

The Illuminator

“A light in a world of darkness”

October 2009
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2009 BDSRA CONFERENCE RECAP

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Special Child



Thomas Christian Anderson - INCL



Australian Chapter (story on page 9)

Batten Disease Support &
Research Association
166 Humphries Drive
Reynoldsburg, OH 43068
1-800-448-4570
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Another BDSRA Conference has come and gone. Our time spent in St. Louis was one for the record books with many memorable moments including the Kids' Parade and the opportunity to learn about research first hand from the scientists conducting it. As always, families had the opportunity to catch up with one another, and new families had the opportunity to meet fellow BDSRA families for the first time.

During Saturday night's award banquet, BDSRA was honored to pass out research awards in the amount of **\$273,826!** The research abstracts will be available on the BDSRA website. Those individuals who received the awards are as follows:

J Alfred Rider Memorial Research Award
Shannon L MacAuley (Mark Sands, PhD)

Washington University in St. Louis
\$40,000

“Activated Astrocytes Therapeutic Targets in INCL”

(Also a BDSRA Postdoctoral Fellowship)

Sandra Hofmann, MD, PhD

University of Texas Southwestern
\$60,000

“Enzyme Replacement Therapy for Palmitoyl Protein Thioesterase Deficiency (INCL)”

(Purpose of study: To develop enzyme replacement therapy for Infantile Batten Disease.)

Anil B. Mukherjee, MD, PhD

NIH/NICHHD
\$65,000

“The development of a treatment strategy for patients with INCL, caused by PPT1 nonsense mutations”

Krystyna Wisniewski Memorial Research Award

Su Xu (Peter Lobel, PhD)

Center for Advanced Medicine and Biotechnology/Rutgers University
\$31,500

“Intrathecal Enzyme Replacement Therapy for LINCL”

(Also a BDSRA Predoctoral Studentship)

Martin L. Katz, PhD

University of Missouri
\$37,326

“A Possible Canine Model for Infantile NCL Therapy Development”

Sunita Biswas, PhD

Massachusetts General Hospital, Boston
\$40,000

“Generation of Human NCL iPS cells for study of NCL disease biology”

(Also a BDSRA Postdoctoral Fellowship)

During Saturday night's banquet, we also awarded families, individuals, and chapters with fundraising awards. We award families in three different categories:

- a) Fundraising for Research
- b) Fundraising for Programs & Services
- c) Combined Chapter Fundraising

In the category of Fundraising for Research, the winners were:

- 1) Mike & Diane Carbrey -- \$51,000
- 2) Eric & Christine Thelen -- \$45,000
- 3) Joni & Pete Metcalf/MN Chapter -- \$30,000

In the category of Fundraising for Programs & Services, the winners were:

- 1) Charlie & Wanda Leffler/Debbie Lowther -- \$10,100
- 2) Lorie Williams -- \$10,008
- 3) Joe & Kathy Allio -- \$8,000

In the category of Combined Chapter Fundraising, the winning chapters were:

- 1) Australia Chapter -- \$116,000
- 2) Southeast Chapter -- \$52,000
- 3) Heart of America Chapter -- \$51,000

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Research Update

University of Iowa News Release - Sept. 21, 2009

UI scientists use blood brain barrier as therapy delivery system

The blood brain barrier is generally considered an obstacle to delivering therapies from the bloodstream to the brain. However, University of Iowa researchers have discovered a way to turn the blood vessels surrounding brain cells into a production and delivery system for getting therapeutic molecules directly into brain cells.

Working with animal models of a group of fatal neurological disorders called lysosomal storage diseases, the UI team found that these diseases cause unique and disease-specific alterations to the blood vessels of the blood brain barrier. The scientists used these distinct alterations to target the brain with gene therapy, which reversed the neurological damage caused by the diseases.

The findings, which were published Sept. 13 in Nature Medicine's Advance Online Publication (AOP), could lead to a new non-invasive approach for treating neurological damage caused by lysosomal storage diseases.

"This is the first time an enzyme delivered through the bloodstream has corrected deficiencies in the brain," said lead investigator Beverly Davidson, Ph.D., UI professor of internal medicine, neurology, and molecular physiology and biophysics. "This provides a real opportunity to deliver enzyme therapy without surgically entering the brain to treat lysosomal storage diseases.

"In addition, we have discovered that these neurological diseases affect not just the brain cells that we often focus on, but also the blood vessels throughout the brain. We have taken advantage of that finding to deliver gene therapy, but we also can use this knowledge to better understand how the diseases impact other cell types such as neurons," she added.

"Our discovery allowed us to test the idea that the brain cells might be able to make use of the reintroduced enzyme to stop or reverse the damage caused by the accumulated materials," said Davidson, who also is the Roy J. Carver Professor in Internal Medicine. "In the treated mice, the affected brain cells go back to looking normal, the brain inflammation goes away and the impaired behaviors that these mice have is corrected."

The team modified a deactivated virus used for gene therapy so that the virus expressed copies of the unique brain-targeting peptide on its outer coat, and also carried the genetic blueprint for the missing enzyme. The study showed that the modified virus targeted the blood vessels in the brain and caused the blood vessel cells to produce the enzyme. Most importantly, the researchers found that the enzyme was secreted into the brain tissue in sufficient quantities to correct the disease symptoms and problems. The team was able to use this approach to treat two types of lysosomal storage disease in mice, suggesting that the approach could be used for other types of lysosomal storage disease and possibly other neurological disorders.

The study was funded by grants from the National Institutes of Health and from the Batten Disease Support and Research Association.

StemCells, Inc. Demonstrates Protection Of

Neurological Function In Batten Mouse

Preclinical Proof-of-Concept Data for the Company's Neural Stem Cells

Published in Cell Stem Cell

PALO ALTO, Calif., September 3, 2009 – StemCells, Inc. (NASDAQ: STEM) today announced the publication of preclinical data demonstrating for the first time that transplantation of its proprietary, purified human neural stem cells delays the loss of motor function in a mouse model of infantile neuronal ceroid lipofuscinosis (NCL). NCL, commonly referred to as Batten disease, is a fatal neurodegenerative disorder in children. This paper, "Neuroprotection of Host Cells by Human Central Nervous System Stem Cells in a Mouse Model of Infantile Neuronal Ceroid Lipofuscinosis," was published online today in the peer-reviewed journal *Cell Stem Cell*, and will be featured in the September 2009 print edition. For the entire story, see: www.stemcellsinc.com/news/090903a.html

More Research News on Pg 9

Fundraising News

Event Updates

Race for a Miracle/Cook for a Cure - Mandeville, LA

The 1st Annual "Race for a Miracle" was held on June 7th to commemorate Batten Disease Awareness Day. The event was a huge success! While they expected 200-250 to turn out for the race, they had 580 registered participants! Total amount raised was **\$14,546**. Way to Go!!! For more information, please visit Mary Payton's Miracle Foundation website at: www.marypaytonsmiracle.com

Jump for a Cure – Plainview, NY

The amazing Elna Faret went skydiving on Batten Disease Awareness Day, June 7. This was her second jump to benefit BDSRA, and she raised **\$600!** She did it to bring awareness for her grandson, Ryan, who has Batten Disease. Way to Go, Elna!

8th Annual Mendota High School Relay for Charity - Mendota, IL

The 8th Annual Relay for Charity began at noon on June 12 and continued until noon on June 13. With over 50 runners participating, another couple dozen walked with a baton, and covered over 500 miles. This year's relay exceeded **\$3,800** to BDSRA! To date the relay has generated over \$45,000 to the Batten Disease Support and Research Association (BDSRA) with all funds directed to research. For more info, please visit their website at: <http://mendotacc.weebly.com/index.html>

Wine Tasting Fundraiser - Eagan, MN

The first annual Wine Tasting event was hosted by Warren & Lois Shuros and Minnesota Chapter and held on June 25. It was very successful for the first event and raised **\$4,200** for BDSRA. Congrats to the Minnesota Chapter & Shuros Family!

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**10th Annual Melissa Froio
Golf Tournament -
Sicklerville, NJ**

The 10th Annual Melissa Froio Foundation Golf Tournament was held on June 19 at the Ramblewood Country Club. The event was a huge success raising over **\$12,000** for BDSRA's research initiatives. Thank you to the Froio Family for their continued efforts that have resulted in such an awesome contribution! For more information, visit their website: <http://melissafroio.org>.

**"Action for Asher" Kick-a-
Thon and AsherBash4 -
Norwalk, OH**

The Nikolajevs Family did a great job again this year with two fundraisers held in June and July. "Action for Asher" Kick-a-thon held on June 20 and the AsherBash4 on July 18 were huge successes and raised over **\$7,000**. AsherBash4 included a Motorcycle Run "Dash to the Bash Dice Run" which concluded with a concert benefit at the Knucklehead Saloon in Huron, OH. Hats off for a job well done!

**6th Annual "Kevin's Fund"
Golf Tournament -
Hagerstown, MD**

The Lumm Family, along with the Maugansville Ruritan Club, hosted their 6th Annual Kevin's Fund Golf Tournament on June 26. All proceeds benefited a special fund to assist children with special needs in Washington County, in addition to BDSRA. Thank you Marty & Vicki for hosting a wonderful golf outing!

**Kick Batten's For Carl -
Arlington, WA**

Lisa Boyle, friend and Physical Education teacher at Post Middle School in Arlington coordinated a fundraising event in honor of Carl Bergam who passed away in February this year. They held a Soccer Tournament and Silent Auction on August 8th and raised **\$2,000!** Thank you to the Pre-K - 5th Graders, family and friends who attended this event and made it such a success. They look forward to doing it again next year.

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Special Child: Thomas Christian Anderson (INCL) from Nevada, Iowa

Thomas was born August 19, 1998. He developed normally but around 15 months we noticed that he wasn't walking the best and was falling a lot; he was also not chewing his table food very good and was spitting it out. We resorted back to stage 3 baby food and he seemed to do better with that but after a few weeks it was taking 2 hours to feed him each time and there was more on his highchair than what went in his mouth. We went to the pediatrician and told him something was wrong. He told us that boys are a little slower and we shouldn't worry, but he did do a blood work-up on Thomas and everything came back normal. After a few more weeks had passed, Thomas is now back on the bottle 75% of the time and only eating a little bit of baby food but he seemed to be spitting up half of what he was taking in. Thomas had started to crawl more than walk and was only babbling and not saying the only 3 words he ever learned: mama, dada, baba. He also started to press his thumbs into his eyes and when he was crawling he would put his head down on the ground and press his face into the carpet. We could not understand what was happening to our little boy. Thomas now cries constantly unless he is watching Teletubbies, listening to musical books, or sleeping.

In April 2000, we met with the pediatrician and he scheduled a MRI. The MRI results were not good, as it showed Cerebral Atrophy. One week later we were sent to Blank Children's Hospital to see a Pediatric Neurologist. He did more blood work and told us to come back in 6 weeks but also made appointments to see an eye doctor and the pediatric Geneticist in Iowa City, but wanted us to come back 2 days later and do an EEG. Those results showed no signs of seizure activity. We did the eye exam and found out he was near-sighted and had stigma's in both eyes & should have glasses. We saw the Geneticist and they did blood work and everything was normal again.

July 2000 we went to the Children's Hospital in Chicago and Thomas cried about the whole time in the car. We met with the pediatric neurologist and they weighed him and he was 27lbs at almost 2 years old. The doctors wanted another MRI, and a muscle, nerve & skin biopsy done. We had the MRI done back home and then drove back to Chicago to have the biopsies done. The biopsies came back normal.

September 2000 we went to Iowa City to have an ERG (ElectroRetinoGram) of his eyes. What little results they got back from the ERG, they ran thru a database of rare disease and Batten Disease was one of those that came back. They started to test for that & worked with the University of Chicago and used the biopsy materials for further testing. By this time Thomas was not walking anymore and was having a hard time crawling. For the next few months we just dealt with the all the regression of Thomas. I was back to giving Thomas a bottle of milk every 4 hours and then he would spit most of it all out afterwards.

February 2001 - (2 ½ years old) Thomas was staying at his grandma's house overnight for the Super Bowl and the next day my mom called me in the afternoon and told me that she thought something was wrong with Thomas. I came right over and called the pediatrician and he said to bring him right in. We took him in and the doctor said that he thought Thomas was having a seizure due to dehydration (remember he was spitting up everything he was drinking). They admitted him into the hospital. He was to have a G-tube placed two days later, but woke up with a fever of 102, did tests and he had Influenza A & RSV. Six days later he had a Nissen-Fundoplication done and a G-tube placed. On that same day I received a call from University of Iowa Hospital and they confirmed that Thomas had Infantile Batten Disease. The pediatrician told me that he might not make it thru the night and probably won't make it thru the next month. **WOW what a shock!** I just found out my only child will die and I have a 25% chance of having another child with this disease. Thomas was in the hospital for 3 weeks and before we left I met with Hospice & was given a tour of the Hospice house. Hospice sent someone to help take care of Thomas for 4 hours each day at the babysitter's house & at our house on Saturdays. Thomas has now pretty much stopped crying and barely makes a sound unless he has a seizure.

April 2001 - We were contacted by Make A Wish and were granted a week long wish to stay at Give Kids the World and go to Disney World and Epcot Center. My parents came with us to help with Thomas. Thomas is now pretty much 'bed-ridden'. He can not hold his head on his own, he can't sit or even roll from his back to his side or vice versa. He doesn't understand anything we say to him but does seem to recognize our voices at times. We have been told that he is deaf and blind but he can see very bright objects (like the sun) and squints at it in the morning & his hearing is probably better than mine - he will jump at a pin drop.

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There hasn't been much change in Thomas since 2001. He has had ALOT of ear infections and has had his second set of tubes in since December 2001. We have had a lot of issues with sleeping at night over the years and have tried several different sleep medications; he also has multiple daily seizures. In August 2009 Thomas turned 11 years old & is currently 4 feet tall and keeps growing lengthwise but has weighed about 46 pounds for the past 6 years. We feed him 4 to 6 ounces of Pediasure thru his G-tube every 4 hours, 100% of the time. I take Thomas to the babysitter's house everyday while I work and she has been taking excellent care of him since he was 2 months old. Since Thomas needs 24 hour care, he is now the only child that she watches. In 2006 we built a house RIGHT next to my parents' house on an acreage and it has been wonderful having them right there when we need help with Thomas or to run errands.

Even though Thomas is not aware of much around him, we can still get him to smile and laugh at times. We are so blessed to have Thomas in our lives. We love him so much and are thankful for every minute we get to spend with him.

~~ Submitted by Mike & Stephanie Anderson, parents

Tracey Wilson, Director of Development, Resigns from BDSRA

We regret to inform everyone that as of August 28th, Tracey Wilson no longer works for BDSRA. She resigned to accept a position of Executive Director for another non-profit organization. Tracey was only here a short time, but she helped the organization build a firm foundation in fundraising and awareness building. We are in the process of looking for a new Director of Development and will notify everyone as soon as a replacement has been hired. We wish Tracey the best of luck in her future endeavors.

Chris Hawkins, 704-590-8226

Teach & Be Taught DVD: Volume 3 – Educating Students with Infantile or Late Infantile Batten Disease

BDSRA Education Specialist, Wendy Bills, PhD, has created a third volume for her Teach and Be Taught DVD series. This newest DVD is specifically for schools and teachers who educate students with INCL and LINCL. It can also help teachers in preschool and daycare settings. It helps to address some of the challenges of a child who cannot speak for his/herself as well as ways to integrate more sensory directed learning techniques. This DVD is also great for students who are in the later stages of JNCL. This DVD was unveiled at Conference in July and has been going out to families around the country. If you would like a FREE copy of Wendy's latest DVD, please email Amy Lombardi at kirka@bdsra.org. If you do not have the other materials available in the Teach and Be Taught series (book and first 2 DVDs) you can also request these FREE of charge. With school back in full swing, this would be a great resource for any teacher of a child with INCL or LINCL.

New Dementia Book Now Available

BDSRA Nurse Nancy Carney has created a new publication called "Dementia in Children, Teenagers and Young Adults: A Guide for Parents, Teachers and Care Professionals". This book addresses the dementia issues that many families experience, especially those who have children with JNCL. The book explains what can cause dementia, symptoms, and different therapy interventions. To receive a copy of this FREE publication, please contact the BDSRA National Office.

New Resource Guide for Parents Helping Siblings Now Available

Amy Lombardi, Coordinator of Family Services, has created book filled with resources from the internet, books, and articles that can help parents address the tough topics that may affect their unaffected children/siblings. Some of the topics covered include talking to your child, of any age, about their sibling's illness, living with a sibling who has special needs, books that talk about grief and loss, etc. The resources provided are not specific to Batten Disease, but help parents navigate conversations about related topics. This publication is FREE and can be obtained by contacting the BDSRA National Office.

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Mostacholli Dinner - Lincoln Park, MI

The Rhodes Family hosted a delicious Italian Mostacholli Dinner at the Living Word Church of God in Taylor, MI, on August 22. The benefit raised **\$900** for BDSRA! Great job, Rhodes family!

Garage Sale & Raffle - Templeton, MA

The Montville Family hosted their first multi-family Yard Sale & Raffle to benefit Late Infantile Batten Disease research on August 22. Included in the raffle were Red Sox tickets and a BMW bicycle. The event raised nearly **\$5,000!** Great job, Montville Family!

2009 Hayden's Hope Golf Classic - Two Rivers, WI

The Thelen Family hosted the Hayden's Hope Golf Classic which took place on August 22 at the Emerald Hills Golf Course. There was a shotgun start at 2 p.m. with dinner and music following at 5. All proceeds are benefiting INCL research. For more information, visit their website: www.help Hayden.com.

Trivia Night for a Cure – Valparaiso, IN

Jim & Linda Smith organized a Trivia Night on August 29 with all proceeds to benefit the BDSRA 2010 Conference on behalf of the Midwest Chapter. The night was a huge success with 175 participants and over **\$7,500** raised for BDSRA! Great job, Smith Family!

BBQ Blues for Batten - Gateway, CO

The Gateway Canyon Resorts, located in southwest CO, hosted a fundraiser for BDSRA. The event brought together the best of summer on August 29 with the first annual BBQ Blues. This event featured a number of well-known blues bands for a day of memorable music, along with a BBQ contest to bring some of the region's best grillers to the festivities. Visit their website: www.gatewaycanyons.com. Great job Melissa Sisson and the Gateway Canyon Resort staff!

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**8th Annual Batten Disease
Golf Tournament –
Vacaville, CA**

The Fairfield Police Officers Association sponsored this annual golf tournament in honor of Police Sergeant Joe Allio's family and his daughters, Catie and Annie, who have Juvenile Batten Disease. The event took place on September 11 in Vacaville, California. Money raised goes to fund BDSRA's programs and services. Thank you. Fairfield Police Officers and Allio family!

**Bash for Batten's -
Maple Grove, MN**

Brandy & Carl Wempner organized a "Bash for Batten's" on September 13 at the Cambria Suites in Maple Grove, Minnesota. The event included an Italian Buffet Dinner and silent auction. Warren Shuros, BDSRA Board Member, and Laura Butenhoff were both key speakers. The event raised over **\$1,200!** Great job, Brandy and Carl!

**Motorcycle Run –
Shellbina, MO**

Tracy Frye held a Motorcycle Run and event on September 17. The event raised funds for BDSRA and Kufs (Adult NCL) research. The event also included an auction and featured the amazing motorcycle displayed at our St. Louis Conference! Overall the Motorcycle Run was able to raise **\$15,000!** We're sure everyone had a terrific time!

**Metro NY/NJ Chapter
Garage Sale**

Fred & Janet Surrey had a huge garage sale the weekends of September 17/18 and 25/26. Proceeds raised will go to Batten Disease Research. Altogether they were able to raise over **\$3,500!** Amazing job, Surrey family!

Garage Sale – Crystal Lake, IL

Kerrie Kurth was Sara Pfaller's teacher. Even though Sara passed away earlier this year, Kerrie has remained committed to raising funds and awareness for BDSRA. On September 25/26, Kerrie held a garage sale. All **\$750** that was raised will go towards the 2010 BDSRA Conference in Chicago, IL. Great job. Kerrie!

From the Nurse's Corner

by Nancy Carney, RN
nancycarney@bdsra.org
1-877-642-5512

Cough is defined as an explosive expiratory maneuver that, under normal conditions, serves to protect the airways and lungs by clearings inhaled material, excessive mucus, and abnormal substances. In certain diseases states, cough can become, excessive, nonproductive, distressing, and potentially harmful to the airway mucosa, and it can be a factor in the spread of infection. Cough is one of the most common symptoms for which the general population seeks medical attention. Cough is also a symptom of other conditions for which patients with advanced illnesses are susceptible, such as the common cold or pneumonia. Because cough serves as a protective function, ineffective cough puts patients at risk for complications such as pneumonia and atelectasis. Also, patients with weakened muscles due to neurological diseases like Batten Disease are often not able to cough effectively.

Causes of cough: colds, bronchitis, pneumonia, asthma, heart failure, reflux disease, and aspiration are a few of the most common. Complications of cough are: Cardiovascular - hypotension, arrhythmias; Constitutional - sweating, anorexia, fatigue; Gastrointestinal - reflux, herniations; Genitourinary - urinary retention; Musculoskeletal - strain to rupture abdominal muscles, rib fracture; Neurological - cough syndrome, dizziness, headache; Ophthalmologic - rupture of the subconjunctival veins; Psychosocial - self-consciousness; Respiratory - dyspnea, exacerbation of asthma, laryngeal or tracheobronchial trauma; Skin - petechiae and purpura.

History and Physical Examination - the etiology of the cough can be determined in 80% of patients with advanced illnesses by a thorough history and physical examination. Some of the questions needed to be asked are: the primary disease, any other disease processes, allergies, exposures to infections or substances that may affect the respiratory system, past surgeries, current medications and recent changes in appetite, weight, sleep, voice, activity tolerance or energy level. Evaluate the cough by asking questions like character of the cough (barking, hacking, brassy honking, dry, wet), sputum

production (onset, color, volume, tenacity, any changes over time), pain related to cough (location, severity and management), aggravating factors (inspiration, activity level and eating), alleviating factors (rest, positioning, and medications), influence of positioning (lying, upright) on cough, temporal patterns (constant, nocturnal, only during day, or variable), effect of cough on quality of life. Then the actual physical examination: overall appearance, level of anxiety, vital signs (fever, respiratory rate), skin color (ashen, flushed, pink, cyanotic) and character (cool, hot, diaphoretic, presence of edema or finger clubbing), breath sounds (wheezing, rales, rhonchi, crackles, friction rub, diminished or absent), upper airways (nasal drainage, throat irritation, or sinus congestion), neck (vein distention, deviated trachea, edema, enlarged lymph nodes), chest (shape, vein distention, accessory muscles, retractions, or abnormal percussion sounds), abdomen (distention, masses, enlarged organs, tenderness, or abnormal sounds).

Diagnostic Testing - There are diagnostic testing that can be done including chest x-ray, echocardiogram, tomography barium swallow and blood work to help diagnose where the cough may be coming from.

Intervention and Treatment - You want to treat the cause to improve the quality of living, but cough serves a protective mechanism and should not be suppressed unless it is excessive, interferes with sleep and rest, or causes discomfort. Cough can be triggered by more than one cause and therefore may require more than one treatment at a time. There are many medications to suppress cough and between your Doctor and the Pharmacist, you will be able to choose one to be effective. There are breathing treating and breathing techniques that also may be very helpful. The best thing to do is to keep a diary or record of how long the cough has been going on and what seems to help or not help the situation. Do not self treat for over one week before seeing a Doctor.

Family Services News

Amy K. Lombardi, MSW, LSW
Coordinator of Family Services

Caregiving for someone who is seriously or chronically ill is a daunting task, and an emergency can be frightening. Taking steps in advance will help you handle a crisis, should one occur.

By Linda Foster, MA
Medically reviewed by Lindsey Marcellin, MD, MPH

Caregiving can be a demanding and intimidating responsibility, especially when you know the person you are taking care of might one day be facing a medical emergency. Should that happen, the situation will go more smoothly for you as the caregiver and for your loved one if you're prepared in advance and know exactly what you'll need to have handy and what steps you'll need to take.

5 Basics Steps

to Emergency Preparedness

Basic training and first aid supplies will go a long way toward giving you a greater comfort level as a caregiver:

1. Take a CPR class. You can use CPR to revive someone whose heart has stopped beating or who has stopped breathing. Bill Guerra, RN, BSN, of the Seven Hills Surgery Center in Henderson, Nev., recommends that all caregivers take a CPR class so you understand the ABCs of emergency response: A (airway), B (breathing), and C (circulation).

2. Learn the Heimlich maneuver. CPR training also involves learning the Heimlich maneuver – how to clear someone's airway in the event a foreign object or food becomes lodged in the throat. Attempting the Heimlich maneuver without proper training can injure your loved one. Visit the American Heart Association Web site (www.americanheart.org)

to learn about the Heimlich maneuver and where to take a CPR class. Also, find more on the warning signs of heart attack and stroke.

3. Maintain a well-stocked first aid kit. Thoroughly read the manual as soon as you buy your first aid kit. Check monthly to make certain you have adequate supplies, and replace any items that may have expired. An ear thermometer may be the best choice if your loved one might accidentally bite down on an oral thermometer. Keep a second first aid kit in your car; keep both out of the reach of children.

4. Create a medical provider list and keep copies handy. "Always have all doctors' numbers in a central, convenient place; put one copy of the list in your purse or wallet and one on the refrigerator. Include a list of all medications and other health facts and conditions. Make another list of family members to notify," says Guerra. When applicable, have phone numbers of people who can watch your children or pets and secure your home, should you need to take your loved one to the emergency room.

5. Buy an automatic blood pressure cuff. You can buy a good one at any local drug store. Learn how to use it and practice using it regularly. On regular doctor visits, take the cuff with you to check its accuracy against the physician's blood pressure monitor and to ensure you are using it correctly.

4 Steps to Take

in an Emergency Situation

Keeping your wits about you will be key, should an emergency arise. Practice these steps so that you'll know exactly what to do in an emergency:

1. Assess the situation. Is your loved one bleeding or did he have a blow to the head, a fall, or an

allergic reaction? Is he experiencing any symptom specifically related to his illness? What were you both doing just prior to the emergency? Is he responsive? Are his pupils enlarged and are they the same size? Was there a complaint of pain or anything else relevant? What is different or unusual about your loved one? "Observe, observe, observe," says Guerra in anticipation of calling 911.

2. Call 911. Do this when you have the even slightest hint that your loved one is facing a life-threatening emergency. "A call to 911 is the best thing in any emergency," says Guerra. Do not attempt to take anyone with a potentially serious problem to the hospital yourself; instead, call 911 immediately. Give the 911 operator as much information as you can, so that emergency personnel can be fully prepared to assist your loved one when they arrive, says Guerra. It's crucial to accurately describe the situation and speak slowly and clearly when talking with the 911 operator. Mention any pre-existing conditions, such as a history of heart attack, diabetes, a bleeding disorder, or asthma.

3. Loosen any tight clothing. Make sure your loved one has nothing constricting the airways, like a restrictive shirt or tie, and keep them in a comfortable position while you wait for help.

4. Comfort and communicate. Talk to your loved one until 911 arrives. Guerra recommends that you keep your loved one awake by talking to them, but don't encourage them to talk. Take slow, deep breaths to help yourself stay calm as well. For a caregiver, an emergency can be both alarming and frightening, but when you're prepared, you can make a tremendous difference in your loved one's well-being

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We also held a Chapter Poster Contest in which each BDSRA Chapter is invited to create a poster for display at the Conference. A small anonymous panel judges each poster and the winning chapters are announced. The winners of this year's Chapter Poster Contest were:

- 1) Ohio Chapter
- 2) Heart of America Chapter
- 3) Australia Chapter

Congratulations to all of our winners and thank you to everyone for your hard work and fundraising throughout the year!

Looking Ahead.....

The 2010 BDSRA Conference will be held **July 29-August 1** at the **Chicago Marriott Oak Brook** hotel in Oakbrook, IL (Chicago suburb). For those of you who attended the 2001 Chicago Conference, the 2010 Conference is being held in the same location. More information about the 2010 Conference will be available in the January and April Illuminators.

For more information on the hotel, please visit their website at: <http://www.marriott.com/hotels/travel/chiob-chicago-marriott-oak-brook/>

New BDSRA Postage Stamp!

Please visit Zazzle.com and research "Batten Disease" to place your order for our new customized U.S. postage stamps. 10% of all proceeds will benefit BDSRA. Great idea to use for thank you letters following a special event.



BDSRA Medical Equipment Exchange

BDSRA maintains a Medical Equipment Exchange for its families. We have many pieces of equipment available including wheelchairs, suction machines, bath aides, feeding supplies, etc. These items are available to BDSRA families at NO COST! The latest list of our inventory can be viewed by going to the BDSRA website under the *Family News* section. If you need a piece of equipment or have equipment to donate, please contact Amy Lombardi (kirka@bdsra.org).

SIBS Fur-Raiser

We rolled out a special "fur-raising" opportunity for our SIBS group at this year's Annual Conference held in St. Louis, MO. Our new Batten Bear is available to sell in your local community. Order forms are available by emailing BDSRA with your interest. Selling for \$5, this is an easy fundraiser to take to schools, youth groups and community events. We hope that all Siblings will participate in this fundraiser. Prizes will be awarded to top sellers.



Good Luck!

You Can Now Follow BDSRA on Twitter and Facebook

BDSRA is now on Twitter! You can become a "Follower" of BDSRA by visiting our Twitter page at: <http://www.twitter.com/BDSRA>

You can also get the latest news from BDSRA on Facebook by visiting the BDSRA Cause Page. To visit our BDSRA Cause Page go to:

<http://apps.facebook.com/causes/7771>

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Cardthartic Event – German Village, OH

Cardthartic, a private label greeting card company, teamed up with Amy Parrish, a renowned professional photographer, to create a special card featuring Celia Betz who is 2 years old and suffers from Batten Disease. The event took place on September 26 at the Book Loft in German Village. Half of all proceeds from the sale of the entire line of cards that weekend will go to BDSRA. The event raised over **\$4,400** for BDSRA! Thank you, Cardthartic and the Book Loft! Please visit the Book Loft's website at www.bookloft.com.

Dance & Chinese Auction – Gardner, MA

Jessica Montville, along with friends and family, hosted a dinner, dance and Chinese auction on September 26. The event benefited LINCL research. Jessica received media attention about the event as well! The event raised approximately **\$2,000!** Great job Montville Family!

Highlandville Fun Fest - Highlandville, IA

The Shuros family hosted their 2nd Annual Highlandville Fun Fest on September 26 in their hometown of Highlandville, IA. The event featured a Duck Race, Cow Chip Bingo, Live Auction and Hog Roast. The event raised over **\$11,000** for BDSRA and Batten Disease research! Great job. Shuros family!

Spaghetti Dinner – Pittsburgh, PA

The Grady family held a Spaghetti Dinner and Chinese Auction on September 27 to benefit LINCL Research. They have been gaining awareness for Batten Disease in the Pittsburgh area. Great job on a successful event!

Batten Disease Research Golf Tournament – Dartmouth, MA

Mo & Jean St. Gelais hosted their first ever Batten Disease Research Golf Tournament on September 28 at the Allendale Country Club. Golfers were treated to 18 holes of golf, refreshments, prizes and a prime rib dinner. You can get more information by going to their website at www.battendiseaseresearch.com. They raised approximately **\$15,000!** Great job, St. Gelais Family!

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Upcoming Events

Take Flight Triathlon Pre-Race Dinner - Charlotte, NC

The Hawkins Family and Southeast Chapter are planning this event designed to raise funds while featuring health, wellness, and disease research. Four-time Olympian Sheila Taormina, a world champion triathlete and gold medalist at the 1996 Atlanta Olympic Games will be the keynote speaker. The dinner will take place on October 3, and they expect 500-600 in attendance. Good luck!

Uff Da Fest '09 – Spring Grove, MN

This annual event scheduled on October 3 is a Norwegian celebration to commemorate Autumn in Spring Grove, MN. "Uff Da" typically is an expression of surprise or "Oh No!". Joining the festivities, the Minnesota Chapter and Bluff County Artists Gallery will host a Hog Roast, Art Auction and Raffle for a one-of-a-kind ceramic pot made by the renowned ceramic artist, Dean Schwartz who donated the art piece. For more event details, please visit www.uffdafest.com

8th Annual Golf Tournament – Jemison, AL

Becky Lucas has been raising funds and awareness for Batten Disease for a long time. On October 3 Becky, along with some wonderful friends, will host their 8th Annual Golf Tournament. Teams of two will have a shotgun start at 8:00am. The event will also include a door prizes and a catered lunch. Good luck to Becky! (This was rescheduled from an earlier date due to a rain-out.)

2nd Annual "Miracles Can Happen" Gala Mandeville, LA

The Mary Payton Foundation is hosting it's Miracles Can Happen Benefit which will be held on October 9 at Benedict's Plantation in Mandeville, Louisiana. A live and silent auction will be featured, along with great food and the band, 4 Unplugged. They are hoping this event will be even bigger and better than last year! For more information about this event visit their site: www.marypaytonsmiracle.com.

You can now follow Dr. Jonathon Cooper and the PSDL on Facebook and Twitter

Dr. Jon Cooper is the head of the Batten Disease/Pediatric Storage Disorders Laboratory (PSDL) at the Kings College of London. He is one of our Batten Disease experts and serves on the BDSRA Scientific Advisory Board. As a way to help keep families informed as to what is going on in his lab, Dr. Cooper has allowed families to follow the lab on both Facebook and Twitter. To follow PSDL:

~ On Facebook, go to:

<http://www.facebook.com/group.php?gid=131604681678>

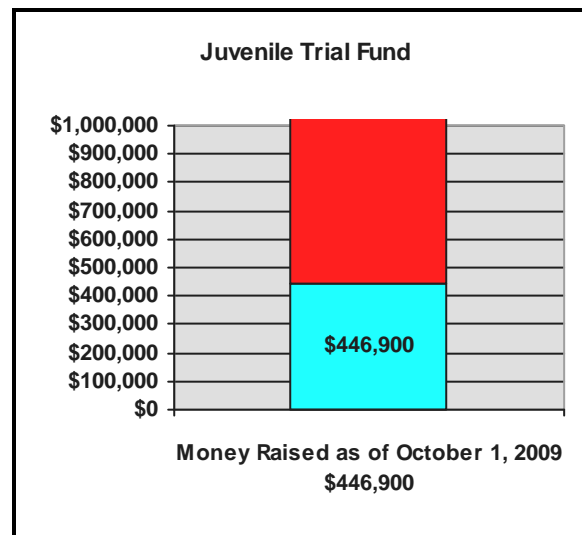
~ On Twitter, go to: http://twitter.com/Batten_PSDL

~ PSDL homepage, go to:

<http://www.iop.kcl.ac.uk/departments/?locator=382>

Update on Clinical Trial for Juvenile Batten disease

The proposal, protocol and budget for the Juvenile Batten disease clinical trial has been received by BDSRA and is being reviewed by members of BDSRA's Scientific Advisory Board and Medical Advisory Board. Funding for the trial is still to be determined. Additional funding by BDSRA may be required.



FDA Sets New Rules on Experimental Drugs -- Wall Street Journal Article

The U.S. Food and Drug Administration finalized new regulations recently designed to provide broader access to experimental drugs for seriously ill people who have exhausted all other commercially available treatments. The new rules, which were posted on the FDA's Web site in August, mostly clarify regulations explaining how patients can receive drugs in development outside of a clinical trial, and set standards for when drug companies or researchers can charge for the treatments.

In hopes of clearing up confusion and potentially making expanded-access programs available to more patients, the FDA issued two new rules that will go into effect in mid-October. The first rule describes how patients can obtain access to drugs outside of a clinical trial and the second rule allows companies and researchers to charge patients for the experimental treatments, potentially allowing more start-up or smaller firms to participate in expanded-access programs. Information for patients is available at www.fda.gov.

The entire article can be viewed at: <http://tinyurl.com/kmdoze>

Dave's HomeRoast Coffees

David Dahl (son Clifford—JNCL) has been in the coffee roasting business for awhile now. His kitchen and ovens become "roasting central" a few times a week. Dave was able to take his coffee roasting business to the internet and has begun serving customers all across the country. Dave sells his premium roasts for \$10.00 per pound. Shipping is \$5.00 for the first bag and \$2.00 for each additional bag. All of the proceeds raised go toward the Juvenile Trial. Please visit Dave's Etsy store at www.daveshomeroast.com and buy a bag of his coffee today! If you have any questions, feel free to email Dave at daveshomeroast@gmail.com.

Australian Chapter Participates in City 2 Surf (picture on page 1)

A Sunday walk or run takes on a whole new meaning when it is shared with 75,000 others in Sydney's annual City2Surf fun run. The route is picturesque and undulating, taking runners from the central city's Hyde Park around the coast to the pristine white sands of Bondi Beach. Along the way there are magnificent sea views including one breathtaking vision of the Sydney Harbour Bridge and Opera House. With clear, sunny skies and blue sea, it takes the runner's mind off the challenge of Heartbreak Hill (which is at about the 7km mark).

Then there is the marvelous crowd support from start to finish - people cheering, live jazz and rock bands, a lone piper, music blaring through speakers. Times by our team members varied between 1.23 hours to 3.06 hours. By the end, runners may have completed 14km but they are also ready to party on the beach. Our fabulous team created awareness with their Battens T-shirts whilst on their journey.



Marty Mailbox wants you to know.....

Have you moved recently? Did you know that if we don't have a current address, the mail we send to you comes back to us and, we are charged fifty cents to get your change of address? Starting in January 2010 that charge can increase!

So it is very important to BDSRA that we know what your new address is as soon as possible and we can get the change put into the database. Won't you help us to save money for other more important things by calling or e-mailing us your new address? Thank you.

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## 2010 Dues Are DUE!!!

The Membership Dues for 2010 are increasing to \$40.00 per family. This is the first time dues have increased since BDSRA's inception in 1987. Membership Dues help pay for things like our Medical Equipment Exchange Program, the Illuminator, and services provided by our Coordinator of Family Services and Family Medical Liaison/Educator. Paying your dues also entitles you to one vote in the annual Board of Directors election.

For your convenience, you can either send a check made out to BDSRA or make payment through our website, [www.bdsra.org](http://www.bdsra.org). Thank you!

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## Ongoing Events

### Cookie Lee Jewelry—Dallas, TX

Jessica Griffith, (in memory of sister Kari Anderson-JNCL) is a Cookie Lee fine fashion jewelry consultant. Jessica has graciously offered to donate a portion of her profits made from her Cookie Lee jewelry sales to BDSRA. To view Jessica's jewelry, visit her website at [www.cookielee.biz/jessicagriffith](http://www.cookielee.biz/jessicagriffith).

When you place an order, be sure to mention "Batten Order" to ensure BDSRA and the Juvenile Trial receives the profits. Jessica is also selling a special line of pink jewelry in memory of Kari. The proceeds raised from this jewelry will also benefit BDSRA. If you have questions, please email Jessica at [jessicagriffith@cookielee.biz](mailto:jessicagriffith@cookielee.biz).

### Kroger Community Rewards

BDSRA is pleased to announce our recent participation in the Kroger Company's Columbus Division (stores located in Ohio and parts of West Virginia) Community Rewards Program. The Kroger Company is committed to helping our communities grow and prosper and will donate funds to BDSRA (up to \$50,000 on a quarterly basis) each time our supporters swipe their Kroger Card for grocery purchases. During the past quarter, BDSRA earned **\$321.00** from the Community Rewards Program. If you live in Ohio or West Virginia and would like to sign up for the Community Rewards Program, please contact Amy Lombardi at [kirka@bdsra.org](mailto:kirka@bdsra.org) or 1-888-379-2546 for instructions.

### Improvements to GoodSearch

Help BDSRA when you shop online and search the web -- download GoodSearch/GoodShop toolbar! Each time you shop at one of more than 1,000 participating stores, BDSRA will earn a percentage of the purchase price -- at no extra cost to you! It also includes a search box and each time you search the web, about a penny is donated to BDSRA. To download toolbar, go to [www.goodsearch.com/toolbars.aspx?charityID=48076](http://www.goodsearch.com/toolbars.aspx?charityID=48076)

## Third Quarter Honor Roll

*BDSRA has been remembered many times in the past three months by family and friends of children with Batten Disease. To all of you we express our deepest appreciation.*

### **HONOR OF ALEXANDRIA & AUDRIANNA ALBAUGH**

M/M Richard Albaugh

### **MEMORY OF TERRELL AMOSAH**

Ms. Eleanor Hodgson

### **MEMORY OF KARI ANDERSON**

M/M Owen Litz

### **HONOR OF THE MARRIAGE OF WILLIAM & KARYN BERTHEL**

Ms. Nicole Heron

### **HONOR OF CELIA BETZ**

M/M James Sheridan

Ms. Betsy Loeb

Ms. Irene Scaramazza

The Columbus Foundation

Chelahnlie Lyons

Stellar Solutions Foundation

### **MEMORY OF DANIEL BREUER**

Mr. Daniel Breuer

### **MEMORY OF PATRICIA CAIN**

The Patricia Cain Family

### **MEMORY OF LULU CALDERON**

The Pincus Family Fund

### **MEMORY OF HANNAH CAULFIELD**

Mr. Michael Caulfield

### **MEMORY OF ALLY CHANCE**

Ms. Marjorie Botellio

Ms. Kimberly Chance

### **HONOR OF CLIFFORD DAHL**

M/M David Dahl

### **HONOR OF NICHOLAS DAINIAK**

Ms. Kathleen McKay

M/M A.W. Hargrove

### **MEMORY OF AARON & ANDY DRAPER**

Ms. Kathleen Schroeder

Ms. Sharon Knutsen

M/M John Abbadusky

M/M Frederick Schauer

Ms. Beverly Haack

M/M Arthur Frett

### **HONOR OF RYAN FARET**

Kevin Anorroun Yvonne Reid

M/M Patrick Cronin Rodnay Schabel

M/M Robert Hasbrouck Debra Reid

M/M John Martakis Bhola Banik M.D.

M/M Bernard Goodman Dennis Olsen

M/M Sverre Andersen M/M P. Calise

Marion Rissman Gunhild Hendrickson

M/M Kenneth Rudin Jo Ann

M/M Lawrence Goldstein

William Wallace & Dr. Jayne Gassman

### **CELEBRATING KRISTINA'S BIRTHDAY**

### **& HONOR OF RYAN FARET**

Ms. Pamela Yourish

### **HONOR OF OUR NIECE,**

### **CANDACE FILECCIA**

M/M Fileccia

### **HONOR OF JACOB GEER AND FAMILY**

Barry Community Foundation Angel Fund

### **HONORING COURTNEY GUNTHER**

M/M Melton Frederick

### **HONOR OF KELSEY HARDIN**

M/M Gary Hardin

### **JACOB'S WISH**

M/M Thomas Basch

### **MEMORY OF JAMIE JERSHA For Her**

### **Birthday**

Ms. Irene Jercha

### **MEMORY OF ELIZABETH JOHNSON**

Ms. Joyce McKay & Mr. Wiley Martin

M/M Lloyd Portis

M/M Gerald Hoyt

Ms. Nelda Mae Anderson

Judy Meyer & John Gray

Ms. Edna Kugelman

M/M Paul Silva

M/M Paul Keyser

Mr. Ernest Jackson

Ms. M. Ann Johnson

### **MEMORY OF ELAINE JOHNSTON**

Ms. Jennifer Thiebert

M/M Dennis Dahl

M/M Dean Wyse

### **MEMORY OF ASHLEY LYNN KIEFER**

M/M Wayne Kiefer

### **MEMORY OF BRETT KILLEN**

M/M John Boer

### **MEMORY OF JJ JONES MATA & HILLARY**

### **JONES MATA**

Mr. Kevin Baldwin

### **MEMORY OF STEPHANIE & ROBBIE MAXIM**

Ms. Madeline Stone

### **HONOR OF ETHAN BLAKE McKINNEY**

Ms. Barbara McKinney

### **HONOR OF ETHAN'S MOM, MINDY WALKER**

Ms. Barbara McKinney

### **HONOR OF ETHANS GRANNY, BARBARA**

### **McKINNEY**

Ms. Barbara McKinney

### **MEMORY OF BILLY MILANI**

M/M William Milani

### **HONOR OF JEFF MONTAVON'S 21<sup>ST</sup> Birthday!**

Ms. Margaret Gilmore

### **HONOR OF NORA MONTVILLE**

M/M Thomas Montville

### **HONOR OF SAM NIKOLAJEV**

Ms. Mary Oakes

### **MEMORY OF NATHEN OLIEN**

Ms. Marla Nelson

Ms. Lorean Hall

### **MEMORY OF MEGHAN O'NEIL & Honor of 50<sup>th</sup>**

### **Anniversary Of M/M EUGENE O'NEILL**

Ms. Sherran Vandemark

M/M Ronald Deutch

M/M James Reason

M/M Melvin Videto

M/M William Stippich

M/M Louis Wisniewski

M/M Charles Cornett

M/M Leo Strominger

M/M Jim Scott

M/M Alan King

M/M Charles Rothers

M/M William Dreyer

### **MEMORY OF SARA PFALLER**

Crystal Lake Lions Club

### **HONOR OF DAVID PFOHL**

Ms. Rachel Keppeler

### **HONOR OF MICK & SHIRLEY RAFKO'S 50<sup>th</sup>**

### **Anniversary**

Ms. Bonnie Thompson

### **MEMORY OF EARL ROY**

M/M James Ferguson

Mr. James Roy

M/M Douglas Roy

### **MEMORY OF SANDY ROYALTY &**

### **HONOR OF AMBER & SARAH ROYALTY**

M/M Mark Koenig

### **MEMORY OF JERED & MADISON SAVOY**

Ms. Jennifer Thiebert

### **HONOR OF MICHELE SHERIDAN**

M/M Casey Zaski

### **MEMORY OF KARLIE SKAVLE**

Mr. Thomas Noel Bnk West

### **HONOR OF KRISTIN SMITH**

Emerson Power Transmission

M/M William Murphy

### **HONOR OF MAKAYLA GRACE SMOAK**

Dorothy Puzio & Kevin Foster

Ms. Lillian Cilano

### **CSX:**

Andrew Woodgear

Roger Huber

Michael Lakel

Alan McCullough

Howard Haga

Ms. Mary Forseth

Ms. Andrea Allio

Ms. Peg Dicapua

Demore Construction, Inc

### **MEMORY OF THOMAS SMYSER**

M/M Charles Luebke

Mr. Victor Schmidt

M/M Dennis Bates

Avenue Rental, Inc.

### **MEMORY OF JOAN SPRIGG & NIKKI DAVIS**

Mr. Eugene Jones

M/M Thomas Ives

M/M Donald McCarney

### **MEMORY OF EMILIO SUTTI**

M/M Don Visconti

Ms. Betsy Peck

Ms. Carol Griffin

M/M Daniel Hynie

Ms. Ruth Hardin

Morgan Stanley

Bill Limneos

M/M Matthew Fortier

Anonymous

### **MEMORY OF EMILY THOMPSON'S**

### **BIRTHDAY**

Ms. Bonnie Thompson

### **MEMORY OF SARAH ROSE WHELTON**

Ms. Margaret Mitchell

### **HONOR OF IRENE WILHELM'S BIRTHDAY**

M/M Edward Hayhow

### **MEMORY OF EDNA WORSTER &**

### **HONOR OF CHELSEY WORSTER**

M/M Earle Bate

### **HONOR OF MADELINE ZELLMER**

Ms. Ruth Zellmer

### **FAMILY SERVICES & OPERATING**

Patricia Harrison

Charles Percy

### **RESEARCH**

Genactis, Inc.

Michael Uribe

Mr. Thomas Cornell

Riverview Baptist Church

Hayden's Batten Disease Foundation

Mr. & Mrs. Tongulen

Ms. Kimberly Fuller

### **STIPEND**

Anonymous



# *In Loving Memory*



## **MARK SZIGETVARI**

Son of Gabor & Marta Szigetvari, Budapest, Hungary  
Born: 10/24/95 - Died: 05/05/09 LINCL

## **SUE REYNOLDS**

Partner of Leslie Hudson, N. Hampton, MA  
Born: 04/18/75 - Died: 05/08/09 Kufs (Adult NCL)

## **SARAH WHELTON**

Daughter of Paul Whelton, Houston, TX  
Born: 11/05/98 - Died: 06/5/09 LINCL

## **TERELL AMOSAH**

Son of Philip & Tanya Amosah, Oxbow, Saskatchewan  
Born: 07/14/01 - Died: 06/13/09 LINCL

## **HILARY JONES**

Daughter of Bobby Jones and Fred & Deb Mata, Callahan, FL  
Born: 04-10-92 - Died: 07-28-09 JNCL

## **JANINE & TRACY BULLOCK**

Twin daughters of Marilyn Bullock, Thornlie, W. Australia  
Born: 01-26-78 - Died: 07-30-09 JNCL

## **STORM CARRETTA**

Son of Lynn Coll & Randy Carretta, Pittsburgh, PA  
Born: 01/06/04 - Died: 07/31/09 Cockayne's Disease

## **DAZLIN ALEXUS BATES**

Daughter of Roderick Bates & Denecra Bates, Nashville, TN  
Born: 10-01- 97 - Died: 08-05-09 INCL

## **ELISABETH JOHNSON**

Daughter of Keith & Alice Johnson, Nevada City, CA  
Born: 03-07-87 - Died: 08-17-09 JNCL

## **SETH HOLDER**

Son of Jocelyn Rowe & Stuart Holder, Mandurah, Australia  
Born: 10-10-02 - Died: 09-01-09 LINCL

## **FIELDING APPLGATE**

Son of William Applegate & Deborah Applegate, Watauga, TX  
Born: 07/01/92 - Died: 09/17/09 JNCL