

Parents, children fight rare disorder

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CHENNAI: She might be living with a rare genetic disorder and its debilitating effects, but that has not stopped 14-year-old Krithikaa Ramesh from dreaming big.

Diagnosed with Lysosomal Storage Disorders (LSD), Krithikaa wants to become a scientist; the one thing she wants to accomplish is finding a remedy for LSD.

At the age of one, Krithikaa was diagnosed with mucopolysaccharidoses, a type of LSD, which is a group of 45 genetic disorders caused due to deficiency of specific enzymes in special compartments (lysosomes) of cells. Since then, she has undergone seven surgeries, including one for knee correction. But her family has ensured that Krithikaa does not lose out on her studies.

"She has completed class VIII in a regular school. But she needs assistance while walking. So, we plan to help her write the SSLC exam as a private candidate next year," says R. Bhuvaneshwari, her mother.

Krithikaa and her family



Actor Karthi interacts with children diagnosed with Lysosomal Storage Disorders at an event organised by a support society — PHOTO: R. RAGU

are not alone in their battle against the disorder. There are nearly 600 registered children with LSDs in India, with 350 in the Tamil Nadu centre of Lysosomal Storage Disorders Support Society (LSDSS).

LSDs occur in about one in 5,000 live births, affects multiple organs, and causes developmental delay, movement disorders, seizures, deafness and dementia.

For many parents, under-

standing LSDs has been a long and difficult process. C. Shanmugam, a farmer from Kutteripattu near Tindivanam, had no idea what his son, S. Sabarish, in class III now, was suffering from. "People think it is like a mental disorder but LSD is completely different. Many organs suffer damage," he said.

Many parents have been fighting a long battle against a lack of awareness, for healthcare facilities that

would facilitate an early diagnosis, treatment in India and above all, supportive care.

V. Selvi recalls how she hunted for help for her son V. Gunal, now aged nine.

"I knew that something was wrong with my son when he was six months old as his chest bone was protruding. I went from one hospital to another but nobody knew anything. He was diagnosed with LSD at the age of three," she said.

Six of the group of disorders can be treated by Enzyme Replacement Therapies (ERT). Around 100 children are receiving ERT through a charitable access programme of a U.S. company, Sujatha Jagadeesh, consultant in Clinical Genetics, Fetal Care Research Foundation, said.

As the treatment involves huge costs, parents have urged the government to come up with a comprehensive scheme to fund treatment. Meanwhile, actor Karthi, who is the cause ambassador of LSDSS, has appealed to the government to waive import duty on medicines treating LSD.