Reports claiming ALS caused by head trauma lacks scientific validation

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A recent study[1] suggesting that amyotrophic lateral sclerosis (ALS) may be attributed to "repetitive head trauma experienced in collision sports" lacks scientific epidemiological evidence to support this claim. In a review of the 12-patient study, several experts specializing in motor neuron diseases challenge the findings as entirely pathological and without clinical merit. Their editorial, which aims to dispel doubts of Lou Gehrig's ALS diagnosis, is now available online in the peer-reviewed journal *Muscle & Nerve*.

ALS, also known as Lou Gehrig's disease, is a progressive neurological disease that attacks nerve cells (neurons) in the brain and spinal cord which control voluntary muscles. As the upper and lower motor neurons degenerate, the muscles they control gradually weaken and waste away, leading to paralysis. Other symptoms of ALS include difficulty breathing, issues with swallowing (gagging, choking), and speech problems. According to the National Institutes of Neurological Disorders and Stroke roughly 20,000 to 30,000 Americans have ALS, and 5,000 patients are diagnosed annually with the disease.

In the editorial, the authors cite two media reports—"Brain Trauma Can Mimic Lou Gehrig's Disease" in the New York Times and "Maybe Lou Gehrig Didn't Die of Lou Gehrig's Disease" in Time magazine—as lacking in scientific validation. These reports have led to numerous inquiries from ALS patients who are seeking answers from their physicians.

"Media coverage generated by the McKee et al. study has caused much concern for our ALS patients who now believe they may be misdiagnosed," said Dr. Stanley H. Appel, Chairman of the Department of Neurology at the Methodist Hospital in Houston, Texas, and one of the foremost experts on Lou Gehrig's disease. "We want to make it clear to physicians and their ALS patients that reports of Lou Gehrig succumbing to anything but the disease which bears his name are inaccurate."

The study in question reported pathological changes of chronic traumatic encephalopathy (CTE) in 12 patients, 3 of whom displayed both the TAR DNA-binding protein (TDP-43) and tau in their spinal cords. The build-up of toxic proteins contributes to a number of neurodegenerative diseases, with TDP-43 found in most ALS cases and tau more commonly associated with dementia. Based on the findings of both proteins in the 3 subjects, the study authors concluded that head trauma and CTE led to an "ALS-like" condition, which one author during media interviews, suggested naming chronic traumatic encephalomyopathy (CTEM).

"There is no clinical or pathological evidence of muscle disease reported in the study that would support a diagnosis of CTEM," added Valerie Cwik, M.D., Executive Vice President and Medical Director for the Muscular Dystrophy Association, and co-author of the editorial. "More likely their 3 patients with CTE and ALS had the two distinct diseases."

The editorial authors maintain that an ALS diagnosis is established when patients meet well-defined clinical criteria rather than identification of pathological findings. Editorial contributor, John W. Day, M.D., Ph.D., Director of the Muscular Dystrophy Clinic at the University of Minnesota Medical Center concluded, "Further studies are needed to fully understand the potential role of trauma in ALS and investigation of TDP-43 and tau as potential targets for therapeutic interventions are warranted."

More information: "Trauma, TDP-43, and Amyotrophic Lateral Sclerosis." Stanley H. Appel, Valerie A. Cwik and John W. Day. Muscle & Nerve; Published Online: November 19, 2010; (DOI:10.1002/mus.21939); Print Issue Date: December 2010.

[1] McKee AC, Gavett BE, Stern RA, Nowinski CJ, Cantu RC, Kowall NW, et al. TDP-43 proteinopathy and motor neuron disease in chronic traumatic encephalopathy. J Neuropathol Exp Neurol 2010; 69:918-929.