THE STATE OF BLACK ARIZONA

# What does the experience of rare diseases have in common with COVID-19? Plenty.

For the tens of millions of people in the United States seeking diagnosis and treatment for rare diseases, the pandemic is a trip down a familiar path of unknowns. Similarly, in so many respects, COVID-19's disproportional impact on people of color is nothing entirely new for the Black community, which continues to feel the impact of a long legacy of inequity throughout the U.S. health infrastructure — research, diagnosis, treatment, access to care.

For African Americans fighting for diagnosis and treatment of rare diseases, including those that primarily impact the Black community, COVID-19 made an already-challenging everyday existence many times worse.

In 2020, the State of Black Arizona made raising awareness of rare diseases part of its mission — gathering data and information about issues affecting African Americans in the state; fostering a collaboration on addressing those issues and empowering the community to advocate for more of the positive changes. We see progress in the diagnosis and treatment of rare diseases, but there is substantial room and an undeniable necessity for improvements.

This white paper continues SBAZ effort to encourage better policies and practices that increase awareness of rare diseases, reduce possibilities of misdiagnosis for lack of awareness of a particular disease, and address the unacceptable consequences of delayed or denied access to care.

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It's not unusual to find African Americans overrepresented in what's called the "diagnostic odyssey". We travel alone on some rare disease journeys.

*Sickle cell disease is among the most well-known of rare diseases impacting the Black community.* It's also a very good illustration of the struggles for people with rare diseases and of the need to strongly advocate for research and treatment options.

The history of sickle cell disease began in 1910 when a physician noticed a "peculiar, elongated sickle-shaped" red blood cells in a patient with anemia. Early on, it was known that the disease occurred only or primarily in people of African origin. It wasn't until the early 1970s, with the civil rights movement calling attention to racial inequality in health care, that the deadly genetic condition received government funding for screening, research, and treatment. With solid information and a spirit of collaboration, African American community leaders also spearheaded public awareness campaigns. New treatments are improving the quality of life and extending the life expectancy of people with sickle cell disease.

*Cardiac amyloidosis is another disease that significantly impacts the Black community. Sometimes called "stiff heart syndrome," cardiac amyloidosis occurs when amyloid deposits take the place of a normal heart muscle.* Most people with cardiac amyloidosis never receive a proper diagnosis and remain undiagnosed for many years after symptoms become severe. Cardiac amyloidosis is over-represented among elderly African Americans with severe congestive heart failure.

Dr. Mathew Maurer, who has been studying cardiac amyloidosis for 15 years, told Cardiology Magazine, a publication of the American College of Cardiology, in a February 2021 article, that the disease is more common than once thought. He said that if you went to a newborn nursery today and screened for hereditary cardiac amyloidosis, about one in 25 babies who are Black would have the gene variant linked to hereditary cardiac amyloidosis. Carrying the mutation does not guarantee the eventual development of the disease, but that information is worth knowing considering the prevalence of cardiac amyloidosis.

In the same Cardiology Magazine article, Dr. Ronald Witteles, co-director of the Stanford Amyloid Center at Stanford University, said if you look in the right places, you'll find cardiac amyloidosis again and again. "When I give talks on cardiac amyloidosis, I tell cardiologists and internists, 'If you haven't seen a patient with amyloidosis in your practice in the last year, it's not because it wasn't there, it's because you missed it,'" Witteles said.

**Knowledge is power along any health journey. It's true for patients and it's true for curious, caring health care professionals. With new advances in rare disease diagnosis and treatment, what's needed now to see real progress for patients and their families, is greater awareness and the resolve to change or create policies affecting health outcomes.**

*A disease is considered rare in the United States when it affects less than 200,000 people. One in 10 people in the United States have rare diseases. That's 30 million people. More people are impacted by rare diseases than cancer and AIDS combined.*

On average, it takes nearly five years to get an accurate diagnosis of a rare disease. The rare disease journey is filled with evaluations, studies, trials, dead ends, roadblocks and brick walls. But more and more, with advances in diagnosis and treatment, there is hope and optimism in the rare disease community. Finding advocacy support, seeing the impact of beneficial research, and gaining access to treatment help smooth the way to better health outcomes.

The State of Black Arizona is committed to creating opportunities for more hope and optimism in the rare disease community. As part of our mission to inform, collaborate and empower, we encourage the health community and others to work with us to help identify rare diseases, particularly those that disproportionately impact the African-American community, under a large umbrella of advocacy. There's room to build upon recent success.

In 2021, Arizona Gov. Doug Ducey championed expansion of telemedicine as a way to improve patient access to health care. He signed House Bill 2454 that makes it easier to continue pamdemic-era use of telemedicine and encourages the health-care industry's continued research and investment in an innovative way of serving patients by, for example, reimbursing providers for a telemedicine visit as it would for an in-person visit.

**Rare disease advocates in Arizona have supported the push for implementation of beneficial public policy, including changes in newborn screening and in step-therapy practices.**

In 2021, the Arizona Legislature approved an expansion of newborn screenings to include all conditions that are federally recommended. The expansion includes rare diseases and disorders. Step therapy is a cost-control effort in which insurers require patients to take one or more alternative medications before they can access the medicine prescribed by their provider. Step therapy is a common concern of individuals and families dealing with rare disease because it is often applied to patients with little regard for their medical situation.

Also in 2021, Arizona enacted a reform that creates step-therapy protections for health insurance policies issued or renewed on or after Dec. 31, 2022. The new law streamlines the step-therapy process and gives Arizonans a pathway to request exceptions to mandated step-therapy treatments.

But opportunities for policies and practices that improve diagnosis and treatment of rare diseases remain. They include improving access to care through more inclusive telehealth medicine and to biomarker testing for precision medicine.

While COVID-19 put telemedicine at the forefront of creating options for access to health care, communities of color and those with lower socioeconomic status were marginalized during the pandemic in part because of the lack of access to remote care. With the proper broadband infrastructure and education, telemedicine could continue to improve access to health care for marginalized communities. People with rare diseases who don't live close to urban centers that are homes to major medical and research centers also would benefit from more robust, reliable telehealth connections.

The same access issues apply to biomarker testing used in precision medicine. Biomarker testing provides information about a person's genes that can be critical to diagnosis and treatment options. Precision medicine holds promise for improving care across a broad range of diseases, including sickle cell. Marginalized communities tend not to have access to precision medicine because their level of insurance doesn't cover it.

Insurance policies continue to affect access to care for individuals with rare or complex or otherwise costly conditions. Both insurance pre-authorization practices and in discriminatory benefit design, in which coverage, intentionally or not, prevents individuals with complex or otherwise costly conditions from obtaining appropriate treatment. Both cause uncertainty and delay quests for optimum health.

Advocacy comes in many forms, including patient or personal advocacy. Patients and caregivers must have the information to be their own advocates. They should feel empowered to trust themselves and the understanding of their situations that there is more to be uncovered than the doctor may perceive.

While self-advocacy is key to patient success in finding answers to unusual health challenges, everyone needs help at some point in accessing good information and to change policies and practices that pose barriers to better health outcomes. Progress on an individual or a systems level, doesn't happen without collaboration.

People with rare diseases need information, focused attention, and respect. They also need a research and medical infrastructure built for discovery. This would lower the odds of missing the signs of diseases that impact our community. They also, need research, medical information and advocacy to shorten diagnostic odysseys and improve chances of enjoying optimal health with conditions that are becoming better understood and managed with research and development less burdened by structural racism and discrimination.

**Knowledge is power to advocate for the proper consideration and the best treatment options available for rare diseases. Knowledge and advocacy of policies and practices focused on improved care, should take some of the treachery out of the diagnosis odyssey.**

**Collaboration is key to sustaining an energetic response to improving conditions for people with rare diseases and their families. The State of Black Arizona stands ready to work with partners willing to do the essential, overdue work of removing barriers to effective, equitable health care.**