8 minutes, and will take place at a fixed time during the 2.5-hour pre-scheduled Abstract Review. Registered participants will be contacted individually by e-mail to confirm the time of their session, and how to access it, a few days before it is due to take place. Participants must ensure that they are available for the entire 2.5-hour time window until they receive this email. Unfortunately, the Nature Masterclasses team will be unable to rearrange the individual sessions if participants do not attend at the allocated time. Please note that the editor will be unable to comment on the suitability of abstracts for publication in specific journals. Please also be aware that the Nature Masterclasses team cannot guarantee that the editor will review participants' abstracts before these individual sessions, although the editor will do their best to do so.

Workshop Requirements

- Participants will be organized in groups of six individuals.
- Five workstations each with a provided laptop.
- Participants each to use their own smartphone
- Participants using their smartphones to download *go-to-training* app. See QR codes in this booklet
- It is recommended that one display the Participant Guide on a separate device during the webinars.
- Room set up with projector, central screen and hybrid audio/visual system as the trainers will be joining online.
- Good internet connection
- Participants need to complete the required pre-workshop activities

More information available (see QR codes in this booklet)

Fellows Abstracts & Biographies

SPEED TALK

QuasiFlow: A Nextflow Pipeline for Analysis of NGS-based HIV-1 Drug Resistance Data

Alfred Ssekagiri* (1,2), **Daudi Jjingo** (3,4), **Ibra Lujumba** (2), **Nicholas Bbosa** (5), **Daniel Lule Bugembe** (5), **David Patrick Kateete** (2), **Irving King Jordan** (6), **Pontiano Kaleebu** (1,5), **Deogratius Ssemwanga** (1,5)

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(5) Medical Research Council (MRC)/Uganda Virus Research Institute (UVRI) and London School of Hygiene and Tropical Medicine (LSHTM) Uganda Research Unit, Entebbe, Uganda. (6) School of Biological Sciences, Georgia Institute of Technology, Atlanta, Georgia, USA.

INTRODUCTION: Next Generation Sequencing (NGS) technologies enable reliable detection of minority HIV-1 drug-resistant variants (MDRVs). MDRV have been associated with an increased risk of virological failure in patients on Non-Nucleoside Reverse Transcriptase Inhibitor based regimens. However, the scarcity of computational skills to effectively and expeditiously identify minority HIV-1 drug resistant variants from NGS-based HIV drug resistance (HIVDR) testing data remains a challenge in resource-limited settings.

METHODS: The pipeline takes paired-end short reads from Illumina platforms as input. The quality of the reads is checked using FastQC which generates a report for each of the input files. FastQC results are aggregated into a single report with MultiQC. Adapter trimming of the reads is done using trim-galore. As part of the Quasitools pipeline Bowtie2 is used to align reads onto the HIV reference genome HXB2 and HyDRA is used for HIV variant calling. Quasitools outputs filtered FASTQ files, amino acid variant call files, a mixed base consensus sequence in FASTA format, and a drug resistance mutation report (consisting of identified drug resistance mutations and corresponding mutational frequencies) in CSV format. The consensus sequence is parsed to sierra-local for scoring of identified drug resistant mutations. The drug resistance report generated by HyDRA is combined with the JSON file (from sierra local) in the R programming environment to generate a comprehensive drug resistance report. The tools are assembled into an automatic workflow using Nextflow. QuasiFlow is distributed with docker containers for all third-party tools.

CONCLUSION: To this end, we developed QuasiFlow, a portable and scalable pipeline for the reproducible analysis of NGS-based HIVDR testing data. QuasiFlow provides a single platform that can run fully offline (with periodic database update) to expeditiously generate user-friendly HIV-1 drug resistance reports from raw NGS data. This tool will improve reporting times of NGS-based HIVDR testing results, especially for researchers with unreliable internet connectivity and in cases where data transfer to remote servers is restricted.

Alfred Ssekagiri

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Alfred Ssekagiri is a third year PhD candidate at the University of Makerere, Uganda in the Department of Immunology and Molecular Biology under the supervision of Dr Daudi Jjingo. Alfred's research interests are in the area of microbial informatics, with interest in developing methods and tools for analysis of microbial communities and relationship between the microbiome, antimicrobial resistance, health and disease. In his PhD fellowship he is developing tools for integrative analysis of microbiome multi-omics datasets. He obtained his B.Sc. from Makerere University where he studied Mathematics, physics with education. He later joined African Institute for Mathematical sciences where he obtained M.sc. in mathematical science. Alfred received an M.Sc. in Bioinformatics from University of Glasgow where he developed an R package for statistical analysis of microbial communities in an environmental context. He has participated as H3ABioNet bioinformatics associate at Uganda Virus Research Institute, co-facilitated bioinformatics trainings hosted at the institute and East African Network for Bioinformatics Training (EANBiT).

POSTER

Establishment of a clinical and genomic database of Breast cancer in Mali

Amoro TRAORE (1), **Oumar SAMASSEKOU** (1), **Madani LY** (2), **Guida LANDOURE** (3), **Mahamadou TRAORE** (4), **Bakarou KAMATE** (5)

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- (6) Department of General surgery, CHU Gabriel Toure, Bamako, Mali**

INTRODUCTION: Breast cancer is the most frequent cancer in women in Mali. Some clinical reports have mentioned the presence of hereditary breast cancer in some Malian families. Gaining knowledge on specific ethnic or genetic predispositions of familial breast cancers is important because it directly affects the clinical and therapeutic management of patients. However, current data on the genetic predisposition of breast cancer comes primarily from Western populations, which can potentially lead to inappropriate care for patients of non-Western ethnicities. It is, therefore, crucial to start studies in regions of the world such as Mali where the genomic profile (tumor and somatic) of patients with breast cancer is not yet known. At present, no clinical database associated with genomic data on breast cancer exists in Mali.

OBJECTIVES: The Aim of this pilot study with a view to creating a clinical and genomic breast cancer database.

METHODOLOGY: We carried out our study over a period of 10 months and enrolled 122 breast cancer patients. We recorded available demographic and clinical data of the patients and then extracted DNA from their peripheral blood.

RESULTS: We found that 122 patients 69.8% were at stage T3 and T4, nodal invasion was present in 95.08% of patients, and the nuclear grade II or III according to SBR was found in 54%. The patient had a median survival of 4.02 years. We were able to obtain DNA in 87.70% of patients with optimal concentration and purity for subsequent experiments. Finally, 76 patients of the 122 patients in the cohort had complete clinical data and DNA of adequate quantity and quality required for a breast cancer biobank. We found that the most challenging part of this pilot study was to record accurate and complete clinical data. A

CONCLUSION AND NEXT STEPS: Although getting a high quality and adequate quantity of DNA is essential, the cornerstone to establish a biobank for breast cancer in Mali is to get high quality clinical data. From this pilot study, we have acquired tangible data to create a clinical and genomic database of breast cancer in Mali. **Keywords:** Cancer, Biobank, breast, clinical, genomic.

AMORO TRAORE

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Amoro Traore is a student of the faculty of medicine who was born in 1993 to mali, 5 years later he entry in school, he has done his primary school at banakabougou in destin-school since 2008, after his primary school he went to high school in 2009 and got his Baccalaureat in 2012, he has been in faculty of medicin since 2013.

SPEED TALK

Benchmarking of Long-read Assembly Tools for Analyzing African Genomes

Andrews Frimpong Adu (1,3), Pandam Salifu (1), Peter Amoako-Yirenkyi (2, 3), Henry Martin (3,4)

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(2) Department of Mathematics, KNUST

(3) National Institute of Mathematical Sciences, NIMS-KNUST

(4) Department of Physics, KNUST

INTRODUCTION: Assembling quality reference, eukaryotic genomes is becoming more feasible for single research teams. This has resulted in developing a plethora of assembly algorithms and packages. Most past benchmarking work has concentrated on how these tools perform for bacterial genomes. Therefore, more extensive benchmarking research for eukaryotes (humans) is needed to assist research communities in deciding which strategy to utilize for assembling human genomes. The wide range of sequencing methods and assembly algorithms available make it difficult in deciding which tool to use and there is no clear consensus on a best-practice and a computational strategy for the de novo genome assembly of eukaryotic genomes. Here, we provide findings from a thorough benchmarking of ten (10) cutting-edge assembly methods for assembling African genomes utilizing PacBio HiFi sequencing (HG02723) data.

METHOD: The assembly tools including ABruijn, HiCanu, Hifiasm, Flye, MBG (Minimizer-based sparse de Bruijn graph building), JumboDB, BCALM, Miniasm/Racon, Raven, and Redbean (Wtdbg2) were assessed based on their

default parameters. We present recommendations for the most efficient tools based on standard evaluation criteria such as assembly contiguity, computational performance, reference reconstruction, and specific genome constraints such as genome representation, including repeat content, percent GC content, genome fraction, and genome size.

RESULTS AND FUTURE DIRECTIONS: Flye produced a reliable assembly with an assembly length closer to the human reference genome (hg38) and the largest contig and alignment length. Moreover, Flye had the most significant mismatches and made the smallest indels. ABruijn and Redbean also produced good assembly results. ABruijn achieved the smallest number of contigs and had the most extended runtimes of all the assemblers tested. HiCanu and Hifiasm HiFiasm produced phased assemblies; however, HiCanu used the least runtime for its assembly compared to the other assemblers. Misassembly detection revealed a higher number of misassemblies in HiCanu with higher transposable elements (TEs) and the largest unaligned length than any of the assemblers tested. Future directions will explore more African genomes and the possibility of combining tools for better performance.

Andrews Frimpong Adu

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I am a Ph.D. student in Biodata Analytics and Computational Genomics at the Kwame Nkrumah University of Science and Technology, Ghana. I am working on de novo genome assembly tools and genome graph building particularly specializing in procedures for representing genome graphs that minimize memory storage with less computational power. I have interested also in developing methods and tools for Health care and personalized medicine.

POSTER

Epidemiology of Bovine Tuberculosis and Its Zoonotic Implication in Addis Ababa Milkshed, Central Ethiopia
**Begna Tulu^{1,2*}, Aboma Zewede³, Mulugeta Belay⁴, Miserach Zeleke¹, Mussie Girma¹,
Metasebia Tegegn⁵, Fozia Ibrahim⁵, David A. Jolliffe⁴, Markos Abebe⁵,
Taye Tolera Balcha⁵, Balako Gumi¹, Henny M. Martineau⁶, Adrian R. Martineau⁴ and
Gobena Ameni^{1,7}**

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2 Department of Medical Laboratory Sciences, Bahir Dar University, Bahir Dar, Ethiopia, **3** Ethiopian Public Health Institute,

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Bovine tuberculosis (bTB) continues to be one of the most widely distributed chronic infectious diseases of zoonotic importance, which causes a significant economic loss in animal production. A cross-sectional study was conducted to estimate the prevalence of bTB and its associated risk factors and type the *Mycobacterium bovis* isolated in central Ethiopia. A total of 65 dairy farms and 654 cattle were tested for bTB using a single intradermal comparative cervical tuberculin (SICCT) test. Data on farm management, animal-related characteristics, and the owner's knowledge of the zoonotic importance of bTB were collected using a structured questionnaire. In addition, a total of 16 animals from different farms were identified for postmortem examination. Lowenstein Jensen (LJ) culture was also conducted, and spoligotyping was used to type the *M. bovis* strains isolated. Chi-square test and logistic regression models were used to analyze the herd- and animal-level risk factors. Herd- and animal-level prevalence rates of bTB were 58.5% (95% CI: 46.2%–69.2%) and 39.3% (95% CI: 35.5%–43.5%), respectively. At the herd level, poor farm management was the predictor for bTB positivity ($p < 0.05$). Animal breed, poor BCS, farm type, and poor farm management conditions were significant predictors of bTB positivity ($p < 0.05$) at an individual animal level. All animals identified for postmortem examination were found to have gross TB-like lesions. A total of 14 *M. bovis* strains were identified from 12 animals that were positive for LJ culture. The strain with the largest number of clusters (five isolates) was SB1176, followed by SB0134 (three isolates), SB0192 (two isolates), and SB2233 (two isolates), and two new strains, each

consisting of only one isolate. The majority (58.5%) of the respondents did not know the zoonotic importance of bTB. The result of this study showed a high prevalence of bTB in the Addis Ababa milkshed and a low level of consciousness of the owners on its transmission to humans. Therefore, the launching of acceptable control measures of bTB and the creation of public awareness about its zoonotic transmission and prevention measures are required.

Keywords: bovine tuberculosis, Addis Ababa milkshed, zoonotic implication, spoligotyping, farm management

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Begna Tulu (Ph.D.) is a senior Tropical and Infectious Diseases and clinical laboratory expert at Bahir Dar University, Ethiopia. His main research interests are in the host-pathogen-environment interaction to develop diagnostics and vaccines, with a particular focus on tuberculosis. Presently, he is involved in the anticov.org clinical projects sponsored by dndi.org. On top of that, as an honorary staff member at Ambo University, Ethiopia, I am also involved in the project Enhancing Public Health Capacity through Academic Networks and Mobility. This is a North-South collaboration initiative between VID University (Norway) and Ambo and Jimma Universities (Ethiopia) aimed at building capacity in public health among African universities.

POSTER

Comparative Genomic Analyses of Helicobacter Pylori Isolates Among Patients with Peptic Ulcer Using Whole Genome Sequences

Biigba Yakubu, Kwame Nkrumah University of Science and Technology, Kumasi, Ghana

Pandam Salifu(phd) Kwame Nkrumah University of Science and Technology, Kumasi, Ghana

It is worth noting that all strains of H. pylori expression virulence are currently treated same way with similar antibiotics across Africa. However, some strains are highly pathogenic and are more often associated with specific clinical outcome, whereas others can be less virulent and are rarely associated with any clinical diagnosis.

As such we sorted for 100 genomes sequences of 100 H. pylori isolates from ten (10) different locations in Africa and employed Bioinformatic pipelines and packages such as Abricate package, MLST, Prokka, Roary Pangenome pipeline, Phandango, R package to compare the virulence genes harbored in each isolate and their degree of association to antibiotic resistance or susceptibility.

Over 9k AMR genes of the bacteria were observed from all the hundred (100) with varying quantities in each isolate. AMR genes which constituted the toxins dominated in south African isolates while the secretions were profound in the Nigerian isolate whereas the adhesins dominated in the north African (Morocco) isolates. Majority of the isolates (95%) did not show any genetic relatedness while almost 90% of the isolates harbored shell genes, 50% harbored cloud and core genes. Interesting some isolates from Angola, Somalia and South Africa did not retain the CagA and vacA genes meanwhile the most frequently used antibiotics such as clarithromycin, metronidazole amoxicillin and tinidazoles were all reported to be resistant to most of the AMR genes (vacA, cagA, oip, bab) observed in this study.

Conclusion: This study found AMR genes in all H. pylori strains in all geographies around Africa. In the African region antibiotic usage could be contributing to the spread of H. pylori antibiotic resistance

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Mphil Biodata Analytics and Computational Genomics, KNUST, Kumasi, Ghana west Africa B.Sc Agricultural science, knust, kumasi diploma in education, Bagabaga college of education, Tamale, Ghana worked in both private and public sectors as field supervisor and classroom teacher. loves sciences but equally vocal on national and international issues. conversant at public speaking and moderation. single and a believer by faith.

SPEED TALK

Impact of COVID-19 on the Sputum Conversion Rate and Adherence of Tuberculosis patients to Directly Observed Treatment at some Diagnosis and Treatment Centres of the Littoral and South West Regions of Cameroon within 2019 and 2021: A hospital-based retrospective study

Fiona Tchouaket Ngueyi^{1,2*}, Kevine T. Tiogou^{1,2}, Nancielle Mbiatong Tchata^{1,2}, Henry Dilonga Meriki^{1,2,3}, Desmond Akumtoh Nkimbeng^{1,2}, Magha Chefor^{1,2}, Jerome Fru-cho^{1,2}, Abdel Njouendou^{1,2}, Samuel Wanji^{1,2}

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BACKGROUND: Tuberculosis (TB) is a disease of public health concern especially in countries like Cameroon with limited resources and accounts for the majority of morbidity and mortality rate among individuals from a single infectious agent. TB is managed by TB control programs (Directly Observed Therapy) where patients are followed throughout their treatment period with their sputum smear test done routinely at specific intervals. This study aimed to examine the dynamics of sputum conversion together with assessing the outcome of patients during effective tuberculosis treatment in nine diagnostic and treatment centers in the Littoral and South West regions of Cameroon. **METHODS:** A hospital-based retrospective cohort study was conducted by reviewing treatment registers of TB patients from January 2019 to March 2021. A structured data collection form was used to extract TB data of interest of patients aged 18 years and above. Data were analyzed using Statistical Package for Social Sciences version 20 and chi-square test was done to establish association between independent and dependent variables. **RESULTS:** A total of 4,460 records of TB patients were reviewed, out of which 2,159 (48.4%) were smear positive pulmonary TB patients and retained in the study. Of the 2,159 patients included in the study, 1356 (62.8%) were males and 803 (37.2%) were females. The sputum conversion rate of TB patients was 95.6% before the COVID-19 era as opposed to 98.3% during COVID-19. With respect to the TB treatment outcome, 72% of patients were cured before COVID-19 as opposed to 72.6% during COVID-19. A higher proportion of patients were lost to follow-up during COVID-19 (13.8%) than before COVID-19 (6.5%). **CONCLUSIONS:** Despite the fact that the COVID-19 does not seem to affect negatively the sputum conversion rate of TB patients attending these treatment centres, a significantly higher proportion of them were lost to follow up during the COVID-19 pandemic. This calls for attention of the health system to improve on strategies to retain patients into the DOT programmes during COVID-19. This can only happen with a reallocation of more resources towards TB during COVID-19 pandemic to ensure good patient compliance to the treatment.

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I am a 23 years old MSc student in Epidemiology and Control of Infectious diseases at the University of Buea. I am a Bachelor degree holder in Microbiology with minor in Medical Laboratory Technology. In 2020, I did an internship at the District Health Service of Buea during which I was actively involved in a cholera outbreak that happened at that time in the locality and I found the experience very much exciting. I am a disciplined, open minded and research oriented young lady passionate about research linked to infectious diseases. Presently, I am undergoing a research on the treatment outcome of tuberculous patients visiting certain diagnostic and treatment centers in my home country.

SPEED TALK

Clinical and genetic aspects of osteogenesis imperfecta in a Malian family

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Introduction: Osteogenesis imperfecta (OI) is a group of genetic disorders affecting connective tissue leading to bone fragility. It is a rare genetic condition inherited in an autosomal dominant or recessive manner and caused by mutations in COL1A1/2 genes encoding for type 1 collagen in 95% cases. OI is characterized by frequent fractures with minimal or absent trauma. Clinical features include bone fragility, bone deformity, short stature, dentigenosis imperfecta, and hearing impairment in adult year. Only few genetic confirmed cases have been reported in Africa, and none in Mali.

Objectives: To clinically characterize a patient with osteogenesis imperfecta and identify the underlying the genetic defect.

Methodology: The patient and her family members were enrolled after an informed written consent and assent were obtained. The patient was seen by a multidisciplinary team including neurologist, rheumatologist,

ophthalmologist and ENT specialist. Bones X-Ray and blood chemistries including Vitamin D were performed. DNA was collected for candidate gene testing.

Preliminary results: A 7-year-old girl from a consanguineous marriage was referred to our neurogenetics for multiple fractures. She is the third child in a sibship of three. It was an uneventful pregnancy and delivery. She first fractured her right clavicle at age 2 without any trauma. Then, she respectively fractured her left clavicle, left humerus (3 times), right arm, left arm, right leg, left leg, and the most recent one was her right arm in a context of minimal trauma. At clinical examination, anthropometric measurements included a weight of 10 kgs, height of 92 cm, head circumference of 46 cm. She presented with bowed and short legs and arms, chest deformity. Limbs X-ray showed multiple fractured and curved long bones and demineralization. Blood chemistries included decreased vitamin D (17 ng/ml) and normal calcium (2.51 mmol/l). According to clinical, radiologic and laboratory findings osteogenesis imperfecta was the more likely diagnosis.

Next step: Candidate gene testing including: COL1A1/2, LEPRE1, CRTAP, PPIB is ongoing to uncover the genetic defect in this family. Additional genetic analysis including WES/WGS might be needed. The genetic testing will uncover the genetic defect in this family.

Key words: Osteogenesis imperfecta, COL1A1/2 gene, WES/WGS, Mali.

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I am a graduate student currently enrolled in the neurology residency and research assistant at the Neurogenetics laboratory. I have been working on recessive ataxias since my internship in 2018. I mainly see patients in clinics, perform DNA extraction and analyse NGS data.

SPEED TALK

Characterisation of pharmacogene allelic variation in African populations and development of a novel diplotype calling algorithm.

David Twesigomwe [1,2], Zané Lombard [2], and Scott Hazelhurst [1,3] on behalf of the Wits-H3A/GSK ADME Collaboration.

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BACKGROUND: Genetic variation is in part responsible for the variability in drug response within and between populations. However, the full catalogue of pharmacogenetically-relevant alleles is yet to be established, in particular for African populations. Therefore, this study aimed to characterise the variation in three core pharmacogenes in drug metabolism i.e. CYP2D6, CYP2B6 and CYP2A6, across diverse continental African populations for whom high coverage whole genome sequence (WGS) data has recently become available through H3Africa and other collaborations across Africa. **METHODS:** Given the known challenges of genotyping these hypervariable genes, we developed a novel graph- and Nextflow-based pipeline (StellarPGx) to facilitate scalable, reproducible and more accurate CYP450 allele calling using WGS data. Thereafter, we applied a consensus star allele calling approach, involving StellarPGx, Astrolabe, Aldy, Stargazer and Cyrius to assess the CYP2D6, CYP2B6 and CYP2A6 allelic diversity mainly across sub-Saharan Africa, based on 962 high-depth African genomes. Targeted Single Molecule Real-Time (SMRT) Sequencing was used to validate novel haplotypes in a subset of our study population. **RESULTS:** For this presentation, the focus will be on results for CYP2D6 and CYP2B6 as the allele nomenclature for CYP2A6 is undergoing an update. (1) We inferred over 46 potential novel CYP2D6 and CYP2B6 haplotypes combined – most of which were rare and African-specific. (2) Our results highlight the frequency distributions of actionable alleles such as *17, *29, *4 and *5 for CYP2D6, and *6, *22 and *18 for CYP2B6. (3) Using high coverage WGS data facilitated calling structural variant alleles for both CYP2D6 (e.g. *5, *1xN, *36 and *68+*4) and CYP2B6 (e.g. *29). (4) Activity score-based phenotype prediction showed that the distributions of CYP2D6 and CYP2B6 metaboliser phenotypes are non-uniform across SSA. **CONCLUSION:** This study presents the StellarPGx pharmacogenomics pipeline, novel African-specific CYP2D6 and CYP2B6 alleles, and exemplifies the utility of multi-algorithm-based allele calling in mining high coverage African WGS data for variation in highly polymorphic CYP genes. Our findings highlight the diverse landscape of CYP2D6 and CYP2B6 star alleles and predicted phenotypes, mainly in SSA populations, which could inform future precision medicine strategies.

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David is a PhD (Human Genetics) candidate at the Sydney Brenner Institute for Molecular Bioscience (SBIMB), Wits University, Johannesburg. He is part of the Wits-H3A/GSK ADME team – studying variation in genes that encode proteins involved in absorption, distribution, metabolism, and excretion (ADME) of medications in the African context (<https://www.wits.ac.za/research/sbimb/research/gsk-adme>). Before joining Wits, David completed his BSc in Biomedical Sciences at Makerere University in 2017 and thereafter joined the Core laboratories at the Uganda Virus Research Institute (UVRI) for a three-month internship in Bioinformatics supervised by Dr. Jonathan Kayondo. David then continued his development at UVRI by doing a one-year strategic internship in Bioinformatics and Computational Biology under the Makerere University/UVRI Center for Excellence in Infection and Immunity (MUII-Plus) Research and Training programme. This unique opportunity enabled him to receive focused hands-on training in Unix, Python and R programming, NGS data analysis, Metagenomics, and pipeline development among other skills. The MUII-Plus strategic internship also offered David an opportunity to receive mentorship from leading Bioinformatics researchers in Uganda and H3ABioNet at large. David is excited about the opportunity to contribute to pharmacogenomics research in Africa and precision medicine in general, starting with the fascinating work under the Wits-H3A/GSK ADME collaboration. He is also very passionate about mentoring fellow young scientists that are considering career choices in Bioinformatics and Computational Biology.

SPEED TALK**Prevalence of advanced HIV disease in people living with HIV on antiretroviral therapy at the Buea Regional Hospital and co-infection rates of *Mycobacterium* spp. and *Cryptococcus* spp.**

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Background: HIV/AIDS remains a significant public health problem, having claimed 36.3 million lives so far. In 2020, approximately 680,000 people died from AIDS-related illnesses worldwide. Despite increased access to antiretroviral treatment (ART), the burden of mortality and morbidity due to advanced HIV disease (AHD) has not significantly changed over the years. AHD individuals are more prone to opportunistic infections– including tuberculosis (TB) and Cryptococcal meningitis (CM). CM has been reported as a significant cause of mortality among people living with HIV (PLHIV), accounting for 15% of the global AIDS-related deaths in 2019. To keep track of the commitments made by the government to end the AIDS pandemic as a public health threat by 2030, knowledge of the burden of AHD and its risk factors is essential. Methods: A hospital-based cross-sectional study was conducted from February 2020 to February 2021. HIV-infected individuals on ART at the Buea Regional Hospital were screened for AHD after obtaining their consent. A pre-tested semi-structured questionnaire was administered to the participants, and blood, sputum, and urine samples were collected. Cryptococcus antigen lateral flow test was performed from serum. Chi-square and regression analysis were carried out on SPSS at a 5% significance level. Results: From a total of 3,229 PLHIV on care in the study period, 327 (10.1%) were suspected of having advanced HIV disease, and these clients were enrolled in the study, most of them females (62.4%). The mean age of the participants was 40.5 ± 24.7 years SD. A total of 130 (39.8%, [95% CI, 34.41-45.29]) participants presented with AHD. Forty-one (35.7%, [95% CI, 26.94-45.12]) and 7 (6.7%, [95% CI, 2.72-13.25]) had TB and Cryptococcal infections respectively. One patient presented with both CM and TB (0.7%). AHD was associated with male gender (aOR; 3.9, 95% CI; 1.40-11.70), and being on ART for more than 12 months was protective (aOR; 0.1, 95% CI; 0.01-0.35). Conclusion: Even when ART uptake has dramatically improved in Cameroon, AHD (35.7%) is far beyond the UNAIDS reduction target of 10% by 2030. Improvement in the management of PLHIV is warranted. Keywords: HIV/AIDS, Advanced HIV disease, *Mycobacterium* spp., *Cryptococcus* spp., antiretroviral therapy.

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SPEED TALK

Gastric microbiome predictive model for gastric carcinogenesis

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Gastric cancer continues to be one of the most significant cancers globally. Changes in the microbiome composition and interaction have been implicated in gastric cancer development. *Helicobacter pylori* continue to be one most important pathogen affecting the pathogenesis of the disease; however, non-h. *Pylori* bacteria have been reported to influence the progression of the disease. Toward the understanding of how the microbiome affects the pathogenesis of the disease, many studies have provided relevant yet varying results. We present a comprehensive analysis of the gastric microbiome in gastric carcinogenesis, focusing on bacterial diversity, co-occurrence pattern, and ultimately identification of potential microbial biomarkers. We combined raw 16s rRNA data from six (6) studies across 985 samples from individuals consisting of healthy, gastritis, intestinal metaplasia and cancer. Batch effects were corrected with the Herman package in R. The gastric microbiome was dominated by Bacteroidetes, Proteobacteria, Firmicutes, Actinobacteria, Acidobacteria, and Fusobacteria. However, the composition of the phyla differs with the development of gastric cancers. Differential abundance testing revealed bacteria species, mainly *Sphingobium* and *Zoogloea*, that can degrade toxic xenobiotic compounds to harmless forms were highly associated with healthy individuals. The Proteobacteria composition and diversity decrease, and the Actinobacteria increase with carcinogenesis. Transient oral pathogenic and intestinal bacteria, *Prevotella*, *Propionibacterium acnes*, *Acinetobacter baumannii*, *Lactobacillus*, *Gordonai* polyisoprenivorans, were highly enriched with increasing carcinogenesis from gastritis to cancer. Microbial co-occurrence analysis revealed important keystone species with *Pseudoxanthomonas spadix* and *Sphingobium* represented as hubs in healthy individuals. *Filifactor alocis* showed significant interaction with pathogenic bacteria *Fusobacterium nucleatum* in gastric cancer communities. LASSO models identified potential microbial signatures that could distinguish healthy, gastritis, intestinal metaplasia samples pairwise with high accuracy (average ROC of 8.0). *Bacteroides dorei*, *Hydrogenophilus hirschii*, and *Propionibacterium granulosum* were discriminative for gastric cancer. This study provides significant insight into the gastric microbial communities and how they could serve as a potential tool for predicting gastric carcinogenesis. However, studies are required to investigate the potential biomarkers identified in this study.

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I am a diligent young aspiring research scientist in areas of molecular biology, genomics, and bioinformatics. I am committed to improving the livelihood of the people in my community by contributing my quota to the improvement of health through the application of bioinformatics tools. I envision myself in the near and distant future as a research scientist with the necessary skills, knowledge and competence to develop and implement effective and efficient research strategies to retain and develop new tools in computational biology to improve healthcare for the benefit of Ghana and Africa at large. My primary research interest is in the application of molecular biology and computational tools to understand diseases affecting Ghana and the African diaspora at large. I am broadly interested in understanding genetic basis of certain disease especially non-infectious diseases, studying disease patterns and microbial profiles associated with certain diseases with much emphasis on molecular biology and computational genomics as a diagnostic tool. My future research interests are shaped by the emerging trends in the use of bioinformatics to improve health.

SPEED TALK

Novel MARVELD2 Variant Associated with Post-lingual Autosomal Recessive Non-Syndromic Hearing Impairment in Consanguineous Ghanaian Family

Elvis Twumasi Aboagye (1,3*), Samuel Mawuli Adadey (1,3), Edmond Wonkam-Tingang (3), Kevin Esoh (3), Lucas Amenga-Etego (1), Isabelle Schrauwen (2), Gordon A. Awandare (1), Suzanne M. Leal (2) and Ambroise Wonkam (3,4)

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Systematic investigation of implicated candidate genes has identified gap junction protein beta 2 genes (GJB2)-p.(Arg143Trp) founder variant as the major contributor (25.9%) to non-syndromic hearing impairment (NSHI) in Ghana. The use of Next Generation Sequencing in hearing-impaired studies is limited in Africa, despite the high population genetic diversity, and genetic heterogeneity of HI. This means that we are missing the full spectrum of the gene(s) variants/allele(s) co-segregating with HI among affected families in the sub-region. Whole-exome sequencing (WES) was used to investigate a consanguineous multiplex Ghanaian family with post-lingual autosomal recessive non-syndromic hearing impairment. The elucidated putative marker was further validated by direct Sanger sequencing and screened in ethnologically matched hearing controls (n=46), and a group of unrelated individuals with simplex NSHI (n=151). The mutant protein was modeled with a resolved wildtype protein (PDB ID: 5N7K) as a template, using homology modeling on SWISS-MODEL. A literature review of the implicated gene variants association with HI was performed. Homozygous MARVEL domain containing 2 (MARVELD2): c.1058dupT (p.Val354SerfsTer5) frameshift variant on exon 2 segregates with the phenotype in this family. This bi-allelic novel MARVELD2 (MIM 610572) variant was not identified in the 46 matched controls, and 151 unrelated sporadic cases screened. Likewise, the variant was absent in hereditary hearing loss homepage (HHL), online Mendelian inheritance in man (OMIM), human phenotype ontology (HPO), and ClinVar among many other available databases. The identified MARVELD2: c.1058dupT (p.Val354SerfsTer5) variant predicted to truncate the encoded protein, is the putative pathogenic variant associated with the intra-family bilateral ARNSHI. A review on the gene associated with ARNSHI identified isolated reports of twelve different MARVELD2 variants from seven countries. The bi-allelic MARVELD2 nonsense mutation [c.1058dupT: (p.Val354SerfsTer5)] identified in this multiplex consanguineous Ghanaian family segregating bilateral, sensorineural, post-lingual ARNSHI, is novel and adds to the global knowledge in refining HI disease-gene pair curation.

Keywords: MARVELD2, Tricellulin (TRIC), Non-syndromic Hearing Impairment; Ghana.

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Aboagye, Elvis Twumasi is a Ph.D. student at the University of Ghana and holds a thesis completion fellowship at the University of Cape Town. He is working on Hearing Impairment Genetic Studies (HI-GENES) in Africa Project. This project seeks to explore functional genomics and African genome diversity to elucidate specific markers linked to human hearing disorders. The goal is to establish a causal link between population-specific genetic variations and epigenetic modifications/dysregulation, to disease etiology or susceptibility. At the West African Centre for Cell Biology of Infectious Pathogens (University of Ghana) and Division of Human Genetics (University of Cape Town), we are focused on elucidating population-specific genetic markers for segregating early on-set of autosomal recessive non-syndromic hearing impairment (ARASHI) in Africa. The target is to create and design a molecular diagnostic tool for newborn screening in Africa. Future investigation will explore i) the use of computational algorithms to correct varied cellular epigenetic dysregulation, 2) epi-transcriptomics and disease/disorders, and 3) single-cell methods that allow the description of the single-cell resolution of DNA modifications.

SPEED TALK

Vitamin D binding protein gene polymorphism and free serum bioavailability among tuberculosis patients and household contacts

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Background: The vitamin D binding Protein gene is highly polymorphic with over 120 variants. Genetic variants influence the distribution of vitamin D in circulation leading to vitamin D deficiency. The two extensively studied nonsynonymous DBP single nucleotide polymorphisms (SNPs) rs7041 and rs4588 are found in diverse populations. The wild-type Gc1F genotype is predominantly found in the African population, with a low frequency of Gc2 and Gc1S, and it is associated with low vitamin D levels in whites. **Objective:** The aim of this study was to examine the association of the DBP gene polymorphism with the free and bioavailable vitamin D levels among active TB patients, individuals with latent TB infection and those without TB infection.

Methodology: This was a cross-sectional study of 56 active tuberculosis patients, 49 latent tuberculosis individuals, and 43 without tuberculosis infection. Sanger sequencing was performed and single nucleotide polymorphisms were identified using BioEdit tool. Percentages were used for frequency distribution. Logistic regression and Spearman's correlation were used to determine associations. **Results:** Frequencies of 98% GCF1, 2% GC2 and 1% GC1S genotypes were reported with no deviation from Hardy-Weinberg equilibrium, $D' = 0$. The median vitamin D levels in the GC1F genotype were 3.8 ng/ml, GC1S 2.2ng/ml and GC2 1.9 ng/ml respectively. Vitamin D binding protein gene and vitamin D levels were not statistically significant, p -value 0.05. Spearman's correlation between the D binding protein gene and free vitamin D levels revealed a negative association ($r = -0.0404$). **Conclusion:** The DBP rs7041T and rs4588C (Gc1F) genotype was predominantly found in active TB patients, LTBI individuals and those without TB infection, with no difference in frequency distribution among the three groups. No significant association was found between the D Binding Protein gene and the free and bioavailable vitamin D levels. However the Gc1F genotype is known for high utilization of vitamin D metabolites, therefore leading to the relatively low median levels observed. **Future direction:** Further research is warranted in a heterogeneous population to detect the minor alleles and therefore associate them with the free and bioavailable vitamin D levels.

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POSTER

Effect of a capacity-building workshop on knowledge about concepts related to ELSI in biobanking and genomics among Ethics Committee Members and Early Career Researchers in West Africa

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Introduction: There is a growing interest in stroke genomics, biobanking and associated Ethical, Legal and Social Issues (ELSI) in sub-Saharan Africa (SSA). However, the requisite knowledge on these fields remains sparse especially among Ethics Committee Members (ECM) and Early Career Researchers (ECR) in West Africa.

Objective: This study documents gain in knowledge among ECM and ECRs following participation in a workshop on ELSI related to stroke biobanking, genomic research and genetic counselling.

Methods: The African Neurobiobank for Precision Stroke Medicine - (ELSI) Project conducted a 2-day workshop for ECR and ECM (N=143) from its seven sites in Ghana and Nigeria on biobanking, genomics research, genetic counselling, and other ELSI-related topics. A 20-item semi-structured questionnaire was used for pre- and post-workshop assessment of knowledge. Data was analysed quantitatively using (STATA version 16) descriptive statistics and Mann Whitney U test for comparison of means at $p < 0.05$.

Results: Majority of participants were males (57.5%), aged 30-39 years (39.8%) at mean age (41.4 ± 9.97); have > 10 years professional experience (35.8%), Master/MPhil degrees (53.0%) and 57.1% have worked with people living with stroke. Overall, the percentage of participants with good knowledge of biobanking and genetics increased from 33.0% in pre-test to 67.0% post-test ($p < 0.042$). When stratified by participant group, the percentage of ECM participants with good knowledge on data sharing and ethical framework increased from 28.9% at pre-test to 71.1% post-test while ECR participants' knowledge on genetic counselling increased from 43.1% at pre-test to 56.9% post-test.

Conclusion: Participation in this workshop increased knowledge in the above fields. This needs to be sustained in the interest of implementation of quality and effective ELSI and Genomic research studies in Africa.

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Ezinne Uvere holds a Bachelors degree in Microbiology from the Federal University of Technology, Owerri and a Masters degree in Public Health (MPH) from the University of Ibadan, Nigeria. She is an early career researcher with over ten years of progressive experience in coordination and implementation of collaborative research initiatives funded by local and international donors. She has an employment history with academia, faith-based local and international not-for-profit organizations. Prior to her current position as the Programme Manager on the African Rigorous Innovative Stroke Epidemiological Surveillance (ARISES) study, she was a Research Associate on the Stroke Investigative Research Education Network (SIREN) and Programme Manager on the Systematic Investigation of Blacks with Stroke (SIBS Genomics) studies respectively. Her areas of research interests include non-communicable disease [cardiovascular risk factor] prevention especially stroke, community engagement, Child/adolescent health. Currently, she is a Ph.D student at the Faculty of Public Health, College of Medicine, University of Ibadan, Nigeria with a research focus on cardiovascular risk factor prevention and also provides assistive support towards the Community Engagement component of the African Neurobiobank/ELSI Project at the College of Medicine, University of Ibadan, Nigeria.

SPEED TALK

Docking of human band 3 anion transporter proteins with their Plasmodium falciparum interactors based on short linear motifs

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BACKGROUND: Plasmodium falciparum (P. falciparum) is a pathogen causing severe forms of malaria. In the blood stage, several protein interactions occur between P. falciparum and Human erythrocytes. Among Human proteins, the Band 3 Anion Transporter (B3AT) is considered as the main invasive pathway for the parasite in erythrocytes causing malaria symptoms. **AIM:** The interactions between P.falciparum parasite and erythrocytes along this receptor have been investigated with specific focus on SLIMs. **METHODS:** The Agile Protein Interactomes DataServer (APID), a protein interaction repository was used to identify interaction between P. falciparum and its Human host. After SLIMs was identified in a set of sequences using SLiMFinder program. SLIMs are Short Linear Mediators sequences involved in several biological processes, acting as mediators of protein interactions. Moreover, PATMATDB program aided to screen the motifs identified by SLiMFinder in the sequences of Plasmodium proteins in order to predict those interacting with B3AT. In the end a guided protein-protein docking based on SLIM motifs was carried out with HADDOCK (High Ambiguity Driven protein-protein Docking) program and COCOMAPS for analyzing, visualizing and comparing the interface in protein-protein complexe. **RESULTS:** We have identified 25 interactions validated at least by two experiments. Among them is the tryptophan-rich protein A5K5E5_PLAVS as an important interactor of B3AT. We identified "[DE] L.R" as a SLIM. Also found out that this SLIM is highly represented in the human (7424 proteins) and P. falciparum (584 proteins) proteomes, emphasizing the importance of the interaction between B3AT and Plasmodium. The best-oriented complex provided with protein-protein docking approach had a very important HADDOCK score of -201.3. The contact map illustrates a very clear pattern of interaction between the 'A5K5E5' and 'B3AT' proteins from the side of the short linear motif (SLIM) of the B3AT. In conclusion, this study showed very interesting results on protein-protein interactions between Human and P. falciparum with the SLIMs involved. And through protein-protein docking we confirmed the potential of our methodology which can be used in other infection studies where molecular mimicry has been demonstrated.

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Fatoumata Gnine FOFANA PhD student in Bioinformatics at the Doctoral School of Science and Technology of Bamako. Her research focuses in 'In silico analysis of protein-protein interaction networks between Human and P. falciparum for the design of vaccine candidates'.

She worked on a project entitled 'Protein-protein Docking of Human B3AT with its Plasmodium falciparum interactors based on Short Linear Motifs (SLIMs)' and funded by NIH Fogarty supported training grants: West African Center of Excellence for Global Health Bioinformatics Research.

Holder of an MSc in Bioinformatics in 2017 from the University of Sciences, Techniques and Technologies of Bamako / African Center of Excellence in Bioinformatics (USTTB/ACE). During my MSc degree, my research focuses on Human and Malaria parasite protein-protein interactions based on Short Linear Motifs (SLIMs) that have been conducted in the Laboratory of Bioinformatics, biostatistics, and biomathematics (BIMs) at the Pasteur Institute of Tunis.

And at the same time, I participated in a research project which consists of the 'Identification of malaria targets and the design of inhibitors using structural bioinformatics tools'.

She followed a 3-month online training course as a participant in 2019 with the University of Cape Town on 'Introduction to Genomic Medicine Training in Africa'.

In 2020 and 2021, she has been involved in online training Course for 3 months as a Teacher assistant organized by the University of Cape Town.

She had the opportunity to participate in various scientific communications and workshops in the field of bioinformatics (Data Sciences, Metagenomics, Grant writing) that took place in Morocco, Nigeria, India, Ghana also in Mali.

POSTER

Bardet-Biedl Syndrome – a case presentation

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Introduction: Bardet Biedl Syndrome (BBS) is a rare genetic disorder. It presents with a group of symptoms which spans all organ system in the body. Key features are progressive pigmentary retinal disease, truncal obesity,

postaxial polydactyly, mental retardation, renal and genital abnormalities. Supporting features span all organ systems in the body.

Objective: To report a case of BBS presenting on account of an ocular complaint and review existing literature highlighting important issues in prognosis and patient counseling

Methodology: A 9 year old boy was evaluated. History was obtained, ocular and systemic examination were performed. Investigations include plasma glucose profile, thyroid function test and electrolytes, urea and creatinine assay

Results: We report a case of a 9 year old Nigerian boy with progressively decreasing vision despite spectacle wear and associated poor vision at night of 3 years duration. There was no family history of similar condition and parents are not consanguineous couple. He was noticed to have polydactyly of both upper limbs at birth. History of rapid weight gain in early childhood and labile emotions were also given.

Examination revealed an overweight child with hyperpigmented macules on the abdomen and bilateral post-axial polydactyly on both hands. He weighed 48kg and waist circumference was 75.5cm which were excess for age (> 95th percentile) with a body mass index of 26.2kg/m². Ocular examination findings of moderate visual impairment, and mid-peripheral punctate lesions and bone spicule pigmentation in both retina. He had a spontaneous penile length of 4.9cm and testicular volume of 1ml each.

Electrolyte, urea and creatinine assay, glomerular filtration rate, thyroid function test and plasma glucose profile were within normal limits.

He was prescribed spectacles and low vision aids and his mother counseled to seek educational support for him.

Next steps: The first presentation of patients with BBS may be to the eye unit on account of poor vision hence a high index of suspicion is needed for early diagnosis. Due to the progressive nature of the disease, optimization of vision at all times is necessary to help them cope, particularly for school children with high visual demands.

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Dr. Folahan Ibukun is a senior registrar in the department of Ophthalmology at the University College Hospital, Ibadan where she provides eye care to people of all ages. She is interested in advancing eye health through research and increasing awareness on prevalent eye diseases through public health campaigns.

SPEED TALK

What do research participants want and what should they be given? Feedback of Genetics Research Findings in the H3Africa Kidney Disease Research Network

Grace Larbie-Gafa^{1*}, Vincent Boima¹, Isabella Rockson¹, Chatio S3, Anita Ghansah², Paulina Tindana⁴, Dwomoa Adu¹ and the H3Africa Kidney Disease Research Network

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Background: Genetics research raise important ethical issues, particularly those related to feedback of findings. The H3Africa Kidney Research Network (H3A-KDRN) has been investigating the genetic causes of chronic kidney disease (CKD) for almost a decade. There is evidence that variants in the Apolipoprotein L1 gene in Africans and people of African descent INCREASE the risk of developing CKD. It is also known that relatives of subjects with CKD are at increased risk of developing CKD. Should participants and relatives be informed about their risk of CKD? What findings should be returned? Who should be involved in the feedback process? The aim of this study was to seek the views of research participants, families and stakeholders on the feedback of genetic research findings.

Methods: This study explored the views of genetic researchers, participants, families and members of research ethics committees on what count as good ethical practice in deciding what, who and how to return genetic findings of the kidney disease research in Ghana. This study employed a qualitative study involving in-depth interviews, focus group discussions and workshops. Data was analysed thematically with the NVivo software 12.

Results: There was a consensus that individual and aggregate genetics results should be fed back to participants and communities. Most participants preferred to receive their personal results from a doctor, genetic counsellor and a research scientist.

Conclusions: There is an ethical imperative to return genetic research results to participants, families and communities. There is the need to train genetic counsellors to support the feedback process.

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A focused young lady with special interest in kidney disease research. She has a Bachelors degree in Biochemistry and a Master of Philosophy degree in Chemical Pathology, both from the University of Ghana. She is currently working as a Research Assistant with the H3Africa Kidney Disease Research Network office under the University of Ghana Medical School, Accra. She is the research assistant on the Community Engagement in Biobanking and Genomics (CEBioGen): Project two. She is optimistic that soon, the root cause of kidney diseases in Africa will be uncovered.

SPEED TALK

Stakeholders' perspectives on community engagement for genomics research in Africa – a literature review

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Introduction: The growth of Genomics research in Africa is timely given the increase in genetic diseases. Genomics is making the prediction, diagnosis, and treatment of diseases more precise and advanced. These advancements, however, become suspect unless their process of scientific discovery is consistent with the interests of individuals or communities participating in genomics research. These interests could be met through community engagement (CE). However, there is limited knowledge on how to effectively carry-out CE in genomics research. Objective: To explore the perspectives of key stakeholders (genomics researchers, communities, and research regulators) to contribute knowledge on how effective CE for genomic research should be. Methods: A search of literature from Google scholar, NCBI and Pubmed, published between 2011 to 2020 was done, using the following words: Community engagement, community, genomics, genetics, stakeholders, effective, meaningful, Africa and ethical Results: Fifty-nine articles were purposively reviewed. Genomics researchers perceived CE to enable understanding of target communities to; inform research questions, study designs, implementation, and dissemination. However, they reported challenges of: limited local and international guidelines on CE for genomics research; difficulty explaining genomics to communities. Community stakeholders appreciate CE as an opportunity to be heard by researchers and to see their input effect change. However, they face challenges of; Inadequate genomic information on the consent form; Resistance by communities due to unfamiliar sampling procedures, and the involvement of healthy participants. Research regulators perceive CE to support informed consent, address potential misconceptions and minimize stigmatization. Research regulators' CE challenge was the limited technical ability of research ethics committees to review proposals for genomics research. Conclusion: When fostering ethical research, central in consideration are the voices and perspectives of different stakeholders that would work with and apply such research. This is important not only to increase the relevance and legitimacy of the research but also because broad stakeholders' input is a key element of ensuring that research fulfills the goals of justice. Next steps: We plan to use these literature findings to facilitate the design of research tools to collect empirical data that will enable the development of a framework for effective CE in genomics research.

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Harriet Nankya, a TrypanoGEN+ Fellow doing a PhD in Applied Ethics from Makerere University, I have a Master of Health sciences in Bioethics and a Bachelor of Science in Biochemistry and Mathematics from the same university. My PhD research is "Towards Effective Community Engagement in Genomics Research in Uganda". My MSc. research assessed "Knowledge, Perceptions and Willingness to participate in Genomics Research in Uganda".

SPEED TALK

Attitudes towards genetic testing and the associated factors among adult eye clinic patients in Ibadan, Nigeria.

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INTRODUCTION

Genetic testing in eye care is becoming increasingly relevant with the elucidation of the human genome and the advent of gene therapies for eye conditions. Patients' attitudes form the basis of their acceptance of testing and may be influenced by various factors.

OBJECTIVE

The aim of this study was to evaluate the attitudes of adult eye clinic patients towards different aspects of genetic testing, and their associated factors, in Ibadan, Nigeria.

METHODOLOGY

The study was a hospital-based, cross-sectional survey using a mixed-methods approach. Consecutively recruited adult patients of the Eye Clinic, University College Hospital, Ibadan were interviewed on their perceptions of genetic testing using an interviewer-administered questionnaire with closed and open-ended questions. Bivariate analysis and multiple logistic regression were used to assess the association between attitudes towards the various aspects of genetic testing and participants' socio-demographic details. A p-value of <0.05 was considered significant. Responses to open-ended questions were coded and thematically analysed.

PRELIMINARY RESULTS

There were 223 participants with a mean (SD) age was 49.4 (18.5) years. A majority (205, 92%) were of Yorubas, 118 (53%) were males, 164 (74%) were married, 148 (66%) were Christians, and 118 (53%) had attained at least tertiary education. About three-quarters were parents (174, 78%) and the average (SD) number of children was 3.9 (1.9). Though 206 (92%) were willing to undergo testing, only 176 (79%) were willing to pay out-of-pocket for it. A majority (148, 66%) believed the government should bear the cost of testing. The maximum amount they were willing to pay ranged from N200 to N20,000 (\$0.49 to \$49). In terms of reproductive planning, only 43 (19.7%) and (15, 6.7%) were in support of prenatal testing and pre-implantation diagnosis respectively and these were significantly associated with having attained tertiary education (p=0.03) and (0.001), respectively. Reasons for low support included religious inclinations and no perceived problems with infertility.

NEXT STEPS

The creation of awareness of the implications, benefits and limitations of genetic testing is important and should be carried out in consideration of the people's perceptions to maximise outcomes.

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Henrietta Ifechukwude Monye is currently an Ophthalmology Senior Registrar at the University College Hospital, Ibadan, Nigeria. Her research interests include ophthalmic genetics and ophthalmic clinical and operational research.

She received her undergraduate medical degree from the University of Nigeria, Enugu, and an MSc in Public Health for Eye Care from the London School of Hygiene and Tropical Medicine. She is a fellow of the International Council of Ophthalmology and a member of the Royal College of Surgeons, Edinburgh.

She is passionate about reducing avoidable blindness and promoting eye health awareness through creative means, which she achieves through the non-profit, EyeHubNigeria Initiative. She enjoys listening to music, spinning creative stories and concepts, and acquiring new skills.

SPEED TALK

Genetic Diversity of Bundibugyo ebolavirus from Uganda and the Democratic Republic of Congo

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Background: The ebolavirus is one of the deadliest viral pathogens which was first discovered in 1976 during two consecutive outbreaks in the Democratic Republic of Congo and Sudan. Since then, six known strains have been documented and caused outbreaks in Africa. Uganda experienced a new outbreak in Bundibugyo district in 2008. This outbreak was constituted with 116 human cases and 39 laboratory confirmed deaths. After 5 years, this strain re-emerged in DRC. This was declared by the WHO on the 17th August 2012. This outbreak had 36 human cases with 13 laboratory confirmed deaths. Despite several research studies conducted in the past, there is still scarcity of knowledge on the genetic diversity of this strain. A research study was therefore undertaken to provide insights into the unique variants of Bundibugyo ebolavirus.

Methods: The approaches used were; Quality Control, Reference Mapping, Variant Calling, Annotation, Multiple Sequence Alignment and Phylogenetic analysis to identify genomic variants as well determine the genetic relatedness between the two epidemics. 41 viral sequences that were retrieved from the NCBI database.

Results: Our analysis identified 14,362 unique variants from the two epidemics. The Uganda isolates had 5,740 unique variants, 75 of which had high impacts on the genomes. These were 51 frameshift, 15 stop gained, 5 stop lost, 2 missense, 1 synonymous and 1 stop lost and splice region. Their effects occurred mainly within the L gene region at reference positions 17705, 11952, 11930 and 11027. For the DRC genomes, 8,622 variant sites were identified. The variants had a modifier effect on the genome occurring at reference positions 577, 5928 and 6587. Examples are C577T, T5928C and T6587C. Phylogenetic reconstruction identified two separate and unique clusters from the two epidemics.

Conclusion: Our analysis provided further insights into the genetic diversity of Bundibugyo ebolavirus from the two epidemics. The Bundibugyo ebolavirus strain was genetically diverse with multiple variations. Phylogenetic reconstruction identified two unique variants. This suggests an independent spillover event from a natural reservoir and not a contact with a past ebolavirus survivor that initiated the resurgence in DRC in 2012. Therefore, the two epidemics were not genetically related.

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I am a final year Bioinformatics MSc student under Nurturing Genomics and Bioinformatics Research Capacity in Makerere University Uganda. My research interests are majorly in Genomic Epidemiology of Infectious Diseases with a key interest in Re(emerging) Viral Zoonoses. Skills that I am focusing at include; Viral Evolution, Variant detection, Metagenomics, Online Bioinformatics and Pipeline development.

In addition to this, I also work as a research laboratory technologist under the department of Arbovirology, Emerging and Re-emerging Infectious Diseases at the Uganda Virus Research Institute, Entebbe. My key roles here is mainly in Real Time PCR analysis and reporting of COVID-19 positive cases.

Outside work, I am a good fun of Manchester United Football Club. I love playing football too. I enjoy watching movies and road trips.

POSTER

Killer cell immunoglobulin receptor diversity and its relevance in the human host's response to HIV infection in African populations.

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Introduction: Host response to the human immune deficiency virus (HIV) involves both the innate and adaptive immune systems. The killer cell immunoglobulin-like receptors (KIRs) found on natural killer cells (innate immune system) and some T-lymphocytes are genetically diverse and play key immune functions in the host response against viral pathogens. In the last decade, there has been substantial growth in sequencing technologies and bioinformatics capacity to understand human host genetics, including KIR. A better understanding of the effects of KIRs on the host's immune response to HIV in African settings is essential to inform strategies to develop more effective therapies and vaccines to improve health among people living with HIV (PLWH). Objectives: To conduct a literature review of the KIR diversity, the role of epigenetics in KIR functional diversity, the role of KIR immunogenetic variation in the human host response to HIV and discuss current perspectives on the studies to assess the relationship between KIR diversity and the HIV disease continuum. Methodology: A focused literature review will be conducted. Databases including EMBASE, MEDLINE, Pubmed, African Journal Online will be used to search for published studies reporting any form of KIR and or HIV associations. Only articles in English will be screened and reviewed by two independent researchers to determine their eligibility based on pre-set criteria and any disagreement will be resolved through discussions with the third reviewer. Data will be summarized into tables, graphs, and text descriptions Next steps: To perform the literature review and data synthesis.

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John Mukisa, MD is a PhD in Bioinformatics BReCA fellow at the Makerere University College of Health Sciences in Kampala, Uganda where he also obtained his undergraduate degree in Bachelor of Medicine and Bachelor of Surgery, and postgraduate training in clinical epidemiology and biostatistics. Currently, John continues to pursue his NIH Fogarty sponsored PhD study training with a focus on understanding the common genetic variants and Killer cell immunoglobulin receptor genotypes among children with varying degrees of HIV disease progression in African cohorts from Uganda, Eswatini and Botswana.

POSTER

Significant attenuation of genetic association signals by fine scale population structure in Cameroon

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Large-scale human genetic studies of malaria in Africa have been generally successful. However, current observations have barely matched expectations; the extensive genetic diversity within the continent having hampered association analyses. In their series of multi-site studies across sub-Saharan Africa, the MalariaGEN observed specific limitations and encouraged country-specific analyses. Here, we investigated the impact of recently characterized fine-scale population structure on large-scale genetic association analyses in Cameroon using the set of genotype data from 1,029 Cameroonian participants that contributed to MalariaGEN consortium projects. We utilized public imputation resources and mixed model approaches to assess the possibility of improving association signals for our highly structured population, and we investigated the possibility of differential

evolutionary paths among Cameroon's major ethnic groups due to malaria, which may impact joint genetic association analyses. Our study confirms significant attenuation of association signals due to ethnicity. We found signals in potentially novel malaria protective loci; SOD2 ($P = 3.85e-09$; FDR = 0.011; OR = 0.31; 95%CI = 0.21 – 0.45; $\lambda_{GC} = 0.99$) specific to Cameroon's Semi-Bantu population, and CHST15 ($P = 1.04e-08$; FDR = 0.002; OR = 0.42; 95%CI = 0.31 – 0.56; $\lambda_{GC} = 1.02$) in the general population. We uncovered heterogeneity within the beta-like globin (HBB) gene cluster among Cameroon's major ethnic populations that underscores age-old fine-scale structure within the country. Together, our findings highlight population-specific variants and disparate genetic association patterns among Cameroon's major ethnic groups that should be important for future genetic association studies in the Country. Our findings also further the understanding of the evolutionary course of Cameroon's major ethnic populations under malaria pressure.

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POSTER

Evaluation of the Impact of the COVID-19 Pandemic on the Dynamics of Enrolment of TB Patients into Directly Observed Treatment (DOT) Program in Selected Diagnostic and Treatment Centres of the South West and Littoral Regions of Cameroon from 2019-2021

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INTRODUCTION: Despite progress made to achieve the WHO targets of the End TB Strategy, the emergence of the COVID-19 pandemic could pose new challenges for global and national TB control efforts. This study aims to evaluate the impact of the COVID-19 pandemic on the dynamics of enrolment of TB diagnosed patients into the DOT program in nine selected diagnostic and treatment centres of the South West and Littoral Regions from 2019-2021. **METHODS:** A retrospective cohort study was conducted on TB patients diagnosed and enrolled for treatment from February 2019 to March 2021. Data were entered and analysed using Microsoft Excel 2019 and SPSS version 25. Poisson regression analysis was used to compare trends during the pre-COVID-19 (February 2019 to February 2020) and COVID-19 (March 2020 to March 2021) periods. The Chi-square test was used to determine associations between independent and outcome variables, and P-values less than 0.05 were considered statistically significant. **RESULTS:** A total of 2503 TB patients were included in the study, with 1344 recorded during the pre-COVID-19 and 1159 patients during the COVID-19 periods. Males constituted a larger proportion of the study population (62.9%), and the majority (31.0%) were between 21-30 years old. The period during the COVID-19 pandemic significantly reduced the monthly number of TB patients diagnosed and enrolled for treatment by 0.79 ($p < 0.001$, CI: 0.73 - 0.85) and 0.84 ($p < 0.001$, CI: 0.78 - 0.91) times, respectively. There was a significant increase in the proportion of rifampicin-resistant TB from 1.6% during the pre-COVID-19 to 4.3% during the COVID-19 period ($p = 0.031$). We found a high proportion (22.7%) of TB patients co-infected with HIV. **CONCLUSION:** Urgent action and innovative solutions need to be implemented to catch up with the delays in diagnosis and treatment during the pandemic so that the world will not lose the achievements of ending the TB epidemic by 2035.

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I am Tiogouo Tezemene Kevine, holder of a BSc. in Medical Laboratory Science and an MSc student in Epidemiology and Control of Infectious Diseases from the University of Buea. I am passionate about research, interested in new findings. I am currently working on a research project entitled "Evaluation of the Impact of the COVID-19 pandemic on the Dynamics of Enrolment of TB Diagnosed Patients into Directly Observed Treatment (DOT) Program in Selected Diagnostic and Treatment Centres of the Southwest and Littoral regions of Cameroon". Imparting in the journey of health programs has always been my dream. I've acquired skills in data

collection, data entering, and management and I am looking forward to specializing in Humanitarian projects and integrating NGOs so as to contribute to promoting health, especially to the vulnerable.

SPEED TALK

Structural characterization of phosphatidylserine decarboxylase a promising therapeutic target and prediction of its potential inhibitors

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Plasmodium falciparum is a protozoan parasite responsible for the most severe and deadly form of malaria. The resistance of this parasite to last resort antimalarial drugs has been reported, so there is an urgent need to identify new therapeutic candidates for the development of new drugs. Bioinformatics is the most appropriate approach to predict therapeutic candidates in a reasonable time and at low cost. In this work, we propose Phosphatidylserine Decarboxylase (PSD) as a promising new target of *Plasmodium falciparum*. It is an enzyme extracted from the TDR Target database which facilitates the identification and prioritization of drugs and drug targets of neglected pathogens according to specific criteria.

It is a member of the lyase family, more precisely the carboxy-lyases, which cut carbon-carbon bonds. Phosphatidylserine decarboxylases (PSDs) catalyze the decarboxylation of phosphatidylserine to generate phosphatidylethanolamine, a critical step in phospholipid metabolism in prokaryotes and eukaryotes. The model of this protein not previously characterized, structurally opens the way to the design of new potential inhibitors for the development of future antimalarial drugs.

The main objective of this work is to build the 3D structure of PSD and to identify new inhibitors of this promising new therapeutic target.

The 3D structure of the target protein was predicted using the AlphaFold server and the ligands extracted from the zinc DB chemical library. The molecular docking was performed using autodock-vina. At the end of this study, we have identified ten (10) PSD inhibitors that should be studied experimentally to evaluate their antimalarial activities.

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I am Mamadou SANGARE holder of a master in Bioinformatics. I work on Computer-Aided Drug Design (CADD), specifically on the identification of therapeutic targets, molecular modeling, virtual screening and the study of protein interaction networks.

The topic of my master project in bioinformatics is the identification of a therapeutic target of *Plasmodium falciparum* and construction of its model by homology.

In this work, under the supervision of Dr. Cheickna CISSE, I established a list of potential therapeutic targets of *Plasmodium falciparum*, selected the most promising and built its model by homology, thesis publicly defended in January 2019.

More than two years ago, under the direction of Professor Mamadou WELE and Dr Cheickna CISSE, I was associated to a research project on "Prediction of the structural model of two parasitic malaria target proteins and insilico screening of their inhibitors",founded by the University of Sciences, Techniques and Technologies of Bamako within which my Master thesis was realized.

In the framework of this project, I benefited from an internship at the Laboratory of Bioinformatics, Biomathematics and Biostatistics of the Pasteur Institute of Tunis where for one month under the supervision of Dr. Alia Benkhala, I studied the Protein-Protein interaction of *Plasmodium falciparum* in order to establish a list of proteins involved in these interactions and to select promising therapeutic targets of the parasite. This work is currently submitted for publication.

I have also done several internships and trainings, including in medical biology and research laboratories, participated in scientific conferences and obtained awards such as:

2016-2018 Participation in the ninth and tenth edition of the Malian Symposium of Applied Sciences. (MSAS); Bamako, Mali

Oral communication: "Identification of a therapeutic target of *Plasmodium falciparum* and construction of its model by homology", first symposium of the African Center of Excellence in Bioinformatics (ACE-B), Bamako, Mali, 11 -12 March 2019.

Submission: "Prediction of the structural model of two parasitic malaria target proteins and insilico screening of their inhibitors".

2020: Identification of a therapeutic target of Plasmodium falciparum and construction of its model by homology
Published in F1000

June 15-17, 2021: Participation in the second annual symposium of the African Center of Excellence in Bioinformatics on the topic Bioinformatics and Data Science for Global Health.

Poster presentation on the topic: Phosphatidylserine decarboxylase, a promising therapeutic target for Plasmodium falciparum.

July 13, 2021 - October 12, 2021: Teaching assistant - H3ABioNet Introduction to Bioinformatics course.

October 7, 2021 - November 25, 2021: Teaching Assistant H3ABionet Training Course.
(INT-IBT_2021)

Honors

2018 : H3abionet Scholarship

November 10 -16, 2019: Best poster award at the ISCB-Africa, ASBCB conference on Bioinformatics.

July 2020: PhD scholarship funded by the West African Center of Excellence for Global Health.

Research training in bioinformatics through grant #5U2RTW010673.

Currently a PhD student in Bioinformatics at the Doctoral School of Sciences and Technologies of Mali (EDSTM) of the University of Sciences, Techniques and Technologies of Bamako (USTTB),

I am working on the characterization of new therapeutic targets of Plasmodium falciparum and the prediction of their potential inhibitors.

This thesis project is funded by the West African Center of Excellence for Global Health. Research training through grant # 5U2RTW010673.

SPEED TALK

Impact of next-generation sequencing (NGS) on HIV-1 drug resistance testing among patients experiencing virological failure at the time of therapy switching in Uganda

Maria Magdalene Namaganda¹, Hakim Sendagire^{1*}, Moses Nsubuga Luutu¹, Isaac Ssewanyana², Chris Okirya² Susan Nabadda², Edgar Kigozi¹, Fred Ashaba Katabazi¹, Jupiter Marina Kabahita^{5,1}, Fredrick Elishama Kakembo⁴, Stephen Kanyerezi⁴, David Patrick Kateete¹, Andrew Kambugu³, Barbara Castelnuevo³, Moses L Joloba^{1,5}, Gerald Mboowa⁴

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Background: The emergence and spread of antiretroviral drug resistant HIV-1 variants is one of the major factors responsible for therapeutic failure in persons living with HIV (PLWH) as it jeopardizes the efforts to reduce the progression of AIDS. While Sanger sequencing is the conventional method for HIV drug resistance testing, it is unable to detect low-abundance variants. This study assessed the impact of next-generation sequencing (NGS) on drug resistance testing before switching of therapy.

Methods: Blood samples were collected from 60 PLWH to detect for the presence of low-abundance drug resistance variants using NGS of HIV protease and reverse transcriptase genes. Sanger sequencing was performed for all the samples for validation purpose. We used the HyDRA bioinformatics pipeline was used to analyze for the drug resistance mutations and Stanford HIV drug resistance database for interpretations of the variants.

Results: Out of the 60 samples, 58 had complete sequence data and were considered for analysis. Low-abundance HIV drug resistance variants were identified in 38 out of the 58 samples (65.5%). Overall, we found 757 variants from the NGS data and 90 variants from the Sanger sequencing data sets. The most prevalent minority variants present in the samples were K65R (65.5%), K14R, K45R, L63P and I64V identified in 63.79% of the samples and I15V, K70R, V77I and L283I identified in 60.3% of the samples.

Conclusion: Findings from the study reveal that more than half (65.5%) of the sampled population harbored low-abundance HIV-1 variants, most of which were previously reported to cause virological failure and consequently antiviral drug resistance.

Key words: HIV, Drug resistance testing, Low-abundance (minority) variants, Sanger sequencing, Next-generation sequencing

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My name is Maria Magdalene Namaganda, a finalist graduate student Masters of Science in Bioinformatics at Makerere University. I hold a Pre-Doctoral in Bioinformatics from the University of Georgia. I work as a Research Scientist at Molecular Biology Laboratory, Department of Immunology and Molecular Biology at Makerere University. I am very passionate about developing my career in Bioinformatics and its application in the Genomics of Infectious diseases that are still a major threat in Sub-Saharan Africa.

SPEED TALK**Popular perception on genomic studies and hereditary diseases in Ari Community, South Omo Zone, Ethiopia**

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Introduction: Advances in genetics and genomics offer many opportunities for improving health and moving towards the era of precision medicine. Uptake of these advances would, depend on the public's understanding of genetics and their perception towards integrating genetics and genomic technologies into individual care and public health measures. Genomic research becomes mainstay in Africa and many countries are warming up towards integrating genomics and genetics into healthcare and public health. It is important to gauge public understanding and perspective towards genetics and genetic medicine. We, therefore, designed a qualitative study to explore public understanding and attitude to genomic research in Ethiopia prior to starting participant enrollment for a main study that aims to unravel susceptibility to Tuberculosis (TB) in Africa. Method: The study setting was South Ari district of the South Omo Zone, southern Ethiopia. Using In-depth Interviews and Focus-Group Discussions, we explored perception and awareness of genetics, genetic diseases and genomic research with members of the community elders, health officials/health extension workers, TB patients and apparently healthy individuals. Data analysis was done using MAXQDA (version 2020). Results: Elephantiasis, asthma, epilepsy, goiter, TB, leprosy, lymph node swelling and umbilical hernia were considered as hereditary in the community. This was because they suspect aggregation of affected individuals in certain families. The community explained inheritance concepts using phenotypic traits encountered in their daily life such as polydactylism and hyperpigmentation of tongue. With the exception of Asthma, the community had limited understanding on the role of gene-environment interaction in shaping one's health, as often they will say that in asthma, their disease symptoms improved upon changing residential locations. Despite their understanding of the role of genetics in health, many of them expressed reservations towards participating in a genomic study because they are afraid that such studies will scientifically prove the genetic basis of diseases that are considered stigmatizing in the community. Conclusion: This study shows limited understanding in the community about what disease conditions are hereditary. Therefore, there is a need for public awareness programs on genetics and genomics and its role in health and healthcare.

Key words, Genomic, Genetics, Heredity, Inheritance, Stigma

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Melaku Tilahun is an associate researcher at the Armauer Hansen Research Institute (AHRI). He received a BSc degree from Jimma University in Medical Laboratory Science and completed his MSc Degree in Clinical Laboratory Science with Diagnostic and Public Health Microbiology Specialty in Addis Ababa University, Ethiopia. He has worked as a Laboratory Manager of the Institute. During his stay at the institute, he was participating in different research projects and he has published his works in different open-access peer-reviewed journals. Currently, he has joined Arba Minch University, Ethiopia to study his Ph.D. Degree in the field of Biotechnology with a specialty of Medical Biotechnology. Melaku's main research interest is studying the epidemiology and

genomics of M.tuberculosis to understand the transmission dynamics and the genetic basis of antibiotic resistance and to provide information for current discrepancies between genotype and phenotype drug resistance.

SPEED TALK

Profile of individual telomere length of a rare chromosomal abnormality in Chronic Myeloid Leukemia in Mali.

INTRODUCTION: Chronic myeloid leukemia (CML) is characterized by the presence of the reciprocal translocation t(9;22)(q34;q11), resulting in a BCR/ABL1 gene fusion on the derivative chromosome 22 called the Philadelphia (Ph) chromosome. CML progresses from a chronic phase (CP) to an accelerated phase (AP), and/or a blast phase (BP) when diagnosed late or untreated. This progression is frequently preceded or accompanied by recurring secondary chromosomal abnormalities which are believed to play a role in the transformation of different phases. In Western countries, the atypical Philadelphia chromosome accounts for less than 5% of CML patients, but they account for 25% of the 101 CML cases in our cohort. Moreover, telomere length shortening is known to be associated with disease progression. The dynamic length changes of individual telomeres might be involved in the remodeling of the genome of the CML cases presenting atypical Philadelphia chromosome. Therefore, it is essential to know the individual telomere length of CML patients presenting atypical translocations. **OBJECTIVE:** The goal of this study is to determine the length of telomeres on each chromosomal arm of CML patients presenting atypical Philadelphia chromosome. **METHODOLOGY:** We used Cy3 labeled peptide nucleic acid probes specific for (T2AG3)₃ sequences as well as centromeric probes as an internal control to perform quantitative fluorescence in situ hybridization (Q-FISH). Following the microscopic acquisition of Q-FISH pictures with Metasystem (Isis), multicolor FISH was performed on the same slide (M-FISH). The Q-FISH helps measure individual telomere length while M-FISH enables characterization of chromosomal translocation. **RESULTS:** We report a rare case presenting an atypical Philadelphia chromosome and a pericentric inversion inv(9)(p22q34) involving the derivative chromosome 9 that resulted from t(9;22)(q34;q11.2) in a patient with Ph-positive CML. We found that the chromosome 1 present the highest intensity 98 of telomere while the shortest was the chromosome 22 with 43. The derivative chromosome 9 presenting the inversion has a lowest intensity 99 of telomere, compare to its homologue the normal. **CONCLUSION:** We reported here a rare chromosomal abnormality in CML and the dynamic changes of individual telomere length in atypical translocations may be associated with primary and secondary therapeutic resistance in CML.

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Mr. Modibo K Goita received his baccalaureate in Exact Sciences from the High School of Yorosso in 2013. He began a doctoral training at the Faculty of Pharmacy (FAPH) of the University of Sciences of Techniques and Technologies of Bamako (USTTB) in 2014. He was then recruited as an intern at the Laboratory of Neurogenetics of the same university with funding from Human Heredity and Health in Africa (H3Africa). His thesis work focused on the assessment of telomere length during myeloid chronic leukemia in Mali. He is also pursuing a master's degree in Bioinformatics at the African Center of Excellence in Bioinformatics of Bamako (ACE-B) of USTTB. He is expected to graduate in 2022.

SPEED TALK

The role of copy number variants in the aetiology of developmental disorders in South Africa - a whole exome sequencing study

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INTRODUCTION: Developmental disorders are rare conditions, causing a mental or physical impairment. Genetic heterogeneity and highly variable clinical manifestations poses great challenges in the diagnosis of developmental disorders. New sequencing technologies have become an important and cost-effective tool in the diagnosis of rare diseases and results can impact clinical practice significantly. The Deciphering Developmental Disorders in Africa (DDD-Africa) study aims to recruit and perform detailed clinical phenotyping and whole-exome sequencing (WES) on 500 African patients with a developmental disorder. No genetic diagnosis has been made in any of these

patients. Copy number variants (CNVs) are responsible for a large proportion of the genetic aetiology of developmental disorders. The introduction of exome sequencing has allowed the detection of CNVs and single nucleotide variants exome wide with a single test. This would be a valuable approach to implement especially in a limited resource setting. The availability of CNV detection tools is increasing, however, at present there is no gold standard for WES-based CNV detection.

AIM: This study aims to identify the most appropriate bioinformatics approach to detect CNVs from exome data. Subsequently, it will be implemented in a developmental disorder variant analysis pipeline for WES data generated by the DDD-Africa study, to establish the role that CNVs play in this African cohort.

METHODOLOGY: InDelible is used to identify structural variation (SV) from WES by using split-read information and was incorporated into our first batch of WES samples (n=288) from South Africa. This includes 60 trios, 46 duos, three extended families and six singletons of patients with developmental disorders. All six primary steps namely, fetch, aggregate, score, blast, annotate and denovo were processed using the CRAM files of these samples.

PRELIMINARY RESULTS AND NEXT STEPS: Findings from InDelible are still being finalised although we have successfully implemented this tool into our dataset of 288 samples. In-depth filtering and data analysis are being carried out to identify putative disease-causing SVs in our dataset. Additional bioinformatic approaches will be incorporated to select the best tool or combination of tools for CNV detection in our African patient cohort.

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Studied Human Genetics undergraduate and Honours at University of Pretoria. After Hons she was appointed as Junior Laboratory Assistant in the Human Genetics section of UP and started a Master's degree which was completed in 2014 whilst working as a lab assistant. In 2018 she was appointed as Research Assistant for the DDD-Africa project and has since changed roles to project coordinator and researcher on the project located in Johannesburg, South Africa. She is currently pursuing a PhD degree in Human Genetics with the University of the Witwatersrand.

SPEED TALK

Apolipoprotein L1 variants and Preeclampsia

Nana Yaa Achiaa Karikari Agyemang^{1,2,3}, Christian Obirikorang³, Linda Ahenkorah Fondjor³, Rachael Abena Bonkuah Duodu^{1,2}, Miriam Gakpey Lawson^{1,2,4}, Mona-Liza Ebiere Sakyi^{1,2,4}, Alexander Nyarko^{1,2}, Anita Ghansah^{1,2}, Charlotte Osafo¹, Graham Lipkin⁵, Dwomoa Adu^{1,4}, H3AKDRN H3AKDRN1, NMIMR2, KNUST³, WAGMC⁴, University Hospitals Birmingham NHS Foundation Trust⁵

Background

Maternal mortality is up to 50 times higher in sub Saharan Africa (SSA) than in Europe or the USA.¹ More recently in Ghana, hypertensive disorders of pregnancy have overtaken obstetric haemorrhage as the major cause of maternal death.² Pregnant African Americans carry more than a 2-fold higher risk of PET as compared to non-Hispanic Whites (6.04% to 3.75%) and Hispanic Americans (6.04% to 2.58%)($P < 0.0001$).³ The high incidence of PET in African Americans and African blacks suggest that these two populations may share common genetic predispositions for such. One possible contributor to preeclampsia are variants in the apolipoprotein L1 gene which are found only in people of African origin and encode for apolipoprotein L1 (APOL1). The G1 (Ser342Gly or rs73885319) and G2 (rs71785313) variants of APOL1 are a major risk factor for non-diabetic kidney disease in African Americans and Africans.^{4,5} Studies, mostly of African Americans in the USA have suggested that foetal but not maternal APOL1 variants are associated with PET.^{6,7}

Methods: We studied a total of 53 women with PET and 53 pregnant healthy controls. Isolated genomic DNA was multiplex-PCR amplified and Ligation-Detection Reaction assayed (LDR) for single-base variants. Gel electrophoresis separation identified SNP-variant ligation products APOL1 G1 (rs60910145, rs73885319), G2 (rs71785313). Association between APOL1 variants and PET was determined using the chi square test and odds ratio was calculated to determine strength of association. 17% of cases with PET had 2 APOL1 high risk alleles as did 13.2% of controls (OR 1.08; CI: 0.37 to 3.18)

We report that in a small sample, maternal APOL1 variants do not influence the risk of PET. Our further studies will investigate the role of foetal APOL1 variants in the risk of PET.

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I have a first degree in Biochemistry from the University of Cape Coast and currently I am a student at the Kwame Nkrumah University of Science and Technology reading MPhil Molecular Medicine. I am a Senior Research Assistant at the Noguchi Memorial Institute for Medical Research, University of Ghana Legon and a team member of H3Africa Chronic Kidney Disease Research Network. My research focuses on the use of genetic mapping to understand and explain diseases or conditions of multifactorial origin specifically Pre-eclampsia during pregnancy. Becoming a biomedical research scientist employing Human medical genetics, molecular biology and evolutionary biology in the design of alternative drug therapy and disease control is my sole research goal specifically using Genomics as a prime focus in translational medicine of non-infectious diseases. My hope is to gain training, research experience and expertise for further studies. This in the near future, I believe would help me become an esteemed research scientist and a lecturer, to equip me in the transfer of knowledge and skills to early career scientists.

Joining policy bodies like the Ghana Health Service (GHS), the World Health Organization (WHO) or other humanitarian organizations to influence worldwide policies to provide efficient and alternative strategies to control and eliminate diseases through innovative techniques and methodologies that are simple, handy and efficient, forms the core of my career objectives

SPEED TALK

Comprehensive Benchmarking of Copy Number Variants Detection Tools Using Whole-Exome Sequencing **Nicholas Opoku (1), Sampson Pandam Salifu (1), Peter Amoako-Yirenkyi (1,2), Henry Martins (1,2)**

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Background: Copy number variation (CNV) has gain significant attention as a kind of genomic variation that contributes to complex phenotypes and disease vulnerability. Among the various approaches in detecting these CNVs, whole exome sequencing is an attractive alternative to both whole-genome sequencing and microarray analysis because of its cost effective nature as well as potential ability to detect CNVs ranging from 1 exon to several mega bases. After the development of next-generation sequencing (NGS) technologies, several tools have been developed in an approach that facilitate the effective detection of CNVs. Due to the number of CNVs that have been developed it becomes necessary to help investigators choose a suitable method based on their objectives to aid in detecting CNVs.

Method: The study performs a review on commonly used CNV detection applications based on read-depth methods, benchmarking the applications by the number of CNVs detected, strength, weaknesses and computational demands. Eight published CNV prediction tools (i.e., cn.MOPS, CODEX, exomeCopy, Canoes, cnvkit, ExomeDepth, deanncnv and contra) and the BAM format of 10 exome datasets mapped to GRCh37 decoy reference genome from Nigeria and Gambia downloaded from the Phase 3 of the 1000 Genomes Project were used.

Findings: Our review is part of the limited number of benchmarking done using datasets from Africa and provides a comprehensive performance evaluation for these selected CNV detection methods. Majority of CNVs detected were found in Nigerian dataset compared to their Gambian counterpart. The best method for detecting CNVs were Canoes, exomeCopy and cn.MOPS. Most cnvs detected were deletions rather than duplication. Although, overlappings were observed among samples for various genomic positions, each algorithm had a certain range of detected lengths. Upon unified comparison, the tools were not equivalent.

Conclusion: The analysis performed allows choosing algorithm(s) most suitable for a specific goal and further facilitate future development and improvement in CNV prediction methods.

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Nicholas Opoku is a Bio-data Analytics and Computational Genomics Ph.D student at the Kwame Nkrumah University of Science and Technology. Before his enrollment, he launched his career as a research assistant for both AIMS-Ghana and International Development for Mathematical Sciences (IDEMS); an international organisation. During this period Nicholas held various key positions which increased his researching skills paving way for him to publish his works in reputable journals. Nicholas is a graduate from both the African Institute for Mathematical Sciences and University of Cape Coast where he holds a masters degree in Mathematical Sciences. On the surface Nicholas is a trained mathematician but knowing there is no single approach that works for everyone, he continues to educate himself on emerging courses to which he seek to transfer this experience into his new field where he plans on developing an algorithm for the detection of copy number variations which is an important evolutionary force and have been associated with complex disease susceptibility.

SPEED TALK

Current status of molecular markers of *Plasmodium falciparum* resistance to Sulphadoxine-Primethamine (SP) and the effectiveness of IPTp-SP among pregnant women in Mutengene, Southwestern, Cameroon.

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INTRODUCTION: Accumulating resistance to Sulphadoxine-Primethamine-intermittent preventive treatment in pregnancy (IPTp-SP) is partly attributed to its failure to improve malaria outcomes in pregnancy. As resistance continues to accumulate, ineffectiveness of IPTp-SP is inevitable. Routine monitoring of parasite strains for the effectiveness of antimalarial is therefore crucial, in disease-endemic areas, for early detection of reduced parasite susceptibilities and emerging drug resistance.

AIM: This cohort study assessed the current status of pfdhps and pfdhfr resistance markers to SP and the effectiveness of IPTp-SP in Pregnant women.

METHODS: Venous blood was collected and malaria parasite infective determined by blood mRDT and light microscopy. DNA was extracted by Chelex-100 method and nested PCR for *P. falciparum* detection. Single Nucleotide Polymorphism genotyping for Pfdhps and Pfdhfr resistant markers in positive *P. falciparum* samples was then undertaken by nested PCR followed by allele-specific restriction analysis (ASRA).

RESULTS: The proportion of malaria parasite infected from 300 pregnant women determined by microscopy, mRDT and PCR was 12.9%, 16.4% and 29.4% respectively and a very low-density infection, averaging 271 parasites per microliter of blood. The proportion of infection was significantly reduced in IPTp-SP users compared to non-users with a significant association ($p=0.001$) between the malaria parasite status by microscopy and IPTp-SP dosage.

ASRA shows that this population harbour 100% mutant alleles for dhfr S108N, 53.9% dhps A581G mutants while dhps K540E substitution was absent.

CONCLUSION: These results suggest an increase prevalence of these molecular markers associated with *P. falciparum* resistance to SP with implication for IPTp-SP in Mutengene, Southwest Cameroon.

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PhD student and Graduate Teaching Assistant Department of Biochemistry and Molecular Biology and a Junior data analysis for PAMGEN-University of Buea, Southwest, Cameroon Ntui Vincent Ntui-Njock, a PAMGEN fellow and PhD student and Graduate Teaching Assistant Department of Biochemistry and Molecular Biology is a holder of an MSc in Molecular Biotechnology and a BSc in Biochemistry at University of Buea. He is currently executing his research in our laboratories at MRC Unit

the Gambia thanks to the excellent molecular biology and genomic laboratory platforms. His current research focuses on Specific strains of *P. falciparum* that persist during pregnancy and IPTs-SP intake and adverse malaria effect in pregnant women and infants in Cameroon. Within the framework of his Master's degree, he had an intensive clinical and community field and laboratory work and analytical experience working on "known molecular markers of *P. falciparum* resistance to Sulphadoxine Pyrimethamine". Ntui had been invited to report for MESA during BioMalPar 2020 and ASTMH 2020 conferences. He is a highly motivated and optimistic PhD Biochemistry student looking forward to a long and productive career as a young African research scientist in malaria with interest in Malaria Epidemiology, parasite diversity and drug resistance.

SPEED TALK

Risk scoring model for predicting Hypertension among indigenous Africans: Findings from the SIREN study

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Background & Objective: Hypertension is one of the dominant risk factors for cerebrovascular and cardiovascular diseases, and over 65% of the global burden of hypertension occurs in low-and-middle-income countries. Early identification of populations at risk of hypertension is an effective mechanism in reducing the burden of hypertension. However, the paucity of cohort studies in Africa has limited the development of a predictive tool for identifying populations at risk of hypertension in this region. This study is aimed at developing a hypertension risk-scoring model for Africans.

Methods: We used 4390 population-based, stroke-free controls from the SIREN study to develop and validate an hypertension risk scoring model for Africans. 80% of the data was used for training the model, while 20% was used for validation. Fifteen risk factors were used to predict hypertension using multivariable logistic regression, and the beta coefficient of significant variables was weighted using a constant and standardized weighting procedure. A standardized score was generated between 0-100%. The risk-score cut-off point was estimated using the receiver operating characteristics curve (ROC), sensitivity, specificity, positive predicted value (PPV), negative predicted value (NPV), and the Cohen's kappa value at $P < 0.05$.

Results: Standardized and constant weight was applied to eight significant risk factors, and the risk-score cut-off was 56% at a maximum Cohen's kappa value of ≥ 0.64 . The model performance had a ROC of 92.0% (95%CI: 91.0, 93.0), sensitivity of 79.2%, specificity of 94.4%, PPV of 95.2%, and NPV of 76.7% in the validating set. Similarly, the male and female stratified models had a similar cut-off of 56%, and the model predictive accuracy was 92.0% (95%CI: 89.0, 95.0), and 91.0% (95%CI: 88.0, 94.0) for the male and female samples, respectively.

Conclusion: The hypertension risk-scoring model was robust and commendable with high predictive accuracy. This model will be useful for the timely detection of persons at risk of hypertension and could aid the development of interventions to mitigate the burden of hypertension among indigenous Africans.

Keywords: Hypertension, Risk-score, Predictive accuracy, Indigenous Africans, SIREN

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I am Osahon Jeffery Asowata from Nigeria, A Biostatistician. I am currently on the SIBS Genomics and CVD working group project at the college of Medicine University of Ibadan.

SPEED TALK

De novo variant in PAX3 gene in a Malian family with Waardenburg syndrome type 1

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Backgrounds: Waardenburg syndrome (WS) is one of the most common type of syndromic hearing impairment (HI), accounting for 2 to 5% of all congenital HI. Only four genes are known to cause WS, mainly found in population with Caucasian ancestry. We report here the first genetically confirmed case from Mali, one of the rare cases from sub-Saharan Africa.

Objectives: To clinically characterize patients with Waardenburg syndrome, and to identify the underlying genetic defects.

Methods: Written consent was obtained from all participants before their enrollment. Patients were examined by a multidisciplinary team including ENT specialists and neurologists. Then, we performed a pure tone audiometry (PTA) and classify the severity according to the Bureau International of Audiophonology (BIAP). DNA was obtained for genetic analysis.

Results: Two individuals, one male and one female from two unrelated families and their relatives were enrolled with the age of 5 and 12, respectively. There was no familial history of WS, and pedigree analysis was consistent with sporadic cases. Clinical examination showed type I in the female and type II in the male patient. Audiometry performed in the probands confirmed the bilateral and sensorineural HI in both patients. WES found a pathogenic, de novo variant in the PAX3 gene (c.C664T; p.R222X) in the female and no candidate variant in any known gene associated with WS in the male. In silico analysis confirmed deleteriousness with CADD=37.

Next steps: Sanger sequencing is ongoing for segregation analysis, and WGS and functional studies are planned.

Keywords: Waardenburg syndrome, de novo mutation, Mali, Africa.

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I am a graduate student and resident At the Neurology clinic/Neurogenetic Departement at the Teaching Hospital of Point G, Bamako, Mali.

SPEED TALK

Study of the Nuclear Organization of Telomeres in Urinary Circulating Cells in Bladder Cancer

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Background: Bladder cancer (BC) is caused by an excessive proliferation of abnormal cells in the lining of the bladder, and the Schistosoma haematobium infection is its major cause in Mali. BC is one of the most common cancers and the first cause of death by cancer in urology worldwide. The poor prognosis of patients with bladder cancer could be due to the presence of aggressive forms associated with genomic instability which can be determined by the telomeric nuclear organization of cancer cells studies.

Objectives: To assess the nuclear organization of telomeres in the urinary circulating cells of patients suffering from bladder cancer.

Methodology: We enrolled bladder cancer patients. We carried out cytological preparation from their urine samples. Then, we did the 3-Dimensional (3D) fluorescence in situ hybridization by using the Cy3 labeled peptide nucleic acid probes specific for telomeric. Then, we captured 3D images of the cancer and the normal cells, and assessed the parameters defining the telomeric nuclear organization of cancer and normal cells by the TeloView software.

Results: We observed a difference between all of the telomeric parameters of cancer and normal cells. For instance, cancer cells had more telomeres and telomeric aggregates, but were shorter telomeres than normal cells. The nuclei of tumor cells were twice as large as those of normal cells. The telomere nuclear distribution index, which corresponds to the cell proliferation index, was also twice higher in cancer cells compared to normal cells. Also, the telomeres of cancer cells had a more central nuclear location than those of normal cells. Furthermore, we found that cancer cells were more heterogeneous than normal cells. Finally, we found a trend towards an association between the advanced stage of bladder cancer and the level of alteration of telomere nuclear organization.

Conclusion: This is the report of the telomeric nuclear organization of urinary circulating cells in bladder cancer, and the telomeric profile of those cells has a potentiality of being a biomarker which could be a decisive turning point in the management of this pathology in Mali and even elsewhere.

Keywords: Bladder cancer, Telomeres, 3D FISH, Mali.

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I am a graduate student and research assistant at the oncogenetics/neurogenetics laboratory. I have been working on the genomic instability of bladder cancer in the Malian population since my internship in 2019.

SPEED TALK

Menopause, HIV and Cardiometabolic Disease Risk: A Cross-Sectional Analysis on Midlife Women from Kenya and South Africa

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Introduction: Menopause and HIV infection are both associated with cardiometabolic disease (CMD). In sub-Saharan Africa (SSA), the health transition, access to anti-retroviral therapy (ART) and improved life expectancy is mirrored by a growing population of midlife women with HIV. It is therefore important to examine the effect of HIV infection on CMD risk in these women during the menopause transition. The aims of this study were to compare CMD risk factors between midlife SSA women with and without HIV and to determine whether the differences in these risk factors between pre- and postmenopausal women were modified by HIV infection.

Methods: The analysis included 1665 HIV negative (733 pre- and 932 postmenopausal) and 414 HIV positive (211 pre- and 203 postmenopausal) women between the ages of 40-60 years from the Africa Wits-INDEPTH partnership for Genomics studies (AWI-Gen) at three sites in Kenya (Nairobi), and South Africa (Soweto and Dikgale). Demographic, anthropometric and cardiometabolic variables were compared between the HIV-positive and HIV-negative women and between pre- and postmenopausal women according to HIV status using multivariable linear regression analyses.

Results: The mean age and HIV prevalence in the study population were 49.1±5.8 years and 19.9% respectively. Age at menopause was lower in HIV-positive compared to HIV-negative women (48.1±5.1 vs 50.9±4.7 years, $p<0.001$) after adjusting for confounding variables.

Compared to the HIV negative women, those with HIV had a lower hip circumference ($\beta = -1.0$, $p=0.002$), BMI ($\beta = -3.4$, $p<0.001$), cIMT ($\beta = -0.14$, $p=0.02$), diastolic ($\beta = -2.0$, $p=0.01$) and systolic ($\beta = -0.04$, $p<0.001$) blood pressure, but elevated triglycerides ($\beta = 0.08$, $p=0.002$) and insulin resistance ($\beta = 0.14$, $p=0.02$).

When comparing CMD risk factors between pre- and postmenopausal women in those with or without HIV infection, only LDL-cholesterol levels differed between menopausal groups being higher in post- than premenopausal women ($\beta = 0.13$, $p=0.03$) but only in those without HIV infection.

Conclusion: HIV infection is associated with earlier age at menopause and has differential associations with CMD risk factors in midlife SSA women. Differences in the levels of CMD risk factors between pre- and postmenopausal women are similar in those with or without HIV infection, with the exception of LDL-cholesterol.

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University of the Witwatersrand

A persistent young scientist with a strong orientation on achieving goals. Enthusiastic about developing as a young scientist by interacting with different professionals and using skills obtained during my education to provide a valuable contribution to the community. Currently a PhD student at the University of Witwatersrand, Johannesburg, South Africa.

SPEED TALK

Evaluating the accuracy of genotype imputation in the Major Histocompatibility Complex (MHC) region in selected African populations.

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Genome-wide association studies typically use genotyping arrays to genotype large sets of individuals and thus determine which SNPs are significantly overrepresented in the cases compared to the controls and thus determine association with disease. Genotyping arrays are cheaper than sequencing, but only measure selected SNPs across the genome. To increase the number of SNPs, genotype imputation is performed. Some regions within the human genome such as the Human Leukocyte Antigen (HLA) region, are highly variable and thus difficult to impute. The HLA region plays an important role in autoimmune and infectious diseases. In view of this, it is important to evaluate the accuracy of HLA imputation, especially in African populations as they have high diversity, and this has not been extensively studied. The aim of this study was to therefore evaluate the accuracy of HLA imputation in selected African populations. The study sets were selected from the Gambian individuals within the GGVP datasets. The Illumina Omni 2.5 array and H3Africa array data were inferred from the GGVP datasets using matching markers. The reference datasets were chosen from the 1kg-All, 1kg-Afr, 1kg-Gwd and H3Africa populations. HLA-A, HLA-B and HLA-C alleles were imputed using HIBAG and SNP2HLA while HLA SNPs were imputed using Minimac4, IMPUTE5 and SNP2HLA imputation tools. The assessment metrics were concordance rate and squared Pearson correlation coefficient. The most preferable software was HIBAG for HLA alleles imputation and IMPUTE5 for HLA SNPs imputation. The 1kg-All reference panel was the best performing reference panel for HLA alleles imputation implying that the reference panel sample size influences HLA alleles imputation. For HLA SNPs imputation, the 1kg-Gwd reference outperformed the other reference panels depicting that population specificity is key when imputing HLA SNPs. The H3Africa array and Illumina Omni 2.5 array performance were comparable for both HLA alleles and HLA SNPs imputation showing that genotyping arrays have less influence on HLA imputation in African populations. HLA SNPs with low minor allele frequencies (MAF) were imputed less accurately suggesting the need to build new algorithms and larger population-specific reference panels with an aim of improving imputation of HLA SNPs with low MAF.

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Nanjala Ruth is an Eastern Africa Network for Bioinformatics Training (EANBiT) fellow. She presently leads the Bioinformatics mentorship and incubation program at the International Centre of Insect Physiology and Ecology (ICIPE), Kenya. She pursued an M.Sc. in Bioinformatics at Pwani University, Kenya and the University of Cape Town, South Africa. Her Masters' research focused on evaluating the accuracy of genotype imputation in the Major Histocompatibility Complex (MHC) region in selected African populations. She holds a B.Sc. degree in Microbiology from Pwani University where she graduated with a first-class honors degree. Ruth is interested in human genetics and genomics research, with particular focus on applying bioinformatics principles to understand the mechanisms underlying human diseases, especially diseases such as cancer which claims many lives in Africa. She hopes to work towards addressing the gap in disease diagnoses, prognosis and treatment and thus improve the health and lives of others.

SPEED TALK

Comparative genomics, antibiotic resistance and virulence determinants of clinical *Pseudomonas aeruginosa* strains in Kenya

Shahiid Kiyaga(1), Cecilia Kyany'a(2,3), Angela W. Muraya(4), Hunter J. Smith(2), Emma G. Mills(5), Caleb Kibet(6), Gerald Mboowa(1,7) & Lillian Musila(2,3*)

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Pseudomonas aeruginosa is a leading cause of nosocomial infections worldwide. It can produce a range of debilitating infections, have a propensity for developing antimicrobial resistance, and present with a variety of potent virulence factors. This study investigated the sequence types, phenotypic antimicrobial susceptibility profiles,

and resistance and virulence genes among clinical isolates from urinary tract and skin and soft tissue infections. Fifty-six *P. aeruginosa* clinical isolates were obtained from six medical centers across five counties in Kenya between 2015 and 2020. Whole-genome sequencing was performed to conduct genomic characterization, sequence typing, and phylogenetic analysis of the isolates. Results showed the presence of globally distributed high-risk clones (ST244 and ST357), local high-risk clones (ST2025, ST455, and ST233), and a novel multidrug-resistant (MDR) clone carrying virulence genes (ST3674). Furthermore, 31% of the study isolates were found to be MDR with phenotypic resistance to a variety of antibiotics, including piperacillin (79%), ticarcillin-clavulanic acid (57%), meropenem (34%), levofloxacin (70%), and cefepime (32%). Several resistance genes were identified, including carbapenemases VIM-6 (ST1203) and NDM-1 (ST357), fluoroquinolone genes, *crpP*, and *qnrVCi*, while 14 and 22 different chromosomal mutations were detected in the *gyrA* and *parC* genes, respectively. All isolates contained at least three virulence genes. Among the virulence genes identified, *phzB1* was the most abundant (50/56, 89%). 21% (12/56) of the isolates had the *exoU*⁺/*exoS*⁻ genotype, while 73% (41/56) of the isolates had the *exoS*⁺/*exoU*⁻ genotype. This study also discovered twelve novel lineages of *P. aeruginosa*, of which one (ST3674) demonstrated both extensive antimicrobial resistance and the highest number of virulence genes (236/242, 98%). Although most high-risk clones were detected in Nairobi County, high-risk and clones of interest were found throughout the country, indicating the local spread of global epidemic clones and the emergence of new strains. Thus, this study illustrates the urgent need for coordinated local, regional, and international antimicrobial resistance surveillance efforts.

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I am an early career scientist/researcher passionate about Bioinformatics especially Microbial Genomics, Antimicrobial Drug Resistance, Infectious Diseases, and Disease modeling.

POSTER

Contribution of Biorepositories to Research: A Case Study of The Integrated Biorepository of H3Africa Uganda

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Background:

Biobanking is a field in science that has rapidly grown over the years, being employed for more than a century (Martina Siwek, 2015). In Biobanking, samples are collected processed stored, and later distributed to different end-users to answer various research questions (Vaught J 2016). Biobanks have a pivotal role in research advancement and have therefore evolved in response to the changing needs of investigators and projects that make use of the biobanks. Biorepositories meet the ever-growing need of researchers by providing highly efficient, safe, and secure capabilities for receiving, processing, storing and distributing biological specimens for clinical trials and scientific innovation. The data associated with stored biospecimens have also increased in complexity, encompassing many aspects of participant or patient information (De Souza YG and Greenspan JS. 2013). In Uganda, the Integrated Biorepository of H3Africa Uganda (IBRH3AU), located at Makerere University, College of Health Sciences, has played a pivotal role in the advancement of science and research in the country and as such laying a solid foundation for biobanking science in Uganda.

Problem statement/Gap: Biobanks have played and continue to play a big role in propelling research in many countries. The Integrated Biorepository of H3Africa Uganda which is the first state-of-the-art Biobank in Uganda is one of them. IBRH3AU has greatly contributed to research over the years from its establishment and this research, therefore, aims to document these contributions and further open the eyes of the scientific community to the value of this biobank.

Objectives: To document the specific contributions the biobank has made to the scientific research community in Uganda.

Methodology: We shall conduct a survey among different scientists/researchers that have made use of the IBRH3AU biobank using a data collection tool that will be designed. We shall also target students of Makerere University who have benefited from the existence of the biobank.

Next steps: Biobanking is a field that is sprouting and yet to reach its full potential in Uganda just as research continues to advance. We intend to continue documenting the successes of biobanking science in Uganda.

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I hold a Bachelor's of Science degree in Biotechnology from Makerere University and currently pursuing a Master's in Immunology and Clinical Microbiology still at Makerere University. I begun my career in biobanking in 2017 when I joined the team at the Integrated Biorepository of H3Africa Uganda where I currently work as the Biorepository Manager.

SPEED TALK

Participant withdrawals from an observational longitudinal study of the respiratory microbiota of Gambian children during a pandemic

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Background

Participant withdrawals during a prolonged longitudinal study can have significant implications on statistical analysis and study conclusions. A study was conducted on school-going children aged 5 to 14 years to determine the effect of air pollution on the pharyngeal microbiome. Commencing January 2019, the two-year recruitment of study participants was extended to three years due to the COVID-19 pandemic, with participant recruitment ending in March 2022. One of the main challenges encountered was withdrawal of participants after the lockdown. Mitigating this challenge was a crucial focus of research during the study.

Objective of the Main Study

To determine the effect of air-pollution on the bacterial respiratory flora in older children.

Methods

The study team numerated houses and generated a digital map. Field nurses collected nasopharyngeal, oropharyngeal, blood and urine samples from healthy children at eight time-points. Community sensitisations were carried out before and during the study to engage villagers. Study objectives were presented to local schools which assisted students undergoing collection of personalised air-pollution data. Parents were interviewed at various time-points during the study and also when withdrawal was requested.

Preliminary Results

547 children were sampled at the start of the study; whereas 400 (73%) were available for sampling during the final time-point. Out of 133 study withdrawals, 40 (30%) occurred pre-lockdown whilst 93 (70%) occurred post-lockdown. A total of 68 participants moved out of study area, 65 withdrew consent whilst 13 missed their sampling appointment. Reasons for withdrawals included childhood stigma associated with carriage of air-pollution device, and misinformation surrounding intravenous blood sampling and the COVID-19 vaccine. The most common reason for parental withdrawal was due to complaints about the duration of the study.

Conclusions

Investigating reasons behind withdrawal requests were crucial in mitigating participant withdrawals during the pandemic, though a small proportion of withdrawals provided no reason. Vaccine hesitation was found to negatively affect acceptance of intravenous sampling. Education of the populace through community sensitisation meetings proved essential in refuting misinformation.

Next Steps

The study plans to undertake metagenomic sequencing of pharyngeal samples and detection of biomarkers that indicate the presence of pollutants in the body.

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SPEED TALK

Identification of Hpas-associated SNPs in children on corticosteroid treatment

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3) Paediatric Endocrine Unit, Department of Paediatrics and Child Health, Stellenbosch University

Background: Corticosteroids are part of standard therapy for asthma management but can lead to hypothalamic-pituitary-adrenal suppression (HPAS). Biochemical evidence of HPAS is documented in about two-thirds of asthmatic children treated with various corticosteroids.

Aim: This study aimed to identify single nucleotide polymorphisms (SNPs) that may be protective against or causal of HPAS.

Methods: Ninety-six (96) asthmatic children on inhaled corticosteroids and nasal steroids underwent an overnight metyrapone test and their post-metyrapone adrenocorticotrophic hormone (PMACTH), 11-deoxycortisol, and 11-deoxycortisol + cortisol measured. HPAS was diagnosed based on three criteria: PMACTH < 106 pg/ml, 11-deoxycortisol (11DOC) < 208 nmol/l and 11DOC+C < 400 nmol/l. Fifty-five children who had measurements for all 3 variables were used in this analysis. Thirty-two (32) were suppressed (cases) while 23 were non-suppressed (controls) and made of 25 males and 30 females. DNA was extracted from saliva and whole exome sequenced on the Ion Torrent Sequencing platform. The SNPs were called with both Torrent Variant Caller and GATK algorithms. A set of candidate genes (470) were defined and SNPs affecting these genes selected for association analysis with both HPAS, BMI and BMI z-scores. A functional annotation and mapping (FUMA) tool was utilised to prioritize significant SNPs and identify genomic risk loci. Logistic regression was performed with age, sex, weight, height, BMI z-scores and drug dose and genetic models assessed.

Results: Overall, 210 SNPs were significantly associated with HPAS at $p = 0.05$. Six of these were prioritised via the FUMA analysis (rs4324305, rs1126417, rs28665675, rs8024276, rs1267542 and rs6503870). One was potentially protective i.e., rs8024276 (OR=0.09, 95% CI=0.02-0.45, $P=0.00047$, gene = SIN3A) while 5 were potentially causal. All SNPs were independent of covariates except rs28665675. All SNPs were statistically significant under the additive, Cochran-Armitage trend test and SNP rs8024276 significant for the dominant model. While rs1267542 & rs6503870 were recessive, the others were inconclusive under dominant and recessive models.

Conclusion: SNPs rs8024276, rs1267542 and rs6503870 if confirmed in a large sample and validated in an independent population, could be genetic markers for HPAS

Next Steps: Publication

Wisdom Akurugu

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Wisdom Alemya Akurugu is originally from Ghana but currently a Ph.D. student in Professor Nicola Mulder's Lab, Computational Biology (CBIO) at the University of Cape Town. He obtained his BSc Biochemistry (Single Major) degree from the University of Ghana in May 2007. Wisdom has over fifteen (15) years of experience in biomedical research from the Navrongo Health Research Centre and the Noguchi Memorial Institute for Medical Research (NMIMR), Ghana. He has been involved with activities of H3ABioNet since July 2013 as a resident bioinformatician for the NMIMR node. Through his work experience, Wisdom developed an interest in understanding genetic factors that place a person at risk of developing disease using modern Molecular Biology and Bioinformatics techniques. He is particularly interested in Bioinformatics discovery of genomic variations (i.e. single nucleotide polymorphisms (SNPs), indels, CNVs etc) that have biological significance. He pursued his MSc Bioinformatics degree from the University of the Western Cape where he assessed nucleotide variations on the structure and function of the human arylamine N-acetyltransferase 1. Since joining CBIO, in June 2018, Wisdom has been mining human genetic exome data for variations that may protect or predispose asthmatic children on corticosteroid treatment to hypothalamic-pituitary-adrenal suppression (HPAS). The study has both family-based trio and population-based case-control components. He is using various Bioinformatics, Computational and Statistical algorithms to identify, annotate and assess the impact of the potential variants on HPAS. Wisdom is enthusiastic about contributing to the effective and efficient treatment of asthmatics on corticosteroids based on his current project. In the near future, he looks to developing Bioinformatics, Computational and Statistical competencies in processing large-scale robust genomic data, interpreting the functional effect and the impact of genomic variation, integrating systems data to relate complex genetic interactions with phenotypes and translating these discoveries into medical practice. He is opened to collaborations in the areas of genomics, proteomics, transcriptomics, structural biology, software development and teaching

Trainer Biographies

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Dr Anita Ghansah is a Senior Research Fellow at the Parasitology Department of the Noguchi Memorial Institute for Medical Research, University of Ghana. Her research focuses on how parasite genetic diversity influences the epidemiology and pathogenicity of malaria and its implication for therapeutic and vaccination strategies as well as malaria vector control strategies. She has a keen interest in using population genetics strategies to identify genetic loci that contribute to drug resistance in *P. falciparum* and the development of genomic tools to characterize *P. falciparum* diversity and drug resistance in the population.

She has also been involved in Bioinformatics, genetics training in Ghana and developing innovative tools for monitoring malaria drug resistance, malaria vaccine efficacy studies that target parasite population dynamics, human genetics, and environmental impact. She is also interested in engaging/educating the research community in genomics, ultimately improving public health using genomics research.

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Mr. Collins Misita Morang'a is a PhD research fellow at WACCBIP, funded by the Wellcome Trust DELTAS Programme at the University of Ghana. He finished his studies in Biochemistry from Egerton University and obtained his Master of Science Degree in Molecular Biology from Maseno University in Kenya. In his Ph.D., he is interested in using artificial intelligence approaches to classify the malaria infections, as well as deconvolution of single cell Transcriptomics data from subclinical and clinical individuals. The secondary focus of his Ph.D. research is to identify regulatory mechanisms that dampen the activation of the Type I interferon pathway by sensing Plasmodium DNA during asymptomatic infections. The regulation checkpoints can

provide opportunities for therapeutic interventions against clinical malaria.

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Dr. Lucas N. Amenga-Etego is a Research Fellow at the West African Centre for Cell Biology of Infectious Pathogens (WACCBIP), Department of Biochemistry Cell and Molecular Biology, University of Ghana. He was a Research Fellow and Head of the Biomedical Science Department at the Navrongo Health Research Centre, Ghana from June 2013 until January 2018. His background is in microbiology and genetic/genomic epidemiology of malaria and his research interest is in infectious diseases with specific focus on the genetic contribution to malaria disease susceptibility (or resistance) and antimalarial drug resistance. For the past 10 years, he has collaborated with colleagues from the Big Data Centre, University of

Oxford and the Wellcome Trust Sanger Institute in the UK to conduct studies into the genetic diversity and population structure of *Plasmodium falciparum* populations and malaria vector mosquitoes in northern Ghana. He also conducts antimalarial drug resistance surveillance studies in northern Ghana. He is a founding member

of the Plasmodium Diversity Network Africa (PDNA) and a co-PI of the H3Africa Pan-African Malaria Genomic Epidemiology Network (PAMGen).

Mr. Vincent Appiah

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Mr. Vincent Appiah is the High Performance Computing Manager at WACCBIP, University of Ghana. He finished his studies in Biochemistry from University of Cape Coast and obtained his Master of Science Degree in Information Technology from Kwame Nkrumah University of Science and Technology, Ghana. His roles at WACCBIP include bioinformatics training and building of data analysis pipelines.

Facilitators

Dr. Rolanda Julius

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Rolanda Julius holds a PhD in Zoology with the University of Pretoria. She is a member of the South African Council for Natural Science Professions (SACNASP) and her research interests include zoonotic disease ecology, parasitology and molecular phylogenetics. She has experience in teaching molecular techniques, facilitating practical course components and has been a tutor for several undergraduate modules in the natural sciences faculty. She has also co-supervised postgraduate honours and MSc degree studies. She is dedicated to promoting public health awareness and life science education.

Dr. Laura Povlich

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Laura Povlich is a Program Director in the Division of International Training and Research at the Fogarty International Center, part of the National Institutes of Health, where she was previously an American Association for the Advancement of Science (AAAS) Science & Technology Policy Fellow. Dr. Povlich administers a portfolio of grants that covers a range of research, research training, and research education projects related to global health technology. Additionally, she is the program director for the H3Africa Global Health Bioinformatics Research Training awards. Prior to working at Fogarty, Dr. Povlich was the 2011-2012 Materials Research Society/Optical Society Congressional Science and Engineering Fellow in the Office of Congressman Sander Levin.

Dr. Povlich earned a B.S.E. in Materials Science and Engineering and a Ph.D. in Macromolecular Science and Engineering, both from the University of Michigan. Her research focused on the synthesis of functionalized conjugated polymers for biological sensor applications and for neural probe and prosthetic device electrode coatings.

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H3Africa Education and Coordinated Training Working Group (ECTWG)
Agenda WG

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Ambulance

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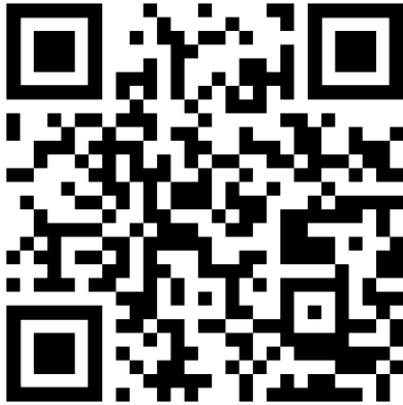
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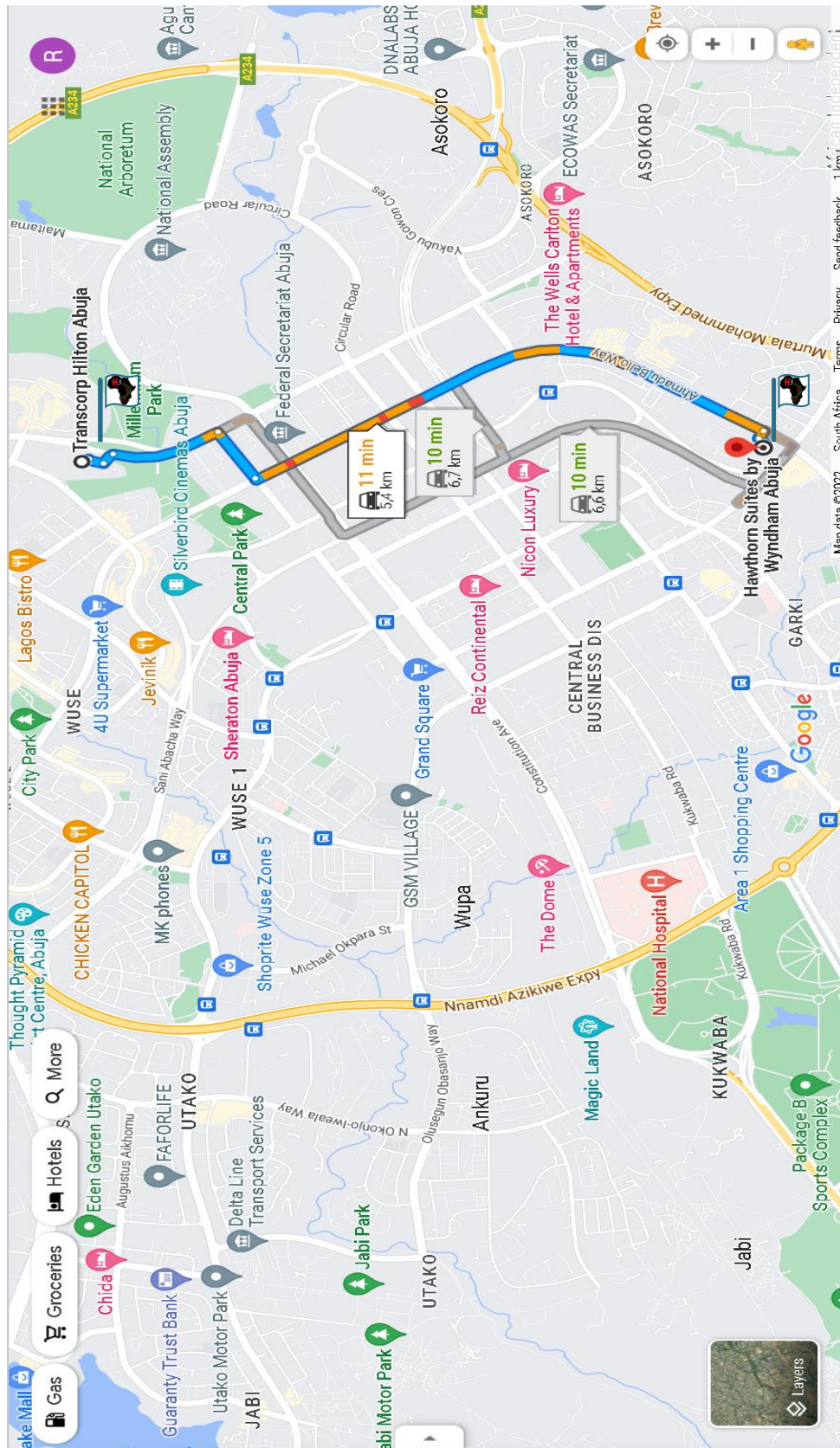


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Bioinformatics Coach

Site Map



Notes

This image shows a single sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

Notes



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