hypertension. Routine laboratories investigations were normal, and there were no renal anomalies. Extensive investigation disclosed no cause for their HTA. Neurologic clinical course was rapidly favourable. The first case was treated for HTA for 3 months by an ACE-inhibitor. The second had a calcium channel blocker for few days

Conclusions PRES is rare in children and often associated with concomitant renal disorders. To the best of our knowledge the two cases are among the first publications of PRES syndrome with a common viral infection as a possible risk factor

PP-047 CONGENITAL MYASTHENIC SYNDROME AND EPILEPSY; A RARE CASE REPORT

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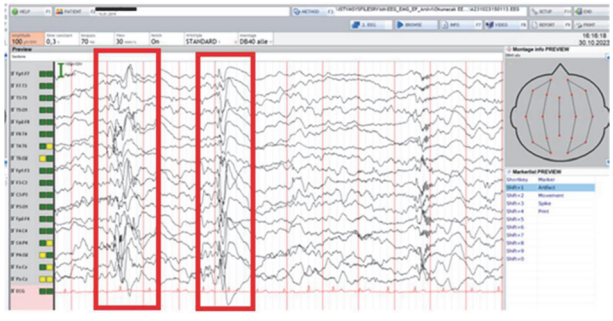
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Aim Congenital Myasthenic Syndromes (CMS) are rare hereditary neuromuscular junction diseases characterized by weakness the ocular, bulbar and skeletal muscles during infancy and childhood. CMS were divided three groups such as presynaptic, synaptic or postsynaptic. Main clinical features of presynaptic types are fatigue, apnea, dyspnea, ventilator dependence, ptosis and bulbar paralysis (1). Cognitive disability, dysmorphism, neuropathy, or epilepsy are rare. (2) Development of Whole Exome Sequencing(WES) leads us to discover much more mutation in CMS genes. (3). CHAT gene mutation is approximately 5% at CMS and a member of the presynaptic types (3). In this case report, we planned to pay attention about clinical features and treatment responses a patient extremely rare CHAT mutation.

Material and Method A 5-year-old male suffered from disability of walking, limping and fatigue during the infantile period. Initial symptoms started at 1 year 4 months aged. On physical examination truncal hypotonisity, muscle weakness, increased deep tendon reflexes and abnormal involuntary movements were detected. Autosomal recessive mutation in CHAT gene was detected in WES. Tracheostomy due to extubation failure during several hospitalizations in pediatric ICU. He is feeding via nasogastric way. His brain MRI showed cerebellar cortical atrophy and ventricular enlargement. (figure 1–2) During neurology follow-up EEG was performed on the patient with movement disorder and suspicious epileptic activity. EEG shows us polyspikes and spike-and-slow-wave pattern. (figure 3) He used pyridostigmine and haloperidol before seizures started. According to EEG results, levetiracetam treatment was also started and significant benefit was obtained.

Results Multiple genes were influence developement of CMS. WES should br performed in these patients to detect the related gene. Although CMS characterized mainly with muscle weakness and respiratory distress, our case diagnosed with epilepsy also.

Conclusions Co-occurrence of CMS and epilepsy reveals the necessity of fallow up in terms of accompanying systemic diseases, and larger studies are needed.



Abstract PP-047 Figure 1

PP-048 PHARYNGEAL-CERVICAL-BRACHIAL VARIANT OF GUILLAIN BARRÉ SYNDROME WITH ANTI GANGLIOSIDE ANTIBODIES

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Aim Guillain-Barré syndrome (GBS) is an acute-onset ascending sensorimotor neuropathy often provoked by a preceding infection. Atypical variants of GBS include; pure sensory variant, pure motor variant, bilateral facial palsy with paraesthesias, pharyngeal-cervical-brachial (PCB) variant, and the Miller



Abstract PP-048 Figure 1 Sagittal T1 Spinal MRI. Spinal MRI revealed enhancement of the root of the spinal nerve and the cauda equine (figure 1).

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Fisher syndrome (MFS). We describe a child with PCB predominant weakness overlapping with classical GBS syndrome.
Material and Method .
Results A 9-year-old boy was referred for acute onset dysphagia, speech, gait difficulty and weakness of limbs of 4 days. He had a history of acute gastroenteritis recently. He had bilateral peripheral facial palsy more prominent on the left side, dysphonia. Shoulder abduction was 2/5, elbow flexion and extension 4/5, distal upper extremity and lower extremity muscle powers were 5/5. Biceps and brachioradialis deep tendon reflexes were hypoactive, patella and Achille reflexes were absent. He had an ataxic gait. Other neurologic and systemic examination of the patient was normal. Brain MRI was normal, spinal MRI revealed enhancement of the root of the spinal nerve and the cauda equine (figure 1). Intravenous immunoglobulin was given at a dose of 2 gr/kg. During follow up, lower extremity weakness developed, and he could not walk without assistance. The EMG revealed demyelination, slow motor and sensory nerve conduction. He was diagnosed with GBS. Anti-Ganglioside antibody panel was tested from the serum and anti-GQ1b and anti-GD1b were found to be positive. His bulbar symptoms were resolved and he was discharged two weeks after the initial presentation. He can walk without support.
Conclusions Atypical variants of GBS can be seen with regional involvement. Anti-ganglioside antibodies are usually found in patients with cranial nerve involvement (2). Our case shares features of PCB variant of GBS and classical GBS with tetraparesis. Early recognition of atypical variants of GBS is important for early treatment.

PP-049 METHYLMALONIC ACIDEMIA IN SIBS: CLINICAL CASE

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Aim Not aplicable for clinical cases.
Material and Method Clinical case.
Results «Methylmalonic acidemia' refers to a group of inborn errors of metabolism associated with elevated methylmalonic acid (MMA) concentration in blood. We observed the family with two boys with MMA. Marriage is consanguineous. Mother had 3 pregnancies. The first pregnancy ended spontaneously. The woman had high risk of miscarriage during the second pregnancy. The boy birthed at 39–40 weeks. The birth weight was 3070 g, APGAR 6–7. The child began to suck poorly on the 2nd day. Measured concentrations of aminoacids was 1302 mkmol/l for Valin (3,6 times higher than normal), 1598 mkmol/l for Leucin (4,3 times higher), the concentration of C3 was 8 mkmol/l (also 2 times higher). Diagnostically MSUD was presumpted as main methabolic error. The child is assigned «Nutrigen 14» (-leu, -ile, -val). The child died at the 12th day of life, due to multiple organ failure. Posthumously obtained the result of a urine test; measured concentration of MMA in urine was 6,292.14 mmol/mol CRE (normal range 0–2). Pathogenic mutation c.360dupT (p.Lys121fs) of the MMUT was identified. During the 3rd pregnancy, there was a high threat of miscarriage at 8 weeks of gestation. Same mutation of the MMUT was identified in the fetus in homozygous state. Childbirth at 40 weeks of pregnancy. Birth weight was 3390 g, APGAR 9–10. At the first day of life, the ammonia in the blood was normal. Measured concentration of C3 in blood was 15,9 mkmol/l (3 times higher than normal value) and level of MMA in urea was 1072,16 mmol/l (normal range 0–2 mmol/l). The child is assigned «Anamix infant MMA/RA», «Nutrigen 14 MMA/RA». Current child's status: Age 5.5 months, weight 7340 g. Routine C3 blood level is 13.2 mkmol/l (normal range 0.13–6.8 mkmol/l), blood ammonia is 94 mkmol/l (normal range 21–50).
Conclusions Clinical case.

PP-050 CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY CASE REPORTING IN KRASNODAR REGION

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Aim This article reports case of carnitine palmitoyltransferase I deficiency, first identified in Krasnodar region as part of extended neonatal screening.
Material and Method Clinical observation.
Results Male infant, born in 2023 in closely related marriage (parents are cousins). It was his mother's 3rd pregnancy (1st – healthy girl, 2nd – frozen pregnancy, 3rd – current). 2 of deliveries were urgent surgical. Birthweight 3520 g, body length at birth 54 cm, Apgar score: 8/9. Type of nutrition - breastfeeding. TMS revealed changes – C0 increase, C18:1, C18:2 decrease. Such changes may be typical for carnitine palmitoyltransferase I deficiency. Confirmatory tests revealed previously undescribed nucleotide sequence in exon 11 of CPT1A gene variant(chr11:68549409A>T) in homo/hemizygous state, causing premature terminating codon to appear (NM_001876.: c.1182T>A (p.Cys394*)). Detected nucleotide sequence variant isn't registered in Genome Aggregation Database control sample(gnomAD v2.1.1). ACMG criteria consider this variant as probably pathogenic. Variants in homozygous and compound heterozygous CPT1A gene were described in patients with CPT deficiency, hepatic, type IA (OMIM:255120). At age of 1,5 months infant's mother didn't complain during examination, no somatic and laboratory changes were found. The infant's feeding started with specialized product for infants from birth and adult patients with long-chain fatty acids oxidation defects, chylothorax and lymphangiectasia, due to which TMS tested carnitine levels normalized. Later patient's mother canceled low-fat formula by her own for 2 months and resumed breastfeeding, which resulted in excessive weight gain, increased liver size, cytolysis syndrome up to 2 norms, hyperammonemia, increased CK at 3,5 months age. Hypoglycemia symptoms weren't detected. TMS dynamics showed more than 2 norms increased carnitine level, increased C0/C18 acylcarnitine ratio. Low-fat formula resumption led to laboratory parameters normalization within 3 days.
Conclusions Our observation showed necessity of early low-fat diet initiation and parents education, which should minimize risk of complications and timely consultation of families for offspring genetic risk assessment.

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