Rare but Real: Singapore's Rare Disease Fund

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Rare diseases are an important but often overlooked part of human health. They are not common, but they are still significant and can be hard to understand or diagnose. The recent story of the brave trio – Thaddeus and his twin brothers Hugard and Reynar, all suffering from a rare condition known as mucopolysaccharidosis type VI (MPS VI) – published in *The Straits Times*, underscored this paradox.[1] This article endeavours to shine a light on the lesser-known Rare Disease Fund (RDF) of Singapore, its vital role in supporting families such as Thaddeus', and how the medical fraternity can contribute to this essential initiative.

The financial burden of rare diseases

Medical professionals grapple with a myriad of health challenges, many of which are commonplace and well understood. However, lurking in the undercurrent are rare diseases that, while seldom seen, have significant impact on those they afflict. MPS VI, also known as Maroteaux-Lamy syndrome, is one such enigma.[2]

MPS VI is a rare genetic disorder resulting from the complete or partial deficiency of the enzyme, N-acetylgalactosamine-4-sulfatase, resulting in the pathological accumulation of complex carbohydrates called glycosaminoglycans (previously known as mucopolysaccharides, hence the name MPS VI). Clinical features include deafness and corneal clouding. Body growth is initially normal but comes to an abrupt halt around age 8. With progressive skeletal changes, progressive functional movement limitation is almost inevitable.[3] Nearly all afflicted with MPS VI also have cardiac complications in the form of valvular dysfunction. There is also reduced life expectancy for individuals with MPS VI. This elusive disease is presently known to afflict no fewer than four individuals in Singapore – Thaddeus, Hugard, Reynar and an adult who does not wish to be identified.

The rarity of these conditions often leads to two significant challenges. Firstly, these diseases are frequently overlooked in public health discourse due to the lower number of patients compared to more common diseases. Secondly, the dearth of research and treatment options combined with the exorbitant costs associated with the few available treatments often place these families in a precarious financial position.

Madam Yang, the mother of the three boys grappling with MPS VI, provides us with a poignant glimpse into the daunting financial reality of managing a rare disease. The good news is that enzyme replacement therapy (galsulfase [Naglazyme™]) is effective and has been approved for use in Singapore.[4] The enzyme is administered intravenously over four hours during each weekly session, and has been shown to improve walking and stair-climbing capacity. However, the bad news is that each session costs approximately $10,000 for Thaddeus and $6,000 for each twin, a sum that would only increase with body weight. With such staggering figures, treatment costs could easily surpass a median Singaporean household's annual income within a few months.[1]

However, the burden extends beyond the cost of medication. The time investment for treatment, the potential for lost income due to caregiving responsibilities, and the psychological toll of dealing with a lifelong disease further exacerbate the situation. After exhausting insurance claims and Medisave, a family like Madam Yang's would still be far from achieving financial stability without additional aid. This sobering narrative illuminates the astronomical financial strain and the subsequent urgency for support mechanisms for families battling rare diseases.

Ethical dilemma: cost of innovation vs equitable access

An ethical conundrum is evident here. Historically, new drugs or treatments like monoclonal antibodies or cancer drugs are initially expensive due to developmental costs but would eventually become more affordable. Drug companies hold onto the patent rights so that profits made can recoup their development costs. If a disease is common, the profits generated by countless patients using them will, over the course of time, result in cheaper prices. The problem with rare diseases is that the expensive development costs cannot be absorbed by the very few users.

There are historical precedents where patent rights were relinquished. When Frederick Banting and Charles Best isolated and produced insulin in the 1920s, they each sold the patent to the University of Toronto for just $1, to ensure its widespread availability.[5,6] Jonas Salk, the developer of the inactivated polio vaccine, chose not to patent it, allowing for broad dissemination.[7]

To date, such generosity has largely not occurred for the treatment of rare diseases.

The RDF

In the grim landscape of high-cost treatments for rare diseases, the RDF was established in Singapore in 2019, and it has emerged as a beacon of hope. Funding comes from donations by public companies or individuals, with the Singapore Government matching each dollar donated threefold. This means that for every $1 the public donates, the Government will donate $3, thus yielding a total sum of $4 for the fund.

Within three years of its establishment, the RDF raised $4.9 million in public donations. With the government matching scheme, the total sum raised totalled $19.2 million by 2022. This financial bolstering illustrates the collective power of societal contributions and government support in providing life-saving treatments to patients and families who would otherwise be mired in financial distress. Through this scheme, Madam Yang's twins were included under the RDF, enabling them to receive the necessary enzyme-replacement therapy.[1]

The ground reality

Despite the RDF's assistance, the everyday reality for families contending with rare diseases remains challenging. From managing hospital visits requiring "needle-poking" investigations and treatment injections, to the uncertainty of their sons' life trajectories, Madam Yang's family navigates an intricate maze of physical, emotional and societal challenges. Yet, with the RDF's support, the boys can continue their treatments, enjoy piano lessons, swimming classes, and maintain a semblance of normalcy.[1]

In short, RDF's intervention extends beyond financial relief – it bolsters hope, normality and possibility for families like Madam Yang's. It paints a brighter future where their children, despite battling a rare disease, can enjoy their favourite foods, partake in loved activities, and simply, be children. ◆

The Rare Disease Fund (Singapore)

To find out more about the fund and how you can contribute, please email development@kkh.com.sg.

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