

# S.D.A. ♥ eNewsletter

Shwachman-Diamond America

Issue 18

Sept/Oct 2010

## Special points of interest:

- Briefly highlight your point of interest here.
- Briefly highlight your point of interest here.
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## Alex Turnquist Memorial Research Grants Available

Shwachman-Diamond America awards Alex Turnquist Memorial grants up to \$10,000 USD. Grant proposals are accepted throughout the year. We do not have a grant request form.

SDA requires that the grant proposal be in writing and include the following:

1. Name of applicant, principle investigator, project title and summary of proposed investigation (include specific aims, significance and background preliminary studies..)
2. A detailed description of your hypothesis/hypotheses and proposed methodologies.
3. Relevance of the research to Shwachman-Diamond Syndrome.
4. Biographical information on the principle investigator and co-principle investigator, if relevant.
5. A detailed budget sheet.
6. Statement of facilities available.
7. A starting date for the project.

For more information on the general conditions for awarding the Alex Turnquist Memorial Research Grants, please visit our website:  
[www.shwachmandiamondamerica.org](http://www.shwachmandiamondamerica.org)



## Kids going through Transplant



Jasmine had her bone marrow transplant in December 2009. This past month, she has been in ICU with a lung infection. Please keep Jasmine in your prayers.



Jasmine in May 2010

Summer 2010



## A Matter of Perspective

I keep waiting for life to slow down, yet it never does. We are lucky to find our slice of normal in everyday life amid the chaos, however. I feel blessed to see my children participating in sports we never thought they would be able to play. Sometimes, as I watch my children participate in sports and watch as other parents seemingly take their own children's ability to play for granted, I want to scream, "Don't you know how blessed you are!?" Finding a balance can be difficult. Knowing what and when to share can be tough. It can feel like walking on a tightrope five hundred feet above a crowd without a net to catch you, should you fall. There are times when I have not been able to keep my mouth shut and others where I was rendered speechless. It really is a matter of perspective.

We had just returned from a week long trip to Cincinnati Children's Hospital where my two youngest had several appointments, testing and their annual bone marrow biopsies. During this trip, their secondary diagnosis of Mitochondrial disease was made firm while visiting the Mito specialists there and my mind was all abuzz with thoughts of treatments and what this diagnosis meant for their futures. As we settled back into our routine of home schooling, piano, baseball and football practices, we also had to find time to make it to follow-up appointments and meetings.

Early one Saturday, I brought Sean to baseball practice. I watched in amazement. The assistant coach was yelling at the boys, especially his own son. He told his son repeatedly, "You're terrible!" and,

"That was terrible!" along with a myriad of other demeaning statements. I remained quiet. He then yelled at my son after he missed fielding a ball, "That is terrible! As punishment, run out and get the ball now!" "Wow," I thought to myself. I still stayed silent. He told another boy in a condescending tone, "You can't catch!" Again, I remained silent. Practice went on for another hour before the boys moved to catching fly balls. That's when things got out of control. His son kept missing fly balls and the coach lost his temper. He screamed, "Jesus! G-d!" and continued to belittle his son in front of the other players. When his son did catch a ball, he said something like, "See where you are catching the ball? That is why so many of the balls roll out of your glove!" I thought, "Wow! That is over the top, his kid caught the ball and he is still not pleased!" The last straw for me was when his son missed yet another fly ball. The coach threw the bat down and yelled the Lord's name in vain. At that moment, I knew I was going to say something in hopes that he might gain a bit of perspective. He dismissed all the players except for his son, at whom more insults were hurled as he was also told, "You will stay out here until you catch three fly balls in a row!" That is when I told Sean it was time to leave and said to the assistant, "In the scheme of things, missing a few fly balls really isn't a big deal. We spent last week at the hospital with our two youngest having various testing and their biopsies. We are just glad that Sean is able to be out here and play baseball." The coach stared at me and turned around. He said nothing. His friend asked what was wrong

## by Pattie Curran

with the boys and I tried to keep it simple as I walked with Sean to the truck, "They have a genetic disease and we have to keep an eye on their bone marrow to be sure they are not developing leukemia." Before getting into the truck I said, "Thank you for volunteering to practice with the boys."

I felt a small twinge of guilt as I drove away. Well, I only felt it for a second. Sometimes, I feel guilty for telling people about our lives and what we deal with on a daily basis. Other times, I think that people might benefit from a little perspective. Not that I am the authority on perspective, but I do think we parents of chronically ill children can help others reflect on the blessings we all have been given in life. Again, it really is all a matter of perspective. Our SDS family recently lost another young adult in this past month, David Smyth. David developed leukemia and did not have a donor in the bone marrow donor registries. Jasmine, another SDS friend, has been battling for her life in the ICU over the last several weeks. Having lost so many friends to Shwachman-Diamond Syndrome over the years (this year we lost a few friends to Mitochondrial disease, as well), we realize that some things in life really are not important enough to become angry over.

Over the years, we've had what I call, "sports challenges". Sometimes, these sports challenges are the final straw on the stress scale. I've been yelled at by a baseball coach in front of players and parents for having the audacity to ask him (privately) to discuss good sportsmanship with the assistants and players.

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## A Matter of Perspective

They were all being nasty to one another and as the season progressed, it became worse. The boys would hurl insults when their teammates made mistakes and coaches were yelling for what seemed an eternity over every mistake. I was rendered speechless as he stood there yelling at me. It was crazy. Other times, I have found courage to speak up. It does take courage and sometimes I am so worn out from our day to day medical adventures, I just sit and say nothing. I remember a mom who yelled at another mom for borrowing a band aid for an injured player. Then there was the lady who yelled at me because the team had run out of orange socks. And we cannot forget the parents who become angry when a team loses. It really makes you want to say, "Get some perspective, people!" Obviously, this matter of perspective carries over to other facets of life. The person screaming at the checkout clerk in the grocery, the person at the coffee shop raging over coffee that is not precisely the "proper" temperature, and the person at the fast food place going ballistic because he has to wait a few minutes longer than he thinks he should.

We find ourselves in an alternate universe, those of us with sick kids. We've seen and met dying children. We've visited these dying children and their families at home and in the hospitals. We know far too many children who have passed away and we know all too well how fragile life is. We know that life can change in an instant. We know all too well the saying, "There but for the grace of God go I." We could be the next family sitting in ICU because of some strange infection or the next family battling the life-threatening after effects of

bone marrow transplant. Our family has been close to transplant before. It is a scary place to be, especially knowing what we know.

We fight for our children and pray for the others who are battling life-threatening complications of this disease. The joy we have when our children can play sports with the medical challenges they face is greater than the joy we feel when our healthy son plays. Both bring joy, but the joy is greater for the one who faces challenges getting to the field for competition because of what we know and have experienced. We realize how blessed we are that our boys have been able to overcome the obstacles they face, because we know too many children who cannot play sports at all. Joseph recently played his first real game of tackle football. We were so proud of him because he did not allow the medical challenges he faces each day to stop him from playing. The challenges are many, but together, we have come up with strategies to help him overcome the problems and tackle the game of football. These are the moments I cherish -the triumph over Shwachman-Diamond Syndrome and Mitochondrial disease. My boys win the competition before they ever step onto the playing field. They continue to win something far greater than most of us will ever know – the victory over two disabling diseases and not allowing those diseases to define their lives.

There are days when I am actually thankful to have Shwachman-Diamond and Mitochondrial disease in my vocabulary. I think God has allowed me to count my blessings and given me the grace to gain perspective from our ex-

## con't from pg 3

periences. While I do not like the diseases and I pray daily for a cure to be found, there really are blessings to be had. The joys are far greater because we have gained perspective and know how important it is not to take the joys in life for granted. I wish that all parents could feel the joy we feel -- the joy of watching your children accepting the disease and not allowing it to define who they are or what they can do in life. I believe that parents of chronically ill children can bring a message to the world. I believe these children can bring a message to the world. Sometimes, we just have to live it, say it and walk away. Do not be afraid to share your perspective with the world, for we live in a world where far too many folks could use a little!

***"Do not be  
afraid to share  
your  
perspective  
with the world"***





## SDS Safari? Of course!

by Christel Timmermans

For years we've been dreaming of a Safari in Africa. With an SDS son this needs a bit more planning than it would normally take, but as you can read it is very much possible and dreams can come true.

We started three years ago with a vacation in Tunisia (a 3 hrs flight so we would be able to be back home in the Netherlands quickly if it would not go well). Jaimz also has rheumatism (during that year he spent 6 months in a wheelchair). After a few pain-free months he could not walk at all when we left home, but the warm water and warm dry climate did wonders. After a short fever during our vacation his knees were painless again. No SDS problems and no food problems – so this first step did go very well!

The following year a bit further away, to the Sinai desert in Egypt, Red Sea coast (Dahab). Again perfect! His DTP jab (diphtheria, tetanus, polio) was all the protection he would need. We had a fantastic time, snorkeled a lot (beautiful coral and loads of colored fishes) and even went to Jordan to visit the Petra ruins. Two more countries conquered by SDS! Last year Jaimz' rheumatism was causing hip problems and he could not walk very well. So we decided to give Egypt another try. This would also give us another year to see how Jaimz' immune system would be holding up. In spite of his rheumatism (Jaimz' father carried him when needed) we were even able to go deep into the desert and climb canyons. And again no SDS related problems.

And after 3 or 4 years of perfect BMB's that told us his bone marrow was showing no abnormalities at all, and besides his rheumatism nothing more serious than a few nose colds, and lots of mental preparations

(from 'you will need to wash your hands very often because of strange bacteria' to 'be sure to listen to our guide when he is telling you what or what not to do when we're around wild animals' and lots of reading and watching documentaries) we asked Jaimz' pediatrician/hematologist/immunologist if Jaimz could go to Kenya and if she agreed on the necessary vaccinations against Hepatitis A/B & Yellow Fever. 'No problem' she said.

Great! So we booked our vacation and went to the 'Area Health Authority' (travel doctor) for our vaccinations. We had to fill in a questionnaire of our health history and of course all the alarm bells went off when she read about SDS (not that she had heard of it, so I had to explain). Jaimz was NOT allowed to get a Yellow Fever vaccination at all, she said, because this would be a weakened live virus and it would most likely give him Yellow Fever (a pretty serious disease, for which only the symptoms can be treated). Of course we protested. Jaimz' hem had said it was ok, besides his immune system had been working well for years and he had reacted well to his MMR jabs (also weakened live viruses). (Note: at the time no one ever told us that these MMR jabs could cause problems for Jaimz/SDS patients!!!) She would not listen to reason and simply refused to give the vaccination. She said she would contact Jaimz' hem first to discuss it with her. So we had to make another appointment. Jaimz' hem phoned us a few days later to tell us to go to another country instead, to avoid any possible risks. Not the reaction we anticipated at all and very different from what she had said before!

After a few more phone calls with her and the travel doctor

things got a bit more nuanced. The travel doctor told us that if we would stay in the South and Coastal areas of Kenya, we could take the risk of not having Jaimz vaccinated against Yellow Fever, because that was also advised to people over 60 years old, for whom a Yellow Fever vaccination was also more risky, and there the chance of actually getting Yellow Fever would be smaller, than the risk of complications due to a reaction to the vaccine.

Jaimz' hem on the other hand agreed with us that the risk of complications for Jaimz (after she had given it a bit more thought) considering Jaimz' health and good reaction to the MMR jabs, would probably be minor (keyword 'probably' so we were not reassured very much). At the same time there were elections in Kenya. The last time (I believe that was in 2007) this resulted in an internal war with lots of death and destruction. If these elections would have the same result this time, we would have to cancel our trip to Kenya. We asked for the last possible date that Jaimz should get the vaccination and agreed that we would postpone our decision until this date. This would give us time to look for answers on the vaccination risk worldwide & see what would happen in Kenya.

I contacted SDS Netherlands and Shwachman-Diamond America to ask for help. Because we were looking for someone who would be able to tell us more about the risks of this vaccination for someone who has a 'less serious' variation of SDS. An American mom with a daughter who has SDS and plans to travel to India gave us the phone number of her ped which I forwarded to Jaimz' hem.

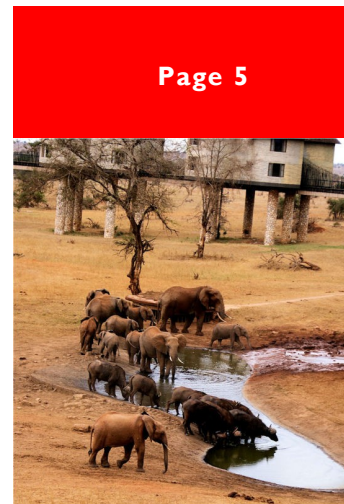
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## SDS Safari con't

Jaimz' hem also posted this question on the international PAGID list (a list of immunologists worldwide). SDS Netherlands could not give us any info (not so many patients, so not so much experience). The PAGID list came with 2 positive reactions from a Spanish and an American Dr. (both with 'less serious' SDS patients) who both said: the risk for Jaimz is minimal so he should be vaccinated. We discussed with Jaimz' hem that we thought that a minimal risk to complications with a weakened virus while still being in the Netherlands would be preferable to a small risk at getting Yellow Fever because of a mosquito bite with a real live virus while being in Kenya (incubation period is 3-6 days). It was a difficult decision for us. However minimal the risk, getting Yellow Fever is a serious problem. But we also did not want to cancel this trip because this would not be good for Jaimz as well. His SDS and rheumatism already influence his life, and we did not want to add a canceled trip because of his SDS to this, because that realization would also harm him. We believe the future holds no guaranties for no one so that we should live life at its fullest and enjoy every day, but even this belief does not make it easier to 'gamble' with your son's health. Meanwhile the situation in Kenya was stable in most parts of the country and Jaimz got vaccinated August 13<sup>th</sup> but you can imagine that the following 10 days were a bit scary while watching him for any reactions, that luckily never came. We were so relieved! Not only for this year's trip, but this also means that we can go anywhere we want now even with SDS (the vaccination will give protection for 10 years, but future vaccinations should be

ok as well). We left for Kenya August 24<sup>th</sup> and it was fantastic! We had booked a resort right at the beach that was supposed to be not only beautiful but also clean and with good quality food, thinking this would give us a healthy base to stay and from where to travel. And it was simply perfect. Small houses (clean & lovely) in a beautiful large garden, filled with beautiful trees and plants and flowers, great food, very friendly people and lots of cats and monkeys. From here we traveled and discovered this beautiful country a bit and found out that it is a great country to visit with a kid. Very relaxed atmosphere and people! The Website of the Dutch Ministry of Foreign Affairs had warned us to be careful and take certain precautions, but we never felt any danger at all. Far from it! We went on a trip with a glass bottom boat and snorkeled from it. Another day we sailed to Wasini Island (at the border with Tanzania) to a Marina Sanctuary to snorkel, unfortunately it was storming at the Indian Ocean, so Jaimz & his dad got sea sick and we did not see whales or dolphins but did see beautiful coral and fish and around noon the weather cleared and we enjoyed a fantastic Swahili Seafood lunch at the island. We've visited a Snake Park and have petted lots of reptiles, visited a Monkey sanctuary and had 3 trees planted in our name as a remaining memory. We've visited a Masai village and saw them dance. We've walked to small villages and talked to lots of people and of course relaxed at the beach and in the pool. And... we've been on a 3 day Safari in Tsavo East (the largest park in Kenya) and Tsavo West. In Tsavo East we spent the night in a fantastic safari tent (with real beds and

shower and toilet) and from the porch we could see elephants & gazelles. We fell asleep listening to the trumpeting elephants. The next morning we even saw big cat-like footprints close to the tent. The second night we spent in a small round house built on a high pole, in the middle of the bush & with a fantastic view on a wide landscape full of zebras, antelopes, elephants, gazelles, buffalos, warthogs, baboon, etc. There was a waterhole where we could watch the elephants drink at 'touching distance'. During our stay at these camps we've made a few game drives and saw lots of animals and even 4x lions (a total of 10). It was so much more beautiful than we could have hoped for; the animals very close, the most beautiful locations & landscapes and Jaimz absolutely loved it. And where ever we went, the food was delicious and we never got sick. In fact, we even never saw a mosquito or Tsetse fly so all the Yellow Fever & malaria worries before we left turned out to be unnecessary (afterwards). All in all it was a fantastic experience.. And again we've experienced that even with SDS you can do lots of things. Prepare well and take lots of medication with you, but then nothing can hold you back! I wonder where we'll go next year?



## **Shwachman-Diamond America's Mission:**

Fund and promote research in all aspects of Shwachman-Diamond Syndrome.

Disseminate current medical literature to families and physicians.

Facilitate the development of a medical management plan.

Promote parent education through a family support network.

Help Fund the Biennial International Congress on Shwachman-Diamond Syndrome

## **A Good Day**

**by Honey Denson**

It was one of those days, at least to me. The children, unfortunately, have all inherited seasonal allergies from their parents and were half sick, so I had decided to keep them home from school. It was a half day anyway. Why in the world would I want to load the miserable crew into the van just to go to school for two or three hours, especially considering that Luke and Joshua's schools are about 25 minutes apart from one another, and there was some mysterious fever going around Luke's school. We've been lucky so far as far as sickness goes, but I did not want to risk it. Anyway, they were fairly grumpy, whiny, and I swear they have mastered the skill of synchronized pooping. I mean I've heard of women living together syncing their cycles, but this has gotten ridiculous. To top it all off, I ran into the kitchen responding to the sound of Josh's cries to find that Lucy had put a laundry basket over him and was sitting on top of it. She looked very proud of herself.

Needless to say by nap time I was over it. I put the younger two to bed and got to work on some much needed cleaning. I have neglected the walls of late and finally decided that it was almost embarrassing the amount of dried chocolate milk droplets and evidence of Lucy's "artwork" that had collected on them over the past few months. I was down on my hands and knees scrubbing when Luke came in and said "Kiss my cheek Mommy!" Of course I can never deny such a sweet request. Then he said "Its been a good day!" What!? I thought. With all the weeping, wailing, and gnashing of teeth? The dirty house and runny noses? Its been a good day? Really? But then I had to think about it. What about the movie we watched with all of them cuddled up on my lap? The good lunch we ate. The music we listened to and the bubbles I blew while the kids danced around me yelling "Pop! Pop! Pop!" From Luke's perspective it had been a good day. He was surrounded by people he loved (most of the time) and doing things that he enjoyed. I had been focusing so much on the negative parts of the day, I had almost forgotten the good. I've been doing that a lot lately about many things in my life. I guess I should look at life from a five-year-old's perspective more often.



**S.D.A.**  
Shwachman-Diamond America

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