

MAKING THE GRADE

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Thank You to Our Volunteers!

In honor of National Volunteer Week (April 18 – 24), I would like to send out a heartfelt note of appreciation to all who made the organization such an important part of so many lives over the past year. Volunteers take many shapes at FIRST, especially during a pandemic. From simple but impactful contacts to those in need, to sharing your story and time with others just beginning their journey. You give FIRST the sense of community even when we can't be together in person.

My thanks include our Board of Directors for the hundreds of hours they dedicate to keeping the mission of the organization moving forward and the diligence they demonstrate in ensuring that FIRST continues to operate in a way we all can be proud.

Thank you also to volunteers who stepped up to present on virtual Zoom or Facebook Live events. Sharing your knowledge and experiences is invaluable to those new to the community. Also, those that contributed to the content and design of our newsletters, electronic and print.

We owe much to our Medical Scientific Advisory Board for always being available to the staff to answer member questions, consulting with other dermatologists less familiar with ichthyosis and keeping our website information fresh and relevant.



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FIRST exists to improve lives and seek cures for those affected with ichthyosis or a related skin type.

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The Foundation for Ichthyosis &
Related Skin Types, Inc.
PO Box 1067, Lansdale, PA 19446

Phone: 215.997.9400

Toll-free: 800.545.3286

Fax: 215.997.9403

Email: info@firstskinfoundation.org

Web: www.firstskinfoundation.org

CEO

Chris Boynton

Editor

Kimberly Cole

Medical Editor

Amy Paller, MD

Editorial Assistant

Bailey Pretak

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“

Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has.

– Margaret Mead

”

Volunteers are the beating heart of any patient advocacy organization, but especially for the smaller ones like FIRST. With limited resources and staff, volunteers make many of the programs and services possible. Please consider joining the volunteer team. We have many exciting initiatives in early planning stages and need your guidance and expertise to make them come to life. We need people with marketing, project planning, sales, communication, and design skills along

with anyone else willing to roll up their sleeves and pitch in.

You decide how much time you can give, and we will customize an opportunity for you. Check out the “Get Involved” tab at the top of our website (www.firstskinfoundation.org) to see if there is something that interests you or reach out to our staff. A little of your time can make a big difference at FIRST!

Thank you, again.

Chris

FIRST HAS A NEW ADDRESS!

Foundation for Ichthyosis & Related Skin Types, Inc. (FIRST)
PO Box 1067
Lansdale, PA 19446-0687

Foundation for Ichthyosis & Related Skin Types

New Logo Celebrates the Power of Shared Connections

In celebration of FIRST's 40th anniversary, we have updated our logo to reflect the power of the shared connections we have in our community.

first

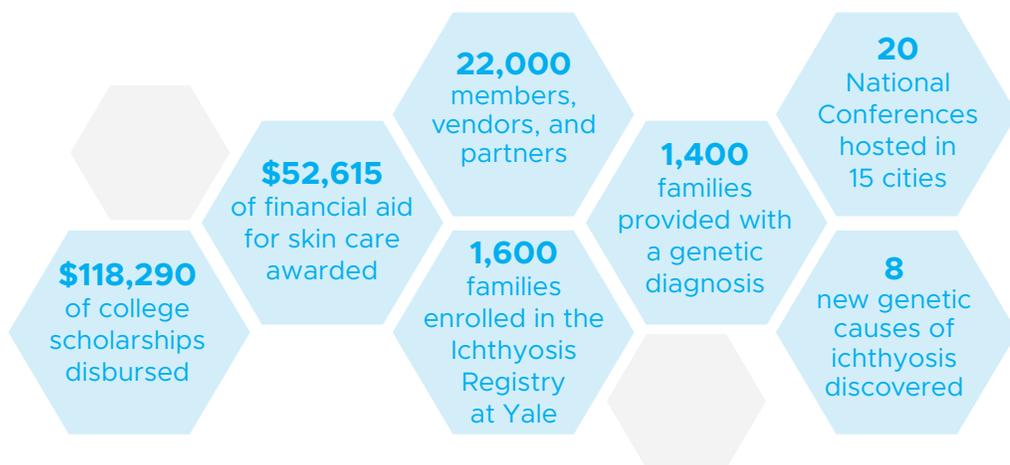
Foundation for Ichthyosis & Related Skin Types

With 40 years of community-building to advance treatments and research of ichthyosis and related skin types, we have much to celebrate. At FIRST, each member is surrounded and supported by our community. From advice between parents, guidance from our research leaders, to welcoming new friends to FIRST, our community is resilient

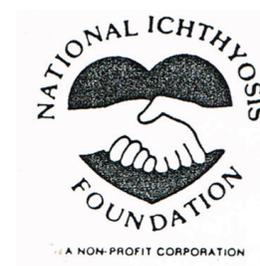
and purposeful. One is never alone.

Making connections and supporting individuals with ichthyosis and related skin types is at the core of everything we do. We are proud to celebrate this important milestone with you.

We are proud to celebrate 40 years of shared connections, including:



1981 - 1986



1987 - 2009



2010 - 2020



National Conference Postponed to 2022

In response to the ongoing COVID-19 pandemic, it was necessary to postpone the FIRST National Conference twice. It is now scheduled for **June 24 – June 26, 2022**, and the location remains the same in **Providence, Rhode Island**.

Christopher Boynton, FIRST's CEO, summarized his views this way in an announcement to members:

"If you had asked me earlier this year, I would have said we are 'all systems go' for a smaller conference with some virtual components for June 2021. However, guidance from several sources have me changing course, even considering positive vaccination news. It has always been important

to have a conference where 'being in a room with others who look like me' and the therapeutic nature of the bonding and camaraderie is the priority. FIRST will continue to create and execute virtual events in 2021 and, if the environment is safe later in the year, some in-person Patient Support Forums."

Stay tuned for further announcements (locations and dates) about FIRST's one-day Patient Support Forums this fall. We hope to see all of you at the National Conference in Providence in June 2022. It has been too long without in-person meetings and we miss you. Next year, we will have all the familiar aspects of the conference and some new aspects, too. You will have

the chance to see your FIRST friends, enjoy social interactions, meet experts in ichthyosis, participate in research studies and so much more. You can be assured that it will be the best National Conference yet!



#FIRSTNC2022

FIRST Retains Financial Health, Supports Members Through 2020 Challenges

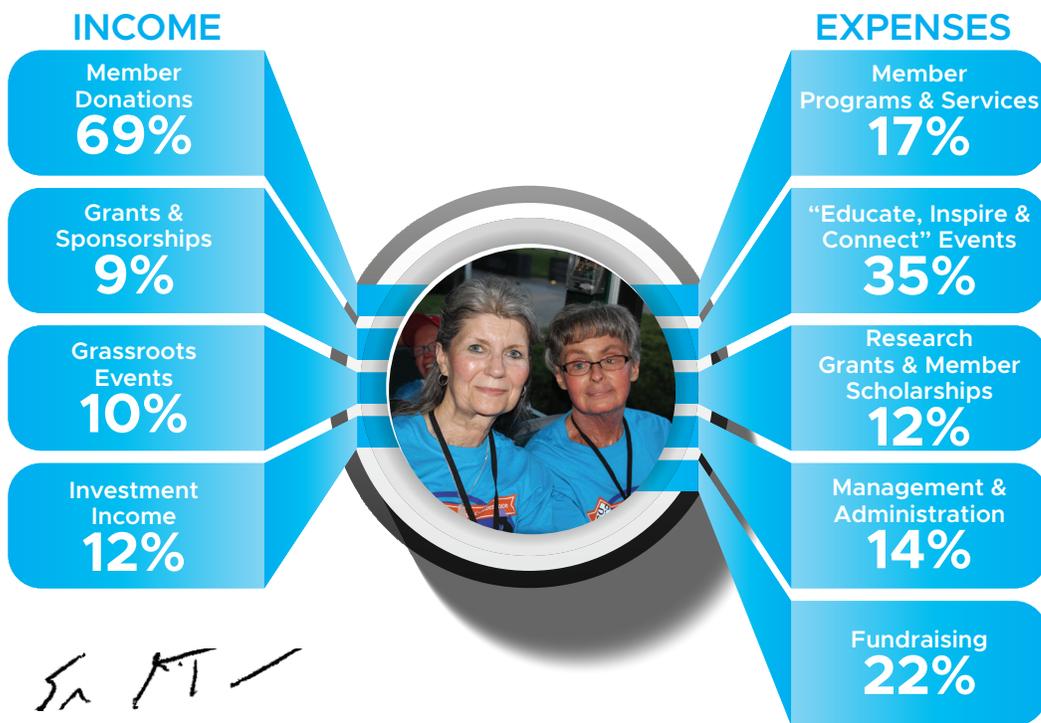
Despite the many challenges our members and organization faced in 2020, FIRST continues to hold a healthy financial position. We remain committed to using our financial resources to provide advocacy, research and specialized programs and services to our members and community.

Last year, FIRST realized its mission by:

- Funding grants to support members with skincare and higher education expenses
- Supporting the National Registry for Ichthyosis and Related Skin Disorders, which conducts genetic testing at no charge to FIRST members
- Responding to emails, phone calls and social media posts seeking information or resources

Our ability to continue serving in this capacity is due to the generosity of our donors and dedication and hard work from our volunteers, staff, Board and Emeriti.

I am pleased to share this high-level summary of our annual income and expenses.

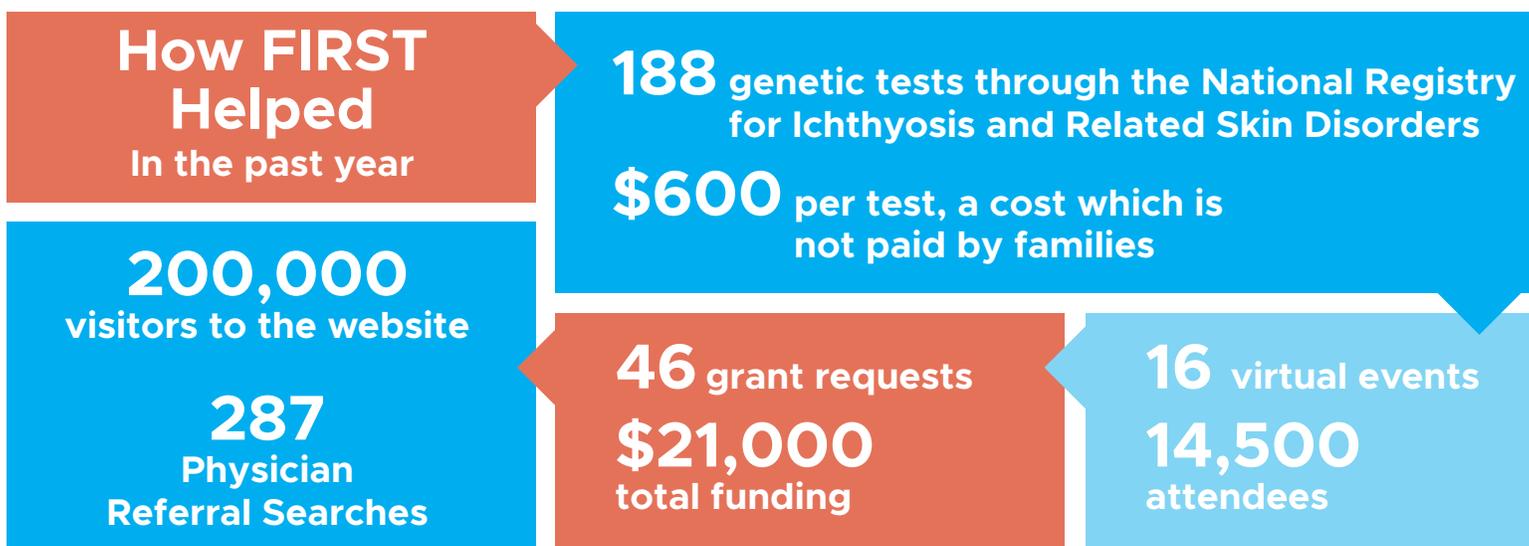


Sean McTernan is FIRST's Chief Financial Officer and leads the Finance Committee. McTernan and his wife Carolyn, have three children, Avery (9), Henry (7) and Claire (5) who has temperature-sensitive lamellar ichthyosis.

"FIRST has played a special role in our lives, particularly so in the early days of our youngest daughter's life, when few others could provide answers or perspective regarding ichthyosis," he said. "We are sincerely appreciative of the role FIRST plays in supporting and advocating for those in the ichthyosis community and are happy to give back to the organization which has done so much for our family."

McTernan works for Brown Advisory where he is a Principal and Regional Investment Consultant. He lives with his family in the Philadelphia suburbs.


Sean McTernan, Finance Committee



May Is Ichthyosis Awareness Month!

Below are some ideas for getting involved. Visit our website for more info and tools!

- **Share stories:** whether your own or others', through social media or in person, sharing the narratives of life with ichthyosis is an important way of owning your story and educating others.
- **Raise funds:** Facebook makes it easier than ever to host a fundraiser. Or, consider setting your Amazon default website to smile.amazon.com where a portion of every sale will go to FIRST.
- **Raise awareness:** Contact local media outlets and let them know they have someone rare right in their midst. We have a draft press release on our website for your use!
- **Volunteer for FIRST:** Get involved by checking out our latest volunteer listings on our website.
- **Advocate for the cause:** Write to your members of congress and advocate for better access to care, treatment, insurance coverage, and research for ichthyosis. For a sample letter, visit: www.firstskinfoundation.org/sample-letter-to-congress
- **Celebrate the ichthyosis in your life:** Try a dose of self-care—consider a special skin care treatment or new product. We'd love to hear how it goes!

New Member Spotlight



A special welcome to new member Merle Ways, age 77, of Sicklerville, New Jersey. Merle recently joined FIRST and said, "Looking back, there are some years I truly don't know how I made it alone with ichthyosis." However, as she grew up, Merle was blessed with many family members who shared her ichthyosis vulgaris / PPK, including three affected children: Melinda, Martha, and Raymond. She also has two affected grandchildren, Rashon and Lakesha. FIRST is delighted to meet all of the Ways family members!

START A
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DURING IAM



UFIRST Program Adds Up to Success for Math Teacher

Submitted by Bailey Pretak

Ryan Balog tries to be a teacher that is student-friendly and fun to be around. He implements as many hands-on or virtual activities as possible to make math come alive for his middle school students. “As much as I love teaching the kids math,” he said, “I really feel my job is to impact these kids on a daily basis and to make them better people.”

Balog is one of 11 UFIRST Scholars awarded in 2020. The program has helped him pay some costs of his education. He has received the award for five years, supporting both his undergraduate and graduate studies. “It has meant a lot to me,” he said. “I know UFIRST gets many outstanding applications every year, and it’s very humbling to get chosen.”

Balog earned his bachelor’s degree in middle level math from LaRoche University in Pittsburgh and is currently in the process of obtaining his master’s in instructional leadership from Robert Morris University. He is not sure of the exact point when he knew he wanted to be a teacher, but he always excelled in math and enjoyed working with middle school kids, so it made sense to go into teaching.

“

I want each (of my students) to follow their dreams and find their passion.

– Ryan Balog

”

Today, Balog teaches 6th Grade Math at Pittsburgh Langley Public School. His favorite thing about teaching is making connections with the kids, especially since many of them face a lot of challenges outside of school. His personal goal is to build trust to not only help them succeed in math, but in the real world.

When students dislike math, he likes the challenge of trying to get them comfortable, so they do not fear it. There are countless career fields needing math, so he works to set a strong foundation of solving problems



of any sort. “As a teacher, my goal is to set my kids up for success in the future,” Balog said. “I want each of them to follow their dreams and find their passion. These kids are the future, and I want that future to be brighter than today.”

One of his most memorable moments this (virtual) school year was singing during class breaks. He gets out his karaoke microphone and starts singing. “I believe some of the kids feel bad for me and start singing along,” he said. “The kids always get a good laugh when I embarrass myself. It keeps the vibe positive.”

Balog encourages other FIRST members heading off to college to apply for the UFIRST Scholarship. He is

grateful for the scholarship but is quick to point out that FIRST has helped him in many ways throughout his life: providing opportunities to meet others, gaining lasting friendships, learning suggestions to care for his condition, and inspiration from countless people he has met through the years. These experiences have given him the confidence to be himself and not be shy with his disorder, which he believes led him to where he is today.

Learn more about the UFIRST Scholars program, and how to contribute to its continued success at firstskinfoundation.org/ufirst.

G16 Skin Repair (recommended for Vulgaris, El, X-Linked, Bullosa and Lamellar types) has been helping people with different dry skin conditions including several types of ichthyosis for many years now. It is changing the lives of many thousands of people all over the globe and is fast becoming the an effective dry skin treatment. G16 Skin Repair has been working with FIRST for several years and many of their members have reaped the benefits of the lotion. It is designed to loosen and remove dead skin cells that become attached to the outer layer of skin. It then manipulates the skin into a regular skin shedding cycle. It stimulates new healthy skin cell growth and deeply hydrates and nourishes the new skin. Here are some stories from G16 customers of how this amazing lotion has changed their lives.

Jessica Kurti's Story – 21 from London, England – Born with Ichthyosis – Using G16 Skin Repair for one month

Life was horrible. Ever since a kid I had to moisturise four plus times a day. I tried everything and nothing worked. I managed to get this amazing body but never showed it off because of my skin. Never wearing crop tops or shorts unless I had a tan, looking at my friends getting changed at the gym who had normal skin and my skin looked like a fish peeling. I always dismissed swimming, as even my legs were so bad. It has made me lose so much in life.

I have tried many products and been to over 25 dermatology hospitals and met lots of different doctors. The products I would use would be effective in keeping my body soft but would not keep the skin from peeling. It would go back to how it was within the hour of having used the cream. My mother was searching on the internet one day and randomly G16 Skin Repair showed up. She looked at the website, saw all the testimonials from other individuals suffering from this condition and ordered it for me. When I looked at their website, I had never come across before-and-after pictures quite like it. This made me actually believe that this would be life changing for me - and it was!

I saw results on the third day and I was shocked. I ran to my mother and just cried, and she did too. I had never seen my skin like this, it had never felt like this! It was amazing!

The best thing for me was that it brought my whole body confidence back! I am no longer shy to sleep round at friends'

houses wondering if they would feel uncomfortable with my skin shedding and making a mess. I no longer worry that I can't wear short tops because people outside would stare; some people would even ask what was wrong with my skin and I would have to explain myself. Now I don't have to do anything like this! I'm free!

I have been using G16 for a month now. I would recommend G16 Skin Repair so much if you want your life to change. It is worth every penny to save your skin. You won't be let down, and one day you could feel just like me, so happy.

G16[®]

Skin Repair



Denice Holliday's Story - 60 from Georgia, USA – Born with Congenital Ichthyosis Erythroderma (CIE) – Using G16 Skin Repair for two years

I came across G16 doing an internet search on lotions for dry skin. I was super skeptical as I know there is no cure for my skin condition but thought I would give it a try. I am always searching for something great to work on my skin. G16 works extremely well for me. My skin is not 'healed' but does look close to 'normal'.

Before I found G16 my life was a constant struggle with worries and insecurities about my skin. I still do have those same feelings but not as much. I have tried lots of products over the years, a few products have worked well but once I try them for a week, (even after a day or so) I can tell they are very 'weak' and do not have any lasting power/effect. I saw an advert for G16 Skin Repair and wanted to give it a try. I am glad I did.

In two days, my skin was much easier to exfoliate. Within a month, I saw some 'anti-aging' effects! After a year or so of use, I had a yearly check up with my dermatologist. She mentioned how great

my skin looked. I told her I believed it was this new lotion I had found, G16 Skin Repair. She had never heard of it. Due to my condition being genetic I have to apply this lotion every day. I go through a bottle every 2 weeks. I can use it more sparingly, a little goes a long way, but I still have to use it every day.

The best thing is that I am happy I found a product that works well for me! The difference it has made in my life is that it has given me a boost in confidence in dealing with the appearance of my skin. I would recommend G16 Skin Repair. This lotion is a MUST TRY for any one with certain types of ichthyosis.

Joe Goodwin's Story – 67 from Western Massachusetts, USA – X-Linked Ichthyosis - Using G16 Skin Repair one year

I first came across G16 Skin Repair in a FIRST newsletter via email. Before I started using G16 Skin Repair I had a lot of discomfort with itchy, dry, flaky skin. A lot of self-consciousness and embarrassment and I made a strong effort to conceal my skin condition. I was unable to play sports in my school years because I didn't want to expose my skin in the locker room. Dating was difficult and social gatherings around pools and the beach were troubling.

Prior to finding G16 Skin Repair I had tried everything I could get my hands on! The testimonials that I read on the G16 website and advertisements were the reasons I decided to give their lotion a try but mostly because it was a product I had not tried before.

When I started using G16 it took about a week to 10 days and then I started seeing results. It is easy to apply and a small amount goes further than most lotions. I now apply the lotion about every two days but when the weather is warm and humidity up, I only use it on my legs where it is most severe. In winter months, I use it every other day, all over my body.

The best thing about G16 for me was that it removed the scales and softened my skin to the point that I could wear shorts and short sleeve shirts in public with confidence, and not worry about leaving skin flakes behind me at the gym and elsewhere. I would absolutely recommend G16 Skin Repair to others with this skin condition. I would urge young people with ichthyosis to use it in order to have a normal life. Use this product as instructed and you will see amazing, life-changing results in a short time! It is so worth the money!

UFIRST Scholar Recipients Share Aspirations



The UFIRST Scholars program awards scholarships to post-secondary students. Congratulations to the 2020 UFIRST Scholars!



NASEER AHMED

ARCI-lamellar ichthyosis
University of Kotli Azad Jammu, Pakistan

"The highest calling for me is to serve the greater good of humanity and the world."



RYAN BALOG

ARCI-CIE
Robert Morris University, Pennsylvania

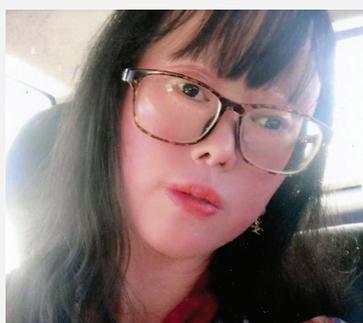
"I want to influence students and be a role model for many. I want to show my future students that nothing can hold you back."



KAYLEE BROWN

Netherton syndrome
Bradley University, Illinois

"I want to go into the medical field to become a pediatric dermatologist. I want to be able to help kids with skin issues like me."



TSHERING CHODEN

ARCI-lamellar ichthyosis
Royal Thimpu College, Bhutan

"My goal is to obtain a bachelor's degree, continue further studies and obtain a master's, go into child counseling and help in the future."



CHRISTINA DASILVA

Palmoplantar keratoderma
Purdue University, Indiana

"I aspire to become certified as a registered dietitian and work in the competitive field of athletics, promoting optimal performance through proper nutrition."



WILLIAM EGAN

Ichthyosis hystrix
Louisiana State University

"My career goals are to have a good paying job in a place where I make a difference or even work to be awarded patents for unique ideas."



EMELIA GREGOR

ARCI-CIE
University of Redlands, California

"My goals are to get my bachelor's degree in liberal studies and Spanish and my master's in education so that I may become a high school Spanish teacher."



KATHARINE HAMEL

unknown
University of New Hampshire

"I aspire to become a nurse or work in healthcare. I want to provide support and help to those in need."



RYLEE IOTT

ARCI-CIE
University of Sioux Falls, South Dakota

"My goals to earn a bachelor's degree with a double major in accounting and business administration. Following that, I plan to work towards my master's."



ADAM KLAFTER

Epidermolytic ichthyosis
Savannah College of Art & Design, Georgia

"My goal is to be more independent and become successful after graduating college"



ALEX YODER

Ichthyosis vulgaris
Indiana Wesleyan University

"I want to get a college degree and teach children in a public school setting. I want to have a part in raising and inspiring the next generation."

Scalp Care Tips from a Cosmetologist

Submitted by Cora Cossel

Being a cosmetologist in the ichthyosis community, one of the biggest questions I am asked is how to care for the scalp. I will admit, even with my training, this had me stumped for the longest time in dealing with my daughter's scalp care. We tried so many different things, some which seemed to help, and some which seemed to help others, but not so much us. I am going to share a few tips and tricks that have helped us, and some of the science behind things that should work.

“

Even with my training, [scalp care] had me stumped for the longest time.

”

First of all, we don't want to lather hair in lotion. It gets greasy enough, and it can be a battle getting ointments out of the hair. Should you ever get in a situation where you feel that you can't get the grease out, I recommend a clarifying shampoo. Some lines will call this "normalizing" or "balancing", but the purpose of all of these is deep cleaning. Paul Mitchell Shampoo Three is a good one that will cut through grime on the hair. But remember, this is clarifying, it is meant to strip things from the hair, so it may also be drying to the scalp. Use it sparingly.



Now let's address the next issue: scaling on the scalp. Think of conditioner as lotion for the scalp. One of the best things you can do is lather the scalp with conditioner, put a shower cap on, and let it sit. It will soften the scales, and allow you to comb them out easier with a fine-tooth comb. I recommend a lice comb.

Let's talk science. The pH in shampoos determine what the shampoo does for your hair. Anything with a lower pH is more acidic -- and that makes it more moisturizing. Most shampoos are not going to list the pH, so there are key words to look for. Shampoos with salicylic acid, such as T/Sal are game changers for the scalp. Nioxin is another line that focuses on creating a healthy scalp. Using these daily will help decrease the scales on the scalp without excessive drying. And the more moisturized the scaling, the more likely the scales are to slough off without forcing them off, resulting in less damage to the scalp.

You want to avoid heavy products that are not intended for the scalp. Coconut oil may make your hair and skin feel amazing, but the compound is too heavy for the scalp. It can clog pores on the scalp, hindering hair growth. It can also trap in bacteria and fungus, which we struggle with enough as it is. For all of these reasons, I do not recommend it for the scalp.

I don't know that there is an exact science to haircare and scalp care with ichthyosis; these are just some of the things I have found to work. I am always here to answer any questions you may have. Contact the FIRST office at info@firstskinfoundation.org to connect with me!

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*Baden, H.P. Management of Scaly Skin with Epilyt. Seminars in Dermatology, 6:55-57, March 1987.

To learn more, check out our recent virtual event featuring Cora Cossel and Toyia Bailey.

Watch at youtube.com/FIRSTskinfoundation.

Clinical Trials: Consider Commitment and Concerns

Submitted by Phoevos Hughes, JD, Interim Head of Medical & Clinical Affairs, Mayne Pharma

Clinical trials are an important component in the development of new pharmaceutical products. They are the primary way researchers find out if a new treatment is safe and effective in people. As such, clinical trial participants are extremely important in the development of new products and their contribution has potential to help current and future patients.

There are many reasons why people choose to join a clinical trial. It may be due to a lack of effective treatment options, or lack of treatment options altogether. Whatever the reason, here are a few things to note when determining if clinical trial participation is right for you:

1. Clinical trials are a commitment. It is important to understand all the details regarding a clinical trial you may be interested in participating in. Details such as visit frequency and duration, treatment dosing, restricted activities or medications, and other factors are important to understand. These details should be considered in light of how study participation will affect your day-to-day life.

2. Ask lots of questions. Whether you are an active participant in a clinical trial or considering participation, you should always ask the study doctor or staff as many

questions as necessary to ensure you are comfortable with participating (or continuing to participate) in a trial. Making an informed decision will help alleviate any fear or anxiety you may have, regardless of your decision.

3. Vocalize concerns and provide feedback. If there is something you find concerning about the trial, openly discuss it with the study doctor. An alternate solution may be available to address a concern, and you will never know unless you ask. If a concern still remains, it is OK to choose not to participate in the trial. Although clinical trials cannot easily be modified, trial designs are commonly updated to account for, among other reasons, participant feedback.

4. You can stop participating at any point. All clinical trials require your informed written consent before you are allowed to participate in the trial. However, this is not the only time you can choose to not participate. You may withdraw your consent at any point for any reason. If you wish to stop a trial you are participating in, you should inform your study doctor in order to determine the safest way to do so.

If you are interested in learning more about participating in clinical trials, talk to your doctor or other healthcare provider, or contact the FIRST office.

Mother Shares Story with X-linked Ichthyosis

My name is Jolien van der Geugten and I live in the Netherlands. My son is 5 years old and has X-linked ichthyosis.

When my son was only nine days old, he got strange spots in his face. They looked like wet yellow crusts. The next morning it was worse. I called the midwife and she advised us to go to the doctor's office. The general practitioner sent us directly to the emergency room.

At the emergency room, the pediatrician thought it was serious: an infection in his face. The dermatologist rushed to the hospital. They thought of impetigo. My son was admitted and got an infusion of antibiotics. He could barely open his eyes because of all the scabs and skin.

I was shocked, sad and angry. Why was I reassured that it was baby acne? How did he get the infection? Could we have prevented it?

After a few days we got answers. The dermatologist thought he had X-linked ichthyosis. That explained the dry, flaky skin he had just after birth, but was not recognized by anyone as ichthyosis.

With ichthyosis, bacteria can more easily cause a serious infection. The advice was to apply fatty ointment at least twice a day.

And that is what we did. But my son experienced a lot of itching and red scratched open skin. He cried at night and was inconsolable. I got in touch with the patient association in the Netherlands. Through the association I got to Prof. Dr. Suzanne Pasmans at the Erasmus Medical Center in The Netherlands. She determined that my son also had eczema. With several ointments we got the eczema under control, and his skin became more stable.

Several times I have felt guilty. Did I give my son the best care? What if I had lubricated him with ointment every day immediately after birth? As a parent, I wanted the best for my child. But with rare skin conditions like ichthyosis, you don't always know what's best. I got a lot of strength from FIRST's Facebook groups, the Dutch patient association and FIRST. The recognition and advice I received were amazing.



My son is now 5 years old. He entered primary school and I realized there is no book to explain about his condition to his new friends. That's why I wrote an adventurous children's book about a boy with ichthyosis for children from about 3 to 6 years old. In May 2021 the book will be published in the Netherlands. There are plans to release the book in English as well at the end of 2021.

Research Provides New Insight on PPK, Harlequin and Growth

Mutations in *ASPRV1* Cause Dominantly Inherited Ichthyosis

American Journal of Human Genetics; original article, July 2, 2020; Boyden LM, Zhou J, Hu R *et al.*

Review: Lamellar ichthyosis featuring palmoplantar keratoderma (PPK) had exclusively been attributed to autosomal recessive mutations prior to this discovery. Investigators sequenced the coding regions of all the genes in the genome to reveal that in four unrelated ichthyosis kindreds, ten subjects with ichthyosis and PPK had heterozygous, novel missense mutations in *ASPRV1* causing autosomal dominant ichthyosis. The mutations arose in utero in two individuals, and mutations in the other eight subjects were transmitted from one generation to the next, indicating that *ASPRV1* mutations cause a dominantly inherited ichthyosis that was previously known to have recessive inheritance. *ASPRV1* encodes skin aspartic protease, an enzyme that breaks down the filaggrin protein. Compared to age-matched controls, those with *ASPRV1* mutations have excess unprocessed filaggrin protein and epidermal differentiation impairment resulting in thick scale and PPK. Given this desquamation defect, keratolytic agents can prove therapeutically beneficial, as two affected individuals who used keratolytics experienced complete resolution of non-palmoplantar scale.

Summary: *ASPRV1* mutations cause dominantly-inherited lamellar ichthyosis with palmoplantar keratoderma, highlighting the importance of aspartic proteases (enzymes that break down filaggrin protein in the skin) in epidermal differentiation. This not only further elucidates the various causes of ichthyosis but also provides a potential target for future therapies.

Mary Sun and Dr. Keith Choate from Yale reviewed recent research published in medical journals to summarize for members and physicians. Read on for more information on developments in ichthyosis research.



Mary Sun



Dr. Keith Choate

3D Model of Harlequin Ichthyosis Reveals Inflammatory Therapeutic Targets

Journal of Clinical Investigation, original article; Aug. 10, 2020; Enjalbert F, Dewan P, Caley MP *et al.*

Review: Harlequin ichthyosis (HI) is a rare, severe ichthyosis with high rates of complications and death in the perinatal period. It is caused by mutations in the gene *ABCA12* which eliminate or severely reduce the function of its encoded protein. Skin inflammation is a well-known component of HI but few studies have investigated the inflammatory processes inherent in the disorder. The authors developed a 3D model of HI using CRISPR/Cas9 (a technique that edits the genome) to engineer an *ABCA12* knockout cell line that closely mimicked the most severe HI phenotype. Using this model and HI skin samples, the authors discovered upregulation of cytokines in the IL-1 family and upregulation of the STAT1/NOS2 signaling pathway, which results in increased accumulation of inflammatory free radicals. Inhibition of NOS2, an enzyme with a central role in inflammatory processes, reversed the barrier defects seen in the HI model. These findings not only provide insight into the pathogenesis of HI but also reveal new therapeutic targets.

Summary: Upregulation of inflammation, in particular proinflammatory cytokines, *STAT1* and *NOS2* signaling drives HI redness.

Malnutrition in Children with ichthyosis: Recommendations for Monitoring from a Multidisciplinary Clinic Experience

Journal of the American Academy of Dermatology; original article; June 2020; Rodríguez-Manchón S, Pedrón-Giner C, Cañedo-Villarroya E *et al.*

Review: Growth failure (not meeting milestones for height and weight among other measures) is common among individuals with congenital ichthyosis but there is a paucity of data evaluating nutritional status in ichthyosis. The authors assessed 50 ichthyosis patients, majority of whom were male and under 18 years of age, in a prospective study at a hospital in Spain. Nearly one-third met WHO criteria for undernutrition. Growth impairment was found in 24% of children, especially those 5 years and younger. Ichthyosis severity positively correlated with rates of undernutrition, and nearly two-thirds of patients had micronutrient deficiencies, particularly deficits in iron, selenium, vitamin D and zinc. These results provide the rationale for early nutritional assessment and support in order to maximize growth potential.

Summary: Children, especially those who are younger and those with severe ichthyosis, are at risk of malnutrition and should have nutritional assessment at diagnosis and during follow-up.

Viewpoint: Mother and Daughter Team Up to Inspire Confidence

Alyssa Loudermilk, 15, and her mother, Krista, reflect on their relationship, facing fears and building strength to overcome adversity.

Alyssa:

It is hard to grow up and be different than everyone else, to not look like everyone else, and to have people judge you for something you can't change. But what you can change is your confidence. It's not easy to be confident; people will always try to bring you down and make you feel less than, just because you are different. But being different is amazing!

I have learned to never let my skin define me and tell me I can't do something. It's hard sometimes to put those bad thoughts behind me and be confident, but being comfortable in my skin feels amazing! I know that even with my skin condition I can do anything or any sport I choose. I hope this has helped and lets you know that just because you have a skin condition doesn't mean you can't be happy. So do that sport! Wear what you wanna wear! And be confident in your skin!

Krista:

At some point acceptance is all you can have. Children grow and learn to cope. We have always had the understanding that if you want to try a sport you have to finish the season - no quitting. In T-ball we thought she was going to pass out in the field because she couldn't regulate her temperature, but we finished the season. We race four-wheelers and decided she could try racing her little four-wheeler. This started a passion to be competitive. She found her love for cheerleading and tumbling, and then track. I was scared to death of her overheating, but she was taking control and she managed on her own and competed. Currently Alyssa has won the Junior class for 3 years in a row racing her junior dragster. Her competitive cheer team will be attending Nationals again this May, like they have done for several years now. Alyssa is also on the Varsity High School track team.

I believe she takes control of her skin condition like a competition. She wants to know if she can beat it. She sets high goals for herself, and competes to complete them. Alyssa has showed us that in this cruel world where not every child is taught to not make fun of another person, and even adults don't know better to make fun of a child, that she has the right to stand up for herself. She is a strong-willed, very smart and beautiful person and she is becoming herself and not letting her skin control her. I hope she will always compete to take care of it and herself.



Alyssa Loudermilk, 15, is confident and strong posing in her cheer uniform

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Hoerle Led FIRST Board Through Years of Change, Growth

Submitted by Mark Evans and Tracie Pretak

Jeff Hoerle has inspired us with his multi-talented, multi-faceted and unwavering dedication to FIRST's members, staff, Board, and Medical & Scientific Advisory Board over so many years. His ability and willingness to operate at both a very high strategic level, as well as rolling up his sleeves to get even the smallest task done, is truly remarkable. Jeff has always been willing to fill any void necessary at FIRST.

He has left FIRST well-positioned to move forward and serve our community from both a staffing standpoint and in an enviable financial position. Although Jeff has completed his term as Board President and we wish him all the best, we also know that he and his family are still a strong and vital part of the ichthyosis community.

We have both truly enjoyed getting to know Jeff while working with him on FIRST's Board and Committees. We can't thank Jeff enough for his leadership, mentorship and steady hand at the wheel of FIRST over so many years.

Evans and Pretak are the co-chairs of the FIRST Board of Directors



Jeff Hoerle served as Chair of the FIRST Board of directors from 2015 - 2020.