

NEWSLETTER

Winter 2004

Happy 2004! Happy 10-year anniversary to SDSI. Along with the start of a new year, comes quite a few changes. I am of the belief that "change is good". One of the first changes is the author of this editorial. I would like to begin by announcing that Nancy Ruick resigned as President of SDSI at the end of December. She has been involved with this organization for five years and has been the president for the last three. I would like to thank her for the endless hours she poured into this group. All of our children have benefited because of her hard work and dedication in furthering the cause. I was nominated and elected president by your board of directors of SDSI. I am honored to be able to step up and do my best for all those affected by SDS. I would also like to thank Kelly Bright and Doris Bull for their efforts on behalf of Shwachman-Diamond Syndrome. Both Kelly and Doris have resigned their positions on the board. As I announce the changes on the board, I challenge you to see where you fit in with this organization. Your board of directors changes leadership every so often and we are looking for future leaders. Get involved whether that means advocating for your child, raising money to find a cure for this life threatening disease, working on the newsletter, helping with the Angel Anna baskets, helping with grant writing, attending conferences to promote doctor education, volunteering to be a contact support person in your area or find out

how the board of directors operates to determine if you would one day fill one of the vacated slots. We have an outstanding group of physicians around the world working at finding a cure for this disease and as parents, grandparents, and friends of people affected, **we all have to do our part.**

For those of you that do not know me, allow me to introduce myself. My name is Debbie Kadel. I am married and live in Colorado with our three children. Brittany, our middle child, is 7 1/2 years old and is our SDS child. She has had a bone marrow transplant and too many other surgeries to mention here. I have been involved with SDSI since 1997. I have been on the board of directors of SDSI since January 2002. I look forward, with the help and support of many, to take this support organization to a new level. And in case you haven't noticed our name has changed from Shwachman-Diamond Syndrome International to Shwachman-Diamond Syndrome Foundation or SDSF. One challenge we still struggle with is when people see our name, they have no idea who or what we are. By adding the word foundation, we are hoping to clarify that from the beginning. Foundation signifies not only a non-profit group but also a group working on raising and donating money through grants to researchers, doctors, and scientists.

**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
Website: www.shwachman-diamond.org

We have had the privilege of supporting and serving many families in countries throughout the world that do not have a support organization in place. Although we have taken away the word “international” we plan to continue supporting these families wherever they are. The name change will not alter anything that we have done in the past or continue to do in the future concerning supporting families, whether it is here in the USA or elsewhere. What it will do is allow us to also look at the possibility of a Global SDS board. We have done a tremendous amount of work furthering the knowledge of this disease. It is my opinion and the belief of others that “many hands make light work”. I believe that in order to find a cure and better treatments for this disease we need to work collaboratively with the support organizations around the world and tackle this disease head on. I would like to congratulate those that have continued this effort in their own country and to encourage those that are thinking of starting up a new support group if there isn’t already one. We would be honored to help in this endeavor.

Thank you to all the physicians on our MSAB as well as physicians, researchers, nurses etc. around the world whom are reading this newsletter. We can do our part but without you, we cannot meet our ultimate goal, and this is finding a cure. I would also like to take time to thank all previous board members of SDSI.

I hope you realize the sacrifices these individuals made in the beginning getting this organization off the ground. We are ten years into the process and I can’t imagine what it will be like in another ten.

So in closing I ask, **what are you going to do this year for Shwachman-Diamond Syndrome?**

Best wishes to you and your families!

Debbie

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. 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 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

M5G 1X8

Angel Anna Baskets a “Smiling” Success!

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-SDS-INTL or contact me personally online at kroppejohn@aol.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 this project! With love, Julie Kroppe

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 and click on Products and Services or call 866-AXCANRX for enrollment information.

Update on Oral Diseases and Shwachman-Diamond Syn- drome

The first phase of Dr. Glogauer’s research on oral diseases in SDS is close to completion. The study has already run for almost two years but only 10% of SDS families have participated so far. Although the response rate is less than ideal, the study suggests that there are indeed oral health issues associated with SDS.

As far as teeth are concerned, SDS patients tend to suffer from more dental decay (cavities) than their non-SDS siblings. The primary dentition (baby teeth) also seems to be more affected by dental decay. Dental development also appears to be delayed. In addition, mouth-sores are also more common among SDS patients. The combined effects from poor oral health conditions clearly have a negative impact on a person’s general well-being. 44% of the SDS participants reported having pain when they eat.

In order to strengthen our conclusions which is important for educating health care professionals, more participants are needed before phase one of the study is closed (June 2004). If you are interested in the study, Dr. Glogauer can be reached by e-mail, michael.glogauer@utoronto.ca, for a new set of questionnaires. Your participation and support are required at this important time as we work to improve treatment and the quality of life of SDS patients.

The second phase of the study focuses on how oral health affects “quality of life”. Similar to the phase one, the current study also compares the results between SDS patients and controls (e.g. non-SDS sibling, friend or neighbor at similar age). The entire questionnaire takes less than 10 minutes to complete and the results can significantly strengthen the overall message. At present, phase two questionnaires are only mailed to those who participated in phase one. If you would like to help out as well, please don’t hesitate to contact Dr. Glogauer.

RESEARCH

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. **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 propensity), 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 at frederick-goldman@uiowa.edu.

PLEASE NOTE CHANGE OF ADDRESS:

**710 BRASSIE DRIVE
GRAND JUNCTION, CO 81506**

SEND ALL CORRESPONDENCE TO THIS ADDRESS

Identification of Genes Involved in Marrow Failure and Malignant Myeloid Transformation by Gene Expression of Bone Marrows from Patients with SDS and other Inherited Marrow Failure Syndromes

Dr. Yigal Dror, Director of the Marrow Failure and Myelodysplasia Program at The Hospital for Sick Children, is conducting a study on the identification of genes involved in marrow failure and leukemia in Shwachman-Diamond syndrome and other inherited marrow failure syndromes. It is unknown why and how these patients develop marrow failure and leukemia. Dr. Dror will use a new technology called DNA microarray. DNA microarray is a powerful technique, which can provide comprehensive information on many genes. He is going to analyze bone marrows from patients with Shwachman-Diamond Syndrome and other inherited marrow failure syndromes after consent is obtained from the patients and families. Using this powerful technique to analyze marrow samples from these patients, Dr. Dror will attempt to uncover mechanisms for the development of marrow failure, preleukemia and leukemia. In addition, many patients with Shwachman-Diamond syndrome and other inherited marrow failure syndromes undergo innumerable diagnostic tests for months or even years until accurately diagnosed. The results may serve as a solid basis for establishing an easy and quick tool to distinguish between the disorders and dramatically simplify the currently very complex process of establishing a diagnosis. Please contact Dr. Dror for more information yigal.dror@sickkids.ca

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

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

Inherited bone marrow failure syndromes (IBMFS) are rare disorders in which there is usually some form of aplastic anemia (failure of the bone marrow to produce blood), associated with a family history of the same disorder. Some of these conditions have typical changes in physical appearance or in laboratory findings which suggest a specific diagnosis. There are several well-described syndromes, which can be recognized by health care experts. There are also patients who are harder to classify, but who appear to belong in this category. Patients with these syndromes have a very high risk of development of cancer [Alter, Blanche (NCI)] (leukemia or solid tumors). At the moment we cannot predict which specific patient with an IBMFS is going to develop cancer. The NCI IBMFS [Alter, Blanche (NCI)] Cohort Study will enroll North American families in which at least one member has or had an IBMFS.

The web page "marrowfailure.cancer.gov" describes the study and provides contact information. By telephone, please contact Lisa Leathwood 1-800-518-8474 or you may also contact SDSF for more information.

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 of individuals with bone marrow failure. The study is still seeking volunteers, and anyone wishing to participate may contact the study coordinator for more information.

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:

Jennifer Ivanovich, M.S., Study Coordinator:
Washington University School of Medicine, Box 8100
660 S. Euclid Ave., St. Louis, Missouri 63110, USA
jen@ccadmin.wustl.edu, Phone 314-454-5076

Monica Bessler, M.D., Ph.D., Co-Director
Division of Hematology
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660 S. Euclid Ave., Box 8125;
St. Louis, MO 63110, USA
Phone 314-362-8807, email: Mbessler@im.wustl.edu

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Division of Pediatric Hematology/Oncology
Washington University School of Medicine;
660 W. Euclid Ave., Box 8208;
St. Louis, MO 63110, USA
email: Wilson_D@kids.wustl.edu

Established Shwachman-Diamond Groups

Shwachman-Diamond Syndrome Support - Australia

Contact: Joan Buchanan
61 03 5427 0645
email: buchanafam@bigpond.com.au
http://www.shwachman-diamond.org

Shwachman-Diamond Support-UK

Contact: Kim Wright
01 522 792039
email: kimwright@tesco.net
http://www.shwachman-diamondsupport.org

Italy Association for Shwachman Syndrome

Contact: Aurelio Lococo
email: aiss@shwachman.it
http://www.shwachman.it

Shwachman-Diamond Syndrome Canada

Contact: Karen Campbell
email: sdscanada@sympatico.ca
http://www.shwachman.org

Shwachman Syndrome - Netherlands

Contact:
email: koster.e@hccnet.nl
http://www.shwachman.nl/

REGIONAL PARENT CONTACTS

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

IN THE USA

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

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

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

Jenny Jenuwine - MI: (810)395-2358 or jengrsls2@bignet.net

Kelly Bright - TX: (409)738-2925

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

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

OTHER COUNTRIES

Kim Wright - England:
01522 792039 or kimwright@tesco.net

Lee-Anne Hayes - Australia
61 02 4968 9117 or cerridwen@kooee.com.au

Reinald Baumhauer - Germany
Fax: 049-089-41902871 or
Reinald.Baumhauer@T-online.de

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



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

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-**

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) or call our toll free number with your new address.

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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.

FOR YOUR INFORMATION

***SHWACHMAN-DIAMOND SYNDROME HAS MADE A
NAME AND ADDRESS CHANGE***

***PLEASE SEND ALL CORRESPONDENCE TO THE NEW
ADDRESS***

SHWACHMAN-DIAMOND SYNDROME FOUNDATION

710 BRASSIE DRIVE

GRAND JUNCTION, CO 81506 U.S.A.

FAX: 970-255-8293

EMAIL: 4SSKIDS@SHWACHMAN-DIAMOND.ORG

WEBSITE: WWW.SHWACHMAN-DIAMOND.ORG

1-877-SDS-INTL (TOLL FREE)