pathology of the eyes, skin appendages, teeth, central nervous and musculoskeletal systems. The disease manifests in the neonatal period and has four stages: vesicular, verruciform, hyperpigmented and hypopigmented. It occurs mainly in girls, since inheritance has an X-linked dominant character.

A 7-year-old girl, appealed to the department in satisfactory condition, with complaints of mental deficiency, reduced memory, behavior disorders, multiple hyperpigmented spots on the limbs and body. The child after birth had multiple vesicles on an erythematous background on the body, localized linearly, mainly on the flexor surface of the limbs, as well as on the skin of the chest and back. After birth, he was diagnosed with progressive erythema and vesicular rash. In connection with skin manifestations, she was examined for intrauterine infections – pathology wasn't detected. The patient had no family history of skin diseases. Subsequently, the appearance of hyperkeratosis in the form of plaques, warts and lichenoid growths along the Blaschko lines was noted in the affected areas of the body. The girl was under the supervision of a pediatrician. During the examination in the Department revealed: mental retardation, hyperpigmented spots of gray-brown color with light edges, on the extremities of the rash in the form of 'mud splashes', on the trunk – in the form of a 'spiral', ring 'or' marble cake', with the location of pigmentation along the lines of Blaschko.

The course of Bloch-Sulzberger syndrome is suspected in a child with clinical manifestations of mental retardation, stages of the skin process with a debut in the newborn period. To confirm the diagnosis, a molecular genetic examination was performed: mutations of the IKBKG gene, which is located on the X-chromosome, were detected.

Typical skin manifestations allow you to clinically suspect the presence of Bloch-Sulzberger syndrome in newborns, to conduct timely molecular genetic diagnostics to make the correct diagnosis. In view, the importance of differential diagnosis in the neonatal period and the rare disease, a multidisciplinary team of neonatologists, pediatricians and neurologists to this disease is necessary.

406

FIRST EPISODE OF SPORADIC HEMIPLEGIC MIGRAINE IN PEDIATRIC PATIENT- DIAGNOSTIC PHANTOM

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The aim of this case report is to describe rare form of migraine in pediatric patient and to point out difficulties of making the right diagnose and classification according to ICHD-III (International Headache Society 2018.)

Case Report A 14-year old girl driven by emergency team to pediatric emergency from school where she had severe unilateral headache with nausea and paresthesia in the right hand and leg about an hour before arrival. At arrival she was confused, unknowing the name of her school and dysphasic. Neurological exam showed dysphasia and right hemiparesis, in the next half an hour she was somnolent, only opening her eyes when shaken and started vomiting.

After two hours (three from the first symptoms) all clinical signs vanished, she was in her full consciousness, ambulatory with no neurological deficits!

-Brain CT, EEG and MRI/MRA were normal, so did D-dimers and coagulogram, we transported her to tertiary level center

Differential Diagnoses 1. cerebrovascular disease- ischemic or hemorrhagic stroke; transient ischemic attack 2. CNS infections/encephalopathy/brain tumor 3. syndrome of transient headache and neurological deficits with cerebrospinal fluid lymphocytosis (HaNDL) 4. migraine

Diagnosis and Classification -Diagnosis is clinical, TIME has major role -It is the diagnosis of exclusion- when we exclude diagnosis from above and time shows that the attack was episodic and reversible, we can than afford ourselves to think about migraine -which type of migraine? According to ICHD -III it does not fulfill criteria for hemiplegic migraine since it takes at least two attacks with motor deficit, so we can only classify this case as 1.5.2: probable migraine with aura.

Conclusion Diagnosis of a first episode of sporadic hemiplegic migraine (severe attack) in a pediatric patient is very challenging for a clinician, especially in a first few hours What makes already dramatic clinical picture even harder to diagnose:

-children and adolescents find it hard to describe their aura symptoms- motor aura is necessary feature for the diagnosis, clinician must establish as clearly as possible whether the patient actually experienced motor weakness as an aura symptom -lack of literature about this cases; no guidelines for the management of acute attacks -signs and symptoms described in all of the other aura types- heterogeneous symptoms Regardless of the new technologies in diagnostic, accessible medical data bases and several headache classifications, some conditions will always remain challenging

407

VESTIBULAR SCHWANNOMA IN A TEENAGER: CASE REPORT

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Relevance Vestibular Schwannoma (VS) – is a benign encapsulated tumor, initially arises in the inner auditory canal from the neurolemma of the vestibular nerve with further growth towards the base of the skull. Basically, they are associated with neurofibromatosis type 2 (NF2). In children sporadic cases of VS are uncommon. We report a case of VS in teenager.

Methods A 15-year-old girl with a viral infection was diagnosed with unilateral acute sensorineural hearing loss (SNHL). There was no NF2 family history. General medical examination revealed no ENT-pathology. MRI showed no tumor and vascular malformations in the brain and temporal region. After the standard course of dexamethasone effect was not obtained. SNHL progressed over the next 9 months. Evaluation of advanced hearing tests showed: tympanometry (probe-ton 226 Hz) demonstrated Type A bilateral, TEOAE were documented as 'Pass' bilateral, pure tone audiometry determined profound hearing loss on the right, Auditory Brainstem Response (ABR) was defined wave V latency on the right, ABR thresholds were detected with intensity stimulus level 85 dB SPL on the right and 30 dB SPL on the left. At the series X-ray CT images of temporal region were determined inner auditory canal deformation and extension. A second MRI scan revealed a bulky tumor coming from the inner auditory canal. The

tumor was removed with Stereotactic Radiosurgery (Gamma Knife system).

Conclusions Unilateral hearing loss in a child is a reason for an extended examination in order to exclude retrocochlear pathology.

408 HYPERKINETIC AND HYPOKINETIC MOVEMENT DISORDERS – IN PEDIATRIC CLINICAL PRACTICE

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To present heterogenous group of motor movement disorders of different etiology and clinical features.

Medical history and clinical exam and assessment, MR neuroimaging, metabolic tests, molecular genetics, lumbar puncture and cerebrospinal fluid exam, antibodies to surface neuronal antigens, EEG monitoring, abdominal ultrasound were performed.

Patients with autosomal dominant torsion dystonia DYT 6 (THAP1 gene mutation) and movement disorders caused by mutations in ATP1A3, SCN8A, CaCN1, PANK2, POLG1 genes and acquired disorders: autoimmune (NMDAR) encephalitis, dyskinetic cerebral palsy, opsoclonus myoclonus ataxia will be presented as well as treatment response.

MD can manifest alone or as part of complex phenotypes of different etiology. Considerable overlap of MD and seizures exists especially in patients with epileptic encephalopathies, ataxias and spasticity in whom clinical diagnosis can be more challenging.

409 HEART OR BRAIN GENETICS?-ICTAL CARDIORESPIRATORY ARREST DURING THE FIRST AFEBRILE EPILEPTIC SEIZURE WITH LEFT TEMPORAL ONSET- LIFE THREATENING EVENT WITH GOOD OUTCOME

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Temporal lobe has important role in the regulation of autonomic function, while the insular cortex is associated with cardiac autonomic functions control. Focal epileptic activity in the temporal lobe may cause cardiac dysfunction.

Patient and Methods A girl at the age of 12 developed cardiorespiratory arrest after she vomited several times during night. She was resuscitated and mechanically ventilated. Her pro-BNP and troponin values were increased, as well as CK, ECG monitoring in ICU was unremarkable, however Holter-ECG showed ST elevation and prolonged QRS complex. Her EEG monitoring showed focal dyscharges from frontotemporal region right. Heart US showed mild dilatation of apical region. Cardioverter defibrillator (ICD) was implanted and carbamazepine treatment was introduced. Her brain MR was normal as well as her heart US after 5 days. She manifested febrile seizures during childhood, attention deficit hyperactivity disorder and joint laxity. Next generation sequencing revealed

heterozygous KCND3 gene mutation :c.916G>A;p.Gly306Ser. KCND3 is reported for Brugada syndrome as well associated with developmental delay, intellectual disability, ataxia, epilepsy and attention deficit hyperactivity disorder.

Conclusion Ictal discharges involving limbic structures may be a cause of both apnoea and ictal asystolia by inhibition of brainstem cardiorespiratory control circuits caused by the mutation in KCND3 gene.

Cardiorespiratory arrest may occur as results of autonomic dysfunction provoked probably by postictal suppression.

Paediatric Pulmonology

410 INFLAMMATORY LUNG DISEASE IN RETT SYNDROME

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Rett syndrome is a progressive pervasive neurodevelopmental disorder that affects female patients and is the second most common cause of mental retardation in the female gender, with an incidence of 1:10,000 to 1:15,000 live born girls. The classic form of Rett syndrome in 95% of cases is caused by de-novo mutation on the X-chromosome-linked gene encoding the methyl-CpG-binding protein 2. It is characterized by mental retardation, ataxia, epilepsy, characteristic stereotyped movements of the hands and loss of their normal function and breathing disorders. There is no effective cure, but early recognition of the disease and early treatment, primarily antiepileptic and respiratory, can delay the onset of further complications. Breathing disorders, including apnea, hyperventilation, rapid and shallow breathing, breath holding and spontaneous Valsalva maneuvers, are due to ventilatory perfusion inequality and are the result of a number of factors, including oxidative stress and chronic subclinical inflammation. The aim of this case report is to present a girl with Rett syndrome who has been treated with antibiotics on several occasions for radiologically persistent changes in the right upper pulmonary lobe as the changes are understood as pneumonic infiltrate. A girl aged 3 years and 2 months diagnosed with Rett syndrome was hospitalized for additional diagnostic treatment of radiologically verified persistent changes of the upper right pulmonary lobe. The girl was treated with antibiotics at the ages of 15, 17 and 18 months as to have had bacterial pneumonia of the right upper pulmonary lobe. In July 2019, esophagogastroduodenoscopy, bronchoscopy, and computed tomography of the thorax were performed. Computed tomography showing in the region of the upper pulmonary lobe the consolidation of the pulmonary parenchyma with visible air bronchogram and deformation of the bronchi in the branches oriented toward the medial and in the basal segments of the upper paracardial lobe. The described changes are within the framework of Rett syndrome and correspond to the contents filled the alveoli with impaired ventilation and the minimal segment deformation of the bronchi by type of bronchiectasis. Changes of the upper right pulmonary lobe, on computed tomography of the thorax, are found in 50% of children with Rett syndrome and are chronic in nature and do not require antibiotic treatment.