

Health system first in nation to offer treatment for hereditary amyloidosis

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Nancy Kim (right) and her father both live with hereditary transthyretin amyloidosis. Kim was the first patient in the United States to receive this new drug, which stops progression of the disease. Credit: Nancy Kim

UC San Diego Health is first in the nation to offer a new injectable medication to patients with nerve damage caused by hereditary

transthyretin amyloidosis, a rare disease that can be fatal without management. Previously, patients with this systemic disease were tethered to long, intravenous infusion therapy sessions every three weeks. Now, they receive one injection every three months to stop disease progression.

Nancy Kim was diagnosed with hereditary transthyretin amyloidosis in 2018.

"I long suspected I had the condition given my [family history](#), but it was different to get an official diagnosis," said Kim, whose seven family members have had the condition. "My symptoms included numbness and tingling in my feet, [heart palpitations](#) and dizziness, and what felt like severe carpal tunnel syndrome in my right wrist, which would go up to my shoulder. This has all subsided since treatment."

UC San Diego Health, the region's only academic health system, began offering the treatment in July 2022.

The injection is specifically indicated for polyneuropathy, a condition where nerves are damaged by hereditary transthyretin amyloidosis. There is an ongoing clinical trial for its use for other manifestations of transthyretin amyloidosis, such as amyloidosis involving the heart.

"This medication fits under a class of drugs called 'silencers,' which decrease production of the protein that creates amyloid," said Marcus Urey, MD, cardiologist at UC San Diego Health and assistant professor of medicine at UC San Diego School of Medicine. "After receiving the medication in [clinical trials](#), polyneuropathy or pain caused by amyloidosis progressed much slower and symptoms improved in some patients."

There are more than 30 types of amyloidosis that can affect different

organs. Hereditary transthyretin amyloidosis occurs when an abnormal protein accumulates in the blood stream and eventually deposits in a person's organs, causing them to become dysfunctional, and in some cases, fail.

Symptoms can range from shortness of breath, numbness in the hands, constipation or diarrhea.

Often affecting multiple organs, patients with amyloidosis require a multidisciplinary team of experts for treatment; however, obtaining and coordinating timely and [effective treatment](#) for a condition that is difficult to diagnose can be challenging. Many individuals may require an organ transplant as a result of this disease.

Kim's father also has the disease and recently underwent heart transplant surgery. Sadly, six of their [family members](#) have passed away due to complications from the disorder.

Kim is hopeful her father will be able to receive this new treatment soon as well.

UC San Diego Health is one of few centers in California that utilizes a comprehensive approach toward treating amyloidosis, and the only one in San Diego. Obtaining the FDA-approved silencer drug, Amvuttra, was made possible by a team of experts ranging from physicians, nurses, pharmacists and patient navigators.

"We all felt a sense of urgency—this is such an impactful therapy in improving the quality of our patients' lives," said Urey. "We are dedicated to providing novel [treatment options](#) and technologies when addressing amyloidosis to ensure our patients have the best options available."

Urey says the goal is to eventually offer the treatment to all UC San Diego Health patients with the disease. He believes this is just the beginning in finding a potential cure for amyloidosis.

For Kim, she is looking forward to the future with a different lens.

"At times, having an [incurable disease](#) can feel overwhelming, especially because there really aren't many treatment options for amyloidosis available," said Kim. "Advancements like this reassure me and give me hope."

"Now, I have the option of planning my life, even if it's three months at a time. It's a new chapter, a new beginning."

Provided by University of California - San Diego

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