

DOWN SYNDROME

A FAMILY'S JOURNEY

Last March, Mary Anne Christo gave birth to twin sons. She and her husband, Eric Waksmundski, anticipated complications as the boys were born 10 weeks early, but were devastated when both boys were diagnosed with Down syndrome.

Shane and Wyatt had a rough start, faced multiple surgeries and spent the first few months of their lives in the hospital. The

twins have dealt with more in their first year, than most people experience in an entire lifetime.

The next few months had their ups and downs, as the Mahoning Valley couple came to grips with the fact that their lives had changed forever. Old dreams were tucked away, and new, fragile dreams have taken their place. Life is now lived one day at a time

and no one looks too far beyond tomorrow.

As the holidays neared, and the family anticipated celebrating the twins' first Christmas, life began to veer off track again. Wyatt's health began to deteriorate, and every breath was becoming more and more difficult.

For most of December and January, Wyatt remained in the hos-

pital, where doctors poked and probed, hoping to find the source of his most recent medical problems.

Then finally, in a hospital operating room, doctors discovered why the strong-willed baby couldn't breathe.

In Chapter Three read about Wyatt's latest medical journey, and learn why he's earned the nickname "Super Wy."

CHAPTER THREE

Breathing a little easier

By KAREN CIMMS and AMY MILLER
kicimms@tnonline.com

Wyatt Waksmundski has a lot. He has a twin brother, Shane, and a big brother, Jesse, 3. He has a mom and dad who love him, and a large extended family. He has nurses to help care for him and lots of high-tech equipment to help him breathe and eat.

He even has some really cool nicknames, like "Super Wy," "Fightin' Irish" and "Medical Marvel."

What he didn't have, until recently, was the left half of his diaphragm.

A rough start

The first 11 months of Wyatt's life have been eventful. Born on March 6, he and Shane were both diagnosed with Down syndrome. Almost all of the first four months of their lives were spent in the hospital, first at St. Luke's in Bethlehem where they were born, and then at Geisinger's Janet Weis Children's Hospital in Danville.

The twins faced multiple surgeries, including fundoplication, a procedure that prevents food and stomach acid from backing up into the esophagus. With the fundoplication, they cannot burp after a meal, nor can they vomit if they are ill.

To insure that they receive sufficient nourishment (via a feeding tube), and also allow the escape of air from the stomach, a feeding port (or MIC-KEY button) was surgically inserted into their stomachs.

Before the fundoplication, Shane and Wyatt suffered with severe episodes of reflux that caused serious heart and lung issues, as well as aspiration pneumonia, which could have occurred from inhaling formula when being bottle-fed.

While the twins have had ups and downs since being released from the hospital in July, including additional hospitalizations, the last couple months have been rough on Wyatt.

A real challenge

Dr. Christopher Coppola, an associate in pediatric surgery at Janet Weis Children's Hospital who has helped treat the twins over the last few months, said that they are "their own medical definition."

"They represented well-described characteristics and features of new-borns with Down syndrome," Coppola said, such as facial features and health problems, but Wyatt provided his doctors with more of a challenge.



Wyatt Waksmundski looks alert and awake shortly after going through major surgery.

"Every time I see that boy, he's bigger, chubbier, taller — just the fact that he's growing is heartwarming. You see he's got the desire to live."

"What is true in children with Down syndrome is that they throw doctors curve balls," Coppola said, adding that common problems include a hole in the heart, blockage in the intestines and malformed arteries and veins. Wyatt didn't have any of these problems.

Instead, he had something very rare. Something only about one in every 20,000 people are born with — agenesis of the hemidiaphragm.

Coppola explained that agenesis, or absence of part of the diaphragm, is something he has handled only four times in his medical career.

"Agenesis was new with Down syndrome. But it didn't surprise me," he said. Unfortunately, this medical complication, masked by a thin, clear membrane that kept Wyatt's intestines from moving completely into his chest cavity, kept the doctors wondering and searching for an answer for months, and kept Wyatt from being a healthy baby.

A difficult diagnosis

In December, Wyatt's unusual condition was beginning to make itself known, and he was soon back in the hospital.

"He was gagging a little bit on his food, and trying to vomit," explained the boys' father Eric Waksmundski. "He went through all the motions, minus the vomit."

While his wife, Mary Anne Christo, remained home with Shane and Jesse, on the evening of Dec. 17, Waksmundski and Wyatt's home health care nurse took Wyatt to a Lehigh Valley-area hospital. Although doctors were unsure what was wrong, after running some tests and tak-

ing a stomach x-ray, he was admitted. "They knew that he had a lot of gas; and his stomach was pretty extended," recalled Waksmundski. "He had a lot of bile coming back through his (feeding port), which could be an indication of a blockage, so they were concerned. The x-ray showed no blockage, so it was just a matter of relieving the gas and trying to figure out what was going on with his stomach."

"That was Friday into Saturday," said Waksmundski, "by Sunday, he was actually worse."

Another x-ray Sunday showed Wyatt had significantly more gas. He was put on a stomach suction through his MIC-KEY button, which removed the gas and gave him some relief.

"It worked," said Waksmundski. "Within a couple hours he appeared to be doing much better, once they were able to relieve some of that pressure."

While there was no real diagnosis and no apparent blockage, the hospital staff suggested Wyatt might have had some type of virus.

As Wyatt's condition started to improve, he was given Pedialyte. When he could tolerate that, formula was introduced in small amounts.

"As the week went on," said Waksmundski, "his breathing became more labored. This was not a problem that he was having before."

While on the outside, for Jesse, it appeared to be a happy Christmas; on the inside, the Waksmundskis were hurting.

"I don't think that you can help it," said Waksmundski. "You're torn, and it's not even in half. You are torn in multiple ways, because you want all your kids home. We didn't go to the hospital that day, and that was a decision we had to make because we really didn't want to take away from Jesse on that day. We knew that Wyatt was being taken care of and he was in good hands with the doctors and the nurses. It was important that we didn't compromise that day for Jesse. It was really about not taking away from him, and I think we did a good job at it, but it's hard as a parent."

Christo, who is a registered nurse, headed to the hospital, hoping to bring her son home. She was stunned when she saw him.

"When I went up there, I almost keeled over, because he had not been like that two days before," she recalled. "He looked horrible, and I said 'You need to take a chest x-ray of him before you release him.'"

She told her husband if Wyatt was released that day, she was taking him directly to Geisinger.

Although the doctor she spoke with did not believe an x-ray was necessary, he relented and ordered one. The results were not what he expected.

"He came in and said 'You need to sit down. We need to talk.' I instantly thought of cancer," said Christo. "I just couldn't even imagine."

The x-ray appeared to indicate Wyatt had a diaphragmatic hernia, and that his intestine may have moved into his chest cavity. Arrangements were made to transfer him to Janet Weis Children's Hospital the next day — Christmas Eve.

Not a merry Christmas
Early that morning, around 2 o'clock,



Hospital staff pause in the hallway with Wyatt on the way to the recovery room, so that his parents, Eric Waksmundski and Mary Anne Christo, can see him, just minutes after surgery during which doctors replaced a missing left diaphragm.

BOB FORD/TIMES NEWS PHOTOS

ing a stomach x-ray, he was admitted.

"I am having a hard time falling asleep tonight," he wrote. "This is not the Christmas I was hoping for and I really feel bad for my son Jesse and my wife. So much we were hoping to do this year, that just won't get done ... You know, it is easier believing in Santa than being Santa."

A few hours later Waksmundski was on his way to the hospital to see Wyatt before his three-hour journey to Danville. Although happy to see his Papa, Wyatt was still having trouble breathing and just wanted to be held.

On initial examination at Janet Weis, it almost seemed like the answer to all their prayers and a real Christmas miracle. There appeared to be no evidence of the diaphragmatic hernia and Wyatt's lungs seemed clear, with no sign of pneumonia.

"The first test, he looked much better and he was off oxygen," said Waksmundski, "and the doctor thought that he had probably had a bad case of pneumonia, and was doing better."

More tests were scheduled for that evening, including a CT scan to get a better look at Wyatt's lungs and diaphragm.

It was after 9 p.m. before Waksmundski left the hospital to head home and play Santa, for the sake of 3-year-old Jesse.

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nia. Surgery was scheduled for the afternoon of Jan. 27.

Minutes before "Super Wy" headed into surgery, Waksmundski captured a sweet photo of his wife and their son, his hair standing up in its regular "mohawk," staring up into his mother's teary eyes, as she stroked his head and cradled his left arm. An IV had already been started in his right arm.

Wyatt was wheeled into surgery around 2 p.m. and the couple was told it could take two to four hours, but within minutes the phone rang in the waiting room, and the family was told it could take even longer.

A shocking discovery
The surgery took a little more than two hours, and when it was over, the Waksmundskis received some shocking news.

Wyatt did not have a diaphragmatic hernia — he had no diaphragm on the left side at all. There was nothing more than a bit of membrane. The doctors were stunned.

Dr. Coppola said that once the team saw the problem, it was like "a light bulb went on" and they instantly knew what was causing Wyatt's health issues.

"The reason it was so hard to determine that Wyatt did not have a diaphragm was because he was fighting numerous conditions that were more life threatening at the same time," Coppola explained, adding that this wasn't the first time he had operated on Super Wy. "We had our hands and eyes in him before and didn't see this. He had a thin little membrane, much like Saran wrap, separating the two cavities (thoracic and abdominal). It had no muscle in it and didn't function right."

"It took us 11 months to figure this out, but we kept at it until we figured it out. When we saw it, we knew it explained why he was having to fight to breathe."

Coppola and his colleagues used a material called AlloDerm® Tissue Matrix, a material produced from donated human skin tissue, supplied by certified tissue banks. The donated skin is processed to remove all epidermis and cells, leaving behind a natural material, free of all donor cells, that the body would not reject.

"We felt that AlloDerm® was the best choice because it will allow Wyatt's own cells to regenerate and grow around the diaphragm I built for him," he said.

To build the diaphragm, which is tubular in shape, Coppola cut a half-circle out of AlloDerm®, and then sewed the tube into place between the chest and stomach.

Super Wy
Following the surgery, Coppola spoke with the Waksmundskis about their little "medical marvel."

"He said usually, you find out in utero, or early on," said Christo, "and there's usually a lot of other issues. And the fact that he wasn't on a ventilator, he was just shocked. I said to Eric, 'All those betamethasone shots I had when I was pregnant to build up their lungs — maybe it did.' It's just unbelievable. The shock that he had."

"If it's going to be Wyatt, he'll surprise you."

The family was able to see Wyatt shortly after surgery and what they saw, was a welcome sight.

He was awake and alert and his color was excellent. Thanks to an epidural, he was not in pain. He was placed in the PICU, and had a bit of a rough start the following morning, but after repositioning his chest tube, he did much better. Follow up x-rays showed vast improvement and his lung was fully expanded.

Wyatt continued to improve and eat well. On Feb. 3, he was released from the hospital.

His prognosis is promising. "Every day that he lives, I think his chances for the future get brighter," said Dr. Coppola. "But coming from birth with medical conditions, I think he's persevered and I expect him to get through childhood. I've seen such a will to live and I'm confident he's going to make it."

"Every time I see that boy, he's bigger, chubbier, taller — just the fact that he's growing is heartwarming. You see he's got the desire to live."

Coppola's "prognosis" for the Waksmundskis is promising as well. "It's an incredible chunk of work, every day to keep a sick child going," said Coppola, who noted that he too has three sons, and knows how hard it is to make sure that they are healthy and safe.

"You've got to take your hat off to what those parents have done."



Eric Waksmundski, center, and his wife, Mary Anne Christo, left, listen closely to Dr. Chris Coppola in the waiting room after Wyatt's surgery.

What is a diaphragm?

By AMY MILLER
amiller@tnonline.com

Wyatt Waksmundski has experienced many issues with his lungs since he was born.

After numerous battles with pneumonia, bronchitis and breathing in general, it was discovered that Wyatt was missing a diaphragm on his left side.

According to Dr. Christopher Coppola, an associate in pediatric surgery at Janet Weis Children's Hospital in Danville, a diaphragm is "an active muscle that moves like a dome." It sinks when a person breathes in, pulling the lungs down so they can be inflated; and pushes up on the lungs when a person breathes out.

This muscle, which divides the thoracic (chest) and abdominal cavities, also keeps the lungs separated from the stomach and intestines. In Wyatt's case, the left side of the chest cavity was only guarded by a thin membrane. This absence of a diaphragm — formally called agenesis of the hemidiaphragm — allowed his intestines to partially migrate into the chest cavity and push on his lung, causing it to not be able to fully expand.

As Wyatt continued to have breathing issues and illnesses, doctors did many tests.

Probing for the answer

Coppola explained that the medical team began testing Wyatt with less invasive tests at first, in the hope of finding the root of his problems. They did x-rays; a CT scan (a more advanced x-ray); and a barium swallow test, which is used to determine how well the muscles in the esophagus are functioning.

Coppola noted that the esophagus



ABOVE: Eric Waksmundski and his wife, Mary Anne Christo, spend time with Wyatt in the PICU at Janet Weis Children's Hospital, where he is surrounded by monitors following his surgery.

LEFT: Wyatt looks up at his mother Mary Anne Christo from his crib in the PICU at Janet Weis Children's Hospital in Danville, soon after surgery to replace his missing left diaphragm.

Follow the TIMES NEWS for future stories on the Waksmundski family and their journey with Down syndrome. You can also log on to the TIMES NEWS website at www.tnonline.com for this and all of the stories in the series, "Down syndrome: A family's journey."