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

Ichthyosis Focus

Fall 2014 A Quarterly Journal for Friends of FIRST Volume 33, No. 3

Doctors Connect Online to Diagnose and Advise

Discover how FIRST is Reaching New Heights in Telemedicine.

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Dear Members and Friends of FIRST,

Jean R. Pickford
Executive Director

Although each issue of the Ichthyosis Focus has its own special message, I am particularly excited to be sharing this fall 2014 issue, as its message is one of connecting far beyond what we ever thought possible. Not only will you be learning about the origin behind our newly revised Tele-Ichthyosis Program, but you will hear from one of the leading dermatologists in the field of telemedicine and share in his vision of global medical care for all those affected with ichthyosis and related skin types. You will also be inspired by the latest updates from the FIRST-funded research team, learn how to best prepare to meet with an ichthyosis medical specialist, and get a unique inside look at all the amazing grassroots events hosted by our wonderful members. We invite you to join us in paying tribute to our UFIRST Scholar recipients and two very special Testimonial Dinner honorees. Last but not least, we have continued to grow our relationship with Positive Exposure and bring you all of the exciting opportunities that Rick and this team have in store in the upcoming months.

I hope you are as inspired and excited by our news, as I am to be sharing it.

Happy Fall!

Jean

Jean R. Pickford

Correspondence Corner

Hi FIRST Team!

This is overdue, but I wanted to send a quick note to say congratulations and thank you for an outstanding conference! Our family had an amazing time and I heard nothing but raves from the others I talked to.

Your long hours, hard work and dedication to FIRST made it all possible. I hope you were able to enjoy the conference yourselves and take pride in making it a fabulous experience for members.

I am so happy to work with you all to achieve FIRST's mission, and I especially enjoyed the chance to finally meet and speak face-to-face with you!

Take care and keep up the great work!

Many thanks,

Kimberly Cole
Wauwatosa, Wisconsin

Dear FIRST,

This is my beautiful granddaughter, Kate. She is doing great in her skin and was recently awarded most popular in school! We are so proud of her.

Sincerely,

Proud Grandfather, Joseph Hugenberg
McQueeney, Texas

Dear Friends at FIRST:

I just finished reading your summer 2014 (Vol. 33, No. 2) edition of Ichthyosis Focus. Great publication; very informative. I regret having missed the recent conference.

I wish to express my gratitude to FIRST for making me aware of Beiersdorf's Aquaphor Program. I started using Eucerin "Professional Repair" lotion earlier this year, and in all my years (I'm 81), it's the first (pardon the pun) lotion that's been effective, and, best of all, it's on the Aquaphor Program. For me, it works – no more flaking, much improved appearance, and a happier wife (no more complaints about flakes on the floor or my "picking"). MANY THANKS, FIRST!!!!!

Again, thanks for being there, my friends at FIRST. The ichthyosis community truly needs someone to be their advocate and to help find ways to rid us of this terrible generic problem. May your research program have outstanding results in the very near future.

Most sincerely,

Ronald M. Landau
Huntington Beach, California

Editor's Note: Mr. Landau is affected with ichthyosis vulgaris

This information is provided as a service to patients and parents of patients who have ichthyosis. It is not intended to supplement appropriate medical care, but instead to complement that care with guidance in practical issues facing patients and parents. Neither FIRST, its Board of Directors, Medical & Scientific Advisory Board, Board of Medical Editors, nor Foundation staff and officials endorse any treatments or products reported here. All issues pertaining to thecare of patients with ichthyosis should be discussed with a dermatologist experienced in the treatment of their skin disorder.

Dear FIRST:

What a lovely surprise it was to receive my membership kit. Words seem inadequate to express just how much joy it brought me to receive the package containing important information leaflets, books, DVD and bookmark regarding ichthyosis. God has been so good to us, it came like a 'manna' to us when we were in the desert...being in a place so devoid of everything that could make things a wee easier for our baby Kenile.

Thanks a whole lot again to the FIRST team for your effort. You have brought so much sunshine to our little world. We feel so much at ease and at peace now, knowing that we are NOT alone in this. God bless.

With sincere love & gratitude,

Kenile Khing parents
Dimapur, Nagaland, India

Grassroots Fundraising *Hits It Out of the Ballpark!*

New York Mets

It was an amazing turnout (not to mention a beautiful spring evening), at CitiField in New York on April 25th, for FIRST Night Out at the Mets! A big shout out to the Benedetto family, whose son Marc is affected with ARCI-lamellar type ichthyosis and the Saccente family whose daughter, Kylie Leigh, is also affected with ichthyosis, for rallying such an enthusiastic group of supporters! Also, our sincere gratitude to the Mets organization for working with us step-by-step 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 the Mets, played throughout the entire arena during the game. FIRST was also mentioned as a sponsor of the game in the post game box score on Sports New York (SNY). Everyone enjoyed a co-sponsored Mets/FIRST hat that was given to all those who bought FIRST Night tickets. A Mets win was enjoyed by all and FIRST donations for the evening totaled $2,550. Thanks to everyone for making difference!

Pittsburgh Pirates

FIRST's Day Out at the Pittsburgh Pirates on May 4th was a smashing success! We'd like to extend a heap of gratitude to the Kocher family, whose son Calvin is affected with ichthyosis, for their great efforts in pulling this event together. In addition to the Kocher family, three other affected individuals and their families joined in the festivities. The Horr family, whose son Laban is affected with ARCI-lamellar type ichthyosis, FIRST board member Tracie Pretak and her daughter Bailey, who is also affected with ARCI-lamellar type, and the Allenbaugh family, whose 9-year-old daughter Emma is affected with Sjögren-Larsson syndrome (SLS) and was meeting people with "skin like hers" for the first time, all joined together to create a great day of awareness, friends, family and, of course, fun. We are so thankful that so many families were able to connect and enjoy a relaxing evening with new friends. The game brought in nearly $4,000 in donations to FIRST. Wow! What a difference you all are making.

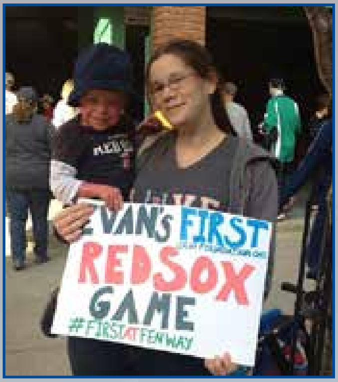
Washington Nationals

It was an amazing turnout in Washington, DC at the FIRST Night Out at the Nationals game on May 5th. Although there were only four "dry" innings of baseball, by all accounts, everyone had a great time meeting new friends and connecting with new families affected with ichthyosis. We'd like to extend our gratitude to the Beard family for all their efforts in making this FIRST Night Out a very special one. Melissa Beard also commented, with regard to the evening, "We even had the chance to explain the disease to others. All in all it was a great experience!" Over $500 was raised for FIRST.

Cleveland Indians

Emma Klima, who was born with epidermolytic ichthyosis (EI), and her family and friends cheered on the Cleveland Indians at the second annual FIRST Night Out at the Indians as the Tribe played the Chicago White Sox on Friday, September 5th at Progressive Field. They brought home a winner for the Tribe, with a bases-loaded walk-off single to win the game 2-1 in the 10th inning and a win for FIRST with donations totaling close to $400. A big thank you to Margot Klima for organizing this wonderful evening and to all who participated!

Boston Red Sox



FIRST Red Sox fans hit the field for some fun and fundraising. The Fascianos, whose son Evan is affected with harlequin ichthyosis (HI), the Hamills whose daughter Lauren is also affected with HI, and the Robinsons, whose daughter Sienna is affected with epidermolytic ichthyosis (EI), raised nearly $400 at FIRST Night at Fenway. Plus, the Red Sox honored Lauren Hamill with the “Leader of the Pack” recognition, giving her a game ball right on field! A scoreboard shout out for FIRST and ichthyosis awareness was a wonderful way to cap off an exciting night of baseball - aside from the Red Sox win of course. Much gratitude to these families for their passion and continued support, and for everyone who came out to raise awareness for a very worthy cause.

Houston Astros

A fun time was had by all at the FIRST Day Out at the Houston Astros on Sunday, May 18th. Thank you to the Sanders family, whose son Ruairi is affected with epidermolytic ichthyosis (EI), for helping to raise awareness and over $300, including $85 in online donations and a $5 donation for each of the 46 tickets sold. Also, a sincere thank you to the Astros organization for providing the great seats and all of the support throughout the event planning process, not to mention a winning game

Indianapolis Indians

It never ceases to amaze us how touched people can be by the ichthyosis community. In fact, after being introduced to ichthyosis and FIRST at the National Family Conference in Indiana, photographers Heath and Ashley Shanahan and videographer Daniel Scott felt compelled to get involved and help the cause in any way possible. Ashley, Heath and Scott had never heard of ichthyosis until they were hired by FIRST to take photos and video at the Conference. Shortly after the conference, FIRST learned that the Shanahans and Scotts would be coordinating a FIRST Night Out at the Indianapolis Indians on August 9th. But not only that, they graciously shared information about ichthyosis and the FIRST community online, and throughout their social medial channels as well. As for the game, in Heath’s own words, “The game on Saturday was fantastic. We sold 82 tickets, raising over $400 for FIRST, and at least four FIRST members and their families were in attendance. The weather ended up being perfect, and it was a really great game.” Plus, they were able to connect with FIRST members Beth and Joel Hampshire, and discuss the possibility of planning future activities. We are grateful to the Scholl, Conrad and Curtis families for showing their support and attending the game. And, of course, a big thank you to Shanahans and Scotts…as well as a warm welcome to the FIRST family!

Kansas City Royals

On May 17th, once again a whole crew of Ashlynne Biggs’ army of supporters attended Ichthyosis Day at the K to cheer on the Kansas City Royals, and to raise awareness for ichthyosis. Ashlynne is affected with ichthyosis en confetti, and this is the second year in a row the Cossel family has stepped up and organized Ichthyosis Day at the K. Friends and family enjoyed a pre-game tailgate, followed by a Royals win, where a fan even threw Ashlynne a game ball! “Ashlynne felt like a little queen again,” said mom Cora. Plus, this year the Cossel family added T-shirt sales, a penny drive at Cora’s place of business, and a diecast Nascar car for sale on E-bay. Donations totaled over $1,200! Much gratitude to the Cossel family, and to everyone who participated.

More Amazing Grassroots Events!

Thirty-One Gifts

A giant thank you to FIRST mom Mara Jackson, whose son Brady is affected with X-linked ichthyosis, for holding an online fundraiser. As Mara is a Thirty-One Gifts consultant, in August she hosted a “back-to-school” catalog sale featuring the coolest Thirty-One accessories of all – Chillicious Thermal Lunchbags. Twenty-five percent of all sales were donated to FIRST. Thank you Mara for all your support and for your super cool and creative fundraising idea!

Yankee Doodle Craft Fair

Nancy Hamill, grandmother to Lauren Hamill, affected with harlequin ichthyosis (HI), sold handmade items at the Yankee Doodle Craft fair in Billerica, Massachusetts, giving a percentage of the proceeds to FIRST. Thank you for all you do Nancy!

Online Bracelet Sale

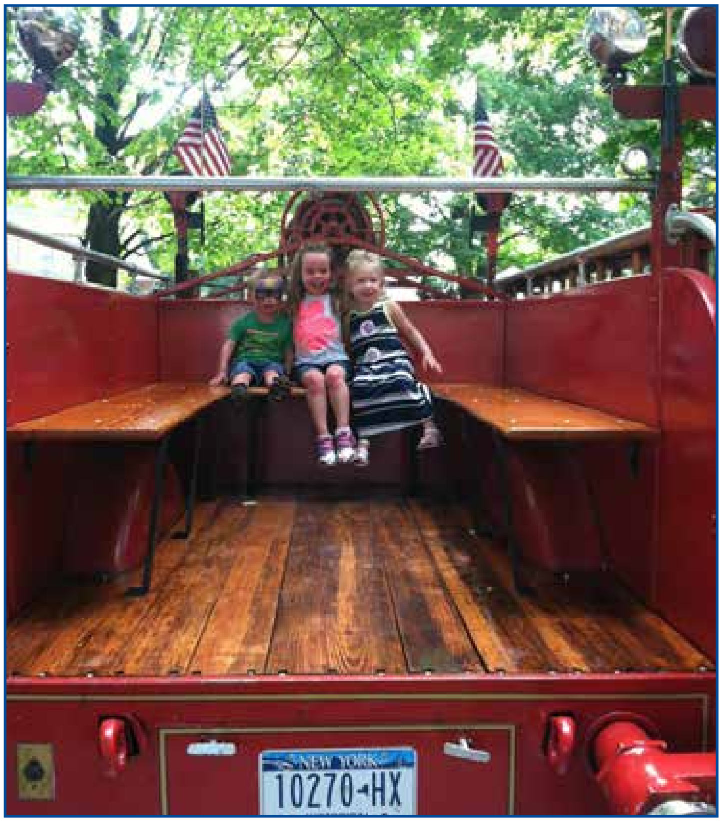
Member Sam Zavitz, owner of the The Pretty Chicken, held an online fundraiser with her beautiful “Be Brave” bracelets, donating 50% of the proceeds from the entire month of August to FIRST. Thanks so much for your continued support!

7th Annual Driving for a Cure Tournament

Members Christopher and Michelle Dugan once again coordinated a successful golf outing on June 28th in Spencerport, New York to benefit FIRST. Donations totaled over $1,300. Thank you so much for all of your hard work and continued support!

Car Show

A special thank you to the Straight Family of Troy, New York, whose son Aaron, and daughter Quinn are both affected with ichthyosis. For the fifth year, they have participated in the South Troy Timing Association Classic Car Show – this year alone raising $1,000 for FIRST (In total, they have raised nearly $5,000)! Way to go!



Why Join a *FIRST to Know* Call?

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 and talk with 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.

Become A Regional Support Volunteer

FIRST is building a stronger program within the Regional Support Network. In addition to being a support volunteer, FIRST members will be able to make an impact in areas of interest and specialty. Everyone is encouraged to be a Regional Support Volunteer (RSV), and to support each other through the sharing of knowledge, experience and advice through personal experiences. The role of a RSV strengthens the community overall. If you have an interest or experience in the areas of education, health care/insurance, physician recruitment, advocacy and fundraising, or if you want to host a small get together with other members in your area, we encourage you to sign up to be a FIRST Ambassador, and work together to strengthen programs and resources for the ichthyosis community.

For more information about these programs, visit the Regional Support Network page on FIRST’s website or contact Moureen Wenik, Program Director at mwenik@firstskinfoundation.org or at 1.800.545.3286.

A Spectacular Evening of Hope at the Release the Butterfly Benefit Concert

It was a magical evening in Johnsonburg, Pennsylvania, on September 27th, as singers and dancers took the stage at the Johnsonburg Area High School for "performances of hope," and a community united for the Release the Butterfly benefit concert. The concert was hosted by FIRST members and self-advocates, Tracie and Bailey Pretak. A special friend of the Pretaks and of FIRST, Hunter Steinitz, affected with harlequin ichthyosis (HI), participated in a unique "talk show" interview, along with Bailey. The interview focused on "living with ichthyosis," and was conducted by former student of Tracie Pretak and recent journalism graduate, Sara Vallone.

"Sara was so great at leading the conversation about ichthyosis. The questions and answers were amazing! The audience definitely learned a lot, and they were noticeably moved. They were changed forever because of the presentation," said Tracie.

There were over 60 people in attendance at the concert, raising $1,800 for FIRST. "Not bad at all for a "'village in the middle of nowhere!'" said Tracie. FIRST member Evan Melton affected with congenital ichthyosis, traveled from over an hour away to attend the event. "It's moments like that (noticing the Meltons in the audience) that remind us why it is so important for us to keep doing this," added Tracie.

Performances included:

Bailey Pretak (dancer, vocalist); Hunter Steinitz (vocalist); Tracie Pretak (vocalist, pianist); Ross Bish (vocalist); Megan Fannin (vocalist); Alexa Marciniak (vocalist); Carley Chiesa (vocalist); Sami Ferragine (vocalist); Haley Freeburg (vocalist); Anna Stanko (vocalist); Lily Deane (vocalist); Hannah Ross (vocalist); Megan Inghram (vocalist); Todd Fannin (vocalist, guitarist); Ginette Watts (vocalist); Ashton Watts (vocalist); Sara Vallone; Dancers from April School of Dance and Studio K. There was also a Chinese Auction with 16 items donated by local businesses and the Pretaks.

We are so very grateful to the Pretaks for their steadfast enthusiasm for the mission of FIRST. We'd like to also offer a special note of gratitude to all who participated in their spectacular Release the Butterfly event! Your support makes all the difference.

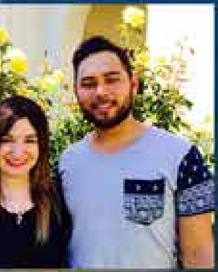
Dane’s Friends for FIRST

It was an evening of music and magic, wrapped in the bond of a community that cannot be broken. From the very first moment, it was clear that all of the attendees at the Dane’s Friends for FIRST concert at the Heritage Theater in Campbell, California, were not only abuzz about the incredible silent auction and upcoming performances, but each had been personally touched by the story of Dane Phelps. It was hard not to hear the sharing of special memories amongst the crowd, or notice the curiosity and hope that has captivated this community. They were eager to learn more about the rare condition that affected Dane, the mission of FIRST, and how they could best be of service to the cause of FIRST.

Dane’s Friends for FIRST is a fundraising concert event that was inspired by the life and memory of Dane Christian Phelps. Dane was affected with ichthyosis along with heart issues, tragically leading to unexpected heart failure in 2008, at the precious age of 3 ½ years old. For the past three years, the Phelps family has hosted a concert near their community of San Jose, California, to honor Dane’s memory and to serve as a fundraiser for FIRST.

As in the past two years, the 2014 Dane’s Friends for FIRST Concert was as enchanting an evening as ever. The tone for the night was set with an inspiring video montage of the 2014 FIRST Family Conference, followed by an enticing orchestra performance. Soon after, a parade of dancers, including Chandler Phelps, and singers and musicians, including Cade Phelps, filled the stage for the rest of the evening. And in the midst of all the wonderful entertainment, the lovely Suzanne Phelps introduced a young woman named Grace Ryan, who had flown in from Hamilton, New Zealand. But unlike any of the other performers at this concert, or the two previous events, Grace is affected with epidermolytic ichthyosis (EI). Yet, when Grace took center stage, it was hard to notice anything but her angelic voice, as she sang the heartfelt song “Beautiful” by Christina Aguilera

Videos were shown throughout the evening highlighting the FIRST-funded doctors who have dedicated their careers to ichthyosis research, and special broadcast interviews of our members. The overall presentation offered a true picture of the kind of support and advocacy provided by FIRST to the ichthyosis community.



We’d like to thank all of the performers and the caring community of Campbell, California, and to offer our deepest gratitude to the Phelps family, for all their hard work and passion, and for their unwavering commitment to the mission of FIRST. Together, we will make a difference. Find out more about the Dane’s Friends for FIRST Concert and underwriting campaign at www.danesfriendsforfirst.com.

Performances and concert participants included:

Mentors/Teachers/Choreographers: Valley Christian Strings Ensemble; Valley Christian Jazz Ensemble; Angelic Harp Academy; Capitol Dance Company Choreographers and the Capitol Dance Company Dancers.

Musicians: James and Katie Nobriga with Cade Phelps; Ryan Sims, Ethan Newman and Andy Varner; Howi Dietz; Four Guys and a Girl; Eden in Exile; Mario Minardi; Mary Flinsbaugh; Joyce Randolph; Grace Ryan.

PLEASE CONTACT DEVELOPMENT COORDINATOR, CHRIS WASSEL, FOR ASSISTANCE IN PREPARING AND PROMOTING YOUR GRASSROOTS EVENT!

Doctors Connect Online to Diagnose and Advise

Discover how FIRST is Reaching New Heights in Telemedicine

Telehealth is the delivery of health care using technology to link patients and care providers who are in separate locations in either a synchronous or asynchronous fashion. It is an increasingly utilized and incredibly promising way in which to deliver care in situations where access is a problem.

Thanks to members of our Medical & Scientific Advisory Board (MSAB) and the Lennox Foundation, the FIRST Tele-Ichthyosis is now available to dermatologists and other health care professionals to upload questions, documents and images for input and consultation from ichthyosis and related skin type experts. The site uses a store-and-forward teledermatology approach in a secure, HIPAA compliant environment to facilitate communication between dermatologists and other medical professionals, dealing with this rare set of skin diseases.

But Tele-Ichthyosis offers more than a portal for doctors to connect and collaborate, it offers the promise that, one day, patients with ichthyosis and related skin disorders around the globe will have access to the care they need, at the time they need it the most. Below, Dr. Jonathan Dyer, a member of FIRST's Board of Directors and MSAB, and one of the key physicians behind the development of this telemedicine program, discusses the evolution of the Tele-Ichthyosis site and the potential and vision for the future of telemedicine.

"In the case of rare disorders, three of the most common complaints I hear from patients are:

1."Doctors never know what I have"; 2."I know more about my condition than my care provider," and 3. "There isn't anyone in my region who knows about my disease."

The Tele-Ichthyosis Program was created to try to address those specific issues. It grew directly out of my experiences at the FIRST National Family Conference, and the fact that the University of Missouri, where I trained and now practice, was one of the first universities in the country to have an ongoing and functioning telehealth network. As a faculty member I established one of the very few live interactive teledermatology clinics in the country.

After attending a FIRST National Family Conference and seeing what an amazing resource the screening days were for both the patients and their families, as well as the expert team of physicians so committed to learning more about their disorder and how to better care for these patients, I wondered if store-and-forward teledermatology might offer a way to use technology to have a "family conference-like" visit, when it was needed - rather than having to wait for the next meeting. Several of the members of the FIRST MSAB discussed this online concept at that very conference, and the idea grew into reality soon after.

I believe the Tele-Ichthyosis program has been an incredibly worthwhile endeavor. It is a resource that is relatively unique - even among patient support organizations. At present it serves as a resource for physicians from all over the country (and beyond) and has helped many patients.

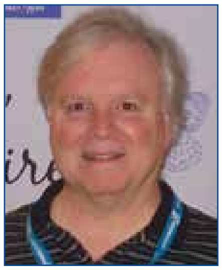
However, currently I envision what we presently have as a starting point. I think the technology can go much further. The old model of care delivery with a patient driving into a clinic (or flying, as I've had patients fly from quite far away to see me because of their skin conditions) is now just one way of doing things. More efficient modalities such as telehealth offer the potential of linking patients and providers (or teams of providers) all of whom may be physically separated by hundreds or thousands of miles.

Perhaps, one day, we'll even be able to coalesce systems so that patients will eventually have an "online medical home"– a network of care providers across the country who not only see patients in their physical clinics but also weigh in on more complicated clinical issues as a group, much like we do on the site. Imagine a monthly or quarterly online "grand rounds-type" event where doctors are able to connect and weigh in on patients, or a 24-hour 7-day-a-week harlequin hotline for that rare surprise of the birth of a harlequin infant (or severe collodion). There is much potential and possibility with this type of technology and much hope for the future of telemedicine."

Educate

Ichthyosis Research Update…

Dr. William Rizzo, Ichthyosis and 12R-Eicosanoid Metabolism in Sjögren-Larsson Syndrome

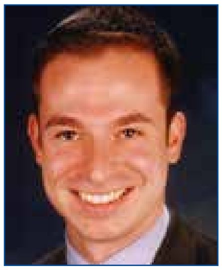


In 2006, Dr. William Rizzo of the University of Nebraska Medical Center in Omaha, Nebraska, was funded by FIRST for his project focusing on Ichthyosis and 12R-Eicosanoid Metabolism in Sjögren-Larsson Syndrome (SLS). This research investigates a defective lipid (fat) metabolic pathway that is seen in SLS and several other genetic forms of ichthyosis.

"Our research may lead to new approaches for cutaneous therapy for selectively bypassing the metabolic block in lipid metabolism and providing the metabolites that cannot be made by SLS patients," said Dr. Rizzo.

Most recently, Dr. Rizzo has partaken in the STAIR Consortium, an international multi-center, collaborative research project focusing on genetic diseases that are caused by defects in Sterol (cholesterol) And IsopRenoid metabolism. "The funding from FIRST in 2006 helped us to bring the research to a new level and to the point where the NIH was interested in funding this unique, collaborative effort."

The STAIR Consortium's goal is to establish the natural history of rare diseases, identify biomarkers for future therapy studies, investigate new treatments, discover new diseases, and train new physicians/researchers to work on rare diseases. Dr. Rizzo, one of the world's leading researchers of Sjögren-Larsson syndrome explained, "Access to biological data from as many patients as possible is critical for understanding the disease." He further emphasized that the input from patient advocacy groups and their membership base is a necessary part of this type of research collaboration. STAIR is currently working with seven patient advocacy groups worldwide, including FIRST.



Dr. Keith Choate, Yale University Gene Discovery Project

In 2008, FIRST awarded Dr. Keith Choate of Yale University our inaugural Clinical Scholar Award, enabling him to continue his work, eventually receiving additional funding from other foundations including the Doris Duke Charitable Foundation. FIRST's award and support has been instrumental in the sustained development of Dr. Choate's Gene Discovery Project. In fact, Dr. Choate's project began in a small room at the 2010 FIRST National Family Conference in Orlando, Florida. Since that time, the research has incurred tremendous growth, recruiting 375 total families and, so far, determining a genetic diagnosis for 247 of those families. Since 2012, Dr. Choate and his team have also identified three new genes which cause ichthyosis.

Additionally, recent advances in genetic sequencing technology has made genetic diagnoses faster and much less expensive. Of the 247 families who were able to obtain a genetic diagnosis from Dr. Choate and his team, 80 percent of them were able to get that diagnosis through a "pre-screening" process, which looks at the 11 most common genes that cause ichthyosis. Learning more about the specific genetic causes of ichthyosis will enable future research to develop effective therapeutic pathways for treating ichthyosis," says Dr. Choate.

Dr. Amy Paller, Topical Delivery of Keratin 10 Mutation-Specific siRNA-Gold Nanoparticles for Epidermolytic Ichthyosis (EI)

In 2012, Dr. Amy Paller and her team at Northwestern University were awarded a $75,000 grant, allowing further research to deliver an siRNA that specifically recognizes the common mutation of EI, R156H, with the intent to turn down production of the abnormal protein, while maintaining production of the normal protein. Dr. Paller, has inspired new hope for silencing the underlying mutation of EI and its changes to the skin surface. She and her team have discovered that siRNA, attached to a "central 13 nm gold nanoparticle" can be rubbed into the skin in a simple topical ointment.

As of late, Paller reports "we are using a new nanotechnolgy-based technique called 'spherical nucleic acids' (SNAs) to suppress the production of the abnormal keratin 10 gene that is the most common change leading to epidermolytic ichthyosis (EI). We continue to screen candidate SNAs to find a few that clearly suppress the abnormal keratin 10 gene much more than the normal keratin 10 gene. In the meantime, we have developed several tools towards this effort, which can also be used by other researchers. Most recently we've developed a special 'lentivirus reporter construct' in which we can see through changes in fluorescence whether or not our SNA works."

"We just received a grant from the National Institutes of Health (NIH) to continue this effort based on our preliminary data collected with FIRST's funding support,"Paller added.

Aldeyra Therapeutics Planning a Clinical Trial for Skin Disease in Sjögren-Larsson Syndrome

ALDEYRA THERAPEUTICS™

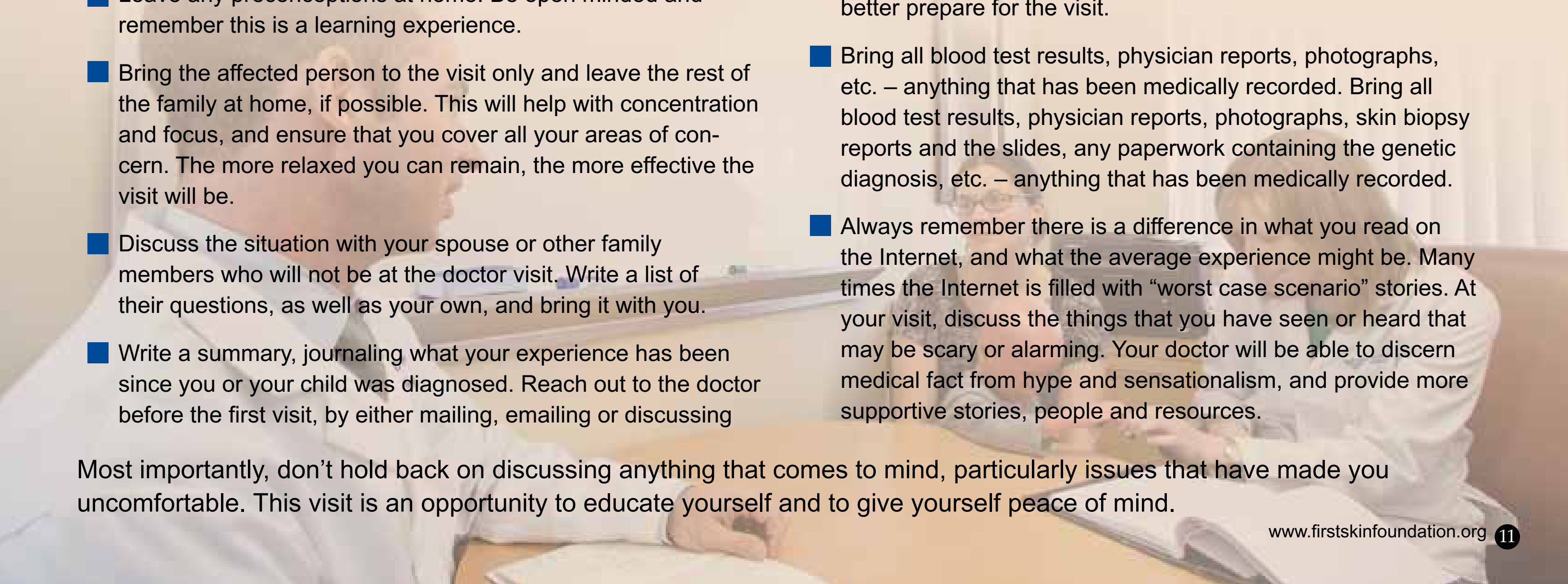
Aldeyra Therapeutics is a biotechnology company focused primarily on the development of products to treat diseases thought to be related to endogenous free aldehydes, a naturally occurring class of toxic molecules. One of these diseases is a rare genetic condition called Sjögren-Larsson syndrome (SLS). Unfortunately, there are no FDA-approved therapies for SLS patients. However, Aldeyra is in the process of preparing for a clinical investigation for the topical treatment of a skin condition known as ichthyosis secondary to SLS.

SLS is a rare inherited disease characterized by congenital ichthyosis (scaly, thickened skin), neurological problems and retinal disease. SLS is the most widely recognized form of neuro-ichthyosis, and is caused by the deficiency of an enzyme called fatty aldehyde dehydrogenase (FALDH), which breaks down naturally formed chemicals known as fatty aldehydes. In SLS patients, FALDH activity is deficient, resulting in an accumulation of fatty aldehydes that are toxic. The end result is disruption of the normal function of the skin and the development of ichthyosis associated with SLS. It is thought that the removal of the excess fatty aldehydes in patients with SLS will lead to an improvement in ichthyosis and other cutaneous disease symptoms.

Aldeyra is in clinical development of a drug called NS2 that is designed to trap free aldehydes, and plans to file an Investigational New Drug (IND) application for clinical testing of NS2 in 2014 for the treatment of SLS. NS2 has not been approved for sale in the Unites States or elsewhere. Aldeyra plans to conduct a treatment study in patients with SLS, which is currently expected to be initiated in 2015 in the United States. SLS patients will be treated topically with NS2 cream for two months, and safety and preliminary efficacy will be evaluated.

How Can I Prepare for My Visit with an Ichthyosis Medical Expert?

So, you've finally scheduled an appointment with an ichthyosis medical specialist, but have no idea as to how to prepare? No worries, our physicians will guide you step-by-step on how to have the most effective visit possible. At the FIRST National Family Conference in Indianapolis this past June, we sat down with Drs. Keith Choate and Philip Fleckman and asked them that very same question. Below is a summary of what they shared regarding best practices for preparing to meet with an ichthyosis specialist:



- Educate yourself as much as possible beforehand. The FIRST website is a wonderful resource with abundant information on both the clinical and emotional aspects of many types of ichthyosis and related skin types.
- Leave any preconceptions at home. Be open minded and remember this is a learning experience.
- Bring the affected person to the visit only and leave the rest of the family at home, if possible. This will help with concentration and focus, and ensure that you cover all your areas of concern. The more relaxed you can remain, the more effective the visit will be.
- Discuss the situation with your spouse or other family members who will not be at the doctor visit. Write a list of their questions, as well as your own, and bring it with you.
- Write a summary, journaling what your experience has been since you or your child was diagnosed. Reach out to the doctor before the first visit, by either mailing, emailing or discussing it with them over the phone. Let them know the exact genetic diagnosis if you have that information, symptoms, concerns, and specifically how ichthyosis is affecting your lifestyle. Writing it down may also take some of the emotion out of the story, so you can remain focused, and also help the doctor to better prepare for the visit.
- Bring all blood test results, physician reports, photographs, etc. – anything that has been medically recorded. Bring all blood test results, physician reports, photographs, skin biopsy reports and the slides, any paperwork containing the genetic diagnosis, etc. – anything that has been medically recorded.
- Always remember there is a difference in what you read on the Internet, and what the average experience might be. Many times the Internet is filled with "worst case scenario" stories. At your visit, discuss the things that you have seen or heard that may be scary or alarming. Your doctor will be able to discern medical fact from hype and sensationalism, and provide more supportive stories, people and resources.

Most importantly, don't hold back on discussing anything that comes to mind, particularly issues that have made you uncomfortable. This visit is an opportunity to educate yourself and to give yourself peace of mind.

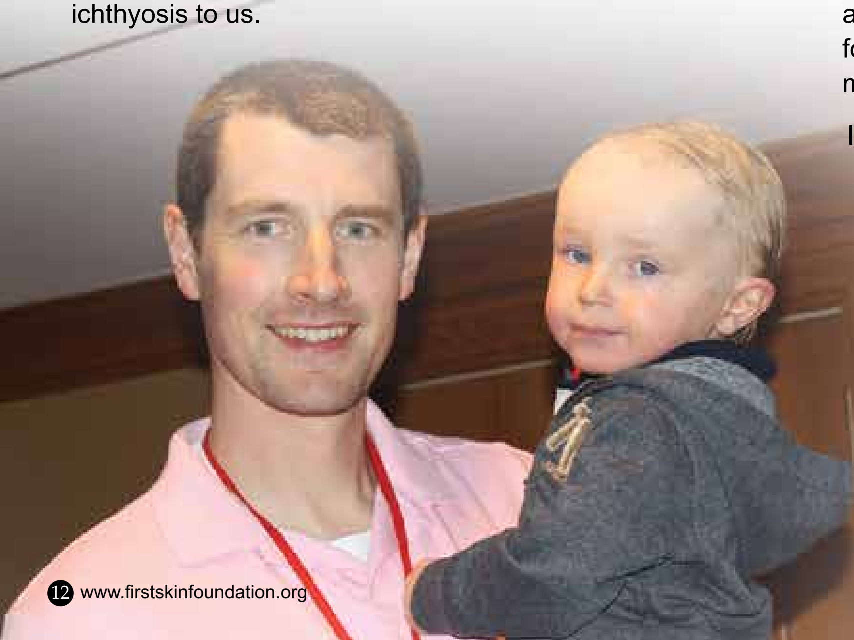
Meet Our Members

Andrew Sanders Running Aramco Houston Half Marathon for FIRST! -- January 18, 2015

It is always exciting at FIRST when new members are inspired in such a way that they immediately embrace our community, making the most of our services, resources and opportunities to connect. In June 2013, we met Andrew and Heather Sanders for the first time, along with their son Ruairi, who is affected with epidermolytic ichthyosis (EI), at a regional support meeting in Dallas, Texas. Ruari was just 6 months old. In a little over one year's time, they have made numerous connections within the FIRST community, providing support for others and enthusiasm for our advocacy efforts, in every possible way. Today we are sharing the story of why Andrew, a man who self-reportedly does "not have legs for running," will run 13.1 miles, to say thank you…

"Heather and I had decided we would try to run a marathon when we moved over to the United States. We had originally applied to do a full marathon in 2012, but our arrival to the states was delayed by a few months, so we decided to defer. I ran the Houston half marathon in 2013, not long after Ruairi was born. It was particularly hard to keep my training going on with little to no sleep in the early months! Obviously, Heather couldn't run in 2013, because it was only a couple of months after Ruairi was born. She did run her first marathon in 2014 and managed to beat my time! So, needless to say, I have extra motivation for 2015!

But our story with FIRST began after Ruairi was born in 2012. We were obviously aware of the fact that his skin wasn't as we'd expected. It was very red in places and sort of paper-like in others. The doctors ran over a number of possible explanations, many of which were deeply concerning, and told us that he would be taken to Texas Children's Hospital in downtown Houston. Before he was taken by ambulance, one of the neonatologists mentioned the possibility of ichthyosis to us.



I was aware of ichthyosis, having seen a documentary back home in the United Kingdom about a family in England who had two daughters with harlequin ichthyosis. When we Googled the term ichthyosis, the image results primarily showed babies with harlequin, but we knew that Ruairi didn't have that particular form of ichthyosis. But we were obviously still very worried about him. Heather quite quickly found her way to both FIRST and the UK ichthyosis support networks and we have found them to be an amazing resource. Indeed Heather is very active in the Facebook community, both with friends whom we've met at FIRST National Family Conferences and with new members. We had a great time at the 2014 FIRST Family Conference - Indianapolis this past summer. We met some amazing people and learned a great deal.

We were even motivated to organize a fundraiser day at the Houston Astros a few months ago, which we were more than pleased to do. It's really great that MLB teams do this for charities. It both brought some of the local families who are affected by ichthyosis together, while raising some awareness among those who came to the table and picked up a wristband or some literature. Obviously, there is still a long way to go. It was just this weekend that I had some harmless but frustrating comments from strangers that Ruairi looked like he'd gotten too much sun. My stock response is to tell them that he has a skin condition and, no, I haven't let my 2-year-old get a second degree burn, although I tend to only think the latter part of that line!

In all honesty, I'm running the 2015 Houston Half Marathon, in hopes that I can raise some money to help support FIRST by way of thanks for the support they have given us. I'm sure I can get some of my friends to sponsor me on the basis that a half marathon is a challenge for anybody, but particularly a former basketball player of 6'8"! My long legs are not really made for running.

In the longer term, I'm sure like most people reading this, I really hope for a cure. I'm confident that a good amount of research into genetic conditions is already happening and that people who deal with ichthyosis can benefit from scientific discoveries elsewhere. Obviously, this all comes down to money. Perhaps the money I raise can also be put toward research. But perhaps just raising a little awareness will also do some good! A donation link has been set up so you can join us in supporting FIRST and making a difference. Visit FIRST's website to make a donation."

-Andrew Sanders
Cypress, Texas

FIRST Honors David and Valerie Scholl at 2014 Testimonial Dinner

Over fifty guests joined FIRST, as we proudly honored David and Valerie Scholl, on September 6th, at FIRST's Annual Testimonial Dinner at the Heritage Club in Mason, Ohio. The Scholls have been fiercely committed to the mission of FIRST since their very first introduction to the organization nearly 10 years ago when they became the proud and loving grandparents to their first-born grandchild, Payton Scholl. Payton was born with ARCI-CIE type ichthyosis. From that day forward, they both have taken an extremely active role in the FIRST organization.

Dave has been a dedicated and enthusiastic member of FIRST's Board of Directors for nine years, serving as Board President from 2004–2011. Valerie, a kind, caring and compassionate advocate for FIRST, has been a dedicated volunteer in our Ambassador Program, as well as an active participant in the Regional Support Network for Region 4. As individuals they are outstanding advocates; as a couple, they possess a truly unique blend of compassion and drive… like no other. ***FIRST was delighted to present them with this honor.***

Patient Support Forums

Come out for a day of networking with other families affected by ichthyosis and related skin types. Meet other families in your area and learn how to build a stronger network of connections, advice and support in the ichthyosis community. These 1-day forums are filled with medical discussion, research updates, networking, product sharing and skin care routines. The forum includes a light breakfast, lunch and childcare for families with small children. Registration information and cost will be posted on FIRST website and Facebook page.

Meetings for 2015 are planned for: Arizona, Florida, Minnesota and Ontario. Details coming soon…

Jane & Henry Bukaty Skin Care Fund

Thanks to the generosity of Jane and 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 application can be downloaded from FIRST's website and requests the specific product/treatment for which funds are needed, and a demonstration of the financial need for this product/treatment.

Awards will not exceed $200. Applications will be awarded two times per year as determined by the Review Committee. Applicants will be eligible to receive one award every two years.

Please download the application from FIRST's website and email to the national office at jpickford@firstskinfoundation.org, fax to 215.997.9403 or mail to the attention of:

Jane & Henry Bukaty Skin Care Fund Foundation for Ichthyosis & Related Skin Types, Inc.®
2616 N. Broad Street, Colmar, PA 18915
The deadline to submit an application is January 31, 2015. The next cycle of funds will be awarded in February 2015.
You will be contacted by the office if you have been awarded aid from this fund.

Congratulations to 2014 UFIRST Scholars!

FIRST is proud to announce this year's 10 scholarship recipients funded by the UFIRST Scholars Program. Congratulations to everyone. We wish you the best of luck as you continue your education.

Atique Ahmed

ARCI-lamellar type ichthyosis

Al Hikmah Institute of Sciences

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, i.e. AIDS, ichthyosis."

Hannah Allen

Congenital ichthyosiform erythroderma (CIE)

College of the Ozarks

Goals and aspirations: "I plan to graduate college in 2015 with a bachelor's degree in psychology. I want to get a job in the human resources field."

Sarah Beamish

Netherton syndrome

University of Delaware

Goals and aspirations: "I want to be able to understand as much about life as I possibly can. I hope that by studying biology and psychology, I will be given answers that satisfy my curious mind."

Maria Gad

ARCI-lamellar type ichthyosis

Ohio State University

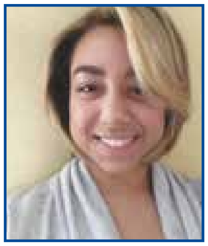
Goals and aspirations: "I aspire to spend every day of my life helping others. My career goal is to become a pediatric dermatologist that can help others with rare skin diseases and encourage them to persevere."

Elizabeth Joyner

Ichthyosis vulgaris

Union University

Goals and aspirations: "I look forward to teaching or working with students in whatever setting God calls me to. I can use a degree in education as a teacher in the classroom as well as a youth leader at a church. I hope to be able to use what I have learned to encourage students and let them know they are all special in God's eyes."



Taylor Mahoney

Ichthyosis vulgaris

Ohio Dominican University

Goals and aspirations: "After college I plan to move to a big city and work in fashion and entertainment. I also want to share my story in some powerful way."

Gina Messer

ARCI-lamellar type ichthyosis

University of California, Davis

Goals and aspirations: "I would like to go to grad school so I can earn my PsyD, then go on to become a licensed psychologist. My goal in life is to help others and to live life to the fullest!"

Kathleen Smith

ARCI-lamellar type ichthyosis

Lawrence Memorial Regis College

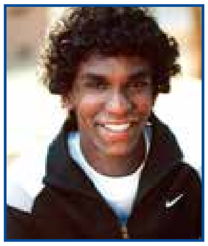
Goals and aspirations: "I want to be the best nurse I can possibly be for my patients. I want to work in the places that are most in need around the world and improve access to health care for women and children."

Hannah Suda

ARCI-lamellar type ichthyosis

Bemidji State University

Goals and aspirations: "My goals are to attend Bemidji State University to receive a BSN degree in nursing."



Mani Woodward

X-linked ichthyosis

University of Oregon

Goals and aspirations: "I would like to earn degrees in human physiology and Chinese at the University of Oregon and then continue to medical school where I would like to pursue a career as a pediatrician or a dermatologist."

Change How You See: Positive Exposure and the PEARLS Project

Many of you are familiar with FIRST friend Rick Guidotti, our keynote speaker at the 2014 National Family Conference - Indianapolis, and the Founder and CEO of Positive Exposure (PE). Not only has our community been inspired and uplifted by Rick and Positive Exposures "Change How You See. See How You Change." message, but the synergy between the FIRST community and PE continues to blossom, creating opportunities for both organizations to thrive. Most recently, our community has been invited to participate in their PEARLS (Positive Exposure Ambassadors Real Life Stories) project, which takes a very unique approach to teaching about living with differences and rare disease.

The PEARLS Project is a ground-breaking educational tool which invites students to learn about their peers living with genetic, physical and behavioral differences through an image gallery and safe online blog. This trailblazing interactive program enables student audiences to gain a deeper understanding and respect for human diversity, while empowering their peer group living with difference to become self-advocates and teach the world about life from their unique perspective.

How can I get involved?

PE is looking for volunteer PEARLS ambassadors. During your commitment of being a PEARLS Ambassador, you will be expected to blog a minimum of once a week for 16 weeks. These blog entries are essentially "journal entries." Topic ideas for blog entries include; a day in your life, thoughts about the world around you and events in your life. Photos, videos, artwork, poetry and other creative additions can also accompany your blog entries. They can be as long or as short as you like. Family members are also welcome to submit a blog on your behalf. With your submissions, PE will create a web page specifically for you. You will be given your own username and password. Additionally the entire PEARLS database is password protected, as it is an interactive program enabling students from selective host schools to gain a deeper understanding and respect for human diversity. Through a series of projects, lower, middle and high school students can better relate to their peer group living with differences.

The PEARLS project also has programs for medical students and other health care professionals-in-training, professional development for teachers and administrators, parents, siblings, families, communities and the broader public. For more information or to fill out an application to become a Pearls Ambassador, please contact Maureen Neville, Communications Director, at mneville@firstskinfoundation.org.

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