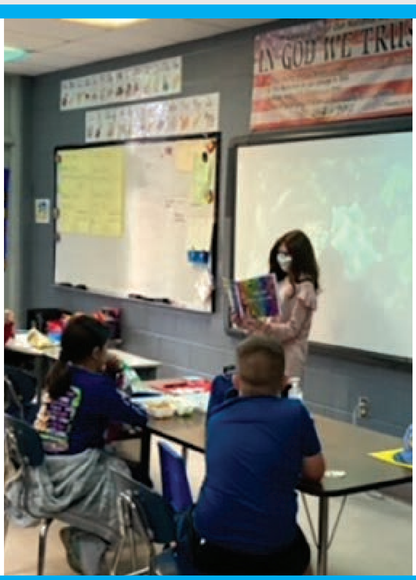
Foundation for Ichthyosis & Related Skin Types

SPRING 2023 • Volume 42, No. 1

Ichthyosis Focus

A Journal for Friends of FIRST



CROWNING ACHIEVEMENT

Carlie Foulks shines in the spotlight....6

Rising to the Challenge

Dear FIRST Members, Family and Friends

Beth Hampshire

I am so excited to have started this year as the new chairperson of the FIRST board. I intend to serve this community of amazing people with an open heart and steadfast commitment to providing our members with the services and resources they need. I am looking forward to continuing to work with the FIRST staff, board members, Medical and Scientific Advisory Board, and volunteers who make this organization as special as it is.

I recently had the opportunity to participate in the Timber clinical trial. Over the years, I have participated in a few clinical trials. The first was an overwhelming experience for many reasons. I had to completely change my skin care routine, which was incredibly scary. I didn't know how my skin would react to the change. I was uncomfortable for a short period of time, but then my skin got used to the new treatments and I was able to go through the rest of the trial with ease. I learned some new things about my skin that helped me manage it even better after the trial was over. I also overcame the fear that I had about changing my skin routine, and it opened me up to being willing to participate in other trials. This is a subject that I am now passionate about. By participating in the clinical trials, I realized I am not only helping myself but also everyone else in our community. It is one more way that I can advocate for people with ichthyosis.

In this issue, you'll read about remarkable members who are challenging themselves by hiking across the Grand Canyon or building awareness from the pageant stage. I ask everyone to challenge themselves to advocate for the ichthyosis community this year. You can participate in a clinical trial, donate or volunteer for FIRST. Start a fundraiser or reach out to another member of our community to share information. The more that we advocate for ourselves the more resources will become available to us.

It means so much to me that, as a person who has ichthyosis, I get to help lead this organization that has had such an impact on my life. I am so proud to be able to give my time and my resources to such an amazing group of people.

My warmest regards to you.

Beth M Hampshire

Beth Hampshire
Chairperson, FIRST Board of Directors

Vol. 42, No. 1 **Spring 2023**

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

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.

Foundation for Ichthyosis & Related Skin Types

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

Grand Canyon Hike a New Frontier for FIRST

In May, a group of 12 FIRST members and supporters will embark on a hike through the Grand Canyon to challenge themselves, inspire others and raise funds during Ichthyosis Awareness Month. They will travel 33 miles and ascend a combined 9,300 feet over the two-day excursion.

"This is a powerful grassroots fundraiser for FIRST but, aside from the funds raised, this is important because a group of affected people will complete an athletics endurance event together -- facing heat, sun, elevation and many miles -- to raise awareness for ichthyosis." said FIRST Director of Development Denise Gass, an affected adult who is joining the hike. "This massive hike will be so inspiring to the FIRST community! The support and participation of family members and the medical community is uplifting and ensures we will all get through it together."

The hike is led by Climb for Hope founder Andrew Buerger, who proposed a fundraising effort with his organization after speaking at the 2022 National Conference. Since 2007, Climb for Hope has taken more than 250 people safely to summits around the world, raising $750,000 for medical research in the process.

The campaign aims to raise $36,000 for FIRST. You can donate to an individual, a team (affected individuals, family members, medical professionals), or to the campaign as a whole at https://givebutter.com/IAM2023.

#ClimbforFIRST
Follow Along on Social Media!

Meet the Team

Sarah Kniskern Aughenbaugh, 43

Minneapolis, MN

Mother of affected child

The Foundation for Ichthyosis and Related Skin Types (FIRST) has supported my family with invaluable resources and relationships. I want to make sure this legacy of care and community continues.

Jackie, 45, and Rick Barrett, 50

Overland Park, KS

Affected individual and spouse

Through the FIRST relationships I have formed, I found out more ways to take care of my skin which has improved my quality of life. This is why I am taking a hike through the Grand Canyon to raise funds for FIRST.

Dr. Cheryl Bayart, 40

Cincinnati, OH

Pediatric dermatologist, MSAB member

I am excited to have the opportunity to support the ichthyosis community through this beautiful adventure.

Continued on next page

Continued from previous page

Meet the Team

Andrew Buerger, 58

Baltimore, MD

Founder of Climb for Hope

This is a life changing experience for people. We've taken breast cancer survivors and people with MS on these hikes. They had no idea they could accomplish such challenging physical feats. I'm excited to have some of our hikers learn what they're fully capable of.

Sean Cina, 47

West Caldwell, NJ

Father of affected children

I have been involved with fundraising for FIRST for the last 15 years, including hosting a yearly wine tasting at my home and running the New York City Marathon. This seemed like a great next step and opportunity!

Denise Gass, 38

New Orleans, LA

Affected individual

I'm excited to do something epic AND raise important funds for people living with ichthyosis. We will all be challenged on this journey, but ichthyosis will intensify the challenges for those of us who are affected hikers. I hope we can inspire people and make the community proud!

Rylee Iott, 21

Flandreau, SD

Affected individual

I love hiking and am always looking for ways to challenge myself. The Grand Canyon hike is the perfect combination of the two while also an amazing fundraising opportunity for a wonderful organization.

Hugh Keenan, 19

Robbinsville, NJ

Affected individual

As a person with ichthyosis, I am very invested in finding new treatments and cures. Additionally, I want to see more research toward effects on carriers that are suspected to be present. As a hiker and outdoorsman, I am hoping to test my limits beyond anything I've ever experienced!

Lisa McTernan, 62

Cortlandt Manor, NY

Grandmother of affected child

Of course my main reason for attempting to do this hike would be to help raise money and awareness for FIRST. But I would also like to challenge myself with doing this as I realize many people do hard things every day.

Sean McTernan, 41

Lower Gwynedd, PA

Father of affected child

I'm excited to participate in this challenge to support FIRST, the ichthyosis community and my daughter, Claire! FIRST is a fantastic organization that has helped our family and many others. Through this hike, I hope to be able to do my part to support our community and raise awareness with the added huge bonus of doing so with my mom, Lisa!

Robert, 53, and William, 10, Wagmiller

Mountain Lakes, NJ

Father and affected son

We are participating to help raise money for FIRST to support other families of affected children and adults. Will is also participating because he wants to show other affected kids that doing something really hard is possible, if they work and train for it.

Donate Now to Support FIRST and Our Team of Adventurers!

Shine Bright Like a Diamond: Members Gain Confidence Through Pageants *by Bailey Pretak*

Beauty, grace, glitz, glamour, poise. These are all words that can come to mind when one thinks about a pageant. Some feel that pageants are superficial and outdated for today's world, but some members of the FIRST community disagree. Through competing in pageants, they have grown in confidence, built strong friendships, and found a forum to bring awareness to issues they care about. Let's meet Carlie Foulks.

At age 12, while in the audience watching the Miss Greene County pageant, Carlie realized she wanted a turn on stage. Since then, she has competed in several different pageant systems including Miss Tennessee Volunteer, Royal International Miss, and National Elite Miss.

Carlie has grown to love the interview portion because the judges get to know her and her heart.

Pageants have several categories to compete in: evening gown, talent, fashion, and interview. Carlie has grown to love the interview portion because the judges get to know her and her heart. She admits her favorite part is talent, however, because it allows her to show off her clogging or pointe skills.

The highest title she has earned is Miss East Tennessee Royal International Miss Teen Role Model. She went on to place 15th out of 57 girls in her age division from around the world. What an amazing accomplishment!

There are many stereotypes of pageants being superficial, catty, and only about outer beauty. Unfortunately, that still exists within pageant systems. However, pageants weight the interview portion as the largest percentage of the overall score, allowing for highly intelligent, well-spoken, and driven women to compete for the title. These ladies are continually giving back to serve the communities they are in and have a strong desire to make a difference in the world. Even within categories like evening gown or fashion, the judges look for how the girls carry themselves and how confident they are, not only how they look.

Carlie placed in the top 15 in the world for her age division

Carlie was crowned Miss East Tennessee Role Model

One thing that Carlie really appreciates about pageants is that they care about what you stand for. Contestants must have a platform – a social issue or organization they are passionate about and will promote throughout their reign. Carlie's platform is called Beauty in Our Differences.

"As someone who has grown up with a few visual differences, I want to be the role model I never had," she said. "I hope that being on stage will help show girls, and boys, of any age that you can do whatever you put your minds and hearts to!"

> **"As someone who has grown up with a few visual differences, I want to be the role model I never had."**

With her platform, she uses her voice to talk about how we are all unique and the beauty to find within that. Her favorite thing to do as a titleholder is visit local schools and read books about differences to the students.

Having ichthyosis, Carlie is a rare contestant. But she has never let it hold her back. She grew up on stage dancing ballet and has a strong support system back home, so there was no fear about how others might react to her when she went to compete in her first pageant. She has found many wonderful friendships and has inspired others through her experiences.

Pageants are evolving to be more welcoming to beauty in all shapes and sizes, and more contestants with disabilities are competing than ever before. Some pageant systems even provide scholarships to help pay for college. The Miss America Organization is our country's largest scholarship pageant and has enabled many women to go to college for free or at a significantly reduced cost.

Carlie encourages anyone who has an interest in pageants to go for it! Pageants are a great way to boost confidence and can be great for everyone no matter what age.

Pageants Provide Space to Sparkle

Beth Hampshire

Beth Hampshire, chair of the FIRST Board of Directors, has been volunteering with the Miss America pageant system since 1997 through choreographing, judging, and directing at the local level.

Beth is also a pageant coach and loves helping young women achieve their goals. Beth has always had an amazing support system in her life, and she says this is her way to give that back to others. Her favorite category to coach is the interview portion. She loves watching the contestants become more comfortable speaking in a room full of people. Plus, it is good practice for future job interviews.

Her biggest challenge as a coach is watching a young woman full of potential not meet her goals because of self-doubt. Sometimes your toughest judge as a contestant is yourself , she says Her proudest moment has been the growth of one of her high school girls who finished in the top 5 at the state competition.

Bailey Pretak

Bailey Pretak, a FIRST board member, has also competed in pageants. It was something she always wanted to do, so in 2015 she began her pageant journey. She has competed with Pure International Pageants and International United Miss.

Her favorite part of the competition is the interview. She enjoys sharing her story and passions with the judges. She also loves the evening gown portion and the elegance she feels walking the stage in her best dress. Bailey's most recent title was Ms. Pennsylvania for International United Miss in 2018-2019. She says pageants have helped her confidence to grow immensely and get over her fear of public speaking.

Zoey Seboe

Zoey Seboe was a top 30 finalist for Miss South Africa in 2022. She says she entered the pageant to inspire others and educate people about rare skin conditions.

Zoey, who has lamellar ichthyosis, says she feels she is here for a purpose: to "speak to young girls and make them aware they are worthy of seeing themselves in any space."

She hopes her visibility in pageants and in her modeling will help create a better life for others. "They say knowledge is power," she said. "Once people become aware of the condition and other rare skin conditions, there will be less discrimination and more respect and love for others."

Summarized from an article by Qhama Dayile that originally appeared in DRUM, a South African online publication. Read the complete story on the FIRST website.

Evan Mayone's Legacy Lives On

Evan Mayone won the FIRST Frances Bernstiel Junior Award in 2020. Then he won it again in 2021. This award recognizes the member under the age of 18 who was top fundraiser for the year. Evan is the only person to win it twice.

Evan passed away Dec. 29, 2022, from chemotherapy complications due to leukemia. He was 17.

FIRST is proud to announce this award will now be named the Evan Mayone Award to honor his life, spirit, and commitment to others.

In remembering Evan's short yet full life, his family shares these wise lessons.

Find community

The Mayone family found support in their friends and neighbors in Portland, Maine, and within FIRST. In a Facebook post following Evan's death, his mother Kimberly Mayone wrote, "Just as we have been since Evan's birth, we will be buoyed by your kindness, love and support."

Donate blood

Cancer patients, especially children, require a lot of blood products. During treatment, Evan received many transfusions, which were possible thanks to blood donors.

Fundraise as a family

Evan worked with his mother and grandmother to plan annual fundraisers. Following his grandmother's death, he continued, writing: "Typically, I do this fundraiser in coordination with my grandmother, but due to her passing, I want to carry on her dreams and continue this wonderful tradition we had made."

Be the kind of friend you want to have

Evan had a dedicated group of friends who shared his interest in fashion, social media, food and other trends. He served as class president his freshman and sophomore years. In his final days, Evan's family played him more than 100 audio messages from friends and loved ones.

Make an impact, no matter your age

From the first family fundraiser for FIRST when he was 1, Evan was proud to fundraise for special causes. As he raised thousands of dollars over the years, he always maintained a grateful spirit.

Never give up on your dreams

Evan loved to travel and planned to study aviation and become a commercial airline pilot. He had taken his first flight lessons with his grandmother in Texas. As he fought his illness, he made plans for the places he would go and the foods he would eat when he returned home. He looked forward to the hospital staff blowing bubbles in a "bubble parade" as he left the intensive care unit – a celebration that was instead incorporated at his funeral.

To read an extended tribute, visit www.firstskinfoundation.org/news/tribute.

Tips for the Tub: How to Enter and Exit Safely *by Abby Evans*

Baths are a critical element of skin care for many people with ichthyosis. But slips and falls in the bath are common and can lead to injury. To promote independence in bathing for all ages, we recently sat down with an occupational therapist to better understand tips for bathing safely.

First, be sure to have a bathmat outside of your bathtub to reduce slipping when entering and exiting the tub. You also may want to purchase a nonslip mat for inside the bathtub, depending on your comfort and personal preference.

When entering the bathtub, use the wall to steady yourself as needed. You may want to consider entering the bathtub sideways. Put both hands against the wall and step one leg in at a time. Depending on your level of mobility, personal comfort, and physician suggestions, you may also sit on the ledge of the bathtub, gently swing one leg over at a time, then lower yourself into the tub. For even more security, consider installing grab bars within your bathtub.

When you are ready to exit the tub, you may want to consider turning onto your knees and standing up. This promotes balance and steadiness in a slippery environment. You can also use your hands to push yourself up. If it works better for you and your personal comfort, use the ledge of the bathtub to sit on and then swing your legs out of the bathtub.

These tips from a board-certified occupational therapist are appropriate for children and adults. Please contact your physician if you have specific concerns.

First Research Grant: Exploring KID Syndrome

FIRST is pleased to congratulate Thomas W. White, PhD, Professor of Physiology & Biophysics at Stony Brook University School of Medicine, for being selected as the 2022 FIRST Research Grant Recipient.

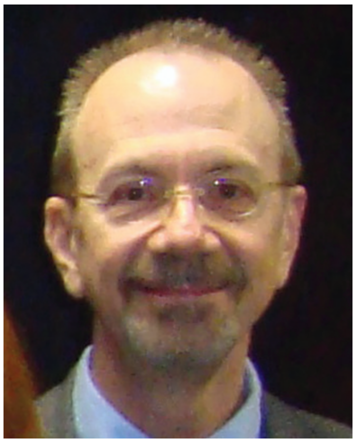
White's research focuses on increased hemichannel activity related to mutations in connexin26 (Cx26) that cause keratitis ichthyosis deafness (KID) syndrome.

Mutations in the genes encoding connexins cause several human skin diseases, including KID syndrome. These genetic diseases have no cure, and current treatment aims at providing symptomatic relief, when possible. In a recent study, White showed that a monoclonal antibody targeting the activity of the connexin protein linked to KID syndrome greatly improved skin related pathology when administered to mice.

Blocking connexin activity with specific monoclonal antibodies offers a new approach for potential treatment of KID syndrome. It also provides a valuable tool to probe why connexin mutations are so disruptive to the skin, knowledge that could lead to more novel therapeutic approaches to treating connexin skin disorders in the future.

In Memoriam

Dr. John DiGiovanna



FIRST is sad to share that John DiGiovanna, MD, died Feb. 6. Dr. DiGiovanna held several positions within the National Institutes of Health, including Head of the Dermatology Clinical Research Unit in the National Institute of Arthritis and Musculoskeletal and Skin Diseases.

He served on FIRST's Medical & Scientific Advisory Board for 23 years. His expertise and warm approach were welcomed by FIRST members at numerous National Conferences.

Dr. Phil Fleckman

FIRST learned of the passing of Dr. Phil Fleckman at press time. A tribute to the great work that Dr. Fleckman accomplished on behalf of FIRST will be printed in our next issue.

Research tests treatment with isotretinoin, secukinumab

Micah Pascual, MD
Program Class of 2024

Anna Bruckner, MD, FIRST
Medical and Scientific
Advisory Board

Micah Pascual and Dr. Anna Buckner from the University of Colorado School of Medicine summarize research published in medical journals.

Safety, tolerability, and efficacy of a novel topical isotretinoin formulation for the treatment of X-linked or lamellar congenital ichthyosis: Results from a phase 2a proof-of-concept study

Paller AS, Browning J, Parish LC, Bunick CG, Rome Z, Bhatia N., J Am Acad Dermatol. 2022;87(5):1189-1191. doi:10.1016/j.jaad.2022.02.060

Review: Oral retinoids have more risk for adverse side effects than topical retinoids due to systemic absorption. This study tested the tolerability, safety and efficacy of topical isotretinoin in patients with lamellar or X-linked recessive ichthyosis. Isotretinoin has been available as an oral medication for many years. A topical formulation is not available for patients at this time.

In the first 8 weeks of the study, patients applied either a low (0.1%) or high (0.2%) concentration of the medication to one area of skin, and ointment without the medication to another similar area of the skin. In the last 4 weeks, both areas of the skin received medication. Patients did not demonstrate abnormalities of routine lab tests or evidence of absorption of isotretinoin. Skin side effects included irritation and itching, which was rated mild to moderate in most cases. At the end of the study, more patients using the 0.1% concentration than the 0.2% concentration had improvement of scaling.

Summary This study of topical isotretinoin ointment provides preliminary evidence suggesting this medication can reduce scaling for patients with lamellar and X-linked ichthyosis, although skin irritation may limit use. Additional studies in a larger group of patients are needed.

Secukinumab responses vary across the spectrum of congenital ichthyosis in adults

Lefferdink R, Rangel SM, Chima M, et al., Arch Dermatol Res. 2022;10.1007/s00403-022-02325-3. doi:10.1007/s00403-022-02325-3

Review: Secukinumab is a medication that is used for the treatment of psoriasis, an inflammatory skin disease. The purpose of this study was to determine whether the use of secukinumab could improve ichthyosis severity (skin redness, scaling and symptoms) in individuals with congenital ichthyosis. The rate of bacterial and fungal skin infections was also assessed. Twenty adult patients with epidermolytic ichthyosis (EI), Netherton syndrome (NS), lamellar ichthyosis, or congenital ichthyosiform erythroderma (CIE) were treated in this study.

Although biomarkers of the implicated inflammatory pathways were reduced with secukinumab, the treatment did not improve ichthyosis severity overall. However, five patients (with EI, NS, and CIE) had notable reductions in redness and scaling, as well as self-reported improvements in itch, pain, and skin care management. These patients elected to continue treatment with secukinumab after the study ended. An increased risk of bacterial or fungal skin infections was not noted.

Summary Treatment with secukinumab did not lead to skin infections but did not improve ichthyosis severity for most patients with ichthyosis. It may be beneficial for some patients with certain subtypes of ichthyosis. Additional studies are needed to help identify which patients are most likely to benefit from this treatment.

Read more research reviews at
firstskinfoundation.org/quarterly-research-literature-review

first
Foundation for Ichthyosis & Related Skin Types

Ichthyosis Focus

SPRING 2023 • Volume 42, No. 1

A Journal for Friends of FIRST

PO Box 1067 • Lansdale, PA 19446-0687

Events Offer In-Person Connections and Learning

FIRST held the first of our patient meet-ups for 2023 on April 1. Nine families gathered in St. Louis, MO, to meet others and share ideas and concerns. Eleven of the 15 participants in attendance were affected adults, many of whom had never met another affected individual.

Dr. Keith Choate from Yale University joined in via Zoom and presented an overview of ichthyosis and its genetics. He then answered questions from the group.

The day ended with each person sharing their story and supporting the other attendees. It was a very educational and uplifting day for everyone in attendance.

Two additional meet-ups are planned for this year. Visit www.firstskinfoundation.org/patient-support-forums to register.

2023 Patient Support Forums

- St. Louis, MO: April 1, 2023
- Tampa, FL: May 6, 2023
- Seattle, WA: September 2023 (TBD)

2024 NATIONAL CONFERENCE

Join us in

Albuquerque, New Mexico!
June 28-30, 2024

Welcome reception June 27

More details coming soon!