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 PALLISTER-HALL SYNDROME/ GREIG CEPHALOPOLYSYNDACTYLY SYNDROME (*GLI3*) 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)

☐ 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

Has the patient undergone previous DNA testing for PHS/GCPS? ☐ 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 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.

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*-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)

☐ **2011424 *GLI3*-Related Disorders (*GLI3*) Deletion/Duplication:** For known familial deletion/duplication.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member.

Master Label

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