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

ICHTHYOSIS FOCUS

Vol. 21, No. 2 | A Quarterly Journal for Friends of F.I.R.S.T. | Summer 2002

Current Ichthyosis Research

Ichthyosis diseases result from mutations in genes that encode structural proteins or enzymes that are needed to build a normal epidermis on the outermost layer of our skin. Every one of us has two copies of each gene (one gene copy per chromosome) in all of our cells, including epidermal cells. Some diseases arise when a mutation occurs in one gene copy. These are called autosomal dominant diseases, because the bad protein/enzyme made from one mutated gene interferes with the normal copy, and thereby destroys the function of an epidermal cell. Very often autosomal dominant diseases involve genes for structural proteins. An example is epidermolytic hyperkeratosis (the keratin 1 or keratin 10 genes). Other ichthyoses occur when both gene copies are mutated, so that the products from both copies are bad. Many of these are autosomal recessive diseases, and commonly involve enzymes. An example is lamellar ichthyosis (the transglutaminase 1 gene). We all have 23 pairs of chromosomes. Each chromosome carries hundreds or thousands of genes. And each gene consists of a code made up of strings of thousands of DNA nucleotides. When the cells in our bodies divide for normal growth, the entire chromosome and all of its DNA is copied. Even though the error rate of copying DNA is very very low, errors do occur. If copying errors occur in reproductive cells (a sperm or an egg), then there is a chance that a mutation will be passed along to our children. Probably each one of us carries several mutant genes caused by copying errors. Mercifully, many errors are 'silent', so that we live perfectly normal lives blissfully unaware of their existence. Very rarely however, the errors affect an epidermal gene, and so unfortunately, a new or 'spontaneous' occurrence of an ichthyosis disease appears simply due to a copying error in germ cells (the reproductive cells). Also, the natural gene copying process is so efficient that defective genes from our parents are faithfully but regrettably copied and can be passed along to our children. Based on the simple chance of which gene copy is passed along, the possibility of inheriting an autosomal dominant disease from our parents can be more than 50%, and an autosomal recessive disease, 25% or more. These germline mutations will affect every epidermal cell over our entire bodies. That's why most of the skin is affected in an ichthyosis disease. Of course DNA copying errors also occur after fertilization in 'somatic' cells (all other cells in the body), but usually these somatic mutations are never noticed because the single sick epidermal cell simply falls off without affecting the whole skin at all.

Dr. Peter Steinert

Genes for making a normal epidermis fall into three types. The first are the specific genes, like keratins, transglutaminases, and many others. (Liver, bones, nerves, etc, each need hundreds of specific genes also, but they are different from epidermis-making genes.) Then there are other genes (we don't have any idea how many yet, but it may be hundreds) that are used to control, like traffic lights, the actions of the specific genes. I bet that some of these controllers can become mutated too, resulting in an ichthyosis disease. Finally, there are "housekeeping" genes, which are used for routine non-specific stuff in all cells, like copying DNA, making proteins, shuttling things around inside our cells, gossiping with neighboring cells, etc. If there were a mutation in any of these, a cell will quickly perish.

continued on page 6

ICHTHYOSIS AWARENESS WEEK

October 6
October 13

See page 7 for more about how you can be involved in this important initiative.

Ichthyosis Focus

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

Correspondence Corner

Dear F.I.R.S.T.,

I would like to thank you very much for sending my son and I the Ichthyosis Focus. I very much enjoy reading it and find it very helpful.

I would also like to say thank you for sending me a free sample of SBR-Lipocream to try out. I am willing to try anything that could help my son. Thomas's skin is still very bad; new problems pop up all this time. Over the last six to eight months his skin has started blistering for no reason. He also goes through a stage where the skin on the whole palms of his hands, and soles of his feet, peels off. Thankfully it doesn't hurt him, but it doesn't look very nice.

Thomas's condition changes all the time. That is why I want to thank you. I am very grateful for any help or information you can offer me. I will do anything to make things a little better for him. Thomas is my special little hero.

Yours sincerely,
Amanda & Thomas O'Dea, Swansea, Wales, UK

Dear Friends:

I am looking for solutions to Darier's Disease, in Australia. I am searching for solutions to severe constant itching (caused by any condition) which makes thinking, concentrating, and therefore, work, almost impossible. I would also like to find doctors, or naturopaths, or any health professional, who understands the very severe, psychological repercussions of such a condition.

If you have any helpful information, please contact Carla, at ckosi@hotmail.com. Please put "Re: Darier's" in the subject line. Or in Australia, call 0438 640052 (Brisbane) and I will call you back.

Carla Kospartov, Queensland, Australia

Dear Friends,

The World Association for Children and Parents (WACAP) is a non-profit, child assistance and adoption agency. Over the past twenty-five years, we've found permanent, loving homes for 7,400 children in need.

WACAP is currently seeking a family for an international adoption of a little girl diagnosed with Ichthyosis. She is two years old, and is described as very sociable and friendly. She has a good appetite and particularly enjoys eating chicken, and chocolate pudding! She is doing remarkably well with the care that she is receiving. She understands what is expected of her and has extremely good manners. She understands what is said to her and gives replies, using sentences of four to five words. This cheerful little child readily poses for photographs and thanks you when you're done. She receives daily treatments for her skin, although her vision has been impacted by her condition.

While doctors expect her vision to return to normal, she desperately needs a family who understands her special needs. We are looking for any and all avenues in which to advocate for this child. For more detailed information this child, please contact WACAP's Family Finders Project at 206-575-4550 / FamilyFinders@wacap.org or visit our website at www.wacap.org.

Thank you for helping us make a better world . . . one child at a time!

Sincerely,
Karen Halverson, WACAP Family Finders Project

continued on page 3

The National Ichthyosis Registry Needs You

The editors of The Focus regularly tell you, our members and readers, that it is important for you to register yourselves or your families with the National Registry for Ichthyosis and Related Disorders. Enrolling in the Registry offers you a number of personal benefits as well as benefits to the whole 'ichthyosis community.' Some of those benefits include: confirmation or clarification of your diagnosis; genetic testing and counseling; and promoting the understanding of ichthyosis, and how it affects individuals with the disease, in the medical and research communities. The Registry's connection to the medical and research communities is particularly important right now. Why? Because this year our government made a big commitment to rare disease research with the drafting of two critically important laws that are companion legislation to The Rare Diseases Act of 2001, and with the passage of the Loan Repayment Program.

The Rare Diseases Act of 2002 will create an official Office of Rare Diseases at the National Institutes of Health, which will promote and coordinate research on rare disorders. The Rare Diseases Orphan Product Development Act of 2002 will provide millions of dollars a year for clinical trials of new drugs, diagnostic tools, medical devices and medical foods for rare diseases. (See page 13 of this issue of The Focus for more information on these pieces of legislation.) Congress has already passed the Loan Repayment Program, which will award money to doctors to repay their student loans if they commit to participating in clinical research. These pieces of legislation are critically important to people with rare diseases who are waiting for new treatments and cures for their conditions. Passage of these laws will create an environment where, at the highest level of our government, research into rare diseases is considered important.

The National Registry for Ichthyosis and Related Disorders stands ready for this focus on rare diseases research with an extensive database of information from individuals affected by the ichthyoses and how they are affected, and a list of well-characterized individuals willing to consider being research participants. Please consider joining the Registry and helping to provide a valuable resource to investigators interested in studying and treating people with ichthyosis. To contact the Registry, call 1-800-595-1265, or email info@skinregistry.org or visit their website at www.skinregistry.org <http://www.skinregistry.org> .

Donor Recognition

The Foundation wishes to thank Dr. and Mrs. Eugene Van Scott for their generous contribution of $85,000. to the Foundation and it's mission. Dr. Van Scott has been a member of the Foundation for many years and is a current member of the Medical Advisory Board. Dr. Van Scott's concern for people with ichthyosis led him to develop the formula for NeoCeuticals Problem Dry Skin cream (PDS).

Correspondence Corner

continued from page 2

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

We would like to share Tiffany's healing with everyone. Tiffany was born with Congenital Ichthyosiform Erythroderma (CIE). This skin condition made her skin dry, red, and thickened in some places. We didn't understand the condition because no one in our family had dry skin. Dermatologists told us that it came from a recessive gene and Tiffany inherited this gene from both of us. The Foundation for Ichthyosis & Related Skin Types helped us understand the disease more, but we still knew there was no cure. We used Aquaphor on her skin from the time she was born, and bathed her in clear water. We never had any other children because we knew there was a one in four chance that another child could be affected.

When I learned that a member of my church was healed of leukemia, I began to believe that Tiffany could be healed too. On Sunday, February 24, we attended church and asked Brother Herb to pray for Tiffany. Brother Herb anointed my hand with oil. He explained that healing was process and that our faith was Tiffany's road to becoming a miracle. We kept the faith for Tiffany and every day her skin began to improve.

We no longer have to put Aquaphor on her skin and she is sweating, where before she couldn't. Both my husband and I have been deeply touched by this experience. We have witnessed a move of God's power and glorious works that we have never experienced before.

God Bless,
Vonnesia & Dwayne Brown, Bethpaige, TN

I am looking for other children with lamellar ichthyosis in Alabama. My five year-old granddaughter has this disorder and I would like her to meet other children that look like her. She lives in Demopolis, AL.

My name is Sandra Blair. If you are interested in having your child meet my granddaughter please contact me at the email address or phone number below.

Sandra Blair, Eutaw, AL
squirleyjean@oal.com
205-372-3754

Spotlight On...

PJ & Zach Rittiger

My Son Has Ichthyosis Vulgaris / This is what I tell him when he asks why?

By PJ Rittiger, copyright April, 2002 Pittsburgh PA

As I glance out the kitchen window, I note in an absent-minded way that it is snowing again. Not something unusual for southwestern Pennsylvania in February, which is sometimes colder than Anchorage. Zach, my nine-year old son, is pouring over his math homework and I am stirring a pot of pasta that will be turned into dinner before the hour is out. The house is cozy around the two of us and this pleases me in a boy-am-I-lucky sort of way.

"Mom, why do I have Ichthyosis?" Zach's question is sudden this afternoon but never wholly unexpected. He is at the age of reason and it is natural for him to question this particular subject endlessly. "You were born with it, Zach, and we don't know why some kids are born with Ichthyosis and most are not," I respond softly, turning to look at his upturned face, with his enormous, green eyes and yard-long lashes. He has heard this many times before but there it is again.

"Why can't a surgeon just cut me open and put little oil glands into my skin? "Probably because you'd end up with little cuts all over your body, which would be painful and then you'd also be at risk for infection and think of how long such a surgery would take and....." "But it could work, right? I mean if a doctor was willing to do that?" "I doubt it Zach."

Zach slams his pencil onto the table, splitting the point and scaring the dog. I sit down at the table and touch his cheek with the tip of my finger. "You're right. It stinks, but I can't lie to you to make you feel better, honey. There are other forms of Ichthyosis that are much harder to live with, than the form you have. I can only take care of you and teach you to take care of yourself," I say, wishing that neither of us had ever heard of Ichthyosis in any of its awful forms.

"Well, when is somebody going to find a cure?" he asks in his most demanding tone. "Why don't you grow up and find the cure? You're smart enough to find it for yourself." At this suggestion, Zach slams his math book shut, gathers his papers and stalks off to complete the homework in his room, where he will calm down. I return to my pasta and the falling snow that is now drifting through a dark winter sky. "Damn," I whisper to myself. "Damn, damn, damn!"

When Zachary was born on a blistering hot June morning in 1992, I knew that I had accomplished the most awesome task ever assigned to a woman by God, but I'd never heard of Ichthyosis. As a nurse placed my new son into my arms, I noted a small red patch on his plump, little cheek. I recall thinking that I'd heard that all newborns have gorgeous skin. What could that be, I wondered silently, touching the innocent looking patch? It would take me five years to get an actual diagnosis. By then, Zach was starting to take gym classes with his fellow Kindergarten students and I knew I had to get answers fast.

As the years passed, I took my son to a variety of pediatric specialists but no one, including the pediatric dermatologist who had come highly recommended, could tell us why Zach became over heated so easily or why his skin seemed to become inflamed almost without any cause. When Zach was about four years old, I started looking (in desperation) at exactly what all of his symptoms were. I also started taking a hard look at what was available to me on the market to correct these problems, but there was no single product that addressed all of the issues. Then, I got creative by mixing several over-the-counter products together to finally come up with what we now call "Mom's Cream." Although this is not something I'd ever advise someone else to use , it helped Zach a lot and we still use it; however, I still did not have answers on the issue of overheating. With a growing sense of urgency, I obtained an appointment with a well known pediatric endocrinologist, thinking that perhaps Zach's problems were hormonal. The pediatric endocrinologist examined Zach and declared him free of hormonal problems. As he was leaving the room, the doctor paused to give Zach's dry shin a rub. "We don't see much Ichthyosis, you know. That's what that looks like to me."

I made the endocrinologist write the word out for me. Two weeks later, I had a conclusive diagnosis of Ichthyosis Vulgaris, the mildest form of all of the Ichthyoses diseases. The new dermatolo-

continued

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gist walked into the exam room, looked at Zach's arms, shins and scalp and said: "Yep, that's Ichthyosis. What are you doing for it?" I told her about "Mom's Cream" and she told me to keep making it. The dermatologist also talked with me about over-heating and what we could do to prevent it, which was not much, especially since Zach was very opposed to using a cooling vest. OK, I thought, marching back to the car, after the appointment. Now, I know what I am dealing with. The next day, I hit the Internet and learned more.

Today, Zach is a good student, plays roller hockey, is an excellent public speaker, has taken karate, played Little League baseball, takes gym class regularly and goofs off as all nine-year-olds should with a bunch of neighborhood kids. To look at him, you would not guess that he has a problem, but I know what his skin can look like and feel like if I drop the ball. Every year I write a letter to each of his teachers alerting them to the fact that Zach has Ichthyosis and telling them what must be done for him if he appears over heated and I am not present.

Zach is now slowly learning to care for his own skin and take necessary precautions, which is a milestone. He is unconsciously admitting that he is a bit different from other children. This is a good thing because Zach's Ichthyosis is not going to ever go away. Someday, I will not be here but Ichthyosis goes on forever.

Ichthyosis has also made my son more empathetic to others. He understands discomfort better than a lot of kids his age. I have told him that I think he is one of the bravest people I know and he will need his courage to face unusual challenges that most people do not understand. He will also need to harness his anger and use it. I am not kidding when I urge him to find a cure himself.

"So, you don't think a surgery like that, where a doctor transplants oil glands into my skin, would work?" Zach asks me again, calmly probing once more at the idea of a fantastical, surgical cure.

The evening has worn on and we are now munching pasta that is loaded with tomatoes and garlic.

"Think of all those little incisions," I insist. "Maybe, there is a better way to fix Ichthyosis. What do you think?"

"I think someone should find a cure for this, Mum. That's what I think. How come nobody has yet?"

"Because I think you're the guy to do it and you have not grown up yet," I answer soberly, choosing the words like step stones across a pond. "Maybe none of the doctors researching Ichthyosis right now actually have it so you already know so much more than they do," I say hopefully.

"Yeah," he says quietly, "Maybe I do."

Current Ichthyosis Research

continued from page 1

Current basic research in ichthyosis diseases falls into three broad categories. The first is what I shall call 'discovery' research. Here, scientists are actively trying to discover all of the genes that are required to make a normal epidermis. When the first draft of the human genome sequence was published recently, we found out that there are large numbers of epidermal genes no one knew about before. Many of these are grouped together on chromosome 1 with known epidermal genes such as filaggrin and loricrin. At present no one has a good idea on what all these new guys do. So the goal of 'discovery' researchers is to try to figure out what they do. Are they always used in normal epidermis? Are they backups that are used when a normal epidermal gene is mutated? Do they protect us from the damaging effects of ultraviolet light and sunburn? Another aspect involves discovery of the nature and function of the genes that control the specific genes. Who are these? What do they do? When do they do it? You may have heard or read about DNA chips. These are tiny high-tech devices used to identify all of the genes used in a particular tissue during its growth and function. Several teams worldwide are working on epidermal genes. My understanding is that thousands of genes are used to make the epidermis. This great complexity will take time to sort out. Potentially, mutations in any one of these could cause disease, or offer a window of opportunity for treatment.

There have been great advances made just within the last 10 years in the identification of the genes that cause some types of ichthyoses. However, many still need to be solved, such as ichthyosis vulgaris, and many types of diseases grouped together as congenital recessive ichthyoses. Thus the second category of current research is to continue this work for all ichthyosis diseases. Some of them may be 'complex' diseases. Such diseases occur because there are mutations in more than one gene affecting the epidermis at the same time. Psoriasis is an example of a notoriously complex multi-gene skin disease. Once we know these details, physicians and scientists can design specific diagnostic tests. The third category of current basic research concerns what scientists are now trying to do to treat ichthyosis diseases for which the involved gene is already known. The favored approach by many investigators worldwide involves gene therapy. In principle, this is a simple idea: why not just replace the defective gene with a normal copy? In practice, however, this is very hard to do. Yes, we can clone a normal gene, but how do we effectively get it into the affected epidermal cells? Scientists have made progress with modified harmless viruses that can 'infect' cells with the normal gene. One difficulty with this method is that the epidermis is a constantly renewing tissue, so that cells would have to be infected with these viruses every few days. But epidermal cells grow from stem cells hidden deep inside the epidermis. Thus one hot research topic is to try to specifically find and infect the stem cells so that all new epidermal cells will always have two normal gene copies. Another problem we have to deal with in autosomal dominant ichthyosis diseases is that the product of the one bad gene copy is often enough to destroy the normal life of the epidermal cell. So, we have to find a way to switch off the bad gene, or switch off the cellular machinery that makes the bad protein. We already know that the 50% of protein or enzyme made from the one normal gene copy is usually enough for the epidermal cell to live comfortably. So scientists are hard at work and there are promising ideas on the horizon. One is the use of an artificial thing called a ribozyme. This is a small piece of synthetic RNA which can act like a specific knife. When all cells decide to make a protein from a gene on a chromosome sitting in the nucleus, the gene is first copied as a piece of RNA. This RNA is then decoded into a protein or enzyme. Ribozymes can be designed to specifically destroy a piece of RNA bearing an evil mutation. In theory, this will stop the production of a bad protein or enzyme, so that the other normal 50% can work without interference. A ribozyme to cure an occurrence of epidermolytic hyperkeratosis could be added to the skin in some sort of liposome cream, similar to what are used in cosmetics. A third approach is being tested for autosomal recessive enzyme diseases where both copies are bad. Scientists have learned how to make buckets full of normal epidermal enzymes, such as transglutaminase 1. The challenge now is to find how to resupply it back to the epidermal cells. Again, they are trying all sorts of liposomes to package the normal enzyme for proper delivery.

Many of us in the research community have committed our entire research careers to not only finding the genes at fault in various ichthyosis diseases, but also to finding the simplest and best way for effective treatment.

Peter M. Steinert
Chief, Laboratory of Skin Biology
Building 50, Room 1523
NIAMS, NIH, Bethesda MD 20892-8023

See page 14 for more on the Human Genome Project.

Senior Forum

Dear Readers:

The Foundation and the editorial board of the Ichthyosis Focus would like to institute a Senior Forum in our next issue of the newsletter, Fall 2002, that will address the unique needs of our senior members. We are not sure what format the forum will take, but we are open to suggestions and recommendations from you. It could be a question or answer format with questions generated from our senior readers. It could resemble our current Ask the Readers column. Or we could create a Focus On Seniors where members could submit their personal profiles and talk about how they manage their life and disease.

If you have a suggestion about what kind of format this new forum should take, please contact Maureen in the national office, 1-800-545-3286, or email us at info@scalyskin.org and share your ideas.

Ichthyosis Awareness Week

Mark your calendars! This year National Ichthyosis Awareness Week is scheduled for the week of October 6 through 13. All members of the Foundation are encouraged to promote ichthyosis awareness in your community. While the staff in the national office continually works to promote knowledge and awareness of ichthyosis, you are our best ambassadors for extending that knowledge and awareness into your community.

There are several ways to be involved in Ichthyosis Awareness Week. You can contact your local media with your own personal story about ichthyosis. You can write letters to the editors of your local newspapers, or offer to speak during a local school assembly, science class, or health class. Hosting a fundraiser accomplishes two goals, supporting the Foundation and increasing awareness of ichthyosis. Easy community fundraisers include: Auctions, Bake Sales, Bingo or Bowl-a-thons, Coin Harvests, Community Dog or Car Washes, Garage Sales, Raffles, Run/Walks, or sporting events. You can contact the national office for more information or direction on how to accomplish any of these suggestions. All ideas are good ideas; be creative, have fun and get your family involved.

Be sure to contact the national office to let us know what you are doing. We would like to recognize and share your efforts with the other members of the Foundation.

Ichthyosis Support Network

The Ichthyosis Support Network provides our members with moral support, practical advice, guidance, education, and resource information through telephone and email contact with volunteer peer counselors. Volunteer peer counselors are individuals with ichthyosis, or parents of children with ichthyosis, who feel they have the time and experience to offer support to others in the same situation. Our peer counselors live all over the country and represent all age groups and levels of experience.

The Ichthyosis Support Network is always in need of more volunteers. The time commitment is minimal, but the rewards are great. If you feel you have something to offer someone else struggling with these diseases, please call Maureen in the national office, 1-800-545-3286, to find out more about how you can help. The network is particularly in need of people to represent the more rare forms of ichthyosis, such as Palmoplantar Keratoderma, Pachyonychia Congenita, Pityriasis Rubra Pilaris, and Epidermal Nevus. We are also looking for people who are fluent in a language other than English, particularly Spanish.

Correction

Camp Discovery

The location given for Camp Discovery in the last issue was incorrect. Camp Discovery meets at Camp Knutson in Crosslake, Minnesota. The editorial staff of The Ichthyosis Focus apologizes for any confusion this may have caused.

Executive Director's Report

Dear Friends and Members of The Foundation:

I want to formally thank and recognize the Strategic Planning Committee for their hard work and commitment toward the development of our Strategic Plan. The Board of Directors approved the plan at the March board meeting. The objective of the Strategic Planning Committee was to determine how the organization could have the greatest impact on behalf of its members in the next three to five years. In a process that began more than one year ago, the committee sought input from members of the Board of Directors, past leaders of the organization, members of the Foundation, and professional planners. The information collected from these sources was used to help build a strategic plan that will meet the future needs of the Foundation's constituency.

I will continue to keep our members informed of the plan's progress in my quarterly reports in this newsletter. However, if you have specific questions or would like more information, please do not hesitate to contact me at the office.

In addition to my annual April trip to Washington, DC, for NIAMS (National Institute for Arthritis, Musculoskeletal and Skin Disease) Day, I attended a meeting with Dr. Stephen Katz, Director of NIAMS, along with other representatives of the Coalition of Patient Advocates for Skin Disease Research. Dr. Katz explained that NIAMS will be hosting a "Burden of Skin Disease" Meeting on September 4-5, 2002 in Washington, DC. Representatives from the Coalition are invited to this meeting. The purpose will be to identify ways to address the issue of the burden that skin diseases place on Americans. Dr. Katz stressed that this initiative will not only focus on the financial burden but the psychological burden as well. Since the psychological burden will be difficult to quantify, this meeting will explore the best way to do this.

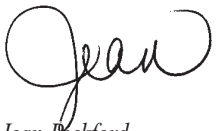
Dr. Katz also announced that NIAMS will award its first set of applicants under the Loan Repayment Program this year. The repayment program will grant up to $35,000 per year, plus taxes, for a two-year contract to young doctors whose careers will focus on research. Dr. Katz also reminded our group that NIAMS is a resource for funding for scientific meetings. An application, commonly called an R13, can be submitted for consideration if organizations are planning a scientific meeting that is specific and relevant to the latest information about the disease.

While in Washington, I visited several legislative offices to encourage their support of increasing the NIH and NIAMS budgets. This is the last year of the plan to "double the NIH budget in five years." The legislative assistants commented that they do not foresee a problem with this increase for the last year of the plan, but cannot promise that this type of increase will continue in the future. Due to the current state of the country, much more funding will be spent on bioterrorism and homeland security.

The Foundation received a wonderful opportunity to print a new brochure for an incredible price. Thanks to Maureen Tierney, our Program Director, we secured a volunteer graphic designer, Janet Felton, and she created a very professional color brochure with many descriptive pictures of our members.

The Family Conference will be here shortly. Both Maureen and I are excited about flying west and meeting everyone in Seattle. Registration forms continue to arrive at the office and we are anticipating over three hundred attendees. If you have not registered yet, please visit our website at www.scalyskin.org and download the form. This year's conference promises to be one of the best. There are exciting programs for all age groups and we will have the leading medical experts available to answer your questions about ichthyosis.

Sincerely yours,



Jean Pickford

I am pleased to welcome our newest employee, Lorraine Daniels. Lorraine replaced Kelly Strother as Office Assistant, and joined our staff on April 30. Lorraine jumped right in and is busy familiarizing herself with our office systems. Lorraine has excellent interpersonal and administrative skills and will be a great asset to our staff.

The Only One

Standing beneath the desert sun,
Wondering why I have to be the only one…
As a man I feel I should be strong,
But no matter how much I want it,
I know I can't stay long
Under the sun,
The only one…

Up on my feet,
Feeling my fast heartbeat,
I know that I soon will need to retreat.
I gave it my best,
It wasn't enough,
It's hard to digest
I can't be that tough.
Under the sun,
The only one…

Now I'm alone
Missing the fun,
But I need to release
My hate for that sun—
The sun is our friend,
It's really my skin
That I need to accept,
And it's time to begin.

- Eric Beeler 8/15/00

"In general, the goal in taking care of ichthyosis is to hydrate (moisturize) the skin, hold in the moisture, and keep scale thickness to a minimum."*

***Foundation for Ichthyosis & Related Skin Types, http://www.scalyskin.org**

Aquaphor® Healing Ointment helps heal dry skin associated with ichthyosis. Its unique petrolatum-based formulation combines a moist environment with the benefits of a semi-occlusive barrier that allows skin to breathe and absorb fluids.

Aquaphor is ideal for daily use because it is hypoallergenic, non-comedogenic, fragrance and preservative-free. Aquaphor Healing Ointment is safe enough for even the most sensitive skin.

Special Offer

For Readers of this F.I.R.S.T. Newsletter

MANUFACTURER'S COUPON | EXPIRES 06/30/03

Save $2.00 on any Aquaphor® Product

(NO TRIAL SIZES)

Consumer: Coupon good on the purchase of any Aquaphor® Product, no trial sizes. Limit one coupon per item purchased. Any other use constitutes fraud. Not good with any other special offer. Consumer must pay sales tax.
Retailer: You are authorized to act as our agent to redeem this coupon and we shall reimburse you at face value plus 8¢ handling in accordance with our redemption policy. Cash value 1/100¢. Copies available upon request. Offer void if copied and where prohibited, taxed or otherwise restricted. Mail to: Beiersdorf Inc, PO Box 880504, El Paso, TX 88588-0504. Good only in USA.

What's New, What's Hot & What Works…

Hello and FYI,

I am an ichthyosis sufferer and have come across a product, which has helped my dry, cracked feet. It is The Body Shop Hemp Foot Cream. I hope this can be of help to somebody as it has been to me.

Regards,
Bethany Calsy
Madrid, Spain

Editor's Note: Try www.the-body-shop.com to find a store near you, or call 1-800-BODYSHOP (263-9746) to receive a catalog or place an order.

Dear Members of FIRST:

Our son Patrick, who is fourteen years old, has lamellar ichthyosis and is doing very well! We've never really liked many creams and always made our own with a recipe that was given to us by another family, who's son had lamellar. The homemade cream was made of glycerin, unibase, and water. We have been unable to get any pharmacy to supply us with the unibase. They gave us other compounds that were similar, but none of them were the same when mixed with glycerin and water! Then a friend of mine was talking about Emu oil and how it cleared up her niece's acne. So I contacted the people who were dealers and we now use this oil on Patrick. It has made some major differences in his skin, especially on the nape of his neck!

The oil, after a few uses, loosens the scales and they slough off easily. The neat thing about the pure oil is that you only need a very small drop to do so much. If the skin is wet, then a small amount can cover a large area. The people who sold me the oil said if you can feel the oil on your skin then you used too much. We also use the body lotion, but personally I like the pure oil better.

Emu oil is not a cure or medicinal drug, but it does keep his skin hydrated nicely, and the nape of his neck no longer looks like dried out alligator skin! We've been using this oil for six months and I like it. But more importantly, Patrick likes it too. So I'm asking if you could put my information in the newsletter for others, especially if others are having problems finding unibase for the homemade cream. And, if someone knows where or how to get a hold of unibase, please have them contact us!

Marlene, Joe and Patrick Huffman
Hutchinson, MN

Dear F.I.R.S.T.,

I have intense itching on certain parts of my body that eight different dermatologists could not help. I found an amazing product called Natural Relief 1222. I suffered for years before I found it. It really takes the itch away. And over time you can use less and less, but get the same relief.

I think it was originally made for dermatitis and eczema. It's expensive, but you only need a little to get relief. You can order it directly at 315-697-8116. Also, Dermarest for psoriasis is excellent for itching. I use Lac-Hydrin 12% too, which is wonderful.

Debra Isula
Tonowanda, NY

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For more information contact:
Lou Dretchen, R.Ph.
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News & Notes

Rare Diseases Legislation Alert

Two critically important laws were introduced in the House of Representatives to strengthen research programs on rare disorders at the National Institutes of Health (NIH) and the Food & Drug Administration (FDA). The two bills are companion legislation for the Rare Diseases Act of 2001 (S.1379). The House bills separate the two initiatives for the NIH and FDA by making each a separate piece of legislation.

One bill in the House of Representatives is named The Rare Diseases Act of 2002 (H.R. 4013). It will establish an official Office of Rare Diseases at the NIH. Congressman John Shimkus (R- IL) and Henry Waxman (D- CA) introduced this bill. The Office will promote and coordinate research on rare disorders, and will create academic Centers of Excellence for research on these conditions. The NIH Office of Rare Diseases would receive $24 million per year for this program.

The second bill in the House is named The Rare Diseases Orphan Product Development Act of 2002 (H.R. 4014). Congressman Mark Foley (R- FL) and Henry Waxman (D - CA) introduced this bill. The law would provide $25 million per year for the FDA's Orphan Products Research Grant Program, which supports clinical trials of new orphan drugs, diagnostics, medical devices and medical foods. This program received only $12 million from Congress this year, which is less than the funds appropriated for these research grants in 1995. To date, twenty-seven new products (twenty-four orphan drugs and three medical devices) have been developed and are currently on the American market because academic scientists and small companies received grants from this program to support clinical trials.

These two bills are critically important to people with orphan, or rare, diseases who are waiting for new treatments and cures to be developed for their health conditions. In order to ensure that Congress will enact these laws, interested people are urged to contact their congressional representatives and ask them to co-sponsor H.R. 4013 and 4014. More than half of the House of Representatives must sign on to the bills before the entire House will vote on them. If we want these laws to be enacted this year, this must occur before Congress recesses before the election season this autumn.

It is best to phone your congressional representative either at their state office, or their Washington D.C. office (phone the Capitol switchboard at 202-224-3121 and ask for your Congressman's office). Remember that mail to Washington is delayed by four or more weeks due to security concerns. If you wish to send a letter, either mail it to your congressman's local state office, or FAX it to their Washington, D.C., office, or you can go to: < http://www.house.gov/writerep/ > and email your representative.

Reprinted from the National Organization for Rare Disorders On-line Bulletin.

AAD / CPA-SDR Partnership

The Academy of Dermatology (AAD) recently hosted a Partnership Conference with the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR) with the goal of helping patient advocate groups communicate effectively. Twenty patient advocate group representatives participated in the Chicago conference, including the Foundation for Ichthyosis & Related Skin Types.

Presentations given by AAD staff included: "Developing a Marketing Strategy," "Defining Your Message," "Producing Effective Materials," and "Effective Boothmanship". AAD 2002 President Fred F. Castrow II, MD, urged representatives of the patient advocacy groups to continue to participate in Academy of Dermatology activities, and he encouraged patient advocates to have regular communication with him and the Academy. "We want to hear from you," Dr. Castrow told the group. "We are working together, we're in this together."

"The most valuable point of the whole meeting was that the AAD is accessible and that the staff is willing to address the questions we might have to create a more effective relationship between the organizations," said CPA-SDR Co-Chair Karen Ball, executive director of the Sturge-Weber Foundation.

The overriding concern voiced by CPA-SDR members who attended the meeting was the need to increase awareness of their groups to both physicians and patients. Several members expressed concern that patients are not receiving information about these support groups from their dermatologists. "One of our biggest goals is education for patients and physicians," said attendee Judy Jones of the Mycoses Fungoides Foundation.

According to Cheryl K. Nordstedt, deputy executive director of the Academy, the partnership conference is part of a larger ongoing effort by the AAD to partner with patient advocacy groups in a number of areas including patient advocacy in Washington, research for skin disease issues, lobbying efforts, and broadening the group's involvement in government affairs issues that affect patients.

Excerpted from Dermatology World, March 2002

News & Notes continued on page 15

Coalition members gather at the AAD/CPA-SDR partnership conference.

Understanding The Human Genome Project

By Jean Pickford

Does the phrase 'gene sequencing' mean organizing your jeans according to shades of blue denim in your drawer? Is the Human Genome Project a discussion topic at your dinner table? Or does it seem so complicated and overwhelming that it's hard to comprehend? Well, if this describes you, you are not alone. Many individuals know very little about the recent advances that have been accomplished with the human gene. I hope this article will help explain, in an easy-to-understand way, the subject of genetics, the Human Genome Project, and genomics in today's society.

I had the privilege of attending a meeting at the National Institutes of Health (NIH) where Alan E. Guttmacher, MD, gave an informative presentation about genetics and the Human Genome Project. Dr. Guttmacher is the Senior Clinical Advisor to the Director at the National Human Genome Research Institute (NHGRI) at the NIH. So I was learning from the best.

Our genes are the basic building blocks of the human body. They determine who we are as individuals. Genetics is the study of the genes and how they function. Tradtionally, the term genetic condition refers to conditions wholly caused by an extra or missing complete chromosome or part of a chromosome (e.g., Down Syndrome), or a mutation in a single gene (e.g., cystic fibrosis, tuberous sclerosis).

These conditions are of great importance to the people they affect. But even when they are added together, they are still relatively rare. Since the majority of people are not affected, genetics plays a small role in the health care industry and society. But, did you know that nine out of ten leading causes of mortality have genetic components? Conditions such as heart disease, cancer and diabetes are all affected by genetics.

So what is the Human Genome Project? Begun formally in 1990, the Human Genome Project is a thirteen-year effort, coordinated by the U.S. Department of Energy and the National Institutes of Health. Project goals are to:

- *Identify* all the approximately 30,000 genes in the human DNA.
- *Determine* the sequences of the 3 billion chemical base pairs that make up the human DNA.
- *Store* this information databases.
- *Improve* tools for data analysis.
- *Transfer* related technologies to the private sector, and
- *Address* the ethical, legal and social issues that may arise from the project.

The human genome consists of three billion base pairs of the amino acids, Adenine, Cytosine, Guanine and Thymine. If you would print out the first letter of these amino acids in their order of four, A,C,G,T, the combinations would fill over 150,000 telephone book pages. Disease is often caused by a single variation in these three billion bases. It can be compared to one incorrect letter out of all those 150,000 telephone book pages. Genomics is the study of the human genome sequence.

Genomic Medicine refers to conditions partly caused by mutation(s) in gene(s) (e.g., breast cancer, Alzheimer disease) and prevented by mutation(s) in gene(s) (e.g., HIV). These conditions are of great importance to individuals and families who are affected by them because they are quite common and affect virtually everyone. This will create a large role for genetics in the health care industry and society.

Genomic medicine will change the health care industry by creating a fundamental understanding of the biology of many diseases, even the "non-genetic" ones. This understanding will lead to defining disorders by the mechanism that causes them, rather than by symptoms. Genomic medicine will change health care by providing knowledge of individual genetic predispositions and other technologies. Having the knowledge of genetic predispositions will allow individualized screening (e.g., mammography, colonoscopy), individualized behavior changes and presymptomatic medical therapies (diet, exercise and antihypertensive agents before hypertension develops). Genomic Medicine will change health care by creating pharmcogenomics. Pharmacogenomics will allow new approaches to drug design, individualized medication based on genetically determined variation in effects and side effects, and new medications for specific genotypic disease subtypes.

Genomic Medicine will change health care by providing a better understanding of non-genetic (environmental) factors in health and disease, allowing genetic engineering, and emphasizing health maintenance rather than disease treatment.

All of these incredible advances are bringing us into the "New Genetics" era. New genetics will include knowledge about traits that most of us see as human characteristics, rather than diseases (e.g., height, intelligence, athletic ability, etc.). New genetics will change our lives by knowing our own (and others) diseases dispositions, knowing our (and others) characteristic dispositions, and showing us that we are all mutants!

New genetics may also change society by genetic stratification (e.g., in employment and marriage), genetic engineering against diseases and characteristics, cloning and an increased opportunity for 'private eugenics." All of these issues raise new concerns for all Americans such as discrimination against individuals and groups, nature vs. nurture (genetic determinism), genes running in families, and confidentiality/privacy issues. More concerns regarding fairness in access, the right not to know and not to act, what is an appropriate informed consent process, and patenting and licensing will also become areas of concern.

The National Human Genome Research Institute funds and conducts very little disease-specific research, knowing that other institutes and centers at the NIH are ably utilizing a genomics approach. The NHGRI attempts to fund and conduct research that provides cross-cutting scientific and technologic tools that further such disease- and tissue-specific research. The NHGRI also funds

continued on page 15

Human Genome Project

continued from page 14

and conducts some scientific research that promises to establish new ways of looking at research and clinical care.

As completion of the sequencing of the human genome draws near, the NHGRI is enthusiastically planning for the era of genomic research and genomic medicine. Dr. Guttmacher explained that the entire human genome sequence will be mapped by April 2003. Ironically, that date will mark the 50th year anniversary of the discovery of DNA by Drs. Watson and Crick. The NHGRI is in the midst of developing a new plan which will look at the future of genomics and what role the NHGRI will play in helping to develop that future. Clearly translational research will be a primary focus that emerges from this planning process. Translational research means translating the discoveries made into a medicine or therapy that someone can use to treat, prevent or cure a disease or condition. If you are interested in learning more about the planning process, log on to www.nhgri.nih.gov/ planning.

There are two important areas of skin-related research that investigators at the NIH are pursuing. Dr. Jeffrey Trent's lab is using new genomics-based knowledge and techniques to look at the biological mechanisms underlying the spread of melanoma. Dr. Julie Segre's Epithelial Biology Section focuses on the dynamic process by which the epidermis maintains the proper balance between proliferation and differentiation.

Why is the Human Genome Project and the resultant genomic medicine important for clinical research in the new millennium? As President Bush stated at the White House on April 10, "Our age may be known to history as the age of genetic medicine, a time when many of the most feared illnesses were overcome." And former President Clinton stated on June 26, 2000 after the announcement of the completion of the human genome draft sequence, "It is now conceivable that our children's children will know the term cancer only as a constellation of stars."

New Publications

New Brochure

Thanks to the generosity of Felton Design and Clare Printing the Foundation now has a new four-color brochure, which we will be using as our primary tool to educate the public about ichthyosis and the Foundation's services.

Our printer, Clare Printing, Sayre, PA, offered to print a new four-color brochure at a deeply discounted rate. Janet Felton of Felton Design, Princeton, NJ, volunteered her services in designing the new brochure. The result is a new colorful brochure that offers an accurate depiction of people with ichthyosis. Several of our members agreed to let us use their photographs so that we could provide that accurate depiction.

The Foundation is very grateful to Felton Design, Clare Printing, and our members for their generosity and cooperation in providing us with this valuable new tool.

Resource Fact Sheets

The Foundation has compiled useful information and helpful hints on the following subjects related to ichthyosis as an additional service to our members:

- Overheating
- Retinoids
- Scalp Scale
- Ear Wax and Scale

For many individuals with ichthyosis these topics can be a real concern. Having accurate, concise, and helpful information at your fingertips can be useful.

The Resource Fact Sheets are available to all Foundation members. Simply go to our website, www.scalyskin.org, to order your complimentary copy. Please provide your name, address, and phone number and the name of the fact sheet you are requesting. Someone from the office staff will send it out immediately. Or call Maureen in the national office, 1-800-545-3286, and provide the same information. Other topics will be added to the Resource Fact Sheet series. Please let us know if you have a suggestion for a topic.

New & Notes continued

Don't Laugh at Me

The Don't Laugh at Me Project (DLAM) is working to transform schools, camps, and other youth organizations into more compassionate, safe, and respectful environments. Founded by Peter Yarrow of the folk group, Peter, Paul and Mary, the project offers educational resources that are designed to promote a climate that reduces the emotional and physical cruelty that some children inflict on each other by behaviors such as ridicule, bullying and — in extreme cases — violence.

The Don't Laugh at Me Project has developed three curricula, one for grades two to five, another for grades six to eight, and a third for summer camps and after school programs. The programs all utilize inspiring music and video as well as materials based on the well-tested, highly regarded, conflict resolution curricula developed by the Resolving Conflict Creatively Program (RCCP) of Educators for Social Responsibility (ESR). Don't Laugh at Me is a gateway program; it is designed to provide teachers, school counselors, social workers, administrators and other professionals with an entry point for year-round social, emotional, and character education programs, as well as to interface with and energize other efforts of this kind.

The Don't Laugh at Me Project is a project of Operation Respect, Inc. (OR), 2 Penn Plaza, 23rd. Floor, New York, New York 10121, www.dontlaugh.org.

Reprinted from Let's Face It 2002 Spring Resource Update and the Don't Laugh at Me website.

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