



Cure For A Future

# Shwachman-Diamond Syndrome Foundation

## NEWSLETTER

**Winter 2006/2007**

HAPPY 2007

We are more than a month into the New Year and I am ever reminded how fast time passes. I would like to apologize for the oversight of not including the article by Dr. Elizabeth Kerr in the last edition of this newsletter. I am very pleased to announce that **it is** included in this one.

Some changes have been taking place within the board of directors of SDSF. Sharon Lamb will be vacating her position on the board. Sharon has been involved with this foundation for many years and has served on the board of directors since 2001, her most recent position as Treasurer. Sharon is a high school teacher, basketball coach, mother of two teenage children and is extremely busy. I would like to thank Sharon for her dedication to SDSF and for her years' of service to this foundation. She is definitely going to be missed. It is a pleasure to announce that we have two new members joining the board, Kelly Bright and Theresa Henle have both agreed to join our efforts for this worthwhile cause. You can look for more personal information about them in the next newsletter edition.

We are very pleased to announce that the first meeting

on the North American SDS Registry has taken place. Having an International/National Registry for patients with SDS has been a goal of this foundation for many years. I would like to thank all of the physicians who attended that meeting and for their ongoing commitment to the work that will need to be done to see this through to fruition. I would especially like to thank both Dr. Durie and Dr. Harris for their work and dedication to making sure this meeting happened.

Last but not least, I want to thank all the families, friends, and physicians who supported SDSF with a donation during our annual fund drive. Every dollar counts! It might seem that individually we can't do much, but together we can do A LOT!

Debbie Kadel and the board of SDSF



Cure For A Future

Shwachman-Diamond  
Syndrome Foundation

710 Brassie Drive  
Grand Junction, CO 81506 U.S.A.  
1-877-SDS-INTL (Toll Free)  
Fax: 970-255-8293  
E-mail: [4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org)  
Website: [www.shwachman-diamond.org](http://www.shwachman-diamond.org)

# **4TH INTERNATIONAL CONGRESS ON SHWACHMAN-DIAMOND SYNDROME**

**BOSTON, MASSACHUSETTS**

**JUNE 10 - 12, 2007**

Please save the date for the Fourth International Congress on Shwachman-Diamond Syndrome, the premier meeting for this disease. Bringing together expert clinicians and researchers from around the world and representing a broad range of different clinical and scientific disciplines, the congress will provide a forum to discuss the latest advances in scientific and clinical research. The meeting will be held in the Conference Center at Harvard Medical School on June 10-12, 2007. Our keynote speaker is Dr. David Nathan, a world-renowned hematologist noted for his work as both a clinician and a scientist.

For more information visit: [www.sdscongress07.com](http://www.sdscongress07.com). Online registration opens February 1, 2007.

Please forward this newsletter article or email link, [www.sdscongress07.com/save-the-date/](http://www.sdscongress07.com/save-the-date/), to anyone who might be interested in attending this important meeting. If you would also be kind enough to forward this email to your physicians for greater outreach, we would appreciate it.

---

## **Provisional Program**

**Welcome Address:** Introduction and Welcome:  
Richard J. Grand, MD

**Keynote Address:** "Congenital Bone Marrow Failure: No longer Enshrouded in Mystery": David G. Nathan, MD

### **Session 1. Clinical Features**

- Discussion Leader: Kim Smith-Whitley, MD
- Gastro-intestinal Features: Peter R. Durie, MD  
FRCPC
  - Hematological Issues: Akiko Shimamura, MD,  
PhD
  - Skeletal Features: Outi Makitie, MD, PhD
  - Endocrine Evaluation: Ingrid A. Holm, MD, MPH
  - Oral/Dental Features: Carol Mason,  
BDS(Hons),FDSRCS(Eng), ILT(M)
  - Neuropsychology Features: Elizabeth Kerr,  
BASC, MA, PhD
  - Genetic Counseling: Ellis J. Neufeld, MD, PhD
  - Malignancy: Blanche P. Alter, MD, MPH

### **Session 3. Workshop: Shwachman-Diamond**

**Workshop:** Shwachman-Diamond Syndrome:  
Clinical Diagnosis and Work-up: Johanna Rommens/Akiko Shimamura

### **Session 2. Surveillance, Management, and Treatment**

- Discussion Leader: M. James Lopez, MD, PhD
- Psychosocial Issues: Nancy Cincotta, M.S.W.
  - Nutrition and Pancreatic Supplementation:  
Mark E. Lowe, MD, PhD
  - Malignancy: Surveillance, Treatment, and  
Out comes: Jeffrey M. Lipton, MD, PhD
  - Hematopoietic Stem Cell Transplantation:  
Charlotte Niemeyer, MD

## **Syndrome Registries**

Discussion Leader: Peter R. Durie, MD FRCPC

- Marco Cipolli, MD
- David C. Dale, MD
- Jean Donadieu, MD, PhD
- Yigal Dror, MD
- Elene Psiachou-Leonard, MRCPCH, MRCP(I), FGPA, FGHA
- Corneila Zeidler, MD

## **Session 4. SDS Genetics**

Session Chair: Taco W. Kuijpers, MD, PhD

- Johanna Rommens, PhD
- Shiro Ikegawa, MD, PhD

## **Session 5. SBDS Function**

Session Chair: Johanna Rommens, PhD.

- Yigal Dror, MD
- Johnson M. Liu, MD
- Emanuela Maserati, MD, PhD
- Akiko Shimamura, MD, PhD
- David R. Soll, PhD
- Alan J. Warren, PhD FRCP FRCPath

## **Session 6: SBDS and Hematopoiesis**

Session Chair: Yigal Dror, MD

- George Q. Daley, MD, PhD
- Taco W. Kuijpers, MD, PhD
- Daniel C. Link, MD

## **Session 7. SBDS and Leukemia**

Session Chair: Alan J. Warren, PhD FRCP FRCPath

- A. Thomas Look, MD
- Kevin Shannon/Jasmine C. Y. Wong, PhD
- Daniel G. Tenen, MD
- M. Tarek Elghetany, MD

## **Session 8. Links Between SDS and Other Bone Marrow Failure Syndromes**

Session Chair: Jeffrey M. Lipton, MD, PhD

- Johnson M. Liu, MD
- Steven R. Ellis, PhD
- U. Thomas Meier, PhD
- Inderjeet Dokal, MBChB, MD, FRCP, FRCPCH, FRCPath
- Davide Ruggero, PhD

## **Session 9. Organ Development and Failure**

Session Chair: Mark E. Lowe, MD, PhD

- Raymond J. MacDonald, PhD
- Sanna Toiviainen-Salo, MD

## **Session 10. Novel Diagnostics and Therapeutics**

Session Chair: Johnson M. Liu, MD

- Wan F. Ip, MASC
- Frederick D. Goldman, MD
- Christopher Walsh, MD, PhD
- Adrianna Vlachos, MD

## **Session 11. Late Breaking Stories/Selected Abstracts**

*The provisional program is subject to change at anytime without notice*

*In partnership with Shwachman-Diamond Syndrome Foundation and Children's Hospital Boston*

# ***DOCTOR DATABASE***

It is finally here!! The list below is a compilation of the doctor's names that have been involved with patients of SDS. These names have been given to us by the patients and/or their families in order to help others with SDS find a doctor in their area. We originally wanted to call this listing CareSearch but decided to keep it simple by calling it Doctor Database. We are contacted every month by a new family in need of a doctor with some knowledge of SDS and it's treatment or how to refer that patient for treatment. Thus have created this list which you see here and on our web site. This list is not an approved and/or recommended list from our board but instead, a listing of doctors who have been recommended by their patients and families.

Please feel free to email me ([blairvanbrunt@comcast.net](mailto:blairvanbrunt@comcast.net)) with the names of your doctors (please list their title, address, phone number and most importantly their specialty, if you can) if you feel they are doctors who others would benefit visiting. The larger the list, the more we help our fellow SDSers. Who knows? You could even help an existing patient find a doctor closer to home. Thanks for helping.

## **Regional Clinics**

### **New England :**

The Bone Marrow Failure Program at Children's Hospital Boston  
Dr. Akiko Shimamura  
300 Longwood Ave  
Boston, MA 02115  
617-335-8246  
[www.childrenshospital.org/clinicalservices/Site1970/mainpageS1970P0.html](http://www.childrenshospital.org/clinicalservices/Site1970/mainpageS1970P0.html)

### **Mid-Atlantic:**

National Institutes of Health  
Principal Investigator: Blanche P. Alter, M.D., MPH  
800-518-8474  
[www.marrowsfailure.cancer.gov](http://www.marrowsfailure.cancer.gov)  
Ask this clinic for a doctor referral in the area.

Although this clinic is for research only, it can be an important tool for those who live in the area.

### **West Coast:**

Oregon Health & Science University  
Division of Pediatric Hematology/Oncology  
Mail code CDRCP  
3181 S.W.Sam Jackson Park Road  
Portland,OR 97239-3098  
Appointments: 503-418-5150  
Coordinator: Kelly Brady,R.N.503-418-5341  
Peter Kurre,M.D.  
Assistant Professor,  
Pediatrics, and Cell and Developmental Biology  
[www.ohsuhealth.com/dch](http://www.ohsuhealth.com/dch)

### **Midwest:**

Cincinnati Children's Hospital Medical Center  
Bone Marrow Failure Clinic  
MLC 11013  
3333 Burnet Avenue  
Cincinnati, OH 45229-3039  
[www.cincinnatichildrens.org/svc/alpha/b/blood/programs/bmf-clinic](http://www.cincinnatichildrens.org/svc/alpha/b/blood/programs/bmf-clinic)  
\*\*For more information, or for a consultation, please contact the Bone Marrow Failure Coordinator, Sue Heist, RN, at 513-636-3570, or toll-free at 1-800-344-2462, extension 3570.

### **New York:**

Bone Marrow Failure Program And Fanconi Anemia Comprehensive Care Clinic  
Lipton, Jeffrey M, M.D., Ph.D.  
Pediatric Hematology/Oncology  
Pediatrics  
  
Pediatric Hematology/Oncology And Stem Cell Transplant  
269-01 76th Ave  
New Hyde Park, NY 11040  
718-470-3470

**Near Toronto:**

Dr. Peter Durie  
Division of Gastroenterology and Nutrition  
The Hospital for Sick Children  
555 University Ave  
Toronto, Ontario, Canada. M5G 1X8  
Phone: 416-813-6185  
Fax: 416-813-6531

**For those who don't live near these clinics, here are other names to contact. These are names from our families' recommendations. Please feel free to contact these doctors for consultation or referrals:**

Dr. Sawaf  
Pediatric Hematology/Oncology  
19229 Mack, Suite 28  
Grosse Pointe Woods, MI 48236  
313-647-3200

Amarnath, Rathna MD  
Palmetto Pediatric Gastro  
2113 Adams Grove  
Columbia, Sc 29203  
803-254-1006

Dr. Roger H. Giller  
Specialty: Hematology/Oncology - Pediatrics  
The Children's Hospital  
1056 E. 19th Avenue  
Denver, CO  
303-861-6776

Gaia Georgopoulos M.D.  
Specialty: Orthopaedic Surgery  
The Children's Hospital  
1056 E. 19th Avenue  
Denver, CO  
303-861-6600

Michael S. Kappy M.D.  
Specialty: Endocrinology - Pediatrics  
The Children's Hospital  
1056 E. 19th Avenue  
Denver, CO  
303-861-6128

Ronald J. Sokol M.D.  
Specialty: Gastroenterology - Pediatrics  
The Children's Hospital  
1056 E. 19th Avenue  
Denver, CO  
303-861-6669

Dr. Ashok Raj  
Pediatric Hematologist  
Louisville, KY  
502-629-7750

Dr. Thomas Stephen  
Pediatric GI  
Louisville, KY  
502-629-5796

Steven Werlin MD  
The Children's Hospital of Wisconsin  
9000 Wisconsin Ave  
Milwaukee, WI  
Contact: Lillian Sablan RN  
414-266-3379

---

***THE SHWACHMAN  
DIAMOND SYNDROME  
UK REGISTRY,  
MREC APPROVAL***

The above project has now received favorable Multicentre Research Ethics Committee (MREC) approval. For information about registering a patient contact Dr. Elene Psiachou-Leonard, Chief Investigator, on 01162585309 or email [elene.psiachou-leonard@uhl-tr.nhs.uk](mailto:elene.psiachou-leonard@uhl-tr.nhs.uk).

# ***LEARNING PATTERNS OF CHILDREN WITH SHWACHMAN-DIAMOND SYNDROME***

At the 2006 SDS Family Conference, Dr. Elizabeth Kerr gave a very interesting talk focusing on the learning patterns of children (ages 6-17) with Shwachman-Diamond Syndrome. Based on the findings of her research, Phonological Processing, Visual Processing, Attention, Flexible Problem Solving, and Behavior were among the types of difficulties that can be experienced. As such, the following recommendations were made:

If you notice specific difficulties, the earlier your child receives support or intervention the better. With a “wait and see” approach, you may miss many months of valuable support.

## **Phonological Processing/ Reading/Spelling:**

Phonological processing is related to the use of sound structures in processing written and oral language and involves rapid naming, phonological awareness (i.e., the sound structures), and phonological memory. Chronic middle ear infections almost always infer with hearing. In turn, delays in language development may be evident earlier on with continued delays in phonological processing. Weaknesses in phonological processing are related to delays in reading and spelling achievement. Research at the Hospital for Sick Children and their colleagues (e.g., “Putting struggling readers on the PHAST track” m. Lovett et. al., Journal of Learning Disabilities, 2000 Vol 33 (5), pgs 458-476) have discovered three essential components to remediation:

- (1) Development of pre-requisite skills which includes letter sounds associations, identification of sight words, recognition of vowel, variant vowel sounds, and affixes. These skills are best developed if a specific block of time is set aside each to focus on them. Direct Instruction can be extremely beneficial. Lessons are highly structured and scripted. Responses are modelled for the student. The structure and repetition of the lessons leads to on-line processing of information. Teaching is reciprocal, in that, the instructor is constantly checking in with the student to ensure that he or she is learning. “Direct Instruction Reading” by Douglas Carine, Jerry Silbert, and Edward Kameenui (Prentice Hall, 1996) is one resource. Other suggestions include the grapheme level activities outlined in “Phonological Awareness Kit-Intermediate” and activities such as deletions (i.e., Rosner activities) and substitutions (i.e., Lindamood activities).
- (2) Practice in word identification strategies. Specific strategies include (a) Rhyming or being taught a list of relatively simple words containing common spelling patterns (e.g., look) and then being taught how to compare new, unfamiliar words to the list (e.g., book, nook, shook, brook, etc); (b) Vowel alert or being taught that each vowel sounds has a short and a long sound and how to use different sound to make a real word (e.g., “I see the vowel ‘o’ in this word. First I’ll try ”o” as in “go”. Next I’ll try ‘o’ as in “dog”. That sounds like a read word.”); (c) I Spy or being taught how to look for small words or parts for words in longer, more difficult words (e.g., identify ‘bad’, ‘in’, and ‘on’ in the word “badminton”); and (d) Peeling off, or being taught how to identify and remove prefixes and suffixes before using another strategy to read the root word.
- (3) Use of learning to learn strategies. These strategies include verbal scripts that an individual can use when approaching a new task or word. The highly scripted approach to direct instruction exposes a child to this as do the examples outlined in number 2 above.

## Visual Processing:

Visual processing is related to the ability to process, interpret, and organize visual and visual-spatial information. When an individual has weaknesses in these areas, it can cause difficulties at school and sometimes in the social realm. For a given individual, they might display some (but not necessarily all) of the following: poor internal and external organization, difficulty coping with changes in routine, difficulty with generalizations, making literal translations, being overwhelmed, having difficulty with directional concepts and co-ordination, and being readily distractible.

“The Source for Non-Verbal Learning Disorders” by Sue Thompson (lingsystems) and “Educational Care: A system for understanding and helping children with learning problems at home and in school” by Mel Levine (Educators Publishing Service, Inc) are two resources which provide information for understanding and assisting with weaknesses in visual processing. If a child’s language skills are stronger, then teaching them to put labels on what they are seeing and doing can help solidify information.

## Attention:

Attention is multifaceted. It refers to a number of processes including: (1) how much information a person can hold in his mind and process at one time (i.e., attention span and working memory); (2) whether an individual is able to focus on, or search for, a specified target (i.e., selective attention); and (3) how well s/he can pay attention during a mundane or boring task (i.e., sustained attention). A child who has difficulties with attention may have trouble listening when someone talks, waiting her or his turn, completing a task, or returning to a task if interrupted. By the age of 5, a child needs to be able to pay attention for a least 25 minutes in order to perform adequately in school.

TeachADHD ([www.teachadhd.ca](http://www.teachadhd.ca)) is a new resource developed by researchers at the Hospital for Sick Children and their colleagues for use by teachers at school. It instructs on various types of attention and helps the teacher develop strategies to harness attention within the classroom.

At school and at home it will be important to:

- Follow a structured daily routine.. School and household routines help the inattentive child to accept order. Keep the times for wake-up, meals, snacks, chores, naps, and bed as regular as possible. Try to keep your environment relatively quiet because this encourages thinking, listening, and reading at home. In general, leave the radio and TV off. Predictable daily events help your child’s responses become more predictable.
- Maintain firm discipline. Children with attention difficulties need more carefully planned discipline than the average child. Rules should be formulated mainly to prevent harm to your child and to others. Aggressive behavior, such as biting, hitting, and pushing, should be no more accepted in an inattentive or hyperactive child than in the normal child.

At home:

- **Stretch your child’s attention span.** Encouraging attentive behavior is the key to preparing your child for school. Increased attention span and persistence with tasks can be taught at home. Be sure to praise your child when he plays independently. Set aside several brief periods each day to teach your child listening skills by reading to him. Teach games to your child, gradually increasing the difficulty by starting with building blocks and progressing to puzzles, dominoes, card games, and dice games. Later, consequence games such as checkers or tic-tac-toe can be introduced. When your child becomes restless, stop the activity and return for another session later. Plan to have your child do homework and other tasks that require concentration in short blocks of time with breaks in between. Try having your child study with low-level background sound such as white noise or instrumental music. Do homework and studying away from the sounds of television, radio, or others talking but where adults can supervise.

## **Flexible Problem Solving:**

This skill typically develops with age and is required more at school when a child reaches his or her teen years. Flexible problem solving requires the ability to generate ideas, as well as to maintain and/or shift thought processes. Individuals with difficulties in this area may have trouble dealing with ambiguity or getting “unstuck” from an automatic way of responding.

Teachers and parents can help students in this area by making the steps involved in an assignment or daily activity more explicit (e.g., provide “cheat” sheets or templates that outline the steps required to write the essay, to analyse and solve math problems etc). Embed questions designed to prompt the individual into using analytical skills. Questions can include “How did you solve that problem?” “Can you think of another way of doing that?”, “What can you do to help remember that information”. Teach the individual a set of questions to ask him/herself when confronted with a problem, such as: “What is my problem? What is my plan? Am I following my plan? How did I do?”. In other words have the individual identify the problem, develop a solution strategy, self-monitor his or her performance and evaluate the outcome.

The Learning Toolbox website ([http://coe.jmu.edu/Learning\\_Toolbox/](http://coe.jmu.edu/Learning_Toolbox/)) is designed for secondary students with specific learning challenges (e.g., organization, problem solving, attention etc) as well as for their teachers and their parents. The student section includes tools to help the student improve in many areas (e.g. Study skills, test taking, advanced thinking, organization) by providing templates to follow.

## **Behavior:**

Children and adolescents who are not doing well in school may not feel good about themselves. If they feel they can't cope, they may withdraw from their friends and social activities. Social difficulties were raised by parents of children and adolescents with SDS on questionnaires. Two resources to foster social competence are:

- “No one to play with: Social problems of LD and ADD Children” by Betty B. Osman, and
- “Raise Your Child's Social IQ” by Cathi Cohen.

# ***UNDERSTANDING CBC RESULTS***

Ever have trouble deciphering all those numbers 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](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 Adobe Acrobat reader installed in order to open it.

---

# ***DONATE FROM YOUR IRA THIS YEAR!***

Tell your friends and family that they can donate funds from their IRA to SDSF without having to pay income tax on the distributions for 2006 and 2007 tax years only or if you are over 70 1/2 years old. The new Pension Protection Act of 2006 allows up to \$100,000 to be donated directly from an Individual Retirement Account and avoid any income taxes on the withdrawal. This is only for IRAs and not other qualified retirement plans. If you have another such plan and want to donate tax free to SDSF, it has been suggested that you create a new IRA and roll assets from the other retirement plan into the new IRA. As with all articles in this newsletter, please consult your own advisor to determine if this new provision in the Pension Protection Act can apply to your situation.

# ***FAMILY SHARING PAGE***

---

Hello SDSF Community. My name is Chris and I'm a nineteen year old student at a very large public University, five hours away from home. My mom suggested that I write a short letter about what it is like to have bone marrow failure and be away from home and responsible for my own health for the first time, so here it is!

This is actually my second year being away from home. I originally planned on going to a large University a thousand miles away from home. In preparation, my mom arranged for me to be followed by a doctor there that did pediatric bone marrow transplants. We were able to meet with him ahead of time and set up a plan and protocol. I felt very comfortable going away; sure everything would go as planned. Unfortunately, Mother Nature had a different idea! My first college experience lasted all of 4 hours, just enough time to move into my dorm. We were then told to put our evacuation plan into place and Hurricane Katrina followed us up to the evacuation location and hit us dead center! My plan to attend Tulane University destroyed, I then ended up at a medium sized University about 150 miles from home. I had a private room in the dorm and amazingly enough, even with all the stress of everything I had been through my first year of college, I didn't get sick once! I thought this would be a piece of cake.

I transferred to another school for my second year. This school, although in-state, is actually further away from home and much larger than the one I had just finished. I know a lot of kids here so I am enjoying this year more. I still have a private room (the one thing my hematologist insisted on) in the dorm. My mom tried to be proactive and set up an appointment for a local hematologist, but this time ran into more roadblocks and nothing was finalized ahead of time. Overall I've still been very healthy. My first illness was some sort of stomach virus. My RA (Resident Assistant) heard me being sick and called the ambulance (they are very proactive here!) and carted me off to the ER. This was the first time

I've had to try and explain my illness to a doctor. They did counts, which were good, kept me for 12 hours for IV hydration and kicked me loose. At this point, I still felt pretty good about how it all went and I didn't even call my parents until the next day.

Five days later though was a different story, I woke up with

a fever, headache, and chills. I felt awful! I called my mom and she said I should get an appointment at the University Health Center. There I had to try and explain AGAIN to another doctor about SDS and bone marrow failure! I felt awful and really couldn't answer all his questions, but I think I did a pretty good job. This time I did sign the HIPAA papers to allow my parents to talk to the doctors and got the doctor's phone number to give to my mom. The doctor gave me antibiotics and sent me back to my dorm, stating he REALLY wanted me to have a local hematologist because he didn't feel comfortable treating me.

The second illness really threw me for a loop. I missed several days of classes, felt awful, and was tempted to just go home. I stuck it out and once I arranged to make up all my missed classes and assignments felt better about the experience. My mom finally got an appointment with a local Oncologist (only about 5 miles from campus) and we met him last week. He didn't really remember anything about SDS but is interested in learning about it. My counts were amazing, so things are looking up again. I feel better knowing that if I get sick I can just tell the Health Center or ER doctors to call him. In fact, most of the time I will be able just to go to him and get what I need and bypass the Health Center/ER altogether.

I've been really lucky; as I've gotten older my health has improved. My parents have, for the most part, treated me just like any other teenager. If I have any advice to others in my situation it would be, make sure you arrange to have a local doctor who knows you and how to treat your illnesses. I still see my home hematologist as my primary specialist, but it takes a lot of pressure off me to have someone here at school that I can go to if I do get sick.

Chris G.

# SPOTLIGHT ON RESEARCH

## *RECENT RESEARCH DONE IN SDS*

In recent years exciting research has been and continues to be done for patients with Shwachman-Diamond Syndrome. The discovery of the gene responsible for SDS in December, 2003 was of utmost importance; however, ongoing research enables scientists and doctors to better understand this disease and will hopefully lead to both a cure and to improved treatment regimens for patients. For parents and patients interested in following the research, an online search can be made using PubMed, which is maintained by the National Library of Medicine and the National Institutes of Health. Upon opening the PubMed website (by using your search engine to do a search for PubMed) simply enter Shwachman-Diamond Syndrome in the disease search and a list of research in chronological order will appear. The research appears with date, author, title, journal name, volume, and pages listed. An abstract of each article can be viewed by clicking on it. The abstract will briefly explain the research conducted and will give possible conclusions and suggested future areas of research. Related links are also listed, as well as a PubMed identification number. With this number, a full text article can often be obtained through the publisher or through a medical library. Although the information is often written in scientific or medical terminology, it can provide us with information to better understand the disease and to share with our own doctors.

Below is a list of a few of the recent studies you will see listed on a PubMed search:

Sauer M, Zeidler C, Meissner B, Rehe K, Hanke A, Welte K, Lohse P, Sykora KW.

Substitution of cyclophosphamide and busulfan by fludarabine, treosulfan and melphalan in a preparative regimen for children and adolescents with Shwachman-Diamond syndrome.

Bone Marrow Transplant. 2007 Jan 8; [Epub ahead of print]

Ruggiero A, Molinari F, Coccia P, Attina G, Maurizi P, Riccardi R, Bonomo L.

MRI findings in Shwachman diamond syndrome.

Pediatr Blood Cancer. 2006 Dec 20; [Epub ahead of print]

Shimamura A.

Inherited bone marrow failure syndromes: molecular features.

Hematology Am Soc Hematol Educ Program. 2006;:63-71.

Rosendahl J, Teich N, Mossner J, Edelmann J, Koch CA.

Compound Heterozygous Mutations of the SBDS Gene in a Patient with Shwachman-Diamond Syndrome, Type 1 Diabetes Mellitus and Osteoporosis.

Pancreatology. 2006 Nov 10;6(6):549-554 [Epub ahead of print]

Kawashima H, Ushio M, Aritaki K, Kashiwagi Y, Watanabe C, Nishimata S, Takekuma K, Hoshika A, Taneichi H, Kanegane H.

Discordant endocrinopathy in a sibling with shwachman-diamond syndrome.

J Trop Pediatr. 2006 Dec;52(6):445-7. Epub 2006 Oct 19. No abstract available.

Erdoş M, Alapi K, Balogh I, Oroszlan G, Rakoczi E, Sumegi J, Marodi L.

Severe Shwachman-Diamond syndrome phenotype caused by compound heterozygous missense mutations in the SBDS gene.

Exp Hematol. 2006 Nov;34(11):1517-21.

Macipe Costa RM, Javierre Miranda E, Lou Frances MG, Heredia Gonzalez S, Calvo Martin MT.

[Shwachman-Diamond syndrome. A case report]

An Pediatr (Barc). 2006 Jul;65(1):79-82. Spanish.

Zhang S, Shi M, Hui CC, Rommens JM.

Loss of the mouse ortholog of the Shwachman-Diamond syndrome gene (Sbds) results in early embryonic lethality.

Mol Cell Biol. 2006 Sep;26(17):6656-63.

Taneichi H, Kanegane H, Futatani T, Otsubo K, Nomura K, Sato Y, Hama A, Kojima S, Kohdera U, Nakano T, Hori H, Kawashima H, Inoh Y, Kamizono J, Adachi N, Osugi Y, Mizuno H, Hotta N, Yoneyama H, Nakashima E, Ikegawa S, Miyawaki T.

Clinical and genetic analyses of presumed Shwachman-Diamond syndrome in Japan.

Int J Hematol. 2006 Jul;84(1):60-2.

Shimamura A.

Shwachman-Diamond Syndrome.

Semin Hematol. 2006 Jul;43(3):178-88. Review.

Anyone interested in help viewing the abstracts or in obtaining a full text of an article can contact Susan Utz at [sutz123@insightbb.com](mailto:sutz123@insightbb.com) for assistance.

Shwachman-Diamond Syndrome Foundation (SDSF) is committed to both supporting ongoing research and providing family support. Our organization is currently providing funding to three researchers studying aspects of SDS. For more information about these studies and other ongoing research, see reviews contained in this newsletter.

---

## RESEARCH SUPPORTED BY YOUR DONATIONS

### STUDY USING YEAST GENE AS A MODEL FOR THE SBDS GENE

Dr. David Pearce of the University of Rochester Medical Center is using Baker's yeast as a model to study Shwachman-Diamond Syndrome. According to Dr. Pearce, many of the genes or proteins that are associated with human disease are also present in yeast. In fact many of the normal cellular processes that occur in cells are similar to those in yeast. The yeast cell contains a gene designated SDO1, which stands for Shwachman Diamond Ortholog 1, ortholog meaning that if yeast were human, it is the same gene. Thus, SDO1 is essentially the same as the human SBDS gene, which when defective causes Shwachman-Diamond Syndrome. Using yeast has many advantages, as it is simple to grow and easy to manipulate. Dr. Pearce is studying SDO1 on the assumption that the information gained on the role SDO1 has in the yeast cell will be directly applicable to studying what the function of SBDS will be.

The first question being investigated is what happens to yeast that lacks the SDO1? Yeast DNA can easily

be altered, so they have specifically removed its SDO1 gene. These yeast are called the “Shwachman Diamond yeast”. They have been studying what effect an absence of SDO1 has on yeast, with respect to normal function. So far they have found that yeast that do not have SDO1 grow very poorly. This enables them to focus discovering ways to “suppress” or fix this using tricks of yeast genetics to uncover other genes that work with SDO1.

The second question is to ask what SDO1 or SBDS does exactly in the cell. In human cells, it has been shown that SBDS is found in the nucleus, a subcompartment in the cell that generates and processes information for distribution around the cell to ensure proper function. The yeast SDO1 also localizes to the yeast nucleus. However, Dr. Pearce has found that some of the SDO1 actually exits the nucleus and aims to understand what SDO1 does outside the nucleus.

### **SBDS mRNA EXPRESSION IN PERIPHERAL BLOOD LEUKOCYTES**

Dr. Elena Nicolis and her colleagues at the Azienda Ospedaliera di Verona, in Verona, Italy are interested in the study of mRNA levels of SBDS in patients. The SBDS transcript (mRNA) is widely expressed in many tissues and mutations cause a wide variety of abnormalities and symptoms. SDS affects the exocrine pancreas, bone marrow and skeleton. Patients also exhibit hematological abnormalities such as neutropenia and are predisposed to the development of myelodysplasia and acute myelogenous leukemia.

The three most frequent SBDS gene mutations appear to be severe as they are predicted to generate truncated SBDS proteins. Recent studies indicate that the clinical phenotype of SDS patients is associated with expression of hypomorphic SBDS alleles; such that low levels of protein and function occur in all patients and that complete absence of SBDS is not compatible with life.

Their research project entitled “SBDS mRNA Expression in Peripheral Blood Leukocytes” is intended to detect and quantify the SBDS transcripts obtained from leukocytes derived from peripheral blood of SDS patients and healthy subjects. Moreover they are

planning to compare transcripts and protein expression and match transcript expression levels between different SBDS genotypes in SDS patients, in order to investigate the possible relation between SBDS mRNA levels, SBDS protein content and phenotypic variability, which is notable in the SDS population.

This work is being done in collaboration with the project by Peter Durie, Johanna Rommens, Wan Ip (The Hospital for Sick Children, Toronto) and Akiko Shimamura (Children’s Hospital, Boston), entitled “SBDS Protein Expression in Peripheral Blood Leukocytes”.

### **INVITATION TO PARTICIPATE IN A RESEARCH PROJECT ENTITLED:**

#### **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:

Lynda Ellis, RN

[lynda.ellis@sickkids.ca](mailto:lynda.ellis@sickkids.ca)

Phone: 416-813-5515

### **NEW RESEARCH STUDY ACCEPTING PARTICIPANTS:**

#### **STUDY INVESTIGATING THE RELATIONSHIP BETWEEN THE SBDS GENE, DNA REPAIR AND TELOMERE ELONGATION**

The research of Drs. Neal Young and Rodrigo Calado of the Hematology Branch of the NIH focuses on defining the function of the SBDS gene product, specifically as it relates to DNA repair and telomere elongation. Through their study they hope to define the role of the SBDS gene in telomere repair. The results of this study will potentially be relevant to the diagnosis and treatment of patients with many marrow failure syndromes.

Through their preliminary investigations they have found mutations in the SBDS gene in some patients with apparently acquired Aplastic Anemia, who also have very short telomeres. In addition, they have already described mutations in other genes that help repair telomeres in this same patient population. Therefore, they feel that SDS, like Dyskeratosis Congenita, appears to have an adult form, and that SBDS may be involved in repairing chromosomes after cells replicate.

To participate in the study a small amount of blood (5-10 ml, anticoagulated), is required from patients with SDS. A special phlebotomy need not be performed, but an extra tube of heparinized blood could be obtained along with routine blood counts or other laboratory studies. Any patient with a clinical diagnosis of SDS is invited to participate. The study will pay any shipping costs associated with sending the specimen.

For further information contact: Rodrigo Calado at 301-496-5093 or [calador@nhlbi.nih.gov](mailto:calador@nhlbi.nih.gov)

## **SBDS STUDY UPDATES:**

### **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 tests on the samples. Dr. Elghetany will also be testing bone marrow samples from Dr. Blanche Alter's study above.

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, email [melgheta@utmb.edu](mailto:melgheta@utmb.edu). Dr. Elghetany's research is an ongoing study and he is still accepting bone marrow samples.

## ***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](http://www.shwachman-diamond.org). Sorry no COD's or credit cards. Please allow 2-3 weeks for delivery. Make your check payable to Jenny Jenuwine. All proceeds go to SDSF. 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, 15028 Hough, Allenton, MI 48002  
810-395-2358 or [jengr1s2@klondyke.net](mailto:jengr1s2@klondyke.net)

## 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@yahogroups.com](mailto:shwachmandiamond-subscribe@yahogroups.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)

---

## 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.

Haverhill, MA

Petersburg, PA

National City, CA

Littleton, CO

Edmond, OK

Hicksville, NY

Mitchell, SD

## 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](mailto: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, 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 September 20, 2006 - February 10, 2007)

Diana & David Murphy  
Joseph F. Olivo, Jr.  
Bernard & Andrea DiFalco  
John & Joyce Wall  
Stephen & Peggy Lentz  
Jan James  
John & Jeanette Brooks  
Frederick & Trudy Goldman  
Pediatric Gastroenterology Consultants, P.C.  
Solvay Pharmaceutical  
The Louise H. and David S. Ingalls Foundation  
Southeastern Michigan Area CFC  
Heart of West Michigan United Way  
Fort Polk-Central Louisiana CFC

**Allstate Giving Campaign**  
Shana Hovey

**In Honor of Aidan Wilson**  
Kelley, Casey & Moyer, P.C.

**In Honor of Patrick Kroppe**  
Craig A. Colby  
Troy Athens High School Student Body

**In Honor of Gavin Miller**  
Adam & Amber Sanchez

**In Honor of Kaitlyn Bright**  
Eastgate United Pentecostal  
Church Ladies Auxillary

**In Appreciation of Julie Kroppe**  
Mr. & Mrs. Donald McNew

**In Honor of Gracie Van Brunt**  
Gracie Fund

**In Honor of Brittany Kadel**  
T. Kent & Elizabeth Harbert

**In Honor of Clint Johnson**  
Kathy Poth

**In Honor of Heather Pendergast**  
Estate of Stephen Sohan

**In Honor of Collin Brown**  
Mike & Shannon Yarbrough  
John & Barbara Dickson  
Mary L. Anderson  
Bryan & Gloria Norman  
Wayne & Nancy Heape  
Robert & Betty Letzig  
Bob & Mary Jane Nelson  
Morris & Judy Salter

**In Honor of Katie Ruick**  
June C. Ruick

**In Honor of Marissa Avroch**  
Bryan & Jennifer Avroch

**In Honor of Ryan Miller**  
Nancy Miller

**In Honor of Owen Bannett Murphy**  
Thomas & Betty Lewellen

**In Memory of Ima F. Smith**  
Bettie & Michael Papajohn

**In Memory of William J. MacKenzie, Jr.  
In Honor of Gracie Van Brunt**

Mr. & Mrs. Bradford Briggs  
Virginia & Tom Citron  
Louise S. Walsh  
Thomas B. Slaughter  
Jane K. Morgan  
Richard B. White  
Mrs. Arthur R. Kneibler  
Carol Webb  
Caroline & Walter Whetstone  
Samuel & Elizabeth Thorne  
Donal & Katharine O'Brien  
Robert & Campbell  
Moccasin Lake Foundation  
Caroline Cole  
Janey & Stuart Symington  
Marie Roberts  
Peter B. Cannell & Co., Inc.  
Thomas L. Cassidy  
Eric & Elizabeth Johnson  
Gayle A. Findlay  
George & Jill Stevens  
Janecke C. Madsen  
Florence Durfee  
Jonathan Isham  
Shirley Mck Tanner  
Ann Cohen  
Melissa Widdifield  
Amy & Steven Roberts  
Donald & Nancy Kipp  
Richard K. Leblond  
James & Carol Hollyday  
Kenneth F. Mountcastle, Jr.  
Ervin F. Bickley, Jr.  
Edwina S. Millington  
Donald & Helen Douglass  
Harley & Carolyn Stimmel  
Richard & Leslie Pierpont  
Jean B. Isler  
David J. Feldman  
Richard & Mary Harris  
Robert & Nancy Oden  
Jonathan K. Spole  
Ada Weld Osborn  
Sandra L. Traff  
John & Lucille Carter  
Sandra & Michael Chism

Scott & Amy Stevens  
Hanna & Kris Henderson  
Horace I. Crary  
Delano W. Ladd, Jr.  
Frank & Rose Canonico  
James Rogers  
Marion Baldwin

**In Honor of Dylan Kolar**  
Friends from South Shore Home Health Services

**BRACELET PURCHASES  
(Sept. 20, 2006 - February 10, 2007)**

**In Honor of Logan Stone**  
Leah Gentry  
Nancy Molnar  
Adrienne Molnar  
Katie Herfel  
Bonnie Allphin  
Leslie Rajchel  
Susan Utz  
Jennifer McDonogh  
Kaitlyn Sisco  
Lara Schillizzi  
Robin Whitesell  
Linda Webb  
Lu Ann Avery  
Jana Lemon

**In Honor of Tyler Knoble**  
Vicki Brown  
Sue Hill  
Judy Corbit

**In Honor of Marissa Arvoch**  
Nicole Carpiello

**In Memory of Marc Strumm**  
Anita Strumm

## Established Shwachman-Diamond Groups

### Shwachman-Diamond Syndrome Support - Australia

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

### Shwachman-Diamond Support-UK

Contact: Sharon Clusker  
Tel: 024-76345199 Fax: 024-76345199  
email: [enquiries@shwachman-diamond-support.org](mailto:enquiries@shwachman-diamond-support.org)  
<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

email: [koster.e@hccnet.nl](mailto: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 at the address or e-mail them to: [4sskids@shwachman-diamond.org](mailto:4sskids@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](mailto:opcrccdb@aol.com)

**Jenny Jenuwine - MI:** (810)395-2358 or [jengrls2@netzero.net](mailto:jengrls2@netzero.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

**Sharon Clusker - England:**  
[Sharwk60@aol.com](mailto:Sharwk60@aol.com)

**Lee-Anne Hayes - Australia**  
61 02 49608428 or [hathor@bigpond.net.au](mailto:hathor@bigpond.net.au)

**Reinald Baumhauer - Germany**  
Fax: 049-89-41902871 or  
[r.baumhauer@mnet-mail.de](mailto:r.baumhauer@mnet-mail.de)

**Aurelio Lococo - Italy**  
Tel. e Fax: +049 8736130 or  
[aiss@shwachman.it](mailto: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 **1-877-737-4685** or e-mail us at [4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org) with your changes.

## Medical Scientific Advisory Board

**Blanche Alter, MD, MPH**  
National Cancer Institute  
Bethesda, MD

**Lynda Ellis, RN**  
GI/Nutrition Dept. Hospital for Sick  
Children, Toronto, Canada

**Michael Glogauer, DDS, PhD**  
University of Toronto  
Canada

**Alan Warren, MD**  
Laboratory of Molecular Biology  
Cambridge, England

**Peter Durie, MD**  
The Hospital for Sick Children  
Toronto, Canada

**Thierry Leblanc, MD**  
Hospital Saint Louis  
Paris, France

**Tarek Elghetany, MD**  
University Texas Medical Branch  
Galveston, TX

**Outi Makitie, MD, PhD**  
Helsinki University Hospital  
Finland

**Johanna Rommens, PhD**  
Hospital for Sick Children  
Toronto, Canada

**Marco Cipolli, MD**  
Gastroenterologist  
Cystic Fibrosis Center  
Verona, Italy

## SDSF

**Founder**  
Joan Mowery

### Board of Directors

Debbie Kadel - **President**

Blair Van Brunt - **Vice President /  
Fundraising Chair**

Kelly Bright - **Secretary**

Theresa Henle - **Family Service  
Coordinator**

Susan Utz - **Medical Librarian**

Alice Johnson

Jenny Jenuwine

Mailing Directory  
Peggy Lentz

Attorney  
Ann Bodewas Stephens

## 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.



*Cure For A Future*

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

ADDRESS SERVICE REQUESTED

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.