Diagnostic Deserts: Community-Level Barriers to Appropriate Genetics Services

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The limited geographic spread of medical genetics across the country has been well-documented.[1] The majority of the populace still lives outside of the catchment area of medical geneticists, with these providers often densely concentrated in large metropolitan areas.[1] Moreover, it has recently been reported that the United States has 2.5 times fewer clinical geneticists than the estimated need for this specialty,[2] which has been described as a "critical shortage."[1] Access to appropriate molecular diagnostic services is therefore a significant issue facing medical genetics and American healthcare more broadly.

The concept of food deserts has been used since the early 1990s[3] to explore the community-level barriers people face in acquiring affordable and nutritious food. The concept has been expanded more recently to investigate justice issues as diverse as access to legal services[4] and access to the internet.[5] Of particular relevance here, the concept of healthcare deserts has begun to gain purchase in the literature on inequalities.[6–8] Additionally, domain-specific healthcare deserts have been described, including, for example, medication[9] and surgical deserts.[10]

While as of yet, there has not been a systematic or scoping review of the literature on healthcare deserts, these areas can broadly be said to mirror traditional food deserts in being defined along two primary dimensions. One dimension is the distance or time it takes for a person to arrive at the nearest food market or healthcare facility. The second dimension consists of the socioeconomic factors that may impact a person's ability to acquire nutritious foods or appropriate healthcare services, irrespective of the physical location at which they are obtained.[11]

However, unlike traditional food deserts, healthcare deserts are affected by a wide variety of other barriers, and their subtypes are heterogeneous in nature. It is for this reason that several healthcare specialty deserts have been enumerated. For instance, barriers to access to one healthcare service such as pharmacies may or may not mirror barriers to access to another healthcare service, such as primary care, in any given community. In the same way, having access to good primary care does not necessarily mean that one has access to competent genetics services. Thus, given healthcare deserts' heterogeneity, there is the important potential for issues regarding access to one sort of healthcare service to be *unique to* that service.

In what follows, we propose the concept of *diagnostic deserts* to analyze the distinctive set of barriers that communities of patients with undiagnosed rare genetic disorders face on their journey to a molecular diagnosis for their symptoms. Diagnostic deserts prevent these communities from accessing expedient, affordable, and appropriate care. Such deserts are distinguished from other forms of healthcare desert by a specific and unique constellation of

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issues entailed in the diagnostic odyssey. Moreover, they pose particular ethical issues of justice as they affect not only individuals but also communities of individuals, as members of specific classes. Our conceptualization of *diagnostic deserts* is based on a series of interviews we conducted with patients seen in the Undiagnosed and Rare Disease Clinic (URDC) at Indiana University and an extensive review of the literature on issues in accessing genetics services and on healthcare deserts. We believe that this conceptualization will make visible the unique, community-level barriers that affect patients in need of molecular diagnostic services. Following Shaw,[12] we break these barriers down into three superordinate categories: barriers of abilities, assets, and attitudes.

BARRIERS OF ABILITIES

Physical or spatial inequalities are possibly the most common impediments to diagnosis. The widely acknowledged shortage of genetics specialists[1,2] means that many Americans live far outside the catchment area of appropriate diagnostic services. Patients may live in rural communities, dozens or hundreds of miles away from the closest medical geneticist. Mobility issues characteristic of certain patient populations, however, may prevent individuals and their families from accessing even nearby clinics. For instance, patients unable to drive or suffering from chronic pain may find it difficult or practically impossible to utilize geographically proximate services. Other families may require nonemergency medical transportation but live in regions where it is difficult to schedule such services. And some patient populations suffer from physiologic limitations to their mobility.

On the diagnostic odyssey, many patients find themselves needing to travel to multiple different clinics and multiple different specialists in order to uncover a label for their condition. Living in a diagnostic desert can complicate a patient's diagnostic odyssey, as searching for an appropriate diagnosis can require numerous hours and miles of travel. In some cases, diagnostic deserts may be vast enough that a patient's odyssey becomes an international journey: Lacking adequate genetics support in their home country, they may have to arrange travel abroad in search of a genetic diagnosis and appropriate care.

BARRIERS OF ASSETS

Many patients face financial barriers when pursuing a diagnosis for themselves or a loved one. Most straightforwardly, patients often struggle to cover the cost of medical genetics visits and indicated genetic testing.[13,14] In medical genetics, insurance coverage for seeing multiple specialists and for diagnostic genetic testing may be particularly salient issues applying to the rare disease community writ large.[15–17] Such costs make diagnostic deserts not only geographic in nature, but also financial. Patients seeking genetics services must also account for the costs associated with travelling to frequent appointments, as mentioned above. This includes the cost of transportation and lodging, as well as the professional cost of spending vacation days or going without pay while on the road. Reducing or renouncing one's job often entails losing insurance coverage, thus compounding the barriers of assets. For some patients navigating a diagnostic desert, searching for a diagnosis for themselves or someone for whom they care becomes a full-time job.[18]

BARRIERS OF ATTITUDES

Attitudinal barriers to access also exist in the form of values and beliefs that oppose genetic testing or cause reticence to participation in medical procedures. The long duration of the diagnostic odyssey may lead to frustration and cause some patients to give up their search. In such cases, diagnostic deserts arise from a loss of motivation in patients who are faced with an emotionally burdensome and prolonged diagnostic odyssey. Low health literacy also stands as an attitudinal barrier that can make navigating the healthcare system challenging. Patients with low health literacy may struggle to uncover a successful path from their symptoms to a genetic diagnosis, and so they may find themselves trapped in a diagnostic desert. In addition to this, low health literacy can weaken a patient's legibility when describing symptoms and health events to providers. One study found that patients with a rare disease who have difficulty acquiring information about their condition are five times more likely to receive a misdiagnosis for their condition.[19] When dealing with rare diseases, the relevant level of literacy needed to understand the condition is often higher for both patients and clinicians.[20] In this way, simply having access to a genetics specialist may not be sufficient to navigate a diagnostic desert successfully (hence our emphasis on the need for patients to travel).

Attitudinal barriers on the part of clinicians also contribute to the formation of diagnostic deserts. Patients often rely on primary care physicians to recognize the signs of an underlying genetic disorder. This means that local nonspecialists must have a high enough degree of knowledge about genetics and must be willing and able to refer patients to an appropriate genetics clinic. Another such attitudinal barrier is that local clinicians may have varying personal and research interests in specific types of rare disorders, making certain conditions better understood and supported than others, irrespective of patient numbers or phenotypic overlap.

CONCLUSIONS

It is our hope that in conceptualizing community-level barriers to genetics services under the banner of *diagnostic deserts*, patterns in unjust access to such services may be identified and addressed. It is not merely that we require more genetics specialists in the United States to increase catchment.[1] A simple calculation of diagnostic deserts on the basis of specialists by square mile or by population size proves insufficient in describing these disparities. Other issues are critical to understanding the unique constellation of barriers represented by diagnostic deserts, such as transportation, insurance coverage, and health literacy. Solutions to the problem of diagnostic deserts, therefore, must be multifactorial.

One potentially promising partial solution is heralded by the rapid growth of telehealth. Given that telehealth does not require travel on a patient's behalf, it is possible that virtual clinics may serve as oases for families and patients living within diagnostic deserts, avoiding painful and difficult transportation and missing work. However, given that accurate genetic testing may require careful phenotyping and physical examinations, screen-mediated virtual visits may prove insufficient for some patients.[21]

Entire communities – be they geography- or disease-based – can suffer from the effects of diagnostic deserts. This constellation of barriers can increase the time to diagnosis, incidence of misdiagnoses, and financial and mental health burden of the diagnostic odyssey. More

research is needed to improve our understanding of diagnostic deserts in order to achieve equitable access to appropriate genetics care for all patients in the United States.

DISCLOSURE

The authors declare no conflict of interest.

ETHICS DECLARATION

The interviews that informed this commentary were part of a study approved by the Indiana University School of Medicine Institutional Review Board (IRB). All participants provided informed consent.

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