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**Year in Course**: 3rd (Senior)

**Topic**: Biomedical Sciences, Biotechnology, Genetics

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**Title**: The Feasibility of Improving Positive Predictive Values for MCAD Deficiency through the Use of Additional Acetyl-carnitine Markers

**Abstract**

Fatty-Acid Oxidation disorders (FOD) are a subcategory of metabolic disorders that affect the functionality of fatty acid transport and mitochondrial β-oxidation (4). Complications due to fatty-acid oxidation disorders cause individuals to go into a metabolic crisis. This often requires hospitalization and emergency medical treatment. To prevent these crises, a screening program was implemented in 2002 in order to serve as an early detection system for these rare genetic disorders (1). This screening program identifies infants who are at risk of having a rare genetic disorder. Within the last five years an increase in false-positive results have been recorded by David Axelrod Institute, the center for the newborn screening program in New York. This study addresses this sudden increase in false-positive through a review of current procedures conducted and the analysis of past samples in order to determine if a secondary biomarker could be identified. Samples analyzed were from archival data preserved by New York State’s Department of Health Division of Genetics at David Axelrod Institute. Results showed a statistically significant correlation, with an r-value of 0.887, between the levels of carbon fourteen (C14) and 3-hydroxypalmitoyl carnitine (C16:1-OH) in the blood of infants who were afflicted with glutaric-acidemia type two. Additionally the carbon eight (C8) biomarker was found to not be significantly correlated to the levels of C14 or C16:1-OH. These additional biomarkers are crucial in the differentiation of diagnosis between medium-chain acyl-CoA dehydrogenase deficiency and multiple acyl-CoA dehydrogenase deficiencies, which significantly decreases the risk of sudden unexpected death in infancy.