A Gift for Nicholas

The Cure for Cooley's Anemia

BY MILES RYAN FISHER

A week after little Nicholas Jannetti was born at Philadelphia area's Bryn Mawr Hospital, his mother, Catherine, received a call from nearby St. Christopher's Hospital. At first, Catherine thought the lady on the other line was soliciting a donation.

"Look, if you don't mind," Catherine said politely, "I have a newborn baby here. I don't have time to make a donation right now, but if you call me back in a month, I'd be more than happy to donate."

"No, no, no," the lady said, "I'm not calling for a donation. I'm calling in regards your son, Nicholas."

Catherine's heart plummeted.

In the next moments, the lady confirmed her worst fear. Nicholas's screening had come back, and it was clear that his body was not producing hemoglobin, the oxygen-carrying component of red blood cells. Nicholas was diagnosed with Cooley's anemia.

Catherine and her husband, Carl, had known this was a possibility. Both are trait carriers—which is required to pass it on—and had their first son, Carl, tested one year after his birth. That this call came just a week after Nicholas's birth caught Catherine completely by surprise.

After hanging up the phone, she did what any mother would do—she went online to learn more about the disease. What she found first was a lot of frightening information. So she contacted the Cooley's Anemia Foundation (CAF), where an individual allayed her fears, explaining that the information she'd found was outdated and that CAF would immediately get her the *right* information. The individual also explained that one of the top treatment hospitals for Cooley's anemia happened to be nearby: Children's Hospital of Philadelphia (CHOP).

The Sons of Italy Foundation made the Cooley's Anemia Foundation one of its official charities in 1974. Today it is one of three Sons of Italy National Charities, which also includes the Alzheimer's Association and the Doug Flutie, Jr. Foundation for Autism.

In order to provide Nicholas with the hemoglobin that his body needed, he began receiving blood transfusions when he was just six weeks old. He received one every three weeks, each taking eight hours to complete. Without a more aggressive form of treatment, this was the only life he would know.

The blood transfusions were always a struggle for both Nicholas and his mother, who had to listen to the screams of her infant son as nurses poked him several times in search of his vein. Every night, Nicholas slept with a pump, which required a needle to be stuck in his stomach or thigh for ten hours. The pump delivered a medicine that extracted the iron from the donated blood, since although a normal amount of iron is good, the amount of iron a body gets from regular transfusions can lead to organ failure. In the morning, Nicholas's urine would be the color of rust—proof that the iron was leaving his body.

As Catherine began researching further options for her little boy, her father-in-law mentioned the condition to his neighbor, Joe Marcasciano, a member of the Order Sons and Daughters of Italy in America's (OSDIA) 'Vincin' al Mare Lodge #2601 in Margate, New Jersey. Upon learning of Nicholas's condition, Joe recognized this disease immediately, having helped with numerous Grand Lodge of New Jersey fundraisers for the Cooley's Anemia Foundation, one of OSDIA's national charities. Joe immediately reached out to his friend, Ralph Colasanti, who has lived with Cooley's anemia for more than 50 years and also sits on the board of CAF. He put the family in touch with Ralph, who helped guide and support the Jannettis in dealing with and making decisions about Nicholas's condition, including when it came to discussing further forms of treatment.

Catherine found out that there were two treatment options beyond lifelong blood transfusions for her son: a bone marrow transplant—which requires a match—or gene therapy (which is still in clinical trials and has yet to be submitted to the FDA for approval). In October 2018, Catherine received a call from a hematologist at CHOP, informing her that they had identified five potential donors. A bone marrow transplant was, however, a risky option—one that is usually not performed unless the donor, frequently a sibling, is a perfect match. Without a perfect match, the transplant may not take, which leads to Graft-versus-host disease, whereby the donated bone marrow perceives the recipient's body as foreign and begins to attack it. Although Nicholas had five potential donors, none of them was a perfect match, giving him a 95% chance of being cured. The other 5% was the chance that the transplant would result in fatality.

Nicholas receiving blood transfusions, one every three weeks for eight hours at a time.

Catherine told 3 ½-year-old Nicholas about this possibility of a cure when they first learned about it that fall. As best as she could, she described to him what it would involve, and little Nicholas agreed that he would do it. Still, that didn't convince Catherine, the 5% chance of fatality terrifying her. So she didn't mention it to Nicholas again,

as she struggled with the decision she and Carl should make. She solicited medical opinions from across the country—from Pennsylvania to California—and even the world—particularly Italy, where treatment for Cooley's anemia is more common.

As months passed, the decision grew more and more agonizing. While she wanted her son to live a life free of anemia, she couldn't bear the thought of losing him. In March 2019, Catherine and Carl were scheduled to meet with CHOP about their decision to either go forward with the transplant or hold out hope for gene therapy developments. The day before the meeting—with her husband at work; Carl at school; and Nicholas at preschool—Catherine sat at home alone, rereading all the material. That's when she collapsed into tears, sobbing uncontrollably on her couch in the family room. "I need your help, God," she pleaded. "And I need it *right now*."

Nicholas's donor, Jessica, whom he absolutely adores.

That night, she put both of her sons to bed. Nicholas blew out his candle and said, "Jesus heal me," just as he did every night. As Catherine walked down the hallway, she heard Nicholas call out to her.

"Mama," he said, "what if I died right now?"

She hurried into his room and sat beside his bed. "Oh my God, Nicholas, don't ever say that."

Still, he repeated it. "But what if I died right now?"

"Nicholas, you're not going to die," Catherine told him. "But if you did, you would go to heaven."

"Do I have to use my pump and go to the hospital in heaven?" he asked.

"No, buddy," she said. "You don't."

Nicholas looked at his mom and said, "Well then, I want to go to heaven."

Upon hearing her son say this, Catherine had to fight the tears that wanted to pour out of her. "Nicholas," she said, "Mama's not ready for you to go to heaven yet. This is why we pray to Jesus every night to heal you—so that you don't have to go to the hospital anymore and you don't have to use your pump, but you can still stay here with Mama."

"Well then, how about I go and stay at the hospital and those people heal me?" Nicholas said, referencing the treatment that he'd been told about four months earlier.

Catherine told her son that she and his dad were going to the hospital the very next day to discuss that. Then she kissed Nicholas goodnight, and the first thing she did after leaving his room was text herself everything he'd said so she wouldn't forget it.

Nicholas comforted by his parents, Catherine and Carl, as he undergoes a bone marrow transplant.

"There's no doubt in my mind that God heard me crying that day, and He spoke through Nicholas," Catherine reflected. "I told my husband that when we make our decision, I need to know that we don't have any regrets. After that night, it became very clear what Nicholas wanted. He told me, 'I want to be cured or I want to be in heaven.' By him telling me that, I knew that if we went through with the decision, I would not have any regrets because ultimately Nicholas would get what he wanted. Either he would be cured or he would be up in heaven, free of any needles and any hospital visits."

Catherine and Carl met with the hospital the following day and arranged for Nicholas's transplant. It was scheduled for July, Nicholas's birth month, meaning he would turn four years old while undergoing the

Since 1974, the Order Sons and Daughters of Italy in America's local and grand lodges have raised and donated millions of dollars to the Cooley's Anemia Foundation.

Nicholas (left) with this brother, Carl.

"The Cooley's Anemia Foundation is incredibly blessed that OSDIA has been such an incredible ally over the years. The support OSDIA brothers and sisters have given has made an enormous difference in the lives of everyone in the Cooley's anemia community. Without OSDIA, the Jannettis and all of our families would have faced even more hardships and challenges."

- Craig Butler, National Executive Director, Cooley's Anemia Foundation

procedure. By the time he entered the hospital for the transplant, he'd already had 73 blood transfusions in his young life.

After nine days of chemotherapy, which wiped out Nicholas's immune system to allow the bone marrow cells to develop, Nicholas had to spend another four weeks in the hospital. The staff monitored his levels to ensure that the transplant was taking.

But one day, on the way home from seeing him, Catherine got a call from her husband.

"Catherine, you have to come back to the hospital," he told her.

Nicholas's blood work had taken a dive, and the doctors were afraid that the transplant had failed. As Catherine drove back to the hospital, the thought entered her mind that her little boy may never be coming home. Catherine and Carl stayed with their son through the night, a night that Catherine can only describe as the worst night of her life. If the bone marrow transplant had failed, the only resort would be to immediately find another transplant.

The very next day, Nicholas's engraftment came back—he had his donor's cells. The transplant had taken.

In July 2020, Nicholas celebrated his "new" birthday, a day on which—one year after his transplant—he could officially be considered cured. Since the transplant, he has not needed a single blood transfusion.

Upon meeting Nicholas and learning about his condition at the 2018 Grand Lodge of New Jersey's State Convention, Mary Ann Ragone DeLambily—then President of the Enrico Fermi Lodge #2229 of Cherry Hill, New Jersey—took this experience back to her lodge. On June 23, 2018, the lodge hosted a blood drive through the American Red Cross to show their support for Nicholas and the Jannetti family.

"We all felt that we could play even the smallest part in helping this little boy beat this illness," MaryAnn said.

(L. to R.) Diana Cannatella (Enrico Fermi Lodge) holding Andrew Simone; Catherine Jannetti holding Nicholas; Andy Simone (Mario Lanza Lodge #2308 of Berlin, New Jersey) with Carl Jannetti; MaryAnn Ragone DeLambily (Enrico Fermi Lodge); Arlene and Jack Montesjardi (Enrico Fermi Lodge); and Frank Masso (Enrico Fermi Lodge). Not pictured but attended: Bob DeLambily (Enrico Fermi Lodge) and Carl Jannetti, father of Nicholas Jannetti.

To celebrate his "new" birthday, as well as his actual birthday, the Jannetti family rented a house at the New Jersey shore. During the week, Catherine checked her phone incessantly—hoping for a certain message. One year out from the transplant, and they were permitted to be in contact with Nicholas's donor, something that had to be mutually agreed upon by both parties. The Jannettis had signed the paperwork right away and anxiously waited for word if Nicholas's donor had done the same.

"Ultimately, it goes back to the Sons of Italy because the Sons of Italy, over the years, has been one of the biggest supporters of the Cooley's Anemia Foundation. Because of their generosity, so many things have happened as a result. Without the funding and love and support of them, Cooley's Anemia wouldn't be able to do everything that they do. One of those things is advances in medicine."

- Catherine Jannetti

On the final evening at the beach, while the family sat out on the deck and enjoyed the warm air, Catherine received the message: Nicholas's donor, Jessica, had signed the release as well.

Nicholas would get to meet the young lady who gave him the greatest gift he'd ever received.

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What is Cooley's Anemia?

Thalassemia is the name of a group of genetic blood disorders. To understand how thalassemia affects the human body, you must first understand a little about how blood is made.

Hemoglobin is the oxygen-carrying component of the red blood cells. It consists of two different proteins, an alpha and a beta. If the body doesn't produce enough of either of these two proteins, the red blood cells do not form properly and cannot carry sufficient oxygen. The result is anemia that begins in early childhood and lasts throughout life.

People whose hemoglobin does not produce enough alpha protein have alpha thalassemia. It is commonly found in Africa, the Middle East, India, Southeast Asia, southern China, and occasionally the Mediterranean region. There are four types of alpha thalassemia that range from mild to severe in their effect on the body.

(L. to R.) Carl Jannetti, Grand Lodge of New Jersey State President Nick Burzichelli, MaryAnn Ragone DeLambily, and Catherine Jannetti with Nicholas in her arms.

People whose hemoglobin does not produce enough beta protein have beta thalassemia. It is found in people of Mediterranean descent, such as Italians and Greeks, and is also found in the Arabian Peninsula, Iran, Africa, Southeast Asia and southern China. There are three types of beta thalassemia, including Cooley's anemia, that range from mild to severe in their effect on the body.

Thalassemia Major, or Cooley's anemia, is the most severe form of beta thalassemia where the complete lack of beta protein in the hemoglobin causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. These extensive, lifelong blood transfusions lead to iron-overload which must be treated with chelation therapy to prevent early death from organ failure.

"Cooley's anemia, which is a form of thalassemia, is disproportionately represented among some people of certain backgrounds, including people of Italian heritage. Although no one knows exactly why, it's thought that the trait developed in the Mediterranean and other areas as a way of protecting against malaria. In some places, it's even referred to as Mediterranean anemia. This is one reason that the Italian-American community has been so ardent in spreading information about Cooley's anemia."

- Craig Butler, National Executive Director, Cooley's Anemia Foundation

For more information, visit the Cooley's Anemia Foundation website at www.thalassemia.org