

Propionic Acidemia Questionnaire
Propionic Acidemia Foundation
Coriell Institute for Medical Research

Please take a few minutes to have your physician fill out the following questionnaire. The information here will be of help for scientists who will be using the cells and DNA prepared from the samples that are being submitted. The clinical, demographic and laboratory information will be placed in the Coriell database. Any personal identifiers, such as name or date of birth, will be maintained only by the PA Foundation, so that you are able to be re-contacted. Please contact Jill Franks of the PA Foundation or Barbara Frederick at the Coriell Institute, if you have any questions about this form.

Coriell Reference # _____

PA # _____

Subject Profile:

Name of individual: _____ Date of Birth: _____

Race: Caucasian Black Asian East Indian Mixed Race/Other _____

Country of birth: _____

Ethnicity: _____

Hispanic Non-Hispanic

Gender: Male Female

Age at child's diagnosis of PA: _____

Neonatal Data:

Was this child newborn screened? Yes No

If yes, was the result prior to hospitalization? Yes No

Weight at birth _____ kg Length at birth _____ cm

Was child breast-fed: Yes No If yes, duration _____

Failure to thrive: Yes No

Molecular/Enzyme Laboratory Test Results:

Mutations: Allele 1 _____ Allele 2 _____

Lab that determined mutations _____ Method (if known) _____

Which Gene: PCCA _____ PCCB _____

Enzyme activity _____ % of normal; Absolute Amount _____

Lab that determined enzyme activity _____

Are fibroblasts available? Yes No Unknown

If so, which laboratory _____

Family History:

Are there other family members with PA? Yes No

of siblings with PA _____ # living _____

Other affected relatives? Yes No Relationship: _____

Has the individual passed away? Yes No

If yes, age at death: _____ Date of death: _____

Clinical Evaluation:

Current age: _____ Current Weight: _____ kg Current Height: _____ cm

Respiratory:

Apnea: Yes No Unknown Tachypnea: Yes No Unknown

Cardiovascular:

Cardiomyopathy: Yes No ; mild moderate severe heart transplant

Unknown Age of diagnosis _____ Age of transplant _____

Long QT: Yes No Unknown ; Chronic Acute Age of diagnosis _____

Abdomen/Gastrointestinal:

Pancreatitis: Chronic Acute Never Noted

Kidney problems: Yes No Unknown

Liver transplant: Yes No Unknown

If yes, age at transplant _____ date of transplant _____

Port-a-cath: Currently in place Not currently in place Never had one

Gut motility: Slow Normal Requires medication

List medications _____

Reflux: Yes No Unknown

List medications _____

Eating by mouth: 100% ; 51-99% 1-50% 0%

Tube Fed: NG tube G-tube GJ-tube J-tube Other _____

Vomiting: Daily Once a week or more Once a month or more Infrequently

Are anti-emetics used for vomiting? Yes No Unknown

List medications: _____

Neurologic:

Basal ganglia damage: Yes No Unknown If yes, determined by MRI CT

Seizures: Yes No Unknown Type _____ Frequency _____

Autism Spectrum Disorder: Yes No Unknown

ADD/ADHD: Yes No Unknown

Optic Nerve Damage: Yes No Unknown Age at detection _____

Hematology/Immunologic:

Neutropenia: Chronic Acute Unknown Treatment? _____

List medications: _____

Anemia: Chronic Acute Unknown

Immune deficiency: Yes No Unknown Treated with IVIG: Yes No

Low Platelets: Chronic Acute Not Applicable

Asthma: Yes No Unknown

Skeletal:

Secondary Hip Dysplasia Yes No Unknown

Broken Bones: Yes No Unknown Type of fracture _____ Location _____

Short Stature: Yes No Unknown

Growth hormone treatment: Yes No Unknown

Osteoporosis: Yes No Unknown

Developmental Evaluation:

Walking: Not at all 25% of the time 50% 75% 100% of the time

Age when first walked? _____

Language: Age appropriate Slightly below age level

Significantly below age level Uses assistive technology (Device, PECS or sign)
No communication

Age when first talked? _____ (Years/Months)

Cognitive Ability: Age appropriate Mildly impaired

Moderately impaired Severely impaired

IQ: Known (give value or age level) _____ Untested/Unsure

Metabolic:

Is the individual biotin responsive? Yes No Unknown

Chronic hyperammonemia Yes No Unknown

Currently Elevated: < 2 times normal 2 times normal > 2 times normal

Is the individual currently on metabolic formula? Yes No Unknown

If yes, check all that apply: Propimex SHS XMTVI OA

Pro-phree PFD 80056 Duocal Polycose Vitaflo Other: _____

Is the individual taking levocarnitine? Yes No If yes, _____mg/kg.

Other supplements: Co-Q10 DHA Vitamin E B-6 Biotin Thiamin

Multi-vitamins Iron Other _____

Please add any other information you think may be relevant

If you have any questions please contact:

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