From anonymity to recognition: The challenge of underdiagnosis and misdiagnosis in rare diseases

As we highlight Rare Disease Day, it's crucial to focus on the often-overlooked issue of underdiagnosis and misdiagnosis in conditions like Arginase 1 Deficiency (ARG1-D), a progressive genetic metabolic disorder characterized by elevated plasma arginine levels, leading to serious health issues. ARG1-D presents with progressive neurological impairment, developmental delay, progressive spasticity and intellectual disability. The invisibility of rare diseases like ARG1-D in both public awareness and medical practice results in delayed interventions or even the omission of relevant treatments, significantly worsening the lives of affected patients.

"We raise awareness for Rare Disease Day, since it's crucial to address the widespread issue of underdiagnosis and misdiagnosis, notably in conditions like ARG1-D. These rare diseases often remain overlooked, leaving patients and families to confront their challenges alone. By amplifying awareness, enhancing education, and implementing standardized screening, we can bridge the gap between anonymity and recognition, ensuring every patient receives the care and support they require." says Anders Edvell, CEO of Immedica.

Recent studies [1] indicates that ARG1-D's birth prevalence is substantially higher than what newborn screening studies previously suggested, especially in North America and Europe. This finding highlights the urgent need for enhanced education about ARG1-D's signs and symptoms, broader testing, and standardized screening to identify patients early and prevent disease progression. The challenge of structural underdiagnosis is unfortunately a common occurrence within the realm of rare diseases.

The burden on caregivers due to the lack of a diagnosis for rare diseases is significant. Families grappling with these conditions not only face emotional and financial stress but also take on the role of primary caregivers while navigating complex medical systems. Challenges such as underdiagnosis and misdiagnosis further complicate matters, prolonging the time required to receive an accurate diagnosis. Consequently, caregivers often feel compelled to reduce their work hours or leave the workforce entirely with the subsequent burden that this implies for families and society.

Marking Rare Disease Day, Immedica reaffirms our dedication to increasing awareness and improving screening for ARG1-D and similar rare diseases, aiming for a healthcare system that promptly and equitably recognizes and addresses the needs of all patients. Immedica have developed the websites www.ARG1Dinfo.com and www.UCDandYou.com to help drive this awareness. Please visit the sites to learn more.

About Immedica
Immedica is a pharmaceutical company, headquartered in Stockholm, Sweden, focused on the commercialization of medicines for rare diseases and specialty care products. Immedica's capabilities cover marketing and sales, compliance, pharmacovigilance, quality assurance, regulatory, medical affairs and market access, as well as a global distribution network serving patients in more than 50 countries. Immedica is fully dedicated to helping those living with diseases which have a large unmet medical need. Immedica's therapeutic areas are within genetic & metabolic diseases, hematology & oncology and specialty care.

Immedica was founded in 2018.Today Immedica employs more than 100 people across Europe and the Middle East.

For more information visit www.immedica.com

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[1] Catsburg et al., (2022)

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