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MUSCLE DISORDERS

BRAIN DYSTROPHIN IN DUCHENNE DYSTROPHY

To define the potential pathogenic role of dystrophin deficiency in the cognitive impairment characteristic of Duchenne muscular dystrophy (DMD), the protein in brain cortical synapses of an 8-year-old patient examined at autopsy and an age-matched control subject dying of myelogenous leukemia was analysed in the Department of Neuroscience and Cell Biology, Rutgers-The State University of New Jersey, Piscataway, NJ. Western blot analysis of protein in total homogenate, synaptic membrane, and the highly purified postsynaptic density (PSD) disc beneath the postsynaptic membrane, showed 427-kd dystrophin normally expressed in the PSD of the control tissue but was undetectable in the PSD from the DMD cerebral cortex. (Kim T-W, Wu K, Black IB. Deficiency of brain synaptic dystrophin in human Duchenne muscular dystrophy. *Ann Neurol* September 1995;38:446-449). (Respond: Dr Kim, Laboratory of Genetics and Aging, Department of Neurology, Massachusetts General Hospital East, 13th Street, Charlestown, MA 02129).

COMMENT. Human brain dystrophin is normally present in the cortical synapse but is absent in the brain of a child dying with Duchenne muscular dystrophy. Dystrophin deficiency in DMD may be a factor in both muscle and brain synaptic dysfunction. The patient examined in this report had no obvious cognitive impairment but a subclinical deficit was not excluded. The possible relationship between the dystrophin deficiency in the synapse and cognitive function was undetermined. That brain cortical dysfunction needs further study in patients with DMD is indicated by this report and by occasional observation of Babinski and other abnormal central nervous system signs.

ADHALIN DEFICIENCY AND MUSCULAR DYSTROPHY

Muscle biopsy specimens from 30 muscular dystrophy patients were examined for a deficiency of adhalin, the 50-kd dystrophin-associated protein,

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