

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)

- | | | |
|---|---|--|
| <input type="checkbox"/> Abnormal EEG | <input type="checkbox"/> Head growth deceleration | <input type="checkbox"/> Normal perinatal history |
| <input type="checkbox"/> Autistic features | <input type="checkbox"/> Hypotonia | <input type="checkbox"/> Period of normal development |
| <input type="checkbox"/> Breathing abnormalities | <input type="checkbox"/> Intellectual disability | <input type="checkbox"/> Recurrent respiratory infection |
| <input type="checkbox"/> Current microcephaly | <input type="checkbox"/> Loss of purposeful hand movement | <input type="checkbox"/> Repetitive hand movements |
| <input type="checkbox"/> Developmental regression | <input type="checkbox"/> Loss of speech | <input type="checkbox"/> Seizures (age of onset: _____) |
| <input type="checkbox"/> Encephalopathy | <input type="checkbox"/> Non-ambulatory | <input type="checkbox"/> Spasticity |
| <input type="checkbox"/> Gait ataxia | <input type="checkbox"/> Non-verbal | |
| <input type="checkbox"/> Growth retardation | <input type="checkbox"/> Normal head circumference at birth | |
| <input type="checkbox"/> 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 rearrangements; clinical sensitivity up to 95%.
- 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.



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