Treatment gives hope to teen with disorder known as childhood Alzheimer's

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The mother and father watched as their 16-year-old daughter ate her pork chop and green beans across the kitchen table of their Mount Prospect home, grateful each time she swallowed without struggle.

She paused in between bites to sing a few lyrics of her favorite song. "We're all in this toge-e-ether," Hayley Koujaian crooned the theme from "High School Musical," a movie she loved as a little girl, before anyone could tell she was sick.

Hayley has a rare genetic disorder called Niemann-Pick Disease Type C, often dubbed childhood Alzheimer's because its symptoms are similar to those of adult dementia, though it's not the same disease. Memory, speech and mobility fade. It gets harder to eat and drink unaided. There is no treatment approved by the Food and Drug Administration, and young children with the disorder typically don't live past their teens.

"We all knew what the outcome was going to be if the disease progressed," said her father, Harry Koujaian. "There is only one outcome. That's why we were so desperate."

Yet the family has found hope in a promising experimental drug - and the local physician who helped them gain access.

Every two weeks, Dr. Elizabeth Berry-Kravis injects a treatment called cyclodextrin into Hayley via a spinal tap at Rush University Medical Center as her father holds her steady and her mother silently prays.

Hayley is one of roughly 40 patients across the country using the investigational drug, hoping for future FDA approval.

Berry-Kravis, a pediatric neurologist, said there's initial evidence the injections could be staving off the disease's progression. Some skills even appear to be improving.

Before starting treatment a little more than two years ago, Hayley was functioning at the level of an 18-month-old, based on language and cognitive testing. Now her abilities closer match a 2 {- or 3-year-old, Berry-Kravis said. Swallowing is much easier. Her gait is smoother, and she's less apt to fall.

Prior to treatment, speech therapists found Hayley had difficulty putting together a few words. Now she reads simple sentences, responds to short questions and even sings some of the songs from early childhood, before her mind began slipping away.

While the drug is still in clinical trials, it has significantly extended the lives of mice and cats with the disorder compared with those left untreated.

The Koujaians wonder whether it could be extending Hayley's life, too.

"We would not be here, like this," said her mother, Gail Koujaian, gesturing to her family sitting around the dinner table. "Maybe we would be pushing her wheelchair. Maybe we would be cleaning her feeding tube." Not like this, she said.

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Hayley was born Aug. 10, 1999, a chubby-cheeked baby with heart-shaped lips and no hint of any abnormality.

Gail and Harry Koujaian recall their daughter was just like any other toddler, running around with ease and learning how to read and write alongside the rest of her kindergarten friends.

Then in first grade, she mysteriously began falling behind. First teachers noticed she wasn't retaining information. There was odd clumsiness, bumping into walls and desks without explanation. Most alarming, she appeared to be losing abilities she once mastered.

Her mother noticed during Brownie troop meetings that all the other girls could walk alone across a long, thick tube on the playground. Gail Koujaian always had to hold her daughter's hand.

Then Hayley began erupting in seizures, sometimes several in one day. Medication after medication failed, and, at 11 she had brain surgery and her tissue was sent to a pathologist for testing.

Six months and a litany of tests later, the Koujaians got the devastating diagnosis: Niemann-Pick Type C, a fatal disease often appearing in children where naturally produced cholesterol accumulates and becomes toxic. There are about 500 cases diagnosed internationally.

Gail Koujaian recalls days when she would drop off her daughter at school and then cry in her car alone. The Koujaians started meeting other families with the genetic disorder from across the country, which was both comforting and terrifying.

"You've got kids who are dying, kids in wheelchairs, kids who can't eat, kids on feeding tubes," Harry Koujaian said. "It's a cruel disease."

It was another mom who told them about cyclodextrin, which appeared to help her own twin daughters. The National Institutes of Health had just begun an exploratory trial of the drug. The Koujaians jumped at the

chance for Hayley to join the study, but she was found ineligible because of her uncontrolled seizures.

The only other option was to find a physician to file a compassionate-use request with the FDA, a complicated and lengthy process. The Koujaians weren't sure anyone would be able or willing to take on the burden. But they had just started seeing a new pediatric neurologist, Berry-Kravis, who found the most recent research for cyclodextrin so compelling, she wanted to give it a try.

"I can do this," the Koujaians remembered her saying.

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Every other week, the Koujaians drive a few miles to pick up Berry-Kravis at her Des Plaines home, and they all travel together to the clinic at Rush.

Berry-Kravis usually discusses Hayley's progress, typing up her reports on a laptop in the front passenger seat as they sit in traffic on the Kennedy Expressway. It's like a house call in the family's 2006 green Honda Pilot.

Last week at Hayley's 56th treatment, she was put to sleep with melatonin, her thick brown ponytail spilling over the edge of the hospital table, as Berry-Kravis numbed her back with lidocaine.

The neurologist inserted a 3-inch needle with a pencil-point tip into Hayley's spinal canal, first removing 10 milliliters of fluid to be used for research. Hayley didn't open her eyes or appear to be in any pain.

The process has become routine for them. They intermittently joke and laugh softly so as not to wake her.

During these procedures, Gail Koujaian asks God to make sure Hayley doesn't have a seizure; Harry Koujaian wraps his arms around Hayley as a precaution, just in case she does.

Berry-Kravis started injecting the drug through the spinal tap when a car alarm went off seven floors below. Hayley fidgeted a little, and her parents quickly steadied her as Berry-Kravis finished. Then they reclined the table, positioning Hayley's feet up, hoping the drug will drip from her spine to her brain as she continued to sleep.

The first procedure in December 2013 took around 12 hours, with rigorous testing before the injection and prolonged monitoring after. Now the whole visit takes about 2 { hours. Afterward, the Koujaians hang out in the waiting room, talking with the families of other patients with Niemann-Pick Type C.

After Berry-Kravis was approved to treat Hayley, others with the disorder started trickling in to get the injections, too. She is now administering cyclodextrin to 12 patients, some traveling from as far as Minnesota and Texas. Some, like Hayley, are receiving it under compassionate use, others as part of an ongoing clinical trial with a Maryland-based biotechnology company called Vtesse Inc., which is developing the drug.

Questions still linger. Will the drug work long term? Is there a less invasive method? What's the optimal dose?

Outside of Berry-Kravis' office, colorful charts detail some of her patients' progress. If the disorder is untreated, symptoms might plateau for a little while before they worsen, but they don't get better, Berry-Kravis said.

But Hayley's graphs, labeled Subject 1, show improvement in areas like

swallowing, gait and speech.

"There is hope," Gail Koujaian said.