Transcript of the Human Heredity and Health in Africa Press Conference in London

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(11.00 am)

NEW SPEAKER: Good morning, ladies and gentlemen, welcome to the Wellcome Trust Human Heredity and Health in Africa media briefing conference call. My name is Dave and I will be your coordinator for today's call. For the duration of this call, you will have the opportunity to ask questions. If at any time you need assistance, please press *0 on your telephone and you will be connected to an operator.

I'm now handing you over to Dr Eric Green for today's conference.

ERIC GREEN: Good morning, my name is Eric Green, I'm the director of the National Human Genome Research Institute Part of the US National Institutes of Health. This is being videocast and those of you who are interested in calling in later to ask questions of any of the participants, let me give you that number now, if you want to call and ask questions, it's 44 203 003 2666. That's 44 203 003 2666.

Today the National Institutes of Health and the Wellcome Trust are announcing a new partnership, to fund and conduct population-based studies in Africa. The initiative is called the Human Heredity and Health in Africa project, or H3 Africa. It has several goals, including using new research tools to help us understand the relationship between genes and the environment, in health and disease.

The effort also intends to build capacity on the African continent so that African researchers can conduct these kinds of studies. The partnership, as you will hear, also includes the African Society of Human Genetics, which is helping get the effort off the ground. I'm here from the National Human Genome Research Institute because my Institute will manage NIH's portion of the partnership. But now, to help you understand the NIh's involvement in this, I'd like to introduce the director of the National Institutes of Health, Dr Francis Collins.

FRANCIS COLLINS: Thanks Eric. It's a pleasure to be here this morning and I think this is a significant announcement of a major project which in many ways is timed nicely at the tenth anniversary of the draft of the human genome sequence, here we are, with major advances in technology that have occurred, with the intention, as a joint effort between the National Institutes of Health and the Wellcome Trust and the African Society of Human Genetics, to try to put together a really bold program to understand genetic and environmental contributions to common diseases in sub-Saharan Africa.

The US recognises that the global is not the opposite of domestic, and domestic is not the opposite of global, we live in a global world and if we want to understand the causes of illness, we need to investigate them all over this globe, and Africa is a special place to carry out those kinds of studies. There's more genetic variation there than anywhere else on earth and Africa is the cradle of humanity, and things that we learn there will undoubtedly have broad implications for peoples in all other parts of the planet.

And so we're delighted to be part of this. The focus is going to be both to try to understand genetic contributions to infectious diseases, but also to non-communicable disorders, like diabetes, heart disease, and cancer, which represent the most rapidly growing causes of morbidity and mortality in Africa.

Because these are important goals, and the pay-off is potentially large, NIH has already invested $750,000 this year just to get this project started. The project is going to be called H3 Africa, for Human Heredity and Health in Africa. Starting in October of this year, the US government will invest $5 million a year over a five-year period for a total of $25 million in this program.

We expect that out of this research there will be broad studies carried out to understand the genetic contributions to both infectious and non-infectious disorders as well as surveying the initial ways in which those diseases come about. We hope that this will also provide an opportunity for African investigators to collaborate with each other, the intention here is to empower scientists in Africa to take the lead on this program. This means that the resources will be also including efforts to improve training in that area, and out of this, this will become a community resource project. The idea is that the data that comes forward from this project will be broadly accessible so that all the bright brains on the continent of Africa can work together on the insights that we hope to obtain about health and disease. Again, I'm delighted to be here on this signal moment of announcing a program that I don't think we could have imagined even a few years ago, and the time is right now to get this going, and with our partners at the Wellcome Trust we're delighted to have a chance to support it. Thank you.

ERIC GREEN: Thank you, Dr Collins. I'd like to now introduce the director of the Wellcome Trust, Sir Mark Walport.

MARK WALPORT: Thank you very much indeed. First of all, I'm delighted that we're announcing this program today -- and of course we've worked in close partnership with the National Institutes of Health from the beginnings of the human genome project, so this is a very logical continuation. The last ten years have seen amazing progress, the technology has now changed, so the first human genome took about ten years to do and cost roughly $1 billion, they're now sequencing more than one human genome every day, and the challenge now is to take that from the laboratory to the clinic.

This fits very well with the work of the Wellcome Trust, the work that we have been supporting, and in a similar way to NIH we have been supporting work in Africa for very many years, and there are different elements of it: Firstly, the study of the diseases themselves, the major killers in Africa, malaria, tuberculosis, HIV, childhood infection, but, as Francis has said, non-communicable disease is rising up the agenda as well, so diabetes, obesity, heart disease and cancer are all becoming major problems, as well. So one element is obviously looking at the disease; the other -- and this is highly relevant to public health, actually -- is the study of cohorts of people through time, and it's very important that studies similar to the Framingham study in the United States, and the ...bank study in the United Kingdom. Studies like these happen in Africa, as well, and that's highly relevant to the health of the population, because information from those studies contributes not only to studies of how the human genome

inereacts with the environment, variation, to increase succeptibility to disease, but this also tells you about the health of populations and will direct health interventions, as well.

It's also about people. It's about developing capacity, in order to make the most of human genetics now one needs people who are skilled in bio ... so it's about training people, and about building institutional capacity, the capacity to provide an environment for this sort of research for first-class health research, and so this is in fact an extremely important initiative, and it builds on the type of investment that we've made in institutional capacity, where we've now funded seven networks of institutions across sub-Saharan Africa, involving more than 50 institutions.

The Wellcome Trust will be contributing approximately £8 million over the next five years to this initiative, and I think probably rather more over time, and this will include fellowships, training, capacity development, the phenotyping and genotyping, and we will be allocating that through a transparent grant funding mechanism, so it could not be more timely in the tenth anniversary week of the completion of drafting the genome, and I'm really excited to be making this announcement today.

ERIC GREEN: Thank you. And I'd now like to introduce Dr Charles Rotimi, who's been a driving force in creating H3 Africa. He has an unusual background, he was born in Nigeria and earned his professional degrees in the United States and is now an intramural researcher at the National Human Genome Research Institute, where he directs the centre for research on Genomics and Global Health; he is the president of the African Society on Human Genetics.

CHARLES ROTIMI: Thank you very much, I'm going to try to contain myself here because of the excitement level, this is something that we have been talking about, H3 Africa, under the big umbrella we're thinking in terms of the human genome project, for Africa, but the discussion really started under the umbrella of the African Society of Human Genetics, in 2007, in Cairo, where we felt, as geneticists, and epidemiologists and clinicians and other types of researchers from Africa, that we would like to make sure that whatever benefit that we're going to accrue from using genomics to understand health and human history that doesn't go past Africa, as other revolutions have done in the past.

So started this discussion without really knowing where the money was going to come from and how we're going to do this, we wanted to make sure that we at least try to put this in place and hopefully we'll get the attention of key funding agencies and I'm very, very happy to be here today to say that we are now launching Human Heredity and Health in Africa, H3 Africa, and this is extremely exciting to me, and we're going to get the attention of the National Institute of Health and also the Wellcome Trust, to provide the initial momentum and significant funding for this effort.

So under this umbrella, although we now have expanded this initiative to go beyond just understanding genetics, like Malcolm Francis said, we're going to give need large cohorts of thousands and thousands of people, if you heard research about how genomics is being used, you know it requires a lot of people, people to understand genetic variation and how that relates to health, so we're going to set up clinical cohorts and centers around Africa, to be able to gather this kind of huge number of individuals to do this kind of study, this is again extremely exciting, so we're going to set up resources to enable us to do gene and environment interactions, there are unique environmental characteristics of Africa that you have to

do it within Africa to be able to understand how these interractions were caused, so again, very exciting. Lastly, I'd like to talk about the fact that this funding is really going to start to change things in the way we do research in Africa, traditionally, what occurs is that Africans ... participate in the development of scientific projects and most of the time those resources end up outside of Africa. We are hoping under this umbrella that we are going to do things differently. Those resources are primarily going to remain in Africa, but they will be open to global collaborations, and to create that sense, and to foster intraAfrica collaborations between scientists, so the issue of biological repositories, the issue of building centres of excellence, the issue of the training of young investigators to be able to do the science in the way that they can truly be first authors of these major types of publications. Right now if you look at science and genetics, most of the research is coming from Africa, and indeed not led by African scientists, I don't think that's because of lack of intellectual capacity, but not having the resources to do that at the present time, and I'm hoping that under this umbrella we can begin to do things very, very differently.

Again, to end my comment, I'd like to really thank the Wellcome Trust and the NIH for stepping up to create this kind of big umbrella that we can begin to put things in place to really change the way research is done on the continent, so I'm extremely excited about this opportunity.

ERIC GREEN: Thank you, Dr Rotimi.

So it's important to recognize that the efforts to date in planning H3 Africa have been sort of a tripartite collaboration between the African Society of Human Genetics, the National Institutes of Health, and the Wellcome Trust, and various of us have been trying to plan what some of the activities might be, and we've divided the task in terms of formulating the scientific plan into two working groups, one focused on communicable and one on non-communicable diseases, so I'd like to turn this over to one of the chairs of one of the working groups, to describe what has been done so far. Dr Bongani Mayosi is chairman of a working group focused on non-communicable diseases, he is Professor of Medicine and Head of the Department of Medicine at the University of Cape Town.

BONGANI MAYOSI: Thank you very much, Dr Green, for the opportunity to be at this meeting. I also want to share my excitement with this initiative from the National Institutes of Health and the Wellcome Trust, and I want to point out, as a person working in Africa, that it actually indicates a very, very important shift in the way science is done is Africa. Up until now, I think we have been operating almost in a colonial mode of doing science, people from outside have been coming to collect samples, processing them outside and publishing the papers and advancing the careers of people outside Africa. What is significant about this initiative, as Dr Rotimi has said, is that it seeks to do science in Africa, by Africans, and for Africans, and I think that's a key shift, it's a historic step forward, I think, in the science making of the world, and is a response to the calls that many of us have been making about what needs to happen to effect change, the fortunes of Africans.

For most people, when they think of health problems in Africa they think about malaria, AIDS, and tuberculosis. It is true that Africa is still blighted by these communicable diseases, but of late, we have observed a rise in non-communicable diseases, so that whilst we still do battle with communicable

diseases we must now begin to focus our understanding on the role of genes and environment and the rise of disorders such as heart disease, obesity, diabetes, cancer, as well as mental health. The committee that I chair seeks to identify the diseases which will be most fruitful for work through the H3 Africa projects. These are very difficult questions, and we are still working to identify the sorts of populations, and the sorts of diseases, that will be amenable to a genetic approach.

The web groups which are under the auspices of the auspices of the African Society of Human Genetics, and will provide a network through which researchers will learn as well as develop. My working group, which is working on communicable diseases, has already met several times and we've identified a range of possible studies that can be tackled. We are busy formulating the road map for this project, there is a meeting that is planned in Oxford in early August, where both the people working on non-communicable diseases, as well as those working on communicable diseases, will meet to share their thoughts on how best to take this opportunity, it is likely that there will be quite a lot of overlap between those two groups, working on communicable as well as non-communicable diseases, but it is also likely that there will be unique topics that will become obvious, for example, in sub-Saharan Africa there are certain diseases that are unique to the region that still persist, such as rheumatic heart disease, which crosses the divide between communicable and non-communicable, that still exists there, and humanity has an opportunity to understand its genetic origins, so that as we try to eradicate it we can learn its secrets that it keeps for biology.

So overall, I think this project is not only significant for the people of Africa, for investing in the intellectual capital of Africa, but I think it's also to go to be important for biology and for lessons for the global community at large.

Thank you.

ERIC GREEN: Okay. I would now like to open the floor for questions. Please introduce yourself and let us know who your question is directed to, and if you're asking a general question, I will find someone to answer it for you.

NEW SPEAKER: Mark Henderson from the Times. I have a few questions here, probably for different people. First of all, could I ask Dr Rotimi and Dr Mayosi, perhaps. We've learned an awful lot about the genome in the last ten years but the majority of that has been focused on populations in European and to a lesser extent Asian origian. Do you feel that the genomic revolution has to an extent passed Africa by a little bit, and how do you hope the project will address that? That's the first question. The second question is, what kind of genomic study do you have in mind, are we talking about (GEWOS)?are we talking about further resequencing studies, do we need to do a new version of the thousand genomes project, with very large numbers of African genomes, given the diversity of the African population? And further, for Dr Collins, when I interviewed Howard (Armers?) last year, he mentioned that he was very keen on the idea of science as diplomacy, and as a way of promoting the US, the UK, et cetera, abroad. How does this fit into that?

ERIC GREEN: Let's start with Charles.

CHARLES ROTIMI: I think you can make a very strong case that it is until now, that at least the way we have applied the genomic science to understand health has for the most part really been -- Africa has been left out of that. A good example to look at that is only one genome-wide association study has been published so far that is solely based on the African population, and that's a malaria gene publication, you know, so out of, you know, hundreds of pieces of work that have been done, one can say it's pretty tragic to think that the whole of Africa, only one has been done so far. So to a very large extent we've not equally applied the tools of genomics in terms of trying to understand health. There have been various attempts to try to rectify that, from some people, for example, in thousand genome project has now included more African populations, but again, that's science, like, you know, my colleagues and everybody on the table has been indicating, most of those have been driven in the West and in Asia, so again, this umbrella that we are creating here, we're hoping again to begin to change the dynamics in the way that we can actually apply genomics to diseases that are relevant to African populations, and taking advantage of a unique environment, a cultural environment that is there, to understand genomic interactions. And just as important is to study neglected deseases, to use genomics to understand neglected diseases, for example with funding from NIH and Wellcome Trust we're currently looking at podoconiosis, which occurs in rural parts of Ethiopia, which is a disease that looks like elephantiasis but it's not, it's a completely neglected disease, it used to be in Europe but it's not here any more, and we're trying to use the genome wide association approach to understand the disease. There's genetic supceptibility but you don't get it because you don't wear shoes, so it's a classic geno-environmental interaction that cannot be studied anywhere but in that environment. That's the kind of opportunities that exist on the continent to understand that African problem, and we hope, again, to solve global problems, because Africa is the trunk and root of human evolution and history, so what we get from there is going to be equally important to different parts of the world.

ERIC GREEN: Your second question was what exactly are you going to do: are we doing genome-wide association studies? are we sequencing genomes. I'll take the first part and just point out that, you know, advances in genomic technologies and, as Dr Walport mentioned, the plummeting cost, the fact that the costs are plummeting, are opening up opportunities, so that the answer to your question is we're going to do all those things, that's the bottom line, because we can; and importantly, though, we're going to build a broader based approach for collecting individuals, getting good phenotyping done on the individuals, and bringing technologies to bear on those collections, which is going to provide the opportunities that you heard two of our speakers in particular talk about, projects that otherwise have been neglected but now they're empowered by genomic advances.

MARK WALPORT: I agree with that completely, I think the most difficult bit is the phenotyping, so that the sequencing is changing dramatically, and it's changed in the last five years and it's likely to change equally dramatically in the next five, I suspect, in terms of cost reduction, as Eric says, it's all of the above. The phenotyping, the actual really detailed analysis, is very hard, and I think that's going to be the biggest challenge, actually.

FRANCIS COLLINS: There is a scientific point here that hasn't been made yet, that there are unique aspects of the African population that empower this ability to track down genetic contributions to common diseases. The African population is older than that of Europe and Asia, which means that the

neighborhoods where genetic variations traveled together are smaller. That turns out to be really useful, because if you're a seeking a genetic variation that's functionally involved as a risk factor for diabetes or high blood pressure and you find that in an European or an Asian population, you're finding generally dozens of those variants that are all equivalent, in terms of their predictive power, because they're all in lock step, in this larger neighborhood, and your resolving power is limited by that. In Africa, because the population has been around longer, the neighborhoods are smaller, and therefore the ability to shine a bright light on what the functional variant is that's actually responsible for that diabetes risk or that high blood risk is substantially better, and that is a resource that will help in tracking down these ancient variations that are probably present across the world but in Africa will be more easily delimited to a more precise interval. That's going to help us a lot in the current circumstances, where we have hundreds of these variants that have been identified in general, but for very few of them we actually know which specific letter of the DNA code is responsible for the risk. Africa will answer the question.

Let me answer your question about science and diplomacy. That's a very important question, and that was one of the motivations when I decided what the five areas were of greatest opportunity in science right now. At NIH global health is one of those five, and it's because the science is exciting and because things have come along in the last few years that have made it possible to tackle global health problems in ways that we could not have contemplated, but it's also this opportunity to reach out to the rest of the world with what has been called soft power, or maybe smart power, to be able to draw the peoples of the world together in a shared effort to try to understand the causes of illness and the means to prevent and treat disease. Surely that emphasizes our shared humanity, and surely that is part of what a society should try to do for other societies, surely that fits with what the Wellcome Trust has been doing for decades, and what the National Institutes of Health stands for, but we have a special opportunity to do that and I think H3 Africa is a wonderful example of how to take resources and apply them in that way.

ERIC GREEN: Other questions from individuals in the room? Yes.

NEW SPEAKER: Kate ... from Reuters. You mentioned that you might be setting up similar things to the UK biobank. Do you have any details yet on where those kinds of things will be set up? Also you were talking about collaboration within the continent. Collaboration is quite limited at the moment, as far as I know, and I think South Africa is actually way ahead of the rest of Africa in terms of this, so how are you going to be able to spread that, that network?

ERIC GREEN: I might suggest Dr Rotimi take the first question and Dr Mayosi the second.

CHARLES ROTIMI: Your first question was --

ERIC GREEN: Where is the biobank , where is the repository --

CHARLES ROTIMI: We've set up two working groups that one of the terms of reference for them is to help us to identify first of all to understand what is currently on the continent, in terms of infrastructure, and as part of that evolution process we're going to make recommendations that will guide us to say

these are areas where something like the repository can be facilitated, because there's something that's already there that can be improved upon or that the environment lends itself to that kind of support, to support that kind of infrastructure. But it has to be located in such a way that we are creating ... that all African investigators feel that they can send their samples there, in a way that they can still have ownership but it will be valuable for large collaborative effort, and I think that will go a long way to foster intraAfrica collaboration, between African scientists.

NEW SPEAKER: Sorry, you don't worry that that's going to basically focus the new stuff where the old stuff is already, you're looking at where the infrastructure is already in place, isn't there then a risk that -- you know.

CHARLES ROTIMI: Yes, there's definitely that risk, but we are sensitive to that, and that's not just to continue to give to those that already have, it all depends on the activity, for example, if we are setting up clinical centers, which is going to be critical, as I have indicated, phenotyping is really going to be the limiting step in all of this, and if we set up clinical centers, it will enable us now to be able to engage more African institutions and countries, because we need large numbers, so we are going to set up these centers around, so the the molecular labs may not be as widely spread around, the biology repository, we can do, maybe one or two, strategically located, but there are going to be opportunities to engage more institutions, more centers, based on different activities. The best one, I think, in terms of developing this large, clinical-type cohort that really is needed, most of the data we get out of Africa today is really challenging, because we don't have this large cohort that really says how many people really die from certain conditions and why, and we have a systematic way to track this, so there've a lot of emphasis here on genetics; but I do see a very, very important aspect of this study, that's not necessarily going to be genetic, setting up this large cohort that will enable us to understand causes of mortality and morbidity in a very comprehensive manner and that's going to engage multiple centers across the continent.

MARK WALPORT: There is more infrastructure than you might think, there are demographic surveillance sites across Africa, many of which are looking at quite large populations of up to half a million people, so there is quite a lot and you can't build infrastructure out of nowhere, you have to build science on strength, and I think that the network of demographic surveillance sites are very important, they collect vital information about health and disease, the causes of mortality, in countries where there isn't always vital registration so they're a very important place.

FRANCIS COLLINS: The point is very well taken and I think H3 Africa has the opportunity to build upon a number of other programs where African scientists and clinicians are being encouraged to participate in research in new ways, the Wellcome Trust has their network already of such centers of excellence, we have just started in the US something called the Medical Education Partnership Initiative, which is partly sponsored through (PEPPHAR?) and partly through the NIH, which is aiming to try to build capacity in terms of research capabilities and training, there is an effort through the G8 meeting going on in Canada this week to focus on the needs for more research and other efforts in sub-Saharan Africa, the global health initiative, President Obama's effort is potentially going to include some research activities focused on sub-Saharan Africa. It's a moment where a lot of things are coming together and I think H3

Africa can serve as a really helpful umbrella to bring many of these ideas in a coordinated way, instead of disconnected.

ERIC GREEN: So the second question related to how we're going to sort of change the collaborative style of African scientists, Dr Mayosi?

BONGANI MAYOSI: I just want to pick on the issue of the strength of genetic science in Africa, and point out that although there appears to be areas of activity such as South Africa, when you look at South Africa's contribution, although it accounts for 30 per cent of all the publications on genetics from Africa, but actually, South Africans, over the last few years, have only been producing an average of a hundred papers on genetics a year, that is not a lot, so the existing genetic capacity, even in those areas that are considered to be strong, are, relatively, in the early stages of development and in need of strengthening. When it comes to networks, as I think has been pointed out by Sir Mark, the demographic surveillance systems are there, and there's been a lot of good work done by the African society of Human Genetics in terms of improving intraAfrican collaboration and there have been more progressive funding initiatives, that have led to institutions within Africa working together, universities, such as, you know, the initiatives from the Wellcome Trust to build research capacity, the initiative from (PEPPHA?) and the NIH to try to encourage and build capacity within medical schools. So more recently there's been quite a lot of intraAfrican cross talk, which I think is to be encouraged the universitites are organised, the academies are becoming more ogrganised as a voice so that this particular initiative will find an environment that is in fact ready to move forward.

ERIC GREEN: Other questions from the floor? If not, are there any questions from any phone callers?

NEW SPEAKER: We don't have anyone on line just at the moment, no.

NEW SPEAKER: I have something.

ERIC GREEN: Sure.

NEW SPEAKER: Will there be any pathogen sequencing as part of this?

CHARLES ROTIMI: I would imagine definitely that there will be, again, if you're dealing with the genetics of infectious diseases I think you have to look at both the host and the pathogen, I would definitely imagine that would be part of this effort, yes.

FRANCIS COLLINS: And maybe even the vectors.

CHARLES ROTIMI: Yes.

NEW SPEAKER: Can I come back, as well, if that's okay.

MARK WALPORT: Keep it going.

NEW SPEAKER: I wanted to ask about the chronic disease elements, I mean, you said quite clearly that one of the things you think you're going to get out of this is the more original causes of some of these

chronic diseases. Do you think you're going to find, you know, key differences in the way that these diseases develop in different populations as well? Is that what you're -- I mean, I know you don't know the answer yet, but you've kind of got a suspicion of what the answer is, is that the kind of thing that you're going to be investigating? As well as -- I mean, you know, again, are we looking for things that will help the rest of the world or are we looking for only things that will help African populations in dealing with what is a very fast-growing problem.

ERIC GREEN: So the answer is going to be both, but somebody could --

CHARLES ROTIMI: I could try to add some more to that. I think that, again, it will depend on the disease that you're looking at, there are certain diseases that will have unique environmental factors that you can't do it, you know, in another place, you have to be in the local environment, to take advantage of what is it -- for example, if it is diet, that has implications for what you're studying, and then you need to collect dietary information within that environment to understand, if you want to, for example, see the impact of salt on hypertension, you need to see how people are cooking their food, and one of the things that is really interesting in the context of doing this kind of work in Africa, we did a trial in rural Nigeria, to try to see how reducing salt can lower blood pressure, and it turned out that it was actually a much easier study to do in rural Africa than in London, or somewhere in the US, because the source of salt in the diet is limited, and mostly it comes as a result of adding stock cubes to your soup; and teaching women how to reduce that, or eliminate it, significantly lowered blood pressure. That's an unique environment in which we can do that, and also, the fact that, like what Francis and others have indicated, is at a genetic level you do have this opportunity to fine map, to use the African, you know, small ... structure to better localize the signals that may otherwise be not so clear in European or in Asian populations, so the answer is that we're going to do things that will directly benefit the African population but because we all share common history as humans, that information, I think, for the most part, will directly be relevant to other human populations.

ERIC GREEN: Other questions?

NEW SPEAKER: We have no questions coming from the phone line at the moment.

ERIC GREEN: Have we not mentioned anything that anybody on the panel wanted to make sure that we said, have we got across all the points? If not, I thank all of you for participating, we'll look forward to talking with you more about this as the program progresses, and thanks again.

(11.40 am)

(The webcast concluded)