



# ICHTHYOSIS FOCUS

Vol. 12, No. 3

A Quarterly Newsletter for Friends of FIRST

Summer 1993

## SPECIAL NATIONAL CONFERENCE EDITION CHICAGO '93

### OVER 200 F.I.R.S.T. MEMBERS GATHER IN CHICAGO FOR THE LARGEST NATIONAL CONFERENCE IN THE FOUNDATION'S HISTORY

More than 200 F.I.R.S.T. members met in Schaumburg, Illinois, just outside Chicago, over the weekend of June 25-26 for what proved to be by far the largest national conference in Foundation history, and by all accounts the most successful. Joined by researchers, physicians, and health care professionals from around the country, the group met for one and a half days of general sessions, seminars and discussion groups, and informal meetings on topics as diverse as retinoid therapy, nutrition and growth, genetic breakthrough, and topical treatment strategies. Several pharmaceutical firms joined the conference as technical exhibitors, sharing samples of their products and joining a running dialogue with F.I.R.S.T. members about treatment products and strategies for their use.

The conference kicked off Friday evening with a reception and opening session. Conference attendees heard from Board President Frances McHugh, who welcomed

everyone and introduced the Board of Directors. Regional Support Network National Coordinator Heather Gattuccio introduced regional coordinators and spent a few moments outlining RSN goals. Executive Director Nick Gattuccio then presented a review of F.I.R.S.T. over the past ten years, since the time of the Foundation's first national conference in 1983, which was also held in Chicago. The comparison offered a rich opportunity to measure the strides F.I.R.S.T. has made over the past decade. (You can read more about this in the *E.D.'s Desk* column on page three.)

This issue of *FOCUS* is devoted to extensive coverage of the Chicago conference. Although we cannot replace the conference experience for those of you who could not join us in Chicago, we'll try to give you a full overview of the conference and a rundown on the weekend's highlights. It was a powerful weekend, and one we will not soon forget.



Joshua Bates and John Beuhler typify the spirit of the conference.

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# Ichthyosis Focus

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The editor invites correspond-  
ence of any kind. Please offer  
advice, criticism, and by all  
means help us with ideas for  
stories. Tell us what you want  
to hear about, whom you wish  
to hear from, and what kinds  
of information you would like  
to see in *Ichthyosis Focus*.

Send your letters, suggestions,  
criticisms, ideas, and com-  
ments to the F.I.R.S.T. office  
in Raleigh. We'd like to offer  
the community an oppor-  
tunity to learn from your ex-  
periences, and we'd like to  
hear your opinions and views  
on subjects of importance to  
us all.

## C O R R E S P O N D E N C E C O R N E R

From Sapula, Oklahoma, one of  
F.I.R.S.T.'s founding members, Betty  
McMasters, writes: "Enclosed is our  
form and money to attend the '93 Na-  
tional Conference. We are all excited;  
our girls have never been on a plane be-  
fore!"

"I am so happy to see how this  
Foundation has grown. Katy was born  
on February 8, 1979, and I joined the  
Foundation and helped start things up  
along with two other families and two  
doctors back in 1980. I have the very  
first newsletters up to present date and  
how wonderful it is to read and see the  
growth. God bless you all and see you in  
Chicago."

Several people wrote to thank us for  
assisting with the distribution of samples  
of Dermal Therapy Research's urea-  
based lotion in March. Following is a  
sampling of the letters we received. Ja-  
nine Kiely of Jericho, New York, writes:  
"Thank you so much for sending the re-  
cent sample. It appears good for my  
needs." From Mohrsville, Pennsylvania,  
Janet Greusel writes to tell us, "Arrived  
home to Pennsylvania the middle of  
May from our winter home in Florida  
and was surprised to find a sample from  
Dermal Therapy. Found it a terrific lo-  
tion. Just placed an order today .... Can-  
not make the convention in Chicago this  
year, but my thoughts are with everyone.  
You are all doing a terrific job." To or-  
der, Dermal Therapy can be reached at  
1-800-668-8000.

"First let me thank you for the sam-  
ple of Dermal Therapy," writes Mary

Toth of Stratfort, Connecticut. "I use it  
and do like it. At present I am using Lu-  
briderm and NutraPlus together. I find  
the mixture does the trick ... it makes  
my skin feel human again. I use Lac-  
Hydrin on my feet, for it heals the  
cracks, which can be very sore."

Milton M. Crowell of Darmouth,  
Nova Scotia, Canada, writes: "Thank  
you very much for sending me the Win-  
ter 1993 newsletter. In it I read com-  
ments made by a Walter Ciszek of Cal-  
ifornia concerning a moisturizer he  
began using, apparently with excellent  
results. I took the newsletter to our fam-  
ily druggist and he mixed me a special  
lotion. Within days I noticed a marked  
improvement, and two months later  
there is absolutely no trace of scaliness.  
Thank you Mr Ciszek for the in-  
formation about a treatment that after  
seventy-four years is the only thing that  
corrected a very embarrassing skin con-  
dition. I am so happy with the result I  
might go so far as to apply for member-  
ship in a nudist colony."

Marjorie Locke kindly writes:  
"You're doing a super job and the news-  
letter is wonderful. I look forward to re-  
ceiving each issue. The newsletter intro-  
duced me to Aqua Glycolic lotion and I  
like it a lot. Keep up the good work."

Herald Pharmacal is still offering  
members of F.I.R.S.T. a special case  
price on their product, Aqua-Glycolic,  
of \$48.00 for 12 8-ounce bottles. Herald  
may be reached at their toll-free num-  
ber: 1-800-253-9499.

☛ Deadline for submissions to the next issue of *FOCUS*: October 1, 1993 ☚

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# NOTES FROM THE E.D.'S DESK

by Nick Gattuccio

It was ten years ago, in 1983, in Chicago that F.I.R.S.T. held its first ever national conference. The Foundation was then two years old. The conference was presided over by then executive director and board co-president, Charles Eichhorn. Fifty-five people attended. The conference was produced on a budget under \$400, while the Foundation as a whole lived on under \$10,000 for the



year. There was no significant discussion of biomedical research. The big topic was talk of impending plans for a new institute at the National Institutes of Health to be devoted in part to skin-disease research. There was also talk at the time of F.I.R.S.T. helping form a coalition of skin-disease groups into a Patient Advocacy Group to help foster this and other efforts.

It was fitting that our conference just past should also be in Chicago. Rather than completing a cycle, our returning to Chicago in 1993 signifies taking up a vantage point from which to assess the tremendous strides this Foundation has taken over the past ten years. Not 55, but 223 people attended this year's conference; and it was produced not on \$400, but rather on \$10,400—larger than the Foundation's annual budget in 1983. In 1993 advances in biomedical research occupied the lion's share of the general sessions, for there were so many advances to speak of, so many scientists to speak. Several of the most distinguished researchers presently working in the field of ichthyosis and keratinizing disorders were on hand to speak to us, to teach, and to learn from us, too.

Ten years ago the National Institute

of Arthritis, Musculoskeletal & Skin Disease (NIAMS) of the National Institutes of Health was only a rumor. Today it is a pillar of biomedical research in the ichthyoses and related disorders. And what in 1983 was a newly conceived Patient Advocacy Group, of which F.I.R.S.T. was a charter member, has grown into the Coalition of Patient Advocates for Skin Disease Research (CPA-

SDR), a coalition of 14 skin-disease advocacy groups representing the interests and speaking with the collective voice of some three and a half million people. Our presence on Capital Hill is prominent, and our commitment to the hot debates of the coming years is a full plate of critical health-care issues.

Indeed, 1993 in Chicago was not a mirror of 1983; instead, it was a looking post. From it we measure our progress. More important, though, we measure the richness of the opportunities before us in the decade ahead.

So what about the next ten years? What is in the cards for the year 2003? How will our conference look then? Who will be leading this organization then, if, indeed, the organization exists? What treatments for the ichthyoses will be in hand at that time, and how effective will they be? Will we still be managing symptoms, largely, or will viable and safe therapies exist? For how many of the ichthyoses will the genetics and biochemistry be understood? And what will American health care look like ten years from now? How expensive will it be, and what form will it take?

These are unanswerable question, of

*(Continues on Page 18)*

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# NATIONAL CONFERENCE COVERAGE

## CHICAGO '93

On the following pages we shall attempt to offer in words and photographs as full a coverage as possible of our 1993 national conference. Included are articles reporting on most of the conference sessions. Where possible, we offer transcripts of what actually occurred. In all cases we try to offer a flavor of the actual event.

### CONFERENCE KEYNOTE SPEAKERS: DR. PEYTON WEARY & JANET WEARY

**Editor's Note:** For keynote speakers we were honored to welcome Dr. Peyton Weary, Chairman of the Department and Professor of Dermatology at the University of Virginia School of Medicine in Charlottesville, Virginia. Dr. Weary is president-elect of the American Academy of Dermatology, and throughout his career has devoted himself to health-care policy issues and the politics of health care. Dr. Weary's wife, Janet Weary, is also a long-time supporter of F.I.R.S.T. Mrs. Weary offered a powerful presentation reflecting on the personal and familial aspects of living with ichthyosis. Her presentation left not a dry eye in the house Saturday morning. We felt it was worth offering a full transcript of her keynote address. Dr. Weary offered a perfect complement to Janet Weary's presentation, electing to focus on the most focal and critical political aspects of health care in America in the 90s. Dr. & Mrs. Weary are parents of a grown daughter with epidermolytic hyperkeratosis, and of a son with EHK who survived only three days.



Janet Weary

When our daughter, Terry, was born in 1954, my obstetrician and pediatrician did not know what was wrong. Finally, our pediatrician diagnosed epidermolysis bullosa, because of the extensive blistering. But Edward P. Cawley, then chairman of dermatology at the University of Virginia, called her condition congenital ichthyosiform erythroderma. Neither Peyton nor I had the condition, nor was there any record of it in either family. It was assumed to have been caused by recessive genes in the fortunately rare incidence of two parents each carrying the gene.

In 1954 genetic mutations were not every day news as they are today, and it was only years later that Terry's condition was recognized as having been caused in this latter manner, and was further identified by the name epidermolytic hyperkeratosis.

You would probably not be surprised at the number of doctors, including dermatologists, who continued to disagree or be unconvinced. Dr. Cawley became a pillar of strength for us, even from a distance, as we left the University of Virginia area for some years. I will never forget his kindness and patient council, then and throughout Terry's childhood. I would wish for each of you a doctor whom you can trust and the development of a true partnership between you.

When Terry was born my husband was still a student and we had to learn a lot of things you've had to learn and some you already know. We learned that cortisone cream (the newest miracle drug at the time) wasn't going to work and couldn't have been used continuously anyway. We learned about the need for prompt antibiotic treatment for infected blisters; and to this day Terry often has trouble getting antibiotics when she knows a strep infection is winning but her doctor doesn't.

We had so many trials and errors as far as treatment went that I no longer re-

#### Keynote Address by Janet Weary

I want you to know that I feel great admiration for you all. I am no longer parenting a child with ichthyosis, for she has, by any standard, achieved a large measure of contentment and success, and of course I never personally had to cope with the physical discomforts associated with ichthyosis which those of you who have the condition do every day of your lives.

It never occurred to me to try to create a national foundation, which you have done, and I stand in awe of your accomplishments in a world that is often not very supportive. Your knowledge now is much greater than ours was during the 50s, 60s, and 70s, and of course some of you are actively involved in sophisticated research in the field of skin disorders. But I know where you are coming from, as they say, because of the four children Peyton and I had, two were born with epidermolytic hyperkeratosis—a daughter and first child, who is now 38, and a son, our last, who survived only three days.

*We had so many trials  
and errors as far as  
treatment went that I  
no longer remember  
them all. I do re-  
member some foolish  
indulgence in day-  
dreams about magic  
cures.*

member them all. I do remember some foolish indulgence in daydreams about magic cures on my part, and some flirtations with treatments that were risky, impractical, or uncomfortable. Through them all Terry persevered.

We had to learn about her intolerance to heat and very cold, dry temperatures, and how to keep a swim in the ocean from being a misery for her while a pleasure for the rest of the family. We had to learn to accept her limitations as far as physical activity and we had to learn how important it was to teach the people in her life about them, and also of our concern for her emotional well-being.

We weren't always successful. We had some sitters who were afraid they would "catch it," children whose mothers were afraid they would "catch it," but for the most part intelligent people can handle what they can understand, and it is important to make the effort, even when it is not easy, to help them.

At the beginning of each school year, I went to the school first, before classes began, and talked with Terry's teachers about her condition, how it might affect her physically and emotionally. Without exception, I found her teachers to be cooperative, supportive, and very appreciative of the help this was to them. I did this for a number of years, until we felt Terry could handle these things herself, but even when she was nearly an adult, her father provided her with a letter of explanation—quite simple—just the name of her skin condition, that it was congenital and not contagious, and suggesting medications should she need a prescription from a strange doctor. She carried it with her to Switzerland one summer when she was 17 and had it at college if needed.

I am not going to tell you life was easy. You know it wasn't. We tried to teach our daughter that those who discriminated against her by words or actions needed to build themselves up because of their own insecurities. This is not an easy lesson for a little child to understand, but she tried. Our generation was also not brought up to be very open to others about our feelings, especially to our children (talk shows and self-help books were totally unknown to us), and we muddled through with our copies of *Dr. Spock* as best we could.

I knew absolutely no one who shared my problem. My support system was our wonderful parents, especially my mother who was never too busy to give me a break. They and our close friends treated all our chil-

dren with loving acceptance, and my husband always made me feel that he would take care of us.

It wasn't easy for our other daughters, either, and I wish that they had had a kind of "Ichthy-Anon," like Al-Anon, for siblings, because we were not able to discuss and share our feelings as much as we know now we should have. It was particularly difficult for them when our son died. If there is one thing I wish I could do over it would be to have been more aware of their pain in the midst of my own.

But now I see in these daughters the kind of compassion and empathy for others that seems rare among people, and they in turn will pass this rare quality on to their children. I see also that there is a more open communication between parents and children, more freedom to express thoughts and feelings on the part of both generations.

*I am not going to tell you life was easy. You know it wasn't. We tried to teach our daughter that those who discriminated against her by words or actions needed to build themselves up because of their own insecurities. This is not an easy lesson for a little child to understand, but she tried.*

I think it not surprising that as Terry grew up she became interested in medical fields and worked several summers as a candy-striper in our hospital. Certainly she became aware at that time of the vast numbers of people who have physical imperfections, ranging from acute and temporary, to the permanently disabling. Later, while she was studying at Bowman Gray to become a physician's assistant, she qualified to work as a nurse's aid. Eventually she worked briefly in Virginia as a physician's assistant. There were always friends, and she, like most of us, has remained in contact with the closest of them from high school and college.

Now she is married to a fine man and they have two children, whom they adopted, and who are two of our six, soon to be seven, grandchildren. The decision to adopt came after a long and difficult consideration of all aspects of their beliefs, feelings, and plans for their future, and is not a recommendation to others. It was simply the answer for them. After her children arrived, Terry decided to go back to school to take the necessary classes to qualify for an M.S., and eventually a doctorate. She is now a candidate for a Ph.D. in molecular genetics. She is involved in the lab in the uses of DNA and truly understands the work in this field which has given us new knowledge about epidermolytic hyperkeratosis. I know that she has been in contact with some of you involved in the research. I'm sure that even should she never directly contribute to a Big Cure, her involvement in this field brings her great satisfaction.

I asked our daughters what they would like to say

*(Continues on next page)*

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## Keynote: Janet Weary

Continued from Page 5

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to you who have ichthyosis, or to you family members.

Our youngest said, "Tell them that siblings with normal skin, because they were born physically 'perfect', can feel that they have no excuses not to be perfect. And when they are not, they can feel they have somehow failed. So-called 'perfection' can also be a burden."

Our middle daughter said, "If there is one thing I would change about how we dealt with ichthyosis as a family, it would be to increase the openness among us, because ichthyosis, by being unspoken of, can become omnipresent. Questions can become worries, and answers sought without council can suggest to the sister or brother that she or he is somehow responsible."

And this: "It is much easier, I think, to be grown up than to grow up. Maturity brings strength and the ability to shape good things from the stuff you are given."

And finally, Terry said, "Tell them how important it is for a person with ichthyosis to not feel identified *only* by his or her skin, but by the recognition of some positive thing, be it a special talent, ability, accomplishment, experience, service to others, adventure, achievement, etc. Look for your special thing in life, whatever it may be. This means, of course, that you must work for it, just as everyone else does. And it also means that those of you who are parents, siblings, and friends, must be supportive while also giving your child (sister, brother) the space to find his or her own independent life."

I think I see all of the necessary elements for this achievement in this room today, and I wish you success in reaching out more and more to others, wherever they may be.

Thank you.

# KEYNOTE ADDRESS TO THE F.I.R.S.T. NATIONAL CONFERENCE

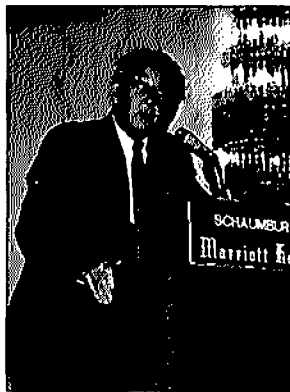
by

Dr. Peyton Weary, M.D.

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*Editor's Note: Following is a summary of, with excerpts from, Dr. Weary's Keynote Address to the Foundation. In the past, the Foundation's political agenda has been largely limited to participating with the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR) in efforts to educate*



*Congress and to advocate on behalf of increased funding for skin disease research, primarily for NIAMS. But times are changing. In the face of serious attempts at health-care reform, and with the emergence in some states of previously untested strategies for managing the distribution (or rationing) of health care services, new challenges fall upon the patients themselves, and upon patient advocacy groups like ours, to enter the debate in order to ensure satisfactory compliance with our own best self-interests. This is the subject of Dr. Weary's address.*

Dr. Weary issued the Foundation a political call to action at our national conference in Schaumburg. The call, he told us, is the result of several factors. On the one hand, there is a very real threat to continuing funding for biomedical research. "It is clear to me," he said, "that biomedical research support will be among the many government activities which may languish severely underfunded for the foreseeable future, as a result of the economic crunch caused by our budget deficit and huge national debt. Discretionary spending programs such as NIH, which account for a relatively small fraction of the over-

all budget, are the first to be cut and the last to be restored in tight financial times."

With this for a backdrop, Dr. Weary introduced a "matching funds" concept which he first developed in 1980. "The concept is to ask the Congress to provide matching funds to the

NIH for monies contributed by patient advocate groups and other philanthropic organizations for biomedical research." The concept is based on these matching funds *not* being used to reduce existing or future federal appropriations to NIH.

"The selling point here," he argued, "is that if the Congress were to provide dollar for dollar matching funds, this would create a substantial incentive for contributors to patient advocate groups, such as your Foundation, to increase their support and thereby raise additional funds for research."

Dr. Weary then turned to the issue of health care reform, which presents many formidable challenges to patient advocacy groups. He began by offering the opinion that "the American public has become persuaded that there is a health care *crisis* when in fact a true crisis does not exist." He acknowledges, however, that there exist "major problems, such as inadequate insurance coverage ..., and escalating costs of care fueled in large measure by a substantial increase in the elderly population, increasing availability of very expensive technology, and failure of the public and the physicians to reach any

(Continues on Page 15)

# BREAKOUT WORKSHOP: MOM'S GROUP

by Heather Gattuccio

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*Editor's Note: Among the most successful and well received of all workshops, the Mom's Group was led by Heather Gattuccio, with the help of Joann Villamarin, MD, a dermatology resident and mother of a child with ichthyosis.*

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At the Chicago conference I had the privilege, along with Dr. Joann Villamarin, of leading a mother's discussion group. About 30 women attended. The mission of the workshop was to provide a safe environment for mothers to discuss their feelings about raising children with ichthyosis, and to share their life experiences with the other women present. The only guidelines were that everyone should have the opportunity to speak, and that everyone recognize that all feelings are valid and that judgements should be kept at bay.

We began with introductions, followed with a free discussion of the best and worst aspects of raising a child with ichthyosis.

Most agreed that the most positive aspect of raising a child with ichthyosis is simply the children themselves—the joy they give us, their love, their laughter, their inquisitiveness—in short, their just being children, like all children. All of the moms present were proud of their children and grateful for their unique personalities and special talents. We discussed the importance of helping our children realize that they are not defined by ichthyosis, and that they



*Heather Gattuccio, with daughter Caitlin Irene.*

should find, and that we should focus on, the talents they excel at.

One mom mentioned that her family's experience with ichthyosis had brought them closer to God, while another said her dealings with the disorder allowed her to look beyond skin and to re-examine our society's quest for perfection. An-

other mother mentioned that, while she would never be happy that her child has ichthyosis, she was continually grateful for the many strong and supportive people who had entered her family's life because her daughter was born with it.

Emotions were soon palpable in the room, particularly when we began discussing some of the difficulties of day-to-day life with ichthyosis in the family. Luckily, many women came armed with tissue, which many of us used.

I discussed my fear for the day my two-year-old daughter, who is extremely self-confident and strong-willed, understands in a quick, cruel instant the reason for the stares

and the rude comments that strangers thoughtlessly toss at her, or me in her presence. Now she is oblivious, but one day all of the hypocrisy will click, and her self-confidence will be instantly crushed.

Tears were free as one by one some moms described the intense pain they felt watching helplessly as their children were hurt by the callousness and cruelty of ignorant, careless strangers.

One mom told of her young daughter's year-long struggle with an abusive classmate. This woman told how she reached for strength each day when her daughter came home from school with her feelings hurt and her self esteem in jeopardy. She told how she would save her weeping for the privacy of her bedroom when she knew her daughter was sleeping. She told of her ambivalence about involving herself in the classroom struggle, as she knew that her daughter would need to learn to fight her own battles. In fact, her daughter knew this, too, without explanation. The mom reinforced that it was the cruel classmate who in fact had the problem. Fortunately, many of her daughter's classmates joined forces and stuck up for her against the agitator.

Another mother, who has ichthyosis herself, described how difficult high school was for her because of the isolation and name calling. She told how she was barred from going on her senior class trip to Europe because the school administration decided that her skin was too severe a handicap. It was a crushing blow. This woman encouraged mothers to educate their schools and stand up for their children's rights.



*Madonna Taylor and Jill Wood.*



*Diane Beuhler with new daughter, Joshua with mom Cynnie Bates, and Bera McDonagh.*

*(Continues on next page)*

We talked for some time about handling questions from strangers, and all agreed that any given day's response depended largely on the day's mood. Some days we feel like being kind and informative; on other days we want to simply walk into a grocery store and buy our diapers or shampoo and not be hassled. We agreed that our response to strangers also depends on the approach.



*Joann Villamarin, Tiffany Karst, Debra Piercey, and Brian Karst with daughter Jessa.*

If the stranger is curious in a sensitive way, we tend to be kind in turn. However, if a stranger is accusing and cruel, as is so often the case, we have no problem telling them to leave us alone. One mom said she carried F.I.R.S.T. brochures in her purse to hand out to inquisitive strangers. Another mom said she likes to kill with kindness. With someone particularly thoughtless or relentless, she says, "Thank you for your kindness. I had no idea you were so interested in ichthyosis. Let me give you a card so you can send a donation to F.I.R.S.T."

In general, we agreed that the best approach was to keep it simple. It is exhausting and rarely productive to enter a conversation on the genetics of ichthyosis while picking up one's dry cleaning.

Soon, the conversation shifted to positive, creative ways we can help our children cope, not so much with their differences, but with what their society is going to make of them. One mother told us that she noticed over time that her daughter tended to fall into negative behavior to break up conversations in which she



*Betsy Bates-Freed speaking with Janet Weary.*

was discussing her daughter's ichthyosis. Once she understood this, the mom alleviated the problem by telling her daughter to take her by the hand and squeeze it twice any time the conversation becomes too personal or uncomfortable.

This mom reports that her daughter rarely squeezes her hand, and yet knowing that she can if she feels the need to has given her an important sense of control over her own life and the boundaries of her privacy.

Another mom described working with her toddler's pre-school teacher to make certain that her son never feels isolated or self-conscious during lotioning. When time came for lotioning, his class had a lotion party. The children delighted in slathering a mild lotion on their arms and legs while her son, inconspicuously, received his essential treatment. Another mom shared a similar story. Her daughter, whose EHK occasionally prevents her from wearing shoes, relies on the teacher to declare for the whole class a "no shoes day" at the appropriate times.

One mom proudly recounted her daughter's deciding to lecture her class on ichthyosis during their science hour. This brave talk answered many of her schoolmates' questions and alleviated a lot of unnec-

cesary curiosity. This mother, who also has ichthyosis, is amazed at how strong and sensitive her daughter is about living with ichthyosis.

The piece of advice we heard over and over again from experienced mothers was to TALK. The message: keep the lines of communication open—for family, friends, and school. Avoid making ichthyosis a taboo subject in the home. One mom especially encouraged open dialogues within the family. Her son, she said, began internalizing many of his negative feelings, acting out his anger in destructive ways. This mother felt strongly that if we sensed that our children did not feel comfortable talking to us about their ichthyosis, then we should find a means for them to speak to someone else.

We also recognized the importance of our having the opportunity to talk, whether it be to other mothers, a spouse, friends, or professionals. We discussed the pressures of being forced into the role of paraprofessional—



*The Mom's Group*

that is, being required to be quasi-doctors, nurses, advocates, defenders, housekeepers, entertainers, and, of course, moms on top of everything else.

After only an hour and forty minutes our time was up. I think we could have continued talking most of the day, and maybe even into the night. For many of us, it was empowering to surround ourselves with other women who share our unique experiences, and who can relate so deeply to our most subtle feelings. All of our lives are different, of course, but they are the same.

We connected in this room, and as most of us know, half the battle of ichthyosis is knowing that one is not alone.



# BREAKOUT WORKSHOP: YOUTH GROUP & PANEL DISCUSSION

by Dr. Sharon Berry, Ph.D.

*Editor's Note: Among our attempts to draw our young people into the Chicago conference was the youth group. Dr. Sharon Berry, a pediatric psychologist with Children's Memorial Hospital in Chicago served as moderator and facilitator for the panel. Following are comments by Dr. Berry, and following that are excerpts from the panel itself. Individual speakers are not identified so that their privacy may be preserved. About thirty-five young people and a handful of adults attended the group session. Youth panel members were Wendy Vaughn, Tim Ohlwiler, Tonya Richardson, Eric Beeler, and Sandeep Bhatt. All did a fantastic job sharing their experiences and teaching those younger than themselves. Hats off to all of them!*

The Youth Panel at the F.I.R.S.T. national conference in Chicago was impressive and exciting. This stellar group of young people provided information which was both informative and motivating. I felt moved beyond words with many of



*Youth Panel (from l. - r.): Sandeep Bhatt, Tonya Richardson, Wendy Vaughn, Tim Ohlwiler, and Eric Beeler. Standing is Dr. Sharon Berry.*

the stories they told about their first-hand experience living with ichthyosis and related skin disorders.

The young people on the panel offered many helpful suggestions, and their comments focused on the skin disease playing a role as only a minor aspect of their lives and personalities. Everyone was reminded that no one completely understands what they go through, even the specialists, and each plays a unique role in educating others. They all agreed, very adamantly, that parents and family members should not treat them "special or different."

Each person was reminded that the doubts of others challenge them to believe in themselves even more. Suggestions were offered as to how to handle the most embarrassing or difficult questions from others. They recognized that they needed to be "bold" and work to believe in themselves even when given feedback such as "you'll need to find a career in the back somewhere, where you are not so noticeable."

A school principal in the group (and grandfather of a child with ichthyosis) encouraged the young people to use their school principals and teachers for advocacy, encouragement, and protection. Most agreed that it was helpful to start out each school year by informing teachers and other students about their medical condition. Younger children would likely need the help of their parents, but older students described successful exper-

iences with this open approach.

All agreed on the need to have good friends who could be well-informed and supportive. For those encounters that are embarrassing or humiliating, Wendy has developed a very helpful and enlightening method of providing a business card (which she shared with those in attendance) which provides information about herself and a number to call for questions (and also put the person in their place in a nice sort of way!) Everyone loved this idea.

I was honored to be part of this moving experience, and proud for each of the panel members and those in attendance for their strength and courage and for their motivation to become the best person they can be.

*Following are excerpts from the transcript of the Youth Panel. Only minor editing has occurred, and this only to make the text more easily readable.*

• The comment I get most often is the "sunburn" one. I just tell people I have a skin condition. I don't bother to go into detail. On a bad day, when people say the wrong thing, I deal with it by playing the piano or listening to music, or I go jog. It's important to get the feelings out of my system. *(Girl, 15 years old)*

• I'm going off to college next year. I've been very lucky by the reaction I've had from friends and family. They are a very accepting community, so I always feel free to speak out about my feelings, to keep a dialog going. This has helped me avoid the instinct to be an outsider. Don't depend on your peers; take advantage of your parents' and relatives' life experience. *(Boy, 18 years old)*



*Crystal Rose Tumlinson*

• I have a job as a checker at a large department store, so I have to deal with people every day, and this can be frustrating. People don't care what they say. They blurt our rude things, like "Looks like we've stayed in the sun too long." I say, "No, I'm not that stupid." Sometimes I feel like I should wear a sign saying

*(Continues on Next Page)*

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## YOUTH GROUP

(continued from page 9)

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"Sunburned/Burned in a Fire/None of the Above." But it gets hard. Some days I can deal with it, some days I can't; then I get angry at customers and blow up. I deal with it by writing poetry. People might not know what the poems mean, but I do. Listening to music helps, too. I become a stronger person as I get older. Some days I want to lay back and not do anything because I'm so sick of it. Sometimes it's so hard I don't want to live this way anymore—deal with people's cruelty. When it gets bad I sometimes go to a professional, but they don't understand. People always say they understand, but they don't.

I think I'm becoming a better person, though—I mean, better at handling this. I'm excited to be out of high school, though, because there was so much peer pressure—having to be pretty, popular, to play sports and all of that. It was very hard in high school. I'm excited to be going to college. (*Girl, 18*)

• I think ichthyosis is what got me into college. I wrote an essay about the difficult experiences I have had and how I handled them. My worst experience ever was when I was young and I went to the hospital with my parents to see my new cousin who had just been born. But at the hospital they said I couldn't see the baby. They were afraid of my skin. My mom argued with the nurse, but it was no use. This really affected me. I thought that if anyone should understand, it should be doctors and nurses. I thought they should have confidence in what you know about ichthyosis. The nurse made me question what I knew was

true. She made me doubt myself. (*Boy, 18*)

• [from adult]: I worked in public schools for 32 years, and I can tell you that you must be very bold. You must stand out instantly. You must hone your skills and talents. You should go to the principal and assistant principal and make people hear you at your school. It is very important to educate the people around you. You have rights, and you must make people at your school face you. (*Grandfather of child with ichthyosis*)

• Middle school is the toughest age group because in middle school everyone wants to be alike, wear the same clothes, be the same. This will pass after a while. Soon, nobody wants to be like anyone else.

• Because of my ichthyosis and the difficult times it has made for me, I feel more mature than others my age. I feel like I know more about life than all my friends whose biggest worry is their hair.

• Truly cruel people are the ones who have the real problems. Different isn't bad.

• Sometime you just don't feel like dealing with it. But then some rude stranger comes up and wants to know what's "wrong" with you. If he wants to know if it's contagious, tell them "yes," and then give them a hug!

• Keep looking for people to talk to who understand. They may not understand exactly, but they can understand your feelings. If people you talk to don't understand, educate them, or find someone else. You don't have to get stuck talking to somebody who isn't listening the way you want them to.

• *A survey of the stupidest questions we get:*

- Do you have cancer?
- What's *wrong* with you?
- Are you on steroids?
- Didn't you take a bath?
- Are you sunburned?
- Were you burned in a fire?

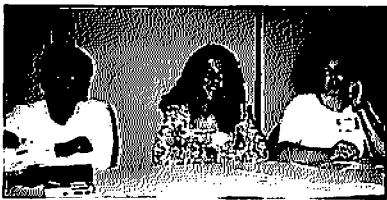
• Sometimes I'm straight up with people. Other times I'm not. It's a lot better now than when I was younger. The only one who can stop us from doing anything is ourselves. (*Adult*)

• Speak up right away. Nip curiosity in the bud. Try to stop rumors quickly. It might be hard at the time to talk to your class about ichthyosis, but in the long run you will end up

getting fewer hassles.

• Sometimes teachers can be rude and that bugs me. It's very disappointing when adults shame me. I somehow expect to be protected by adults. You have to do a lot to educate adults.

• Knowledge is the best thing. Everyone has individual differences genetically. Kids have to know concrete answers so they don't begin to believe something is really wrong with them.



Panelists Sandeep Bhatt, Tonya Richardson, and Wendy Vaughn.



Attendees of the Youth Group make ready.

# THREE OF THE COUNTRY'S MOST DISTINGUISHED RESEARCHERS IN ICHTHYOSIS ADDRESS CONFERENCE

Drs. Peter Steinert and Sherri Bale of NIH, and Dr. Elaine Fuchs of the Howard Hughes Medical Institute Share Their Important Work

F.I.R.S.T. members had the unique opportunity to hear from three of the country's most distinguished researchers in the field of keratinizing disorders, among them ichthyosis. Drs. Peter Steinert and Sherri Bale of the Laboratory of Skin Biology at NIH/NIAMS, and Dr. Elaine Fuchs of the Howard Hughes Medical Institute at the University of Chicago, represent two of



Left to right: Dr. Peter Steinert, Ph.D., Director of the Laboratory of Skin Biology at NIH; Dr. Sherri Bale, Ph.D., Chief of Genetics Section, Laboratory of Skin Biology, NIH; Dr. Elaine Fuchs, Amgen Professor of Molecular Biology, Howard Hughes Medical Institute, University of Chicago.

the country's three laboratories which, in the fall of 1992, virtually simultaneously announced the discovery of the location of the gene for epidermolytic hyperkeratosis (EHK). (The third lab is that of Drs. Dennis Roop and Joseph Rothnagel of the Baylor College of Medicine in Houston, Texas.)

The presentations were so interesting because of the study in contrasts. While the NIH team (led by Drs. Steinert and Bale) arrived at their findings by way of "classical genetics" techniques (that is, statistically evaluating linkage analysis studies of genetic material obtained in blood samples from large families in which EHK runs through several generations), the team from the Howard Hughes Medical Institute, led by Dr. Fuchs, arrived at virtually the same conclusions from a very different direction. Rather than genetic material from humans, Dr. Fuchs' team actually engineered in mice the human genetic defect which they hypothesized would account for EHK, and then evaluated the outcome of this mutation in the skin cells of offspring of the altered mice. Indeed, these "transgenic" mice exhibited the predicted outcome—the symptoms of EHK.

In fact, there is not one but two mutant genes which can account for symptoms of EHK. To further complicate the findings, either of these two genes may have mutations at any of several points along the gene itself. To understand why this is so, the researchers pointed out that the basic biological defect which accounts for EHK is a disruption of the structure of skin cells which lie in a thin layer just above the base layer of the epidermis. The defect lies in thin strands of keratin filament. These keratin filaments, which give the cells

their structural integrity, are actually made up of two types of keratin protein (known by the numbers K1 and K10). A defect in either one of these two ingredients of the keratin filaments will lead to the structural abnormalities which result in the symptoms of EHK. Because the genes which control the production of each type of keratin are located on separate genes, a defect on either

one of these genes can result in defective keratin filaments, which in turn results in the symptoms of EHK.

Nearly everyone familiar with EHK knows there are enormous variations in severity from one affected person to another. The second part of the work done by these researchers addresses this issue, and has to do with the surprising finding that not only can the mutation exist on either of two separate genes (i.e., the K1 gene or the K10 gene), but that the mutation can be located at any of several points along either of the respective genes. The researchers have offered a working hypothesis that variations in the way EHK is manifested among different individuals may be explained, at least in part, by differences in the precise location of the mutation along these two culprit genes.

This is only a hypothesis, of course. Nothing conclusive has been shown on this point to date, although investigations are underway. As the genetic information on more and more people with EHK is analyzed, ultimately researchers will have enough information to catalog the various point mutations and compare these with variations in physical symptoms. If the hypothesis is correct, a correlation will emerge between the symptoms and the genetics of the disease.

The Chicago conference provided an ideal opportunity for the research community to mix with the lay community, for both to learn from one another, and for each to increase their understanding of our separate worlds. In the laboratory both worlds come together daily in the "test tube"; however, it was important for all concerned that the two worlds meet face to face, for a change—for one day at least.

# A PORTFOLIO OF IMAGES FROM CHICAGO '93



Maggie the Clown entertains children in the child care room with balloons and stories, games and all 'round good cheer.



Wendy Vaughn entertaining young Meade Piercy of Ogdenburg, New York.



Host physician for this year's national conference, Amy Paller, M.D.



New friends—Rita Tanis of Fairfield, Connecticut, Elise Johnson from Seattle, and Michelle Petersen from Metuchen, New Jersey.

In the main auditorium during the keynote address.



John Beuhler and Caitlin Gattuccio size up one another's balloons during a lull in the day's events. In the background, l. to r., Lori and Mark Rogers of Lenoir City, Tennessee, and Barbara and Dave Rockwell, of Mississauga, Ontario Canada.

Four women enjoying a break in the action, modeling the Chicago '93 commemorative T-shirt.



Cathy Sipper of Rutledge, Alabama, with her son, Zebulun.

Revelers at the Saturday night party. In front is Sandeep Bhatt and behind him his father, Kanu.



Heather Gattuccio with Dave Purcell of Houston, Texas.



Haley Rice, looking things over!



Board meeting Sunday morning: (l to r) Ginna Franz, Tom Beuhler, Georg'Ellen Betts, Virginia LaBrant, Nick Gattuccio, Frances McHugh, Lynne Alba, Cynn timer Bates, and Deb Vilas. Not pictured are Pam Stockton and Betsy Bates-Freed (taking the photo).



The McDonagh family: Vera and John with daughters Sonia, 11, and Cassandra. Cassandra's t-shirt reads: Skin Disease / Contagious / NOT!"



Peter Gass with two of his five children, daughter Emily, 3, and son Eric, 5.



Keynote speakers and special guests Janet and Dr. Peyton Weary.

Lynne Alba reads a story to children in the Child Care room. Beside her is one of the Eastin twins, Tamara or Jalene (only her mom knows for sure). Lynne & Harry Alba were stars of the show for making the day's child care a huge hit for all!



Louis A. Semento, President of Neo-Strata, one of our wonderful group of pharmaceutical exhibitor's at this year's national conference.



Board member Georgie Betts handing grand prize to Eric Beeler of Tucson, Arizona. Eric won two round trip tickets to any destination in the U.S., courtesy of Travel Agents International, our official conference booking and travel agent.

The group enjoys a crowded Saturday night party after being rained out of the outdoor circus tent. The room was small, but we had fun regardless.



Little Larissa Martin with one of our child care workers.

Right: The Lauters family plus one: (l. to r.) Kathy Lauters, Rita Tanis, Gary 3rd and Kevin Lauters, and Gary, Sr.



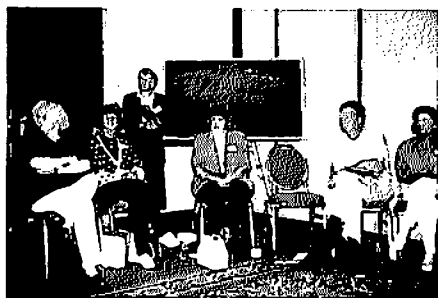
# BREAKOUT WORKSHOP: ADULTS WITH ICHTHYOSIS

by Deborah B. Vilas

Twenty-five joined in the discussion group for adults with ichthyosis. We were lucky to have some spouses, parents, friends and partners of adults with ichthyosis present as part of the group. I served as

facilitator of the group, less because I am a member of the Board of Directors of F.I.R.S.T. than because I myself am an adult with ichthyosis—congenital ichthyosiform erythroderma (CIE).

We got the conversation started by asking each person to pair off with someone else in the room whom they did not know. For ten



Part of the Adults With Ichthyosis discussion group. Standing is Deb Vilas.

minutes, the pairs interviewed one another, collecting some personal data, such as type of ichthyosis (or what their relationship was to someone with ichthyosis), where they hailed from, and what positive things had happened to them through having ichthyosis, as well as challenges that they found difficult.

When the interviews were completed, each person was asked to introduce their new friend to the group. I made a list on a blackboard of both the positive aspects and the challenges of having ichthyosis while people shared their stories.

Several points came up again and again. Those adults who had supportive friends, spouses and families were extremely grateful for their love and support. Significant others shared how special it was to be close to someone with ichthyosis.

Under the heading of challenges, it was fairly unanimous that we all get tired of answering the ever present questions which the public always seems to have about our appearance. However, several participants shared humorous, upbeat accounts of how they cope with the incessant questioning.

To be sure, living with ichthyosis is a challenge on one level or another for everybody. But when people come together to share their experiences, a common bond is forged that makes each person feel that they are not alone in their struggle. Those who joined in this discussion should be congratulated for their openness, warmth and courage in reaching out to others like themselves. We are not alone.

## NUTRITION & ICHTHYOSIS: A CONFERENCE WORK GROUP

by Donna Rice

A quick look around the Chicago conference room revealed a startling number of small children. I knew the growth and nutritional concerns that we are currently experiencing with

our 17-month-old daughter, Haley, were not unique for children with lamellar ichthyosis, but I did not realize how widespread the problem was. As I opened the nutrition discussion group parents began to talk of the long term problems they've



Donna Rice and daughter Haley.

experienced with their children's poor growth and nutrition. Apparent was the lack of solutions, the seriousness of the problems, and a lack of research literature or other information that might help resolve these issues.

Our own search for solutions to Haley's growth problems began when her slow but steady weight gain began to fall off when solid foods were introduced at 7 to 8 months. We became desperate to

find ways to help her gain weight when at one year of age she weighted just 12 pounds (the average weight for a six month old), and her developmental skills were significantly delayed. A literature search, discussions with other F.I.R.S.T. parents and Haley's physicians revealed that failure to thrive in children with ichthyosis can be caused by an increased metabolic rate resulting from excessive skin production, loss of protein through the skin, poor iron absorption, and fine-motor and feeding problems. We saw that Haley's nutrition problems were multifaceted, and that our experience feeding our other children would be of little help.

Shortly after Haley's first birthday, we consulted with Dr. William Klish and his staff at Texas Children's Hospital's Nutrition Clinic in Houston, Texas. This clinic takes a

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**Keynote Address: Dr. Peyton Weary**  
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agreement on acceptable levels, or targets, for utilization of these expensive services."

The question, though, is how these problems will be addressed in the next few years. There is a great deal of speculation on this count, but Dr. Weary outlined what he termed the "four emerging patterns [for health care reform] under consideration":

1. **Radical Reform Proposals** (also called *single-payer systems, national health insurance, or Canadian-style system*). "Under this system, as in Canada, the government would regulate the health care system, budget for hospitals, eliminate private insurance, set physician fees, and theoretically hold down costs by capping health care expenditures." The system would be financed by taxes. Although the Clinton Administration has reportedly rejected this concept, many powerful individuals, among them Congressman Pete Stark and some other legislators, favor it.

2. **Employer-Based Reform** (also called *play or pay, or employer-mandated insurance*). "This is basically the pattern adopted some years ago by the state of Hawaii, and which is now being instituted or considered by some other states.... It would require all employers to either provide health insurance for their employees, or, if unwilling to do so, pay a tax to help support a risk pool for purchase of insurance by a state or federal agency for uncovered employees.... To cover the unemployed, Medicaid would be expanded."

3. **Incremental Adjustments**. "There are a number of incremental reforms which have been proposed, some of which are aimed at correcting a single problem, and some designed to correct several problems." Among

the health care reform issues dealt with singly or in groups by this method are "insurance reforms, such as requiring portability of health insurance from one job to another, community rating instead of experience rating..., elimination of exclusions for pre-existing diseases, and prohibition of prolonged waiting times before coverage commences.... Other proposals would deal with such things as malpractice reforms, creation of administrative simplicity, and reduction in expensive regulations."

"There are many who feel that an incremental reform approach may be the best and most practical one in the long term given the lack of consensus on many issues at this time. Furthermore, many states are already moving ahead with various incremental reforms, and there are those who feel this is probably a good thing as a way to pilot-test some of these changes.... I would direct your attention to the existence of minimum benefits packages and also to the proposals for Medicaid expansion, both of which are now underway in the state of Oregon."

4. **Managed Competition**. "Although not enough information is yet available about the Clinton Administration proposal [forthcoming from the Health Reform Task Force, led by Hillary Clinton], most of the conventional wisdom suggests it will be a hybrid of what was originally termed 'managed competition'."

Based on a proposal for health care restructuring made by the so-called *Jackson Hole Group* led by economist Alain Enthoven and a physician, Paul Elwood, managed competition would promote, through legislation, the creation of massive Health Maintenance Organizations (HMOs) based on regional delivery networks. "These organized delivery systems, which would be, in effect, large HMO conglomerates, would be expected to

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**NUTRITION & ICHTHYOSIS**  
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multi-disciplinary approach to nutrition problems and includes a gastroenterologist, dietitian, occupational therapist, physical therapist, and speech therapist to evaluate all aspects of nutrition, growth and feeding. Evaluation of Haley's metabolism and daily food consumption revealed an increased resting metabolic rate and a daily calorie intake insufficient for growth. Although all children have different rates of metabolism and calorie consumption, Haley's increased metabolism required her to eat 50% more

calories per day than was average for a child her age. A feeding evaluation indicated the possibility of poor oral-motor integration, or a "hyper-sensitive" mouth, and that her feeding schedule was not allowing her to get hungry and thus be motivated to eat. Tests to determine vitamin and mineral levels are pending.

Dr. Klish and staff recommended a nutritional supplement (Pediasure) to increase calories and protein without requiring Haley to eat more. A rigid feeding schedule was designed to allow Haley to be motivated by hunger instead of "grazing" all day, and an oral-motor stimulation program to "desensi-

tize" her mouth.

I'd like to report that we went home armed with these results and that Haley began to eat and grow. However, as noted by other parents in the discussion group, providing our daughter with proper nutrition is time consuming, requiring constant attention and patience. We continue to search for answers to this complex problem, and like many parents we spoke with in Chicago, we continue to be concerned by the lack of medical research about what is to us a very serious problem affecting all aspects of our children's health and well being.

compete with one another for health insurance contracts from industry and from the public, presumably by offering *cost-effective, high quality, reasonably-priced basic benefit packages*, and also higher-option packages for those willing to pay extra. In effect, these would become the supermarkets of health care, virtually eliminating the solo, or small group, practitioner. An independent *National Health Board* would be created to oversee the program, determine what benefits would be included in the standard benefit package, what deductibles would be required, and to monitor the quality and outcomes of care." There are a great many other details associated with the plan, particularly fiscal issues keyed to the so-called "global budgeting" concept. Essentially, though, "it is this hybrid which combines managed competition with global budgeting which is said to comprise the Clinton Administration plan.

"There are many detractors as well as supporters of managed competition. Many are skeptical of the acceptance by the public of the supermarket approach to health care and the disappearance of the independent practitioner. Many are skeptical of the ability of managed care to control costs, and there is *significant* societal resistance to forcing a restricted-access "gatekeeper" pattern upon the majority of our citizens who are accustomed to free choice of physician and direct access to specialist care.

"This brings me now to the two areas in which I feel the patient advocate organizations *must* become involved and pro-active because health policy decisions in these two areas will have a potentially *very serious impact* upon your constituents."

#### *Minimum Benefits Packages*

"The first of these is the trend toward development of minimum benefit packages, or standard benefit packages." This is essentially a list of medical conditions and procedures detailing which will, and will not, be reimbursed. "Most of the health care reform proposals indicate that they will develop and define minimum benefit packages. This is the centerpiece of the Clinton proposal as well, and reportedly that proposal will offer a very generous package as an inducement.

"It is predictable that cosmetic procedures and treatment of purely cosmetic disorders will not be covered under any minimum benefit package, and this is one of the areas that should be of concern to the Coalition of Patient Advocates for Skin Disease Research. What is cosmetic? How do you define it? Who defines it? What is the boundary between cosmetic and disabling? All of these are questions that must be addressed if your constituents are to be treated fairly."

The so-called "Oregon Plan" results from an initiative in the State of Oregon to provide Medicaid coverage for a broader segment of the poor and near poor.

The state established a system of assigning priority numbers to virtually all common diseases or disease groups—745 in all. By Oregon's system of prioritizing these diseases, ichthyosis was rated 740, just five slots from the bottom, and nearly 150 slots below the exclusion line. Dr. Weary analyzes weaknesses in the system employed by Oregon officials in prioritizing diseases, and offers modifications which could minimize unfair exclusions, particularly for diseases which may be *interpreted* to be cosmetic. "Finally," he said, "I believe it is essential for patient advocate groups to be at the table when minimum benefit packages are being developed because they are the ones who are *best* able to evaluate the disability, disfigurement, and socioeconomic consequences of the disorders of their constituents."

#### *"Gatekeepers" & Direct Access to Dermatologists*

"The second area in which I feel the patient advocate groups must be proactive relates to yet another basic health policy issue. Most managed care systems are structured about the primary care physician defined as family doctor, pediatrician, and general internist as the so-called gatekeeper. The patients are required to consult the gatekeeper first for any and all problems, and can be referred on to other specialist *only* with the gatekeeper's approval.

"Now, some gatekeepers are ready to refer to dermatologists with little prompting, particularly if they have little interest in, or knowledge about, skin disease. Others are reluctant to refer, particularly if they have a strong financial interest in limiting referral (and I might add that *most* gatekeepers do have a financial incentive to limit referrals to specialists). Thus, the patient may be denied direct access to the specialist best able to diagnose and treat the patient's disease." Dr. Weary identifies four essential reasons why "dermatologists have long been critical of the gatekeeper pattern," which include visual experience, familiarity with latest treatments, and minimum need for expensive diagnostic and therapeutic interventions.

"Because patients with diseases represented by our Coalition members have disorders which are more complex, chronic and disabling, and would be poorly served by the imposition of impediments to direct access to dermatologists, we believe your organization should want to speak out in favor of allowing patients to have *direct access to dermatologists as first contact physicians*. We will be soliciting your support over the next few years as we attempt to foster the concept that providing direct access for patients to dermatologists is both good and cost effective medical care.

"Again, my sincere congratulations to you for the leadership you have already shown and which I know you will continue to provide."



# LEGISLATIVE UPDATE

## *In A Difficult Budget Year Spiced With Deficits And Partisan Squabbles, Funding For NIH and Biomedical Research May Suffer*

As reported in the last issue of *FOCUS*, we are in the midst of a most difficult federal budget season. Appropriations for the National Institutes of Health (NIH), and particularly to its National Institute of Arthritis, Musculoskeletal & Skin Disease (NIAMS) for biomedical research appeared dismal in April. The outlook has improved somewhat.

The House and Senate appropriations committees (and their respective subcommittees) are right now completing work on fiscal year (FY) 1994 appropriations legislation. (The 1994 fiscal year begins October 1, 1993).

President Clinton's budget, which was released in early April, dealt a hard blow to biomedical research. His budget request for NIAMS called for a nearly \$2 million funding cut, or about one percent below current funding levels. Combined with biomedical research inflation, which runs at about 4% per year, NIAMS stood to lose about 5% in FY 1994.

The cuts reflect two overriding

factors. First, the well publicized effort to attack the federal deficit has led to extremely conservative spending goals. Second, while overall spending has been constrained, limiting total dollars available to NIH, strong pressure to enhance federal support of certain high-priority research issues (specifically, AIDS research, women's health issues—particularly breast cancer research—the tuberculosis crisis, and the Human Genome Project) has resulted in a shifting around of the fixed dollars available to NIH.

In short, the budgets of some NIH Institutes like NIAMS were undercut in order to free up research funds for these high-priority research projects.

Of course, no one questions the high priority given these most critical American health-care issues. The disappointment comes from the "rob-Peter-to-pay-Paul" method by which the Clinton budget seeks to fund them.

However, a presidential budget is only a budget *request*. Congress, not the President, controls the

country's purse strings. And the House and Senate Appropriations Committees, and their respective subcommittees, play a dominant role in the funding process.

On June 8th, the House Appropriations Subcommittee that controls funding for NIH (the Subcommittee on Labor, Health and Human Services, Education, and Related Agencies), which is chaired by Congressman William Natcher of Kentucky, began their "mark up" of FY 1994 appropriations legislation for NIH. This House version restores many of the President's cuts, offering NIAMS a budget of about \$223.3 million, which is an increase of nearly \$13 million over the President's request, and a little over \$11 million above FY 1993 funding—a funding increase of just over 5.7%. This figure is slightly exceeds the biomedical inflation rate. The House appropriation, then, although not offering significant gains for NIAMS, at least holds off the real-dollar losses found in the President's budget.

However, the process is far from finished. The Senate has yet to act. The Senate bill will almost certainly differ in some respects from the House bill, and the two bills will then need to be reconciled. The final version of the FY 1994 appropriations bill should appear in the early fall. We'll keep you posted on developments.



## WRITE TO CONGRESS



It is most important that your Senators and Representatives hear from you. They do read your letters, and they do listen to what you say.

Listed here are members of the House Appropriations Subcommittee which oversees funding of NIH and NIAMS. They play a most powerful role in the funding of biomedical research in America, and ultimately, in funding research in ichthyosis. Write and tell them every dollar spent on re-

search returns over \$10 back to the economy. Tell them you support biomedical research, and that it is an essential investment in America's future. Tell them that part of the solution to America's health care crisis begins in the research lab.

Address your letter:

The Honorable John Doe  
U.S. House of Representatives  
(Office number/building)  
Washington, D.C. 20515

### House Appropriations Subcommittee for Labor, HHS, & Education

William H. Natcher (Chairman) 2358 Rayburn	[Dem, KY]	Jose Serrano 336 Cannon	[Dem, NY]
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Louis B. Stokes 2365 Rayburn	[Dem, OH]	C.W. Bill Young (Ranking Republican) 2407 Rayburn	[Rep, FL]
Steny H. Hoyer 1705 Longworth	[Dem, MD]	John E. Porter 1026 Longworth	[Rep, IL]
Nancy Pelosi 240 Cannon	[Dem, CA]	Helen D. Bentley 1610 Longworth	[Rep, MD]
Nita M. Lowey 1424 Longworth	[Dem, NY]	Henry Bonilla 1529 Longworth	[Rep, TX]

## Region Five Conference Slated for October 9

Joe and Marlene Huffman, along with Paula Lange, are presently bringing together another Regional Conference for Region Five. The Conference is set for Saturday, October 9, 1993, at the Holiday Inn South in Rochester, Minnesota. For more information contact Marlene or Joe at 612-864-4508.

## Let's Face It Publishes Resource Catalog

Let's Face It, an international organization dedicated to providing information and support to people with facial differences, their families, friends and professionals, has published a resource catalog. Following two items drawn from it. In future issues of *FOCUS* we'll provide further listings.

### *The Exceptional Parent Magazine*

Published eight times a year for parents of special-needs children. Articles are written by parents,

health care and legal professionals, and individuals with disabilities. Each issue provides an extensive "Forum" section (input and information from readers), and an annual guide to products, services, schools, camps, state resources, etc. Subscriptions are \$18 per year. For information call 1-800-247-8080.

### *Paying the Bills: Tips for Families on Financing Health Care for Children with Special Needs*

This 56-page detailed and clearly written guide is considered an "excellent resource" for parents of special needs children. Those living in New England can receive a copy of it free. Those outside of New England may receive a copy by sending a 9 x 12 inch self addressed stamped envelope (\$1.05 postage) with a letter of request to:

Paying the Bills  
New England SERVE  
101 Tremont Street, Suite 812  
Boston, MA 02108  
Phone: 617-574-9493

## FROM THE E.D.'S DESK

(continued from page 3)

course, but each one is of great importance. They are important because the quality of the questions we ask today will shape the debate we partake in tomorrow.

And the debate will be most critical for us all. Life for a family living with a chronic disorder is not unlike a palm tree in a hurricane. There is great commotion and peril, yet little is expected but that we bend. Indeed, much will change—*must change*—in the coming decade. We may elect to bend and let it happen to us, unwitting and passive, or we can choose to participate in the debate. We are duped if we believe we have no voice, and paupers if we take no stake in our views and opinions.

It *will* be an interesting ten years. I hope to meet you all at our national conference in 2003 (perhaps it will be in Chicago again) so that we can once again measure a decade past, compare our notes, and look toward to ten years more years.

## *In Memorium*

### Harold Wiens

Last month we heard from F.I.R.S.T. member Janice Richmond, of Marion, Kansas:

"My father, Harold Wiens, died unexpectedly on April 17, 1993. His granddaughter (my daughter, Lindsey), who is 3 years old, has lamellar ichthyosis. The family has decided to donate part of his memorial money to F.I.R.S.T. I know it would have pleased my father greatly to know some of the money was donated to this organization."

Our sincere condolences to you, your family, and to the friends of Harold Wiens. Please accept our gratefulness at your having thought of us—our community—at this most difficult time.

## Ellen Rowe Receives F.I.R.S.T. Distinguished Service Award

Ellen Rowe, past President of the Board of F.I.R.S.T., received at a special ceremony in Chicago a the 1993 *F.I.R.S.T. Distinguished Service Award* to recognize her extraordinary contribution to F.I.R.S.T. over the past five years. In addition to a commemorative plaque, Ellen was presented with a special resolution from the sitting Board recognizing the impact on the organization of her years of service.

Indeed, at a time when this organization suffered from a serious energy drain, Ellen moved F.I.R.S.T. from Oakland, California, to Raleigh and helped stabilize the Foundation in its present form. Recent efforts to carry this Foundation to even greater prominence stand on the shoulders of Ellen's important contribution.

# N . O . R . D . R E P O R T

*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.*

## Quote of the Day:

"Most members of Congress are still driven by that letter from home. You know the kind of letter that is written on the paper with blue lines and five holes and a coffee mug ring on the upper corner ... and the first consultant that can get real people to say what they really want is the one who's going to win the health care issue, not the slickest commercials." (Ex-Rep. Dennis Eckart, D-OH)

## N.O.R.D. Opens Washington Office

NORD recently opened an office in Washington, D.C., to educate policy makers and advocate for people with orphan diseases during the current health care reform debate. NORD's new Director of Policy is Glenda Booth.

To help convince Congress to address the needs of people with rare disorders, NORD's Washington office needs examples of problems patients have had with the health care system, such as denial of Medicare, Medicaid, or private insurance for the following reasons:

- Pre-existing conditions;
- Refusal of reimbursement for "off label" use of prescription drugs;
- Expenses associated with therapies;
- Seeing an appropriate specialist for treatment of their rare disorder (especially refusals of HMO's to refer outside of their networks).

## Orphan Drug Update

Proposed amendments to the

Orphan Drug Act are expected to reach Congress again this year. Senators Howard Metzenbaum and Nancy Kassenbaum and congressman Henry Waxman are preparing revisions to the Orphan Drug Act. The amendments are aimed at closing unintended loopholes in the law which open the door to excessive profits. Prior efforts at enacting these amendments have failed in the past. Then President Bush vetoed

one such effort, and last year's initiative died in Congress.

On a related issue, NORD is presently preparing suggestions for a bill that will create an Office for Rare Diseases in the National Institutes of Health. Senator Mark Hatfield is expected to introduce this legislation soon.

## Profits, Profits, Profits ...

A recent study by Harvard University's School of Public Health found that Boston's non-profit tax-exempt teaching hospitals "have amassed billions of dollars in surplus cash since 1984." In 1992, they ended the year with \$1.1 billion in discretionary funds which "could have reduced daily hospital rates by up to \$281."

# G R A S S R O O T S

The Dystrophic Epidermolysis Bullosa Research Association of America (D.E.B.R.A.), and organization much like ours representing the interests of patients and families living with epidermolysis bullosa (EB), recently completed a week-long nationwide public-education and grassroots fund raising campaign which they called *EB Awareness Week*. In addition to a media blitz of articles in local media prompted by D.E.B.R.A. members in their local communities, many families successfully promoted and secured local resolutions and proclamations on the part of their local governments promoting awareness of EB. Another part of EB Awareness Week involved a flowering grassroots fund raising effort. I'll let them tell the story themselves:

"EB Awareness Week also spurred families and friends to conduct creative fundraisers on behalf of EB awareness and research. Tim

and Jean Anderson of Albin, Wyoming, held a bowl-a-thon to 'strike out' EB and raised over \$1,600. Connie Swett and her students in Tennessee raised over \$2,000 in a 'piano practice-a-thon,' and Rita and Jeff Slifer of Richmond, Indiana, held a 'putt-putt for D.E.B.R.A.' and raised over \$4,000. Our Michigan chapter held a canister drive and concert. Throughout the country, other D.E.B.R.A. members held bake sales, flea markets, and walk-a-thons to promote an awareness of EB and to raise funds for research. [Reprinted from the *D.E.B.R.A. newsletter*, EB Currents.]

Congratulations to Miriam Feder, executive director, and to all the folks at D.E.B.R.A. on their successful effort. Their dedication and hard work serves as an example to us all. I think they will not be offended if we consider 'borrowing' a few of their ideas in the future.



*FOCUS is printed on recycled paper.*

## Regional Support Network

The Regional Support Network is in place to facilitate communication between members of the ichthyosis community. Parents can exchange practical tips and child-care information. Adults can support one another by sharing experiences in job hunting, self-treatment, medical issues, social coping strategies, and other issues. If you wish to join the support group in your area, please contact the representative for your region. *If you wish to join your area's Network, call your Regional Coordinator or F.I.R.S.T.'s toll-free number to receive the "pink" registration form.*

### REGION ONE

NY, PA, VT, NH, NJ, CT, RI, MA, ME  
Deb Vilas  
7 East 88th Street, #4-B  
New York, NY 10128  
(212) 427-5904

### REGION FOUR

KY, OH, IN, IL, MO, MI  
Cynnle Bates  
133 Elam Park  
Lexington, KY 40503  
(606) 276-0142

### REGION SEVEN

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

### REGION TWO

DE, MD, WV, VA, NC, SC  
Robin Joyce  
Route 1, Box 189-F  
Sandy Ridge, NC 27046  
(919) 871-3277

### REGION FIVE

WI, IA, MN, ND, SD, NB  
Joe & Marlene Huffman  
1326 DeSoto Avenue  
Glencoe, MN 55336  
(612) 864-4508

### REGION EIGHT

CA, NV, UT, AZ, HI  
Mark & Claudia MacNaughtan  
610 East 800 North  
Genola, UT 84655  
(801) 754-3064

### REGION THREE

TN, GA, FL, AL, MS, LA, AR  
Jennifer Tomasik  
880 Lake Overlook  
Roswell, GA 30076  
(404) 587-1886

### REGION SIX

TX, OK, NM, CO, KS  
Donna Rice  
2902 West Elm Circle  
Katy, TX 77439  
(713) 391-4407

### NETHERTON'S SUPPORT GROUP

Cathy Sipper  
P.O. Box 127  
Rutledge, AL 36071  
(205) 335-6827

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# Y E S !

*I Want To Help F.I.R.S.T. Help Others  
In The Fight Against Ichthyosis*

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Mail to: F.I.R.S.T. / P.O. Box 20921, Raleigh, N.C. 27619