# West London toddler becomes youngest UK patient to benefit from pioneering gene therapy for rare brain disorder



A three-year-old girl from west London has become the youngest patient in the UK to receive a revolutionary gene therapy aimed at treating a rare and life-threatening inherited disorder. Gunreet Kaur, diagnosed with aromatic L-amino acid decarboxylase (AADC) deficiency at just nine months old, underwent treatment in February 2024 at Great Ormond Street Hospital (GOSH), the only UK hospital currently offering this pioneering therapy.

AADC deficiency severely impairs children’s physical, mental, and behavioural development by disrupting the brain’s dopamine production. This neurotransmitter is crucial for controlling movement and autonomic functions such as head control, blood pressure, and heart rate. Without a functional AADC enzyme, children experience profound developmental delays, weak muscle tone, and painful episodes, often facing a grim prognosis with many not surviving into adulthood.

Gunreet’s mother has reported remarkable improvements since the gene therapy, which involves a precise, minimally invasive procedure that administers a virus carrying a working copy of the AADC gene directly to the brain. This allows nerve cells to produce the enzyme themselves, restoring dopamine production. Gunreet has begun to reach developmental milestones previously thought unattainable—she holds her head up, smiles more, rolls over, and has improved coordination and vocalisation.

The treatment, known commercially as Upstaza or eladocagene exuparvovec, represents a major advance in gene therapy for rare neurological diseases. It was approved in the UK and Europe for patients over 18 months with severe AADC deficiency. Delivered via robotic-assisted neurosurgery to the putamen region of the brain, it is the first gene therapy of its kind commissioned by NHS England with direct brain infusion. Clinical trials have shown significant motor function improvements, with some treated children gaining independent walking ability and enhanced communication over time.

Experts at GOSH emphasise the transformative potential of this treatment in reducing the painful and disabling symptoms of AADC deficiency. The progress of patients like Gunreet challenges the previously accepted natural history of the disorder, which generally saw little developmental progress after infancy. Moreover, the introduction of such novel therapies is only possible through multidisciplinary collaboration, involving surgeons, neurologists, physiotherapists, and speech therapists, alongside partnerships with pharmaceutical companies supplying the treatments.

This development comes after the National Institute for Health and Care Excellence (NICE) recommended the therapy as a cost-effective option for the NHS, following an agreement ensuring its value to taxpayers. Meanwhile, in the United States, the Food and Drug Administration approved a similar gene therapy, highlighting the global momentum in treating this and other neurological disorders via gene therapy.

While challenges remain—including the high costs and complexity of brain-targeted gene therapies—this approach is seen as a promising frontier not only for AADC deficiency but also for other debilitating diseases such as Parkinson’s, Alzheimer’s, and Huntington’s. Ongoing studies and clinical trials continue to explore expanded applications, potentially heralding a new era of treatments for conditions previously considered incurable.

For Gunreet and her family, the gene therapy has brought hope where there was little before. Her mother reflects on the progress as a profound relief and a glimpse of the possibilities this treatment opens—not just for delaying deterioration but for real growth and quality of life improvement in children born with this severe genetic condition.

### 📌 Reference Map:

* Paragraph 1 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html)
* Paragraph 2 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html), [[5]](https://en.wikipedia.org/wiki/Eladocagene_exuparvovec)
* Paragraph 3 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html)
* Paragraph 4 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html), [[2]](https://www.nice.org.uk/news/articles/nice-recommends-life-changing-gene-therapy-for-children-with-ultra-rare-genetic-disorder), [[5]](https://en.wikipedia.org/wiki/Eladocagene_exuparvovec), [[6]](https://europepmc.org/article/MED/37824694), [[7]](https://www.io.nihr.ac.uk/techbriefings/eladocagene-exuparvovec-for-aromatic-l-amino-acid-decarboxylase-deficiency/)
* Paragraph 5 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html)
* Paragraph 6 – [[2]](https://www.nice.org.uk/news/articles/nice-recommends-life-changing-gene-therapy-for-children-with-ultra-rare-genetic-disorder)
* Paragraph 7 – [[3]](https://www.fda.gov/news-events/press-announcements/fda-approves-first-gene-therapy-treatment-aromatic-l-amino-acid-decarboxylase-deficiency), [[4]](https://www.apnews.com/article/79c0b20862f3f9e2bb4ee977c050489d)
* Paragraph 8 – [[4]](https://www.apnews.com/article/79c0b20862f3f9e2bb4ee977c050489d), [[7]](https://www.io.nihr.ac.uk/techbriefings/eladocagene-exuparvovec-for-aromatic-l-amino-acid-decarboxylase-deficiency/)
* Paragraph 9 – [[1]](https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html), [[7]](https://www.io.nihr.ac.uk/techbriefings/eladocagene-exuparvovec-for-aromatic-l-amino-acid-decarboxylase-deficiency/)

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## Bibliography

1. <https://www.independent.co.uk/life-style/health-and-families/health-news/aadc-treatment-gosh-hospital-nhs-b2775937.html> - Please view link - unable to able to access data
2. <https://www.nice.org.uk/news/articles/nice-recommends-life-changing-gene-therapy-for-children-with-ultra-rare-genetic-disorder> - The National Institute for Health and Care Excellence (NICE) has recommended eladocagene exuparvovec, a gene therapy for children with aromatic L-amino acid decarboxylase (AADC) deficiency. This rare genetic disorder leads to severe motor and cognitive impairments. The therapy, administered via a minimally invasive procedure, has shown potential in improving motor development and offering long-lasting benefits. NICE's recommendation follows a confidential commercial agreement between NHS England and PTC Therapeutics, the therapy's manufacturer, ensuring its value for the NHS and taxpayers. The final guidance is expected in April 2023.
3. <https://www.fda.gov/news-events/press-announcements/fda-approves-first-gene-therapy-treatment-aromatic-l-amino-acid-decarboxylase-deficiency> - The U.S. Food and Drug Administration (FDA) has approved Kebilidi (eladocagene exuparvovec-tneq), the first gene therapy for treating aromatic L-amino acid decarboxylase (AADC) deficiency. This rare genetic disorder affects neurotransmitter production, leading to motor and cognitive delays. Kebilidi is administered through four infusions into the brain's putamen, enabling the production of the missing AADC enzyme. Clinical studies demonstrated improvements in motor function, with some patients achieving head control and the ability to sit unassisted two years post-treatment.
4. <https://www.apnews.com/article/79c0b20862f3f9e2bb4ee977c050489d> - A groundbreaking gene therapy, eladocagene exuparvovec, has been administered directly into the brain to treat a rare genetic disorder, AADC deficiency. This therapy has shown promising results, with patients experiencing significant improvements in motor functions. The treatment, approved in Europe and the UK, is expected to seek U.S. approval. At least 30 U.S. studies are underway for brain-delivered gene therapy for various disorders, including Alzheimer's, Parkinson's, and Huntington's. Despite challenges for more complex diseases and high costs, this approach shows promising evidence, potentially pioneering treatments for several brain disorders.
5. <https://en.wikipedia.org/wiki/Eladocagene_exuparvovec> - Eladocagene exuparvovec, also known as Upstaza, is a gene therapy product for treating aromatic L-amino acid decarboxylase (AADC) deficiency. It involves infusing the gene encoding the human AADC enzyme into the brain's putamen region. This process leads to dopamine production, improving motor function in individuals with AADC deficiency. The therapy is administered via a minimally invasive procedure and has been approved in the European Union and the United Kingdom for patients aged 18 months and older with a severe phenotype of the condition.
6. <https://europepmc.org/article/MED/37824694> - Gene therapy using one-time targeted delivery of a gene vector (AAV2-AADC) directly to the brain has been developed for AADC deficiency. Studies have investigated different brain target sites, including the putamen and midbrain. Eladocagene exuparvovec (Upstaza) was approved in 2022 for patients aged 18 months and older with a severe phenotype of AADC deficiency. Clinical trials have reported improvements in symptoms and motor function, with some individuals gaining the ability to walk independently after treatment.
7. <https://www.io.nihr.ac.uk/techbriefings/eladocagene-exuparvovec-for-aromatic-l-amino-acid-decarboxylase-deficiency/> - Eladocagene exuparvovec is a gene replacement therapy developed for patients with aromatic L-amino acid decarboxylase (AADC) deficiency, a rare inherited disease affecting neurotransmission. The therapy involves transferring the gene encoding the AADC enzyme into the brain's putamen via a surgical procedure. By increasing AADC enzyme production, it enhances dopamine and serotonin levels, improving motor and cognitive symptoms. If licensed, eladocagene exuparvovec will provide the first medicinal treatment option for adult and child patients with AADC deficiency, addressing a significant unmet clinical need.