ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
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 _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American

☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply)

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

☐ Other symptom(s): _____

Is there any relevant family history of intellectual disability/autism/Rett syndrome? ☐ No ☐ Yes ☐ Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms/diagnosis:

_____

_____

Has *MECP2/CDKL5* testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's laboratory result (REQUIRED for familial mutation testing).

Check the test 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 mutation previously identified in a family member; copy of relative's lab result is REQUIRED.

Master Label

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