ARUP LABORATORIES

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

PATIENT HISTORY FOR PALLISTER-HALL SYNDROME/GREIG CEPHALOPOLYSYNDACTYLY SYNDROME (*GLI3*) 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?[ ] No [ ] Yes; please check all that apply

[ ] Bifid epiglottis
[ ] Hydrocephalus
[ ] Hypertelorism
[ ] Hypothalamic hamartoma
[ ] Imperforate anus
[ ] Intellectual disability
[ ] Macrocephaly
[ ] Non-polydactyly skeletal anomalies
[ ] Polydactyly (__ postaxial/central __ preaxial)
[ ] Pulmonary segmentation anomalies
[ ] Renal/genitourinary anomalies
[ ] Seizures

Does the patient have a FAMILY HISTORY of Pallister-Hall syndrome (PHS)/Greig Cephalopolysyndactyly syndrome (GCPS)? [ ] No [ ] Yes [ ] Unknown
If yes, attach a PEDIGREE or specify the relatives' RELATIONSHIP to the patient. Please list their symptoms.

_____

Has DNA testing been performed for these family member(s)? [ ] No [ ] Yes [ ] Unknown

Has the patient undergone previous DNA testing for PHS/GCPS? [ ] No [ ] Yes [ ] Unknown

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

Circle the test you intend to order OR write the test name and number below:

Recommended testing for PHS/GCPS	
2011465	***GLI3*-Related Disorders *(GLI3)* Sequencing and Deletion/Duplication:** Clinical sensitivity is 90% for PHS and 75-85% for GCPS
2011470	**GLI3*GLI3*-Related Disorders (*GLI3*) Sequencing:** Clinical sensitivity is 90% for PHS and 70% for GCPS
2011424	***GLI3*-Related Disorders *(GLI3)* Deletion/Duplication:** Clinical sensitivity is unknown for PHS and is 5-10% for GCPS
Targeted testing for known mutation (laboratory report from family member REQUIRED)	
2001961	**Familial Mutation, Targeted Sequencing:** targeted testing for a known familial sequence mutation
2011424	***GLI3*-Related Disorders *(GLI3)* Deletion/Duplication:** for known familial deletion/duplication

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

Master Label