“Caring for Hong Kong Rare Disease Patients”

Our Position Paper

A. Rare Diseases

A rare disease is any uncommon disease with a very low prevalence rate. There are over 6,000 diseases\(^1\) 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.

The definition for rare diseases differs among different organisations and jurisdictions, as follows: 6.5 to 10 cases in every 10,000 by World Health Organization (WHO); 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. (See Appendix 1)

Some commonly known rare diseases identified in Hong Kong include Mucopolysaccharidosis, Pompe Disease, Infantile Cutaneous and Articular Syndrome, Tuberous Sclerosis Complex, Myelofibrosis, Marfan Syndrome, Spinocerebellar Ataxia, Muscular Atrophy, Fabry Disease, Gaucher Disease, Achondroplasia, various forms of Osteochondrodysplasias, Inborn Errors of Metabolism and Mitochondrial Diseases. Most of the rare diseases are genetic in nature. In spite of a very few reported cases, rare diseases present a real threat to patients’ health, and bring heavy burden to their families.

Hong Kong has in place a world-renowned public health policy, and a healthcare system of high standard. However, we lag far behind in terms of prevention, diagnosis, treatment and protection of rare disease patients, as compared with many countries and places in Europe, US, and Asia. Government has not come up with a clear

\(^1\) Rare Disease Day website [http://www.rarediseaseday.org/article/what-is-a-rare-disease](http://www.rarediseaseday.org/article/what-is-a-rare-disease)

European Organization for Rare Disease (EURORDIS) [http://www.eurordis.org/about-rare-diseases](http://www.eurordis.org/about-rare-diseases)
definition of rare diseases, or formulated related policies. The general public, including some healthcare professionals and social workers, have inadequate understanding and knowledge about rare diseases.

**B. Problems Faced by Rare Disease Patients in Hong Kong**

The Hong Kong Alliance of Patients’ Organizations held the first Roundtable Meeting on the Development of Rare Disease Policy in Hong Kong on the eve of Rare Disease Day 2014 (28th February 2014). The Meeting provided a platform for various stakeholders, including experts from medical, academic, social welfare and political sectors, as well as patients and caretakers, to exchange views. Various points of views were expressed during the Roundtable Meeting. The following is a summary of problems faced by rare diseases patients and their families (See Appendix 2 for more details):

1. **Long duration for diagnosis:** 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.

2. **Lack of clinical experience:** 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.

3. **Lack of support to rare diseases patients:** 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\(^2\) are receiving support from the Expert Panel on Rare Metabolic Diseases\(^3\), often through stringent criteria, yearly case by case review scheme. The majority of rare disease patients cannot benefit from the existing mechanism.

4. **High cost of medication:** 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.

5. **Economic burden and stress:** 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\(^4\). Some have to give up their jobs, which will worsen their financial situations, and psychological health. A HKU

\(^2\) Mucopolysaccharidoses I, II and VI, Pompe Disease, Fabry Disease, and Gaucher Disease

\(^3\) Hospital Authority Expert Panel on Rare Metabolic Diseases

http://www.info.gov.hk/gia/general/201012/08/P201012080320.htm

\(^4\) Speech of Professor Cecilia CHAN Lai-wan, Head and Professor, Department of Social Work and Social Administration, The University of Hong Kong in the First Round Table Meeting on the Development of Rare Disease Policy in Hong Kong
A scholar once indicated that, the unemployment and poverty rates are much higher among families with rare disease patients than average level.

C. Overseas Experience

Many countries had laid down clear definitions for rare diseases since the 1980s. They passed clear legislations and formulated evidence-based long-term policy, which enabled rare diseases patients to have proper treatment and timely care, in such areas as early approval of orphan drugs, subsidy and more.

US, EU and neighbouring jurisdictions, such as Singapore, Japan, Australia, Taiwan and Korea have adopted the following rare diseases policies: (1) Defining rare diseases; (2) Formulating supportive policy and measures for rare disease patients; (3) Establishing rare diseases database; (4) Requiring insurance schemes to cover the expenses of rare diseases treatment, and (5) establishing legal framework to encourage pharmaceutical companies to develop drugs for rare diseases. (See Appendix 3)

In 2000, Taiwan passed the Rare Disease Control and Orphan Drug Act. Then, regulations such as the Enforcement Rules of the Rare Diseases and the Orphan Drugs Act were also implemented to meet with the challenges of rare diseases. According to the People with Disabilities Rights Protection Act, rare diseases patients are entitled to enjoy the welfare, support and protection as a person with disabilities. As at 2013, 197 diseases were identified as rare diseases with a population of more than 4,000 patients in Taiwan. 78 drugs have been classified as orphan drugs and more than 1,000 patients are receiving the treatment. Through the National Health Insurance Scheme, low-income families would be fully subsidised with medical treatment, drugs and special nutrient products. Other families would get a subsidy to cover 80% of the expenses.

In 2003, Korea passed a legislation that defined rare diseases. Korea also issued a guideline on rare diseases, providing drug subsidy and funding a national survey on
rare diseases. At present, there are 500,000 \(^5\) rare disease patients in Korea. As the Korean National Health Insurance (NHI) covers rare disease cases, patients only need to pay for 10% of their medical expenses; the rest would be covered by NHI. People with low income would enjoy full coverage by NHI. In early 2014, the launch of a USD540 million post-genome project\(^6\) was announced to develop and commercialise new genomic technologies. (See Appendix 4)

Although there is no official definition of rare diseases in Mainland China, the Chinese National Birth Defects Monitoring Network\(^7\) had been established since 1986 to monitor rare diseases, including relevant prevention, diagnosis and research works. The government also drafted the Provisions for New Drugs Approval giving priority status to rare diseases-related new drugs; established Hemophilia Home of China – a patient group; adopted the Action Plan of China to Increase the Quality of Newborns and Reduce Birth Defects and Deformity; classified birth defects as non-genetic diseases and genetic diseases; established Rare Disease Relief Public Fund and Shandong Province Rare Disease Association. In addition, the National Natural Science Foundation of China has allocated additional resources in support of relevant research correspondingly every year. Regarding drug control, the foundation set up a negotiation mechanism on orphan drug prices. It also suggested using the entry system of orphan drug market to encourage its research and development to improve diagnosis, with reference to experiences from other countries and regions.

**D. Our Requests**

The International Covenant on Economic, Social and Cultural Rights (Article 12) of the International Bill of Human Rights, states the rights of “every individual to the enjoyment of the highest attainable standard of physical and mental health”, with timely medical treatment. Moreover, the United Nations Convention on the Rights of Persons with Disabilities (Article 25) also reinforces “the rights of disabled individuals to the enjoyment of the highest attainable standard of health” to ensure the

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\(^5\) Speech of Dr Gu Hyun-min, Head of information Management Division, Korea Orphan Drug Centre in the First Round Table on the Development of Rare Disease Policy in Hong Kong

\(^6\) Rare Disease website of China [http://www.hanjianbing.org/content/details_9_1505.html](http://www.hanjianbing.org/content/details_9_1505.html)

\(^7\) Speech of Professor Li Ding Guo, Chairman, Rare disease specialist division, Shanghai Medical Association in the First Round Table on the Development of Rare Disease Policy in Hong Kong
provision of required healthcare services including rehabilitation, early diagnosis and inception for the prevention of recurring disability. The above two Bills are both applicable to Hong Kong Special Administrative Region.

We urge the Government to establish patient-centered healthcare policies, which honour and are driven by the principles of human right and equality, regardless of the rarity of the disease types. Early engagement of patients and their families at the policy-formulation and implementation processes is vital. Proper policy consultation, planning and review meetings should be well participated by affected patients and their families, for effective service planning and delivery in the long run.

Our recommendations are as follows:

1. **Call for a Clear Definition of Rare Disease**

Hong Kong Government/Hospital Authority should clearly define “rare disease” making reference to WHO standard, and encompass rare diseases in the local healthcare policy and system.

While the US, EU, Singapore, Japan, Australia, Taiwan and Korea all have a clear definition of rare disease and put in place related healthcare policies in support of rare disease patients, Hong Kong still lacks far behind. With better knowledge and increasing number of cases identified annually, it is opportune that the Government take action and put forward a much-needed definition and long awaited policies to support rare diseases patients and their families.

2. **Establish a rare diseases registry**

An integrated and comprehensive rare diseases registry should be established in Hong Kong. This meets the criteria for disease registry from a public health point of view. Rare diseases have definite diagnostic criteria, and collectively they affect a significant proportion of the population. Patients with rare diseases will be significantly affected by their diseases lifelong. The registry will serve essentially as a repository of past rare disease cases, with regular updates, in order to inform and
facilitate proper diagnosis and treatment. The registry will also facilitate clinical trials and other research activities involving patients with rare diseases.

3. **Set up “Rare Disease Centre”**

In light of the genetic inheritance of rare diseases, genetic counselling, neonatal screening and new-born screening are important for early detection of rare diseases. We urge the Government to establish a “Rare Disease Centre” at the new Children’s Hospital, which will be put in service in 2018. The Government should also strengthen medical training for rare disease diagnosis and treatment, adopting a multi-specialist treatment mode if needed, and consider special approval procedure for orphan drugs. As rare diseases are serious and often life-threatening to sufferers, the Children’s Hospital should extend its service to adult rare diseases patients to ensure continuous care.

4. **Strengthen rare diseases genetic speciality service**

The Royal College of Physicians suggested a ratio of two genetic specialists and four genetic counsellors to every one million of population. Hong Kong has a severe deficiency of speciality service. Specialist doctors with training in clinical genetics are lacking. Resources allocation in professional training and manpower establishment is crucial to meet pressing demand for clinical genetic services among rare disease patients.

5. **Launch related support and social security policy**

Government should put in place corresponding support policies and measures, such as information centre, awareness building in the community, social support in areas of medical, rehabilitation, education, employment and social participation. For severe rare disease cases, the need of a case manager should be considered.

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E. Future Prospect

We will collaborate with various stakeholders, including patients and their families, alliances, medical practitioners and overseas stakeholders for policy advocacy. The Alliance will soon initiate meetings with various Government departments such as Food and Health Bureau, Labour and Welfare Bureau, Hospital Authority, Department of Health, Equal Opportunities Commission, to reflect patients’ needs and urge for better support policies in the aspects of medical, rehabilitation, education, social welfare, employment and social participation.

Last but not the least, we are launching public education activities in the community to promote the understanding and assistance towards patients and carers, in hope for a fulfilling life for the patients with the help from the government, related organisations and the general public.

Signatories:

| Hong Kong Alliance for Rare Diseases          | 香港罕見疾病聯盟               |
|                                           | Hong Kong Paediatric Rheumatism Association       |
| Little People of Hong Kong                   | 香港小腦萎縮症協會               |
| Hong Kong Spinocerebellar Ataxia Association | 香港肌健協會                 |
| Hong Kong Neuro-muscular Disease Association | 香港罕見疾病關注組             |
| Hong Kong Rare Disease Concern Group         | 香港紅十字會甘迺迪中心校友會 |
| Hong Kong Alliance of Patients’ Organization | 香港病人組織聯盟              |
| Hong Kong Marfan Syndrome Association        | 香港香港馬凡氏綜合症協會       |
| Hong Kong Association of Squint and Double Vision Sufferers | 香港斜視重影病患者協會 |
| Hong Kong Mucopolysaccharidoses & Rare Genetic Diseases Mutual Aid Group | 香港結多糖暨罕有遺傳病互助小組 |
| The Hong Kong Society for Rehabilitation     | 香港視網膜病變協會             |
| Retina Hong Kong                             | 夏約書孤兒症基金會             |
| Joshua Hellmann Foundation for Orphan Disease | 遺傳性心律基金會               |
| Sudden Arrhythmia Death Syndromes Foundation | 香港卓護義工協會             |
| Hong Kong Credible Care Volunteers Association Limited | 周權棣 – 結節性硬皮症病人的照顧者及家長 |
| Mr. Ronald Chow – parent of a patient diagnosed of Tuberous Sclerosus Complex | Mr. Patrick Fuk Wu Chan – a diagnosed Myelofibrosis patient |
| 陳福和 - 香港骨髓纖維化病人               | 香港紅十字會甘迺迪中心校友會 |
| 香港病人組織聯盟 |
Appendix 1: Definitions of Rare Disease in Major Countries / Regions

WHO\(^9\) Rare, debilitating or even fatal diseases with 6.5 to 10 cases in every 10,000

US\(^10\) Diseases threatening less than 200,000 patients in the country (less than 7 cases in every 10,000)

EU\(^11\) Diseases threatening less than 5 cases in every 10,000

Taiwan\(^12\) Genetic diseases with diagnostic difficulty threatening less than 1 case in every 10,000

Japan\(^13\) Diseases threatening less than 50,000 patients in the country (less than 4 cases in every 10,000)

Australia\(^14\) Diseases threatening less than 2,000 patients in the country (less than 1 case in every 10,000)

South Korea\(^15\) Diseases threatening less than 20,000 patients in the country (less than 4 cases in every 10,000)

Mainland China Under discussion. Medical sector suggests the definition of rare disease which threatens less than 0.02 case in every 10,000, or less than 1 case in every 10,000 new-born babies

\(^9\)WHO Standard
\(^11\)Introduction of Approval of Drugs for Rare Diseases in European Medicines Agency http://www.ema.europa.eu/ema/index.jsp?curl=pages/special_topics/general/general_content_000034.jsp&mid=WCOb01ac058002d4eb
\(^12\)Definition by the Health Promotion Administration, Ministry of Health and Welfare, Taiwan http://www.hpa.gov.tw/BHPNet/Web/HealthTopic/TopicArticle.aspx?id=201109150001&Class=2&parentid=200712250007
Appendix 2: Problems Faced by Hong Kong Rare Disease Patients (views expressed during the Roundtable Conference on 27 February 2014)

1. Long diagnosis period

Patient with Myelofibrosis

- Doctors were not familiar with Myelofibrosis. The patient was diagnosed with the disease after endless check-ups on stomach, liver, duodenum, bone marrows in a period of two years.

Patient with Marfan Syndrome

- The patient could only be diagnosed with the disease until a cardiac surgery when he was 30 years old. He also experienced delayed and different diagnosis.

Caretaker of Squint and Double Vision Patient

- Lack of guidelines, and it took 5 years for the diagnosis.

Patient with Pompe Disease

- A prolonged diagnosis after numerous checkups. The condition was only confirmed after a few years when he started to walk unsteadily, like his brother.

2. Lack of clinical experience

Patient with Marfan Syndrome

- There were only a few lines or a PPT slide about this syndrome in medical literature.
- It is hard to have patients sharing. It is suggested that the government could ride on the opening of the Children’s Hospital and organise patients sharing.

Patient with Myelofibrosis

- The symptoms are common and patients are a few. There are misunderstandings towards this disease.

Professor Chan from the Nethersole School of Nursing at the Chinese University of Hong Kong
Nursing staff are stressed due to shortage of resources, manpower and space.
Nursing staff has very little knowledge about rare diseases. Sometimes, it is embarrassing that patients know more than they do.
Nursing staff needs training in three areas: education, placement and scientific research.
The government should fund scientific research and support in caring of rare diseases patients. It is inappropriate not to treat due to few cases found.

Caretaker of Rett Syndrome Patient
- Gene mutation causes continuous body degradation and triggered several symptoms.
- Mostly doctors only know the existence of this rare disease but have no relevant clinical experience. Caretakers sometimes know more than doctors do.
- Was diagnosed as Autism in the early stage.
- Doctor provided antispasmodics to alleviate seizure but did not work.

Caretaker of 史立體病患
- Diagnosis is very difficult since it is an inborn metabolic disorder.
- It was hard to find suitable doctors. Patient samples were sent overseas for diagnosis.
- As the symptoms are common such as vomiting and doctors use normal treatment only.
- Caretakers needed to find doctors for diagnosis.

Patient with Muscular Atrophy and His Caretaker
- Continuous body degradation and also triggers off several symptoms.
- It brings a heavy economic and mental burden to the family as he needs different medical auxiliary equipment at different degradation stages.
- He was diagnosed with the disease when he was very young. He was acknowledged of his possible lifespan and was threatened.
- According to some foreign patients groups, they reveal that the living quality has positive impact toward lifespan. It is inappropriate that doctors adopt solely the medical approach towards patients in Hong Kong.
3. **Lack of medical support**

Patient with Marfan Syndrome

- Under Hong Kong hospital clusters system, guidelines to various diseases are either inconsistent or outdated.
- The latest guideline on Marfan syndrome should be dated in 2010, summarising its diagnosis, management and more updates. But the information in the public hospitals is not consistent and outdated.
- There is no comprehensive mechanism to review the current clinical guidelines in Hong Kong.
- Information is not shared among clusters. Inconsistent records are found in different hospitals and the diagnosis is inappropriate. Patients need to wait for at least 3 to 4 months for another treatment, which is resources and time consuming.
- No central registers or database for gauging patient records which leads to delayed diagnosis and treatment.
- In England, medical record is systematically recorded which also serves as clinical experiences sharing.
- There are genetic counsellors and nurses in England. They provide a great support to patients and their families.
- In US, some articles share how to deal with the syndrome from children’s perspective which is useful to the healthcare sectors and patients. It is suggested that patients groups or government professionals should work as the contact point to receive the information from these international organisations.

4. **High cost of the medication**

Caretaker of Tuberous Sclerosis Complex Sufferers

- Medication is highly costly at HK$30,000 to HK$40,000 per monthly, adding on family economic burden which many families cannot afford.

Caretaker of Children with Rheumatism

- Some treatments for Chronic Infantile Cutaneous, Articular Syndrome and Systemic Arthritis, are self-financed drugs rather than general drugs and cost over a million.
Patient with Pompe Disease

- The patient can only continue with the treatment if they pass the HA medical review every year which cause huge stress to the patients and their families. Therefore, they hope HA to extend the review to once every two years.
- The drug review process lacks transparency. Patients only receive the results without full explanation. It is suggested that HA should upload the results like some foreign countries.

5. Economic burden and stress

Caretaker of Squint and Double Vision Patient

- The disease causes psychological problems to the victim’s family, such as loneliness, hopelessness and frustration.
- It leads to academic difficulties and social issues if no any clinical psychologist is referred.

Caretaker of Tuberous Sclerosis Complex Sufferer

- There is no official definition on “rare disease”. This makes social resources provided to the patients (and caretakers) scarce.
- During the period of summer vacations and school holidays, it is challenging to choose suitable extra-curriculum activities for the patient. The parents hope to have summer courses which can provide special care services.
- Caretakers need to take care of daily lives and homework of the patient, in addition to the heavy job duty and economic burden. They are psychologically and mentally stressed and would like to receive social support in various aspects.
Appendix 3: Legislative Status on Rare Diseases in Foreign Countries

<table>
<thead>
<tr>
<th>Country</th>
<th>Legislative Action</th>
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<tbody>
<tr>
<td>US</td>
<td>Orphan Drug Act was signed in 1983</td>
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<tr>
<td>Singapore</td>
<td>Medicines (Orphan Drugs) (Exemption) Order was established in 1991</td>
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<tr>
<td>Japan</td>
<td>Revised orphan drug regulations in 1993 on promoting research and development of drugs for rare diseases</td>
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<tr>
<td>Australia</td>
<td>The Australian orphan drugs policy was reserved in 1997. In order to speed up the approval and introduction of orphan drugs</td>
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<tr>
<td>European Union (EU)</td>
<td>The European Union Regulation on Orphan Medicinal Products passed in 1999 with effect from 2000</td>
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<tr>
<td>Taiwan</td>
<td>The Rare Disease Control and Orphan Drug Act was passed in 2000</td>
</tr>
<tr>
<td>Korea</td>
<td>The Orphan Drugs Guideline was introduced in 2003</td>
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Appendix 4: Legislative Details on Rare Diseases in Foreign Countries

Korea 2003
Launched the Orphan Drugs Guideline.
1. Exclusive marketing rights for 6 years
2. Medical expense reimburse by the government
3. Nationally funded research programs support from the government
4. There is no fast track on drug approval process

Taiwan 2000
Established “The Rare Disease Control and Orphan Drug Act” and “Enforcement Rules of the Rare Diseases and Orphan Drugs Act”. The government subsequently included rare diseases patients into the “Physical and Mental Disable Protection Act”. Rare disease patients can all receive the same protection under this act, including financial assistance, social welfare services, medical care, rehabilitation, special education subsidies.

Support system
1. A 100% reimbursement for low-income families and 80% for others on medical, drugs and special nutrition through health insurance plan
2. Providing subsidies on home medical care equipment rental, domestic diagnostic testing, nutritional counselling, emergency medical expenses which are essential for the maintenance of patients’ life
3. 10 years exclusive marketing rights for Orphan drug
4. A separate programme on subsidising for rare diseases patient expenses to avoid being marginalised from other medical expenses
5. If a medicine not yet registered but qualified as an orphan medicine for the treatment of rare disease, a 30-day special fast-track on drug approval process applied

Strengthen prevention and early stage detected, included:
1. New-born screening for 11 rare diseases
2. Early health assessments is recommend for high risk groups included family member have suffered from rare disease, have an infant with health problems. They all receive a subsidy amount to NT $1,500 to NT $2,000
3. Suspected cases would transfer to central authorities for diagnose then refer to Genetic Counselling Centres for patients counselling
Passed The European Union Regulation on Orphan Medicinal Products

The Committee for Orphan Medicinal Products (COMP) is the committee at the European Medicines Agency that is responsible for reviewing applications from people or companies seeking “orphan-medicinal-product” designation. Incentives for the designated product as below:

1. 10 years exclusive marketing rights for Orphan drug
2. Eligible for incentives made available by the European Community and the Member States to support research into orphan medicinal products
3. The Committee for Medicinal Products for Human Use (CHMP) is the committee at the European Medicines Agency that is responsible for preparing opinions on questions concerning medicines for human use. It is not necessary to go through all EU members.

Support system
1. Improving recognition and visibility of rare diseases
2. Ensuring that rare diseases are adequately coded and traceable in all health information systems
3. Supporting national plans for rare diseases in EU member countries
4. Strengthening European-level cooperation and coordination
5. Creating European reference networks linking centres of expertise and professionals in different countries to share knowledge and identify where patients should go when expertise is unavailable in their home country
6. Encouraging more research into rare diseases
7. Evaluating current screening population practices

Revised the Australian orphan drugs policy, in order to speed up the approval procedure and introduction of orphan drugs.

The benefit of the orphan drug policy in Australia are:

1. This orphan drugs programme aims to ensure the availability of a greater range of treatments for rare diseases and allows the Australian Therapeutic Goods Administration (TGA) to use information from the US Food and Drug Administration (FDA) Orphan Drugs Program as part of the Australian evaluation process
2. Waiver of application and evaluation and no annual registration fees
3. A five-year exclusivity marketing rights for Orphan drug (similar to other drugs).
Revised orphan drug regulations on promoting research and development of drugs for rare diseases

Drug companies that are granted orphan drug designation are eligible to receive the following benefits:

1. The Ministry of Health, Labour and Welfare (MHLW) has a consultation service specifically for orphan drug designation applicants, which is free of charge.

2. The applicant may receive financial aid from the Japanese government for the collection of supporting data. The applicant may receive financial aid for as much as 50% of the cost of the clinical trials.

3. The application will be placed on a fast-track approval process, which takes 10 months while the approval for regular drugs takes at least 12 months.

4. Exclusive marketing rights for 10 years.

Support system:

1. Grants-in-aid for research programs.

2. Medical expense reimbursement for 56 diseases, with 30% of expenses paid by insurance companies and the rest paid by national and prefectures governments.

3. Price-control policies negotiated by Japanese National Health Insurance (NHI) and pharmaceutical companies.

Intended Medicines (Orphan Drugs) (Exemption) Order

According to the order, orphan drug means a medicinal product which has been identified by any doctor or dentist as an appropriate and essential remedy with no effective substitute available for the treatment of any rare disease, priority approval on applications for orphan drug.
United States President signed the Orphan Drug Act in January 1983, in order to encourage research and development of drugs for rare diseases.

In 2002, the Congress passed the Rare Diseases Act, establishing the National Institutes of Health established under the Office of Rare Diseases, now known as the Office of Rare Diseases Research.

According to Orphan Drug Act, the manufacturer or the sponsor of a drug may request the Secretary to designate the drug as a drug for a rare disease or condition. Incentives to orphan drugs providers in terms of R&D, intellectual property and marketing:

1. A seven year period of marketing exclusivity following the marketing approval. Other institutions may not be sold similar diseases, unless they prove that their drug is relatively more effective than in the exclusive market

2. A tax credit on the cost of clinical trials undertaken in the US

3. A fast-track procedure for the FDA to evaluate registration files (takes 6 months while the approval for regular drugs takes at least 10 months.

4. Launched funding scheme to subsidise clinical trials of drugs for rare diseases

The goals of the Office of Rare Diseases Research (ORDR) are to:

1. Supports with NIH Institutes and Centers (ICs) the Rare Diseases Clinical Research Network (RDCRN)

2. Supports the Global Rare Diseases Patient Registry and Data Repository (GRDR), a patient registry and data repository with fully developed Common Data Elements

3. Supports the development and implementation of a publicly accessible database of human biospecimen repositories, the Rare Diseases Human Biospecimens/Biorepositories (RD-HuB)
4. Identifies and responds to scientific opportunities and builds international research collaborations
5. Works with other organisations for more opportunities in rare diseases research
6. Provides opportunities for patient support groups to become partners with the NIH, to better understand NIH research programs, and to gain better access to NIH research opportunities

Orphan Drug Act is widely considered as effective. Between 1973 and 1983, there are less than 10 drugs for rare diseases in the market. Since the commencement of the legislation in 1983 till 2013, over 400 drugs for rare diseases introduce to the market.

For the financial subsidies on medicine, there is not much difference between rare disease and others. The health care system of US is primarily provided by insurance companies and supplement by public insurance programs by the government.

Drug expenses are covered by both insurance plans provide by private insurance companies and the government.