Ichthyosis Focus

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# Genetic Testing for the Ichthyoses

Sherri Bale, PhD, and Cheryl Scacheri, MS, CGC GeneDx, Inc.

## Introduction

Perhaps you or a family member have a diagnosis of ichthyosis and are looking for answers to questions like, "What type of ichthyosis do I have," or "What are the chances that I will pass ichthyosis on to my child?" Sometimes these answers may be provided by routine tests, skin biopsy, or physical exam. At other times genetic testing provides the information that may not be available through any other test or exam. Specifically, genetic tests may be able to determine precisely which type of the many sub-types of ichthyosis a person has. Or it may be used to pinpoint the way ichthyosis is inherited in a family, providing information about each relative's chance to have a child with ichthyosis. Lastly, genetic test results can open the door to a number of reproductive options, including testing embryos before a woman becomes pregnant and other procedures that can be performed during pregnancy. This article will explain what genetic testing is, the reasons one might choose to have a genetic test, what testing can and cannot tell you, and resources you can use to find out if a test is available and where it can be done.

## What is genetic testing?

Genetic tests involve an examination and analysis of a person's DNA. DNA is a chemical in each cell of the body, which provides instructions for making the proteins and other molecules we need for normal body function. To create this massive textbook of instructions, 3 billion bits of DNA (referred to as "bases") are strung together. Most of these bases are the same from one person to the next. In fact, 98% of my DNA is identical to yours! It is the other 2% that makes us look different from each other, and some of those differences are responsible for causing genetic diseases. If a change is present in a gene and it causes a molecule to be made incorrectly, that change is called a "mutation." Genetic testing is a means of looking at a person's DNA to identify the mutation that causes their condition. However, since there is so <u>much</u> DNA to look through, researchers must already know which of the many genes to examine in a particular disease before genetic testing can be offered. Also, a person's doctor must have a good idea of what the clinical diagnosis is for that patient. Genes have not yet been identified for every condition, but there are many genes for the ichthyoses for which we can test. Laboratories often

*continued on page 3*

**Tests currently available for the ichthyoses include:**

| Disease name | Gene(s) involved | Inheritance pattern |
|---|---|---|
| CHILD Syndrome | CDPX2, NSDHL | X-linked |
| Conradi-Hunermann Syndrome | EBP | X-linked dominant |
| | PEX7 | Autosomal recessive |
| Autosomal recessive congenital ichthyosis (lamellar or CIE) | TGM1 | Autosomal recessive |
| Ichthyosis Bullosa of Siemens | KRT2e | Autosomal dominant |
| Epidermolytic Hyperkeratosis | KRT1, KRT10 | Autosomal dominant |
| Sjogren-Larsson Syndrome | FALDH | Autosomal recessive |
| Vohwinkel Syndrome | GJB2 | Dominant and recessive |
| Erythrokeratoderma Variabilis | GJB3, GJB4 | Autosomal dominant |
| Darier Disease | ATP2A2 | Autosomal dominant |
| Hailey-Hailey Disease | ATP2C1 | Autosomal dominant |
| KID Syndrome | GJB2 | Autosomal dominant |
| Multiple Sulfatase Deficiency | SUMF1 | Autosomal recessive |
| X-linked Ichthyosis (steroid sulfatase deficiency) | ARSC1 | X-linked |
| Epidermolytic Palmar-Plantar Keratoderma | KRT9 | Autosomal dominant |
| Pachyonychia Congenita | KRT16, KRT17 and KRT6a, KRT6b | Autosomal dominant |
| Netherton Syndrome | SPINK5 | Autosomal recessive |

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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 Friends:**

I am 57 and have ichthyosis vulgaris, diagnosed at age 8. It is not "severe," but if I leave it untreated in winter my skin stiffens and cracks. I use *Xenolin Lotion,* 5% lanolin and 6% propylene glycol. It minimizes my scale build-up and improves flexibility. And it doesn't feel oily or gummy. It's also inexpensive to use if you need it all over, $5.00 for 8 oz. Not in stores, available online from arbilab@earthlink.net.

**Rick Bibbs**

**Dear Jean:**

I wanted to drop you a short note. I have been a member since May 1981, when I attended my first meeting of the National Ichthyosis Foundation in New York. I have found F.I.R.S.T. a valuable source of information and research over the years.

Recently, I have been experiencing health problems that require specialized medical treatment. What I wanted to pass on to you is that I have been working closely with the Dermatology departments at Milton S. Hershey Medical Center in Harrisburg, PA, and Johns Hopkins University in Baltimore, MD. I was pleased that both organizations were aware of, and more importantly, referred me to F.I.R.S.T.!

Of course, I have known about F.I.R.S.T. longer than the doctors recommending this valuable support network have had their medical degrees! I actually diagnosed myself with Congenital Ichthyosiform Erythroderma (CIE) based on information from Dr. Mary Williams' research, and had it confirmed at the National Institutes of Health in 1986. But the word is getting out there; keep up the good work!

Sincerely,

**Barbara C. Straw**
**Mechanicsburg, PA**

## Genetic Testing

*(continued from page 1)*

add tests as researchers identify new genes. Up-to-date information about available testing and the contact information for laboratories can be found at an excellent government-supported Web site: www.genetests.org.

**What a genetic test can tell you, your family, and your doctor.**

Your physician may have reasons for ordering genetic testing and may discuss it with you, or you may wish to bring it up with your doctor yourself. Sometimes, when a patient has ichthyosis, they have features that could fit with several different diagnoses because there are overlapping symptoms between the subtypes. Doctors frequently use genetic testing to help define which ichthyosis a person actually has. This may help them to treat and manage the patient.

Another reason to have a genetic test is if you or a family member wants to have children. Genetic testing, which would ideally be performed first on the person with ichthyosis, is often helpful in determining a person's, and their relative's, chances to have a baby with ichthyosis. In some cases, the doctor can determine this by looking at the family tree. However, genetic testing may be recommended if the inheritance pattern is unclear or if you or a family member is interested in reproductive options such as genetic diagnosis before implantation or prenatal diagnosis.

**How is genetic testing performed?**

All genetic tests are very specialized and require highly trained scientists and technicians. In this article, we discuss only DNA-based molecular genetic testing (and do not address the microscope-based test commonly performed for steroid sulfatase deficiency). Samples must be specially prepared to separate out the DNA from a blood, skin, or cheek brush sample from the patient. Genes are usually tested one at a time and sometimes certain regions of a gene, called "mutation hot spots," are tested first. Mutation hot spots are places in the gene where the majority of mutations are found. For most tests, the gene is first prepared for testing by using a technique to make millions of copies of the different regions of the gene. Then, the laboratory may use other tests to look at hot spots. In most situations, however, much of the entire gene must be analyzed, base by base. This is done by a technique called "sequence analysis." With sequence analysis, another piece of equipment is used which reads each letter of the DNA pattern, or sequence. Remember there are 3 billion of these in total, and a single gene usually contains several thousand bases. The recipes that genes encode are quite complex! The sequence in the patient is compared to the sequence that is usually observed in healthy people. This is done by scientists who go through the thousands of DNA letters, meticulously comparing the patient's DNA pattern to the normal sequence. It is almost like editing the chapter in a book, letter by letter. A change is identified when there is a difference between the normal published sequence and sequence observed in the patient. Changes that haven't been seen before must meet rigorous criteria before they are deemed responsible for causing ichthyosis in a patient. The reports issued by laboratories are written for the referring physicians, and an interpretation of the findings is always provided. Once a mutation is identified in a family, other family members can have carrier testing specifically to look for that mutation, and the reproductive options outlined below can be discussed.

**Testing on a pregnancy or before a pregnancy is achieved.**

Genetic testing during a pregnancy, called "prenatal diagnosis," has been around for decades and usually involves one of two procedures to obtain a sample from the pregnancy. Before becoming pregnant, it is critical to get started by testing the person in the family who has ichthyosis. Once the mutation in the family is known, genetic tests are performed on the DNA from the developing baby and can tell you if the baby has inherited the mutation. Both of the procedures have a small chance of complications, which should be discussed with your doctor or genetic counselor. Amniocentesis is the more common procedure. It is performed around 16 to18 weeks of pregnancy and the doctor uses a needle to obtain amniotic fluid, which has some of the baby's cells floating in it. Chorionic villus sampling can be performed earlier in pregnancy, at around 11to13 weeks. The doctor obtains a piece of the placenta, which has the same genetic material as the baby, for testing. Genetic testing on a pregnancy may be done to obtain information about the baby that might be helpful for delivery or decision-making about continuing the pregnancy. "Pre-implantation diagnosis" is a relatively new technique in which embryos are created outside of the body through in vitro fertilization. Genetic testing is performed on the embryos. Only the embryos that did not inherit the mutation are transferred into the woman's womb to create a pregnancy. This method is performed at only a few specialized centers in the world. Because each of these procedures is invasive and have both benefits and limitations, it is best to have a thorough discussion with your physician and the specialists who perform them if you are considering prenatal diagnosis or pre-implantation diagnosis.

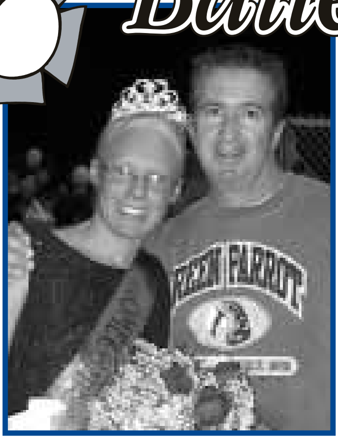
**What genetic testing cannot tell you.**

Results of genetic tests, even when they identify a specific mutation, can rarely tell you how mild or how severe a condition will be in any particular individual. There may be a general presentation in a family or consistent findings for a particular diagnosis, but it's important to know that every individual is different. The result of a genetic test may be "negative," meaning no mutation was identified. This may help the doctor exclude certain diagnoses, although sometimes it can be unsatisfying to the patient. "Inconclusive" results occur occasionally, and this reflects the limitation in our knowledge and techniques for doing the test. But we can be optimistic about understanding more in the future, as science moves quickly and new discoveries are being made all the time.

*For more information about genetic tests performed at GeneDx, visit our Web site: www.genedx.com. If you have any questions about this article or genetic tests, please feel free to contact GeneDx, Inc. at genedx@genedx.com or 301-519-2100.*

# Spotlight On ...

# Bailey Jones



*Bailey and her elementary school principal, Bob Grumley. He said he knew she would win.*

On Friday, September 5th, the Johnsonburg Area High School Senior Class chose Bailey Jones as their 2003 Homecoming Queen. Bailey, a 17-year-old senior at JAHS, has lamellar ichthyosis. She is the daughter of Roger and Tracie Pretak of Wilcox, PA.

## Bailey's Perspective

"The Homecoming Queen for 2003 is … Bailey Jones! When my name was announced over the speaker, I was overwhelmed and in total shock. Every girl's dream is to receive a crown and it was coming true for me at that moment. A mix of emotions flooded my soul. It's hard to put my feelings into words. I will forever cherish the night of September 5th.

You see, I was never popular in school, probably because of my lamellar ichthyosis. Before I started kindergarten, a couple of parents called the principal to ask him if he would take their children out of my classes. Thankfully he said, "No!" I have been called names and laughed at. Every public place I walk into, people stare at me like I'm some strange creation. It hasn't been easy.

That is why winning the title of Homecoming Queen was such a huge honor for me. At my school, the teachers vote to choose the candidates for the Homecoming Court. Then on the day of the crowning, the senior class members vote for the Queen. I thought I had a chance to get on the Court because I'm a good student, ranked second in my class. But I never dreamed that I could be chosen as the Queen! I felt inferior being on the court with five very popular girls, but I continued being my social, fun-loving, kind and friendly self. It must have paid off!

My life hasn't always been filled with great accomplishments. I've had my struggles along the way. But through it all, I've learned to love myself, love others, and love my skin disorder! I wouldn't want my skin to be "normal" because it wouldn't be me! I believe that God gave me ichthyosis to teach me compassion and to be a role model for others. I want to be a missionary to Romania and work in orphanages helping kids who are lonely and feel unloved. I am thankful that God gave me a family who brought me up like a "normal" child, and I appreciate my close-knit group of friends who don't even notice my skin anymore.

My advice to other teens and kids with ichthyosis is, "hang in there, learn to love yourself just the way God intended you to be, and love others even though they make fun of you." If you do, your life will be one exciting journey with many surprises along the way!

Bailey Jones and her escort Brandon Parana.

## Mom's Perspective

Wow! How can I put into words something so special, so amazing, and so unbelievable? What an incredible moment! The flood of emotions was so intense; joy, shock, relief, pride, thanks, and so much more. Bailey's crowning was the completion of a long journey to "Release the Butterfly." Tears of joy flowed freely from everyone who had been a part of the process.

Bailey was born on June 15, 1986, with lamellar ichthyosis. She was covered in a collodion membrane. The doctor who delivered her was not sure what was wrong, so at 20 minutes old Bailey got her first ride in a helicopter to a neonatal intensive care unit. The next day the doctor made the diagnosis of ichthyosis and she stayed in the NICU for two weeks.

Bailey was quite the attraction when we got home. Rumors spread like wildfire around here, and everyone had to check her out. Everywhere we went, people stopped and stared. As Bailey got older, she became aware of this and would hide behind me. I grew tired of the constant stares and insensitive comments, so I started being just as rude to these nosy strangers. But one day I realized that my angry reactions were only hurting Bailey! I also realized that if she wasn't my child, I might do the same thing. After all, it's a natural reaction to stare at something you've never seen before. So we tried something new; when someone stares, we smile and wave! Most people are embarrassed when they get caught staring. And for those who won't quit, we introduce Bailey, explain what she has and give them a brochure about ichthyosis (available from F.I.R.S.T.).

It hasn't always been easy. Bailey has been stared at, pointed at, and laughed at to the point of tears more than once. But it has mad her stronger and more compassionate. Through it all, I kept reminding her that NO ONE is perfect, and that true beauty comes from within. God doesn't make mistakes, and she is a beautiful, special,

*continued on page 5*

*Spotlight On ... continued from page 4*

and unique person created in His image – just like everyone else! Bailey has grown into a truly beautiful person who is kind, caring, and encouraging to everyone she meets.

My advice to other parents is to love your child and accept them just the way they are. Remember that no one is perfect. They won't learn to love themselves if they sense that you are bothered by their skin or want them to be "normal." Everyone is good at something. Find your child's strengths, talents, and gifts, and then do all you can to develop and encourage them. Get support from other parents who have been through this. Get involved in educating your community about ichthyosis. The more people know the less likely they are to stare. Teach your child how to handle questions, stares or rude comments in a kind positive way. Remind them (and yourselves) that most people are just curious. Sometimes miracles hide. It is my prayer that you will realize the miracle you are holding in your arms and "Release the Butterfly" in your own child.

("Release the Butterfly" is a phrase we found in a booklet available from F.I.R.S.T. Since then, it has been our goal to "Release the Butterfly" in every child with ichthyosis!)

# Foundation Resources

**Jane Bukaty Membership Assistance Fund**

The Foundation is now accepting applications to the Jane Bukaty Membership Assistance Fund for the next review period, which will end in July. Here is an opportunity to alleviate some of the financial burden that may be facing you or your family. It is easy to apply and you will receive a cash award, if your application is accepted. Awards from the fund are given to help with the treatment of ichthyosis only; funds cannot be used toward membership fees or Family Conference fees.

To apply: Submit a letter stating your need for help. State the amount of money you are requesting. Awards generally do not exceed $100.00. Indicate the product or treatment for which you need the funding. Describe your financial need for this product or treatment.

Email a request for an application form to info@scalyskin.org, or call 1-800-545-3286. Completed forms may be mailed to the attention of the Jane Bukaty Membership Assistance Fund, 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446. **The deadline for applications is June 30, 2004.** The awards will be given in July 2004.

*Donations to the Jane Bukaty Membership Assistance Fund are always appreciated and enable the Foundation to make this fund available to more of our members.*

**X-linked Ichthyosis Support Network**

The Ichthyosis Support Network is seeking more volunteers to act as peer counselors in the X-linked networks. A new prenatal screening test, which measures maternal estriol, in addition to other important pregnancy and fetal health markers, is detecting an increased number of suspected X-linked ichthyosis cases. Expectant mothers and their genetic counselors are contacting us for more information about this disease type. And it would be helpful to have more experienced parents to respond to their concerns and questions.

If you have X-linked ichthyosis in your family and think you could be a good support person for an expectant mother, please contact Maureen in the Foundation office, 1-800-545-3286 or email info@scalyskin.org. We need families with young school age or pre-school age children in particular.

# A Bid for a Cure

On March 20, over 150 friends, neighbors, and relatives of the Medlock Bridge community in Alpharetta, GA, gathered for a Saturday evening of fun, entertainment, and shopping. The event was entitled “A Bid for a Cure,” and included a live and silent auction. The best part about this special gathering was that all proceeds raised from the auction were designated for ichthyosis-related research. The event raised $40,000; a phenomenal success, compared to the original goal of $5000!


*Donated auction items filled more than five tables.*

Last fall, Mark and Kelly Klafter, parents of two-year-old Adam, affected with Epidermolytic Hyperkeratosis, decided they wanted to raise money for research. After meeting with friends, the group agreed to host an auction in their neighborhood clubhouse. They hoped to raise about $5000. Mark, Kelly, and their friends went to work and began soliciting auction items and the floodgates opened! Community businesses donated food, beverages, and supplies. Friends of friends were calling to donate items for the auction. The response was overwhelming! Donations of trips, art, household items, autographed sports memorabilia, plus much more came pouring in. Even the professional auctioneer donated his time.

A big part of this grassroots fundraiser was the donation of a $2500 gift certificate to a resort in Myrtle Beach, NC. The Klafters and their committee decided to raffle this prize and raised more than $12,000 in raffle donations alone. Combining this remarkable success with the bids for over 85 auction items, the group raised $40,000 for ichthyosis-related research.


*Special thanks to Mark & Kelly Klafter and their committee. Pictured, left to right: Helen Blase, Gwen Levit, Victoria Carter, Kelly Klafter, Mark Klafter and M.J. Jelleme.*

Congratulations to the Klafters, their committee of friends, neighbors and family members! Both Kelly and Mark welcome the opportunity to talk with other Foundation members about their experience and share what they learned though their first grassroots fundraiser.

## Seniors Speaking

**Dear FIRST:**

Thank you for the lead article in the Fall 2003 issue of “Ichthyosis Focus” on Lamellar Ichthyosis (LI) and CIE. You very accurately described my almost 69 years of living with LI.

My older brother was the only one of my five siblings that also had the condition. He died at age 64 of causes unrelated to LI, I think.

You correctly related how the condition causes the tightness of skin around the eyes and eyelids. My brother’s eyes were irritated for about 5 years before his death. Just this summer I underwent plastic surgery to place skin grafts under my eyes to correct ectropion.

As I age with the condition, both good and bad things have happened. The massive calluses that were on my hands and the bottom of my feet have lessened and become more comfortable. But, about 15 years ago, the blood flow to my fingertips almost ceased to exist during cold weather. As a result the fingertips, under the fingernails become very irritated and crack, besides becoming greatly thickened. I do not know if this condition is associated with LI.

I probably am the biggest individual user of mineral oil in history, since I have used it on my skin since birth. About 15 years ago, my dermatologist introduced me to Ammonium Lactate and that has been wonderful. Now my self-care involves daily showers with a rough glove scrubbing.

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# Annual Report Fiscal Year 2003

## Executive Director's Report

**Dear Members and Friends of the Foundation,**

Thank you to all our members and friends who supported the Foundation through our winter holiday campaign. As you may know, our Foundation reaches out to the ichthyosis community for financial support three times per year, spring, summer and winter. This spring, the Foundation will conduct its annual membership renewal campaign. You should be receiving your annual membership renewal envelope in your mailbox within the next month. Please be sure to support the Foundation with your annual membership in order to remain a member in good standing and continue to receive our newsletter.

I extend another thank you to all those who took the time to complete the Foundation's on-line survey. The response to this survey was tremendous. Your comments and feedback will provide our board and staff with the necessary information to shape our future programs and services for you, our members.

In February, I attended the annual American Academy of Dermatology (AAD) Convention in Washington, DC. As a member of the Coalition of Skin Diseases, our Foundation exhibited in the convention hall and provided literature to the thousands of dermatologists in attendance. During the convention, I also visited several pharmaceutical companies to learn about new products that may be helpful for ichthyosis patients.

The day before the AAD convention, I had the privilege of participating in the First Annual Skin Disease Day on Capitol Hill, sponsored by the American Academy Association of Dermatology Association. Partnered with an NIH-sponsored scientist and a practicing dermatologist, each of us had a different perspective on how and why funding to the NIH, particularly NIAMS, is critical to skin diseases. Our congressional visits were well received and beneficial to everyone in the skin disease community.

Every year at the annual AAD convention, our Medical Advisory Board comes together for a meeting to discuss the Foundation's activities and ways for our physicians to collaborate on ichthyosis-related issues. This year was especially exciting for members of this group. The Medical Advisory Board was expanded to include leading scientists and researchers in the field of dermatology, particularly disorders of cornification (i.e. ichthyosis). The Medical Advisory Board changed its name to the Medical & Scientific Advisory Board and welcomed Sherri Bale, PhD, Theodora Mauro, MD, Dennis Roop, PhD, and William Rizzo, MD to their distinguished group. Future plans for generating more ichthyosis-related research were discussed. The level of enthusiasm from this group of doctors was incredible to witness. Plans for the future are very exciting and hopeful for those affected with ichthyosis.


*Drs. Sherri Bale, John Digiovanna, Phil Fleckman, and Alan Moshell.*

In March, I traveled once again to Washington DC to participate in the annual NIAMS Day. This day is organized for supporters of arthritis, musculoskeletal and skin diseases to meet with their congressmen and encourage their support of increased funding to the NIAMS branch of the NIH. Once again, it was a very successful trip.


*Jean Pickford with Congressman Joe Hoeffel (D, PA) outside the Capitol.*

Each year, The Foundation supports research into ichthyosis by providing grant money to the Dermatology Foundation. This year the Foundation provided $40,000 for two ichthyosis-related research grants. The grant recipients were announced in February. Dr. Anna Lee Bruckner from the University of California, San Francisco received a grant for the Utility of Ultrastructure in the Diagnosis of Ichthyosis. Dr. Jiang Chen from Baylor College of Medicine received a grant for Testing Gene Therapy Approaches for Epidermolytic Hyperkeratosis. Congratulations to both of these outstanding researchers. I will keep you informed of their progress.

The Foundation received a $5000 grant from Sovereign Bank to reprint our two educational booklets, "Release the Butterfly: A Guide for Parents and Caregivers of Children with Ichthyosis," and "A Guide for Teachers." Our staff and volunteers are busy making edits and updates to the original booklets, which were printed in 1994. Our goal is to have these booklets ready for our membership late this summer.

Working with our Fund Raising Committee Co-Chairpersons, the Foundation has been busy recruiting and working with volunteers on the following four areas:

- Soliciting major pharmaceutical/ corporate support
- Planning large special events
- Development of an official Planned Giving Program
- Grant Solicitation

I am pleased to report that we are off to a great start, but we are looking for more volunteers to help our co-chairs with all these special projects. Please contact me to discuss how you can get involved.

Sincerely yours,

Jean

Jean R. Pickford

# Annual Report *Fiscal Year* 2003

## President's Report

**Dear Members and Friends of the Foundation:**
These are exciting times for the Foundation and for each of you. The Foundation is the strongest it's ever been by almost every measure—financially, size of paid staff, involvement of volunteer expertise, and quality and extent of program and services. The Board of Directors and the staff are committed to further improving the Foundation's governance, including the development, updating, and constant focus on a comprehensive five-year strategic plan. Funding more ichthyosis-related research is at the top of Foundation's strategic priorities. Collectively, we are all making great strides in achieving ambitious funding goals related to research. As part of this effort, the Foundation's Medical and Scientific Advisory Board has recently been expanded to include even more of the nation's top experts who are involved in ichthyosis-related research. Consistent progress is being made every year regarding ichthyosis; we're moving closer and closer to a day when we can begin to seriously discuss not only highly effective treatments but a cure. I hope a record number of you will be able to join us in Kansas City in July for our 2004 National Family Conference. The program we have planned is the most comprehensive agenda that has ever been presented at such a conference.

There is much to celebrate in our recent accomplishments. I am especially grateful for the outstanding commitment and dedication of the Foundation's staff and for all of our stalwart members and friends. I am continually amazed by the passion and tenacity demonstrated by the members of our Medical and Scientific Advisory Board in their professional endeavors related to ichthyosis. I wish you could all see first-hand how well and how hard the Foundation's Board of Directors works to advance the Foundation. Although there is still much important work to be done, growth to achieve, people to help, I hope you all share my sense of achievement in how far we've come.

Sincerely,

Laura J. Phillips
President, Board of Directors

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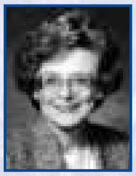

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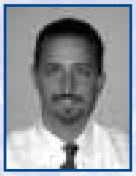

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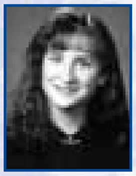

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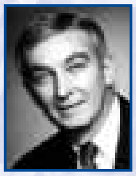

**Peyton Weary, MD**
Father of Children, EHK
Former President of American
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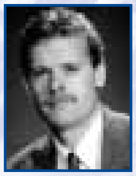
Professor of Dermatology,
University of Virginia
Charlottesville, Virginia

**Mary Williams, MD**
*Founding Member*
Adjunct Professor of Dermatology &
Pediatrics
Chief of Pediatric Dermatology
Service, University of California
San Francisco, California



**Peter Woodford**
Affected, Ichthyosis Vulgaris
Departmental Manager 15
State of Michigan, Department of
Management & Budget
Northville, Michigan

# Annual Report *Fiscal Year* 2003

## Chief Financial Officer's Report

Dear Members and Friends:

The Foundation continued making great strides in fiscal year 2003. Spending on program services increased almost 43%, while spending on administrative and fundraising decreased slightly. Contributions were up 16% from the previous year, primarily as a result of the Foundation's increased efforts to raise funds for research. You, our members, have been telling us that research is your top priority. As a result, spending on research also increased significantly. The Foundation awarded $80,000 in research grants in fiscal year 2003.

Several improvements to office operations were made during fiscal year 2003, including a major database overhaul, the development of ichthyosis fact sheets, a redesign of the FOCUS newsletter, and major changes to the Foundation's website. In fact, the Foundation's website won a Gold Triangle Award from the American Academy of Dermatologists. We are fortunate to have such a dedicated and hard-working office staff making the most of your donations and membership contributions.

The Foundation also continues to build on its relationships with pharmaceutical companies. In 2003, their financial contributions provided support to the Ichthyosis Support Network, helped to create a comprehensive physician and patient brochure about ichthyosis that was mailed to every practicing, non-cosmetic dermatologist in the U.S., and provided our members with several product samples. Several of these companies also made donations towards our 2004 National Family Conference. These contributions help make the conference more affordable for our members. I hope to see you in Kansas City this summer!

Our goal is to raise and spend $100,000 for ichthyosis-related research each of the next five years. We need your help to meet this goal! For those of you who are interested in raising money for research activities, the Foundation's staff has created an easy-to-follow manual to assist you in organizing a grassroots fundraiser. It contains great information about how to contact your local media and the essential steps needed to create a successful event, plus much more. To request a copy of the fundraising guide, visit our website at www.scalyskin.org.

I appreciate the opportunity to serve you and feel fortunate that our Foundation has the backing of such a diverse group of committed members and partners. Your ongoing support will allow us to continue these programs and make further advancements possible.

Sincerely,

Elizabeth A. Gray
CFO, Board of Directors

* * *

* *This Statement of Financial Position is excerpted from the Foundation's audited financial statements as of September 30, 2003. A complete copy of the audited financial statements and the independent auditors' 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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Eugene Van Scott, MD

## $10,000 +

Dermik Labs
Neostrata
Peyton & Janet Weary

# Conference Chatter

## 2004 National Family Conference: *A Fountain of Knowledge*

### July 2 – 4, 2004

### The Westin Crown Center, Kansas City, Missouri

**Registration** – See pages 13 and 14 for the official registration form. The registration form can also be completed online at www.scalyskin.org.

**Clinical Screening** – Experienced, knowledgeable dermatologists will examine conference attendees with ichthyosis. If you are interested in a consultation with one of the doctors, be sure to complete Section III of the registration form so that an appointment can be made for you. There is no charge for the clinical screening.

**Conference Program** – The conference program will feature four tracks of programs to accommodate each age group. Age-appropriate material will be included in each track. There will be a strict policy that no exceptions will be made to any age group.

Track 1 (Adults, ages 18+)
- What's new in research?
- Breakout discussions on specific disease types and treatments.
- Breakout discussions for women, men, mothers, and fathers.
- Sibling talk (for parents) deals with concerns often experienced by brothers and sisters, such as peer issues, resentment, and concerns about the future.
- Specialist panel will talk about eyes, ears, and nutritional issues.
- Breakout discussions to share information and helpful hints.
- Breakout session addressing depression in children.
- The ABCs of advocacy.
- School, workplace, and insurance issues.

Track 2 (Teenagers, ages 13-18) & Track 3 (Pre-teens, ages 9-12).
- Self-defense techniques.
- Making healthy choices in nutrition (teens only).
- Stress relief techniques.
- Affected teens and pre-teens talk (moderated by a clinical psychologist).
- *Sibshop* for unaffected siblings (moderated by a specialist in sibling issues).
- Career panel discussion, preparing for the future (teens only).
- Creative expression using art (pre-teens only).
- Exercise, stretching, and health.
- Teen and Pre-teen Jeopardy!

Track 4 (Children, ages 1-8)
- Program specifically designed for young children. Games, toys, art supplies, books, etc., will encourage children to interact. This track will be supervised by the licensed, trained, and caring professionals of A1 Tiny Tots, a professional childcare company.

**Discounted Travel to Kansas City, Missouri -**

US Airways – Receive 5% off lowest applicable published fares or 10% off with 60 days advance reservations/ticketing required from all points on US Airways' route system. Certain dates, rules, and restrictions apply. To obtain this discount, call US Airways' Group and Meeting Reservation Office toll-free at 1-877-874-7687, 8:00 am – 9:30 pm EST. Refer to Gold File Number 64162913.

Delta Airlines – Points of origin include US, Bermuda, Caribbean, Mexico, and Canada. Receive 5% off US Domestic published fare or 10% off published YO6/YR06 fare. Certain dates, rules, and restrictions apply. To obtain this discount, call Delta Airlines at 1-800-241-6760 and refer to Meeting Identifier Code DMN200484A.

National Patient Travel Center - You may be eligible for free transportation using the Angel Flight America Program. If you live within 1000 miles of Kansas City, Missouri, Angel Flight will fly a family in a 4-seater or 6-seater plane to and from the conference at no cost. Contact the National Patient Travel Center at 1-888-675-1405 and refer to the 2004 Foundation for Ichthyosis Family Conference Special Lift Program.

**Accommodations** – Attendees must make their own room reservations at the Westin Crown Center. Westin's discounted room rate is $89.00 per room, per night, flat occupancy + applicable taxes. Call 1-888-627-8538 to reserve your room at the Westin Crown Center. The Reservations Center is open 24 hours a day. Be sure to name the "Foundation for Ichthyosis" to receive the discounted rate in the room block. If you are interested in extending your stay in Kansas City, this rate is available from June 28 through July 7, 2004. However, you must make your reservation by June 4, 2004 to guarantee this discounted rate. After June 4, 2004, the rooms will be available on a first-come, first-served basis.

* * * * * * * * * * * * *

THE FOUNDATION FOR ICHTHYOSIS & RELATED SKIN TYPES

A FOUNTAIN OF KNOWLEDGE

FAMILY CONFERENCE • KANSAS CITY • 2004

**Westin Crown Center Hotel**
**Kansas City, Missouri**
**Friday, July 2 – Sunday, July 4, 2004**

## Section I – Contact Information

Name: ____________________

Address: ____________________

City: ____________________ State: __________ Zip: __________

Phone: ____________________ Email: ____________________

## Section II – Registrant's Information

This section must be completed with registrant's complete name, age, and appropriate track for each attendee. Please print clearly; nametags will be provided.

| Name | Age | Type of Ichthyosis | Adult<br>18+ | Teen<br>14-17 | PreTeen<br>10-13 | Child<br>1-9 |
|---|---|---|---|---|---|---|
| | | | ☐ | ☐ | ☐ | ☐ |
| | | | ☐ | ☐ | ☐ | ☐ |
| | | | ☐ | ☐ | ☐ | ☐ |
| | | | ☐ | ☐ | ☐ | ☐ |
| | | | ☐ | ☐ | ☐ | ☐ |
| | | | ☐ | ☐ | ☐ | ☐ |

## Section III – Clinical Screening Appointments

This is an opportunity to be seen by knowledgeable dermatologists to answer any questions or concerns that you may have. These appointments will take place throughout the day on Saturday, July 3. Your appointment will be assigned and posted at the registration table.

| Name of person for appointment | Age | Male/ Female | Type of Ichthyosis | Confirmed by a dermatologist? |
|---|---|---|---|---|
| | | | | Yes No |
| | | | | Yes No |

## Section IV – Other

| | Yes | No |
|---|---|---|
| • Do we have your permission to take photos during the conference? | ☐ | ☐ |
| • Do we have your permission to place photos of you and your family on our website? | ☐ | ☐ |
| • Do we have your permission to include your contact information on a conference roster to be distributed to all registrants? | ☐ | ☐ |
| • Have you ever attended a conference before? | ☐ | ☐ |
| • Can you volunteer some time at the conference to help? | ☐ | ☐ |

## Section V – Payment Information

The Foundation has received several sponsorships, which allows us to offer the discounted registration fees of $95 per adult and $50 per child. The actual cost per person at the conference is $150.00. Conference fees help offset the cost of food, beverages, room rental, supplies, etc.

Number of adults attending? (18 or over) __________ x $95.00 = $____________

Number of children attending? (17 or under) __________ x $50.00 = $____________

Additional Donation (optional) $ ____________

**Grand Total** $____________

☐ My check is enclosed, made payable to FIRST.

☐ Please charge my credit card: MasterCard, American Express, Visa (circle one)

Credit Card Number ______________________________________________

Expiration__________________ Authorized Signature________________________________

## Miscellaneous Information

- ✓ The conference will begin on Friday, July 2, at 2:00 pm CST and end on Sunday, July 4, at 12:00 pm CST.
- ✓ Breakfast, lunch, and dinner will be provided on Saturday, July 3, and breakfast will be provided on Sunday, July 4.
- ✓ On Friday evening, July 2, there will be free time to visit Kansas City. Transportation will be available from the hotel to deliver guests to the famed Country Club Plaza, home to a wide variety of shops, boutiques, restaurants, and theaters.
- ✓ Conference attendees are responsible for making their own room reservations at the Westin Crown Center. Call toll-free 1-888-627-8538, 24 hours a day.
- ✓ For those who are driving, there is a parking fee of $12.50 per night, with a $10.00 weekend max.
- ✓ For driving directions to the hotel, use www.mapquest.com or http://maps.yahoo.com/. The hotel address is 1 Pershing Road, Kansas City, MO 64108.
- ✓ KCI Shuttle provides transportation from the airport to the hotel. The cost for a one-way transport is $14.00 or $23.00 round trip. Simply report to the nearest KCI Shuttle ticket counter adjacent to most airline baggage claim areas.
- ✓ Mail this form to FIRST, 650 N. Cannon Ave., Suite 17, Lansdale, PA 19446. Registration closes June 11, 2004.

# Seniors Speaking

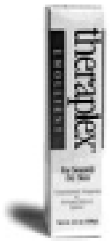
*(continued from page 6)*

I then spray myself with the Ammonium Lactate and use mineral oil on the most sensitive parts. The Ammonium Lactate prevents the large scales from accumulating and the skin sheds in smaller particles.

In my career, I served in a very public position that was always in the fishbowl of public interest. I served 4 cities in 4 different states. I'll bet that 99% of the people in those cities would be greatly surprised if they were told of my LI condition. Only my closest friends and family know. Sometimes at the YMCA someone would comment on the bad sunburn I had, but I'd just smile and say "sure."

Young people out there need to understand that self-pity just won't help. Get over it being an overwhelming factor in your life; don't let it define you. I went through all the psychological and social outcast treatment from peers and strangers when I was growing up, but you just have to ignore it. Now, when I attend school reunions and see the girls who wouldn't touch me, and the guys who used to tease and taunt me, I realize that I have had a comparatively very successful and happy life. It's been a great life with a wonderful family of five very successful children, (none with LI) and a satisfying career. Having LI is a minor inconvenience at the most.

I would love to hear from other older persons with LI. What other changes in my life will occur because of LI?



Thanks again for a great article and, more importantly, thanks for serving as our conduit to let us know that we are not alone in having this condition.

Very truly yours,

**William D. Frueh**
**Oshkosh, WI**

Dear Senior Friends,

I too want this column titled "Seniors Speaking" continued in the Ichthyosis Focus. Maureen and the staff are so generous in allowing our voices to be heard. If you, in reading this most informative journal, have this same goal **please** submit articles or thoughts that you want other seniors to view. Let us be the AARP of Skin Seniors. If we want items of interest for our age group, we have to be the contributor. And we can do it; we just need a little prodding!

I read articles about people worried about wrinkles, and face lifts. We have had this since the beginning of our time. Our skin is not our choice to deal with; it is the cards we have been dealt. Our facelifts are here every day, as we have to put on lubrication to make our skin comfortable for the rest of the day. Wrinkles and scales are with us everywhere we roam, and we often leave a trail of dry skin behind. The "normal" person doesn't have to think about a dark carpet showing our scales, or the upholstery on our car being dark or the bathroom rugs not being dark, especially in homes we visit as we leave a trail taking our clothes off and on. How about the color of clothes we choose; I personally am never at the liberty of wearing the beautiful black or dark blues I love. Oh yes, suggestions of the right shampoos do the trick on a perfect day. Most other days, I am self conscious of this embarrassment.

The word, self-conscious, couldn't we write a book on this subject? Since being very little, I knew I was different, and not in a pleasant way. My parents were always asked about me, and later I was asked. So many embarrassing questions, you sometime wonder how people can say the words. How have we handled this difficulty for 50 plus years? What can we share with younger F.I.R.S.T members and their families?

The years are going by fast. How about when we are in assisted living facilities? Will caregivers ever know how important it is for our skin to be lubricated? It is not just **dry skin**, it is every nerve ending needing to be lubricated. The great feeling of relief after this happens, only we can understand. How do we share this information with care professionals?

So I have given you two topics to respond to, but don't stop there. You share your views! This is all we have left at our age, support systems for each other. It is never to late to communicate. So look into yourself and share, and ask questions.

Happy Scales to you,
Beverly Browne
Italnhugger@comcast.net
El Dorado Hills, CA

# News&Notes

## Ichthyosis Research Study

Dr. Anna Bruckner, Dr. Peter Elias, and Dr. Mary Williams from the Department of Dermatology at the University of California, San Francisco, are conducting a study to identify certain changes that are seen when the skin from individuals affected by an inherited ichthyosis is examined using an electron microscope. These changes will be evaluated to see if they can be used as a diagnostic test for ichthyosis.

Participants in the study will have basic information regarding their disease collected and a skin biopsy will be taken. This biopsy will be sent to the San Francisco V.A. Medical Center where it will be analyzed using an electron microscope.

The study focuses on patients with Harlequin Ichthyosis, Lamellar Ichthyosis, Congenital Ichthyosiform Erythroderma (CIE), Netherton Syndrome, Vohwinkel Syndrome, and Epidermolytic Hyperkeratosis (EHK), as well as several other ichthyoses in which the underlying abnormalities are poorly understood.

*If you are interested in participating, or for more information, please contact:*

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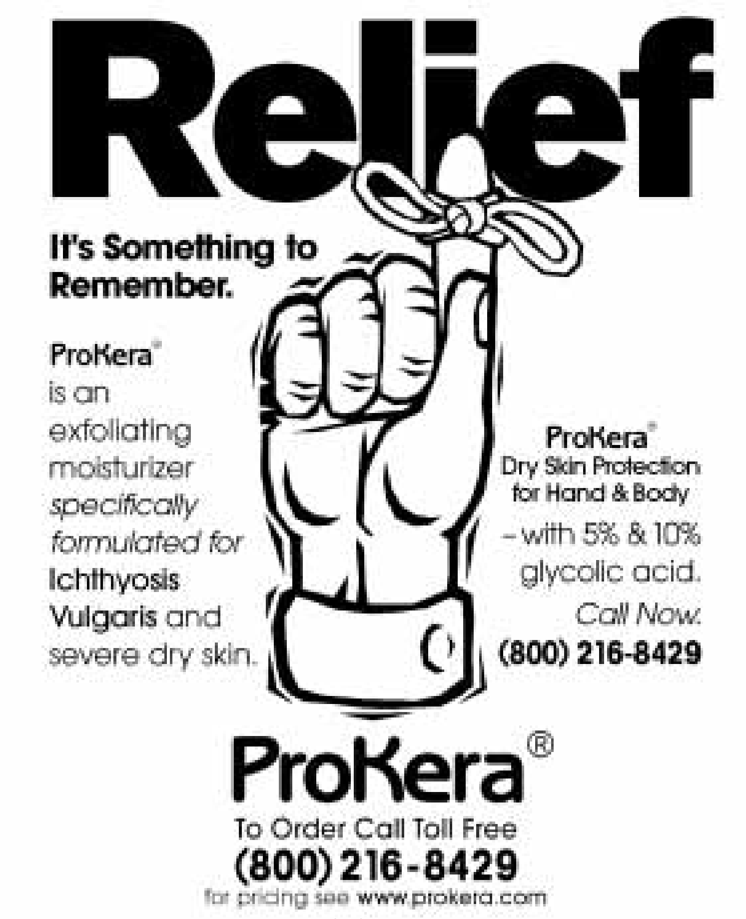
Email: abru5473@itsa.ucsf.edu
Phone: 415-353-7989
Fax: 415-353-7850

Mary L. Williams, MD
Email: williamsm@derm.ucsf.edu
Phone: 415-221-4810

Peter Elias, MD
Email: elaispm@itsa.ucsf.edu
Phone: 415-221-4810, Ext. 2092



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