Clinical History:

A 40 year old G2 P2 mother delivered a full term male infant by repeat Cesarean section, at an outside institution, after an uncomplicated pregnancy. The mother’s first pregnancy was uncomplicated but a Cesarean section was performed for breech presentation. Prenatal labs were unremarkable. At delivery the infant was noted to have poor cry and decreased extremity tone. Apgar scores were 8 and 9 at 1 and 5 minutes, respectively. The patient was transferred to level 2 nursery two days after birth for work up.

On admission the infant had a weak cry and was severely hypotonic in all extremities. The face was symmetric, but flaccid in tone. All cranial nerves were intact and a smile could be elicited. Respiratory effort continued to be labored and sternal retractions were noted. Breath sounds were clear.

During the hospital course the infant was initially on room air with some dusky episodes that seemed to improve, however, 27 days after birth breathing became more labored due to the improper handling of secretions. The infant was treated with frequent suction and placed in a mist trough with oxygen to maintain saturations over 95%. An oral gastric feeding tube was placed due to an inability to swallow without aspiration. Barium swallowing studies were attempted but discontinued in progress due to aspiration. Cytogenetics revealed a normal male karyotype. MRI showed right temporal lobe polymicrogyria. A muscle biopsy was performed.

Material submitted: Two kodachromes of H&E and modified Gomori trichrome stained frozen sections from the right quadriceps muscle.

Points for discussion:
1. Diagnosis
   2. Pathogenesis