CASE 10

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Ref. No. NP-72-30.

The slide submitted is from a deltoid muscle biopsy in a 4-1/2-year-old boy. The boy had some difficulties at birth due to protracted labor and consequent anoxia. There is questionable slight mental retardation. He was found to be weak and rather immobile at the age of 14 months. Neurologic evaluation on repeated occasions has revealed a fairly static muscle weakness. The child finally was able to walk at 20 months of age but not well and with a waddle and swayback motion. Repeated electromyography was interpreted as normal; repeated enzyme examinations reveal mildly elevated CPK's, last reading 28 Sigma units (January 1972).

The disease is identical with that found in a six-year-old sister, who had normal delivery and early history and was noted to be weak at about 1-1/2 years of age. Again the only change in her was a slightly elevated CPK, last recorded 26 Sigma units (normal 16). The parents and more remote members of the family are all without neurologic disease. There are six other sibs, both older and younger, all well. Due to the fact that these children were abandoned at various times by either or both parents the history obtained when they moved to Indianapolis was not complete. It is now obvious that both children had been previously submitted to muscle biopsy at UCLA in 1968; the results of this examination have been published by Dr. Cancilla et al. in Neurology 27: 579-585, 1971.

A cylinder of muscle tissue was carefully excised. Four micron sections, serial, were studied by enzyme histochemistry. The sections submitted are from various blocks and are stained with the modified rapid trichrome technique of Engel and Cunningham (Neurology, 13: 919, 1963). There may be an occasional annoying bright red stain precipitate.