

## *Invisible Project*

2018 Pediatric



### **Cole Swanson**

#### *Staying cool in the face of adversity*

As an infant, Cole Swanson always wanted to be barefoot; he managed to figure out the zippers and snaps of his footie pajamas months before he should have been developmentally able, and always refused to keep socks or shoes on more than a minute or two.

As a toddler, he began to put words to what he was experiencing: his feet were “tingly,” and no matter what he and parents Carl and Carolyn tried, he would burst into tears over things like the feel of his legs on the mattress. Treatment for his initial diagnosis, restless leg syndrome, proved useless, and when little Cole’s feet began turning bright red and the tingles turned to pain, his parents were referred to a specialist.

Luck was on their side. They visited an experienced rheumatologist at the end of a long career who, on their very first visit, diagnosed Cole with a condition he’d only seen twice before: primary erythromelalgia (EM). The average amount of time before diagnosis for most EM patients is nearly a decade, but Cole was diagnosed just after he turned four.

The Swanson family was relieved to know there were answers. They eventually did genetic testing and confirmed a rare SCN9A genetic mutation, “meaning the condition is just a part of who he is,” explains Carolyn. Unfortunately, learning about what he was facing, and the treatment options out there for him, didn’t make them feel lucky at all.

## *Heading towards crisis*

Erythromelalgia is a neurovascular disorder thought to result from dysfunction in the normal constriction and dilation of certain blood vessels, leading to abnormal blood flow and severe burning pain in the small fiber sensory nerves. Attacks can be caused by something as simple as wearing socks and shoes; other triggers include heat, pressure, mild activity, exertion, insomnia, or stress. Essentially, many of the things that make up being a kid!

“When he was very young, he pointed at a TV screen full of static and said, ‘My feet feel like that,’” explains Carolyn. “But at the height of the crisis he used words like ‘fire’ and ‘lava.’”

The only treatments the medical establishment had to offer focused on alleviating his symptoms; there was no “fix” available.

“At that point, he was practically a textbook case. He would have extreme discomfort in response to even really low heat stimulus, room temperatures even,” recalls Carolyn. “He refused to ever take a bath, he was always turning his shower colder, and his feet would get really, really red. He checked all the boxes.”

One thing that really helped was soaking his feet in cold water. It gave immediate relief, and Cole began relying on it—but the Swansons soon learned why doctors advise against this treatment for people with EM.

“His feet became like little addicts, where they’d get their hit—in this case it was the cold—to feel better. But the next time, his feet needed to be in the cold water longer, and it would need to be colder, and they’d need it again sooner.”

Cole’s condition continued to worsen. His skin was breaking down from being in the water so much. “I’d find him in the middle of the night sitting in the bathtub, soaking his feet,” remembers his mom. “And we got to the point where he could not be out of the cold water for more than a couple of minutes, so he went through a stretch where he didn’t sleep at all. For four days he didn’t sleep at all. At that point our family was just in crisis, and we knew we had to take him to the hospital.”

That marked the beginning of his month-long hospital stay, when he was just shy of five years old. They tried treatment after treatment. A nerve block didn’t help; nor did ketamine and other aggressive infusions. They went through a dozen prescriptions. Opiates did nothing for his pain, “because it’s his actual nerve channels that are programmed to send a pain response when they shouldn’t,” explains Carolyn.

Instead of soaking his feet, doctors showed him how to use a type of orthopedic cold therapy machine that used membranes to protect him. This saved his skin, but he was still in the same position with the cold: he needed more and more. “It actually escalated to the point where he got frostbite on one of his toes, which isn’t good in the middle of summer,” says Carolyn. “He lost part of that toe. That’s when we knew we were headed down a really bad path, that what we were doing wasn’t sustainable. He had absolutely no quality of life. So we brought him home.”

## ***Taking control***

They had run out of options; the medical establishment said that there were no treatments left to try. So the Swansons made up their own.

The plan was to wean off of all medication, and use rigid environmental controls to get Cole functioning at near-normal temperatures. They eliminated the cold water and cold therapy machines, but turned their home’s air conditioning down to 60 degrees, elevated Cole’s feet, and pointed multiple fans at them. Every couple of weeks, they’d take a fan away or turn the thermostat one degree warmer.

It was a slow process, but it worked. Once Cole was OK with 70-degree temperatures indoors, they started letting him go outside when the temperatures were within a certain range. And they incorporated other treatments, like swimming, that helped him blow off steam and regain strength, while regulating his temperature. “It’s really a matter of trying to help him regain a normal life, while not exposing him to the high or low end of the spectrum,” says Carolyn. “We never go anywhere without a fan. Ever.”

The family recently built a custom home that incorporates several features that make Cole’s life easier.

“Sometimes [when I need to cool down] I just take a shower,” Cole, now eight, explains. “To help my feet stay cool I have foot sprayers, and I can adjust them so they get where I want them to.” The foot sprayers are on a separate temperature control from the normal shower head,” Carolyn explains, so his feet can stay cool while the rest of his body is warm. “Yeah, so my body doesn’t get freezing!” Cole interjects.

“You never really took a warm shower before we moved here,” observes his mom.

## ***Alternative therapies and the future of treatment***

Encouraged by a family member who practices acupuncture, they also began trying alternative therapies to cope with the physical and psychological side effects of what he was going through. The family tried acupressure, reiki, and eventually found unexpected benefits in energy reading.

“You come away thinking, ‘I’m not sure I believe that, but I’m not sure I don’t believe it, either,’” says Carolyn. “We were already doing environmental controls at the time we began incorporating alternative therapies, so it’s difficult to say what one thing did versus another, but he did start to get better. His sleep was less troubled, and he began putting on some of the weight he’d lost.”

In terms of America’s “opiate crisis,” Cole is in an unusual place: though opioid medications give him no relief, his disorder involves an unusual mechanism for pain. The big societal push to develop non-opioid pain medication means that a lot of research is being done into the channels they’ve discovered because of people like Cole. If researchers could discover a pain medication that helped him, it would be an entirely new class of pain meds that work in an entirely new way.

### *Just a cool kid*

Above all, Cole is just a kid. Although he can’t always play outside in the sun, he has a group of trusted friends who are happy to stay inside during recess and play Legos with him. Dealing with strangers can be difficult, however; there’s often a well-meaning busy-body happy to weigh in on why Cole shouldn’t be wearing flip flops when it’s cold outside, or how to treat the “sunburn” that must be causing his red feet and legs.

“A defining moment came when we were made to stand out in the sun and explain Cole’s condition to a police officer, because someone saw us get out of our car, assumed we were using a handicap tag illegally, and reported us,” recalls Carolyn. “It’s surprising how much other people—often well-meaning—can have a really negative impact on Cole simply by not understanding that just because the condition isn’t always visible, doesn’t mean it isn’t there. He suffered an hours-long pain episode that day, because a stranger made him wait in the sun a minute longer than he should have. And that person has no idea.”

Even though his condition does sometimes restrict what he can do, with the help of his parents, his sister Gemma, his friends, and a wonderfully supportive school, he manages to live the life of a fairly typical kid.

“The moral of the story is that we kept trying and kept trying and things did get better, which is not normal for his diagnosis,” says Carolyn. The family knows people who have chosen voluntary amputation rather than live with the pain of the condition. “Our story isn’t like that. He got better. There is hope of getting better.”







