



H3Africa

Human Heredity and Health in Africa



The Human Heredity and Health in Africa Initiative

2nd H3Africa Consortium Meeting

16-18 May 2013, Accra, Ghana

Audrey Duncanson

Wellcome Trust

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TrypanoGEN – genetic determinants of susceptibility to trypanosomiasis

Enock Matovu
College of Veterinary Medicine
Makerere University, Uganda



- Issa Sidibe, CIRDES, **Burkina Faso**
- Dieudonne Mumba, INRB, **Democratic Republic of Congo**
- Mathurin Koffi, University of Abob-Adjame, **Côte d' Ivoire**
- Gustave Simo, University of Dschang, **Cameroon**
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- Bruno Bucheton, IRD Marseille, **France**
- Neil Hall/Christiane Hertz-Fowler, University of Liverpool, **UK**
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Co-applicants

Collaborators

RHDGen Network – Genetics of rheumatic heart disease

Bongani Mayosi
 Department of Medicine
 University of Cape Town



- Raj Ramesar, University of Cape Town, **South Africa**
- Nicola Mulder, University of Cape Town, **South Africa**
- Andrew Whitelaw, New Groote Schuur Hospital, **South Africa**
- Stephen Ogendo, University of Nairobi, **Kenya**
- Ana Mocumbi, Instituto Nacional de Saude, **Mozambique**
- Christopher Hugo-Hamman, CBMH, **South Africa**
- Okechukwu Ogah, University College Hospital, **Nigeria**
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- Charles Mondo, Uganda Heart Institute, **Uganda**
- John Musuku, University of Zambia, **Zambia**
- Guillaume Pare, McMasters University, **Canada**
- Bernard Keavney/Heather Cordell, Newcastle University, **UK**
- James Dale, University of Tennessee, **USA**
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Burden, spectrum and aetiology of type 2 diabetes in sub-Saharan Africa

Albert Amoah

University of Ghana, Ghana



- Clement Adebamowo, Institute of Human Virology, **Nigeria**
- Pontiano Kaleebu, Uganda Virus Research Institute, **Uganda**
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- Charles Rotimi, NHGRI, **USA**
- Manjinder Sandhu, Wellcome Trust Sanger Institute, **UK**
- Eugene Sobngwi, University of Yaoundé, **Cameroon**
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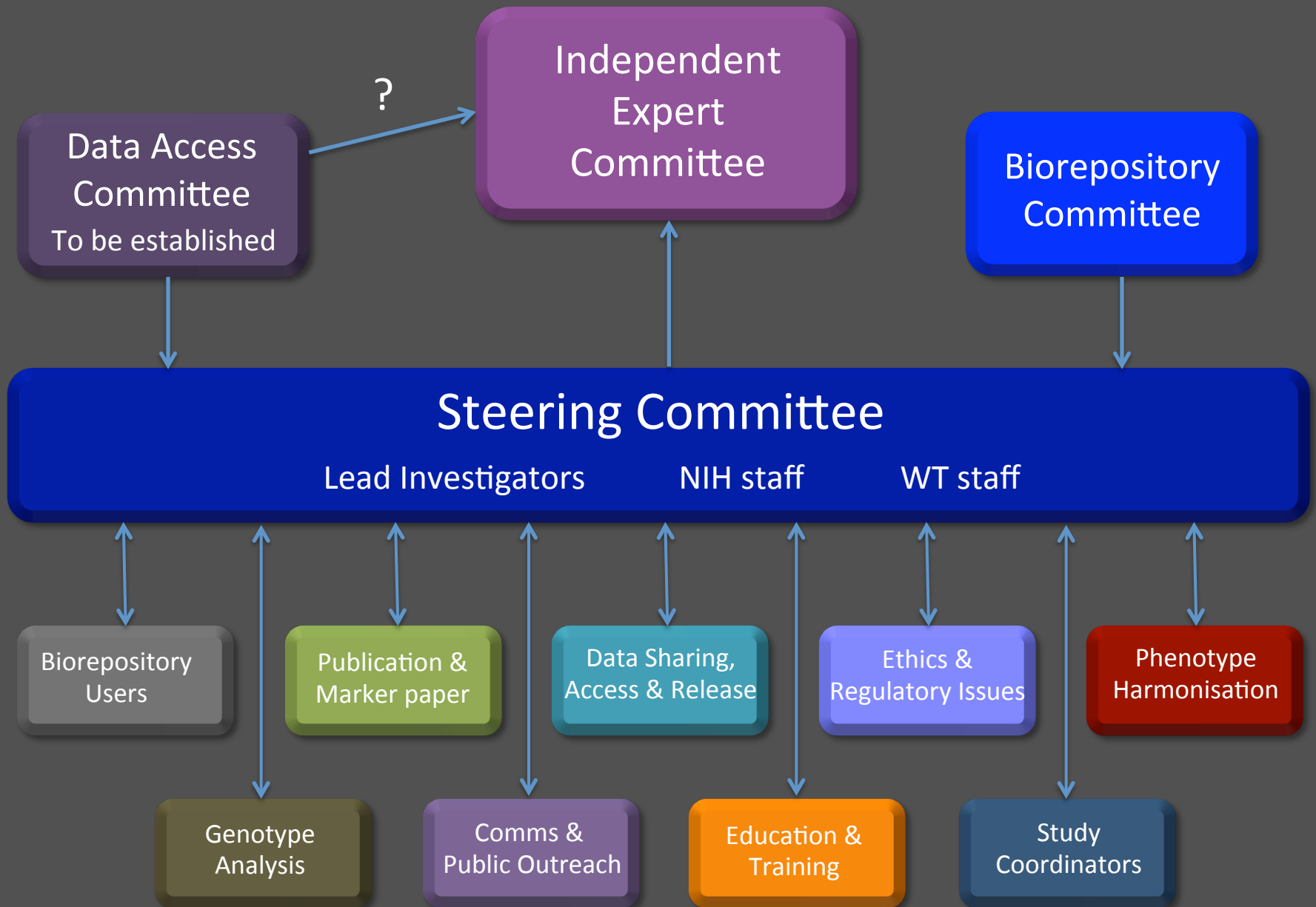
H3Africa as a Consortium....

Consortium approach

- Individual projects funded under H3Africa would come under the H3Africa ‘umbrella’
- Funders envisioned a Consortium approach, such HapMap, WTCCC and the 1000 Genomes Project
- Maximise the benefit and impact of the H3Africa Consortium

Could be thought of as a ‘Participatory Democracy’

‘Aims to create opportunities for all members of a Consortium [population] to make meaningful contributions to decision-making and to obtain as broad a range of views as possible’



Challenges

- Participants need to buy-in to the structure and overarching goals
- Can be burdensome for participants, eg Working Groups, Steering Committee calls
- Individual preferences or ways of working may need to adapt and change
- Consortia need to be flexible and change over time
- Communication essential – internally and externally

Benefits

- Shared resources
- Exchange of ideas
- Centralised resources, *eg* H3ABioNet
- Standardized operating procedures, methods and analysis schemes
- Access to training, bioinformatics and analysis pipelines

Benefits cont' d....

- Added value from cross-project and cross-consortium collaborations
- Shared risk

What do we need from you?

- Participation!
 - Working Groups
 - Steering Committee

Consortium Coordinating Centre

- Provide support for trans-H3Africa Consortium activities
-
- Will employ an administrator and coordinator
-
- Will provide support for the Working Groups and H3Africa Steering Committee
-
- Reports to the Steering Committee

Samples deposition

- Deposition of biological samples into NIH-funded biorepositories is not a condition of WT award
- Concern there would be challenges in using biorepositories, such as moving samples across borders,
- It is the expectation that Trust-funded samples will be deposited, if no barriers exist

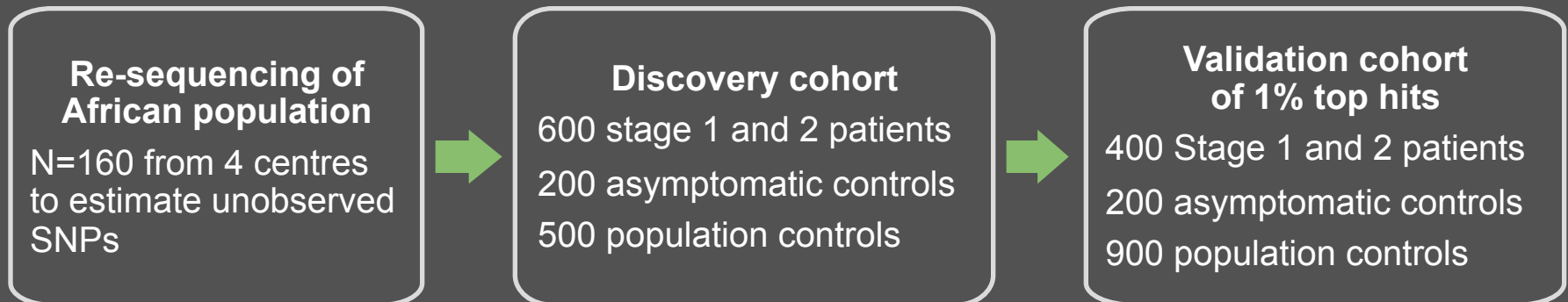
Questions?

“TrypanoGEN – genetic determinants of susceptibility to trypanosomiasis”



Enock Matovu
College of Veterinary Medicine
Makerere University, Uganda

- *Vision:*
 - Develop an integrated study of human, parasite and vector genetic variation in trypanosomiasis
- *Initial high level objectives:*
 - Create a biobank of retrospective and prospective samples
 - Generate a database of human genetic variation across Africa



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“RHDGen Network – Genetics of rheumatic heart disease”



Bongani Mayosi
Department of Medicine
University of Cape Town

- *High level aim:*
 - Broaden understanding of RHD pathogenesis in sub-Saharan Africa by identifying resistance and susceptibility genes

Case-control study

1500 cases
1500 controls
...to discover SNPs associated with RHD



Family-based GWAS

1000 independent cases
2000 parents
...to confirm or refute significant hits



Combined analysis

2500 cases
2500 controls/pseudo-controls
...to identify associations of smaller effect size

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“Burden, spectrum and aetiology of type 2 diabetes in sub-Saharan Africa”



Albert Amoah
University of Ghana, Ghana

- *High level aim:*
 - Assess burden and aetiology of type 2 diabetes and its complications in adult populations across sub-Saharan Africa
- *Initial development of epidemiological and genomic resource:*
 - Recruitment of up to 1000 cases from each of 12 centres (diabetes clinics, hospitals, referral centres, tertiary clinics)
 - Population-based cross-sectional survey
- *Possible genomics projects:*
 - Re-sequencing of candidate regions at high coverage of 4000 participants
 - Sequencing of 500-600 genomes at low coverage
 - Whole genome sequencing of 1000 participants
 - Genotyping of 1500 participants

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- Co-applicants
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Disease burden in Africa

Poverty

+

Limited access to affordable modern health care

=

Disproportionate burden of disease

Differences in disease prevalence*

Environmental factors

+

Human genomic factors

=

Vast differences in disease prevalence between

- Countries
- Geographic regions within a country
- Ethnic groups in the same geographic region

* all individuals affected by a disease within a particular period of time

Contemporary genetic and genomics research in Africa?

- African populations and their genetic variation in the context of disease are **understudied**
- African researchers are **underrepresented** in modern genetics and genomics research
- African research capacity to undertake population-based genetics and genomics research is **underdeveloped**



Human Heredity and Health in Africa = H3Africa

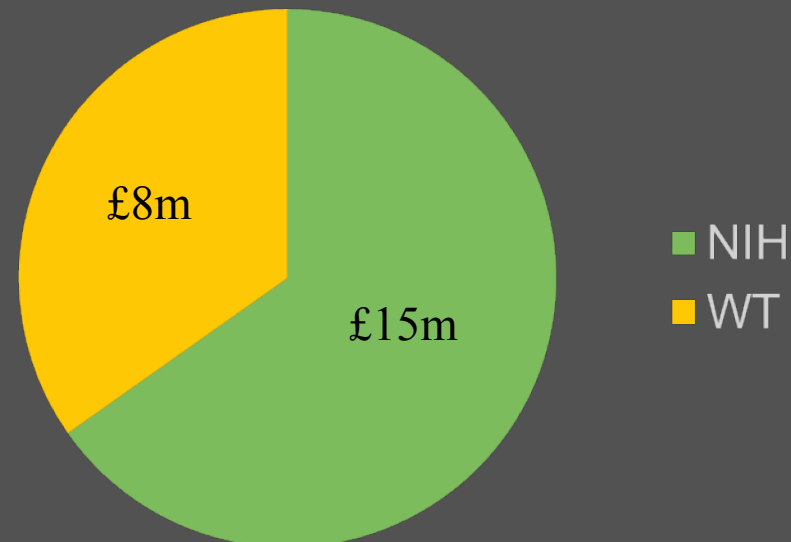
The vision of H3Africa

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



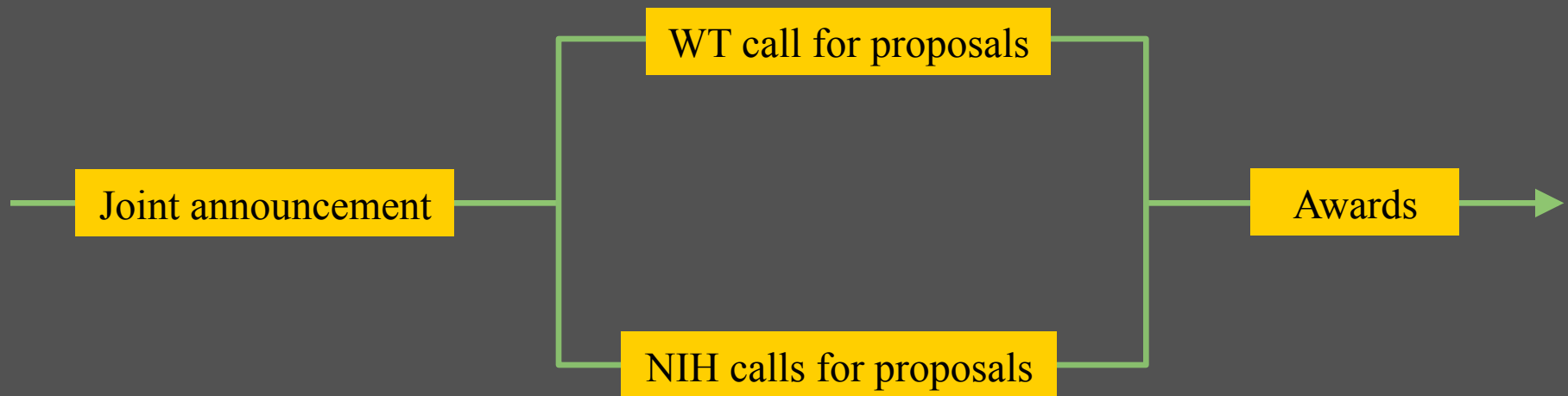
NIH and Wellcome Trust join forces

- Announcement of partnership between the NIH and the Wellcome Trust (June 2010)
- White Paper “Harnessing genomic technologies toward improving health in Africa” (February 2011)
- H3Africa conference in Cape Town (March 2011)



One initiative, multiple calls...

- Themed Strategic Awards to support research networks



- Bioinformatics Network
- Biorepository Grants
- Collaborative Centres of Excellence

Building on existing capacity

- Cohort studies and demographic surveillance systems
- Networks
 - MalariaGEN
 - African Bioinformatics Network
 - INDEPTH
- Training
 - African Regional Training Centre for Bioinformatics and Applied Genomics



Building on NIH and WT investments





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THANK YOU.

<http://h3africa.org>