

Cardiac Manifestations in Propionic Acidemia

Saleem Z., Bhole V., Chakrapani A., Timmins J., Grossman S., Chikermane A., Birmingham Children's Hospital, Birmingham, UK

Introduction: Propionic acidaemia (PA) is an autosomal recessive disease that results from deficiency of propionyl-CoA carboxylase (PCC). In the majority of reported cases, the phenotype includes metabolic acidosis and/or neurological deficits. Our aim was to evaluate the relationship between propionic acidemia (PA) and cardiac involvement, which has been identified in literature.

Materials and Method: 12 year retrospective analysis of data in a single centre of patients with PA; electrocardiographs (ECG) and echocardiograms of all patients with PA analysed looking for patterns of presentation and progression of cardiac disease.

Results: 11 patients with confirmed diagnosis of PA. 10 had ECGs, all had echocardiograms. In our cohort, all the patients were of Asian origin, and there were 4 girls, and 7 boys. 2 children died, one following an attempted septal ablation for hypertrophic obstructive cardiomyopathy, and the other following a liver transplant. ECG analysis was undertaken on 10 patients as one child did not have an ECG. Of these, 4 had a normal corrected QT interval (QTc); 4 others had a normal ECG to start with but progressed to have QT prolongation, while the other two had prolonged QTs on their first ECG. In two of the children, we identified intermittent QT prolongation on the 24-hour holter when their baseline 12-lead ECG was normal to start with. On echocardiography, one child had a small muscular VSD, while two others had small PFOs. All of them had a normal global systolic function with a normal fractional shortening; two had hypertrophic cardiomyopathy of whom in one, the pathology was present at the first cardiac assessment and in the other, it developed at 3 years of age. Both the children who died had LV hypertrophy.

Conclusion: We have shown that there can be progression of ventricular repolarisation abnormalities and cardiac function. It is also important to undertake regular holter monitoring as in some cases the QT prolongation is intermittent.