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

ICHTHYOSIS FOCUS

Vol. 12, No. 1 A Quarterly Newsletter for Friends of FIRST Winter 1993

F.I.R.S.T.'S 1993 NATIONAL CONFERENCE SET

Planning Underway For F.I.R.S.T.'S Largest & Most Ambitious National Conference Ever: Chicago, June 25 — 26

Planning is well underway for FIRST's 1993 National Conference in Chicago. The Conference site is the Chicago Marriott at Schaumberg, in Schaumberg, Illinois, a Chicago suburb.

The Conference is still five months away, and a great many details have yet to be finalized, but many of the broad outlines of the Conference are in place. Indeed, the 1993 National Conference is shaping up to be the largest, most innovative and ambitious Conference in FIRST's 12-year history.

The configuration of the meeting space allows us unprecedented flexibility in planning. In addition to the main ballroom, we'll be using four large anterooms, so we can conduct several small seminars and focus group sessions at once. We'll also have childcare facilities—one for the very young, and a room for older children. The Friday and Saturday night social periods will be in a huge outdoor circus tent.

Here are a few highlights of the Conference as we forsee them now:

- **Focus/Discussion Groups**: We'll have several of these, including a special Mother's group session, a Father's group, groups for ichthyosis types, etc.
- **Teen Group**: An opportunity for our young adults to get together to discuss their own special topics. We also look forward to some teens talking to groups of pre-teens about their ex-

(Continues on Page 5)

NUTRITION & ICHTHYOSIS: NEW FINDINGS

by Nick Gattuccio

Researchers In San Diego Document Link Between Protein Nutrition and Physical Development in a Harliquin Ichthyosis Patient

For nearly two years we have attempted to search out documentation of a link between nutrition and growth and development in newborns with ichthyosis, and for two years we have drawn blanks. Not infrequently physicians would address the issue informally, off-the-record, offering hypotheses about hyperproliferative skin and resulting demands on the young body's protein reserves.

And there is no shortage of anecdotal accounts on the part of parents of newborns with ichthyosis about the perceived relationship between nutrition and their babies' growth (or failure to thrive). But until now their has been no systematic study of the issue—no documentation, no measurements, nothing in print.

In a recent issue of the medical journal *Dysmorphology and Clinical Genetics*, (see citation at end of article), five San Diego reseachers and physicians document the influence of nutrition on the growth and development of a young boy with harlequin ichthyosis. The researchers followed the boy's progress from birth to age five years, eight months. As summarized in the article's abstract, "At 15 months, failure to thrive or develop led to nutritional investigation, which indicated that inhibition of growth was a consequence of protein malnutrition that was a consequence of enormous losses of protein in desquamated skin."

It was at 15 months that the boy's failure to thrive became critical. His weight was under five pounds, and all other developmental measurements were consistent with an infant just a few months old. "His developmental age was assessed as 2 months or less at chronologic age 15 months." However,

(Continues on Page 6)

IN THIS ISSUE . . .

Ichthyosis
Focus
Vol. 12, No. 1
Winter 1993

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

***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

Pen Pals, Pen Pals, Pen Pals . . .

Janice Swan, a sixteen-year-old friend of ours in Tatamagouche, Nova Scotia, wants very much to exchange letters with other teens with ichthyosis. She tells us, quite correctly, that "I am sure there are many others who have never had any contact with anyone who has the same skin disorder and would be just as interested as I am in forming a penpal relationship." Janice can be reached at **R.R. #2, Tatamagouche, Nova Scotia, Canada B0K 1V0.**

Janice may wish to write to **Pumehana Poai**, a fourteen-year-old with CIE who also wishes to exchange letters with a penpal. Pumehana lives in Hilo, Hawaii. It is difficult to imagine two more different places to be a teenager than Hawaii and Nova Scotia. Anyone out there wanting to share letters with Pumehana can write to: **62 W. Puainako Street, Hilo, Hawaii 96720.**

Walter Ciszek of Rancho Murieta, California, wants to share a care regiment that is working successfully for him. He mixes 1 part glycerine (which he purchased with a prescription from his physician) to 6 parts of Nature's Family Aloe Vera Vitamin E Moisturizing lotion and applies the compound to his entire body at bedtime. After his first experiment with the compound, he found, "to my surpirse the next day I saw a marked improvement ... and approximately three months after I first applied the lotion, my skin shows no sign of scaliness." Mr. Ciszek tells us that "It took me 75 years to get relief from this malady."

From Charlestown, New Hampshire, **Jackie Treat** reports that her three-year-old daughter, who has CIE, showed marked results with the application of NuSkin moisturizer. "In just a few days we noticed a significant reduction of overall redness and reduced flaking." Adds Jackie, "We also appreciate the reduction in unsolicited public comments. The convincing moment came when we ran out of the lotion and tried something else with poor results." Anyone wishing to speak to Jackie about her treatment regimen may call her at **(603) 826-4021.**

Heather Gattuccio has been collecting lists of useful books for young people with ichthyosis and wishes to share some here. Two are skin books for young adults: *Skin Disorders*, by Lynne Lamberg (New York: Chelsea House, 1990); and *Rheumatic & Skin Disease*, by Wrynn Smith, PhD (New York: Facts on File, 1988). For young people aged 4-10, Magination Press in New York publishes a book entitled *Double-Dip Feelings: A Book to Help Children Understand Emotions*, by Barbara Cain (1990). Another health book for the young is *How & Why: A Kid's Book About the Body*, by Catherine O'Neill, illustrations by Loel Barr (New York: Consumer Reports Books, 1988).

From Laurinburg, North Carolina, we heard recently from **Barbara Poole**, whose daughter **Whitney** was born last November with a rare condition called epidermal nevus syndrome. This disorder falls within the scope of related skin types, is extremely rare, and Barbara would like very much to hear from anyone who may be familiar with this disorder. She can be reached at **12280 Evergreen Lane, Laurinburg, NC 28352 (919) 277-7923.**

From Belgium, **Luc De Batselier** and **Kristin Biesemans** write to tell us they'd like to correspond with parents of children with lamellar ichthyosis. Their child, **Seppe**, was born with lamellar ichthyosis, and the shortage of resources in Belgium make it all the more imperative that they communicate by mail. You can write to them at the following:

Bookmolenstraat 187
9200 Dendermonde
Belgium
Phone: 052 / 477-147

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

Ichthyosis Focus is provided as a service to members of F.I.R.S.T. as a medium for the free exchange of information. Neither F.I.R.S.T., its Board of Directors, its Medical Advisory Board, nor the *Focus* Editor endorse any treatments or products reported on in *Ichthyosis Focus.*
Views and opinions expressed in this publication do not necessarily reflect the views of F.I.R.S.T. or Foundation officials.

FROM THE E.D.'s DESK

by Nick Gattuccio

I have just passed the three month mark behind the ED's desk here in Raleigh, and the transition has been as hectic as it's been interesting. Indeed, a great deal is afoot.

The first week in December took us out to San Francisco, where Board President Frances McHugh and I represented FIRST at the annual American Academy of Dermatology (AAD) convention, where, among a great many other things, we convened our annual meeting of FIRST's Medical Advisory Board. The meeting was attended by officials from NIH and NIAMS (National Institute of Arthritis, Musculoskeletal & Skin Disease), and the discussion focused largely on prospects for developing the ichthyosis patient registry.

As you recall, last year the foundation successfully spearheaded an effort to include language in NIH appropriations legislation highlighting the importance of such a registry to both the research and patient communities. NIH/NIAMS was directed by Congress to study the issue and report back its assessment at budget hearing this spring. Drs. Alan Moshell and Steven Hausman of NIH took the opportunity to discuss the outlines of their outlook on a registry, and had the opportunity to hear the opinions of ichthyosis researchers on our Board.

Frances and I also attended the winter meeting of the Coalition of Patient Advocates for Skin Disease Research, of which FIRST is a member. The session was attended by the AAD's new president-elect, Dr. Peyton Weary, which was an important statement of the Academy's support of groups like FIRST. Dr. Weary is an ardent supporter of the Coalition. The meeting was also attended by Dr. John Gruppenhoff, the AAD's political point man. The AAD is enormously supportive of nonprofit advocacy and support groups like FIRST. The AAD is a strong backer of the Coalition and has allocated considerable resources to our organizations and agendas. The Coalition meeting was devoted largely to preplanning the Coalition's 1993 legislative agenda and coordinating our Washington meeting and legislative visits in March.

The entire week of the San Francisco AAD convention was extremely interesting, particularly to someone as new in the driver's seat as I am. The entirety of the Moscone Convention Center was covered by an enormous interwoven blanket made up of threads of distinct, yet complementary, interests: medical practicioners, biomedical reseachers, agency administrators, pharmaceutical manufacturers, political mavins, and nonprofits. It was a fascinating blanket and an excellent education.

Turning from old to new business, the big plate, of course, is our upcoming National Conference in Chicago this June. The event is covered extensively in this issue, so all I wish to add is that I'm striving to make this the most interesting, innovative, and productive conference ever. I encourage everyone to give serious thought to coming out for it. We're trying to offer something for nearly everyone, and we're particularly interested in our children and young adults. Incidentally, I've had a call from a woman in Tasmania, Australia, who'll be coming to Chicago with her daughter.

With the inaguration of the new democratic administration in Washington, the buzzword of the season is "change," and we're not immune to it here, either. I'm pleased, then, to welcome on behalf of the foundation membership four new members of our board of directors: Pam Stockton of Raleigh, Betsy Bates-Freed of Los Angeles, Ginna Frantz of New York, and Virginia LaBrant of Redington Beach, Florida. All four are accomplished professionals who will bring to the board a welcome bredth of experience and expertise.

And none too soon. Over the past two years FIRST has more than doubled its membership rolls, tripled its budget and revenues, and has embarked on member services programs with increasingly ambitious stripes. At the same time, signifcant research findings are emerging from labs around the country and the shape of American health care will certainly change drastically in the coming years. We are indeed in the midst of profound changes as an advocacy and support organization, and we will face some tough decisions as we consolidate those changes and plan for the coming decade. The prospects are of great opportunities, not uncertainties, and of an optimistic, not cloudy, future.

NEWS FROM NIAMS

The National Institue of Arthritis & Musculoskeletal & Skin Diseases of the National Institutes of Health

by Sherri J. Bale, Ph.D.

Research Team Studies C.I.E. and Lamellar Ichthyosis in Families in Egypt

On November 15th, Dr. John Compton, Dr. John DiGiovanna, and I boarded a plane at Washington National Airport in the nation's capitol. Twenty-four hours later we arrived in Cairo, Egypt, where we spent the next 10 days working with an Egyptian geneticist in her clinic at a major Cairo hospital seeing patients with a variety of ichthyoses.

Our special interest in Egypt developed because marriage between cousins is a common occurrence in that society, and therefore there is a relatively high incidence of recessive disorders. Since lamellar ichthyosis (LI) and congenital ichthyosiform erythroderma (CIE), both recessive ichthyoses, are research interests of ours, it seemed likely that the patient population in Egypt would be very beneficial for our studies of the genetic basis of these disorders.

Our collaborator reviewed her files on patients seen in the clinic over the last two decades, and contacted many families to see if they would be interested in participating in our research. As a result, we met more than a dozen patients with ichthyosis and their families. Many of these people are willing to have blood drawn and undergo skin biopsies, because of their interest in contributing to the knowledge of the genetic basis of ichthyosis.

Although Egypt is a beautiful country, full of history and excitement, it is also a poor country. Many of the ichthyosis patients we saw had severe skin disease, and yet had never had the benefit of treatment. Even common, over-the-counter creams and lotions which provide such relief for dryness and itching are often unavailable, especially outside of the major city.

We plan to retun to Cairo in the Spring or early Summer, at which time we hope to have all the details in place to be able to carry out our research plans. We will continue to update the members of F.I.R.S.T. on our progress.

¶ ¶ ¶

Skin Biology Lab Requesting the Assistance of Patient Families for Lamellar Ichthyosis and CIE Study

Dr. Bale and her colleagues at the skin biology lab, NIAMS, are requesting the assistance of patient families in the U.S. in their study of lamellar ichthyosis and CIE.

The study will involve patient families in with two or more affected siblings. Both natural parents will need to take part.

If your family meets these criteria, and if you are intersted in participating, you may reach Dr. Bale at NIAMS, at **301-402-2679**.

F.I.R.S.T. ELECTS FOUR NEW MEMBERS TO ITS BOARD OF DIRECTORS

At it's final 1992 board meeting in December, FIRST's Board of Directors elected to three-year terms four new members. All are professional women who bring a wealth of new expertise and experience to our board of directors at a time when the foundation faces difficult decisions about FIRST's policies and goals as we grow and expand toward the 21st century.

Virginia (Ginna) Frantz: Owner of her own V. M. Frantz & Company, a New York public relations and marketing firm, Ginna Frantz brings to our board a broad range of skills. A graduate of Georgetown University's School of Foreign Service, Ginna brings seventeen years' experience in public relations and marketing, including media relations, television production, events management, publication production, and much more.

Elizabeth (Betsy) Freed: Betsy has a long-standing record of service on health-related boards and community service programs. A media professional and specialist in science writing and public relations, she has worked for the USC Health Sciences Public Information Office as writer and publications director; as a reporter for the *Los Angeles Times*, and as Director of News Planning for a L.A. television station.

Pamela Stockton: With extensive experience in finance, accounting, and business, Pam brings much appreciated business savvy to the board at a time when its role as policy making body for the Foundation will surely be tested. A Raleigh resident, Pam will be able to assist with local foundation development, as charitable giving is among her financial specialties.

Virginia S. LaBrant: A resident of Redington Beach, Florida, Virginia adds to our foundation considerable experience with nonprofit boards and health-related community service. As a former vice-president of corporate communications for a large financial institution, and presently a consultant in the same area, Virginia adds valuable expertise in board organization and foundation strategic planning.

We welcome our new members of the board, and we look forward to enjoying the fruits of the enormous wealth of professional experience they bring to FIRST. They will add an important new slant to the policy making process within the foundation, and we all look forward to their influence on the few short years between now and the next century.

National Conference Set for Chicago

(continues from page 1)

periences with ichthyosis. In short, we hope to give our young adults an opportunity to teach as well as learn.

- **Mini Seminars**: Recognizing that not all members at the Conference will be interested in all available topics, we'll cover many subjects in small open seminars. Among topics under consideration for these mini seminars are the genetics of ichthyosis; the ABCs of retinoid therapy; current advances in ichthyosis research; ichthyosis and nutrition; and self-image and ichthyosis. Others are under consideration.
- **Children's Art Show**: Among other activities for the children during the Conference, they'll be creating art for their own and for our enjoyment and pleasure. Additonally, we'll be bringing art work collected by one of our board members to illustrate her book about ichthyosis, and that will be included in the art show too.
- **General Session**: We'll be inviting a distinguished guest speaker to address our assembled group at a general session on Saturday afternoon.
- **Social Gatherings**: On Friday evening we'll be having an informal registration session and social hour (or two) offering everyone the opportunity to meet old acquaintances from the Williamsburg or Orlando Conferences, or meet new ones.
- **Technical Exhibitors**: For the first time ever, FIRST is inviting our acquaintances in the pharmaceutical community to participate in the Conference. Representatives from several of the companies whose products are no doubt familiar to you will be available to answer questions, offer samples, and present you with an opportunity to offer input on the product's use and effectiveness.

We're striving to make this conference both interesting and informative for the broadest possible array of people. And we're putting special emphasis on issues relating to our children and young adults. Conference attendees will have a broad array of choices at hand and will be able to tailor the conference to their own interests and needs. I look forward to seeing all of you there.

Conference registration forms will be available in April. The next issue of *FOCUS*, due out in early May, will have a full rundown on the Conference itinerary.

SPECIAL AIR TRAVEL ARRANGEMENTS FOR F.I.R.S.T. MEMBERS

Travel Agents International here in Raleigh, the firm which negotiated our arrangement with the Marriott in Chicago, is offering to help FIRST members who will be flying to Chicago obtain the lowest possible fare.

Everyone booking flights through this firm will have the opportunity to rebook at any time up to the travel date in order to take advantage of any drops in ticket prices.

Additionally, the travel agent will be donating back to FIRST 2% of all fares booked to help underwrite the cost of the National Conference.

CONTACT:
Linda Puckett
Travel Agents International
919-846-8221
800-727-6338

PLANNING FOR THE CHICAGO CONFERENCE

When: June 25 - June 26, 1993

Where: Chicago Marriott at Schaumberg
50 North Martingale Road
Schaumberg, Illinois 60173

Hotel Reservations: (708) 240-0100

Hotel Note: We've negotiated a special rate for individuals & families attending the conference—**$68.00 per night**, unrestricted occupancy. This means a family of two, three, or four can stay for the same flat rate. Be sure when you make your reservation that you identify yourself as a FIRST National Conference attendee.

ICHTHYOSIS SUPPORT GROUP STARTS UP IN AUSTRALIA

Annette and Paul Cahill of Abbotsford, New South Wales, Australia, have started a support group for ichthyosis. In the eight years since his birth, their young son Ben Cahill, who suffers from lamellar ichthyosis, has yet to met another person with his condition. But his parents are hopeful this will change soon. Since beginning their ichthyosis support group, their local newspaper wrote a length article about Ben, his condition, the isolation, and their hopes for the success of their group, and they've heard from a great many people. They're attempting to raise money for a computer (they presently handle all of their correspondence by hand), and look forward to periodic meetings.

We encourage all of our Australian readers (physicians as well as patients) to contact Annette and Paul Cahill at:

1 Florey Place
Abbotsford, New South Wales
Australia.

F.I.R.S.T. PERSON

Harlequin Ichthyosis: Letters from a Friend

On May 18, 1992, Lauren Nicole (Nikki) Purcell was born to Dave and Ann Purcell. Nikki was born with Harlequin Ichthyosis. Over the past months we have communicated with Dave Purcell on several occasions, and we've received from him two very moving letters. Dave has granted me permission to edit and reprint here excerpts from his letters to us. Nikki is under the care of Dr. Moise Levy at Texas Chilren's Hospital in Houston.

You probably understand why it has taken us so long to get around to answering your letter. Thanks so much to you and the Board for your card expressing congratulations and kind thoughts.

Since Nikki was a Harlequin baby, she spent her first two months in the hospital. We thought she was probably going to die. But Nikki had other ideas!

She was six weeks premature, spent three weeks in intensive care at Texas Children's Hospital, and then six weeks at Katy Community Hospital. Her progress during her hospital stay was consistent with other survivors in the literature, with the exception that she never contracted any serious infections. She began taking oral etretinate in her second week, but had already begun to shed the thick yellow skin herself. Despite pronounced eclabion, Nikki had a very strong suck reflex from birth, and by three weeks she took all of her feedings by bottle. We stopped the etretinate at two months.

Nikki was able to make progressively longer visits home during her stay at Katy, and came home permanently at age two months. By this time she had shed all of the thick yellow skin associated with the Harlequin Syndrome (as I prefer to call it).

Nikki has been home for about six months, and she is a vigorous but well-behaved baby. Her skin has progressed to severe non-bullous CIE. We keep her lubricated with Eucerine, and we are experimenting with some of the alpha-hydroxy acids. We recently began using a Retin-A cream for her hands, where the skin gets very think.

But Harlequin has left its scars. Nikki has no eyelids, and although she has a blink reflex and can scrunch her eyes closed, she sleeps with them open. We lubricate her eyes constantly. We are consulting various opthamology professionals about surgery to create eyelids for her, and would be very interested in hearing from anyone who has experience with this surgery. Her ears are almost normally formed, but are fused to her scalp.

Both of her hands were deformed by the thick Harlequin skin. She lost the ends of her fingers on her right hand, and what remains of her fingers is encased in a single mass of tissue. Her left hand has similar but lesser damage. Both thumbs are intact, although slightly deformed. All muscles and nerves seem intact, and we are hopeful that in a few years we can begin to correct some of her hand problems.

The message I want to convey is that we as a family have survived that terrible period of Nikki's first two months when the was in the hospital, prognosed to die and suffering some pain, and the following months of having her home with extra care demands. She is thriving, cheerful, and well loved and protected by her two older brothers, Devon (4) and Matthew (2).

The next major medical decision is what to do about her eyes, and when. We would be thankful to hear from patients and parents about the successes they have had with surgery to correct ectropion.

Anyone wishing to contact Dave Purcell may write him at 12023 Nova Drive, Houston, TX 77077. We ask everyone to respect Dave's request that he NOT be called at home.

ICHTHYOSIS & NUTRITION

(Continues from Page 1)

the boy's nutritional history indicated that he was taking in sufficient calories to thrive (227 calories/kg/day, and 4.2 grams per kg/day of protein). "It was evident that it was not physically possible for him to eat any more than he already was."

On the basis of nitrogen balance and other biochemical studies, the researchers concluded that the problem lay in the enormous "turnover of protein to make keratin." It was decided to elevate the boy's protein intake—in fact, to double it to 8 grams per kg per day with special protein supplements to his formula.

The response was dramatic. He gained 690 grams (just over 1½ pounds) in 12 days. "His growth response was an essentially vertical acceleration of weight. Growth in height followed in a less dramatic but predictable fashion. His head grew quite rapidly. He continued to grow at this rate following discharge. Dramatic results occurred both physically and developmentally."

By 24 months, he could sit independently and support himself standing; "... developmental age was estimated at 1 year. At this point the daily dietary goals were 200 calories per kg, and 10 grams/kg protein."

Conclusions for the reseachers were clear and unambiguous:

> "Expereience ... indicated that the enormous loss of desquamated skin produces a requirement for protein and calories that may be impossible for even a very hungry infant to meet.... Recognition that the loss was virtually pure protein led to a later regimen sufficiently high in protein to promote growth and development.... The conclusion that protein had been limiting was drawn from our experience on two occasions when change in protein intake without alteration in total calorie intake led to enhanced growth. The amounts of protein required were enormous, amounting to 8 to 10 g/kg, and similar to those seen in children with severe burn injury .... Once protein needs were met, adequate growth and development followed."

It is very important to keep in mind that these findings are derived from a very extreme case of ichthyosis, and from only *a single case study*. One should resist the temptation to generalize. However, this study does clearly document a relationship between hyperproliferative ichthyotic skin and protein deficiency. If you are concerned about this issue, you should speak to your pediatrician or dermatologist.

Citation:

Brian Saunders, Charles Freedman, William L. Nyhan, Marylynne Rice-Asaro, and Frank Mannino. "Influence of Nutrition on Growth and Development of a Long-Surviving Harlequin Fetus," *Dysmorphology and Clinical Genetics*, vol. 6, no. 1, pp. 2-8 (1992).

KID Syndrome:

A Review of Recent Literature

by Dr. Roni Wechsler, M.D.,
University of Maryland School of Medicine

A number of syndromes exist in which ichthyosis is part of the clinical presentation. One of these syndromes is termed the "KID Syndrome" (for **k**eratitis, **i**chthyosis, and **d**eafness). While this syndrome is not common, a review of the medical literature identified five papers written on this subject in 1990. The following article briefly summarizes this most recent information.

The KID Syndrome is an uncommon ichthyosis-like skin disorder which consists of keratitis (inflammation of structures within the eye), ichthyosis, and neurosensory deafness. Ichthyosis, by the classical definition, implies universal skin involvement. As the KID Syndrome involves the skin in a patchy manner, it is not categorized as a true ichthyosis, but rather an ichthyosis-like disorder.

This disorder was first recognized in the literature as early as 1915, but was not fully described until 1981, when Skinner coined the term "KID Syndrome." Overall, fewer than 40 cases have been reported. Originally the KID Syndrome was thought to be acquired, not hereditary. However, several cases have been reported recently where a mother-daughter, a father-daughter, and a father-two sons (in three separate families) were all affected, so some mode of genetic transmission is likely. The type of inheritance is still not certain.

All patients with KID Syndrome have skin, eye, and ear abnormalities. The skin is usually abnormal at birth, and has been described as red, dry and leathery. Scaling is not a feature initially. During the first year of life, verrucous (warty), ichthyotic plaques develop symmetrically on the face and extremities, which give children a prematurely aged appearance. Thickened plaques are usually found on the elbows, knees and backs of the hands and feet, and a fine scale with horny spikes occurs over the rest of the body. Thickening of the skin on the palms, soles, and palmar pits also occur. In addition, many of these patients have thin, unmanageable hair, sparse or absent eyebrows, eyelashes and body hair, and abnormally thickened finger- and toenails.

The eye disease usually consists of chronic inflammation of the cornea, and neovascularization of the cornea (i.e., a growth of abnormal blood vessels over the front of the eye) which results in progressive visual impairment, often leading to blindness. The eye changes usually begin at birth or in childhood, but can occur later in life.

Ear abnormalities are usually present at birth; however, they are often not detected until later in life when a delay in speech development is noticed. The defect is almost always a bilateral neurosensory deafness (i.e., defective auditory nerves in both ears).

Other abnormalities have been noted in some, but not all, patients with KID Syndrome. These include increased susceptibility to infection (particularly of the skin, ears, eyes, and mouth), the development of multiple skin cancers (especially squamous cell carcinomas), dental defects, defects in sweatiing, a shortening of the achilles tendon, and underdevelopment of the cerebellum in two cases. Intellectual development is almost always normal. Growth delay does occasionally occur.

Treatment of the skin component of KID Syndrome has proved to be difficult. Current forms of therapy include treatments used for other ichthyoses, such as topical keratolytics and emollients (which decrease skin thickness and scale and increase moistrue content of the skin), and oral retinoids (synthetic vitamin A derivatives), such as Accutane and Tegison. Overall, mild to moderate improvement of the skin abnormalities have been reported with retinoid therapy. In many patients, however, the degree of improvement was not significant enough to warrant the side effects observed with long-term retinoid therapy. One patient treated with Accutane did show significant skin improvement; however, the medication had to be stopped due to worsening of her eye disease. Ultraviolet light therapy was tried in at least one patient; however, its use was limited by the occurrence of photosensitivity. Vitamin D preparations (both oral and topical) have also been used experimentally on patients with ichthyosis generally without success.

Dr. Roni Wechsler, M.D., is a resident in dermatology at the University of Maryland, working under the direction of Dr. Gary L. Peck of FIRST's Medical Advisory Board.

F.I.R.S.T. SET TO UPGRADE COMPUTERS

by Chip Dram

F.I.R.S.T. Seeks Help of Members In Effort To Bring the National Office Into The '90s

A move is presently underway to upgrade the F.I.R.S.T. national office's computing capabilities. While the size of our active membership has more than doubled, our budget nearly tripled, and our range of member services has expanded enormously, our computing capabilities have remained in the early dark ages.

"It's both a hardware problem and a software problem," says Exectutive Director Nick Gattuccio. "The learning curve is steep, the equipment unreliable, and the software largely outdated—particularly our critical database software."

The office presently runs with an ancient, generic 286-based DOS machine. "The beast is *so* generic that it doesn't even have a name on it," says Gattuccio. "I can't even tell you what brand of computer it is!"

The software problem is even greater. The Foundation's member database is committed to a relational database program called *dBase III*. This is complicated software requiring sophisticated programming skills. F.I.R.S.T. paid out nearly $2,000 in 1992 for a consultant to help restructure and program this complex software. Previous attempts by Foundation employees to struggle with it in the dark resulted in a database structure that was so entangled it became virtually worthless. "So now we're paying a consultant to help undo all of the well-intentioned errors of the past years," says Nick.

But there is a solution to this entire problem—a long-

(See "Computer WISH LIST," page 9)

Q. & A.

by Melodie Buxman, M.D.

Q. I recently gave birth to a baby diagnosed as a "Harlequin Baby" and was told that this was fatal and the baby would die within a few days or weeks. The dermatologist was consulted and said several Harlequin babies have in fact survived well into childhood recently and that treatment with new drugs has improved their outlook. What is true?

A. Prior to the late 1970s Harlequin Ichthyosis, the most severe of the ichthyoses, was always fatal. The infants succumbed to infection, or severe restriction of eating and breathing due to the excessively tight skin around the mouth and chest. However, with the new well-equipped neonatal units in large hospitals, and with new antibiotics to combat infection, we have been able to save most of these babies. One is 9 or 10 years old and doing well. Several have been treated with the new vitamin A drug Etretinate, with dramatic results. Two or three of these Harlequin babies have survived for a number of months and have had sudden unexplained crib death. We are collecting data on these and hope to be able to discover why. Another possibility is that of misdiagnosis. Since many types of ichthyosis are very rare, infants who actually have one of the other forms of recessive ichthyosis may have tight skin when born (the so-called collodion baby). This goes on to peel nicely in a few weeks and the true nature of the ichthyosis becomes apparent. These infants are sometimes improperly called "Harlequin" babies.

Q. I have ichthyosis vulgaris and would like to know if either of my two children (neither of whom has ichthyosis) could be carriers of the lamellar or other more serious types of ichthyosis.

A. Since we believe that each form of inherited ichthyosis is caused by a single, separate factor, your children would have no greater chance of carrying factors for the other types of ichthyosis than anyone else in the general population. Although ichthyosis vulgaris is a "dominantly" inherited trait, its expression is highly variable and many who have the factor do not clinically express it, or express it only under certain conditions (cold, dry weather, for example).

Melodie M. Buxman, M.D., a dermatologist on the FIRST's 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. (anonymously, if you wish) at FOCUS: P.O. Box 20921, Raleigh, NC 27619-0921.

CALLING ALL COOKS

Do you have a cobbler recipe that to wow the masses? A soup to chill a snowman? Maybe a pasta sauce or a seafood platter to die for? Do you handle chocolate like Donatello handled bronze?

Well, start collecting your recipes, because FIRST is undertaking development of a cookbook to raise funds and strengthen public awareness of our foundation and its underlying disorder. We'll be collecting recipes for appetizers, soups, main courses of all kinds, desserts, salads, and any other specialty items you may have up your sleeve. We'll be collecting recipes for the some time—likely the better part of 1993—but we'll be featuring selected recipes in future issues of *FOCUS*.

Be sure to include all ingredients and special cooking instructions. Offer any tidbits about the history of the recipe, if you like, and by all means include a note about the cook!

Any Artists Out There?

We'll be including artwork in the cookbook, small line drawings and other subtle items, so please send along your cuisine art, or rope in the services of a friend or neighbor.

Give it some thought and send your recipes to the FIRST office:

FIRST Recipes
P.O. Box 20921
Raleigh, NC 27619

Send your recipes and artwork to the attention of Heather Gattuccio.

F.I.R.S.T. DISTRIBUTES SAMPLES

Some of you in the states of Michigan, New Jersey, and California will soon receive (if you haven't already) a complimentary sample of a urea-based skin lotion. F.I.R.S.T. participated in the distribution of this sample, and the program was conceived as a service for our membership. We may from time to time provide other product samples.

Be aware, however, that our cooperation in distributing these samples ***does not*** imply an endorsement on the part of F.I.R.S.T. for this or any other product. You should also be assured that names and addresses of foundation members who received this sample were ***not*** given out to the manufacturer of the firm whose product we helped distribute.

F.I.R.S.T. never has, and never will, distribute or sell or in any way compromise the proprietry and privacy of its membership rolls.

N.O.R.D. REPORT

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

Who Are The Uninsured?

Not who you might have thought. A study by "Physicians for a National Health Program" and "Public Citizen" shows that 1.07 million Americans with incomes between $25,000 and $50,000 lost their insurance in 1991, compared to less than half that number for the same income bracket in 1990.

The study reports that today 14.1% of the population (about 36 million people) is uninsured. Interestingly, 90% of these reside in just five states: Texas, Indiana, Florida, North Carolina, and Massachusetts. Eleven percent of whites, 32% of Hispanics, and 21% of blacks have no health insurance.

The study also found that only two occupational groups exist in which no one lacks health insurance: legislators and judges.

Clinton & American Health Care Agenda

Among the most important of Bill Clinton's health-care campaign priorities was health insurance reform. He is expected to send to Congress reform legislation within his first 100 days in office. However, both Democratic and Republican leaders of Congress agree that health insurance reform is one of the most contentious issues the new administration will face. While there is almost unanimous agreement that our current system is appalling and that it *must* be changed, there is no consensus whatsoever as to *how* it should be changed. There are obviously no easy answers. The difficulty of the dilemma stymied the Republican administration/Democratic Congress pairing. It will be interesting to see how reform legislation plays out in our new Democratic/Democratic coupling.

Impact of Malpractice on Health Costs

The Bush administration advocated control of malpractice awards as a means of containing spiraling health-care costs. However, studies have shown that most patients harmed by negligent hospitals and doctors do *not* sue, and the total cost of malpractice settlements is negligible in the overall scheme of health care expenses. A study published in a November 1992 issue of *The Annals of Internal Medicine* found that unjustified malpractice awards are very rare, and that in most cases when a patient wins an award for injuries, "the care was, in fact, substandard." This findings, based on 976 cases in which there was a jury verdict (from a pool of 8000 cases studied), found that in 76% of the cases the verdicts favored the doctor. The author of the study is Dr. Mark I. Turgin.

F.I.R.S.T. Develops a Computer "Wish List"

(continued from page 7)

term solution that will not only simplify the foundation's computing, but will build a foundation for FIRST's computing needs well into the 21st century. Says Nick, "The solution is simple: scratch the whole pile of it, go back to the starting block, and set up a system we can more than just live with—one that we can grow with."

Over the past months the E.D.'s been studying this—talking to experts, reading reviews, assessing the Foundation's long-term needs—and he's developed a computing "wish list."

"I don't really like the term 'wish list'," he says. "These are *Goals*. And we'll reach these goals in 1993. For years our computer has been the Foundation's adversary, and our database of members has always been our jailor, in a manner of speaking. But this is going to change this year."

The new system will be an Apple Macintosh system, based on one of the company's new 68030 or 68040 computers (see sidebar). It will have the computing power to handle the Foundation's requirements for ten or more years to come. More important, though, its software will be both powerful, useful, and "user-friendly."

"A nonprofit Foundation like ours has always had a problem with complicated computers and software," says Gattuccio. "We make a mistake when we expect volunteers to be computer experts. That's the beauty of the new system. You won't need to be an expert to become productive with it."

But we need the help of our community to make this goal a reality. Because we can't afford to buy this system, we need to target a donor—a corporation or computer dealer or interested individual who will help us achieve this ambitious goal. We're asking all of you to think about this. And send us your ideas.

We *CAN* make this happen.

F.I.R.S.T.'S 1993 COMPUTER WISH LIST

HARDWARE

Computer:	
Mac II ci with 12Mb of ram and 80-120 Mb hard drive, including grey-scale monitor	$4,500
Printer:	
Postscript (300 dpi)	2,500
Fax-Modem	
9600 bps.	500
TOTAL HARDWARE:	**$7,500**

SOFTWARE

Database: 4th Dimension	$525
Spreadsheet: Excell	295
Word Processor: Word	295
Graphics: Canvas	275
Page Layout: Quark Express	575
Account Manager: Quicken	55
Contact Manager: [undecided]	99
Utilities: [assorted]	250
Statistical: StatView	375
TOTAL SOFTWARE	**$2,744**
TOTAL	**$10,244**

All prices are approximate to within about 10%, reflecting "street prices" at the time of this writing.

FIRST'S 1992 HONOR ROLL

A ROLL CALL OF FIRST'S MOST PROMINENT DONORS

PRIVATE DONORS

$2,000 - $5,000

Dr. Eugene & Mary L. Van Scott

$1,000 - $1,999

James V. & Else Phillips
Dr. Ruy Yu, M.D.

$500 - $999

William & Claire Amwake
Mr. H. J. Bukaty
Sammy & Mary Williams

$200 - $499

John V. & Sandra Ann Adams
Shirley & Carl Anderson
Bruce Batley
Randolph Beimel
Georg'Ellen & Dr. Wilmer C. Betts
Jane Graves Blackford
Melodie M. Buxman
Joseph Chester, Jr.
Jean A. & John J. Cox
Mr. & Mrs. David E. Cromartie
Jessie Doyle Deely, Esq.
Karoline Freed-Briggs
Margaret A. & Ray F. Haywood
Dr. & Mrs. Harold R. Hudson
Ken & Arlen M. Krips
Leonard M. Milstone, M.D.
Dr. Arthur & Mona W. Norins
Gwenda K. Parker
Mr. & Mrs. French Schonyers
Janet L. Showers
Russel & Margaret Vernet

$100 - $199

Dr. Harvey & Elizabeth Adams
Lynne & Harry Alba
Col. & Mrs. Hubert Allen
Howard Baden, M.D.
Candace & Larry Ballantyne
Cheryl Baxter
Reba Benson
Gary & Mardel Bierwagen
Bill & Darlene Blackwell
Patricia & Allen Blackwell
Ronald J. Bongo
Dr. & Mrs. Thomas W. Brown
Elizabeth R. Buehler
Roger I. Ceilley, M.D.
William & Elizabeth Clendenning
Rebecca & Randall Couk
Luc DeBatselier & Kristin Biesemans
Mr. & Mrs. Charles A. Dennery
Denise & Schuyler Eastin
(In memory of Mary Bukaty)
Peter M. Elias, M.D.
Gloria Fickert Estes
Mr. & Mrs. John C. Ferguson
Linette Finstad
Vera M. Finzel
Norene M. & Walter Foxe
Mr. & Mrs. Michael S. Franklin
Carol I. & Thomas E. Frost
Nona Galey
Joseph A. Galluccio, Jr.
Nicholas & Heather Gattuccio
Lowell A. Goldsmith, M.D.
Stephen D. Goodman
Jim Griffin
Mary P. & Theodore Gross
Margaret T. & Roger L. Hall
Larry J. & Nita Halvorson
Ronald C. Hansen, M.D.
Timothy S. Hickey
Dr. & Mrs. Charles H. Hicks
Paul J. & Janice Hudson
Thomas C. Hudson
Charles C. & Lois Jacobs
Carey Kell
Paul Kelley
Richard A. & Jean Kelley
Mr. & Mrs. Joe T. LaBarbera
Wilmer S. & Virginia LaBrant
Justine LaFemina
Jane Henderson & Harry Lancaster
Lucille & Edward Leyba
Gaetano & Mary Licursi
Dr. & Mrs. Raymond Madry
Jack & Nancie Martin
Mr. & Mrs. Brian Martin
Betty & David McMasters
Julie S. Mellum
Lawrence H. Miller, M.D.
Frank J. & Isabel Mosunic
Diane M. Novotny
Herdis L. & F. Pierce Olson
Mr. & Mrs. T.W. Ovbey
Mr. & Mrs. Albert V. Palmiere III
Francene & Hugh B. Parham
William & Estelle F. Patrick
Don & Leslie A. Patterson
Gary L. Peck, M.D.
Blair W. Pride
David K. & Anna Purcell
Mary M. Pyndus
Ieva Rasmussen
William B. Rizzo, M.D.
Mrs. Dudley Hill Sargent
Lawrence Schachner, M.D.
Linda & David L. Schell
Linda Schmidt
Dr. & Mrs. James Semans
Karon & Kenneth R. Shouse
Robert A. Silverman, M.D.
Susan G. Snyder
Marian M. Stanton
Mr. & Mrs. Marvin Stark
Karen & Edward A. Stone
Dr. & Mrs Nicholas E. Stratas
Barbara C. Straw
Pauline T. Suter
Virginia P. Sybert, M.D.
Simon & Magdalena Tarlovsky
Norma C. Treat
Mr. & Mrs. Neil A. White
Walter H. & Mary Louise Wiewel
Mary L. Williams, M.D.
Alice F. Henson Workman
Marilyn & David M. Zinn

CORPORATE & INSTITUTIONAL DONORS

$2,000 - $5,000

Chesebrough-Pond's
Hoffmann-LaRoche
Sandoz Pharmaceutical Corporation
Westwood-Squibb Pharmaceuticals

$1,000 - $1,999

Bristol-Meyers Squibb
Dermik Laboratories
Fraternal Order of the Eagles of Austin, OH
Twin Stars Enterpirses

$500 - $999

Owen/Galderma Laboratories

$200 - $499

Genuardi's Supermarkets
Johnson & Johnson/Consumer Products Div.
March of Dimes of Bloomington, MN
National Organization for Rare Disorders (N.O.R.D.)
National Psoriasis Foundation
Neutrogena Corporation
Sween Corporation

$100 - $199

Schering Corporation

• • • • •

N E W S & N O T E S

Book Titles For Parents of Children With Special Needs

The Association for the Care of Children's Health announces several interesting titles that parents of children with special needs may wish to own. One is an annotated bibliography of over 400 books for toddlers through young adults (includes an extensive subject index) entitled ***Books for Children and Teenagers About Hospitalization, Illness, and Disabling Conditions*** (L. Redburn, editor; #5053-M, $13.50). Also available is a mother's discussion of how parents and families cope with a disabled child in the family—***A Difference in the Family: Life With a Disabled Child***, by H. Featherstone (#3007-1 $7.95).

An interesting twist on the parents' point of view is provided in ***Parents Speak Out: Then & Now*** (#3427-1, #26.00), by A. & H.Turnbull, parents who are also health professionals speaking out on how their families coped with having a child with a disabling condition and the difficulties in obtaining appropriate services.

Titles are available from ACCH, 7910 Woodmont Ave., Suite 300, Bethesda, MD 20814; (301) 654-6549.

Attorney Specializes in Problem Claims Against Insurers.

A Chicago attorney, **Leonard Saphire-Bernstein**, wants us to know that he specializes in representing individuals pressing claims against health insurance companies. Mr. Saphire-Bernstein reminds us how unreasonable insurers can sometimes be when confronted with sizeable claims. He acknowledges that few attorneys are willing to take on cases of this kind, particularly on a contingency basis (that is, attorney fee recovered from the claim itself). About his experience, he tells us "Since graduating from Harvard Law, I have worked with a major Chicago law firm, and then for the federal governement, where I handled disability insurance cases. In 1989 I joined a firm which represented primarily hospitals ... against health care insurers."

Mr. Sapphire-Bernstein may be reached in Chicago at (312) 338-6400.

Useful Child Nutrition Books Available

How to Be Your Own Nutritionist, by Stuart M. Berger, MD (New York: Morrow, 1980). ***Nutrition Smarts: A Cookbook and Guide from Toddler to Teen***, by C. Moore, M. Kerr, and R. Shulman, MD (Hauppauge, NY: Barron's, 1990). ***Child of Mine: Feeding with Love & Good Sense***, by Ellen Satter, RD (Palo Alto: Bull Pub. Co., 1986.)

IN MEMORIUM

Luke John Moran

7 July 1990 - 13 July 1992

We join Helen & John Moran of Victoria, Australia, in grieving the loss of their son, Luke John. He is pictured here with his older sister, Erin. Both have ichthyosis. John Luke was killed in an automobile accident. His parents have made a special garden of roses in his honor.

The FIRST community joins in extending our heartfelt sympathy to the family and loved ones. He will be missed by all.

A SPECIAL THANKS FOR SOME VERY GENEROUS HELPING HANDS

by Frances McHugh
President of the Board

We have another generous donation from the employees of **Hibbard Brown & Company** office in Pittsburgh, Pennsylvania. The managers there are our very good friends **Joe Chester** and **Randy Beimel**. Over the last three years the employees at Hibbard Brown have raised donations for F.I.R.S.T.totaling over $4,000. We are forever grateful for their support. We really could not continue the work we are doing without the generous concern of people such as the Hibbard Brown staff.

• • •

We are very happy to welcome another group of supporters from the Pittsburgh area—the employees of the **L. C. Wegard & Company** office in Monroeville, Pennsylvania. The managers at Wegard—**Jack Adams, Ron Bongo** and **Neil White**—conducted a fund raiser for F.I.R.S.T. which netted almost $1000. We really appreciate the generous support of each and every one of the employees in the L.C. Wegard office. Bless you all.

• • •

Claire Amwake, the aunt of young **Ryan Licursi**, is a real estate agent in Pittsburgh. She and her husband, **Bill**, have been generous supporters of F.I.R.S.T., and it is through Claire that the staffs of L.C. Wegard and Hibbard Brown have been made aware of ichthyosis, of F.I.R.S.T., and of the important work we are doing.

• • •

We should all reach out—all of us together—to thank this Foundation's single most generous donor for the year of 1992, **Dr. Eugene Van Scott** and his wife **Mary**. A member of our Medical Advisory Board, Dr. Van Scott was a pioneer in research into effective treatments for ichthyosis, and remains one of this Foundation's most ardent supporters. We are grateful for the depth of Dr. Van Scott's commitment and its powerful impact on our small community.

FIRST
P.O. BOX 20921
Raleigh, N.C. 27619-0921

NonProfit Org.
U.S. Postage PAID
Raleigh, NC
Permit No. 2209

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, dealing with depression, and other social coping strategies. **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
Harry & Lynne Alba
917 Pierce Road
Norristown, PA 19403
(215) 584-6366

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

Region Three:
TN, GA, FL, AL, MS, LA, AR
Jennifer Tomasik
880 Overlook
Roswell, GA
(404) 587-1886

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

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

Region Six:
TX, OK, NM, CO, KS
Donna Rice
2902 West Elm Circle
Katy, TX 77493
(713) 391-4407

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

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

Netherton's Support Group
Cathy Sipper
P.O. Box 127
Rutledge, AL 36071
(205) 335-6827

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

• • •

All contributions of $100 or more are acknowledged in FOCUS

Y E S !

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

☐	Single	$20 / year
☐	Family	$35 / year
☐	Sponsor	$100 / year
☐	Patron	$500 / year
☐	Benefactor	$1000 / year
☐	Please accept my donation of $ ________	

Name: ____________________

Address: ____________________

City: ____________________ State: ________

Zip: ____________ Phone: ____________

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