NO SLOWING HER DOWN

Five-year-old Hampton Bays girl with rare genetic disorder continues to inspire

BY KYLE CAMPBELL

Julia Scourakis, 5, scoots around the Hampton Bays Elementary School gym with her classmates Friday, March 21. KYLE CAMPBELL

Julia works on an assignment in June Eaton's kindergarten class on Friday, March 21.

While the other students in June Eaton's kindergarten class walk from the gym back to their classroom in two single-file lines, Julia Scourakis kicks herself through the halls of the Hampton Bays Elementary School, seated on a miniature, three-wheeled Radio Flyer scooter.

Standing just 15 inches tall, the 5-year-old is about half the size of most of her classmates, and because of a permanent bend in her tiny legs, she cannot walk upright, leaving the scooter as her only means of getting around without being pushed or carried.

Although she has not yet received a definitive diagnosis about her daughter, Jill Scourakis of Hampton Bays said Julia most likely has a condition known as Escobar syndrome, a rare genetic disorder that disrupts prenatal development.

Very little is known about the disorder itself, also called multiple pterygium syndrome, including the number of people who are born with it, as only 50 cases have been documented in medical literature and in several countries, including England, France and Germany, according to online records. The disorder, which has proven fatal before birth or very soon after birth in the more severe cases, often leaves those suffering from it with webbing of the skin and can cause joint deformities, called contractures, that restrict the movement of limbs.

The physical manifestations of the disorder have required a steady flow of appointments with doctors, surgeons and specialists since Julia was born. The condition also means a lifetime of physical limitations, including her small stature and, potentially, the inability to walk upright.

Julia has already become painfully aware of some of the limitations she faces when compared to her classmates, but rather than sulk, the plucky girl in the pink glasses and braided pigtails has demonstrated a sense of quiet determination that has inspired her family, teachers and classmates.

“She’s made such an impact on our class,” Ms. Eaton said. “Julia represents perseverance and determination, but also kindness. She’s really had an amazing effect on all the other students.”

The Early Years

The first two years were the most difficult for Julia and her mother. The complications were evident early, Ms. Scourakis recalled, as several red flags popped up in a 20-week ultrasound that showed spinal regression and nuchal fold thickening, but was unable to determine a sex—all potential signs of trouble.

Further testing put Ms. Scourakis at ease for the final few months of her pregnancy, until she went into labor, which lasted three days.

“She wouldn’t come out, so they did a C-section,” she said. “When she came out she wasn’t breathing that well and she was sort of frozen, her neck wouldn’t turn to one side, her arms were webbed, her legs were webbed.

"They didn't tell me [anything was wrong] until after I had the C-section," she continued.

Julia was born with severe scoliosis and webbing along the inside of her arms, the back of her legs and elsewhere on her body that prevented her from being able to extend her limbs. Her muscles were underdeveloped, as was her sense of hearing.

She spent the first three months of her life in the natal intensive care unit at Stony Brook University Hospital. Ms. Scourakis described the daily trips between the hospital and her home in Southold as her "fulltime job." Because of her extended time off, Ms. Scourakis, who has been the primary guardian of Julia and her 6-year-old sister Nicole, since her divorce in 2010, eventually lost her job as a medical assistant, a position she had held for nearly a decade.

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When she was one, Julia had surgery to remove the growth plates in her backbone to prevent further curvature of her spine, Ms. Scourakis said, meaning she will likely stay roughly the same height for the rest of her life.

The first two years of Julia's life were full of visits from nurses, physical and occupational therapy appointments and cryinglots of crying, Ms. Scourakis recalled. The tears were caused by the pain in Julia's back, joints and limbs, as well as the discomfort of a feeding tube inserted directly into her stomach when she was just two months old. Even at age 2, Julia was still unable to crawl and struggled to move her arms and legs.

But, at about two-and-a-half years of age, Julia "turned the corner," her mother said, as she began eating solid foods and learning to communicate with the help of speech therapy.

"All of a sudden it was like a light bulb went off in her head, like, 'Oh, I'm supposed to eat and talk and walk and learn how to do all this stuff,'" Ms. Scourakis said. "It was like 'click.' But, until she was about two-and-a-half, she was miserable every day. I was beside myself, didn't know what to do with myself."

A Mysterious Illness

Escobar syndrome does not affect mental development and Dr. Joseph Quinn of Southampton Pediatrics, Julia's primary care physician, said the condition primarily hampers quality of life, but not usually one's lifespan.

Aside from being more prone to respiratory infections, people with Escobar syndrome are not usually more susceptible to illness than those without it. Although he is not an expert on the syndrome, nor has he had a patient with even a similar condition in the past, Dr. Quinn said Julia has handled her situation admirably.

"She's very sweet, she's got a nice disposition," Dr. Quinn said. "When she's sick, she's like other kids. She's not smiling because she doesn't feel well, but most of the time she's happy. She's amazing in her own way, the way she's handled everything."

Julia regularly sees several other doctors in New York City to monitor the condition of her spine and gastrointestinal tract, among other problem areas. Ms. Scourakis has recently been talking with a leg specialists about a surgical procedure that could help Julia walk upright.

Currently, Julia's legs have a slight bend at her knees and although she is able to waddle in a crouched position, she cannot fully extend them to walk normally. She has a yellow wheelchair that her aide, Lori Florimont, can push her in while she's in school, but Julie prefers to be as independent as possible, so she favors the scooter.

Julia and her mother were also able to obtain an electric wheelchair two years ago through their insurance, but technical difficulties have kept it lightly used, as did the lack of space within their home and car.

Being in school and around children her own age has made Julia yearn to be able to do what others can do, according to her grandfather, John Milowski of Riverhead.

“She knows she can’t do a lot like the other kids,” Mr. Milowski said. “She told her grandma that, ‘I can’t walk and I can’t run and I can’t play like the other kids can play,’ and that’s what she wants to do. She wants to do that, so she does the best she can.”

Whether it’s sitting on her dining room table, meticulously selecting bright-colored clips to put in her hair, or insisting that she turn her own work in to her classroom’s assignment bin rather than having an aide do it for her, Julia has a desire to be self-reliant as often as she can.

As much as she values her independence, however, there are daily tasks that Julia is unable to do on her own because of her size, like using the restroom, sitting at a table or going through the school lunch line. For these tasks she needs to be picked up and carried, generally by her mother at home or Ms. Florimont at school. Ms. Eaton will even carry her to the classroom’s Smartboard to answer questions during lessons.

While being able to walk correctly is a goal for both Julia and her mother, it could also prove to be a medical necessity. Ms. Scourakis explained that her youngest daughter needs to walk to avoid long-term damage to her hips, legs and feet from constantly sitting and crawling in a cross-legged position.

But surgery is not a guaranteed solution, as the webbing is known to grow back, which would leave amputation and prosthetics as the next best alternative, according to Ms. Scourakis.

In the meantime, Julia enjoys having play dates and sleepovers with her classmates, playing with dolls and board games with her older sister, tinkering with apps and taking selfies on her mother's iPhone, and drawing. But, more than anything, she loves swimming and the free range of motion it provides her.

"This girl," Ms. Scourakis said, "once you get her in the water, you can't get her out."

The Road Ahead

Julia continues to receive physical, occupational and speech therapy about three times a week in school. At home, she likes to finish all of her homework in one sitting to free up the rest of her week for more entertaining ventures.

"I like math because it's easy and I don't need any help," Julia said.

Ms. Scourakis said she's not quite sure how she and her daughter will face future challenges, particularly as Julia's classmates grow taller and become able to do more things that Julia cannot. The total number of cases of Escobar syndrome is not known but, from what Ms. Scourakis and her family can tell, they are few and far between. Therefore, there are very few support groups and, at least locally, no other parents to turn to for answers.

For now, Ms. Scourakis, a volunteer with the Hampton Bays Volunteer Ambulance Corps, is more focused on making ends meet. She's managed to be thrifty, furnishing her home with second-hand rugs and furniture—most of which look as good as new.

She also entered an essay contest and hopes to win a wheelchair-accessible van that can fit Julia's electric wheelchair, which would enable Ms. Scourakis to take her youngest daughter with her while running errands without having to carry or push her in a stroller.

Ms. Scourakis noted that the caretaker role is a natural fit for her, from her previous career, to her volunteer duties, to her home life.

"I'm so used to doing it," she said of volunteering for the ambulance company, which she's done for seven years. "You think I'd want to run away from that sort of thing, but I embrace it."

Julia has embraced her own role at school. Her classmates are drawn to her—as evident by the swarm of children huddled around her during a gym class drill on Friday afternoon—as are staff members, who clamor to talk to her during her lunch period.

But few have gotten to know Julia quite as well as Ms. Florimont, a four-year employee of the Hampton Bays School District who has worked side by side with Julia all year. For her, aiding Julia has been both unique and rewarding.

"It's been a fantastic experience," Ms. Florimont said. "She's an amazing child and she has a wonderful personality."