Complex Child E-Magazine

www.ComplexChild.com

Joshua's Journey with Shwachman-Diamond Syndrome

by Tania Nelson

Shwachman-Diamond Syndrome (SDS) is a rare bone marrow failure syndrome that mainly involves the pancreas, bone marrow and skeleton, but other organs may also be affected. It is a genetic disorder that affects one in 75,000 people. Primary to SDS are hematological abnormalities, usually a decreased number of at least one type of blood cells, most commonly the white blood cells called neutrophils. Many SDS children have more than one blood cell line affected, while some have all three blood cell lines affected: the white blood cells, red blood cells, and platelets. Neutropenia, or a low neutrophil count, is the most common hematological abnormality found in SDS.

The signs and symptoms of SDS include:

- Diarrhea that is greasy and foul smelling due to pancreatic insufficiency. This usually improves once pancreatic enzymes are started.
- Abnormal bone marrow
- Hematological issues including a decrease of any or all types of blood cells:
    - neutropenia (low white blood cells)
    - thrombocytopenia (low platelets)
    - anemia (low hemoglobin)
- Failure to thrive and growth problems leading to short stature
- Frequent infections
- Dental issues
- Puberty delays
- Lung disease
- Cardiac lesions
- Liver abnormalities
- Bone lesions
- Behavior problems
- Eating difficulties

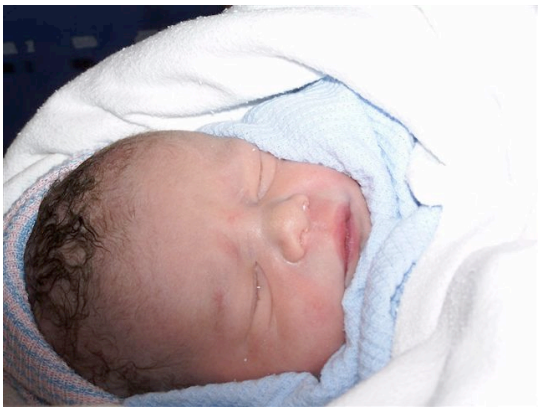
Diagnosis is determined by blood counts, pancreatic testing (which can be fecal fat tests and/or a pancreatic stimulation test done by a GI), the SBDS gene test, bone marrow biopsy, and lots of other lab tests to rule out other causes of the child's symptoms. A GI doctor may also order tests measuring pancreatic function and absorption of food, including fecal elatase, serum trypsinogen and serum isoamylase tests. Gene testing may show a patient to have gene mutations consistent with SDS, but not all patients with SDS have known mutations. Therefore, a negative gene test does not rule out SDS.

SDS is still a fairly new diagnosis and there are only a few known mutations. Patients who clinically fit the SDS criteria for diagnosis but do not have the known mutations are diagnosed as SDS-like. This is the diagnosis of my four-year-old son, Joshua.

Whether a child is diagnosed as SDS or SDS-like, bone marrow biopsies must be done at least annually to watch the marrow since any of the SDS or SDS-like children have a 30 percent chance of developing Leukemia. Bone marrow biopsies indicate how well the body is producing blood. Along with the bone marrow biopsies, blood draws are done on a routine basis to check how the bone marrow is producing white blood cells, red blood cells, and platelets.

Joshua's Story: Early Signs

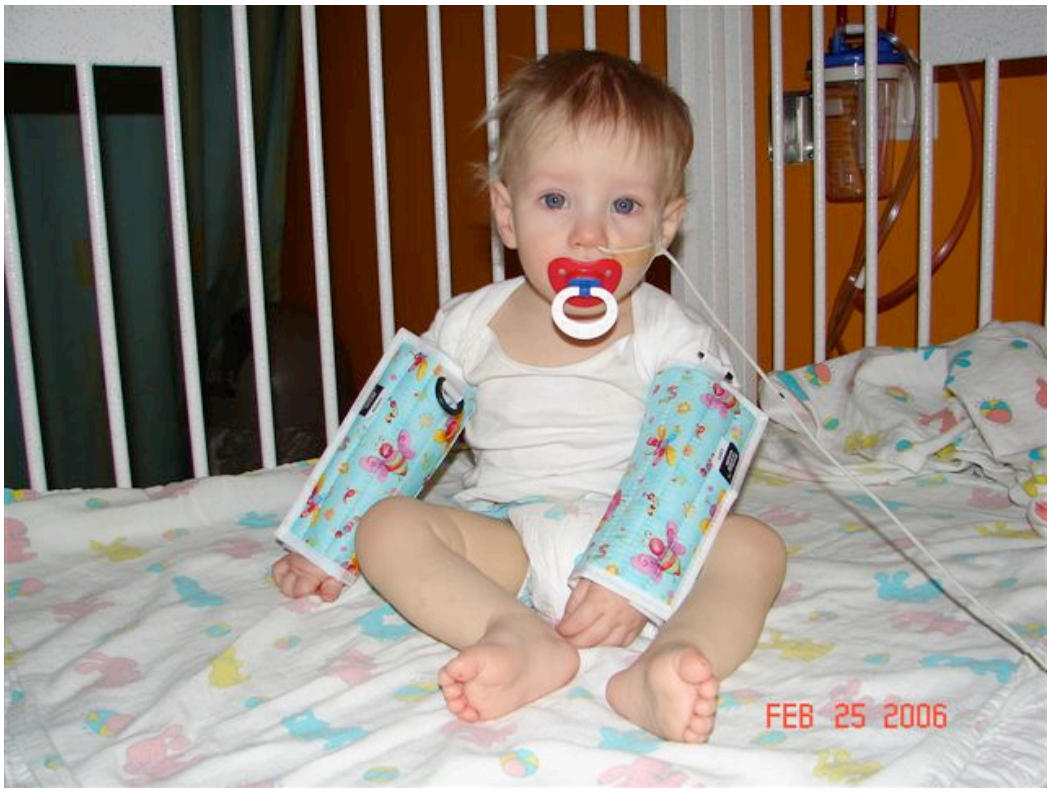
Joshua Scott Nelson was born on March 28, 2005, by C-Section. He spent several days in the special care nursery until he was four days old, at which time he was airlifted to a higher level NICU three hours away. At 18 days old, Joshua was discharged with a special blanket to address his jaundice. When Joshua was six weeks old, he was seen for possible reflux. He was started on reflux meds to try to get it under control. He would only be able to nurse a little bit at a time or else he would throw up, so daily and nightly routines were based on when Joshua would have to nurse.



Joshua's first main symptom was weight loss and failure to thrive. Joshua was 18 pounds at eight months old and 17 pounds at ten months old. His Pediatrician decided to run a sweat chloride test, celiac labs, a renal ultrasound, a urinalysis and have an upper GI X-ray scan done. At almost 11 months old, he dropped to 16 pounds, and that is when he was referred to a Pediatric GI (gastroenterologist) and had a nasogastric feeding tube (NG tube) put in to supplement his feeds.

Joshua had a gastric emptying scan (GES) done at one year old that verified what we were concerned about. Joshua did in fact have delayed gastric emptying (DGE), which is where food doesn't process through his stomach as quickly as it should. It can mean increased vomiting, as foods or formula sits in the stomach for too long and becomes too much to keep down.

Joshua was admitted back into the local children's hospital the day after his first birthday. He weighed 15 pounds 12 ounces and was a sick little boy. We actually had to beg the doctor to admit us that day, because we were really concerned about his health. His reflux meds were increased, but I still knew that there was more to it than that. The problem was that Joshua did not follow the textbook and his doctors were stumped by his continuing weight loss. When Joshua was discharged, there still was no answer as to what was going on with our little boy.



Searching for Answers

When we were discharged home, I called our pediatrician and told her my concerns and asked for a referral to a larger regional hospital, Seattle Children‘s Hospital. She called Children’s and talked to one of the GI doctors who agreed to see him. After seeing the Seattle GI, we were admitted again. For ten days, Joshua went through test after test. Nothing obvious showed up except reflux and the delayed gastric emptying. Joshua was discharged with the NG tube still in, in hopes that the tube feeds would help him grow. Later that month, the GI scheduled Joshua to have a feeding tube surgically placed in his stomach (G-tube) instead of the temporary NG tube.

Shortly after the G-tube was placed, we had Joshua back to eating some food orally! We were tube feeding him for 12 hours a day plus letting him eat anything he wanted by mouth. He was finally starting to look healthier--small, but healthy. Nonetheless, throughout the summer of 2006, Joshua’s weight was still a roller coaster. It was very

frustrating because nobody knew what was wrong. We had doctors communicating across the state and they were all stumped!

When Joshua was about 18 months old, we started pancreatic enzymes on a trial basis just to see if he would respond to them, which he did. Our pediatrician was still thinking that maybe Joshua had a case of atypical Cystic Fibrosis (CF) and was in communication with a CF doctor. Joshua did gain weight while on enzymes. The month that we didn't give him enzymes, he developed an intestinal blockage and lost weight. That right there was a determining factor to his doctors that the enzymes were helping.

Once we got the enzyme dosage right for Joshua and his weight started maintaining and actually gaining, we started to have problems with him always feeling hungry. We have talked to every doctor that we can think of about the issue of him always being hungry, but still battle that issue daily. Joshua doesn't understand that if he eats too much he will become sick. More times than not, he cries and cries because he thinks that he is still hungry, when in reality he has eaten as much as, if not more than, an adult would eat.

Finally Heading Toward a Diagnosis

When Joshua was two, we saw a Pulmonary/CF doctor at Seattle Children's and talked in great lengths about Joshua's medical history. He said that he didn't think it was CF, especially since we had a negative gene test done for CF, which tests for all known CF mutations. He did mention Shwachman-Diamond Syndrome (SDS) as a possibility. He said that it was a long shot. Joshua's blood work looked okay except for anemia, and his

pancreatic tests were okay even though clinically he showed improvement with pancreatic enzyme supplementation.
Of course, after getting home from Seattle, I got on the Internet and started researching Shwachman-Diamond Syndrome (SDS). Thanks to finding Shwachman Diamond America, I quickly learned that Dr. Harris at Cincinnati Children's Hospital was the place to go for a workup on Shwachman-Diamond Syndrome.

It took me awhile to get up the courage to e-mail Dr. Harris to see what he thought, because I didn't want to take the time and spend the money to travel across the country if it wasn't going to point us in the right direction. Finally, I sent him an e-mail, and he responded saying that he would see Joshua if we wanted to make the trip.

We traveled to Cincinnati Children's Hospital, which proved to be a very busy week. In addition to Dr. Harris, we saw a Pulmonologist, Gastroenterologist, Endocrinologist, and had an anesthesia consult. They drew more blood than anyone could imagine and ran numerous tests, including another sweat chloride test, since Joshua's symptoms are so similar to CF.

After leaving Cincinnati, it was a waiting game to get all of the results of Joshua's tests. After a few weeks, I got a phone call from Dr. Harris with all of the results. He said that Joshua's SBDS genetic test was negative, but the results from his bone marrow biopsy indicate that he should be classified as SDS-like. They will follow his DNA closely and he will need annual bone marrow biopsies.

The "like" part of the diagnosis is the hard part, because some doctors don't take that as a diagnosis. I have been told that he will be followed just like any SDS kid would be followed, but I have also been told by other doctors that it isn't a diagnosis and we still don't have answers. It is frustrating, because Joshua simply isn't a textbook case.

What SDS-Like Means for Us

His last bone marrow biopsy showed some red flags. This means more frequent blood draws and a bone marrow biopsy done at six months instead of waiting the usual 12 months.

Since being in Cincinnati, Joshua has gained more weight and is actually back on the growth chart for both weight and height. We are still trying to get his GERD and Asthma under control, and trying to figure out what we can do for him, so that he doesn't think that he has to be eating constantly.

There are the nights that I stay awake and wonder what we can do to figure out exactly what is wrong with our little boy. Where can we take him? Who could we see? What test or tests can be run? But, at the end of the day, we know that he has some type of bone marrow failure issue. His abnormal marrow and his response to enzymes, along with his many other medical issues, point us right to SDS.

It is hard to look at my little boy, knowing that even though he looks so good now that he does have a life-threatening medical condition. The comments that people make about him not looking sick make us feel good, because we work so hard to keep him looking healthy. However, they don't understand that what they can't see that is going on inside of his body is the real problem.

It is difficult on Joshua as well. The anxiety that Joshua has with the hospital is horrible! He loves his doctors, and he has a good team of them, both locally at home and at Seattle Children's Hospital. However, seeing his fear increase with each trip to the hospital is difficult.

For me, having a child with SDS is a roller coaster. To have a child who is at a higher risk than healthy kids can be exhausting. As parents, my husband and I have to find the right balance between doing what is best for Joshua and letting him be a normal four-year-old boy who lives on a farm.