



Back to School

Back to school time will be here before you know it!

Beginning a new school year can be a very exciting time. For some it may also cause anxiety. To alleviate any anxiety that beginning a new school year can bring, make sure you have all of the information that you need to make the transition as easy as possible.

FIRST is happy to provide you with the resources that you will need to make sure that your child's new teachers have all of the information necessary to understand how ichthyosis will affect your student.

Visit our website at www.firstskinfoundation.org and click on the *Back to School* link on our home page.

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Pathogenesis-Based Therapy Reverses Cutaneous Abnormalities in an Inherited Disorder of Distal Cholesterol Metabolism

Summary of article appearing in the *Journal of Investigative Dermatology* (2011), Volume 131

by Amy Paller, M.D., Prof. & Chair, Dermatology/Prof., Pediatrics, Northwestern University Medical School

Although the scaling that characterizes ichthyosis often occurs as the body's attempt to provide a better barrier against water loss and protection from the outside, peeling agents and moisturizers are the way most ichthyosis is treated. During the past decade or two, however, scientists have discovered the genetic and biochemical basis of most forms of ichthyosis. These discoveries provide an opportunity to use mechanism-targeted topical therapy. One form of ichthyosis, called Congenital Hemidysplasia with Ichthyosiform erythroderma and Limb Defects (CHILD) syndrome, is a disorder primarily in girls (X-linked dominant) in which there is an enzyme missing in the pathway towards production of cholesterol. Although most of the public thinks of cholesterol as unwanted, it is actually critical to the function of cells, including skin cells. In the case of CHILD syndrome, the combination of cholesterol deficiency and the accumulation of intermediates because of the missing enzyme leads to red, scaling skin and developmental defects (especially of limb formation). What is intriguing about CHILD syndrome is that the skin and bone changes tend to occur on only one side of the body ("lateralization"), and no one had previously tried to understand why this occurs.

Because of our understanding of the basis for the features of CHILD syndrome, we hypothesized that the combination of blocking the formation of the toxic intermediates by using a statin and adding back cholesterol to the skin could improve the skin condition. We formulated 2% lovastatin/2% cholesterol and applied it to

the skin of two affected women twice daily. Topical treatment with lovastatin/cholesterol (but not cholesterol alone) virtually cleared skin lesions by 3 months. Biopsies of treated skin showed that it had virtually normalized, as viewed both by routine microscopy and under the high power electron microscope. Both women were able to taper the use of the topical cholesterol/lovastatin to once or twice a week for maintenance and have remained relatively clear. This exciting result validates the use of "pathogenesis-based therapy" and suggests that this approach could be considered for other forms of ichthyosis, particularly those of lipid metabolism.



Amy Paller, M.D.

We also began to investigate why the skin and bone abnormalities occur only on one side of the body. We took biopsies of skin from areas with abnormal-appearing skin and from normal-appearing skin, and we grew cells in culture from the outer layer of skin (keratinocytes) and the deeper layer of skin (fibroblasts). We found that the DNA-based genetic change that leads to the missing enzyme occurs in every cell on both sides of the body. To understand the lateralization of CHILD syndrome, one first has to understand "functional mosaicism," a normal situation in girls. Females have two copies of the X chromosome, one from the mother and one from the father. In every female during the first weeks of life, a decision is made in each cell to turn one X chromosome on and turn the other

Continued on page 14

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The editor invites your correspondence. We welcome your comments, observations, and suggestions. Please send your letters to *Ichthyosis Focus* at the address listed above.

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Testimonial Dinner Honors Dr. Dennis Roop and his Ichthyosis Research

One hundred and fifty-five people joined FIRST in honoring Dennis R. Roop, PhD on Thursday, June 21 at the Denver Country Club. Dr. Roop and his team at the Charles C. Gates Center for Regenerative Medicine and Stem Cell Biology at the University of Colorado are making amazing research advances in Epidermolytic Ichthyosis (formerly known as EHK). FIRST was privileged to honor him amongst his family, friends, and colleagues. Although he was very humbled by this honor, his selfless goal was to raise funds to support FIRST, the only patient advocacy organization in the country devoted to helping individuals and families touched by ichthyosis.



Dave & Valerie Scholl, Dennis & Betty Roop, Joel & Beth Edelman, Jerry & Ann Winterrowd



Dr. Roop takes it all in.

Dr. Roop is an internationally recognized leader in skin disease research. His laboratory has had a long-standing interest in studying genetic pathways required for normal skin development and in the identification of genetic alterations that occur in inherited skin diseases and in acquired skin diseases. Dr. Roop has devoted a major part of his career to studying EI and is making novel advances using stem cell therapy.

The event raised over \$121,000, which included a live and silent auction for sports memorabilia, exotic trips, and handcrafted jewelry. Guests mingled to classic jazz entertainment provided by *Just Friends*, followed by dinner and the tribute to Dr. Roop. Dr. Moise Levy, Vice President of FIRST and a long-time friend and colleague of Dr. Roop, shared some wonderful and funny stories, and was followed by Mark Klafter, the father of Adam, a 10-year-old boy affected with EI. Mark spoke about how Dr. Roop's research is providing hope to his family and others affected by this skin disease. Next, Jerry Winterrowd, retired Episcopal Bishop of Colorado, paid tribute to Dr. Roop and his lab for using stem cell therapy for many other medical conditions. The evening ended with an amazing serenade of "Dream the Impossible Dream" sung a capella by Dr. David Norris, the chairman of Dermatology Department at UC Denver and long time friend of Dr. Roop.

Correspondence Corner



Dear FIRST,

I bought this cooling towel and it's the best thing I've ever seen for cooling down. Here is the website, <http://www.froggtoggs.com/#cooling/details/CP100/>.

I soaked it, wrung it out, and wore it like a scarf on my shoulders. It feels heavenly in hot weather.

I got mine at Dick's Sporting Goods and it was \$15.

I took it on my vacation to the Mediterranean and it saved my life (probably literally) on the walking tours. I can't say enough good things about this product. I think the company should use this stuff to make loose t-shirts.

Terry Melton
State College, PA

Dear Moureen,

Please accept this donation for the wonderful work your foundation is doing. Without your foundation, I might still be lost.

In December 2008, I was experiencing a lot of skin irritation. It hurt to wear clothing or take a shower and there was terrible itching. In February 2009, I had a skin biopsy and received a letter from my doctor saying I had Darier's disease and there was no cure. It was after hours and I couldn't call my doctor, so I immediately went to the Internet and learned all kinds of terrible things about this disease. When I called my doctor the next day to make an appointment for a consultation, the receptionist acted like I was stupid, but she made one for me anyway. The doctor was very gentle and kind, but when I checked out, the receptionist told me I took too long with the doctor. I left crying and I still had unanswered questions and not a lot of information. I felt alone and continued to rely on the Internet for all of my information.

For over two years, this consumed me because I was constantly looking for information on the Internet and trying oils, gels, and anything that seemed to help other people that I read about, so I wouldn't have to take steroids. I spent a lot of time crying, because of the pain or because of my unknown future, and I didn't know how to deal with it. I had absolutely no one who understood because no one ever heard about this disease. I was told that it was genetic, so I called both my children to let them know this was something they could pass on to their children and I felt so horrible that I could have passed this on to them.

In the summer of 2011, I took a day off work to search and make phone calls. I was determined to find someone who could give me answers and help put me in touch with anyone who knew of this disease. I happened across the FIRST Foundation. You answered my call and told me you knew exactly what Darier's disease was and knew what I was talking about. Not only that, you were going to send me some information and put me in touch with a doctor in Chicago who might be able to help me.

After talking to you, I immediately called the doctor in Chicago, who was skeptical about some of my symptoms and felt I was misdiagnosed. He said Darier's disease and Grover's disease are very similar under a microscope and felt it would be a good idea for me to see a doctor at Barnes/Jewish Hospital in St. Louis to re-test me. I made my appointment at Barnes/Jewish Hospital, and their lab work confirmed that I was, indeed, misdiagnosed and that I was suffering from Grover's disease, which is not genetic and much less serious than Darier's.

I still experience some painful breakouts, but not as bad. I do have some scarring, but I was able to wear a beautiful dress for my daughter's wedding in October and comfortably show some of my skin. I am so thankful every day that I found the FIRST Foundation. I've been reading many emails lately about meeting children who have ichthyosis. The pain I went through seems so small compared to what these children experience in their daily lives, not to mention the parents who are learning to deal with their conditions. It makes me feel guilty when I'm thankful for my misdiagnosis, and I want to do something to help your Foundation and the wonderful work you do for these families. I know many people who support cancer research or St. Jude's Children's Hospital, but ichthyosis is so rare that there doesn't seem to be much awareness about it. Your Foundation and the people you serve are close to my heart and I hope my small donation will help.

P.S. I have to apologize for taking so long to send this donation to you. Today is my birthday and my wish was to take the time to sit down to write this letter and send off my donation.

Sandy Paulus
Perry, Missouri



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Ichthyosis Awareness Month

FIRST celebrated Ichthyosis Awareness Month in May, and what a celebration it was!

This year, FIRST began a new awareness initiative. Several of our members were featured in an online campaign.

Nineteen people wrote their story or the story of their child affected by ichthyosis. The stories were then emailed, put on our facebook and twitter pages, and put on FIRST's website throughout the entire month. This idea was so well-received that we hope to continue with a monthly feature throughout the year! If you missed the stories and would like to read them, they are available on our website under Ichthyosis Awareness Month.

We kicked off the month with FIRST's message Ichthyosis Awareness – www.firstskinfoundation.org scrolling on the top of Philadelphia's PECO Building for 3 nights. This is the second year that ichthyosis awareness was seen in the nighttime skies over Philadelphia.



Lauren Hamill was one of those featured during FIRST's Ichthyosis Awareness Month email campaign.

FIRST Night at the Phillies Game



The Licursi family, from left, Ryan, Paul, Shelly, and Kevin

FIRST meeting at Citizens Bank Park to watch the Phillies take on the New York Mets. Everyone was welcomed at the Licursi family's pre-game tailgate to enjoy great food, fun and games, and mingling with each other. While the home team ultimately lost the game, it was still a great evening!

The McCoy family enjoy pre-game ice cream cones.

The festivities continued with our 2nd Annual Night at the Philadelphia Phillies. **Paul and Shelly Licursi** and their son **Ryan**, who is affected with EHK, led the charge selling 115 tickets to family and friends. Other members from around the Philadelphia area joined in the festivities, totaling 195 members of



Francine Mondini and her father, Nicholas brave the cold to support FIRST.



Awareness Blogging

Members across the country participated in their own awareness-raising campaigns. Many members have ichthyosis blogs and featured ichthyosis awareness on their blogs during the entire month. **DeDe Haggis**, featured FIRST on her wonderful blog "Our Young Warrior – Evan," where she writes about her adorable son, **Evan Fasciano** who is affected with Harlequin ichthyosis. **Susan Anadale**, whose young son **Peter** is affected with lamellar ichthyosis, featured ichthyosis and FIRST on her blog, "Homeschooling Hearts & Minds." **Courtney Westlake**, mother of beautiful **Brenna** who is affected with Harlequin ichthyosis, also featured Ichthyosis Awareness Month and FIRST on her blog, "Blessed by Brenna." In addition, FIRST member **Carly Findlay** from Australia posts regularly on her blog, "Tune Into Radio Carly." Carly is affected with Netherton syndrome.

It Makes Cents to Help FIRST Campaigns

Several members participated in the *"It Makes Cents to Help FIRST"* campaign that began last year. They went above and beyond with decorating canisters and collecting change from family, friends, and local businesses. Some of the highlights include:

DeDe Haggis is the mother of adorable 2-year-old **Evan Fasciano**, who was born with Harlequin ichthyosis. DeDe held her campaign in her hometown of Goshen, Connecticut. Her family, friends, and local businesses responded tremendously. DeDe's efforts raised more than \$4,000!

Jolie Cina, a wonderful advocate for FIRST, held a Penny Challenge at Bright Horizons pre-school in Roseland, New Jersey, where her children are students. Jolie decorated canisters with photos of her adorable children, **Portia** and **Myles**, and placed them in classrooms at the school. Students and faculty alike made donations and raised more than \$200 for FIRST.



DeDe and Evan with the many canisters that DeDe created.

Portia and Myles pose with the decorated canisters.

Girls Go Out in Georgia



Sam selects a raffle winner.

FIRST member **Sam Zavitz** and her husband **Brian** are the proud parents of 4 ½ -year-old **Cate** who is affected with lamellar ichthyosis.

Sam held an event for FIRST last May, called *High Heels & Handbags ~ a Girls' Night Out* at Glazed & Fired Pottery & Art Studio. It was such a terrific success, that she planned another event for this year's awareness month celebrations.

This year's event was even bigger and better than last year! There were more purses, more shoes, more attendees, more money raised, and much more awareness about ichthyosis and FIRST!

The event was held at the clubhouse of a local subdivision. Approximately 70 people attended this Girls' Night Out and purchased like-new handbags and shoes. Sam even created a permanent Facebook page for the event called High Heels & Handbags - Atlanta. An additional 35 people participated in the Facebook auction. There was also a

raffle with many great items donated for participants to win.

FIRST's Program Director, Moureen Wenik, happened to be in the Atlanta area for the Georgia Dermatology Physician's Assistants-Dermatology Pearls meeting. After the meeting, Moureen was able to attend Sam's event, which was a great way for the entire town of Cumming, Georgia to get to learn more about FIRST and ichthyosis.

Sam has a close group of friends who wanted to help her with the event. They were instrumental helping make the evening so successful. Stephanie Campbell donated her professional photography services for the event; Ashley Spenner handled all of the food; Marisa Cope was in charge of location and logistics; and Nicki Waits and Randi Arnett were instrumental in so many ways! Sam extends a huge thank you to these women who dedicated hours of their time because of their commitment to her family.

This event is becoming so well known in the Cumming area that Sam has already started receiving donations for the 2013 event! More people have volunteered to work on the executive committee for next year, and FIRST members in other states have reached out to learn how to run this event in their own state.



Sam, Brian, and Cate Zavitz greet Moureen before the event.

Collecting Donations and Creating Awareness in Michigan

Member *Nancy Osentoski* has been a long-time advocate for FIRST. She has held many awareness and fundraising activities over the years. This year, Nancy created an awareness poster and decorated canisters with FIRST information. Nancy displayed her information at the local Pamida's store in Imlay City, Michigan and spent the day encouraging people to donate and informing the public about ichthyosis and FIRST. Nancy's efforts paid off, raising more than \$500!



Nancy is prepared to offer information about ichthyosis and FIRST.



The Josh Ridings Band performs

Josh Ridings Band performs in South Carolina

FIRST member *Kitty Wall* and her husband *Jimmy* went to the Regional Conference in Richmond, Virginia last year. They left with a strong desire to help their son-in-law, *Trey*, and their granddaughters, *Addison* and *JoElla*, all of whom are affected with erythrokeratoderma variabilis (EKV). After much thought, the Walls decided to hold a benefit concert. Kitty's nephew, Josh Ridings, has a band and is the worship leader at a local New Testament Church. Grammy-winner Laura Storey sings back-up on one of the songs on the Josh

Ridings Band blog. Josh was very eager to help Kitty. Mountain View Church in Boiling Springs, South Carolina, where Kitty and Jim are members, graciously let the family host the concert. They made flyers and advertised in their community. More than \$2,300 was raised at the concert.

Chick-fil-A

Josh Ridings is also the Public Relations representative for Chick-fil-A in Hillcrest, South Carolina and suggested that the *Wall* family host a spirit night. More than \$100 was raised when family members and friends enjoyed their dinner at the Chick-fil-A on a Tuesday evening in May. Some friends and family members who could not attend either event, made donations, amounting to a grand total of \$200 raised that evening.



Trey, on left, and Jessica, right, join Josh Ridings at Chick-fil-A



Francine sings during the show.

More Than Skin Deep

FIRST member *Francine Mondy* is affected with lamellar ichthyosis. Francine received information about Ichthyosis Awareness Month and decided that she wanted to participate and create awareness.

Francine spent many hours coming up with a list of songs focusing on dealing with differences, overcoming adversity, and ultimately celebrating life. She wanted people to understand that there are many different types of issues out there, whether physical or emotional, but in the end we should be grateful to be alive and celebrate how lucky we are. Once she had the song list in place, Francine reached out to the Villagers Theatre

in Somerset, New Jersey about scheduling the event.

When people ask Francine about why she wanted to do the fundraiser, the reason is quite simple. "Over the years, I have been stared at and/or questioned about the way I look. In the past, I've ignored such ignorance. However, I felt it was time to start raising awareness so that someday in the future, others who suffer from this condition won't have to endure what I did."

Approximately 60 people attended the event. Early estimates are that Francine's efforts raised more than \$1,300 with the show itself and the direct donations that were collected.



Francine and her cast mates during the performance.

Congratulations 2012 UFIRST Scholars!

FIRST is proud to announce this year's five scholarship recipients from the UFIRST Scholars Program. Congratulations to everyone. We wish you the best of luck as you continue your education.



Bridget Smith

Ichthyosis Vulgaris

William Jessup University

Goals and aspirations:

"My goal at this time is to complete my Bachelor's Degree and obtain a career in counseling and spiritual coaching."



Mani Woodward

X-linked Ichthyosis

University of Oregon

Goals and aspirations:

"I hope to graduate from the U of O with a double major in Biology & Chinese. I then want to attend medical school to pursue a career as a pediatrician or a pediatric dermatologist."



Ashley Cloud

Ichthyosis Vulgaris

Agnes Scott College

Goals and aspirations:

"I want to make an impact in the world by studying anthropology and helping people to understand and accept other cultures."



Elizabeth Joyner

Ichthyosis Vulgaris

Union University

Goals and aspirations:

"To teach elementary school and encourage students that God made each one special and to strive to do their best."



Darcy Crawshaw

Lamellar Ichthyosis

Kansas State University

Goals and aspirations:

"To help others with anxiety and other debilitating psychological disorders."



UFIRST Scholars was established in 2010 with a seed gift from Valerie & David Scholl. The Scholls are grandparents to an affected granddaughter and wanted to provide an opportunity for affected students to advance their post-secondary education in partnership with FIRST. Their inspiration is to provide the opportunity for students affected with a form of ichthyosis to achieve their highest educational potential. Donations are graciously accepted to help grow the fund.

Applications are made available on February 1st of each year. Applications may be downloaded from FIRST's website, www.firstskinfooundation.org. Completed applications are due in March, with a specific deadline announced each year. Scholarship recipients are announced in May.

A committee of volunteers evaluates each scholarship application. The applications are scored using the following six criteria: 1) demonstrated academic ability; 2) a written essay (topics vary each year); 3) extracurricular activities/community activities; 4) financial need; 5) recommendation letters; and 6) involvement with FIRST.

NEWS ON THE HILL



On June 28, 2012, the U.S. Supreme Court caught many observers by surprise by upholding the sweeping health-care reform package signed into law in 2010. The health reform bill aims to provide health insurance coverage to an additional 32 million people. The law requires all Americans to purchase health insurance or pay a penalty to the Internal Revenue Service.

Twenty six states sued the federal government claiming that the so-called individual mandate was unconstitutional. The Supreme Court ruled that the individual mandate is a constitutional exercise of Congress' taxing authority. The Court also held that Congress could not withdraw existing Medicaid funds from states for failure to comply with the Affordable Care Act's expansion of Medicaid, but federal funding remains available to any state that affirms its willingness to participate.

So what does all this mean to FIRST? The existing law stands. Over the next few years, key provisions will continue to be implemented. By 2014, virtually all Americans will have to obtain coverage or pay a tax penalty. For a single person, the penalty will start at \$95 per year or up to 1 percent of income, whichever is greater. The penalty becomes progressively larger from 2014 through 2016, when it will reach full strength.

Medicaid coverage will stay the same for now. If you are living in a state that was offering Medicaid to a larger share of the poor than required by federal standards, the law has required your state to lock in those more generous eligibility rules through 2014. States will have to decide whether to participate in the law's expansion of Medicaid. If they choose not to participate, they could opt to scale back eligibility rules for the program.

Key provisions of the law are the following:

- the law forbids insurance companies from discriminating against patients for pre-existing conditions
- children under 26 years of age can stay on their parents' insurance
- many preventative screening tests must be covered by insurance with little to no co-pay
- insurance companies cannot use small loopholes in insurance contracts to cancel your plan if you become ill.



News on the Hill is a column to keep members current with the legislation in Washington, DC. This column is written by Angela Godby, Assistant Vice Chancellor for Federal Relations for the University of Texas System. She is affected with Lamellar/CIE.

FIRST NIGHT AT THE CHICAGO WHITE SOX

With the tremendous success of the Philadelphia Phillies and New York Mets baseball games, FIRST is reaching out to the Midwest with a Night at the Chicago White Sox.



Please join members of FIRST as the
WHITE SOX take on the KANSAS CITY ROYALS
on Wednesday, August 8
7:10 pm



The tickets are in section 162 and priced at \$34 each, with 50% of the sale price being donated back to FIRST.

For more information, please contact FIRST's office at 215.997.9400 or visit our website at www.firstskinfoundation.org.

THE RIGHT PLACE AT THE RIGHT TIME

Jean Pickford, Executive Director

Have you ever had a moment when you knew you were in the right place at the right time? One of those life events that was “meant to be”? Well, that’s what happened to Tracy McCaffery and her son, Ned, on a cold day in January in the hometown of our national office, Colmar, PA.



Tracy and Ned McCaffery

That particular morning, Ned, age 10, had a doctor’s appointment. They traveled from their home in Ambler, about two stops on the local train, to Colmar. Here’s where things get interesting.

As they are taking the 10-minute walk down Broad Street to the doctor’s office, they pass by a small sign in a window that reads, “FIRST – Foundation for Ichthyosis & Related Skin Types – 215.997.9400 – www.firstskinfoundation.org.” Tracy says, “Ned, you have ichthyosis! Let’s call them.”

As I sat in my office near the front of the building, I heard talking and laughing. This is quite unusual, because the only noise I ever hear from the busy street are cars, buses, and the occasional honk of a horn. Then the phone rings and a few minutes later, Moureen announces that we will be receiving two visitors in about an hour.

Tracy and Ned stopped in to visit FIRST on their walk back to the train station. What a win-win for both of us! We asked a lot of questions to find out their history. It turns out that Ned was diagnosed as a baby with ichthyosis vulgaris. His uncle has it too. He says his skin doesn’t bother him very much, but gets much worse in the winter. Tracy works for a local skin product entrepreneur, so she has a lot of knowledge about creams, lotions, and additives. Although Tracy had heard about FIRST a long time ago, life got in the way, and she never pursued getting in touch with us. Until this day...

And we loaded them up! We put together a package of materials, filled them in about the free Aquaphor program, the national family conference in Denver, and gave them an application for the Jane & Henry Bukaty Skin Care Fund, among other things. It was so nice to meet them and be able to make that connection. As they left our office, we all felt good about making new friends and helping Ned as he continues down life’s path with ichthyosis.

More Local Connections....

As I was writing this story about Ned and Tracy, I began thinking about other local connections that I’ve encountered over my years at FIRST. When I first started working here in 1999, my husband and I had a very close friend who was always lovingly teased in college as having “old man hands.” We were talking shortly after I started working for FIRST about how his skin used to affect him as he was growing up. He commented about how he hated gym class because he had to change clothes in front of his friends. He said he had to apply lotion on his skin every night and every morning after his shower or it would get really dry and flaky. He said his two brothers and a few uncles had it too. I told him he must have ichthyosis and he quickly said, “Icky-what?,” and blew me off. Of course, I mailed him some info a few days later. The following month he called me and said, “Jean, you were right. I have X-linked ichthyosis. I read your materials and that’s exactly me.”

How about the connection I wrote about last year? When Janet Burns went to the local Trader Joe’s and saw our thank you letter hanging on the community bulletin board near the restrooms? She and her brother both have ichthyosis vulgaris and never even heard of FIRST! Janet is now one of our best volunteers and donates her time in the office regularly.

A third connection was made when I moved into my new neighborhood. The neighbor from across the cul-de-sac came over one morning to introduce himself and say hello. As we were talking, the conversation came up about what I did as a profession. He stopped me immediately (as I can go on and on and on about FIRST) and said, “I have ichthyosis. It’s really mild so no one ever really knows. But I’ve had it all my life.” Of course, I put info in his mailbox the very next day.

And finally, last year, we made a local connection with Karen Tanner. Our local paper did a feature story about FIRST and our staff. Karen, who lives one town over from our national office, read the article over breakfast one morning. The article stuck out in her mind because she found it interesting that no one who worked for FIRST had ichthyosis or an affected relative, but they were working hard to help these families. Little did she know that six weeks later, her grandson would be born with ichthyosis in Pittsburgh. She immediately remembered FIRST and paid us a personal visit one afternoon in October. Now their whole family are members and strong advocates for FIRST.

In a world of a rare disease, the world is not so small after all!

Grassroots Fundraising

Another Beading Bonanza in Maine

Kim Mayone has once again held a very successful event at Caravan beads. Kim is the mother of 6-year-old *Evan*, who is affected with EHK. Several years ago, she began a fundraiser with her local Caravan Beads in Portland, Maine. This year was truly a family affair with Kim's son Harrison manning the information table that offered literature about FIRST. Daughter Sophia joined Kim and the staff at Caravan Beads assisting event attendees.

These fundraisers are great for Kim as they don't require a tremendous amount of time. The store donates 30% of all sales to FIRST and opens exclusively for Kim's event. This year was her best year yet, raising \$500!



Kim and Evan at Caravan Beads.

Jeans for Genes in North Carolina

Erin Edwards teaches at Ledford Middle School in Thomasville, North Carolina. The teachers made a donation to wear jeans to school. The campaign took place in support of "Wear Jeans for Genes: Rare Disease Day." The students joined in the fun by wearing jeans too! The day raised awareness about ichthyosis and \$500 for FIRST.



The students and teachers wearing their jeans.

ATOPALM™

We would like to introduce our ATOPALM™ patented skin barrier repair technology. To learn more about ATOPALM and its benefits, visit our website at www.atopalm.com.

We are offering a 40% discount on the purchase of any ATOPALM product...just enter the code **FIRST** when placing an order.

40% Discount



Ray Jubela displays his photography

GeneDx Sponsors Art Show

Staff members at GeneDx joined together to display their artistic talents and support FIRST. Board of Directors and Medical & Scientific Advisory Board member *Sherri Bale* is the managing director at GeneDx and very committed to FIRST. There was a large space in their building about to undergo renovations. Sherri asked Ray Jubela, the Accessions Supervisor and a budding photographer, if he would like to have a show in that space. He agreed and encouraged others to participate with him. GeneDx staff members and friends and family members with artistic talents ranging from photography, to painting, to pottery and metalwork joined the show. Mr. Jubela selected FIRST as the recipient of any funds raised through this evening. Some 50 people attended the evening and enjoyed light refreshments along with the variety of artwork displayed. It was a terrific evening of sharing, raising more than \$500 for FIRST.

Grassroots Fundraising

Beach Run & Walk in New Jersey

Ace in the Hole Foundation was founded by **Greg LiCalzi**, a FIRST member affected with lamellar ichthyosis who lost his twin brother, 1st Lt. Michael LiCalzi (“Mike”), in Iraq on May 11, 2006. Mike was born and raised in Garden City, NY, a proud graduate of Chaminade High School & the U.S. Naval Academy, and surfer at Lido West Beach. Mike was 24 years old and had only been in Iraq for six weeks when he died for our country. The Ace in the Hole Foundation, Inc. 501(c)3 is a charitable foundation established to raise, receive, administer, and disburse money and property for charitable, scientific, and educational purposes in the name and honor of 1st. Lt. Michael LiCalzi. The Ace in the Hole Foundation was founded in 2008. The term “Ace in the Hole” derives from Mike’s service with the 2nd Tank Battalion of the USMC. 2nd Tank’s slogan is “Ace in the Hole.”

On a beautiful Saturday in May, the Ace in the Hole Foundation held its 3rd Annual Beach Run & Walk. More than 400 people ranging from seasoned runners to families out for an afternoon stroll joined the festivities on Lido West Beach. This event raised money for several organizations in addition to FIRST. Proceeds were given to Marine Corps Tankers Association, which provides scholarships for Marine Tankers and their families. Huntington’s Disease Society of America and the Wounded Warrior Project were also recipients. FIRST’s portion of the funds raised totaled \$15,000! We are so grateful to the Ace in the Hole Foundation for this generous donation.



Greg LiCalzi presents FIRST Executive Director, Jean Pickford with the funds raised at the Ace in the Hole Event.



Pictured from left to right: Mark Van Deusen, Liz LiCalzi, Greg LiCalzi Sr., Carol LiCalzi, Luke LiCalzi, Greg LiCalzi Jr., Kim LiCalzi, Town Supervisor Kate Murray, Tom Montgomery, Donald Clavin

Teachers Unite in Florida

Olga Marrero’s daughter, Olivia, attended Marjory Stoneman Douglas School in Miami, Florida during her elementary years. Olga is also a teacher there. There is such a great relationship with the staff at the school that Olga went to her fellow teachers and the principal this year to ask them for a favor. Fourteen-year-old Olivia is affected with EHK. The teachers at the school participate in the United Way campaign. Olga asked those that don’t have a specific charity designated to consider designating FIRST for their donation. Mr. Rodriguez, the school principal, and 15 teachers joined together and collectively raised \$2,500. After the campaign, Olga treated the participants to a breakfast and Olivia and Olga personally thanked everyone. What a wonderful tribute to Olivia and her family that so many teachers responded so well to their plea. Olga wishes to extend special recognition to the people who donated on behalf of Olivia.

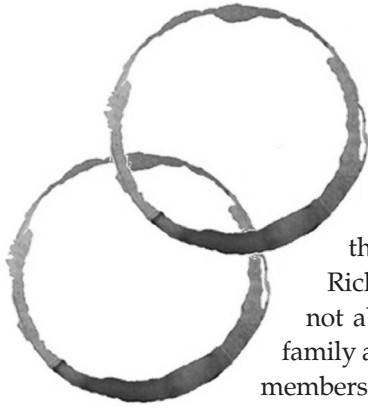
Elizabeth Alfonso
Ibis Bovo
Aimee Capote
Barbara Fernandez
Leslie Gonzalez
Patti Guadagno
Agustin Hart
Veronica Hart
Pablo Hernandez

Maria Jimenez
Rita Lopez-Phillips
Elena Martinez
Estrella Mellon
Olga Mena
Isbel Munoz
Todd Phillips
Ana Rodriguez
Rodolfo Rodriguez



Olga and Olivia thank the teachers and principal for their contributions.

Wine Tasting in New Jersey



Now a West Caldwell tradition, the **Cina family** hosted their 3rd annual Wine Tasting at their home. Sean and Jolie are the proud parents of adorable 5-year-old **Portia** and 2-year-old **Myles**, both of whom are affected with ichthyosis en confetti. The Cina family have been strong advocates for FIRST and ichthyosis awareness over the years. This year's wine tasting was a terrific success with more than 25 attendees sampling the wine, and enjoying each other's company and the wonderful hors d'oeuvres. Senator Richard Codey, whom Jolie has cultivated an advocacy relationship with over the years, was not able to attend this year, but he continues to be a strong advocate on behalf of the Cina family and FIRST. In addition to those who attended the evening, many other friends and family members of Sean and Jolie made donations to FIRST to support the event.

Dinner and Silent Auction in California

George and Robyn Swan are the parents of beautiful, 2-year-old **Geordyn Lily Swan** who is affected with recessive congenital ichthyosis. The Swan family hosted a benefit dinner and silent auction in Santa Rosa, California to raise money and awareness for Ichthyosis Awareness Month and for FIRST. La Rosa Tequileria & Grill donated the space for a cocktail party, attended by more than 90 people. There were appetizers and a silent auction, which featured two trips to Mexico; an overnight trip for 4 was also raffled off. The restaurant also hosted a dine and donate for guests in the main dining room. A local artist, Robert Herrera, entertained the crowd by playing the guitar and singing. After the cocktail party, participants enjoyed music on the outside patio on the warm May night. More than \$10,000 was raised for FIRST on this wonderful evening of connection and awareness.



Robyn, center, with her mother and sister, left and right respectively.



Geordyn being held by her daddy, George.

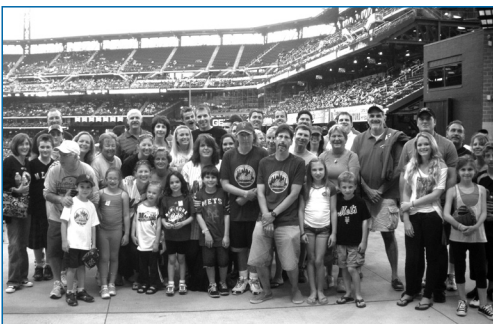
FIRST Night at the New York Mets



The Benedetto family and the LiCalzis accept the Spirit Award from the Mets.

Ichthyosis Awareness Month wrapped up on a high note with 220 New York-area members joining in the fun as the New York Mets hosted the Philadelphia Phillies. The evening kicked off with the presentation of a Spirit Award to FIRST. Members on hand to receive the award were the **Benedetto family**, Marc, Denise, Marc Jr., and Eric. Young Marc is affected with lamellar ichthyosis. Also present on the field to accept the award were **Greg LiCalzi**, who is also affected with lamellar ichthyosis, and his wife, Kim. **Denise Benedetto** led the charge, selling tickets to more than 100 of her friends and family members. Member **Brooke Saccente**, whose daughter **Kylie** is affected with an unknown type of ichthyosis, also sold tickets to 80 of her friends and

family members. The Saccente group even had t-shirts created for the evening. The festive purple shirts featured Friends of Kylie on them and made it very easy to spot the Saccente group at the game. While the home-team Mets lost their game, it was still a fantastic evening of community and awareness-raising.



Denise Benedetto and her friends and family members enjoy the game.



Friends of Kylie Saccente show their support.

National Family Conference

FIRST's 17th Biennial National Family Conference was held in Denver, Colorado over the weekend of June 22-24. More than 300 people representing 102 families, from 35 states and seven countries, participated in the weekend events, highlighted by the Disorders Of Keratinization Project conducted by Dr. Keith Choate and the Yale University team. The goals of this study are to identify the genetic causes of ichthyosis and to understand how mutations cause skin disease. The Yale team enrolled 84 conference participants in the study, which took place all day on Friday of the conference weekend.

Registrants from as far as Australia, Ireland, and Zambia checked in on Friday morning and had the opportunity to see the friends that they haven't seen since the Orlando Conference, renew old friendships, and make new connections. The conference activities kicked off with the Opening Session. Veteran conference attendees shared their experiences with the crowd, almost half of which were first-time conference attendees. They offered information and ideas for what to expect over the course of the weekend. The atmosphere was then relaxed while everyone participated in an ice-breaker activity to help people get to know each other. After lunch, participants enjoyed workshops about lotion tips, navigating the school system, and a highlight, a workshop on diet, nutrition and exercise facilitated by long-time FIRST member and conference veteran Paula Heinzman Ryan.

After enjoying a Friday evening on their own to see Denver or just relax, everyone was back together Saturday morning to hear the latest on the gene discoveries and research that are taking place at Yale University. Drs. Leonard Milstone and Keith Choate shared the exciting work being done by the group of researchers at Yale. Attendees then learned of grassroots fundraising opportunities from a panel of volunteers active with FIRST and veteran grassroots coordinators.

After lunch, registrants took part in more workshops, had the opportunity to ask questions from members of FIRST's knowledgeable Medical & Scientific Advisory Board, and shared their experiences during the group networking sessions.

During Saturday afternoon, the teens were treated to a field trip to Denver's Museum of Nature and Science. They enjoyed the lizards and snakes exhibit, the exhibits of Africa, Australia, and many other regions, the Mummy exhibit, the Dinosaur exhibit, a Tornado Alley movie in the IMAX theater, and the black hole presentation in the planetarium.

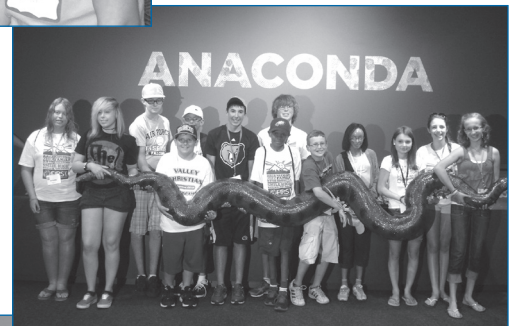
Once again, the highlight of the weekend was the Saturday night dance party. This year's party was, without a doubt, the best one yet. Each year, more and more youngsters show their courage and share their talents with the audience. With wonderful performances from Ema Hodgkinson, Abigail Mast, Rylee Iott and her friends, Portia Cina, Aklya



"I really enjoyed being able to not worry about the stares. I also really liked having doctors here who we didn't have to educate about my condition; they already knew what I had."



"FIRST does an excellent job. The physicians that are here are excellent, as well as the faculty and advisors."



"I most enjoyed networking with others and realizing how important it is that our population interact collectively with the researchers so that connections toward research can be made."



Photo by Meredith Rizzo

Photo by Meredith Rizzo



Franklin, Treasure White, and Sama Mulhern, our children showed their talents and their wonderful spirit. Taylor White rocked the house with a wonderful performance of *Bound to You* by Christina Aguilera from the movie *Burlesque*. Grace Ryan answered the audience demands and performed an astounding rendition of *Beautiful* that brought the crowd to its

feet. Everyone danced, played games, and eagerly anticipated hearing their names for the raffle drawings. One and all had such a great time that they lingered in the ballroom long after the DJ left, for no one was ready for the evening to end.

The conference ended on Sunday afternoon after disease-type workshops and focus groups. Everyone gathered over lunch to reminisce about the weekend, share phone numbers and emails, promise to keep connected, and look forward to seeing each other again in 2014. The powerful emotions and the strong bonds that were created will last a lifetime.

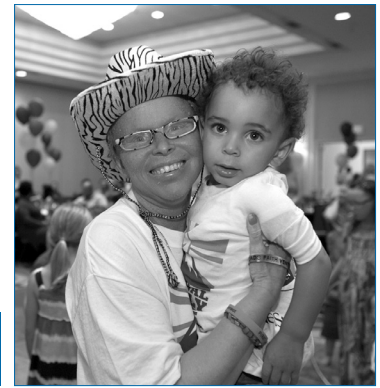


Photo by Meredith Rizzo

Pathogenesis-Based Therapy *Continued from page 1*

X chromosome off. From that time on, as each cell divides to populate the skin and other organs, it retains the original code that dictates which X chromosome is turned on and which is turned off. Normally, that early decision to express the genes from one X chromosome or the other in a given cell is a random one, leading to a random pattern. Because both X chromosomes usually express normal gene products, we don't usually notice the difference. When the product of one X chromosome is abnormal because of a gene mutation that affects a skin cell, however, it may manifest as a patterned skin abnormality. Chondrodysplasia punctata or Conradi's syndrome is a form of ichthyosis in which the gene change occurs on the X chromosome and stripes of skin changes can be seen in a random distribution. The gene that is mutated in CHILD syndrome is also located on the X chromosome, and thus affected females have one normal and one mutant X chromosome. However, the skin (and bone) change in CHILD syndrome does not present in expected stripes in a random distribution on both sides of the body, suggesting that the X chromosome inactivation is not randomly distributed. Indeed, when we examined the blood cells of one of our patients, we found that almost 90% of the cells showed activation of the normal X chromosome. This situation provides evidence of a selective disadvantage to cells with activation of the abnormal gene. When we examined keratinocytes from the ichthyotic skin, we found activation only of the abnormal X chromosome; in keratinocytes from unaffected skin, we found activation of only the normal X chromosome.

While we do not yet understand the mechanism by which abnormal skin and bone cells are cleared from one side of the body, but persist on the other side in the developing embryo, further studies could both explain the lateralization of CHILD syndrome and help us to understand normal spatial organization in developing babies.

Conference 2012

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Photo by Meredith Rizzo



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REGIONAL MEETINGS ARE PLANNED FOR 2013

Locations are:

North Jersey | Columbus, Ohio | Kansas City, Missouri
Dallas, Texas | Los Angeles, California

Dates and details will be posted on our website,
www.firstskinfoundation.org, when they are confirmed.





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Combined Federal Campaign (CFC)