

NEWSLETTER

Summer 2004

Spring is gone and summer is in full swing. Thinking about time I often find myself wondering “where did the time go?” This edition of the newsletter is delayed and I am truly sorry! This is a very big job and I do worry about doing my best. I know, however, that I am a mother of a very special little girl and that she will always come first! I also know that all of you with children (adult or child) understand. It is amazing what we do for our kids. Our family has been at the mercy of a hip spica body cast for the last ten weeks. Brittany is 6 years post bone marrow transplant this July, but she is not cured from Shwachman-Diamond Syndrome. I think about that and I have to wonder what it must be like for the rest of the SDS parents and adult patients. We are currently at the mercy of this disease in one-way or another. We have a huge job!

I am very happy to announce the addition of three new board members, Blair Van Brunt, Susan Utz, and Alice Johnson. Blair Van Brunt will be our new Fundraising Chair, Alice Johnson will be our new Secretary, and Susan Utz will be family support. Blair and her family have raised over \$150,000 for SDS research. Alice organized the fundraising t-shirts that our group sold. Susan wrote an article for a previous newsletter about IEP's. I look forward to working with these wonderful ladies and am sure they will be a huge asset to this foundation.

This year I am proud that we as parents have funded several research projects. You can find those listed

on the financial page included in this newsletter. As more knowledge is gained, we are going to need more and more money to fund the scientists interested in further study and eventually cure. **I strongly urge you to see how you can become involved in this effort.**

If every family did just **one** thing a year, can you imagine what we could do? That one thing might be as simple as selling donuts. It could be sending in a donation of **any size** or sell “Dream For a Cure” bracelets that Jenny J. is making. You could do a letter writing campaign to friends and family. **If you don't know where to start but would like to help, please call 1-877-SDS-INTL.** Blair Van Brunt is available for any questions or ideas you may have regarding fundraising. She is also putting together “how to” packets of information for any families that would like them.

In closing, **don't hesitate. Get involved.** Wouldn't it be something if it were our generation that helps fund research that finds the cure? Think of what that would mean to our children/grandchildren's future. I extend to you and your families my warmest thoughts and best wishes!

Debbie

**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

Welcome Susan Utz

Hi! My name is Susan Utz and I have recently accepted a position on the board of SDSF. I am humbled by the prospect of following in the footsteps of those who have previously served or are continuing to serve on the board. Until now, I did not realize the amount of time and effort that others have put into this organization in order to provide funds for ongoing research and parental support for SDS. My gratitude goes out to all those who have worked so hard for this organization over the years. I am the mother of a 15 year old son with SDS, named Logan. We live in southern Indiana near Louisville, KY. I work full-time in the public schools as a Speech/Language Pathologist, and I teach piano lessons part-time. My hobbies include: traveling, spending time with friends and family, playing the piano, and going to shows and concerts. My husband (Logan's stepfather) is named Curt. I became involved in SDSF several years ago and have appreciated getting to know many other parents of children with SDS. I have also been grateful for the chance to learn about ongoing research and obtain valuable information through SDSF. We were fortunate to have had the opportunity to attend the SDSF family conference this past summer and to have met many of the wonderful people involved in this organization. I look forward to serving on the board and hope to be able to contribute to its operation.

Welcome Blair Van Brunt

My name is Blair Van Brunt and I am honored to have just joined the Board this month after many years of working with the Board on fundraising. I am married and am an at-home-mother of three with a clothing and jewelry business (www.chakra-tees.com) that I run out of our home. I am on two other boards right now (one town groundwater protection committee and one Peace Abbey non-profit charity) that keep me busy but not overwhelmed and I am ready to contribute all that I can to this important endeavor.

Our children are ages 15, 11, and 9 and it is our 9
2 year old daughter, Gracie, who has SDS. She

year old daughter, Gracie, who has SDS. She was diagnosed at age 2 due to marrow failure after many illnesses and hospitalizations. She recovered from the failure but continued to be hospitalized for many different infections and illnesses until the age of 5. And then she started to improve month by month. It is now 3 years and 4 months that she has been free of hospitalizations (not illnesses) and our lives are certainly easier.

But it was back in January 1997 that we found SDS on the internet (thanks to Joan Mowery and the first board members), which helped us learn more about and understand this disease. From there we attended the first family conference, met Johanna Rommens (our genius geneticist) and the rest is history for us. We have dedicated our efforts to help our daughter and all others with this disease by fundraising for medical research towards a possible cure one day. Starting in the summer of 1998, we have raised between \$20,000 - \$40,000 each year for SDS medical research projects and we will continue to do so because we believe that we all have a role in our children's lives and this is ours.

I can think of no better way to spend my time than towards an organization, which benefits not only my daughter but all the people with SDS. After all, that is what this is all about - THE SDS PATIENTS! As they say, it is the unlocked information of the rare diseases that help determine keys for the diseases that afflict the larger populations. I am excited to work with Debbie Kadel, whom I have much respect for, and together with the rest of the Board members and the members of the Medical Advisory Board we hope to make a difference in your child's life and yours. So find your niche and go for it! Is it speaking with other parents, writing articles, bookkeeping, grant writing, web site managing, fundraising? Let us know. I look forward to serving on this Board.

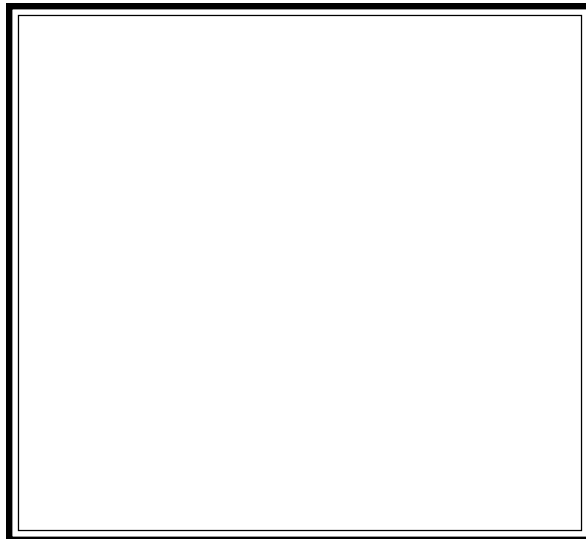
Thank you,
Blair Van Brunt

Welcome Alice Johnson

Hello, my name is Alice Johnson. It is an honor to be asked to join the Board and I have accepted the position of Secretary. I am very excited about my roll in the foundation and look forward to contributing to it and the families.

I have been involved with SDS since 1997 when our son Clint, 9 years old, was first diagnosed. I am married to Mike and we also have a younger son, Luke, 7 years old. We live in Lannon, Wisconsin, outside of Milwaukee. I work part-time at an auto dealership. I love to travel, sew, go to the movies and spend time with family.

**SUPPORT SDSF
WITH
PURCHASING
BRACELETS
MADE BY
JENNY JENUWINE**



**GO TO PAGE 5
FOR MORE INFORMATION**

United Way or Combined Federal Campaign

An easy way to donate to SDSF

If you would like to help our SDS children by donating to your local United Way or if you are a federal employee your local CFC, it is easier than you think. It would be a great opportunity for you to contribute and help our cause. If you already donate to the United Way or CFC in your area, you might check with your local chapter and see if they will add SDSF as your charity. Our Combined Federal Campaign number is 1329. You may have to start now in order to get everything in order for the start of your yearly campaign. Some companies offer to match any funds and that might be an option. You could also post or email a memo to fellow employees asking if they would be interested in writing in SDSF as their charity of choice also. With this you could include a little information about SDS and how it affects your child.

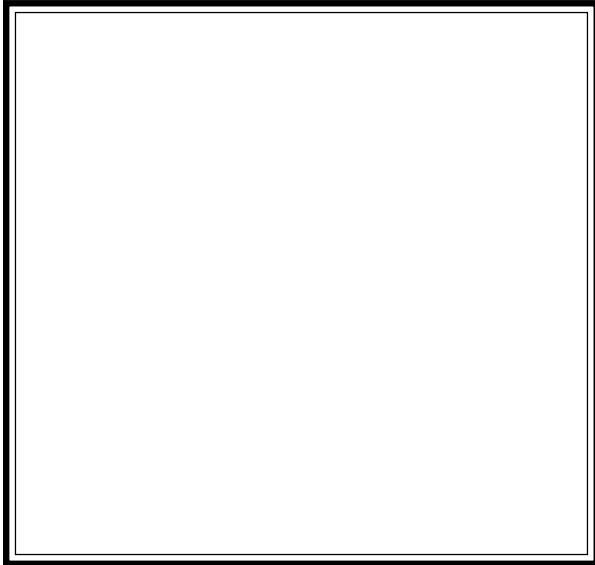
If you have any questions, please don't hesitate to contact us. You can either call 1-877-SDS-INTL or email us at 4sskids@shwachman-diamond.org.

NEW ADDRESS FOR SDSF

SEND ALL CORRESPONDENCE TO:

Shwachman-Diamond Syndrome Foundation
710 Brassie Drive
Grand Junction, CO 81506

Fax: 970-255-8293



Alex S. Turnquist
May 7, 1991 - March 19, 2004

Alex was first diagnosed with Shwachman-Diamond Syndrome when he was 9 months old. He was in the sixth grade and doing well. For many years we were worried about his bone marrow because he had decreased cell counts, decreased cellularity, increased blasts and chromosomal abnormalities. We always imagined that if something happened to Alex, it would be related to his having Shwachman-Diamond Syndrome. On March 26, 2004, Alex slipped and fell on the ice. Our lives were changed forever. He spent the rest of his days fighting to survive. The doctors removed a blood clot in his brain, placed a shunt to relieve the pressure. He fought until the end. His brain pressure kept going up and he died peacefully in his Mom and Dad's arms on March 19, 2004.

Alex loved to build things with legos and K'nex! He was always building! He also enjoyed his game boy, puzzles, listening to music, helping his Dad with projects, and keeping Mom organized in the house. He was part of the youth group at Gethseman Lutheran Church in Upsala, MN. He brought so much joy to so many lives. He helped out at the camp-ground and touched the lives of so many people. He would help get firewood for people and even run to get them soda from the machine (as long as he could keep the change!).

Alex brought joy to many lives and will always be in our hearts. He will always be our hero. His life was full of doctors, pokes and illness yet he still lived life to the fullest. Nothing could stop him from doing the things he loved. Here is a poem we used as part of his memorial.

We love you Alex,
Jay, Laura, Carlee and Drew

I'm Free

*Don't grieve for me, for now I 'm free
I'm following the path God laid you see.
I took His hand when I heard Him call
I turned my back and left it all.*

*I could not stay another day
To laugh, to love, to work or play
Tasks left undone must stay that way
I found the peace at the close of the day.*

*If my parting has left a void
Then fill it with remembered joys--
A friendship shared, a laugh, a kiss
Oh yes, these things I too will miss*

*Be not burdened with times of sorrow
I wish you sunshine of tomorrow
My life's been full, I savored much
Good friends, good times, a loved one's touch.*

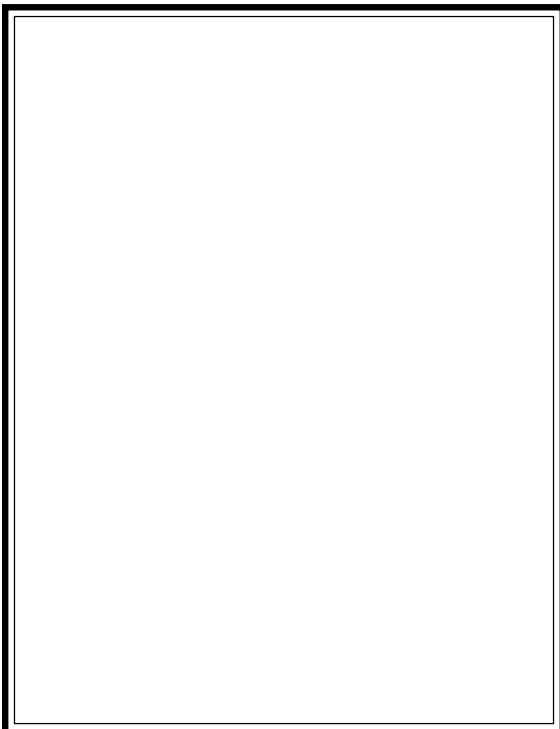
*Perhaps my time seemed all too brief--
Don't lengthen it now with undue grief.
Lift up your hearts, and peace to thee--
God wanted me now; He set me free.*

FUNDRAISER FOR SDSF

I am making SDSF bracelets made of Swarovski Crystal and Sterling Silver with the SDSF logo as a fundraiser. Each bracelet is priced at \$30.00 plus shipping. I have several colors to choose from and can customize for size. I have available Siam (red), Garnet (dark red), Montana (dark blue), Light Blue, Light Sapphire, Padparadscha (peach/pink), Tanzanite (green/black), Topaz, Rose, Light Rose, and of course Clear. These bracelets are beautiful and help show support for SDSF. The money will go to help SDSF families and to research. Please try to sell these. The pictures will be available on our web site.

Thank you in advance for your support.

Jenny Jenuwine
jengrls2@bignet.net
810-395-2358



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.

Axcanscandipharm, 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.

Financial Status of SDSF from January 1, 2003 - June 30, 2004

Income		Expenses	
Beginning Balance:	\$ 32,168.96	Conferences:	
Donations:		ASH 2002 Conference	\$ 910.93
Anna Angel Baskets	\$ 500.00	ASH 2003 Conference	\$ 3,128.02
Ebay Proceeds	\$ 81.00	Family Conference	\$ 18,072.54
Patrick Kroppe	\$ 957.78	Scientific Conf. - June 2003	\$ 10,247.44
Research	\$ 550.00	Family Support:	
United Way & CFC	\$ 6,660.54	Newsletters	\$ 9,946.00
Goff Children	\$ 78.00	Oral Project	\$ 101.25
General	\$ 15,219.61	Anna Angel Baskets	\$ 1,370.98
Fundraising:		New Family Packets	\$ 1,342.28
2003 Superbowl	\$ 9,500.00	General Postage & Delivery	\$ 1,172.87
2004 Superbowl	\$ 7,011.00	Printing & Reproduction	\$ 1,714.44
Brittany Kadel Birthday	\$ 1,665.00	Research Grants:	
Dance For A Cure	\$ 5,070.00	Dr. Kerr Research Grant	\$ 20,000.00
Dylan Kolar	\$ 3,270.00	Dr. Liu SBDS Research Grant	\$ 30,000.00
Brett Riley	\$ 2,520.00	Yigal Dror Research Grant	\$ 25,000.00
Gracie Fund	\$ 34,975.00	Hematopathologic Grant	\$ 13,200.00
Michael's Goal	\$ 40,000.00	Soll Research Grant	\$ 30,000.00
Troy DeBoer	\$ 5,642.50	Other Business Expenses:	
Jenuwine Bracelets	\$ 570.00	Board Conference Calls	\$ 2,350.62
Golf Outing Proceeds	\$ 950.00	Office Supplies	\$ 274.99
T-shirts and cards	\$ 485.00	Dues & Subscriptions	\$ 490.00
Johnson's	\$ 1,600.00	Telephone	\$ 1,325.68
Grants:		Misc. Business Expenses	\$ 1,653.84
Newsletter	\$ 8,100.00	Bank Fees and late charges	\$ 80.00
Unrestrictive Grants	\$ 15,000.00	Payroll, Legal & Prof. Services	\$ 5,035.00
2003 Family Conf. Registration	\$ 1,269.00	Payroll	\$ 1,309.06
		Federal Taxes	\$ 239.44
Total Income:	\$ 193,843.39	Total Expenses:	\$ 178,965.38

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.

Identification of Genes In- volved in Marrow Failure and Malignant Myeloid Transfor- mation 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

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

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

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

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

Research on SDS white blood cells at Iowa

During the past 20 years, the Soll laboratory has been interested in how animal cells migrate, and has developed computer-assisted systems to quantitate these behaviors. Recently, with funds from the National Institutes of Health and SDSF, we have used these systems to examine the behavior of neutrophils of SDS patients. We hypothesized that because SDS patients have an increased incidence of infections, often independent of a low white blood cell count, the neutrophils may be defective in behavior. Because we have learned a great deal about which molecules are involved in particular behaviors in chemotaxis, we have used this information to elucidate and define the defect in SDS neutrophils, which we believe will lead to predictions of defective molecular mechanisms in SDS neutrophils. We have now found a particular defect in SDS neutrophils. While SDS neutrophils are normal in all aspects of basic chemotactic behavior, they cannot read chemical gradients. This suggests that SDS neutrophils are less capable of being attracted to sites of inflammation, which could explain why many SDS patients have an increased rate of infections. The defect in reading chemical gradients was observed in every SDS patient tested, and was absent in every control patient tested. The defect is specific to a class of molecules involved in polarizing (orienting) the cell in the correct direction in gradients of signals (toward the site of inflammation). To study this defect at which such polarity has been studied at the molecular level, we identified and cloned the SDS gene in two model systems in which such polarity has been studied at the molecular level, a yeast and an amoeba. The SDS gene in each organism has been mutated to reflect the defects in the same gene in SDS patients, and the normal genes in the yeast and amoeba are now being replaced with the mutated gene. We can thus study the function of the SDS gene in these models to gain insights into the defects in SDS patients, and possible methods for reversing the defect. We thank SDSF for continued support, and SDS patients for their participation in this research, and hope to have more to report in the near future.

David R. Soll
Fred Goldman
The University of Iowa

Update on Oral Diseases and Shwachman-Diamond Syndrome

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.

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 jengrls2@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-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) 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**
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**The children and adults you are helping THANK YOU for caring.
Your generosity in giving is greatly appreciated.**

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

Thank You to our Donors

(donations January 21, 2004 - June 19, 2004)

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Third International Congress on Shwachman-Diamond Syndrome
June 26-29, 2005
Robinson College, Cambridge, England

Sponsored by Shwachman-Diamond Support (UK)
with support from the University of Cambridge

What are your plans for summer 2005? Come to the city where Watson and Crick announced the DNA double helix structure in 1953, and learn how much that discovery has influenced our understanding of this rare disease!

Since the identify of the gene SBDS was published 50 years later in 2003, there has been rapid progress in defining the role of its protein product in the cell, with important and wide-ranging implications for scientists and clinicians in haematology, oncology, gastroenterology, nutrition, growth and development, and psychology, among other disciplines.

Robinson College has modern facilities and is centrally located. Cambridge is at its best in summertime. You can soak up the history and keep right up to date with the science, all at the same Conference! The inclusive registration fee will be reasonable and the scientific and social programmes exciting. Look for the Second Announcement and Call for Abstracts later in 2004 but put the dates in your diary now.

Topics (Papers are invited on the following topics)
Oral and Poster presentations, discussion, roundtables

1. What have we learned about SDS? Clinical Features|Genetic Diagnosis
2. Where are we now? Epidemiology|Molecular biology|Management of clinical problem: Gastrointestinal|Nutritional|Blood & Bone Marrow|Growth and skeletal|Oral & Dental|Developmental and psychological
3. Where are we going? International collaboration|Registries & Databases|Prospects for new treatments:
Genetic|Immunogenetic|Pharmacological

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NEWSLETTER RELEASE
REMEMBER WHEN IT WAS SAID THAT COMPUTERS WOULD SAVE
PAPER?

Well, we want to prove that teory correct by switching to an electronic newsletter!! Don't panic if you don't have access to email and the internet, we will still send you a printed copy. Everyone, with or without internet access, please fill out the form below to let us know what your status is. We will publish this announcement for the next two newsletters to make sure that we don't miss out on anyone. After that, if we haven't heard from you, we will assume that you do not want the newsletter at all and we will not attempt to send it either snail mail or email. Electronic newsletters will go out effective January 2005.

THINK ABOUT IT! Saving trees, conserving our environment, saving money to be spent on medical research or family support or whatever is needed for the organization. We can even add many more people, doctors, hospitals, etc. to the "mailing list" without adding extra costs in printing, paper, and postage. This means more education for others about SDS and that can translate into many positive benefits for all of us. We are excited to pursue this. We hope that you are too. If you have any questions or concerns, feel free to call 1-877-SDS-INTL (737-4685) toll free. Or email 4sskids@shwachman-diamond.org.

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710 BRASSIE DRIVE
GRAND JUNCTION, CO 81506

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

ADDRESS SERVICE REQUESTED

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