DNA detection on Guthrie-card resulted positive and a novel gene mutation in PLP1 was found at whole-exome-sequencing.

In both cases neurological impairment differential diagnosis between cCMV and genetic syndrome appeared particularly challenging. bMRI suggested for both a cCMV causative role: in case-1 radiologic imaging revision revealed a possible fetal clastic damage, in case-2 grey matter degeneration with normal white matter appeared inconsistent with PLP1 mutation. Moreover, a detailed perinatal history revealed early signs of cCMV, which were ignored: in case-1 intrauterine-growth-restriction, in case-2 pathological audiological screening.

In conclusion, congenital cytomegalovirus infection should always be considered in cases of infant with unexplained neurological impairment, particularly when signs suggestive of congenital infection are present at birth. In fact, an early screening of possible neurological sequelae allows precocious treatment whose efficacy is documented only if started within the first month of life.

386 EPILEPSY IN PEDIATRIC PATIENTS – EVALUATION OF ANATOMIC STRUCTURES' VOLUME OF THE BRAIN

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Epilepsy is a disease of the central nervous system with somatic, vegetative and psychological symptoms that appear as a result of various morphological and metabolic changes in the brain. Epileptic seizures are the manifestation of temporary problems with communication between nerve cells. About 1% of the world's population suffers from epilepsy. Published articles have focused so far on evaluating changes in adult patients. The aim of the study was to evaluate anatomic structures' volume of the brain in pediatric patients with epilepsy.

A group of 42 pediatric patients with clinical symptoms of epilepsy (study group) and 16 healthy patients (experimental group) aged 3 months-17 years were enrolled in the study. Brain MR imaging was performed in all children according to a dedicated protocol (epilepsy specific protocol). Individual anatomical structures of the central nervous system were analyzed on the basis of T1-weighted 3D isometric 1 mm sequence and volume changes of specific structures were compared between the epilepsy group and the control group.

In the study group, the ratio of brain tissue to CSF was 89,08% to 10,92%, while in the control group it was 90,99% to 9,01%. In the research group compared to the control group, the volumes of each brain structure were: cerebrum – 77,99%/79,68%, cerebellum – 9,65%/9,87%, brainstem – 1,41%/1,44%, lateral ventricle – 1,41%/0,49%, caudate – 0,54%/0,55%, putamen – 0,61%/0,68%, thalamus – 0,82%/0,91%, globus pallidus – 0,18%/0,19%, hippocampus – 0,45%/0,50%, amygdala – 0,11%/0,10%, accumbens – 0,04%/0,05%.

During the course of epilepsy in pediatric patients, there is a decrease in the volume of brain tissue, with particular emphasis on the cerebrum, cerebellum, brainstem, caudate, putamen, thalamus, globus pallidus, hippocampus and accumbens, moreover an increase in the volume of lateral ventricles. The study indicates cortical and subcortical atrophy in pediatric patients with epilepsy. The data obtained have important

clinical and prognostic significance, however they need to be confirmed on a large study group with taking into account changes in the volume of anatomical structures of the brain in relation to age and disease duration

387 EVOLUTION OF FOCAL EPILEPTIFORM DISCHARGES IN THE ELECTROENCEPHALOGRAPHY OF THE PREMATURE NEWBORN WITH WHITE MATTER LESIONS

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Advances in neonatal medicine has dramatically improved survival rate of preterm infants that are frequently born with numerous neurodevelopmental disabilities including epileptic seizures and epilepsy. In this dissertation the hypothesis was that localization of white matter injury is associated with epileptiform EEG changes and motor development.

In 64 preterm infants we analyzed EEG, brain MRI and ultrasound and motor development. We evaluated the relationship of brain white matter lesion with EEG and lesions of certain brain localization (segment II – crossroads than parietal, temporal, occipital lobe, basal ganglia, cerebellum and ventriculomegaly) with epileptic seizures and neurodevelopmental outcome.

The total number of white matter lesions was in a positive correlation with the pathological EEG findings and epileptic seizures in infants. Infants who did not have visible anterior and posterior crossroads frequently had epileptic seizures during newborn period and worse neurodevelopmental outcome.

The obtained results indicate that white matter damage is associated with epileptogenesis. Among individual localization, the visibility of segment II as the place where commissural, projection and associative fibers are crossing has a prognostic value for the future neurodevelopmental outcome.

Further studies are needed in order to determine the pathogenic factors that contribute to epileptogenesis in infants with white matter lesions

388 HSV-1 ENCEPHALITIS MIMICKING BILATERAL MCA STROKES IN A YOUNG TODDLER

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A 15-month-old girl presented with a 2-day history of altered behaviour, lethargy and vomiting. She developed a focal left motor seizure that responded to buccal midazolam. She was encephalopathic and apyrexial at presentation but had fever while in the ward. Initial biochemical investigations were not suggestive of infective or metabolic conditions. A CT angiogram and an MRI head showed evidence of multifocal infarct within the middle cerebral artery territories bilaterally and a small area of haemorrhagic transformation on the right frontal region. There were also older bilateral thalamic infarct noted. Echocardiography study was normal. There was no evidence of thrombus from any of the imaging modalities.

Cefotaxime, aciclovir and low-molecular-weight heparin were commenced early.

EEG revealed generalised encephalopathy and epileptic seizure predisposition on the right hemisphere, consistent with the clinical and radiological findings. Lumbar puncture was not immediately performed in view of the intracranial bleed and anti-coagulant treatment. Due to her poor clinical response, steroid treatment was started for suspicion of an autoimmune or inflammatory condition. A CT head done after 48 hours didn't show progression of the haemorrhage. We proceeded with lumbar puncture after withholding a dose of LMWH, screening for infection, metabolic and autoimmune panels. HSV-1 was detected in the CSF, despite no cutaneous or serological evidence of HSV infection. Concurrent cell counts showed pleocytosis confirming the clinical suspicion of encephalitis. She remained on aciclovir for 21 days as a definitive treatment.

Discussion Ischemic stroke and haemorrhage are increasingly recognised CNS manifestations of HSV infection. The brain ischemia is mostly related to multifocal cerebral large vessel vasculitis. HSV-related infarction is a rare but potentially treatable cause of stroke. Steroid treatment may be considered even in the absence of confirmation of vasculitis on neuroimaging. A systematic review showed up to 50% of cases with HSV ischemic manifestations presented with encephalitis while 30% presented with stroke-like symptoms.

Performing lumbar puncture is crucial to differentiate encephalitis from other forms of encephalopathy. Ideally it should be undertaken prior to initiation of aciclovir treatment, however this is usually delayed by other investigations, due to the nature of presentation. Therefore, high index of suspicion is needed to start aciclovir empirically as early treatment could reduce mortality and morbidity significantly.

Conclusion Cerebrovascular events in children should be recognised as a possible manifestation of HSV encephalitis. Aciclovir should be commenced early and CSF sampling should be undertaken at the earliest opportunity once safe to perform.

389 COGNITIVE DISORDER IN CHILDREN WITH EPILEPSY

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Evaluation of different factors influence on cognitive function (CF) in children with epilepsy 20 children with epilepsy: 15 girls (75%) and 5 boys (25%), from 6 to 17 years old (mean age 11 years 9 months). The investigation included: standard neurology status, neuroimaging by CT or MRI, EEG, cognitive function examination by Epi Track Junior it was test. Anamnesis of Life and disease (duration and debut age of epilepsy, type and frequency of seizures, antiseizures therapy), delivery anamnesis and early motor and speech development.

There were several groups in Epi Track Junior results testing: good results 0%, mild disorder of CF – 25% (5 children), average disorder of CF – 20% (4 children), severe disorder of CF – 55% (11 children).

In light asphyxia (Apgar less than 7 degree) delivered 5 children (25%), in the rest cases (75%) Apgar was normal. Early development was normal in 19 children (95%) and 1 child (5%) with average speech retardation.

Unknown epilepsy etiology has 12 patients (60%), genetic – 3 (15%) and structural – 5 (25%). Focal seizures without consciousness impairment were in 2 cases (10%), generalized in 14 (70%) and focal with secondary generalization in 4 (20%). In remission were 3 children (15%), with rare seizures were 9(45%), 1 (5%) were with frequent seizures and 7 children were (35%) with very frequent seizures.

All patients (3 (15%) in polytherapy had severe CF disorder. Children with frequent seizures (8 patients (40%) had mild CF disorder in 2 cases (10%), average CF disorder in 2 cases (10%) and severe CF disorder in 4 cases (20%).

Debut of seizures up to 7 years was in 9 children (45%) and 7 (35%) had severe CF disorder.

Duration of epilepsy from 1 to 3 years was in 4 children (20%) and 2 children (10%) of them had severe CF disorder. Duration epilepsy from 3 to 7 years were in 7 children (35%) and all of them had severe CF disorder.

Severe and average CF disorder were showed in children with frequent seizures, early (up to 7 years) debut of seizures, duration of disease more than 3 years and polytherapy cases.

There was no influence on CF: Apgar score, early development, neurology deficit, structure changes in neuroimaging, type of epilepsy and seizures, EEG changes.

390 EFFICACY OF GENE THERAPY WITH ELADOCAGENE EXUPARVOVEC IN PATIENTS WITH AADC DEFICIENCY COMPARED WITH NATURAL HISTORY CONTROLS

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Aromatic L-amino acid decarboxylase (AADC) deficiency is a rare autosomal recessive disorder resulting in marked or complete loss of dopamine, impeding normal motor development. Eladocagene exuparvovec, a recombinant adeno-associated virus containing the human cDNA encoding the AADC enzyme, is in clinical development for treatment of AADC deficiency.

Eladocagene exuparvovec was administered via bilateral infusion into the putamen of 28 children with AADC deficiency in 3 clinical trials (AADC-CU/1601 [8 patients, completed], AADC-010 [10 patients, ongoing], and AADC-011 [10 patients at data cutoff of 26 February 2020; ongoing]).

Patients received a total dose of 1.8×10^{11} vg (n=21) or 2.4×10^{11} vg (n=7; AADC-011). Motor milestone achievement was assessed using the Peabody Developmental Motor Scales, 2nd Edition. Improvements in motor function from treated patients in the full analysis set were compared to 49 subjects from a comprehensive Natural History Database (NHDB). This database was derived from a systematic review of literature reporting data from patients with AADC deficiency. From an original pool of 237 patients, 49 were chosen as matched natural history controls. These natural history controls were chosen because they met the criteria of unique patients with confirmed AADC deficiency who had not participated in clinical trials of eladocagene exuparvovec, had documented lack of motor milestone achievement, and had a similar disease phenotype to patients in clinical trials.

As early as 12 months after receiving eladocagene exuparvovec, 44% of patients achieved head control and 20% of