RDDC
RARE DISEASE DIVERSITY COALITION

HEALTH EQUITY AND RARE DISEASE

It's Rare

A condition affecting fewer than 200,000 people in the US is considered "rare".

Rare diseases are more common than they sound. It is estimated that 25–30 million Americans—nearly 1 in 10—suffer from a rare disease.[1, 2] The rare disease community comprises approximately 400 million people around the world and represents over 7,000 individual diseases.

It's Rare

It can take an average of 5-7 years to get a diagnosis of a rare disease, even longer for people of color.

Even for those who have access to adequate medical care, the lack of information and knowledge about rare diseases by most medical experts and researchers makes it difficult to get a diagnosis. Delays in diagnosis and treatment can result in serious disease progression. Studies already show that health outcomes are strongly tied to social determinants of health. For the non-white population (people of color) with a deep-rooted mistrust of the medical establishment or the lack the time or insurance for doctor visits, diagnosis may be delayed even further. Many patients are misdiagnosed multiple times before getting an accurate diagnosis. Moreover, not being able to share previous family history of a rare disease or results of genetic testing often contributes even further to delays in diagnosis and treatment.

It's Rare

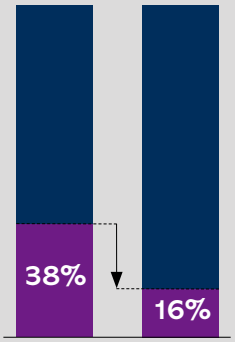
Only 10% of rare diseases have FDA-approved treatments, and people of color have less access to specialists, clinical trials and innovative therapies.

Medical professionals may underestimate the level of interest or capability that people of color have to understand their own disease. They often decline to offer the same access to clinical trials or treatment options as they do to their white patients because they incorrectly assume a person of color will find the process too complicated, require too much explanation or not have the family or financial resources to participate in a clinical trial.

Rare disease specialists are limited in number and are geographically scattered, compounding the challenge of securing a timely diagnosis and treatment from knowledgeable professionals. Access to these specialists and to the limited therapies available can be prohibitively expensive for many families and geographically out of reach. Patients and caregivers of color are also sadly under-represented in the rare disease advocacy groups that have been so important in educating and providing resources to those newly diagnosed with a rare disease.

Underrepresentation in Clinical Studies

People of color are less represented in research studies, leading to a lack of understanding about effective treatments. Despite making up more than **38% of the U.S. population, people of color comprise only 16% of research study participants.**



It's Rare

Since 80% of rare diseases are genetic, properly understanding the genomics of diverse ethnic populations is critical to increasing the speed of diagnosis.

Rare diseases are often caused by a genetic mutation and although a particular disorder may be seen more frequently in one ethnic group, there are people of color who have many rare genetic disorders.[3] Although some genetic disorders, such as sickle cell anemia, thalassemia, and hATTR amyloidosis, are more likely to occur among people who trace their ancestry to a particular geographic area.[4] Unfortunately, 80% of the participants in genome-wide association studies are of European descent. This has devastating implications for diverse populations and the rare diseases that disproportionately impact them.

Rare Disease Diversity Coalition is supporting patients and families living with rare diseases, advocating for research, resources, and improved healthcare access, and spreading awareness and breaking down the barriers surrounding rare diseases. **Through the work of their 3 working groups:**

Diversity in Research and Clinical Trials

Government Regulation, Legislation & Policy

Patient, Provider, Caregiver Journey

Rare Disease Diversity Coalition Mission

Rare Disease Diversity Coalition is dedicated to addressing the extraordinary challenges faced by historically underrepresented rare disease patients as encompassed by social determinants of health (SDOH). The Coalition brings together rare disease experts, patients, health care professionals, diversity advocates, and industry leaders to bring about evidence-based solutions that alleviate the disproportionate burden of rare diseases on marginalized populations.

The time is NOW to align resources and advocates to address the challenges faced by underrepresented populations with rare diseases in accessing adequate medical care. Together, we can make a difference for the millions living with rare diseases. Visit RDDC at www.rarediseasediversity.org to learn more and get involved.

REFERENCES

1 National Institutes of Health. "Rare Diseases FAQ." National Human Genome Research Institute. 10 Jan 2020, https://www.genome.gov/FAQ/Rare-Diseases. Accessed November 2020.
2 United States Census Bureau. U.S. and World Population Clock. https://www.census.gov/popclock/. Accessed February 2020.
3 Ando, Y., Coelho, T., Berk, J.L. et al. Guideline of transthyretin-related hereditary amyloidosis for clinicians. Orphanet Journal of Rare Diseases 8:31 (2013). https://doi.org/10.1186/1750-1172-8-31
4 Mayo Clinic. "Thalassemia." Mayo Clinic, Mayo Foundation for Medical Education and Research, 22 Nov 2019, www.mayoclinic.org/diseases-conditions/thalassemia/symptomscauses/syc-20354995.