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Often neglected, rare diseases now have a collective voice

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India is home to an estimated over seventy million patients affected with one of more than seven thousand rare diseases. The number of patients with known and novel rare diseases is increasing every year and it is estimated that 1 in 20 Indians are affected by one of the seven thousand rare diseases. Due to the inadequate infrastructure or the lack of awareness at various stakeholder levels, rare disease management is growing into a major healthcare concern in India.

There is much to be done at the policy level to address the needs of rare disease patients. The absence of budget allocation for research is a major bottleneck in developing a quality healthcare delivery mechanism for rare disease patients in India. Since there are seven thousand types of rare diseases and each of these diseases affects a very small population there are severe constraints in conducting scientific and clinical research and development of a nationwide newborn screening regimen. This has lead to limited therapeutic and diagnostic modalities for rare disease patients.

Although there have been individual organizations and persons working in the area but the need for a strong collective voice was important for pursuing pending issues with the government.Therefore to address that, the Organization for Rare Diseases India (ORDI) was formally launched by the founding team members in a function held at the constitution club of India on February 18, 2014. The launch ceremony was attended by eminent scientific and medical advisors of ORDI, representatives of the rare disease stakeholders and key opinion leaders in health care policy and adminstration in India.

ORDI's vision is to make rare diseases in India diagnosable and treatable just like other common diseases such as diabetes, breast cancer and cardiovascular disease etc.

ORDI's mission is to be the collective voice of every rare disease patient, family member, and healthcare professionals in India by pooling together expertise, efforts, and resources to help early diagnosis, treatment and management of rare diseases in India.

Commenting on the launch, Dr Vijay Chandru, founder member ORDI said, "There is a genetic origin for at least 80 % of rare diseases and in 50 % of cases the onset occurs in childhood. Rare diseases also include rare cancers, autoimmune diseases, congenital malformations and infectious diseases among others. A majority of these diseases lack proper diagnosis and treatment options. Through ORDI we will try to empower rare disease patients and their families in India with access to national and international resources and help improve their quality of life. This would also mean the development and delivery of affordable diagnostics and treatments for rare diseases through innovative collaborations and partnerships among multiple

stakeholders."

ORDI will open doors for patients to access facilities and opportunities by registering with patient advocacy groups within India and overseas, such as Jain Foundation for diagnostic testing, patient registries and clinical trials. Jain foundation is a patient advocacy group focused on Limb Girdle Muscular Dystrophy 2B and is heavily involved in creating opportunities in India for clinical trial readiness. Establishing patient registries, and systematic treatment protocols to finding a cure for rare diseases is the ultimate goal of these joint efforts," said Dr. Madhuri Hegde, founder member ORDI.

"Patients suffer physically, mentally and financially as the families knock on the doors of various hospitals seeking diagnosis. Even after proper diagnosis, there is little hope for cure. It is in this context ORDI has an important role to play in pooling together expertise, efforts, and resources to help patients and health professionals share information on rare diseases across India," said Prasanna Shirol, founder member ORDI and former President of Lysosomal Storage Disorders Support Society.

"The whole education and healthcare system is more geared to cater to common diseases such as infectious diseases, diabetes, cardio vascular disease and cancer. This lack of awareness has created a dearth of resources and clinical care for patients with rare diseases," said Sangeeta Barde founder member ORDI.

"Information technology (IT) has emerged as a greatest enabler of recent times. ORDI is determined to leverage IT in serving rare disease cause by being an umbrella organization for information contribution, collection, consumption and propagation on rare diseases. ORDI also serves as a platform that unites, educates and connects rare disease patients, doctors, scientists, volunteers, evangelists and organizations with the help of latest technological breakthroughs." Ravinandan ME, founder member ORDI quoted.

"Rapid advances in applying molecular diagnostics tools such as next generation sequencing and bioinformatics to diagnose patients worldwide across the human disease spectrum will have a highly beneficial impact in the diagnosis, treatment and management of rare disease patients. ORDI sees exciting real opportunities emerging out of the molecular diagnostics revolution in enabling personalized medicine for rare disease patients in India", said Dr. Preveen Ramamoorthy, founder member ORDI.

"Our review of the state of rare disease management in India identified the various challenges and opportunities in India (currently under review by a peer-reviewed journal). The need for an umbrella organization to engage all rare disease stakeholders in India became evident. ORDI serves as this common platform to initiate open engagement among and strengthen the unified voice of rare disease stakeholders in India." continued Dr. Harsha Karur Rajasimha, founder member ORDI "We are very encouraged by the enthusiasm and support of several national and international rare disease organizations for our mission. Rare Genomics Institute (RGI) represents one of our early partners in USA. RGI is helping to facilitate crowd funding for genomic-based research to help undiagnosed and economically disadvantaged patients with rare disease in India. Several Indian disease-specific patient advocacy organizations and biotech industry bodies have confirmed their collaboration with ORDI." - he concluded.