

Genetic disorders go undiagnosed

DC CORRESPONDENT
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Jothi (38) of Pallavaram has waited more than a decade to get her 18-year-old daughter treated for a congenital disorder that was not diagnosed. Sandhiya has the growth of a six-year-old, weighs only 22.5 kg and cannot perform daily tasks by herself.

Dr Sujatha Jagadeesh, genetics consultant, Fetal Care Research Foundation, MediScan said, "More than 80 per cent of parents of children with genetic disorders were found to have married their relatives." Sometimes, the disease also comes from their ancestors.

"When there are only around 600 registered genetic disorder patients in the country, it is feared that the country at present has over 20,000 who were not diagnosed for the disease. While the foundation has registered more than 350



Actor Karthi Sivakumar giving away a cheque for ₹5 lakh to children affected by genetic disorders at an event in Chennai on Thursday.

patients from the four southern states, around 15 to 20 children were feared to be born with the disease in Tamil Nadu every year. Out of 45 types of genetic disorders, treatment is done only for four types," she said.

Dr Sujatha said it is difficult even for doctors to

diagnose the problem. And so, training should be given to physicians. She also insisted that newborn screening and treatment for all types of disorders be brought to India.

The treatment comes at a high cost. It costs ₹15 lakhs per month for the medicine which has to be taken

throughout one's lifetime and ₹10,000 per month for symptomatic treatment alone. Only around 100 children have access to the treatment in the country free of cost through various charitable access programmes from abroad, said Manjit Singh, president of Lysosomal Storage Disorder Support Society.

The Central and State governments should create a corpus fund for the treatment of these children. Early detection (probably soon after the child is born) and early medication would avoid many complications in the child, he added.

The Fetal Care Research Foundation, in association with the LSDSS, convened a programme in Chennai to observe 'Rare Disease Awareness Month' and 'World Rare Disease Day' (February 28). Actor Karthik Sivakumar, who participated in the programme, donated ₹5 lakhs.