

Ichthyosis FOCUS



Volume 27, No. 2

A Quarterly Journal for Friends of F.I.R.S.T.

Summer 2008

Ichthyosis Through the Decades

Third Part of the Series

Eustolia Perez, age 73, and Rosalie Carter, age 81, are two women who have so much in common. Both women share a strong faith in God, have been through many years of trial and error, and now are excited to share their stories with you.

Eustolia Perez - Age: 73
Edinburg, TX

What type of ichthyosis are you affected with?
Lamellar Ichthyosis



What treatments do you use for your ichthyosis? Polysorb hydrate blended with hot salt water and sometimes I add Lac-hydrin Lotion. A strong religious faith and closeness of extended family and friends have been very helpful to my coping with this condition.

How have these treatments changed over your lifetime, and has your ichthyosis itself changed? At birth, the local doctor told the family that I would not live more than three days. Immediately, they began experimenting with any suggestions recommended by doctors and healers. Ten years later, the first dermatologist came to South Texas. When I was young, my parents used homeopathic remedies including olive oil, lard, aloe vera and any other options. My parents would boil and try every plant anyone suggested; lantana, sage, mesquite, and citrus leaves were just a few. The bath had to be very warm. After each bath, I applied cream and ointment to contain the moisture in my skin, to this day, I continue the process. Scales have become larger and harder, and my skin has become harder and tightened, my eyes have changed quite a bit. The tightness has caused the lower lids to roll down, placing the tear duct on the outside.

Tell us about the challenges you face, at this decade of your life, dealing with ichthyosis. Besides my vision, my sense of touch is almost non-existent. The hardness and sleekness of my palms make it difficult to handle papers; often they just slip off my hands along with anything I hold. Holding a pen or pencil is very difficult as well. My eyes tear all the time, often blurring my vision. If my eyes are not tearing, they are very dry, often burning my eyes. During the hot weather, if I overheat, my head begins burning, my scalp dries and cracks, and I feel like my head is so tight and hot it seems like it will explode.

What seems important now that you are older compared with what seemed important when you were younger? I have always been concerned with the appearance of my skin and hair. These days I wear a wig in public. Ever since I was young, the heat has caused my body to swell to the point that my skin splits and bleeds. Now I know to avoid the heat as much as possible. When I get over heated, my body gets red and bloated. If I am not careful, my skin can stretch so much that it breaks and bleeds.

If you knew "then" what you know "now" what would you do different? I would not change the way I dealt with my problem. I experimented alongside the doctors and healers to find what would or would not work to benefit my condition.

What hopes do you have for the future, taking your condition into consideration? I would like to write an article about my life dealing with ichthyosis so that my family and others can benefit from my experience and gain a better understanding of the condition.

What advice would you have for others affected? I would like the families of children born with this condition to encourage and support them to lead as normal a life as possible. I would encourage families to build a strong religious and loving foundation. Along with a good education, they will lead a very long and fulfilling life. I also would like to thank God for my life. He has provided me with the ability to cope with this condition. He gave me a very caring family. My parents, grandparents, siblings, cousins, aunts, uncles and friends were there to guide and protect me.

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Correspondence Corner

Dear Jean,

I am a member of FIRST and have one of the most severe cases of EHK in the U.S. March of this year I had my gallbladder taken out laparoscopic. About that same time, my nerve endings became very sensitive. Since having my surgery my skin has improved greatly. No one seems to have an answer as to why, but I was encouraged to let your organization know of this. I obviously still have it, and it's still noticeable, but everyone has commented on how much better it looks. It has thinned out and become much smoother. Hope this information helps others.

Thanks,

Lisa Kehrt, Indiana

Dear Jean

My 6-year-old son, Brian, has Harlequin Ichthyosis. I have been struggling for years to maintain his skin integrity to prevent infection from having a port of entry into his bloodstream. He has been hospitalized numerous times with MRSA. His skin care regime has been full of extensive bathing and exfoliation of the dead cells and the application of Aquaphor to his entire body. The difficulties of maintaining his hands and feet without cracks have been trying, especially when he gets sick or in cold weather. I was recently introduced to a line of products by a company named Arbonne; being 40 years old the anti-aging line was suppose to be for myself. I couldn't believe how wonderful my skin felt. Brian's skin lacks lipids and is very similar to an older persons skin especially the frailness. Researching the ingredients, I learned they are all natural and blend 9 key elements to improve the skin. I decided to try it on him! I started using the NutriMinC foaming body wash which helped to exfoliate his skin in addition to soften it. I then incorporated the NutriMinC Hydrating body lotion to Brian's hands and feet in combination with the Aquaphor after. The results were incredible within 1 week. I then added the NutriMinC body serum. Brian's toes have separated and there is no build up or cracks on the bottom of his feet. I have seen changes in the texture of his skin for the better and a softening of his eye lids. He is actually asking to use these products. In the past he has never wanted or allowed me to use anything but Aquaphor on him. The retail cost is slightly high but if you like the products and join as a member for \$29.00, there is a 35% discount which makes it more manageable. The product definitely works to improve the skin. Feel free to contact me at ejaguden@gmail.com for more information.

Denise Merrill, New York

Dear Jean,

I am so excited! My husband and I have been trying different avenues in hopes of getting help for my lamellar ichthyosis and other health challenges. Finally we found something that made a big difference. Believe it or not it is water – hexagonal water! I drink it, bathe in it, and it had made my skin so soft, less scaly, and easier to care for. I feel so much better and am a lot less tired. This water also has other uses. Contact me to find where to purchase a machine that converts tap water into high alkaline, antioxidants, and super hydrates your body, which can resolve many other health issues. We can be contacted at 479.736.2503 or 479.524.2720.

David and Nola Unruh, Arkansas

Grassroots Fundraising

Harry Caray's Charity Event

Congratulations to Frank Osowski, aka Frankie O, on the successful Harry Caray's fund raising event. The charity benefit was held Thursday, June 26, 2008, the eve of the 15th Biennial Family Conference. Hundreds of people filled the world-famous Italian steakhouse in downtown Chicago for a night of great food, drinks, prizes, and comedy. Frankie O's Chicago connections were successful in obtaining donated raffle and door prizes. Bids were taken on many items such as signed sports jerseys and memorabilia, tickets to sporting events, weekend getaways, and guest appearances on Comcast Sportsnet. Raffles were drawn for gift certificates to area restaurants and a beautiful handmade quilt. A unique addition to this event was the opportunity for conference attendees to join in the festivities. The event raised over \$40,000; way to go Frankie O!



Jean stands with
Jacky & Frank Osowski

Take it to the Court for F.I.R.S.T.

J.R. Drouhard loves basketball and therefore turned his love into a fund raising event. J.R. organized a basketball tournament in his home town. He had over 75 players in four age groups ranging in ages 8 to adult. Each player received a shirt, and the players on the winning team received a special benefit basketball. This was a great turnout and raised \$200 for F.I.R.S.T.. Keep up the great work J.R.!

Nancy Osentoski Raising Funds for F.I.R.S.T.

Congratulations to Nancy Osentoski, 2007 Volunteer of the Year, for collecting over \$700 during her butterfly can collection. Nancy is dedicated to fund raising and creating awareness for the Foundation. She spends countless hours sitting at many locations to educate others about ichthyosis.



Running for F.I.R.S.T.

Martina Cheung's nephew, David, was patient number 20 to be registered with ichthyosis in Ireland. David's specific strain has still not been identified. She felt compelled to raise money to help with research for ichthyosis. During the month of April, she ran a 5K road race each weekend. By using the website firstgiving.com, she set up a fundraising page for a fast and totally secure way to donate to her event. She exceeded her goal and raised over \$5000 for the Foundation. Martina is warming up for the event for next April!



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Treatment of ichthyosis—There is always something you can do! In Memoriam: Wolfgang Küster

Heiko Traupe, MD,^a and Walter H. C. Burgdorf, MD^b
Münster and Munich, Germany

We have lost a good friend, dermatology has lost a cherished colleague at a far too early age, and patients with ichthyosis and other disorders of keratinization have lost one of their staunchest supporters, Wolfgang Küster (Fig 1).

Wolfgang was born in Kassel, Germany, on June 17, 1953, and spent the first two decades of his life there. He studied medicine in Münster, Germany, and in Vienna, graduating in 1980. Stimulated by the famous human geneticist in Münster, Widukind Lenz, who had identified the thalidomide embryopathy, Wolfgang first studied human genetics in Münster and then Düsseldorf, where he became board certified. Not satisfied to diagnose genetic disorders, Wolfgang also wanted to help these often-neglected patients and thought dermatology would be the best route to this goal. He trained in Düsseldorf with Gerd Plewig, did his habilitation on lipids in disorders of keratinization, and then joined Rudi Happle in Marburg where he was on the faculty for 6 years. The last 9 years of his life, Wolfgang was director of the TOMESA Specialty Clinic for Dermatology in Bad Salzschlirf, a large private clinic where he was able to develop an extensive practice in caring for disorders of keratinization and other genetic disorders and severe skin diseases such as psoriasis and atopic dermatitis.

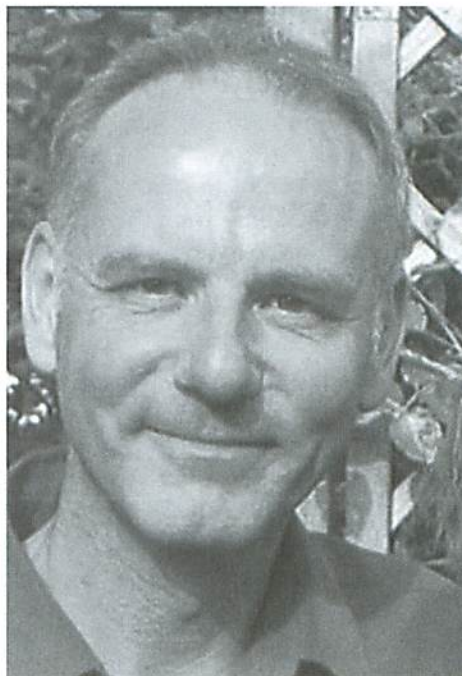


Fig 1. Wolfgang Küster, 1953-2006.

Wolfgang's dedication to his patients influenced everyone who had contact with him. He had almost limitless energy when it came to studying and caring for patients with ichthyosis. Part of this engagement came from his family; Wolfgang's second wife, Sabine Wiegandt, was for many years chairperson of Selbsthilfe Ichthyosis e.V. (Self-help Group for Ichthyosis) in Germany. Two of her three children have lamellar ichthyosis with transglutaminase deficiency; thus, Wolfgang had two stepchildren with whom he enjoyed working daily to perfect his therapeutic approach to this genodermatosis. In addition, he worked tirelessly with the self-help group and other professional organizations to correct a travesty of false economy in dermatologic therapy. Wise administrators decided that urea-based compounds were skin care products, not medications, and thus need not be paid for by health insurance companies. Through much political activity, this decision was reversed so that at least patients

From the Departments of Dermatology at University of Münster^a and Ludwig Maximilian University, Munich.^b

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Conflicts of interest: None declared.

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with ichthyosis could receive urea products on a prescription basis.

Wolfgang had considerable intellectual curiosity. His training in two disciplines had convinced him of the need for cooperative or interdisciplinary research. He was one of the driving forces behind the establishment of the Network for Ichthyoses and Related Keratinization Disorders (NIRK), a federally sponsored program established in 2003 as part of a drive to devote more resources to rare diseases.¹ The network attempts to link basic science researchers, clinical researchers, practicing physicians, and patient groups into an effective team to address the many outstanding questions in this area. NIRK today is addressing projects dealing with CHILD syndrome, palmoplantar keratoderma (PPK), lamellar ichthyosis, and gene therapy for Sjögren-Larsson syndrome.

Wolfgang left behind a written legacy of around 100 scientific articles, almost all dealing with some aspect of genodermatoses. One of his most important articles clarified the cloudy situation surrounding Unna-Thost and Vörner PPK. It had long been believed that Vörner PPK with the distinctive histologic finding of epidermolytic hyperkeratosis was a relatively rare disorder, whereas the most common form of PPK was Unna-Thost variant in which there was only marked hyperkeratosis and acanthosis. No physician or patient enjoys a palmar or plantar biopsy, so it was easiest to follow this conventional wisdom, but fortunately Wolfgang was curious and produced some surprising results. He identified members of the original family studied by Thost and biopsied them, finding epidermolytic hyperkeratosis and confirming that Thost and Vörner PPK are identical.² Later he and others identified very similar mutations in keratin 9 for patients from both Thost's and Vörner's original families.³

His last article, published in November 2006, dealt with a peculiar medical mystery that he and NIRK helped solve. Bathing trunk ichthyosis is a peculiar form of lamellar ichthyosis in which the patient is born as a collodion baby, and then has widespread ichthyosis that persists on the trunk but resolves on the extremities. Members of NIRK combined forces to show that the explanation is a temperature-sensitive mutation in transglutaminase-1, the enzyme responsible for the most common type of lamellar ichthyosis. The enzyme is effective enough in cooler body regions, but in warmer areas it cannot meet the metabolic demands and keratinization is abnormal.⁴

In our estimation, Wolfgang's greatest contribution to dermatology was his insistence that every patient with ichthyosis or PPK could be helped by highly individualized therapy. He was actively opposed to the usual dermatologic approach of

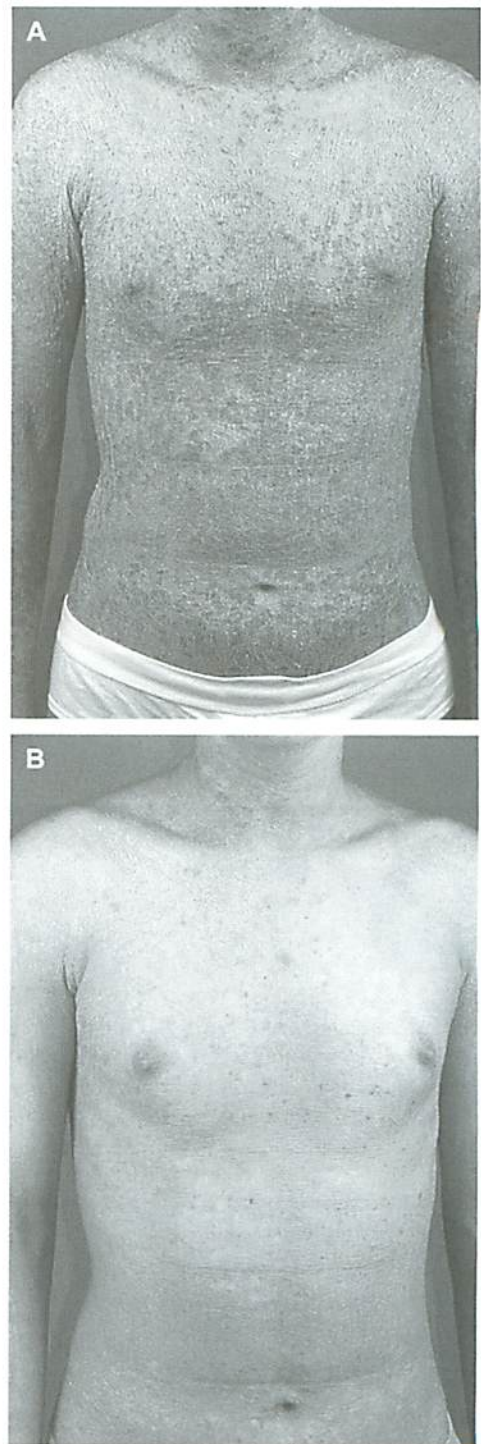


Fig 2. Patient with lamellar ichthyosis before therapy (A) and after using only topical methods discussed in this article (B). Reproduced with permission of *Deutsches Ärzteblatt*.⁵

explaining to the patient that they have a rare genetic disease for which there is no cure and only limited therapeutic options. Instead, he worked tirelessly with each of his patients to try to find a treatment plan that worked. One of his last articles appeared in the

Executive Director's Report




Dear Members & Friends of F.I.R.S.T.,

What a wonderful time to be a member of F.I.R.S.T.! We just completed our 15th bi-ennial family conference in Chicago, and it was the best one yet. Record-breaking attendance of 400 people will make this an event to remember for the next few years. Highlights from the conference are on pages 12 to 14. We are now beginning the planning for the next conference in 2010, which will be announced in an upcoming newsletter and on our website.

The "Ichthyosis through the Decades" feature column has been extremely well received. Many members have commented on being "able to relate." The idea for this column came from a member. So, please let us know what you would like to see in the newsletter and send us your ideas. You can email me at jpickford@scalyskin.org.

This October 5 – 12 is Ichthyosis Awareness Week. It's an opportunity for members from all over the country to create awareness in their neighborhoods and local communities. F.I.R.S.T. has brochures and materials for you to distribute. You can also conduct a grassroots fund raiser to generate more excitement and funds for the Foundation. If you are unsure of what you want to do or how to get started, don't hesitate to contact us. We can help you get started and provide guidance. Just think, if every member did something for Ichthyosis Awareness Week, more people would be educated about these diseases and the better the lives would be for our affected members.

Sincerely,


Executive Director

One week out of the year our members host awareness campaigns and fund raising events to raise money and educate the public about ichthyosis.

Ichthyosis Awareness Week will be officially celebrated during **October 5-12, 2008**. In actuality, Ichthyosis Awareness Week can occur anytime during the year – when you can devote your energy to raising awareness or funds for the Foundation. Educating your local community can make life a little easier for those who are affected. The simple knowledge that ichthyosis is a genetic skin disease which is not contagious, a bad sunburn, or the result of poor bathing habits can help the public better understand the disease and its symptoms.

"Everyone has something they love to do; take that love interest and turn it into a fund raiser."

Fundraising ideas:

- Auction
- Bake Sale
- Basketball Tournament
- Bingo
- Book Sale
- Candy Sale
- Car Wash
- Dance
- Donations in lieu of birthday gifts
- Dress Down Day at Work or School
- Garage Sale
- Golf Tournament
- Halloween Costume Contest
- Raffle
- Skate-a-thon
- Walk-a-thon or 5K Run



**Ichthyosis
Awareness Week
October 5-12, 2008**



How you can help...

The possibilities are endless. Start by contacting your local newspaper to let them know about Ichthyosis Awareness Week. Share your personal story with them, or let them know that you will be hosting a fundraising event.

For those of you who have always wanted to do something to help raise awareness and money for the Foundation, but have little experience...we have the answer. The Foundation staff has created an easy-to-follow manual on how to organize a grassroots fundraiser. It is available free of charge to anyone who requests it. It contains great information about how to contact your local media and the essential steps to create a successful event, plus much more. Please call the office at 1-800-545-3286 or email us at info@scalyskin.org with your mailing address to receive a copy.



News & Notes

F.I.R.S.T.'s 2008 Clinical Scholar Award



The Foundation is excited to name Keith A. Choate MD, PhD as the recipient of the 2008 Clinical Scholar Program, funded by the Lennox Foundation. The goal of the Clinical Scholar Program is to identify and train the next generation of clinicians who will treat patients with ichthyosis

and who, through their own clinical and laboratory investigations, advance understanding of this group of diseases. Dr. Choate is a highly-recommended, talented, and committed person who has a longstanding interest in ichthyosis.

The National Organization for Rare Disorders (NORD) and rare-disease

patient organizations across the United States are celebrating the 25th anniversary of the signing of the Orphan Drug Act (ODA) during 2008. This groundbreaking legislation brought real hope to the



more than 25 million Americans living with one of the nearly 7,000 diseases considered to be rare.

In the decade before the Orphan Drug Act was passed by Congress and signed by President Ronald Reagan, only 10 treatments had been developed for rare diseases by the pharmaceutical industry. In the 25 years since then, more than 1,100 treatments for rare diseases have entered the research pipeline, and more than 300 have been approved by the U.S. Food and Drug Administration for marketing. In addition, the Act has proven to be a potent catalyst to the growth of the pharmaceutical and biotechnology industries in the United States.

A rare or "orphan" disease is defined by the U.S. Food and Drug Administration (FDA) as a disease or condition that affects fewer than 200,000 Americans. In the past, these diseases of low prevalence were overlooked by drug and medical device developers. The Orphan Drug Act provides financial incentives that help companies recover the cost of developing a drug for small patient populations. Special events are planned for the year. Information about these special events, and other news related to rare diseases and the development of new treatments for them, will be posted throughout the year on NORD's web site at www.rarediseases.org.

Jane Bukaty Membership Assistance Fund

The Foundation is now accepting applications to the Jane Bukaty Membership Assistance. Email a request for the guidelines and an application to:

info@scalyskin.org or call 1-800-545-3286

Completed forms may be mailed to the attention of the Jane Bukaty Membership Assistance Fund, 1364 Welsh Road, Suite G2, North Wales, PA 19454. The deadline for applications is October 31, 2008. The awards will be given in December 2008.

Donations to the Jane Bukaty Membership Assistance Fund are always appreciated and enable F.I.R.S.T. to make this fund available to more of our members.

In Memory of Dane Christian Phelps



The Foundation sadly writes about the sudden passing of Dane Christian Phelps. Dane passed away on June 25, 2008 at the tender age of three-and-a-half years. Dane had a very, very rare form of some unknown type of ichthyosis, which had never been officially diagnosed. In addition to his ichthyosis, other medical complications caused his heart to enlarge and he suffered cardiac arrest. He had a smile for everyone and loved so many. Through the numerous tests and procedures he underwent during his short life, Dane was called a little warrior. He fought until the very end. People who met Dane will never forget him and those who got to know Dane will have their lives forever changed by his precious spirit. Dane's parents, Suzanne and Eric Phelps, are members of F.I.R.S.T. and were instrumental in providing the Foundation with the opportunity to apply and ultimately receive a grant from their family foundation. Suzanne is active on the ichthyosis.com board and often gives comfort and advice to other parents with affected children. Dane was a little brother to Cade and Chandler, great-grandson to Louise Phelps, grandson to David and Loretta Norris, Janice and James Babcock, and Jeriann and Dennis Phelps. Dane was loved by many uncles, aunts and cousins. He will be sadly missed by everyone.

News on the Hill

The Fight for Increased Funding for NIH

The National Institutes of Health (NIH) is the lead biomedical research agency of the federal government. Support for NIH is key to ensure that researchers continue to make strides in genetics and treatment research for ichthyosis and other diseases. NIH received less than a 1% increase this fiscal year. This represents the fifth year of stagnant growth and almost an 11% drop in purchasing power for the NIH.

However, there is some hope out there. Included in the recently passed supplemental funding package to pay for the Iraq war is an additional \$150 million for NIH. The NIH could also see increased funding in fiscal year 2009, which begins October 1, 2008. While the President proposed essentially flat funding, the House and Senate Appropriations Committees are working to add over \$1 billion, or roughly a 3.5% increase. Unfortunately, the appropriations process has been thrown off track by procedural partisan wrangling in an election year. It is unlikely that the funding bills will be finalized until early next year.

F.I.R.S.T. supports a 6.6% (\$1.9 billion) increase in NIH. This amount would allow NIH to keep pace with biomedical inflation and continue to fund vital research and attract the best and brightest researchers. Please urge your congressional representatives to support a 6.6% increase in NIH. For information on how to reach your federal representatives go to www.house.gov and www.senate.gov.

Genetic Non-discrimination Act Signed Into Law by President

In response to growing concerns over discrimination on the basis of genetic information found through genetic testing, Congress passed the Genetic Information Non-discrimination Act. The bill was signed into law on May 21, 2008. The bill prohibits discrimination on the basis of genetic information with respect to health insurance and employment. The measure also establishes basic legal protections that will enable and encourage individuals to take advantage of genetic screening, counseling, testing, and new therapies. Furthermore, the bill prevents health insurers from denying coverage or adjusting premiums based on an individual's predisposition to a genetic condition, and prohibits employers from discriminating on the basis of predictive genetic information. Additionally, the bill stops both employers and insurers from requiring applicants to submit to genetic tests, maintains strict use and disclosure requirements of genetic test information, and imposes penalties against employers and insurers who violate these provisions.

Health Information Technology Legislation Approved

On Wednesday, June 25, the House Energy and Commerce Subcommittee on Health approved the "Protecting Records, Optimizing Treatment, and Easing Communication through Healthcare Technology Act of 2008." The legislation promotes the nationwide adoption of a Health Information Technology (HIT) infrastructure and establishes incentives for doctors, hospitals, insurers, and the government to exchange health information electronically across the country. Additionally, the bill makes permanent the Office of the National Coordinator for Health Information Technology at the U.S. Department of Health and Human Services and encourages the use of an electronic health record for each person in the United States by 2014. It also strengthens the law protecting the security and privacy of an individual's health information through provisions such as requiring notification when personal health information is breached.



News on the Hill is a new column to keep members current with the legislation in Washington, DC. This column is written by Angela Godby, Assistant Vice Chancellor for Federal Relations for the University of Texas System. She is affected with Lamellar/CIE.

Table I. Guidelines for individualized therapy*Congenital ichthyosis (neonatal intensive care)*

- Monitor fluid balance, electrolytes; watch for skin infections
- Incubator with high humidity but somewhat lower temperature; remember infants may have trouble with sweating; monitor temperature
- Never bathe without relubricating; apply nonmedicated cream thinly 6-8 times a day
- Ectropion management by ophthalmology
- Watch for flexural contractures
- Check vision and hearing; get neurologic evaluation
- Put family in touch with ichthyosis self-help group

Newborns and infants

- Bathe twice daily with sodium bicarbonate
- Rub lightly with soft washcloth or microfiber towel
- Apply nonmedicated cream 2-3 times daily
- No urea or salicylic acid
- Ectropion management by ophthalmology; cleaning of external ear by otorhinolaryngology
- Physical therapy to avoid flexural contractures

Children

- Bathe twice daily with sodium bicarbonate
- Rub with soft washcloth, microfiber towel, or pumice stone
- Apply 5% urea cream twice daily
- Treat scalp with stronger urea cream 7%-10%
- Ectropion management by ophthalmology; cleaning of external ear by otorhinolaryngology
- On warm summer days, measure temperature, force fluids, cool environment, no occlusive ointments; play outside earlier or later, not in midday
- No vacations in warm climates

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Deutsches Ärzteblatt (a journal distributed to every German physician) in June of 2006, and was entitled "Ichthyosen: Vorschläge für eine verbesserte Therapie".⁵ One of us (W. H. C. B.) contacted Wolfgang to help translate this article into English, but never received an answer. With the help and permission of Dr Christopher Baethge, Medical-Scientific Editor of the *Deutsches Ärzteblatt*, we now present paraphrased excerpts of Wolfgang's last article translated into English to make this important clinical concept available to more readers. The material taken from the German article is printed in italics.

ICHTHYOSES: SUGGESTIONS FOR IMPROVED THERAPY

Introduction

A MEDLINE search for "ichthyosis therapy" from 1990 to 2005 revealed 262 citations but only 4 controlled studies. Only a few clinical centers concentrate on ichthyoses, which are both rare and heterogenous. Following up patients for years gives one insight into the effectiveness and safety of a given therapy. In the past 20 years in the university dermatology departments in Düsseldorf and Marburg and the TOMESA Clinic in Bad Salzschlirf, I have treated 692 patients with ichthyosis and had the opportunity to observe them, their problems, and

their response to therapy. Despite the marked differences in both gene defect and clinical manifestations of the many forms of ichthyosis,⁶ the same general approach to therapy can help almost all these patients. I have tried many approaches not previously reported that have enabled me to develop a beneficial treatment plan for almost all my patients. Table 1 summarizes my approach.

Newborns

Newborns with congenital ichthyosis are often premature. Many present as a collodion baby surrounded by a thin membrane that splits soon after birth and is lost in the first weeks of life. Left behind is erythematous skin that, during a period of weeks, begins to develop scales of varying intensity. These patients require intensive nursing care, as their skin is not capable of normal water retention or temperature control. Setting the incubator temperature requires a fine touch. If it is set too high, they may become erythematous and have a fever that is mistaken for an infection; if too low, they get cold. Their abnormal skin barrier can also lead to electrolyte imbalance and infections. If there are no nondermatologic contraindications, they should be bathed 1 to 2 times daily and lubricated with a thin, nonocclusive cream 6 to 8 times daily to restore the

Table II. Keratolytic or hydrating substances with usual maximum concentration

Agent	Maximum concentration (%)
Urea	10
NaCl	10
Lactic acid*	5*
Salicylic acid only for stubborn areas	10
Tretinoin	0.05
Glycerin	10
Vitamin E acetate	5
Dexpanthenol	5
Propylene glycol	15

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*Lactic acid much better in optimized buffered commercial preparations where 12%-14% concentrations can be achieved that are well tolerated.

normal moisture and reduce scale formation to a minimum. Creams with salicylic acid cannot be used in this setting; absorption is dramatic and metabolic acidosis can occur rapidly and be life threatening.

Bathing

Regular bathing, even more than once daily, is important for many reasons. Both scales and residual medications are loosened, while at the same time, the skin is tanked up with the water it has lost because of the imperfect barrier. The loosened scales can then be removed mechanically. Neither oil nor salt bath additives are ideal; the former is messy and may be occlusive, whereas the latter can be irritating. Instead, I have found sodium bicarbonate (NaHCO_3), commonly known as baking soda, most effective.

Several handfuls in a tub of water are required. By denaturing the keratin and making the water alkaline, it helps remove scales. Of the more than 350 inpatients I treated with NaHCO_3 baths, more than 70% profited from this approach. Other agents that can be tried if NaHCO_3 is not helpful include wheat starch, corn starch, and rice starch.

A steam bath is even better for softening scales but is not widely available. Patients with bullous ichthyoses such as congenital ichthyosiform erythroderma Brocq tend to have foul-smelling areas of maceration, which is a major social problem. Antiseptics such as 0.1% octenidine or 0.1% polihexanide can be added to bath water or applied afterward.

Mechanical scale removal

Mechanical removal of scales after bathing is more efficient than the use of keratolytics. After

Table III. Two favorite prescriptions

5% urea cream for body (water in oil)	
Urea	5.0 g
Lactic acid	1.0 g
Sodium lactate 50%	4.0 g
Water	35.0 g
Glycerin	5.0 g
Aquaphor Original*	ad 100.0 g
Shelf life: 6 mo	
Urea scalp cream	
Urea	7.0-10.0 g
Lactic acid	1.0 g
Sodium lactate 50%	4.0 g
Propylene glycol	10.0 g
Water	40.0 g
Basic cream DAC	ad 100.0 g
Shelf life: 6 mo	

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DAC, Deutscher Arzneimittel-Codex.

*Manufactured by Beiersdorf, Norwalk, CT.

trying many different ways of rubbing away scale, I have found that microfiber household towels, pumice stones, and special silk from China are most effective. Effective, relatively painless scale removal by repetitive gentle rubbing can be achieved in the bathtub once the patient has soaked for 30 minutes. The process is easier to describe than to do—it requires training, is physically exhausting, and lasts at least 1 hour if the entire body is affected. The required interval is between twice daily and twice weekly. A bland cream must be applied immediately after getting out of the tub.

Topical therapy

Newborns and small children should be treated with a medication-free vehicle base. The ratio of body surface area to weight is 2.5-fold larger than in adults, so that the resorption of topical substances is proportionately higher. In addition, the skin is more sensitive so that most keratolytics are not tolerated. Later a number of compounds can be considered for use (Table II). Topical corticosteroids are not in this group; their use is contraindicated for treating ichthyosis.

Urea is the most important active ingredient in ichthyosis therapy. It is a classic humectant, binding water and facilitating epidermal barrier regeneration, with additional keratolytic and antimicrobial effects. It should not be used in the first year of life because of irritation and risk of elevated blood levels.

Salicylic acid is an effective keratolytic but is too well absorbed. It is contraindicated in infants and

small children and is best used in older children and adults to treat limited stubborn areas.

Tretinoin (0.025%-0.05%) is also keratolytic but irritating, especially because after a few applications it causes fine barely visible but painful fissures. In addition, with widespread use, there is enough absorption to raise the issue of teratogenicity and make its use in females of childbearing age inadvisable.

Additional agents are considered in the Table II. Lard (*Adeps suillus*) can be used to loosen thick crusts on the scalp, palms, and soles. Glycerin is helpful to improve skin flexibility for those patients with stiff or armorlike scales.

Because of the tremendous individual variation in skin sensitivity and response, the therapy must be optimized for each patient. The best way of doing this is to do side-by-side comparisons, treating each half of the body differently. To increase the effectiveness of keratolytics, troublesome areas can be wrapped with clear household wraps for a few hours or overnight. Because of problems with temperature control, large areas should never be so occluded.

The scalp requires special attention. Many patients have thick plates on the scalp that are often secondarily infected, malodorous, and lead to scarring alopecia. The trick is prolonged occlusion, using a washable urea cream at night under occlusion. Finally the scalp can be soaked in the tub by older children, who enjoy diving or the contortions required. After gentle washing, the scales and crusts are teased off.

Most patients with ichthyosis require 4 different topical agents: a washable cream for the scalp, a low-concentration cream for the face, a midconcentration cream for routine use on the body, and a special high-strength preparation for intermittent use in problem areas. The reality is that most patients must bathe twice daily and frequently rub their skin for debridement. A teenager or adult will require 2 to 3 kg monthly of their basis midconcentration cream or ointment. Patients with ichthyosis require so much topical medication that considerable cost savings can be achieved by compounding (Table III).

Quite remarkable improvement can be achieved by following this regimen (Fig 2). Wolfgang went on to discuss in similar detail the following fine points of ichthyosis therapy that we have summarized in telegraphic style:

Ocular problems: Ectropion can often be avoided by regular lubrication and use of artificial tears. Surgical correction is possible.

External ear: Accumulation of debris often leads to impaired hearing, a totally unnecessary disability

for these children. The eardrum may also be affected by the disease.

Contractures: Regular physical therapy and later activity is essential to avoid contractures secondary to stiffened skin.

Systemic retinoids: Some forms of ichthyosis benefit dramatically from systemic retinoids. For example, harlequin ichthyosis is no longer universally fatal because of the early use of acitretin. The acute problem is always irritation, especially for patients with erythematous forms of ichthyosis, whereas chronic problems include teratogenicity, lipid abnormalities, and irreversible skeletal changes.

Anhidrosis: Problems with sweating are often overlooked for patients with ichthyosis. When the outside temperature is above 70°F, patients should be careful, avoiding strenuous activity and increasing fluid intake. Both heat stroke and febrile convulsions are very real risks.

Inpatient training: Wolfgang and his coworkers devoted considerable energy to teaching patients with ichthyosis and their parents how to care for the disease.

Genetic counseling should be offered to all affected families.

Psychosocial issues: Ichthyosis is a stigma for every patient, as almost invariably the visible skin surfaces are affected, leading to social rejection. Therapy is difficult, expensive, and time-consuming, and produces improvement, not cure. These patients deserve social support, as offered by NIRK, and in many instances require professional psychologic care. In Germany, patients, even small children, are estimated at 70% to 100% disabled and then eligible for financial help for medications and nursing support.

The article was much longer than this perhaps too brief summary, completely demonstrating Wolfgang's devotion to patients with ichthyosis, considering care at every age group, emphasizing a multidisciplinary approach, and expressing concern for social and financial issues. It is his legacy to a group of patients who meant so much to him.

Wolfgang Küster died suddenly, probably from a cardiac arrest. His wife, Sabine, told one of us (H. T.) that he had been restless in the weeks before his death, full of plans for new projects but still uncertain about the future. One can almost imagine that he had somehow anticipated that he was running out of time. H. T. had dinner with Wolfgang 2 weeks before his death, at a meeting in Freiburg, and found him in good spirits and pleased with the progress of dermatologic genetics. He was looking forward to participating in Germany's oldest and most-famous

2008 National Family Conference

Conference Highlights



Chicago, Illinois was a wonderful city to host the 15th Family Conference. The city proved to be a friendly one, as our families ventured out to explore the zoo, the aquarium, and the many quaint neighborhoods surrounding the city days before the conference, during Friday night, and at the conclusion of the conference.



We are proud to announce that this conference is the best attended to date with 400 attendees! Of those registrants, 50% are new families who have never attended a conference before!

Families traveled from as far as Bhutan in S. Asia, Bermuda, Australia and Belgium to meet each other for the weekend.

The 96 clinical screening appointments kept the dermatologists busy all day Friday.

This was an invaluable opportunity for families to meet the leading experts in the country. For some families, this was the first time meeting a knowledgeable dermatologist who could provide accurate and experienced medical advice!



The opening session was filled with energy as Jean Pickford spoke about the Foundation and introduced the programs and services offered, as well as how F.I.R.S.T. *educates, inspires, and connects* members. "Getting to know you" activities brought laughter to the room as beach balls with crazy questions were passed around to help "break the ice." Pearls of wisdom were given by members to inspire others to get the most of the conference.

Thanks to the wonderful services provided by Corporate Kids Events, 76 children enjoyed a weekend of crafts, playing, and having their own fun in child care camp.



The teenagers came together to learn how to express their feelings through art and how to handle the bullies they come across throughout their lives. They also enjoyed seeing all of Chicago by way of the Sears Tower's Skydeck. The day was beautiful

and clear, which allowed them to see for miles! Bowling followed the Skydeck! The teens also helped put together a slide show which was held at closing. This was a great way to recap the events of the entire conference.



While the teens were out on the town, the adults had their own fun during free time. They could be found playing Texas Hold'em, relaxing with chair massages, or participating in a yoga session.

Goodie bags, filled with a variety of lotion samples, and a conference T-shirt were given to each registrant.

A panel of nine F.I.R.S.T. members inspired others to do what they love and raise funds and awareness for F.I.R.S.T. Members spoke about the grassroots fundraiser they held, and how they found success.

Jennifer See and Laura Hogan spent countless hours prior to the conference soliciting raffle prizes, 96 in all! They also spent many hours at the conference selling raffle tickets. Their hard work and dedication raised \$3,000!

The Nikko room, filled with bamboo and festive ponds, was a grand setting for our Hawaiian dance party. Everyone received colorful leis as they entered the party. The talent review was held during the party, with talents of singing and keyboarding.



Volunteer of the Year Awards for 2006 & 2007

Presented to a member(s) whose leadership and heartfelt dedication to F.I.R.S.T. best exemplifies the spirit of giving.

2006 Recipient—Denise Benedetto, Wantagh, NY

2007 Recipient—Nancy Osentoski, Almont, MI

Frances Bernsteil Memorial Awards

Presented to a member(s) whose outstanding grassroots fund raising efforts help to contribute to the financial strength of F.I.R.S.T.

Amie Jo & Josh DeGarmo, Napa, CA
Dawn & Joe Johnson, Friendswood, TX
Kelly & Mark Klafter, Alpharetta, GA
Jacky & Frank Osowski, Mokena, IL
April Sproull, Montgomery, TX

Appreciation Awards

Mike Briggs
The Estate of Edna & Myron Curl
Suzanne Phelps
The Estate of Jean Giroux



Distinguished Service Awards

Presented to a member(s) for their extraordinary contribution or service to the F.I.R.S.T. by helping us grow in strength, membership, or recognition.

James Borden—Schaghitcoke, NY
Stephanie Dexter—Cranford, NJ
Hardeep Dosanjh—Ontario, Canada
JR Drouhard—Conway Springs, KS
Chris & Michelle Dugan—Rochester, NY
Sharon Enting—Valley View, OH
Paul & Betty Ann Foulks—Midway, TN
Matthew Gray—Elkhorn, NE
Sarah Hodgkinson—Ontario, Canada
Elizabeth Kahn-Jaguden—Huntington, NY
Lisa Klima—Cleveland, OH

Matt & Kathy Kouba—Planfield, IL
Kim Mayone—S. Portland, ME
John & Agenia McCarthy—Bakersfield, CA
Janet McCoy—Lititz, PA
Gary Mills—Newport News, VA
Eustolia Perez—Edinburg, TX
Virginia Scully—Island Park, NY
Susan Suda—Grafton, ND
Shelley Trojanowski—Hillside, IL
Eddie Wohlgang—Mayfield Village, OH
Lisa Wright—Laurel, MD

THANK YOU to all our sponsors for your generosity and support.



Conference Kudos

"Thank you so much for all that you do for F.I.R.S.T. As a corporate office manager, I attend many seminars, symposiums, etc. Never have I attended one that was so perfectly run and so easy and enjoyable. What a wonderful job you did!"

Thank You,
Glenda H. Swindle, TN

knowing that there are other moms that feel the same way I do was breath taking and very emotional. I thank God everyday for the F.I.R.S.T. Foundation, and my family that I have within the world because of it! Thank you for the AWESOME job that you do at the office for all of us!"



We love you all,
Paula and Michael Wetterlund, NE

"Thank you! Thank you for a fantastic conference and a wonderful time. You've done it girls.....your hard work, loving dedication and genuine love

and good will paid off. It was a grand affair and it is all so very much appreciated.

Thanks a million again and again,"

Edie Wohlgang, OH

"I just wanted to say that I enjoyed meeting you, and also wanted to thank you for the opportunity to help out with and attend the conference. I enjoyed it very much, and appreciated the chance to meet and talk with patients, plus I learned a lot from the focused sessions. And the tweens/teens field trip was so much fun! Thank you for the work that you do, and I hope to see you again in the future."

With warm regards,
Dr. Diana Lee, IL

"WOW! I want to let you know how wonderful it was to meet everyone! I also want to say F.I.R.S.T. is the most awesome family we could ever ask for! Michael and I both took so much away from the conference in knowledge, and have been sharing it with everyone we know! The first thing that I tell everyone and anyone that asks me how the conference was is, 'I can describe it in two words and then I can tell you about it! It was Educational and Emotional!' Wow, what an awesome learning experience! The MOMS group was the most uplifting,

"Thanks for the tremendous work you all put towards an incredibly successful and fun Family Conference. 'You guys rock' as Maggie would say!"

Janet McCoy & family, PA



Educate



Inspire



Connect



Rosalie Castle - Age: 81
Burlington, KS

What type of ichthyosis are you affected with? CIE

What treatments do you use for your ichthyosis? I use Lanaphilic Ointment (1lb jar) with 20% Urea mixed with a 13 oz jar of Vaseline and a 9.5 oz Pond's Cold Cream. I use it after my daily baths, at night and on any part of me that's been in water during the day. I order the Lanaphilic Ointment in a 24 lb jar once a year through the Medicine Shop

How have these treatments changed over your lifetime, and has your ichthyosis itself changed? Yes, my parents used Vaseline until I was about 12 years old. I used Pond's until about 65 and then needed stronger treatment, thus adding the Lanaphilic and later the Vaseline. My skin was very bad during childhood, then was better until 65. It was reverting back like in childhood but the Urea product keeps it in the best shape possible.

Tell us about the challenges you face, at this decade of your life dealing with ichthyosis. My challenges are being able to reach my entire body – I do 45 minutes of range of motion exercises a day to be able to do so. I am concerned if a nursing home becomes necessary; they will not understand or have time for my skin care. Another is getting into a tub to soak up and loosen scales. Another concern is availability of the products I use. I buy a year supply at a discount so I have it available when it is needed.



What seems important now that you are older compared with what seemed important when you were younger? Comfort from heat (I don't perspire) and dryness is more important to me now. When I was younger, I had more concerns with my peers reactions and remarks and feelings of being different and inferior.

If you knew "then" what you know "now" what would you do different? I'd have liked myself and felt secure. I would have bathed everyday; having well water and heating it over a wood stove made it impossible. I would have let the taunts of schoolmates hurt less. I definitely would not have tried every way suggested to tan when I was young. There was no way I would ever have tanned with red hair and my skin! Instead I had several skin cancers in later years from early sun exposure.

What hopes do you have for the future, taking your condition into consideration? I hope to realize what I can and can't do, and to find ways to comprise. Keep educated about new things discovered and to **enjoy life to its fullest!**

What advice would you have for others affected? I am so thankful for F.I.R.S.T., especially for the young parents to have support. My father had no support and did not know what was wrong. I didn't know until I was 38, and then was told there were only two kinds of ichthyosis, vulgaris and X-recessive. Even after visiting many different doctors, it was not until I was 60 years old that I learned I had CIE. Raising money for research helps a lot. Get in touch with others affected and **KEEP** in touch. Attend all the family conferences possible. I was only able to attend the one in Kansas but learned so much. If one type of treatment doesn't work try others; I realized I would have learned more by trial and error in my 81 years then I knew!

continuing medical education program for dermatologists, the 20th Fortbildungswoche für praktische Dermatologie und Venerologie in Munich on July 23 to 28, 2006. The title of his last talk—one that he never gave—was "Therapie der Ichthyoysen: Es geht immer was." In English, this would have been "Treatment of ichthyosis—there is always something you can do!" This is a fitting legacy for Wolfgang Küster, our friend and colleague, who died on July 24, 2006, just a few days before his talk. We will always remember him for his tireless devotion to patients with ichthyosis and disorders of keratinization.

Sabine Wiegandt, Wolfgang's widow, enthusiastically supported this work. Dr Christopher Baethge, Medical-Scientific Editor of the *Deutsches Ärzteblatt* allowed us to translate Wolfgang's article and reproduce the before-and-after photographs. Dr Marie-Luise Preil, Wolfgang's associate in Bad Salzschlirf, miraculously found the original photographs and provided them to us. Dr Gerd

Wolf, an expert in pharmaceutical compounding, helped us "Americanize" Wolfgang's favorite compounded prescriptions.

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