

# Strengthening Ashley

## A Young Family Finds Hope in IG Therapy

By Jim Trageser

Like any young couple, Jenn Sullivan and her husband were thrilled to welcome a new child into their family. Jenn had just turned 19, her husband was still shy of 21, and when baby Ashley was born in April 2000, she was perfect. Life couldn't have been any better for the young family living in upstate New York.

But as Jenn explained in a series of emails, Ashley's health took a serious turn for the worse between the ages of 20 months and 2 years.

"Up until that point, there had been some odd things we had noticed, but nothing seemed to fit any set pattern, per se.

"For example, she had 'rattly' lungs, tachycardia, purplish feet, a continual clear runny nose, terrible GERD (gastroesophageal reflux disease), a diaper rash so severe it was treated with burn therapies like SSD ointment and whirlpool therapy at the hospital. In fact, her vaginal area was completely devoid of the epithelial layer of skin—her urine or some virus had burned it off, so she would bleed and scream horribly upon soiling a diaper. It was so awful! We wept with her.

"She vomited constantly for no apparent reason, [and] she was quite late in hitting her growth milestones—walking, crawling, sitting, etc. As odd as all of this was, her doctors appeared unalarmed."

More confusing for the family was that Ashley seemed perfectly healthy when she was born—and Jenn's pregnancy had been perfectly normal.

Still, as Jenn wrote, things got even tougher for Ashley when she was 20 months.

"She was diagnosed with psoriasis—she had awful cradle cap that just never went away—and hypothyroidism, and she was put on 50 mcg of Synthroid. There were literally hundreds of tests and scans done in this timeframe



but nothing more could be determined. But there was something wrong! She would go days without eating, she would sleep 16 to 18 hours straight and she developed a very 'barky' cough. She did have a few ear infections, but nothing abnormal, and most kids do get them. She was getting her vaccinations on time, as scheduled.

"The doctors were perplexed! Something was wrong—but what?"

In December 2002, Ashley was admitted to the hospital with double pneumonia.

"We were so terrified," Jenn wrote. "She did eventually recover, but not for long. All of 2003 she fought one lung infection after another. She was completely off of the ➤

charts at this point for her height and weight, as she had stopped growing long ago. She ended up hospitalized again after what we thought was an asthma attack in November 2003—at age 2 1/2. She was sent home later that weekend and we were told she had another pneumonia—we were so sick of hearing that by now!”

Like many families facing an unknown diagnosis, the Sullivans wanted answers as to what was wrong with their little girl.

“This poor baby needed help and it seemed no one could give it to her. That week we were back at the hospital and we refused to leave! I will forever remember one of the residents there, who had remembered us from our previous admission, taking my hand and saying, ‘Something is wrong with your daughter and I promise you, we will find out what it is this time.’”

Before they did, though, Ashley had to fight for her life.

“That next morning, we almost lost her—her blood pressure was up to 180 (sleeping!) and her pulse was down to 90 on full oxygen. The doctor in the room was ready to intubate but, fortunately, Goddess bless, it did not come to that.

“Still, we knew our little girl did not have much time. The doctors had to act—fast! Again, while she was pumped full of fluids, steroids, antibiotics, etc., she had hundreds of tests, scans, samples and even a bronchoscopy-thoracoscopy done to her small little body.”

“When children do ask questions she tells them, ‘I am a SCID kid’ and walks away. She makes no apologies for being the wonderful, free spirit that she is, in her own way.”

After a month of hospitalization, doctors finally ran a test that showed Ashley had no immune system. But even that didn’t tell them why she was sick.

“Finally, we met with the chief of immunology, a wonderful doctor who said that, on a whim, he wanted to test for SCID (severe combined immune deficiency). He said he was almost certain she did not have it, as

most undiagnosed children pass away by the age of 2 and at this time she was 3 1/2! Well, sure enough, that was it! She had SCID, according to a doctor at New York University.”

SCID is caused by a genetic abnormality, and so doctors then ran a genetics background on Jenn and her husband to try to pinpoint Ashley’s condition.

“As it turns out, Ashley has one of the most rare mutations of SCID—ADA, virtually unheard of,” Jenn wrote.

The adenosine deaminase (ADA) subtype of SCID, specifically, is caused by lack of the adenosine deaminase enzyme, which leads to a compromised lymphatic system. SCID affects about one in 100,000 babies, and ADA, only a tiny sliver of those.

Still, as any family who has been down this road knows, any answer is better than uncertainty.

“As frightening as it was, at least we had a name and some treatment options. Since SCID-ADA kids do not generally do well under chemo for bone marrow transplants, Ashley was put on IVIG and an IM injection known as PEG-ADA, or Adagen, two times a week. Under this, she began to improve.”

Not that Ashley’s day-to-day life can be considered normal. Three years ago, doctors found she has pulmonary fibrosis. She also has adrenal insufficiency and hypoglycemia, and she had a mediport put in. In 2005, she was diagnosed with autoimmune hemolytic anemia and delta agranulocytosis (a blood clot disorder). In the summer of 2006, doctors took a bone marrow sample, and it came back fine, giving the family some good news. But in September 2007, she developed keratitis in her right eye, causing serious degradation of her vision.

Still, Jenn says the family refuses to allow Ashley to be defined by her medical challenges. As Jenn put it in an email, “She has SCID-ADA; SCID-ADA does not have her!”

As much as possible, the family is mainstreaming Ashley.

“She attends a regular public school with the assistance of a personal aide, a vision therapist, an occupational therapist, a speech and language therapist and a modified phys-ed program. She loves to play Barbie and school and with her baby dolls. She loves visits to her family members’ homes, she loves dogs, she loves to color, she loves Nickelodeon. She is every other kid, just a little extra special.”

Jenn wrote that she and her family know they’re lucky Ashley was born today, when there are options for treating SCID.

“SCID-ADA and people with SCID-ADA variants (like Ashley has) are less likely to receive the bone marrow transplants that have helped ‘cure’ so many patients, as

their bodies do not handle the required post transplant chemotherapy as well as other primary immune deficiency patients. Therefore, they, like Ashley, must rely on IVIG and medications like PEG-ADA (Adagen)."

In the past, a diagnosis of SCID was a death sentence. Today, though, with treatments such as immune globulin, patients are living longer than ever. And Jenn wrote that Ashley's approach to her challenges is positive and upbeat.

"Because of all she has been through, Ashley is a very empathetic, thoughtful child. She is happy and talkative and very easy and lovable to be near.

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"Since she did have so many random illnesses prior to her official diagnosis, she never got to experience playgroups, playdates, daycare or any of the other activities young children enjoy in their baby-toddler years. She was constantly with doctors, nurses, extended family (all adults), and that kind of shaped her into a 'mini-adult' with a pretty extensive vocabulary.

"She is very good about communicating how she feels, whether it is a physical pain, fear, happiness, nervousness, etc. She is an old pro (unfortunately) when it comes to accessing her mediport and starting IV lines, so she doesn't get too nervous about that—mostly just new doctors or clinics that she is unfamiliar with. And even that tension eases away quickly! The Women and Children's Hospital of Buffalo is great!

"As far as being in more kid settings now, like school, she has just learned to accept her limits. She knows she has to wear her glasses and medic ID tag, she cannot play in any way that could affect her bleeding disorder (playgrounds, etc.), she needs to eat well so her blood sugar doesn't drop, she has to use sanitizers religiously, etc.

"To her, it's just life and she copes with it wonderfully.

When children do ask questions she tells them, 'I am a SCID kid' and walks away. She makes no apologies for being the wonderful, free spirit that she is, in her own way."

For other families facing a diagnosis of SCID—or any life-altering condition—Jenn wrote that parents need to be willing to lean on others, no matter how difficult it can be to swallow your pride and admit you need help.

"There is no 'Martyr Medal.' Accept help! This is an overwhelming, frightening time, and you need people to rally around you. Do not be afraid to ask for, or accept, any and all forms of help offered.

"For help with monetary concerns, such as meal vouchers, parking passes and toiletries for you, ask your RN or a social worker—they are usually great resources! When people do come to visit, do not feel as though you have to keep up appearances. Cry if you need to, vent, laugh, whatever you feel! There is no right or wrong emotion. Each person is different.

"With this in mind, be sure to keep your fears, feelings and emotions separate from your ill child. Focus on activities to do together, like card games, play dough, watching movies, etc.

"Inevitably, the tough questions do arise: What's wrong with me? Why am I sick? I always told Ashley that we don't always know the answers, but, if we enjoy each day and do our best to get through the rough times, the Goddess would see us through. Be honest with your child, but optimistic!"

In addition to tending to your family's—and your own—emotional and spiritual strength, Jenn also advised that staying organized is a huge help.

"Keep a notebook of all medications your child is given, which doctors are involved in their care, and brief notes from each consult and procedure. This makes communication easier and more effective.

"Keep an overnight bag handy—a change of clothes for you, games, books, puzzles, small toys for your child—in case of emergency trips to the hospital. ... Invest in a small, portable DVD player for ER admissions."

And for families facing a primary immune deficiency, Jenn advised staying informed, but letting the medical professionals be the experts.

"Do not over-research—each variant of primary immune deficiency is different. Ask your doctor or RN for reliable books, publications and websites. There is a great web support group through Duke University for patients and families ([www.scid.net/facilities.htm](http://www.scid.net/facilities.htm))."

Her final bit of advice, though, may be the most critical: "Most important, never be afraid to ask, ask, ask questions." ■