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1.NINO TOGONIDZE [1], TAMTA KHUJADZE [1], TINATIN DAVITAIA [1,2], NAZI TABATADZE [3]

AUTOSOMAL RECESSIVE COMBINED METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CLBC: RENAL MANIFESTATION

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INTRODUCTION: Defects in vitamin B_{12} metabolism are inherited as autosomal recessive disorders and are classified into the eight genetic complementation groups (cblA-G and mut). Among them, cblC type is stated to be the most common form. This disorder exhibits a wide spectrum of clinical manifestations, spanning the prenatal period through late adulthood. Early-onset disease typically presents within the first year of life with nonspecific systemic, neurological and hematological abnormalities. Survivors of early-onset form may have severe neurological impairment despite treatment. Late-onset disease, defined by onset after four years of age, most commonly presents with acute neurological deterioration and generally associated with a better outcome when treated promptly. ClbC disease may manifest with a range of kidney disorders, with thrombotic microangiopathy being the most frequently reported manifestation.

CASE DESCRIPTION: We describe 15-year-old female with a history of arterial hypertension, stage II admitted to our hospital. On admission, laboratory tests revealed advanced renal failure (Creatinine – 11 mg/dl; Urea – 24 mmol/l). Patient was hospitalized in the department of Uro-nephrology and work-up for the evaluation of etiology was undertaken. Doppler ultrasonography revealed thrombus in right renal artery. Total MRI and thorough thrombophilia testing (Protein S activity – 90% (N – 62-126); Protein C activity – 89% (N – 70-131): APC resistance – 4.2 (N - >3): Lupus anticoagulant – 1.37 (>2 positive); anti – β 2glycoprotein IgG – 2.90 u/ml (N - <5.00); anti - β 2glycoprotein IgM – 1.30 u/ml (N - <5.00); cardiolipin IgG – 3.00 u/ml (N - <10.0); Cardiolipin IgM – 1.50 u/ml (N - < 7.0); Genetic testing: PAI – 1 4G/5G – heterozygous 4G/5G; Factor II G20210A – neg; Factor V leiden – neg; MTHFR C677T – heterozygous WT/MUT; MTHFR A1298C – neg was performed. WGS revealed homozygous variant c.276G>A p. (Glu92Glu) in *MMACHC* gene, consistent with the diagnosis of autosomal recessive combined methylmalonic aciduria and homocystinuria type cblC. The patient is currently undergoing continuous renal replacement therapy, awaiting kidney transplantation, managed with antihypertensive medication and is followed by metabolic specialist.

CONCLUSION: Awareness of the diverse clinical presentations of cblC disease, especially its potential renal manifestations is essential for preventing irreversible kidney damage through timely diagnosis and intervention. The early detection of cblC disease by newborn screening provides a new opportunity to improve the clinical outcome of affected patients.

2.NINO SOLOMONIA [1,2], TATIA MUKBANIANI [1]

CRIGLER-NAJJAR SYNDROME

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INTRODUCTION: Crigler-Najjar syndrome (CNS) is an uncommon autosomal recessive disorder of bilirubin metabolism, resulting from mutations in the UGT1A1 gene, which induces deficient activity of the hepatic

enzyme UDP-glucuronosyltransferase 1A1. Type I CNS is marked by complete enzyme deficiency, while II type retains partial activity and is often responsive to enzyme inducers. Crigler-Najjar syndrome clinically presents with jaundice, typically in the neonatal period. CNS type I carries a significant risk of kernicterus and, thus, irreversible permanent neurological damage if untreated, while type II usually manifests with milder symptoms. The approximate incidence is 0.6–1 per million live births worldwide. Diagnosis relies mainly on molecular genetic testing. The prenatal diagnosis is also possible through amniotic fluid analysis. Management is meant to lower bilirubin levels, primarily through intensive phototherapy and, when necessary, exchange transfusion. Liver transplantation remains the only life-saving option for type I so far. Long-term prognosis varies; Even with survival into adulthood, approximately 30% of type I patients may develop permanent neurological impairment.

CASE DESCRIPTION: We report the case of a 9-day-old male term neonate who presented with significant unconjugated hyperbilirubinemia. The newborn displayed noticeable jaundice beginning shortly after postnatal hospital discharge. However, over the following days, the yellow discoloration of the skin and sclera became increasingly evident. Consequently, the infant was referred to the pediatrician for an ambulatory visit. By the time of admission, the infant appeared increasingly lethargic, prompting urgent evaluation. Laboratory analysis revealed a markedly elevated total serum bilirubin level of 450 μmol/L - a critical threshold associated with a high risk for bilirubin encephalopathy and kernicterus, and due to the worsening state, hospitalization was necessary. Upon hospitalization, intensive phototherapy was initiated immediately. Despite continuous treatment, the serum levels of unconjugated, indirect bilirubin remained persistently high. The infant's clinical condition fluctuated significantly during the course of admission, alternating between periods of relative stability and episodes of critical deterioration. Over time, the patient also developed bilateral cataracts, raising further concern for systemic complications. Infectious causes were investigated and confirmed, and comprehensive diagnostic workup was undertaken to rule out acute surgical conditions, as well as inborn errors of metabolism. Given the persistent hyperbilirubinemia and lack of response to conventional treatment, a genetic etiology was suspected. Whole exome sequencing ultimately confirmed the diagnosis of Crigler-Najjar syndrome. The analysis identified two homozygous pathogenic variants in the UGT1A1 gene: NM_000463.2:c.1133T>A, p.(Val378Asp), and NM_000463.2:c.-3275T>G. Interestingly, these genetic variants have been previously linked to both type I and type II forms of the disorder, which contributes to the exceptional rarity of this clinical case.

CONCLUSION: Crigler-Najjar syndrome is an exceptionally uncommon condition, and identifying it during the neonatal period can be particularly challenging due to the broad range of potential causes of unconjugated hyperbilirubinemia. Prompt detection and management are critical, as sustained elevated bilirubin levels can lead to permanent neurological impairment. This case underscores the need to include Crigler-Najjar syndrome in the differential diagnosis when a newborn presents with persistent jaundice that does not respond to conventional treatment. Furthermore, this case highlights the critical role of early counseling for parents. Offering families detailed and understandable guidance regarding the condition, available treatment strategies, and potential long-term effects is crucial for facilitating informed, shared decision-making and enhancing the quality of care provided. Equally important is enhancing healthcare providers' awareness to facilitate early recognition, timely initiation of appropriate interventions, and referral to specialized services when needed.

3.DAVIT KATAMADZE, DAVIT TSAKADZE

"VICTORY OF LIFE OVER DEATH" – POLYTRAUMA IN CHILDREN

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INTRODUCTION: Polytrauma (from the Greek: "poly" means many and "trauma" means injury) is a condition in which the same patient has at least two or more severe injuries to different parts of the body or organs at the same time. These injuries pose a threat to life and often require rapid, complex medical intervention.

CAUSES OF POLYTRAUMA IN CHILDREN: 1. Traffic accidents (approximately 50%): 2. Falls from heights (approximately 20-25%); 3. Violence (among adolescents) and family incidents; 4. Sports or play-related injuries. Statistics in children – based on the United States case: 1. In the U.S there are more than 150000-200000 cases of polytrauma in children per year; 2. Hospitalization rate: 14.5 cases per 100000 children; 3. Mortality rate among children with severe polytrauma is 6-10%. Highest risk age groups: 1. Ages 1-4 - household cases; 2. Ages - 15-17 traffic injuries and violence.

MEDICAL MANAGEMENT AND OUTCOMES: 1. 25% of children with polytrauma require intensive therapy (ICU care); 2. More than 15% undergo surgical intervention; 3. In the long term, 30% develop physical or psychological complications (for example, neurological damage, etc.).

CASE DESCRIPTION: patient V.A 14 years old while riding a bicycle and attempting to cross the road, the patient was stuck by car. He was thrown off and landed approximately 30 meters away. He lost consciousness immediately after falling and his heart stopped beating. Right there, an unknown person, an intensivist - performs cardiopulmonary resuscitation (CPR), resulting in the restoration of the heart's function. About 300 meters from the scene, an ambulance crew from Emergency Service 112 arrives and immediately transports the patient to the hospital. Upon hospital admission, the patient is in extremally critical condition. He is in comatose state with critical vital signs. Three minutes after arrival, the patient is placed on mechanical ventilation. A CT scan with intravenous contrast is performed under a polytrauma protocol and the patient is then taken to the operating room.

The patient exhibited the following injuries: the spleen is completely severed from its vascular pedicle, which is the main source of bleeding. The liver is partially severed from the ligamentous attachments, a laceration is observed between segments V-VI, bleeding wound, left diaphragmic rupture, the stomach and loops of the large intestine are rotated into the pleural cavity, bilateral pneumothorax and hemothorax, bilateral pulmonary contusion, fractures of ribs I, II, III, IV, V, VI, VII, VIII and IX on the left side, fractures of rib IV on the right side. Approximately 1,5 liters of hemorrhagic effusion in the abdominal and thoracic cavity. Also noted left tibial fracture and multiple superficial injuries.

SURGICAL INTERVENTION/TREATMENT: The patient underwent surgical procedures in 1 hour and 50 minutes: Laparotomy, transabdominal total splenectomy, Liver suturing, transabdominal diaphragmatic repair due to rupture, bilateral pleural cavity drainage, abdominal cavity sanation and drainage, suturing of multiple wounds. The patient was removed from mechanical ventilation on the 6th day, stayed in the pediatric intensive care unit for 14 days and was discharged from the clinic after 20 days. They completed a three-month rehabilitation course in Georgia and are now fully healthy.

CONCLUSION: Polytrauma in children is a serious and complex problem. Timely diagnosis, rapid medical intervention and preventive measures are essential to safeguard children's health and survival. Statistics show that polytrauma remains one of the leading causes of pediatric mortality and hospitalization.

4.EKATERINE KIPIANI, IRINA MARGOSHVILI, TAKO ADEISHVILI, TEONA GHONGHADZE

ENTEROVIRAL ENCEPHALITIS AS AN UNDERRECOGNIZED CAUSE OF FEBRILE SEIZURES IN A PEDIATRIC PATIENT: A CASE REPORT

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INTRODUCTION: Febrile seizures are a common neurological event in childhood, with a variety of infectious and non-infectious etiologies. Enteroviral encephalitis, despite its relatively low incidence (3–6 per 100,000 annually), remains an important but often underrecognized cause, especially in atypical clinical presentations. Enteroviruses (EVs) have emerged as one of the important etiological agents as a causative organism for encephalitis, especially in children and adults. After the first report of EV encephalitis cases in 1950s, there have been increasing reports of regular outbreaks of EV encephalitis worldwide. Enteroviruses are RNA viruses of the family Picornaviridae that consists of more than 100 serotypes, which are characterized by a single positive-strand genomic RNA. The clinical features are pleomorphic and can be accompanied by mucocutaneous manifestations or isolated encephalitis only. The incidence of encephalitis in EV infection is reported to be about 3% and is associated with high mortality and morbidity. A number of newer therapeutic agents have been used in EV encephalitis with variable results. This review will focus on clinical features, pathophysiology, and newer treatment modality in EV encephalitis.

CASE DESCRIPTION: 5-year-old male admitted with a febrile seizure following a 3-day prodrome of rhinorrhea, low-grade fever, and mild cough. Upon admission, the patient was hemodynamically and respiratory stable, with negative meningeal signs and normal laboratory values. Three hours post-admission, the child experienced a generalized tonic-clonic seizure lasting 10 minutes, controlled with anticonvulsants. Neurological consultation and cranial CT scan revealed no abnormalities. On day four, multiple brief seizure episodes recurred under a low-grade fever. revealed focal epileptiform activity in the right posterior temporal region. Valproic acid (Depakine) was initiated. Two days later, a febrile episode was again complicated by a prolonged (18–20 minute) tonic-clonic seizure. CSF analysis detected enterovirus. The patient was managed symptomatically under close neurological observation and discharged in stable condition.

CONCLUSION: This case highlights the importance of considering enteroviral encephalitis in the differential diagnosis of febrile seizures, particularly when EEG findings suggest focal activity and conventional imaging is unrevealing. Early suspicion, diagnostic confirmation, and supportive management can significantly influence clinical outcomes.

5.GVANTSA ARVELADZE, TEIMURAZ MIKELADZE

MITOCHONDRIAL DISEASES – KEARNS-SAYRE SYNDROME FROM PATHOGENESIS TO THE LATEST METHODS OF TREATMENT AND MANAGEMENT

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INTRODUCTION: Mitochondrial disease is a clinically heterogeneous group of diseases caused by the genetic pathogenic variants underlying primary mitochondrial diseases, which can arise from the mitochondrial genome or the nuclear genome, (mtDNA) mutation may be due to somatic or germline mosaicism. The type

of inheritance of (nDNA) mutations can be either autosomal recessive or autosomal dominant. Mitochondrial DNA can only be passed almost exclusively from the maternal line. Mitochondrial myopathies generally affect predominantly or exclusively skeletal (striated) muscle, but in some cases cardiac and/or smooth muscle can be involved. The different distribution of mitochondria in organs and the varying degrees of mutation determine the clinical diversity of mitochondrial diseases. Despite the fact that the involvement of the nervous and muscular systems is the most common, mitochondrial diseases can affect any organ, begin in any age group, both in early and late childhood and adolescence, and manifest precisely with conditions such as asthenia, easy fatigue, weakness, ptosis, ophthalmoplegia, neurosensorial hearing loss and etc., Various stress factors can become a trigger for the development or worsening of symptoms of mitochondrial diseases.

KEARNS-SAYRE SYNDROME (KSS) is a rare neuromuscular condition. It impacts eyes and other parts of your body, including heart. The diagnostic criteria for KSS include the triad of CPEO, pigmentary retinopathy, and onset before age 20 years. At least one other feature should be present for diagnosis: cardiac conduction defect, cerebellar ataxia, or raised cerebrospinal fluid protein (>100 mg/dL). Additional features may include short stature, anemia, diabetes, deafness, and cognitive deficits or intellectual disability.

CASE DESCRIPTION: K A, 11 yo, *Main complaints*: hand tremor, hearing loss, ataxia, ptosis. *Anamnesis*: G1P1, pregnancy and delivery uneventful, born term with C-section, BW – 3350 gr, BL – 52 cm. She was considered healthy till 7 years. From 7 years – astigmatism, from 8 years – hearing loss (progressive), audiometry – 25% hearing loss in one year and 50% in another, wears hearing aids. She is disturbed with noise lately and prefers to remove the aids periodically during the day. Small mandible also became obvious from 8 years – wears braces for correction. Mouth is often open, causing dryness and cracking of lips. From 9 years – ptosis, diagnosed with Ascher syndrome, photosensitivity. From February 2024 – hand tremor, which is progressive. Hand gets tired easily during writing or drawing, then she takes some rest and continues the task slowly. Calligraphy also worsened. Intention tremor is obvious while using a spoon/fork during eating. She has difficulty walking up or down the stairs independently, and needs support. Her regular walking at that point was not ataxic, but running and jumping is difficult. She went to school and attended the classes, has normal cognitive development, performs well academically, but periodically failed down while walking. Lately she prefers to be seated during the day, rather than being physically active. She eats and chews food very slowly with small portions. If a mother insists on eating slightly more food, then she wants, she'll vomit. She is prone to constipation. Current weight -19 kg (below 3rd percentile), height – 120 cm (below 3rd percentile), Heart US – N. *Diagnosis/testing:* In blood: Lactate – 245mg/l (<300), CK – 282 (<154). *Brain MRS*: symmetrical in the thalamus, in the inner capsule, in the midbrain, dorsally in the pons, in the cerebellum and in the white matter of big hemispheres there are hyperintense signals (T2tse, trim), which give limited diffusion in DW1 regime, but ADC is high. Spectroscopy shows high lactate peak. 1.3 ppm, radiological picture suggestive of Leigh syndrome.

Whole exome sequencing: Identified in mtDNA (chrM:7495-15519). Single large-scale mitochondrial DNA deletion syndromes (SLSMDSs) comprise overlapping clinical phenotypes including Kearns-Sayre syndrome (KSS), KSS spectrum, Pearson syndrome (PS), chronic progressive external ophthalmoplegia (CPEO), and CPEO-plus.

TREATMENT AND MANAGEMENT: Targeted therapy: Coenzyme Q10 and antioxidants-Evicap, L-carnitine, Alpha-lipoic acid, Folinic acid, supportive care can help reduce the risk of complications, which includes occupational and physical therapy. Mitochondrial transplantation is a promising therapeutic

approach for the treatment of mitochondrial diseases caused by mutations in mitochondrial DNA, as well as several metabolic and neurological disorders Mitochondrial replacement therapy (MRT), which is already being implemented in clinics in several leading countries around the world, is mainly carried out through tunneling nanotubes (TNTs) and extracellular vesicles (EVs).

CONCLUSION: The patient presented with clinical signs that are typical for an 11-year-old.: Severe ataxia manifests - Astasia, Abasia. Dysmetria, Dysdiadochokinesia, Complete sensorineural deafness, Severe ophthalmoplegia, bilateral ptosis, dementia, Dysphagia, Peripheral retinal degeneration with dystrophic foci. Also, laboratory data show an increase in Lactate and Creatine kinase (CK), decrease in parathyroid hormone concentration (Hypoparathyroidism), and metabolic imbalance. As well whole exome sequencing: Identified in (mt DNA) (chrM:7495-15519) overlapping clinical some clinical phenotypes of mitochondrial disease including Kearns-Sayre syndrome (KSS) Allows us to confirm the rare mitochondrial disease Kearn-Sayre syndrome.

6.MAKA TEVZADZE, IRMA KAKAURIDZE

SUCCESSFUL ADMINISTRATION OF NITRIC OXIDE IN MULTIPLE ORGAN FAILURE

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INTRODUCTION: Multiple organ failure in pediatric patients is a life-threatening condition with limited therapeutic options. This case report describes the successful administration of inhaled nitric oxide in a 5-month-old girl presenting with MOF secondary to severe pulmonary hypertension, confirmed by echocardiography, with additional complications including (renal dysfunction, cardiovascular instability). Inhaled nitric oxide was initiated as a rescue therapy to address pulmonary hypertension and improve oxygenation. Over the course of treatment, INO administration led to significant improvements in pulmonary artery pressures, oxygenation indices, and stabilization of hemodynamic parameters. The therapy was well-tolerated, with minimal adverse effects. The patient's condition stabilized, allowing for weaning from mechanical ventilation and eventual recovery from MOF. This case highlights the potential role of INO as an adjunctive therapy in managing MOF associated with pulmonary hypertension in pediatric patients, emphasizing the need for careful monitoring and individualized treatment strategies.

CASE DESCRIPTION: We report the case of 5-month-old previously healthy girl, who was brought to the ER at 03.12.23 with a 3-days history of fever, rhinorrhea, eye discharge, intermittent vomiting and diarrhea. The parents reported a recent upper respiratory infection but no significant past medical history, prematurity, or family history of pulmonary or cardiac disease.

On admission-the patient's condition was moderately severe (APACHE II- 15- 25%mort.) Vital sign: HR-172 RR-44 T-39,5c Sa02- 98% room air. Laboratory investigations revealed elevated infection markers: CBC- wbc-28.32 X10^9/L, NEUT abs 23.27x10^9/L, PLT 124X 10^9/L, ESR 24mm/hr, CRP- 164 mg/l, Urinalysis- N, ABG-N. Chest x-ray- Without infiltration. A/B therapy-started ceftriaxone, symptomatic treatment. Diagnosis- Bacterial infection, unspecified (A.49.9). The patient's condition rapidly deteriorated (1-3 days), requiring intensive care management, worsening respiratory symptoms, septic shock clinic was revealed. Blood, sputum, urine culture was sent. Changed a/b therapy with meropenem/vancomycin. Icu management

continued with fluids, pressor. Clostr. Defficile, Bordetella pertussis, covid a/b and nazo-pharyngeal swab test were sent. LP-CSF- negative protein and wbc count. Urynalisis-N. *Head CT -N. chest x-ray*- acute respiratory distress revealed. *abdomen US*-N. *Cardiac US*: patent foramen oval, mitral valve stenosis, mild. EF%- 54%. PASP - 25mmHg. High flow nasal canula and non-invasive ventilation were initially attempted but failed to maintain adequate oxygenation. Intubated due to worsening respiratory failure. Started mechanical ventilation with Pres SIMV/PC, RR-30,PIP-26cmH2O, PEEP-7.0cmH2O, fiO2-1. Continued oxygenation/ventilation deterioration, changed regimes and parameters SIMV/PC--> SIMV/VC, RR-40, Vt-120, PEEP-12.0cmH2O, fiO2-1.--> HFO. Oxygenation index -15—>20-- > 25 (OI=MAPxFio2x100/PaO2) Continued tissue hypoperfusion, oliguria, hypotension. Revealed elevated renal function, hypo coagulation. Rapidly increasing biomarkers: WBC-58.57 10^9/L, PCT-10 ng/mL, *chest US*-bilateral pleural effusion. On the *Cardiac US – pulmonary hypertension* (PASP - 60mmHg). Concilium was assembled- started INO with dose 20-40 ppm.

RESULTS: Administration of INO resulted in a significant improvement in oxygenation, with an increase in partial pressure of arterial oxygen (PaO2) and a reduction in oxygenation index within. (timeframe, e.g., 30-60 minutes). No significant adverse effects attributed to INO. Within 45-minute oxygen saturation increased SpO2-85-->94%. within 8-hour pulmonary pressure decreased (PASP -- 50mmhg, -- 40 mmHg (of systemic pressure 1/3). 24 hour later INO - temporary removed (40-->25-->15ppm.). gradually removed sedation-analgesia-relaxation. Ten days later extubating. All cultures were negative. 03.01.24 was discharged with no neurological sequelae. Laboratory parameters and developmental status remained normal at a three-month follow-up.

CONCLUSION: This case highlights the potential utility of INO as a rescue therapy in infants with multiple organ failure complicated pulmonary hypertension, improving oxygenation in critical settings. However, its impact on overall mortality and long-term outcomes in such complex cases requires further investigation. Careful monitoring for adverse effects is essential.

7.MAKA TEVZADZE, TAMAR BERUASHVILI

CLINICAL CASE MANAGEMENT OF THE HEMOLYTIC UREMIC SYNDROME (HUS) IN A MULTI-SPECIALTY HOSPITAL

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INTRODUCTION: The hemolytic uremic syndrome (HUS) is defined by the sudden onset of microangiopathic hemolytic anemia, thrombocytopenia, and acute kidney injury (AKI). It is a form of Thrombotic microangiopathy (TMA) most commonly caused by Shiga toxin-producing *Escherichia coli* (STEC). Classification has been developed based on pathophysiologic considerations and triggering factors. Hereditary causes of HUS: Complement gene mutations; Inborn errors of cobalamin C metabolism; Diacylglycerol kinase epsilon (DGKE) gene mutations. Acquired causes of HUS is Infection: Shiga toxin-producing Escherichia coli (STEC), Streptococcus pneumoniae and Human immunodeficiency viral infection, rare occurrences in pregnant patients or those with autoimmune disorders

(eg, systemic lupus erythematous), Autoantibodies to complement factors, Drug toxicity, particularly in patients with cancer or solid organ transplant recipients.

EPIDEMIOLOGY: Shiga toxin-producing E. coli (STEC) hemolytic uremic syndrome (HUS) accounts for over 90 percent of cases of HUS in children, Pneumococcal-associated hemolytic uremic syndrome (HUS) has been reported in 5 to 15 percent of all childhood cases of HUS, and in 40 percent of non-STEC HUS cases, Complement-mediated HUS is a relatively rare disorder, prevalence of 7 per 1 million children in Europe. Most complement-mediated HUS cases are due to gene mutations of complement factors, Antibodies to complement proteins have been implicated in the etiology of 6 to 10 percent of patients with complement-mediated HUS.

CLINICAL MANIFESTATIONS CHARACTERISTICS OF HUS: Microangiopathic hemolytic anemia, Thrombocytopenia and Acute kidney injury. Some patients with HUS may present with one or more of the following complications: Neurologic abnormalities – such as seizures, strokes, and decreased level of consciousness occurring in 10 percent of cases are predictors of poor outcome. In any patient with HUS who presents with serious neurologic dysfunction (e.g., seizure and coma), Gastrointestinal complications – e.g., hemorrhage, pancreatitis, Respiratory complications – e.g., acute respiratory distress syndrome, respiratory failure, Hypertension – Particularly in patients with complement-mediated HUS, Cardiac complications – Cardiomyopathy and myocardial ischemia. Manifestations of CNS involvement include altered mental status, seizures, coma, stroke, hemiparesis, and cortical blindness. Major CNS abnormalities are typically seen in up to 20 to 33 percent of cases. In patients with severe neurologic findings, brain magnetic resonance imaging reveals bilateral hypersignal on T2-weighted and hyposignal on T1-weighted images in the basal ganglia, thalami, and brainstem. Severe CNS involvement is associated with increased mortality. In addition, severe hypertension may result in CNS symptoms and require emergent therapy to decrease blood pressure.

CASE DESCRIPTION: Herein we report the case of a 7-year-old female with 3-days history of subfebrile temperature, recurrent vomiting and persistent bloody diarrhea. The patient was transferred from a regional hospital to M. Iashvili Central Children's Hospital. At the emergency department: vital signs upon admission: T – 36.8°C; HR – 139; RR– 26; Blood pressure – 115/80 mmHg; SpO_2 – 98%. Objectively: asthenia, somnolence, petechiae on the chest, swelling of the face, abdomen, and limbs; history of 24-hour anuria. Laboratory tests confirmed acute kidney injury: Creatinine – 6.61 mg/dL (Normal: 0.3–1.0); Urea – 29.22 mmol/L (Normal: 2.5–8.3). Complete blood count: WBC – 19.9 x 10^9/L; Hemoglobin – 10.3 g/dL; RBC – 3.44 x 10^{12}/L; Hematocrit – 26.5%; Platelets – 75 x 10^9/L; Liver function tests: ALT – 277 U/L; AST – 168 U/L; LDH – 10,215 U/L. The patient was referred to Nephrology ward and treatment was initiated with the diagnosis of hemolytic uremic syndrome and acute renal failure. Shiga toxin-producing *Escherichia coli* (STEC) was confirmed by laboratory tests. The patient experienced a seizure during the first dialysis session and was transferred to the PICU. We performed a brain MRI scan, where findings showed metabolic changes, which is characteristic of HUS. Anticonvulsant therapy was instituted. On the 21th day after the onset of the disease developed seizures and anisocoria. The patient was placed on mechanical ventilation. A brain CT scan performed: On the left side, in the frontal-parietal-occipital lobe, there was an intracerebral hemorrhage with marked perifocal edema and mass effect on the midline structures, measuring 3.1x5.0x4.5cm, Midline shift is 0.5cm. The patient underwent neurosurgical intervention - Left-sided decompressive craniectomy, evacuation of intracerebral hematoma. On the ninth day after the operation, the patient was extubated. The treatment provided by our team: ten times RBC and blood component transfusions; 23 hemodialysis sessions, antibiotic therapy:

Meropenem + Vancomycin, Colistin, Fosfomycin + Linezolid; Vasopressor/inotropic support; antihypertensive and anticonvulsant therapy: Sol. Diazepam, Sol. Midazolam, Sol. Phenobarbital; Neurosurgical intervention: Left-sided decompressive craniectomy and evacuation of hematoma.

RESULTS: Dialysis was discontinued on the 27th day after the onset of the disease. The patient was discharged on the 53rd day of hospitalization with full recovery of kidney function and mild right-sided paresis.

CONCLUSION: Considering the severe course and the prognosis of the disease, we report the efficient multidisciplinary (Nephrologists, Intensivists, Anesthesiologists, Neurologists, Neurosurgeons, Infectious disease specialists, Cardiologist, Transfusion specialists, Radiologists and Ophthalmologist) management of the patient with favorable outcome.

8.NINO MCHEDLISHVILI [1],MARIAM GUGUNISHVILI [2,3],TAMAR ADEISHVILI [2],NINELI CHKHAIDZE [3]

RECURRENT PAROXYSMAL LARYNGOSPASM ASSOCIATED WITH LARYNGOPHARYNGEAL REFLUX

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INTRODUCTION: Episodic laryngospasm is most commonly observed in children aged 6 months to 3 years. In older children, recurrent croup may be more frequent and is often associated with gastroesophageal reflux (GERD), allergies, and laryngeal anomalies.

MATERIALS AND METHODS: We present the case of a 6-year-old girl followed at the Center of Allergy and Immunology, whose episodes of laryngospasm became more frequent with age. The child suffered from paroxysmal, occasionally barking cough, dysphonia, and desaturation, leading to frequent emergency room visits. In several instances, she required treatment in the intensive care unit due to respiratory distress. She was evaluated by ENT, gastroenterologist, and pulmonologist. Laryngoscopy and chest CT revealed no pathological findings. At age 4, specific IgE test for aeroallergens was negative. One episode of bronchospasm was documented, and inhaled fluticasone propionate 125 mcg via spacer was prescribed. Despite treatment, she experienced four exacerbations in one month. Significant improvement was observed only after adrenaline nebulization in the ICU. Spirometry and exhaled nitric oxide at age 6 were within normal limits. Repeated skin prick testing remained negative. Brain MRI and EEG were normal. A repeated consultation with a gastroenterologist was conducted, and esophageal barium swallow revealed gastroesophageal reflux both in upright and supine positions, including under mild compression.

RESULTS. Antireflux therapy was initiated with proton pump inhibitors (PPIs), antacids, and H2 receptor blockers. The patient showed marked clinical improvement following treatment.

CONCLUSION: This clinical case highlights laryngopharyngeal reflux (LPR) as a significant and often underdiagnosed cause of recurrent, paroxysmal laryngospasm in children. In the presented case, allergic, anatomic, neurologic, and infectious causes were excluded, and only repeat gastroenterological evaluation confirmed LPR. Active screening for LPR and individualized management strategies are recommended to reduce complications and improve quality of life in similar pediatric patients.

9.NINO SIRADZE, SOPIO TSERTSVADZE, DAVID MAKHATADZE

ALAGILLE SYNDROME (ALGS) CASE PRESENTATION

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INTRODUCTION: Alagille syndrome (ALGS) is a multisystem autosomal dominant disorder with a wide variety of clinical manifestations. It is known as arteriohepatic dysplasia, Alagille-Watson syndrome, Watson-Miller syndrome, or syndromic bile duct paucity. It most commonly results from pathogenic variants in the JAG1 gene or, less frequently, NOTCH2. The severity of the disease can range from a subclinical presentation to a life-threatening condition, with a mortality rate up to 10%.

CASE DESCRIPTION: We report a rare case of a 2- years-old female infant with persistent jaundice from the third month of age, accompanied by hyperbilirubinemia, hemolytic anemia, hepatosplenomegaly and ascites. Extensive laboratory and imaging evaluations ruled out infectious, hematologic, and metabolic etiologies such as G6PD deficiency, pyruvate kinase deficiency, Wilson's disease, and galactosemia Imaging revealed a cirrhotic liver, splenomegaly, agenesis of the gallbladder, and abnormal hepatic arterial vasculature. Echocardiography showed bilateral pulmonary artery branch stenosis, and ophthalmological examination revealed posterior embryotoxon and iris hypoplasia. Genetic testing identified a pathogenic heterozygous variant in the JAG1 gene, confirming the diagnosis of Alagille syndrome.

CONCLUSION: This case illustrates the diagnostic challenges associated with Alagille syndrome, particularly in infants with atypical and overlapping features. It emphasizes the necessity of early genetic testing in patients with cholestasis, hepatic anomalies, congenital heart defects, and characteristic facial or ocular features, even when initial metabolic and infectious screens are inconclusive.

10.SALOME CHIGHLADZE, TEONA SHATIRISHVILI, TINATIN TKEMALADZE, EIRIK BRATLAND, KAKHA BREGVADZE, NINO TATISHVILI, ELENE ABZIANIDZE, GUNNAR HOUGE, SOFIA DOUZGOU

MSMO1 DEFICIENCY: A POTENTIALLY PARTIALLY TREATABLE, ULTRARARE NEURODEVELOPMENTAL DISORDER

M.Iashvili Children's Central Hospital, Georgia

INTRODUCTION: MSMO1 deficiency is an autosomal recessive disorder of cholesterol metabolism, characterized by developmental delay, microcephaly, immune dysfunction, and psoriasiform dermatitis. Only five cases have been previously reported. We describe two Georgian siblings with novel clinical findings - polydactyly, alopecia, and spasticity - and a homozygous c.548A>C (p.Glu183Ala) MSMO1 variant.

MATERIALS AND METHODS: Clinical evaluation, biochemical profiling, and brain MRI were performed. Cytogenomic microarray confirmed regions of homozygosity. Whole-exome sequencing identified a novel likely pathogenic variant. Protein modeling and in-silico analyses supported its damaging effect.

RESULTS: Both siblings had early-onset spasticity, developmental delay, psoriasiform dermatitis, and alopecia. One had preaxial polydactyly. Brain MRI was normal. The variant p.Glu183Ala disrupts enzyme catalytic structure. A treatment regimen using cholesterol supplementation, rosuvastatin, bile acids, and topical cholesterol/statin formulation led to significant dermatological improvement and mild hair regrowth.

CONCLUSION: MSMO1 deficiency should be suspected in patients with neurodevelopmental delay and dermatological findings. Early diagnosis and targeted treatment can improve dermatologic symptoms, though neurologic benefit remains limited. This study expands the phenotypic spectrum and highlights treatment strategies in resource-limited settings.

11.TAMTA KAPANADZE, NINO KHELADZE, NINO TOGONIDZE

VAN WYK – GRUMBACH SYNDROME IN A CHILD WITH DOWN SYNDROME: WHEN HYPOTHYROIDISM MASQUERADES AS HEMATURIA AND OVARIAN CYSTS

M.Iashvili Children's Central Hospital, Georgia

INTRODUCTION: Van Wyk–Grumbach syndrome (VWGS) is a rare complication of untreated juvenile hypothyroidism, characterized by isosexual precocious puberty, multicystic ovaries, and delayed bone age. Children with Down syndrome (DS) are particularly prone to thyroid dysfunction, especially autoimmune hypothyroidism, which may present atypically due to overlapping clinical features of DS.

CASE DESCRIPTION: We report the case of a six-year-old girl with Down syndrome who presented with abdominal distension, recurrent episodes of macrohematuria, pancytopenia, and mildly elevated creatinine levels. Initial imaging revealed hepatosplenomegaly and a large, thin-walled ovarian cyst. The clinical picture raised concerns for a possible urologic or oncologic condition. However, further evaluation revealed profound hypothyroidism (TSH: 1088 μIU/mL, FT4: <5.4 pmol/L), elevated estradiol and FSH levels, and delayed bone age. The bleeding episodes were ultimately identified as metrorrhagia, and the ovarian findings were consistent with multicystic ovarian enlargement - both hallmark features of Van Wyk–Grumbach syndrome (VWGS). Strongly positive thyroid autoantibodies confirmed the diagnosis of autoimmune thyroiditis.

MANAGEMENT AND OUTCOME: The patient was initiated on high-dose levothyroxine therapy (75 μg/day), which led to rapid clinical improvement. Metrorrhagia resolved within two weeks of treatment initiation, and serial laboratory evaluations demonstrated a progressive decline in TSH levels. Bone marrow aspiration excluded hematologic malignancy as the underlying cause of pancytopenia. The ovarian cyst, consistent with a hormonally driven functional cyst, was managed conservatively without the need for surgical intervention.

DISCUSSION: This case underscores the importance of considering VWGS in the differential diagnosis of early vaginal bleeding and ovarian cysts in prepubertal girls. It also highlights the diagnostic complexity in DS patients, where atypical presentations of endocrine disorders are common. The hematologic abnormalities, while uncommon, may also be attributed to severe hypothyroidism. Early recognition and treatment of VWGS can prevent invasive diagnostics and surgeries.

CONCLUSION: VWGS should be considered in prepubertal girls with ovarian cysts, abnormal bleeding and growth delay - especially in patients with DS. Routine thyroid screening is essential in this population to prevent such complications. This case emphasizes the systemic consequences of undiagnosed hypothyroidism and the reversibility of symptoms with timely hormonal replacement.

12.TORNIKE KLDIASHVILI, ZVIAD MALAZONIA, TSOTNE GVASALIA, GIORGI KORKOTASHVILI, DAVIT LALIASHVILI, GIORGI LALIASHVILI, ANANO GABRITCHIDZE

BRAIN ABSCESS IN A 3-YEAR-OLD CHILD FOLLOWING SINUSITIS: A CASE REPORT

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INTRODUCTION: Contiguous infections like sinusitis or otitis media are often the cause of brain abscesses, which are rare but potentially fatal conditions in children. In order to avoid serious neurological consequences, prompt diagnosis and treatment are essential. Targeted antimicrobial therapy is made more difficult by the fact that a sizable fraction of cases are still culture-negative.

CASE DESCRIPTION: We describe a 3-year-old boy who had neurological impairments, fever, and lethargy. He had chronic sinusitis in the past. Microbiological studies, including CSF cultures and PCR panels, were negative, but laboratory tests showed elevated inflammatory markers (CRP 48 mg/L) and a CSF white blood cell count of 162/μL. A left frontal ring-enhancing lesion that was consistent with a brain abscess was discovered by brain MRI. Despite surgical drainage, intraoperative cultures were unable to detect a pathogen in the patient. For four weeks, cefepime and vancomycin were given as part of an empiric intravenous antibiotic regimen.

OUTCOME: The postoperative course was favorable, with resolution of neurological symptoms and normalization of inflammatory markers. Follow-up MRI at 3 months confirmed complete resolution of the abscess. The child remained neurologically intact and in good health.

CONCLUSION: This case highlights the necessity of maintaining a high index of suspicion for brain abscess in pediatric patients with sinusitis and neurological symptoms. Even in culture-negative cases, prompt surgical intervention and empiric broad-spectrum antibiotic therapy can lead to complete recovery and excellent neurological outcomes.

13.TAMAR MICHITASHVILI, NATIA NATROSHVILI

NONKETOTIC HYPERGLYCINEMIA IN NEWBORNS

Iv.Bokeria University Hospital

INTRODUCTION: Nonketotic hyperglycinemia (NKH) is a rare metabolic disorder caused by a defect in glycine metabolism. It is inherited in an autosomal recessive pattern. NKH typically presents with severe encephalopathy that rapidly progresses and can ultimately lead to respiratory failure. Routine laboratory tests are often normal - there is no acidosis, hypoglycemia, hyperammonemia, or specific evidence of organ dysfunction. Clinically, the condition frequently presents with persistent hiccups, seizures, and neurologic deterioration.

DIAGNOSIS: While elevated glycine in plasma is a hallmark, it may be less prominent in neonates due to immature renal reabsorption mechanisms. Urinary amino acid analysis can be especially helpful, as it may reveal elevated glycine excretion.

CASE DESCRIPTION: A term male newborn, G1P1, delivered via spontaneous vaginal delivery. Clear amniotic fluid. Gestational age: 40+2 weeks, Birth weight: 3270 g, Length: 51 cm, Apgar scores: 8/8. GBS

status: negative. Received Hepatitis B vaccination after birth. Maternal history: 20 years old, with a history of bronchial asthma. Paternal history: 24 years old, has seizures, currently undergoing treatment. Parents deny consanguinity. At 48 hours of life, the newborn developed profound hypotonia, adynamia, and was not sucking. Transferred to the neonatal intensive care unit (NICU). On examination: Marked hypotonia and adynamia, opened eyes to stimuli; facial grimace to pain, no crying, weak suck reflex, absent Moro reflex, Vital signs: stable, did not require oxygen support initially. Laboratory findings: Normal infection markers, normal glucose, normal lactate, normal electrolytes. Later that same day: Clinical deterioration with persistent hiccups, followed by clonic seizures, bradycardia tendency, apnea, worsening hypotonia and adynamia. Lumbar puncture was performed to rule out neuroinfection: Cytosis: 30/3, Protein/glucose: normal, CSF culture: sterile. By 12 hours after onset, respiratory depression developed with desaturation. The infant was started on non-invasive ventilatory support and later transitioned to mechanical ventilation (SIMV mode) on day 3 of life. A metabolic disorder was suspected. Enteral feeding was stopped, and amino acids were removed from parenteral nutrition. Ammonia level: 110.3 μmol/L (normal: 16–60). By 2 weeks of age, the infant was weaned from mechanical ventilation, transitioned to non-invasive respiratory support (HFlowNC), and later to mask oxygen. Neurological status remained severely impaired: Opened eyes spontaneously but did not fix gaze, no response to environment, persistent severe hypotonia, adynamia, chaotic limb movements observed periodically, no head control, absent age-appropriate reflexes, no sucking, feeding via NG tube. At 4 months of age, amino acid analysis in plasma showed: Glycine: 3086 mg/L (normal <32), Urine glycine: 65 mg/L (normal <32). Genetic testing results: KCNA1 gene: c.394G>T p. (Glu132*) - known pathogenic variant associated with episodic ataxia type 1GLDC gene: c.2276C>G p. (Pro759Arg) - results in substitution of proline by arginine at position 759; classified as likely pathogenic, associated with nonketotic hyperglycinemia (NKH).

TREATMENT CONSIDERATIONS: Sodium benzoate and hydration may help reduce plasma glycine levels, but do not affect CSF glycine accumulation. Respiratory function may recover, but neurological injury is irreversible. The long-term prognosis is poor. Only rare transient forms of NKH may have a relatively milder course.

CONCLUSION: recognizing and diagnosing rare diseases is extremely important, as only after diagnosis do treatment possibilities become available.