

# Relief rare for children with genetic disorders

## Parents, Doctors Seek Govt Aid For Treatment

TIMES NEWS NETWORK

**Chennai:** R Madhesh's cry annoyed many in the packed room but for his distraught parents, watching him was excruciating. "He seems to be in a lot of pain, but he's struggling to tell us what's causing him so much discomfort," said Alamelu, mother of the 8-year-old who suffers from a rare genetic disorder.

When Alamelu's efforts to pacify him failed, others in the room — women who have been in her place at some point in their lives — came to help. "We get used to this when we bring up children with rare genetic disorders," said Srikala P, cradling Madhesh. Her own 6-year-old son, suffers from mucopolysaccharidosis — a progressive disorder that mainly affects the brain and spinal cord — tugged at her sari.

On Thursday, the Lysosomal Storage Disorders Support Society (LSDSS) in association with foetal care research foundation organised a programme to create awareness and to highlight the problems faced by parents of children various lysosomal storage disorders. LSDs are a group of

### GENE FLAW

#### What are LSDs?

Lysosomal Storage Disorders are a group of more than 45 rare, genetic disorders that lead to abnormal cellular function

**Effects** | Cause progressive physical and mental deterioration like movement disorders, dementia, heart problems and abnormal growth of bones

### INCIDENCE

- ▶ 1 in 5,000 births
- ▶ Treatable LSDs range from 1 in 30,000 to 1 in 4,00,000 births

more than 45 rare genetic disorders that occur in about one in 5,000 births.

Doctors said, despite various advancements in medical science, diagnosis and treatment of these genetic disorders continue to leave doctors stumped. "Only six of the LSDs can be treated with enzyme replacement therapies," said Dr Sujatha Jagadeesh, consultant clinical geneticist at foetal care research founda-

tion. Most of these diseases are frequently chronic, progressive, degenerative and often life-threatening.

With people suffering from LSDs few in number, parents face several hurdles in locating appropriate healthcare facilities, said doctors.

Parents and doctors at the programme appealed to the government to offer financial assistance for treatment of these debilitating disorders. "As of now, only a few doctors and parents are mustering funds for treatment. Medicines are expensive and cater only to a handful," said LSDSS president Manjit Singh.

A few parents requested the government to step up efforts to undertake genetic screening at birth. "This will also help create awareness about genetic disorders. For over five years, I've heard people in my village say my family is cursed which is why my son was born this way," said Shanmugam, whose son Sabarish suffers from LSD. "I want to tell them there are several parents out there who go through the same struggles I do and, not for a moment, do they stop loving their child," he said.