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

Ichthyosis Focus

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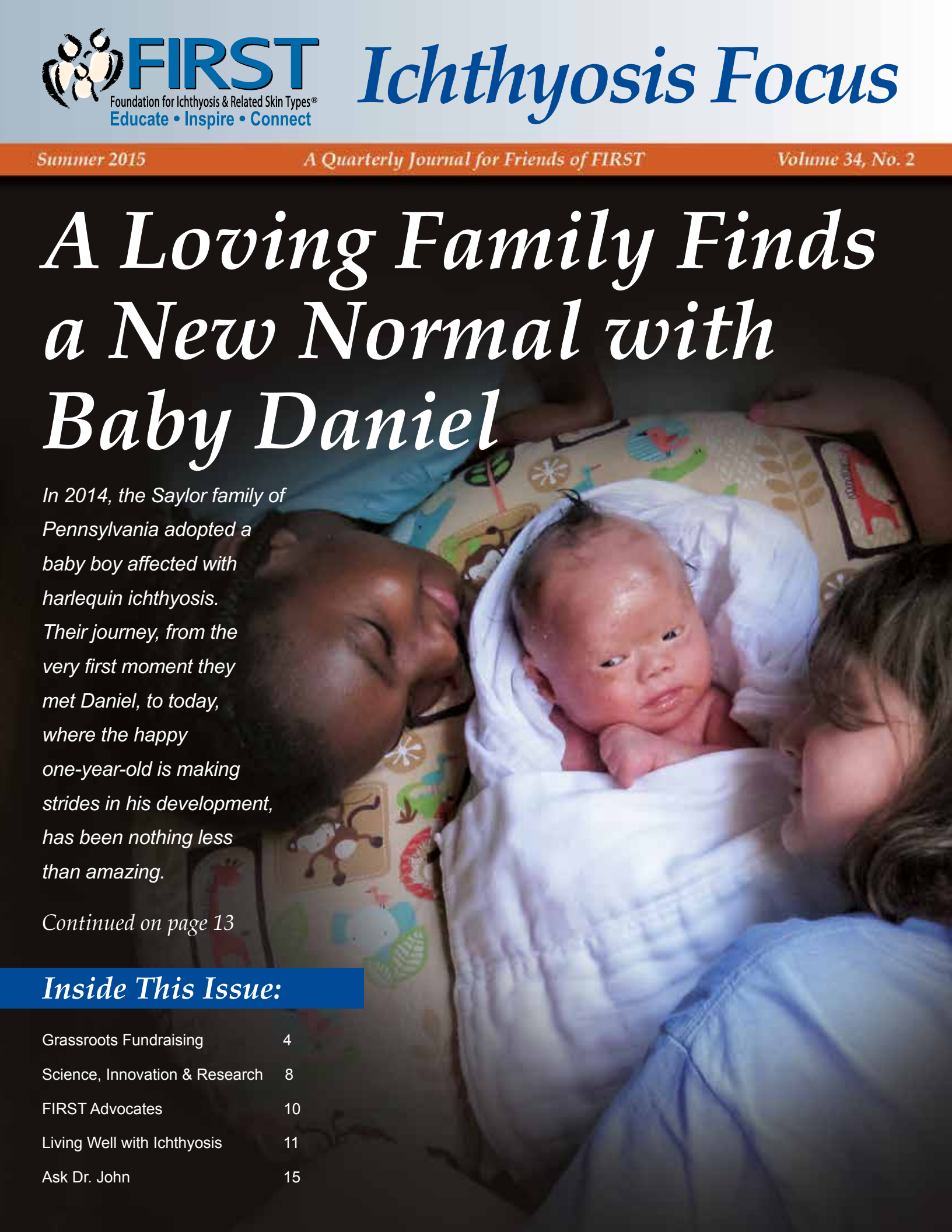
A Loving Family Finds a New Normal with Baby Daniel



In 2014, the Saylor family of Pennsylvania adopted a baby boy affected with harlequin ichthyosis. Their journey, from the very first moment they met Daniel, to today, where the happy one-year-old is making strides in his development, has been nothing less than amazing.

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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.

Editor's note: In the Winter 2015 issue of Ichthyosis Focus, the GE Foundation was mistakenly left out of the annual report listing for FY2014. We kindly thank them for their donation of $1,600.

Chief Executive Officer Report

Dear Members and Friends:

Our awareness-raising efforts this quarter have truly topped the charts! Our community not only embraced and shared a strong Ichthyosis Awareness Month (IAM) message -"Be Inspired. Use Your Voice. It Matters to Someone." - but they have also enthusiastically engaged in our very first online personalized RAISE fundraising campaign. This member-driven campaign inspired 53 fundraisers to raise over $60,000! We are deeply grateful to each participant, and for every single dollar raised.

In fact, this issue of Ichthyosis Focus is an entire celebration of fundraising, awareness and innovation. You'll learn all about the exciting IAM events that took place around the globe, like the first stop on the Release the Butterfly Benefit Tour, as well more ways to get involved, self-advocate and make a difference. Plus, don't miss our new features, Living Well with Ichthyosis and Science, Innovation & Research, where you'll find the story of FIRST's pivotal role in the discovery of the ichthyosis en confetti gene mutation, submitted by Dr. Leonard Milstone. And, we are truly delighted to introduce baby Daniel, affected by harlequin ichthyosis, and recently adopted into the kind and loving Saylor family of New Cumberland, Pennsylvania. Last but not least, we are gearing up for the 2016 National Conference - San Diego. Stay tuned for all the exciting details. We hope to see you there!

Enjoy and be well!

Jean R. Pickford
CEO, FIRST

Correspondence Corner

Dear FIRST Foundation,

I wanted to thank you very much for the scholarship you have given me. I can't tell you how much I appreciate it and need it. The $1,500 you have awarded me is a great blessing. I plan on going to University of Colorado, Colorado Springs in the fall. I am not sure what I will major in, but my interests are in the sciences. Thank you again, I am truly grateful.

Austin Milam

Dear FIRST,

I am so grateful to have been able to attend the patient support forum today in Bloomington, Minnesota! My son had such a great time and I learned a ton and connected with some really amazing families. It helped me and my son so much to know that we're not alone in the day-to-day struggles of having a child with ichthyosis. Seeing both dermatologists take their time to come and share their expertise this weekend really speaks volumes about the commitment they have to their patients.

Minneapolis, Patient Support Forum attendee, 2015

Dear Diya, Aliya, Ms. Pickford and FIRST staff & members,

From the bottom of our hearts and depths of Liam's skin, thank you! At 3 months old, our son was diagnosed with ichthyosis and it's been a challenging journey. We thank you for considering us for this honor and choosing to bless us with the Diya & Aliya funds to continue to manage this rare disease. You have given us hope and for that we are grateful. This foundation has been a wonderful support and we are very proud to be members, and our son's advocate.

God Bless You,

The Hubart Family

Heat Up Your Summer with a Grassroots Fundraising Event

Fun, fun and more fun! That is the bottom line for grassroots FUNdraising events. If you are planning a special fundraising activity, it's best to choose an event which you and your friends will enjoy! However, it's also important to realize that a grassroots fundraising "event" can be as simple as asking your local coffee shop if you could leave a FIRST donation can next to the register, or even set up a table out front with informational pamphlets for the afternoon. Every dollar counts just the same. Perhaps you are more of a talker than a planner. Ask your local community center if you could give a brief talk about ichthyosis and FIRST. Perhaps leave a basket for anyone who'd like to make an offering.

As for soliciting volunteers, it is likely that volunteers will be more engaged if they are enjoying what they are doing. This will create the most energy, enthusiasm and fundraising dollars. Plus, it will be a more rewarding (and likely easier) experience for you. Most importantly, FIRST can assist you every step of the way. Contact Director of Development, Madeline Bergman at mbergman@firstskinfoundation.org. Her job is to make it easier for you.

IAM Celebrations Continued…

Wine Tasting Event in Ohio

Not a single snowflake was in the forecast for this year's annual wine tasting held at the beautiful Grand River Cellars Winery and Restaurant in Madison, Ohio. Hosted by Susan Schumacher, the event honors her nephew Mark Klima and his family. Mark and Lisa are proud parents of Emma Klima, who is affected with epidermolytic ichthyosis.

More than 60 people attended and the event raised nearly $3,700. Guests enjoyed beautiful scenery, fantastic raffles, delicious food and fabulous wine from Grand River Cellars. Thank you to Susan for her continued support of FIRST.

Wine Tasting Event in New Jersey

One of FIRST's favorite annual fundraisers was held this May. The Cina family, whose daughter, Portia, and son, Myles, are affected with ichthyosis en confetti, hosted their 7th Annual Wine Tasting Event on Sunday, May 9. Friends and family gathered at the Cina residence for some delicious wines and appetizers. Adding a special touch to the event, Portia played piano for the guests and her artwork was displayed at the event. This event raised over $3,800 for FIRST. Thank you to the Cina family for their ongoing support of FIRST and Ichthyosis Awareness Month.

Awareness Dinner in India

As a testament to the fact that FIRST is the leading global patient organization for those affected by ichthyosis and related skin types, Dr. Victoria and Mr. Shalo Khing held an awareness dinner on Saturday, May 9, in Dimapur, India. The dinner was held in honor of their daughter, Kenile, who is affected with ichthyosis.

Over 55 guests joined the Khings for a delicious dinner and educational presentations by Dr. Leishiwon Kumrah and the Khing family. Dr. Khing said, "It was overwhelming to see such a huge response from our community, the few we handpicked, whom we felt would understand and value such an event and who in their place of work could spread awareness." The event even received coverage in the local paper. Thank you to Victoria and Shalo and their entire family for hosting this "Evening of Hope." You are helping us to spread awareness of ichthyosis throughout the world.

Release the Butterfly Tour Stops in Mechanicsburg, Pennsylvania!

On Saturday, May 30, the incredibly talented families of FIRST performed the Release the Butterfly Benefit Concert and it proved to be an evening of pure enchantment. Performed at the Capital Area Christian Church in Mechanicsburg, Pennsylvania, the Pretak, Saylor, Murphy, Steinitz and Evans families, along with numerous friends and participants, presented a multi-purpose, highly entertaining production. The concert included over 20 inspirational performances by vocalists, instrumentalists and dancers from Harrisburg, Pittsburgh, Philadelphia, Lancaster, northwestern Pennsylvania and Maryland. A special highlight, for the second year in a row, was the "Behind the Butterflies" talk-show style Q&A segment, focusing on questions regarding ichthyosis and FIRST. Participants in the talk show included four extraordinary young women - Bailey Pretak, Abby Evans, Hunter Steinitz, and talk show host and FIRST friend, Natalie Hann.

Brand new to the concert this year was the location (a.k.a the first stop of the tour)! FIRST Board Member, Tracie Pretak, mother of Bailey Pretak, affected with autosomal recessive congenital ichthyosis-lamellar (ARCI-LI), said, "We have been performing the concert in Johnsonburg and surrounding communities for over 15 years. As our audience has grown to include more and more affected families, it made sense to take it on the road and share it with their communities." And take it on the road they did.

With the help of FIRST members Mark and Nicole Saylor of New Cumberland, Pennsylvania (*story on page 13*), who have adopted baby Daniel, affected with harlequin ichthyosis (HI), the first stop of the Release the Butterfly Tour performed to an audience of nearly 80 people. Among other remarkable moments, some particularly emotional highlights of the show were the beautiful violin solo by FIRST member Mikela Murphy, and the Saylor's three daughters taking the stage to sing "It's You I Like" by Fred Rogers, in honor of their new brother Daniel.

Tracie also notes, "It is the first of hopefully many concert stops for the Release the Butterfly tour. We are already discussing a 'tour stop' in Lancaster! The tentative date (pending an open venue) is Saturday, September 19th. We believe we can make a difference with our message of hope and courage. We already have. And if we can keep connecting with more ichthyosis families and meeting others in the audience, it is so worth our time, money and effort."

The 2015 Release the Butterfly benefit concert raised $1,800. FIRST is sincerely grateful to all who attended, participated and generously donated. The commitment and enthusiasm of the FIRST community never ceases to amaze! Stay tuned for upcoming dates and information regarding the next stops on this incredible journey.

Performers and Participants (in alphabetical order)

Amanda Bertsch
Ross Bish
Sophie Duffield
Abby Evans
El Elmet Dancers of
Reverence Studios (Alysha Argot, Sophie Gray, Callie McCaffrey, Leah Noss, Hannah Spangler, Emily Uber, Kayla Warren)
Sami Ferragine
Haley Freeburg
Natalie Hann
Mikela Murphy
Oakwood Baptist Praise Band
(Dominique Benninger, Geren Fisher, Mark Saylor, Nicole Saylor)
Bailey Pretak
Tracie Pretak
Brandon Rexrode
Julian Saylor
Melody Saylor
Nicole Saylor
Savannah Saylor
Hunter Steinitz

Ace in the Hole Foundation Beach Run/Walk

On Saturday, May 9, over 450 people attended the sixth annual Ace in the Hole Foundation Lido Beach Run/Walk in Long Island, New York. The event was held in honor of 1st Lt. Michael LiCalzi who died in action in Iraq on May 11, 2006. His twin brother, Greg LiCalzi, Jr., the President and Founder of Ace in the Hole Foundation, serves on the Board of Directors of FIRST and is affected with ARCI-lamellar type ichthyosis.

The event was a two-mile run/walk beginning at the "1st Lt. Michael LiCalzi Path" at Lido West Beach. This year, several special guests joined in on the fun and beautiful weather. Major General Brian Beaudreault, Commanding General of 2nd Marine Division and Lt. Col. Rob Bodisch, Commanding Officer of 2nd Tank Battalion, both attended the run. Marines from Camp Lejeune also were there to support the Ace in the Hole Foundation.

"Since 2008, Ace in the Hole Foundation has worked to raise money in Mike's memory to help those affected by rare diseases or medical conditions," said Ace in the Hole president and founder Greg LiCalzi. "Having ichthyosis, I understand the need to support FIRST. The positive work done by FIRST within the ichthyosis and related skin types community is immeasurable. We invite all those affected to please join us next year in May on Lido West Beach to remember Mike and his sacrifice, as well as support FIRST."

This year's event raised $65,000! FIRST is one of the beneficiaries and will receive a $15,000 unrestricted donation in the memory of 1st Lt. Mike LiCalzi. Thank you to the Ace in the Hole Foundation for their continued support of FIRST and congratulations on another successful event.

About the Ace in the Hole Foundation

1st. Lt. Michael LiCalzi was born and raised in Garden City, a proud graduate of Chaminade High School and the United States Naval Academy, and a 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. It happened when the M1A1 Battle Tank, in which he and three other Marines were traveling flipped off a bridge into a canal in Al Anbar province, Iraq. All four of the young Marines in the tank drowned.

Ace in the Hole Foundation is a NY State 501(c)3 not-for-profit organization that was founded to properly remember and honor the sacrifice of 1st Lt. Michael LiCalzi USMC. The Foundation provides financial aid, material assistance and succor to charitable organizations and causes. The Foundation's financial aid and material assistance is administered directly to deserving recipients or through contributions to charitable organizations with which the Foundation has working partnerships. Particular emphasis is given to the support of Marine tankers, especially those of the 2d Tank Battalion, 2d Marine Division, as well as other armed service personnel and their families.

FIRST Night Out Events

Pittsburgh Pirates

April 19 turned out to be a great day for baseball, friends and family. Lauren and Matt Kocher, whose son Calvin is affected with ARCI-lamellar type ichthyosis, hosted their annual FIRST Day Out at the Pittsburgh Pirates. The day consisted of a tailgate and basket raffle, and then over 160 people joined the Kochers to watch the Pirates take on the Milwaukee Brewers.

In addition to the Kocher family, two other affected individuals and their families joined in the festivities. FIRST board member Tracie Pretak and her daughter Bailey, who is also affected with ARCI-lamellar, and Anthony Lecocq, joined to cheer on the Bucs.

We are so thankful to all those who attended and enjoyed a day of fun and raising awareness of ichthyosis and FIRST. The day raised over $6,000 for FIRST. Go Bucs!

Philadelphia Phillies

The Fahey family Organized FIRST Night at the Philadelphia Phillies, where they took on the Cincinnati Reds on May 13. Although the game was not victorious for the Phightin' Phils, the Fahey family enjoyed their time together and supporting FIRST.

Lakeshore Chinooks

FIRST Board member Kimberly Cole and her family and friends headed to the Lakeshore Chinooks on Friday, June 5, to raise awareness and funds for FIRST. Kim has held many fundraisers for FIRST in honor of her daughter, Harper, who has erythrokeratoderma variabilis (EKV). Thank you Cole family for continuing to raise awareness of FIRST in your community.

Minnesota Twins

The Kimmelman family hosted a FIRST Night Out with the Minnesota Twins on Sunday, May 31. Longtime supporters of FIRST, Sarah, who has EI, Jonathan and their son, Alex, who has CIE, watched as the Twins played the Toronto Bluejays. The Sinclair family, whose daughter Genevieve has EI, also joined in on the fun.

Tampa Bay Rays Game

More than 40 people gathered at Tropicana Field to cheer on the Tampa Bay Rays and raise awareness of FIRST. The Rotondo Family hosted the game on Saturday, June 13 in honor of their daughter, Mary Elizabeth who is affected with ichthyosis. Thank you Rotondo Family for organizing this fun event!

Washington Nationals Game

It was a beautiful night for baseball for the FIRST Night Out with the Washington Nationals on Wednesday, June 24. The Beard Family invited friends to support FIRST and honor their son, Blake who has Loricrine Keratoderma. Through the generosity of the entire Beard Family and friends, the game raised over $1,000 for FIRST!

NY Mets Game

It was another great year for the annual FIRST Night Out with the New York Mets! The Benedetto family, whose son Marc is affected with lamellar ichthyosis and the Saccente family whose daughter Kylie Leigh is also affected with ichthyosis, gathered another large group of supporters to cheer on the NY Mets and raise awareness of ichthyosis at Citi Field on June 26, 2015. The Benedetto and Saccente Families were honored with an NY Met's Spirit Award on the field before the start of the game. A big thank you to the Mets organization for working with FIRST to bring as much awareness to ichthyosis and the mission of FIRST as possible. Awareness was raised throughout the entire game including FIRST logos displayed on LED boards and an informational ichthyosis video, produced by FIRST, played throughout the entire arena.

More Grassroots Events

FIRST is sending out a round of high-fives to Kendall and Jeremy Neurohr and their family. The Neurohrs and family members ran the Big D Half Marathon on April 12 in Dallas, Texas, to raise funds and awareness for FIRST. Kendall and Jeremy's daughter, Emerson, is affected with congenital ichthyosiform erythroderma (CIE). Kendall and the Neurohr team raised over $3,600 for FIRST. According to Kendall, the race day was beautiful and, by wearing ichthyosis-themed T-shirts, participants and spectators learned about FIRST and ichthyosis. A big thank you to Kendall, Jeremy and the Neurohr family.

Several organizations and companies also supported FIRST by hosting a casual day at the office. These 'Jeans Days' are a great way to raise awareness in your workplace. A big thank you to:

Bergseth Bros. Co., Inc. in Fargo, North Dakota for raising $300 in honor of the Linman family, whose daughter Isabelle is affected.

Paine Intermediate School in Trussville, Alabama for raising $210 and celebrating the Ekonen family, whose daughter Annalie has ARCI-lamellar type ichthyosis.

Paychex in East Providence, Rhode Island for raising $397 and having a Jeans Day for Rare Genes, in honor of the Robinson Family, whose daughter Sienna has epidermolytic ichthyosis.

Auburn Elementary School in Shelby, Ohio, wore green and jeans in support of student Brennan Lewis, who has ichthyosis. Staff and students were invited to make donations in support of FIRST. All together, they raised over $200.

FIRST's Pivotal Role in the Ichthyosis With Confetti Story

by Leonard Milstone, MD

[Please note that this article is an abbreviated version of the full length submission. The submission in its entirety can be found at firstskinfoundation.org.]

Major advances in the ichthyosis with confetti (IWC) story were the result of a fortuitous intersection of people, timing and technology. ***Discovery of the mutant gene and identification of the cause of the spots in IWC was, in no small part, the result of asking the right question.*** While others had sought but failed to identify the cause of the red, scaly skin in IWC, we started by asking what caused the distinctive spots of apparently normal skin? From that moment forward, FIRST played significant roles in this effort.

The story began in 1998 when I saw an infant who had uniformly scaly red skin and slow growth compared to his fraternal twin. He looked like an infant with typical congenital ichthyosiform erythroderma (CIE). He returned one year later with a single spot of normal-appearing white skin on his cheek and, in retrospect, became my personal introduction to ichthyosis with confetti. At this point in time, only two additional cases had been reported, making it an exceedingly rare form of ichthyosis. Shortly after I saw that first case, two girls, one from Texas, one from Maine, with apparent CIE and dozens of normal-appearing white spots, came to see me. One girl, Haley Rice, was the daughter of the then-president of FIRST Donna Rice. Donna had been instrumental in setting the stage for FIRST's organizational stability and recent rapid growth. When Mary Williams, MD, a founder of FIRST, was attending the FIRST family conference with me in 2002 and said, "Len, you have to figure out what's going on with Haley," I knew that it would become a long-term personal mission.

As we began our investigation, we carefully asked our patients about the confetti spots. All reported that they were not noticed until late in the first year of life and that the spots dramatically increased in size and number over time. Most remarkably, the spots were normal-appearing skin both by the naked eye and under the microscope. The story that the spots appeared after birth and increased in size and number over time suggested that they were not the usual kind of mosaicism familiar to dermatologists and pet lovers. Rather, we hypothesized that they must represent revertant mosaicism. Revertant mosaicism occurs by spontaneous correction of a mutant gene leading to acquired patches of normal skin. In talking with colleagues, we learned that several groups in the 1990s had looked for mutations in the half dozen genes known at that time to cause dominantly inherited ichthyosis, including several keratin genes, and had found no evidence of mutations in them.

The Perfect Storm of Technology, Perseverance & Passion

About the time I began collecting cases with IWC, high density SNP (single nucleotide polymorphism) arrays had made it to the forefront of gene mapping. One day I asked Keith Choate, MD, PhD, a first year resident in dermatology, whether he thought SNP arrays would facilitate finding the gene for this disease and whether it was a project in which he might be interested. Keith had already done research in ichthyosis as an undergraduate at Stanford and had earned his PhD with Richard Lifton, MD, PhD, chair of the genetics department at Yale. Though he was a busy resident and had not worked in skin science for eight years, he suggested we meet with Dr. Lifton. The information we presented at that meeting about IWC was concise: a rare, likely dominantly inherited form of ichthyosis in which normal spots of skin appear after birth and increase in number and size over time. Dr. Lifton suggested that loss of heterozygosity (LOH) was the simplest explanation for the normal spots, and that using SNP arrays to compare a confetti spot with an affected spot on the same individual should facilitate mapping the gene. The value of looking specifically at cells in the epidermis, rather than whole pieces of skin, became clear in the brief discussion that followed. ***In most genetic studies, the focus is on what is happening in the diseased tissue. In ichthyosis with confetti, the focus became what is happening in the normal tissue.*** The timing of Dr. Choate's re-entry into ichthyosis research was both fortuitous and critical.

Within a few months of embarking on this project, Dr. Choate had a stunning result. He had used high density SNP arrays to compare DNA in keratinocytes from diseased skin to DNA in keratinocytes from normal-appearing confetti spots. In each of the confetti spots in each of our patients, he found long stretches of loss of heterozygosity on chromosome 17 that could have only occurred by a mechanism never before reported in revertant mosaicism – mitotic recombination. Other cultured cells from the confetti spots, namely fibroblasts, showed no such changes. The importance of having patients who were willing to travel long distances to New Haven to donate skin from many red and white spots

cannot be overemphasized. Yet, after two and a half years of carefully analyzing many, many "candidate" genes, the responsible gene remained a mystery.

Sometimes stalled science is rescued by technology. For 40 years, the gold standard for determining gene sequences was Sanger sequencing, a method used to reveal the first complete human genome sequence. As a result of the human genome project, Sanger sequencing had become faster, more reliable, cheaper and automated. Yet limits on all those improvements became obvious and during the 1990s totally new technologies that would be faster and cheaper were developed.

After months of planning and discussion, Dr. Choate selected one of the new sequencing methods and devised several technical adaptations that made it useful for identifying disease-causing mutations. And after years of uninformative Sanger sequencing, he identified the mutant gene in the very first family analyzed using this new approach. The mutation was in the gene for keratin 10, and in every one of our families the mutation appeared in a small region of the keratin 10 gene that was rich in guanine/cytosine (G/C) base pairs.

One of the last patients to enroll in our initial study helped us solve one of the inconsistencies in the literature that had troubled us: why were normal-appearing keratohyalin granules present in the diseased skin of the first French patient reported but absent in the diseased skin of the other patients – including all our patients who had IWC and KRT10 mutations? The affected skin had no mutation in KRT10, but confetti spots from this individual did demonstrate LOH, but this time on chromosome 12. Chromosome 12 is where the KRT1 gene resides. Further investigation resulted in the identification of a mutation in KRT1, the gene that codes for the protein that normally pairs with KRT10 to form the keratin filaments in the epidermis.

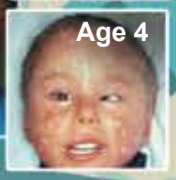
Scientific breakthroughs should clearly and definitively answer the question at hand, but they also often raise even more difficult questions. In many ways, the IWC saga is one of those stories. Why don't all patients with EI and KRT 10 (or KRT1) mutations develop white spots? Why has revertant mosaicism caused by mitotic recombination not been seen in other dominant disorders of keratinization? Is there something about the particular mutations in the keratin 10 or keratin 1 proteins in IWC that cause mitotic recombination to happen more frequently than expected? These broad questions and the novelty of the mechanism by which the confetti spots arise generate excitement in the scientific community and explain, in part, why this particular gene discovery was thought to merit publication in high-profile journals [1]. The new questions raised will keep us going for many years.

Keith Choate was the first recipient of FIRST's Clinical Scholar Award. Support for that award came from a generous donation from the Lennox Foundation at a time when it was most needed. It provided Dr. Choate with protected time to concentrate on this research early in his career. Many other people contributed to this work. Several are members of FIRST's MSAB: Drs. Mary Williams, Peter Elias, Amy Paller, Amy Nopper, and Alanna Bree. We received advice and encouragement from many other colleagues along the way. The patients are all members of FIRST. They and their families are an ongoing source of inspiration and support to these investigations, and to whom no words of thanks adequately express our appreciation.

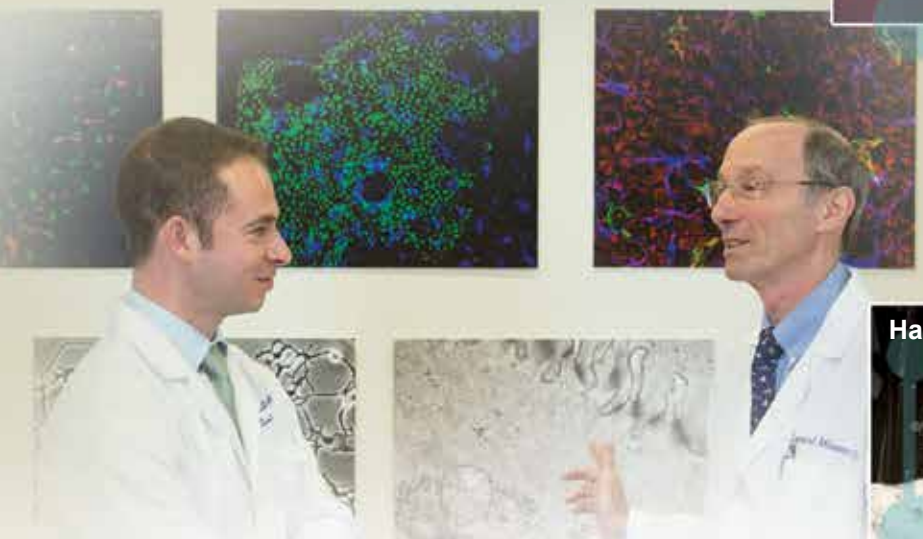

Shawna Grady


Randy Saunders


Age 1


Age 4


Haley & Donna Rice



List includes references to full-length article.

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FIRST ADVOCATES

What Is the Difference if I Advocate...or Not?

Advocacy is the act of pleading or arguing in favor of something, such as a cause, idea or policy.

WHY should I participate? Since it is a rare disease, ichthyosis is not well-recognized, understood or researched. Increased education of our communities and elected officials will help lead to more research, funding, advancements in treatments and possibly a cure.

WHAT can I do?

1. ***Write, call or visit your local, state and federal elected officials***
   - Your elected representatives are most likely not going to hear about ichthyosis unless you educate them.
   - Introduce yourself to your representative and concisely tell them how ichthyosis has affected you. Write a letter, asking for a meeting in the local district office or attend a town hall meeting in your area.
   - Call the local district offices of your elected representatives or check out their websites to learn more about their town hall meeting schedule.
   - When you meet with the representative, leave behind a FIRST brochure for their reference.
   - Don't forget to personalize your story, tell them as much about your personal experience as possible.
2. ***Ask your elected representatives to support a proclamation supporting ichthyosis awareness.***
   - Many local and state governments issue proclamations on topics important to their community. The proclamation is simply a formal announcement that raises awareness of the topic in your area and hopefully, beyond.
3. ***Write a letter to the editor of your local paper***.
   - Raising awareness of ichthyosis will help lead to scientific advancements and better treatments.
   - Consider writing to a local weekly paper or a neighborhood newsletter that may be more likely to publish human interest stories.
4. ***Follow #FIRSTADVOCATES on social media and stay informed of new opportunities to get involved.***

Time to TAKE ACTION

Access to 21st Century Cures Act for Individuals With Rare Diseases

The House Energy and Commerce Committee unanimously approved H.R. 6, the "21st Century Cures Act," on May 21 after reaching a bipartisan agreement to fund the legislation (Cures bill). Often described as a "landmark medical innovation" bill, key provisions of the initiative are intended to speed up the pace at which FDA approves new medicines for conditions currently lacking cures, as well as a lengthy list of other proposals. One of the key provisions of the act is the funding for the National Institutes of Health (NIH) - the Cures bill provides $10 billion over the next five years to the NIH, as well as $550 million in additional funds for the FDA. A vote on the Cures bill by the House of Representatives is expected this summer. The full Senate is not expected to vote on the legislation until the end of the year.

How can you USE YOUR VOICE? Find out more about the "**21st Century Cures Act**," and

Join the Rare ActionNetwork: https://rarediseases.org/advocate/take-action-locally/join-rare-action-network/

State by State Info: https://rarediseases.org/advocate/take-action-locally/state-state-information/

Living Well With Ichthyosis

Travel Tips:

Taking a summer vacation? Traveling with children can certainly present its own unique set of challenges. Add in the fact that most FIRST families are also coping with children who are affected with a rare skin condition and the challenges become even trickier! From moisturizing, to waterparks, to hesitation about using hotel bathtubs, to overheating, our FIRST Ichthyosis Parents Facebook Group weighed in, offering their very best advice for traveling with kids and ichthyosis.

Tub Tips:

- Bring along a blow up baby tub. It's easy to pack and you don't need to use the hotel tub.
- Always carry a battery-powered fan to keep kids cool, especially when activities are outside.
- Bring a big plastic container to pack things, and then use it as a tub as well when you are at your destination.
- Magic erasers and bleach wipes could also do the trick for a bath.
- Bring antibacterial wipes with you when you travel. You can use them for cleaning stations and bathtub.
- Consider switching to showers when traveling and resign yourself to the fact that kids may look a little "rough around the edges" without their same bath routines.

Swim Time:

- If you are going to a waterpark, check ahead to see if it's salt water. Chlorine may sting, but salt water could be great for the skin.
- Consider putting a time limit on "swim time," so your child's skin doesn't get too irritated. A maximum of 30 minutes was suggested.
- If it turns out your child is not enjoying the waterpark or other outdoor activities, be sure you have some back up indoor activites, too.

Keep Cool & Lotion Up:

- Squirt water bottles are a must!
- Be sure to lotion the child up immediately upon getting out of water. It will prevent tightening and peeling.

Interested in joining the conversation? Ask to join our FIRST Ichthyosis Parents Group. https://www.facebook.com/groups/FIRSTforParents/

Regional Support Network

Have you benefited from FIRST's Regional Support Network (RSN)? The RSN is a community of people who come together and support each other through the sharing of knowledge, experience and advice. These connections and relationships provide moral support, information, practical advice, guidance and resource information. One of the key components of the RSN is our Patient Support Forums which are one-day support meetings taking place throughout the United States and Canada. For more information contact FIRST's Sr. Director of Programs & Research, Moureen Wenik, mwenik@firstskinfoundation.org.

2015 Patient Support Forums

From Toronto to Minneapolis to Phoenix to Miami, FIRST's 2015 Patient Support Forums were a huge success - over 150 attendees in all! Patients, family members, doctors and FIRST staff gathered together for these unique one-day meetings filled with medical discussion, research updates, networking, product-sharing and more. FIRST would like to extend our deepest gratitude to all of the families, vendors, and medical professionals who participated in the meetings. Thank you for your commitment, enthusiasm and support. Please check our calendar of events at firstskinfoundation.org for upcoming ways to connect.

What You Can Expect at the 2016 FIRST National Conference-San Diego

An interview with FIRST member, Bhugwant Sethi, from 2014 National Conference in Indianapolis.

Q. What has the conference been like for you?
A. It's been a really emotional but also very hopeful and enlightening. We are seeing kids having such a good time and speaking with adults that are going through, or have gone through, the whole experience of having an affected child. It brings me lots of hope. As a parent, we always worry about our children, whether they have ichthyosis or not. You worry about how their future will be. This conference really reaffirmed that I had been thinking too negatively. It changed the way I think. There is no point to worry about all those things. Just take it one day at a time and everything will work itself out.

Q. Who have you met at the conference?
A. Meeting everyone made me feel even more hopeful about Ruhbaani's future. I met a granddad whose daughter is affected and their grandchild had no skin issue. I also met many of the veteran dads who have been around for many years, and they had so many good, positive things to say about their children. It's just beautiful, you know? To hear all of the stories, it just makes me feel that it will all be OK. And who knows, my daughter may get married one day, she might have children one day. I feel like no matter what, it will all be OK. It's been very comforting.

Q. Any final thoughts about the conference or your connection to FIRST?
A. This whole thing has been such a wonderful experience and I'm so happy we found FIRST. You know it's been our lifeline since Ruhbaani was born, ever since the beginning. I don't know what we would do without it.

2016 National Conference – *San Diego!*
June 24 - 26, 2016

Visit firstskinfoundation.org/financialaid for a financial aid application to the 2016 National Conference - San Diego.
The deadline for applications is February 1, 2016.

A Loving Family Finds a New Normal With Baby Daniel

In 2014, the Saylor family of Pennsylvania adopted a baby boy affected with harlequin ichthyosis. Their journey, from the very first moment they met Daniel, to today, where the happy one-year-old is making strides in his development, has been nothing less than amazing. We are excited to share the remarkable story of this kind and loving family, and delighted that they have connected with the FIRST community.

Mark and I first heard the word "ichthyosis" on July 17, 2014. About six months earlier, we had felt the call to adopt again. We had discovered there was a need for families who are willing to adopt infants with serious, even fatal, medical conditions. We felt that this was the perfect way to blend our desire to "look after orphans...in their distress" (James 1:27) with our desire for another child. We completed the mounds of paperwork, received our approval, and had only been waiting for a few weeks when we came across a Facebook post describing an infant boy in California with a "potentially life-threatening skin disorder." When we requested more information, we learned that this baby with harlequin ichthyosis (HI) was almost ready to be discharged from the hospital, so we needed to decide quickly. We prayed, we researched (Blessed By Brenna and FIRST were both very helpful resources!), and we said "yes!" On Tuesday, July 22, we received a call from California saying we had been "chosen" and could one of us get to California tomorrow?! After a flurry of reservations, babysitting arrangements, and appointment cancellations, I boarded a plane to Los Angeles.

After a harrowing rush-hour cab ride from LAX, I walked nervously down the hospital hallway, bursting with excitement at the thought of meeting my new son. I heard little whispers of, "She's here! The mom from Pennsylvania…she's here!" as I passed the nurses' station. Someone told me to don a gown and gloves, and I smiled awkwardly at the two Asian women in the hospital room, wondering which one was my son's first mom. I peered over the edge of the hospital crib and cried as I took in my beautiful baby boy dressed only in a diaper. He was covered in dark patches of thick skin, sleeping with his eyes open, and he was making those familiar baby squeaks as he stretched. Over the next week, I sat in on all the "rounds," learned about his medical care, signed several stacks of paperwork, and began the process of bonding with my baby boy. We decided on his name and the nurse changed the name tag on the crib from "Baby Boy Z…" to "Daniel." Daniel means "God is my judge," a sentiment that will be useful for Daniel as he grows up with ichthyosis.

That first week in the hospital, I talked on the phone with Courtney Westlake, whose daughter Brenna is affected with HI, and Hunter Steinitz, who is also affected with HI. They were both so helpful to us. Courtney gave little tips such as how to help loosen and remove those stubborn head scales that were the source of Daniel's "malodorous" state. She directed me to FIRST, to the Fasciano family, whose son Evan is also affected with HI, to Carly Findlay's Ichthyosis Awareness Month project, and to the Steinitz family. Hunter Steinitz answered my phone call so cheerfully and showed me that this fragile newborn in my lap might some day be a bubbly college student with a bright future.

One year old, Daniel is making strides in his development (rolling over, sitting unassisted, self-feeding little crackers) and entertaining us with his cute and silly personality. We have developed a "new normal," integrating Daniel's feedings, skin care, and doctor appointments into the family routine. There have been a few hospitalizations, but overall, Daniel is surprisingly healthy. We have been surrounded by love and support from our local medical community, our family, and our church family (who came together to fund Daniel's entire adoption!). We consider it an honor and a privilege to have been chosen for him, and we are so grateful that he was chosen for us. If you are on Facebook, you can follow Daniel's story at facebook.com/chosenfordaniel.

Congratulations to 2015 UFIRST Scholars!

FIRST is proud to announce this year's 11 scholarship recipients funded by the UFIRST Scholars Program. Congratulations to everyone and we wish you the best of luck as you continue your education.

Atique Ahmed

ARCI-lamellar ichthyosis

Government Gordon College Rawalpindi

Goals and aspirations: "I have a clear goal to achieve expertise in chemistry that can make me able to produce such drugs which are important for the treatment of cureless [sic] diseases, e.g., AIDS, ichthyosis."

Ryan Balog

congenital ichthyosiform erythroderma (ARCI-CIE)

LaRoche College

Goals and aspirations: "I would like to graduate college with my education degree in math and to become a teacher and to influence people's lives."

Erica Fedewa

epidermolytic ichthyosis

Eastern Michigan University

Goals and aspirations: "I want to receive my Bachelor's degree in nursing and eventually get my Master's degree and become a nurse practitioner and get involved with FIRST."

Maria Gad

ARCI-lamellar ichthyosis

Ohio State University

Goals and aspirations: "I aspire to learn as much as I possibly can and use that knowledge to help those around me. I aspire not to let anything, especially ichthyosis, to get in the way of living a life full of gratitude and happiness."

Quincy Harrison

x-linked ichthyosis

The Columbia University

Goals and aspirations: "I hope to become a psychiatrist and specialize in treating early-onset dementia and aiding teens with mental disorders caused by family related stress."

Elizabeth Joyner

ichthyosis vulgaris

Union University

Goals and aspirations: "I look forward to teaching, whether in a classroom or as a youth leader. My teaching degree will allow me to be certified for K through 8th grade and I hope to use what I have learned to encourage children in this age group that they can overcome obstacles and pursue their own goals, whatever they may be."

Jessica Lewis

lamellar ichthyosis

Georgia Regents University

Goals and aspirations: "I would like to become a Clinical Nurse Leader. I would like to work as a flight nurse and make a positive impact on those around me."

Austin Milam

ARCI-lamellar ichthyosis

University of Colorado, Colorado Springs

Goals and aspirations: "I want to attend a four-year university and possibly major in Biology. After college I plan on going into a career in Plant Science."

Kayla Murray

congenital ichthyosiform erythroderma (CIE)

Stanly Community College

Goals and aspirations: "I want to obtain my Associate of Arts degree from Stanly Community College and then further my education at a four-year university. I plan to obtain a Master's degree in library science."

Kyle Pappas-Adamson

epidermolytic ichthyosis

University of Kansas

Goals and aspirations: "I'm going to KU to study Sports Medicine and it is my goal to become an athletic trainer for a professional sports team."

Makaveli Williams

pachyonychia congenita

Davenport University

Goals and aspirations: "I would like to go to college and get at least a Master's degree and make the Dean's List in college."

Ask Dr. John

John C. Browning, MD, FAAD, FAAP
Assistant Professor, Baylor College of Medicine
Chief of Dermatology, Children's Hospital of San Antonio

Have a question about living with ichthyosis? Now you can ask Dr. John. Each issue of the newsletter will feature a common (or sometimes not-so-common) question regarding living with ichthyosis. The questions are submitted directly from a member, and an answer will be provided by FIRST Medical and Scientific Advisory Board member, Dr. John Browning.

Do you have question for Dr. John?
Send your inquiry to Maureen Neville at mneville@firstskinfoundation.org.

Ichthyosis and Exfoliation:

I often get lots of questions from my ichthyosis patients about exfoliation: Should I exfoliate? Is it good for my skin? How often? The information below might help to shed some light on this very topic.

Answer:

First of all, what is exfoliation? Webster's Online Dictionary defines exfoliate as "to cast off in scales, laminae, or splinters." So exfoliation is the act of removing the outer scales from the skin.

For those individuals with hyperkeratotic ichthyosis (i.e., thickened skin), exfoliation may make you more comfortable. These might be people with CIE, harlequin, or epidermolytic ichthyosis. Of course, with exfoliation you remove the scale, but it often brings out the underlying erythema or redness. So you will trade the scale for the redness. This is not an issue if the scale is more bothersome to you, but it is important to keep in mind if you do not want to be red. In other cases, such as Netherton syndrome, the skin barrier is impaired and should not be further exfoliated. Exfoliation will only lead to increased sensitivity and irritation.

Some individuals with CIE or epidermolytic ichthyosis prefer to exfoliate by first soaking in a tub for an hour or longer. They can then use a washcloth or loofah brush to gently remove the outer layer. Others might prefer a cream or lotion with ammonium lactate, salicylic acid, or urea to help soften and remove the outer layer. Those with thick scale under their eyes have found that topical tazarotene can loosen the scales and allow their eyelids to close more fully. Whatever the method, it is an individual choice, and you need to find what works best for you. Best wishes to all of you for a happy and comfortable summer!

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

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Registration information will be available in January 2016. We'll continue to share updates throughout 2015 and 2016.

San Diego Marriott Mission Valley
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San Diego, CA 92108 619.692.3800