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 ANGELMAN SYNDROME DNA 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 and describe)

☐ Abnormal EEG ☐ Inappropriate happy demeanor ☐ Scoliosis

☐ Developmental delay ☐ Microcephaly ☐ Seizures

☐ Excitability ☐ Minimal or absent speech ☐ Sleep disturbance

☐ Gait ataxia and/or tremor of limbs ☐ Normal head circumference at birth ☐ Tongue thrusting; feeding problems

☐ Hand flapping ☐ Obesity ☐ Strabismus

☐ Other symptom(s): _____

Has the patient undergone previous DNA testing for Angelman syndrome? ☐ No ☐ Yes ☐ Unknown

☐ DNA methylation analysis; result: _____

☐ FISH analysis; result: _____

☐ Array CGH; result: _____

☐ UPD testing; result: _____

☐ Imprinting center defect testing; result: _____

Is there any relevant family history of severe developmental delay/MR/Angelman syndrome? ☐ No ☐ Yes ☐ Unknown

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

_____

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

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

Check the test you intend to order.

☐ **2005077 Angelman Syndrome and Prader-Willi Syndrome by Methylation:** The most sensitive test to confirm a diagnosis of Angelman syndrome and Prader-Willi syndrome; clinical sensitivity is approximately 78% for Angelman syndrome and 99% for Prader-Willi syndrome.

☐ **2005564 Angelman Syndrome (*UBE3A*) Sequencing:** To confirm a diagnosis of Angelman syndrome in individuals with a normal DNA methylation test; clinical sensitivity is approximately 11%.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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