



Incontinentia Pigmenti International Foundation

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Volume 9 2004

LETTER FROM THE EXECUTIVE DIRECTOR

In keeping with my previous assumptions that those who read this newsletter are most interested in learning the latest news in the field of research of IP, I have asked David Nelson, Ph.D., of Baylor College of Medicine (a leading researcher in the IP International Research Consortium) to write an article on the current work being done in research, now that the gene has been identified. His laboratory assistant at the time the Nemo gene was identified has left the laboratory and Dr. Christine Shaw has taken his place. Dr. Nelson has written a short biography of Dr. Shaw which I think you will find interesting.

I also have requested Ashley Badgwell, MS to do an update on the Natural History Project that was started in 2003. Ms. Badgwell, who conducted the survey and compiled the results, has now done an update. This is a project which will continue for many years to come and we will report updates on current findings periodically in this newsletter. I was delighted that IPiF was the recipient of some very imaginative fundraising projects. I've asked those whose events these were to write stories about them. It is hard to describe how very grateful IPiF is for the time and effort that went into making sure that these undertakings were successful.

IP NATURAL HISTORY UPDATE

Ashley Badgwell, MS and

Judith Willner, MD

Mount Sinai School of Medicine

Last spring, a summary of the

Incontinentia Pigmenti Natural History

Study was published in this newsletter.

For those of you unfamiliar with the

project, Dr. Willner, Director of Clinical

Genetics at Mount Sinai School of

Medicine and I, then a graduate student

at Mount Sinai in New York City, saw

the need for an IP natural history

analysis based on physician and patient

reports. A questionnaire was developed

to assess the affected individual's

experience with IP. It was sent to IPiF

members, with the help of Susanne

Emmerlich and made available on the

website. As of March 2003, 152

completed surveys were returned and

included in our initial analysis. Since

that time, we have received 42 additional

completed surveys, bringing the total

number of participants to 194. Again, we

are very grateful to those who

participated. In this article, we present an

update of the data along with a

discussion of interesting findings.

First, a review of the statistical tools

employed in the analysis may help

readers to better understand the data and

appreciate the importance of sample size.

We understand that the 194 people

included in this study represent only a

fraction of the worldwide population

with IP. Of course, as the number of

participants increase, our understanding

of IP becomes more accurate. Because of

the nature of our data, we decided to use

95% "confidence intervals" to determine

the statistical significance of the

findings. The definition of confidence

interval is "the range within which the

true magnitude of effect lies within a

certain degree of assurance". In this

study, you will find the confidence

interval as two numbers in parentheses,

following a frequency or average. For

example, when you read that 49%

(45%,53%) of the participants in this

study have missshapen nails, that means

that we expect that the true percentage

(Continued on page 4)

CONTENTS

1.	Natural History Project update.	Page
1.	Research Update	
2.	Dr. Christine Shaw	
2.	Krispy Kreame Donut Sale	
2.	Holiday cheer With a Purpose	
2.	Nick & Toni's Celebration	
3.	To Find a Good Doctor.	
3.	How to Get Treated Like a Doctor	
6.	CNS Table	
7.	Photographs	
8.	Need for Funding	
8.	Membership Form	

Finally, new families with unusual mutations in various tissues.

mutations, in addition to those with the

common deletion mutation, continue to

come to the attention of researchers.

These mutations help to define the parts

of the NEMO protein that are important

for function. In addition, more mutations

that lead to the ectodermal dysplasia and

immune deficiency disorder have been

defined. Again, these help researchers to

understand the role of different parts of

(Continued on page 2)

afflicted with this disorder. One challenge we are constantly faced with is the ramifications of IP as she decides to have her own children. The only way we feel we can support her in this situation now is to help raise money that may support future research and advanced prenatal testing possibilities.

Every year we have a group of twelve couples that gets together for a holiday progressive dinner party. As with many holiday gatherings, many of the couples bring host and hostess gifts to the party. Two years ago we decided, as a group, to use the money we would have spent on those types of gifts and donate it to a charitable organization of the host and hostess' choice. This year the dinner portion of the evening was held at our home, therefore we chose IPFF as the charitable organization for the evening.

Using much of the information from the newsletter, I created a form explaining IPFF and the need for funding. I sent a form to each of our friends attending the evening noting that IPFF was to charitable organization for the evening. Much to our surprise, most of the couples that attended the event donated a generous amount to IPFF. In addition, everyone had a wonderful time enjoying friendship and the holiday season.

We cannot thank our dear friends enough. We know that this donation to IPFF is just the tip of the iceberg for what is really needed to continue further research. But, it did encourage us, and hopefully some of you, to take on small fundraising efforts to support our children and their lives with IP.

NICK & TONI'S CELEBRATION

Nick & Toni's is one of the most popular restaurants on the East end of Long Island in New York State in an area known as "The Hamptons". All summer it is overflowing with socialites, movie stars, and business tycoons as well as ordinary folks. As you may have read in previous newsletters over the years, the owners have very generously given one night a year to raising funds for IPFF. This year the restaurant celebrated it's 15th anniversary with a big party, open to all, as a fund raiser benefiting 3 charities, one of which was IPFF. They charged \$100 to enter where one was then able to have drinks, food, dance to a great band, and best of all, children were particularly welcome and were encouraged to join in the dancing. An amazing 600 people showed up. Articles were written about it in several newspapers and it was a rousing success for all.

How did I come up with my idea? It was really easy. I have been wishing for some time that I could contribute in a more substantial way towards finding a cure for IP. I have long thought that this would be the most substantial gift that I could give to my oldest daughter, who was born with IP. She has many years ahead of her until she is of childbearing age; yet, inevitably that day will be here and I very much hope that a cure has been achieved by then. My husband and I make our most substantial charity donation to IPFF, yet I very much wanted to do more than we alone could afford.

I went to Krispy Kreme one day with my daughters (a typical cold rainy Seattle day) and we needed indoor amusement to watch the making of the donuts. I saw flyers on a table explaining how to raise funds for non-profit organizations and took one. I thought about it for a long time instead of acting: the initial outlay of money had to be mine, and finances were a bit tight; I was worried about being stuck with \$250 worth of Krispy Kreme cards. However, donuts are really hot commodities! I purchased 20 Krispy Kreme cards at \$10 each, a net profit of \$10.00 per card. Each card allows the purchaser to get a free dozen donuts for each purchased, up to a maximum of 20 times. At \$6 a dozen, that means the purchaser gets \$120 of donuts for \$20. This is a great deal for people who often purchase snacks for work, clients, neighbors, preschools, scouting trips, etc... Krispy Kreme has less expensive options as well.

I planned to go door to door through my neighborhood but had no need as all my friends bought all 25 cards, and felt like they got a great deal. I plan to buy more Krispy Kreme cards and go door to door, and also give them as Christmas gifts. There are also restaurants who advertise having "benefit nights" and "donate" a certain percentage of proceeds to a given charity; I am thinking of trying one of those next. In the meantime, I hope to send IPFF more money soon!

HOLIDAY CHEER WITH A PURPOSE
Amy Stuurma
Grand Rapids, Michigan

We have a daughter, Hannah, who is four years old with IP. She was diagnosed three weeks after birth by skin biopsy, and later by genetic testing. She is doing very well and lives a very active and healthy life. We are very thankful for that, but continually feel like there is not much we can do to help others

(continued from page 1)

Research Update

the protein. The origin of the common mutation, and of the unusual structure of the NEMO gene and pseudogene continue to be investigated. This structure can be found in chimpanzees and gorillas, suggesting an ancient origin. Of interest is the role of this structure, and of variation in the structure in the human population, in generating the common deletion mutation found in some 80% of patients with IP.

This remains an exciting time in IP research, with advances being made on several fronts. Each of these areas of basic research can be expected to lead to improved diagnosis, better understanding of the normal role of NEMO, and possibly to improved treatment and prevention.

DR. CHRISTINE SHAW
Baylor College of Medicine

Dr. Shaw is currently in the process of identifying novel mutations in the NEMO gene in patients with Incontinentia Pigmenti (IP). Additionally, she is studying the expression pattern of the Nemo protein in various tissues throughout development in the mouse.

Dr. Shaw is a graduate of the University of Wisconsin, where she received a B.S. in Genetics in 2000. She attended graduate school at Baylor College of Medicine in the department of Molecular and Human Genetics. There, her thesis work in the laboratory of Dr. James Lupski involved study of the mechanisms of genomic rearrangements involving proximal chromosome 17p, including the Charcot-Marie-Tooth disease type 1A and Smith-Magenis syndrome regions. She had an illustrious career as a graduate student, contributing to over one dozen peer reviewed publications, including one on Incontinentia Pigmenti that resulted from her short rotation in the Nelson laboratory in her first year. She received her doctorate in April of 2004 and began a postdoctoral position with Dr. David Nelson shortly thereafter.

KRISPY KREME DONUT SALE
Paige Ryan
Seattle, Washington

I forwarded a check to PPF in the amount of \$250, the profits I realized from my sales of Krispy Kreme donuts. I intend to try and do another fundraising event in the near future for IPFF. I realize that \$250 is not very much money, but I wanted to start small as I have no experience in fundraising. Actually, I found it fun and easy and am looking