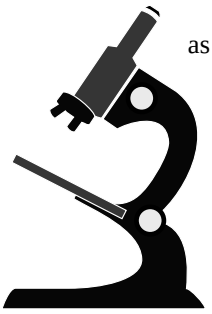
F.I.R.S.T.
Foundation for Ichthyosis and Related Skin Types

ICHTHYOSIS FOCUS

Vol. 19, No. 2 | A Quarterly Journal for Friends of F.I.R.S.T. | Spring 2000

F.I.R.S.T.'s 2000 Grant Recipient



Congratulations to Peter J. Koch, Ph.D. as this year's recipient of the foundation's $10,000 annual research grant. Dr. Koch's research project, *Desmocollin 1: An Epidermal Development and Homeostasis*, is being conducted at Baylor College of Medicine in Houston, TX. The project's summary is that desmocollin 1 is a major protein component of desmosomes, cell adhesion structures that are crucial for the mechanical stability of certain tissues such as skin. Dr. Koch proposes to analyze the specific contribution of desmocollin 1 to cell adhesion by generating and analyzing genetically engineered mice that are deficient for the expression of this protein. We look forward to hearing more about this project in a future issue of the Focus.

It's not too late... Register now for 2000 Family Conference

The 2000 Family Conference is less than two months away! We urge you to make your reservation today. We have changed the hotel location to the beautiful Doubletree Hotel in downtown Philadelphia. There are plenty of rooms still available at our special discount rate ($80.00). Plans are being finalized and we're looking forward to an exceptional National Conference this year. The program will include general sessions and breakout workshops on a broad variety of topics and child care is available for the little ones. We hope you can join us. It is a great opportunity to meet other members, learn more about the disease and speak with physicians who specialize in ichthyosis.

See pages 17 & 18 for more details and registration information.

F.I.R.S.T. Goes to Washington

On April 5, Matt Gray traveled from his home in Nebraska to testify before the House of Representatives' Subcommittee on Labor, Health and Human Services, Education and Related Agencies. Matt was accompanied by his mother, Beth, brother, Mitch and grandfather, Roger Blobaum. Matt and his family were able to make this trip into a "mini" vacation since his grandfather lives in the area. Once at the capitol building, the Gray's were met by Jean Pickford, Executive Director of F.I.R.S.T.

Matt spoke to Chairman John E. Porter (IL) and other members of the subcommittee about his life with ichthyosis. Matt is nine years old and was born with lamellar ichthyosis. He is the author of Matt's Message for Kids, an internet accessible story about what it is like to be a child with ichthyosis. Matt talked about his daily routines of bathing and applying lotions. He shared a personal experience about how he forgot to apply lotion to his hands at school one day. His hands cracked open, bled and he could not hold his pencil.

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Correspondence Corner

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

First of all I want to thank all of you at F.I.R.S.T. for a great newsletter. I'm very thankful for getting it. I have erythrokeratodermia variabilis and I'm the only one in Finland having this particular disease, so it's quite lonely down here! I have never seen or heard about anybody having the same disease as me anywhere, because to be honest with you my doctors never answer my questions about other similar diseases in Finland or elsewhere.

It was great to read Sofia Mamais letter in the last issue of Focus. I must say that I completely agree with you. Today, I'm 22 years old, studying and living a satisfying life with friends seeing me as myself and not as some weird girl with a terrible skin and red spots all over her face and body. For the past four years I've been taking one Neotigason per day (probably just Tegison in the U.S) and as you all know the medicine isn't allowing you to live a really normal life... though my skin is in good condition and I feel great. It's quite good that we didn't have a very cold winter this year here in Finland, because winters are unbearable to me when it's cold and windy.

If there is someone having the same disease as me, I would be very grateful if you would contact me either by e-mail or snail-mail. I'm also very interested in different treatments and gene-connections. For the past three years I've questioned both my medical treatments and my doctors interest in this disease. I usually have to explain to my doctors what this disease is all about and what I've been treated with and honestly I think they've lost interest in my disease because of the rareness.

Sunny greetings from Finland,

Malin Berglund
Stora Robertsgatan 14 A 6
FIN-00120 Helsingfors
e-mail: malin.berglund@eduskunta.fi

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

I have had lamellar ichthyosis since birth and am now 24 years old. I control the flaking by using petroleum jelly, however I have not had success with controlling the redness which is really giving me an inferiority complex. Any hints regarding the redness? Please let me know at berman@echo-on.net. Thank you.

Jerry Joseph Berman
Ontario, Canada

Dear Friends:

We want to thank the ichthyosis group! I still recall our first meeting in Schaumberg, Illinois. We met many caring and wonderful people who truly have the interest of those afflicted with this disease at heart. Just knowing that there are people who can share one's concerns and worries is a great comfort. As much as Melissa and I would love to join you in Pennsylvania this summer for another conference, we find that we will be unable to afford to make the trip. I know how much effort and hard work goes into planning such an event. Believe me, your work is much appreciated! I know you must get several requests and suggestions as to where to hold the next conference, and I know it is impossible to please everyone. Hopefully, we can attend a future conference. Our thoughts and prayers are with you.

Kathy Lauters
West Bend, Wisconsin

F.I.R.S.T.:

I think that your web site is truly fabulous! It looks great and is easy to use. F.I.R.S.T. has been here for us even before we adopted my daughter in 1997. She has lamellar ichthyosis. Your organization sent us information on ichthyosis and referred us to adults and parents that adopted children with ichthyosis. It was a wonderful support system to prepare us for her arrival.

She came to us at 9 months old. Your organization lead us to Michelle's dermatologist, Dr. Amy Paller. She is a wonderfully knowledgeable doctor who has truly helped Michelle get to where she is now. Just a note of "Thanks!" We appreciate you being there to make a difference!!

Jennifer McMillan
Chicago, Illinois

Editor's Note: Visit our new website at www.scalyskin.org.

Donor Acknowledgement

In our last newsletter, Winter 2000, F.I.R.S.T. listed the names of donors who contributed over $100.00 in our 1999 fiscal year. When the computer conducted its search for donors, the following names were inadvertently omitted from the list. F.I.R.S.T. would like to acknowledge these donors and apologize for any inconvenience this error may have caused.

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

F.I.R.S.T. goes to Washington continued

Matt's mom, Beth, a new board member of F.I.R.S.T., thanked the subcommittee for their strong support over the past few years. As a result of that support, we have a national registry for ichthyosis and related disorders and tremendous progress has been made in the area of human genetics and gene therapy.

On March 1, Leonard Milstone, M.D. and Jean Pickford visited Capitol Hill for NIAMS Day 2000. As part of the Coalition of Patient Advocates for Skin Disease Research (CPA-SDR), F.I.R.S.T. and members from other organizations met with the director of the National Institute of Arthritis, Musculoskeletal and Skin Diseases (NIAMS), Dr. Stephen Katz. Then, members of the CPA-SPR spread out on the hill to meet with senators and representatives to lobby for increased funding for NIAMS and the NIH.

F.I.R.S.T. recognizes this subcommittee's strong history of bipartisan support for medical research funding and the National Institutes of Health (NIH). Recently the identification of the genetic defects underlying both Darier and Hailey-Hailey diseases have been identified. Although both diseases are relatively rare, the fact that both are due to mutations in genes that regulate cellular levels of calcium in the epidermis has greatly increased our understanding of the regulation of cellular differentiation. Additional genetic mutations that cause other forms of ichthyosis have been identified and scientists and physicians have a much better understanding of the disease process.

There is much excitement about this progress, and about the current research into gene therapy. There is hope about the possibility for an effective treatment or cure on the horizon, but at this point it is still just hope. We continue to be frustrated by the lack of effective treatment options.

Biomedical research is the foundation upon which all medical care is based. Our Nation's biomedical research infrastructure is an intricate relationship of academia, industry, and the federal government. The NIH serves as the primary source for basic research through universities and independent research institutions. This synergy has alleviated suffering for millions of Americans by fostering the development of innovative treatments, including drugs and vaccines as well as fostering hope for those who continue to suffer.

If the NIH is to unlock the mysteries of disease and to translate the recent discoveries of the research bench into new treatment for the bedside, it is necessary that the appropriation for the NIH be a sizable, sustained and stable effort. We hope that Congress will keep the faith with its constituents, and increase the NIAMS, a branch of the NIH, budget to $405 million.

These two trips to Washington went a long way toward increasing awareness for ichthyosis and the need for skin disease research funding.

You can help…

By writing a letter to your senator or state representative, you can make a difference. Our government needs to hear from people who will benefit from skin disease research to appreciate the importance of this issue. Contact your local state offices to obtain your senators or representative's address.

Darier disease gene: A most unusual suspect encoding a calcium pump

by Alain Hovnanian, MD, PhD, Wellcome Trust Senior Clinical Fellow, Wellcome Trust Centre for Human Genetics, University of Oxford, Roosevelt Drive, Oxford OX3 7BN, UK tel: (44) 1865 287 511; fax: (44) 1865 287 501; e-mail: alain.hovnanian @well.ox.ac.uk

Clinical features of Darier disease

Darier-White or Darier disease (DD) was first described by Darier and White in 1889. They reported a specific inherited skin disorder transmitted through families in an autosomal dominant manner. The disease is characterized by the development of itchy warty papules and plaques in seborrheic areas and is associated with distinctive nail abnormalities and frequent palmo-plantar pits (1). The keratotic lesions affect the upper trunk and scalp, are often malodorous and are precipitated by heat, sweating, sunlight and stress. The nail abnormalities are constant and highly diagnostic of the disease, consisting of red and white longitudinal bands with a distal notch. The prevalence of the disease has been estimated at 1 in 55,000, but a recent study in Scotland suggests that it may be as high as 1 in 30,000. Onset is usually between 6 and 20 years, often at puberty. Severity of the disease is variable between and within families, and the social impact of the disease can be devastating. Oral retinoids may improve the condition. However, in the absence of knowledge of the cause of the disease, no specific treatment is currently available.

The quest for the Darier disease gene

The cause of DD has eluded clinicians and scientists for many years. Microscopy studies of skin samples obtained from patients with DD have revealed separation between cells of the epidermis (superficial layer of the skin) and thickening of the epidermis. These observations suggested that molecules which mediate adhesion between epidermal cells might be involved in DD. However, genetic studies did not confirm implication of known genes encoding cell adhesion molecules in the disease. In the absence of obvious clues, the quest for the DD gene relied on finding its chromosomal location prior to searching for possible candidate genes in a limited region of a chromosome. The first step was achieved in 1993 when the DD gene was mapped to chromosome 12 by two independent groups, in a region which was not known to contain any obvious candidate gene (2, 3).

Subsequently, several groups, including ours, reported further refinement of this region. Several genes were known to map to this limited chromosomal region, but none of them appeared to be an obvious candidate. However, one of these genes encoded a calcium pump and was highly expressed in the heart, muscles and brain. We found that this pump was also highly expressed in the skin, which led us to search for mutations in this gene in DD patients. In fact, all the DD patients that we studied showed abnormalities in this pump, thus demonstrating that this gene is the defective gene in DD (4). These results disclosed a key and unsuspected role for this calcium pump in the skin.

The Darier disease gene encodes a calcium pump

The gene which is defective in DD is called ATP2A2. It encodes a particular type of pump which transports calcium ions from one compartment of the cells to specialized calcium stores where calcium is kept at high concentration. We and others have now identified more than 70 mutations in DD patients, the majority of which are different from family to family (5, 6). These mutations predict absence of synthesis of the pump, or production of an abnormal pump which is non-functional. The precise mechanism by which abnormal function of this pump leads to the disease is unclear. The pump is involved in maintaining highly concentrated calcium stores and in generating calcium oscillations upon external stimulation of the cell. These variations in calcium concentration are thought to lead to specific cellular responses which are currently being intensively investigated.

Patients with localized DD are mosaic for ATP2A2 mutations

Patients with lesions similar to DD but with a segmental distribution on one side of the body only have been reported. These lesions follow lines of migration of embryonic cells, which led to the hypothesis that these patients may have mutations in the DD gene in a small proportion of their cells. In support of this possibility, we have recently shown that such patients can carry a mutation of ATP2A2 in epidermal cells from affected areas only, and therefore are "mosaic" for these mutations (7). The risk of transmission of generalised DD is currently unknown, and will depend on whether the germline is affected.

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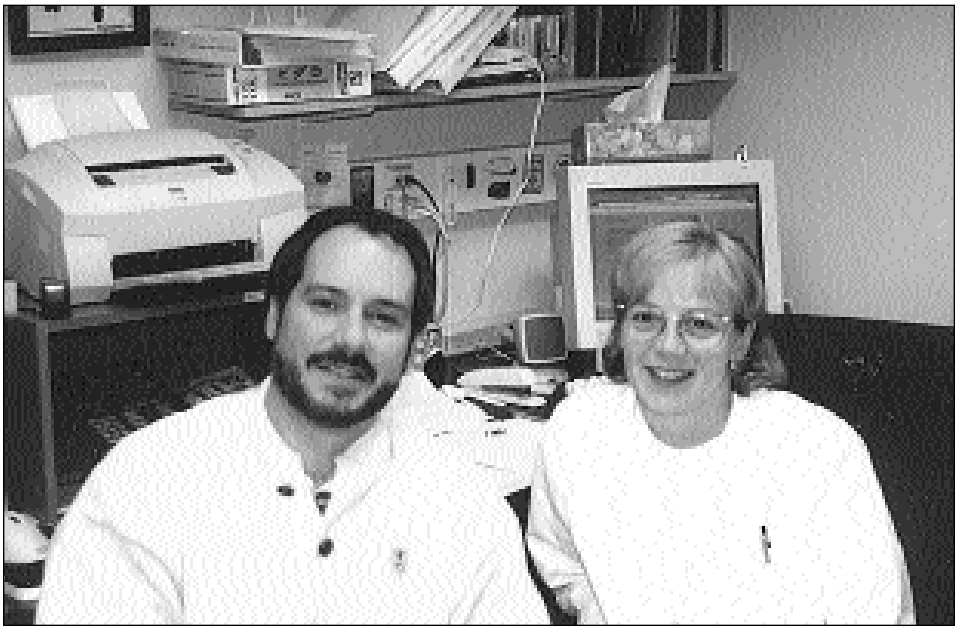
Molecular Diagnostic Services Are Now Available to the Ichthyosis Community



On March 6, 2000, Drs. Sherri Bale and John Compton opened the doors to their new business, GeneDx, Inc. in Rockville Maryland. GeneDx, Inc. is a full service genetic testing and diagnosis company dedicated to serving the diagnostic and genetic counseling needs of individuals and families with rare hereditary disorders.

You may remember Dr. Bale and Dr. Compton who, until opening GeneDx, were at the National Institutes of Health (NIH), in the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS). The two spent a combined total of more than 25 years at the NIH, where, with their colleagues (especially Dr. John DiGiovanna and Dr. Gabriele Richard) they have been leaders in ichthyosis research. With the participation of many F.I.R.S.T. families, their laboratory identified the genetic basis of Lamellar Ichthyosis, Epidermolytic Hyperkeratosis, a type of Palmar/Plantar Hyperkeratosis, and Erythrokeratodermia Variabilis.

The results of many years of research in the ichthyoses has lead to a better understanding of the molecular basis of scaling skin diseases, and will one day, we hope, lead to better treatments or even cures. But along the way, an important side benefit of the genetics research developed. This was the ability to use molecular methods to confirm clinical diagnoses in ichthyosis and to identify mutation carriers in families with recessive ichthyoses (like lamellar ichthyosis and Sjögren-Larsson syndrome). In addition, pre-natal diagnosis for many of these disorders also became feasible.

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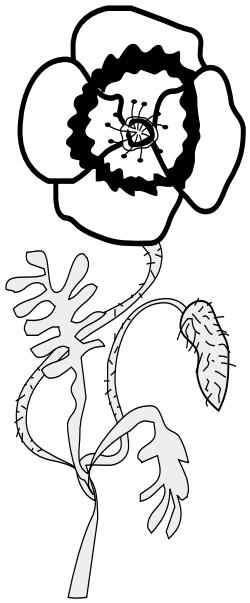
At the FIRST Family Conference two years ago, Dr. Bale explained why the research laboratories that were studying ichthyosis could not provide diagnostic testing services or even release results of mutation analysis to persons who had participated in genetics research. The restrictions are due to (1) a Federal law (CLIA '88) that permits only specially certified laboratories to report clinical test results which can be used for diagnostic or treatment purposes, and (2) a lack of resources, both personnel and financial, in the research laboratories to perform service functions. Dr. Bale investigated many different avenues in her efforts to make these services available to the ichthyosis community. Because of political, financial, and other restrictions, it was clear that the NIH was not going to be able to do this. Large diagnostic companies were not interested because of the small market and the high complexity of the tests. After carefully examining the options, Dr. Bale and Dr. Compton decided to leave NIH and start their own company that will specialize in molecular testing for hereditary skin disorders and other "orphan" diseases for which no other laboratory offers the service.

Of relevance to the ichthyosis community, GeneDx, Inc. offers molecular diagnostic services for the congenital recessive ichthyoses (mutation testing in the TGM1 gene), epidermolytic hyperkeratosis, ichthyosis bullosa of Siemens, epidermolytic and non-epidermolytic palmar/plantar hyperkeratosis, erythrokeratodermia variabilis, Sjogren-Larsson syndrome, Darier disease, Hailey-Hailey disease, and Vohwinkel syndrome. The company also offers testing for several other, non-ichthyotic skin diseases, as well as for several other rare genetic disorders. Tests are done either on DNA from cheek swabs, or from small skin biopsies. Patients, dermatologists, or genetic counselors may obtain information about the cost of a test, shipping of samples, time to receive results, and genetic counseling services by calling 240-453-6285, sending e-mail to genedx@genedx.com, or checking the company's website at http://www.genedx.com.

Both Dr. Bale and Dr. Compton would like to thank the membership of F.I.R.S.T. for their strong support over the years, and hope that they may serve you even more in the future.



Grassroots Fund Raising

The weather is getting warm... the smell of spring is in the air... this is the perfect time of year to start planting a seed to conduct your own grassroots fund raising event. Grassroots fund raising is when you, the members of F.I.R.S.T., work in your local communities to hold your own fund raisers to help raise much-needed funding for the foundation. By conducting a small fund raising event, you can make a big difference. Some ideas for fund raisers are bowl-a-thons, car washes, garage sales, bake sales, golf tournaments, craft sales, and bike-a-thons! If you need help with ideas or are not sure how to go about hosting your own fund raiser, contact Jean at the F.I.R.S.T. office by calling 800.545.3286 and she would be happy to help you. Have fun and good luck!

Spotlight On...

Toni Roberts: A Story About Her Life with Ichthyosis

My name is Toni Marie Roberts, AKA Toni Marie Casale before I was married. I almost ended up being Tony Michael Casale, but that was before my parents found out I was a girl. For 15 years of my life however I was never called anything but "scales". Or, skin rot, carp, snake, etc. Any word describing a scaly, lizard-like creature would suffice.

I was born with ichthyosis. The term actually means fish skin, a point my seventh grade biology teacher felt was important enough to share with my entire class. This congenital skin disorder essentially makes my body produce more skin cells than can be shed, therefore leaving me with a scaly, shell-like exterior. It was discovered the day my mother took me to the doctor with a severe case of cradle cap; a case so bad it had extended down to my feet. The doctor patiently explained to my mother that I had ichthyosis. That was in 1969 and there was not much known about the disease. He only offered her at home treatments to alleviate the soreness of the dry skin. He also told her that I would probably suffer with it for the rest of my life.

My mother began a diligent campaign to "cure" me. She soaked my head in baby oil to loosen the skin so it could be combed out. She applied medicated lotion to the cracked skin until my screaming became intolerable. She fed me a diet high in fats and oils in the hopes that my body would somehow figure out what to do with the excess and use it to soften my skin. But as hard as she scrubbed and rubbed, I would wake up the next day with another scaly coating of skin.

I did manage to live a normal life. I had no idea that my skin was any different than anyone else's. I was content to ride my tricycle up and down the sidewalk for hours on end. I loved going to the park just like every other kid I knew. It wasn't until I turned 5 and entered kindergarten when it became apparent that I was out of the ordinary.

"What is that on your neck?" I was asked by one of the other 5-year-old girls. I didn't answer because I didn't know. It had never been an issue up until that time. Soon the school started receiving calls from angry parents shocked that the district would allow a child with a skin disorder to attend with all of the other "normal" children. The other kids refused to sit by me or touch me when we played tag during gym class. Eventually my classroom performance began to slip. I went from a child who started reading at 4 to one that would huddle up in a corner and throw temper tantrums whenever we had to play king of the mountain. Finally, my mother had to intervene on my behalf. She had a conference with my teacher and other faculty members to inform them of exactly what was wrong with her daughter. She told them that I was not contagious, that she did in fact bathe me on a regular basis despite the appearance of my skin. Eventually everyone calmed down enough to move on to more important things. But, the stigma I received stuck with me all the way through the remainder of my school years.

Soon it was time for junior high which meant more kids and more questions. But, now that I was becoming a young woman I was painfully concerned with other people's opinion of me, especially boys. When I was called a name in grade school I would simply ignore it or stick my tongue out at the offending person. But, now whenever my difference was pointed out to me I would regularly end up running to the bathroom to cry behind one of the locked stalls. I became obsessed with the appearance of my skin. I would slather myself in baby oil. I dressed in turtlenecks and long sleeve shirts even in the heat of the summer. I did not don a pair of shorts until I was in my early twenties. Mandatory swimming classes were the worst. While everyone was parading about in their latest Speedo swimwear I was hiding under the diving board praying for the class to be over.

Despite my complete lack of self-esteem, I did manage to develop a few good friendships during those years. Although my focus had always been on those who would not or could not accept me, there were a surprising number of kids who couldn't have cared less. Most of the people that I knew in junior high and high school are still good friends of mine.

Oh yes, high school. Lest we forget that all-important time in an adolescent's life. I think it can go without saying that it didn't get any easier for me. The kids got bigger...and meaner. Four more years were spent being an easy target for the masses.

Fortunately, by this point all of my diligence in skin care started to pay off. My skin was hardly noticeable from a distance, a point that served me well as long as I stayed at a distance. It was about this time that I discovered another facet to the disease. In gym class we were always required to run "The 12 minute" each quarter. A simple exercise where we would run around the gym for 12 minutes and see how many laps we could do. I was running it on one particularly warm spring day. Afterwards as I was walking down the hallway I started to feel dizzy. I began hyperventilating and then developed nausea. Soon everything went black. I found out later that I had passed out. My mother rushed me to the doctor to find out why.

"Have you ever seen her sweat?" The doctor asked her. Of course, she had never even thought to check. As it turns out, I

am unable to control my internal body temperature because sweat cannot break through the hard shell of skin that covers my body. Up until that day it had never been a problem because whenever I had become too warm I simply stopped what I was doing. No one had ever forced me to run around for 12 minutes before. On warm summer days I would become incredibly cranky and tired, but my mother had always assumed it was due to low blood sugar. From that point on I no longer had to run "The 12 minute". Instead, I got to hold the clipboard for the next three years for the gym teacher, which as you can imagine, made me even more popular. Now I was no longer the girl with the funny skin, but I was also lame.

As I grew older my intolerance to the heat grew worse. I had to visit the ER on several different occasions because I had underestimated the temperature or the time that I would be outside. I learned to adapt, even after I moved to Phoenix, a city known for its incredible temperatures. Walking out of an air conditioned building and to the car would be enough to start me on the long spiral downward into heat stroke. I developed a routine of dipping my feet into icy cold water to cool off and holding an ice pack between my eyes.

I'm now a married woman with a 5-year-old daughter who thankfully was not born with icthyosis. I've surrounded myself with people that I love and who love me back, and better yet, accept me for who I am. Long gone are the days when I was called "scales". I now wear shorts whenever I want regardless of the condition of my skin because it is simply more comfortable. I have even gone to the public swimming pool on occasion and lived to tell about it.

What I have learned from this disease is that being born with such an obvious distinction was actually a gift. It has given me an empathy and compassion for other people that I would just not have been able to develop on my own. I appreciate all I have in my life because I know that when I was growing up I never thought I would even have a boyfriend let alone marry someone who was loving and accepting of me.

As for the bitterness I used to have towards those kids in school who made a sport out of teasing me, that is long vanished. I want to thank them actually. Without their harsh words I would not be the determined and driven person I am today. I guess someone would say that I am a person who has a lot to prove. And, so far I have proven everyone wrong.

Visit our
new website at
www.scalyskin.org

Darier disease gene

continued from page 5

Perspectives

The identification of ATP2A2 as the defective gene in DD provides completely new insights into the role of calcium in maintaining skin integrity. This opens a new field in research for the understanding of the disease mechanisms and will lead to the developement of new pharmacological approaches. This finding has guided the search for the gene for another genetic skin disease (namely Hailey-Hailey disease), which turned out to be another calcium pump, thus emphasizing the key role of calcium in skin biology. Finally, several features of Darier disease remain to be understood. In particular, further studies are needed to provide insights into the late onset, focal cutaneous distribution, aggravation by external factors, variability of the disease and limitation of the clinical symptoms to the skin despite ubiquitous expression of this gene.

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Acknowledgements

We are grateful to the families, Anavaj Sakuntabhai, Sue Burge, Peter Bonis, Tom Strachan and Anthony Monaco for their participation in the project and to DEBRA UK for their support.

"In general, the goal in taking care of ichthyosis is to hydrate (moisturize) the skin, hold in the moisture, and keep scale thickness to a minimum."*

Foundation for Ichthyosis & Related Skin Types, http://www.scalyskin.org

Aquaphor® Healing Ointment helps heal dry skin associated with ichthyosis. Its unique petrolatum-based formulation combines a moist environment with the benefits of a semi-occlusive barrier that allows skin to breathe and absorb fluids.

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Consumer: Coupon good on the purchase of any Aquaphor® Product, no trial sizes. Limit one coupon per item purchased. Any other use constitutes fraud. Not good with any other special offer. Consumer must pay sales tax.
Retailer: You are authorized to act as our agent to redeem this coupon and we shall reimburse you at face value plus 8¢ handling in accordance with our redemption policy. Cash value 1/100¢. Copies available upon request. Offer void if copied and where prohibited, taxed or otherwise restricted. Mail to: Beiersdorf Inc, PO Box 880504, El Paso, TX 88588-0504. Good only in USA.

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Executive Director's Report

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

By now everyone should have received the appeal to renew your annual membership. A large part of F.I.R.S.T.'s financial strength comes from our members and I encourage everyone to renew your commitment to the foundation by sending in your donation. In the membership mailing, I inserted a bright green notice to alert everyone to the change of location for our family conference. I hope the last minute change of location has not inconvenienced you.

Plans for the conference are going very well. Hard to believe that it is less than six weeks away! Our ad hoc conference committee is working very hard to secure items for our gift bags, select entertainment and other details to ensure that this is one of the best conferences yet. I look forward to meeting our members and families this July. It will be hard to miss me...I'll be eight months pregnant with my second child (a baby girl).

For the first time in our history, F.I.R.S.T. will be hosting a Continuing Medical Education (CME) course at Children's Hospital in Philadelphia on July 7. Our CME course is directed toward pediatricians, internists, family practioners, geneticists, dermatologists and allied healthcare providers. The objective of the CME course is to educate physicians on how to identify the different types of ichthyosis, develop a diagnostic approach and understand how to manage and treat patients. A special note of thanks to Dr. Ho Jin Kim of Children's Hospital of Philadelphia for helping coordinate this program.

I am pleased to announce that four new members were elected to our Board of Directors. Elizabeth Gray, a parent of a child with lamellar ichthyosis, Glenn Oclassen, a corporate leader in the pharmaceutical industry, Dan Siegel, Esq., a Philadelphia attorney and parent of two young boys with X-linked ichthyosis, and Peyton Weary, M.D., a prominent physician in the world of dermatology and a parent of an affected child will add their expertise to advance the mission of the foundation.

Since the last newsletter, I had the opportunity to attend the American Academy of Dermatology conference in San Francisco. In addition to meeting many dermatologists and pharmaceutical representatives, I had the privilege of meeting most of our Medical Advisory Board (MAB). The MAB's annual meeting took place very early Saturday morning at the conference with overwhelming attendance. Three new members were nominated to the board and two members whose terms were expiring were renewed and will serve again.

Once again, F.I.R.S.T. was selected to testify before Congress about ichthyosis and increased funding for the NIH (see cover story). Our new board member, Beth Gray and her son Matt spoke on behalf of the foundation. One of our other members, Angela Godby and her husband, Carlos, took good care of the Gray's by showing them around capitol hill. Angela, who has lamellar ichthyosis, is the chief staffer to Representative Henry Bonilla (TX), a member of the Subcommittee on Labor, Health and Human Services.

Thanks to Beiersdorf, manufacturers of Eucerin and Aquaphor, F.I.R.S.T. has received funding to hire a part-time professional Program Director to manage the Ichthyosis Support Network (ISN). Beiersdorf has donated $10,000 to help fund the position and I am currently interviewing candidates to run the ISN. Look forward to meeting our new employee at the family conference and in the next newsletter.

On a final note, I am asking everyone to help F.I.R.S.T. by participating in a grassroots fund raising activity to promote awareness about ichthyosis in your community and help raise funds for the foundation. I have many great ideas and would be willing to help you, so please call me if you are interested.

Sincerely yours,

Jean

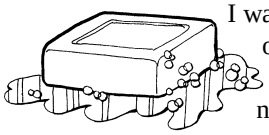
What's New… What's Hot

New Distributor for Theraplex®

Bioglan Pharmaceuticals, located in Wayne, PA, has taken over distribution of all Theraplex products. Bioglan is selling these products by the case only. To order call 1-888-246-4526 and identify yourself as a F.I.R.S.T. member and you will receive a discounted price.

New Soap Product

I want to share some great news about something that has worked for my family.



I was sweepstaking one day on the internet connected to www.soapmeister.com. I commented that I had two boys with ichthyosis and the people at soapmeister sent me some free samples of their soap. My son used this soap only five times and I would say his ichthyosis is 80% gone. I am so amazed and so thankful for gaining back some of his self-esteem. I wanted to tell anyone who has this ichthyosis about this product. I would also be interested in knowing if it works for others and what the long-term use of the soap can do. My thanks to www.soapmeister.com for what they have done for my son. You may contact me at michelleholmes@webtv.net or Cheri at www.soapmiester.com.

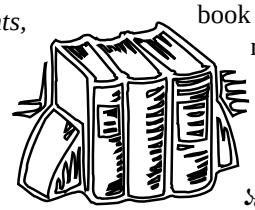
Computer Donation

A big thank you to Paul Meeker, a member of F.I.R.S.T. and father of a child with CIE, and his company, Computer Science Corporation, for donating two new-but-used computers to replace our older and slower ones. F.I.R.S.T. is very grateful for your generosity.

Check it Out!

Don't forget to check out our new website www.scalyskin.org. There are few sections that are still "under construction" but it contains a lot of valuable information and links to help individuals and families affected by ichthyosis. We continually update the site so stop in and take a look!

New Books Available



Raising A Child With a Neuromuscular Disorder: A Guide for Parents, Grandparents, Friends, & Professionals, by Charlotte Thompson, M.D. Dr. Thompson addresses all of the questions and difficulties that parents of children with neuromuscular diseases face from the initial diagnosis to adulthood. Available through Oxford University Press. ISBN 0-19-512843-5.

Raising a Handicapped Child: A Helpful Guide for Parents of the Physically Disabled, by Charlotte Thompson, M.D. A helpful guide for anyone who cares for or works with handicapped children. This book includes agency names and phone numbers, as well as a glossary of medical terms. Available through Oxford University Press. ISBN 0-19-513253-X.

Cooling Vest Available

A child's used cooling vest has been donated to the office and is available to a member of F.I.R.S.T. The vest is manufactured by Thinsulate 3M, Model 15 Ice Pack Vest P/N ST10V2637 Code ID# 74897. Please contact the national office at 800.545.3286. The vest is available on a first-come first-serve basis.

The Registry is here and at the Conference!

Hello! to everyone in FIRST!

Kim, Dr. Fleckman and I want wanted to thank all of you who have taken the time to enroll in the Registry, and I look forward to seeing everyone at the conference in July! I thought I'd offer a reminder that we will be screening people at the conference to help enroll people in the Registry, and want to encourage everyone who reads this to call us to see if you have enrolled. Remember, being a member of FIRST doesn't mean you are enrolled in the Registry!

Out of the 572 people who have started to enroll in the Registry, 387 people have finished, and another 107 are almost done (the others were not enrolled, usually because they did not have an appropriate diagnosis or they moved and we have lost track of them). We really need people to let us know when they move, and to call us if they aren't sure if they've enrolled or not. It's important!

To enroll, just sign a consent, have your dermatologist fill out a form, and talk with me on the phone! That's it! You can contact us for information and enrollment forms, and we can help with referring you to a dermatologist if needed.

When you have finished enrolling, we have a very complete picture of what your disorder is, how it has affected you personally, and we know if you are interested in having us forward to you information about any research projects you might be able to participate in. The bigger the Registry is, the more we can do to promote better understanding about these disorders, and that leads to better treatment!

Please help us make this next few years with the Registry a BOOMING success. Call us and see if you are enrolled. Call your friends or family who are affected and ask THEM to enroll in the Registry. We will send forms and information to ANYONE in the USA who is interested, so don't be shy - use us! And please be sure to mail back your annual update from the Registry with the consent signed!

Thanks again to everyone for your interest and support, and I hope to hear from you or see you in July!

Geoff Hamill, RN Registry Coordinator

Letter from the CFO

Dear F.I.R.S.T. members:

This past year was a very eventful one for F.I.R.S.T. The Foundation hired a new executive director and moved its offices to the Philadelphia suburb of Lansdale, Pennsylvania. In spite of the changeover in staffing, the Foundation closed out its 1999 fiscal year with a 7% increase in net assets over last year.

The Foundation continued to expand its services and sphere of influence in fiscal 1999. The creation of the Jane Bukaty Membership Assistance Fund represents an important addition to the Foundation. This endowed fund will directly benefit our members and we are grateful for the opportunity to administer this program. Also in fiscal 1999, F.I.R.S.T. committed to funding another $10,000 research grant through the Dermatology Foundation, providing additional research efforts into a cure for ichthyosis. We consider these two accomplishments among the major achievements of the Foundation in fiscal 1999.

Last year, I shared with you the new investment policy that was implemented in fiscal year 1999. Since the adoption of that investment policy, I am pleased to report that the Foundation has received a modest increase in investment returns. As we continue to grow, adherence to our investment policy has the potential to significantly improve the long-term health of the foundation.

As we move into the next century, we expect the Foundation to significantly extend its range of influence in every respect—number of members, services offered, support of research, stature and awareness in the community. The Foundation couldn't be positioned for this kind of growth without the continued support of our members, corporate donors and other friends. I hope you'll all continue to follow the Foundation as it advances, and look forward with me to another great year.

Sincerely,

Laura J. Phillips
CFO, Board of Directors

FOUNDATION FOR ICHTHYOSIS AND RELATED SKIN TYPES
STATEMENT OF FINANCIAL POSITION
SEPTEMBER 30, 1999
(WITH FINANCIAL INFORMATION FOR SEPTEMBER 30, 1998)

	1999	1998
ASSETS		
CURRENT ASSETS		
Cash and cash equivalents	$ 30,474	$101,260
Marketable securities	69,725	-
	100,199	101,260
CASH - PERMANENTLY RESTRICTED	10,000	-
FURNITURE AND EQUIPMENT	1,545	2,099
OTHER ASSETS	3,086	2,661
TOTAL ASSETS	$114,830	$106,020
LIABILITIES AND NET ASSETS		
CURRENT LIABILITIES		
Accounts payable	$ -	$ 1,107
Accrued payroll	2,047	-
TOTAL LIABILITIES	2,047	1,107
NET ASSETS		
UNRESTRICTED		
Operating	90,492	91,356
Board designated for research	10,718	9,807
	101,210	101,163
TEMPORARILY RESTRICTED	1,573	3,750
PERMANENTLY RESTRICTED	10,000	-
TOTAL NET ASSETS	112,783	104,913
TOTAL LIABILITIES AND NET ASSETS	$114,830	$106,020

This Statement of Financial Position is excerpted from the foundation's audited financial statements as of September 30, 1999. A complete copy of the audited financial statements and the independent auditors' report are available and can be obtained by calling the national office at 215.631.1411.

Program
2000 National Conference
F.I.R.S.T. Puts Me First

CROWN CORK & SEAL

Special Thanks to Crown Cork & Seal for its generous sponsorship of the National Family Conference

FRIDAY, JULY 7, 2000

Registration	4:00 - 6:00 p.m.		
Reception	6:30 - 9:30 p.m.	Light snacks, cash bar, time to mingle.	Sponsored by Bell Atlantic

SATURDAY, JULY 8, 2000

Breakfast	8:00 - 8:45 a.m.	Continental breakfast and late registration	
Children's Program	8:30 - 12:00 p.m.	Free child care available for children over 1 year of age, staffed by Kiddiecorp®	Sponsored by AmeriHealth Administrators and Health Data Management Corp.
General Session	8:45 - 10:30 a.m.	Opening Remarks	Michael Dunleavy, VP, Crown Cork & Seal
		What is Ichthyosis? How & who treats it?	Leonard Milstone, MD Mary Williams, MD
Breakouts	10:45 - 12:00 p.m.	A. CIE/Lamellar B. EHK C. Vulgaris/X-linked D. Forms of Ichthyosis with Secondary Symptoms	Leonard Milstone, MD Ho Jin Kim, MD Philip Fleckman, MD Mary Williams, MD
Lunch	12:15 - 2:00 p.m.	Symphony Ballroom	
Children's Program	1:45 - 5:00 p.m.	Free child care available for children over 1 year of age, staffed by Kiddiecorp®	Sponsored by AmeriHealth Administrators and Health Data Management Corp.
General Session	2:00 - 3:30 p.m.	Panel Discussion with Questions & Answers	Nannette Zale, MD, Opthalmologist Udayan Shah, MD, Otolaryngologist
Discussion Breakouts	3:45 - 5:15 p.m.	A. Mom's Discussion Group B. Dad's Discussion Group C. Women's Discussion Group D. Teenager's Discussion Group	
Open Session	4:00 - 6:00 p.m.	A. Ichthyosis Registry & Clinical Screening B. Ichthyosis Support Network C. Make-up Demonstration	Philip Fleckman, MD, Geoff Hamill, RN, Leonard Milstone, MD, Mary Williams, MD Lori Schreiber, MA Linda Graham, Kelly Jefferson
Dinner & Entertainment	6:30 - 9:30 p.m.	Symphony Ballroom	

SUNDAY, JULY 9, 2000

Breakfast	8:00 - 8:45 a.m.		
Children's Program	8:30 - 12:15 p.m.	Free child care available for children over 1 year of age, staffed by Kiddiecorp®	Sponsored by AmeriHealth Administrators and Health Data Management Corp.
General Session	9:00 - 9:30 a.m.	Focus on F.I.R.S.T.	Mary Williams, MD Jean Pickford, Executive Director Donna Rice, President
Breakouts	9:45 - 11:00 a.m.	A. Education discussion B. Tips for talking with your doctor C. Ask the Doctors (limited to affected adults only)	To be determined Geoff Hamill, RN Leonard Milstone, MD, Mary Williams, MD, Ho Jin Kim, MD, Philip Fleckman, MD
General Session	11:15 - 12:15 p.m.	Insights & Inspiration	To be determined
	12:15 p.m.	Closing Remarks	Michael Dunleavy

F.I.R.S.T. Puts Me First
2000 National Conference – July 7, 8 & 9
Philadelphia, PA

Registration Form

Name: _____

Address: _____

City: _____ State:_____ Zip:_____ Country: _____

Phone (day): _____ (evening): _____ Email: _____

Name of those attending	Adult	Child	Age	Check if affected with ichthyosis
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐
_____	☐	☐	_____	☐

Type of ichthyosis, if known: _____

Number of adults (18 & older) _____ x $95 per person = _____

Number of children: (1 through 17)* _____ x $50 per person = _____

TOTAL = _____

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

Name	Age (date of birth)
_____	_____
_____	_____
_____	_____

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

Name _____

PLEASE MARK ALL APPROPRIATE BOXES

☐ This is my first National Conference

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

☐ I/We will be staying at the Doubletree Hotel

☐ I/We are combining the National Conference with a family vacation

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

Please return registration fees with completed form to Jean Pickford, F.I.R.S.T., 650 N. Cannon Avenue, Suite 17, Lansdale, PA 19446. Kindly make checks payable to F.I.R.S.T. in US funds.

Cancellations will be honored with full refunds until Advance Registration Date of Wednesday, June 7, 2000.

2000 National Conference
July 7, 8 & 9

Hotel Accommodations

Please make a note...the conference location has changed since our last newsletter! Our new hotel facility is the Doubletree Hotel in downtown Philadelphia.

For hotel information and reservations, call the reservations department at 215.893.1600 or 1.800.222.TREE and identify that you are attending the Foundation for Ichthyosis & Related Skin Types, Inc. (F.I.R.S.T.) family conference. The room rates have remained the same at $80.00 per night, any occupancy. This rate does not include applicable taxes, which currently total 14%. Rates guaranteed from July 2 through July 9, 2000. Check-in time is 4:00 p.m. and check-out is 12:00 noon.

Individual attendees will be charged for one night's room and tax if they cancel within 48 hours of arrival; cancellations made prior to 48 hours will not be penalized.

All rooms must be reserved by June 8, 2000! Rooms not reserved by that time will be accepted on a space and rate availability basis.

See the Sites

With the F.I.R.S.T. family conference falling between Independence Day and the Republican National Convention, Philadelphia will be in perfect shape for sight seeing! The city is filled with fun activities to entertain the whole family.

Kids will love The Philadelphia Zoo, Franklin Institute and Please Touch Museum which are all fun and educational. Experience the historical significance of the city by visiting The Philadelphia Museum of Art, Independence Hall, and the Liberty Bell Pavilion. For anyone who loves to shop, The Gallery at Market East is one of the nation's largest shopping centers and is conveniently located near the Doubletree Hotel. The Philadelphia Phillies will be playing home games that weekend, so it is a great opportunity for the baseball fan to catch a game.

If you are looking for a place to eat, you will have no problem choosing from one of Philadelphia's numerous restaurants. From the famous Pat's or Geno's Steaks to the lesser known restaurants and cafés, a variety of culinary creations can be found throughout the city. And don't forget, Atlantic City and The New Jersey State Aquarium are only a hop, skip, and jump away!

For a free Philadelphia getaway planner and a value book filled with tons of savings, call the Philadelphia Visitor's Bureau at 1.877.334.2238. The Visitor's Bureau can also be accessed through the Internet at www.pcvb.org.

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 July 1, 2000 to July 15, 2000:

* 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 (877) 874-7687 and refer to Gold File Number 74671373. There are also discounts on First Class and Business Class fares. Certain rules and restrictions apply. Call US Airways from 8:00 am to 9:30 pm EST for details.

RESERVE EARLY, AT LEAST 60 DAYS PRIOR TO DEPARTURE DATE, TO RECEIVE THE MAXIMUM DISCOUNT.

Airport Shuttle

Tropiano, 1-800-559-2040, will provide transportation between Philadelphia International Airport and the Doubletree Hotel for a discounted rate of $15.00 per adult, 1/2 price for children age 10 and under, and free for children age 2 and under, each way.

Advanced reservations are recommended, but not required. Upon arrival at the airport, travelers can also make arrangements from the "Ground Transportation Center" in the baggage claim area (Dial #19 on the white phones at the counter). Indicate that you are participating in the F.I.R.S.T. family conference.

Here's an idea:
Why not combine the F.I.R.S.T. conference with a family vacation and celebrate our country's Independence Day, July 4, in historic Philadelphia!

Gene Therapy Correction

In the last issue of the Focus, we reprinted a statement from NORD (National Organization of Rare Diseases) about gene therapy. One part of that statement was misleading and another part contained factual errors.

First, it is important to appreciate that there are many ways of administering gene therapy. No two gene therapy trials are the same, either in the goals they try to achieve or in the methods they use. It is particularly important to understand that not all forms of gene therapy utilize viruses or parts of viruses to deliver new genes to target tissues. Clearly, then, any concern about viral vectors does not apply to the many protocols that do not employ such vectors. It is misleading to raise concerns about gene therapy, in general, in the context of raising concerns about the viral vectors used for certain forms of gene therapy.

Gene therapy still holds great promise for many diseases.

Second, it is not correct to say that "we know that viruses...used to insert new genes into cells can form new, potentially dangerous viruses that can escape into the environment." While such an event may be theoretically possible, the probability for it happening is extremely low. There is not a single example of a gene therapy vector turning into a dangerous virus and escaping into the environment. Viruses have two properties: they can infect living cells, and once inside a living cell they can reproduce themselves. The "vectors" used in gene therapy protocols are designed to get new genes into cells, but these vectors have been carefully modified and tested so that they are completely unable to reproduce themselves. When a vector gets into a cell, the cell becomes its final stop because it cannot reproduce itself.

Gene therapy still holds great promise for many diseases. We may be disappointed with the pace of progress or disturbed by human errors of omission or commission. There are certainly risks to gene therapy but, to date, there is no evidence that gene therapy of any kind poses risk to anyone except the recipient of the therapy. Most experts believe that the risks of gene therapy are inherently no different than the risks of more conventional medicinal treatments.

News & Notes

Dr. Weary Receives Award

Peyton E. Weary, M.D., a member of F.I.R.S.T.'s Board of Directors, was honored with the Janssen Pharmaceutica's Masters in Dermatology at the American Academy of Dermatology conference this past March. Dr. Weary, the only dermatologist ever to serve as president of the American Board of Medical Specialties (ABMS), has championed the field of dermatology in Washington, testifying before countless committees and agencies. His efforts have paid off in greatly increased funding for dermatology research and greater visibility and respect for dermatology among all medical specialties. Dr. Weary has the greatest respect for patient advocate groups, in part, because of his family's personal experience. His first daughter was born with Epidermolytic Hyperkeratosis (EHK) and his fourth child, a son, died in infancy from the same disease.

Alliance Name Change

The Alliance for Genetic Support Groups has recently changed its name to the Genetic Alliance. The Alliance is a non-profit coalition of support groups, consumers and professionals dedicated to promoting the common interests of children, adults and families with, or at risk for, genetic disorders. The Alliance specializes in linking people interested in genetic conditions with organizations that can provide support and information. The Alliance is still the very same organization with the same mission and services, but with a new name. Their website address is http://www.geneticalliance.org.

Please help us to minimize our printing costs. Call us at 1-800-545-3286 if you do not wish to receive any future issues of *Ichthyosis Focus.*

IRS Grants Deduction for Illness Conferences

(Times Staff and Wire, Thursday, May 11, 2000; http://www.hotcocoa.com/health/stories/irs_20000511.htm)

Parents with chronically ill children may now deduct many of the costs related to attending medical conferences and meetings, the Internal Revenue Service ruled this week. Community groups pressed for the change, saying families with ongoing medical expenses had trouble paying for the national meetings that help them stay on top of research and treatments that could help their children.

Rep. George Miller, D-Martinez helped shepherd the new rule through the IRS regulatory process. "Today's ruling means that parents who must often expend a great deal of money to learn about innovative treatment for their children will be able to deduct some of the costs of attending these conferences," Miller said in statement. "Virtually everyone else attending meetings can deduct the cost –physicians, vendors, salespeople. I thought the parents of the patient should get a little help too, and I am gratified the IRS agreed."

The rule is explained in Internal Revenue Bulletin 2000-19, released May 8.

Credit Cards

F.I.R.S.T. is now accepting credit cards. The national office is equipped to accept Mastercard, Visa and American Express. Please consider this form of payment the next time you make a donation or purchase materials.

F.I.R.S.T.
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Lansdale, PA 19446

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