

Propionic Acidemia Questionnaire
The University of Illinois at Chicago and Children's Memorial Hospital
Propionic Acidemia Foundation

The following questionnaire is part of a research study conducted through the University of Illinois at Chicago and Children's Memorial Hospital, in Chicago, Illinois, with the help of the Propionic Acidemia Foundation (PAF), to gather clinical information regarding patients with propionic acidemia (PA). This study has been reviewed and approved for enrollment by the Investigational Review Board (IRB) at both institutions. Please take a few minutes to fill out the following questionnaire to the best of your knowledge for yourself if you are affected with PA and live independently. If you are a parent or guardian of a person with PA, please fill out the survey providing information for the affected person. 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. The investigators will make every attempt to keep your information confidential and both the surveys and any personal identifiers, such as name, date of birth, and contact information, will be maintained separately from this questionnaire at a secure location by the investigators. Only the study investigators (Dr. Pena and Dr. Burton) will have access to the completed questionnaires. Please contact Jill Franks from the PAF or Loren Pena at the University of Illinois at Chicago 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
Is the patient 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 Not known
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 (include measures of each ingredient, if known):

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:

Has an echocardiogram been done? Yes No Unknown

If yes, please specify age at which last echo was done:

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:

Has an EKG (electrocardiogram) been done? Yes No Unknown

If yes, please specify age at which last echo was done:

Abnormal heart rhythm (i.e. arrhythmia or long QT): Yes No Unknown

If yes, please specify the type of abnormal rhythm and 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, # of episodes and age at each: Treatment (please specify if one time or ongoing):

Did it resolve? Yes No Unknown

Low red blood cell counts (anemia): Yes No Unknown

If yes, # of episodes and age at each: Treatment (please specify if one time or ongoing):

Did it resolve? Yes No Unknown

Low platelet counts (thrombocytopenia): Yes No Unknown

If yes, # of episodes and age at each: Treatment (please specify if one time or ongoing):

Did it resolve? Yes No Unknown

Immune deficiency: Yes No Unknown

If yes, # of episodes and age at each: Treatment (please specify if one time or ongoing):

Did it resolve? Yes No Unknown

Other symptoms:

Decreased bone density (osteopenia/osteoporosis): Yes No Unknown

If yes, please specify age at diagnosis: Treatment:

Broken bones (fractures): Yes No Unknown

If yes, please specify age and circumstance when each fracture 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.

**Loren Pena, MD, PhD
Department of Pediatrics, Division of Genetics (MC 856)
University of Illinois at Chicago
840 S. Wood Street, 12th floor
Chicago IL 60612
lpena@uic.edu**

If you have any questions, please contact:

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paf@pafoundation.com

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Addendum: Please complete this portion *if you previously completed a survey*. These are additional questions that will clarify previous answers. You do not need to complete this portion if you are participating after July 1, 2009 – these questions have been incorporated to the current survey. Please submit the answers to the email or postal address for Loren Pena listed on page 4 of this document. Thank you for your time.

PA reference # _____

Name: _____ Date of Birth: _____

Cardiovascular:

Have you had an EKG? Yes No Unknown

If yes, please indicate age at which last EKG was performed:

Have you had an echocardiogram? Yes No Unknown

If yes, please indicate age at which last EKG was performed:

Hematologic:

If you previously answered yes to any of the questions below, please indicate if this is an episodic or chronic problem and specify whether the patient received treatment each time the problem presented or if treatment is ongoing (ie, chronic).

Low white blood cell counts (neutropenia):

If yes, # of episodes and age at each:

Did it resolve? Yes No

Treatment (please specify if one time or ongoing):

Unknown

Low red blood cell counts (anemia):

If yes, # of episodes and age at each:

Did it resolve? Yes No

Treatment (please specify if one time or ongoing):

Unknown

Low platelet counts (thrombocytopenia):

If yes, # of episodes and age at each:

Did it resolve? Yes No

Treatment (please specify if one time or ongoing):

Unknown

Immune deficiency:

If yes, # of episodes and age at each:

Did it resolve? Yes No

Treatment (please specify if one time or ongoing):

Unknown

Other symptoms:

If you have had fractures, please indicate the age and circumstance in which each fracture occurred: