Report on the Third H3Africa Consortium Meeting
Johannesburg, South Africa
3-6 October 2013

Content

1. Overview of H3Africa

The Human Heredity and Health in Africa (H3Africa) initiative is a partnership among the National Institutes of Health (U.S.A.), Wellcome Trust (U.K.) and the African Society for Human Genetics. H3Africa aims to enhance the use of genomic approaches to the study of the genomic and environmental determinants of disease in Africa. The initiative supports African population-based genomic studies of common, non-communicable disorders such as heart and renal disease, as well as communicable diseases such as tuberculosis. The studies, are led by African scientists, and use genetic, clinical and epidemiologic methods to identify hereditary and environmental contributions to the risk of illness.

The goals of H3Africa are to (1) significantly increase the number of African scientists that are internationally competitive in genomics and population-based research; (2) establish collaborative networks of African investigators pursuing genomics-based, disease-oriented projects; and (3) create or expand genomics research infrastructure. The H3Africa Consortium currently funds twenty programmes which includes research projects, pilot biorepositories, a bioinformatics network and the study of the societal implications of genetic\genomics research in order to achieve its goals.

2. Meeting Aims and Objectives

Aims

- To provide the H3Africa Working Groups (WGs) an opportunity to meet face-to-face to finalize policies and guidelines, and present these to the Consortium for final approval.
- To provide an opportunity for the H3Africa investigators and the Steering Committee to meet and consider the progress and direction of the Consortium.
- To update the H3Africa Consortium, it's Independent Expert Committee and NIH and Wellcome Trust staff about the progress of the H3Africa grants.

Objectives

- To provide an opportunity for H3Africa investigators to give updates on the achievements of their projects.
- To share start-up experiences, successes, and lessons learned across projects.
- To provide an opportunity for the H3Africa working groups to meet and fine-tune policies, materials and recommendations to standardize and support activities across the Consortium.
- To identify opportunities for collaborations across the Consortium.
- To identify opportunities for collaborations outside of the Consortium that are generating genomic resources or reagents that are of potential value to H3Africa.
- To discuss how to build research capacity across the initiative and access new opportunities.
- To provide an opportunity for awardees to discuss specific issues with their funders.

3. Format of the meeting

The Third H3Africa Consortium provided a supportive platform for working groups and investigators to report back on their challenges and progress; community engagement was facilitated by open sessions and the consortium members and the broader scientific community had an opportunity to engage with internationally renowned experts in the fields of population genetics and genomics, with an emphasis on viable high throughput platforms appropriate for African studies.

The first two days were dedicated to the working group presentations, discussions and consolidation of keys tasks needed to move forward. On the third day the first phase grantees reported on their progress, and the new members of the consortium were introduced and presented their projects. On the last day of the meeting experts in the field of African population genetics and genomics gave presentations on their cutting edge research and current genomics tools; consortium members and broader scientific community also had a Q & A session with the panellist.

4. Internal Sessions

4.1. Biorepository Users Working Group

Introduction
Akin Aboyami started the session by giving a brief overview of the agenda: Moratorium for Biospecimen Release from H3Africa Biorepositories presented by Alan Christoffels; Biospecimen Access Committee presented by Akin Abayomi and Biospecimen Fees presented by Ute Jentsch and her team.

Moratorium on biospecimen release
The Biospecimen Release Policy and Procedures document formulated by Sub-group for Discussion on Sample-specific policies and noted that this document has to be viewed in the context of the broader policy document guidelines formulated by the Biorepository WG. The overarching points considered in this document are: a) release of samples (mainly DNA) from the research site to the biorepository and b) Control of access to samples by external users once at the biorepository.

It was suggested that each PI formulates a sample submission timeline in conjunction with the funders. It would be more economical to ship larger quantities simultaneously.

The sample and data submission and associated moratorium could extend over at least seven years, a potential stumbling block for the viability of biorepositories.

Once a sample submission schedule has been agreed upon, it would be very useful to monitor submission targets with a harmonized LIMS system.

Unresolved issues:
- At which point in the timeline can PIs share samples with collaborators and what are the implications for publication policy within the consortium.
- The PI could hold back on sample submission which freezes the countdown of the moratorium.
- The issue of commercial request/use of samples still has to be debated.

Biospecimen Access Committee (BAC)
This session was used to propose the formulation of the BAC, their qualifying prof essional characteristics and working framework.
- The BAC was suggested to have four to five members
- The majority of members should be from the African Continent (at least two thirds) and should include
    - Senior scientists, preferably in the field of genomics
    - Biobank specialist
    - Human research ethics specialist
    - Legal expert
    - Community representative

  The committee could serve a three-year term; however, a mechanism ensuring continuation has to be in place such as perhaps by the chair being replaced and the next chair being groomed during the run up period, and a new member joining.

The BAC must not have any conflicts of interest; individual members have to be totally unrelated to the consortium.

Function of the BAC:

Adjudication of sample requests

It was suggested that all requests should propose a mechanism for capacity building in Africa linked to it.

Hierarchy of criterion would be considered for each request:

a) H3Africa consortium members
b) African scientist in Africa
c) African scientist abroad,
d) International researchers with African collaborators
e) International scientist

Biospecimen Fees
Detailed fee schedules were given in the presentation. Please see the ppt.

4.2 Genome Analysis Working Group

Introduction
Zane Lombard chaired the internal working group session. There were six agenda items that were discussed: Assess genome analysis objectives and progress of H3A research projects; Improve communication, information sharing and skills transfer relevant to genome analysis between research groups; Assess desirability and feasibility of designing a customized standardized genotyping array; Guidelines in data QC, imputation and meta-analysis; Teaching and Training and Laboratory infrastructure in Africa.

Assess genome analysis objectives and progress of H3A research projects
The minimum information required for all projects has been defined previously at the study level. Surveys have been conducted and some data remains outstanding on existing projects. In addition, new projects have to be included. This data was collected by H3ABioNet and is still accessible.

Improve communication; information sharing and skills transfer relevant to genome analysis between research groups
An interactive forum should be utilized on the H3Africa website, which should encompass Q&A, progress reporting and a monthly newsletter. Interaction with the Phenotype harmonization working group should be actively pursued.

Assess desirability and feasibility of designing a customized standardized genotyping array
It was agreed that it was necessary to pursue the development of an Afro centric genotyping chip. The Genome Analysis working group would explore using existing and prospective data within the consortium and data available through AGVP, and possibly accessing other networks with similar objectives.

Guidelines in data QC, imputation and meta-analysis
Draft guidelines based on platforms which adequately capture African variation, GWAS QC and imputation and standardized QC procedures for genotyping and sequence data should be done

by May 2014 and finalized by October 2014. The Meta analysis and combined analysis of minimum data should be done by October 2014.

Teaching and Training
Training will be facilitated by the Genome Analysis working group in cooperation with H3ABioNet with an emphasis on consortium scientists being empowered to analyse their own data. The original idea was for researchers to work on their own data. However the data is not forthcoming presently and an alternative may be to use simulated data to expedite the training.

Laboratory infrastructure in Africa
It was felt that this task was particularly challenging and that a sub-group could be formulated to tackle the issue of setting up GWAS and NGS infrastructure. This would require elements of human resources as well as equipment, back-up power, and reagents. Objectives have to be clearly defined.

4.3. Ethics and regulatory Issues Working Group

Introduction
This session was chaired by Jantina De Vries. In the context of data sharing, informed consent and community engagement, there was a consensus from the working group that trust was the central thread. Trust between researchers and participants; researchers and their institutions and researchers and their funders. The success of the consortium and its objectives is dependent on fostering trust, and community and public engagement is key to this. It was clearly articulated that the onus of ensuring that research is carried out ethically rests firmly on the investigators and not with ethics committees or the ethics working groups.

The H3Africa consortium has the potential to change the way in which collaborative research in and with Africa is done; doing research should be taken seriously and done responsibly. Stakeholders should be engaged at all levels including ethics committees, ensuring that policies, materials and agreements take into consideration/are responsive to concerns at all these levels. Limited funding for ELSI within the current H3Africa funding scheme with respect to the intensive levels of engagement required is problematic.

Major challenges were identified:

- A real challenge that needs to be discussed much more broadly in the context of H3Africa studies is ‘what do people minimally need to understand the genomic project in order for their consent to be valid’.
- Does H3A need a policy on the feedback of incidental and individual findings and if so, what would that policy be?
- Perhaps the Ethics WG could contribute to understanding what would constitute ‘benefit’ in the H3A projects, and to monitor whether benefits are in actual fact generated. For instance, if researchers say that benefit will be capacity building, perhaps the WG could monitor this aspect by exploring how many individuals have been trained, where they find employment, etc. However, it was not clear whether it would be feasible for the WG to pursue this.
- A concern in the context of H3A is that there is very limited funding available for empirical ethics work, whilst there is an acute need for empirical data to inform policy development.

Recommended action items established by the working group:

- Develop clear regulatory guidelines for the implementation of best practices within consortium;
- Develop the definition as to what constitutes benefit, monitor capacity building and build trust in the research enterprise.
- Develop materials that would help participants understand genomics better. Examples of such materials would be PowerPoint presentations, online information, brochures and so forth. Coordinate with other working groups to prevent repetition.
- Sustained feedback from projects could be facilitated by the ethics working representative within each project and regular questionnaires about ethical challenges the project encounters.

Suggestions:
- Ideally, each project should have a dedicated ethics person empowered to make real contributions to their projects. Could this be achieved through an administrative supplement?

4.4. Education and Coordinated Training Working Group

Introduction
This working group has recently been formed; they have established their goals and elected co-chairs, Sheryl McCurdy and Misaki Wayengera.

Goals
- Establish a website for educational opportunities including workshops with beginners' and advanced courses, mentor programs, etc.
- Ensure that the training materials are available online
- Develop a list of trainees and cohorts
- Follow-up and assess efficacy of training experiences within one, five and 10 years of initiation.
- Set guidelines/MOP for creating training courses and establish advanced courses

While the research groups are already engaged in short-specialised, longer term, and cross disciplinary training programs, the working group sees its role as coordinators of training resources so as to avoid duplication. The WG will also actively engage organizations committed to investment in African development.

4.5 Study Coordinators Working Group

Introduction
This session was chaired by Widaad Zemanay. Since the group was recently formed, the internal session was mostly used to discuss the goals of the group, progress to date, and future plans. The two co-chairs are Widaad Zemanay and Mark Engel.

Goals

- Facilitate inter-project discussions to optimize grant function
- Review draft policy documents and provide practical feedback
- Aid and advise in the launch of new sites
    - Discuss experiences and lessons learnt
    - Provide a platform for problem solving, sharing of documentation and templates
    - Advise the implementation of H3Africa policies on site
- Provide support and facilitate full utilization of available resources

Progress
Review of several consortium documents

- Biorepository SOP master list
- Biospecimen release policy and procedures
- Guidelines for informed consent
- Guidelines for community and public engagement

Future Plans
Continue to review and provide inputs with respect to H3Africa policy documents as they are drafted. Maintain open communication with all working groups and PIs across respective grants.

4.6 Data Sharing, Access, Release working Group

Introduction
This session was chaired by Nicola Mulder. After a brief review of the agenda items, specific documents developed by the working group were reviewed and a brief overview of each document was presented. An overview of the flow of data from the primary investigators to H3ABioNet and then to the EGA with associated timeline was presented. Quality control, data submission timelines and data access were discussed and potential solutions were formulated.

Data Quality Control
The working group felt that the allotment of time for quality control which was originally two months should be reviewed in the context of each project. As the type of data is specific for each project, this issue should be referred to the funders for further discussion to determine when the clock starts ticking for each project.

Quality Control workshops should be held:

- To assist investigators with their QC issues to ensure they are compliant and adhere to their timeline.
- To train on QC of next generation sequencing and chip based genotyping data ,and
- To entail video streaming and an onsite trainer.

Data Submission to H3ABioNet and the EGA

It was acknowledged that not all data sets are the same and although some may have components that can stand alone, others will require all partial data sets to be combined to be meaningful. It was conceded that this could considerably alter the timeline for data submission and that once again the details should be worked out with the program officers from the funding agencies.

A concern was raised about the relatively short time that new trainees in genomics will have to move from data collection to publication. Another view was presented indicating that not releasing data in a timely fashion is equally unacceptable/unethical.

Data Access

- **Internal Access**
  It was suggested that consortium members should be able to access the data using an internal mechanism set up by H3ABioNet separate from the external mechanism (through the EGA). This mechanism would have to be transparent to safeguard against 'scientific nepotism'.
- **External Access**
  - **Data Access Committee Structure**
    - Consists of 10 members.
    - H3Africa Consortium PIs ineligible.
    - Majority of the members should be from Africa.
  - **Request arbitration**
    - Requests should be linked to an element of capacity building within the Consortium and recorded in a data access document (agreement).
    - Evidence of tangible benefits for the African populations should be a criterion
    - Matters arising could be referred back to the PIs.
    - Unanimous decisions would be made. If ethical issues did arise this could be referred back to the relevant ethics committee (original IRB, special ethics committee)
- **Data Sharing, Access and Release and Biorepository Users working groups**
  - Should these working groups be merged or should the Biorespository Access and the Data Access committees be merged?

4.7 Phenotype Harmonization working group

Introduction
This session was chaired Alia Benkahla. Nine of fifteen research projects participated in this session. The working group based their model on a previous model developed by the GENEVA consortium (Bennet et al., 2011).

Recommendations for phenotype harmonization

- Phenotype standards (the way in which the questions are asked).
- A set of phenotypes to be collected in all studies.

PhenX standards would be implemented and adapted to suit the H3Africa Consortium studies where appropriate. Thus far, they have received CRFs from four research projects and compared phenotypes being collected. To improve this feedback, the document was re-circulated during the meeting and would also be sent out via email to the working group by October 23, 2013 with a response deadline of October 30, 2013. The response should indicate a selection of questions to be included in the questionnaire, comments and agreement/disagreement with regards to existing questions.

The responses would then be integrated into the CRFs before submission to expedite the feasibility of cross-study analysis.

4.8 Publications and Marker Paper Working Group

Introduction
The chairs of this session were Michele Ramsay and Enoch Matovu.

Guidelines for authorship
Several examples of publications from other consortia were presented and formed the basis for discussions. The types of papers expected from the H3Africa consortium were categorised. There were discussions on authorship and as to which would require submission of manuscript concept documents or submission of the manuscripts or abstracts for review by the steering committee. The table below summarizes the consensus reached in the workshop.

Overall the guidelines for authorship discussed were embraced. However the guidelines for single grantee group publications was amended as follows: e.g. Authors, followed by " for the X research group, as members of the H3Africa consortium".

Marker Paper and publications
It was unanimously agreed that information about newly funded projects should be included so that the paper would be representative of the current situation. An updated map including primary award institutions and collaborating sites would be needed. Members were urged to share information about challenges and experiences so far, such as consenting participants for genomics research.

Science was earmarked as the journal of choice and considerable discussion was already underway. There was agreement that the Marker Paper should be open access and that this should be negotiated with the editors of Science.

Would theses/dissertations arising from graduate students work be obligated to acknowledge the consortium? It was agreed that these need not be submitted for review before candidates can get their academic awards, but manuscripts arising should be categorized and subjected to the same principles as above, giving due acknowledgement to the consortium.

Publication category	Authorship	MCD	Manuscript
H3A Consortium	Marker paper: The H3Africa Consortium – all members listed at the end of the paper Consortium-wide papers: Members of the research group as authors followed by "and The H3Africa Consortium", followed by all members of H3Africa listed at the end of the paper	Yes	Submit to CC review by SC prior to submission
H3Africa Consortium Working Group	Authors followed by "and The H3Africa Consortium" followed by the remainder of the WG members at the end of the paper	Yes	Submit to CC Review by the SC prior to publication
Multiple grantee group	Authors followed by "as members of The H3Africa Consortium".	Yes	Submit to CC following publication
Single grantee group	Authors followed by "as members of The H3Africa Consortium".	No	Submit to CC following publication
Consortium Collaborations (including external partners)	H3Africa Consortium members collaborating with external partners –MCD completed publication guidelines as above	Yes /No	Submit to CC following publication

4.9 Communication and Outreach Working Group

Introduction

This session was chaired by Akin Abayomi. The group will focus on Communication Outreach Advocacy (not Community Engagement) and is aimed at facilitating the vision and goals of H3Africa through multiple tiers of ambassadors in the community to ensure community and national ownership of H3Africa. It is more of a top down approach than a bottom up approach. Stakeholders include politicians and other research groups. Media, television, articles, public talks, documentaries, social media, and conferences would be used as mediums of communication.

Primary function is Advocacy targeted at African Governments to:

- Increase financial investment on scientific and genetic/genomic research
- Increase public awareness
- Build research capacity
- Improve research infrastructure on the continent

Three levels of ambassadors were defined:

- high level ambassadors (former ministers of Health or Science, high-level officials, deans of science academies, philanthropists, ex-presidents, nobel laureates
- Popular icons (musicians, poets, film stars)
- Youth ambassadors

The plan to identify and recruit high profile ambassadors, engage them, determine terms of reference, recruit, and prioritise communication and publicity mechanisms.

Marketing the consortium needs to be prioritized, with an emphasis on engaging for governments and the Africa Union, so that they understand that the project is a international asset.

5. Joint Sessions

5.1 Data, Phenotype, and Biorepository users joint session

Introduction
The aim of this session was to help the three working groups identify and discuss overlapping issues. The discussion was focussed on what amount of data (participant information) should accompany a sample for biorepository purposes, for phenotype harmonization purposes and data access purposes. Would there be separate lists? The phenotype harmonization list (CRF) covers individual level variables, study level variables, and sample specific variables. It was agreed that this list would be used.

A recurring issue which may resolve many overlapping problems requires the merging of the BAC and DAC.

5.2 Ethics and Study Coordinators working group joint session

Introduction
Having a clear oversight of the rules relating to the movement of materials per country would be very relevant to the Consortium. Creating resource information 'repository' relating to existing, country- or institution-specific MTAs, rules and regulations regarding sample import and export. There are existing web based tools www.clinicalresearchregs.org (BETA).

An important question was how to deal with restrictions on the movement of samples. For instance, the RHD group has currently got permission for samples to be sent from Uganda to South Africa, for sample processing in Cape Town only. But samples are often sent abroad for some genotyping (service only) and the question is whether this would be a deviation from the original permission. The group felt that it would be, and that permission for sending samples elsewhere needed to be obtained. Due to the real potential of sample and data analysis being done abroad, should this awareness be included in the consent form?

MTA should include assurance that analysis cannot be done locally and there analyses have to be done elsewhere and the export of the samples will benefit health in Africa and contribute capacity development in Africa.

Appendix 1 – Summary of Action Items

1. **Biorepository Users Working Group**
   - The moratorium on biospecimen release will be affected by project specific needs: PIs will liaise with funders for project specific timelines.
   - Initiate the formulation of the Biospecimen Access Committee (BAC) (which will potentially be merged with the Data Access Committee (DAC).)

2. **Genome Analysis Working Group:**
   - Recirculate minimum study level survey information to project representatives (including new projects) within the working group.
   - Create an interactive discussion forum on the H3Africa website for genome analysis working group.
   - Form a task team for the development of an Afro-centric chip.
   - Develop a draft document by May 2014 and finalize the document by Oct 2014.
   - Collaborate with H3ABioNet on training programs.
   - Form an NGS/GWAS infrastructure sub group.

3. **Ethics & Regulatory Issues WG**
   - Develop clear regulatory guidelines for the implementation of best practice within consortium;
   - Develop the definition to constitute benefit, monitor capacity building and build trust in the research enterprise.
   - Develop materials that would help participants understand genomics better. Examples of such materials would be PowerPoint presentations, online information, brochures, etc... Coordinate with other working groups to prevent repetition.
   - Ethics Working Group's representatives from each project will facilitate sustained feedback from their projects and regulate questionnaires about ethical challenges their projects encounter.
   - Ideally, each project should have a dedicated ethics person empowered to make real contributions to their projects. The Ethics Working Group will consider the possibility of asking for an administrative supplement?
   - Establish what the legislation is around importing and exporting biospecimens nationally (for all African countries concerned).

4. **Education & Coordinated Training WG**
   - Coordinate training resources to avoid duplication
   - Engage organizations committed to investment in African development

5. **Study Coordinators Discussion Group**
   - Continue reviewing H3A policy documents
   - Maintain open communications with all working groups and PIs across H3Africa grants.
   - Include in the Material transfer agreement (MTA) document assurance that analysis cannot be done locally and the export of samples will benefit health in Africa and contribute to capacity development in Africa.

6. **Data Sharing, Access and Release WG**
   - Plan QC/NGS/Genotyping workshops for projects.

- Continue the discussion of and ratify the mechanisms for internal and external data access.
- Finalize the decision on merging the DAC and the BAC.
- Finish discussions on Data Sharing policies.

7. Phenotype Harmonization WG

- Finalization of the Case Report Form.

8. Publications & Marker Paper WG

- Engage with Science on the point of open access of the marker paper.
- Finalize the Publications policy and circulate the document to the H3Africa Steering Committee.

9. Outreach & Communications WG

- Engage with high level officials to become H3Africa Ambassadors and promote H3Africa as a valued national asset.

Appendix 2 – Meeting Agenda

Thursday, October 03, 2013 Third H3Africa Consortium Meeting Day 1		
07:00-08:30	**Registration & Welcome Coffee/Tea**	**VENUES**
08:30-10:00	**PARALLEL SESSIONS** **Biorepository Users Working Group** Chairs Akin Abayomi and Alan Christoffels Discussion Facilitators Sue Penno **Genome Analysis Working Group** Chairs Debo Adeyemo and Zane Lombard Discussion Facilitators Jeff Struewing **Ethics &Regulatory Issues Working Group** Chairs Jantina De Vries Discussion Facilitators Ebony Bookman and Katherine Littler **Education and Coordinated Training Working Group (08h30 – 09h30)** Discussion Facilitators Maria Giovanni and Lara Bethke **Study and Coordinators Discussion Group (09h30 – 10h00)** Discussion Facilitators Lara Bethke and Robin Mason	**Main Station One** **Main Station Two** **Main Station Three** **Union Station** **Union Station**
10:00-10:30	**Tea/Coffee break**	**Outside Meeting area**
10:30-12:30	**PARALLEL SESSIONS** **Biorepository Principle Investigators** Chairs Sue Penno Discussion Facilitators Alash'le Abimiku **Genome Analysis Working Group** Chairs Debo Adeyemo and Zane Lombard	**Main Station One** **Main Station Two**

	Discussion Facilitators Jeff Struewing **Ethics &Regulatory Issues Working Group** Chairs Jantina De Vries Discussion Facilitators Ebony Bookman and Katherine Littler **H3ABionet Infrastructure and User Support Working Group**	**Main Station Three** **Union Station**
12:30-14:00	**Lunch**	
14:00-15:00	**PARALLEL SESSIONS** **Data Sharing, Access, Release Working Group** Chairs Nicola Mulder and Bamidele Tayo Discussion Facilitators Maria Giovanni and Audrey Duncanson **Phenotype Harmonization Working Group** Chairs Alia Benkahla Discussion Facilitators Jeff Struewing **Publications & Marker Paper Working Group** Chairs Michele Ramsay, Charles Rotimi, Enock Matovu Discussion Facilitators Mark Guyer and Audrey Duncanson **Communications & Outreach Working Group** Chairs Akin Abayomi Discussion facilitators Jane Peterson & Lara Bethke	**Main Station One** **Main Station Two** **Main Station Three** **Union Station**
15:00-15:30	**Tea/Coffee break**	
15:30-16:30	**PARALLEL SESSIONS** **Data Sharing, Access, Release Working Group** Chairs Nicola Mulder and Bamidele Tayo Discussion Facilitators Maria Giovanni and Audrey Duncanson	**Main Station One**

	Phenotype Harmonization Working Group Chairs Alia Benkahla Discussion Facilitators Jeff Struewing **Publications & Maeker Paper Working Group** Chairs Michele Ramsay, Charles Rotimi, Enock Matovu Discussion Facilitators Mark Guyer and Audrey Duncanson **Communications & Outreach Working Group** Chairs Akin Abayomi Discussion Facilitators Jane Peterson & Lara Bethke	**Main Station Two** **Main Station Three** **Union Station**
16:30-17:30	**Steering Committee Meeting**	**Main Station One**
17:30-18:00	**Discussions** Principle Investigators and Independent Expert Committee Principle Investigators and Project teams Principle Investigators and Funders	**Main Station Two**
18:00-19:00	**Social hour**	**Pool deck**
19:00	**Dinner**	

Friday, October 04, 2013 ***Third H3Africa Consortium Meeting Day 2*** *(All meetings take place in the Gautrain room unless stated otherwise)*	
07:30-08:30	**Welcome Coffee/Tea**
08:30-08:35	**Opening Remarks –** *Audrey Duncanson*
08:35-09:30	**Biorepository Working Group** Chairs Charmaine Royal Discussion Facilitators Akin Abayomi, Alash'le Abimiku
09:30-10:30	**Data Sharing, Access and Release Working Group** Chairs Val Sheffield Discussion Facilitators Nicola Mulder, Bamidele Tayo
10:30-11:00	**Tea/Coffee break**
11:30-13:00	**Ethics and Regulatory issues Working Group** Chairs Ruth Chadwick

	Discussion Facilitators Jantina De Vries
13:00-14:00	**Lunch**
14:00-15:00	**Genome Analysis Working Group** Chairs Carlos Bustamante Discussion Facilitators Zane Lombard
15:00-16:00	**Phenotype Harmonization Working Group** Chairs George Mensah Discussion Facilitators Alia Benkahla
16:00-16:30	**Tea/Coffee break**
16:30-17:30	**Publications & Marker Paper Working Groups** Chairs Phillip Awadalla Discussion Facilitators Michele Ramsay, Enock Matovu
17:30-18:30	**17:30 - 17:50: Education & Coordinated Training Working Group** **17:50 - 18:10: Communications & Outreach Working Group** **18:10 - 18:30: Study Coordinators Discussion Group** *Facilitator: Lara Bethke*
18:30-19:30	**Discussions** Principle Investigators and Independent Expert Committee Principle Investigators and Project teams Principle Investigators and Funders
18:30-19:30	**Social Hour –** *Pool Area, Radisson Hotel*
19:30	**Dinner**

Saturday, October 05, 2013 **Third H3Africa Consortium Meeting Day 3** (All meetings take place in the Gautrain room unless stated otherwise).	
07:30-08:00	**Welcome Coffee/Tea**
08:00-08:30	**Welcome & H3Africa Overview** *Kay Davies*
SESSION I	
Bioinformatics - *Chair: Kay Davies*	
08:30-08:50	**H3ABioNet: A Sustainable African Bioinformatics Network for H3Africa** Principle Investigator : Nicola Mulder
SESSION II	
Non-infectious Diseases - *Chair: Val Sheffield*	
08:50-09:10	**H3Africa Kidney Disease Research Network**

	Principle Investigator : Dwomoa Adu
09:10-09:30	**Genomic and Environmental Risk Factors for Cardiometabolic Disease in Africans** Principle Investigator : Michele Ramsay / Osman Sankoh
09:30-09:50	**Burden, spectrum and etiology of type 2 diabetes in sub-Saharan Africa** Principle Investigator : Clement Adembamowo
09:50-10:10	**The RHDGen Network: Genetics of rheumatic heart disease and molecular epidemiology of Streptococcus pyogenes pharyngitis** Principle Investigator : Bongani Mayosi
10:10-10:30	**Stroke Investigative Research & Educational Network (SIREN)** Principle Investigator : Mayowa Ojo Owolabi
10:30-11:00	**Tea/Coffee break**
11:00-11:20	**Clinical and genetic studies of hereditary neurological disorders in Mali** Principle Investigator : Guida Landoure
11:20-11:40	**Genomics of Schizophrenia in the South African Xhosa** Principle Investigator : Raj Ramesar
SESSION III	
Microbiome - *Chair: Carlos Bustamante*	
11:40-12:00	**African Collaborative Center for Microbiome and Genomics Research (ACCME)** Principle Investigator : Clement Adebamowo
12:00-12:20	**The nasopharyngeal microbiome and respiratory disease in African children** Principle Investigator : Mark Nicol Patrick
12:20-13:30	**Lunch**
SESSION IV	
Biorepository – *Chair: Ruth Chadwick*	
13:30-13:50	**IHVN H3 African Biorespository (I-HAB) Initiative** Principle Investigator : Alash'le Abimiku
13:50-14:10	**Development of H3 Africa Biorepositories to facilitate studies on Biodiversity, Disease & Pharmacogenomics of African Populations** Principle Investigator : Akin Abayomi
14:10-14:30	**Integrated Biorepository of H3Africa Uganda - IBRH3AU** Principle Investigator : Moses Joloba
14:30-14:50	**Establishment of an H3Africa Biorepository at Contract Laboratory Services** Principle Investigator : Ute Jentsch
SESSION V	
Ethics - *Chair: Charmaine Royal*	
14:50-15:10	**Exploring Perspectives on Genomics and Sickle Cell Public Health Intervention** Principle Investigator : Ambroise Wonkam
15:10-15:40	**Tea/Coffee break**
SESSION VI	
Infectious Diseases - *Chair: Philip Awadalla*	
15:40-16:00	**Host and Microbial Genetic Determinants of Febrile Illness in West Africa** Principle Investigator : Christian Happi
16:00-16:20	**TrypanoGEN: An integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis** Principle Investigator : Enock Matovu
16:20-16:40	**Reprogramming of the Trypanosoma brucei epigenome during human infection: opportunities for new therapies** Principle Investigator : Hugh-George Patterton

16:40-17:00	**Collaborative African Genomics Network (CAFGEN)** Principle Investigator : Gabriel Anabwani
17:00-17:20	**RAFAgene: Contribution of genetic variation to pharmacokinetic variability and toxicity in patients undergoing multi-drug tuberculosis treatment in Sub-Saharan Africa** Principle Investigator : Dissou Affolabi
17:20-17:40	**Systems Biology for Molecular Analysis of Tuberculosis in Ethiopia** Principle Investigator : Gobena Ameni
18:00-19:00	**Steering Committee Meeting**

Sunday, October 06, 2013 **H3Africa Genome Analysis Session** **The Maslow Hotel**		
TIME	**SPEAKER**	**TOPIC**
8:30 – 8:35	Zané Lombard	Introduction to the session
8:35 – 8:45	Audrey Duncanson	Overview of H3Africa
8:45 – 9:25	Himla Soodyall & Mattias Jakobsson	Population genomic variation across Africa
9:25 – 10:00	Manj Sandhu (AGVP)	Current arrays for capturing variation in African populations & whole genome sequencing based approaches
10:00 – 10:25	Dominic Kwiatkowski (MalariaGEN)	Imputation, meta-analysis & population stratification
10:25– 10.50	Carlos Bustamante	Admixture and African populations
10:50 – 11:40	Major platform & technology companies Chair: Michele Ramsay	Technologies and platforms for genomics in Africa:
11:40 – 12:10	Panelist Manj Sandhu Dominic Kwiatkowski Mattias Jakobsson Himla Soodyall Chairs: Carlos Bustamante and Michele Ramsay	Panel Discussion
12:10 – 12:15		Closing remarks

Appendix 3 – Project abstracts and Investigators

Wellcome Trust H3Africa Research Projects

RHDGen: The genetics of rheumatic heart disease network
PI: Bongani Mayosi
Organization: University of Cape Town, South Africa

There are four proposed activities for the RHDGen Network:

1) To build a clinical and laboratory network for the phenotyping of patients with rheumatic heart disease (RHD) and controls;
2) To identify genetic variants affecting susceptibility and resistance to RHD;
3) To train a group of scientists and clinicians in genomic studies of multifactorial disease; and
4) To address ethical, legal, and social issues (ELSI) that are relevant to Africa.

The key goals of the network are:

1) To recruit 2,500 patients with echocardiographically-confirmed RHD and 3500 controls.
2) To conduct a case-control genome-wide association study by genotyping 5 million single nucleotide polymorphisms in 1,500 RHD cases and 1,500 unrelated controls using micro-array technology, followed by replication in a further 1,000 independent cases and 2,000 family-based controls, and combined analysis of genotype data from all 2,500 cases and 2,500 controls or pseudo-controls to detect rarer alleles or alleles of smaller effect.
3) To train 16 scientists and clinicians in genomics and ELSI at masters, doctoral and postdoctoral levels.

Bongani Mayosi
University of Cape Town
Bongani Mayosi is the Professor of Medicine and Head of the Department of Medicine at Groote Schuur Hospital and University of Cape Town. He qualified in medicine from the University of KwaZulu-Natal in Durban, and trained in internal medicine and cardiology in Cape Town. He was as the Nuffield Oxford Medical Fellow in cardiovascular medicine at the University of Oxford from 1998 to 2001. His research interests include genetics of cardiovascular traits, treatment of tuberculous pericarditis, prevention of rheumatic fever, and epidemiology of heart disease in Africa. He is the Chairman of the South African National Health Research Committee, President of the College of Physicians of South Africa, President of the Pan African Society of Cardiology (PASCAR), Chairman of the Rheumatic Fever Council of the World Heart Federation, and Associate Editor for Africa for *Circulation*. In November 2009, President Jacob Zuma bestowed upon him South Africa's highest honour, the *Order of Mapungubwe in Silver*, for excellent contributions to medical science.

Jantina De Vries
Department of Medicine, University of Cape Town
Dr. Jantina de Vries is the Chair of the H3Africa Working Group on Ethics and Regulatory Issues. She is a senior researcher in bioethics at the Department of Medicine, University of Cape Town and involved in Prof. Bongani Mayosi's RHD GEN (H3Africa) project. Dr. de Vries obtained a DPhil at the Ethox Centre, University of Oxford. Her DPhil research focused on examining ethical issues in the use of ethnicity data in genomics research in Africa. She was previously the ethics coordinator of MalariaGEN

Maia Lesosky
University of Cape Town
Dr. Maia Lesosky is a Senior Lecturer and biostatistician in the Department of Medicine at the University of Cape Town and an Extraordinary Lecturer in the Faculty of Veterinary Sciences at the University of Pretoria. Maia was educated at the University of Guelph, Canada, with degrees in mathematics (BSc Honours 2004) and statistics (MSc 2005, PhD 2009) and has held previous positions at Aalto University, Finland, Roslin Institute, UK and the Centre for Immunity, Infection and Evolution at the University of Edinburgh. Dr. Lesosky has conducted research in a wide range of fields, including evolutionary biology, ecosystem change and infectious disease transmission but now primarily focus' on biostatistical aspects of human medical research including genetic association studies for complex disease traits.

Babu Muhamed
University of Cape Town
Babu Muhamed is currently registered for a PhD in cardiovascular genetics at the University of Cape Town. He completed his undergraduate degree in microbiology at the University of Zululand followed by honours and master's degrees at the University of Pretoria and University of Cape Town, respectively. His area of focus is on genetics of Rheumatic Heart Disease.

Nakita Laing (Nee Verkijk)
University of Cape Town
After completing my undergraduate degree at Stellenbosch University, I registered at the Division of Human Genetics at UCT for my Honours degree in 2009. My research project was on familial colorectal cancer and looking at potential modifiers for the disease. I was then accepted into the MSc genetic counselling programme at UCT in 2010 and completed my internship in March 2013. My research project for my MSc involved doing a qualitative study into the reasons why women from a family affected by ATR-X syndrome did not present for carrier testing. During my internship I was exposed to various area's in genetics, working with the retinal degeneration project through funding from RetinaSA, and with the Department of medicine at Groote Schuur hospital assisting with cardiology research, together with the usual genetic clinics that our division supports. I currently work Part-time in the Division of Human Genetics mainly counselling patients in various clinics (both Red Cross Children's hospital and Groote Schuur Hospital) and also aiding in teaching of medical students, nurses and other professionals. I also still work part-time for the department of Medicine at Groote Schuur Hospital assisting with cardiology research by extending and counselling known families affected by inherited cardiomyopathies as well as determining the eligibility of patients with Rheumatic heart disease for new genomic research that is going to be performed through H3Africa.

Roxi-Lynn Vergotine
University of Cape Town, Dept.of Medicine
Database management/administation for IMPI Trial. Been with Mayosi group for four years.

Gasnat Shaboodien
University of Cape Town
For our H3Africa project, I have been appointed as the person in charge of our Biorepository as well as shipping of all biological samples from and to the various sites.

Mark Engel
University of Cape Town
Mark Engel is based in the Dept of Medicine at UCT as a senior Researcher. He holds an MPH in epidemiology and conducted his doctoral studies in the Epidemiology of rheumatic heart disease.

TrypanoGEN: an integrated approach to the identification of genetic determinants of susceptibility to trypanosomiasis
PI: Enock Matovu
Organization: Makerere University, Uganda
The over-arching aim of this network is to improve the health of people living in some of the poorest countries in the world that carry a disproportionate burden of infectious diseases. Despite their importance, the study of many tropical diseases has lagged behind that of diseases of developed countries. This network will redress the balance by performing high quality research into the neglected tropical disease of human African trypanosomiasis.

High level objectives:
1. To create an extensive biobank of both retrospective and prospective samples. In order to deliver this scientific objective, it will be necessary to achieve underpinning capacity building objectives; to establish a pan-African, interdisciplinary research team incorporating parasitologists, geneticists, genome analysts, clinicians, ethicists and bioinformaticians; to train personnel in diagnosis/sampling and depositories for both retrospective and prospective and to provide underpinning infrastructure.
2. To generate a database of human genetic variation from different African countries that will be available to the wider scientific community for research on other diseases and analysis of human genetic diversity and evolution. In order to deliver this scientific objective, it will be necessary to achieve the capacity building objective of enhancing local research capacity via development of training in advanced genomics.

Enoch Matovu
Makerere Univeristy
Ass. Prof. Enock Matovu obtained his PhD in Molecular Parasitology from the University of Bern, Switzerland in 2001, while he worked as a Research Officer at the Livestock Health Research Institute, Tororo, Uganda. Since then has continued his work on drug resistance and later diagnostics for African Trypanosomiasis. In 2004, he relocated to the Makerere University School of Veterinary Medicine, where he was first employed as a Lecturer. In 2008, Enock received the prestigious Royal Society Pfizer Award in recognition of his work on molecular mechanisms of drug resistance in African trypanosomes. The previous year (2007) he had obtained the Joint Third World Academy of Science Award for Young Scientists, for his contribution to the field of Molecular Parasitology. Enock Matovu has vast experience in HAT ranging from surveillance, diagnostics, drug resistance and clinical trials.

John Chisi
College of Medicine, University of Malawi
Professor & Head of Basic Medical Sciences, Department. College of Medicine, University of Malawi
University of Malawi, Chancellor College BSc 1984-1986 Bachelor of Science.University of St Andrews BSc Hons 1986-1990 Human Anatomy.University College, Lon. UNIMA, College of Medicine MBBS 1990-1993 Clinical Medicine.University of St. Andrews, Scotland PhD 1995-1998 Pathology (Haematology)

Christiane Hertz-Fowler
University of Liverpool
Christiane graduated from the University of St Andrews in 1995 with a degree in Biochemistry and Microbiology. During her undergraduate degree she developed an interest in parasitology, which led to doctoral work on the life cycle differentiation and cytoskeleton of the African Trypanosome, completing a PhD and post-doctoral work in 1999 in Professor Keith Gull's group. Christiane subsequently moved to the Wellcome Trust Sanger Institute in 2001, where she was involved in, and more recently lead, the management, annotation and analysis of trypanosomatid genome projects as well as the development of accompanying database resources. In 2009, Christiane moved to the University of Liverpool, where she now manages the Centre for Genomic Research (http://www.liv.ac.uk/cgr/).

John C.K. Enyaru
Makerere University
Professor in Biochemistry at the department of Biochemistry and Sports Science, College of Natural Sciences, Makerere University, Uganda. Iam currently carrying out research on the development and application of diagnostic tools for the detection of trypanosomes in tsetse flies, the vector for African Trypanosomiasis.

Issa Sidibe
Centre International de Recherche Développement sur l'Elevage en zone Subhumide
Issa started to work on trypanosomiasis in 1984, at CRTA (Centre de Recherches sur les Trypanosomes Animales) in Burkina Faso. He initially worked on immunology, studying trypanotolerance versus susceptibility to trypanosomiasis in different cattle breeds. Building on this work, he undertook epidemiological studies in different agropastoral zones of Burkina Faso and went on to work in the Kénédougou area studying the resistance to trypanocides of different trypanosome strains. Issa defended his PhD on the genetic variability of T. congolense: taxonomical and epidemiological implications in 1996 at the University of Montpellier II. Building on this, he went on to work on improving the molecular diagnosis of trypanosomes using PCR (polymerase chain reaction) techniques. Issa was chairman of the ISCTRC from 2001 to 2003 and remains a member of its Executive Committee as well as a member of the PAAT advisory group. In CIRDES (formerly CRTA), Issa was head of the Biological and Integrated Disease Control Unit. In April 2005, he was appointed scientific director of CIRDES to August,2011. In 2006, cumulatively with his function of Scientific Director of CIRDES, Issa SIDIBE was appointed as the National Coordinator of the Pan African Tsetse and Trypanosomiasis Eradication Campaign (PATTEC) of Burkina Faso. Issa SIDIBE is the Investigator for CIRDES of the TRYPANOGEN Project of H3Africa funded by Wellcome Trust.

Gustave Simo
Department of Biochemistry, University of Dschang
Senior Lecturer at the Department of Biochemistry of the Faculty of Science of the University of Dschang, Cameroon. Gustave Simo received his PhD in 2001 from the University of Yaoundé 1, Cameroon. He serves as a Senior Lecturer at the Faculty of Science of the University of Dschang, Cameroon. He served as a Visiting Scientist at Bay Paul Center of the Marine Biological Laboratory,Woods Hole. Simo was Alexander Von Humboldt Fellow at the Division of Functional Genome analysis of DKFZ, Heidelberg. He conducted studies on the animal reservoir of T.b. gambiense, characterization of trypanosomes, transcriptomics of T. brucei. He is collaborating with UMR177 (France), IMT (Belgium), DKFZ and ZMBH (Germany), IRET (Gabon), CIRDES (Burkina Faso), INRB and University of Kinshasa (DRC). His research involves field studies and basic research. His current ongoing projects are focused on the genetic of tsetse and trypanosomes, drug resistance in trypanosome, miRNA expression and human susceptibility to trypanosome infections. He is coapplicant of the TrypanoGEN project of the H3Africa initiative.

Burden, spectrum and aetiology of type 2 diabetes in sub-Saharan Africa
PI: Albert Amoah
Organization: University of Ghana, Ghana
Our primary aim is to assess the burden and aetiological characteristics of T2D in adults in SSA using large scale population based approaches. To achieve this, we aim to develop a large scale epidemiological and genomic research resource comprising up to 12,000 cases of T2D and a population based cross-sectional study of up to 12,000 participants drawn from diverse sampling frames across SSA. Scientific objectives:

1)To assess the burden and spectrum of T2D in adults;
2)To investigate the environmental and genetic determinants of T2D in SSA;
3)To characterise the prevalence and distribution of microvascular complications associated with T2D; and

4) To study the environmental and genetic determinants of microvascular complications associated with T2D.

The research will be supported by cross-cutting research activities, which will be common to the work of most, if not all, the scientific objectives, including:
1) networking and management;
2) epidemiological design and statistical analysis;
3) bioinformatics;
4) genomics and statistical genetics;
5) capacity building; and
6) bioethics.
To achieve these scientific objectives—and as part of the vision for a sustainable research network—local capacity building, including researchers and infrastructure, will be a fundamental component of the proposed research programme.

Albert Amoah
University of Ghana Medical School
Albert Amoah is a Professor of Medicine at the University of Ghana Medical School(UGMS), the Director of the National Diabetes Management and Research Centre(NDMRC) at the Korle Bu Teaching Hospital, and Head of the Diabetes Research and Chronic Diseases Reference Laboratory. UGMS, Accra, Ghana. He is a Consultant Physician to the Korle Bu Teaching Hospital. He holds a doctor of Medicine and Surgery(MB.ChB) degree from the University of Ghana, a PhD in Biochemistry from the University of Surrey and a number of fellowships including the European Society of Cardiology and the Ghana Academy of Arts and Sciences. Professor Amoah has a research focus on the pathophysiology and genetics of Type 2 Diabetes and aetiopathogenesis of atherosclerosis. He has served as Vice Dean of UGMS, Deputy Provost/Acting Provost of the College of Health Sciences, University of Ghana, President of the Pan African Society of Cardiology and on the Executive Boards of the World Heart Federation and the International Society on Hypertension in Blacks(ISHIB). He is the PI of the Wellcome Trust funded H3A Diabetes Study.

Kenneth Ekoru
MRC/UVRI Uganda Research Unit
Kenneth is a Visiting scholar at the International Health Research Group at the Department and the Genetic Epidemiology Group based at the Wellcome Trust Sanger Institute. His research focusses on methods of aggregating data from multiple surveys with differing clustering structures. In particular he is interested in maximizing the utility of data from multiple surveys in identifying environmental and genetic determinants of selected non-communicable diseases in African populations. For this work, Kenneth is meta-analysing data from fifteen sites across the region. He is also part of the H3Africa daibetes study team that is set to conduct large scale population based studies to quantify the burden of and study risk factors for Type 2 Diabetes in Sub-saharan Africa.

Paulina Tindana
Navrongo Health Research Centre
I am a bioethicist by training and a senior research officer/postdoctoral researcher at the Navrongo Health Research Centre in northern Ghana. I recently completed my doctoral thesis at the University of Oxford which examined the ethical issues rising in the collection, export and storage of human biological samples in Africa. Prior to this I was a consulting bioethicist on the Ethical, Social and Cultural (ESC) program of the Grand Challenges in Global Health. My work has involved conducting empirical research on informed consent, community engagement in health research, reproductive health issues as well as drug advertisements on health seeking behavior. I am a member of the H3africa Ethics and Regulatory Issues working group. My current research interests lie in understanding the ethical dimensions of international collaborative research, and particularly the ethics of genomic research in LMIC settings.

Eugene Sobngwi

University of Yaounde 1
Professor Eugene Sobngwi (MD, MPhil, PhD) is affiliated with the University of Yaoundé 1, Cameroon and the University of Newcastle upon Tyne, UK. He is also a Consultant Endocrinologist at the Central Hospital in Yaoundé.In 1996, Dr. Sobngwi graduated from the University of Yaoundé with honours from the Faculty of Medicine and worked as a GP. He later trained to become an Endocrinologist in Paris, France and obtained an MPhil in NCD Epidemiology from the University of Newcastle upon Tyne. He has also worked as a lecturer in Endocrinology and a Registrar Specialist at the University of Paris VII, France where he additionally obtained his PhD in Metabolism in 2005
.
His main research focus is the epidemiology of diabetes and other non-communicable disease in populations of African descent, and the pathophysiology of ketosis-prone atypical diabetes. Currently he is investigating the physiological and epidemiological determinants of diabetes in Africa, diabetes care in resource-limited settings and general endocrinology of African populations.

CharlesRotimi
NIH/NHGRI
Charles Rotimi, PhD, a genetic epidemiologist and a biochemist, is a senior investigator in the Inherited Disease Branch of the NHGRI intramural program. He is the Director of the Center for Research on Genomics and Global Health (CRGGH). The mission of this new trans-NIH center is to advance research into the role of culture, lifestyle, genetics and genomics in disease etiology and health disparities. Dr. Rotimi develops genetic epidemiology models and conducts population genetics research that explores the patterns and determinants of common complex diseases in human populations with particular emphasis on populations of the African Diaspora. As a senior investigator and director of the CRGGH, Dr. Rotimi leads a team of researchers across multiple disciplines (Medicine, genetics/genomics, epidemiology, statistics and informatics) to understand the complex interactions between inherited characteristics and environmental factors in disease susceptibility, variable drug response and health disparities. For example, his team conducted the first GWAS of an African American cohort that identified several genetic variants including PMS1, SLC24A4, YWHA7, IPO7, and CACANA1H underlying susceptibility to Hypertension and blood pressure control. His lab also conducted the first and only genome-wide linkage study of Type 2 Diabetes in West Africans (the AADM Study). Dr. Rotimi's lab continues to contribute to the global understanding of human genetic variation and its implication for differential disease distribution, variable drug response and human migration history.

Ayesha Motala
University of KwaZulu-Natal
Professor and Head, Department of Diabetes and Endocrinology, Nelson R Mandela School of Medicine, University of KwaZulu Natal, in Durban and South Africa.
Clinician whose research has focussed on the epidemiology of diabetes in Africans and Indians in the province of KwaZulu-Natal in South Africa.
Member of the H3A Diabetes group (Amoah Group) and represents this group on the phenotype Harmnization Working Group.

David Adeyemi
Awaiting bio

NIH H3Africa Research Projects

Contribution of genetic variation to pharmacokinetic variability and toxicity in patients undergoing multi-drug tuberculosis treatment in Sub-Saharan Africa
PI: Dissou Affolabi
Organization: National Hospital for Tuberculosis and Pulmonary Diseases, Benin

In 2010 there were an estimated 8.8 million incident cases of tuberculosis (TB) globally, with 2.3 million of these reported in Africa, 1.1 million deaths among HIV-negative cases of TB and an additional 0.35 million deaths among people who were HIV-positive. The complex relationship between TB pathogen, host, and drug exposure in the pathogenesis of TB is poorly understood. The treatment regimen that is currently recommended by WHO for new cases of drug-susceptible TB is highly efficacious, with cure rates of around 90% in HIV-negative patients. However, even if all new TB cases were treated and patients were adherent to the treatment, there would still be 10% of patients (i.e. 880,000 patients worldwide, 230,000 patients in Africa) who fail to respond to treatment. Even if adherent to treatment, a proportion of patients, with rifampicin sensitive TB, are slow to respond to medication or are non-responders. The problem is even more complex and serious in HIV infected patients where the efficacy of the current treatments appears to be lower. Still other patients can be treated successfully, but will experience toxicity and thus treatment interruptions. While several potential determinants of the variable response to drug treatment are recognised (e.g. sex, age, ethnicity), much of the variability in response to anti-tuberculosis drugs remains unexplained. In recent years there has been a rapid development in the understanding of the genetics underlying interindividual differences in drug metabolism and treatment efficacy. The field of pharmacogenetics encompasses the study of the heterogeneity in genes related to drug transporters, drug metabolising enzymes and drug targets, in the context of efficacy of treatment and adverse drug reactions. Few studies have been conducted to explore this field for TB disease. Through this study we aim to explore and determine host genetic factors contributing to pharmacokinetic (i.e. drug concentration) and dynamic (i.e. treatment outcome) variability in TB patients. The “RAFAgene” study is a 5 year project which will be nested within two multi-country randomised phase III tuberculosis treatment trials, the OFLOTUB and RAFA trials (reg numbers NCT00216385 and PACTR 201105000291300) conducted in Sub-Saharan Africa. Patients enrolled in the pharmacokinetic studies within these 2 trials will be sampled for genetic analysis (genome-wide and targeted SNPs screening with in vitro confirmation of the biological plausibility of the association between pharmacokinetic and genetic characteristics). The proposed project is led by Dr Dissou Affolabi at the National Hospital for TB and Pulmonary (NHTPD) with partners from the National TB program in Senegal, the University Ignace Deen in Guinea, the University of Cape Town (SA), the Medical Research Council in Durban (South Africa), the University of Liverpool UK and the London School of Hygiene and Tropical Medicine UK.

Systems Biology for Molecular Analysis of Tuberculosis in Ethiopia
PI: Gobena Ameni, Ethiopia
Institute: Addis Ababa University
Mycobacterium tuberculosis (Mtb) is estimated to have infected one third of the world’s population based on reports from surveys on positive skin tuberculin tests. There are 22 high-burden countries globally, among them Ethiopia, accounting for 80% of all active tuberculosis cases. The clonal relatedness of strains circulating in humans and other potential reservoirs is poorly understood. Molecular epidemiology is gaining importance in tracking strains and addressing key public health challenges to prevent and control communicable diseases in Ethiopia, including tuberculosis (TB). Ethiopian pastoralist populations have been neglected despite their vulnerability to various infectious diseases. Surveillance of TB in these areas is also minimal. As pastoralists rely on livestock products, a significant, largely unexplored challenge is the potentially high level of transmission of tuberculosis between livestock and people. There is currently no effective vaccine protecting humans against TB. Another challenge is the need of prolonged antibiotic treatment which, if not properly completed, accelerates the development of multiple-drug resistance of Mtb.

Infectious bacilli are inhaled as aerosolized droplets derived from infected individuals with active TB and potentially livestock. Bacilli reach the lungs, are phagocytosed by alveolar macrophages, invade sub-epithelial layers of lung tissue and cause localized inflammation. Infiltration with immune cells and formation of lung tissue granulomas are hallmarks of TB pathology. After 2-3 weeks of rapid bacterial growth, a containment phase sets in where granulomas develop a markedly fibrous sheath, are diminished in blood vessels and develop hypoxic regions. These morphological changes often result in a persistent, immune tolerant, non-replicative state of Mtb, the so-called latent infection. CD4 T-cells are essential cells for the anti-Mtb defense, knowledge that has emerged with the rising HIV/AIDS epidemic. Active TB is more prevalent in HIV-infected than in immune-competent individuals, and weakening of the human immune system due to malnutrition and chronic helminth infections is expected to aggravate TB outcomes among Ethiopia's pastoralist populations. Modern genomics tools could considerably impact the knowledge of transmission dynamics, strain diversity and molecular interactions of TB with its host environments. In the proposed partnership between Addis Ababa University (AAU) and J. Craig Venter Institute (JCVI), the objective is to build genomics capacity at AAU and apply newly developed skills towards understanding the relationships of active TB disease with Mtb strain type diversity and host components such as the human microbiome and immune responses in the respiratory tract.

Hypothesis: Molecular interactions between TB progression and the host environments are not well understood. Multiple health-compromising factors affecting Ethiopian pastoralists contribute to higher TB-associated morbidity and mortality. Via an integrated system biology approach (characterizing genotypes of Mtb and *M. bovis* strains, surveying immunological responses, profiling the microbiome and host defense proteome in the airways of infected patients), we expect to gain insights into geographical strain diversity and their antibiotic resistance traits, and to generate hypotheses pertaining to the involvement of the respiratory tract microbiome in susceptibility to and progression of TB.

Specific Aim 1. Understand Mtb transmission dynamics among Ethiopian pastoralist populations and their livestock in two geographical regions. We hypothesize that an innovative approach surveying the variable IS6110 chromosome region of 400 to 500 Mtb isolates from patients and spoligotyping of 50-150 *M. bovis* isolates from the livestock of pastoralists will elucidate novel disease transmission rules. Non tuberculosis mycobacteria will be identified from both humans and livestock using HAIN DNA STRIP®-technology**.** Whole genome analysis of selected strains will enhance such data at a higher resolution level. Drug sensitivity patterns of the Mtb isolates will be determined using line probe assay. Such epidemiological data and information on drug sensitivity of Mtb will also be considered to eventually improve public health outcomes.
Specific Aim 2. Obtain preliminary data to understand the influence of the human respiratory tract microbiome on the susceptibility to Mtb infection in the context of a weakened immune system. Ethiopian pastoralists are exposed to conditions weakening the immune system including malnutrition, chronic helminthiasis and other zoonotic pathogens. We hypothesize that such conditions alter the microflora in the respiratory tract that can be surveyed effectively via 16S rDNA profiling. Microbiome data at the genus resolution level will be correlated with indicators of compromised immune states of *circa* 100 surveyed Mtb patients and control subjects.
Specific Aim 3. Obtain preliminary data to understand and characterize the antimicrobial and immune response occurring locally in the respiratory tract of the cohorts described in Aim 2. We anticipate being able to identify antimicrobial and immune response protein signatures from 25 surveyed Mtb patients and control subjects associated with changes in the microbiome and the severity of tuberculosis, using an innovative metaproteomic methodology.

Gobena Ameni
Addis Ababa University
I was borne on September 30,1972 at Ambo District of central Ethiopia. I did my elementary and high school studies in the same District. Thereafter, I joined the Faculty of Veterinary Medicine of Addis Ababa University in September 1990 and obtained DVM degree in July 1996. I obtained phD in Microbiology from the Imperial College of Science and Technology in February 2009. I also got

Diploma of Imperial College in Immunology in 2009. I joined Addis Ababa University as an Assistant Lecturer in May 1997 and presently, I am a professorial candidate at the Addis Ababa University.Tuberculosis and histoplasmosis are my research interests and I have published over 80 articles on peer-reviewed articles. I led several national and international projects. Among others, I was the PI and coordinator of NUFU (Norwegian),Melinda and Bill Gates, and two work-packages of the Wellcome Trust projects, all three on tuberculosis (human and animal). I taught over 12 different courses to undergraduate and graduate students of the Addis Ababa University. Furthermore, I supervised 54 DVM, 80MSc and 4 PhD thesis. Presently, I am supervising 8 PhD students.

Val Sheffield
University of Iowa
I am a medical and molecular geneticist with broad expertise in the molecular genetics of human genetics diseases, as well as clinical genetics. My laboratory has a long history of identifying and understanding the functions of genes that cause a variety of human disorders, including but not limited to retinal diseases and glaucoma. My research efforts have focused on the molecular genetics of monogenic disorders that have phenotypic overlap with common complex diseases. This approach provides insights into the types of genes, mutational events, and gene product interactions that contribute to common complex disorders. A long- term project in his laboratory has focused on the study of Bardet-Biedl syndrome, a project that helped lead to extensive interest in the scientific community in the signaling role of primary cilia. My laboratory has had an active role in the human and the rat genome projects, and uses gene transfer technologies and knockout mouse models to understand how mutations in the genes identified by the laboratory lead to disease in the affected tissues. Recent efforts in my laboratory include using mouse models to develop treatments with the eventual goal of developing interventions for human disease.

Host and Microbial Genetic Determinants of Febrile Illness in West Africa
PI: Christian Happi
Institute: Redeemer's University, Nigeria
This study aims to investigate host and microbial genomic determinants of febrile illness in West Africa by establishing the West African Genomic Research (WAGER) Network. The WAGER Network consists of West African Francophone and Anglophone centers of excellence in Nigeria, Sierra-Leone and Senegal. The partners in these three countries have a long-term track record of successful collaboration with one another and with US partners at the Broad Institute, Harvard University, and Tulane University studying the devastating diseases Lassa fever and *Plasmodium falciparum* malaria. Efforts in this project will be devoted to using the next-generation sequencing and microbial metagenomics approaches to uncover new microoraganisms and expand our understanding of disease-associated (fever) pathogens in Sub-Saharan Africa. As new microbes are identified they can be incorporated into field-deployable tools to continuously enhance diagnostic tools, creating a positive cycle that supports local and international efforts. We will develop, validate, and implement a qPCR-HRM diagnostic panel of ~100 previously identified FUO-causing organisms in West Africa and new microbes identified in this project. Where possible, we will conduct further study of genetic diversity and clinical manifestations of identified microbes.

In this project we will greatly enhance efforts for training, supporting and promoting cutting-edge genomics research on health in Africa. We will carry out regular and rigorous trainings at the Broad Institute and at WAGER sites, developing the WAGER program into a larger training program that can be used by all H3 Africa partners. We will create a core genomics center at Redeemer's University with an Illumina MiSeq and lay the groundwork to collect 400 high quality samples from febrile patient per year from each of the three WAGER field sites. With these foundations in place, we will use both field-deployed and state-of-the-art genomic technology to identify pathogens driving febrile illness and to examine the genetic basis of infectious disease susceptibility.

Onikepe Folarin
Redeemer's University
I'm a senior lecturer of Department of Biological sciences, College of Natural Sciences, Redeemer's University. I received training in molecular Biology at the Malaria Research Laboratories, IAMRAT, University of Ibadan from 2000- 2009 during my PhD and post-doctoral training. I have a track record of being involved in molecular biology and molecular epidemiology of antimalarial drug resistant parasites by monitoring polymorphisms on the molecular markers of resistance. In the past 5 years, I have been involved in the human genomics and Lassa fever genomics, through NIAID and NIH funded project as well as the 1000 Genome project in which I am a research team leader in Nigeria. The genomics research all aim at developing new diagnostics tools and therapeutics for the Lassa Virus as well as unravelling the basis of human genetic susceptibility to Lassa virus. I have used acquired knowledge in the field of molecular biology and Genomics to train undergraduate and graduate students on molecular biology techniques and applications.

Clinical and Genetic Studies of Hereditary Neurological Disorders in Mali
PI: Guida Landoure
Institute: University of Bamako, Mali
Hereditary neurological disorders affect millions of people worldwide, present with all modes of inheritance, and can start early or later in life. However, despite the vast diversity of African populations, genetic studies in Africa have been limited. African populations, Malians in particular, have a high rate of intra-ethnic and consanguineous marriage, resulting in increased prevalence of autosomal recessive diseases. Family-based genetic studies can be limited in developed countries due to small sibships. The average fertility rate in Mali is over 6 births per woman, offering a unique opportunity to find new disease genes or mutations that can then be studied in other populations. Hereditary neurological diseases are very debilitating diseases, and developing countries, particularly in Africa, pay a high price in terms of disability-adjusted life years. Although most are currently untreatable, increasing awareness about hereditary neurological disorders can reduce this burden.
This study will help to identify and characterize novel hereditary neurological disease genes in Mali. These genes are likely to be important in the normal function of the nervous system and to have important pathophysiological implications for African and other populations. This study will also train physicians and students in the characterization of neurodegenerative diseases as well as in genetic technology and molecular biology, and build a suitable research environment that will retain them.
Guida Landoure
Universite des Sciences, des Techniques et des Technologies de Bamako
I am a medical doctor trained in Mali in general medicine (2002), and moved to NIH (2004) at the Neurogenetics Branch/NINDS for a 3-year training. In Januray 2008, I enrolled in a PhD program at the University College London, London UK in Neurology and Medical Genetics which I completed in July 2011. Since October 2011, I returned to Mali to settle a lab working on hereditary neurological disorders. I also work at the Neurology Department of the Teaching Hospital of Point "G", Bamako, Mali.

Mahamadou Traore
Institut National de Recherche en Sante Publique
I have got my PhD in Medical Genetics in 1987 in Budapest, Hungary. Soon after, I was hired by the National Institute of Research in Public Health (INRSP) as Research Director. In 1990, I was appointed as Associate Professor at the Faculty of Medicine, Pharmacy and Dentistry, and became Associate Professor in 2001. My research interest is in cytogenetics and reproductive biology.

The Nasopharyngeal Microbiome and Respiratory Disease in African Children
PI: Mark Nicol
Institute: University of Cape Town, South Africa

The central role of the infant nasopharyngeal (NP) microbiome in the development of pneumonia or wheezing illness has been demonstrated by analysis of specific components of the microbiome in

children in developed countries. No comprehensive, longitudinal study of the NP microbiome has yet been undertaken, nor related to these outcome measures. We will apply sequence-, culture- and specific PCR-based approaches to define the composition and dynamics of the infant NP microbiome from birth to 2 years of age and to determine the association between the NP microbiome and pneumonia or wheezing in African infants. We will further investigate the association between risk factors for pneumonia or wheezing and the NP microbiome.
Studies will be nested within an existing, funded birth cohort study. All 500 children enrolled in the cohort will have NP sampling at birth, 6 weeks and 6-monthly for the first 2 years. In addition, NP sampling will be done at two-week intervals in a sub-group of 300 children over the first year of life. Samples will be archived for later retrieval and analysis in nested case-control studies. All cases of pneumonia, first episode wheezing or recurrent wheezing will be prospectively identified and investigated for etiology. Case-control studies of children with pneumonia or wheezing will be done. Controls matched by age, clinic site and HIV status will be selected from the same study population. Archived NP samples from cases and controls will be retrieved to perform detailed assessment of the composition and dynamics of the NP microbiome. For pneumonia cases we will focus on the samples collected in the 3 months preceding pneumonia to identify changes associated with near-term progression. For recurrent wheezing we will focus on the association with the composition of the NP microbiome at birth, 6 weeks, 6 months and 12 months of age. Techniques used to define the microbiome will include culture for common respiratory pathogens, specific multiplex molecular detection of 33 different viral, fungal and bacterial pathogens as well as sequence-based microbiome analysis for bacterial microorganisms.
Analysis will use a time-series approach which will allow adjustment for seasonal variation and assessment of the evolution of diversity. In order to identify the determinants of pneumonia or wheezing at each time point we will use weighted generalized ridge regression methods, which are able to select variables in a high dimension setting. We will further gauge the strength of the predictors thus identified as a function of time. In order to integrate viral and bacterial data, both components of the microbiome will be included in the SARIMA model.

Capacity building is a major focus. In particular, we propose to perform the complete microbiome pipeline within South Africa, including high throughput sequencing. Substantial emphasis is placed on training including training visits both to and from JCVI, a training workshop and training of postgraduate students. By focusing on training and ensuring that the complete pipeline (from sequence generation to final association analysis) is performed in South Africa we aim to build independent and sustainable capacity in this field.

Mark Nicol
University of Cape Town
Mark Nicol is head of the Division of Medical Microbiology, University of Cape Town and holds a joint appointment with the National Health Laboratory Service of South Africa. His primary research interests are in respiratory illness in children and novel diagnostics for tuberculosis.

Widaad Zemanay
University of Cape Town
Training: PhD in Medical Biochemistry (Focus on proteomics)
Currently:Research Manager in the Division of Medical Microbiology at the University of Cape Town

Reprogramming of the Trypanosoma brucei Epigenome during Human Infection: Opportunities for New Therapies
PI: Hugh Patterton
Institute: University of the Free State, South Africa

The aim of this study is to map any reprogramming of the *Trypanosoma brucei* epigenome that may accompany movement of this trypanosome from the insect to the human host. This will be undertaken in two projects:

1) The first project involves the genome-wide mapping of nucleosome positions in *T. brucei*. Nucleosome positions will be mapped by paired-end sequencing of 147 bp nucleosome fragments isolated from procyclic fly form (PF) and from human bloodform (BF) cultures. The sequenced fragments will be aligned to the *T. brucei* genome, and the nucleosome positions mapped relative to the polycistronic transcription units and silent genomic loci. It was suggested that in the absence of a defined transcription start sequence in *T. brucei*, the correct positioning of an RNA polymerase at the start of a polycistronic unit was conferred by local chromatin structure and histone modifications. It is therefore expected that nucleosome positions and histone modifications may play a major role in the epigenetic control of transcription in *T. brucei*.

2) Intriguingly, the *T. brucei* histone H3 tail lacks K9, generally associated with transcriptional repression in the tri-methylated state. In the second project, the type and position of modifications of the H3 tail, as well as the genome-wide distribution of select modifications, will be determined by LC-MS/MS and by ChIP-seq in PF and BF cultures. Particular attention will be paid to modifications associated with silent genomic loci and with silent Expression Sites (ES). A single Variable Surface Glycoprotein (VSG) is mono-allelically expressed from one of approximately 15 ESs. Switching of the ES and the expressed VSG allows continual evasion of the human immune system by *T. brucei*.

It is likely that insight into the epigenomic reprogramming of *T. brucei* associated with human infection will identify novel epigenetic targets for future drug development.

Hugh Patterton
University of the Free State
Graduated with Ph.D. in Biochemistry in 1991. Performed post-docs at NIDDK, NIH and Penn State in chromatin structure and gene regulation. Developed research career in epigenomics and bioinformatics.

The Genomics of Schizophrenia in the South African Xhosa People
PIs: Dan Stein, Ezra Susser and Mary-Claire King
Institutes: University of Cape Town, South Africa, Columbia University, U.S.A. and the University of Washington, U.S.A.
Co-investigator: Raj Ramesar
Institute: University of Cape Town, South Africa
The goal of this multisite, collaborative project is to identify genes responsible for schizophrenia in the Xhosa population of South Africa. The vast majority of the genetic basis for schizophrenia has yet to be explained. Strong evidence suggests that individually rare, severely deleterious mutations are responsible for a substantial portion of cases. We hypothesize that critical genes will harbor multiple mutations leading to the disorder, with each affected individual harboring a different severe mutation. We propose a two-step approach to identify these genes. In a discovery series of 200 Xhosa individuals with schizophrenia and 200 age- and sex-matched Xhosa controls, we will sequence complete exomes and related regulatory regions to identify all rare point mutations and indels. In parallel, we will evaluate the same individuals for copy number variants (CNVs) genome-wide (Aim 1). Next, we will validate promising variants and use bioinformatic strategies to prioritize approximately 250 candidate genes most likely to be relevant to schizophrenia. We will develop custom solution pools to capture these candidate genes, and characterize their mutational spectra in an independent series of 900 Xhosa individuals with schizophrenia and 900 Xhosa controls (Aim 2). We will identify genes enriched for deleterious mutations (Aim 3). This project will be the first to use massively parallel genomic sequencing to study schizophrenia in any population of sub-Saharan African lineage. If successful, our approach will identify multiple genes important for the disorder in populations worldwide. Each of these genes will stimulate future efforts to develop more effective treatment and prevention strategies.

Dan Stein
Dept of Psychiatry and Mental Health, University of Cape Town
Dan Joseph Stein, BSc, MB ChB, FRCPC, FRSSAf, PhD, DPhil is Professor and Chair of the Dept of Psychiatry and Mental Health at the University of Cape Town, and Director of the MRC Unit on Anxiety and Stress Disorders.

Rajkumar Ramesar
University of Cape Town
Raj Ramesar is Professor and Head of the Division of Human Genetics at the University of Cape Town (UCT) and its Allied Hospitals in South Africa. As Director of the MRC Human Genetics Research Unit at the University of Cape Town, the emphasis of Raj's research has been on disease susceptibility in African populations, progressing from the commonly recognised inherited diseases, to those that are more complex yet more common and relevant to our large burden of disease, e.g. hypertension, bipolar disorder, amongst others. The richness of African population biodiversity has led to Raj's latest quest to establish a research programme, 'Heritage' that crosses all academic boundaries and celebrates our origins, our diversity (lineages, cultures, languages), while using this opportunity to identify those genomic fragments which predispose us to disease. This quest will contribute to a more proactive and preventative approach to health. Apart from being on the editorial board of several international journals, Raj serves on the Executive of the African Society for Human Genetics, and is its Liaison Officer to the International Federation of Human Genetics Societies.

Morne Du Plessis
University of Cape Town
I am currently based at the University of Cape Town where I am working on a project titled: The Genomics of Schizophrenia in the South African Xhosa People. I have a research background in Human Molecular Genetics as well as Genomics. My interest lies in the use of Next Generation technologies and the associated Bioinformatics tools to unravel the complexity of Human Genetic Disorders.

NIH H3Africa Collaborative Center Research Projects

African Collaborative Center for Microbiome and Genomics Research (ACCME)

PI: Clement Adebamowo
Institute: Institute of Human Virology, Nigeria

The African Collaborative Center for Microbiome and Genomics Research (ACCME) implements 3 interlinked scientific projects, a training and capacity developing project and a grants administration and management project focused on understanding the associations between high risk HPV infection, vaginal microenvironment, HPV genomics, germline and somatic mutations in the etiology of cervical cancer. Cervical cancer is the most common cancer in Africa but significant regional variation in incidence suggest that the risk factors and their interplay with genetic factors vary from one part of Africa to the next. Our preliminary studies already suggest that the most prevalent high risk HPV infections – a necessary, but not sufficient cause of cervical cancer – may be different in African populations compared to the rest of the world. There are also opportunities to identify specific germline risk factors, specific patterns of somatic genomic and epigenomics as well as community state types of vaginal microbiota in African women that may be associated with risk of persistent infection
The center will implement projects including Epidemiology and Biomarker Discovery of Persistent high risk HPV (hrHPV) infection and Cervical Cancer in African women at risk of cervical cancer (The EPIC-Biomarker Study); Discovery of Biomarkers of the association between the Vaginal Microenvironment and persistent high risk HPV (hrHPV) infection (The Maverick Study) and the Gene Discovery of Risks of persistent hrHPV infection (hrHPV), cervical cytokines, patterns and stability of vaginal microbiome and CIN2+ (The Gene-Disc) Project.

These projects will provide an integrative approach to modern epidemiology research that studies the multifactorial etiology of a complex disease like cancer of the cervix and serve as a paradigm for studies of similar diseases in future. String linkages are established between the projects and a Training, Capacity Development and Technology Transfer Project (TRACAD) that provides post-doctoral training to highly qualified African candidates through a rigorous and competitive pan-African candidate selection process implemented through the African University of Science and Technology system will be established. In addition, there are short- and medium-term, local and international sandwich trainings in research methods, epidemiology, bioinformatics, biostatistics and data management, laboratory methods, social and behavioral research for African scientists at the IHVN, Abuja, Nigeria; CIDRZ, Lusaka, Zambia; and the AUST, Abuja, Nigeria.

Clement Adebamowo
Institute of Human Virology, Nigeria
Dr. Adebamowo studied Medicine, Surgery and Epidemiology. He is Director of Research at the Institute of Human Virology, Abuja, Nigeria and his research interests are AIDS Associated Cancers, Breast and other cancers, Bioethics. He is PI of 3 NIH funded training programs in Nigeria

Johnson Obilikwu
Institute of Human Virology, Nigeria.
Johnson Obilikwu coordinates major finance functions in the Institute of Human Virology, Nigeria as Associate Director, Finance. He also generates and coordinates the budget spending including the compilation of financial reports of the Institute. As Unit head, he also supervises the Regional financial operations of the organization. Johnson is responsible for preparing the annual financial statements of the Institute. Having worked with the Institute since February 2005, he has acquired more experience and skills in financial administration of multiple grants.Johnson is also a member of two professional bodies: the Institute of Chartered Accountants of Nigeria and Nigeria Institute of Taxation. Before joining Institute of Human Virology, Nigeria in February, 2005, Johnson worked with Setraco Engineering and Construction Company Limited for six years, as an Accountant. He was responsible for maintaining financial records of the Company including preparation of bank reconciliation, the administration of wages and salaries and financial reporting.

Jesse James
Institute of Human Virology, Nigeria
Scientist & Researcher

Eileen Dareng
Oluranti Famooto
Emilomo Ogbe
Sally Akarolo-Anthony

H3Africa Kidney Disease Research Network Organization

PIs: Dwomoa Adu and Lolu Ojo
Institutions: The University of Ghana Medical School, Ghana and the University of Michigan, U.S.A.

It is estimated that more than 500,000 individuals succumb to end stage renal disease annually in sub-Saharan Africa with an additional 50 million people suffering from pre-dialysis chronic kidney disease. Advanced genome-based analysis strategies, such as Mapping by Admixture Linkage Disequilibrium (MALD) in African Americans, have identified a strong association between single gene variants (e.g., MYH9 and AP0L1) and kidney disease. In addition, nearly 20 genetic variants have been linked to childhood onset nephrotic syndrome. Most of these genetic advances in elucidating the etiology of kidney disease have occurred outside sub-Saharan Africa where there is a shortage of genetic experts and the infrastructure for human genomic research is as sparse as the Sahara Desert itself. In this application, we propose to rapidly increase the capacity to conduct

genomic studies of kidney disease in sub-Saharan Africa through a collaborative research network comprised of investigators based at 10 institutions in five African countries - pop. 362 million (Ethiopia, Ghana, Kenya, Nigeria and South Africa) and four North American institutions. The Network will accomplish the following seven objectives: (1) phenotype 8,000 kidney disease cases and controls (1:1); (2) conduct four genetic research projects addressing single gene mutation kidney disorders in affected families, genetic variants of single genes associated with kidney diseases in the populations and genome wide association studies; (3) establish one low-capital intensity, rugged and sustainable genomics research laboratory each in Ghana (University of Ghana) and Nigeria (University of Ibadan); (4) implement a customized six-track training and career development plan for African-based genomic researchers; (5) establish and maintain a Network-wide biospecimen repository that will harmonize seamlessly with the H3Africa Biorepository Grants (RFA-RM-11-011); (6) establish and maintain a Network-wide data management and bioinformatics facility that will effectively integrate with the H3Africa Bioinformatics Network (RFA-RM-11-010) and (7) cooperate and coordinate the activities of this Network with the H3Africa Consortium and the NIH Program Scientists/Staff. This application is submitted by the University of Ghana with substantial institutional support from the University of Michigan.

Dwomoa Adu
University of Ghana Medical School
Consultant Nephrologist. Co-PI on H3Africa Kidney Research Network

Akinlolu Ojo
University of Michigan
Dr. Akinlolu Ojo is a Professor of Medicine and Epidemiology and the Inaugural Florence E. Bingham Research Professor in Nephrology at the University of Michigan. Dr. Ojo received his medical education from the University of Lagos, Nigeria and residency training in internal medicine at the University of Kentucky. He completed nephrology fellowship, PhD in Epidemiologic Science and the Masters of Business Administration (MBA) at the University of Michigan. Dr. Ojo research interest includes clinical transplantation and chronic kidney disease in Black populations. He has over 130 peer-reviewed publications and has served on the editorial boards of several journals and on NIH study sections. Dr. Ojo maintains an active clinical and translational research program with a portfolio of both NIH and industry funding. Active research collaboration includes investigators in Austria, Brazil, Ghana, Jamaica, South Africa, Spain and Taiwan. Dr. Ojo directs the Department of Medicine Global Health Research Initiative and the joint University of Michigan-International Society of Nephrology (UM-ISN) fellowship program to promote the training of nephrologists from developing. Dr. Ojo is an elected member of the American Society of Clinical Investigation (ASCI), the Association of American Physicians (AAP) and the American Clinical and Climatological Association.

Philip Awadalla
Sainte Justine Hospital
Philip Awadalla, Ph.D., is an Associate Professor of Population and Medical Genomics in the Department of Pediatrics at the University of Montreal, and is the Executive Scientific Director of the CARTaGENE project. He was trained at the University of Edinburgh, and has now developed a population and medical genomics group whose main research interests have focused on the development of next-generation genomics approaches, model-based tools and population-based approaches to study disease. These tools have been used in analyses of the Human HapMap project and the 1000 Genomes Project publications. New tools focus on systems genomics approaches, including genomic, transcriptomic and other molecular approaches to capture signal in population-based samples as well tools to capture rare or de novo variants potentially critical to disease phenotypes. His main research interests include: 1) genomic epidemiology of chronic diseases in Quebec through the CARTaGENE project; 2) the genetic and environmental control points of developing world disorders such as sickle cell anemia and infectious diseases such as malaria; and 3) developing -omic approaches to identify causal variants and discover biomarkers for childhood neurodevelopmental disorders, immunodeficiencies and cancers. He currently holds a

Genome Quebec/MDEIE Recruitment Award and the 2012 Joe Doupe, Canadian Society for Clinical Investigation, Investigator of the Year Award.

Charlotte Osafo
University of Ghana Medical School
Consultant physician and nephrologist, Dept of Medicine, UGMS/Korle-Bu Teaching Hospital, Accra- Ghana Graduated from Kwame Nkrumah University of Science and Technology 1994. ISN Fellow 2005, Fellow of West Africa College of Physicians . Co-investigator with H3Africa Kidney Disease Research Network.

Ifeoma Ulasi
College of Medicine, University of Nigeria Enugu Campus
Professor of Medicine and Nephrology, College of Medicine, University of Nigeria.Co-PI: H3 Africa Kidney Disease Research Network, Enugu, Nigeria Centre.Research interests: Preventive Nephrology; Genetics of kidney disease and hypertension; Ethics in clinical trials

Bamidele Tayo
Loyola University Chicago Stritch School of Medicine
Dr. Bamidele Tayo is an Assistant Professor with educational background in Human Nutrition, Genetic Epidemiology, Statistical Genetics, Epidemiology and Community Health. Dr. Tayo is a Fellow of the Royal Statistical Society. He is also a professional member of the International Genetic Epidemiology Society, African Society of Human Genetics, American Society of Human Genetics, American Heart Association, and the New York Academy of Sciences. Dr. Tayo's research interests include genetic association mapping of complex diseases, development and evaluation of statistical methods for genetic linkage and association studies.

Nikki Tiffin
Awaiting Bio

Collaborative African Genomics Network (CAfGEN)
PIs: Gabriel Anabwani, Kekitiinwa Rukyalekere, Moses Joloba, Oathokwa Nkomazana, Sununguko W. Mpoloka, Graeme Mardon
Institute: Botswana – Baylor Children's Clinical Centre of Excellence, Botswana; Baylor College of Medicine Children's Foundation, Uganda; Baylor College of Medicine, U.S.A.; Makerere University, Uganda & University of Botswana, Botswana
Advanced genetic and genomic technologies promise to transform our understanding and approach to human health and disease. Such genomic analyses are now common in Western populations of European descent. Studies of host genetic factors underlying long-term non-progressors of HIV infection have led to new therapies through the identification of loci that are important to *in vivo* control of virus pathogenicity. Similar studies of host genetic factors influencing active TB infection have also identified important loci that could significantly impact the future development of more effective therapeutic and prophylactic strategies. Most of these studies were undertaken *in non-African, adult populations*, although there are more than 2 million new cases of HIV and HIV-TB in Sub-Saharan Africa every year, including more than half a million children. HIV-infected children - who differ from their adult counterparts in their route of acquisition, clinical course, and pathophysiology – have been conspicuously absent, although they potentially have more to ultimately contribute and gain from therapeutic advances.

The Collaborative African Genomics Network (CAfGEN) aims to redress this scientific imbalance by integrating genetic and genomics technologies to probe host factors that are important to the progression of HIV and HIV-TB infection in sub-Saharan African children. The network will incorporate five sites – the Botswana and the Uganda Children's Clinical Centers of Excellence will provide clinical expertise for patient recruitment; Makerere University and the University of Botswana will provide local molecular genetic expertise; and Baylor College of Medicine will provide access to genomics expertise and resources that will ultimately be transitioned to African

researchers and institutions in a sustainable manner. The CAfGEN research agenda includes the recruitment of prospective and retrospective cohorts of HIV and HIV-TB infected children; the development of core genomic facilities for sample processing and storage; candidate gene re-sequencing, HLA allelotyping and whole-exome sequencing of patients at the extremes of HIV disease progression; and integrated genomic analyses of active TB progression and associated clinical outcomes using expression quantitative trait loci. These projects will be undertaken through an extensive training and career development plan that will also see significant upgrades in local genomics infrastructure. In so doing, CAfGEN will create a unique, highly synergistic African alliance that can contribute novel and important mechanistic insights to pediatric HIV and HIV-TB disease progression while establishing sustainable genomics technology, expertise, and capacity on the African continent.

Gabriel Anabwani
CAfGEN
Graduated in medicine at the University of Nairobi in 1975 and later completed specialized training in paediatrics (Nairobi, 1980), paediatric cardiology (Glasgow, 1983) and clinical epidemiology (McMaster, 1990). Taught paediatrics and child health at the University of Nairobi and at Moi University (1980-1996) before relocating to Botswana in 1997 when became involved in paediatric HIV work. Currently professor of Paediatrics at Baylor College of Medicine and Executive Director, Botswana-Baylor Children's Clinical Centre of Excellence, Gaborone, Botswana.

Oatlhokwa Nkomazana
University of Botswana
Doctor Oathokwa Nkomazana MBChB FCOphth MSc CEH, is Associate Program Director at the University of Botswana School of Medicine, and an Adjunct Associate Professor of Medicine at the University of Pennsylvania. She received her MBChB from the University of Glasgow in 1993 before interning in Surgery and then Medicine in Scotland. She then served as a Medical Officer in Internal Medicine, Accident and Emergency, and Opthalmology at Princess Marina Hospital in Gabarone, Botswana before becoming a Resident in Opthalmology in Cape Town and Bloemfontein South Africa from 1996 through 2000. In that year, she received her FCOphth from the College of Opthalmologists, RSA. She continued her education later, receiving an MSc CEH from the University of London in 2006. From 2001 through 2006, Dr. Nkomazana worked as a Specialist, Consultant, and Senior Consultant in Opthalmology in Gaborone, becoming the Head of Opthalmology at the Princess Marina Hospital in 2002. From 2005 through 2006 she served as Hospital Superintendent at the same hospital. Internationally, Dr. Nkomazana is a member of the ongoing WHO Guidelines Development group for the WHO/PEPFAR initiative on scaling up transformative medical nursing and midwifery education, as well as the International Consensus Group on AIDS-related CMV retinitis: developing guidelines on the management of CMV retinitis in resource limited settings. She was a member of the Princess Marina Hospitals Ethics Committee from 2007 through 2010. Within Botswana, she chairs the Botswana Eye Care Providers Society and the Technical Committee on the Prevention of Blindness. Up until recently she was the Chair of her institution’s Graduate Medical Education Committee. She is the PI for a number of funded research projects: Botswana MEPI project ($10 000) 2010-2015, Human Resources for Primary Care in Africa (HURAPRIM) (€298 532.00.), 2011-2015; A Comprehensive Mixed-Methods Evaluation of Botswana's National Antiretroviral Therapy Program to Inform Policy on Efficiency and Quality of Care ($500 000) per year for two years 2012-2014.

Adeodata Kekitiinwa
Baylor College of Medicine Children's Foundation Uganda
I am a Ugandan born certified physician, an Associate Clinical Professor of Paediatrics at Baylor College of Medicine and the Executive Director of Baylor College of Medicine Children’s Foundation-Uganda (Baylor-Uganda) since 2004. At Baylor Uganda, I lead a team of over 370 personnel some of whom offer comprehensive HIV care at the Children Clinical Centre of Excellence at Mulago Hospital, Kampala and majority give technical assistance to 23 districts across the country to improve service delivery of comprehensive family centered paediatric and

adolescent HIV/AIDS services. My major focus over the last 7 years has been to reduce the inequities of paediatric HIV service delivery, work closely with the Ministry of Health to design strategies for eliminating paediatric HIV and bridging the gap in human resources for health targeting midwives, a key cadre to elimination of mother-to- child HIV transmission, and laboratory cadres. As the lead Paediatrician at the Children's Clinical centre of Excellence (COE) at Mulago Hospital, the clinic grew from 700 children in 2005 to the current 6000 HIV clients (80% children) and 2771 HIV exposed infants; increased availability of antiretroviral drugs and several other interventions to improve the health outcomes. Under my leadership, Baylor Uganda has expanded comprehensive HIV/AIDS care and treatment to 480 health facilities in 23 districts of Uganda and advocated for children infected and affected by HIV in Uganda. Currently Baylor-Uganda is the lead technical organization in provision of paediatric HIV care and treatment in Uganda. As a member of the PMTCT subcommittee and chair of the Early Infant HIV Diagnosis (EID) subcommittee, Baylor Uganda pioneered EID services at Mulago Hospital. I have since worked with MOH to expand EID services from about 30 HIV specialized clinics in the capital city to over 2000 clinics country wide. My current research efforts have been in the field of paediatrics and Adolescent HIV/AIDS. I was the local Principal Investigator (PI) on the recently concluded largest paediatric trial, the ARROW (Anti-Retroviral Research for Watoto) trial and results from this study were published in the Lancet. I have also been a Principal Investigator on several other studies, published in several peer reviewed journals and supervised both local and international students' research at graduate, post graduate and doctorate levels

Betty Nsangi
Baylor College of Medicine Children's Foundation Uganda
I am an Epidemiologist currently working with Baylor College of Medicine Children's Foundation Uganda (Baylor Uganda). I graduated with a Bachelor's degree in Medicine and Surgery (MBChB) from Mbarara University of Science and Technology (Uganda) in 2002 and thereafter worked with the Infectious Disease Institute as a medical officer where I was involved with managing children and adolescents with HIV. I was an NIH-funded AIDS International Training and Research Program (AITRP) scholar at Baylor College of Medicine in Houston, Texas (D43 TW001036, Mark W.Kline, PI) between January 2006 and December 2010. During the five years of my training, I completed both Masters of Public Health (MPH) (2008) and Doctor of Philosophy (PhD) in Epidemiology (2011) degrees through this funding mechanism. I am currently working with Baylor Uganda as a Research Coordinator and I have an adjunct position as a Clinical Assistant Professor of Pediatrics at Baylor College of Medicine, Houston, Texas. At Baylor Uganda, I'm entrusted with contributing to the building of the research unit at Baylor-Uganda by building the research infra-structural and human resource capacity at Baylor-Uganda as well as attracting research grants. Earlier this year, I was awarded a Global Research Initiative Program (GRIP) grant from NIH to study the incidence and outcomes of tuberculosis in HIV-infected children in Uganda. In addition, I supervise, train, guide and mentor staff in research related activities including proposal writing, abstract and manuscript writing. I also currently supervise two Master of Public Health students' research.

Vincent Tukei
Baylor College of Medicine Children's Foundation Uganda
I am a Ugandan trained medical doctor with a Bachelors degree in Medicine and Surgery (MBChB) obtained from Mbarara University of Science and Technology (Uganda) in 1995; a Masters of Medicine (MMed) degree in Pediatrics and Child Health obtained from Central South University (China) in 2004; and a Masters of Public Health (MPH) degree from the University of Texas (USA), May 2010. I am a Pediatrician and Public Health specialist currently working with Baylor-Uganda; I have over 15 years experience in clinical medicine working in government and non-government health facilities in rural and urban parts of Uganda. For the last 8 years I have worked as an HIV/AIDS specialist at Baylor-Uganda - the largest pediatric HIV/AIDS treatment center in Uganda, and was appointed to senior management positions in this organization. I am currently employed as Manager for Research at Baylor-Uganda and as a trial Pediatrician for an ongoing multicenter clinical trial. In addition, I supervise, train and mentor medical officers and other allied health professionals involved in studies at Baylor-Uganda. I also guide and mentor staff on research

related activities including proposal writing, abstract and manuscript writing, and offer research related strategic direction to the organization with the aim of building capacity in HIV/AIDS related operational and clinical research. I have also published in peer reviewed journals.

Chester Brown
Baylor College of Medicine
We seek to understand gene functions that have important effects on energy metabolism by influencing the metabolic rate, fat cell differentiation, growth and function. We use mouse model systems and cell culture approaches to understand these processes. Clinically, we are now applying emerging genomic technologies to benefit patients, and our collaborations will allow us to develop several lines of translational study, not only related to obesity and metabolism, but also to better understand the genomic basis of HIV and TB susceptibility and progression and other conditions in African children, while helping to build the capacity and infrastructure for African scientists to carry out such studies independently through the H3 Africa initiative. These efforts hold substantial promise to establish collaborations with several African institutions and to benefit patients on many fronts. Human subjects research is a significant new focus for our lab, a decision made because of the clinical importance of the problems and the importance of ethical and culturally sensitive practices for genomic science abroad as we seek to impact global health. Collectively, our team has many years of experience in the clinical, molecular and statistical aspects that are critical for the success of future projects both at home and abroad.

Neil Hanchard
Department of Molecular and Human Genetics, Baylor College of Medicine
Assistant Professor of Molecular and Human Genetics at Baylor College of Medicine in Houston Texas. I am a clinical geneticist and molecular geneticist by training, with a research interest in complex trait mapping in paediatric populations, particularly in populations of African descent. My research background also includes training in population genetics.

Graeme Mardon
Baylor College of Medicine
Dr. Graeme Mardon received his B.S. in 1980 with a double major in Biology and Chemistry from Haverford College. Following a four-year research associate position with Dr. Harold E. Varmus studying the viral oncogene src at the University of California in San Francisco, Dr. Mardon began graduate school at the Massachusetts Institute of Technology in 1984. He received his Ph.D. in 1990 in the laboratory of Dr. David C. Page where he studied genes located in the sex-determining region of the mouse Y chromosome. Dr. Mardon then conducted his postdoctoral work with Dr. Gerald M. Rubin at the University of California in Berkeley from 1990 to 1994 studying genes required for normal eye development in the fruit fly Drosophila melanogaster. Dr. Mardon joined the faculty at Baylor College of Medicine in 1994 where he has established a research program studying the molecular genetics of developmental neurobiology using both the fruit fly Drosophila and the mouse as animal model systems. Over the last decade, Dr. Mardon has employed genomics approaches to study retinal development in Drosophila and humans.

Marape Marape
Botswana-Baylor Children's Clinical Centre of excellence
Marape Marape MB, BCh, BAO, MPH, PhD
Assistant Professor, Pediatrics, Baylor College of Medicine
Public Health Specialist and Director of Research, Botswana-Baylor Children's Clinical Centre of Excellence - Gaborone, Botswana
Co-Investigator , CAfGEN.
Research Interest: Cinical, Psychosocial and Genetic Determinants of variable disease expression among children, especially Tuberculosis and HIV/AIDS.

Masego Tsimako-Johnstone
University of Botswana
I am a lecturer at the university of Botswana My background is biochemistry and Molecular Sciences. I am involved in numerous research projects where I collaborate nationally and internationally. I am also a I am a co-investigator in the CAFGEN H3 Africa grant proposal.

Ishmael Kasvosve
Awaiting Bio

Sununguko Wata Mpoloka
Awaiting Bio

Stroke Investigative Research & Educational Network (SIREN)
PI: Mayowa Owolabi and Bruce Ovbiagele
Institute: University of Ibadan, Nigeria and Medical University of South Carolina, U.S.A.

Although stroke is a discrete phenotype, it is a clinical culmination of several complex and interacting biological processes, precipitated by various genetic and environmental factors, thereby making ready analyses of its underlying mechanisms a challenge. Nevertheless, a better understanding of the predisposing risk factors for stroke in Africa is imperative for prediction of its occurrence, subtype, severity and prognosis, as well as the formulation of successful tailor-made prevention programs. The overall goal of SIREN (Stroke Invclementestigative Research and Education Network), is to evaluate the premier genetic and environmental risk factors for stroke in Africa, while simultaneously training professionals and building sustainable capacities in phenomics, biobanking, genomics, biostatistics and bioinformatics for the high-level investigation of stroke . We will accomplish the goal of SIREN via a concise network of research and educational collaborations within Africa, along with inputs from renowned content experts in United States and United Kingdom. SIREN will be comprised of three interwoven projects geared at a comprehensive investigation of stroke and a training core aimed at building research capacity that will outlast the study period. The Systematic Investigation of Blacks with Stroke (SIBS) will involve three interconnected projects: SIBS-Phenomics, SIBS-Genomics and SIBS-Bioinformatics. SIBS projects will be implemented at eight sites in three countries (Nigeria, Ghana, and South Africa).
Specific Aims

1) To evaluate the qualitative and quantitative contributions of established and novel sociodemographic, clinical, biochemical, radiological, and genetic factors to stroke occurrence, subtype and outcome patterns in Africa.
2) To elucidate the current knowledge, attitude, beliefs and practices about genetic testing for stroke in Africa.
3) To collect and bank genomic material for future analysis using emerging next gen techniques.
4) To compare environmental and genetic risk factors for stroke among Black Africans in Africa to Black Americans.

Mayowa Ojo Owalabi
University College Hospital
Dr. Mayowa Owolabi, *MBBS,MWACP,FMCP, Cert. Neurol., Dr. med.-magna cum laude (Berlin), Cert Epid& Glob Health (Dundee), MSc(distinction)* is a Consultant Neurologist and the Subdean(Postgraduate), Faculty of Clinical Sciences, University of Ibadan, Ibadan, Nigeria. He has more than 10 years' experience in collaborative studies (Germany, Canada, USA) in vascular neurology including research on risk factors for stroke (INTERSTROKE) and stroke prevention. As the site PI for SIRROWS and SIRRACT, he participated in innovative multicenter randomized controlled trials for rehabilitation after stroke. In a multidisciplinary study (of which he was the PI) funded by a special Mac-Arthur grant, he pioneered research into carotid atherosclerosis in Nigeria. Dr. Mayowa Owolabi is the pioneering and current regional vice president of the World Federation for NeuroRehabilitation in East, West and Central Africa and has garnered experience in stroke

epidemiology and outcome measurement. As an innovative researcher, he developed the Seed of Life Model, the Health-Related Quality of Life in Stroke Patients (HRQOLISP) questionnaire, the Stroke Levity Scale and stroke recovery cycle. He also propounded the cervical vertigo tetrad. In 2008, he won the American Academy of Neurology International Scholarship Award. He got the AU-TWAS (African Union-Academy of Sciences for the developing world) Young Scientist National Award in 2011. In the same year, he won 2 additional awards: the American Academy of Neurology – Palatucci Advocacy Leadership Award as well as the InterAcademy Medical Panel Award for Young Physician Leaders.Dr. Owolabi has over 100 scientific publications to his credit. He serves on the Editorial Board of the Journal of Neurological Sciences, World Journal of Neurology and West Indian Medical Journal. He is a member of the American Academy of Neurology (AAN) as well as the American Society of Human Genetics. On the need for collaborative translational neuroscience research in Africa he wrote a WFN-president acknowledged review on mapping Africa into prominence in Neurology and a recent review on taming the exploding scourge of stroke in Africa: stroke quadrangle to the rescue. A critical component of the stroke quadrangle is prevention. For this pillar, he is the Co-PI of a NIH-funded multi-pronged intervention study (THRIVES). However, stroke prevention efforts rely on full understanding of genetic and environmental risk factors for stroke. Therefore, he plans to establish this crucial pillar of the stroke quadrangle by filling the knowledge gap while building capacities and careers for translational genomic research through the SIREN (Stroke Investigative Research and Educational Network) Projects.

Genomic and environmental risk factors for cardiometabolic diseases in Africans (AWI-Gen)
PI: Michèle Ramsay
Institute: University of the Witwatersrand & NHLS, South Africa
Co-PI: Osman Sankoh
Institute: INDEPTH, Ghana
The long-term vision of the Collaborative Centre (AWI-Gen) is to build sustainable capacity in Africa for research that leads to an understanding of the interplay between genetic, epigenetic and environmental risk factors for obesity and related cardiometabolic diseases (CMD) in sub-Saharan Africa. AWI-Gen will be consolidated under the auspices of the University of the Witwatersrand (Wits) and the International Network for the Demographic Evaluation of Populations and Their Health in Low- and Middle-income Countries (INDEPTH). It will capitalize on the unique strengths of existing longitudinal cohorts, including the urban Soweto and rural Agincourt studies in South Africa (Wits based), and the well established INDEPTH demographic health and surveillance centers in Kenya, Ghana, Burkina Faso and South Africa. The centers offer established infrastructure, trained fieldworkers, long-standing community engagement, and detailed longitudinal phenotypic data, focusing on obesity and cardiometabolic health. Key strengths are harmonized phenotyping across sites, building on strong existing cohorts, and representation of the geographic and social variability of African populations. We aim to: 1. Build sustainable infrastructure (biobanks and laboratories) and capabilities (well characterized population cohorts, genotyping and bioinformatics) for genomic research on the African continent; 2. Understand the genomic architecture of sub-Saharan populations from west, east and south Africa to guide genomic studies (genome sequencing and high throughput SNP and CNV arrays using unrelated individuals and family trios to improve the accuracy of haplotype analyses) and; 3. Investigate the independent and synergistic genomic contributions to body fat distribution (BMI, hip/waist circumference, subcutaneous and visceral fat) in these populations considering the relevant environmental and social contexts (rural/urban communities, quickly transitioning obesity prevalence, differential HIV, TB, and malaria infection histories). We will investigate the effect of obesity and fat distribution on the risk for CMD in the longitudinal cohorts. AWI-Gen will draw upon a wide group of highly experienced African scientists and international collaborators to ensure the success of its vision.

Michele Ramsay
Wits and NHLS
Professor in the Division of Human Genetics, NHLS and University of the Witwatersrand (Wits), and Interim Director of the Sydney Brenner Institute for Molecular Bioscience (SBIMB). Her research

interests include African population genetic and epigenetic diversity and their role in diseases exacerbated by adverse lifestyle choices, including obesity and cardiometabolic disease and foetal alcohol spectrum disorders (FASD). Research collaborations include studies on obesity, hypertension, bone development, HIV related kidney disease and glaucoma in South African populations. She is joint PI for an NIH funded training program "Wits Non-Communicable Disease Research Leadership Program", chair of the Southern African Society for Human Genetics and chair of the Wits Bioinformatics Steering Group. She is PI for an H3Africa NIH funded Collaborative Centre, AWI-Gen, which is a Wits-INDEPTH partnership to study genomic and environmental risk factors for cardiometabolic diseases in Africans. The views expressed in this email are, unless otherwise stated, those of the author and not those of the National Health Laboratory Service or its management. The information in this e-mail is confidential and is intended solely for the addressee. Access to this e-mail by anyone else is unauthorized. If you are not the intended recipient, any disclosure, copying, distribution or any action taken or omitted in reliance on this, is prohibited and may be unlawful. Whilst all reasonable steps are taken to ensure the accuracy and integrity of information and data transmitted electronically and to preserve the confidentiality thereof, no liability or responsibility whatsoever is accepted if information or data is, for whatever reason, corrupted or does not reach its intended destination.

Daniel Achinko
National Health Laboratory Systems, Division of Human Genetics, South Africa
I am currently a Post-Doctoral student With the Sydney Brenner Institute of the University of Witwatersrand. I am undertaking a Human genetics program looking at GWAS in regards to obesity disease in the black African population of South Africa

Kay Elizabeth Davies
University of Oxford
Kay Davies is the Dr Lee's Professor of Anatomy and, Associate Head (Development, Impact and Equality) Medical Sciences Division, Honorary Director of the MRC Functional Genetics Unit and co-Director of the Henry Wellcome Building of Gene Function at the University of Oxford. Professor Davies's research interests lie in the molecular analysis of human genetic disease, particularly the genetic basis of neuromuscular and neurological disorders in children. She first became interested in muscular dystrophy more than 20 years ago and much of her research group is dedicated to finding effective treatments for Duchenne muscular dystrophy, a devastating muscle wasting disease. She works very closely with the Muscular Dystrophy Campaign and other charities in the USA, France and Italy. In 1999, she set up the MRC Functional Genetics Unit aimed at exploiting genome information for the analysis of the function of genes in the nervous system. In 1999 she co-founded the Henry Wellcome Building of Gene Function with Professor's Ashcroft (Physiology) and Donnelly (Statistics) to bring together genetics, physiology and bioinformatics in a new multidisciplinary building which was completed in 2003. Professor Davies has always been closely associated with the human genome project. From 1993 to 1995 she was Vice President of the International Human Genome Organisation (HUGO). She has an active interest in the ethical implications of genomics research and in promoting public understanding of science. She is a founding fellow of the Academy of Medical Sciences and was elected a Fellow of the Royal Society in 2003. She was awarded a CBE in 1995 and a DBE in 2008 in recognition of her many contributions to medical research. She will be Deputy Chairman of the Wellcome Trust from October 2013

Zane Lombard
University of the Witwatersrand
I am a Senior Lecturer in Bioinformatics at the University of the Witwatersrand. I completed my PhD in Human Genetics & Bioinformatics in 2008, under the supervision of Prof Michele Ramsay (Title of Research: Computational prediction of genetic targets for fetal alcohol spectrum disorders). This study focused on the use of data-mining techniques to prioritize genes in a candidate list, or in the absence of such a list, could be used to rank all genes in the genome, in order to identify putative candidate genes for complex diseases. Following the completion of my PhD I worked at the Division

of Human Genetics (National Health Laboratory Service, Johannesburg) for four years as laboratory manager and postdoctoral researcher, where I managed the genotyping facility and DNA biobank, and participated in several genotyping projects investigating complex traits in African populations. I joined Wits Bioinformatics in 2012, where I currently teach undergraduate and postgraduate courses in Bioinformatics, and continue my research into risk factors for chronic disease in Africans. I am currently involved in the H3Africa AWI-GEN project (Pis – Ramsay & Sankoh) as co-investigator, and building my own research group focused on biocuration of complex datasets and epigenetics of obesity.

Nigel Crowther
National Health Laboratory Service
Joint appointee with NHLS and University of the Witwatersrand and head of research in Department of Chemical Pathology. Research interests include: adipocyte cell biology; genetics and epidemiology of obesity and diabetes; metabolic side effects of HIV infection and anti-retroviral therapy; role of early life events in the aetiology of metabolic diseases; body fat distribution and disease.

Nadia Carstens
University of the Witwatersrand
After completing my M.Sc and PhD in human genetics at Stellenbosch University, I joined Wits Bioinformatics as a postdoctoral fellow as part of the NIH Fogarty Wits Non-communicable disease research leadership program in June 2012. I have a strong interest in complex genetic disorders and will be involved in projects centered on public data mining, genome-wide association analysis and the bioinformatic analysis of next-generation sequencing data to investigate the contribution of genetic variation to non-communicable diseases in African populations during my postdoctoral research. In addition, I participate in AWI-Gen, an H3Africa funded partnership between the University of the Witwatersrand and the INDEPTH network. This project aims to investigate genome structure in sub-Saharan African populations in west, east and South Africa and to identify genetic variants that influence body composition and contribute to susceptibility to cardiometabolic disease in these populations.

Alisha Wade
University of the Witwatersrand
Dr Alisha Wade is a Senior Lecturer in the School of Public Health at the University of the Witwatersrand in Johannesburg, South Africa. She was the Clinical Gold Medalist in her graduating medical class at the University of the West Indies and obtained her Doctor of Philosophy degree in Clinical Medicine from the University of Oxford, which she attended as a Rhodes Scholar. This was followed by a residency in Internal Medicine at the Johns Hopkins Bayview Medical Center, United States. Dr Wade then completed a Fellowship in Endocrinology, Diabetes and Metabolism at the University of Pennsylvania. Her research focuses on the determinants of the chronic disease epidemic in developing countries and the development of strategic interventions.

NIH H3Africa Biorepository Pilot Projects

Development of H3 Africa Biorepositories to facilitate studies on Biodiversity, Disease & Pharmacogenomics of African Populations
PI: Akin Abayomi
Institute: National Health Laboratory Services, Stellenbosch University Faculty of Medicine, South Africa.
The objective of this application is to develop a plan towards a full scale H3 Africa Central Biorepository and service facility in a joint collaboration between the National Health Laboratory Services of South Africa and the Faculty of Medicine and Health Science, University of Stellenbosch, in the Division of Haematological Pathology to support H3 Africa and other large research projects on the Continent in a sustainable manner. In phase I, we will set up governance, operations and test protocols towards biorepositories for nucleic acids, blood, haematological

malignancies, cultured stem cells and mesenchymal stromal cells and any other human derived sample as the case may require from the H3 Africa consortium projects. This approach builds upon existing infrastructure and capacity for processing the above sample types that exists in this institution. New perspectives in our approach will be to evaluate room temperature storage of human samples as a sustainable energy efficient option for Africa. Furthermore, we will evaluate the possibility of automation for sample preparation and processing of nucleic acid, storage and cryopreservation in order to meet the expected service demands of 100,000 samples a year in phase II. Our proposed approach to create renewable cell lines would be by both conventional methods and induced Pluripotent Stem Cells (iPSC).There are several key challenges to overcome in this later approach. Although there are many large scale Biorepositories internationally, there are no large human specimens Biorepositories in the African continent, this therefore poses as a challenge to evaluate what is feasible on the African continent.
Generating iPSC is not new in Africa, but it is not readily available and the ethics of this technoilogy is not fully understood, therefore it is essential that a thorough evaluation of the feasibility of our approach be conducted. We have convened a panel of indigenous and international experts in the biobanking and allied fields to develop an agenda for full scale biorepositories in Phase II. They are known as the Governance Advisory Panel (GAP). We have benchmarked international biorepositories and stem cell biobanks, and set up governance that will set the technical and ethical guidelines as well as long term sustainability planning. Furthermore, we are in the process of evaluating current and prospective biorepository operations at the University of Stellenbosch in the Division of hematological pathology against the industry best practices adherence to a set of best practices such as those set by the International Society for Biological and Environmental Repositories (ISBER) and the National Cancer Institute (NCI). We will develop a Laboratory Informatics Management System (LIMS) for all aspects of biorepository operations with our collaborative bioinformatics centre, SANBI at the University of the Western Cape in conjunction with the H3 Bioinformatics network. We will develop automation, iPSC reprogramming and room temperature biobanking with our collaborative centres, the RUCDR in New Jersey, IFASEMB, the SCRIPPS Research Institute, Centre for Regenerative Medicine in San Diego California and the Cape Haematology Stellenbosch University Satellite Bone Marrow Transplant Unit in Cape Town. We will explore the possibility of a partnership with these institutes to promote an agenda for stem cell and nucleic acid large scale biobanking for studies on the health, diseases and pharmacogenomics of African populations. A common theme in our approach will be stringent governance, harmonization and sustainability. Creation of a full scale biorepository will be an important part of scientific capacity building on the African continent and will support large scale genomic studies that will be performed on the continent. These studies and supporting infrastructure are necessary to address the health needs of the continent that is plagued with infectious disease such as HIV/AIDS and its numerous complications as well as an ever increasing burden of non communicable diseases such as the metabolic disease syndromes and cancer. Biorepositories will also function as a repository to preserve representative samples of the vast human biodiversity of the continent, harmonize sample collection efforts, be a training centre for African scientists and a community outreach portal to educate the public about the implications of biobanking and genomics for the health of African populations.

Akin Abayomi
University of Stellenbosch
Prof Emmanuel Akin Abayomi Position: Head of Department of Haematopathology Tygerberg Academic Hospital and Associate Professor of Haematology University of Stellenbosch, Cape Town. Qualifications: MBBS (London), MRCP (UK), FCPath Haem (SA), MPhil (UP), FRCP(Edin) PhD Project: Stem cells in HIV disease. My current position is as Chief Pathologist and Head of the Division of Haematology and Associate Professor Faculty of Health Sciences, University of Stellenbosch University, Cape Town, South Africa (www.sun.ac.za/haema). I am a specialist in internal Medicine and Haematology, I studied at the Royal Medical College of St Bartholomew's Hospital in the University of London where I attained my first graduate degree in Medicine. Specialized in Internal Medicine and Haematology, obtaining fellowships from both Royal College of Medicine in the United Kingdom and the College of Medicine of South Africa. I have worked in

several countries around the world in both Internal Medicine and Haematology and have been exposed to a variety of geographical variations and disease patterns within the discipline of Internal Medicine and Haematology. My focus has mainly been on the complications of HIV and the development of laboratory and clinical capacity to rise to the challenge of the HIV epidemic in the Developing world and Africa.

Ravnit Grewal
Stellenbosch University
Dr Ravnit Grewal is a Board Certified Hematologist, and is currently working at Cape Haematology, Constantiaberg Medi-clinic, Cape Town, South Africa. She was working as a consultant/specialist for the National Health Laboratory Services in Cape Town, Tygerberg Hospital from June 2010 until March 2013. The Division of Hematopathology at Tygerberg Hospital offers both a diagnostic and research platform. She has been involved in most of the routine diagnosis of haematologic conditions for the past 8 years. As a specialist in the Division of Hematopathology, her duties included routine diagnostic work, training and teaching undergraduate medical students and post graduate students (medical and scientists), outreach both nationally and internationally and research. She is currently involved in 4 major research groups.Her areas of expertise involve Hematological diagnosis and reporting including bone marrow flow cytometry and interpretation of molecular tests, teaching of undergraduate students, post graduate students in Hematopathology, Organising outreach programmes in South Africa and other African countries. As a co-investigator on the H3A biobank project, her skills as a pathologist and clinician, fulfil the role of the pathologist in the team.

Beverley van Rooyen
University of Stellenbosch
Dr van Rooyen is a research scientist at the Division of Haematology, Faculty of Medicine and Health Sciences, University of Stellenbosch, South Africa. She has a background in molecular genetics and biochemistry, and completed her PhD in Medical Biochemistry at the University of Cape Town in 2011. As biorepository manager at the NSB-H3A (NHLS Stellenbosch Biobank - H3Africa)she is mainly involved in the development of biobanking infrastructure and technical operations.

Carmen Swanepoel
National Health Laboratory Services/Stellenbosch University
Dr. Carmen Swanepoel is a Molecular Biologist in the Division of Haematology at the National Health Laboratory Services and Stellenbosch University at Tygerberg Hospital, Western Cape, South Africa. She has been concerned with research, development of diagnostic laboratory tests and the teaching and training of staff and students in laboratory skills and quality management. She has been instrumental in the development of the new flow cytometry research and training laboratory as well as the cell culture facility in the Pathology department and her experience in various molecular based techniques, cell culturing and genetic association studies adds to her role as scientist on the NIH H3Africa Biorepository Project. Other research interest includes Mesenchymal Stromal cells and its application in Regenerative medicine as well as Flow cytometry and its application in cytokine detection.

IHVN H3Africa Biorepository (I-HAB) Initiative
PI: Alash'le G. Abimiku,
Institution: Institute of Human Virology, Nigeria (IHVN)
The proposed IHVN H3Africa Biorepository (I-HAB) initiative is directed by Dr. Alash'le Abimiku, a highly experienced African laboratory scientist with two decades of research and repository experience in Africa, and the Principal Investigator of the current proposal. The goal of Phase I implementation is to: *Advance the capacity of the IHVN Biorepository to achieve International Society for Biological and Environmental Repositories (ISBER) best practices required for Phase II implementation.* To achieve the Phase I goal the I-HAB partners with the Coriell Institute to implement three Specific Aims: 1) Assessment of current practice and identify strengths and gaps;

2) Upgrade repository practice and infrastructure; 3) Conduct pilot Phase II implementation. This two year Phase I process engages an iterative quality assessment-based interaction between experienced African scientists and technicians at IHVN and their counterparts from Coriell Institute who provide objective assessment, interactive didactic and mentored capacity building to instil ISBER best practices for Phase II implementation drawing upon Coriell proven models. The goal of Phase II is to: *Expand the capacity of the I-HAB to support multiple H3Africa investigators to conduct high quality genomics and translational research in Africa using well processed, preserved and quality controlled and redundantly protected human biological samples accessible to the H3Africa and larger research community.* To achieve the 5 year Phase II goals the I-HAB targets 5 Specific Aims: 1) Implement a high quality biorepository of primary human biologic samples; 2) Develop, implement, manage and support robust cloud computing based bioinformatics tool to support biorepository capacity; 3) Establish administrative governance and Quality Assurance/Quality Control (QA/QC) procedures and sustainable funding strategies; 4) Conduct short, medium and long term training and mentoring of staff on Biorepository and Biobanking Sciences; 5) Integrate best practices in biobanking and biorepository ethics. This 5 year plan implements a staged expansion of staffing and infrastructure to support multiple African genomic research partners including Dr. Clement Adebamowo, an African scientist also at the IHVN who served as the Principal Investigator of the African Phase I HapMap Project and is a pioneering leader in genomic research and in addressing the ethical challenges of genomic research on the African Continent on the Continent. I-HAB will provide reliable sample processing support, secure shipping, rapid accession and documentation of sample quality, accessible information on clinical and epidemiological data and sample quantity and quality, reliable retrieval and proactive facilitation of collaboration to achieve best science and sustainable practice and funding. Continuous quality improvement for reliable repository function and ongoing feedback from end users fosters trust in service delivery and product quality.

Alash'le Abimiku
Institute of Human Virology Nigeria
Dr. Alash'le Abimiku has been pivotal to the establishment of a long-term collaborations between Institute of Human Virology Nigeria (IHVN) and other Institutions in Nigeria and investigators at the Institute of Human Virology University of Maryland School of Medicine, Baltimore where she is an Associate Professor, the National Institutes of Health and research institutes around the world. Dr. Abimiku has been highly successful in growing portfolio of grants including the Institute of Human Virology-Nigeria PEPFAR program where she directs the activities of the laboratory clinical services and research. As a trained Medical Microbiologist with specialization in Retrovirology, Dr. Abimiku first demonstrated the unique nature of the HIV strain prevalent in Nigeria in 1993 as subtype G during her postdoctoral training at NIH, and provided the first reliable HIV research laboratory in central Nigeria. In addition to being a successful fully funded independent researcher for several years, Dr. Abimiku has trained a cadre of pre and post-doctoral Nigerians who are engaged in advanced laboratory science and research through the UM-IHV NIH Fogarty AIDS International Training and Research Program Grant. She is internationally recognized in her role on the board of the African Society of Laboratory Medicine, and the WHO HIV Vaccine Advisory Committee. She is the Principal Investigator of the IHVN H3Africa Biorepository (I-HAB).

Ndidi Agala
Institute of Human Virology, Nigeria (IHVN)
A medical laboratory Scientist working with the IHVN, in the I-HAB Lab as a supervisor. I have my interests in molecular Biology, Haematology, Immunology and genomics.

Talishiea Croxton
Institute of Human Virology Nigeria
Joint appointee with NHLS and University of the Witwatersrand and head of research in Department of Chemical Pathology. Research interests include: adipocyte cell biology; genetics and epidemiology of obesity and diabetes; metabolic side effects of HIV infection and anti-retroviral therapy; role of early life events in the aetiology of metabolic diseases; body fat distribution and disease.

Charles Mensah
Institute of Human Virology, Nigeria
A Chartered Accountant, a Certified Public Accountant, Health Care Executive, and Grants Administrator trained in Nigeria and U.S.A. with over 28 years of continuous working experience. A well organized, analytically minded leader able to develop and motivate high-functioning teams and optimize interdepartmental communication to ensure the cohesive operation of large-scale international non-profit agencies; strong focus on cost control, resource management, and accountability. Currently serving as the Chief Operating Officer/Managing Director of the Institute of Human Virology, Nigeria supporting two H3 Africa Principal Investigators (Alash'le Abimiku and Clement Adebamowo).

Establishment of an H3Africa Biorepository at Clinical Laboratory Services
PI: Ute Jentsch
Institute: Wits Health Consortium (Pty) Ltd
H3Africa is an important Africa-wide initiative to build capacity in genomics-based research in order to improve health in African populations. The goal of the Clinical Laboratory Services (a Division of the Wits Health Consortium) in Johannesburg South Africa is to become a fully operational H3A Biorepository that receives, processes, stores, and distributes samples for research in Africa, in accordance with international and local operational and ethical standards. Based on its Biorepository experience in the last decade, CLS is equipped to support the aims of H3 Africa by providing Biorepository capacity and expertise. In order to be a key stakeholder in the H3 Africa mission, the project funding will be used to expand and optimize CLS' sample storage infrastructure in order to accommodate samples from H3Africa research sites in a dedicated state of the art facility. The project will also assist with the implementation of important CLS activities in order to better serve the needs of the H3Africa initiative, including the upgrading of its laboratory and sample management software, the potential of using novel, yet cost-effective, sample identification methods to reduce human error relating to sample management and relevant staff training.

The establishment of this repository will be undertaken in two phases. In the first phase, the following aims will be addressed. In the first year CLS will focus on assessing the H3 Africa biorepository needs and enhance its current infra-structure and operations to ensure the H3 Africa Biorepository needs can be met. This includes the final scoping of the required storage facilities, evaluation of new software for data management and sharing, feasibility of proposed robotics and sample labeling. The second aim is to provide training to CLS H3Africa staff, to ensure operations are in keeping with the required standards. The training will include: LDMS, LIMS, IATA, and GCLP, sample handling and equipping staff with the required theoretical knowledge and practical exposure relating to the repository, health and safety and H3 Africa policies and procedures. Thirdly, CLS will endeavor to develop and strengthen networks and collaborations with other H3Africa biorepositories and research sites.

The key aims of the second phase is the full commissioning and operation of the dedicated H3A Biorepository to handle the full scope of H3 Africa research samples in terms of receipt, storage and distribution. This is likely to require the expansion of CLS infrastructure to accommodate the H3A Samples, including the purchase of a dedicated -80℃ robotics freezer system and liquid nitrogen freezers, new air conditioners, and computer hardware, and implementation of process improvement projects such as the upgrading of systems for database management, a robotic sample management system, sample labelling and data sharing.
In all stages specific care will be taken to maintain the high standards of sample protection, as required by CLS research partners.

Dr Ute Jentsch
Clinical Laboratory Services
I, Dr Ute Jentsch, am the Chief Executive of CLS. In that capacity I am responsible for developing and executing the CLS business plan and strategy. I also strive to fulfill our mission statement; which is to be to support state of the art teaching and research for the University of the Witwatersrand, School of Pathology. I have many years experience in the fields of serology and microbiology and its application to clinical trials. I have also facilitated the set-up of TB diagnostics in the Biosafely Level 3 laboratory at CLS. Between 2003 and 2009, I was responsible for coordinating the central laboratory role and activities in the Microbicides Development Program. Together with other CLS Project Managers, I currently collaborate in several major studies, involoving the following networks, namely IAVI, PrEP, GSK, MVI, MTN, ACTG and the TB Alliance. A recent interest of mine is the field of sample storage. The CLS Biorepository has grown significantly over the last years as most clincial trial protocol require sample storage facilities and we had built up biorepository experience in the operational aspects thereof. The H3 Africa Biorepository initiative will allow CLS to be a more visible Biorepository Partner within a greater intitive of studying health and disease in the African population.

Garth Swartz
Clinical laboratory services
I am the repository manager for clinical laboratory services. My interest is in the biorepository data group.

Shiksha Reddy
Clinical Lab Services
Shiksha Reddy is the Chief Operations Officer at CLS, a division of Wits Health Consortium. She is qualified as a medical scientist, and has 11 years of experience in a clinical pathology environment and for the last 8 years has managed the operational aspects of a commercial pathology laboratory. She has an MBA from the Open University: Harvard and provides operational support and strategic leadership to the CLS management team including advice, guidance, and direction on operational policies and procedures, business initiatives, and management of existing relationships.

Mukthar Kader
Clinical Laboratory Services
I am a qualified and registered Medical technologist (Clinical Pathology) in South Africa.
My Experience includes being the Laboratory Manager of a Clinical Pathology and a PCR laboratory. I have 16 years of clinical pathology laboratory work experience and have been involved in the accreditation process (SANAS) of several laboratories.

I am trained at GCLP compliance, IATA certification and completed the Protection of Human participants course. At CLS I hold the position of Project manager and my responsibilities involved overseeing all aspects of the Gates funded PrEP Study for Laboratory sites in Kenya and Uganda for the past 3 years. Part of the PrEP study was to ensure that the different sites in Kenya and Uganda were study compliant with regard to maintenance of the freezer works computer system, ensuring participant samples were traceable, easily retrieved, stored and shipped to the coordinating centre in accordance to study requirements. Duties also included site assessment to GCLP standards and study specific requirements, training of staff and quality assurance EQA and IQC monitoring and training. Currently I am part of the H3Africa team within Clinical Laboratory Services (CLS) and involved in the CLS BR operation.

Integrated Biorepository of H3Africa Uganda – IBRH3AU
PI: Moses Joloba
Institute: Makarere University College of Health Sciences, Uganda
Over the last ninety years, Makerere University College of Health Sciences (MakCHS) Kampala, Uganda formerly Faculty of Medicine, has established itself as a reputable center for research on infectious and non-infectious diseases. On the basis of the existing research and service provision-

infrastructure at MakCHS, as well as a multidisciplinary team aiming to establish and maintain leadership in genetics/genomics and environmental related research, we are proposing a two phase approach to build capacity for a biorepository at MakCHS to strategically support the overall goals of the H3Africa projects. That is to bridge the existing gap in the knowledge and practice on genetics/genomics and environmental determinants of health. The overall goal of the Phase I (UH2) is to develop capacity and conduct a feasibility study for MakCHS to host a biorepository for the H3Africa, while that for Phase II (UH3) is to scale-up (building on the plan and feasibility data generated during Phase I) and develop a fully functional biorepository for H3Africa investigators, other researchers in Africa and internationally. We propose an integrated biorepository that will bank a broad range of samples that are available in our set up. The resultant Integrated Biorepository of H3Africa Uganda ($IBRH_3AU$) will support the activities of the H3Africa awardees, whose objective is to enhance the capability of African scientists and research institutions to use genomics and other powerful novel approaches to address problems of African health and disease. The specific aims contingents to the above goals are:

Phase I

1) Aim1: Capacity development required for a full-scale biorepository – the $IBRH_3AU$. 1(a) Develop a plan for the organization structure of the IBRHAU. 1(b) Train $IBRH_3AU$ personnel in biorepository management and science. This training will equip them with the necessary skills and knowledge to manage a project of this magnitude and innovation in Uganda and within Africa. 1(c) Develop plans for improving the existing infrastructural for the biorepository. 1(d) Develop protocols for sample receipt, processing, storage and distribution, with rigorous quality assurance and control procedures as established by international standards.
2) Aim 2: Conduct a feasibility study to demonstrate the ability of the $IBRH_3AU$ to host and maintain a large-scale biorepository that will meet the needs of the H3Africa Consortium, and the greater community of African and International scientists who will use human biomaterials for research.

Phase II

1) Aim 1: Improve the existing infrastructure to establish the $IBRH_3AU$ that will function as a local, regional and international biorepository. The $IBRH_3AU$ will be a highly annotated and curated world class biorepository serving the local, regional and international scientific community to be able to conduct a wide array of scientific investigation.
2) Aim 2: Build more capacity in biorepository science and management through training.
3) Aim 3: Perform centralized and standardize biorepository operations. The processing methodologies will ensure fit-for-purpose operations such as high throughput high-quality automation procedures, and robust data trail. The methods will avoid approaches that inherently preclude some future analyses. Maintain a detailed, quality assured sufficient and secure data audit trail through the use of a Laboratory Data Management System (LDMS).

The overall specific deliverable of this proposal is a structure that is capable of collecting 100,000 specimens per year in the $IBRH_3AU$ and biospecimen distribution to H3Africa Consortium researchers. The achievement of the specific aims of this proposal will focus on the local and international efforts to provide simplified access to high quality specimens that can be used to generate genomic and non-genomic data serving the H3Africa, African and international research/scientific community.

Moses Joloba
Makerere University College of Health Sciences

Dr. Joloba has developed skills in conducting clinical microbiology, laboratory-based research and training as well as building laboratory capacity in Uganda and other countries. Initially he graduated as a physician at Makerere Medical School in 1994 and later as a clinical microbiologist at Case Western Reserve University (graduated in May 1996, MS Degree) after which he returned to Uganda and took a faculty position at Makerere university Department of Medical Microbiology. He also helped to establish a TB laboratory for the Tuberculosis Research Units (TBRU) and became its technical director and later a scientific director site (1996 – 1999). During that time a number of laboratory based studies were conducted and published many with Dr. Joloba as the first author.

These involved research on Early Bactericidal Activity (EBA), quantitative bacillary response to therapy and evaluation of new markers to therapy. In addition, the TB laboratory was successfully involved in collection, processing, storage and shipment of samples for the World Health Organization Biorepository for TB diagnostic samples in 1998 -1999. The samples handled included sputum, saliva, serum and urine. In 2000 – 2003, Dr. Joloba undertook a Ph.D course at Case Western in Microbiology and Molecular Biology. After his return Dr. Joloba, with support of NIH and Gates Foundation, he established a Molecular Biology, immunology and mycobacteriology laboratories at the University. He became the Director of the National TB Reference laboratory now a supranational laboratory. All the laboratories have successfully established capacity to receive, process, store and ship various samples. Over 50,000 samples are either received or shipped from these laboratories annually. Dr. Joloba is a TB laboratory consultant for World Health Organization. He is a monitor for clinical laboratories and a trainer of Good Clinical Laboratory Practice. Using his previous experience in biobanks, he has since been a key monitor in selection of sites and handling of samples for a successful WHO TB biobank in Kenya and South Africa in 2005. Dr. Joloba has won over 8 grants and published over 70 peer reviewed papers, he is a reviewer for 6 journals and been an author of a book chapter. Dr. Joloba has developed skills in setting up standard laboratories, running and monitoring biobanks, initiating and participating in successful North to South and South to South collaborations, conducting laboratory-based research as well as training of graduate students. Dr. Joloba has trained 33 Masters and 14 Doctoral students. Dr. Joloba is the Program Coordinator of the World Bank East African Public Health Laboratory Network – EAPHLN – as is in charge of Laboratory Accreditation and Networking in East Africa. This project is worth US$100 million.Research Interest: Dr. Joloba's Research interests are in infectious disease mainly tuberculosis His focus is mainly on pathogenesis, diagnostics and strain characterizations. He uses both phenotypic and genotypic approaches.

Misaki Wayengera
Makerere University College of Health Sciences
I am a medical doctor (MBChB, 2004) with postgraduate (MSc) training in immunology and clinical microbiology. I hold tenure as an assistant lecturer of genetics/genomics & immunology at Makerere University, Kampala, Uganda in the Depts. of pathology and medical microbiology. My research interests center on the immunology of HIV and other viral vaccines, as well as emerging genetic/genomic approaches for the cure of all persistent viral infections. I am a member of the steering committee of the young and early careers investigators (YECI) of the Global HIV Vaccine Enterprise. I am also non-executive member of the IAS "Towards an HIV Cure" group. I have worked on the R-M system model for an HIV cure since 1999, and the proposed zinc finger nuclease (ZFN) gene-therapy for HIV is a direct result of these efforts. I am active member of the African Society for Human Genetics/Genomics, and the International Academy of Pathology (IAP). I am actively involved in Clinical Service at the Mulago National Referral hospital where spear-head the making of diagnoses for children born with genetic abnormalities.

Eddie Wampande Mujjwiga
Makerere University
I am a Ugandan by citizen born in 1971, holds a bachelors degree in Veterinary Medicine, Ms in Molecular Biology and now pursuing a PhD in Molecular Epidemiology of TB. I will be pursuing my Post Doc in Human genetics.

Samuel Kyobe
Makerere University College of Health Sciences
The aim of the proposed research is to identify host genetic factors associated with non-progression of disease after HIV infection in a coohort of HIV-infected African children. It also seeks to identify genetic factors associated with the progression of TB infection. I am an NIH-funded MEPI-MESAU scholar of Medical Microbiology at Makerere University College of Health Sciences (5R24TW008886 OGAC, NIH and HRSA, Sewankambo N PI) since August 2011 under the mentorship of Prof. Moses Joloba. I completed a Bachelor of Medicine & Bachelor of Surgery in 2009 after which I worked as a Resident in Rubaga Hospital for a year. Then I joined the

Department of Microbiology as a junior staff, I was elected to manage the molecular biology laboratory tasked with the production of quality basic science research as cloning, gene knock out and site directed mutagenesis. I possess skills in performing molecular epidemiological studies for tuberculosis using techniques such as MIRU-VNTR, RFLP, and SNP genotyping. Have competencies in using molecular methodologies for pathogen detection and identification as well as genotypic drug resistance detection and treatment monitoring. I have recently trained in Pyrosequencing a low through-put method of sequencing. I will perform the role of Project coordination for the Makerere University site on this project. I am expected to participate in whole exome sequencing and analysis. With additional training in these technique I will be in position to direct the project on attaining its goals. Eventually, I will gained the knowledge, expertise, confidence and leadership required for future research in genomics/genetics studies which is much needed in Africa. My research and leadership experience has adequately prepared and equipped me with the minimum skills I need to manage and co-ordinate project site specific activities.

NIH H3Africa Ethical, Legal and Social Implications (ELSI) Project

Exploring Perspectives on Genomics and Sickle Cell Public Health Interventions

PI: Ambroise Wonkam

Institute: University of Cape Town, South America

The H3Africa initiative as a whole draws increased attention to a number of longstanding and emerging issues in genetic and genomic research, such as informed consent, community engagement, privacy and confidentiality, (mis)use of genetic information, governance of biorepositories, reciprocity/benefit sharing, and ownership (http://h3africa.org/h3africa_whitepaper.pdf). Sickle cell disease (SCD) research encounters most, if not all, of these issues, as well as others (e.g. genomic data sharing, return of research results and incidental findings, and obligation to family members) highlighted by genome-wide association studies (GWAS) and whole exome/genome sequencing that have been carried out in some setting like Tanzania. Research on SCD also will also grapple with issues related to the inclusion of children in genomic research. There is a dearth of published research on the views of Africans and African communities concerning participation in genomic research, and virtually no information on their perspectives regarding several of these issues.

As technological advances, well-established in Ghana and not yet available in Cameroon and Tanzania, contribute to the global expansion of newborn screening, the values and practices of health professionals and community members must be considered in order to ensure culturally appropriate and cost effective implementation of strategies to improve awareness, early detection, and prevention of complications. The concept of early detection and prevention are also applicable to prenatal diagnosis (PND), in which case disease is detected even earlier than newborn stage, allowing for the option of preventing disease through termination of pregnancy. While PND accompanied by termination raises an array of ethical, legal and psychosocial issues, previous research has shown a high level of interest in and uptake of these procedures among African parents with an SCD-affected child. We propose to build on our preliminary work on newborn screening in Ghana and work related to attitudes and practices of SCD PND in Cameroon as well as genomics work performed in Tanzania, to gather information about the complex cultural, religious, economic, educational and political dimensions of public health interventions for SCD.

Empirical data from a broad spectrum of stakeholders are essential to the development of effective policies and programs. A major objective of this project is to advance our understanding of the perspectives of research scientist, health professionals, SCD patients and community populations within our collaborative network concerning both genomic research and the public health aspects of SCD.

We will employ qualitative research methods to pursue the following specific aims:

Aim 1: Explore perspectives and attitudes regarding genomic research and its implementation and implications in Cameroon, Ghana, and Tanzania.
Aim 2: Assess perceptions about public health interventions to increase awareness, early detection, and prevention of SCD-related complications.

This research is the first phase of a series of longitudinal, mixed method studies exploring individual, family, community, and professional perspectives on genomic research and SCD-related public health interventions. This formative research will help us to define the most effective strategies for: 1) implementing genomic research and addressing the pertinent issues and 2) insuring informed decision-making about and optimal uptake of newborn screening, other public health interventions for SCD, and the related services.

Ambroise Wonkam
University of Cape Town
Prof Ambroise Wonkam is a medical geneticist, associate professor/senior specialist in the Division of Human Genetics, Faculty of Health Sciences, and University of Cape Town, South Africa.After a MD from the Faculty of Medicine and Biomedical Sciences, University of Yaoundé I (Cameroon), Prof Wonkam complete a thesis in Cell Biology in the department of Morphology, University of Geneva (Switzerland), followed by a specialization in medical geneticist.His interests are reflected in his more than 40 peer-reviewed publications, which are in laboratory, clinical, ethical and educational aspects of medical genetics. His major ongoing research projects are on monogenic conditions in African populations; specifically his laboratory is exploring the phenotypic correlation of sickle cell anemia to genotypic variations, and genetic of congenital hearing loss in Africans. Hi is member of the H3Africa Consortium and PI of an H3Africa NIH ELSI proposal.His has recently established the medical genetics practice in Cameroon. Prof Wonkam is board member of the African Society of Human Genetics.

Sheryl McCurdy
University of Texas Houston Health Science Center
Sheryl A. McCurdy, PhD is Associate Professor at the School of Public Health, University of Texas-Houston Health Science Center. She has conducted National Institute on Drug Abuse and Social Science Research Council/American Council of Learned Societies funded research as well as CDC HIV Prevention programming for heroin users in Tanzania. Her publications are in public health, medicine, and African Studies journals and books. She is co-editor and contributor to _'Wicked' Women and the Reconfiguration of Gender in Africa_. During the last decade her work focused on HIV prevention and the ethical, legal, and social implications (ELSI) of research on the human microbiome. She is a co-investigator on a newly funded H3Africa project examining ELSI issues related to the genomics of sickle cell disease in Cameroon, Ghana, and Tanzania.

NIH H3Africa Bioinformatics Network Project

H3ABioNet: a sustainable African Bioinformatics Network for H3Africa

PI: Nicola Mulder
Institution: University of Cape Town, South Africa

H3ABioNet aims to create a sustainable African Bioinformatics Network to support H3Africa researchers through the development of bioinformatics capacity on the continent. Specifically, it aims to: 1) engage with the H3Africa Consortium, providing a framework for integration and communication amongst its members to ensure progress towards the common goal of full exploitation of our genomic and environmental resources for translation into improved health in Africa; 2) develop a core bioinformatics infrastructure (hardware and human resources) to aid research in genomic medicine, high throughput biology, systems biology, genetics, and medicine, for the study of human heredity and health; S) develop tools and bioinformatics solutions appropriate for exploitation and interpretation of biological information in Africa, and make these

more accessible to H3Africa researchers; 4) develop research partnerships and promote interaction between bioinformaticians, clinicians, molecular geneticists and ethics researchers to ensure integrative research into health issues in Africa; 5) develop the bioinformatics capacity of people in Africa to empower them to perform cutting edge research, and to ensure retention of bioinformatics skills on the continent; 6) facilitate secure, high-fidelity storage and management of data generated within the H3Africa framework and their deposition in public databases, so that maximum value can be derived from these data; and 7) ensure that the increasing amount of information from genomic analyses and high-throughput molecular biology is accessible to all H3Africa researchers, to promote health research, scientific progress and global competitiveness in Africa. These aims will be achieved through four major activities: user support, training and capacity development, research and tools, and outreach and communication. These activities will result in a bioinformatics support structure, continuous specialized training for bioinformaticians and researchers, development and accessibility of new tools appropriate for the African setting, and a framework for communication flow within the H3Africa Consortium. Through the pooling of existing expertise and training of the next generation of researchers, we will build critical mass in bioinformatics for H3Africa.

Nicola Mulder
University of Cape Town
Associate Prof Mulder heads the Computational Biology Group at the University of Cape Town (UCT) (http://www.cbio.uct.ac.za). After her PhD in Medical Microbiology, she spent 8 years at the European Bioinformatics Institute (EBI) in Cambridge, moving into the area of bioinformatics. At the EBI she was a Team Leader, responsible for development of one of the most heavily used Bioinformatics resources at the Institute. At UCT A/Prof Mulder works in the area of bioinformatics of infectious diseases, including pathogen and host genomics and biological networks, human variation studies and disease associations. The group also provides bioinformatics support and training for postgraduate students and local researchers. A/Prof Mulder is President of the African Society for Bioinformatics and Computational Biology and is coordinating the H3Africa Bioinformatics Network.

Alia Benkahla
Institute Pasteur de Tunis
Dr Alia Benkahla is a bioinformaticist she did her PhD studies at the IGS-CNRS in Marseille with Jean-Michel Claverie as PhD supervisor, did her Post-Doc at the Max-Planck Institute in Berlin, and moved to LIVGM at Institut Pasteur de Tunis in January 2005. She initiated the Group of Bioinformatics and Mathematical Modeling and invested her initial period at IPT in capacity building in the field of Bioinformatics by: training students; starting a research group; co-organizing international events in Africa; and looking for funds in Bioinformatics for pathogen and disease vectors. She was the chair of the third ISCB Africa ASBCB Conference on Bioinformatics 2013. She was the PI of 5 research projects. Her research is mainly in the area of systems biology aimed at understanding processes of regulation, interaction, signaling mechanisms activated in response to infections (i.e., leishmaniasis and/or tuberculosis).She is the principle investigator of 2 research grants, co-author of 13 publications among which 3 Nature and co-author of 1 chapter.

Scott Hazelhurst
University of the Witwatersrand, Johannesburg
Scott Hazelhurst is Acting Director of Wits Bioinformatics and an associate professor in the School of Electrical & information Engineering at the University of the Witwatersrand, Johannesburg. He received his BScHons and MSc degrees from Wits and his PhD from the University of British Columbia. His research interests are high performance computing and bioinformatics.

Chisomo Msefula
College of Medicine, University of Malawi
My name is Chisomo Msefula from Malawi. I am a Lecturer in Microbiology at the College of Medicine, University of Malawi and a Research Scientist based at the Malawi-Liverpool-Wellcome Trust in Blantyre. I graduated with a PhD from the University of Liverpool in 2009, working on the

antibiotic resistance and genetic diversity of Multidrug resistant Salmonella. I have continued with pathogen genomic studies branching to E.coli and Klebsiella sp genomes. I am now working on establishing genetic host-susceptibility studies to infection under H3 Africa initiative.

Seydou Doumbia
University of Bamako, USTTB
Dr. Doumbia earned his MD degree from University of Bamako, Mali and a Ph.D in epidemiology of infectious diseases in 2002 from Tulane University in New Orleans, USA. Dr. Doumbia is currently the Chair of the Department of public Health of the Faculty of Medicine of University of Bamako (USTTB) and Deputy Scientific Director at the Malaria Research and Training Center of NIH-International Center of Excellence in Research (ICER-Mali). He has long standing experience in epidemiology and public health research in infectious disease for more than 15 years. His research areas cover infectious disease, particularly malaria, leishmaniasis and HIV. He works closely with basic scientists and public health practitioners to felicitate the test of scientific innovation and cutting edge technology in the field. Dr. Doumbia has been involved in several capacity building initiatives in Africa and contributing to the establishment of an MPH training program in Mali supported by NIH-Fogarty International and a Center for training in Bioinformatics and Functional Genomic of disease vectors supported by NIH and WHO. He is Program Director or Co-PI several NIH extramural and intramural (DIR) projects on malaria and leishmaniasis. He is Program Director of NIH funded Tropical Disease Research Center (TMRC) on neglected tropical diseases and Project Leader of International Center of Excellence in Malaria Research (ICEMR)- West Africa a consortium also funded by NIH. He is Co-PI of H3ABioNet (African Bioinformatics Network). Dr. Doumbia collaborates with several African and international research institutions and Universities including but not limited to Tulane University, Harvard School of Public Health, Columbia University, UCAD, University of Ghana, IP of Tunis, UCT, NIAID (LPD and LMVR), ISPED of Bordeaux and WHO/TDR.

Oyekanmi Nashiru
National Biotechnology Development Agency, Abuja. Nigeria
Oyekanmi Nash, PhD - Director, Molecular Biotechnology and Bioinformatics Department, National Biotechnology Development Agency, Federal Ministry of Science and Technology (NABDA/FMST), Abuja, Nigeria. He coordinates the NIH H3ABioNet Project at NABDA/FMST, Abuja. An Associate Professor with the Nigerian Defense Academy, he coordinates the Key Laboratory for Translational Research on Emerging Infectious Diseases at the NABDA-Southwest Biotechnology Center, University of Ibadan, Ibadan. He was a Postdoctoral Fellow with the Canadian Protein Engineering Network Center of Excellence, University of British Columbia, Vancouver, Canada, and a Research Associate with the Department of Microbiology and Immunology, Albert Einstein College of Medicine, New York. He is the African Regional Director for the International Consortium in Anti-Virals (ICAV)

Dean Everett
Malawi-Liverpool-Wellcome Trust Clinical Research Programme, Malawi/ Institute of Infection and Global Health, university of Liverpool, UK
Senior lecturer in Molecular Microbiology, with a focus on pathogen genomics, particularly respiratory pathogens. A molecular microbiologist based in Africa for the last 15 years, with an interest in pathogen and host genomics.

Judit Kumuthini
Centre for Proteomics and Genomics Research
Judit received her BSc in Biomedical Science and MSc in Bioinformatics in the UK. She completed her PhD from University of Cranfield, UK in Bioinformatics in genetic network (GN) extraction using Bayesian belief framework. During her PhD Judit established novel processes to extract genome-wide Genetic Network extraction for E. coli, yeast and humans, from microarray data. Judit later joined drug discovery group at GSK (Glasgow Smith Kline, UK) as a drug target bioinformatician. She is currently the Bioinformatics Manager at CPGR (Centre for Proteomic and Genomic

Research) after completing her fellowship at UCT (University of Cape Town) and is leading her team to provide expertise in various fields in bioinformatics. This includes providing service, support and R&D through collaboration to life scientists in "omics" field, addressing a wide range of biological questions from genomics to system biology. Judit is the associated node manager for EMBNET, where she was elected as the PRPC (public relation and publicity) committee member. Judit is committed to human capital development in the bioinformatics arena to enhance the knowledge base in Africa. She is also co-PI, node manager and the chair of bioinformatics user support working group for H3ABioNet project, part of H3A initiative (http://h3africa.org). At the CPGR, Judit started and manages the mini internship programme (MIP) and Knowledge transfer programme (KTP) aimed at developing specific skill sets required for next generation of in silico biologists. She has trained and supervised many postgraduate students in Africa and Europe. Her broad research interests include African Genetic rare diseases, process optimization, algorithm development, systems development, information management and visualization and e-learning.

James A.M. Brandful
Noguchi Memorial Institute for Medical Research (NMIMR), University of Ghana.
Dr. James AM Brandful is immediate past Head of the Department of Virology of Noguchi Memorial Institute for Medical Research (NMIMR), University of Ghana, one of Ghana's prestigious biomedical research institutes. Dr. Brandful is a molecular virologist and holds a BSc Biology, MSc in Molecular Biology, PhD in Medical Microbiology His work in the late 80s focused on enteroviruses responsible for acute haemorrhagic conjunctivitis (AHC). He isolated and molecularly characterized the agent of a massive 8-month epidemic of AHC. For the first time in Ghana, a coxsackie virus was identified. He has also worked extensively on the molecular epidemiology and characterization of HIV strains in Ghana, the monitoring of drug resistance in persons on treatment or naïve for antiretrovirals (ARVs) and implications for antiretroviral therapy. Aspects of his work also looked at human papilloma virus (HPV) and association with cervical cancer in Ghanaian women, some serum biomarkers of HIV/AIDS progression and has been involved with studies on Hepatitis viruses in Ghana. He has interest in viral haemorrhagic and emerging diseases, active surveillance of such diseases and the development of antivirals against these pathogens. He has also been a strong advocate for researching herbal-based products/medicinal plants for the management of HIV/AIDS. Currently he collaborates with Japanese scientists on investigation of some Ghanaian medicinal plants for anti-HIV properties. He has great interest in Bioinformatics and a PI for the NMIMR Node of the H3ABioNet Consortium on developing bioinformaticians to support H3Africa/Wellcome Trust Projects

Ozlem Tastan Bishop
Rhodes University
Ozlem received her BSc degree in Physics from Bogazici University, Istanbul, Turkey. Then she moved to the Department of Molecular Biology and Genetics at the same University for her MSc degree. She obtained her PhD from Max-Planck Institute for Molecular Genetics and Free University, Berlin, Germany in 2003. She was co-founder and the first President of the student association of Max-Planck Institute in Berlin. While doing her PhD, Ozlem became interested in structural biology, and during her postdoctoral positions (Texas University, UWC and UP) she gained experience in structural bioinformatics as well as structural biology. In October 2009, Ozlem took up a senior lecturer position at Rhodes University, with the responsibility to develop postgraduate studies in bioinformatics at the University. She established Rhodes University Bioinformatics research group (RUBi) in 2010, and started one-year MSc programme in bioinformatics by coursework and research thesis in 2011. Ozlem is the president of the South African Society for Bioinformatics (SASBi) since September 2012. Ozlem's broad research interest is comparative genomics and structural bioinformatics.

Dan Masiga
International Centre of Insect Physiology and Ecology (ICIPE)
Dan Masiga is a Research Scientist and Head of the Molecular Biology and Bioinformatics Unit (MBBU) at ICIPE. He leads a research group with interests in the ecology of infectious diseases,

including interactions between vectors and pathogens. Within the context of H3ABioNET, together with Anne Fischer, he is providing a platform for training in bioinformatics and supporting other initiatives, such as the Glossina Genome Initiative and activities of the ISCB-ASBCB Regional Students Group (RSG). The group at icipe is composed of Students, Postdoctoral and Research Scientists, with an active collaboration within icipe and with other groups globally.

Ellis Owusu-Dabo
Kwame Nkrumah University of Science and Technology. Kumasi Centre for Collaborative Research in Tropical Medicine
Dr. E. Owusu-Dabo earned his medical degree at the KNUST and his specialist professional degree at Ibadan. He holds additional qualification (PhD) from the University of Nottingham. His areas of expertise include: epidemiology of noncommunicable disease, population genetics of pulmonary tuberculosis and Health systems research.He is currently the Scientific Director for the Kumasi Centre for collaborative Research in Tropical medicine. He is a Fellow of Ghana College of Physicians, member of West African College of Physicians and Principal Investigator of many research grants including H3ABionet for bioinformatics. He has served on several review committees, and a member of West African Health Research Network's scientific committee since 2010.

Victor Jongeneel
University of Illinois
Victor Jongeneel heads the High-Performance Biological Computing Group at the University of Illinois. He previously served as the Founding Director of the Swiss Institute of Bioinformatics, the Director of the Vital-IT HPC facility and the Vice-President for Research of the Cyprus Institute. His training is in microbiology and molecular genetics.

Faisal M. Fadlelmola
Centre for Bioinformatics, Future University of Sudan
Faisal M. Fadlelmola, PhD, Fellow of the British Computer Society (FBCS), Chartered Information Technology Professional (CITP, UK), and the Scientific Director for the Centre for Bioinformatics, Future University of Sudan. Dr. Fadlelmola is an Associate Professor at the Faculty of Science, Future University of Sudan. He coordinates the NIH H3ABioNet Project at Sudan Bioinformatics Node. He holds a PhD in Molecular Genetics and Bioinformatics from the University of Wuerzburg, Germany, awarded in 2003 and two masters of Science from UK in Medical Molecular Biology (1999) and Information Systems (2000). Prior to joining the Future University, Dr. Fadlelmola was a Postdoctoral Research Fellow at the Centre for Translational and Applied Genomics (CTAG) at the British Columbia Cancer Agency (BCCA), Vancouver, Canada from 2004-2006. He also worked as Research Associate at the CTAG at the BCCA from 2007-2008. He has been involved with a variety of research projects on such topics as cancer genomics, bioinformatics, molecular epidemiology of cancer, translational and applied genomics, computer sciences and IT, mobile health and mobile learning, among many others.

Sylvester Lyantagaye
University of Dar es salaam
Holds a PhD in Biochemistry and underwent 2 years postdoctoral training in bioinformatics at the South African National Bioinformatics Network (NBN). Research interests are on Screening natural products and mineral complexes for the presence of bioactive compounds of medicinal importance, and on Identification of Unique Genes of Cancer, TB, HIV and Malaria Parasite and vector as potential new drug targets, by Comparative Genomics using Bioinformatics tools. One of the current projects is developing a malaria database called UDSM Malaria information system (MIS). The system aims to integrate malaria vector, parasite and host (human) related –omics data and ontologies from public sources to enable large-scale analysis of the data. MIS user can examine relationships of the Plasmodium data with that of human, mosquito and other species whose data have been loaded into the system. MIS will allow for easy integration of a new type of experimental data into the MIS as required. This integrative model will give more insight to scientists researching

on anti-malaria vaccine and/ or drugs to better understand the relationships between the genotypes and phenotypes related to the disease. Also involved in regional project as a: Coordinator of the Southern Africa biochemistry and informatics of natural products (SABINA) project funded by Carnegie-IAS –RISE, Co-PI of the Industrial enzymes for sustainable bioeconomy (Bio-Innovate project 8) fundd by Sida) and Co-PI - H3ABioNet-a sustainable African Bioinformatics Network for H3Africa (NIH-USA). A regular reviewer of the Tanzania Journal of Science (TJS) since 2008 and African Journals of Microbiology Research since 2009

Appendix 4 – Independent Expert Committee

Name	Appointment	Institution
Barry Bloom	NIH appointed, Co-Chair	Harvard University
Kay Davies	WT appointed, Co-Chair	University of Oxford
Philip Awadalla	WT appointed	University of Montreal
Carlos Bustamante	NIH appointed	Standford University
Ruth Chadwick	WT appointed	Cardiff University
Rex Chisholm	NIH appointed	Northewestern University
Fred Nakwagala	NIH appointed	Makerere University, Mulago Hospital
Solomon Nwaka	WT appointed	World Health Organization
Ayoade Oduola	NIH appointed	Institute of Infectious Diseases of Poverty
Charmaine Royal	NIH appointed	Duke University
Val Sheffield	NIH appointed	University of Iowa

Appendix 5 – NIH and Wellcome Trust staff

National Institutes of Health

Name	Position
Dr. Francis Collins	Director
Dr. Eric Green	Director National Human Genome Research Institute
Dr. Mark Guyer	Deputy Director National Human Genome Research Institute
Dr. Jane Peterson	Senior Advisor to the Office of the Director National Human Genome Research Institute National Institutes of Health
Dr. Charles Rotimi	Director, Center for Research on Genomics and Global Health National Human Genome Research Institute National Institutes of Health

Wellcome Trust

Name	Position
Dr Jimmy Withworth	Head of International Activities
Dr. Audrey Duncanson	Science Portfolio Manager, Molecular and Physiological Sciences, Science Funding
Katherine Littler	Senior Policy Advisor
Lara Bethke	Science Portfolio Advisor

Appendix 6 – Speakers and Observers

Speaker	Affiliation
Carlos Bustamante *PhD*	Stanford University
Manjinder Sandhu *PhD*	University of Cambridge
Dominic Kwiatkowski *PhD*	St Johns College
Mattias Jakobsson *PhD*	Uppsala University
Himla Soodyall *PhD*	University of the Witwatersrand