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01 September 2014 | News | By Rahul Koul Koul

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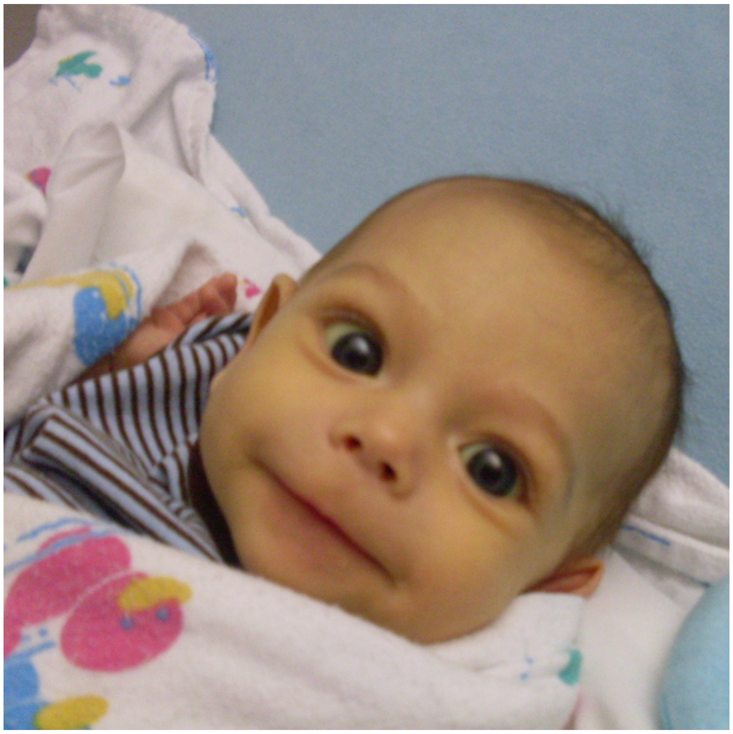

Recently in New Delhi, experts from across the globe came together for the first South Asia Meet on Lysosomal Storage Disorders (LSD) which result becasue of the missing or dysfunctional enzymes from the lysosome of a cell. LSDs are a group of around 45 rare genetic disorders, found in about one in every 5,000 live births in the world. Most LSDs are managed through disease-specific supportive care measures in the absence of any treatment or cure.

While most patients in India remain unaware of the fact, the prominent doctors informed the audience that 6 out of 45 rare and debilitating genetic diseases are now completly treatable with enzyme replacement therapy (ERT). These treatable rare diseases include Gaucher Type I, Fabry's disease, Pompe disease, MPS I (Hurler disease), MPS II (Hunter disease), and MPS VI (Maroteaux-Lamy syndrome).

Present at the event held on August 30, was Dr V M Katoch, secretary, Department of Health Research, and director general, ICMR, who was the chief guest. Dr Katoch accepted that India, due to its competing priorities in healthcare has neglected the rare diseases. "Their cause is currently being promoted by few individuals in the medical community, but we need a proper and permanent organizational structure and network in the government and healthcare system to identify these diseases, conduct research, put together documentation and draft a practical plan to tackle rare diseases. There is also a need for collaboration with SAARC countries because people in the region have the same genetic makeup and dietary habits. Not many medical companies have moved into the space of rare diseases in India because of miniscule volumes and lack of incentives from the Government. This has to be rectified. Another challenge is the lack of diagnostic tools to identify most of the rare diseases. We need a structured approach to the problem to make a difference. It is a difficult task but can be done in the next few years," explained Dr Katoch.

Currently, there are 8 to 10 centers of excellence in the country for the management and treatment of LSDs, and 6 to 8 centers have diagnostic capabilities. In Delhi-NCR, two hospitals - AIIMS and Sir Ganga Ram Hospital - provide free counseling and treatment to 30 patients of LSDs, who are registered with LSDSS, a non-profit organization that works for the cause of patients with rare diseases. Genzyme, a part of Sanofi, has been running an initiative called the "India Charitable Access Program" under which it provides free treatment to around 100 children suffering from LSDs.

As per Dr I C Verma, director, Centre of Medical Genetics, Sir Ganga Ram Hospital, and patron, Society for Indian Academy of Medical Genetics, "Enzyme replacement therapy (ERT) is critical to improve the quality of life for patients suffering from Lysosomal Storage Disorders and is a new hope to these patients. Without treatment, there is a progressive accumulation of partially degraded compounds which stealthily leads to multi-organ dysfunction."

The South Asia Meet on Lysosomal Storage Disorders has been organized by the Indian Society for Inborn Errors of Metabolism (ISiEM), Society for Indian Academy of Medical Genetics (SIAMG) along with the Genetics Chapter of the Indian Academy of Pediatricians (IAP) and supported by Genzyme. Expert clinicians, practitioners, academicians, and researchers from across India and South Asia participated in the two-day event (August 30-31, 2014) to discuss latest advancements in the diagnosis and treatment of LSDs.

Prominent speakers at the meet included Dr Shubha R Phadke, professor and head, Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow; Dr R Premlatha, director, Indira Gandhi Institute of Child Health, Bangalore; Dr Jayesh Seth, director, Institute of Human Genetics, Ahmedabad; Dr Pramod K Mistry, professor of Medicine, Yale University; and Dr Alberto Ortiz, professor of Medicine, University Autonoma of Madrid (UAM), Spain.

Dr AP Dubey, professor and head, Department of Pediatrics, Maulana Azad Medical College, said, "Misdiagnosis and delay in treatment can play havoc in the lives of these patients. Lifelong treatment with ERT allows them to lead a near normal and productive life, but unfortunately most of them cannot afford it as the treatment is very expensive and beyond the means of almost everyone."

Experts at the meet agreed that the major roadblock in tacking LSDs is the high cost of treatment with biological drugs. These drugs are different from chemical drugs and entail considerable research and safety benchmarks. Their R&D and manufacturing costs need to be recovered from the patient population, which is miniscule compared to that of common diseases like diabetes and cancer. Moreover, all the medicines of rare diseases are imported into India, adding to the costs.

Ms Meryl Sarah, a Delhi-based patient of Pompe disease, brought cheer to the audience when she narrated her story. She said that initially she faced a lot of hardship because of misdiagnosis but is happy today. With access to ERT, Ms Sarah is able to lead an active and normal life and shows no visible symptoms of the disease. "I am lucky to have access to this life-saving treatment. I am optimistic about the future as my condition is much better now. I hope all patients of these rare diseases get access to timely ERT treatment to lead normal lives," she added.