Shwachman-Diamond Syndrome Foundation

Cure For A Future

NEWSLETTER

SUMMER 2009

Happy Summer everyone!!

So far this year we have managed to accomplish many things. We are still going through the research granting process and are reviewing 2 very exciting proposals!! It is a 6 month process that is guided by our Medical Advisory Board (listed at the bottom of the newsletter.) You all probably received the email that stated that we raised the amount of our grants from $30,000 to $65,000 in order to attract more proposals and more researchers to SDS. I also attended the Shwachman Diamond Syndrome International Congress in late June in Amsterdam. It was an excellent meeting that attracted scientists and researchers from around the world to discuss SDS, its diagnosis, symptoms, treatments and management. We hired a bookkeeper this past winter but she is donating her time to SDSF and thus we are not paying for her expertise. The web site still has changes and updates to make but we are progressing. Every week we receive emails from families recently diagnosed with SDS and we welcome them into our SDS family. It is so hard to get the diagnosis of SDS and not understand what it means – yet! SDSF strives to give the most up to date disease information, referrals to regional family contacts, clinics to go to and other doctor referrals, and information about the registry that we encourage you to participate in.

We are also saying goodbye to Susan Utz. Susan is leaving the SDSF Board of Directors after dedicating 5 years of service to the Foundation and our community. We are so thankful for her help and her work over these last 5 years and have enjoyed getting to know her and her wonderful family. Susan's son, Logan, with SDS, has contributed to our newsletter a couple of times and even held his own fundraiser for the Foundation. He is off in college now and doing very well and Susan has 2 younger children to care for as well as working full time. We will miss her and wish her and her family well. THANKS SUSAN!

We are discussing many ways to fundraise to pay for research and welcome any ideas you may have. Please contact me.

Until the fall,

Blair Van Brunt, President, SDSF

Shwachman-Diamond Syndrome Foundation

127 Western Avenue
Sherborn, MA 01770 U.S.A.
1-888-825-SDSF (7373) (Toll Free)
FAX: 1-888-825-SDSF (7373)
E-mail: info@shwachman-diamond.org
Website: www.shwachman-diamond.org

PLEASE CONTRIBUTE TO THE ANNUAL FUND DRIVE

We accept checks, stocks, contributions from IRAs, payment through our web site on the Get Involved page, matching contributions from your employers using our #9588351, and our tax ID # 43-1709945, ask friends and family to donate through the CFC or United Way campaigns (the numbers are listed in the newsletter) asking family to donate to SDSF in lieu of flowers upon the passing of a loved one, and of course any fundraiser that you can think of. To request information on any of these items email or write to us at:

SDSF, 127 Western Ave, Sherborn, MA 01770

Don't forget that you can sell the rubber bracelets or purchase the crystal bracelets to help in this fundraising. Perhaps sending out the letter below that was in the Winter Newsletter? You can copy, edit to your liking and print it out on our stationary or email it to all your friends and have them pay through Paypal through the web site. Please email SDSF info@shwachman-diamond.org or call 1-888-825-SDSF (7373) to request any of these items.

Here is a sample letter for your use:

Dear Family and Friends:

As you may know, our (family member, relation to you, and name) has Shwachman Diamond Syndrome. This disease is diagnosed in patients with bone marrow dysfunction (anemia, neutropenia or pancytopenia, multiple infections, bone marrow failure and higher risks for MDS, AML or ALL leukemias), pancreatic insufficiency (children are unable to digest food without pancreatic enzyme supplements), skeletal abnormalities (bone growth doesn't match the age growth thus giving rise to bone abnormalities that can require surgery), and other lesser reported symptoms involving the lungs, heart, liver.

We are writing to you to ask you to send in any amount of money that you can to help us help (name of patient) through the efforts of Shwachman Diamond Syndrome Foundation.

Shwachman Diamond Syndrome Foundation was started in 1994 by Joan Mowry, a mother of an afflicted child and it has grown to serve over 300 patients and their families, plus hundreds of medical professionals who ask for and receive information from it. It was the first SDS organization in the world and it spawned numerous other support groups due to its leadership and vision. With only 7% of the revenue going to administrative costs, 93% of every dollar donated goes to the activities listed below.

Activities supported and funded by SDSF are sending New Family Packets comprising of pertinent information for new families to learn about SDS, advocating grants for and fund research for treatment and possibly a cure, baskets to hospitalized patients, providing email/phone family support, publishing tri-annual newsletter, attending pertinent medical conferences to educate and raise awareness in the medical community, hosting and maintaining an international web site, and hosting a bi-annual, free-for- the- family conference. The gene was discovered December 2002 through the financial help of SDSF and this has given way to a better understanding of the disease. It has led to diagnostic tests that have been developed to help with faster diagnosis as well as to insight into how the gene works. This information can lead to possible therapeutic treatment agents and potentially a cure.

Our family has benefited from many of this support and activities and wants to continue to do so. Please help by going online to www.shwachman-diamond.org and donating through Paypal on the Get Involved page or send a check in to: SDSF, 127 Western Ave, Sherborn, MA 01770. We will have your name published in the newsletter as having contributed in (name of patient) honor.

This is not a case of 'what if" (name of patient) takes a downturn, but 'what if" we help fund a cure!

Don't forget to fill out the Matching Gift fund paperwork from your employer to maximize your

donation and the number is 9588351. The tax exempt number is 43-1709945

Thank you for your consideration to donate. Please feel free to call or email me with any questions.

Sincerely,

HAVE FUN FUNDRAISING!

Hi! My name is Julie and my husband, John, and I have done many fundraisers. We have raised money for important research for SDSF that benefits ALL families dealing with this disease. I have to say that my favorite fundraiser of all is working with local high schools! Almost all high schools every year pick a charity that they raise funds for, and I feel that this may not be known to many of us SDS families looking to pitch in with funraising for SDS research. I have found that just by calling your local high schools, most will say that they DO raise money for charity. I always ask if I could give a small talk or presentation about SDS to the school or student council in the hopes that they will choose SDS as their charity to work for. In four years, I have had 4 high schools raise money for SDS!

Three years ago, Athens High School choose to sponsor Shwachman-Diamond Syndrome, these students raised over $15,000. This year I went in and talked to the student council again, and they seemed to love to help needy charities, especially like ours where we do not get government funding. Showing them Patrick's picture and making it personal always helps the students to really connect with what SDS is all about. Next week is their "charity week" and their goal is to raise $20,000 for SDS research this year!! For me, it is the easiest way to fundriase. The students do all of the work planning and just keep me updated on how things are going. In the end, SDSF and all SDS families benefit from all their hard work. Thank you Athens High School!!!

Happy fundraising!
Julie Kroppe, jkroppe@wowway.com
(reprinted from earlier newsletter)

SYNOPSIS OF THE 5TH INTERNATIONAL SCIENTIFIC CONGRESS ON SDS

written by Blair Van Brunt

I have just returned from the 5th International Scientific Congress where approximately 85 doctors and scientists who have been studying SDS or are interested in learning more about SDS gathered in Amsterdam this past June 18th-20th. In addition to the aforementioned were representatives of many of the international support groups as well as a smattering of family members of SDS patients from various countries such as Holland, Denmark and the USA. Countries that were represented through their Foundations were: Canada, the UK, Italy, Holland of course, and Norway.

The Congress was organized by Dr. Liesbeth Siderius and Dr. Taco Kuijpers, both of the Netherlands and was financially supported by collected registration fees and by Shwachman Diamond Syndrome Foundation (USA), Shwachman Diamond Syndrome Canada, Shwachman Syndrome Italian Association, Stichting Shwachman Syndroom Support Holland and Amgen (Amgen is a manufacturer of medical drugs such as Neulasta and Neupogen). These kinds of conferences are very expensive to organize and we on the board of SDSF are proud to be sponsors of a meeting that gathers so many brilliant doctors and researchers who are diligently working on our behalf to create benefits for our SDS community. As one attendee, who is fairly new to the SDS medical community said, "Going to Amsterdam...let me learn more than I could by reading for six months."

The topics covered at this meeting included: genetics and phenotype variation asnd the known SDS gene

(SBDS) with its up-to-date information on its function, modeling and expression. Various aspects related to growth and abnormalities, gastrointestinal features, bone metabolism, psychological and behavioral issues, hematological and bone marrow transplant issues were also presented. Discussions highlighted many remaining questions including when and what causes a patient's bone marrow to transform resulting in aplastic anemia, MDS or leukemia. Refinement of diagnostic guidelines and the medical and psychological issues pertaining to the growing adult population were also discussed. Sub-topics including how mouse and other animal models can help determine the function of the gene and find other responsible genes; research on finding other mutations and other genes for the SDS-like patients were also mentioned. There was also a session with 20 posters giving more detailed information on ongoing research. On the Thursday prior to the actual meeting, two additional working meetings were held: one to discuss consensus guidelines for diagnosis and management, and the second to provide an update on efforts to obtain national and international patient Registries. These meetings correspond to collaborative work that is ongoing by doctors and researchers and involve several countries.

We are not publishing the detailed program of the Congress because this would jeopardize the potential of each of the presenters to publish their work in a medical publication. We all benefit when one of "our" researchers publish in the medical literature because then more doctors and scientists from around the world can learn about SDS. There are increasing efforts by our government and others to promote public access to all medical literature, so stay tuned. The more awareness we have of this disease, the more that your local doctor will discover about SDS diagnosis, treatments and management. This conference has helped to create more awareness for SDS in terms of attracting more research and researchers and possibly more funding as well. Please continue to financially support your support group or foundation, whatever country you are in, as we support these international, medical research conferences. Every SDS patient and his/her family benefits from this important work. There are very few drug companies and corporations who are willing to donate funding to this rare disease and we are counting on you and your family and friends to support SDSF which in turns helps pay for these meetings and of course research.

The next scientific congress will be held in the New York, USA area.

The list of presenters was follows:

Dr. Johanna Rommens, The Hospital for Sick Children, Toronto, Canada; **Dr. Heather Ball**, The Hospital for Sick Children, Toronto, Canada; **Dr. Elena Nicolis**, University Hospital, Verona, Italy; **Leslie Steele**, MSc, Dept. of Pediatric Lab Medicine, The Hospital for Sick Children, Toronto, Canada; **Dr. Nikolaus Trede**, University of Utah; **Dr. Ivo Touw**, Erasmus Medical Center, Rotterdam, Netherlands; **Dr. Jean Donadieu**, AP-HP Hospital Trousseau, Paris, France; **Hanming Wang**, The Hospital for Sick Children, Toronto, Canada; **Dr. Alan Warren**, MRC Lab of Molecular Biology, Cambridge, UK; **Christine Hilcenko**, MRC Lab of Molecular Biology, Cambridge, UK; **Rastislav Horos**, Erasmus Medical Center, Rotterdam, Netherlands; **Dr. Sanna Toiviainen-Salo,** Helsinki University Hospital, Helsinki, Finland; **Dr. Liesbeth Siderius**, pediatrician, the Netherlands; **Dr. Johnson Liu,** The Feinstein Institute for Medical Research, Manhasset, NY; **Dr. Peter Durie**, The Hospital for Sick Children, Toronto, Canada; **Dr. Jutta Koeglmeier,** Chelsea and Westminster Hospital, London, UK; **Dr. Marie Helfrich,** University of Aberdeen, Aberdeen, UK; **Dr. Arif Manji**, The Hospital for Sick Children, Toronto, Canada; **Marina Tourlakis**, MSc, The Hospital for Sick Children, Toronto, Canada; **Dr. Jan Booij**, Academic Medical Center, Amsterdam, Netherlands; **Dr. Elizabeth Kerr**, The Hospital for Sick Children, Toronto, Canada; **Sandra Perobelli,** Cystic Fibrosis Center, Verona, Italy; **Dr. Hans Joenje**, Free University, Amsterdam, Netherlands; **Christopher Allen,** BSc, The Hospital for Sick Children, Toronto, Canada; **Dr. Gulay Sezgin**, Schneider Children's Hospital, New Hyde Park, NY;

Saswati Sen, BSc, The Hospital for Sick Children, Toronto, Canada; **Dr. Robbert Bredius,** Leiden University Medical Center, Leiden, Netherlands, **Dr. Akiko Shimamura**, Fred Hutchinson Cancer Research Center, University of Washington, Seattle, WA;

Group at SDS Congress

Dr. Akiko Shimamura, Pediatric Hematologist, USA and Dr. Johanna Rommens, Geneticist, Canada

Dr. Peter Durie, Pediatric Gastroenterologist, Canada on the microphone introducing Dr. Sanna Toiviainen-Salo, Radiologist, Finland

Dr. Lisbeth Siderius, Pediatrician, Holland Support Group (left) with Dr. Peter Durie, Pediatric Gastro-enterologist, Canada, (middle) receiving a Delft plate from another Dutch support group representative, Irma Dijk

Dr. Johnson Liu, Adult Hematologist, USA (left)

Dr. Jean Donadieu, Hematologitst, France (right)

Families & International Support Group Representatives

Dr. Liesbeth Siderius, Peditrician, the Netherlands

Priniciple organizer of SDS Congress 2009

SHWACHMAN DIAMOND UNITED

Amsterdam, June 18, 2009

Every other year representatives from the international support groups get together at the SDS Scientific Congress to discuss how things are going, what the important issues are for each group, how everyone is handling all the different aspects of their own SDS community, current research involving SDS and more. This year we met for a few hours in Amsterdam, one day prior to the 5th SDS Scientific Congress. In attendance were representatives from the support groups from Norway, the United Kingdom, Italy, USA, Canada and Holland. There were also family members from New York, Holland and Italy.

Discussion in the meeting started out with what the International Medical Advisory Board is presently doing. They are re-formulating diagnostic and treatment consensus guidelines for a new booklet/ web document that every country can adapt for their support group. Also discussed was the hope for an international registry as many countries are implementing their own registries in order to eventually merge with everyone. Lastly we all submitted general questions pertaining to family support and research for the Scientific meeting that could be answered by the doctors/scientists in attendance.

We created and approved the following document which was read during the Congress concerning the importance of an international registry:

Shwachman Diamond United supports an International Registry because it will:

- Define the size and demography of affected populations so the appropriate services can be planned.

- Share clinical knowledge and experience about Shwachman Diamond Syndrome (SDS) natural history, variability and optimal management, which can be modified as experience develops.

- Improve health care by understanding different aspects of SDS and enable early diagnosis, prevention and therapy.

- Identify an adequate population of potential subjects for research into SDS with a view to understanding of its pathophysiology which may lead to improving management and finding a cure.

The registry should be:

- A cooperation of all health care providers and family representatives.

- Based on consensus of diagnostic criteria and management of patients and research needs.

We anticipate a conference call to take place sometime in the fall to update each other on many of these issues mentioned. SDSF will keep you posted on these talks.

NORTH AMERICAN SDS REGISTRY

SDSF would like to encourage every patient with SDS to register in the North American Shwachman-Diamond Syndrome Registry. The more data that is available to doctors and researchers, the faster they can help with diagnosis, treatments and hopefully a cure!

Here are some answers to frequently asked questions that may help you decide if registration is right for you:

What is the North American SDS Registry (NASDSR)?
Since SDS is a rare disorder, our understanding of the medical complications and best treatments for SDS are limited. For this reason, the NASDSR was established to collect medical information and clinical samples on all individuals with SDS and who are SDS-like acroos the United States and Canada with the goal of improving diagnosis and treatment. The

NASDSR is headquartered at the Fred Hutchinson Cancer Research Center in Seattle with collaborating centers in Toronto and Cincinnati. The SDSR is a research organization dedicated to gathering and analyzing information about SDS and sharing any new knowledge with the SDS community and medical professionals. Scientific studies using collected blood and bone marrow cells will focus on elucidating the causes of SDS and its complications in order to develop better treatments.

How can you participate in the NASDSR?
Patients can enroll in the registry by contacting the Registry coordinator, Melissa Alvendia at 206-667-6965 (toll-free 1-866-792-5876).

After signed informed consent is obtained, the patient or parents (if the patient is a minor) will receive a brief questionnaire which will be updated annually. Additional information is obtained from medical records, laboratory and pathology reports. Clinical samples are obtained when blood, bone marrow or other tissues are being obtained for clinical care, so no extra procedures are involved.

In the near future, enrollment will also be available through additional centers including Toronto and Cincinnati.

How much will this Registry cost me?
There is no charge to you for your participation in this Registry.

For more information, please contact
Melissa Alvendia
Fred Hutchinson Cancer Research Center
1100 Fairview Ave. N
Mailstop D2-100
Seattle, WA 98109-1024
malvendi@fhcrc.org

BE THE MATCH℠ IS NEW NAME FOR NATIONAL MARROW DONOR PROGRAM® (NMDP) REGISTRY

NMDP seeks to raise awareness and participation in life-saving mission

The National Marrow Donor Program (NMDP) is a health care nonprofit committed to connecting critically ill patients with people who can potentially offer the gift of life. Yet despite its stature and vital work, independent market research conducted last year showed the general public had low awareness of the organization.

That's why Be The Match was born. Be The Match is the new name for NMDP's registry of volunteer marrow donors and donated umbilical cord blood units. But Be The Match is more than just a name – it's a call to action in the battle to help thousands of patients in need of a marrow or cord blood transplant.

"Seventy percent of patients who need transplants do not have a matching donor in their family. The Be The Match Registry is there to help those patients find the donor they need," explained Jeffrey W. Chell, M.D., chief executive officer, NMDP. "By changing the registry's name to Be The Match, we're making our mission and our message loud and clear to the public. Every individual holds the potential of providing the precious gift of life to someone who desperately needs it."

These patients include people with leukemia, lymphoma and more than 70 other life-threatening

diseases. Today, medical advances are making transplants available to more patients of all ages than ever before. NMDP introduced Be The Match to meet this need by engaging a growing community of people inspired and committed to helping patients in any way they can — by joining the registry, donating umbilical cord blood, contributing financially or volunteering.

The National Marrow Donor Program is still the name for the organization that operates the Be The Match Registry and partners with a global network of leading hospitals, blood centers, public cord blood banks, laboratories and recruiters. The NMDP facilitates transplants worldwide, conducts research to improve survival and quality of life, and provides education and support services to health care professionals and patients.

How to Get Involved: Visit BeTheMatch.org to learn more about the many ways to get involved. There are several ways to help support patients:

- § Grow the registry by becoming a member or donating cord blood
- § Make a tax-deductible contribution
- § Volunteer your time

Our NEW Combined Federal Campaign contribution number: 10799

Our NEW United Way Campaign contribution number : 1329

DIRECT-TO-CONSUMER GENETIC TESTING

Genetic tests are increasingly being offered directly to consumers and include at-home tests and those provided by private companies. The National Society of Genetic Counselors has issued a statement recommending that anyone considering undergoing genetic testing ask the following questions:

o Is a genetics professional or a knowledgeable healthcare provider involved in the process of ordering or interpreting a genetic test?

o Is the consumer provided with comprehensive information regarding what the test can and cannot say about his or her health?

o Will results be given in a manner understandable to the average consumer, with a clear explanation of their clinical implications, if any, and including resources providing appropriate follow-up?

o Is the scientific evidence on which a test is based clearly stated?

o Is the clinical testing laboratory accredited by CLIA, the State and/or other applicable accrediting agencies?

o Are privacy concerns addressed?

SDSF and the head of our Medical Advisory Board, Dr. Johanna Rommens, suggests to consult www.genetests.org . She has this to say about the organization: "I worked with them to get everything in Shwachman up-to-speed, including a review and description. They are excellent and a central resource for genetics disease - for professionals and the public."

WHAT IS A GENETIC COUNSELOR?

Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical genetics and counseling. Most enter the field from a variety of disciplines, including biology, genetics, nursing, psychology, public health and social work.

Genetic counselors work as members of a health care team, providing information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions. They identify families at risk, investigate the problem present in the family, interpret information about the disorder, analyze inheritance patterns and risks of recurrence and review available options with the family.

Genetic counselors also provide supportive counseling to families, serve as patient advocates and refer individuals and families to community or state support services. They serve as educators and resource people for other health care professionals and for the general public. Some counselors also work in administrative capacities. Many engage in research activities related to the field of medical genetics and genetic counseling. (Adopted by the National Society of Genetic Counselors, Inc. 1983)

http://www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf

To find a genetic counselor in your area, for information about a genetic counseling specialty, or for information about genetic counseling as a field, visit www.nsgc.org . This is a link to a great booklet that is currently online but you can also write to request one.

COPING WITH SDS: TRANSITIONING INTO ADULTHOOD

Part II

by Corky DeBoer

In the last SDS newsletter, I focused on some of the dynamics which families face when their child is transitioning into adulthood. I asked you to consider one or more of the following questions:

- "How do you view your teenage children?"
- "What are the biggest barriers, or do you think will be the biggest barriers, for you to view your son's/daughter's approaching adulthood, let alone being an adult?"
- "What has been most painful for you as your son/daughter is approaching or going through this transition into adulthood?"; "Most painful for your son/daughter?"
- "Who / what has been the most helpful or supportive person / resource for you as your son/daughter is approaching or going through this transition into adulthood?"; "Most helpful or supportive for your son/daughter?"

Today, I'd like to make a few comments on one aspect of this important transition into adulthood, namely, creating the space so that our young adults can function as independently and as interdependently as possible.

For this article, I will be focusing mainly on dynamics that arise in young adults who are primarily impacted by the physical health aspects of SDS, who have no or little cognitive and/or emotional impairment. In a future article, I will focus on issues and dynamics present in young adults with SDS who struggle just as much, if not more, with cognitive and/or emotional impairments.

One of the more difficult dynamics of any chronic condition or illness, especially as our children transition into adulthood, is compliance:

Compliance in consistently taking physician ordered medication.

Compliance with regard to physician recommended follow-up tests.

As part of this transition into adulthood, the young adult is trying to establish her/his own identity. In order to do this, decisions are sometimes made as a reaction against (that is, the exact opposite) what the young adult's parent(s) would have made on her behalf. In other words, there are often power struggles that take place on several levels, as the young adult seeks to set out on her own.

Since physicians are also seen as authority figures, young adults may consciously or unconsciously choose to stop taking certain medication (such as pancreatic enzymes), or to delay or altogether avoid regular bone marrow biopsies that the physician may deem important. If the physician has built a rapport with the young adult over the years, this can be a real benefit, since the young adult may be willing to listen to and more objectively consider the benefits of taking their medication or undergoing intrusive tests.

As parents, it is important not to get hooked by our young adults' behaviors. I know this is much easier said than done. One way not to get hooked is by being aware of and acknowledging the real tension between wanting to educate (or even convince) our young adult about the benefits of medications or tests, while at the same time wanting to honor their developmental desire and need to make their own decisions.

Hopefully, our young adults will not be reactive (striving only for their independence), but will act interdependently, by seeking out advice or support from their peers or other adults that they trust as they sort out their feelings and make decisions regarding their medical care.

I conclude with one of my favorite prayers, which may be helpful to you, whether you are a parent or a young adult, during these times of transition into adulthood, the well-known Serenity Prayer:

God Grant me the Serenity to:

Accept the Things I Cannot Change,

Courage to Change the Things I Can

And Wisdom to Know the Difference

If there are other topics you would like me to comment on, feel free to drop me a line at opcrccd@aol.com.

Rev. Corky DeBoer

ACPE Supervisor

Manager, Spiritual Services

Mission and Spiritual Care

Advocate Christ Medical Center

4440 W. 95th St., Oak Lawn, IL 60453

FAMILY SHARING PAGE

AN ADULT'S PERSPECTIVE ON SDS

by Michele Ellebracht as told to Joan Mowery

I'll be 47 years old in September which means I am one of the older SDS patients. I think my age gives me a different view about SDS then the younger patients have. My experience has been very different. We have all had a number of challenges to deal with because of SDS and each of our experiences is different. I am going to just talk about some of the experiences I have had.

First of all, when I was born I weighed 5 pounds and 7 ounces and was 17 inches long. For the first 3 years of my life the doctors had no idea what was wrong with me. I was sick for the first time when I was 6 weeks old. The doctor gave me antibiotics but they didn't work so they put me in isolation in the hospital. I was hospitalized 50 times by the time I was 3 years old both for infections and failure to thrive. I only weighed 10 pounds at one year old despite constant feedings. Each time I was sick the doctors put me in an isolation room because my blood counts were bad but they didn't know why. They didn't even know why I had 10 to12 bowel movements a day.

When I was 3 years old, my pediatrician happened to read an article in the New England Journal of Medicine that described a new syndrome that sounded like what I had been going through. I was again in the hospital when Dr. Louis Diamond happened to come to St. Louis for a medical conference. My pediatrician contacted him and asked him if he would come by and look at me. Dr. Diamond examined me and reviewed my records and determined that I had the syndrome that they were calling Shwachman syndrome at the time. Unfortunately although we now had a name for what was wrong with me that was all we had.

Drs. Shwachman, Diamond and Khaw had identified this new syndrome while treating patients at Massachusetts General Hospital who had Cystic Fibrosis. They noticed that about 17 of their young patients had some of the features of CF but also had other problems like blood abnormalities. That first medical article was simply a description showing how this new syndrome differed from CF.

Over the next several years, for no apparent reason, my blood counts improved somewhat and I was sick less often. I did start having other problems that the doctors said were not related to SDS—like learning disabilities, diabetes, osteoporosis, dental problems and orthopedic issues. We now know that these things are part of SDS.

Everything stayed pretty good until I was about 34 years old when my blood counts started dropping again and since then I have been sick and in the hospital more often again. By the time I was 43, my absolute neutrophil counts had dropped to zero, and I was in the hospital with one infection after another. The hematologists determined that I had to start neupogen shots and I have been taking them since. I have not been in the hospital since starting the shots for an infection. I have had some changes in my bone marrow which are concerning and I have seen the bone marrow transplant team recently.

Obviously I don't know what the future holds, but none of us do. SDS has come a long way. I can remember when doctors did not even know how to spell Shwachman much less know anything about it. I am happy for all of you younger patients that they now have tests to tell you if you have SDS and treatments to help.

If there is one thing I want you all to know it is—do NOT think that you are going to outgrow SDS, just because you are well and don't have any obvious problems. It may sit back and be quiet for a long time but it is always there. Keep vigilant and keep

seeing your specialists and stay on top of all the research and go to the conferences so you are informed on the latest treatments. We all may have SDS but we don't have to let it have us.

JOIN OUR "CLUB" FOR ADULT SDS PATIENTS

SDSF is becoming increasing aware that many of our children are reaching or soon will be reaching an age when they are no longer able or wish to be seen within the pediatric medical community. Older patients also have additional and/or different social and other concerns from children with SDS. For this reason, we are starting a "club" for older and adult patients. Some of the areas we will be approaching are establishing referrals within the adult medical community of physicians who have knowledge and/or experience in treating adult SDS and other BMF patients. In addition, the club plans to set up a referral file to direct adult patients to those who can assist them in obtaining health and life insurance as well as their medications.

We would like input from both the patients themselves and/or their parents with any information or suggestions you might have about what else can be done. Please send your thought and ideas to SDSF by either calling 1-888-825-SDSF (7373) or emailing us at info@shwachman-diamond.org.

SPOTLIGHT ON RESEARCH

STUDIES ON THE MOLECULAR MECHANISMS OF BONE MARROW FAILURE

Bone marrow failure (BMF) syndromes such as aplastic anemia or myelodysplastic syndrome (MDS) may develop by a number of different mechanisms. We believe a genetic predisposition to aplastic anemia and MDS is much more common than currently appreciated, and a significant proportion of individuals thought to have "idiopathic" aplastic anemia or myelodysplasia may have a genetic alteration as the underlying or predisposing cause.

Drs. Monica Bessler, Philip Mason, and David Wilson at Washington University in St. Louis, have begun a new study to identify alterations in genes that may predispose a person to the development of bone marrow failure and how these alterations cause disease. We are collaborating with researchers at several other institutions throughout the United States.

Our study seeks to identify genes, their mutations, and their role in the development of bone marrow failure and the genes contributing to leukemic transformation. By understanding the genetic contribution, we hope to gain a better understanding of the course of the disease and ultimately factors that predict leukemic transformation and response to treatment.

Our study is open to all children and adults who have or had aplastic anemia (inherited or acquired), paroxysmal nocturnal hemoglobinuria (PNH), or MDS.

Advancing our knowledge of how these conditions develop is only possible because of the participation of individuals with bone marrow failure. The study is still seeking volunteers, and anyone wishing to participate may contact the study coordinator for more information.

Our study uses a comprehensive approach to the evaluation of participants, which is necessary to truly understand the genetic contribution to the development of disease. Individuals who wish to participate will be asked to:

* Sign a consent form indicating your desire to participate,

* Complete a written medical and family history questionnaire,

* Submit a sample of blood (we can provide kits so a physician can draw your blood), and

* Undergo a physical examination (for families in the St. Louis area only).

Individuals are not responsible for any costs associated with the study. The confidentiality of all study related materials will be maintained in accordance with State and Federal laws. To learn more about the study please visit our website at http://bmf.im.wustl.edu/, or contact the study coordinator:

Jennifer Ivanovich, M.S., Study Coordinator

Washington University School of Medicine

Box 8100, 660 S. Euclid Ave.

St. Louis, Missouri 63110, USA

314-454-5076, jen@ccadmin.wustl.edu

Monica Bessler, M.D., Ph.D., Co-Director

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David Wilson, M.D., Ph.D. Co-Director

Division of Pediatric Hematology/Oncology

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Box 8208; 660 S. Euclid Ave.,

St. Louis, MO 63110, USA

Wilson_D@kids.wustl.edu

RESEARCH STUDY NEEDS YOUR PARTICIPATION

Dr. Akiko Shimamura, MD, PhD., Associate Professor of Pediatrics at the University of Washington in Seattle, is investigating the molecular functions of the SBDS protein. The goal of her research is to understand the causes of bone marrow failure and leukemia in Shwachman-Diamond Syndrome and to develop new therapies. Samples of blood and bone marrow from patients with Shwachman-Diamond Syndrome are critical for these studies. These samples may be collected when they are being drawn for clinical care. Dr. Shimamura or Dr. Trisha Wong would be happy to discuss the study further with anyone interested in sending a sample. Dr. Shimamura's e-mail is, shima2@u.washington.edu.

ONGOING RESEARCH NEEDS YOUR CONTINUED SUPPORT

SBDS Protein Expression in Peripheral Blood Leukocytes

Shwachman-Diamond Syndrome (SDS) is a rare genetic condition which causes a number of problems in different body organs, particularly the bone marrow (blood producing cells), pancreas (digestive gland) and bones. These problems may vary considerably from person to person, which sometimes makes it difficult for doctors to diagnose SDS. The mutated gene which causes SDS has recently been identified. To have SDS, one has to have a copy of the mutated gene on each chromosome. So far, three common mutations as well as about 50 rare mutations have been identified. Approximately 60% of SDS patients carry common mutations on both chromosomes. Others carry a common mutation on one chromosome and a rare mutation on the second chromosome. However, in about 5-15% of people with clinical findings of SDS, mutations cannot be found, even after extensive laboratory testing.

Each gene in our body acts as a code for making a unique protein. Each protein has a definite function. When a gene is defective, it either produces no protein, very little protein or produces a protein that doesn't work properly. We have now developed a way of measuring the normal SDS protein in blood cells from people without SDS. In addition, we have shown in a small number of people with SDS, that the level of this protein is absent or greatly reduced. These preliminary results suggest the possibility that we may be able to use this test to diagnose SDS. We also think that the amount of protein in blood cells might help to explain why some people with SDS have worse or different problems from other people.

To do a more extensive study of the SDS protein in blood cells, doctors at the Hospital for Sick Children, in Toronto and the Children's Hospital in Boston are seeking volunteers to join this research project.

We are seeking the following individuals to join our research study:

· People with SDS who carry the uncommon (rare) mutation on at least one allele.

- Parents of people with SDS who carry the rare mutation and/or an adult sibling who is a known carrier of the rare mutation.

· People with a confirmed clinical diagnosis of SDS but no SBDS mutation have been identified.

· People who are suspected to have SDS in whom testing remains inconclusive. This might include people who have: (a) a problem in the bone marrow but no known problem in the pancreas, or; (b) a problem in the pancreas and/or skeleton but no evidence of a bone marrow problem.

Individuals who wish to participate will be asked to:

- Sign a consent form indicating willingness to participate,
- Complete a brief written medical questionnaire,
- Submit a blood sample (we will provide a kit for your lab or physician)

Individuals will not be responsible for any costs associated with the study. The confidentiality of all study related materials will be maintained and no information that discloses the identity of the subject will be released or published without consent unless required by law. The results of the tests described above will be used for research purposes only.

To learn more about the study or to see if you are eligible, please contact the study coordinator:

Wan Ip at 416-813-6183 or wanip@sickkids.ca

Etiologic Investigation of Cancer Susceptibility in Inherited Bone Marrow Failure Syndromes (IBMFS)

The National Cancer Institute Institutional Review Board has given its approval to open a study entitled "Etiologic Investigation of Cancer Susceptibility in Inherited Bone Marrow Failure Syndromes." The principal investigator responsible for this study is Blanche P. Alter, MD, MPH. This study is open to patients with SDS, along with their immediate families. Individuals with one of the inherited bone marrow failure syndromes, and their parents, brothers, sisters, and children, are all invited to participate. Those who come to the NIH Clinical (CC) will belong to the "CC Cohort," and those who do not will belong to the "Field Cohort." Individuals who choose to participate in the NCI IBMFS [Alter, Blanche (NCI)] Cohort Study will be asked to complete a family history questionnaire and an individual information questionnaire. Physical examinations and samples of blood, bone marrow (from those affected with the disorder), and other tissues may be requested for research studies.

Inherited bone marrow failure syndromes (IBMFS) are rare disorders in which there is usually some form of aplastic anemia (failure of the bone marrow to produce blood), associated with a family history of the same disorder. Some of these conditions have typical changes in physical appearance or in laboratory findings which suggest a specific diagnosis. There are several well-described syndromes, which can be recognized by health care experts. There are also patients who are harder to classify, but who appear to belong in this category. Patients with these syndromes have a very high risk of development of cancer [Alter, Blanche (NCI)] (leukemia or solid tumors). At the moment we cannot predict which specific patient with an IBMFS is going to develop cancer. The NCI IBMFS [Alter, Blanche (NCI)] Cohort Study will enroll North American families in which at least one member has or had an IBMFS. The web page "marrowfailure.cancer.gov" describes the study and provides contact information. By telephone, please call 1-800-518-8474 and ask for the Referral nurse or you may also contact SDSF for more information.

University of Texas Medical Branch-Galveston, Texas

Dr. Tarek Elghetany, Division of Hematopathology at the University of Texas Medical Branch in Galveston, Texas is studying the bone marrow and blood of patients with Shwachman-Diamond Syndrome for early signs of myelodysplastic syndrome and leukemia. If you or your child have a bone marrow study performed, Dr. Elghetany can perform several research studies on the samples. Dr. Elghetany will also receive some bone marrow samples from Dr. Blanche Alter.

Dr. Alter is the principal investigator for the Etiologic Investigation of Cancer Susceptibility in Inherited Bone Marrow Failure Syndromes (IBMFS) that is taking place at the National Cancer Institute. The specific aims of these studies are to study similarities and differences between SDS bone marrow, other bone marrow failure disorders, and RA bone marrows; to characterize all SDS patients with regard to presence or absence of AA or MDS; to classify SDS patients with MDS and to study MDS features in SDS; to also identify early markers of clonal evolution and to correlate MDS grade or early clonal markers with the development of acute leukemia; and to evaluate different MDS scoring systems regarding their predictive value for survival and development of acute leukemia in SDS patients. Dr. Elghetany will study patients with SDS and follow them up for several years. Their bone marrows will be studied for a variety of markers and will be compared with 40 patients with other inherited bone marrow diseases, 20 patients with refractory anemia (RA), 10 patients with acquired aplastic anemia (AA), and 10 with normal bone marrows.

These long-term goals require several years of follow up. This study will address and clarify the significance of the diagnosis of MDS in SDS. Dr. Elghetany's studies are not intended to take the place of the usual studies done by your doctor(s). For more information on how to participate and/or to obtain the needed forms, please contact Dr. Elghetany at 409-747-2468, e-mail melgheta@utmb.edu. Dr. Elghetany's research is an ongoing study and he is still accepting bone marrow samples.

RUBBER BRACELETS FOR FUNDRAISING AND COMMUNITY AWARENESS!!!!

I f you would like to help increase awareness of Shwachman-Diamond Syndrome in your community and at the same time assist in raising funds for medical research and family support, order some of our new Shwachman-Diamond red rubber bracelets! You can buy them in sets of 10 directly from our web site at www.shwachman-diamond.org and jsut click "Get Involved" and they will be mailed to your home. Please email us at info@shwachman-diamond.org to let us know that you have purchased any of these bracelets in honor or memory of anyone and we will publish the information in the newsletter. They are great to use as conversation starters to help family, friends, and medical professionals in your area understand about SDS and its impact on families. In addition to that, they are cool!!!!!!!!! You can give them to your friends and family or re-sell them to others. THANKS ALL!!!

SDSF DREAM BRACELETS FUNDRAISER

I am making Swarovski Crystal bracelets with Sterling Silver beads with SDSF charms as a fundraiser for SDSF. I have several colors to chose from and can customize for size. I have added a Premier Bracelet style along with the Standard Bracelet style. The Premier includes different shaped Swarovski Crystals, in clear only, with the colored stones. The shapes are cubes, cones, disc, and larger stones. They really add to the bracelet. Colors available are clear, black, dark blue, sapphire, light blue, pink, light amethyst, amethyst, tanzanite (lavender), light red, ruby, garnet, peridot (light green), emerald, white pearl, black pearl, pink pearl and birthstone colors. Include the size you wish.

The Standard bracelet is still $30.00 and the Premier is $33.00. There is still a $3.00 shipping charge per order to the same address, additional addresses will be an extra shipping charge. You can purchase these online at: www.shwachman-diamond.org through the donate button. Please email us at info@shwachman-diamond.org to let us know that you have purchased any of these bracelets in honor or memory of anyone and we will publish the information in the newsletter. Please allow 2-3 weeks for delivery.

The bracelets are beautiful and make great gifts. What a great way to support all SDS children. Thank you for your support in our dream to find a cure. Any questions, please contact me directly.

Jenny Jenuwine
810-395-2358
jengrls2@banyanol.com

E-MAIL SUPPORT GROUP

Would you enjoy e-mailing other Shwachman-Diamond families? Have you ever thought your child seems to have something you may not think is related to the syndrome? Why not sign up for our e-mail support group through Yahoo. It is a good way to stay in contact with other SDS families and also a great venue for asking questions you may have.

If you would like to subscribe to this support group, the link is: shwachmandiamond-subscribe@yahoogroups.com

If you would like to look at the guidelines for our e-mail support group, the link is: http://groups.yahoo.com/group/shwachmandiamond/?yguid=79215263

If you have any questions, please contact Julie Kroppe at jkroppe@wowway.com

WELCOME NEW FAMILIES

Each year many new families from all over the United States have children diagnosed with SDS. Some of these families may be in your area and we would like to welcome them into the Shwachman-Diamond Syndrome Foundation circle of support.

Berrysburg, PA
Newman, GA
Lyden, WA
Franklin, OH
Baline, MN
Covington, TN
Oxford, PH
Pocatello, ID
Churchville, PA
Phoenix, AZ
Denver, CO
Fort Worth, TX
Menifee, CA
Seattle, WA
Hydro, OK

REQUEST A BASKET FOR YOUR CHILD OR FAMILY MEMBER IF THEY ARE IN THE HOSPITAL

The Angel Anna Baskets are filled with gifts tailored specifically to each sick child's age and needs, and are sent out to the hospital or the child's home, upon learning of a lengthy hospitalization. Balloon bouquets are also sent out to those children who are temporarily in the hospital or who are going through a particularly rough time medically. It is our way to let these families and children know that we care and are thinking of them during their difficult time. I believe it is a wonderful addition to the family support that SDSF gives to each of our SDS families!

If you would like to request an Angel Anna Basket sent to a sick and/or hospitalized SDS child, or if you would like to make a tax deductible donation to our Angel Anna Basket Project (material or monetary donation), please call SDSF at the toll free number 1-877-737-4685 or contact me personally online at **psbishop1@yahoo.com** **or call me at (515)252-7445. I will be glad to answer any questions and I appreciate any and all input. Thank you to the many families who have contributed to this project!**

F.Y.I.

Axcan Scandipharm, the makers of Ultrase enzymes, ADEK vitamins, Scandishakes and many other products has included Shwachman-Diamond Syndrome in their patient support program. SDS patients who use their products may qualify for free and/or discounted products and information. Please note that Axcan Scandipharm patient support program has changed. Patients are no longer required to mail in receipts and forms in order to receive program benefits. The new card, AXCAN Rx COMPLETE card, will allow you to receive your program benefits more efficiently. To take advantage of this exciting new program card or to ask questions about it, please call the AXCAN Rx COMPLETE Program line toll-free, at 1-866-AXCAN-RX (1-866-292-2679), Monday-Friday, between 8:00 a.m. and 8:00 p.m., EST.

Thank You to our Donors

(donations January 12, 2009 - July 10, 2009)

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In Honor of Alex & Trevor Fisher
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In Honor of Logan Stone
Susan & Curt Utz
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In Memory of Jennifer Cromwell
Fran Cromwell

Established Shwachman-Diamond Groups

Shwachman-Diamond Syndrome Support - Australia
Contact: Joan Buchanan
61 03 5427 0645
email: buchanan.joan@gmail.com
http://www.shwachman-diamond.org

Shwachman-Diamond Support-UK
Contact: Sharon Clusker
Tel: 02476-345199 Fax:: 02476-345199
email: mail@sdsuk.org
http://www.sdsuk.org

Italy Association for Shwachman Syndrome
Contact: Aurelio Lococo
email: aiss@shwachman.it
http://www.shwachman.it

Shwachman-Diamond Syndrome Canada
Contact: Heather Norton
email: sdscanada@sympatico.ca
http://www.shwachman.org

Shwachman Syndrome - Netherlands
email: koster.e@hccnet.nl
http://www.shwachman.nl/

NEWSLETTER IDEAS

Do you have ideas for our newsletter? Do you have a question you would like to ask a doctor? Want to share your story?

Please send your stories and/or questions to SDSF to our address or e-mail them to: **info@shwachman-diamond.org**

We appreciate ALL input! We will print stories and answers in future newsletters.

Thank you.

REGIONAL PARENT CONTACTS

In a effort to help increase family support, these parents have volunteered to help with questions and concerns:

IN THE USA

Corky DeBoer - IL: (708)532-4954 or opcrccdb@aol.com

Jenny Jenuwine - MI: (810)395-2358 or jengrls2@banyanol.com

Kelly Bright -TX: (409)738-2925

Donna Garfield -VA (703) 731-7889 or dkgarfield@verizon.net

Michelle Noble - CA: (760)947-4283 or MNoble2day@aol.com

Cyndi Smith - SC: (803) 781-7100 or Chs5099@aol.com

OTHER COUNTRIES

Sharon Clusker - England:
Sharwk60@btinternet.com

Lee-Anne Hayes - Australia
61 02 49608428 or hathor@bigpond.net.au

Reinald Baumhauer - Germany
Fax: 049-89-41902871 or
r.baumhauer@mnet-mail.de

Aurelio Lococo - Italy
Tel. e Fax: +049 8736130 or
aiss@shwachman.it

CHANGE OF ADDRESS OR E-MAIL

Please forward your change of address or e-mail to continue receiving your newsletters.

If your newsletter is sent by regular mail, the post office will not forward it to you due to "Bulk Rate" postage being used.

Either call us at **1-888-825-SDSF (7373)** or email us at **info@shwachman-diamond.org** with your changes.

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127 Western Avenue, Sherborn, MA 01770 U.S.A.

Credit Card donations can be made through our website - www.shwachman-diamond.org

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The children and adults you are helping THANK YOU for caring.
Your generosity in giving is greatly appreciated.

Shwachman-Diamond Syndrome Foundation is a tax exempt organization as described under the Internal Revenue Code, Section 501 (c)(3). Our Tax ID number is 43-1709945.

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