Natural History, Physiology, Microbiome and Biochemistry Studies of Propionic Acidemia

NCT02890342

Status	RECRUITING
Sponsor	National Human Genome Research Institute (NHGRI)
Enrollment	1,045 participants

Key Eligibility Criteria

Inclusion (6)

• Patients 2 years of age or older, of any gender and ethnicity, with propionic acidemia are eligible to enroll in this protocol. Patients diagnosis will be confirmed based on biochemical and/or molecular and enzymatic testing. Participants of any gender and ethnicity over 1 month of age are eligible to enroll remotely for collection of outside records and natural history data. They will be eligible to enroll in the full study for in-person evaluation at 2 years of age.

• Unaffected family members over 1 month of age, of any ethnicity or race, may be included in the study as household controls for microbiome studies and/or for genetic analysis. Studies in unaffected family members may include collection of medical and family history; if necessary completion of physical examination; drawing of blood for research purposes include testing of DNA; collection of stool samples for microbiome studies; collection of dietary history using pen-and-paper or electronic food diary and questionnaires; collection of saliva for metabolite and DNA analysis. In some unaffected family members without a known familial cause of propionic acidemia, exome sequencing or genome sequencing could be performed. Unaffected family members will not receive direct benefit from taking part in the study.

• If a participant becomes pregnant while on study, the participant can remain on study. The only way to learn more about the critical biological differences in those who affected with propionic acidemia who are pregnant is to continue to follow pregnant women on study.

• However, no tests or procedures that are greater then minimal risk will be performed. Affected subjects who are pregnant may undergo procedures as part of their clinical care, including blood draws, genetic studies, and consultations, according to the clinical judgement of the clinical team. However, pregnant participants will be excluded from procedures such as organ tissue collection, stable isotope studies, GFR testing, and brain or cardiac MRI until the pregnancy is concluded.

• Healthy volunteers may be eligible to participate in the study if they are between 12 - 40 years of age, must meet specific BMI criteria (similar to affected individuals studied).

... and 1 more (see full listing online)

Exclusion (2)

• A subset of participants may be enrolled in the tissue collection part of the study only (i.e. if they are too sick to travel). We can may also arrange limited remote consultation with our research team and NIH consultants, the participants referring physician and the participant/their legal guardian through the telephone or an NIH supported telehealth platform for participants who are unable to safely travel to NIH. This would not replace a study visit but would be used when travel isn t possible due to extenuating circumstances (e.g. pandemic). Participants would be encouraged to follow-up for a more thorough in-person evaluation when they are able to travel to NIH.

• For the healthy volunteers, they will be excluded if they have halitosis, cavities, dental or gingival problems, respiratory diseases (for example, asthma or recent history of COVID19), use tobacco products (for example, cigarette smoking or chewing tobacco), or use electronic nicotine delivery systems (for example, use of e-cigarettes or vaping devices), as this may interfere with accurate measurement of their volatile organic compounds. NIH staff and their family members will be eligible to participate in the healthy volunteer portion of the study.

Locations (2 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States
Children's Hospital of Pittsburgh, Pittsburgh, Pennsylvania, United States