

Gene therapy helps teens with rare genetic disorder

Kyodo

Nov 11, 2015

Gene therapy has dramatically improved the conditions of two bedridden teens suffering from a rare disease, Jichi Medical University said Tuesday.

The Tochigi Prefecture-based university said that the finding could lead to the development of treatment for other rare neurological diseases.

The two teens have a deficiency in an enzyme called aromatic L-amino acid decarboxylase (AADC), which is necessary to produce the neurotransmitter dopamine. Patients with AADC deficiency, which emerges within a month after birth, have only minimal motor skills.

There are only six patients diagnosed with the disease in Japan. Patients suffer from repeated seizures and become bedridden before they can hold their heads up. There has not been an effective treatment so far.

In June and July, the university administered an adenovirus containing the AADC gene to four places in the brains of a 15-year-old boy and his 12-year-old sister.

After two months, the siblings no longer suffered from seizures and were able to sit up with help. Their facial expression became calmer. The sister is now able to roll over in bed and take off her socks by herself. She has even started learning to walk.

“We had hoped it would stop the agonizing seizures, which were unbearable to watch. But the result of the treatment was beyond that, and we are surprised,” said their parents in a statement.

Jichi Medical University said there may be similar patients who have been wrongly diagnosed with cerebral palsy, and called on possible parents of such children to seek medical attention at a proper institution.

The university is also conducting clinical research on gene therapy for Parkinson’s disease, which also causes low AADC.

<http://www.japantimes.co.jp/news/2015/11/11/national/science-health/gene-therapy-helps-teens-rare-genetic-disorder/>