allied academies

International Conference on

Health Care and Neuroscience

April 08-09, 2019 | Zurich, Switzerland

Progressive cavitating Leukoencephalopathy in an infant baby: A case report

Rayya A Almarwani
King Faisal Specialist Hospital and Research Center, Saudi Arabia

Cavitating leukoencephalopathy is a group of disorders caused by multiple mitochondria dysfunction syndrome (MMDS). Multiple genetic mutations have been identified in its etiology including a mutation in the IBA57 gene on chromosome 1q42. We present the case of a 5-month-old boy who was diagnosed with progressive cavitating leukoencephalopathy at King Faisal specialist Hospital, Riyadh city, Saudi Arabia. The patient presented with frequent excessive crying since birth. He also had a history of abnormal movement described as tonic spasm of the upper limbs for few second, delayed developmental milestones, and regression of the achieved milestones. Neurological examination of the patient was positive for horizontal eye nystagmus and optic nerve atrophy, exaggerated gag response, spastic tone that was more prevalent in the upper limbs, and bilateral clonus. The patient showed abnormal findings on MRI including cavitation and cystic leukodystrophy, and homozygosity mapping showed the IBA57 gene mutation. The patient died at the age of 7 months from progressive respiratory failure. MMDS is a rare condition, and according to available data, this case of MMDS is the first to be reported in Saudi Arabia and in the Gulf region.

Speaker Biography

Rayya A Almarwani is senior pediatric neurology resident at King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia. She has completed her medical school 2014 from Faculty of Medicine, Tabuk University.

e: rayya.aljohani@gmail.com