PS2.026

West syndrome - a diagnosis is not everything

Daniel Carquejo(1), A Alves(2), E Fernandes(1), C Jorge(2), F Ribeiro(1), R Magalhães(1)
(1) ULS Nordeste, UCSP Mirandela 1, Mirandela, Portugal
(2) ULS Nordeste, UCSP Sé, Bragança, Portugal

Corresponding author: Dr Daniel Carquejo, ULS Nordeste, UCSP Mirandela 1, Vila Real, Portugal. E-mail: nicarquejo@gmail.com

Background: West syndrome or infantile spasms syndrome refers to a severe infantile epileptic encephalopathy, whose etiology often remains unknown. It starts in the first year of life, and is characterized by a triad including spasms, usually in clusters and often involving the extremities and head/neck, followed by regression of psychomotor development, and has in most cases a pattern of hypsarrhythmia in the electroencephalogram (EEG). This is a rare disease difficult to treat, whose first-line treatment is hormone therapy or vigabatrin and may be used alternatively other antiepileptics or ketogenic diet. The prognosis is poor, since most patients have neurocognitive delay, although it can be improved with early identification and treatment.

Case description: A 3-month-old girl began to present episodes of trunk flexion spasms accompanied by eye reversal occurring in several clusters per day. The objective examination showed a vague and weak interaction look. The family doctor referred the patient to a pediatric consultation. Suspecting that this is West syndrome started a multidisciplinary research in order to determine the etiology. The EEG revealed hipsiarritmia. Several treatments have been used with apparent partial control of seizures without objective quantification. Currently under treatment with vigabatrin and ketogenic diet, it keeps daily seizures but more controlled, it has a delay in psychomotor development, and is still unknown etiology.

Discussion/Conclusions: This case highlights the importance of regular monitoring of the clinical course, quantifying the frequency and duration of attacks in order to evaluate the response to the different treatments used. Primary health care can have an important role given the proximity and greater accessibility, being in these situations a support assurance and monitoring the whole family in all its needs, despite the failure to meet their expectations regarding the treatment of patients with West syndrome.