Hypoparathyroidism in Aicardi-Goutières syndrome (AGS)— A Descriptive Case Report

Abdulhafiz Hakmi, M.D., Nawal Al Mohamad, M.D.

Abstract: Aicardi-Goutières syndrome (AGS) is a genetically inherited autosomal recessive disorder characterized by progressive encephalopathy, predominant basal ganglia calcifications, and elevated cerebrospinal fluid interferon-alpha (IFN-α) levels. The clinical manifestations often include seizures, cognitive impairment, and generalized dystonia. While the prevalence of AGS remains uncertain, pathogenic variants linked with AGS have been detected in affected individuals from various ethnic backgrounds. AGS, recognized as a monogenic hereditary disorder, is divided into nine distinct types based on specific pathogenic genes, with SAMHD1 commonly associated with milder phenotypes. The disease presentation can be challenging due to its variability across stages, making it easily mistaken for other conditions such as congenital TORCH infection or metabolic disorders. Endocrinopathies in AGS are acknowledged but not extensively described. This report discusses a unique case of a SAMHD1 variant causing AGS in an 18-year-old female college student, presenting with recurrent hypocalcemia secondary to hypoparathyroidism. The patient experienced various neurological and non-neurological symptoms over time, highlighting the complexity of AGS diagnosis. Imaging findings, especially through Computed Tomography, play a crucial role in identifying characteristic brain calcifications specific to AGS. This case underscores the importance of ongoing research to better understand the correlation between AGS and hypoparathyroidism, and the necessity for more defined management guidelines in this context.