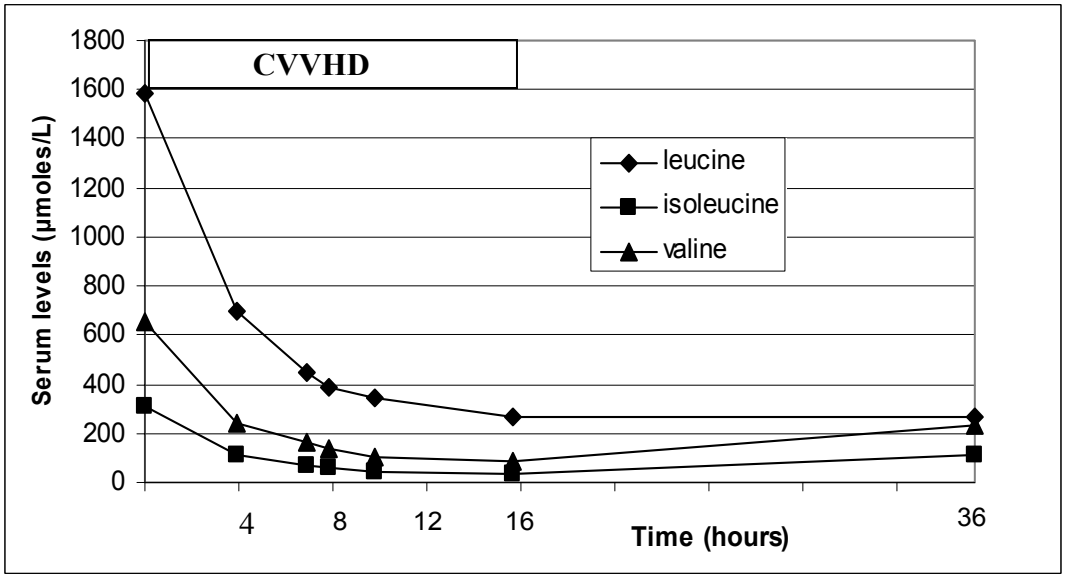
Continuous Veno-venous Hemodiafiltration Therapy for Acute Decompensation with Cerebral Edema in Maple Syrup Urine Disease

JJ Zaritsky [1], JA Martinez [1], OYadin [1]

[1]David Geffen School of Medicine, Mattel Childrens Hospital, UCLA, Los Angeles, United States

Maple syrup urine disease (MSUD) is caused by an inheritable deficiency in 2-ketoacid dehydrogenase and results in the systemic accumulation of branched chained amino acids (BCAAs) and their metabolites. During episodes of metabolic decompensation characterized by extremely high serum levels of BCAAs, affected children have severe neurological deterioration often accompanied by life-threatening cerebral edema. Because endogenous renal clearance of BCAAs is limited, therapy has focused on the rapid extracorporeal removal of BCAAs often via intermittent hemodialysis (IHD). However the rapid diffusive clearance of IHD may lead to solute disequilibrium potentially worsening cerebral edema. In this report, a symptomatic 7yr old male with MSUD who developed cerebral edema underwent CVVHDF with a small molecule clearance rate of 37.5ml/min/m2. This approach resulted in a nearly 50% decrease in BCAA levels within the first 4 hours of treatment. Therapy was discontinued at 16 hours due to the rapid improvement in neurological status. At that time, serum leucine levels had fallen from 1581 to 268Œºmoles/L, isoleucine levels had fallen from 313 to 110Œºmoles/L and valine levels had fallen from 652 to 83Œºmoles/L. Leucine, isoleucine and valine clearance rates measured during the last four hours of therapy were 30.4, 30.9 and 30.7ml/min respectively. This case demonstrates that CVVHDF is an effective and safe therapy of severe acute decompensation in MSUD accompanied by cerebral edema.



Corresponding author:
JJ Zaritsky
UCLA, Pediatric Nephrology
Mattel Childrens Hospital
A2-383 MDCC
10833 LeConte Ave
90095 Los Angeles
CA, United States
jzaritsky@mednet.ucla.edu
Phone: 310-206-6987