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Urgent call for comprehensive reform of rare disease care in Hong Kong

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To the Editor—We write in response to an article that highlighted the experience of two Cantopop artists whose son was diagnosed with a rare disease.[1] Because of its complexity, immediate discussion is warranted of the critical aspects of managing rare diseases in Hong Kong.

Rare disease, which impacts about 300 million individuals globally, encompasses a number of medical conditions across different specialties.[2] Neurological disorders and metabolic causes account for 40% and 10% of rare diseases, respectively, with tuberous sclerosis and spinocerebellar ataxia being examples in Hong Kong. In general, there is a lack of awareness about rare diseases among healthcare professionals and the public, with consequent delayed diagnosis and treatment.[3] Families who cope with diseases often encounter emotional and psychological problems that are compounded by a lack of specialised psychosocial support and palliative care access.[4] Moreover, the financial strain of managing diseases is substantial; drug costs for rare diseases are reportedly up to 13.8 times higher than those of more common ailments.[3] It is crucial to provide comprehensive care for rare disease patients and their families.

The healthcare system for rare diseases in Hong Kong is not as advanced or well equipped as comparable centres in the US[2] and Mainland China.[5] It faces challenges at different levels. Key issues include insufficient patient support, absence of a specific registry, limited availability of genetic testing, and a high financial burden for patients. To bridge these gaps, Hong Kong could learn from the well-established networks and care models of the US, as well as the central registry in Mainland China, and adopt supportive policies and financial assistance programmes. As a starting point, the Hong Kong Genome Institute (https://hkgp.org/en/) provides a strong platform from which to promote public awareness of rare diseases in Hong Kong. In addition, with the newly established Genetics and Genomics (Medicine) Fellowship of the Hong Kong Academy of Medicine,[6] genetic testing and counselling that target rare disease could be streamlined.

Author contributions

All authors contributed to the letter and critical revision of the letter for important intellectual content. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

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