ARUP LABORATORIES

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform kabuki syndrome testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR KABUKI SYNDROME TESTING

Patient Name _____ Date of Birth_____ /_____/_____ Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

Genetic Counselor _____ Counselor Phone (_____)_____

Patient's Ethnicity (check all that apply)

[ ] African American [ ] Ashkenazi Jewish [ ] Asian [ ] Caucasian
[ ] Hispanic [ ] Middle Eastern [ ] Native American [ ] Other_____

Does the patient have SYMPTOMS of KABUKI SYNDROME (KS)? [ ] No [ ] Yes [ ] Unknown

If yes, check all that apply:

[ ] Abnormal dentition
[ ] Cardiac malformations
[ ] Cryptorchidism
[ ] Dermatoglyphic pattern abnormal
[ ] 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
[ ] Distal interphalangeal flexion crease absent or attenuated

[ ] Other _____

Has the patient undergone previous DNA testing for KS? [ ] No [ ] Yes
If yes, please describe test(s) and results_____

Does the patient have a FAMILY HISTORY of KS? [ ] No [ ] Yes [ ] Unknown
If yes, please specify the RELATIONSHIP of the family member(s) to the patient and describe the symptoms in each symptomatic relative. _____

Has DNA testing for the *KMT2D* gene been performed for these family member(s)? [ ] No [ ] Yes [ ] Unknown
If yes, please attach a copy of the laboratory result (REQUIRED for familial mutation testing)

Circle the KS 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: Targeted sequencing for a *KMT2D* mutation previously identified in a family member. A copy of relative's DNA laboratory result is REQUIRED for familial mutation testing.

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

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