Case 1993-1

Submitted by: Uma P. Kalyan-Raman, M.D., Robert Cruse, M.D., Tim Miller, M.D. University of Illinois College of Medicine, Peoria, Illinois and Saint Francis Medical Center, Peoria, Illinois 61637

Clinical Summary: This newborn male was born to a 26 year old female in good health. She was on no medications. The pregnancy had a reported history of polyhydraminos without any fetal distress. It was not clear whether there was any intrauterine mobility during pregnancy. He was delivered at thirty-eight weeks gestation by induced labor with vacuum extraction. His weight was about 3,000 gm. His initial APGAR scores were 2 and 7. There was hypotonia at birth and he required supplemental oxygen. Shortly after birth he was transferred to our institution.

Initial examination showed marked hypotonia. He also had a curious tremor of his chin which could be elicited and seemed to be clonus. This persisted for about 24 hours. It also occurred during EEG monitoring and was considered not to be seizure activity. The EEG was normal. Other features of this initial examination included a high arched palate, bland facies and narrow chest, mild flexion of the fingers and limitation of knee extension. This was considered to be consistent with mild arthrogryposis. There was questionable cardiomegaly. The diaphragm appeared to be high bilaterally. There was no evidence of organomegaly. The initial CPK was 1,740 but when repeated four days later was 169. SGOT was 140. His lactic acid was 1.1 mm./l., and pyruvic acid was 0.9 mg./dl. His electrolytes were normal and an EKG was normal. There was no evidence of cardiomyopathy on ultrasound. An EMG was not done. Ultrasound studies of the brain and kidneys were normal. A muscle biopsy was done within four weeks of birth. No definite conclusion could be reached on the muscle biopsy. However, the patient continued to deteriorate and a second muscle biopsy and nerve biopsy were done six weeks later. Soon after the second biopsy, the patient's condition deteriorated and he expired. An autopsy was performed.

Material submitted:
1) Muscle from the second biopsy (cryostat frozen section stained with H&E).
2) Kodachrome photomicrograph of Trichrome stain

Points for discussion:
1. What is your diagnosis?
2. What else needs to be done for a definitive diagnosis?