Press Release
February 18th 2014

***Organization for Rare Diseases* India (ORDI)**
officially launched today…

New Delhi: 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.

To bridge this gap and work as a catalyst that can bring about a change in the manner in which rare diseases are diagnosed and treated, a group of eminent healthcare leaders, NGO's, doctors and government bodies have come together to launch a first-of-its-kind 'Organization for Rare Diseases' in India, ORDI

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. We dream of a day when all Indian citizens affected by rare diseases will have equal access to diagnosis, treatment and management of their condition.

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.

How many rare disease patients are there in India?
Reproduced with permission from Dr. Navneetham (http://rarediseasesindia.org).

Table 1

Countries in South Asia	Rare Diseases and Disorders Population [1-8]
Afghanistan	1,530,006
Bangladesh	9,151,081
Bhutan	44,099
India	72,611,605
Maldives	19,037
Nepal	1,589,670
Pakistan	10,999,800
Sri Lanka	1,216,656

Table 2B

Union Territories (India)	Rare Diseases and Disorders Population[4]
Andaman & Nicobar Islands	22,797
Chandigarh	63,281
Dadra & Nagar Haveli	20,571
Daman & Diu	14,575
Lakshadweep	3,866
NCT of Delhi	1,005,194
Puducherry	74,668

Table 2A

States (India)	Rare Diseases and Disorders Population[4]
Andhra Pradesh	5,079,932
Arunachal Pradesh	82,957
Assam	1,870,156
Bihar	6,228,278
Chhattisgarh	1,532,412
Goa	87,463
Gujarat	3,623,018
Haryana	1,521,185
Himachal Pradesh	411,391
Jammu and Kashmir	752,936
Jharkhand	1,977,974
Karnataka	3,667,842
Kerala	2,003,261
Madhya Pradesh	4,355,854
Maharashtra	6,742,378
Manipur	163,305
Meghalaya	177,840
Mizoram	65,461
Nagaland	118,836
Orissa	2,516,841
Punjab	1,662,254
Rajasthan	4,117,261
Sikkim	36,461
Tamil Nadu	4,328,337
Tripura	220,262
Uttar Pradesh	11,974,891
Uttarakhand	607,005
West Bengal	5,480,864

ORDI accomplishments:

- ORDI has initiated a national rare disease hotline number **8892 555 000 a valuable resource to the rare disease patients seeking support and care.** We have already received hundreds of calls on this hotline and are keen to help the patients who call us. We have developed a full proposal to develop this hotline into a comprehensive rare disease care coordination center. **Rare Disease Care Coordination Center** will improve the traceability of patients

suffering from rare diseases, guiding them to the right doctors for correct diagnosis followed by treatment and management. One proposal is ready and is being considered by corporate entities

- We have initiated a corporate council to engage with the industry in India and in a short span we have a few companies that have pledged sponsorship for ORDI.
- We are in the process of sending out a letter inviting all existing rare disease organizations and NGOs to partner with ORDI in working towards our common goals.
- Reviewed the state of rare diseases in India in a manuscript under review by a peer-reviewed Journal.
- ORDI will establish collaborations with national and international rare disease organizations. In partnership with **Rare Genomics Institute, USA,** ORDI facilitates the enrollment of economically disadvantaged rare disease patient groups in India for crowd funding for genomic research and patient-care.

These initiatives are aimed at providing both patients and health professionals with improved access to medical information, treatment centers, patient support groups and epidemiological/research data.

Steps to be taken by ORDI

- **ORDI will assert itself as a national advocate capable of influencing policies** on rare diseases. It will be strongly involved in the preparation and follow up of the National Plan on Rare Diseases. ORDI will offer to work with government policymaking agencies to help formulate a version of (Orphan Drugs Act) ODA for India.
- **ORDI will build a rare disease portal** that integrates disparate databases, bio repositories, and patient registries to enable rare disease translational research and recruitment for clinical trials.

Name of the founder members and their brief biographies

Harsha K. Rajasimha, MS, Ph.D.

Serves as a founder member of the Organization for Rare Diseases India and is academically affiliated as a faculty in the School of Systems Biology at George Mason University, Fairfax, VA, USA. Harsha has over a decade of experience working on various inter-disciplinary projects involving systems biology, genomics, medicine and BigData computing for academia, USA government and industry**.** He is the founder CEO of a Bioinformatics services company – Jeeva Informatics Solutions that he founded in 2013. Most recently, Harsha joined Strand Genomics Inc. and focuses on implementing clinical genomics services in community hospitals in USA.

Madhuri Hegde, PhD, FACMG
Is an American Board of Medical Genetics certified clinical molecular geneticist and is the Executive Director of Emory Genetics Laboratory and a Professor of Human Genetics and Pediatrics at Emory University, GA, USA. She is a founder member of the Organization for Rare Diseases in India. Dr Hegde is current of Chair of Genetics subdivision of Association of Molecular Pathology and serves on several committees of the American College of Medical Genetics and Genomics, American Society of Human Genetics, and College of American Pathologists. Dr. Hegde has several grants from NIH and other private foundations to develop novel approaches to implementing precision medicine as it relates to genetics and its integration in medicine.

Prasanna Kumar, MBA

Founder President of LSD Support Society (Lysosomal Storage Disorder Support Society), the first National Level Parents support group for children suffering from Rare Diseases in India. The society initiated the observance of International Rare Disease day for the first time in India on a national scale under his leadership. He has worked extensively to create awareness on LSDs and Rare diseases in India. He has represented India in various conferences and meetings internationally and developed a strong network of international Parent and Rare disease groups.

Prasanna Kumar has Post Graduate degree in Business Management has over 16 years of sales experience in telecom and consumer industry. Worked with companies like Sony, Reliance, Bharti etc. He has experience in launching product and services. Personally, he is married to Sharada and has a 14-year daughter. His daughter Nidhi is India's first Pompe Patient and also runs Pompe Foundation supporting Pompe Patients in India.

Preveen Ramamoorthy, MS, PhD

Serves as a founder member of the Organization for Rare Diseases in India and is Vice-President, Clinical Diagnostics at Strand Life Sciences. He serves on numerous advisory boards of biotech companies and non-profit organizations in the USA Dr. Ramamoorthy founded the Molecular Diagnostics department at National Jewish Health, one of the world's leading centers for respiratory and immune diseases and currently serves as an adjunct professor for translational research at National Jewish Health. Prior to joining National Jewish Health Dr. Ramamoorthy was involved in the development of vaccines and monoclonal antibody therapeutics at Medimmune. He is passionate about enabling personalized medicine in the clinic through the application of integrated molecular diagnostics approaches.

Ravinandan, ME

A Technologist, Dreamer, Doer. Loves to be in thick of things. Wishes to make a positive impact with the help of technology. On a mission to democratize technologies. Evangelist of Lean principles and rare diseases awareness.

Sangeeta Barde, MMM

A pathfinder of Sorento with ever-futuristic vision, ability to think beyond, inquisitive mind and deep commitment to healthcare. With an ability to conceptualize and execute complex solution architects has devised new products and services for Sorento. Over 22 years' experience in Pharmaceutical Marketing and Healthcare Communications

Trained as a Microbiologist and with a Master's in Marketing Management Sangeeta has crafted strategies for more than 100 healthcare brands

While continuing with her commitment to healthcare devotes time to the cause of rare diseases and keeps the vision of changing a state of rare diseases in India

Vijay Chandru, PhD, FASc, FNAE
Dr. Vijay Chandru is a founder CEO of the translational biology company Strand Life Sciences for which the World Economic Forum has recognized him as a Technology Pioneer in 2006. Dr. Chandru was a founder of Picopeta Simputers, which commercialized the handheld computer called the Simputer. He received the Dewang Mehta award for innovation in information technology from the Ministry of Communications and Information Technology in 2001 for this work with the Simputer.

Dr. Chandru serves on the Global Advisory Board of the MIT Enterprise Forum and

the Governing Board of the Indo-USA Science and Technology Forum. In 2003, he helped start the Association for Biotech Led Enterprises (ABLE), the apex body of the Indian Biotech Sector, and has served as the Founder Secretary (2003-2006) and as President (2009-2012). Dr. Chandru is a member of the Vision Groups for Biotechnology and Science & Technology for the state of Karnataka. He is a fellow of the Indian academies of science and engineering and an adjunct professor of the International Centre for Theoretical Sciences (TIFR) in Bangalore.

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Rare Diseases Overview

By definition, a rare disease occurs infrequently in a population, but there is no universal definition. In the USA, a rare or orphan disease is defined strictly according to its prevalence, specifically "any disease or condition that affects less than 200,000 persons in the United States," or about 1 in 1,500 people. It is estimated that approximately 7,000 rare diseases affect over an estimated 30 million Americans. European Union (EU) considers diseases affecting fewer than 1 in 2,000 persons to be rare. Between 7000 and 8000 different rare diseases affect about 29 million people in the EU alone. In Japan a rare disease is defined as affecting not more than 1 in 2500 people. Other countries have developed definitions that are appropriate in the context of their own health care system. India, like most countries, currently has no standard definition. Considering the population of India the suggested threshold for India for a disease to be defined as rare is 1 in 10,000. This may seem like a small number but it is important to note that when all rare diseases are taken together, the number of patients affected is significantly large. Roughly, over 70 million patients with rare diseases are estimated to live in India with about 3.7 million patients in the state of Karnataka alone.

The exact number of rare diseases is not known but is estimated to be around 7000-8000 worldwide. With the rapid advances in genomic technologies in the last decade, the number is steadily increasing each year with new diseases and associated genes being identified. About 80% of rare diseases are genetic disorders, many of which are thought to be monogenic. Rare diseases also include rare cancers, autoimmune diseases, congenital malformations, and infectious diseases amongst others. All rare diseases taken together affect about 6-8% of the population. About half of the rare diseases affect children causing significant social and economic burden, while the remaining manifest in adulthood. Some examples of rare diseases include Hemangiomas; Hirsch sprung disease, Gaucher’s disease, Cystic Fibrosis, muscular dystrophies and Pompe disease.

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