



The National Ichthyosis Foundation

ICHTHYOSIS FOCUS

Send all correspondence to:
Barbara M. Landwehr
The National Ichthyosis Foundation
151 Toyon Drive
Vallejo, California 94589
Tel. (707) 644-5205

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FROM A MEDICAL POINT OF VIEW

The Ichthyoses: Recent Research Update

Peter M. Elias, M.D.
San Francisco, CA 94121

In previous numbers of our newsletter we have read a lot about the inheritance, classification, and treatment of ichthyosis. These articles helped to clarify what both you as patients and we as doctors already knew, namely that the ichthyoses are a very heterogeneous group of diseases, no more related to one another than are ulcers and constipation.

Now, there is also a growing body of evidence that the ichthyoses are distinctive pathologically, functionally, and biochemically. There is much active research in all of these arenas, fueled by one strong suspicion that understanding the ichthyoses, which are characterized by the worst scaling, will provide valuable clues into the mechanisms of normal skin shedding. Moreover, knowledge of the ichthyoses may provide clues into the mechanism of scaling in more common diseases like psoriasis and eczema.

Lamellar ichthyosis, which afflicts a goodly number of our members, is similar to psoriasis in that it is accompanied by increased turnover of skin cells. That is, as shown by Drs. Phillip Frost and Gerald Weinstein, skin cells are dividing at at least twice the normal rate. Moreover, both lamellar ichthyosis and psoriasis are characterized by red skin (erythroderm) and increased water loss from the skin. Although to some extent increased cell turnover may explain the scaling in lamellar ichthyosis, it is probably not the whole story. Increased stickiness of stratum corneum cells (the outermost layers in the skin) also contributes to scaling. Recent evidence from the laboratory of Drs. Mary Williams

and Peter Elias, supports that abnormal lipids (fat substances) may accumulate in the stratum corneum thereby leading to increased stickiness. However, since lamellar ichthyosis is an autosomal recessive disorder, it is likely that a single cause will be found for all of the abnormalities in lamellar ichthyosis, namely red and "leaky" skin as well as increased stickiness.

X-linked ichthyosis (RXLI), is another recessive form of ichthyosis, which is more common than lamellar ichthyosis, but less severe. Therefore, we don't have many members with RXLI. Yet, RXLI is also providing new insights into the causes of scaling. Dr. Larry Shapiro first found that patients with RXLI lack steroid sulfatase, an enzyme of steroid hormone and cholesterol metabolism. Later, Drs. Howard Baden and Ervin H. Epstein, Jr., found that this enzyme was missing in the scale and blood cells of RXLI patients, and Drs. Mary Williams and Peter Elias found that lack of the enzyme leads to accumulation of cholesterol sulfate in scale. This is yet another clue that fat substances may be important for the skin stickiness in the ichthyoses.

Although these new findings in lamellar ichthyosis and recessive x-linked ichthyosis are very exciting, we still have no idea how altered lipids might lead to increased stickiness.

Epidermolytic hyperkeratosis (EH), or **bullous congenital ichthyosiform erythroderma**, afflicts a goodly proportion of our patients. Since EH is an autosomal dominant

(familial) disorder, it is likely that it will be more difficult to find a single cause (missing enzyme) for this disease. EH is also characterized by increased turnover of skin cells and by abnormally increased skin water loss. The microscopic pathology is very distinctive in EH, and this has permitted pre-natal diagnosis of EH by skin biopsy through a fetoscope. This exciting development may presage the use of pre-natal diagnosis for several other skin diseases, as well.

By electron microscopy, Dr. Anton-Lamprecht in Heidelberg has identified abnormal-appearing protein fibers inside EH skin cells. Therefore, in contrast to lamellar ichthyoses and recessive x-linked ichthyosis, EH may prove to be a protein, rather than a lipid, abnormality.

Ironically, we know least of all about the most common form of ichthyosis, **ichthyosis vulgaris**. This autosomal dominant (familial) disorder is very common, but usually not severe; and therefore, patients often ignore their disease. Often they think they simply have bad "dry skin", and indeed, it is often difficult to distinguish severe dry skin from ichthyosis vulgaris. Both the turnover of skin cells and skin water loss are normal in ichthyosis vulgaris. Dr. Anton-Lamprecht and others feel that ichthyosis vulgaris also is a protein abnormality. They have found a striking absence of protein globules, called keratohyaline granules in this disorder, in contrast to all other forms of ichthyosis.

In future issues of the newsletter we will try to keep our members alerted to new discoveries in the ichthyoses.

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CHAPTER NEWS

MICHIGAN CHAPTER

Nothing could be more shocking than to find your newborn child has been afflicted with a condition you never heard of. There was no one to talk to. NOBODY knew what our daughter had. It wasn't until weeks after Quinn was born that we finally found someone able to diagnose it.



At this time, we are not going to rehash what happened or ask ourselves "Why", etc., etc. It has happened and that's that.

We decided to become as active as humanly possible in the Ichthyosis Foundation so that maybe someday we, or someone in our chapter, would

be able to offer comfort and understanding to a family that has experienced the same as us. We believe nothing could be more important than to have a successful Foundation. We have just begun and we can only imagine the amount of work necessary to do it RIGHT. The fellowship and availability to someone in need is priceless. That alone, though, we don't believe will keep a foundation going. We need "Go-Getters" and "Hard-Chargers" to make this happen.

Money is very important. We, personally, cannot wait to start generating funds for the foundation. Barbara Landwehr informs us every time we call of the great need for money. Everything costs! We have out-of-pocket expenses already and haven't even started.

Our first telephone conversation with Barbara Landwehr in February of 1981 was very enlightening. We are especially proud to have her as President of the national foundation. She is what we referred to earlier as a "go-getter" and a "hard-charger". We only hope that we can be as successful and energetic on the regional level.

We are looking forward to the responsibility of developing and operating a chapter, and are especially proud to be members of the National Ichthyosis Foundation.

--Quinn, Carol & Dick DeLoughary

OKLAHOMA CHAPTER

The following story was submitted by Betty McMasters, president of the Oklahoma Chapter and member of our National Board of Directors. Many of us, as parents, have had these same feelings and anxieties. Congratulations David & Betty on the birth of your new daughter.

Ichthyosis Families,

The 9 months wait was very hard to take at times, but it was worth it. We had a baby girl, 7 lb. 14 oz. Pamela Jean McMasters. Not even a trace of dry skin.

Now to tell you my deep feelings of how I felt through these 9 months.

I didn't want another child because of the risk of facing another child with Ichthyosis. Most of all, if this baby did have Ichthyosis, he or she would have to go through life with all the rude people out in the world who can be so harmful. As Katy, our 2 year old who has Lamellar, gets older I have noticed that it's not really the children who are harmful but their PARENTS!

When Katy and I made our first trip to the University of Seattle in Washington, they ran tests on us informing me that the chances of having a second baby with mild Ichthyosis would be 50-50, with a 25% chance of severe Ichthyosis. Just having that thought go through our heads was bad enough. On the other hand, we thought that Katy needed a brother or sister to grow up with. Of course, we were in hopes of a baby boy, but our new girl will do just fine!

I could have had a test done ahead of time telling me if the baby had Ichthyosis or not. The procedure involves examining the fetus during the 16th - 18th week of pregnancy. This is done with an instrument called a fetoscope, but the procedure carries a 5-10% risk of miscarriage and only three places in the country will do this test! David & I didn't want to go that way.

I believe there is a reason for everything that happens in this world. If you have the faith in God, he has answers to our many questions. It's just a matter of time.

Hope everyone had a nice Thanksgiving Holiday. One of my dear family came to Oklahoma from Kansas for Thanksgiving. We had a nice visit on Saturday.

Happy Holidays to all of you and may God bless.

DO YOU REALLY WANT A WORKSHOP?

The cancellation of the December Workshop was a tremendous disappointment to a great number of you. There were a lot of questions you wanted answered and you've waited a long time for such an opportunity.

A workshop of such intensity takes a lot more than just one person to do all the work. A committee needs to be formed - a committee of people who are dedicated to the common cause of **understanding and dealing with ichthyosis**.

To volunteer means to attend at least two organizational meetings at your own expense ... and a lot of hard work!

If this Workshop is really important to you and you wish to be on the committee, **please** write to Barbara Landwehr in care of this newsletter no later than February 28, 1982.

A LIVING MEMORIAL

So often money is wasted on flowers that quickly die. Make your remembrance of your loved one a gift that will live on, giving hope to those afflicted with Ichthyosis. Such a gift will help the National Ichthyosis Foundation reach others with Ichthyosis, educate the public, and assist in finding treatments.

To make such a gift, just send us the name and address of the person you wish to honor and remember. We will acknowledge all gifts and notify the family of this special tribute. The amount of the donation will be confidential.

Give the gifts of love and hope! The memory of your loved one will live on in the lives of those with Ichthyosis as they live more comfortable lives. What could be more thoughtful or lasting?

The National Ichthyosis Foundation

A New Year Message From Your President

Hi Friends!

I really apologize for the lateness of this issue of Ichthyosis Focus, I've been dragging my feet trying to put off the inevitable. It saddens me to have to write this message, but it must be done.

We have made numerous pleas to you, as members, to support your Foundation and still a great many of you have not even sent in your \$10.00 subscription fee.

We have had wonderful support from many pharmaceutical companies, as you have seen. Without Hermal Labs you wouldn't have such a beautiful newsletter. Just in December, an agreement was made with Roche Laboratories to print the literature about Ichthyosis that all of you have been crying for. Do you realize the great expense these companies are going to because they believe in us and want to help? No, I really don't think you do! And, what's more, I am doubting whether or not you really care! It seems that as long as you don't have to give from the pocketbook, your support is great. What you have to realize is that the responsibility for the success or failure of this Foundation is **yours** - not the pharmaceutical companies.

We are now broke! As I am writing this, we don't even have the money to renew our bulk mailing permit.

Other directors of various organizations warned me that this sort of thing would happen, but I refused to believe them. I was so sure that you would not let your organization down. I really believed it was important to you. **Was I so wrong to believe in you?**

It's sad to say, but this could possibly be your last issue of the Ichthyosis Focus unless we get money very soon.

It was very expensive for us to go to the American Academy of Dermatology in December. That's where all the rest of our money went. Had we not gone, we wouldn't have connected with Roche Laboratories for your Ichthyosis literature.

Please organize your various chapters into **immediate** fundraising events (i.e. garage sales, raffles, auctions, etc.) Do it today. Tomorrow will be too late!! And in the meantime, cut out the form below and mail it with your pledge and/or contribution, **TODAY!**

Best Wishes for a Happy
and Prosperous New Year,

Barbara Landwehr
Barbara Landwehr
President

I DO CARE ABOUT THE NATIONAL ICHTHYOSIS FOUNDATION !!!

Name _____ Phone () _____

Address _____

Enclosed is my immediate contribution in the amount of

\$500.00 _____ \$250.00 _____ \$100.00 _____ \$75.00 _____ \$50.00 _____ other \$ _____

I also pledge \$ _____ per month for the next _____ months.

Amount

Number

Amount

Date

Signature

The National Ichthyosis Foundation is a charitable organization. All the money collected by it will be used for charitable purposes, such as education, counselling, and ichthyosis research. The organization's major expenses are mailing its newsletter and printing educational material. Articles of Incorporation as a non-profit organization have been approved by the California Secretary of State and its Tax Exempt Status has been approved by the California State Franchise Tax Board. All contributions to the organization are deemed tax-deductible under both California and Federal law.

National Headquarters
Nat'l Ichthyosis Found.
151 Toyon Drive
Vallejo, Calif. 94589
(707) 644 5205

Regional Representatives
San Francisco Area
Ms. Judy Droste
710 Laurel Ave.
Apt. C12
San Mateo, Calif. 94401
(415) 348 3309

Colorado/Utah/Wyoming
Mrs. Teri Thompson
349 Silver Springs Court
Colorado Springs,
Colorado 80919
(303) 598 8020

Connecticut Chapter
Mrs. Alese Kummer
69 Church St.,
Seymour, Conn. 06483
(203) 888 0037

Michigan Area Chapter
Dick & Carol DeLoughary
24685 West Highland Ct.
Farmington Hills, MI 48018
(313) 879 8022

Carolina Chapter
Mrs. Karen Stone
459 Bull Creek Rd.
Asheville, N.C. 28805
(704) 298 6499

New York Chapter
Claudia Kennington
12 Cresthill Court
Huntington Station
New York NY 11746
(516) 423 0277

Ohio Chapter
Gene & Kitty Arick
985 Palmer Rd.
Grandview, Ohio 43212
(614) 488 4285

Kansas/Oklahoma
Mrs. Betty McMasters
1838 So. Muskogee
Sapulpa, Okla. 74066
(918) 224 9099

Pennsylvania Chapter
Pat Giuliana
12536 Deer Run Rd.
Philadelphia, Pa. 19154
(215) 637 7220

Virginia/Maryland
Mrs. Donna Tormey
116 N Harrison Rd
Sterling, Va. 22170
(703) 430 0585

Ichthyosis Focus — Editor
Barbara M. Landwehr
151 Toyon Drive
Vallejo, Calif. 94589
(707) 644 5205

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The National Ichthyosis Foundation
151 Toyon Drive
Vallejo, California 94589
(707) 644-5205



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Outpatient Building
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Ann Arbor, Michigan 48109

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2300 Children's Plaza
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Building 10, Room 12N238
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Bethesda, MD 20205

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Chapel Hill, NC 27515

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Salt Lake City, UT 84112

Sidney Hurwitz, M.D.
2 Church St. South
New Haven, CT 06519

Gerald Weinstein, M.D.
Department of Dermatology
University of California
Irvine, CA 92717

Mary L. Williams, M.D.
V.A. Medical Center
Clement St
San Francisco, CA 94121

Howard P. Baden, M.D.
Department of Dermatology
Massachusetts General Hospital
32 Fruit Street
Boston, MA 02114

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N.I.H.
Bethesda, MD 20205

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Piscataway, NJ 08854

Eugene J. Van Scott, M.D.
Skin & Cancer Hospital
Department of Dermatology
Temple Univ. Health Sciences Center
3322 N. Broad St
Philadelphia, PA 19140

Peter Elias, M.D.
V.A. Medical Center
Clement St.
San Francisco, CA 94121

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