

*Center for Research in Biochemistry and Molecular Biology
Laboratory of Lipids and Human Brain Development*

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TO WHOM IT MAY CONCERN:

When Matthew Wulf came to us in April, 1998, diagnosed with infantile Refsum's disease, he had a typical deficiency of docosahexaenoic acid (DHA). However, his DHA levels were not so low as in other patients because he had been taking a DHA preparation (Neuramins) for a month. At the end of April, Matthew had steatorrhea (++++), very altered liver tests and failure to thrive. He was hypotonic and his developmental milestones were delayed. He was deaf and his vision was limited to strong light. Treatment with DHA ethyl ester was immediately initiated (300 mg/day). His diet was progressively changed to a complete, normal diet for the age, rich in fish and meat. In about 3 weeks, Matthew's steatorrhea virtually disappeared (+), transaminases fall abruptly and he gained weight (960 g).

After a year, Matthew has made further improvement. In special his vision is much better and he can now track objects. Hearing is very poor but it improves a great deal with the hearing aids. Matthew is very active and playful. His muscle tone is now clearly better, although he still has a slight degree of axial hypotonia. Matthew sits unsupported and stands up with help. He has hepatomegaly and his liver tests are slightly increased. On MRI, he has an estimated age of about 12 months.


I think it is very important that a neuropediatrician sees Matthew regularly. He is having a low dose of vitamin K (7.5 mg/weeks). In my opinion he should take more, a minimum of 2.5 mg/day. He needs a high dose of vitamin E (about 200 mg/day), as an antioxidant. Also, Matthew probably needs some vitamin D supplement, since his bone development is delayed. It would be nice to know Matthew's levels of liposoluble vitamins before increasing the dosages. Peroxisomal patients usually have low levels of those vitamins.

Matthew may benefit a great deal from multiple stimulation, physical as well as visual and auditory. It is important to strengthen his muscles as much as possible so that he can walk. The periodical control of an orthopedist and/or a physiotherapist is very important. Few of these patients can walk or speak or but I hope that Matthew can do both things if we help him as much as possible.

DHA therapy must be followed, for the moment, indefinitely. I would appreciate that you keep in contact with me referring Matthew's evolution. He will come here twice a year but, as your local doctors, you are most important.

Please, do not hesitate to contact me on the phone, by e-mail or fax. My e-address is: <mmartinez@mail.cinet.es. My phone: 34-93-4894065. My fax: 34-93-4894064.

Sincerely,



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