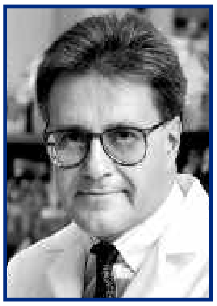
Ichthyosis Focus

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

Vol. 24, No. 1 *A Quarterly Journal for Friends of F.I.R.S.T.* *Spring 2005*

Gene for Harlequin Ichthyosis Found

By Jouni Uitto, MD, PhD



Harlequin ichthyosis is a devastating congenital skin condition which often leads to death of the newborn shortly after birth. Harlequin ichthyosis is also known as “harlequin fetus” because the affected children, in most cases, are born prematurely. The infants are encased in a thick skin, described as a “coat of armor,” that severely restricts their movement. As this thick skin dries out, it forms hard diamond-shaped plaques, which severely affect the facial features, distorting the lips, eyelids, and ears. In general, newborns with Harlequin ichthyosis die within a few days of birth because of respiratory distress, bacterial infections, and feeding difficulties. However, as a result of improved neonatal intensive care, some of the affected individuals survive and the skin comes to resemble severe non-bullous congenital ichthyosiform erythroderma, i.e., red and scaly skin without blisters.

The molecular basis of Harlequin ichthyosis was not known until very recently. Two groups have now been able to demonstrate mutations in the ABCA12 gene in patients with this disorder. First, a group of scientists at the Centre for Cutaneous Research in London set out to identify and map minute variations in the DNA sequences in families with affected and unaffected children with Harlequin ichthyosis. Evaluation of these families placed the probable site of the gene to the long arm of chromosome 2. Microsatellite markers narrowed the focus to the location of 6 identifiable genes. Within this relatively small region on the chromosome, the gene ABCA12 was considered as a candidate gene for the following reasons. First, a characteristic feature of the cells of the epidermis in Harlequin ichthyosis is abnormality in the way that lipids are transported and discharged into the top layers of the skin, together with abnormal lamellar granules. (Lamellar granules are tiny spherical grains that migrate upwards through the skin, depositing lipids into the intercellular spaces of the epidermis.) At the same time, ABCA12 may encode a transmembrane transporter protein involved in the transport of epidermal lipids. Secondly, the ABCA12 gene has been associated with mutations in a milder form of ichthyosis, lamellar ichthyosis, which resembles the expression of Harlequin ichthyosis in patients who survive beyond the early newborn period. Sequencing of ABCA12 gene allowed identification of mutations in both alleles in 11 out of the 12 cases studied. With one exception, all cases identified identical gene pairs for mutations that were predicted to result in shortened or missing protein.

Independently, a research group at Hokkaido University in Japan took the candidate gene approach to study ABCA12 in Harlequin ichthyosis patients. Similar to the studies by the British investigators, the Japanese scientists were able to identify mutations in ABCA12, which were predicted to result in shortening or deletion of highly conserved regions of the protein. These investigators further demonstrated the effect of ABCA12 protein on lamellar granules and in areas of lamellar granule-cell membrane fusion in normal epidermal cells. The Japanese team observed expression of ABCA12 in the normal keratinization of human epidermis, and they confirmed defects causing congested lipid secretion in Harlequin ichthyosis patients' skin. Finally, the lamellar granule-mediated lipid secretion was resumed in patients' cultured keratinocytes upon transfer of the wild-type ABCA12 gene. Thus, based on these two studies, it is very clear that mutations in the ABCA12 transporter underlie Harlequin ichthyosis.

What are the translational consequences of finding the gene harboring mutations in Harlequin ichthyosis? In other words, is there something that benefits the patients and their families? An immediate benefit is the possibility of DNA-based prenatal diagnosis, which has not been possible without the knowledge of the mutated gene. Considering the severity of Harlequin ichthyosis, which frequently has a lethal outcome during the early newborn period, there has been a demand for

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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.:

Thank you for publishing my letter. I have been getting a lot of responses from people all over the world. Just yesterday, I got mail from someone in India who has a 4 year-old girl with lamellar ichthyosis. It is great to be in touch with so many people and share information. It is only possible because of F.I.R.S.T.

Tsering and I are planning to attend the F.I.R.S.T. conference in 2006. We just heard that it is taking place in Atlanta. We will be there if everything works out with our visas.

Tsering is currently using a gel called Tazorac, which is available in the U.S., and it is really working wonders. I use it sparingly on her face and neck area once a week, and already the dead, stubborn skin there has come off and she is looking better.

Thank you. Much love from Bhutan to all at the F.I.R.S.T. office.

Ugyen Pelden
Bhutan

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

Hi. For those of you who don't know me, my name is Becky Butler. I am a 24-year-old New Yorker living with lamellar ichthyosis. I am also an Occupational Therapist who works with children and adults that have special needs. My joy comes from helping others and watching them conquer their challenges and achieve their goals. It is now my turn to achieve one of my goals, and conquer the many challenges along the way. My goal is simple, really; I would like to raise money for ichthyosis research while also exposing and educating society on the world of ichthyosis. The plan involves my sending letters and information to well-known authors, actors/actresses, and/or musicians and asking them for their help to educate society and raise money for the F.I.R.S.T. foundation through book signings, autograph signings, and eventually, hopefully, a benefit concert.

Upon meeting with them, I would like to present to them what and whom F.I.R.S.T. represents in a personal way. So I am asking for your help. I would like to compile a variety of materials; photographs (you, family, friends met through F.I.R.S.T., hands, feet, you with your favorite pet, a particular memory that sticks out, or you accomplishing a goal, etc.), hand-drawn pictures, letters, poems, collages, etc. Anything you feel expresses what it is like to live with ichthyosis, the upsides, downsides, the challenges, your greatest achievements, family support, which will then be used to create a scrapbook that represents and educates in a truly personal way. So, if you could email me at BeBeWicket@hotmail.com (please label subject F.I.R.S.T., so I don't accidentally delete as junk) or write to me, Becky Butler, 647 Latta Road, Rochester, NY 14612, that would be great! I will deeply and truly appreciate any and all contributions and, hopefully, it will lead to further exposure, societal education, and increased funding for ichthyosis research.

And who knows, from this F.I.R.S.T. may find someone who will annually assist us in raising money or advocating in front of Congress, or someone that might be able to educate large masses of society or just be a friend to the Foundation. You never know until you try, right? Now don't get me wrong, I am prepared for a lot of rejection letters; however, I have to believe that there are “popular” or “famous” people that would like to use their popularity and the influence they have over others to do good. Julia Roberts advocates for Rett Syndrome, the late Princess Diana greatly support AIDS research, the late Christopher Reeves hope and support for research on spinal cord injuries, and so many more, help to educate society plus fundraise in positive and hope inspiring ways. I truly believe that if I give it my all, we may be able to do just that.

Thanks everyone. If you have any questions, feel free to call me at 585-368-8544.

Kind regards,
Becky Butler
Rochester, NY

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FDA Issues Public Health Advisory on the Use of Elidel® and Protopic®

The American Academy of Dermatology's Response and What the Advisory Means to Individuals with Ichthyosis

The FDA recently issued a public health advisory informing healthcare providers and patients about a potential cancer risk associated with the use of Elidel® (pimecrolimus) Cream and Protopic® (tacrolimus) Ointment. Elidel cream and Protopic ointment are topical immunosuppressant calcineurin inhibitors that are applied to the skin for treatment of atopic dermatitis, or eczema. They are the only approved drugs in this class.

Concern about the use of these products was based on information from animal studies, case reports in a small number of patients, and information on how these drugs work. Animal studies showed that three different species of animal developed cancer following exposure to these drugs. These studies were conducted at doses much higher than generally used by patients and the risk of cancer increased with increasing drug dose and duration. These products work by suppressing the body's immune system. A form of tacrolimus known as Prograf® is used in pill form or by injection to prevent organ rejection in patients with liver and kidney transplants. Prograf is known to cause both skin cancer and lymphomas in humans by suppressing the body's normal defenses against cancer. The cancer risk increases with higher doses and longer treatment courses. Since the approval of Elidel and Protopic, FDA received reports of lymphoma and skin cancer in children and adults treated with these drugs, but whether the reported cancers are associated with these products has not been clearly established. Both Elidel and Protopic are sometimes absorbed through the skin, although usually in very low amounts. Occasionally, children who have been treated with Elidel or Protopic have shown blood levels of these drugs.

Based on these concerns, the FDA is recommending: using minimum amounts of Elidel and Protopic for short periods of time; using these products as second-line agents not first choice treatments; and avoiding use in children younger than age 2. FDA also plans to add a black box warning to the health professional label on these products and issue a medication guide for patients outlining these restrictions on their use.

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Correspondence Corner

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

I am interested in ordering silicone bracelets that are like the ones currently being worn for breast cancer awareness (pink) or Lance Armstrong's Livestrong yellow bracelets. I've been researching companies, colors, costs, etc., and I am only able to order a minimum amount of 500. I'm interested in knowing how many of you would be willing to sell bracelets and participate in this fundraiser with me. My hopes are that with enough of your support and help we can make this fundraiser work, and in the process, spread awareness and education of what F.I.R.S.T. is all about.

If you wish to be a bracelet seller, please send me your name and the amount of bracelets you would like. No amount is too few to participate in this fundraiser, it will all add up! My idea is that the bracelets will be blue and white. I figure that because they will be 2-colors the cost would be $1.50 per bracelet ($.85 per bracelet at the bulk rate). I think the bracelets should say, “Butterflies of FIRST,” but if you have a better idea, please feel free to share!

You can contact me by email at BeBeWicket@hotmail.com (please label subject F.I.R.S.T., so I don't accidentally delete it as junk) or mail Becky Butler, 647 Latta Road, Rochester, NY 14612. You are also welcome to call me by phone, 585-369-8544. I look forward to hearing from all of you.

Ambitiously yours,
Becky Butler
Rochester, NY

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prenatal testing in families at risk for recurrence of the disease. In the past, prenatal diagnosis has relied on fetal skin biopsy that is performed late during the second trimester of pregnancy. It is predictable that in Harlequin ichthyosis, as has been the case in many other severe skin diseases, DNA-based prenatal testing will replace the fetal skin biopsy in short order. Such testing can be performed as early as the tenth week of gestation by chorionic villus sampling, or perhaps even earlier by non-invasive analysis of the fetal cells and DNA in maternal circulation. Finally, knowledge of precise mutations in these devastating diseases forms the basis for the application of preimplantation genetic diagnosis.

Dr. Uitto is the Chair of the Department of Dermatology and Cutaneous Biology and the Director of the Jefferson Institute of Molecular Medicine, Jefferson Medical College, Thomas Jefferson University, Philadelphia, PA.

For a complete copy of Dr. Uitto's article with the accompanying references, please contact Maureen in the Foundation office, 1-800-545-3286 or info@scalyskin.org.

Definitions of terms, in order of appearance:

Plaques - patches or flat areas.
Respiratory distress - trouble breathing.
Molecular basis - genetic defect.
DNA sequences - the order of nucleotides, the building blocks of DNA, in a gene.
Microsatellite - a segment of DNA with variable sequence that can be sequenced to follow the inheritance of a copy of a gene.
Epidermis - the outermost layer of skin.
Transmembrane transporter - a protein that carries substances across cell membranes.
Expression - the outward appearance of a person's disease as a result of a genetic defect.
Sequencing - reading the order of nucleotides in DNA.
Allele - a copy of the gene, one inherited from the mother and the other one from the father.
Keratinization - the normal conversion of keratin, the protein that makes up hair, skin, and nails, to resilient layers.
Chorionic villus sampling - a doctor obtains a piece of the placenta, which contains the same genetic materials as the baby, for testing of mutation.
Preimplantation - genetic diagnosis embryos are created outside the body by in vitro fertilization and genetic testing is performed on the embryos. Only the embryos that did not inherit the mutation are transferred to the woman's womb to create a pregnancy.

Executive Director's Report

April 2005

Dear Friends and Members of the Foundation,

A lot has been happening at the Foundation over the past few months. We are now in full swing with our annual membership campaign. Everyone in our database should have received a mailing from the office with a membership renewal form. Renewals and new registrations are beginning to arrive at the office. Thank you very much for your continuing support of the Foundation.

This past February, the Foundation's Medical & Scientific Advisory Board (MSAB) met in New Orleans at the annual American Academy of Dermatology (AAD) meeting. The MSAB, chaired by Dr. Leonard Milstone, welcomed its newest members, Dr. Anna Bruckner and Dr. Sancy Leachman. This 23-member board is such a vital resource to the Foundation. We rely on these exceptional doctors for accurate medical information, physician referrals, member inquiries, advancement of ichthyosis-related research, plus much more. It is an amazing experience to sit among the leading experts in ichthyosis and discuss how we can improve the lives of those affected with these diseases.

Jean Pickford (center) visits with Dr. John DiGiovanna, MSAB member, and Beth Gray, board member, at the Coalition's booth at the 2005 AAD meeting.

This year at the AAD meeting, a board member, Beth Gray, accompanied me. Beth graciously agreed to volunteer some time with me to man the Coalition of Skin Disease booth in the exhibit hall. The booth is a collaboration of various other skin disease organizations. The Coalition's booth showcases photos of affected individuals and provides disease-specific literature and resources to the thousands of dermatologists in attendance.

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Gene Therapy

By Joann Boughman, PhD

Gene therapy refers to the treatment of human disease by transfer of genetic materials (DNA or RNA) into a patient to replace a deficient or non-functional gene. In most medical treatments, a drug is used either to replace a missing or defective molecule (for example, thyroid hormone), to increase or stimulate production of a molecule, or to counteract or inhibit an adverse bodily production or condition (for example, cholesterol reducing pills, an antibiotic to fight an infection, or medications to lower blood pressure). In gene therapy, the concept is that a *gene* is introduced to produce the substance needed or to reduce a substance that is overproduced. While gene therapy holds great promise for the treatment of genetic diseases in the future, several challenges remain to be resolved through scientific research and clinical trials.

Because the control of expression of genes is not completely understood, targeting the gene to the right cells in the body and into the DNA of those cells is extremely difficult, leaving in question the safety and efficacy of gene therapy. To begin with, gene therapy can target two types of cells, germline cells or somatic cells. Germline gene therapy introduces genetic material into all cells of the body or a subset of cells, including the germ cells (reproductive cells, e.g., eggs or sperm). As a result, the genetic modification can be passed on to offspring, thereby permanently altering the genetic material. This type of gene therapy is not permitted in the United States. In contrast, in somatic gene therapy, genes are introduced into cells of various parts of the body (e.g., muscle, heart, bone marrow) but not the germ cells. This form of treatment affects only the patient and does not result in transmission of the genetic modification to future generations.

For gene therapy to be successful, several things must occur: 1) the gene must be known; 2) the gene must be able to be expressed (turned on); and 3) the gene must be introduced into the appropriate host tissue. Thanks to many years of genetic research and most recently the Human Genome Project, the genes responsible for many genetic diseases have been identified and studied extensively. However, introducing the gene into the target host cell and making sure it is expressed has proved to be more difficult. The most common approach to transmitting a gene into a cell involves using a virus as a transporter. The gene of interest is packaged into the virus, which has been rendered harmless. The virus carrying the gene of interest is then injected into the body and targeted to specific cells. Sometimes naked DNA (without a carrier) can be taken up by host cells such as muscle cells, but often times the DNA is degraded or does not efficiently integrate into the DNA of the host cell. Other carrier molecules or packages are also being examined.

Next, the DNA (gene) of interest must integrate into the DNA of the host cell and be activated. But sometimes the DNA can integrate at an important site in the genome, disrupting other important cellular functions and possibly resulting in serious adverse responses or even another disease. In addition, even if the gene is successfully integrated and expressed, the time of expression may be brief and require additional gene therapy treatment.

Gene therapy strategies may vary depending on the disease being treated and the type of vector used. In ex vivo gene therapy, cells are first removed from the patient and then mixed with the viral vector carrying the corrected gene. This type of gene therapy occurs “outside” of the patient. Once the cells have taken up the virus and the new gene, the cells are inserted back into the patient. Another strategy would take place “inside” the patient, known as in vivo gene therapy. In this approach, the viral vector is given to the patient and targeted to the affected tissue.

While the concept of gene therapy seemed straightforward when it was first tested in 1990, subsequent studies have revealed just how complicated the process is, requiring an exquisite understanding of the gene, the control of its expression, and the delivery of material that will not be rejected to the precise site of needed action. As many genetic disorders are the result of very early developmental alterations that result in long-term problems, not every genetic disorder can be treated through gene therapy.

Nonetheless, there are hundreds of gene therapy studies ongoing, and gene therapy trials are underway in many places across the world. Serious adverse effects remain a major safety concern for all gene therapy trials. Although the widespread use of gene therapy for many genetic disorders is years away, with each new understanding and study on gene therapy, scientists are advancing to the day when gene therapy can be used as a successful treatment.

Information about gene therapy trials registered with the NIH may be found by accessing the Genetic Modification Clinical Research Information System (GeMCRIS) available at http://www4.od.nih.gov/oba/RAC/GeMCRIS/GeMCRIS_public.htm

Dr. Boughman is Executive Vice President of the American Society of Human Genetics. She is a board certified medical geneticist and was on the faculty of the University of Maryland, Baltimore.

Executive Directors Report

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Representing eleven skin diseases, Jean Pickford and colleagues from the Coalition of Skin Diseases gather together to compare their organization's stories.

Beth also accompanied me on my visits to the pharmaceuticals booths in search of new products that may be beneficial for ichthyosis. We also expressed our thanks in person to the companies who continue to support our important mission.

As is typical in the spring, I traveled to Washington, DC, to participate in the annual Skin Disease Research Day on Capitol Hill. This year's event took place on April 5-6 and was organized by the American Academy of Dermatology Association. Skin Disease Research Day is an event where physicians, researchers, and patient advocates come together to promote increased research funding for the National Institutes of Health (NIH).

Along with my colleagues from the Coalition of Skin Diseases, members of the American Academy of Dermatology and the Society for Investigative Dermatology joined together for an advocacy briefing/training and then attended pre-arranged appointments with our senators and representatives. The purpose of our visit was to thank the senator or representative for their past support of increased funding to the NIH. We were also asking for his or her support of a 6% increase for research funding in 2006 in order to meet the needs of patients who are struggling with an incurable and often debilitating skin disease.

Jean Pickford and other representatives from the CSD, AAD, and SID meet in the lobby of the Senate Hart Building for appointments with Senator Arlen Specter (R-PA).

From 1998 through 2003, Congress provided a consistent 15% increase in order to meet the mandate of doubling the NIH's budget in five years. This was an incredible effort and significantly stimulated research in all health areas, including skin diseases. Many new projects and promises were made possible during this doubling phase. Since then, funding increases have been declining. The NIH received only a 2% increase this past year. These decreases are having a huge impact on health research because there is less money available to award to scientists and promising young investigators with fresh ideas. Some of the projects and research advances that have been started and maintained over the past few years are being dismantled due to the lack of funding. Our message on Skin Disease Research Day was to tell our senators and representatives that it is vitally important that Congress not stymie the efforts of physician researchers who are in the midst of cutting-edge medical research by under-funding the NIH's obligations.

Representative Jim Gerlach (R-PA, 6th District), Jean Pickford, and Dr. Jouni Uitto spoke in his office about increased funding for the NIH.

The President's proposed increase for the NIH for fiscal year 2006 is only 0.7% and only 0.4% for NIAMS, the branch at the NIH responsible for skin disease research. You can see we had our work cut out for us during our visits.

At this point, it is hard to predict what next year's congressional budget will appropriate to the NIH. We certainly did our best to educate our senators and representatives about the need for more research money. There is one thing that remains constant - children continue to be born with skin diseases, families continue to be affected, and our work continues.

Sincerely,

Jean

Jean R. Pickford
Executive Director

Filming for Teen DVD Completed

Alyssa Kosmer, Shawna Grady (and Shannon Tormey) smile happily when their interviews are completed.

Eleven teenagers and their families traveled to North Wales, PA, on a rainy weekend in April to be filmed for the Foundation's newest project, a DVD that addresses issues facing teenagers affected with ichthyosis. Thanks to a special grant funded by Operation Good Neighbor, the Foundation will be producing one thousand DVDs, which will be available this summer.

Bailey Pretak relaxes in the sunroom as she awaits her turn in front of the cameras.

After consulting with the production company, ASAP Productions, Maureen Tierney's home was chosen as the location for filming. Maureen, the Foundation's Program Director, is also the mother of two teenagers, and her house provided the perfect combination for the filming. Her home is equipped with plenty of entertainment for the teens and her decorating style provided various backdrops for each interview. The Foundation is grateful to Maureen and her family for opening their home for the filming.

Diana Grady, Nicole McMillian, and Katie Smith take a break from watching a movie to pose for the camera.

Parents Bill Grady, Debra Bowie, and Linda Kosmer listen to an entertaining story from an outgoing teen.

Each teen was interviewed on camera for approximately 45 minutes. Armed with a list of questions that were specifically chosen for each teen, Justin Tormey, the producer/director, sat across from each teen and settled into a conversation. The cameras, microphones, and lighting were strategically placed to get the best angles and shots as they answered questions and chatted. Once the nervousness of being a "star for a day" wore off, the teens spoke candidly about their ichthyosis and being a teen. Many issues were addressed, including relationships with peers, making new friends, college plans, skin care routines, school and extracurricular activities, and dealing with the public.

Justin Tormey, producer/director, focuses on his camera angle for the next teen interview.

Margaret Frost is all smiles as she arrives and is greeted by friends from past conferences and camp.

Matt Gray knows his interview went very well.

Half of our teen participants were local, while the other half traveled from other states. Some families traveled from as far as Nebraska, Tennessee, and Alabama. A grant provided by LaRoche-Posay, makers of Lipikar Baum, covered the costs of hotel accommodations and food during the weekend. In all, it was a great experience for everyone. One teen remarked, "It was really special to be a part of this filming. I know that what I said will affect a younger person with ichthyosis. I am glad to be able to help someone else who is dealing with our disease."

Cameraman Steve Thorne adjusts the lighting, making sure everything is just right.

Zebulun Sipper and his mom, Cathy, get directions from Maureen Tierney before they head out to downtown Philadelphia for some sightseeing.

Annual Report Fiscal Year 2004

President's Report

Dear Members and Friends of the Foundation:

Fiscal year 2004 was highly successful for the Foundation by many measures. As I've mentioned before, both the Board and the staff are committed to further improving the Foundation's governance. Last year, that commitment included the adoption of a new conflict of interest policy covering the Board and staff and the completion of a survey of our membership, which was used to develop a member profile, in accordance with the demands of the Foundation's strategic plan. The Board will meet for a two-day retreat in November 2005 to assess, among other things, the Foundation's progress against the strategic plan and to update the plan. All of the Board members feel a keen responsibility to the Foundation's members and reflect that focus in our oversight of the Foundation's operations.

We reached our goal of raising $100,000 for ichthyosis-related research, and the Board participated in fundraising education, including each Board member's committment to raise at least $1,000 in 2005. The Foundation also created and implemented a new planned giving program. We, in concert with several other organizations, were able to privately fund the maintenance of the Ichthyosis Registry for the next two years, an important commitment after the Registry's government funding ceased. My hope for a record number of participants at our 2004 National Family Conference in Kansas City was realized - it was our best-attended conference in the Foundation's history.

Once again, we all have much to celebrate in the Foundation's accomplishments. I continue to be especially grateful for the outstanding commitment and dedication of the Foundation's staff. I am particularly pleased that the Foundation's offices relocated to a larger facility this past year. This move should, on several levels, result in a better work environment for our talented staff. I remain in awe of the professionalism and passion that my fellow Board members bring to all their efforts, as well as the members of our Medical and Scientific Advisory Board. Collectively, I believe that the Foundation is poised to serve its members better than ever, especially in program services and research. I hope you will join me both in expecting great things from the Foundation and in assisting in the achievement of those great things yourself.

Sincerely,

Laura J. Phillips
President

Chief Financial Officer's Report

Dear Members and Friends of F.I.R.S.T.:

As Chief Financial Officer of your Foundation, I have the privilege to report on our financial status for fiscal year 2004, which ended September 30, 2004. I am happy to report that the Foundation is in solid fiscal shape and has been well stewarded by the Executive Director and your Board of Directors.

In 2004, our contributions rose by 57% versus the prior fiscal year. This does not include monies received for registration fees, in-kind contributions, or similar revenues associated with the national conference held in Kansas City. We have had several years in a row of solid growth for contributions received.

The most significant factor in our revenue increase is related to our research goals. As mentioned in previous communications to our membership, the Foundation has a goal of $100,000 per year to be raised for research. In fiscal year 2004, the Foundation saw a number of members reach out to their communities and business contacts to solicit funds. One grassroots effort garnered over $40,000. Another member's letter writing campaign resulted in a $50,000 grant from United Parcel Service.

During fiscal year 2004, our total expenses decreased by 5%. However, in fiscal year 2003, there was $80,000 awarded in research grants that the Foundation did not duplicate in fiscal year 2004. Also, in 2004 the Foundation saw a significant expense

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Chief Financial Officer's Report

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for the national conference which was not incurred during fiscal year 2003. Removing those two events from consideration, year-to-year comparable expense items only show an increase of 5% in 2004 versus 2003. Based on the exceptional contributions reported above, this modest increase is well within acceptable limits.

Much of the increased expenses came from a variety of basic items incurred by the Foundation's national office, such as telephone, printing, and postage expenses. The Foundation's office also relocated in 2004, which generated a small increase in rent expenses. The trade off is much better quality space for the Executive Director and her staff, which will enable them to continue producing great information and materials for our membership.

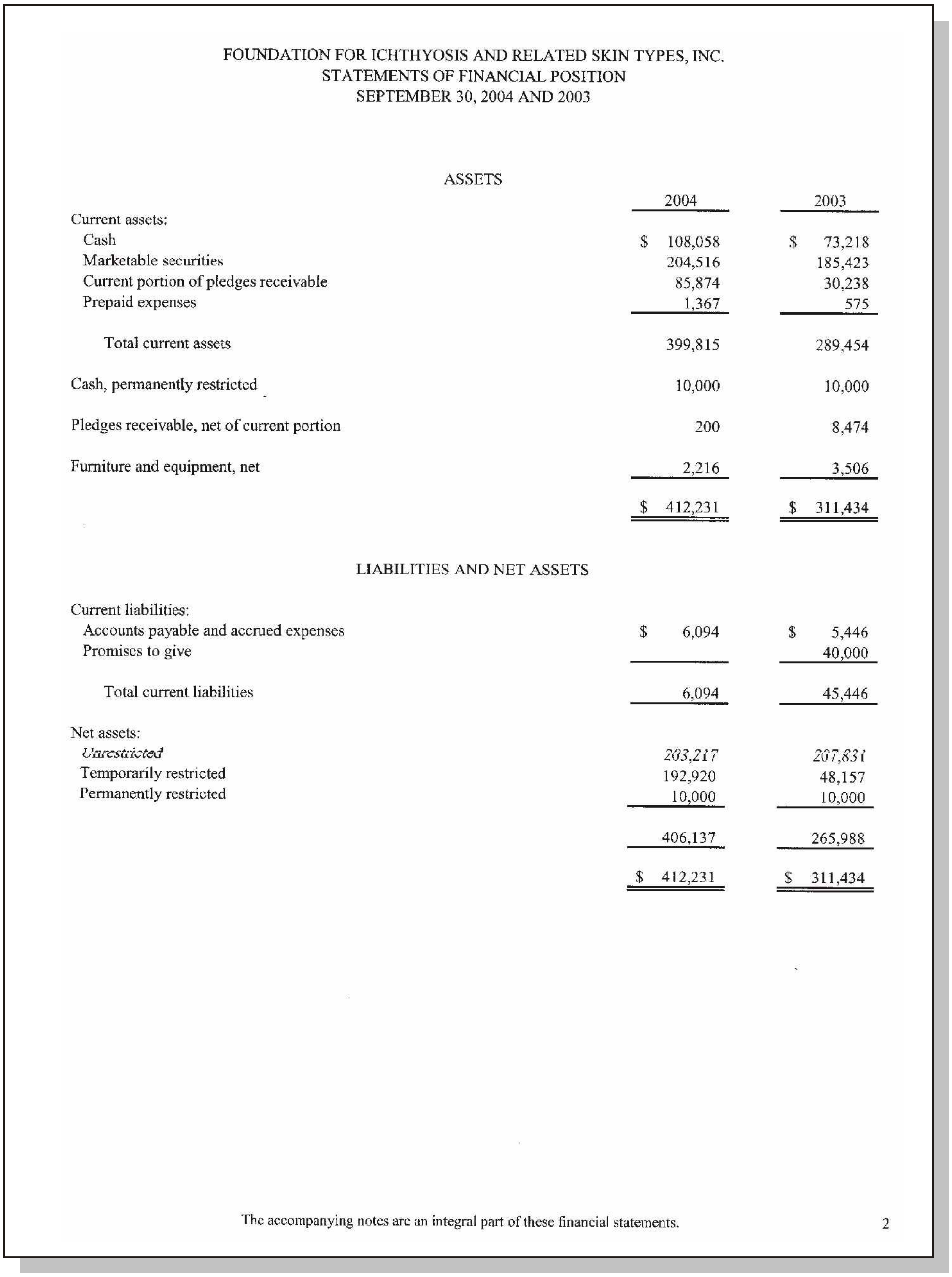
In 2004, many of you had the opportunity to experience another outstanding family conference. The event was well supported by members, friends, and corporate partners. In fact, 350 members and friends traveled to Kansas City last summer. That's the most ever! Prominent pharmaceutical companies continue to help support our functions, too, as over $9,000 was contributed to subsidize the expense of the conference.

As we move forward, I urge all members to be as active as possible. We, as an organization, are only as strong as you make us. Fundraising is critical if we are to attract researchers worthy of granting, who will take on the challenges of ichthyosis research. We will continue to seek those opportunities that will benefit our members and the entire ichthyosis community.

Sincerely,

Mark E. Klafter

Mark E. Klafter
Chief Financial Officer



FOUNDATION FOR ICHTHYOSIS AND RELATED SKIN TYPES, INC.
STATEMENTS OF FINANCIAL POSITION
SEPTEMBER 30, 2004 AND 2003

ASSETS

	2004	2003
Current assets:		
Cash	$ 108,058	$ 73,218
Marketable securities	204,516	185,423
Current portion of pledges receivable	85,874	30,238
Prepaid expenses	1,367	575
Total current assets	399,815	289,454
Cash, permanently restricted	10,000	10,000
Pledges receivable, net of current portion	200	8,474
Furniture and equipment, net	2,216	3,506
	$ 412,231	$ 311,434

LIABILITIES AND NET ASSETS

	2004	2003
Current liabilities:		
Accounts payable and accrued expenses	$ 6,094	$ 5,446
Promises to give		40,000
Total current liabilities	6,094	45,446
Net assets:		
Unrestricted	*203,217*	*207,831*
Temporarily restricted	192,920	48,157
Permanently restricted	10,000	10,000
	406,137	265,988
	$ 412,231	$ 311,434

The accompanying notes are an integral part of these financial statements.

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* *This Statement of Financial Position is excerpted from the Foundation's audited financial statements as of September 30, 2004. A complete copy of the audited financial statements and the independent auditor's report are available and can be obtained by calling the national office at 215-631-1411, or 1-800-545-3286.*

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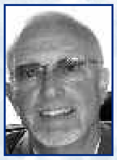
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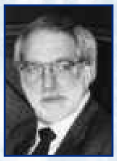
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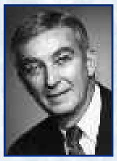
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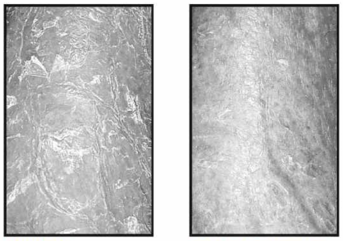
When Timothy* grows up, his parents want him to be ordinary. Just ordinary.

Problem Dry Skin (PDS) Symptoms of Lamellar Ichthyosis
BEFORE

AFTER 4 WEEKS

PDS Symptoms of Lamellar Ichthyosis



BEFORE

AFTER 4 WEEKS
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* Ficticious name. Consult a physician before using on children.

PAID ADVERTISEMENT

FDA Issues Public Health Advisory

continued from page 3

The American Academy of Dermatology responded to this action by the FDA in a statement issued on March 10, 2005. " The American Academy of Dermatology is disappointed that the FDA has taken this action, despite the fact that there is no data that proves proper use of pimecrolimus and tacrolimus is dangerous in people," said dermatologist Clay J. Cockerell, MD, president of the American Academy of Dermatology. "Because these medications are applied to the skin, virtually none of it gets inside the body. It's not the same as taking a pill. These are valuable medications, and if used properly, they significantly reduce the debilitating impact of eczema and allow millions of our patients to live normal lives."

On February 15, 2005, dermatologist Robert A. Silverman, MD, of Fairfax, VA, spoke on behalf of the Academy at an FDA pediatric advisory committee hearing, urging the FDA not to impose a black box warning or other labeling restrictions because such steps could limit access to these medications, or limit treatment options if qualified patients decide not to use these medications based on fear of cancer risk.

Advocates representing eczema patients also spoke out against more restrictive labeling. "These medications have been the only treatments that have given my children anything resembling a normal quality of life," said LaDonna Williams, executive director, Inflammatory Skin Disease Institute (ISDI) and mother of two children with severe atopic dermatitis. "Every day we see first hand how these medications improve the lives of patients and their families," said Ruthann Newton, support group coordinator for the ISDI.

"The health, safety, and welfare of our patients being treated with these topical medications is of paramount importance to dermatologists," said Dr. Cockerell. "We are concerned that these warnings will confuse and unnecessarily worry our patients. We urge patients to get the facts on how to appropriately manage their eczema from their dermatologist."

What does all this mean to individuals with ichthyosis? Dr Amy Paller comments: "In summary, there is no human evidence that these topical medications cause any serious health risk in humans. The lymphoma type of cancer that is caused by high doses of these medications, when taken internally, is a specific type that has not been described in patients using the topical form, despite a few reports among the large patient population applying topical medications of other forms of lymphoma. The animal studies with very high dosages and systemic levels of medication are the basis for concern. These studies have not been performed with the alternative agents, topical steroids, but topical steroids have been available for several decades. Although the long-term experience in humans may well be quite different than in animals, it is important to monitor individuals using these topical medications and to use good sun protection, at least for the next few decades until we can be sure that there are no important side effects. Of note, individuals with ichthyosis (and especially Netherton syndrome) may have greater absorption through their skin than persons with normal skin or even atopic dermatitis. Many individuals with ichthyosis are itchy and use Elidel or Protopic to control the itching and/or eczema that can accompany ichthyosis. Given the possibility that more Elidel or Protopic is absorbed through the skin in persons with ichthyosis, these agents should be used only selectively for ichthyosis and with regular monitoring by a knowledgeable physician. Assessment of blood levels can be performed as a part of the monitoring process."

Sources: U.S. Food and Drug Administration Website, Center for Drug Evaluation and Research - www.fda.gov/cder/drug/advisory/elidel_protopic.htm

American Academy of Dermatology Website - www.aad.org/public/News/NewsReleases/fda_decision.htm.

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News&Notes

FDA Announces Creation of Oversight Board

On February 15, the Food and Drug Administration (FDA) announced the creation of a new independent Drug Safety Oversight Board that will be responsible for the management of safety issues relating to FDA approved drugs currently on the market. The Board will provide physicians and patients with emerging information about the risks and benefits of medications.

The move by the FDA comes on the heels of criticism of the agency over its handling of reports that linked popular arthritis drugs to increased risk of heart attack and stroke. “The public has spoken and they want more oversight and openness,” said Department of Health and Human Services Secretary Mike Leavitt, during a meeting with FDA employees. “They want to know what we know, what we do with the information, and why we do it. We will address their concerns by cultivating openness and enhanced independence.”

The new Board will be charged with recommending information and updates for placement on the Drug Watch, resolving disagreements over how to approach drug safety issues, and assessing the need for MedGuides. It will also oversee development and implementation of Center-wide drug safety policies. Board members will include individuals drawn from within the FDA and medical experts from other HHS agencies and government departments, such as the Department of Veteran Affairs. The Board will also consult with outside medical experts as well as representatives of patient and consumer groups.

In addition to creation of the Board, the FDA administration announced its intention to share drug safety information with the public sooner and more broadly and conveniently. This includes the launch of a new Drug Watch Web page and dissemination of drug safety information sheets to patients and health care professionals.

“FDA understands that the public expects better and more prompt information about the medicines they take every day,” said Acting FDA Commissioner Lester M. Crawford, MD, who was recently nominated by President George W. Bush to assume the commissioner post permanently. “Our goal is to prepare the agency for these new demands by improving the way we monitor and respond to possible adverse health consequences that may arise regarding drugs that have been approved for sale to U.S. consumers.”

Reprinted from Dermatology World, *Vol. 15, No. 3, March 2005, p. 4.* Dermatology World *is a publication of the American Academy of Dermatology Association.*

Camp Wonder 2005

The Children's Skin Disease Foundation is now accepting applications for Camp Wonder 2005, an incredible camping experience being offered to children with skin disease, ages 6 to 16, at no cost to the family or child. The week offers the opportunity to participate in exciting activities, form lasting friendships, and ultimately develop a stronger self-esteem while under the loving supervision of an excellent medical staff and counselors.

The two sessions will be at Camp Wonder, Livermore, CA, June 19 to June 25, and at Victory Junction Gang Camp, Randleman, NC, July 31 to August 5.

For more information, please contact Christine Tenconi, 925-947-3825, or email ctenconi@hotmail.com. Applications are being accepted on a first come basis this year.

Web Site of Interest, Family Health History

www.hhs.gov/familyhistory/

Most Americans believe that knowing their family health history is important, but only about one-third have actually tried to gather and record this information, according to a study by the Centers for Disease Control and Prevention (CDC). Furthermore, the discussion of family history between patient and physician typically lasts just a few minutes.

“But knowing your family health history can save your life,” U.S. Surgeon General Richard H. Carmona, MD, said at a recent press conference to launch an online Family History Initiative. As part of this project, a computerized tool has been created to walk people through the process of creating a family history.

The site guides users through a series of screens to record health data for family members. All personal information is maintained on the user's computer. No information is given to the government. Once the history is complete, it can be printed and shared with a physician. People who don't want to use the online version, or who don't have a computer, may call the HRSA Information Center at 888-275-4772 for a print version.

Reprinted from Orphan Disease Update, *Volume 23, Edition 2, Spring 2005, a publication of the National Organization for Rare Disorder, Inc.*

Your Medical Information Bureau (MIB) Record

If, during the past seven years, you have applied for life, health, or disability insurance, it is likely that your health record is on the website of the Medical Information Bureau (MIB). MIB is an association of insurance companies that tracks medical records. When you sign a form allowing an insurance company to check your medical records, it is likely that they check your records on MIB.

Now the *Fair and Accurate Credit Transaction Act,* which gives us our annual free credit reports, will also give us access to our own health record report once each year at MIB. If the information is inaccurate, you can let them know.

You can reach MIB at www.mib.com. Go to "Consumer Site." Click on "Request your record," or you can phone MIB at 866-692-6901.

Reprinted from NORD ON-LINE, April 2005, a publication of the National Organization for Rare Disorders, Inc.

National Coalition for Health Professional Education in Genetics Survey

The National Coalition for Health Professional Education in Genetics (NCHPEG) and the Genetic Alliance are partnering in a survey. The purpose of the survey is to ask individuals living with relatives or friends with genetic conditions about their perceptions of their health care providers' knowledge of genetics. The information from this survey will be used to develop genetics education for specific types of health care providers.

This is an opportunity to be heard and to have our members' voices heard. Please use this link to respond to the survey, http://www.nchpeg.org/SFGC_Survey/Survey.asp.

The coalition would like to hear from many different people representing experiences with many different diseases.

The Genetic Alliance is a coalition of patient advocate groups who represent individuals with a variety of genetic diseases. The Foundation for Ichthyosis is a member of the Genetic Alliance.

Have you moved recently or are planning on moving soon? Please let us know your change of address so you can continue to receive the Focus. Postage for the newsletter does not include forwarding.

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