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

ICHTHYOSIS FOCUS

Vol. 11, No. 4 A Quarterly Newsletter for Friends of FIRST Fall 1992

AMERICANS WITH DISABILITIES ACT

by Nick Gattuccio

Congress Passes Law Ensuring Civil Rights Protections to Persons With Disabilities

SCENARIO: A woman with ichthyosis vulgaris is hired as a food server in a local restaurant. Her manager is an old friend. Although the restaurant's uniform calls for a short-sleeve shirt, all agree that this woman will wear long sleeves to cover signs of ichthyosis on her arms. The woman's hands are also affected. However, within three days a customer complains vociferously that she will ***not*** *be served by this woman. A scene erupts. The manager supports the employee and attempts to explain the situation to the outraged customer, who then storms out of the restaurant. The manager calls the restaurant's owner to appraise him of the situation. Surprisingly, he orders the woman with ichthyosis be fired. The manager is dumbfounded and quits her job in protest.*

Prior to July of this year, the woman with ichthyosis had no civil rights protections against job discrimination of this kind. Now, such discrimination is against the law. We're happy to report that the waitress has an attorney and the restaurant is on the run.

Signed into law on July 26, 1990, and set to begin going into effect in July, 1992, the Americans with Disabilities Act (ADA) guarantees America's 43 million citizens with disabilities the same wide-ranging civil rights protections that were extended to women, minorities and others in the Civil Rights Act of 1964. Covered by the ADA are those with physical or mental impairments that "substantially limit activities such as working, walking, talking, seeing, hearing, or caring for oneself." The law also protects individuals who have a history of such an impairment, and those who are *regarded* as having an impairment. In short, conditions which people *perceive* as being disabilities (for example, mild cases of ichthyosis) qualify under the law.

The ADA bars discrimination in employment and requires employers with 25 or more employees (15 or more beginning in 1994) to make "reasonable accommodations" for "qualified employees with disabilities." (See definitions of key terms in sidebar.) Also covered by the law are accommodations for disabilities in the realms of public services and public transportation; the design of public accommodations (hotels, motels, theaters and concert halls, shopping malls, etc.); and in the future design of telecommunications equipment.

There are numerous stories of in-

(Continues on page 4)

TEACHING TEACHERS

by Deborah Brewster Vilas

Any parent of a child with ichthyosis can tell you: an informed, sensitive and communicative teacher can make all the difference in your child's success at school. However, educating school staff about the medical and psychosocial needs of your child is a challenging task. Most lay people have never heard of ichthyosis, and they may find it difficult to understand the unique challenges and limitations that the disease may involve.

With this challenge in mind, I am in the process of writing a manual for teachers. The manual will have classroom teachers as the intended audience, including those teaching at the preschool, elementary and high school levels. However, the information included will benefit every level of school employees, from the bus driver to the nurse and phys-ed coach. The manual will include a brief overview of ichthyosis, its symptoms and treatment, as well as practical advice for teachers looking to meet the physical and developmental needs of children with ichthyosis.

FIRST has graciously offered to publish and distribute this manual to parents, so that you will have a tool with which to approach the schools involved in your child's education. The expected date of publication is early 1993. In the meantime, I need your help.

Many parents and children have already contributed generously to the content of the manual by participating in phone interviews and by sending me artwork, some of which will hopefully be part of the finished product. But I am still looking for more input. I am very intent

(Continues on Page 9)

IN THIS ISSUE:

Ichthyosis Focus
Vol. 11, No. 4
Fall 1992

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

P.O. Box 20921
Raleigh, North Carolina
27619-0921
*
919-782-5728
800-545-3286

President of the Board
Frances McHugh,
Acting

Executive Director/
***Focus* Editor**
Nicholas Gattuccio

F.I.R.S.T. is a 501(c)(3) charitable organization supported by private donations.

The editor invites correspondence 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 comments to the F.I.R.S.T. office in Raleigh. We'd like to offer the community an opportunity to learn from your experiences, and we'd like to your opinions and views on subjects of importance to us all.

CORRESPONDENCE CORNER

Ann Hicks in Pulaski, Virginia, writes: "I have a son named Clinton Dwayne Meadows who was born March 13, 1991. He was a collodion baby. I would love to hear from other people who have children with this disorder. Please write me at P.O. Box 2461, Pulaski, VA 24301, or call (703) 980-5526."

Beth Dzilinski in Tallahassee, Florida, wants to know if anyone has experience with seborrheic keratosis (horny growths that, when removed, leave unsightly white spots or reddish bumps). Write Beth at Route 3, Box 579J, Tallahassee, FL 32308.

From Walnut Creek, California, **John and Dorothea Sanderson** send FIRST a donation and write to tell us that Herald Pharmaceuticals' Acqua-Glycolic Lotion "...clears my arms of scale very well. If I let it go it takes about two weeks to clear, but if I use it daily the scale keeps clear."

Alice Workman of Great Falls, Montana, sends FIRST a donation and tells us about her daughter, **Annastasia**, whom the Workmans adopted at two months (she is now 17): "I will never forget young children coming over to see my baby exclaiming, 'Oh, her hands are dirty,' or 'What's wrong with her'? When she was in kindergarten her teacher called me to investigate 'how she had been burned.' Annie had told her teacher that she had burned her hands because she was uncomfortable with the teacher's questions about them. I think her worst hurt to date over her ichthyosis must have been her eighth grade ballet recital. At the end of the recital all the troupe lined up for a final bow. They were all to join hands. The girl next to Annie dropped her hand as soon as it touched Annie's and pulled away. I know Annie must have been both hurt and embarrassed. Annie is a strong individual and has developed a toughness where her ichthyosis is concerned. She has had to do this."

Lydea Mansfield of Fremont, Michigan, who has ichthyosis vulgaris, writes to tell us that she recently discovered a most helpful product, Avon's new Cellulite Contour. She writes: "I'm not sure if it has done anything for my cellulite, but I was amazed how soft and smooth my skin was and the dry scales were gone ... It is not greasy, it smells good and a little goes a long way."

From Aztec, New Mexico, **Lee Lawrence** writes to tell us that he uses a small-toothed dog grooming brush (a flicker comb) before shampooing to remove scales to allow the shampoo to penetrate. Scales and flakes don't return for two days, he reports.

We heard recently from young **Tonya Knuesel**, from Maplewood, Minnesota: "You sent me some information for my local science fair. I would just like to thank you. I won 1st place at my science fair and Grand Prize and was invited to the Twin Cities Regional Science Fair at Augsburg College. At Regionals I came away with 2nd place!

I not only came away with the red ribbon, but also knowing I have informed a lot of people. I interviewed an 11-year-old boy with ichthyosis. Many of his classmates had questions for me, showing that they had never been told why his skin was different. The parents of these children also appeared to be learning."

Congratulations, Tonya, from all of us at FIRST for your great contribution to educating your community about ichthyosis.

From Roselle, Illinois, we received a delightful letter from eighteen-month-old **Tomas Buehler** (with a little help from his parents **John T.** and **Dianne Buehler**) who attended the Region IV Support Network Conference in Indianapolis in August (see a full report on this conference on page 6). Thomas writes:

"I am pleased to enclose donations from my family and friends in the amount of $1,245.00 to support the FIRST Foundation, and to aid in the current research project in Lamellar Ichthyosis, which is the form of Ichthyosis I have.

"Hopefully these donations will help to find a cure or at least relief for ichthyosis. I am eighteen months old and I am looking forward to the day that more information is available.

☛ Deadline for submissions to the next issue of *FOCUS:* **January 5, 1993** ✍

"This is our first effort to raise money for FIRST, and next year we hope to raise even more for the Foundation. My parents and I appreciate having an organization such as FIRST to help us in dealing with this condition and the information you've given us regarding treatment and research."

All of us at FIRST extend our great appreciation to the Buehler family and friends for their hard work and extraordinary generosity!

Kudos to **Karen Jones** of Johnsonburg, Pennsylvania, who continues to raise money for FIRST, year after year, with small projects such as craft sales at the annual Christmas bazaar ($250 in 1991). This year Karen raised $50 through an aluminum can recycling project. Small innovative efforts such as these go farther than anyone can imagine toward keeping FIRST afloat.

Thanks to you, Karen, from all of us at FIRST, for your generous, unselfish efforts!

TELL ME DOCTOR

by Melodie Buxman, M.D.

Q. I have Ichthyosis vulgaris. I was diagnosed with this about two months ago; until then I was told I had "dermatitis." My skin is very sensitive and I break out and turn red often. I have tried many lotions and creams and nothing seems to help until I get a cortisone shot. That makes my skin temporarily almost normal. Please help. It's very depressing, and I've had a hard time dealing with this.

A. You undoubtedly have both Ichthyosis vulgaris and what is known as "atopic dermatitis." These conditions occur together fairly frequently and often there is a family history of eczema, hay fever, or asthma.

In atopic dermatitis the skin does not hold water normally and dries out easily, leading to cracking, inflammation, and severe itching. Superficial skin infections with bacteria such as staphylococci are common. Sometimes atopic dermatitis is difficult to treat effectively. Keeping the skin hydrated with emollients, and even increasing the humidity of your environment, say with a humidifier, will help to prevent outbreaks. We try to maintain the red breakouts with mild corticosteroid ointments like hydrocortisone, increasing the strength as necessary to control intermittent flares. If the dermatitis is out of control, and not responding to these measures, antibiotics orally may be of help.

Be sure your bath or shower is short and warm rather than hot, and apply moisturizer immediately upon exiting the shower rather than waiting until the skin is dry. Also, make sure your soap is very mild—glycerin soap is both inexpensive and effective. Avoid deodorant soaps as they may be too harsh.

One exciting area of research in atopic dermatitis is the use of derivatives of the thymus gland to treat severe atopic dermatitis. Some individuals have been able to get remission of six months or more with injections of these derivatives. Hopefully, relief is on the way!

Melodie M. Buxman, M.D., a dermatologist on the F.I.R.S.T. Medical Advisory Board, will be happy to answer your medical questions about ichthyosis and secondary problems. Please send your questions to Dr. Buxman care of the F.I.R.S.T. office (anonymously, if you wish) at FOCUS: P.O. Box 20921, Raleigh, NC 27619-0921.

F.I.R.S.T.'s 1993 NATIONAL CONFERENCE SET FOR CHICAGO

Planning is well underway for the 1993 FIRST National Conference to be held in June, 1993, in Chicago. The Next issue of *FOCUS* will offer a full rundown of itinerary, schedule, location, prices, and room rates and availability. Because the final details of the Conference have yet to be firmed up with our prospective meeting site, these specifics were not available at the time this issue went to press.

It has been two years since our last National Conference, in Williamsburg, Virginia. A great deal has occurred over these two years, both within the Foundation and in the world of biomedical research. The Chicago conference offers a vital opportunity to all members of the Foundation to meet fellow members, to meet the people behind the names in the Foundation's leadership, and to also meet some prominent members of the medical field who specialize in the ichthyoses.

Over the next few months the Foundation will work aggressively toward developing funding of a membership scholarship fund to help enable some members to attend the Conference who might not otherwise be able to make it for financial reasons. We hope to be able to offer at least partial cost offsets. Stay tuned to *FOCUS* for more on this.

THE AMERICANS WITH DISABILITIES ACT

(Continued from Page 1)

dividuals with ichthyosis encountering discrimination in the workplace. This usually stems from employers unreasonably assuming that physical appearance might somehow compromise their company's operations. However, under the new ADA, employment decisions (including job assignment, salary, and promotions) that are based on such assumptions are against the law.

Furthermore, an employer is required by the ADA to make "reasonable accommodations" to make the workplace or job assignment more accessible to persons with physical impairments. This may include air-quality and temperature regulation in the workplace. It may also include "restructuring jobs, setting up part-time or modified work schedules, purchasing or modifying equipment or devices, or modifying examinations, training material or policies."

These "reasonable accommodations" are required of employers ***unless*** they result in "undue hardship"—that is, unless they result in significant difficulty or expense to the employer. Factors considered in establishing undue hardship include "the nature and cost of the accomodation, and the financial resources and overall size of the business in terms of the numbers of workers, the number of facilities, and the structure and function of the workforce."

An employer may ***not*** submit a job applicant to a pre-employment medical exam ***unless*** such exams are required of all job applicants—that is, without regard to disability. Physical exams may ***not*** be used to determine whether a person has a disability or to evaluate its nature or severity. However, the employer may inquire whether the applicant can perform job-related functions.

In short, federal law now protects individuals with disabilities from unreasonable behavior on the part of employers. Not only must those with disabilities be offered the same opportunities and afforded the same rights as employees without disabilities, but employers are required to make an extra effort to make such opportunities and rights accessible to those with disabilities.

DEFINITIONS OF SOME KEY TERMS

DISABILITY

- A physical or mental impairment that substantially limits one or more of the major life activities, for example, walking, seeing, speaking or hearing;
- A record of such an impairment, for example, a person who has recovered from cancer;
- Being regarded as having such an impairment even when no limitations exist, for example, a person who is scarred from burns.

QUALIFIED INDIVIDUAL WITH A DISABILITY

- An individual with a disability who, with or without **reasonable accommodation**, can perform the essential functions of the employment position that such individual holds or desires.

REASONABLE ACCOMMODATION

- Making existing facilities used by employees readily accessible to and usable by individuals with disabilities;
- Job restructuring, part-time or modified work schedules, reassignment to a vacant position;
- Acquisition or modification of equipment or devices;
- Appropriate adjustment or modifications of examinations, training materials or policies;
- Other similar accommodations.

UNDUE HARDSHIP (for employer)

Means an action requiring significant difficulty or expense. Factors to be considered in determining whether an accommodation would cause an undue hardship include:

- The nature and cost of the accommodation;
- The resources and size of the business as a whole and of the facility making the accommodation;
- The type of business operation, including the composition, functions and structure of the workforce;
- The impact that the accommodation would have on the facility making it and on the business as a whole.

In general, a larger employer will be expected to make accommodations requiring greater effort or expense than a smaller employer.

This information provided in a publication of the President's Committee on Employment of People with Disabilities.

IF YOU FEEL THAT YOU'VE BEEN SUBJECTED TO DISCRIMINATION IN THE WORK PLACE

The Equal Employment Opportunity Commission (EEOC) has been charged with enforcing employment discrimination provisions of the ADA. You may receive additional information from the EEOC: 1801 L Street, NW, Room 9024, Washington, DC 20507. 1-800-669-3362. There is also an EEOC Regional Office in your area. Check your phone book.

You may also be in a position to file civil suit against an employer you suspect of being in violation of ADA provisions. You should consult an attorney who specializes civil rights/employment discrimination law. Consult your state's Bar Association for information and referral.

The ADA ensures that "no person can be discriminated against because he or she has made a charge, testified, assisted, or participated in an investigation, proceeding, or hearing under ADA." In other words, you are protected from recriminations made by an employer you seek to challenge under provisions of the ADA. Furthermore, the winning party in an ADA action "may be awarded a reasonable attorney's fee, including litigation expenses and costs."

F.I.R.S.T. PERSON

F.I.R.S.T. MEMBERS TELL THEIR OWN STORIES
IN THEIR OWN WORDS

Ichthyosis & Discrimination

by Chandra Foote

I have never viewed myself as handicapped or disabled. I am just as capable as any member of society. But that statement in itself reveals a prevailing attitude in society—that the handicapped and disabled are not as capable as people without a disability.

I have congenital lamellar ichthyosis, but I have never let my skin condition slow me down. I graduated from high school after being a member of the speech team and editor of the school newspaper. I am now a sophomore at the University of Oregon, where I am a full-time student, a manager of the U of O's campus radio station, and where I do free-lance writing for the college newspaper and a campus magazine. I also work part-time. But I have had to put forward an enormous effort to get where I am today.

While I have only been a member of the work force for five years, I have already experienced discrimination in the workplace. This first occured in high school when I was looking for an after-work job. A friend told me that a video store where she worked was hiring, so I applied that afternoon. The manager told me she wasn't hiring anyone right then, but I was welcome to fill out an application. I mentioned this to my friend a week later, and she told me they'd hired three people just the day before. I hadn't even been called for an interview. Applying at another job, the manager told me she couldn't hire me because the other employees would be disturbed by my appearance.

I sometimes feel as if I am a member of a silent minority. Unfortunately, our minority doesn't have a Mahatma Ghandi or a Martin Luther King, so people's perceptions of the handicapped and the disabled are changing at a snail's pace. However, there is something new that may help. The Americans with Disabilities Act (ADA).

Although the intent of the law is to discourage employers from discriminating against disabled applicants, the law does not automatically guarantee that a handicapped applicant will get a job, even if he or she is equally as qualified as another candidate. Instead, the ADA is a "strategy to provide the job applicant information after the fact," according to Larry Smith, Director of the Career Planning and Placement Office at the University of Oregon. In short, the law offers handicapped applicants legal recourse in situations where they feel they may have been discriminated against.

What is needed for the ADA to be truly effective, though, is for people to go out and apply for jobs. The law won't be effective if people aren't out there not just looking for work, but also finding out why they're not getting hired. According to the Spring 1992 issue of *Field Guide to Computer Careers*, there are 16 million disabled people of working age, but only four million are working.

If you don't get a job, find out why. Smith commented that you should "call them up, say 'help me understand the difference in the qualifications of the person you hired compared with mine.' They may not tell you anything, but they also may." This serves two ends. First, it can help you find areas where you might be able to make improvements in the way you present yourself as an applicant. Second, it might help you uncover situations where you truly should have been hired, and that's were the ADA comes in.

All in all, if the disabled don't go out and apply for jobs, discrimination won't be stopped, the ADA won't be effective, and worst of all, we won't have jobs. We are just as qualified and just as capable as anyone else, and now all we have to do is make people realize this.

Chandra Foote is a sophomore at the University of Oregon in Eugene, where she continues an active schedule and her fight against discrimination.

WITH GROWTH COMES CHANGE

A Report on Changes in FIRST's Leadership

If the good news to report upon completion of an active 1992 is tremendous growth and remarkable developments for the Foundation—a doubling of our membership rolls, unprecedented gains on the legislative front, and spectacular advances on the biomedical research front—then the flip-side is continuing change within the Foundation itself.

Over the past months FIRST has lost the services of two key planks in our foundation. Ellen Rowe, for several years a cornerstone of our organization, has stepped down as Board President. We thank her sincerely for the pivotal, invaluable service she's rendered over the years. Frances McHugh has assumed the role of acting president. Also departing the organization is Susan Snyder, our first Executive Director. Susan shepherded the Foundation through those critical early months of growth and change with a strong and steady hand. We all owe Susan our gratitude for her unswerving commitment to our cause.

Departing FIRST's Board of Directors is Heather Gattuccio. Fortunately, though, Heather's services will not be lost. With her husband, Nick, and daughter, Caitlin, Heather has moved from Portland to Raleigh, where she will assume the role of Coordinator of FIRST's Regional Support Network. In September the Board appointed her husband, Nick, to the position of Executive Director, and he assumed his duties in Raleigh on November 1st. Heather resigned from the Board due to Nick's appointment, to ensure no conflict of interest.

Finally, let's welcome to the Board Deborah B. Vilas, of New York, New York. Deb is presently completing work on her Masters degree in special education with the goal of becoming a child life specialist. She has completed her manuscript, *Teaching Teachers*, a handbook for teachers of pupils with ichthyosis (to be published by FIRST in 1993). She is on the staff at the Sloan-Kettering Cancer Center in New York. Deborah will surely prove to be an invaluable addition to our Board of Directors.

The F.I.R.S.T. Regional Support Network

INSIDE THE RSN

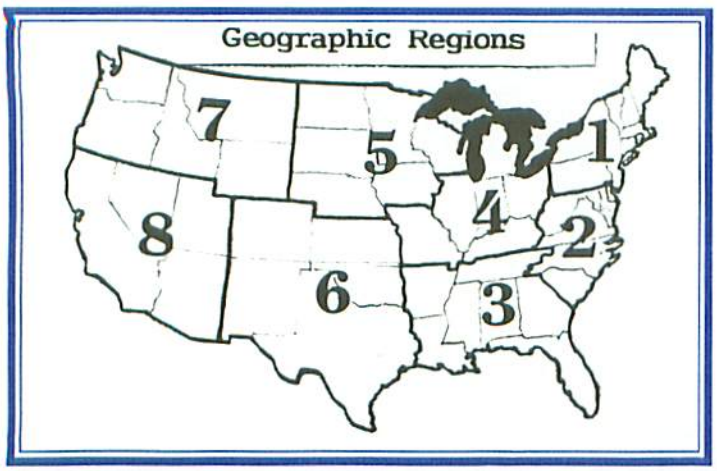
by Cynnie Bates

The Regional Support Network (RSN) is the helping hand of F.I.R.S.T. outstretched to its widely dispersed community. Reaching out to serve the ichthyosis community with information and support would be impossible without the volunteers of the RSN. We recognize that as the Foundation grows and matures, the price we pay for this growth is an organization that is in danger of being less accessible to the community we serve.

Initiated in August, 1991, the RSN has strived to offset this danger, going into the community, acting as a localized resource, providing information, reducing isolation, and offering opportunities to give and receive support, practical advice and encouragement.

We have divided the country into eight regions. For each region there is one Coordinator and one or more Representative available to serve the region. Coordinators and Representatives are volunteers, each of whom has personal experience with ichthyosis—either they have ichthyosis themselves, or they have a close family member with it.

We recognize that the word "support" in the program title means different things to different people. Individuals need different types of support at different times. Parents of a child newly born with ichthyosis often need more complete information about the disorder. They need practical day-to-day caregiving information, and they may need emotional support while adjusting to this new turn in their lives. A school-age child, on the other hand, wants to know there are others in the world like him or her, who also have to put on lotions, and who also deal with awkward or painful situations at school. Finding a pen-pal is one way that children with ichthyosis can both give and receive support. Sometimes siblings of a child with ichthyosis wish to talk with another sibling about what it is like to grow up with a brother or sister who has ichthyosis. Finally, we know that adults, too, often wish to communicate with one another, to share skin-care tips and compare notes, and the RSN strives to bring each region's adults together too. We understand that "support" comes in a variety of forms, and we strive to provide for them all.



Region I: NY, PA, VT, NH, NJ, CT, RI, MA, ME
Harry & Lynne Alba
917 Pierce Road
Norristown, PA 19403
(215) 584-6366

Region II:
DE, MD, WV, VA, NC, SC
Trice Ovbey
803 Westmont Drive
Fayetteville, NC 28305
(919) 433-2777

Region III:
TN, GA, FL, AL, MS, LA, AR
Cathy Sipper
P.O. Box 127
Rutledge, AL 36071
(205) 335-6827

Region IV:
KY, OH, IN, IL, MO, MI
Cynnie Bates
133 Elam Park
Lexington, KY 40503
(606) 276-0142

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

Region VI:
TX, OK, NM, CO, KS
Donna Rice
22714 Royal Arms Court
Katy, TX 77449
(713) 347-0345

Region VII:
WA, OR, ID, MT, WY, AK
Shari Gelevich
1344 S.E. Rex
Portland, OR 97202
(503) 236-3203

Region VIII:
CA, NV, UT, AZ
Mark & Claudia MacNaughtan
610 East 800 N.
Genola, UT 84655
(801) 754-3064

However, we also understand that not everyone needs support. If you don't need this program for yourself, you might consider serving others in your region with your positive attitude, your understanding and sympathetic ear, and the strategies you have learned for meeting the challenges of life with ichthyosis.

If you are not already participating in our Regional Support Network, we invite you to join your regional network. The only requirement is your F.I.R.S.T. membership. Call the F.I.R.S.T. national office at 1-800-545-3286 and ask for an RSN Participation Form. Complete the form and return it to the national office. If you prefer to serve by supporting your fellow ichthyosis sufferer, please note this on your Participation Form. Your form will be forwarded to your regional coordinator. And please feel free to call your the Regional Coordinator in your region to discuss the RSN and your participation.

CYNNIE BATES of Lexington, Kentucky, a member of FIRST's Board of Directors, is the founder of the Regional Support Network. She has developed the RSN into the vital, strong organazation it has become this past year. Cynnie is stepping down as the national coordinator of the RSN at the end of this year to devote time to her family—her sons William and Joshua (who has CIE) and her husband John. We owe Cynnie our gratitude for her hours of work and her devotion to FIRST, the RSN, and the community at large. Good luck, Cynnie, and thanks!

Region IV Conference: *A PORTRAIT*

On August 29, 1992, FIRST's RSN Region IV held an enormously successful conference in Indianapolis. Eighty-nine people attended. Congratulations to Mark and Jill Woods who handled the bulk of organizing in Indianapolis, and to Cynnie Bates, Region IV Coordinator. Following is a spread of selected photos taken at the conference.

ABOVE: Dr. Arthur Norins, Caroline Paulson, geneticist & medical student, with RSN & Region IV Coordinator, Cynnie Bates.

LEFT: Conference organizers, Jill and Mark Woods with their children, Matthew, 4, and Leah, 2.

ABOVE: Lunch at the Conference. At center is Irving Goldberg.

RIGHT: Tom Buehler with his son, John, 19 mos. old. Tom raised over $1200 in donations for FIRST which he presented to the Foundation at the Conference.

LEFT: Madonna Taylor (l.) with Wendy Vaughn, age 17, of St. John, Indianna.

RIGHT: Sandy Smollinger, one of several childcare volunteers, with Mark Wood.

LEFT: Beth Engerman, age 11, of Kendallville, Indiana.

RIGHT: Becca and Frank Ost, of Princeton, Indiana.

ABOVE: Part of a lamellar ichthyosis discussion group.

LEFT: Part of a EHK discussion group.

A N N'S P R A Y E R

a short story
by Polly A. Johnson

He came into the store where Ann worked looking for a particular book. She waited on him at the counter. As he paid for the book and she handed him his change, he noticed her hands. He asked her, "May I pray for you?" She was not surprised at his question, since this was a Christian bookstore, but she was taken aback for a moment. But at last she said, "sure."

As they went toward the back room for privacy, he asked her about her hands. She explained to him that her condition was hereditary and that her father and two other sisters had the same condition. She didn't know at the time that her condition was called epidermolytic hyperkeratosis. All Ann knew was that she lived with dry skin on the palms of her hands and the soles of her feet and in all of the folds of her skin.

The man said, "God can break through hereditary barriers." Ann didn't disagree. She believed, as he did, in the power of God. They closed their eyes. Holding hands the man prayed for Ann. They were both fervent—he in his prayer, and she in the hope of a miracle.

The prayer was over. They both looked expectantly at Ann's hands. Nothing had changed. She didn't feel tingling or any special sensations. All was as before. She thanked the man and he left. She never saw him again.

Ann believed that the miracle, which was not apparent on the outside, must have taken place within her body. When she met people she knew well she would share her story of the man who came into the bookstore to pray for her healing. She would tell these friends "... I believe my genes are healed. I don't think my children will have this skin condition."

Some days Ann pitied herself and thought life unfair. However, she recognized all the other blessings that God allowed in her life. One of these blessings turned out to be Arthur. Arthur loved her dearly and accepted every part of her. He told her that her hands were softer than those of anyone he knew. Ann was grateful for this support. Arthur became her husband.

After a year in a wonderful marriage, they decided to have a child. A son arrived, perfectly formed with skin soft and unblemished. He became Arthur's joy and Ann's delight. Ann secretly smiled and said, "Thank you Lord, my genes are truly healed." The son grew into a fine toddler. He needed a brother or sister, so another child was planned and on the way.

It was a warm spring day when Mariah made her way into the world. She arrived two weeks earlier than expected, ready to get a head start in life. In the delivery room the doctor said, "I see the head; push on the next contraction, Ann, and we will have a baby." He spoke correctly. The next contraction and a push expelled a pink and red eight-and-a-half pound ball with a dark fuzzy head.

But the doctor appeared momentarily confused as he held the child. He placed Mariah on the warming table and conferred with his nurses. The two nurses rushed frantically about speaking in hushed tones while furtively looking at Ann. Ann looked at her baby and declared to the doctor, without much feeling, "I know what is wrong with her. She has skin like mine, and there is nothing you can do about it."

One of the nurses wrapped the crying child in a warmed blanket and handed her to Ann. Ann looked at her daughter up close for the first time. Seeing Mariah's skinned face brought tears to her eyes. She unwrapped her child's blanket partway and saw with great disappointment that Mariah's entire body resembled a skinned knee, all red and raw. She held her quietly and waited until she was settled in her room to grieve.

The hospital staff didn't quite know how to care for Mariah. She couldn't be clothed, so blankets were continually replaced and draped loosely around the tender skinned infant. Instead of staying another night at the hospital, Ann had Arthur come and take her and the baby home. She'd feel more comfortable there. She couldn't bear the thought of visitors coming to see Mariah. The thought of being pitied was overwhelming. At this time the stress of the situation released too many fragile feelings. Even a kind word became a cue for tears.

... The doctor appeared momentarily confused ... two nurses rushed frantically about while furtively looking at Ann. Ann looked at her baby and declared, "I know what is wrong with her. She has skin like mine, and there is nothing you can do about it."

It took several months for Mariah's skin to toughen up to where it looked nearly normal. Ann spent long days and sleepless nights comforting her child. Mariah had to be held carefully. Movements that most people take for granted would hurt her and make her cry. But under all of the scabs and skinned patches, Mariah's beauty came through. The fine features of her round cheeks glowed with health.

Ann and Mariah became close. The sharing of the same condition created a bond that could not be severed. Ann relived her infancy as she watched her daughter grow. This heart rending experience would not allow her to lose faith in God. She didn't understand why it all had happened, but she accepted it.

One day, a year after Mariah's birth, a letter came in the mail. It was from the Ichthyosis Foundation, which was a support group for those with epidermolytic hyperkeratosis and other related genetic diseases. In amazement, Ann read that extensive research now in progress might eventually lead to a cure for this genetic disease.

The prayer for Ann that day so long ago had not provided the healing she had sought. The answer to that prayer would come in a future cure. Ann believes in and hopes for that day.

Polly A. Johnson, a school teacher, lives in Morgantown, West Virginia, where she lives with her husband, her daughter Cassie (17 mos.), and son Robby (3 yrs.). Polly and her daughter Cassie have EHK.

N.O.R.D. REPORT

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

Orphan Drug Amendments Update

The Orphan Drug Act Amendments (S.2060) is stalled in the Senate due to heavy pharmaceutical industry lobbying. Senators Metzenbaum & Kassebaum are trying to get more sponsors so they can force a vote on the Bill. Senator Metzenbaum (D-OH) met with presidential candidate Bill Clinton and elicited a promise that should he win the election he will push for pharmaceutical price constraints and will not veto the Orphan Drug Amendments. President Bush vetoed an earlier version of this bill.

On the same note, NORD's Executive Director, Abbey Meyers, appeared on the *Good Morning America* show on July 22 to debate this issue with a representative of the biotechnology industry. Abbey focused on the new cancer drug, Taxol, an example of a blockbuster drug presently sheltered from competition through the Orphan Drug Act. Although Taxol will be therapeutic for a wide range of prevalent types of cancer, the manufacturer (Bristol Myers) nevertheless received orphan drug status for the drug by seeking FDA approval for Taxol's application to only one rare type of cancer. A few days after the show was broadcast, Bristol Myers relinquished its orphan drug designation for Taxol, saying it would not be within the spirit and intent of the ODA to continue the designation.

Drug Prices & Politics

It has been reported that the pharmaceutical industry is frightened of a Clinton Presidency, in part because Senator David Pryor (D-AR) may play a significant role in the Clinton administration. Senator Pryor is the leading congressional advocate for reducing pharmaceutical tax credits and limiting the industry's ability to raise prices beyond inflation. The Democratic platform specifically mentions pharmaceutical inflation several times, promises to reign in drug costs, limit the ability of the drug industry to credit lobbying and marketing expenses to their R & D (research & development) expenses, limit their Puerto Rican tax credits, and impose some kind of pricing constraints.

The Industrial Biotechnology Association (IBA), a biotechnology manufacturers trade group, printed an interview with Governor Clinton and President Bush. Both were asked point blank: *"Do you support federal controls on drug pricing and drug price increases?"*

Bush: "I oppose regulation of drug prices."

Clinton: "A Clinton Administration will support Senator David Pryor's proposal to eliminate special tax breaks now on the books for drug companies that raise their prices faster than Americans' incomes go up."

Medical Bankruptcy on the Increase

A study by the Consumer Bankruptcy Project of 24,000 debtors in the states of California, Illinois, Pennsylvania, Tennessee and Texas showed that 12.4% of bankruptcies filed in 1991 were due to unpaid medical bills. Ten years earlier, in 1981, this figure was 3%. This represents greater than a four-fold (413%) increase over ten years.

Patient Privacy—Fact or Fiction?

Although it is true that people's direct medical records are protected by privacy laws, true medical privacy hardly exists because much of our personal medical history is on the Medical Information Bureau's (MIB) database. Subscribers to the MIB database (mostly insurance companies) can find out what diseases we have and how much money insurance companies have paid out for our care. The MIB is the insurance industry means of enabling them to find out whether you are telling the truth on medical or life insurance applications.

How do they get their information? Past medical insurance claims is one way. Another way is that pharmacists, physicians and health insurance companies are *selling patients' prescription histories* to drug manufacturers, who then use the data for marketing purposes. Rep. Pete Stark (D-CA) has introduced legislation that would make this illegal. His bill is patterned on the law which prohibits your local video store from selling your video tape rental history. As the law now stands, your movie rental history is protected. Sadly, the same protection does not extend to your prescription medication history.

TEACHING TEACHERS

(Continued from Page 1)

on ensuring that this manual is representative of the needs of ***all*** children with ichthyosis. So, please, if you are the parent of a school-age child, contact me to share your hopes and concerns regarding your child's education.

If you are a child or teen with ichthyosis, I'd love to hear your perspective as well. I am also still in need of artwork done by children. If you like to draw or paint, please get out those crayons, markers or paint, and send me some of your artwork. I am particularly interested in self-portraits, pictures of you and your family, and pictures of you doing something that you really like.

I welcome phone inquiries. My home phone number is (212) 427-5904. I have an answering machine, so feel free to leave a message and I'll get back to you. Artwork should be mailed to my home address: 7 East 88th Street, Apt. 4-B, New York, New York 10128-0504.

Thank you in advance for your support. I look forward to the publication of the manual so that I can give back a bit of what you all have shared with me.

Deborah B. Vilas, a newly elected member of the F.I.R.S.T. Board of Directors, lives in New York, New York, where she is completing work on her Master's degree in special education.

NEWS & NOTES

Video About Teenagers Coping with Chronic Illness Available

Duke University Medical Center offers a video focusing on teenagers with chronic illnesses. Interviewed are teens with different illnesses, including cancer, sickle cell disease, asthma and cystic fibrosis. These young people talk frankly about their problems and address issues such as friendship, independence and the possibility of death. Cost is $35.00.

The video is available from Julia Gaskel, Duke University Medical Center, Box 2916, Durham, NC 27710; (919) 684-3401.

Variety of Useful Newsletters Available

Readers of *FOCUS* surely understand the importance of obtaining information from numerous sources on a wide variety of topics. Any number of nonprofit agencies publish newsletters that may contain valuable information. Following is a list of some of these, which you may find useful:

CHOICES, published by the Shriners Hospitals for Crippled Children. Stated goal is "to improve access to comprehensive, coordinated, family-centered, community-based health care through the establishment of linkages between public and private agencies serving children with special health needs." Inquiries to Betty Presler or Ken Massanari, c/o Shriners Hospital, 1900 Richmond Road, Lexington, KY 40502; (606) 266-2101.

CONNECTIONS: The Newsletter of the National Center for Youth with Disabilities. The stated mission of the NCYD is "to improve the health and social functioning of youth with disabilities through providing technical assistance and consultation, disseminating information, and increasing coordination of services between the health care system and others. Center activities are directed at enabling youth to become full participants in their communities." Inquiries to: National Center for Youth with Disabilities (NCYD) Box 721, University of Minnesota Hospital and Clinic, Minneapolis, MN 55455; (800) 333-6293 or (612) 626-2825.

The NASP Newsletter, a publication of the National Association of Sibling Programs "discusses programs in all stages of development which focus on the well-being of brothers and sisters of people with special needs." Inquiries to: Donald Meyer, editor, the Sibling Support Project, Children's Hospital and Medical Center, 4800 Sand Point Way N.E., PO Box C5371, Seattle, WA 98105.

NETWORK NEWS, published by the Center for Child Health and Mental Health Policy, Georgetown University's Child Development Center, focuses on children and youth in high-risk situations. Inquiries to Suzanne Bronheim, Georgetown University Child Development Center, 3800 Reservoir Road N.W., Washington, DC 20007.

We'll periodically list more of these important information sources in future issues of *FOCUS*.

Researcher Asks for Assistance in Study of Neutral Lipid Storage Disease

Also known as Chanarin-Dorfman Syndrome, Neutral Lipid Storage Disease is one of the more uncommon of the ichthyoses. In addition to its dermatological symptoms, people with NLSD may have an enlarged and fatty liver, lipid droplets in their white blood cells, weak muscles, an enlarged heart, and/or mild deafness. **Professor Rosalind Coleman, MD**, of the University of North Carolina, Chapel Hill School of Medicine, writes to tell us that she has been studying this disease "with the goal of determining the underlying genetic defect." She continues: "A major difficulty for my research has been obtaining skin cells from patients and their families. I would be very grateful if you could let your readers know of my interest."

Professor Coleman may be contacted at the following address/phone number: CB# 7400, McGavran-Greenberg Hall, The University of North Carolina School of Medicine, Chapel Hill, NC 27599-7400; (919) 966-7213.

Medical Advisory Board Members Contribute to FIRST Newsletter

In our last issue of *FOCUS*, we acknowleged the generous donations of members of our Medical Advisory Board who helped to underwrite the cost of producing and distributing the revamped Summer issue of the Newsletter. The donations of three members of the Board arrived too late to include in the last issue, and we'd like to thank them here:

Dr. Lawrence Schachner
Miami, Florida

Dr. Amy Paller
Chicago, Illinois

Dr. Robert A. Silverman
Annandale, Virginia

Many thanks to these and all other members of our Medical Advisory Board for their generosity which has helped make the new *FOCUS* such a success.

Neutrogena a FIRST Corporate Sponsor

Also last issue we overlooked listing the **Neutrogena Corporation** as a Foundation Corporate Sponsor in our tabulation of contributors. Neutrogena is a long-time supporter of FIRST and we regret having failed to acknowledge their generous contribution. Apologies first, then thanks.

Any information you'd like to see in the News & Notes section of the newsletter, please send it along to us in Raleigh.

IN MEMORIUM

Nollaig Casey
November 19, 1991 — July 19, 1992

The entire ichthyosis community with FIRST join hands and bow heads in silent prayer for Nollaig Casey, who succumbed to Netherton's Disease after a seven-month struggle for life. The beloved daughter of Brian and Jacinta Casey of Enniscrone, County Sligo, Ireland, little Nollaig battled pervasive infections and struggled vainly to gain weight throughout her seven months. Cathy Sipper, FIRST's Region III Coordinator, whose own son Zebulin has Netherton's, remained in close contact with the Casey family throughout their ordeal.

Brian Casey wrote us a moving letter thanking everyone in FIRST for their help and their thoughts. He tells us: "We will be thinking of you all in FIRST and we are sure that Nollaig will also be watching over all of you."

Three Research Teams Locate EHK Gene

Researchers Foresee Development of Prenatal Testing & Treatments for Sufferers of Epidermolytic Hyperkeratosis

by Nick Gattuccio

We reported in our last issue of *FOCUS* that researchers at the National Institutes of Health/NIAMS (Dr. Sherri Bale and colleagues) had narrowed the search for the gene which causes EHK to a narrow region on chromasome 12. In the short span of time since then, three separate research teams have identified the precise location of the genetic defect and explained its function in this blistering and scaling disorder. The remarkable speed with which breakthroughs are occurring on this front underscores the tremendous efforts of the researchers dedicated to this work.

Dr. Joseph Rothnagel and Dr. Dennis Roop, both of the Baylor College of Medicine in Houston, Texas, and their colleagues, reported their findings in the August 21, 1992, issue of *Science* (vol. 257). Virtually the same week, in the September 4th issue of the journal *Cell*, two teams of reseachers (one led by Dr. Elaine Fuchs of the Howard Hughes Medical Institute at the University of Chicago, and the second led by Dr. Peter Steinert of the Laboratory of Skin Biology at NIAMS) reported similar findings.

The research findings conclude that in fact two genes cause the blistering symptoms of EHK. Both of the culprit genes produce defective keratin proteins in the skin's epidermis. These critical keratin proteins are essential for creating structural integrity in our skin cells. They help build the cell's cytoskeleton. Without this cellular "skeleton," skin cells near the base level of the epidermis are very easily destroyed, and their destruction leads to severe blistering with even mild rubbing or abrasion.

In their respective studies, the three research teams located genetic defects on one of two genes in all individuals in their studies who had EHK. These defects were located on the genes that code for the keratin proteins K1 and K10. The gene for the former resides on chromasome 12, that for the latter on chromasome 17. Because keratin proteins always work in pairs (and since the K1 and K10 proteins are a natural pairing), defects in either one of the genes which encode for the respective keratin proteins create defects which result in the blistering symptoms of EHK. All three research teams note the similarity between the genetic defect which causes EHK, and that which causes a similar disorder, epidermolysis bullosa simplex (EBS). In the case of EBS (see *FOCUS*, Spring 1992), the same cellular cytoskeleton error occurs, but in that case the error is in the pair of genes which encode for the pair of keratin proteins K5 and K14.

Perhaps the most significant short-term outcome of this discovery of the genes responsible for EHK will be the "improvement of pre-natal screening for couples seeking genetic counseling," according to Drs. Rothnagel and Roop. In their view, "identification of specific mutations within affected family members will now allow accurate diagnosis at 10-11 weeks gestation." Dr. Fuchs agrees: "the development of genetic probes for prenatal testing would be a significant improvement over current methods."

As for long-term benefits of the discovery, Rothnagel and Roop suggest that it "paves the way for better and more systematic molecular approaches for the treatment and amelioration of EHK and related keratin disorders of the skin."

However, the researchers caution that their work is far from finished, and note that a possibility exists that some other protein in addition to the keratins K1 and K10 may cause symptoms of EHK. The long-term goal is to continue studying genetic material from large numbers of EHK patients in order to end up with a "catalog" of specific mutations that cause EHK. Says Dr. Fuchs, "A deeper understanding of the location of mutations in EHK genes should help us not only in developing improved methods for diagnosis, but also in exploring whether it may be possible to treat this disease by gene therapy." Dr. Steinert agrees, adding that "once we have a catalog of mutations, we can design treatments that are directed toward correcting the molecular defects rather than treating the symptoms."

We congratulate the stunning efforts of all the researchers involved and look forward to further advances.

Meade Piercey Wows 'em At Seaway Festival!

FIRST members Debby and Parker Piercey of Ogdensburg, New York, sent us a press report announcing their daughter Meade's stunning victory of sorts in their area's annual Seaway Festival Lip-Synch competition. Meade, a six year old with CIE, and her cousin, Morgan Fletchall, brought down the house with a "sizzling rendition" of Roy Orbison's "Pretty Woman." Meade took the part of Roy Orbison, while her cousin Morgan played the part of the pretty woman. Meade's mother writes to say, "May this serve to remind others like Meade to go out and show their talent. All children are special, but especially those with ichthyosis."

Meade, who just started school this year, enjoys tap dancing and swimming. As with many children with ichthyosis, beginning school, entering a new environment, posed a challenge. Meade and her mother want us to know about a book that helped her meet the challenge. It is entitled, *Pobody's Nerfect* (ISBN # 15683-2), published by Current, Inc., of Colorado Springs, Colorado. Meade used this book as the basis for a presentation she made to her class on the first day of school to introduce them to her ichthyosis, and to emphasize that, indeed, nobody's perfect. But that would have been tough to explain to the judges, who awarded Meade and her cousin a perfect score, for a Seaway Festival record 180 points. Congratulations to Meade and Morgan, and to all of our children who meet the challenge of ichthyosis and win!

Meade Piercey (l) as Roy Orbison, with "pretty woman" Morgan Fletchall.

F.I.R.S.T.
P.O. BOX 20921
Raleigh, N.C. 27619-0921

Non-Profit Org.
U.S. Postage PAID
Raleigh, NC
Permit No. 2209

FOR CHRISTMAS, 1992 . . .
A F.I.R.S.T. GIFT OF LIFE

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

FIRST offers you an opportunity to give a gift of life this Christmas. Perhaps you've considered giving your neice or father-in-law or your boss a toaster or dust buster, or maybe just a gift certificate to an Italian restaurant. Or maybe they're one of those relatives who is just impossible to shop for. Consider this . . . Consider giving them, instead, a FIRST Gift of Life Certificate. Offer those on your Christmas list a FIRST Gift of Life Certificate — a $50.00 donation in their name to FIRST. For this, they will receive a personally inscribed Gift of Life Certificate and a subscription to FIRST's newsletter, FOCUS.

(Detach here and send with your Gift of Life Purchase -- Please Print Clearly)

GIFT OF LIFE (1)
FOR: _____
Address _____
City/State/Zip _____

GIFT OF LIFE (2)
FOR: _____
Address _____
City/State/Zip _____

GIFT OF LIFE (3)
FOR: _____
Address _____
City/State/Zip _____

Enclosed is my payment for ____ FIRST Gift of Life Certificates (@ $50.00 each) = $_____

Be sure to list the name(s) of your recipients in the way you wish it to appear on the Gift of Life Certificate.
Use additional page if necessary.

Return this form with your check made out to FIRST by
December 10, 1992
to ensure delivery by Christmas

FIRST will provide you with a receipt for your tax-deductible donation.

Mail to:
FIRST/Gift of Life
P.O. Box 20921
Raleigh, N.C. 27619-0921

Your Name _____
Address _____
City/State/Zip _____
PHONE: ( ) _____