

From: Manuela Martinez, M.D. <[REDACTED]>
To: Dietrich Matern, MD <[REDACTED]>
Cc: Brian Wulf <[REDACTED]>
Date: Saturday, July 24, 1999 5:57 AM
Subject: Re: Warning: could not send message for past 5 days

Dea Dr. Matern,

In the particular case of Matthew, I also prefer to say that he has a generalized peroxisomal disorder, without further specifications. There are certain things in Matthew that do not agree with the IRD phenotype. For example, the levels of the very long chain fatty acid (VLCFA) 26:1 were initially higher than those of 26:0. This admittedly happens in Zellweger's syndrome. In NALD, 26:0 and 26:1 are about equal. In IRD, 26:1 is much lower than 26:0 while phytanic acid is higher than in the other two phenotypes. In Matthew, phytanic acid was only mildly increased. Again, these are only biochemical approximations, and I think that all signs and symptoms can appear mixed-up in every peroxisomal patient.

My personal contribution to the peroxisomal field has been that related to docosahexaenoic acid (DHA). In 1987, I found that a patient with the Zellweger syndrome had extremely low levels of DHA in the brain and other tissues. Later on, I could confirm this finding in many other patients with generalized peroxisomal disorders. DHA is believed to play a fundamental role in the brain and retina and, being deficient in Zellweger's syndrome, this could be related to the pathogenesis of the disease. So a second part of the research started in 1991, when I

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