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

ICHTHYOSIS FOCUS

Vol. 18, No. 2 A Quarterly Journal for Friends of F.I.R.S.T. Fall 1999

EKV: A Connection with Connexins

In the recent past, enormous progress has been made in understanding the molecular cause of inherited skin disorders, especially in epidermolysis bullosa and congenital ichthyoses. Little is known however, about the group collectively called erythrokeratodermias (EK). This group forms a broad spectrum of overlapping disorders characterized by co-existence of two distinct features: thickening of the outermost layer of the skin called epidermis (hyperkeratosis), and redness (erythema). The best-recognized and most common disorders are erythrokeratodermia variabilis (EKV), and progressive symmetric erythrokeratodermia (PSEK). Four years ago, I was part of Dr. Sherri Bale's research group at the National Institutes of Health in Bethesda. We, together with Dr. John DiGiovanna from the Department of Dermatology of Brown University/ Rhode Island Hospital, Providence, decided to join our research efforts, and to embark on clinical and molecular genetic studies of EK. We now have discovered the first gene whose mutations underly EKV. I am currently at the Department of Dermatology and Cutaneous Biology at Jefferson Medical College, Philadelphia, and my laboratory is continuing the research on EK.

EKV is a chronic skin disorder with autosomal dominant inheritance, which was first described in the Netherlands. To date, more than 200 cases with diverse genetic backgrounds have been reported worldwide. The hallmark of EKV is well demarcated, figurate outlined, red patches of variable intensity that may involve any part of the skin surface. These patches last only minutes to days and may be preceded or accompanied by a burning sensation, which may cause serious discomfort for patients. The remarkable variability of the red patches in number, size, shape, location and duration are a typical feature of EKV that is reflected by the name of the disease.

Independent of the erythema develops hyperkeratosis, a localized or generalized thickening of the skin. The localized type is characterized by sharply demarcated, geographically outlined, fixed, yellow-brown hyperkeratotic plaques. The plaques are almost symmetrically distributed over the limbs, buttocks and trunk, often with sparing of the flexures, face and scalp. They are usually relatively stable, and last for month to years.

The severe, generalized type of hyperkeratosis presents as persistent, yellow-brown-gray thickening of the skin with accentuated skin markings, and fine scaling. In about half of EKV families, hyperkeratosis involves also palms and soles as a patchy or diffuse palmoplantar thickening of the skin, often associated with peeling. Frequently, either erythema or hyperkeratosis predominates, and occasionally one may be missing. Skin lesions in EKV may constantly change their appearance, and vary tremendously between different persons. They can be triggered by internal and/or external factors, including stress, sudden temperature changes, mechanical friction and sun exposure. EKV usually presents at birth or during infancy and persists throughout life. It may be severely disfiguring and can have tremendous psychosocial impact, although improvement and periodic clearing of the skin are not unusual. The treatment of EKV is symptomatic, and aimed to diminish the hyperkeratosis and minimize the discomfort. It includes the regular use of emollients, moisturizing creams and lotions, and keratolytics such as urea, alpha-hydroxy acids, propylene glycol, and topical retinoid preparations. In severe cases with generalized hyperkeratosis, the systemic treatment with retinoic-acid derived drugs (Soriatane, Accutane) is very efficient.

To elucidate the genetic basis of EKV, we collected and examined an exceptional number of 53 patients with EKV from 12 unrelated multigenerational families, a unique resource of this rare disorder. All participants gave informed consent to our clinical and genetic studies, for which we obtained DNA from either blood or buccal cells. In the 1980's, EKV had been mapped to the short arm of chromosome 1 near the Rhesus Factor blood group locus. Hence in a first step, we selected the 6 largest EKV families for genetic linkage analyses to refine the gene localization.

Linkage studies compare in a given family the inheritance of a disease (EKV) with the inheritance of anonymous DNA marker loci. These markers are amplified from DNA samples by polymerase chain reaction (PCR) and analyzed by gel electrophoresis. If a DNA marker is located near the disease gene, it will be transmitted from the parents to the children together

continued on page 3

2000 Family Conference
See page 8

Correspondence Corner

Dear F.I.R.S.T.:

For many years my brother has given me the Saturday Evening Post as a Christmas gift. In one of the doctor's columns, they talked about ichthyosis. That was the first time I have ever seen anything about it, so I sent for the information and your foundation sent it out to me. I am from a family of five children, three boys and two girls…only myself and my sister were born with ichthyosis.

In 1996, my friend Helen asked me to be in the Ms. Senior Connecticut Pageant, and I said "Me, oh no!" But she kept asking, so I said, "Let me pray on it and I will give you an answer." I have been blessed with my deep faith in the Lord, my family and friends. I prayed, and finally it came to me…if you are given a gift, as we all are, they should be shared with others and not hidden. To my surprise, I won the Ms. Senior Connecticut Pageant!

No, I didn't win the Senior America Pageant, but I felt I had already won, with the courage it took me to do this, and all the people who have touched my life. I have always been a shy person, but at sixty-two in 1996, I found it's never too late to try to do the things you always wanted to do, with the love that comes from deep within your heart.

Claire Okon
Windsor Locks, CT

Dear F.I.R.S.T.:

Yours is a wonderful organization. I became acquainted with you via an Ann Landers or Dear Abby column some years ago. Since then your newsletters made me aware of the many people, especially children, afflicted with ichthyosis. Also, I never knew, before contacting you that there were other types of ichthyosis besides vulgaris, which I have. I found out how fortunate I am compared to others.

From 13 months old to about 21 years of age I also battled severe weeping excema. As a child I would fantasize that my skin had a zipper that I could unzip and step out of with smooth clear skin! My reason for mentioning this is to assure you that if I can be of any assistance to parents/children I would be happy to do so. Childhood and adolescence can be an uphill battle in the best of times without having to cope with medical problems seen by one's peers.

However, I do not want my name, address, etc. on any list sold, rented or given to businesses or fund raising organizations. I receive too much of that kind of mail as it is!

Thank you for the great work you do.

Patricia Blackwell
Naples, FL

Editor's Note: F.I.R.S.T. does not sell, rent or give our member's names and addresses to any other organization or business.

Dear Friends:

Let me first introduce us and explain our interest in F.I.R.S.T. We are the parents of a wonderful child born with ichthyosis in 1951. She has lived with this skin condition and all its effects and we think she has accomplished much, which I will cover later in this letter.

Our first child, a son was born with ichthyosis in the then small town of Orange, Texas. The attending physician and no one else in the hospital had ever seen a child or anyone else with this skin condition. Treatment for our child was makeshift at best and as a result our son lived only 21 days. The hospital and staff did all they knew to do at the time. Our next child, a son, still has normal skin, after 49 years. Our third child, Teresa, was born with ichthyosis. It was touch and go for several months of Teresa's early life. Infection and all that goes with it were always present and frustration by the caregivers and us strained everyone's patience and nerves from time to time. The best part is that Teresa lived and has been an inspiration to us and to many others.

Teresa was born in Houston, TX, and attended school there until we moved to California. It was there that Teresa was introduced to the Stanford Hospital Dermatology Department where the staff knew something of ichthyosis and exhibited an interest and support in treating and improving the patients well being.

Teresa graduated from high school, obtained a B.S. degree in nursing from San Jose St. University and commenced working in a hospital. She got married and gave her parents a healthy normal grandson whom she has raised for the most part as a single parent. Teresa decided that nursing wasn't all she wanted to do. She has since earned her MBA and worked in various non-medical fields.

You may have gathered that Teresa has been a challenge and a joy in our family and our life and we are indeed proud of her accomplishments. Teresa has more experience and knows more about taking care of this condition than most. She advised me of your organization and your web site and I believe I can be a valuable contributor to F.I.R.S.T.

Isabell & Merle Blanton
Sedona, AZ

EKV continued

with the disease gene (the marker is "linked"). In contrast, if the DNA marker is distant from the disease gene, it has a different inheritance pattern (the marker is "unlinked"). We analyzed in all individuals of our 12 families 20 different DNA markers distributed across the short arm of chromosome 1, several of which we found to be linked to the EKV locus. Statistical linkage analyses determined a critical region of about 2.6 megabases (2.6 Million DNA base pair) harboring the gene causing EKV in chromosome band 1p35.1. In our search for the EKV gene, we identified within this interval a cluster of connexin (Cx) genes. Connexins are a family of transmembrane proteins that assemble to form intercellular channels at gap junctions in the cell membrane. These gap junctional channels allow the exchange of ions, small molecules and messengers between neighboring cells, and hence act as a fast intercellular communication system. We cloned and characterized 2 novel Cx genes encoding connexin-31 (Cx31) and connexin-31.1 (Cx31.1), and demonstrated their expression in human epidermis, where they are probably involved in the growth and differentiation of keratinocytes, the major cell type of the epidermis.

To determine if these genes are causally involved in EKV, we screened the DNA sequences encoding the connexins Cx31 and Cx31.1 in one affected individual from each of our 12 EKV families. While we failed to detect any sequence variations in Cx31.1, direct DNA sequencing of the gene encoding Cx31 (designated GJB3) revealed three different mutations in four EKV families. Each mutation changed a single nucleotide in one strand of the patient's DNA (missense mutation), which is predicted to alter the conserved amino acid sequence of Cx31 and its structure and/or

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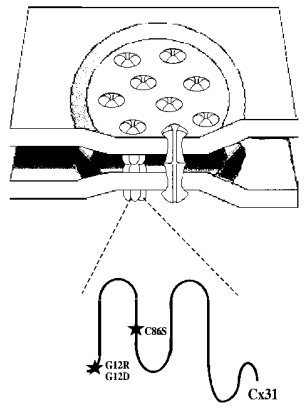

Figure Legend: Top: Schematic view of a gap junction showing the clustering of several gap junctional channels at the cell membranes of 2 neighboring cells, through which signals and molecules can be exchanged. Bottom: Schematic view of the protein structure of connexin-31. Stars indicate the position of mutations found in EKV.

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

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Executive Director's Report

Hello to everyone! I am Jean Pickford, the new executive director at F.I.R.S.T. I have been working in the non-profit world for 10 years and have been employed by the American Heart Association and the National Tay-Sachs & Allied Diseases Association of Delaware Valley. I have been married for six years to Stephen and have one child, Matthew who is 15 months old. Having a toddler doesn't leave much time for myself, but when I do have some free time I enjoy refinishing furniture, decorating my home and shopping.

My most sincere thanks to all of you who contributed to the foundation through the Summer Appeal mailing. As of this writing, F.I.R.S.T. has received over $10,000! So far, it has been a terrific campaign and I keep receiving donations every day! This is a great beginning to our fundraising efforts for our fiscal year 2000.

Next, I want to introduce the office staff who are working hard to continue our valuable mission. Welcome to our new administrative assistant, Kelly Strother, who started in early October and will be with us on a regular part-time basis. Another hearty welcome to our Support Network consultant, Michele Bolles, who will be "re-energizing" the Support Network for the next three months. Look forward to some exciting changes!

Ichthyosis Awareness Week (IAW) took place during October 3-10. As you will see on page 3, many of our members took part in promoting awareness and/or fund raising. Thank you for all your efforts. I would love to see even more participation next year so we can continue to increase the public's awareness about this disease.

The 2000 Family Conference plans are going well. Thanks to Lynn Alba and her assistance in locating a great hotel, we are booked and ready to get started for July 2000. If anyone is interested in helping with the conference, please contact me. Remember, this conference is for you so your input is greatly appreciated!

Since this is my first of many reports in the newsletter, I want to take a quick moment to share with you my enthusiasm and commitment to the foundation. I foresee great things happening at F.I.R.S.T. and look forward to working with all our members.

Best regards,

Jean

P.S. If there is something you would like to see featured in an upcoming issue of the newsletter, please call me! I welcome new ideas!

Agreement under U.S.-Canada treaty facilitates cross-border charitable contributions (Notice 87-47, 1999-36 IRB 344)

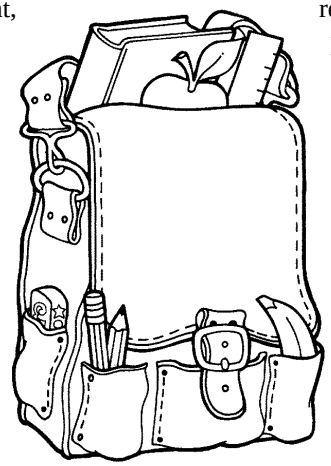
The U.S. and Canada Competent Authorities have entered into a mutual agreement that implements Article XXI as contemplated by the diplomatic notes. Under the terms of the agreement, recognized religious, scientific, literary, educational, or charitable organizations that are organized under the laws of either country will automatically receive recognition of exemption without having to apply for it in the other country. U.S. organizations must be recognized as exempt under Code Sec. 501 (c) 3 in order to qualify for this treatment. Similarly, Revenue Canada must recognize Canadian organizations as Canadian registered charities. Recognized charitable organizations resident in one country will be eligible to receive deductible charitable contributions from residents of the other country.

Source: Excerpt reprinted from Research Institute of America: Federal Taxes Weekly Alert, September 9, 1999.

Kids and the Classroom

Am I going to find the right classroom? Will the other kids make fun of me? What will my teacher be like? Will I have homework the first night? Where's the cafeteria? What bus do I take home?

The first day of school is a very nerve-wracking experience for parents and their children. The anxiety is even greater if you are the parent of a child with ichthyosis. As a parent, what can you do to ensure the most nurturing, profitable learning experience for your child? The following are steps recommended to set your child on a path for success in their school experience.



Schedule a meeting with your child's teacher, school support personnel (guidance counselor, etc.) and the principal. If at all possible try to arrange this meeting before school starts, or as close to the beginning of the year as possible. Explain the unique adaptations to the learning environment that your child will require. If you are unsure of what may be required, a booklet called *Ichthyosis, A Guide for Teachers*, is available at the F.I.R.S.T. office to help explain the challenges your child may face in their school environment. There is also a professional video called *Butterflies: The Children of Ichthyosis* that will further enlighten the school personnel to the challenges of your special child. This meeting could be all that you need to guarantee the success of your child.

Most school administrators, teachers and other support personnel will go above and beyond the call of duty when it comes to making sure that your child receives the right kind of support to become a successful student. On the other hand, if the school will not adapt the curriculum or learning environment to meet the needs of your child, there are other steps you can take.

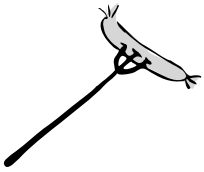
If your child is experiencing developmental delays due to their condition, you can request an Individual Education Plan (IEP) tailored to meet the needs of your child. There are several steps involved in this course of action, but in the long run your child's needs will be documented and the school will be required by law to meet those needs.

If your child's needs require different attention and are still not being met by the school personnel, you can set up a meeting to discuss Public Law 94-142 and Section 504. This Federal Law requires that a child be educated according to "assessed needs." This means that an assessment must be conducted in a timely fashion, the parents must be included in the decision making process, and that the plan for correction of problems found (if any) must be documented in writing and implemented promptly. Section 504 of the Rehabilitation Act of 1973 allows modification of traditional programs to meet special needs of children. Selected portions from the act states "no otherwise qualified handicapped individual in the United States, as defined in section 706(6) of this title, shall, solely by reason of his handicap, be excluded from the participation in, be denied the benefits of, or be subjected to discrimination under any program or activity receiving Federal financial assistance or under any program or activity conducted by any Executive agency or by the United Postal Service….(B) a local educational agency (as defined in section 198(a)(10) of the Elementary and Secondary Education Act of 1965), system of vocational education, or other school system."

Your child deserves the best education possible. There are laws established to protect your student in the school system. As a parent, you need to ensure the most nurturing, profitable learning experience for your child. Don't be afraid to exercise your rights!

Every school should have a copy of the 504 law available to the public. You can also log onto http://www.mindspring.com/~ncatp/p4.htm or http://www.thompson.com/tpg/fed_gts/hand/hand for more information.

CAMP HAPPINESS



by Lydia Preston

CAMP HORIZON OPENS on a warm Sunday in August. Cars pull into the campground in the rolling farmland of central Pennsylvania, unloading kids, backpacks and bulky duffel bags in the parking lot. The newcomers hang back, clinging to their parents; returning campers race off with barely a backward glance to join friends from previous summers.

It's a typical first day at any summer camp, anywhere. But many of these children would not feel at home in other camps. Some are swathed in bandages. Other kids are completely bald. Some walk with short, painful steps; a handful are in wheelchairs. And most of them have some kinds of rash, inflammation or growth on their skin. This is the tie that binds them, and what makes Camp Horizon unique.

"When people hear about this place, the first thing they often say is 'Why do you need a camp for that? It's only skin. How bad can it be?'" says Julie Winfield, M.D., a pediatric dermatologist from Minneapolis, who is part of the all-volunteer camp medical staff. "Well, it can be really bad. Some skin conditions are very disabling - they can even be fatal - and they are all socially devastating. For some of these kids, camp is one of the few places on Earth where they're not made to feel like total outcasts."

"Kids at school call me 'Fish' - you know, because I've got scales," says twelve-year-old Vicky Murray, a pretty, blue-eyed brunet who has had severe psoriasis since she was a toddler. "They stare at me when I walk down the hall, and if I touch something, they won't touch it."

"They laugh at me, and they call me 'Mummy' because of the bandages," says Lorien Johnson, thirteen. Lorien has a very rare form of the genetic condition called epidermolysis bullosa (EB), which makes the skin extremely fragile. The slightest pressure can raise a painful blister or open a dangerous wound, so Lorien must wrap herself from head to toe daily with protective gauze dressings. Accumulated scar tissue has fused her fingers into mittenlike fists, a common consequence of her type of EB.

"When people first see me, they go, 'Whoa! What's that?'" says Chris Overstreet. The thirteen-year-old was born with ichthyosis, a genetic disorder that causes the body to produce skin cells faster than they can be shed. In its most severe form, ichthyosis can render the skin as stiff as an alligator's hide. In Chris's case, large brownish plates of scale give his skin a rough dappled appearance.

"When we go someplace like a mall or an amusement park, the way people point and stare is obnoxious," says his mother, Sue Overstreet. "It's so hard to watch - to see your child try to ignore something like that."

"The worst thing for me," adds Chris, "is knowing that I was born this way and there's nothing I can do about it. People seem to think there's some cream or medicine I could use that would make it go away. That really ticks me off."

ISOLATING DISEASES

There is no real cure for any of the conditions that affect these campers. Just over half of the eighty children have rare genetic disorders, for which only modern genetic engineering offers some possible hope of a future remedy. The others have severe cases of more familiar skin ailments. Some 6.4 million Americans - about 10 to 15 percent of them under the age of ten - have psoriasis, and an estimated 10 percent of all children have atopic dermatitis, the worst form of the chronic rash-producing disease eczema.

For reasons doctors still don't fully understand, in some patients the diseases rage out of control, enveloping large areas of their bodies for weeks or even years.

Often, these children are prone to serious infections. They may not be able to sweat efficiently, which can be life-threatening in hot weather. Some disorders, like EB, cause internal scarring, hampering the ability to eat. For children with excessively fragile or stiff skin over their joints, just holding a pencil can be agonizing. Some find it too painful to walk.

Only a tiny percentage of all children with dermatological ailments are so severely affected. But, adds Amy Paller, M.D., professor of pediatrics and dermatology at Northwestern University's Children's Memorial Hospital, in Chicago, "That's one of the reasons a camp like this is so important. Most of these children never meet anyone else like themselves.

Photo courtesy of David Graham © '98

Children with skin diseases live with teasing, stares and loneliness. But here, they can forget about being patients and just be kids.

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EKV continued

function. Mutations in two EKV families resulted in changes of codon 12 replacing the invariant glycine to a positively charged arginine (G12R), or to a negatively charged aspartic acid (G12D). These changes affecting the amino terminal domain of Cx31are likely to interfere with the normal assembly or gating function of the protein. The third missense mutation replacing cysteine 86 with serine was identified in 2 unrelated families, and occurred in the conserved second transmembrane domain of Cx31 known to be critical for the regulation of voltage gating. Neither mutation was detected in unaffected family members and198 Caucasian control alleles, confirming that these mutations are the proximal cause of EKV in these families. Our data published in the prestigious journal "Nature Genetics" in December of 1998 (Richard et al., Nature Genetics 20: 366-369, 1998) constitute the first report linking a human skin disorder to mutations in a Cx gene. The finding of mutations in GJB3 suggests that the clinical manifestations of EKV can be caused by impaired gap junctional intercellular communication due to a defect in gap junctions. They also emphasize the critical role of Cx for proper epidermal differentiation and response to external factors. However, about 2/3 of all EKV families as well as several patients with PSEK do not carry identifiable mutations in Cx31. These results suggest that there is yet another gene whose mutations could give rise to EKV and possibly PSEK, which we are currently investigating in my laboratory. In addition, current and future studies aim to elucidate the nature and effects of known mutations in GJB3. We use different experimental approaches to study the influence of specific Cx mutations on gap junctional intercellular communication in different cell types. This research will have major implications for our understanding of the specific functions of gap junction communication in skin and the pathomechanims leading to the disease.

We like to use this chance to thank all EKV families for their generous participation in our studies, and all physicians, the Foundation for Ichthyosis and Related Skin Types and the National Registry for Ichthyosis for their continuous support and patient referral. We continue to solicit additional patients and families with EK for our studies. If you would like to participate, I can be contacted at the following address: Gabriele Richard, M.D., Department of Dermatology and Cutaneous Biology, Thomas Jefferson University, 233 S. 10th Street, BLSB, Suite 409, Philadelphia, PA 19107 (Tel: (215) 503-8259, Fax: (215) 503-5788, e-mail: Gabriele.Richard@mail.tju.edu)

In Loving Memory...

Our sincere sympathies to the family of Floral Emerson who passed away on August 10, 1999 from congenital heart failure. Ms. Emerson was affected with Epidermolytic Hyperkeratosis (EHK) as her daughter Reba, and granddaughter Becca. They have all been members of F.I.R.S.T. for many years. The family requested all donations be sent to F.I.R.S.T. and the foundation received over $600 in her memory. Our sincere thanks to Ms. Emerson's family for thinking of F.I.R.S.T. during this most difficult time.

Thanks to Generosity of Anonymous Donation, Membership Assistance Fund to be Created

The Foundation for Ichthyosis & Related Skin Types (F.I.R.S.T.) is pleased to announce a Membership Assistance Fund will soon be available for applications from F.I.R.S.T. members. This fund was made possible by the enormous generosity of an anonymous donor who contributed $10,000 to form an endowment. The interest from this endowed fund, along with a second contribution of $1,300 from the same donor will be used for direct support of F.I.R.S.T. members in need of financial assistance. F.I.R.S.T. will begin accepting applications in the new year. Further details of the application process will be included in the next issue of Focus.

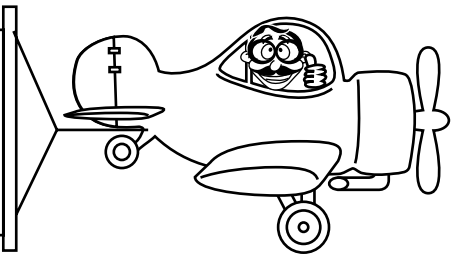
If you are interested in making a donation to help build the Membership Assistance Fund, please contact Jean Pickford at the F.I.R.S.T. office.

Our sincere gratitude to the anonymous donor for your extreme generosity. It is people like you that help make our world a better place.

SAVE THE DATES!!!

2000 Family Conference

July 7, 8 & 9, 2000
The Park Ridge at Valley Forge
King of Prussia, Pennsylvania



Hotel Accommodations
480 North Gulph Road, King of Prussia, PA 19406
610.337.1800 Reservation Desk, 800.337.1801Toll free

For hotel information and reservations, call one of the above numbers and identify yourself as a member of F.I.R.S.T. to receive our special conference rate of $80.00/night, any occupancy. This rate does not include 6% Pennsylvania sales tax and 2% local room tax. This rate is guaranteed from July 10 through July 17. All reservations must be canceled by 4:00 p.m. on the day of arrival in order to avoid a no-show charge of one night's room and tax. Check-in time is 3:00 p.m. and check-out time is 1:00 p.m.

This special conference rate of $80.00 is only guaranteed for rooms booked a minimum of one month prior to the conference and you must call during EST business hours to make your reservation. If you have any questions or experience any difficulty, please call the F.I.R.S.T. office at 800.545.3286.

Airline Reservations, Airport Shuttle and Agenda Information have not been finalized yet….so look forward to seeing more information in the next issue of Ichthyosis Focus.

REGISTER EARLY!

Name: _____

Address: _____

City: _____ State: _____ Zip: _____

Phone: (Day) _____ (Evening) _____

Attendees	Adult	Child	Age	Check if affected with ichthyosis
_____	❑	❑	_____	❑
_____	❑	❑	_____	❑
_____	❑	❑	_____	❑
_____	❑	❑	_____	❑
_____	❑	❑	_____	❑
_____	❑	❑	_____	❑

Type of ichthyosis, if known: _____

Number of adults _____ x $95 per person = _____
(18 & older)

Number of children: _____ x $50 per person = _____
(1 through 17)*

TOTAL = _____

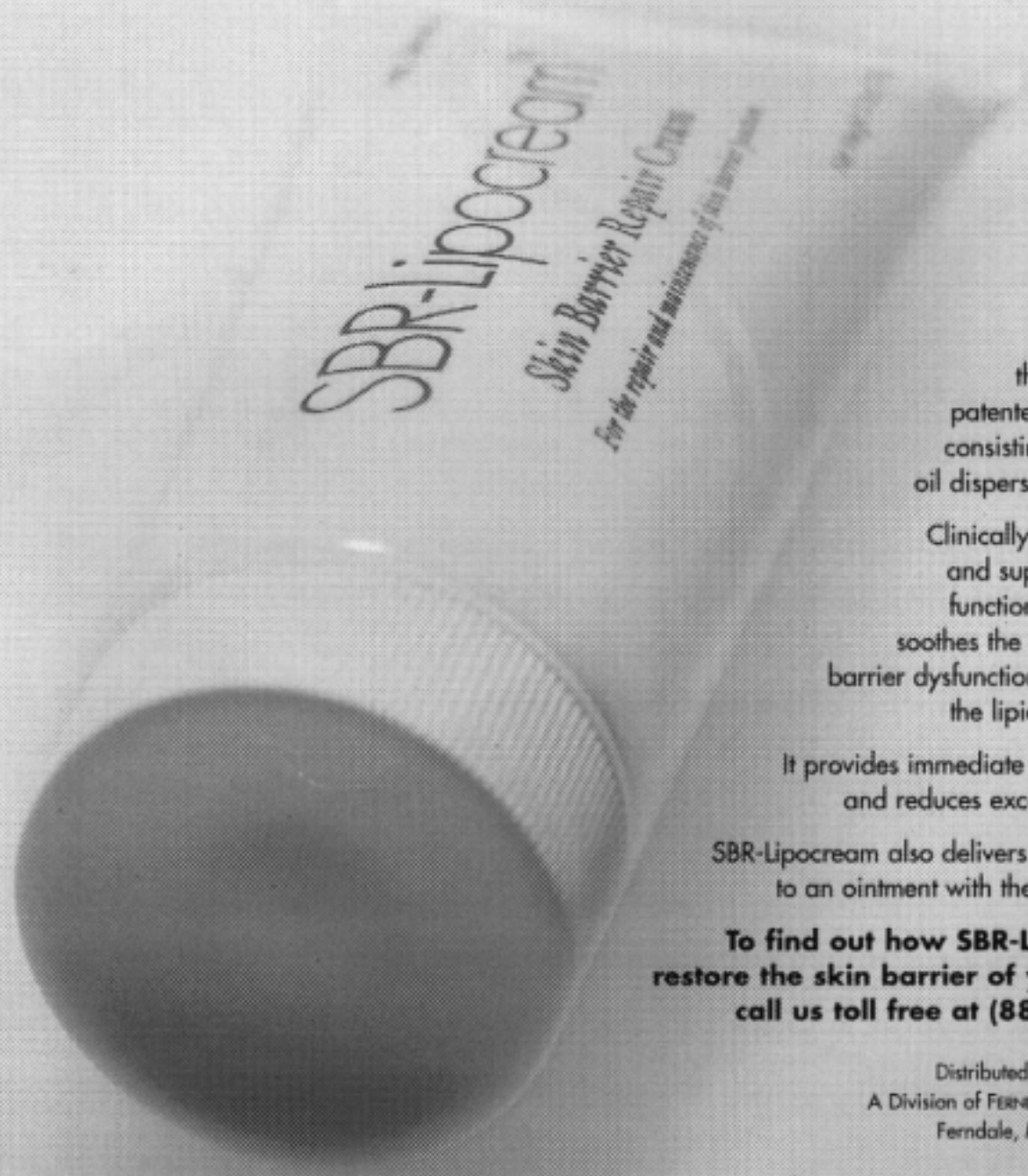
*There is no registration fee for children under one year of age

Please return registration fees with complete form to Jean Pickford, F.I.R.S.T., 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446. Kindly make checks payable to F.I.R.S.T. in US funds.

Cancellations will be honored with full refunds until Advance Registration Date of Wednesday, June 14, 2000.

SBR-LIPOCREAM®

The Power to Restore and Maintain Healthy Skin Barrier Function



SBR-Lipocream therapy features a patented, scientific base consisting of nearly 70% oil dispersed in 30% water.

Clinically proven to repair and support skin barrier function, SBR-Lipocream soothes the symptoms of skin barrier dysfunction by replenishing the lipid film of the skin.

It provides immediate soothing therapy and reduces excessive water loss.

SBR-Lipocream also delivers occlusion similar to an ointment with the feel of a cream.

To find out how SBR-Lipocream can restore the skin barrier of your patients, call us toll free at (888) 548-0900.

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Ferndale, Michigan 48220 USA

CAMP HAPPINESS

continued from page 6

They feel so isolated and lonely."

Camp Horizon opened in 1995 outside rural Millville, Pennsylvania, under the direction of Howard Pride, M.D., a pediatric dermatologist at Penn State Geisinger Medical Center in nearby Danville. It was inspired by a similar facility, Camp Knutson, in Minnesota. Today, both weeklong skin-disorder camps operate under the aegis of the Camp Discovery program of the American Academy of Dermatology, which provides funding and whose physician members refer young patients from throughout the U.S., Canada and elsewhere. There is no cost to the children's families.

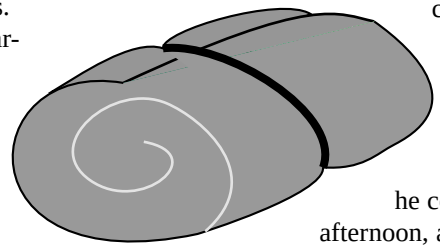
The eight- to thirteen-year-olds at Camp Horizon bunk in eight roomy cabins, and are divided into groups of four to six who eat and participate in scheduled activities together. From the start, camp veterans Chris Overstreet, Matt Sweeney and Alex Khauslender, all thirteen, unite in a rowdy posse - ribbing their two counselors, checking out the girls in the mess hall and happily plotting water-balloon fights. Alex has alopecia, a type of hair loss that comes and goes without warning. Last summer, he wore a baseball cap to cover his bald patches; this year, he sports a full head of dark hair.

The fourth in their group, thirteen-year-old newcomer Anthony Discepola, is reserved and wary on the first day. That afternoon, while the other boys race off to the pond, Anthony heads for the Med Shed, which houses both the camp clinic and the office. His psoriasis is acting up - his arms and legs, covered with angry red skin and silvery scales, itch like crazy.

It's hard to predict what Anthony's future holds. In general, the more severe an inflammatory skin condition is in childhood, the more likely it is to be a lifelong problem. Experience has left Anthony with little faith in doctors - "They can't do anything," he says dismissively - or in alternative therapies: "I've tried, like, every cream and medicine in the world. Nothing ever lasts."

Scratching his arms, Anthony carries on a glum conversation with a counselor. Why did he come to camp? "I dunno." What's he looking forward to here? "Not much." What does he like to do with his friends back home in Atlanta? "I really don't have any friends," he says curtly. His mood picks up after dinner, though, as he is drawn into a punt, pass and kick competition, displaying an impressively strong and accurate throwing arm. Later, he joins the other kids for a raucous evening of get-acquainted games, followed by bingo and a bedtime snack of s'mores.

On Monday morning, Anthony becomes completely absorbed in a knot-tying class. At the heavily stocked fishing pond, he proves adept at fly casting, too, and he enthusiastically joins in the mock-betting at "Casino Night" that evening. "It's actually pretty cool here," he concedes. Tuesday afternoon, at an advanced knot-tying class, he becomes the first Horizon camper ever to master a complicated eye splice weave. At dinnertime, wearing the rope slung across his chest, Anthony stands on the mess-hall stage, beaming, as his feat is announced, The campers, led by Chris, Alex and Matt, cheer.



"This place is like heaven on Earth for me," Anthony declares later, with a broad smile.

A PLACE TO FORGET THE PAIN

The breakneck round of activities at Camp Horizon is the typical mixture of sports, crafts and outdoor skills: swimming, paddle-boating, fishing, archery, tie-dyeing, woodcraft and the like. After dark, there's stargazing, campfire songs and the option of sleeping out in the woods. Even the most severely disabled campers are never left on the sidelines. Those who have trouble walking are ferried in golf carts; softball, volleyball and basketball games slow to a snail's pace for kids who can't tolerate rough play.

For the most part, tedious daily skin care is unobtrusively woven into camp routine. The children with EB check into the Med Shed daily for dressing changes, and those with feeding or tracheostomy tubes are tended by medical personnel. Counselors keep a sharp eye out for children who might be overheating, especially those with ectodermal dysplasia, a condition in which the skin has no functioning sweat glands.

Most of the kids appear to be blissfully oblivious to their conditions after the first day, although a few do find kindred souls. Ichthyosis patients Bailey Jones and Laura Ashton joke about vacuuming skin flakes off their beds and furniture every day.

On a more sober note, Laura wishes she knew how the texture of her skin feels to other people. "No one will tell me," she says with an edge of frustration in her voice. "When I ask my mother, she says my hands feel normal. But I know that's not true." She extends her hand. "Will you tell me what it feels like?"

Laura's hands seem a little dry, kind of work-worn. The plaquelike scales are waxy, probably due to the heavy creams she uses. "But they look so sharp," she says doubtfully. "Don't they hurt when you touch them?" No, they don't. Her mother is right - Laura's skin feels pretty normal.

Few people are willing to discover that for themselves. "Most skin diseases are not contagious, but people think they are," explains John Koo, M.D., a psychiatrist, dermatologist and associate clinical professor of dermatology at the University of California, San Francisco. "In general, that makes skin disease more stigmatizing than a handicap. If you're in a wheelchair, plenty of people will come forward to help. If you have psoriasis, they pull back."

That stigma extends to Camp Horizon, too - at least in the beginning. On the first day, kids accustomed to their own conditions recoil from campers with unfamiliar disorders. Counselors who expected to be caring for children with mild conditions are shocked at all the bandages, rashes and scaly skin. Some privately admit being reluctant to touch the youngsters or go swimming with them.

And yet, by Wednesday, kids and adults alike are jumping in the pool and hot tub and linking hands around the fire. "I don't even notice their skin anymore," marvels counselor Yale Han, a Bucknell University premed student.

FRIENDS FOREVER

At the end of the week, the atmosphere at camp is giddy. Friday, the last full day, features a rowdy after-supper talent show in which four performers stand out. Eight-year-old Michael Allen, who has the corn-silk hair and pale skin typical of ectodermal dysplasia patients, struts onstage shirtless and has the audience roaring with laughter at his cocky bodybuilder poses.

Josh Nelson, whose epidermolysis bullosa has left him small for his twelve years, zips up to the microphone in his motorized wheelchair and holds up some pictures he has drawn: "A scary fish; a Tyrannosaurus rex." He gets a standing ovation. Twelve-year-old Novelette Munroe, who also has severe EB, brings the audience to its feet again with an impassioned recitation of Lady Macbeth's "Out, damned spot" soliloquy. Finally, Lorien Johnson, wearing a flowery dress over her bandages brings down the house with a rap number.

Later, as water-balloon-wielding campers party on outside, Novelette, Lorien, Vicky Murray and Vicky's friend Lauren Schwartz sit quietly in their cabin, reflecting on the week and their friendship. "What I like most about being here is that you can communicate with people who understand you," says Novelette softly. "You feel like you're not alone."

"I used to think I had it bad," says Vicky, reflecting on her psoriasis. "I complained all the time. But when you come here, you know what all these other kids go through, and you see they're happy. It gives me confidence. It makes me think I can do stuff I didn't think I could do."

Lauren, who has a less severe form of EB than Lorien and Novelette do, agrees: "I like it here because people don't look at you and go, 'Eewww.'"

What would they like to say to their tormentors? Novelette replies, "I'd say that we're different on the outside, but not on the inside." Vicky has no suggestions. "People don't care," she says. "If they don't like your looks, they don't care what you are or what you can do."

The group spends more than an hour exchanging stories, drying tears, comparing notes and comforting one another. Finally, counselor Kathryn Zeoli, M.D., a dermatologist from Fort Lauderdale, Florida, interjects gently, "But you feel happy and comfortable here."

"Yes," agrees Novelette. "I don't feel like anybody's staring at me."

"They get used to you real fast here!" says Zeoli. They laugh, the grim mood broken. The girls joke, hug, exchange addresses and vow to stay friends forever.

Once again, Camp Horizon seems like any other summer camp, on any other final night.

Lydia Preston is currently working on a book about the physical and psychological effects of acne.

News & Notes

Online Genetics Glossary: The National Human Genome Research Institute (NHGRI) at the National Institutes of Health provides an online multimedia glossary appropriate for professionals and the general public. This glossary contains user-friendly, downloadable explanations of genetic terms. A limited number of CD versions are available to educators and libraries free of charge. Visit http://www.nhgri.nih.gov/DIR/VIP/Glossary.

Reprinted from Alliance Alert, September 1999, a publication of the Alliance of Genetic Support Groups.

Medical Privacy & Discrimination: The Ganske Amendment: The House of Representatives passed an amendment offered by Representative Greg Ganske (R-IA), which was added to the recently passed banking reform legislation (HR.10). Although the law has nothing to do with health, the Ganske amendment will allow all financial institutions (banks, insurance companies, brokerages and lenders) to share or sell personal medical information about customers without their knowledge or consent. This would allow a bank, for example, to delve into confidential medical information when a person applies for a car loan or mortgage.

After the amendment passed on a party line vote, it caused significant controversy. The President "strongly urged" that it be stricken from the banking bill, and the AMA protested loudly. Now the House and Senate will be meeting in a Conference Committee to iron out the differences between the House and Senate passed banking bills. Since the Senate (S.900) does <u>not</u> contain the Ganske amendment, it is hoped that it will be deleted from the final bill. We urge NORD's member agencies to ask their Congressperson and Senators to delete the Ganske amendment from the banking reform bill.

Reprinted from NORD Online, July 1998, a publication of the National Organization on Rare Disorders.

Looking Ahead with the Support Network *Michele M. Bolles, CHES*

The Foundation for Ichthyosis & Related Skin Types' (F.I.R.S.T.) Support Network was formed to break the isolation and show that you are not alone! The Support Network can serve to provide resources for information, emotional support, practical advice, guidance, and understanding. The volunteer peer counselors know, care and can give emotional support to families and individuals affected by ichthyosis.

Currently, the Support Network is made up of eight regions throughout the United States. A Regional Coordinator and a host of volunteer peer counselors manage each region. By contacting F.I.R.S.T's Support Network program you are starting the process that matches you with a peer counselor. When your inquiry is received, F.I.R.S.T. refers your contact information to a Support Network Coordinator and a "match" is made. A "match" is based on many factors including the profile of the individual/family, type of ichthyosis and geographic area. The coordinator then forwards the name and contact information to the volunteer peer counselor. The volunteer counselor will be in touch with you to assist in addressing your questions or concerns. They can give practical advice and provide information on other available resources. Many times, an individual may just need to talk to someone who has "been there." Knowing that you are not alone can make all the difference in the world.

F.I.R.S.T.'s Support Network is made up of members who know first-hand how stressful and frightening the days ahead may be. They have been down the same path and want to share the knowledge and experiences that guided them through their journey. Remember, these are people just like you who can offer support to others experiencing similar concerns.

Over the next year, F.I.R.S.T. will be working to make an already successful program even better! Volunteer peer counselors will be getting newly developed training materials, including educational handouts and helpful resource information. Additionally, we're planning to coordinate information sharing among our volunteers to highlight the invaluable lessons learned from our members. F.I.R.S.T. also sees the Internet as a wonderful tool to help support our members. In the past, many were only able to interact via phone or mail, now the Internet makes the world a little smaller and helps people to feel much less isolated.

If you would like to become part of the Support Network or know someone who would make a great Support Network counselor please contact us at (800) 545-3286 or email at ICHTHYOSIS@aol.com.

Ichthyosis Awareness Week

October 3 - 9, 1999

Thanks to the following members for their generous efforts in promoting awareness about ichthyosis. Through our strong membership, F.I.R.S.T. continues to raise awareness about the disease and the people it affects.

Thanks to...

Ann Minarick .....for volunteering clerical assistance at the national office

Barbara Molica.....for writing to the Oprah Winfrey Show

Dan Siegel .....for selling tickets for local fund raiser

Denise Benedetto .....for conducting the "Butterfly Fund Raiser" at her school where she teaches

Dona & Tracy Hoar.....for conducting the "Butterfly Fund Raiser" and distributing brochures in their local community

Donna Rice.....for writing an awareness letter about her child's school

Dorothy Cordle.....for writing to her congressmen asking for more research money

Elena Levitan.....for selling tickets for local fund raiser

Francis & Albina Dunleavy .....for selling tickets for local fund raiser

Jeff Gerber.....for organizing a United Way campaign at work

Lisa & Christopher Short .....for selling tickets for local fund raiser

Liza Santamina.....for running in the Human Race

Lori Florian .....for organizing a garage sale

Lynne Alba .....for helping plan the 2000 Conference and selling tickets for local fund raiser

Nicole & Grace McMillian .....for selling tickets for local fund raiser & working the door on the day of the shopping benefit

Pat Mateson.....for selling tickets for local fund raiser

Frank Meli.....for donation from Unico National

Susan Suda .....for writing an awareness article to major newspaper

Tracie Pretak .....for conducting an annual Spiritual Concert with her daughter and distributing butterflies to other volunteers

Tracy Garcia .....for conducting the "Butterfly Fund Raiser" at two school carnivals

Spotlight On...

Shelby Riggs & Lori Florian

Shelby Riggs was born with Lamellar Ichthyosis. Her mom, Lori, remembers the nurses saying "your baby is just not normal." While in the intensive care unit, Lori was able to get the diagnosis through a skin biopsy. Since this disease is so rare, the majority of the doctors had not heard of it, much less how to treat it. So the first year of Shelby's life was a learning period for her family and doctors.

Shelby was about four months old when her family received a pamphlet about F.I.R.S.T. Two months later, Shelby and her family attended a national conference in San Diego. Shelby was the youngest one there. Lori learned more about this disorder from other parents raising children with ichthyosis than she did from any of Shelby's doctors.

"Comments made by the general public are the hardest part of dealing with Shelby's skin disorder," Lori says. "We rarely are able to go shopping or just for a walk down the street without comments or stares. So we are learning how to deal with them better. As Shelby gets older, she will have a better understanding of her skin and be able to explain it on her own. For now, we are in the process of getting business cards made, and they will say 'I have ichthyosis, a genetic skin disorder and any further information can be obtained by contacting F.I.R.S.T.'" Lori also had the editor of their local newspaper write two articles about Shelby. She had them written to inform her community about Shelby's skin disorder because people in her town would call the police department accusing Shelby's family of leaving her out in the sun too long.

Lori Florian and Shelby Riggs

Today, Shelby is 4½ and having a great time! Shelby loves playing with her older half sister Shauna, riding her bike, roller-skating, riding horses, jumping on her trampoline and watching Disney movies. She has been dancing for two years, and each May she participates in a dance recital. Shelby attends their local elementary school where she is enrolled in the early intervention pre-kindergarten class.

Shelby surrounds herself with tons of friends. Besides her mom, Shelby's father, Tom Riggs is her biggest protector. Tom loves to play with her and treats her like a little princess. This past summer, Shelby and her mom went to live in northern California to be closer to Tom, while he worked a job in Truckee. Together they enjoyed the mountain scenery, hiking and fishing. If you would like to contact Shelby, Lori, or Tom, please e-mail them at LFO456@AOL.COM.

If you would like to submit an article about someone you think we should spotlight in an upcoming issue of Focus, please feel free to send it to us. We always love to receive success stories from our members.

LET'S FUND RAISE!

F.I.R.S.T. is very committed to raising more funds for our programs and services. We need to make contact with our members to establish more leads for fund raising opportunities. You may not realize that your company, benevolent organization, social groups, etc. would be willing to make a donation to F.I.R.S.T. Making the request can sometimes be very difficult and uncomfortable, so Jean Pickford, our new executive director, would be willing to help you. Please complete the following questionnaire and Jean will contact you to discuss how we can work together to fund raise.

Please keep in mind that in-kind donations are greatly appreciated too. An in-kind donation includes items for gift give-aways at the Family Conference, office supplies, printing, marketing/ public relations or accounting services, etc.

Name: _____

Address: _____

City _____ **State** _____ **Zip** _____

Phone _____ **Email** _____

❑ I would be happy to deliver a proposal to my company/organization requesting funding for F.I.R.S.T.

❑ I work for a company who donates to charity, but am uncomfortable asking for a donation. I would be happy to put Jean Pickford in touch with the right people.

❑ I have some ideas for an in-kind donation.

❑ I would like to do my own fund raiser in my local community but need ideas and/or guidance.

The Ichthyosis Registry Needs YOU!

by Geoff Hamill, R.N.

Hello to everyone at F.I.R.S.T.! I just got back from another great week at Camp Discovery, and it reminded me of how important it is that the Registry continues its work encouraging research into diagnosis and treatment of the ichthyoses.

We are in our fifth year, and have assisted investigators like Dr. Sherri Bale (NIH), Dr. Gabriele Richard (TJU), Dr. Matthew Stiller (Harvard), and Dr. Peter Elias (UCSF) in their studies of different types of ichthyotic disorders. We would not have been able to assist these investigators without the participation of many of you who have taken the time to enroll in the Registry. It's really a valuable resource we have created for finding answers to the questions about how these problems occur, and what can be done to try and help, treat, or fix them. BUT YOU ARE THE RESOURCE!

I'm hoping you will call me (or e-mail, write, fax, send smoke signals, whatever!) and enroll in the Registry. Please enroll yourself or encourage your family, your friend, or whomever you know that is affected. If you're not sure if you have enrolled, do the easy thing and call! The more people that enroll, the better a resource we have to offer to promote research. It's as simple as that. Thanks for all of your support and participation. Call me anytime, I'll be here! Geoff Hamill, R.N.

P.S. from the Registry Secretary:

Hi! This is Kim. Some of you know my voice as the fellow who leaves friendly messages on your answering machines. Part of my job is to gently remind folks that they need to tidy up a loose end or two to complete their enrollment. If I call you, it doesn't mean you've been negligent or naughty (we know that daily life and things like childcare, work, and other things take time). I'm just calling to let you know that we care about you and developing the resource that is the Registry. Geoff and I are really friendly, nice, and helpful guys who look forward to speaking with you on the phone. We hope to answer your questions and help you to complete your enrollment. (Remember, to enroll all you need to do is: Sign your consent; have a dermatologist fill out a short evaluation and send it in; and call to let us know when it's convenient for you to talk with Geoff on the phone. Once the two of you have chatted, that's all! You're enrolled! See - it doesn't hurt a bit!).

Like Geoff said, call us. We really would love to hear from you. Kim Pineda

Share your story in an upcoming book

Mark Elzey of Treehouse Publications is trying to put together a book of inspirational stories about people with a facial disfigurement. Mark is doing this book to bring awareness to the general public and to dispel some common misconceptions about facially disfigured people. There is no book on the market today that provides inspiration to families and individuals affected by a facial disfigurement. In addition, a film documentary is being planned to complement the book and some of the people profiled in the book will be profiled in the documentary.

Mark needs stories of people whose lives have been affected by a facial disfigurement. If you have a story or know of someone who has a story that will inspire others please send it in (remember: it doesn't have to be a literary masterpiece)! Keep in mind, if you share your experiences, the story could make the road a little smoother for the next person. They will know that they are not alone.

Submission Guidelines

1. Stories must relate to facial disfigurement and the constructive way that some people, with or without a facial disfigurement, have helped.
2. Stories must depict courage, unwavering support, or any upbeat testimonial that you feel needs to be shared.
3. Submit stories on a voluntary basis only. There will be no compensation for story submissions.
4. After your story has been accepted, you must sign a release form giving Mark the copyrights to this story.
5. If your writing skills are not up to par, you may be a little apprehensive about sending a story. Please don't worry! Mark or one of his editors will work with you. Together you will develop the story. Just send in the best story you can!
6. You will receive credit as the author.
7. All stories must be based on true events.
8. Please keep stories under 5,000 words.
9. Please include your name, address, phone number, and email address. It is critical that someone is able to get in touch with you.

You can submit stories by mail to Treehouse Publications, c/o Mark Elzey, P.O. Box 79, McDanials, KY 40152-0079 or email at TreehouseStories@aol.com. If you want to contact Mark for more information about this project, please write to either of the addresses listed above or call (317)848-4947.

RESEARCH STUDIES

Study for Multiple Family Members with Ichthyosis Vulgaris
Researchers at the National Institute of Arthritis and Musculoskeletal and Skin Diseases/NIH in Bethesda, Maryland, in collaboration with researchers at the University of Washington, Seattle are continuing their efforts to understand the genetic basis of Ichthyosis Vulgaris. Families in which multiple members have Ichthyosis Vulgaris are eligible for study. Please contact Dr. Sherri Bale at NIH (301-402-2679; you may call collect) if you and your family are interested in participating. All expenses for participating in the study, including travel and hotel costs, will be covered.

Clinical Delineation of the Keratitis-Ichthyosis-Deafness Syndrome
Principal Investigator: Dr. Ethylin Wang Jabs
Johns Hopkins University School of Medicine

Investigators: Dr. Laura Russell
National Institutes of Health

Dr. John DiGiovanna
Brown University

Dr. Peter Elias
University of California, San Francisco

The Keratitis-Ichthyosis-Deafness syndrome is a rare inherited disorder in which affected persons have 1) deafness present at birth, 2) gradual destruction of the cornea of the eye, possibly leading to blindness, 3) localized areas of disfiguring reddish skin thickening, and 4) thin or even absent scalp hair.

The investigators listed above seek to recruit patients with KID syndrome for a clinical study that will define different subtypes. These studies will be performed at no cost to the patient and their results will be shared with the patient.

If you have KID syndrome and might be interested in participating in this study, please contact: Dr. Laura J. Russell 514-934-4427

Netherton Syndrome, Ichthyosis Linearis Circumflexa, and Peeling Skin Syndrome
We are studying the clinical spectrum of Netherton Syndrome, Ichthyosis Linearis Circumflexa and Peeling Skin Syndrome, and try to elucidate their molecular basis. We are looking for single cases as well as families with multiple affected persons for clinical examination and genetic studies.

The studies are conducted on a collaborative basis between the Thomas Jefferson University, The National Institutes of Health, and Brown University/Rhode Island Hospital. Patients diagnosed with these disorders, who are interested to learn more about, or participating in our studies, please contact Dr. Gabriele Richard at the address below:

Please contact: Gabriele Richard, M.D., Department of Dermatology and Cutaneous Biology, Thomas Jefferson University, 233 S. 10th Street, BLSB, Suite 409, Philadelphia, PA 19107 (Tel: 215-503-8259, Fax: 215-503-5788, e-mail: Gabriele.Richard@mail.tju.edu)

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