

Ichthyosis Focus



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

Winter 2007

Recent Ichthyosis Research

by Philip Fleckman, MD

This article is a summary of a presentation that Dr. Fleckman gave at the National Family Conference in Atlanta, Georgia.



Ichthyosis is a term for a large group of different scaling skin disorders. Because the term refers to a large group, the disorders are referred as the ichthyoses. Most of the problems in the ichthyoses occur in the epidermis, the top layer of the skin that forms a barrier between our bodies and the outside world.

While much is going on in ichthyosis research, it will be helpful for our purposes to focus on just three topics:

Profilaggrin mutations in ichthyosis vulgaris.

A new name for lamellar ichthyosis/CIE and the new mutations that support the new name and suggest a common metabolic pathway underlying their cause.

Animal models of the ichthyoses.

Before beginning, let's review a little about the epidermis:

The epidermis forms the barrier between our bodies and the outside world.

The epidermis can be divided into four layers based on how it appears when a biopsy of skin is viewed under the microscope. From the top (outside layer) down, these are:

1. The cornified layer.
2. The granular layer.
3. The spinous layer.
4. The basal layers.

The epidermis is attached to the middle level of the skin (the dermis) at the basal layer.

Profilaggrin mutations in ichthyosis vulgaris.

Ichthyosis vulgaris (IV) gets its name from the Latin word for common (vulgar) because it occurs more often than any other form of ichthyosis. It occurs in about one in every 250 individuals in the world. Until this year, ichthyosis vulgaris was thought to be an autosomal dominant disease. All of us have 23

pairs of chromosomes, the structures that carry genetic information (genes), in every one of our cells. One of each chromosome pair comes from our mother, the other comes from our father. One of these pairs determines our sex. The other 22 pairs determine the rest of us; these 22 pairs are called autosomal chromosomes. Autosomal dominant means that a change (mutation) in a gene on one of the two chromosomes in any autosomal chromosome pair results in disease. (Autosomal recessive means that mutations in the same gene on both of the chromosomes in the pair must occur for the disease to occur.)

This year Dr. Frances Smith and Dr. Irwin McLean, investigators in Dundee, Scotland, along with eighteen other investigators from across the world, showed that ichthyosis vulgaris is actually an autosomal semidominant disease. Autosomal semidominant means that a mutation in one gene of the autosomal chromosome pair results in a mild version of the disease, while mutations in the gene on both chromosomes results in the full-blown disease.

It has been known for several years that different degrees of severity have been seen in ichthyosis vulgaris, but those individuals with moderate to severe disease have no visible granular layer in their epidermis. In humans, the granular layer is made up of small granules in the cells that are composed of a protein called profilaggrin. Profilaggrin gets its name because it is a precursor of filaggrin, a protein that performs at least two functions in the cornified layer. Filaggrin means filament aggregating protein, because it helps to join together structural proteins in the cornified layer to form the barrier. It is then degraded to substances that bind water in the top part of the cornified layer. Those people with ichthyosis vulgaris with no granular layer have almost no profilaggrin in their epidermis.

A few years ago, Drs. John Compton and Sherri Bale, Dr. John DiGiovanna, and I showed that, in one large family, ichthyosis vulgaris with no granular layer was linked to an area of chromosome 1 where a number of genes involved in formation of the epidermis, including the gene for profilaggrin, are located. We suspected that the absence of profilaggrin reflected a mutation in the gene for profilaggrin, but we were unable to prove this.

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Correspondence Corner

Dear F.I.R.S.T.,

I am somewhat pleasantly amazed about a product and thought you should hear about it! I have been using Détente for just one week and I have noticed several things that I want to share.

First, if you have not seen a person with Darier disease during a period of remission, the disease remains visibly obvious because there are darkened discolorations beneath the skin's surface. They are always present where the papules have erupted or will eventually erupt when the disease is active.

I have noticed that, after just one week of using the Détente, the discolored areas on my chest, shoulder, and back are beginning to fade (or lighten) significantly. Also, where the disease exists actively year-round at and in the hairline on the back of my scalp, the pimple-like papules have been diminishing in size. Since the first day of using Détente, these papules are no longer "erupting, weeping, burning or itching" during the nighttime hours while I'm sleeping. The past three nights I have enjoyed uninterrupted sleep for the first time in recent memory because I have not needed to seek relief for the burning and itching.

While this is certainly a preliminary observation of Détente's impact, it is the first time in the 25 years I have had this disease that I have (a) noticed a change in coloration beneath the affected skin areas, and (b) experienced any sustained remission of the disease around the hairline on my scalp. Although I cannot declare that Détente has a long-term positive effect on my disease, I did want to report that, in the short term, I have noticed nearly immediate significant changes that have never occurred with any previous topical treatments, prescription or over-the-counter (OTC) product. In addition, I am also quite pleased at how small an amount of Détente is required to bathe and to apply to the affected areas.

Doug Vermillion of Indiana
d.vermillion@insightbb.com

*PS – If you are interested in
emailing me, please put
"Dariers" or "Détente" in
the subject so it is not deleted
as spam.*

Editor's Note:

*Individuals interested in
trying Détente can visit
www.detentednt.com, or call
Cindy Cooper, 610-520-0102,
or Pam Bonds, 610-296-
2404, for an information
package and sample.*

*Détente is a cleanser and
moisturizer formulated for
sensitive skin*

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Recent Ichthyosis Research

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The gene for profilaggrin is quite complex and difficult to study. Drs. Smith, McLean, and coworkers worked out a way to study the gene and showed that mutations in profilaggrin underlie ichthyosis vulgaris. They found that people with mutations in the profilaggrin gene on both copies of chromosome 1 have ichthyosis vulgaris, while those with mutations in only one copy have much milder clinical findings. This means that ichthyosis vulgaris is a semidominant disease. Smith, McLean, and colleagues showed that mutations in the profilaggrin gene are very common, which explains why the disease is seen in so many people. They also showed that mutations in the profilaggrin gene are very strong predisposing factors for atopic eczema and that there is a very strong association of profilaggrin mutations with asthma occurring with atopic eczema.

Lamellar ichthyosis/CIE - New concepts, new mutations.

This topic deals with the importance of lipids in epidermal function. Lipids are essential for all cells and for proper barrier function of the epidermis.

Many members of F.I.R.S.T. have been diagnosed with lamellar ichthyosis or CIE. Lamellar comes from the Latin word for plate, describing the plate-like scales. CIE is short for congenital (you were born with it), ichthyosiform (ichthyosis-like), erythroderma (red skin), or the condition where one is born with red, scaling skin. The scale in lamellar tends to be large and dark, while the scale in CIE tends to be small and white.

At the extreme, people with this group of diseases have the clinical findings either of lamellar or CIE. However, it has long been known that the distinction between the two is often blurred, and people can have features of both. Both are autosomal recessive diseases (the individual has a mutation in the same gene on both chromosomes) and are rare, occurring in about 1 in every 200,000 to 300,000 people.

These disorders not only have varying appearances clinically, they result from mutations in a number of different genes. The first mutation identified in these disorders was in the gene for transglutaminase type 1 (TMG1). Although mutations in this gene are found in about one-third of people with lamellar ichthyosis, they can also be seen in individuals with CIE. Since the description of transglutaminase 1 mutations, investigators in France and elsewhere have identified mutations in seven other genes that result in clinical findings of lamellar ichthyosis, CIE, and overlaps between the two.

Because almost all of these disorders are autosomal recessive, they are now being grouped into the large group of Autosomal Recessive Congenital Ichthyosis (ARCI). So, hello ARCI, goodbye lamellar and CIE. The French investigators speculate

that all these gene codes for proteins are involved in a new, common pathway of lipid metabolism. If this turns out to be true, it might explain the common clinical presentations of what were thought to be two different disease types.

Animal Models of the Ichthyoses.

No one argues that mice are the same as humans. However, both are mammals and have many genes in common. Many interesting tools have been developed to test on mice the effects of mutations in genes that are common to both mice and humans. One interesting model has been developed in the laboratory of Dr. Dennis Roop, a member of the Medical and Scientific Advisory Board of F.I.R.S.T. In this model, investigators are able to induce the findings of epidermolytic hyperkeratosis (EHK). They are now using these mouse models to test therapeutic drugs to treat this disease.

In summary, ichthyosis vulgaris is an autosomal semidominant disease resulting from mutations in the profilaggrin gene. Lamellar ichthyosis and CIE are now being grouped into Autosomal Recessive Congenital Ichthyosis (ARCI) and result from mutations in what might be a common metabolic pathway. And, animal models of ichthyosis are useful tools for understanding what causes and how to treat these disorders.

Correspondence Corner

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Dear Jean, Maureen and Staff:

We wanted to drop you a line after your recent letter. We are excited about the in-house research program and specifically that the Foundation granted Dr. Rizzo research funds around ichthyosis and Sjögren-Larsson syndrome. We know that monies could go many places but especially appreciate any help to Dr. Rizzo. Kayla is so itchy!

Our Best,
The Kopps
Highlands Ranch, CO

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Calling All Members! First Annual National Fund Raiser The Phantom Tea



The Foundation for Ichthyosis & Related Skin Types is pleased to announce our First Annual “Phantom Tea” national fund raiser. The Phantom Tea is a make-believe tea party that you invite your family and friends to attend. Using an amusing and heart-felt verse on a cute invitation, you ask them to help you raise the funds necessary to carry on the important work of the Foundation. F.I.R.S.T. will supply you with the tea bags & invitations to be given to your family, friends, and coworkers. In the comfort of their own home, they will sip a cup of tea, make a donation, and toast to a cure for ichthyosis and the related skin types.

As a national organization where our members are located in all different parts of the country, our biggest fund raising challenge has been trying to get everyone involved in a “grassroots” event. There are many of you who may have the best intentions to help raise money, but with family and work commitments, it can be overwhelming and hard to get started. This fund raiser is simple, non-threatening, and easy to do. Other similar organizations have been doing this event for years and, for them, it has grown into their largest fund raiser.

So be on the lookout for a letter from the office inviting you to participate in our first annual Phantom Tea. Your participation will guarantee our success.

What To Do If Your Child Is Being Bullied



What is Bullying?

Bullying among children is aggressive behavior that is intentional and that involves an imbalance of power or strength. A child who is being bullied has a hard time defending him or herself. Usually, bullying is repeated over time. Bullying can take many forms, such as hitting or punching (physical bullying); teasing or name-calling (verbal bullying); intimidation using gestures or social exclusion (nonverbal bullying or emotional bullying); and sending insulting messages by phone or computer email (cyber bullying).

Effects of Bullying

Bullying can have serious consequences. Children and youth who are bullied are more likely than other children to:

- Be depressed, lonely, anxious;
- Have low self-esteem;
- Be absent from school;
- Feel sick; and
- Think about suicide.

Reporting Bullying to Parents

Children frequently do not tell their parents that they are being bullied because they are embarrassed, ashamed, frightened of the children who are bullying them, or afraid of being seen as a “tattler.” If your child tells you about being bullied, it has taken a lot of courage to do so. Your child needs your help to stop the bullying.

What To Do if Your Child Is Being Bullied

1. First, focus on your child. Be supportive and gather information about the bullying.

Never tell your child to ignore the bullying. What the child may “hear” is that *you* are going to ignore it. If the child were able to simply ignore it, he or she likely would have not told you about it. Often, trying to ignore bullying allows it to become more serious.

Don't blame the child who is being bullied. Don't assume that your child did something to provoke the bullying. Don't say, “What did you do to aggravate the other child?”

Listen carefully to what your child tells you about the bullying. Ask him or her to describe who was involved and how and where each bullying episode happened.

Learn as much as you can about the bullying tactic used, and when and where the bullying happened. Can your child name other children or adults who may have witnessed the bullying?

Empathize with your child. Tell him or her that bullying is wrong, not their fault and that you are glad he or she had the courage to tell you about it. Ask your child what he or she thinks can be done to help. Assure him or her that you will think about what needs to be done and you will let him or her know what you are going to do.

If you disagree with how your child handled the bullying situation, don't criticize him or her.

Do not encourage physical retaliation (“Just hit them back”) as a solution. Hitting another student is not likely to end the problem, and it could get your child suspended or expelled or escalate the situation.

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Foundation Research Grant Recipient to Head Regenerative and Stem Cell Biology Program



Dr. Dennis Roop, ichthyosis researcher and member of the Foundation's Medical and Scientific Advisory Board, has been selected as the first Charles C. Gates Chair in Regenerative Medicine and Stem Cell Biology. This program, funded by the Charles G. and June S. Gates Family Fund, is a comprehensive new program that will conduct cutting-edge research that could lead to cures for debilitating diseases. The program is housed at the School of Medicine at the University of Colorado at Denver and Health Sciences Center (UCDHSC).

Dr. Roop was recently selected by the Foundation for Ichthyosis to receive a research grant in the amount of \$75,000 for his project, "Testing Therapeutic Approaches for Epidermolytic Hyperkeratosis Using a Preclinical Mouse Model." Dr. Roop has moved his entire lab, his co-investigator, Dr. Jiang Chen, and this particular project to the University of Colorado as of January 1, 2007.

As director of this program, Dr. Roop will recruit additional scientists and medical researchers to the program. In addition, he will oversee the comprehensive stem cell biology program, overseeing faculty who will focus on biomedical research from a wide variety of disease specialties. Richard Krugman, MD, Dean of the School of Medicine, said of Dr. Roop, "Dennis will complement the substantial existing research efforts by the School of Medicine Faculty. This gift is the largest gift to date for research at the School of Medicine at UCDHSC and we are very grateful to Diane Gates Wallach and her brother, John Gates. It is a wonderful tribute to their father." Charles C. Gates was former chairman of the board and CEO of the Gates Corporation and Gates Rubber Company.

Reprinted from UCDHSC press release, August 23, 2006.

Correspondence Corner

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using the basic diaper and doubling up on the liners. Disposable liners are also available, but I don't recommend them for children with fragile skin because they can cause irritation, especially for newborns.

Four different sizes are available from premie to toddler, and they come in many different colors. They can be ordered by phone or over the Internet and the people are very nice to work with. They recommend not using bleach on the diapers. I use a detergent that is dye and perfume-free, no fabric softener, and I run the rinse cycle two times to get rid of any detergent residue.

Here are some websites and numbers to call. I hope that all you parents will have as much success in using these diapers as I have. (I don't recommend the ultra diaper; it has a vinyl-like liner on the outside that made my son blister, and it's a thicker diaper that made it hard for him to walk.)

www.kushiesonline.com

Customer Service 1-800-841-5330

www.babybungalow.com

1-866-769-2229 (1-866-7-MYBABY)

www.thebabylane.com

1-888-387-0019

Sincerely,
Jenny Grider
McDonough, GA

Dear F.I.R.S.T.:

I have ichthyosis, but I do not follow what is going on with new treatment developments, so maybe someone has already discovered this.

I broke my finger recently and reluctantly let the doctor wrap my finger. I have never been able to stand band aids or any other wrapping before. But the doctor used Vetrap. It sticks to itself, not to skin. It is used for horses and can be purchased at veterinary supply or animal feed stores.

I have now had my finger wrapped for 14 days and am doing well. I bought rolls of the Vetrap at my local feed store for \$1.95 each. It comes in many great colors that would appeal to kids. Vetrap is called Coban when you buy it at a medical supply store, but it is much more expensive that way. Funny how people who can afford horses get charged less than people who need some specialized medical care.

Sincerely,
April Kear
Sellersville, PA

Editor's Note:

There are several Internet sites that sell Vetrap at a variety of prices. Rolls are 4 inches by 5 feet. Use Vetrap in your search box to find these sites and the best prices. Or try your local veterinary supply or feed store. Check Feed Supply in your Yellow Pages.

Spotlight On...

Janet and Maggie McCoy Everyone is Different and Different is Good

By Deborah Pulver, Sovereign Bank



Janet and Maggie McCoy

Six years ago, Janet McCoy, director of Mid-Atlantic planning and administration at Sovereign Bank, Reading, Pennsylvania, and her husband, John, welcomed their first child, Maggie, into the world. When Maggie developed Congenital Ichthyosiform Erythroderma, a form of the rare genetic skin condition ichthyosis (ick-thee-oh-sis), the McCoy's had to significantly alter their lives to care for their

daughter. They have since become experts on ichthyosis, and Janet is active in the Foundation for Ichthyosis and Related Skin Types (F.I.R.S.T.).

Because she was initially a healthy baby, the McCoy's did not realize how much little Maggie would change their world. At six months old, Maggie developed spots on her skin which turned very red and began peeling. After consulting with numerous specialists over the next year, doctors at Johns Hopkins Hospital in Baltimore, Maryland and Children's Hospital of Philadelphia confirmed the diagnosis.

Ichthyosis is a set of rare genetic diseases that cause the skin to build up and scale, making it extremely dry, among other problems. Ichthyosis is a life-long condition and there is no cure, only treatments.

"We had a very difficult time getting her diagnosed, and we weren't allowed to take Maggie out anywhere except to her doctor's appointments for months while being diagnosed," said Janet. She explained that doctors initially thought Maggie had an immune deficiency, which meant a limited life span. "We were elated to learn that her condition isn't life threatening."

In Maggie's case, she can't regulate her body temperature, so her parents and teachers must ensure she does not get overheated. She normally looks like she has a sunburn, but when she gets sick, her skin turns bright red for a day or two and then her entire body peels.

Janet and her husband have had to modify their lives to accommodate Maggie's skin condition. They installed central air conditioning in their home. Their morning and evening routines are a bit different from other families because Maggie needs to soak in the bathtub at least one hour each day and must have a

thick lotion applied to her skin twice a day. Because Maggie's skin is fragile, the McCoy's must always carry antibiotic ointment to help prevent cuts and scratches from becoming infected. In addition, a spray bottle of water is always on hand to help Maggie cool down when she begins to get warm.

When they travel, the McCoy's typically get accommodations with two bathrooms because of the amount of time Maggie must spend soaking in the tub. Trips to the beach are not possible during the summer, but they do spend time at the shore during the spring and fall months. They are in the process of building a new home, designed with Maggie in mind, where she will have her very own bathroom.

Janet and her husband have learned as much as they can about ichthyosis and now help educate others and raise funds for F.I.R.S.T. Janet began as a committee member and has been a member of the F.I.R.S.T. board for nearly three years. As a board member, Janet participates in meetings, helps raise funds, and provides guidance to other parents in similar circumstances.

She was involved in creating the organization's privacy policy and some of its literature. F.I.R.S.T. provides programs and services including publications, a support network, information from physicians, advocacy and awareness efforts, and their own research funding program to develop better treatments and, hopefully one day, a cure.

F.I.R.S.T. hosts a global conference every two years for individuals with ichthyosis and their families. The McCoy's attended the conference in Kansas City, Missouri in 2004 and again last summer in Atlanta. They have learned that Maggie's ichthyosis is mild compared to others, some of whom have severe health issues, appear to be burn victims, and/or have physical deformities.

Through F.I.R.S.T., the McCoy's have learned about treatment options and how to help prepare Maggie and her classmates for school. For Maggie's recent birthday, she asked her friends to make a donation to F.I.R.S.T. in lieu of presents.

The McCoy's try to keep their lives as normal as possible. Maggie simply has some daily issues and a few more doctor visits than most kids her age. Janet says her daughter has "probably met about 300 doctors in her six years." Maggie's treatments will remain the same for the foreseeable future but when she reaches her teens, there are retinoids and stronger medications she may try.

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Foundation Resources

Caregivers Manual 2006 Edition Now Available

At long last, *Release the Butterfly: A Handbook for Parents & Caregivers of Children with Ichthyosis* has been updated and is available to our members and the public. Thanks to generous funding from Sovereign Bank, the booklet was updated with the help of several parents and dermatologists. The 40-page booklet focuses on the early stages of dealing with a newborn to practical matters of everyday care. This booklet will be helpful to anyone who is caring for a young child with ichthyosis, including day care providers, grandparents, and other relatives. The cost of the booklet is \$5.00 plus shipping. To place an order, go to our website, www.scalyskin.org, and click on The Kiosk to place a credit card order. Or call 1-800-545-3286 to place a phone order or request an order form.

Questions Kids Ask

The brochure, "Ichthyosis: Questions Kids Ask," is now available. This helpful brochure was created by Ronit Ovadia, Genetic Counseling intern, Northwestern University. Written in an easy to understand format, this publication answers questions such as, "What is ichthyosis?" and "How do I get ichthyosis?" There is also helpful advice on how to talk to other kids about ichthyosis.

This brochure is available to all Foundation members. Simply go to the website, www.scalyskin.org, and click on Free Publications to receive your complimentary copy. Please include your name, address and phone number. Or call 1-800-545-3286.

Skin Care Products List

The Foundation's skin care products listing has been updated for 2007. Some products have been discontinued and new favorites have been added. The list is a comprehensive collection of creams, lotions, cleansers, shampoos, cooling devices, and other products recommended to us by our members.

The listing is free to all members in good standing. Please email or call the office for your copy; info@scalyskin.org, 1-800-545-3286.

Spotlight on Janet and Maggie McCoy

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Because Maggie looks a little different, she is often asked why. Her parents have taught her to respond politely in a conversational tone.

"Maggie will always have this condition, so we're trying to get her used to talking about it," said Janet. "Our motto is "everyone is different and different is good," and so far she agrees."

The McCoy's also have a son, Conor, 2, who does not have ichthyosis.

Reprinted with permission from Sovereign Bank employee newsletter.

Filing an Appeal with Medicaid to Cover Over-the-Counter Items

By Erin Burke Edwards

We recently were successful in completing the process to have many of my son's over-the-counter medications covered by Medicaid. It took a great deal of persistence not only on my part, but also his physicians'. After three long months, our hard work paid off. It is such a relief to have basic necessities (Aquaphor Healing, Eucerin ointment, Puralube Eye Ointment, and Aquaphor Baby Wash) covered by Medicaid. Be prepared for a great deal of effort, but hopefully these steps will help get you started on the right path:

Read your Medicaid booklet carefully. In North Carolina, it specifies that Medicaid must cover any medication that cures/ameliorates (makes better) a medical condition. Lotions, etc. can ameliorate ichthyosis.

Speak to your/your child's physician about the need for items to be covered. Our physician was amazed at the vast quantities of these items we were purchasing each month. Ask if he or she would be willing to contact Medicaid to file an appeal and send pertinent medical records. In addition, my son's physician was asked by Medicaid to write a letter explaining what medications, what quantity, and why they are needed (what do they prevent from happening?).

Call Medicaid and request a TAR form for an appeal. Explain why you/your child need(s) these items. When I made this first phone call, I was on the phone for upwards of four hours before I finally reached the person I needed to speak with.

After sending in the completed form, I called my contact in the pharmacy division weekly to get an update. It really helps if you have someone there on your side when the case is brought to the physician in charge of making the decision. They also understood how important it was that my son receives these medications.

As you near the completion process, find a sympathetic pharmacy. We use a small, independent pharmacy that was willing to stay on the phone all day, literally, with Medicaid. Because some of the over-the-counter products do not have NDC numbers, it made the process difficult.

Make sure when the process is complete to thank everyone who helped you along the way.

Make sure to read your paperwork carefully where you can request the medications again in a timely fashion with another form (sometimes it is in 6 months, sometimes in 1 year).

I hope that this is helpful. If I can be of further assistance in this process, please email me at: info@ichthyosis.us

Executive Director's Report

Dear Members & Friends of the Foundation,



Happy New Year to everyone! Thanks to all of our members, friends, and corporate supporters the Foundation had a tremendous year in 2006. Some highlights include launching our first in-house research grant program, hosting a successful Testimonial Dinner fund raiser and family conference, hiring a new Executive Assistant, and moving the office to larger quarters. A personal highlight for my family was the arrival of our third child, Erika, on November 1. Erika was welcomed by her big brother and sister and adds so much joy to our family.

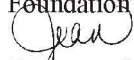


We are poised for another great year in 2007. All of our growth and success would not be possible without you, our members. The upcoming months will, once again, be very busy for the Foundation. As you will see on page 9, we are launching a new, national fund raiser called the Phantom Tea. I am also excited to announce that Requests for Proposals are being accepted for another cycle of research grant funding in 2007.

We will also be very active in February in Washington, D.C., the site of the 2007 annual American Academy of Dermatology meeting. The Foundation is hosting a second Testimonial Dinner on February 1 to honor Drs. Gloria & James Graham and Dr. & Mrs. Peyton Weary, long-time Board members. We will also be hosting our annual Medical & Scientific Advisory Board meeting on Saturday, February 3. Finally, the Foundation will be meeting with our friends in the Coalition of Skin Diseases for our annual meeting on February 5.

You may recall that, in 2002, the Board of Directors adopted a five-year strategic plan. Time flies and we are gearing up to create a new plan for 2008. A Strategic Planning Committee is in place and ready to review our successes and areas of improvement for the future of the Foundation. This committee will be meeting throughout the next six months, and I'll be ready to share the new five-year strategic plan with you by year's end.

As always, please do not hesitate to contact the office if you have an idea, concern, or news to share with the membership. Remember, the Foundation is here for you! I encourage you to pick up the phone or send me an email at jpickford@scalyskin.org.


Best regards,

Jean R. Pickford
Executive Director

A Fond Farewell to Maureen Tierney



After six years of working for the Foundation and its members as our Program Director, Maureen Tierney is moving on to explore new adventures. Maureen, a.k.a. "Mo," has been a tremendous asset to the Foundation. She has been responsible for managing the Ichthyosis Support Network, writing and producing the quarterly newsletter, and, most recently, helping to launch the in-house Research Program. In addition to these projects, Maureen used her talents in enhancing our resource library by writing the Ichthyosis Fact Sheets, Resource Sheets, and implementing many other services for our members. Over the years, some of you may have had the pleasure of meeting Maureen at the family conferences in Seattle, Kansas City, and Atlanta.

Maureen has been an integral part of our success over the past six years and she will be greatly

Correspondence Corner

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Dear Members and Friends of F.I.R.S.T.:

I am a licensed real estate agent associated with Realty One - the number one real estate company in the state of Ohio. I want to share with you a new program that can help raise money for the Foundation. It is called "The Realty One National Outbound Relocation Program." If you are planning to move your home, this program can be a tremendous resource for you and, at the same time, be a fundraiser for F.I.R.S.T.

As part of our services, Realty One offers a relocation assistance program whereby they procure the best possible real estate agents nationwide to service their clients. The program is set up as follows: anyone, anywhere, who is planning to sell or buy a home, can access their website and avail themselves of this service. In this manner, all buyers and sellers are assured that they will be provided with the best, most knowledgeable, and accomplished real estate agents specializing in particular areas throughout the nation. There are no hidden expenses and/or fees incurred either by the buyer or seller in these transactions.

In keeping with my commitment to F.I.R.S.T., I will be delighted to volunteer to manage and oversee this program. All proceeds and/or commissions realized personally by these endeavors will be immediately earmarked, directed, and donated in their entirety to F.I.R.S.T.

If you are contemplating a move, be it from one street to another, city to city, or even state to state, please contact me. My information is listed below.

Looking forward to hearing from you,

Edie Wohlgang
Mayfield Village, OH
Phone: 440-449-4124 or 216-407-7714
Email: ediew@adelphia.net

Dear Members and Friends of F.I.R.S.T.:

I wanted to share an idea that several of us are using to create awareness about ichthyosis and the Foundation. Most of us email our family, friends, and others on a regular basis. Why not add a "signature" at the end of your email that mentions ichthyosis? It's simple and easy to do.

Signatures are basically an image or images that give information about something, or just compile pictures and graphics in a way that looks nice. Anyone can make one. There are a ton of photo editing programs available. I happen to use Adobe Photoshop. But if you don't want to spend a lot of money, most PC computers come equipped with a program called "Paint." I'm not sure what it is called on a MacIntosh computer. These programs allow you to make fliers or business cards, pretty much anything!

Signatures (a.k.a. "siggies" in online chat forums) can be used for anything. As I mentioned, you can attach them to your email so each time you send a message, it will show up at the bottom. You can use it in a similar way in chat forums so every time you post a message, it will show up. You can even send them to your friends and family so they can use them too. The effort is minimal and the benefits are enormous.

I have one design below to give you an idea. Keep in mind, on a computer it is full-color. If you have questions or need help in creating one, let me know. You can contact me at Courtney.shell@gmail.com.

Good luck!
Courtney Shell
Seaside, CA



Dear Members and Friends of F.I.R.S.T.:

Three of my five children and I have EHK. I wanted to share with you an awesome diaper that has literally changed our lives in just a few weeks.

I have been struggling with disposable diapers and the effect they have on my children's skin for 10 years. Even my children without EHK were irritated by disposable diapers.

Kushies diapers are 100% cotton, very soft, machine-washable, and dryer safe. Cotton liners are available that you can double up for extra absorbency. These diapers are the best choice that I have made for my children's skin in a long time. They may seem a little expensive for the first purchase, but because you are reusing them, they are worth the price in the long run. I suggest

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Research Grant Program Moves Into Year Two

January 2007 marked the start of year two of the Foundation's Research Grant Program. The Foundation will again be encouraging investigators who are working on ichthyosis to submit their projects for review and possible funding. The projects must focus on one of the following priorities:

Original investigations leading to the improved understanding of the molecular pathogenesis of the various ichthyoses, particularly lamellar ichthyosis/CIE and EHK.

Novel approaches to the management and treatment of the various ichthyoses, particularly lamellar ichthyosis/CIE and EHK.

Investigations that address the treatment and prevention of secondary complications or quality of life issues for patients with disorders of cornification.

High priority will be given to projects that focus on the translation of known genetic information and established pathogenic pathways into curative therapies or new therapies with significant impact. These priorities have been developed jointly and are reviewed annually by the Foundation's Board of Directors and Medical and Scientific Advisory Board.

In year one of this program, the Foundation chose four projects to receive funding. Those projects focused on EHK, Lamellar/CIE, Sjögren-Larsson Syndrome, and Ichthyosis-Related research, respectively.

Interested investigators should go the Foundation's website, www.scalyskin.org, and search the Research section for the guidelines and application forms for 2007, or contact Maureen Tierney in the office at 215-619-0670.

What To Do If Your Child Is Being Bullied

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Check your emotions. A parent's protective instincts stir strong emotions. Although it is difficult, a parent is wise to step back and consider the next steps carefully.

2. Contact your child's teacher or principal.

Parents are often reluctant to report bullying to school officials, but bullying may not stop without the help of adults.

Keep your emotions in check. Give factual information about your child's experience of being bullied including who, what, when, where, and how.

Emphasize that you want to work with the staff at school to find a solution to stop the bullying, for the sake of your child as well as of other students.

Do not contact the parents of the student(s) who bullied your child. This is usually the parent's first response, but it sometimes makes matters worse. School officials should contact the parents of the child or children who did the bullying.

Expect the bullying to stop. Talk regularly with your child and with school staff to see whether the bullying has stopped. If the bullying persists, contact school authorities again.

3. Help your child become more resilient to bullying.

Help to develop the talents or positive attributes of your child. Suggest and facilitate music, athletics, and art activities. Doing so may help your child be more confident among his or her peers.

Encourage your child to make contact with friendly students in his or her class. Your child's teacher may be

able to suggest students with whom your child can make friends, spend time, or collaborate on work.

Help your child meet new friends outside of the school environment. A new environment can provide a "fresh start" for a child who has been bullied repeatedly.

Teach your child safety strategies. Teach him or her how to seek help from an adult when feeling threatened by a bully. Talk about whom he or she should go to for help and role-play what he or she should say. Assure your child that reporting bullying is not the same as tattling.

Ask yourself if your child is being bullied because of a learning disability or lack of social skills? If your child is hyperactive, impulsive, or overly talkative, the child who bullies may be reacting out of annoyance. This doesn't make the bullying right, but it may help to explain why your child is being bullied. If your child easily irritates people, seek help from a counselor so that your child can learn the informal social rules of his or her peer group.

Home is where the heart is. Make sure your child has a safe and loving home environment where he or she can take shelter, physically and emotionally. Always maintain open lines of communication with your child.

These and other materials are available online at: www.stopbullyingnow.hrsa.org, a resource from the Health Resources and Services Administration of the U.S. Department of Health and Human Services.

Camp Discovery 2007

Upcoming dates for Camp Discovery

Teen Camp

July 7 - 13, 2007 Crosslake, MN
(Ages 15 - 16)

Junior Camp

July 14 - 20, 2007 Crosslake, MN
(Ages 10 - 14)

Camp Dermadillo

August 5 - 10, 2007 Burton, TX
(Ages 9 - 16)

Camp Horizon

August 11 - 18, 2007 Millville, PA
(Ages 8 - 13)



See www.campdiscovery.org for more information, or www.aad.org Public Resources / Parents and Kids. Or call the American Academy of Dermatology at 847-330-0230

Jane Bukaty Membership Assistance Fund

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

Thanks to the generosity of an anonymous donor, the Foundation's Jane Bukaty Membership Assistance Fund helps to 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 MAF aid.

The applicant must be a member in good standing.

The applicant is required to submit a letter indicating his or her need for funding.

The letter 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 \$100.00. Applications will be awarded two times per year as determined by the Support Network & Member Assistance Committee.

Requests can be emailed to the national office or mailed to the attention of the Jane Bukaty Membership Assistance Fund at 1364 Welsh Road, G2, North Wales, PA 19454. You will be contacted by a member of the Support Network & Member Assistance Committee if you have been awarded aid from this fund. The deadline for applications for the next cycle of funding is June 30, 2007.

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.

Non-Profit Recycles Durable Medical Equipment

Foundation member Christy Kopp, of Highlands Ranch, Colorado has helped to launch a non-profit organization that recycles durable medical equipment (DME) such as walkers, wheelchairs, gait trainers, and therapeutic tricycles to children in need. Because children often outgrow their equipment long before its useful life is exhausted, Kids Mobility Network collects used DME, reconditions it, and provides it to appropriate children to enhance their lives.

Christy, who is the executive director of Kids Mobility Network, says, "DME provides greater safety, mobility, and independence for children in their important development years. It is so important for children to have the necessary equipment that allows them to optimize interaction with their peers. It helps them to develop important relationships, social skills, and self-confidence."

Christy's daughter, Kayla, is affected with Sjögren-Larsson syndrome, a rare form of ichthyosis. For more information about Kids Mobility Network, go to www.kidsmobility.org, or email Christy at Christy.kopp@kidsmobility.org. Christy can also be reached at 303-242-8281.

Emergency Information Forms for Children with Special Medical Needs

In the wake of such disasters as Hurricane Katrina and the earthquakes in Hawaii, the American Academy of Pediatrics and other organizations that support children's medical services are recommending that parents prepare Emergency Information Forms for their children with special medical needs so that first responders and emergency room doctors know what issues to consider when assessing medical needs. This form can be used as a "medical passport" to provide all the information necessary to start or update a medical file.

Emergency Information Forms are available online from the American Academy of Pediatrics, www.aap.org. Write *Emergency Information Forms* in the Search box. Forms adapted by the County of Los Angeles Children's Medical Services with more user-friendly language and Spanish translations are available from the website for the Genetic Alliance, www.geneticalliance.org. Click on Resource Repository and use the alphabetical index to access the form.

Parents and caregivers should work together with their health care provider to fill out the form, which should then be stored in a

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place where emergency personnel can find it such as the refrigerator or freezer (emergency personnel are trained to look in the refrigerator or freezer for this information). This location also protects the information from fire. Parents should also keep copies at their primary care provider's office, with the school nurse, in their car and workplace and with their child's belongings when he or she is traveling. The form can also be filed in a central repository with Medic Alert, from which medical professionals anywhere can access it. Medical Alert jewelry is also an important item to consider if your child has a serious condition, such as a food or drug allergy, diabetes, or asthma. Your local pharmacist can assist you in choosing a design that will appeal to a child and help you decide what and how much information should be engraved on the jewelry.

Correction:

A piece of information was missing from *Donate through the United Way or Combined Federal Campaign Programs* in the Summer 2006 issue. If you would like to designate your United Way donation to the Foundation, please put our name, address, and Tax ID number on your Donor Choice Form. The Foundation's Federal Tax ID number is 94-2738019. This number may also be used when soliciting donations from businesses for grassroots fundraisers or for asking for grants from local foundations and corporations.

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