As my children are settled into another school year, I realize summer is over and fall is just around the corner. The mornings are chilly and in the mountains of Colorado I can already see the changing trees. I have to reflect on the summer just a little bit. For those of you that weren’t able to attend our family meeting at Camp Sunshine, I hope you enjoy some of the pictures and a couple of stories in this newsletter. It was a beautiful place and a wonderful time. As a member of the board, I have to admit, it was a little more difficult to meet and spend time with other families than I would have liked. However, it was still nice to spend what time I could with them to get to know them better and find out more about their children. It was amazing when talking to their parents how many similarities there are with all of our children. It was also so nice to be able to see the young adults and adults affected by SDS and I have hope for the future for our little ones. I also loved visiting with the parents of the adult children; their vast knowledge and experience in having a child impacted by a rare disease is invaluable. The scientific sessions were very informative. Again, I have to say that I am so thankful for the team of physicians that attended and their diligence in studying Shwachman-Diamond Syndrome. They were also such fun to be with at the evening functions scheduled by the camp and always available to be pulled aside and offer advice or answer questions. Look for the picture of Lynda Ellis on the dress-up night you won’t want to miss it.

Please know that the board of SDSF is doing its best for those affected by Shwachman-Diamond Syndrome and their families. We would like very much to offer another meeting such as the one held this summer in the summer of 2008. We value your input and suggestions. We did receive feedback from those that attended, but we would like to hear from those that did not. What can we do that would make this happen for your family next time? Please drop us a line and let us know your thoughts on what you might like to see in a family meeting. You can reach us at 4sskids@shwachman-diamond.org. We are also available to answer any questions you might have.

With the start of school some parents might be looking at having an IEP in place. We can offer you suggestions or help with that. There is an informative article in this newsletter by Dr. Elizabeth Kerr who did a study on children affected by SDS and the impact it has on their behavior and learning. This article was presented at camp.

Another thing we are trying to put into place is a physician referral service called CareSearch. One of the questions we get a lot from new families or a family that has moved is, what doctor can we call in our area with experience in treating Shwachman-Diamond Syndrome? We would love for you to
e-mail us the names of hospitals/clinics and of the physicians who you feel are helpful to you in whatever specialties to blairvanbrunt@comcast.net. If you are a physician reading this newsletter and would like to be put on a list for possible referrals, please send us your information as well.

Finally, I would like to thank those of you that are out there fundraising on behalf of SDSF. By holding fundraisers, donating through the United Way or Combined Federal Campaign, or just sending in donations big or small, it ALL makes a difference. We can’t do what we do without your help and support. THANK YOU!

My best to each and every one of you and your families, Debbie Kadel

A SPECIAL THANK YOU TO JULIE KROPPE!

Julie Kroppe has been involved with our foundation since her youngest son was diagnosed with SDS. She has raised a significant amount of money for this foundation in so many ways. She has held a “Dance for a Cure” a few years in a row, a “Quarters for a Cure” campaign and was accepted twice by her local high school to receive funds from their annual charity week, on behalf of SDSF, raising between $15,000 and $25,000 each time. She has also been the coordinator behind the Angel Anna Basket program for the past seven years. She has found partial funding for each and every basket that is sent out as well as putting her heart into making each one special for each SDS recipient and their family. In addition to all of that, she is also the moderator and a great support to families on one of the SDS list serves.

I am writing this to thank Julie for all of her hard work and dedication to SDSF and to our children affected by this disease. She is going to take some time off from the Angel Anna Basket program after many years of service and I couldn’t let that happen without thanking her. Thank You, Julie for all that you have done for SDSF and for putting an immeasurable amount of smiles on the faces of the children who have received your baskets!!

The Angel Anna Basket project will continue. I am happy to announce that another parent of an SDS child, Staci Bishop, has agreed to take over this very worthwhile program.

Thank you both, Debbie Kadel

STACI BISHOP,
ANGEL ANNA COORDINATOR

My name is Staci Bishop and I am the new Angel Anna Coordinator. My oldest son, Davison, is 6 1/2 years old and was diagnosed with SDS when he was 3 months old. He has a brother Zachary and a sister Kylie that are not affected. We have received a lot of love and support through SDSF and I feel privileged to be able to give back to families by helping with the Angel Anna Baskets. Please feel free to call me directly or through the foundation, 515-252-7445.

DONNA GARFIELD
NEWSLETTER COORDINATOR

My name is Donna Garfield and I am the new newsletter coordinator. I was/am a registered nurse (I say was because I am not “working” as a nurse, although, I certainly use my “training” on a daily basis!). I do work outside the home full time but have been blessed over the years to have had understanding bosses that have let me adjust my hours or work from home on occasion and for that I am extremely thankful!
I have two sons, both with different but “rare”
potentially life-threatening chronic genetic diseases. My youngest son has a metabolic disease which although is chronic, is now well controlled and not progressive. His diagnosis was my first personal introduction into family life with a child with a chronic disease. My older, nineteen year old son Chris, is one of those “gray area” or “SDS-like” kids. No definitive diagnosis, but he has the short stature, chronic neutropenia, and bone marrow changes of SDS. Chris does not currently have PI (pancreatic insufficiency) but it is possible he outgrew it (by history he probably was pancreatic insufficient). He was never tested for PI during infancy, but fell off the charts at 6 months old and would never have gotten back on without the help of growth hormone shots. He was not diagnosed formally with a hematologic problem until he ws 14, although looking back at all of his CBC’s (complete blood counts) it has been an intermittent problem all of his life. As he has aged his neutropenia has become more severe and constant. He definitely had the frequent illnesses, infections, etc. that SDS’ers have during their childhood, even though his neutropenia was intermittent at the time.

I have heard many of the parents of older children and adult SDS’ers say that in general the number of infections has decreased as they got older. For us this has certainly been true. In fact last year Chris went away to college and stayed completely healthy the entire year!

Really, enough about me, so one parting thought. If anything good has come out of Chris’ illness it was the ability to meet so many compassionate and caring people. The doctors that we have met along our journey have been wonderful. Chris’ hematologist is not only a great doctor but a wonderful person. Even Dr. Alter and Dr. Elghetany, as busy as they are with their studies, have taken the time to answer my gazillion questions, even though Chris doesn’t have a definitive diagnosis. And last but not least are the other parents and our children as well. I am constantly amazed at what wonderful people our children are growing up to be. The stories I have heard about the good they do in the world and how they continuously fight to overcome the challenges placed in their paths are inspiring to say the least. That says a lot for them and the wonderful job all you parents are doing.

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GENETIC ALLIANCE PUBLISHES
“UNDERSTANDING GENETICS: A GUIDE FOR PATIENTS AND HEALTH PROFESSIONALS”

On May 25, 2006, Genetic Alliance announced that it published “Understanding Genetics: A Guide for Patients and Health Professionals” in collaboration with the District of Columbia Department of Health (DC DOH). This comprehensive manual provides a wealth of genetic education material for patients and health professionals for all readers. It is designed to help patients, their families and health professionals understand the place of genetics in healthcare. The straightforward and intuitive guide covers basic genetic concepts, complemented with in-depth information about diagnosing genetic conditions, newborn screening, family-history collections, genetic counseling and genetic testing. Although there is a section on local information that focuses on patient care, patient and provider education, and genetic services in the Washington, DC area, there is much more other helpful, useful information for people living anywhere in the world. The 120-page manual is published in print and electronic formats and is being distributed by the DC DOH Washington area healthcare clinics. You can download it for free by accessing the web site listed below.

http://www.geneticalliance.org/ws_display.asp?filter=resources_manual
WHY IS THE GENETIC INFORMATION NONDISCRIMINATION ACT NEEDED?

HOW IS THE SHWACHMAN-DIAMOND SYNDROME FOUNDATION INVOLVED?

There are a few states with strict protections against genetic discrimination, but most states have little to no protection. This leaves individuals with little knowledge about how much their genetic information is protected from state to state. In addition, companies who deal in any kind of health information are left with no national framework to guide how they handle genetic test results and genetic privacy information.

Genetic information is not properly covered under HIPAA privacy guidelines

Current HIPAA guidelines do not prohibit insurers from requiring genetic testing or from denying coverage based on genetic information. In fact, GINA (Genetic Information Nondiscrimination Act) is filling holes in HIPAA, making the whole legislation more consistent with regard to data protection. Genetic information is becoming increasingly more ingrained in medicine and as such will serve to complicate privacy law if steps are not taken now to close gaps in policy.

Genetic discrimination cases are already appearing and are sure to exponentially increase with so many new genetic tests coming out

In 2006, the Coalition for Genetic Fairness is continuing to work with lawmakers to ensure that the Genetic Information Nondiscrimination Act (H.R.1227) passes this year. Today, H.R.1227 has more than 203 cosponsors in the U.S. House of Representatives. To find out what you can do to help, visit: www.geneticfairness.org/action.html

Dr. Frances Collins, Director of the National Human Genome Research Institute at NIH, articulated how vital this legislation is to the scientific community, saying, “genomics today is where the computer industry was in the 1970’s - at the beginning of a technology revolution.” Former Speaker Newt Gingrich echoed those thoughts, asserting, “To not have this bill is to cripple our ability to save lives.”

Protect Americans from Genetic Discrimination. The Shwachman-Diamond Syndrome Foundation has signed on as official supporters of this bill to help ensure the passage of effective and responsible genetic information nondiscrimination legislation. As a member of the Coalition, our organization will play an integral role in educating Congress about the importance of this bill.

REPORT FROM THE INTERNATIONAL SHWACHMAN-DIAMOND SYNDROME DATABASE WORKSHOP BY PETER DURIE ON BEHALF OF ALL THE DELEGATES

Eighteen delegates from eight countries met in Verona, Italy in February, 2006 to participate in a highly successful workshop. As a result, plans have been formulated to establish an International Shwachman-Diamond Syndrome Database during 2006. This workshop was made possible by a generous grant from Altus Pharmaceuticals, Inc. who has been a long-term supporter of the medical and scientific activities concerning Shwachman-Diamond Syndrome (SDS). Altus has been a corporate sponsor of the last two International SDS Congresses, and now the company has committed to provide a three-year grant to support the important task of establishing the International SDS Database. I wish to convey a special word of thanks to Mr. Bob Gallotto and Dr. Alana Kimura, of...
Altus Pharmaceuticals, who attended the meeting as observers.

The workshop was divided into three half-day sessions. The first session examined how each country has progressed to establish a National SDS Database. National delegates offered a unanimous commitment to actively participate in the International SDS Database Consortium. Delegates provided concrete plans to complete or initiate the establishment of a national database in each country and have agreed to work on plans to transfer anonymous medical and genetic information from the National databases to the International database. Inaugural member countries of the International SDS Database Consortium include Australia (unable to send a delegate), Canada, Finland, France, Germany, Holland, Italy, the United Kingdom and the United States.

The second session was devoted to specific plans on how to establish the International Database. This included agreeing on the specific information that should be included in the database as well as formulating what we consider to be the most important medical and scientific questions an International database might be able to answer.

During the final session three working groups established a frame work for the International database, which included: ownership; governance and funding; day-to-day management and use of the database; and last but not least the legal and ethical implications of establishing a database. The latter working group addressed the important matter of securing and maintaining anonymity of information within the database as well as respecting the privacy of each individual’s health information.

While we are optimistic about the future, a lot more work needs to be done for the International Database to get off the ground. The most important and pressing need is to develop strong databases in individual countries. We sincerely hope the vital work that needs to be done in each country will be supported by the SDS families as well as all of the national SDS support groups.

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**CALLING ALL YOUNG PEOPLE**

Hey, kids! We want to hear from you! Do you have a story, joke or a picture you want to share with us? If so, please send it to us to put in an upcoming newsletter for others to see and enjoy. We know that you have many talents to share and things to say that other kids would enjoy hearing.

Hey teenagers! We want to hear for you as well! Please send us your stories, poems, pictures or reports about what is going on in your lives. Your experiences will undoubtedly inspire all of us and we would love to hear them.

Send any items to share to: dkgarfield@garfieldhome.net. We are looking forward to hearing from you!!!

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**FIREFLY**

**BY KELSEY JENUwine, AGE 12**

Once upon a time there was a very lonely firefly. The firefly wanted to be noticed by someone, so he went to the other side of the country. There he met the sun and told him his problem. The sun then gave him some of his light. The firefly thanked the sun and hurried back home.

When he got home no one knew who he was because he shined so bright. Then they knew because he is small. They asked him where he got it and he said from the sun. They all thought that was wonderful. That’s how fireflies got their light, when you see them at night.
FAMILY CONFERENCE AT CAMP SUNSHINE

What a fabulous family conference we had at Camp Sunshine in Casco, Maine from July 9-14, 2006. We wish that every family could have attended for all the fun, support and information the kids and parents received. Every age group was catered to. The 70 volunteers and fantastic staff were an integral part of making the families feel very welcomed and a part of every activity and discussion group. We had a blast!

We started off with a “meet and greet” on Sunday night followed by a children’s entertainer and family bonfire. On Monday and Tuesday during the day, the 9 doctors from our Medical and Scientific Advisory Board that SDSF invited, presented their latest research and information on SDS. They also helped open wonderful discussions between themselves and parents. While the parents were busy during the day, the older kids were busy with “camp”. Camp included archery, mini golf, waterfront activities (canoeing, kayaking, paddleboats, and swimming), climbing the rock wall, volleyball, arts and crafts, playing on the playground, computer time, and the challenge course. The toddlers and nursery age kids were lovingly taken care of by the volunteers in their own play spaces which provided for their every need.

At night there were special activities to reconnect the families to each other after a long day as well as to other SDS families. There was a masquerade dance with a DJ, carnival night, talent show, and a celebration ceremony put on by the kids and their counselors. There was also free time for the parents to participate in games organized for them or they could just relax in their individual suites that provided perfect accommodations for up to six. All of the meals were taken together in the dining hall which provided a much needed relief from cooking for 5 days. There was even an adult-only dinner served by candlelight with karaoke music for entertainment that gave parents a little respite and time to themselves.

Camp Sunshine thought of it all. They found a way to give families a vacation from cooking, cleaning, and worrying about doctor’s appointments, etc. and giving them time with their loved ones as well as new friends and doctors in a fun, supportive, loving, connecting environment.

PERSPECTIVE ON CAMP SUNSHINE FROM

MIKE, DEBBIE AND COLLIN BROWN

For our family, the conference at Camp Sunshine was very fulfilling. This past fall, our son, Collin, at 18 months old was diagnosed with SDS. Therefore, we are still learning so much about this disease. It was so nice to have the opportunity to speak one-on-one with Specialists about current studies being done to try and find a cure. Equally rewarding were our conversations with other Moms and Dads that provided us with irreplaceable information. Although we felt we were always on the go at Camp Sunshine, we still felt at ease, thanks to its relaxing environment. All in all, we came away with so much more than we arrived, a great deal of information and several new friends.
SYNOPSIS OF DOCTOR/SCIENTIST’S SEMINARS FROM CAMP SUNSHINE

Two of the five days at Camp Sunshine in Casco, ME were dedicated to a series of seminars presented to families by invited doctors/scientists by SDSF. These were wonderfully informative and enlightening as usual, whether you were a first time attendee or a five timer. The whole session was opened by Dr. Peter Durie, a tireless and dedicated doctor/scientist who has been working on SDS medical issues for over ten years. He was the first chair of our SDSF Medical and Scientific Advisory Board and is still on the board heading up the organization for an international SDS database. He is a gastroenterologist but has his hand in many aspects of this disease. He reviewed the basics of the pancreatic insufficiency issues within SDS while saying that SDS isn’t necessarily a nutritional disorder because the patients’ growth curves (height and weight) are typically in good ratio with each other.

Dr. Roberto Mendoza spoke next from the Hospital For Sick Kids in Toronto, Canada. He reviewed the skeletal dysplasia that can accompany SDS. He spoke of the osteoporosis issues as well as growth plate issues that are both affected by the malabsorption of vitamins D and K. He does not think that higher doses will correct the osteoporosis due to the basic defects in the osteoblasts. He also believes that although most patients have some skeletal issues, most improve and/or get rid of them as they age. He believes that more studies certainly need to be done to provide more information.

Dr. Blanche Alter, a member of the SDSF Medical Advisory Board and employee of the National Cancer Institute where she is running a bone marrow failure clinic spoke next. Her seminar focused on the morphology of SDS into leukemia, myelodysplasia and marrow failure. She also spoke about the use of steroids and GCSF/neupogen (neutrophil elevating drugs) and how they affect different patients.

We also received information from Dr. Elizabeth Kerr, psychologist from the Hospital For Sick Kids in Toronto, Canada. She visited many patients in the hospital and their homes to give them a series of psychological testing based on the premise that there are certain learning patterns of kids with SDS. She presented her results at the conference and made recommendations as to what parents might do to help their kids fare better in school. (Recommendations presented in following article).

Dr. Fred Goldman from Children’s Hospital of Iowa followed Dr. Kerr with information about bone marrow transplants and the role of GCSF (medication) therapy to elevate neutrophils. There is much controversy surrounding this drug and Dr. Goldman gave us his view of the positives and negatives of how the drug works. If he had a choice of using prophylactic antibiotics or GCSF, he would use GCSF. But this does cause the platelets to go down and there is a risk of the patient potentially morphing into having clonal abnormalities and/or leukemia that has been mentioned. Thus there is risk with everything and each patient is different.

Dr. Glogauer, D.D.M., from Toronto was unable to attend the meeting but sent along his data with Dr. Durie to explain. His findings have shown that people with SDS do get mouth sores from gum disease, more tooth decay, recurrent canker sores, bleeding gums and can have congenitally missing teeth and dysplastic teeth which means they haven’t fully formed the enamel. There can also be internal resorption of these teeth. Dr. Glogauer is trying to determine whether these issues arise from having low neutrophils or a congential defect stemming from the SBDS gene. He is continuing to study these issues.

Next up was Dr. Johanna Rommens, PhD, who is currently the chair of the SDSF MSAB and has been a member for many years now. She spoke to us about her studies with the SBDS gene and mouse models. The SBDS gene, founded by Dr. Rommens and her team back in December of 2003, is located on the 7th chromosome and is present in every living species except bacteria. It seems to be present where genes are
growing very quickly. Dr. Rommens is trying to create a mouse with SDS in order to then figure out how to rid the mouse of the disease. It has been a tricky process and she is constantly working with different proteins to figure out how to complete this task.

Dr. Richard Harris who chairs the transplant unit at Cincinnati Children’s Hospital has performed many bone marrow transplants for SDS patients and has had great success. He listed his main reasons for transplanting a patient: when a patient has severe, chronic neutropenia and does not respond to high doses of GCSF; severe pancytopenia; high grade MDS; and leukemia. He also spoke of the organ toxicity that can accompany the use of cytotoxan, the chemotherapy drug used as a preparative therapy prior to a transplant, and radiation, both of which can be damaging to the heart and lungs.

Our last seminar was Dr. Durie speaking to us about the international database/registry organizing meeting that he held in Verona, Italy back in February. This meeting helped to create a consortium and agreement on establishing and maintaining national databases on SDS patient data in each country which would contribute to an international registry. Many details are still being worked on including the legal and ethical aspects of obtaining the data, ownership, management, governance and funding but the work being done so far is important to the future of all aspects of this disease. By having a registry in place, doctors can access data for clinical markers and information for their patients, and scientists can access the data to enhance their research which can ultimately help everyone down the path towards a cure.

But lest you thought that was all, there were afternoon break out sessions. We broke into smaller groups and had more intimate discussions with the doctors about their specialties. Dr. Durie and Lynda Ellis (nurse extraordinaire and organizer of all things SDS in Toronto at the Hospital For Sick Kids) spoke about enzymes and nutrition; Dr. Alter and Dr. Elghetany discussed hematology/pathology reports and how to read them; Dr. Kerr took individual questions about learning behaviors and needs; Dr. Goldman and Dr. Harris addressed bone marrow transplants; and Dr. Mendoza and Dr. Rommens discussed genetics and genetic counseling.

Although I could go into detail for pages, I hope that you have gained an insight of what was presented during the family conference. Everything we heard was incredible and it was fantastic that these doctors took the time out of their busy summer schedules to share their knowledge with us. If you have any questions about the conference, please feel free to contact our board and we will be happy to answer them to the best of our knowledge. 4sskids@shwachmandiamond.org
FAMILY CAMP PICTURES

07.12.2006

JUL 13 2006
SDS PHYSICIAN/SCIENTIST PROFILE

DR. JOHANNA ROMMENS
Program in Genetics & Genomic Biology

My name is Johanna Rommens. I have been a researcher at The Hospital For Sick Children and the University of Toronto for most of my professional career.

From early days, I recall liking school a lot with math and science as my favorite subjects. My parents tell me that I tried their patience many times with the plants, caterpillars and other living things that I used to keep and study in our house. They gave up complaining when I promised not to study anything contagious and eventually became supportive of all my projects. My formal training has not been in medicine but in basic science including areas of chemistry and biology, especially genetics. Genetics deals with inheritance of genes from one generation to the next of all living things, and human genetics specially applies to people.

There are two major components to the work I do. One is to carry out research and the second is to teach university students. Both components are related as post-graduate university students work and learn directly in my laboratory and help to carry out the experiments for our studies on Shwachman-Diamond Syndrome (SDS). These students are earning Masters and Doctoral degrees and will go on to carry out jobs related to scientific or medical research in the future.

SDS is a rare disease but it occurs in populations from all over the world. As genetic diseases go, SDS was described late, only in the nineteen sixties. However, it was not really until the seventies and eighties that advances in science gave methods to understand genetic diseases in detail. With this ability to study chromosomes (these are the structures in our cells that carry genes from one generation to the next), one of the earlier steps of our research was to collect blood samples from families like yourselves.

These contributions were very important, as they were needed to help begin to sort out the cause or causes of SDS. We could then track down on which chromosome and subsequently which of our 28,000 genes was affected.

We found that the major gene that causes SDS, or ‘SBDS’ as we call it occurs on our seventh, largest chromosome. While we are able to find this major gene, we also found that it had not been studied before. It does not look like other human genes that we know so it remains unknown just exactly what its role is in our tissues. One very obvious feature though, is that this gene or a very similar one is found in many living things, this includes all animals, plants and yeast, in fact, everything but bacteria and viruses.

Presently, we are trying to figure out exactly what the gene and its protein product does in our organs. This is important to know so we can then try to determine what happens when it does not work. One way to do this is to build model systems that can be studied in detail. We can add or remove the SBDS gene from cells that are growing in the laboratory to see what happens. We also can mimic changes found in the SBDS gene in cells or in an animal such as a mouse, and then study what happens throughout its life. From work carried out to date, it appears the SBDS is involved in allowing the building blocks of our tissues (or cells) to interpret genes and the genetic program. It also appears that SBDS is most necessary when cells need to grow quickly.

I have always considered the work that I do as a great privilege. I have the pleasure of working with people that are very committed to what they do and I am able to think about and learn new things. These are just a couple of reasons why I work on SDS, the major reason relates to how much there is to do and there remains so many aspects of SDS that need to be investigated and improved ... so back to work for me.
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 psbishop1@yahoo.com or call me at (515)252-7445. 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!

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

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.

Grand Prairie, TX
Reynoldsburg, OH
Wheaton, IL
Churville, PA
Dallas, TX
Lake Winnebago, MO
Baltimore, MD
LaMesa, CA
Gouldsboro, PA
Stamford, CT
Anoka, MN
Blackstone, MA

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.
RESEARCH

University of Texas Medical Branch-Galveston, Texas

Dr. Tarek Elghetyan, 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 Figoeroa 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.
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

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Division of Hematology
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660 S. Euclid Ave., Box 8125;
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St. Louis, MO 63110, USA
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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.
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.

SDSF DREAM BRACELETS FUNDRAISER

I am making Swarovski Crystal bracelets with Sterling Silver beads with SDSF charms as a fundraiser for SDSF. I have several colors to chose from and can customize for size. I have added a Premier Bracelet style along with the Standard Bracelet style. The Premier includes different shaped Swarovski Crystals, in clear only, with the colored stones. The shapes are cubes, cones, disc, and larger stones. They really add to the bracelet. Include the size you wish. The Standard bracelet is still $30.00 and the Premier is $33.00. There is still a $3.00 shipping charge per order to the same address, additional addresses will be an extra shipping charge. You can order your bracelet in honor of your child and a special card will be sent. It will also appear in the newsletter. An order form can be found on the web site for your convenience, www.shwachman-diamond.org. Sorry no COD’s or credit cards. Please allow 2-3 weeks for delivery. Make your check payable to Jenny Jenuwine. All proceeds go to SDSF. Thank you for your support in our dream to find a cure. Any questions, please contact me directly.

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

COMMUNITY NEWS FUNDRAISER

The Van Brunts have held their 9th annual Barn Dance fundraiser In September and raised $35,215 for SDSF.

Don’t forget to let us know about any upcoming fundraiser big or small to add to our community page on the web site. We want to let our SDS community know what wonderful things you are doing.
Thank You to our Donors
(donations March 10, 2006 - September 20, 2006)

Student Council of Norman J. Levy, Lakeside Elementary School in Merrick, NY

Kiwanis Club of Eastpointe

Solvay Pharmaceuticals

Chicago Area Combined Federal Campaign
Trident Area Combined Federal Campaign
CFCSENC Fund - 0656
Southeastern Michigan Area CFC
Combined Federal Campaign of Kitsap & Mason Counties
CFC of South Hampton Roads
Combined Federal Campaign of Maricopa County
Combined Federal Campaign of Louisville, KY
Heartland Combined Federal Campaign
Monadnock United Way
Global Impact CFC Overseas
CFCNCA of Alexandria, VA
United Way of Southeastern Connecticut
Fort Polk - Central Louisiana CFC
CFC Fall 2004
Global Impact CFC of the National Capital Area
Orange County’s United Way
United Way of Berkeley & Morgan Counties
United Way of Metropolitan Dallas, Inc.
United Way of Rhode Island
United Way - PCFO
United Way of the Midlands
United Way of Mesa County, Inc.
United Way of Delaware

Eastern Niagara United Way

Carol Bush
Jeffrey Costello
Judith DeMaillie
Robert Gilsinan
Mary Grabowski
Nancy Harrod
Virginia Hughes
Sharon Lamb
Louise Mangan
Carol Rinaldo
Robert Roth
James Stephens

In Honor of Gracie Van Brunt

Gracie Fund

In Honor of Malcom Scates

Cary Scates
Betty Scates
Joan More
Marty More

In Honor of Troy & Kelsey DeBoer

Jake & Lois DeBoer

In Honor of Dylan Kolar

Town of Islip, Dept. of Parks, Recreation & Cultural Affairs Summer Playground Program
Shwachman Diamond Project

In Honor of Ashley Chiochetti

Thomas & Deborah Chiochetti

Allstate Giving Campaign

Shana Hovey

Deutsche Bank Americas Foundation
Matching Gifts Program

Mark McHugh
BRACELET PURCHASES
(March 10, 2006 - Sept. 20, 2006)

In Honor of Brittany Kadel
Debbie Kadel

In Honor of Tyler Knoble
Sindy Knoble
Vicki Brown

In Honor of Dylan Kolar
Donna
Lilliana
Debra
Kathy
Nancy
Carol
Heather
Jen
Kristine

In Honor of Danny Rohe
Pat Whatlen
Debi Ratterman
Alecia

In Honor of Amanda Lamb
Sharon Lamb

In Honor of Logan Stone
Susan Utz

In Honor of Gracie Van Brunt
Blair Van Brunt

In Honor of Erin Johnson
Joni Johnson

In Honor of All SDS Children and Adults
Nancy Cincotta
Anna Gould

Thank you to all families that purchased bracelets at Camp. If I have forgotten your information, please contact me at jengrls2@klondyke.net
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
Tel: 024-76345199 Fax.: 024-76345199
email: enquiries@shwachman-diamond-support.org
http://www.shwachman-diamond-support.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
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 opcrecdb@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

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.

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.
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.