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

Ichthyosis Focus

Winter 2017 A Quarterly Journal for Friends of FIRST Volume 36, No. 1

Photo Courtesy of Nashville Convention & Visitors Corporation

Music City... Here We Come!

20th Biennial National Conference

What better backdrop for FIRST's largest "conference of connection" than a town whose soul is synonymous with the word music, like Nashville?

Continued on page 5

Inside This Issue:

Vol. 36 No. 1 Winter 2017

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

Requests to reprint information
contained in the *Ichthyosis Focus*
should be directed to the editor.

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

Chief Executive Officer
Jean Pickford

Editor
Maureen Neville

Medical Editor
Amy Paller, MD

Editorial Assistant
Kimberly Cole

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.

Happy New Year to all our members and friends of FIRST!

Did you know that FIRST is the leading global patient advocacy group for ichthyosis and related skin types in the world? Our membership is reaching 6000 families and $2.1 million has been funded toward research and is still climbing. We are helping more families and connecting more individuals with each other and expert medical care than ever before.

Have you experienced the impact of connecting with another family or individual affected with ichthyosis or a related skin type? If yes, then you will agree that 2017 is poised to be a very powerful year within the FIRST community. All year long, we are celebrating the power of connecting by hosting 12 (yes, twelve) Patient Support Forums from coast to coast! On page 6, you will learn all the details about these very special support meetings, as well as 12 compelling reasons to attend.

Also, before you know it, Ichthyosis Awareness Month (IAM), which is celebrated throughout the month of May, will be at our doorstep. This year the awareness and fundraising efforts are reaching far and wide, as we've been connecting with our international partners to create a universal campaign of awareness, advocacy and collaboration. We invite you to join us by getting involved in this global initiative. We'll be sharing all the ways to participate on our website and social media channels. Stay tuned!

In this quarter's Ichthyosis Focus, you'll learn why sweating can be a complicated issue for those affected with ichthyosis, and you'll meet Dr. Chris Bunick, our 2017 FIRST Research Grant recipient, and learn why his research in x-ray crystallography is so important to our community.

And, with much fanfare and hoopla….we are announcing the location for the greatest way to connect that FIRST has to offer! Our 2018 National Conference - Nashville, here we come (June 29-July 1, 2018)! Here's to connecting with you in 2017 and beyond!

Jean R. Pickford

Jean

Chief Executive Officer

Correspondence Corner

“Families and children can benefit from supportive networks of other families with similar conditions. FIRST is a great example of this.”

J. Teng, MD

“My angel Elliott was born 6 weeks early and diagnosed with ichthyosis at birth. He is now almost 3 months old. My partner has lived by FIRST’s website for the past 12 weeks. Thank you so much for having such wonderful resources.”

A. Britt

“Thank you so much FIRST for your help with all of this stuff! You made me feel like I wasn’t alone and crazy. That means so much to me.”

K. Young

Meet FIRST 2017 Research Grant Recipient

On September 25, the Board of Directors approved funding of Dr. Christopher Bunick’s work in determining the structural mechanisms of ichthyosis-causing mutations by x-ray crystallographic analysis of keratins 1 and 10. The FIRST research grant will provide funding in the amount of $50,000 for a period of one year, which began October 1.

Dr. Christopher Bunick is a unique physician-scientist among dermatologists because of his application of x-ray crystallography to the field of dermatology research, bringing an expansive view to the notion of what can scientifically be achieved to help patients. “X-ray crystallography technology enables determination of a 3-D image of what a protein looks like in living cells, therefore, I am able to see the structures of the proteins that constitute the human skin barrier. This technology is going to greatly improve our understanding of the science behind skin function,” he explained. When asked about his futuristic vision, Bunick stated, “What I imagine is creating a niche of dermatology research, ‘molecular or structural dermatology’ so to speak, and one by one, determine 3-D structures of proteins highly relevant to normal and diseased skin. I am excited to tackle many disorders in the field of dermatology from this structural perspective, and my hope would be translation of this work into new classes of therapeutics to help patients with skin disorders.” Please join the FIRST community in congratulating Dr. Bunick and wishing him all the best as he continues his endeavors into the field of ichthyosis research. Stay tuned for updates on Dr. Bunick’s progress at firstskinfoundation.org.

New Clinical Trial Conducted by Dermatologists at Northwestern University (Chicago) and Mt. Sinai School of Medicine (New York)

If you are 18 years or over with congenital ichthyosis (lamellar, CIE, netherton, or epidermolytic ichthyosis), please consider participating in a new research study focusing on treatment for ichthyosis. There is no cost to participate and enrollees will be compensated for their time.

The new research suggests that skin redness and possibly scaling of ichthyosis may be related to skin immune system imbalances. “We are testing a therapy to correct these imbalances, which we suspect will improve ichthyosis redness and possibly scaling,” says Dr. Amy Paller of Northwestern University. The study will enroll 40 adults (ages 18 and above) with the forms of ichthyosis listed above who have at least moderate redness.

Participants will receive a shot of either secukinumab or placebo for 16 weeks (about once monthly), then all will receive secukinumab for the rest of a year. Bathing and use of moisturizers can be maintained throughout the study. Participants must be able to travel to Chicago or New York approximately monthly, for the duration of the trial (some travel allowance available).

Participants must stop oral retinoids 4 weeks before participating in the study, and topical peeling medications 2 weeks before participating in the study.

Trial is posted at:
https://clinicaltrials.gov/show/NCT03041038

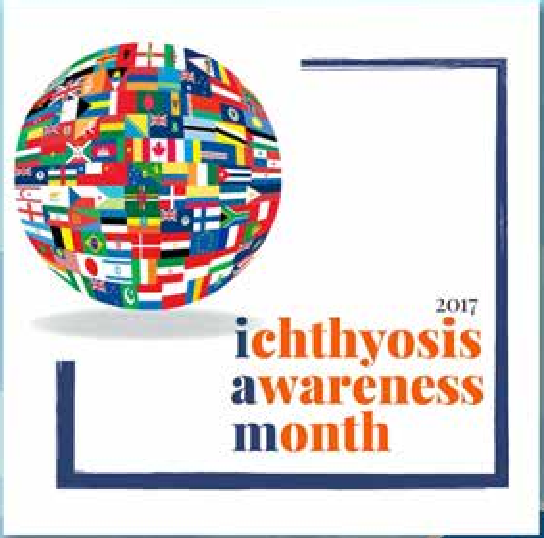
Contact Dr. Erin Ibler and Dr. Amy Paller for information and interest: erin.ibler@northwestern.edu; Phone: 312-695-2203

RARE DISEASE DAY - February 28

Rare Disease Day takes place on the last day of February each year. The main objective of Rare Disease Day is to raise awareness among the general public and decision-makers about rare diseases and their impact on patients' lives. The campaign targets primarily the general public and also seeks to raise awareness among policy makers, public authorities, industry representatives, researchers, health professionals and others who have a genuine interest in rare diseases. Many members of the community get involved in advocacy efforts at the state or local levels on Rare Disease Day and throughout the year. Many share information about their rare condition on social media, as a quick, easy and far-reaching way to get the word out. For more information go to **www.firstskinfoundation.org/rarediseaseday.**

IAM International - MAY 2017

Be a part of the worldwide effort.



May is Ichthyosis Awareness Month (IAM). During May, we encourage families and friends around the world to join together, use their voices, and raise awareness about the hopes and challenges of the ichthyosis community. Simply handing out educational information, or taking the time to talk to neighbors, friends, co-workers and local communities, is one of the best ways to show you care about those affected with ichthyosis.

Are you on Facebook? Twitter? Instagram? IAM is going global!

Social media is the quickest and easiest way to be a part of the worldwide IAM effort. Just post a photo of you, your friend, relative or child with ichthyosis, or a related skin type using **#IchthyosisAwareness**.

Be sure to include an IAM message, and a symbol of your home town, state or country! Let the world know that you support someone with ichthyosis, or a related skin type.

Music City... Here We Come!

20th Biennial National Conference 2018

Nashville, TN, June 29 - July 1, Nashville Airport Marriott

Music is a common thread connecting the hearts and minds of people throughout the world. And what better backdrop for FIRST's largest gathering of individuals and families connecting in one place, than a town whose soul is synonymous with the word music like Nashville? Not only will you meet lots of new friends and families affected with the very same rare skin condition as you, but you'll also have the opportunity to meet one-on-one with the world's most renowned medical and research experts in the field of ichthyosis — all while enjoying the zest and culture of America's beloved Music City. Plus, Nashville has a lot more going on lately than guitar riffs and late-night crooning. There is something for the whole family! The city continues to rise as a foodie destination and the new Riverfront Park completed last summer holds 10 acres of parklands, an amphitheater, gardens and the ultimate downtime essential — free WiFi.

It's not too early to start planning and saving for your trip to FIRST's 20th Biennial National Conference in June 2018...

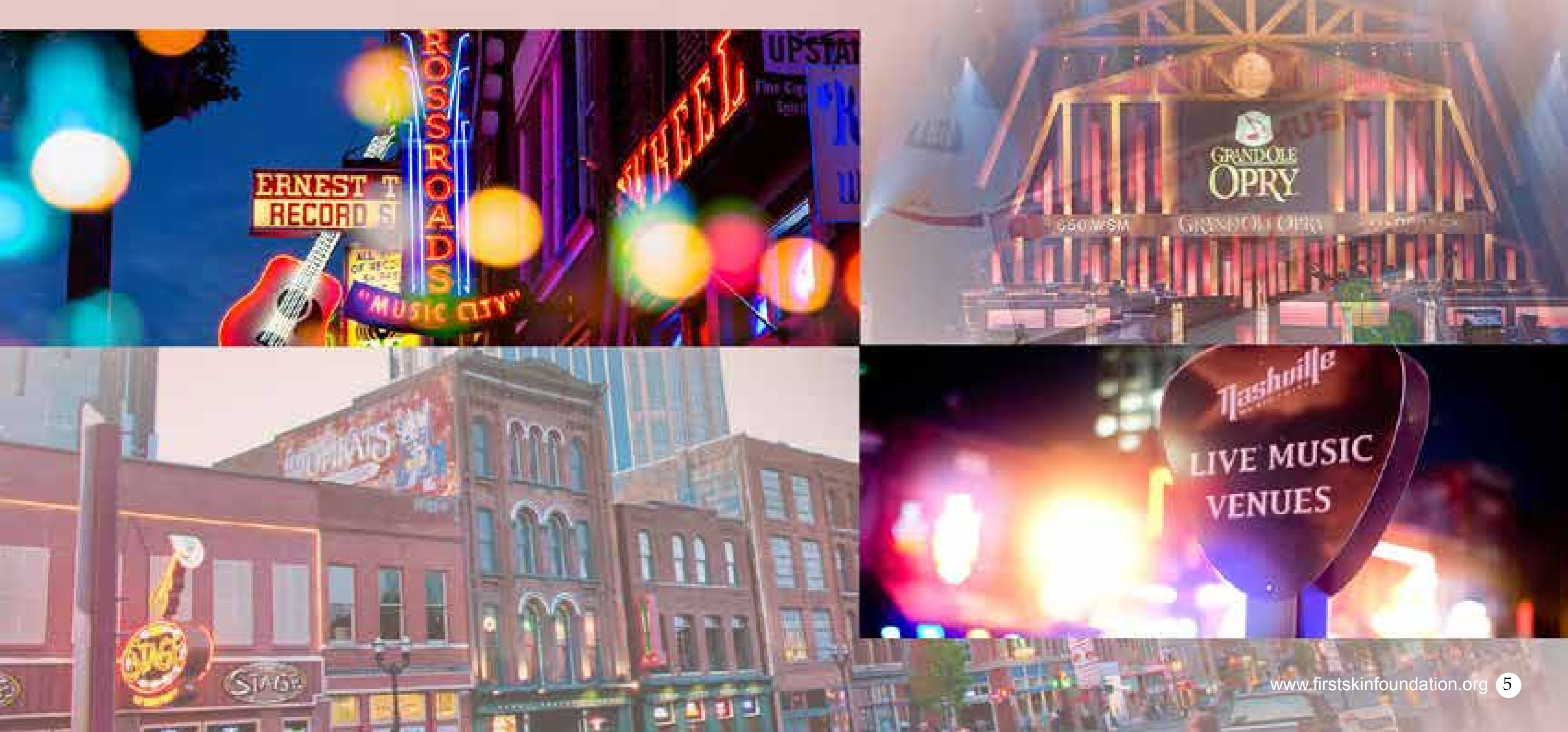
Here are a few ideas from FIRST member Mark Klafter, to ensure you and your family can enjoy this unique, inspiring and educational experience that only FIRST can provide.

- Start a change jar for all your daily spare change. You'd be surprised how quickly this adds up!
- Find one thing you spend money on regularly that you can either cut back on or cut out altogether. Skip the grande mocha latte and brown bag lunch it for a while…an extra $5 a day means $150 extra in a month. Yup!
- Walk instead of driving if you can. Figure out how much you've saved in gas money and put it right in the jar!
- Raise the money through a small fundraising activity. Get the whole family involved. Perhaps you or the kids can walk the neighbor's dogs, babysit, hold a bake sale or have a neighborhood car wash.

There is an endless number of ways you can do it. And you'd be amazed how people are willing to help when they know what you're trying to accomplish.

More details regarding the conference will be available on the FIRST website in upcoming weeks (firstskinfoundation.org). Please feel free to contact Senior Director, Programs & Research, Moureen Wenik with any questions at mwenik@firstskinfoundation.org.

Here's to seeing you in Nashville in 2018!



13 Ways to Connect

FIRST has numerous ways to connect with other families, individuals, and ichthyosis experts. Our national conferences, patient support forums, FIRST to Know calls, and Facebook Groups are some great ways to find tips, ask questions, make friends and meet medical experts that are truly "in the know" about your condition. Connecting with others can make all the difference in coping and living with your condition. And this year, there are more opportunities than ever before. In fact, there are 13 Patient Support Forums all over the country!

13 Benefits of Attending a PSF...

1. Make new friends with members from your region
2. Connect one-on-one with ichthyosis medical experts
3. Learn how to find out about the genetics of your condition
4. Receive emotional support from individuals and families who truly understand
5. Come to a warm, safe place to share your story
6. Meet FIRST staff and learn more about FIRST, the only patient advocacy organization in the United States
7. Learn skincare tips from others
8. Hear stories about coping and living with ichthyosis, for all stages of life
9. Be inspired by others and leave feeling empowered
10. Sample skincare products
11. Join in the medical discussion
12. An evening in the city!
13. Have fun!

Patient Support Forums for 2017:

- **March 25** - Raleigh, North Carolina
- **April 1** - San Antonio, Texas
- **May 6** - Philadelphia, Pennsylvania
- **May 20** - Omaha, Nebraska
- **June 10** - Portland, Oregon
- **July 15** - Milwaukee, Wisconsin
- **July 22** - Billings, Montana
- **August 19** - Boston Massachusetts
- **September 16** - Denver, Colorado
- **September 23** - Las Vegas, Nevada
- **TBD** - Jackson, Mississippi
- **TBD** - Louisville, Kentucky
- **TBD** - Canada

A registration form is now available online at firstskinfoundation.org

Call In and CONNECT

Sharing tips, stories and challenges with others, makes living with ichthyosis just a little bit easier. The ***FIRST to Know*** calls are a great way to "meet" other families and individuals.

Each call is centered on a topic or subject and lasts for one hour. You can decide which call is interesting to you and phone in. You can be an active participant, or call in and listen to what others are saying. All calls are held at 8:00 PM Eastern Time. Please remember to register for the call at firstskinfoundation.org/first-to-know-conference-calls. Do you have an idea for a ***FIRST to Know*** topic? Contact Moureen Wenik, mwenik@firstskinfoundation.org.

2017 FIRST to Know Call Schedule

March 19 - ARCI-Lamellar: Sharing Information
April 23 - Netherton Syndrome: Sharing Information
May 21 - Palmoplantar Keratodermas (PPK): Sharing Information
June 25 - Erythrokeratoderma Variablis (EKV): Sharing Information
July 16 - X-Linked Ichthyosis: Sharing Information
August 27 - Ichthyosis Vulgaris: Sharing Information
September 24 - National Conference: What Are the Benefits of Attending?

Sweating in Ichthyosis Skin

Sweating is important for human thermoregulation.

Sweat glands are distributed broadly in human skin. Sweating is critical for regulating our body temperature through heat dissipation when water in the sweat evaporates from the skin surface. Abnormal sweating, termed hyperhidrosis (too much) and hypohidrosis (too little), cause various health issues ranging from social embarrassment to discomfort, and overheating to heat stroke (1-3). People who are born with a genetic disorder called Hypohydrotic Ectodermal Dysplasia (HED), for example, are born with no or underdeveloped sweat glands, and suffer from severe difficulty in regulating their temperature. Because of their inability to sweat and to dissipate body heat efficiently, these patients need to stay in a well air-conditioned environment or to constantly spray water mist onto their skin surface to cool down. It is very dangerous and potentially life-threatening to perform outdoor activities under heat exposure. In addition to secreting sweat, sweat glands also produce various proteins that can fight infectious organisms (antimicrobial peptides) (4), as well as substances that are important to maintain the acid-base balance (pH) of the skin within a normal range. As such, insufficient sweating is also a cause of dryness and propensity to infections in the skin.

Sweat glands are made from the outer layer of the skin (epidermis). Early in development (in the embryo), the sweat gland starts to bud from the epidermis and continues to grow into the deeper part of the skin (called the dermis) (5, 6). As the gland matures, its bottom part forms a tightly coiled tube-like structure (called a duct) that is responsible for sweat production and secretion (Fig. 1, left). The sweat will be squeezed out, first through a straight region of the duct in the dermis, then in a spiral-shaped region of the duct in the epidermis (termed the acrosyringium), eventually reaching the opening in the skin surface.

Ichthyosis patients cannot sweat efficiently.

Many patients with ichthyosis also suffer from a similar inability to efficiently regulate their body temperature. As opposed to the situation in HED, it is unlikely that the reason in ichthyosis is because sweat glands don't form normally – although certainly there has been little study to explore the reason for the decreased ability to sweat. New research could help us to understand, at least in part, why ichthyosis is associated with decreased sweating.

Sweat cannot reach to the surface in wounded skin.

In wounded skin, the acrosyringium (that region of the duct in the epidermis) is damaged. As a consequence, sweat cannot reach the skin surface (Fig. 1, Right) and the benefits of sweating, heat dissipation and moisturizing of the skin, are temporarily lost until the ducts are repaired. Research using animal model and human skin (7, 8) discovered that cells in the deeper part of the sweat duct can be activated to migrate into the epidermis to repair the acrosyringium region in response to epidermal injury. Ichthyosis can be thought of as "wounded skin" that is under stress – and it is possible that there is an abnormality in this repair mechanism. Recent work by Perknimitr et al. (9) demonstrated sweat duct obstruction in mouse model with decreased levels of filaggrin, a protein that is important for the skin barrier and that is reduced in people with the most common form of ichthyosis, ichthyosis vulgaris (IV) (10). It is possible, then, that this sweat gland obstruction is responsible for the heat intolerance that several individuals with more severe ichthyosis vulgaris have reported to FIRST Filaggrin levels are not reduced in other forms of ichthyosis (11), and in fact is often increased; however, all forms of ichthyosis share the issue of barrier abnormalities and associated thickening of skin, and a similar mechanism may be occurring. Further investigations using skin samples from ichthyosis will help us to understand if sweat glands are developed normally and the mechanism for inefficient sweating, including the possibility of these abnormal repair mechanisms. Better understanding of these issues related to sweating could improve the quality of life of our patients.

Catherine Lu, PhD Postdoctoral Fellow, Laboratory of Mammalian Cell Biology and Development Rockefeller University, New York Dec. 2016

Reference

1. K. Sato, W. H. Kang, K. Saga, K. T. Sato, Biology of sweat glands and their disorders. I. Normal sweat gland function. J. Am. Acad. Dermatol. 20, 537–563 (1989).

2. W. P. Cheshire, R. Freeman, Disorders of sweating. Semin Neurol. 23, 399–406 (2003).

3. C. Lu, E. Fuchs, Sweat gland progenitors in development, homeostasis, and wound repair. Cold Spring Harb Perspect Med. 4 (2014), doi:10.1101/cshperspect.a015222.

4. B. Schittek et al., Dermcidin: a novel human antibiotic peptide secreted by sweat glands. Nat. Immunol. 2, 1133–1137 (2001).

5. C.-Y. Cui, D. Schlessinger, Eccrine sweaat gland development and sweat secretion. Exp. Dermatol. 24, 644–650 (2015).

6. C. P. Lu, L. Polak, B. E. Keyes, E. Fuchs, Spatiotemporal antagonism in mesenchymal-epithelial signaling in sweat versus hair fate decision. Science. 354 (2016), doi:10.1126/science.aah6102.

7. C. P. Lu et al., Identification of stem cell populations in sweat glands and ducts reveals roles in homeostasis and wound repair. Cell. 150, 136–150 (2012).

8. L. Rittié, D. L. Sachs, J. S. Orringer, J. J. Voorhees, G. J. Fisher, Eccrine sweat glands are major contributors to reepithelialization of human wounds. Am. J. Pathol. 182, 163–171 (2013).

9. **P. Rerknimitr et al., Decreased filaggrin level may lead to sweat duct obstruction in** filaggrin mutant mice. J. Invest. Dermatol. 137, 248–251 (2017).

10. F. J. D. Smith et al., Loss-of-function mutations in the gene encoding filaggrin cause ichthyosis vulgaris. Nat. Genet. 38, 337–342 (2006).

11. A. Paller et al. An IL-17-dominant immune profile is shared across the major orphan forms of ichthyosis. J Allergy Clin Immunol. 139, 152-165 (2017).

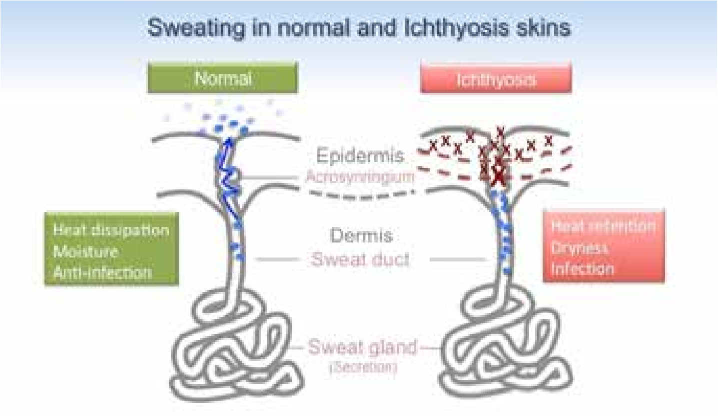


Figure 1. Sweating in normal and ichthyosis skins.
Blue dots indicate the sweat droplets, secreted to skin surface through the acrosyringium regions in normal skin, but retained in the tissue in wounded skin due to obstruction (red cross) in the epidermis.

Physician Scientists & Patient Advocates Collaborate at PeDRA Annual Conference

A research network can be a powerful force in changing care for pediatric skin disorders, including ichthyosis. FIRST's relationship with PeDRA (Pediatric Dermatology Research Alliance), demonstrates how valuable it is to be involved in this network. Senior Director, Programs & Research, Moureen Wenik attended PeDRA's Annual Conference, where the interest in disorders of keratinization has a large working group of extremely dedicated and talented clinical researchers, basic scientists, and experts in research tools. FIRST was represented on a panel, along with Dr. Keith Choate, to discuss the research currently conducted at Yale University including the Gene Discovery Project led by Dr. Choate's lab, and the Ichthyosis Newborn Study conducted by Dr. Britt Craiglow. FIRST also participated in a poster session, which was a unique opportunity to highlight the important work FIRST does, and the research needs for our members. There was so much energy and enthusiasm at this meeting, as well as excitement for future research that is emerging from the PeDRA network.

Diya & Aliya's Friends (DAF) Fund

Thanks to the generosity of the Shahnaz Kraybill family (Aliya, affected with ichthyosis, her sister, Diya, and their parents Durreen and Robert), and their family and friends, FIRST has established the Diya & Aliya's Friends (DAF) Fund to help alleviate some of the financial burden that may be facing families with affected children. This special fund is available for children around the world affected with ichthyosis and is intended to provide financial assistance to purchase lotions, medicine, and treatment necessities.

To apply for an award, please complete the DAF application form. All forms must be completed in English and submitted to the FIRST office. Individual awards will not exceed $250.00. Applications will be awarded two times per year; all applications must be received by April 30 or October 31 each year to be considered for each cycle. For non-US residents, the grant award will be sent via Western Union; it is the applicant's responsibility to locate the nearest Western Union office to receive the award. Applicants will be eligible to receive one award every two years.

This fund is available to affected children from around the world. Two-thirds of the fund disbursements have been designated for non-US families; the other one-third is designated, but not limited, to US families. You will be contacted by the FIRST office if you have been awarded funding.

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

Foundation for Ichthyosis & Related Skin Types, Inc. ® (FIRST)
Diya & Aliya's Friends (DAF) Fund
2616 N. Broad Street, Colmar, PA 18915 USA

The deadline to apply for funds is April 30, 2017

Donations to Diya & Aliya's Friends (DAF) Fund are always appreciated and enable FIRST to make this fund available to more deserving families. Donations can be made at www.firstskinfoundation.org.

An Unsung Hero for FIRST

Meet FIRST's legal counsel and longtime member, Dan Siegel

FIRST is only as strong as it is because of the passion and commitment of our volunteers, members and community. Today we'd like to introduce you to what FIRST considers a volunteer "behind the scenes," who has been quietly and loyally supporting the mission of FIRST for nearly 17 years. We are delighted and privileged to introduce you to FIRST's legal counsel, Daniel J. Siegel, Esq.

CEO Jean Pickford, who has worked alongside Dan for her entire career with FIRST, says, "Dan was one of the first members who I personally met shortly after I was hired. He lived local to the national office, and was someone I could really count on in the early days of my employment at FIRST. I can remember meeting his wife and two sons, who are affected with X-linked ichthyosis, when they were just little boys. Fast forward 17 years, and FIRST is still so lucky to have Dan on our team. His boys have grown into amazing young men, not to be hindered by their skin. Dan's commitment to FIRST remains unwavering. He's one of those quiet but mighty volunteers who mean so much to FIRST!"

We are delighted to share with you a recent interview with Dan, who says he has always been impressed by FIRST's "member-first" perspective.

How did you initially find out about FIRST?

I saw an ad for FIRST in Dorland's, which was then a huge print publication that listed all of the doctors and hospitals in the Philadelphia area. Because I had only recently learned that my sons had X-linked ichthyosis, I contacted the organization, asked for information and joined.

What inspired you to get more involved?

Actually, it was FIRST that asked me. I had been running for a local elected position (Township Commissioner) and lost the election. Shortly after the election, I received a call from another local member who served on FIRST's board asking if I wanted to volunteer to serve as counsel for the organization. She also said that she waited until after the election to ask me, pending the outcome. Though I did lose that election, I ran again and was elected Commissioner in 2008. I'm currently running for my third full term this year.

To what capacity have you volunteered for the organization? How long have you been involved with FIRST?

I have served on the Board of Directors and provided consulting on technology matters, as well as serving as general counsel for all 20 years of my involvement. During that time, I have provided completely free legal services to the organization, ranging from analyzing contracts, to providing legal advice on a wide range of issues. My wife and I also encourage friends to donate to FIRST, and we regularly donate to honor someone who has recently passed or to honor someone for a life event.

What changes have you witnessed since you first connected with the organization?

The growth of the organization has been dramatic. When I first became involved, FIRST had a tiny office in a small building in Havertown (where I live), with a pizza parlor below. Although the pizza parlor is still in business, FIRST moved a bit farther north, and now has a much larger staff and budget, and has broadened its ability to serve members and affected persons significantly. In addition, the staff has grown, allowing Jean Pickford to focus her efforts on the broader issues and allow others to focus on members, research grants and fundraising.

I would be remiss if I did not state that the organization, and its Board in particular, is remarkably collegial and has never devolved into one where personal agendas compete with governance. Second, Jean Pickford has been a steady leader who handles whatever issues she faces with professionalism and cordiality. Finding both of these in one organization is very rare, and that comes from a person who has served on or advised boards and nonprofits for more than 30 years.

What are your hopes for the future of FIRST? Ichthyosis?

That the organization continues to grow and, consequently, I also hope that the organization retains its member-first perspective. As for ichthyosis, I hope that FIRST can work with researchers to cure the condition or alleviate the discomfort (physical and emotional) suffered by those with the condition.

It Takes Only One Day to Make a Difference

by Maddy Bergman

Want a way to get you through the winter blues? Thinking of something fun to do for a day with your family? Host a fundraiser for FIRST! Here's our guide on how to hold a rewarding and enjoyable event for FIRST in only one day.

It only takes one day to make a difference to the ichthyosis community. That's 24 hours to help connect families of this rare skin condition; 1,440 minutes to raise awareness of ichthyosis and the FIRST community.

Every time I work with a FIRST member to plan a grassroots fundraising event, I am in awe of their creativity and passion. Each event we plan is more ingenious than the last! From ***FIRST Night Out*** events with a local sports teams to bake sales, car shows, or pumpkin painting, I love to see how our grassroots organizers plan an event that also speaks to their own personal passions and interests.

Tracie Pretak, organizer of the Release the Butterfly tour, said it best. "I love to sing, Bailey loves to dance and we both love FIRST. That's why developing a musical tour as our fundraiser works for us. If I was to plan a golf outing, I would not be as passionate. Golf is not my forte," she said.

Regardless of your interest, you and I can work to come up with a way to raise funds. The most important thing you need to have is the desire to help FIRST and the ichthyosis community. So, what can you do in 86,400 seconds? Contact me, Madeline Bergman at mbergman@firstskinfoundation.org today and we can start planning your next fundraiser for FIRST.

4 WAYS TO GET STARTED

1. Contact FIRST

Madeline, FIRST's Director of Development, is here to help you organize your fundraiser each step of the way - from brainstorming what kind of event to hold, to helping promote your event in the community.

2. Do something that interests you

Do you love bingo? Do you love to bake? Perhaps a rousing game of kickball is more your speed! When thinking about what type of event you should organize, always pick something that interests you, your family and friends. This adds even more fun to the event planning process.

3. Get the word out

Invite your friends, family, school or church community. A grassroots fundraiser is the perfect way to reconnect with old friends and make new ones. FIRST will help promote your event to local FIRST members in your hometown too!

4. Party on FIRST members!

It's party time, excellent! Everyone loves a party. Not only will you have a fun and rewarding day with your family and friends, but you will also be spreading awareness about ichthyosis in your hometown. FIRST will be happy to send you awareness materials and FIRST giveaways to share with your guests.

Difference Made

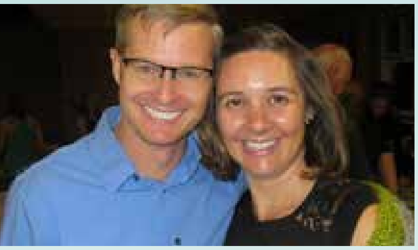
The fun of the event may be over, but the impact your fundraiser has on the FIRST community is just beginning. All fundraisers, regardless of the amount raised, will help FIRST enhance its educational resources, connect more families affected by this rare skin condition, and make further advancements in research.

CONGRATULATIONS TO FIRST CHAMPIONS, KYLE AND KELLY ROBINSON!

On October 22, over 100 people gathered at the Providence Picture Frame and Dryden Galleries in Providence, Rhode Island, to celebrate with Kelly and Kyle Robinson as they were named FIRST Champions. The afternoon featured a delicious wine tasting, fantastic raffle items and food. Adding to the celebrations, the Release the Butterfly Tour performed for the guests, including the Pretak family, Steinitz family, Abby Evans, Mikela Murphy and the Hamill family.

Kelly and Kyle were celebrated for their support of FIRST and the ichthyosis community. They contacted FIRST in 2012 after their daughter, Sienna, was born with epidermolytic ichthyosis (EI). Both Kelly and Kyle have organized multiple fundraisers, including an annual Jeans Day for Rare Genes and a FIRST Night Out with the Boston Red Sox. The Robinsons have attended three FIRST National Conferences. It was great to see Sienna and her brother, Teagan, this summer in San Diego.

The night raised over $4,800 for FIRST programs and services. We extend a heartfelt thank you to the Robinson family and their kind and generous community.



2nd Annual FIRST Night on the Dance Floor with the Schell Family!

Over 80 people danced the night away in Fort Collins, Colorado, at the 2nd Annual FIRST on the Dance Floor event, hosted by Allan and Michelle Schell. The night featured some good tunes, merriment and a silent auction. Thanks to Allan and Michelle's hard work in soliciting auction items, the event raised $1,500 over their previous year. FIRST is so thankful to the Schells for their fundraising efforts and continued support of FIRST.

It is quite a list of grassroots organizers when you put them all together! Thank you to the following individuals and families for hosting a fundraiser for FIRST this fall. You are all superstars! Hopefully your dedication is inspiring others to join with you to raise awareness and funds for FIRST.

Event	Location	Amount Raised	Organizer
Jeans Friday	Florham Park, NJ	$265	Savoy and Jolie Cina
Dig Blue for Liam Volleyball Tournament	Vestal, NY	$721.77	Vestal Varsity Club
Release the Butterfly Tour	Lancaster, PA	$2,668.75	Stern Family
LuLaRoe Fundraiser	Online	$290.43	Erin Edwards
Pumpkin Paint & Bake Sale	Flandreau, SD	$1,225	Iott Family
Dance Palooza at MUW	Columbus, MS	$300	Kelly Benford
FIRST on the Dance Floor	Fort Collins, CO	$5,029.77	Allan & Michelle Schell

WHEN YOU GIVE TO FIRST…You Give a Hug

Members and their community gave BIG on Giving Tuesday

Giving Tuesday is a global day of giving celebrated on the Tuesday following Thanksgiving (in the United States) and the widely recognized shopping events, Black Friday and Cyber Monday. Giving Tuesday kicks off the charitable season, when many focus on their holiday and end-of-year giving. This year, a record number of individuals reached out and gave our members a big, warm, caring hug with their kind and generous donations, totaling $6,034.80! Each dollar reassures those affected that someone really cares about their issues, needs, and lives. Thank you with a great BIG HUG from FIRST!

FIRST ADVOCATES

21st Century Cures Act Signed Into Law

On December 13, former President Obama signed into law the 21st Century Cures Act, a game-changing bill for medical innovation. This legislation has the potential to help nearly every American family, including the 1 in 10 Americans with rare diseases who desperately need treatments and cures. The bill includes many provisions that will improve the discovery, development, and delivery of therapies for rare disease patients, including:

- Extension of the Rare Pediatric Disease Priority Review Voucher Program, which incentivizes the development of new therapies to help the more than 15 million children with rare diseases;
- Streamlining of U.S. Food and Drug Administration (FDA) review of genetically targeted and protein variant therapies for rare diseases;
- Creation of funds in the amount of $4.8 billion over 10 years for the National Institutes of Health (NIH) to fund the Precision Medicine Initiative, BRAIN Initiative, and the Cancer Moonshot;
- Expansion of the Patient-Focused Drug Development Initiative and requirements for the FDA to report on how patient experience data was used in regulatory review.

Member Highlight

Meet member Bradley Auw

Sometimes a little encouragement, along with a foolproof strategy to avoid the heat, is all you need to follow your heart. That was the case with Bradley Auw, whose mother Amy submitted this story to show other families that ichthyosis and athletics can actually be a winning combination. Just watch the temperature, prepare for warmer weather, and have an alternate plan for days when it is simply too hot.

Bradley Auw is 13 years old. He was diagnosed with Congenital Ichthyosiform Erythroderma (ARCI-CIE type) by Dr. Amy Paller. We wanted to write and share with FIRST and your readers and members that, while it was a warm fall here in Chicagoland, Bradley, an 8th grader, participated in his 3rd year of cross country at his middle school.

As the story goes, Bradley has always really enjoyed sports. Unfortunately, he has been relatively restricted from participating in many physical activities, especially in warm weather. But he decided to give middle school cross country and track a try, after watching his older brother, Stephen, who is not affected, participate in the sport. We figured that the distances were short enough (1 and 2 miles) and that Bradley could handle the physical activity.

There have been times during the seasons, where the outdoor heat did prevent him from participating with his team. On the really hot days, Bradley would practice at home on the treadmill. He did have to skip a meet this season because it was just too hot. There were other days at practice, where he would have to take breaks in the shade. Sometimes, he would even take his shoes and socks off to help cool down. Most days he would bring a jug of water to dump over his head to avoid overheating. For many of the meets, Bradley would put his entire head under a running sink, just before race time, to help stay cool.

In Bradley's own words, "My first meet this season was very tough to compete in because of the heat and humidity. I had a bad run and felt pretty defeated. I honestly didn't know if I wanted to finish the season, but on the car ride home my Dad reminded me that the weather would eventually cool down, and my running times would improve."

As promised, the weather did cool down and Bradley ended up being a major contributor to his team. Bradley's team earned second place at sectionals, which qualified them for the state meet. At state, Bradley ran as 4th man for his team. He shaved 15 seconds off his previous best time and finished with a solid 12:24 two mile run. - Amy Auw

So the good news is that sports are definitely possible, but not always easy. In summary, here is a handy list of five tips to remember for "athletes with ichthyosis."

1. Be aware of shady spots around the playing field when a break is needed.

2. Bring plenty of water.

3.Taking off shoes and socks can help to quickly cool down.

4.Sometimes you may have to sit out a game or meet. No big deal!

5.If outdoors is just too hot for training, train indoors on a treadmill or weight set.

FIRST highly recommends that you consult with your or your child's physician prior to beginning any athletic routine.

AAD Camp Discovery Dates Announced

The American Academy of Dermatology's (AAD) Camp Discovery program is for children ages 8-16 who have a chronic skin disease.

Under the expert care of dermatologists and nurses, Camp Discovery gives campers the opportunity to spend a week with other young people with skin conditions having fun and participating in activities such as swimming, horseback riding, arts and crafts and many more. There is no fee to attend; all costs, including transportation, are provided by the AAD through generous donations from its members, outside organizations and individuals. All campers must be initially referred by their dermatologist.

This year the Academy is proud to offer five camping sessions:

June 18-23: Crosslake, Minnesota (ages 8 – 13)

July 2-7: Crosslake, Minnesota (ages 14 – 16)

August 6-11: Burton, Texas (ages 8 – 16)

August 12-18: Millville, Pennsylvania (ages 8-16)

August 13-19: Andover, Connecticut (ages 8 – 16)

For more information about attending, volunteering or donating, please visit campdiscovery.org or contact Janine Mueller at jmueller@aad.org.

Heading out for a Winter Pedicure?

Check out these tips from FIRST Member Denise Eiser...

Denise Eiser, Member of FIRST Board of Directors

1. Work on your feet ahead of time so they don't have much skin to remove. For me, it usually takes an hour with a foot file. Then you can tell them not to worry about it (the pedicurist I had yesterday went way overboard and I should have told her to stop. She couldn't see when it was time to stop because our skin acts differently.)

2. If the pedicurist uses a foot file, only let them do it when your feet are DRY. If they use one after soaking, it can easily take off too much.

3. The most important thing to remember is to bring your own foot cream. The lotions they use will never keep my feet adequately moisturized.

4. If you have a spot where you think a crack might develop, as I usually know where/when one is about to form, put a bandaid on it so they don't even go there.

Enjoy! Denise

Have You Heard? It's a new GENEration.

Join the new GENEration of ichthyosis research

Have you received a genetic diagnosis?

Now is the time to get diagnosed and join FIRST in playing an important role in ichthyosis research. Since 2016, FIRST has been co-sponsoring a transformational scientific endeavor with one of the world's leading institutions in ichthyosis research, Yale University. We encourage you to participate. The National Registry for Ichthyosis & Related Disorders, or just "The Registry," is available to investigators worldwide, and is the largest resource of ichthyosis patient information - critical data that holds the key to better, more targeted treatments, and eventual cures. The Registry, co-sponsored by FIRST is the next evolution of ichthyosis research. There is no cost for participating in The Registry. If you do not already have a laboratory diagnosis, Dr. Choate's lab will perform the DNA testing under his research funding at no cost to you/your insurance. And there is no need to travel to Yale University. We will work with you/your physician to obtain all relevant medical records and consents via email or fax. Any costs of shipping will be covered by us. To participate, please email maryann.ackerman@yale.edu or call the Yale lab at 203.785.5364. A member of the team will promptly respond.

Join The Registry and the new GENEration, and play your part in unlocking the future of ichthyosis research.

Research Study for Congenital Ichthyosis

If you or a loved one has congenital ichthyosis, consider participating in a research study.

We are conducting a research study of an investigational topical ointment for ichthyosis (X-Linked or Lamellar). Study participants must be 12 years of age and up, and in otherwise good health to participate. Qualified participants will receive compensation for time and travel.

Neal Bhatia, MD
Therapeutics Clinical Research
9025 Balboa Ave., Ste. 105, San Diego CA 92123
Contact: Christina West, Recruitment Supervisor
cwest@therapeuticsresearch.com
(619) 512-3376

John Browning, MD
Texas Dermatology and Laser Specialists
3320 Oakwell Ct., San Antonio, TX 78218
Contact: Carla Ilie, Clinical Research Coordinator
carla@texasdls.com
(210) 660-2949

Amy Paller, MD
Ann & Robert H. Lurie Children's Hospital
225 E. Chicago, Chicago, IL 60611
Contact: Thy Huynh, MD. Subinvestigator
(312) 227-6485

www.ichthyosistrial.com

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

UFIRST
Scholars
Education Scholarship Program

UFIRST Scholars Program Accepting Applications Beginning February 1st - March 31st

The UFIRST Scholars program is an opportunity for affected students to advance their post-secondary education in partnership with FIRST. The 2017 UFIRST Scholars applications will be accepted beginning February 1. Deadline to apply is March 31. Please go to firstskinfoundation.org/ufirst-scholars-program for more information.

20th Biennial National Conference 2018

Nashville, TN, June 29 - July 1
Nashville Airport Marriott