

Survey of Health Status and Complications Among Propionic Acidemia Patients

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Propionic acidemia (PA) is a rare organic acidemia that is due to deficiency in the enzyme propionyl-coA carboxylase. Complications are currently described mostly in the form of case reports. We sampled a population of affected individuals in order to estimate the frequency of complications amongst the sample. The study is a cross-sectional retrospective review with a survey instrument and recruitment through the Propionic Acidemia Foundation. Responses for 58 individuals were tabulated for each question as how frequently the complication was reported among responders. Commonly reported findings included seizures, arrhythmia, leucopenia, and anemia. Developmental and cognitive disabilities were reported in the majority of individuals. Heart failure or cardiomyopathy was reported in over half of deceased individuals at time of death. Pancreatitis was reported in a minority of the sample, yet more than half of these reported a recurrence. These results update and extend our current knowledge of recognized complications among individuals with PA. The results also provide new information regarding developmental outcomes and previously unreported morbidity from cardiac and gastrointestinal complications. Longitudinal studies exploring associated biochemical and clinical parameters are necessary to further our understanding of the pathophysiology of PA and its complications. © 2012 Wiley Periodicals, Inc.

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