Jesus Navarro – President, Mexican Organization for Rare Diseases (OMER)

We need to treat all people with dignity, diagnose them properly, and give them the best care possible

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Dr Jesus Navarro explains how his personal experience with his daughter's rare disease led him to form the Mexican Organization for Rare Diseases (OMER) to support others facing similar challenges. Dr Navarro highlights the lack of a comprehensive registry for rare diseases in Mexico and the need for specific legislation and a dedicated commission to improve diagnosis, treatment, and resource allocation for rare diseases. He also emphasises the crucial role of the pharmaceutical industry in providing information, training, and support for public policy and fundraising, advocating for closer collaboration to ensure patients receive necessary treatments.

What motivated you to specialize in rare diseases and how has your career developed?

My experience with rare diseases spans 19 years. The need to delve into this topic arose with the birth of my second daughter, Ximena. She was born with a severe metabolic condition, and at ten months old, we managed to obtain a diagnosis and access to treatment, which was uncommon at the time. The diagnosis was mucopolysaccharidosis type 1, Hurler variety.

Once diagnosed, the next critical step was ensuring my daughter maintained the treatment, which we managed to do.

When Ximena was born, we found that her life was at risk due to this disease. We were fortunate to get a quick diagnosis and access to specific treatment the next day. It was a process full of challenges and much luck; we had to form a civil association to defend the rights of people with rare diseases to access and maintain their treatment, as their lives depend on it. Understanding this situation and how it affected our daughter's life made us put in all efforts to learn about the rights of these people and demand their fulfilment. This had very good results: Ximena resumed her treatment, along with other people in the country, which gave us the certainty that we were doing the right thing.

How did this influence your work in the Mexican Organization for Rare Diseases?

We realized that other rare diseases, despite being considered of low prevalence, were also not receiving adequate treatment, even though it was more accessible than ours. Leaders and people who needed treatment started consulting us, and we saw the need to form a conglomerate of rare disease associations. This is how the Mexican Organization for Rare Diseases was born, offering information, advice, and support to those in need.

How would you summarize the work done by the Organization?

At the Mexican Organization for Rare Diseases, we have created a structure to offer guidance, information, and support. Guidance is provided when someone contacts us from anywhere in the country, region, or the world. We tell them where there are associations in their area that can help and inform them if the pathology has treatment or not. We guide regionally to avoid unnecessary travel.

We have also developed a resource sheet indicating where specialists are located. We guide them on where to find associations and pathologies so they can receive information and create a community. If patients don't find the information they need and want to form an association for a specific pathology, we guide them. We consider ourselves facilitators and trainers.

With the International Rare Diseases Congress that we have created, we organize an annual scientific event for training doctors, first-contact doctors, pediatricians, general practitioners, gynecologists, and researchers. This year, we will invite experts from the United States and Spain. These doctors will lecture on diagnosis, available treatments, and necessary tests, and give references to local associations.

We do not provide treatments, but we guide families on what to do. We have scientific and family references. If someone suspects their child has a rare disease, we refer them to the appropriate expert.

According to the WHO, a rare disease is defined as one that affects one person per 10,000 births. What is the prevalence of these diseases in Mexico, and how does it compare globally?

In our region, we have not been able to agree on how to classify rare diseases. While in Colombia, it should be five per 10,000 inhabitants, in Mexico, we have adopted the UN and Orphanet classification, which is one per 10,000. Another difference is that in Colombia, Peru, and Ecuador, they are called orphan diseases, not rare diseases.

Considering this classification, in our country, diseases like cystic fibrosis and hemophilia are no longer considered rare due to their higher prevalence.

How well developed are the processes for registration and treatment of these diseases in Mexico?

In our country, we have been denied the opportunity to have a registry of people with rare diseases. In 19 years, we have made many attempts, even with the General Health Council, to prioritize this need, as is done in Europe and other parts of the world, but we have not succeeded. Therefore, we lack vital statistical, population, and epidemiological data to plan and know how many rare diseases affect Mexicans, where we are, the ages of those affected, and what they are being treated with.

We are spending a lot of money treating undiagnosed or misdiagnosed rare diseases. There is no timely diagnosis; we never get there in time. Imagine this: globally, there are 7000 rare diseases indexed in a well-organized and classified database called Orphanet. In our country, we don’t know how many of these diseases affect Mexicans.

And in terms of legislation?

We do not have specific legislation that considers this type of disease, although in the General Health Law, we worked on articles 223bis and 224 to include rare diseases and orphan drugs. This classification already exists, but undiagnosed people are being treated, spending resources without benefit for their pathology.

Access to these medications has many regulations and basically must be through the National Health System. There are cases where private insurers cover these treatments, but few people have this type of insurance. Although there is legislation, it is not enough for everyone, and improvements are needed.

What improvements do you think are necessary to support people with rare diseases?

To improve this, it will be crucial to create a Commission for the Follow-Up and Registration of Rare Diseases. This collegial group of rare disease experts would have the most valuable opinion on the incorporation of new molecules and would be responsible for the registry of people with rare diseases, allowing better planning.

This Commission would also oversee where and how these diseases are treated, in addition to pharmacovigilance and the evaluation of new technologies. They would guarantee therapeutic success and make budget planning based on the people who need treatment. This way, we could know how many people with rare diseases there are, how many receive treatment, how many could not receive it, and why.

Globally, no country has registered all 7000 rare diseases. Spain is the closest, with 4000 or 5000 registered pathologies, but it does not reach 7000. Of these, only 3% have specific treatment, that is, around 400 treatments.

Talking about rare diseases is talking about many pathologies, but not all have treatment. However, all need something, whether specific treatment or palliative care to improve the quality of life. We need to treat all people with dignity, diagnose them properly, and give them the best care possible. At the Mexican Organization for Rare Diseases (OMER), we work to address this issue and improve the situation of people with rare diseases in Mexico.

This requires a lot of work and coordination. How do you see the role of the pharmaceutical industry in this context?

The pharmaceutical industry plays an important role by offering information and training to associations. They help us with public policy and fundraising workshops and connect us with global experts. However, we need closer collaboration to ensure that patients receive the necessary treatments.

What are the biggest challenges you have faced in these 19 years?

In my 19 years with the organizations, the biggest challenge has been getting health officials to understand the need to plan and manage resources efficiently for rare diseases. The solution lies in forming a collegial team that helps with the traceability, follow-up, and pharmacovigilance of treatments.

I don’t think creating a National Institute of Rare Diseases is feasible; we should take advantage of the installed capacity with diagnostics and the existing National Institutes. By applying what is already written in the legislation, we could resolve many things.

What message would you like to leave to our readers?

We need to work together to build a collegial body that gives order and structure to the treatment of rare diseases. Health is a fundamental pillar of the economy and national security. If we have a healthy and working population, we can move forward as a country. We must focus on early diagnosis and neonatal screening to prevent long-term problems. Additionally, we should take advantage of our installed capacity and make efficient use of our resources.

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