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Ichthyosis Focus

Winter 2015 A Quarterly Journal for Friends of FIRST Volume 33, No. 4

Use Your Voice

How one family has taken self-advocacy to a whole new level.

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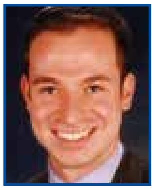
Happy New Year!

Jean R. Pickford
Executive Director

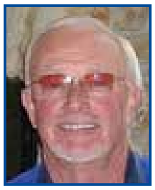
As I reflect on the accomplishments of a spectacular 2014, I am also looking ahead to a world that holds more promise for those affected with ichthyosis than ever before. In this issue, you'll meet a family whose 13 year commitment to fundraising has inspired the entire FIRST community. Plus, you'll learn the basics of gene therapy, and read about the fascinating 'scaling pathways' research being conducted by one of our 2014 Research Grant Recipients, Dr. Ryan O'Shaughnessy. You'll also find the latest discovery from the Yale Disorders of Keratinization Project, and meet a longtime member of FIRST who has made his own new discovery: acceptance.

Jeff Hoerle

I would like you to join us in congratulating Jeff Hoerle, the new President of FIRST's Board of Directors and Dr. Keith Choate, who will be joining the Board. And, as we welcome our new volunteers with open arms, we are also bidding a heartfelt farewell to former Board President, Mike Briggs, and former CFO, John Schoendorf, both to whom FIRST is eternally grateful for their commitment and outstanding contributions. Although Mike and John will no longer be serving FIRST in a Board capacity, they will continue to be some of our best advocates and fundraisers. Both Mike and John concur, "I have considered it a privilege to have served with such a fine, dedicated group of people and will be available in the future for whatever capacity or assistance you may need!"



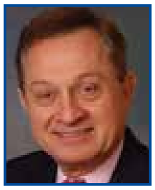
Dr. Keith Choate



Mike Briggs

I'm very much looking forward to 2015, as it is shaping up to be another banner year for FIRST. Our next four-year strategic plan will be finalized, four Patient Support Forums are scheduled, and our focus on research has never been stronger!

Best wishes for a healthy and wonderful new year,



John Schoendorf

Jean

Jean R. Pickford

Correspondence Corner

Dear Friends at FIRST:

I would like to thank you for the financial support to purchase Dermal Therapy products for my ichthyosis vulgaris. I can`t thank you enough. I am 52 years old and I`ve had ichthyosis all my life. All the while I`ve only been using lotions or creams from the drugstore or grocery store. And they don`t help. I hope others will find the help and support they need for this condition. I am truly grateful and glad to know that there are others out there who care for people with this skin condition.*

Maria Lourdes Salas
Los Angeles, California

***Editor's Note:** To apply to receive funds from the Jane & Henry Bukaty Skin Care Fund, visit FIRST's website, www.firstskinfoundation.org.

UFIRST Scholars Program
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"We have been using it about 6 days a week. It has done wonders for her! Nothing else I've used even comes close to getting the amount of dry skin exfoliated from her body. I've tried spa mitts, loofah pads, and scrub brushes. The Mitt works SO MUCH BETTER than anything I've used before. Since using the MicrodermaMitt, her skin doesn't dry out as fast and she doesn't get nearly as itchy during the day."

"The MicrodermaMitt is AMAZING!! I cannot tell you enough how happy I am that I found this mitt for my daughter. After our first bath using it, I was speechless! Her skin was soft, smooth, shiny and so healthy looking. Her skin has been wonderful since we started using the Mitt. It looks so much smoother and virtually flake free. The skin also comes off so easy without much effort."

"After just one use, my 8 year old smiled and said "look at my new skin." It just works perfectly. Prior to using the Mitt, we were using acid-based cream and sand paper or manually peeling and cutting the skin off. Her skin is now smoother, shinier and healthier."

PAID ADVERTISEMENT

Use Your Voice!

How one family has taken self-advocacy to a whole new level.

What is self-advocacy? Self-advocacy is individuals speaking up for themselves, speaking up for others, and taking charge of their own future in any way possible. And, although FIRST is there every step of the way, there is not a single more resounding voice than that of an affected member or family.

We are proud to say that there are so many families in the FIRST community who embrace self-advocacy by making incredible efforts to use their voice, raise awareness, and raise funds for a cause that is so very close to home. They are heroic and inspirational, to say the least. However, what we are now discovering is that new families in the FIRST community can benefit greatly from families who have been self-advocating through unique special events and local media connections, for many years.

For the past 13 years, one family in particular, the Johnsons of Friendswood, Texas, have not only educated more and more individuals about ichthyosis and grown their community following, but through trial, error and sheer will, they have been able to gain the crucial attention of the media, inspiring them to share their story-one of hope, commitment, and living with rare disease.

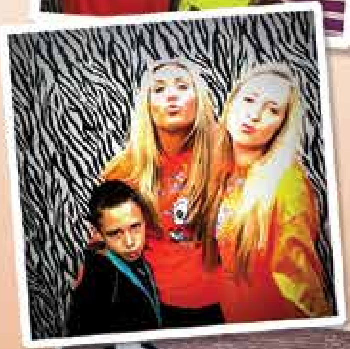
Monster Dash, 2014

This year marks the 13th year that Dawn Johnson, her husband Joe, and children, Jordan and Dawson, have hosted a Monster Dash/Fun Run 5K to raise money for FIRST. Dawn's daughter Jordan is affected with epidermolytic ichthyosis.

On October 25th, their incredible army of friends, neighbors, and family members, nearly 300 in total, gathered together at Friendswood High School. This year's event was not only a huge success but proved to be a day of endless fun! A photo booth added a unique new twist that kept smiles on faces all day long, in addition to the goody bags, well-branded Monster Dash T-shirts, a fun run, and the fact that everyone was gathering for a very worthy cause. This year's Monster Dash raised $25,000, which was their highest amount to date!

Dawn's success in promoting her event to the media included a robust social media campaign along with securing a KHOU TV, Houston segment, which was a fantastic human interest piece that aired after the event on the 10PM news. "The fun run has grown and evolved in ways I simply can't put into words. Each year has its own unique touch and leaves an impression on our hearts." stated Dawn.

FIRST would like to extend a giant heap of gratitude to the Johnsons and the caring community of Friendswood. Your commitment and generosity has made a difference in the lives of so many.


Grassroots Fundraising

Shopping with Santa!

'Tis the season to shop with Santa! Rhianon Miller, mother of 3-year-old daughter Evelyn, who is affected with ichthyosis, hosted a bake sale at the Shop with Santa craft fair in New Berlin, Wisconsin. Members of the Green family, whose three children, Michael, Aurora, and Matthew, are affected with ARCI-lamellar, traveled from their home in Illinois to join in the fun as well. The event raised $450 for FIRST! Many thanks to the Greens for supporting the event, and also to Rhianon for her efforts planning the day.

Pumpkin Painting in South Dakota

The Iott family held their 7th annual Pumpkin Paint and Bake Sale on October 18 at the Flandreau Aquatic Center. Pumpkins were donated by Iotts Greenhouse, whose owners are the grandparents of Rylee Iott, affected with ARCI-CIE. Participants painted pumpkins and enjoyed sweet goods that were donated by friends and family members. Friends and family members donated the baked goods for sale.

In addition to their annual Pumpkin Paint, the Iott family also hosted their annual coed softball tournament, held at the Flandreau City Park. Teams entered the tournament for an entry fee and played softball throughout the day. After the tournament, close friends of the family grilled pork ribs, chicken, bratwurst, and potluck. A free-will donation bucket was placed at the serving line. All totaled, the Iotts and their family and friends raised almost $1,800 for the Foundation.

Many thanks for the Iott family to their continued dedication and hard work on behalf of FIRST.

Beading for Research

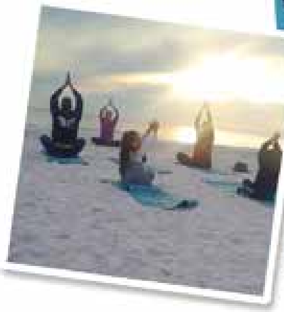
The Mayone family has continued their fantastic partnership with Caravan Beads in Portland, Maine, to host another fundraising event for FIRST. Each year, the event is held in honor of Mark and Kim's son, Evan, who is affected with epidermolytic ichthyosis. Grandpa and Grandma Patti Sundik always help out and can't resist purchasing some of the beautiful beads.

A Bonfire on the Florida Beach

Several families in Florida combined efforts to create a fun and memorable Beach Bonfire and Yoga event at Santa Rosa Beach. Jessica Nicolosi, Cara Tundidor, her sister Amber Rodriguez, and Tamika Green joined forces on this memorable evening in support of their children. Gabby Nicolosi and Teofilo Tundidor are both affected with ARCI-CIE type, and Mykail and Zaria Green are both affected with ARCI-lamellar type ichthyosis.

Participants enjoyed hot dogs, drinks, s'mores, crab hunting and music on the beach. Jessica also led a sunset yoga session. Families had a wonderful time around the bonfire, raising funds for FIRST and awareness about ichthyosis.

The event raised almost $2,000, of which Kohl's Department Stores donated $1,000. Thank you so much to all of the families who worked so diligently to make the evening such a success.



Volleyball Tournament in Vestal, New York

Nicole Ewing is a teacher at Vestal High School in Vestal, New York. Her son, Liam is affected with ichthyosis. For the second year in a row, the students at the high school hosted a volleyball tournament during November to raise awareness about Liam and ichthyosis, but also to raise money to support FIRST's efforts. This year's tournament raised more than $1,000. Kudos to the students and staff at Vestal High School for a job well done!

Tea Sales in the TUB

Longtime FIRST supporter and advocate Hunter Steinitz, affected with harlequin ichthyosis, is attending Westminster College in New Wilmington, Pennsylvania. One of the clubs that Hunter belongs to is the Disability Awareness Club. The group decided to host a tea fundraiser during the fall, selling cups of hot tea to students during lunch at the hangout known as the "TUB." The tea sales went so well, that they expanded it for the entire week! Many, many thanks to Hunter for her continued work on behalf of FIRST, and also to her fellow students at Westminster College.

Go Jacky O Go!

Kudos and praise to FIRST member and veteran volunteer Jacky Osowski, who ran the Chicago Marathon on October 12th. Jacky and her husband Frank are the parents of 12-year-old Ben, who is affected with epidermolytic ichthyosis. Frank and Jacky created a social media campaign around the marathon - *Even though she won't finish first, Jacky's gonna finish for FIRST! Go Jacky O Go!!*

With Frank leading the way, friends and family members came out in full to support Jacky, both at the race and online, raising $4,000. What a fantastic accomplishment for both Jacky and FIRST!

For more ideas on how to create and promote your special event, or reaching out to the media to self-advocate and raise awareness, contact Chris Wassel at cwassel@firstskinfoundation.org. Be sure to also view our newly updated Grassroots Fundraising Guide on the Grassroots Fundraising page of FIRST's website -www.firstskinfoundation.org

Gene Therapy and Rare Disease

Today, one can hardly read the health and science section of a national newspaper, let alone any medical journal, without seeing a story related to the field of gene therapy, targeted drug therapy or genomics (the branch of molecular biology concerned with the structure, function, evolution, and mapping of genomes). But what exactly is gene therapy, and how does it apply to rare disease?

In simple terms, gene therapy is an experimental technique that uses genes to treat or prevent disease. In the future, this technique may allow doctors to treat a disorder by inserting a gene into a patient's cells instead of using drugs or surgery. Researchers are testing several approaches to gene therapy, including:

- Replacing a mutated gene that causes disease with a healthy copy of the gene.
- Inactivating ("knocking out") or suppressing ("knocking down") a mutated gene that is functioning improperly.
- Introducing a new gene into the body to help fight a disease.

Additionally, there is an increasing interest in the research of gene therapy as it pertains to rare genetic disease. According to a recent article on Medscape.com, Targeting Drugs for Rare Disease, Gayatri R. Rao, MD, JD, Director for the Office of Orphan Products Development (OOPD), states "…increasingly in the routine practice of medicine, we are seeing more patients with rare diseases. Part of the reason for that is an increase in the diagnosis of rare diseases. Before, we would cluster groups of patients into syndromes, and now we are realizing that there are actually different disease states."

Additionally, Rao notes, "Not only are more patients being diagnosed, but from a therapeutic standpoint, interest has increased in the development of therapies for rare diseases as a result of an increased understanding of genomics. We are able to target therapies, which has expanded interest in the treatment of rare diseases."

It seems the more gene mutation targets that are discovered, the more main stream diseases are fitting into the "rare disease" category.

With regard to the effect this evolving focus on orphan diseases will have on medical research in general, Rao states, "This will have an impact on every field of medicine. Moving forward, the lessons being learned from the orphan products group will be applied across the rest of the regulatory landscape and to medicine in general."

Although gene therapy is a promising treatment option for a number of diseases (including inherited disorders like ichthyosis, some types of cancer, and certain viral infections), the technique remains risky and is still under study to make sure that it will be safe and effective. Gene therapy is currently only being tested for the treatment of diseases that have no other cures.

Ref; Summar ML, Dorman DE. Why You Should Advocate for All Rare Diseases, Not Just One. Medscape. Published Dec 8, 2014. Available at: http://www.medscape.com/viewarticle/835741

Ref; http://ghr.nlm.nih.gov/handbook/therapy/genetherapy - Genetics Home Reference, Your Guide to Understanding Genetic Conditions

FIRST Research Grant Awarded to Dr. Ryan O'Shaughnessy, UCL Institute of Child Health, London, UK


Dr. Ryan O'Shaughnessy

Dr. Ryan O'Shaughnessy received $50,000 from the FIRST Research Grant Program to continue his work in targeting the scaling pathways in ichthyosis. The research specifically focuses on understanding the mechanisms that cause scaling, and subsequently increasing the options for treatment.

As explained by Dr. O'Shaughnessy, "Hyperkeratosis, or scaling, is a very common symptom in skin disease, with around 150 genetic ichthyosis and ichthyosis-related skin diseases leading to this thickening of the outermost layer of the skin. In many skin diseases this scaling is the most outwardly visible effect of the disease, and the one that patients would most want to treat. Current treatments for hyperkeratosis such as retinoids, although effective, are used without a full understanding of the molecular machinery behind the process. By better understanding the basis for scaling ("molecular mechanism") in ichthyosis, more directed protein or gene therapies could be developed that benefit patients, regardless of their specific genetic defect. Also, if possible, it would be particularly attractive to find therapeutic targets that already have a range of drugs that could be rapidly re-tooled to treat ichthyosis, providing dermatologists multiple therapeutic options to treat ichthyosis beyond the first-line treatment of retinoids.

Our previous studies on autosomal recessive congenital ichthyosis, the most severe form of ichthyosis, have identified key components of the molecular machinery that causes scaling. We have already determined a key cellular signal, interleukin-1 alpha, which plays a critical role in the process. We think this signaling pathway represents an attractive new target in the treatment of scaling. In this project, we will test three different drugs, including retinoids and aspirin, on their ability to reduce interleukin-1 alpha signaling and skin scaling in skin cells and 3D reconstructed skin models from ichthyosis patients. The drugs are tested either on their own or in combination. Our long term aims of this research are to more fully understand the mechanisms that cause scaling and, as a result, to increase the options available to treat scaling, not only in ichthyosis, but potentially in a wide range of skin diseases."

The Latest Update from the Gene Discovery Program at Yale


Dr. Keith Choate


Dr. Brittany Craiglow

Many, if not most human diseases have a genetic component. Thanks to advances in next-gen sequencing, a number of studies in recent years are beginning to unravel the role of de novo mutations — genetic variants that arise in a child but are not present in either parent, in human disease.

Drs. Keith Choate, Brittany Craiglow and their research team at the Yale University Disorders of Keratinization Project are employing the genetic tools to identify new genes causing ichthyosis and related skin types (aka "disorders of keratinization"). With collaborators Drs. Amy Paller at Northwestern, and Kim Morel and Christine Lauren at Columbia University, Dr. Choate's group has discovered a new genetic cause of the disorder erythrokeratodermia variablis et progresiva (EKVP). In three families, central to this discovery was finding novel dominant, de novo gene mutations in a gene called GJA1. Mutations in GJA1 had previously been found to cause a very different disorder, oculodentodigital dysplasia (ODDD), which involves faulty development in the tissue of eyes, teeth, fingers and nervous system. The EKVP subjects showed none of these ODDD features. GJA1 encodes connexin 43 (Cx43), the most widely expressed gap junction protein in the body, but the mutations found in EKVP appear to have a special role in the skin without effects on other organ systems.

"These findings reveal a critical role for Cx43 in epidermal homeostasis, and provide evidence of organ-specific pathobiology resulting from different mutations within GJA1," said Dr. Choate. "Especially important is the finding that this disorder has a relatively late age of onset, with completely normal skin until about 6 months of age. If we can understand how these mutations cause skin disease, we are hopeful that we might be able to restore normal skin function for our patients."

Other contributors to this work include Lynn Boyden, Jing Zhou, and Rong-Hua Hu, all part of the Yale team. The work was partly supported by the Yale Center for Mendelian Genomics.

Abstract available from Journal of Investigative Dermatology: www.nature.com/jid/journal/vaop/naam/pdf/jid2014485a.pdf

Atopic Dermatitis, Ichthyosis, Melatonin and Sleep Disorders

Whether it’s an itchy, uncomfortable child keeping the household on high alert throughout the night, or an adult with ichthyosis who simply can’t get comfortable, lack of sleep is among many of the issues facing those affected with ichthyosis. Recent results published by the American Academy of Pediatrics investigated the sleep disturbance of individuals suffering from atopic dermatitis (AD) also called eczema, a disorder which is commonly linked to ichthyosis vulgaris and Netherton syndrome), and found that levels of melatonin and allergen-specific immunoglobulin E (IgE) may play a role in the disturbance (see abstract below).

Understanding the similarities between AD and forms of ichthyosis is a current area of investigation. At the FIRST National Family Conference in Indianapolis, Dr. Amy Paller and her team from Northwestern University obtained blood and skin biopsy samples from volunteers. Dr. Paller’s team is collaborating with the Guttman-Yassky lab at Mt. Sinai to look at shared “biomarkers” that may drive the redness, thickening and itch of both eczema and various forms of ichthyosis and are expanding their investigation of sleep issues in eczema to ichthyosis. Dr. Britt Craiglow from Yale has found that the majority of individuals with ichthyosis are plagued by itching, which can translate into poor sleep. In addition, Dr. Gil Yosipovitch and his team at Temple is interested in itch of ichthyosis in comparison with eczema and psoriasis. We will provide updates as more research regarding treatment plans become available.

ABSTRACT:

BACKGROUND AND OBJECTIVES:

Sleep disturbance is common in patients with atopic dermatitis (AD). However, studies have largely been questionnaire-based, and the pathophysiology remains unclear. The aims of this study were to determine objective characteristics of sleep disturbance in children with AD and explore contributing factors and clinical predictors.

METHODS:

Sleep parameters were measured by actigraphy and polysomnography in 72 patients with AD and 32 controls ages 1 to 18 years. Urinary 6-sulfatoxymelatonin levels, serum cytokines, and total and allergen-specific immunoglobulin E (IgE) levels were also measured.

RESULTS:

The patients with AD had significantly reduced sleep efficiency, longer sleep onset latency, more sleep fragmentation, and less non-rapid eye movement sleep. Results from actigraphy correlated well with those from polysomnography. The AD disease severity was associated with sleep disturbance (r = 0.55-0.7), and a Scoring Atopic Dermatitis index of ≥48.7 predicted poor sleep efficiency with a sensitivity of 83.3% and a specificity of 75% (area under the curve = 0.81, P = .001). Lower nocturnal melatonin secretion was significantly associated with sleep disturbance in the patients with AD. Other correlates of sleep disturbance included pruritus, scratching movements, higher total serum IgE levels, and allergic sensitization to dust mite and staphylococcal enterotoxins.

CONCLUSIONS:

Poor sleep efficiency is common in children with AD and can be predicted by the Scoring Atopic Dermatitis index. Melatonin and IgE might play a role in the sleep disturbance. Further studies are required to explore the mechanisms and clinical implications, and actigraphy could serve as a useful evaluating tool.

Pediatrics. 2014 Aug;134(2):e397-405. doi: 10.1542/peds.2014-0376. Epub 2014 Jul 14. Atopic dermatitis, melatonin, and sleep disturbance.

Author information - Chang YS1, Chou YT2, Lee JH3, Lee PL4, Dai YS5, Sun C3, Lin YT3, Wang LC3, Yu HH3, Yang YH3, Chen CA3, Wan KS1, Chiang BL6.

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FIRST Member Finds Acceptance – Meet Kenny Krips


"I would rather have people acknowledge the 'difference,' than just stare or ignore."

Over the summer, FIRST member Kenny Krips had contacted the FIRST office to check in, say hello, and inform us of one important fact, "It was time to tell his story." After a few failed attempts to connect during a very busy fall, FIRST finally had the pleasure to meet with Kenny.

Kenny was born in the small southern New Jersey town of Glendora in 1970, a time period when very little was known about his skin condition (ARCI-lamellar ichthyosis), and even less known about its treatment. His childhood, once depicted in a documentary entitled "Kenny," was admittedly not ideal, as he not only was singled out for his skin condition, but was the child of divorce at a very early age.

However, to his good fortune, Kenny was surrounded by the love of family, an older brother and mother with whom he has a close and supportive relationship. "My brother was always there for me. Raising a child with a rare condition that no one has ever heard of, virtually on her own, my mom never once let on that she was anything less than strong and confident," he said. And, she taught him one of the most valuable lessons of all: Do not pity yourself. Respect yourself, and always present well.

And present well he does. Although Kenny admitted over the years he has experienced dark periods of frustration and low self-esteem, he now feels his life experience, including his ichthyosis, has brought him to a place of acceptance, with a good dose of wisdom. He is a friendly, well-spoken gentleman, with a new perspective on life, and an enviable passion for his work.

"In a way, I am blessed to be around trauma," Kenny proclaimed.

Although Kenny's degree is in interior design, he feels compelled to help people on a deeper level. As the business supply coordinator for the trauma unit at Cooper University Hospital, in New Jersey, he has ample opportunity to connect with people in a very deep and meaningful way.

"This woman came into the trauma unit, and she was in a car accident. She had Alzheimer's and she was very confused and agitated. I sat next to her and rubbed her foot. She did not ask what happened to me (lifting his hands up to show that they are very affected). She just sat, looked me in the eyes, and listened. And then she started to calm down. She seemed to really be affected by what I was saying," he said, further explaining why his job is so fulfilling. "The other day a man in the trauma unit stopped me and said, 'Thank you so very much.' For what? I asked. 'You asked me if I was OK and let me know you were here to help me and that made all the difference.' It really made me feel good."

Kenny credits his job as being a major influence for his newly found perspective. "There are so many parts of my job that make me feel good about being alive. And as my mom always said, "If you help one person in 44 years, you did your job."

He has also come to terms with the notion that "everyone suffers" in one way or another. "I do still get frustrated, sometimes. Like when I want to wear black or be outside on a hot day. But everyone struggles with something once in a while…finances, career, health issues. Everyone faces the fear of the unknown," he said as he explained that this realization has left very little room to feel sorry for himself. "I am lucky that I have a strong family unit, a job I love, and amazing friends."

With regard to how he now handles adverse reactions to his condition, Kenny adds, "I would rather have people acknowledge the "difference," than just stare or ignore. But if they do stare, or make a face, I just have to pardon them. Life is too good to worry about it."

When asked how his life would change if he suddenly did not have ichthyosis, Kenny joked, "Well, I'd never be out of a Speedo!" But then he added, "I am absolutely OK with who I am. I am not mad at who I am anymore, no longer stuck. I feel good about what I do and I want to continue to do that. I don't want to stop. I think the man upstairs has it all mapped out for me. Whatever time I do have here, I want to make the best of it...ichthyosis or no ichthyosis."

Why Donate to FIRST?

It's a fair and valid question. And, in fact, one of the most rewarding and exciting parts of the job for the staff at FIRST, is to provide the answer. Likely you are already aware that donating to FIRST will ensure the continuation of the Research Grant Program, which supports the top investigators in the field of ichthyosis research in their pursuits to find better treatments and an eventual cure. However, as the global leaders in ichthyosis advocacy and research, it is not only our job to bring public attention to this rare condition and support the advancement of research, but to also embrace those living with ichthyosis today, and to help them live a rich, fulfilling, and healthy life. Your donations make this possible.

Did you know that one of the programs supported by your donation is our Regional Support Network (RSN)? The RSN offers numerous connection opportunities including regional meetings, family conferences, *FIRST to Know* conference calls, and access to an extensive network of ichthyosis experts. It is one of the many programs funded by individual donations, corporate appeals, fundraising events, United Way and CFC donations, and private grants. FIRST does not receive any governmental funding; it is solely funded by individual and private donations.

Perhaps you have attended one of our events, been connected to a doctor or family in your region, or gained some insight about ichthyosis on a *FIRST to Know* conference call? If so, you may have a deeper understanding of the critical need for these types of personal connections, as well as the rare opportunity to build relationships with the top doctors in the field of ichthyosis. The RSN offers life-changing connections that may not occur in any other situation. Your donations make these connections possible.

When asked about the importance of the RSN, and how it has changed the lives of so many, Program Director, Moureen Wenik, said, "The ichthyosis community is a small and very close-knit community. I have witnessed the relief people feel when they have someone to talk to who really understands what it is like to have a child with ichthyosis, or be an affected adult. It is great to talk to someone who 'gets it' and doesn't have to explain ichthyosis. The RSN provides a safe place for families to connect, whether through social media, by telephone, or face-to-face at a meeting. Often they have never met another person who shares the disorder. The feelings of isolation for new moms, parents of children with ichthyosis, or affected teens or adults with ichthyosis, can be overwhelming. This support network gives people the opportunity to talk to others who have experience living with ichthyosis and to share their feelings with others who understand." Your donations help improve the lives of those in the ichthyosis community.

The RSN is only one of the programs supported by your donations. You can find more amazing ways your support is making a difference at www.firstskinfoundation.org.

FIRST encourages you to take advantage of the many opportunities and resources available through our foundation. And, we thank you wholeheartedly for helping us to provide them for you, each and every day.

Ask Dr. John

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

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

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

Question:

"I'm curious, do people with ichthyosis have trouble absorbing vitamins or problems digesting food properly?"

Answer:

For most types of ichthyosis, affected individuals have a higher metabolic rate and need far more calories than the average person. This is due to increased synthesis of skin as well as increased loss of heat from the skin. Loss of heat (from being red) also leads to loss of water and this can lead to constipation. Therefore, it is very important for those with ichthyosis to consume extra calories and fluids. It is important that your child is followed regularly by a pediatric dermatologist or other physician who has a thorough understanding of ichthyosis and can evaluate his digestive issues further if needed.

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

It's not too early to start planning for the 2016 National Family Conference-San Diego!

San Diego Marriott Mission Valley
8757 Rio San Diego Drive
San Diego, CA 92108 619.692.3800

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

Participants needed for Itch Study at Temple University

School of Medicine
TEMPLE UNIVERSITY®

Research Study: Phenotyping itch in atopic eczema, psoriasis, and ichthyosis

Temple University Hospital, Philadelphia Pennsylvania, sponsored by the Leo Foundation.

About the Study: The purpose of this research study is to examine the genetic differences involved with itch in atopic eczema, psoriasis, and ichthyosis patients. Participants with the symptom of itch and without the symptom of itch are both needed. Participants must be willing to undergo a blood draw and two skin biopsies.

Status: Currently recruiting patients until July 2015.

TO BE PART OF THIS RESEARCH STUDY YOU MUST:

- Be 18-65 years old
- Have psoriasis, atopic eczema, or ichthyosis (must have genetic diagnosis)
- Have intense itch for more than two months or no itch
- Be willing to undergo a blood draw and two skin biopsies
- Be able to speak English
- NOT be pregnant or nursing
- NOT be involved in another research study

Please contact Temple Dermatology at 267.838.1094 or at TempleDermatology@gmail.com.

How Will You Celebrate Rare?

World Rare Disease Day is February 28, 2015

World Rare Disease Day is an annual observance held on the last day of February to raise awareness for rare diseases and improve access to treatments and medical representation for individuals with rare and genetic diseases and their families. This year it will be held on Saturday, February 28, 2015. On this day, various activities take place in the United States, Europe, Russia, Japan and around the world to raise awareness for rare and genetic diseases, and to raise donations for the foundations that support and serve these communities. Many individuals plan special events on February 28th, large and small, to support the cause and show they care. For more information on how to get involved in world Rare Disease Day go to firstskinfoundation.org.

RAISE. Awareness. Funds. Hope.

One hundred. That's how many committed members FIRST needs to **RAISE.** *the bar!* **RAISE.**, *is our new national fundraising campaign, enlisting the support of members around the country to work together and raise awareness, funding and hope for all those affected with ichthyosis. Will you join us in our efforts to…***RAISE.**? *Here's how it works…*

Request: Upon request, FIRST will provide each **RAISE.** participant with a link to an online donation page and step-by-step instructions on how to easily customize content. You can keep your message simple and straightforward by sharing personal stories, photos and videos about your life with ichthyosis, or a related skin type, and your connection to FIRST. Or, if you are more inclined to plan a fundraising activity, like a walk, car wash or any type of 'athon,' you can feature your event on your donation page too!

Hi,
I'm Jean, the Executive Director of FIRST. This organization is so important to me because of the support programs and research that is taking place to help those affected by ichthyosis. Won't you please consider supporting the important work of FIRST with a

Action: Once your donation page is ready, take action and tell the world! We'll provide you with messaging suggestions for initial emails and social media posts, so you can quickly and easily send your personalized donation page to family, friends, neighbors and beyond.

Information: Keep your family, friends, and neighbors informed about ichthyosis and your progress toward reaching your goal, all the way up to the deadline of July 3, 2015. We'll provide you with the wording for follow up emails and social media posts, so you can simply cut, paste and share!

See results: You'll see results in real time. There's a built-in fundraising measuring tool on your donation page, so as your donations go up, so does the fundraising thermometer! This will inspire all your family, friends, and neighbors to help you reach, and even exceed, your goal!

Exceed: Help FIRST to exceed expectations and make this the best National Fundraising Campaign ever. **Why are we looking for one hundred participants?** It's a simple mathematical equation: 100 x $1,000.00 = $100,000.00 = exceeding expectations!

Don't wait! The **RAISE.** campaign needs your to join us now and through July 3, 2015. With a little luck, and one hundred committed participants, we'll be announcing that we've raised $100,000…or more!

Contact Chris Wassel at cwassel@firstskinfoundation.org, or call 215.997.9400 to request your **RAISE.** donation page link and start raising funds today!

Ready. Set. **RAISE.** #RAISEFIRST

Why Donate to FIRST?

Donating to FIRST will not only ensure the continuation of the Research Grant Program, whereby the top investigators in the field of ichthyosis will be supported in their pursuits, but your donations also help FIRST to connect with and embrace those living with ichthyosis today. Through specials programs, scholarships, meetings, and ongoing support from the FIRST staff, your donations help the ichthyosis community to live rich, fulfilling, and healthy lives. Learn more on pg 11 Why Donate to FIRST.

Diya & Aliya's Friends (DAF)

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 jpickford@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, 2015

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.

Register for 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 one-day forums are filled with medical discussion, research updates, networking, product sharing and skin care routines. The forum includes a light breakfast and lunch. Registration information and cost will be posted on FIRST website and Facebook page.

Dates & Locations for 2015:

Toronto, Canada – Saturday, April 18

Minneapolis, Minnesota – Saturday, April 25

Phoenix, Arizona – Saturday, June 6

Miami, Florida – TBD

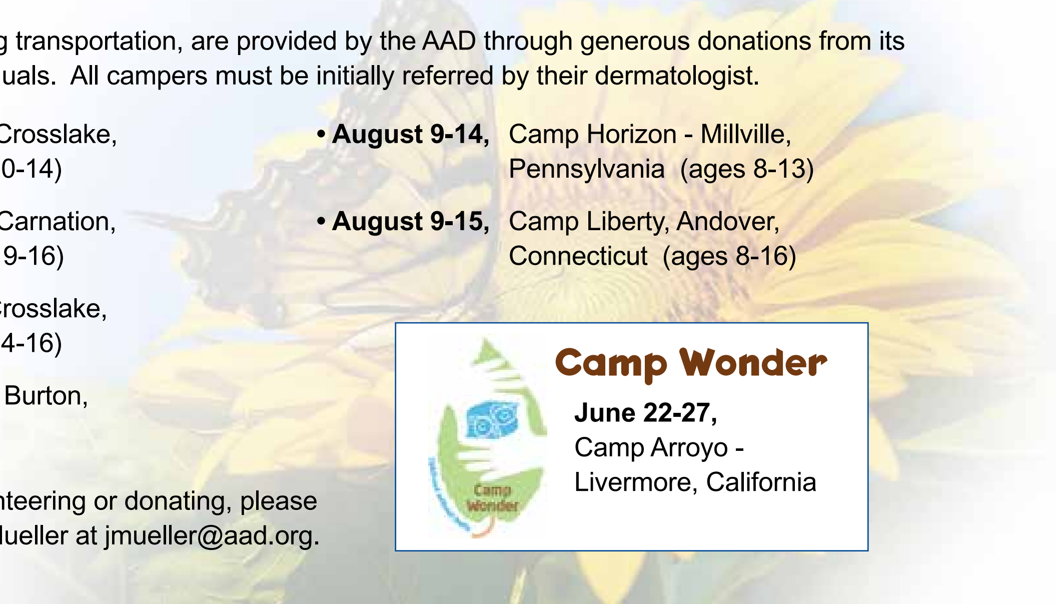
For details, contact Moureen Wenik at mwenik@firstskinfoundation.org.

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

Are You Seeing Our Posts?

*Be sure and check ✓**Get Notifications** when you Like us on Facebook at https://www.facebook.com/FoundationforIchthyosis.*
Follow us on Twitter @FIRST_Skin

Camp Discovery 2015 Dates!



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.

- **June 21-26,** Camp Little Pine - Crosslake, Minnesota (ages 10-14)
- **June 22-26,** Camp Reflection - Carnation, Washington (ages 9-16)
- **July 5-10,** Camp Big Trout - Crosslake, Minnesota (ages 14-16)
- **August 9-14,** Camp Dermadillo - Burton, Texas (ages 9-15)
- **August 9-14,** Camp Horizon - Millville, Pennsylvania (ages 8-13)
- **August 9-15,** Camp Liberty, Andover, Connecticut (ages 8-16)

Camp Wonder

June 22-27,
Camp Arroyo -
Livermore, California

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