ipif
Incontinentia Pigmenti International Foundation

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LETTER FROM THE EXECUTIVE DIRECTOR

We can look back on the past seven years of **IPIF**'s growth and accomplishments with enormous pride. In some years achievements have been a great deal more spectacular than in others, but no year goes by without progress. The year 2001 saw the foundation gain new members from all parts of the world and has become better known to health providers in all the medical specialties that are potentially affected by IP. Hopefully this results in fewer cases of misdiagnoses and more recognition of IP's various symptoms. Research is constantly making headway, although in some years less spectacularly than in others. 2001 was, however, very difficult for fundraising, an experience many charities shared as a result of the terrorist attacks on September 11 in New York City where the foundation is based. IPIF suffered a large drop in revenue, which we will try to make up in 2002.

In each newsletter I try to address the concerns that people, who have contacted the foundation over the past year, have expressed to me. One of the major questions has been about statistics. As IP has been unrecognized for so long by so many physicians, accurate numbers are hard to come by. A project I have tried to initiate for some time is finally being undertaken. The story on page 1
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IP NATURAL HISTORY PROJECT

Ashley Badgewell
Genetic Counseling
Mount Sinai Medical School

Dear IP families and IPIF supporters,

I would like to introduce myself and present the upcoming IP natural history project. I am a first-year genetic counseling student at Mount Sinai Medical School in New York City. For those of you unfamiliar with the profession, genetic counselors help parents and patients at risk for genetic disease to organize and understand genetic tests, diagnoses and disease management. Before entering graduate school, I completed my undergraduate work at the University of Texas at Austin. Then, I spent two years as a technologist in a diagnostic
cytogenetic lab at Stanford University Hospital in California. I am interested in the way parents of affected children encourage scientific research. The role of scientists and doctors is crucial but no one has a greater interest in eradicating a disease and alleviating its symptoms more than parents of affected children have. I have heard of several situations where dedicated family members have accelerated research by pressuring scientists, raising money, sharing information and spreading awareness.

As you probably know, the manifestations of IP are variable and there is a great range in severity of symptoms from patient to patient. This can cause a new diagnosis of IP to be extremely traumatic because parents have little idea what their child's experience with IP will be. With the help of Dr. Judith Willner, Director of Clinical Genetics at Mount Sinai and Susanne Emmerich, Executive Director of IPIF, I will be initiating a study to compile a natural history of IP. In other words, we would like to find out which traits many IP patients' display which have been
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THE IMPORTANCE OF RARE DISEASE RESEARCH

People with rare disorders know how important these disorders are because an "orphan disease" can profoundly impact one's life and one's family. Scientists who study these diseases suggest patients can offer unique knowledge that will inevitably enhance the understanding of more prevalent health conditions. However, it seems that politicians of all nations ignore the importance of rare diseases because public health policy, and the money that governments spend on public health, is usually aimed at "major" diseases that affect large numbers of people. Unfortunately, the neglect of rare health threats is a consequence of minimal resources and lack of political and scientific interest.

It is up to us, individuals with rare diseases and patient organizations; to remind politicians, scientists, and the public that orphan diseases are not "minor" health threats. They demand attention; they need resources; and they deserve recognition of the pain and disability that they cause. It is wrong for any society to deny resources to a minority, and minorities should speak up and make their needs known.
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Spanish Support Group Meeting in Barcelona