



ICHTHYOSIS FOCUS

Vol. 13, No. 1

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

Winter 1994

RESEARCHERS AT BAYLOR COLLEGE OF MEDICINE REPORT FIRST SUCCESSFUL PRENATAL DIAGNOSIS OF EHK BY DIRECT GENE SEQUENCING

An article in the January, 1994, issue of *The Journal of Investigative Dermatology* reports on the first ever prenatal diagnosis of epidermolytic hyperkeratosis (EHK) through direct sequencing of genetic material obtained from the fetus. The breakthrough occurred in the laboratory of Drs. Dennis Roop and Joseph Rothnagel at Baylor College of Medicine in Houston, Texas, who successfully diagnosed the affected status of twins from genetic material obtained at 15 weeks gestation. According to Dr. Roop, prenatal diagnosis of EHK by gene sequencing is feasible as early as eight weeks gestation.

Until now, fetal skin biopsy, an invasive procedure posing significant risk to the fetus, was available for prenatal diagnosis of EHK. However, fetal skin biopsy is unreliable before the 20th week of pregnancy, and even then is not perfectly reliable. Attempts to diagnose EHK prenatally by analysis of amniotic fluid is "not considered reliable enough to exclude the possibility of a false-negative diagnosis."

This work is a direct outcome of discoveries over the past

eighteen months by researchers from several research centers who have mapped the sites of genetic mutations accounting for EHK. (In addition to the Baylor group, this includes Dr. Elaine Fuchs and colleagues at the University of Chicago, and Drs. Sherri Bale, Peter Steinert and colleagues at NIH)

The locating and mapping the specific mutation in a given family lays the framework for diagnosing EHK in utero. This is so because it is known that mutations at any of a large number of sites on either of two genes may account for EHK.

In the present report, then, the first step was to sequence (or map) the genetic mutation in the affected parent. With this map in hand, the Roop and Rothnagel team prepared a second sequence analysis mapping genetic material from the fetus. The two were then compared, and because the fetus bore the same genetic mutation as the affected parent, the diagnosis was positive. To reinforce their findings, the researchers also analyzed corresponding segments of genetic material from non-affected members of the same family and found that none bear this same mutation. They also report that neither is the mutation found in the non-affected population at large.

(Continues on Page 7)

F.I.R.S.T. GETS A NEW COMPUTER

Cellegy Pharmaceutical Corporation of Novato, California, in November provided F.I.R.S.T. with a substantial development grant earmarked for upgrading and enhancing our database management and general computing capabilities. The grant was applied to the purchase of a new Macintosh Centris 650 computer.

We'll continue through 1994 developing our new computing center. We are designing a new database management environment, and by year's end will have the entirety of Foundation records and systems ported over to the new Macintosh platform from our present outdated

DOS system.

But there are many needs yet to meet. We're presently negotiating with software firms for access to expensive programs at affordable prices, and we have yet to build the remainder of our hardware network. Our needs include a high speed, wide carriage dot matrix printer, and a postscript laser printer. Also slated for 1994 is going online with electronic communication via the Internet. Stay posted.

Our sincere thanks to Cellegy CEO, Jerry Simmons, for his most generous commitment to F.I.R.S.T.

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The editor invites correspondence. Please offer us your comments, criticisms, observations and suggestions. Tell us what you wish to see and whom you wish to hear from in *Ichthyosis Focus*.

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

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

My daughter Megan is now two and a half years old, and she is a beautiful and healthy child. It is thanks to you people at F.I.R.S.T. that my husband and I were able to make it through all the hard times and experience the many wonderful times we've had with Megan.

During the holiday season, we often take time to reflect on the past year. This year was especially important to my husband and I because our Megan has passed the "danger" point with her condition. During that time, she has been totally infection free. I guess the real reason I am writing this letter is to let other new parents of these special children know that it's worth the effort. All the doctor visits, the bandaging, the sometimes frightful times. I just have to take one look at my daughter's beautiful blue eyes and I know she is the best part of my life. She is a source of inspiration for everyone around her.

Thank you, F.I.R.S.T., for your newsletter. The letters of encouragement I've read in it sometimes make me cry and sometimes make me smile when I read how strong others with EH are when dealing with the ignorance of the public. But always, always give me the feeling that my Megan will never be alone. She'll always have a group of wonderful friends to turn to when she faces the tough times.

So, happy holidays to everyone at F.I.R.S.T. Keep up all the good work!

P.S. We have had wonderful luck with a new product from Jergens. It's called Advanced Therapy Dual Healing Cream. Just wondering if anyone has tried this.

Cori Pyndus
Cohoes, New York

I find much helpful information in *Ichthyosis Focus*. I've tried all the samples sent

me, but still get the most relief from Eucerin cream. I started using it many years ago when I paid \$1.60 for a pound jar. The work you are doing is wonderful. Keep it up.

Dorothy L. Keene
Westbrook, Maine

I have been diagnosed with EHK. My mother and daughter, who is seven years old, also have it. I am very interested in meeting more people with EHK. Outside of my family I've met only one other person with it. I believe meeting others would benefit my daughter and allow her to see that she's not the only one who has this condition.

I'm interested in hearing about other people's experiences—physically, mentally, and socially. I would also like to offer a word of advice. Even though society doesn't want to accept people with EHK, the more they learn about it the easier it is to accept and the less frightening it is. There will always be unpleasant people in the world, but just remember that you can't change those who can't get past their ignorance.

If people are left to draw their own conclusions about EHK they will only come up with a very distorted picture of what it really is. So always welcome questions. If you don't it will only fuel their fears and concerns.

Karen L. Carey
Fort Smith, Arkansas

My nephew, Ethan, is approaching three years of age. He has EHK. His parents borrowed a television from my in-laws. They liked the television so much that they offered to buy it. My mother and father-in-law insisted that they just keep it, but my sister and her husband sent them \$50.00 anyway. So my in-laws just

Letters continue on page 11

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Deadline for submissions to the next issue of FOCUS: April 1, 1994

NOTES FROM THE E.D.'S DESK

by Nick Gattuccio

This fall has been a busy period of meetings and conferences. Lay organizations such as ours that are members of the National Organization for Rare Disorders (NORD) and the Alliance of Genetic Support Groups met in November, and the professionals under the canopy of the American Academy of Dermatology (AAD) met in December. The Coalition of skin disease patient advocacy organizations (CPA-SDR), of which F.I.R.S.T. is a member, also met at the AAD conference in Washington in December.

It should be no surprise that the spectre of impending health care reform legislation overshadowed all of these meetings and conferences. Shuttling back and forth, as I did, between meetings of patient advocacy organizations and professional medical groups created a bizarre sort of double-vision.

Interestingly, the issues which most concern the two groups (patients and professionals) are remarkably similar. What differs—and differs significantly—is the point of view. What I mean is, both groups are greatly concerned about a set of issues which, taken together, amount to preserving the relationship between dermatologists and their patients. However, what differs between patients and professionals is the set of obstacles which health care reform may throw in the way of the patient-physician relationship.

The big concern from the point of view of professionals is the “gatekeeper.” In any HMO-style health care system (the Clinton plan, as well as other plans presently or soon to be in Congress, provides for an HMO-style health care system), a patient must pass through a “primary care physician” (a general practitioner or pediatrician) whose place it is to evaluate a patient’s needs and, if the primary care physician deems it appropriate, refer the patient on to a specialist.

This system disturbs dermatologists for two reasons. First, there may be financial disincentives for primary care physicians to refer patients to specialists except in the most extreme cases. Patients are a physician’s lifeblood, after all, and it is in no physician’s self-interest to be sending his or her patients to another physician. With specialties like surgery

this is not an issue, since referral circumstances are so clear cut and obvious. With dermatology, though, things are often far less obvious.

The second reason for dermatologists’ concern over the gatekeeper system pertains to diagnosis. Dermatologic diagnosis is frequently visual, and the depth and breadth of most dermatologists’ visual experience can not be matched by that of most primary care physicians. This increases the probability of misdiagnosis, and also increases the likelihood of primary care physicians electing not to refer to a dermatologist.

For ichthyosis patients the gatekeeper system is troublesome, too. No one needs to tell an ichthyosis patient that they require a dermatologist’s care—and not just any dermatologist, but one with a special knowledge of, and interest in, ichthyosis. Ichthyosis patients are capable of self-referral. Furthermore, ichthyosis is a rare and complex disorder, and most of us know from experience that not all physicians (and not all dermatologists, for that matter) are equally equipped to treat it.

For patients, then, the dangers of a gatekeeper system are many, including duplication of services (multiple visits to ultimately see a dermatologist), misdiagnosis, and inexperienced care.

At stake for patients, we see, is our freedom to refer ourselves to dermatologists and elect the physician of choice. At stake for physicians is access to the broad pool of dermatology patients. In a sense both appear to be the same issue, since the outcome is identical—the patient/dermatologist relationship. They are different, though, in that patient concerns are driven by the need to preserve the continuity and quality of their medical treatment, while physicians’ concerns are driven largely by economic self-interest. This is not to say that physicians are not deeply concerned with the welfare of their patients; I mean only to underscore the terms of the debate.

There is a great deal more to say about terms of the various health care reform proposals now under consideration. We shall continue discussing the central issues of the health care reform debate in coming issues of *Focus*. Stay tuned.

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F.I.R.S.T. JOINS IN THE FIFTY PEAKS PROJECT: CLIMBING FOR A CAUSE

Five Climbers, All With Disabilities, Will Climb the Highest Peak in Each of the Fifty States to Call Attention to Serious Disease, Disability & National Health Concerns

It will be the busiest 100 days in the lives of five disabled climbers who will attempt to set a record for scaling the highest peak in each of the nation's fifty states. The previous record is 101 days.

The purpose of the Fifty Peaks Project is to call attention to serious diseases, the people who have these diseases, and to help change the public's attitudes about perceived limits these people face.

Beginning on Mt. Mauna Kea in Hawaii on April 24th, the adventure will end with the nation's highest peak, Mt. McKinley on August 4th. The entire last month of the expedition is scheduled for climbing this difficult peak.

Each Peak Dedicated to a Specific Cause

Each of the fifty peaks is dedicated to a specific disease cause. For example, Mt. Rainier in Washington is dedicated to Down Syndrome, Backbone Mountain in Maryland is dedicated to epilepsy, and Mt. Marcy in New York is dedicated to AIDS. Representatives from organizations representing each disease group will participate in ceremonies at the trailhead at the beginning of each climb. Someone with the designated disability will pass to the climbers a symbolic emblem that will be carried to the summit of the mountain. All fifty emblems will be carried along on the final climb of Mt. McKinley, the highest point in the U.S.

In the words of Project Coordinator, Rick Porter, "The Fifty Peaks Project is a grand symbolic

gesture, not of disabled climbers climbing mountains, but about the challenges of life. The 100 day record attempt symbolizes the time each of us have in this life to deal with our own challenges—the time we are all given to walk our own individual paths and ascend whatever mountains or obstacles are before us."

F.I.R.S.T. Set to Represent Skin Disease

The highest peak in the state of Tennessee has been designated for skin disorders, and F.I.R.S.T. has been selected to serve as representative for this broad group. The state's highest point, Clingman's Dome (6,643 ft.) will be climbed on April 30th. Represent-

tatives from F.I.R.S.T. will join the climbing and support teams, local dignitaries and the press at the trailhead to participate in ceremonies and pass the symbolic emblem to the climbers. Clingman's dome is located in the Great Smoky Mountains just west of the North Carolina state line.

Anyone interested in participating in the activities on April 30th should call Nick Gattuccio at the F.I.R.S.T. office in Raleigh.

We look forward to this being a golden opportunity to raise public awareness about skin disease in general, and about ichthyosis in particular. We hope to bring together a full contingent of people to represent our cause and our group.



The Climbers: (rear) Todd Huston, Helen Klein, Mary Ann Racanelli; (front) John Goggin and Erik Safewright.

John Goggin, 42, is a chronic asthmatic. He lives in Boulder, Colorado, where is director of the Boulder Mountaineering School.

Mary Ann Racanelli, 25, lost her sight at the age of five due to congenital glaucoma. She lives in Chicago where she can be found ice skating, parasailing, jet skiing, or playing piano, drums or bass guitar.

Todd Huston, 32, lost his leg as a teenager in a boating accident. He is a psychotherapist and director of the Amputee Resource Center in Brea, California.

Helen Klein, 71 year old, is a retired nurse, mother of 4, grandmother of 9, and great-grandmother of 3. She is presently holder of the world record for her age group in the 100 mile run.

Erik Safewright, 14, lives in Charlotte, North Carolina, and suffers from muscular dystrophy. Erik is an honor student and his love of mountains has taken him from the Blue Ridge Mountains to the Swiss Alps.

CAMP KNUTSON II

For the Second Year, the American Academy of Dermatology Sponsors a Special Camp for Children with Skin Disorders

August 6 - 13, 1994

If last year's experiences are any indication, this year's summer camp at Camp Knutson will be a blast! Last year forty-nine young people with skin disorders, representing 26 states and Canada, attended the week-long summer camp on the Whitefish lakes north of Brainerd, Minnesota. Sponsored by the American Academy of Dermatology, the special week included many young people from the ichthyosis community. In fact, I learned recently that young people with ichthyosis comprised the largest group at the camp.

This year the camp will once again host 25 boys and 25 girls between the ages of 10 and 13. Campers will be selected regardless of their financial means, geographic location or skin disorder. There is no fee for the camp, and scholarships for transportation will be available for those at need. Bus transportation from the Minneapolis

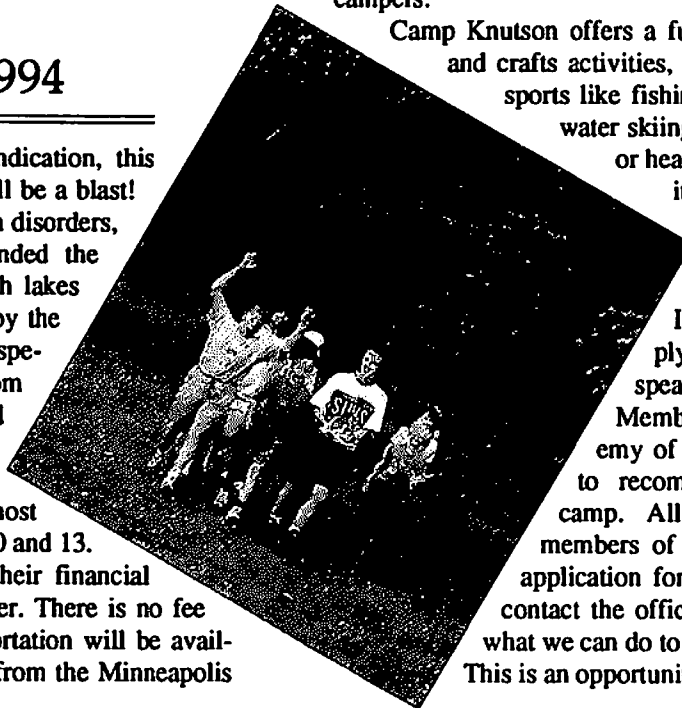
airport to the camp will be provided.

The camp is equipped to handle children with special needs and each camper's medical requirements will be individually monitored. The camp will be professionally staffed with trained counselors, volunteer dermatologists, a camp nurse and a dermatologic nurse. A group of young adults with skin disorders will act as counselors and friends, to offer support and advice to campers.

Camp Knutson offers a full range of sports and arts and crafts activities, with an emphasis on water sports like fishing, swimming, boating and water skiing. For children who are sun or heat sensitive, waterfront activities are scheduled for late afternoon, and many alternative indoor activities are available.

If you are interested in applying for this year's camp, speak to your dermatologist. Members of the American Academy of Dermatology will be asked to recommend candidates for the camp. All dermatologists who are members of the Academy should have application forms. If you have difficulty, contact the office in Raleigh and we'll see what we can do to help.

This is an opportunity not to be missed!



I was born September 13, 1961 to Allen and Mildred Essick Horton of Pilot Mountain, North Carolina. I am the oldest of four girls. While growing up my family and I raised Tobacco. My father died when I was ten. Though times were hard, my mother seen to it that we had the necessities. I look back to this part of my life and realize I had much more than a lot of children. We had our health, a mother who kept us together, and most of all a love for each other that many never share.

During high school, I met a wonderful young man named Darrell Joyce. He soon became my best friend. We were married June 30, 1979. Though we were very young, we have grown up together and made a wonderful life.

On January 31st, 1981, I gave birth to our daughter. Brook soon became the center of our lives. Darrell and I had planned to have only one

PERSONAL PROFILE

Robin Joyce
RSN Region Eight Coordinator



Robin Joyce with her husband, Darrell, daughter Brook, and son Ethan.

child, but could not imagine Brook without a brother or sister. She had so much love to give a sibling. So on February 21st, 1991, Ethan was born. I took for granted that because Brook was healthy, Ethan would also be healthy.

Ethan was born with EHK.

The first year was tough. I remember thinking, how could a skin disorder have such an effect on all of our lives. I honestly believe that everything that happens in life is for a reason. I have learned so much about the little person I was before Ethan's birth. I have learned to look beyond the appearance of someone and see their true beauty within.

At the present time, I have returned to school and I am in the first year of an associate degree program in nursing. Upon completing this program and passing the state board examination, I hope to be employed as a registered nurse in North Carolina.

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.

Gene Therapy Protocols Underway

As of mid-1993, 59 gene therapy/transfer protocols had been approved by the NIH Recombinant DNA Advisory Committee (RAC). Of these 59, 36 were for therapeutic experiments, while 23 were for gene marker experiments with not therapeutic value. The 36 "therapy" protocols involved 26 experiments for cancer, 4 for cystic fibrosis, 2 for AIDS, 2 for Gaucher's disease, 1 for severe combined immune deficiency (SCID-ADA), and 1 for familial hypercholesterolemia.

Approximately three-quarters of gene therapy/gene transfer experiments are for cancer. Few experiments are being pursued on genetic disease.

Interestingly, companies involved in the manufacture of "vectors" (altered viruses that carry genetic material into the cell) have been applying for (and receiving) orphan drug designations for their vectors. This means that competitors will not be permitted to manufacture similar vectors for seven years after the FDA grants marketing approval.

How Much Does That Prescription Cost?

Don't ask your doctor. A University of Missouri study that surveyed 100 primary care physicians found that most do not know the prices of the drugs they commonly prescribe. The Missouri study found that doctors underestimated prices of 18 out of 25 popular prescription drugs. Some estimates were as much as 70% lower than actual retail cost.

New Report on the Uninsured

The non-elderly uninsured population in the United States increased by 6.1% during 1992. The U.S. Census Bureau's March 1993 report, *Current Population Survey*, found that 38.5 million Americans under the age of 65 had no health insurance during 1992. The 1991 figure for the uninsured in the same age group was 36.3 million. This is an increase of 2.2 million people. The recession, which caused massive job losses, had a lot to do with loss of insurance benefits, but the rising costs of health insurance also led to many small businesses dropping their insurance policies.

Who's Hurting?

According to the January 3rd issue of *Forbes* magazine, the health care industry in general "has been the most profitable industry in the country over the past five years." Health care companies bumped food, drink and tobacco out of the top spots. Seventy-four of the health care companies tracked by *Forbes* showed an 18.3% return on equity. The group was led by 23 pharmaceutical companies showing 26% return on equity.

The American Way

Rep. Ron Wyden (D-OR) has been investigating the implantable birth control device known as Norplant, made by the firm Wyeth-Ayerst. The drug was developed in the U.S. by the Population Council and was licensed to Wyeth-Ayerst, but it sells at much lower prices in other countries. Norplant costs \$365 in the U.S. (not counting physician charges) and yet it sells in Sweden for just \$23. The top price in any country outside the U.S. is only \$120. The cost to manufacture each device is only \$16.

At a recent congressional hearing, according to the *New York Times*, a Wyeth-Ayers official said, "the primary reason the price has been kept high was to prevent the drug from becoming known as a 'poor woman's drug', and thus perhaps be shunned by middle class women."

NEW DIRECTOR ELECTED BOARD MEMBER & FORMER DIRECTOR OF THE RSN DEPARTS

At its December, 1993, board meeting, F.I.R.S.T.'s board of directors in turn expressed their thanks for many years of strong and committed service to departing board member and former director of the RSN, Cynnie Bates of Lexington, Kentucky. If this is not enough, Cynnie is one of the co-authors of F.I.R.S.T.'s forthcoming handbook for parents of children with ichthyosis.

Cynnie's has always been a strong and forthright voice speaking on behalf of the ichthyosis community. During her three-year tenure on the board she was steward over period of greatest growth and change in the Foundation's history. Her leadership, her forthright views, and her voice tuned to the membership will be sorely missed. We wish you good luck!

Elected to a three-year term on the board at the same meeting was Michelle Petersen of Metuchen, New Jersey. We first met Michelle at the national conference in Chicago last June.

Michelle has experience serving with several service organizations, including the PTA and Metuchen Organization of Women. She has a B.A. in Business Administration and an M.A. in Industrial Psychology, and is presently vice president and Manager of Trading Support for the Republic National Bank of New York.

We welcome Michelle to the board and look forward to progress and success in the coming years.

GRASSROOTS

An organization is only as strong as its members. F.I.R.S.T. continues to be fueled by the energy and dedication of its friends and family in the ichthyosis community. We are growing in strength and numbers, and we have ambitious goals for 1994.

With the new year upon us, it is time to create new strategies to generate the funds we'll need to achieve these goals. We ask all F.I.R.S.T. members, as you have in the past, to recognize the significant role you play in strengthening and furthering the mission of this Foundation. We are a grassroots organization, and you are our greatest resource. No effort at fund raising or raising public awareness is too small in the ongoing fight against ichthyosis.

The very idea of raising money can be daunting. What makes it so fearsome is that we think that big efforts raising large sums of money are the only efforts that really matter. This couldn't be farther from the truth. It is the accumulated sum of a

great many small efforts that make the big difference.

We're throwing out a few ideas that have proven to be both productive and fun in the past, both for members of F.I.R.S.T. and for grassroots fund raisers at other organizations like ours. The important thing is, one doesn't need to be a professional fund raiser, have a lot of money, or know "important" people to raise money. You simply need a good idea, a workable plan, and some friends and family willing to help you pull it off.

Grassroots fund raising can open doors to new people and new experiences. It can give people in your community an opportunity to prove they support your cause. Most of all, it can raise public awareness of ichthyosis and the obstacles our community faces while raising funds to further the programs and services of the Foundation.

Have some fun in the name of F.I.R.S.T. Happy new year!

Prenatal Diagnosis (continued from page 1)

In conversation, Dr. Roop remarked that parent screening is this procedure's key. Parents affected with EHK who feel they may wish prenatal evaluations should themselves be evaluated to establish a baseline sequence analysis.

Unfortunately, few sites in the U.S. are presently able to do this procedure. In addition to the Baylor College of Medicine, though, we know of two other sites whose research teams have published results of their studies of the genetics of EHK and who are equipped to perform sequence analyses of patients with EHK. One is Dr. Elaine Fuchs' lab at the University of Chicago; the other is the Laboratory of Skin Biology at the National Institute of Arthritis, Musculoskeletal and Skin Disease (NIAMS) of the Na-

tional Institutes of Health in Bethesda, Maryland.

The implications are far-reaching. According to the article, diagnosis by gene sequencing "can be used to screen *in vitro* fertilized embryos, from affected couples, prior to implantation. Once screened, unaffected embryos can be selected for transfer to the uterus, thereby eliminating the need for risky surgical procedures for prenatal diagnosis later in the pregnancy." Dr. Roop added in conversation that just such a procedure is used by families known to be at risk for cystic fibrosis and muscular dystrophy.

Rothnagel JA, Longley MA, Holder RA, Küster W, and Roop DR. "Prenatal Diagnosis of Epidermolytic Hyperkeratosis by Direct Gene Sequencing." *The Journal of Investigative Dermatology*, vol. 102, no. 1, pp. 13-16; January, 1994.

GRASSROOTS FUND RAISING EVENT IDEAS

- Special sale, like a book sale or plant sale
- Holiday dances or bazaars
- Special movie night -- a special screening of a film in cooperation with a local theater
- Bake sale
- Garage sale. This can also be a neighborhood event
- Quilt/Afghan raffle
- Car wash
- Sewing sale
- Bike-A-Thon
- Bowling Tournament, or bowl-A-thon
- Golf tournament
- Cook-off
- Haircut-A-Thon
- Dress-down day at work
- Bridge party
- Marathon or other kind of race
- Auction
- Bingo (where legal)
- Celebrity lecture or reading by noted author or poet

Be creative. Frequently it is possible to tie your event in with a local civic group, club, school or church.

And always feel free to touch base with the office in Raleigh for suggestions or assistance of any kind with your event.

Thanks from all of us here to all of you out there!

Q & A

EDITOR'S NOTE: This issue marks the introduction of a new format for our Q & A feature. Following nearly four years of dedicated service, Dr. Melodie Buxman has relinquished her role as columnist for this feature. We thank her sincerely for her years of committed service. Beginning with this issue, we will employ the services of our new, seven-member board of medical editors who will address reader questions on a teamed and rotating basis. While on the one hand lightening the work load for any given physician, this will offer the added advantage of giving us a broader range of physician viewpoints on some topics of concern to our readers. We sincerely hope you appreciate the change. NG

Q. My 16-month-old daughter, who we believe has lamellar ichthyosis, has thick, leathery patches of skin on her scalp. We have tried various products to help get them to come off more easily, but haven't been very successful. Our dermatologist suggested we try Retin-A gel (0.01%) on her scalp once a day. It does seem to help the patches come off more easily, but I have begun to worry about possible side-effects since she is so young. Should I be concerned?

A. Dr. Philip Fleckman, MD:

No one knows the absolute answer to your question, but based on a few assumptions, your child is probably at very little risk for systemic side effects from the Retin-A. Absorption of topical Retin-A gel in adult skin is about 5-10% of what is applied. Absorption in young children with normal skin is probably slightly greater. However, experience is limited, and might not be the same in your daughter's skin. Nonetheless, if one made the assumption that *all* of the applied Retin-A were absorbed, the normal circulating levels of retinoic acid would be increased only twofold. This is probably not significant, since "normal" levels vary in adults by as much as a factor of five.

Systemic side effects of orally administered retinoids in young children appear to be limited to occasional, unpredictable premature closure of the growth plates of long bones. This is with doses that result

in approximately 100 times the normal levels. There could always be other, unknown side effects. But even with the conservative estimates listed above, the topical Retin-A would probably not affect your child adversely.

The gel form of Retin-A contains a fair amount of alcohol and irritates some peoples' skin. There may be less irritation with the cream form.

A. Dr. Mary Williams, MD:

The major side effect of Retin-A creams and gels is skin irritancy, usually experienced as a chapped skin-like reaction. People vary tremendously in how sensitive they are to irritancy reactions to Retin-A. Young children may be no more sensitive than many adults. Skin on different parts of the body also varies in how sensitive it is to irritancy reactions. Usually the scalp is not a particularly sensitive area. Irritancy, if it does occur, is not a serious side effect; it's more a nuisance and will resolve on its own when you stop using the Retin-A. You can usually safely resume its use once the reaction has resolved, if you use it less frequently (e.g., every other day), or use a weaker formulation (in many instances the 0.025% cream is milder than the 0.01% gel).

You have probably heard something about the side-effects of Accutane and Etretinate, which, like Retin-A, are derivatives of vitamin A. When taken orally in very high (supraphysiologic) doses, all vitamin A

related drugs have the potential for producing bone toxicity; in children there is particular concern that long term exposure to these drugs will arrest the growth of bones. This, however, is a very uncommon side effect, and only occurs after long-term therapy (years) and very high cumulative doses. There is no reason to believe that enough Retin-A could be absorbed across your daughter's scalp skin to cause such internal side-effects.

So, if the Retin-A helps and if your daughter tolerates the treatment without too much irritancy, I see no reason not to use it on her.

Q. I am male, 60 years of age, and I have had ichthyosis since birth. My son also has ichthyosis, and I am writing this letter for him. Unfortunately, he was recently in an auto accident. He broke both legs and had on casts and a Hoffman apparatus on both legs. Of course, he became extremely dry and cracked and irritated beneath the casts and he is miserable with itching. Can you recommend a way to ease the discomfort.

A. Dr. Neil Prose, MD:

The management of itching beneath a cast is a very difficult problem. Until the casts are removed and your son can go back to using his emollients, the only treatment option is the use of oral antihistamines.

A number of different classes of antihistamines are available by prescription from a physician. Unfortunately, all of the antihistamines that benefit itching also cause some drowsiness.

Finding the dose and type of antihistamine that suppresses the itching and causes the least degree of sedation is sometimes a process of trial and error. If he is able to work together with his doctor, your son should be able to get relief from his itching in this way.



F.I.R.S.T.'s Regional Support Network Turns Three

by Heather Gattuccio
National RSN Coordinator

F.I.R.S.T.'s Regional Support Network (RSN) is entering its third year. Dividing the U.S. into eight regions (and presently developing a Canadian branch), the RSN's goal is to execute on a local, person-to-person level the mission of the national organization. Each region is run by a regional coordinator, who is assisted by a regional representative plus a network of individuals and families experienced with ichthyosis. We have also developed a special Netherton's Syndrome support group.

While the RSN began small, with just a handful of dedicated individuals, it has grown into an active, thriving network comprised of several hundred participants. Within this person-to-person network the RSN provides an invaluable source of support, guidance, practical advice, and general information. Parents can exchange practical skin care tips and child care information. At the same time, children can seek out other children and exchange visits, phone calls and letters. Adults with ichthyosis can

support each other and share the outcome of their valuable experience with treatment, job hunting, and social coping strategies, as well as serve as role models for children and adolescents with ichthyosis.

Frequently, members of the RSN can offer a broader base of useful information than even most physicians, since their experience runs so much deeper.

Many regions have hosted successful regional meetings and conferences which bring area families and individuals together with one another and with physicians to discuss important medical and treatment issues in a congenial setting. These regional meetings offer those who can't attend the national conference a means of gathering up-to-date information and finding others near by who share similar life experiences.

F.I.R.S.T. receives countless letters from individuals who want to "get involved," but don't know how. The RSN is an excellent opportunity to offer your personal contribution to F.I.R.S.T., gather useful information, and establish wonderful friendships in the ichthyosis community.

We invite you to contact your regional coordinator and join us today. Please check the back page of this issue of *Focus* to see who your regional coordinator is. You can receive a Participation Form by calling the office in Raleigh at 800-545-3286.

Why Does RSN Participation Require A Separate Membership Enrollment?

We're often asked why you need to fill out a separate Participation Form for the RSN. Why doesn't membership in F.I.R.S.T. automatically enroll me in the RSN, and vice versa?

The answer is that F.I.R.S.T. maintains a strict, ironclad policy of never, under any circumstances, releasing member names or other information *to anyone*. On your RSN participation form, however, you are asked to sign a waiver that allows us to share your name and number with other members of the RSN network (but *only* to other members of the RSN). Without these signed waivers, we could never have created the RSN in the first place.

To receive your RSN Participation form just call the F.I.R.S.T. office in Raleigh, or contact your regional coordinator listed on the back page of this issue of *Focus*.

DONOR BILL OF RIGHTS

On the following page we present our annual honor roll of cherished donors to the Foundation for Ichthyosis. This occasion marks an ideal opportunity for us at F.I.R.S.T. to acknowledge that philanthropy is a two-way street. This Foundation bears obligations to its donors—those of you who keep our doors open. We present here the Donor Bill of Rights, which was drafted by a consortium of several organizations, including the American Association of Fund Raising Counsel and the National Society of Fund Raising Executives, among others. We present this Bill of Rights to write in stone what we feel to be our obligation to you. Please feel free to contact us if you ever feel we are deficient in any way regarding your following rights as donors:

I *To be informed of the organization's mission, of the way the organization intends to use donated resources, and of its capacity to use donations effectively for their intended purposes.*

II *To be informed of the identity of those serving on the organization's governing board, and to expect the board to exercise prudent judgment in its stewardship responsibilities.*

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IV *To be assured that your gifts will be used for the purposes for which they were given.*

V *To receive appropriate acknowledgment and recognition.*

VI *To be assured that information about your donations is handled with respect and with confidentiality to*

the extent provided by law.

VII *To expect that all relationships with individuals representing organizations of interest to the donor will be professional in nature.*

VIII *To be informed whether those seeking donations are volunteers, employees of the organization, or hired solicitors.*

IX *To have the opportunity for their names to be deleted from mailing lists that an organization may intend to share.*

X *To feel free to ask questions when making a donation and to receive prompt, truthful and forthright answers.*

Any questions or concerns about this Donor Bill of Rights should be directed to the executive director in Raleigh. Our donors are our lifeblood, and we respect our obligation to each of you.

FIRST'S 1993 HONOR ROLL

A ROLL CALL OF FIRST'S MOST ARDENT SUPPORTERS

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Books, Books, Books ...

Several books that may be of interest to the community have come to our attention over the past months:

Prenatal Tests: What They Are, Their Benefits and Risks, & How to Decide Whether to Have Them or Not, by Robin J.R. Blatt (Vintage, 1988/ISBN 0-394-75887-0). A comprehensive resource, for pregnant women and their partners that also addresses the emotional and ethical aspects of decision-making in prenatal testing.

Developing Social Skills: Issues for Adolescents with Chronic Illnesses & Disabilities. A new bibliography produced in the CYDLINE series from the University of Minnesota. The guide explores the programs and materials available to help adolescents deal with relationships. Prepayment is required, and the cost is \$6.00. Make check payable to University of Minnesota-NYCYD and send to Box 721,

University of Minnesota, Minneapolis, MN 55455; (612) 626-2825.

Also available from the CYDLINE series from the University of Minnesota is a resource entitled *Legal Issues for Adolescents with Chronic Illnesses and Disabilities and their Families*.

The bibliography may be obtained in the same manner as the title above; also \$6.00 (prepayment required).

F.I.R.S.T. Set to Appear Before Congress

On February 9th, executive director Nick Gattuccio, wife Heather, and daughter Caitlin will appear before the House Appropriations Subcommittee which oversees funding of NIH to testify for enhanced funding for biomedical research. One of the feared outcomes of the present health care reform debate is a threat to research funding. Cuts in federal spending for research nearly occurred in the 1994 federal budget, but Congress restored these proposed cuts.

RSN SEEKS VOLUNTEER IN CALIFORNIA

Regional Support Network membership is growing by leaps and bounds in the state of California, and we are looking for a volunteer to serve as a California State Coordinator. The position will offer a great way to meet people in your state as well as make your personal contribution to F.I.R.S.T.

Duties include serving as first contact for newly referred Californians dealing with ichthyosis. Working with the Region Eight Coordinator and other members of the RSN, you will offer support, encouragement and information. We all know how easy it is to feel alone in the fight against ichthyosis, so if you feel you can make a difference in people's lives, please consider serving.

Anyone interested in this position should contact Heather Gattuccio through the office in Raleigh.

Letters

(Continued from page 2)

decided to donate that money to F.I.R.S.T. in honor of Ethan. So please accept this donation from Bobby and JoAnn Simmons on Ethan's behalf.

Ethan is such a loving and giving child and such a special part of our family. Everyone that meets him agrees that he is an extraordinary child. I am so thankful that Ethan came into my life. He has taught me to look on the inside to find a person, and he has helped me to value my life a little more.

Thank you for your continuing work. It means a lot to many that you probably never know it affects. And thank you, Lord, for Ethan. His wonderful personality and sense of humor and big heart make this world a much nicer place to be!

Jenny Simmons
Westfield, North Carolina

Calling All Cooks ... Again!

In the Winter, 1993, issue of Focus, F.I.R.S.T. members were asked to contribute their favorite recipes for a Foundation cookbook to be used for fund raising. While we received a few heart stirring recipes, the general response was bleak. But we don't give up easily here in Raleigh, so we ask you again to dig into those recipe boxes and drawers or ask a friend or family member to share the recipe of a family treasure.

We're collecting recipes for appetizers, soups, meat and vegetarian main courses, salads, breads and desserts. We'll also be including an international section, so you folks in other countries help us out with an exotic recipe or two.

Be sure to include all ingredients and cooking instructions. Offer tidbits about the history of the recipe and, by all means, a note about the cook. Dedicate a special recipe in honor of a friend or family member with ichthyosis.

All artwork and creative thoughts on content and theme are welcome. Many thanks, and send your contributions to Heather Gattuccio c/o F.I.R.S.T.

I just wanted to write and wish you all a wonderful holiday season and say thank you for your conference in Chicago. It was very professional and educational. It was the first time I had been to a conference. My parents didn't want me to go (I was 16). They thought it would be traumatic. I went. Sometimes it was

scary, but still it was a positive experience. I am sending a small gift to show my thanks.

Tim Ohlweiler
Kaysville, Utah



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Regional Support Network

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

REGION ONE

NY, PA, VT, NH, NJ, CT, RI, MA, ME
Lianne Busby
8001 Miller Road
Avoca, NY 14809
(607) 566-8403

REGION TWO

DE, MD, 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 Lake Overlook
Roswell, GA 30076
(404) 587-1886

REGION FOUR

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

REGION FIVE

WI, IA, MN, ND, SD, NB
Joe & Marlene 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 77439
(713) 391-4407

REGION SEVEN

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

REGION EIGHT

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

CANADIAN SUPPORT GROUP

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

NETHERTON'S SUPPORT GROUP

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

Y E S !

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