B'lureans who live with them, and daily uncertainties, campaign for research into better treatment options

WITH RARE DISEASES, TREATMENT IS RARER

• Akhila Damodaran

Gagandeep Singh Chandok has thalassemia and seen many of his friends die from this genetic condition. "Raj Kishore, Ali, Imran, Mona Lisa, Manjushree, Asma...", he names them in his online petition asking the Government of India to develop gene therapy, which could save lives of people suffering from several rare diseases.

He writes that he and his friends were thrilled when the research into this therapy was first started in India, but Gagandeep says that it has come to a standstill from lack of incentives and support. He urges the medical community to restart this "critical treatment".

Dr Vijay Chandru, chairman and managing director of Strand Life Sciences, says that the gene therapy may help treat rare genetic disorders because they can modify genetic code. Dr Chandru hastens to add that it is still in experimental stage.

Large Population Affected

While there is no reliable record of people living with rare diseases, Dr Chandru says that a large part of the population has genetic conditions. Of the 1,200 cases that his team had tested, over 60 per cent were genetic and neurological disorders, and about 180 cases were of skeletal issues and the rest skin diseases.

"Fifty per cent of genetic disorders have sympathetic symptoms - the ones that appear at infantry - and the other fifty per cent show symptoms over the course of life," he says.

Principal Secretary to the Government Gaurav Gupta, ITBT, Science and Technology, estimates that nearly 7.2 crore Indians are living with a rare disease and "Karnataka has its fair share of perhaps 30 to 40 lakh patients". He adds, "A drafting committee was appointed to formulate a policy for the state on rare diseases and orphan drugs."

Vision Group of Biotech, headed by Kiran Mazumdar Shaw, is part of this committee and has recently submitted the Karnataka Policy on Rare Diseases to the state government. The policy aims to have an equitable and accessible health system and have in place best practices in diagnosis, care, treatment and management of orphan/rare disease patients in the state. Dr Chandru is also a member of the Drafting Committee and says, "At present the recommendations are under the purview of the state health secretariat and the team is working very closely to refine the policy for its implementation."

In these recommendations as well, the committee has asked for more research into treatment methods.

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– Dr Vijay Chandru

Karanveer with his parents

Muscular Dystrophy

Ravdeep Singh Anand is aware that his son will not live for long. Affected by muscular dystrophy, a rare genetic disorder, his 16-year-old child Karanveer is confined to a wheelchair because of muscle weakening.

"When he was around 2, we noticed that he had difficulty in sitting up on bed or getting up from the floor," says Ravdeep. "In March 2003, when he was a little over 2 years, he was diagnosed with muscular dystrophy." Ravdeep's family flew to the US for a diagnosis because "the technology available in India was not advanced enough". The tests identified Karanveer's condition as Duchenne Muscular Dystrophy. "DMD is caused by the absence of a protein dystrophin that helps keep muscle form of muscular dystrophy."

As a child grows older, he loses his ability to run or climb stairs. It affects the entire body and functioning of all vital organs. In most cases, the child has a cardiac failure before he turns 25.

Ravdeep adds, "We got in touch with a paediatrician from Chennai in 2004 who suggested that Karanveer take steroids." To date, there is no cure or treatment available for this condition. Therefore, Ravdeep quit his job and started the Dystrophy Annihilation Research Trust (DART) in 2012. Along with scientists and a few parents, he researches on therapies that can help treat children with such rare disorders.

His son undergoes physiotherapy regularly. Also, as he takes steroids, it causes side effects like mood swings and weight