Unveiling the unseen: Rethinking Rare Diseases

In the vast landscape of healthcare, the term "rare disease" may suggest scarcity, but the impact is anything but rare. Within the European Union, a rare disease is officially classified as one affecting fewer than **1 in 2000 individuals**.[1] However, this seemingly low prevalence statistic paints only a fraction of the picture.

Estimates reveal that 6-8% of the European population will suffer from a rare disease during their lifetime. In the **Netherlands**, this translates to over **1 million people** (1 in 17),[2] and for **Belgium**, the figure **surpasses 500,000**.[1] Astonishingly, most of these patients—over **70%—will manifest** with initial symptoms during **childhood**, underscoring the urgency for early detection and intervention.[2]

What defines a rare disease in Europe extends beyond statistical rarity; it encompasses conditions deemed "life-threatening or chronically debilitating". Amidst this classification, a staggering **6000-8000 different rare diseases** have been identified, each presenting unique challenges.[2] Whereas certain rare diseases affect thousands of people (e.g. cystic fibrosis), for others only a few cases are known worldwide.[1]

Low prevalence in combination with limited knowledge about rare diseases facilitates diagnostic delays.[3,4] **Diagnostic delays** of several years (averaging 5 years), inflict profound physical and psychological tolls on patients and their families.[3,4] Misdiagnoses are not uncommon, leading to **inappropriate treatments** and even surgeries, as revealed by Dutch research.[2]

As approximately **80%** of rare diseases **have a genetic origin**, hope beckons through advances in genetic diagnostics.[5] Genetic testing emerges as a pivotal tool in the diagnostic arsenal, offering a beacon of clarity for patients and healthcare professionals alike.

The ripple effects of rare diseases extend beyond personal struggles, casting a substantial societal burden. Recent research by Chiesi Rare Disease and IQVIA illuminates the economic impact, demonstrating that the **indirect cost*** for a rare disease can constitute an average of 29% of the **total burden**[#] when treatment is available.[6] This figure rises to 45% in the absence of treatment, highlighting the value of accessible interventions to alleviate (financial) strain on families and healthcare.

Information and education emerge as powerful allies in the quest for early diagnosis and improved patient outcomes. Even when treatment options are limited, a correct diagnosis provides a compass for both patients and healthcare professionals to navigate their unique paths.[4] In Belgium and the Netherlands, a spectrum of initiatives and resources stands ready to support healthcare professionals in this pivotal endeavor.

In essence, rare diseases, far from being rare, beckon us to unravel their complexities, fostering a collaborative and informed approach for the benefit of patients, families, and the healthcare community. This significance is highlighted on **Rare Disease Day**, on **February 29th**, as a dedicated platform to raise awareness, and unite efforts in the pursuit of solutions for these often-complex medical challenges.

*Indirect cost include patient and caregiver productivity loss, work loss, home changes, traveling and accommodation for medical visits.

Total cost includes direct cost (cost of treatment, medical procedures, hospitalizations, physician visits, home healthcare and other medical cost), indirect cost and mortality cost (based on the value of statistical life and the difference between average life expectancy and that for people with a rare disease).

References:

1- https://health.ec.europa.eu/european-reference-networks/rare-diseases_en. Accessed 20-1-2024.

* 2-Deden AC, Alma MA, and Van Zelst-Stam WAG. Zeldzame aandoeningen sneller diagnosticeren. NTVG 2020;164:19

3-Alma MA, Verheij NP, van der Mei SF, et al. Scherperzicht op diagnostische vertraging bij zeldzame aandoeningen. Groningen: Universitair Medisch Centrum Groningen; 2018

4 - https://www.eurordis.org/our-priorities/diagnosis/ accessed 20-1-2024

5 - https://www.sciensano.be/en/health-topics/rare-diseases/numbers#rare-diseases-in-general Accessed 20-1-2024

6- https://www.chiesi.com/img/CGRD%20Rare%20Disease%20Burden%20DE%20FR%20IT_final.wodc.pdf