P-103 - DYSTONIA TREATMENT IN PATIENTS WITH PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (PKAN)

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BACKGROUND: Pantothenate kinase-associated neurodegeneration (PKAN) is a type of neurodegeneration with brain iron accumulation (NBIA). The phenotypic spectrum of PKAN includes classic PKAN and atypical PKAN. Classic PKAN is characterized by early childhood onset of progressive dystonia, dysarthria, rigidity, and choreoathetosis. Pigmentary retinal degeneration is common. Atypical PKAN is characterized by later onset (age >10 years) and more gradual progression of disease. Treatment for NBIA disorders remains symptomatic. OBJECTIVE: To describe clinical response of dystonia in PKAN patients treated with pharmacological and surgical treatment. METHODS: Retrospective descriptive study, analysis of clinical records and revision of videos of pediatric patients with PKAN. RESULTS: 11 genetically confirmed patients (8 males / 3 females). 6 presented typical presentation and 5 atypical PKAN. Average age of onset 6 years. The initial symptoms were gait disorders associated with frequent falls (7/11) and focal dystonias (4/11). Dystonia with prominent cranial involvement and cognitive impairment were present in all patients. 7 patients showed spasticity. The eyes of the tiger sign T2-weighted MRI was seen in 10/11 patients. Symptomatic treatment for dystonia included oral treatment with trihexyphenidyl, benzodiazepines, baclofen, tetrabenazine and levodopa showed mild or no response. Botulinum Toxin-A Injection was used for focal dystonia with transient response. Two patients were treated with deep brain stimulation (DBS), both with good response, one patient recovered gait and one patient was treated as management of dystonic storm with significant reduce of dystonia. No adverse effects were reported. CONCLUSIONS: In our serie of PKAN patients, poor response to pharmacological dystonia therapy were observed. Surgical treatment with DBS showed significant reduction of dystonic symptoms, improving quality of life in our patients.