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Empowering Lives: Rare Disease Diagnostics and Awareness

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Petra Furu, General Manager of Revvity's Reproductive Health Business shares insights in view of "Rare Disease Awareness Month and Day, February 28th"



Rare diseases, often overlooked, affect a significant portion of the global population. However, early detection and intervention can pave the way for improved outcomes and quality of lives.

What are rare diseases?

Rare diseases, also known as orphan diseases, are conditions that affect a small percentage of the population. In various regions, a disease is typically considered rare when it afflicts fewer than 1 in 2,000 individuals. However, the exact prevalence thresholds may vary from one country to another.

A couple of the most common diseases that are universally screened for include congenital hypothyroidism (CH) and phenylketonuria (PKU). CH is defined as thyroid hormone deficiency, which can lead to irreversible neurological deficits as well as poor appetite and breathing problems. PKU is caused by a change in the phenylalanine hydroxylase (PAH) gene that can quickly cause infants to exhibit irritability, posturing, increased deep tendon reflexes. Today more than 50 rare diseases

are included in newborn screening (NBS) programs globally, and new diseases are added. Recent rare disease NBS expansioninclude spinal muscular atrophy (SMA), duchenne muscular dystrophy (DMD) and mucopolysaccharidosis type II (MPS II).

There are currently 7,000 known rare diseases. According to the World Health Organization (WHO), rare diseases affect roughly one in fifteen people globally with a majority of these diseases affecting children. An estimated 200 million people in the Asia-Pacific region, and 400 million people worldwide, are living with a rare disease.

Challenges in diagnosis and treatment

What makes rare diseases particularly challenging is the diversity and complexity of these conditions. There are thousands of identified rare diseases, each with its own unique set of symptoms and characteristics. Many of these conditions are genetic in nature, resulting from mutations or alterations in an individual's DNA.

Due to their infrequency, rare diseases often pose significant diagnostic and therapeutic challenges. Misdiagnosis or delays in diagnosis are not uncommon, leading to prolonged periods of uncertainty for affected individuals and their families. This underscores the crucial role of early detection and intervention, a mission that Revvity wholeheartedly embraces.

While individual rare diseases may affect only a small number of people, collectively, they impact millions worldwide. One of the objectives of the Asia-Pacific Economic Cooperation's (APEC) Action Plan on Rare Diseases is to raise public and political awareness of rare disease issues amongst APEC economies. Raising awareness and fostering understanding of rare diseases are vital for ensuring that those affected receive the attention and support they need.

What is Revvity doing?

Among the 7,000 rare diseases, 80% of them are genetic. Therefore, NBS plays a pivotal role in the early identification of these conditions. By employing advanced screening technologies, healthcare professionals can detect markers and indicators associated with various rare diseases shortly after birth, enabling timely interventions and personalized treatment plans.

Revvity is continually developing new technologies to meet the demands of NBS today and for the future. Through our research, collaborations, and partnerships we are helping to decipher the role of next generation sequencing in NBS, both in terms of testing and in the delivery of digital solutions and data management. With Revvity's NBS solutions, 33 million babies around the world were screened just in 2023, and over more than three decades, Revvity has helped save 700,000 babies born with life-threatening diseases. Within China, over 50% of the prenatal and neonatal R&D and production is now completed domestically to serve local families across the country.

The Revvity NBS World View Summit in 2022 helped foster collaboration and knowledge-sharing among stakeholders in the newborn screening community. The company is hosting another World View Summit in 2024, aiming to inspire and engage the next generation of thought leaders advancing the education and scientific agenda for newborn screening. It's through such partnerships and initiatives that progress and positive change in healthcare globally can be achieved.

Recognizing Rare Disease Awareness Month

Each February, the world recognizes the strength and resilience of those facing rare diseases and emphasizes the importance of awareness.

As innovative technologies and screening solutions continue to redefine the standard of care, ensuring that no rare disease goes unnoticed, healthcare professionals are able to make early, informed decisions, offering hope and support to families navigating the challenges of rare diseases.

Revvity's unwavering dedication extends beyond technology. We actively collaborate with healthcare communities, advocacy groups, and researchers to foster a holistic approach to rare disease management. By fostering awareness, we contribute to a global effort to transform the landscape for those affected by these conditions. Through our collective efforts, we can raise awareness, promote early detection, and ultimately make strides toward a brighter, healthier future for all.

Together, let's make a difference in the lives of those touched by rare diseases, today and every day.