P-111 - CASE SERIES OF PEDIATRIC PATIENTS WITH DEFICIT OF CEREBRAL CREATINE SECONDARY TO GUANIDINOACETATE METHYLTRANSFERASE (GAMT) DEFICIENCY IN ANTIOQUIA AND SANTANDER, COLOMBIA

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INTRODUCTION: Cerebral creatine deficiency is an inborn error of metabolism that may be secondary to L-arginine glycine amidotransferase (AGAT) deficiency, guanidinoacetate methyltransferase (GAMT) deficiency, and the creatine transporter (CRTR) deficiency, the latter one being the most common of the three. There are few reported cases in literature around the world, by 2015 less than 20 cases of AGAT deficiency had been reported. OBJECTIVE: Describe the phenotypic and genotypic findings in two patients with refractory epilepsy and autism spectrum disorder, in which creatine deficiency was suspected and then confirmed. MATERIALS AND METHODS: Describe two cases of confirmed cerebral creatine deficiency in Colombian population. RESULTS: Two pediatric patients form different geographic regions of Colombia presented a history of refractory epilepsy and autism spectrum disorder. Innate errors of metabolism were suspected on both patients and finally the diagnosis of cerebral creatine deficiency was made by molecular demonstration of mutations on the guanidinoacetate methyltransferase (GAMT) gene. One of the patients had a brain nuclear magnetic resonance with spectroscopy that showed absence of creatine peak in the basal nuclei and semioual centers. Both patients are currently being treated with nutritional restriction of proteins of animal origin and nutritional supplement with monohydrate creatine and L-ornithine. CONCLUSION: Cerebral creatine deficiency is a rare condition that should be suspected in children with developmental delay, hypotonia, drug resistant epilepsy and behavioral problems. Although the most common cause of this condition is the creatine transporter deficiency, all three causes should be studied when this entity is suspected. Brain magnetic resonance with spectroscopy is of great help when this diagnosis is suspected.