

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
☐ Clinodactyly
☐ Simian Crease
☐ Other: _____

☐ Slender/Small Feet
☐ Clubbed Fingers
☐ Pes Planus

☐ Fetal Pads
☐ Tapered Fingers
☐ Pes Valgus

Respiratory:

☐ Abnormal Breathing Patterns
☐ Apnea
☐ Other: _____

☐ Intermittent Breathing
☐ Aspiration

☐ Hyperventilation
☐ Sleep Difficulties

Cutaneous:

☐ Hyperpigmentation
☐ Other: _____

☐ Hypopigmentation

☐ Dry Skin

Gastrointestinal:

☐ Constipation
☐ Other: _____

☐ Gastroesophageal Reflux

Genitourinary:

☐ Small Penis
☐ Other: _____

☐ Cryptorchidism

Cognitive/Behavioral:

☐ Intellectual Disability:____(IQ/DQ)
☐ Stereotypical Movements
☐ Autism
☐ Other: _____

☐ Happy Personality
☐ Anxiety
☐ Sensory Processing Disorder

☐ Aggression
☐ Behavioral Problems

Assistive Devices:

☐ Wheelchair
☐ Orthotics
☐ Service Animal
☐ Other: _____

☐ Walker
☐ Hearing Aid
☐ Communication/Learning Device

☐ Braces
☐ Glasses

Treatment and Management:

☐ Physical Therapy
☐ Speech Language Therapy
☐ Other: _____

☐ Occupational Therapy
☐ Special Education Services

☐ Psychological Therapy

☐ 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 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.