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Scientists race to unravel deadly mystery of Amanda Gellel's one-in-a-million disease

KEVIN VAN PAASSEN / NATIONAL POST

In January of 2001, Diane Gellel died of Lafora disease, a hereditary ailment that causes seizures beginning in the teenage years and growing progressively worse. Three months earlier, Diane's younger sister, Amanda, had her first seizure. She too has Lafora. Amanda, shown above with her parents, is now 18 and doing much better than Diane was at the same age. Diane was 23 when she died.

They found the problem – but how do they fix it?

Funding difficult for research on rarities

BY BRAD EVENSON

The first seizure hit Diane Gellel on the volleyball court. Of the endless thousands that followed, her father still remembers that seizure in 1991 like a fresh nightmare. Sam Gellel was a supervisor at a wheelchair factory in Toronto. He got the call at work.

"At first, we thought it was just epilepsy," he said. But after conducting a battery of tests on the 13-year-old, doctors at Toronto's Hospital for Sick Children had more ominous news.

Diane had Lafora disease, an incurable form of epilepsy so rare most neurologists never see a case in their entire careers.

"You don't hear much about this disease," says Stephen Scherer, a University of Toronto geneticist. "We know of maybe 200 cases in the world."

Neurologists at the hospital confessed there was little they could do to help. A few photocopied pages from the *New England Journal of Medicine* told Sam and Rita Gellel what to expect. At first, the seizures would strike more often. Soon they would come weekly, finally cascading into one another, unstopping, jerking their daughter's body like a marionette and eventually destroying her mind. Along with seizures, Lafora patients suffer dementia, losing their memory, powers of speech and sometimes their vision.

There was worse news. It runs in families.

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