Ichthyosis Focus

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

Vol. 25, No. 3 | A Quarterly Journal for Friends of F.I.R.S.T. | Fall 2006

Foundation Research Grant Program Funds Four Projects

The Foundation's Research Grant Program, officially launched in January of 2006, has completed its first year with the receipt and review of several worthy projects and the decision to fund four of those projects. Projects have been selected in the categories of Epidermolytic Hyperkeratosis (EHK), Lamellar/Congenital Ichthyosiform Erythroderma (CIE), Sjögren-Larsson Syndrome, and ichthyosis-related research; contracts are being negotiated with the investigators' institutions.

Dr. Dennis Roop and his colleague Dr. Jiang Chen at Baylor College of Medicine in Houston, Texas have been selected to receive $75,000 for their project titled “Testing Therapeutic Approaches for Epidermolytic Hyperkeratosis Using a Preclinical Mouse Model.” Drs. Roop and Chen have developed a genetically engineered mouse in which they have been able to generate areas of EHK on the mouse's skin and paws. They will use this mouse model to test strategies to suppress or correct the expression of one of the mutant genes that causes EHK. This project is relevant to the mission of the Foundation and the interests of its members because, according to Dr. Roop, “Besides symptomatic care, no effective treatment is available for EHK. If the preclinical studies described above are successful, these strategies could be used to treat EHK patients.”

Dr. Judith Fischer of the Centre National de Génotypag in Evry, France, has been selected to receive $75,000 for her project, titled “Genetic and Clinical Studies of Autosomal Recessive Congenital Ichthyoses Belonging to the 12(R)- lipoxygenase Pathway.” Dr. Fischer will be working to identify mutations in the seven genes that are known to cause the autosomal recessive ichthyoses (ARCI), specifically lamellar ichthyosis and CIE. Dr. Fischer is collaborating with our own medical advisors, Dr. Philip Fleckman and Dr. Leonard Milstone, and will be utilizing genetic material from the National Registry for Ichthyosis and Related Disorders. Dr. Fischer's project is relevant to the mission of the Foundation and the interests of our members because: “The results of this project should lead to a better understanding of the metabolic pathways involved in the pathogenesis of ARCI and should provide potential targets for treatments.”

Dr. William Rizzo of the University of Nebraska Medical Center, Omaha, Nebraska, has been selected to receive $50,000 for his project, titled “Ichthyosis and 12R-Eicosanoid Metabolism in Sjögren-Larsson Syndrome.” Dr. Rizzo is looking at a defective lipid (fat) metabolic pathway that is seen in Sjögren-Larsson Syndrome (SLS) and several other genetic forms of ichthyosis. Dr. Rizzo's project is relevant to the mission of the Foundation and the interests of our members because: “Therapy of the ichthyosis in SLS is non-specific. Our research may lead to new approaches for cutaneous therapy for selectively bypassing the metabolic block in lipid metabolism and providing the metabolites that cannot be made by SLS patients.”

Dr. Eli Sprecher and his colleague Dr. Dina Ron of Technion Israel Institute of Technology, Haifa, Israel, have been selected to receive $50,000 for their project titled “SNAP29 - A Novel Regulator of Epidermal Differentiation.” Drs. Sprecher and Ron will be investigating the role of a small protein molecule in the physiological pathway that is important to normal skin development and function. Drs. Sprecher and Ron's project relates to the mission of the Foundation and the interests of our members because: “The data gleaned in the course of this research are expected to establish, for the first time, the importance of SNAP29 during epidermal differentiation and to shed light on its specific roles during this process. Establishing the role of SNAP29 during cornification may point to possible use of SNAP29 as a diagnostic marker as well as a therapeutic target for a number of inherited and acquired disorders of cornification.”

The Foundation wishes to thank all of our medical and scientific advisors who helped develop the research grant program and guided us through this first year. In particular, we would like to thank those scientists and physicians who volunteered their time to serve on the Research Review Committee. These talented and experienced individuals had the daunting task of reviewing the work of their peers in the ichthyosis community and deeming it worthy of the Foundation's mission and the funds that so many of you have generously contributed over the past few years.

Please see page 3 for a brief biography of each of our selected investigators.

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.

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Correspondence Corner

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

Hi. My name is Michelle and I live in Newport, NC. I am posting this because I also have ichthyosis. I do not know of anyone else who lives near me that has this, so you can imagine how alone I feel.

I am very seriously thinking about starting a support group for people, like myself, who have no one in their area to talk to or to share their thoughts and frustrations with. If we do have a support group, it will be in the Havelock, Beaufort, and Morehead City, NC area.

If anyone is interested in coming together as a group to support one another, please let me know by contacting me by email. I would like to see how many people would be interested in this idea before I pursue it any further. Please note that this group will be for our children as well. So it will be a family and friend support group for people of all ages. It does not matter what type of this disease you have, you are welcome to participate.

Thank you and God bless all of you!

Michelle Cassity
Newport, NC
Audry@ec.rr.com

Dear Friends and Members of the Foundation:

We would like to extend an invitation to all grandparents of children with ichthyosis to participate in a Grandparents' Forum on the bulletin board on www.ichthyosis.com. We will use this forum to share our experiences and to share our expertise and mount a campaign to fund raise on many different levels, to fund research to help find a cure. We can pool our resources, bounce around ideas, outline and detail all our efforts, and thus, expand our efforts.

Log on to www.ichthyosis.com and click on Bulletin Board to participate. You do need to register with the ichthyosis.com site to use the bulletin board function. For those grandparents who do not have access to the Internet, but want to participate, please contact Maureen in the Foundation office (1-800-545-3286 or 215-619-0670) and she will put you in touch with one of us. Together, we can do great things.

Donna Clare
Marietta, GA

Edie Wohlgang
Mayfield Village, OH

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20 % urea + AH lotion for F.I.R.S.T. member

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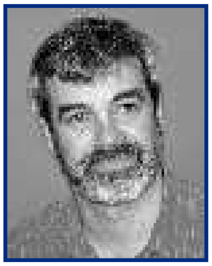
2006 Research Grant Recipients

Dr. Dennis Roop is a professor of Molecular and Cellular Biology and Dermatology at Baylor College of Medicine in Houston, Texas. He is also the Director of the Center for Cutaneous Molecular Biology at Baylor and the leader of the Cancer Biology program, Cancer Center, Baylor College of Medicine. Dr. Roop received his education at Berea College, Berea, Kentucky, the University of Tennessee at Knoxville, and Baylor College of Medicine. Dr. Roop enjoys a distinguished career in cellular and molecular biology research and is the recipient of several notable research and lecture awards. Dr. Roop's work has contributed to the scientific community's knowledge of skin function, genetic skin disease, and skin cancer.

Dr. Jiang Chen is an instructor at Baylor College of Medicine. Dr. Chen received his education at Henan Medical University, the University of Heidelberg, and Baylor College of Medicine. Dr. Chen's work has focused on tumor development, wound healing, and genetic skin disease. Dr. Chen received a grant from F.I.R.S.T in 2003 through the Dermatology Foundation Grant Program for his preliminary work on “Testing Gene Therapy Approaches for Epidermolytic Hyperkeratosis.”

Dr. Judith Fischer is the Head of Dermatologic Disease Projects at the Centre National de Génotypag in Evry, France. Dr. Fischer received her education at the Faculty of Medicine Homburg and Faculty of Medicine Marburg, St. Louis Hospital in Paris, and the Centre National de Génotypag. Dr. Fischer's research career has focused on genetic studies of psoriasis and rare genetic skin diseases.

Dr. William Rizzo is a Professor of Pediatrics at the University of Nebraska Medical Center, Omaha, Nebraska. Dr. Rizzo received his education at Northwestern University, Evanston Illinois, the University of Illinois at Chicago, and Johns Hopkins University, Baltimore, Maryland. A pediatrician by training, Dr. Rizzo has a strong interest in medical genetics and in Sjögren-Larsson (SLS) syndrome in particular. His research into the genetic and metabolic pathways for SLS spans almost twenty years.



Dr. Eli Sprecher is the Head of the Laboratory of Molecular Dermatology, Department of Dermatology, Rambam Medical Center, Haifa, Israel, and is on the Faculty of Medicine, Technion, Israel Institute of Technology in Haifa.
Dr. Sprecher received his education at the Hebrew University, Jerusalem, Hadassah Medical School, Jerusalem, Thomas Jefferson Medical School, Philadelphia, Pennsylvania, and Rambam Medical Center. Dr. Sprecher's research has focused on a variety of genetic skin diseases and inherited conditions. He is the recipient of a variety of research awards and prizes.

Dr. Dina Ron is an Associate Professor on the Faculty of Biology, Technion, Israel Institute of Technology, Haifa, Israel. Dr. Ron received her education at Ben-Gurion University of the Negev, Israel, and did her post-doctoral training at the National Cancer Institute of the National Institutes of Health, Bethesda, Maryland. Dr. Ron's research had focused on cellular and molecular biology.

Correspondence Corner

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Dear Jean:

Thank you very much for the Distinguished Service award. I am honored to receive it. I'm sorry that I wasn't there to accept it in person. My husband and I, along with our children (Samuel, age 7, and Jenna, age 5), have been selling bracelets for EHK research to family, friends, and members of the community. We will be sending a check to you next month for the profit from the sales. We figure that every little bit helps!

Thanks again,

Julie Pratt
Brattleboro, VT

Foundation Receives Two Gold Triangle Awards

The American Academy of Dermatology (AAD) awarded the Foundation for Ichthyosis & Related Skin Types two Gold Triangle Awards at its summer meeting, July 27, 2006 in San Diego, California.

The prestigious Gold Triangle Awards are given to health community organizations, the media, industry, and public service organizations, as well as individuals, for effort in promoting public awareness about dermatology and encouraging the care of skin, hair, and nails. The Foundation received awards in the Health Community Organization category for our online Health Education Modules for Healthcare Professionals and for our new teen DVD, "*Living with Ichthyosis: A Teenage Perspective*." Dermatologist Stephen P. Stone, MD, president of the American Academy of Dermatology and host of the 2006 Gold Triangle Awards said, "Our Gold Triangle Award winners are to be commended for their efforts in increasing public awareness of dermatology and skin disease, and greatly impacting the lives of those affected physically, emotionally, and financially by these conditions."

Local Foundation members Jennifer and Alan Leblang accepted the awards on the Foundation's behalf at an elegant dinner ceremony held at the San Diego Marriott. The Leblangs have a son affected by EHK.

Alan and Jennifer Leblang accept a Gold Triangle Award from actor Robert Wagner.

In accepting the awards, Jennifer told the audience that her son has a rare skin condition, EHK, and that when he was born the doctor told her that he would live in a bubble. Jennifer expressed thanks to the Foundation for having educational materials available to help parents understand their child's disease and how to treat it.

Jennifer and Alan purchased the teen DVD shortly after it became available as a resource for their son. Their son has since made friends with one of the teens in the video. Jennifer said, "They now correspond by email and phone. When my son is having a bad day, he can call someone that totally understands what he is going through. We may be one family, but these resources mean a lot to us. It is just another way the Foundation is connecting its members."

Jennifer and Alan thanked the AAD for recognizing the hard work of the Foundation.

Alan and Jennifer Leblang accept two awards on the Foundation's behalf.

The Foundation sincerely thanks Alan and Jennifer for accepting the award on our behalf and would like to thank again all the individuals who participated in the creation of these valuable resources.

<u>Online Education Modules for Health Professionals</u>

Dr. Bari Cunningham and Dr. Mary Williams provided the slides, photos, and audio lectures for this series. Terry Tormey introduced us to ePharmaLearning, an online technology company based in Conshohocken, PA. ePharmaLearning provided the tools, production, and hosting service for these educational modules.

<u>Living with Ichthyosis: A Teenage Perspective</u>

Jim Bode, Jasmyn Bowie, Margaret Frost, Shawna Grady, Matt Gray, Alyssa Kosmer, Ryan Licursi, Bailey Pretak, Tom Seman, Zebulun Sipper, and Katie Smith offered their perspectives on living with ichthyosis for the creation of our teen DVD. These wonderful young people traveled with their families to Pennsylvania for the weekend filming at the home of Program Director, Maureen Tierney.

Director / producer Justin Tormey, of ASAP Productions, New Hope, PA, and his crew spent many hours filming and editing to get the best possible resource for teens affected by ichthyosis.

A grant from Operation Good Neighbor Foundation underwrote the production costs of the DVD, as well as some other expenses. An additional grant from LaRoche-Posay, makers of Lipikar Baum, covered the cost of hotel accommodations and food during the weekend.

Wonderful, collaborative relationships like these help the Foundation offer award-winning resources and services to families affected by ichthyosis.

F.I.R.S.T. Volunteers Excel at Grassroots Fund Raising

Several Foundation families achieved amazing success this year with a variety of local fundraising efforts. Their creativity and hard work generated over $75,000 for ichthyosis.

Project Save Our Skin

Project Save Our Skin, a non-profit organization founded by Texas attorneys, Cort DeHart and Elizabeth Berry, hosts annual events to raise money for ichthyosis research. Cort and Elizabeth's daughter is affected with lamellar ichthyosis.

Each spring, the Broken Oar Tarpon Invitational is held in Key West, Florida. The event is a four-day, guided catch-and-release fly fishing tournament. This year's event raised approximately $28,000.

Each year, Cort's law firm, DeHart Crockett, P.C., hosts its own charity event called Crawfish in the Park. Held in April, the event boasts a huge crawfish boil, live music, and children's activities. This year's proceeds amounted to $10,000. Attendance and donations for both events continue to increase each year.

Ice Cream for Ichthyosis

The F.X. Hegarty Student Council of F.X. Hegarty School in Island Park, NY, sponsored a fund raising campaign for ichthyosis during the month of May that concluded with an ice cream sale on June 1, 2006.

The student council is made up of third and fourth graders, but the whole school participated in the event. Every class had a collection container in the classroom and students made donations of cash, checks, or coins for a "Funds for F.I.R.S.T." campaign. An "Ice Cream for Ichthyosis" sale brought the accumulated proceeds to $2, 672.86!

"Ice Cream for Ichthyosis" whets everyone's appetite.

Foundation members, Denise and Marc Benedetto, were an integral part of this event. Denise is a kindergarten teacher at F. X. Hegarty and a co-advisor to the student council. Denise has been a coordinator in the Ichthyosis Support Network for many years and is a dedicated volunteer. Her son, Marc, (who will be entering fourth grade in the fall) is affected with lamellar ichthyosis.

J.R. Drouhard's Basketball Benefit Tournament

J.R. Drouhard coordinated a basketball tournament in his hometown to help raise the money he and his mother needed to attend the National Family Conference in Atlanta, Georgia. J.R. is fifteen years old and has lamellar ichthyosis.

J.R.'s efforts were so successful, he and his mother, Jill Kimple, were able to attend the family conference and donate an additional $300 to ichthyosis research.

J.R. and his friends in Conway Springs, Kansas, had a great time while raising money for a good cause. J.R.'s mom, Jill, says, "We are so glad to have F.I.R.S.T. as a resource. Thank you all for all you do."

A Man - A Plan - A Bike

Jake Brukhman and his friend, Eyal Wellisch, set off on July 3, 2006, on a journey that would take them 2000 miles and 6 weeks to accomplish. Their plan was to bike a route along the Atlantic Coast, starting from Cranford, New Jersey, and ending in Miami, Florida. Besides the personal accomplishment for both men, their goal was to raise funds for ichthyosis research and raise awareness of ichthyosis.

Jake Brukhman's close friend, Courtney Shell, and her husband gave birth to a daughter with Netherton syndrome last year. Soon afterward, Brukhman and Wellisch decided the bike trip would double as a fund raiser for research.

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Executive Director's Report

Dear Friends & Members of F.I.R.S.T.

The past few months have been very busy for our Foundation. The family conference in July, in Atlanta, was a huge success. Close to 300 members attended this year and I am excited to begin planning the 2008 event. Please visit our website or look for the 2008 location in an upcoming issue of the newsletter.

On August 1, we officially moved to a bigger, better and more professional office space, allowing us to expand our operation. And we didn't waste any time! I am pleased to announce the addition of our newest employee, Bonnie Thompson. Bonnie officially joined our team in mid-September as our part-time Executive Assistant. Bonnie's role is to assist the Foundation in many areas, but with a special emphasis on fund raising. The success of our in-house research grant program in its first year clearly demonstrates that there are many interested investigators ready and willing to study the ichthyoses. Our Foundation must meet that need by raising the necessary dollars to fund these worthy projects. And, as usual, we must continually raise money to maintain the exceptional services and programs provided to our members and the ichthyosis community. Bonnie will provide that extra help to generate more funds, assist our members with grassroots events, and strengthen relationships with existing donors while building new ones.

More exciting things will be taking place in the next few months. I will be taking a few weeks off for the birth of my third child, due to arrive at the end of October. Plans are underway to develop a new five-year strategic plan. We adopted our first plan in 2002 and will have achieved all of the major goals we set out to accomplish by 2007. It is time, once again, to poll our members and map out which areas the Foundation should concentrate on and establish goals to achieve by 2013.

Over the past few years, there has been some confusion among our members and friends about the two main ichthyosis websites on the Internet. To try to alleviate some misconceptions, I wanted to take the time to clarify each website's main objective. The websites are not in competition with each other in any way. In actuality, they complement each other in different ways, each serving a specific purpose.

> www.scalyskin.org – This website is owned and managed by our national office. Our objective is to provide accurate information about the ichthyoses and related skin types and ways to utilize and/or support the Foundation's programs, resources, and services. As a national 501(c)(3) organization recognized by the IRS, the Foundation must be sure to comply with their regulations so as not to jeopardize our non-profit status. The Foundation must also be sure not to favor any company, product, or treatment.
>
> www.ichthyosis.com – This website is privately owned and managed by a generous husband-and-wife team in Washington, DC. This site serves the personal side of ichthyosis. It provides recommendations of skin care treatments, failures and successes through personal stories, and individualized perspectives into living with ichthyosis. This website also provides a forum to exchange thoughts, ideas, and suggestions through bulletin board postings and a chat room.

A research appeal should have recently arrived on your doorstep. This appeal is an opportunity to contribute directly toward ichthyosis research. Please know that any size donation is greatly appreciated and will make a difference. I hope to receive your support.

Finally, our Board Development Committee will be reviewing applications for the 2007 Board of Directors. If you are interested, or know someone who would make a good candidate for the Board, please let me know. The Foundation is only as strong as its membership, so I encourage you to contact me at jpickford@scalyskin.org or call at 215.619.0670.

Jean

With warmest regards,

Jean R. Pickford

Are you interested in receiving the newsletter via email? Let me know by emailing me at jpickford@scalyskin.org or calling 215-619-0670.

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The men reached their destination on August 26, 2006. To date they raised $970 for ichthyosis research.

Even though Courtney Shell and Jake Brukhman have been friends for years, Courtney said she was overwhelmed by the gesture and by how giving people could be.

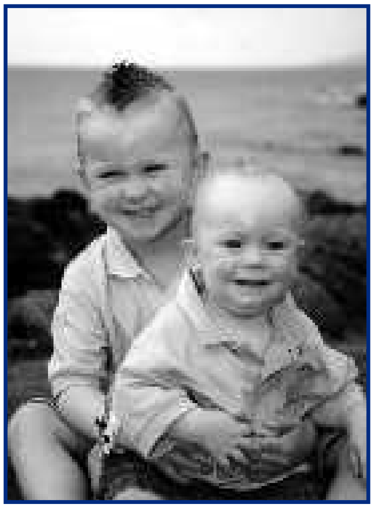
Napa Valley Style

Napa Valley residents and Foundation members Josh and Amie DeGarmo hosted a golf tournament to benefit ichthyosis on July 12, 2006, at the Silverado Country Club in Napa, California. The DeGarmo's sons, Jaydn and Carson, are affected with lamellar ichthyosis.

Key volunteers Richard and Lois DeGarmo (Josh's parents) and (friend) Robert Larson helped to organize the event and attract local businesses as sponsors.

Sixty-three golfers headed out into the California sunshine in noontime shotgun start. A Silent Auction and dinner followed the rounds of golf with 103 guests in attendance. Jim Griffin, a California native and founding member of the Foundation, delivered heartfelt remarks. This event raised over $12,000 in proceeds.

Josh and Amie plan for this to be an annual event and would be happy to help other Foundation volunteers plan a similar event in their communities.



Jaydn and Carson DeGarmo

Illinois Golfers Bear the Heat for Ichthyosis

Foundation members Kathy and Matt Kouba joined forces with their friends, family, and community in Plainfield, Illinois, to host a golf outing at High Point Golf Club, Essex, IL, on July 15, 2006. Sixty-four golfers enjoyed the play on all 18 holes in spite of the ninety-degree weather.

Kathy and Matt's two children, Alicia and Ben, are affected with CIE. Their intention in hosting this event was not just to raise money for the Foundation but also to educate their community about ichthyosis. The Koubas have had many of the experiences that are common to parents of children with ichthyosis; the lack of knowledge in area doctors, the danger of overheating in warm weather, and the stares and comments from people in public places. Two local newspapers interviewed the Koubas about the golf outing and the family's experience with ichthyosis. As a result, their community is more informed, local businesses were motivated to generously support the golf outing, and the family has connected to other people in their area who are affected by ichthyosis.

The Kouba kids get in on the golf fun.

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

Awareness &
Fundraising
Guide

The Foundation's Grassroots Fundraising Guide can help you plan and carry out these and other kinds of community fund raising events. Please contact the office for your copy today.
info@scalyskin.org
215-619-0670
1-800-545-3286

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F.I.R.S.T. Volunteers Excel

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Patience and Persistence

Edie Wholgang, Foundation member and grandmother to a grandson, Adam, with EHK, took on the daunting task of volunteer grant-writing. Writing grants to foundations and corporations is difficult work with the receipt of many more "no" answers than "yes' answers. And sometimes that yes answer never comes through.

Edie's persistence paid off, however, with the receipt of a $2,500 grant from the Stiefel Foundation.

Edie Wholgang receives recognition at the Family Conference

'Can Do' Volunteer

Foundation member and Georgia resident, Donna Clare, solicited Atlanta businesses and merchants for donations to the Foundation's National Family Conference held in Atlanta this summer. Not only did Donna receive so many donations that almost every family attending the conference went home with a door prize; she also acquired a lovely array of prizes that went into a cash raffle. Lucky winners went home with a beautiful gold bracelet, a Razor scooter, hair care and accessory basket, or a one-week stay in a Florida condominium. The raffle proceeds totaled $1,254.

Donna is the proud grandmother of Grace, who is affected with lamellar ichthyosis. We found out that Donna's nickname is "Donna Do." She and all our other grassroots volunteers are great examples of what the power of one person can do.

The Foundation, its staff, and the Board of Directors are very grateful to all of these volunteers for their hard work and generous contributions.

Correspondence Corner

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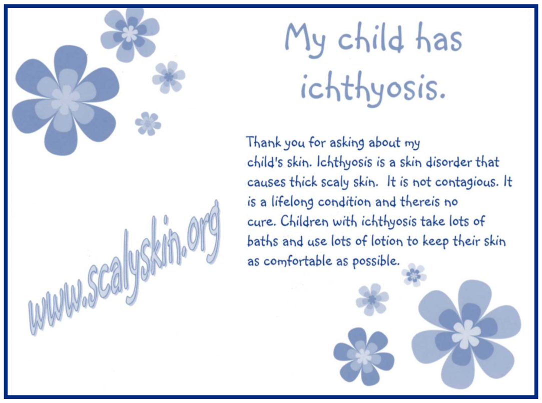
Dear Jean:

I printed up some cards for myself and Evan. I get tired of all the questions sometimes. Vista Print (www.vistaprint.com) sent me a great offer, so I printed up some general cards for other moms. I hope they are helpful.

Sincerely,

Kimberly Mayone
S. Portland, Maine



My child has ichthyosis.

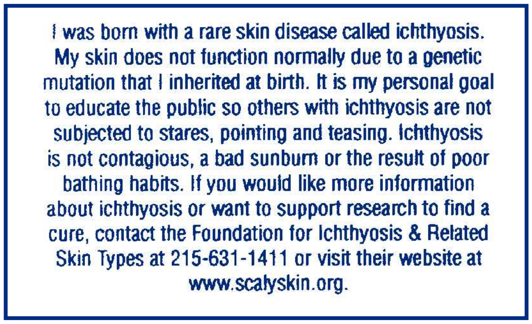
Thank you for asking about my child's skin. Ichthyosis is a skin disorder that causes thick scaly skin. It is not contagious. It is a lifelong condition and thereis no cure. Children with ichthyosis take lots of baths and use lots of lotion to keep their skin as comfortable as possible.

www.scalyskin.org

Kim's card

Either card can easily be adapted to your family's needs with computer programs designed to create postcards or business cards. You can find printable paper for postcards or business cards at your local office supply store.

Please call Maureen in the office, 215-619-0670, 1-800-545-3286, or e-mail info@scalyskin.org, for copies of either card.



I was born with a rare skin disease called ichthyosis. My skin does not function normally due to a genetic mutation that I inherited at birth. It is my personal goal to educate the public so others with ichthyosis are not subjected to stares, pointing and teasing. Ichthyosis is not contagious, a bad sunburn or the result of poor bathing habits. If you would like more information about ichthyosis or want to support research to find a cure, contact the Foundation for Ichthyosis & Related Skin Types at 215-631-1411 or visit their website at www.scalyskin.org.

Foundation awareness card

Patient Advocates, Investigators and Physicians Are Important Allies

Coalition of Skin Diseases Remains an Important Society for Investigative Dermatology Partner

The Coalition of Skin Diseases (CSD) is a voluntary coalition of patient advocacy groups (which includes the Foundation for Ichthyosis & Related Skin Types) addressing the needs and concerns of millions of people whose lives are affected by skin diseases. The CSD works to accomplish its mission of advocating on behalf of individuals with skin disease by:

- Supporting basic science and clinical research.
- Fostering physician and patient education.
- Generating awareness of skin disease.
- Supporting the growth of member organizations through the sharing of mutual concerns which may increase the pace of discovery or a cure while improving the quality of life for those affected.

The CSD works closely with the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) to coordinate research efforts for all skin diseases. Members of the coalition meet annually with members of Congress and testify when selected before the appropriations committee to educate Congress and the public on the seriousness of skin diseases while stressing the need for increased medical funding.

The Society for Investigative Dermatology (SID) has long benefited from its work with patient support and advocacy groups. The CSD, originally the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR), was formed in 1984 as a voluntary coalition. The coalition was originally composed of 24 national lay skin disease organizations representing people with skin diseases throughout the United States and the world. Together with members of the American Academy of Dermatology (AAD), CPS-SDR and SID representatives began to work together, lobbying on Capitol Hill to fund skin research.

Today, the CSD, AAD and SID conduct Skin Disease Research Day, now in its third year, to speak to elected officials in Washington. This advocacy event leverages the synergies in the missions of all the organizations, focusing nearly 60 participants who are passionate about finding treatments and cures for the nearly 80 million patients suffering each year. The partnership confirms that patients are the best possible advocates to leverage the skin disease research agenda.

Coalition partners translate the scientific terminology of cutaneous biology into a language that penetrates far deeper than journal articles, abstracts or posters into the minds and hearts of Congress. They transform the scientific pursuit of skin research into the most important language, that of humanity. As the voice of patients from around the country, they eloquently express the reality that , unless major research efforts are undertaken, important advances in research may go undiscovered, leaving so many with chronic illness sentenced to a diminished quality of life.

To see the complete list of CSD groups and links, please visit the SID website at:
www.sidnet.org/Coalition_Skin_Disease.asp.

Reprinted from SID*news, a quarterly publication of the Society for Investigative Dermatology, early summer 2006.*

The Foundation for Ichthyosis & Related Skin Types has been an active member of the Coalition of Skin Diseases for 10 years and participates annually in Skin Disease Research Day.

2007 Testimonial Dinner Honoring Drs. Gloria & James Graham and Dr. Peyton & Janet Weary

On Thursday, February 1, 2007, the Foundation will host a Testimonial Dinner fund raiser in honor of Drs. Gloria & James Graham and Dr. Peyton & Janet Weary. The Graham's and Weary's have been devoted physicians and advocates for patients affected with ichthyosis for many years. Although they are humbled by this recognition, their main objective is to help individuals and families affected with ichthyosis and raise money to support the mission of the Foundation.

The event will take place at the exclusive Cosmos Club in Washington, DC on the eve of the 65th annual American Academy of Dermatology convention. If you would like to honor them by attending the dinner or making a contribution, please email the office for more information at info@scalyskin.org.

Camp Horizon 2006

By Paula Heinzmann, Lamellar Ichthyosis
Paula.Heinzmann@gmail.com

> "I don't want to go home. I like it here. Nobody stares at me; nobody teases me. I want to stay here forever."
>
> "Won't you miss your family?"
>
> "They can come here. My family and friends and all of you can live here."

A conversation with one of my campers on the last night of another magical week at camp reminded me what a sacred place I have been a part of for the last several years.

Imagine someone with ichthyosis being transported from her everyday life of striving to look and feel "normal" to a world where she *is* "normal". This teleport, from the world as we know it, to a place that, to me, could very well resemble my heaven, happens all in a few short hours on the bus ride from Harrisburg, PA to Camp Horizon.

Camp Horizon is a camp for children with skin disorders. The American Academy of Dermatology provides these children with the opportunity to feel like just another kid at a summer camp for one week a year.

The minute the bus arrives at Camp Horizon, campers scatter around the grounds, reuniting with the best friends they haven't seen in a year and meeting new friends. Talk of the adventures to come that week can be heard: fishing, biking, rope climbing, arts and crafts, baking, and even a theme park.

The 'ichthyosis gang' poses for a picture to add to their memory books.

There are, of course, a few campers, hesitant to be there for the first time, or maybe even a little homesick. But those same shy children you see the first day of camp are almost always the ones least willing to leave camp only a short week later. The transformation is amazing and is represented by new-found confidence and higher self-esteem.

Hunter Steinitz and Paula Heinzmann

Not everyone at Camp Horizon has ichthyosis, but all of the campers and a majority of the counselors have some type of skin disorder: Alopecia, epidermolysis bullosa, and eczema to name a few. The variety of skin disorders challenges the campers to see beyond the disorder, just as they want their peers at home to see beyond theirs.

Camp Horizon is one of the few places where best and lifelong friends are made in a week. The shared experience of growing up "different" brings everyone at camp together. The growth and development experienced at camp is truly magical. I encourage everyone to share in the experience.

I was 17 when I first went to camp. I had never met anyone else with ichthyosis until my first day at Camp Horizon. I distinctly remember walking with a girl similar in age to me, looking down, and saying out loud, "Your toes wrinkle the same way mine do when I walk". It's the little things like that, things that could only happen at camp, that get me through the year.

Caregivers Manual 2006 Edition Now Available

At long last, the *Release the Butterfly: A Handbook for Parents & Caregivers of Children with Ichthyosis* has been updated and is available to our members and public. Thanks to generous funding from Sovereign Bank, the booklet was updated with the help of several parents and dermatologists. The 40-page booklet focuses on the early stages of dealing with a newborn to practical matters of everyday care. This booklet will be helpful to anyone who is caring for a young child with ichthyosis, including day care providers, grandparents, and other relatives. The cost of the booklet is $5.00 plus shipping. To place an order, visit our website, www.scalyskin.org and click on the "Kiosk" or contact the office at 215-619-0670, or email at info@scalyskin.org.

A RESEARCH STUDY OPPORTUNITY

The experiences of patients with <u>obvious physical differences</u>, when participating as a teaching case in the genetics clinic

You are invited to be part of a study sponsored by the Johns Hopkins Medical Institutions and the National Institute of Health. This study involves a one-time interview of patients regarding their experiences in the genetics clinic or with a genetics professional. The interview will be conducted by telephone.

Specifically, the study will explore the following:

- The experiences of being involved in informing others about one's condition
- What types of factors make the genetics clinic experience more or less comfortable for the patient
- Better/different ways that genetics health professionals could conduct clinic practices

To participate in this study, you must have an **obvious physical difference**. You must also have attended a genetics clinic or been seen by a genetics professional in the last **5 years**.

If this describes you and you are interested in learning more about the study, please call Jamie Dokson at 1-866-294-9078 or email (jdokson@jhsph.edu) with your name, phone number or email address, and the best time to contact you.

We hope that you will consider being in this study.

Sincerely,

Barbara Bowles Biesecker, M.S., C.G.C
Genetic Counselor & Associate Investigator
Director, Genetic Counseling Training Program
Social and Behavioral Research Branch
National Human Genome Research Institute, NIH

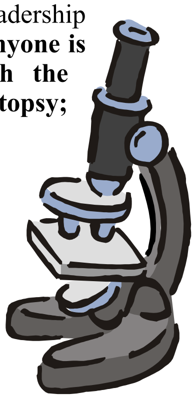
Jamie K. Dokson
Genetic Counseling Student
Johns Hopkins University
National Human Genome Research Institute

Netherton Syndrome Research Study

Netherton syndrome (NS) is a very rare inherited disease. Patients have severe skin disease starting early in life (known as congenital ichthyosis), hair abnormalities (called bamboo hair), and severe food allergies. Taking care of several children with NS in our immunology outpatient clinic at Children's Hospital in Seattle, we realized that, especially in early childhood, children with NS are more often sick with infections than healthy children. Because of sepsis and recurrent infections of skin, ears, and the gastrointestinal and respiratory tracts, we started to evaluate the immune system of NS patients. We found that patients have a problem making specific antibodies to some antigens. Antibodies are made against antigens (term for germs causing infections) to defend us from getting sick. Therefore, we started several of our patients on immunoglobulin therapy. Intravenous supplementation with immunoglobulin (IVIG) provides antibodies collected from healthy human donors, in order to prevent infections in patients who are unable to make sufficient antibodies on their own. Our NS patients responded with improvement of their skin abnormalities and decreased frequency of infections after initiation of IVIG treatment.

In our research program, our aim is to understand the basic mechanism of this abnormal immune status. What is known about the underlying cause of this disease so far? A few years ago a gene identified in the human DNA (called *SPINK5*) became associated with NS. Patients that have mutations (defects) in this gene cannot produce functional amounts of a protein that is normally produced by this gene. The lack of expression or function of this protein (called *LEKTI*) is most likely the cause of the dysregulation in the immune system. Not all cell types of the human body produce this protein. *LEKTI* is known to be mainly found in the surface of the skin, with its function not fully understood. Another organ where *LEKTI* is expressed is the thymus. The thymus is located in the chest in front of the heart. The thymus has a central role in immune regulation and education of different lymphocyte types (cells in the blood responsible for the function of the immune system). To assess in more detail the function of *LEKTI* in the thymus, we have to study the thymus in individuals with NS who died and had an autopsy, since biopsies of the thymus are difficult to obtain from living individuals.

Therefore, we would like to ask you and the readership of the Ichthyosis Foundation magazine: **if anyone is aware of an individual who died with the diagnosis of NS and had a post mortem autopsy;** please, contact us with the info given below. Pathology departments store tissue for a long time. This would make it worthwhile to check for stored samples even if the patient had died many years ago. We would contact the pathology department after obtaining permission.



We know that this is an uncomfortable request. However, we ask because of tremendous potential contribution to the understanding of NS leading to the potential of enhanced quality of life for NS patients in the future and would greatly appreciate your cooperation!

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Please, feel free to contact us also in regards to any other questions you might have related to NS.

Kind regards,

Hans D. Ochs, MD
Ellen D. Renner, MD
Professor of Pediatrics

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Genetic Skin Disease Clinic Opens in Alabama

The University of Alabama at Birmingham has opened a genetic skin disease clinic. The clinic, which is open one Thursday a month, offers evaluation of family history, examinations, specialized diagnostic testing, comprehensive treatment programs, genetic counseling, and opportunities to participate in clinical research projects.

For more information, contact Dr. Bruce Korf or Dr. Amy Theos at 1-800-UAB.MIST (1-800-822-6478), or email mist@uabmc.edu. Read more about the clinic at www.health.uab.edu/default.aspx?pid=83239. If you do not have access to the Internet, please contact Maureen in the Foundation office for a copy of the information, 215-619-0670 or 1-800-545-3286.

We've Moved

We are settled into our new office space.
Please make note of our new contact information:

F.I.R.S.T.
1364 Welsh Road, G-2
North Wales, PA 19454
215-619-0670
215-619-0780 Fax

Our toll-free number, website, and email remain the same.
1-800-545-3286
www.scalyskin.org
info@scalyskin.org

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.

F.I.R.S.T.
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