

NEWSLETTER

Fall 2004

Happy Holidays~

As I think about the upcoming months and how busy they are, I try to enjoy the quiet times. There is so much to be thankful for it is hard to know where to start. First, I would like to thank all of you that read this newsletter. We have received a great response from people who would like to continue to read the newsletter either in the current format or would like to save a few trees and receive it electronically. Not only did we receive the newsletter form, we also received a number of donations along with it.

Secondly, I am thankful everyday for this organization. Without the many that came before the current executive board and laid the groundwork and foundation to what this organization has become, I can't imagine where this disease would be today. We provide many families and physicians with information and support in dealing with Shwachman-Diamond Syndrome. This organization has helped launch other support groups around the world so that we can work together to better support individuals dealing with this disease.

Thirdly, I am thankful for all the families and individuals that donate to this worthwhile cause. We have always prided ourselves on not charging families for our services and without all of you that would not be possible. From time to time we have had some very low years and have been forced into charging a small fee for our last family conference, but we are hoping

that won't have to happen again. We all need to do our part to keep this organization running and to fund the research necessary to one day find a cure for this disease. Therefore, we are going to start having a yearly one-time fund drive and hope families will consider contributing to SDSF. Enclosed in this newsletter is a self addressed envelope for you to send in your donation. You might ask your company if they have a donor match program.

I would like to give special recognition to the families that have had fundraisers on behalf of SDS this year. The Kolar family had a first annual 5K run that raised \$35,000, they also had a block party, a golf outing, and ordered 24 bracelets in honor of Dylan Kolar. The Van Brunt family held their annual Barn Dance and raised \$30,000, you can read more about their fundraiser on page 4 of this newsletter. The Jenuwine family held their annual Super Bowl party which is always a success. The Kroppe family held their annual Dance For A Cure, the Johnson family had a Sock Hop and silent auction, the DeBoer family had their "Shoot for Shwachman" basketball fundraiser, and Joan Mowery worked with Ebay and sold some items to benefit this foundation. **Thank you families for working so hard to help us fund necessary research and provide support to families all over the world!!**

May your holidays be filled with much laughter, love, good friends, and fond memories. God Bless each and every one of you, Debbie

**Shwachman-
Diamond
Syndrome
Foundation**

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Grand Junction, CO 81506 U.S.A.
1-877-737-4685 (Toll Free)
Fax: 970-255-8293
E-mail: 4sskids@shwachman-diamond.org
Website: www.shwachman-diamond.org

MEDICAL LIBRARY DEVELOPMENT

Have you ever wondered about the research being done toward finding a cure for Shwachman-Diamond Syndrome? Have you ever wondered about studies on any of the various symptoms of SDS? Are you interested in reading more about the studies that led to the discovery of the SBDS gene or the implications of this discovery?

The Shwachman-Diamond Syndrome Foundation (SDSF) is working toward helping you and/or your child's physicians find answers about research topics relating to SDS. We are currently developing a medical library which will contain articles and/or abstracts on various aspects of research in the field. This database of information will have a variety of categories such as pancreatic function, bone marrow failure, bone marrow transplantation, gene studies, skeletal complications, and disease management. We hope to be able to direct you toward studies of interest that help you fully understand your child's disease. We continually strive to update our webpage and that of others in the medical field, such as WebMD and MedLine Plus, so they may have the most recent information available on SDS.

Further information will be in our newsletters regarding the developments and retrieval of information about current research.

MEDICALERT BRACLET FOR PEACE OF MIND

We all sleep well at night knowing that our 9 year old daughter wears a MedicAlert bracelet. She is allergic to 5 classes of antibiotics. We know that if she ever is in an emergency when we are not with her, the doctors at hand will call the toll free number listed on her bracelet to find out more about her condition and allergies. I have listed with the service not only the specific antibiotics that she is allergic to but what the name of her disease is, what medications she is taking, and what her typical blood counts are for reference. Because she is

chronically neutropenic, it is important that the doctors know this is a normal condition for her.

You can list whatever is important to a doctor treating this patient when the parent might not be there. In our case, we couldn't fit a lot on the bracelet so we listed the name of the disease and "please call for list of allergies". Teachers, coaches, school nurses, and friend's parents have ease of mind that she will be well taken care of if there is an emergency.

Below are some of the services provided by MedicAlert and you can call 1-800-625-3780 for more information. The cost is \$35.00 for the first year and \$20.00 each year thereafter.

- * **24-hour Emergency Response Service** - supported by on-site Registered Nurses, MedicAlert will relay your vital medical information to emergency personnel.

- * **24-Hour Member Services Contact Center** - Make free, unlimited updates to your file, order a new emblem, or ask a question at any time. Someone who cares will always answer the call. Providing support for our members 24/7, around the clock.

- * **Family Notification** - MedicAlert calls your family contacts and notifies them of your situation, so you won't be alone in an emergency.

- * **Medication Dosage Service** - MedicAlert stores your medication dosages, which emergency medical personnel often need.

- * **Online Access and Updating** - Make instant changes to your personal record, including change of address, adding/discontinuing medications, and new physician information.

- * **Information Storage** - Store information such as the brand, model, and deactivation instructions for medical devices and implants.

- * **Advance Directives** - You can also store your state-recognized written Advance Directives and Do-Not-Resuscitate orders with MedicAlert for an additional \$20.00 per year.

- * **Privacy** - All medical and personal information is stored in a secure electronic health record database, and is never distributed to unauthorized personnel or organizations.

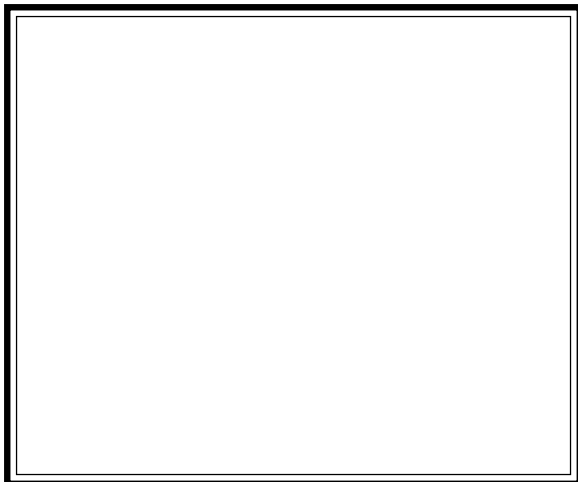
WHAT A SUCCESS!! DREAM FOR A CURE BRACELETS

I want to thank everyone who has helped make this a huge success - keep up the good work!! We have already raised over \$1,800.00!!!

The bracelets are made of Swarovski Crystal and Sterling Silver beads, it also has a dream message bead and the SDSF logo on a silver charm. I have several colors to chose from and can customize for size. The colors available are Siam (red), Garnet (dark red), Montana (dark blue), Light Blue, Light Sapphire, Padparadscha (peach/pink), Tanzanite (green/black), Topaz, Rose, Light Rose, and Clear. I also have birthstone colors available. These bracelets are beautiful and make great gifts. You can order your bracelet in honor of your child and a special card will be sent with the bracelet as well as being listed in our newsletter. What a great way to support all SDS children and help with our Dream For A Cure.

To order just send a check or money order for \$33.00 (shipping and handling included) with the color and size of the bracelet to me. Please allow 2-3 weeks for delivery . Sorry NO COD's or credit cards accepted. **All profits go to SDSF.** Thank you for your continued support!!

Jenny Jenuwine
15028 Hough
Allenton, MI 48002
jengrls2@bignet.net
810-395-2358



ELECTRONIC NEWSLETTER UPDATE

In our zeal to save money and trees.....

Effective **MARCH 2005**, SDSF will start to send the newsletter electronically or snail mail (USPS) for those of you who have requested to continue to get the newsletter.

In order to receive the newsletter, either electronically or thru the postal service, you must complete the form on page 15 to let us know how you would like to receive the newsletter. **We have recently learned that anyone who has either HOTMAIL or YAHOO as their email hosts may not be able to receive the newsletter electronically. Our newsletter will be OVER 1 megabyte in size and some email hosts will not let you have attachments that big!!!** Please check with your internet service provider first and then let us know what your choice is. After March 2005 if we have not heard back from you, you will not be receiving the newsletter at all.

Everyone, with or without internet access, please fill out the enclosed form (on page 15) to let us know what your status is.

If you have any questions or concerns, feel free to call 1-877-737-4685 (toll free), or email 4sskids@shwachman-diamond.org.

Please take a moment to fill out the form on page 15 the trees will sing and the bank account will soar!!!

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 1-877-737-4685 or contact me personally online at jkroppe@wowway.com or call me at (248) 619-9316. I will be glad to answer any questions and I appreciate any and all input. Thank you to the many families who have contributed to 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

BARN DANCE FUNDRAISER

In our daughter’s school application she called it a family tradition, our friends call it the party of the year, and we call it a fundraiser. I am talking about the annual Barn Dance we have at our home (barn) to raise money (\$25,000-\$40,000) for Shwachman-Diamond Syndrome Foundation. It all started when my husband, Brad, and I learned in 1997 at the first family conference in St. Louis that Dr. Johnanna Rommens needed \$100,000 over three years to get the research started for finding the SDS gene. We were there on behalf of our daughter, Gracie, who is now 10. Brad talked with his band (Sabaka) about it and voila.....a plan was beginning to emerge. In a few months and many phone calls and meetings, we had picked a date, created a guest list, and planned an evening that we now look forward to every year.

We live in an old 1850 house with a separate barn that is just perfect for setting up a band, bar, and dinner in with a donated tent attached (we do this ourselves). We invite approximately 570 friends and usually welcome 160-180ish to our house. Out of those invited, I would say about half donate, whether they come or not. Everything has become a well-oiled machine by this point, but in the beginning I had to put a lot of parts together to form the whole. Almost everything is donated, whether it is food or time. The band is called Sabaka and my husband, Brad, is a lead guitarist and vocalist and I am a chick singer on a few songs. We have friends who own a restaurant (Tennessee’s BBQ) and they donate the dinner, another friend is the brew master of a microbrewery and he donates the beer, another friend donates flowers for the centerpieces on the tables, and another friend owns Polar Seltzer and donates mixers. I ask about 10 people each year to donate one bottle of booze to create the makings of a stocked bar (we buy the rest) and I get two-five cases of wine from two local liquor stores. I then get couples to man the bar and they find their own replacements when they are ready to dance. We pay for the porto-potties and table and chair rentals plus any extra things that always seem to crop up.

The printer (a local Sir Speedy) donates the printing (I buy the paper) for the invitations and the “Program

Of Thanks” that thanks everyone who helped with the event. I ask friends to help serve the dinner, help clean up, and just help me survive.

No doubt about it - throwing a fundraiser is a lot of work. Many details to keep track of, many people to organize, and many thank you letters to write. But, we wouldn't trade any of this work in for an easier time. By bucking up and asking our friends for help, we all feel better. They feel good about contributing to a cause where 100% of their donation goes to a streamlined, effective organization and helps their friends, and we obviously feel good about helping scientific progress for SDS. We probably spend \$1,000 of our own money and we receive a tax deduction letter from SDSF for these expenses. But there are many ways to keep the costs down or non-existent. This type of fundraiser just works for us.

My husband and I look at Gracie's illness as a positive opportunity to live, learn, and love. God bless all of you with children and adult children with SDS. We realize that everyone has a part to play and we all play them differently. It isn't easy when a family is in the middle of an acute time of illness or is chronically tending to their child in need to think about fundraising. We just wanted to share how we have raised money and what motivates us. Thanks for reading this article about how we have been playing our part.

United Way or Combined Federal Campaign An easy way to donate to SDSF

It is a lot easier than you think to donate to your local United Way or CFC. It would be a great opportunity for you to contribute and help our cause. If you already donate, check to see if your local chapter will add SDSF as your charity, our CFC number is 1329. Some companies offer matching funds, you could post/email a memo to fellow employees asking to contribute to SDSF and include a little information about SDS and how it affects your child. If you have any questions, please don't hesitate to contact us at 1-877-737-4685 or 4sskids@shwachman-diamond.org.

CALLING ALL DONATIONS

Have you ever felt that you wanted to contribute to this terrific organization but not sure how? Everyone can play an important part and it doesn't have to be thru a big fundraiser, cash donation, or asking anyone for money. Let us help you with some easy ideas. SDSF is always in need of:

Angel Anna basket items

matchbox cars coloring books & crayons
art supplies new stuffed animals
children's movies and books for all ages
small toys for boys and girls

Pre-Paid phone cards

for families with members in the hospital
(\$25.00 and \$50.00 are most popular)

Please send any Angel Anna basket items and Pre-Paid phone cards to:
Julie Kroppe
3714 Sandburg
Troy, MI 48083

Pre-Paid Card to Staples or Office Max with instructions for the card to be used for SDSF only.

Please send office supplies to Administrative Assistant to the Board,
Kim McDowell
2334 Rolling Ridge Drive
Avon, NY 14414

Please be assured that we are an accredited and audited 501(c)(3) organization and uphold all requests honorable. You will receive a letter describing your donation with appropriate amount listed for tax deduction purposes.

Thank you for considering pitching in. We are all in this together to help all patients with this disease. Many hands make light work!!

RESEARCH

SKELETAL FEATURES OF SHWACHMAN-DIAMOND SYNDROME

Outi Makitie, Lynda Ellis, Peter Durie, Johanna Rommens, William Cole
Hospital for Children and Adolescents, Helsinki University Hospital, Helsinki, Finland and The Hospital for Sick Children, Toronto, Canada

Shwachman-Diamond Syndrome (SDS) is a complicated disorder which affects a number of different organs, including the bone marrow, pancreas and skeleton. Changes in the skeleton were first reported in 1967. Later reports suggested that skeletal changes were only present in some patients with SDS. Since the previous findings were based upon limited information in a small number of patients with SDS, we completed a study to clarify the characteristic skeletal features in SDS, to find out how commonly they are present and what changes are happening with increasing age. We analyzed all available X-rays from 15 patients with SDS whose diagnosis had been confirmed by genetic testing. In 10 patients repeated X-rays were available as the patients grew up. Unlike all previous reports, we found that skeletal changes can be seen on X-rays of all patients with SDS. However, the type and severity of changes varied a lot from person to person, and also changes with increasing age. The typical X-ray changes include:

Delayed bone maturation

In the first years of life the growth plates, which are at each end of the long bones, appeared later than normal and looked immature. This is an important observation, because growth plates are responsible for making bones grow in length. However, delayed bone maturation tended to normalize as the children grew older.

Abnormal appearance of the growth plates

Generally, the bone at the base of the growth plates was wider than normal and irregular in appearance. While, the growth plate abnormalities were very variable in severity, they tended to become more prominent with advancing age. In early childhood, similar changes were observed in the ribs which often appeared abnormally short. In older children the growth plate changes were most prominent at the knees and hips.

It is important to stress that, for most people with SDS, X-ray changes that are seen in the skeleton cause no problems. In a minority of people with SDS, however, changes in the skeleton can have consequences. For example, in some patients, abnormal growth plate development can lead to a change in the angle of the hip bone which may require surgical treatment. In others, abnormal growth of bones at the knees can cause curving of the legs. The X-ray appearance of the growth plate appears to be unique for people with SDS. Because it differs from those seen in other conditions affecting this region of the skeleton, X-ray changes provide important diagnostic information.

Osteoporosis

In almost all of the patients included in this study, the bones appeared thin and low in mineral content. This is called osteoporosis. In a few adult patients we examined, X-rays of the spine also showed signs of osteoporosis. A crush fracture of a vertebra has been described in occasional patients with SDS. Osteoporosis in SDS may partly be due to the nutritional problems but it is more likely to be another poorly understood feature of the defect in bone development.

Summary

The results of our study suggest that X-ray changes of the skeleton are present in virtually all patients with SDS. However, these changes vary considerably from person to person and with age. Therefore, in addition to the known pancreatic and bone marrow problems, we now feel that these unique skeletal changes are useful for establishing a diagnosis of SDS. Since some of the skeletal features change with advancing age, we now recommend that all people with SDS have complete bone X-rays at diagnosis and that regular screening be continued as part of routine care. Sufficient intake of calcium and vitamin D is important in the hope of preventing osteoporosis. Further studies of a larger number of people with SDS are needed to work out: how frequently complications occur; when needed what surgery is best; and whether treatment can help prevent osteoporosis.

This article is a brief summary of a recent publication "Skeletal Phenotype in Patients with Shwachman-Diamond Syndrome and Mutations in SBDS" *Clinical Genetics* 2003;65:101-112

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.

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,

(continued from page 7)

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.

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.

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 W. Euclid Ave.,
St. Louis, Missouri 63110, USA
Phone: 314-454-5076
jen@ccadmin.wustl.edu

Monica Bessler, M.D., Ph.D., Co-Director
Division of Hematology
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Washington University School of Medicine;
660 W. Euclid Ave., Box 8208;
St. Louis, MO 63110, USA
email: Wilson_D@kids.wustl.edu

New SDSF Brochures Now Available

The new Shwachman-Diamond Syndrome brochures have arrived and are available for anyone who needs them. They are a good tool for fundraising and for people who are interesting in finding out what we are all about.

If you would like to have some brochures, please contact us at 1-877-737-4685 or email us at 4sskids@shwachman-diamond.org, with your address and how many you would like sent to you.

We would like to extend to all our SDS families

a safe and healthy holiday season.

Merry Christmas

and

Happy New Year

Best Wishes

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 an 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.

Medical Scientific Advisory Board

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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 theory 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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1-877-SDS-INTL

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