Working Every Day for the Rare Disease Community

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Approximately 400 million people around the world are living with a rare disease.[1] Though the definition of a rare disease varies among countries, people living with these conditions have much in common.

People with rare diseases face significant challenges, such as a lengthy diagnostic journey, lack of treatment options, and limited research into and awareness of their conditions. These challenges weigh even more heavily on racial and ethnic minorities and marginalized groups, leading to economic hardship, difficulty accessing care, and poorer outcomes for both patients and caregivers.

As we mark Rare Disease Day on February 28, we must be aware of the important changes that can improve health equity and access for people living with rare diseases, as well as their families and caregivers.

First, the diagnostic odyssey for rare disease patients must be shortened. Rare diseases are difficult to diagnose, often taking up to five years and involving numerous visits to multiple doctors, specialists, and consequently, many misdiagnoses.[2] In rural or underserved areas, this problem is compounded by a lack of medical clinics, physician experience with rare diseases, and transportation options. At Alexion, AstraZeneca Rare Disease, we are working to address these challenges by raising awareness about rare diseases among health care providers and funding transportation for rare disease patients who need assistance in traveling to a medical clinic for diagnosis.

Second, we must also encourage access to powerful diagnostic tools like newborn screenings and genetic testing. Around 80 percent of rare diseases are genetic,[3] and nearly 50 percent of rare disease patients are children.[4] Diagnostic tools have been proven to speed diagnosis and reduce health care costs. In fact, pilot genetic screening programs in the US states of California[5] and Florida[6] have saved millions of dollars through fewer unnecessary procedures, tests, and hospital stays. In the United Kingdom, the National Health Service is undertaking an ambitious effort by enabling all seriously ill children with an unexplained disorder to be eligible for genome analysis.[7] Yet, widespread access to these important tools, even within countries like the UK and the United States, is hampered by a variety of laws governing their use and insurance coverage. Broader access should be a priority – not just in select states or nations, but everywhere, if possible.

Third, we must conduct outreach and education within minority and ethnic communities and with their health care providers. Engaging early and often with patients to understand their journeys is crucial to uncovering what health inequities they face. Health care providers often lack knowledge of rare diseases and may even have their own biases, making it difficult to diagnose and provide appropriate treatments for patients. Raising awareness of rare diseases among these communities can make a meaningful difference in care.

Finally, we must ensure that underrepresented populations have a fair chance at inclusion in clinical trials. Today, less than 5 percent of rare diseases have an approved treatment option.[8] New treatments are critical but with rare diseases, patients are scarce and the presentation of a disease may differ among individuals. For these reasons, regulatory pathways in different countries should allow for flexibility in study design protocols for rare diseases, and we should identify the relevant clinical trial sites across a broader geographical spread so we can increase diverse participation in clinical trials.

At Alexion, AstraZeneca Rare Disease, we are committed to working with the patient community and health care stakeholders worldwide to address these challenges. As is often said in the rare disease community, “rare is many, rare is strong, rare is proud.” Together, on Rare Disease Day — and every day — we can make a powerful difference.

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[1] Global Genes (n.d.). Homepage - Global Genes. Retrieved February 8, 2023, from https://globalgenes.org/rare-disease-facts/

[2] National Organization for Rare Disorders (2020, November 19). Barriers to Rare Disease Diagnosis, Care and Treatment in the U.S.: A 30 Year Comparative Analysis. National Organization for Rare Disorders | NORD. Retrieved February 8, 2023, from https://rarediseases.org/wp-content/uploads/2020/11/NRD-2088-Barriers-30-Yr-Survey-Report_FNL-2.pdf

[3] Global Genes (n.d.). Homepage - Global Genes. Retrieved February 8, 2023, from https://globalgenes.org/rare-disease-facts/

[4] Global Genes (n.d.). Homepage - Global Genes. Retrieved February 8, 2023, from https://globalgenes.org/rare-disease-facts/

[5] Project Baby Bear. Rady Children’s Hospital – San Diego. June 2020. Available at https://radygenomics.org/wp-content/uploads/2021/04/PBB-Final-Report_07.14.20.pdf.

[6] Advanced Genomics for Critically Ill Children. Nicklaus Children’s Hospital: Project Baby Manatee. June 2020. Available at https://www.nicklauschildrens.org/NCH/media/docs/pdf/research/Final-report-State-Appropriations-NCH-PMI.PDF.

[7] NHS England (2023). NHS Genomic Medicine Service. NHS England and NHS Genomic Medicine Service. Retrieved February 15, 2023, from https://www.england.nhs.uk/genomics/nhs-genomic-med-service/

[8] Global Genes (n.d.). Homepage - Global Genes. Retrieved February 8, 2023, from https://globalgenes.org/rare-disease-facts/