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Explanatory Notes for the Draft Rare Diseases Bill

1. A rare disease is any uncommon disease with a very low prevalence rate. There are over 6,000 diseases identified as rare diseases worldwide. Most rare diseases are caused by genetic defects or mutation and are found during childhood or even early infancy. In most of the cases, they pose serious threat to a patient's entire life with extreme disabilities affecting quality of life. Some of them are even degenerative and life-threatening.

2. The definition for rare diseases differs among different organisations and jurisdictions, as follows: less than 7 in every 10,000 in the US; less than 5 in every 10,000 in the European Union (EU); and less than 1 in every 10,000 in Taiwan.

3. Hong Kong has in place a world-renowned public health policy, and a healthcare system of high standard. However, the Government has not established any official definition of rare diseases, nor has it set out any specific policy on provision of support for rare disease patients. Indeed, some key stakeholders (particularly patient groups) have criticized the Government for being unable to provide adequate support to rare disease patients, as evidenced by (a) the delay in the time to diagnose rare diseases; (b) limited number of patients receiving subsidies to help cover the high-cost medication; (c) the lack of a comprehensive patient registry to facilitate the provision of evidence-based treatments to patients; and (d) insufficient provision of social care services to patients and their carers.

4. Given the diseases' rarity and a lack of awareness and experience in clinical genetics, many patients have to go through a prolonged period of clinical investigations and follow-ups by different specialists in different hospitals before a proper diagnosis is made. Due to insufficient facilities, some laboratory investigations for the diagnosis of rare diseases are not available in Hong Kong. Patients' specimens need to be frozen and sent to overseas laboratories for examination, which is very costly and time-consuming. Frozen specimens also reduce the accuracy of the test. Unable to afford the high cost of investigations, many patients are made to suffer hopelessly from the delay of diagnosis, lack of proper treatment, and deteriorating health.

5. In Hong Kong, "clinical genetics" is not yet included in the list of specialists by the Medical Council of Hong Kong. Therefore, at present, no doctor can claim to be a specialist doctor in clinical genetics. Limited public resources are apportioned for research and development on rare diseases, leading to inadequate medical experience, and lack of timely, accessible and proper medical support. About 80% of rare diseases are genetic in origin, but there are grossly insufficient specialists in this field.

6. Without an official definition in Hong Kong, the needs of rare diseases patients are often neglected. Many of them cannot benefit from the current safety net namely the Samaritan Fund, or denied access to publicly-funded medication. There are over 6,000 types of rare diseases identified in the world, it is estimated that there are thousands of rare disease cases in Hong Kong. The authority has yet to provide more support to rare diseases patients.. At present, only patients from 6 diseases are receiving support from the Expert Panel on Rare Metabolic Diseases , often through stringent criteria, yearly case by case review scheme. The majority of rare disease patients cannot benefit from the existing mechanism.

7. The rarity of rare diseases has led to small number of patients using orphan drugs. Although some pharmaceutical companies are willing to invest on R&D, orphan drugs are very costly and most families cannot afford to pay for these drugs. Most rare diseases cannot be cured and patients require continuous medication to manage and control the conditions, sometimes at higher than normal dosage. But these supporting drugs are not included in the HA Drugs Formulary.

8. According Audit's reports No. 67, the Audit Commission pointed out that only a few HA hospitals and clinics, mainly the leading hospitals, had regularly applied for new drug listing. During the audit visit to a medium-sized hospital, Audit was informed that the hospital had never applied for new drug listing. Audit also noted that no applications for listing on the HADF had been made for 45 non-HADF registered drugs used by public hospitals and clinics in 2015-16, although some were in regular demand.

9. Besides, the Audit Commission observed that applications for new drug listing not made for many non-HADF drugs in regular demand.Of the 95 non-HADF registered drug items (involving 73 drugs) used in 2015-16, applications for new drug listing had not been made for 57 items (involving 45 drugs — see para. 2.11(a)). Audit noted that 12 of these 57 drug items, being in regular demand, had been acquired through bulk contracts (standing offer agreements) with drug suppliers over a one-year period (see para. 3.5). 2.34 In Audit's view, for non-HADF drugs intended to be used for an extended duration, the due process for putting up the drugs for listing on the HADF should be followed. The listing of cost-effective drugs of proven safety and efficacy on the HADF helps ensure that patients attending different public hospitals and clinics have equitable access to the drugs (see para. 2.28 for the audit recommendations on non-HADF drugs).

10. Discrimination and misunderstanding on rare diseases can isolate the patients. Patients not only suffer from sickness and lack proper, timely diagnosis and support, but also have to deal with sense of guilt, despair and harsh feelings, and adverse impact on their employment and study, or even marital, social and psychological problems. Some have to give up their jobs, which will worsen their financial situations, and psychological health. A HKU scholar once indicated that, the unemployment and poverty rates are much higher among families with rare disease patients than average level.

11. Different overseas places have passed legislation to promote development of treatments for rare diseases. For example, US was the first to pass specific legislation (the Orphan Drug Act) designed to promote development of treatments for rare diseases. The Act contains provisions governing the designation of orphan drugs and granting of incentives and assistance in the regulatory process to encourage pharmaceutical companies to develop orphan drugs. Subsequently, Japan, the EU and Taiwan have also passed legislation to establish a similar orphan drug designation system and provide incentives to address the issue about the limited availability of drugs.

12. As a contracting region to the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD), Hong Kong has a responsibility to formulate and implement relevant policies as required by the Convention. According to the Convention Article 25, "States Parties recognize that persons with disabilities have the right to the enjoyment of the highest attainable standard of health without discrimination on the basis of disability. States Parties shall take all appropriate measures to ensure access for persons with disabilities to health services that are gender sensitive, including health-related rehabilitation."

13. The International Covenant on Economic, Social and Cultural Rights also includes basic obligations of contracting regions regarding the enjoyment of the highest attainable standard of health. Article 12 points out that

> "1.The States Parties to the present Covenant recognize the right of everyone to the enjoyment of the highest attainable standard of physical and mental health.
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> 2. The steps to be taken by the States Parties to the present Covenant to achieve the full realization of this right shall include those necessary for:
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> (a) The provision for the reduction of the stillbirth-rate and of infant mortality and for the healthy development of the child;
>
> (b) The improvement of all aspects of environmental and industrial hygiene;
>
> (c) The prevention, treatment and control of epidemic, endemic, occupational and other diseases;
>
> (d) The creation of conditions which would assure to all medical service and medical attention in the event of sickness. "

14. The object of this Bill protect and promote the right to health of persons afflicted with rare disease so timely access to health information and adequate medical care are available for persons afflicted with rare disease, as with any others; and to provide the premises for a comprehensive and integrative policy targeting to prevent, diagnose and cure rare diseases as well as to ensure the well-being of persons afflicted with rare disease so that rights of a person afflicted with rare disease guaranteed under the United Nations Convention on the Rights of Persons with Disabilities can be properly achieved.

15. The proposed legislative provisions deal with the following areas of rare diseases: (a) Commission on Rare Diseases Policy, (b) Evaluative Committee on Rare Diseases (c)Recognition of a disease or malfunction as rare disease, (d) Register of rare disease drugs, rare disease treatments or rare disease products, (e) a statutory scheme of subsidy to subsidize costs a person afflicted incurred for a rare disease drugs or rare disease treatments, and (f) Rare Disease Information System.

16. Commission on Rare Diseases Policy is to perform functions include: (a) to advice the Government in the strategic development of the policy concerning rare diseases; (b) to monitor the implementation of the policy concerning rare diseases; (c)to monitor the work of Evaluative Committee on Rare Diseases, and (d) when required by the Chief Executive to do so, to provide to the Chief Executive in Council a report and its recommendations about the policy concerning rare diseases.

17. Evaluative Committee on Rare Diseases is set up for functions include:(a) to evaluate any disease or malfunction on its own motion or on application, so as to determine if that disease or malfunction qualifies as a rare disease for the purpose of this Ordinance; (b) for any disease or malfunction that qualifies as rare disease, to make recommendation to the Food and Health Bureau for its recognition; and(c) to maintain a register of rare disease drugs, rare disease treatments or rare disease products.

18. Recognition of a disease or malfunction as rare disease will be recommended if, and only if (a) it is clinically definable; and (b) it affects no more than one in 10,000 individuals in Hong Kong. Where a disease or malfunction is clinically definable but do not have any applicable clinical data concerning the Hong Kong population, the Committee is entitled to (a) analyse and consider any data or situation concerning the same disease or malfunction in other countries; and (b) recommend the disease or malfunction to be provisionally recognised as rare disease for a specified term of not more than 5 years.

19. When a drug, treatment or product have indications for the cure or alleviation of a rare disease or its symptoms, the Committee may register that drug, treatment or product as a rare disease drug, rare disease treatment or rare disease product.Before any drug, treatment or product is registered as a rare disease drug, rare disease treatment or rare disease product, the Committee must be satisfied as to its safety, quality and effectiveness.

20. The Government is responsible for launching and maintaining a scheme to subsidize the expenses on rare disease drug or rare disease treatment of a person afflicted with rare disease. The said scheme must endeavour ensure the provision of safe, quality, effective and affordable rare disease drugs and rare disease treatment to a person afflicted with rare disease, whereas cost-efficiency shall not be the foremost concern.

21. The Secretary for Food and Health must establish and maintain a Rare Disease Information System. The Rare Disease Information System must include, inter alia, —(a) an updated list of all rare diseases; (b) data on the prevalence of rare diseases and demographic information of persons afflicted; (c) data on the use of rare disease drugs, rare disease treatments or rare disease products.

22. In conclusion, the currently proposed legislative provisions will be the important step to protect and promote the right to health of persons afflicted with rare disease. It is hoped that further support measures will be explored , to enhance the awareness, early identification, prevention and treatments of rare diseases.