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Towards government-funded special biomedical research programs to combat rare diseases in China

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Summary **Rare diseases are rarely conditions that are often debilitating and even life-threatening, which was identified by the World Health Organization (WHO) with a prevalence of 0.65-1‰. 5,000-7,000 rare diseases are thought to exist, which account for around 10% of diseases for individuals worldwide. It is estimated that over 10 million people were patients with rare disease in China. During the past years, public awareness of rare diseases has in fact heightened with the launching of campaigns by patients' organizations and spontaneous efforts by members of the public, not only in developed countries and regions including United States of America (USA), the European Union (EU), and in Japan, but also in China. However, the features of missed or delayed diagnosis, shortage of effective drugs, and the high cost of currently available drugs for rare diseases make it an important public health issue and a challenge to medical care worldwide. To combat rare disease, the government should assume the responsibility of taking on the important task of promoting the sustained development of a system of medical care for and research into rare diseases. Government-funded special biomedical research programs in the USA, EU, and Japan may serve as a reference for China coping with rare diseases. The government-funded special biomedical research programs consisting of leading clinicians and researchers to enhance basic and applied research on rare diseases were expected to be launched in China.**

Keywords: Rare diseases, orphan drugs, public health, medical care

1. Introduction

February 28, 2015 marks the eighth international "Rare Disease Day" coordinated by EURORDIS. On and around this day, over 650 awareness campaigns have been held by patients' organizations in more than 80 countries and regions worldwide in line with this year's theme, "Day-by-day, hand-in-hand" (*1*), with the purpose of launching a blast of upsurge for the attention on patients with rare diseases.

Public awareness of rare diseases has in fact heightened over the past ten years with the launching of campaigns by patients' organizations and spontaneous efforts by members of the public. One example is a charity effort known as the "Ice Bucket Challenge" that went "viral" via social media in the United States of America (USA) and then spread to the rest of the world in the summer of 2014 (*2*). This effort firmly established awareness of a rare disease known as "amyotrophic lateral sclerosis (ALS)" in the public consciousness. Even though "Rare Disease Day" campaigns are held every February since 2008 and events like the "Ice Bucket Challenge" occasionally surface like a flash in the pan, the serious challenge begins when the fever of spontaneous non-governmental efforts eventually dies down, who will fill the "vacancy" and maintain attention on and provide support to patients with ALS or other rare diseases?

2. Rare disease as an important medical and social issue

Rare diseases are rarely conditions that are often debilitating and even life-threatening, which was

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identified by the World Health Organization (WHO) with a prevalence of 0.65-1‰. It is estimated that the combined number of patients suffering from rare diseases in USA and the European Union (EU) exceed 55 million, and 5,000-7,000 rare diseases are thought to exist, which account for around 10% of diseases for individuals worldwide (*3,4*). It is well-known that rare diseases are an important medical and social issue (*5*). With the features of missed or delayed diagnosis, shortage of effective drugs, and the high cost of currently available drugs (*6,7*), the non-governmental spontaneous activities are clearly not enough to improve the plight for patients with rare diseases. Given this reality, the government should assume the responsibility of taking on the important task of promoting the sustained development of a system of medical care for and research into rare diseases.

In addition to efforts to specifically define and classify rare diseases, specific legislation has been drafted to encourage discovery and development of orphan drugs, and the health insurance system for rare diseases has been improved in many countries and regions, such as the USA, EU, Australia, Japan, and South Korea (*8*). Government-funded special biomedical research programs to enhance basic and applied research on rare diseases should also receive full attention. Evidence has shown that biomedical research on rare diseases could provide insight into the pathologies of these diseases and revealed their underlying mechanisms (*9-11*). Such work may ultimately reveal avenues to potential therapeutics. Moreover, once biomedical research identifies suitable drug candidates and becomes more translational, it will garner industry attention and potentially lead to safe and effective orphan drugs.

3. Government-funded special biomedical research programs in USA, EU, and Japan

In Western countries, many research centers or projects have been established to support special biomedical research programs on rare diseases and development of orphan drugs, such as the Office of Rare Diseases Research (ORDR) established in the USA in 1993 within the National Institutes of Health (NIH) and the Rare Disease Task Force (RDTF) established in the EU in 2004 within the European Commission Public Health Directorate (*8*). In Asian countries, biomedical research on rare diseases has made great advances in Japan due to the system known as the Specified Disease Treatment Research Program established in 1972 with the support of the Ministry of Health, Labor, and Welfare (MHLW) (*12*).

Over the past 5 year, new efforts have been instituted to combat rare diseases through government-support biomedical research in Japan. A project entitled the "Early Exploratory Clinical Trial Bases for Specific Research Areas" was launched in 2011 in order to promote innovative drugs and medical devices from Japan for treatment of rare diseases. Pursuant to this project, 5 academic institutions were selected as national early exploratory clinical trial bases for specific research areas including cancer, cerebral and cardiovascular diseases, neuropsychiatric disorders, and immunological rare diseases. Two of the bases - the School of Medicine in Keio University and the University of Tokyo Hospital - have formulated specific plans to develop new drugs to treat immunological and neuropsychiatric rare diseases, promoting the translation of orphan drugs from basic studies to clinical use. For each base, the Japanese government invests up to 500 million yen for infrastructure construction and 150 million yen for clinical trial research led by physicians (*13*). Furthermore, the "Revision of Measures to Combat Intractable Diseases" was approved by the MHLW on January 25, 2013. This ordinance highlights "government-funded special biomedical research programs to enhance basic and applied research on rare diseases" as one of three pillars with which to combat rare diseases in Japan. One hundred and thirty diseases have been targeted by special research programs and research grants from the government, and allocated funds increased to 11.3 billion yen in 2013 (*14*).

4. Rare diseases research in China: current status and future perspectives

In China, although rare diseases have yet to be officially defined due to a delay in legislation, over 10 million patients have rare diseases based on to the WHO definition of rare diseases and the incidence of rare diseases in other countries (*15*). During the past ten years, with the efforts from patients with rare diseases and their families, patients' advocacy organizations, health-care professionals, lawyers, and representatives of the People's Congress, the public awareness of rare diseases has indeed increased in China, however, the official definition of rare diseases, the specific legislation to encourage research of rare diseases and development of orphan drugs have not been established until now.

In China, support for special biomedical programs on rare disease research comes mainly from the National Natural Science Foundation of China (NSFC). Data show that 366 projects (involving 32 rare diseases) were funded by the NSFC from 1999 to 2007 with total funding of 89.358 million RMB and annual funding of about 10 million RMB; this amount represents just 1/10th of similar funding in the USA (*16*), which lead to the lag in benefiting Chinese patients with rare diseases through better diagnosis and more treatment choices.

To combat rare diseases in China, the measures of legislation to confirm the definition and classification of rare diseases, assembling accurate epidemiological data

on rare diseases, incentives to encourage manufacturers to develop orphan drugs are urgently needed. The government-funded special biomedical research programs performed in USA, EU, and Japan have showed that it could benefit to prompt research on the prevalence, diagnosis, treatment, and management of rare diseases, then to improve the quality of life for patients with rare diseases. The government-funded special biomedical research programs consisting of leading clinicians and researchers to enhance basic and applied research on rare diseases were expected to be launched in China.

5. Conclusion

The features of rare diseases make these conditions an important public health issue and a challenge to medical systems worldwide. To combat rare diseases, the government should assume the responsibility of taking on the important task of promoting the sustained development of a system of medical care for and research into rare diseases. Government-funded special biomedical research programs in the USA, EU, and Japan may serve as a reference for China coping with rare diseases. The government-funded special biomedical research programs consisting of leading clinicians and researchers to enhance basic and applied research on rare diseases were expected to be launched in China.

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