To: Wisconsin Neurologists, Clinical Laboratory Directors, and Infection Preventionists

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Laboratory Testing Options for Prion Diseases

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Human prion diseases, whether suspected or confirmed, are reportable in Wisconsin. Creutzfeldt-Jakob disease (CJD) is the most common form of human prion disease; other forms include fatal familial insomnia and variant CJD (vCJD). They are all 100% fatal and currently the only method of definitive diagnosis is by post-mortem testing of the brain.

Ante-mortem laboratory testing for prion diseases has historically relied on detection of non-specific markers of neurodegenerative disease in patient CSF specimens, typically the 14-3-3 or tau proteins. Since 2015, the real-time-quaking induced conversion (RT-QuIC) test, which specifically detects the abnormal prion protein found in prion diseases, has been available from the National Prion Disease Pathology and Surveillance Center (NPDPSC) at Case Western Reserve University. This is the only laboratory in the U.S. which performs the RT-QuIC. Recent publications report a sensitivity of 85-89% and specificity of 99% for the RT-QuIC. This is compared to a sensitivity of 86% and specificity of 80% for 14-3-3 (Acta Neurologica Belgica 2018, 118:395-403; Neurology 2018, e331-338). All CSF tested at the NPDPSC is also tested for tau and 14-3-3 proteins.

This memo encourages laboratory directors to ensure testing of CSF at the NPDPSC is available to providers caring for patients with suspected CJD, or other prion diseases. Prion protein-specific ante-mortem testing is a critical step in helping to establish a more accurate ante-mortem diagnosis for the patient and their family, and may encourage families to pursue post-mortem brain autopsy, which is the only means of differentiating familial and sporadic forms of the disease.

For more information regarding testing protocols or submission to the NPDPSC:
https://case.edu/medicine/pathology/divisions/national-prion-disease-pathology-surveillance-center/resources-professionals

For information about reporting a suspect case of prion disease to DPH:

For assistance with brain autopsies, or any other questions, contact Lorna Will, Prion Disease Epidemiologist (lorna.will@wi.gov / 608-267-0401) or Rachel Klos, State Public Health Veterinarian (rachel.klos@wi.gov / 608-266-2154).

The recommendations in this memo were developed in accordance with Wis. Stat. ch. 252 and Wis. Admin. Code ch. DHS 145.