

Rare Diseases Hong Kong

Submission on the Public Consultation for the 2024 Policy Address

(September 2024)

Established in December 2014, Rare Disease Hong Kong (RDHK) is the first patient group in Hong Kong comprising cross-rare-disease patients and their families with the support of experts and academics in the field. Representing the patients and caregivers, RDHK is committed to enhancing public awareness of rare diseases in collaboration with the stakeholders. We aim to improve rare disease policies and services, and to ensure equal respect and protection for patients in terms of fundamental rights such as healthcare, social support, education and daily needs.

Over the past year, the Hospital Authority (HA) and RDHK have met regularly to discuss and exchange views on strategic issues, clinical services and inclusion of orphan drugs, and have made progress in deepening understanding, enhancing mutual trust and addressing some specific issues properly. RDHK is pleased to learn that in response to patient demands, the HA has set up a “Working Group on Rare Diseases” to co-ordinate different professional teams, establish a patient database for dozens of rare diseases, and study and formulate clinical service strategy. RDHK is grateful to the HA team for their efforts made, as well as acknowledged and appreciated their accomplishments.

For more than two years, the current-term Government has not proposed purposive strategies and measures for rare diseases. RDHK expects that the Government will draw on the responsibilities and actions taken by the Central Government over the past decade in improving the diagnosis and treatment of rare diseases, and will take a macro perspective, proactive and forward-thinking approach to policy development so that more systematic, comprehensive and integrated short- and long-term measures can be launched. RDHK hopes that the Government will consider and adopt the following 8 recommendations for combating rare diseases in the coming year:

- I. Fulfill the role of “the first person responsible” and take the lead in establishing a cross-sectoral coordination mechanism on strategy for rare diseases;**
- II. Keep up with the Mainland’s list of rare diseases and formulate an integrated that suits Hong Kong;**
- III. Provide humanized clinical services for patients with rare diseases;**
- IV. Remove the age restriction on the subsidy for patients with spinal muscular atrophy to access drugs;**
- V. Make dispensing of foods for special medical purposes a regular service;**
- VI. Adopt horizon scanning to set priorities for the allocation of resource in advance;**
- VII. Collaborate on rare disease diagnosis and treatment in the Greater Bay Area; and**
- VIII. Include Herpes Zoster vaccination in the Vaccination Subsidy Scheme.**

The recommendations are elaborated as follows:

I. Fulfill the role of “the first person responsible” and take the lead in establishing a cross-sectoral coordination mechanism on strategy for rare diseases

In response to a written enquiry from a LegCo member, the Health Bureau stated that the HKSAR Government is the first person responsible for taking care of the health of Hong Kong citizens and will continue to provide quality healthcare services to Hong Kong citizens. RDHK fully agrees and supports such role and commitment and hopes that the Government will put it into practice in tackling the challenge of rare diseases.

One of the effective strategies for strengthening the diagnosis and treatment of rare diseases in the Mainland in recent years is to establish the "China Alliance for Rare Diseases", which is composed of various stakeholders such as government departments, experts and scholars, hospitals and universities, business enterprises and patient organizations, to serve as a national platform for communication, coordination and implementation of government policies and measures.

In recent years, rare diseases have been gaining more and more attention from not only the general public, but also the various sectors such as universities, research institutions, innovation & technology enterprises, and healthcare & pharmaceutical industries. With the accumulation of experience in screening, diagnosis and treatment of rare diseases among the HA staff from management to frontline clinical specialists, the Working Group on Rare Diseases has been established. The government officials should also have a more comprehensive and in-depth understanding of rare disease issues. On the existing foundation, if the Government can take the lead to gather the passion and strength of different parties, and coordinate the professional knowledge and resources effectively, the outcomes of tripartite collaboration among the Government, the business sector and the community can certainly be maximised, so that the efforts of the Government and stakeholders can be multiplied with half the efforts. RDHK expects that the current-term Government can become the driving force for the establishment of a cross-sectoral coordination mechanism on strategy for rare diseases. It is suggested that the coordination mechanism be led by the Government and be composed of government officials, HA representatives, scholars from the two local medical faculties, rare diseases specialists, members of the Legislative Council, representatives from patient groups and the business sector, etc. The main functions of the mechanism are:

1. To make recommendations on policy goals and medium to long term strategy for rare diseases in Hong Kong to the Government, based on the existing practical experience and the experience of Europe, the US and the Mainland.
2. To devise and implement in phases the “Hong Kong Rare Disease Action Plan” in accordance with the situation in Hong Kong, including issues of human resources, patient registries, centres of excellence, cross-border collaboration, rehabilitative support, etc.; and
3. To coordinate and implement various measures for rare diseases proposed by the Chief Executive, and communicate the implementation progress with government departments.

II. Keep up with the Mainland’s list of rare diseases and formulate an integrated that suits Hong Kong

RDHK has always advocated that Hong Kong should have a local definition and list of rare diseases to facilitate the effective formulation of relevant policy initiatives and the implementation by various stakeholders.

In 2018 and 2023, the National Health Commission (NHC) and other four departments announce the first and second lists of rare diseases, covering a total of 207 rare diseases. Meanwhile, both the public and private sectors have actively studied how to define rare diseases. Some experts have proposed that the definition of rare diseases is both a matter of science and values, reflecting the government’s attitudes to the vulnerable social groups and its civilization level.

Last year, in reply to RDHK’s enquiry about the progress of providing a local definition for “rare diseases”, the authorities concerned indicated that they would make reference to the 207 rare diseases announced by the Mainland. We think this is a feasible measure at present, at least it provides reference for the stakeholders.

It is hoped that the Government should keep up with the Central Government’s efforts and commitments in dealing with rare diseases when making reference to the Mainland’s catalogue of rare diseases. Based on repeated investigations and in-depth analysis, the Central Government has set out an overall strategy and formulated purposive policies. To ensure that the relevant policy measures can be effectively implemented and achieve results, the Central Government has strengthened and established organisational settings, and implemented multiple measures by multiple departments to respond to the rare disease patients’ demands in all aspects such as diagnosis, treatment, screening, drug research and development, drug security, etc.

Professor Zhang Shuyang, President of Peking Union Medical College Hospital, pointed out in her speech at the symposium held by RDHK last year that China’s response to rare diseases is comprehensive, involving the government, enterprises and the community, and aims to ensure that patients with rare diseases receive early diagnosis and treatment. In tackling the problems of rare diseases, all stakeholders should play their part to the best of their ability. However, the

government's policy is the most critical and fundamental. Without the government's policy and leadership, non-governmental organisations cannot be of much help.

RDHK deeply believes that the SAR Government can make reference to the actions taken by the Mainland and combine Hong Kong's strengths in the healthcare system, scientific research, talents and international connections, etc. It should strengthen its policies and lead an all-round collaborative network among the Government, the business sector and the private sector, so as to minimize the gap between Hong Kong and the Mainland.

In addition, for unknown reasons, the term "rare diseases" seems to be the SAR Government's taboo. In the official documents, rare diseases are referred to as "uncommon disorders". In fact, the United Nations, the World Health Organisation, and the Mainland and Taiwan Governments adopt the term "rare diseases". So, it is suggested that unless there is a good reason for it, the SAR Government should replace "uncommon disorders" with the internationally accepted terminology "rare diseases".

III. Provide humanized clinical services for patients with rare diseases

Most of the rare disease cases are followed up by multiple specialties, and for many patients, follow-up consultation is a difficult challenge. For many patients, follow-up consultations are a difficult challenge, as they have to travel to different specialties and undergo multiple tests at different times on different days. In order to cope with follow-up consultations, patients and their carers have to spend a lot of time and energy on running around, which is physically and mentally exhausting.

According to the HA, the Hong Kong Children's Hospital (HKCH) will become the clinical centre for all paediatric patients with rare diseases in the future, and when they reach adulthood, the patients will be referred according to the needs of different specialties or their personal wish to other hospitals such as the Princess Margaret Hospital (PMH), which specializes in the management of metabolic diseases for follow-up. This arrangement will undoubtedly help improve clinical services for patients and clinical management of rare disease cases.

It is understood that individual specialties of the HKCH have tried to focus on follow-up consultations for certain rare diseases, such as tuberous sclerosis, during a certain period of time. On this basis, it is hoped that hospitals providing follow-up consultation services for rare diseases will try to concentrate the various specialties related to a certain rare disease on providing follow-up consultation to patients at the same time (for example, half a day or one day), so as to minimize the need for patients to travel for many days, and to help the relevant specialties to comprehensively observe and keep abreast of the latest clinical changes of the patients' cases. Hence, more appropriate treatment and care can be provided.

This year, RDHK has proposed to the HA that three rare diseases, namely epidermolysis bullosa, neurofibromatosis and neuromyelitis optica, should be included in the pilot scheme of one-stop cross-specialty follow-up consultation, in the hope that the HA will consider the proposal and respond to it.

In addition, patients should be allowed to go to clinics near their homes for various tests such as taking blood and stool samples, radiological examinations, etc. prior to the follow-up consultation, instead of being mandatorily required to go to designated hospitals. It is learnt that the New Territories East Cluster has already allowed the cancer patients to choose preferable clinic or hospital to go for blood sampling(抽血自由行). RDHK hopes that this practice will be extended to other clusters and rare diseases, and that more “patient-oriented” and humanized clinical services will be introduced in the future.

IV. Remove the age restriction on the subsidy for patients with spinal muscular atrophy to access drugs

Since last year, a small number of patients with spinal muscular atrophy (SMA) over the age of 25 have reluctantly taken the risk to seek medical treatment in the Mainland in order to manage their condition because they are ineligible for the drug subsidy under Community Care Fund. Accompanied by their caregivers, they travel long distances to seek medical treatment across borders in order to prolong their lives. This is by no means an ideal situation at all, for the patients and caregiver, or for Hong Kong that always takes pride in its public healthcare system.

In its explanation of the reasons for not subsidising medication for patients aged 25 and above to the LegCo members, the Health Bureau said, “The HA Expert Panel on SMA (the Expert Panel) has noted and reviewed the findings of an international preliminary observational study published after adult patients with the disease had been treated with the drug Risdiplam. The Expert Panel considers that, at present, the relevant scientific data and clinical evidence are still limited, and there is no empirical evidence to show that such drug can bring significant effects to adult patients with the SMA.”

It can be seen that the relevant decision of the Government is only based on a “preliminary observational study” without mentioning the publication date of the report. It is claimed that scientific data and clinical evidence are limited, but it’s all relative as there is no objective criterion available. Ultimately, it is the standpoint of the decision-makers that matters. In fact, we approached the members of the Expert Panel a few months ago and their views differ from those of the Health Bureau.

Internationally, most of the countries where the above-mentioned drug is currently used, such as Japan, Australia, Canada and the Mainland, do not have age restrictions for users, and there are data showing the different levels of drug effectiveness in SMA type 3 patients over 25 years old. Therefore, RDHK urges the government to earnestly fulfill its role as the “first person responsible” by being receptive and keeping pace with the times, and open the door to medication for SMA patients over 25 years old as soon as possible.

V. Make dispensing of foods for special medical purposes a regular service

As some rare diseases lack effective drug treatments, foods for special medical purposes (also known as medical foods) are used as the primary means of disease prevention and management. This kind of food is equivalent to the General drugs and Special drugs for other patients, and are only available by prescription. But since medical foods are not classified as drugs, they are not dispensed by the Department of Pharmacy like prescription drugs. According to the current practice, the HA will not provide medical foods except for inpatients. Patients have to solve the problem by themselves upon discharge.

Medical foods are specially formulated and intended for patients with distinctive nutritional needs, and are much more expensive than general foods. Therefore, some families facing rare diseases (including phenylketonuria, pyruvate dehydrogenase deficiency and glutaric aciduric type 1) sought assistance from RDHK.

In view of the fact that medical foods are essential in helping patients manage their disease and maintain quality of life, we strongly urge the Government to make “dispensing medical foods to patients in need” a regular service, like the way general and special drugs are dispensed.

It is believed that if the Government is resolved to fulfil its role as the “first person responsible”, it will provide these rare disease patients with the prognostic information and care they needed, and will be able to find ways to adjust the current practice to meet the needs of patients. In fact, in the past year or so, some hospitals in the Mainland for the diagnosis and treatment of rare diseases have recognised the needs of patients and assigned hospital pharmacies to dispense medical foods.

RDHK understands that it may take some time to make operational adjustments. Prior to this, medical social workers could assist some families with financial difficulties to apply for financial assistance as a transitional expedient measure. After the operational adjustments, the provision of medical foods can be made a regular service of the HA.

VI. Adopt horizon scanning to set priorities for the allocation of resource in advance

Due to the rapid development of health technologies and the ever-rising healthcare costs, horizon scanning has been adopted by many advanced countries such as the United Kingdom, the United States, Canada, Australia, etc. to identify new and emerging health technologies that may have impact on the health system so that policymakers can make prearrangements and early planning for regulatory environment, financial management, workplace preparedness, training and education, ethics and so on. According to the Agency for Healthcare Research and Quality in the United States, horizon scanning aims to identify, differentiate, filter and prioritise new and emerging health technologies in order to assess or predict their potential impact on health, costs, society and the healthcare system.

In the long run, if horizon scanning is adopted in the Hong Kong healthcare system, the authorities concerned will be able to grasp the information on what new drugs are going to be registered in Hong Kong, specifications and clinical use of the drugs, etc.; and start financial planning early, which will enhance the effectiveness of drug inclusion and funding. Besides, the adoption of horizon scanning can also prompt the pharmaceutical companies to plan in advance and prepare the information and related data required for drug registration as early as possible, which will further accelerate the registration and drug inclusion process. Thus, patients can receive treatment as soon as possible. It will be a triple win situation for the Government, the business sector and the patients.

VII. Collaborate on rare disease diagnosis and treatment in the Greater Bay Area

The Central Government and various provinces and municipalities in the Mainland have launched many policy measures to improve the diagnosis and treatment of rare diseases in recent years. For instance:

- ✧ in 2015, the National Health and Family Planning Commission established the “Expert Committee on Rare Diseases Diagnosis, Treatment and Medical Security”, reflecting that rare diseases have entered the field of national policy;
- ✧ in 2016, the National Rare Diseases Registry System was established to consolidate national rare disease data;
- ✧ in 2018 and 2023, the NHC released the first and the second lists of rare diseases respectively, including a total of 207 rare diseases; and
- ✧ in 2019, the NHC established a national collaborative network for rare disease diagnosis and treatment which consists of over 300 hospital nationwide (13 of which are located in the Greater Bay Area, including the Hong Kong University-Shenzhen Hospital which is familiar to the people of Hong Kong), and published the first “*Guidelines for Diagnosis and Treatment of Rare Diseases*”.

Hong Kong has excellent healthcare facilities and human resources, but the Government has always lacked proactive and holistic strategy and long-term planning for the diagnosis and treatment of rare diseases. Cross-border collaboration, especially with the Greater Bay Area, will bring about complementary benefits to local healthcare services and patients.

The following are the preliminary recommendations for close collaboration between Hong Kong and the Greater Bay Area on rare disease diagnosis and treatment:

1. Designate some hospitals in Hong Kong (Hong Kong Children’s Hospital, Queen Mary Hospital or Princess Margaret Hospital may be considered) to participate in the national collaborative network for rare disease diagnosis and treatment, and work closely with the 13 designated rare disease hospitals in the Greater Bay Area in clinical diagnosis and treatment. There can be cross-border referrals as necessary and when conditions permit.
2. Include HA’s rare disease cases in the National Rare Diseases Registry System, and

encourage intensive cooperation and exchange of data and scientific research with the 13 designated rare disease hospitals in the Greater Bay Area.

3. The internationally recognised Clinical Trial Centres of the two faculties of medicine in Hong Kong should collaborate with the 13 designated rare disease hospitals in the Greater Bay Area to attract multinational and Mainland pharmaceutical companies to conduct clinical trials for innovative drugs in Hong Kong.
4. Review the differences between the drug regulatory systems in Hong Kong and the Mainland to facilitate effective coordination and promote integration.
5. Explore the pros and cons of working with the Greater Bay Area on medical insurance mechanisms, and explore the possibility of making strategic procurement of orphan drugs.

As cross-border collaboration between Hong Kong and the Greater Bay Area on rare diseases is a new thing, it is expected that the government departments and relevant experts will discuss and study the above recommendations as soon as possible, and launch initiatives as far as they are ready.

VIII. Include Herpes Zoster vaccination in the Vaccination Subsidy Scheme

In recent years, there has been increasing concern about the long-term burden of the healthcare system caused by herpes zoster (also known as shingles) and its related complications. According to publicly available information, the average health expenditure per outpatient amounted to over \$2,400, and the average health expenditure per inpatient amounted to about \$38,000.

Patients with rare diseases and immune system diseases such as rheumatoid arthritis, lupus erythematosus, psoriasis, blood cancer, etc. have poor immunity, and they are at higher risk of getting shingles.

The burning pain or tingling on the skin caused by shingles is often unbearable, and some studies even say that it is more painful than childbirth. Although most patients will get better within two to four weeks, about 30% of patients will subsequently develop *postherpetic* neuralgia, which will last for weeks or years, severely affecting the patient's physical, psychological and social health, as well as the quality of life; and some patients will get eye shingles that can lead to blindness. Compared with normal people, if those with weak immunity get infected with shingles, they are likely to develop more severe symptoms and complications like pneumonia, hepatitis, meningitis, etc., and therefore the risk of hospital admission is much higher.

At present, herpes zoster vaccination in Hong Kong is not subsidised by the Government, and the more protective recombinant zoster vaccine generally costs over \$5,000. RDHK recommends that the authorities include the herpes zoster vaccination in the Vaccination Subsidy Scheme and encourage the high-risk citizens to get vaccinated.

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