THE KOREAN SOCIETY OF NEPHROLOGY
APCN & KSN 2024
June 13(Thu) - 16(Sun), 2024 Coex, Seoul, Korea
Promoting Sustainable Kidney Health: The Asia-Pacific and Beyond

Abstract Submission No.: A-0419

Gitelman-like Syndrome Manifested by Dysarthria and Dystonia in Early Childhood: A Case Report

Hyunseung Noh, Jinwoon Joung, Heeyeon Cho
Department of Pediatrics-Nephrology, Samsung Medical Center, Korea, Republic of

Case Study : Gitelman syndrome is a hereditary salt-losing tubulopathy with SLC12A3 mutation, characterized by hypokalemic alkalosis and hypomagnesemia. However, about 10% of Gitelman syndrome remain genetically unsolved. We report a pediatric case of Gitelman-like syndrome caused by pathogenic variants in mitochondrial DNA manifesting neurologic symptoms. A 9-year-old boy presented with dysarthria and dystonia of neck and arms. The neurologic symptoms were first observed at the age of 6 years after a febrile event with diarrhea and resolved gradually through a year. Yet, the symptoms relapsed with another febrile event at the age of 9 year and he was referred to this center. His height and weight were inadequate for the age. No significant findings were noted from neurologic evaluations including magnetic resonance imaging (MRI) of brain and electromyography. Whole genome sequencing (WGS) revealed a pathogenic variant of mitochondrial tRNA for phenylalanine (MT-TF m.591C>T) gene which causes Gitelman-like syndrome. Sequentially, hypokalemic alkalosis with hypomagnesemia was detected so that potassium and magnesium was supplemented orally. His sister with a history of dystonia was additionally diagnosed as Gitelman-like syndrome and the maternal inheritance was confirmed with pedigree analysis. Pathogenic variants in MT-TF can cause Gitelman-like syndrome and particularly the m.591C>T and m.4291T>C variants are the hotspot mutations. The clinical phenotypes include hypomagnesemia-related neurologic symptoms and hypokalemic metabolic alkalosis which are similar to that of Gitelman syndrome. The pathologic findings demonstrate tubulointerstitial kidney disease with abnormal mitochondria in distal tubule. Different from Gitelman syndrome caused by SLC12A3 mutations, MT-TF mutations are known to be associated with the development of chronic kidney disease (CKD). Clinicians should be aware that the unexplained neurological deficit can be the first presenting symptoms of Gitelman-like syndrome and the screening of electrolyte imbalance is recommended for such patients.