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

between individuals: some patients accumulate minimal disability over their lives, whereas others experience a rapidly disabling disease course. A part of patients with multiple sclerosis presents also seizures that lead to epilepsy. Several clinical series reported an association between multiple sclerosis and epilepsy. The most studies show an increased comorbidity between multiple sclerosis and epilepsy. The cumulative incidence of epilepsy by 10 years after diagnosis of MS was 1.9%. The probable anatomic basis for the seizures is areas of inflammation, edema, and/or demyelination in the cerebral cortex and the juxtacortical white matter generated by a mechanism that is not completely understood; the fact that these plaques are very common suggests that other factors must operate in view of the rarity of seizures in MS. In most cases, however, the prognosis of epilepsy was good and there seemed not to be any clear correlation between the severity of MS and epilepsy.

Case report. A patient V. male, 41 years , came at a neurologist in April 2019 with the following complaints: facial hyperemia, heat sensations, alterations of consciousness with convulsive components in the anamnesis. At the same time: walking instability, recurrent diplopia, frequent urination, sleeping disorders, memory loss and decrease in body mass. Anamnesis: In 2005 patient has an acute respiratory infection, possible a flu. After a half a year had appeared diplopia, diplopia and frequent urination. In 2007 the diagnosis of multiple sclerosis was established. The diagnosis was confirmed in Moscow and the patient started the treatment with Galatimer acetate (Copaxone) that he administered for 5 years with the improvement of the evolution of the disease. Subsequently administered Acsoglatiran till present but without any obvious effect. In 2015 the patient has a seizure for the first time with unconsciousness but without warning signs. Other signs and symptoms associated with unconsciousness the patient doesn't remember. A similar episode was in 2017. In 2019 the patient received symptomatic treatment in the neurology department for diagnosis: Multiple sclerosis clinically and imagistic defined, recurrent remissive form, in exacerbation, with pronounced atactic syndrome. Structural epilepsy-mesial temporal sclerosis on the right associated to multiple sclerosis plague with focal seizures with bilateral passage treated with Carbamazepine retard 300 mg/day. Now the antiepileptic treatment is Timonil 750 mg/day with a very good outcome.

Conclusions. MS is a risk factor for developing epilepsy. Patients with MS have a threefold increase in risk for developing epilepsy when compared with that expected in the general population. The reason for this increased risk is unclear and needs further investigation.

Key words: Multiple sclerosis, epilepsy, seizures, disability.

DEPARTMENT OF ONCOLOGY

34. COLLECTING DUCT CARCINOMA APPEARING AS A HEPATIC HYDATID CYST. A RARE CASE REPORT

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Background. Collecting duct carcinoma is located in the renal medulla and it originates from the collecting duct epithelium. It involves about 1% from all renal epithelial malignancies.