

Professor Shi-Wen Wu: One City, One Doctor—building up the national DMD registry network

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Professor Shi-Wen Wu (*Figure 1*), MD & PhD, is director of the Department of Neurology in General Hospital of Chinese Armed Police Forces, Beijing, China. After completed his doctorate degree in General Hospital of the People's Liberation Army in Beijing, Prof. Wu continued his education as a research fellow in National Center of Neurology and Psychiatry (NCNP) in Japan during 2004 to 2005. He has more than 50 publications with him listed as the first author or the corresponding author, some of which are published on prestigious international journals such as *Brain* and *Neurology*.

Prof. Wu's expertise has been focusing on neuromuscular diseases, neurological genetic diseases, and so forth. Among all the neuromuscular diseases that Prof. Wu has been dealing with, muscular dystrophy, particularly the Duchenne Muscular Dystrophy (DMD), has become his main research field and makes him a well-known professional in China.

Over the past 3 years, Prof. Wu and his team has been endeavoring to improve the life quality of DMD patients in China. In addition to the establishment of a DMD multidisciplinary clinic, Prof. Wu has also pioneered the Chinese Genetic Disease Registry (CGDR), a China-wide registry of people diagnosed with a genetic disease, especially for DMD/BMD and SMA. So far, there are more than 1,000 DMD/BMD patients registered. To facilitate the registry and follow-up process, a nationwide training program named "One City, One Doctor", is also under preparation. CGDR is the one and only official organization in China that is recognized by TREAT-NMD Neuromuscular Network.

In June 28th, 2015, I was honored to interview Prof. Wu in Beijing and invited him to share more of his expertise, experience and the stories behind.

DMD: a tragedy only falls on boys

Caused by an absence of dystrophin, a protein that helps keep muscle cells intact, DMD patients suffers from



Figure 1 Professor Shi-Wen Wu.

progressive muscle degeneration and weakness. It is a recessive X-linked form of muscular dystrophy, though in rare cases it can affect girls, it primarily affects boys.

The onset of symptoms happens in their early childhood, usually between ages 3 and 5, characterized by weakness of muscles of the hips, pelvic area, thighs and shoulders. Later, muscle weakness also occurs in the arms, neck, and other areas. By the age of 8 to 10, these patients will have difficulty walking. Because the heart and respiratory muscles also are affected, they will suffer heart dysfunction and be in need of cardiac and respiratory support. Most of the patients eventually die because of heart and lung failure.

"Boys with DMD normally did not survive much beyond 20 years old before. But thanks to advances in cardiac and respiratory care, as well as other systematic treatments, their life expectancy is increasing. In Europe, boys with DMD may live up to the age of 45 years old. However, in China the number is much lower." Prof. Wu told me.

When I asked whether there was a specific average number for it, Prof. Wu was not able to give the answer. Why?

The one and only: establishment of DMD registry in China

“Like ALS (widely known due to the “Ice Bucket Challenge”), DMD is also one kind of rare diseases. But unlike ALS, DMD remains barely known in China. Governmental support hasn’t been enough to finance research, neither does social support for DMD patients.” Prof. Wu later explained. “It is difficult to come out with a precise number of these patients’ life expectancy in China as Western countries do, because we need to do long-term follow-ups.” Considering that there was no registry system in China until 2012, it is now quite clear why we do not have an answer to the question yet.

According to TREAT-NMD, an international network with one of its goals focusing on the partnership with many countries in the registry of neuromuscular diseases, China and Africa are two main areas that do not established the national-wide registry system in 2010.

“The Chinese Genetic Disease Registry (CGDR) was established in September 1st, 2012 and was soon accepted by TREAT-NMD as the official registry system in China. So far, there are more than 1,000 DMD/BMD patients and 100 SMA registered,” said Prof. Wu, “with all the data in the registry system, we will be able to know the number of DMD patients and their current condition in China. Most importantly, it will help us learn more about the disease and seek for more effective therapy.”

To emphasize the profound significance of the registry system, Prof. Wu gave me an example. “As a rare disease, the number of DMD patients is not as big as common diseases like hypertension or stroke. In Japan, it has to take 10 years to have 1,000 cases registered. But since the population base is huge in China, we reached to that number in less than 3 years. If a big clinical trial is recruiting patients, only four cases per year are from Japan, partly due to the limitations of the recruitment. Under such circumstance, the trial may need 10 years to be completed. But since now China has the registry system, if they work with us, the time circle of the trial will be largely shortened. The more cases registered, the more choices we have. The strength of one single center is weak, while of a multi-center is strong. After all, the registry comes to end for the good of patients.”

One City, One Doctor: connect the dots and build up the network

When the registry first started, Prof. Wu encouraged

patients to submit the required materials by themselves. To ensure the accuracy of all data, a registry office was set up to check all the materials, and then helped finally upload them to the system. But one important issue occurred: it was easy to upload the primary data, but hard to keep them updated.

“The register office will make calls to catch up the patient’s updated status. We also suggest patients should come to our multidisciplinary clinic in Beijing every half a year, so that we can gather new data. But for families with low incomes, the cost is too much. In addition, because there are not many doctors in China specialized in neuromuscular diseases, it leaves them with very few choices. In other words, the long-time follow-ups will take too much time and cost both from the registry office and the patients.”

Prof. Wu told me.

That’s how the “One City, One Doctor” project comes to the plan. The goal of this project is to facilitate the registry and follow-up process by having at least one doctor specialized in neuromuscular diseases in one major city. *“In this way, it will be easier for patients to have their periodical checks. The doctor will be responsible for renewing the patients information in the registry system, as well as entering information of new cases.”* Prof. Wu said.

The “One City, One Doctor” project is still under preparation. In cooperation with Chinese Research Hospital Association and DXY (the largest physician community with 1.5 million active registered users), Prof. Wu expected this project would officially initiate in early 2016. By connecting the dots of every major city in China, a systematic registry network will be established and definitely work with more efficiency.

Catching up with the advanced: researches on the way

With the registry system, Prof. Wu and his team are capable of doing more studies on DMD, one of which is the use the sniff nasal inspiratory pressure (SNIP), a simple invasive test to detect early degeneration of inspiratory muscle of DMD patients. In another research, Prof. Wu found out that DMD patients had microstructural changes in splenium of corpus callosum, which, for the first time, was suspected of relating to the cognitive changes of DMD patients. *“Though further studies are still needed, the result of this study opens a new door for us to know more about the disease.”* Prof. Wu told me.

Currently, there is no known cure for DMD. Steroid drugs can slow the loss of muscle strength, but they have many side effects. Other treatments aim to control symptoms to improve quality of life. There are several

new treatments being studied in trials. *“In China, however, we do not have any DMD related clinical trials being done or undergoing.”* Prof. Wu said. *“We have two clinical trials in plan and hopefully they will bring good results to our patients. In addition, we are also planning to build up a precision medicine laboratory next year, because gene therapy plays a very important role nowadays.”*

To breathe is to live: fighting for the patients

As one kind of rare diseases, DMD does not catch too much attention from social welfare or governmental financial support in China. *“I saw a big gap between China and Japan in taking care of DMD patients. By the age of 8 to 10, these DMD kids will have difficulty walking. In Japan, government offers every DMD family two chances to install a lift for free in their house (most houses in Japan has two floors). Japanese social welfare also includes free provision of respiratory machine and wheelchair for these families.”* Prof. Wu said. *“We performed a survey on the life quality of DMD patients in China this year and found that the gap is even bigger. In China, these patients are more likely to be ‘Left-behind Children’ without parental care. In some cases, they are the reason of a broken home. Without enough care, no wheelchair and no respiratory machine, most of them die in their teenage years.”*

Prof. Wu wants to do more for the patients. However, it is never easy to carry on the research work of a rare disease. In most of the time, it is quite lonely. What drives Prof. Wu

to strive?

“Every 2 years, we will organize an international symposium to discuss advances in DMD rehabilitation and treatment. The symposium will receive attendees like healthcare professionals, both international and national, as well as families of DMD patients. Prof. Jes Rabbek, director of the Danish National Rehabilitation Centre for Neuromuscular Diseases, was invited to the symposium in 2014. He shared with us a survey that I will never forget.” Prof. Wu said. *“When being asked if there is any life quality left for a DMD boy with his trachea open and intubated, breathing only with the help of a respiratory machine, only 10% of the majority said they might live a life with quality, only 20% of doctors opted the positive, but 80% of the patients said ‘Yes, this is life’. As long as they are breathing, their hearts beating, they feel life. I think that’s the ultimate destination of all my work goes—to extend these boys’ life and make them enjoy what’s left for them.”*

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Footnote

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