POOLING OF EXPERTISE : THE NEW MODEL OF CARE BORN OUT OF NECESSITY IN THE FIELD OF RARE DISEASES

DECEMBER 2022

Interview with Professor Marshall Summar - Director, Rare Disease Institute Laboratory, National Children's Hospital in Washington DC, USA

MEET MARSHALL SUMMAR

The needs of Persons Living with a Rare Disease (**PLWRD**) are linked specifically with the rarity of their condition, for which there is often a **lack of research and expertise**. The scarcity of experts, which translates into limited knowledge, directly impacts the quality of care provided and therefore, the life expectancy of the 300 millions PLWRD worldwide.

Over the past few years, the pooling of expertise among experts in clinical and research networks has gradually become the new model of care in the field of rare diseases. **How was this made possible?**

Professor Marshall Summar, well-known for his pioneering work in caring for children diagnosed with rare diseases and leader of the Division of Genetics and Metabolism at **the National Children's Hospital in Washington DC, USA,** accepted to provide further insight into why pooling of expertise has become a central part of caring for Persons Living with a Rare Disease.

This conversation is the fourth of a series of interviews with members of the Panel of Experts, a group of over 200 clinicians, researchers, administrators, academics and civil society leaders, supporting the development of the Global Network for Rare Diseases.

INTERVIEW

Could you tell us more about the context in which the need to establish a pool of expertise in rare diseases emerged?

Over the past 20 to 30 years, we have seen an **explosion of information in the field of rare diseases**, as many previously undiagnosed diseases have now been described at the molecular level and can now be linked to an actual clinical phenotype(*). Thanks to this breakthrough, we are able to identify an average of four new diseases per week, **raising the number of recognized rare diseases to more than 7500**. And this number continues to grow rapidly.

(*) the term phenotype is used in clinical medicine for speaking about the presentation of a disease.

This **progress in research creates both opportunities and challenges for experts in rare diseases**. As it is impossible for one center to handle all the information alone, it has required them to collaborate and embrace digital technologies to be able to exploit all this new data. This is why the networking of experts, notably through the implementation of **digital platforms,** has become essential to provide care in the field of rare diseases.

What are the benefits of pooling expertise in the field of rare diseases?

There are many outstanding instances of pooling expertise. Let's take the example of the beneficial impact of the **development of centers of expertise on cystic fibrosis.** By pooling expertise, the **life expectancy** of patients with this rare disease has nearly tripled, without the need for a new drug, but simply by making everyone aware of the information that was available in the world on this disease.

This has led the way for others to follow. **More and more expertise centers started connecting** with each other to share best practices, protocole, and knowledge on specific identified rare diseases. Networking and collaboration at national, regional and global levels are not only important, but **essential** to build an evidence base and provide the best possible care for all PLWRD.

How has this new model of care helped create a stronger evidence base for rare diseases?

Providing timely diagnosis and appropriate care to PLWRD involves **working on treatment guidelines in a new way** in which pooling of expertise is central.

To understand why, we need to **step back from the traditional approach** to building the database for determining disease characteristics, which usually includes the following steps. First, initial publications describing a disease and the molecular change rely on many patients to establish a pattern. Only then do most subsequent publications tend to focus on variations or exceptions encountered by the practitioners. The classical way has value when you have large patient cohorts for research, which is, as you know, rarely the case in the field of rare diseases. **So how can we proceed when the experience of a few practitioners becomes not only the best evidence, but the only evidence you have available?**

Creating a **digital space for experts** to foster dialogue and discussion on a case-by-case basis appeared as the best solution to build strong evidence bases in the field of rare diseases. It allows experts who are working on the same condition to connect to share their experiences, letting them all know what worked well or did not, and allowing all the practitioners to be aware and able to comment. **A sort of 'Wikipedia meets Reddit'** where the experts can put down in real time the different approach they are adopting to create an interactive database and progressively building a protocol together.

IIn terms of the use of technological information, have you noticed a shift toward virtual care and telehealth?

Before the pandemic, we were treating about 5% of patients a year using telehealth. This number quickly reached 100% during the pandemic, and has now stabilized at **about 40% of patients preferring to be received virtually**. This new tool is **especially important in remote and low-and-middle income countries,** where patients have less access to experts . There is simply not enough healthcare workforce for patients in those regions (1 expert for a million of people to sometimes only 1 for 4 millions) and these digital tools help to address this shortage by extending the expertise of practitioners beyond their clinic and countries.

How do you see pooling of expertise developing in the future?

I hope to see more and more formalisations of existing collaborations between experts, and centers themselves.

This has been led in the United States by **NORD (National Organization for Rare Disorders),** an independent and third party to the hospitals. The national organization have designated 31 centers of excellence as rare disease centers, based on a certain amount of expertise, personnel, training activities and facilities. Our goal now is to enable patients and clinicians to easily find the centers with the expertise they are looking for, for their particular rare disease by extending this practice to all regions of the world.

For more info on Marshall Summar work for PLWRD

- Visit Children's National Website: **https://childrensnational.org/visit/find-a-provider/marshall-summar**
- Read 'Marshall Summar, M.D., receives Lifetime Achievement Award for rare disease work' : **bit.ly/3BwSVzu**

Please find this article and others to come on the **Rare Diseases International Website**

Stay tuned for the next Global Network for Rare Disease expert article

Follow us on social networks to be informed of the release of the next articles

#GN4RARE

RARE DISEASES INTERNATIONAL