

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: 06/09/98

CLINIC NAME: METABOLIC CLINIC

DISCIPLINE: PEDIATRICS

Matthew is a 6-month-old infant with infantile Refsum disease who was seen for a follow-up visit today in Metabolic Clinic. He was accompanied by his mother, Joyce, and was evaluated together with Dr. Robert Steiner and Judy Tuerck, R.N.

This is the first follow-up visit for Matthew since his month-long stay in Barcelona, Spain. He is participating in a research study via the Center for Research and Biochemistry and Molecular Biology through Dr. Manuela Martinez at the University Maternity Childrens Hospital. During his month-long stay there he was started on Docosahexaenoic acid ethylester (DHA-EE) with doses increasing from 100-300 mg per day. His steatorrhea markedly improved, as did his liver function studies. His evaluation there included an ophthalmologic evaluation with ERG. The ERG was abnormal, but the funduscopic exam did not show retinitis pigmentosa. Visual and brainstem-evoked potentials were abnormal.

Joyce is quite pleased with Matthew's improvements on the DHA. She states he is much more content and will play up to one hour babbling and entertaining himself. This is in marked contrast to his earlier days when he was very fussy and inconsolable. Developmentally, he is visually alert and auditorially attentive. He babbles and plays with his hands. He is able to sit for a few seconds without support. His head control is much improved. He is now sleeping through the night. He began rolling one month ago. The family is in the process of getting him enrolled in Early Intervention, and he scheduled for an assessment either later this month or some time in July.

DIET: Matthew continues to breast feed. Joyce is supplementing her diet with fish 3-4 times a week to increase the DHA content of her breast milk. Per Dr. Martinez' recommendations, he is not eating any animal fat or any green, leafy vegetables which may contain higher levels of phytanic acid. He is on skim milk.

MEDICATIONS:

1. DHA-EE 300 mg per day.
2. Vitamins E, K, and multivitamins.

Matthew will be receiving an audiologic evaluation today with Dr. Plapinger following our clinic visit.

PHYSICAL EXAMINATION: In general, Matthew is a mildly dysmorphic but alert and attentive baby in no distress. Weight today is 6.3 kg (now at the 5th-10th percentile, up from below the 5th percentile), height is 65 cm, and head circumference is 42.5 cm. Blood pressure is 104/57.

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ORIGINAL-MEDICAL RECORD

CLINIC DATE: 06/09/98

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Med Rec No.: 01-38-94-88

Name: Wulf, Matthew A

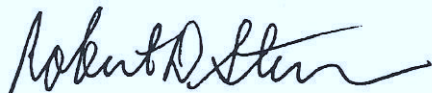
HEENT: Anterior fontanel remains open and flat, but is decreasing in size. The posterior fontanel is now closed. Hair is curly blond, of normal texture and distribution. Matthew's mildly dysmorphic features include flattened nasal bridge and a shortened upturned nose. Ears are normally formed and placed. Pupils are equal, round and reactive to light with a positive red reflex bilaterally. Gaze is conjugate. CHEST: Respirations are quiet and unlabored. ABDOMEN: Soft without hepatosplenomegaly. NEUROLOGIC: Deep tendon reflexes are 1+ and symmetric. Matthew's tone has markedly improved. He has slight head lag when pulled to sit, but is able to maintain his head position when upright. He consoles readily in the mother's arms. He babbles and coos.

ASSESSMENT: Matthew is a 6-month-old with infantile Refsum's disease who appears to be doing very well on the research protocol through Dr. Manuela Martinez which includes DHA-EE. Per Joyce, Dr. Manuela ^{recommends monthly} ~~would like to check labs on a monthly basis~~ ^{monitoring} which we will ~~try to~~ coordinate through this clinic. Laboratory studies obtained today include plasma for pipercolic acid, phytanic acid, and plasminogens. Blood for essential fatty acid studies; red blood cells and plasma; very long chain fatty acids, vitamin A and E levels; liver function studies and PT/PTT.

Pending the results of the liver function and coagulation studies, we may consider weaning Matthew off of vitamin K.

We would like to see Matthew for a follow-up clinic visit in two months. He will return next month for a blood draw only. We will probably continue to see Matthew on a 2-3 month basis in clinic, but he will return for monthly blood draws ^{for disease monitoring} ~~per the study protocol~~. Additionally, Matthew is scheduled for a brain MRI with a Spectroscopy ^{to look for white matter involvement} ~~to look for~~ or signs of intracranial hemorrhage.

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ORIGINAL-MEDICAL RECORD

<p>Oregon Health Sciences University Consultative Report</p> <p>CHILD DEVELOPMENT AND REHABILITATION CENTER</p> <p>P.O. Box 574, Portland, OR 97207-0574</p>	<p>Account No. Medical Record No. 01-38-94-88 Name Wulf, Matthew A Birthdate 12/03/1997</p>
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CLINIC DATE: 10/20/1998
CLINIC NAME: METABOLIC CLINIC
DISCIPLINE: PEDIATRICS


We saw Matthew in the Metabolic Clinic at OHSU on October 20, 1998. Matthew has infantile Refsum disease. He was diagnosed at approximately six weeks of age. He has been followed since then by us and by Dr. Martinez in Spain. Dr. Martinez has Matthew participating in a research protocol to evaluate the efficacy of DHA in the treatment of paroxysmal disease. Matthew currently takes DHA as part of that study.

Matthew has been doing quite well. He has not been sick since his last visit. Developmentally, he continues to gain skills. He is not yet walking, pulling to stand, or cruising, but does have much better head control and is sitting up. He has significant hearing loss, but does seem to be able to hear with hearing aids. He also has visual loss. These are to be expected with his paroxysmal disorder.

On examination, he is hypotonic with some facial dysmorphic features. His liver is not significantly enlarged. He is well appearing.

IMPRESSION: Matthew is almost 1 years old with infantile Refsum disease. I think he is doing quite well. I am encouraged that the DHA treatment may be having some effects in preserving hearing and Vision and improving development above what would be expected. In order to monitor his condition, we collected blood for very long chain fatty acids and phytanic acid.

We would like to see Matthew back in the clinic in 4-6 months.


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