



**Save Babies Through Screening Foundation, Inc.**

# **Newborn Screening NEWS**

Raising the Awareness of Newborn Screening and Related Disorders

[www.savebabies.org](http://www.savebabies.org)

Fall 2005 Volume 5, Issue 2

## **New Foundation Launched in Canada**

After many months of preparation the Save Babies Through Screening Foundation of Canada was launched officially in September. Its founder, Tammy Clark, lost her 9-month old daughter, Jenna, to undetected MCAD Deficiency in November, 2002. Tammy and her husband Roger were appalled and angered to learn that if this disorder had been detected in their daughter's newborn screening, she would most likely be alive and well today.

The Save Babies Through Screening Foundation of Canada hopes to put newborn screening on an equal "footing" for all infants born in any province. While independent of the Save Babies Through Screening Foundation in the United States, the new Canadian non-profit organization operates similarly through volunteers. Their focus is to raise awareness of the need for comprehensive newborn screening for all infants born in Canada.

Despite a universal healthcare system, newborn screening in Canada is similar to that in the United States in that geography plays a deciding role as to which disorders are screened. For instance, Ontario—the first province to begin newborn screening forty years ago—screens infants for only two conditions, whereas Saskatchewan screens for 29. Jenna Clark was born in Ontario.

According to recent media attention in Ontario, approximately 160 additional cases of genetic disorders would be detected yearly if the province were to screen for the disorders recommended in the U.S. by the American College of Medical Genetics. In August Ontario's Health Minister, George Smitherman, indicated that within two months the government would have a plan to improve newborn screening. At press time Ontario was planning to add nineteen disorders to its panel.

## **Children's Stories to Educate Physicians**

SimulConsult, a medical diagnostic software provider, has quoted several stories from [www.savebabies.org](http://www.savebabies.org) on its website [www.simulconsult.com](http://www.simulconsult.com). The purpose for using these stories is to educate physicians and other medical professionals about diseases with which they may not be familiar and to help them reach a timely diagnosis regarding diseases that might be considered rare.

The stories quoted by SimulConsult are those of Tyler Mize (GALT), Stephen Monaco (IVA), Michelle Murry (MCADD), and Michael Sandoval (GA1). SimulConsult also links from these

quoted stories to the Save Babies Through Screening Foundation website.

Also, to aid diagnosis, SimulConsult has added a new resources page to its website titled "The Differential Diagnosis of Child Abuse" by Michael Segal, MD, PhD, and Chief Medical Officer of SimulConsult. Dr. Segal became concerned about false accusations of child abuse in cases involving certain metabolic disorders after reading about such cases on [www.savebabies.org](http://www.savebabies.org). Investigating further, he collected information about

these and other medical conditions that can be confused with child abuse. Quoted stories on this page include those of Sean Calisto (TFP) and Caden Richards (GA2) from [www.savebabies.org](http://www.savebabies.org).

Dr. Segal hopes that this new resource page [www.simulconsult.com/resources/abuse.html](http://www.simulconsult.com/resources/abuse.html) will provide additional considerations for physicians and abuse teams in hospitals while preventing heartache to parents who might be subject to false accusations of causing their child's condition.

## Alexis' Story

by Kris Smith

**Alexis Nicole Knapton** was an active, seemingly healthy 2nd grader who had just turned 8 years old when the silent and deadly killer, undiagnosed MCAD Deficiency, took her life without warning. Her "Birthday Girl" ribbon still hung on her bedroom door as flowers and condolences began arriving at the house that echoed her laughter.

Alexis had stayed home from school on Thursday, March 6, 2003, with a stomach virus. She couldn't eat much that day and preferred to sleep. Nobody knew her body could not convert stored fat into energy, and very early the next morning she was found unconscious and unresponsive in her bed.

She was rushed by ambulance to the PICU at Children's Mercy Hospital in Kansas City, Missouri. Her condition worsened. Her brain was swelling and collecting

fluids, her heart raced, and medication to slow her heart rate caused her blood pressure to plummet dangerously. Her kidneys and liver were failing. She was having seizures, and the doctors couldn't stabilize her. All her vital organs were shutting down.

The doctors had no answers as we waited and prayed that she would open her eyes and smile that contagious smile. By mid-day Sunday she was declared 100% brain dead, and that afternoon she died. Many weeks later we learned that undiagnosed MCADD had taken her life.

"Undiagnosed what?" we asked.

Like most people, we'd never heard of MCADD or supplemental screening. We naively thought Alexis had been screened at birth for anything that could hurt her. Searching for more information, we found it scarce and hard to understand.

Finally, we located stories of other families with similar experiences on [www.savebabies.org](http://www.savebabies.org).

Kelly Peters, Alexis' mother, is now involved with the Save Babies Through Screening Foundation to help increase education and awareness of supplemental screening. Being Alexis' maternal grandmother is one of my greatest blessings. Shortly after she died I started a memorial website with the help of Leon Smith, my brother-in-law, a very talented webmaster. On [www.mcadangel.com](http://www.mcadangel.com) people can find solace, support, and information regarding MCADD.

For such a little girl, Alexis Nicole Knapton left huge footprints in the hearts of everyone who knew her. Because she died, we share what we've learned about MCADD in the hope that other children like Alexis, with undiagnosed MCADD, will never have to endure what she did.

### Is your info current with us?

Please notify us when any change occurs in your address, email address, or telephone number so that we can stay in touch with you. The volunteers who maintain our records will be happy to receive your new contact info. Call toll free, 1-888-454-3383; email to [email@savebabies.org](mailto:email@savebabies.org); or fax 1-610-647-5757. Regular mail can be sent to Save Babies Through Screening Foundation, 4 Manor View Circle, Malvern, PA 19355. Please allow time for our multi-tasking volunteers to update your contact info. Thanks!

### State NBS Program Notes

*[Editor's note: There is no uniform counting system to number the disorders for which states are screening. For consistency, Save Babies Through Screening Foundation uses the figures below to show a balanced comparison, using traditional counting practices.]*

**California** – screening for 41 disorders since Aug. 1, 2005

**Iowa** – now screening for Cystic Fibrosis

**Kentucky** – conducting a roll-out of additional disorders between July 1 and Dec. 31, 2005, for a total of 28 expected to be fully implemented by Jan. 1, 2006

**Louisiana** – piloting for ASA Lysase Deficiency, Citrullinemia, and MCADD

**Maine** – offering supplemental screening for 17 disorders

**Missouri** – screening for 25 disorders since July 1, 2005

**New York** – screening for 43 disorders since spring, 2005

**North Dakota** – mandating screening for 7 disorders and requiring reporting for an additional 29

## Hunter's Legacy

Eight-year-old Hunter Kelly, son of Jim and Jill Kelly who co-founded Hunter's Hope in their son's name, passed away on August 5, 2005. Diagnosed with Krabbe Disease at 4 months of age, Hunter defied his doctor's expectation that he likely would have only a few more months to live. Instead, Hunter lived to wear his famous quarterback father's No. 12 jersey and to share in some of the public recognition that his Hall of Fame dad attracted.

Hunter's Hope Foundation, established in 1997, has raised millions of dollars for research and awareness of the disease that eventually took Hunter's life. In 2004 the foundation created the Hunter James Kelly Research Institute at the University of Buffalo.



This brave little boy, who suffered greatly throughout his short time on earth, left a legacy of hope—hope that children yet to be born with Krabbe Disease and other leukodystrophies will benefit from early detection, effective treatment, and, possibly, one day, a cure.

*Ed. note: Visit [www.huntershope.org](http://www.huntershope.org) for more information.*

## Applause!

- For **Laura Clow** and **Corrina Arnold** who ran a newborn screening booth at the Richardson Regional Medical Center Great Expectations Baby Fair in Dallas, Texas, on February 5, 2005.
- For **Judy Dow** who dispensed information about newborn screening at the Hospital Jamboree in the Milwaukee, Wisconsin, area in March of this year.
- For **Mary-Ella Frederich** who provided newborn screening pamphlets and information to parents and expectant parents at the 7<sup>th</sup> Annual Family Café Conference, Caribe Royale, Orlando, Florida, on June 3-5, 2005.

## Spreading the Word

*by Kris Smith*

On June 8, 2005, Kelly Peters addressed an audience of approximately 80 pediatric healthcare providers at the "Common Problems in Pediatrics" seminar at Westminster University in Salt Lake City, Utah. Her topic, *The Importance of Supplemental Screening*, covered many aspects of newborn screening and reflected her personal experience with undetected MCAD Deficiency that claimed the life of her 8 year old daughter, Alexis. [See the story of Alexis in this issue.]

Stressing that early detection of deadly disorders is key to saving lives, Kelly

described the value of supplemental newborn screening, where to obtain the screening kits, and how the specimens should be handled. She spoke about the need to develop and fund a plan to provide overnight courier service to eliminate delays in transporting specimens to the lab and to lessen the occurrence of damage to specimens from improper handling.

Kelly quoted statistics on the incidence of some diseases that can be detected through supplemental screening, challenging the notion of how "rare" they are

and noting that health care providers and parents have a responsibility to learn more about metabolic disorders. As a member of the Board of Directors and volunteer with the Save Babies Through Screening Foundation, she described the activities of the foundation to educate and increase awareness of newborn screening. Kelly knows firsthand that newborn screening is not provided equally in all states, and she welcomes the advances that are underway in many states to improve their programs.

## Avon for Babies

Lori Michaud hosted an Avon fundraiser in recognition of NBS Awareness Month in September, during which time she donated half her profits to the Save Babies Through Screening Foundation. Lori is the mother of Jordan who died from an unclassified FAOD.

She has extended the fundraiser and will continue to donate a portion of her profits through December. To help Lori save babies, go to [www.savebabies.org](http://www.savebabies.org) and click on "Fundraisers" in the menu on the left side of the screen.

## With Gratitude and Recognition

Donations of money, time, and talent are the mainstay of the foundation's programs. Thank you for all that you do on behalf of babies. Noted here are the special contributions *received between January and June of 2005.*

### Fundraising with Awareness

Rick Schraitle  
*Online greeting cards*  
*Greeting Cards International*  
Jennifer Hermon  
*"Mother's Bracelet"*  
*JennyLou Jewelry*  
Marshella Merritt  
*SBTS Fdn. logo-ware*  
*CafePress online*  
Jill Fisch  
*Campaign Manager*  
*Awareness Bracelets*

### Presentations

Laura Clow, Corrina Arnold  
*Exhibitors*  
*Great Expectations*  
*Baby Fair*  
*Dallas, Texas*  
Judy Dow  
*Presenter*  
*Hospital Jamboree*  
*Milwaukee, Wisconsin area*  
Mary-Ella Frederich  
*Exhibitor*  
*7<sup>th</sup> Annual Family Café Conference*  
*Orlando, Florida*  
Kelly Peters  
*Speaker*  
*Common Problems in Pediatrics*  
*Conference ~ Salt Lake City, Utah*



### Monetary Gifts

**Patrons:**  
**\$500 and above**  
Amyx, Inc.  
Jack C. Hall/*NYL PAC Matching*  
*Charitable Gifts Program*  
Stephanie and Andrew Plaisted

**Supporters:**  
**\$250 and above**  
James Hilke/*Microsoft Giving Campaign*  
Catherine and Keith Lafferty

### Memorial donations

Mary E. Ashby  
Noah Cooper  
Storm Hall  
Tiger Hall  
Ben Haygood  
Tyler Wayne Mize  
Andrew "Drew" Plaisted  
Nora Waananen  
Emma Grace Zagariello

### Honor Donations

Erik Crouch  
Danny Dobbs  
Nathan Gariepy  
Robin Haygood  
The Haygood Family  
Cory and Mary Hilke  
The Monaco Family  
Alex Nawn  
Kevin Nawn  
Justice Quent  
Mattina Vitale

## AAP Endorses ACMG Report

In a press release dated May 12, 2005, the American Academy of Pediatrics (AAP) endorsed the Newborn Screening Report from the American College of Medical Genetics (ACMG). Noting that the ACMG examined best evidence on screening for certain conditions before compiling its recommendations, the AAP acknowledges that the recommended screening would eliminate the geographical disparity currently present in state programs. The AAP also stressed that an explicit follow-up system must be in place to support the effects of expanded screening on pediatric practices.

*Be proactive,  
not a passenger.  
Make something  
happen.*

## Positive <NBS> Negative

The positive aspect of newborn screening news is that states are developing their programs to include more disorders and additional follow-up services. The negative aspect includes the potential for liability when a hospital, or state, fails to meet national recommendations.

Whether the glass is half-full or half-empty, we must not forget the most negative aspect of all: the death or disability of a child when those consequences could have been prevented.

## NBS Advocates Honored in Virginia

In a ceremony on June 30, 2005, Jana and Tom Monaco of Woodbridge, VA, were commended by the Prince William County Board of Supervisors for their efforts in expanding newborn screening in their state.

"Though it is always bittersweet for us, we feel proud to make a difference for others," says Jana. "We take comfort in knowing that the babies in Virginia are going to have a better start in life."

The couple's son Stephen was 3 years old when he experienced metabolic crisis because of undetected IVA. Suffering severe disabilities, Stephen, now age 8, has been the inspiration for the Monacos' advocacy. His 3 year old sister Caroline also has IVA, but early

detection and a special diet have prevented the consequences experienced by her brother. Two older boys are unaffected.

The Monacos testified before legislative committees, educated lawmakers about the need to expand the state's newborn screening program, and willingly told their story to the media on several occasions. These efforts contributed to the state's new mandate. Effective March 1, 2006, Virginia will significantly expand its newborn screening panel from 9 to 28 disorders in keeping with the enhanced Virginia Newborn Screening Act. The act was signed into law by Gov. Mark Warner in April of this year.



*The Monaco Family on a recent vacation in the Great Smoky Mountains*

### Editorial:

#### Follow the Leader...please!

Wisconsin has redefined its newborn screening panel to be consistent with the nomenclature in the recommendations set by the American College of Medical Genetics. While methods for counting disorders vary among the states and have become a numbers game in some instances, Wisconsin holds to a higher standard—a standard that reflects the truth of its newborn screening status in terms of national recommendations.

We acknowledge that among the states there is disagreement as to the ACMG's recommendations, but we see the value of establishing a uniform approach in naming and counting the disorders. States that screen infants for disorders not included in the national recommendations can identify those additional disorders as "also screened" when they present their panel for public scrutiny. We urge all states to follow Wisconsin's lead in adopting the nomenclature of the ACMG's recommendations.

**To help metabolic families affected by Katrina send *MONETARY ONLY* donations to the National Coalition for PKU and Allied Disorders, P.O. Box 1244, Mansfield, MA 02048.**

### Advocacy Corner

*Tips from Tera Mize*

When contacting newspapers or magazines about reporting on newborn screening, or radio and TV stations about airing newborn screening segments, request that they provide contact information for Save Babies Through Screening Foundation (1-888-454-3383 and [www.savebabies.org](http://www.savebabies.org)).

Be sure to ask to review the final draft of what they intend to publish/air. I was once interviewed by a newspaper that said MCADD is a machine!

## Education: Key to Success!

Having attended and provided public comments at all meetings of the [federal] Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, I can tell you that education is widely recognized to be one of the keys to success of newborn screening expansion! I have seen this issue being addressed very proactively all along, and the last Subcommittee on Education meeting provided much insight to wonderful progress in this area.

Many of the relevant physician and advocacy groups were at that meeting, and what I see coming up for education looks very promising, from the OB-GYN's to pediatricians and on... We will be hearing MUCH about newborn screening education and the benefits of its invaluable importance!

*-Micki Gartzke, Director of Education and Awareness, Hunter's Hope Foundation  
www.huntershope.org*

## Speaking Out

Kileen Hall, newborn screening advocate and member of the Board of Directors of Save Babies Through Screening Foundation, participated in a panel on newborn screening at the Mountain States Genetic Network annual conference, held July 14-16 in Englewood, Colorado. Others on the panel were Jeffrey R. Botkin, MD, MPH, Professor of Pediatrics and Medical Ethics, University of Utah; Joseph D. Marinec, JD, Consumer; and Bradford Therrell, PhD, Professor and Director, National Newborn Screening and Genetics Resource Center, University of Texas Health Science Center, San Antonio.

The Mountain States Genetic Network is comprised of Arizona, Colorado, Montana, New Mexico, Texas, Utah, and Wyoming. Kileen Hall lost two infant sons, Storm in 1996 and Tiger in 1999, to VLCAD Deficiency which was not included in their newborn screening in New Mexico. The boys' stories appear in the Family Stories section of [www.savebabies.org](http://www.savebabies.org). New Mexico screens for six disorders and has not adopted the use of tandem mass spectrometry.

*From the sad-but-true file:*

## We know that more education is needed when...

...a geneticist incorrectly advises a parent that her 2 ½ year old son cannot possibly have MCAD Deficiency because that disorder affects only babies;

...newborn screening is called the PKU test;

...a reporter writing about newborn screening refers to MCADD as a machine;

...a high rate of false positive results from newborn screening is accepted as unavoidable;

...a state legislative body enacts law to increase the newborn screening panel and sets a target date of nearly a year in the future—with no provision to

safeguard newborn health through the services of a private laboratory in the meantime.

## In Memoriam

### Wendy Thorson

*Sept. 20, 1978 - Sept. 10, 2005*

Daughter to Mary Thorson of Conyers, GA, and sister to Tracy, Valerie, and Brad

Wendy lived with **Trifunctional Protein Deficiency** for nearly 27 years.

## The Word on Campus

Pre-med and other science and nursing clubs on college campuses are a good place for newborn screening presentations. In addition to the obvious educational benefit for a course of study and a potential career, the information has relevance to these students as future parents.

Contact Save Babies Through Screening Foundation for newborn screening pamphlets to accompany these presentations. Information from the foundation's website can be copied provided that the foundation is cited as the source. Also, please tell us when and where your event will occur so that it can be noted in our records of newborn screening awareness activity. Write to [email@savebabies.org](mailto:email@savebabies.org) or call 1-888-454-3383.

## NBS AWARENESS BRACELET ORDER FORM

To view bracelets visit [www.savebabies.org/fundraising/Programs.php](http://www.savebabies.org/fundraising/Programs.php)

Quantity at \$2.00 each: \_\_\_\_\_ adult size \_\_\_\_\_ child size

Ship to:

Name \_\_\_\_\_

Address \_\_\_\_\_

Make check or money order payable to:

**Save Babies Through Screening Foundation, Inc.**

For credit card purchase:

Credit Card # \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_

Expiration Date \_\_\_\_/\_\_\_\_

Signature \_\_\_\_\_

*as it appears on credit card*

Mail to:

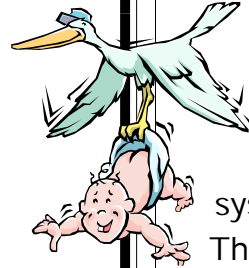
**Jill Fisch**  
205 Delhi Rd.  
Scarsdale, NY 10583

## The Follow-up Argument

One of the arguments against expanding newborn screening is lack of follow-up care. Follow-up care issues should not be prohibitive to supplemental newborn screening.

Affected children will be born, anyway, whether or not a follow-up system is in place.

The follow-up they will receive from being comprehensively screened will be much less complicated than the follow-up that results from not being comprehensively screened.



*Please detach and enclose with your donation.*

## Your Donations Make a Difference!

**Won't you help Save Babies?**

Donation Amount \$ \_\_\_\_\_

If paying by credit card:

Name \_\_\_\_\_

credit card number \_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_/\_\_\_\_\_

Address \_\_\_\_\_

expiration date \_\_\_\_/\_\_\_\_

signature \_\_\_\_\_

*as it appears on credit card*

This gift is in \_\_\_Memory of \_\_\_Honor of  
*Full Name/address of designated person or family:*

date \_\_\_\_\_

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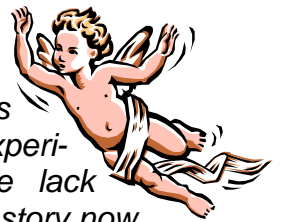
## Back Issues of NBS Newsletter Available Online

When you wish you'd kept the back issues of the NBS Newsletter because there is an article you could use right now, check online at [www.savebabies.org/newsletter.php](http://www.savebabies.org/newsletter.php) to find the issue you need. New subscribers might like to take a look back, too!

### Pictures & Stories Needed

## Share your story with SBTS Foundation

*The need for new stories and pictures is ongoing. If you haven't shared your experience with newborn screening, or the lack thereof, please consider submitting your story now. Remember to include a completed release form. If you send in professional photographs, you will need to include a release from the photographer, as well. Release forms can be downloaded from our website at [www.savebabies.org/share.php](http://www.savebabies.org/share.php) or email [terilyn@savebabies.org](mailto:terilyn@savebabies.org) to request a form.*



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*Formerly Tyler For Life Foundation*

## **Save Babies Through Screening**

**Foundation, Inc.**

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