CASE 1

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Ref. No. U.H. 1005140 A64-2

This 9 month old boy was the product of a normal full term pregnancy. At about three months the patient developed progressive weakness, hepatomegaly, cardiomegaly, and cardiac failure which was terminated by a fatal aspiration pneumonia.

The electromyogram was interpreted as showing muscle denervation secondary to anterior horn cell or axonal destruction. The glucose tolerance test was normal, but the mumps skin test was markedly positive at three months of age.

At autopsy the brain and spinal cord showed no significant gross abnormalities.