I told the parents that there are no cures or treatments for infantile Refsum's disease. I did explain that several groups are pursuing experimental therapies including DHA and arachidonic acid supplementation. These patients clearly have DHA deficiency usually and the hope is that it is the DHA deficiency that contributes to much of the pathology in this condition and that supplementation might help prevent blindness, deafness, and some developmental problems. I explained that there are research trials in Spain and at Johns Hopkins University in Baltimore, and in addition that we are interested in studying treatments for infantile Refsum's disease here in Portland at Oregon Health Sciences University. In fact, we had discussed putting together a research protocol in this direction with Dr. William Connor and Dr. Richard Weleber even before Matthew was born.

In order to confirm Matthew's diagnosis of infantile Refsum's disease and to look for progression of hepatic dysfunction, we collected blood for free fatty acids and lipoprotein profile, blood pipecolic and phytanic and very long chain fatty acids, PT and PTT, liver panel, vitamin A, 25-hydroxy vitamin D, and vitamin E levels as well as calcium, phosphorus, and alkaline phosphatase. I told the family that we will need to schedule a BAER to check hearing, MRI of the brain with spectroscopy to look for any evidence of intracranial hemorrhage, or abnormalities consistent with infantile Refsum's disease, and finally an ophthalmologic appointment with Dr. Weleber to look for signs of retinitis pigmentosa. We probably would want to have a baseline electroretinogram (ERG) performed to look for subclinical retinal involvement. Finally, I wrote a prescription for vitamin K1 2.5 mg p.o. q.o.d. that Matthew should take to try to prevent bleeding from vitamin K deficiency secondary to cholestasis and liver disease associated with infantile Refsum's disease. Finally, I would like to see Matthew back in clinic in approximately two months for follow-up. During the next two weeks, we will find out if the research study at Johns Hopkins is open for enrollment, what it would take to enroll, what the costs are, and we will find out how long it would take us to obtain a source of DHA and get our protocol approved. I explained to the family that these are research protocols, that there is no guarantee that DHA supplementation will help in this condition, that it is not likely to be harmful, and that the family would have to consent to this experimental therapy before we could consider it.

Dr. Layne, thank you for allowing me to participate in the care of your patient, Matthew Wulf. Please feel free to call if you have any questions or wish to discuss his care.

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