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DIAGNOSES AND REFERENCES

MODERATOR: E. Tessa Hedley-Whyte, M.D.

EDITOR: Leroy R. Sharer, M.D.

Case 2007-4

Submitted by: Brian H. Le, M.D. and James W. Mandell, M.D., Ph.D., Department of Pathology, University of Virginia, Charlottesville VA 22908-0214

Diagnosis: Congenital hypomyelinating neuropathy

Comment: The infant had a gastrocnemius muscle and sural nerve biopsy. The muscle, on myofibrillar ATPase at pH 4.3, had fiber type grouping, but no grouped atrophy, also with many central nuclei. On electron microscopical examination of the nerve, there was no myelin, no evidence of myelin breakdown, no duplication of the basal lamina, and no onion bulbs. MPZ (P0) studies, done at a commercial laboratory, were reported as normal, but other gene studies have not yet been performed. The baby died at age 3.5 months, and an autopsy was not performed. On MRI, central myelin was thought to be normal.

References:

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Szigeti K, Saifi GM, Armstrong D, et al. Disturbance of muscle fiber differentiation in congenital hypomyelinating neuropathy caused by a novel myelin protein zero mutation. Ann Neurol. 2003;54:398-402.