NIGMS HUMAN GENETIC CELL REPOSITORY

PITT-HOPKINS SYNDROME CLINICAL DATA ELEMENTS FORM

Sample ID#:_____ **Age at Onset of Symptoms**: _____ **Age at Diagnosis**: _____

Genetic Testing *(please attach a copy of the results, if available)***:**
List *TCF4* gene mutation or describe deletion/translocation:

_____
Test methodology (PCR, array CGH, sequencing, etc):

_____

Clinical Information (*please check all that apply*)

Pregnancy:

☐ Abnormal Serum Screen	☐ Advanced Maternal Age	☐ Fetal Abnormality *(note below)*
☐ IUGR	☐ Oligohydramnios	☐ Polyhydramnios
☐ Increased Nuchal Translucency	☐ Cystic Hygroma	☐ Hydrops (Unknown or Infection)
☐ 2 Vessel Umbilical Cord	☐ Premature Delivery: ____Weeks	☐ Prior Affected Pregnancy
☐ Breech	☐ Decreased Fetal Movement	

☐ Other: _____

Neurological - Structural Findings:

☐ Microcephaly	☐ Atrophy of Frontal/Parietal Cortex	☐ Ventricular Asymmetry
☐ Bulging Caudate Nuclei	☐ Agenesis/Hypoplasia of Corpus Callosum	

☐ Other Structural Brain Anomaly: _____

Neurological - Clinical Findings:

☐ Absent or Sparse Speech	☐ Hypotonia	☐ Hypertonia
☐ Clonus	☐ Seizures	☐ Delayed Motor Development
☐ Limited Walking Ability	☐ Unstable, Ataxic Gait	☐ Incoordination
☐ Strabismus	☐ Defective Vision (Myopia, Astigmatism, etc.)	

☐ Other: _____

Craniofacial:

☐ Coarse Face	☐ Bitemporal Narrowing	☐ Deep-Set Eyes
☐ Square Forehead	☐ Upslanting Palpebral Fissures	☐ Broad Nasal Bridge
☐ Beaked Nasal Bridge	☐ Flaring Nostrils	☐ Downturned/Pointed Nasal Tip
☐ Short Philtrum	☐ Full Cheeks	☐ High Cheek Bones
☐ Wide, Open Mouth	☐ Thick, Fleshy Lips	☐ Tented/Cupid Bowed Upper Lip
☐ Cleft Lip	☐ Cleft Palate	☐ Widely Spaced Teeth
☐ Cupped Ears	☐ Thick/Fleshy Helices	☐ Short Neck

☐ Lower Face Prominence/Well-Developed Chin

☐ Other: _____

Musculoskeletal:

☐ Slender/Small Hands ☐ Slender/Small Feet ☐ Fetal Pads
☐ Clinodactyly ☐ Clubbed Fingers ☐ Tapered Fingers
☐ Simian Crease ☐ Pes Planus ☐ Pes Valgus
☐ Other: _____

Respiratory:

☐ Abnormal Breathing Patterns ☐ Intermittent Breathing ☐ Hyperventilation
☐ Apnea ☐ Aspiration ☐ Sleep Difficulties
☐ Other: _____

Cutaneous:

☐ Hyperpigmentation ☐ Hypopigmentation ☐ Dry Skin
☐ Other: _____

Gastrointestinal:

☐ Constipation ☐ Gastroesophageal Reflux
☐ Other: _____

Genitourinary:

☐ Small Penis ☐ Cryptorchidism
☐ Other: _____

Cognitive/Behavioral:

☐ Intellectual Disability:___(IQ/DQ) ☐ Happy Personality ☐ Aggression
☐ Stereotypical Movements ☐ Anxiety ☐ Behavioral Problems
☐ Autism ☐ Sensory Processing Disorder
☐ Other: _____

Assistive Devices:

☐ Wheelchair ☐ Walker ☐ Braces
☐ Orthotics ☐ Hearing Aid ☐ Glasses
☐ Service Animal ☐ Communication/Learning Device
☐ Other: _____

Treatment and Management:

☐ Physical Therapy ☐ Occupational Therapy ☐ Psychological Therapy
☐ Speech Language Therapy ☐ Special Education Services
☐ Other:_____

☐Medication(s):_____
☐Surgeries: _____

Please describe additional dysmorphology, behaviors, or other clinical features below (or attach relevant clinical documents and/or test results):

_____
_____
_____

<u>Once this form is complete</u>:

- Please include form and the General Clinical Data elements form (and other relevant documents) with the sample in the shipping box.
- You can also email the form/documents to nigms@coriell.org or fax it to 856-437-5638.