Correlation between severity and SMN protein level in spinal muscular atrophy

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Spinal muscular atrophy (SMA) is a common autosomal recessive neuromuscular disorder characterized by degeneration of motor neurons of the spinal cord. Three different forms of childhood SMA have been recognized on the basis of age at onset and clinical course: Werdnig-Hoffmann disease (type-I), the intermediate form (type-II) and Kugelberg-Welander disease (type-III)[1]. A gene termed 'survival of motor neuron' (*SMN*) has been recognized as the disease-causing gene in SMA[2ú6]. *SMN* encodes a protein located within a novel nuclear structure and interacts with RNA-binding proteins[7]. To elucidate the molecular mechanism underlying the pathogenesis of the disease, we examined the expression of the *SMN* gene in both controls and SMA patients by western blot and immunohistochemical analyses using antibodies raised against the SMN protein. The present study shows a marked deficiency of the SMN protein in SMA.

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