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.

ANGELMAN SYNDROME DNA TESTING PATIENT HISTORY FORM

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) ☐ Unknown

☐ 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 and age of onset:

_____

_____

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; copy of relative's lab result is REQUIRED.

Master Label

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