

# S.D.A. ♥

Shwachman-Diamond America

Shwachman  
Diamond America  
eNewsletter

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## Inside this issue:

When A Child is Born 2

More than a Gift A Lesson 3

SDS Registry Update 4

Hello from Over the Pond 5-8

## An Update on Jasmine

Jasmine Binette had her bone marrow transplant on December 29<sup>th</sup>, 2009. She is doing well at 4 days post transplant, she even ate a four-egg omelette! Please keep Jasmine and her family in your thoughts and prayers. Jasmine's brother, Jacob, passed away due to bone marrow transplant complications in 2003.



If you would like to send Jasmine a card while she is in the hospital, please contact Shwachman-Diamond America for the address. Jasmine is expected to be in the hospital until the end of January early February.

Merry Christmas!

## When A Child is Born

When a child is born, she joins a family.

A family of grandparents, parents, brothers, sisters, uncles, aunts, and cousins.

Grandparents tell the child where she came from.

Parents teach the child where she is and how to be.

Brothers and Sisters teach the child how to cope with love and with hate.

Uncles, Aunts, and Cousins are there to support the child and to remind her to not take life so seriously.

Then the child grows up. Mostly, she feels loved, cared for, respected, and understood.

But many times, she doesn't.

She feels lost.

Because she is part of a world that is different from others.

She makes "friends." She grows, she learns, she hurts. But she doesn't tell.

She knows she doesn't quite fit, but the others believe she is like them. They don't understand, they can't understand, and they don't want to understand.

The Ones called "friends" try, but they can't understand.

Because she is different. She is strong. But she needs help to survive and to thrive.

Help from family, doctors, nurses, researchers, and friends.

They keep the child well. They keep her safe. Safe from the world that is not safe for her.

But sometimes they can't.

She is surrounded by well-meaning people who try, who don't understand. They say she is the same. They say she is special. She knows that she is different.

She treasures those who taught her where she came from, where she is, and who are with her as she moves forward. She knows they care. She knows they try. She is who she is because of them.

She still feels something missing. She moves on.

She changes as she grows. She can look outside of herself now. She is NOT alone. There are others like her. There are others who are different, who are strong, and who need help.

There is nothing missing. She discovered how to see.

They have been where she has been, experienced what she has experienced, and felt what she has felt. They share their experience with her. They share their anger, their heartache, their passion, and their strength.

They share with her the strength to know that she can choose. She has power in her life.

She is safe to feel. She is safe to be.

And she knows. She is not alone. She is special. She is different. She is accepted and accepting.

She will become who she will become because of the Ones who know. The Ones who will always be with her. And the Ones who will always understand.

They are her Chosen Family. They are her bond.

By Felima Hohn

In honor of Arianna and Sadie

## More than a Gift... A Lesson

On April 27, 1998 our third child was born, another little girl, Jeryca Lynn Christenberry. As I lay there on the table fear took hold of me not a sound filled the room. Lying there temporarily paralyzed my voice strained as tears filled my eyes "What's wrong with my baby?" "I don't hear her!" Just then a nurse brought my girl to my side and her tiny little face stared back into mine. Her eyes wide open and not a peep as if she was soaking in her new surroundings.

Born full term by C-section, She weighed in at 5 pounds 14 ounces. She was 17 ¾ inches in length. She was no bigger than a doll you would buy at a toy store. Soon after birth the Doctor noticed Jerycas bilirubin levels were above normal and she spent the following three days under the lamps . The nurses all assured me that this was a normal thing, that it was nothing to worry about. I was not so sure I somehow knew from the beginning of my pregnancy that something was not right.

Three weeks after bringing Jeryca home she had her first ear infection. I began asking questions I was concerned, again the doctor reassured me that all was okay. At six weeks old Jeryca was again treated for an ear infection. The doctor was puzzled this time, not only did she have an ear infection but also there was a boil like growth behind Jerycas ear, we again treated her with antibiotics. Doctor Wildbaughn then referred us to Doctor Kale a pediatrician in Galesburg Illinois. It was then that we did her first CBC and Dr. Kale noticed something not right with Jerycas White blood count. A few weeks later Jeryca came down with another ear infection and this time we had severe diarrhea that smelled horrible, her tummy also seemed to be distended and she cried a lot as if she was uncomfortable. This time Dr. Kale admitted Jeryca into the hospital. He again ran a CBC and this time noticed she had no neutrophil count. It was at this point he sent us to Iowa City Children's hospital. It was here that we were introduced to Dr. Tanoos and Dr. Fredrick Goldman. It was at this point that our lives began a long journey that we could have never imagined. There were tests after tests and several doctors and nurses. It was frightening to be in this unfamiliar place with a child so sick . To look around you could see the faces of exhaustion and little hope. The mothers and fathers of these babies lying in beds and wondering around the hospital unit. My heart was broken and my body ached with fear. Over the next few weeks we were plagued with more questions than answers. All the while our child lay in a hospital bed hooked up to IVs growing increasingly more ill. Jeryca was swollen twice her size and orange from all the infection and Jaundice. After several antibiotics and attempting to treat a mystery illness, we now had C diff to add to the list of ailments to treat. Before I knew it we had spent three months of our lives in this hospital which had now become home. As I sat in Jerycas room with her one night I remember feeling lost. I sat there rocking her to sleep as tears welled in my eyes, my thoughts drifted to the reality that our children do not belong to us and that they are only lent to us for a period of time and she was gods child . It was at that point that I came to terms, so I thought with the fact that I may not walk out of this hospital with my child. I placed her in her bed that night as I was getting ready to leave to go get some dinner I kissed her and told her I loved her. Her father and I got as far as the parking garage and our pager went off, we were called back to Jerycas room she was having a seizure that would last for nearly 30 minutes. We spent the next 7 days in PICU, a few days later back in the pediatric unit we finally got some good news, they had narrowed it down to Kostmans syndrome or Shwachman Diamond Syndrome. Doctor Goldman had also noticed that Jeryca was not producing neutrophils. We were approached with GCFS and now had to learn to give shots. A few more weeks went by and we were finally released to go home armed with GCFS. We had several other hospital stays and sicknesses along the way it seemed at times that we were in a never ending battle against this sickness. Then came the next hard hit, Jeryca had stopped responding to the GCFS and we were faced with a decision to let nature takes its course or to go through with a bone marrow transplant. After testing everyone for a match there was not a match close enough to risk it. Dr. Goldman came to us with the idea of a stem cell transplant . We would use the stem cells from an umbilical cord donor. This was still experimental at the time; we decided to go for it. On September 9<sup>th</sup> 1999 Jeryca being 11/2 years old at this time was the first stem cell transplant to be done at Iowa City Children's hospital. On October 13<sup>th</sup> 1999 we were released to go home, within six months Jeryca was off all medications and within a year she had a complete functioning immune system, her bone marrow was now functioning as it was suppose to be. Jeryca is now 10 years old.

## SDS Registry Update

December 14, 2009

To Shwachman-Diamond America,

We are happy to report that, with the concerted efforts of the entire group, the SDS Registry opened to patient accrual in December 2008. Since the primary site in Seattle was required to open before Toronto and Cincinnati could proceed with their IRB/ethics board submissions, of necessity patient accrual has thus far centered on Seattle. Conditional IRB/ethics approval has been obtained at Cincinnati and Toronto so the SDS Registry is poised to open soon at those sites. In the meantime, all three sites have been active in recruiting and referring patients to the Registry. As a result of the combined work of all the investigators, patient accrual (35 patients consented) has already exceeded our projections for the first year of the Registry. Our thanks to Shwachman-Diamond America for your efforts to get the word out to the SDS community regarding the Registry!

We are also fortunate that an abstract describing the SDS Registry was presented at the latest annual American Society of Hematology meeting in New Orleans, LA. It was a wonderful opportunity to educate pediatric and adult hematologists about SDS and the Registry. In addition to the 35 registered patients, we have received 10 peripheral blood samples and 12 bone marrow aspirates for the repository. We have received complete medical records for 16 patients.

This year, efforts focused on designing the data extraction forms and the database after extensive research into other successful Registries currently in operation. We would like to thank all the many outside investigators who graciously shared with us their experience and expertise. We also sought input from biostatisticians experienced in Registry-based research and from database designers experienced in clinical research. In the spirit of international cooperation, we shared the data extraction forms developed for this Registry with the worldwide SDS research community. We hope that capturing a common set of data in a similar format will strengthen the efforts of each country.

In year 2 of the Registry, we will open the Registry for accrual at all three sites. We will also pilot the data extraction forms and entry into the database with the goal of running a test run query of the database. We will continue development of the database for the patient questionnaire. We are consolidating our efforts with that of the SCNIR, our partner Registry, to streamline costs and workload. Another important goal for year 2 is to have the Registry website go live. To be consistent with the SDS Consensus Guidelines, we will defer full completion of the educational entries for the web site pending final consensus. In the meantime, we will provide a link to the Shwachman-Diamond America web site for family educational materials.

Thank you for your continued efforts to support this important project.

Sincerely,

David C. Dale  
Peter Durie  
mens  
Akiko Shimamura

Stella Davies  
Richard Harris

Yigal Dror  
Johanna Rom-

## HELLO FROM OVER THE POND!

Eventually I get tapping after a very honourable request to write an article. We feel very humbled to be asked to write an article for what we class as the best resource for SDS sufferers around the world.

Our family journey started even before our immediate family sufferer was born. Some 11 months before our youngest daughter Kimberley was born, one of my sisters gave birth to her daughter Charlotte. It was not a particularly good pregnancy and an even more traumatic birth, including her first resuscitation. Eventually my sister came round to no child at the side of her and said the immediate question of ..."where is my child? Is it alive?". Fortunately she was, but up in the high dependency unit struggling to breath, feed and visibly showing head trauma of the birth.

Charlotte was nearly 2 months old before the hospital felt she was stable enough to go home. Eventfully she got to see her new home. In the meantime, I became pregnant with our second child. Charlotte's plight continued with seemingly no understanding from the doctors as to why she was struggling so much, and included the introduction of a feeding tube to ensure her nutrition intake due to her continual failure to be able to feed normally. While I was pregnant, I recall us being called to her final farewell on more than one occasion! One event, following a supposedly simple investigation of her airways, I recall having to sing to her to keep her calm and focused as the doctors yet again fought for her life; Mum & Dad where, as you can imagine very traumatic at this stage, once again. After that episode it was confirmed that she was allergic to being under antithetic.

Shortly after this, our second daughter was born, not a very memorable pregnancy or birth; until the concern from the doctors that she was cold! Dad promptly helped her to raise her body temperature. Soon, we where shortly on our way home with our new 6lb 3 parcel. The joke of she never stops crying, used to amaze people when I would talk about her – the times she did sleep – until they met her and realised I was not exaggerating. When I was not attempting to force feed her with a prescribed syringe due to her inability to latch on and her constant crying.

I spent a lot of the time visiting and supporting my sister and her, seemingly, continual stay in hospital. Failure to thrive comments started to be bounced around about Kim's lack of weight gain and then the label of paranoid status of a mother began to appear with the constant visit to the doctors with Kimberley's inability to be without pain and her very loose stools. "She has colic..." I recall was mentioned – despite the instinct chant that this is not colic. Her first Christmas and New Year was spent in hospital.

## Over the Pond (Cont.)

After a day of whatever went in – eventually – came out the other end as if it had just come out of the tin/jar! A cold jumped to pneumonia within 24hrs – doctors could not believe it! Then I felt myself watching my daughter as I had earlier watched Charlotte; whom was in another hospital in the area! Eventually Kimberley recorded on the hospital consultant chart and the tests began.. Crohn's, celiac and sweat test for CF.. all negative.

In the meantime, Charlotte was in hospital and back in November 2001, she appeared to be back on the journey to the light! Knocked out with morphine and her tiny body piped up to too many machines to get close to her, I tried to comfort my sister as much as I could. Our parents had just left the country for their winter holiday and I recall seriously asking the head nurse “ do I need to call them to tell the to come back!?”

Fortunately that day, there was a nurse whom had transferred from Manchester ( other side of the country for us) and she recalled a child similar to Charlotte displaying failure to respond to treatment appropriately and they had Shwachman Diamond Syndrome... well off they all went to pull it up on their medical search engine! At the time I could not believe their actions of getting answers from the computer! But as I understand now, no one had ever seen it or knew about it – it was just one of those things in a text book! Treatment was identified that seemed to stabilise her – in the meantime fortunately the World DNA test was discovered & so her blood was sent off to confirm the visual call. I then trawled the internet to attain information for my sister of the syndrome and filtered a lot!

Annoyingly our local consultant aware of Kim's' cousin s' diagnosis thought she had the same.. we were shocked that he could jump to such a thought– yes I was related and maybe a carrier, but not my husband! However tests were taken – over a messy month of NHS dis-organisation and the waiting game started! Over a month later; 31 March 2002 , my last day of maternity leave , I got the call at home to confirm the initial consultants thoughts as correct. Fortunately I was not holding Kim as I collapsed in shock – having watched Charlotte and thinking of all the things we would have to go through yet!

So, our journey began for real! After much blaming, crying and questioning of what did we do wrong... we – as I am sure we all do – just got on with every day at a time!

After the first year of denial we eventually completed the documents to register Kim disabled and in so attain financial help with her day to day care. I then felt, though I was not of the medical groups wanted and needed to be doing something for my daughter, niece & other sufferers - and so my journey of fundraising and awareness started.

## Over the Pond (Cont.)

After some years with the National UK group, I decided to work on a more local ground, and with my sister accidentally set up an group known as KC Dream (K Kimberley ; C Charlotte). Following a mad idea to jump out of an aeroplane, as well as 4 other mad friends, & a small article in the local press asking for support ;KC Dream came into existence with the need to deposit the offerings of cheques for the cause focus somewhere. This initial article took our journey to the nearby City paper, then the local city BBC radio Show , big article in National Paper and eventually TV news coverage that grew to an half an hour programme of “ A day in the life of ... “. We hope to continue to encourage our support in the numerous rafle events we do in the local area – usually resulting in me been strapped to a bike for a day for up to 8 hours or leading 2 hour fitness classes, collecting mobile phones, aluminium cans, internet shop & search engine! I know it is not a lot but I feel it helps us deal with Kims’ suffering and others; as we raise the awareness of this rare disorder and try to help even out the balance of, because we are part of such a rare disorder the research in it seems to be so far down the National agenda.

I recall when we discovered that nonw of the family members where a bone marrow match for Kim’s, we asked for the support of a designer baby – in the UK no way! After a year of searching, a clinic in Belgium maybe be interested to help at our cost and a choice of bone marrow match but maybe with SDS or not SDS but maybe not bone marrow match. All this after a 2 year wait of researching the disorder as no one had ever done it in the world before! We choose not to go ahead, but I did offer to be a test start to help this research but it was deemed as not medically accepted – I just could not believe no other family had asked and wanted to get the ball going!

Anyway our journey with Kim & Charlotte , at times, as I feel the seeming “ one in a Million Chance” as the media label the cousins’ deal; has put more distance between us than if they did not both have it! Each scared of saying or asking the wrong question and the constant fight and comparing the girls. We are well aware that though Kim has her moments they are far fewer than Charlotte and other sufferers. That drives me on that while we are able to maintain a normal working family life to the full - even if it means watching Mum do silly things , then we will continue 110% while we can. We find that the diagnosis and possible expected short life span makes us do things than we may not have normally thought of. We continue to take holidays abroad and see as much of the world as possible, we have been lucky to get to your great state of Florida, (especially Orlando) and hope to go visit the original resort in California next year! Do mad things while away, take those excursions that maybe we would save for the next time we come! The most recent that got me up in hospital and under medication for a year, was our visit a couple of years ago to Tenerife, Canary islands and our (my) gung-ho! spirit of lets get up that volcano! Dad and oldest daughter stopped half way, but stubborn mum proceeded to carry Kim up determined she saw it “just in case” nearly to the top – got told to go down by a ranger ( appeared from nowhere) as the



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## **Over the Pond (Cont.)**

park was closing! I was a few more feet from the edge! Came down with a headache that lasted a further 2 days! Then later that year and with failing eyesight and thinking too much PC work, went for an eye test to be rushed to hospital scared to death of a potential massive brain tumour to be operated in that night – had planned to go shopping that afternoon! Wasn't, but after further tests and been hospitalised 3 brain clots were discovered due to, they think now altitude sickness through climbing the volcano under stress (carrying Kim) and dehydrated! I am pleased to report on the mend and can put back on the fundraising plan the London to Paris cycle. Kim's health maintenance is good, though challenging and comments of "do not know how you do it" but as we all know we do and will continue to. We wish you all the best for the times ahead and give our wishes of good memories to those whom have lost their angels and as Pocahontas said in your great resort "I'd rather die tomorrow than live a hundred years without knowing you" and also as Grandmother Willow said as she dipped her branch into the water "Look the ripples so small at first, but see how they grow. Someone has to start them!" So let us continue to create that great big wave. I have our family surf board ready!

Kimberley (6 this year), Georgina (8 this year), Paul (best Dad in the world) & Jules (maddest Mum in the world!) x