F.I.R.S.T.
Foundation for Ichthyosis and Related Skin Types

ICHTHYOSIS FOCUS

Vol. 20, No. 1 | A Quarterly Journal for Friends of F.I.R.S.T. | Spring 2001

An Ichthyosis Update – Part II

This is adapted from a talk given by Dr. Mary L. Williams at the Academy of Dermatology Annual Meeting in San Francisco in March, 2000 and edited by Rita Tanis.

Genetic Heterogeneity

Confusing enough? I find refuge in the "Rule of Heterogeneity": When in Las Vegas, put your money on heterogeneity of genotypes (specific gene mutations) and phenotypes (how the gene is expressed; its clinical features). Corrollary 1 to the rule is: multiple genes may be found to produce a common phenotype. Corollary 2 is: multiple phenotypes will be associated with mutations affecting a single gene.

The lamellar ichthyosis group of autosomal recessive, primary ichthyosis is an example of heterogeneity. The nomenclature of this group remains confusing. Some US authorities lump these together under a single name, lamellar ichthyosis, while others subdivide into two phenotypes, **lamellar ichthyosis** and **nonbullous congenital ichthyosiform erythroderma** (CIE); and in Europe they are grouped as **ichthyosis congenita**, with at least 4 subtypes recognized. At least 3 genetic loci have been implicated in causing these disorders, using genetic linkage analysis, although only one gene, encoding the enzyme, keratinocyte transglutaminase 1, has been identified to date. This enzyme is unique to epidermis, hence the skin is only abnormal in these patients. The enzyme is involved in crosslinking proteins, such as loricrin, to form the cornified envelope. The cornified envelope forms a shell that surrounds the cells of the outer skin layers. The cornified envelope, together with the keratin filaments within the interior of these cells, is responsible for the mechanical strength of the outer skin layers.

Patients in this group also show considerable clinical heterogeneity, but to date efforts to sort out the phenotypes with and without transglutaminase mutations have been conflicting. In my rather limited personal experience, and corroborated by others[1], the classic lamellar ichthyosis (LI) phenotype with large plate-like scales is typically associated with transglutaminase mutations, while the nonbullous congenital ichthyosiform erythroderma (CIE) phenotype with a finer, lighter scale pattern is not. However, others have reported no correlation between TG1 mutations and CIE or LI phenotypes[2], and the final word is not yet in. If we invoke the rule of heterogeneity, we would predict at least some overlap of phenotypes in association with transglutaminase mutations.

Improved Diagnosis:

Netherton syndrome is characterized by ichthyosis, structurally weak hairs that easily break, and atopy (the tendency to be

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F.I.R.S.T. Proudly Celebrates our 20th Anniversary

Thank you to our friends and supporters for helping us reach this important milestone. Please be assured that F.I.R.S.T. will continue to do its best to earn your continued confidence and generous support. It is our hope that our relationship with you will continue for many years to come. See pages 8 and 9 for a timeline of important events in our 20 year history.

Family Conference Mark Your Calendars!

Family conferences are a great way to meet other people whose lives are affected by ichthyosis.

Regional Family Conference

April 7, 2001 in Houston, TX (see back cover for details on how to register!)

2002 National Family Conference

The National Family Conference will be held in Seattle, Washington during the summer of 2002. Stay tuned for more details and start planning now so you can join us!

Ichthyosis Focus

Vol. 20, No. 1
Spring 2001

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Request to reprint information contained in Ichthyosis Focus should be directed to the editor.

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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.

Correspondence Corner

Editor,

The latest issue of Ichthyosis Focus (Vol 19, No. 4) was by far the best issue ever published.

The article by Dr. Mary L. Williams was meaty and informative. It was a treat to hear about the topic of ichthyosis directly from an experienced physician, in the Focus. Please keep this in mind for future issues, perhaps with other authors. I do look forward to the remainder of her talk in the next issue, and it is just too bad we couldn't get all of it out in one issue.

In addition to that, I was happy to see that three people wrote letters to the Focus editor this issue. Do you think folks realize how much the readers love hearing their comments, stories, questions, and suggestions?

Although I am settled into my treatment regimes and don't so much need the "Ask the Readers" section, even there I was glad to see the focus of the Focus rightly placed on ichthyosis and not on fluffier topics such as fund-raising, convention photos, shmooze stories, etc. Even the presentation of the Board of Directors in the Focus was informative. Of course, the story by Merritt Andrews showed a lot of work and care given to writing to those of us with ichthyosis. The photos of her made a nice study, and told more than her words, too. The personal stories are by far my favorite part of the Focus.

Even the ads were strongly done and welcomed by me. Apparently there is an increasing amount of products, and increased attention being given to ichthyosis. All of this is good news, and I can only say:

Keep Up the Good Work!
David Morris, San Antonio, TX

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

I was born in Trenton, NJ on January 16, 1918, so my 83rd birthday has just arrived! Efforts to help my ichthyosis vulgaris began with my dad taking buckets of ocean water and traveling 50 miles back to my home in Trenton. He thought the ocean water made it better. The truth was that the hot summer weather perspiration made it appear normal. My own first efforts to find a solution to my problem began with a trip to visit some doctors at Columbia University. I was told to rub clear vaseline on it, which did not help. Ten years ago, a now retired dermatologist at the Medical College of Ohio, had me try a prescription of 12% Lac Hydrin. You can get an over-the-counter bottle of 4% Lac Hydrin. After six weeks I could see how wonderful it worked and made my skin look normal. Ten years of daily use of 12% Lac Hydrin has led me to the following procedure that I would like to share with you. Doctors said take only one shower a week but after extensive reading, I discovered that idea was wrong. I now shower daily, and after a quick toweling, I apply 12% Lac Hydrin on my legs, arms and a dab on my back. Next I take a three foot piece of rope, or line, with a knot at each end. I put the 12% Lac Hydrin in my hands and rub it on the full length of the rope. Then I take the rope and rub it up and down my back. This picks up the dab that I had placed on my back earlier. This process gets the 12% Lac Hydrin all over my back where I could not reach. Then I dress for whatever (3 hours of daily exercise and guitar playing). My skin gets bad in the winter, when the furnaces dry out the house humidity. But otherwise, this skin is just about perfect from head to toe—not too bad for an 83 year old man! P.S. You may print this letter if you wish or use any part of it. Perhaps my years and years of research and thought put into action could help others.

Conrad N. Marriott, Toledo, OH

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

My name is Tracy Wood. I receive your newsletter and I just received a free sample of NeoCeuticals cream from you. I can't express just how happy that made me. I have had ichthyosis since birth and it is so hard to deal with, although I have become alot more understanding of other people's ignorance to this disorder in my 29 years. It was so touching to receive the cream, because it shows me that there are really people out there who care and who want to make life a little easier for those of us afflicted with this disorder. Thank you very much again.

Sincerely,
Tracy Wood, Cleveland, OH

Deadline for Jane Bukaty Membership Assistance Fund: April 15, 2001

The Jane Bukaty Membership Assistance Fund has been established to help members of F.I.R.S.T. with some of the expenses of their skin care. The Support Network and Member Assistance Committee recently granted awards to 5 members of F.I.R.S.T. Applications are now being accepted for more grant awards this spring. Take this opportunity to help alleviate some of the financial burden that may be facing you and your family. The application process is simple and easy to do. Submit a letter indicating your need for funding and indicate the amount of money you are requesting (awards will not exceed $100.00). Please demonstrate your financial need and indicate what product or treatment for which you are requesting the funds.

Email your request to info@scalyskin.org or mail your application to the attention of the Jane Bukaty Membership Assistance Fund at 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446.

Once your application has been received at the F.I.R.S.T. office, it will be submitted to the committee for review. You will be notified by Jean Pickford, Executive Director, if your application is approved.

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.

Discovering Fun and Friendship at The American Academy of Dermatology's Camp

Children ages 10 through 13 are invited to attend Camp Discovery in Crosslake, Minn., July 14-21, 2001. Due to increasing demand, the Academy also uses facilities at a camp in Millville, Penn., to accommodate additional campers. Campers, ages 9 through 13, are invited to the Pennsylvania camp on August 11-18, 2001. Campers are assigned by space availability.

All campers receive full tuition and transportation scholarships through the generous contributions of The American Academy of Dermatology members, corporations, individuals and organizations. Since its inception in 1993, more than 500 children have participated in Camp Discovery.

Referrals for campers must come from a dermatologist. If you would like to attend Camp Discovery, please contact your dermatologist. For more information about Camp Discovery, call the American Academy of Dermatology at (847) 330-0230.

Expanded Role of Registry

The National Registry for Ichthyosis and Related Disorders has expanded its role. While still identifying and enrolling individuals affected with disorders of keratinization, we have obtained funding to allow for molecular diagnosis to identify the specific genetic defect for some individuals who have enrolled and had their diagnosis confirmed. We plan to continue to:

- encourage investigators to study the ichthyoses and related disorders. Since most of these disorders are very rare, it is difficult to find people who are affected and whose disorder can be studied. Our job is to make this task easier.
- find better ways of diagnosing disorders and better treatments than are available now.
- collect as much information and data as we can about every person who enrolls in the Registry. In this way, we can provide an extensive database of anonymous information about how these disorders affect individuals, any other health problems that they may have, other affected family members, and different ways that people try to care for their skin.
- ask about your interest in participating in research studies so we can inform you confidentially about investigators studying these problems, their projects, and how to contact them if you are interested in participating.
- take posters and make presentations at meetings of dermatologists and other scientists each year to educate them about the Registry as a resource and it's availability for their patients.
- publish notices in medical journals to get the word out about the Registry to the medical and scientific communities.
- follow up with any affected family or friends who have asked to receive information.
- try to think up even more ways to get people involved.

Our new tool is molecular diagnosis, which describes a technique used to identify a specific genetic mutation that is the cause of a particular disorder. It is a very precise tool, and it works extremely well when we know where to look for a specific mutation. As you might expect, there are limits as to who can be tested, but it is a definite advance in the knowledge we can obtain and share with many of you who have enrolled in the Registry. In order for the Registry to perform molecular diagnostic testing, certain criteria have to be met. The following "rules" apply. (See table for eligible diagnoses).

1. An individual must complete their enrollment in the Registry.
2. For all eligible disorders, a clinical examination and description by a board-certified dermatologist will be required.
3. For EHK, IBS, epidermolytic PPK, Darier, and H-H, the diagnosis must be confirmed by histopathology (having a biopsy slide read by a dermatopathologist) of at least one enrolled family member.
4. For all eligible disorders, analysis will include genetic counseling.
5. The Registry will not participate in prenatal diagnosis. This will not preclude affected individuals from interacting with the lab performing the molecular diagnosis either directly or through third party insurers.

We have begun enrollment for over 600 people and, of those, over 400 have finished the process. Our best guess suggests that if only half of the membership of FIRST enrolled, we would triple the size of the Registry almost overnight! What a huge resource we could be for finding a better answer to the questions of 'What is this?' and 'What should I do?'.

Won't you consider enrolling? Please call, e-mail, or visit our website. It's not hard to enroll, and the benefits are great. Thank you to everyone who has already enrolled. We appreciate it and will be sending our annual mailing in June. If you have moved or have not heard from us, please contact us and let us know so we can keep your address current. If ANYONE thinks we can be of some help, please call! We will do our very best for you, we promise!

To contact the registry, call 1-800-595-1265.

Disorders in which mutation detection or diagnostic testing will be undertaken by the Registry:

TYPE OF DISORDER	ASSOCIATED GENE
Keratin Mutations	
Epidermolytic hyperkeratosis (EHK) (includes extensive epidermal nevus (EEN) with epidermolytic change)	KRT1 and KRT10
Ichthyosis bullosa of Siemens (IBS)	KRT2e
Epidermolytic palmar/plantar keratoderma (epidermolytic PPK)	KRT9
Pachyonychia congenita (PC)	KRT6a, KRT6b, KRT16, and KRT17
Other Mutations	
Erythrokeratodermia variabilis (EKV)	GJB3 (Cx31)
Darier disease (Darier)	ATP2A2
Hailey-Hailey disease (H-H)	ATP2C1
Lamellar ichthyosis (LI)	TGM1
Netherton syndrome (Netherton)	SPINK5
Sjögren-Larsson syndrome (S-LS)	ALDH10 (FALDH)

(Blood) Serum Testing
Confirmation of X-Linked Recessive ichthyosis will still be done by measuring the level of cholesterol sulfate in serum.

Executive Director's Report

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

Thank you to everyone who supported the foundation through the Holiday Mailing appeal. Once again, our membership has responded generously. Your financial support is very much needed and appreciated.

F.I.R.S.T. welcomed two new board members since the last newsletter. Dr. Philip Fleckman, Principal Investigator/Director of the Registry for Ichthyosis & Related Disorders and Associate Professor of Dermatology at the University of Washington School of Medicine in Seattle, WA has joined our board. Also joining our board is Louis Giuliana, whose daughter is affected with Epidermolytic Hyperkeratosis (EHK). Mr. Guiliana is a Director in the Careers Center at Holy Family College in Philadelphia, PA.

F.I.R.S.T. will have a booth at the American Academy of Dermatology's annual conference in Washington, DC on March 2-7. I will be attending the 5-day event with our informational literature to distribute among the 17,000 dermatologists, researchers and geneticists who will be in attendance. As in the past, the foundation will host its annual meeting of the Medical Advisory Board on Saturday, March 3, 2001 at the AAD conference.

F.I.R.S.T. is a member of the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR) and the month of March will be busy with activity. First, I will be attending the annual meeting of the CPA-SDR on March 4 in Washington, DC. Then on March 26, I will be visiting the skin laboratories at the National Institute of Arthritis, Musculoskeletal and Skin Diseases (NIAMS), a branch of the National Institutes of Health (NIH). I will also be meeting with Dr. Steve Katz, the director of NIAMS and having dinner with representatives of CPA-SDR, the American Academy of Dermatology and the Society of Investigative Dermatology. On March 27, I will be heading to Capitol Hill with other representatives of the CPA-SDR to lobby for an increase in research money for NIAMS.

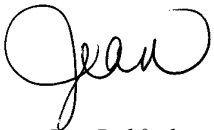
If you have access to a computer and the internet, I encourage you to visit our website (www.scalyskin.org) You will see that we have made some changes and added more information. F.I.R.S.T. believes that our website will become an increasingly important way to disseminate information to our members and is likely to be the initial way in which many potential members become aware of our existence.

I hope you received samples of products from some of our pharmaceutical partners during the past year. F.I.R.S.T. works with these companies to disseminate products that may aid in your comfort or skin care. We send these samples to you as a service and we carefully manage the mailing to protect your confidentiality. Many of you have written to me sharing your positive results from trying these samples. I'm glad we're able to help.

This past January, we had a personnel change. Lori Schreiber, the former Program Director, left the foundation to pursue a new opportunity. Lori was instrumental in getting our Ichthyosis Support Network (ISN) up-and-running again. She will be greatly missed. Lori handed over the reigns to Maureen Tierney, who joined F.I.R.S.T. as our new Program Director on January 19. Maureen comes to us with a lot of experience and will be a great asset to the foundation *(see page 12).*

I am very excited to begin working on plans for the 2002 Family Conference. Thanks to the Site Selection Committee (Les Avakian, Lori Florian, Glenn Oclassen and Dr. Mary Williams), our next conference location has been chosed as Seattle, WA. I will be working with the hotels to choose the best dates and facility for our members. The conferences provide you with an opportunity to meet others affected by ichthyosis. So start to plan now so you can join us in Seattle.

Sincerely yours,



Jean Pickford

ASK THE READERS: What Do You Do?

At the national office we continually receive questions from our members about many different issues. We know that many of you have your own "tried-and-true" methods for your skin care. We also know that many of you have great answers to those difficult questions that are frequently asked by the general public.

We have developed **ASK THE READERS:** What Do You Do? to share this valuable information with others. Each issue will ask a question of our readers and we will print your answers. If you have any advice you would like to share, please take a moment and send in your comments.

In response to your Winter 2000 question...How do you keep your skin moist?

I would like to share a recent discovery of ours: Mango Body Butter for very dry skin. This product is truly amazing! In three days, we saw visible results. It softens thickened skin allowing it to exfoliate without harsh hydroxy acids. It does not sting and smells mango-licious. I butter my son up twice a day after his shower/bath. I can't say enough about this product. It is available at a chain called The Body Shop and comes in various scents. It costs about $13.00 a tub and lasts about one week. I am in the process of contacting the headquarters to inquire if it may be purchased in bulk for a greater savings and convenience. The products not only keeps my son's skin super moist, it succeeds in eliminating any unpleasant odor commonly associated with the build up of dead skin. I love this stuff...it is awesome. **Lisa D., Medfield, MA**

PS Neighbors and relatives have also commented on how great my son's skin appears, despite the harsh winter climate. My son even butters himself and no longer resists his "creaming up."

Hi FIRST!

I have the EHK form of ichthyosis and when I was a mere 2 months old, my mother called my Greek grandfather in the middle of the night. I was crying and so was my Mom. My wise grandfather suggested she rub olive oil over my skin...that was the first time I slept through the night. Our 2nd daughter was born with EHK and I told the doctor about the olive oil incident...he thought that would make a great "liquifyer". As soon as we used it, the raw redness disappeared. Obviously, using olive oil would be too greasy and too smelly, but I have recently come across a fantastic soap "Made in Greece" distributed by Kiss My Face, which is olive oil based. I have only used this product for 3 weeks, and already my skin is softer and just as clear as if I were using prescription drugs! Check out their website kissmyface.com I have also used Albolene Cream Liquifyer for as long as I can remember after my bath time. **Andrea D.**

My son is two and the way we keep his skin so moist is putting Crisco oil in his bath water and putting Crisco shortening on his body every two to four hours. **Paula W., Belleview, NE**

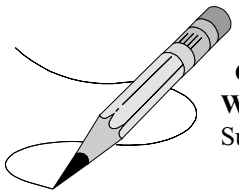
I have X-linked ichthyosis. My 12 year old grandson also has it. I suggest he tell others, "It's just dry skin, not catching. Most people's dry skin powders off or washes off, ours tends to stick and look different, that's it." I use baby oil in my bath water, apply Aqua Glycolic lotion daily, count my many, many blessings and live with it! (I'm 74) **Mr. Weaver, Garland, TX**

The best way we have found to keep my grandson Morgan's skin moist is to bath him twice a day using a soap called "oatmeal and honey" that is made by The Soapmeister. It is a specially made soap made from oils of olive, coconut, cottonseed, soybean, oatmeal, honey and most importantly tea-tree oil…the website is www.soapmeister.com and is very informative about the different kinds of soaps and reasons used. This seems to moisten his skin and lift scales from his head that are difficult to remove. Of course, after the bath we apply Aquaphor ointment. This keeps him very soft and huggable. He is 16 months old and we try and make the bath time as fun as possible. **Jennetta, the grammie, Muldrow, OH**

Here's our next question:

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

Send us your comments. We will print the responses in the next newsletter. Here's how to contact us:



E-mail: info@scalyskin.org
Fax: 215.631.1413
Call: 800.545.3286
Write: 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446

"In general, the goal in taking care of ichthyosis is to hydrate (moisturize) the skin, hold in the moisture, and keep scale thickness to a minimum."*

Foundation for Ichthyosis & Related Skin Types, http://www.scalyskin.org

Aquaphor® Healing Ointment helps heal dry skin associated with ichthyosis. Its unique petrolatum-based formulation combines a moist environment with the benefits of a semi-occlusive barrier that allows skin to breathe and absorb fluids.

Aquaphor is ideal for daily use because it is hypoallergenic, non-comedogenic, fragrance and preservative-free. Aquaphor Healing Ointment is safe enough for even the most sensitive skin.

Special Offer

For Readers of this F.I.R.S.T. Newsletter

MANUFACTURER'S COUPON | EXPIRES 12/31/01

Save $2.00 on any Aquaphor® Product

(NO TRIAL SIZES)

Consumer: Coupon good on the purchase of any Aquaphor® Product, no trial sizes. Limit one coupon per item purchased. Any other use constitutes fraud. Not good with any other special offer. Consumer must pay sales tax.
Retailer: You are authorized to act as our agent to redeem this coupon and we shall reimburse you at face value plus 8¢ handling in accordance with our redemption policy. Cash value 1/100¢. Copies available upon request. Offer void if copied and where prohibited, taxed or otherwise restricted. Mail to: Beiersdorf Inc, PO Box 880504, El Paso, TX 88588-0504. Good only in USA.

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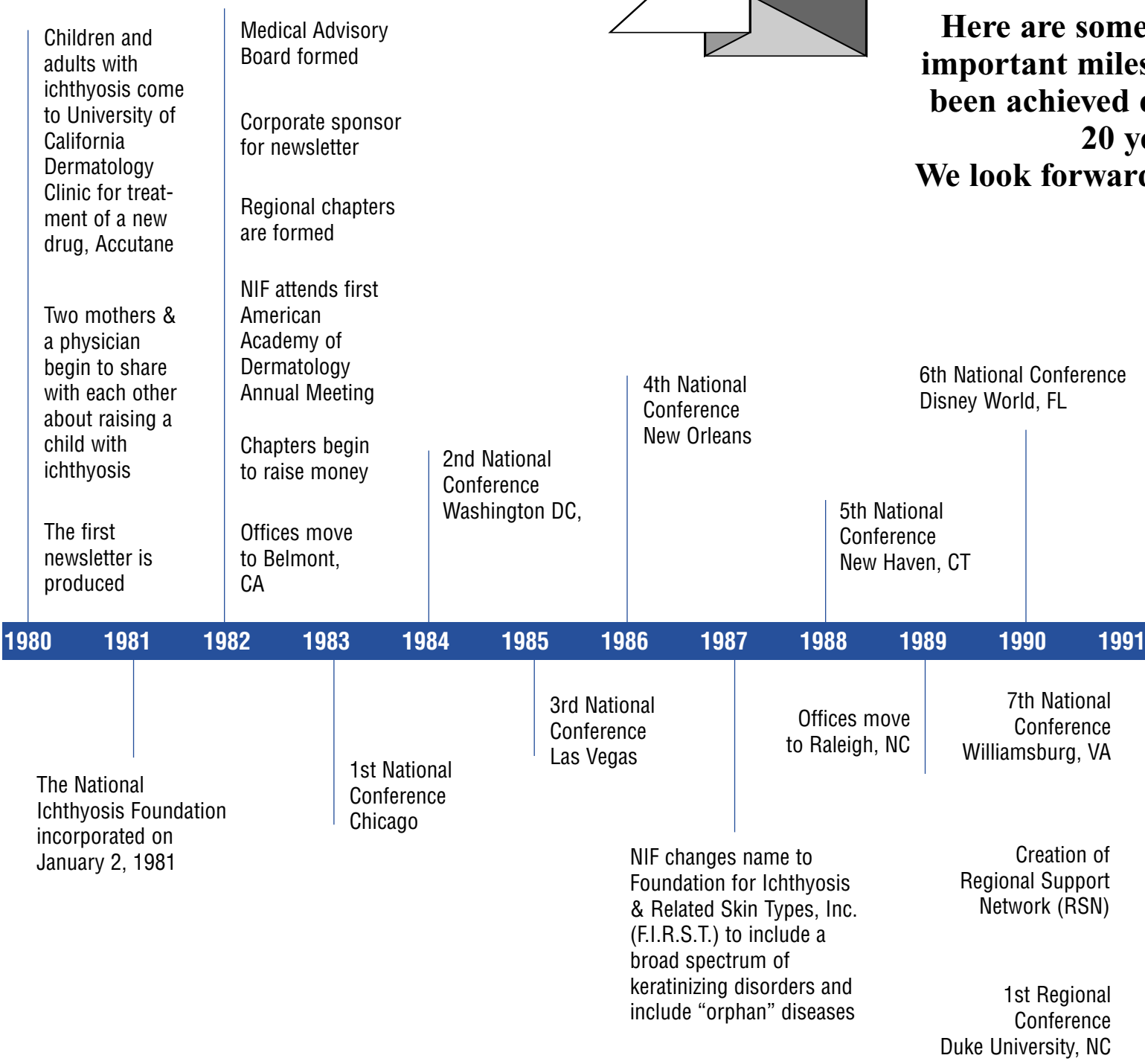
Congratulatio

Here are some important miles been achieved 20 y We look forward



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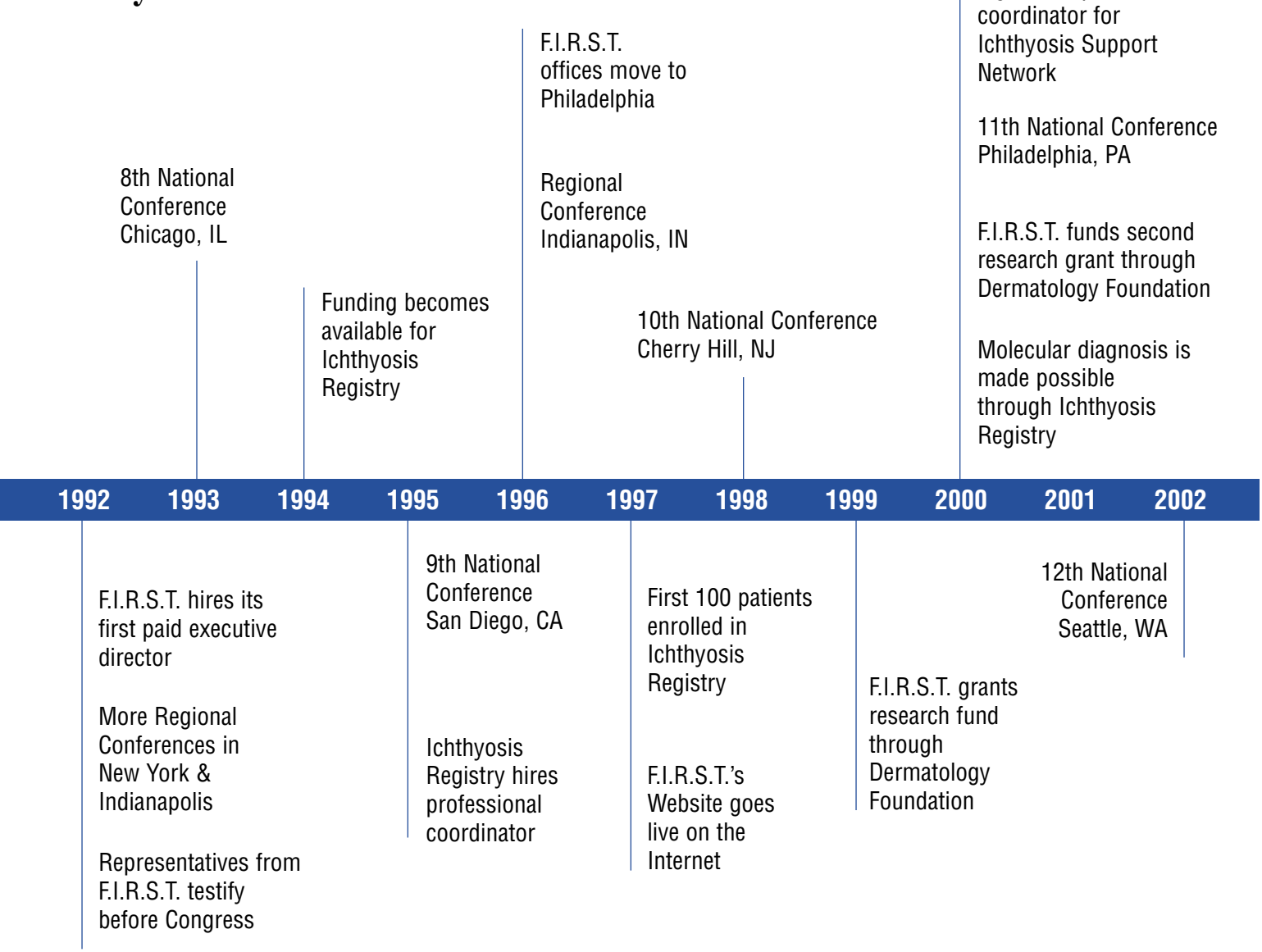
of F.I.R.S.T.'s ones that have uring the past ars. to many more.



Spotlight On...
Gerri Farnworth

Photo courtesy of The Reinbold Gallery, Anaheim, CA.

My daughter's name is Morgause (pronounced MORE-GAY-SAH) and she is four and a half months old. It has taken a lot of determination, sheer will, prayer and your basic hoping for a miracle to get her this far. She is amazing to us…if I had been her, I would have given up by now but she trudges on, slowly making herself the center of everyone's universe that she comes into contact with. I never cease to look upon her with wonder.

She was born May 17, 2000 in the year of the dragon. Normal vaginal delivery…everything fine. The only thing I remember the doctor saying before she had actually been delivered was that she appeared to have a scratch on her head. When she finally was delivered, all were dumfounded as to what to say, much less what it was. It looked to me like she had been stripped of her skin, like she had been burned with acid. I cannot properly explain the emotion that comes over you when you are witnessing the happiest moment of your life and in the next moment filled with a dread that defies explanation. You have tunnel vision, all around you is gone. For a split second all there was in that room was my baby and I. I held her briefly, for they wanted to get her into the neonatal intensive care unit (NICU) as soon as possible, and I looked at her quiet, lethargic little body so wracked with pain. Immediately a wave of grief swept over me…liken it to panic, dread and a feeling that she was going to die right there in my arms all rolled into one. I think back to how very helpless I felt that there was nothing on God's green earth that I could do for her to make it better.

She was sent to the NICU and there we went right behind her hugging her incubator, crying and trying to comfort each other. No one was notified of her birth for over 48 hours, because we couldn't bare it.

She was transferred to Children's Hospital of Orange County and a skin biopsy was done to diagnose what she had. In the meantime, the nurses improvised to make her as comfortable as possible. It was the longest week of my life. We found out through the biopsy that she had Epidermolytic Hyperkeratosis, a form of ichthyosis…that it was a lifetime sentence and she would never "get over it."

There she remained for almost a month. Because the epidermis layer of skin was completely destroyed and what little was left was coming off or was covered with blisters, her temperature kept dropping. She couldn't keep any liquids in her body, so she had a hard time urinating. I heard talk of her kidneys failing because of the liquid loss through her skin. We noticed that her heart rate was erratic, jumping up and down. Because of all these problems they wrapped her up in what looked like a mummy suit. All we could see were her eyes and mouth. These bandage changes took 4 1/2 hours to complete. She was kept on morphine around the clock and it was doubled (sometimes tripled) during the bandage changes. She screamed senselessly during the simplest of things like a diaper change. They would often have to give her a dose of morphine before the diaper change was done. We were not able to hold her for over a week after she was born and then only on an air mattress to cushion our arms from hurting her.

We were at her side everyday of her stay. Don, my husband, sometimes slept in the car (he works nights) just so he could come up and see her now and then. There I learned how to do a sterile diaper change, change bandages, lance blisters, look for infections, bath her, hold her, lift her, feed her…you name it. I had to learn how to be a mother all over again, an ichthyosis mother. So I am part nurse, part maid and part mother. Not much time left to play and love on her. She was finally able to come home on June 12, 2000. And so it begins…

She screamed for almost 24 hours straight almost everyday for a whole month. Don works nights, I work days so we were taking care of her in shifts and surviving on very little sleep. Our jobs have suffered some, we were absent and tardy often, but they were very understanding. (The people at my work even raised over $1600.00 to help with expenses.) We had no other option. She is too expensive for one of us to quit our jobs and we couldn't exactly take her to a daycare, due to the possibility of infections. On top of all that, we were keeping doctor's appointments and all the other things that had to be done on a daily basis for her care. Morgause also has GERD, gastroesophageal reflux disease, so she is constantly vomiting on herself and her clothes. I was always soaking something! GERD causes problems with her ichthyosis as well. She would stop eating and her skin would fall apart. She has actually gone back to the hospital twice because of it. We have her on a high calorie diet now, so she isn't losing as much weight. She requires a lot of calories to re-create her skin every two weeks, so even though this probably would not have been such a problem with a normal baby, it was a big problem with Morgause. Another problem with the constant vomiting is that it would get caught in her neck. Aside from the blistering, thickening of skin, scaling and extreme dryness, there is also a problem of the skin getting too moist. If it does get too moist in the crevices of her skin, it will split open to the dermis level causing her pain and potentially an infection. So we are very careful to keep the vomit out of her neck area. Morgause's skin also rips like paper, which we found out the hard way. Don lifted her up with his hands on both sides of her chest leaving her skin ripped open where his thumbs had been. Even barely bumping her can rip her skin.

This has been a learning experience for us and we are constantly finding new things out about it.

Morgause has this disease but she is getting better. Little steps. We are overjoyed with even a little progress. She is blistering less, her skin is starting to keratinize and thicken. The scaling is becoming more noticeable but she seems to be a little more at ease than she was.

She is an amazing child. She blows my mind each and everyday. She smiles all the time, laughs too. She is starting to mimic sounds that we make and grasp on to things. Her progress is remarkable. She has changed our lives in a way that no one around us thought would happen. Not once have we felt sorry for ourselves, even though this has been a strain on our family and destroyed us financially. She has given us a gift that we would have never seen otherwise. I now realize how fortunate my life is. We never realized how many dear friends that we have and they have been a great support to us. I appreciate my family so much more, as they have rallied around us since the day she was born. My 13 year old daughter, Veronique, has been a constant help to me. She can do everything from changing a sterile diaper to mixing her formula. My father, stepmother and sister all came to offer their assistance two days after she was born and they live out of state. I had a life all along that I always dreamed of, but it took her to make me see that.

I wish you could see her, she has such light inside her. She comforts us instead of the other way around. She makes my life worth living. I wake up in the morning and go to sleep thinking of her constantly. I am so fortunate to have her and I tell her so everyday. I never dreamed I would give birth to my hero…and so she is…and so she shall remain.

Smurfit-Stone Container Corporation, for whom Gerri works, will also be running an article on her efforts to raise awareness for ichthyosis. The article will appear in the June issue of Smurfit-Stone's company magazine, Connections, which has a circulation of close to 50,000 and is produced in both English and French versions for employees and friends of the company worldwide.

Genetic Heterogeneity

continued from page 1

allergic). (Although its genetic cause was not known at the time of this presentation, in June of 2001 the underlying mutation affecting a serine protease inhibitor, SPINK 5, was reported. This mutation presumably results in increased activity of some proteolytic enzymes in the epidermis). In the past decade we have come to recognize that these infants commonly present with the picture of a red scaly baby with failure to thrive. These infants are at significant risk for mortality due to fluid and electrolyte imbalance, and severe infection (septicemia). The correct diagnosis is often delayed, because the diagnostic hair abnormality of "bamboo hairs" may not develop until later on. An earlier diagnosis may be permitted by analyzing electron microscope changes in the stratum corneum. Also skin biopsy can be helpful in distinguishing this entity from other causes of red scaly babies with failure to thrive, such as severe congenital immunodeficiency or severe metabolic or nutritional disorders, such as cystic fibrosis. (Finally, with definition of the underlying genetic cause, diagnosis at the genetic level is now possible.)

New Treatments?

Sjögren-Larsson syndrome is characterized by ichthyosis, spasticity and mental retardation, and is caused by an enzymatic defect in fatty alcohol metabolism. Previous attempts to treat the disorder with dietary modifications have been discouraging. In this report, 2 patients were treated with a low fat diet supplemented with both medium chain triglycerides and essential fatty acids. One patient, in whom treatment was begun in infancy, showed significant clinical improvement on the diet, accompanied by reduction in blood fatty alcohol levels. The authors urge that efforts at dietary treatment not be abandoned and early treatment may be critical. If so, it will be important for physicians to recognize the cutaneous phenotype prior to onset of neurological symptoms.

Topical n-acetylcysteine was proposed as new treatment for ichthyosis[3]. The report concerns use of a 10% emulsion with significant improvement in one patient with lamellar ichthyosis, in comparison to vehicle. The authors note that n-acetylcysteine is an antioxidant thiol, previously used to thin pulmonary secretions and to protect skin after radiation therapy. While the authors propose that it works by inhibiting keratinocyte proliferation, this may well not be the primary mechanism of action. Whatever the mechanisms, we need safe, non-irritating and effective topical treatments for the ichthyosis. If this proves to be the case for n-acetylcysteine, it will be a welcome addition to our therapeutic options. It must be keep in mind, however, that to date only a single patient has been treated and reported in the literature. Until further evidence of safety and efficacy are forthcoming, this should be considered an experimental, non-FDA approved treatment.

Keeping Up:

This is a fast moving field. We all want to give our patients the best and latest information. One mechanism is to access the frequently updated, Online Mendelian Inheritance in Man (http://www3.ncbi.nlm.nih.gov/omim/) — the online version of mcKusick's text. Then, to find out whether and where genetic or enzymatic testing is available, you can inquire though the national registry, Genetests (Genetests@genetests.org). However, one of the frustrations of the past few years has been general unavailability of diagnostic testing for genetic skin diseases. New genes have been identified, often to great fanfare, but with only a couple of notable exceptions testing has not been available other than on a research basis. It is with great enthusiasm that I report that help is on the way. A new company GeneDx (GeneDx@genedx.com) has just been launched that will make genetic testing for many of our orphan skin diseases commercially available. (Finally, since this presentation, it was announced that the National Ichthyosis Registry has received renewed funding by the National Institutes of Health; the Registry will be able to do genetic testing on enrolled subjects who have one of the ichthyosis types in which the genetic defect is known.)

Making Sense Of All This:

How can we understand the incredible array of diverse genes causing scaling skin diseases? We've come a long way from the "bricks and mortar" model of these diseases, in which they were attributed to defects either affecting the structural proteins within the cells of the stratum corneum (the "bricks") or the lipid membranes between the cells (the "mortar"). Now, in addition to mutations affecting structural proteins and enzymes of lipid metabolism, we must also consider defects in cell to cell communication, and the regulation of differentiation and proliferation, and even gene transcription and translation. I find it useful to think of the epidermis as having a limited injury response. It can fall apart, as it does in epidermolysis bullosa simplex or Hailey Hailey disease. It can eliminate a bad cell, or "apoptose", as it does in Darier's disease. Or it can become hyperkeratotic (develop a thickened stratum corneum), as it does in the ichthyosis and keratodermas. Sometimes it does so purely through a failure to desquamate (outermost cells fail to shed normally), but more often there is a component of hyperproliferation (i.e., overproduction of stratum corneum cells, usually accompanied by incomplete maturation of these cells). This hyperproliferation could be a primary abnormality, as caused perhaps by abnormal cell communication, as in the connexin disorders; or it can occur secondarily through a primary defect that results in a faulty skin barrier. When the barrier is impaired, homeostatic signals are generated that instruct the epidermis to make more cells, as a means to overcome the barrier defect.

The Future?

We can soon expect to have finished finding the genes that cause the remaining unmapped ichthyoses. Then we will need to turn to the task of better understanding the pathogenic sequence from genotype to phenotype. This will be important not only for our understanding of these, often rare, genetic diseases, but will also teach us a lot about how the epidermis works. At the same time, our attention should turn to better treatments, based not on serendipity, but on designer therapeutics. This rationalistic approach will include gene-based therapies. All of these advances will be dependent on access of investigators and patients to one another. This can be accomplished through support by clinicians and patients of the National Registry for Ichthyosis and Related Disorders (info@skinregistry.org or 800-595-1265).

1 Laiho et al, Eur J Hum Genet &:625, '99
2 Heenies et al, Am J Hum Genet 52: 1052, '98.
3 Redondo and Bauzá, Lancet 354: 1880 '99.

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

It has been a pleasure to work for the foundation over the last several months. I have gotten to know many of you either in person at the family conference or through email and telephone calls. I enjoyed every minute of that time. An opportunity for new challenges has occurred in my life and I will be moving on to a new position elsewhere. I have been fortunate enough to meet the terrific new Program Director, Maureen Tierney, which makes this move just a little easier. My hope is that I will continue to have contact with the foundation in the future and will continue to hear of its successes. I wish all of you happiness and good things for the future.

Lori

Lori Schreiber

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

Hello everyone. I am so pleased to be a part of F.I.R.S.T., and am working to pick up where Lori left off.

I am very glad to be working as an educator and advocate for you and your families. My background is in the health sciences. I spent many years working as a medical technologist, and I hold a Master of Science degree in Health Education. My last position was Juvenile Arthritis Programs Coordinator for the Eastern PA Chapter of the Arthritis Foundation. My responsibilities encompassed many of the same things I will be doing for F.I.R.S.T.; information, support, advocacy, and helping with family conference planning. I also had the privilege of developing and directing Camp JRA: Juveniles Reaching Achievement, a residential summer camp program for children with rheumatic diseases. That was truly an awesome experience. If you are considering a special needs camping experience for your child, I highly recommend it. See page 5 for more information about camps.

My husband, John, and I will celebrate our 20th anniversary this spring. We have two great kids, a fifteen-year-old daughter and a twelve-year-old son. Maybe you'll get to meet them at the family conference, Summer 2002. I look forward to talking with, and meeting, many of you and your families.

Maureen

Maureen Tierney

ISN Education & Training Listening Skills

A volunteer peer coordinator's first responsibility to someone calling for support or information is to be a good listener. Feeling that they are being heard helps the caller to 'hear' their real concerns, as well as some possible solutions.

BEING A QUALIFIED LISTENER:

A good listener turns their whole attention to the other person, filtering out your own thoughts or environmental factors that may prevent you from paying complete attention.

The accepting listener hears what the caller is saying and respects what the person is saying even if they don't agree.

The active listener makes sure they understand by re-stating or summarizing the problem. Letting the caller know that you understand not only the words but also the feelings of what they are trying to convey.

- Re-stating the problem enables the caller to clarify anything that may have been misunderstood.
- Ask open-ended questions and show a genuine interest in the answers.
- Recognize that sometimes the silence and empathy are the best kind of support.
- Don't impose a solution on the caller, facilitate the caller to problem solve.
- Don't ask questions that require yes or no answers, one-word answers enables the listener to talk more than the caller.
- Avoid interrupting, changing the subject or using clinches.

For example, if someone is worried about their daughters latest crisis don't say "Oh she will be fine" (even if you think she will be fine). Discuss exactly what she is worried about and why, this will lead the person to use problem-solving skills to come to a positive conclusion.

THE CALLER BENEFITS FROM EFFECTIVE LISTENING BY:

- feeling accepted and not judged.
- building a rapport and increased comfort level with the volunteer.
- gaining a clearer understanding of what their "real" issues are and how to address them.
- realization that they are not alone and that there are successful ways to cope with a difficult situation.
- decreased anxiety levels.

F.I.R.S.T. Members Continue to Raise Awareness

Even after Ichthyosis Awareness Week this past October, members are still working to raise awareness about ichthyosis and fund raising for F.I.R.S.T. These events are a great way for the public to learn about and understand ichthyosis. F.I.R.S.T. is very appreciative of these efforts and encourage other members to get involved. Contact the national office for advice on planning a fund raiser or raising awareness through your local media.

F.I.R.S.T. would like to thank and recognize:

Denise Benedetto and her son, Marc, who participated in a one-day bake sale at school that turned into a two-day event! With the help of the Site Based Management Team at Francis X. Hegarty Elementary School in New York, the principal, Gloria Maffettone and the school nurse, Barbara Gould, this event raised over $2300 for F.I.R.S.T. Denise's main goal was to educate the children, teachers and community about ichthyosis and she succeeded!

Shauna and Ryley Johnson in Ogden, UT for raising over $500 by participating in the "Release the Butterfly" fund raiser. A story about Ryley was featured in their local paper and two people in their area with lamellar ichthyosis contacted them.

Nancy Noe in Roseville, MI for raising awareness about ichthyosis by participating in the "Release the Butterfly" fund raiser. Apple Annie's Restaurant and Roy O'Brien Ford sold the butterflies and combined donations totaled over $250. Nancy was also successful in raising awareness about F.I.R.S.T. and ichthyosis with a feature article in her local newspaper.

Photo courtesy of Douglas County Post-Gazette

The students at Skyline Elementary in Elkhorn, NE for collecting over $100 in donations during a lunch-hour sale of lollipops. This was an amazing feat since the lollipops sold for only 25 cents each! The donation was presented to Matt Gray, a fourth-grader at the school and his mom, Beth. The support from Mr. Baere, the principal and staff at the school was terrific and this event was publicized in their local paper.

On April 16, **Carl Whalen Jr.** of St. Mary's, PA will be running for F.I.R.S.T. in the Boston Marathon. This is Carl's 6th marathon and his second trip to Boston. Joining him will be **John Sherry**, also of St. Mary's, who is running his 5th marathon and **Dave Cavalline** of Ridgway, PA, who is participating in his 4th marathon. These men are raising awareness about F.I.R.S.T. and collecting donations in their honor for the event. If anyone is interested in making a donation to show your support of these runners, please contact the F.I.R.S.T. office at 215.631.1411.

Lori Florian in Boulder City, NV for participating in The Festival of Trees in her local community. Lori worked the Santa booth for two days, sharing time with the Down Syndrome Association. Both organizations split the proceeds and F.I.R.S.T. received $200. Lori was able to pass out information about ichthyosis and F.I.R.S.T. during this event.

Thank you to all our members who raise awareness about ichthyosis and the foundation. Keep up the good work!

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Eugene Van Scott, M.D.

A sincere thank you to everyone who made a donation to F.I.R.S.T. in 2000. Our success would not have been possible without your support. F.I.R.S.T. looks forward to your continued commitment and financial support in the future.

Regional Conference Houston, Texas

FIRST is sponsoring a one-day, regional conference in the Houston area on Saturday, April 7 from 10 a.m. to 5 p.m. This conference is designed to give FIRST members in this region an opportunity to meet others affected by ichthyosis and to gain additional insight into their disease; however, it is open to all FIRST members. The program will include a morning session about treatment, research and genetics, as well as a question answer period. Moderated discussion groups will convene in the afternoon after a picnic lunch in the park (weather permitting). Registration fees of $10 per person (children under 3 are free) include conference materials, children's activities and lunch. Conference attendees may also attend a 5:30 p.m. dinner and social time together at a local restaurant. This is optional and not included in the registration fees.

Pre-registration is required and must be received by Friday, March 30. Please complete the form below and mail with your check made out to F.I.R.S.T. to 5160 Franz Road, Suite G, PMB #102, Katy, TX 77493.

If you would like additional information about this conference please contact the national office at 800-545-3286 or email Donna Rice at drice@pdq.net.

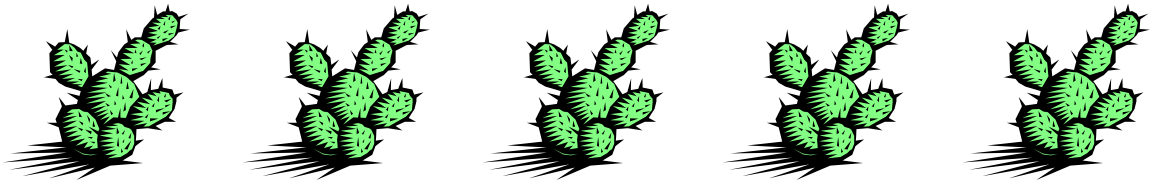
Hope to see you in the Lone Star State!!

Name: _____

Address: _____

Phone: _____

Number of individuals attending, _____ adults, _____ children



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