MAYO CLINIC LABORATORIES

NEW TEST

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

Rapidly Progressive Dementia Evaluation, Spinal Fluid

Test ID: RPDE

Useful for:

Evaluation of individuals presenting with rapidly progressive dementia of uncertain disease etiology and a differential diagnosis of Creutzfeldt-Jakob disease and rapidly progressive Alzheimer disease

Ordering Guidance:

In individuals with a high clinical suspicion of Alzheimer disease, order ADEVL / Alzheimer Disease Evaluation, Spinal Fluid.

This test can only be performed on specimens collected and transported in polypropylene tubes. If this test is ordered and a polystyrene tube is received, it will be canceled and automatically reordered by the laboratory as CJDE / Creutzfeldt-Jakob Disease Evaluation, Spinal Fluid.

For cases where there is high suspicion of human prion disease supported by clinical or paraclinical magnetic resonance imaging features, order CJDE / Creutzfeldt-Jakob Disease Evaluation, Spinal Fluid.
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 this test.

Profile Information:

Test ID	Reporting Name	Available Separately	Always Performed
RPDEI	RPD Eval Interp, CSF	No	Yes
RTQPC	RT-QuIC Prion, CSF	No	Yes
TTPTQ	t-Tau/p-Tau	No	Yes
ADRTQ	Alzheimer's Disease Evaluation, CSF	No	Yes

Methods:

RPDEI: Medical Interpretation
RTQPC: Real-Time Quaking-Induced Conversion (RT-QuIC)
TTPTQ: Calculation
ADRTQ: Electrochemiluminescent Immunoassay (ECLIA)

Reference Values:

RT-QuIC PRION, CSF:
Negative

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

p-TAU/ABETA 42:
< or =0.028

BETA-AMYLOID (1-42) (Abeta42):
>834 pg/mL

TOTAL TAU:
< or =238 pg/mL (Alzheimer disease)
< or =393 pg/mL (Creutzfeldt-Jakob disease)

PHOSPHORYLATED TAU 181:
< or =21.6 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 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.** **Note: Polystyrene collection tubes are not acceptable.** Exposure of CSF to polystyrene tubes may result in falsely low Abeta42 concentrations. The Alzheimer's Association consensus protocol for handling of CSF for clinical measurements of Abeta42 and tau recommends using the drip method for CSF collection and directly collecting into a low-bind polypropylene tube. Although some clinicians prefer the syringe pull method due to speed of collection, the drip method reduces the risk of Abeta42 binding to the plastic of any syringe used. 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 the real-time quaking-induced conversion (RT-QuIC) assay.

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

The presence of fluorescent substances may interfere with testing and prevent the 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, and in atypical sporadic prion disease subtypes that have slower indolent disease progression.

Improper specimen handling or interindividual differences in overall concentration of Abeta peptide production may yield an abnormally low Abeta42 in the context of a normal p-Tau181/Abeta42 ratio. Results should be interpreted in combination with other clinical information.

Exposure of cerebrospinal fluid to polystyrene tubes can reduce concentrations of the amyloid Abeta42 by as much as 20% to 50% due to adherence of the sticky amyloid protein to polystyrene tube surface material, potentially altering clinical interpretation, including the p-Tau181/Abeta 42 ratio. P-Tau181 and total Tau protein do not substantially adhere to polystyrene collection tubes.

Failure to adhere to the specimen collection instructions provided may result in falsely low Abeta42 concentrations and potential misdiagnosis of Alzheimer disease.

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 3

Day(s) Performed: Monday through Friday, Sunday **Report Available:** 3 to 8 days

Questions

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