Hitoshi Nakajima, J Gastrointest Dig Syst 2018, Volume 8
DOI: 10.4172/2161-069X-C6-080

Antepartum ornithine transcarbamylase deficiency: A case report

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Ornithine transcarbamylase deficiency (OTCD) is the most common type urea cycle enzyme deficiencies. This syndrome results from a deficiency of the mitochondrial enzyme ornithine transcarbamylase, which catalyzes the conversion of ornithine and carbamoyl phosphate to citrulline. Our case was a 28-year-old female diagnosed with OTCD following neurocognitive deficit during her first pregnancy. Although hyperammonemia was suspected as the cause of the patient's mental change, there was no evidence of chronic liver disease. Plasma amino acid and urine organic acid analysis revealed OTCD. After combined modality treatment with arginine, sodium benzoate and hemodialysis, the patient's plasma ammonia level stabilized and her mental status returned to normal. At last, she recovered without any damage remained.

Biography

Hitoshi N has completed his PhD at the age of 29 years from Hirosaki University and postdoctoral studies from Hirosaki University School of Medicine. He is now the Professor of General Medicine Toho University, Toho Medical Center Omori Hospital Faculty of Medicine Toho University. He has published more than 10 papers in reputed journals and has been serving as a society councilor member of reputed gastroenterology related societies.

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