ATTENTION

JYOTI KAPOOR

Deepak Kumar with his 11-year-old daughter Palak.

WHERE PEOPLE FIND SUPPORT THEY NEED

By **Harsha Chawla**
in New Delhi

A GLIMPSE into the struggles of the parents who have set up the first LSD Support Society of India will help you realise that the recently released film *Extraordinary Measures* based on a highly-motivated man's quest to find the treatment for his children suffering from the rare disease Pompe is more true to life than imagined.

Prasanna Shirol struggled for eight years, making rounds of over 45 hospitals in the country to obtain the correct diagnosis of his daughter's illness. This journey compelled him to set up a group for parents facing similar struggles.

Luckily, he wasn't alone in his endeavour to start the first of its kind patient-group network: Many like-minded parents and doctors were keen to come together to create a strong voice for LSD patients in India. "Most of us bumped into each other during our rounds at different hospitals. All of us wanted to do something for those who lack access to specialised doctors or treatment," says Prasanna Shirol, president of the society.

Prasanna Shirol

The LSD society organised the first International Rare Disease Day in the Capital recently, bringing together parents and doctors to share their experiences at Sir Ganga Ram Hospital. Sponsored by pharmaceutical company Genzyme Corporation, the occasion drew about 90 parents from different parts of the country. "We were relatively lucky to get in touch with doctors such as Dr I.C. Verma and Dr Ratna Puri who guided us in the best possible way. One of the main priorities of the society is to put the parents in touch with the right doctors," says Deepak Kumar, a core member of the society.

Kumar considers the society a leverage group to create awareness in parents as well as doctors, connect parents to specialised doctors and make treatment accessible. The LSD society is also working towards arranging corporate sponsorships for treatments that are both unavailable and unaffordable in India.

TIMELY DIAGNOSIS AND TREATMENT SAVED HIS LIFE

By **Manu Moudgil**
in New Delhi

Suyog at age 17.

HE WAS only three years old when he was diagnosed with Gaucher's disease, a lysosomal storage disorder (LSD). Enzyme deficiency causes Gaucher's, leading to the buildup of a fatty substance in the spleen, liver, lungs, bone marrow and sometimes the brain too. "I was lucky that only my spleen was affected. I had a large tummy but skinny legs. I was also low on energy because of anaemia which is one of the fallouts of Gaucher's disease. I studied and played indoors," Suyog recalls.

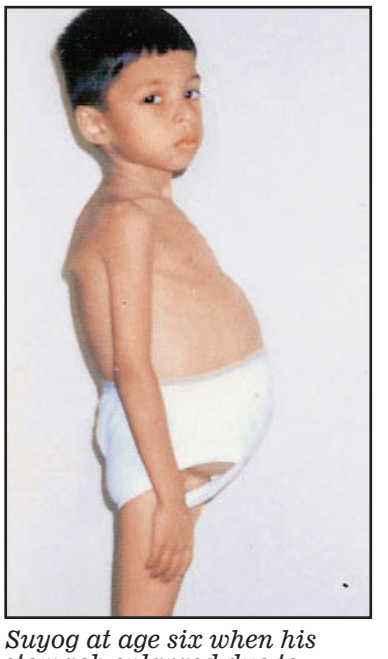
The real breakthrough came after his spleen was surgically removed at the age of six. He later contacted a US-based pharma company Genzyme Corporation, which provided him a free supply of drugs. "Fortunately, my treatment was started at a young age otherwise there could have been irreversible changes

Drugs are out of reach because of high cost

like stunted growth and low bone density. The medicine is now doing the job of maintenance by breaking down the fat molecules which tend to accumulate," says Suyog, who is now a healthy 17-year-old studying engineering.

Suyog is one of the lucky few with access to treatment. While rare diseases affect millions across the world, very little progress has been made on the treatment front. Only six out of 45 LSDs are treatable right now. While medicines do increase the life quality and expectancy of those affected, very few companies produce them, due to high production cost and low return on investment since the patient group is extremely small. The impossibly high cost of these medicines (Rs 25 lakh to Rs 1 crore annually) make them inaccessible to most.

The only ray of hope remains



Suyog at age six when his stomach enlarged due to Gaucher's disease.

the compassionate access programme of Genzyme Corporation that Suyog reached out to. This provides drugs for four rare diseases free of cost to 70 Indian patients. "It's not feasible to help everyone but we are committed to provide these drugs for free to selected patients throughout their lives," says Sandeep Sahney, managing director (India & South Asia), Genzyme Corporation.

This means people like Suyog are very few and far between and pharma companies blame the authorities for this state of affairs. While in all developed countries, tax relief, regulatory concessions and even financial assistance are given to companies manufacturing drugs for rare diseases, there is no such provision in India.

"We have raised this issue several times with the Union government, but there has been no positive response yet. There are companies which would like to manufacture these drugs but because of high cost and low return, they require government support," says N.R. Munjal, president of the Indian Drug Manufacturers' Association. In the absence of such a policy, millions of families continue to suffer under the burden of unknown and untreatable diseases every day.

PAINTING GREETING CARDS HELPS HER DEAL WITH HER ILLNESS

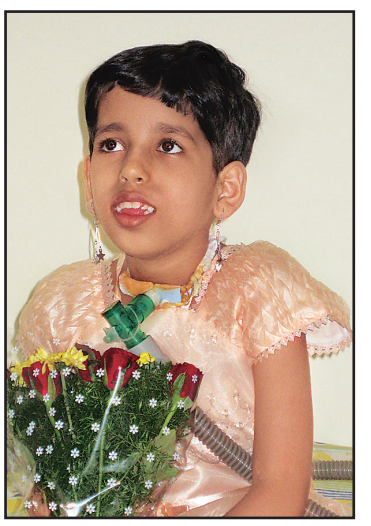
By **Harsha Chawla**
in New Delhi

THIS plucky nine-year-old used the time she spent on a ventilator to learn painting. Today her creative efforts have been turned into greeting cards by the company which sponsors her medicines. Nidhi Shirol is one of a handful of people suffering from Pompe, an extremely rare disease that disables the heart, respiratory muscles and skeletal muscles.

She was born normal, but missed on the key milestones, which doctors named developmental delays.

"Our lives changed when Nidhi's symptoms worsened. We stopped attending family functions or meeting friends. The only time we went out was to visit the doctor," says Nidhi's father Prasanna Shirol. Five years passed before a doctor in Devangare, Karnataka, broke the news to the parents that Nidhi was suffering from Pompe, which is caused by mutations in a gene that makes an enzyme called alpha-glucosidase.

Nidhi has been on a ventilator for the past two years. But her condition has improved since she began myozyme, an enzyme replacement therapy for Pompe disease. About 5,000 to 10,000 people are believed to be suffering from this worldwide.



Nidhi Shirol and her painting (left) in which she expressed her desire to play outdoors.