CORIELL INSTITUTE FOR MEDICAL RESEARCH

NIGMS HUMAN GENETIC CELL REPOSITORY

Propionic Acidemia Clinical Data Elements Form

Sample ID or PA ID #: _____
Age at Diagnosis: _____ **Age at Symptom Onset:** _____

Neonatal History:
Was newborn screening performed? ☐ Yes ☐ No
If yes, what was the result: ☐ Positive ☐ Negative
Was the newborn screen result known prior to hospitalization? ☐ Yes ☐ No
Birth weight: _____kg Birth length: _____cm
Was child breast-fed? ☐ Yes ☐ No; If Yes, duration: _____ Failure to thrive? ☐ Yes ☐ No

Molecular/Enzyme Laboratory Test Results:
Gene: PCCA _____ PCCB _____
Mutations: Allele 1: _____ Allele 2: _____
Lab that determined mutations: _____ Test Method: _____
Enzyme activity: _____% of normal; Absolute Amount: _____
Lab that determined enzyme activity: _____
Are fibroblasts available? ☐ Yes ☐ No ☐ Unknown; If Yes, which laboratory?: _____

Clinical Evaluation:
Current weight: _____kg Current height: _____cm

Respiratory Evaluation:
Apnea: ☐ Yes ☐ No ☐ Unknown Tachypnea: ☐ Yes ☐ No ☐ Unknown

Cardiovascular Evaluation:
Cardiomyopathy: ☐Yes ☐ No ☐ Unknown; If Yes: ☐ Mild ☐ Moderate ☐ Severe; Age at diagnosis: ___
Heart transplant: ☐Yes ☐ No ☐ Unknown; If Yes, Age at transplant: _____
Long QT: ☐ Yes ☐ No ☐ Unknown; If Yes: ☐ Chronic ☐ Acute; Age at diagnosis: _____

Abdominal/Gastrointestinal Evaluation:
Pancreatitis: ☐ Yes ☐ No ☐ Unknown; If Yes: ☐ Chronic ☐ Acute
Kidney problems: ☐ Yes ☐ No ☐ Unknown
Liver transplant: ☐ Yes ☐ No ☐ Unknown; If Yes, age at transplant: _____
Port-a-cath: ☐ Currently in place ☐ Not currently in place ☐ Never used
Gut motility: ☐ Slow ☐ Normal ☐Requires medication, list medication(s):_____
Reflux: ☐ Yes ☐ No ☐ Unknown; If Yes, List medication(s): _____
Eating by mouth: ☐ 100% ☐ 51-99% ☐ 1-50% ☐ 0%
Tube feeding: ☐ NG tube ☐ G-tube ☐ GJ-tube ☐ J-tube ☐ Other:_____
Vomiting: ☐ Daily ☐ 1x week or more ☐ 1x month or more ☐ Infrequently
Nissen fundoplication ☐Yes ☐ No
Anti-emetics: ☐ Yes ☐ No; If Yes, list medication(s):_____

Neurological Evaluation:
Basal ganglia damage: ☐ Yes ☐ No ☐ Unknown; If Yes, determined by ☐ MRI ☐ CT
Seizures: ☐ Yes ☐ No ☐ Unknown; Type: _____ Frequency: _____

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Autism spectrum disorder: ☐ Yes ☐ No ☐ Unknown ADD/ADHD: ☐ Yes ☐ No ☐ Unknown
Optic nerve damage: ☐ Yes ☐ No ☐ Unknown; If Yes, age at detection: _____

Hematologic/Immunologic Evaluation:
Neutropenia: ☐ Yes ☐ No ☐ Unknown; If Yes, ☐ Chronic ☐ Acute; List medication(s):_____
Anemia: ☐ Yes ☐ No ☐ Unknown; If Yes, ☐ Chronic ☐ Acute
Immune deficiency: ☐ Yes ☐ No ☐ Unknown
If Yes, treatment with immunoglobulins? : ☐ Yes ☐ No ☐ Unknown
Low platelets: ☐ Yes ☐ No ☐ Unknown; If Yes, ☐ Chronic ☐ Acute
Asthma: ☐Yes ☐ No ☐ Unknown Food or environmental allergies: ☐ Yes ☐ No ☐ Unknown

Skeletal Evaluation:
Secondary hip dysplasia: ☐ Yes ☐ No ☐ Unknown Osteoporosis: ☐ Yes ☐ No ☐ Unknown
Broken bones: ☐ Yes ☐ No ☐ Unknown
If yes, type of fracture(s):_____ Location(s):_____
Short stature: ☐ Yes ☐ No ☐ Unknown Growth hormone treatment: ☐ Yes ☐ No ☐ Unknown

Developmental Evaluation:
Walking: ☐ Not at all ☐ 25% of the time ☐ 50% of the time ☐ 75% of the time ☐ All the time
Age when began walking: _____
Language: ☐ Age appropriate ☐ Slightly below age level ☐ Significantly below age level
☐ Uses assistive technology (Device, PECS or sign) ☐ No communication
Age when began talking: _____
Cognitive ability: ☐ Age appropriate ☐ Mildly impaired ☐ Moderately impaired ☐ Severely impaired
IQ: ☐ Known (give value or age level) _____ ☐ Untested/unknown

Metabolic Evaluation:
Responsive to biotin? ☐ Yes ☐ No ☐ Unknown
Ketoacidosis: ☐ Yes ☐ No ☐ Unknown If Yes, frequency: ☐ 1x week ☐ 1x month ☐ Infrequently
Chronic hyperammonemia: ☐ Yes ☐ No ☐ Unknown
If currently elevated: ☐ <2x normal ☐ 2x normal ☐ >2x normal
Metabolic formula: ☐ Yes ☐ No ☐ Unknown; If Yes, list formula: _____
Levocarnitine: ☐Yes ☐ No ☐ Unknown; If Yes, _____mg/kg
Supplements: ☐ Co-Q10 ☐DHA ☐ Vitamin E ☐ B-6 ☐ Biotin ☐ Thiamin ☐ Multi-vitamin ☐ Iron
☐ Other: _____
Please describe any additional clinical features: _____
_____

Please direct all questions regarding this request to:

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