

NEWSLETTER

Winter 2005/2006

As the year winds down there is still much work to do for Shwachman-Diamond Syndrome and this Foundation. It is time again for our annual fund drive and my hope is that we can meet or exceed our previous amount raised of \$17,000. This **can** only be done with your help. This is a **very** exciting time for SDS and we need more funds to support the research projects that are submitted to our organization every year. This organization has a board of dedicated doctors/scientists who make up the Medical Scientific Advisory Board (MSAB) and we are so fortunate that they are interested in working with us on our disease. They not only read over and decide to grant or not grant approval for the research projects submitted to SDSF for funding but they are all involved in some kind of research on SDS themselves. I have to reiterate the energy and excitement I felt at the last Scientific Congress because of all these wonderful researchers. If we are forced to continually turn away research due to lack of funding, ***the scientists/researchers will find another disease to work on - we've seen it happen.*** I know it must seem that to give one more donation this year is too much. With everything that has happened in our nation alone this last year, I can imagine that there are many organizations that need your funds. It is unimaginable what families here and around the world are going through that have been affected by the hurricanes and other natural disasters. Organizations that help in these circumstances are important, but **so is this foundation.** For most of you who are reading this, you have a family member with SDS and it is ok to take care of family first.

When my daughter was diagnosed almost nine years ago, I read all that I could about this disease. I lived in denial and didn't want to believe what I read could happen to us. I knew there were children and young adults that passed

away, I also knew this was a bone marrow failure disease. My choice was to bury my head in the sand. I am not going to condemn myself because we all have different strategies of coping. BUT when my nightmare became reality and she became very sick and was in complete bone marrow failure, I went to this organization **first.** I knew they would have the history that wasn't published (like, how many bone marrow transplants had succeeded and where they were done), the doctor advice that was specific to this disease, the family support that came in the way of phone cards and phone calls, and so much more. My daughter made it through the transplant, but it **did not** cure her disease. She still has had numerous surgeries on her hips and she still has malabsorption. Bone marrow transplant only cures one part of the disease because it is a multi system disorder. We **have** to find the key to unlock the mysteries of how this gene affects so many systems.

As it stands, there are less than a handful of families that raise **all** the funds to continue this mission. Where do you fit into all of this? When and how are you going to get involved? Contact us if you have an idea but don't know how to get started, we can help. SDSF is here to help all the SDS patients and their families however we can.

Happy and healthy Holidays to all of you from all of us!
Debbie Kadel

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**Shwachman-
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Syndrome
Foundation**

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ANNUAL FUND DRIVE

What does SDSF do for you?

SDSF has begun its 2nd Annual Fund Drive to support all patients and their families as well as research for Shwachman-Diamond Syndrome. Ask your friends, family, and yourself to donate in helping find a Cure For the Future. All donations help SDSF to strive to accomplish our mission statement:

- ** Advocate and support research towards a cure and improve medical management of symptoms**
- ** Educate the medical community and general public about Shwachman-Diamond Syndrome**
- ** Provide emotional support to patients and their families**
- ** Link families through medical/family conferences to share experiences and ideas**
- ** Disseminate current medical information**
- ** Support an International Patient Registry and International Medical Conferences**

For every dollar donated only 7% goes toward administrative costs and \$.93 helps to accomplish our mission (according to the Combined Federal Campaign - the federal employee, annual workplace giving campaign). Each research project costs \$30,000 and we want to make sure that we always have at least triple that amount of money in the bank for at least 2 research proposals each year as well as reserves for the family conference coming up (more details to follow) and the next scientific/researchers meeting. We need to make sure that we can fund all that we need to fund as our needs change each year.

Please take a moment to send your donation in the envelope provided and also ask your friends and family to donate to this worthy cause. Don't forget to fill out the Matching Gift fund paperwork from your employer to maximize your donation. Our number is 43-1709945.

We are working hard to help all those affected by Shwachman-Diamond Syndrome.

WHY IS SDSF SO IMPORTANT TO YOUR SDS FAMILY'S HEALTH?

According to NORD (National Organization of Rare Disorders) the federal government defines a rare disease as one which affects fewer than 200,000 people and it is estimated that approximately 6,000 rare disorders collectively affect more than 25 million people in the United

States. The common issues that face all of these patients and families are: Diagnosis (or misdiagnosis), adequate information about the disease, available resources, insurance issues, and treatments.

Perhaps the greatest barrier to the timely and effective treatment of rare diseases is the lack of knowledge among both patients and providers. Rare diseases are difficult to diagnose and many doctors are unfamiliar with their symptoms. Consequently, rare diseases often go undiagnosed, or misdiagnosed, for long periods. A 1989 Congressional Report which summarized the needs of individuals with rare disorders, and of the providers and researchers who service this community, indicated that slightly more than half (52%) of rare disease patients who went undiagnosed for three months or longer felt that the delay in receiving a diagnosis was due to insufficient physician knowledge to make a correct diagnosis. Patients also cited confusing symptoms (27%) and improper or incorrect initial diagnosis (18%) as additional reasons for delays in diagnosis. Patients also reported a need for understandable information about rare disorders. When faced with a rare disease diagnosis, patients are often scared and confused - they are anxious to obtain understandable information about their condition and about the resources which are available to them.

NORD successfully lobbied for the passage of two bills which were signed into law by President Bush last fall: The Rare Diseases Act of 2002, which established the Office of Rare Diseases as a permanent entity within the National Institutes of Health and establishes centers of excellence for rare disease research. The second bill is the Orphan Product Development Act of 2002, which authorizes appropriations under the Act through FY 2006. Both of these bills are important for us to know about as we head down the trail of caring for a rare disease patient because they give us hope that we aren't in this alone. We are not only a part of the Shwachman-Diamond Syndrome Foundation community but of the larger rare diseases community as well.

While the rare disorders community shares in the needs of the general patient population (eg: affordable health insurance), they have a distinctive subset of needs. The major, and interrelated, needs of this community are the same as they were in 1989: early, effective diagnosis and treatment; and ready access to reliable, understandable information. SDSF has been involved in doctor education since its inception through attending medical conferences and through helping to host or financially support scientific conferences specifically for SDS. Through our newsletter, web site, and family conferences, we have worked hard to disseminate information to the families, patients, doctors and scientists to ease these problems of where to turn to for financial and medical help. By bringing

the scientific community together through research funding and sharing, we can help push the idea of creating treatment protocols that can standardize and improve care throughout the country and the world. We all have to participate in the medical research studies, educate our own doctors, send in or fundraise for donations while being proactive with your doctor as a partner for information. We want to work hard for SDSF to help our community but we need your help too. We can all keep each other healthy!

This article has used information from Marlene G. Krammer, a Sarah Lawrence grad student reporting an Evaluation and Assessment for NORD, January 14, 2003 as well as information from the NORD web site.

CHARITABLE MEDICAL AIR TRANSPORTATION

Do you have a family member needing long-distance travel for specialized medical evaluation, diagnosis or treatment?

If so, you need to call the National Patient Travel Center, a 501(c)(3) charity specializing in helping patients find free or low-cost travel and hospital hospitality for family members in distant cities far from home.

The National Patient Air Transportation Helpline is the only service of its kind in America. It can be reached 24/7 everyday of the year. It has been in operation for 15 years and helps thousands of patient families annually. Call them on 800-296-1217 east coast time from 9am to 5pm. There is no charge for their referral services.

The NPATH Helpline is operated by Mercy Medical Airlift in support of Angel Flight America (which does 90% of long-distance charitable medical air transportation in the U.S.), Joe's House, the National Association of Hospital Hospitality Houses and the American Cancer Society.

Note: If you have any questions about this service call Mr. Steve Patterson, Executive Vice President, Mercy Medical Airlift on 757-318-9174.

Mercy Medical Airlift
National Patient Travel Center
4620 Haygood Road, Suite 1
Virginia Beach, Virginia 23455
mercymedical@erols.com

MAKE-A-WISH FOUNDATION

The Make-A-Wish Foundation grants the wishes of children between the ages of 2 1/2 and 18 with life-threatening medical conditions to enrich the human experience with hope, strength, and joy.

Born in 1980 when a group of caring individuals helped a young boy fulfill his dream of becoming a police officer, the Foundation is now the largest wish-granting charity in the world, with 73 chapters in the U.S. and its territories and 28 international affiliates on five continents. Granting more than 11,500 wishes a year and more than 144,000 wishes worldwide since inception, the Make-A-Wish Foundation celebrates "25 Years of Making Dreams Come True" in 2005.

The Foundation's mission reflects the life-changing impact that a Make-A-Wish experience has on children, families, referral sources, donors, sponsors, and entire communities. The Make-A-Wish Foundation has fulfilled its mission thanks to the millions of individuals, organizations, and corporations that have supported the organization by volunteering, donating, sharing the Make-A-Wish message, and participating in the wish experience. Individuals interested in supporting the Make-A-Wish cause will find opportunities to help, regardless of location, income, or time commitment.

For more information about Make-A-Wish Foundation, visit www.wish.org

TELL US YOUR STORY

If you would like to share your family's story about your experience with a wish-granting agency such as Make-A-Wish, Dream Factory, Wishing Star, or any of the other agencies that provide wishes for children with chronic illnesses, we would love to read about it in our SDSF newsletter. Likewise, if you have an experience with a long distance medical air transportation agency such as Angel Flight, we want to hear about it. Please send your story to 4sskids@shwachman-diamond.org for a future edition of our newsletter. Your family's experience may provide encouragement or inspiration to another family of a child with SDS and will allow us to know a little bit about your brave son or daughter.

FAMILY SHARING PAGE

PATRICK MAKES A WISH!

When Patrick was four years old, the doctors told us that Patrick's SDS disease was progressing. We were devastated. Nothing in the world hurts more than watching your child suffer, knowing that it may get worse. Shortly after our meeting with Patrick's doctor, he phoned us with an upbeat tone to his voice and said that Patrick had been chosen to participate with the Make-A-Wish Foundation (MAW). Patrick was so thrilled, as was the whole family!

The MAW volunteers came to our house and brought gifts for Patrick and his older brother, one thing we learned quickly was that MAW always included the siblings in everything! They also explained to us that MAW gives wishes to children with life threatening diseases not only terminally ill children. I think many people have that misconception about MAW, as I did. The volunteers came over several times and answered questions and discussed with us that Patrick could wish for ANYTHING he wanted. That was hard for us to comprehend and put Patrick into overdrive of dreaming up all kinds of outstanding things a four year old could dream of!

Patrick first asked the MAW volunteers for a real red Porsche then he asked for his favorite cousin to move into **our** house forever. But after those two wishes were impossible to grant (with a little chuckling from the volunteers!), Patrick finally decided that going to Disney World as a family would be totally "cool"! We had never been to Disney before.

I can't tell you how we felt taking that once in a lifetime trip. MAW took such great care of our family, they paid for everything and pampered us to no end. We stayed at Give The Kids The World, a place for children like Patrick, and it was amazing. There was a movie theatre on the premises as well as an all you can eat ice cream palace! Right near our condo, we rode horses, a train, and a carousel too. It was a child's dreamland come true. Best of all, because Patrick was a "wish kid", our family was able to be FIRST in line at EVERY ride at all the theme parks! No waiting! Patrick went on the Buzz Lightyear ride 10 times in a row and was able to meet Buzz and Spiderman up close, what a treat!

We keep a scrapbook of our trip to Disney and look through it often remembering all the wonderful, fun things we did. I know Patrick will never forget it and we will never

be able to thank MAW enough for their pure generosity and kindness. Now, nearly three years later, MAW is still part of our lives. They call us often to ask Patrick and our family to attend professional hockey and baseball games at no cost. They want our family to have a break from the daily grind of life, especially dealing with a child with a chronic illness like SDS. We attend those sporting events and have a blast! THANK YOU MAKE-A-WISH!!!!!!

Sincerely, the Kroppe family

LOGAN STONE'S STORY

My name is Logan Stone and I am a sixteen year old with Shwachman-Diamond Syndrome. I was born on May 21, 1989 but wasn't expected until June 27th. I surprised my parents by coming early, my Dad was up near Greenland or Iceland when I was born. I weighed 4lbs. 12 ozs. and was around 17 inches long. My parents thought something might be wrong since I wasn't growing and I had many (8-10) messy diapers a day. When I was a year old I weighed only 14lbs. and I had many ear infections and colds. My first of many surgeries was when I had tubes put in my ears at one years old.

My parents took me to the pediatrician and many specialists trying to figure out what was wrong. The first person who mentioned SDS was a gastroenterologist, she ordered many biopsies, such as liver and intestines. They went ahead and put me on pancreatic enzymes, they also tried to do a pancreatic stimulation test but they couldn't get the tube down my throat because I was too small. Many years passed with doctor changes, surgeries, and different types of biopsies. Finally at age 8, we went to Dr. Chong at Riley Children's Hospital and he was able to perform the pancreatic stimulation test successfully. They found out that I was definitely pancreatic insufficient and they discovered that my normal blood counts weren't so normal. Doctors finally gave me a clinical diagnosis of SDS. Many years later the gene was discovered and I tested positive for it. I get bone marrow biopsies done 1-2 times a year and blood work done nearly every month. I have chromosomal abnormalities and very hypocellular bone marrow on my biopsies.

SDS has affected me in huge ways. There are not many positives for SDS, not any in fact, but I have adjusted to it. I have often been bullied by other students, mainly because of my size, stomach problems, and the fact that I am not as athletic as people my age. My self esteem has

suffered as well. I have also had to put up with a lot of pain and disruption in my social life since I spend a lot of time at doctor appointments and in the hospital. I get tired of being treated like a science experiment. I don't necessarily get tired of the pokes I go through, but several times a day my veins harden on me and hurt from the scar tissue. I also get tired of my stomach being too predictable at times it hurts. The only positive I can think of are that I am a very serious, hard driven student, very sympathetic to others, and that I hope to go into the medical profession from these experiences. Even though I have had a lot of problems growing up, I have learned to cope pretty well, I wouldn't know any other life now.

I love to read, write, play video games, listen to music and ponder about anything. I like being in touch with other young people with SDS especially people my age. I have played soccer for 12 years and just finished my second year on the JV soccer team at my high school. I have also played the violin for 12 years and I am in two different orchestras. I have been in Boy Scouts since Kindergarten. I have decided to make my Eagle Scout Project a blood drive through the American Red Cross because of all I have been through. My blood drive will be on November 22, 2005 and I hope for a good turn out. My Eagle Rank will come sometime after that.

HOLIDAY HUMOR

"HOLIDAY EATING TIPS"

** Avoid carrot sticks. Anyone who puts carrots on a holiday buffet table knows nothing of the Christmas spirit. In fact, if you see carrots, leave immediately. Go next door, where they're serving rum balls.

** Drink as much eggnog as you can and quickly. Like fine single-malt scotch, it's rare, in fact it's even rarer than single-malt scotch. You can't find it any other time of the year but now, so drink up! Who cares that it has 10,000 calories in every sip? It's not as if you're going to turn into an eggnog-aholic or something. It's a treat. Enjoy it. Have one for me. Have two. It's later than you think, it's the Holidays!

** If something comes with gravy, use it, that's the whole point of gravy. Gravy does not stand alone. Pour it on. Make a volcano out of your mashed potatoes. Fill it with gravy. Eat the volcano. Repeat.

** As for mashed potatoes, always ask if they're made with skim milk or whole milk. If it's skim, pass. Why bother? It's like buying a sports car with an automatic transmission.

** Do not have a snack before going to a party in an effort to control your eating. The whole point of going to a Holiday party is to eat other people's food for free. Lots of it. Hello?

** Under no circumstances should you exercise between now and New Year's. You can do that in January when you have nothing else to do. This is the time for long naps, which you'll need after circling the buffet table while carrying a 10-pound plate of food and that vat of eggnog.

** If you come across something really good at a buffet table, like frosted cookies and pastries, position yourself near them and don't budge. Have as many as you can before becoming the center of attention. They're like a beautiful pair of shoes. If you leave them behind, you're never going to see them again.

** Same for pies. Apple. Pumpkin. Mincemeat. Have a slice of each. Or, if you don't like mincemeat, have two apples and one pumpkin. Always have three. When else do you get to have more than one dessert? Labor Day?

** Did someone mention fruitcake? Granted, it's loaded with the mandatory celebratory calories, but avoid it at all cost. I mean, have some standards.

** One final tip: If you don't feel terrible when you leave the party or get up from the table, you haven't been paying attention. Reread tips; start over, but hurry, January is just around the corner.

Remember this motto to live by:

"Life should NOT be a journey to the grave with the intention of arriving safely in an attractive and well preserved body, but rather to skid in sideways, chocolate in one hand, eggnog in the other, body thoroughly used up, totally worn out and screaming "WOO HOO what a ride!" Have an amazing holiday!"

WELCOME NEW FAMILIES

Each year many new families from all over the United States have children diagnosed with SDS. Some of these families may be in your area and we would like to welcome them into the Shwachman-Diamond Syndrome Foundation circle of support.

Roanoke, VA

DUTCH SDS PEDIATRICIAN SPEAKS TO CAPITAL HILL

ABOUT SDS

Our kind associate, Liesbeth Siderius from Holland, has sent along an article for this newsletter concerning her trip to Washington D.C. this past October. She is tirelessly working on behalf of the patients with SDS and we truly appreciate her dedication to this disease.

“To all involved in Shwachman-Diamond Syndrome,

During the 75th Anniversary meeting of the American Academy of Pediatrics in Washington, pediatricians advocated protection of Medicaid for children.

On behalf of the Shwachman-Diamond Syndrome Foundation and Dr. Frederick Goldman of the University of Iowa Children’s Hospital, the statement listed below was delivered to the Washington office of the Representative Tom Latham of Iowa. I had the opportunity and pleasure to meet his staff member, Mr. Parker, and discuss the need of Medicaid and Genetic non-discrimination policies in the US.

I urge you to have direct contact with members of the House of Representatives in Washington or at your home state about the importance of the translation of genetic knowledge and accessible health care which will hopefully give insight to the needs of all children and adults with genetic based health risks.”

Liesbeth Siderius, M.D.

Pediatrician in the Netherlands, and member of the advisory board of the Shwachman-Diamond Support Group Holland

Document presented to Rep. Tom Latham’s office:

Protecting Medicaid for Children:

- All children should have equal access to health care independent of their social status, diagnosis, state of health and risk factors.

- Preventive care is necessary and cost effective. Early and periodic screening for rare genetic diseases leads to early diagnosis and treatment and offers health gains and reduces health care costs.

- Advances in Genetic knowledge (Genomics) can be transformed into preventive medicine. To make this happen, an accessible health care system based on solidarity is required. Genetic knowledge (Genomics) offers opportunities for early diagnosis which can provide children the care they need. However, this information could be problematic as it potentially may lead to discrimination by insurers or employers when these children reach adulthood. Genomics offers promising opportunities for treatment adjusted to ones individual genes, which can help to save lives. For many genetic diseases, there needs to be well established phenotype-genotype correlation to provide optimal care and predict outcome.

-The **Shwachman-Diamond Syndrome** has a prevalence of 1:50,000-70,000 births but approximately only 1,000 children are diagnosed in the U.S. at this time. This means that there are thousands of children who are either undiagnosed or misdiagnosed. The gene has been found and easy accessible testing is being developed. It is imperative that these children have access to this testing and are not penalized by their insurers or Medicaid. In fact insurers will benefit from early knowledge of diagnosis. First, there will be cost savings in that there will be fewer medical bills trying to figure out what is wrong with them and second, treatment can begin earlier. The clinical features of this disease are age dependant. The features may include bone marrow dysfunction with a risk of developing leukemia, pancreatic exocrine insufficiency, developmental delays and skeletal abnormalities.

Families involved with rare genetic disorders face the following problems: 1) delayed diagnosis; 2) inadequate treatment often due to limited access to care; and 3) sometimes poor knowledge base of diseases and its current treatments by local medical provider. International collaboration is essential for understanding the natural history of rare genetic diseases through patient registries and collaborative clinical trials with similar treatment protocols.

Children living in countries with an accessible health care system based on solidarity will profit more of genetic knowledge (genomics) than others.

SAVE THE DATE FOR THE NEXT FAMILY CONFERENCE IN JULY, 2006

The plans for the next family conference are starting to come together and we wanted to alert you to some of the details we have so far. It is going to be held at Camp Sunshine on Lake Sebago in Casco, Maine where the whole family can come and enjoy themselves. “Camp Sunshine supports children with life threatening illnesses and their families. Free quality services are provided at the camp including accommodations and meals, onsite medical services, counseling services, and recreational facilities. Breakfast, lunch, and dinner are served each day. A physician is located at the camp for the entire session so that families can be secure in the knowledge that the camp offers 24 hour onsite medical support. In addition, a hospital with full-time emergency room physicians and pediatricians is only 25 minutes from the camp.” All you have to pay for is travel expenses to and from the camp. The rest is paid for by the camp.

We will be providing more information on our web site, www.shwachman-diamond.org, as we formulate more details. To check out Camp Sunshine for yourself, go to their web site, www.campsunshine.org, for more information on the benefits of this new kind of family conference for our Shwachman-Diamond Syndrome community. You can meet other families and some of the doctors involved in SDS care as well as researchers of better treatments, GI issues, and molecular genetics.

So while we are mentioning this, we would like to send out a request for pictures of your SDS child/children with their pets, families, friends, etc. We will be putting together a slide show for the conference and want to include as many families as we can. Please go through your pictures and send in your favorite 5 to: Sharon Lamb, 7339 Lake Road, Appleton, NY 14008. If you would like to have these pictures back, please include a self addressed, stamped envelope and mark the back of each picture with your name. Thank you for your participation in helping to make this conference a great one. Every little bit counts.!

**YOU CAN DO IT!! BE OUR NEWS-
LETTER COORDINATOR**
not writer, not producer, not editor
It won't take up much time - really!

SDSF is looking for a person to help with coordinating articles and proofreading of this newsletter 3 times per year. It is a relatively small commitment as far as newsletters go and we could really use the help. You will be coordinating with the board members and any contributions via e-mail as well as with the assistant to the board who already knows how to manage this task and is ready for some help with production. There is no production involved for you - only coordinating the type and number of articles as well as tracking whether they have been sent to the assistant. It is easy and you can help with ideas of what goes into each newsletter.

Please say yes you will help. Must have an e-mail account as well as adobe acrobat 7 (easily downloadable for free from the internet) knowledge. Please e-mail the web site address, 4sskids@shwachman-diamond.org and tell us about your interest in this really important job. **We send this newsletter to families and doctors all around the world who count on the information they receive from us.** Please consider volunteering for this job. You CAN make a difference.

E-MAIL SUPPORT GROUP

Would you enjoy e-mailing other Shwachman-Diamond families? Have you ever thought your child seems to have something you may not think is related to the syndrome? Why not sign up for our e-mail support group through Yahoo. It is a good way to stay in contact with other SDS families and also a great venue for asking questions you may have.

If you would like to subscribe to our support group, the link is: shwachmandiamond-subscribe@yahoogroups.com

If you would like to look at the guidelines for our e-mail support group, the link is: <http://groups.yahoo.com/group/shwachmandiamond/?yguid=79215263>

If you have any questions, please contact Julie Kroppe at jkroppe@wowway.com

CONGRATULATIONS TO THE WASHINGTON UNIVERSITY SCHOOL OF MEDICINE AND THE NATIONAL INSTITUTES OF HEALTH

(each center has SDS research studies underway in their hematology departments - refer to page 10 & 11 of this newsletter)

Human Research Protection Award Recipients Announced

Bethesda, Maryland (December 8, 2005) - Winners of the 2005 Award for Excellence in Human Research Protection were announced today by Dr. Peter G. Goldschmidt, President and Founder of the Health Improvement Institute.

Winners were:

** ***Family Health International***, for its best practices (2 awards):

** *Research Ethics Training Curriculum*

** *Ethics Training Curriculum for Community*

Representatives

** ***Department of Clinical Bioethics, National Institutes of Health***, for its innovations, *Framework and Benchmarks for Evaluation of Research*

** ***Partners in Healthcare Systems***, for its best practice, *Human Research Quality Improvement Program*

** ***Washington University, School of Medicine and Human Studies***, for its best practice, *St. Louis IRB Consortium*

Awards are given for demonstrated excellence in promoting the well being of people who participate in research. The Health Improvement Institute created this awards program. The Office of Human Research Protection was the founding sponsor of the awards program. Sponsors of the 2005 awards program included American Diabetes Association and Pfizer. Judges and Committee and Award Advisory Board members are volunteers.

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.

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

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.

REQUEST A BASKET FOR YOUR CHILD OR FAMILY MEMBER IF THEY ARE IN THE HOSPITAL

The Angel Anna Baskets are filled with gifts tailored specifically to each sick child's age and needs, and are sent out to the hospital or the child's home, upon learning of a lengthy hospitalization. Balloon bouquets are also sent out to those children who are temporarily in the hospital or who are going through a particularly rough time medically. It is our way to let these families and children know that we care and are thinking of them during their difficult time. I believe it is a wonderful addition to the family support that SDSF gives to each of our SDS families!

If you would like to request an Angel Anna Basket sent to a sick and/or hospitalized SDS child, or if you would like to make a tax deductible donation to our Angel Anna Basket Project (material or monetary donation), please call SDSF at the toll free number 1-877-737-4685 or contact me personally online at 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

Thank You to our Donors

(donations August 1, 2005 - November 30, 2005)

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Angel Anna Baskets

Jeffrey & Lisa DeGriek

DREAM BRACELETS

I am making Swarovski Crystal bracelets with Sterling Silver beads, and SDS charms on them as a fundraiser for SDSF. 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 their families.

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 \$6,000.00 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
jengrsl2@netzero.net

BRACELET PURCHASES (August 1, - Nov. 30, 2005)

In Honor of Chilton Price

Becca Price
Miss Averitt

In Honor of Logan Martin

Carrie Martin

In Honor of Corinne Savulich

Mary Alice & Stephen Savulich

In Honor of Michele Ellebracht Mowery

Mamie Robinson
Faye Watt
Colleen Apple
Mary Walke
Jean Burke

In Honor of Danny Rohe

Shelly Friedhoff
Pat Wissinger
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Nikki Williams
Marlene West
Zola Craft
Amanda Brown
Sharon
Shirley Grawe
Sharon Jansen
Kim Watson
Denise Smith
Lauren
Rachel

In Honor of Dylan Kolar

Maureen Dolan
Nancy Doxee
Jennifer Kolar
Sandra Waiters
Violet Bischoff
Marion Lucarello

Established Shwachman-Diamond Groups

Shwachman-Diamond Syndrome Support - Australia

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

Shwachman-Diamond Support-UK

Contact: Sharon Clusker
01 522 792039
email: philcoxes@blueyonder.co.uk
<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

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

Jenny Jenuwine - MI: (810)395-2358 or jengrls2@netzero.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

Sharon Clusker - England:
Sharwk60@aol.com

Lee-Anne Hayes - Australia
61 02 49608428 or hathor@bigpond.net.au

Reinald Baumhauer - Germany
Fax: 049-89-41902871 or
r.baumhauer@mnet-mail.de

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

NEWSLETTER IDEAS

Do you have ideas for our newsletter? Do you have a question you would like to ask a doctor? Want to share your story?

Please send your stories and/or questions to SDSF at the address or e-mail them to:
4sskids@shwachman-diamond.org

We appreciate ALL input! We will print stories and answers in future newsletters.

Thank you.

CHANGE OF ADDRESS OR E-MAIL

Please forward your change of address or e-mail to continue receiving your newsletters.

If your newsletter is sent by regular mail, the post office will not forward it to you due to "Bulk Rate" postage being used.

Either call us at **1-877-737-4685** or e-mail us at **4sskids@shwachman-diamond.org** with your changes.

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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.

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

ADDRESS SERVICE REQUESTED

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Disclaimer: The views expressed in this newsletter do not necessarily represent the views of the Board of Directors, Professional Advisory Board or members of the organization. We do not promote or recommend any particular treatment, etc. The relevance of any medical information in this newsletter should be discussed with your physician.