MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: May 2, 2024
Effective Date: May 2, 2024

Creutzfeldt-Jakob Disease Evaluation, Spinal Fluid

Test ID: CJDE

Useful for:
Assessment of Creutzfeldt-Jakob disease or other human prion disease in patients with rapidly progressive dementia

Ordering Guidance:
In cases where there is high suspicion of human prion disease supported by clinical or paraclinical (MRI imaging) features, this test should be ordered.

Early in the disease course, or in atypical cases, the disease progression may be slower and include significant clinical overlap (dementia, rigidity, myoclonus) with other potential causes of rapidly progressive dementia, including Alzheimer disease. In the latter case, it would be more appropriate to order RPDE / Rapidly Progressive Dementia Evaluation, Spinal Fluid.

Profile Information:

Test ID	Reporting Name	Available Separately	Always Performed
CJDEI	CJD Eval Interp, CSF	No	Yes
RTQPC	RT-QuIC Prion, CSF	No	Yes
ADCJD	Tau CJD Evaluation, CSF	No	Yes

Methods:
CJDEI: Medical Interpretation
RTQPC: Real-Time Quaking-Induced Conversion (RT-QuIC)
ADCJD: Electrochemiluminescent Immunoassay (ECLIA)

Reference Values:
RT-QuIC PRION, CSF:
Negative

t-TAU/p-TAU:
< or =18

TOTAL TAU:
< or =393 pg/mL

Specimen Requirements:

Supplies:	CJD/RPD Evaluation Kit (T966)
Container/Tube:	
Preferred:	2 Sarstedt CSF False Bottom Tubes 63.614.625 (2.5 mL)
Acceptable:	Sarstedt 72.703.600 (1.5 mL) or Sarstedt 72.694.600 (2 mL)
Specimen Volume:	2 tubes, each containing 1.5 mL to 2.5 mL
Collection Instructions:	1. Perform lumbar puncture and discard the first 1 to 2 mL of cerebrospinal fluid (CSF). 2. Collect two tubes of CSF directly into an acceptable collection tube until the tube is at least 50% full. 3. Send CSF specimen in original collection tube. **Do not aliquot.** 4. Collection instructions can also be found on Spinal Fluid Specimen Collection Instructions for Creutzfeldt-Jakob Disease and Rapidly Progressive Dementia Evaluations (T974).
Minimum Volume:	See Specimen Required

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
CSF	Frozen (preferred)	28 days	BlueTop SARSTEDT
	Ambient	12 hours	BlueTop SARSTEDT
	Refrigerated	14 days	BlueTop SARSTEDT

Cautions:

These test results should be interpreted in the appropriate clinical context along with other clinical and paraclinical findings. Only through neuropathological assessment of brain tissue can a definitive diagnosis of sporadic prion disease be established.

Some molecular subtypes of prion protein have been reported to have lower detectability by real-time quaking-induced conversion (RT-QuIC) assays.

Even small quantities of blood in cerebrospinal fluid (CSF) can result in false-negative RT-QuIC results.

The presence of fluorescent substances may interfere with testing and prevent an accurate interpretation of the RT-QuIC assay.

Careful consideration of the differential diagnosis is advised when RT-QuIC test results are unexpectedly negative. Repeat testing with RT-QuIC may be warranted if there is high suspicion of prion disease. A small subset of initially negative cases by RT-QuIC may become positive as the disease progresses. However, a small proportion of patients with definitive prion disease may be persistently negative by RT-QuIC. False-negative RT-QuIC results are most often encountered in cases of genetic prion disease, such as fatal familial insomnia and Gerstmann-Straussler-Scheinker disease, and in atypical sporadic prion disease subtypes that have slower indolent disease progression.

In rare cases, some individuals can develop antibodies to mouse or other animal antibodies (often referred to as human anti-mouse antibodies [HAMA] or heterophile antibodies), which may cause interference in some immunoassays. The presence of antibodies to streptavidin or ruthenium can also rarely occur and may interfere in this assay. Caution should be used in interpretation of results, and the laboratory should be alerted if the result does not correlate with the clinical presentation.

CPT Code:
84999
83520 x 2

Day(s) Performed: Monday through Friday, Sunday

Report Available: 3 to 8 days

Questions

Contact Amy Ennis, Laboratory Resource Coordinator at 800-533-1710.