



# ICHTHYOSIS FOCUS

Vol. 14, No. 1

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

Winter 1995

## RESEARCHERS AT NIH LOCATE GENETIC BASIS FOR LAMELLAR ICHTHYOSIS

*Researchers trace gene for lamellar ichthyosis to a site on chromosome 14 which controls production of the enzyme transglutaminase 1*

In Collaboration With Egyptian Researchers, The NIH Team Traces The Genetic Defect To a Skin Enzyme Whose Activity is Critical to Formation of the Outermost Layer of Epidermis, the *Stratum Corneum*

Researchers at the Laboratory of Skin Biology at the National Institute of Arthritis, Musculoskeletal & Skin Disease (NIAMS) of the National Institutes of Health, reported in the March issue of the *Nature Genetics* that they have located the gene whose mutation accounts for lamellar ichthyosis. This culprit gene, which they found on chromosome 14, controls production of an enzyme called transglutaminase 1 (TGMI).

The research team made several visits to Cairo, Egypt, and worked in concert with Dr. Nemat Hashem, Director of the Ain-Shams clinic. Dr. Hashem has assembled a genetic

database over the past 25 years which includes information on more than 16,000 individuals with inherited disorders, including several rare forms of ichthyosis.

"The Egyptian families were important for these studies because of the high rate of intermarriage in Egypt," explained Dr. Sherri Bale, one of the NIH researchers. "This results in increased prevalence of recessive diseases such as lamellar ichthyosis." Having DNA samples from intermarried families helped the initial genetic analysis greatly.

The researchers first used DNA from

- Continues on page 13 -

Laura J. Russell, John J. Di-Giovanna, Geraldine R. Rogers, Peter M. Steinert, Nemat Hashem, John G. Compton & Sherri J. Bale. "Mutations of the Gene for Transglutaminase 1 in Autosomal Recessive Lamellar Ichthyosis." *Nature Genetics*, March 1995.

## SPECIAL NATIONAL CONFERENCE EDITION

SEE PULL-OUT INFORMATION SHEET AND  
REGISTRATION FORMS BEGINNING ON PAGE 8

JUNE 16<sup>th</sup>, 17<sup>th</sup> & 18<sup>th</sup> 1995

San Diego, California

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Please send your letters to us c/o *Ichthyosis Focus* at our office in Raleigh.

# LETTER FROM THE PRESIDENT

by Deborah B. Vilas

Dear Members,

Greetings to you all from the Board of Directors. It may be the middle of winter, but I assure you, June is not that far away. It is with this in mind that I look forward to the upcoming F.I.R.S.T. National Conference in San Diego. Looking forward to the coming conference, I can't help but reflect on the conference I attended two years ago in Chicago.

For me, the Chicago '93 Conference marked the first time I had found myself in the company of so many people with ichthyosis. It was an experience that changed my self perception in many ways. I realized that I was not alone.

Having ichthyosis had always made me feel separate from those around me. It was incredibly uplifting and comforting to speak with others who knew so much about living with ichthyosis. It was liberating to be with people who required no explanation about the redness of my face or wrinkliness of my hands. I felt accepted in an extremely profound way. It was a bit like coming home to a family I had never met.

Granted, attending the conference brought up a bit of anxiety for me. What would these people be like? What would they look like? What would they think of me? Would the experience depress me? However, because I had met several individuals with ichthyosis before the conference, the ex-



perience was less overwhelming for me.

Other F.I.R.S.T. members have also expressed trepidation at the thought of meeting 250 people with ichthyosis. If you are considering attending the conference but have never met anyone else with the condition, and if you would like to meet

someone with ichthyosis before heading to San Diego in June, the Regional Support Network can hook you up with others in your area. You can contact the office in Raleigh for a referral (800-545-3286). If a face-to-face meeting is not possible, speaking with someone else on the phone or staying in touch through letters may do the trick.

If you are the parent of a child with ichthyosis, please know that friendships that are forged between children (as well as adults) at this conference may last a lifetime. Children, more than anyone, perhaps, need to know they are not alone in the world with different skin.

Whether you are a newcomer or have attended previous conference, we look forward to seeing you and your family or significant others in San Diego. Just remember, we were all newcomers once. There will be someone to reach out and welcome you when you arrive. I hope that someone will be me.

With Warmest Regards,  
Deb Vilas

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# CORRESPONDENCE CORNER

*In the last issue of Focus, a letter from Mary Anne Brame asked readers, "What have I got to look forward to?" The question was directed to older adults with ichthyosis, and Mary hoped to promote a dialog among older adults about their quality of life and other issues related to their lives with ichthyosis. Following are some responses.*

Dear Mr. Gattuccio,

This is in response to the letter by Mary Anne Brame. Since I'm turning 60 in a few weeks and have X-linked ichthyosis, I'd like to respond.

First, don't let it ruin your life, and don't let it make excuses for you! My brother passed away this year and ichthyosis made him a life-long bachelor and a hermit during his declining years. Too many times I was tempted to agree with him in saying and thinking that I can't do something because of this skin disease!

I became a civil engineer, got married and have five of the most beautiful children you could imagine. I now have four grandchildren. I went into city government and became a City Manager in 1968. I have been Oshkosh Wisconsin's City Manager since 1976. I thank God for the wonderful and deeply rewarding career I've had so far. I've been given the opportunity to do some things that have made the quality of life better for the citizens in four cities. So please don't think your life is inhibited because of ichthyosis.

Really, the skin condition gets better as you get older. The only slightly distracting thing is that the circulation isn't quite as good as when I was younger and that causes more cracking and discomfort at the fingertips.

All in all, do not let it get you down. Do not let it be an excuse for failure. You can live a full and productive life. As my hero Lou Gehrig said, "I consider myself the luckiest

man on the face of the earth."

William D. Frueh  
Oshkosh, Wisconsin

Dear FIRST,

In response to Mary Anne Brame's letter in the Fall *Focus*: I am 44 years old and I think my lamellar ichthyosis gets better the older I get. The scale production has slowed down as I age and my oil and sweat glands work better as the skin gets thinner. Also, as I age I don't worry about my looks or what others think of my looks as much as when I was young.

Name withheld by request  
Henderson, Kentucky

Dear FIRST,

My daughter and I are so pleased with the new F.I.R.S.T. book, *Release the Butterfly*. We now look forward to publication of *Ichthyosis: A Guide for Teachers* [available now; see back page of this *Focus*]. Whenever we can we both try to educate the public about ichthyosis. Keep up the good work! Happy Holidays!

Dolores Heikkila  
Jackson, Michigan

Dear Nick,

I enjoy reading *Focus* and keeping abreast of all the wonderful work the Foundation is accomplishing. I especially like reading Correspondence Corner, where I keep up with old friends from F.I.R.S.T.

I want to thank you for appreciating my efforts on behalf of the National Registry for Ichthyosis & Related Disorders [see *Focus*, Fall 1994]. I am so pleased that Dr. Fleckman's grant application won the contract to develop the registry. Congratulations to all those who participated in that effort.

I also want to say that while I did speak

*(Continues on page 12)*

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# Learning To Inhabit My Skin

*A first-person account by*

*Cheryl E. Ryder*

“Following is a painful account that I hope will be educational for parents nurturing children with skin disorders. It is also written especially for women with ichthyosis who see something of themselves in this story.”

As the New Year begins, I sit and ponder where I have come in this past year. For a moment, I think that nothing has been accomplished, that it has been a year spend in limbo; but I think a while, and I realize that a great deal of progress has been made. It is this journey that I want to share with F.I.R.S.T. readers.

The following is a painful account that I hope will be educational for parents nurturing children with skin disorders. It is also written especially for women with ichthyosis who see something of themselves in this story. From it all comes a rebirth, a merging of my *self* and my skin for the first time: a self-knowledge and honesty that I have never known; a desire to touch and be touched.

It is the story of my relationship with the therapist who saved my life. It is the story of coming to value my fiancé as I learned to value myself.

It started on New Year's Day of 1994, when I sat my parents down and told them I needed help with my anorexia. My problem was not only that I was too thin—that was a symptom of a larger concern. Instead, I was trying to cope with the vicissitudes of lamellar ichthyosis by seeking control in what I ate. In January I traveled to Florida and packed up my life there, ended a career as a marine conservationist, terminated a five-year engagement, and returned home to Vermont to start over.

It is the new year again and there is so much more to do. I still feel frightened and uncertain sometimes. People say I am too thin and I am still learning, quite literally, to live within my skin. But I believe that I can continue to move forward.

§ § §

When I was young my parents felt that the best way to help with my skin disorder

was to treat me the same as my siblings, so I would not feel different. I know this made sense to them, but the problem was, I *was* different. I never remember anyone once saying the word ichthyosis to me. It was not a family problem; it simply did not exist. As a consequence, I never learned how to talk about my skin disorder openly and honestly.

The message I received was that it was not acceptable to talk about ichthyosis. It was somehow unmentionable. Although they dealt with the physical manifestations with topical treatments, my parents never acknowledged that there was an emotional side to this disorder, which could only be dealt with through open communication. So I was unable to establish a solid sense of self, to deal in a healthy way with intimacy and sexuality in my teenage years or to realize that my skin did not define my self.

§ § §

In spite of this lack of communication and any sort of support network, I managed to get through high school as a three-season athlete, an honors student, a member of band, orchestra, and the student council. I even got the date I wanted for the junior prom, which seemed important at the time.

Then, slowly, everything started to erode. By my senior year I started wearing black all the time and dropped out of many activities. I was sending out all kinds of signs for help, but none was forthcoming.

In this state of mind I went off for four years at a fine liberal arts college in Maine, where I got a degree in Environmental Studies and Art History. I lived a very isolated, lonely and, at time, self-destructive existence, seeking solace in the beauty of the coast and mountains. Unfortunately, at that age appearances are perceived to be very important, without anyone to instill a more meaningful

standard of achievement and self-worth (and unable to define this for myself), I was left feeling inadequate, different, and depressed. Everyone around me was becoming socially active, establishing relationships, and exploring newfound independence. I felt increasingly ashamed and untouchable, more likely to hide behind unattractive clothing and go off by myself.

§ § §

At the end of my freshman year in college, my dermatologist introduced me to Accutane—the drug that would be my “cure.” In retrospect, I would have been better off without it.

With 160 mg/day, I had perfect skin for the first time in my life. I had skin that was better than normal. I could sweat. I liked myself. I felt beautiful and touchable. I felt worthy. But I wasn't ready for the Pandora's box of issues that this overnight transformation opened up. Emotionally, I hadn't caught up with my metamorphosed body.

Then, I would take three months off the Accutane and revert to my previous untouchable, “disgusting” self. Soon, the intervals off Accutane became longer, until I was only on Accutane for three months and off for the rest of the year.

I never had an opportunity to experience my body as a continuum. It was always changing. I could never count on it to be the same. As my physical self alternated, so did my emotional state and self-image. I lived for the times when I could be on Accutane; but, when I was on it, I felt that the body I was presenting to the world was a lie. I felt dishonest and fraudulent. I would have been better off to either be on the drug or off it. That fluctuating existence led to a schizophrenic sense of self.

§ § §

Eventually, I found something that I *could* control: my eating. In a final effort to avoid dealing with my ichthyosis and its emotional manifestations, I became anorexic. By focusing 100% of my energy on eating and exercise, I was able to avoid every issue in my life. If I couldn't have perfect skin, then I would be thinner than everyone else.

I also thought this might deflect attention from my skin problem, or that people might think my dry skin was due to some other disease, something respectable. This worked to the extent that I became very isolated and alone. I removed all of the threats to my self worth. I became an unfeeling, numb person without any emotions, burying everything that hurt until there was little left emotionally or physically.

Somehow during this time I was able to get a Master's degree and es-

...my parents felt that the best way to help with my skin disorder was to treat me the same as my siblings, so I would not feel different. I know this made sense to them, but the problem was, I *was* different.

tablish myself in the field of marine turtle conservation and biology. My research and work took me to Georgia and Florida, the West Indies and Central America, where I frequently suffered from heat stroke because I was unwilling to admit my physical limitations. I also became engaged to a kind, caring man who accepted my skin disorder from the outset. I could never achieve that acceptance of myself. IN spite of the love and support that Steve gave freely, I was unable to engage fully in that relationship.

§ § §

After starving my body to the point of digestive failure, I landed myself in the emergency room in February of 1994. Fortunately, my body somehow

retained sufficient restorative strength to come out of this medical crisis. Also fortunately, I was not allowed to leave the hospital, but was instead transferred to the psychiatric ward.

I asked my therapist what I was supposed to do there. She suggested that I try and sit still with my thoughts. I thought that was the most ridiculous thing I had ever heard, but as I sat there in that room I did have thoughts for the first time in a long time about myself, my life, my relationships, and everything I had never dealt with. That was the beginning of the healing process.

It was a very long time before I attached any emotions to anything I was feeling. When I did, when I confronted a lifetime of buried hurts, I became very depressed. My therapist was with me, and over the past eight months we have worked together to bring me back to

life. We are not doing the work we could have done at age eleven. Because that work was not done then, we have had a much more arduous undertaking. I wish it could have been done twenty years ago.

There are moments that stand out in our journey together. She was the first person to ever use the word ichthyosis with me; the more we used it, the more the word lost its power. There was the day I realized that my parents had simply not been equipped to have a child with special needs, and I cried over all the pain that need not have been. There was the last transition off Accutane, when I allowed myself to shed tears of frustration for the first time—to mourn the passing of my “normal” skin.

One day I realized that maybe “bad skin days” were days when I should treat myself a little better instead of acting self-destructively. Perhaps a bad skin day was not defined by how my

**F**unded through September, 1999 by the National Institutes of Health (through its National Institute of Arthritis and Musculoskeletal and Skin Diseases—NIAMS), the aim of the National Registry for Ichthyosis & Related Disorders is to provide a coordinated means of confidential contact between individuals affected by these disorders and medical professionals who study them. The goal is to enroll as many affected individuals as possible.

Several people and institutions contributed to the formal proposal for the Registry or offered endorsements for the proposal that Dr. Fleckman was preparing. Dr. Leonard Milstone was key among these, as were other members of F.I.R.S.T.'s Medical Advisory Board and executive director Nick Gattuccio. Endorsements were provided by F.I.R.S.T.'s Board of Directors, a number of prominent skin scientists and dermatologists throughout the nation, members of the pharmaceutical industry, as well as the American Academy of Dermatology.

The overriding purpose of the Registry is to improve diagnosis and treatment of the ichthyoses and related disorders. Because the ichthyoses are so very rare, it is very difficult for a single researcher to accumulate a great deal of information about any single type of ichthyosis. The Registry is designed to help surmount this obstacle.

Principal Investigator on this project is Dr. Philip Fleckman, MD, Associate Professor of Medicine, Dermatology, at the University of Washington in Seattle. Co-Investigator and the Registry's pathologist is Dr. Michael Piepkorn, MD, also of the University of Washington. Dr. Cedric Shackleton, of Oakland Children's Hospital, will conduct certain of the chemical studies in his research laboratory. The database itself will be designed and maintained by the Systems Analyst for the Uni-

# THE NATIONAL REGISTRY FOR ICHTHYOSIS & RELATED DISORDERS

by  
Geoffrey W. Hamill, R.N.

Research Assistant for the newly formed National Registry for  
Ichthyosis & Related Disorders

versity of Washington Clinical Research Center, Mr. Ron Ling. He will work closely with me, Geoffrey Hamill, RN, who will be the person of first contact, enrolling people in the Registry.

Dr. Fleckman has established an Advisory Committee to oversee the implementation and operation of the Registry. The Advisory Committee has many key

responsibilities, including suggesting modifications or changes in enrollment criteria over the lifetime of the Registry, reviewing and publishing annual synopses of Registry data to help stimulate research interest in this area, and reviewing research protocols from scientists wishing to use enrolled membership of the Registry.

## REGISTRY ADVISORY COMMITTEE

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- Mary L. Williams, MD, University of California at San Francisco.

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## LEARNING TO INHABIT MY SKIN

(continues from page 5)

skin looked, but by how it felt to me. Whether it was functionally comfortable seemed more significant than whether I could go out wearing a skin exposing, black silk shirt—the sort of fashion item someone with flaking skin dreams of wearing comfortably!

There was a day when I said I enjoyed my skin care rituals, and that I liked the feel of my skin, that it was mine. When I started therapy I told myself that I could not change my skin, but that I could change the way I felt about it. I discovered that the better I feel

about it, the better it gets, and I create a positive feedback loop. Shortly after this, I found myself talking openly about my skin disorder with a new acquaintance. I found that, once the topic of ichthyosis was out of the way, I was free to enjoy being with the person.

It seems to me that now I have freed up resources to focus on more important things. Before, by striving to avoid dealing with ichthyosis, I was consumed by it. Now, by facing it, my ichthyosis has assumed its proper place in my life. Indeed, who would I be

without ichthyosis? I would simply be a person without ichthyosis, and that is all. I have never judged others by their skin, and now I do not judge myself. I am surrounded by images of skin every day. I notice the texture and beauty of skin all around me. I enjoy the feel of skin in the simple act of a handshake, and I am no longer self-conscious about how my hand feels to someone else as they shake mine. This is the first thing that comes between me and other people, but it is not the only thing.

§ § §

It is a new year. I hope to complete this stage of my therapy. I know that I will continue to take better care of my skin. I hope to maintain an adequate

Perhaps the most critical issue in developing the Registry is that of defining diagnostic criteria for each of the ichthyoses and related disorders. Because all are rare, and can be difficult for even trained dermatologists to identify with precision, we will ask that affected individuals see a dermatologist for evaluation. In cases where a sample (i.e., skin biopsy) is taken for microscopic and chemical evaluation, these will need to be sent directly to the Registry. We will also be sending questionnaires to all potential enrollees to obtain information about the onset and course of that individual's case. Following are those diagnoses recognized by the Registry:

- Bullous congenital ichthyosiform erythroderma (BCIE), or epidermolytic hyperkeratosis (EHK)
- Non-bullous congenital ichthyosiform erythroderma (CIE) & lamellar ichthyosis
- Recessive X-linked ichthyosis
- Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD syndrome).
- Chondrodysplasia punctata syndromes
- Harlequin ichthyosis
- Ichthyosis & deafness syndromes:
  - (1) KID (Keratitis-Ichthyosis-Deafness)
  - (2) HID (Hystrix-like ichthyosis with deafness)
- Ichthyosis & trichothiodystrophy (Tay, PIBI[D]S)
- Ichthyosis follicularis, atrichia, & photosensitivity (IFAP) syndrome
- Ichthyosis linearis circumflexa (ILC)/Netherton's syndrome
- Neutral lipid storage disease
- Peeling skin syndromes
- Refsum's syndrome
- Sjögren-Larsson syndrome
- Erythrokeratoderma variabilis
- Progressive symmetrical erythrodermatoderma
- Darier's Disease (keratosis follicularis)
- Hailey-Hailey Disease (familial benign chronic pemphigus)
- Inherited pityriasis rubra pilaris
- Inherited scaling skin disorders lacking specific diagnosis

It will not be easy identifying individuals to enroll in the Registry. We'll work with dermatology societies like the American Academy of Dermatology and the Society for Investigative Dermatology, and with organizations like F.I.R.S.T. Three of

the major dermatology journals have agreed to periodically run information about the Registry, so practicing dermatologists can inform their ichthyosis patients about it. Once the Registry is well established, we'll advertise regionally to reach people who no longer use the established health care system.

CONFIDENTIALITY is the most important cornerstone of the Registry's design. Contact between enrollees and researchers *can only be made by the enrollees themselves*. Studies that use information in the Registry database (for example, epidemiological studies) will be entirely anonymous. Other types of scientific studies will require approval of an NIH-endorsed Investigational Review Board before the plan's submission to the Registry Advisory Board.

When the Registry Advisory Board approves a research proposal to draw on information in the Registry, the appropriate enrollees (for example, those with a specific type of ichthyosis) will be contacted *by the registry only*. The enrollees will be given information on how to contact the sponsors of the approved research project. *Under no circumstances will names or any identifying information about individuals in the Registry be given out by the Registry.*

We would like to thank all of the many individuals involved in helping to obtain funding for establishment of the National Registry for Ichthyosis & Related Disorders. From the early discussions at F.I.R.S.T. through the long process of preparing and shepherding the proposal through all of the required steps, it has been a collaborative effort. We hope that this Registry will stimulate research interest, and ultimately a better understanding of these disorders, resulting in better treatment options and a better quality of life for those affected with the ichthyoses. For more information feel free to contact us

Philip Fleckman, MD or Geoffrey Hamill, RN  
 National Registry for Ichthyosis & Related Disorders  
 Dermatology, RM-14  
 University of Washington  
 Seattle, WA 98195  
**1-800-595-1265**

weight. When I start to focus on my skin or on eating, I know to try and identify what is really wrong. I am working on quieter ways of thinking through my feelings.

I would like to resume a full-time career. If I don't continue with endangered species conservation, I would like to find a way to help women with disabilities or eating disorders. I would like to write something that will help adolescents with ichthyosis as they struggle through the difficult teenage years.

I hope to be married to my fiancé of six years and I would like to have a child when I know that I am healthy enough. I believe that I can do all of

these things. I have rediscovered the sentient parts of myself and the inspiration I have always derived from the natural world. I step out onto the frozen lake outside my cabin here in Vermont on cross country skis and it is snowing. I am alone and I stop and look around me, overjoyed at my *self* here this moment. I'm alive and it feels good.

§ § §

Finishing this article has been difficult. My feelings about ichthyosis continue to evolve and are always in flux. the journey continues and every day I learn more.

I would like to thank F.I.R.S.T. for giving me this opportunity. I am indebted to my parents for doing the best

that they knew how when I was young. I am grateful for my fiancé, Steve Swanberg, for his continued love and guidance. Most of all, I would like to thank Dr. Mary E. Willmuth. In her own words, she seeks to help women with disabilities live their lives more richly and fully. Because of Mary, I know that I can. I look forward to hearing from parents or women who have comments or questions about what I have written. I hope my words can help some of you. Please feel welcome to contact me.

Cheryl Ryder  
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# F.I.R.S.T.'s 1995 NATIONAL CONFERENCE

## JUNE 16, 17 & 18 -- SAN DIEGO

### A PERSONAL INVITATION FROM THE EXECUTIVE DIRECTOR

I want to offer a personal invitation to all of you to join us in San Diego. While putting on our last successful conference in Chicago in 1993, I learned a lot from those who attended about what F.I.R.S.T. members most want out of their National Conferences. On the basis of these experiences, there will be some new features seen for the first time in San Diego.

The biggest change is that this will be the longest-running and most ambitiously scheduled of any previous conference, running over the course of an entire weekend.

Although registration fees may seem expensive, please keep in mind that *the registration cost will include five meal functions* — two continental breakfasts, one luncheon, one dinner banquet, and the opening night reception.

I've already begun scheduling our professional attendees. We have commitments so far from Dr. Philip Fleckman, director of the new National Registry for Ichthyosis; from Drs. Sherri Bale and John DiGiovanna, both of the National Institutes of Health; and I have tentative commitments from several others.

I've heard from representatives of our sister organization in Rotterdam, Holland, and latest word is that five of them will be joining us in San Diego. Please come meet them.

This will be the best and most productive National Conference in this Foundation's history. We're projecting attendance exceeding 300 members, a dozen professionals, and pharmaceutical exhibitors, too. We hope to see all of you there.

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#### Contact Agent: Linda Puckett

The Travel Agents International office here in Raleigh is the official travel & ticket agent for the F.I.R.S.T. National Conference in San Diego. The agency is offering F.I.R.S.T. the following amenities:

- 5% off already discounted/economy fares;
- All bookings held until final 30 days prior to departure to take advantage of fare drops between booking and departure dates;
- Agency donates 10% of commissions to F.I.R.S.T.

### THE 1995 F.I.R.S.T. NATIONAL CONFERENCE

**When** June 16th, 17th & 18th, 1995 (Fri, Sat & Sun)

**Where** The Bahia Hotel & Resort  
998 West Mission Bay Drive / San Diego, CA  
92109

**619-488-0551**

**Room** \$68.00 per night (family rate)

**Family rate** means two adults and all children under 18 stay for the flat \$68.00 rate. For more than two adults (18 and over) in a room, add \$15.00 per person.

This rate is guaranteed for 3 nights prior to, and 3 nights following the scheduled conference dates., for a total of 9 nights (June 13th-June 21st). Note that this special rate on the extended days is on a *space available basis*, so be sure to book your rooms well in advance.

*The F.I.R.S.T. National Conference rate of \$68.00 is only guaranteed for rooms booked a minimum of one month in advance of the conference date — by May 15th, 1995.*

You may book rooms at any time by calling the Bahia Hotel at 619-488-0551.

If you have questions, or if you experience difficulties booking your room, please call F.I.R.S.T. executive director Nick Gattuccio in Raleigh, at 800-545-3286.

### OUTLINE OF CONFERENCE SCHEDULE

June 16, 17 & 18 (Fri, Sat & Sun)

Fri 6/16	Registration	5:00 - 6:30 pm
	Reception	7:00 - 10:00 pm
Sat 6/17	Breakfast & late registration	9:00 - 9:30 am
	Morning session	9:30 - noon
	Afternoon session	2:00 - 5:00 pm
	Banquet & Social	7:00 - 10:30 pm
Sun 6/18	Breakfast & plenary session	9:00 - 10:00 am
	Morning session	10:00 - 11:30 am
	Afternoon session	12:30 - 2:00 pm
	Closing Session	2:00 - 2:30 pm

- For those of you planning on getting flights out of San Diego on Sunday, you may check out of the Bahia Hotel that morning and store your luggage until your departure time.
- Registration on Friday afternoon should only take a few minutes to check in and pick up your Conference materials. If you have a late flight in on Friday, you can pick up your conference material any time that evening or the next morning.



# F.I.R.S.T.

## 1995 NATIONAL CONFERENCE

June 16, 17, 18

Bahia Hotel

San Diego, California

Please Print

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City: \_\_\_\_\_ State: \_\_\_\_\_ Zip: \_\_\_\_\_

Phone: \_\_\_\_\_ Country: \_\_\_\_\_

NAMES OF THOSE ATTENDING

adult child

Age of Each Child

Check here for those affected with ichthyosis

_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>
_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>
_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>
_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>
_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>
_____	<input type="checkbox"/>	<input type="checkbox"/>	_____	<input type="checkbox"/>

Type of ichthyosis, if known: \_\_\_\_\_

Your Registration fees will include five meal functions — two continental breakfasts, one luncheon, one dinner banquet, and the opening night reception. Children's fees include all child care costs.

Number of Adults: \_\_\_\_\_ x \$75 = \_\_\_\_\_  
(18 & older)

Number of Children: \_\_\_\_\_ x \$35 = \_\_\_\_\_  
(4 through 17)

Infants & Toddlers: \_\_\_\_\_ x \$20 = \_\_\_\_\_  
(0 through 3)

TOTAL = \_\_\_\_\_

Please Return Registration Fees With Your Completed Form

Make Check Payable to F.I.R.S.T. In U.S. Dollars, Please

Cancellations Will Be Honored With Full Refunds

UNTIL The Advance Registration Deadline:

MAY 15, 1995

<input type="checkbox"/> I will need child care for _____ children	NAME	AGE
	_____	_____
	_____	_____

<input type="checkbox"/> I am a teen and I'd like to help take care of the children for an hour or two	NAME	AGE
	_____	_____
	_____	_____

<input type="checkbox"/> I am an adult willing to help at the conference for an hour or two (child care, reception table, etc).	NAME
	_____
	_____

<input type="checkbox"/> I am a teen or young adult and I'd like to participate in a discussion group with others my age.	NAME	AGE
	_____	_____
	_____	_____

**PLEASE MARK THE APPROPRIATE BOXES**

- This is the first National Conference I have ever attended
  - I have attended National Conferences in the past (City/year): \_\_\_\_\_  
 \_\_\_\_\_  
 \_\_\_\_\_
  - I will be staying at the Bahia Hotel in San Diego
  - We are combining the National Conference with a family vacation
- We're traveling the following number of miles to attend this Conference (mark the appropriate box)
- 0 - 150     150 - 500     500-1000     1000 - 1500     1500 - 2500     over 2500

For hotel information & reservations, call the Bahia Hotel at **619-488-0551**. Identify yourself as a F.I.R.S.T. member to receive our special conference hotel rate of \$68.00 per night. This rate is good from 3 days prior to the Conference to 3 days following — from June 13th through June 21st.

***Your reservation must be made by May 15th, 1995***

Questions about the conference or trouble booking a room? Call Nick at F.I.R.S.T. at 800-545-3286

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# N.O.R.D. REPORT

*The National Organization for Rare Disorders (NORD) is an umbrella organization representing the interests of groups like F.I.R.S.T. in the difficult arenas of political lobbying and health-care advocacy. "N.O.R.D. Report" is an ongoing digest of highlights from NORD's newsletter, NORD ON-LINE.*

## The GOP & Health in America

Insiders expect the GOP to target government research funds for drastic reductions. The Republicans' "Contract With America" has already identified \$19 billion in science related research that they would like to cut over the next five years. The Heritage Foundation, a conservative think tank, has called for a 15% reduction in NIH appropriations. Such cuts would cripple skin disease research in America.

## The GOP & The FDA

Speaker of the House, Newt Gingrich, has targeted Food and Drug Administration Commissioner David Kessler. At the Fall conference of the Biotechnology Industry Organization, Gingrich called the FDA "the leading job killer in America." Gingrich fails to mention that before the FDA existed earlier this century, unscrupulous "snake oil" salesmen and manufacturers were among the leading *people* killers in America.

According to the *Wall Street Journal*, hopes to redesign the FDA, "dramatically increasing the role of the private sector in the testing and certification process." A leading force in this effort is the Progress and Freedom Foundation, which is funded largely by drug companies.

According to Sidney Wolf, a member of Ralph Nader's Health Research Group, turning the drug and medical device approval process over to the "scientific entrepreneurs" (as Gingrich puts it) "It's not just turning over the chicken coop to the foxes, but getting rid of the chicken coop altogether."

## The Bottom Line on Generics

When the Waxman-Hatch Act was signed into law in 1984 [promoting consumer access to generic drugs], experts predicted that generic drugs would save U.S. consumers \$1 billion per year over the next ten years. They were wrong. Generics saved American consumers five times that amount — \$50 billion over the 10 year period. Generic drugs are now used to fill 40% of all U.S. prescriptions. The number is expected to rise to 50% within the next few years.

## Health Reform: Will It Happen Now?

Health reform does not appear on the Republican's "Contract with America." However, Senator Bob Dole noted that health reform still scores high as a priority on opinion polls. Therefore, "Health care is still a problem," said Dole. "I'm not sure how much we can fix, but we ought to do the best we can."

Sceptics do not expect to see serious attempts at large-scale health care reform.

Meanwhile, Republicans are determined to return power to the states.

Many states are trying experiments with health reform, but they complain that local health reform efforts are being blocked by a federal law called ERISA. This statute permits self-insured employers [usually large, multi-state corporations] to design their own health insurance plans and allows them to ignore state health insurance laws.

Public opinion polls indicate that health reform is the top issue that voters most want the 104th Congress to act on. In order of importance, these issues are: health care (33%), crime (28%), and taxes (23%). [Source: Kaiser Family Foundation, Harvard Univ., KRC Communications, and Voters News Service Polls.] Even a poll by the Health Insurance Association of America (HIAA) indicated that 48% of the public will be disappointed if health reform is not enacted this year.

## Quotes of the Day

• "Most of us have exercised our constitutional right to develop no meaningful opinion about the health care issue. This is a right that can never be taken away from us, even if we change jobs." -Achenbach, Washington Post

• "The Republicans and Democrats sometimes act like little boys playing marbles." - Former Surgeon General Joycelyn Elders

## F.I.R.S.T. WELCOMES NEW "VOLUNTEER"

Nick, Heather and Caitlin Irene Gattuccio proudly announce the arrival of F.I.R.S.T.'s newest volunteer staff member — *Lorenzo O'Dea Gattuccio*. Little Enzo was born December 21st without complication. Heather did wonderfully and everyone came home the next morning. Caitlin has proved to be a top-notch big sister and is completely in charge.

to many legislators of the importance of establishing a Registry, there were many others on the Medical Advisory Board who helped. But most importantly, it was Stephen and Lianne Busby of Avoca, New York, who made the very long road trips to Washington, DC with their three small children with lamellar ichthyosis. In spite of extremely warm temperatures in the nation's capital at the time, the Busbys made personal visits to many Senators and Representatives to introduce their children and speak of the difficulties they've had to overcome in obtaining adequate care for their children in all sectors of their lives.

Please extend the Foundation's appreciation to these and all other volunteers who gave of themselves to benefit all.

Susan Snyder

Raleigh, North Carolina

*Susan Snyder is the former executive director of F.I.R.S.T.*

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

I cannot begin to express how much I enjoy receiving your newsletters. For years I was the only one I knew with this kind of skin. Over and over different doctors would label it ichthyosis, often calling over other doctors to look at it. I often felt like a freak. They were most interested when I had lost control of it.

It runs in my father's family. I do not know what kind we have; we examine our newborns, and if their hands are wrinkled up like an old man's we can be pretty sure they will have it. At the age of 18 months, I was taken from my parents because authorities thought the ichthyosis was a sign of neglect. After about a year, when my skin was worse than ever, they gave up and gave me back to my parents.

I remember my father going to school and talking to the teacher because other kinds did not want to hold my hand when we made a circle and played a game. My teacher was great. She would take my hand and instruct another child to take my other hand. If someone at school became cruel, I would explain to them that God gave them their wonderful skin and He also gave me mine, and they had no reason to think they were better because they did nothing special to get their good skin, and I didn't do anything to get mine. I know I learned tolerance and sensitivity to others at an early age because of my skin.

So here I am, age sixty-five. I had been without serious problems for a few years, but then, with a little stress in my life, and lo and behold, here we go again. Back and forth to the doctor. I belong to an HMO, and they discourage referrals. The internists do not always know what they're dealing with, and they tend to take it too lightly. Finally, I decided I didn't need to have the insurance pay for a dermatologist, and I just went out and paid it and got on with my life.

I am in a new city, and the first doctor that attempted to treat it here told me to use Crisco to "cream" it. He obviously was turned off by the looks of it, but had to act like he knew it

all. That is always a clue—when a doctor pretends that he "has a sure cure." I have found that treating ichthyosis is trial and error that goes on your whole life. While one product might work at one time, something else may work better another time.

I want to thank you at F.I.R.S.T. for all the hard work that I know is involved in what you're doing. Know that you are an encouragement. Just knowing I am not alone is a good feeling.

Joyce Schram

Kentwood, Michigan

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

I am the mother of a 9-month-old who has trichothiodystrophy with ichthyosis. I am interested in receiving your newsletter and any information you may have on our daughter's illness.

She has a problem with a very dry, scaly scalp, and this seems to be her most pronounced problem area. I would love to hear any suggestions for this problem. Also, I'm looking for a parent match and wondered if you could forward my name to anyone you know of who has, or whose child has, this illness.

Julie Creek

Williamsport, Indiana

*As we all know, the scalp is frequently the most difficult of all body areas to treat for those with ichthyosis. We wish to encourage all of you out there with your thousands of years of collective experience treating scalps to share your strategies with Julie and the rest of us here in Focus. -Ed.*

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

*[Editor's note: The following letter is being referred to a member of our Board of Medical Editors for professional review in the next issue of Focus. However, we wished to throw it open to our X-linked ichthyosis community (particularly the carrier moms) for their responses.]*

My husband has recently had his ichthyosis diagnosed as recessive X-linked (RXLI). This, I understand, makes our daughter a carrier. On re-reading the Summer, 1992 issue of *Focus*, I was surprised to read of an RXLI carrier referring to her "symptoms" in her letter on page 6 of that issue.

I was unaware that RXLI carriers have any related symptoms, and so, I would be extremely grateful for any information about possible "symptoms" in our daughter.

Mrs. John Burton

Nottingham, England

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*We welcome your letters to Correspondence Corner and encourage you to write. The Editor reserves the right to edit letters for space and clarity. If you wish your letter to be published without your name, please note this in your letter. Address letters to the Focus Editor at the Raleigh address.*

# NEWS & NOTES

## Water Filtration System Heralded

Member Chandra Foote in Lake Oswego, Oregon, writes to tell us about a water filter system that's had a big impact on her lamellar ichthyosis. "I have recently started using a water filter that connects to my shower pipe. It removes chlorine, ammonia, lead, and other inorganic chemicals. The effects on my skin have been wonderful." For more information you can write Chandra at Box 200-257, Lake Oswego, OR 97035, or call 503-795-9437.

## Sun Protection Hats & Clothing

Mary Fitzpatrick writes to tell us about a clothing company called Sun Precautions, which sells their sun protection hats and garments via cat-

alogue. This "Solumbra" line is made of a special fabric, Mary tells us, "which has a sun protection factor (SPF) of 30—they really work and actually keep the wearer cooler." They are in Washington State, and the catalogue can be ordered at 206-441-6688.

## Pen Pals in Sweden

The Secretary of our Ichthyosis sister organization in Sweden has asked if any of our members wished to have Swedish pen pals. If so, please write:

Agneta Ganemo  
Iktyosforeningen  
Ganarp 2517  
S-260 80 Munka Ljungby  
Sweden

## F.I.R.S.T. SEEKING INFO ABOUT THE OREGON PLAN AND OTHER STATE HEALTH LEGISLATION AFFECTING PEOPLE WITH ICHTHYOSIS

F.I.R.S.T. is seeking out individuals with ichthyosis, *particularly those in the State of Oregon*, who have been denied state mandated benefits (for example, Medicaid or SSI), or have had these benefits reduced or altered because of new state laws or regulations. We are particularly interested in the impact of the so-called *Oregon Plan* on Oregon residents who have ichthyosis and who may have had SSI or Medicaid benefits amended or eliminated altogether. However, similar experiences in other states are also of great interest. Let us know!

Please call Nick Gattuccio at F.I.R.S.T. (800-545-3286).

## RESEARCHERS FIND GENE FOR LAMELLAR ICHTHYOSIS

(Continues from page 1)

both U.S. and Egyptian patients to narrow their search for the lamellar gene to a specific region of chromosome 14. They examined this region of the chromosome because they knew that TGM1 was one of several proteins involved in formation of the stratum corneum. In fact, TGM1 was the eighth such "candidate" gene that researchers screened for a possible connection to lamellar ichthyosis. Results of these initial genetic linkage studies were reported in the December 1994 issue of the *American Journal of Human Genetics*.

Transglutaminase 1 (pronounced *trans-gloo-TAM-in-ace*) serves to cross-link cellular proteins which form a rigid scaffold in the stratum corneum—the flattened layer of "dead" skin cells making up the outermost, surface layer of the skin. These cells initially form at the base layer of the epidermis, then slowly migrate outwards toward the surface. This takes about 14 days. As the cells migrate outward, their structure and shape changes. The nucleus of the "living" cell eventually disappears and the cell membrane is transformed into a "cornified cell envelope." The cells become flattened and ultimately form an overlapping and interlocking fabric—the surface of our skin, the stratum corneum. This complex process of forming the stratum corneum is called "terminal differentiation."

"The terminal differentiation process is somehow abnormal in people with lamellar ichthyosis," says Dr. John Compton, a member of the NIH team. "We now know a major cause of scaling is failure of this one component of the process. We know that the function of TGM1 is to produce crosslinks among cells of the stratum corneum..., but exactly what role these crosslinks play in producing a normal stratum corneum is still a mystery." The research team plans to continue identifying and cataloging mutations that occur in the TGM1 gene in their patients.

As with findings for EHK reported in *Focus* over the past two years, it appears that lamellar ichthyosis may result from mutations at any number of sites along the length of the gene. Drs. Bale and DiGiovanna have already begun trying to correlate symptoms of EHK with specific point mutations in patients. Ultimately, the same kinds of differences in severity and appearance among patients with lamellar ichthyosis may be traced to these patients having mutations at different locations on the TGM1 gene.

Regardless, this work will significantly increase knowledge about the biology of the skin. "The most important function of the skin is to make a normal stratum corneum," says Dr. DiGiovanna. "Knowing more about this will not only increase our understanding of lamellar ichthyosis, but also of many other skin diseases."

# FIRST'S 1994 HONOR ROLL

## A ROLL CALL OF FIRST'S MOST ARDENT SUPPORTERS

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I had reserved this space to report on the outcome of an ambitious fund raising effort conducted by Frances Bernstiel, of Collegeville Pennsylvania. Instead, the space becomes a memorium on her sudden passing this past Christmas week.

A longtime supporter of F.I.R.S.T. (her daughter, Lynne Alba, is on F.I.R.S.T.'s Board of Directors), Mrs. Bernstiel purchased \$10,000 worth of gift certificates to Genuardi's Supermarket in Norristown, Pennsylvania and then sold them to friends, family and supporters in her area. Her effort netted the Foundation \$1,000 — one of the

## In Memorium Frances Bernstiel 1930-1994

most successful grassroots fund raising efforts by a F.I.R.S.T. member.

We were stunned to learn of Mrs. Bernstiel's sudden passing. A bright, vivacious woman, and always thoughtful and generous toward F.I.R.S.T., Mrs. Bernstiel's unexpected death caught us all off guard—as death so often does.

We offer our most sincere condolences to Frances's husband of 39

years, Elwood Bernstiel, and her daughter, our good friend Lynne Alba. She is also survived by two sons, James and Mark, and by two precious grandchildren—Lynne's son and daughter—Matthew and Brittany Alba.

The Bernstiel family, in a remarkable gesture to honor Mrs. Bernstiel's commitment to F.I.R.S.T., asked that memorial contributions in her memory be made to F.I.R.S.T. To date, friends and family have contributed \$2,500 to F.I.R.S.T. in the name of Frances Bernstiel. I know that all of you join me in offering condolences to family and friends.

## Regional Support Network

### REGION ONE

NY, PA, VT, NH, NJ, CT, RI, MA, ME  
open

### REGION TWO

DE, MD, WV, VA, NC, SC  
Robin Joyce  
Route 1, Box 189-F  
Sandy Ridge, NC 27046  
(910) 871-3277

### REGION THREE

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### REGION FOUR

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133 Elam Park  
Lexington, KY 40503  
(606) 276-0142

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Katy, TX 77493  
(713) 391-4407

### REGION SEVEN

WA, OR, ID, MT, WY, AK  
Sheri Gelivich  
1344 S.E. Rex  
Portland, OR 97202  
(503) 236-3203

### REGION EIGHT

CA, NV, UT, AZ, HI  
Chris Beeler  
1002 E. Gifford Drive  
Tucson, AZ 85719  
(602) 887-7188

### Canadian Contact

Barbara Rockwell  
3080 Ballydown Crescent  
Mississauga, Ontario L5C 2C8  
Canada  
(416) 896-9620

### Netherton's Syndrome Contact

Cathy Sipper  
Rt. 1, Box 300  
Laverne, AL 36049  
(205) 335-6827

### National Coordinator

Heather Gattuccio  
3726 Bellevue Road  
Raleigh, NC 27609  
(919) 781-6671

### STATE CONTACTS

#### Kansas

Tiffany Karst  
913-268-3692

#### Mississippi

J. Charlene Wiggins  
601-769-2454

#### Indiana

Mark & Jill Wood  
317-841-9923

F.I.R.S.T. is developing a system of state contact persons. This is in addition to our network of Regional Coordinators. Most likely, future Regional Coordinators will be drawn from the pool of acting state contact persons.

Anyone wishing to become a state contact person should call the RSN National Coordinator, Heather Gattuccio, to obtain an application. Openings are also presently available in some Regional Coordinator slots. Please speak to Heather about these as well if you are interested in filling the position.

F.I.R.S.T.  
P.O. BOX 20921  
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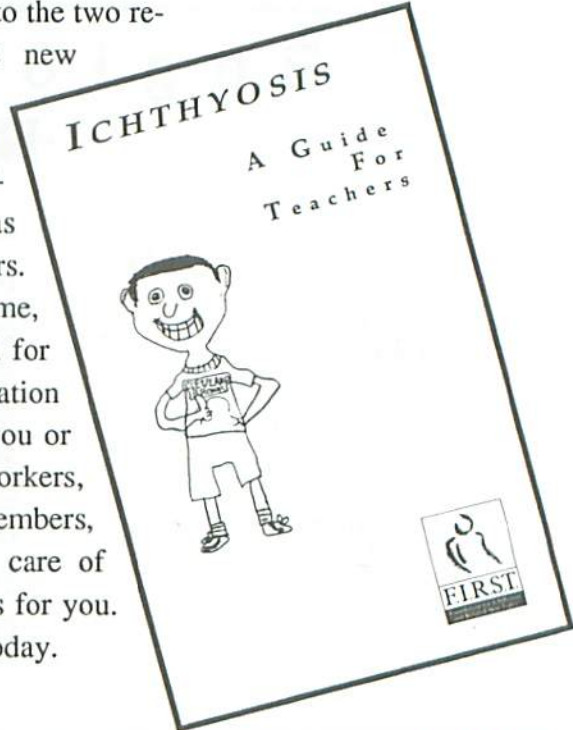
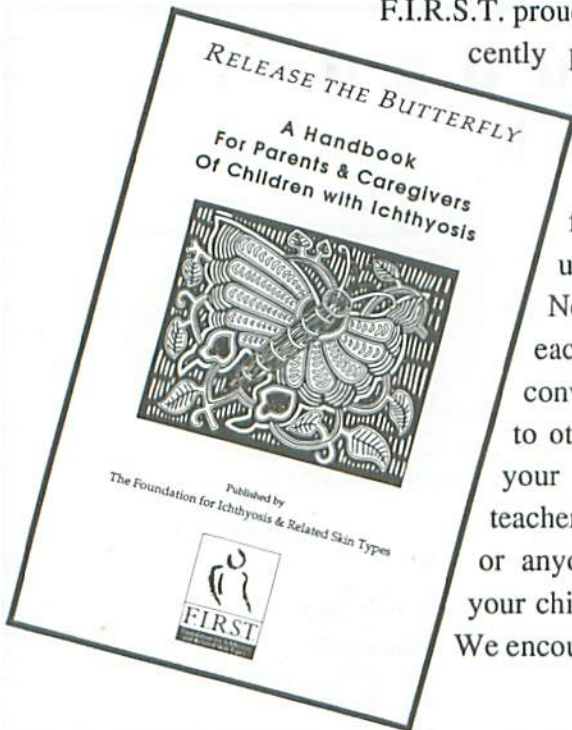
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F.I.R.S.T. proudly introduces members to the two recently published books in its new

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Not only useful in the home, each can be a valuable tool for conveying important information to others who interact with you or your child—child care workers, teachers, employers, family members, or anyone else for whom the care of your child is as important as it is for you. We encourage you to order both today.



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P.O. Box 20921  
Raleigh, NC 27619

TOTAL: \_\_\_\_\_