POVERTY, HUNGER AND DISABILITY - THE MISSING LINK

From Policy to Practice with People with Rare Diseases

The sixth global webinar in the series sponsored by IFRA and IDF on 7th Sept 2023

The Honorary Convener of IFRA and IDF, Balakrishna Venkatesh (venky), gave a warm welcome to all participants and presenters to Webinar #6 in the series on Poverty, Hunger and Disability – The Missing Link. The previous five webinars, where good practice examples of interventions in resource scarce communities had been showcased, were held in 2021-22. This webinar introduces examples of good practice across the spectrum with people with a wide variety of rare diseases. Venky emphasized little work was being done in this area and that much still had to be done to reach those persons with rare diseases, especially those living in resource scarce communities.

He identified that two recent resolutions passed by the World Health Assembly do not explicitly recognize persons with rare diseases as a distinct group. He acknowledged that advocacy by the disability movement had helped draft the "UN General Assembly Resolution on Persons Living with a Rare Disease and their Families." This webinar was requested by those living with and working with persons with rare diseases to help influence WHO, the UN focal agency on health, to include "Persons Living with a Rare Disease and their Families" in its strategy and program. Some facts for why WHO should act now include:

- Rare diseases affect 1 in 2,000 people, 400 million people worldwide.
- The exact definition of a rare disease varies from country to country.
- Due to the small number of people affected by each disease, research and development of treatments has been limited, making it more difficult for patients to receive adequate care.
- 95% of persons identified as having a rare disease do not have access to USFDA approved drugs.
- 80% of rare diseases occur at birth and are genetic; 50% are children with a 35% mortality in the first year.
- High costs of identification and care have an economic impact and can lead to poverty; "orphan" drugs are prohibitively expensive.
- There is around 40% error in diagnosis during a patient's first visit with little collaboration among healthcare ecosystems.
- There are negligible specialty diagnostic labs and few doctors & paramedics trained on rare diseases.
- There is a lack of specialty in healthcare infrastructure.
- There is a lack of patient support/advocacy.

Given this scenario, IFRA and IDF pledge their efforts to work with others to change this situation so that the concerns of people with rare diseases become more visible at all levels, leading to inclusion of rare disease treatment in health systems.

Four speakers shared examples of how community-based care is being provided for people with rare diseases in Singapore, Vietnam, India and China. View the welcome address at: https://youtu.be/WM-JNtBXx1A?si=ZP2ZhzCRy_pWslLz

The first speaker was **Aparna Mittal,** CEO and Founder of PatientsEngage, a social enterprise and online platform based in Singapore and India for patients dealing with chronic disease and their caregivers where they can share personal experiences, information and learn from each other – Inform, Engage, Empower. Rare disease management is complicated and filled with uncertainty. It helps persons with rare diseases and their families navigate the health care system and the challenges in the diagnostic process for care and

treatment. Her topic, *"Rare Diseases and Rehabilitation — A lived experience perspective,"* provided examples of the benefits of providing spaces for people with rare diseases and their families to share their stories and information with each other. She described the experiences of three persons with rare diseases, all of whom had suffered the consequences of spending 10 years or more without an accurate diagnosis – years of not knowing, difficulties navigating treatment and health professionals, all in search of help. She affirmed many diseases are genetic so require constant and supportive rehabilitative care. To have a better quality of life, persons living with rare diseases need defined protocols, better information, and access to others who share their journeys. Mittal concluded, "No one should have to deal with their condition alone." View Aparna's presentation at: https://youtu.be/nTg3zyh2h7A?si=r4G5jhLCS7XPpbBD

The second presenter was **Dr. Huy Phuong Do**, CEO and Co-Founder of the Vietnamese Organization for Rare Diseases (VORD). Established in 2021, VORD is a patient support and social enterprise group which promotes research, diagnosis and treatment for people with rare diseases. They provide education, advocacy and patient services in urban, semi-urban and rural communities. His topic, *"So Rare So Care: Helping rare disease patients and families to live a better quality of life,"* provided examples of the importance of collaboration among patients, their communities, and global partners. He described how VORD supports persons with rare diseases through activities such as patient care, research collaboration, policy advocacy, and raising public awareness about rare diseases. VORD works with those who are experienced with rare diseases, including patients, families, medical professionals and organizations, to accelerate diagnosis, treatment, genetic solutions, and patient advocacy. He gave examples of the support given to three organized groups of people with particular rare diseases and how VORD has helped them improve the quality of their lives by providing education and rehabilitation classes, as well as access to free medical equipment such as ventilators and wheelchairs. He summarized by reinforcing the importance of getting community and policy directions in place to support persons with rare diseases. View Huy Do's presentation at: https://youtu.be/Es2GnttMzMc?si=3mf7WnBEjBh1tO-R

As the third speaker, **Smitha Sadasivan** shared her lived experience as a person living with a rare disease, multiple sclerosis, and the advocacy she has been leading for cross disability and inclusive health rights in India for the past 19 years. Her topic, *"People with Rare Diseases — the India Experience,"* gave a powerful insight into the situations faced by persons with rare diseases and their families in India, where there is no official definition of what can be called a rare disease. There has been some movement on this by the government — some funding support is now in place for research at the National Research Development Program and the Indian Council for Medical Research. As a result of significant advocacy by civil society organizations, the government has also created 11 Centres of Excellence in hospitals for new-born screening, thus improving diagnosis time and increasing awareness of rare diseases. Civil society continues to work collaboratively with governments, patients and their families to bring people together, to sensitize and create hope for persons with rare diseases. View Smitha's presentation at: https://youtu.be/NRwltrcMNRE

The final speaker of the webinar was **Lijia (Mary) Sun.** She is a child and family psychologist who has served as the technical director of Our Family China since 2012. She chairs the child psychology committee of the China Association for Rehabilitation of the Disabled and is a member of many regional professional committees. Our Family China provides professional rehabilitation and social inclusion services to children with rare diseases living in a variety of geographical and socio-economic contexts using a holistic and community-based approach. She spoke about *"Empowering Rare Disease Patients: A journey of rehabilitation, innovation and social inclusion."* She described the early support given to persons with rare diseases through international co-operation between organizations in Italy and China. As with other speakers, she reiterated there is no unified definition of a rare disease in China, yet almost 72% of rare diseases are genetic and manifest in childhood. Using a community-based rehabilitation (CBR) approach, and in collaboration with other partners, teams travelled to cities in different parts of China to provide CBR

training to local people, and during COVID started an online service to provide rehabilitation guidance which continues today. This wholistic approach brings together families, civil society, and communities, to provide services such as hydrotherapy camps, family support groups, and organizing a Rare Disease Day across the country to focus on awareness and social inclusion. View Mary's presentation at: https://youtu.be/tGBSpPcwmwE

The final moments of the webinar were dedicated to a conversation between participants and speakers, and a round of thanks to the presenters, interpreters, organizers, participants and funders of the webinar. A single recording of the entire webinar can be found at: https://us02web.zoom.us/rec/share/LuV27JZTocBqfD7TAb4MAbDE08aKuLoxBJrX5MhINYUbODz0lfY1Ec1JfW7XVnNJ.Er9tt_KsdJgYajFj