first
Foundation for Ichthyosis & Related Skin Types

Ichthyosis Focus

FALL 2025 • Volume 44, No. 2

A Journal for Friends of FIRST

LIFE ON HER TERMS

Dawn Trombley lives fully....8

Strong Foundation, Exciting Future

Hi FIRST Community,

As I write this, several important projects and new programs aimed at improving quality of life, treatment, and research for members of our community are making progress. These projects are the result of the strategic thinking of your Board of Directors and staff team, hours of volunteer time from our committees and Medical and Scientific Advisory Board, and the important funding coming from strong supporters like Beiersdorf, The Lennox Foundation and individuals like you.


Chris Boynton

Just to give you a little preview, work is nearly complete on a consensus statement on the treatment of newborns with ichthyosis. This document will provide definitive treatment guidance to physicians and other care team members, around the world, on how to best manage the new patient and family at the time of birth through the first few months of life.

The Board of Directors will also deliver the next strategic plan that prioritizes efforts and the investment of our resources to make the largest possible impact on this condition over the next five years. In addition to the strategic plan, the first FIRST Research Roadmap will make recommendations about how the organization can best direct its own research funds, assist the research of academic institutions, and play a role in encouraging, de-risking, and advancing research in the private sector. The purpose of all this is to accelerate the development of new treatments.

Work is just beginning on new tools for patients and medical professionals, including a toolkit for adults and one for those living with ichthyosis in the workplace. In addition, continuing education classes will develop skills and awareness around ichthyosis for dermatologists and non-dermatologists like family medicine doctors, pediatricians, and other healthcare professionals.

Thanks to all who support these efforts through their volunteer time and financial support!

Chris Boynton
CEO

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

Vol. 44, No. 2 **Fall 2025**

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

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first®
Foundation for Ichthyosis & Related Skin Types

Frequently Asked Questions About the National Conference

Q: When and where will the conference be held?

The conference will take place in Minneapolis, Minnesota, from June 26 to 28, 2026, at the Hyatt Regency Minneapolis, 1300 Nicollet Mall, Minneapolis, MN 55403. The hotel's phone number is 612-370-1234. The venue is fully accessible and located near dining, attractions, and transportation options.

Q: Who can attend?

The conference is open to individuals affected by ichthyosis or related skin types, families, caregivers, medical professionals, researchers, and anyone interested in learning more and supporting the ichthyosis community.

SAVE THE DATE FOR THE 2026 NATIONAL CONFERENCE!

The FIRST National Conference is heading to the City of Lakes!
We hope you'll join us in

Minneapolis, Minnesota, June 26-28, 2026

Visit firstskinfoundation.org/minneapolis-2026 for details.

Q: What is the purpose of the conference?

The National Conference is an opportunity to come together with people from across the country and the world to share experiences, learn from each other, hear the latest medical updates, and build lasting connections and friendships within the ichthyosis community.

Q: What kinds of sessions and activities will there be?

You can expect a mix of expert-led medical presentations, small-group discussions, hands-on workshops, and social activities for all ages. Topics include skin care advances, mental health, advocacy, and tips and tricks for caring for the skin. There are also many opportunities to connect with others with the same type of ichthyosis or in a similar stage of life.

Q: Will there be programs for children and teens?

Yes! We offer age-appropriate activities for children through the option of childcare (for ages up to 12). This opportunity provides a safe space for children to connect and have fun while their parents attend educational sessions and connect with other families. The teen program has not been finalized, but we will share it on our website and on social media when it is complete.

Q: How do I register?

Registration opens on October 1, 2025, through the FIRST website.

- Early Bird Registration: Opening Soon!
- Standard Registration: December 1, 2025, to April 30, 2026

We encourage early registration to secure your spot and take advantage of discounted rates.

Q: Is financial assistance available?

We understand that travel and registration costs can be challenging. FIRST hopes to offer some travel and registration scholarships to help families attend. If financial aid becomes available, all details will be provided on FIRST's website.

Q: What safety and accessibility measures are in place?

The Hyatt Regency Minneapolis is wheelchair accessible, and sessions are planned with the comfort of individuals with skin disorders in mind, including climate control. Additionally, if you have any dietary concerns, please indicate those concerns when you register.

Q: What if I've never attended before?

First-time attendees are warmly welcomed! We host an orientation session to help you get familiar with the schedule, meet the FIRST team and feel comfortable from the start.

Q: How can I get updates about the conference?

Sign up for FIRST's email list and follow our social media channels on Facebook and Instagram and check for updates on the website and in the semi-monthly eNews.

UFIRST Scholars Look Ahead to Academic Future

FIRST is proud to announce the scholarship recipients from the UFIRST Scholars Program.

These 13 students received a total of more than $50,000 in awards for their post-secondary education.

The UFIRST Program is partially funded by Beiersdorf, makers of Aquaphor and Eucerin.

Congratulations, scholars!

UFIRST
Scholarship Program

Amelia Bumpus

Ichthyosis vulgaris

Tennessee Technological University

Freshman pursuing an architecture major and early childhood development minor

Goals & Aspirations: My dream is to be able to help people make sure their needs are met in their homes or buildings. I hope that by pursuing these areas of study, I can improve the lives of people I meet.

Meghna Cacatte

Ichthyosis Vulgaris

George Washington University

Pursuing Executive Masters in Healthcare Administration

Goals & Aspirations: My long-term goal is to move back to India and set up and manage an intergenerational care facility with senior citizens and daycare-aged children to bring joy to both.

Portia Cina

Ichthyosis with confetti

Savannah College of Art & Design

Freshman pursuing 2D animation, concept design

Goals & Aspirations: I'm passionate about animation, storytelling and music, especially when it comes to representing diverse voices and normalizing rare conditions like ichthyosis. I want to create content that educates and inspires, giving kids with rare conditions - and underrepresented communities in general - someone to relate to.

Maxwell Claydon

Epidermolytic ichthyosis

University of Nevada Las Vegas

Junior pursuing music composition (vocal) and jazz piano minor

Goals & Aspirations: I want to have my own music as an artist and producer and be able to collaborate with other artists.

Mackenzie Dortch

ARCI lamellar ichthyosis

Indiana State University

Freshman pursuing art education

Goals & Aspirations: I plan to get a degree in art education. While teaching, I will also pursue my love of creating art.

Lauren Hamill

Harlequin ichthyosis

Salem State University

Senior pursuing early childhood education

Goals & Aspirations: Enrollment in graduate school with employment as a special education instructor upon graduation.

Brennan Lewis

Epidermolytic ichthyosis

Ohio State University, Mansfield

Sophomore pursuing a degree in English

Goals & Aspirations: I have freelanced for local news journals and sports in the area part-time. I also just got an LLC for my own business doing sports reporting. I want to work on sports writing in my local community the rest of my life.

Andrej Lozevski

ARCI-lamellar ichthyosis

University of North Carolina at Chapel Hill

Senior pursuing a degree in Biology

Goals & Aspirations: To become a physician with a focus on researching, treating and assisting patients who struggle daily with rare diseases and illnesses.

Devin Mong

X-linked ichthyosis

Westminster College

Junior pursuing a degree in Biology

Goals & Aspirations: I would like to attend graduate school and pursue a career in genetic/developmental research.

Abigail Schaffer

Netherton syndrome

University of Scranton

Freshman pursuing a degree in pre-med/biology

Goals & Aspirations: After completion of college, attend medical school to become a physician.

Hannah Soucek

ARCI-CIE

Iowa State University

Sophomore pursuing a degree in genetics

Goals & Aspirations: After my undergrad, I plan to pursue a PhD in a genetics or public health related field and pursue research related to rare genetic diseases, cancer, precision medicine, neurodegenerative disorders or other related areas.

Continued on page 7

Members share their perspectives on childhood, teen years, college and adulthood

Ellie W.

Shelby Township, MI

The day after my daughter Ellie was born the nurse gave her a bath and said, “I had a tough time getting all the vernix off.” I didn’t know then how those words were kicking off a new path in life: ichthyosis. By sound it seems like something icky, disgusting, gross. I hate the name ichthyosis, and I hate what it’s done to my daughter. But ichthyosis doesn’t define who our little Ellie is. Ellie is silly, funny, cuddly. She loves music and dancing. She likes going for walks, swinging, and playing with her brothers. She gets excited to draw and get out from behind the four walls of her home. She’s a typical two year old in those ways.

Read more of Ellie’s story at firstskinfoundation.org/ellie-w-2025.

Calvin K.

Pittsburgh, PA

Calvin is a teenager with ARCI-lamellar ichthyosis. He recently submitted an essay to convince his school community to donate money from a basketball fundraiser to FIRST.

Imagine waking up every day to a body that feels like it’s covered in scales. Even the simplest touch or movement could be painful, while also having to take half an hour out of the day to constantly moisturize one’s skin. But for some people diagnosed with ichthyosis, this is their reality. However, one organization helps fight the battle for those affected and that is the Foundation for Ichthyosis and Related Skin Types. The [school] community must donate the money raised from the Student-Faculty Basketball Game to The Foundation for Ichthyosis and Related Skin Types for its efforts to improve the lives of people affected by ichthyosis and to find cures for ichthyosis.

…

One way FIRST improves people’s lives is by holding biennial national conferences in rotating locations. At these conferences, guest speakers discuss their experiences and ways to combat ichthyosis. Many people say that these conferences are the first time they have seen someone with the same skin as them. One of these instances is a girl named Sarah who said “The first time I saw another person with skin that looked like mine was at my first FIRST National Conference in Chicago as a full grown and married adult. I cannot describe the feeling that washed over me and then stayed for the whole weekend – and beyond. I hadn’t even realized how alone I had felt, even surrounded by those who love me. FIRST gave me that and, ever since, I’ve tried to give back and be that person for someone else. FIRST improved this person’s life and countless others because they went to the conference and did not feel as alone.

Read the rest of Calvin’s essay at firstskinfoundation.org/news/member-essay-calvin-pittsburgh-pa.

Payton S.

Sarasota, FL

As I prepare to transition from my identity as a student at Marquette University into the next stage of life, I've begun to reflect more intentionally on the kind of person I want to become. My years here—shaped by the values of Catholic Jesuit education—have significantly influenced that vision. I no longer see my education as simply a collection of credits and classes, but as a formation process aimed at cultivating not only intellect but also character, compassion, and conscience.

A central theme at Marquette is cura personalis, or care for the whole person. That idea has reshaped how I view success and purpose. I don't want to become someone who chases achievement at the expense of self-awareness, integrity, or the needs of others. The Jesuit tradition calls us to be reflective, discerning, and committed to justice. It asks us to become the kind of people who not only think critically but also act compassionately.

Read more about Payton's hopes for her future at firstskinfoundation.org/payton-s-2025.

Cassandra K

Monroe County, TN

My name is Cassandra. I'm 34 and live in Monroe County, Tennessee. I have a wonderful husband and three beautiful kids. I have Darier disease which is a rare skin disease not many people know about. I started getting it when I was 10 years old. I'm originally from Ohio but I moved to Tennessee when I was 13 and went to a dermatologist here. I had a biopsy done to confirm that I had Darier disease.

Read more about Cassandra's experience at firstskinfoundation.org/cassandra-k-2025.

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Eileen Uthuppan

Chanarin-Dorfman syndrome

Valparaiso University

Senior pursuing physician assistant studies

Goals & Aspirations: Derm PA, blending my passion for science and art by growing SKINRARE magazine and working with skin health companies.

Addison Webber

Erythrokeratodermia variabilis

Spartansburg Methodist College

Freshman pursuing an associate of arts degree

Goals & Aspirations: To be a productive member of society, to be successful in my career, and to live a godly life, reflecting Jesus Christ.

UFIRST scholarships are proudly supported by Beiersdorf, makers of Aquaphor and Eucerin

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Life on Her Terms: Dawn Trombley Lives Fully as an Athlete, Teacher, Grandmother, and Affected Adult

Submitted by Anne Kaier

Dawn and her husband Joseph

Dawn Trombley, who has ARCI- lamellar ichthyosis, seems to have lived her life pretty much as she wants. She's an ebullient, outgoing woman with a lot of energy. Her own personality shines through her conversation. "I never met a wall I couldn't talk to," she says.

A true extrovert, Dawn has always enjoyed getting out there and mixing it up with people. She played volleyball and basketball in college. To this day, you can find her on the tennis court wearing an ice vest. Living in a small town near Austin, Texas, she tries to play in the early morning or inside, but she loves the game, loves feeling her body move in a sport she's good at.

Dawn was into athletics from an early age, swimming competitively as a kid. Her parents were in the mission field and she and her brothers grew up in "little bitty towns, all over," she remembers. That meant she had to make new friends often. Occasionally, that led to trouble. "Some parents didn't want me playing with their kids," she remembers.

In the 1950s and 60s when she was growing up, kids with ichthyosis, she says, generally weren't offered counseling or treatment for depression if they wanted it. Even at the Mayo Clinic, she reports, all the doctors could offer her was Vaseline and wrapping up with Saran Wrap. "I developed a thick skin," she laughs. "I'm just not easily bothered—and what am I gonna do? Change?"

> **As a special education teacher, Dawn "wanted kids with a variety of experiences, so they could learn from each other**

This practical, positive personality helped get her through some tough years in high school in Texas. She concentrated on academics and sports in public school and dated, but it wasn't easy. When she got to college at Kansas State, though, she was elected homecoming queen. "The farmers' kids liked me," she remembers, "because I didn't think I was better than they were."

She earned a bachelor's degree in education at Kansas State and, later, a master's in family counseling from the University of Kansas and a doctorate from the University

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Foundation for Ichthyosis & Related Skin Types

FY 2024 FIRST ANNUAL REPORT

About Us

OUR MISSION STATEMENT

Our mission is to improve lives and seek cures for those affected by ichthyosis and related skin types.

OUR LEADERSHIP

The Foundation for Ichthyosis and Related Skin Types (FIRST) is governed by a volunteer Board of Directors dedicated to supporting our mission with their time and talents. FIRST's Board members are active participants in the various activities of the Foundation and are representative of our diverse population.

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EMERITI

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Amy Paller, MS, MD, Chicago, IL,
Dave Scholl, PhD., Mason, OH
Mary L. Williams, MD, San Francisco, CA

Year in Review: A Snapshot of Impact Membership & Community Engagement

118 **New Members Joined:** A significant expansion of our community.

127 **People Used the Physician Referral Service**: Demonstrating the value of our support network.

Digital Reach & Impact

125,239

New Website Visitors: A strong indicator of growing interest and awareness.

Top 10 Most Visited Webpages:

1. FIRST
2. What is Ichthyosis
3. Harlequin
4. PPK
5. Bathing/ Exfoliation
6. SLS
7. Types
8. Collodion Baby
9. Itching
10. Albuquerque 2024

Education & Support

268 **Attendees at the National Conference:** A successful event for learning and networking.

81 **First-Time Attendees:** Highlighting increased engagement and reach.

119

Financial Aid Recipients: Providing crucial support to those in need.

$146,000

Awarded in Financial Aid: A testament to our commitment to accessibility and empowerment.

Research

TWO grants totaling **$100,000** were granted to Drs. Keith Choate and Amy Paller for their project "Employing spatial transciptomics to identify drivers of scaling and erythema in ichthyosis" and Dr. Cory Simpson titled "Evaluating novel therapeutic strategies for Darier disease".

How We Invest Your Money

We believe our donors deserve full transparency on how their money is allocated in pursuit of our mission to improve the lives and seek cures for those affected by ichthyosis and related skin types.

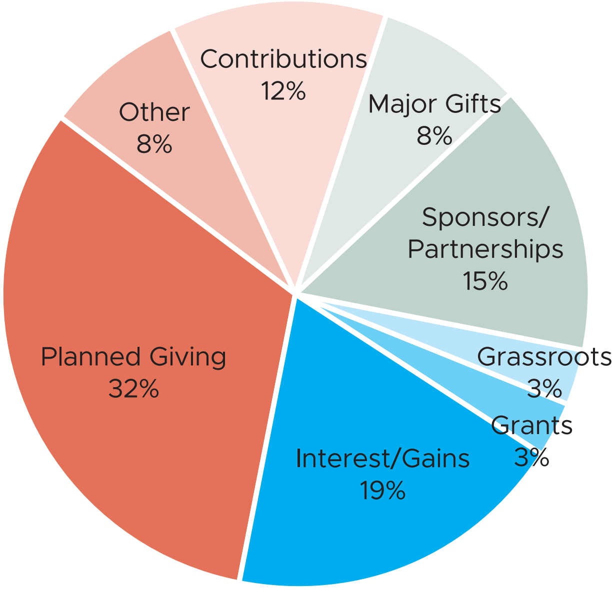
The following pages contain information from our audited financial statements to show how we raised and stewarded donor dollars in FY '23-'24 to empower affected individuals and their families.

REVENUE

Total revenue, gains and other support for the year ended September 30, 2024

	FY '23-'24
Contributions	$286,657
Major Gifts	$180,186
Sponsors/Partnerships	$356,488
Grassroots	$64,885
Grants	$75,950
Planned Giving	**$758,423**
Interest/Gains	$447,954
Other	$182,054
Total Support Services	$135,300
Total Support	**$2,362,597**

TOTAL SUPPORT



Impact of Your Donations

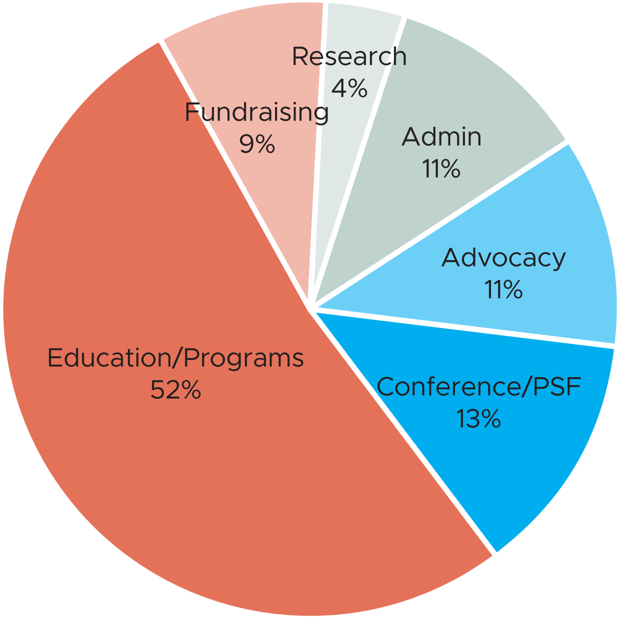
The following pages contain information from our audited financial statements to show how we raised and stewarded donor dollars in FY '23-'24 to empower affected individuals and their families.

EXPENSE ALLOCATION

Total Mission program and support expenses for the year ended September 30, 2024, were as follows:

	FY '23-'24
Advocacy	$124,847
Conference/PSF -- Conference/Meet-Up	$158,702
Education/Programs	$619,140
Research	$47,705
Total Mission Program Services	**$950,394**
Admin -- Management and General	$131,207
Fundraising	$100,911
Total Support Services	**$232,118**
Total Mission and Support Services	**$1,182,513**

YOUR IMPACT



Sustainability for Our Future

FIRST prudently invests reserves in a well-diversified portfolio to ensure consistent delivery of services regardless of the economy or investment environment. It is our goal to maintain one year of operating expenses in reserves.

NET ASSETS

	2024				
	Without Donor Restrictions	Temporarily Restricted	Permanently Restricted	Total	2023 Totals
Support and operating revenue					
Contributions	$1,218,343	$602,550	$28,520	$1,849,413	$676,431
Pass-Through (Movie)	$65,555			$65,555	
Net assets released from restrictions	$252,272	-$252,272	$0	$0	$0
Total support and revenue	**$1,536,170**	**$350,278**	**$28,520**	**$1,914,968**	**$676,431**
Operating expenses					
Program services	$950,395	$0	$0	$950,395	$620,790
Supporting Services					
Management and general	$131,207	$0	$0	$131,207	$73,153
Fundraising	$100,911	$0	$0	$100,911	$62,147
Total operating expenses	**$1,182,513**	**$0**	**$0**	**$1,182,513**	**$756,091**
Changes in net assets before non-operating revenues and expenses	$353,657	$350,278	$28,520	$732,455	-$79,660
Non-operating revenues and expenses					
Investment income (loss)	$447,955	$0	$0	$447,955	$207,988
Change in net assets	**$801,612**	**$350,278**	**$28,520**	**$1,180,410**	**$128,328**
Net assets, beginning of year	**$1,172,894**	**$643,915**	**$237,850**	**$2,054,659**	**$1,926,331**
Net assets, end of year	**$1,974,506**	**$994,193**	**$266,370**	**$3,235,069**	**$2,054,659**

2024 Donor Listing *(donations made during the 2024 calendar year)*

$100 - $499

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2024 Donor Listing *(donations made during the 2024 calendar year)*

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Continued from page 8

of La Verne in California. This education surely helped her talk skillfully with affected families and understand their concerns about how life as an affected person can be lived.

Dawn met her husband, Joseph Trombley, through friends. "We just clicked," she says. "He's a radiologist and is quieter than me," she remarks. "Maybe opposites do attract." They raised their five kids in California, where she taught special education. She taught a variety of kids—some blind, some deaf, and some with learning disabilities. But she also insisted on having non-disabled kids in her classroom, too.

Ever the athlete, she coached volleyball and swimming in a Catholic school as well. "I got all the kids who didn't get picked for the teams," she remembers. "I had to go to the priest and say, you gotta let my people go to bat the first time up—or they'll never get to bat." Once again, she was looking out for the underdog.

> **"Having ichthyosis has made me more empathetic, I think," she says, "less quick to judge. When I see someone different like me, my heart goes out to them."**

When Dawn talked to FIRST, she and her husband were traveling near the Artic Circle, where, in mid-summer, it was light all the time. Maybe that perpetual summer light mirrored her personality and her positive outlook.

When her kids were younger, she said, they used to be a bit protective of her. If they were out and someone in the street made a rude comment such as "do you know you're sunburned?" or "you could lead Santa's sleigh," her kids might ask Dawn if these comments hurt her feelings. "Not much," she'd tell them. "It's okay for people to learn about something new like ichthyosis." She says all her grandkids know she has "something genetic."

"Some people are born smart," she tells them. "Some people are born athletic. Some people are born with dry skin." Some people, like Dawn, are born with all three.

Another trait Dawn shaping Dawn's personality? Kindness.

Dawn (right) with her daughter and granddaughter

"Having ichthyosis has made me more empathetic, I think," she says, "less quick to judge. When I see someone different like me, my heart goes out to them."

Dawn didn't know about FIRST until late 2005. It was a good find. She's grateful for the group because it puts her in touch with other people and families affected by ichthyosis.

She's served as an Ambassador, talking mainly to parents and grandparents of kids with ichthyosis. "I ask them what their concerns and hesitations are," she says. She talks about her own experiences as a way to show parents that their child can grow up to have a satisfying life.

"I'm as normal as I can be," she says.

She's also, needless to say, a great advocate, especially as an affected adult, for FIRST.

About the Author: Anne Kaier

Kaier's memoir, *They Said I Couldn't Have a Love Life*, was a Finalist for the Association of Writers and Writing Programs' 2024 Sue William Silverman Prize. Her essays have appeared widely in venues such as *The New York Times, The Kenyon Review, 1966journal, Alaska Quarterly Review*, and the anthology *About Us: Essays from the Disability Series of The New York Times* for which she appeared on NPR's Radio Times. "Maple Lane" was mentioned on the list of notables in an edition of Best American Essays.

Her poetry collection, *How Can I Say It Was Not Enough?*, won the Propel Poetry Award. It's out from Nine Mile Press, 2025 (distributed by Syracuse University Press). Poems have appeared in several anthologies including the 2012 ALA Notable Book *Beauty is a Verb: An Anthology of Poetry, Poetics, and Disability*. She is a Virginia Center for Creative Arts Fellow and has served on a Fulbright screening committee for creative writers. She has an MA from the University of Oxford and a Ph.D. from Harvard University.

Make Your Giving Go Further

Gifts of non-cash assets are smart ways to benefit your 2025 tax return while making a powerful impact on FIRST's work.

Read on to learn about four types of gifts offering tax advantages. To make a gift, contact Denise Gass at dgass@firstskinfoundation.org or 504-434-5325.

Donor-advised funds (DAF)

For people who have a DAF, contributions offer an immediate tax deduction, plus:

- ✓ Simplified record-keeping with one tax receipt
- ✓ Ability to donate appreciated assets
- ✓ Option to remain anonymous
- ✓ No minimum distribution requirements

If you are interested in starting a DAF, check out the tool from GoFundMe (gofundme.com/c/givingfunds), which offers easy set up and distribution with zero fees.

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For people with IRAs who are 70 1/2 or older, these distributions:

- ✓ Count toward IRA required minimum distributions (RMDs)
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Cryptocurrency donations

For people who own crypto, giving digital assets allows these benefits:

- ✓ Receive tax deduction for the fair market value
- ✓ Avoid capital gains tax on appreciated crypto

Gifts of stock

For people who own appreciated stocks, bonds, or mutual funds, these gifts can provide double tax benefits:

- ✓ Avoid capital gains tax on appreciation
- ✓ Receive a tax deduction for the full market value

> **"Giving to FIRST through my donor-advised fund couldn't be easier, and knowing I'm helping improve lives for those with ichthyosis makes every contribution meaningful. Honestly, it's the easiest way I've ever supported a cause I care about. A few clicks online, and I'm helping fund the search for treatments that could transform the lives of people living with ichthyosis."**
>
> **- -Tejal Kamdar,**
> **Secretary of FIRST**
> **Board of Directors**
> **and parent of an**
> **affected child**

Participating in Ichthyosis Clinical Trials: a Vital Step Toward Brighter Futures

Research into rare diseases faces unique hurdles due to small patient populations and a lack of geographic concentration of potential participants. A recent increase in interest from pharmaceutical companies offers hope for those living with ichthyosis. The development of new and improved treatments hinges on the success of clinical trials, making patient participation a critical factor in shaping a brighter future for the ichthyosis community.

Living with a rare genetic skin condition like ichthyosis can be incredibly challenging. The daily reality of managing dry, thickened, and scaling skin, coupled with potential complications, significantly impacts the quality of life for individuals and their families. It also makes the decision to stop a regimen that works, even temporarily to participate in a trial, weightier.

Why participating in ichthyosis clinical trials matters

Clinical trials play a crucial role in advancing medical knowledge and developing new treatments for rare diseases like ichthyosis.

- **Access to novel therapies**: Clinical trials offer the opportunity to access investigational treatments that are not yet widely available, potentially offering a better treatment option than current standards of care.
- **Advancing medical knowledge**: Participation in trials helps researchers deepen their understanding of ichthyosis, its mechanisms, and the impact of potential treatments on symptoms and disease progression.
- **Contributing to future treatments**: The data and insights gathered from trials are invaluable in developing targeted therapies and interventions that can improve the lives of future generations with ichthyosis.
- **Improving diagnostic methods and severity measures**: Research driven by clinical trial data and participation in surveys, focus groups, photography, the Ichthyosis Registry, and biobanks can lead to improved diagnostic accuracy and increase access to genetic testing, enabling better understanding and management of the condition for individuals and their families.

What is FIRST doing?

In its soon-to-be-released new strategic plan and research roadmap to better treatments, FIRST specifically addresses the importance of creating a community that is knowledgeable about clinical trials and ready to participate. We are also supportive of the Ichthyosis Registry at Yale, which plays a significant role in identifying potential candidates with specific genes and contributes to research conducted by academic institutions and pharma.

Members of our Medical and Scientific Advisory Board including Drs. Amy Paller, Keith Choate and Joyce Tang are readying a very comprehensive grant application to the National Institutes of Health that includes “clinical trial readiness” for academic institutions and patient communities like ours.

Current research and hope on the horizon

Quoin Pharmaceuticals and Azitra are currently looking for patients living with Netherton syndrome to participate in their trials. There is also other research occurring at academic institutions that needs your help.

Overcoming challenges

Rare disease clinical trials face unique challenges in recruiting from limited patient populations. However, the commitment of FIRST, our MSAB, researchers, and pharmaceutical companies is to drive efforts to overcome these obstacles. Recruiting to fill the trials as quickly as possible goes a long way towards ensuring the trial can make it across the finish line.

Even trials that do not meet their clinical goals provide useful information, advancing science towards possible treatments. However, trials that do not meet their recruitment goals, set by the FDA, end prematurely and dramatically prolong the drug discovery process.

A call to action

Participating in ichthyosis clinical trials is a powerful way to accelerate the development of new treatments and improve the lives of individuals living with this condition. By embracing the opportunity to participate, you contribute directly to the advancement of medical knowledge and offer hope to those affected by ichthyosis. There simply are not enough people within this community to count on others to carry the load of participation. To advance new, better treatment options, we need more people to step forward for themselves and for others diagnosed in the future.

If you are not already participating in the Ichthyosis Registry at Yale, please look at how you can get involved at firstskinfoundation.org/ichthyosis-registry

If you are interested in learning more about clinical trials for ichthyosis, visit firstskinfoundation.org/clinical-trials.

Hikers Experience Epic Adventure, Make Enormous Impact

Submitted by Tracie Pretak, Former Board Member Co-Chair

Maybe it was a mid-life crisis moment. Maybe I needed a new goal to achieve. Maybe it was the enthusiasm that past hikers shared in their stories that inspired me. Whatever the reason, when I listened to a group of dads speaking at the 2024 National Conference about their experiences hiking the Grand Canyon, I literally became obsessed with doing it myself.

But when you take on a challenge of this magnitude – I mean seriously, this isn't just any hike, it's the Grand Canyon! – you are going to have moments of doubt. I returned home and started taking long hikes just to convince myself that this grandma could handle the long mileage.

I also began recruiting other hikers because I know that going through any challenge with someone who truly "gets it" makes the effort easier. Everyone needs a friend like Lyndsey. She's the kind who just says, "Sign me up!" Knowing I had her by my side pushed me through the rough times during training and the hike.

When I saw the canyon for the first time, I was in awe. My excitement turned into a massive adrenaline rush that made sleep nearly impossible. After being awake for 22 hours (due to travel and the time change), I only slept for one hour. Our hike began at 5 am. We were blessed with a beautiful sunrise. We took our time and took in the world around us. When we reached the bottom, we were rewarded with a cool swim in the green water of the Colorado River.

I ended up hiking with two of our affected members on the uphill climb. A partial trail closure forced us to take a connecting trail across a plateau in the direct sun. It was supposed to be 5 miles, but it had an unexpected detour making it a couple miles longer. The temperature in the canyon was nearly 100 degrees Fahrenheit. I was so impressed with our guides and the things they did to keep all of us safe – holding umbrellas over our affected hikers to give them a little shade, carrying extra liters of water to squirt down hikers who were getting hot, monitoring our heart rates and stopping to rest when needed, sprinting to rescue our lead group of hikers from an aggressive 6-foot rattlesnake, and saving the life of a hiker we met on the trail who tried this hike unprepared and was experiencing serious heat illness. The humanity and compassion I witnessed in all of us was heartwarming and good for my soul. Overcoming obstacles and working together has bonded us for life through our shared experiences on the trail.

We were still climbing at nightfall. When we reached the last rest house, we took a long break. We saw what few people will ever see – the

canyon lit up by a full moon. It was so beautiful and the perfect ending to an epic day!

Denise Gass, FIRST development consultant, hiked down to meet us and walk with us for the final steps of the journey. Having done this hike herself, she knew the encouragement that was needed at the end. On the ride back to camp, she asked all of us to give a word to describe the day – resilient, perseverance, amazing, accomplished, teamwork, adapting, awe-inspiring, together, and delightful. It was all of that and more.

I am so proud of our team of hikers and what we accomplished together. We didn't just complete a huge challenge, we also raised a bunch of awareness and nearly $40,000 for FIRST!

There is so much power in community and believing in yourself. I proved that I can do anything and that has given me freedom in ways I never thought were possible. I will do this hike again! And Lyndsey? As always, she said, "Sign me up!"

Researchers Propose New Names, Advance Understanding of Risks

A proposal for a new pathogenesis-guided classification for inherited epidermal differentiation disorders

Reference: Hernández-Martín Á, Paller AS, Sprecher E, et al. Br J Dermatol. Published online March 28, 2025. doi:10.1093/bjd/ljaf065

Nonsyndromic epidermal differentiation disorders: New classification and nomenclature based on disease-associated genes leading to targeted therapy. Akiyama M, Choate K, Hernandez-Martin A, et al. Br J Dermatol. Published online May 1, 2025. doi:10.1093/bjd/ljaf154

Syndromic epidermal differentiation disorders: New classification towards pathogenesis-based therapy. Paller AS, Teng J, Mazereeuw-Hautier J, et al. Br J Dermatol. Published online April 4, 2025. doi:10.1093/bjd/ljaf123

Riddhi Patel

Predoctoral research fellow at Northwestern University

Amy Paller, MD

Northwestern University FIRST MSAB Chair

Palmoplantar epidermal differentiation disorders: a new classification towards pathogenesis-based therapy. Sprecher E, Ishida-Yamamoto A, Schwartz J, et al. Br J Dermatol. Published online March 19, 2025. doi:10.1093/bjd/ljaf054

Four landmark articles have been published this year in the British Journal of Dermatology, in which top experts in ichthyosis from around the world propose a new way to name the various forms of ichthyosis and related skin types. The group working on the new classification system, called the Reclassifying Epidermal Differentiation Disorders Initiative (REDDI), included 15 expert physician-scientists and two affected individuals. Over more than two years, the team met virtually and in person to achieve these landmark papers.

The new names are meant to be more accurate in their grouping than the old names of "disorders of cornification" or "disorders of keratinization" for the entire group. In addition, the new names get rid of terms that have been hurtful to people with ichthyosis, like "vulgar" in ichthyosis vulgaris, "harlequin" referring to a clown, "hystrix" meaning porcupine, or even "ichthyosis" itself which means fish.

The new naming system discards the names of people whose names tell us nothing about the skin problem (for example, Netherton syndrome). Historically, these conditions also used names based on what the skin looked like, which led to confusing or outdated terms.

The new classification follows trends in naming of genetic issues by including the underlying gene that is affected. It also groups the disorders based on function, with the goal to find new treatments based on understanding the gene affected and how that gene change causes disease in the skin.

The authors replace the word "ichthyosis" with "epidermal differentiation disorder" or EDD, which truly encompasses all of the different types. There are three main categories:

- Nonsyndromic epidermal differentiation disorders (nEDDs) affecting just the skin, hair, nails, and/or sweat glands
- Syndromic epidermal differentiation disorders (sEDDs) affecting the skin and its structure plus at least one other body system (most often the nervous system or eyes)
- Palmoplantar epidermal differentiation disorders (pEDDs) affecting mainly the palms and soles

Some of the new names are shorter (like FLG-nEDD, formerly ichthyosis vulgaris), while other names will be longer or harder to remember. Other ones that affect many in FIRST with their new names are: STS-sEDD (X-linked ichthyosis); TGM1-nEDD (most with lamellar ichthyosis); ALOX12B-nEDD (most common form of CIE); ABCA12-nEDD (harlequin ichthyosis); KRT9-pEDD (epidermolytic palmoplantar keratoderma).

These names will be more scientific to help doctors understand what causes the disease at a molecular and functional level. By classifying these conditions by the gene that is affected, doctors will be better able to counsel families about prognosis and develop more precise treatments.

The articles include an introduction to this new concept and one article on each of the three different subtypes. Each article has detailed tables with old names and new names, key features and detailed descriptions of each of the almost 150 subtypes now listed, and many photographic images to help practitioners recognize these EDDs and plan the best treatment. As such, these articles will be the new encyclopedia for ichthyosis and other similar disorders.

We recommend that you tell your doctor to keep each of these handy as a reference in their clinics. Clearly, the old names will be around for a while as each person has a form with a label that becomes part of one's identity. The same is true for FIRST itself as an organization.

But during the next decade, these names will likely transition, as getting a test that tells the gene causing EDDs becomes the norm, and new individuals born with EDDs are told the new names rather than "ichthyosis". The FIRST-associated registry has long offered free testing to find genes for FIRST members – and all are encouraged to take advantage of this offer, which simply involves providing consent to registry personnel and sending a sample of saliva. Taken together, this is an opportunity to be part of a transition that focuses on what causes the skin scaling, thickening, and redness and that will accelerate discoveries towards treatment and cure.

Bullying in children with congenital ichthyosis.

Reference: Rustad AM, Soltani H, Yang LJ, ...Paller AS. Pediatr Dermatol. 2025;42(3):499-505. doi:10.1111/pde.15920

Review: Most of the rare forms of ichthyosis have scaling and often red skin from birth that involve the whole body and are easy to see. In addition, affected children often show behaviors that make them seem different, such as scratching and reluctance to join in sports because of issues with sweating. These differences make children with ichthyosis at increased risk of bullying. Indeed, bullying is a frequent concern for both affected individuals and their families as discussed at support meetings. However, research on the subject has been lacking. This study, performed at Northwestern and the Ann and Robert H. Lurie Children's Hospital of Chicago, surveyed children with ichthyosis and their caregivers about the children's bullying experiences. FIRST members actively participated in this survey.

Forty-nine caregivers and children completed the survey, which asked questions about bullying, stigma, and quality of life. There was also an open-ended question in which caregivers and children could include other comments about their experiences. Thirty-three percent of the children with ichthyosis who completed the survey had experienced moderate bullying. This is higher than the 15-19% in the general United States high school population between 2021 and 2023. Of all affected body parts, only facial involvement was associated with significantly increased bullying, as reported by children. Bullying was done by other children in 80.7% of cases, but could be from adults, such as teachers or even strangers. Children experienced bullying by being excluded, called names, and asked rude questions. Children reported feeling sad, embarrassed, worried about the future, and "less human."

Caregivers suggested strategies to manage negative attention, including encouraging the child to avoid thinking about ichthyosis as shameful or a secret. They also suggested avoiding appearance-based comments at home and instead focusing on action-based and character-based comments. Strategies to address and reduce bullying in children with ichthyosis proposed in this study were increasing awareness and education related to ichthyosis in the school setting to build a strong support network. Screening for bullying during health appointments was encouraged for early identification of bullying and to identify if the child worried about feeling different. Children's books featuring characters with visible differences can act as anti-bullying tools for families and are listed online through the FIRST website (firstskinfoundation.org/bullying).

The importance of cardiac screening in X-linked ichthyosis - a plea.

Davies W. Clin Exp Dermatol. Published online May 16, 2025. doi:10.1093/ced/llaf221

This author from Cardiff University in the United Kingdom looked at a dataset of almost half a million participants from the UK and found that adults with X-linked ichthyosis had a four times increased risk of atrial fibrillation/flutter, in which the upper chambers of the heart have a fast or irregular beat. When the author then surveyed boys and men with X-linked ichthyosis and adult females who were carriers of the mutation, they found a higher prevalence of parent- and self-reported heart rhythm abnormalities. The cause of increased heart rhythm abnormality risk is unclear and still needs further investigation. This paper recommends that asking about cardiac problems should be part of the routine clinic visit. If palpitations, chest pain, breathlessness, extreme anxiety, dizziness or fainting, and fatigue are frequent or continuous, affected individuals should be evaluated by a cardiologist.

Increased risk of anxiety and coping strategies in patients with selected genodermatoses with cornification disruption.

Fryze M, Mlak R, Kulbaka A, Wertheim-Tysarowska K, Matosiuk D, Pietrzak A. Sci Rep. 2025;15(1):14013. Published 2025 Apr 23. doi:10.1038/s41598-025-98535-6

This study from the Medical University of Lublin in Poland compared anxiety levels between adults with ichthyosis or palmoplantar keratoderma with unaffected adults using surveys and tools that assessed emotional state and personality traits. High or moderate levels of trait anxiety were four times more common in those with ichthyosis or palmoplantar keratoderma when compared to the unaffected group. The authors highlighted the importance of stress-coping strategies in these individuals, noting that avoidance as a coping mechanism (trying to distance oneself from problems and avoiding social interactions) was a common and ineffective strategy. Doctors need to pay attention to the patient's mental health state and refer to professional psychological/psychiatric therapy to improve their ability to cope, which has an impact on one's quality of life.

Congenital ichthyosis is associated with cutaneous infections in a case-control study of 2260 patients.

Curtis KL, Zeldin S, Lipner SR. Clin Exp Dermatol. 2025;50(2):429-431. doi:10.1093/ced/llae382

This study review from Weill Cornell Medicine used the All of Us Database through the National Institutes of Health to analyze the risk of developing skin infections in 226 people with congenital ichthyosis and 2034 unaffected controls. The study found a fivefold increase in the risk of developing fungal nail infection, a threefold risk of body fungal infection, and a twofold increase in the risk of a fungal foot infection in individuals with congenital ichthyosis. Participants with congenital ichthyosis also had a threefold increased risk of warts. Skin infections such as fungal infections and warts, which show localized scaling and thickened skin, may be hard to identify in patients with congenital ichthyosis because of the associated scaling and thickening of skin. The authors suggest that a poor skin barrier in ichthyosis could increase the potential penetration of the fungi and viruses that cause fungal infection and warts, respectively, and recommend that fungal infection be considered in those who do not respond to ichthyosis treatments.

Biologics in congenital ichthyosis: are they effective? Mazereeuw-Hautier J, Granier Tournier C, Hernandez-Martin A, … Paller AS. Br J Dermatol. 2025;192(2):327-334. doi:10.1093/bjd/ljae420

During the last decade, numerous medications called biologics have become available to treat two common red, scaling skin disorders, eczema and psoriasis. Based on studies from skin biopsies and tape strips of people with ichthyosis, including many members of FIRST, evidence emerged that there may be shifts in the immune system of skin that could be treated by these biologics. Indeed, at least 30 papers have shown the benefit of a biologic for a patient with ichthyosis. However, it is common that only those with positive results are reported. In this multicenter international study, the authors analyzed change in ichthyosis severity in patients with congenital ichthyosis who were treated with at least one biologic for 3 months. More than half of the 98 patients treated with a biologic (average age 20 years) had a form that was associated with very red skin (Netherton syndrome or congenital ichthyosiform erythroderma (CIE)) and severe or very severe disease. Overall, 45 patients (46%) responded to treatment to some extent; however, only 18 (18%) had a moderate to excellent response and all had forms with very red skin. There was no one biologic drug or clinical sign with improvement (that is, skin redness, scaling, or itch) that seemed superior to others. None of the patients with lamellar ichthyosis or epidermolytic ichthyosis had a good response to biologics. The main takeaway of this study is that biologics may lead to improvement in a subset of patients with ichthyosis, particularly in those with redder skin. Further studies are needed to evaluate the effects of biologics in specific forms of congenital ichthyosis.

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Captain's Council Featured Member

John Schoendorf (left) poses with Holly Friddle and Angelo Mantione at the 2010 FIRST National Conference

Captain's Council recognizes donors who decide to make a gift to FIRST in their will. If you would like to create a free will that includes a gift to FIRST, you can visit www.freewill.com/first It's an easy way to protect your loved ones and make sure future generations have access to the same connection and care you've found here. If you have already included First in your will, please email dgass@firstskinfoundation.org so we can add you to Captain's Council.

John Schoendorf
Forensic Accountant
Vero Beach, Florida
Epidermolytic Ichthyosis

What does FIRST mean to you? Everything, both as treasurer for seven years helping transition FIRST to a new business model and being part of an organization that continues to provide tremendous value to affected individuals and families.

Why did you decide to include FIRST in your legacy planning? I believe in paying forward. I have been blessed with good fortune and need to share with others in a meaningful way.

Do you have a favorite memory of FIRST? I just love to see the upbeat attitude of the young ones at family conferences, especially with the fun dancing and show competitions.

Captain's Council is a FIRST legacy society recognizing those who are making a gift to FIRST in their will.