

Propionic Acidemia Questionnaire
Children's Memorial Hospital & The University of Chicago
Propionic Acidemia Foundation

The following questionnaire is part of a research study conducted through Children's Memorial Hospital and The University of Chicago, in Chicago, Illinois, with the help of the Propionic Acidemia Foundation (PAF), to gather clinical information regarding patients with propionic acidemia (PA). Please take a few minutes to fill out the following questionnaire to the best of your knowledge for each patient affected with PA. The information will help us improve our understanding of the symptoms, natural history, and complications of propionic acidemia. Leave blank any information that you do not understand or for which you have no information, and feel free to consult with your geneticist regarding any conditions that you are not sure may have occurred. You may attach additional sheets with formula or medication information if there is not enough space. Your answers will be kept confidential, and any personal identifiers, such as name and date of birth, will be maintained separately from this questionnaire at a secure location. Please contact the PAF Board of Directors (Jill Franks, Janice Boecker, Brittany Smith, or Jennifer Mouat) or Loren Pena at Children's Memorial Hospital if you have any questions.

The completed questionnaire should be returned electronically or by regular mail to Loren Pena at the email or postal address included at the end of the survey. Thank you for your participation.

PA reference # _____

Subject Profile:

Name: _____ Date of Birth: _____
Race: Caucasian Black Asian East Indian Mixed Race / Other
Are you Hispanic? Yes No
Country of birth: _____
Gender: Male Female
Age at patient's diagnosis of PA: _____
Is this patient deceased? Yes No

Newborn information:

Was this patient detected through the newborn screen? Yes No
If yes, was the result available prior to the initial hospitalization? Yes No
Weight at birth: kg / lb Length at birth: cm / inches
Was the patient hospitalized within a month after birth? Yes No
If yes, how was the patient treated? Intravenous fluids
Intravenous carnitine Dialysis Other (please specify)

Molecular / Biochemical laboratory results:

Mutated gene: PCCA PCCB
Mutations: 1. _____ 2. _____
Level of enzyme activity: _____ % of normal Not known

Family History:

Are there other family members affected with PA? Yes No
If yes, number of siblings with PA: _____ Number alive: _____
Are there any other affected relatives? Yes No
If yes, relationship to patient: _____
Is this relative alive? Yes No
If no, age at death: _____

Clinical Information regarding patient with PA:

Current age (if living): Current weight (if living): kg/lb
Current height (if living): cm/inches
Number of lifetime hospitalizations:
Age at first hospitalization:
Current treatment:
Grams of daily protein from regular food:
Grams of daily protein from medical formula:
Name of medical formula:
Recipe used:
Carnitine dose per day:
Is the patient taking biotin supplements? Yes No
If yes, daily dose: mg
Does the patient take any other supplements or medications? Yes No
If yes, please specify the name and dose of supplements or medications:

Does the patient have any other health conditions in addition to PA? Yes No
If yes, please specify:

Cardiovascular symptoms:

Enlargement or dysfunction of the heart (i.e., cardiomyopathy) Yes No Unknown
If yes, mild moderate severe Heart transplant
Age at diagnosis: Age at transplant:
Abnormal heart rhythm (i.e. arrhythmia or long QT): Yes No Unknown
If yes, age at diagnosis:
Other heart problems: Yes No Unknown
Please specify the type of heart problem:

Gastrointestinal symptoms:

Pancreatitis: Yes No Unknown
If yes, how many episodes:
Liver transplant: Yes No Unknown
If yes, age at transplant:
Does the patient eat by mouth? Yes No
If yes, 100% of total intake 50% 25% Less than 25%
If no, does the patient have a G-, G-J, or NG tube? Yes No
Age at placement of tube:
Type of tube:
Does the patient receive any other type of nutrition? Yes No
Please specify type:

Neurologic symptoms:

Seizures: Yes No Unknown
If yes: Type Frequency
Age at diagnosis:
Metabolic stroke: Yes No Unknown
If yes, please specify age:
Optic nerve damage: Yes No Unknown
If yes, age at diagnosis:

Basal ganglia damage: Yes No Unknown
 If yes, age at diagnosis:
 Movement disorder: Yes No Unknown
 If yes, age at diagnosis:
 Autism Spectrum Disorder: Yes No Unknown
 If yes age at diagnosis:
 Attention Deficit Disorder or Attention Deficit and Hyperactivity Disorder (i.e., ADD/ADHD):
 Yes No Unknown
 If yes age at diagnosis:

Hematologic symptoms:

Low white blood cell counts (neutropenia): Yes No Unknown
 If yes, age at diagnosis: Treatment:
 Low red blood cell counts (anemia): Yes No Unknown
 If yes, age at diagnosis: Treatment:
 Low platelet counts (thrombocytopenia): Yes No Unknown
 If yes, age at diagnosis: Treatment:
 Immune deficiency: Yes No Unknown
 If yes, age at diagnosis: Treatment:

Other symptoms:

Decreased bone density (osteopenia/osteoporosis): Yes No Unknown
 If yes, please specify age at diagnosis:
 Broken bones (fractures): Yes No Unknown
 If yes, please specify age at which fractures occurred:
 Blisters and unusual rashes: Yes No Unknown
 If yes, please specify age at diagnosis and describe:
 Treatment:
 Hair loss: Yes No Unknown

Developmental information:

Cognitive ability (i.e., ability to understand and perform relative to the patient's age):
 Age-appropriate Mildly impaired
 Moderately impaired Severely impaired
 IQ (if known, please give value or age level):
 Language skills: Age-appropriate Mildly impaired
 Moderately impaired Severely impaired
 Uses assistive technology No communication
 If applicable, please specify the patient's:
 Age at first word:
 Age speaking with full sentences:
 Gross motor skill (i.e., ability to perform tasks such as sitting, walking, climbing stairs):
 Age-appropriate Mildly impaired
 Moderately impaired Severely impaired
 If applicable, please specify the patient's:
 Age when patient sat unassisted:
 Age when patient walked unassisted:
 Fine motor skills (i.e. ability to perform tasks such as holding a pencil with two fingers, buttoning a shirt):
 Age-appropriate Mildly impaired

Moderately impaired Severely impaired

If applicable, please specify the patient's:

Age when patient was able to feed self with a spoon:

Age when patient was able to use a crayon or pencil:

Activities of daily living:

Is the patient toilet trained? Yes No

If yes, age that toilet training was achieved:

Is the patient able to dress self? Yes No

If yes, age that patient was able to dress self:

Can the patient brush his/her teeth? Yes No

If yes, please specify at what age:

Does the patient attend school? Yes No

If yes, please specify mainstream or special education classes

Grade level:

Deceased patients:

Age at death:

Cause of death:

Can we contact you if we need more information? If so, please indicate the name of the person filling out this questionnaire and a telephone number where we can reach you:

Please feel free to add any other information that may be relevant:

Please return the completed questionnaire by email or regular mail to Loren Pena at the following email or postal address.

Children's Memorial Hospital

Division of Genetics, Birth Defects, and Metabolism

2300 Children's Plaza, Box 59

Chicago IL 60614

lpna1@bsd.uchicago.edu

If you have any questions, please contact:

Board of Directors
Propionic Acidemia Foundation
1963 McCraren Rd.
Highland Park IL 60035
877-720-2192
paf@pafoundation.com

Loren Pena, MD, PhD
Division of Genetics, Birth Defects, and Metabolism
2300 Children's Plaza, Box 59
Chicago IL 60614
773-880-4462
lpna1@bsd.uchicago.edu