Letter

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Establishing a rare diseases center: Experiences from Western China

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SUMMARY Rare diseases pose unique challenges to health care delivery. In August 2016, the West China Hospital of Sichuan University (WCHSU) established a rare diseases center. This center has created a multidisciplinary team of rare disease experts. The center provides expedited pathways online and offline for patients with rare diseases to save them time and money, to improve their experience, and to increase the hospital's efficiency. At the same time, the center regularly organizes public education campaigns and it offers free consultations to enhance awareness of rare diseases. Establishment of the rare disease alliance and facilitation of 5G-based remote multi-disciplinary consultations will help to improve the level of diagnosis and treatment and to solve problems with diagnosis and treatment encountered by local patients with rare diseases. WCHSU's rare diseases center has been feasible, acceptable, and effective in Western China and it should benefit patients, doctors, and hospitals. The center should lead to significant improvements in treatment for patients with rare diseases. The successful establishment of a rare diseases center here may be a useful reference for other parts of the world.

Keywords rare diseases, center, experiences, China, platform

Rare diseases refer to diseases with a very low prevalence. There are more than 7,000 rare diseases recognized internationally, accounting for about 10% of human diseases; 80% of rare diseases are genetic, and 50 to 60% occur in childhood (*1-3*). Because of the large number of people affected, patients with rare diseases are actually "not rare" in China (*4*). Due to the complex etiology, heterogeneous symptoms, and the limited forms of examinations, rare diseases are often undiagnosed and there are few treatment options (*5*). Other challenges include a lack of information and resources, the financial cost of care, and difficulty in accessing appropriate medical expertise, which is compounded by a lack of specialist training programs for medical professionals (*6*). The current situation needs to be improved urgently (*7*).

In August 2016, the West China Hospital of Sichuan University (WCHSU) established a rare diseases center in order to provide better medical care for patients with rare diseases in Southwest China and better treatments and therapies for patients with rare diseases in China. WCHSU is a prestigious and well-known medical center located in the City of Chengdu, Sichuan Province. This is the first medical facility to establish a rare diseases center among hospitals in China. The center has created a team of multidisciplinary rare diseases experts, devised procedures for center operations, and provided patients with rare diseases with an expedited pathway for faster diagnosis and treatment. Specialists specifically provide patients with one-stop care.

In February 2018, the center officially launched an online rare disease platform to disseminate information about rare diseases and to facilitate remote diagnosis and treatment – "Huaxi Rare Diseases," the center's official WeChat account. This is the first online platform built specifically for patients with rare diseases in Chinese hospitals. The platform's features include submission of applications for diagnosis and treatment of rare diseases, online consultations, introductions to experts in various departments, a rare disease encyclopedia, and special medical treatments. Patients with rare diseases can submit diagnosis and treatment applications on the platform. If the patient's application is approved, the center will make an appointment with a specialist clinic for the patient. In addition, doctors also provide patients with online outpatient diagnosis and treatment services through video and audio online so that patients with rare diseases can see a doctor without leaving home.

In November 2018, the center took the lead by establishing the Rare Disease Committee of the

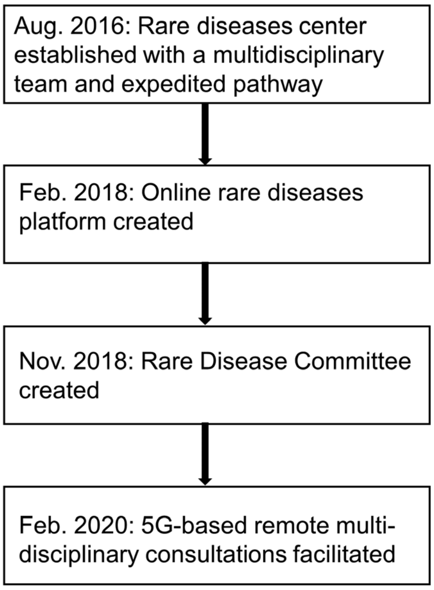

Figure 1. Flowchart depicting establishment of the rare diseases center.

Sichuan Medical Association. The committee regularly provides continuing education to doctors to promote and encourage the spread of new technologies, new drugs, and new findings related to the diagnosis and treatment of rare diseases. The committee also regularly organizes public education campaigns in order to disseminate medical information about rare diseases, to improve the public's awareness and level of self-care, to standardize the treatment of patients with rare diseases, to provide more information to patients with rare diseases and their families, to encourage a positive attitude among patients with rare diseases, and to improve their quality of life.

In February 2020, the hospital launched a new 5G web-based, real-time video telemedicine system for consultations. A multidisciplinary team deals with cases of rare diseases to serve patients and to help improve the level of diagnosis and treatment. The hospital plans to coordinate with hospitals at all levels to regularly conduct remote rare disease consultations and case discussions.

From August 2016 to February 2020, the center helped a total of 1,185 patients with rare diseases on-site, and it provided assistance to 2,169 including patient registration and admission to hospital. Two campaigns online reached more than 60,000 people. The WeChat account has 7,531 followers, it has published a total of 81 messages on rare diseases, and its messages have been read 100,209 times, with an average of 1,237 times per article. The online platform has received 866 patient applications, including 431 patients with rare diseases and 435 patients who do not meet the center's requirements. Of the 431 patients with rare diseases who applied for diagnosis and treatment online, 306 (71.0%) visited offline. Submitting an application *via* the platform to a consultation offline takes 0-7 days, and the average time to a consultation is 4.31 ± 2.18 days. The rare disease committee has organized three large-scale academic conferences on rare diseases to describe the latest advances in clinical and scientific research on rare diseases to more than 3,000 people, and it has organized more than 80 public events and free clinic visits. In February 2020, a survey of 100 patients with rare diseases and 50 doctors at primary hospitals and this hospital found that all of the respondents were 100% satisfied with the center.

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