

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Rett Syndrome (MECP2) or (CDKL5) testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR RETT SYNDROME (MECP2) OR CDKL5-RELATED DISORDERS (CDKL5) TESTING

Patient Name _____ **Date of Birth** ____/____/____ **Gender** F M

Physician _____ **Physician Phone** (____) _____ **Practice specialty** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

PATIENT'S ETHNICITY (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African-American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Does the patient have SYMPTOMS? No Yes If yes, check all that apply

- | | |
|---|--|
| <input type="checkbox"/> Normal perinatal history | <input type="checkbox"/> Encephalopathy |
| <input type="checkbox"/> Normal head circumference at birth | <input type="checkbox"/> Abnormal EEG |
| <input type="checkbox"/> Head growth deceleration | <input type="checkbox"/> Seizures (age of onset: _____) |
| <input type="checkbox"/> Period of normal development | <input type="checkbox"/> Spasticity |
| <input type="checkbox"/> Developmental regression | <input type="checkbox"/> Non-ambulatory |
| <input type="checkbox"/> Current microcephaly | <input type="checkbox"/> Gait ataxia |
| <input type="checkbox"/> Growth retardation | <input type="checkbox"/> Non-verbal |
| <input type="checkbox"/> Loss of purposeful hand movement | <input type="checkbox"/> Loss of speech |
| <input type="checkbox"/> Repetitive hand movements | <input type="checkbox"/> Hypotonia |
| <input type="checkbox"/> Mental retardation | <input type="checkbox"/> Breathing abnormalities |
| <input type="checkbox"/> Autistic features | <input type="checkbox"/> Recurrent respiratory infection |
| <input type="checkbox"/> Other _____ | |

Does the patient have a **FAMILY HISTORY** of mental retardation/autism/Rett syndrome? No Yes Unknown

If yes, specify the **RELATIONSHIP** of the symptomatic family member(s) to the patient and detail the symptoms/diagnosis in each.

Please attach a copy of the relative's *MECP2/CDKL5* laboratory result (REQUIRED for familial mutation testing).

Circle the test below you intend to order.

0051614 Rett Syndrome (MECP2) Sequencing and Deletion/Duplication - Sequencing of *MECP2* coding regions and deletion/duplication analysis for large genomic rearrangements; clinical sensitivity up to 95%.

0051378 Rett Syndrome (MECP2) Sequencing - Sequencing of *MECP2* coding regions with 80% clinical sensitivity.

2004935 CDKL5-Related Disorders (CDKL5) Sequencing and Deletion/Duplication - Order to exclude a diagnosis of atypical Rett syndrome or X-linked spasm syndrome. Clinical sensitivity of ~17% for females with early onset seizures/spasms.

2004931 CDKL5-Related Disorders (CDKL5) Sequencing - Clinical sensitivity of ~17% for females with early onset seizures/spasms.

2001961 Familial Mutation, Targeted Sequencing - Tests for a *MECP2* or *CDKL5* sequence change previously identified in a family member; copy of relative's lab result is REQUIRED.

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141

Master Label