Research Office
Legislative Council Secretariat

Fact Sheet

Supplementary information on rare disease policies in selected places

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1. Introduction

1.1 Rare diseases are a large group of diseases that are characterized by a low prevalence in the population.[1] While individually rare, there are more than 7 000 rare diseases collectively affecting 1 in 15 persons worldwide.[2] While nearly all genetic diseases are rare diseases, not all rare diseases have a genetic origin since they also cover very rare forms of infectious diseases, auto-immune diseases and rare cancers.[3]

1.2 Rare diseases are serious chronic diseases and are often life-threatening. They pose challenges to the medical sector in view of the diverse types of rare disease conditions that exist, insufficient knowledge or training on the diseases, and the high costs and risks of research and development of drugs (commonly referred to as "orphan drugs") for treating the diseases given the small and dispersed patient base for each disease. As a result, medical care for patients suffering from rare diseases may be hampered by delayed diagnosis, unavailability of treatments, and/or limited access to costly drugs and treatments. These patients and their families are facing immense physical, psychological and economic burdens.

1.3 At its special meeting on 11 April 2017, the Panel on Health Services requested the Research Office to study (a) the policy on the provision of support measures for rare disease patients in overseas places; and (b) the rare diseases covered under their orphan drug designation systems to facilitate comparison with the rare diseases/uncommon disorders covered under the drug subsidization schemes in Hong Kong. This fact sheet studies the United States ("the US"), the European Union ("the EU"), Australia, Japan,

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1 Rare diseases are often referred to as orphan diseases due to the lack of financial incentives for pharmaceutical companies to develop drugs that would treat a small target patient population.

2 See De Vrueh, R. et al (2013), Shafie, A. et al (2016) and Song, P. et al (2012).

3 See De Vrueh, R. et al (2013).

Taiwan and South Korea which have devised medical care policies over the years to address issues faced by rare disease patients. The paragraphs below give an overview of the rare disease policies in Hong Kong and the overseas places studied, and the salient features of the latter are compared in the **Table**.

2. Rare disease policy in Hong Kong

2.1 In Hong Kong, the Clinical Genetic Service of the Department of Health provides clinical diagnosis, counselling and prevention services for families possibly affected by genetic related diseases. Meanwhile, the Hospital Authority provides medical services for patients suffering from genetic diseases. Furthermore, the Department of Health and the Hospital Authority started the Pilot Study of Newborn Screening for Inborn Errors of Metabolism in October 2015, in an effort to prevent and reduce severe problems arising from inborn errors of metabolism. The number of inborn errors of metabolism covered under the Pilot Study increased from 21 to 24 in April 2016.

2.2 Since 2008-2009, the Government has also subsidized patients who suffer from six specified types of lysosomal storage disorder[4] and meet the specific clinical criteria to obtain enzyme replacement therapy. Recently, the Government has planned to provide drug subsidies to eligible patients suffering from specified rare diseases/uncommon disorders (e.g. Paroxysmal Nocturnal Haemoglobinuria) through the Community Care Fund.

2.3 Notwithstanding the above-mentioned medical services provided, 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.[5]

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[4] The six types of lysosomal storage disorders are Gaucher disease, Pompe disease, Mucopolysaccharidosis Type I/Type II/Type VI and Fabry disease. Up to December 2016, the Hospital Authority had provided enzyme replacement therapy to 27 patients with lysosomal storage disorders.

[5] See Minutes of Meeting of the Panel on Health Services of the Legislative Council (2014) and Hong Kong Alliance for Rare Diseases (2016).

3. Rare disease policies in selected places

3.1 The US, the EU, Australia, Japan, Taiwan and South Korea have devised policies over the years to address issues faced by rare disease patients. These places have set out a definition of rare diseases and put in place an orphan drug designation system to encourage the development of orphan drugs for treating rare diseases. They have also implemented other measures to support the medical and/or social care of rare disease patients under their respective policy framework.

Definition of rare diseases

3.2 All the overseas places studied have defined rare diseases in terms of a prevalence rate measured by the total number of rare disease patients (less than 200 000 in the US, fewer than 20 000 in South Korea and fewer than 2 000 in Australia), the number of rare disease patients in 10 000 persons (fewer than five in the EU and fewer than one in Taiwan), or the proportion of rare disease patients in the total population (less than 0.1% of the population in Japan). In addition, Japan, Taiwan and South Korea have considered other criteria in their definition of rare diseases such as diseases that are difficult to diagnose and treat, or no appropriate treatment is available.

Orphan drug designation system

3.3 Among the overseas places studied, the US was the first to pass specific legislation (i.e. 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. Following the US, Japan, Australia, the EU, Taiwan and South Korea 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 and treatments for rare diseases. The incentives granted by the overseas places studied generally include financial subsidies and tax relief on research expenses, fast-track marketing approval process, reduced or waived application fees, and marketing exclusivity for a certain number of years after obtaining marketing authorization of an orphan drug.

3.4 As a result of the introduction of the orphan drug designation system, all the overseas places studied have experienced increased orphan drug development activities and greater availability of authorized/approved orphan drugs for treating rare diseases. For example, between 2000 and 2016, the EU has designated 1 805 orphan drugs and granted marketing authorizations to 128 orphan drugs for treating 101 conditions. Likewise in the US, the number of designated orphan drugs totalled 4 078 as at May 2017. The orphan drug designation system has also successfully enabled the development and marketing of over 600 drugs and biologic products for rare diseases since 1983. In contrast, fewer than 10 such products supported by industry came to market between 1973 and 1983.

3.5 The orphan drug designation systems in all the overseas places studied cover all of the seven uncommon disorders that are or to be covered under the drug subsidization schemes of the Hong Kong Government.[6]

Other support measures for rare disease patients

3.6 In addition to the orphan drug designation system, all the overseas places studied have adopted one or more of the following common measures to enhance the awareness, early identification, prevention and treatments of rare diseases: (a) providing relevant information on rare diseases through an online information centre; (b) implementing a newborn screening programme; (c) establishing a patient registry or a repository of registries to facilitate information sharing for patient care and research purposes; [7] and (d) committing resources on research and development on rare diseases. It is noteworthy that Japan and Taiwan have specifically addressed the need for social care services among rare disease patients in their policy framework. In particular, they have expanded the definition of persons with disabilities to cover patients suffering from designated rare diseases and provided them with social care services that are stipulated in the relevant legislation.

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[6] Comparison made is based on the latest information available on the orphan drug designation systems of the respective places.

[7] In Australia, there have been calls for the federal government to formulate a comprehensive national plan on rare diseases covering the establishment of a national registry and provision of coordinated care to rare disease patients. At present, some research institutions are collecting rare diseases patients' information but none of them is reportedly resourced adequately.

Table – Rare disease policies in selected places

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Definition and prevalence of rare diseases						
Definition of rare diseases in terms of prevalence criterion	• Less than 200 000 patients.	• Fewer than five in 10 000 persons.	• Not more than 2 000 individuals at any time.[8]	• Less than 0.1% of the country's population.	• Fewer than one in 10 000 persons (less than 0.01%).	• Fewer than 20 000 patients.
Other criteria considered for designation of rare diseases	• Not specified.	• Not specified.	• Not specified.	• Other criteria include: (a) causes of diseases not being identified; (b) lacking effective treatments; (c) requiring long-term treatments; and (d) existence of objective diagnostic criteria.	• Other criteria include: (a) having a genetic origin; and/or (b) being difficult to diagnose and treat.	• No appropriate treatment is available.
Number of rare diseases/ designated rare diseases affecting the population	• About 7 000 rare diseases.	• About 5 000 to 8 000 rare diseases.	• Information not available.	• 330 designated intractable/rare diseases.	• About 210 designated rare diseases.	• Over 110 rare diseases.

[8] The Australian Government Department of Health is reviewing the rare diseases threshold under its Orphan Drug Program with a view to broadening the scope of medications that is qualified for orphan drug status. Besides, other proposals such as the introduction of additional eligibility criteria for designation of orphan drugs and the modification of the designation process have also been considered under the review.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Definition and prevalence of rare diseases (cont'd)						
Prevalence of rare diseases	• Between 25-30 million persons in the US suffering from rare diseases.	• About 30 million persons in the EU suffering from rare diseases.	• An estimated 1.2 million persons in Australia suffering from rare diseases in 2015.	• 943 460 patients in Japan suffering from designated intractable/rare diseases as at end-2015.	• 7 625 patients in Taiwan suffering from designated rare diseases in 2015.	• An estimated 500 000 persons in South Korea suffering from rare diseases in 2013.
Policy framework						
Responsible authorities	• The United States Department of Health and Human Services, the United States Food and Drug Administration, and relevant state authorities.	• European Medicines Agency and relevant authorities of individual member states.	• The Australian Government Department of Health and the Australian Government Department of Human Services.	• Ministry of Health, Labour and Welfare ("MHLW").	• Ministry of Health and Welfare ("MOHW").	• Ministry of Health and Welfare and Ministry of Food and Drug Safety.
Relevant legislation	• The Orphan Drug Act of 1983 and the Rare Diseases Act of 2002.	• The European Union Regulation on Orphan Medicinal Products.	• Therapeutic Goods Regulations 1990 as amended in 1997 to provide for the establishment of the orphan drug designation system.	• The Pharmaceutical Affairs Act; and the Act on Medical Care and Social Supports for Patients with Intractable/Rare Diseases.	• Rare Disease and Orphan Drug Act《罕見疾病防治及藥物法》.	• Regulation on Designation of Orphan Drugs.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Policy framework (cont'd)						
Policy scope identified	• Promoting development of orphan drugs. • Supporting research and development on rare diseases.	• Promoting development of orphan drugs. • Supporting member states to ensure their effective and efficient recognition, prevention, diagnosis and treatments of, and research on, rare diseases.	• Promoting development and marketing of orphan drugs.	• Developing effective treatments, and enhancing medical and social care services for rare disease patients. • Establishing a fair and consistent subsidization mechanism. • Enhancing public understanding of rare diseases.	• Improving the awareness, prevention, diagnosis, and treatment of rare diseases. • Enhancing medical and social care services for rare disease patients.	• Promoting development of orphan drugs. • Improving quality of life of patients with rare diseases. • Promoting research and development on rare diseases.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Orphan drug designation system						
Criteria of defining an orphan drug	• The drug is to treat a disease which (a) affects less than 200 000 persons in the US; or (b) affects more than 200 000 persons in the US but cost of developing the drug is not recoverable from sales in the country.	• The drug is intended for treating a life-threatening disease that meets the prevalence criterion and no satisfactory treatment is available.	• The drug is (a) intended to treat, prevent or diagnose a rare disease that affects not more than 2 000 individuals in Australia at any time; or (b) not commercially viable to supply to treat, prevent or diagnose another disease or condition.	• The drug must meet three criteria: (a) to be used by less than 50 000 patients in Japan; (b) indicated for the treatment of serious diseases and no alternatives are available; and (c) with a scientific rationale to support the need for the drug.	• The drug should have major indications for the prevention, diagnosis and treatment of designated rare diseases.	• The drug must meet the following criteria: (a) treating a disease which affects 20 000 patients or less in South Korea, or there is no available treatment for the disease in South Korea; and (b) the total production or sales value of the drug should be lower than the amount specified under the Regulation on Designation of Orphan Drugs.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Orphan drug designation system (cont'd)						
Provision of financial incentives and assistance in regulatory process to facilitate research and development activities	• Including financial subsidies/tax credit, fast-track marketing authorization and a seven-year period of marketing exclusivity.	• Including a 10-year period of marketing exclusivity, research grants and reduced fees for marketing authorization applications.	• Including waiver of fees for the application, evaluation and registration of drugs.	• Including financial subsidies/tax relief, a 10-year period of marketing exclusivity and fast-track marketing authorization.	• Including a 10-year period of marketing exclusivity, and allowance for special application for usage reimbursement of designated orphan drugs prior to market approval.	• Including reduced application fees for drugs developed domestically and a six-year period of marketing exclusivity for both domestically developed and imported orphan drugs.
Number of designated orphan drugs	• 4 078 as at May 2017.	• 1 805 between 2000 and 2016.	• 287 between 1998 and 2013.	• 318 as at May 2015.	• 98 as at January 2017.	• Information not available.
Number of designated orphan drugs granted with marketing authorization or approval	• 614 since 1983.	• 128 between 2000 and 2016.	• 144 between 1998 and 2013.	• 238 as at May 2015.	• Information not available.	• 341 as at 2016.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Orphan drug designation system (cont'd)						
Reimbursement of drug costs incurred by rare disease patients	• Drug costs are covered by (a) the public or private insurance schemes that the patients enrolled in; and (b) co-payments by patients.	• Reimbursement of drug costs is made in accordance with the healthcare financing systems and reimbursement arrangements put in place by individual member states.	• Patients who meet specific clinical criteria for using orphan drugs listed in the Pharmaceutical Benefits Scheme Schedule can recover part of the drug costs under the government-funded Pharmaceutical Benefits Scheme ("PBS"). • The Life Saving Drugs Programme provides patients, who suffer from eight specified types of rare diseases and meet specified eligibility criteria and conditions, with free access to 12 expensive and life-saving drugs that are not available through PBS due to failure to meet the cost-effectiveness criterion.	• Costs of using orphan drugs granted with marketing approval can be reimbursed under the health insurance system.	• Reimbursement of drug costs can be arranged for those drugs which are approved to be on the list of drugs covered by the National Health Insurance Administration. • Reimbursement of the costs of using unlisted drugs is allowed if approval is sought prior to drug usage.	• Costs of orphan drugs approved to be on the reimbursement drug list of the Korean National Health Insurance Service can be partially reimbursed.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Orphan drug designation system (cont'd)						
Coverage of the seven uncommon disorders which are/will be covered under the drug subsidization schemes of the Hong Kong Government ("the seven uncommon disorders")[9]	• All of the seven uncommon disorders are covered.	• All of the seven uncommon disorders are covered.	• All of the seven uncommon disorders are covered.	• All of the seven uncommon disorders are covered.	• All of the seven uncommon disorders are covered.	• All of the seven uncommon disorders are covered.

[9] Currently, the Hong Kong Government provides drug subsidies to eligible patients suffering from six specified types of lysosomal storage disorder, i.e. Gaucher disease, Pompe disease, Mucopolysaccharidosis Type I/Type II/Type VI and Fabry disease. It has also planned to provide drug subsidies to eligible patients suffering from Paroxysmal Nocturnal Haemoglobinuria through the Community Care Fund.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Other support measures for diagnosis and treatment of rare disease patients						
Information provided to enhance key stakeholders' awareness and knowledge of rare diseases	• Current and easy-to-understand information provided through the Genetic and Rare Diseases Information Center.	• Comprehensive and updated information provided through the Orphanet portal.	• Not specified under the national policy framework.	• Information provided through an online resource centre (i.e. the Japan Intractable Diseases Information Center).	• Information provided by MOHW through a specific portal and public education programmes.	• Information provided through a nationwide portal (i.e. the Helpline).
Facilitation measure for early identification of rare diseases	• Newborn screening programmes implemented by individual states.	• Newborn screening programmes implemented by individual member states.	• Newborn screening programmes implemented by individual states/territories.	• Not specified under the government's policy framework.	• Newborn screening programme covering 11 metabolism disorders.	• Newborn screening programme is implemented.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Other support measures for diagnosis and treatment of rare disease patients (cont'd)						
Facilitation measures for accessing medical care services	• Passage of the Patient Protection and Affordable Care Act in 2010 to remove various discriminatory insurance practices against rare disease patients.[10]	• Subject to the policy framework of individual member states.	• Not specified under the national policy framework.	• Patients only have to bear 20% of the medical costs as co-payment, capped at a monthly limit set by MHLW.	• Patients of designated rare diseases are provided with 80% reimbursement for medical and medication costs under the National Health Insurance programme. Low-income patients can receive 100% reimbursement. • Patients are subsidized to access overseas diagnostic services in case these services are not available locally.	• The government subsidizes medical expenses for low-income patients suffering from 133 specified rare diseases. • The government funds a regional hospital network dedicated to providing effective patient care and enhancing counselling services for genetic and rare diseases in four provinces.

[10] The Patient Protection and Affordable Care Act has been opposed by some stakeholders as it has brought about issues such as increase in insurance costs from better insurance coverage. Currently, the federal government has been reviewing the health insurance system with a view to repealing the Act.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Other support measures for diagnosis and treatment of rare disease patients (cont'd)						
Facilitation measures for accessing social care services	• Not specified under the government's policy framework.	• Not specified under the policy framework of the European Union.	• Not specified under the national policy framework.	• Provision of social care services through the intractable/rare diseases consultation and support centres. • Expanding the definition of persons with disabilities to cover persons suffering from most designated intractable/rare diseases and providing them with relevant social care services.	• Expanding the definition of persons with disabilities to cover persons suffering from designated rare diseases and providing them with relevant social care services.	• Not specified under the government's policy framework.
Establishment of a patient registry/ reporting system	• Yes, through the Global Rare Diseases Patient Registry Data Repository to store patient information from different registries set up by patient advocacy groups or researchers.	• Yes, member states develop their own patient registries.	• The federal government has not established a comprehensive rare disease registry.	• Yes.	• Yes.	• Yes.

Table – Rare disease policies in selected places (cont'd)

	The United States	The European Union	Australia	Japan	Taiwan	South Korea
Other support measures for diagnosis and treatment of rare disease patients (cont'd)						
Committing resources on research and development ("R&D") on rare diseases	• The Office of Rare Diseases Research under the National Institutes of Health is tasked to promote R&D on rare diseases.	• The European Commission funds collaborative R&D projects conducted by institutions across Europe and other countries.	• The Australian government sponsors the Australian Paediatric Surveillance Unit to conduct research for facilitating national surveillance of rare childhood conditions. • There have been calls for a coordinated research strategy and targeted research funding on R&D on rare diseases.	• MHLW commits resources on R&D projects related to rare diseases.	• MOHW provides incentives to encourage institutions to engage in R&D on rare diseases.	• The Ministry of Health and Welfare promotes research on rare diseases through (a) providing the infrastructures (e.g. establishing the Korean Mutation Database and organizing the clinical study networks); and (b) leading basic research.

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Research Office
Information Services Division
Legislative Council Secretariat
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Tel: 2871 2143