## عنوان مقاله:

Maple Syrup Urine Disease Induced Grand Mal Seizures: A Case Report


## محل انتشار:

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## خلاصه مقاله:

Background Maple Syrup Urine Disease (MSUD) is a rare autosomal recessive metabolic error, characterized by Branched Chain α-Keto-acid Dehydrogenase Complex (BCKDC) deficiency. Mutations in 3 genes can lead to abnormal metabolism and accumulation of leucine, isoleucine, valine and corresponding keto-acids. MSUD affects 1 in 185,000 infants globally. Seizure is a common presentation among neonates. However, in intermediate MSUD, seizures have a delayed and insidious onset, along with developmental Case Report We report a case of grand mal seizures in a patient with intermediate MSUD, presenting with multiple episodes of seizure, dystonia, spastic quadriplegia, involuntary micturition and oculogyric crisis.Seizures were managed successfully with intravenous lorazepam and other supportive measures. The patient was advised to strictly adhere to branched chain amino acid restricted diet. Conclusion This case report emphasizes on the importance of medication adherence and dietary .restrictions to prevent permanent psychomotor damage or death



## کلمات کلیدی:

GTCS, Genetic, MSUD, Seizures, Quadriplegia