



### Èva Nadeau, 8 years old

Diagnosed with a genetic mutation on the CDKL-5 gene  
(Atypical Rett syndrome)

**“You always hear it, but it’s true: when a child is sick, the entire family’s life is turned upside down. Opération Enfant Soleil donates locally so that children can have their exams, treatments and follow-ups close to home. And that makes a huge difference!”**



Èva was two months old when the first signs of the disease appeared. It began with shivering for one or two seconds. Over time, these tremors intensified and began lasting longer and longer. Eventually, she was hospitalized and taken to the intensive care unit at the CHUS - Hôpital Fleurimont. The shivering continued to become more severe, each episode lasting up to 15 minutes several times a day. It turned into severe convulsions and epilepsy. The medical team conducted numerous tests, including blood tests, spinal taps, CT scans and magnetic resonance imaging on Èva over the following weeks to understand what was wrong with her. They weren't able to pinpoint a diagnosis at the time. After spending two months in the hospital and undergoing various treatments to control her epilepsy, Èva finally went home. However, her parents had to administer several medications morning, noon and night, in addition to feeding her a ketogenic diet. “Her symptoms were under control, but we still didn't know the underlying cause,” said Catherine.

Èva was 6 months old when she was given an official diagnosis. She had a rare condition caused by a spontaneous genetic mutation on the CDKL-5 gene. This genetic mutation causes serious hypotonia, intellectual disability, inability to speak and use her hands, inability to move around and severe, drug-resistant epilepsy. There's no cure and no hope for improvement. Èva will never be able to speak and will be dependent on an adult for the rest of her life.

Her parents were devastated by the news. Their lives were forever altered. However, Èva's father decided to “stand up and carry the family,” “I said to myself, nothing will be the same as before,” recalls Catherine. Despite the immense pain and questions, her parents learned everything they could about the disease. They did everything they could to support Èva and allow their two eldest children to live as normally as possible.

“Together, we've faced a thousand griefs for what our daughter will never be able to do or experience. But we've been able to pick ourselves up and reinvent ourselves as people, as a family. Today, I'm very fortunate to have my beautiful Èva with me. Despite her disabilities and limitations, she teaches us to see and appreciate life differently. I accept the situation as it is and not as I would have liked it to be,” says her mother.

Today, Èva's epileptic seizures are under control. However, she still takes four anti-epileptic drugs three times a day to limit the convulsions, which still sometimes occur. However, she can play, participate in activities and do physiotherapy, occupational therapy and speech therapy. Although other health problems have recently arisen, Èva's determination is boundless. She's a fighter and works tirelessly to gain muscle strength and perform certain movements. She can now sit up on her own and move around on the floor, rolling a short distance. Recently, she propelled herself by pulling with her arms to grab an object. “Èva surprises us every day she defies the diagnosis, and I'm sure she has other wonderful surprises in store for us,” says her father, Jean-François

**“Having our daughter as an ambassador for the cause gives meaning to what we're experiencing. If our story can inspire and help other families, encourage them to persevere and not give up, we're proud to have shared it.” – Catherine**

**Catherine Roy, Jean-François Nadeau, Félix (12 years old) and Laurie (11 years old)**

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