NEWSLETTER

Winter 2007/2008

Reflecting upon the Thanksgiving holiday I realize the many things that I have to be thankful for. There are too many to mention here but a couple that come to mind are; the family that surrounds and supports me and this organization! I am so thankful for Shwachman-Diamond Syndrome Foundation and what it has meant to me and my family. I can recall the exact moment I was told that my daughter had this disease and what it felt like. I remember how thankful I was that there was an established organization that freely gave me information about a disease my child had and that I knew nothing about. I found out that we were not alone on this journey and more importantly that there were people that cared about me and my family who were also somehow impacted or affected by this disease.

I vividly remember writing my first editorial when I took over the position of president a few years ago and how nervous I was. Looking back, it seems like it was just yesterday. So much has happened between then and now! I am proud to have been asked and was able to serve in this capacity and I am at peace with passing on this torch to Blair Van Brunt. I will be stepping down from the Board of Directors in January after many years of service, first as secretary and then as president. I am proud of the work that this foundation has accomplished and for the research we have supported, and the many innumerable things that we have done. I am thankful for all of the women that served before me that built the foundation for this organization so that it could continue to grow into what it is today and yet still continue to grow and change with new leadership. I am thankful for the support of foundations in other countries, the Canadians, Italians, United Kingdom, Netherlands, and Australia. There have been leadership changes in some of those foundations as well and I am thankful for all the previous board members and those that are stepping up to continue the global effort of learning more and hopefully someday curing this disease.

I would also like to announce that Jenny Jenuwine resigned her position on the board a couple of months ago. The Jenuwine's have been instrumental in fundraising for this disease. They started with coffee mugs and raffle tickets and have never stopped raising money for this foundation and this disease. I would like to thank them and their extended family for all the years of hard work they have put into their fundraisers. Jenny will continue to make the beautiful bracelets and I am sure they will still continue their super bowl fundraiser. With that, I would like to announce that Bryan Sample has joined our board in the position of treasurer. We are very excited that he has offered his help to fill the role of this important position. You can read Bryan's story in this edition.

I have truly enjoyed the many email, phone and in-person conversations and friendships that I have made over the years. I have learned and have been able to share so much with you in this journey that we are on. Thank you all for touching our life!

All the best, Debbie Kadel

Shwachman-Diamond Syndrome Foundation

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

WELCOME, BRYAN

My name is Bryan Sample and I am pleased to be joining the Board as treasurer. My wife Elvira and I are parents of Andy (14), Libby (11) and John Michael (6) who happens to be our SDS child. Since his diagnosis, we have relied on SDSF for information through the newsletters and family conferences to keep us up to date on the latest research and developments. We are happy to be involved in a more visible manner with SDSF and look forward to working on the board.

DOCTOR DATABASE

Don't forget to email us at 4sskids@shwachman-diamond.org with the names of your doctors if you feel they are doctors who others would benefit visiting. Please list their title, address, phone number and most importantly their specialty.

The larger the list, the more we help our fellow SDSer. Who knows? You could even help an existing patient find a doctor closer to home or perhaps help a doctor find another doctor who he/she can call for advice.

Thanks for helping.

For Those of You Who Didn't Get The Email Or Accidentally Deleted It

Urgent Request About the Congressional Bipartisan Bone Marrow Disease Resolution

The Shwachman-Diamond Syndrome Foundation needs your help today to move the Bipartisan Bone Marrow Disease Resolution to the floor of the House!

Earlier this year, Representatives Jim McGovern (D-MA) and Mary Bono (R-CA) introduced H.Con.Res. 81, the Bipartisan Bone Marrow Disease Resolution, to encourage the federal government to fund research and to engage in public health initiatives that give patients greater access to more treatment options and, ultimately, cures for bone marrow failure diseases.

For this resolution to pass, we need to have a broad level of support from individual Members of Congress. Specifically, the Energy and Commerce Committee, which has jurisdiction over this resolution, requires the support of 100 Members of the House to send the resolution to the floor of the House. **Now is the time to call or to email your U.S. Representative to urge him/her to co-sponsor H.Con.Res. 81.** If you do not know who your Representative is, or need contact information, please access www.house.gov and enter your zip code under the heading "Find Your Representative."

We appreciate the support of the 30 Members of Congress who have already signed on as co-sponsors (arranged by state):

Raul Grijalva (AZ) Brian Bilbrary (CA)

Mary Bono (CA) Michael Honda (CA)

Doris Matsui (CA) Pete Stark (CA)

Henry Waxman (CA) Rosa DeLauro (CT)

Chris Murphy (CT) Christopher Shays (CT)

Eleanor H. Norton (DC) Madeleine Bordallo (GU)

Jerry Costello (IL) DannyDavis(IL)

Chris Van Hollen (MD) Michael Capuano (MA)

William Delahunt (MA) Barney Frank (MA)

Ed Markey (MA) Jim McGovern (MA)

Marty Meehan (MA) Donald Payne (NJ)

Timothy Bishop (NY) Maurice Hinchey (NY)

Michael McNulty (NY) Charles Rangel (NY)

Betty Sutton (OH) Robert Brady (PA)

Sheila Jackson-Lee (TX) Frank Wolf (VA)

If you do not see the name of your representative above, please contact him/her immediately and urge him/her to co-sponsor H.Con.Res. 81!

For your convenience, a sample script that you can use when you contact your Member of Congress is below. **(Please note that we are only contacting U.S. Representatives at this time and not U.S. Senators.)**

Members of Congress do not co-sponsor these types of resolutions unless they hear from their constituents. Spread the word and contact your elected Representative today—thank you!

LANGUAGE FOR CONTACTING YOUR MEMBER OF CONGRESS

Whether you send an email or call your member's office, please give your name and address so that the office knows you are a constituent.

"Hello, I am contacting you today to urge you to co-sponsor H.Con.Res. 81, the bipartisan "Bone Marrow Disease Resolution." (*tell why you care about this resolution, i.e., "My child has SDS…, I suffer from SDS....."*)

This resolution will help to raise greater awareness of SDS, along with aplastic anemia and MDS- two diseases that can affect patients with SDS. Enactment of this resolution will encourage the federal government to fund research and to engage in public health initiatives that will improve treatment and lead to cures for these diseases.

Thank you for your help!!

SDS and IEP's

By Nancy Miller and Susan Utz

While some children with Shwachman-Diamond Syndrome (SDS) may never require special services in order to benefit from their education, a significant number of them may. Before receiving these services, the child must be determined to have a disability in one or more areas. Recognized categories of disability generally include: autism, communication disorders, dual sensory impairment, early childhood delays, emotional handicaps, hearing impairment, learning disabilities, mental handicaps, multiple handicaps, orthopedic impairments, **other health impairments,** traumatic brain injury, and visual impairments.

Children with Shwachman-Diamond Syndrome may present with medical concerns **only** or they may exhibit additional characteristics from any of the above listed disabilities. The primary disability impacting his/her education would need to be determined, as well as any related problems. In cases where health impairment was determined to be the primary or the only disability, it would need to be determined that the health impairment adversely affects the child's educational performance. This may be apparent in limited strength, vitality, or alertness as a result of chronic or acute health problems. Eligibility for special education services for this disability would be determined based on an extensive evaluation of the child's learning capability, academic strengths and weaknesses, present level of functioning, social and developmental history,

adaptive behavior, and as needed, fine and gross motor skills, and communication. It would also depend upon a review of the medical history and would require a diagnostic statement from a physician with an unlimited license to practice medicine describing the health impairment. This should include a medical plan listing any medications, procedures, and special medical needs.

In cases in which a child is identified as having a disability of any type, an Individualized Education Program/Plan (IEP) is to be written and implemented in order for the child to gain from his/her education. On its website at http://www.ed.gov/offices/OSERS/OSEP/Products/IEP_Guide/, the Office of Special Education and Rehabilitation Services (U.S. Department of Education) offers an excellent guide to the IEP process. In that document, both the process for identifying students with disabilities and the development of IEP's are explained in detail. Some of that information will be reviewed in this article.

The process involving the development of the Individual Education Program consists of the following steps:
1) The child is identified as possibly needing special education and related services through a referral or request for evaluation. This may be generated by the parent and/or teacher. Parent permission must be obtained before an evaluation can be completed.
2) The child is evaluated in all areas related to the suspected disability. The evaluation must be completed in a reasonable amount of time. If the parents disagree with the assessment, they can take the child for an independent evaluation and may ask the school system to pay for it.
3) Eligibility is determined. After reviewing the evaluation, the team (parents and school professionals) decide if the child has a disability according to the definitions stated in federal and state special education laws. Parents may challenge eligibility decisions.
4) If the child is found to be eligible for services an IEP must be written within 30 days.
5) An IEP meeting is scheduled. The parents are to be notified in advance of the time and place of the meeting, and it must be agreeable to them and the school.
6) The IEP meeting is held and the IEP is written. The team gathers to discuss the child's needs and to write the IEP. Parents may discuss their concerns with the team if they disagree with the IEP. The team must attempt to work out disputes. If the parents still disagree with the IEP, they may request mediation or a due process hearing.
7) Services are provided. The school professionals must assure that the IEP is carried out as it was written. Each of the professionals involved should know their responsibilities including the accommodations, modifications, and support they are to provide. The IEP must be implemented in no later than 10 days after it is completed.
8) Progress is to be measured and reported to the parents on a regular basis. The reports are to be given as often as that of the nondisabled children's reports.
9) The IEP is to be reviewed by the team at least once a year. This can take place more often if the parents and/or school request it. The IEP may be revised if necessary. At the end of the year, a new IEP is to be created based on the child's needs at that time. At any time, parents can make suggestions and can agree or disagree with the goals or placement. The process for disagreeing may include, as necessary: presenting concerns, requesting additional or independent testing, requesting mediation, requesting a due process hearing, or filing a complaint with the state education department.
10) The child is to be re-evaluated every three years.

Although IEP forms and paperwork may vary from one state or school system to another, they are based upon the nation's special education laws as delineated in the Individuals with Disabilities Education Act (IDEA). Specifics to be included are: 1) A statement of current performance, 2) annual goals and short term objectives, 3) the type and amount of special education and related services to be received, 4) the amount of participation the child will have with nondisabled peers, 5) how the child will participate in state/district tests, 6) the dates and places services will be given, 7) the plan for transi-

tion services for children 14 and over, 8) a statement of rights being transferred at the age of majority, and 9) the method for measuring the child's progress.

The IEP is a legally binding written document created specifically for an individual child. It is to be written by a team including, but not limited to, the parents/guardians, regular and special education teachers, school representative, and individuals with knowledge of the student or special expertise (e.g. diagnostician, Occupational Therapist, Physical Therapist, Speech/Language Therapist, psychologist, social worker, etc.). The committee may also include the child, representatives from transition agencies, family friends and relatives, and other professionals.

If the child needs specific related services in order to benefit from his/her special education services, these should also be included in the IEP and the personnel involved should be included in the process. Related services may include: audiology, counseling, medical services, occupational or physical therapy, orientation or mobility training, parent counseling, psychological services, social work, speech/language therapy (if not already considered as part of the disability), transition planning, etc. If the child's medical condition warranted it, a medical plan should be included indicating medications required and any other special medical needs listed.

The IEP team must consider certain other factors to determine their impact on the child's ability to gain from his/her education. These include things such as the child's behavior, whether he/she has limited English proficiency, whether or not he/she is blind or visually impaired, and whether or not the child requires assistive technolgoy. Assistive technology may include low, mid, or high technology equipment that allows the child to participate in the educational process (adapted computer software/hardware, communication devices, switches to activate equipment, environmental controls, mobility equipment, architectural adaptations, etc.).

In addition to the goals and short term objectives in the child's IEP, a listing should be included that gives necessary modifications/adaptations/ and supports in order for the child to benefit from his/her education. These may include suggestions on the means of communicating with or eliciting communication from the child (such as, speaking slowly to him/her, providing visual and/or auditory cues, waiting a sufficient time for the child's response, providing alternative communication systems, utilizing peer models, encouraging output, avoiding abstract language, gaining the child's attention prior to communication, responding positively to attempts, avoiding open ended questions, etc. Also included may be strategies for presenting materials, such as the use of written words/pictures/objects, the provision of information in sequential steps, the opportunities for repeated practice, the use of prompts/cues, the use of peer tutoring, the use of low or high technology equipment,etc. Assessments and assignment modifications may also be listed, for example, modifying the length or complexity of assignments, allowing for extra time, modifying question format, providing rehearsal, allowing choices, utilizing highlighting, or having allowable parts read aloud. Behavior issues may be addressed within modifications or if warranted, a separate behavior plan may be written. Strategies to assist with behavior may include methods such as the use of reinforcements or contracts, the use of visual and/or auditory cues, the use of timers, the presentation of choices, permission for frequent breaks, the use of redirection, and the provision of frequent feedback. The committee can discuss and include these or other strategies they think will allow the child to gain from his/her special education program.

By providing in Individualized Education Program to the child with a disability, he/she can be given the support needed in order to learn to the best of his/her ability. If a parent suspects that his/her child has a disability which may be adversely affecting his/her education, the school should be contacted and an evaluation should be requested to determine eligibility. This especially applies to children with chronic health problems, such as SDS, who may already be facing a variety of challenges in their young lives. As their parents, we can and should advocate for their

educational needs just as we have learned to do so for their medical needs. There are many excellent resources and individuals available to us as we learn the process for assisting our children in the world of education.

Works Cited:
"A Guide to the Individualized Education Program". 11 Jul 02. http://www.ed.gov/offices/OSERS/OSEP/Products/IEP_Guide>
"Title 511, Indiana State Board of Education, Article 7, Rules 3-16, Special Education Rules". 17 Aug 02. http://babyindstate.edu/iseas/art71.html

Additional Suggested Reading:
"What Makes a Good Individual Education Plan for Your Child?". 11 Jul 02. http://www.pacer.org/parent/iep.htm

BLOOD TESTS AND INFECTIONS

A common question for many SDS patients and their parents relates to the complete blood count (CBC) and what its findings reveal. A CBC test is used in a variety of situations as it gives information about the cells in a patients' blood - including the concentrations and other details of white blood cells, red blood cells and platelets.

A CBC may be useful if a doctor suspects an infection because of the information specifically gained about the white blood cell components. The interpretation must always be considered in context of the patient, their illness history and other medical findings. In general, infections usually lead to an elevation of white blood cells numbers, but different infectious agents affect different types of white blood cells.

The following is modified and updated from a Q&A article 'give title' originally published in 'the Nursing Journal/Newsletter'. We thank Dr. Peter Durie of The Hospital for Sick Children and our MSAB members for their summaries, suggestions and updates.

Q: How do complete blood count results help to determine whether an infection is bacterial or viral in origin?

A: The aspects of the CBC findings that support the presence of an infectious process are the white blood cell (WBC) and white blood cell differential (WBC differential) counts. The differential count gives the percentage of the various types of white blood cells that are present. White blood cells include polymorphonuclear leukocytes (also referred to as neutrophils, granulocytes, POLYS or SEGS), lymphocytes (LYMPHS), monocytes (MONOS), eosinophils and basophils.

Bacterial infections are often accompanied by an increase in the numbers of neutrophils. Immature forms of neutrophils are also often seen (referred to as BANDS), especially with acute infections. The appearance of these younger cells in the circulation is referred to as a 'SHIFT TO THE LEFT'. This occurs with many bacterial infections such as in the lung (pneumonia), blood (septicemia), spinal column (meningitis), and bone (osteomyelitis). A classic example of this type of infection is a patient presenting at the Emergency Department with abdominal pain, fever, nausea and vomiting. In this case an increase in neutrophils and BANDS support the diagnosis of acute appendicitis and a trip to the operating room can be anticipated.

Viral infections are often accompanied by an increase in lymphocytes. The WBC differential can also identify lymphocytes as being 'reactive or atypical', indicating that these cells have likely responded to a virus. This occurs in many viral infections such as influenza (caused by flu viruses) or infectious mononucleosis (caused by Epstein-Barr virus). With a viral infection, a frequently encountered scenario is a patient that complains of feeling ill with a fever and swollen glands (lymph nodes). In this case an increase in WBC counts and the WBC differential may reveal 'atypical' lymphocytes. A mononucleosis screen can be considered to rule out

or confirm an Epstein-Barr virus infection, but many other viruses will show the same findings. Analysis of white blood cells in the spinal fluid of patients suspected of having meningitis is critical, as this will help to distinguish between bacterial (presence of neutrophils) and viral (presence of lymphocytes) forms of meningitis.

Additional points to keep in mind when considering findings:

Bacterial infections will not always lead to increases in WBC counts. The lymphocyte count may also be increased in some chronic bacterial infections. Tuberculosis, for example, will show an increase in lymphocytes and monocytes, as well as neutrophils.

A variety of non-infectious inflammatory conditions, other than infection, can cause increases in WBC counts; these can include trauma, severe arthritic conditions and leukemia.

The normal WBC and WBC differential counts, and their ranges, depend on the age of the patient. The normal WBC count is highest at very young ages and decreases during childhood. Local infections, such as abscesses or skin wounds will not necessarily result in changes in the WBC or WBC differential counts as determined by the CBC.

ANOTHER BORING FUNDRAISING PLEA??????

Matching Gift Fund # 43-1709945

Not here!! The Family Camp is coming up this summer and it isn't boring at all. But it is expensive for the Foundation (free to the families) and will cost somewhere around $15,000. The Camp asks for a contribution from SDSF and we need to pay for the doctors to travel and stay there as well. So, we are in need of many financial donations. Ask any family who went two summers ago – it was fantastic!! They can't wait to go back! Please email 4sskids@shwachman-diamond.org if you would like to be put in touch with a family who has attended the camp for more details. We have families that will be happy to talk to you.

Only 7% of our revenue goes to administrative costs. So for every dollar, just $.93 is helping the patients and their families in the SDS community. Most charity consultants will tell you not to donate to a charity that uses $.80 or less for direct services to the patients and their families because that will be an irresponsible charity. We are not only responsible but effective!! We care to do all that we can to help the SDS community.

So shoot for the stars and see what happens when you ask others for financial donations. People love helping their friends, families and co-workers. You will be amazed! And don't forget to mention that we have a **MATCHING GIFT FUND.** Ask your employer if they contribute to matching gift funds and get the paperwork to maximize your donation. Your friends/family can also do this. Our number is **43-1709945.**

Thank you on behalf of all the Board members of SDSF. Please call us at 1-877-737-4685 or email us at 4sskids@shwachman-diamond.org.

FAMILY SHARING PAGE

WILL'S STORY

(with a little help from my friends)

My name is Will Leonberger, and I am 8 years old. I am like any other 8 year old boy, except I was born with Shwachman-Diamond Syndrome which affected my bone marrow. When I was 5 years old the doctors started looking for a donor because they felt I was going into bone marrow failure. My doctor started me on a drug called neupogen (GCSF shots) which worked for a while. Then I had another bone marrow biopsy and it showed that my pre-leukemic cells were quickly multiplying and the doctor said I would need to have a transplant soon. At first we thought I would need a donor from the National Marrow Donor Program (NMDP) because there were no matches in my family. But then God blessed me with a little brother who had the same bone marrow as me so he was my donor. I went to Cincinnati Children's Hospital in April, 2006 for my transplant with Dr. Harris. My brother, Jeb, who was 1 years old, came to the hospital in May. The bone marrow was taken from his hip bones and given to me. On May 11, 2006 ny new healthy life began. I went home from the hospital in late June, 2006.

While I was in the hospital for my transplant, I found out that there was only one other child there (out of 16 BMT patients) that had a sibling donor. This made me sad that there were kids that didn't have a brother (or sister) like my little brother, Jeb, to save their life. My Mom and Dad explained to me that there was something called a National Morrow Donor Program and that strangers unselfishly donate their bone marrow to save the lives of other people. I was really excited about that, until I met a friend who couldn't find a donor on the registry. I told my Mom and Dad that I wanted to help my friend and other people like her.

So for my one-year transplant anniversary and in celebration of my new healthy life, I decided to get as many people to register on the National Marrow Donor registry, so that they could in turn give the gift of life.

Will's Wish Bone Marrow Drive was held on Friday, May 11, 2007 at Middletown United Methodist Church in Louisville, KY. It was part of the National Marrow Donor Program and was sponsored by the James Graham Brown Cancer Center. We had a lot of food and the kids that came got to bounce in inflatables, see clowns, have their faces painted, and get balloons. There were door prizes for the grown ups. Horse jockey, Pat Day, came out to the drive to help us. I had met him one Halloween when I was trick-or-treating dressed as a jockey. When I rang his doorbell he really liked my costume and started talking to me and my parents. He has kept in touch with us ever since. So he came to help us with the bone marrow drive.

We were able to get 160 racially diverse donors in just three hours! Also with the help of my family and friends, we have raised and donated more than $6,000 in funds which will be used to pay for future bone marrow drives.

Some facts about bone marrow donation that you may find interesting:

- Approximately 70% of patients in need of a tranplant do NOT have a family member who is a suitable match.
- Each year, more than 30,000 children and adults in the United States are diagnosed with life-threatening blood diseases. Many have to turn to the NMDP to find a life saving match.

It's easy to donate to the NMDP Registry. It only takes a small sample of cheek cells from the donor. This is done with a sterile cotton swab. Any healthy person between the ages of 18-60 can do it. Minorities are strongly encouraged to join, because a patient's most likely match is with an individual of the same racial or ethnic heritage. Expanding the diversity of potential donors improves the chance a match could be provided for miniority patients.

You can read more about the donor registry at http://www.marrow.org/.

SDS PHYSICIAN/SCIENTIST PROFILE

Dr. Akiko Shimamura, MD, PhD

Dr. Akiko Shimamura is a doctor in the forefront of SDS research and knowledge. Dr. Shimamura completed her residency in Pediatrics at Johns Hopkins Hospital in Baltimore and completed her fellowship in the Hematology/Oncology Program at Children's Hospital Boston. In 2002, she started a research project about SDS while working for Children's Hospital and Dana Farber Cancer Institute. She is also credited with being a key proponent in starting the Bone Marrow Failure Program at Children's Hospital Boston. Her commitment to SDS is shown by her attendance at the Scientific Congress in 2005 in England and then chairing the Scientific Congress in 2007 in Boston. Because of her commitment she was asked and is currently a member on our MSAB. Dr. Shimamura has written and co-written numerous research articles about Shwachman-Diamond Syndrome, Fanconi Anemia and other bone marrow failure syndromes. Her interest and commitment in research for SDS and our children is invaluable.

Her contact information is:

Akiko Shimamura, MD, PhD

Associate Professor of Pediatrics

Division of Hematology/Oncology

University of Washington

815 Mercer St., Rm. 356

Seattle, WA 98109

Tel: 206-685-5282

Fax: 206-616-4082

shima2@u.washington.edu

SPOTLIGHT ON RESEARCH

EXCITING NEWS ABOUT RESEARCH PERTAINING TO SHWACHMAN-DIAMOND SYNDROME !!!

The amount of research published yearly in the last five years which pertains to Shwachman-Diamond Syndrome (SDS) doubled that which was published yearly prior to 2002. The explosion of interest in SDS was set off by the discovery of the SBDS gene and has resulted in the efforts of many scientists who are attempting to understand this syndrome. Researchers around the world have become increasingly interested in understanding SDS and the other bone marrow failure syndromes. During the past three years alone, over twenty new articles addressing various aspects of SDS have been published each year in medical journals. For those of us with loved ones with SDS, this gives us a realistic hope for improved medical management and ultimately for a cure for this disease.

The following research has been published just since July 2007:

1. Alter BP.

Diagnosis, genetics, and management of inherited bone marrow failure syndromes.

Hematology Am Soc Hematol Educ Program. 2007;2007:29-39.

PMID: 18024606 [PubMed - in process]

2. Costa E, Santos R.

Hematologically important mutations: Shwachman-Diamond syndrome.

Blood Cells Mol Dis. 2007 Oct 2; [Epub ahead of print]

PMID: 17916435 [PubMed - as supplied by publisher]

3. Gohring G, Karow A, Steinemann D, Wilkens L, Lichter P, Zeidler C, Niemeyer C, Welte K, Schlegelberger B.

Chromosomal aberrations in congenital bone marrow failure disorders-an early indicator for leukemogenesis?

Ann Hematol. 2007 Oct;86(10):733-9. Epub 2007 Jul 25.

PMID: 17653548 [PubMed - in process]

4. Rawls AS, Gregory AD, Woloszynek JR, Liu F, Link DC.

Lentiviral-mediated RNAi inhibition of Sbds in murine hematopoietic progenitors impairs their hematopoietic potential.

Blood. 2007 Oct 1;110(7):2414-22. Epub 2007 Jul 17.

PMID: 17638857 [PubMed - indexed for MEDLINE]

5. Toiviainen-Salo S, Mayranpaa MK, Durie PR, Richards N, Grynpas M, Ellis L, Ikegawa S, Cole WG, Rommens J, Marttinen E, Savilahti E, Makitie O.

Shwachman-Diamond syndrome is associated with low-turnover osteoporosis.

Bone. 2007 Sep 5; [Epub ahead of print]

PMID: 17920346 [PubMed - as supplied by publisher]

6. Ganapathi KA, Austin KM, Lee CS, Dias A, Malsch MM, Reed R, Shimamura A.

The human Shwachman-Diamond syndrome protein, SBDS, associates with ribosomal RNA.

Blood. 2007 Sep 1;110(5):1458-65. Epub 2007 May 2.

PMID: 17475909 [PubMed - indexed for MEDLINE]

7. Calado RT, Graf SA, Wilkerson KL, Kajigaya S, Ancliff PJ, Dror Y, Chanock SJ, Lansdorp PM, Young NS.

Mutations in the SBDS gene in acquired aplastic anemia.

Blood. 2007 Aug 15;110(4):1141-6. Epub 2007 May 3.

PMID: 17478638 [PubMed - indexed for MEDLINE]

8. Hesling C, Oliveira CC, Castilho BA, Zanchin NI.

The Shwachman-Bodian-Diamond syndrome associated protein interacts with HsNip7 and its down-regulation affects gene expression at the transcriptional and translational levels.

Exp Cell Res. 2007 Jul 10; [Epub ahead of print]

PMID: 17643419 [PubMed - as supplied by publisher]

9. Costa E, Duque F, Oliveira J, Garcia P, Goncalves I, Diogo L, Santos R.

Identification of a novel AluSx-mediated deletion of exon 3 in the SBDS gene in a patient with Shwachman-Diamond syndrome.

Blood Cells Mol Dis. 2007 Jul-Aug;39(1):96-101. Epub 2007 Mar 21.

PMID: 17376717 [PubMed - indexed for MEDLINE]

Abstracts for these articles can be reviewed on PubMed's website at www.pubmed.gov. For any assistance in obtaining research articles, contact SDSF at 4sskids@shwachman-diamond.org.

Ongoing Research Needs 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 uncommo (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 questionnaire,
- 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:

Lynda Ellis, RN at 416-813-5515 or
lynda.ellis@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.

University of Texas Medical Branch-Galveston, Texas

Dr. Tarek Elghetany, Division of Hematopathology at the University of Texas Medical Branch in Galveston, Texas is studying the bone marrow and blood of patients with Shwachman-Diamond Syndrome for early signs of myelodysplastic syndrome and leukemia. If you or your child have a bone marrow study performed, Dr. Elghetany can perform several research studies on the samples. Dr. Elghetany will

also receive some bone marrow samples from Dr. Blanche Alter.

Dr. Alter is the principal investigator for the Etiologic Investigation of Cancer Susceptibility in Inherited Bone Marrow Failure Syndromes (IBMFS) that is taking place at the National Cancer Institute. The specific aims of these studies are to study similarities and differences between SDS bone marrow, other bone marrow failure disorders, and RA bone marrows; to characterize all SDS patients with regard to presence or absence of AA or MDS; to classify SDS patients with MDS and to study MDS features in SDS; to also identify early markers of clonal evolution and to correlate MDS grade or early clonal markers with the development of acute leukemia; and to evaluate different MDS scoring systems regarding their predictive value for survival and development of acute leukemia in SDS patients. Dr. Elghetany will study patients with SDS and follow them up for several 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, e-mail melgheta@utmb.edu. Dr. Elghetany's research is an ongoing study and he is still accepting bone marrow samples.

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 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 SDSF and mail to 710 Brassie Drive, Grand Junction, CO 81506.

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.

Louisville, KY
Auburn, NE
Santa Rosa, CA
Sweetwater, TX
Madisonville, KY
Palisado, CO
Mundelein, IL
Walla Walla, WA
Island of Mauritius
Hertzillia, Isrel
Lapeer, MI

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 16, 2007 - November 30, 2007)

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BRACELET PURCHASES

In Honor of Marissa Avorch
Nicole Carpiniello

In Honor of Kaitlyn Bright
Lori Ess

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: sharwk60@btinternet.com
http://www.shwachman-diamondsupport.org

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

Shwachman-Diamond Syndrome Canada
Contact: Heather Norton
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 opcrccdb@aol.com

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

Kelly Bright -TX: (409)738-2925

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

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

OTHER COUNTRIES

Sharon Clusker - England:
Sharwk60@aol.com

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

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

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

NEWSLETTER IDEAS

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

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

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

Thank you.

CHANGE OF ADDRESS OR E-MAIL

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

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

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

Medical Scientific Advisory Board

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Please send you tax deductible gift to: **Shwachman-Diamond Syndrome Foundation**
710 Brassie Drive, Grand Junction, CO 81506 U.S.A.

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

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The children and adults you are helping THANK YOU for caring.
Your generosity in giving is greatly appreciated.

Shwachman-Diamond Syndrome Foundation is a tax exempt organization as described under the Internal Revenue Code, Section 501 (c)(3). Our Tax ID number is 43-1709945.

710 Brassie Drive
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