Researchers search for defective gene in 5-in-1 million case

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When she was 8 years old, Rachel Ducusin had sore throats all the time, and a swelling on the right side of her neck.

When doctors took her tonsils out, they discovered a rare cancer lodged in a mass of nerves near the base of her skull. It was a ganglioneuroblastoma, which strikes about 500 children a year.

For Ducusin, now 32, it was the start of a harrowing three years of treatment - chemotherapy, radiation and bone marrow transplant preparations at Sloan-Kettering Hospital in New York City.

Ducusin was often sick from the treatment, missing months of school, and recalls it as a time of loneliness and fear. But she survived the grueling regimen and grew up to marry, have two sons, and work as a patient care technician in pediatrics at St. John Hospital Detroit, where she was treated as a child.

The ordeal she endured is nothing she would wish on anybody. Then, last fall, Ducusin noticed a slight swelling on the right side of her son Joey's neck. Joey, a bright fifth-grader at DeKeyser Elementary in Sterling Heights, Mich., and a defensive guard with the city youth league basketball team, seemed in good health. But the lump nagged at Ducusin; she pressed doctors to test her son.

"Joey had absolutely no symptoms. He had not been sick, not even like a cold. But his neck just kept swelling and swelling," Ducusin said. "It's

always in the back of my head that it was going to catch me again, or catch one of my kids."

Tests revealed that the menacing malignancy that afflicted Ducusin was now threatening her son - an occurrence doctors say is exceedingly rare. Joey had a ganglioneuroblastoma in the exact spot his mother's illness manifested itself some 24 years ago. And, suddenly, the doctor who had treated Rachel's cancer became the doctor treating her son's cancer.

"It's the first time that I've treated a parent and child," said Dr. Hadi Sawaf, a pediatric oncologist at St. John.

"When cancer strikes a child, it's usually on a parent's mind - Is it something genetic, and is it something I passed on to my child? And usually the answer is no. Only a small number of cancers are genetically based," said Sawaf. "The great majority of it doesn't run in the family, and that's the unusual thing about this case."

Despite what they suspect is a genetic connection, tests to determine whether Rachel and her son have either of two gene defects associated with the cancer have come up negative. That leads Sawaf to surmise that other genes are involved, and other tests are under way to see if researchers can further isolate another defective gene.

Ganglioneuroblastoma is a rare tumor, occurring in fewer than 5 out of 1 million children per year. Its exact cause is unknown. The tumor is usually found in a child's abdomen, but can occur elsewhere, as it did in the Ducusins' cases.

Compared to the difficult course that laid Ducusin low, Joey's treatment has been easier for him to undergo and for his family to face.

To look at Joey is not to see a kid beset by cancer. The operation in late

October removed a mass that was 7 by 4 centimeters long from the base of his neck. Although it was a large mass, it was confined and had not spread. After three days in the hospital, Joey was back at home in time for a very short foray out for candy on Halloween night.

Joey said he was scared about what was happening to him, but because his mother survived the same thing, he figured he would, too. "She was telling me how everything was going to be OK when this whole thing is over with," he said.

Joey returned to school after a few weeks. And he and younger brother Alex, who will turn 8 on Monday, are never too tired to wrestle on the family room floor. There was no need for chemotherapy, or radiation, or experimental bone marrow treatment. The long-term survival rate of patients with Joey's type of tumor, said Sawaf, is 90 percent.

"There's been a lot of progress in the curability of ganglioneuroblastoma," said Sawaf. "He does not need the chemotherapy, and we were able to get to it earlier and excise the entire tumor."

Mother and son shared some of the same aftershocks from their operations.

"Now my son has the same scar, the same side effects," Ducusin said. There is a numbness in his face. The pupil of his right eye is smaller than that on the left - imperceptible to strangers, but obvious to his mother and doctors, who say it occurs because of nerves that were cut during the surgery to remove the tumor.

"I can swallow better now that the thing is out," Joey said.

Ducusin has turned to the resiliency she developed during her bout with

cancer to contend with the unimaginable now. And she and her husband, Joe, have found comfort and support for their son from the same place that lifted her up once.

When Ducusin was ill, Make-A-Wish arranged for her and her family to visit Disneyland in California. They also arranged for her to go on the set with the cast of the TV show "Growing Pains." The show's pubescent lead was Kirk Cameron, the object of Ducusin's Hollywood infatuation.

"It was a nice little perk that let me forget about it all," she recalled. "Somebody took care of me. There were surprises every day."

In November, a knock on the Ducusins' door announced the arrival of "wish granters" - two volunteers with the Michigan Make-A-Wish Foundation.

It's the first time the Make-A-Wish Foundation of Michigan is helping a child whose parent also was a Make-A-Wish recipient, said Laura Varon Brown, a spokeswoman for the charity that will grant more than 365 wishes to seriously ill children in the state this year.

"If you could wish for anything in the world - ANYTHING - what would you wish for?" a wish granter asked Joey.

Although Joey really would like to meet John Cena, a headliner with World Wrestling Entertainment, he decided to wish for a trip to Disney World, "so my whole family could go. "

Ducusin is grateful for the blessings that have come to her family through their ordeal.

"They were there for me. And now to be here for him is special," ducusin said of Make-A-Wish.

A trip is tentatively scheduled for early summer.

A healthy childhood, adolescence and adulthood is on the horizon, too. Joey's prognosis, said his doctor, "is excellent."