ARUP LABORATORIES

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)* TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male
Physician: _____ **Physician Phone:** _____
Practice Specialty: _____ **Physician Fax:** _____
Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian/White ☐ 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.

☐ **2001961 Familial Mutation, Targeted Sequencing** Tests for a sequence variant 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.