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

ICHTHYOSIS FOCUS

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RECENT RESEARCH ON THE GENETICS OF ICHTHYOSIS: WHERE ARE WE NOW, AND WHERE WILL (OR SHOULD) WE GO?

Sherri J. Bale, Ph.D.

Dr. Bale received both her M.S. in Human Genetics and Genetic Counseling (1981) and her Ph.D. in Human Genetics (1984) from the University of Pittsburgh Graduate School of Public Health. She took her Fellowship training in Medical Genetics at the National Institutes of Health from 1984-1987, and was Board Certified in Medical Genetics in 1987 (American Board of Medical Genetics). She has been involved in research into the genetic basis of hereditary systems throughout her career. She is currently the Chief, Genetic Studies Section, Laboratory of Skin Biology, National Institute of Arthritis and Musculoskeletal and Skin Disease (NIAMS), NIH. Dr. Bale Holds a black belt in judo as well as regional referee certification in the sport. She is a member of the Board of Directors of Maryland Judo, Inc. She is married to Eric Spears, N.B.A., M.A.I., and has a 15 year old son, Nathan.

Research into the genetic basis of hereditary skin disorders has become hot, hot, hot! in the past five years. And the group of disorders coming under the umbrella of "ichthyosis" has definitely reaped the benefits of that attention and excitement. In this article, I will briefly review the recent advances in our understanding of the genetic cause of the ichthyoses. I will discuss how these findings can be used to benefit the specific families who participated in the research leading to the breakthroughs. I will address, also, the broader issue of "translational research" -- how research results can be accessed and utilized by the many families and patients who are affected with the disorders.

Recent enlightenment about the genetics of ichthyosis was actually launched by the finding that mutations in keratin genes caused a skin disease called Epidermolysis Bullosa Simplex (EBS). This disorder causes blistering with minimal trauma in affected persons, and is actually unrelated to the ichthyotic group of disorders. However, because of knowledge about the particular site within the epidermis (upper portion of the skin) that keratins do their job, other investigators were able to extrapolate the findings from EBS to Epidermolytic Hyperkeratosis (EHK), an autosomal dominant type of ichthyosis. The scientists hypothesized that different keratins, expressed in the portion of the skin affected in EHK, might be responsible for that disease. And in 1992, the first mutations in the genes for keratin 1 and 10 were identified in patients/families with EHK.

Once mutations in keratins 5 and 14 were found in EBS, and mutations in keratins 1 and 10 were found in EHK, things really got rolling! Mutations in keratin 9 were found in a type of ichthyosis that affects only the palms and soles (Palmar/Plantar Keratoderma, or PPK); mutations in keratin 2e were found to cause Ichthyosis Bullosa of Siemens; and mutations in keratins 6a, 16, and 17 were found in Pachyonychia Congenita (PC; considered either to be an ichthyotic disorder or an ectodermal dysplasia). This work really opened up the study of keratins, themselves, and then mutations in other keratins were found in other disorders that affect epithelial cells, but do not result in ichthyosis, including keratins 4 and 13 in white

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Dr. Bale will be discussing genetic research at F.I.R.S.T.'s National Conference, June 19th - 21st in Philadelphia. See pages 9 - 12 for complete program and registration information.

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The editor invites correspondence. We welcome your comments, observations and suggestions. Please send your letters to us c/o *Ichthyosis Focus* at the address listed above.

CORRESPONDENCE CORNER

Dear F.I.R.S.T.,

I'm writing to you on behalf of my son, Rhyan, who is nearly two and has CIE. The information and literature provided to us by F.I.R.S.T. have been so helpful and we wanted to say "thank you". We also wanted to pass some of our knowledge on to others.

Rhyan has been seen by some of the best in the field of dermatology and genetics, including Dr. DiGiovanna and Dr. Bale at National Institutes of Health. Along with his other two dermatologists, Dr. Peck and Dr. Cohen, we have received so many suggestions, some that worked and some that didn't. The lotion and cream that I absolutely swear by are Carmol 10 and Carmol 20 (OTC). These products contain urea, so test a patch of skin first to make sure it doesn't sting. I apply the Carmol 10 first and the Carmol 20 over it. On Rhyan's face, I mix Vaseline and Carmol 40 (prescription) together in my hands and apply. I use a mixture of salicylic acid and mineral oil (your dermatologist must write for this) on his scalp to remove his scales, then I scrub with a fingernail brush, and shampoo off with Baker's P&S. I alternate the mixture every other day with Baker's P&S liquid.

I hope that some of these suggestions may help you or your child. I have seen truly remarkable results on Rhyan. Make sure to speak to your dermatologist first, especially about the salicylic acid/mineral oil mixture because it can be harmful if absorbed by the skin. I hope to see all of you at the conference, bye!

Kirsten Brennan
Waldorf, MD

Dear Friend at F.I.R.S.T.,

Thank you for the information you have posted out to our group. I cannot tell you how excited we were to find that a group such as yours existed. The type of information you have provided is exactly what we were hoping for and we have already formulated ideas from the booklets and newsletters for disseminating to our members. We were also very impressed by the size, medical involvement and political standing that you seem to have. Our group is really only in its infancy and it is very supportive to know that we can access information and knowledge from a body such as yours.

Yours Sincerely,

Dianne Josephs on behalf of the
S. Australia Ichthyosis Group

Dear Friends at F.I.R.S.T.,

Before my Women's Club Christmas luncheon, a short meeting and discussion were held. The subject of charitable donations was brought up and we voted on three of them - one of them was my motion to donate $250.00 to F.I.R.S.T. Of course a lot of questions were asked about it as they had never heard of it. Some of my closest friends know our son, Randy, who has CIE. They spoke up and said to donate $500.00!! So here is the check!!

I would like to challenge other members of F.I.R.S.T. to speak up at their respective Woman's Club to donate also. Together We All Win!!

Sincerely,

Joe, Chris & Randy LaBarbera
Jacksonville, FL

SECOND ANNUAL BOARD RETREAT

by Donna Rice

I was so excited as I walked into the lobby of the Cherry Hill Hilton in New Jersey for the second annual Board of Director's retreat on a clear, crisp day in November. Excited to be away from home for a weekend, excited actually to SEE friends on the board and meet the new members, excited by all I knew we would accomplish, but mostly I was excited to imagine returning there with my family in June for the F.I.R.S.T. National Conference. It was so easy to imagine the lobby filled with members and friends of F.I.R.S.T., seeing familiar faces in the restaurant or hearing the laughter of our children in the childcare rooms and at the pool. Planning the 1998 Conference (a significant portion of the retreat agenda) was an important task made easier by being at the conference site.

The very ambitious retreat agenda included electing new members, Rita Tanis of Connecticut, Laura Phillips and Lewis Horowitz of New York, and Dr. Mary Williams of California, as well as thanking Mike Pinnisi, whose term ended in December, for his service to F.I.R.S.T. The new slate of Board officers approved for a two year term were:

Donna Rice, President
Michelle Petersen, Vice President
Elise Johnson, Secretary
Laura Phillips, Chief Financial Officer

The board was fortunate to be able to thank Mike Dunleavy in person for his leadership during the last year as Board president. His continued commitment to F.I.R.S.T. puts the Board in a unique position of having the most recent presidents, Mike and Deb Vilas as active board members. Francis McHugh, another previous president, attended the retreat as advisor for the second year in a row.

In addition to the conference, we addressed two areas, which we will continue to focus on in the coming year. The development and fundraising session resulted in the establishment of a research committee chaired by Dr. Leonard Milstone. Our membership and services discussion resulted in a commitment to increase the accessibility of F.I.R.S.T. programs, including increased circulation of *Ichthyosis Focus* and expansion of the Regional Support Network by hiring a part-time paid RSN coordinator.

Renewing the Board's commitment to F.I.R.S.T. is never officially on the retreat agenda, but is always the end result. Thanks to all Board members for their attendance, insight and commitment.

WHERE ARE WE NOW, AND WHERE WILL (OR SHOULD) WE GO?

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sponge nevus (a disorder of mucous membranes) and keratins 3 and 12 in Meesman's corneal dystrophy (an eye disease). Every one of these findings resulted from the study of many families in which the disorders were occurring, and could not have been accomplished otherwise.

The next important type of ichthyosis to succumb to the efforts of the geneticists was Lamellar Ichthyosis (LI). Studying families from both Egypt and the United States, researchers were able, in 1994, to identify the chromosome on which the LI gene resided. Very soon thereafter, the gene for an enzyme that is active in the epidermis, transglutaminase-1 (TGM1), was found to be mutated in persons with LI.

The genetic basis of at least one other disorder that includes ichthyosis as one of several major clinical problems (Sjogren-Larrson syndrome) has also been discovered in the past few years. Mutations in the gene for fatty aldehyde dehydrogenase (FALDH) were shown to cause Sjogren-Larrson syndrome in 1996.

So the next issue is: How can knowing the specific gene that is involved in EHK, LI, etc. benefit the families who participated in the research that led to those discoveries? In many of the participating families, the exact mutation in the keratin gene (for EHK or PPK, for example) was identified during the course of the research. Depending upon the agreement between the research subjects and the investigator (as outlined in the consent document signed by all participants and approved by the Institutional Review Board where the investigator carried out the research), the patients may be given that information. Knowledge of the specific mutation could then, potentially, be used for purposes of family planning and genetic counseling (risk of recurrence in future births, carrier detection for unaffected relatives of persons with recessive ichthyosis), and even pre-natal diagnosis (i.e. determination of a fetus' disease status prior to birth).

Although the specific disease-causing mutation was identified in many of the families who participated in genetic studies, there are many other families where this information is still not available. For example, researchers still find that they are unable to identify causative mutations in keratin 1 or 10 in 20%-30% of people with EHK.

If you or your family participated in genetic research, how can you find out what your results are? I would suggest that you call the investigator with whom you participated. If the results are available, AND if the research was conducted with the agreement that the information would be given back to the families, you *will* probably be able to get it.

What if you participated in genetic research, and the investigator says he/she has no results available? Why is that? There are two reasons. In LI, where the gene that has to be screened (TGM1) is quite large, it is a great deal of work. And since LI is autosomal recessive, a mutated gene must have come from each parent and therefore two mutations have to be identified. And worse, the mutations seem to occur all over the gene, so there is no high yield portion of the gene to screen. In EHK, there are two different genes to screen (keratin 1 and keratin 10), although clinical features can give a good clue as to which gene to work with first. At least in the keratin genes, the mutations tend to occur in particular parts of the gene, so one can design a scheme to investigate the most likely portions first and therefore have a higher likelihood of success in mutation identification.

Beside the fact that the work can be difficult, there is a more important reason why there may be no information available for a family that participated in genetic research. This is due to the nature of research, itself. That is, research is supposed to find *new information* that leads to greater knowledge of the function of biological systems. Once it is known that mutations in TGM1 cause LI, for example, the cataloging of a large number of mutations in TGM1 is no longer of such great interest to the scientific community. In fact, a researcher's funding to do his/her work is dependent on always learning *new* things. Researchers often have no resources (money for supplies or personnel) to do exhaustive searches to identify specific mutations in all the patients they have studied, once the basic discovery has been made.

In my opinion, this may be a major problem for some families who become involved in research, and it should also be something that makes the researcher uncomfortable. But what is the solution?

The answer to the above question brings in the third issue mentioned at the beginning of this article. "Translational research" refers to the act of bringing research findings from the research laboratory to the clinical setting. With respect specifically to the issue of mutation identification in genetic skin disease, translational research addresses the issue of providing genetic information to the many families with EHK, LI, PC and PPK that are no longer of "research interest", but who can benefit from the knowledge gained in the research setting.

There are two major restrictions to using the research laboratories to provide this information. The first is the lack of resources and the different purpose of research laboratories as discussed above. The second is that most research laboratories have not necessarily been certified as meeting the federal regulations that govern provision of results that will be used by physicians for the purposes of diagnosis or management.

The 1988 Clinical Laboratory Improvement Act (CLIA) is the relevant federal regulation that governs these facilities. Only CLIA-certified laboratories are supposed to perform such tests and provide the information to the clinician.

Are there CLIA-certified laboratories available to perform mutation detection in the keratin genes, TGM1, or FALDH? A search of HELIX, the genetic laboratory diagnostic database on the internet, turned up three laboratories that will test for X-linked ichthyosis. However, all testing for EHK or LI is being done only on a research basis. I could identify no service laboratories testing for keratin 1, 2e, 9, 10, 6a, 16, 17, or TGM1 mutations. Although there are many service laboratories that do genetic testing and mutation identification, apparently there is not a large enough patient population with ichthyosis for any laboratory to believe that it is cost-effective for them to set up the necessary assays for these genes.

How can this problem be solved? It is my personal opinion that this issue can only be brought to light by the patient community. You, the members of F.I.R.S.T., are the ichthyosis patient community. You will have to speak up if you believe the rare genetic disorders should not be ignored. You need to publicize the fact that the relevant research information is available and that a way must be found for it to become available in a clinical setting. Without the tremendous cooperation of families with these disorders, we would have none of this new knowledge about the genetic basis of the ichthyoses, or any of the fascinating information we have gained about skin structure and function from our work. It is clearly now time to give something more back to the community. But you, the patient community, will have to proclaim your needs!

This article presents the personal views of Dr. Sherri J. Bale. This viewpoint is not necessarily that of the National Institute of Arthritis and Musculoskeletal and Skin Diseases or of the National Institutes of Health.

F.I.R.S.T. Advocates for Skin Disease Research

In the previous article, Dr. Bale discussed recent advances in our understanding of the genetic causes of ichthyosis, and some of the hurdles involved in making the results of this knowledge more widely available. She urged us all to become more active in increasing awareness of ichthyosis and the research needs of our community.

On Wednesday, February 4th, Shelly Licursi, Regional Support Network coordinator for the north-Atlantic States, and her son Ryan, a twelve year old who has Epidermolytic Hyperkeratosis (EHK) traveled to Washington D.C. to testify on behalf of F.I.R.S.T. They appeared before the House Appropriations Subcommittee on Labor, Health and Human Services, Education and Related Agencies. Much of their testimony focused on the need for additional research that will translate this new scientific knowledge into practical applications for better testing and more effective treatment.

Each year, F.I.R.S.T. requests an appearance at a public hearing before this committee to discuss appropriations for skin disease research. We are delighted when we are invited to testify (as we have been these last two years). We do recognize that we are just one of many healthcare organizations, each with their own funding priorities, that are competing for the attention of this committee.

In March, therefore, we will also travel to the capital as part of a larger group, the Coalition of Patient Advocates for Skin Disease Research. We will meet with Dr. Steve Katz, the Director of the National Institute for Arthritis, Musculoskeletal and Skin Disease (NIAMS) which is part of the NIH. We will join with other skin disease associations to visit the congressional offices of Appropriations committee members, where we will focus on the importance of skin disease research and increased funding for NIAMS and NIH. Finally, we will visit the offices of our local senators and representatives.

The Foundation can be an effective advocate for our members (previous efforts played a key role in government funding for the Registry for Ichthyosis & Related Disorders) but we need your help. Our elected officials need to understand what it is like to struggle with ichthyosis on a daily basis. A personal appeal for better treatment and testing can have an enormous impact on reinforcing the advocacy efforts of the Foundation.

You can help ensure that our message is heard!

Please contact your elected officials to educate them about ichthyosis and the research needs of our community. F.I.R.S.T. can provide copies of our testimony, legislative contact tips sheets, and proper forms of address for letter writing. Contact the National Office for more information.

Focus On...
Pityriasis Rubra Pilaris

Mary L. Williams, M.D.

Dr. Mary Williams is currently an Adjunct Professor of Dermatology and Pediatrics at the University of California San Francisco. Her interest in the problems of ichthyosis dates back to her dermatology residency in the late 1970's, when she participated as an investigator in the first clinical trials of oral retinoids in disorders of cornification. During this study, many of her patients were able to meet someone with "their disease" for the first time. The benefit to children, in particular, through the opportunity to meet successful individuals with ichthyosis was particularly touching. In her waiting room, through the efforts of these parents and patients, F.I.R.S.T. was born. Dr. Williams continues to be closely involved with this family of disorders, both in her clinical practice, in her teaching and publications, and in her research laboratory.

If the ichthyoses may be considered "orphan" diseases, because of their rarity[1], pityriasis rubra pilaris (PRP) is an orphan of orphans. Like the ichthyoses, the disorder is one of scaling skin, and like most individual types of ichthyosis, PRP is quite rare. Yet, unlike most of the ichthyoses, PRP is not inherited or genetically based[2], and also unlike most ichthyoses, the disorder is neither lifelong nor congenital (present at birth). PRP appears to come in several forms and its classification is confusing even to the experts. The most common form begins in adult years, usually starting as focal red, scaly bumps and patches, often on the scalp and upper body. At this stage PRP may be difficult to diagnosis, as skin biopsy findings are not absolutely diagnostic, and a variety of more common disorders, particularly psoriasis, but also one of the eczemas, and even a cutaneous T-cell lymphoma, may be considered. But over time, the correct diagnosis usually declares itself, as PRP typically continues to progress over weeks and months until most of the body is involved in a generalized red scaly rash (exfoliative erythroderma), distinguished by skip areas or islands of normal, uninvolved skin, a marked, waxy thickening of palm and sole skin, and a roughened, "nutmeg grater" quality of skin over the hands, knees or elbows.

The vast majority (~80%) of the adult patients with the "classic" form of the disease will enter remission within 3 years; and this form of the disease is typically very responsive to retinoids (Tegison® or Accutane®). Rarely, adults may have an atypical form in which onset is more gradual, disease is poorly responsive to treatment and of longer duration. Children may also develop PRP. Preschool children in particular may develop a more localized form of the disease, often with patches predominantly on elbows and knees. Older children may have a more adult pattern of disease expression. A rarer, familial form appearing in childhood and resistant to treatment is also recognized, although some experts feel that this may be another inherited disorder of cornification (i.e., an ichthyosis variant). Finally, unusal cases of a severe PRP-like rash have been observed in a few patients with Acquired Immunodeficiency Disease.

The cause of PRP is unknown. Deficiency in vitamin A or a defect in vitamin A metabolism has been suspected, but is not proven. Because of the variety of clinical courses in different patients, it is likely that the causes may also be multiple. In children, an association with preceding viral infections has been observed.

In addition to the synthetic retinoids and vitamin A[3], many therapies used for recalcitrant psoriasis have been used to treat PRP[4]. In children with mild or localized disease, treatment may be limited to gentle skin care and emollients.

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1 (and, therefore, lack of national economic incentive to find their causes and effective treatments unless banded together with other "orphan diseases")

2 At least there is no evidence currently for a genetic cause; however, many diseases may be modified or caused in part by genetic factors and this could be true of PRP.

3 Vitamin A is generally no longer used because of concern about toxicity at higher doses.

4 See Cohen and Prystowsky. Pityriasis rubra pilaris: a review of diagnosis and treatment. J. Am Acad Dermatol 20: 801-807, 1990.

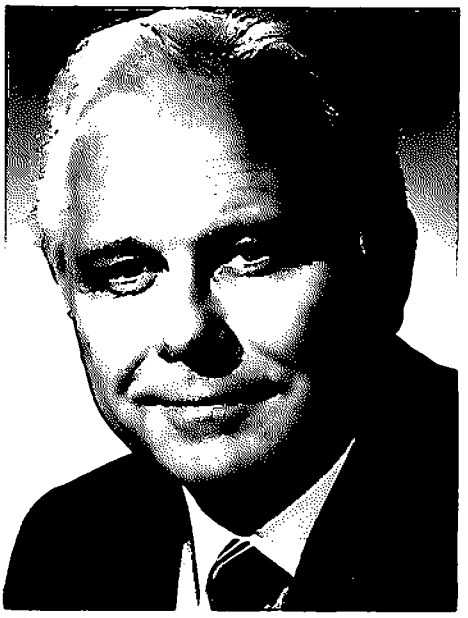
A Journey Through PRP Valley

by Tom Stuelpnagel

My journey through PRP valley started with fleeting shadows and a few boulders by the road. Gradually the valley walls grew steeper, blocking the sun, and the road narrowed to a path. Why me? And why so late in life? These were my laments as the disease took over one of my golden years at age 71.

At first folks thought I had just returned from Hawaii. Then, scaly blotches of about one-half inch in diameter formed on my shoulders and progressively merged. This was originally diagnosed as Pityraisis Rosea--a skin disease that lasts about two months in the shoulder and trunk area and disappears on its own. I sought more information in a dermatology book in the library. On the page next to the "Rosea" was a photo of a patient with PRP. My reaction was one of horror--and relief that at least I didn't have that problem.

As my condition continued to worsen, my doctor began to suspect PRP. He arranged to have me examined by a team of local dermatologists. Eventually my skin turned to a mottled red and salmon color from head to toe, and the secret was out. I learned to refer to PRP as a 'disorder' because friends and acquaintances seemed to be more comfortable with this term. In any event, it's definitely " not catching". PRP acts like a deep sunburn with continuous burning, itching and scaling--month after month, 24 hours a day. My journey through PRP valley lasted 16 months, during six of which I spent much of the day in a plastic suit with ointment slathered on my body.



Thomas R. Stuelpnagel is retired, age 73 and former president of a helicopter company. He resides in Avila Beach, California with his wife. Shirley.

My doctor prescribed Tegison (synthetic vitamin A), Triamcinolon .1% ointment, Hydroxyzine HCL for the control of burning and itching, and the plastic suit. The first brand pulled apart after a week of use. Two other brands didn't do much better and all were too small for my six foot body. A friend in the iris farm business suggested that a zip-up plastic suit used for spraying pesticides might be a better body bag. Sure enough, it was much roomier and easier to get into. It was so comfortable I tended to stay in it for up to eight hours per day--and reasonable in cost at $8 each. At one point, I tried the bright yellow color thinking it would make my day sunnier. This proved to be a mismatch with red skin, so I retreated to gray. Each suit lasted about two weeks and could be turned inside-out and washed daily to an acceptable (less than icky) condition for reuse. Another aspect of this body bag was its lightweight fabric. It is made from "Tyvex" material and weighs only 8 ounces with the hood cut off. In general, clothes fit nicely over the bag, although I didn't try a business suit. Just getting into the suit was a chore and the re-entry day after day was icky to say the least. Getting ready in the morning took an hour, but the resulting comfort level made it time well spent.

Another discovery was the use of thin plastic bags, as supplied in grocery stores, to wrap my feet. This was a better solution than the Saran Wrap I had struggled with initially. I put socks on over the bags. My bag supply proved inadequate from normal marketing --and I began to feel guilty about stealing them--so I bought a whole roll from the market for $8.00. Then I felt safe. Then there was the need for footwear that did not rub on my sensitive toes. The solution was to cut the top area of my athletic shoes.

Gloves were another need because, like my feet, the skin had cracked and they needed continuous moisture. I went through dozens of white cotton gloves and latex gloves before finding that the thin plastic gloves worn by food handlers were ideal. I used several sets per day, but the cost at 2 1/2 cents per pair was quite tolerable.

I credit the zip-up body bag, foot bags, modified shoes and plastic gloves for my relative comfort and ability to continue everything I had been doing--except my daily swimming. Later, I was able to play some nine-hole golf with golf gloves on both hands. Continuing with normal activities made the days go faster.

Skin creams and ointments were another adventure. I tried dozens of brands and ran side-by-side comparisons. Interestingly, I found that plain old Vaseline was as good or better than any other and there was no risk of absorbing chemicals through my skin such as the steroid in Triamcinolon.

The PRP or my medications caused the loss of some hair on my head and all body hair. My barber no longer had me as a regular customer and a daily shave was unnecessary except for morale. My finger and toe nails eroded severely and my lower eye-lids protruded (ectropion). Leg swelling occurred (edema).

see **PRP** page 8

The PRP Valley

PRP from page 7

At one point I could only walk a block due to muscle pain. My body cycled erratically from hot to cold, forcing me to take a blanket and a cap along to the movies.

When after six months on Tegison, my doctor and I did not see any progress, we agreed to switch to a modest dosage of Methotrexate (MTX). We were being cautious because MTX is a chemotherapy drug, used principally for breast cancer and rheumatoid arthritis, and its side effects are worse than Tegison. Studies show that it can be tolerated in the typical patient at a cumulative level of two grams before a liver biopsy might be needed. This computed to about a four year period for my dosage so I felt comfortable with the medication.

All told, I was on the MTX medication for about six months with reduced dosages overtime. My condition started to improve after about a month. At that time I stopped the use of the plastic suit. After two months on MTX, I could sleep through the whole night without re-lubeing. Finally after three months with MTX the peeling stopped and I started to gain confidence that the end might be in sight. It turned out that I had no side-effects from Tegison or MTX either physically or as measured in monthly blood tests.

Progress was so good that my wife and I proceeded with our plans for a cruise we had booked before I became ill. The cruise toured the "Spice Islands" departing from Singapore. The path through PRP valley had become wider. The cliffs were gone and the sun was out. On Valentine's Day aboard the ship I stopped the MTX and proclaimed myself cured (cautiously!) and celebrated with a dry martini. I was now a year older and wiser and very grateful to be back on the highway to health!

I even acquired some modest benefits. My sensitive digestive system has become robust, and my head and body hair have returned in greater quantity and are darker in color. Moles, age spots, and other skin defects have vanished. My finger and toe nails are growing back slowly. It is a pleasure to get dirt under my nails again and to open my pocket knife with a thumbnail instead of a dime. Forced by PRP to abandon my electric razor, I found today's razor blade does a better job in less time so I am continuing with it.

The cause of my recovery will remain in doubt since two different medications and possible self healing were involved. Sustained recovery is also unproven, but I am viewing the results I had with MTX as a reason for optimism. My encouragement to those suffering with PRP is that when recovery from Pityraisis Rubra Pilaris finally starts, it can proceed rapidly.

What's New, What's Hot, What Works!

... a regular feature about skin care problems and solutions.

The following letter was submitted by Kevin and Gary Allard-Mendelson in response to the question from Karen Rusch, who asked for tips on treating skin and wax build-up in her son Matthew's ears.

Our son Richard had a similar problem. In fact, when we adopted Richard, he could barely hear because the skin and wax build-up had almost completely blocked his ear canal. At the time, Richard was four and not yet able to speak due to his inability to hear.

When we took him to our pediatrician, he cleaned his ears and suggested we place two drops of liquid Colace, a mild stool softener, in his ears once every other day. While we thought this was quite an odd recommendation, it has proven very effective at loosening the skin and wax build-up in his ears, allowing us to keep his ear canals quite clean and clear in between doctor visits.

We would recommend conferring with Matthew's pediatrician before you try this method, but for us it has worked quite well.

Sophia Mamais is 24 years old and has ichthyosis vulgaris. She reports that, after initial trial and error, she has had success with the following treatments:

My doctor found that Retina A and petroleum jelly (which he mixes), helped my skin. The flakes and lines practically dissolved and the skin looked better and hurt less. This is what I have been using for 21 years and my skin looks better than ever. I apply this ointment twice a day, immediately after a shower. For my face, I apply either Eucerine or Almay Cold Cream, which has helped relieve the tightening. I also have found that Bath and Body Work's has some great body soaps which are not only soothing but smell nice.

F.I.R.S.T.'s 1998 NATIONAL CONFERENCE
JUNE 19, 20 & 21 -- PHILADELPHIA

CONFERENCE FACULTY

Sherrie J. Bale, Ph.D., Chief, Genetic Studies Section, Laboratory of Skin Biology, National Institute of Arthritis and Musculoskeletal and Skin Diseases NIAMS at the National Institutes of Health. Dr. Bale is involved in research into the genetic basis of hereditary disorders, particularly those affecting skin and musculoskeletal systems.

John DiGiovanna, M.D., Director Dermatopharmacology Division, Department of Dermatology, Brown University, School of Medicine in Providence, R.I. and Adjunct Investigator, Genetic Studies Section, Lab. Skin Biology, NIAMS. Dr. DiGiovanna has been intensively involved in both clinical (improving treatment) and basic science (better understanding causes) research of skin disorders, and has a particular interest in retinoid therapy. He serves on F.I.R.S.T.'s Board of Medical Editors, the Advisory Committee of the National Registry for Ichthyosis & Related Disorders, and has recently joined F.I.R.S.T.'s Medical Advisory Board.

Philip Fleckman, M.D. A member of F.I.R.S.T.'s Medical Advisory Board, Dr. Fleckman is Principal Investigator/ Director of the National Registry for Ichthyosis & Related Disorders. An Associate Professor of Dermatology at the University of Washington School of Medicine in Seattle, Washington, he also serves as Chief Editor of Dermatology Online Journal. Research interests include the pathophysiology of ichthyosis vulgaris and other inherited disorders of keratinization.

Geoffrey Hamill, R.N., Registry Coordinator for the National Registry for Ichthyosis & Related Disorders in Seattle, Washington, Mr. Hamill has worked with investigators on a variety of clinical research protocols, including trials for new medications, and began his career in 1986 as a neonatal/ antepartum nurse.

Amy Paller, M.D. Head of the Division of Dermatology Children's Memorial Hospital in Chicago, and Professor of Pediatrics and Dermatology at Northeastern University Medical School. Dr. Paller restricts her clinical practice to children with skin disorders and specializes in genodermatoses, immunodeficiency and collagen vascular disorders. She is a member of F.I.R.S.T.'s Medical Advisory Board.

Howard B. Pride, M.D. Department of Dermatology, Geisinger Medical Center. Dr. Pride is chairman of the American Academy of Dermatology's Camp Discovery Committee, which organizes three camps for children with chronic skin disorders; Camp Knutson (MN), Camp Horizon (PA) and a new camp in California. He is also President of the Camp Horizon Steering Committee.

Eugene Van Scott, M.D. A clinical investigator, Dr. Van Scott has established a private practice and research laboratory in the Philadelphia area and continues to focus on conditions, including ichthyosis, not helped by available therapies. Dr. Van Scott is a member of F.I.R.S.T.'s Medical Advisory Board. He discovered that alpha hydroxy acids (AHAs) produce a response in skin conditions with faulty keratinization processes. He is a co-founded of NeoStrata Company, Inc., which has developed a line of AHA products.

HOTEL ACCOMODATIONS

Hilton at Cherry Hill
2349 West Marlton Pike
Cherry Hill, NJ 08002

For hotel information and reservations, call toll free at 1-800-HILTONS or the reservation desk in Cherry Hill at 609-665-6666 . Identify yourself as a F.I.R.S.T. member to receive our special conference hotel rate of:

- $80.00/night, single or double occupancy
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New Jersey state sales tax of 6% not included.

This rate is guaranteed from June 16th to 22nd. For extended stays, the rate is available on a *space available basis* so be sure to book your rooms well in advance.

There is an early check-out fee of $50.00, which may be avoided by notifying the hotel of any change in your reserved check-out date on or before check-in.

The F.I.R.S.T. National Conference rate of $80.00 is only guaranteed for rooms booked a minimum of one month in advance of the conference date - by May 18th, 1998.

If you have questions, or if you experience difficulties booking your room, please call F.I.R.S.T.'s National Office in Ardmore, PA at 1-800-545-3286.

AIRLINE RESERVATIONS

US Airways has the most flights into and out of Philadelphia International Airport. As the official carrier for F.I.R.S.T.'s National Conference they are offering the following discounted fares on roundtrip travel from June 14, 1998 to June 26, 1998:

- **5%** off the lowest applicable published fares
- **10%** off the same fares with **60 days advance reservations**

To obtain these discounts, you must call US Airways' Meeting and Convention Reservation Office at (800) 334-8644 and refer to Gold File Number 34630417. There are also discounts on First Class and Business Class fares. Certain rules and restrictions apply. Call US Airways for details.

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Sky Shuttle, 1-800-825-3759, will provide transportation between Philadelphia International Airport and the Hilton at Cherry Hill for a discounted rate of $10.00 per person, each way.

Advanced reservations are recommended, but not required. Upon arrival at the airport, travelers can also make arrangements from the "Ground Transportation Counter" in the baggage claim area (Dial #15 on the white phones at the counter). Indicate that you are participating in the F.I.R.S.T. family conference.

PRELIMINARY PROGRAM
NATIONAL CONFERENCE 1998 - PHILADELPHIA

This is a preliminary schedule only. Meeting times may vary slightly in the final program, which will be distributed prior to the conference to all registrants. Individual sessions and time slots may also change.

Friday June 19th

4:00 - 6:00 p.m.	Registration	
6:30 - 9:30 p.m.	Reception	Light snacks, cash bar, and plenty of time to mingle.

Saturday June 20th

8:30 - 9:15 a.m.	Breakfast	Continental breakfast and late registration
9:15 - 10:15 a.m.	General Session	♦ *Opening remarks* ♦ National Registry for Ichthyosis and Related Disorders - Philip Fleckman, M.D. ♦ Camp Discovery - Howard Pride, M.D.
10:30 - 11:45 a.m.	Breakout Session	A. Registry Enrollment Workshop - Geoff Hamill, R.N. B. The Genetics of Ichthyosis - Sheri Bale, Ph.D. C. Camp Discovery - Howard Pride, M.D. D. To be determined
12:00 - 2:00 p.m.	Lunch	*with Awards Presentations*
2:15 - 3:00 p.m.	General Session	Treatment of Ichthyosis - Moderated by Amy Paller, M.D.
3:15 - 4:30 p.m.	Breakout Session	A. Mom's Discussion Group B. Women's Discussion Group C. Dad's Discussion Group D. Men's Discussion Group E. Panel Discussion: insurance, employment, and legal issues.
6:00 - 9:00 p.m.	Dinner	*followed by entertainment*

Sunday June 20th

8:30 - 9:15 a.m.	Breakfast	Continental breakfast
9:30 - 10:15 a.m.	General Session	Genetics: Past, Present, and Future - Sheri Bale, Ph.D.
10:30 - 11:45 a.m.	Breakout Sessions	A. The ABC's of Retinoids - John DiGiovanna, M.D. B. Talking with your doctor - Geoff Hamill, R.N. C. To be determined D. To be determined
11:45 - 12:15 p.m.	Closing Remarks	

F.I.R.S.T.

1998 NATIONAL CONFERENCE

June 19, 20, 21

Philadelphia, PA

Please Print

Name: _____

Address: _____

City: _____ State: _____ Zip: _____

Phone: Day_____ Evening _____ Country: _____

NAMES OF THOSE ATTENDING	adult	child	Birthdate of Each Child	Check here for those affected with ichthyosis
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐

Type of ichthyosis, if known: _____

Number of Adults: _____ (18 & older) x $95 = _____

Number of Children: _____ (1 through 17)* x $50 = _____

TOTAL = _____

**There is no registration fee for children under one year of age*

Please Return Registration Fees With Your Completed Form

Make Check Payable to F.I.R.S.T. In U.S. Dollars, Please

Cancellations Will Be Honored With Full Refunds Until The Advance Registration Deadline:

MAY 18, 1998

☐ I will need child care for _____ children (over one year of age).

NAME	BIRTHDATE

☐ I am an adult willing to help at the conference for an hour or two (child care, reception table, etc).

NAME

_____

_____

☐ I am a teen or young adult and I'd like to participate in a discussion group with others my age.

NAME	AGE

PLEASE MARK THE APPROPRIATE BOXES

☐ This is the first National Conference I have ever attended

☐ I have attended National Conferences in the past (City/year): _____

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☐ I will be staying at the Hilton at Cherry Hill in New Jersey

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$80.00/night, single or double occupancy

$10.00/night, each additional person.

Your reservation must be made by May 18th, 1998

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Children's Success Story

By Bailey Rae Jones

Hi! My name is Bailey Rae Jones. I'm eleven and have been living with Lamellar Ichthyosis all my life. I take tap and ballet from April's School of Dance, and flute lessons from Pat Luckenbill. I also take voice and piano lessons from Asa Carns. I'm also in the Youth Choir that my mom directs at my church. In my spare time I love to read, shop, watch ice-skating, write stories and eat tacos. I'm in sixth grade and a straight-A student. I love school most of the time. My favorite subjects are Spelling and English. This year the sixth grade is putting on the play "The Nutcracker". I'm one of the sugarplum fairies. I also get to play a piano solo at the beginning of the show.

This past summer I went to Camp Horizon. The reason I went was because my penpal from Bermuda (Laura) was going. It was cool to meet her for the first time! Camp Horizon was really fun. I got to do things I normally don't get to do at home, such as archery, paddleboating, fishing, and we even had a nature program! So parents, let your kids come to camp, it was a lot of fun!

Laura Ashton (left), Jean Cahill, and Bailey Jones.

I get stared at just about everywhere I go. Sometimes I'm almost on the verge of tears because of that. Other times me and my mom get mad and smile and wave at the kid. Sometimes that works to stop them, other times the kid waves back. I'm pretty much used to getting stared at by now.

When I grow up I want to become a famous ballerina, or an author, or maybe even an ice-skater. Even though I have ichthyosis, I still am a kid on the inside, but just look a tiny bit different than everyone else.

Camp Discovery 1998

July 11-18 at Camp Knutson, Minnesota
August 16-22 at Camp Horizon, Pennsylvania

Camp Discovery, a camp for children with chronic skin disorders sponsored by the American Academy of Dermatology, will be held for the sixth summer in 1998 at two locations. Camp Knutson, in Minnesota, accommodates 50 campers each summer on a beautiful lakeside property which has a sandy beach and several boats. Camp Horizon, in Pennsylvania, features a fishing and boating pond, outdoor swimming pool, and covered pavilions. Both camps offer a wide range of organized, supervised activities and plenty of opportunities to meet and make friends with other children suffering from various skin diseases.

The camps are staffed by experienced dermatologists, pediatricians, and nurses to make sure that the medical care of the campers is not neglected. Routine and emergency care are readily available. Help in performing dressing changes and supervision of oral and topical medical regimens are facilitated by the medical staff.

A staff of counselors oversees the campers' daily activities. The ratio of campers to counselors is kept intentionally small to assure close surveillance. Many of the counselors and volunteer helpers have skin diseases themselves and serve as excellent role models for the campers.

Children 10-13 years of age may attend Camp Knutson and 8-13 years of age Camp Horizon. Counselors must be at least 18 years of age, and there is limited room for younger junior counselors. Inquiries regarding camp may be directed to Howard Pride, M.D. at 717-271-8050 or Debbie Kroncke at 847-330-0230.

A third camp site will be starting in the Los Angeles, California area in 1998. Children 8-13 years of age may attend this shorter camp on August 27-30. More information can be obtained from Jenny Kim, M.D. at 310-825-5420.

National
REGISTRY for
ICHTHYOSIS
and Related Disorders

Registry Director: **Philip Fleckman, M.D.**
Registry Coordinator: **Geoff Hamill, R.N.**

University of Washington
Dermatology/Box 356524
Seattle, WA 98195-6524
E-mail: ichreg@u.washington.edu
URL: http://weber.u.washington.edu/~geoff/ichthyosis.registry/

1-800-595-1265

The National Institutes of Health (through its National Institute of Arthritis, Musculoskeletal and Skin Disease—or NIAMS) is sponsoring a National Registry for Ichthyosis & Related Disorders. The goal of the Registry is to develop a clinical database of individuals affected with the ichthyoses and other rare skin disorders (that is, a roster that compiles information about people with these disorders).

The Registry's purpose is to improve methods of diagnosis and treatment of the ichthyoses. We do this by creating a list of affected individuals. This should have a profound impact on research in the ichthyoses and related disorders, since one of the greatest obstacles facing researchers doing studies on rare diseases is locating affected individuals willing to help them out.

The Registry should solve this problem by creating a central resource containing diagnostic information about the individuals who have enrolled. Researchers can then easily locate affected individuals, as well as obtain information about the frequency and involvement of their disorders. In addition, even if you choose never to participate directly in research studies, the information you provide the Registry should prove valuable in better understanding these disorders.

The Registry will NOT give identifying information out to researchers. Instead, those enrolled in the Registry will be contacted ***by the Registry*** and informed of any appropriate research projects. At that time, you can decide for yourself if you wish to contact the researcher involved.

Please contact the Registry Coordinator, Geoff Hamill, to ask more questions and to obtain enrollment forms.

Talking With Your Doctor

by Geoff Hamill, R.N.
National Registry for Ichthyosis and Related Disorders

For many of us, our doctor is our point of access to health care. We go to him or her for information about problems that we have, and for advice and instruction on how to take care of our family and ourselves. This can be an intimidating process, especially in these days of HMO's and health care plans. The days of having one doctor for all of our care for most of our lives are pretty much gone, so the chance of getting to know and trust a single individual over a long time rarely happens anymore. So how can a person get to feel comfortable with the people they depend on for care? It is possible, and I'd like to share with you some suggestions that can help.

To start with, let's acknowledge a few truths. When we go to see a doctor, we usually want to talk about something that concerns us. We are in a position where we feel vulnerable, and at times we can feel a little desperate. Any information we can get has got to be better than what we don't know now, right? And these are busy people, with a lot of patients to see and a lot on their minds, and we're pretty lucky to get to talk with them at all! Does this sound at all familiar? It's OK if it does, because we all have things we are scared about, and asking for help from someone you don't know can be very hard sometimes. If you've ever felt like this, I can tell you you're not alone.

So how can we get our concerns addressed in a way that satisfies what we feel we need? By being honest and direct about what those concerns and needs are, and by asking for specific help in meeting them.

I know it sounds trite, but communication is everything. If your doctor doesn't know what you are worried about, then no matter what he or she does you may not get the information and advice that you were looking for. If you are willing to share your concerns with your doctor, you will provide them with a clearer picture of the reasons you came to ask their help. For some people, making a list to bring with them to their appointment can help. For others, practicing for a moment what they want to say before they are in the office is a good way to organize their thoughts. Even having a list of questions with space to scribble answers below them can be a useful tool in keeping your thoughts together during that short time when all that information is going back and forth between you.

I know this is nothing new to many of you, and that even with the best of planning and intentions you might be receiving care from someone you feel "uncomfortable" with. It happens (and for people with unusual or rare problems it may seem like it happens a lot!), and it helps to have some strategies to deal with it.

Even if you are not comfortable, start by realizing that your doctor has had a rigorous and thorough education in dealing with health problems, and that (all cynicism aside) they wouldn't be doing what they do without having a real desire to help people. So it's likely that you both have the same goal in mind: solving your problem. The next step is to try to be clear in communicating with them about your concerns. When they ask questions about your problem, try to be as specific and thorough as possible. Let them know that you will share all of the information that you can, and that you expect them to let you know when they have a thorough understanding of what's

involved. They can't give you their best effort if you don't provide them with all the clues to the puzzle.

Perhaps the hardest part of all is when you've tried everything you can, and because of your health plan, or the size of your town or your budget, you still need to receive care from someone you feel is not meeting your needs adequately. These limiting situations can happen more frequently when your issues involve unusual or rare disorders like ichthyosis. My suggestion is that you try and use whatever resources you have to enlist the help of other people in addressing your concerns, and that you are as clear as you can be about whatever specific needs you still feel have not been addressed. If you don't understand a term or the reason for a type of treatment, then ask. Be honest about not understanding what might be going on, and be clear that the reason you are there asking for help is because you feel you can't deal with your issue by yourself. Try to control your temper, but remember it's a good thing to share your frustrations or fears with your doctor because that's the reason you went there in the first place!

Always bear in mind that you don't have to be alone in this. Call F.I.R.S.T. for someone to talk to that's been there as well, or for a referral to a dermatologist. The Registry may also be able to suggest a local or regional 'expert' who can work with you and your doctor in finding solutions. It's easy to get caught up in feeling alone and helpless, but you aren't. We are all in this together to help each other, and given half a chance that's exactly what we will do - help each other.

Geoff will be discussing this topic at our National Conference in June, where he will also provide information about enrollment in the National Registry for Ichthyosis & Related Disorders.

Our heartfelt sympathies to Stephen and Deanna Simonette of Parkersburg, West Virginia who lost their two week old son, Dominic Michael, to complications of Harlequin's Ichthyosis.

F.I.R.S.T. neither recommends, nor endorses, products or services advertised in *Ichthyosis Focus.*

REGIONAL SUPPORT NETWORK (RSN)

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Cathy Lopez
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South Carolina
Nancy Bryant
803-559-9369

Wisconsin
Theresa Tremmel
414-538-4922

RSN Profile

Shari Gilevich
Region Seven Coordinator

Hello to members of the F.I.R.S.T. organization. My name is Shari Gilevich and I have been volunteering as the Region 7 coordinator for a few years now. I was born in San Antonio, Texas (way back in 1951) and grew up in southern California with three sisters and two brothers. I was the only one in the family with a skin condition. While the condition created some miserable times when I was young, I often felt special because of all the extra attention I would get! It was not until I was in my early 30's that my condition was diagnosed as EHK.

I attended college at the University of California in Riverside, majoring in anthropology. After one and a half years working in the VISTA program (anyone remember "Volunteers in Service to America," our local Peace Corps?), I attended the University of Oregon and earned an M.A. in geography. At the University of Oregon I met Manning Welsh, my husband of nearly 18 years. We have two wonderful daughters, Madeleine age 12, and Laura age 9. I currently work as a land use planner and find myself, like most other folks I know, always on the *run with* kids, work and volunteer work.

Shari with her daughters Madeleine and Laura.

The F.I.R.S.T. group has been invaluable for meeting other people experienced with skin conditions as well as keeping me apprised of the latest information. Our daughter Madeleine, who inherited EHK, has made several pen pals through the group and I am grateful to all those who share their time and friendship with us.

. . . out of the darkness, into the light . . .

N.O.R.D. REPORT

The National Organization for Rare Disorders, Inc. (N.O.R.D.), dedicated to helping people with rare (orphan) diseases.

FDA MODERNIZATION AND ACCOUNTABILITY ACT

Despite an attempted 34-hour filibuster by Senator Edward Kennedy, the FDA Modernization and Accountability Act (S.830) passed the Senate, and three similar bills passed the House of Representatives. The House bill was split into three bills, each covering one topic: pharmaceuticals, medical devices and foods.

Despite the hoopla in Washington about this law, it will not speed the approval of new breakthrough drugs any faster then they currently are. The law simply codifies procedures that the FDA has implemented for several years that prioritize drugs for serious and life threatening diseases over ordinary drugs. Among other things, the bill would:

- Change the standards for testing new drugs. Currently manufacturers must perform two or more well-controlled clinical trials before applying to the FDA for marketing approval. The new law requires only one or more clinical trials.
- Allow drug companies to make economic claims without regulation or validation of the claims, such as claiming an expensive drug actually saves medical expenses because of reduced hospitalizations, etc. The FDA would have the power to force a company to stop making claims that the FDA deems false or misleading.
- Allow pharmaceutical companies to claim that their drugs are safe and effective on diseases for which they have not been tested (off-label claims). They can make off-label claims for up to five years. Unfortunately, the bill would allow companies a waiver so they never have to perform research if the disease is rare. Thanks to Henry Waxman (D-CA), the language of the House bill is such that a company can get a waiver if they prove the required research is economically unfeasible. Thus, if the House language is adopted, a waiver will not be based on the low prevalence of the disease.
- Require drug companies to give the public six-month notice if they no longer want to manufacture a drug. This NORD-supported amendment is important as it will give physicians time to switch the patient to another drug and/or allow for us to locate another manufacturer who may be willing to adopt the drug.

NORD fears this bill foreshadows a regression to the early 1900s when food and medicine manufacturers could freely proclaim they cure everything from cancer to baldness without a shred of proof, nor any guarantee their products are safe for their intended use. The FDA assures us, however, that these changes are benign, and they will do their utmost to monitor the claims of companies to reduce any adverse impact on public health.

HEALTH INSURANCE COVERAGE SLIPS AGAIN

The U.S. Bureau of the Census reported on September 29, 1997, that the number of uninsured children under 18 grew to 10.6 million in 1996. This amounts to 15.8% of all children in the U.S. This is up from 13.8% in 1995. The total number of uninsured Americans was up 1.1 million to 41.7 million, or 15.6% of the entire population.

Excerpts from President Clinton's State of the Union Address

Tuesday, January 27, 1998

"And while we honor the past, let us imagine the future. Think about this -- the entire store of human knowledge now doubles every five years. In the 1980's, scientists identified the gene causing cystic fibrosis -- it took nine years. Last year, scientists located the gene that causes Parkinson's Disease -- in only nine days. Within a decade, "gene chips" will offer a road map for prevention of illnesses throughout a lifetime...

Tonight, as part of our gift to the millennium, I propose a 21st Century Research Fund for path-breaking scientific inquiry -- the largest funding increase in history for the National Institutes of Health, the National Science Foundation, the National Cancer Institute...

As important as all this scientific progress is, we must continue to see that science serves humanity, not the other way around. We must prevent the misuse of genetic tests to discriminate against any American..."

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