ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

500 Chipeta Way
Salt Lake City, Utah 84108-1221
phone: (801) 583-2787 | toll free: (800) 242-2787
fax: (801) 584-5249 | www.aruplab.com

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 KABUKI SYNDROME (KS) 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)

☐ Abnormal dentition
☐ Cardiac malformations
☐ Cryptorchidism
☐ Dermatoglyphic pattern abnormal
☐ Distal interphalangeal flexion crease absent or attenuated
☐ Early breast development in infant girls
☐ Facial features of KS
☐ Feeding difficulties
☐ Gastroesophageal reflux
☐ Hearing loss
☐ Hypospadias
☐ Hypotonia
☐ Intellectual disability/developmental delay
☐ Joint dislocations/hypermobility
☐ Microcephaly
☐ Ocular abnormalities
☐ Palatal malformations
☐ Persistent fetal fingertip pads
☐ Renal malformations
☐ Repeated infections
☐ Seizures
☐ Short stature
☐ Skeletal abnormalities
☐ Structural brain abnormalities

☐ Other symptom(s): _____

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

If yes, describe the test(s) and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown

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

_____

_____

Has DNA testing for the *KMT2D* gene 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.

☐ **2009306 Kabuki Syndrome (*KMT2D*) Sequencing:** Clinical sensitivity ~ 70% in patients with a clinical diagnosis of KS.

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

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

Master Label