

THE USA IP SUPPORT GROUP

Anne K. Ryan

Please allow me to introduce myself and invite you to join my newly founded USA IP Support Group based out of upstate New York.

First, let me tell you about me and why I had to take on this labor of love. My name is Anne Ryan and simply stated, I am a mom. I have a wonderful husband, a beautiful 2-year-old daughter Willow, and a blessing of another daughter, 9 month old, Kateri. Kateri was born in August, 2002. She was diagnosed with IP within one week of birth. I can still feel the fear and hear the words as the doctor read me pages off the Internet on what *incontinentia pigmenti* was. I have never in my life felt so alone. There was no one to sympathize with the grief my entire family was feeling and I knew that someday I had to do something to help.

My background before being a full time mom was in the medical field as an Office Manager/Lung Transplant Coordinator for a pulmonary practice. My husband works two, yes two, full-time jobs to enable me to be home with the girls. He is a Youth Division Aide at a Maximum Secure Facility for Juveniles, and a Personal Fitness Trainer. We are short on time with one another, short on money, but rich with love. Our children are our life, and outside of them, nothing else matters to us. I am also a professional singer and actress, and hope to be cutting a demo CD this summer.



Anne K. Ryan and Kateri (continued on page 2 col. 1)

INCONTINENTIA PIGMENTI: A NATURAL HISTORY STUDY COMPILED FROM PATIENT REPORTS

Ashley Badgwell, MS and Judith P. Willner, MD

For the past year, a research project to better document the natural history (symptoms and clinical course) of IP has been underway at the Mount Sinai School of Medicine. Because the types of symptoms and their severity vary greatly among affected individuals, even within the same family, the prognosis is difficult to predict. The last large-scale clinical study of IP was in 1976. As familiarly with this disorder has increased among physicians, milder cases of IP have been described. Based on the cases seen in our practice, we suspected that the more severe cases might be over-represented in the literature. We felt a revised natural history of IP was called for. This is particularly timely since, with the recent DNA-based testing allows confirmation of diagnosis in milder cases.

Dr. Judith Willner, Director of Clinical Genetics at Mt. Sinai, and I, then a graduate student in Genetic Counseling, proposed to compile a natural history of IP based on patient and physician reports. We developed a six-page questionnaire, consisting of questions regarding patients' family histories and medical histories relevant to IP. It was translated into four languages. With the assistance of Susanne Emmerich of IPiF, it was sent to all members of IPiF and posted on the IPiF website. We had received 152 completed surveys by March when the initial analysis was performed. Completed questionnaires have continued to arrive, and we plan to update the analysis continually. We are very grateful to all those who participated.

Because the report on our findings is almost 30 pages long, it is not possible (continued on page 2, col. 2)

LETTER FROM THE EXECUTIVE DIRECTOR

In each of the previous newsletters I have made an effort to include articles on subjects that are likely to be among the most important issues facing people with IP. Presently one of the most eagerly awaited is the result of the questionnaire that was sent out on the natural history of IP. This tried to determine the frequency of specific symptoms experienced by those with IP. A subject that is always uppermost in the minds of women with IP who are contemplating having children, and of women who are wondering what the consequences will be for the children of their daughters with IP.

Although you will read that 700 questionnaires were mailed out, please keep in mind that many went to families with several members with IP, and to physicians who have many patients. The questionnaire is also posted on the web site and was read and filled out by many more individuals (exactly how many we have no way of knowing). Therefore the number 700 really represents many more people. As the identification of patients was coded, we don't know how many people actually (continued on page 2 col. 1)

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