Study examines delayed, misdiagnosis of sporadic Jakob-Creutzfeldt disease

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A medical record review study of 97 patients with the fatal, degenerative brain disorder sporadic Jakob-Creutzfeldt disease (sCJD) suggests that a correct diagnosis of the disease was often delayed by a variety of misdiagnoses, according to a report published Online First by *Archives of Neurology*.

The disease is often misdiagnosed because of a variability of early symptoms and signs, a variability in disease duration and a lack of recognition of the condition in the medical community. Often, sCJD is mistaken for other neurodegenerative conditions such as Alzheimer disease and dementia with Lewy bodies, according to the study background.

Ross W. Paterson, M.R.C.P., and colleagues from the University of California, San Francisco, retrospectively reviewed all cases referred to the UCSF Memory and Aging Center rapidly progressing dementia and CJD clinical research program between August 2001 and February 2007. They identified 97 patients with pathology-proven sCJD for whom they had sufficient medical records (40 women and 57 men who ranged in age from 26 to 83 years).

The 97 patients had received a combined total of 373 alternative diagnoses prior to their diagnosis of likely CJD, with an average of 3.8 misdiagnoses per patient. The physicians who most commonly made the misdiagnoses were primary care physicians and neurologists. In the 18 percent of patients (17 patients) who were correctly diagnosed at their

first assessment, the diagnosis was almost always made by a neurologist. The average time from onset to diagnosis was almost eight months, an average of two-thirds the way through the disease course, according to the study results.

"In any patient with a rapidly progressive dementia who has been given multiple potential diagnoses, sCJD must be considered," the authors comment.

Researchers note that "early and accurate" diagnosis of sCJD is valuable for public health reasons and to allow for potential treatments to be tested as early as possible in the disease course.

"It would therefore be valuable to improve early and accurate diagnosis of sCJD premortem to identify at-risk persons, allowing for public health measures that would prevent transmission to healthy individuals through blood donation, infected surgical equipment and or other medical procedures," the authors conclude.

In an editorial, Richard J. Caselli, M.D., of the Mayo Clinic Arizona, Scottsdale, writes: "Unquestionably, when confronted with the syndrome of rapidly progressive dementia, our first concern must be the search for reversible causes, a point that Paterson et al discuss in this issue of Archives, but their main point is to illustrate the diagnostic journey patients with CJD travel on their way to a final and very unfortunate diagnosis."

"There is also a societal dimension to the timely diagnosis of CJD. At this time of global economic crisis, national health care reform, escalating medical expenditures and an aging population (creating projections of accelerating health care-driven financial gloom and doom), the financial burden posed by prolonged and duplicative testing that results from diagnostically ambiguous cases of rapidly progressive

dementia must also be considered," Caselli continues.

"As we attempt to rein in health care costs while sacrificing no degree of medical accuracy or compassion, we owe it to our patients and to society to have the necessary knowledge to consider the diagnosis, establish the diagnosis as efficiently as possible, and then work with patients, families and palliative care providers to maximize the quality of life for our dying patients," Caselli concludes.

More information:

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