20-114 AMINO ACID ANALYSIS AND URINARY ABNORMAL ORGANIC ACIDS IN PATIENTS WITH MAPLE SYRUP URINE DISEASE . K.J.Hsiao, S.H.Sheu*, S.J.Wu, M.S.Chen, S.H.Chiang, T.T.Chang, Y.C.Chung[1], C.H.Lin[2].
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Maple syrup urine disease (MSUD) is an autosomal recessive inherited disease, caused by branched-chain oxo-acid dehydrogenase deficiency. Symptoms consist of refusal of feeding, vomiting, muscular hypertonicity, respiratory difficulties, mental retardation, coma and metabolic acidosis. A 3 years old female and a 2 months old male patients were recently detected by us in Taiwan. Their urinary 2,4-Dinitrophenylhydrazine screening test were positive. The plasma concentration of the leucine (1124-1361 umol/L), isoleucine (329-606 umol/L), valine (593-631 umol/L), are 4 times higher than the normal ranges. The urinary amino acids (Leu:1015-6391 umol/g creatinine, Ile:226-1679 umol/g creatinine, Val:335-4355 umol/g creatinine) are also higher than the normal. A lot of abnormal organic acids accumulated from the metabolism of the branched-chain amino acids, were also identified in their urine by Gas Chromatography-Mass Spectroscopy. The nature of the mutation of these patients remains to be illustrated.

20-115 PRENATAL DIAGNOSIS OF PROPIONIC ACIDEMIA WITH AMNIOCYTES.
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Propionic acidemia (PA) is an inherited autosomal recessive life threatening disease caused by a deficiency of Propionyl CoA Carboxylase (PCC) activity. Prenatal diagnosis of PA were performed by two methods in our laboratory: 1). Incorporation of radiolabel from propionate-1-$^{14}C$ into protein (trichloroacetic acid, insoluble cell material) in amniotic cells for detecting defects in propionate metabolism, and 2). determination of PCC activity in cultured cell by $^{14}CO_2$-fixation method. A patient with a PA proband child who requested genetic counseling and prenatal diagnosis for third pregnancy. The PCC activity (28 pmol/min/mg protein) and propionate incorporation (34 pmol/hr/mg protein) of cultured amniotic cell were much lower than the reference ranges of 340-570 pmol/min/mg protein (n=5) and 180-390 pmol/hr/mg protein (n=20), respectively, and were similar to the results (PCC 7 pmol/min/mg protein; propionate incorporation 16 pmol/hr/mg protein) of the PA cell line GM0371 (from NIGMS Human Genetic Mutant Cell Repository, USA). After counseling, the pregnancy was terminated at 21 weeks of gestation as requested by the family. Both propionate incorporation and PCC activity in the fibroblast cultured from the abortus confirmed that the fetus was affected with PA as prenatally diagnosis.