Driving health equity for the rare disease community

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Collectively, more than 400 million people around the world are living with a rare disease, defined as conditions that affect a small number of people compared to the general population.[1] The rare disease community is by far the most marginalised patient community, no matter their race, background, socio-economic level or geography, because they face unique challenges, many beyond those that hinder access to health care for people living with more common chronic conditions.

People living with rare diseases often have a lengthy diagnostic journey, lack treatment options, and encounter limited awareness of their conditions. For people living with a rare disease, health inequity remains a major barrier to their care, and it is a top priority that we must address.

There are more than 10,000 rare diseases, more than 90 percent of which do not have an approved treatment.[2, 3] On average, it takes nearly five years —and often longer — to receive a rare disease diagnosis, a journey that often involves multiple doctors, specialists and misdiagnoses along the way.[4] Many rare diseases can be difficult to detect, making misdiagnosis common, particularly for neurological diseases or diseases that present in childhood or adolescence.

Before and after diagnosis, living with a rare condition can make nearly every aspect of life more difficult. People who have rare diseases, along with their caregivers, often experience economic hardship, greater social isolation, significant delays in treatment, and poorer health outcomes. In countries with weak medical, social, and economic infrastructures, the challenges are magnified for people with rare diseases.

Making a meaningful impact for people with rare diseases, everyday

We are committed to taking bold steps to overcome these societal and policy challenges and improve health equity for people living with rare diseases:

- **Reducing the time to diagnosis:** Access to effective screening and diagnostic tools remains inequitable for many rare disease patients. We are working to expand access to newborn screenings and next-generation sequencing, two key ways to provide needed answers more rapidly. We also are developing and testing digital health technologies to make it easier to reach undiagnosed patients in underserved geographic locations.
- **Improving access to care and treatment:** Rare disease patients — regardless of where they live — face significan obstacles to accessing quality health care and treatment. With digital technology expertise and support, we can overcome barriers for people living in rural or underserved areas, who often lack medical clinics, reliable internet connectivity and transportation options that can delay or impede access to necessary care. These technological advances have hastened the discovery of new treatments for patients with rare diseases.
- **Encouraging the enactment of equitable rare disease health policy:** Addressing health equity challenges will require policymakers and health care stakeholders to consider the unique circumstances inherent to rare disease drug development, reimbursement and diagnosis, among other issues, and craft sound policy solutions. Such solutions also should encourage and facilitate patient access to treatments and be supported by a health care infrastructure that encourages appropriate data collection and medical research, the use of real-world evidence and collaboration across communities.

Underpinning these steps is the need to enhance outreach and education, especially among health care providers, because raising awareness of rare diseases in underserved communities can make a difference in care. Health care providers often lack knowledge of rare diseases and have their own biases, making it hard to diagnose and provide appropriate treatments for patients.

A joint effort to transform the lives of people with rare diseases

While we have an important role to play, we cannot solve all the issues faced by rare disease patients on our own. We applaud the work of organisations like Rare Diseases International, EURORDIS and others that has been instrumental to furthering the concept of health inequity from a rare disease perspective, leading the United Nations to take steps to recognise and address the broad challenges facing people living with rare diseases, and urging UN Member States to take action.

We also have further affirmed our commitment to change through our support of the Global Health Equity Network Zero Health Gaps Pledge. Now we need country-level action to bolster this progress so that those living with rare diseases have the same opportunities as the rest of society. We call on policymakers, health care providers, patients and other stakeholders to join us at every step of the process.

We are determined to make a difference -- reducing time to diagnosis, improving access to care and treatment, and engaging global policymakers to advocate for broader support for the rare disease community. That’s because rare disease patients and their families around the world are counting on us to meet their needs, not only on Rare Disease Day, but on each and every day.

References

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