Pipecolic acid as a diagnostic marker of pyridoxine-dependent epilepsy.

Abstract:

Pyridoxine-dependent epilepsy, although described some decades ago, may still be an underdiagnosed disorder. We have recently described isolated pipecolic acid elevations in the plasma and/or CSF of three patients with pyridoxine-dependent epilepsy with an intriguing inverse correlation to the oral intake of pyridoxine. We have now confirmed these findings in a further 6 unrelated patients with pyridoxine-dependent epilepsy. Pipecolic acid in plasma was 4.3- to 15.3 fold elevated compared to the upper normal range before pyridoxine and remained in the mildly elevated range while on pyridoxine. Pipecolic acid was even more markedly elevated in CSF. The extent of pipecolic acid elevation in CSF exceeded that of plasma by a factor of 2.2 to 4.8. This clearly discriminates pyridoxine-dependent epilepsy from other possible defects with elevated pipecolic acid. Determination of pipecolic acid in plasma and/or CSF should be included in the diagnostic work-up of patients with therapy-resistant seizures. It will in addition prevent patients with pyridoxine-dependent epilepsy from experiencing potentially dangerous pyridoxine-withdrawal, which until now has been necessary to prove the diagnosis.