

## 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:**

- |  |  |   |
|--|--|---|
| <input type="checkbox"/> Abnormal Serum Screen         | <input type="checkbox"/> Advanced Maternal Age         | <input type="checkbox"/> Fetal Abnormality (note below) |
| <input type="checkbox"/> IUGR                          | <input type="checkbox"/> Oligohydramnios               | <input type="checkbox"/> Polyhydramnios                 |
| <input type="checkbox"/> Increased Nuchal Translucency | <input type="checkbox"/> Cystic Hygroma                | <input type="checkbox"/> Hydrops (Unknown or Infection) |
| <input type="checkbox"/> 2 Vessel Umbilical Cord       | <input type="checkbox"/> Premature Delivery: ____Weeks | <input type="checkbox"/> Prior Affected Pregnancy       |
| <input type="checkbox"/> Breech                        | <input type="checkbox"/> Decreased Fetal Movement      |   |
| <input type="checkbox"/> Other: _____                  |  |   |

**Neurological - Structural Findings:**

- |  |   |  |
|--|---|--|
| <input type="checkbox"/> Microcephaly                          | <input type="checkbox"/> Atrophy of Frontal/Parietal Cortex     | <input type="checkbox"/> Ventricular Asymmetry |
| <input type="checkbox"/> Bulging Caudate Nuclei                | <input type="checkbox"/> Agenesis/Hypoplasia of Corpus Callosum |  |
| <input type="checkbox"/> Other Structural Brain Anomaly: _____ |   |  |

**Neurological - Clinical Findings:**

- |  |   |  |
|--|---|--|
| <input type="checkbox"/> Absent or Sparse Speech | <input type="checkbox"/> Hypotonia                                    | <input type="checkbox"/> Hypertonia                |
| <input type="checkbox"/> Clonus                  | <input type="checkbox"/> Seizures                                     | <input type="checkbox"/> Delayed Motor Development |
| <input type="checkbox"/> Limited Walking Ability | <input type="checkbox"/> Unstable, Ataxic Gait                        | <input type="checkbox"/> Incoordination            |
| <input type="checkbox"/> Strabismus              | <input type="checkbox"/> Defective Vision (Myopia, Astigmatism, etc.) |  |
| <input type="checkbox"/> Other: _____            |   |  |

**Craniofacial:**

- |  |  |   |
|--|--|---|
| <input type="checkbox"/> Coarse Face                               | <input type="checkbox"/> Bitemporal Narrowing          | <input type="checkbox"/> Deep-Set Eyes                |
| <input type="checkbox"/> Square Forehead                           | <input type="checkbox"/> Upslanting Palpebral Fissures | <input type="checkbox"/> Broad Nasal Bridge           |
| <input type="checkbox"/> Beaked Nasal Bridge                       | <input type="checkbox"/> Flaring Nostrils              | <input type="checkbox"/> Downturned/Pointed Nasal Tip |
| <input type="checkbox"/> Short Philtrum                            | <input type="checkbox"/> Full Cheeks                   | <input type="checkbox"/> High Cheek Bones             |
| <input type="checkbox"/> Wide, Open Mouth                          | <input type="checkbox"/> Thick, Fleshy Lips            | <input type="checkbox"/> Tented/Cupid Bowed Upper Lip |
| <input type="checkbox"/> Cleft Lip                                 | <input type="checkbox"/> Cleft Palate                  | <input type="checkbox"/> Widely Spaced Teeth          |
| <input type="checkbox"/> Cupped Ears                               | <input type="checkbox"/> Thick/Fleshy Helices          | <input type="checkbox"/> Short Neck                   |
| <input type="checkbox"/> Lower Face Prominence/Well-Developed Chin |  |   |
| <input type="checkbox"/> Other: _____                              |  |   |

**Musculoskeletal:**

- |  |   |  |
|--|---|--|
| <input type="checkbox"/> Slender/Small Hands | <input type="checkbox"/> Slender/Small Feet | <input type="checkbox"/> Fetal Pads      |
| <input type="checkbox"/> Clinodactyly        | <input type="checkbox"/> Clubbed Fingers    | <input type="checkbox"/> Tapered Fingers |
| <input type="checkbox"/> Simian Crease       | <input type="checkbox"/> Pes Planus         | <input type="checkbox"/> Pes Valgus      |
| <input type="checkbox"/> Other: _____        |   |  |

**Respiratory:**

- |  |   |   |
|--|---|---|
| <input type="checkbox"/> Abnormal Breathing Patterns | <input type="checkbox"/> Intermittent Breathing | <input type="checkbox"/> Hyperventilation   |
| <input type="checkbox"/> Apnea                       | <input type="checkbox"/> Aspiration             | <input type="checkbox"/> Sleep Difficulties |
| <input type="checkbox"/> Other: _____                |   |   |

**Cutaneous:**

- |  |   |                                   |
|--|---|-----------------------------------|
| <input type="checkbox"/> Hyperpigmentation | <input type="checkbox"/> Hypopigmentation | <input type="checkbox"/> Dry Skin |
| <input type="checkbox"/> Other: _____      |   |                                   |

**Gastrointestinal:**

- |                                       |  |
|---------------------------------------|--|
| <input type="checkbox"/> Constipation | <input type="checkbox"/> Gastroesophageal Reflux |
| <input type="checkbox"/> Other: _____ |  |

**Genitourinary:**

- |                                       |   |
|---------------------------------------|---|
| <input type="checkbox"/> Small Penis  | <input type="checkbox"/> Cryptorchidism |
| <input type="checkbox"/> Other: _____ |   |

**Cognitive/Behavioral:**

- |   |  |  |
|---|--|--|
| <input type="checkbox"/> Intellectual Disability:___(IQ/DQ) | <input type="checkbox"/> Happy Personality           | <input type="checkbox"/> Aggression          |
| <input type="checkbox"/> Stereotypical Movements            | <input type="checkbox"/> Anxiety                     | <input type="checkbox"/> Behavioral Problems |
| <input type="checkbox"/> Autism                             | <input type="checkbox"/> Sensory Processing Disorder |  |
| <input type="checkbox"/> Other: _____                       |  |  |

**Assistive Devices:**

- |   |  |                                  |
|---|--|----------------------------------|
| <input type="checkbox"/> Wheelchair     | <input type="checkbox"/> Walker                        | <input type="checkbox"/> Braces  |
| <input type="checkbox"/> Orthotics      | <input type="checkbox"/> Hearing Aid                   | <input type="checkbox"/> Glasses |
| <input type="checkbox"/> Service Animal | <input type="checkbox"/> Communication/Learning Device |                                  |
| <input type="checkbox"/> Other: _____   |  |                                  |

**Treatment and Management:**

- |  |   |  |
|--|---|--|
| <input type="checkbox"/> Physical Therapy        | <input type="checkbox"/> Occupational Therapy       | <input type="checkbox"/> Psychological Therapy |
| <input type="checkbox"/> Speech Language Therapy | <input type="checkbox"/> Special Education Services |  |
| <input type="checkbox"/> Other: _____            |   |  |

- Medication(s): \_\_\_\_\_
- Surgeries: \_\_\_\_\_

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

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

**Once this form is complete:**

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