

**Dichotomy Between Social Identity and Ancestry in Large Population Studies**  
*Charles N. Rotimi, Ph.D.*

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DR. TUCKSON: Let's turn now to Charles Rotimi, who will share his thoughts on the dichotomy between social identity and ancestry in large population studies.

Charles, thank you. Again, Charles is Acting Director of the National Human Genome Center at Howard University.

DR. ROTIMI: Thank you. Thanks for inviting me.

What I thought I would do today is share with you some of my thoughts, some of my biases, and how I think about some of these issues in relation to how we do large population studies, and how we try to represent different groups, or not represent different groups for various reasons.

One of the first comments I wanted to make is that depending on what we are doing, we desire different levels of resolutions. For example, if we are trying to identify how common alleles, at least 5 percent or higher, impact on disease, we will define our study in such a way that we have a level of resolution to get at that. For example, HapMap.

If we want to identify people who eat beef, that is one level of resolution. If we want to identify people who not only eat beef, but eat it in a certain way, cook it in a certain way, that's another level of resolution, and you may have to go to some parts of the world, and not other parts of the world.

So again, depending on how we are defining ourselves and our identity, we do stop at different parts of this. If you really look in terms of our own history, one can say that we are indeed Africans, and that we started somewhere in terms of the roots and trunk of human evolutionary history from somewhere in Africa.

But of course time did not stop, and we are migrating to different parts of the world. Depending on your socialization, and depending on what you are willing to accept, how you want to define yourself, and indeed sometimes it is the question of survival, the identity you want to put forward. Your level of resolutions do differ, and we have to always bring that to bear.

That is why it is extremely important when we are defining large-scale studies like what we are planning here, that is capable of impacting on health for a very long time, we need to be extremely careful as to who is at the table, and who is making decisions.

Not just in terms of science, but in terms of how is this representing the people. Especially if you are using taxpayer's money. So again, it is extremely important for us to appreciate all of that. And indeed scientists were socialized before they became scientists. We bring all of our baggage to these issues.

Also I want to again, make some distinction here. That is in terms of when we are talking about understanding etiology, and when we are talking about eliminating her disparity. Sometimes we say these things and say they are the same, and sometimes there is overlap. I actually wanted to make this overlap a little bigger, but I couldn't figure it out in the PowerPoint.

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It is indeed a little bigger than that, but there is not a complete overlap. For example, if you are interested in eliminating her disparity, you may be interested in how people get access to care. That may have nothing to do in terms of etiology. So again, we need to be clear as to what is it that we want to do.

Looking at her disparity may have more involvement in strategy at a social level. Again, typically we look at a diagram like this, and we usually use this to represent her disparity, and sometimes to point out etiology.

One of the things I wanted to point out here is when you look at a 50 percent prevalence of Type 2 diabetes among Pima Indians, one has to wonder within the same United States as to what is going on. The gene hasn't changed that much. It doesn't mean genetics is not involved, but it hasn't changed that much over the years.

One of the things that we do know is that characteristics have changed. So again, looking at this, you can be looking at etiology, you can be looking at her disparity, and at the same time, you may be addressing both.

Now, this is on account of her disparity. This is looking at populations of the African diaspora. Again, this is where I used to stay when I was working at Loyola Medical Center in Chicago. It is 84 percent African American. This whole cohort here is over 10,000 people from different parts of the Diaspora.

What you do see, again, is that this is clearly her disparity issue among people who have African ancestry. About 14 percent here, about 34 percent here. You do see a dramatic increase in body mass index. So clearly how heavy you are and the environment where you find yourself has serious implications for hypertension.

This is a new study that is extremely important in terms of how we address some of these issues, what we are calling disparity, and how it plays out in different ethnic groups in different parts of this continuum in terms of human experience with the problem of hypertension. This was done with Richard Cooper and his colleagues recently.

What did you see? Again, clearly depending on where you are, you do have very different rates. What I want to point out here, when you look at whites, the group we called whites within the United States in relation to other ethnic groups, typically we see it as a huge disparity.

Yes, there is a huge disparity, but if you place all of these populations and you look at it together, you see that it is truly a human experience. When you are in Germany, your rate of hypertension is really, really high. The U.S. whites tend to be quite healthy in relation to other European populations.

Therefore, it exaggerates, to a large extent, how we think about the issue of who is getting hypertension, and who is not. So again, this slide here is really important when we are doing a large-scale cohorts like this, that we have to bring to bear cross-culturalized and international experiences, so that when we are defining our variables and strategy, that we take those into consideration.

This is the same sort of study. Now, if you group all of your opinions, the populations and all African populations, you do see that the Europeans have a much higher level of diastolic blood pressure. But you don't hear this when you hear people talking about experiences of high blood

pressure and hypertension. So again, cross-cultural comparisons are extremely important, and international experience is extremely important in doing these large-scale studies.

Also, in what we want these large-scale studies to answer, we also have to define this study. Do we want it to just stop at a level of who gets diabetes, yes/no? Who is reacting to drugs, yes/no? Or are we also wanting to tell some stories about who we are, where we are from, and are we related. It may be useful. If indeed it is, then we need to bring to bear a design strategy that will help us to see those things in the way that we are not reinforcing old notions about who we are. So in that regard, ancestry, in my opinion, becomes a very critical thing for us to consider.

I like these slides a lot, because every time people talk about the issue of race/ethnicity, I am getting so tired of the whole issue, but I always ask myself, where do we draw boundaries, and how do we draw boundaries? Again, it really just depends on where you grew up, how you were socialized, the things that you are afraid of, and the things that you like.

So who is black? This is a whole spectrum of who is black. This spectrum is indeed also limited. You can expand this. There is no limit to it.

One of the best pictures I have seen so far is on the PBS website where they actually show that you can see all the variations of human complexion right there in Africa. All of it. I'll show you some of my experiences when I was in Brazil. I'll tell you a story in a minute. But you do see that these all would be considered black. But again, they have a radically different ancestral history from the Aborigines, to Ethiopia, and different parts of the world.

I put this slide here to tell a story about what we are doing in terms of Type 2 diabetes in the African Diaspora. This is a study we are doing in Nigeria and Ghana, but the real intention here, what we are trying to get at, is why the high rate of Type 2 diabetes in African Americans.

We felt compelled to really get at that. We need to go back to the source population of African Americans. We all know the ugly history of the Middle Passage, and that most African Americans, again, came from this part of West Africa, and again, Mozambique.

The story here I really want to point out is when we started writing the manuscript reporting the results of this study, one of the things that reviewers took us to task on is how you are sure that you can combine all of these groups together, because these are an affected pair design.

We analyzed the cohort. There were about 400 affected pairs with Type 2 diabetes. We analyzed this cohort as a uniform group, as one group. But repeatedly the reviewers gave us trouble and said, why do you think you can combine all of these groups together?

But the point here is that I have done similar work in African Americans, and no reviewer has taken me to task that why do I think African Americans are a uniform group? You see the way we are socialized impacts even on the way we review the work and what we fund, because indeed, this kind of work, if you are writing a grant, it can be killed based on that reason only, that reasoning, but you know that even the ancestral history of African Americans is even broader than what we have here. But nobody takes us to task on it, because the assumption is we are dealing with a uniform, homogeneous group. So we need to be very conscious about what we're talking about. The problem I see is that group identity is confused with ancestry, and self-identification is confused with more complex ancestry.

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Now, when I prepared the slides for this talk, I wondered about this issue. But if you think the issue of African Americans is confusing, not to talk about the history of the Hispanic population, or what we would call Hispanic, that is completely mind-blowing when you look at it where we classify who we put under that umbrella. How we approach it, with some notion of uniformity, to me really begs the question of what are we doing.

It may indicate why we are not getting some consistent results in some of the work that we've been doing, because we lump people together based on some very interesting groupings.

For example, when we look at the Census, the Census is pretty clear. I think this is one of the issues that confuses it. We say we're not doing anything that deals with biology, we are just looking at it where society has designed itself, and we are collecting information on that. But what we do as scientists, we impose biology on that, or want to impose biology on that. Sometimes it works, sometimes it doesn't work. So I say Hispanic, but you can be of any race.

So this is just to point out some of the groups we call Hispanic. Mexican, South America, Cuba, Puerto Rico. This is a whole list of people who have radically different ancestry if you really go into the history.

I put a slide here. I took this picture on my last and only visit so far to Rio. It was friendly and informative for me, and I enjoyed myself quite a bit.

I was flabbergasted when I drove on a major road going to the university in Rio, and I saw this junction. It took me back to my young elementary school days when I was in Nigeria going to school. We used to put our school bag -- ours was made out of a metal box, and we put them in on our heads. We were so good, we could play soccer on the way to school.

But what it turns out is that this is a sacrifice made to the Gods in Rio, and it followed the tradition. I was extremely surprised by that. What you have is these are the feathers of a chicken, pots, oil, and wine, making offerings to the God for protection.

This is three years ago in Rio. Now, talk about gene/environment interaction. If you are studying this group, then you had better take into consideration the African ancestry and history, and why this group has kept this experience over the years. What does it mean, therefore, to have Cuba, Mexico, and Brazil as Hispanic in studying the group?

This is, again, to show you again how we lump people and sometimes lose quite a bit of information. If you look at people who are under 18 and 65 plus, you do see that depending on which population, the Hispanic population that we are sampling, you could be doing yourself a service or a disservice.

The same thing also here in terms of education. There are radically different education experiences.

I think the same story is true when we look at Asians. We do group all of these groups, and we call it Asian. Now, for example, HapMap is looking at Japanese and Chinese. Now, how does that represent the experiences of these people and the ancestral history of these people. And if indeed there is something that has been selected over the years and these are the only experiences, it may indeed not be well captured. I don't know. But again, for us to just be conscious of who we are calling Asians.

One of the other extremes in this experience in working, and actually I live in the United States, is that depending on how you see yourself and how you relate to your environment, you tend to lose some of the social identity that you have. It's not important anymore to be German American. It doesn't offer you any extra advantage, okay? Whereas it may be extremely important for you to identify yourself as Native American, or Hispanic, or however it is you want to do it.

But again, this shows that depending on the group, who is sitting at the table, they might see the relevance of setting things and not the relevance of all this. So we need to begin to be very careful as to why we are using this and how this came about, and what is their present relevance.

Now, to sort of wrap up here, looking at ethnicity identity in terms of Africa. One of the things that has happened over the years, and this is just one of the issues I take with cultural anthropologists, and I tend to single them out, but they are not the only guilty one.

It is this whole notion of things which end up in part of the world, or in a remote environment, sort of static and that they don't change, or that we don't want them to change. So if people are cooking in one particular way, we want them to continue to cook, whereas in our environment, we are creating jets that can carry 800 people now and things like that. We are lots of society to evolve, and one part is to stay static.

I don't know the rationale behind that, but the point is that just like anywhere in the world, identity changes. How we look at ourselves changes. Those things have been based on economic, political, and whatever else ways for us, especially the issue of survival.

I would say that we are extremely efficient in the way we identify differences, because I do believe somewhere down the road that we need it to be so. We need it to be known who is family, who is friend, and who is outside of that cycle. So we are very, very good at seeing differences that may not actually be the reality.

So the message here really is that things have not remained static, that identity changes. It is multi-layered. Depending on where you are looking, genetics may be important, and they may not be. Making the sacrifice at the junction on the road may be more relevant in terms of the issue.

So I'd like to end by just again bringing us to some areas in terms of who is telling the story. Depending on who is telling the story, depending on who is designing the study, depending on who is present, who is funding this study, you can tell stories and history in a very, very different way.

For example, during the earlier interactions between Europeans and Africans, there were some various surprises that were not anticipated, and because of the biases that came or preconceived notions, certain things were very difficult to assert.

By the way, this is where I grew up. So I know this history quite well, and some of the issues that we have, again, we are still trying to get some of the artwork that went away a long time ago. But the take-home message here for this particular slide is that we need to think more comprehensively if we are going to design very large studies, especially if we are going after gene/environment interactions.

Again, this is the same set of points. I'm just going to skip these.

But where do we sample? Again, it becomes very, very relevant. Very interestingly, only European Americans, again, that's a very broad term, no question who is under that umbrella. You can sample anywhere in the United States for that group.

But if you are interested in American Indians, Eskimos, Asians, blacks, or Hispanics, you have to go to different parts of the United States. For example, you do see most African Americans here. The people we would call Hispanics are here.

So again, it is very, very important if you want to emphasize efficiency that you go, and depending on also who you are putting under that umbrella of Hispanic, it may do you better to be in Florida and to be in California. Again, just for us to be conscious of that.

This is something that we did recently at Howard University with Nature Genetics and some of the people that are here who actually contributed to that effort.

It is really to try to get at how do we explain the fact that, yes, there is variation at the genome level, and that variation needs to be studied. How do we do it in such a way that we don't bring our whole notions on it? Let it tell its own story so we can really know how we are related.

But the point I also want to make with this slide is depending on where you draw circles here, here, or here, the genetic variation will tell you a story. If you move, it will tell you a story. There will be overlap. There might be some differential frequency. But usually what happens is you don't have uniqueness. It is just a gradation.

So in terms of large scale, I look at large scale as this big umbrella, and that we are trying to fit a lot of things under this big umbrella. Depending on how many things we want to fit under this umbrella, it would determine the level of compromise that we are going to have to make. This could be prostate cancer, heart disease, or something within heart disease. Again, this could be infectious diseases, HIV, whatever.

So depending on what are the things that we want to put under this umbrella, we are going to compromise. We are going to have to make some compromises. I want to say at this point that the really critical thing here is the cost of phenotyping that is going to drive all of this effort.

At some point in the very near future, five years or so down the road, we are probably going to have all of our genetic variants on a chip and put it on our neck like an I.D. card.

But the environment is interesting, because it is ever changing on us, and it would depend on how we feel today. My blood pressure can be high or it can be low. Just looking at you, I can be smiling, and things are happening to my physiology. How do we capture that in a way that we can relate it to genes that are supposed to be under the influence of this environment? I think we need to think carefully how many things we want to put under this umbrella, and what we want it to answer.

So as the final note here, the whole point I'm trying to make in my presentation, or tried to make, was this point here. "The historical, anthropological, and linguistic definition of populations, within which genetic finders are correlated to represent superficial understanding of the dynamic history of presenting ethnic populations or high-risk populations were developed."

The future use of drug therapy will not depend on the (inaudible) race/ethnicity, but on the individual patient. I think David Goldstein made this point earlier. The idea then is not to

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eradicate or ignore differences, but to redefine or move beyond social group labels such as (inaudible) to more precise categories of differences with justification for establishing such differences.

Thank you very much.

DR. TUCKSON: Thank you very much as well. We very much appreciated that. Thank you.