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

ICHTHYOSIS FOCUS

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SCIENTISTS LINK GENE TO EHK

Dr. Sherri Bale and Colleagues at NIAMS Link A Keratin Gene To The Ichthyosis Epidermolytic Hyperkeratosis

National Institues of Health/NIAMS
Bethesda Maryland (June 30, 1992):

Researchers at the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) have linked a hereditary skin disorder, epidermolytic hyperkeratosis (EHK), one of the ichthyoses, to a cluster of genes that code for keratins. Keratins are structural proteins found in the outer layer of skin–the epidermis. The work, performed by Sherri J. Bale, Ph.D., and colleagues at NIAMS and the National Cancer Institute, was published in the July 1, 1992 issue of *Nature Genetics*.

"This finding sheds more light on the structure and function of the skin and should ultimately lead to improved diagnosis and treatment of epidermolytic hyperkeratosis and other inherited skin diseases," says Dr. Lawrence E. Shulman, Director of NIAMS.

EHK causes thickening and scaling of the outermost layer of the epidermis–the stratum corneum–as well as blistering. Some 3,000 Americans suffer from this chronic disease, one of a family of scaling skin disorders known as the ichthyoses. "There are definitely relationships among all the ichthyoses," Dr. Bale points out. "Once we've figured out what the genetic defect is in one type, we'll have clues to be able to solve some of the others."

Microscopic examination of skin samples from EHK patients reveals a characteristic pattern that includes abnormal clumps of keratin filaments. Keratins, which are the most abundant proteins in the outermost layer of the skin, are divided into two families—type I and type II keratins. In skin, pairs of keratin proteins consisting of one each of a type I and type II keratin are the building blocks for long strands of keratin called intermediate filaments. These strands normally form part of the internal "skeleton" of the cell—the cytoskeleton—a web-like network of molecules that reinforces the cell's structural integrity. Defects in keratin proteins and in their assembly into intermediate filaments could thus lead to structural defects in cells that might account for their fragility in disorders such as EHK.

The results reported by Dr. Bale and her colleagues provide strong support to the hypothesis that defective keratins can contribute to the skin abnormalities found in EHK. The researchers found a close link between the disease and a region on chromosome 12 that is known to contain a cluster of genes that encode several type II keratin proteins. A particularly strong link was found with one of these genes, the keratin K1 gene.

Researchers on EHK Gene Discovery Team: Standing(l to r): Dr. Peter Steinert, Sandra Santucci R.N., Dr. John Compton; Seated: Dr. Christopher Amos and Dr. Sherri Bale. Not pictured: Kathleen Kearns, Dr. Bernhard Korge, Dr. John DiGiovanna, Dr. Donia Abangan, Dr. O. Wesley McBride.

The researchers studied 20 people with EHK from three generations of a single family. Samples of DNA (genetic material) from each of these patients were used for genetic linkage analysis. This technique involves looking at how often certain genes are passed on from parent to child along with the disease. "In this family the disease gene and the keratin K1 gene are always inherited together, which makes us think K1 *is* the disease gene," explains Dr. John G. Compton, NIAMS, the lead author on this work. Linkage analysis also enabled the scientists to *rule out* linkage of the EH gene with other genes that were candidates for causing the disease.

It is still possible that another type II keratin gene that lies very close to the K1 gene on chromosome 12 may actually be the gene that

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IN THIS ISSUE:

A Report From the Executive Director

Welcome to all of the new friends of F.I.R.S.T. As a result of last year's Public Awareness Campaign and Frances McHugh's letter to Ann Landers, F.I.R.S.T. has doubled its number of contributors. We have found people who never knew that anyone else had skin like theirs. To all of you newcomers, please sit back and enjoy your first edition of *Ichthyosis Focus*. We welcome comments and questions. Our initial Information Form does not tell us much about you. So, speaking for myself, for the Board of Directors, and for Support Network Volunteers, let me say that we want to hear your stories. We want to know who you are, what you do, and how your life is going. Please write us!

One of the many things our friends can do is provide the medical professionals with information about our disorder. Since most physicians have only a few patients with one of the many forms of ichthyosis, they are unable to notice problems that many or all ichthyosis patients have. For example, how many of you were premature babies? How many of you have an adverse reaction or are allergic to the sulphur drugs? How many of you female patients notice a flare-up of skin problems related to your menstrual cycle? How many parents notice your child spike a fever just before a skin flare-up?

These are things we need to know and share. If you have any peculiar symptoms or want to share your questions, please write us. Our combined knowledge is the wealth of information about our disorder that the professional health care workers need to hear.

Many people ask how they can become involved in F.I.R.S.T.'s activities. Our organization is only able to do its work because of the many volunteers who function in diverse and sometimes very small ways that make a tremendous difference for all of us. Call the office and join us in accomplishing our mission of education, patient support, public awareness, and research advocacy.

And **THANKS** to all of those volunteers on the Regional Support Network, the Board of Directors, the Medical Advisory Board, and friends at large who help us grow and continue our strong presence both here and abroad.

P.S. Our National Office is in need of an IBM-compatible laser printer. If you know of any company or individual who is replacing a similar piece of equipment and who would like to make an in-kind contribution, please contact us.

Susan Snyder flanked by Dr. Lawrence Schulman, Director of NIAMS, and his wife

Dr. Marlene Huffman, Director of FDA Orphan Products Development, with Susan Snyder

A Note From The New *Focus* Editor

It is with great pride and heady optimism that we offer this, the first issue of *Ichthyosis Focus* produced under my editorial hand. With this issue I have introduced many editorial changes. Some of the changes should be readily apparent; others are likely less so. Most important, however, is that most of what has made *Focus* useful to F.I.R.S.T. members in the past remains in tact: the personal tone and flavor—the sense of one hand outstretched to touch another. The human dimension shall remain.

Instead, the strategy for revising *Focus* was designed to expand on the publication's existing strengths. If the cornerstone of the newsletter's usefulness has always been its slant on caregiving and support, then I've striven to add to this, not take it away. And what I shall strive to add (this will be a long process, spanning many issues of *Focus*) is a far broader base than ever before. Additionally, I will strive for a cleaner, more readable design and layout of the newsletter, hoping to make *Focus* more inviting to its readers.

This broader base that I hope to introduce into the newsletter will stand on three legs:

1. Caregiving and support, which has always been a *Focus* strength.
2. Concrete medical information—not just about caregiving opportunities, but about the world of medical research and treatment issues that surround the ichthyosis community.
3. Coverage of commercial, economic and political issues that are critical to *any* medical support organization, including F.I.R.S.T.

You will see much of this applied in this issue. In addition to a full report on the status of the Orphan Drug Amendments presently before congress, there is a report on issues highlighted by our sister organization, N.O.R.D. (see "NORD Report," on page 9, and Susan Snyder's report on F.I.R.S.T.'s congressional testimony on page 7). At the same time, none of the old flavor of *Ichthyosis Focus* has been lost.

I am looking forward to hearing from you, our friends and readers, for your comments and criticisms, observations and opinions. I need your help to make *Focus* the best it can be for us. We are, after all, the point of the publication.

How Will the Kassebaum Amendments Change the Orphan Drug Act?

Senate Bill S.2060, "The Orphan Drug Amendments of 1991," sponsored by Senators Kassebaum and Metzenbaum, leaves the Orphan Drug Act of 1983 largely in tact. What it does, however, is close the blockbuster loophole. It does this by creating what is called a sales "trigger" of $200 million in cumulative sales of orphan drugs. A company's seven-year monopoly on orphan drug sales would remain in tact **until** total sales of the drug reaches this $200 million trigger. At that point the market would be opened up to unrestricted competition.

The 1991 Amendments provide an appeal procedure to protect a company who can show that development costs for an orphan drug exceed the the $200 million trigger. Under these circumstances the exclusivity rights would remain in force, protecting the rightful profits of companies who undertake development of particularly challenging (and expensive) drug research projects. However, the Senate Antitrust Subcommittee studied this issue and found that development costs for the six blockbuster orphan drugs presently on the market were significantly less than $200 million. In fact, "the range of development costs is from $10 million to $150 million, with most of the blockbuster orphan drugs coming in below $50 million." In short, drug developers will easily recoup development costs **and** make handsome profits, even with the Amendments in place.

Another provision of the Amendments would create an Office of Orphan Diseases and Conditions within the Department of Health and Human Services. This Office will "coordinate the activities within the federal government concerning the development of drugs. The Amendments provide for an eleven-member advisory committee comprised of representatives of organizations of persons with rare disorders and conditions (5 members), research scientists (3 members), and representatives of health-related companies (3 members)."

Why Are Some Companies Exploiting the Orphan Drug Act

The need for amendments to the ODA would never have arisen had not certain companies, particularly biotechnology firms, begun exploiting the Act to shield their profits. At first glance, then, it appears that simple corporate greed is at the heart of the trend, but this is not altogether true. In fact, a peculiarity of the U.S. patent laws is the true root cause of the trend toward seeking orphan drug status for the products of recombinant DNA technology.

Under present law, a company may not normally receive a patent for "substances found in nature." For mainstream pharmaceutical companies this is not a problem, because drugs are normally composed of synthetic chemical compounds, which they may patent. For biotechnology firms, however, who are using remarkable new recombinant DNA technology to produce unique biological substances for treating rare diseases, the U.S. patent laws do not hold, since DNA is clearly a substance found in nature. The upshot is that, because U.S. patent laws make these products frequently unpatentable, some biotechnology firms (and pharmaceutical manufacturers using this biotechnology) have begun using the Orphan Drug Act as a means to create "back-door" patents on their products.

But the Orphan Drug Act was not enacted by Congress, nor signed by the President, to solve patent law problems for the nation's biotechnology companies. The ODA was enacted to offer government incentives for the sole purpose of encouraging the development of drugs of little commercial value in order to help save the lives of people suffering from rare diseases. If these firms are having difficulties with patent laws, they should be working vigorously to amend these laws, not hiding behind the protection of the Orphan Drug Act. The great danger of this is that the Orphan Drug Act itself may become sullied, corrupted, and ultimately become ineffective. The ODA has proven to be a good law, and it would be an American tragedy if it were to be corrupted by corporate self-interests.

Why Is This Debate Important to the Members of F.I.R.S.T.

Although treatments exist for **symptoms** of ichthyosis, there is as yet no therapy for the genetic basis of the diseases known collectively as the ichthyoses. In fact, at this writing researchers have not yet located the gene or genes responsible for the ichthyoses (except for X-linked ichthyosis). But this is changing (see the cover story for a report on the location of the gene for the ichthyosis EHK). Perhaps five years in the future, or ten, or maybe twenty years or even more, gene therapies for some or all of the types of ichthyosis may very well be available.

Protecting the integrity of the Orphan Drug Act (and that's really the issue here, protecting the integrity of the letter and the spirit of the law) is important to members of F.I.R.S.T. for two reasons. First, when gene therapies for ichthyosis arrive on the scene, almost certainly they will result from breakthroughs offered by recombinant DNA technology, and these products will likely be manufactured by biotechnology firms. For that reason, even though no products presently affecting the ichthyosis community are involved in the present debate, the technology by which they eventually *will* be developed is at the very heart of it.

Second, because ichthyosis is an orphan disease, members of the ichthyosis community have an obligation to stand up with the orphan disease community at large on any issue which threatens the present or future welfare of any part of that community. Today it is Gaucher disease, pituitary dwarfism, AIDS, and end-stage renal disease. Tomorrow, according to the Senate Antitrust Subcommittee, it will be cystic fibrosis, transplant patients, and patients with certain cancers. Next week or next year or next decade, however, it very well may be us.

Will Senate Bill S.2060 Pass?

Last year a bill very similar to the Orphan Drug Amendments of 1991 passed Congress on a unanimous vote. Surprisingly, though, President Bush vetoed the bill and it died in the Oval Office.

The same thing might happen this year. FDA Commissioner Dr. David Kessler testified at a March 3 hearing before the Senate Labor and Human Resources Committee. As reported in *Health News Daily*, a Capitol Hill health-industry newsletter, Kessler told committee members "'The Administration strongly opposes S.2060,' and if it were referred to President Bush, Health and Human Services Secretary Louis Sullivan 'would recommend that it [S.2060] be vetoed.'"

The pharmaceutical and biotechnology industries are lobbying hard to defeat S.2060. The industry's position is that weakening existing exclusivity provisions would discourage

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F.I.R.S.T. PRESENTS TESTIMONY BEFORE HOUSE AND SENATE

by Susan Snyder

Almost every year F.I.R.S.T. joins the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR) in participating in a presentation of testimony before Congress. The CPA-SDR has been instrumental in increasing funding to the National Institutes of Health, specifically, the National Institute of Arthritis, Musculoskeletal and Skin Diseases (NIAMS), which has resulted in tremendous breakthroughs in skin-disease research, including the discovery (reported in this issue of *Focus*) of the gene responsible for the ichthyosis Epidermolytic Hyperkaratosis.

THE BUSBYS: Stephen & Lianne, Rochelle, Evan & Lawrence

In December of 1991, F.I.R.S.T.'s Board of Directors decided to initiate advocacy for a National Ichthyosis Patient Registry. We applied to Congressman William H. Natcher (KY), Chairman of the Subcommittee, to present testimony by F.I.R.S.T. Our request was met favorably, and we began planning our testimony. After discussions with Ellen Rowe, President of the Board, We contacted Stephen and Lianne Busby of Avoca, New York, who have three small children with lamellar ichthyosis, to ask if they would be willing to come to Washington with their children to testify. They were honored to be asked and excited to be volunteering for F.I.R.S.T.

Following telephone interviews with the Busbys, Nick Gattuccio wrote the testimony while we made appointments with the offices of six Congressmen to arrange a round of visits between them and the Busbys, accompanied by Doctors Len Milstone and Lowell Goldsmith, both of F.I.R.S.T.'s Medical Advisory Board, and myself.

The Busbys were great! Rochelle, age 5, was dressed in her Easter best, with a straw bonnet and purse. Evan, age 3, was decked in a "spiffy" black suit and printed red tie, with small fire trucks bulging from his trouser pockets, while Alan, 15 months, wore a sailor suit. The children were precious and their spirited, warm and friendly personalities had the congressional aides cooing and tickling. Needless to say, we all made lots of friends that day, and certainly left a mark on the hearts of all who met with us. Stephen and Lianne Busby did a tremendous job presenting the testimony with all three children on their laps at the witness table. Days afterward we were still getting phone calls from people who either had been in the hearing room or who'd received second-hand reports on the testimony. The Busbys returned to Washington in July to offer similar testimony before a Senate Committee

Many, many thanks to Dr. Milstone, Dr. Goldsmith, Nick Gattuccio, Mr. & Mrs. Larry Kline, Laurie Kline, and all five Busbys for their efforts on behalf of the more than one million ichthyosis sufferers across the country. F.I.R.S.T. will continue to be a strong voice in Washington to educate policy makers about our conditions and our personal lives.

Please take a few minutes to write your Congressmen or members of the Appropriations Committee, who make decisions about how tax dollars are spent. (You will find them listed in the last issue of *Focus*.) Tell them you want to see more money invested in research and improvements in our health care system. Let them know who you are. After all, they are in their positions to represent you and your interests. If not your opinions, they'll be listening to some else's.

F.I.R.S.T. ON THE ROAD

by Susan Snyder

Through a scholarship awarded by the Food & Drug Administration, F.I.R.S.T. was able to attend the annual meeting of the National Organization for Rare Disorders (NORD), of which F.I.R.S.T. is a member. The four-day conference offered a range of presentations on subjects such as the American Disabilities Act, Genes and the Human Genome, Gene Therapy, Activities at the FDA, Family Support Systems, Public Relations Tools, Fund Raising, Board Development, and Grant Writing, to name just a few. An awards banquet honored pharmaceutical companies who have contributed to the development of new and not very profitable drugs for rare disorders. Dr. David Kessler, Director of the FDA, opened the banquet with remarks about his own experiences as a physician in the delivery room where some very "special" children were born.

In June, F.I.R.S.T. featured an exhibit at the 18th World Congress of Dermatology meeting in New York, where nearly 6,000 dermatologists from around the world gathered to share information of interest to the world of dermatology. Perhaps the most interesting part of the meeting for me was listening to the many research presentations, case studies, and poster exhibits on ichthyosis. It is exciting to know that so many scientists in the U.S. and around the world are studying ichthyosis and other disorders of keratinization. Several members of F.I.R.S.T.'s distinguished Medical Advisory Board were among those scientists presenting their research findings to the assembled body.

Through the joint efforts of tri-lingual New York volunteers Carmen Delsol and Cathy Lopez, we were able to speak with physicians in four different languages. We made many fruitful contacts, answered innumerable questions, and distributed much of the literature we'd brought from Raleigh. Many thanks go to volunteers Carmen Delsol, Cathy Lopez, as well as their families for allowing them time away from home for this important contribution to our cause. Additional thanks go to in-kind contributors Deb Vilas and Jeff Krauss. Well done, team!

stopped looking at her as a patient, as someone afflicted with a dread disease, and we began looking at Caitie instead as our daughter—a beautiful girl with different skin. We became, at last, a family.

It's been one year, now, and I watch Caitie in her high chair surrounded by partyers, the center of attention, as usual. She frolicks, excited, screaming with delight. We blow her first birthday candle out for her, and then she goes for the cake herself—not with a hand, but with her face. I stand there and I watch her demolish her first birthday cake and I realize how far we have come as a family.

Caitie does have a relatively severe case of lamellar ichthyosis, but the ichthyosis does not consume our lives as it did in those difficult first months. In fact, days go by when I don't even think about it. Often, we lotion Caitie on auto pilot. It is just another part of our lives. Her skin looks very dry and her eyelids pull away from her eyes somewhat because of the tightness of her skin, but Caitie is a beautiful child with icy blue eyes, curly red hair and a fiery personality. She has a deep laugh and will wrestle you for a grape popcicle. She has an iron will, too, and we encourage it, because we know how desperately she will need her strength and her will throughout her life.

Some days are difficult, though. Just last week, Caitie and I were grocery shopping and I heard a young child comment to her mother how "horrible" Caitie looked. Tears came to my eyes instantly because I realized this child's comment was just a preview of the many thoughtless comments Caitie will hear in her lifetime. We know the road ahead is not going to be easy.

So Caitie is a year old, now, and Nick and I are one year stronger. All along we had thought it was us who cared for her, but now we've learned it's the other way around. Caitie gave *us* this strength. Our future is bright and optimistic, and this is so because her future is in her own hands, we've learned, not in ours or a thoughtless world's. And so I realize, now, that Caitie can go to her prom, that she can go to law school and that she can get married, because it is her world, not mine, and I'm humbled by this lesson she has taught me. But like all daughters, she will likely ignore her mother's dreams, exert her already blossoming strong will, and do exactly as she pleases. Nothing would make me happier.

N.O.R.D REPORT

The National Organization for Rare Disorders (NORD) is an umbrella organization representing the interests of groups like F.I.R.S.T. in the difficult arenas of political lobbying and health-care advocacy. F.I.R.S.T. is a member of NORD. "NORD REPORT" is the first of an ongoing digest of highlights from NORD's newsletter, NORD ON-LINE.

Bookstores offer special collection for children with special needs.

Barnes & Noble, Inc., the nation's largest bookseller, announced a new program for children with disabilites: The Children with Special Needs Collection, which will be offered in some 700 B. Dalton and Barnes & Noble book stores nationwide. Most selections in this collection had previously been difficult to find in bookstores, including special content books for children with disabilities, as well as for their families, friends, teachers, and health care professionals.

Participating B. Dalton and Barnes & Noble bookstores will prominently display the books in the top two or three shelves of the "Family & Child Care" sections of the stores. A brochure about the collection is available at all participating stores.

The FDA scrutinizes health-insurance industry practice of denying reimbursement for "off-label" uses of drugs.

You should be aware that many rare disorders are commonly treated with drugs that are approved by the FDA for other, far more common conditions. Because laws require drug manufacturers to **prove** that drugs are both safe and effective for diseases or conditions which the manufacturer claims the drug is suitable for treating, and because obtaining such proof requires enormously expensive testing and trials, manufactuers usually only seek FDA approval for just one or two very specific applications for any given drug. At the same time, however, physicians become aware over time of secondary, or "off-label" uses of some drugs, and these off-label applications frequently become very common weapons in physicians' arsenals, frequently in treating rare disorders.

This situation creates a gap between technically "approved" uses, and medically common "applied" uses of drugs. The health-insurance industry has stepped in to exploit this gap by sometimes withholding reimbursement for these off-label uses, regardless of how common their use may be within the medical community. The health insurers often deny reimbursement on the grounds that these drugs are "experimental." This is particularly true of expensive drugs.

The FDA is looking in to this problem, largely due to vocal protests on the part of the AIDS and cancer communities, whose patients are particularly vulnerable to this problem. FDA Deputy Commissioner for Policy, Michael R. Taylor has created a task force to study the problem.

Anyone having health insurance reimbursement problems because their disorder is not listed on the label of a drug that a physician deems medically appropriate for therapy should send a letter to the FDA's task force director. Because the letter must include documentation of the off-label efficacy of the drug (e.g., journal articles reporting the findings of clinical studies), the letter will probably require the assistance of the physician who prescribed the drug in question. The letters should be addressed to:

Michael R. Taylor
Deputy Commissioner for Policy
Food and Drug Administration
5600 Fishers Lane
Rockville, MD 20857

Senator David Pryor releases a report on excessive inflation for pharmaceutical products

In September, 1991, Senator David Pryor, Chariman of the Senate Special Committee on Aging, released a very revealing report entitled ***The Drug Manufacturing Industry: A Prescription for Profit***. Following are a couple of highlights:

- Through the 1980s, while the consumer price index rose 58%, pharmaceutical prices in the U.S. rose 152%, or nearly triple the inflation rate.
- Americans pay, on average, 62% more for a prescription than do Canadians for the same drug, and 54% more for the same item than do Europeans.

Stay tuned for a fuller report on Senator Pryor's report in an upcoming issue of *Ichthyosis Focus*.

A VERY SPECIAL THANK YOU

F.I.R.S.T. wishes to thank everyone who has contributed to the Foundation through June of 1992. Some have given in the form of personal time and energy. Others have supported us with notes of encouragement and with many helpful suggestions. And many have contributed financially to help keep our programs running. Thanks to all of you, word is spreading about ichthyosis..

Special appreciation goes to those who gave $100 or more. If we have inadvertantly overlooked you, please let us know. We want to thank each and every one of your for your continued support.

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Researchers Link Gene to EH

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is defective in EHK. Definitive proof for a role of the keratin K1 gene in EHK will require more experiments, such as looking for evidence of a specific mutation (defect) in the K1 gene of this family.

It is of particular interest that mutations in two other keratin genes have been linked recently to the hereditary blistering skin disease epidermolysis bullosa (EB) simplex. [See the last issue of *Focus* for an article on the discovery of the gene for the disease epidermolysis bullosa simplex, whose workings are remarkably similar to those reported on here.] Like EHK, EB simplex is a dominantly inherited skin disorder involving the epidermis and is probably caused by keratin defects. It is distinctly possible that, as in the case of EB simplex, in different families defects in different keratins may be found to contribute to EHK. Genetic studies of other families and individuals suffering from EHK are planned by Dr. Bale and her colleagues which may provide the answer.

Elucidating the role of various structural proteins of the skin in hereditary skin disorders is yielding great insights into both the normal and abnormal structure and function of the skin. Besides leading to better diagnosis, this knowledge may help researchers develop ways to treat these and other skin disorders.

We will follow these and other research developments in future issues of Focus.