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Inherited Metabolic Diseases

153. DETERMINATION OF PROPIONYL CoA CARBOXYLASE AND ITS APPLICATION ON PRENATAL DIAGNOSIS OF PROPIONIC ACIDEMIA

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Propionic acidemia (PA) is an inherited autosomal recessive life threatening disease which is caused by the deficiency of propionyl CoA carboxylase (PCC). The PCC activity could be determined in cultured cells by $^{14}CO_2$-fixation method; ie, the incorporation of $NaH^{14}CO_3$ in the presence of propionyl CoA into nonvolatile ^{14}C-labeled products at alkaline condition, and the removal of unreacted residual $NaH^{14}CO_3$ as $^{14}CO_2$ by acdification and drying on filter paper. The remained redioactivity was determined by liquid scintilation counter. A pregnacy at risk for PA has been monitored by assaying the PCC activity in cultured amniotic cells by this method. The PCC activity of the cultured amniotic cells (574 pmol/min-mg protein) is similar to that of normal control (274-765 pmol/min-mg protein, n=4). The healthy male infant was born at term. After five months of birth, the diagnosis was confirmed by postnatal assaying of PCC in cultured fibroblast. The PCC activity in the cultured fibroblast of the infant and normal control were 654 and 505-600 (n=3) pmol/min-mg protein, respectively, which were about 100 times of the PA cell line GM0371 (from NIGMS Human Genetic Mutant Cell Repository, USA). The results indicated that the $^{14}CO_2$-fixation method permited analysis of PCC in cultured cell and could be applied to prenatal diagnosis of PA in our laboratory.