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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:
https://www.dhs.wisconsin.gov/publications/p01610.pdf

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.