



**Shwachman-Diamond  
Syndrome Foundation**

“Help Us Grow”

## NEWSLETTER

### Summer 2005

Happy Summer! I hope this issue finds you well and enjoying the warmth and beauty of summer.

I would like to begin by thanking the UK support group and physicians from their MSAB (Medical and Scientific Advisory Board) for hosting the Third International Scientific Congress on Shwachman-Diamond Syndrome. I can imagine the amount of time and energy that was put into this very important and successful meeting. The main point I would like to convey to the families is that we have some of the best, if not the best, researchers, physicians, and scientists researching towards a cure and better treatments for this disease! I was absolutely floored by the amount of information that was presented. It has only been two years since our last scientific meeting was held in Toronto and so much more has been learned. **I can't go on any further without acknowledging and thanking Johanna Rommens and everyone at the lab at The Hospital For Sick Kids.** Without the gene, I know a lot of what has happened and will happen from here hinged on that discovery. Please stay tuned to the next issue of our SDSF newsletter for a full recap of this meeting from Dr. Alan Warren. I would like to announce that the next scientific meeting is going to be held in the United States during the summer of 2007.

We will also be having our family meeting in Denver, Colorado in July of 2006. More details

will be available soon through this newsletter and our website at [www.shwachman-diamond.org](http://www.shwachman-diamond.org).

These two meetings are going to mean a lot of work for the Board of SDSF. Not only will there be a great deal of planning involved, it is also going to be a costly endeavor. With funding research as one of our top priorities (we spent \$60,000 on research last year alone), we will need to secure funds above and beyond what we set aside for research to meet these goals. Our Board is working on soliciting grants through various venues. As parents and family members of children with this disease, we all need to work together to make these goals a reality. Please think about how you can fundraise, get involved and help!

Finally, I would like to thank everyone for sending in their response sheets for how they would like to receive the newsletter. I would like to stress that we don't mind sending the newsletter snail mail AT ALL. We just thought we could save some trees and money if we offered an alternative delivery. Please don't feel hesitant to state that you like to have the hard copy delivered to your mailbox! We believe in sharing information and our main goal is to support those affected with this disease and this is just one of the ways we can do that!

My best to you and your families for a happy, healthy summer!

Debbie Kadel



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# **DIGESTIVE DISEASE WEEK MEDICAL CONFERENCE A SUCCESS FOR SDSF**

The three members of the Board of SDSF attended the Digestive Disease Week medical conference in May 2005. This conference is geared towards providing scientific and educational information in the fields of gastroenterology, hepatology, endoscopy, and gastrointestinal surgery and included 5,000 oral presentations, poster sessions, and booths that were set up by many companies looking to sell their products. Doctors, clinicians, nurses, researchers, and publishing houses made up the variety of people who attended and it was very enlightening for us to learn more about this world and how we can tap into it for the benefit of our Shwachman-Diamond Syndrome patients and families.

We already owned a booth from previous conferences so we spent a morning setting it up and then manning it for three full days. It was quite an experience for us Board members who were there. Approximately 16,000 people attended this conference and we were one of 294 booths. Our booth was located in the back aisle near other non-profit organizations but we managed to see a fair amount of traffic anyway.

Our goal going into this conference was to advertise the need for adult GI's to learn more about Shwachman-Diamond Syndrome because SDS is perceived as a pediatric disease and we know that these kids grow up and need adult GI's. We managed to talk to quite a few and were able to give them the most up-to-date information and medical research articles. There were many doctors from other countries who sought us out because they have SDS patients and this was most gratifying to be able to meet them face to face and help them with issues or give them referrals as needed. But most of all, we feel that the bulk of the success came from having made contact with a few companies who make products in this digestive field who are interested in helping us out financially for our family conference next year and a scientific conference

to be held in the US the following year. We will be following up on this as the year progresses. Although it does cost money to attend these conferences, we feel that this was a successful expenditure of our time and all of our dollars.

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## **SAVE THE DATE FOR THE NEXT FAMILY CONFERENCE**

### **JULY 2006**

We are planning our next family conference for the end of July, 2006 in Denver, Colorado. We still have many details to work out between now and then, but wanted to let you know in advance for planning purposes.

We will be putting together a slide show for the conference and want to include as many families as possible. Please go through your pictures of your SDS child/children with their pets, families, friends, etc. Send in your favorite 5 to: Ms. Sharon Lamb, 7339 Lake Road, Appleton, NY 14008. If you would like these pictures back, please put your name on the back of each picture and include a self addressed, stamped envelope.

Thank you for your participation in helping to make our family conference a great one. Every little bit counts!

## ARE YOU OUR NEXT NEWSLETTER COORDINATOR?

We need your help! We are looking for someone to help coordinate articles and proofreading the newsletter 3-4 times per year. You will be working with the board members, doctors, families, and any contributors via email. Our assistant to the board already manages this task but needs help with the production and layout.

The only requirements for this very important job is, an email account and knowledge with Adobe Acrobat 7 (easily downloadable for free from the Internet).

Email us at [4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org) with any questions and interest. Please consider volunteering for this job. **We send this newsletter to families and doctors all around the world who count on the information they receive from us.**

Thank you for thinking about it!!!

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## FAMILY VOICES WEB SITE HELPFUL INFORMATION FROM STATE TO STATE

I have recently come across this web site that many of you may know already but wanted to share with those of you who don't know it. We, as a board, have not done any work with this organization but thought that we would just share the information in case some of you were interested.

“Family Voices is a national, grassroots clearinghouse for information and education concerning the health care of children with special health needs. We stay on top of public and private sector health care changes that affect our children and families through the collective efforts of our families: a volunteer Coordinator in every state; 10 Regional

Coordinators; and a small staff working in several locations around the country. Together, we share the expertise and experiences of families from around the country with state and national policymakers, the media, health professionals, and other families. We work in public and private hospitals, public health programs, in state capitals, in Washington, DC, serving on boards and task forces, working in partnership with health professionals and policymakers, bringing the family perspective to policy discussions and decisions. There are almost 40,000 Family Voices members - families of children with special health needs and friends and professionals who know and love our children.”

To learn more about specific national projects please visit Projects. To learn more about the work of state Family Voices organizations, please visit Family Voices in Your State.

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Family Voices, Inc.  
2340 Alamo SE, Suite 102  
Albuquerque, NM 87106  
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Fax: (505)872-4780  
Email: [kidshealth@familyvoices.org](mailto:kidshealth@familyvoices.org)

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**THE SDSF BOARD  
WOULD LIKE TO WISH  
YOU AND YOUR FAMILY  
A SAFE AND HAPPY  
SCHOOL YEAR**

## **AQUA-E NEW PRODUCT AVAILABLE FOR SDS PATIENTS**

Aqua-E is a new product formulated for absorption by adults and children with conditions associated with malabsorption of lipids due to liver or pancreatic insufficiency or inflammation or infection of the gut. Aqua-E was recently introduced by Yasoo Health to overcome fat-soluble vitamin malabsorption.

Aqua-E is a unique, clinically-tested formulation of liquid water-soluble complete vitamin E that can be absorbed even by children with impaired digestion or impaired liver function. Aqua-E contains the entire vitamin E family of alpha-, beta-, gamma-, and delta-tocopherol and alpha-, beta-, gamma-, and delta-tocotrienol, like the vitamin E from food sources, while its competitor, Liqui-E contains only alpha-tocopherol. Aqua-E is particularly rich in natural gamma-tocopherol and the tocotrienols. These other members of the vitamin E family have different properties than alpha-tocopherol and are depleted when using alpha-tocopherol supplementation alone. Gamma-tocopherol, for instance, is an important scavenger of nitrogen free radicals and shows anti-inflammatory properties.

Aqua-E is odorless and has neutral taste. It is very light and with no aftertaste, and it mixes easily with most beverages and water. Aqua-E is available in 4 oz. and 8 oz. sizes, and dosing may be easily adjusted to meet specific needs.

Aqua-E is sold in pharmacies, health food stores, and by mail order distributors. Patients can also order directly from us by telephone, fax, or our web site at [www.yasoo.com](http://www.yasoo.com). Any pharmacy or health food store can also order directly from us at wholesale prices. We also offer a "ME Club" for people who must supplement with large dosages: with a script or physician's note we can set them up on a buy 3, get 1 free plan.

*Submitted by Barbara Schreiber, Yasoo Health, Inc.*

## **DREAM BRACELETS**

I am making Swarovski Crystal bracelets with Sterling Silver beads, and SDS charms on them as a fundraiser for SDSF. I have several colors to choose from and can customize for size. The colors available are clear, black, dark blue, light blue, sapphire, pink, light amethyst, amethyst, tanzanite (lavender), light red, medium red, garnet, peridot (light green), emerald, and birthstone colors. These bracelets are beautiful and make great gifts. You can order your bracelet in honor of your child and a special card will be sent with the bracelet as well as being listed in our newsletter. What a great way to support all SDS children and their families.

To order just send a check or money order made payable to Jenny Jenuwine for \$33.00 (shipping and handling included) with the color and size of the bracelet. Please allow 2-3 weeks for delivery. Sorry NO COD's or credit cards accepted. All proceeds go to SDSF to help our dream.

Thank you for your continued support!! To date approximately \$4,000.00 has been raised. You can view a sample bracelet on our website. If there are any questions, please contact me directly.

Jenny Jenuwine  
15028 Hough, Allenton, MI 48002, 810-395-

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## **LAUGH FOR THE DAY**

The CEO of a large HMO dies and goes to heaven. St. Peter shows him to a lovely villa, wonderful music, great views, full staff of servants, gourmet meals, etc.

The CEO says, "This is terrific!!"

St. Peter says, "Don't get too comfortable, you're only approved for a three-day stay."

# FAMILY SHARING PAGE

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## THE GOFF FAMILY STORY

*Life is the heart of it, waste any part and the world passes you by, Don't ask for how long or why, just celebrate life.....*

I am grateful for my three SDS children who live everyday celebrating life. Recently, the State of Idaho offered an art contest to all elementary students in South Eastern Idaho in conjunction with the Eagle Rock Arts Museum. There were 11,000 entries including two entries from my sons, Spencer (10 years) and Samuel (7 years). They spent many hours deciding on their medium and subject matter and many more hours perfecting their art. Finally the deadline for all entries arrived and they proudly carried their finished work to school where it was transported to the museum to be judged. To our great surprise, both boys were selected as winners! Only 95 students out of the 11,000 were chosen to have their art displayed in the beautiful museum. A special open house was held in honor of the student artists and the boys were selected to be the honorary children photographed for the brochure and advertising the museum and further contests. They were very proud of their work and felt very special to be a part of something so great! We have since learned that their art pieces have been chosen, from the 95 winners to be a part of a traveling art show, showcasing children's artistic talents. The show will be going to major museums around the United States, Canada, Europe, and Asia.

Shelbie began private voice lessons two months ago, a dream she has always had but was afraid to admit until one day she confided in me. It only took me an entire day and alot of phone calls and begging to find a teacher who would take a vocal student so young with so many breathing issues, but by the next day she began her first lesson with a wonderful teacher.

She had to try out before the teacher would make any commitments and was told she had to sing a foreign song and a folk song. She began with a Latin song we practiced all night and before Shelbie had completed singing the second verse, the teacher was in tears. Her voice is so pure and melodic. The two bonded right away. Before the first month was over, Shelbie had to sing in her first serious vocal festival where she was judged by one of the toughest voice professors from the University. She had to sing two solos, one a folk song and the other a Broadway song and both pieces had to be memorized. She studied hard, and received the highest score of any of her teacher's other 45 students who were much older and had been singing for two years or more. She got 99%. The judge fell in love with her voice. She has since sung in recitals, and many other functions and continues to excel despite her breathing difficulties.

There are many things my children are not able to do like normal children but they don't see that as a detriment. They don't worry about a bubble that has just burst, they set out to blow another one! It's not important that they make me proud, it's more important that they are proud of themselves, and they truly are! We are looking forward to another concert featuring Shelbie's breathtaking voice and the boys have been chosen to feature their tumbling and skills on the rings in an upcoming Gymnastics Hallmark. You can not steer a river, you can only ride it through.

Sincerely, Kathy Goff, mom to Shelbie, Spencer, Samuel - all SDS  
Expect...Invite...Receive...Enjoy

# RESEARCH

## Dr. Johnson Liu's Bio

Dr. Johnson Liu received his medical degree from the University of Michigan Medical School. After residency training in internal medicine, he joined the National Heart, Lung, and Blood Institute in 1988, where he remained for 14 years in the Hematology Branch, before leaving to New York in 2002.

His research interests have focused on elucidation of the molecular biology of the congenital bone marrow failure syndromes, including Fanconi Anemia (FA) and Shwachman-Diamond Syndrome (SDS). These disorders share common pathophysiologic features with each other and may transform to acute myeloid leukemia. Thus, while rare, these diseases serve as genetic models of hematopoietic failure and may reveal insights into fundamental stem cell biology. Importantly, all these disorders can have a devastating impact on both patients and their families.

Dr. Liu has authored or co-authored 86 papers, reviews, monographs, and book chapters in the scientific and medical literature, of which 40 contributions are devoted to Fanconi Anemia. His work has focused on understanding the hematopoietic defect in FA, through the use of "knock-out" and transgenic mice, as well as by molecular methods such as protein-protein interaction screening. He was also responsible for the development of the first clinical protocol aimed at FA gene therapy, guiding this project from the initial bench experiments in 1994, through the manufacturing and regulatory process to completion in 2000.

Dr. Liu has recently joined the Schneider Children's Hospital in New Hyde Park, NY. His current research interests have continued to focus on developing novel therapies for congenital bone marrow failure syndromes and acute myeloid leukemia. One aspect of this work centers on elucidating the function of the newly discovered SBDS gene product. His second long-term objective is to develop effective gene therapy strategies for diseases such as SDS.

## SEARCHING FOR THE FUNCTION OF THE SHWACHMAN-DIAMOND SYNDROME GENE PRODUCT, SBDS

Abdallah Nihrane, Schneider Children's Hospital, North Shore-Long Island Jewish Health System, New Hyde Park, NY, USA, Steven R. Ellis, University of Louisville School of Medicine, Louisville, KY, USA, and [Johnson M Liu](#), Schneider Children's Hospital, North Shore-Long Island Jewish Health System, New Hyde Park, NY, USA

Shwachman-Diamond Syndrome (SDS) is an autosomal recessive disorder characterized by pancreatic exocrine dysfunction, skeletal abnormalities, and bone marrow failure, which can evolve to leukemia. Mutations in SBDS have been shown to cause SDS, and SBDS's yeast ortholog, SDO1, appears to play a role in ribosomal RNA processing. Ribosomes are cellular organelles made up of RNA and proteins that are responsible for synthesizing proteins. Several bone marrow failure syndromes including dyskeratosis congenita, cartilage hair hypoplasia, Diamond Blackfan anemia (DBA) have already been linked to defects in ribosome synthesis. In order to clarify the function of SBDS, we have performed studies on the effect of depleting SBDS (or SDO1) in yeast, human, and mouse cells by using various molecular methods. The growth of human and mouse cells that lacked normal amounts of SBDS was markedly hindered, and this growth abnormality appeared to be related to an increased propensity to programmed cell death. In order to understand differences between normal and SBDS-depleted cells (both yeast and mammal), we analyzed corresponding patterns of gene expression and of ribosome syntheses (always comparing the normal cell to SBDS-depleted cell). When considered together, our data suggest a unique function of SBDS and its orthologs in making complete and working ribosomes. A better understanding of SBDS may enable us to target specific biochemical pathways to effectively diagnose and treat SDS patients in the future.

## 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 20 patients with SDS and follow them up for 2 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, email [melgheta@utmb.edu](mailto:melgheta@utmb.edu). **Dr. Elghetany's research is an ongoing study and he is still accepting bone marrow samples.**

## Research on Motility and Chemotaxis in SDS Neutrophils

Dr. Fred Goldman and Dr. David R. Soll, of the University of Iowa, are studying neutrophil motility and chemotaxis in SDS patients using advanced computer-assisted 2D and 3D motion analysis systems. A recent study completed last year in Dr. Soll's laboratory demonstrated a very specific defect in chemotaxis that was reproducible in all SDS patients that were examined. This is also consistent with several earlier reports of neutrophil motility defects in SDS. The proposed studies are important to SDS in many ways. First, it will shed light on this disorder and may lead to predictions as to the underlying molecular basis of SDS. Second, it may help explain certain clinical circumstances (e.g. infection propensisty), and offer the potential for developing strategies to correct this defect (e.g. lithium therapy). For more information contact Dr. Goldman's immunology nurse coordinator, Catherine Figueroa RN at (319)384-8101, or you may email Dr. Goldman

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## Update from Toronto: Genetic Testing for SDS

The research aims of the genetic testing in SDS families will no longer include active recruitment of additional patients. The research will now focus on the function of the gene and establishment of models of disease in order to understand what happens in the affected organs. Genetic testing, including pre-natal testing, is now being performed at the Molecular Lab at the Hospital for Sick Children (HSC). Information about the lab can be found on the web site: [www.sickkids.ca/molecular](http://www.sickkids.ca/molecular). The web site is currently being updated to include an announcement of testing for SDS and will include requisitions, general information about SDS and the cost of the analysis. Until the web site is updated, questions can be directed to Ms. Leslie Steele by e-mail: [leslie.steele@sickkids.ca](mailto:leslie.steele@sickkids.ca) or by phone 416-813-6590. A reminder for those who wish to receive the results from the genetic research study: We require written authorization to release the results to your Doctor. Please send the letter with your Doctor's contact information to: Dr. Peter Durie, GI/Nutrition, Hospital for Sick Children, 555 University Avenue, Toronto, Ontario, Canada

## 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 that a genetic predisposition to aplastic anemia and MDS is much more common than currently appreciated, and that 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 or influence the course of the disease. We are collaborating with researchers at several other institutions throughout the United States including St. Louis University, Boston Children’s Hospital, the University of California at San Francisco, the University of Iowa, Children’s Hospital of Pittsburgh, Oregon Health Science University, Duke University, and other collaborating centers. 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, or MDS. Advancing our knowledge of how these conditions develop is only possible because of the participation

## Participation in the Studies of the Molecular Mechanisms of Bone Marrow Failure

Our study is taking 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 will not be 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 contact the study coordinator:

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St. Louis, Missouri 63110, USA  
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David Wilson, M.D., Ph.D. Co-Director  
Division of Pediatric Hematology/Oncology  
Washington University School of Medicine;

## **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 contact Lisa Leathwood 1-800-518-8474 or you may also contact SDSF for more information.

## **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 [jkroppe@wowway.com](mailto:jkroppe@wowway.com) or call me at (248) 619-9316. 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

### **F.Y.I.**

Axcan Scandipharm, the makers of Ultrase enzymes, ADEKs vitamins, Scandishakes and many other products has included Shwachman-Diamond Syndrome in their CareFirst for CF Program, Comprehensive Care Program and RX Cost Reduction Program. SDS patients who use their products qualify for free and/or discounted products and information. For more information go to their website at [www.axcanscandipharm.com](http://www.axcanscandipharm.com) and click on Products and Services or call 866-AXCANRX for enrollment information

# Thank You to our Donors

(donations March 12, 2005 - June 30, 2005)

Rachel Anderson  
Daisy Morton  
Harold & Phyllis Duchan  
Mark McHugh  
Peggy & Stephen Lentz  
John & Joyce Wall  
Mr. & Mrs. Joseph Galko

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Larry Rinaldo  
Robert Roth  
Andrea Smith  
James Stephens  
Terence Upton  
Pilar Voutour

Angel Anna Baskets

Jeffrey & Lisa DeGriek

In Honor of Dylan Kolar

Nicole Zaugg

In Honor of Michelle Ellebracht

Florissant Elks Ladies Club, Inc.

In Honor of Patrick Kroppe

MMGS Troop 1549

In Memory of Steven Sohan

Florence T. Lenda  
Christine Price  
Rita & Richard Barry

In Memory of Charles Ruick

Andrew & Carol DeCarlo  
Norma Pitcairn  
Russell & Mary Jo Kieffer  
Cynthia Piper  
Doris Stob  
Thomas & Molly Onstott  
Irs & Pamela Sharfin  
Ruick Family Trust

**SDS bracelets purchased  
In Honor of Dylan Kolar**

Nancy Doxsee  
Kellie Smith  
Maureen Dolan  
Monica Beato  
Doreen Barile  
Carole Quigley  
Margaret Herrmann  
Violet Bischoff  
Kathleen McCarthy

**SDS bracelets purchased  
In Honor of Logan Stone**

Carolyn Middleton

**SDS bracelets purchased  
In Honor of Michele Mowery**

Caralmae Knickmeyer

**SDS bracelets purchased  
In Honor of Aiden Wilson**

Angela Durham

**SDS bracelets purchased  
In Honor of Danny Rohe**

Tricia Rohe  
Jennifer Wills  
Alecia Grawe  
Shari Etris  
Judy Rohe  
Janet Grawe

Brenda  
Debbie Zwick  
Michele Merk  
Tehresa Duerring  
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Kim Watson  
Denise Smith  
Lauren  
Rachel

**SDS bracelets purchased  
In Honor of Kaitlyn Bright**

Rita Sussko  
Michele Fontenst  
Kelly Bright  
Taylor Bright  
Donna Thompson  
Katherine Forse  
Velma Dudley

**SDS bracelets purchased  
In Honor of Chris Garfield**

The Holloway Family

SDS bracelets purchased  
In Honor of Michelle Ellebracht  
  
Robert & Caraimae Knickmeyer

## 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.

North Cambria, PA

Florence, KY

Burlington, WI

Macomb TWP, MI

Clintontown, MI

Pueblo, CO

Brandon, FL

Batavia, OH

Acworth, GA

Yucaipa, CA

Fort Wayne, IN

W. Harrison, IN

Wichita, KS

Hanahan, SC

Ostburg, WI

Orange, TX

## 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 our support group, the link is: [shwachmandiamond-subscribe@yahoogroups.com](mailto: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](mailto:jkroppe@wowway.com)

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## SCIENTIFIC CONFERENCE ABSTRACTS

### AVAILABLE VIA E-MAIL

If you would like a copy of the abstracts from the Third International Congress on Shwachman-Diamond Syndrome from June 2005, please e-mail us at [4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org). We will be happy to e-mail you a copy.

## Established Shwachman-Diamond Groups

### Shwachman-Diamond Syndrome Support - Australia

Contact: Joan Buchanan  
61 03 5427 0645  
email: [buchanafam@bigpond.com.au](mailto:buchanafam@bigpond.com.au)  
<http://www.shwachman-diamond.org>

### Shwachman-Diamond Support-UK

Contact: Kim Wright  
01 522 792039  
email: [kimwright@tesco.net](mailto:kimwright@tesco.net)  
<http://www.shwachman-diamondsupport.org>

### Italy Association for Shwachman Syndrome

Contact: Aurelio Lococo  
email: [aiss@shwachman.it](mailto:aiss@shwachman.it)  
<http://www.shwachman.it>

### Shwachman-Diamond Syndrome Canada

Contact: Karen Campbell  
email: [sdscanada@sympatico.ca](mailto:sdscanada@sympatico.ca)  
<http://www.shwachman.org>

### Shwachman Syndrome - Netherlands

Contact:  
email: [koster.e@hccnet.nl](mailto: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

**Doris Bull - UT:** (801)825-1734 or [nobull@xmission.com](mailto:nobull@xmission.com)

**Nancy Ruick - OH:** (614)855-0407 or [nruick@aol.com](mailto:nruick@aol.com)

**Corky DeBoer - IL:** (708)532-4954 or [opcrcdb@aol.com](mailto:opcrcdb@aol.com)

**Jenny Jenuwine - MI:** (810)395-2358 or [jengrsl2@bignet.net](mailto:jengrsl2@bignet.net)

**Kelly Bright -TX:** (409)738-2925

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

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

### OTHER COUNTRIES

**Kim Wright - England:**  
01522 792039 or [kimwright@tesco.net](mailto:kimwright@tesco.net)

**Lee-Anne Hayes - Australia**  
61 02 4968 9117 or [cerridwen@koee.com.au](mailto:cerridwen@koee.com.au)

**Reinald Baumhauer - Germany**  
Fax: 049-089-41902871 or  
[Reinald.Baumhauer@T-online.de](mailto:Reinald.Baumhauer@T-online.de)

**Aurelio Lococo - Italy**  
Tel. e Fax: +049 8736130 or  
[aiss@shwachman.it](mailto:aiss@shwachman.it)

## NEWSLETTER IDEAS

Do you have ideas for our newsletter? Want to share your story? Please send your suggestions and stories to SDSF at the address or email them to:

**[4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org)**

We appreciate ALL input! Thank you.

Do you have a question you would like to ask the doctor? We will print answers to questions in future newsletters. Send your questions to SDSF or email your questions to: **[4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org)**

## MOVING????

Please remember that we will need your new address if you are planning to move. Because our newsletter is sent "Bulk Rate" the post office will not forward it to you even if you have provided them with a forwarding address. Also, the newsletter will not be returned to us so we have no way of knowing you have moved. You can email us ([4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org)) or call our toll free number with your new address.

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Finland

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ogy  
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## WE NEED YOUR HELP PLEASE!!!!

Please send you tax deductible gift to: **Shwachman-Diamond Syndrome Foundation**  
710 Brassie Drive  
Grand Junction, CO 81506 U.S.A.

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.



**"Help Us Grow"**

710 Brassie Drive  
Grand Junction, CO 81506  
1-877-SDS-INTL

ADDRESS SERVICE REQUESTED

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**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.