



TOGETHER AGAIN

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This issue celebrates the National Conference in Rhode Island in June! It was wonderful to meet, in person and face-to-face, for the first time in many years! The energy at the event and the feeling of connectivity was overwhelming. It was obvious that an in-person event was much needed by our community.



Mark Evans



Tracie Pretak

We know how much this event means to our members because of the opportunity to connect, re-connect, provide and receive support, obtain helpful information, and meet with doctors who have experience treating patients with ichthyosis. We know our own families have greatly enjoyed past conferences, as well as this one. We send a big thanks to our staff, physicians and volunteers who made it happen!

At the conference, we announced that Beth Hampshire, a long-time FIRST member with ARCI-lamellar ichthyosis, will take over as Board chair when our term concludes at the end of the year. We are excited for the ways Beth will drive our mission forward. Get to know more about Beth on the next page.

Timber Pharmaceuticals has advanced their clinical trial to a Phase 3 study to investigate the efficacy and safety of a topical isotretinoin treatment (read more on p. 12). The results of the first two phases are promising. Also on the research front, we continue to receive a robust volume of research grant requests that are being evaluated by a committee of experts.

Regarding fundraising, grassroots events have always been important to us. We recently had our first baseball game fundraiser in quite a while (see pictures on p. 4). These grassroots events provide a relaxed and fun atmosphere for our members, friends, and family to connect and raise awareness. If you are interested in hosting, please reach out to Denise Gass (dgass@firstskinfoundation.org). We continue to hold a wide variety of virtual events that offer the opportunity for you to learn and connect digitally.

As we prepare to step aside as Board Chairs, we are proud to know that FIRST is well-positioned for the future with a strong financial outlook, dedicated staff members, an engaged Medical and Scientific Advisory Board (MSAB), and our committed volunteer Board of Directors, who all collaborate to accomplish FIRST's mission to improve lives and seek cures for those affected by ichthyosis and related skin types.

Mark Evans & Tracie Pretak
Co-Chairs, FIRST Board of Directors

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

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

Phone: 215.997.9400
Toll-free: 800.545.3286
Fax: 215.997.9403

Email: info@firstskinfoundation.org
Web: www.firstskinfoundation.org

CEO

Chris Boynton

Editor

Kimberly Cole

Medical Editor

Amy Paller, MD

Editorial Assistants

Bailey Pretak
Abby Evans

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Hampshire to Continue Giving Back as New Board Chair

Giving back has been a common thread through Beth Hampshire's involvement with FIRST.

As a child, FIRST National Conferences were central to Beth's life. As an adult, she leaned on FIRST for resources and to connect with other women. In 2018, wanting to ensure that FIRST provided value to affected members of all life stages, Beth moved from member to volunteer leader.

Beth joined the Board of Directors with an interest in serving affected members of the ichthyosis community, by giving them a voice in the organization and inspiring younger members to see themselves in positions of leadership. She will assume the role of Board Chair in January.

Beth recalls recognizing her impact as a role model after serving as a counselor at Camp Discovery, a summer camp for children with skin conditions. The young campers dreamed of getting married and having a family – things

Beth, who is affected with ARCI-lamellar ichthyosis, was realizing in her own life. "I saw the importance of engaging young adults in our community and giving them meaningful ways to give back," she said. "Whether this is through mentoring, connecting at the National Conference, or being a friend on social media."

During her term as Chair, Beth plans to encourage research and participation in clinical trials. Over the past five years, Beth has participated in many clinical trials herself. She believes this is an important way members can give back to the community, potentially, helping better the lives of those affected with ichthyosis in the future.

She also hopes to expand the membership and involvement of affected individuals. She will look for new ways that FIRST can engage and connect all members.

Beth is married with three children, and works as deputy controller for the



Beth Hampshire

city of Fishers, Indiana She has served on the Board since 2018, and currently holds the position of 2nd Vice President. She also leads the Strategic Planning Committee and Board Development Committee.



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Weary Fund Provides Cool Relief

FIRST is excited to announce The Weary Fund, which began from a generous donation from Terry and Robert Melton to supply air conditioner units for those with ichthyosis who do not have access to them.

Terry, an affected adult with epidermolytic ichthyosis, has been a FIRST member for many years. She and her parents, Dr. Peyton and Janet Weary, have all served on the Board of Directors. Dr. Terry was an academic dermatologist at the University of Virginia for over 50 years. Terry wished to create The Weary Fund in honor of her parents and their commitment to FIRST.

Terry recalls difficult experiences of overheating and being unable

to cool down. She remembers her father carrying a window air conditioner up several flights of stairs into her college dorm on many occasions in North Carolina.

After college, Terry became a physician's assistant and earned her PhD in genetics, working in forensics. Now retired, she enjoys volunteering for local organizations.

Terry felt motivated to start the fund through her work with the environment and understanding of and concern for climate change. For those with ichthyosis who cannot regulate their body temperatures, a warming world is a worrisome prospect. She hopes this fund makes the world accessible, safer, and cooler for those with ichthyosis



who do not have access to air conditioning.

The Weary Fund will be a part of the FIRST Aid skin care program. Applications will be available in the spring and forms will be available on FIRST's financial aid page, firstskinfoundation.org/financial-aid.

FIRST Night Out at the Dodgers Game

The Coates family once again hosted their LA Dodgers game for FIRST. There were 100 people in attendance on a beautiful night in July. Ticket sale profits and additional donations added up to more than \$1,700 to help advance FIRST's mission. Thank you, Coates family and supporters!



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FIRST Partners with Climb for Hope in Grand Canyon Crossing

Participants at the recent National Conference learned about purpose-driven adventure from keynote speaker Andrew Buerger. When his sister was diagnosed with breast cancer, Andy got the idea to raise money for research by taking people up mountains. The challenge of climbing a big mountain is an apt metaphor for the struggles of dealing with a disease or chronic condition. In 2018, Grand Canyon crossings were added to the list of endurance expeditions offered through his Climb for Hope organization and are now some of the most popular trips.

FIRST is excited to offer a Grand Canyon crossing in May 2023 to benefit ichthyosis research. This trip is certain to also bring awareness and inspiration in the form of stories of perseverance over many miles in challenging conditions.

Participants can choose one of two climb options. The double crossing is a challenging, bucket-list item

for outdoor fitness enthusiasts. Covering over 50 miles, the trip begins with a descent into the canyon from the South Rim. After a few miles along the canyon floor, the hike heads up over 4,000 feet to the North Rim. The return the next day, covers the same number of miles with slightly less elevation gain. Those seeking a less intense experience can choose to hike only the second day.

The expedition is not technical but requires excellent physical conditioning. Climb for Hope will provide an 18-week training program similar to a marathon training schedule. The trip is at a higher elevation, temperatures can range from the 70s to 100s, and the distance covered over two days is long.

Spectacular views, a great physical achievement and the satisfaction gained from raising much needed funds for FIRST while serving as a role model for our community will reward participants.



Because of the intense logistics required for this expedition, a **\$3,000 fundraising minimum is required of all participants.** Sponsorship from friends, families, co-workers and companies is encouraged. To learn more details, please contact Chris Wassel at cwassel@firstskinfoundation.org.

Putting on a Show for FIRST

Bailey Pretak has a passion for everything Broadway. For the second year in a row, she celebrated her birthday with a Broadway-themed event to benefit FIRST! She gathered some of her closest theater friends, as well as some of her mother's music students, and together they performed their favorite songs from Broadway shows.

Tables were decorated with taxis, NYC street names, and Playbills, and a beautiful backdrop of the NYC skyline decorated the stage. Attendees could order food from the venue and bid on several different auction baskets like Girls' Night Out, Kids' Summer Fun, Movie Night, and a NYC Theme. The event raised over \$2,100!



Let's give a standing ovation to Bailey for using something she's passionate about to create a great fundraiser. We can't wait to hear how your birthday bash goes next year!



Paller Selected for Hall of Fame Award

Dr. Amy Paller received the Society for Pediatric Dermatology's first Hall of Fame Award earlier this year. The criteria for this honor include being an SPD member for at least 30 years and having made substantial contributions to the organization and field of pediatric dermatology.

Dr. Paller is the Walter J. Hamlin Professor and Chair of Dermatology, Professor of Pediatrics, and Principal Investigator of the NIH-funded Skin Biology and Diseases Resource-based Center at Northwestern University in Illinois. She serves on FIRST's Medical and Scientific Advisory Board and is an emeritus member of the Board of Directors.

An author of almost 600 peer-reviewed publications, Dr. Paller has also edited several major textbooks in dermatology. She is an NIH-funded investigator in

both her laboratory-based and clinical research. She has directed the Pediatric Dermatology Clinical Trials Unit at Northwestern/ Lurie Children's for the past 30 years and has been the lead investigator on several landmark trials in pediatric skin disease, including genetic skin disorders like ichthyosis.

Her laboratory focuses on cell-cell communication in inflammatory skin diseases and impaired wound healing, as well as on topically delivered gene regulation through nanotherapy. She has defined skin and blood biomarkers for children with atopic dermatitis and ichthyosis towards pathogenesis-directed therapy and has developed new patient-related outcomes scales for itch and stigma in children.

Dr. Paller has served in leadership roles for several dermatology



Amy Paller

associations, as well as mentored hundreds of students, residents, fellows, and junior faculty. Her greatest pleasure is to watch young people gain knowledge, get excited about concepts and discoveries, and advance their own careers.

In September, Dr. Paller received an additional honor -- the 2022 Tripartite Legacy Faculty Prize in Translational Science and Education. This award honors a Northwestern University faculty member who has demonstrated excellence in research that emphasizes translational approaches, teaching and mentoring, and leadership.

Possible Skin Cancer Risk for Certain Skin Conditions

Skin cancer is the most common cancer in the United States, with approximately one in five developing skin cancer in their life. When skin cancer is detected early, it is often easily treated.

Certain characteristics increase one's risk for skin cancer, including having a lighter skin tone, skin that sunburns easily, and a personal or family history of skin cancer. Some skin conditions may also increase risk.

Individuals with disorders of cornification including Netherton syndrome, KID syndrome, Huriez syndrome, RSPO1-associated syndrome, CAPOK syndrome and MAUIE syndrome (a subtype of ichthyosis with confetti) should



get more frequent skin cancer screenings. For these subtypes, skin cancer has been reported more frequently and at an earlier age.

Among others with ichthyosis, the risk of skin cancer is not known and seems to be similar to the general population. The frequency of skin cancer in disorders of cornification, however, is an area that needs further study.

A dermatologist is trained to identify and treat all types of skin cancer. It is

important to ask your dermatologist how to check your own skin for signs of skin cancer and if you are due for a full body skin check. A general rule of thumb is if you notice a spot that is different from others, that changes, itches or bleeds, you should make an appointment to see a dermatologist.

The American Academy of Dermatology recommends these daily sun protection strategies:

- Stay in the shade when possible.
- Wear protective clothing like long sleeves, pants, and a wide-brimmed hat.
- Apply sunscreen with broad spectrum coverage, SPF 30 or higher, and water resistance.

Shine Your Light: Conference Inspires and Connects



In late June, 364 members of the FIRST community gathered for the 2022 National Conference in Providence, RI.

The attendees included:

- 63 families with affected children
- 57 affected adults
- 29 medical professionals and researchers

More than 56 families were attending their first conference.

The three-day event included motivational keynote speakers, including Hunter Steinitz, a 27-year-old FIRST member affected with harlequin ichthyosis. Hunter spoke about the power of telling your story, for yourself, for others affected by ichthyosis, and for the world at large.

“The way that we change how the world sees us is by boldly telling our stories, sharing our experiences,” she said. “Because if we don’t, no one outside of this family will know what it is like.”



Hunter Steinitz sparked while delivering her keynote address

Attendees broke into tracks or interest groups to learn more about products and care tips, types of ichthyosis, genetics, clinical trials and other topics. Groups for affected adults, young adults, mothers, fathers and grandparents offered opportunity to connect with and seek support from others.

In a spotlight session, Dr. Len Milstone shared about the intersection of his decades of work in ichthyosis research and the growth of FIRST.

While working on early studies of retinoids in the 1970s, Milstone noted they were breaking ground in an unexpected way – the studies brought together, for the first time, physicians,

patients and families connected by ichthyosis. For physicians, “this meant that learning and teaching about ichthyosis moved from textbooks to a living reality,” said Milstone. “Just as importantly, people with ichthyosis began meeting others who shared their practical, social and medical concerns.”

This was the catalyst for the formation of the Ichthyosis Foundation – the organization now known as FIRST. Dr. Milstone has been a consistent presence from those early days to today, where he continues to serve on the Medical and Scientific Advisory Board and as an emeritus member of the Board of Directors.

Throughout the conference, it was apparent that the lights of individuals glow brightest when they shine together.



Dr. Len Milstone spoke about highlights of his research career

Thank you to our Co-Chairs Mark Evans and Tracie Pretak for their tremendous service to FIRST over the past two years! Through COVID cancelations and new staff leadership, they offered a steady hand and worked to build a sturdy foundation for future growth. As their term concludes, please join us in thanking them for their many contributions to FIRST!







Awards Given for Grassroots Fundraising and Volunteerism

Members who lead fundraisers and volunteer reflect the very core of what we do at FIRST—come together to make a difference, in any way we can.

At the National Conference, award winners were announced in three categories. There were four years of awards to share, due to the cancellation of the 2020 conference.

FIRST is grateful for the efforts of these members.

Bernstiel Award for outstanding grassroots fundraising

- 2018: Denise Benedetto and Brooke Saccente, who raised \$13,745 at a Mets game
- 2019: Jolie and Sean Cina, who raised \$5,320 at a wine tasting
- 2020: Michelle Iott, who raised \$3,205 at a pumpkin painting party
- 2021: Tejal Kamdar and Jason Snyder, who raised \$19,211 with an online birthday campaign

Bernstiel Junior Award for members under the age of 18:

- 2018: Cailyn McCullers, who raised \$1,836 with a lemonade stand

- 2019: Ashlynn Biggs, who raised \$860 with a Facebook birthday fundraiser
- 2020: Evan Mayone who raised \$560 with a Facebook fundraiser
- 2021: Evan Mayone who raised \$778 with a Facebook fundraiser

Volunteer of the Year Award for members who have made the most compelling contribution to FIRST:

- 2018: Jennifer See, for her work at the conference and all she has done to demystify ichthyosis for so many families
- 2019: The Release the Butterfly team, for innovation in fundraising and outstanding advocacy as they created, produced, managed, and toured a musical production
- 2020: Lowell Wright, for remarkable effort in coordinating FIRST diversity, equity and inclusion advancements
- 2021: Jennie Wilklow-Riley for outstanding advocacy, innovative grassroots fundraising, and community leadership



Volunteer Spotlight: Harlequin Diva

After her daughter, Anna, was born in 2017, Jennie Wilklow-Riley quickly recognized the expense of caring for a child with ichthyosis.

Concerned that other families would not be able to afford necessary and potentially life-altering treatments, Wilklow-Riley was determined to help. Soon after, the Harlequin Diva Foundation was created.

To date, the foundation has raised more than \$50,000 to help individuals and families cover the costs of specialized bathing equipment, air conditioning units, cooling vests and lotions.

At this year's conference, Wilklow-Riley offered more than \$16,000 worth of Aquaphor and product donations to attendees. She also distributed dozens of cooling vests and other items to assist those with ichthyosis.

Learn more about the fund at HQDdonations.com.

Great Benefit and a Great Bargain

Attending a FIRST National Conference is a wonderful experience. Between the clinical visits, informative sessions, motivational speakers and opportunities to connect with others in the same boat, the conference is packed with great benefits.

But did you know it is also a great bargain? FIRST heavily subsidizes the cost of attendance to make the conference accessible to all. Financial aid is available to all that apply but even those who don't receive aid are getting a value.

Conferences are an expensive proposition. Food, room rental, logistics, entertainment, presentation costs and extra staffing like childcare add up quickly. Below is a quick breakdown of the most recent conference in Rhode Island.

Total Conference Cost	\$250,000
Number of Registrants	313
Total Cost per Registrant	\$798
Average Fee Paid per Registrant	\$225
Savings per Person	\$573

FIRST's ability to keep this level of subsidy is dependent on support from the membership. This is particularly true now when the impact of inflation means our dollars buy less. Please consider supporting FIRST by becoming a donor member today.

Caring for the Full Rainbow of Ichthyosis Skin Colors

This update is provided as part of FIRST's commitment to Diversity, Equity and Inclusion, and meaningful representation of members of all races and skin colors.

Thanks to input from volunteers, we have many efforts to report.

- **Photos:** We are expanding our photo collection so we can have a library of lifestyle and medical images of diverse skin tones. There are 28+ types of ichthyosis and related skin types, and they all look different depending on skin tone. It is FIRST's mission to educate the medical community for more accurate diagnoses and to depict an accurate reflection of everyone living with ichthyosis. Do you have photos to share? Please email Denise at dgass@firstskinfoundation.org.
- **Ichthyosis Registry:** FIRST and Yale University researchers will determine the racial breakdown of those in the registry. If proportions do not match population data, FIRST will work to improve representation, including exploring race-correlated obstacles to accessing ichthyosis treatment.
- **Academic Study:** Researchers at Yale are writing a research paper on ichthyosis and skin of color. This will explore the appearance of ichthyosis across melanin levels in a committed, academic way. It will utilize the new Eumelanin scores (see inset). FIRST will publicize this effort and make it available on the website. It will also be indexed



Lowell Wright, pictured with CEO Christopher Boynton, was honored for his work to advance FIRST's DEI efforts

- in PubMed as a tool for medical education.
- **Conference:** Every conference element was reviewed through a DEI lens, and reviews and feedback will allow for continued improvement. FIRST recalls the vivid, negative experiences Black members have shared and we must improve.
- FIRST offered more financial aid available than ever before, a leap forward to help with access for all.
- Every speaker/moderator received a DEI training and agreed to create space for attendees of every color, orientation, religion, and knowledge level.
- FIRST reviewed all speaker and moderator positions to ensure as much diversity as possible.
- FIRST added DEI criteria to evaluation of cities for 2023 Patient Support Forums and the 2024 National conference.
- **Product Listing:** FIRST recognizes not all ichthyosis products are good for all skin tones. This information needs to be updated and ready for sharing. Do you have suggestions for products that work for Black hair and non-white skin tones? Which products do not work? Email Denise at dgass@firstskinfoundation.org.

Researchers Propose New Scale for Describing Skin Color

An objective scale for identifying skin color in medical research will enable health professionals and researchers to describe skin color more accurately, compare studies and identify underrepresentation in research.

The Eumelanin Human Skin Colour Scale was developed by members of the British Association of Dermatologists' Lexicon Group. The

scale assigns skin color to one of five categories based on the amount of light reflected from the skin.

"For a long time, healthcare professionals and researchers have been missing a simple tool in their toolboxes: an evidence-based approach to describing skin color," said Dr. Ophelia Dadzie, Chair of the British Association of Dermatologists' Lexicon Group.

Skin color, rather than ethnicity, may be an important variable in research. This scale offers a standard approach for recording skin color, allowing researchers to analyze how diseases affect patients of different skin tones, and combat underrepresentation in research.

Clinical Trial Opportunities

Phase 3 Trial for Isotretinoin Ointment

Many dermatologists and affected individuals are familiar with oral isotretinoin. By formulating isotretinoin into a topical ointment, it may be possible to reduce systemic absorption, potentially allowing for chronic use over larger areas of the body. Researchers from Timber Pharmaceuticals recently announced the start of a Phase 3 trial.

Study participants must be 6 years of age or older and have either autosomal recessive congenital ichthyosis (ARCI) or recessive X-linked ichthyosis. Participation in this study will last approximately 12 to 24 weeks and require six to nine visits to the study center. This is a randomized, double-blind trial, meaning that some participants will receive ointment that does not contain the test formulation for the first 12 weeks.

Learn more at firstskinfoundation.org/tmb-001-ascend.

Growth and Development Study

A study at Yale University will investigate linkages between ichthyosis and growth delay. Results will help families and physicians predict how different types of ichthyosis may affect growth and better determine whether early interventions, such as nutritional supplements or gastrostomy tubes, are needed. This may lead to better health outcomes for these children, as well as reducing some of the stigma surrounding the smaller stature of these individuals.

The study is enrolling individuals with ichthyosis under the age of 25. Participation requires completion of an electronic consent form and a medical record release to obtain childhood growth chart data from pediatricians or other providers. Data on unaffected siblings may also be requested.

For more information, visit firstskinfoundation.org/growth-and-development-study or contact ichthyosisregistry@yale.edu.

Learn more about clinical trial opportunities at firstskinfoundation.org/clinical-trials.

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

FIRST Mom Puts Ichthyosis Research at Center of PhD

Carleen Walsh, whose daughter Lucy is affected with ichthyosis, was recently awarded a PhD after completing research on the implications of caring for a child with ichthyosis.

Walsh's studies explored the connections between ichthyosis severity and caregiver support needs. They also led to the development of a caregiver needs assessment tool to identify unmet needs.

Walsh hopes that findings from this research will improve understanding of challenges faced by caretakers, potentially closing the gap between the level of health care and social services support currently available to families and the level of support

perceived as needed by the caregiver.

Dr. Amy Paller praised the research. "There is no one who understands the ichthyosis experience better than family," she said. "I have been so excited about Dr. Walsh's decision to focus on the unmet needs of the family in terms of support -- and make it her mission to study these needs with guidance from academic experts."

Walsh expressed gratitude to the study participants, many of whom came from the FIRST community. "I am both humbled and privileged to have made new life-long friends, who have taught me so much from their lived experience and wealth of expertise," she said.



CLINICAL STUDY FOR THE TREATMENT OF NETHERTON SYNDROME

FIRST sponsor Quoin Pharmaceuticals Ltd. is an emerging company focused on the development and commercialization of therapeutic products for rare diseases. One of their key areas of concentration is Netherton syndrome and we are pleased to announce they have launched a new clinical study for this disease.

This randomized, double blinded, vehicle-controlled study is being conducted under a U.S. Investigational New Drug (IND) Application and will assess two different doses of QRX003 topical lotion versus a vehicle lotion in Netherton patients. The test materials will be applied once daily over a 12-week period, to pre-designated areas of the patient's body.

QRX003 is a topical lotion that is formulated with a proprietary delivery technology that provides a barrier protection for the skin whilst simultaneously moisturizing it. The active ingredient in QRX003 is a broad spectrum serine protease inhibitor, whose mechanism of action is intended to down-regulate the hyperactivity of skin kallikreins leading to a more normalized rate of skin shedding. If proven to be safe and effective, long term daily application of QRX003 could lead to the development of a more typically functioning skin barrier and a significant improvement in the quality of life of Netherton patients.

If you are interested in participating in the study or learning more please visit our website at www.NethertonSyndromeClinicalTrials.com.



Research Explores X-linked Comorbidities, Affects of Vitamin D and Toll of Caregiving

“Big Data” Exposes Issues Linked to X-linked Ichthyosis

Reference: Brcic L, Wren GH, Underwood, FG, et al: Comorbid medical issues in X-linked ichthyosis. *JID innovations* 2022;2:100109

Review: X-linked ichthyosis occurs primarily in males, although mothers of these boys who carry the abnormal gene deletion on the X (female) chromosome can also show signs and symptoms of dry skin and more. The gene codes for a protein enzyme, steroid sulfatase (STS), that is important for skin integrity and many other important functions of other organs in the body. “Comorbidity” is the word that describes these other medical issues.

Different deletions in the same STS gene can cause different associated comorbidities. Boys may be born with undescended testicles, asymptomatic eye issues called corneal opacities, and neurodevelopmental or mood disorders. Young pregnant women who carry abnormalities in some of their genes have lower placental estrogen hormone that can result in longer gestations beyond term and delayed or prolonged labor. They also have milder dry skin and milder mood disorders.

Ichthyosis investigators in England analyzed a large United Kingdom databank containing medical information on over 500,000 middle-aged individuals from the general population. They uncovered new associated medical conditions in small numbers of affected adults that include heart problems, increased bleeding disorders such as nose bleeds, and placental hematomas in pregnant women. They also discovered an increased risk of palmar fascial fibromatosis also known as Dupuytren contracture. Exactly how the abnormal STS gene is related to these observations is not completely understood.

Patients with X-linked ichthyosis should know about newly discovered comorbidities and inform their primary care physician to be on the lookout for them.



These summaries of recent research are provided by Robert A. Silverman, MD, FIRST Medical and Scientific Advisory Board member.

Don't Forget Your Vitamin D

Reference: Bakshi S, Mahajan R, Karim A, et al: Oral vitamin D versus acitretin in congenital non-syndromic ichthyosis: double blinded, randomized controlled trial.

Review: Vitamin D is very important for day-to-day maintenance of your skin. It regulates cellular proliferation, differentiation, barrier function and immune function – all processes that are affected in patients with most, if not all, forms of ichthyosis. Because there is such a rapid production or turnover of skin cells in ichthyosis, it should not be surprising that vitamin D levels are frequently reduced.

There has been an interest in studying the role of vitamin D in ichthyosis for many years. Researchers performed a controlled, evidence-based evaluation of vitamin D administration in presumably non-vitamin D deficient children with ichthyosis and compared their findings to a similar group of children who were given acitretin, a retinoid that has been used for years for treating ichthyosis. They measured their results with several important validated scales, Visual Index for Ichthyosis Severity (VIIS), the Ichthyosis Area Severity Index (IASI) and the Ichthyosis Quality of Life Index-32 (IQoL-32). They also measured the effects of vitamin D on important cellular pathways in the laboratory and looked for adverse events in the trials.

Subjects that were vitamin D deficient before the study were “normalized” with 60,000 IU vit D given eight weeks before the study began. For the 24-week study, subjects were given either vit D (2,000 iu/day) and an acitretin placebo or low-dose acitretin (.5mg/kg/day) and a vit D placebo.

While both groups improved in clinical parameters, there were no statistically significant differences in improvement between the vit D group when compared to the acitretin group. No serious adverse events occurred in any subject. In the lab, mRNA expression for IL-17 and the retinoid receptor gamma were both significantly reduced. The authors suggested that vitamin D and acitretin both work by the same mechanisms. They interpret these results as vitamin D is as good as acitretin for patients with ichthyosis.

Careful analysis of the study methods could uncover biases that led to these conclusions. Even though the results were mixed, vitamin D is very important and one should consider supplementation for all forms of “red” ichthyosis to keep levels in the normal range.

Interviews Assess Weight of Ichthyosis Caregiving

Reference: Daae E, Feragen KB, Sitek JC, von der Lippe C: It's more than just lubrication of the skin: parents' experiences of caring for a child with ichthyosis. *Health Psychol and Behav Med* 2022;10:335-356.

Review: Researchers from Norway have highlighted and confirmed the troubles, challenges and devastating effects that having a child with ichthyosis has on one's personal and family life. They sought to understand, from the parents perspective, the issues that parents confront on a daily basis as caregivers. This data will help clinicians better support parents with stress management, leading to better health outcomes for their children with ichthyosis.

The authors used semi-structured interviews, predetermined open-ended questions, and participant-led explorations to gather their findings. Participants in the study came from the Norwegian national database of rare disorders and included parents of children and adults with ichthyosis. There were a total of 19 parents and 19 children who participated in the study (48% of parents and 11% of adults in the total population of the patients with ichthyosis in the rare disease database). Interviews were by telephone which may have hampered the effects of nonverbal and visual cues on data collection but may have improved honest collection because of the sense of anonymity this technique provided. This data will help clinicians better support parents with stress management, leading to better health outcomes for their children with ichthyosis.

Results were not surprising. There were four main themes: The reactions of new parents and others to the differences in their child's skin; experiences with health-care services; the difficulties of skin care; and, the impact on relationships. Parents of newborns felt afraid, alone, helpless and in denial. The lack of knowledge of health-care professionals, the lack of guidance and poor communication only served to magnify these distressing feelings. Touching, itching, pain, financial and time burdens were universally mentioned. Overwhelming emotions frequently resulted in poor communication about their worries in even every-day situations.

I recommend that this article be made available to all parents with a child who has ichthyosis and should be provided to, and read by, all of their health care providers to enhance their understanding of the effects of this condition.

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

Raising Awareness Across the Community

The Steinbrunner Family of Harrisburg, Pa., hosted two events this year, where their family and friends came out to support Wendy's daughter, Harley.

On Rare Disease Day, Hershey Medical Center put the spotlights on and "Lit Up for Rare".

During Ichthyosis Awareness Month, they hosted a fundraiser at Hoss' Steak & Sea House where a portion of the proceeds were donated back to FIRST. They raised over \$200!

All events help FIRST to Educate, Inspire and Connect. If you'd like to host an event, please reach out to Denise Gass (dgass@firstskinfoundation.org).



OUR NEW ADDRESS! PO Box 1067 • Lansdale, PA 19446-0687

New Book Features Main Character with Ichthyosis

Meet Cece, a young, spunky, confident girl who loves to sing and dance! She was born with ichthyosis and spends a lot of extra time caring for her skin. Cece doesn't let her skin condition define her—she is a great friend, sister, and performer.

Skin-vincible, a new book by Andrea Rustad, follows Cece on the day of her big performance in her school's talent show, while learning about skin, ichthyosis, and standing up against bullying!

For anyone who has ever felt different or alone, this book was written for you with love.

Author Rustad is a medical student pursuing ichthyosis research at Northwestern University. Part of the proceeds will go to FIRST! Order your book now at <https://mascotbooks.com/mascot-marketplace/buy-books/childrens/picture-books/skin-vincible/>.

