Shwachman-Diamond Syndrome Foundation

Cure For A Future

NEWSLETTER

FALL 2009

Well, it is almost the end of the year and I can happily say that we received many donations this year in spite of the terrible economy and our whole SDS community should be so happy!!! Thank you to all of you who have created fundraisers, bought rubber and crystal bracelets, sent in a check, donated through organized giving due to the passing of a loved one or exercised your stock options. Every little bit counts and it is all going towards great work and research!!!! We still have the holiday season in front of us and if you would like to give a really thoughtful gift to a special person or family member that says you have donated to SDSF in their name, we will send out the card for you along with a label pin. THANK YOU!

Since our last newsletter we have asked Donna Garfield, our newsletter editor, to become a member of the Board and we are delighted that she is joining us!!! Look for an article about her later in the newsletter. We have also been blessed with the donation of time from a long time SDS community mother, Shani Allen, who has offered to be a Family Follow Up Coordinator for us. She has been contacting people who have contacted us long ago to see how they are, do they need anything, and what is going on with them and their families. This is a great service to all of us as she tries to track down people who may have gotten lost in the shuffle of board members who live all across the country, working on a volunteer basis for the board and who do the best they can to keep up with everyone. Also, Kim Zajac has offered to be our new Regional Contact for the Mid Atlantic region and we are so thankful for her help as well!!

We have also posted a memorandum about the H1N1 virus on our web site. After collaborating with our Medical Advisory Board, we all came up with a possible response to how an SDS patient might deal with the virus. As always, please check with your care provider for the medical advice that is the right course of action for you.

And last but not least, we have a generous couple who will donate their time and skills to translate any inquiries that we may receive from Spanish speaking families or their doctors for them into English. Read more about them later in the newsletter.

FUNDRAISING CONTEST: We have decided to hold a fundraising contest starting in January. We will send out specifics via email in December but we wanted to get you thinking now about how you and your family can raise the most money for SDSF research, doctor and family education, family support via the family camp/conference and materials, and more.

Thank you for letting us serve you. By supporting SDSF, you are supporting yourselves.

Blair Van Brunt

Shwachman-Diamond Syndrome Foundation

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SDS REGISTRY

EXCITING STATUS UPDATE

FROM THE

SDS REGISTRY TEAM!

We are happy to report that the SDS Registry opened to patient accrual in December 2008. Since the primary site in Seattle was required to open before Toronto and Cincinnati could proceed with their IRB/ethics board submissions, of necessity patient accrual has thus far centered on Seattle. Conditional IRB/ethics approval has been obtained at Cincinnati and Toronto so the SDS Registry is poised to open soon at those sites. In the meantime, all three sites have been active in recruiting and referring patients to the Registry. As a result of the combined work of all the investigators, patient accrual (30 patients consented) has already exceeded our projections for the first year of the Registry. Our thanks to the SDSF for your efforts to get the word out to the SDS community regarding the Registry!

We are also fortunate that an abstract describing the SDS Registry has been accepted for presentation at the upcoming meeting of the American Society of Hematology. This will be a wonderful opportunity to educate pediatric and adult hematologists about SDS and the Registry.

In addition to the 30 registered patients, we have received 9 peripheral blood samples and 11 bone marrow aspirates for the repository. We have received medical records for 10 patients. This year, efforts focused on designing the data extraction forms and the database after extensive research into other successful Registries currently in operation. We would like to thank all the many outside investigators who graciously shared with us their experience and expertise. We also sought input from biostatisticians experienced in Registry-based research and from database designers experienced in clinical research. In the spirit of international cooperation, we shared the data extraction forms developed for this Registry with the worldwide SDS research community. We hope that capturing a common set of data in a similar format will strengthen the efforts of each country.

In year 2 of the Registry, we will open the Registry for accrual at all three sites. We will also pilot the data extraction forms and entry into the database with the goal of running a test run query of the database. We will continue development of the database for the patient questionnaire. We are consolidating our efforts with that of the SCNIR, our partner Registry, to streamline costs and workload. Another important goal for year 2 is to have the Registry website go live. To be consistent with the SDS Consensus Guidelines, we will defer full completion of the educational entries for the web site pending final consensus. In the meantime, we will provide a link to the SDSF web site for family educational materials.

SDSF would like to encourage every patient with SDS to register in the North American Shwachman Diamond Syndrome Registry. The more data that is available to doctors and researchers, the faster they can help with diagnosis, treatments and hopefully a cure!

How can you participate in the SDSR?
Patients can enroll in the registry by contacting the Registry coordinator, Melissa Alvendia at 206-667-6965 (toll-free 1-866-792-5876).

After signed informed consent is obtained, the patient or parents (if the patient is a minor) will receive a brief questionnaire which will be updated annually. Additional information is obtained from medical records, laboratory and pathology reports. Clinical samples are obtained when blood, bone marrow or other tissues are being obtained for clinical care, so no extra procedures are involved.

In addition, enrollment will soon be available through additional centers including Toronto and Cincinnati.

THE H1N1(SWINE) FLU VACCINE INFORMATION WITH REGARDS TO SDS

According to the CDC, people of any age with certain chronic medical conditions should get the flu/swine flu vaccine but this is certainly a decision that you are your doctor can make together. There are some people who should not get a flu/swine flu vaccine without first consulting a physician. These include:

- People who have a severe allergy to chicken eggs.
- People who have had a severe reaction to an influenza vaccination.
- People who developed Guillain-Barré syndrome (GBS) within 6 weeks of getting an influenza vaccine.
- Children less than 6 months of age (influenza vaccine is not approved for this age group), and
- People who have a moderate-to-severe illness with a fever (they should wait until they recover to get vaccinated.)
- SDS Patients whose blood counts are very abnormal compared to their usual counts
- SDS patients who are on some sort of steroid

If you decide that getting the flu/swine flu vaccine is the right option for you and your family, here is some information that you need to know about the differences between the vaccines delivered by shot and by nasal spray.

It has been advised to our SDS community to get shots only. The nasal spray is a live, attenuated virus and should only be given to healthy people between the ages of 2-49 years of age. One of our Medical Advisory Board members has recommended that no one in the household should get the live attenuated virus if someone who also resides in the household may immunosuppressed..

According to the CDC, "Simultaneous administration of inactivated vaccines against seasonal and novel influenza A (H1N1) viruses is permissible if different anatomic sites are used. However, simultaneous administration of live, attenuated vaccines against seasonal and novel influenza A (H1N1) virus is not recommended."

If you have more specific questions about the flu vaccine and what is right for you, please call your doctor and please refer to the CDC web site http://www.cdc.gov/FLU/protect/keyfacts.htm for the regular flu vaccine info and http://www.cdc.gov/h1n1flu/vaccination/acip.htm for the Swine flu.

IS YOUR CHILD STILL UNDIAGNOSED?

The National Human Genome Research Institute at the National Institutes of Health (NIH) is sponsoring a study that seeks to learn more about how parents of children with an undiagnosed medical condition think and feel about their child's condition. They hope that this knowledge will improve the health care and counseling for these parents. Men and women who are 18 years or older and have at least one child with a medical condition that has remained undiagnosed for more than 2 years are needed to take part in this study. Participation involves one survey that takes about 45 minutes to finish. The survey can be taken online or a paper copy can be mailed to you. For additional information about this study, you may review the Notice to Participants <http://www.surveymo nkey.com/ StudyNoticeUncer tainty> disclosure.

If you have questions, please contact:
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SDSF BEHIND THE SCENES

DONNA GARFIELD

My name is Donna Garfield. I have just recently joined the SDSF Board of Directors, but I'm not new to SDSF. For the last several years I have coordinated the newsletter and been available to those new to SDS as a regional coordinator. I have two sons, my oldest Chris is considered SDS-like, since he does not have any of the "known" SDS genetic mutations. He is 22, recently graduated from Virginia Tech with his BS in Mechanical Engineering and is currently attending grad school at the University of Virginia. I am a registered nurse and currently working at the National Institutes of Health. I look forward to becoming more active in supporting the SDS community.

KIM MCDOWELL

Our invaluable, wonderful assistant!

My name is Kim McDowell and I have been the Administrative Assistant for SDSF for almost 10 years. I live in upstate New York with my husband and 5 children, Morgan 12, Kendra 10, Trinity 7, Sarina 4, and Brody 2, none of who have SDS. I am a stay at home Mom and also the Recreation Director of my community, coordinating extracurricular sports activities for students. I got involved in the Foundation through a co-worker of my husband who has a child with SDS and was on the Board of Directors at that time.

Some of the work that I have been doing for these past 10 years includes having been the bookkeeper for 8 years, layout editor of the newsletter, creating and maintaining the databases for the lists, maintaining and mailing out New Family Packets, answering new family inquiries via email, maintaining relationships with our bank and depositing all checks and writing all the thank you notes for our generous donors. I enjoy working for the SDS community and am here to help in any way I can. If you need to contact me for any reason, please email me at kim@shwachman-diamond.org.

SPANISH TRANSLATION COMING TO SDSF

From time to time we get requests for information from Spanish-speaking families who have little or no English proficiency. In the past, we couldn't offer much but now we are very excited to announce that Tara Orzolek and David Vargas Lowy have volunteered to translate Spanish communications for SDSF as their gift to help our SDS community.

Tara is a writer and professional translator and David is a medical doctor currently doing a research fellowship at the Brigham and Women's Hospital in Boston. Tara and David are parents of two young girls and live in Newton MA. As well as providing translations for emails to SDSF, they have started working on translations of key parts of our website. You should start to see those appear over the next few months.

Meanwhile, if you or someone you know wish to communicate with us in Spanish, you may do so via the usual contact points: info@shwachman-diamond.org or through the web-based forms on our website.

SDSF estrena traducción al español

De vez en cuando recibimos peticiones de información de familias hispanohablantes con dificultades para entender el inglés. Hasta ahora, no teníamos los recursos para ayudarles, pero nos complace anunciar que Tara Orzolek y David Vargas Lowy se han prestado voluntarios para traducir las comunicaciones en español con SDSF en forma de regalo para la comunidad de SDS.
Tara es escritora y traductora profesional y David es un médico que actualmente realiza investigación en el Hospital Brigham and Women de Boston. Tara y David son padres de nos niñas pequeñas y viven en Newton, MA. Además de ofrecer traducciones de correos electrónicos para SDSF, han comenzado a traducir las secciones más importantes de nuestro sitio web. Empezarán a aparecer en los próximos meses.
Mientras tanto, si alguien necesita comunicarse con nosotros en español, puede hacerlo por nuestros medios de contacto habituales: info@shwachman-diamond.org o a través los formularios de nuestra página web.

THE NATIONAL ORGANIZATION OF RARE DISEASES (NORD) NEEDS YOUR HELP TO ABOLISH LIFETIME INSURANCE CAPS!

The following article is reprinted from NORD. SDSF is not taking a formal position on this but wanted to let you know of the opportunity to support this bill. If you decide to support the bill, please adapt the phone script to fit your situation.

As you read this, key Congressional leaders are behind closed doors determining what YOUR healthcare options will look like next year.

The time to act is now, by joining NORD and its Member Organizations in the following:

- Call the switchboard of the U.S. Congress at (202) 224-3121 and ask for your Representative's office.
- Ask your Member of Congress to call Rep. Patrick Kennedy's (D-RI) office to sign on to his letter to abolish lifetime caps.
- If you live in North Dakota and have been affected by lifetime insurance caps, please share your story with Senator Byron Dorgan (D-ND). Sen. Dorgan may also ask the Senate to abolish lifetime caps and will need these stories to explain the issue to his colleagues.

You can use the following phone script in your call to your legislator.

And…remember…the ultimate authority of the U.S. Congress to act resides in YOU! Together, we can make a difference!

PHONE SCRIPT:

Good morning/afternoon. My name is _____ and I am a constituent living in (city, state). I have a rare disease.

I am calling to bring to your attention a letter that Rep. Patrick Kennedy is circulating in Congress. This letter calls for the immediate abolition of lifetime insurance caps in the House health care reform bill, HR 3200.

As this bill is currently written, patients may have to wait up to nine years after reform is enacted to see those lifetime caps eliminated. People like me who have rare diseases face possible financial ruin if lifetime insurance caps are not immediately abolished. This issue is very important to me.

Will Representative (insert name of your Representative) sign on to Patrick Kennedy's letter?

FEDERAL, POSTAL & MILITARY EMPLOYEES - DON'T FORGET TO DONATE TO SDSF THROUGH THE COMBINED FEDERAL CAMPAIGN # 10799

"The mission of the CFC is to promote and support philanthropy through a program that is employee focused, cost-efficient, and effective in providing all federal employees the opportunity to improve the quality of life for all. As the world's largest and most successful annual workplace giving campaign, each

year, more than 300 CFC campaigns throughout the country and internationally help to raise millions of dollars. **Pledges made by Federal civilian, postal and military donors during the campaign season (September 1st to December 15th) support eligible non-profit organizations that provide health and human service benefits throughout the world**. CFC Charities are organizations with status as tax exempt charities as determined by the Internal Revenue Service under 501 (c) (3) of Title 26 of the United States Code that provide health and human services and that are determined to be eligible for participation in the CFC. Organizations may apply and be listed in the CFC brochure as either a local, national or an international unaffiliated organization or as a member of a local, national or international federation. Charities that apply to receive funds through the CFC are required to submit to extensive review of their financial and governance practices prior to acceptance. This eligibility review has helped set standards for participation in giving initiatives that transcend the community."

Although this is an arduous process and application for SDSF to go through each year, it is an invaluable resource for donations. Thank you to all the families who donate through the CFC.

E-MAIL YOUR FRIENDS TO ENCOURAGE MORE CFC DONATIONS

According to Robert Handloff, Ph.D., Manager of the Combined Federal Campaign at the Library of Congress, the main reason people don't contribute to the CFC is: "Nobody asked me." So, how do we encourage federal employees to donate to CFC? Ask them!!!

Here is an e-mail template that you can use to e-mail to 10 friends who can pass it on to 10 friends, etc. urging them to designate SDSF on their CFC form this fall when the designation decisions are called for.

Dear Friend,

If you know someone who is a federal employee, please ask them to **designate #10799** on their CFC form to pledge their donations to Shwachman - Diamond Syndrome Foundation, the patient advocacy, research-granting support organization that my family is a part of due to our child's diagnosis of SDS, a rare bone marrow and pancreatic insufficiency disease. You will be helping not only our child but many children with SDS all over the country. Go to **www.opm.gov/cfc** to see how your federally employed friends can participate. Please e-mail 10 friends and ask them to e-mail 10 friends and together we can support this effort. Somewhere amongst all the e-mail addresses, there will be a federal employee who will be willing to help. Thank you for your kindness.

Just cut, paste and edit to your liking and hopefully we will see our yearly donations rise. In fact, we can send our non-federally employed friends e-mails reminding them to donate through our web site too. You just need 10 e-mail addresses. Thank you for all your support.

DONATE YOUR FREQUENT FLYER MILES TO HELP A FAMILY GET TO CAMP SUNSHINE

by Joan Mowery

In light of the current economic situation, I am starting a collection of frequent flyer miles to help send families to the next SDSF family conference (summer 2010) at Camp Sunshine in Casco, Maine. The week at Camp Sunshine is completely free for all families- they only have to pay travel to and from Maine. For you new families, the conference is a chance to meet other families, meet many members of the SDSF Medical Advisory Board and let the kids be with other SDS kids - invaluable! Any family who has gone to camp can tell you how they have benefited from the experience.

In this effort to help, I am asking anyone who has frequent flyer miles or other free transportation vouchers to donate them so that more families have

the opportunity to attend this important conference. If you are in contact with any organizations that might help a family get to camp, I could use their help too. Please contact me, Joan Mowery, at jmlmowery@yahoo.com

Many thanks for your consideration of this wonderful way to help our SDS community.

FRIENDSHIP DRAWS RESEARCHERS TO GET INVOLVED WITH SDS!

Blair recently asked 2 researchers who have been involved with SDS for many years how they initially became interested in SDS research.

This excerpt below was co-written by Dr. James Huang, Director of Pediatric Hematology, University of California, San Francisco, San Francisco, CA and Dr. Paul de Figueiredo, Ph.D., Faculty of Genetics, Program in Biotechnology, Faculty of Molecular and Environmental Plant Sciences, Department of Veterinary Pathobiology, & Department of Plant Pathology and Microbiology, Texas A&M University, College Station, Texas. Here is their story:

Jim and Paul grew up together in Houston, and have been close friends for more than thirty years. They separately received training on the coasts (New York, Baltimore, Boston, Seattle), and were excited to return with their families to Texas after many years away. After residency at Johns Hopkins and Fellowship at the Dana Farber and Boston Children's Hospital, Jim took a faculty position at Baylor College of Medicine. Paul returned to Texas a few years later to establish a research lab at Texas A&M.

New faculty members are sometimes provided "start-up" packages to help jump-start their research careers. The package that Texas A&M provided Paul was very generous, and he became excited about using a portion of it to pursue a project with his good friend Jim, who was now living only 90 miles away. At the same time, Jim began bombarding Paul with stories about his research on Shwachman Diamond Syndrome. Jim had trained as a yeast cell biologist in fellowship because he was convinced that Baker's yeast is a powerful model for studying human disease.

At the time that the Shwachman gene was cloned, Jim was primarily studying cell cycle regulation in yeast. He was immediately drawn to the Shwachman field for a number of reasons: he had begun taking care of a number of Shwachman patients after he moved back to Texas; he had been inspired to enter Pediatric Hematology Oncology in part because he trained with Dr. Frank Oski (one of Drs. Shwachman and Diamond's coauthors in their original description of the syndrome) during his residency; and his good friend from residency and fellowship, Dr. Akiko Shimamura (now at the Fred Hutchinson Cancer Research Center in Seattle), encouraged him to develop a yeast model since the gene function was unknown, and he believed that he might have a unique approach to studying the disease. His approach involved inducing the "disease state" into a very simple organism (Baker[1]s yeast), and then using insights garnered from his yeast experiments to discern why SDS mutations lead to disease.

Though Paul had never heard of the disease (unlike Jim, Paul had neither received training in hematology nor worked with the powerful Baker's yeast system), he could tell from Jim's enthusiasm that this project was perfectly suited to his rare combination of skills. They had such fun exchanging ideas on the yeast project that they quickly recognized that Jim's SDS research project might provide a unique route for two old friends to work together and perhaps impact patients' lives."

Dr. de Figueiredo has recently completed an SDS project with the Molecular Discovery Center at Emory University and is currently working through The Broad Institute in Cambridge, Massachusetts, to advance potential SDS therapeutic lead compounds toward clinical application. Dr. Huang, now

the Director of Pediatric Hematology at the University of California San Francisco School of Medicine, is poised to take promising compounds uncovered in the laboratory directly into clinical trials. They are excited about their collaborative effort and are hopeful that together they will be able to develop new approaches and therapeutics to help SDS patients.

LEARNING PATTERNS OF CHILDREN WITH SHWACHMAN DIAMOND SYNDROME

In 2000 SDSF funded a research study by Dr. Elizabeth Kerr on the behavioral phenotype in SDS. Her research has recently been accepted for publication. In anticipation of its imminent release we are reprinting our earlier newsletter article (originally printed in our 2007 Winter edition, http://www.shwachman-diamond.org/newsletters/Winter2007.pdf) which summarized her early research. An abstract of the upcoming article can be found at http://www.ipeds.com/article/S0022-3476(09)00901-9.

At the 2006 SDS Family Conference, Dr. Elizabeth Kerr gave a very interesting talk focusing on the learning patterns of children (ages 6-17) with Shwachman Diamond Syndrome. Based on the findings for her research, Phonological Processing, Visual Processing, Attention, Flexible Problem Solving, and Behaviour were among the types of difficulties that can be experienced. As such, the following recommendations were made:

If you notice specific difficulties, the earlier your child receives support or intervention the better. With a "wait and see" approach you may miss many months of valuable support.

Phonological Processing/ Reading/Spelling:

Phonological processing is related to the use of sound structures in processing written and oral language and involves rapid naming, phonological awareness (i.e., the sound structures), and phonological memory. Chronic middle ear infections almost always infer with hearing. In turn, delays in language development may be evident earlier on with continued delays in phonological processing. Weaknesses in phonological processing are related to delays in reading and spelling achievement. Research at the Hospital for Sick Children and their colleagues (e.g., "Putting struggling readers on the PHAST track" m. Lovett et. al., Journal of Learning Disabilities, 2000 Vol 33 (5), pgs 458-476) have discovered three essential components to remediation:

(1) Development of pre-requisite skills which includes letter sounds associations, identification of sight words, recognition of vowel, variant vowel sounds, and affixes. These skills are best developed if a specific block of time is set aside each to focus on them. Direct Instruction can be extremely beneficial. Lessons are highly structured and scripted. Responses are modelled for the student. The structure and repetition of the lessons leads to on-line processing of information. Teaching is reciprocal, in that, the instructor is constantly checking in with the student to ensure that he or she is learning. "Direct Instruction Reading" by Douglas Carine, Jerry Silbert, and Edward Kameenui (Prentice Hall, 1996) is one resource. Other suggestions include the grapheme level activities outlined in "Phonological Awareness Kit-Intermediate" and activities such as deletions (i.e., Rosner activities) and substitutions (i.e., Lindamood activities).

(2) Practice in word identification strategies. Specific strategies include (a) Rhyming or being taught a list of relatively simple words containing common spelling patterns (e.g., look) and then being taught how to compare

new, unfamiliar words to the list (e.g., book, nook, shook, brook, etc); (b) Vowel alert or being taught that each vowel sounds has a short and a long sound and how to use different sound to make a real word (e.g., "I see the vowel 'o" in this word. First I'll try "o" as in "go". Next I'll try 'o' as in "dog". That sounds like a read word."); (c) I Spy or being taught how to look for small words or parts for words in longer, more difficult words (e.g., identify 'bad', 'in', and 'on' in the word "badminton"); and (d) Peeling off, or being taught how to identify and remove prefixes and suffixes before using another strategy to read the root word.

(3) Use of learning to learn strategies. These strategies include verbal scripts that an individual can use when approaching a new task or word. The highly scripted approach to direct instruction exposes a child to this as do the examples outlined in number 2 above.

Visual Processing:

Visual processing is related to the ability to process, interpret, and organize visual and visual-spatial information. When an individual has weaknesses in these areas, it can cause difficulties at school and sometimes in the social realm. For a given individual, they might display some (but not necessarily all) of the following: poor internal and external organization, difficulty coping with changes in routine, difficulty with generalizations, making literal translations, being overwhelmed, having difficulty with directional concepts and co-ordination, and being readily distractible.

"The Source for Non-Verbal Learning Disorders" by Sue Thompson (lingisystems) and "Educational Care: A system for understanding and helping children with learning problems at home and in school" by Mel Levine (Educators Publishing Service, Inc) are two resources which provide information for understanding and assisting with weaknesses in visual processing. If a child's language skills are stronger, then teaching them to put labels on what they are seeing and doing can help solidify information.

Attention:

Attention is multifaceted. It refers to a number of processes including: (1) how much information a person can hold in his mind and process at one time (i.e., attention span and working memory); (2) whether an individual is able to focus on, or search for, a specified target (i.e., selective attention); and (3) how well s/he can pay attention during a mundane or boring task (i.e., sustained attention). A child who has difficulties with attention may have trouble listening when someone talks, waiting her or his turn, completing a task, or returning to a task if interrupted. By the age of 5, a child needs to be able to pay attention for a least 25 minutes in order to perform adequately in school.

TeachADHD (www.teachadhd.ca) is a new resource developed by researchers at the Hospital for Sick Children and their colleagues for use by teachers at school. It instructs on various types of attention and helps the teacher develop strategies to harness attention within the classroom.

At school and at home it will be important to:

- Follow a structured daily routine. School and household routines help the inattentive child to accept order. Keep the times for wake-up, meals, snacks, chores, naps, and bed as regular as possible. Try to keep your environment relatively quiet because this encourages thinking, listening, and reading at home. In general, leave the radio and TV off. Predictable daily events help your child's responses become more predictable.
- Maintain firm discipline. Children with attention difficulties need more carefully planned discipline than the average child. Rules should be formulated mainly to

prevent harm to your child and to others. Aggressive behavior, such as biting, hitting, and pushing, should be no more accepted in an inattentive or hyperactive child than in the normal child.

At home:

- Stretch your child's attention span. Encouraging attentive behaviour is the key to preparing your child for school. Increased attention span and persistence with tasks can be taught at home. Be sure to praise your child when he plays independently. Set aside several brief periods each day to teach your child listening skills by reading to him. Teach games to your child, gradually increasing the difficulty by starting with building blocks and progressing to puzzles, dominoes, card games, and dice games. Later, consequence games such as checkers or tic-tac-toe can be introduced. When your child becomes restless, stop the activity and return for another session later. Plan to have your child do homework and other tasks that require concentration in short blocks of time with breaks in between. Try having your child study with low-level background sound such as white noise or instrumental music. Do homework and studying away from the sounds of television, radio, or others talking but where adults can supervise.

Flexible Problem Solving:

This skill typically develops with age and is required more at school when a child reaches his or her teen years. Flexible problem solving requires the ability to generate ideas, as well as to maintain and/or shift thought processes. Individuals with difficulties in this area may have trouble dealing with ambiguity or getting "unstuck" from an automatic way of responding.

Teachers and parents can help students in this area by making the steps involved in an assignment or daily activity more explicit (e.g., provide "cheat" sheets or templates that outline the steps required to write the essay, to analyse and solve math problems etc). Embed questions designed to prompt the individual into using analytical skills. Questions can include "How did you solve that problem?" "Can you think of another way of doing that?", "What can you do to help remember that information". Teach the individual a set of questions to ask him/herself when confronted with a problem, such as: "What is my problem? What is my plan? Am I following my plan? How did I do?". In other words have the individual identify the problem, develop a solution strategy, self-monitor his or her performance and evaluate the outcome.

The Learning Toolbox website (http://coe.jmu.eud/Learning Toolbox/) is designed for secondary students with specific learning challenges (e.g., organization, problem solving, attention etc) as well as for their teachers and their parents. The student section includes tools to help the student improve in many areas (e.g. Study skills, test taking, advanced thinking, organization) by providing templates to follow.

Behaviour:

Children and adolescents who are not doing well in school may not feel good about themselves. If they feel they can't cope, they may withdraw from their friends and social activities. Social difficulties were raised by parents of children and adolescents with SDS on questionnaires. Two resources to foster social competence are:

- "No one to play with: Social problems of LD and ADD Children" by Betty B. Osman, and
- "Raise Your Child's Social IQ" by Cathi Cohen.

FAMILY SHARING PAGE

GRACIE VAN BRUNT

USES HER VOICE FOR SDSF!

Hi. My name is Gracie Van Brunt. My mom, who is the president of the Shwachman Diamond Syndrome Foundation, asked me to write a short piece about how I am using my passion of creating music and singing to raise money for SDSF. I am 15 years old now and was diagnosed with SDS at the age of 2. I was in and out of the hospital so much that I knew pretty much every nurse and doctor who worked there. Up until I was 6 or 7, I entered the hospital at least every month with some sort of infection whether it was pneumonias, meningitis, bone marrow failure, chicken pox or even a simple bug bite because my immune system was that weak. But I have been healthy (no hospitalizations) since then and have been enjoying having music in my life.

Although I have been doing music theater since I was 7, it wasn't until my wish came true through the Make A Wish Foundation that my drive to create music started. My wish was to meet Kelly Clarkson and we met in June of 2007 in LA. I was able to attend a really small concert, ask her questions, sing for her and have pictures taken with her. Things just changed from then on. I came home and started writing songs. From there, I have been asked to sing for many venues and events – a radio station, the Make A Wish Foundation's black tie gala, the ABC affiliate TV station here, guest artist in a few bands in the area and eventually I got my own gigs at a music hall in downtown Cambridge, MA where they have all ages shows (no alcohol, don't worry).

With having my own gigs now, I am getting paid a little bit of money and I have pledged to donate 5% of it to SDSF to help pay for research to cure my disease. Although this isn't nearly what my parents raise each year to help SDSF, I just want to do my part and raise what I can. So far I have donated $53.00. You can read about all of this on my web site www.myspace.com/gracievanbrunt and if you have any questions, you can email me through the site. You can also join my email list to see where I am going next. I am so excited about my future!! Not only am I doing what I love to do, but I am contributing to a cause that helps me and so many other people!!

P.S. I just entered a local talent contest last night and part of the way that a winner is picked is through email votes. I don't yet have the link, but as soon as it's ready, it will be posted on my Myspace page. If I win, I receive $100, 5% of which will be donated to SDSF.

JOIN OUR "CLUB" FOR

ADULT SDS PATIENTS

SDSF is becoming increasing aware that many of our children are reaching or soon will be reaching an age when they are no longer able or wish to be seen within the pediatric medical community. Older patients also have additional and/or different social and other concerns from children with SDS. For this reason, we are starting a "club" for older and adult patients. Some of the areas we will be approaching are establishing referrals within the adult medical community of physicians who have knowledge and/or experience in treating adult SDS and other BMF patients. In addition, the club plans to set up a referral file to direct adult patients to those who can assist them in obtaining health and life insurance as well as their medications.

We would like input from both the patients themselves and/or their parents with any information or suggestions you might have about what else can be done. Please send your thought and ideas to SDSF by either calling 1-888-825-SDSF (7373) or emailing us at info@shwachman-diamond.org.

SPOTLIGHT ON RESEARCH

YOU CAN MAKE A DIFFERENCE! THESE ONGOING RESEARCH STUDIES NEED YOUR CONTINUED SUPPORT!

SBDS PROTEIN EXPRESSION IN PERIPHERAL BLOOD LEUKOCYTES

Shwachman Diamond syndrome (SDS) is a rare genetic condition which causes a number of problems in different body organs, particularly the bone marrow (blood producing cells), pancreas (digestive gland) and bones. These problems may vary considerably from person to person, which sometimes makes it difficult for doctors to diagnose SDS. The mutated gene which causes SDS has recently been identified. To have SDS, one has to have a copy of the mutated gene on each chromosome. So far, three common mutations as well as about 50 rare mutations have been identified. Approximately 60% of SDS patients carry common mutations on both chromosomes. Others carry a common mutation on one chromosome and a rare mutation on the second chromosome. However, in about 5-15% of people with clinical findings of SDS, mutations cannot be found, even after extensive laboratory testing.

Each gene in our body acts as a code for making a unique protein. Each protein has a definite function. When a gene is defective, it either produces no protein, very little protein or produces a protein that doesn't work properly. We have now developed a way of measuring the normal SDS protein in blood cells from people without SDS. In addition, we have shown in a small number of people with SDS, that the level of this protein is absent or greatly reduced. These preliminary results suggest the possibility that we may be able to use this test to diagnose SDS. We also think that the amount of protein in blood cells might help to explain why some people with SDS have worse or different problems from other people.

To do a more extensive study of the SDS protein in blood cells, doctors at the Hospital for Sick Children, in Toronto and the Children's Hospital in Boston are seeking volunteers to join this research project.

We are seeking the following individuals to join our research study:

· People with SDS who carry the uncommon (rare) mutation on at least one allele.

- Parents of people with SDS who carry the rare mutation and/or an adult sibling who is a known carrier of the rare mutation.

· People with a confirmed clinical diagnosis of SDS but no SBDS mutation have been identified.

· People who are suspected to have SDS in whom testing remains inconclusive. This might include people who have: (a) a problem in the bone marrow but no known problem in the pancreas, or; (b) a problem in the pancreas and/or skeleton but no evidence of a bone marrow problem.

Individuals who wish to participate will be asked to:

- Sign a consent form indicating willingness to participate,
- Complete a brief written medical question-

naire,

- Submit a blood sample (we will provide a kit for your lab or physician)

Individuals will not be responsible for any costs associated with the study. The confidentiality of all study related materials will be maintained and no information that discloses the identity of the subject will be released or published without consent unless required by law. The results of the tests described above will be used for research purposes only.

To learn more about the study or to see if you are eligible, please contact the study coordinator:

Wan Ip at 416-813-6183 or wan.ip@sickkids.ca

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 call 1-800-518-8474 and ask for the Referral nurse or you may also contact SDSF for more information.

STUDIES ON THE MOLECULAR MECHANISMS OF BONE MARROW FAILURE

Bone marrow failure (BMF) syndromes such as aplastic anemia or myelodysplastic syndrome (MDS) may develop by a number of different mechanisms. We believe a genetic predisposition to aplastic anemia and MDS is much more common than currently appreciated, and 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 and how these alterations cause disease. We are collaborating with researchers at several other institutions throughout the United

States.

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 (PNH), 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.

Our study uses 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 are not 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 visit our website at http://bmf.im.wustl.edu/, or contact the study coordinator:

Jennifer Ivanovich, M.S.

Study Coordinator

Washington University School of Medicine

Box 8100, 660 S. Euclid Ave.

St. Louis, Missouri 63110,

USA

314-454-5076

jen@ccadmin.wustl.edu

Monica Bessler, M.D., Ph.D., Co-Director

Division of Hematology

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Box 8125; 660 S. Euclid Ave.,

St. Louis, MO 63110, USA

314-362-8807

Mbessler@im.wustl.edu

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

Division of Pediatric Hematology/Oncology

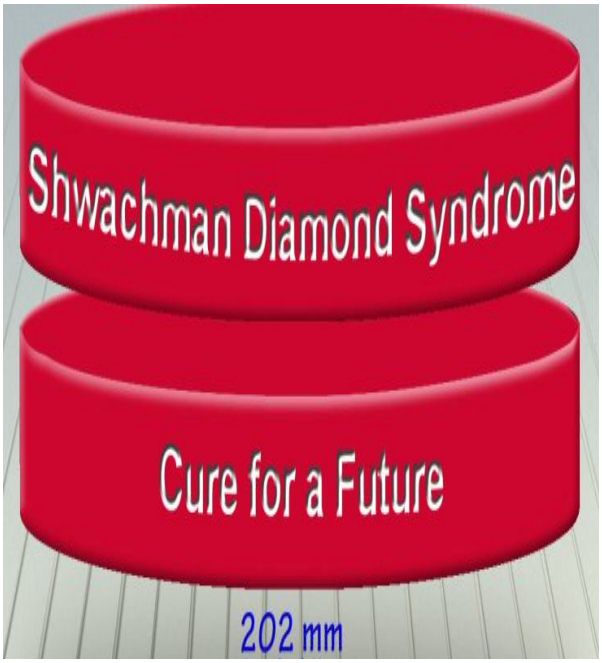
Washington University School of Medicine;

Box 8208, 660 S. Euclid Ave.,

St. Louis, MO 63110, USA

Wilson_D@kids.wustl.edu

RUBBER BRACELETS FOR FUNDRAISING AND COMMUNITY AWARENESS!!!!



I f you would like to help increase awareness of Shwachman-Diamond Syndrome in your community and at the same time assist in raising funds for medical research and family support, order some of our new Shwachman-Diamond red rubber bracelets! You can buy them in sets of 10 directly from our web site at www.shwachman-diamond.org and jsut click "Get Involved" and they will be mailed to your home. Please email us at info@shwachman-diamond.org to let us know that you have purchased any of these bracelets in honor or memory of anyone and we will publish the information in the newsletter. They are great to use as conversation starters to help family, friends, and medical professionals in your area understand about SDS and its impact on families. In addition to that, they are cool!!!!!!!!! You can give them to your friends and family or re-sell them to others. THANKS ALL!!!

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. Colors available are clear, black, dark blue, sapphire, light blue, pink, light amethyst, amethyst, tanzanite (lavender), light red, ruby, garnet, peridot (light green), emerald, white pearl, black pearl, pink pearl and birthstone colors. 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 purchase these online at: www.shwachman-diamond.org through the donate button. Please email us at info@shwachman-diamond.org to let us know that you have purchased any of these bracelets in honor or memory of anyone and we will publish the information in the newsletter. Please allow 2-3 weeks for delivery.

The bracelets are beautiful and make great gifts. What a great way to support all SDS children. Thank you for your support in our dream to find a cure. Any questions, please contact me directly.

Jenny Jenuwine
810-395-2358
jengrls2@banyanol.com

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 this 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.

McKinney, TX
Anderson, SC
Old Bethpage, NY
Rice, TX
Gorham, NH
Millbrae, CA
Ashland, KY
East Meadow, NY
Pulaski, TN
Ludlow, KY
Cleveland, TN
San Clemente, CA
Kendall, NY
Newport, WA
Middletown, NY

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!**

F.Y.I.

Axcan Scandipharm, the makers of Ultrase enzymes, ADEK vitamins, Scandishakes and many other products has included Shwachman-Diamond Syndrome in their patient support program. SDS patients who use their products may qualify for free and/or discounted products and information. Please note that Axcan Scandipharm patient support program has changed. Patients are no longer required to mail in receipts and forms in order to receive program benefits. The new card, AXCAN Rx COMPLETE card, will allow you to receive your program benefits more efficiently. To take advantage of this exciting new program card or to ask questions about it, please call the AXCAN Rx COMPLETE Program line toll-free, at 1-866-AXCAN-RX (1-866-292-2679), Monday-Friday, between 8:00 a.m. and 8:00 p.m., EST.

Thank You to our Donors

(donations July 10, 2009 - November 18, 2009)

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Established Shwachman-Diamond Groups

Shwachman-Diamond Syndrome Support - Australia
Contact: Joan Buchanan
61 03 5427 0645
email: buchanan.joan@gmail.com
http://www.shwachman-diamond.org

Shwachman-Diamond Support-UK
Contact: Sharon Clusker
Tel: 02476-345199 Fax:: 02476-345199
email: mail@sdsuk.org
http://www.sdsuk.org

Italy Association for Shwachman Syndrome
Contact: Aurelio Lococo
email: aiss@shwachman.it
http://www.shwachman.it

Shwachman-Diamond Syndrome Canada
Contact: Heather Norton
email: sdscanada@sympatico.ca
http://www.shwachman.org

Shwachman Syndrome - Netherlands
email: koster.e@hccnet.nl
http://www.shwachman.nl/

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 to our address or e-mail them to: **info@shwachman-diamond.org**

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

Thank you.

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 opcrccdb@aol.com

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

Donna Garfield -VA (703) 731-7889 or dkgarfield@verizon.net

Michelle Noble - CA: (760)947-4283 or MNoble2day@aol.com

Theresa Henle - CA: (949) 858-5662 or Theresahenle@mac.com

Cyndi Smith - SC: (803) 781-7100 or Chs5099@aol.com

Kim Zajac - PA: (570) 350-1354 or rkczajac@ptd.net

OTHER COUNTRIES

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

CHANGE OF ADDRESS OR E-MAIL

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

If you have moved please forward your current mailing address to keep our databases updated.

Either call us at **1-888-825-SDSF (7373)** or email us at **info@shwachman-diamond.org** with your changes.

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Please send you tax deductible gift to: **Shwachman-Diamond Syndrome Foundation**
127 Western Avenue, Sherborn, MA 01770 U.S.A.

Credit Card donations can be made through our website - www.shwachman-diamond.org

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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.

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