

The influence of health disparities on individual patient outcomes: What is the link between genes and environment?

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A 20-year-old African American woman with diabetes mellitus has been hospitalized many times for asthma. She has seasonal allergic rhinitis. Many of her family members have allergic rhinitis and asthma, and an uncle recently died of asthma. She has missed several appointments and returns today having run out of medications. When she reaches the doctor's office, she is asked to reschedule because she is late.

The patient has a new job working in a cafeteria in a distant suburb that requires her to take 2 buses with 1.5 hours of travel time each way. Her job provides health insurance but no coverage of medications. She has been unable to make recent doctor's appointments because her doctor only has hours from 9 to 5, and the patient is worried about taking time from her job, which she has had for only a few months. She is late for this appointment, for which she took a vacation day, because one of the buses did not arrive on schedule.

Are the primary influences on this patient's asthma morbidity related to physical or social environmental exposures or genetic susceptibilities? We are told the patient's uncle died of asthma. Has she inherited very severe asthma? Does she have an inherited polymorphism of a gene or genes that predispose her to severe asthma and also diabetes? Is her asthma a result of exposures to allergens and irritants, such as pollution from living in a poor neighborhood next to a highway? Is her morbidity a result of her socioeconomic status that results in an

Abbreviations used

ED: Emergency department

IOM: Institute of Medicine

SIRE: Self-identified race or ethnicity

inflexible work schedule and inadequate medical coverage often characteristic of low-paying jobs? Or is it the result of the physician's policies, which include turning away patients who are late? In other words, what are the relative contributions of genetic susceptibilities, exposures of the physical environment, and the social and cultural climate to the patient's asthma state? This case highlights the importance of the excellent perspectives in this issue dedicated to health disparities, one with an epidemiologic perspective, but one focused on patients' experiences in the health system,¹ and the other on genetics.² This review will provide the background for these perspectives, beginning with the relevant demographic trends.

RELEVANT DEMOGRAPHIC TRENDS IN THE UNITED STATES RELATED TO ASTHMA

The cultural and ethnic diversity of the United States is increasing (Fig 1). By 2050, it is estimated that ethnic minorities will comprise about 50% of the US population.³ Limited resources for health care accompany our growing population and expanding diversity.

Not only does the prevalence of diseases like asthma differ among population groups but so does its morbidity and mortality. As indicated by Joseph et al¹ in the current issue, the prevalence of asthma is higher in African Americans and other minority groups. More striking than the prevalence differences in such patient groups are the differences in trends in emergency department (ED) visits and hospitalizations. According to the National Hospitalization Discharge Survey, hospitalization and ED visit rates for asthma have been reasonably stable from 1980 through 2002, even during a period of increasing asthma prevalence (Fig 2).⁴ However, the

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The racial and ethnic diversity in the US is increasing

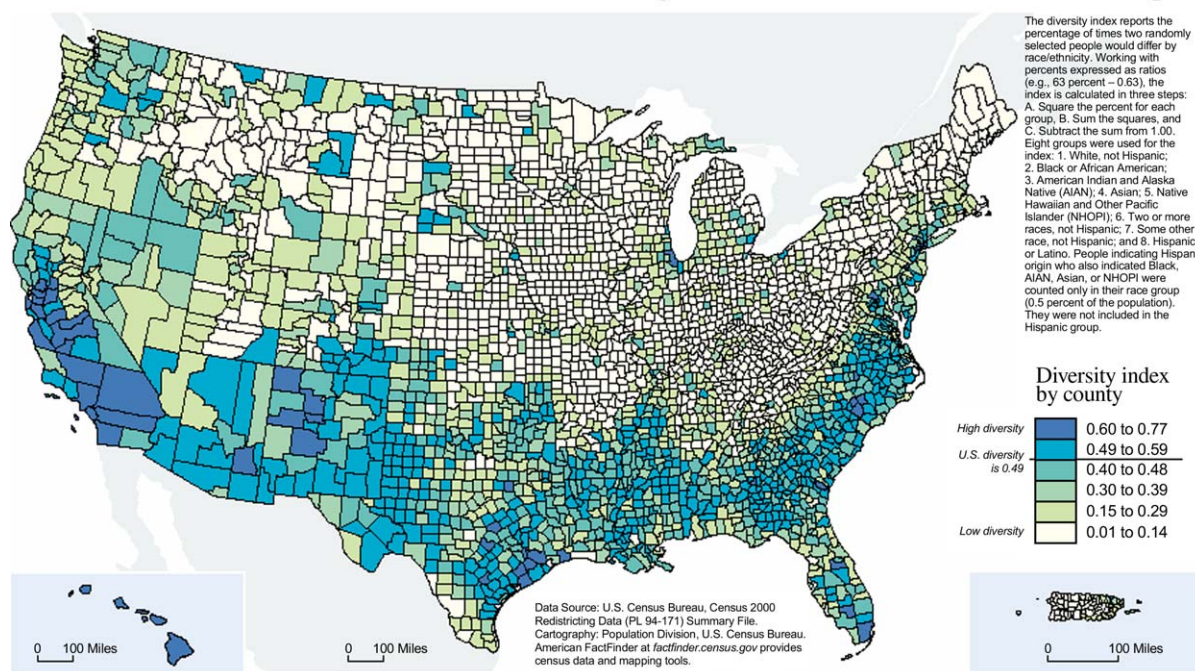


FIG 1. The diversity index for year 2000 by United States county, from the United States National Census Bureau. The colors indicate the percentage of times any 2 randomly selected individuals will be of different race-ethnicity. The 2 darkest colors indicate a probability of 50% or more. (U.S. Census Bureau, Census 2000 Redistricting Data [PL 94-171] Summary File. Cartography: Population Division, U.S. Census Bureau).

hospitalization rate for blacks greatly exceeds that for whites. For example, in 2002, it was more than 3 times that for whites, and the ED visit rate was almost 5 times higher than for whites. These differences are proportionally larger than the milder increase in prevalence in blacks over whites (Fig 3). Although absolute mortality for asthma is low, similar patterns of excess mortality are seen for blacks. The larger differences in hospitalization and mortality suggest that there might be differences in health care received by such patient groups.

DIFFERENCES IN HEALTH CARE ACCORDING TO RACE-ETHNICITY

Because of these and similar findings in many areas of medicine, Congress charged the Institute of Medicine (IOM) to “assess the extent of racial and ethnic differences in healthcare not otherwise attributable to known factors such as access to care, and evaluate potential sources of racial and ethnic disparities.”³ The resulting report, “Unequal Treatment,”³ published in 2003, includes a meta-analysis of relevant literature. The authors found differences in health care provided to patients on the basis of their race-ethnicity. That is, some of the poorer health outcomes seen in minority groups could be attributed to differences in health care.

There are many studies, including some relating to asthma, that support the finding of unequal treatment of minority patients.^{3,5} Of particular interest is a study by Phelan et al,⁶ who found wider health disparities in diseases for which there are proved therapies, providing support for the notion that those who are more affluent and powerful are more able to avail themselves of the best health care. Joseph et al¹ make the important point that “within class strata, measures of income and education seldom translate into the same level of wealth for minorities compared to whites.” Thus racial and ethnic disparities lead to socioeconomic disparities and compound the difficulty of minority patients in accessing the best care.

Joseph et al¹ also stress the importance of approaching the elimination of disparities by targeting primary, secondary, and tertiary prevention. All of these preventive efforts require satisfactory communication between patient and provider.

The IOM report places great emphasis on the importance of the doctor-patient relationship, what has always been considered the essence of the art of medicine, for eliminating health disparities. It acknowledged that patient attributes, such as refusal of treatment and poor adherence, might contribute to health disparity. However, the authors concluded that these patient behaviors might contribute to poorer health outcomes as a result of “a poor culture match between minority patients and their providers,

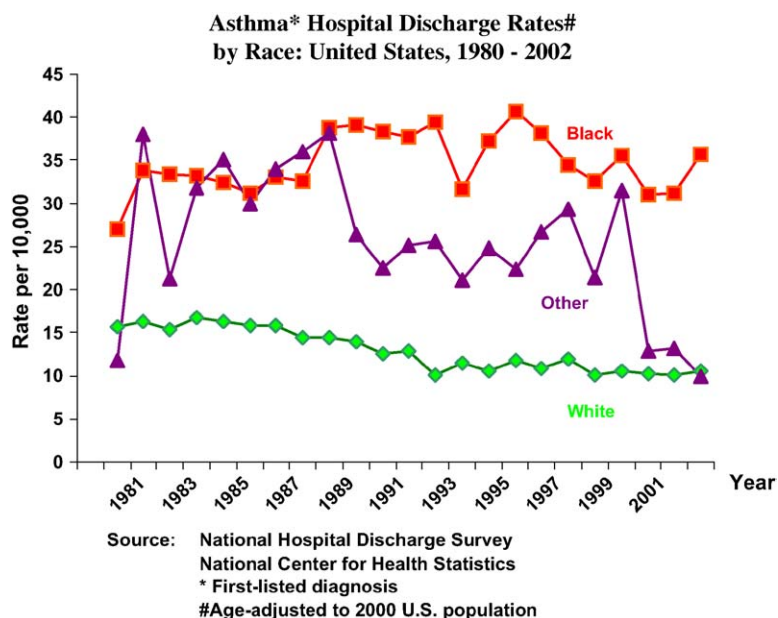


FIG 2. United States asthma hospital discharge rates per 10,000 population comparing blacks, whites, and “others” from 1980 through 2002, from the National Hospital Discharge Survey of the National Center for Health Statistics. The rate of discharge for blacks is about 3 times that for whites. The rates for the “other” group (other than black or white) are more variable because of the relatively smaller size of the population.

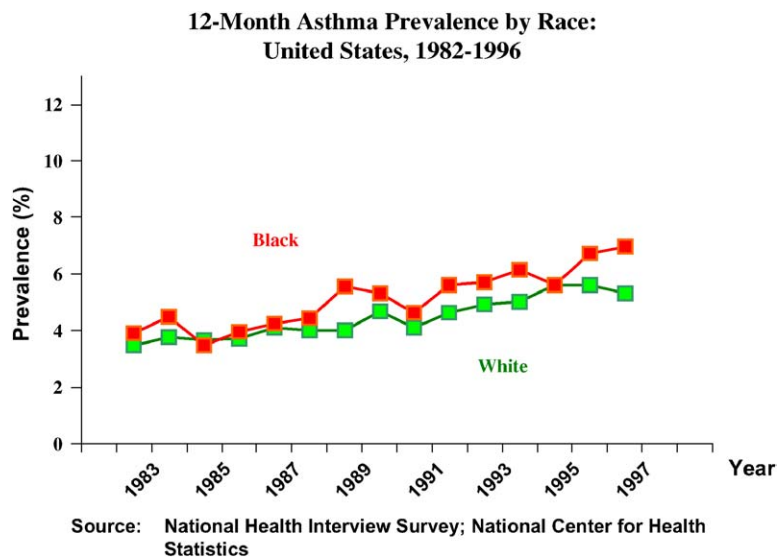


FIG 3. Twelve-month asthma prevalence in the United States per 10,000 population by race from 1982 through 1996. In 1997, the survey was redesigned, and the questions were changed.

mistrust, misunderstanding of providers instructions, poor prior interactions with the healthcare systems, or simply from a lack of knowledge of how to best use health care services.”³ Thus the report concluded that patient-related attributes are not a major source of health care disparities but were mostly part of a response to a cycle, depicted in Fig 4, of unsatisfactory communication and experiences in the health care setting.⁷ The patient

presented at the beginning of this article illustrates how this could happen: She was turned away for being late without an inquiry as to the reason, and the patient did not think the office would care why she was late.

The IOM report recognized the need for further research while finding evidence of stereotyping according to race-ethnicity. Among studies cited was a survey conducted by Van Ryn and Burke⁸ of 193 physicians and 618 physician-

Hypothesized mechanisms through which provider factors influence race/ethnicity disparities in healthcare

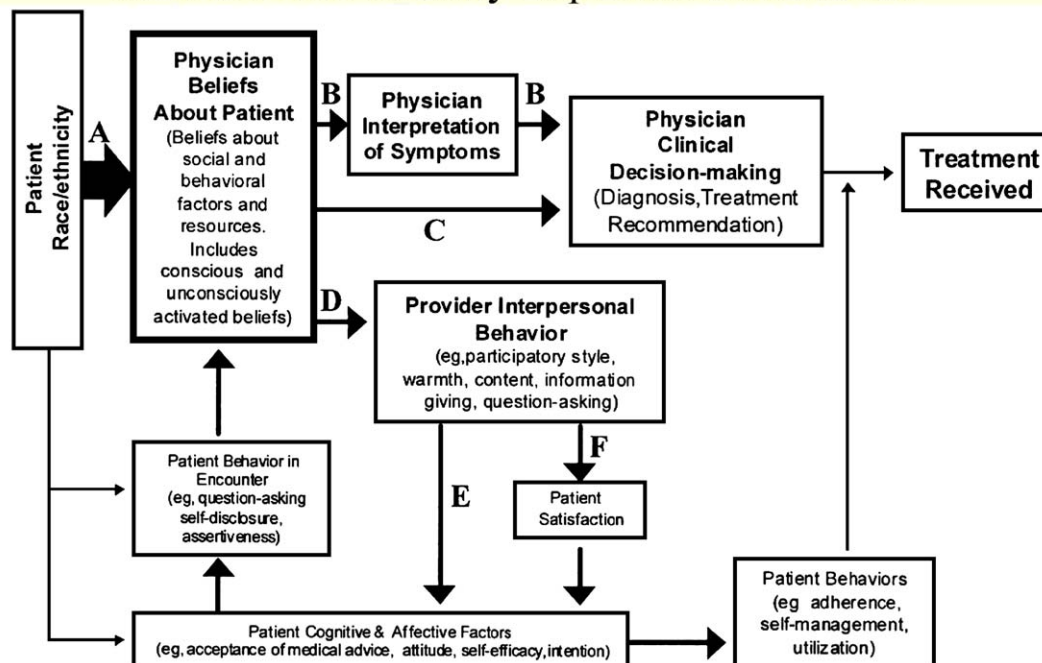


FIG 4. Hypothesized mechanisms through which provider factors influence race-ethnicity disparities in treatments received independent of clinical appropriateness, payer, and treatment site. The thickness of each arrow indicates its relative estimated importance by which patient-provider behavior might contribute to race-ethnicity disparities in medical care. Reprinted with permission from van Ryn.⁷

patient encounters after a cardiac angiogram. Controlling for patient age, sex, socioeconomic status, and health status, as well as physician age, race, sex, and specialty, the investigators found that physicians tended to perceive black patients less favorably on a number of characteristics, including intelligence, pleasant personality, adherence to medical advice, and likelihood of abusing alcohol or other drugs. Johnson et al⁹ examined the association between patient race-ethnicity and patient-physician communication by using audiotape and questionnaire data from 458 African American and white patients who visited 61 physicians. Patient-centered communication was computed by dividing the amount of socioemotional talk and questions asked by the patient by the biomedical elements of talk during an interview. Physicians were more verbally dominant and engaged in less patient-centered communication with African American patients than white patients. Both African American patients and their physicians displayed lower levels of positive affect than white patients and their physicians. The investigators acknowledged potential limitations to their study, such as that only 50% of recruited physicians participated, and those who did tended to have different demographics than those who did not (ie, participating physicians were younger, more likely to be female, and had fewer years in practice). These

investigators concluded that targeting both patients (finding ways to empower patients) and providers (education and training) for intervention should be addressed in subsequent studies.

WHAT IS RACE?

Our previous discussion has used the term as if it is a well-defined characteristic, but it is not. Some have called it a genetic construct, others a social construct. Is a person with one black and one white parent black or white? Most black individuals have evidence of white ancestry.¹⁰ In many clinical and genetic studies, race-ethnicity is self-reported. In fact, genetic researchers use the term “self-identified race or ethnicity” (SIRE) to describe how race is defined in a genetics study. Yet in one study one third of individuals reported their race-ethnicity differently than they had the previous year.^{11,12} We do know that genetic variability correlates with SIRE, although only to a minor extent, explaining only about 13% of the genetic variability in individuals. SIRE is also associated with social and cultural factors.¹¹

Are there genetic differences of significance among racial and ethnic groups that result in increased morbidity from asthma? Or is the health disparity associated with

asthma completely a result of the social and physical environment? Barnes² excellent article addresses these complicated questions in the context of recent genetic research. Importantly, and as in the perspective by Joseph et al,¹ the effect of environment is considered.

ASTHMA AND GENETICS

With the mapping of the human genome, there has been an explosion of research trying to understand the genetics of complex diseases like asthma and the interactions of genetic and environmental influences. In her outstanding perspective, Barnes² lays out the principles of the new field of genetic epidemiology and discusses the relevance of race-ethnicity. Important for our discussions, she explains study designs, such as association studies and family-based studies, concepts of linkage disequilibrium, and population stratification. She goes on to present how concepts of race are used in clinical research.

Asthma is what geneticists call a complex disease, one in which many genes likely each play a very small role. Additionally, the exposures of the environment influence the expression of these genes. Barnes² points out that linkage disequilibrium, the nonrandom association of genes that might have been conferred by ancestry, is eroded over time by genetic drift, migration, and the increasing mobility of society. Environment, social or physical, continues to influence phenotype both directly and through modification of genetic expression.¹¹ As Barnes² observed, no asthma genes are uniformly associated across races, supporting the importance of consideration of the social and physical environment. It is important to remember that social scientists frequently express the fear that in pursuing genetic studies, taking account of social factors will be neglected, and stereotyping will be perpetuated. On the other hand, determining significant genetic contributions to phenotypes with information derived from considering environment might lead to better therapies.

In performing genetic epidemiologic studies, how is race-ethnicity to be taken into account as a marker of the social and physical environment or a marker for genetic homogeneity? Barnes² indicates in her perspective that genetic homogeneity differs between SIREs. But as Rebbeck and Sankar¹¹ state in a recent editorial, ethnicity, race, or ancestry should not be ignored. SIRE is a marker for dietary preferences, health beliefs and practices, and other environmental exposures and not an absolute characteristic of an individual, and might change depending on how questions on race-ethnicity are framed. With our increasing diversity, SIRE will become increasingly complex. Thus Rebbeck and Sankar recommend that in research race-ethnicity should be described carefully and used consistently and that consideration should be given to how this information contributes to the study hypothesis, operationalizing it in a way that best fits the concepts being tested.

Both the perspectives by Joseph et al¹ and Barnes² emphasize the need to consider a wider range of patient

groups, particularly underserved minorities, patient groups with the highest morbidity. Barnes comments that the higher than expected asthma prevalence among Puerto Ricans living both in the mainland and on the island supports a possible role for genetics. Interestingly, an unidentified social cause could be responsible for the increased morbidity in Puerto Ricans when compared with other Hispanic groups. For example, whereas all Puerto Ricans are United States citizens, Hispanics from other groups are not uniformly citizens. The poorest of these, which are likely to be the most in need of health care, might include illegal aliens who would be less likely to present for medical attention for fear of being deported. Whether a social effect like this accounts for the differences observed among Hispanic patient groups needs to be studied.

WHAT ARE THE IMPLICATIONS FOR US AS PRACTICING ALLERGIST-IMMUNOLOGISTS?

What does all of this mean for the patient described at the beginning of this article and for our patients? The IOM report found that stereotyping and bias increase in the patient-physician encounter as time allotted for a visit decreases and financial constraints and diagnostic uncertainty increase. Cultural diversity results in patients having different ways of describing symptoms, giving a medical history, and attributing significance to temporally or geographically associated events. Thus more time is required for a medical history and examination at a time when there is increasing pressure to take less.

Returning to our patient described at the beginning of this article, what is the primary influence on the state of her asthma? Most likely there are many influences. She might have asthma susceptibility genes that increase her risk. She also might have environmental exposures that make her disease worse, although we cannot determine this from what we know. Her SIRE and her race-ethnicity as perceived by others determine her social environment, an environment that without a doubt makes it very difficult for her to receive optimal care. Both perspectives make it clear that for any individual patient the effect of the myriad of various susceptibility genes and cultural and environmental exposures are unique. The individual differences are more important than the differences among socially defined groups like racial groups. For the physician, we are best able to have an effect on some environmental exposures but most importantly in improving the social climate, improving access to care, and focusing on the patient-physician interaction.

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