49th ANNUAL DIAGNOSTIC SLIDE SESSION, 2008 DIAGNOSES AND REFERENCES

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Case 2008-3

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Diagnosis: Familial amyotrophic lateral sclerosis, with extensive leptomeningeal fibrosis

Comment: Muscle examination during life (muscle biopsy) exhibited marked grouped atrophy involving entire muscle fascicles, with some fibers having basophilic sarcoplasm and increased internal nuclei. SMN gene deletions were not detected in this patient, nor was there a SOD-1 mutation. Immunocytochemical studies at autopsy showed some ubiquitin staining of anterior horn cells, but no evidence of tau. No corticospinal tract changes could be discerned. Fibrosis was present in the spinal leptomeninges, surrounding the cord.

<u>From the Presenter</u>: Immunohistochemical studies for TDP-43 were negative. Meningeal fibrosis was not present in all slides.

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