

# NEWSLETTER

Spring 2005

Happy 2005~

What an honor and privilege it is to announce the amount of money raised through our first annual fund drive. It was our hope to raise between \$5,000 and \$10,000, we never expected to reach \$17,014! What an amazing group of people you are!! Thanks to each and every one of you for your help in the continuing success of this organization. Without you, we couldn't do what we do for this disease.

I would also like to congratulate the Jenuwine's on another successful super bowl fundraiser, this years total is over \$8,000. Thank you for your continued support.

Last year we funded several research projects, that are key to unlocking the mysteries of this disease. We funded Dr. Soll and Dr. Liu both for \$30,000. We also approved continued funding for Dr. Dror, without your donations this would not be possible.

This summer we will help sponsor the Third Scientific Congress on Shwachman-Diamond Syndrome hosted by the United Kingdom support group. We are very excited to be an active part in such an important meeting. Physicians and Scientists from around the world will attend to discuss this disease that affects so many of our children and loved ones.

There will be an opportunity for the support groups to meet and work on furthering our relationships to bond and work together for those affected by SDS. For more information on the conference, you can contact Kim Wright, President of SDS UK, at [kimwright@tesco.net](mailto:kimwright@tesco.net) and this newsletter and our website.

If you are wondering why you recieved a copy of the newsletter snail mail this time, we decided to do one more mailing. **This is absolutely the last chance to stay on our mailing list.** Thank you to those who have sent back the response forms, **if you have not responded you will be removed.**

Please take a moment to fill out the form on page 15 and either mail it back or at [4sskids@shwachman-diamond.org](mailto:4sskids@shwachman-diamond.org) with your preference for receiving the newsletter.

Best wishes to you and your families~  
Deb Kadel

**Shwachman-  
Diamond  
Syndrome  
Foundation**

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

## SDSF TO EXHIBIT AT DIGESTIVE DISEASE WEEK

Two or three representatives of the Board of SDSF will be attending and exhibiting at Digestive Disease Week, a national trade show for gastroenterologists being held in Chicago from May 15-18, 2005. SDSF has never attended nor exhibited at this conference and we are looking forward to the opportunity to meet and greet GI's from all over the country. Hopefully this will give us the chance to get SDS information into the hands of doctors who have patients that either have been diagnosed or can be properly diagnosed with SDS. We will be handing out our brochures and medical articles, we want to arm these doctors with as much information as possible to give to every patient who comes to them with any symptoms of SDS for a chance to get a proper diagnosis. The secretary of the board, Alice Johnson, and her husband are working hard to put together CD's with pertinent medical articles transcribed into them to hand out, and Kim McDowell, assistant to the board, will be amassing hundreds of brochures to hand out as well. Jenny Jenuwine has tapped upon her sister to create a sign for the booth as well as other signage that will help attract attendees to our booth. We feel fortunate that even more awareness will be brought to SDS (which is great for not only attracting researchers but funding as well) by Dr. Peter Duri, an advisor on the Medical Advisory Board of SDSF, who will be holding a symposium on SDS during this conference. Dr. Durie has this to say about the conference "DDW is the gastroenterology equivalent of ASH (American Society of Hematologists). Approximately 7,000 gastroenterologists are expected to attend the conference, primarily from the US and Canada, but there is good representation from around the world. The majority of attendees are gastroenterologists who care for adults--most of whom don't even know how to spell Shwachman-Diamond let alone know anything about it!! For this reason--if you manage to "nab" a few of them, you will help to raise awareness. In addition there will be a fair number of pediatric gastroenterologists attending, only some of whom are familiar with SDS."

If anyone speaks with their GI prior to May, let him/her know that Dr. Durie is presenting his symposium on SDS and that we will be exhibiting at the DDW in Chicago and **urge them to attend**. DDW will be held at McCormick Place, Lakeside Center, Chicago, IL., May 15-18, 2005. It is jointly sponsored by the American Association for the Study of Liver Diseases, the American Gastroenterological Association, the American Society for Gastrointestinal Endoscopy, and the Society for Surgery of the Alimentary Tract. For more information visit [www.ddw.org](http://www.ddw.org).

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## WHAT IS NORD AND GENETIC ALLIANCE?

The National Organization for Rare Disorders (NORD) is a unique federation of voluntary health organizations dedicated to helping people with rare "orphan" diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and service. It is also involved in the Medication Assistance Programs that help needy patients obtain certain drugs they could not otherwise afford. NORD was established in 1983 by patients and families who worked together to get the *Orphan Drug Act* passed. One in every 10 individuals in this country has received a diagnosis of a rare disease. NORD has serviced as the primary non-governmental clearinghouse for information on rare disorders and also provides referrals support groups and other sources of assistance.

Genetic Alliance is an international coalition comprised of more than 600 advocacy, research and healthcare organizations that represent millions of individuals with genetic conditions and their interests. As a broad-based coalition of key stakeholders, we leverage the voices of millions of individuals living with genetic conditions. With an 18-year history, the Alliance identifies solutions to emerging problems and works to reduce obstacles to rapid and effective translation of research into accessible technologies

and services that improve human health. It helps to advocate for robust healthcare systems; educate affected individuals and families to ensure informed decision-making; and empower advocates to partner with academia, industry and government to achieve their missions.

The Board has determined these two organizations worthwhile investments of our donated dollars because of the important work they do for patients and families like ours. While we are in the trenches of learning about our disease, offering patient care, handling insurance issues, providing emotional and financial support, etc., NORD and Genetic Alliance are busy helping us with the bigger picture of education, advocacy for public policy and research, drug and treatment policy and more. We pay \$50.00 a year for NORD and nothing for Genetic Alliance memberships, a bargain for the work they do to educate and influence Congress, the President, and other governmental agencies about rare disease like SDS. To find out more about these organizations either visit their web sites or call them. NORD: [www.rarediseases.org](http://www.rarediseases.org), (203) 744-0100 and toll free (voicemail only) (800) 999-6673. Genetic Alliance: [www.geneticalliance.org](http://www.geneticalliance.org), (202) 966-5557.

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## **NIH CALLS ON SCIENTISTS TO SPEED PUBLIC RELEASE OF RESEARCH PUBLICATIONS**

The National Institutes of Health (NIH) announced today, February 3, 2005, a new policy designed to accelerate the public's access to published articles resulting from NIH-funded research. The policy--the first of its kind for NIH--calls on scientists to release to the public manuscripts from research supported by NIH as soon as possible, and within 12 months of final publication.

The NIH policy will achieve several important goals, including: creating a stable archive of peer-reviewed research publications resulting from NIH-

funded studies to ensure the permanent preservation of these vital research findings; securing a searchable compendium of these research publications that NIH and its awarress can use to manage more efficiently and to understand better their research portfolios, monitor scientific productivity, and ultimately, help set research priorities; and making published results of NIH-funded research more readily accessible to the public, health care providers, educators, and scientists.

As part of on-going efforts to implement this new policy, NIH plans to establish a Public Access Advisory Working Group, as a subgroup of the National Library of Medicine's (NLM) Board of Regents. The Working Group will include representatives of the patient advocacy, scientific, library, and publishing communities, and will provide advice on implementation issues and assess progress in meeting the new policy's state goals. Additional information on the new policy and related documents, including a "Questions and Answers" fact sheet can be found at: <http://www.nih.gov/about/publicaccess/index.htm>.

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## **GENETIC NONDISCRIMINATION ACT PASSES IN THE SENATE**

The US Senate unanimously passed S.306, the Genetic Information Nondiscrimination Act of 2005 on Thursday, February 17, 2005. The bill is almost identical to S.1053, which passed in the last Congress.

Genetic Information Nondiscrimination Act of 2005: In health insurance, this act prohibits a group health plan or a health insurance issuer from: adjusting premiums on the basis of genetic information; or requesting or requiring an individual or a family member of such individual to undergo a genetic test. In employment information, this act prohibits an employer, employment agency, labor organization, or joint labor-management committee for discriminating against an employee, individual, or member on the basis of genetic information.

## DREAM FOR A CURE BRACELETS

The bracelets are made of Swarovski Crystal and Sterling Silver beads, it also has the SDSF logo on a silver charm and a Dream message bead. I have several colors to choose from and can customize for size. The colors available are clear, black, dark blue, light blue, sapphire, pink, light amethyst, amethyst, tanzanite (lavender), light red, medium red, garnet, peridot (light green), emerald, and birthstone colors. These bracelets are beautiful and make great gifts. You can order your bracelet in honor of your child and a special card will be sent with the bracelet as well as being listed in our newsletter. What a great way to support all SDS children and help with our Dream For A Cure.

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

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

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

## 7th ANNUAL SUPER BOWL FUNDRAISER

The seventh year for our football fundraiser proved to be SUPER, thanks to the great effort of friends and family. This year we were able to raise \$8,561.00, our running total is about \$50,000, not bad for a little Knights of Columbus hall party. We had around a hundred people show up for this year's bash, it seems to be growing every year. I would like to thank Norm & Mary Brideau, Jim & Rita Cole, and Steve Guarini, who have been very generous and donated to our cause for a number of years. Having friends and relatives like these, help make our efforts that much better. Others who have

been there each and every year are my parents, Ed and Doris Jenuwine, my father has sold an amazing amount of raffle tickets every year. A special thanks goes out to my sister-in-law, Peggy Lentz, for all of her hard work. Even though Peggy just had a baby three months ago, she still went around to different businesses to get items to raffle. She even got a company to make us a banner with the Shwachman logo on it. There are many others who have helped with selling raffle tickets and their efforts are very much appreciated. Each year we raffle off a big screen TV, this year it was a 51 inch Toshiba high definition, the lucky winner was Deanne Smith.

Deanne just happens to live close by us and actually rode to the party with Jenny. Think we didn't hear about that one! The winner of our 50/50 raffle was Shaun Ranney, this year's pot was worth \$2,000.00 and he is also local to our town. We sure heard a lot about the outcome of this year's drawings, but of course it was just a coincidence. Congratulations to both of you, and to all who showed up to help out with our fundraising event. A good time was had by all!!

I would just like to conclude with a thought about fundraising. Over the years our group has certainly gotten bigger, and we fortunately have more individuals getting involved in fundraising. There is always room for more of us to do something and get involved with helping our support group. Just think of all the information that has become available to all of us through the doctors and the board of directors. Someone is always there to answer questions, give advice, or refer you to someone who just might be able to help you and get the information you are looking for. Fundraising can be a lot of fun, as our event has turned into, but it certainly makes you feel like you are giving back a little bit for the efforts of a lot of caring people. Think about what might work for you in your area. The ways of contributing are endless! As always, I would be happy to speak to anyone if they have a thought or idea regarding FUNDRAISING!

Sincerely,  
Al Jenuwine

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

## Research on Motility and Chemotaxis in SDS Neutrophils

Dr. Fred Goldman and Dr. David R. Soll, of the University of Iowa, are studying neutrophil motility and chemotaxis in SDS patients using advanced computer-assisted 2D and 3D motion analysis systems. A recent study completed last year in Dr. Soll's laboratory demonstrated a very specific defect in chemotaxis that was reproducible in all SDS patients that were examined. This is also consistent with several earlier reports of neutrophil motility defects in SDS. The proposed studies are important to SDS in many ways. First, it will shed light on this disorder and may lead to predictions as to the underlying molecular basis of SDS. Second, it may help explain certain clinical circumstances (e.g. infection 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](mailto:frederick-goldman@uiowa.edu).

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## Update from Toronto: Genetic Testing for SDS

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

## Studies on the Molecular Mechanisms of Bone Marrow Failure

Bone marrow failure (BMF) syndromes such as aplastic anemia or myelodysplastic syndrome (MDS) may develop by a number of different mechanisms. We believe that a genetic predisposition to aplastic anemia and MDS is much more common than currently appreciated, and that a significant proportion of individuals thought to have “idiopathic” aplastic anemia or myelodysplasia may have a genetic alteration as the underlying or predisposing cause. Drs. Monica Bessler, Philip Mason, and David Wilson at Washington University in St. Louis, have begun a new study to identify alterations in genes that may predispose a person to the development of bone marrow failure or influence the course of the disease. We are collaborating with researchers at several other institutions throughout the United States including St. Louis University, Boston Children’s Hospital, the University of California at San Francisco, the University of Iowa, Children’s Hospital of Pittsburgh, Oregon Health Science University, Duke University, and other collaborating centers. Our study seeks to identify genes, their mutations, and their role in the development of bone marrow failure and the genes contributing to leukemic transformation. By understanding the genetic contribution, we hope to gain a better understanding of the course of the disease and ultimately factors that predict leukemic transformation and response to treatment. Our study is open to all children and adults who have or had aplastic anemia (inherited or acquired), paroxysmal nocturnal hemoglobinuria, or MDS. Advancing our knowledge of how these conditions develop is only possible because of the participation 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 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  
Washington University School of Medicine;  
660 S. Euclid Ave., Box 8125;  
St. Louis, MO 63110, USA  
Phone 314-362-8807  
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

## **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.

## **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](mailto:jkroppe@wowway.com) or call me at (248) 619-9316. I will be glad to answer any questions and I appreciate any and all input. Thank you to the many families who have contributed to 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](http://www.axcanscandipharm.com) and click on Products and Services or call 866-AXCANRX for enrollment information

# **THIRD INTERNATIONAL CONGRESS ON SHWACHMAN-DIAMOND SYNDROME**

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

**Sunday June 26, 2005 - Wednesday June 29, 2005  
Robinson College, Cambridge, UK**

On behalf of the members of the Conference Organizing Committees, I am pleased to invite you to the third International Medical and Scientific Conference on Shwachman-Diamond Syndrome (SDS), which this year will be located at Robinson College in Cambridge. The Conference is expecting to attract worldwide participants, including specialists in genetics, hematology, particularly those interested in neutropenia and bone marrow failure, oncologists, and gastroenterologists with an interest in pancreatic function, malabsorption and nutrition.

This third bi-annual Conference will be held over 3 days in the delightful surroundings of England's most beautiful university city. The meeting will provide a vital forum for research presentations, discussion and the exchange of ideas, and encompass stimulating scientific and social programs with parallel sessions for support and parent groups.

Robinson College is situated just outside Cambridge City Centre. Cambridge is within easy reach of London, Heathrow and Stansted airports, with good motorway, rail, and bus connections to other parts of the United Kingdom. Cambridge Airport has some domestic services and facilities for charter flights. Detailed travel information can be found at <http://www.robinson.cam.ac.uk/info/findingsus.php>.

## **IMPORTANT DEADLINES:**

<b>Submission of Abstracts:</b>	<b>April 22, 2005</b>
<b>Early Bird Registration Fee:</b>	<b>April 25, 2005</b>
<b>Requests for refunds on Cancellations:</b>	<b>April 25, 2005</b>
<b>Hotel Bookings:</b>	<b>May 27, 2005</b>

The Conference program will be presented in English. There will be no simultaneous translation.

## **PRELIMINARY PROGRAM**

Sunday, June 26, 2005

Registration

“Why are the clinical phenotypes of single-gene disorders so varied and complex?” by Professor Sir David Weatherall MD FRCP FRS Hon FRCPCH

Buffet Reception and Speaker's Dinner

Monday, June 27, 2005

- Registration
- Congress opening by Professor John Dodge, University of Wales, Swansea (UK)
- Clinical and Laboratory features of SDS:
  - Pancreatic/Growth by Peter Dale (UK)
  - Hematological features by Georgina Hall (UK)
  - Skeletal features by Outie Makitie (Finland)
  - Oral and Dental features by Carol Mason (UK)
  - Behavioral Phenotype by Mandy Bryon (UK)
- Genetics and Genotype phenotype:
  - Common Mutations by Martin Schwarz (UK)
  - Rare Mutations - Is there another Gene? by Johanna Rommens (Canada)
  - Presentations on genotype and phenotype by T Kuipjer (Holland)
  - Genetic Counseling by Peter Scambler (UK)
- Panel Discussion - Does genetic testing confirm the diagnosis? Chaired by Peter Scambler (UK)
- Late Breaking Science/Research papers

Tuesday, June 28, 2005

- Research Papers - Titles to be announced later. Presented by Alan Warren (UK), G Boocock (Canada), A Shimamura (USA), Siyi Zhang (Canada), T Menne (UK), Y Dror (Canada), and Elena Nicolis (Italy)
- Research Papers - Presented by Johanna Rommens (Canada), J Donadieu (France), SK Loftus (USA), C Liu (USA), and Shiro Ikegawa (Japan)
- Panel Discussion - Chaired by Alan Warren (UK) and Johanna Rommens (Canada)

Poster Review and Commentary

Wednesday, June 29, 2005

- Ethics of research into rare genetics disorders by M Parker (Oxford, UK) - to be confirmed
- Workshops: An International Registry - Chaired by Peter Durie (Canada). Facilitators: M Cipolli (Italy), E Siderius (Holland), P Durie (Canada), C Zeidler (Germany), Elene Psiachou-Leonard (UK), Sarah Ball (UK), and Patricia Petaros (Italy)
- Consensus on current management:
  - The role of Granulocyte Colony Stimulating Factor (rG-CSF) by J Donadieu (France)
  - The role of recombinant Growth Hormone (rGH) by E Psiachou-Leonard (UK)
  - The management of MDS by M Freedman (Canada)
  - BMT when and how by P Veys (UK)
  - Pancreatic supplementation: How much and for how long by P Durie (Canada)

# Thank You to our Donors

(donations December 1, 2004 - March 11, 2005)

Stephen & Peggy Lentz  
Kathryn M. Poth  
Maxine L. Borghesi  
Ali & Avishan Elmi  
John & Jeanette Brooks  
Thomas & Betty Lewellen  
John & Joyce Wall  
Mark & Susan Hanson  
Donald & Deborah Buterbaugh  
Lambda Chi Omega Sorority  
Allstate Giving Campaign  
Pediatric Gastroenterology Consultants, PC  
The Boston Foundation  
Center Line Lions Club  
La Mariposa School PTA

## **Gracie Fund**

### **Angel Anna Baskets**

Robert & Elizabeth Aloisi  
Jeffrey & Lisa DeGriek

### **Kolar Golf Charity**

Jennifer & Kevin Kolar  
Daniel Mangan  
John & Patricia Egan  
513 Third Restaurant LTD  
The Butcher's Block  
Wood Street, Inc.  
43-17 Corp. DBA The Gaslight  
PLK Vending Inc.

### **Super Bowl Fundraiser**

Elizabeth Jenuwine  
James & Rita Cole  
Steven & Renee Guarini  
Larry & Nancy Alvarado  
Peter Tranchida  
Dennis & Donna Timpf  
Paul Van Den Branden  
Edward & Donna Tessmer  
Harold & Phyllis Duchan

Michael & Ericka Haag  
Sandra Ingesoulia  
David Yangoutian  
Michael & Anne Marie Lentz  
Crooked Creek Farm Dairy  
PVS Transportation, Inc.  
Greenfield Cabinetry Inc.  
Schoenherr & Cahill, PC  
Ford Funeral Home, Inc.

### **In Honor of Warren Oliver**

Kathryn & Jeff Farmer  
Tasa & Frank Anderson  
James & Kathryn Oliver  
Leigh Pauling  
James & Elizabeth McCoy  
Elizabeth Murphy Jones

### **In Honor of Dylan Kolar**

Robert & Kristine Kolar  
Raymond & Kathleen McCarthy  
Bernard & Carol Kolar  
Jennifer & Kevin Kolar  
Karen Scaramucci

### **In Honor of Michele Elebracht**

Florissant Elks Ladies Club, Inc.

### **In Honor of Melissa Henle**

Patrick & Marie Cillo

### **In Honor of the Curran Boys**

Douglas & Amy Avrit

### **In Honor of Corinne Savulich**

Mr. & Mrs. Daniel Towers

**In Honor of Baleigh & James Closson**

Caroline Street School

**In Honor of Erin**

Monty & Jan Mayo

**In Honor of Kaitlyn Noel Bright**

Noson & Michele Fontenot

**In Honor of Katie Ruick**

Carol Lee

**In Honor of Troy & Kelsey DeBoer**

Corky & Ros DeBoer

**In Honor of Ashleigh Orosz**

George & Valeria Vezakis

**In Memory of Ima Smith**

Herbert & Vivian Crouthamel

Norman & Mary Wilson

Carroll & Beverly Papjohn

David & Karen Mueller

Cara & Forrest O'Neal

Summer Canteen, Inc.

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

Jackie Holt

Katherine Forse

Donna Thompson

Michele Fontenst

Taylor Bright

**SDS bracelets purchased  
In Honor of Christopher B. Garfield**

Hllie Knighton

Donna Garfield

KristinHolloway

Gelnda Ludaback

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

Caralmae Knickmeyer

Betty Cronin

**SDS bracelets purchased  
In Honor of Ryan Miller**

Nancy Miller

Nancy Holohan

Rachel Adamski

**SDS bracelets purchased  
In Honor of Gavin Miller**

Amber Sanchez

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

Linda Osborne

Coleen Eshman

Joyce Eshman

**SDS bracelets purchased  
In Honor of Brooke Lindgren**

Mrs. David Lindgren

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

Nancy Molnar

Rita Utz

Susan Utz

Alice Swango

Linda Cody

Jenni Herfel

Ruth Robinson

Anchor Club/Jeffersonville High School

**SDS bracelet purchased  
In Honor of Katie Ruick**

Nancy & Gary Ruick

## 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

## NEWSLETTER IDEAS

Do you have ideas for our newsletter?  
Want to share your story? Please send your suggestions and stories to SDSF at the address or email them to:  
**4sskids@shwachman-diamond.org**  
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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Toronto, Canada

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Research Coordination  
Bethesda, MD

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Laboratory of Molecular Biology  
Cambridge, England

## SDSF

**Founder**  
Joan Mowery

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Attorney  
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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.

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

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FAMILY NAME ON NEWSLETTER: \_\_\_\_\_

ADDRESS ON NEWSLETTER: \_\_\_\_\_  
\_\_\_\_\_

PHONE NUMBER: \_\_\_\_\_

PLEASE SEND ME THE ELECTRONIC NEWSLETTER, MY EMAIL ADDRESS IS:  
\_\_\_\_\_

I DON'T HAVE ACCESS EMAIL OR THE INTERNET, PLEASE SEND ME A PRINTED COPY OF THE NEWSLETTER AT THE ABOVE ADDRESS \_\_\_\_\_

PLEASE SEND FORM TO: SHWACHMAN-DIAMOND SYNDROME FOUNDATION  
710 BRASSIE DRIVE  
GRAND JUNCTION, CO 81506

710 Brassie Drive  
Grand Junction, CO 81506  
1-877-737-4685

ADDRESS SERVICE REQUESTED

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