

GRAND CHALLENGE

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A closer look at advocacy

Research, education and advocacy comprise the three-legged stool of patient-focused organizations like FIRST.

Research is the pursuit of better treatments or even cures. FIRST pursues this part of its mission through the direct funding of research projects at universities in the United States and around the world. We work closely with the Ichthyosis Registry at Yale University and promote participation to make this important research resource more valuable. We work with private industry as they seek to develop new treatments and bring them to market. All these parts work together.

Education, through information we provide on our website, publications, meetings (virtual and in-person), and social media, aims to provide information that helps improve quality of life. We also facilitate members educating each other. Some of the most valuable information and education is shared between members in our Facebook groups.

Advocacy is where we band together, combine our voices, and collectively let those in decision-making roles know the importance of our perspective, the shared experience of those living with this rare condition. That collective voice is powerful when pushing for legislation that impacts access to treatments, the cost of those treatments or the speed at which they become available.

Advocacy can dramatically improve the strength of the other two legs. This spring, FIRST advocates met with members of congress to encourage increases in the National Institutes of Health (NIH) budget to fund more research, funding far beyond what we could support as an organization. Those same advocates, and those writing their congressional representatives from home, requested increased funding for the Centers for Disease Control and Prevention for a program that grants funds to organizations like FIRST to better educate their communities. Articles in this issue highlight other examples of FIRST members and staff building knowledge and support across key audiences.

Advocacy can also mean sticking up for each other. When one is being treated unfairly, we band together to right the wrong. With FIRST, you are never alone. You have a powerful force behind you that will always have your back. That's the power of advocacy and the truly most important part of FIRST.



Chris Boynton
CEO



Chris Boynton

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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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Dr. Fleckman remembered for kindness, integrity

Dr. Mary Williams recalls conducting clinical visits at a FIRST National Conference alongside Dr. Phil Fleckman. After a long day of seeing children affected by ichthyosis and their families, he raised the question – why are so many of them so small for their age?

Williams credits Fleckman's observation with igniting future research. "We all agreed that there was no data on which to answer this important question, and the conversation stimulated me and my coworkers to try to find out why," she said. "This led to the work which revealed the tremendous caloric burden that having a severe form of ichthyosis with an impaired skin water barrier imposes on the growing infant and young child."

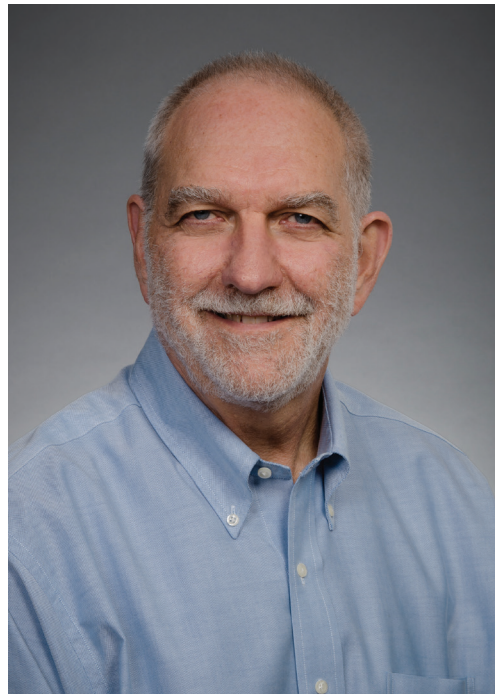
Fleckman died Sunday March 19, 2023. He had been suffering with a refractory cancer, advanced mycosis fungoides, for some time.

Fleckman was a devoted volunteer in FIRST leadership. He served on the FIRST Board of Directors from 2001 to 2009, including as Vice President from 2006-2008, and continued to advise the organization as an emeritus board member up until the time of his death. He also served on the Medical Scientific and Advisory Board from 2000 to 2021, where he was chair of the Research Committee.

He will be remembered in our ichthyosis community for the impact he had on patients and his colleagues.

Hunter Steinitz said her parents met Dr. Fleckman at their first FIRST conference, where he helped answer their questions about her harlequin ichthyosis with warmth and care – and encouraged them to keep asking the questions he could not answer. Over the years, he became a familiar face to them at conferences.

"I felt seen and understood whenever I spoke to him," Steinitz said. "While he was clearly interested in my skin, he was also just interested in me, my passions, and my regular life. He was approachable and encouraged me



Dr. Phil Fleckman

to approach other researchers for the benefit of other affected people. He helped me and my family to see beyond the daily dribble and to imagine the possibility that lay ahead."

Dr. Leonard Milstone shared a lab with Fleckman at Yale University. "Phil was a worker and a quiet leader who never sought the limelight. He cared deeply about his patients and his local and academic communities," Milstone said. "He would get a twinkle in his steely blue eyes when asked about his family. He had no hard edges and always could be counted on for an honest, straightforward opinion. Ellen and I, and many others, will miss him."

Williams added: "When I think of one word that describes who Dr. Phil Fleckman was, the word 'integrity' comes first to mind. Phil was upright, straight forward, and uncomplicated in his approach to all issues. The next term that comes to mind is 'kind'. He offered an unstinting kindness to everyone he encountered – his professional colleagues, his patients, his coworkers and associates, everyone. He was the best of physicians for his patients and colleagues and his presence at FIRST will be deeply missed."

From Dr. Fleckman's Family

Phil Fleckman's daughter Mahri Leonard-Fleckman shared these memories of her father:

At my sister Morgen's wedding in 2009, our dear family friend JR cornered me at the reception and said, "Mahri, get to know your father. He's the most understated person I know." JR's comment struck me, because while I loved my dad, I didn't really know him, or so I thought at the time. I spent the next 11 years or so trying to "get to know" my dad, meaning that I grilled him with intense questions about life, forced him sit down while I tape-recorded him, and made other awkward attempts to dig deep (poor guy).

He always went along with my attempts—he was gentle and kind, after all, and he loved me dearly—but we never really got anywhere, as you can imagine. My dad was an understated, humble person. He did not make himself the center of attention. He was a listener, always present. He said what needed to be said in the moment. Plus, I already knew him.

It's impossible to express how much I loved my dad, how much I have learned from him, and how heartbreaking his death has been. He was one of the best human beings I have known. It is also a relief that his suffering is over. I want you to know that we (his family) are so grateful for all of you—his colleagues and friends—because you are the ones who perhaps knew him best, or at least knew him in his other element, in the thing he loved most outside of his family. Thank you all for your comments and for your love. It is deeply meaningful to read them and to get to know him through you.

FIRST advocates and raises awareness during IAM

Health advocacy groups partner on shared issues at global summit



FIRST CEO Chris Boynton participated in the Global Patient Partnership Summit 2023 meeting hosted by Boehringer Ingelheim during Ichthyosis Awareness Month in May. The meeting was truly global as it took place simultaneously in Miami, Vienna and Singapore.

Health advocates from North and South America including the United States, Canada, Peru, Columbia, Brazil and Argentina, and representing many health conditions, met for important collaboration, networking and to discuss several challenges facing patients and patient advocacy organizations.

The meeting was highly participative and dealt with four challenges:

- **Empowering patients** – There has been a proliferation of information and advocacy groups, but patients report a lack of involvement in individual decision-making. The empowered patient of the future lives “with” their condition, not in spite of it. They have the resources, knowledge, confidence and access to do this.
- **Integrated care** – Care outcomes can improve through better coordination among the various healthcare practitioners patients deal with, communication, technology, and healthcare

literacy.

- **Patients with multiple chronic conditions** – As treatments improve, a significant percentage of patients are dealing with multiple chronic conditions such as hypertension, diabetes, arthritis, anxiety and obesity. Many of these are linked and people living with ichthyosis are likely to be living with multiple conditions, as well. Coordinating treatments, prescriptions and doctor visits can be a very real burden.
- **Patient experience data** – Real-world evidence of life with these conditions is an important consideration in the drug approval process. Organizations like FIRST can play an important role in gathering this information and ensuring the patient experience is properly considered and valued.

Attendees split into multiple groups to workshop solutions to the challenge statements and select ideas to move forward. Much work remains to fully develop the ideas and bring them to fruition.

UPDATE YOUR CONTACT INFORMATION

Scan this code to update your contact information **if it has changed**. FIRST would also like to collect contact information for affected young adults living with you or outside of your home.



GRASSROOTS FUNDRAISING

Did you know you can fundraise for FIRST anytime by sharing a link to our donation page on your social media channels or by email? Or use the QR code below for in-person events. Make sure to ask donors to include your name or event name in the Gift Notes section so FIRST can credit the donations to you!



FIRST advocates and raises awareness during IAM

Beiersdorf celebrates Ichthyosis Awareness Month with FIRST

FIRST partner Beiersdorf, the makers of Aquaphor and Eucerin, celebrated Ichthyosis Awareness Month at their headquarters in Wilton, CT, on May 23. Christine Wassel, FIRST community engagement director, along with FIRST members Sacha Schenker and Hunter Steinitz, and author and dermatologist Andrea Rustad, MD, presented topics to 50 Beiersdorf employees to educate them about ichthyosis.

Wassel provided an overview of ichthyosis and how FIRST serves our community. She was followed by Steinitz, who shared what it's like to live with ichthyosis. Schenker provided a live demonstration of yoga with ichthyosis and Dr. Rustad gave a reading of her book, "Skin-vincible: A Story of a Superstar with chthyosis".

The day ended after Q & A and time for the entire group to interact and learn from each other. Later, Beiersdorf utilized their social media channels to raise awareness and money for FIRST.

The presentations are posted to FIRST's YouTube channel, youtube.com/firstskinfoundation.

FIRST looks forward to continuing this important partnership in the years to come.



FIRST representatives Christine Wassel, Sacha Schenker, Hunter Steinitz and Andrea Rustad, MD

Joining forces with CSD on Capitol Hill

CEO Christopher Boynton and FIRST members Eric Schweighoffer and Jennie Wilklow-Riley attended the Coalition of Skin Diseases Hill Day to advocate for all those living with skin disease, including those with ichthyosis and related skin types.

People with skin disorders sometimes come up against barriers to care. The CSD supports state and federal initiatives that expand patients' access to health coverage and health care; remove financial, administrative, and legal barriers to care, providers, and treatment alternatives; and give patients a voice in their care.

CSD supports legislation to:

- Limit insurance practices** that act as barriers to care
- Establish guidelines** to protect patients from high out-of-pocket costs
- Improve access** to care
- Promote equity** in the nation's healthcare system
- Expand covered benefits** to include co-morbid conditions
- Ensure coverage** for medical equipment devices
- Preserve protections** for patients with insurance coverage
- Increase coverage options** for uninsured Americans
- Modernize health insurance policies** to better protect patients



Eric Schweighoffer, Jennie Wilklow-Riley and Christopher Boynton advocated for all FIRST members at the CSD Hill Day.

Members connect at Tampa forum



Over 30 people gathered in Tampa in May to meet, share skin care tips, learn and gather and give support.

Dr. Sharon Albers, from The University of South Florida, presented an overview of ichthyosis and answered questions from attendees. Jess Raiz from Timber Pharmaceuticals explained what to expect when participating in a clinical trial and why it's so important to do so.

Prior to the meeting, a few attendees had never met another person with ichthyosis. Many exchanged contact information so that they could continue to support each other moving forward.

The final patient support forum of 2023 is scheduled for Sept. 30 in Seattle. For more details and registration info, visit firstskinfoundation.org/patient-support-forums. We'd love to see you there!



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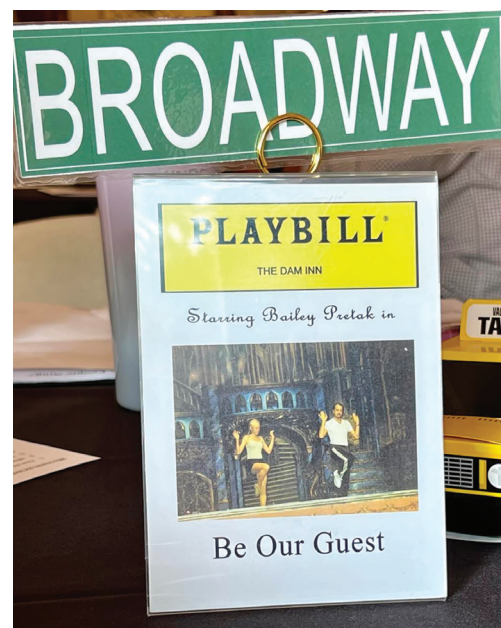
Grassroots: Members support FIRST through fun events

Bailey's Broadway Birthday Bash

Bailey Pretak held her annual birthday fundraiser "Bailey's Broadway Birthday Bash" on Saturday, June 17, at The Royal in Ridgway, PA.

Pretak came up with the idea during the 2020 pandemic. Gatherings were limited when her birthday rolled around, so she asked several of her talented, musical theater-loving friends to record themselves singing their favorite Broadway tune. They submitted the videos and, on her birthday, everyone gathered virtually to watch the performances. The following year an in-person concert was held, and it has become an annual tradition since.

This year's show featured over 20 performers that were a mix of professional musicians, college students studying musical theatre, and some of Tracie Pretak's (Bailey's mom) voice students. They presented songs from musicals such as *Six*, *The Phantom of the Opera*, *The Sound of Music*, *Wicked* and many more. Guests paid for a ticket to the show and had the opportunity to bid on themed auction baskets. It was a great evening and Pretak was able to raise over \$3,000 for FIRST!



Night at the ballpark in New York

Over 300 friends and family of Marc Benedetto and Kylie Saccente gathered on June 2nd to support and raise awareness for ichthyosis at a Mets game. Over the years, FIRST Night Out at the Ballpark events have taken place across the United States and continue to be a way for friends, families, and FIRST members to connect. FIRST members, along with family and friends, have been gathering at Citi Field, the Mets Stadium, each year for many years.

On this particular night, the Mets may have lost, but the ichthyosis community celebrated many wins, as affected members raised awareness. The Benedetto and Saccente families were honored via a special ceremony on the field prior to the start of the game. Additionally, the section where everyone was seated together received some extra special attention throughout the game.

FIRST thanks the Benedetto and Saccente families – and all the families who have supported FIRST through Night Out at the Ballpark events – for raising awareness and funds through these fun celebrations!



Grand Canyon hike challenges members, supports FIRST

May 2023 was the first time FIRST combined fundraising and programming in a way that would physically challenge and exponentially inspire and connect our community. Most people with ichthyosis and related skin types sunburn and overheat easily while struggling to sweat. This made climbing the Grand Canyon an especially challenging mission.

The group included 17 people. There were 12 fundraising hikers including four affected members, one affected FIRST staffer, six family members and one dermatologist. They were led by guides from Rare Earth Adventures and Andy Buerger, who advised the expedition.

The hiking team assembled from across the country beginning in the fall of 2022. Slow and steady, they began planning, fundraising and training. They ranged in age from 10 to 62, with a diverse range of fitness.

The first challenge was filling the group. FIRST wanted to fill the capacity of 12 spots but was prepared to share the expedition with another group if necessary. By the end of Dec 2022, all 12 spots were filled with people wanting to hike for FIRST and who agreed to meet the fundraising

minimum of \$3,000 per person.

The next challenge was meeting that fundraising minimum. Some participants were experienced fundraisers while others were learning the basics for the first time. Participants created their own fundraising page and set about emailing, texting, posting and talking about their mission to their networks. Their fundraising efforts reached far and wide, spreading awareness of ichthyosis and bringing messages of hope, dedication and community support. The leading fundraiser was Sarah Aughenbaugh, who raised more than \$12,600. Ultimately, the hike raised more than \$45,000 for FIRST!

Then, the challenges grew more specific to the hike itself. In mid-May, the National Parks Service announced that the North Rim of the Canyon would not reopen in time for the trip. This meant the guide team had to re-route the group to stay at the South Rim. The group originally planned to go from one rim to the other, and back again. The new route would be shorter, but still grueling.

FIRST was committed to making the hike as safe as possible, with all potential planning and precautions

Continued on next page



The hikers

Twelve FIRST members, ranging in age from 10 to 62, comprised the team: Sarah Aughenbaugh, Jackie Barrett, Rick Barrett, Cheryl Bayart, Sean Cina, Denise Gass, Rylee Iott, Hugh Keenan, Lisa McTernan, Sean McTernan, Bob Wagmiller and Will Wagmiller.

In addition, three FIRST members fundraised from home in support of the mission: Sheila Boardman, Jane Goff and Beth Hampshire.

Tips from the canyon

“We got started at dawn to maximize trail miles before the hottest part of the day.”

-Connor Henzel, lead guide

“Will wore a cotton t-shirt under his UPF shirt. We soaked both because cotton stays wet and cool longer than wicking fabrics, while the UPF shirt provides strong UV protection.”

-Bob Wagmiller, father to affected son Will, age 10, both hikers

“During training, I noticed my heart rate and conversational ability indicated when I was approaching overheating. I watched my heart rate and communicated it to the guide I was hiking with. After a few hours of this, he was able to infer my heart rate from his own, and when our conversation died down, he knew we needed shade and a break.”

-Denise Gass, affected hiker and FIRST staffer



taken for those hiking with ichthyosis. The FIRST Medical and Scientific Advisory Board conferred on topics such as blistering, electrolytes and hydration. FIRST worked with Rare Earth Adventures to explain the ins and outs of life with ichthyosis, and to ensure all guides were prepared to support the team. The guides and hikers connected remotely to prepare, discussed details upon arrival in Arizona, and then worked throughout the hike to constantly monitor and support all those hiking.

On May 20, the hikers and guides flew to Phoenix from across the country. Packing into three vans, the team drove to the Grand Canyon campground where they bedded down in tents by pairs. The two-person tents were a challenge, with Sean Cina joking “I don’t sleep this close to my wife!”

The first day of the hike, May 21, the guides rose at 3 am to start coffee and breakfast. By 4 am, the hikers were all awake, lacing boots and applying sunscreen in the dark. It was about 45° but would later climb to near 100°. After a quick breakfast and a ride to the trailhead (blaring “Eye of the Tiger”), the group descended into the Grand Canyon. They each carried trekking poles and wore backpacks with at least three liters of water, snacks and sunscreen. One hiker carried the names of her donors, another carried artwork from her kids, and another carried the group’s #IAM23 flag, worn like a cape.

Unfortunately, within the first mile, hiker Jackie Barrett rolled her ankle resulting in a fracture and an early departure from the trail. The bad luck in getting injured so early was mitigated by the fact that she had just a “short” trek out of the canyon with a guide after pressure wrapping her foot.

The rest of the group journeyed on, moving into three different pace groups. The route was a total of about 18 miles and descended via South Kaibab Trail for 7 miles of descent. At the bottom of the canyon, the group traveled 1.5 miles of flat trail crossing the Colorado River to Phantom Ranch,



a tiny, remote spot to buy a postcard and lemonade. After a brief respite, the hikers returned across the river and up Bright Angel Trail. The last leg was a grueling 10 miles uphill. The afternoon high temperature was around 100° with direct sun.



See videos from the hike at [youtube.com/firstskinfoundation](https://www.youtube.com/firstskinfoundation).

No one suffered overheating, heat exhaustion or even sunburn!

Heat and sun precautions were many. Guidelines while hiking included drinking a liter of water per hour and applying sunscreen every hour. Beiersdorf sponsored the hike and provided SPF products for face, body, and lips. The hikers were careful about clothing choices, as well, with many wearing sun-protective shirts,

long pants, and wide-brimmed hats. Coolibar generously provided long sleeve shirts. To manage the heat, the group doused cooling towels, hats and shirts in streams and even in hydration bladders. Special thanks to Frogg Toggs for providing cooling towels and Heliocare for providing sun protection supplements.

Several water spigots along the route were closed, meaning the hikers had to plan in advance and carry more water than usual. Finally, the hikers were advised to bring snacks amounting to about 200 calories per hour.

Other physical challenges included blisters, asthma, altitude adjustments, cramps, fatigue, chafing and more. Of course, there were immense mental challenges, as hikers pushed up against their limits and faced fears. For many of them, it was their first time moving at those inclines for that length of time. Some listened to headphones, while others engaged in moving meditations to pass the time.

During the trek, ichthyosis was top of mind. The affected hikers were keenly aware of the additional challenges ichthyosis posed to their trek and remained vigilant about safety

Continued on next page

precautions. The family members on the hike contemplated anew the challenges facing their loved ones. A dermatologist on the trip, Dr Cheryl Bayart, commented about how inspired she was to witness the perseverance of the whole group. In particular, the triumph of the affected individuals moved her, as she recalled counseling new families afraid their children with ichthyosis would never be able to play sports.



The first hikers exited the canyon in just over 9.5 hours. The last of the group finished in about 15 hours. Upon leaving the trail, each hiker was greeted by others on the team with cheers, high fives, and memories to last a lifetime.

The second day, the group was given options to do the same trail in reverse, or hike and sightsee around the rim. Two intrepid hikers journeyed back in the canyon for another grueling day. The other 15 people hiked several miles around the canyon and learned about the history of canyon geology and indigenous peoples of the region.

As hiker Sean Cina wisely surmised, the trek was a testament to the three pillars of FIRST's mission – educate, inspire, and connect. The staff and board at FIRST applaud the efforts of the hikers in taking forward the mission of FIRST!



FIRST is planning a similar fundraiser in the future!
If you are interested in participating, please email Denise at dgass@firstskinfoundation.org.



FIRST thanks Beiersdorf, makers of Eucerin and Aquaphor, for sponsoring this event.

Additional thanks to Coolibar, FroggToggs and Heliocare for generous donations of supplies.

FIRST-Aid offers financial support

FIRST members may sometimes have a tough time navigating the emotional, psychological and logistical pressures of life associated with ichthyosis. Financial stress can add to the struggle.

FIRST-Aid is our new program to help provide financial support to individuals affected with ichthyosis in the management of their skin. Grants can be used to support skin care needs, including purchasing products, devices or services.

Grants may be awarded during two application cycles each year. The deadlines for each cycle are January 31 and July 31. For more details or to submit an application, visit firstskinfoundation.org/first-aid.

FIRST-Aid grants are awarded based on the availability of funds. If you would like to support this new fund, please visit the website to become a donor.

FIRST-Aid is just one of the financial assistance programs offered to members. Help is available from several financial aid funds for those who need it.

Diya & Aliya's Friends (DAF) Skin Care Fund

Thanks to the generosity of the Shahnaz Kraybill family and their family and friends, FIRST has established this fund to help alleviate some of the financial burden that may be facing families with affected children. This special fund is available for children under the age of 18 years around the world affected with ichthyosis and is intended to provide financial assistance to purchase lotions, medicine, and treatment necessities.

National Conference Financial Aid Fund

A financial aid fund is available for families to apply for financial assistance to attend the FIRST National Conference. Since 2008, FIRST has awarded \$98,000 for 111 families to attend the conference.

Making an Impact, Creating a Legacy

Many people ask FIRST how they can make an impact for others with ichthyosis and related skin types. There are so many ways to make a difference in addition to donating or hosting an event, such as:

Estate Planning/Leaving a Legacy

Many planned gifts originate from those with only modest incomes. Individuals with wealth are also good candidates.

Bequests are made in wills, living trusts, and other estate plans.

Life Income Gifts This plan offers the donor income in return for making an irrevocable gift.

Life Estate This provides the Foundation with the gift of a residence or farm while allowing the donor the right to continue to live there for the remainder of his or her life.

Charitable Lead Trust provides the Foundation with income for a term of years and leaves the remainder to the donor's heirs.

Life Insurance This designation

provides the foundation to receive benefits upon the death of the insured.

Corporate Donations and Matching Gift Program Most large corporations budget for charitable contributions. Many prefer to give to causes that help their employees or families, so please ask.

Please contact Christopher Boynton for more information.
cboynton@firstskinfoundation.org.



UFIRST Scholars Program

The UFIRST SCHOLARS Program was established in 2010 by a seed gift from Valerie & David Scholl for costs associated with post-secondary education. This program has provided 127 awards to 72 students totaling more than \$147,500.

The Weary Fund

Terry and Robert Melton have created The Weary Fund through a generous contribution in honor of Terry's parents, Dr. Peyton and Janet Weary, and their strong commitment to FIRST. This fund provides window air conditioning units to people with ichthyosis who do not have access to them.

Requirements and application deadlines vary by program. Learn more at firstskinfoundation.org/financial-aid.



Ichthyosis contributes to mental health concerns

Submitted by Angela J. Luo, medical student at the Yale School of Medicine

In addition to physical symptoms, individuals with ichthyosis can experience social challenges due to looking different from others. This quality-of-life impact can contribute to a common set of mental health symptoms.

In a 2020 study, researchers found that both depression and anxiety are associated with ichthyosis in adults and children, with increased quality of life burden due to disease correlated with a greater likelihood of depression and anxiety. Among adults diagnosed with any form of ichthyosis, 34.4% screened positive for depression and 27.3% screened positive for anxiety ^[1]. Among children diagnosed with any form of ichthyosis, 30.2% screened positive for depression and 37.7% screened positive for anxiety ^[1]. Although we recognize that depression and anxiety can be affected by multiple factors, such as life stressors and family history, the high prevalence of depression and anxiety in individuals with ichthyosis underscores the need for clinicians to provide depression and anxiety screening regularly to those affected with ichthyosis.

This association of skin disease and increased prevalence of depression and anxiety is not exclusive to ichthyosis. Other skin diseases with visible disease manifestation, such as psoriasis and atopic dermatitis (eczema), are also both independently associated with increased rates of depression and anxiety in both adults and children ^[2] ^[3].

Unique challenges of rare diseases

As a rare skin disease, there is still much to discover about ichthyosis. Treatments are often time-consuming and require a multi-pronged approach to alleviate uncomfortable symptoms, such as itch and dryness, and to improve quality of life. In contrast to other disorders, for which targeted therapies can lead to near-complete resolution of symptoms, therapies for most forms of ichthyosis remain under development. As a result, the mental health burden in ichthyosis may be alleviated at a slower rate compared to other skin diseases.

What are some efforts to address these struggles?

Medical support

Understanding ichthyosis as a disease and learning to live with its associated symptoms may improve quality of life and treatment adherence in individuals with ichthyosis. A dermatologist who diagnoses ichthyosis and develops a treatment plan with the patient can best support patients to manage the symptoms of ichthyosis. The managing dermatologist can provide the most up to date information about ichthyosis treatment and collaborate with other medical professionals to suggest appropriate clinical trials. Researchers have suggested that screening for symptoms of anxiety and depression should become standard of care in routine management by dermatologists as a part of an integrative management plan ^[1].

In addition, talking with one's primary care doctor can provide referrals to mental health providers to help manage the possible psychological sequelae of ichthyosis. Mental health providers include social workers and psychologists who can provide psychotherapy (talk therapy), as well as psychiatrists who can prescribe medications which can be helpful depending on the severity of mental health symptoms.

Caregiver support

Living with a skin disease not only affects the individual, but it also affects caregivers. Studies have found that caretakers of individuals with psoriasis and atopic dermatitis experience psychological strain and report feelings of helplessness, exhaustion, and frustration ^[4]. While caregivers play an important role in supporting individuals with ichthyosis, it is important that caregivers also receive their own form of support, such as through a caregiver support group, as caregiving can be a mentally taxing responsibility.

Community support

Lastly, connecting with a support community, both in-person and online, has been shown to be beneficial in mitigating the mental health impact of skin diseases, including ichthyosis. Living with a rare disease can feel isolating and be an experience that few peers can relate to. In a study in atopic dermatitis individuals, researchers showed that children in support groups had improved quality of life and mood, highlighting the importance of advocating for a support community in any skin disease treatment plan ^[5]. Because ichthyosis is a rare disease, when in-person support groups are unavailable, online social forums may foster a sense of belonging to a community. These social media opportunities can facilitate meaningful interaction with others also living with ichthyosis, thus allowing individuals to feel less alone ^[6].

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Member stories: Dealing with unwanted attention

A perfectly pink person

At this point in my existence, when anyone makes a comment, I laugh, giggle, smile, nod and move on. I don't waste my breath on educating people with the technical terms and descriptions of what I actually have, but if I'm feeling especially goofy, I make up a crazy story about how I got lost on an island during a vacation and had to wait hours until the next boat came out to get me, heating up under the sun of paradise, making giant S.O.S signs made of rocks and sticks. But, whatever I say, I can't seem to please their expectations, it's like they want me to tell them what it's like being thrown in the sun.

I've learned through the years that it doesn't matter what people think happened to me; they could think I'm an adventurous teenager who has no regard for skin health or protection from UV rays, or they could think I've had an unfortunate life ever since the fire that took everything. Whatever they believe, I know that I'm doing the best I can and that their opinion on how I look will not, and does not determine who I am.

Emma Hodgkinson, Woodstock, Ontario



Appearance doesn't tell the story

People stare all the time. People comment frequently about Asher's dry skin and the redness. We get comments about "ooh poor baby is sunburned" like we don't protect his skin from the sun. When in fact, he is red as a baseline. He turns red quickly from warmth or cold even when protected by SPF clothing or shade. His body can't sweat, nor can it regulate its own temperature. None of these things can be known or seen from the outside. Same goes for anyone with any physical "difference" from what you perceive is typical. You don't know the why or their story.

So, the best thing to do is not comment on it. If you know us, you know we pour 200% of ourselves into caring for Asher's skin. I'm sure he will deal with more than his share of kid comments in school. As adults, let's set a better example!

Katie Berrey, Eureka, CA



Moving beyond a bad experience

It can be difficult to be different. I took Miles with me to the mall, and he wanted to play at the playground area. Kids were running around having a blast and Miles couldn't wait to join. However, when Miles began to play the staring and the whispering started from everywhere.

My heart broke because this was the first time I actively saw Miles was bothered. Bothered by the staring, the questions, and the kids who refused to leave him alone. I didn't let him know what I was feeling. I let him finish playing and told him it was time to go and then we had a talk. I told him that it can feel overwhelming when others stare and ask questions. If you want to answer you can but if you don't want to you owe them nothing more than this is my special skin and I was born this way. The most important thing is that you love yourself.

I figured if they are going to stare let's give them something to stare at so we took turns yelling louder "I LOVE MYSELF!" down the mall corridor until we got outside.

Emily Otteni, Pittsburgh, PA



The stories above are excerpts from member submissions. Read the full stories online at firstskinfoundation.org. If you have a story to share, contact Chris at cwassel@firstskinfoundation.org.

FIRST announces 2023 Research Grant recipients

FIRST is pleased to announce the recipients of the 2023 FIRST Research Grant, Dr. Cory Simpson and Dr. Benjamin Nanes. Read on for information about the research they are leading.

Evaluating novel therapeutic strategies for Darier disease

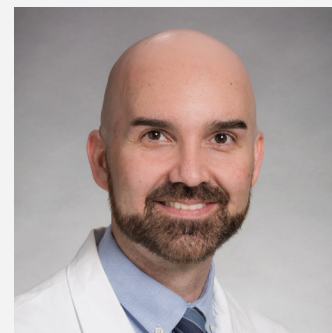
Cory Simpson, MD, PhD, Assistant Professor of Dermatology, University of Washington, Seattle

Award: \$50,000

Evaluating novel therapeutic strategies for Darier disease Cory Simpson, MD, PhD, Assistant Professor of Dermatology, University of Washington, Seattle Award: \$50,000

Darier disease is an incurable genetic skin disorder caused by impaired keratinocyte adhesion and cornification. Though linked to SERCA2 protein deficiency more than 20 years ago, there is still no FDA-approved specific treatment for this rare disease available. Dr. Simpson's research work recently highlighted that epidermal growth factor (EGF) receptor signaling is hyperactivated in Darier disease. The proposed study will assess the therapeutic potential of inhibiting this pathway. In addition, the study will leverage new platforms to identify other compounds for treating Darier disease.

Dr. Simpson states: "As a physician-scientist specializing in rare skin disorders, I see firsthand the need for innovative research aimed at identifying new treatments for patients with inherited dermatologic diseases. Commercial entities are often reluctant to invest in early pre-clinical studies for such uncommon disorders, which is why FIRST funding is critical. Our proposed studies focus on Darier disease, which causes immense suffering in patients due to chronic skin break-down and recurrent infections. As an understudied disease that lacks any FDA-approved therapy, patients with this disorder are treated using off-label medications having side effects (oral steroids) or teratogenicity (retinoids). We established in vitro human cellular and organotypic tissue models of Darier disease and are poised to identify novel treatment strategies for patients."



Functional consequences of keratin isoform switching during epidermal differentiation

Benjamin Nanes, MD, PhD, Instructor of Dermatology, UT Southwestern Medical Center, Dallas, TX

Award: \$35,000

In epidermolytic ichthyosis, mutations affecting keratin 1 (K1) or keratin (K10) disrupt the keratin mechanical scaffold. Understanding K1/10-specific functions that may not be rescued by other keratin isoforms expressed in epidermis is critical for many potential treatment strategies. This proposal aims to overcome our gaps in understanding if and how these keratins associate with signaling networks to modulate epidermal differentiation and barrier formation, which may reveal entirely novel therapeutic approaches for the wide variety of skin diseases where epidermal architecture is disordered.



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Name of affected individual _____

Grant drives progress in KID syndrome research

2022 award leads to two advances

With the support of our grant award, we have recently published a study in the scientific journal EBioMedicine using an antibody to inhibit the activity of the mutated form of Connexin 26 (Cx26) that causes KID syndrome¹. Mutations in Cx26 that cause KID syndrome significantly increase a specific type of functional activity that can be blocked by an antibody developed by Dr. Fabio Mammano's research group in Italy. In collaboration with Dr. Mammano's group, we used this antibody in a mouse model of KID syndrome. When mice were administered the blocking antibody, their skin disease symptoms were greatly reduced compared to mice that did not get the antibody. These results confirmed the idea that the specifically increased activity blocked by the antibody contributes to skin disease in KID syndrome. They also suggest that blocking antibodies may have potential as a future treatment for KID syndrome, although this will require a lot of additional experimentation.



We have also published a study in the International Journal of Molecular Sciences showing that mutations in Connexin 43 (Cx43), a protein related to Cx26, cause a similar change in functional activity as the mutations in Cx26 that cause KID syndrome². Mutations in many different connexin genes cause a variety of skin diseases. Mutations in Cx43 cause several rare genetic disorders, which can include skin disease. To determine if Cx43 mutations caused skin disease in the same manner as Cx26 mutations, we studied the functional consequences of a Cx43 mutation in the lab by expressing the mutated gene in cultured cells. We found that the mutated Cx43 had the same increase in the specific type of functional activity shown by the Cx26 mutations causing KID syndrome. These data suggest that the specifically increased activity could be a common feature of all connexin mutations that cause skin disease.

1. Antibody gene transfer treatment drastically improves epidermal pathology in a keratitis ichthyosis deafness syndrome model using male mice. Peres C, Sellitto C, Nardin C, Putti S, Orsini T, Di Pietro C, Marazziti D, Vitiello A, Calistri A, Rigamonti M, Scavizzi F, Raspa M, Zonta F, Yang G, White TW, Mammano F. EBioMedicine. 2023 Mar;89:104453. doi:10.1016/j.ebiom.2023.104453.
2. Increased Hemichannel Activity Displayed by a Connexin43 Mutation Causing a Familial Connexinopathy Exhibiting Hypotrichosis with Follicular Keratosis and Hyperostosis. Crouthamel OE, Li L, Dilluvio MT, White TW. Int J Mol Sci. 2023 Jan 22;24(3):2222. doi:10.3390/ijms24032222.

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

Conference registration and financial aid opens Oct. 1

The 2024 National Conference will be held June 28-30 in Albuquerque, New Mexico. We hope you will join us for a weekend of connection, learning and fun!

Registration and financial aid applications will be accepted beginning Oct. 1. Register early for the best rates. Early bird registration is open through Nov. 30.

The conference location is the Hotel Albuquerque at Old Town. The conference room rate is \$189 per night**.

Additional conference details will be released as they are confirmed. Check firstskinfoundation.org/albuquerque-2024 for updates

2024 NATIONAL CONFERENCE

Join us in
Albuquerque, New Mexico!
June 28-30, 2024

Welcome reception June 27

firstskinfoundation.org/albuquerque-2024

** (plus taxes and fees)

