

Boy's rare disease inspires support group

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Evan Penny, who uses a wheelchair, has a mitochondrial disease. (Courtesy Penny family)

A Calgary family whose son has a rare genetic disease is trying to bring together other Canadians suffering the same experiences.

Evan Penny, 5, has been diagnosed with a mitochondrial disease.

He was a happy, healthy child before going in for a routine appendectomy two years ago. He didn't recover properly and is now a quadriplegic. He can't talk or eat and no one knows what his future holds.

Evan's father Blaine Penny said it took a year for his son to be diagnosed with a mitochondrial disease. After the surgery, Evan fell into a coma and had severe brain damage by the time he woke up. He was diagnosed with a neurological disease called acute disseminated encephalomyelitis, but later tests indicated he has an underlying mitochondrial condition. "I had never heard of this disease a year ago. Mitochondrial disease? What is it? So you go to Wikipedia, you look it up. It doesn't paint a pretty picture."

The Pennys are helping organizing the first Canadian conference, set for Hamilton, Ont. in April, aimed at bringing others with mitochondrial diseases, and their families, together. They are also trying to incorporate a not-for-profit organization called MitoCanada. A U.S.-based support group exists, but it doesn't address what's happening in Canada, said Blaine Penny.

"We think having that support is one piece that's really going to help people out of the gate when they are diagnosed with something like this."

Not enough people diagnosed: doctor

Mitochondrial diseases occur when mitochondria, the specialized parts of cells responsible for generating energy, fail, producing less and less of the energy needed to sustain life and support growth.

Depending in which type of cells the mitochondria fail, the central nervous system, liver, pancreas or other organs can be affected. No two cases are the same.

Dr. Aneal Khan, an assistant professor of medical genetics and pediatrics at the University of Calgary, knows of about 35 people in Calgary who have been diagnosed.

"We estimate there probably should be hundreds of people, but because it is so difficult to diagnose, that's probably leading to a much smaller number of people that actually get to a specialist and have that diagnosis," he said.

Starting a Canadian support group could also help raise awareness and lead to more people being properly diagnosed, Khan said.

"With other conditions, support groups give families a home ... where they can do some research for the illness that's affecting their family. So information is one big benefit from having a support group. Support groups are also very good at mobilizing resources. Often times the resources are different in different communities."

Two years after Evan Penny's surgery, he is going to school, and although he can't communicate, he is usually a happy child, his father wrote in his blog.

"The day-to-day challenges of Evan's needs have become the norm now. We are not sad as often and look back less and less as time goes on. It is easier that way. Every time I see a picture of walking, talking Evan I get teary-eyed."