ARUP LABORATORIES

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)

[ ] African-American	[ ] Ashkenazi Jewish	[ ] Asian	[ ] Caucasian
[ ] Hispanic	[ ] Middle Eastern	[ ] Native American	[ ] Other_____

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

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

Master Label