31. A CASE OF TEMPORAL LOBE EPILEPSY WITH HIPPOCAMPAL SCLEROSIS

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Background. Temporal lobe epilepsy (TLE) is the most common form of focal epilepsy. Approximately 6 out of 10 patients with focal epilepsy have TLE. Hippocampal sclerosis (HS) is a frequent pathological abnormality underlying the TLE.
Case report. A 34-year-old man was admitted to the epileptology clinic with seizure episodes that start focally with an unpleasant feeling in the stomach, unusual smells, unmotivated fear followed by repetitive jerky movements of the left part of body and complex automatic behavior, accompanied with altered consciousness, which periodically progressed to a bilateral tonic-clonic seizure. The postictal period was characterized by confusion and amnesia. The seizures started at the age of 12 years after a traumatic brain injury. Carbamazepine, valproic acid, and clonazepam have been used (in monotherapy or polytherapy) to treat seizures, however, with inadequate response. His video electroencephalogram (EEG) monitoring revealed focal right temporo-frontal epileptiform discharges (spike, sharp wave, sharp and slow-wave) in wakefulness; right temporal, temporo-frontal epileptiform discharges in drowsiness and slow sleep. The high-density EEG (256 channels) revealed the onset of epileptiform activity in the hippocampus (parahippocampal gyrus) with subsequent propagation to the temporal lobe (superior temporal gyrus). MRI scan showed that the inferior horn of the right lateral ventricle measured 5.0 mm (left 1.5 mm) and the right hippocampus had a reduced volume. After the neurological evaluation, patient was started on carbamazepine extended release (15 mg/kg/bid), lamotrigine (3.5 mg/kg/bid) and clonazepam 1 mg/qd. With this combination of drugs, his seizures are partially controlled.
Conclusions. Seizure semiology, video-EEG, high-density EEG, and MRI results confirm the diagnosis of TLE with HS. Taking into account the inadequate control of seizures with medication and the presence of a confirmed structural cause, the patient could be considered eligible for the pre-surgical evaluation. TLE with HS is refractory for treatment in as many as 60% to 80% of cases. However, with the aid of MRI, high-density EEG, and neuropsychological evaluation, patients can now be timely selected for a surgical resection, a procedure that leads to seizure control and improvement in disabling psychiatric symptoms with minimal need for medication. Studies show a better long-term outcome in patients with HS after surgery (up to 90%) in comparison with antiepileptic drug therapy.
Key words: Temporal lobe epilepsy, hippocampal sclerosis, drug resistance, high-density EEG

32. CHARGE SYNDROME

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Background. Introduction. In this descriptive study the clinical and neurological issues related to CHARGE syndrome (C – coloboma, cranial nerves; H – heart defects; A – atresia of the choanae; R – retardation in growth, mental development, G – genital abnormalities, E – ear malformation / hearing loss) were assessed. The study presents the clinical examination of one case with typical form of pathology, along with the identification of diagnosis and treatment particularities. Aim of study. Being a relatively rarely encountered disease, it requires a separate attitude from both patients and medical staff. The aim of the study is the identification of typical existing forms of the disease, in order to determine the principles and methods of diagnosis and treatment.

Case report. Materials and methods. A 19 years old boy was admitted to the Institute of Neurology and Neurosurgery, Chisinau, Republic of Moldova in February, 2020 being evaluated according to clinical methods (investigation, anthropometry) and laboratory tests. Ressults. The patient's complaints were: hearing impairment, memory loss, pain in thoracic and lumbar spine, headache, asthenia, myalgia. Neurological examination: hyposmia; the presence of hearing loss in left ear, and hypoacusis in the right ear; unsteady Romberg's position; diffuse hypotonia. Somatic examination: BMI = 14,7 kg/m2 (hyponutrition), regular pulse, BP = 120/90 mmHg. Patient presents major criteria: atresia of choane, cranial nerve dysfunction – I, VIII, IX, and minor criteria: rhomboencephalic dysfunction including sensorial deafness, hypothalamo-hypophyseal dysfunction (gonadotropin or growth hormone deficiency) - genital hypoplasia and growth deficiency, characteristic facial features, intellectual disability, feeding difficulties, skeletal anomalies – thoracic and lumbar scoliosis grade 2 with rib block T8-T10. Atypical signs: immunodeficiency, gastroesophageal reflux, sleepiness, vestibular abnormalities. Prior to establishing the final diagnosis, the differential diagnosis was: Rubinstein-Taybi syndrome and Oppenheim amyotonia. The CHARGE syndrome is an autosomal dominant genetic condition caused by a mutation in the CHD7 gene. The patient has 2 sisters, 24 and 21 years old, who are also diagnosed with CHARGE syndrome. They both are pregnant, and the risk of passing on the syndrome to their offsprings is very high. Early appropriate investigations of the syndrome facilitate a correct diagnosis and proper management. Given the number of affected systems in CHARGE syndrome, we believe that a multidisciplinary clinical model is beneficial in the management of these children: the general paediatrician, genetic diagnosis, otolaryngologist, ophthalmologist, cardiologist.

Conclusions. The patient manifests a typical phenotype of CHARGE syndrome according to the Verloes's criteria. The diagnosis is usually made on clinical grounds. It requires a genetic test to confirm the CDH7 mutation in order to identify the individual's and their offsprings' risk and to initiate an early targeted therapy.

Key words: charge syndrome, criteria, clinical features, genetic testing

33. EPILEPSY IN PACIENTS WITH MULTIPLE SCLEROSIS

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Background. Multiple sclerosis (MS) is a central nervous system disorder characterized by inflammation, demyelination and neurodegeneration, and is the most common cause of acquired nontraumatic neurological disability in young adults. The course of the disease varies