48th ANNUAL DIAGNOSTIC SLIDE SESSION 2007

CASE 2007-4

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Clinical History: The patient is a 2 month-old Hispanic male born at 40 weeks gestation via Caesarian section to a 23 year-old, G1P0 mother whose pregnancy had been uneventful. Apgar scores were 6 and 8 at one and five minutes, respectively. Supplemental oxygen was administered to the neonate from 1 to 4 hours of life.

The baby showed dysmorphic facial features and contractures at the hips, elbows, and fingers (arthrogryposis). Hypotonia and areflexia were observed, in addition to poor latch and suction with attempts at feeding. MRI of the brain showed immature sulci development, along with hypoplasia of the corpus callosum in the regions of the posterior body and splenium. The myelination pattern appeared appropriate.

With electrodiagnostic testing, no motor or sensory responses could be obtained in the arms or legs. Needle exam revealed fibrillation potentials and absent/reduced motor unit potentials in the distal and proximal muscles. The interpretation rendered was that of a severe neuropathic process causing profound denervation.

Laboratory data included a creatine kinase level of 144, a CSF protein level of 51, and a karyotype of 46, XY. Combined muscle (left gastrocnemius) and nerve (left sural) biopsies were performed, with the muscle biopsy showing essentially nonspecific myopathic changes (increased fiber size variation with atrophy), but no evidence of grouped atrophy or of fiber type grouping.

Material submitted: Toluidine blue stained cross section of sural nerve

Points for Discussion:

1. Differential diagnosis?
2. Diagnosis?
3. What additional molecular genetic testing could be informative?