Journal of
Gynecology and Women's Health
ISSN 2474-7602

Juniper Publishers
key to the Researchers

Mini Review
Volume 6 Issue 4 - August 2017
DOI: 10.19080/JGWH.2017.06.555691

J Gynecol Women's Health

To Bridge the Gap: Implementation of Rare Disease Screening Program in Hong Kong

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Submission: February 21, 2017; **Published:** August 24, 2017

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Introduction

The main objective of this article is to raise awareness amongst the public and decision-makers about rare diseases and their impact on patients' lives in Hong Kong. Rare diseases affect many people , however only few of these diseases have an effective drug treatment available. Hong Kong is relatively late to implement inborn errors of metabolism (IEM) screening program. This is a world recognized cost effective public health program aiming at reducing the morbidity and mortality associated with rare diseases. Therefore, universal IEM screening program should be implemented in Hong Kong as soon as possible. There is an urgent need to bridge the knowledge gap by research, educating and connecting patients, families, physicians and specialists in rare disease in Hong Kong. So as to advance the awareness, diagnosis, treatment and research of IEM and other rare diseases. As result to improve the welfare of patients with rare diseases in Hong Kong. Moreover, it is necessary to launch a comprehensive rare disease centre in Hong Kong which aims at early identification and provision of timely treatment, quality health care and support to patients and families with special needs.

Rare Diseases is not Rare

Henrard & Arickx [1] reviewed that each disease is rare but collectively, rare diseases affect many people, however only few of these diseases have an effective drug treatment available. According to Rare Disease Day 2017 [2], about 30 million people in the European Union may be affected by one of over 6000 existing rare diseases. 80% of rare diseases have identified as genetic origins, however, 50% affected children.

Beginning in 2008, International Rare Disease Day [2] hold events every year have raised awareness worldwide. At the beginning only 18 countries has taken place, however, in 2016 there were 84 countries has held events about rare disease (including Hong Kong). It states that 'Research is key. It brings hope to the millions of people living with a rare disease across the world and their families'.

Reduce Costs for Healthcare Systems

Rare diseases are a large group of rare, genetic diseases involving disorders of metabolism. The lack of scientific knowledge and quality information on the disease often results in a delay in diagnosis. According to Direct Relief [3], for patients with rare disease, the mean length of time from symptom onset to accurate diagnosis was 4.8 years, however, treatment exists for these diseases is less than 5%.

But & Hui [4] concluded that the ongoing implementation IEM screening program may led to the development of clinical and scientific research about rare diseases. Moreover, which may led to the development of new diagnostic and therapeutic procedures for rare diseases. IEM screening reduces the time for accurate diagnosis so that a rare disease may be diagnosed earlier and properly treated. Therefore, these patients no longer need irrelevant tests, ineffective treatment or hospital visits. As a result to reduce costs for healthcare systems. These may result in the development of appropriate public health policies. In addition, research on specific rare diseases often gives a brighter and more promising future for patients with rare diseases.

Hong Kong's Situation

According to Policy Address 2017 [5], starting from 2017, HA will progressively expanded newborn screening of IEM (inborn errors of metabolism) as complementary screening for all eight public hospitals with maternity wards.

Inborn errors of metabolism (IEM) are a large group of rare, genetic diseases involving disorders of metabolism. According to Poon [6] there were an estimated 2,814 to 9,170 patients and 14 new cases with IEM in Hong Kong in 2013. According to the Chinese University of Hong Kong (CUHK) 2016 [7], about 1 in

4,700 Hong Kong babies is affected by inborn errors of metabolism (IEM), which are caused by genetic defects leading to failure to produce certain essential enzymes for normal metabolism. The affected babies will suffer from chronic illnesses including mental retardation and even fatal. Poon [6] explained that these diseases with broad diversity of disorders and symptoms that vary from disease to disease. The relatively rare diseases without obvious symptoms are likely to be overlooked by clinicians and governments. Yau [8] supported that early recognized and diagnose for IEM patients with treatable neurological disorders may result in better neurological outcome. Therefore, irreversible damages to patients' body may arise from delayed diagnosis and treatment. IEM Screening Program is a newborn screening program targeting at a panel of IEM of amino acids, organic acids and fatty acids. Infants affected by IEM can appear normal at birth. If delayed to diagnose and treat, may result in permanent neurological damages and even fatal. The goal of the screening program is to identify the affected neonates at the earliest moment, before they develop any signs or symptoms of the diseases and treat them early so as to maximize the efficacy of treatment. However, Chong [9] comment that although most practicing clinicians may not be actively involved in the care of rare disease patients, however, they play an important role in early recognition and referral of patients for further diagnostic workup and treatments.

Chong commented that compared to other countries of Asia Pacific region and Hong Kong is relatively late to implement inborn errors of metabolism screening. She concluded that wide universal rare diseases screening program should be implemented in Hong Kong as soon as possible.

Since 1984 newborn screening program was initiated by using cord blood to screen for Glucose-6-phophate dehydrogenase deficiency (G6PD) and congenital hypothyroidism by the Clinical Genetics Service of the Department of Health. Hearing screening was added since 2007. In 2013, the Chinese University of Hong Kong (CUHK) [9] launched the first screening program for inborn errors of metabolism (IEM) in Hong Kong for 30 IEM conditions. It included departments of Chemical Pathology, Obstetrics and Gynecology, and Pediatrics jointly developed this screening program. Then in October 2015 the Pilot Study on Newborn Screening for Inborn Errors of Metabolism implemented in two public hospitals for 24 IEM conditions. According to Policy Address 2017 [5], the Department of Health (DH) and the Hospital Authority plan to extend and expanded the screening service to all eight public hospitals with maternity wards in phases from the second half of 2017-18. It has provided hope and support to the patients with rare diseases and their families. Furthermore, this create further public awareness about rare diseases.

Poon [5] summarized that since 1997 the Department of Pediatrics at the Prince of Wales Hospital of The Chinese University of Hong Kong was the first to establish a Joint Metabolic Clinic, similar joint metabolic clinics have been established in other regional hospitals in Hong Kong namely the Princess Margaret Hospital since 2002, the Queen Mary Hospital since 2003 and the Queen Elizabeth Hospital since 2009. These four hospitals established as specialist centers, run by pediatricians, chemical pathologists and dietitians, providing a comprehensive and multidisciplinary care to patients with IEM for treating lysosomal storage disorders with enzyme replacement therapy.

Poon [6] comments that although recent enzyme replacement therapy (ERT) have been shown to be effective in dealing with the symptoms of some Mucopolysaccharidoses (MPS) and other types of lysosomal storage disorders including Gaucher disease, Fabry disease and Pompe disease. However, ERT is very expensive and involves lifelong treatment. This often results in heavy social and financial burdens on patients and their families.

Poon [5] and Yau [8] expressed that early recognize the signs and symptoms and a timely referral to a metabolic specialist and early diagnosis and treatment are crucial in the management for rare diseases patients. Although But and Hui [4] stated that there are more and more treatment available for previously incurable conditions. However, Mucopolysaccharidoses and rare genetic diseases mutual aid group (HKMPS) was disappointed that the current screening program does not include MPS ,Pompe Disease, Gaucher's Disease and Fabry Disease.

In a Press Releases [10] Kwok question that the Hospital Authority (HA) not only has been slow in investigating and approving for the applications for subsidies but also subsidies insufficiently to cover the expenses on treatment and medications required by patients with rare diseases to slow down their diseases or even to sustain their lives . The Secretary for Food and Health, Dr Ko replied that since 2008-09, HA has provided ERT for 20 patients suffering from lysosomal storage diseases (LSD) and there are 16 patients receiving ERT in 2013. The Hong Kong Government expanded $45 million each year to provide enzyme replacement therapy (ERT) for patients suffering from only six types of LSD including Pompe disease, Fabry disease, Gaucher disease, and Mucopolysaccharidosis Type 1, Type 2 and Type 4. However, ERT only given to those early in the stage of illness with no irreversible harm to the body which may ensure the best possible treatment outcome.

Kwok [10] argued that these given rise to inequalities and difficulties in access to treatment and appropriate quality health care for patients who are late in the course of illness or with some degree of irreversible harm to the body. HKMPS [11] and National organization for rare disorders (NORD) [12] advocated that the government should subsidies the expensive bill for effective drugs for patients with rare diseases.

However, in Hong Kong some physicians may not know where to refer patients with rare disease. There is lack of a comprehensive rare disease centre among different providers

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of metabolic specialist, pediatricians, chemical pathologists and dietitians, nurses, social services and support group such as Mucopolysaccharidoses and other rare genetic diseases to provide a comprehensive and multidisciplinary care. Through fostering communication and collaboration among different providers to provide a comprehensive and multidisciplinary care, support and share information to patients and families suffering from rare diseases. It is necessary to launch a comprehensive rare disease centre in Hong Kong which aims at early identification and provision of timely treatment and support to patients and families with special needs. HKMPS hope that further expanding the IEM screening program to more treatable rare disease screening in Hong Kong. These may reflected their needs to relevant decision-maker, strive for a reasonable allocation of community resources and have equal opportunities in the society.

Recommendation

For the current IEM screening program only 24 IEM conditions are included. Is it enough? Of course not. It is only a good starting step. It is necessary for further expanding the IEM screening program to more and more treatable rare disease screening in Hong Kong. Furthermore, to launch a comprehensive rare disease centre in Hong Kong which aims at early identification and provision of timely treatment and support to patients and families with special needs is also very important. Moreover, reflected their needs to relevant decision-makers, strive for a reasonable allocation of community resources and have equal opportunities in the society. There is an urgent need to bridge the knowledge gap by research, educating and connecting patients, families, physicians and specialists in rare disease in Hong Kong. Government urged to sponsor pricey drugs for rare cancer 00:03:02.

References

1. Henrard S, Arickx F (2016) Negotiating prices of drugs for rare diseases. Bull World Health Organ 94(10): 779-781.
2. Rare Disease Day. What is a rare disease?
3. Direct Relief Access to Therapies for Rare Diseases.
4. But WM, Hui J (2014) The Hong Kong medical diary 12: 1-2.
5. Policy addresses (2017) Health care. Disease prevention. p. 242.
6. Poon WK (2014) Overview of inborn errors of metabolism in Hong Kong. The Hong Kong medical diary 12: 10-12.
7. The Chinese University of Hong Kong (CUHK) (2017) Joshua Hellmann Foundation Newborn Metabolic Screening Program. pp. 1-20.
8. Yau KC (2014) Paediatric neurotransmitter diseases (PNDs) due to disorders in the biogenic amine pathway. The Hong Kong medical diary 12: 23-26.
9. Chong SC (2014) Expanded newborn metabolic screening: working towards a mandatory screening programme in Hong Kong. The Hong Kong medical diary 12: 5-7.
10. Press Releases LCQ 21: treatment of rare disease 2013.
11. Mucopolysaccharidoses and rare genetic diseases mutual aid group (HKMPS).
12. National organization for rare disorders (NORD). AdvocatePolicy Issues.

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