



Save Babies Through Screening's

Newborn Screening NEWS

Raising the Awareness of Newborn Screening and Related Disorders

www.savebabies.org

Summer 2002 Volume 2, Issue 3

Study Results Clearly Show Screening for MCAD is Cost Effective

The results of a study examining the cost-effectiveness of newborn screening for Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) were recently reported by the researchers from the Children's Hospital of Philadelphia. The oral presentation, delivered at the 2002 Pediatric Academic Societies' Meeting in May, was entitled "Newborn Screening by Tandem Mass Spectrometry for Medium Chain Acyl-CoA Dehydrogenase Deficiency: Is It Cost Effective?"

The event was held in Baltimore, Maryland and had a general pediatric research agenda, with a wide audience, and was a great forum for presentation of the study results.

Save Babies Through Screening helped to facilitate the research efforts by including information about the study in our Summer 2001 newsletter and encouraging our members to participate in the study.

"Thanks again to you and all the

members for your continued interest and support," said Dr. Chuck Venditti, about the participation of SBTS members in the study. "We had quite a few families mail in surveys and their participation was greatly appreciated and very important in helping verify some of our study variables."



Three-year-old Alex, pictured above, has MCAD. Because he was supplementally screened, he lives a normal, healthy life.

"Our study clearly shows that screening for MCAD is cost-effective. We hope this work will provide a reference that the newborn screening community can use to help institute expanded newborn screening," said Dr. Venditti, who is one of the primary investigators of the study.

The work is being submitted to a pediatric journal for review prior to publication. Dr. Venditti invites any interested party to contact him by email (cvenditt@mail.med.upenn.edu) or phone (1-215-898-2080) if more information is desired.

Second Annual Mother's Day Drive Raises More Than \$12,000

The 2002 annual Mother's Day Fundraising Drive was a great success and is the most successful fundraising event for Save Babies Through Screening to date. The drive raised more than \$12,000, which will be used in newborn screening advocacy, education and research projects.

Save Babies Through Screening relies upon dedicated, caring and concerned parents and friends who work tirelessly to organize critical fundraising events every

year. The events range from massive corporate events to local, family planned outings. If you are interested in helping raise funds to help save babies through screening, contact fundraising@savebabies.org or call 1-888-454-3383.

Many thanks to all who participated in the Mother's Day Drive and helped make it such a tremendous success!

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Dallas and Tera Mize

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HISTORY

The Tyler For Life Foundation was created in 1998 by a family who lost their infant son, Tyler, to a treatable inherited disorder detectable through newborn screening. Had Tyler been screened properly, he would have likely lived a normal, healthy life. In 2001, the Tyler For Life Foundation adopted the name Save Babies Through Screening in an effort to better connect with the general public.

Since its creation, SBTS has strived to provide information on newborn screening to parents and the medical community. The organization is a non-profit organization operated solely by volunteers.

SBTS provides services such as a toll free information line, a quarterly newsletter, a monthly online newsletter, announcement email list, discussion list servs, informative publications, public awareness campaigns, national public service announcements, advocacy materials, NBS unity quilt, and supporting research.

Letters to the Editor

Just wanted to let you know that you guys are doing a fantastic job. I need some info on how to receive 100-200 of the new brochures that you had made. I have printed up a couple hundred of your old blue ones but I just received a copy of the color one and it looks great. I go through them at my dental office quite quickly as patients tell me they are pregnant or being a grandparent or great-grandparent soon. We also leave them in the waiting room for patients to read and if they are interested we also carry the parent packets from NeoGen Screening and give them one. Hope to see some of you at the MSUD Symposium in July

-Seth H., Springfield, NJ
Joshua and Matthew, MSUD

My name is Laurie and I have a 21 month old daughter with PKU. We found out she tested positive at 5 days old. Thanks to the newborn screening, my daughter is very healthy and very bright for her age. It has been a challenge for the past 21 months. I guess like other parents with children with a metabolic disorder, I was in shock. I thank GOD each and everyday that I have her and she is as perfect as a child that does not have PKU or another disorder.

Last Sunday, I was reading our paper and I came across an article titled "Screening program helps save babies" I would like to send you the article. I will briefly tell you what it is saying. In our state (Louisiana) they screen for 4 screenings. We have a hospital that has/will start screening for 50 rare metabolic disorders. I just thought about you and your organization. I would love to send this to you to read via email or regular mail if you are interested. Please just let me know.

I would love to become active in NBS or PKU in the future. If I can ever do anything to help, please let me know.

-Laurie P., Shreveport, LA
Morgan, PKU

Minnesota Now Testing Newborns for Expanded List of Dangerous Metabolic Disorders

Minnesota is one of a handful of states now using new technology to expand the list of potentially dangerous metabolic disorders that can be detected in newborn infants - and these stepped-up testing efforts have already begun to pay off.

Since May of 2001, the public health laboratory at the Minnesota Department of Health (MDH) has been pilot testing the use of tandem mass spectrometry technology to screen

newborns for these potentially life-threatening genetic disorders.

As a result, the lab can now detect up to 20 or more metabolic problems for which screening previously wasn't available. Included are conditions like MCAD deficiency, a potentially life-threatening disorder resulting from the inability to process fatty acids in the child's diet.

So far, the expanded screening effort has identified nine infants with potential problems that wouldn't have been detected previously. All nine were referred to metabolic disease specialists at the Mayo Clinic or the University of Minnesota for diagnosis and treatment.

"Our experience so far has supported what many experts in the field had already suspected," said Dr. Norman Crouch, who heads the state public health lab. "These conditions are a significant public health problem - a 'hidden' public health problem which, until now, often went unrecognized. It's now believed that these conditions may account for a significant number of previously unexplained deaths and illnesses."

Some of these conditions may not cause problems until later in life, Dr. Crouch noted. While they can be life-threatening, these disorders can be successfully managed through diet and appropriate medical monitoring. "That makes early detection critically important," Dr. Crouch said.

Prior to phasing in the new technology, the MDH lab had already been screening newborns for a basic list of

metabolic disorders that included five different types of conditions - phenylketonuria (PKU), congenital hypothyroidism, galactosemia, hemoglobinopathy and congenital adrenal hyperplasia.

Screening for these conditions has been a legal requirement in Minnesota for a number of years.

Roughly 65,000 Minnesota infants are screened every year for congenital metabolic problems. About 140 infants each year are

referred for medical evaluation, and about 50 of those infants are subsequently diagnosed with metabolic conditions.

The 20 or so conditions that can be detected using the new technology represent an addition to the basic list of five. The same blood samples used for the required tests are also being used to run the new screening procedures.

Conditions that can be detected using tandem mass spectrometry generally fall into three broad categories - fatty acid oxidation disorders (a group that includes MCAD), amino acid metabolism disorders and organic acid metabolism disorders.

It's also possible that additional conditions may be added to the initial list of 20, according to analytical chemist Mark McCann, who is the technical expert in charge of the expanded screening program.

"We're still in the pilot phase in terms of using this technology," McCann said. "Tandem mass spectrometry allows us to pick up, in a blood sample, many different chemical compounds that might be a marker for some type of metabolic problem. Based on the work that's been done so far, here at MDH and elsewhere, we believe that we have identified around 20 to 24 conditions that we can detect using this procedure. But that list may not be exhaustive."

For more information, contact Buddy Ferguson, MDH Communications, at (651) 215-1306 or Louise Liao, MDH Environmental Laboratory, at (612) 676-5706.

-Minnesota Dept. of Health Press Release, 2/18/02

"These conditions are a significant public health problem - a 'hidden' public health problem which, until now, often went unrecognized."

Joint Metabolic Conference to be Held in Orlando

The National Coalition for PKU and Allied Disorders will hold a conference October 3-5, 2002, in Orlando, Florida. The conference coincides with the World Congress & Exposition on Disabilities (WCD), which will be held at the Orange County Convention Center.

Cost for the NCPAD conference will be \$50.00 per person and will include lunch on Thursday, October 3. Cost also includes access to all WCD sessions and exhibit hall. In addition, there will be daycare available for children whose families plan to attend the conference. The NCPAD conference will feature disorder specific educational sessions for Phenylketonuria (PKU), organic acid disorders, Tyrosinemia, fatty-acid oxidation disorders, Homocystinuria, and Maple Syrup Urine Disease (MSUD).

This year's WCD has more than

160 scheduled breakout sessions, including several sessions on newborn screening. Topics for the newborn screening related sessions include What Is UNBS and Why Is It So Important? (Piero Rinaldo, Mayo Clinic), Detection, Screening & Treatment (Piero Rinaldo, Mayo Clinic), Screening Methodologies (Donald Chace, NeoGen Screening), Why Screening is Important to Our Nation (Charles Hehmeyer, attorney), The Role of Genetic Counseling (Mary Davidson, Genetic Alliance), and Genetic Disorders and Counseling (Paul Fernhoff, Emory University).

More information about the National Coalition conference can be found at www.pku-allieddisorders.org or by calling 1-877-996-2723. Further information about the WCD can be found at www.wcdepo.com.

Co-Developer of Screening Technology Recognized for Outstanding Achievement

Donald Chace, Ph.D. M.S.F.S. was the recipient of The Sigi Ziering Award for Outstanding Contribution for a Publication in the Journal Clinical Chemistry.

The American Association for Clinical Chemistry (AACC) presents several awards each year to clinical chemists and other clinical scientists who work in the field of clinical laboratory science. These awards are presented to recognize outstanding achievements, to increase awareness of contributions made to the development of the science and improved health care, and to stimulate other scientists to accelerate their efforts.

Dr. Chace is one of the primary investigators that developed the newborn screening application of tandem mass spectrometry. Dr. Chace previously served as the assistant medical research professor in the department of Pediatrics at Duke University Medical Center. He



Donald Chace, Ph.D., M.S.F.S.

currently is the Section Chief of the Division of BioAnalytical Chemistry and Mass Spectrometry at Neo Gen Screening in Pittsburgh, Pennsylvania. In addition,

Dr. Chace is currently working with the Center of Disease Control to develop quality assurance systems for newborn screening of inherited metabolic diseases as well as new mass spectrometry methods to detect additional diseases to improve the quality of life for newborns and their families.

The American Association for Clinical Chemistry is an international scientific/medical society of clinical laboratory professionals, physicians, research scientists and other individuals involved with clinical chemistry and other clinical laboratory science-related disciplines. The AACC was founded in 1948 and has 11,000 members at the present time. More information about the AACC can be found at www.aacc.org.

Senators Urge Review Of State Newborn Screening Programs

Seeking to improve critical detection of potentially life-threatening genetic disorders in infants, Senators Chris Dodd (D-Connecticut) and Mike DeWine (R-Ohio) called for a study of each state's newborn screening programs in a recent letter to the General Accounting Office (GAO). The letter from Dodd and DeWine, requested the GAO to report on individual state's efforts; how they test for disorders; the coordination among families, physicians, and laboratories involved; and the procedures and quality of data. The letter also asks for a report on how states protect the privacy of this sensitive and critical health information.

"A child's health shouldn't be influenced by a line drawn on a map," said Dodd. "Newborn screening is a valuable tool in efforts to treat children's health problems, and - as such - we need to better explore how it is being implemented nationwide. This is one test where failing shouldn't be an option."

"Newborn screening programs have unlimited potential. This is an excellent example of preventative medicine at its best and families in all states should be able to reap the benefits," said DeWine. "Clearly, detecting and treating disorders early in life produces tremendous benefits--by both decreasing the cost of care and increasing the quality of a child's life."

About 3,000 infants screened each year are identified with having health conditions that could be harmful in the future. Some of the disorders can be treated simply by a change of diet or increase in certain vitamins, as long as the condition is caught at an early stage.

Currently, each state develops and administers their own plan to check for disorders and the programs can vary greatly between states. At this time, there is no comprehensive data on the initiatives of each state.

-Senator Chris Dodd Press Release, 2/6/02

State Program Updates

GEORGIA

Georgia has begun a four year partnership with the Georgia Chapter of the March of Dimes to make tandem mass spectrometry newborn screening available to all babies born in Georgia. The Georgia MOD will be providing funds to purchase the additional equipment needed to implement the testing and to help provide parent and professional education. However all plans are at a standstill until the state can provide funding for the personnel to run the equipment. The first MS/MS machine arrived in May 2002 and is being placed at Emory University for diagnostic testing. Georgia currently screens for 8 disorders: Congenital Adrenal Hyperplasia, Galactosemia, Homocystinuria, Hypothyroidism, Maple Syrup Urine Disease, Phenylketonuria, Hemoglobinopathies such as Sickle Cell Disease, and Tyrosinemia (Type 1 & 2).

MISSOURI

Although Missouri's recently-passed House Bill 279 mandates that the Department of Health expand the newborn screening requirements, the new law includes the phrase "subject to appropriations." And, as in many other states, Missouri is in a budget crisis and funding has not been made available to expand the screening program even though the law has been passed. Congenital Adrenal Hyperplasia (CAH) is the only disorder among the expanded list appearing in the bill to receive funding so far. Currently Missouri tests for 5 disorders: Congenital Adrenal Hyperplasia, Galactosemia, Hypothyroidism, Phenylketonuria, and Sickle Cell Disease.

CALIFORNIA

In January 2002, the state of California started using tandem mass spectrometry (MS/MS) screening in a MS/MS Evaluation Project to screen babies for more than 25 additional disorders. Previously, California only screened for four disorders in its newborn screening test. Parents of babies born in California are offered the option of having the expanded testing. There is no additional cost to receive the new expanded testing, however, only birthing facilities that have enrolled in the new program are offering the added testing. Biotinidase deficiency and congenital adrenal hyperplasia are still not currently screened for in California even with the new expansion.

Visit www.savebabies.org/states.htm for information about other state programs.

SBTS Salutes Heroes That Work To Save Babies

Some people wonder if they can really make a difference in the world, but we know it's possible because we've seen the difference made in the lives of screened children by the diligent volunteers who have taken up the cause. The volunteers of Save Babies Through Screening don't just talk about making the world a better place, they do something about it by reaching out and giving their time. They are wonderful examples of what caring people should be, and in a world that sometimes seems a bit cold and impersonal, their warmth and compassion really shine through. Every volunteer is a very special person, and we would like to take this time to say thank you to all the people who give of themselves to help children affected by diseases detectable through newborn screening.

2002 Save Babies Through Screening Volunteers:

Aalberts, Joan	Hendricks, Judy	Ruff, Melanie
Abbott, Jessica	Hoek, Cindy	Ruter, Paula
Arjona, Michelle	Honey, Wendy	Speelman, Betty
Arnold, Corrina	Hubbard, Therica	Spraggins, Lisa
Bedney, Anne	Jaquez, Lisa & Gonzo	Stephens, Mikki
Blenet, Patricia	Kelly, Sue	Sweeney, Pam
Boiko, Michelle	Kolenkiewicz, Linda	Szabo, Dr. Alexandra
Bradford, Jacque	LaGoy, Jeanette	Taylor, Paul
Brown, Heather	Lazzaro, Jamie	Thomas, Margaret
Bulcher, Sandy	Lazzaro, Jim	Thompson, Holly
Burns, Michelle	Leach, Anita	Thurston, Janet
Campbell, Melissa	Leight, Kelly	Vandam, Wendy
Carmichael, Stacy	Leslie-Brown, Sharia	Vengrenyuk, Yuliya
Casas, Randie	Liner, Gabe	Vikstrom, Karen
Chong, Monica	Magol, Marsha	Wallace, Angus
Clay, Janine	Maguire, Michelle	Wallace, Maria
Clow, Laura	Masterson, Louis	Walton, Jennifer
Cook, Trina	Mathews, Shantel	Watkins, Michelle
Cooper, Sandy	Merkel, Sarah	White, Darci
Cornette, Teresa	Merrill, Sheri	Williams, Christi
Daniel, Lakeisha	Miller, Michelle	Wilson, Jane
Defoe, Stephanie	Millett, Susan	Wilson, Tiffany
DePaolo, Terilyn	Mize, Cliff	Wilson-Hancock, Diana
Dougherty, Dawn	Mize, Dallas	Wood, Jill
Dove, Kandi	Mize, Tera	Workman, Shea
Dunahoo, Marcia	Monaco, Tom	Young, Tiffany
Eskew-Sneddon, Julia J.	Moore, Patty	
Farmer, Laurie	Mueller, Ann	
Field, Attny. Tracy	Murry, Theresa	
Finn, Carolyn	Nawn, Wendy L.	
Finn, Terence	Olstad, Carlene	
Fisher, Sharon	Olstad, Todd	
Foster, Kelly	O'Reilly, Susan	
Franks, Jill	Ottinger, Debbie	
Gill, Indermeet S.	Parker, Richard	
Grant, Wanda B.	Pennington, Kimberly	
Greichunos, Julie	Pietryga, M. Paula	
Hall, Kileen	Pycior, Rhonda	
Hamburg, Deborah	Raccosta, MaryAnn	
Harnos, Liz	Revinski, Agnes	
Harris, Peggy	Revinski, Gina	
Haris, Tiana	Riddell, Judy	
Haygood, Robin	Roberson, Sonja	
Heberer, Jennifer	Royal, Dena	
Heinze, Monica	Ruff, Dell	

Call Your CBS TV Station to Run PSA

Don't forgot to call your local CBS TV station and ask them to run the public service announcement done by Guiding Light actor Michael O'Leary. Here's what to do:

- Ask for the Community Relations Director.
- Ask them to air the tape around the Guiding Light's air time as frequently as possible.
- Contact SBTS at 1-888-454-3383 to tell us where to send the tape.

Celebrating Children's Lives Through Quilt Panels

Quilt panels for the Newborn Screening Unity Quilt are still being collected. The quilt is a visual testimony of the lives touched by newborn screening and gives a lasting face to many disorders that are not routinely screened for in state-mandated screening programs.

Families with children who have detectable disorders are encouraged to submit panels to celebrate the lives of their children. Participation is open to anyone who has a disorder currently detectable through newborn screening. For more information about the NBS Unity Quilt project, please email quilt@savebabies.org or call 1-888-454-3383. More information

is also available at www.savebabies.org/ quilt.

The quilt panels were displayed last September and October by the hospitals of Virtua Health System in New Jersey. The panel of Ben Haygood (MCAD) was also showcased at the National Society of Genetic Counselors education conference in Washington, D. C., and the American Academy of Pediatrics conference in San Francisco, California. The quilt is scheduled to be displayed again this September in celebration of Newborn Screening Awareness Month. A full display schedule will be released at a later date.

School Project Teaches Many About NBS

The Tupelo, Mississippi Public School District students embraced the spirit of giving as they worked to raise awareness about and funds for charity during the fifth annual Students Helping Others Planet-wide (SHOP) event. SHOP is intended to teach children how to help others.

The event took place November 12-16, 2001, at Gloster Creek Village shopping center. The students set up more than 25 booths representing charities from St. Jude Children's Hospital to the local library to the Tyler For Life Foundation (now know as Save Babies Through Screening).

More than 900 children from grades 2-8 participated in the SHOP event. Students began preparing for the event early in the school year by choosing an organization to support and researching it.

Several students chose Tyler For Life Foundation as their charity. The children built a booth that represented the Tyler For Life Foundation in which they accepted donations, handed out educational materials on newborn screening and talked with visitors to the event about NBS. The students raised almost \$1,000 in donations for NBS.

Newborn screening advocates Robin Haygood and Sharia Leslie-Brown were instrumental in bringing TFL/Save Babies Through Screening to the attention of the SHOP school children for consideration in the event. Robin Haygood's son, who is also Sharia's grandson, died from complications of undiagnosed MCAD. Robin and Sharia both formerly worked for the Tupelo Public School District. Robin is also a SBTS board member.

The students in Jenny Decker's class who put together the exhibit were Joseph Koon, Rob Ellis, Elizabeth Anderson, Mary Byars, Heaven Adams, Brittany Vaughn, Robert Stroup, Tyler Rardin, Mandy Collins and Tyler Berry.

Louisiana Hospitals Begin Supplemental Screening

The Willis-Knighton Health System of Shreveport, Louisiana, recently started supplemental screening on all babies born at their facilities. Willis-Knighton's supplemental screening program extends the routine screening of 4 conditions to more than 55.

The cost of the supplemental screening is included in the hospitals' routine neonatal charges, and the same blood sample is used for both the required and supplemental newborn screenings.

Since implementation of the program, one suspected case of Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) has already been detected.

Willis-Knighton Health System boasts that the supplemental newborn screening program there is "another example of Willis-Knighton's commitment to not simply treat problems, but to prevent them."

Willis-Knighton uses the nation's largest independent screening laboratory, NeoGen Screening, for the additional testing. Since its founding in 1994, Neo Gen Screening has analyzed blood samples from more than 1,300,000 newborns. NeoGen Screening reports five Louisiana birthing hospitals now offer supplemental screening through their laboratory.

If you or someone you know is expecting a baby, please contact your local birthing hospital to see if they are offering supplemental newborn screening to their patients. For more information about obtaining supplemental screening and advocating that your local hospitals offer the services, visit www.savebabies.org or call 1-888-454-3383.

Educational Handouts Available for Expectant Families

Newly revised newborn screening pamphlets are available for distribution. The handouts, entitled "A Parent's Guide to Newborn Screening", feature the eye-catching headline "A Simple Test Could Save Your Baby's Life".

The handouts supply important information about newborn screening and are written with families in mind. Specifically, the handouts are intended to be given to expectant parents during the third-trimester of pregnancy, as this is considered the most opportune time to educate expectant parents about newborn screening.

The handout can be downloaded in English or Spanish from www.savebabies.org for immediate access. Hard copies are currently available in English only and can also be ordered from the website or by calling 1-888-454-3383. Cost for hard copies is \$10 per pack of 100.

Visit us online @
savebabies.org

Putting Faces Behind the Statistics

The Save Babies Through Screening website features many personal stories from affected families. The purpose of this project is to bring people into the lives affected by detectable disorders, as these families are much more than just another number on a public health agency report. They are much more than just statistics.

By sharing their stories, affected families are associating personal lives with detectable diseases so that decisions in screening can be made with only one thing in mind - the well-being of affected children. In essence, they are putting a face on newborn screening.

Families are always needed to share their story for this important segment. The stories are told through numerous avenues such as the savebabies.org website, quarterly newsletter, public service announcements and legislative testimonies.

If you would like to share your story to help promote newborn screening, please e-mail stories@savebabies.org or call 1-888-454-3383 for more information.

Thank you to all the families who have already shared their stories. Save Babies Through Screening thanks you, as do all the affected babies born in the United States every day.

Germany May Screen for 20 Disorders

Germany could be the first country in the world to adopt universal tandem mass spectrometry newborn screening, after more than 170 children were saved from serious disability or death during a three-year trial program there.

Upcoming Media

'Parents' magazine is scheduled to include an article about parent activists that will include SBTS President and Co-Founder, Tera Mize. Although the article is still in its preliminary stages, it is reported the article may be included in the September 2002 issue.

Upcoming Events

JULY 2002

July 22-24, 2002

National Healthy Mothers Healthy Babies Coalition - Biennial Partnership Conference

More Info: (813)974-6682

Web: www.hmhb.org

Clearwater Beach, Florida

AUGUST 2002

August 15-16, 2002

National Institute of Health (NIH) -

Secretary's Advisory Committee on

Genetic Testing (SACGT) Meeting

Web: <http://www4.od.nih.gov/oba/sacgt/sacgtmtg.htm>

Bethesda, Maryland

SEPTEMBER 2002

September 1-30, 2002

Newborn Screening Awareness Month

Sickle Cell Awareness Month

September 17-22, 2002

Sickle Cell Association of America -

Annual Meeting

Meeting will be held jointly with the NIH.

Web: www.sicklecelldisease.org

Washington, D.C.

OCTOBER 2002

October 3-6, 2002

National Coalition for PKU & Allied

Disorders - Metabolic Conference

Web: www.pku-allieddisorders.org

Orlando, Florida

October 4-6, 2002

World Congress and Exposition on

Disabilities - Annual Conference

Web: www.wcdexpo.com

Orlando, Florida

NOVEMBER 2002

November 9-13, 2002

American Public Health Association -

130th Annual Meeting & Exposition

Contact: Lynn Schoen

More Info: (202) 777-2479

Web: www.apha.org

Philadelphia, Pennsylvania

November 21-22, 2002

National Institute of Health (NIH) -

Secretary's Advisory Committee on

Genetic Testing (SACGT) Meeting

Bethesda, Maryland

Every September is NBS Awareness Month

It is possible to reduce the deaths and disabilities that occur from undetected metabolic disorders, but we need your help to let everyone know. Every September, Save Babies Through Screening celebrates Newborn Screening Awareness Month. This September help increase public awareness about newborn screening by wearing a NBS lapel pin and by having the media broadcast educational messages about newborn screening.

NBS supporters are encouraged to wear NBS lapel pins every day in September to help raise the public awareness of newborn screening. The pins have become a symbol of newborn screening and more than 30,000 people have worn the pins over the last two years. Pins are available at a cost of \$3 each and can be ordered by filling out the order form to the right.

In addition, supporters are requested to contact the news media to have

them present stories about newborn screening and air NBS public service announcements. Below is a pre-written radio announcement. Please have announcers read it as much as possible.

Radio PSA Script - 30 seconds

A simple \$25.00 blood test could save your baby's life. Many doctors don't know about this test...maybe your doctor is one of them...maybe your child is at risk...many of these disorders are silent killers...death occurs with little or no warning. However, children with these conditions can live normal, healthy lives if they are detected and started on treatment early. To learn how to get the extra screening that your baby needs, call 1-888-4LIFE-83 or visit savebabies.org.

More PSA's are available from www.savebabies.org/psa.htm.

March of Dimes Adds MCAD to Recommended List of Disorders To Screen For

Disparities among states in health screening tests given at birth mean too many babies with serious birth defects are not being diagnosed and treated in time, the March of Dimes says.

"Parents are often unaware that, while nearly all babies born in the United States undergo newborn screening tests for genetic birth defects, the number and quality of these tests vary from state to state," says Dr. Jennifer L. Howse, President of the March of Dimes, noting that January is national Birth Defects Prevention Month.

"These tests, done immediately after birth, may mean the difference between a healthy life or a severe disability for a baby," Dr. Howse says. "Tragedy can sometimes be avoided by quickly identifying a problem and providing the appropriate medical treatment."

José F. Cordero, M.D., M.P.H., Assistant Surgeon General and Director of the National Center on Birth Defects and Developmental Disabilities, notes, "We need to develop a national strategy for ensuring safe and effective newborn screening tests for all infants, regardless of where they are born." Approximately four million infants are born annually in the United States, and of these, an estimated 150,000 are born with serious birth defects. Dr. Howse also says the March of Dimes has increased its core list of recommended newborn screening tests for metabolic disorders to nine, to include the condition known as medium-chain acyl-CoA

dehydrogenase (MCAD) deficiency.

MCAD deficiency affects about 1 in every 15,000 infants born in the U.S., the March of Dimes says, and can lead to mental retardation or death if not identified shortly after birth. It is an inherited disorder of fatty-acid metabolism caused by the lack of an enzyme required to convert fat to energy. Seemingly well infants or children can suddenly develop seizures, respiratory failure, cardiac arrest, coma, and death. Identifying affected children before they become ill is vital to preventing a crisis and averting these consequences. Treatment includes steady food or glucose intake and avoidance of fasting.

Accurate testing for MCAD deficiency requires the use of a relatively new scientific instrument called a tandem mass spectrometer. This instrument electronically "weighs" the compounds found in blood samples and can precisely identify unusual levels of certain molecules not detectable by other means.

"The March of Dimes has been reviewing the use of tandem mass spectrometry in laboratories nationwide. We're now convinced that this technology has reached the point where we can recommend it for MCAD deficiency testing for every baby," says Nancy Green, M.D., Associate Medical Director of the March of Dimes.

"The consequences of MCAD deficiency, including death or permanent neurological disability, are completely

preventable by newborn screening and follow-up to educate the family about how to prevent the fasting that occurs during childhood illnesses. I am extremely pleased that the March of Dimes has included MCAD deficiency among the core group of disorders for which every baby in the United States should be screened," says Edward R.B. McCabe, M.D., Ph.D., Physician-in-Chief, Mattel Children's Hospital at the University of California at Los Angeles.

The other metabolic disorders on the March of Dimes-recommended list are: phenylketonuria (PKU); congenital hypothyroidism; congenital adrenal hyperplasia (CAH); biotinidase deficiency; maple syrup urine disease; galactosemia; homocystinuria; and sickle cell anemia.

The March of Dimes also advises a hearing test for all newborns but remains concerned about the current level of technology and intervention. Significant hearing impairment is one of the most common developmental abnormalities present at birth. Undetected, the condition will impede speech, language, and cognitive development.

For more information, visit the March of Dimes Web site at www.marchofdimes.com, its Spanish Web site at www.nacersano.org, or call 1-888-MODIMES.

-March of Dimes Press Release, 1/1/02

Order NBS Lapel Pins

Name: _____

Address: _____

City, State, Zip Code: _____

Phone Number: _____

Quantity: _____ x \$3.00 each = \$ _____

Please mail this form along with check or money order for total amount due to:

Save Babies Through Screening
5335 N. Helton Road
Winston, GA 30187



Treatability Of Disorders Should Not Be Criterion For Inclusion Of Screened Disorders

Tandem mass spectrometry (MS/MS) has catapulted newborn screening into a new era of "screening ethics". In previous years, disorders were added to newborn screening panels when they met certain criteria. This criteria was noted in an August 2000 report by the American Academy of Pediatrics' Task Force on Newborn Screening (NBS) to be:

- a) the condition is an important health problem that occurs frequently enough to justify screening an entire population
- b) the treatment for the condition is effective when initiated early, accepted among health care professionals, and available to all screened newborns
- c) the test is simple, safe, precise, validated and acceptable

The new ethical issue seen by some is that screening using MS/MS technology allows for identification of some disorders that may not necessarily be "treatable" in conventional terms of the word. There is currently national controversy over this issue. Some individuals feel one should not screen for a disorder that doesn't consistently respond to treatment. However, Save Babies Through Screening recommends that 55 disorders be screened on all babies born in the United States.

"These children will have these disorders whether screening is in place or not," says Save Babies Through Screening President Tera Mize. "Families deserve to know what is truly wrong with their children, and their physicians deserve an opportunity to try to help them."

Knowing that a child has a condition that may not respond consistently to treatment is important to families in a number of ways.

Families may choose to avoid having more children after having a child diagnosed with an untreatable inherited disorder. Without screening for these rare metabolic disorders, however, many infants die of unexplained causes, or are wrongly diagnosed with non-recurrent conditions. Parents are then advised that what was wrong with their child won't happen to future children they may have. With screening, parents are aware of the possibility of having more children with the disorder and can make educated family planning decisions.

Early identification also helps families avoid emotional torment and financial burden, which can stem from

years of going from doctor to doctor trying to find out what is wrong with their child. Without screening, some affected children have incurred years of suffering from invasive procedures and families have incurred millions of dollars in costs for unnecessary medical tests to determine what is wrong with the child. Even if the disease doesn't have known treatment options, most parents are extremely gratified to know a diagnosis and spend their time searching for treatment for their child instead of just searching for a diagnosis.

Furthermore, additional knowledge of cases of these diseases will help physicians develop better treatments. "Until we start identifying affected children through screening and giving the doctors an opportunity to intervene early, we won't have treatments," says Wendy Nawn, Vice President of Save Babies Through Screening. "In addition, gene defects are quite variable, or can be expressed in different patterns, thus the result is that in any given disorder there is a range of mild to very serious symptoms. Therefore, it can't be said with any certainty that a disorder is 100% untreatable. And I have never met a parent that would just let a child die if there was any chance he or she could live a normal, healthy life."

"In previous years, conditions such as cancer were thought to be untreatable, yet families were still afforded the courtesy of an accurate diagnosis," says Tera Mize, whose infant son died from a disorder detectable through newborn screening. "People with cancer survive the majority of the time because physicians were allowed the opportunity to learn to treat it. It is time we learned more about 'untreatable' disorders detectable through newborn screening and use this information to devise effective treatment plans. Families believe that these children deserve the opportunity to live normal, healthy lives as do children with diseases such as cancer."

SBTS recommends that all expectant families obtain supplemental screening for the disorders that are not routinely screened for in their state. Supplemental comprehensive screening can be obtained nationally from Neo Gen Screening (1-866-463-6436; www.neogenscreening.com). Screening for disorders detectable through tandem mass spectrometry only can be obtained from Baylor Medical Center (1-800-422-9567; www.baylorhealth.com/newbornscreening) or Mayo Medical Laboratories (1-800-533-1710; www.mayo.edu/mml).

Tyler For Life Foundation dba

Save Babies Through Screening

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5335 N. Helton Road Winston, GA 30187

1-888-454-3383 Fax: 770-947-8507

www.savebabies.org

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