

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's 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.

0051618 Rett Syndrome (*MECP2*), Deletion and Duplication- Detects large deletions or duplications of *MECP2*; clinical sensitivity up to 15%.

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.

2004927 *CDKL5*-Related Disorders (*CDKL5*) Deletion/Duplication- Clinical sensitivity unknown.

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

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