

A Critique of Race-Based and Genomic Medicine

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ABSTRACT

Now that a composite human genome has been sequenced (HGP), research has accelerated to discover precise genetic bases of several chronic health issues, particularly in the realms of cancer and cardiovascular disease. It is anticipated that in the future it will be possible and cost effective to regularly sequence individual genomes, and thereby produce a DNA profile that potentially can be used to assess the health risks for each person with respect to certain genetically predisposed conditions. Coupled with that enormous diagnostic power, it will then depend upon equally rapid research efforts to develop personalized courses of treatment, including that of pharmaceutical therapy. Initial treatment attempts have been made to match drug efficacy and safety to individuals of assigned or self-identified groups according to their genetic ancestry or presumed race. A prime example is that of BiDil, which was the first drug approved by the US FDA for the explicit treatment of heart patients of African American ancestry. This race-based approach to medicine has been met with justifiable criticism, notably on ethical grounds that have long plagued historical applications and misuses of human race classification, and also on questionable science. This paper will assess race-based medical research and practice in light of a more thorough understanding of human genetic variability. Additional concerns will be expressed with regard to the rapidly developing area of pharmacogenomics, promoted to be the future of personalized medicine. Genomic epidemiology will be discussed with several examples of on-going research that hopefully will provide a solid scientific grounding for personalized medicine to build upon.

Key words: race-based medicine, BiDil, pharmacogenomics, genomic epidemiology, personalized medicine

Introduction

Taking a broad perspective, Western Medicine generally has sought to deliver the very best and latest health care cognizant of individual and group variability. For example, in recent years clinical trials have been much more attendant to better represent women in light of possible differing health decisions and policies to that of men. So too, race and/or ethnic group has been increasingly addressed for some of the same reasons. Numerous studies and reports have been devoted to the topic of race-based medicine¹⁻⁶.

However, biomedical research as well as clinical practice has endured an apparent uncomfortable and unsettled relationship involving human races. This is understandable given the ubiquitous lack of adequate definition and clarity for applying a workable concept of classification into racial categories. For the most part, medical science has resorted to self-identification and self-reporting in its recruitment for clinical trials. Hence, recruits assigned themselves to already existing categories. It is probable that the general public in the USA has been exposed formally to racial categorization through several

applications including the Federal Census Forms, law enforcement agencies, and health care studies and health delivery services.

Indeed, considering the last of these, mainstream news media continually broadcast the disparities in health care delivery that are based upon race/ethnic group designations, such as White, African American, Hispanic, Asian and others. These categories are found in the Federal Census forms and also are required to be used by biomedical researchers receiving NIH funding¹.

Since assignment to these racial categories is often by individual or personal choice, it can be expected that decisions are made according to a combination of biological variables and sociocultural characteristics. Hence, the conjoined label »race/ethnic group« seems entirely appropriate, at least in this instance. As a consequence, throughout much of the biomedical research literature, race and ethnic group appear to have equal and nearly interchangeable status. A major question that has been raised in that literature, and serves to focus this paper, is

whether disparities in health care delivery and resultant differences in treatment success are only due to socioeconomic variables or are there possible underlying biological/genetic influences that in part explain both variation in susceptibility to certain diseases and the efficacy and safety of administered pharmaceuticals or drugs?

Recently, the US Federal government has recognized that this question should be addressed from a sound scientific approach that attempts to sort out the relative contributions of environment and heredity by establishing the NIH Center to Study Genomics and Health Disparities (NCSGHD). Whether this particular effort will be successful is, of course, yet to be determined. However, considering the state of affairs with regard to current pharmaceutical research based upon racial categories, there is good reason to be skeptical about any major breakthroughs in dispelling health care disparities.

Race-Based Medicine

Pharmaceutical companies routinely have utilized racial categories in investigating drugs that can be tailored toward selected populations, and thereby has been referred to as race-based therapeutics⁷ or race-based medicine⁸. Their aim is to develop and market new medicines to treat diseases that disproportionately affect certain groups or races. Their concept of race presumably is founded on self-identification or self-reporting but is purported to be indicative of the genetic variation and ancestry of the self-defined group, for example, African Americans. For this purpose, race is thus viewed as a proxy or placeholder for yet to be determined genetic information and specification. It might be mentioned that this commercial interest in targeted drug development by pharmaceutical companies is not restricted to race but to several demographic divisions including gender and age groups.

Hence, it is perhaps reasonable to surmise that drug companies fundamentally are driven by a profit motive along with their benevolent search for targeted medicines, which, of course, is an acceptable business practice under a capitalistic economy. However, there is an unsavory instance of one drug that appears to have followed this tailor-made development and marketing pathway but unfortunately was subjected to very questionable science^{7,9–10}. This is the case of BiDil. BiDil is taken in pill form that actually is composed of two generic drugs, hydralazine and isosorbide dinitrate. Its primary physiological function is vasodilatory (expands blood vessels) and it has been shown to have clear therapeutic value in the treatment of congestive heart failure. Therefore, it was entirely legitimate to claim that BiDil was efficacious. However, it is the developmental pathway itself that led to its approval as a targeted drug for African Americans that raises troubling questions and hopefully also raises some caution flags⁹.

A synopsis of the BiDil case can begin by noting that about 20 years ago initial studies were done to investigate the effectiveness of vasodilating drugs on congestive

heart failure. The two drugs used separately in the research were hydralazine and isosorbide dinitrate. In one trial, V-HeFT II, these two drugs were compared with an ACE inhibitor that proved to be more effective, and this placed the vasodilating drugs as just a backup treatment for patients who didn't do well on ACE inhibitors. Not to be deterred, the principal investigator persisted research efforts and was able to secure a patent in 1987 that defined the method for using the two vasodilating drugs, hydralazine and isosorbide dinitrate, that were then combined in the 1990s into the single pill called BiDil. A first attempt to receive FDA approval for BiDil was rejected because the initial drug trials did not meet FDA standards⁹. This rejection occurred in 1997 or ten years after patent approval and thus halfway through patent rights protection period. It is surmised that the pharmaceutical company that supported the BiDil research and held the patent rights backed out of the project at this time because of the FDA rejection and shortened patent life, both of which would have adversely impacted profitability of BiDil⁹. Apparently, again not to be deterred, the principal investigator went back to the original V-HeFT I and »data mined« the results that showed vasodilating drugs were especially effective in African Americans, at least in the 49 patients in the trial. This positive finding was sufficient for him to attain another patent in 2000, which then extended protection from competition until 2020. Shortly thereafter, he engaged a second drug company to underwrite another clinical trial known as African-American Heart Failure Trial (A-HeFT)^{7,9}.

In the A-HeFT study involving a self-identified African American sample of 1050 patients, BiDil was found to reduce fatality by 43% and to decrease hospitalization by 39%, when compared to the placebo group. In fact, the A-HeFT trial was stopped early because of such positive results. The next most significant step was when BiDil received US FDA approval in June of 2005 and was defined as race-specific, the first such drug ever to receive that designation⁹.

Needless to say, this action generated controversy along with some support from certain quarters. Issues that surround commercial interests of drug companies are certainly profound and warrant a continued vigilance against what in the case of BiDil appears to place profit in the forefront. For instance, in order to properly address the criticism that BiDil might be just as effective in non African Americans would require a full prospective study that compares adequate samples of all so-called racial/ethnic groups. This would be a costly, time-consuming endeavor that probably will not attract copious drug company investment. Of course, »off label« use of BiDil for a patient of any ancestral background can be done with no additional testing. In the end, it is fully expected that the development of drugs will continue to follow an economic benefits model and a set of marketing strategies that enhance the financial status of companies, but hopefully will also adhere to reasonable profit motives, and most importantly to sound scientific practices.

Beyond troubling commercial maneuvers, there are numerous additional faults within the BiDil case that essentially rest upon compromises of scientific integrity. At this point we can generalize to offer a critique of race-based medicine overall that may have explicit application in BiDil but has been implicated throughout medical research and practice both before 2005 and persists up to the present time¹¹.

Critique of Race-Based Medicine

Race as a social construct

There has been an ongoing and spirited discussion of whether human races are biologically real or are entirely social constructed^{4,12}. In the latter view, based on the ideology of postmodernism, race does not have objective reality, or more succinctly this notion sometimes translates as races do not exist. This statement certainly is supportable if it refers to the outmoded typological thinking that defined human races as mutually exclusive, genetically homogenous (»pure«) categories. On the other hand, acknowledging numerous constraints on any classificatory use of race, does allow restricted definitions of biological/genetic races to be made operational, albeit with dubious scientific value or practical application. Additionally, the act of classifying races is possibly less problematic than any attempts to satisfactorily assign all individuals to those defined race categories.

However, even if the concept of biological race classification is accepted formally within selected academic circles, this does not remove the major obstacles facing race-based medicine. Furthermore, race labeling could quite possibly do harm in the eyes of the public that often wrongly equates races with invariant genetic determinism. A corollary to this public opinion is that race-specific labeling of medicines, directly infers that certain diseases are therefore race-specific and inevitable, and this could lend itself to possible stigmatizing and discrimination. Insurance coverage is one area where a discrimination concern appears to have legitimacy⁹.

Race category assignment

As already noted, assignment to race/ethnic groups in medical research and practice is usually done through personal choice and self-reporting. This means that categories may contain individuals who have little or possibly even no direct ancestral connection to those categories. Perhaps of some interest, the US Census Bureau has permitted persons to identify themselves into more than one race category, and in actuality this is the situation of a large portion of the US population. With respect to race-based medicine, because of a lack of clarity or control of the genetic makeup of and variation within categories, any pretext of race serving as an adequate proxy for genetic ancestry is unfounded^{3,13}.

Genetic basis of disease

Given the above limitation, it seems fair to conclude that all medicines that are deemed race-specific do not necessarily match up with underlying genetic causality or even predisposition for the disease being treated or the drug dose being regulated. Later in this paper the field of pharmacogenomics will be discussed, which does attempt to associate precise nucleotide sequences with specific diseases. However, race-based medicine by itself does not do this. More particularly, BiDil is not a pharmacogenomic drug, it is not targeted to a detectable genetic variant.

Genetic variation within and differences between so-called racial groups

A fundamental flaw of race-based medicine is a failure to fully appreciate that a large degree of genetic variation that exists within self-reported racial groupings, in part due to the historical mixing among US populations, but more importantly as an expected condition of any reasonably large demes or gene pools. This flaw parallels historically earlier misguided attempts to classify biological races as homogeneous (»pure«) and distinct. As a consequence of genetic variation among individuals, prescribing a race-specific drug might increase the probability of efficacy but by an unknown degree. There is a final matter to consider with regard to the lack of knowledge of variability at the genetic level, and this has to do with gene expression related to gene × gene and gene × environment interactions¹³. This is more appropriately taken up when we consider the next topic of pharmacogenomics.

In summing up this section, it must be said that the controversy stemming from the marketing of BiDil as a race-specific drug might well be resolved through concerted research to determine what if any gene variants are associated directly with individual, not racial group, responses to the use of this treatment in congestive heart failure. That, in essence, is the direction pharmacogenomics claims to be moving toward.

Pharmacogenomics

A major aim of pharmacogenomics is to ascertain a genotype association with pharmacology in order to provide a more personalized drug treatment. As would be expected much of the research in this field is carried out within or with the support of pharmaceutical companies¹⁴. However, it can be pointed out that one of the US Federal NIH agencies, the National Institute of General Medical Sciences, hosts the Pharmacogenetics Research Network (PGRN) that states a vision – To lead discovery and advance translation in genomics in order to enable safer and more effective drug therapies¹⁵. Its mission is to promote innovative research in pharmacogenetics and also serve as a clearing house for disseminating knowledge about the impact human genetic variation has upon drug responses, for example, results of genotype associations noted above.

The usual research method for determining genotype associations is through sequencing of Single Nucleotide Polymorphisms or SNPs. One such study is that of Rafael, et al.¹⁶ in which they were able to show, among other findings, that efficacy and safety of different cancer treatments varied across race/ethnic groups according to SNP polymorphisms. SNP sequencing methodology also, of course, applies to initially establishing an association or marker between a particular condition or disease entity and a specified gene variant or locus, which when discovered subsequently can be tested against various drug treatments, as was done in the aforementioned cancer study¹⁶.

Multiple laboratory strategies have been employed ranging from whole genome DNA sequencing for SNPs down to RNA transcriptomes, the later method being more efficient and less costly. To expedite this approach, the National Human Genome Research Institute (NHGRI) has undertaken an effort to generate a haplotype map (HapMap) of the human genome that will consist of commonly occurring SNPs that define a rather smaller number of ancestral haplotypes. Whole genome sequencing does generate a very large amount of genetic variability results that only roughly correlates with designated race/ethnic groups.

On the other hand, once particular SNP allele associations have been ascertained they may be subjected to race/ethnic group distribution comparisons, as in some of the earliest reported associations for hemochromatosis, sickle cell anemia and cystic fibrosis. However, these broad group associations could hardly be considered very direct and so far have not led to successful development of personalized drug treatments for any of them.

One of the more intriguing associations has been found with respect to an ethnic/race group distribution of the ApoE4 gene variant that is a risk factor for Alzheimer disease. The ApoE4 variant ranges in frequency of 9% in Japanese to 19% in African Americans, yet the actual risk for expressing Alzheimer disease is reversed to show a relatively lower six times increase for African Americans, while in Japanese the risk markedly increased to 35 times¹⁷. Genetic or environmental modifiers of the gene were implicated in this finding. Clearly, possessing the SNP variant associated with a particular condition does not necessarily predict the outcome. Rather, SNPs might better be viewed as evidence of a genetic predisposition that may or may not be realized depending upon a number of factors, most importantly gene x gene and gene x environment interactions¹³, and also of significance to note here is an ever-increasing research interest in how epigenetics affects gene expression.

A further consideration is that knowledge of a particular genetic risk profile, such as presence of the ApoE4 marker, may be unsettling for some, especially those in their early adult years, due to the current inability to offer much in the way of treatment for Alzheimer disease.

Another caution flag was raised in a report by Rotimi and Jorde (2010)¹¹ that showed ethnic group variation with an HLA SNP and hypersensitivity to the drug

Abacavir. Abacavir is administered in the treatment of HIV and AIDS. Negative reaction to this drug, in the form heart attacks, is found in European Americans at an average rate of nearly 6%. In two African populations the hypersensitivity SNP allele did not appear at all in the Yoruba from Nigeria, but showed an average frequency of 13.6% in the Masai of Kenya. In this case, race/ethnic group labels such as African or Black would not be at all meaningful with regard to informed genomic understanding or in medical practice. It is important to note, however, that the US FDA recommended that all groups be screened for the HLA hypersensitivity allele irrespective of their ethnic affiliation or ancestry¹¹.

Genomic Epidemiology

These kinds of problems in genomic medicine are prospectively resolvable as continued research efforts are made especially in the area of genomic epidemiology. Major initiatives along this direction are being taken by the NIH funded National Human Genome Research Institute (NHGRI) and the Genome-Wide Association Study (GWAS) both of which are scanning the entire genome in search of SNP markers that are significantly associated with diseases and conditions in individuals. Positive results or »associations« thus far have been found for Type 2 diabetes, Parkinson's disease, heart disorders, obesity and prostate cancer¹⁸.

As an update regarding Alzheimer disease, new research has gone beyond the ApoE4 marker to recently discovering several additional SNPs through genome-wide association studies showing putative risk genetic variants or candidate genes¹⁹. Hopefully, this work will lead to better diagnosis and possibly even eventual preventative treatment measures for Alzheimer disease²⁰.

As already noted, genotype associations do not necessarily identify the causal gene, a search that may be facilitated by conducting exome screening rather than the whole genome. This approach could lead to highly informative candidate gene markers, or »driver mutations« in the parlance of oncogenetics²¹. Another of these large projects within NHGRI is that of the 1000 Genomes Project, which to date actually has sequenced over 2000 genomes, and has been making their data readily available online, along with a tutorial for how to access and use the data sets²².

Still another project under NHGRI sponsorship is underway at Howard University, in Washington, DC²³. A main component of this research deals with Africans and African population descendants who were involuntarily dispersed to other world areas including the Caribbean and USA. The focus of this project is on genetic epidemiology in a multigenerational search for associations with diabetes, hypertension and obesity, all conditions that have adversely impacted the health of peoples of African descent. Additional projects at Howard University are directed toward a genetics x environment study of asthma in African Americans and also a genetic epidemiology study of breast cancer in African American women²³.

A final project to mention here is housed at the MalariaGEN Resource Centre, University of Oxford, UK²⁴. Four initiatives have been defined, including; a) genome-wide association with severe malaria, b) genetic determinants of immune response, c) genetic diversity in malaria endemic regions, and d) genetic linkage studies of resistance to malaria. All of these hopefully will shed some light on the vexing issue of genetic variation that is found among individuals who are assigned or assign themselves to presently defined racial categories. This MalariaGEN project, as well as others noted above, are also addressing ethical considerations with respect to genome research, particularly the vexing problem of informed consent.

Looking Ahead

For the foreseeable future it would appear that race-based medicine will continue to find an awkward and at times, contestable, place in medical research and in clinical practice. An optimistic viewpoint is that race categories can prove meaningful in health care and research with careful training of medical personnel^{25,26}. However, as a contrary move, an advisory has been made by opponents of race-based medicine to abstain from participation in racial category studies and to refrain from the use of available race-specific drugs²⁷. Perhaps this move will encourage an even more rapid transfer of genetic identity from that of a dubious race label to individual genomes. Along that direction, Crews and Gerber (2008) suggested that race categories used in medicine be replaced by already well-studied Ancestry Informative Markers (AIMs) but referenced to an individual level and thereby referred to as IGA (individual geographic ancestry)³. In a related line of inquiry, commercial enterprises are offering individual DNA testing of percentages of ancestry for ancestral populations, such as BioGeographical Ancestry (BGA)²⁸. It is not yet evident whether these DNA lineage approaches have been incorporated directly into medical research or practice.

Regarding the cost involved of screening, it would appear that genomic medicine will be taking full advantage of rapidly advancing and cheaper methods for DNA and RNA sequencing and testing (including high throughput and nanotechnology), and will become increasingly dependent on these methods for diagnosing and protecting patients in terms of drug appropriateness and dosage level. Pharmacogenomics as a mainstay of personalized medicine heralds in a potentially safer and more effective delivery of health care to the individual.

However, it is very important to appreciate that neither race-based nor genomic medicine will replace the time-honored and highly successful other diagnostic and therapeutic insights available to the clinician, derived from such factors as gender, age, lifestyle and family history. From a clinical perspective, these constitute risk factors. To single out one of these, family history has in fact been found in a comparative framework to be more informative than personal genomic screening in a sub-

study of the Framingham Genetic Research Study dealing with atrial fibrillation²⁹, as well as in a risk assessment for breast, colon and prostate cancer³⁰, both study results reported late this past year.

Recommendations

The major problems discussed in this review regarding the diagnosis and treatment plans for conditions of a genetic nature might be effectively addressed through concerted educational efforts and professional development. There are several partially overlapping and integrated avenues to this approach, such as more intensive training of physicians in genomic medicine, expanded inclusion of genetic counselors in patient interactions between physicians and patients, an introduction of patient advocates who are able to assist persons in making decisions more directly than is customarily expected of or ethically permitted by genetic counselors, and to locate support groups and online resources that can provide useful information and guidance.

The complexity and crush of ever-expanding amounts of genetic information may well be overwhelming to most persons seeking medical assistance, and probably even to a high proportion of health professionals as well. Yet intensifying genomic training into a medical school curriculum would impose a substantial burden and is not likely to occur. Thus, the role of genetic counselors should be much more routinely integrated into the care of patients and parents of patients dealing with genetically influenced conditions. Patient advocates fill a somewhat different niche to that of the genetic counselor. Traditionally, and for good reason, ethical standards expected genetic counselors to remain nondirective in their role of providing complete, accurate, and up-to-date genetic information to clients³¹. In more recent years the non-directive stance has been challenged by a psychosocial approach³² that seems to establish a more active and interactive relationship between the counselor and client. Here is where a patient advocate, who would be fully prepared to process and impart the complexities of genetic information with clients, would assist by offering professional advice, but clearly without coercion or imposing their own values, and in consort with a physician's guidance. Other health professionals, including nurses and physician associates also might be anticipated to carry some of the load imposed by genomic medicine applications. Finally, presuming that shared experiences do offer empathetic consolation, prospective clients of genomic medicine might well benefit from interacting with online support groups. Furthermore, taking full advantage of current technological developments, web-based resources could provide very personal guidance through the labyrinth of genetic diagnoses and prognoses that may or may not have any or even adequate courses of treatment.

These recommendations essentially deal with educating both professionals and public regarding the promise and practice of genomic medicine. Just as importantly, it

is necessary to reign in the overstating of immediate benefits derived from personal genomics. A viewpoint expressed recently by a physician and geneticist³³ is that expanding genetic knowledge in time will be very significant in terms of understanding the hereditary nature of some diseases, and thereby enhance diagnostic power, while providing potentially valuable information to persons who might be at high risk for developing certain dis-

eases themselves, or for prospective parents to guide in their family planning decisions. He added, however, that at the present time personal genetic information can play only a small role in the overall picture for assessing individual health³³. Unfortunately this suggests that race-based medicine will persist in some fashion into the future prior to a transition to a more realized practice of genomic-based personalized medicine.

REFERENCES

1. BRAUN, L, FAUSTO-STERLING A, FULLWILEY D, HAMMONDS EM, NELSON A, QUIVERS W, REVERBY SM, SHIELDS AE, PLoS Med, 4(9) (2011) 1. — 2. MONTOYA MJ, Curr Anthropol, 22 (2007) 94. — 3. CREWS DE, GERBER LM, Genes, Geographic Ancestry, and Disease Susceptibility: Applications of Evolutionary Medicine to Clinical Settings. In: TREVATHAN WR, SMITH EO, MCKENNA JJ (Eds): Evolutionary Medicine and Health. New Perspectives (Oxford University Press, Oxford, 2008). — 4. SCHWARTZ RS, N Engl J Med, 344 (2001) 1392. — 5. BURCHARD EG, ZIV E, COYLE N, COMEZ SL, TANG H, N Engl J Med, 348 (2003) 1170. — 6. TISHKOFF S, KIDD K, Nat Genet, 11 Suppl (2004) 521. — 7. BLOCHE MG, N Engl J Med, 351 (2004) 2035. — 8. YU JH, GOERING S, FULLERTON SM, Camb Q Healthc Ethics, 18 (2009) 57. — 9. KAHN J, Available from: URL: <http://www.scientificamerican.com/article.cfm?id=race-in-a-bottle> (2007). — 10. KAHN J, Soc Stud Sci, 38 (2008) 737. — 11. ROTIMI CN, JORDE LB, N Engl J Med, 363 (2010) 1551. — 12. ANDREASEN RO, Phil Sc, 67 (2000) 653. — 13. KITTLES RA, WEISS KM, Ann Rev Genomics Hum Genet, 4 (2003) 33. — 14. The Pharmaceutical Research and Manufacturers of America (PhRMA) Website, Available from: URL: <http://www.phrma.org>. — 15. NIH Pharmacogenomics Research Network Website, Available from: URL: <http://www.pgrn.org>. — 16. ROFAIEL S, MUO EN, MOUSA SA, Pharmacogen Pers Med, 3 (2010) 129. — 17. BURCHARD EG, ZIV E, COYLE N, GOMEZ SL, TANG H, KARTER AJ,

MOUNTAIN JL, PÉREZ-STABLE EJ, SHEPPARD D, RISCH N, N Engl J Med, 348 (2003) 1170. — 18. Genome Wide Association Studies Website, Available from: URL: <http://www.genome.gov/gwastudies>. — 19. BETTENS K, SLEEGERS K, VAN BROECKHOVEN C, Hum Mol Genet, 19 (2010) 4. — 20. KIM S, SWAMINATHAN KS, SHEN L, Neurol, 76 (2011) 69. — 21. STRATTON MR, CAMPBELL PJ, FUTREAL PA, Nature, 458 (2009) 719. — 22. 1000 GENOMES PROJECT WEBSITE, Available from: URL: <http://www.1000genomes.org/>. — 23. HOWARD UNIVERSITY NATIONAL HUMAN GENOME CENTER WEBSITE, Available from: URL: http://www.genomecenter.howard.edu/research_programs.htm. — 24. Malaria-GEN Resource Centre Website, Available from: URL: <http://www.malaria-gen.net>. — 25. TASHIRO CJ, Pediatr Nurs, 31 (2005) 208. — 26. TASHIRO CJ, Pediatr Nurs, 31 (2005) 305. — 27. Macalester College Environmental Studies Department Website Available from: URL: <http://www.macalester.edu/environmentalstudies>. — 28. Available from: URL: <http://www.ancestrybydna.com>. — 29. BEALS JK, Available from: URL: <http://www.medscape.com/viewarticle/734249> (2010). — 30. LEACH CE, Amer Soc Hum Gen Meeting Abstract, 147 (2010). — 31. BENNETT RL, HAMPPEL HL, MANDELL JB, MARKS JH, J Clin Invest, 112 (2003) 1274. — 32. WEIL J, J Genet Couns, 12 (2003) 199. — 33. EVANS JP, Sc News, 179 (2011) 36.

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KRITIKA RASNO-BAZIRANE I GENOMSKE MEDICINE

SAŽETAK

Nakon sekvenciranja kompletnog ljudskog genoma, znanstvenici su stavili naglasak na otkrivanje genetičke baze nekoliko kroničnih zdravstvenih pitanja, većinom s područja tumora i kardiovaskularnih oboljenja. Anticipirano je kako će u budućnosti biti moguće i novčano prihvatljivije redovito sekvencirati individualni genom, te shodno tome isproducirati DNA profil koji se potencijalno može koristiti za otkrivanje zdravstvenih rizika svakog pojedinca u odnosu na njegovu potencijalnu genetičku predispoziciju. Praćeno s tom snažnom dijagnostikom, jednako će ovisiti i o jednako uložnim naporima razvoja personaliziranih tretmana liječenja, uključujući i farmaceutsku terapiju. Početni pokušaji tretmana bili su usmjereni ka povezivanju učinkovitosti i sigurnosti lijeka za pojedince određene skupine na osnovi njihovog genetičkog naslijeđa ili pretpostavljene rase. Glavni primjer je lijek pod nazivom BiDil, prvi koji je odobren od US FDA za izričitu upotrebu srčanih bolesnika Afričkoameričkog porijekla. Ovo rasno bazirano liječenje s pravom je podleglo kritici, posebno na etičkoj razini koje je dugo vremena bilo poput kuge, te se koristilo za neopravdane klasifikacije unutar ljudske vrste. Ovaj rad želi odrediti rasno bazirana medicinska istraživanja i praksu u svjetlu cjelokupnog razumijevanja ljudske genetičke varijabilnosti. Dodatna zabrinutost iskazana je u odnosu na brzo rastuće područje farmakogenomike – budućnosti personaliziranih lijekova. Genetska epidemiologija biti će diskutirana kroz nekoliko primjera tekućih istraživanja. Ovaj rad će procijeniti rasno bazirana medicinska istraživanja i praksu u svjetlu detaljnijeg razumijevanja ljudske genetske varijabilnosti. Dodatna pažnja posvećena je ubrzanom razvoju području farmakogenomike, koja se smatra budućnošću personalizirane medicine. Genomska epidemiologija diskutirana je kroz nekoliko primjera kontinuiranog istraživanja koje će pružiti solidne znanstvene temelje za izgradnju personalizirane medicine.