nipf
National Incontinentia Pigmenti Foundation

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LETTER FROM THE EXECUTIVE DIRECTOR

I am pleased to present the second issue of the NIPF newsletter. Much has occurred in the past year. Please note that the *Foundation has moved* to new offices,. The address, telephone and fax numbers are listed above. The post office has been requested to forward the mail, but they have been slow to do so, therefore if your letters were slow in being responded to, I apologize.

One of the proudest achievements of NIPF is the extent of its outreach, through various means: internet, genetic conferences, membership affiliations, internet, etc. NIPF is now in contact with physicians, IP families, etc. in eighteen countries. This has been an amazing accomplishment in such a short period of time. As a result, membership has increased to include over 450 families and individuals.

This issue will introduce the first in a series of articles dealing with the very complex issues faced by those with a genetic disorder or are the parents or siblings. I will not attempt to solve these very complex feelings, but I will attempt to identify them so those affected realize they are not alone in experiencing these emotions. In the first article we will attempt to deal with the feelings of a mother having given birth to a baby with a disorder.

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INTERNATIONAL RESEARCH COLLABORATION

In the last newsletter it was reported that "*the first international research meeting*" would take place on September 27 in New York City. This meeting was canceled and the following meeting took place instead. Three laboratories joined forces on December 5th and 6th, 1996, for the purpose of accelerating the search for the defective gene(s) which cause IP. This international symposium was the first of its kind for Incontinentia Pigmenti (IP). Once the gene is isolated, the next step will be to attempt to eradicate the symptoms which are caused by this genetic disorder. At the meeting, each of the participating laboratories presented case histories, accompanied by slides and diagrams, which were followed by a discussion in which all participated.

The importance and rarity of such a collaboration cannot be emphasized strongly enough, It is the wish fulfillment of all foundations, such as NIPF, to isolate and subsequently resolve the problems caused by a genetic disorder. It is also well known that funding is extremely difficult to come by. None of the laboratories involved receives funding specifically for IP. The bench work is done in the laboratories alongside many other projects. Meetings, such as the one described above, must be funded by the NIPF. The expenses include airfares to and from the meeting site (from various parts of the world), hotel accommodations, meals, and many other costs. Expenses are kept to a minimum, but they exist.

It is also possible that the foundation may eventually be required to pay the salary of a Ph.D. graduate who would do the bench work in a laboratory, under the direction of one of the leaders of the collaboration. This would require approximately $30,000 per year.

If you can assist in this effort, by making a contribution, it would be greatly appreciated

THE VALUE OF HUMAN FETAL SPECIMENS IN IP RESEARCH
by Richard A. Lewis, M.D.

In our current state of medical knowledge, little is known about how the gene(s) for Incontinentia Pigmenti of the classical type causes the changes in development of a female fetus that lead to the distinctive skin rash, altered teeth, altered migration of nerve tissues in the brain, and the malformed blood vessels in the retina that become proliferating tissues and retinal detachment. Similarly, of course, nothing is known about how or why the product of this gene is invariably lethal in a developing male fetus. Eventually, when any gene for Incontinentia Pigmenti is cloned, research geneticists, cell biologists, and physicians will want to answer these questions. In what tissues does the protein product of this gene act: Why, if the gene is present in every cell in the body is it "turned on" only in selected tissues? What causes the variable severity of the disease in different members of the same family? Why do some families seem to express the disease more severely as it passes from one generation to the next?

To assist the answers to these and other questions, we must begin to salvage the products of conception that are lost or discarded during a spontaneous or selective termination of pregnancy. These specimens offer an unique opportunity to study both normal and abnormal human