Race, Genomics and Medicine

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In 2000, President Bill Clinton said, “I believe one of the great truths to emerge from this triumphant expedition inside the human genome is that in genetic terms, all human beings, regardless of race, are more than 99.9% the same.”¹ For hundreds of years, the concept of ‘race’ has intruded into the lives of millions. The preamble of UNESCO’s Constitution, adopted in 1945, specifically named racism as one of the social evils which the new organization was called upon to combat.² As early as the 1950s, Ashley Montagu and his colleagues proposed that race is a social construction that is developed by the ideas of a particular society. The “Race Question” was written by an international consortium of scientists for UNESCO and edited by Ashley Montagu, who later wrote extensive commentaries on the draft. The 1950 UNESCO statement says, “For, like war, the problem of race which directly affects millions of human lives and causes countless conflicts has its roots ‘in the minds of men.’”³ Approximately twenty years later, Richard Lewontin supported Ashley Montagu’s claims with his research on population variability and on blood group polymorphisms in differing races. In his work

³ ibid, p. 1.
“The Apportionment of Human Diversity” in 1972,\textsuperscript{4} Lewontin proved that individuals with a ‘race’ differed more within that group than they differed from individuals in differing groups.

In 2003, the Human Genome Project was completed. Many Americans, including President Clinton, had an abundance of optimism following the genomic revolution, although eugenics and the concept of ‘race’ were even more apparent within the American ‘racialized’ society, and new claims for racial differences immediately began to appear based on genomics. In this paper I will argue that the concept of race is not supported by genomics. Race, scientifically-speaking, is the identification of a particular set of markers due to geographical isolation, but Americans socially construct ‘race’ to accommodate to the stigmas of society and the standards we believe should be upheld within these developed norms.

\section*{1.1 Ashley Montagu & Social Expectations/Language}

Many scientific facts about ‘race’ are known, but within American culture, the word is normally being used to refer to a social construct not a scientific one. Before Lewontin, there was Ashley Montagu (1905-1999). Montagu was an anthropologist who particularly gravitated towards popularizing topics such as race concerns in politics. He also worked as an investigator for UNESCO in the 1950s in the investigation of the \textit{Race Question}. He advocated the elimination of ‘race’ in science and the replacement of ‘race’ by ‘ethnicity’ in most contexts. ‘Race,’ although used in scientific contexts to signify ancestry perhaps, is manipulated in our vernacular language, as if it was a biological

marker, although at best it is a social construct which imposes an identity.\(^5\) Ethnicity, on the other hand, signifies that an individual belongs to a particular group; it is culturally learned and a person embodies this as their self-identity rather than because of an assumed biological marker. Ethnicity is adopted rather than imposed. An example is the young man in this video, who physically looks Chinese, but is ethically Italian. Although society pressures him to admit to a physical identity, which predominantly appears oriental, he claims his ethnicity as Italian because he was raised under Italian traditions in Italy by his mother \(\rightarrow\) [http://youtu.be/aDz3BJDPXHA](http://youtu.be/aDz3BJDPXHA) [location 3m 15sec].\(^6\) According to “The Race Question,” the word ‘race’ should be omitted completely in our language and we should replace such a word with ethnicity, which is related to who the person considers they are rather than how we as Americans want to label them. Essentially, race is a discredited scientific concept.

### 1.2 ‘Race’ is Real? The Archaic Insult to Social Scientists

Nicholas Wade, a science writer with a specialty in evolution, has recently suggested that race does have a biological basis, but racism does not. In his book, *A Troublesome Inheritance: Genes, Race and Human History*, he suggests that understanding of the genetic differences between human groups should not lead to a society that enables racism. In his *TIME* article “What Science Says about Race and Genetics,” Wade says, “In each of these races, a different set of genes has been changed

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by natural selection. This is just what would be expected for populations that had to adapt to different challenges on each continent. The genes specially affected by natural selection control not only expected traits like skin color and nutritional metabolism, but also some aspects of brain function. Though the role of these selected brain genes is not yet understood, the obvious truth is that genes affecting the brain are just as much subject to natural selection as any other category of gene.”

He proposes that during a period 30,000 to 5,000 years ago there was a racial divide of three principal races: Africans (those who live south of the Sahara), East Asians (Chinese, Japanese, and Koreans), and Caucasians (Europeans and the peoples of the Near East and the Indian subcontinent). Unlike Lewontin, Wade believes he is using the patterns of human variation in populations as claiming that race is a valid, biological taxonomic category in science.

Wade’s arguments are supported by the research of California economics professor, Gregory Clark, and according to Matt Ruben, perhaps even the work of Francis Galton. Ruben says, “Galton founded the pseudo-science of Eugenics. He used a bastardized version of his cousin Charles Darwin’s theory of Natural Selection to argue what Clark argues: that genetics explains why elite families and groups remain on top from generation to generation, while the rest of us have a tougher time.”

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8 Ibid.

Clark first intrigued Wade with his ‘plausible yet unexamined possibility: that productivity increased because the nature of the people had changed.’ Clark’s proposal was unique, and a challenge to most forms of conventional thinking because economists ‘tend to treat people everywhere as identical, interchangeable units.’ The research of Clark’s that Wade speaks about was published in Gregory Clark’s *New York Times* column titled “Your Ancestors, Your Fate” on February 21st 2014, which proposes that for most individuals one can predict status by the earnings of people within their ancestry. Clark had estimated that fifty to sixty percent of variation in overall status is determined by an individual’s lineage. This experiment was conducted “…after examining reams of data on surnames, a surprisingly strong indicator of social status, in eight countries — Chile, China, England, India, Japan, South Korea, Sweden and the United States — going back centuries. Across all of them, rare or distinctive surnames associated with elite families many generations ago are still disproportionately represented among today’s elites.” After making it clear that wealth is passed own generationally, just like someone’s genes, Clark leaves a statement of optimism: “Does this imply that individuals have no control over their life outcomes? No. In modern meritocratic societies, success still depends on individual effort. Our findings suggest, however, that the compulsion to strive, the talent to prosper and the ability to overcome failure are strongly inherited. We can’t know for certain what the mechanism of that

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inheritance is, though we know that genetics plays a surprisingly strong role.\textsuperscript{12} This is very reassuring.

What problems arise with Gregory Clark’s argument? Much like Galton, Clark completely misuses evolution. In her book review of Wade’s \textit{A Troublesome Inheritance: Genes, Race and Human History}, Margery Lucas does agree that wealth and fertility have been linked, but she corrects Clark’s absence of addressing that linkage in traditional, foraging societies as well as agrarian societies.\textsuperscript{13} Lucas says, “Hard work and patience, such as that required to track a large animal over time and distance, are also rewarded in these cultures, although the pay off may be in a different currency than the kind measured by Clark (e.g., number of wives or amount of food shared).\textsuperscript{14} Also, I know of no studies linking wealth, fertility, and traits for social competency, let alone studies identifying a genetic component. Even Wade admits that it is unlikely that a genetic component will be found since there are no doubt many genes underlying these traits and they probably each have very small effects. “ Even with the support of Gregory Clark, Wade’s book makes no substantial contribution to the discussion of ‘race.’ In Wade’s book, he introduces Clark’s research by that stating that “…when a civilization produces a distinctive set of institutions that endures for many generations, that is the sign of a supporting suite of variations in the genes.”\textsuperscript{15} Lucas adequately responds to this statement

\begin{itemize}
  \item \textsuperscript{12} \textit{Ibid.}
  \item \textsuperscript{14} \textit{Ibid.}
  \item \textsuperscript{15} Nicholas J. Wade, \textit{A Troublesome Inheritance: Genes, Race, and Human History}. Penguin Group (USA), 2014. 150.
\end{itemize}
as being one “…That is quite an unwarranted inferential leap”. Even if one were unacquainted with Nicholas Wade’s book, Gregory Clark’s research speaks volumes about the controversial, and speculative ideas of the two men.

This brings about another important question: What is race, and does race exist at all? Although this is a legitimate question and with the support of genomics it is proven race is more social than it is scientific, people still attempt to categorize one another. For example, in the United States, Americans are identified by five racial categories: American Indian/Native American or Alaskan Native, Asian, Black or African American, Native Hawaiian or other Pacific Islander, and White/Caucasian. There are also two categories which questions one’s ethnicity as ‘Hispanic or Latino’ and ‘Not Hispanic or Latino.’ This classification was introduced in 1997 by the Office of Management and Budget, and adopted by the Census Bureau. All this occurred before the completion of the Human Genome Project in 2003, however the five categories still remain the identifiers of race in the United States.

2.1 Richard Lewontin and his Supporters: We are 99.9%

identical

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16 ibid.
http://www.census.gov/topics/population/race/about.html
As early as the 1970s, Richard Lewontin researched the blood group polymorphisms in differing races, and proved that groups identified by ‘race’ differed more between individuals within that group that they differed from individuals from differing groups. His results were published in his work “The Apportionment of Human Diversity” (1972). According to Lewontin, the concept of race is far more complex than advanced biochemistry, because there is no definite understanding of when one race begins or ends. In Lewontin’s research, he has seventeen polymorphic loci and seven blood groups, which included Caucasians, Black Africans, Mongoloids, South Asian Aborigines, Amerinds, Oceanians, and Australian Aborigines.\(^{18}\) Note that Lewontin used seven categories of ‘race’ in contrast to the U.S. Census Bureau’s five. After Lewontin’s measurement of his fixation indexes using the seventeen polymorphic loci, Lewontin found that 85.4% of genetic variation is found within a single population of similar ‘race,’ 8.3% is found between the varying populations, and 6.3% is accounted for by his racial classification. Overall, the majority of the total genetic variation between individuals is a mere 0.1%, so humans are 99.9% identical in regards to genomics.\(^{19}\) Lewontin says, “it is clear that our perceptions of relatively large differences between human races and subgroups, as compared to the variation within these groups, is indeed a biased perception and that, based on randomly chosen genetic differences, human races and populations are remarkably similar to each other, with the largest part by far of human variation being accounted for by the differences between individuals.”\(^{20}\) More recently in research, Arthur G. Steinberg and Charles E. Cook came to the same


\(^{19}\) ibid, 396.

\(^{20}\) ibid, 397.
conclusion as Lewontin. In *The Distribution of the Human Immunoglobulin Allotypes*, they examine Gm haplotypes commonly present in eleven races.\textsuperscript{21} There is no master list of what is considered scientifically a race. The examples considered so far do not even agree on the number of races; are there five, seven, or eleven? Even if there was a master list, research from Lewontin to the Human Genome Project makes it apparent that genetically we do not differ greatly.

2.2 Lewontin’s Fallacy

Up until about the 1990s, Montagu and Lewontin’s critique of race was widely accepted. One of the promises of the Human Genome Project was to progress to a system of personalized medicine, but instead an uprise of racialized medicine has taken control. As this uprise began to occur, a group against Lewontin’s concept of race had also begun to form. In A.W.F. Edward’s “Human Genetic Diversity: Lewontin’s Fallacy (2003),” he claims that Lewontin’s conclusive findings were unjustified because Lewontin’s form of argument overlooks the fact that most of the viable information that differentiates populations from one another is hidden within the correlation structure of that data and not particularly in the variations of individual factors, per se.\textsuperscript{22}

The reality is, genetic variation exists at a continuum across geographic regions. In the blog, “How to Paint your Panda,” (titled “Lewontin’s Fallacy” and Race’), the writer Alexis, says that many anthropologists agree that if there are no definite categories, race simply cannot be usefully be applied to humans. She goes onto to say that this is much like the concept, coined the “Fallacy of the Beard,” which refers to an analogy.


about the status or type of an individual’s facial hair. Alexis says, “The argument would suggest that because you cannot assign a differential category between 100 hairs on a beard from 102 hairs, or so on, that statuses of beard lengths (can be simple as long/short, or can refer to things like five o’clock shadow) do not exist. The reason this is not the case is because beard lengths have extremities, such as being completely clean shaven, implying that while nominal in nature, the partitioning of such categories of beard status rely on the ‘number of hairs’ in a ratio level of measurement, where there is a meaningful zero (lack of any hairs). This cannot be done for human genetic variation.” 23 This may appear silly in comparison to the debate about race, but by and large, it’s not that race does not exist, but instead that the definition of race is subjectively classified. The definition of ‘race’ relies more on societal context as opposed to any clear biological differences.

Another example of an anthropological approach can be illustrated in Lise Funderburg’s article “The Changing Face of America” featured in National Geographic (Photography by Martin Schoeller), which discusses the concept of ‘race’ in the United States. As previously discussed, the U.S. Census Bureau identifies by five racial categories: American Indian/Native American or Alaskan Native, Asian, Black or African American, Native Hawaiian or other Pacific Islander, and White/Caucasian. Funderburg says, “The multiple-race option has been lauded as progress by individuals frustrated by the limitations of the racial categories established in the late 18th century by German scientist Johann Friedrich Blumenbach, who divided humans into five “natural

varieties” of red, yellow, brown, black, and white. Although the multiple-race option is still rooted in that taxonomy, it introduces the factor of self-determination. It’s a step toward fixing a categorization system that, paradoxically, is both erroneous (since geneticists have demonstrated that race is biologically not a reality) and essential (since living with race and racism is). The tracking of race is used both to enforce antidiscrimination laws and to identify health issues specific to certain populations.”

Surprisingly, the U.S. Census Bureau is very aware that “racial categories are flawed instruments, disavowing any intention ‘to define race biologically, anthropologically, or genetically.’ And indeed, for most multiple-race Americans, including the people pictured here, identity is a highly nuanced concept, influenced by politics, religion, history, and geography, as well as by how the person believes the answer will be used.”

There are too many factors to adequately create a list of definite categories of ‘race.’ Like many of the issues seen in the YouTube video, there is a pressure within American society to identify with what individuals phenotypically appear in regards to ‘race,’ rather than claiming loyalty to their cultural upbringing which would be better defined as their ‘ethnicity.’ For example Yudah Holman, 29, self-identifies half Thai and half black, but when prompted to categorize his ethnicity he marks Asian. He predominantly self-identifies with his Thai background first “because my mother raised me, so I’m really proud of being Thai.”

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25 Ibid.
3.1 The Human Genome Project

The Human Genome Project remains to this day the largest collaborative biological project ever, performed in approximately twenty research facilities all over the world.\textsuperscript{26} The Human Genome Project was first funded by the United States Department of Energy in the 1980s with intentions of protecting the genome from the possibility of mutagenic effects caused by radiation, but later it became the project we are familiar with today.\textsuperscript{27} The overall goals of the Human Genome Project entailed the process of mapping, sequencing, and identifying each gene within the \textit{Homo Sapiens} genome, and storing and analyzing that data to identify and treat genes that could be responsible for disease or disorders. According to Francis Collins, the director of the National Human Genome Research Institute, “This is the single most important genomic resource for understanding human disease, after the sequence.”\textsuperscript{28} The results of the Human Genome Project supported the theory that humans, regardless of ‘race,’ were 99.9\% identical across their genomes.\textsuperscript{29} If this is true, why are we developing race-specific drugs? Why is it relevant to test medications in clinical studies by race, or by someone’s ethnic background? David Lawrence, author of “A Rational Basis for Race,” says that the Human Genome Project promised to “…dispel deep-rooted myths about differences conferred by race, and the health disparities attributed to such divisions.”\textsuperscript{30}

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acknowledges that genetic variation does exist, it’s what makes us individuals, and it does correspond to geographically isolated groups—-but we need to be reminded that humans are not so distinctively different that they can be categorized by ‘clean color-based lines.’  

### 3.2 How was Genomics supposed to eliminate racism?

The Human Genome Project had hoped to discover many of the genetic markers that linked to diseases, such as Alzheimer’s and heart disease. Today, genomes are still being sequenced for exactly these reasons, but the construction of databases that analyze genomes are participating in discriminatory practices. In *ScienceFriday*’s news program, “Does your Genome Belong to your Family, Too?,” Christopher Intagliata, Susan Wolf, and Robert Green discuss the issues of autonomy and the ownership of one’s sequenced genome. Years ago, sequencing a genome would cost millions of dollars, but today it is at a feasible cost of approximately $1000. This is still quite expensive, which may explain why most biobanks consist of predominantly white patients.  

How were these genetic markers significant in comparison to the rest of the genome? In Michael Kenny’s “Genomics, Genetic Identity, and the Reconfiguration of ‘Race,’” he discusses how DNA sequencing has broken down cultural barriers because the genetic markers have identified Phoenician ancestries that before were unrecognizable. Even before the completion of the Human Genome Project, genomic technological devices

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31 ibid.  
were employed to categorize people into ‘types,’ or as we will discuss into ‘haplotypes,’ which were based upon an individual’s relative disease risk. Kenny says, “A ‘haplotype’ is such a kind, and is defined as ‘the combination of allelic states of a set of polymorphic markers’ on a chromosome or chromosome region ----However, many markers cohere in groups that are inherited as a block, thus giving rise to ‘haplogroups’ that reflects population history (e.g., migration, founder effects, demographic bottlenecks, natural selection, genetic drift).“ 33 The fact that humans fall into such groups has important implications for medical research, deep-time studies of genetic ancestry, and the future of race. With the completion of the Human Genome Project, it was believed that on a large scale the elimination of racism would result, but as we have observed, that is not the case.

3.3 Racialized Medicine & Cultural Reaction

The Human Genome Project, although originally intended to seek protection from the mutagenic effects of radiation, should have dissipated all belief of there being significant differences between ‘races’. Even more shockingly, this profound concern with race appears to linger even more strongly in the United States. I am not suggesting that Americans would ever dare to repeat horrifying research such as the Tuskegee Experiments, but I am suggesting that America pessimistically concentrates more on the 0.1 percent difference rather than embracing one another in the 99.9 percent similarities we share genetically. An example of such as case can be observed in Jonathan Kahn’s

Race in a Bottle: The Story of BiDil and Racialized Medicine in a Post-Genomic Age, which covers the development of BiDil (first encountered in March 2002). BiDil was developed in the 1980s as a single pill to treat heart failure for any patient prescribed, but because of two patents, it was ultimately decided to treat “black” patients.\(^{34}\) Kahn says that the drug is effective for males of every ‘race,’ but researchers oversaturated a small correlation that suggested it more greatly benefited perhaps a select group of their African American participants compared to any other group of people. The drug is now marketed as a race-specific medication. However, there is no medical evidence to compel companies to market the drug as race-specific.\(^{35}\) This brings about many questions, such as what is racialized medicine? According to pharmaceutical companies, it is the optimistic beginning to a route of personalized medicine.\(^{36}\) The entire marketing model for BiDil, including its request for FDA approval, has been framed by the appeal of personalized medicines.

Another particularly striking example of using race to differentiate a product in the marketplace is advertisement of the beta-blocker nebivolol (marketed as Bystolic by Forest Laboratories), which is common prescribed in the United States to treat hypertension and other cardiovascular conditions, such as heart failure.\(^{37}\) Why is this important? Kahn believes Forest and its surrogates wrongly emphasized two aspects of Bystolic’s profile to distinguish the drug from the eighteen other beta-blockers on the market. Firstly, nebivolol was believed to have had fewer side effects, such as fewer

\(^{34}\) Jonathan Kahn, Race in a Bottle: The Story of BiDil and Racialized Medicine in a Post-Genomic Age (New York: Columbia University Press, 2013): 4

\(^{35}\) ibid, p. 49


\(^{37}\) Ibid, 112.
instances of erectile dysfunction, and secondly, it worked remarkably well in African Americans. Much like BiDil, Forest Laboratories based their claims of Bystolic upon a race-specific trial, meaning it was exclusively tested in a trial of self-identified African Americans.\textsuperscript{38} Race can admittedly be useful whilst attempting to understand how diseases manifest in certain populations of individuals with relative sets of genetic markers, but unfortunately the research within medical communities are being distracted by identifying the social construct ‘race’ rather than focusing on the underlying causes of health disparities. Drugs such as BiDil and Bystolic are sending dangerous messages to the public about race being somehow genetic, and it’s hindering the opportunities that were set forth after the completion of the Human Genome Project.

If Kahn is indeed correct about the lack of evidence in racialized medicine, why does racial profiling persist in medical research? In Kahn’s article, “The Troubling Persistence of Race in Pharmacogenomics,” he proposes two reasons. Firstly, Kahn says, “Race is evolving into a ‘residual category’ that is being used to explain any variation in drug response that is not captured by genetics.” Anywhere in the near future, it is certain that genetics will never certifiably explain one hundred percent of variable drug response. There are far too many factors to consider, whether it be environmental, dietary, or behavioral factors, or perhaps all three in regards to drug response. Geographical origin is only the beginning. Secondly, Kahn says, “there is an inertial force to race in biomedicine.”\textsuperscript{39} What does this mean exactly? Kahn is being quite literal; once

\textsuperscript{38} Ibid, 113.
researchers are introduced into a “…conceptual system for evaluating biological
differences, it very difficult to dislodge race.” Even when Montagu and Lewontin’s idea
that ‘race’ was a social concept was widely accepted, there were still dissatisfied people
who believed races could be justified by scientific evidence. Ironically, ‘race,’ as a
scientific concept, has not disappeared as many believed would result from the Human
Genome Project, although it continues to lack a firm scientific foundation. As humans,
we have an inherent, but unnecessary, desire to categorize things as we see them, and in
this case, there are too many factors to be accounted for. Race may be irrelevant, but it
iscertainly taking the world by storm. “Dr. Francis Collins, the director of the National
Institutes of Health, put it, “a weak and imperfect proxy” for genetic differences. But it is
also a familiar concept — and asking people what race they are is substantially cheaper
than genotyping them.”40 In Science Friday’s news program, “Does your Genome Belong
to your Family, Too?,” which we previously discussed, it is reported that the first human
genome cost about 2.7 billion dollars whereas in present day an entire genome sequence
could cost upwards of one thousand dollars.41 Although pharmaceutical companies have
somewhat accomplished masking racialized medicine as personalized medicine, perhaps
having a race-specific medication has given individuals the empowerment of believing

41 Intagliata, Christopher, prod. "Does Your Genome Belong to Your Family, Too?"
they are doing what they can in the extent of personalized medicine, without paying the one thousand dollar figure for such a service? It is important to consider in the future of personalized medicine individuals would not be required to have their genome sequenced since personalized medicines would be developed to benefit specific haplogroups rather than one particular person.

Is racialized medicine here to stay in the United States? How is society reacting to this push for race-specific drugs/ racialized medicine? In Katrin Weigmann’s “Racial Medicine: Here to Stay?, ” she says, “Adverse drug reactions are an underestimated but serious problem in medicine, and are the fourth leading cause of death in the USA after cancer, coronary heart disease and stroke (Lazarou et al, 1998). Although any given drug, if prescribed correctly, is beneficial for most patients, it may have unexpected results in a small subset of users.”42 This is quite alarming. Even after the Human Genome Project was completed, research projects are still prevalent today with the hope of identifying genetic variants that would link to the metabolic differences between patients. Several follow-up projects include the notorious HapMap initiative, which has high hopes of identifying and cataloging haplotypes. In the midst of all this optimism, Weigmann says that the one major hindrance in the possibility of an effective system of personalized medicine is that physicians and scientists have “…found a comfortable interim solution: racial profiling.” The first step most physicians take in prescribing an “individualized treatment” usually lies under the assumption that specific traits cluster by race, and in most cases, this is not particularly true. Weigmann also proposes that depending upon

how the data of the HapMap is used, it may reinforce racial thinking rather than 
eliminating it because the HapMap and other genome projects provide the tools to search 
for genetic differences between races (Lee, 2005). Sandra Lee, as quoted by Katrin 
Weigmann, says, “If you look for differences, you find differences. What becomes the 
central question is whether a difference is significant.”43 This desire to explore the 
differences rather than the similarities explains why the fraction of 0.1% is held to be 
significant, although it is not biologically defensible. This presents a larger danger to the 
medical community because scientists might misuse the data indicating genetic 
differences with disproportional frequency between different ethnic groups and 
‘incorrectly extend these results to broader racial groups; thereby strengthening the 
misconception that race is a biological category.’44

**Conclusion**

Whether racialized medicine is here to stay is a mystery. Hopefully there is a 
future for personalized medicine, but I hope that it is not based entirely on ‘race.’

Anthropologists and social scientists from the 1950s onwards attacked the concept of 
‘race’; this attack was strongly supported by work in genetics beginning in the 1970s. 
The Human Genome Project was hailed as even stronger evidence against the scientific 
acceptability of ‘race’. An immediate consequence of the Human Genome Project was 
the advent of ‘personalized medicine’, and this led, ironically, to new claims for 
‘racialized medicine’ in the case of the heart medication BiDil and the beta blocker

43 Ibid.
http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1456889/.
Bystolic. From the evidence presented, it is overwhelmingly understood that the concept of race is not supported by the scientific findings of genomics and dismissals of the concept of ‘race’ from the middle of the twentieth century onward are still valid.

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