



ICHTHYOSIS FOCUS

Vol. 20, No. 3

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

Fall 2001

National Ichthyosis Awareness Week

September 30 - October 7, 2001

This year Ichthyosis Awareness Week will be celebrated during the week of September 30 - October 7. All members of F.I.R.S.T. are encouraged to promote ichthyosis awareness in your community by contacting your local media and/or hosting a fund raiser. See page 4 for ideas on how you can help educate your community about this disease or raise money for the foundation.

continued on page 4

Why Should I Enroll in the Ichthyosis Registry?

The National Registry for Ichthyosis and Related Disorders identifies and enrolls individuals affected with disorders of keratinization. They also have received government funding to perform molecular diagnosis to identify some of the specific genetic defects for enrolled individuals. Here's your opportunity to discover which specific genetic mutation is the cause of your particular disorder. Unfortunately, there are limits as to who can be tested and what tests can be offered. However, this testing is a huge improvement in the knowledge that the Registry can obtain and share with its enrollees.

Over 700 individuals have already enrolled or are in the process. Besides the obvious benefit of molecular diagnosis, there are many other reasons to join the Registry. The Registry:

- encourages investigators to study the ichthyoses;
- finds better ways of diagnosing disorders and better treatments;
- collects information for their database as to how these disorders affect individuals, other health problems that may be associated to the ichthyoses, how other family members are affected, and different ways individuals treat their skin;
- seeks your interest in participating in research studies;
- takes posters and demonstrates at scientific meetings to educate dermatologists and scientists about using the Registry as a resource;
- publishes notices in medical journals;
- provides a doctor's forum on their website for doctors to ask questions about ichthyosis.

To begin the enrollment process, contact Geoff Hamill, Registry Coordinator, at 800.595.1265, e-mail at info@skinregistry.org or visit their website at www.skinregistry.org.

Please Give to the United Way

It's simple...

just write in Foundation for Ichthyosis & Related Skin Types (F.I.R.S.T.) on the Donor Choice Option Form and your funds will be designated to F.I.R.S.T.



A United Way
System Partner

Thanks to all who contribute through the United Way....
your help goes a long way!

Regional Meeting

October 13th, 2001
9 a.m. - 4 p.m.

Bloomington, GA

See page 10 for meeting
information

2002 Family Conference

Seattle, Washington

July 5 - 7, 2002

See page 8 for conference
information

Ichthyosis Focus

Vol. 20, No. 3
Fall 2001

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Ichthyosis Focus is published quarterly by the Foundation for Ichthyosis & Related Skin Types (F.I.R.S.T.)

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

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F.I.R.S.T. is a 501 (c) 3 charitable organization supported by public and private donations. All contributions to F.I.R.S.T. are tax deductible to the full extent of the law.

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

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Correspondence Corner

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

In response to your letter about annual membership, let me please say that I am more than happy to be a member of this foundation, so please continue to send me all of the information. Please take a minute to read my story, so that you may understand why you have not heard from me before.

My pregnancy was normal, but when I gave birth to my son I saw the horror on the face of the nurse as I heard her say, "What is that doctor?" It was then I knew my son was different. He was sent to Children's Hospital in San Diego, and the doctors told me that my son had a very uncommon disease. The dermatologist at Children's Hospital told me my son had ichthyosis and that so few cases of it were known that we would just have to live with it. At one of our doctor's appointments, another dermatologist saw my son and after reading his file asked why my son had not been sent for a skin biopsy, and why we had not been referred to the foundation. This was when I learned that F.I.R.S.T. existed.

I feel much better knowing that I am not alone, and that my baby is not the only one affected.

Virginia Cortez
Calexico, CA

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

Since I went through the skin biopsies and testing with the dermatologists from Washington, D.C., I have learned a lot, tried different lotions, experimented with different scrubs to exfoliate the skin and I feel the best I have about my skin in all my 66 years. I am very glad I went through the F.I.R.S.T. Foundation. I also enjoy the Focus very much, thank you!

Rosella Olsen
Alexandria, MN

MATCHING GIFTS :

Does your company have a matching gift program? If you donate to F.I.R.S.T., your company can match your gift just by completing and submitting a matching gift form. Check with your company to see if they have a matching gift program and see how you can double your support to the foundation.

Deadline for Jane Bukaty Membership Assistance Fund: December 31, 2001

The Jane Bukaty Membership Assistance Fund has been established to help members of F.I.R.S.T. with financial support for expenses involved in the care of ichthyosis. The Support Network & Membership Assistance Committee awarded three applicants with funds from this program during the last round of applications. Applications are now being accepted for the third cycle of applicants. The application process is simple and easy to do. Simply complete an application form and send it in! The form is available in the "members only" section at our website, www.scalyskin.org, or one can be mailed to you at your request. Just call the national office at 800.545.3286. The deadline for applications is December 31, 2001.

Mother and Daughter Team are Honored with Gracie Allen Award

Congratulations to Heather and Caitie Gattuccio on winning the prestigious Gracie Allen Award. Heather is a former RSN Coordinator for F.I.R.S.T. and her husband Nick, is a past executive director. This award is given each year by The American Women in Radio & Television. For over 25 years the AWRT has dedicated itself to programming by, for, and about women. They strive to encourage positive and realistic portrayals of women in today's society. This past May, the awards ceremony was held in New York City, where the winners gathered to celebrate their achievements. Some of this year's other winners include: Rosie O'Donnell, Debbie Allen, ABC News - 20/20, and Dateline.

The Gattuccio's radio production, entitled "Caitie's Story," was written and produced by Heather Gattuccio. Speaking about her daughter Caitie, this documentary essay depicts the physical, emotional and mental impact lamellar ichthyosis has had on their family. Heather describes what it was like when Caitie was born and how they are coping with the difficult situations facing their family as Caitie grows up. Nine-year-old Caitie shares her feelings about what it is like to have ichthyosis and how she envisions her future.

Cassettes of "Caitie's Story" are available through SoundPrint Media for \$12.50 each. You can listen to Caitie's story at http://www.soundprint.org/radio/display_show/ID/29/name/Caitie%27s+Story or order your own copy at www.soundprint.org, by calling 1.888.38.TAPES, or writing to Soundprint Media Center, Inc., 525 Main Street, Suite 105, Laurel, MD 20707.

In Memory of Joseph William Robertson Jr.

F.I.R.S.T. extends sympathies to the family of Joseph William Robertson Jr., otherwise known as "Bill," who died at the age of 53 from a massive heart attack. Bill was the father of three children, Jenny, Bonnie, and Joseph. Bonnie has Darier's disease. He is survived by his ex-wife, Dianna Berg, a sister, Shellie Robertson, brother, Jim Robertson, daughter-in-law, Sara, and grandson, Joseph William Robertson IV. Bill was extremely dedicated to his family and had a great sense of humor and love for others. He was Chief Financial Officer for Weingarten Realty, a consultant for Live Oak Capital, and a full-time golfer. F.I.R.S.T. is grateful to receive donations in Bill's memory from family and friends, totaling over \$3100.00.

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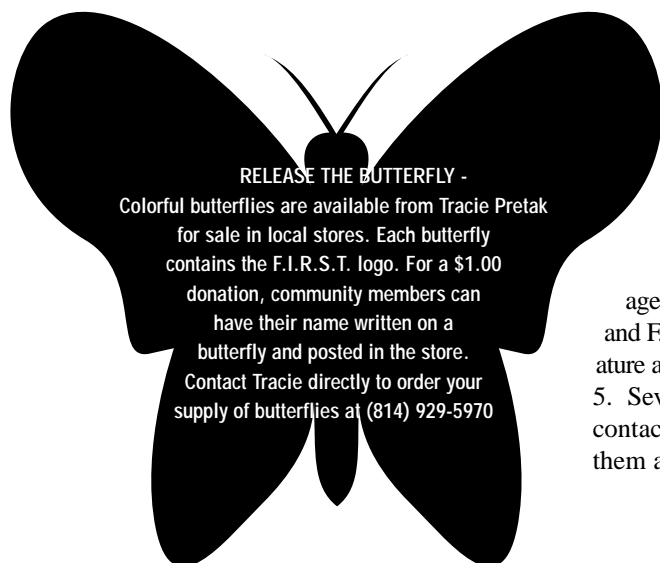
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Ichthyosis Awareness Week — Get Involved September 30 - October 7

continued from page 1

Thanks to our members, last year's Ichthyosis Awareness Week (IAW) was a huge success. This year our goal is to surpass last year's success and create even more exposure about ichthyosis. Even if you don't have a lot of time, there are plenty of ways you can help. All ideas are good ideas. Be creative, have fun, and get your family and friends involved.



Here are some helpful hints to organize your contact with the local media. Most communities welcome human interest stories from their residents, so don't be shy. The more media exposure about ichthyosis and F.I.R.S.T., the more people will become educated about the disease.

1. Make a list of local newspapers, radio and TV stations.
2. Contact the places on your list and ask for the name and address of where to send your human interest story.
3. Write your own personal story about ichthyosis and how it has affected your life.
4. Send your story (and photo, if possible) together with a package of information about ichthyosis and F.I.R.S.T. (brochures and other literature are available at the national office)
5. Seven to 10 days later, make voice contact with your media contacts. Tell them about Ichthyosis Awareness

- Week and F.I.R.S.T. Ask them when they will be printing or airing your story. Make a personal, sincere request.
6. If you don't receive a favorable response, follow up again. Depending on the news of the day, you may need to make repeated requests to bring attention to your story.

Here are some other ideas for fund raisers: Auction, Bake Sales, Bingo, Bowl-a-thon, Car Wash, Coin Harvest, Community Dog Wash, Garage Sales, Raffle, Sports Events, Walk/Run-a-thon.

Remember to contact the office to let us know what you are doing. We would like to recognize and share your efforts with other members of F.I.R.S.T. We are always here to help you with your project, so don't hesitate to call us.

Note: If your schedule is hectic during this week, don't worry. You can plan your event for another time during the year when your schedule is a little more convenient. We encourage anyone to get involved at anytime.

Italian Ichthyosis Conference

On June 24th, the Italian Ichthyosis Group held its first conference in Rome, Italy. The day-long conference was attended by almost 100 participants, in addition to the many doctors and researchers who spoke. The conference was a great success, as it was the first time many of the participants could meet and speak with one another.

Professor Rino Cavalieri, head of the Istituto Dermopatico dell' Immacolata (I.D.I.), opened the conference by praising sci-

entists and therapists for their diligent efforts. Dr. Mauro Paradisi explained the pathology, while Dr. Eleonora Candi discussed the aspects of the molecular biology and the methods through which samples are analyzed to obtain a precise diagnosis. Dr. Biagio Didona talked about the therapeutic approaches for treating various types of ichthyosis. Research and future gene therapy were discussed by Dr. Gerry Melino, a Molecular Biology Researcher.



Executive Director's Report

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



Thank you to everyone who donated through our annual membership campaign this past spring. Donations exceeded our expectations. As usual, our membership continues to show their dedication and generosity in support of the services and programs F.I.R.S.T. has to offer.

I am excited to report that the foundation is currently engaged in the development of a 3-5 year Strategic Plan. This plan will help to shape the future of the foundation. Last month, Strategic Planning Committee Members and I attended a one-day face-to-face retreat in Philadelphia to define the future goals of F.I.R.S.T. These dedicated committee members will return to Philly this month to determine the objectives to achieve these goals. Input from telephone and newsletter surveys from our members were an integral part of helping to outline these goals and, ultimately, the future direction of F.I.R.S.T. The final written strategic plan will be presented to the Board of Directors at our annual board retreat in November. More details about this plan will be forthcoming.

Last month, I attended a meeting at the University of Pennsylvania's Center for Bioethics on intellectual property rights. A few lay advocacy groups, researchers, and ethicists attended the meeting. The discussion focused on gene patenting and issues surrounding this subject. In summary, it is the hope of this group that even if a company, researcher, or university patents a gene, it is still available, accessible, and affordable to the general public.

I also had the opportunity to volunteer for a day at Camp Discovery in Millville, PA last month. Camp Discovery is one of three camps in the country that is sponsored by the American Academy of Dermatology for kids with skin diseases. What an awesome experience! Please turn to page 14 to read about the camp and see photos.

Since the last printing of the newsletter, there has been a change of officers on the Board of Directors. Laura Phillips resigned as CFO due to a conflict in her professional life and was replaced by Beth Gray. Laura has taken over Beth's position and now serves as Secretary of F.I.R.S.T.

Please feel free to contact me within the next few weeks about any ideas or plans you may have for Ichthyosis Awareness Week, September 30 - October 7, 2001. I would be very happy to help.

Sincerely yours,

Jean R. Pickford



Strategic Planning Committee Members (clockwise from top left): Charles Eichhorn, Beth Gray, Laura Phillips, Leonard Milstone, MD, Elena Levitan, Donna Rice and Lynne Alba. (not pictured: Mike Dunleavy and Deb Vilas)

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Good Sense Helps Defuse Suspicions of Child Abuse

Reprinted from the Baltimore Sun, April 6, 2001, by Dan Rodricks

When I asked that Rhayne Evans spell her name — as you can see, a necessary and reasonable request — she flinched a little, laughed nervously and said, “I’m not going to end up on that list, am I?” She was referring to the state’s Child Abuse Registry, which includes the names of about 100,000 people never criminally charged with abuse but the subject of complaints filed by suspicious neighbors, teachers, doctors and others.

Because this list is secret — accessible only to state and county social workers — I had no way of knowing the answer to that question. But given what is known — that some names stay on the registry for up to 25 years, that mere suspicion of child abuse can get you listed — Evans hesitated about attaching her name to today’s column. It was understandable. Her concern — that she’d be associated with child abuse for years merely because someone in a Burger King had had a suspicion — floats in that crevice (or chasm, depending on your view) between the public’s desire to protect children and its desire for personal privacy.

While I couldn’t guarantee anything, I told Evans that I believed no reasonable person, upon reading this column, would tap her name into a computer at the state Department of Human Resources.

Her cute little girl, Abby, is not the victim of abuse. She was born nearly three years ago with a rare genetic skin disorder called lamellar ichthyosis. It’s evident by dry skin and patches of dark, scaly areas on her body. It occurs in one of every 300,000 children. It has no cures, only treatments.

One day last month, Evans and a friend took their children to a Burger King in Cockeysville. They had lunch and let the children loose in the adjoining play area. They were there for about an hour.

Long into that hour, a young Baltimore County police officer stepped into the restaurant and, after making some observations, approached Evans. He qui-

etly told her that another BK patron had reported seeing a little girl who was severely bruised.

“Fortunately,” Evans says, “the officer was very nice and intelligent enough to realize that Abby was not bruised. He never approached my daughter, thereby saving her some trauma. He only watched her from a distance. He said, ‘I hope you understand, we have to investigate.’”

“This wasn’t the first time people said thoughtless, stupid things or gawked at Abby, only the first time they have gone so far as to contact the police.”

While Evans was grateful for the police officer’s sound judgment and professional conduct, she was miffed that someone in the Burger King — possibly a woman with whom she’d had brief and pleasant conversation — would call the police.

“I taught elementary school in Baltimore City for seven years and understand the importance of being aware of possible child abuse,” Evans wrote me in an e-mail. “But I would never have presumed to call the police about someone I happened to see in a restaurant unless I was witness to actual abuse.”

That’s where our conversations got even more interesting.

I told Evans I would not have to witness assault to contact police. Maybe everyone has a different definition of child abuse. I think most humans instinctually know it when they see it.

If I saw it, I’d have to do something — get a license tag number maybe, or a description of the child and the suspected abuser. I felt compelled to do just that a few years ago in Baltimore County.

Outside a supermarket, I saw a stressed-out guy grab a small boy by the hood of his winter jacket, hoist him 2 feet off the ground and carry him across the parking lot. The little boy appeared to be trussed inside his own coat; he looked as if he was choking. I yelled at the guy, and he yelled something back. I got his tag number and reported the incident to police. I don’t know whether anything was done. Maybe that stressed-out guy’s name is on the Child Abuse Registry now. And maybe he thinks that’s a terribly unfair

thing. But you know what? It doesn’t bother me one bit.

Rhayne Evans’ case is way different.

Upon my visit to the Evans home in Carroll County, it was instantly obvious that there’s something psoriasis-like about the skin on Abby’s hands and forehead, nothing more. The creased, dry skin reminded me of that of a boyhood friend who must have had one of the 20 varieties of ichthyosis that dermatologists have identified over the years.

“This wasn’t the first time people said thoughtless, stupid things or gawked at Abby, only the first time they have gone so far as to contact the police.”

Still, someone in the Burger King must have seen it differently. But I don’t fault someone for making a report to police.

What’s important is that the police officer did his job in a gentle and discreet manner and that he made a sound judgment. It’s what we count on cops, social workers and judges to do every day as they make decisions about the welfare of children, and if we’re lucky, they get it right most of the time.

“I wish I had thought to ask the officer’s name, as he was very kind and professional,” Evans says. “After I had collected myself and gotten over the shock that something like this could happen, I would have liked to have been able to thank him.”

That’s all Evans wanted from this column — to thank the police officer. I talked her into going a little further with it — to make more people aware of ichthyosis, for one thing, and to remind everyone about the impact of their 911 calls about suspected child abuse. Make sure you’ve made the best judgment you can make, then make the call. And even if you’re not sure, make the call. If everything works the way it’s supposed to — and I think it did at the Burger King in Cockeysville — innocents have nothing to fear.

What's New, What's Hot & What Works...

I would like to let the readers know that out of all the creams and lotions I have used, Nivea Cream has worked the best. I would like to hear from some people who have had good results from other creams and lotions. I know the weather is getting cold. I don't get along with hot or cold weather, so I would like to know what some people do to get comfort from the heat. I do not have air conditioning as I am on a fixed income and can't afford it.

Raymond Reed Jr.
Delanco, NJ

I use Avon Moisture Therapy for Hands all over my body and have had incredible results. My ichthyosis has dramatically improved and the scales on my legs are barely noticeable now. It has worked very well for me and I wanted to share this tip with your readers.

Phyllis Parsons
Brook Park, OH

My wife wrote this at the suggestion of a friend. We carry this card to hand out to someone who comments about our son's skin. Hopefully it will help other parents when they have to deal with the community like it has helped us.

While we appreciate the obvious concern you have regarding our child's skin, he is of the age where he understands what is being said, and we will not discuss this condition continually in front of him. Though you may feel you are the only one to ask, we address this question daily. Thus, here is some information that you may deem helpful in understanding his condition. Our son has a rare skin disease called Pityriasis Rubra Pilaris. This disease causes our son's skin to appear chapped and/or sunburned. It is a genetic disorder that has been passed on to him, so it is not contagious. Since we noticed signs of the disease when he was four (4) months old, he has been under the care and supervision of leading pediatric dermatologists. We are doing everything in our power to keep him comfortable and assist him in leading as normal a childhood as possible. His treatment consists of a variety of topical creams. UltraViolet lamp treatments are suggested for when he is older, but for now exposure to the

sun is recommended. If you have children, please discuss this with them and assure them that it is fine to play with him. If you are a parent, you can imagine how it feels to have your child teased or made to feel bad every day if something is different about them. Your understanding is appreciated. Thank you.

Tim & Tamer Holzer
Cumming, GA

Taking Care of Dry Nails — Reprinted from SKINFACTS, from the American Skin Association

Many of us are troubled by nails that split or tear easily. What causes this problem? How can we prevent or treat it? Dermatologists have found that nails split because they're dry due to a lack of water in the nails. Extended stays in unusually dry atmospheric conditions and severe droughts (as experienced in many parts of the U.S.) contribute to the problem. The best treatment for this is to soak the nails in warm water — either by submerging them or wrapping them in wet cloths — for 10 minutes, followed by the application of oils. Wearing cotton dermal gloves nightly helps to keep the nails from losing moisture. Do not use topical "nail strengtheners" or hardeners (especially those with formaldehyde) as they coat the nail and prevent water from penetrating. They also cause the nails to be abnormal. In addition to the therapy, nutritional options may also help some individuals. Consult your physician. Whatever approach you take, don't expect overnight results. Adherence to a regular program will bring results gradually.

Bathing Glove

Last year, I discovered a fantastic bathing glove product, which has been extremely helpful to me with my lamellar ichthyosis. I was so pleased with this product that I shared it with my sister, Jean, who lives in Florida and also has lamellar ichthyosis. She loved the gloves so much, she told me she actually looks forward to bathing! This is important to note, since the degree of severity of our condition is definitely not the same. This product works equally great for both of us.

This product is a pair of loosely knit gloves made from an abrasive type of mate-

rial, which appears to be nylon or some type of plastic. You can use these gloves on just one hand or both hands, but they will greatly speed up your bathing time and save your fingernails. They will clean your skin really, really well. And the astounding thing about them is that they will last a long time. Also, you can even increase their longevity by turning them inside out and wearing on the opposite hands.

Just put the gloves on and soap them with liquid or bar soap, as you would with a washcloth. With a glove on each hand, you will be amazed at how well they clean — and in less time. They rinse out quickly and clean and you can hang them in your shower or bath. Before your next bath time, they will be completely dry and ready to go again. Of course, periodically, you should wash them out very well and dry.

These gloves can make a great holiday stocking stuffer or any other type of gift. They are great for anyone, whether you have ichthyosis or not. I would have loved it if someone had presented me with a pair — perhaps along with a bar of great soap or a bottle of bath gel.

I have managed to find a supply source for these gloves and want to share them with anyone who would like to try them. These gloves come in about (3) color categories: various pinks, teal and white. I will do my best to come as close as possible to the color you want, but I cannot guarantee the color, as I will have to accept a supply of various colors due to the limited number being ordered. These gloves hug your hand, but they will stretch and only come in one size.

I will be running all orders I receive through my small business, which is unrelated to this item. If you would like to try a pair of these bathing gloves, please forward a check or money order for \$11.00 for each pair you desire (shipping, handling and taxes included). For USA only. For any foreign address orders, you need to call me first.

Please print your name, address and phone number and send with your check payable to: Elaine Enterprises, Ltd., P.O. Box 7755, Greenwich, CT, 06836.

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203.661.7026 (phone & fax)

2002 Family Conference

Seattle, Washington • July 5-7, 2002

CONFERENCE CHATTER

Program

Below is a tentative list of what has been planned for the conference program.

ADULTS: Ask the Doctors — Pediatric Concerns; Ask the Doctors — Adults Concerns; Importance of Family; Mom's Discussion; Dad's Discussion; Women's Discussion; Men's Discussion; ISN Sign-up and Training; Treatment Options (AHA's, retinoids, etc); How to support your kids through a medical procedure; Information sharing & helpful hints; Make-up demonstration.

TEENS: Healthy choices in a fast food world, diet supplements, nutri-

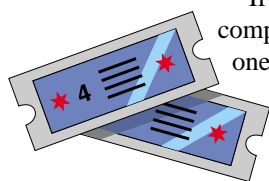
tion issues; Career Panel Discussion; Opportunity to talk with doctors who specialize in ichthyosis; Strategies for coping with social challenges; Simple strategies for personal safety; Make-up demonstration.

TWEENS: Creative outlets for personal expression; Simple strategies for personal safety; Healthy choices in a fast food world, diet supplements, nutrition issues; Strategies for coping with social challenges; Crafts, art, music.

CHILDREN: KiddieCorp. — Professional child care for children ages 9 and under.

A CLINICAL SCREENING by physicians who specialize in ichthyosis will be available for all attendees. These screenings will be scheduled by appointment only. Appointments can be made at the registration table on Friday, July 5.

Raffle Items Needed



If you work for a company or know someone in business who would be willing to donate an item in support of our raffle, please let

Jean know at the office. Items such as a DVD player, trips, electronics, jewelry, housewares, gift certificates, etc., would be greatly appreciated. All donations are fully tax-deductible to the extent allowed by law. Call the office at 800.545.3286 or email at info@scalyskin.org.

Registration

Registration will not begin until January, 2002. A registration form will be included in the next issue of the Focus and will be available online at www.scalyskin.org.

Room Reservations

Call the Seattle Marriott Sea-Tac Reservation Office at 800.643.5479 to make your room reservations. F.I.R.S.T.'s discounted price is \$89.00 + applicable taxes per room, flat occupancy. Be sure to mention this group rate is reserved under Foundation for Ichthyosis. The deadline for room reservations is June 13, 2002.

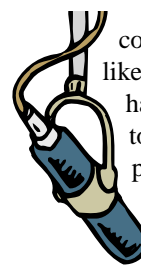
Seattle Tour



F.I.R.S.T. is organizing an optional social tour around Seattle on Sunday, July 7, after the conference ends.

Coaches will take our group for a 3-hour tour of Seattle and its famous sights. There will be a small fee for this tour. Information about this tour will be available in the next issue.

Talent Show



On Saturday evening, the conference committee would like to host a Talent Show. If you have something you would like to perform for the audience, please contact Jean at the office. Here's your chance to show off your talent.

Whether you can dance, sing, play a musical instrument, tell a good joke, juggle or whatever, we want to hear from you. Don't be shy!

Transportation

For air transportation assistance and/or information, contact the National Patient Travel Center at 888.675.1405. This organization makes use of Angel Flight America programs and commercial airline programs. F.I.R.S.T. has established a unique plan for our members called *2002 F.I.R.S.T. Family Conference Special Lift Program*.

Advice

Haley, my 9-year-old daughter with ichthyosis, and I were recently approached by a middle aged woman with a homemade cure. Because our native languages were not compatible, we're uncertain of the ingredients beyond vinegar. Haley found the encounter amusing and we began to list all the unsolicited and unwelcome advice we've received. Some of our favorites are:

"You need to get that sunburned child in the shade" (and YOU need to get some manners)

"You need to find some time for yourself" (does sleep count?)

"All she really needs is love"(yea and a pound of Aquaphor®)

"You ought to try to get rid of that scaly stuff"(duh!)

"It'll go away"(glad we didn't hold our breath)

We've also gotten some great advice over the years from books, friends, family and professionals. Some of you may recognize your words, if so, thank you. Some of you may recognize the words of your favorite authors, if so, don't tell their publishers.

You may want to change the world, but you don't have to do it every time you go to Wal-Mart.

You have room to make mistakes and it's a good thing cause you're gonna make lots of 'em.

Humor is an emotional Swiss Army Knife: tool, weapon, vehicle, cushion, and medicine, all in one.

Don't try so hard, there's a place between pity and heroic martyrdom.

Stop obsessing, have a little party for the stuff that went away.

Life is what happens while you're busy making other plans.

Not every bad thing that happens is because of ichthyosis.

If you're not going to try this new cream tell me now.

It doesn't have to be the worst to be enough.

It's just a different way to define beauty.

Sorrow is like wind, it comes in gusts.

Make friends with your child's doctor.

Severity is in the eye of the beholder.

Ichthyosis is a disease, not a career.

Moisturize, moisturize, moisturize.

One bath is good, two's better.

Not everything has a solution.

Blame is a waste of time.

Never refuse help.

Joy is a choice.

Skip the guilt.

Lighten up.

Play nice.

Never lie to your mother. (I just threw in that one for my kids.)

If you've received some really great advice, bits of wisdom or words to live by, send them to us; we'd love to add them to our collection.

*Donna and Haley Rice
drice@pdq.net*

Heritable Skin Disorders

Reprinted from Progress & Promise, a publication produced by the National Institute of Arthritis and Musculoskeletal and Skin Diseases

Our largest and most highly visible organ, the skin, literally holds us together. And, like other organs, it is susceptible to disease, which can be disfiguring, debilitating, or even deadly. Among the major research focuses of the NIAMS are how skin functions, grows, and repairs itself and how we can treat the diseases that affect it.

Researchers have found specific genetic defects that cause inherited disorders affecting the structural integrity of the skin. These disorders include the ichthyoses, a group of scaling skin disorders. The discoveries are resulting in more specific diagnostic tests, making possible prenatal diagnosis and genetic counseling, as well as opening the door to treatments, including gene therapy, to correct these defects.

Scientists have:

- Identified a gene mutation linked to the region of chromosome 1 that is likely to impair normal epidermal differentiation. This study demonstrated the power of genetic analyses in evaluating rare diseases. As researchers develop a better understanding of epidermal differentiation, insights into more specific diagnoses and treatments of disease are expected to follow.
- Shown that a protein involved in developing the outer layer of skin is reduced in people with ichthyosis vulgaris, the most common form of the disease.
- Discovered that lamellar ichthyosis, characterized by severe scaling that greatly affects a person's ability to sweat, is caused by mutations in the gene that regulates the protein transglutaminase 1.

Readers should recognize that there is a cluster of genes that encode proteins that are involved in the maturation or "differentiation" of epidermis (the outer layer of skin) on the first chromosome (1q21-25). Chromosome abnormalities at this site were first described years ago in patients with hyperproliferative skin disorders, and it is likely that mutations in several of these genes may someday be linked to specific disorders.

Transglutaminase I is the first gene to be associated with lamellar ichthyosis, and is probably the most common gene. However, readers should recognize that there are at least two other genes that have been linked with lamellar ichthyosis, although at present the identity of these genes is unknown.

ASK THE READERS: What Do You Do?

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 our Summer 2001 question... During hot summer months, how do you keep your body cool?

During the summer, I keep cool by wearing a hat when I garden or mow the yard. I do not garden during the hot summer afternoons, I leave it for the cooler evenings when there is shade. When I go for my walks, I go in the evening when it has cooled down or walk in the mall, and I always carry a jug of water with me. If the temperature is going to be high I do not go to functions where I could get overheated, and I am okay with that. When I do get too warm, I splash on some cold water and stay inside. We are fortunate that we have central air. I also only go golfing when it is cool. All these adjustments have worked out well for me and I am so grateful to F.I.R.S.T. for all the knowledge I have obtained in regard to handling my ichthyosis vulgaris. Thank you. **Rosella O., Alexandria, MN**

Sometimes in the summer, I let my child play in wet T-shirts, and I use wet bandanas to wrap around her head and neck. That usually keeps her cool for a little while. I also keep a wading pool in the backyard for frequent dips and the sprinkler on. **Jennifer S., Camden, DE**

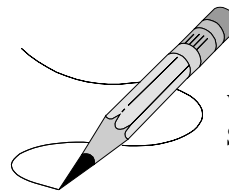
For my nine-year-old, I carry a small ice chest in the car with ice packs, bottled water, and wet cloths. I also have a cool pad for the car along with a cool vest. **Donna R., Katy, TX**

When we go places that are hot, I always take my "MistyMate" or spray bottle with a fan. This helps keep me cool when I start to get overheated. And I never leave the house without a spray bottle of ice water. **Karen S., Hellertown, PA**

Here's our next question:

When your child gets a scrape or cut, what do you do about a band-aid that just won't stick?

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

Regional Meeting

October 13th 2001

9am-4pm

Bloomington Alliance Church

501 W. Hwy 80, Bloomington, GA 31302

Contact Patrice Russell at (912) 748-8396

The program will include a morning reception with coffee, juice, bagels, doughnuts, and registration for door prizes. After the reception, a dermatologist will join us to speak about ichthyosis and any new treatments available. We'll break for lunch to enjoy some barbecue with all the fix'ins. (Please specify if special arrangements are needed for any health conditions, such as diabetes.) Lunch will be followed by a question and

answer session with the dermatologist and open discussion within our group. Dinner reservations will be made upon request.

There will be children's activities, which include pony rides, crafts, T-shirt painting, puppet show, and a nursery for infants. We have a group of a dozen teens and adults who will be donating their time and talents to make your children enjoy their time at the meeting. There will also

be hot dogs, chips, and lots of snacks for your little ones.

The cost is \$12.00 for adults and \$8.00 for children. Pre-registration is required. A block of rooms has been reserved at a local national hotel chain, so please let me know if you would like me to hold one of these rooms for you. Contact Patrice Russell at (912)748-8396 or e-mail at Tresey1@Juno.com.

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.



ISN Education & Training

Coping and Crisis Information

Part 2 — *Helping an Hysterical Caller*

There may be times when support network coordinators and volunteer peer counselors in the support network find their caller becoming hysterical or overwrought.

If the caller is crying hysterically, and you've tried to console him without success, you may need to take a short break. Explain to the person that you really want to talk to him, but you want to give him a break. Tell the caller you will call him back in ten minutes. Suggest he take a few minutes to go splash his face, take some deep breaths, get a drink of water. Then you take a break, too. Call another member for some support for yourself.

CALL BACK WHEN YOU SAID YOU WOULD! Time may allow the caller to collect himself and his thoughts, and to be able to speak more clearly. If the per-

son is still crying on the second call, try to ask specific open-ended questions like:

“What specifically is upsetting you?”

“What happened today that upset you?”

“Who has been the best support person for you in this, and why?”

Find out if there is another family member or friend nearby on which the caller can rely if he needs additional support. If you feel it is appropriate, give the caller a way to contact you. Don't give information that you are uncomfortable giving.

Keep in close contact (daily if possible) until the caller seems more in control. If you feel the person needs more support from other network members or the national office, make the appropriate calls. If the caller experiences frequent

crying spells over a period of time, it might be useful for the person to talk with a professional counselor. Appropriate therapists can be found through family doctors, employee assistance programs, church or synagogue, or the county mental health office usually listed in the blue pages of the phone book.

Recognize your limitations. If you feel uncomfortable handling the caller's situation, seek help from the national office or other support network members. Do not attempt to counsel or advise someone who needs more than you can give. You might do more harm than good. Remember, you are a volunteer counselor, not a professional.

Let's See What Our Members Have Been Up To...

John Little, of Webster, NH, sponsored a demolition derby raising \$400.00 for the foundation. The winning drivers, Lee Day, Chris Rose, and Chris Wesoja, also donated their winnings to the foundation. The money was donated on behalf of Nichi Hoar, an eight year old with lamellar ichthyosis.

Liza Santamina participated in the 2001 Human Race, raising \$380.00 for F.I.R.S.T. The Human Race is an annual event in which walkers and runners raise money for non-profit agencies.

Tiffany Karst hosted her annual garage sale which raised over \$1500.00. Tiffany and her friends join together each year to raise money for F.I.R.S.T. by selling garage sale items to the community.

On July 25th, **Tracie Pretak and Bailey Jones** held a “Christmas in July” concert. Over 170 people attended the summer concert in which twenty talented people, including Kelly Walters and Bailey Jones, told the story of Christmas through song and dance. A free-will offering for F.I.R.S.T. was collected, and

videos were sold with a percentage of the proceeds going to the foundation. The Johnsonburg Rotary Club and Aid Association for Lutherans generously assisted with financial donations for the production. So far the concert has raised \$2,160.00 for F.I.R.S.T.

Upcoming Fund Raisers

Entertainment Books are for sale once again this year. These books can be purchased for \$25.00 and contain coupons to local businesses in your area. Books are available for all states in the country. The books became available as of August 24th. For each book purchased, F.I.R.S.T. will receive \$5.00. Contact Kelly Strother at the national office at info@scalyskin.org or call 800.545.3286.

Dawn Johnson and Stephanie Concialdi, two mothers of affected daughters, will be hosting their 2nd annual Monster Dash. Proceeds from this 5K race are donated to F.I.R.S.T. This event will take place on Saturday, October 27th in

Friendswood, TX. Please contact the office if you are interested in supporting this event.

Rod Hoover is hosting “Tour of the Flinthills,” a 60-mile bicycle ride touring the scenic beauty of the rolling Flinthills in Kansas. The event will take place on September 29th. All proceeds raised from this event will be donated to F.I.R.S.T.

Tracie Pretak and Bailey Jones are planning their 5th annual “Release the Butterfly” concert for October 7. They will continue with their usual format of sacred music with God's message of “Love One Another.” Their talented team of family and friends are excited to perform again for their generous community.

Rick and Kim Maurer of Morgan, MN, are planning a Bowl-a-thon on September 22 to raise funds for F.I.R.S.T. Their four-year-old son, Preston, has CIE. The Maurers' story about Preston and ichthyosis was also covered in a local newspaper, The Redwood Gazette. Rick and Kim hope to receive more coverage in other area papers.



Spotlight On... Nancy Rusin

About our daughter...
by Mary and Frank Burgo

Our youngest child, Nancy, was born on July 2, 1957. The doctor delivering her was almost speechless because he had never seen a baby who looked like her before. (She was born with lamellar ichthyosis.) He recommended that we not see the baby. He thought her strange appearance was an indicator of serious medical problems and that she might not live. In spite of these doubts, the doctor and our pediatrician set out to research this strange condition and care for Nancy. After two weeks we brought our daughter home.

We were totally unprepared to care for a baby with lamellar ichthyosis. We found she needed constant care to improve the very thick, dry, scaly skin, which seemed to impair her movement and make her uncomfortable. We bathed her as many as five times a day and used Vaseline, Eucerin, and other creams to help soften her skin and relieve her discomfort. A big concern was that she could not close her eyes; we worried that they would be damaged and she would not be able to sleep.

Over the years, we saw several different doctors and tried a variety of treatments: ointments, oils, vitamins, occlusion suits, and even Methotrexate injections. None offered any lasting solutions. Nancy took over her care as she got older, always on a quest for a lasting treatment, or hopefully a cure. Through this time she completed grammar school and graduated from high school. Her fellow students often subjected her to cruel words. Thankfully, Nancy had a group of very close friends, who continue to be her friends today, that protected her and helped her fight her battles.

Today Nancy is married to a very nice man and lives and works in Florida, where she is loved by a number of friends and co-workers. She takes excellent care of herself and is able to maintain a very normal appearance. Although there is still not a cure or greatly improved treatment for Nancy's condition, we all continue to search for help. The defective gene that causes lamellar



Nancy at age 7.

ichthyosis has been identified and hopefully a way to correct the gene will be found. Government funding and public contributions to research will expedite the cure.

About my sister...
by Janet Dooley

I was five years old when my sister, Nancy, was born. My mother came home from the hospital with no baby. My sister was alive, but she was in an incubator fighting for her life because of a rare skin disorder called ichthyosis.

Eventually, Nancy came home and the challenge our family would face really began. Nancy needed care beyond normal baby care. She was constantly uncomfortable and her skin needed constant care. My parents not only cared for her twenty-four hours a day but also took care of my brother and me. We never felt neglected or that there was no time for us. As small children, we never realized the full impact this must have made on our lives. Inside our home we lived normal happy lives. It was when we ventured outside our home that life got tough. We were constantly stared at or approached by strangers asking what was wrong with Nancy. They made us so uncomfortable that we gave up going out in public. But my parents made fun for us in other ways.

A swimming pool in our backyard created hours of entertainment for us and our neighbors and friends. Holidays and birthdays were made special by family traditions. Camping was a wonderful family experience, too. We would take my grandfather's camper and go to the Adirondacks or Thousand Islands. These trips involved a considerable amount of work for my parents. Because Nancy's skin did not allow her to sweat, she



Nancy at age 43 with her husband Jerry.

would run high fevers in the car while we were traveling. My parents would have to bring cool washcloths and ice packs to help bring her temperature down. We had a great time, but there was never any vacation for Nancy or my parents from the everyday routine of this disorder. Nancy still needed to keep her skin lubricated and had to sleep in a plastic suit to contain the moisture. She would lose her hair at times, and due to the tightness of her skin and eyelids, she was rarely able to close her eyes when she slept.

I often wondered and worried what Nancy's life would be like and whether she would have any of the wonderful experiences life has to offer. She was a genuinely happy child. She didn't seem to worry about tomorrow and pretty much dealt with what was happening today. I credit my parents with raising us all with a sense of normalcy. Nancy, also, was an inspiration to us. She fought battles every day to survive; some we were aware of, some we will never know. She tolerated unending physical pain, along with mentally and emotionally trying times. Hospital stays and cruel classmates were a fact of life. My parents, of course, never stopped looking for a cure.

It takes a tremendous amount of strength and a very strong family unit to endure the challenges this, or any other handicap, sets before you. We always believed that you are never given more than you could handle and that with prayer and a strong will you will succeed. Well, succeed Nancy did. She is married, living in a wonderful home in Florida, and has a great job. All due to the determination and perseverance of both Nancy and my parents.

WHAT I DID ON MY SUMMER VACATION

by Merritt Andrews, age 31

I was fortunate enough to attend Camp Discovery for Teens with rare skin disorders, July 28 - August 3, at Camp Knutson in Crosslake, MN.

Although I was there acting as a counselor, I was also a camper. I was never able to attend camp when I was younger. I was born with lamellar ichthyosis, so in the summer when my friends would go off to camp, I would stay behind. My parents worried about my suffering from heat exhaustion, public ridicule, and any other medical problems that might happen. I always felt like I had missed out on the camping opportunity. When I heard about Camp Discovery I jumped at the chance to be a part of it. Camp Discovery in Crosslake, MN, is sponsored by the American Academy of Dermatology and is under the guidance of Dr. Julie Winfield. This camp hosts teens, from 14-17 years old, and counselors with different skin conditions from all over the world. There are

two other camps sponsored by the AAD, both for children from 9 - 13. There is never a dull moment with activities running from 9:30 a.m. - 10 p.m. Teens enjoy horseback riding, canoeing, swimming, tubing, paddle boating, playing basketball, and doing arts and crafts. Every day has a different theme and there is a different activity held each night. There is trained medical staff on hand to help with bandage changes or emergencies. There are also dermatologists who can answer any questions. The facilities at Camp Knutson are so beautiful, with rustic cabins, hundreds of tall trees and huge lakes on either side of the peninsula.

At Camp Discovery I was "in charge" of seven girls, along with fellow counselor Missy. Most of them had lamellar

ichthyosis, one had Netherton's Syndrome, and one had Epidermolysis Bullosa (EB). We bonded quickly and became immediate friends. Some of the campers knew each other from previous years so, for some, it was an emotional reunion. I was touched by the fact that no one stood out as being different; we all had a unique sort of kinship, and everyone was accepted. We felt "comfortable in our own skin" and did not have to hide or worry about what others thought. We learned so much about each other and our struggles to fit in, and realized that we are not alone.

I encourage parents to send their kids next year, the lessons learned and the friendships made at Camp Discovery will last a lifetime. Contact the American Academy of Dermatology at 847-330-0230 (phone), 847-330-0050 (fax), or www.aad.org for details about how you can apply to attend the camps.



Camp Discovery in Millville PA

I was really excited to volunteer for a day at the camp. Now that I've had this experience, I plan on staying for more than one day next year. It was great. When I arrived, some of the campers were getting rides on the back of motorcycles by volunteers from the local Harley Davidson chapter. I was quickly assigned to the job of assisting campers with the cake decorating activity. I handed out cupcakes and the campers smoothed on their own colored icing and added candy of their choice. There were gummy bears, chocolate kisses, sour-patch worms, M&M's, Reese's Pieces, sprinkles, plus much more. How lucky I was to have been assigned to this activity; I was nibbling all day. The campers and counselors with ichthyosis were kind enough to pose for this photo. I had to interrupt their dinner of ham and cheesy potatoes, so I appreciate their patience. Can't wait to go back next year. **Jean Pickford**

Dr. Peter Elias Receives Award

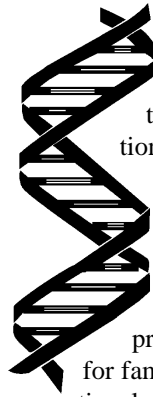


Dr. Peter Elias, a founding and current member of FIRST's Medical Advisory Board, was awarded the William Montagna Lectureship by the Society for Investigative Dermatology at the May, 2001 annual meeting. The lectureship honors and rewards investigators with primary emphasis in skin biology. Dr. Elias' lecture was entitled *From Basketweave to Biosensor: Evolving Concepts of Stratum Corneum Structure and Function*.

A pioneering researcher of the epidermal permeability barrier, Dr. Peter Elias was the first to propose that its structure is analogous to "bricks-and-mortar." His theory is now generally accepted in medical circles; he is extending his research beyond structural definitions to the barrier's functional and metabolic properties.

"The barrier function drives everything underneath; it's not just an end-product," Dr. Elias says. "It's the most important function of the skin and what allows us to live in a terrestrial environment."

Dr. Elias is a staff physician at the Veteran's Administration Medical Center Dermatology Service in San Francisco, California, as well as the head of its Dermatology Research Laboratory. He also serves as a Professor of the Department in Dermatology at the University of California, San Francisco.



People's Genome Celebration

The People's Genome Celebration was held June 8, 9, 10 in Washington D.C. Reflecting the theme, *Genetics is About ALL of us*, this free, educational forum brought individuals with genetic conditions, families, and representatives from government and industry together to focus public attention on the universal impact of genetics in the 21st Century. The celebration also observed the mapping of the human genome and highlighted the promises and implications of our historic genomic age for families and society. The program included informational sessions such as "Everything You Wanted to Know About DNA, But Were Afraid to Ask," as well as artistic exhibitions including "Genome Music."

In Memory of Janine Giuliana



Every day of her 23 years, our little daughter Janine's wonderful, agonizing life was filled with grace and pain, hope and suffering, and, most of all,

love. Love she gave; love she engendered. Janine gave the example of how to appreciate little things, how to suffer bravely, how to be concerned for others while your own physical life is a tragedy. Janine left this earthly life on May 25, 2001, surrounded by the love and sadness of her brothers and sister. Her mother and father had the honor of holding her little hands as she departed on the first trip she would take alone. Our deep sadness was tempered by the knowledge that our little treasure would, in moments, be running and jumping, free of pain, with her Heavenly Father.

Having suffered tremendously with a

grace and humor that hid her pain, Janine was ready to go to her eternal home. She had said she was tired of fighting, after her recent six-month battle with a barrage of illnesses at Children's Hospital in Philadelphia. Actually, epidermolytic hyperkeratosis had to get in line with her other problems, including, most recently, convulsions, fractured bones, osteoporosis, meningitis, kidney failure, ARDS, and a blood infection.

With all this, she loved life intensely. In the midst of her suffering, her focus was on people. Janine loved children, especially her nephews Justin and Nicky. Possibly her greatest joy was her godson Lito. She was so proud at his Baptism, her pain buried in happiness. One of her great desires was to volunteer in the Child Life playroom at St. Christopher's Hospital in Philadelphia. She interviewed and had her ID badge, but then became too ill to work there. A very special friend was her "companion dog," Vader. (She loved animals, too!)

Our days are longer now and our home seems much larger. Caring for Janine was a 24-hour job for 23 years. Gone are her hospital bed and Hoyer lift,

wheelchairs, crutches, and walker. Her oxygen generator no longer hums. The IV poles and pumps, the aspirators, the Nivea skin oil-gone. Syringes, tubing, bandages, eyewashes are no longer there. The drawer that was home to her 38 daily medications is empty. Visits to our pharmacist are fewer now. The van that gave Janine some pleasure in the last year of her life is gone from our driveway. The home care nurses, our constant companions over the past 5 years, no longer appear at our door.

Yet, thank God, so much of Janine is still here. Our memories of her are vibrant. We still marvel at her humor, her happiness, her strength. She is still here, in her books, crafts, dolls, and Beanie Babies. We still stop at her favorite stores in the malls. Janine is there. Her voice still answers our voice mail.

We learned love and strength from Janine. We learned caring, too. We admire people who, like Janine, accept their lives and work to succeed. We understand, now, how minor our "major" problems really are. Attitude is important. How we accept the challenges God gives us defines character. Character — it fits Janine so well. She had it. We must learn it from her. Thank you, Janine.

News & Notes

Funds Sought on Rare Diseases

Reprinted from the Boston Globe, Aug. 8, 2001



WASHINGTON — Senator Edward M. Kennedy has introduced legislation that would dramatically increase federal spending on rare diseases, including the establish-

ment of regional “centers of excellence” where research on the 6,000 rare disorders would be conducted. Boston is among the possible sites for such a center.

While many rare diseases are little known and affect only a small number of people each, the overall impact is large, with an estimated 25 million affected.

In the bill, Kennedy cited several examples of rare diseases, including cystinosis, a metabolic disease that causes havoc in kidneys and other organs and affects about 600 people in the United States. The Globe on Sunday profiled an 11-year-old Burlington boy, Shea Hammond, who has cystinosis and whose mother, Colleen Hammond, has been leading a fight for more funding for rare disease research.

Advocates for more funding have noted that research on

rare diseases has the potential for discoveries that can help uncover information on common diseases. Medical specialists said some of the most important discoveries about heart disease and stroke have come from the study of rare diseases, but funding of research on the lesser-known disorders has been minuscule compared with the amounts devoted to the most widespread diseases.

Kennedy called for doubling the funding for the Office of Rare Diseases at the National Institutes of Health to \$4 million annually; increasing drug research grants for rare diseases from \$12 million to \$25 million; and setting aside \$20 million to create the centers of excellence, where clinical trials could be conducted.

The location of the centers would be determined by independent specialists, with Boston among the possible sites, a Kennedy aide said. A companion bill would accelerate tax credits for “orphan drugs,” which are developed for rare diseases, a measure that may attract the most interest from biotechnology companies.

Kennedy, who filed the bill on Friday, said in a statement yesterday that he hoped the increased funding would “greatly enhance the prospects for developing new treatments and diagnostics, and even cures for literally thousands of rare diseases and disorders.”

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