A letter to our Leukodystrophy Families

From: Professor Alfried Kohlschuetter

Would you have liked to have known earlier?

"Newborn screening" is only looking for well-treatable conditions

Dear Families:

Have you ever asked yourself: Why it took so long to make the diagnosis of Leukodystrophy in your family? And what would it have been like if we had known everything much earlier?

Leukodystrophies are metabolic disorders of the nervous system. They are presently incurable, although in many cases their progression can be stopped and relief is most frequently possible.

Medicine knows countless other congenital metabolic diseases. They can be divided into three groups.

(1) For a small group of diseases, efficient therapies have been developed. When the diagnosis is made early, the development of severe symptoms can be completely prevented. For such diseases *newborn screening* has been instituted for more than 40 years. A drop of blood is taken from the newborn's heel, put on a strip of filter paper and sent to a laboratory. If an easily treatable metabolic disorder is recognized in the blood, all measures can be taken to protect the child from damage. An example of such disorders is phenylketonuria (PKU), which is treated with a special diet.

(2) For the second group of diseases, therapies are also available, but their efficiency is not so convincing as in the first group. An example is Adrenoleukodystrophy (ALD). Such diseases are not included in the newborn screening programmes.

(3) A third group consists of genetic diseases for which there is presently no effective treatment. This is true for many Leukodystrophies. The lack of any treatment prevents such diseases from being considered in the newborn screening programme.

Nevertheless, an increasing number of voices presently emphasise other advantages of the early recognition of such diseases: the way to diagnosis would not be that long and full of twists and turns, the chances would become greater should new treatments become available, the risk of having further affected children would be recognized in time for choices to be made and research into rare diseases in general would be boosted.

Newborn screening is complicated, expensive and strictly regulated by law. I have been responsible for the programme in a part of Germany for many years. I am presently going through a new experience listening to discussions about the inclusion into the newborn screening of diseases that can be treated only with restricted efficiency. Opinions vary between extremes. "If you cannot do anything, I would like to enjoy the time with my child before the onset of a severe disease, without thinking of the future." On the contrary, people, mainly those who have closely experienced severe suffering, frequently say: "Had we known earlier, many things would have been easier for us."

I believe this discussion should be carried further into the society and I have been discussing this vital concern with Bob Wyborn from the Leukodystrophy Resource & Research Organisation in Australia. We both agree that the voice of people like yourselves whose opinion is based on personal experience, is of critical importance. We are eager to be able to talk with you about these questions shortly and I encourage you to send your comments to either or both of us please.

Please include your preferred method of return contact to either of the following;

Alfried Kohlschütter, M.D.
Professor of Paediatrics
Department of Paediatrics
Clinic for Degenerative Brain Diseases
University Medical Centre UKE, Martini St. 52,
20246 Hamburg, Germany
kohlschuetter@uke.uni-hamburg.de

Bob Wyborn
President
Leukodystrophy Resource & Research
Organisation Inc.
PO Box 209
Clontarf Beach
Queensland 4019
Australia
Email: bobwyborn@bigpond.com
www.leukodystrophyresourceresearch.org