Rare Diseases Hong Kong
Submission on the Public Consultation for the 2023 Policy Address
(September 2023)

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

Over the past year or so since the current-term HKSAR Government took office, the Health Bureau (HHB) has made many impressive and commendable accomplishments, such as effective control of the COVID-19 epidemic and restoration of people's livelihoods, orderly implementation of the primary healthcare blueprint, promotion of the development of Chinese medicine and integration of Chinese and Western medicine, facilitating the first cross-border organ donation and transplantation between the Mainland and Hong Kong, and continuing to explore the establishment of a regular organ transplant mutual aid mechanism between the two regions, etc.

However, the work of the HHB in dealing with the challenges of rare diseases and responding to the demands of patients with rare diseases is somewhat inferior. Within the first year of the new-term Government, RDHK had written the Secretary for Health and Under Secretary for Health five letters requesting an information exchange meeting on the implementation of the former Government's rare disease measures and the current-term Government's approach to rare diseases, before eventually being granted a chance to meet the Under Secretary for Health and Hospital Authority (HA) representatives on 3 August this year. Please refer to the "*Meeting summary*" attached for the key points discussed in the meeting.

To sum up, in its first year in office, the current-term Government mainly focused on gradually implementing some of the rare disease measures proposed by the last term of Government, dealing with its unfinished business without putting forward any new ideas and strategies for tackling rare diseases. RDHK has always advocated tripartite coordination among the Government, the business sector and the community to address the challenges of rare diseases, and the Government should take the leading role. RDHK hopes that the current-term Government will strive to create a new situation in the fields of economy, housing, innovation and technology, etc. without forgetting to conceive innovative ideas in dealing with rare diseases, in order to better reflect the core values of an international metropolis like Hong Kong – keeping improving, being caring and civilised.

The measures for rare diseases proposed by the former Government have helped to address some specific problems of individual diseases. However, these measures were only reliant on clinical means, sporadic, fragmented, unsystematic and unplanned. RDHK hopes that the current-term Government will draw on the responsibilities and actions of the Central Government of the Mainland over the past decade in strengthening the diagnosis and treatment of rare diseases, and will take a macro perspective, proactive and foreword-thinking approach to policy development so that more systematic, comprehensive and integrated short- and long-term measures can be launched. It is hoped that the Government will consider and adopt the following six recommendations in the coming year for combating rare diseases in Hong Kong:

I. To form a government-led cross-sector coordination mechanism on strategy for rare diseases;
II. To commence a study on the local definition of rare diseases;
III. To provide rare disease patients with humanised healthcare services;
IV. To optimise and improve the access mechanism for orphan drugs by cutting the red tape;
V. To collaborate on rare disease diagnosis and treatment in the Greater Bay Area; and
VI. To include Herpes Zoster vaccination in the Vaccination Subsidy Scheme

The recommendations are elaborated as follows:

I. To form a government-led cross-sector coordination mechanism on strategy for rare diseases

One of the effective strategies of the Mainland in recent years to strengthen the diagnosis and treatment of rare diseases is the establishment of the "China Alliance for Rare Diseases", which comprises multiple stakeholders from government departments, experts and scholars, hospitals and universities, commercial enterprises and patient organisations, as a national platform for communication, coordination and implementation of government policies and measures.

In recent years, rare diseases have been receiving increasing public attention in Hong Kong. The introduction of new measures by the Government in consequent years has contributed to the boosted attention and commitment to rare diseases among various sectors such as universities, research institutions, innovation & technology enterprises, and healthcare & pharmaceutical industries; accumulation of experience in screening, diagnosis and treatment of rare diseases among the HA staff from management to frontline clinical specialists; and a more comprehensive and in-depth understanding of rare disease issues among the government officials. On the existing foundation, if the passion and strength of different parties are gathered, and professional knowledge and resources are coordinated effectively, the outcomes of tripartite collaboration among the Government, the business sector and the community can certainly be maximised, so that the efforts of the Government and stakeholders can be multiplied with half the efforts. RDHK expects the current-term Government will launch a cross-sector coordination mechanism on strategy for rare diseases.

It is suggested that the coordination mechanism be led by the Government and chaired by the Secretary for Health, with government officials and representatives from the HA, two medical faculties, rare diseases specialists, Members of the Legislative Council, patient groups, the business sector, etc. as members. The core responsibilities of the committee are:

1. To suggest the policy goals and medium to long term strategy for rare diseases in Hong Kong based on the existing practical experience as well as the experience of Europe, the US and the Mainland for consideration and reference by Government decision-makers;

2. To devise and implement in phases the "Hong Kong Rare Disease Action Plan" in accordance with the situation in Hong Kong, including issues of human resources, patient registries, centres of excellence, cross-border collaboration, rehabilitative support, etc.; and

3. To coordinate and implement various measures for rare diseases proposed by the Chief Executive, and communicate the implementation progress with government departments.

II. To commence a study on the local definition of rare diseases

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

In May 2018, five national bodies including the National Health Commission jointly issued the *First National List of Rare Diseases* including a total of 121 rare diseases. Meanwhile, both the public and private sectors have actively studied how to define rare diseases. Some experts have proposed that the definition of rare diseases is both a matter of science and values, reflecting the Government's attitudes to the vulnerable social groups and its civilization level. There are always more solutions than difficulties. There are different types of experts in Hong Kong, and the GDP per capita is much higher than that of other major cities such as Beijing, Shanghai and Shenzhen. As long as the Government takes action, it is completely possible to formulate a local definition of rare diseases.

It is understandable that defining rare diseases locally is a complex task that cannot be accomplished overnight. However, the current-term Government should no longer evade the issue. Instead, it should demonstrate an accountable and responsive governance style by bringing together the stakeholders to commence a study. The Government should draw on the experience of those countries and regions that have effectively dealt with rare diseases, especially the ideas and practices put forward by the Mainland Government and the fields related in recent years, and draw conclusions that are in line with the actual situation in Hong Kong.

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

III. To provide rare disease patients with humanised clinical services

Most of the rare disease cases are followed up by multiple specialties, so attending follow-up consultations is a big challenge to many rare disease patients. They need to travel to various specialties and undergo multiple tests at different times. Both patients and caregivers have to spend a lot of time and energy to cope with follow-up consultations, which can be exhausting.

According to the HA, Hong Kong Children's Hospital will become a treatment centre for all children with rare diseases in the future. When reaching adulthood, patients will be referred to other hospitals such as Margaret Hospital specialising in metabolic disorders, for follow-up according to their needs or personal wishes. This arrangement will undoubtedly help to improve the clinical management of rare disease cases, but RDHK expects to have improvement in clinical services for patients as well.

It is known that some specialties of Hong Kong Children's Hospital have tried to focus on a certain rare disease (such as Tuberous Sclerosis Complex) at a certain period of time. On this basis, hospitals that hope to provide follow-up consultations for specific rare disease patients will try to concentrate various specialties related to a rare disease at the same time period (such as half a day or one day) to follow up for patients. This arrangement not only helps to spare the patients from travelling to different hospitals on different days, but also helps the relevant specialties to fully integrate the latest clinical changes of patients' cases in a timely manner so as to provide more appropriate treatment and

care. Therefore, it is hoped that this practice will be extended to other types of rare diseases. We understand that it takes extra time and effort to gather different specialties for the same group of rare disease patients at a certain time, and certain administrative change may be required. However, this change is beneficial to both clinical services and clinical management, more humane to patients and caregivers, and the difficulties incurred are not insuperable.

In addition, patients are currently required to go to the designated hospital for some check-ups before follow-up consultations. RDHK suggests that patients be allowed to go to the nearby clinics for blood taking, submission of urine and stool samples, radiological examinations, etc. It is reported that New Territories East Cluster has already allowed the cancer patients to choose where to go for blood taking. So, we hope that this practice will be adopted by other clusters and apply to all rare disease patients, and more "patient-oriented" and humanised clinical services can be launched in the future.

IV. To optimise and improve the access mechanism for orphan drugs by cutting the red tape

For those rare disease patients with declining physical function, their greatest concern is whether they can receive timely treatment. According to the usual process of drug registration and inclusion, patients usually wait for years before having the opportunity to take new drugs that have already come onto the market. Fortunately, the Government has begun to recognise and propose new measures dealing with rare diseases in recent years. For instance, since August 2017, eligible patients have been provided with subsidy to purchase ultra-expensive drugs (including those for treating uncommon disorders). Two of the ultra-expensive drugs (Tisagenlecleucel and Tafamidis) currently funded by the Community Care Fund (CCF) were incorporated into the CCF assistance programme in 13 to 14 months on average after registration; whereas Nusinersen was included in the Safety Net in only 4 days after registration, so that the patients in need could receive treatment as soon as possible.

As a patient group, RDHK welcomes the flexible approach adopted by the HA. The precedent cases in recent years have shown that accelerating the inclusion of orphan drugs in the HA Drug Formulary and Safety Net is feasible. Therefore, RDHK handed in a submission on "Optimising and Improving Access Mechanism for Orphan Drugs by Cutting the Red Tape" to the Government and HA last year, recommending the seven measures below to optimise and regularise the access mechanism for orphan drugs:

1. Optimise the documentation requirements for registration and registration process of orphan drugs, such as granting conditional registration of orphan drugs with one Certificate of Medicine (CPP);
2. Strengthen collaboration between the Chief Pharmacist's Office (CPO) of the HA and the pharmaceutical industry;
3. Omit the non-essential work of the Drug and Therapeutics Committee (DTC) in the drug inclusion process;
4. Adjust the schedule and mode of the Drug Management Committee (DMC) meetings;
5. Solve the chicken-and-egg dilemma by coordinating the two mechanisms of medical testing and drug inclusion;
6. Adopt early awareness and alert systems (also known as horizon scanning) to set priorities for the allocation of resource in advance; and
7. Adopt "Orphan Drug Trial" policy and systematically collect and use real-world data to generate empirical evidence which helps to accelerate patient access to orphan drugs.

RDHK is aware that some of the above recommendations have been implemented and some will be implemented soon. It is hoped that the relevant departments will continue to take action to effectively improve and accelerate patient access to orphan drugs.

V. To collaborate on rare disease diagnosis and treatment in the Greater Bay Area

The Central Government and various provinces and municipalities in the Mainland have launched many policy measures to improve the diagnosis and treatment of rare diseases in recent years. For instance, in 2015, the National Health and Family Planning Commission established the "Expert Committee on Rare Diseases Diagnosis, Treatment and Medical Security", reflecting that rare diseases have entered the field of national policy; in 2016, the National Rare Diseases Registry System was established to consolidate national rare disease data; in 2018, the National Health Commission (NHC) released the first list of rare diseases, including 121 rare diseases; and in 2019, the NHC established a national collaborative network for rare disease diagnosis and treatment which consists of 324 hospital nationwide (13 of which are located in the Greater Bay Area), and published the first "*Guidelines for Diagnosis and Treatment of Rare Diseases*".

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

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

1. Designate some hospitals in Hong Kong (Hong Kong Children's Hospital, Queen Mary Hospital or Princess Margaret Hospital may be considered) to participate in the national collaborative network for rare disease diagnosis and treatment, and work closely with the 13 designated rare disease hospitals in the Greater Bay Area in clinical diagnosis and treatment. There can be cross-border referrals as necessary and when conditions permit. (Action to be taken by: HHB, HA)

2. Include HA's rare disease cases in the National Rare Diseases Registry System, and encourage intensive cooperation and exchange of data and scientific research with the 13 designated rare disease hospitals in the Greater Bay Area. (Action to be taken by: HHB, HA, Faculties of Medicine of HKU and CUHK)

3. The internationally recognised Clinical Trial Centres of the two faculties of medicine in Hong Kong should collaborate with the 13 designated rare disease hospitals in the Greater Bay Area to attract multinational and Mainland pharmaceutical companies to conduct clinical trials for innovative drugs in Hong Kong. (Action to be taken by: Faculties of Medicine of HKU and CUHK)

4. Review the differences between the drug regulatory systems in Hong Kong and the Mainland to facilitate effective coordination and promote integration. (Action to be taken by: HHB, Department of Health)

5. Adopt strategic procurement and initiate cross-border collaboration on medical insurance system in the Greater Bay Area so that more reasonable orphan drug prices can be achieved. (Action to be taken by: HA)

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

If the diagnosis and treatment of rare diseases in Hong Kong and the Greater Bay Area are integrated, it is expected that local industry and patients will benefit in the following ways:

1. The number of clinical cases will be largely increased by real-time clinical exchange through the collaborative network for rare disease diagnosis and treatment. Consequently, the experts can gather more data and clinical experience which helps to accelerate diagnosis and treatment planning process.
2. If more and more clinical trials for innovative orphan drugs are conducted in Hong Kong, the local patients will gain early access to new drugs, whereas local cumulative data on drug use can be collected.
3. Under the national medical insurance system, the Mainland has strong bargaining power. Therefore, the adoption of strategic procurement in the Greater Bay Area will help the patients in Hong Kong gain access to high-quality and more affordable drugs.

VI. To include Herpes Zoster vaccination in the Vaccination Subsidy Scheme

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

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

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

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

— End —

ATTACHMENT

Rare Disease Hong Kong
Follow-up meeting on the implementation of the measures on rare diseases proposed in the Policy Addresses
Meeting Summary

Date : 3 August 2023 (Thu)
Time : 10:05 – 11:00 am
Venue : Room 1801, Central Government Offices
Attendees:

1. Dr Libby LEE, Under Secretary for Health, Health Bureau
2. Ms Ellen CHAN, Principal Assistant Secretary for Health, Health Bureau
3. Ms Suki WONG, Assistant Secretary for Health, Health Bureau
4. Dr Simon TANG, Director of Cluster Services, Hospital Authority
5. Dr Larry LEE, Chief Manager (Cluster Performance) , Hospital Authority
6. Dr Venus SIU, Chief Manager (Quality and Standards), Hospital Authority
7. Dr Benjamin LEE. Chief Pharmacist, Hospital Authority
8. Mr Clement CHAN, Senior Manager (Allied Health), Hospital Authority
9. Dr Oliver CHAN, Senior Manager (Operation and Cluster Service), Hospital Authority
10. Mr KP TSANG, Chairman, Rare Disease Hong Kong
11. Ms Rebecca YUEN, Rare Disease Hong Kon
12. Ms Iris CHAN, Rare Disease Hong Kon
13. Mr Henry TAM, Rare Disease Hong Kon
14. Ms May HO, Rare Disease Hong Kon
15. Ms Amy CHAN, Rare Disease Hong Kon

Summary of the meeting:

Rare Disease Hong Kong (RDHK) requested a meeting with the Health Bureau (HHB) and Hospital Authority (HA) representatives for the main purpose of ascertaining the progress of the implementation of the rare disease measures in the Policy Addresses and to convey views to the authorities.

1. HA's responses

1.1. Genetic counselling service

1.1.1. Since 1 July 2023, the HA has taken up the Clinical Genetic Service which was under the Department of Health, including the Genetic Counselling Clinic, the Genetic Screening Clinic, Neonatal Screening and Genetic Laboratory. The services provided by the Genetic Laboratory will also be handed over by the end of this year. The aim is to strengthen the provision of one-stop multi-disciplinary support for patients with genetic diseases and their families. Services available include genetic diagnostic service, treatment, subsequent management, counselling and prevention, etc.

1.1.2. The team providing services includes specialists in genetics and genomics, paediatricians, allied nurses, genetic counsellors and scientists. It is expected that 6,000 cases can be handled per year.

1.1.3. To meet the development needs of genetics and genomics, the Hong Kong Genome Institute (established and wholly owned by the Government) has added the post of "Genetic Counsellor" and will provide training. The HA will also gradually create more positions.

1.2. Information platform

1.2.1. Smart Patient Website – There are links to different rare diseases for the reference of the public. In the future, more information about rare diseases, such as short videos, will be added, and a link to the RDHK's website may also be added.

1.2.2. The website of Hong Kong Children's Hospital – Introduction to genetic testing, counselling services, and information on individual rare diseases.

1.3. Patient registries

1.3.1. Over the past few years, the HA has been focusing on the Patient Registry Scheme, and the Hong Kong Children's Hospital (HKCH) has gradually established a database for individual rare diseases to assist in clinical diagnosis and treatment since 2021/22. At present, HKCH has established a database of 30 rare diseases, including 25 congenital metabolic diseases, 4 brain diseases and 1 endocrine disease, the detailed list of which is shown in the appendix.

1.3.2. In the long run, HKCH will become a treatment centre for all children with rare diseases. When reaching adulthood, patients will be referred to other hospitals such as Margaret Hospital specialising in metabolic disorders, for follow-up according to their needs or personal wishes.

1.4. Responses to RDHK's requests and suggestions

1.4.1. Cancer patients in the New Territories East Cluster have been allowed to choose where to have blood drawn. This practice will be extended to the Department of Medicine later. Patients are no longer required to have their blood test in designated hospitals. Instead, they can choose to go to the nearby clinic for blood taking, which is time-saving and convenient. But so far, such practice will be applied within the Cluster only. The ultimate goal is to apply it to all Clusters.

1.4.2. In order to facilitate the seamless handover of pediatric patients to adult hospitals when they grow up, HKCH currently has two case managers, each responsible for about 50 cases, and will increase the number of staff in the future. At the same time, Princess Margaret Hospital has set up a "transition liaison clinic" to follow up 30 metabolic disease cases and conduct video consultations. However, the HA has reservations about the suggestion to concentrate all adult patient cases in one hospital, as different diseases involve different specialties and different hospitals have different specialties, so it would be more practical to refer individual diseases to hospitals that specialize in handling them.

1.4.3. As at March 2023, the Community Care Fund covers 7 ultra-expensive drugs for the treatment of different diseases, including rare diseases such as spinal muscular dystrophy and neuroblastoma, with a total of 219 applications approved in the past. In addition, 64 applications for congenital metabolic diseases were approved.

1.4.4. RDHK reported that after the Community Care Fund optimized the financial means test mechanism in 2019, most patients were satisfied. However, the practice of paying the drug fee in a lump sum had caused financial pressure on the patient's family. For example, some drugs can cost a few hundred thousand to 1 million dollars per year, and most families have cash-flow problem if they have to pay in a lump sum. HA expressed its understanding and promised to review the issue.

2. HHB's responses

2.1. The Health Bureau has not changed its policy on rare diseases and will continue to support and care for rare diseases.

2.2. At present, the focus is on how to implement and accelerate the implementation of the policies and measures that have been put in place, such as the above-mentioned HKCH measures to concentrate on the treatment of all children with rare diseases and adult rare diseases in designated hospitals.

2.3. Apart from rare diseases, the HHB has also maintained communication with the Mainland on

various aspects, such as the drug utilisation mechanism. During the current Government term, Hong Kong's drug inclusion mechanism will be adjusted from the current practice of requiring two Certificates of Pharmaceutical Product (CPP) to only one CPP, with sufficient clinical evidence, so that drugs can be introduced into Hong Kong as long as Hong Kong specialists believe that they are needed. If there are more available drug options, the price of drugs may also be reduced, so that more people can benefit.

2.4. In the long run, it is hoped that Hong Kong will be able to review drugs independently. But the first step to achieving this vision is to train or recruit enough people who know how to review the effectiveness of drugs and understand scientific data. Therefore, within one to two years, it is planned to implement a drug inclusion mechanism that requires only one CPP plus local clinical research data.

2.5. Responses to RDHK's requests and suggestions

2.5.1. RDHK provides reference materials on "Policies Related to the R&D and Marketing of Rare Disease Drugs in China" (《中國罕見病藥物研發上市相關的政策》), showing that the policies and measures to deal with rare diseases in the Mainland start from the top-level design, involving different government departments and non-governmental collaboration. In addition, there are more than 300 hospitals forming a rare-disease network in the Mainland, and it is suggested that Hong Kong may also consider adding HKCH or other designated hospitals to the network.

2.5.2. According to the HHB, HKCH has already had a lot of exchanges and collaboration with the Mainland at the specialist level, such as exchanging with Peking Union Medical College Hospital in September this year, and exchanging with HKU-Shenzhen Hospital on a regular basis. In the future, it also plans to form sister hospitals with some hospitals in the Mainland.

2.5.3. On the whole, Hong Kong doctors are very willing and often go abroad to communicate with the outside world. However, the situation in the Mainland and Hong Kong is very different. In the first place, the Mainland has far more rare diseases than Hong Kong, so the policy will be easier to implement. Secondly, the Mainland's healthcare system is mainly supported by medical insurance, which subsidises patients in the form of tax reductions. However, Hong Kong relies mainly on the HA, which means that the Government bears the main responsibility to help patients by increasing drug options and reducing the price of drugs.

2.5.4. The Health Bureau agrees that there are many opportunities for co-operation between Hong Kong and the Mainland, and one of the major directions is to make use of the advantages of Hong Kong's scientific research to help the Mainland go global.

2.5.5. At present, the list of rare diseases in the mainland includes 121 rare diseases, but it is also being adjusted, and the 1:10,000 criterion may be adopted. Hong Kong will use the list of rare diseases in the Mainland as a reference standard, to identify rare diseases such as spinal muscular dystrophy and tuberous sclerosis complex.

2.5.6. RDHK reported that patients with rare diseases would like to go to HKCH for treatment, but their main hospital would not release them. The HHB responded that the future policy is still to gather all pediatric patients in HKCH, but in some individual cases, the actual needs of patients have to be considered. For instance, those with stable condition, only need to obtain prescribed drugs or read reports, do not necessarily go to HKCH. The arrangement of video consultation will not be excluded as long as the situation is appropriate.

3. RDHK thanked the HHB for arranging the meeting and promised to maintain regular communication with RDHK via video or physical meetings in the future.

Appendix

Hong Kong Children's Hospital has established a database for 30 rare diseases which are shown as follows:

1. 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
2. 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency
3. Argininaemia
4. Argininosuccinic Acidaemia
5. Beta-Ketothiolase Deficiency
6. Biotinidase Deficiency
7. Carnitine Palmitoyltransferase II Deficiency
8. Carnitine Uptake Deficiency
9. Carnitine Acylcarnitine Translocase Deficiency
10. Citrullinaemia Type I
11. Citrullinaemia Type II
12. Classic Galactosaemia
13. Classic Phenylketonuria
14. Congenital Adrenal Hyperplasia
15. Dravet Syndrome
16. Duchenne Muscular Dystrophy
17. Glutaric Acidaemia Type I
18. Glutaric Acidaemia Type II
19. Homocystinuria
20. Isovaleric Acidaemia
21. Maple Syrup Urine Disease
22. Medium chain Acyl CoA Dehydrogenase Deficiency
23. Methylmalonic Acidaemia and Homocystinaemia (Cobalamin C Deficiency)
24. Methylmalonic Acidaemia
25. Multiple Carboxylase Deficiency
26. Propionic Acidaemia
27. Spinal Muscular Atrophy
28. Tuberous Sclerosis Complex
29. Tyrosinemia Type I
30. Very Long chain Acyl CoA Dehydrogenase Deficiency