FIRST
Foundation for Ichthyosis & Related Skin Types™
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Ichthyosis FOCUS

Fall 2010 | A Quarterly Journal for Friends of FIRST | Volume 29, No. 3

Through the Coalition of Skin Diseases, we have learned of a book that might be helpful to children. The book is Different Just Like Me and it was written by Lori Mitchell, whose daughter has vitiligo.

There is no mention of vitiligo in the book, only meeting people who are different than the main character, April. The story is told from the point of view of a little girl anticipating a visit to her grandmother's house. Each day that she is waiting for the trip, the little girl encounters someone who is different, someone who is either older, speaks another language, has a disability, or is of a different race. But, in each situation, the person is doing the same thing that April is.

The book is available on Amazon and most likely also at your local library. You can visit the book website at www.differentjustlikeme.com.

INSIDE Ichthyosis Focus...

YALE TEAM FINDS A GENETIC RARITY: A Mutation That Restores Health

Errors in the copying of genes during cell division can cause numerous diseases, including cancer. Yale School of Medicine scientists, however, have unraveled the secrets of a much rarer phenomenon with potential therapeutic implications-disease-causing genes that show a high frequency of self-repair.

In the August 26 issue of Science Express, the Yale team describes how one mutated copy of a gene called keratin 10 causes a severe skin disease known as ichthyosis with confetti. However, amidst the diseased skin, these patients also have hundreds to thousands of spots of normal skin. This phenomenon, the researchers report, occurs by the recombination of chromosomes prior to cell division. Instead of producing one normal copy of the gene and one dominant, disease-causing mutation, the exchange between chromosomes results in cells with either two mutant copies or no mutant copies. If the latter occurs, spots of normal, disease-free skin will form. The investigators used these recombination events in spots of normal skin to map and, ultimately, identify the disease gene.

"Usually, you have a disease-causing mutation, and you are stuck with it," said Keith Choate, assistant professor of dermatology and first author of the paper. "But we demonstrate that in this disease, there is an unusually high frequency of the appearance of mutation-free clones of cells." The reason these particular mutations revert to normal so frequently is not clear, note the scientists. However, in all affected patients, the normal tail end of the keratin 10 protein is replaced by a protein sequence enriched for one amino acid, arginine. This causes the mutant keratin 10 to end up in the wrong part of the cell. "We believe the mis-localization of keratin 10 contributes to both the severity of the disease and the appearance of the clones of normal skin," said Richard Lifton, senior author of the paper and Sterling Professor and chair of the department of genetics.

Dr. Keith Choate

Dr. Leonard Milstone

The researchers say that knowing that these particular mutations can revert with high frequency gives them hope that they might find a way to mimic this process to develop treatments for other genetic diseases.

"Perhaps, rather than directly correcting disease-causing mutations, we might be able to recombine them away, similar to what happens in this disease," said Leonard Milstone, emeritus professor of dermatology and member of the research team.

Researchers at the University of California, San Francisco, Children's Mercy Hospitals and Clinics, and Texas Children's Hospital contributed to the study. Other Yale authors on the paper are Yin Lu, Jing Zhou, Murim Choi, Anita Farhi, and Carol Nelson-Williams.

The work was funded by the NIH and the Howard Hughes Medical Institute.

Additional participants in this project include: Amy Nopper, Alanna Bree, Peter Elias and Mary Williams

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The editor invites your correspondence. We welcome your comments, observations, and suggestions. Please send your letters to *Ichthyosis Focus* at the address listed above.

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Treasures Found...

When we do spring cleaning in our attics and basements, we never know what we might find. The roller skates we used as a child, old report cards, or a family heirloom that may or may not have financial value, but will most definitely have sentimental value. In the midst of all of the items that collect dust and create clutter, often lie dreams or memories of days past.

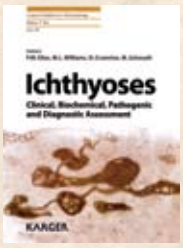
That is precisely what happened to **Anne Kaier** when she helped her mother clean out the attic of her Philadelphia home. Anne discovered letters that she had written to her mother more than 40 years ago. Anne was a student at Oxford University and lived in England in the spring of 1968. She received treatment for her Lamellar Ichthyosis in Newcastle, a wonderful town north of where she was living.

When Anne read the letters, they brought back an old voice, and soon Anne was writing about those times. Anne wrote of the clinical visits to dermatologists, of sitting on the examination table and being afraid to ask questions, and also afraid of what the answers might be. Anne's regular dermatologist while in England was Dr. Renwick Vickers, professor of dermatology at the Oxford Medical School. Her parents found the name of Dr. Sam Shuster, a specialist at the Royal Victoria Infirmary, who recommended new treatments. After trying treatments of Vitamin A and steroids that did not work for her, Anne was willing to consider a treatment of methotrexate. Dr. Shuster was a strong advocate for methotrexate, but her local dermatologist, Dr. Vickers, was cautious with this treatment option. We don't want to give away the entire story here. We will, hopefully, have the story available to you soon.

Anne also wrote about her friends and relationships while in England. They became a strong support for her as she navigated her treatment options.

Anne submitted her story to "The Gettysburg Review," published by Gettysburg College. She received the very exciting news that her story was indeed selected for publication in their Summer 2010 edition. The FIRST office is in the process of seeking permission from "The Gettysburg Review" to provide a link to the article so that our members may also enjoy Anne's work. Stay tuned for more information.

FIRST Leaders Publish a New Book



FIRST is very excited to announce that Medical & Scientific Advisory Board members Drs. Peter Elias and Mary Williams, along with Debra Crumrine and Dr. Matthias Schmuth, have published a book on ichthyosis titled, ICHTHYOSES *Clinical, Biochemical, Pathogenic and Diagnostic Assessment*. The authors hope that the book will prove to be a useful resource for families who would like to learn more about their condition. It will also be a useful tool for the staff at FIRST and is available on amazon.com for purchase. Throughout their careers, Peter and Mary have been tireless advocates for patients with ichthyosis. FIRST is very grateful for their efforts on behalf of those affected with ichthyosis.

NEWS ON THE HILL

Health Reform-What Does It Mean To You?

News on the Hill is a 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.

After years of political posturing and intense debate, new healthcare reform legislation was signed into law earlier this spring. So, what does that mean for FIRST patients and families?

This fall, a number of provisions of importance to FIRST members went into effect:

- **Children with pre-existing medical conditions can no longer be denied health insurance coverage.**

  This means that a FIRST family whose son Jimmy was born with EHK, and who has been unable to find an individual policy because of the baby's pre-existing condition, will now be able to get their son covered. This "pre-existing condition" has meant the family has had to pay out of pocket anytime Jimmy needed to go to the doctor.

- **Children will be able to stay on their families' policies until they reach age 26.**

  While some policies do currently allow coverage of full-time students, now, sons and daughters can be covered even if they are not students. So, Susie Q with Lamellar Ichthyosis will have her derm visits and creams covered under her parents' insurance plan until she is 26.

- **Annual and lifetime limits on health insurance coverage will no longer be allowed.**

  Insurance companies will no longer be able to place lifetime limits on coverage, which will ease consumers' worries about running out of coverage or facing catastrophic out-of-pocket costs.

In addition to these key provisions, an easier appeals process will be required for denial of medical coverage.

Additional insurance reform provisions and coverage benefits will be rolled out gradually over the next four years.

Additional Resources to Learn How Health Reform Affects You

The Genetic Alliance has published information in its weekly bulletin to help you stay informed and updated on the milestones of the Patient Protection and Affordable Care Act. The Kaiser Family Foundation launched a new online tool, the Health Reform Source (healthreform.kff.org), to help the public better understand the various provisions contained in the Affordable Care Act. The new website provides explanations of the provisions included in the new law, an analysis of issues surrounding implementation of the law, public opinion polling, and information from the states as provisions in the new law begin to take effect.

FamiliesUSA (www.familiesusa.org) created a blog series to collect materials and tools of their own and those of many of their partners. They have gathered fact sheets, flyers, videos, brochures, and much more.

Be sure to check out their Six-Month Anniversary Blog Series at www.standupforhealthcare.org/learn-more/quick-facts/9-23.

Executive Director's Report

Dear Members and Friends of FIRST,

FIRST recently closed its 2010 fiscal year (October 2009–September 2010). Although our fund-raising revenue is lower than in previous years, as a whole, we did not fare badly in this economy. Thank you to everyone who made a contribution during this time. Whether it was large or small, it was greatly appreciated.

Congratulations to everyone who participated in raising awareness and/or funds during Ichthyosis Awareness Week in October! Because of your combined efforts, we were able to reach new audiences, including new dermatologists, other physicians, and community leaders.

One of the new initiatives being considered at NIAMS (National Institutes of Arthritis, Musculoskeletal and Skin Disease) is itch research and treatment. As most of you know, itch is often difficult and sometimes a debilitating symptom of ichthyosis. The primary purpose of this initiative is to stimulate and accelerate research in understanding itch, with the ultimate goal of developing effective treatment modalities to control itch and pruritus. This is an exciting project with outcomes that will hopefully help our members. I look forward to sharing more information in the future.

Check us out! FIRST has a huge presence on Facebook. Be sure to become "friends" by finding us at "Foundation for Ichthyosis & Related Skin Types, Inc." Our friends receive updates and postings from other FIRST friends, so don't miss out.

FIRST will begin hosting a series of teleconferences this year, called *"FIRST to Know,"* featuring discussions on specific topics. Members are invited to join these conference calls, which will last about one hour. Members will be notified by email announcements, Facebook posts, and Events & News on our website. Space is limited, so keep checking in for dates and times.

As a reminder, FIRST provides an invaluable service for your doctors. The **Tele-ichthyosis Program** was created to assist local dermatologists with the treatment and diagnosis of patients with ichthyosis and related skin types. The program allows your doctors to upload questions, documents, and images for input and consultation from ichthyosis experts. The site uses a store-and-forward teledermatology approach in a secure, HIPAA compliant environment to facilitate communication between doctors dealing with this rare set of diseases. Please make them aware of this program so that, when they need assistance in a diagnosis, treatment option, or any other unanswered question or concern, he/she can simply go to our website and click on the tele-ichthyosis link to upload your question/case. With a few clicks, your question/case will be reviewed by our expert panel, and a reply will be sent back in a short period of time.

Finally, we are busy researching and polling members for their preference for the 2012 conference location. We have narrowed our choices to three wonderful cities and I will be making a final decision in the next few weeks. Keep checking our website for the final choice!

With best wishes,

Jean R. Pickford
Executive Director

UFIRST Scholars
Education Scholarship Program

FIRST recently announced our new college scholarship fund called U FIRST Scholars. The U FIRST Scholars program was made possible by a seed gift from Valerie & David Scholl. The Scholls are grandparents to an affected granddaughter and wanted to provide an opportunity for affected students to advance their post-secondary education in partnership with FIRST. The college scholarship fund will begin to officially accept applications in January. Visit our website for more information on program guidelines and how families and their college-bound students can apply.

In Memoriam

Janet Gregory Weary

It is with great sadness that we announce the passing of ***Janet Weary.*** Janet passed away on July 26, 2010 at her home in Charlottesville, Virginia, after a short battle with cancer.

Janet is survived by her daughters, Terry Melton of State College Pennsylvania, affected with EHK and a long-time member of our board of directors, Conway Weary of Asheville, North Carolina, and Carolyn Brandt of Crozet, Virginia.

Janet had been involved with FIRST almost since it began. Along with her husband, Dr. Peyton Weary, who passed away last year, they had been staunch advocates for Terry, FIRST, and the ichthyosis community. Janet and Peyton were honored at a testimonial dinner for FIRST in Washington, DC in February 2007. Janet also presented a very moving speech during the 1993 National Family Conference in Chicago, which was very well-received and is still talked about today.

In addition to her commitments to the Foundation, Janet was an artist whose portraits and landscape paintings are proudly displayed in many homes and institutions in her community. She was also a member of the Virginia Art Museum and The Garden Club of Virginia.

Janet loved art, music, books, and nature and passed that love on to her family. She was a devoted mother and grandmother to her 7 grandchildren and one great-grandchild.

The Weary family has generously selected FIRST as a recipient of memorial donations.

Aquaphor®
HEALING OINTMENT

CHANGES TO THE FREE AQUAPHOR PROGRAM

In the Summer 2010 Issue, we printed information about the free Aquaphor program. There are changes in the process for obtaining Aquaphor. Please follow the steps below:

Beiersdorf, makers of Aquaphor, provides a program for families that use large amounts of Aquaphor. The company will send a free case of the product, as long as the instructions below are followed. ***Please note, this has been recently changed:***

- The member/patient must contact customer service at: 1-800-227-4703 to establish an account with Aquaphor, prior to the physician faxing a request.
- Once an account is established, a doctor's letter (on doctor's letterhead), with patient/member contact information, diagnosis, and treatment must be faxed to 1-800-645-1391.
- You have to renew the request every 3 months.

The customer service phone number is 800-227-4703
The fax number is 800-645-1391

Research Projects Funded!

The 2010 Research Grant Program has concluded and FIRST announces the funding of two grants.

Dr. Anders Vahlquist

Dr. Hans Törmä

Studies on novel therapeutic options for epidermolytic ichthyosis affecting the skin barrier.

Anders Vahlquist, MD, PhD and ***Hans Törmä, PhD***, from the Department of Medical Sciences/Dermatology at University Hospital in Uppsala, Sweden, have been awarded $75,000 through FIRST's research grant fund.

Epidermolytic Ichthyosis (EI, or Epidermolytic Hyperkeratosis) is a rare inherited disease characterized by blistering in the suprabasal layers of epidermis. The affected patients suffer life-long problems from a stiff, painful, and malodorous skin that is easily infected. No drugs are known to significantly or consistently improve the widespread blistering and scaling in EI.

Vahlquist and Törmä are studying EI due to mutations in keratin genes (KRT1 and 10) which code for structural proteins constituting the cell skeleton in skin cells. The model system used is cultured skin cells (keratinocytes) from a number of EI patients with different keratin mutations and different clinical symptoms. The clinical severity of the patients has been shown to be reflected in the cultured cells. They have found that such cells, when exposed to external heat stress, show a collapse of the cell skeleton, which is similar to that in EI skin. Pretreatment of the cells with certain compounds (chemical chaperones) preserves an intact cytoskeleton, also, after heat-stress. The protective effect of these compounds not only suggests that they are putative drugs for treating EI, but also that EI resembles other protein misfolding disorders, e.g. neurodegenerative disorders, that have been experimentally treated with systemic administration of chemical chaperones.

In the continued search for improved therapeutic options in EI, they will utilize patients´ cells and a library of more than 8,000 chemicals to screen for other drugs which protect the cytoskeleton.

Methods:

Cultured EI cells are exposed to the compounds and subsequently heat-stressed. The cells are fixed and stained with protein markers that are involved in the collapse of the cytoskeleton.

The expression of selected markers is studied by automated high-content screening methodology using fluorescence microscopy, which allows us to rapidly search for compounds that prohibit the collapse in vitro.

Importantly, the safety and pharmacokinetics of such new candidates need to be tested in pre-clinical trials before controlled clinical trials in patients can be commenced.

Dr. Dennis Roop

Generating immortalized cell lines and iPS cells from EHK patients

Dennis Roop, PhD, from the University of Colorado Denver in Aurora, Colorado, along with Ganna Bilousova and Jiang Chen, have been awarded $75,000 from FIRST's research grant fund.

One of the goals of this FIRST-funded project is to generate induced pluripotent stem cells from Epidermolytic Hyperkeratosis (EHK) patients. What are induced pluripotent stem (iPS) cells? iPS cells are reprogrammed adult skin cells that have essentially all of the properties of embryonic stem cells. Embryonic stem cells can give rise to all cell types in the human body, thus they are called pluripotent cells because of this property. The fact that embryonic stem cells are pluripotent has created a lot of excitement for their enormous potential to regenerate all types of tissues and revolutionize the treatment of diseases and trauma. The potential use of embryonic stem cells has also been hotly debated over the last decade

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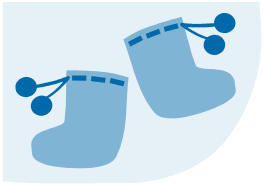
Getting Ready...

FIRST is always looking for a fun and easy way to engage all our members in a national fundraising event—***one exciting event where everyone fundraises the same way with the same tools at the same time.*** We have a few ideas brewing, so be on the alert. Our plan is to announce the national fund raiser in our next issue! It'll be fun, rewarding, and easy! Remember FUN is the first three letters in fundraising!

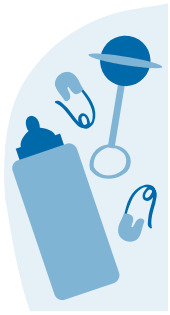


Birth Announcements

FIRST is so happy to announce some new arrivals to FIRST families.

Austin Wolfgang Knabe was born to proud parents ***Adam and Meghan Knabe*** on July 14, 2010. Austin is affected with Lamellar Ichthyosis, just like big sister Emma.

Caleb George Keller arrived on September 15, 2010 to proud parents ***Jennell and Michael Keller.*** Caleb is the baby brother of Chad Erickson, who is affected with x-linked ichthyosis.



Myles Dillon Cina arrived on October 16, 2010 to proud parents ***Sean and Jolie Cina.*** Myles, baby brother of Portia Cina, is also affected with CIE.

FIRST Research Grant – Study Update

*In 2007, **Dr. Suephy Chen** and her colleagues at Emory University received a $49,000 grant from FIRST's Research Grant Program to study "The Economic Burden of Cutaneous Disease in Ichthyosis Patients and Families." Following is an update of their study.*

In a recent study conducted by Emory University's Dermatology Clinical and Outcomes Research Unit, ***Dr. Chen*** and her collaborators sought to evaluate quality of life among patients with congenital ichthyosis (CI). Study subjects were asked to complete an online survey regarding their disease and its impact upon their quality of life and resource utilization.

Disease severity was determined using the previously validated severity scale developed by Chen et al. The severity scale examines a patient's level of redness, scaling, and hair loss. Resource utilization was determined by the amount of time subjects spent daily treating symptoms of CI (for example applying emollients and other topical treatments), as well as the number of ichthyosis-related dermatology visits patients attended each year.

Of 235 study subjects, 60% were female, 84% were Caucasian, 42% had a family history of CI, and the mean age was 29 years. Analysis showed that increased scaling, redness, age, and time spent daily treating symptoms of CI were predictive of worsened quality of life. Younger patients with increased scaling and hair loss attended more annual dermatology visits, as did those patients with no family history of ichthyosis. Younger patients with increased redness who reported worsened quality of life spent more time daily treating their symptoms.

Taken together, these findings suggest that increased disease severity has a significant negative impact upon quality of life in patients with CI. Furthermore, increased disease severity leads to greater resource utilization, whereas increased age and a positive family history of CI lead to decreased resource utilization. These findings imply that better therapies and greater patient education may lead to improved quality of life and decrease resource utilization among patients with ichthyosis.

Dr. Suephy Chen

ICHTHYOSIS AWARENESS WEEK 2010

Ichthyosis Awareness Week took place October 3–10, 2010. Many members from across the country took part in activities that focused attention on ichthyosis, FIRST, and our work on behalf of the ichthyosis community.

Erin Burke Edwards, of Thomasville, North Carolina, and ***Laurie Ann Fiore*** of Wynnewood, Pennsylvania, each set up displays with information about FIRST and ichthyosis at their local libraries to educate the general public. Erin has a 4-year-old son, Ethan who is affected with Lamellar Ichthyosis. Laurie Ann has two children affected with CIE. In addition to her display, Laurie Ann has been actively sending messages through her Facebook page alerting news media and agencies about Ichthyosis Awareness Week and FIRST.

Jeff and Tammy Gerber, of Hanover, Indiana have two daughters, McKenzie and Kennedy, affected with Lamellar Ichthyosis, and another daughter, Kendall, who is not affected. The Gerbers set up a table at the girls' school during Ichthyosis Awareness Week and held a candy and bake sale. There was also popcorn available. They made a great sign with FIRST's logo on it to display behind the table. The entire Gerber family worked together, manning the table, and raised more than $200 for FIRST. This was truly a family affair.

Daniel J. Siegel, Esq., FIRST General Counsel, has two grown sons affected with X-linked Ichthyosis. Dan sends out an e-newsletter to other attorneys. Included in his most recent newsletter, was information about ichthyosis and FIRST. He provided a link to our website so that recipients could make donations.

Sarah Hodgkinson, of Woodstock, Ontario, Canada is an avid crafter/scrapbooker and has a very active blog on the web. She entered a contest, Elmer's Crafting Challenge, in which the prize is $1,000 for the educational charity of her choice. If she wins, Elmer's will donate $1,000 for FIRST's new educational scholarship (see page 4 for more information about the scholarship).

In addition, Sarah was the featured designer at Scrapbook & Cards Today magazine twice during Ichthyosis Awareness Week. The President and Founder of Scrapbook & Cards made a point of directing people to Sarah's blog about Ichthyosis Awareness Week.

You may remember that Sarah created beautiful awareness bookmarks that FIRST sold at the Family Conference in Orlando this past summer. Sarah's 6-year-old daughter, Ema, who is affected with CIE, took the bookmarks to school and gave them to students and teachers to raise awareness. They were terrific gifts to give out. These bookmarks are also available for sale at the Marketplace on FIRST's website.

Marc Benedetto of Wantagh, New York, is an 8th grader at Wantagh Middle School. His Home and Careers class needed to develop a product and sell it, donating the proceeds to charity. A classmate of Marc's suggested the Ronald McDonald House. Marc presented a case for choosing FIRST. After a vote, the class almost unanimously voted to have the proceeds donated to FIRST. They made posters announcing the sale and sold brownies during the school day. Their efforts raised $107.00! This is a great way to educate the children about ichthyosis and FIRST.

Continued on page 14

Research Identifies New Mechanism by Which TGM1 Mutations May Cause Ichthyosis

It has been known for nearly a decade that the type I transglutaminase gene is mutated in at least 50% of all cases of autosomal recessive congenital ichthyosis (ARCI). However, understanding how these mutations lead to disease pathology is not well understood. As part of an effort to better understand the cause of this disease, Dr. Richard Eckert and colleagues at the University of Maryland School of Medicine in Baltimore recently studied the subcellular processing of normal and mutant transglutaminase in human skin cells. These studies, which were spearheaded by Dr. Haibing Jiang of the Eckert laboratory, have led to potentially important new insights regarding the pathology of this disease.

Dr. Richard Eckert

Dr. Haibing Jiang

Autosomal Recessive Congenital Ichthyosis (ARCI) is a debilitating genetic inherited skin disorder that affects 1 in every 200,000 to 300,000 people. It includes Lamellar Ichthyosis (i.e. the plate-like scales) and CIE (i.e. Congenital Ichthyosiform Erythroderma). Mutations of several genes are observed in this group of diseases and one of the genes is type I transglutaminase (TGM1). TGM1 protein is required for skin maturation, and TGM1 mutation results in deficient epidermal cornification leading to an abnormal epidermal surface, including the production of plate-like "fish" scales. TGM1 mutations are found in a significant percentage of ARCI patients.

To study the role of TGM1 in this disease, these investigators delivered TGM1 mutants to normal human skin cells and found that these mutant proteins accumulate at an abnormal location inside the cell. Dr. Jiang remarks that "the finding that mutant forms of TGM1 are abnormally distributed inside skin cells was a surprise." This accumulation is in the endoplasmic reticulum (or ER), which is a processing and quality control center for new proteins being synthesized in the cell. When mutant proteins are not properly folded, they accumulate within the ER for removal from the cell. Sometimes, mutant proteins accumulate within the ER and cannot be successfully removed, and this leads to cell death.

Appropriate folding is required for TGM1 function. "We believe that mutant TGM1 accumulates in the ER because it is not properly folded and that this accumulation of TGM1 in the ER leads to reduced TGM1 function and alters skin cell maturation and may also cause cell death," comments Dr. Eckert. The authors predict that using chemicals to restore normal folding of mutant TGM1 may alleviate some of the disease processes and symptoms. To test this idea, Dr. Jiang showed that treatment with chemicals that enhance protein folding reduces accumulation of mutant TGM1 in the ER.

Although additional research is necessary, we believe that identification of this novel mechanism is a significant step forward in our understanding of ichthyosis and that it may lead to therapies that enhance appropriate folding of mutant TGM1 leading to restoration of TGM1 function. This may help alleviate the ichthyosis-related symptoms. This work was presented in July at the prestigious *"Transglutaminases in Human Disease Processes"* Gordon Research Conference at Davidson College in Davidson, North Carolina; and a scientific manuscript describing this work will soon appear in the *Journal of Biological Chemistry* (Jiang-H, Jans-R, Xu-W, Rorke-EA, Lin-CY, Chen-YW, Fang-S, Zhong-Y and Eckert-RL, Type I transglutaminase accumulation in the endoplasmic reticulum may be an underlying cause of autosomal recessive congenital ichthyosis, J. Biol. Chem., in press). Richard L. Eckert, Ph.D. is the Chair of the Department of Biochemistry and Molecular Biology at the University of Maryland School of Medicine in Baltimore.

FIRST is a proud supporter of this conference.

Jane Bukaty Skin Care Fund

FIRST realizes that the fight against ichthyosis is not only a medical one but also a financial one. As families of affected individuals maintain their daily routine of treatments, it also can be complicated by the ongoing costs of medical supplies and other comforting aids and procedures.

Thanks to the generosity of an anonymous donor, FIRST has established the Jane Bukaty Skin Care Fund to help alleviate some of the financial burden that may be facing our members. Here's your opportunity to apply for some financial assistance for ichthyosis treatment. Since the fund is limited, the following criteria must be met by the applicant in order to be eligible for a grant.

- The applicant must be registered in our database.
- The applicant is required to submit an application indicating his/her need for funding. (The application can be downloaded from FIRST's website.)
- The application must include the amount of funding requested, the specific product/treatment for which funds are required, and a demonstration of the financial need for this product/treatment.

Awards will generally not exceed $200.00. Applications will be awarded two times per year as determined by the Review Committee. Applicants will be eligible to receive one award every two years.

Requests can be downloaded from FIRST's website and emailed to the national office at info@firstskinfoundation.org, faxed to 215.997.9403, or mailed to the attention of:

Foundation for Ichthyosis & Related Skin Types, Inc.™, Jane Bukaty Skin Care Fund
2616 N. Broad Street, Colmar, PA 18915

You will be contacted by the office if you have been awarded aid from this fund.

Donations to the Jane Bukaty Skin Care Fund are always appreciated and enable FIRST to make this fund available to more of our members.

The 2010 Research Grant Program *Continued from page 6*

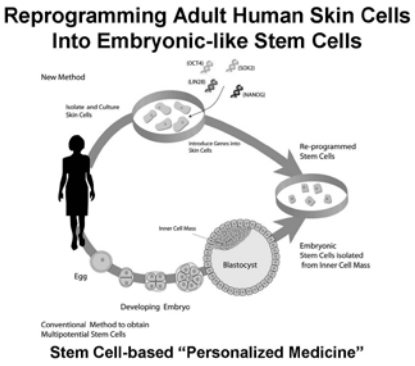


Figure 1

because of the ethical issues associated with the necessity to destroy early stage embryos to harvest these cells *(see fig. 1)*. The ability to generate iPS cells from a biopsy of adult skin potentially solves many problems associated with the use of embryonic stem (ES) cells. This strategy would not only eliminate the need for generating ES cells from fertilized human embryos, but also avoid the complication of immune rejection, which might occur with the transplant of cells derived from ES cells onto an unrelated individual. Therapies based on the use of iPS cells would truly be "personalized medicine", since the iPS cells would be generated from the same individual who would ultimately be treated with these cells. The process of reprogramming, or inducing pluripotency, basically involves taking a committed adult skin cell and introducing factors into these cells that are capable of removing all of the cell's memory and reverting that cell back to an embryonic state, a process analogous to stripping everything stored on the hard drive of a computer.

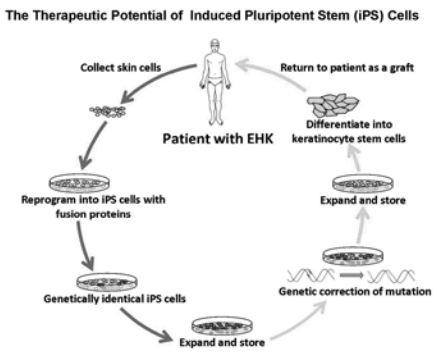


Figure 2

There has been success taking a biopsy from the skin of an EHK patient, placing the skin cells in culture, and reprogramming these cells into iPS cells *(see fig. 2)*. Like embryonic stem cells, iPS cells can be grown in culture in the laboratory indefinitely. Now that there is an unlimited supply of iPS cells generated from an EHK patient, the goal is to see if the defect can be corrected genetically; in this case a mutation in the keratin 1 gene. There has been recent success differentiating iPS cells into keratinocyte stem cells and showing that these cells can form a normal epidermis when grafted onto mice. Therefore, if the keratin 1 defect in the EHK-derived iPS cells can be safely corrected, the ultimate goal would be to differentiate the corrected iPS cells into keratinocyte stem cells and return these cells to the EHK patient as a skin graft *(see fig. 2)*.

FIRST Members Serve as Ambassadors

The Ambassador Program is growing stronger, and FIRST is thankful to everyone who has volunteered to serve as an Ambassador. Serving as an Ambassador is a way to get to know other FIRST members, as well as the local medical community. The goal of the Ambassador program is to foster connections and relationships with key medical personnel throughout the country, creating an awareness of FIRST and the Regional Support Network. The roles of the Ambassador are to build connections between and among individuals and families affected with ichthyosis; be available as a local resource for individuals seeking information regarding ichthyosis and FIRST; and be available to connect with families who have just given birth to an affected infant. FIRST supplies Ambassadors with materials to leave behind at the medical practice visited, as well as resources as support when talking to a new family. If you are interested in becoming an Ambassador, contact Moureen at mwenik@firstskinfoundation.org, or call the National office at 800.545.3286.

The current Ambassadors are listed below and can be reached by email.

REGION	AMBASSADOR	EMAIL
1	Shannon Hamill North Billerica, MA	hamr44@verizon.net
	Jennifer Hotchkiss Warrington, PA	jennifer_hotchkiss@yahoo.com
2	Kacy Leach Ellicot City, MD	kacy825@aol.com
	Jennifer See Henrico, VA	jennifersee@briansee.com
4	Mark & Suzanne Dunkin Kokomo, IN	dunkin6@comcast.net
	Mike & Denise Green McHenry, IL	elloine73@comcast.net
	Lisa Klima Westlake, OH	mlklima@att.net
	Kristi Mack Arnold, MO	Mackmommyandwife@yahoo.com
	Valerie Scholl Athens, OH	vscholl@columbus.rr.com
5	Jonathan & Sarah Kimmelman Bloomington, MN	shlyttle@hotmail.com
6	Amy & Andy Coolidge Frisco, TX	amycool@141.com
	DawnTrombley Austin, TX	trombleyd@yahoo.com
	Christy Kopp Highlands Ranch, CO	christy.kopp@kidsmobility.org
7	Michael Roberto Moscow, ID	homefry84@gmail.com
8	Cathy Bissett Sedona, AZ	cathybissett@yahoo.com
	Lisa Cummins Herriman, UT	lisadcummins@gmail.com
	Adam & Meghan Knabe Moorpark, CA	anmknabe@yahoo.com
9	Scott & Lana Deeter Surrey, BC, Canada	lanadeeter@telus.net
10	Kunle & Abimbola Osunkunle Lagos, Nigeria	iseloshe@yahoo.com

Spotlight On …

Love the Skin You're In

Carly Findlay

As many of our readers know, FIRST has a strong presence on Facebook. Through our facebook page, FIRST has met a wonderful young woman from Australia, ***Carly Findlay.*** *Carly is a young, active twenty-something who loves to write. She posts on FIRST's facebook page and also posts regularly on several blogs, including her own, www.carlyfindlay.blogspot.com/. Carly's blogs are very interesting and provide humor and comfort for those who read them. Following is Carly's post on DiVine in Australia.*

I always try to look my best. I like buying and wearing nice clothes. They can help me feel beautiful. But some people have told me that they would hate to look like me. I think they have been influenced by images of "perfect" models in magazines. There needs to be a bigger variety of body types shown in the media. This includes people with a disability. People also need to remember that being a beautiful person has nothing to do with appearances.

My dad once told me that I should always take pride in myself. He said I should always present myself in the best way possible. I should be well dressed and groomed. Walk with good posture. Always smile. Be a nice person. He told me this because he believed it would help people look past my confronting chronic illness.

His advice came many years ago. But I still think about it now. I love fashion and shopping. I try to present myself well at all times. (My dad now thinks I should spend less on my wardrobe and put more toward [retirement]!) I feel most beautiful when I'm wearing great clothes and having fun. These are the times when I can forget about my illness.

A few people have told me they would hate to look like I do. They said they could not cope with looking so different. They could not cope with the comments and stares. I think this is a reflection of their own personalities, not my appearance. I think their insecurities come from how the media emphasizes and defines external beauty and perfection.

This article was previously published on DiVine in Australia.

ASK THE READERS: *What Do You Do?*

Dear Readers,

We are resurrecting a column that was popular many years ago. At the National Family Conferences, members continually ask about many different issues from skin care to bullying. We also know that many of you have great answers to some of these questions.

We are bringing back **ASK THE READERS:** ***What Do You Do?*** to share this valuable information with others. Each issue will ask a specific question of our readers and we will print your answers in the next issue. If you have any advice you would like to share, please take a moment and send in your comments. Your questions and comments should be e-mailed to FIRST at lbreuning@firstskinfoundation.org or you can mail to: FIRST, 2616 N. Broad Street, Colmar, PA 18915.

The first question is:

How do you balance the needs of your affected child(ren) with the needs of your unaffected child(ren)?

In our Winter 2011 Issue, we will print your answers and the next question to be answered. If you have a question you would like asked, please send it to the office and we will try to print it.

Grassroots Fundraising

Our members across the nation have been very busy holding Grassroots Fundraisers for FIRST!

Softball Tournament in South Dakota

Participants enjoy a day of softball.

Chad and Michelle Iott, of Flandreau, South Dakota are the parents of 9-year-old Rylee Iott, who is affected with CIE. Over Labor Day Weekend, the Iotts hosted their second annual co-ed softball tournament. Six softball teams from the area participated in a day filled with fun, food, and friendly competition. After the tournament, the 80 participants enjoyed grilled ribs, chicken, and hot dogs, grilled by a family friend. As family and friends gathered to eat, a free-will donation bucket was placed on the serving line. Additional festivities of the day included a campfire, horseshoes, and bean bag toss. More than $600 was raised to support the efforts of FIRST.

Iott family members ***Sara (Iott) and Jory Steiner,*** of Sioux Falls, South Dakota generously donated the proceeds from the liquor sales at their August 21, wedding to FIRST. This amounted to more than $500 raised for the Foundation.

Garage Sale in New York

Jennifer Hillman, Foundation Board member from New Hyde Park, New York who is affected with Lamellar Ichthyosis, and her mother, ***Regina Femminella*** of Plainview, New York, held a garage sale, donating the proceeds to FIRST. This idea is great because everyone benefits. The person holding the garage sale is able to clean out and de-clutter, and FIRST is able to continue its mission. Thank you to Jennifer and Regina for raising more than $400.00 for FIRST.

Frank with son, Ben at the 2010 Family Conference in Orlando.

Football Raffle in Illinois

Frank Osowski, of Mokena, Illinois is the father of 7-year-old Ben, who is affected with EHK. Frank raffled off a tremendous package for the September 11, 2010 Notre Dame vs Michigan football game. Included in the package were 4 tickets to the game, pre-game sideline passes, a tour of the football stadium locker room, and a personalized football autographed by Notre Dame Head Coach, Brian Kelly. Frank publicized his raffle on local radio stations and blogs. His raffle ultimately raised more than $3,000.00 for FIRST!

Golf Outing in New York

Chris and Michelle Dugan hosted their 4th Annual Drive for a Cure Golf Tournament at the Brockport Country Club in Brockport, New York. They are the aunt and uncle of six-year-old Mattingly Dugan, who is affected with CIE. More than $2,500 was raised for FIRST.

The Foundation is very thankful to all of our wonderful members for their hard work. Grassroots fundraisers are a great way not only to raise money for FIRST, but also to raise awareness about ichthyosis in your community.

Grassroots Fundraising

Horseshoe Tournament in New Jersey

Participants enjoy the day.

Kasey Williams, of Audubon, New Jersey, has organized the 2nd Annual Chaddie's Beef n' Beer to benefit FIRST. Kasey, whose nephew, 7-year-old Chad Erickson, is affected with X-linked Ichthyosis in addition to other medical conditions, hosted her event at the Barrington Rod & Reel in Barrington, New Jersey on Saturday, July 31. Approximately 140 people attended this fun-filled afternoon, which featured an inflatable slide for the younger children, a Chinese auction, door prizes, a 50/50 raffle, and a basket of cheer. Attendees also had the opportunity to participate in a horseshoe tournament for an additional fee. Part of the funds raised were used to purchase an adaptive bike for Chad to use; the $1,700 remainder of the funds were donated to FIRST.

Kasey Williams (left) and Jennell Keller (right) with Chad and a family friend.

ICHTHYOSIS AWARENESS WEEK 2010 *Continued from page 8*

Susan Anadale of Emmitsburg, Maryland, has a 5-year-old son, Peter who is affected with Lamellar Ichthyosis. Susan has a very active homeschooling blog on the web. During Ichthyosis Awareness Week, she posted three different items about ichthyosis and awareness week, and provided a link to FIRST's website. This is a great way to educate people all over the world about FIRST and ichthyosis!

Movie Premier in North Carolina

Denver Hollingsworth, from Wallace, North Carolina, has a 13-year-old brother, Jackson, who is affected with Lamellar Ichthyosis. Denver and several friends made a movie titled "Dog Gone." The future film stars generously donated all of the proceeds from the premier of their movie to FIRST. In addition to the admission fee of $2 to attend the premier, the group sold individual copies of the DVD of their movie and donated a portion of those proceeds to FIRST. Their efforts generated $1,000.00 for the Foundation. During the weeks prior to and after the premier, the group appeared on local morning radio shows, and in the local newspaper, "The Daily News," out of Jacksonville, North Carolina. Ichthyosis was a major portion of the conversation in these interviews, and FIRST's website was mentioned often as a resource for more information.

Denver making movies and getting the word out.

Photographer John Althouse/The Daily News

Wine Tasting in New Jersey

Sean and Jolie Cina opened their home in West Caldwell, New Jersey to family and friends on October 2 for their 2nd Annual Wine Tasting event. The open house event was a great way to share an afternoon together, creating awareness about ichthyosis while raising funds for FIRST. This event, which raised more than $1,300.00, was close enough for FIRST Program Director, Moureen Wenik, to attend. Moureen had the opportunity to meet the Cinas' friends and family and to share the mission of FIRST. New Jersey State Senator Richard Codey also attended the event to show his support for Jolie and Sean. The Cinas have built a relationship with the Senator thanks to Jolie's chance meeting with him in November 2008.

Sean, Jolie & Portia Cina with Senator Richard Codey

Enjoying an afternoon with friends.

Monster Dash/ Fun Run in Texas

Dawn Johnson, ever committed to FIRST, once again held her annual Monster Dash/Fun Run at Friendswood High School in Friendswood, Texas. Dawn's daughter, 12 year old Jordan is affected with EHK. On a beautiful Saturday in October, friends, family, and newcomers came out to support Dawn and Jordan, help raise funds for FIRST, and enjoy the festivities of the day. Each participant was given a Monster Dash t-shirt, which they proudly donned. There was a race for children, and walks/runs for adults. Prizes were awarded for 1st, 2nd, and 3rd places in each category. Every participant left the day with a smile! Dawn's efforts raised more than $12,000 for FIRST!

Dawn Johnson and Jordan address the crowd before the race.

Runners take your marks!

FIRST
Foundation for Ichthyosis & Related Skin Types™
Educate • Inspire • Connect
2616 N. Broad Street • Colmar, PA 18915

FIRST Goes Green!

Send us your email address.

We are going greener and can send you an electronic copy of *Ichthyosis Focus*.

If you would like to receive the *Ichthyosis Focus* online and stop receiving a hard copy in the mail, please send your e-mail address to lbreuning@firstskinfoundation.org.

You will also receive the most current news for FIRST, including:

E-newsletters • Events in your area

RSN "FIRST To Know" Teleconference Updates

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