香港罕見疾病聯盟
Hong Kong Alliance for Rare Diseases

Written Submission to the HKSAR Government in response to the Public Consultation for the 2017 Policy Address and 2017–18 Budget

Hong Kong Alliance for Rare Diseases

(December 2016)

The Hong Kong Alliance for Rare Diseases (hereinafter "HKARD"), established in December 2014, is the first patients' group in Hong Kong comprising cross-rare-disease patients and their families with the support of experts and academics in the field. Its objectives are to spearhead and improve related policies and services, promote public education on rare diseases, and strengthen the community's support for patients, in order to ensure respect and protection for patients in terms of such fundamental rights as healthcare, social support, education and daily needs equal to other citizens.

Hong Kong not only boasts a world-class economy and fiscal reserves in excess of $800 billion, but also has relatively sound and fair medical and social welfare systems. This naturally is cause for pride to local government officials. However, once the harsh realities of healthcare and social support for rare-disease patients are juxtaposed with existing systems and policies, the rigid and apathetic, if not cold and heartless, side of the systems is completely exposed. The way the government has been handling rare diseases has not only fallen short by failing miserably to address patients' needs, but also pales into insignificance against the commitment to serving such patients in neighbouring regions.

HKARD wrote two submissions in relation to the Policy Address and the Budget in 2015 and 2016 respectively to put forward rare-disease patients' demands, but has not received any response ever since. This year, HKARD is making this submission to reflect

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the gap between patients' demands and existing systems and policies. Our requests fall under the following eight areas:

1. Diagnostic Tests
2. Drug Treatment
3. Patients Registry
4. Prevention and Screening
5. Case Managers
6. Application for Comprehensive Social Security Assistance (CSSA)
7. Employment Incentive
8. Respite Services

We hope the government will listen and respond to us this time.

1. Healthcare:

Approximately 80% of rare diseases are genetic in origin. A patient may display systemic disorders in all their body structures and organs, causing serious physical and intellectual disabilities or even death. Cases of such diseases are rare, and clinical data and evidence are insufficient. Diagnosis is time-consuming and complex and requires the support of all sorts of specialists.

The recurrent expenditure on medical and health in 2016–17 was $57 billion. However, in recent years, investment in healthcare has been increasingly determined by such criteria as patient numbers, clinical evidence and cost-effectiveness. When such criteria are given precedence over citizens' lives and health, there are bound to be patients neglected by the system. The condition of rare-disease patients is a case in point.

1.1 Diagnostic Tests:

Compared with other diseases, diagnosis of rare diseases is much more time-consuming and may, in certain cases, take up to five years or even over a decade. In the meantime, patients and carers have to waste much time and energy toing and froing

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between home and the hospitals and specialist clinics. It is not difficult to imagine the mental stress brought on by all the pent-up frustrations.

The diagnostic process involves lots of tests. As some of the test items are not available in local medical institutions, patients have to go abroad for the tests. The high costs involved may not be affordable for all patients' families.

At present, while drug treatment is available for only 500-plus rare diseases or 7% of the total number of such diseases, confirming a patient's condition as soon as possible is conducive to maintaining proper healthcare and quality of life for patients. Therefore, better-coordinated multi-specialist diagnosis is an important aspect of healthcare for rare-disease patients. HKARD's requests in this respect are as follows:

– To set up clinical genetic services at Hong Kong Children's Hospital, which is under the governance and management of the Hospital Authority, to provide better-coordinated multi-specialist diagnosis for suspected cases of rare disease.

– To ensure provision of specialist service for patients with suspected rare disease to save patients and carers the trouble of toing and froing between hospitals.

– To entrust Hong Kong Children's Hospital with the task of compiling data on suspected rare-disease cases to provide cost-effective systematic tests.

– To entrust Hong Kong Children's Hospital with the task of coordinating the shipment of items abroad for tests, with costs of which to be borne by the Hospital Authority.

To address the requests above, the Hospital Authority should create clinical genetics-related medical staff posts, and provide the staff with proper training and professional development opportunities so as to build up a talent pool for diagnosis and treatment of rare diseases.

1.2 Drug Treatment:

To prolong their lives, keep disorders in check, and relieve symptoms, patients may need to use costly drugs on a long-term basis. However, most patients will not be able to afford such drugs in the long run.

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Subject to the simple, paradigmatic thinking along the lines of patient numbers, clinic evidence and cost-effectiveness, the Hospital Authority has so far only relied on an expert panel for rare genetic diseases to provide subsidised treatment, based on assessment on the clinical conditions of individual patients, for 6 enzyme-replacement drugs and 2 rare cancer drugs. While subsidy is available for patients as well as the drugs elsewhere, the patients in Hong Kong are still a long way from being able to get similar support. What awaits them is the prospect of serious physical and intellectual disabilities and death.

Take Taiwan for example, in 2015, the National Health Insurance Administration paid a total of NT$4.1 billion (approximately HK$1 billion) for rare disease drugs, representing around 0.27% of health insurance medical expenses totalling NT$150 billion, benefitting 7,625 patients in all. Taiwan's population is roughly three times that of Hong Kong. Based on these figures, should Hong Kong's annual expenditure on rare disease drugs match that of Taiwan, it would only be around HK$300 million, representing merely 5% of the Hospital Authority's total dispensing expenditure on drugs for 2015–16 (HK$5.71 billion in total). Yet as many as some 2,500 patients would stand to benefit from this measure as a result.

HKARD's requests in this respect are as follows:

– To make better use of the $45 million fund earmarked each year by expanding the current 6 enzyme disorder drugs and 2 rare cancer drugs to cover other rare diseases.

– To expand coverage of the Health and Medical Research Fund (HMRF) under the Food and Health Bureau to include a rare-disease medication clinical research project. With an aim of building clinical evidence and data, the study will encourage and enable medical schools and the Hospital Authority to carry out clinical trials on rare disease medications.

– To ensure a more active utilization of the Community Care Fund by using it to sponsor rare disease medications with initial evidence to further collect and build clinical evidence.

– To set up an "orphan drug" mechanism taking into account the scarcity of rare-disease cases and clinical evidence, the costly nature of rare-disease medications, the clinical evidence requirement for rare-disease medications, and the special threshold for

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the means test for sponsorship (the Community Care Fund and the Samaritan Fund) of patients.

1.3 Patients Registry:

At present, Hong Kong is lacking in a comprehensive set of evidence on rare-disease epidemiology, and clinical and medical economics. All the universities, medical departments and patients' groups have their own sets of data, making it difficult to estimate the overall medical and social burdens on patients. This is cited by the government and the Hospital Authority as a reason for their unwillingness to provide evidence-based treatment and healthcare for rare-disease patients.

Given that quality medical service and diagnosis hinge on accurate recording of genetic information and incidence rate, it goes without saying that compiling a patients registry is indispensable when it comes to dealing with rare diseases. An integrated and comprehensive patients registry, incorporating a wealth of pertinent experience in rare-disease diagnosis and treatment results, is not only part and parcel of a standard public health policy. It is also conducive to clinical tests and other patients-related studies and to long-term improvements in clinical study and patients' condition.

HKARD's requests in this respect are as follows:

– The government should earmark resources in the next financial year to enable the Children's Hospital (managed by the Hospital Authority) to take the lead in coordinating a joint effort by the two medical schools in Hong Kong, in collaboration with the other hospitals, to pilot a rare-disease patients registry for selected rare diseases.

– To make effective use of an electronic health-record sharing platform to gradually build a comprehensive rare-disease patients registry.

1.4 Prevention and Screening:

Newborn screening tests can detect whether newborns have any genetic or metabolic disorders likely to compromise their long-term health or cause death, and are conducive to timely discovery, diagnosis, and intervention, thus preventing severe complications, disability, and death.

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In the 2015 Policy Address, it was announced that a pilot screening programme for newborn babies for inborn errors of metabolism (IEM) would be launched. Introduced in October 2015, the pilot scheme covered only Queen Elizabeth Hospital and Queen Mary Hospital. During the first 6 months (the first phase of the scheme), 21 kinds of IEM were tested, and 3 more were included in the second phase.

HKARD's requests in the respect are as follows:

– To provide for acceptance of cross-cluster referral arrangements before expanding the screening programme, and to sponsor, through the public-private partnership model, the screening of newborns at The Chinese University of Hong Kong and other private medical institutions that offer self-financed services, so that the programme can benefit more newborn babies.

– To capitalize on technological advances and review the number of items to be screened under the programme from time to time to ensure timely diagnosis and intervention for more rare diseases.

– The government should not only further expand the newborn screening programme and launch the expanded programme in phases, but also make newborn screening a standard procedure in hospitals when the Children's Hospital becomes operational in 2018.

2. Social Support:

Like all other local citizens, every rare-disease patient should be entitled to the rights to live with dignity and enjoy quality of life. To protect these rights, proper and efficient social support is indispensable to all patients.

2.1 Case Managers:

Rare-disease patients require all kinds of social support when it comes to medical consultation, follow-up consultation, rehabilitation, and other daily needs. All the complex procedures and sequences are the cross to bear for patients and their families.

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There are all kinds of rare diseases and most rare-disease patients suffer different degrees of disorder in their organs and bodily functions. They are likely to go back and forth between at least several if not a dozen specialists at the hospital. Follow-up consultation alone takes extra time and effort, not to speak of booking the appointments in advance, and all this seriously compromises their quality of life.

HKARD would like to make the following suggestions in this respect:

– To pilot and gradually promote a "Rare-disease Patient Whole-person Case Manager" service to take care of patients' whole-person support needs in terms of healthcare, follow-up consultation, rehabilitation, schooling, employment, marriage, community life, and mental health, and to produce relevant guidelines for proper implementation by front-line Case Managers.

– The main responsibilities of Rare-disease Patient Whole-person Case Manager are: evaluating the medical and social support needs of patients and their families; arranging and coordinating various interdepartmental and cross-professional social support services; and making service adjustments in line with changing needs of patients.

Take arrangements for above-mentioned follow-up consultation for example, a Case Manager should first and foremost have a general idea of patient's condition, the specialists involved in the consultation process, and patient's daily schedule, so as to be able to properly handle advance booking of follow-up consultation appointments and avoid exhausting the patient during the inevitable toing and froing for the appointments. The Case Manager should take the initiative to communicate with medical staff of the specialists and, after devising personalized care solutions based on individual patients' needs, coordinate implementation of the solutions by the specialists and departments involved. Similarly, the Case Manager has the same role to play in other aspects of patient's life in terms of daily care, schooling, employment, mental health, and community life, etc.

The Social Welfare Department launched the Handbook on Case Management Service in September 2016. HKARD suggests that the government should use this as a blueprint and, after optimizing it based on patients' needs, pilot the proposed "Rare-disease Patient Whole-person Case Manager" service as soon as possible. HKARD should be glad to provide further information and specific suggestions in this respect.

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2.2 Optimizing the CSSA Mechanism:

In addition to basic needs expenses, long-term medical equipment and life-support medical equipment, which easily cost thousands or even tens of thousands of dollars per month, form another economic burden around patients' necks. This is often the last straw that leaves patients no choice but to apply for CSSA, causing them to rely on the safety net to support their life-saving expenses.

Under the present requirements of the CSSA Scheme, not only must patients submit their applications together with their families as joint applicants, all the family members living together are also subject to an asset test, and receive CSSA payments in accordance with CSSA ordinance provisions. In other words, patients are not allowed to apply for CSSA individually and family members living together are discouraged from working because of the CSSA income limit. While seriously affecting family income, this will put social welfare funds under further strain. To avoid compromising their families, patients are forced to file transfer applications or move to residential care homes. To cover their life-saving expenses, they have no choice but to trade their right to live with their families for the right to apply for CSSA. This goes to show how the rigid application procedures ride roughshod over the social needs of patients without regard to their right to choice over their community life.

HKARD would like to make the following suggestion in this respect:

– To allow patients living with their families to apply for CSSA individually. This would satisfy the need of patients for support for their life-saving expenses without compromising their families' living standards and potential productivity.

2.3 Providing Employment Incentive:

With increasing access to education, most rare-disease patients have graduated from secondary school and some of them may even have completed tertiary or a higher level of education. They would have been motivated to get a job to support themselves and live independently had they not been bogged down by the huge expenses on peripheral equipment required for life support. Large equipment such as medical bed, custom-made mattress, wheelchair, suction machine, and ventilator, etc., needs to be replaced within a few years. With wages earned from employment, they will exceed the

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income limit for subsidies. As for charitable funds in the community, the time-nsuming application process and long wait for payments simply become too prohibitive. To secure subsidy for the life-saving medical equipment, patients have no choice but to give up employment and hang desperately on to the CSSA safety net.

Evidently, under the existing system, there is no medical life support to complement CSSA, making it impossible to address the need for social support for patients with both the intention and the ability to work. If feasible transition solutions can be worked out to cover the expenses on medical life support, thus creating an employment incentive, patients would be in a better position to get a job with higher pay. This way they would not only be able to fend for themselves, but they would also find it possible to achieve self-realization and improve their upward mobility efforts.

HKARD would like to make the following suggestions in this respect:

– To set up a "Life-support Medical Equipment and Rehabilitative Aids Subsidy Scheme" to offer proper support to patients who are employed but are unable to afford expenses on the equipment and aids, so as to alleviate their financial problems in this respect and provide them with an employment incentive.

– The Community Care Fund has launched a three-year "Pilot Scheme on Raising the Maximum Level of Disregarded Earnings for Recipients with Disabilities under the Comprehensive Social Security Assistance Scheme". However, the Pilot Scheme only covers CSSA recipients and does not address the needs of people outside the safety net who are employed but in need of help. Apart from offering employment incentive within the safety net, the government should look beyond the net and focus more on helping the rare-disease patients and other chronic patients who are outside the net but have similar needs for healthcare. A bridging programme with the same philosophy as the Pilot Scheme will lighten the financial burden of the patients who want to earn their own living.

2.4 Optimizing Respite Services:

Young patients with disabilities resulting from rare diseases are usually referred to special schools, which do not necessarily provide hostel accommodation. After leaving the schools, typically they have to wait around 6 years before being admitted to residential care homes for persons with disabilities. Owing to the turn-taking arrangements for taking care of the young patients as well as their own work

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commitments or occasional sickness, parents or carers may not be on hand to attend to atients' daily needs 24 hours a day and 7 days a week.

The home-care service launched by the Social Welfare Department in recent years, to a certain extent, serves to address the needs of rare-disease patients' carers. But still the service falls short in a number of ways. For example, it is necessary to book one month in advance, daily service hours are limited to 8 a.m. to 6 p.m., and home workers are not in a position to help patients with emergency situations, etc. These are some of the areas that remain to be optimized.

HKARD's requests in this respect are as follows:

– To introduce contingency booking to provide respite services at short notice to take the place of carers in case of emergency.

– To provide such services 24 hours a day and 7 days a week, so that parents and carers of patients unavailable to help during holidays or at night can have peace of mind.

– To improve carers' emergency handling skills, e.g., the ability to administer first aid before ambulance arrival in case a patient suffers muscle spasms and shortness of breath.

3. Conclusions:

When it comes to rare diseases, the government and the Hospital Authority have always maintained that healthcare in Hong Kong covers all kinds of diseases and there is no need to distinguish between rare and common diseases.

The logic of the government and the Hospital Authority is simple and easy to understand because to all patients seeking health-care services, no public hospital (at least officially) will say no. However, given the rigidity and apathetic side of the existing system and policies, this is tantamount to sidestepping and neglecting the needs and requests of rare-disease patients, shutting the door to any opportunities the patients might have of finding the help they need.

As outlined in this submission, rare-disease patients possess a number of characteristics that are absent in the vast majority of other patients. They include: potential disorder in body structures and organs causing serious disabilities or even death; scarcity of rare-disease cases and of clinical data and evidence; and time-consuming and complex diagnosis involving support from all kinds of specialists. To prolong their lives, keep the disorders in check, and relieve the symptoms, many rare-

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disease patients are reduced to long-term dependence on costly drugs beyond their means. The varied nature of health-care and social-support needs of different patients is such that personalized and properly-coordinated multidisciplinary support and services are indispensable.

Rigid adherence to such criteria as patient numbers, clinical data and evidence, and cost efficiency in the allocation of healthcare resources are now the order of the day in Hong Kong. When policymakers give precedence to such criteria over patients' health and lives, rare-disease patients are practically denied healthcare services, thus becoming virtual orphans of the system. The social support mechanism has also become so rigid and automatous that it has failed to protect patients' dignity and their right of quality of life.

The United Nations proclaimed the Universal Declaration of Human Rights in 1948 and further elaborated its principles in international treaties covering the rights of such groups as women, children, and people with disabilities in the following decades. This goes to show that different groups have different characteristics. In addressing their individual characteristics, while universal norms apply, it is also necessary to devise specific norms for specific groups.

That is why HKARD has been calling on the government to devise specific policies for rare diseases in Hong Kong. Since the existing system and policies have failed to address rare-disease patients' characteristics, the health and lives of such patients have not been accorded sensible and reasonable attention in equal measure to those of other local citizens, nor have such patients received the same level of dignity and quality of life. To tackle the above issues head-on, devising specific policies in a sensible and reasonable manner, culminating in a clear definition of rare diseases, though not necessarily the only viable option, is definitely a great start.

We hope the government will give careful consideration to our requests and suggestions above and offer us a favourable response.

– End –

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