Oregon Health Sciences University
Consultative Report

CHILD DEVELOPMENT AND REHABILITATION CENTER

P.O. Box 574, Portland OR 97207-0574

Account No.
Medical Record No. 01-38-94-88
Name Wulf, Matthew A
Birthdate 12/03/1997

CDRC

CLINIC DATE: 03/30/98

CLINIC NAME: METABOLIC CLINIC

DISCIPLINE: [redacted] PEDIATRICS

We saw Matthew Wulf in the Metabolic Clinic at Oregon Health Sciences University on March 30, 1998. We had just seen Matthew on March 9 for initial counseling. Matthew has infantile Refsum's disease, a peroxisomal disorder characterized by blindness, deafness, developmental delay and hypotonia, all to varying degrees. Matthew's brother, Adam, has the same condition. We brought the family back today since they were going to be seen in Gastrointestinal Clinic anyway to give them follow-up on the laboratory tests that we did last time and follow-up on our progress in attempting to come up with a therapeutic plan for Matthew.

I showed Matthew's parents his essential fatty acid results. Briefly, he is deficient in DHA and arachidonic acid. He also had a deficient vitamin D level, and Dr. Terry prescribed a double dose of liquid baby vitamins and vitamin K, one-half tablet every other day, for his fat soluble vitamin deficiencies. I did show them also Matthew's abnormal transaminases and alkaline phosphatase indicating cholestasis that is often found in this condition.

IMPRESSION: Four ~~year~~ month old with infantile Refsum's disease and cholestasis. There is no proven effective treatment with infantile Refsum's disease. There are several ongoing research products designed to determine whether supplementation of DHA and/or arachidonic acid is helpful in this condition. I read some of the studies by Dr. Martinez in Spain, and the results look quite promising that DHA might help prevent the blindness and even some of the developmental delays seen in this disorder. I have been pursuing with Dr. William Conner and Dr. Leesa Linck the possibility of supplementing DHA as part of a research protocol. We continue to pursue this and are actively writing a protocol to study the use of DHA in peroxisomal disorders. In the meantime, however, since DHA is available as a food additive rather than an investigational medication in some forms and starting DHA sooner rather than later would be encouraged if there is any hope that it is to help, I will look into the possibility of obtaining DHA from a company that makes it. The research study then would be designed to look for changes in ERG, MRI, development, etc., with DHA treatment and to try to evaluate which form of DHA is best; i.e., pure DHA available in health food stores, DHA from the NIH, DHA plus arachidonic acid, fish oil and/or linolenic acid. In addition, we would like to look at metabolism of DHA in patients with peroxisomal disorders by stable isotope dilution technology.