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

Ichthyosis Focus

Summer 2013 | A Quarterly Journal for Friends of FIRST | Volume 32, No. 2

Few & Far Between

An interview with Meredith Rizzo Photo/Video Journalist

Every once in a while, "perfect timing" is more than just an eruption of coincidental moments…it is, in fact, destiny. At least, that was the case when young photo journalist Meredith Rizzo set forth to create her final thesis for a Master of Arts degree in New Media Photo Journalism from the Corcoran College of Art & Design.

"The assignment for our thesis was to submit a long form body of work," said Rizzo when describing the very first steps of her journey. "Around that same time, I learned that there would be a FIRST family conference in Denver. So, I decided to go to the conference and see if I could document someone affected with ichthyosis."

Continued on page 8

Inside This Issue:

Vol. 32 No. 1 Summer 2013

Ichthyosis Focus is published quarterly
by the Foundation for Ichthyosis &
Related Skin Types, Inc.®

The Foundation for Ichthyosis
& Related Skin Types, Inc.®
2616 N. Broad Street
Colmar, PA 18915

Phone: 215.997.9400
Toll-free: 800.545.3286
Fax: 215.997.9403
Email: info@firstskinfoundation.org
Web: www.firstskinfoundation.org

The Foundation for Ichthyosis & Related Skin Types, Inc.® is a 501(c)3 charitable organization supported by public and private donations. All contributions to the Foundation are tax deductible to the full extent of the law.

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

Ichthyosis Focus is provided as a service to members of the Foundation as a medium for the free exchange of information. Neither the Foundation for Ichthyosis & Related Skin Types, Inc.® its Board of Directors, its Medical Advisory Board, nor the Focus Editors endorse any treatments or products in Ichthyosis Focus. Views and opinions expressed in this publication do not necessarily reflect the views of the Foundation or Foundation officials.

A Fresh Face, A Fresh Look

Jean R. Pickford, Executive Director

FIRST embarked on another "first-time ever" event — adding to our constantly evolving history. With the help of a wonderful group of board leaders, we searched and hired a much-needed Communications Director for FIRST. This past April, we introduced Maureen (Mo) Neville on our website, along with Facebook and Twitter announcements, welcoming her to our dedicated staff and wonderful ichthyosis community. It's been quite a whirlwind for everyone, especially for Mo. Her "to do" list grows longer every day. Hopefully, you've seen the changes and influence Mo has had in our public communications in just the few short months she's been at FIRST. If you haven't read any of her work please see pgs. 8-12. There is also one particular change I want to highlight, which is relevant today as you are reading our summer issue of the Ichthyosis Focus. Mo has secured a new design team and printer for the newsletter, which continues to be one of FIRST's most popular resources. Not only do we have a new sleek and inviting look, but the production costs went down as well! Needless to say, Mo is shaking things up, and you can look forward to more compelling news and community outreach through the communications generated from our office. It's just another way we are keeping FIRST as the leading ichthyosis authority in the country, if not the world.

Have an idea or want to connect with Mo? She'd love to hear from you– mneville@firstskinfoundation.org.

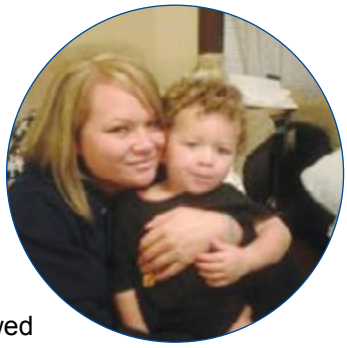
Look at Us Grow!

Volunteerism & grassroots fundraising efforts truly blossomed with the arrival of May, Ichthyosis Awareness Month. At home, online, and everywhere in between–members were more enthusiastic than ever about getting involved! Kudos to all who rolled up their sleeves, got creative, and tackled the tasks of spreading the word and local fundraising.

Tastefully Simple Online Fundraiser

FIRST member Kelly Pelton-Robinson took a fun and creative approach to raising donations, by holding a month-long Tastefully Simple fundraising event on Facebook. Not only did participants purchase delicious treats, from tasty appetizers to luscious desserts, but Kelly also raised $1,077!

Bake Sale



FIRST member Mannessa Elliott held a scrumptious bake sale at the Pub on the Cedar Restaurant in Charles City, IA. Her fabulous baked goods were the talk of the tavern and raised a whopping $1,053! She was also interviewed at KIMTTV! Thanks, Manessa!

E-bay Vacuum Auction!

Going once…going twice…sold! A unique fundraising tactic, FIRST member Susan Schumacher auctioned a high-end vacuum cleaner off on Ebay. And although her original goal was to raise $650, she actually exceeded it, raising a total of $750! Great idea, Susan!

Evan's FIRST T-Shirt Sale

FIRST member DeDe Fasciano held her son Evan's first FIRST Fundraiser–a t-shirt sale! The t-shirts featured a singular, heartfelt phrase to live by: "Sometimes We Live No Particular Way But Our Own." Donations totaled over $1,000!

FIRST Family Dance-a-Thon

The Hodgkinson Family held a FIRST Family dance-a-thon, preceded with wide-spread local press, including television, radio and newspaper. In the end, they raised over $5,000! "The attendees had a blast! One boy even asked his dad to check the newspaper for other dance-a-thons they could go to the next weekend," said Sarah Hodgkinson with regard to the event's success. Way to go, Sarah!

Indoor Garage Sale

Needless to say, there were plenty of hidden treasures at member Allison Lyons' garage sale, as funds raised for the day were over $6,000! Great job Allison! "It feels great to increase awareness as well as help with fundraising," said Allison with regard to her success. Peyton Lyons and friends also raised $76.75 at their duct tape fundraiser that same day. Great job!

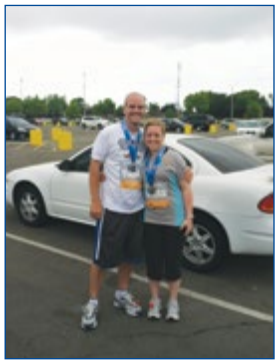
4th Annual Wine Tasting event

The Cina Family hosted a spectacular Wine Tasting Event, right in their own home. Friends and family enjoyed sipping wine, learning about its taste & texture, and, most of all, making a difference. Donations for the evening totaled $2,585.

FIRST Collection Jars & Community Outreach

A tremendous thanks to FIRST members Nancy Osentoski, Curtis Tober and Alicia Morales who spread the word door-to-door by placing collection jars throughout their community. Collectively, they raised over $25,000 in May! And a special thanks to Diana Gilbert, who spread awareness by handing out FIRST brochures throughout her entire community.

Running For Research



FIRST member Jessica Barney planned an Orange County Marathon fundraiser with the first mile of the marathon taking runners along the beautiful Pacific Ocean, winding through the seaside village of Corona del Mar and eventually greeting the finish line at the bluffs that overlook the Upper Newport Bay Estuary Reserve. Runners were not only accompanied by the breathtaking terrain, but were also greeted by cheers of neighborhood residents. Donations totaled nearly $1,500! Congratulations Jessica!

Enchanted Ball

It was an evening of sparkles, charm, and enchantment. Amanda Jillson planned a gala to remember in Springfield, IL. Guests of all ages were invited to join the fun and fairytale at the Enchanted Ball. Two hundred were in attendance. Proceeds benefited FIRST & St. John's Children's Hospital. It never ceases to amaze how supportive a good friend can be.

Ace in the Hole Foundation

For the second year in a row, Ace in the Hole Foundation, a charity founded to remember and honor the sacrifice of 1st LT Michael LiCalzi USMC, named FIRST as one of the official beneficiaries for their Lido West Beach, NY run. This year, there were 400 participants and the foundation raised over $80,000, to be distributed amongst the benefiting organizations. Congratulations to all who participated! We feel extremely honored and privileged to be chosen as one of the beneficiaries. For more information on this extraordinary foundation: www.aceintheholefoundation.org.

Upcoming Events

2nd Annual Dane's Friends for FIRST Concert

Saturday, September 7, 2013

The Phelps Family will be holding their Annual FIRST Concert from 4:30 pm until 9:30 pm, at the Flint Center for the Performing Arts, in Cupertino, CA 95014. Dane's Friends for FIRST Concert is a fundraising event that was inspired by the life and memory of 3 1/2 year old, Dane Phelps. Once again, there'll be some amazing performances and fabulous raffle/auction items planned for the night. This is an event you won't want to miss. Contact Cwassel@firstskinfoundation for details.

Correspondence Corner

Dear Jean,

The work that is being done by FIRST is so wonderful. Our son, David, was born with a collodion membrane and subsequent mild ichthyosis. Needless to say, this has affected our lives ever since. At that time, very little was known or available to us. The progress you have spearheaded is remarkable! We will certainly continue to support FIRST and all its wonderful works. The newsletter is always read and appreciated.

Thank you again.
Anonymous

Dear FIRST,

I was touched when the Foundation for Ichthyosis & Related Skin Types asked to run one of my favorite blog posts to date on their own blog. This month, I got to speak with two different staff members at FIRST-out of the 6 they have on staff! I learned more about FIRST, its mission and how I can become more involved, and I hung up the phone as inspired as ever to continue to raise awareness about ichthyosis and physical difference. I also feel gratitude in knowing that we have a wonderful, very dedicated foundation behind families like ours who are affected by ichthyosis.

Courtney Westlake
Springfield, Illinois

Members Took Social Media by Storm!

Kitty Morris took a unique approach to IAM by posting daily "day in the life" posts to Facebook about her adorable ichthyosis princesses. Here's one of our favorite posts…

Tech- savvy members raised awareness to new heights.

The Blogosphere was on fire all month long! The already popular blogs from members Dede Fasciano, Courtney Westlake, Carly Findlay, Rachel See, were all the rage! From informative and enlightening to provocative and heartwarming, their posts, photos, and videos, raised awareness, funds, and inspired new connections all over the world. Great job, bloggers!

Member Patty Sundik created a special–editorial style Pinterest page, including posts of all the special member stories shared by FIRST, throughout the month. **http://pinterest.com/lovesparis/iam-ichthyosis-awareness-month-first/**. Plus, Patty sells posters of her grandson's drawing (SMILE) on her online Etsy shop **www.lovesparisstudio.etsy.com** to help raise funds for EI research. One hundred percent of the sales from the posters goes to FIRST, and ten percent of her total sales goes to FIRST every year. We thank you for your creative, and generous spirit, Patty!

Take Me Out To The Ball Game
(and don't forget Tommy!)

FIRST's friend Tommy is the MLB's biggest LITTLE FAN!

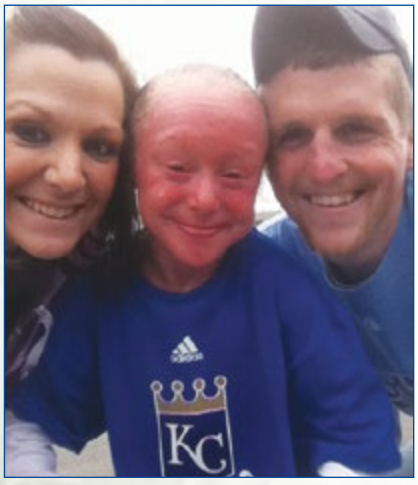
Tommy is on the move! In an effort to broaden ichthyosis awareness, Suzanne & Eric Phelps have enlisted the help of FIRST to reach out to every team in both the National Football League and Major League Baseball. FIRST has even taken Tommy along to every industry conference this year!

Tommy, "the MLB's biggest little fan," is currently traveling via the US Post Office to every MLB team with the goal of gaining each team's support and a donation of a team jersey or other item. After all donations are received from both the NFL and MLB, they will be auctioned off at the Dane's Friends for FIRST 2013 event. (See: 2nd Annual Dane's Friends for FIRST Concert September 7, details on pg. 4.)

First Nights with MLB

New York Mets – The Benedetto and Saccente families spearheaded the entire evening, inspiring participation and gathering on the field with the Mets before the game. They generated $2,925 from 350 tickets sold ($4 per ticket back to FIRST) and many donations. Way to go, team Benedetto and team Saccente!

Pittsburgh Pirates – The Kocher Family "hit it out of the park!" with their first FIRST fundraiser. The pre-game tailgate party was a big success and member Hunter Steinitz represented FIRST on the field that night. The event generated $4,200 ($5 per ticket back to FIRST) including a donation match by the president of the Pittsburgh Pirates, Frank Coonelly. Well done Kocher, Family!



Kansas City Royals – This event was led by Russ and Cora Cossel who generated $2,200 ($10 per ticket back) and several donations. In fact, they sold out an entire section of Kauffman Stadium to watch the Kansas City Royals take on the Chicago White Sox! Thank you kindly, Cossel Family!

Medically Speaking

Bathing Completes the Shedding Process

It's true! Bathing may even be more important to the shedding process than previously considered, as it does not only cleanse our skin of dirt and other external debris, it completes the natural process of desquamation, sweeping away spent and finished epidermal cells.
So…what exactly is desquamation?

The epidermis is a self-renewing system. Old cells (or "squames") are shed from the skin surface as new cells ("keratinocytes") are produced in the underlying epidermis and pushed outward into the stratum corneum to become "corneocytes". This process is known as desquamation.

In the recent past, it was thought that the process of desquamation initiated the progressive breakdown ("proteolysis") of proteins forming structures called 'corneodesmosomes'. These protein forming structures link adjacent 'corneocytes' to one another. We now show that this prior model does not fully account for desquamation, because the cells detach well above the sites where these junctions are degraded. In other words, proteolysis of corneodesmosomes may be necessary for desquamation, but it is not sufficient to complete the entire shedding process.

Instead, recent studies show that cells detach following hydration! Water swells the extracellular spaces and separates adjacent corneocytes. As the water evaporates from between corneocytes, it is replaced by air, which eventually allows individual cells to detach from the skin surface.

What does this mean for the ichthyosis community?

There's no question–for those affected by ichthyosis, frequent bathing is even more important, as it is not only the best way, but the most natural way to remove dry scales and skin. Most importantly, frequent bathing may make people with ichthyosis much more comfortable.

Resource: Peter M. Elias, MD. **http://eliasandwilliams.com/** what-allows-skin-to-shed/ For more information on the dermatology research, see **http://eliasandwilliams.com/?s=FIRST**, a website devoted to the permeability barrier, the most critical, life-enabling function of skin.

Who Knew? A Neural Circuit Just for Itching

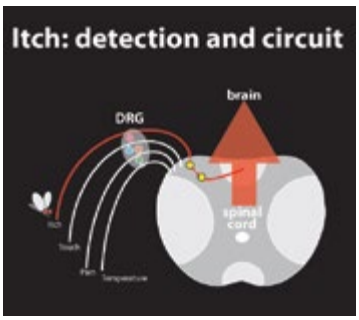
By Dr. Francis Collins, Director of NIH on May 23rd, 2013

Itch-inducing agents activate a discrete population of peripheral sensory neurons that produce a signaling molecule called natriuretic polypeptide b (Nppb). The release of Nppb from these primary pruriceptive neurons triggers a dedicated itch biocircuit to generate the sensation of itch.

The occasional itch—be it a bug bite or rash—is annoying. But there are millions of people with chronic itching conditions, like eczema, psoriasis, and forms of ichthyosis who are constantly scratching their skin. This is more than a little irritation—it drastically reduces their quality of life and is a perpetual distraction. Current anti-itch treatments include topical corticosteroid creams, oral antihistamines, and various lotions. But researchers at NIH have gone beyond the skin's surface and discovered a critical molecule at the root of that itchy feeling [1].

Until now, it's been unclear how the sensation of itching was carried to the brain. Was this a separate system, or did it use the same nerve pathways as pain, touch, or temperature? To find out, the researchers focused on a special group of nerve cells called "TRPV1 neurons," which extend to the skin and detect temperature, various types of pain, and itch. These neurons use several chemicals to transmit signals; one of those is a neurotransmitter called Nppb. When the researchers created mice lacking Nppb, the mice became immune to itching. Even when the researchers exposed the mice to several itchy substances, such as histamine, the mice refused to scratch!

When the researchers actually removed specific spinal cord nerves that receive the Nppb itch signals, other sensations—like pain, touch, and temperature—remained intact. So we now know that in mice, and possibly in humans, there are specific nerve cells and brain circuitry that are entirely devoted to that itchy feeling. That means blocking Nppb could turn out to be a safe and effective strategy to cure itching, especially in chronic cases.



[Images courtesy of Mark Hoon, National Institute of Dental and Craniofacial Research, NIH]

Reference:

[1] The cells and circuitry for itch responses in mice. Mishra SK, Hoon MA. Science. 2013 May 24;340(6135):968-71.

NIH support: National Institute of Dental and Craniofacial Research

Few &

"In the end what I learned is that the story was really not about our differences, it was about our shared connection."
– Meredith Rizzo

Far Between

...continued

Meredith's father, Dr. William Rizzo, a current member of FIRST's Medical Advisory Board, is a geneticist specializing in inherited metabolic diseases at the University of Nebraska Medical Center. He has been researching Sjögren-Larsson Syndrome for the past 25 years.

As ichthyosis is one of the symptoms of Sjögren-Larsson Syndrome, the skin disorder itself was not unfamiliar to Rizzo, and in fact, she worked at the research lab with her father for an entire summer as an intern. But now, as a curious and conscientious journalist–she was interested in hearing about the disorder, directly from the source. She hoped that one of the affected families would be open to sharing their routine, their challenges; and the bull's eye of all inquiries: how the diagnosis of ichthyosis has changed their lives.

"I met the Chappells on the very first day of the conference. Their daughter Mia not only has ichthyosis, but also has Sjögren-Larsson Syndrome–the very same syndrome my dad has researched for all these years. So it was really great to meet someone whom my dad's research has affected. The disease is so rare, that doesn't really ever happen."

As described in Rizzo's essay:

Sjögren-Larsson Syndrome is a syndrome so rare its incidence is unknown, but doctors estimate around 100 people in the United States live with it. For Mia, neurological impairment associated with the syndrome means that she is still learning to walk, climb stairs, and hold herself upright. Her mental progress is two years behind that of other children her age, and she has ichthyosis: dry, scaly skin– a lifelong condition that will require a daily regimen of scrubbing and lotions.

Meredith Rizzo

Rizzo would soon discover that the syndrome, itself, may have changed the daily routine and future plans for the Chappells–and yes, they are learning to deal with the constant boomerang of ups and downs, but in truth, the disorder has really nothing to do with *their life.*

Fate Takes its Next Cue…

"The Chappells were so open to the idea right from the beginning," said Rizzo. And then fate stepped in again. The young woman whose father, a gifted geneticist, was researching one the rarest syndromes known to man, was being led to use her own gifts and raise awareness for that very same syndrome. "Coincidentally, they live only 45 minutes from me, so I was able to really connect with them pretty often over the course of the next ten months."

Rizzo spent time with Mia in nearly every aspect of her life: at her school, capturing the interaction with classmates; at one of her physical therapy appointments; and, on a few occasions she even quietly accompanied Mia during her morning and evening skin care regimen with Mom and Dad. "Whatever she was doing or wherever they were going when I went to their home, they invited me to come along and document what was happening. I even tagged along on Halloween!"

One particularly painful milestone was the day Mia visited the Orthotic Prosthetic Center in Fairfax, VA for the very first time. "The photos from the day she went to the doctors and got fitted for her new leg braces really capture some of the medical challenges they are dealing with."

The story behind the story was beginning to appear. "It was remarkable how much of their lives they shared with me. But then I realized, I was giving them a chance to talk about their situation, because, so often, they are simply not asked."

The Universal Lesson…of Love

Ten months later, she had gathered the pages of a story that had become not only a school project, but a mission.

During the final stages, Rizzo reached out to a connection she had made at Corcoran. "A mentor of mine is actually the photo editor for National Geographic Magazine. She introduced me to their graphic designer, who helped to format the whole photo essay."

Meredith's desire to tell an untold story and infuse it with "the energy of the subjects themselves" was quickly manifesting into a uniquely personal piece of work. Combining text, video and photos, and crafting an online photo essay, and e-book, that reflects not only the deep and profoundly human experience of the Chappell family, but of the journalist herself.

"I went into the experience thinking I would write about how the family deals with symptoms, like itching or immobility. I would reveal how their lives were so different than everybody else's. But in the end, what I learned is the story was really not about our differences; it was about our shared connection. The story of their lives isn't about Sjögren-Larsson Syndrome; it's about a family's love for their daughter…and that's universal." ●

Photos by Meredith Rizzo. To view Meredith Rizzo's photo essay, visit fewandfarbetween.net, or download the e-book: http://www.fewandfarbetween.net/the-ebook-download/

Participant Recruitment for Research Study on Sjögren-Larsson Syndrome

Sjögren-Larsson syndrome (SLS) is a rare form of ichthyosis characterized by the co-presence of neurologic symptoms, such as spasticity and intellectual disability. Not enough is known about SLS and its complications, and therapeutic options are limited. The Rare Diseases Clinical Research Network, funded by the National Institutes of Health, is supporting a research study on SLS to gather clinical information about its natural history and search for biomarkers (tests) that can be used to monitor future therapy. We invite you to participate. SLS patients and at least one parent will be asked to travel to one of three participating study centers [Omaha, Portland (Oregon) and Pittsburgh] and take part in an extensive 3-day clinical evaluation. Costs for travel, housing, and medical tests will be covered as part of the study. This is a unique opportunity for SLS families to contribute to research on this disease. Please contact Dr. William Rizzo at the University of Nebraska Medical Center for more information (telephone 402-559-2560; email wrizzo@unmc.edu).

Jane & Henry Bukaty Skin Care Fund

Financial Assistance for Ichthyosis Treatment

Thanks to the generosity of donors Jane & Henry Bukaty, FIRST 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 applicant will specify the product/treatment for which funds are needed and demonstrate his/her financial need for this product/treatment.

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

Applications can be downloaded from our website at www.firstskinfoundation.org and emailed to the national office or mailed to the address below.

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

The next cycle of funds awarded will take place in July 2013.
The deadlines for submitting an application are July 31 and January 31 each year.
You will be contacted by the office if you have been awarded aid from this fund.

Making Connections

As seen in our new blog!
Blog.firstskinfoundation.org

By Mo Neville, Communications Director, FIRST

Raising Awareness for Rare Disease… One Cape at a Time

If you search the internet for the origin of the "superhero cape," as I just did, you might find yourself lost in a cyber-sea of Superman quotes, Twilight tween-tweets, and an oddly expansive selection of Zorro fan pages and Batman vs. Green Lantern public debates. I suppose now I am well equipped with unique and witty cocktail party conversation–particularly on Halloween–but more importantly, I am also certain that the superhero cape, regardless of its origin–has grown into a universally iconic symbol, summed up in a single word: adventure.

For Robyn Rosenberger, founder of "Tiny Superheroes," life, lately, has been nothing less. I had the great pleasure of speaking with this unlikely seamstress, who is quickly becoming an icon in her own right.

"I am not a seamstress by any stretch," Robyn said when describing her epiphanic moment. "During the summer of 2012, I made three capes. One for my son, my nephew, and my dog–just out of the blue, just for fun, and they loved it." That same summer, she had also been following the blog, Blessed by Brenna–a weekly chronicle, authored by a young mother who happened to be an old schoolmate of Robyn's husband–and who had also just given birth to a baby affected by Harlequin Ichthyosis.

Then, one day while perusing the Blessed by Brenna blog, it happened–the "Aha" moment that would change Robyn's life….

Brenna needs a superhero cape because she is truly extraordinary!," Robyn thought as she rolled up her sleeves and began to assemble the fourth little cape she had ever sewn in her life.

However, when the story of Brenna's cape broke in the blogosphere on both Robyn and Brenna's blogs, it was as if the whole world was watching. Unbeknownst to Robyn, when she had sewn that last little stitch on Brenna's little cape, she had also begun the thread of something truly extraordinary herself–an organization that would empower children affected by rare diseases all over the world.

"We decided to set up a page on our blog where we could take nominations for the next Tiny Superhero to receive a cape." Her blog, Tiny Superheroes, now receives nearly 20 nominees a day, from a vast range of rare genetic disease communities, the world over.

"We seem to have a lot of interest from the ichthyosis community. It has been so wonderful getting to know these kids. And what's been really amazing is that the children are all so different, so special in their own way."

Tiny Superheroes has been featured on Today.com, Fox News, Evening Magazine, the Huffington Post, and World News Tonight. But the most surprising fact? The entire organization officially opened its doors just four months ago, in January 2013. "Now we're even getting messages from people all over the world who'd like to volunteer and help us sew capes. I'm trying to figure it all out as it comes–it's really incredible how quickly it's grown–but it seems to be all working so far."

The second most surprising fact? In those mere four months, Robyn, and a small army of volunteers, have sewn nearly 500 capes–(although Robyn alone sewed 300 of them.)

Admittedly, this was not a lifelong dream of Robyn's, as she had no experience, nor grandiose vision of one day helping to raise awareness for rare diseases. "It's like the kids chose me, and it's taken on a life of its own. I feel so privileged and I couldn't be happier about where everything is going."

By all accounts, the kids chose well.

However, Robyn now knows that she may not have been chosen to simply sew hundreds and hundreds of tiny capes. "What I realized is that this work can help kids raise awareness for their own disease - which is really powerful. It's much more than giving them a cape. It's letting these kids know they can change the world. And I believe they can."

And there you have it–no searching, nor super power, necessary. Sometimes a hero really does just…come along.

For more on volunteering, donating, or nominating a Tiny Superhero go to **www.Tinysuperheroes.com.**

What is Ichthyosis?

The answer may surprise you.

By Mo Neville
Communications Director
FIRST, Foundation for Ichthyosis and Related Skin Types [COOLIBAR GUEST BLOG]

When someone discovers that I am the Communications Director for FIRST, the Foundation for Ichthyosis & Related Skin Types®, typically, I am greeted with the same three questions: "What is ichthyosis?"; "Is someone in your family affected?"; and…"What brought you to FIRST?" From a clinical perspective, I may respond by saying: ichthyosis is a family of genetic mutations of the skin characterized by dry, scaling skin that may be thickened or very thin, and most forms of the disorder are very rare. Each year, more than 16,000 babies are born with some form of ichthyosis, and it affects people of all ages, races, and genders. Ichthyosis may also severely affect someone's emotional health, and it can lead to a host of other illnesses, such as bacterial infection, heat stroke, and immobility.

And, until these past few weeks, I have not known anyone on a personal level that has been affected with ichthyosis.

However, there is more to the story – much more. So what is ichthyosis? It is a genetic skin disorder, yes, but it has also been the catalyst that has led me on a worldwide journey of meeting the bravest and most inspirational people of my life – a young mother from Illinois, whose baby was born with ichthyosis, who has not only accepted her fate of grueling skin care routines, a daily parade of doctors and therapists, and caring for an infant through unprecedented surgeries – but considers it a wondrous blessing; one that she is delighted to share, daily, with the entire world. The young woman from Australia with ichthyosis who has turned her strife into a worldwide "appearance advocacy" effort, changing the way people perceive disability by offering words of empowerment and guidance to all those who "look different" everyday. The teenager from Canada with ichthyosis who only recently shared with us his moment of enlightenment – when he finally realized that because he beat the odds of not "dying by age three," as doctors had predicted, his life was not a merely ordinary existence, but a destined one. Or, the twenty-year old woman from Arkansas who is the second oldest living individual, with one of the most severe mutations of the disorder – who has not let ichthyosis steal a single moment of her life – and in fact, has just given birth to her first child. And of course, the team of medical doctors, stowed away in a clinical laboratory at Yale University, for nearly 31 years, fueled by one and only one thought: finding a cure. Each day, they defy the odds and travel their paths alongside a disorder for which, presently, there is no cure. And they never lose sight that accompanying their every step is hope, support, and the tireless team of advocates at FIRST.

In fact, it is their hope that is the driving force behind the mission of FIRST – to educate, inspire, and connect all those affected by ichthyosis. It is their hope that inspires this 31 year-old organization, that has affected so many lives, to be the only national non-profit foundation located in the United States dedicated to assisting families affected by ichthyosis; the only patient advocacy organization funding medical research specifically targeted toward finding advanced treatment as well as a cure, and an organization that has funded 13 projects granting over $1.4 million in research since 2006.

And it is their hope that expands the definition of ichthyosis to the far corners of the human experience – far beyond the diagnosis of a rare genetic disorder.

So what is ichthyosis? It is a message; a call for human compassion; an invitation to experience the authentic awareness of "other." It is the doorway to unprecedented medical advancements and, most remarkably, it has become an opportunity for worldwide connections…and the chance for global unity.

But the question that is my absolute favorite to answer is this, "What brought you to FIRST?" If only in these moments I had glamorous, compelling tale of high seas adventure that led me to their door – but actually, I do not. In truth, the position for Communications Director was actually posted online. The beginning, clearly, is not exactly what you may call riveting. However, as I would not know for several months whether or not the position would be mine, I had the opportunity to discover that this organization was filled with the extraordinary. And what I can also tell you is that I never dreamed, when I was first invited to join the staff, a mere seven weeks ago, having neither friend nor family member affected with ichthyosis – that I would be brought into a community that has felt like the warmest embrace a family could offer.

It is a privilege to work alongside this team of intelligent, passionate and unwavering optimists, and I will be eternally grateful for the opportunity to meet all of the unforgettable individuals with ichthyosis; to answer questions about the disorder and to spread the word about our work, our mission and, above all, to raise awareness for this rare disease, so that someday I may be answering the question: When did they find a cure for ichthyosis?

The Foundation for Ichthyosis & Related Skin Types, Inc.® (FIRST) is invited as a guest blogger and does not endorse or serve as a product representative for Coolibar.

A New Beginning

RE: Ayra Ghorbani

(from our Ichthyosis Awareness Month member story series.)

A New Beginning by Ayra Ghorbani

On the second of July 1996, I was born with Netherton Syndrome. Doctors had observed and predicted I wouldn't make it past the age of three. They also thought that I would be put on tube feeding for the rest of my life. I'm glad to say …they were wrong.

Now that I look back after sixteen years, I realize how simple it could have been for me to die rather than live. I mean, I could have been with the others that didn't make it, missing the experiences this world offers. But my life must have been, and is, pre-destined. There must be a reason that I'm alive and writing this to you today.

"...my life must have been and is predestined"

Throughout my life I've always wondered why I was born this way. There are so many kids with "normal" skin. They always seemed like they were happier than I was, along with their friends. It was tough. Everywhere I went kids my age, and younger, stared at me, saying things like "Mommy why is his skin red?" Or maybe something like, "Wow he is so red!" These things really hurt me inside, lowering my self-esteem and confidence by a mile. I just couldn't seem to find any good that came out of my life. It hurt me more so when I was with my friends outside; watching my friends' reactions to all the people staring in their direction, knowing the people were actually looking at me.

"Throughout my life I've always wondered why I was born this way."

It was only in 2012 that things changed a lot for me. I came to focus on all the good in my life, as opposed to the bad. I started to see all the pieces fit together for my benefit. Sylvester Stallone said in his movie Rocky, "Let me tell you something you already know. The world ain't all sunshine and rainbows. It's a very mean and nasty place and I don't care how tough you are it will beat you to your knees and keep you there permanently if you let it. You, me, or nobody is gonna hit as hard as life. But it ain't about how hard ya hit. It's about how hard you can get hit and keep moving forward. How much you can take and keep moving forward. That's how winning is done!" It was as if the sun appeared after years of rain and storm. This all happened when I surrendered and faced the fact that this was how I was born. In simpler terms, my mind-set completely changed from going in reverse, to accelerating a hundred miles an hour ahead. I felt like what should've taken me years to figure out happened in only a small portion of that time.

"It was as if the sun appeared after years of rain and storm."

Let me just embolden you to not look back to yesterday; look to your future. There's a saying, "yesterday was history, tomorrow is a mystery"–look forward and never turn back to the old. Instead, pursue the dreams that have taken root in your heart. It's never too late to change. I promise you'll never regret it.

Currently, I am in grade eleven in my school at Lions Gate Christian Academy. I've been playing the piano for almost nine years now and have a black belt in kickboxing. I know I'll go on conquering obstacles, continuing to walk in the fullness of blessings, and you, will too. Shake off the misery and unhappiness. Start each morning giving thanks for all the good in your life. Believe and hope that something awesome is coming your way. I've learned, just before something great happens in life, there are usually hardships and obstacles we face. When those thoughts come to you, don't meditate on them. Your future is too bright to waste on negativity and low confidence level. If you focus on all the good in your life, believing for the greatest, then I know and declare: dreams will come true, healing will take place, and that all of you will become world-changers. Your best days are yet to come. This is a new beginning, a new chapter in your life. God Bless you.

"...pursue the dreams that have taken root in your heart. It's never too late to change."

-Arya Ghorbani

UFIRST Scholars

Congratulations to 2013 UFIRST Scholars!

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

Atique Ahmed *Lamellar Ichthyosis*
New Age Scholar Science College
Goals and aspirations: "I am affected with ichthyosis and it's been a major barrier for me. I want to break this chain and want to show my worth. I want to research in chemistry that can benefit human kind."

Elizabeth Joyner *Ichthyosis Vulgaris*
Union University
Goals and aspirations: "I look forward to teaching children in an elementary school setting. As I have had the opportunity this first year of college to observe in local classrooms. I hope to be able to use what I learn to encourage students and let them know they are all special in God's eyes and each one can learn in their own special way."

Jack Leahey *Lamellar Ichthyosis*
Princeton University
Goals and aspirations: "I would like to be accepted to the Woodrow Wilson School of Public Policy at Princeton and walk on as a member of the cross country team."

Jessica Lewis *Lamellar Ichthyosis*
Georgia Regents University
Goals and aspirations: "The main goal in life that I wish to accomplish is to make a positive impact on every life that I come in contact with day to day. I hope to one day become a Flight Nurse because my passion in life is helping those around me and making a positive difference in their lives."

Gina Messer *Lamellar Ichthyosis*
UC Davis
Goals and aspirations: "Become a successful psychologist, help others through my career and in general, get married, and just live a good, fulfilling life."

Brandon Romero *Lamellar Ichthyosis*
University of Arkansas for Medical Sciences
Goals and aspirations: "To find a cure for ichthyosis, whether through my research this summer with Dr. Keith Choate or later in my career; maintain a 4.0 GPA throughout medical school; challenge myself every day to continue learning and to become the best in my field that I can possibly be; make a difference in others' lives."

Kathleen Smith *Lamellar Ichthyosis*
Lawrence Memorial Regis College
Goals and aspirations: "To be a good person. To become a Nurse Practitioner and open a hospital and nursing school in developing countries. To help increase access to medicine in underserved areas and empowering and educating local people as health workers in their own communities."

Hunter Steinitz *Harlequin Ichthyosis*
Westminster College
Goals and aspirations: "I want to study theology in college and use that along with my ichthyosis to help people understand that it is good to be different."

Mani Woodward *X-linked Ichthyosis*
University of Oregon
Goals and aspirations: "I would like to earn degrees in Human Physiology and Chinese at the U of O and then continue to medical school where I would like to pursue a career as a pediatrician or a dermatologist. "

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

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

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

Meet Stephanie Turner's Healthy BABY BOY!

"This is a very exciting time not only for the Turners but for the entire Ichthyosis community. It offers hope that those affected with Harlequin ichthyosis are certainly leading happy and fulfilling lives." – Jean Pickford, Executive Director.

Many of you are aware that Stephanie Turner of Little Rock, Arkansas has the rare and life threatening skin disorder, Harlequin Ichthyosis, for which there is, at present, no cure. But Stephanie Turner has something that is quite possibly, even more rare: a life-loving spirit, for which there is presently no limit! In fact, one look at her mile-wide smile and there is no doubt this sweet mom from Little Rock knows, despite her skin disorder, she is one lucky lady.

"I haven't let my skin hold me back at all. I'm proud to say I was a cheerleader for four years, in girl scouts, ballet (for a whopping 2 weeks!), and homecoming maid 4 years in a row! I'm very outgoing and I go out often (that was before I got married, now I'm like an old woman!)," she recently joked on her blog, lifeasusdotcom.blogspot.com.

The blog–born from the perspective of the second - oldest person, known to FIRST in the USA to be living with Harlequin Ichthyosis, whose days are filled with complexities that most of us will never know–is an unexpected burst of positivity and, by all accounts, is the quintessential diary of "the girl next door". Stephanie is loving life, finding faith, and greeting the ups and downs with arms open as wide as the sun.

The words, "Between a life threatening skin disorder, being married and being so young, we have our work cut out for us! Join us throughout our journey," are splashed across the top of the page, as Stephanie is happy to spread awareness for her disorder and invites the world to peek in at the day-to-day lives of her and husband, Curt Turner. These days, she has something even more to share. Like the rest of the social-media-savvy ichthyosis community, FIRST learned via Facebook, in the early hours of the morning on May 25th, that this little lucky lady with the never-ending smile became the first known woman affected with Harlequin Ichthyosis to give birth. Their healthy, beautiful boy, William Curtis Drake, weighed 7 pounds, 10 ounces.

FIRST is delighted to extend a warm and heartfelt congratulations to the entire Turner family on the birth of their beautiful son–affectionately called "Little Will" by his joyous new parents. We look forward to following their ever-inspirational journey as they continue their work as advocates for ichthyosis and begin their exciting new lives as parents to Little Will.

All the Best!

Little Will

Save The Date!

Patient Support Forums

August 03, 2013 • New York/North Jersey
August 10, 2013 • Cincinnati, Ohio
September 07, 2013 • San Jose, California
September 21, 2013 • Kansas City, Missouri

18th Biennial National Family Conference

June 20, 21 & 22, 2014
Hilton Indianapolis