



ALBUQUERQUE AWAITS

*Find connection, information at
National Conference in New Mexico....7*

UFIRST Scholars.....5

*Remembering
Patti Brennan....13*

Research reviews15

Dear FIRST Members, Families and Friends,

2024 is going to be an exciting year for FIRST. I can hardly wait to see everyone at the National Conference in Albuquerque, New Mexico, this June. The FIRST staff and volunteers are working hard to put together an amazing conference. I can tell you that attending the conference can be life-changing in so many good ways. I want to encourage all that can to attend. FIRST is proud to partner with Beiersdorf this year to provide financial aid to many of our members to assist them in attending the conference. Along with the conference, FIRST staff and board members are working hard to update and complete our organization's strategic plan. This document will help provide a roadmap for the our future. We also welcomed two new board members this year. Hopefully, everyone will have a chance to meet them at the conference this summer.



Beth Hampshire

FIRST has other financial aid opportunities. The UFIRST Scholars program, also supported by Beiersdorf, provides scholarship funds to affected students who want to advance their post-secondary, graduate, or vocational education. Since the program's establishment FIRST has provided 127 awards to 72 students! Also available are the Diya & Aliya's Friends Skin Care Fund and the FIRST-Aid Skin Care Fund. These funds provide opportunities for members to apply for assistance to purchase lotions, medication and treatments resources. Visit the FIRST website for more information and details on applying.

Programs like these are important to FIRST and our members. These programs were started by – and continue to be supported by – our members' fundraising. Fundraising is a great way to help FIRST with its mission to support people affected by ichthyosis and raise awareness of ichthyosis in your own community. FIRST has resources on our website to help anyone interested in raising funds for FIRST. Fundraising can be as easy as an online birthday fundraiser (see Brenna's story on page 4). Or, if you are ambitious, try something like a wine tasting event. Whatever format you choose, Ichthyosis Awareness Month in May can be the perfect time to try. Please share your results with us so we can thank you.

I look forward to seeing everyone this summer at the National Conference!

My warmest regards to you.

A handwritten signature in black ink that reads "Beth Hampshire".

Beth Hampshire
Chairperson of the Board of Directors

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

Copyright © 2024
by the Foundation for Ichthyosis & Related Skin Types, Inc.

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

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

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 Assistants

Bailey Pretak
Abby Evans

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

The logo for the Foundation for Ichthyosis & Related Skin Types, featuring the word "first" in a bold, lowercase, blue sans-serif font. A registered trademark symbol (®) is located at the top right of the letter "t".

Foundation for Ichthyosis & Related Skin Types

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

Ichthyosis awareness and advocacy at AAD Annual Meeting

FIRST enjoyed a unique presence at the annual meeting of the American Academy of Dermatology in March. Members of FIRST's Medical and Scientific Advisory Board hosted the conference's first-ever ichthyosis general session, while FIRST staff and member Hunter Steinitz promoted FIRST's mission to physician attendees, pharmaceutical companies and potential partners.

General session sparks interest, awareness

Dr. Sarah Asch, MSAB President, and Dr. Cheryl Bayart, MSAB member, hosted a general session open to the conference's 18,000+ dermatological professionals. This year's general session marked an exciting expansion, as FIRST's MSAB often gathers at AAD for a closed-door session on ichthyosis cases. The goal was to increase awareness of ichthyoses and hopefully get one or two dermatologists started down the path to becoming the next generation of dermatologists with a deep abiding interest in ichthyosis.

The session, called Think Like an Ichthyosis Expert, featured:

- Sample cases presented by MSAB members Dr. Evelyn Lilly, Dr. Chris Bunick, and Dr. Jason Meyer. Audience members participated using a live survey tool, submitting their input on diagnoses and treatments.
- The patient experience, shared by FIRST Director of Development Denise Gass, who has ARCI-CIE ichthyosis. Gass drew on her own experiences, with anecdotes from other FIRST members. This was an engaging and unique opportunity as patients rarely appear and speak at the AAD meeting.
- Audience discussion of cases presented by Dr. Jennifer Hand and Dr. Keith Choate, MSAB members.

This format was a departure from scientific meeting sessions in the past, which are typically straight lecture.



FIRST member Hunter Steinitz

Each case received comments from various experts. The discussion was supportive, informed, and friendly.

The audience consisted of experienced doctors, pediatric dermatologists, general dermatologists, and resident physicians and medical students.

"We would like to get more doctors open and less intimidated by caring for patients who have a form of ichthyosis," said Asch. "The overall message was that if general dermatologists are curious, willing to reach out to FIRST for help, and willing listen to their patients and families ...they can be very successful at caring for a person with a rare condition."

Building relationships across industries

The AAD reported that 19,867 dermatologists and healthcare professionals attended its annual

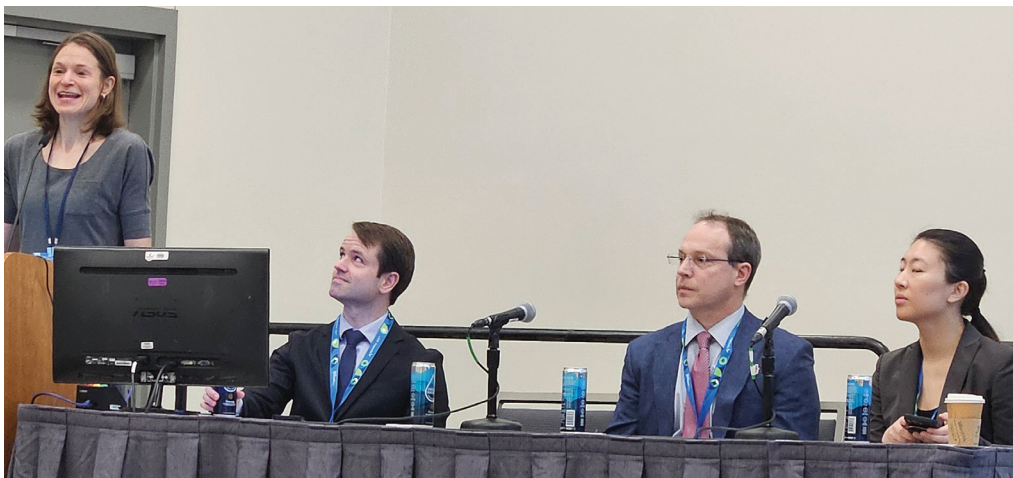
meeting, making it likely the largest-ever dermatology meeting.

Meetings with pharmaceutical partners confirmed deep commitments to clinical trials and seeking FDA-approved treatments for ichthyosis. LEO Pharma, which recently acquired Timber Pharmaceuticals, presented FIRST with a check for \$25,000 and a promising update on their Phase 3 clinical trial for ARCI ichthyosis.

FIRST also met with other patient support organizations for rare diseases and skin diseases. Themes included health equity, growth, and increasing impact for patients.

The meeting provided a chance for FIRST to meet with current and prospective industry partners. Longtime FIRST partner Beiersdorf, the makers of Eucerin and Aquaphor, featured a prominent FIRST banner at their booth, and hosted interviews with FIRST CEO Chris Boynton and FIRST member Hunter Steinitz.

FIRST visited with brands ranging from the well-known to the up-and-coming, looking for products that might help in managing ichthyosis. Advancements in skincare include better SPF options for all skin tones and preferences. One promising trend in the exhibit hall was innovating skincare application. In addition to the de facto tubs and tubes, brands are including sticks, sprays, roll-ons, and gels. These delivery options can be particularly useful for children, itch, layering product, and convenience.



Ichthyosis general session

Members find unique ways to support FIRST

Birthday fundraiser a success for Brenna

Submitted by Brenna Westlake

For my 12th birthday on December 19th, I decided to raise money for FIRST to help people with skin like mine. I wanted to help FIRST because it teaches me about my condition and connects me to friends, especially at the national conference. I thought of the idea because of my business called Kittysitting By Brenna, which allows me to earn money by taking care of other people's cats. When I get paid, I set aside 10 percent to donate.

For a while, I saved up this "giving money" and then kicked off my fundraiser with \$50 of my own money.

On my birthday, I created an online fundraiser on Facebook and asked people to give \$12 because I was turning 12. I also described what FIRST was, so people donating could know what they were supporting. When the final amount was collected, my parents matched the total.

When the fundraiser was done, I had earned over \$4,500! That was WAY more than my original goal of \$500. I was so surprised when it just kept going up and up! I really hope this money will help families in need of support. I was so happy to see how much I had raised.



Celebrating grassroots heroes



Dominique Romer - Stamford, Connecticut

FIRST member Dominique Romer ran the SoNo Half Marathon in Norwalk, Connecticut, in October. She raised over \$13,000 in support of FIRST and to celebrate her 3-year-old son, James!



Tejal Kamdar & Jason Snyder - Oakland, California

FIRST Board Member Tejal and her husband Jason have announced they are making a \$6,000 gift, and they are asking their network to match it!



Schlitt Family - Livonia, Michigan

First-time fundraisers Adam and Sam Schlitt decided to host a Cornhole Tournament benefiting FIRST. They were amazed at the dollars they raised and the connections made. They met four new friends with ichthyosis and raised \$14,000!

UFIRST Scholarships awarded to five members

FIRST thanks Beiersdorf, makers of Eucerin and Aquaphor, for generously providing funds for our UFIRST Scholars program. FIRST received 16 applications in this award period. We are proud to announce these five scholarship recipients:



Meghna Cacatte

ichthyosis vulgaris

George Washington University,
Washington, DC

Pursuing an executive masters in
Healthcare Administration

Goals & Aspirations: With the
knowledge I attain through this
degree, I plan to set up and
manage an intergenerational care
facility in India.



Andrej Lozevski

ARCI-lamellar

University of North Carolina –
Chapel Hill, Chapel Hill, NC

Pursuing a degree in Biology

Goals & Aspirations: To become a
medical doctor and researcher for
rare diseases and develop genetic
therapies.



Devin Mong

X-linked ichthyosis

Westminster College, New
Wilmington, PA

Pursuing a degree in Biology

Goals & Aspirations: My goal is
to obtain a degree in Biology and
pursue a career in research.



Eileen Uthuppan

ARCI-lamellar

Valparaiso University, Valparaiso, IN

Pursuing graduate physician
assistant studies

Goals & Aspirations: To become
a derm PA, potentially become a
digital designer on the side (or full
time in the future) and help develop
the design layouts of medical/
derm apps, grow my Simply Skin
Magazine and raise awareness for
ichthyosis.



Gabrielle White

Netherton syndrome

Cabrillo College, Aptos, CA

Pursuing a nursing degree

Goals & Aspirations: Masters
of nursing, to help facilitate and
further Netherton syndrome/
ichthyosis research.

Beiersdorf

Aquaphor[®]
HEALING OINTMENT

Eucerin[®]

Grand Canyon challenge supports IAM

Last year, 12 FIRST members completed a strenuous two-day hike of the Grand Canyon to raise funds and awareness for ichthyosis. The event was so successful that FIRST is returning for a new challenge in 2024 during May's Ichthyosis Awareness Month.

Five hikers and their guides plan to descend from the South Rim, cross the Colorado River, and climb back to the South Rim in one day. The round-trip covers 18 miles and 4,700 feet of elevation

gain in the harsh desert climate. The hike is scheduled for May 5.

The hikers are Ben Aughenbaugh, parent of an affected child; Matt and Calvin Kocher, parent and affected child; Adam Schlitt, parent of affected child; and Chris Boynton, FIRST CEO.

Join us in wishing these hikers a safe and successful adventure! To donate in support of the team, please visit givebutter.com/IAM2024.



Ben Aughenbaugh



Calvin and Matt Kocher



Adam Schlitt



Chris Boynton

DERMAL
therapy[™]

Helping people with Ichthyosis for 30 years

Alpha Hydroxy + Urea Moisturizers
for FIRST Members

Hundreds of FIRST members use Dermal Therapy's odor free, non-greasy moisturizers to improve skin and scalp instead of expensive prescription products.

MONEY BACK GUARANTEE

Alpha Hydroxy Lotion 16oz <small>(10% Urea, 10% Lactic Acid)</small>	\$28	15% off* + Free Shipping Over \$100
Body Wash 16oz <small>(5% Urea, Excellent for body & scalp flaking)</small>	\$18	
Dry Spot Treatment Roll On 3 oz <small>(25% Urea, 5% AHAs)</small>	\$15	

* Exclusive FIRST member discount of 15% for orders of 3 or more pieces

Call us to order today
and receive your FIRST member discount!

1-800-668-8000

dermaltherapy.com

IAM offers collective chance to raise awareness

For the entire month of May each year, Ichthyosis Awareness Month is dedicated to raising awareness about ichthyosis and related skin types! By posting your story, hosting a fundraiser, reaching out to your congressperson, or educating your community, you can rally support and encourage a deeper understanding of the issues that affect those with ichthyosis.

IAM plays a pivotal role in uniting communities and inspiring advocacy. By acknowledging the challenges of living with ichthyosis, IAM empowers people to make a difference.

Don't forget to tag FIRST in your social media posts using #IAM24. Together we can make a difference!



2024 NATIONAL CONFERENCE

Friday, June 28, to Sunday, June 30
Albuquerque, New Mexico

#FIRSTNC24



Registration

Rates (after May 1): \$450 adults / \$200 children 4-12 / \$50 children 1-3

Includes most meals, all conference sessions and social events

Childcare (ages 1-12): \$50 per child

Location

Hotel Albuquerque at Old Town
800 Rio Grande Boulevard NW
Albuquerque, NM 87104

Room rate: \$189 per night

Hotel: 505-843-6300

Reservations: 1-866-505-7829

hotelabq.com

The FIRST National Conference is quickly approaching. We hope to see you in Albuquerque, New Mexico, in June!

FIRST members travel from across the world to attend the three-day National Conference. Lectures, breakout sessions, research initiatives and special events are designed to provide education and inspiration for people of all ages living with ichthyosis or a related skin type, their families, and physicians that care for them.

The conference schedule and setting encourage connections among members. Attendees also have the unique opportunity to schedule a clinical visit and meet one-on-one with our ichthyosis medical experts.

For conference details and registration, visit firstskinfoundation.org/albuquerque-2024.

Ichthyosis influences art for keynote speaker Pierre Brault

As keynote speaker at the National Conference, FIRST member Pierre Brault will tell his story of how ichthyosis has affected his personal life and how it has informed his career as an actor, playwright and stand-up comedian.

Having overcome many personal demons to finally come to an acceptance of ichthyosis, Brault's career has become the perfect outlet to explore the emotional depth that his condition can bring. He will also discuss the challenges of writing an autobiographical play about life with this unique and challenging imperfection.

Brault, who has epidermolytic ichthyosis, says he is inspired by the courage and resilience of the FIRST community and is looking forward to finally meeting his brothers and sisters.

Brault is an Ottawa-based award-winning actor, playwright, comedian, and arts instructor. He has appeared on stages throughout his home country of Canada and internationally. He is also a veteran of film and TV, most recently in a starring role in Hallmark's "Catch Me If You Clause". He has written and appeared often on Canadian radio and television and wrote and starred in his own award-winning Bravo television special.



Financial aid applications were received from 30 families. Thanks to support from Beiersdorf, FIRST is proud to provide significant scholarships to every family that applied, supporting a total of 58 individuals. Half of the recipients are affected with ichthyosis. Eight families will be traveling from outside of the United States.

10 reasons to attend the National Conference

Are you unsure about what to expect from the FIRST National Conference? On the fence about whether to attend? Or just looking to make the most of a new experience? We've assembled this list of 10 great reasons to join us in Albuquerque.

- 1 Meet someone else like you.** Ichthyosis is so rare that you might have felt alone and that no one else understands. Here, you will meet others who “get” you and what you go through on a daily basis, whether you have ichthyosis or are a parent raising a child.
- 2 Be inspired.** You will meet others who have already been through your current stage of life and provide inspiration for you on how to get through it.
- 3 Let your guard down.** Imagine a place where you can be yourself with no fear of judgment, stares, rude comments, or having to explain your condition. It is freeing!
- 4 Make lifelong friends.** You will meet new people who you will feel like you've known your whole life. The friendships forged here are lifelong. Some say it feels like a big family reunion and that you gain a family that you never knew you needed.
- 5 Meet the experts.** You will have the opportunity to meet and learn from the lead doctors in the ichthyosis field, ask them questions, get updates on research, and participate in studies. You will also learn from the experts living it – those with ichthyosis and those raising them.
- 6 Learn about skin care.** You will gain insights on the best products for your skin and the best ways to handle certain situations. You'll even get some free products to try!
- 7 Meet the FIRST staff and board of directors.** This is your opportunity to meet the people who make FIRST happen, to ask them what the future looks like, and to provide feedback on what you would like to see from FIRST.
- 8 Find a support group.** A highlight of every conference is the breakout sessions, where everyone comes together to discuss their life experiences and perspectives. There's a group for almost everyone: young adults, adults, moms, dads, grandparents, and spouses.
- 9 Be a role model.** So many members return year after year to pass on their wisdom to the next generation. It is remarkable to watch their confidence grow with each conference.
- 10 Visit a new place.** Many families turn the conference into a mini vacation and tack on an extra day or two before or after the conference to explore the city we are in. Your family may have never considered visiting before, but now the opportunity is there for you to take!

“There's always a good reason to go. You'll find yours when you get there.”
- Lowell Wright

Conference agenda offers variety for all

The National Conference is packed with educational sessions and opportunities to connect with members. Here's a peek at the tentative agenda.

THURSDAY, JUNE 27	
6:30 PM – 8:30 PM	Registration
6:30 PM – 8:30 PM	Welcome Reception Sponsored by BioCryst
FRIDAY, JUNE 28	
7:00 AM – 8:00 AM	Yoga with Mui
7:30 AM – 9:00 AM	Registration
7:30 AM – 9:00 AM	Breakfast
7:30 AM – 8:30 AM	FIRST Timers Welcome and Orientation for Parents, Children & Adults
9:00 AM – 10:30 AM	Conference Opening and Keynote Speaker – Pierre Brault
10:30 AM – 10:45 AM	Coffee Break
11:00 AM – 12:00 PM	Taking Your Show on the Road
12:00 PM – 1:00 PM	Lunch
1:00 PM – 1:45 PM	Sessions:
	Insurance Issues & How to Solve Them
	Self-Advocacy - Cultivating Resilience
	Nail, Hair and Scalp Care for Adults
2:00 PM – 2:45 PM	Sessions
	Self-Care While Parenting a Rare Child
	Nail, Hair and Scalp Care
	Seeing Other Specialists
3:00 PM – 3:45 PM	Sessions
	Parenting Young Children with Ichthyosis
	Dealing with Difficult People
	Normalizing and Increasing Access to Mental Health Resources
4:00 PM – 4:45 PM	Sessions
	The Camp Experience
	Genetics and You - Understanding Your Genetic Diagnosis
	Ichthyosis Life Hacks
6:00 PM – 8:30 PM	Potential Evening Activity (TBD – separate registration required)

SATURDAY, JUNE 29	
7:00 AM – 8:00 AM	Yoga with Sacha
7:30 AM – 9:00 AM	Breakfast
9:00 AM – 9:15 AM	Morning Briefing - Preparing for Day 2
9:15 AM – 9:45 AM	Keynote Speaker - Alex Howard - Being Your Best Advocate
11:00 AM – 12:00 PM	Physician Panel Discussions
	Children
	Young Adults
	Adults
12:00 PM – 1:00 PM	Lunch
1:00 PM – 2:00 PM	Types of Ichthyosis Sessions
	Ichthyosis With Confetti
	Epidermolytic Ichthyosis & PPK
	KID, CHILD, PRP, TTD & Related Skin Types
	ARCI-Harlequin Type
	ARCI-Lamellar & CIE Types
	Ichthyosis Vulgaris & X-Linked Ichthyosis
	Sjogren-Larsson Syndrome (SLS)
	Netherton Syndrome
2:00 PM – 2:30 PM	Break
2:30 PM – 4:30 PM	Sharing Sessions
	Parents (Groups for Moms, Dads, Grandparents)
	Young Adults (Groups for Pre-teens, Teens, Young Adults)
	Adults (Groups for Women, Men, Family)
4:45 PM – 5:30 PM	Keynote Speaker - Lowell Wright
6:30 PM – 10:00 PM	Celebration Dinner sponsored by Quoin Pharmaceuticals
SUNDAY, JUNE 30	
7:30 AM – 9:00 AM	Breakfast
9:00 AM – 9:45 AM	Ichthyosis Research Update
9:50 AM – 10:45 AM	Participating in Clinical Trials: What to Expect
10:45 AM – 11:00 AM	Break and Check Out
11:00 AM – 11:15 AM	Closing Remarks

Don't forget to download the WHOVA app to make personalized plans, including meetups with members. Extend your connections outside the sessions to a meal, the lobby, or the pool.

New this year: Research poster session

Researchers will highlight ichthyosis research and answer attendee questions.

Enjoy all that Albuquerque offers

Submitted by Eleanore Nestlerode

Albuquerque is an amazing place, with many unique corners to explore. As a resident of New Mexico for 30 years, I am excited to welcome the FIRST community to my home state. I hope you'll get a chance to experience all Albuquerque has to offer!

Here are some suggestions for making the most of your visit:

- **Visit Old Town:** Explore the historic heart of Albuquerque with its adobe buildings, galleries, shops and restaurants – all just steps away from the Hotel Albuquerque!
 - **Admire the view:** Ride the Tram up Sandia Peak to the observation deck at more than 10,000 feet. There are short hiking trails and a restaurant at the peak.
 - **Explore the Petroglyph National Monument:** Hike or bike through this vast area filled with ancient petroglyphs.
 - **Wander Albuquerque BioPark:** Explore the zoo, aquarium, and botanical garden all in one place.
 - **Take a Breaking Bad tour:** Visit filming locations from the popular TV series and learn about its production.
 - **Discover the Indian Pueblo Cultural Center:** Learn about the history and culture of the 19 Pueblos of New Mexico.
 - **Tour a museum:** Find the fit that's right for you. Nearby options include Anderson-Abruzzo
- Albuquerque International Balloon Museum, Explora! (an interactive children's museum), the Albuquerque Museum of Art and History, and New Mexico Museum of Natural History and Science.
 - **Explore the Rio Grande Nature Center State Park:** Enjoy birdwatching, hiking trails, and educational exhibits about the Rio Grande ecosystem.
 - **Take a college tour:** Let your teen get a sense of campus life. The University of New Mexico is located just a mile outside downtown.
 - **Check out the nightlife:** Music, dance, theater and more await you after dark in Albuquerque.
 - **Enjoy Southwestern cuisine:** Enjoy delicious New Mexican dishes such as green chile stew, carne adovada, and sopaipillas at local restaurants. Here's a sample of options that offer a taste of the vibrant culinary scene in Albuquerque:
 - **Sawmill Market:** A food hall featuring 27 local merchants and a central outdoor dining space.
 - **El Pinto Restaurant & Cantina:** Known for its authentic New Mexican cuisine and beautiful patio dining.
 - **Frontier Restaurant:** A local favorite, famous for its hearty breakfast burritos and New Mexican dishes.
 - **The Range Café:** Offers a diverse menu with Southwestern flavors and a cozy atmosphere.

- **Sadie's of New Mexico:** Known for its flavorful and spicy New Mexican dishes, especially their salsa.
- **Garcia's Kitchen:** Family-owned restaurant serving traditional New Mexican dishes in a casual setting.
- **Farm & Table:** Offers farm-to-table cuisine highlighting local ingredients in a charming setting.
- **Los Poblanos Historic Inn & Organic Farm:** Known for its upscale dining experience featuring farm-fresh ingredients.
- **Mary & Tito's Café:** Famous for its mouthwatering carne adovada and other New Mexican specialties.
- **Poki Poki Cevicheria:** Offers fresh and flavorful poke bowls and ceviche options.
- **Nob Hill Bar & Grill:** A trendy spot known for its diverse menu and craft cocktails.

These are just a few of the many activities to do in Albuquerque. Depending on your interests, you'll find plenty of opportunities for outdoor adventures, cultural experiences, and culinary delights in our vibrant city!

Enjoy Albuquerque and New Mexico!

Recommendation:
Don't forget to apply – and reapply – sunscreen! New Mexico is a very sunny state. The use of strong sunscreen is highly encouraged.

Childcare offered at the National Conference

FIRST is happy to offer childcare to families attending the National Conference in June. This service is provided for children between the ages of 1 year and 12 years. FIRST covers most of the cost of childcare. The cost to families is only \$50 per child for the entire conference weekend.

FIRST has partnered with Corporate Kids Events since the 2008 conference in Chicago. Corporate Kids Events provides age-appropriate games and activities for the children in their care.

There are typically separate areas for three age categories: infants and toddlers, younger children and pre-teens.

Parents are encouraged to drop their children off at the childcare room in the morning, before sessions start. This allows adults to get the most out of the sessions. If there is a session that you want your older child to join, parents are welcome to sign their child out of childcare to participate in the session. Children can also be removed from

childcare at any point in the day for naps, skin care or any other reason. Families eat meals and enjoy evening social activities together.

Each year, children participating in childcare at the conference enjoy their time playing together and making new friends. To register for childcare, select the option during online registration or contact Lisa Breuning, director of operations, at lbreuning@firstskinfoundation.org or 215-642-2364.

Member voices: Stories of support, strength and hope

“A shoutout to my mom Jorja Watts, who doesn't get enough credit for being an advocate for her kids and grandkids. I'm glad my mom is making sure more people know about ichthyosis, one person or pamphlet at a time.”

- Jessica Watts, Pittsburgh, Pennsylvania



“The time FIRST spends helping kids and parents is phenomenal and I applaud their belief that the only thing worse than suffering from ichthyosis is suffering alone. But the research? The research was definitely something I was interested in, and through FIRST I connected with Timber Pharmaceuticals and their study. ... For the first time in my life, the disease that has largely defined my life is gone. The scales are gone. Worrying about covering up is gone. I can sweat and my skin can breathe.”

- James Ellis, Irving, Texas



Timber Pharmaceuticals is now a part of Leo Pharmaceuticals.

“After Marc was born, the doctor in the NICU explained about the collodion membrane, but I really didn't understand. All I kept thinking was that I did not read about any of this in those baby books. Marc was still crying and finally a nurse took him out of the incubator and handed him to me. Once he was in my arms, Marc stopped crying. It then became my mission to find out whatever I could about ichthyosis.”

- Denise Benedetto, Wantagh, New York



“The first time I saw Anna's eyes... I remember this moment so clearly because I was finally with her and when the swelling went down and she opened her eyes she just locked into mine. The pain I felt from the surgery was stabbing but I had to stand and I had to stay there because she wouldn't look away so neither would I. ... In that moment no amount of exhaustion or pain would have kept me from letting her know I was right there.”

- Jennie Wilklow-Riley, Highland, New York



The stories above are excerpts from member submissions. Read the full stories online at firstskinfoundation.org/meet-our-families. If you have a story to share, contact Chris Wassel, Community Engagement Director, at cwassel@firstskinfoundation.org.

FIRST advocacy leads to film change

When a movie negatively portrayed people with ichthyosis, the FIRST community snapped into action. Through advocacy of members and FIRST leadership, and with a boost from media coverage including the Washington Post, the popular streaming service Netflix responded by removing scenes that many found objectionable.

The movie, Gandeevadhari Arjuna, was released on Netflix last fall. Scenes in the movie used photos of children without consent and depicted their ichthyosis as a result of toxic waste exposure. An online petition demanding the film's removal circulated quickly in FIRST social media groups. It would eventually collect over 12,000 signatures.

Holly Johnson, a mother of a child with ichthyosis, created the petition. By streaming the film, she wrote, "Netflix is indirectly endorsing discrimination against individuals living with ichthyosis. This goes against their commitment to promoting inclusivity, diversity and awareness in their programming."

FIRST CEO Christopher Boynton followed with an open letter to Netflix, calling on the company and the filmmakers

to edit the film to remove the images used without consent and delete scenes with misleading depictions of people with ichthyosis.

"A clear mandate for any patient advocacy organization, particularly for one that represents people with visual differences, is to harness the passion of individual voices to become one loud, clear voice to speak for capable of making real impact," Boynton said. "In the case of the Netflix movie, several volunteers worked behind the scenes to create talking points to present to the movie's producers and Netflix. Their collective efforts to reach out to those most directly affected by the movie, smartly placed media relations efforts and strong advocacy skills turned outrage into action."

Read Boynton's open letter to Netflix at firstskinfoundation.org/news/open-letter-to-netflix. Johnson's petition can be found at change.org/p/remove-offensive-movie-band-from-netflix.

STOP SCALY SKIN
with
Epilyt[®]
LOTION CONCENTRATE

Softens and Smooths
Rough, Scaly,
Dry Skin

4 Fl oz / 118 mL

WAMBIER[®]
DERMATOLOGICS

- ✓ Loosens and Removes Scales*
- ✓ Softens and smooths rough, scaly, dry skin*
- ✓ Non-greasy, clear, odorless moisturizing lotion

To order call 1-888-996-9754 or visit wambiermd.com

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

Brennan propelled KID support

Patricia Darlene (Hovanetz) Brennan

March 17, 1943 - January 7, 2024



Patti with grandson Logan

Patti Brennan was driven to fund research, support, and treatment for Keratitis-Ichthyosis-Deafness syndrome. Brennan's grandson Logan Hovanetz, a 14-year-old from Zimmerman, Minnesota, is affected with KID.

In partnership with FIRST staff and medical advisors, Brennan funded a new research effort that has been advancing understanding of KID syndrome since 2022.

Patti Brennan passed away in January after a battle with colon cancer. The ichthyosis community mourns the loss of a visionary benefactor.

The grant Brennan established has allowed FIRST to support KID in many new ways. Research among KID members examined the lived experience of people with KID. The grant funded research, including a study led by Dr. Thomas White that has already yielded a published paper. IT has also enabled FIRST to

share summaries and de-identified information with pharmaceutical and academic investigators.

In 2022, Brennan and her family journeyed to Providence for their first FIRST Conference. In the session for people with KID, the room was buzzing with new hope and excitement, punctuating the trademark support and understanding FIRST members offer one another.

Brennan's contribution to hope for a better tomorrow--and support for today--will not be forgotten.

KID (Keratitis-Ichthyosis-Deafness) syndrome is marked by dry, scaly skin. This type is extremely rare with only about 100 cases having been published. KID affects multiple organ systems. Common struggles include deafness, eye issues, baldness, and abnormal nails.

If you include FIRST in your plans, please use our legal name and federal tax ID.

Legal Name: The Foundation for Ichthyosis and Related Skin Types

Address: PO Box 1067, Lansdale, PA, 19446

Federal Tax ID Number: 94-2738019

A Great Way to Make a Difference

Do you want to make a difference in the lives of thousands of families living with ichthyosis and other skin types?

Most would say "of course!" Many people touched by FIRST throughout the years would love to ensure that the resources, expert advice and connection are available to more people, for many more years. But how can you put that desire into action? If you care deeply about FIRST, you may wish for your passion to benefit future generations.

Thankfully, there is a way. Best of all, it's easier than you may think. Naming the people, causes or organizations you care about as a beneficiary is simple and doesn't cost a thing. Many times, it can be done using a simple form online. All it takes is a few minutes. You can also designate percentages so that your loved ones remain protected.

These types of assets allow you to designate a beneficiary or beneficiaries:

- Retirement plans
- Life insurance accounts
- Individual retirement accounts (IRAs)
- Donor advised funds
- Bank or brokerage accounts

By naming FIRST as a beneficiary, you will leave a legacy that could help parents of an infant born with ichthyosis, provide scholarship money to college students, create connections at the National Conference, or support research to develop better treatments and cures.

Once you have created your gift, let us know. Share the news of your generosity so we can thank you and show our appreciation in your lifetime. Please contact Lisa Breuning, director of operations, at lbreuning@firstskinfoundation.org or 215-642-2364.

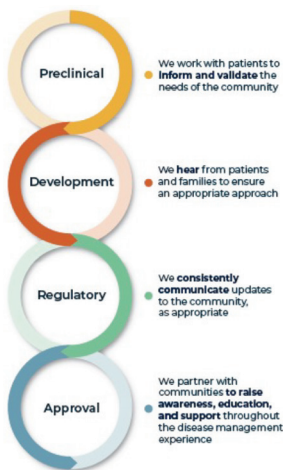


Who We Are at BioCryst

At BioCryst Pharmaceuticals, we work tirelessly, not just for, but with patient communities. At the heart of who we are and what we do is a dedication to understanding and elevating patient community voices through meaningful engagement and collaborative partnerships.

BioCryst is dedicated to advancing novel therapeutics for patients with complement-mediated and rare diseases, including Netherton syndrome. We are committed to a people-focused approach as we develop and bring first-in-class or best-in-class therapeutics to patients around the globe. Our team of researchers developed the first and only oral, once-daily therapy designed specifically to prevent hereditary angioedema (HAE) attacks (a rare, potentially life-threatening genetic condition that causes spontaneous episodes of swelling) in adult and pediatric patients 12 years and older. The therapy is approved and available in the United States and many other countries around the world. We work together across all parts of our organization - from those of us in the field or behind the bench in our labs, to those in our offices and around the globe - to bring medicines to patients who are eagerly waiting for solutions to help restore a sense of freedom for them and their loved ones.

Our research and development team based at our Discovery Center of Excellence in Birmingham, Alabama, uses a unique structure-guided approach to discover and develop potential therapies, including five new programs announced during our Research & Development Day in November 2023. These include investigational therapies that could potentially treat Netherton syndrome, diabetic macular edema (DME), and multiple different complement-mediated diseases, in addition to expanding our therapy for HAE to pediatric patients under the age of 12. Our investigational therapy being studied for Netherton syndrome, called BCX17725, is currently in the pre-clinical phase and has shown rapid distribution to the epidermis (outer layer) of the skin when administered in nonclinical studies. This year, we plan to begin evaluating BCX17725 in healthy individuals. Our hope is to achieve ‘proof of concept’ in patients (early-stage clinical trials providing evidence of the expected response in patients) with Netherton syndrome in 2025 and share our findings from this study in 2026. Our goal is to create an injectable medication that can be taken every two weeks or less frequently in both pediatric and adult patients.



BioCryst’s Global Patient Advocacy and Clinical Development teams collaborate closely with patient communities we serve to incorporate patient and care partner voices into our drug development processes. We listen to and understand the needs of patients and their communities, incorporating their perspectives into every step of the drug development process. From every decision we make to every action we take. We partner with patient organizations at every opportunity to learn from them – including convening advisory boards and listening sessions, spreading awareness about these conditions, attending conferences, and so much more! Now, we are looking to the FIRST Foundation patient, care partner, and healthcare provider community to collaborate with to ensure that we are meeting the true needs of the Netherton syndrome patient community while achieving the extraordinary.



We look forward to meeting you in person at the 2024 FIRST National Conference in Albuquerque, NM. Please stop by our table to say “hello” during the Welcome Reception on Thursday, June 27 from 6:30pm - 8:30pm.

Learn more about our commitment to patient communities!

Website: <https://www.biocryst.com/patient-advocacy/>

Email: PatientAdvocacy@biocryst.com

Scoring system reliability reinforced

The Ichthyosis Scoring System (ISS) was developed in 2019 to aid dermatologists in assessing the severity of their patients' ichthyosis. Specifically, the ISS allows physicians to provide a standardized severity rating of an individual's redness and scale across 10 body sites. Standardized tools help physicians to better understand a patient's response to treatment and help clinicians with less experience caring for people with ichthyosis accurately assess the severity of their disease. The ISS was made available as a free mobile application, allowing for widespread use from remote clinics to academic centers.

In 2021, the ISS was validated as a reliable tool for assessing ichthyosis severity using high-resolution photographs of patients with ichthyosis. We were encouraged by the reliability of the ISS in rating a patient's redness and scale burden using photographs, as the COVID-19 pandemic had moved many medical appointments to virtual platforms. The next step was to determine if the ISS could reliably assess ichthyosis severity in-person. At the 2022 FIRST National Conference, sixty-six ichthyosis patients were recruited to participate in the in-person validation of the ISS.

We found that the ISS was equally reliable in scoring ichthyosis severity in-person compared to using high-resolution photographs. Because scales on the palms and soles can appear yellow, fissure, and shed heavily, these two body sites proved to be the most difficult areas to assess using the ISS. Future in-person assessments by our team will focus on validating the ISS among patients with darker skin. Assessing redness and scale among these patients is challenging for many physicians.

Overall, the ISS is equally reliable in scoring ichthyosis severity in-person compared to using well-lit, high-resolution photographs. Having good tools to assess ichthyosis severity is very important for clinical trials to be able to assess response. We look forward to dermatologists and investigators using the ISS as a tool to advance ichthyosis research and treatment.



Research summarized by
Caroline Echeandia-Francis,
Yale University.

Distinct Skin Microbiome Community Structures in Congenital Ichthyosis

Khek-Chian Tham, Rachel Lefferdink, Kaibo Duan, Seong Soo Lim, X.F. Colin C. Wong, Erin Ibler, Benedict Wu, Hajar Abu-Zayed, Stephanie M. Rangel, Ester Del Duca, Mashkura Chowdhury, Margot Chima, Hee Jee Kim, Bennett Lee, Emma Guttman-Yassky, Amy S. Paller, John E. A. Common; British Journal of Dermatology, 28 May 2022

This study looked at the microorganisms living on the skin of people with ichthyosis. Disorders with similar barrier impairment show changes in the bacteria, viruses and fungi found on the skin, including ones that exist on normal skin and protect against infection.

Researchers collected samples from the scalp, upper arm, and lower back of people with healthy skin and those with ichthyosis of a variety of subtypes. Not surprisingly, the types and amounts of bacteria, fungi, and viruses on the skin of people with ichthyosis were different from those on the skin of healthy individuals. Specifically, some microorganisms associated with a healthy skin barrier and normal skin fats were reduced (like Cutibacterium acnes, a component underlying acne, and Malassezia, a normal yeast on our skin, both of which require active oil glands to survive). Also reduced were some "commensal" bacteria that are able to kill disease-causing bacteria. Bacteria that were increased included Staphylococcus aureus (which causes skin infections in ichthyosis) and Corynebacterium, which tends to be increased as an attempt to kill the Staphylococcus aureus. These changes in the skin's microbiome may contribute to the immune responses observed in ichthyosis.

In summary, this research provides a foundation for further investigation into the relationship between the skin's microbiome, the body's lipid (fat) composition, and gene expression in people with ichthyosis. Understanding these relationships could potentially lead to therapeutic interventions to correct the microbial imbalance in the skin of individuals with ichthyosis.

<https://onlinelibrary.wiley.com/doi/abs/10.1111/bjd.21687>

Association of Somatic ATP2A2 Damaging Variants With Grover Disease

Devin Seli, Katharine T Ellis, Mohamad Goldust, Khadim Shah, Ronghua Hu, Jing Zhou, Jennifer M McNiff, Keith A Choate; JAMA Dermatol; July 2023

Researchers showed that the skin of patients with Grover's disease had mutations in the ATP2A2 gene in 80 percent of cases. The mutation in the gene was only present in the affected area of skin for each patient, suggesting a strong link between the mutation of ATP2A2 and Grover's disease. This result also highlights the contribution of acquired mutations to dermatological conditions such as Grover's disease.



Research summarized by
Devin Seli, Yale University.

2023 FINANCIAL AID BY THE NUMBERS

13

COLLEGE
SCHOLARSHIPS

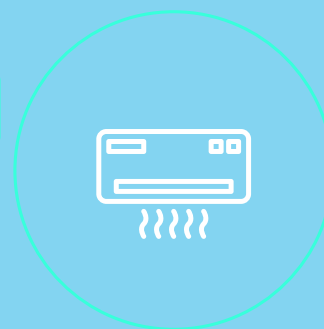
Thirteen students were awarded scholarships thanks to the UFIRST fund and Beiersdorf.



1

AIR
CONDITIONER

The newly-established Weary Fund made its first award--an AC unit to a young man with ichthyosis.



30

CONFERENCE
SCHOLARSHIPS

More families were awarded more aid than ever before! Funding provided by generous FIRST members and Beiersdorf.



22

SKINCARE
AWARDS

The Diya and Aliya Fund awarded skincare relief to 12 families around the globe. FIRSTAid awarded skincare relief to 10 US families.

