# Trad. Revista Cubana de Neurología y Neurocirugía

**2/2020 P1**

# A336

## Possible Variant of Type II Pascual Castroviejo Syndrome

Posible variante del síndrome de Pascual Castroviejo tipo II

# ABSTRACT

# Objective: To describe the clinical and imaging characteristics in a patient with a possible variant of type II Pascual Castroviejo syndrome.

# Clinical case report: A 44-year-old female patient with spastic congenital hemiparesis, congenital microcephaly, mild intellectual disability, structural focal epilepsy (mesial region of the temporal lobe), and a flat hemangioma port wine stain on the hand. Skull magnetic resonance imaging allowed to diagnose schizoencephaly with open lip or type 2, with bilateral frontal-parieto-temporal polymicrogyria and left periventricular heterotopias. After reaching the therapeutic dose of carbamazepine, the patient improved and her attacks were spaced out in such a way that they occurred approximately once or twice a year. In addition, she began rehabilitative treatment for her spastic hemiparesis. Conclusions: The association of cutaneous hemangioma and abnormalities in the development of the central nervous system in this patient suggested the diagnosis of Type II Pascual Castroviejo Syndrome. However, in this case, the highest number of brain malformations was contralateral to the hand hemangioma. There was no malformation in the posterior cranial fossa, which differs from most of the published cases.

**Keywords**: epilepsy; schizencephaly; hemangioma; heterotopia; congenital hemiparesis; polymicrogyria.

# A342

# Diagnostic and Therapeutic Algorithm in Embolic Stroke of Undetermined Cause

Algoritmo diagnóstico y terapéutico en el infarto cerebral embólico de causa indeterminada

### ABSTRACT

### Objective: To describe a diagnostic algorithm that facilitates individualizing the pillars of secondary prophylaxis in patients with indeterminate embolic stroke. Acquisition of evidence: A systematic review of the studies published from 2014 to 2019 on the clinical, imaging and therapeutic aspects of the spectrum of indeterminate embolic stroke was carried out. We consulted Medline (PubMed), SciELO, Cochrane and Google Scholar databases. Original articles, published in peer-reviewed journals, which included patients older than 18 years with an indeterminate embolic infarction diagnosis were taken into account.

### Results: Indeterminate embolic stroke represents 21% of cerebral infarcts in people younger than 50 years. It is defined as a non-lacunar infarct, with no evidence of significant arterial stenosis or cardioembolic source. Mild cortical infarcts associated with paradoxical embolism are more likely in young people without vascular risk factors. In advanced ages, cardiac embolism and the artery-artery mechanism predominate due to cervical and aortic substenotic plaques. Two opposite etiological phenotypes are distinguished: those with an atherothrombotic substrate and white thrombus formation that benefit from antiaggregation, and those due to the production of red thrombi where anticoagulation is the therapy of choice.

**Conclusions:** Including indeterminate embolic strokes, as a clinical phenotype, makes it possible to perform a stratified diagnostic algorithm with more advanced complementary tests in patients who suffer from it, to reduce the cases that remain cryptogenic. However, these patients represent a large and heterogeneous group that does not allow generalization of the use of anticoagulation.

**Keywords**: embolism; cryptogenic stroke; substenotic atheromatosis, paroxysmal atrial fibrillation; atrial heart disease; foramen oval permeable.

# A348

# Intraoperative Neurophysiological Monitoring In a Case of Benign Giant Dorsal Intraspinal Schwannoma

Monitoreo neurofisiológico intraoperatorio en un caso de schwannoma benigno intrarraquídeo dorsal gigante

# ABSTRACT

# Objective: To prove the efficacy of intraoperative neurophysiological monitoring to reduce the risk of neurological injury in surgery for benign dorsal intra-spinal schwannoma.

# Clinical Case Report: We report a case of a 16 years old female patient, with decreased strength in her lower limbs, as well as fasciculations of one month of evolution. A dorsal spinal tumor was determined. Surgical treatment of the lesion was performed, which was anatomopathological compatible with benign schwannoma. Intraoperative neurophysiological monitoring was used, by somatosensory evoked potentials recording of the posterior tibial nerve bilaterally, and a bilateral pre-surgical evaluation of the P40 component was performed. The tumor excision process was guided by the electrophysiological warning signs. Reversibility was achieved for the changes in the P40 component somatosensory evoked potential record of the bilateral posterior tibial nerve. The postoperative evolutionary records established the normality of the somatosensory evoked potential record values. The patient had a favorable clinical evolution. This is the first patient who underwent neurophysiological monitoring, specifically in pediatric neurosurgery, in Holguín province.

**Conclusions**: Intraoperative neurophysiological monitoring contributed to the subtotal excision of the intra-spinal tumor at the dorsal level; there were no intraoperative neurological complications. The patient evolved favorably after surgery.

**Keywords**: intraoperative neurophysiological monitoring; spinal surgery; benign schwannoma.

### A356

**Behavior of Anxiety, Depression and Dissociation in Patients with Epileptic Seizures and Non-Epileptic Psychogenic Seizures**

Comportamiento de la ansiedad, depresión y disociación en pacientes con crisis epilépticas ycrisis psicógenas no epilépticas

# ABSTRACT

**Objective:** To evaluate the behavior of anxiety, depression and dissociation in patients with epileptic seizures and non-epileptic psychogenic seizures.

**Methods**: A descriptive and correlational study was carried out. Thirty eight (38) patients participated (15 with epileptic seizures and 23 with non-epileptic psychogenic seizures), who were evaluated in the Neuropsychology and Clinical Neurophysiology Services, at the International Center for Neurological Restoration, from January 2001 to February 2018. Demographic variables were analyzed, as well as the type of psychogenic crisis, anxiety, depression and dissociation from the application of a structured interview, the dissociative experiences scale, the generalized anxiety disorder scale and the major depressive episode scale. Non-parametric statistics were used with statistical significance p≤ 0.05.

**Results**: Differences were identified between the groups in relation to chronological age (p = 0.04) and suspicion of generalized anxiety disorder (p = 0.03). A negative relationship was obtained between age and dissociative experiences in the group of non-epileptic psychogenic seizures, specifically, in the group with hypermotor seizures.

**Conclusions**: In the group with epileptic seizures, there is a suspicion of generalized anxiety disorder and a higher age range than the group with non-epileptic psychogenic seizures. The type of non-epileptic psychogenic seizures and their relationship with dissociative experiences offer practical information to redirect the comprehensive care of this group of patients. This diagnosis could help to develop rapid interventions that benefit the quality of life of these patients and their families.

**Keywords**: epileptic seizures; non-epileptic psychogenic seizures; anxiety; depression; dissociation.

# A366

**Autoimmune Encephalitis Due To Antibodies against N-Methyl-D-Aspartate Receptor with Neuropsychiatric Presentation**

Encefalitis autoinmune por anticuerpos contra receptor n-metil-d-aspartato con presentación neuropsiquiátrica

# ABSTRACT

**Objective:** To describe the clinical manifestations of the first case of autoimmune encephalopathy due to antibodies against n-methyl-d-aspartate receptor, confirmed in Pastaza, Ecuador.

**Clinical case report**: A 34-year-old male patient is reported because of insidious behaviour disorders associated with progressive impairment of cognitive functions. He underwent psychiatric evaluation and tests to detect autoimmune diseases, which resulted negative. Due to the presentation of these neurological and psychiatric manifestations in a young patient, a surface electroencephalogram and detection of antibodies against the n-methyl-d-aspartate receptor were indicated for the final diagnosis and to prescribe therapy. The patient was found to have autoimmune encephalopathy. After treatment with steroids, the patient evolved favorably.

**Conclusion:** The clinical manifestations of this case were mental dysfunctions such as insomnia of conciliation and maintenance of sleep, abnormal behaviors, irritability, impulsivity, aggressiveness, hypersexuality, compulsion, zealoty, rambling language and memory disorder in the short and medium term.

**Keywords**: autoimmune encephalopathy; n-methyl-d-aspartate receptor encephalitis; neuropsychiatric presentation; N-Methylaspartate.

**A374**

**Neurodegeneration Due to Early-Onset Brain Iron Accumulation in Two Sisters**

Neurodegeneración por acumulación cerebral de hierro, de inicio precoz, en dos hermanas

# ABSTRACT

Objective: To describe the clinical and imaging characteristics of neurodegeneration due to early onset brain iron accumulation in two sisters.

Clinical cases report: We report the case of two patients (female siblings), 8 and 3 years old, respectively. The oldest child had onset of manifestations in infancy, with neurodevelopmental involvement; she was diagnosed, at that time, with infantile cerebral palsy. Subsequently, neuropsychiatric alterations, deterioration of motor, language, cognitive functions and severe dystonic spasms appeared. After an apparent normal psychomotor development, the second child began to show language disorders and ataxia, with rigidity of progressive evolution. The older sister's previous diagnosis of neurodegeneration due to brain accumulation of iron, in its early form, and the tiger's eye images on magnetic resonance imaging in both patients, facilitated the younger sister's diagnosis.

Conclusions: The appearance of clinical manifestations of dysfunction of the nervous system of different topography, with slow and progressive evolution in early ages, and the finding of tiger's eye images on magnetic resonance, allowed the diagnosis of accumulation neurodegeneration iron brain in both cases. The family history facilitated the diagnosis of the younger child.

Keywords: genetics; neurodegeneration; pantothenate kinase; Pediatrics.

# A377

**Kleeblattschädel Syndrome**

Síndrome de Kleeblattschädel

# ABSTRACT

**Objective:** To describe the clinical manifestations, behavior and postoperative evolution of an infant with Kleeblattschädel syndrome.

**Clinical Case Report:**  We report the case of a two-month-old female infant, daughter of young parents, with no history of genetic or chronic disease. Her mother had prenatal care since the first weeks of pregnancy. Ultrasound studies during pregnancy did not report alterations. At birth, the infant showed mild respiratory distress and dysmorphic features. She was referred to Octavio de la Concepción y de la Pedraja Pediatric Hospital in Holguín, with diagnosis of craniosynostosis. The physical examination revealed a wide anterior fontanelle, bilateral exophthalmos, hypertelorism, maxillary hypoplasia, and a cloverleaf-shaped skull configuration. In the computed tomography report described cloverleaf skull, total closure of sutures and bilateral exophthalmos. The cerebral ventricles were dilated with the temporal horns of the lateral ventricles. Cranial correction was performed by means of linear parasagittal craniectomies at three months of birth. As complications of the surgery, she had cerebrospinal fluid fistula and anemia. After the procedure, the infant had satisfactory evolution.

**Conclusions**: Kleeblattschädel syndrome is an unknown-origin rare variant of craniosynostosis. Early surgical treatment is ideal to meet surgical goals, in addition to preventing deterioration and improving the child's psychomotor development. The patient underwent surgery at three months of age and, despite the postoperative complication she evolved satisfactorily.

**Keywords:** cranial correction; craniosynostosis; Kleeblattschädel syndrome.

# A379

# Clinical Features of Amyotrophic Lateral Sclerosis in Patients with Novo Switches

Características clínicas de la esclerosis lateral amiotrófica en pacientes conmutaciones de Novo

#### ABSTRACT

#### Objective: To describe the clinical and epidemiological features of amyotrophic lateral sclerosis in patients with Novo mutations.

**Methods**: A descriptive and longitudinal study was carried out from January 2005 to December 2019, at the Institute of Neurology and Neurosurgery. The sample consisted of 53 patients diagnosed with clinically defined amyotrophic lateral sclerosis, in whom Novo mutations were identified. The variables studied were genes with Novo mutations, age at onset of the disease, sex, skin color, clinical forms, family history of this disease, and survival. The results were expressed in measures of summary and dispersion.

**Results**: The most frequent genes with Novo mutations were ATXN2, SETX, NEK1, TBK1, ERBB4, NEFH and hnRNPA1. The variable which predominated were age of onset of symptoms ranging 41 and 60 years, male sex (54.7%) and white skin color (58.4%). In patients with ATXN2 gene, survival was less than one year in those with the bulbar clinical form. The hnRNPA1 gene was identified in the white subjects, with a family history of the disease, spinal clinical form and survival higher than five years.

**Conclusions**: ATXN2 gene was the most reported with Novo mutations. It predominated in male patients aged ranging 41 and 60 years, with rapidly progressive evolution, both for those with a bulbar clinical form and for those with a spinal variant.

**Keywords**: amyotrophic lateral sclerosis; epidemiology; genetics; mutations.

\*\*\*/ Traducción: Gretchen González Nieto

Servicio de traducción

CNICM-Infomed

13 de agosto 2020