FIRST
Foundation for Ichthyosis & Related Skin Types™
Educate • Inspire • Connect

Ichthyosis FOCUS

Fall 2011 A Quarterly Journal for Friends of FIRST Volume 30, No. 2

Start Planning to Attend the

2012 NATIONAL FAMILY CONFERENCE

FIRST's 17th bi-ennial national conference will be held in beautiful Denver, Colorado at the Renaissance Denver Hotel. Please plan to join us on ***June 22, 23, and 24*** for our signature event! Registration information will be printed in our next issue and posted on our website www.firstskinfoundation.org. ***Mark your calendar today.***

INSIDE Ichthyosis Focus...

FIRST RESEARCH PROGRAM UPDATE

Long-term Faithful Recapitulation of Transglutaminase 1-Deficient Lamellar Ichthyosis in a Skin-Humanized Mouse Model

Below is the update from Heiko Traupe, MD on his project which received funding through FIRST's Research Grant Program.

By Heiko Traupe, MD and Karin Aufenvenne, PhD
University Hospital Münster, Department of Dermatology

Heiko Traupe, MD

Transglutaminase 1 is a crucial epidermal enzyme. It facilitates the formation of the epidermal barrier, which prevents dehydration due to the loss of body water, poisoning from the absorption of noxious substances, and systemic infection from invading surface microorganisms.

Loss of transglutaminase 1-activity results in the severe genetic skin disease lamellar ichthyosis (LI). Most patients with transglutaminase 1-deficiency exhibit a life long pronounced scaling with increased transepidermal water loss (TEWL). Additional complications include episodes of sepsis, fluid and electrolyte imbalances due to impaired skin barrier function, and failure to thrive, especially during neonatal period and infancy. Transglutaminase 1-deficient LI still is associated with increased mortality in the neonatal period and has a dramatic impact on quality of life. No efficient treatment is available; current therapy only relieves some symptoms.

In vivo studies in human skin are limited by ethical and practical considerations. Although an animal model of transglutaminase 1-deficiency exists, it does not recapitulate the human disease and presents several drawbacks. *Tgm1* knockout mice lack clinically visible hyperkeratosis and display an extreme impairment of the epidermal barrier so that TEWL is dramatically increased. The fact that these mice die 4-5 hours after birth due to impaired barrier function is the strongest validation of the importance of transglutaminase 1 for CE-formation.

Our work in the project supported by FIRST has had the aim to develop a stable, long-lived pre-clinical skin-humanized mouse model which accurately recapitulates the human skin phenotype. Using previously established optimized tissue engineering and surgical conditions enabling stable human skin engraftment in immune-deficient mice, we succeeded in developing a robust humanized model of transglutaminase 1-deficient lamellar ichthyosis.

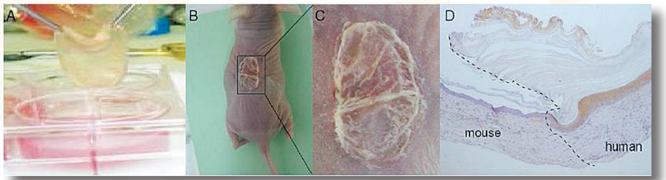

Fig. 1: Phenotype and histology of the skin-humanized mouse model for transglutaminase 1 deficiency.

A: Bioengineered skin equivalents prepared with previously isolated keratinocytes and fibroblasts from patients suffering from transglutaminase 1-deficiency; B,C: athymic nude mouse with graft eight weeks post grafting; D: staining with an antibody specific for human involucrin showing the humanized skin graft. Furthermore, in comparison to the mouse skin, the graft shows a very thick and packed stratum corneum.

Punch biopsies obtained from two patients suffering from transglutaminase-deficiency were used to isolate keratinocytes and fibroblasts. Together with the working group of Dr. F. Larcher (Madrid, Spain) we prepared so-called "bioengineered skin

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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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DR. LEONARD MILSTONE HONORED

FIRST Board member emeritus & Medical and Scientific Advisory Board Chairman Leonard Milstone, MD was honored by FIRST at a Testimonial Dinner. The dinner was held on Friday evening, July 8 at the Top of the World Observation Deck in Baltimore, Maryland. Baltimore was hosting the 37th Annual Meeting of the Society for Pediatric Dermatology (SPD). Close to 100 friends, colleagues, and family members attended this memorable evening which raised more than $45,000 to support FIRST's research fund.

Jean Pickford and FIRST President Dave Scholl present Dr. Milstone with a token of appreciation from FIRST

The evening featured tributes by family, friends, and colleagues of Dr. Milstone, including Anne Lucky, Lowell Goldsmith, Mary Williams, Amy Paller, Elena Levitan, Keith Choate, Len's son Aaron Milstone, and FIRST President Dave Scholl.

Dr. Milstone, a dermatologist with Yale Medical group, was invited to become involved with FIRST in 1981, when it was still known as the National Ichthyosis Foundation (NIF). He served on the medical board for seven years and then was asked to be the chairperson, which he has remained ever since. Dr. Milstone served on FIRST's Board of Directors from 1997 until 2006, when he was asked to remain as a board member emeritus, a lifetime position. Currently, Dr. Milstone serves on the Advisory Committee for the National Registry for Ichthyosis & Related Disorders and is the Chairperson of FIRST's Research Grant Committee.

From left: Keith Choate, Leonard Milstone, Jean Pickford, and Brian See enjoy the evening

FIRST is very grateful to Dr. Milstone. As one of the original devoted dermatologists who founded, nurtured, and has grown FIRST to what it is today, he is so deserving of this honor. It was a privilege to honor this man who has held many leadership positions within our organization over the past thirty years and devoted a major part of his career to studying and treating patients with all forms of ichthyosis.

Board members Ken & Elena Levitan and Janet & John McCoy joined the festivities

Ellen Clemmer (left) and Jean O'Brien congratulate Dr. Milstone

NEWS ON THE HILL

DEFICIT REDUCTION DOMINATES DC

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.

It was a long hot summer in Washington, DC as Congress and the White House negotiated a deficit reduction deal to avert financial catastrophe. Without action, the federal government faced imminent default on this country's borrowing authority. Amidst unprecedented partisan wrangling, the White House and Congress put the federal government on a path to reduce the federal deficit while also reducing federal spending.

In August, the President signed the Budget Control Act of 2011. The bill established a bipartisan, bicameral committee of twelve legislators. This group is charged with the daunting task of coming up with a plan to reduce federal spending by at least $1.2 trillion over the next ten years. The so-called "Super Committee" has a very quick timeline – any plan they develop must be signed into law by the end of the year. If the committee fails to come up with a plan that can pass both houses of Congress and be signed into law, then automatic, across-the-board cuts are triggered.

What does this mean to you? First, almost everything is on the table for cuts. Medicaid, Social Security, and veterans' benefits are protected, but Medicare and the National Institutes of Health (NIH) are not. Medicare would face at least a 2% cut across the board if automatic cuts are put in place, and potentially more from a Super Committee plan. While the NIH is supported by many members of Congress, when the budget will be slashed by cuts of this magnitude, there is no doubt that the pain will be felt everywhere.

What can you do? Continue to educate your member of Congress and the White House about ichthyosis. Using your personal example, share why medical research is so important to ichthyosis patients as well as other rare conditions. Visit the FIRST website at www.firstskinfoundation.org using the "Advocacy" tab under "Research and Support" to learn more about how to help.

DEFICIT REDUCTION
"Super Committee"

Democrats	*Republicans*
HOUSE	
Xavier Becerra (CA)	Dave Camp (MI)
James Clyburn (SC)	Jeb Hensarling (TX)
Chris Van Hollen (MD)	Fred Upton (MI)
SENATE	
Max Baucus (MT)	Jon Kyl (AZ)
John Kerry (MA)	Rob Portman (OH)
Patty Murray (WA)	Pat Toomey (PA)

It Makes Cents to Help FIRST!

During Ichthyosis Awareness month in May, FIRST launched a campaign called ***It Makes Cents to Help FIRST.*** The premise of the campaign was to have friends and family members decorate canisters with information about FIRST, and perhaps a photo of your affected loved one, and then place the canisters in your community or school to collect change. This was both a great tool for generating awareness and also raising funds. Many of our members and their families participated, increasing the discussion of ichthyosis in their communities.

Denise Benedetto leads a group of students in the Winner's Walk

Denise Benedetto, mother of 14-year-old **Marc,** from New York, combined the change campaign at her school, Francis X. Hegarty School, with the school's annual Winner's Walk. The Physical Education Department and the Student Council joined forces to raise money for a charity. This year, during May, the school chose FIRST. Denise went into school to educate the students about ichthyosis and placed coin collection buckets in each classroom. The walk was held as the culminating activity after the fundraising. Music was provided by the school music teacher who became a DJ, and the students walked or jogged around the school yard course for about an hour. After the walk, the students gathered together to congratulate each other for a job well done.

Lani Coates is the mother of 5-year-old **Sophia** who is affected with EHK. **Sophia** attends the Montessori Children's House school in Cypress, California. Lani's husband, Roland, who is currently deployed in Afghanistan, emailed the principal of the school, Dr. Anne Perrah, about participating in the change campaign. Dr. Perrah enthusiastically supported this campaign. A bear-shaped container was set up in the classroom and the children were challenged to "stuff the bear." In addition to collecting change, FIRST literature was distributed and the children and parents were educated about ichthyosis and how it affects Sophia's routine.

FIRST awarded prizes to the member who raised the most money, and also for the member who raised more than $100.

Sophia with Dr. Perrah and the bear used for collection

Congratulations to Denise Benedetto for collecting the most change and to the Cina family for their efforts. Both winners will receive a voucher toward the 2012 National Family Conference.

Executive Director's Report

Dear Members and Friends of FIRST,

It's hard to believe that the summer is coming to an end and we will be approaching cooler temperatures and fall weather. That means most of you will experience changes in your skin. If you're looking for new products, remember that FIRST has an extensive list of products that has been compiled over the years from suggestions from other members. A copy can be made available to you by contacting the office.

The fall will also bring our Board of Directors together for our bi-ennial retreat. We will be meeting in Philadelphia in early November to adopt our new three-year strategic plan. This plan is our roadmap for the future. Our leadership determines the most important areas of focus to keep FIRST strong and meeting the needs of our community. If you have any ideas or suggestions, please feel free to contact the office and share them. FIRST is in business to help YOU, so your input is key to our success.

FIRST operates on a fiscal year calendar, so our books will be closing at the end of September for 2011. I know the economy is still recovering, so our donations are lower than in recent years. Typically FIRST solicits gifts from our entire database three times per year: 1) annual appeal/membership (spring); 2) research campaign (early fall); and 3) end-of-year/holiday mailing (December). Please consider any size gift to support any or all of these campaigns in the upcoming year. Remember, even the smallest gifts add up!

In the next few months, our Marketing Committee will be working on reaching out to dermatologists, pediatricians, and other medical professionals to promote FIRST and the services we can offer to their patients. Thank you to everyone who responded with the names of your doctors. We've been able to expand our Physician Referral Service and will be including these medical professionals in our future communications.

The National Family Conference in Denver will be here before we know it! We are busy making plans for all the attendees and setting up our program. We've received lots of great ideas from our members who attended our four regional meetings this year in Seattle, Richmond, Atlanta, and Chicago. These regionals were a great way to meet local families, interact with local dermatologists, and exchange ideas and support. The national conference will continue to offer that same experience–just on a larger scale. I hope you are planning to attend! Check our website in the next few months for more details and registration.

Best wishes for a great fall season!

Yours truly,

Jean

Jean Pickford, *Executive Director*

Thanks to FIRST's grant from the Lennox Foundation, we have the opportunity to place advertisements in the Society for Pediatric Dermatology quarterly journal. These advertisements will provide information to pediatric dermatologists about what FIRST can offer to them and their patients. This ad can be used for multiple audiences. Let us know if you have an idea or lead for more awareness.

Eee our new ad on page 15!

Ichthyosis and the Workplace

Ichthyosis and the related skin types can raise difficult issues in the workplace. Communication is the key: whether it is letting co-workers and managers know about the disorder, or preparing the work environment to ease your disability and increase your productivity.

Like friend and family relationships, work relationships require a certain amount of communication and honesty. Co-workers may be curious about your skin, and employers may wonder how your ichthyosis might affect your work. Because of this, it is important for people with ichthyosis to understand their disorder, their employment rights, and the resources available in the workplace.

The law with which people are most familiar is the Americans With Disabilities Act of 1990 (ADA), which was amended in 2008. Title 1 of the ADA covers employment. Since 1994, it has required that employers of more than 15 people must make reasonable accommodations that allow a qualified job applicant with a disability to complete the application process or a disabled employee to carry out the duties of his or her job. According to the Americans with Disabilities Act, "an individual is considered to have a disability if he or she has a physical or mental impairment that substantially limits one or more major life activities, has a record of such an impairment, or is regarded as having such an impairment."

It is illegal to require a job candidate to take a medical examination prior to a job offering. The employer cannot try to ascertain whether a job candidate has a disability. Therefore it is sometimes up to the employee to decide whether to disclose his or her disability to the employer. If your ichthyosis is mild and not readily apparent, it is your choice if you want to disclose it. However, while one may have reasons for keeping a disability a secret from an employer, revealing it may require the employer to provide certain accommodations that will allow you to perform better at your job.

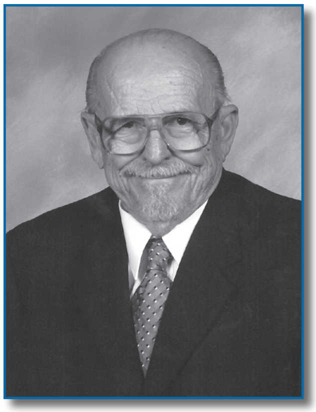
• • • Tips for Workplace Communication • • •

1. Plan ahead. Plan carefully how and when to discuss your ichthyosis with co-workers and supervisors. Education about ichthyosis and its symptoms can help change their perceptions and expectations of people with ichthyosis. Begin by researching all the changes that could make your job as productive as possible.
2. Meet with your supervisor. Schedule a meeting with your supervisor at a time when neither of you is under pressure. Describe simply and plainly the ways your ichthyosis may affect your work (stiffness, dry eyes, overheating, etc.) The goal is not to generate sympathy, but to find ways to solve the problem that will benefit the company, your co-workers, and yourself.
3. Offer alternatives. Be prepared to offer suggestions for possible changes, such as using assistive devices or accommodations (air conditioner, private area to apply lotions, hand-held vacuum to remove skin scales, etc.) These assistive devices and accommodations are items you may need to help you do your job more easily. Chances are any changes you may need will not cost much. Tax deductions and/or tax credit may be available to certain employers who provide accommodations and/or jobs for people with disabilities.
4. Communicate with your co-workers. Understand that co-workers can become resentful if they feel you are not doing your share of the work. Explain to them how ichthyosis affects your body and may, at times, affect your ability to work, but that you are making reasonable accommodations.
5. Listen to your body. You may be tempted to "work through the pain" of ichthyosis, but overdoing it may cause exhaustion, overheating, increased risk of infection, and dehydration. Instead, set your priorities and pace yourself. List your tasks in order of importance and do the most important ones while you feel strongest and most energetic.
6. Create a healthy work environment. Arrange your workday or work area to limit the amount of lifting, carrying, holding, or walking. Vary activities to avoid sitting in one position or repeating one action too long. Go to bed at a regular time and get enough rest to carry you through the next day.
7. Develop a support network. Other people in the workplace may have physical disabilities and can offer support and understanding. Establish friendships with co-workers whom you can trust. If you are not feeling well or are having a difficult time with a particular issue or co-worker, take a break and talk it out with a trusted co-worker. Just having someone to listen can make you feel better and change your perspective on the situation.
8. Maintain a positive attitude. Remember that you are allowed to have low-energy days, but that you are in control of how you relate to the disorder. Share your thoughts with other people with ichthyosis through FIRST's Regional Support Network. Contact the national office at 800.545.3286 or email info@firstskinfoundation.org

In Memoriam

Henry "Harry" Bukaty

September 6, 1926 - June 13, 2011



It is with great sadness that we announce the passing of Harry Bukaty, a long-time friend of FIRST.

Harry was born in Kansas City, Kansas on September 6, 1926, the sixth of seven children born to Daniel and Antoinette Bukaty. After graduating from high school, he enlisted in the US Navy. He served on Guam as a supply clerk during World War II. After the war, he moved to Los Angeles where he attended Compton Community College and later Cal State Los Angeles, majoring in business.

In 1953, Harry met Jane Webster at a Bastille Day Celebration. They were married the following year. The next year, the couple bought their home in the brand-new community of La Mirada. In 1956, their only daughter, Denise, was born.

In 1961, Harry began working as a salesman for Ernest Paper Products (now known as Ernest Packaging Solutions,) where he would remain for the next fifty years. His passion for helping people, problem solving, and anticipating his customer's needs helped him to become an outstanding salesman.

Harry was a devoted husband, father, and grandfather. During their 37 years of marriage, Harry and Jane loved to travel, spend time with friends and family, and go out for a nice dinner. Denise, as a child, would wait at the sliding glass door every afternoon for her daddy to come home from work and play with her. As an adult, she continued to look forward to their Thursday lunches. Harry and Denise's husband, Sky, shared a strong bond. Grandpa Harry also loved to shower his grandchildren with his affection. They especially enjoyed the annual trips to Universal Studios.

In addition to his love for his family, Harry was a very generous and established member of his community. He made friends everywhere he went and was well-known for his kindness, helpfulness, and his sense of humor. Although he was pretty good at keeping a straight face, the twinkle in his eye was the clue that he was "just pulling your leg."

Harry became actively involved with FIRST after the birth of his three grandchildren, all of whom are affected with lamellar ichthyosis. He was very passionate in his dealings with FIRST, and the staff enjoyed chatting with him during his regular calls to the office. Harry was an ardent supporter of the Foundation and our mission.

After two difficult months following a broken hip, Harry passed peacefully in his La Mirada home on the morning of June 13. He is survived by his daughter Denise, her husband Sky, and their children Schuyler, Jr. "Seej," Jalene, and Tamara.

JANE & HENRY BUKATY SKIN CARE FUND

Thanks to the generosity of Jane and Henry Bukaty, the foundation has established the Jane & Henry Bukaty Skin Care Fund to help alleviate some of the financial burden that may be facing our members. Here's your opportunity to apply for some financial assistance for ichthyosis treatment. Since the fund is limited, the following criteria must be met by the applicant in order to be eligible for a grant.

The applicant must be registered in our database and is required to submit an application indicating his/her need for funding. The application can be downloaded from FIRST's website. The specific product/treatment for which funds are needed, and a demonstration of the financial need for this product/treatment.

Awards will not exceed $200.00. Applications will be awarded two times per year as determined by the Review Committee. Applicants will be eligible to receive one award every two years.

Requests can be downloaded from FIRST's website and emailed to the national office at jpickford@firstskinfoundation.org, faxed to 215.997.9403, or mailed to the attention of:

Foundation for Ichthyosis & Related Skin Types, Inc.™
Jane & Henry Bukaty Skin Care Fund
2616 N. Broad Street, Colmar, PA 18915

You will be contacted by the office if you have been awarded aid from this fund.

Regional Meetings Continue to Bring Families Together

by Moureen Wenik, Program and Research Director

This summer, members in regions 3, 4, and 7 had the opportunity to come together for education and networking. The meetings were met with much success, as the attendees shared product information, told stories, and heard presentations from leading dermatologists.

Participants at the Region 7 meeting

Drs. Branding and Sybert address the Seattle group

Region 7 Meeting Held in Seattle

The **Region 7** meeting was held on June 11, 2011 in Seattle, Washington with University of Washington's Dr. Virginia Sybert and Dr. Heather Brandling-Bennett hosting the morning session. Their interactive genetics presentation and candid discussion made this meeting enjoyable. Families who attended this meeting traveled from Washington, Utah, Oregon, and California. The parent breakout session brought much needed discussion, as well as reassurance to the parents in the group, while the adults met and shared some of the challenges they face.

Participants share ideas at the Region 7 meeting

Participants listen to Dr. Sybert in Seattle

All of the attendees at the Region 3 meeting

Region 3 Meeting Held in Atlanta

The meeting in **Region 3** was hosted in Atlanta, Georgia on July 16, 2011 with Drs. Mary Spraker and Leslie Lawley serving as medical speakers for this event. This was a well-attended meeting with participants from Tennessee, Mississippi, Georgia, and Florida. This meeting was also filled with wonderful medical information for the participants, lively discussions for the parents and adults, and recommendations from fellow meeting attendees on the products they like to use.

Many of the meeting participants attending the regional events have never been to a FIRST event prior to coming to one of these meetings. These smaller meetings are a great introduction for these members, and a wonderful introduction to FIRST. The regional meetings give first-time attendees the understanding that they are not alone and that there are others willing to share advice and stories.

Steve Gondusky and Paul Foulks at the Region 3 meeting

Newlyweds Amanda Joy and Todd Houston

Region 4 Meeting Held in Chicago


Chicago meeting participants listen to Dr. Amy Paller's presentation

The 2011 Regional Meetings came to a close with the Region 4 meeting held on September 10, 2011. This Chicago meeting was well attended, with 58 people in attendance including 7 doctors from Northwestern University and Dr. Jennifer Hand from the Mayo Clinic in Rochester, MN. Dr. Amy Paller spoke in the morning session with a general overview on ichthyosis, the genetics of the skin disease, and skin treatment options. The afternoon included product sharing among meeting attendees and a breakout for the parents and adults. The children attending the meeting spent the day in the childcare room having a great time meeting new friends. The team from Northwestern came prepared to draw blood from families interested in participating in Dr. Keith Choate's study that aims to discover genetic causes of ichthyosis. Families filled the room to take part in this opportunity.

The states represented at this meeting included Illinois, Indiana, Kentucky, Michigan, Minnesota, and Ohio! Half of those attending the regional meeting had never been to a FIRST event before, making this their very first event. Other meeting participants have been involved with FIRST since they were born! The day brought great stories, much laughter, and a new connection to those who attended. If you missed a regional meeting in 2011, consider attending the Family Conference, June 22-24, 2012 in Denver, Colorado. Coming together in a face-to-face meeting with others is a wonderful way to learn more about ichthyosis and how to manage your skin.


Ben Osowski enjoys lunch


Holly Friddle (left) and Isabelle Townsend chat between sessions


Frank Osowski, Tom Buehler, and Steve Flury connect at the Chicago meeting

ORGANIZE A GRASSROOTS EVENT

You can help FIRST by hosting a fundraiser. Many of our members have hosted successful grassroots events, and are very willing to share their knowledge and successes.

Types of Events:

Tea Party
Car Wash
Run/Walk/Marathon
Set up a booth at a flea market or a car show
Used clothes/shoe sale
Organize a dress down day at work or school
Organize a penny challenge or change collection at school, work or in your community
Golf Tournament
Dinner Party/Silent Auction
Host a black tie or casual event
Organize a fundraiser at a local restaurant for a percentage of sales
Sell items of jewelry, quilts or art

The options are endless and every little bit helps FIRST!
Contact Chris Wassel at the FIRST office for more information.

ADVERTISE WITH FIRST!

Ichthyosis Focus welcomes your advertisements! If you provide a product or service that you feel would benefit our members, place an ad in a future issue of the newsletter.

For information about placing an ad, contact Lisa Breuning, *Ichthyosis Focus* Editor, at 800.545.3286, or email lbreuning@firstskinfoundation.org.

2012 Family Conference Scholarship Fund Guidelines & Criteria

A scholarship fund is available for families to apply for financial assistance to attend the 2012 National Family Conference in Denver, Colorado. The Family Conference Scholarship Fund, funded by the Jean Giroux Charitable Trust, will provide funds for registration and accommodations. Funding is limited; all applications will be evaluated based on scholarship guidelines.

Scholarships Available

- Scholarships are available for registration fees for two people, with one adult or child being affected. $215.00 Adult (13 years and older) $90.00 Child (2-12)
- Scholarships are available for hotel accommodations for three nights. $119.00 ++ per night

Eligibility for Applicants

- Each applicant must be a member of FIRST.
- Based upon availability of funds, priority will be given to applicants who clearly demonstrate a need for financial assistance based upon income and family circumstances.
- All applications will be strictly confidential.
- Each applicant must commit to volunteer at the conference for a period of time and attend all conference workshops.

Disbursement of Funds

- The award recipients will be reimbursed for their accommodations at the conference, either on the last day or by mail immediately after the event. Registration fees will be waived when you submit your registration form.
- The award recipients must bring receipts for reimbursement; no costs will be reimbursed without written proof.

Application

- Completed applications must be submitted by **February 1, 2012.** They can be faxed to 215.997.9403 or mailed to 2616 N. Broad Street, Colmar, PA 18915, ATTN: Family Conference Scholarship Fund.
- Applications must be submitted with:
  1. Confirmation of hotel reservation
  2. Copy of most recent IRS Form 1040

All applications will be strictly confidential.

FIRST Research Program Update

Continued from page 1

equivalents” for grafting onto immunodeficient mice. Then 4-6 weeks after grafting of “bioengineered skin equivalents,” the devitalized mouse skin used as a biological bandage spontaneously peels off, and efficiently regenerated human skin grafts became visible. We observed that LI-regenerated skin persists in the recipient animals for periods longer than 20 weeks, indicating stable engraftment of epidermal stem cells.

Macroscopically LI-regenerated skin showed the main phenotypic characteristics of patient skin, such as epidermal hyperplasia and compact hyperkeratosis. Using a human specific antibody against involucrin, we could confirm the human origin of the grafts and clearly delineate the border between mouse and human skin. Further investigations of diagnostic markers like the in situ monitoring of TG1-activity and immunohistochemical staining of TG1 using specific TG1-antibodies show a complete absence of TG1 in the grafts. Ultrastructural investigations display cholesterol clefts which are characteristic for transglutaminase 1-deficient lamellar ichthyosis.

In cooperation with Dr. Robert H. Rice from the University of California in Davis, our model was validated by proteomic analysis. Proteomics is the large-scale study of proteins. Proteins are vital parts of living organisms, as they are the main components of the physiological metabolic pathways of cells. The term “proteomics” was first coined in 1997 to make an analogy with genomics, the study of the genes. The proteome is the entire number of proteins of a tissue, including specific modifications. This will vary with time and distinct requirements, or stresses, that a cell, tissue or organism undergoes. In this study, we investigated skin of LI patients and of regenerated grafts and could show that the results in our grafts closely correspond to the results obtained in the skin of LI patients.

We conclude that this skin-humanized mouse model accurately recapitulates the human disease phenotype and concomitant molecular changes and can be used as an excellent tool for testing of novel and meaningful therapeutic approaches for this up-to-date untreatable genodermatosis.

Robert Rice, PhD

FIRST
Foundation for Ichthyosis & Related Skin Types™
Educate • Inspire • Connect

2012 Family Conference
Denver, Colorado
Scholarship Application
Application Deadline: February 1, 2012

Name: ____

Address: ____

City: ______________________ State: ________ Postal Code: ______________

Province: ______________________ Country: ______________________

Home Phone: ____________________ Work Phone: ____________________

Cell Phone: __________________ Email: ______________________________

Scholarships are available for registration fees for two people, with one adult or child being affected.

Name: ____________________ Age: _____ Type of Ichthyosis: ______________

Name: ____________________ Age: _____ Type of Ichthyosis: ______________

Have you attended a FIRST Family Conference before? ☐ Yes ☐ No If so, when? _______

Annual Family Income:	___ Under $20,000	___ $51,000 - $60,000
(please include a copy of	___ $21,000 - $35,000	___ $61,000 - $75,000
Your most recent W-2)	___ $36,000 - $50,000	___ Over $75,000

Why do you want to attend the FIRST Family Conference? (Use additional paper if necessary)

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Please return this form to the FIRST office by February 1, 2012.

Applications received after February 1 will not be considered.

FIRST and its officials reserve the right to disqualify any application that is incomplete.

Foundation for Ichthyosis & Related Skin Types, Inc.™ ● 2616 N. Broad Street ● Colmar, PA 18915
Phone: 215.997.9400 ● 800.545.3286 ● Fax: 215.997.9403 ● E-mail: info@firstskinfoundation.org
Web Address: www.firstskinfoundation.org

Grassroots Fundraising

Dinner at Souplantation in California

Adam and Meghan Knabe from California are the proud parents of three-year-old *Emma* and *Austin,* one year old, both of whom are affected with lamellar ichthyosis.

During Ichthyosis Awareness Month in May, Adam and Meghan decided that they wanted to have an event to raise funds for FIRST and awareness about ichthyosis.

The Knabe family had attended fundraisers at Souplantation, a family-friendly salad bar and buffet, in the past and thought it would be a good opportunity for them to try. The basis for the event is that participants bring a flyer to Souplantation on the night of the event, and a portion of the proceeds generated from those sales will benefit FIRST. They planned their event for May 19, and began handing out flyers to friends and family hoping to have 30 or so participants attend. However, those family and friends also gave out flyers to additional friends. More than 275 people attended the event on behalf of the Knabe family. On a normal Thursday evening, the restaurant serves approximately 300 patrons, but on this evening, the store doubled their sales, serving almost 600 people. The Souplantation staff and the Knabes were overwhelmed by the turnout! The line was out the door almost the entire evening.

Many thanks to Souplantation and the Knabe family for their efforts and raising more than $550.


Emma plays hostess at the event

The Knabe Family

High Heels & Handbags


Sam assists a participant with a purchase

FIRST member *Sam Zavitz* **and her husband,** *Brian,* are the proud parents of 3-year-old *Cate.* Cate is affected with lamellar ichthyosis.

Sam and Brian were so thankful for the help and support they received after Cate's birth that they wanted to give back. Sam held an event in May, called High Heels & Handbags ~ a Girls' Night Out at Glazed & Fired Pottery & Art Studio. The studio generously offered the location for the evening at a discount and then donated a percentage of any pottery painting proceeds from the evening. Sam created a facebook page for the event and advertised with friends and family. There were dozens of donations from friends, family, and businesses of handbags that could be sold the night of the event. In addition, Sam put several handbags up for auction on the facebook page. Facebook friends bid on the handbags right up until the 9:00 pm deadline.

The evening was a terrific success! Approximately 60 people attended this Girls' Night Out and purchased like-new handbags and shoes. An additional 125 people participated in the Facebook auction. In addition, many items were donated for a raffle. Participants purchased tickets in hopes of winning an item.


Participants enjoy the event.

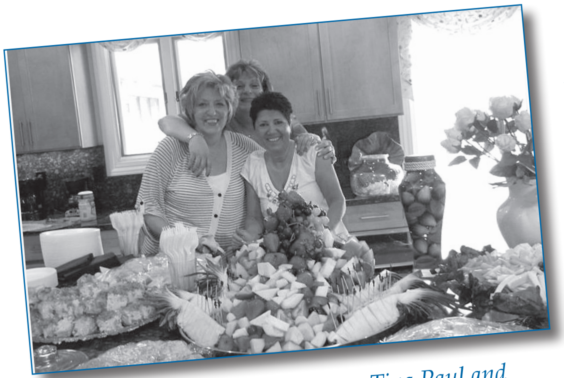
Sam has a close group of friends who wanted to help her with the event. They were instrumental in helping this evening become the success that it was.

The efforts of Sam and her friends raised more than $2,000 for FIRST! We are so appreciative of everyone's hard work on our behalf.

Grassroots Fundraising

The Foundation is very thankful to all of our wonderful members for their hard work. Grassroots fundraisers are a great way not only to raise money for FIRST, but also to raise awareness about ichthyosis in your community.

A Girls' Night Out in Ohio


from left are: JoAnn Marra, Tina Paul and Carmella Corbo

Edie Wohlgang is the grandmother of 9-year-old ***Adam Klafter,*** who is affected with EHK. She has held many fundraisers for FIRST over the years. On Wednesday evening, June 8, Edie hosted a Girls Night Out at the home of her close friend Carmella Corbo, which was attended by approximately 100 people and raised more than $2,800 for FIRST.

Edie is very grateful to her friends who worked tirelessly to make this event so successful. She extends her sincere thanks below.

GOD did not give me sisters, he did much better. He gave me angels, as friends. Very recently we girls came together just for a little fun and to play cards and within two months, these wonderful ladies opened up their homes and their hearts to FIRST. Carmella Corbo opened up her beautiful home for this evening. Joann Marra and Tina Paul spectacularly catered the event. I also extend a special thank you to Michelle Marra and Melissa Marra for their wonderful efforts. All the ladies worked tirelessly to procure the wonderful donations for our raffle. We showcased and offered unique, one-of-a-kind pieces from Edie's Beadies.

FIRST is also very grateful to Edie and her wonderful friends for their efforts on our behalf.


from left are: Michelle Marra, Edie Wohlgang, Melissa, Gia Rose, and Lisa Marie Marra

Dance Recital in Ohio

On June 5, the students and faculty of ***Emjaez Dance Studio*** in Bay Village, Ohio participated in a recital entitled, "In a Perfect World." Each routine of the show was dedicated to a special cause, and the dancers were asked to raise money for their specific cause. The students raised money for FIRST in honor of former Emjaez student Emma Klima. Emma is 7 years old and affected with epidermolytic hyperkeratosis (EHK). The recital raised more than $200 for FIRST. The foundation is very grateful to Emjaez for their donation.

Brother helping brother in Ohio

Jackson mans the drink stand

When ***Denise Lewis*** of Shelby, Ohio decided to have a garage sale in June, her oldest son ***Jackson*** decided to have a drink stand and donate the proceeds to FIRST, because he wanted to find a way to help his younger brother. The Lewis' youngest son, ***Brennan,*** is five years old and affected with Congenital Bullous Ichthyosiform Erythroderma (CBIE). Brennan's ichthyosis appears to be a spontaneous mutation, as neither of his parents has the gene. Denise also decided to donate the proceeds of her garage sale to FIRST. The 2-day sale was held in mid-June and more than $500 was raised by both ventures.

Photographer Lucas Wright/SDG Newspapers

Jackson and Brennan prepare for their sale

A New Website Links Givers and Charities

by Diane Mastrull

Following is an article that appeared on philly.com on August 22, 2011.
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Like many people the day after Thanksgiving, Blair Souder wasn't feeling well. But his distress wasn't related to overeating.

It was from watching television footage of bargain-hungry buyers on Black Friday, the launch of the Christmas shopping season.

"People are fighting each other for DVD players at Target," the 48-year-old Chester County father of two recalled of that scene last November.

It was a disturbing contrast to the few weeks he had just spent in Nepal, where he had done some hiking and had become enthralled with the simple, peaceful existence of the Sherpa people.

"It just really struck me that all this stuff that we think we need or that we think we need to give . . . has really nothing to do with how happy and peaceful we can feel," said Souder, a former General Electric Co. marketing executive with degrees in chemical engineering, business administration, and psychology.

That realization has led to Souder's forming a Web-based business with a brother in California, Kirk, that Souder hopes will "create a new vernacular for how we celebrate things."

ShiftMyGift.com launched last Monday, offering people a way to celebrate any occasion - birthday, baby shower, wedding, graduation, new home, the sun rising - by rerouting money that would have been spent on gifts to needy causes. So, for instance, instead of buying your husband an iPad, you visit his registry page on ShiftMyGift.com and make a donation in his name to any of the nonprofit groups he has listed there as his preferences.

Among the motivational reasons the site offers for donations instead of gifts:

"Your friends don't have to slog through the malls finding you stuff you don't want. Your friends get a tax deduction. The environment loves you for not using gas, packaging, and wrapping paper."

Souder says his site is different from donation sites such as Facebook's Causes in that ShiftMyGift does not raise money. Rather, it aims to serve as a simple-to-use digital link between donors and organizations that rely on charitable giving.

"I see it as a movement as much as I see it as a website," Souder said last week at his home office in Lincoln University, where he also runs a consulting business to help create more-engaging work environments.

What influenced the physical design of the ShiftMyGift site, Souder said, was his own experience last year of not being able to find a donation site that was simple to use and allowed him to find any nonprofit organization in the country in one place.

As a result of a partnership the site has with Guidestar, which maintains a database of registered U.S. nonprofit groups, users of ShiftMyGift.com will have access to a list of 1.2 million organizations.

If already in the Guidestar database, nonprofit groups do not have to register with ShiftMyGift to qualify as a gift recipient. However, registering will enable them to create their own ShiftMyGift landing page on that website.

Souder also plans to feature a variety of charities on his company's website at any one time. The current criterion for a nonprofit to qualify for such recognition, he said, is the ability to promote the site to more than 50,000 people. He said he had no plans to sell advertising on the site. Its only income is from a $1.49 service fee tacked on each donation.

"I'll be happy if the site breaks even and allows me to hire people to help support it and grow the awareness and movement, and therefore the impact," Souder said.

The nonprofit community, always in search of new funding streams, is enthusiastic.

"We try to stay on the cutting edge of social media and provide our donors and followers any avenue they want to use," said Melanie Mullinax, communications manager at Project HOPE, an international health-education and humanitarian-assistance organization. She said she intends to widely promote ShiftMyGift to Project HOPE's more than 100,000 donors.

Philadelphia-based Back on My Feet, which uses running to build physical and emotional well-being among the homeless, is about to open its eighth chapter, in Atlanta, and considers ShiftMyGift "a unique call-to-action and a one-stop-shop for occasion-based giving," said spokeswoman Rachelle Damminger.

At the National Wildlife Federation, Kristin Johnson, senior manager of online editorial, said she was "really excited" about the more personal touch ShiftMyGift brings to fund-raising.

Having supporters encourage family and friends to skip gift-buying and divert that money instead to the federation is "more powerful than if we ask their friends and family for a donation," Johnson said.

Souder said success for ShiftMyGift will be defined in stages - for instance, when major giving milestones are reached, such as $1 million in shifted gifts.

And when he stops getting wallets for Christmas.

"I have four," Souder said.

FIRST is one of the charities listed on the Shift My Gift website. Please watch our website for ways that you can help FIRST through Shift My Gift.

Educating Physicians, Families and Patients for Life.

Ichthyosis is More Than Skin Deep.

Ichthyosis is a family of genetic skin disorders characterized by dry, cracked, scaling, and thickened skin affecting people of all ages, races, and gender. There is no cure for ichthyosis but physicians, researchers, parents, and those affected are collaborating on ways to manage the disorder. Managing it presents many medical and psycho-social issues for the entire family.

FIRST, the nation's only foundation dedicated exclusively to the unique needs of the ichthyosis community, has been educating, inspiring, and connecting those touched by these disorders since 1981.

Here are some ways we can help you and your patients...

- An interactive tele-ichthyosis website where physicians can submit cases for consultation
- Diagnosis and treatment tools
- Robust publication library
- Individual family support

To further explore these resources and more contact FIRST at 800-545-3286 or visit us at www.firstskinfoundation.org.

Look for more ways we can help you and your patients in the next issue.

FIRST
Foundation for Ichthyosis & Related Skin Types™
Educate • Inspire • Connect
2616 N. Broad Street, Colmar PA 18915

www.firstskinfoundation.org

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Donating to FIRST through the **United Way** or **Combined Federal Campaign** is an easy and convenient way to support the important work of the Foundation. Depending on your pay schedule, a small deduction of $5.00 per paycheck can add up to over $200 per year for the Foundation. It's simple and convenient and can really make a difference.

The United Way of America is the national organization dedicated to leading the United Way movement in making a measurable impact in every community across America. The United Way movement includes approximately 1,400 community-based United Way organizations. Each is independent, separately incorporated, and governed by local volunteers. The Foundation for Ichthyosis is the recipient of funds from many of the United Way community-based organizations. Through the Donor Choice Program, you can designate all or a portion of your donation to the Foundation. Simply write in "Foundation for Ichthyosis & Related Skin Types, Inc.™" on the Donor Choice Option form, and your gift will be sent to our office. Be sure to include our current mailing address and phone number, 2616 N. Broad Street, Colmar, PA 18915, 215.997.9400, 800.545.3286

The (CFC) is the annual fund-raising drive conducted by federal employees in their workplace each fall. Federal employees and military personnel raise millions of dollars each year through the CFC to benefit thousands of non-profit charities. The Foundation is the recipient of funds from many CFC organizations throughout the country. The Foundation's CFC code is 10322, which is listed in the charitable organizations directory. Contact your Human Resources Department to find out how you can support the Foundation using United Way or Combined Federal Campaign.