



Cure For A Future

Shwachman-Diamond Syndrome Foundation

NEWSLETTER

Fall 2008

Another fabulous family conference was held at Camp Sunshine, <http://www.campsunshine.org>, in Casco, Maine from July 20 - 25!! Although the weather wasn't great it didn't stop the fun and it was informative and supportive for both the kids and the parents. The 60 or so volunteers and the fantastic staff were an integral part of making the 30 SDS families feel very welcomed and a part of every activity and discussion group.

We started off with a "meet and greet" of familiar and new faces on Sunday night followed by a children's entertainer and family bonfire. On Monday and Tuesday during the days, six doctors, many from our Medical and Scientific Advisory Board and some that aren't, were invited and paid for by SDSF to give presentations of their latest research and information. The doctors were: Dr. Richard Harris, Director of the Bone Marrow Failure Clinic, Cincinnati Children's Hospital Medical Center; Dr. Peter Durie, Director of Cystic Fibrosis Research, Gastroenterologist and Professor at the University of Toronto, Hospital For Sick Kids, Toronto. ON; Lynda Ellis, R.N., Clinical Research Nurse Coordinator - Gastroenterology, Hepatology and Nutrition at the Hospital for Sick Children; Dr. Johanna Rommens, founder of the gene for SDS, professor, scientist, University of Toronto; Dr. Akiko Shimamura, MD, PhD, Fred Hutchinson Cancer Center, Seattle Children's Hospital; Dr. Elizabeth Kerr, Psychologist

at the Hospital for Sick Children, Toronto, Ontario; and Nancy Cincotta, L.C.S.W. from Camp Sunshine. They all helped open wonderful discussions between themselves and the parents during the morning and afternoon sessions. There were afternoon break out sessions where we broke up into small groups and had more intimate discussions with the doctors about their specialties. Here are the topics that were covered during these two days of sessions: Clinical Diagnosis of SDS, Gastroenterology, Nutrition and Growth, Hematology Basics, Skeletal and Dental Features, Behavioral, Social and Learning Features, Hematology & Pathology Reports, what do they say?, Genetics & Genetic counseling, Management of Hematological Issues, Bone Marrow Transplant Strategies, Review of Recent Medical & Scientific Findings, SDS Registry. Although I could go into detail for pages and pages, I hope that you have gained an inkling of what was presented during the family conference. Everything we heard was incredible and it was fantastic that these doctors took the time out of their busy summer schedules to share their knowledge with us.

While the parents were busy during those two days, the older kids were busy with "camp". Camp included archery, swimming, mini golf, waterfront activities (canoeing, kayaking, paddleboats and swimming), climbing the rock wall, volleyball, arts and crafts,

continued on page 2



Cure For A Future

Shwachman-Diamond
Syndrome Foundation

127 Western Avenue
Sherborn, MA 01770 U.S.A.
1-877-737-4685 (Toll Free)
E-mail: 4sskids@shwachman-diamond.org
Website: www.shwachman-diamond.org

continued from page 1

playing on the playground, computer time, and the challenge course. The toddlers and nursery age kids were lovingly taken care of by the volunteers in their own play spaces which provided for their every need.

At night there were special activities to reconnect the families to each other after a long day as well as to other SDS families. There was a masquerade dance with a DJ, carnival night, talent show and a celebration ceremony put on by the kids and their counselors. There was also free time for the parents to participate in games organized for them or they could just relax in their individual suites that provided perfect accommodations for up to 6. We also had a parents-only gathering for sharing ideas, observations and stories. There was even a dads-only meeting!! Both of these were a success and will be offered at the next Family camp!

All of the meals were taken together in the dining hall which provided a much needed relief from cooking for 5 days. It was also a nice feeling for the kids who take enzymes to know that everyone else in that dining room either took enzymes or had a family member who took them and thus there was a feeling of confidence and unity for them. There was even an adult-only dinner served by candlelight with karaoke music for entertainment that gave parents a little respite and time to themselves.

Camp Sunshine thought of it all. They have found a way to give our families a vacation from the daily stresses of cooking, cleaning, and worrying about doctors' appointments, etc. The Camp Sunshine staff supplied it all, they even had an expert medical staff available 24/7 to handle any medical situation that happened to arise. Camp Sunshine staff enabled families to have time with their loved ones as well as new friends and doctors in a fun, supportive, loving, connecting environment.

Check out some pictures from the family conference inside this newsletter!!!

NEW WEB SITE

<http://www.shwachman-diamond.org/>

We have been working hard to revamp our web site and we hope that you will be pleased with the look, design, organization and content. The site hasn't gone live yet due to some hosting transfer issues so please check periodically for it's launch! SDSF is still focusing on funding research towards better medical management and a cure and we are still offering the many family support services that we have offered in the past. We are just trying to make it a little easier to find everything.

Let us know what you think about it. We are still looking for a web manager with knowledge of HTML language that can offer their services as a donation to the cause, so at this time it isn't easy to make the changes. However, if you find something you think needs adjusting we will take into account any comments we receive and work hard to adjust the site so that it says all that we want it to say.

WELCOME DR. BESSLER

Shwachman-Diamond Syndrome Foundation Board of Directors is pleased to announce the addition of Monica Bessler, M.D., PhD, Co-Director, Division of Hematology, Washington University School of Medicine, St. Louis, MO to our Medical Advisory Board.

Dr. Bessler will help SDSF in its effort to represent and support the entire SDS community by focusing on the unique hematological needs of the adult SDS community.

CHECK OUT THESE WEBSITES

Healthfinder.gov

Coordinated by the Office of Disease Prevention and Health Promotion (ODPHP) and its National Health Information Center, the redesign of healthfinder.gov was informed by health literacy and usability principles and tested with more than 650 users. This is a consumer health Web site that's easy to understand and navigate, especially for people who have limited health literacy.

You can visit healthfinder.gov's **Quick Guide to Healthy Living**, <http://www.healthfinder.gov/prevention/> a new resource that uses everyday language and examples to:

- Tell users how to take action to improve their health using a "small-steps" approach
- Give users positive reasons to change their behavior
- Provide tools and encouragement, such as personal health calculators, menu planners and recipes, tips for caregivers, and printable lists of questions to take to the doctor

There is the new "myhealthfinder" tool, which provides personalized health recommendations based on sex, age, and pregnancy status. This feature offers evidence-based recommendations from the U.S. Preventive Services Task Force, an independent panel of experts in prevention and primary care sponsored by the Agency for Healthcare Research and Quality.

For instructions on how to link to healthfinder.gov, please visit

<http://www.healthfinder.gov/aboutus/linking.aspx>.

Survivorshipatoz.org

Supported by grants from Johns Hopkins Medicine and the City of New York, this website is a resource for all those living with a serious illness. The mission of the website is to provide practical information to all people living in that "new normal" that exists after a life-changing diagnosis.

With practical health information, personalized Individual Action Plans, and a customizable Symptom Diary, the website It also has Health Plan Evaluators to help choose the health insurance plan right for you as well as Interactive charts to help manage your healthcare bills and financial situation.

Please visit the website for more information:

<http://www.survivorshipatoz.org/>

CONGRATULATIONS ARE IN ORDER:

DR. SHIMAMURA

Congratulations are in order for Dr. Akiko Shimamura, a member of SDSF Medical Advisory Board, because she was named a "Top Doctor" by Seattle Metropolitan Magazine for 2008. Very impressive for a doctor who only arrived in Seattle in 2006! We have known for a long time that she is a special, wonderful doctor and we are so glad that Seattle knows now too. Dr. Shimamura not only is doing research on SDS but is taking control of the management of the upcoming Registry of SDS data as well as seeing patients of SDS. We are grateful for her commitment to SDS and we offer thanks and congrats to her!!

NATIONAL ORGANIZATION FOR RARE DISORDERS (NORD) UPDATES

Social Security Administration Seeks Input from Physicians on Rare Diseases:

The Social Security Administration (SSA) conducted hearings last December as part of an effort to better understand rare diseases so that patients will encounter fewer delays and frustrations when they apply for Social Security disability assistance. Currently, individuals with rare diseases often receive initial denials that later are reversed. The result is unnecessary expense and delay. To address this problem, SSA is asking physicians who treat patients with rare diseases to help establish criteria for these diseases. Three questions are posted on the NORD Web site (www.rarediseases.org). Physicians may submit their responses to ssa@rarediseases.org.

Lifetime Insurance Caps Bills Introduced in Congress:

Representative Anna Eshoo (D-CA) and Senator Byron Dorgan (D-ND) have introduced legislation in the House and Senate to address the aggregate spending limits placed on many insurance policies. This issue is of particular interest to those with rare diseases who often require high-cost orphan drug therapies and chronic care.

Read NORD's Advocacy Alert on this topic:

<http://www.rarediseases.org/nord/washington/office>

COALITION TO END THE TWO-YEAR WAITING MEDICARE PERIOD FOR PEOPLE WITH DISABILITIES

Recently the Shwachman-Diamond Syndrome Foundation received and accepted an invitation from NORD to join the Coalition to End the Two-Year Waiting Medicare Period for People with Disabilities and help bring an end to unjustifiable delay in health coverage for people with disabilities. According to NORD, nearly 7 million people under age 65 in the United States qualify for Medicare due to severe and permanent disabilities. However, these individuals must wait two years from the date they are deemed eligible for Social Security Disability Insurance to receive Medicare coverage. Tighter Medicaid eligibility requirements and the reduced availability and higher cost of employer-based coverage mean that many people with disabilities are uninsured or underinsured during the two years they wait for Medicare coverage. NORD is hopeful that forming a coalition dedicated to ending the Medicare two-year waiting period, they can coordinate their grassroots advocacy and media outreach and build support for legislation that will end the waiting period.

UNDERSTANDING

CBC RESULTS

Ever have trouble deciphering all those number on the CBC results from your bloodwork? The NIH has produced a publication entitled “Understanding Your Complete Blood Count” that just might help! The publication is available on-line at:

http://clinicalcenter.nih.gov/ccc/patient_education/pepubs/cbc97.pdf

This publication not only explains the different parts of the CBC and their normal values it also includes important information such as medications that can affect your bodies ability to produce platelets as well as red and white cells.

The publication is in “pdf” format so you will need to have an Adobe Acrobat reader installed in order to open it.

Calculating your ANC

What if your lab sheet doesn't have the ANC listed? Here are some tips on determining the ANC for yourself. Calculating ANC Example from Cincinnati Children's Hospital Medical Center.

The following example will help calculate the ANC when lab results return WBC in complete count, rather than in decimal form.. For this example, a Patient's WBC is 4,000 and he has 20 Neut and 10 Bands. To find out what the ANC is, add the Neut plus bands and write this number as a decimal 20% +10% 30% or .30 Multiply the WBC number by the sum of the(WBC) (%Neut + %Bands) = ANC
 $(4,000) (.20 + .10) = ANC$
 $(4,000) (.30) = 1,200$

Still not sure that you've calculated it right? Try one of the online calculators, just plug in the numbers from the lab sheet, and click the CALCULATE button and viola the ANC is figured out for you!

<http://www.mylan-clozapine.com/ANCCalc.asp>

http://www.curehodgkins.com/hodgkins_resources/anc_calculator.html

http://www.fazaclo.com/ANCCalculator_v3.htm

<http://www.globalrph.com/anc.htm>



FAMILY SHARING PAGE



FAMILY RAISES MONEY FOR DOCTOR EDUCATION

In April 2008 we learned that our 3 year old daughter, Jordyn, was diagnosed with Shwachman-Diamond Syndrome (SDS). Like many SDS families our journey to diagnosis was long as we found the knowledge among the medical community was lacking. Dealing with the diagnosis of SDS has been more than difficult. I was in need of a way to channel my emotions effectively; and so my mission began...

Our story spans nearly three years and begins like so many of your own. We knew there was a problem when I was 17 weeks pregnant, and at that time it was determined there was Intrauterine Growth Restriction due to a failing placenta. After countless ultrasounds and fetal non-stress tests twice weekly, I gave birth at 38 ° weeks to the most beautiful five pound baby girl. Many unexplained symptoms started early on, and Jordyn was tested for everything from Downs Syndrome to Cystic Fibrosis. We saw Gastroenterology for her pancreatic insufficiency and Infectious Disease for her immune deficiency. Each symptom continued to be treated separately.

I was a frustrated mother determined to find the

answers, and with the help of Google, that's exactly what I did. When her symptoms were "Googled" together, Shwachman-Diamond Syndrome was the first thing I read. I quickly printed what I found; comparing it to Jordyn's medical record and making my own notes. I took that information and my notes promptly to her Infectious Disease specialist, Michael Ryan DO, a physician with 33 years of pediatric experience. We sat and reviewed my notes with Jordyn's chart and finally my concerns were validated. For the first time in Jordyn's life I felt like we were moving in the right direction. Shortly after a referral to The Children's Hospital of Philadelphia, Jordyn had an SDS diagnosis.

Our local healthcare facility, Geisinger Health System, is one of the nation's leading fully integrated health services organizations. Founded in 1915, this organization serves more than two million residents throughout central and northeastern Pennsylvania. And though it employs more than 100 pediatric physicians and specialists, through no fault of their own, no one has enough knowledge on Shwachman-Diamond Syndrome to make a diagnosis. This is a large facility and the physicians are eager to learn.

As a mother of child with a devastating disease the lack of knowledge is unacceptable. Too many families travel way too far to receive appropriate care for their SDS child. It wasn't until July, when I was homebound with a bout of chicken pox, that my ideas started to come together and my goal was determined...to raise enough money to send two Geisinger pediatric specialists to the 5th International Congress on Shwachman-Diamond Syndrome scheduled to meet in June 2009 in Amsterdam. Along with the help and support of family and friends, as well as Dr. Ryan, Chairman of Pediatrics at Geisinger's Janet Weis Children's Hospital, I am now planning numerous fund raising events in the upcoming months including a 5K Run/Walk and a Benefit Dinner. Geisinger has even set up a fund specifically for the education of Shwachman-Diamond Syndrome. The support and generosity from our community thus far has been heartwarming.

continued from page 6

Education in itself is invaluable. SDS education is invaluable to these children and their families. The benefits to the community are endless; from reaching diagnosis early on to receiving care close to home. Too little is known. I plan to spread the word; to encourage education for the doctors at Geisinger with the hope of sparking an interest. Because with interest comes research and with research I am confident a cure will be found.

By Kim Zajac, Pennsylvania

JOIN OUR “CLUB” FOR ADULT SDS PATIENTS

SDSF is becoming increasingly 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 thoughts and ideas to SDSF by either calling 1-877-737-4685 or emailing us at 4sskids@shwachman-diamond.org.



CALL FOR DONATIONS!

Ever wonder how you could “give back” to SDSF? Consider donating Angel Anna gifts or office supplies. Here are some ideas that are quick and easy but will bring a huge smile!

Angel Anna Baskets

Art Supplies (markers, crayons, paints, stamps, stickers, glue sticks, colored pencils)

Coloring, Activity & Puzzle books

Play Doh & Craft kits

Stuffed Animals (new)

Children’s movies

Books (for all ages)

Music & DVDs (for all ages)

Matchbox Cars

Blankets (knitted, quilted, tied for all ages)

Dolls

Hair Accessories (barrettes, bows)

Journal (for writing and drawing)

Pre-paid phone cards

Games (all ages)

Gift Cards (Walmart, Target, K-Mart)

Remember, the next time you are buying those goody bags or “keep ‘em quiet” favors for your child’s party or road trip, consider picking up a few extra and donating them to SDSF!

Angel Anna Basket donations should be sent to:

Staci Bishop, SDSF Angel Anna Basket Coordinator

6816 Mill Pond Drive, Urbandale, IA 50322

Administrative Supplies

Heading out to Staples or Office Max for supplies for your home computer/office? Consider picking up a gift card for use by SDSF administrative staff.

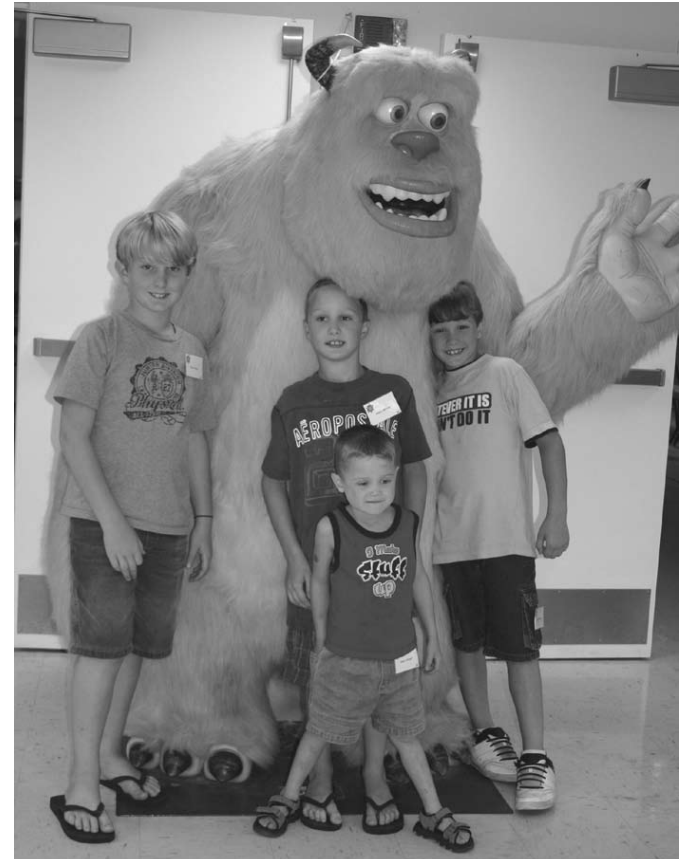
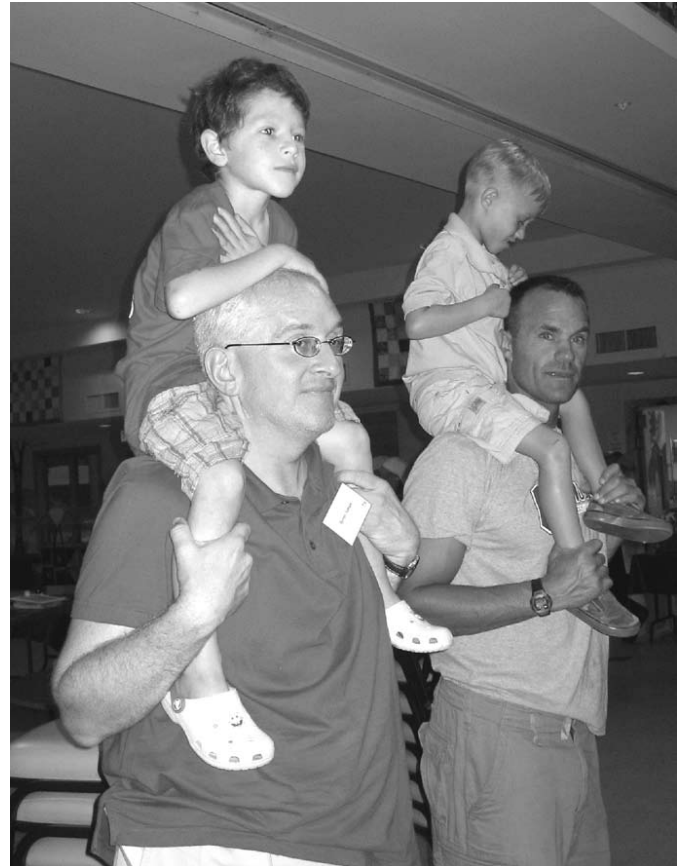
Office Supply donations should be sent to:

Kim McDowell, SDSF Administrative Assistant

2334 Rolling Ridge Drive, Avon, NY 14414

Rest assured that SDSF is an accredited and audited 501(c)3 organization. You will receive a letter describing your donation with appropriate amount listed for tax deduction purposes.

YOUR DONATIONS PLAY A CRUCIAL PART IN SUPPORT OF THE SDSF COMMUNITY.



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
Division of Hematology
Washington University School of Medicine;
Box 8125; 660 S. Euclid Ave.,
St. Louis, MO 63110, USA
314-362-8807
Mbessler@im.wustl.edu

David Wilson, M.D., Ph.D. Co-Director
Division of Pediatric Hematology/Oncology
Washington University School of Medicine;
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



If 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 for \$25.00. Just contact us at 4sskids@shwachman-diamond.org to order them and they will be mailed to your home. 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 choose 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 order your bracelet in honor of your child and a special card will be sent. It will also appear in the newsletter. An order form can be found on the web site for your convenience, www.shwachman-diamond.org. Sorry no COD's or credit cards. Please allow 2-3 weeks for delivery. Make your check payable to SDSF and mail to 127 Western Avenue, Sherborn, MA 01770.

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
jengrsl2@banyanol.com

Thank You Bracelet Purchasers

(purchases May 1, 2008 -
October 10, 2008)

In Honor of Logan Stone

Curt & Susan Utz
Anchor Club of Jeffersonville High School

In Honor of Gracie Van Brunt

Gracie Fund

In Honor of Bailee Thacker

Sarah Dolby

In Honor of Tyler Knoble

Vicki Brown

In Honor of Brooke Lindgren

Shirley Wright

In Honor of Fallon Veicht

In Honor of Hannah Norton

In Honor of Collin Brown



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.

Brookline, MA

Howell, NJ

Philadelphia, PA

Easley, SC

Pocono Summit, PA

Arlington, TX

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 May 1, 2008 - October 10, 2008)

Susan Kolenda
Judith Ann Sherman
United Way of Mesa Cty - Chris Kadel
Don & Dianna McNew
Ali Elmi
Solvay Finance, Inc.
Rose & Frank Canonico
Vickie & Christopher Cox
Carrie Andrews
United Way of Central Iowa

Eastern Niagara United Way

Damise Annable
James Cantella
Jeffrey Costello
Judith DeMaillie
Robert Gilsinan
Virginia Huges
Sharon Lamb
Joan Morgan
Wendy Pellow
Robert Roth
Terence Upton

In Honor of Gracie Van Brunt

Gracie Fund

In Honor of Jayden Wegford

Sunshine Society

In Honor of Erin Johnson

Erin Johnson
Glady Sifter

In Honor of Collin Brown

Jim Brown & Peggy Holt
Richmond Rotary Club

In Honor of Emily & Kelsey Jenuwine

Diana & Dave Murphy

In Memory of Melanie Huh Smailus In Memory of Ralph Ellsworth Kuhn

Mr. & Mrs. Roberts
Richard Hiler
Mildred Crawford
D.L. Reichard
Erin Johnson
Alma & J. Herbert Frantz
The Hershey's Company Information Services and
Operations and Infrastructure Services
Mr. & Mrs. James Coleman
Charles Good
Sharon Finafrock
Ronald & Anne Zappe
Harry & Dorothy Kuhn
Vivian DiCristofaro
Charlene Good
Joan Mowery
Margaret & Charles Smith

In Memory of Alexis Layton McFarland

Donna Solomon

In Memory of Beatrice Ann Low

Ralph Toussie
Marian & Roger Gray
Miriam Blankenship
Kevin Wright

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: 024-76345199 Fax:: 024-76345199
email: sharwk60@btinternet.com
<http://www.shwachman-diamondsupport.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/>

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 opcrcddb@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@garfield-home.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

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: 4sskids@shwachman-diamond.org

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

Thank you.

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 **1-877-737-4685** or e-mail us at 4sskids@shwachman-diamond.org with your changes.

Medical Scientific Advisory Board

Blanche Alter, MD, MPH

National Cancer Institute
Bethesda, MD

Marco Cipolli, MD

Gastroenterologist
Cystic Fibrosis Center
Verona, Italy

Peter Durie, MD

The Hospital for Sick Children
Toronto, Canada

Tarek Elghetany, MD

University Texas Medical Branch
Galveston, TX

Lynda Ellis, RN

GI/Nutrition Dept. Hospital for Sick
Children, Toronto, Canada

Monica Bessler, MD, PhD

Co-Director, Division of Hematology
Washington University School of Medicine
St. Louis, MO

Michael Glogauer, DDS, PhD

University of Toronto
Canada

Johanna Rommens, PhD

The Hospital for Sick Children
Toronto, Canada

Akiko Shimamura, MD, PhD

University of Washington
Seattle, WA

Steve Werlin, MD

Children's Hospital of Wisconsin
Milwaukee

Alan Warren, MD

Professor of Hematology
MRC Laboratory of Molecular
Biology
Cambridge, United Kingdom

SDSF

Founder

Joan Mowery

Board of Directors

Blair Van Brunt - **President**
Matthew Palmer - **Secretary**
Bryan Sample - **Treasurer**
Susan Utz
Theresa Henle

Donna Garfield
Newsletter Editor

Staci Bishop
Anna Angel Basket Coordinator

Kim McDowell
Administrative Assistant

Ann Bodewas Stephens
Herzog, Crebs, St. Louis, MO
Attorney

WE NEED YOUR HELP PLEASE!!!!

Please send you tax deductible gift to: **Shwachman-Diamond Syndrome Foundation**
127 Western Avenue, Sherborn, MA 01770 U.S.A.

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

NAME: _____

BILLING ADDRESS: _____

CITY _____ STATE: _____ ZIP: _____

TELEPHONE: _____

In Honor or Memory of: _____

**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.



Cure For A Future

127 Western Avenue
Sherborn, MA 01770
1-877-SDS-INTL

ADDRESS SERVICE REQUESTED

PRSRT STD
U.S. POSTAGE

PAID
ROCHESTER, NY
PERMIT NO. 546

Copyright Shwachman - Diamond Syndrome Intl., 2001

Disclaimer: The views expressed in this newsletter do not necessarily represent the views of the Board of Directors, Professional Advisory Board or members of the organization. We do not promote or recommend any particular treatment, etc. The relevance of any medical information in this newsletter should be discussed with your physician.