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 complete and submit with the test request form or electronic packing list.

PATIENT HISTORY FOR RETT SYNDROME (*MECP2*) TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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.

☐ **3003144 Deletion/Duplication by MLPA**: Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.

Master Label

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